Everyone is unique:
Dealing with diversity in genetic counselling

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Declaration

I declare that this thesis is my own account of my research.

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Abstract

The rapidly expanding volume and complexity of genetics knowledge, and an increasingly culturally diverse society, have led to changes in genetic counsellors’ roles. The profession continues to evolve from an information-giving medically based model to encompass a broader psycho-social perspective. This thesis examines how Australasian genetic counsellors engage with cultural diversity and their approach to working with culturally diverse clients, in particular those in consanguineous relationships.

The thesis research proceeded in three stages. In the first stage, 69 questionnaires were completed by people in consanguineous relationships who received genetic counselling. The questionnaire examined aspects of their relationship and their counselling experiences. The experiences of 16 participants were further explored through thematic analysis of qualitative interview transcripts. In the second stage, Australasian genetic counsellors’ views on consanguinity and cultural diversity were investigated through 58 postal questionnaires. The transcript of a discussion group with seven genetic counsellors was also reviewed using interpretive content analysis. In the third stage, the dialogue between genetic counsellors and clients was examined through audio-recordings of genetic counselling sessions and post-session questionnaires.

This research reveals that consanguineous couples had positive and negative relationship experiences. The majority were from backgrounds where this is an uncommon marriage form and experienced social isolation. Social pressures outweighed the increased genetic reproductive risks. The data reveals a shift in genetic counsellors’ engagement with cultural difference. Skills in managing diversity were evident, but barriers to application included continuing dominance of the educational model, lack of training and limited resources.
The research highlights challenges inherent in working with diversity. Developing skills and knowledge relevant to particular client groups is frequently suggested; this thesis argues that a model where all clients are recognised as unique individuals could be more effective. Recommendations for enhancing existing skills of genetic counsellors and facilitating bidirectional cultural dialogue are provided.
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Preface

Throughout the thesis I have chosen to use the Australian spelling for counsellor except where the text is a direct quote from a publication that uses the American spelling counselor.

Sections from Chapters Five and Six were presented at the following conferences:

Mountain, H. and Barns, I.
Poster The dialogical model of genetic counselling and transcultural interactions in Western Australia: A quantitative and qualitative study.

Mountain, H. and Barns, I.
Oral The dialogical model of genetic counselling and transcultural interactions in Western Australia: A quantitative and qualitative study.

Mountain, H. and Barns, I.
Poster A survey of Australasian genetic counsellors’ views on cultural difference and consanguinity.
International Congress of Human Genetics, Brisbane 2006.

Mountain, H. and Barns, I.
Oral Consanguineous couples’ experiences of genetic counselling.

Mountain, H. and Barns, I.
Oral An exploratory study of genetic counselling sessions and whether cultural difference affects rapport or communication.
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## Abbreviations

<table>
<thead>
<tr>
<th>Abbreviation</th>
<th>Description</th>
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<tbody>
<tr>
<td>ASGC</td>
<td>Australasian Society of Genetic Counsellors</td>
</tr>
<tr>
<td>ASHG</td>
<td>American Society of Human Genetics</td>
</tr>
<tr>
<td>BRCA1/2</td>
<td>Breast cancer gene 1/2</td>
</tr>
<tr>
<td>CF</td>
<td>Cystic fibrosis</td>
</tr>
<tr>
<td>DR</td>
<td>Doctor</td>
</tr>
<tr>
<td>GC</td>
<td>Genetic counsellor</td>
</tr>
<tr>
<td>GHP</td>
<td>Genetic health professional (genetic counsellor or geneticist)</td>
</tr>
<tr>
<td>GP</td>
<td>General practitioner</td>
</tr>
<tr>
<td>GSWA</td>
<td>Genetic Services of Western Australia</td>
</tr>
<tr>
<td>HGP</td>
<td>Human Genome Project</td>
</tr>
<tr>
<td>HGSA</td>
<td>Human Genetics Society of Australasia</td>
</tr>
<tr>
<td>ND</td>
<td>Nondirectiveness</td>
</tr>
<tr>
<td>NSGC</td>
<td>National Society of Genetic Counsellors</td>
</tr>
<tr>
<td>NSW</td>
<td>New South Wales</td>
</tr>
<tr>
<td>PGD</td>
<td>Preimplantation genetic diagnosis</td>
</tr>
<tr>
<td>QLD</td>
<td>Queensland</td>
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<tr>
<td>REM</td>
<td>Reciprocal-engagement model</td>
</tr>
<tr>
<td>TOP</td>
<td>Termination of pregnancy</td>
</tr>
<tr>
<td>UK</td>
<td>United Kingdom</td>
</tr>
<tr>
<td>URM</td>
<td>Under-represented minority</td>
</tr>
<tr>
<td>USA</td>
<td>United States of America</td>
</tr>
<tr>
<td>VIC</td>
<td>Victoria</td>
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<tr>
<td>WA</td>
<td>Western Australia/n</td>
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Glossary

**BRCA1 and BRCA2**
Human tumour suppressor genes that have a role in DNA repair. When they function correctly they are protective against the development of certain cancers including breast, ovarian and prostate cancer.

**CHROMOSOME**
A chromosome is an organised structure of DNA, protein, and RNA found in cells.

**CLIENT**
A person who receives help or advice from a genetics service.

**CONSANGUINEOUS**
Related by birth; descended from the same parent or ancestor.

**CONSANGUINITY**
A consanguineous relationship.

**CULTURE**
The beliefs, values, behaviour and material objects that constitute a people’s way of life.

**CULTURAL COMPETENCE**
The awareness of, and respect for, cultural differences and in addition the skill to identify differences that facilitate effective counselling.

**CULTURAL DIVERSITY**
The changing population of the world through migration.

**DE FACTO**
A de facto relationship is recognised under Australian law where two people, who are not married or in a registered relationship, are partnered and usually live together as a couple.

**DIALOGICAL PROCESS**
An interaction that is dynamic, relational and engages all parties. The interaction informs and is continually informed by the previous conversations.

**DEOXYRIBONUCLEIC ACID (DNA)**
The molecule that encodes the genetic instructions for the development, structure and function of living organisms and allows the transmission of genetic information from generation to generation.

**ENDOGAMY**
Marriage within a specific class, ethnic or social group.
GENE
A hereditary unit; in molecular terms, a sequence of chromosomal DNA that is required for the production of a functional product.

GENERAL PRACTITIONER (GP)
A medical practitioner who treats acute and chronic illnesses and provides preventive care and health education to patients. Is synonymous with ‘family doctor’ or primary care provider.

GENETIC COUNSELLING
The process by which patients or relatives, at risk of an inherited disorder, are advised of the consequences and nature of the disorder, the probability of developing or transmitting it, and the options open to them in management and family planning.

GENETIC COUNSELLOR
Health professionals with specialised graduate degrees and experience in the areas of medical genetics and counselling who work as members of a multidisciplinary health care team.

GENETIC HEALTH PROFESSIONAL (GHP)
A term that refers to either a genetic counsellor or clinical geneticist.

GENETICIST
A consultant medical doctor who has specialist training in genetics. A geneticist diagnoses, treats, and counsels patients with genetic disorders or syndromes.

MODEL
A representation containing the essential elements of practice.

PSYCHOSOCIAL
Involves aspects of social and psychological behaviour.
1. **Introduction**

Cultural competence is viewing life as fluid and always changing, and being able to change with it. (Barker, 2001, p.5)
1.1. Background

This thesis explores the practice of genetic counselling in an increasingly complex world: where genetic information is complicated and people seeking genetic counselling are more culturally and linguistically diverse. In this world, the role of a genetic counsellor is demanding, increasingly extending beyond one of simply providing information for clients to one of helping a diverse range of clients make complex reproductive choices. The work described here has been undertaken over an extended time period, allowing firsthand experience of these changes. The thesis explores whether genetic counsellors are equipped to meet these demands, the models of genetic counselling process that best describe the situations dealt with in practice, and the applicability of these models.

Clients are referred to a clinical genetics unit to discuss their risk of having a genetic condition or of having a child with a genetic condition. Often they have heard about a genetic diagnosis in a family member, seen a television program mentioning a genetic condition, or had a test indicating they might be at risk. To clarify what this means for each client, the counsellor must explain the meaning of a ‘genetic condition’, genes and chromosomes, the mechanism by which the genes are inherited, the notion of risk and then give a mathematical estimate of the client’s risk. These are complex concepts that require detailed explanations and translation of terminology and jargon.

Understanding how this complex information is integrated into the client’s life requires the counsellor to address the intense emotions that are often aroused in response to the news that there is a genetic condition in the family. Apart from their personal response, clients also have complex connections to others, as part of a couple, as a family member, and as a member of wider society. An appreciation of how these connections influence understanding and adaptation to new genetic
information is essential if counsellors are to be engaged practitioners in the changing and increasingly geneticised society in which we live.

The first contact between client and counsellor is often by telephone – the voice of a stranger asking for detailed personal, and at times confronting, information. If not by telephone, the first meeting commonly occurs in a clinic consultation room. On rare occasions clients arrive at the clinic because they were passing by and drop in for a consultation or a gene test. Whatever the first point of contact with the client is, or their understanding of the reason for the meeting, the counsellor aims to convey the calm and confident demeanour of an empathic professional.

Unless there has been prior personal or family exposure to a genetic service, many clients will not understand the processes of genetic counselling. In some cases, a referring doctor may have explained the reason for referral and the benefit of attending. Others arrive with little knowledge of what the intended outcome may be. Many say they do not need counselling – they just came to get a test done.

Regardless of why clients attend, a consultation usually begins with an exploration of how the client came to genetic counselling, what is understood about their situation, their knowledge of genetics and genetic testing, and what they are seeking from the consultation. This information assists the counsellor to clarify client goals and determine how the consultation might proceed.

1.1.1. A typical working day in my life as a genetic counsellor

I begin with a personal description of genetic counselling and a small window into my practice. During the morning walk towards my office at GSWA I think about what the day ahead will bring, the clients that I will meet in the clinic, and how much of their narrative I will be privy to on our first, and sometimes only, meeting. Amongst the uncertainty there is one thing I can be sure of – today will not be the same as yesterday.
The clients that I meet in the clinic are like a mosaic of the human race – males, females, children, teenagers, adults, elderly, mothers, fathers, sons, daughters, grandparents, English and non-English speakers, from rural areas or the city, healthy, sick, disabled, heterosexual, gay, married, single, university educated, early school leavers and many possible combinations of these attributes.

After mentally preparing on my walk, I reach my office on the third floor of a heritage-listed building. My office contains the tools of my trade – a desk stacked with papers and patient files, a bookcase filled with books, a telephone with a hands-free headset for comfort during long telephone calls and a computer. Increasingly, my computer is used to maintain contact with colleagues and clients and to keep me updated with the latest research.

I locate the files for the three clients scheduled in the morning clinic and scan them for clues to what the consultations will involve. The first belongs to a female and contains details of who referred her for genetic counselling and the reason for referral. The second file contains details of the client’s cancer diagnosis, medical history, and some medical test results. The third file contains only the barest amount of information – the name of the client, his address and date of birth.

In the hour allocated to each client I plan to establish rapport, listen to their narrative, ascertain the issues important to them, and address their questions and concerns. As I prepare for the clinic, I consider the clients, their situations and how each session might progress.

The first client is a 30 year old woman with a family history of breast and ovarian cancer. I have not seen her before. The referral letter states that she wants to have genetic testing to determine if she has inherited the breast cancer predisposition gene identified in her family. A mutation in the breast cancer gene 1 (BRCA1) gene was found in her mother just prior to her death 8 months ago. She has a 50% risk of inheriting the BRCA1 gene mutation which would give her up to an 85% chance of
having breast cancer in her lifetime. I anticipate that my main task will be assisting her to make an informed autonomous decision about whether or not to have pre-symptomatic genetic testing. I plan to discuss the function and etiology of the gene, advantages and disadvantages of either having or not having testing, and the medical, insurance and psychological impact of testing.

We meet in the waiting room, walk along the corridor to the consultation room and sit down. I ask her why she was there and how could I best help her. A few minutes into the consultation she begins crying and openly expressing her grief over the loss of her mother. I quickly adjust my expectations of how the session will proceed and put aside information and decision making for another time. This need to adapt a session quickly and unexpectedly is not unusual.

The second client arrives with little understanding of the reason she has been referred, hence we spend some time exploring my role and the potential benefit of genetic testing for her and her family. A significant proportion of the third consultation involves eliciting the client’s situation and needs because there was little information in the referral and no pre-clinic contact.

After the clinic I return to my office and have telephone conversations, or meet with medical specialists, general practitioners (GPs), nurses, sick and healthy individuals from a range of professional, educational and social backgrounds. I work as a member of a multidisciplinary health care team, often discussing specific aspects of case management with fellow team members, referring people to other services within and outside the hospital, or putting them in contact with support groups or community services. I frequently act as an advocate for clients if they wish to follow a particular course of action or management. The array of topics discussed in these conversations is wide, ranging from discussion of patient management, giving advice about surveillance recommendations, assessment of genetic risk, decision
making about genetic testing, how to inform relatives about their genetic risk, interpretation of test results and information, grief counselling and decision making.

Late in the day I attend the weekly team clinical meeting. We review the clients seen in the past week and booked for the week ahead. We discuss interesting cases and reflect on our practice. Clients’ narratives are re-told, analysed, and the part we play in their genetics journey reviewed. At the end of the day I walk towards home, relaxing and unwinding as I go, reviewing the day’s events and wondering what the next will bring.

1.1.2. The mediating role of the genetic professional

The above description of my typical working day highlights the important role genetic counsellors play in communicating and contextualising medical genetics knowledge. We mediate between the science and the client by interpreting and explaining technical details and scientific knowledge in a way that can be easily understood and used by the client. As genetic counsellors, we also explore how genetic knowledge impacts on the emotional and psychological wellbeing of the client and their family (Rantanen et al., 2008; Resta et al., 2006).

Balancing the complex information, psychosocial and cultural needs of clients and their families is challenging. Rapid assessment, reassessment and adjustment are often required for each client, their needs and goals, and the unique situation they are in. For these reasons, a mediating role is often required (Bennett et al., 2003; Peters et al., 2011; Pour-Jafari et al., 2010).

In order to execute these roles, genetic counsellors need to be skilled in empathic listening, grief counselling, family and interpersonal dynamics, assessing coping styles and exploring psychological processes with clients who may be experiencing these issues for the first time. These roles require a specialised combination of knowledge, experience and skills (Resta et al., 2006; Runyon et al., 2010).
1.1.3. The changing role of the genetic counsellor

I have been involved in the field of human genetics for 23 years, as both a scientist in both research and clinical diagnostic laboratories and as a genetic counsellor. In the 16 years spent as a genetic counsellor at GSWA, I have witnessed many changes in the way the profession views itself and is viewed by others. Both locally and across Australasia, genetic counsellors have promoted awareness of the profession and developed standards, codes of ethics, accreditation and training programs comparable to our international colleagues.

The burgeoning body of knowledge arising from the completion of the Human Genome Project (HGP) in 2003 has placed new demands on genetic counsellors. As interpreters and educators, genetic counsellors communicate increasing amounts of complex information to their clients and other health professionals (Rantanen et al., 2008; Skirton et al., 2010). The rapid increase in the body of genetic knowledge, the concomitant creation of new possibilities for the diagnosis and management of genetic conditions and the amount of information (not always accurate or balanced) available to clients on the Internet have been the most noticeable changes influencing practice (Skirton et al., 2013; Skirton et al., 2010).

There have also been changes in the roles genetic counsellors play within GSWA. The unit originally consisted of three clinical geneticists and three genetic counsellors. The genetic counsellors prepared patient files for consultations, listened to and, when necessary, reiterated the information given by the geneticist, and managed any psychosocial issues outside the consultation. Since 1997, the team has grown considerably and now comprises six geneticists and 13 counsellors divided into three sections: general genetics (adults and prenatal), paediatrics, and familial cancer. The complexity of clients managed solely by each genetic counsellor has increased, with experienced genetic counsellors now seeing clients that in the past would have been seen by the clinical geneticist. These clients include advanced maternal age and consanguinity counselling, first trimester screening results, cystic
fibrosis (CF) carrier testing, abnormal prenatal test results and pre-symptomatic testing.

1.1.4. Engaging with a diverse and complex contemporary society

To adjust to our changing and increasingly diverse society, the genetic counselling profession needs continued evolution and development of a more holistic approach that more actively encompasses cultural difference (Greeson et al., 2001; Pour-Jafari et al., 2010; Warren, 2011). The challenges of multicultural society, resulting from globalisation and migration, access to large amounts of information via the Internet and a greater understanding in the wider community about genetics, requires the development of models of genetic counselling practice that foster engagement with diversity (Abad, 2012; Liu et al., 2011; Skirton et al., 2013).

A number of models of genetic counselling practice, details on how these models can be applied and how they can facilitate measurement of outcomes, have been described in the literature (Bernhardt et al., 2000; Biesecker et al., 2001). Initially, attention focussed on the tension between Kessler’s education model (Kessler, 1997b) and the need for increased focus on psychosocial factors (including ethnocultural aspects) (Lewis, 2002; Wang, 2001; Weil, 2001). More recently, newer models have attempted to overcome the continued dominance of the education model in practice (McCarthy Veach et al., 2007; Shiloh, 2006). These models of practice focus on a more dialogical approach that takes into account the psychosocial processes involved in digesting complex genetic information, the decision making associated with new technologies and genetic testing options, and new research findings.

While research has been directed towards the development of models for improving cultural competence through genetic counselling education, this has been somewhat limited to considering cultural differences involving specific ethnic and minority groups or isolated communities. Research has yet to address the
increasing diversity within mainstream society more generally and the emergence of complex mixed cultures.

1.2. The study

At the time this research commenced in 2002 there was quite a narrow definition of cultural diversity in the literature, with a focus on visible and easily recognised characteristics such as ethnicity and religion. This narrow view prompted me to explore if and why this was held by genetic counsellors in Australasia. This has changed over the period of this study and genetic counsellors now recognise a much broader range of contributing factors to cultural diversity (Saleh et al., 2009).

In this thesis the practice of genetic counselling will be explored through the experiences and reflections of clients and genetic counsellors, using a combination of quantitative and qualitative methods. The dialogue between these two groups will be examined, with a focus on engagement with cultural difference. As responding to cultural genetic diversity through the development of specialist skills for specific ethnic groups may represent a limited approach to providing quality genetic counselling in the broader diverse and complex contemporary society, a particular subset of clients was chosen for investigation, namely those dealing with the issue of consanguinity (marriage between blood relatives). This cohort highlights the diversity and complexity of relationships in contemporary society. Despite greater openness and tolerance of different types of relationships, prejudice towards consanguineous marriage continues and counselling for this group is complex.

1.2.1. Gaps in knowledge

Research exploring genetic counselling between persons of different cultural backgrounds, especially focusing on the perspective of the client, is lacking. Greater attention needs to be focussed on the interaction between the counsellor and client because each brings their own unique cultural influences to the interaction.
My research explores both genetic counsellors’ views on cultural difference and the experiences of consanguineous couples with genetic counselling and cultural difference. By looking at a culturally embedded practice and both parties that participate in genetic counselling and the interaction between them, the following gaps in our knowledge will be addressed.

1.2.1.1. **Consanguinity**

The existing literature on genetic counselling for consanguinity focuses on communities where consanguinity is a commonly practised marriage type. Studies on consanguinity in Australia have focussed on migrant communities and their continued preference for consanguineous unions in their new country. Data published about the unusual nature of the cohort of consanguineous couples in WA (Port, 2007; Port et al., 2005) prompted my interest in the experience of being in a consanguineous marriage in a Western society, where this is an uncommon practice, and the place genetic counselling takes in this experience.

1.2.1.2. **Inside the ‘black box’ of genetic counselling**

Successful application of a more dialogical model of genetic counselling requires a better understanding of the client’s side of the dialogical equation in addition to the experiences of the counsellor. McCarthy Veach et al. (2007, p.717) have highlighted that “research on the processes and outcomes in genetic counseling is in its infancy”. Patient reported outcome measures have been developed that incorporate measures such as empowerment and have improved the ability to evaluate clinical genetics services (McAllister et al., 2008a; McAllister et al., 2011; Williams, 2009). Information on the genetic counselling experiences of discrete groups of clients is still needed.

1.2.2. **Research questions**

The research in this thesis addresses the question of how genetic counsellors have recognised and responded to the challenges of the increasing diversity and
complexity of clients they counsel. Specifically, the research addresses the following five questions:

- How do individuals in a consanguineous relationship perceive their genetic risk?
- Do consanguineous couples find genetic counselling a positive experience?
- What is the lived experience of individuals in a consanguineous relationship in a society where this is not a common marriage type?
- Do the current models of genetic counselling allow for successful engagement with cultural and linguistic diversity?
- Have genetic counsellors recognised and responded to the challenges of the increasing diversity and complexity of clients they counsel?

1.2.3. Methodology

This thesis evolved because of my clinical experience as a genetic counsellor and my curiosity about the narrow view of cultural difference prevalent amongst genetic counsellors at the time the research commenced. An awareness of my dual roles as practitioner and researcher shaped the qualitative approach that was chosen to explore the lived experience of people in consanguineous relationships in Australia and how genetic counsellors engage with consanguinity and cultural diversity.

The research was conducted in three stages. The first stage used questionnaire and interview data to explore the experiences of individuals in consanguineous relationships in Australia. The second stage used a questionnaire to examine the cultural background and work practices of genetic counsellors in Australia and New Zealand, their perception of the risks associated with cousin marriage, how they counselled these clients and whether there was variability in the information given to clients. A discussion group examined how genetic counsellors approach and adjust to the challenges of dealing with greater complexity and diversity, how they perceive, and negotiate, cultural differences between themselves and their clients.
and how this impacts on their interaction during the genetic counselling consultation. The genetic counsellors in the discussion group also considered whether their training equipped them to deal with cultural differences between counsellor and client. The third stage was a preliminary investigation of the communication and interaction between geneticists, genetic counsellors and their clients in Western Australia (WA), conducted through recording actual clinical consultations. Post-session questionnaires provided a rare insight into both client and counsellor perceptions of what had happened during the session, whether any cultural differences were perceived in a positive or negative way, whether clients’ needs were met by their counsellor and whether the counsellor felt they had met the needs of their client.

A detailed explanation of the methodology will be provided in Chapter Four.

1.3. Thesis outline

Chapter Two reviews the literature on the history and development of genetic counselling as a profession and outlines the current models of practice. The evolution from an education model emanating from the discipline of medicine, to a more client-centred model with greater emphasis on counselling is described. The latter model outlines how the cognitive and emotional background of clients impacts on their capacity to understand and apply genetic information. The evolution of these models is explored in the context of the dialogical nature of genetic counselling and trans-cultural practice, and includes the increasing awareness of the impact of cultural background of both clients and counsellors.

Chapter Three reviews the literature on consanguinity – a complex culturally embedded practice. Most cases of consanguineous marriage occur for strong socio-economic reasons and occur in societies where this type of union has been common for centuries. Although in WA there has been an increase in the number of immigrants from countries where consanguinity is common, most consanguineous
couples are not from backgrounds where these relationships are common, and are largely of northern European descent.

Chapter Four introduces consanguinity as a case study of cultural difference and explores how people in consanguineous relationships perceive their genetic risk and experience genetic counselling, and society’s reactions to their relationships. Drawing on insights from face-to-face interviews with individuals in consanguineous relationships, the chapter explores the social, family and genetic issues impacting on these couples, their experiences with genetic counselling and the role of genetic counsellors in providing support and information. Differences in knowledge and understanding of genetics and risk between consanguineous couples that have and have not had genetic counselling are investigated.

Chapter Five investigates genetic counsellors’ knowledge and experience of cultural difference and consanguinity through questionnaire and discussion group data.

Chapter Six explores the interaction between client and counsellor by examining genetic counselling consultations, in particular how different communication styles and cultural differences affect both the counsellors’ and clients’ experiences of the process. Although previous studies have investigated the content of consultations, none have looked at whether cultural differences impact on communication within an interaction. This unique study explores how cultural differences might influence conversation content and flow of information in the session.

The concluding chapter summarises the findings of this study and the implications for continued development of genetic counselling practice. The chapter makes suggestions for future research and recommendations for training and professional development. Current models of genetic counselling, the progression towards more dialogical models of practice, and the search for a model or models that successfully engage with cultural and linguistic diversity are discussed. The chapter addresses the continued dominance of educational models, the need for further development
of skills for reflective practice and dialogical engagement and the contribution of these processes towards true cultural competence.
2. Genetic counselling: models of practice

Genetic counselling has emerged to respond to the individual seeking genetic information and has taken up the challenge of how the knowledge of the genetic contribution of a disease is shared with individuals and families. However, the information is not given as a lecture, but as part of a dialogue between the individual and the counsellor. In the dialogue there is a two-way process with the counsellor and the patient mutually influencing one another. It is a human encounter and the reciprocal interactive field is an important dynamic which needs to be included in the counsellor’s analysis. (Evans, 2006, p.xvi)
2.1. Introduction

Using a broad historical perspective, this chapter outlines the shift from a teaching model of genetic counselling to one which is more dialogical and explores how increasing cultural diversity has been addressed in the theory, training and practice models of genetic counselling. The first part of the chapter outlines how the profession has evolved and defines genetic counselling. The second part focuses on current knowledge of genetic counselling, the models employed to inform practice and the different approaches used by genetic counsellors in their roles as educator, counsellor and mediator.

2.2. Genetic counselling – an historical perspective

2.2.1. Introduction

The genetic counselling profession originated as a result of advances in the field of medical genetics. While medical genetics began primarily as a diagnostic and educational specialty when therapy for most genetic disorders was absent, it is now recognised as a clinical specialty (Epstein, 2006). Scientific advances in medical genetics included the development of an increasing number of new tests, new techniques for prenatal diagnosis, an increase in the amount of information and choices available to individuals and families, and increasing public acceptance of a patient’s right to their own medical information (Harper, 2010). Interlinked with these scientific advances have been increases in the social, cultural, moral and ethical issues (da Rosa et al., 1998; Haan, 2003). The genetic counselling role developed to assist clients and their families in adjusting to the psychological, medical, financial and social aspects of being diagnosed with, or being at risk of genetic diseases (Bennett et al., 2003; Norton, 2008).

The first formal genetic counselling training program was offered by New York’s Sarah Lawrence College and the first counsellor graduated from the Masters course in 1971. This remains one of the largest and most respected training programs.
There are now 29 programs in the USA and over 12 programs worldwide (National Society of Genetic Counselors, 2009; Skirton et al., 2013), including two in Australia.

2.2.2. History of clinical genetics services in Australia and WA

The Human Genetics Society of Australasia (HGSA) was formed in 1977 to provide a forum for those dedicated to the study, investigation and practice of the various disciplines in this region. The first university-based training program in Australia began in New South Wales (NSW) in 1995 with courses later offered in Victoria (VIC) and Queensland (QLD). After completing a science or psychology undergraduate degree, students completed one year of training to obtain a Postgraduate Diploma in genetic counselling and a further year for a Master’s degree. More recently, training and certification in Australasia was reviewed and a Master’s degree is now the recommended minimum qualification (Sahhar et al., 2005). These courses are available in VIC and NSW. A special interest group of the HGSA, The Australasian Society of Genetic Counsellors (ASGC), accredits these postgraduate programs and administers a certification program through a Board of Censors comprised of elected members from each state and area of practice.

The Australia healthcare system is universal, with an optional voluntary contribution to private health insurance. Nationwide, clinical genetics services are provided by teams of specialist health professionals in tertiary hospitals, concentrated in major metropolitan centres. There is a unit each in QLD, South Australia (SA) and WA, six in VIC and five in NSW, each with supporting metropolitan and rural outreach units. Tasmania (TAS) and the Australian Capital Territory (ACT) each have a unit with visiting specialists from VIC and NSW, respectively (Human Genetics Society of Australasia, 1999). Additional units funded through private hospital boards provide clinical genetics services in VIC and QLD.

WA has a single, centralised state-wide clinical genetic service serving a population of over 2.2 million people (Australian Bureau of Statistics, 2008a). GSWA,
established in 1986, now comprises three sections, paediatrics, general (adult and prenatal) and familial cancer, plus associated screening and diagnostic laboratory services in cytogenetics, molecular genetics, biochemistry and maternal serum screening. It has clinics in two outer metropolitan hospitals and outreach clinics in five regional centres. GSWA employs six full-time equivalent (FTE) geneticists, 13 FTE genetic counsellors and additional research and administrative staff.

2.3. Genetic counselling – current practices

2.3.1. The role of a genetic counsellor

Genetic counselling is a rich and complex activity comprising a variety of interactions between genetic counsellors, clients, families and health professionals. Genetic counsellors are members of multidisciplinary genetics teams with individuals from science, clinical genetics, social work and hospital administration disciplines (Platt Walker, 1998). They work in many different settings, including hospitals, private clinics and research laboratories (National Institute of General Medical Sciences, 2006).

Genetic counsellors’ position, sandwiched between medical and scientific experts and the lay community, requires them to understand both complex scientific language and the everyday language used by the general community. In this position they play a key role in translating “gene talk” to clients (Robert, 1998, p.74). As genetic tests and information have become more complex, this role has become more important and more complex (Bennett et al., 2003).

Genetic counsellors develop skills in support, advocacy, education, counselling and co-counselling, therapy and administration (Punales-Morejon, 1997). Genetic counsellors need to be experienced in risk communication, in the facilitation of decision making and the provision of support. They are also often required to work with families and frequently discuss the implications of clients’ actions with family
members (Smets et al., 2007). Genetic counsellors are involved with clients of all ages, from pre-conception through to adulthood, for various lengths of time from one-off short-term crisis intervention to long-term relationships and with individuals or multiple family members (Bennett et al., 2003; Ciarleglio et al., 2003).

Genetic testing and diagnosis requires counselling rather than medical advice, owing to the difference between genetic information and other medical information. Genetic testing has an impact not only on the individual, but also other family members, as genes, and hence genetic information, may be shared. Testing results can impact on reproductive choices, and pre-symptomatic testing can predict whether a condition is likely to develop in the future. Psychological reactions to genetic test results and genetic information are diverse and can impact on the psyche of a client and their family, particularly if treatment is unavailable. Genetic counselling therefore involves a complex interaction of information and psychosocial issues such as anxiety, stress and coping. As Kessler notes in his review of the counselling skills required to apply the goal of nondirectiveness (ND) in genetic counselling:

> Quality genetic counselling is marked by interactions in which counsellor and clients make contact on a human level and leave the latter in a more cognitively and affectively integrated place than when their contact began. This is not to say that a professional can remove the sting of “bad news” or relieve guilt or the pain of various choices. But, at least the client can have the satisfaction of being understood in a compassionate way, of leaving with words of decency and/or hope sounding in their ears. No professional in genetic services needs to be trained in psychotherapeutic techniques to provide such quality counselling; nonetheless, the acquisition of basic skills is a necessity. (Kessler, 2001, p.189)

### 2.3.2. Genetic counselling in a complex world

Advances in the field of medical genetics and the HGP have seen an explosion of genetic knowledge and technologies. Since the inception of the HGP in 1990 and the publication of the complete human genome map in 2003, many new disease
genes have been discovered and the amount and type of genetic information available and the range of possible treatments has increased (Collins et al., 2003; Wang et al., 2004). These genes code for both single gene disorders and genetic susceptibility to complex and adult onset diseases. Genetic tests are now available for an increasing number of conditions across different life-stages, such as preimplantation genetic diagnosis (PGD), prenatal detection of birth defects and chromosome anomalies, newborn screening, carrier screening and the screening of individuals believed to be at increased risk of developing cancer (Catz et al., 2005). It will become increasingly important to consider the impact of these tests on individuals and families (Haan, 2003).

As genetic technology becomes more widespread, the availability of tests and the complexity of issues being dealt with by clients have increased, leading to greater intricacy in the genetic consultation. Of particular concern is direct-to-consumer genetic testing. A variety of genetic tests can now be arranged over the Internet with little control, regulation, quality assurance or knowledge about the science or laboratories behind the tests being offered. There is great concern about the impact of such testing on individuals who may be tested inappropriately, possibly without pre-test counselling, leaving genetic counsellors ‘to pick up the pieces’ (Hudson et al., 2009; Hudson et al., 2007; Patch et al., 2009). Another concern is testing for the hereditary breast and ovarian cancer genes (BRCA1 and BRCA2). Patents for these genes are held by a private company, which raises the issues of cost of testing, the ability of other laboratories to offer these tests, and the reduction in incentives to explore new and more efficient methods and therapeutics (Bennett et al., 2003).

Concerns about geneticisation have also been raised in relation to obstetric care and the routine use of prenatal screening. Despite recommendations that informed practitioners such as genetic counsellors should be available for pre-test consultation (O’Leary et al., 2006), prenatal tests are frequently performed without discussion of options or consequences (Hunt et al., 2005). This may lead to the need
for a definitive diagnosis via chorionic villus sampling or amniocentesis and discussions about termination of pregnancy (TOP). In genetic counselling consultations, an abnormal test would be discussed at length, explaining the nature of the condition, etiology and further testing options and implications of those decisions. In many states in the USA there is a requirement for prenatal testing options to be presented by a qualified genetic counsellor and most women of advanced maternal age are seen by a genetic counsellor for a discussion about their testing options (Hunt et al., 2005; Norton, 2008; Tapon, 2010). In the UK, pre-test counselling is recommended and generally provided by a GP or midwife, and a referral to a genetic counsellor made after the diagnosis of a chromosome abnormality (Tapon, 2010). For many clients, the referral for specialist genetic counselling often comes too late in the pregnancy to allow for prenatal diagnosis of a familial condition or the full range of diagnostic testing options to be offered. The reproductive options of these clients may then be limited as it may be too late to consider TOP. Delays in the provision of medical and psychosocial support may also cause resource and organisational challenges (Schmid et al., 2009).

The role of genetic counsellors will become increasingly important as non-communicable disease becomes more predominant and there is greater awareness of genetics among the general population. Genetic counsellors have a pivotal role in communicating and educating about genetics, and this role will be more challenging as the amount of genetic information and the complexity of available tests increases, clients demand more information and the gap between information and understanding widens. Media and Internet representation of information, underlying attitudes and health beliefs can affect how people use the knowledge gained in decision making (Catz et al., 2005; Shiloh, 1996; Shiloh et al., 1995) and behavioural response to genetic information (Lachance et al., 2010; Lea et al., 2010). The capacity of clients to understand complex health information (Lea et al.,
2010) and the way they obtain and assimilate information from the Internet (Roche et al., 2009) will influence their understanding of genetics and genomics.

2.4. Genetic counselling: practice models

2.4.1. A definition of genetic counselling

The term genetic counselling was first used by the geneticist Sheldon Reed in 1947, when he suggested replacing the previously used terms of “genetic consultation” and “genetic advice” with genetic counselling because he envisaged a form of social work rather than a purely medical encounter (Reed, 1974; Resta, 2006). Against the backdrop of the post-war eugenics debate, he argued that genetic counselling should come from a position of ethical neutrality and focus entirely on the individual interests and needs of families diagnosed with genetic conditions.

As the genetic counselling profession has evolved and genetic counsellors have obtained postgraduate qualifications, the practice has developed its own niche, focusing on diagnostics, communication and prevention (Fraser, 1974; Wertz, 1997b). In addition to counselling clients about traditional Mendelian genetic conditions and birth defects, dysmorphology, chromosomal disorders and inherited metabolic diseases, the profession now encompasses counselling about teratogenic compounds and the genetic aspects of common adult onset or ‘complex’ diseases such as heart disease, diabetes and cancer (Epstein, 2006).

As the genetic counselling role has expanded substantially since Reed’s initial conceptualisation, a more concise definition of the profession is required and new practice models have been developed. Since 1947 a range of definitions have been proposed, reflecting the movement of theoretical frameworks from the medical model, to the teaching model, the counselling model and beyond.

One of the first definitions of genetic counselling was proposed by the Ad Hoc Committee of the American Society of Human Genetics (ASHG):
Genetic counselling is a communication process that deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or the family to:

- comprehend the medical facts including the diagnosis, probable course of the disorder, and the available management,
- appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives,
- understand the alternatives for dealing with the risk of recurrence,
- choose a course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards and act in accordance with that decision,
- make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder. (Hoc, 1975, p.240).

Although it was formulated more than 30 years ago, this definition articulates three salient features of genetic counselling: that genetic counselling is a complex two way interaction; that genetic counselling is a process that takes place over time with the emphasis on the client’s autonomy; and that the impact of a genetic diagnosis is felt throughout the family (Platt Walker, 1998). Additional broad principles that are not evident in this definition but are relevant today include voluntary participation with equity of access, the importance of patient education and full information disclosure, nondirective counselling, confidentiality, privacy and attention to the psychological and social frameworks of clients. Genetic counselling is now seen more than simply transmitting scientific information, but a way of communicating that is empathetic towards the situation of the client (Platt Walker, 1998).

Despite being viewed as a modern addition to the definition of genetic counselling, the importance of psychosocial aspects of genetic counselling was recognised early in the development of the profession. Attention to the psychosocial aspects of counselling is now regarded as the key factor separating genetic counsellors from other health professionals who provide genetic counselling (Resta, 2006). More recent definitions of genetic counselling acknowledge the importance of this
psychosocial role and encompass specific practice goals that represent this expanded view of the profession: promoting understanding, achieving informed consent, facilitating decision making, reducing psychological distress, restoring feelings of perceived personal control and advancing adaptation to stressful events (Biesecker et al., 2001).

In recognition of the prime importance of the psychosocial role that genetic counsellors play, Biesecker and Peters proposed an alternative definition of the profession that emphasises the role as primarily psychological rather than medical:

> Genetic counseling is a dynamic psycho-educational process centred on genetic information. Within a therapeutic relationship established between providers and clients, clients are helped to personalise technical and probabilistic genetic information, to promote self-determination and to enhance their ability to adapt over time. The goal is to facilitate clients’ ability to use genetic information in a personally meaningful way that minimises psychological distress and increases personal control. (Biesecker et al., 2001, p.194)

In 2003, in response to genetic counsellors’ concern that the ASHG definition of genetic counselling was too wordy, a new definition was proposed by the National Society of Genetic Counselors (NSGC):

> Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:
> - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
> - Education about inheritance, testing, management, prevention, resources and research.
> - Counseling to promote informed choices and adaptation to the risk or condition. (Resta et al., 2006 p.79)

This definition was formulated over many months of discussion and peer review with relevant interest groups such as the NSGC membership, NSGC legal counsel, leaders of several national genetic advocacy groups and the NSGC Genetic
Counseling Definition Task Force. The UK and Australia subsequently adopted the NSGC definition (Resta, 2006).

Psychology and counselling theory are now well represented in the genetic counselling literature and practice (Evans, 2006; Weil et al., 2006). It has been suggested that the profession would benefit from a comparable set of contributions from counselling, social work, health psychology, sociology and other allied health professions that have parallels to genetic counselling (Shiloh, 2006; Weil, 2003). Theories of risk, rational and emotional decision making, stress and conflict are all relevant to genetic counselling and raise research questions closely linked to health psychology research and practice (Shiloh, 2006; Shiloh et al., 2006b).

Despite increased recognition of the psychosocial role of genetic counsellors, no single definition or theoretical framework has been developed for genetic counselling that encompasses the diversity of issues and situations faced by practitioners (Eunpu, 1997; Weil, 2003). It is recognised that a more flexible framework is required to allow counsellors to apply their interactive skills-based counselling techniques to the different settings in which they practice (Weil et al., 2006). Whilst the models and approaches that have been applied in the past have had value, it is important to continue to challenge assumptions, augment current practice models and improve understanding of genetic counselling through continued research.

In 2001, an issue of the American Journal of Human Genetics reviewed the evolution of genetic counselling and argued the profession needed to be less introspective and more responsive to the challenges of the rapidly changing and expanding field. The challenges identified included the vast expansion of genetic knowledge and its introduction into mainstream medicine, the growth of genetic advocacy groups, and an interest in techniques and practices that made the profession responsive to growing ethnic diversity. It was noted that genetic counselling research was
becoming process focussed and the consumers of genetic counselling services should be at the centre of these process studies (Baty et al., 2001). Studies examining client perceptions and what they find helpful about genetic counselling were seen as useful in giving client feedback enabling genetic counsellors to modify their practice to address the needs of individual clients more effectively (Evans et al., 2004; McCarthy Veach et al., 1999).

Several years later, Seminars in Medical Genetics addressed the rapid developments in both the practice of, and research on, the field of genetic counselling (Baty et al., 2006). In this issue of the journal it was noted that evidence-based health care was developing, that the roles of geneticists and genetic counsellors in health care provision would continue to evolve, and that the profession would mature beyond service provision into an academic discipline (Baty et al., 2006; Epstein, 2006).

2.4.2. Medical model of genetic counselling

Genetic counselling continues to be located in the broader medical system, within medical genetics, and thus tends to be dominated by the medical model. In the medical system, the client is treated as a patient with a medical problem to be diagnosed and treated. In contrast, the counselling model works with clients to address issues and develop responses together (Williams et al., 1998). Theorists and researchers continue to develop models for genetic counselling that incorporate both medicine and counselling frameworks.

2.4.3. Teaching and counselling models of genetic counselling

The two main functions that define genetic counselling include education (the transmission of information) and psychological counselling (support in reaching decisions about options) (Fraser, 1974). The teaching model was adopted early in the development of the profession in university academic departments, and counselling models were adopted later when non-physician practitioners
commenced practice (Kessler, 1997b). The basic assumptions and goals of these two models are compared in Table 2.1.

**Table 2.1 Comparison of the teaching and counselling models of genetic counselling**
Adapted from Kessler (1997b)

<table>
<thead>
<tr>
<th>Teaching model</th>
<th>Counselling model</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Goal(s)</strong></td>
<td></td>
</tr>
<tr>
<td>Educate counsees</td>
<td>Understand the client</td>
</tr>
<tr>
<td></td>
<td>Bolster inner sense of competence</td>
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<tr>
<td></td>
<td>Promote a greater sense of control</td>
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<tr>
<td></td>
<td>Relieve psychological distress</td>
</tr>
<tr>
<td></td>
<td>Support and raise self-esteem</td>
</tr>
<tr>
<td></td>
<td>Help find solutions to specific problems</td>
</tr>
<tr>
<td><strong>Basis</strong></td>
<td></td>
</tr>
<tr>
<td>Clients come for information</td>
<td>Clients come for counselling for complex reasons</td>
</tr>
<tr>
<td><strong>Assumptions</strong></td>
<td></td>
</tr>
<tr>
<td>Informed clients should be able to make their own decisions</td>
<td>Complex assumptions about human behaviour and psychology</td>
</tr>
<tr>
<td><strong>Counselling task</strong></td>
<td></td>
</tr>
<tr>
<td>Correct misinformation</td>
<td>Complex and requires:</td>
</tr>
<tr>
<td>Provide impartial and balanced information</td>
<td>Assessment of client’s needs limitations, strengths, values and decision trends</td>
</tr>
<tr>
<td></td>
<td>Range of counselling skills</td>
</tr>
<tr>
<td></td>
<td>Individualised counselling styles to suit client’s needs and agendas</td>
</tr>
<tr>
<td></td>
<td>Counsellor attends to own inner life</td>
</tr>
<tr>
<td><strong>Education</strong></td>
<td></td>
</tr>
<tr>
<td>An end in itself</td>
<td>Used as a means to achieve goals</td>
</tr>
<tr>
<td><strong>Relationship with client</strong></td>
<td></td>
</tr>
<tr>
<td>Based on authority</td>
<td>Aims for mutuality</td>
</tr>
</tbody>
</table>

Kessler (1997b) challenged the genetic counselling profession to combine the education and counselling models, and apply both to practice. While Kessler recognised the simultaneous application of these two basic models would be difficult because they had more differences than similarities and their dual application required skills specific to the unique issues that arise in genetic counselling (Kessler, 1997a, 1997b, 1997d), he argued that not enough attention had been given to training genetic counsellors in combining these two models:

The skill demands of the teaching model tend to reinforce attitudes and procedures antithetical to those required in counseling. This may be confusing to students and
interfere with their ability to perfect either pedagogical or counseling skills. As working professionals, many find themselves relying heavily on a teaching approach; further reinforcing behaviours that interfere with the development of the counseling skills needed to help clients reach decisions, deal with couple’s conflicts, manage guilt and shame, provide empathy, and, in general, maintain a nondirective stance. (Kessler, 1997b, p.294)

Evidence in the literature, in addition to my own experience in practice, suggests the predominant mode of practice continues to be the teaching model, and tension remains in applying both models in practice (Butow et al., 2004; Ellington et al., 2006; Kessler, 2000b; Meiser et al., 2008; Pieterse et al., 2005; Roter et al., 2006). Biesecker believes the teaching model predominates because of the influence of the medical model, the priorities in health care delivery and the status of counsellors within the medical genetics team, rather than the constraints of ND or the abilities, training, values, or attitudes of counsellors (Biesecker, 2003). Other reasons include time restrictions in the clinic, assumptions about what clients want to hear and counsellors’ discomfort around dealing with emotional issues (Kessler, 1997a; Michie et al., 1999).

Kessler (1997b) proposed combining the teaching and counselling models to allow information and options to be presented in a value neutral and nondirective fashion. However, questions as to whether they can successfully be applied simultaneously have arisen, as have concerns about validity and inequity in the services received by culturally diverse groups (Kessler, 1997c; Macleod et al., 2002). It has been argued that major limitations in both models mean they fail to recognise and address multicultural issues and while the counselling model may be more supportive of the goals of genetic counselling practice, research into the effectiveness of both models for this profession is limited (Lewis, 2002).
2.4.4. Influence of psychological theories on genetic counselling models

Current genetic counselling training tends to be based on Neo-Freudian and Rogerian theory framed as crisis intervention (Fine et al., 1993; Marks, 2004; McCarthy Veach et al., 2007). In crisis intervention using the psychological framework, counsellors explore clients’ reactions to difficult situations, helping them to clarify values presumed to underlie the feelings they express, using a nondirective approach and support clients to make informed decisions (Hodgson et al., 2005; White, 1999).

The realisation that genetic counselling deals with humans with thoughts and feelings and thus behavioural and psychological states are being modified, has led to a shift towards this ‘psychological paradigm’. Genetic counselling now incorporates aspects of the medical model and preventative medicine as well as the psychological issues that pervade all aspects of genetic counselling including education, communication, emotional support and decision making (Ellington et al., 2011; Kessler, 1997b).

Placing the focus on the concerns of the client being heard enables genetic counselling to become a dialogue between individuals. Learning about an inherited condition is then linked to the psychological processes of coping, and takes place within a framework of personal encounter in which emotional reactions become interwoven with the rational discussion of science (da Rosa et al., 1998; Weil, 2003). This can be extended to encompass the personal experiences of each client owing to cultural differences.

Effective genetic counselling may be enhanced by the employment of a number of different theoretical tenets and techniques, dependent on the practice setting and individual clients’ and families’ circumstances. Genetic counsellors have employed short-term or solution-focussed therapies, have modelled good communication techniques to assist couples having difficulties with decision making, have used
family systems theory and have introduced behaviour-based interventions to assist clients who experience anxiety (Eunpu, 1997; Kenen et al., 1995).

While other theoretical models, such as psychoanalytic theory, may not be suitable for short-term and outcome-focused genetic counselling interactions, this does not preclude these models from being incorporated into practice. Aspects of Rogerian theory, such as genuineness, empathy, unconditional positive regard, and ND are widely used in genetic counselling practice (Eunpu, 1997).

Despite general agreement that attention to psychological and emotional issues is an integral part of practice, resistance to integration of psychotherapeutic theory into genetic counselling practice remains. A survey of 1346 American genetic counsellors in 1995 showed that only 6% were interested in psychotherapeutic interventions in genetic counselling (Eunpu, 1997). Proposed reasons for this lack of interest included limited focus on this area in training, constraints of operating within the medical model, and the increasing complexity of genetics and testing requiring more time be spent providing information.

2.4.5. ND as the central tenet of genetic counselling

One of the founding principles of Rogerian theory is ND. Genetic counselling was one of the first medical areas to adopt a nondirective philosophy and this has been the central tenet of the profession for many years, with other important tenets being autonomy and non-coercion (Weil, 2003). The nondirective approach arose out of concern that genetic counselling may interfere with the decision making of clients. The ND tenet was introduced in the late 1940s in an attempt to separate the profession from the eugenics movement. The focus became public health and disease prevention through a reduction in the number of affected individuals in high-risk families (Resta, 2006).

Nondirective approaches are at the opposite end of the continuum to the medical model, where doctors tend to tell patients what to do and advise them on particular
treatments or courses of action. In contrast, genetic counsellors inform clients about the options available to them and equip them with decision making tools, often to be used under great duress and in times of crisis. Clients have been shown to react differently to this nondirective approach (Shiloh et al., 1995). Many expected a directive, others tried to guess the counsellor’s opinions, and some took neutrality to mean the information being offered was serious.

2.4.5.1. Criticisms of ND

Kessler (2001) has argued that a lack of skills in nondirective counselling hindered the effective application of this form of counselling. While he agreed that some counsellors were skilled in this area, he described others as “conveying little empathy, not responding to clients to indicate understanding, having difficulty hearing beyond the literal level, sometimes saying hurtful or thoughtless things and lacking flexibility to shift between the education and counselling role” (p.189). Kessler advocated the development of more active and effective counselling skills rather than continued discussion of ND. He proposed a minimum number of hours of counselling supervision, the introduction of videotapes and audiotapes in counselling sessions and in supervision work, and argued that this would facilitate a change from the passive role of genetic counsellor to that of facilitator in informed decision making (Kessler, 2001). These suggestions are now included in modern Masters training programs and Australian certification requirements (Dunlop et al., 2011; Sahhar et al., 2005; Sexton et al., 2012).

The importance of the ND tenet to genetic counselling continues to be challenged because it is thought to insufficiently describe genetic counselling and lack defining theoretical principles (Biesecker, 2003; McCarthy Veach et al., 2002a; Weil, 2003). Many question whether ND is even possible, arguing that even balanced information cannot be value free or neutral. Others argue that the principle of respecting autonomous decision making is valid, but providing information in a nondirective manner fails to recognise the human interactions involved in
counselling (White, 1999). As genetic information is complex and there is a range of different circumstances in which it is delivered, and a choice involved in what to information to include, it is argued that genetic counselling cannot be completely neutral or value free and another broader approach is needed (Lippman et al., 1992; Rehmann-Sutter, 2009).

A fresh look at directiveness and ND emerged as the complexity of genetic issues increased, and genetic counsellors struggled to remain neutral while continuing to satisfy the needs of clients, employers and other stakeholders (Kessler, 2001). The debate moved towards accurately defining genetic counselling practice and addressing the questions to maximise effectiveness, including what actually occurs in a consultation and the role of the genetic counsellor (Evans, 2006).

There has been a growing awareness of the fact that all genetic counsellors bring to each situation a set of values, beliefs and ideas about a topic and whilst there may be a desire to be nondirective, embedded cultural values may not make this possible (Oduncu, 2002; Raz & Atar, 2003; Rentmeester, 2001; Suter, 1998). Many counsellors have acknowledged that remaining neutral is a difficult task and at some stage have encountered situations where they felt justified in attempting to influence the decision of a client (Rapp, 2000).

The sociologist Rayna Rapp (2000, p.59) argues that the issue of value neutrality is complex when much of the technology surrounding genetic counselling is “explicitly developed to identify and eliminate foetuses with problem-causing chromosomes (and increasingly genes).” She states that while geneticists argue that they present information and options in a neutral way, the very existence of the technology to screen and terminate a pregnancy makes this consultation anything but neutral. She believes that genetic counsellors are trained to present value-charged information and technology in a neutral way.
Studies by Hunter et al. (2005) and Shiloh and Sagi (1989) found that the subtle influence of counsellors’ values may also have some effect on clients’ choices. In addition, these studies found the way that risk is presented, and who delivers the information (genetic counsellor or obstetrician) affects perception and the choice of options. Further research has been recommended to assess communication, counselling processes and the family, in order to determine the effect that genetic knowledge has on self-concept and family relationships (Burgess et al., 1998; Shiloh et al., 1995).

Perhaps of greater significance is how values and opinions impact on the ability to effectively counsel clients. It is important that through training and ongoing professional supervision genetic counsellors become aware of their beliefs and how they have arisen, and aware of how non-verbal cues (such as mannerisms) might inadvertently convey a message. An understanding of genetic counsellors’ values and how they might impact on the provision of information and options is also important for educators and supervisors engaged in genetic counselling recruitment and training (Pirzadeh et al., 2007).

2.4.5.2. Alternatives to ND

In addition to arguing that many genetic counsellors lack ND skills, Kessler also argues that the narrow and unclear definition of ND inhibits the ability of genetic counsellors to perform their main function: to effectively assist their clients. In 1996 he proposed a broader definition of ND to better reflect the guiding ethos of genetic counselling:

There has been a second aspect of nondirective methods that practitioners recognized, namely their ability to promote the autonomous functioning of the client. This aspect of ND is clearly applicable to genetic counselling and I offer it as a definition: ND describes procedures aimed at promoting the autonomy and self-directedness of the client. (Kessler, 1997d, p.166)
Others agree with Kessler’s views. McCarthy Veach et al. (2002a, p.188) suggests that “Rogerian nondirectiveness is both the profession’s greatest asset and its greatest liability” and that surrendering this would allow for the development of a framework that enables a more proactive position. Weil proposes that “nondirectiveness be replaced as the guiding principle for the clinical practice and professional conceptualization of genetic counselling” (Weil, 2003, p.204). He has suggested the removal of the term rather than the concept, as the underlying principles of providing balanced information and not imposing counsellors’ views are valuable and support client autonomy. He goes on to propose as a guiding principle that “the central ethos of genetic counseling should be to bring the psychosocial component into every aspect of the work” (Weil, 2003, p.207).

Weil’s (2003) proposed change to the central tenet of genetic counselling embodies the fundamental role of genetic counselling, which is to help people to adjust to the many issues that arise after the diagnosis of a genetic disease, all of which are in the psychosocial domain. He argued that technological advances create pressure on genetic counsellors to provide information and that this new central tenet would give weight to the argument for resources to promote the difficult task of attending to the other equally important aspects of genetic counselling. He also pointed out that psychosocial issues must include ethnocultural issues. In practice, Weil recommended adding the psychological aspects of the genetic counselling model to the medical model, and moving towards a dialogical approach to counselling. He suggests that by applying both models, genetic counsellors could act as catalysts for change, moving away from the medical model towards a new holistic multidisciplinary approach that cares for the client and the family as a whole.

Biesecker (2003), in her commentary on Weil’s proposal to change the central tenet of genetic counselling to always include psychosocial aspects, observed that Reed (1974) and Kessler (1980) had made similar proposals in their early writings on genetic counselling. She noted that in order to shift away from the teaching model
and fully attend to the “affective and cognitive meaning of genetic information and their [client] lived experience”, the medical system within which genetic counsellors operate needs to better recognise and value the time and effort required to focus on psychosocial issues (Biesecker, 2003, p.215). Evidence that the teaching model continues to be more prevalent than the counselling model, the reliance on other health care models, a lack of studies providing information on what actually occurs during genetic counselling and a lack of evidence regarding the effectiveness (or otherwise) of interventions has resulted in calls for the profession to develop its own model (or models) and standards (Biesecker, 2001, 2003; Kessler, 2000b; McCarthy Veach et al., 2002a). This needs to take into account the requirements of genetic counselling for both education and psychosocial support and the needs of each individual client.

The issue of ND in genetic counselling was the focus of discussions at the 2003 Annual Meeting of the NSGC. The general consensus was that the profession needed to clarify what genetic counsellors do, describe current model(s) of practice and develop a more proactive model of genetic counselling:

- giving more attention to the wants and needs of the counselees, creating a comfortable setting within which counselees can use as many of their faculties as possible, facilitating active decision making, implementing meaningful informed consent, and identifying and processing genetic counselor’s personal and professional values. (Weil et al., 2006, p.90)

Resta (2006) argues that these developments require ongoing research and development of the definitions and descriptive models by genetic counsellors themselves. A number of theories and models such as the self-regulation theory (Shiloh, 2006), the shared decision making model (Elwyn et al., 2000) and the reciprocal-engagement model (REM) (McCarthy Veach et al., 2007) have been developed in response to calls for models that go beyond ND. These models incorporate psychological and decision making support and address some of the
concerns raised by clients about receiving a more personalised service. They also address issues of culture and diversity, allowing the incorporation of individual and cultural perceptions and beliefs about a genetic condition by focussing on the psychological processes involved in genetic counselling.

2.4.6. The REM

In the first step towards developing an empirically established, comprehensive model for genetic counselling, Veach and colleagues convened a consensus meeting of the Directors of US Graduate Genetic Counselling programs in 2005. The meeting objectives were to describe the clinical practice model that currently existed and in so doing describe the tenets, goals, strategies and behaviours used to address the concerns of clients and identify any variations due to specialities, different genetic conditions or cultural backgrounds. From this they developed the REM of genetic counselling practice (Figure 2.1). This was the first model to articulate a psychosocial basis for exploring genetic information.

The basis of the REM is the assumption of reciprocity, with each element of the model complementing each other and engagement of counsellor and client in a dialogical exchange. Veach described the model as a:

mutual process in which the genetic counsellor and patient participate in an educational exchange of genetic and biomedical information shaped by their unique psychosocial identities. The genetic counsellor-patient relationship is the medium in which these activities occur. The elements of this model are not discrete, they reciprocally affect each other, and each is necessary but not sufficient individually for influencing genetic counselling outcomes. (McCarthy Veach et al., 2007 p.726)
2.5. Diversity in genetic counselling practice

2.5.1. Individual diversity

Genetic counselling is no longer viewed as a process of communication about science but more correctly as an interaction between human beings with thoughts and feelings (Abrams et al., 2002). It is a complex bi-directional process whereby the counsellor and the client explore issues relating to an individual’s or family’s experience with inherited conditions. Although general principles and procedures are followed and certain standard information shared, interactions between a counsellor and client are never identical. Each client and each genetic counsellor
brings with them to each counselling session a unique set of experiences, cultural and social background, understanding and emotions, psychological issues, thoughts and interpretations (Wang, 2001).

Genetic counselling aims to allow clients to use genetic information in a personally meaningful way, but because human variation is vast, individuals experience birth, death, reproductive decisions, chronic illness, and living at risk in different ways. In addition to personal experience and background, there is variation in understanding with genetic health professionals (GHPs) and their patients holding different personal meanings of illnesses and inheritance and different language that they use to describe them. This can cause miscommunication and greatly affect the relationship and quality of care. Medical and genetic information, particularly regarding statistical risks and probability, is often misunderstood or misinterpreted (Austin, 2010; Fransen et al., 2006).

Shiloh (2006) believes it is important that genetic counsellors understand the personal meaning of life changing events and the information that is relevant to each client in order to minimise miscommunication. Rapp agrees that the contribution of individual variation to the consultation is important. She states:

> Each client brings...the light and shadow of her personal biography, family history, and community resources with her to the consulting room, and she hears about new interventions into her aspirations for her own and her child’s futures through these filters. Thus, a deluge of difference and a mosaic of meanings await a genetic counsellor each time she meets a new client. (Rapp, 2000, p.77)

Adopting the concepts contained in the newer models of genetic counselling practice moves it towards addressing the challenges of diversity. In particular, the REM incorporates both counsellor and client in the interaction, provides the basis for taking into account diversity, extends the dialogical process, and thereby allows practitioners to move beyond the teaching model (McCarthy Veach et al., 2012).
The application of models to practice naturally requires a pragmatic approach and therefore genetic counsellors need to be willing and able to apply cross-cultural models of practice. Genetic counsellors are trained to manage difficult situations in an empathic and sensitive manner, and frequently encounter challenges in their attempts to provide relevant information and support to their clients (Alliman et al., 2009; Marks, 2004). Client autonomy is promoted and each client is treated as an individual. Genetic counsellors are often experienced in overcoming language and communication barriers through the use of interpreters, maintaining the central tenet of ND against cultural demands for advice, resistant to institutional and societal pressures to implement screening programs (Raz & Atar, 2003) and can effectively manage the challenge of different cultural constructs of beliefs about genetics and inheritance (Eisenbruch et al., 2004).

Lee and colleagues (2009) noted, however, that greater multicultural awareness and knowledge is required before improvements in genetic counselling practice can be achieved. Genetic counsellors need to adopt a wider concept of sources of difference in order to meet future demands of increasing cultural diversity. The next two sections of this chapter explore possible changes to theory and practice that may further enhance the dialogical approach to genetic counselling.

2.5.2. Cultural diversity

2.5.2.1. Challenges

Ethnocultural issues present new challenges and opportunities for genetic counsellors. Exploring these challenges has the potential to help understand diversity and the different techniques needed to manage diversity within genetic counselling sessions. Many health professions (including mental health, public health, psychology, health education, social work) and ethnocultural studies have explored the issues and could provide useful approaches, knowledge, research and opportunities for collaboration (Lee et al., 2009; Weil et al., 2006).
Culture can be explained as “the ways in which people make sense of the world by deploying shared meanings, attitudes, assumptions and values” (Irvine et al., 2002, p.175). Culture is distinct from ethnicity, although the two terms are often used interchangeably. Ethnicity relates more to common characteristics such as physical traits, religion, history and common ancestry (Greb, 1998). Cultural systems are the key influence over how we respond and behave in the world around us. They can be explicit, apparent and tangible or implicit, invisible and intangible. They can also change over time (Greb, 1998; Schott et al., 1996). According to Schott and colleagues, culture is embedded in every aspect of our lives:

Our culture vitally affects every aspect of our daily life, how we live, think and behave and how we view and analyse the world. But because, like the air we breathe, our culture is all around us from the day of our birth, and because we acquire almost all of it unconsciously early in childhood, most of us grow up unaware that we have a culture at all. (Schott et al. (1996, p.3)

Weil (2001) observed that culture arises from many sources other than geographical or ethnic factors, including gender, training, occupation, sexual orientation, physical or intellectual disability, shared history, lifestyle, life world experience, family situations and religious beliefs. He used the term “ethnocultural” to describe this broader view and pointed out that mixed contributions of a particular identity may mean that individuals have multiple or complex ethnocultural identities. As a result, attention must be given to the bidirectional and interactional aspects of each counselling session:

It is essential that cross-cultural genetic counselling be thought of in terms of a two way interaction between the ethnicity and culture of the genetic counsellor, including the institutions and profession to which he or she belongs, and those of the counselee(s). (Weil, 2001, p.144)

Rapidly increasing cultural diversity, increasing global migration, and differences between the clients’ and genetic counsellors’ perspectives on science, life worlds,
reproductive practices and world view mean that genetic counsellors frequently encounter professional challenges due to cultural diversity (Alliman et al., 2009). Genetic counsellors also face the challenge of applying models of practice that promote engagement with diversity.

2.5.2.2. Cultural diversity within the genetic counselling profession

There is little published data on the cultural diversity of GHPs in Australia, the UK or Europe. In Australia approximately 25.3% of allied health workers are born outside Australia and males currently comprise less than 2% of all genetic counsellors in Australia (Australian Institute of Health and Welfare, 2009; James et al., 2003). However, it is not known how well this data reflects the diversity of genetic counsellors nor is it known how diverse this group is, as the data is not broken down by birthplace.

In the USA, a range of studies have been undertaken into the cultural diversity of the North American genetic counselling profession. In 2008, an NSGC Professional Standards Survey found that 93% of members were Caucasian, 5% were Asian and 1% were Black or Hispanic, and 95% of the group were women (Parrot et al., 2008). Twelve genetic counsellors reported that they had a disability but did not identify themselves as part of a specific disability community (Smith et al., 2009).

Similar to other health and science professions in the USA, minority groups are under-represented in genetic counselling (Smith et al., 1993). Suggested reasons for the lack of diversity in the genetic counselling field include a lack of role models within the health professions, social vulnerability and isolation within minority groups, lack of professional appeal due to low salary, and perceived lack of value of the profession to underserved minorities (Barragan, 2009; Mittman et al., 2008). Minority groups are generally less aware of genetic counselling as a career, and see tuition costs as a barrier to entering the profession (Oh et al., 2005; Schoonveld et al., 2007; Smith et al., 1993).
Large numbers of initiatives to increase diversity within the profession have been launched but these have had limited success. They have been described as being “disjointed, sporadic, and variously lack (sic) realistic goals and effective implementation and evaluation strategies” (Mittman et al., 2008, p.309).

Recognition of the problem and efforts to increase diversity has failed to alter the homogeneity within the profession. In the USA, genetic counselling has the lowest participation of African and Hispanic Americans in the health professions, despite these groups representing almost one-third of the population (Mittman et al., 2008; Schneider et al., 1998; Smith et al., 1993).

It has been argued that the genetic counselling profession in the USA should more accurately reflect the American demographic and to achieve this more counsellors should be recruited into training from under-represented minority (URM) groups. Despite this, a survey of students enrolled in USA Masters programs conducted in 2004 shows that trainees are not representative of the broader population, with 97% female, 87% Caucasian and only 13% identifying themselves as an ethnic minority (Lega et al., 2005). URMs in this sample included Native Americans, African–Americans and Hispanics (Lega et al., 2005).

Genetic counsellors recruited from minority groups report numerous difficulties during their training and early practice, particularly in regard to adjusting to their peer groups and concern that their ethnicity or minority background may have been the reason they were accepted for training (Lega et al., 2005; Schoonveld et al., 2007). While Schoonveld et al. (2007) continue to recommend recruiting students from URMs, they maintain that all students need to engage in a mutual acculturation process, irrespective of whether they come from a URM or not.

Studies of early graduates of genetic counsellor training programs in the USA have shown that these graduates tend to be middle class women returning to the workforce after having children. Gender stereotypes persist within this group, with
older female genetic counsellors displaying ambivalence about the capacities and limits of men in the profession (Rapp, 2000). Schoonveld et al. (2007) note, with some concern, that there is a perception that because the profession was founded by females and dominated by females, the term genetic counsellor implies a female career choice based on principles that appeal more to women.

While there is some evidence to suggest that female clients feel that their questions are addressed more adequately by female counsellors (Zare et al., 1984), other evidence suggests the communication between counsellor and client is largely independent of the gender of the provider (Kessler, 1989). Both counsellors and patients consider the nature of the interpersonal interaction to be extremely important and contributing to the therapeutic nature of the relationship (Bernhardt et al., 2000). Whilst there is a focus on enhancing ethnic and racial diversity in the profession, there is also recognition that each individual contributes to diversity as everyone is different (Lega et al., 2005; Schoonveld et al., 2007; Smith et al., 1993). This suggests that the gender or ethnic background of counsellors may not be as important as the individual qualities they bring to a counselling session.

In the North American literature on health care provision to ethnic minorities, those who argue for greater diversity to reduce disparity between the health of the majority and minority groups use fairness and function arguments to back their case. Fairness arguments refer to the historical exclusion of minority groups from educational, economic and professional opportunities. Function arguments suggest that increased diversity in the health care workforce encourages better access by, and professional relationships with, minority groups (Schoonveld et al., 2007).

These ‘form and fairness’ arguments may apply to the broader health care field but in relation to genetic counselling there are no published data to suggest that genetic counsellors from the URMs are more likely to practice in URM communities. Nor is there data to suggest URMs may have more effective counselling
relationships with clients from the same background, or that Caucasian female genetic counsellors have problems providing quality services to underserved populations (Schoonveld et al., 2007). There is a suggestion that some underserved clients might fear or mistrust the notion of genetic counselling due to an historical fear of eugenics but there are no data indicating a preference for a genetic counsellor of the same race (Laskey et al., 2003; Oh et al., 2005). Furthermore, fairness arguments addressing universal access may not be relevant in Australia, as genetic counselling services are located within public hospitals and almost entirely funded by the government. In the Australian system, affordability is not an issue (Sahhar et al., 2005).

In addition to form and fairness arguments, there is a significant body of literature that argues for increasing diversity within the workforce on the basis that diversity “enriches the educational experience of all as it challenges stereotypes, enhances cultural competence and fosters lasting relationships” and has the potential to reduce inequalities of access to services for minority groups (Mittman et al., 2008, p.302). Whilst I agree that the profession should reflect the ethnic, cultural and gender diversity of the wider community, I present an alternative view that in addition to continuing efforts to increase diversity, improved training opportunities for existing counsellors and students would have more immediate benefits for clients, counsellors and the profession. Developing all genetic counsellors’ skills to enable more effective engagement with diversity could overcome the deficit due to the lack of URMs within the profession. In addition to human genetics knowledge and counselling skills, the range of skills in a genetic counsellor’s tool kit could be expanded to include the ability to apply skills tailored to the individual client in an ethical and culturally sensitive manner, irrespective of the cultural background of either client or counsellor (Pour-Jafari et al., 2010; Tse et al., 2013).
2.5.2.3. **How genetic counsellors manage diversity**

In their commentary on the challenge of responding to an increasingly pluralistic society and engaging with cultural diversity, Irvine and McPhee (2002) recognised the need to understand and respect cultural and ethical pluralism. However, they also acknowledge the difficulties of incorporating understanding and respect into practice when resources are limited. Other authors believe that individuals fit within a cultural framework and genetic counsellors can learn about individual clients by learning about the group they come from. It is impractical to expect to specialise in counselling specific ethnic and cultural groups. Attempts to understand racial and ethnic variation, given the number and complexity of these groups today, may risk bringing into play cultural stereotypes (Hunt, 2005; Hunt et al., 2008).

The ability to speak the client’s first language may be an advantage and some might prefer a health care provider who is of a similar background. However, culture and language have often been melded together inappropriately. In a study of genetic counsellors working with Spanish-speakers in New York, Rapp (2000) found that learning Spanish helped communication initially but ultimately created more problems than it solved because clients incorrectly assumed the counsellors were from the same culture as them and presumed they understood more about their culture than they did.

Every counselling session is unique. During every counselling interaction, both the client and genetic counsellor bring their own values, ethical systems and cultural background, which affects the way medical information is delivered by the counsellor and assimilated into the client’s cognitive framework (McCarthy Veach et al., 2007; Shiloh, 2006; Weil, 2001). A genetic counselling model is needed that takes into account this uniqueness. A model in which the communication process is more dialogical and genetic counsellors themselves become more culturally reflective, working more in conjunction with clients to address their specific and individual cultural and value issues that emerge in consultation.
Training programs moved from a mono-cultural to a multi-cultural perspective when culturally appropriate genetic counselling was first recognised as necessary to cater for the growing population of immigrants and members of different ethnic groups (Weil et al., 1993). The NSGC Code of Ethics was also amended to include a statement that “genetic counselors should strive to respect their client’s beliefs, cultural traditions, inclinations, circumstances, and feelings” (National Society of Genetic Counselors, 2006, p.310). In Australia, research highlighted the ongoing need for professional development and broader policy development to better prepare genetics practitioners for cultural diversity (Saleh et al., 2009). More recently, Warren (2011) developed the Genetic Counseling Cultural Competence Toolkit which is an on-line resource available to all genetic counsellors for training and professional development.

In the same way that genetic counselling practice drew on, and evolved through, various models of practice from other disciplines, it has also drawn on other disciplines for models of multicultural practice. In the following section I will look at the historical progression through these models towards models such as the REM which deals with cultural diversity in a much more collaborative fashion and reflects the increasing awareness of diversity and its attendant issues. Combining aspects of other models with the REM may enable movement towards a truly comprehensive dialogical model that engages fully with diversity.

2.5.2.4. Cross-cultural, multicultural and transcultural genetic counselling

In the early 1990s, Wang and Marsh found that the ethical principles guiding the norms of genetic counselling and how they interact with values or preferences of the different ethnocultural groups in the USA were poorly understood (Wang et al., 1992). Sue and Sue (1990) also found that working with clients of different cultural backgrounds presented a number of unique problems related to issues such as nondirective counselling, informed consent and confidentiality. When these issues are interpreted from two different perspectives (the Western medicine perspective
of the counsellor and the specific ethnocultural perspective of the client) there is potential for a cultural clash.

In the mid-1990s, genetic counselling training began to shift away from culture-specific information provided by experts from different cultural groups and other academic fields. This shift was driven by Wang in a handbook of cross-cultural genetic counselling. The handbook aimed to:

Provide knowledge of cultural concepts and issues, examine issues of self-awareness and transference/countertransference, awareness within the context of group dynamics, and the acquisition of genetic counseling skills within a cultural context. (Wang, 1994, pp.272-3)

Later, evaluation of the effectiveness of the handbook found it was an effective tool for increasing multicultural counselling competence, regardless of when it was used in a training program (Wang, 1998). Students reported an increased awareness of multicultural counselling issues and requested that more time be allocated to training in this area. Wang recommended research to further examine the efficacy of this handbook and determine whether students’ improvement in competence was sustained or changed over time.

Techniques for teaching multicultural competence were further developed by Wang (2001), who wrote of the movement from cross-cultural to multicultural genetic counselling over the past 25 years. She described the past focus of cross-cultural counselling on identifying visible differences and the acquisition of knowledge about specific racial cultural groups in order to attain increased cultural competency. Wang believed that racial and cultural issues were obstacles for people seeking genetic counselling, and that simplistic knowledge resulted in group stereotypes. Wang stated that the literature was “replete with information about specific cultural group norms”, including the need for racial immersion and recruitment of students from diverse backgrounds. She questioned whether this
“had been effective in serving underserved visible racial-cultural groups” and concluded that this approach marginalised these groups and stereotypes ignored individual variation (Wang, 2001, pp.210-211). Historically, genetic counselling training has focussed on the ‘other’ and ignored the counsellor’s own beliefs, values and worldview. More recently, the move from cross-cultural to multicultural genetic counselling has shifted learning from between-group differences to within-group differences, allowing counsellors to develop the ability to understand their own cultural background and respectfully relate to people from all racial-cultural groups (Wang, 2001).

However, along with recognition of the need to change approaches to training has been recognition of the increasing challenge of providing accessible and useful genetic counselling services to an increasingly wide diversity of clients:

From an ethical and moral perspective, the goal of providing equal access and quality of services for all individuals requires that genetic counselors be sensitive, knowledgeable and skilled in working with individuals from diverse cultures and ethnic communities. This is particularly important since genetic counseling addresses issues such as reproduction sexuality, health, disease, and disability, concerning which individuals and group perceptions, beliefs, and practices are fundamentally influenced by culture. (Weil, 2001, p.143)

Weil (2001) recommended improving cross-cultural genetic counselling techniques through changes in education, training and practice (Table 2.2). He also recommended that experience in this area could be developed by working with diverse groups, learning from clients, learning from other professionals, and through improved self-awareness.
Table 2.2  Implementation of effective cross-cultural genetic counselling techniques
Adapted from Weil (2001, pp.143-149)

<table>
<thead>
<tr>
<th>Area</th>
<th>Implementation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Knowledge of relevant ethnic groups</td>
<td>Genetic counsellors are informed about the ethnocultural groups to which the counselee belongs, through many techniques such as consulting the academic literature and interacting with groups outside the institutional setting, the latter approach being much more likely to reflect the diversity present within the group. He cautioned that the information obtained must be used carefully as there is great variation within groups</td>
</tr>
<tr>
<td>Ethnocultural self-awareness</td>
<td>Exploration of one’s own ethnocultural history, identity and world view helps one to better understand the way in which culture is intertwined with beliefs, perceptions, values and ways of thinking and assists the counsellor to more adequately appreciate these influences on the client’s experience of the genetic counselling situation</td>
</tr>
<tr>
<td>Institutional and social barriers</td>
<td>These take many forms including barriers to access to care, communication barriers beyond language such as the use of written materials and professional barriers such as lack of ethnocultural diversity within the profession</td>
</tr>
</tbody>
</table>

Referring to Kessler’s teaching and counselling models of genetic counselling, Lewis (2002, p.209) believed that multicultural genetic counselling could be best supported by the counselling model as this allowed the counsellor to focus on ethnocultural aspects. He proposed that further qualitative research of the “counselling moment” was required to examine what happens when counsellors and clients from different cultures interact in this moment, and to understand the issues that influence genetic counsellors’ choice of counselling practice.

From observing many genetic counselling sessions preceding amniocentesis (prenatal diagnosis) in hospital settings throughout New York, Rapp (2000) had previously shown that the discourse of genetic counselling is very much based on the medical model, using complex medical and scientific language. She described genetic counsellors as inherently bilingual – raised in one language then acquiring science as a second. During counselling she observed “code switching”, with counsellors moving from one language framework to another depending on
perceptions of who the client might be. Another technique the counsellors were observed to use was “lively and familiar metaphors.” These metaphors included “looking for a gene change is like looking for a needle in a haystack”, “50% risk is like tossing a coin”, “chromosomes are like strings of beads and the genes are the beads”, “being in a Catch 22” or “at a crossroads in decision making” (Evans, 2006).

A major goal of genetic counselling is to convey information and to communicate well in order to address the client’s questions and concerns. Rapp (2000) observed that counsellors found conveying standard scientific information concerning birth defects and genetic testing more difficult when a large amount of individual and cultural variation was involved. Some counsellors modified their discussion depending on their assessment of the clients’ scientific background, however, even when counsellors modified their approach a significant gap in the communication process remained. The codes, genres and assumptions of counsellors and their use of scientific terminology contributed to limited conversations with clients. For example, silence or “no” in response to a question often led the counsellor to assume a lack of family history, or that the client was ignorant or uninterested, whereas this response may simply reflect a lack of knowledge regarding diagnostic and medical services information (Rapp, 2000).

Cultural compatibility between counsellor and client has often been suggested as a solution for reducing the effect of cultural and language barriers. Wang (2001) suggested that cultural compatibility may enable the counsellor to challenge misconceptions in a sensitive way. However, knowing a person’s cultural, ethnic or racial background does not necessarily give the counsellor more information about the emotional state of the client or how they will experience genetic counselling (Lewis, 2002). It may be too limiting to assume that clients fit into a cultural group determined solely by characteristics such as ethnicity and religion. In a study of the perspective of clients, many indicated they would like to be treated as individuals with diverse customs, beliefs and cultures (Davidson et al., 2000).
Karbani (2002) believes that cultural compatibility can be used to minimise cultural and language barriers but is not the most important issue in successful transcultural genetic counselling. Rather, genetic counsellors need to have access to information about different cultures to facilitate understanding of their client, an open mind and a willingness to discuss these issues in supervision. Other authors continue to argue that recruitment and training of counsellors from within specific ethnocultural groups is effective in improving transcultural counselling (Raz & Atar, 2003; Weil et al., 1993). An approach that incorporates diversity more fully and enables transcultural dialogue between counsellor and client is needed. This would open the way for effective genetic counselling with ethnoculturally diverse groups, encompassing attention to social structures, communication styles, health care beliefs, migration patterns, experiences of discrimination and attitudes towards authority, and expression of emotions (Raz & Atar, 2003).

Irvine and McPhee (2002) explored the question of how to adequately train genetic counsellors in cross-cultural counselling and proposed ways to improve abilities in this area (Table 2.3). Their recommendations highlight the value of approaching each client as a unique individual and engaging with diversity, suggesting that addressing cultural issues can be achieved by “including all interested parties in negotiations” (Irvine et al., 2002, p.176).

The Self-Regulation Model proposed by Shiloh (2006) gives genetic counsellors a framework for engaging in a dialogue with their clients about their ethnocultural background and beliefs. Suggestions for how to use this model to engage with clients are summarised in Table 2.4.
Table 2.3  Recommendations for improving cultural competence
Adapted from Irvine and Kerridge (2002, pp.175-6)

<table>
<thead>
<tr>
<th>Engaging with diversity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Learning the skills of listening</td>
</tr>
<tr>
<td>Taking the time to listen attentively to patient’s personal narratives</td>
</tr>
<tr>
<td>Being open to learning about other cultures</td>
</tr>
<tr>
<td>Recognising the cultural basis of one’s own beliefs and assumptions</td>
</tr>
<tr>
<td>Including all parties by:</td>
</tr>
<tr>
<td>Explicitly acknowledging the patient’s cultural norms, beliefs and values</td>
</tr>
<tr>
<td>Locating the patient’s individual beliefs and values within that cultural context</td>
</tr>
<tr>
<td>Being explicit about the process of decision making that generally guides doctors’ behaviour (e.g. standard practice)</td>
</tr>
<tr>
<td>Coming to an arrangement between parties regarding how the interaction proceeds</td>
</tr>
</tbody>
</table>

Table 2.4  Model for engaging in dialogue about ethnocultural beliefs
Adapted from Shiloh, (2006, pp.325-337)

<table>
<thead>
<tr>
<th>Ways to engage with clients</th>
</tr>
</thead>
<tbody>
<tr>
<td>Examine the client’s general understating of genetics and heredity before any educational attempt is initiated</td>
</tr>
<tr>
<td>Explore specific representations clients may have before predicting or trying to modify their knowledge or reaction</td>
</tr>
<tr>
<td>Investigate the client’s prior experiences with genetic conditions using a personalised approach that would disclose individual meaning of the experience</td>
</tr>
<tr>
<td>Clarify the role self-representations, types of threat to self-concept and coping behaviours activated in response to these threats and direct counselling to deal with these issues</td>
</tr>
<tr>
<td>Evaluate the costs and benefits of client misconceptions before trying to change them</td>
</tr>
<tr>
<td>Consider interactions and compromises between cognitive and emotional motivations as predictors of client action plans for coping with genetic conditions and risks</td>
</tr>
</tbody>
</table>

Middleton and colleagues (2007) explored the provision of a transcultural genetic counselling service in the UK from the perspective of three genetic counsellors. They concluded that techniques of transcultural genetic counselling still require development and recommended further research, particularly an independent evaluation of the counselling experience from the perspective of the client (Middleton et al., 2007).
2.5.3. Future directions for research and practice

Several studies have identified diversity as a particularly common and challenging issue that arises in genetic counselling sessions. These studies used surveys and focus groups to explore ethical and professional challenges in genetic health professional practice, first in the USA and later in Australia. They found challenges are related to differences between counsellors’ and clients’ cultural, socio-economic, religious or spiritual perspectives and other points of cultural difference including ethnicity and education levels (Alliman et al., 2009; Bower et al., 2002; McCarthy Veach et al., 2001).

Other studies have focussed on service delivery barriers and racial knowledge of counsellors and the frequency and prevalence with which challenges in this area occur. Lee and colleagues (2009) suggested some of the barriers could be overcome by counsellors consulting with colleagues on diversity issues, obtaining knowledge of specific cultural groups and anticipating cross-cultural issues. This approach, however, may risk narrowing the definition of diversity and stereotyping of individuals or groups according to their ethnicity or religion. They also suggest educators and supervisors need to be informed about the most effective ways to pass on knowledge and skills about genetic counselling and cultural diversity. They propose that in addition to supervision, consultation and gathering of culture specific information, further development of cultural competency including cultural empathy, patient-centred practice and recognition of cultural hybridity is needed.

2.6. Monitoring the implementation of practice models

2.6.1. Outcome studies

While genetic counselling can be defined and the practice described, monitoring the implementation of models of practice is required to determine what makes genetic counselling useful or effective (Michie, Axworthy, et al., 1996). The effectiveness of genetic counselling was initially evaluated primarily in early outcome studies that
measured parameters such as educational effectiveness, reproductive intentions, client behaviour, client satisfaction and risk assessment. However, these early studies had considerable methodological problems, for example a lack of controlled study design (Michie & Marteau, 1996).

The first studies of genetic counselling focussed on the educational aspects of the counselling process, investigating clients’ recall of the information given to them. If the material was presented in a way that allowed recall in great detail, the process was considered a success. This approach to measuring the success of genetic counselling reflects the view that the practice embraces a top-down process of education, similar to most other types of medical education. Counselling is seen to take place between clients (who are highly motivated to learn specialised information about the distressing situation they are in) and counsellors (who have mastered scientific and medical knowledge in the area of human genetics and have some skills in patient communication) (da Rosa et al., 1998).

The value of this approach to measuring the efficacy of genetic counselling was challenged by later studies which showed that clients have low retention of scientific information about the risk of having a child with a severe genetic condition (Michie, McDonald, et al., 1997). While the cause of poor retention of information is likely to be poor communication on the part of the counsellor, other mechanisms may be impeding the storage and retrieval of distressing information (da Rosa et al., 1998). In a counselling session, clients are commonly at a time of crisis, under great duress and anxious about the information they may be about to hear. Information uptake by these clients is therefore variable and what is remembered may be different to what the counsellor thinks they have conveyed. Clients’ existing ideas and knowledge, risk perception, emotional interference with information processing and mechanisms (such as denial, self-protection and selective listening) all contribute to the complexity of the counselling session (Michie & Marteau, 1996). If there are psychological factors interfering with the
educational aspect of genetic counselling, it has been suggested that it may be advantageous to address these two issues separately (da Rosa et al., 1998).

2.6.2. Process studies

In order to determine whether genetic counselling makes a difference to outcomes, it is important to establish whether the process of genetic counselling changes the outcome for the client. To do this, a better understanding of the process is required – an understanding of what genetic counsellors do, or report doing, during a session (Michie & Marteau, 1996).

Process studies, designed to explore the content, behaviours and interactions of a genetic counselling session, can be used to examine the relationship between counselling processes and outcomes and may be the only way to determine whether what genetic counsellors say they do matches what they “actually” do in genetic counselling sessions (Kessler, 1992b, p55).

Despite the potential usefulness of process studies, Bernhardt et al. (2000) have highlighted the lack of research examining what actually happens during a genetic counselling session and how genetic counsellors communicate with their clients. Biesecker et al. (2001, p.191) suggest genetic counselling is a “black box” with very little known about what actually happens during a genetic counselling session.

The first study examining what actually occurs during a genetic counselling session was published by Kessler in (1981). This study explored how counselling was provided, what information was given, what issues were emphasised and the impact of the counsellor on the clients. Kessler was also interested in applying qualitative methods to genetic counselling interactions and his studies led to further qualitative studies of counselling styles and how in-session variables impact on outcome measures, including effectiveness and satisfaction. These studies addressed directiveness, examined psychosocial issues and identified effective strategies for counselling and training and development of genetic counsellors.
(Armstrong et al., 1998; Kessler et al., 1982). Sandelowski et al. (1996) used interviews to explore how couples made choices about their pregnancies after the diagnosis of a foetal anomaly.

Since these early studies, additional studies have shown that counselling sessions are often didactic in nature with very little time spent exploring psychosocial and emotional topics. Discussion of psychosocial issues comprises a small percentage of counsellor dialogue and concentrates on emotional reactions, attitudes and preferences, and the impact on family and social relationships relevant to testing and decision making (Butow et al., 2004; Duric et al., 2003; Ellington et al., 2006; Lobb et al., 2005; Lobb et al., 2004; Pieterse et al., 2005).

Failure to elicit clients’ emotional needs and concerns means that counsellors continue to dominate sessions, could fail to display empathy and to draw out all of the reasons why the client is attending. The differing information needs of clients may not be recognised, unvoiced agendas can be present and it is likely that social and emotional agendas are under-represented (Pieterse et al., 2005; Wells et al., 2004). Discrepancies in the perspectives of the two parties may arise because counsellors are unaware of their client’s most important issues, either because they had not made an attempt to find out or they had seriously misjudged their expectations (Michie et al., 1998). These problems may be exacerbated if ethnocultural differences are not taken into account. Despite training programs giving equal attention to clients’ information and psychosocial needs, studies indicate that attention to clinical knowledge predominates within counselling sessions (Roter et al., 2006).

Some of the published process studies of genetic counselling have been criticised for the small numbers of participants included. Given the small number of counsellors within genetic counselling units and the logistical difficulties of coordinating large multi-centre trials, small studies are unavoidable. These
limitations have been overcome in other studies by using video recordings of simulated genetic counselling sessions to examine the communication process and characteristics associated with the teaching and counselling models of practice (Roter et al., 2006). Additionally, qualitative research does not seek to generalise findings methodological and interpretative validity are seen as more important than sample numbers (Spencer et al., 2012).

Most recently, a review of process studies was undertaken (Meiser et al., 2008). This review found that studies have focussed on genetic counselling in the familial cancer setting. Additional findings include: that genetic counselling is often provider driven; a large proportion of communication comprises biomedical information rather than psychosocial dialogue; and the purpose of the consultation, and communication styles, affected communication behaviours. Interestingly, despite the consensus that genetic counselling should embrace a broader range of goals encompassing education and psychosocial support, the broader model of practice has not been fully implemented. Proposed future directions for research included more communication analyses in areas other than cancer genetic counselling, studies of alternate means of communication including telephone and tele-health consultations, more extensive studies on the adoption of teaching and counselling models, and more audio and video recordings of sessions (Meiser et al., 2008).

Qualitative methods have been recognised as one option for achieving a greater evidence base to inform practice. Expanding evidence-based theory on what occurs in the process of genetic counselling has the potential to inform practitioners about the psychosocial processes their clients are navigating, enable the counsellor to predict how the client might behave in response, and to develop interventions that give alternative ways to navigate the reactions (Grubs et al., 2010; McAllister, 2001). Increasingly, qualitative research methods are being employed to examine the process of genetic counselling. It is important to continue using this type of work to examine the perspective of the client and to gain a better understanding of
genetic counselling encounters (Fransen et al., 2006; McCarthy Veach et al., 2002a). The value of qualitative methods in genetic counselling research, and in particular process studies, is explored further in Chapter Four.

2.7. Summary

This chapter has examined the development of genetic counselling practice since its establishment within the discipline of medical genetics. The chapter first followed the progress from the early predominant unilateral medical model towards a more dialogical approach. The development of practice definitions, the role of psychological models and the ND tenet were explored. Diversity and cultural competence in the profession were reviewed and the final section of the chapter explored the role of process studies in evaluating practice.

This review reveals that when genetic counselling was first established, the primary aim was educating the patient and imparting the expert’s knowledge. Development of the profession has led to a shift towards a team approach, more typical of a counselling interaction, where the counsellor assists the client and facilitates movement towards the client’s ideal decision or position. Despite this shift, the overtone of an expert-lay interaction remains, with the implication that genetic counsellors have all the information to give and clients all the knowledge to acquire.

Implicit in the movement towards client-focussed communication is the dialogical process of exchange. In this exchange, genetic counselling becomes truly consultative and the counsellor recognises that the client also has something to give, be that information about themselves and their own thought processes or information about their cultural identity. Genetic counselling has responded well to the increasing diversity in the client base. Many genetic counsellors already have the resources to respond flexibly and empathically. Awareness of each participant’s cultural ‘history’ and the value of exploring clients’ narrative in the context of their cultural identity cannot be underestimated.
It is not useful to view the cross-cultural model of genetic counselling as purely developing specialist skills to work with specific ethnic groups (and thus assuming heterogeneity of such groups). In the context of society’s increasing ethnic diversity and cultural pluralism, rather than specialising in particular stereotypical groups, genetic counsellors may benefit from drawing on educational, counselling and psychological models of genetic counselling. Dialogical exchange is more important than cultural compatibility and permits a successful interaction with every client regardless of their background. Individual and organisational barriers to achieving cultural competence still need to be overcome.

The following chapter investigates the provision of genetic counselling in a culturally diverse community in Australia, particularly in the area of consanguinity. The application of newer dialogical models of genetic counselling for consanguineous couples allows for in-depth exploration of individual needs and preferences and the provision of true cross-cultural genetic counselling.
3. Consanguinity – a culturally diverse practice

In contemporary Western society, the term *inbred* is widely used as a term of denigration, and marriage between biological relatives are often treated, at best with suspicion and frequently with embarrassed astonishment. (Bittles, 2012a, p.7)
3.1. Introduction

Chapter Two explored the need for an approach to genetic counselling that enables practitioners to incorporate diversity of psychosocial and ethnocultural factors into practice. Genetic counsellors need to be equipped to deal with presenting genetic and psychosocial issues in a way that takes into account the wider familial and cultural circumstances. This chapter examines a particular presenting issue that requires genetic counsellors to address diversity, which is the application of cross-cultural genetic counselling to the area of consanguineous unions.

The word consanguineous comes from the Latin words ‘con’ meaning shared and ‘sanguis’ meaning blood. Within medical genetics a consanguineous union is defined as a union between two people who share at least one common ancestor and whose biological relationship is that of second cousins or closer (Bittles, 1998, 2003b). Spouses who are biologically related co-inherit a proportion of their genes from their common ancestor. The most common form of consanguineous union worldwide is between first cousins, who inherit 1/8 of their genes from a common ancestor (Bittles et al., 2010b). Different types of marriage patterns are preferred in different societies and are a function of religious and cultural beliefs, community attitudes and socio-economic factors (Bittles, 2008; Hamamy et al., 2011).

Consanguinity was chosen as an example of a source of cultural difference because it is not traditionally thought of as a cultural variable but rather a practice associated with particular cultural or ethnic groups. It is also an issue that is prone to skewed perceptions, cultural assumptions and stereotypes, and an area of practice where genetic counsellors have to deal with cultural diversity. The study is limited to cousin marriage because this is the most common type of consanguineous union in the world (Bittles et al., 2010b) and in the local cohort (Port et al., 2001).
I developed a particular interest in consanguinity through my practice as a genetic counsellor and my participation in research in this area, contributing to a study that measured the prevalence and outcomes of consanguineous unions in WA (Port et al., 2005). In counselling consanguineous couples, I found that they were not limited to specific ethnic groups and I began to question why there was a social stigma in Western society associated with consanguinity when it is an accepted practice in others (Bennett et al., 2002; Bittles, 2011). The literature on consanguinity tends to focus on genetic risk and ethnic groups where consanguinity is commonly practised (Hamamy, 2012). Consanguineous unions are often criticised in Western society and are seen as an issue that occurs only in specific communities and religious groups (Hamamy et al., 2011).

This chapter reviews consanguinity worldwide with a focus on global patterns and the Australian context. The historical, reproductive, genetic and social factors relating to consanguineous marriage are explored. The chapter then looks at how consanguinity is dealt with in genetic counselling and the complexity of issues surrounding this complex topic.

### 3.2. Global patterns of consanguinity

An estimated 10.4% of the global population are related as second cousins or closer (Bittles et al., 2010a). Up to one billion individuals in consanguineous unions are thought to live in countries where 20-50% of unions are between persons who are related as second cousins or closer. A further 2.9 billion individuals in consanguineous unions are thought to live in countries where 1-10% of marriages are consanguineous. The estimates (Table 3.1) are thought to be conservative as the data on consanguineous marriages in many countries are unavailable or limited at best (Bittles, 2008, 2012a).
Table 3.1  Current global prevalence of consanguineous unions  
Adapted with permission from Bittles (2012)

<table>
<thead>
<tr>
<th>Consanguineous marriages in the population</th>
<th>Population size (millions)</th>
<th>Percentage of total global population</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;1%</td>
<td>1 068</td>
<td>15.5</td>
</tr>
<tr>
<td>1-9%</td>
<td>3026</td>
<td>43.9</td>
</tr>
<tr>
<td>10-19%</td>
<td>35</td>
<td>0.5</td>
</tr>
<tr>
<td>20-50%</td>
<td>814</td>
<td>11.8</td>
</tr>
<tr>
<td>&gt;50%</td>
<td>227</td>
<td>3.3</td>
</tr>
<tr>
<td>Unknown</td>
<td>1 730</td>
<td>25.1</td>
</tr>
</tbody>
</table>

As shown in Figure 3.1, consanguineous unions are most common (20-50% of marriages) in North and sub Saharan Africa, Central and West Asia, and in most parts of South Asia. They are also quite common in South America, Japan and the Iberian Peninsula where they comprise 1-10% of marriages. Rates of consanguineous marriage in the Middle East are high (20-50%) and also vary widely between and within countries (Yunis et al., 2008). They occur to a lesser extent in small isolated communities, such as the old order Amish, Samaritans in Jordan and Israel, Hutterites, Dunkers and Romani Gypsies (Jaber et al., 1998).
Reduced rates of consanguineous unions in some countries have been observed and attributed to industrialisation, greater population movement, a decline in family size, improved economic status and higher literacy rates (Al-Arrayed et al., 2012; Hamamy et al., 2005; Tadmouri et al., 2009). In Western Europe and North America, Japan and Israel where consanguineous unions were once quite widespread, the prevalence of such unions has indeed decreased in recent generations (Bittles et al., 1991; Bras et al., 2009; Yunis et al., 2008). However, on a global scale, the number of consanguineous unions has not decreased, as in Southern and Central Asian and Middle Eastern communities consanguineous marriages remain a common cultural practice (Bittles, 1994; Hussain, 1999; Jaber et al., 2000). Also, rapid improvements in public health facilities in developing countries mean that more children survive to marriageable age and are able to exercise the preference for marriage to a close relative. Bittles et al. (1991)
predicted that a reduction in the frequency of certain types of consanguineous unions will occur but over time there will also be an overall reduction in consanguineous unions, especially in rural communities.

In direct contrast to the factors reducing the number of consanguineous unions are factors leading to possible increases in the global prevalence of consanguineous unions. These factors include the ongoing strong preference for close-kin marriage, the trend towards more fundamentalist doctrines in many Islamic countries and the high rates of population growth in Asia and Africa (Hamamy et al., 2011; Tadmouri et al., 2009). These factors will play an important role in determining the prevalence of consanguinity not only in developing nations but also in developed countries in Western Europe, North America and Oceania due to large numbers of immigrants from Asia and Africa.

3.2.1. Impact of global migration

International migration has changed in both scale and nature, with migration of skilled workers, illegal immigrants and refugees becoming common, many of these from countries where consanguineous marriages are preferred (Bittles, 2008). This pattern has been reflected in Australia, with an increasing proportion of migrants arriving from Asia and Sub-Saharan Africa (Metcalf et al., 2008).

While changes in urbanisation and population mobility initially led to a decline in the practice of consanguineous unions up to the mid-19th Century, many developed countries have seen a reversal in these trends since the middle of the 20th Century (Bittles et al., 2010b). In Western Europe, North America and Oceania, sizeable migration from regions where consanguineous unions are favoured (such as Asia, Africa and the Middle East) have resulted in an increase in consanguinity rates (Hamamy et al., 2011). Studies of immigrant populations in their adopted country have shown the continued practice of marrying close relatives, sometimes in higher proportions than the indigenous population in their country of origin (Bennett et al.,
2002; de Costa et al., 2001; Hamamy et al., 2009; Port et al., 2001; Port et al., 2005; Reniers, 2001). Proposed reasons for this change include: finding a partner from within the community, which may be small and comprise a small number of kindreds; maintenance of community traditions in a new and unfamiliar environment; and preservation of traditional family and cultural values. These reasons recognise that “sociocultural determinants of marriage choice, rather than religion or economic reasons, appear to dominate the preference for consanguineous marriages” (Hussain, 1999, p.458).

The Norwegian Medical Birth registry has kept records for the past 30 years on the biological relationship between parents. Early data from the registry showed an increase in the number of children born to consanguineous parents who migrated from countries where consanguineous unions were common (Stoltenberg et al., 1999). More recent data however, has shown a decrease in the proportion of children born to this group of consanguineous parents (Grjibovski et al., 2009).

This trend towards an increasing number of consanguineous marriages has also been observed among Turkish and Moroccan immigrants in Belgium (Reniers, 1998), the Lebanese community in Australia (de Costa et al., 2001), immigrants from the Middle East in Canada (Hoodfar et al., 1996) and the Pakistani community in the UK (Modell et al., 2002).

3.2.2. Consanguinity in Australia

Australian society is becoming increasingly diverse, with its members coming from a wide range of ethnic, religious and socio-economic backgrounds. With 28% of the current Australian population (22 million) born overseas, immigration has become a significant contributor to Australia’s population growth, comprising at least 45% of population growth almost every year since 1998-99 (Australian Bureau of Statistics, 2014). The variety within second generation Australians reflects past migration and intermarriage patterns. Of all persons born in Australia, 25% have at least one
overseas-born parent and of these 44% have both parents born overseas (Australian Bureau of Statistics, 2008a, 2008b).

Immigrants now come to Australia from more diverse regions of the world and while British migrants accounted for 34.2% of visas issued from 1986 to 1991, Europe is no longer the primary source of immigrants to Australia. For a variety of economic, social and political reasons, the number of European immigrants has been eclipsed by those from Asia and Africa. In the period 1986-1991, Australia derived 46% of its migrants from South and East Asia, while a further 5.9% of visas were issued to migrants from West Asia (Stahl et al., 1993). Sudan was ranked 75th as a source of Australian immigrants in 1985-86 but rose to seventh in 2005-06 (Australian Bureau of Statistics, 2014, 2008b). Significant numbers of migrants have also come to Australia in recent years from Lebanon, Egypt, Cyprus, Israel, Iran, and Syria (Stahl et al., 1993) and North-west Europe, Asia and Africa as a result of the skilled migrant program and the humanitarian program (Australian Bureau of Statistics, 2014).

These figures show that increasing numbers of immigrants come to Australia from countries where there are often relatively high rates of consanguineous marriages. While the prevalence of consanguineous marriage in WA remains low, this may change with continued immigration from these countries. This has been seen in Europe where there are now 10 million residents originating from countries where consanguinity is preferred (Bittles et al., 2010a; Port et al., 2001).

3.3. Consanguinity – genetics and reproduction

3.3.1. Historical studies

The first record of the debate of the ‘safety’ of consanguineous unions was written in 1839 by the physician Thomas Hodgkin. He wrote a pamphlet opposing the opinion of the Society of Friends (Quakers) who prohibited the marriage of first
cousins, and repeatedly denied him permission to marry his first cousin. His pamphlet cited both theological and scientific evidence that there was little risk in marriage between first cousins (Geller, 2002).

One of the first investigations into the effects of consanguineous marriages was undertaken by Charles Darwin (Darwin, 1862). He was one of many eminent persons of the late 19th Century to marry their cousin; he had ten children with his first cousin, seven of whom survived to adulthood. Darwin was interested in investigating the then common belief that children of these unions were at intellectual and biological disadvantage. He asked Sir John Lubbock to petition the British Government to include a question on the number and effects of first cousin marriage in the 1871 Census of Great Britain and Ireland. The request was rejected on the grounds that couples in consanguineous unions might be embarrassed by the question (Bittles, 1994). His son George then devised an alternative method of studying such marriages – using isonomy to establish unions where individuals might be related because they shared the same surname (Bittles, 2003a).

At a similar time, investigations into consanguineous marriages were being undertaken in the USA. In 1855, Reverend Charles Brooks appealed to the American Association for the Advancement of Science to study the effects of cousin marriage (Bittles, 2009). Dr Samuel Bemiss then analysed data on premature deaths and physical and intellectual handicaps in the offspring of biological relatives, from third cousin matings to incestuous matings and concluded that consanguinity was harmful (Bittles, 1994). Later, his views were supported by Lewis Henry Morgan in his book on marriage patterns among Native American Indians (Bittles, 2003a).
3.3.2. Consanguinity and genetic risk

In order to understand the distribution and burden of disease in populations, it is important to have accurate figures on the incidence and recurrence risks of genetic disorders. These figures can be used to guide genetic counselling of clients and their families. While inbreeding over multiple generations may be harmful owing to the expression of commonly inherited recessive genes, this outcome occurs in only a small number of families and individuals within select populations (al Gazali et al., 1995; Bundey et al., 1991). Accurate figures on the incidence of genetic disorders within these populations are not available. Inconsistencies in the available figures suggest that the arguments against consanguinity may be invalid (Bittles, 2011).

Generalisations about the range and incidence of inherited diseases associated with consanguinity are therefore often misleading as they are made on the basis of outcomes for small, inbred populations (Bittles et al., 2010a). Whilst the high rate of perinatal mortality and congenital malformation in these populations may be explained by high rates of consanguineous unions over generations, many studies fail to recognise that social variables can lead to overestimates of genetic risk. Other factors that may contribute to adverse genetic outcomes, such as personal, socio-economic, service-related and environmental factors are often not well explored (Ahmad, 1994; Bittles, 1998, 2001). In addition, many studies are based on comparisons between consanguineous and non-consanguineous groups, with the latter not differentiated on the basis of degree of relatedness (Tadmouri et al., 2009).

3.3.3. Incidence of birth defects and genetic disease

The focus of the early literature on consanguineous marriage was the incidence of rare autosomal recessive conditions, attributed to the inheritance of autosomal recessive genes from common ancestor(s), in communities with high consanguinity rates such as the Pakistani immigrant population in the UK.
In the early 1990s Bundey et al. (1993) suggested that mortality and severe morbidity in the children of British Pakistanis could be reduced by up to 60% if consanguineous marriage was avoided. Studies of Pakistani immigrants living in Norway has shown that children born to these immigrants have double the infant mortality rate (OR=2.16, 95% CI 0.88-5.32) and 1.5 times the perinatal mortality rate (OR= 1.43, 95% CI 0.54-3.78) (Orstavik et al., 1994; Stoltenberg et al., 1999). These figures were mirrored in a study of infant mortality in Yorkshire and whilst data on consanguineous marriages were not specifically collected, the researchers inferred that the higher than expected incidence of recessive conditions resulted from the common practice of consanguineous marriages in the Pakistani migrant community (Bacon et al., 1994).

Other studies have shown that the rates of congenital malformations among the offspring of consanguineous couples worldwide are significantly higher than the rates in unrelated parents (Bennett et al., 2002; Bittles et al., 1994; Bromiker et al., 2004; Jaber et al., 1998; Stoltenberg et al., 1999; Zlotogora, 2002). A number of different structural malformations have been described, including malformations of the cardiac, urogenital, ophthalmic, gastrointestinal, skeletal, cutaneous and central nervous system. Whilst these malformations have not been classified as typical autosomal recessive conditions, it is postulated that they result from increased homozygosity of genes at several loci (Jaber et al., 1992; Stoll et al., 1999).

Many of these studies have been criticised for ascertainment bias as the data comes from populations where consanguineous marriage is common, where there is poor record keeping and no clear definition of what is considered to be a major birth defect. It has been suggested that these studies may not provide an accurate assessment of risk because of the extremely variable nature of the populations studied, the ascertainment methods employed and the variation in definitions of adverse outcomes (Bittles et al., 2010a). Studies where the probabilities of adverse outcomes are calculated on an individual basis using the background population
risk, degree of consanguinity and relevant family history are required (Bennett et al., 2002).

Reports of multigenerational Down syndrome cases in an inbred population in the Gulf region prompted speculation about a recessive gene that controlled non-disjunction (Alfi et al., 1980). Other than this endogamous population, an association between consanguinity and Down syndrome (or other chromosomal disorder) has not been reported (El Mouzan et al., 2008; Zlotogora et al., 2010).

Consanguinity is also proposed to have an adverse effect on cognitive function, with a number of studies indicating that average IQ is lower and the frequency of intellectual and developmental disability is higher in inbred populations (Jaber et al., 1998). Calculations of this risk, however, are often based on studies of non-Western populations and are not necessarily applicable to consanguineous couples in Western countries (Bennett et al., 2002).

### 3.3.4. Common complex diseases

Until recently, few studies have examined the relationship between consanguinity and diseases of adulthood and those that have been published have methodological problems, including difficulty defining control groups and controlling for endogamy (Hamamy et al., 2011). This has resulted in contradictory reports of both positive and negative associations between consanguinity and breast cancer, and consanguinity and heart disease (Bittles et al., 2010a; Jaber et al., 1997; Rudan, Rudan, et al., 2003; Rudan, Smolej-Narancic, et al., 2003; Shami et al., 1991). No increases in common complex disorders, for example diabetes, myocardial infarction, hypertension asthma or duodenal ulcers, have been reported (Jaber et al., 1998; Shami et al., 1991). An increase in susceptibility to multiple sclerosis has, however, been reported (Roberts, 1991). Further standardised research that controls for non-genetic variables has been suggested to resolve the remaining unanswered questions (Bittles et al., 2010b; Tadmouri et al., 2009).
3.3.5. Reproduction and fertility

The effect of consanguinity on reproductive outcomes is equally as unclear as genetic risk, with conflicting reports in the literature and a “notable lack of evidence with respect to the major socio-demographic determinants of fertility” (Hussain et al., 2004, p.1). A higher incidence of sterility, spontaneous miscarriages, stillbirth, perinatal loss, low birth weight and neonatal death is reported in some studies; however, the majority of studies have not found a significant effect of consanguinity on these reproductive outcomes (Tadmouri et al., 2009).

In a large study in Norway using data collected on all births in the country since 1967 (including the relationship between parents), the recurrence of stillbirths and infant deaths among children of consanguineous and unrelated parents was calculated for the first time (Stoltenberg et al., 1999). The risk of stillbirth and infant death was found to be higher for the offspring of first cousin unions in this Norwegian cohort than offspring with unrelated parents.

A study of women attending a Western Sydney antenatal clinic in 1999 also examined the link between consanguinity and reproductive outcome (de Costa et al., 2001). Of the 1331 women attending, 262 (20%) stated that they were in a consanguineous marriage (136 were first cousin marriages) and 157 (60%) of these were born in Australia, the remainder were from the Middle East or South Asia. It is often assumed that in Australia consanguineous unions are more common amongst overseas born couples; however in this study, 60% of the women in the consanguineous group were born in Australia. Previous pregnancy loss, perinatal morbidity and genetic abnormalities were higher in the consanguineous group than in the non-consanguineous control group.

In contrast to these negative reproductive outcomes linked to consanguinity, overall fertility rates are not reduced in consanguineous couples. This has been attributed to factors such as the young maternal age at marriage and first live birth, illiteracy,
and lower contraceptive use (Bittles, 2001; Tadmouri et al., 2009). A study investigating the outcomes of assisted reproductive technology in infertile consanguineous couples in Kuwait (where the incidence of consanguineous marriage is high at 20-50%) found that the pregnancy rate per IVF cycle was lower in the consanguineous group and spontaneous abortion rates were higher than in the control group although the differences were not statistically significant (Egbase et al., 1996). Bittles et al. (2002) conducted an analysis of 30 published studies and combined the data to measure levels of fertility in couples in specific types of consanguineous unions. In addition, they used the Indian National Family and Health survey to compare fertility in consanguineous versus non-consanguineous marriage. This approach allowed correction for socio-demographic variables and found that consanguinity had neither a positive or negative effect on fertility.

### 3.4. Consanguinity – socio-cultural factors

In addition to the impact of consanguinity on reproductive outcomes and genetic disorders, consanguinity is a complex topic involving major social, economic, and demographic influences, differential reproductive behaviour, and early- and late-onset morbidity and mortality (Figure 3.2). This complexity needs to be taken into account when planning services for consanguineous couples. Bittles and Black (2010a, p.3) believe that consideration of these “salient non-genetic variables” is essential if genetic counselling is to address “the concerns of individuals, families, and communities with regard to reproductive choices, and in designing genetic education and genetic counseling programs for consanguineous couples.”
Consanguineous marriages are preferred in culturally, socially, geographically and religiously diverse populations throughout the world (Hamamy, 2012). Family tradition, strengthening of family ties, parental choice of spouse, closer relationship between a wife and her in-laws, greater marriage stability and durability, pride, ease of prenuptial arrangements, financial advantage (relating to lower dowry or bride wealth payments), preservation of family wealth and avoidance of undisclosed health or social problems are the most important and overriding reason for continuing such marriage practices (Bittles et al., 2010a; Hamamy et al., 2009). Despite marked demographic changes, including industrialisation, urbanisation and improved socio-economic status for women, continuation of the practice suggests
that for many communities the social and economic advantages outweigh any genetic disadvantages (Tadmouri et al., 2009).

The highest rates of consanguineous marriage are generally found in more traditional rural communities that are isolated, poor and less educated (Bittles et al., 1993). Exceptions to this rule include some major landowning families, traditional ruling groups and high social classes (Reddy et al., 2007; Schull et al., 1972). One of the main factors contributing to the prevalence of this type of marriage is the lack of a suitable mate outside the family because of geographical, tribal or cultural isolation (Jaber et al., 1998; Jaber et al., 2000).

### 3.4.1. Religion

In Western society, widely held prejudices against consanguineous unions continue and are thought to be attributable to historical religious and secular prejudices dating back as far as the 6th Century AD. The Latin Catholic Church released a papal communication banning marriages up to and including third cousins unless they were granted a dispensation and paid a fee to the Diocese, this was amended in 1917 and limited to first cousins (Bittles, 2003a; Bittles et al., 2010a). In WA, eight applications for permission to marry a first cousin were made to the Roman Catholic Archbishop between 1996 and 2000 (Port, 2007). These biblical prohibitions have had a major influence on Western marriage customs and legislation; however many misinterpretations, additions, deletions and controversies have transpired because of the different cultural contacts and social orders of the societies that have adopted them (Bittles, 2008, 2012a).

It is a widely held misconception that consanguinity is restricted to Muslim communities and whilst the practice is not necessarily encouraged by the faith, rates are high in Islamic society (Bittles et al., 2010a; Hamamy et al., 2011). First cousin marriages are permitted but uncle-niece unions are prohibited in the Quran (IV, v23), which also forbids certain consanguineous unions (mother, daughter,
sister, niece and aunt), fosterage (foster-mother and sister) and affinity (wife’s mother and daughter) (Bittles, 2001; Hussain, 1999; Panter-Brick, 1991). The Prophet Mohammed chose biological relatives as two of his spouses and arranged the marriage of his daughter Fatima to his first cousin Ali (Bittles, 2001; Hussain, 1999). It appears that the preference for first-cousin marriage in most Muslim is influenced more by socio-economic factors (Bittles et al., 2010).

Members of many other religions also practice consanguineous marriage including Christians in Lebanon, Jordan and Palestine, Buddhists, Christians, Jews, Parsees and Druze in South and West Asian countries and Hindu populations of Southern India (Bittles, 1998; Hamamy et al., 2011). In China, as in South India, the marriage of the children of two brothers is prohibited but the children of two sisters, or a brother and sister is permitted (Jaber et al., 1998; Jaber et al., 2000).

3.4.2. Legal regulation

Despite the lack of convincing biological data, the early religious debates in the UK and USA seem to have been the progenitor of the different legal situations in both countries with first cousin marriage being legal in the UK but a criminal offence in many states in the USA (Bittles, 2003a). In the USA, first cousin marriages are a criminal offence in ten states and are illegal in 22 others, despite the recommendation that these laws be rescinded (Bittles, 2003a, 2004). In addition to prohibiting first cousin marriage, some states also prohibit half-sibling and second and third cousin marriages (Schwartz et al., 1997).

Some of the legal restrictions on consanguineous marriage arise from the incest taboo, with almost all US states prohibiting marriage between parent/child, siblings, uncle/aunt, niece/nephew, and grandparent/grandchild. The definition of incest in this legislation includes unions between both biological and non-biological family members and therefore is not always based on biological research or genetic theory (Ottenheimer, 1996).
3.4.3. Acceptability of consanguineous marriages

Whether consanguineous marriages are perceived positively or negatively is dependent on established religious or cultural beliefs (Bittles, 2003b). The attitudes of Western society (including the medical profession) towards consanguinity are often negative and fuelled by a persistent lack of accurate information about adverse outcomes in children. Despite evidence that shows the deleterious effects of consanguinity may be exaggerated (Raz & Atar, 2003; Shiloh et al., 1995), health workers in Western countries often pressure people to go against the custom in communities where such marriages are common and socially preferred (Modell et al., 2002). Images of remote and poor inbred communities, in which a large proportion of their members have physical disorders and intellectual handicap are commonly reproduced (Denic et al., 2011).

In Western countries, consanguineous couples are often the subject of social stigmatisation, rejection and isolation by their family and friends who believe that the relationship should not continue (Hamamy, 2012; Read et al., 2012). In the USA, legal prohibition has led to a fear of prosecution and couples report keeping their relationships hidden from friends, relatives and medical professionals (Bennett et al., 2002). These attitudes are often reflected and perpetuated in the media as illustrated by the headlines such as: “Fears of infant deaths in cousin marriages” The Age 15/10/1991, “Kissing cousins set trend” The Age 15/10/1991, “Go ahead, kiss your cousin” Discover Aug 2003, “Cousins: A new theory of relativity” Time 15/04/2002, “Kissing Cousins” ANU Reporter 28/11/2008.

In the Australian community the incidence of consanguineous unions is low (<1% of marriages) and the majority of consanguineous couples are from communities where it is not a favoured marriage type. As a result, many have suffered owing to isolation and social stigma (Port et al., 2001). In addition, there has been increasing immigration from countries where consanguineous marriage is favoured, and these
couples may experience, for the first time, ignorance and a lack of understanding of this form of marriage.

In Western society, if people have had a close relationship with their cousins in their formative years, they often liken the relationship to that of siblings suggesting a perception that a sexual relationship between cousins could be incestuous. “Incest taboo” is a common feature of nearly all societies and present-day marriage systems (Alvarez et al., 2011). No socio-economic or ethnic group is thought to be exempt from this taboo, with most prohibiting marriage or sexual relations between nuclear family members on moral or legal grounds. Investigations of the biological effects of incest and access to accurate incest figures are hampered by the incest taboo and legal ramifications, resulting in an inability to study large unbiased data sets (Bennett et al., 2002; Bittles, 2004; Denic et al., 2006).

The various explanations for incest taboo are based on social, biological and psychological theories. Social theories focus on the belief that sexual restrictions serve as a form of marriage regulation, serving to maintain roles within the family and encourage development of relationships with external kin groups (Bittles, 2004; Reniers, 2001). This is at odds with the preference for preservation of consanguineous marriages in many communities (Denic et al., 2011).

**3.5. Consanguinity – genetic counselling**

Most of the literature on consanguinity focuses on the genetic risks for couples from groups where consanguinity is the norm and thus ignores the needs of couples from other communities. Other than the NSGC guidelines, there appears to be little or no literature to draw from regarding genetic counselling for more heterogeneous groups. Genetic counselling for consanguinity can be challenging, as in many populations it presents infrequently and requires specialised counselling skills. The counsellor may need to explain the reasons behind Western societies’ opposition to
consanguineous relationship and provide couples with the tools to re-educate their relatives about the risk of adverse reproductive outcomes.

### 3.5.1. Approaches to genetic counselling for consanguinity

The nature of genetic counselling provided to consanguineous couples has been surveyed by Bennett et al. (1999). They concluded there was a large discrepancy in the information and risk figures (0.25 to 20%) given to couples by clinical geneticists and genetic counsellors. In 2002, in response to this research and a lack of published information about consanguinity, the NSGC produced a set of guidelines for counselling consanguineous couples. These guidelines aimed to provide consistency in the approach and information provided to the couples. The guidelines included: providing preconception reproductive options; improving pregnancy outcomes; discussing reproductive options; reducing morbidity and mortality of children in the first years of life, and respecting psychosocial and multicultural issues (Bennett et al., 2002, p.102).

In addition to a detailed medical family history and assessment of risk, the guidelines highlighted a need to address the psychosocial history of the individuals or couple, including: exploring any concerns or feelings; clarifying family myths or misconceptions about risks; family support structures; and any cultural belief systems and traditions regarding consanguineous marriage and the causation of birth defects (Bennett et al., 2002). The authors also suggested that particular attention should be given to specific psychosocial issues relating to consanguinity, such as couples feeling the need to keep their relationship hidden due to fear of ostracisation, stigmatisation, discrimination, or legal prosecution. Guilt and shame reactions and family disapproval and blame have been observed in couples who have children born with congenital anomalies (Bennett et al., 2002).

In order to determine genetic risk during a genetic consultation, genetic counsellors usually enquire whether a couple are related. Questions about relationships
between individuals and consanguinity can be unsettling and have been found to evoke embarrassment, laughter and occasionally disbelief and denial (Rapp, 2000). Even in cultures where cousin marriage is accepted, questions like these may be greeted with suspicion or a desire to not answer the question for fear of further questioning and recrimination.

Despite the guidelines and the availability of genetic counselling services, some consanguineous couples may not access these services either because they choose not to do so or because they are unaware that such services exist (Posch et al., 2012). In a study of 261 pregnant consanguineous couples, de Costa et al. (2001) found only eight (3%) of the couples received genetic counselling. In Israel, consanguineous couples constitute about 4% of referrals to genetic clinics, despite the Israeli Arab population comprising 33% of the total population, with 20-30% of marriages being consanguineous in this subpopulation (Shiloh et al., 1995).

For those clients that do attend counselling, while guidelines are useful, it is important to remember that they each have a unique background and issues for exploration. For example, a second generation Australian couple may be coming at the insistence of their relatives who are against their union, fearing abnormal children, whereas another couple from Lebanon may be referred by their doctor for information on antenatal testing. While they are both seeking counselling about the genetic risks associated with consanguinity, they come for different reasons and with different counselling issues to explore.

3.5.2. Lay beliefs about illness and genetics

The way in which individuals, couples and families understand, and respond to, a genetic condition or the risk of a genetic condition is both variable and individual (Shaw et al., 2008). Lay beliefs are a challenge to genetic counsellors because it may be difficult to replace them with seemingly contradictory genetic information. It is important to determine the client’s prior knowledge and explore these beliefs,
before or during a consultation, as genetic counselling may be less effective if clients misunderstand new information because of their existing beliefs about inheritance (Lanie et al., 2004). Exploring a client’s pre-existing beliefs, and acknowledging the importance of these beliefs, may lead to improved communication and the establishment and maintenance of rapport (Barlow-Stewart et al., 2007; Middleton et al., 2005).

A survey of 36 Saudi Arabian consanguineous couples showed that many beliefs about the neuromuscular illness affecting their children stemmed from religious beliefs (the will of Allah), folk explanations (the evil eye), and to a lesser extent genetics (Panter-Brick, 1991). Some couples who acknowledged a genetic basis for the disease”, did so “without truly believing or understanding it”, and only did so only if the family pedigree made it obvious (Panter-Brick, 1991, p.1296). Other studies have shown the use of religion and folk beliefs allow individuals to adjust to the difficulties they are experiencing, to adduce some individual meaning, and to devise a coping strategy (Shaw et al., 2008). Individuals with a lack of understanding of genetics may find religious explanations easier to comprehend and more palatable.

In the aforementioned study of consanguineous couples, a variety of techniques were used by the couples to assimilate information (Panter-Brick, 1991). Their explanations of genetic disease included Islamic teachings and popular and secular beliefs, which jointly shaped their patterns of health care utilisation and reproductive behaviour. Others rejected advice about their risk of having further children with genetic conditions or disability, as they believed they were “in the hands of Allah” (Gatrad, 1994, p.173). Khlat’s (1986) study of consanguineous couples in Lebanon also showed that couples continue to enter into consanguineous marriages despite being aware of the genetic risks.
3.5.3. Complexity of genetic counselling for consanguinity

There are a number of factors that contribute to the complexity of genetic counselling for consanguineous couples. In addition to variations in ethnicity and lay beliefs, language problems, time pressure and cultural and educational differences may also hamper attempts to provide appropriate services to consanguineous couples (Khan et al., 2010). Clients’ beliefs about the causes of birth defects and genetic conditions, and about health, disability and folk medicines, can also contribute to these difficulties. In addition, some cultures have expectations about the role of medical professionals and authority figures and find the nondirective nature of genetic counselling confusing (Hamamy et al., 2009; Punales-Morejon et al., 1992). In some countries access to genetic services is also a problem, both in terms of available services and possible stigma attached to attending (Alkuraya, 2013). These issues may require the provision of specific training for GHPs working with these additional challenges.

A person’s cultural background is also influenced by a plethora of experiences and ideas about issues such as marriage and de facto relationships, sexuality, gender, and disability. This unique cultural background can impact on their experience of genetic counselling. These varied ideas and experiences prevent people being easily categorised on the basis of simple factors such as ethnicity. However, despite the complexity and uniqueness of individual situations, in some circumstances there is a degree of homogeneity. For example, in WA there are a large number of immigrants from Africa, and in particular Sudan, many of whom have experienced deprivation, refugee camps and upheaval (Mutch et al., 2012). Labelling them all as Sudanese immigrants would ignore their individual experiences, but being aware that they have likely experienced specific and extreme hardships may help genetic counsellors address some of the psychological needs of this group of immigrants.

The broad area of consanguinity can be divided into three categories. There are countries and cultures where consanguineous unions are traditional and
encouraged such as in Southern Asia and the Middle East. There are countries where a large migrant community, with high proportions of consanguineous marriage, exists within a culture where this is not routinely practised, for example Pakistanis in Britain and the Lebanese community in Sydney, Australia. Finally, there are cultures where consanguinity is not a traditionally practised form of marriage, for example Australia and the USA. The presenting issues and the nature of the problem vary depending on which of the three categories predominates. The kind of genetic counselling provided would therefore differ for consanguineous couples in countries where it is a traditional practice from those where it is not.

3.5.3.1. Countries where consanguineous marriages are traditional

In this first category, countries and cultures where consanguineous marriage is prevalent, the availability and effectiveness of genetic counselling and education programs depends largely on the country’s income and the resultant change in focus from nutrition and infectious diseases to genetic disorders (Alkuraya, 2013; Teebi et al., 2006). Programs aimed at reducing the burden of genetic diseases have been effective in Israeli Arab and Bedouin villages and wealthier countries in the Middle East (Bittles et al., 2010a; Raz et al., 2004; Shiloh et al., 1995; Zlotogora et al., 2009). However, in low income countries, a lack of trained scientists, clinicians and genetic counsellors affects the implementation and success of these programs (Alwan et al., 2003; Hamamy et al., 2009; Meyer, 2005). In addition to resource issues, specific cultural factors affect acceptance of genetic counselling such as the expectation that directive advice will be given and the importance of community norms over individual autonomy (Raz, Atar, et al., 2003).

Where consanguinity is widely practiced, genetic counsellors need to approach educating families and communities about the options of screening and prenatal diagnosis with sensitivity to the social, cultural and religious frameworks present (Alkuraya, 2013; Khan et al., 2010). Most commentators agree that the genetic implications of consanguinity need to be seen in cultural context and that criticism
and efforts to disrupt traditional marriage patterns should be avoided (Sathyanarayana Rao et al., 2009). Rather, reducing the impact of genetic conditions can be achieved by more culturally sensitive methods such as family counselling, carrier detection before or during pregnancy, prenatal diagnosis and selective TOP (Alkuraya, 2013; Hamamy, 2012). A non-judgmental and family-oriented, community-based approach is essential in order to establish trust, good communication and mutually advantageous working relationships for couples with a family history of recessive genetic disorders (Hamamy et al., 2009; Sathyanarayana Rao et al., 2009).

Several authors suggest logical strategies for reducing the social and economic burden of consanguinity. These include: educating populations about the genetic diseases and causes of increased risks of congenital malformations (Teebi et al., 2006); identifying the disease caused by founder genes in relevant communities to allow for carrier screening, premarital and prenatal counselling (Jaber et al., 1998); providing preconception, prenatal and genetic counselling services and information; working in a caring sensitive and non-judgmental fashion (Sathyanarayana Rao et al., 2009); and working with religious authorities to clearly define attitudes towards prenatal diagnosis (Jaber et al., 1998).

Jaber and colleagues (1998, p.16) take a more assertive approach by suggesting that “consanguineous unions are not discouraged when love is involved, but this type of marriage should nevertheless not be encouraged” and to “provide prenatal and genetic counselling services and educate as many people as possible to avail themselves of these”. This is contrary to the goals of genetic counselling and fails to take into account cultural and social factors. Bittles (2003b) suggests that the implementation of diagnostic, genetic counselling and treatment programs may be hindered by these “experts” who insist the practice is biologically dangerous and should be avoided.
Reports on genetic counselling to prospective consanguineous couples convey the impression that most find the information reassuring and proceed to have a family (Shiloh et al., 1995). A study of Israeli Jews investigated the effect of genetic counselling on couples’ decisions to marry and have children and also addressed how the information gained in genetic counselling affected clients’ beliefs, attitudes and behaviour (Shiloh et al., 1995). The authors found that genetic counselling did not primarily change decisions, but did affect decisional confidence. A difference in the number of children born to counselled and non-counselled cousins was also observed and the authors directly attributed this to the influence of counselling. However, as this issue was not addressed directly, it is not possible to determine whether this group of counselled cousins had fewer children because they were more worried about adverse reproductive outcomes. The authors recommended further studies to specifically address reproductive decisions.

The majority of the couples (65%) in the Shiloh et al. (1995) study made suggestions about how the counselling process could be improved. The most frequent of these suggestions was to provide tests of couples’ genetic compatibility, suggesting many of these couples desired to reduce reproductive uncertainty when making very important personal decisions. Some couples also suggested the counselling was too general and they would prefer more personalised information, such as information on a specific genetic disease and examples of how families coped with this genetic disease. Distress related to being uncertain about one’s own individual risk, despite having more accurate knowledge of the likelihood of genetic disorders, has been documented elsewhere (Lippman-Hand et al., 1979a). Other couples suggested the counselling process should include directive advice and some wanted fewer people involved to maintain confidentiality. By far the most common suggestion (suggested by 91% of the couples) was the development of a premarital education program that included genetic assessment of individual risks, advice for how to manage the
probable or possible outcomes, approaches to reduce fear and increase confidence, and tools to use for informed decision making (Shiloh et al., 1995).

3.5.3.2. Countries with migrant communities practicing consanguineous marriage

In the second consanguinity category, countries where migrant enclaves commonly practice consanguineous marriage despite the country overall having low levels of consanguinity, approaches to genetic counselling for these enclaves are varied. They include: minimising language and cultural barriers to accessing services; ensuring that the information provided to clients takes into account prior understanding of the causes of illness and inheritance; and avoiding stereotyping on the basis of assumed beliefs or practices (Shaw et al., 2008; Shaw et al., 2009). When counselling clients that fall into this second category, it is helpful to have specific knowledge or skills relating to the community the clients come from, to account for cultural meaning of illness, concepts of kinship and to facilitate engagement (Saleh et al., 2011). The counsellor does not necessarily need to be from the migrant enclave that the client comes from. Rather, acquisition of knowledge about the client and their cultural practices and cultural sensitivity skills can be equally important.

3.5.3.3. Countries where consanguineous marriages are uncommon

In the third category, for example countries such as Australia, with a low background rate of consanguineous marriage, consanguineous couples outside specific migrant enclaves are likely to be from a diversity of communities and exhibit cultural hybridity. Exploration of individual presenting issues and concerns is required in the approach to genetic counselling this group. Genetic risk is an important issue to explore but may be superseded by concerns about the response of the client’s family, particularly if the presenting couple is the first consanguineous union in the family.
Despite consanguineous marriages being uncommon in Australia, there are specific migrant communities in which it is a significant issue. While it may be advantageous for specialist genetic counsellors to address specific cultural and language issues of clients from these communities, this may not be feasible in Australia owing to the relatively small size of the profession and the lack of diversity amongst counsellors. While specialist counsellors may be applicable in specific communities in Australia, in WA this would not be pertinent as the majority of consanguineous couples accessing genetic services in WA are not from communities traditionally associated with consanguineous marriage. There is in WA, however, a clear need for genetic counselling of consanguineous couples. In WA, a review of the records from 1975-2001 showed that consanguineous couples made up about 2.0% of the total numbers of referrals to genetic services. In the six years from 1995-2001 these referrals increased to 38% of total referrals (Port et al., 2005).

3.6. Summary

This chapter has examined the literature on consanguinity, a presenting issue that provides an opportunity for genetic counsellors to utilise cross-cultural counselling skills. The chapter first explored the global prevalence of consanguinity and consanguinity in Australia, and then reviewed historical and current information on the genetic and health aspects of consanguinity and the socio-cultural issues around the practice. Finally, the chapter explored the provision of culturally sensitive genetic counselling services for consanguineous couples.

This review reveals that studying consanguinity from the perspective of cultural diversity is pertinent because consanguinity is often thought to be a practice restricted to certain ethnic groups. Globally, the practice of consanguineous marriage continues to be an important issue for millions of people, and increasingly couples are migrating to Australia from countries where this is a common practice.
Prior studies of consanguinity tended to focus on the genetic implications of consanguineous marriage, whilst from an overall health perspective consanguinity is a much wider and more complex topic. It is essential to explore socio-economic variables in order to provide culturally appropriate genetic education and counselling programs for consanguineous couples (Bittles et al., 2010a). It is also important to find a balance between understanding the effect of detrimental recessive genes that affect a minority of families against a much larger number of families whose children show no identifiable deleterious biological effects and for whom consanguineous unions offer obvious social and economic advantages.

Previous work has shown that couples presenting for genetic counselling on consanguinity in Australia are often individuals from countries that do not routinely practice consanguineous marriage (Port et al., 2005). Whether couples are from communities where consanguineous marriages are commonly practised or not, the provision of culturally sensitive, client-centred genetic counselling is essential. The counsellor may or may not be of the same ethnic background as the clients but their skills do not preclude the delivery of effective counselling. Genetic counsellors are accustomed to working with clients who have “alternative worldviews” and they are trained to maintain their professionalism even if they hold opposing views on the advisability of consanguineous relationships (Cragun et al., 2009, p.557).

Those that support cultural matching of client and counsellor might argue that to be culturally aware the counsellor would need to have direct experience with consanguinity to fully understand the issues involved. Based on their training and skill set, genetic counsellors have the capacity to treat each client as a unique individual or couple, listening, learning and exploring each story, affording them an opportunity to provide client-centred care for all consanguineous couples irrespective of the counsellor’s background or experience. A more dialogical approach applying the principles of the REM (reviewed in Chapter Two) would allow
engagement with any differences and discussion of them in the light of each party’s cultural beliefs and norms.

Research exploring genetic counselling between persons of different cultural backgrounds, especially focusing on the perspective of the client, is lacking (Lewis, 2002). Greater attention needs to be focussed on the interaction between the counsellor and client because each brings their own unique cultural influences to the interaction (Weil, 2001). By exploring a culturally embedded practice such as consanguinity, both parties participating in genetic counselling and the interaction between them, the research presented in this thesis will address some of the questions raised by recent research and publications. The following chapter contains a detailed outline of the methodology employed in undertaking this research.
4. Methodology

Qualitative research involves an interpretive, naturalistic approach to the world. This means that qualitative researchers study things in their natural settings, attempting to make sense of, or interpret, phenomena in terms of the meanings people bring to them. (Denzin et al., 2011, p.3)
4.1. Introduction

The preceding two chapters have highlighted a number of gaps in knowledge about genetic counselling and the uncommon practice of consanguineous marriage in Australia. A shift in approach to genetic counselling towards a more dialogical framework has occurred in response to increasing complexity in genetic counselling because of increased genetic knowledge, treatment options and social and cultural diversity. A better understanding of the capabilities and constraints of genetic counsellors to engage with their clients (encompassing this more dialogical framework) and the innovations in training and practice required to facilitate this approach is needed.

Little is known about the experiences of genetic counselling clients in consanguineous relationships, or how Australasian genetic counsellors work with consanguineous couples, or indeed whether they recognise this as point of cultural difference. Whilst consanguinity *per se* is not a genetic condition, many clients in consanguineous relationships that I met were profoundly affected by societal views about this type of relationship. I wanted to explore the source of these perceptions, what it was like for those experiencing stigmatisation and understand the role genetic counsellors could play in circumventing the prejudice.

My goal was to ascertain knowledge about these previously unexplored questions that could be directly applied to practice. The role of a genetic counsellor involves allowing the client to tell their story and facilitate understanding of the impact of genetic conditions on their lives.

This chapter discusses the qualitative approach that was developed in this thesis, firstly reviewing how qualitative research has been applied in genetic counselling previously, then exploring how the approach I chose and my professional background impacted on the way the research was carried out, my interpretation of
the data obtained and potentially the way the research participants responded to me as a researcher. Finally, the data collection methodology for each of the three stages of the research conducted is described.

4.2. Qualitative research

The acceptance and application of qualitative methods in genetic counselling reflect the flexibility and detailed nature of these methods. They enable engagement with the nature of the issues that arise which are: complex, dynamic, problematic, previously unexplored and often sensitive (Beeson, 1997; Chapman et al., 2002; Sandelowski et al., 1996). Qualitative methods allow ideas and client narratives to be explored in greater depth than with quantitative instruments alone, and enable the researcher to “achieve a depth of understanding with an emphasis on description and interpretive processes” (Hernandez et al., 2006, p. 359). They are particularly useful when the field of interest or human experience is complex and has not been widely examined (Hodgson et al., 2010; Pope et al., 1995; Smith et al., 2011). Participants are able to tell their own stories and experiences in detail and because both facts and emotions are captured the resulting data are often very powerful. This is increasingly realised as being equally as important as facts and figures in understanding a disease or treatment (Austin, 2010; Charon et al., 2002).

The value of using qualitative analysis and narrative approaches to explore previously unexamined dimensions of the interaction between client and counsellor are also recognised (Abrams et al., 2002; Bernhardt et al., 2000; Biesecker et al., 2001; Butow et al., 2004; McAllister, 2001). As previously discussed, qualitative research methods are important in identifying key issues, experiences and responses of groups of individuals to genetic information and genetic counselling.

The techniques used to examine genetic counselling have varied considerably. Many studies have used conversation, discourse and linguistic analysis (Babul-Hirji et al., 2010; Benkendorf et al., 2001; Hodgson et al., 2010; Lehtinen et al., 2005;
Roberts et al., 2005), grounded theory (Hamilton et al., 2009; McAllister, 2001; McAllister et al., 2008b; Skirton, 2001), inductive thematic analysis (Alliman et al., 2009; Schoonveld et al., 2007; VandenLangenberg et al., 2012), ethnography (McAllister, 2001; Peters et al., 2001a, 2001b; Saleh et al., 2011) and focus groups (McCarthy Veach et al., 2001; Saleh et al., 2009). Other studies have examined verbal communication in simulated counselling sessions using descriptive tools designed to capture counsellor processes (Ellington et al., 2011; Roter et al., 2006). These techniques have been used to build evidence-based theoretical frameworks, and to investigate processes occurring in and around genetic counselling including the effect on clients and their families. These studies are valued for what they can add to the theory, practice and strategic planning (Bernhardt, 2008; Grubs et al., 2010; McAllister, 2001).

4.2.1. A constructivist framework

Epistemology is the relationship between knowledge and how the holder of the knowledge understands it. It seeks to ask “what is the relationship between the researcher and that being researched?” (Creswell, 2013, p.21). Different individuals will have different responses and interpretations of knowledge or information depending on their stance or viewpoint. Hence:

Qualitative research begins with assumptions and the use of interpretive/theoretical frameworks that inform the study of research problems addressing the meaning individuals or groups ascribe to a social or human problem. (Creswell, 2013, p.44)

The theoretical paradigms, or different beliefs, “shape how the qualitative researcher sees the world and acts in it” and have been categorised into four major groups: positivist, constructivist, feminist and critical (Denzin et al., 2011, p.13).

In this study I chose to adopt a constructivist framework because of its good fit for research in the areas of psychology and health sciences. A constructivist framework
emphasises interpretation and subjective observation, with the researcher positioned to reconstruct meaning and the experiences of research subjects (Mills et al., 2006).

This constructivist view also fits well with the guiding principles of genetic counselling that inform my position as a clinical practitioner – that is, every individual has their own unique experience or meaning that they attribute to their situation or experience with a genetic condition. In this study, each couple in a consanguineous relationship will have a different experience of what this has been like and how they have negotiated the associated genetic risk. Each genetic counsellor will also have a different experience of working with consanguineous couples and negotiating cultural difference. When genetic counsellors and clients come together through counselling, each experience will be inherently different because of the many variables present.

4.2.2. Phenomenological framework

Phenomenology is the study of the meaning of lived experiences of a phenomenon for one or more individuals and focuses on the common experiences of study participants (Creswell, 2013; Starks et al., 2007). It is a method commonly used for “describing and interpreting participants’ views” where the “overarching aim is to understand the unique meanings and significance of the phenomena as experienced by the participants” (Smith et al., 2011, p.42). Being able to appreciate the ‘life-world’ or lived experience of participants, and their experience or description of it, is the primary purpose of phenomenology (Johnson et al., 2012).

Bernhardt (2008) has highlighted the important role of qualitative methods in medical genetics research, as they enable us to “listen to the voices of our patients”. These methods can capture a broad view of the phenomenon under examination and look at the experiences of clients on a deeper level:
We may find that that although our patients appear to be doing well, on deeper probing, we learn that they have needed to engage in a great deal of emotional or spiritual work in order to be whole and psychologically fit. (Bernhardt, 2008, p.3132)

More specifically, in relation to the method of phenomenology:

Through close examination of individual experiences, phenomenological analysts seek to capture the meaning and common features, or essences, of an experience or event. (Starks et al., 2007, p.1374)

In phenomenology, researchers attempt to get as close as possible to an understanding of the way participants experience the phenomena under study without experiencing it themselves. They immerse themselves in the data but in doing so are expected to practice the reflective practice of “bracketing” where they “recognise and set aside (but do not abandon) their a priori knowledge and assumptions” (Starks et al., 2007, p.1376). Van Manen (2011) reminds us that multiple explanations and interpretations of the data are possible:

Phenomenological inquiry-writing is based on the idea that no text is ever perfect, no interpretation is ever complete, no explication of meaning is ever final, no insight is beyond challenge. It behoves us to remain as attentive as possible to the ways that all of us experience the world and to the infinite variety of possible human experiences and possible explications of those experiences. (van Manen, 2011, n.p.)

Phenomenological inquiry-writing is thus an insightful approach to adopt for this thesis research, which is seeking to develop a deeper understanding of the experiences of cousin couples living in a consanguineous relationship and the experience of genetic counselling for clients and genetic counsellors negotiating cultural diversity.
4.2.3. Reflexivity

Researchers using qualitative methods are often closely engaged with study participants and because of this they need to reflect on how their backgrounds influence their observations (Jacobsen, 2012). Grubs et al. (2010) outline the importance of acknowledging the background, beliefs and values of the researcher undertaking qualitative analysis:

Knowledge is constructed by human beings out of their individual and shared understandings of the world. Within this perspective, the researcher is obliged to strive not for objectivity but for ever-increasing awareness of her/his own assumptions, preconceptions, beliefs and values. This constellation of qualities along with race, gender, and class comprise the position from which a researcher sees the phenomenon under study. An interpretive approach acknowledges that positionality is inevitable and when informed by an in-depth understanding of theoretic literature and professional experience, it contributes to the researcher’s theoretical sensitivity. (Grubs et al., 2010, p.102)

This thesis research arose out of my interest in conducting applied research and exploring the social and ethical implications of genetic counselling. I came to the area of consanguinity because I was involved in a research project investigating the demographic factors and genetic outcomes of consanguineous couples presenting to our genetic service.

To reflect these dual roles, the way in which I came to this research topic, and my continuing practice and interests particularly in the psychosocial issues of genetic counselling, I am describing myself as a ‘practitioner-researcher’. Borrowed from Grubs and Piantanida (2010), the term practitioner-researcher in my research implies an interpretative research orientation with the aim of gaining a deeper understanding of consanguineous couples’ experiences, the nature of the counsellor-client relationship, and the nature and effectiveness of communication between counsellors and clients.
The concept of overlapping and dual roles is being explored in the profession, as a growing number of genetic counsellors are becoming involved in clinical research and undertaking doctoral studies (Wallace et al., 2008). In Australasia the ethical challenges arising from combining the roles of practitioner and researcher have been discussed at the HGSA Annual Scientific Meetings in 2005 and 2008 and a subsequent publication and PhD thesis. Young et al. (2005) explored the ethical challenges that arise when conducting research interviews and experiencing the urge to step into the role of genetic counsellor. Young (2011, p.07.2) describes the roles of researcher and genetic counsellor as “overlapping and complementary” and suggests that genetic counsellors’ training and experience makes them “well placed to undertake qualitative research in genetic counselling”. In 2008, McEwen explored the impact of negotiating the dual roles of practising genetic counsellor and clinical researcher using an insider–outsider framework, concluding that it is vital for genetic counsellors undertaking research to be aware of the tensions and have strategies to manage them (McEwen, 2008).

Developing this awareness of my dual roles of researcher and genetic counsellor was an important step that influenced the study design, conduction and analysis. My knowledge and skills are a result of my professional background and experience, and are intertwined with personal life experience. I am a HGSA certified genetic counsellor with 16 years of experience counselling individuals and families about a range of genetic conditions at a range of life stages. Outside of my work I am married (not to my cousin) and have two school age children.

4.2.4. Insider–outsider continuum

In exploring any phenomenon, question or situation, there are always two views: the insider’s view and the outsider’s view. In the case of research, the researcher is the outsider whose intention is to understand or describe the phenomenon under study. The insiders are the participants who are “living with the problem and have a unique understanding of it” (Streubert et al., 2011, p.310).
The “outsider-alongside” concept developed by McEwen (McEwen, 2011) fits with my position in researching how genetic counselling engages with consanguinity: I am not married to my cousin, but I am a genetic counsellor working with couples who are. Here a major challenge arises in reconciling my dual roles as genetic counsellor and researcher. Following an interview schedule and asking questions, for instance, is straightforward but it can be difficult at times to avoid responding as a counsellor would. However, I feel that counselling skills can enable an interviewer to respond with empathy and understanding and to probe certain issues further. In my research, this could be a positive outcome for the participants, creating a more collaborative environment where they feel comfortable and willing to reveal details about their experiences. I recognise that being an engaged practitioner inevitably affects involvement in research interviews and influences interpretation of the data; however, a positional engagement with the issues could be seen as a positive factor rather than a hindrance. It is this interpretive approach that adds richness and value to the data (Grubs et al., 2010; Young, 2011).

I examined my own views about consanguinity through supervision and discussion with colleagues when, during an interview with a journalist, I became uncomfortable when asked about the type of people who marry their cousin. I replied “people like you and I” meaning to infer ordinary people. I was concerned that he would use the quote, leading readers to assume that I was married to my cousin and this was the reason I was researching in this area. He then compared cousins who marry and “normal people”, which increased my level of discomfort as I viewed consanguineous couples as entirely normal people. I did not have negative feelings about consanguinity, so was surprised by my concern about being condemned by society, reflecting the condemnation that my consanguineous clients experienced. This countertransference of discomfort could potentially affect the way that I interacted with clients in a counselling session. I concluded that it did not,
as I displayed empathy when acknowledging the negative reactions couples encountered and maintained my neutral stance on the issue of consanguinity.

In relation to the genetic counselling aspect of the research, I occupied a different position on the ‘insider–outside continuum’: more of an insider–alongside. As a practising genetic counsellor and the colleague of many of the genetic counsellor participants, I often felt the tension arising from analysing the work of my colleagues. On the other hand, exploring the practice of genetic counsellors from the perspective of one who understands the inner working of the profession added depth that may not have been present if it was an outsider conducting the research.

4.3. Method

The previous sections of this chapter have shown the utility of qualitative research in examining process issues in genetic counselling and highlighted the benefits of qualitative research that involves phenomenological inquiry-writing and reflexivity, and is conducted with a sophisticated awareness of the tensions and ethical issues that confront researcher-practitioners.

As the methodology for this thesis research was developed, it was apparent that the research questions would be best addressed by examining the perspective of both individuals in consanguineous relationships, and of genetic counselling providers. From there, a natural progression was to explore the interaction between the two during live genetic counselling consultations. This lead to the development of the three research stages each designed to examine these three perspectives. Stage 1 of the study examined the perspective of individuals in consanguineous relationships, some of whom were clients of genetic counselling. Stage 2 examined the perspective of genetic counselling providers and Stage 3 explored the interaction between the two during live genetic counselling consultations.
In each of the three stages of this research, previously unexplored phenomena were investigated using both quantitative and qualitative data collection methods. Combining quantitative and qualitative methods provides “access to different levels of knowledge” and “can help to build a wider picture” (Pope et al., 1995, p.44). The collection of data by different techniques enabled comparison of data sources (triangulation) and an audit trail consisting of documentation and an account of the process by which the research was conducted provides the opportunity for external audit and checking for consistency and confirmability (Mays et al., 2000). In addition to the previously discussed issues of reflexivity and the theoretical framework of the study, these design quality criteria were applied during the research.

Quantitative methods were used to recruit participants from the participant pool (a difficult target group in the case of consanguineous couples), obtain preliminary data and inform the development of the interview schedule. Qualitative methods facilitated exploration of previously unexplored topics in greater depth and this was undertaken within the framework of phenomenology.

4.4. Research plan

As explained above, the collection of data took place in three stages and multiple sources of evidence (questionnaires, interviews, discussion group and transcripts of genetic counselling sessions) were used to collect data from participants. The stakeholders and data collection methods are outlined in Table 4.1. The results of each stage of the research will be presented separately in Chapters Five, Six and Seven.
Table 4.1  Stakeholders and data collection methods

<table>
<thead>
<tr>
<th>Stage</th>
<th>Unit</th>
<th>Stakeholder</th>
<th>Data collection method</th>
<th>Chapter</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Client</td>
<td>Consanguineous couples</td>
<td>Questionnaire followed by semi-structured interviews</td>
<td>5</td>
</tr>
<tr>
<td>2</td>
<td>Provider</td>
<td>Genetic counsellor</td>
<td>Questionnaire followed by discussion group</td>
<td>6</td>
</tr>
<tr>
<td>3</td>
<td>Client &amp; provider</td>
<td>Clients and GHPs</td>
<td>Genetic counselling session</td>
<td>7</td>
</tr>
</tbody>
</table>

For all the data generation methods, the ethical issues that may be encountered were considered. Applications for ethics approval were made both to the Ethics Committees of the Women’s and Children’s Health Service, Department of Health WA (Hospital Ethics Committee), and the Human Research Ethics Committee of Murdoch University (Murdoch Ethics Committee). The study was approved by both of these committees (King Edward Memorial Hospital for Women registration number 812/EW and Murdoch University permit number 2002/277). In addition, the ASGC Executive Committee granted permission to use the email listserver to contact and invite genetic counsellors to participate in the research. An application to amend the study by adding the recorded genetic counselling sessions for Chapter Seven was later submitted to both the Hospital and Murdoch Ethics Committees and approved under the same registration numbers cited above.

4.4.1. Stage 1 – questionnaires and interviews with people in consanguineous relationships

This first stage of the research commenced with a postal questionnaire administered to individuals or couples in a consanguineous relationship. A sub-group of participants who completed the questionnaire and expressed a willingness to participate in further research were then interviewed. The interviews provided an opportunity to address the questionnaire themes in greater depth. The recruitment procedures are outlined in Figure 4.1 and the procedures used to administer the questionnaires and interviews are explained in detail below.
4.4.1.1. Questionnaire design

A postal questionnaire was developed to obtain information about demographics and general information about respondents’ consanguineous relationships, risk perception and genetic counselling experiences. The questions were developed based on a literature review, my clinical experience and input from my supervisor and a nurse researcher on the Hospital Ethics Committee. The nurse researcher provided feedback on the draft questionnaire submitted with the ethics application and on the basis of this feedback, minor revisions to the wording and design of some questions were made. The Murdoch Ethics Committee made no suggestions.
for changes to the questionnaire design and content. An experienced health professional completed the survey to check for ease of comprehension and time taken to complete the questionnaire and no further revisions were made.

The questionnaire (Appendix A) contained both open-ended and closed questions as well as questions using a Likert scale. The opportunity to provide written comment was given for several items. The questions were designed to elucidate participants’ experiences of being in a consanguineous relationship, thoughts about genetic risk and whether or not genetic counselling changed their anxiety about genetic risk.

4.4.1.2. Identifying potential participants

Between 1975 and 2004, 286 couples presented to GSWA for advice about their consanguineous relationship (Port et al., 2005). These couples were the target population for this study. Twenty four cases were excluded due to death (themselves or partner), adoption, incest or having a new partner. The remaining 262 clients were invited to participate in the study. The Hospital Ethics Committee stipulated that two groups be created and two different approaches to recruitment applied depending on the length of time since their genetic counselling occurred. Group A comprised 122 clients who were counselled prior to 1993 and group B comprised 140 clients counselled between 1993 and 2004.

A press release (Appendix B) and subsequent media articles (newspaper, television and radio), and a posting about the research on a website for cousin couples (http://www.cousincouples.com/), were used to recruit participants who had not attended GSWA for genetic counselling (Group C). The press release and subsequent media interest generated what could be seen as a negative side effect: the media requested the researcher provide contact details for consanguineous couples (so that they could be asked to participate in media interviews). These media requests were declined.
Sixty individuals interested in participating in the study contacted me following the press release, media articles and website posting; however, 24 were excluded because they were children or grandchildren of consanguineous couples. The remaining 36 individuals were included in the study.

4.4.1.3. Administering the questionnaire

As stipulated by the Hospital Ethics Committee, Group A clients were afforded an ‘opt-in’ approach. In July 2003 the GSWA Head of Department sent a letter inviting them to participate in the study (Appendix C). The letter provided preliminary information about the study and a form (and reply-paid envelope) with the options of indicating: a willingness to participate; obtaining more information about the study; or they did not wish to participate. In comparison, the Group B clients were afforded an ‘opt-out’ approach. In July 2003 they were sent a letter from the GSWA Head of Department (Appendix D) with some preliminary information and a form (with reply-paid envelope) to be returned if they did not wish to participate or wanted more information before deciding. If the form was not returned within three weeks from the date of dispatch a questionnaire and a consent form (Appendix E) was mailed out. Participants in Group C were sent a questionnaire and consent form by mail.

4.4.1.4. Data analysis

Table 4.2 details the number of questionnaires distributed and returned within each of the three study groups. 298 questionnaires were distributed and 69 questionnaires were returned giving an overall response rate of 23.2%.
Table 4.2  Distribution and return of questionnaires

<table>
<thead>
<tr>
<th>Questionnaires</th>
<th>Group A (pre 1993 opt-in)</th>
<th>Group B (post 1993 opt-out)</th>
<th>Group C (press release, media articles, website posting)</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Distributed</td>
<td>122</td>
<td>140</td>
<td>36</td>
<td>298</td>
</tr>
<tr>
<td>Returned</td>
<td>15</td>
<td>27</td>
<td>27</td>
<td>69</td>
</tr>
<tr>
<td>Response rate</td>
<td>12.3%</td>
<td>19.3%</td>
<td>75%</td>
<td>23.2%</td>
</tr>
</tbody>
</table>

Responses to the closed questions were coded and entered into an Excel™ spreadsheet for collation and analysis. Descriptive statistics (mean, median, percentages) were calculated for responses to these items. Responses to open-ended questions were coded and related codes grouped together into conceptually similar themes.

4.4.1.5.  Interviews

In-depth interviewing was used as a means of exploring the phenomenon of consanguinity from the perspective of consanguineous couples, examining their lived experience and allowing them to describe their experiences in their own words (Liamputtong, 2009; McAllister, 2001). This method was considered appropriate, as little is known about consanguineous couples in WA and the study was exploratory in nature.

In addition to exploring the lived experiences of people in consanguineous relationships, the interviews also provided an opportunity to investigate the nature of communication between genetic counsellors and their clients. The interviews focussed on the issues arising directly from consanguinity, and more generally on how clients feel about communication of information and the perceived existence and relevance of cultural issues.

The interviews gave an opportunity to explore in greater detail some of the points that may have been missed by the questionnaire, which restricted participants to abbreviated responses to standardised questions. Participation in the questionnaire prior to the interview afforded participants the opportunity to reflect on or discuss
topics or issues raised prior to attending the interview, thereby adding depth to the data. The interviews were a therapeutic experience for some participants, providing them with an opportunity to tell their story and share their lived experience. As the potential therapeutic benefit of research participation is not often considered a primary aim of research, it is rarely recognised or formally reported (Lakeman et al., 2013). Participants have different motivations for engaging with research studies and the potential perceived benefits should be considered a possible means of mitigating any risks of participating (Clark, 2010).

4.4.1.6. Designing the interview schedule

The interviews were conducted after the questionnaire data was collected, allowing participant responses to inform the interview schedule. As there were no similar studies in the literature and therefore no validated research tool, the questions were designed to address the aims of this study. The development of the tool was guided by an appreciation of the questionnaire data, my clinical experience and input from experienced GSWA clinical staff, my supervisor and members of the Hospital Ethics Committee.

The use of a prepared interview guide allowed for a semi-structured interaction while covering the same general topics and questions with each participant (Johnson et al., 2012). The interview schedule included open-ended questions designed to allow exploration of issues arising from the questionnaire in greater depth. This allowed participants to introduce new topics and the interviewer to follow emergent themes. The 36 questions were designed to guide the participant through a standardised set of questions relating to specific areas of interest (Appendix F). Following a short pilot study of five interviews, additional questions (numbers 12, 16, 20 and 26) were incorporated into the final schedule.

The interview schedule invited participants to talk about their experience of beginning and continuing their consanguineous relationship. The preliminary
questions were designed to put people at ease and allow them to tell their story. A series of questions about the relationship and their experiences of being in a consanguineous relationship, and finally questions about genetic counselling, followed. These were deliberately placed last so that by this time the participants were sufficiently at ease (Bowling, 2005).

4.4.1.7. Identifying potential participants

A form that invited participants to take further part in the research by participating in a recorded interview was attached to the end of the postal questionnaire. Participants were also offered contact by a genetic counsellor if they wished to arrange an appointment. To maintain the respondents’ anonymity, the form was removed and stored separately upon receipt of the questionnaire. Of the 69 participants who returned questionnaires, none requested genetic counselling and 45 indicated they were willing to participate in a recorded interview. Limited resources prevented all 45 of these volunteers being interviewed. I was not the genetic counsellor for any of the interviewed couples.

Between January and April 2005 respondents were approached sequentially (in order of receipt of questionnaire) by telephone confirming their willingness to be interviewed and the interview was arranged at a mutually convenient time and place. English speaking individuals or couples that were, or had been, in a consanguineous relationship were included. Individuals and couples that had been counselled by the researcher (n=7) or resided at a prohibitive distance from the Perth metropolitan area (n=12) were excluded. Ten respondents were not contactable or a suitable time for an interview arranged.

In total, 16 interviews with 14 individuals (all female) and two couples were completed over a 10 month period. All interview participants were, or had been, in a relationship with their first cousin for a period of time. Fifteen were married (one widowed) and one had been in a de facto relationship. Nine of the 16 interview
participants had been for genetic counselling. Several others interpreted genetic counselling as medical advice obtained from other medical professionals and answered the questions in this light. The median time between genetic counselling and the research interview was four years (range 3 to 30).

4.4.1.8.  Conducting the Interviews

Interview participants were offered the option of having the interview at GSWA or in their home. Half the participants attended GSWA and half were interviewed in their home. The flexibility in location was designed to minimise inconvenience and maximise participants’ comfort. Interviews lasted between 45-90 minutes and were, with the participant’s written consent, audio-recorded using a hand held tape recorder. The tapes were stored in a locked cabinet in a locked office and electronic data was stored on a password protected computer.

During the interviews, replies to fixed response items were documented and occasionally notes on responses to open-ended items were taken. At the end of the interview the respondents were invited to raise any other points they felt were important that had not been covered by the questions and discussion. Provision was made for a genetic counsellor (other than the researcher) to see participants if any issues arose during the interview. Follow up information was offered to one participant regarding pregnancy risk.

After the interview a letter of thanks and a copy of the interview transcript were sent to participants. Participants were asked to confirm that the transcript was an accurate description of their thoughts and feelings, invited to write down any additional thoughts and reminded that further genetic counselling was available. No additional comments or written material were received.
4.4.1.9. **Data analysis**

Thematic analysis of interview responses was performed using the approach outlined by Braun *et al.* (2006). This analysis allowed the participants’ experiences, of being in a consanguineous relationship in a society where this is not a commonly practised type of union, to be represented as accurately as possible.

Thematic analysis is a method used to identify, describe, analyse and report themes and patterns within data (Braun *et al.*, 2006). This method was selected for analysis of the interview data because it allows meaning to emerge by eliciting themes and using them to make sense of the data (Liamputtong, 2009). Thematic analysis is “an empirically driven approach for detecting the most salient patterns of content” and is “best suited to elucidating the specific nature of a given group’s conceptualisations of the phenomenon under study” (Joffe, 2012, pp 220 & 212). This type of qualitative research can uncover unexpected experiences of the research participants, as it is they themselves who provide the direction of the study (Douglas *et al.*, 2009).

The interviews were manually transcribed verbatim and checked for accuracy by repeatedly listening to the recordings whilst reading the transcripts. During transcription, all names were replaced with pseudonyms and any potentially identifying information (names and places) replaced with symbols (Table 4.3). Even though it is time consuming, it is recommended that researchers transcribe their own data because transcription is a form of initial data analysis (Liamputtong, 2009). Each transcript was read multiple times, absorbing the content and concepts being conveyed. Immersion in, and intimate knowledge of, the data allows the maximum expression of the analytical skill of the researcher and takes the analysis beyond simply describing data (Beeson, 1997; Pope *et al.*, 2000).
The transcripts were analysed by reading and making notes about the areas of similarity and difference, and common themes were identified using an inductive approach. These common themes generated codes which were assigned to the relevant sections of the transcripts. When appropriate, the same unit of text was included in more than one code. To further enhance rigour and check for consistency, 50% of the de-identified transcripts were read and coded independently by a genetic counsellor experienced in performing thematic analysis. Grubs et al. (2010) support this approach, arguing that a data auditor should read and code at least 10-20% of data is read and coded by an auditor to check for multiple interpretations of this data.

When all transcripts were coded and a tabulated list of codes had been generated, the codes were grouped together based on their conceptual similarity. As the transcripts were analysed, review and modification of the names and groupings continued until all were categorised in a manner that best reflected the data. The data auditor and I discussed and reached consensus on the naming and grouping of the codes and the emerging themes. Differences within and between the codes were continually re-examined. Themes were then developed inductively and each

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**Table 4.3** Transcription key

<table>
<thead>
<tr>
<th>Symbol</th>
<th>Meaning</th>
</tr>
</thead>
<tbody>
<tr>
<td>Country</td>
<td>Country named</td>
</tr>
<tr>
<td>State</td>
<td>State identified</td>
</tr>
<tr>
<td>(.....)</td>
<td>Unfinished overlapping speech</td>
</tr>
<tr>
<td>(.)</td>
<td>Untimed brief pause</td>
</tr>
<tr>
<td>(2.0)</td>
<td>Longer pause estimated to one tenth of a second</td>
</tr>
<tr>
<td>(words)</td>
<td>Material added by author for clarification</td>
</tr>
<tr>
<td>...</td>
<td>Non-relevant material removed to aid coherence</td>
</tr>
<tr>
<td>DR</td>
<td>DR named</td>
</tr>
<tr>
<td>&lt;laughs&gt;, &lt;cries&gt;</td>
<td>Body language, gesture or other non-verbal</td>
</tr>
<tr>
<td>&lt;pause&gt;</td>
<td>brief pause</td>
</tr>
</tbody>
</table>

---
was named based on the major concept illustrated. Towards the end of the analysis no new codes or themes were emerging, indicating that data saturation had been achieved (Braun et al., 2006).

4.4.2. Stage 2 – genetic counsellors questionnaire and discussion group

The second stage of the research involved a survey of Australasian genetic counsellors and a discussion group. The survey, a structured postal questionnaire, explored genetic counsellors’ opinions on cultural difference and consanguinity. The discussion group explored genetic counsellors’ ideas about the effect of cultural difference on their work, how they negotiated diversity issues and whether training in cultural competency was adequate. The recruitment procedures followed during Stage 2 are outlined in Figure 4.2 and procedures used to administer the questionnaires and conduct the discussion group are explained in detail below.

![Figure 4.2  Stage 2 methodology flow chart](image)
4.4.2.1. **Questionnaire design**

A postal questionnaire was developed to obtain information about demographics of Australasian genetic counsellors and their opinions and practice relating to cultural difference and consanguinity. The questions were based on a literature review, my clinical experience and input from my supervisor and a nurse researcher on the hospital ethics committee. The nurse researcher provided feedback on the draft questionnaire submitted with the ethics application and on the basis of this feedback minor revision to the wording and design of some questions were made. The Murdoch Ethics Committee made no suggested changes to the questionnaire design and content. The questionnaire was then piloted with 4 genetic counsellors who were recently graduated and did not participate in the study.

The questionnaire (Appendix G) contained both open-ended and closed questions as well as questions using a Likert scale. Several items included an opportunity to provide written comment. The questions were designed to elucidate counsellors’ experiences in counselling clients in consanguineous relationships and whether or not they modified their genetic counselling in response to cultural diversity.

4.4.2.2. **Identifying potential participants**

Two hundred members of the ASGC were eligible to participate in the study. Emails were sent to each member, via the ASCG list server, with information about the study. In addition, an article about the study was written in the ASCG newsletter *Linkage*. Recipients of the email and Linkage readers were provided with an information sheet about the study (Appendix H) and asked to reply by email and include their postal details if they wished to participate.

4.4.2.3. **Administering the questionnaire**

In April, July and November 2003 questionnaires and consent forms were mailed to the counsellors who consented to participate. An email reminder was sent to those who had not yet returned questionnaires. Sixty one genetic counsellors responded
by email and 58 completed questionnaires were returned giving a response rate of 29%. Upon receipt of the questionnaire, the declaration section was removed and stored separately to maintain the respondents’ anonymity.

### 4.4.2.4. Data analysis

Responses from the completed questionnaires were coded and entered into an Excel™ spread sheet. Descriptive statistics (mean, median, range and percentages) were calculated for responses to fixed items. Written comments with similar content were grouped into categories allowing recurrent themes to be identified (Giarelli et al., 2003).

### 4.4.2.5. Discussion group

The discussion group was held during the 2004 annual HGSA/ASGC conference program. At the conference, I presented an oral paper entitled “The dialogical model of genetic counselling and transcultural interactions in Western Australia: a quantitative and qualitative study”. At the end of my presentation I explained the purpose of the discussion group and invited members of the audience to attend. The discussion group methodology and data analysis is described below.

### 4.4.2.6. Planning and conducting the discussion group

The discussion group for genetic counsellors was conducted after the analysis of the data from the genetic counsellors’ questionnaire. Counsellors’ responses to the questionnaire were used to inform the planning of the discussion group themes and prompts. The discussion group was designed to explore genetic counsellors’ awareness of cultural diversity and their own cultural background, consanguinity as an example of cultural difference, how cultural difference affects the practice of genetic counselling, the resources needed to improve engagement with diversity and ideas for future research. This was explained to participants at the beginning of the discussion group session.
A convenience sample of seven genetic counsellors participated in the discussion. Descriptions of the participants are included in the results section of Chapter Six (Table 6.6).

The discussion group took place in a meeting room at the conference venue. It began with an introduction which described the purpose of the research. The specific aims of the discussion group were also clearly explained to the participants. A warm up exercise, designed to initiate thinking and discussion about the question topics, involved participants writing down their answers to the question – *Tell me about yourself and your cultural background?* This was followed by a brain storming exercise with participants writing down “*The first thing that comes to mind when you think about consanguinity.*” The discussion then moved on to the specific questions, which are listed in the discussion group program in Appendix I. These questions were raised to guide and prompt the discussion, leading to in-depth discussion around the general topics.

4.4.2.7. Data analysis

The hour-long discussion group was audio-taped and transcribed verbatim. Anonymity was maintained by assigning each participant a genetic counsellor number and removing all references to location of practice and ethnicity. Both location of practice and participant ethnicity could be relevant and indeed important in the context of a discussion about consanguinity and genetic counselling, but the small number of participants meant that maintaining anonymity outweighed any benefit of including this information. Minor corrections to the participants’ quotes were made to improve clarity. Disfluencies such as “um” and “ah” were removed and where meaning was not altered, some quotes were truncated for ease of reading and is denoted in the quotation by “...”.

Analysis of the transcript from the discussion was reviewed using interpretive content analysis responses (Braun et al., 2006; Giarelli et al., 2003). This enabled
topics and responses to be organised into common themes, based on their conceptual similarity, and then named in a way that best captured the theme.

4.4.3. Stage 3 – genetic counselling consultations

Having explored the perceptions of consanguineous couples and providers of genetic counselling, the third stage of the research involved the interaction between clients and providers of genetic counselling. The two step approach of this part of the study (recorded counselling sessions followed by post-session questionnaires) is outlined below.

Kessler (1992a) first argued that observation studies of this nature are required. Biesecker et al. (2001, p.191) suggested the need for process studies to “provide a glimpse into the black box” of genetic counselling. Since then, some researchers have provided a glimpse into this previously unknown world. Gale and colleagues (Gale et al., 2010) used Interpersonal Process recall (IPR) following video recorded genetic counselling sessions to explore information provision. This is a method that has been used in training and professional development to facilitate reflective practice, and aspects of this technique have been incorporated into current certification requirements for Australasian genetic counsellors. Bylund et al. (2012, p.293) recorded genetic counselling sessions to explore how genetic counsellors communicated uncertainty to mothers at risk of BRCA1/2 mutations and commented that “rarely have studies captured the actual conversations (genetic counselling sessions) in which this uncertainty emerges”. These studies highlight the value of analysing actual genetic counselling sessions to provide data about how this complex activity is undertaken.

Other qualitative methods of exploring the conversations between GHPs and clients include discourse analysis (Hodges et al., 2008). This approach can be used to analyse a wide variety of data sources and determine the nature of conversations between health professionals and patients. It has been applied to various types of
professional-client interactions in the general health field and has also been used in
the analysis of genetic counselling sessions (Benkendorf et al., 2001; Brookes-
Howell, 2006; Lehtinen et al., 2005; Roberts et al., 2005; Roter et al., 2008).
Although the methodology is relatively unstructured, it allows for analysis of the
conversation or sequential turns of talk in the interaction, takes into account the
sociological uses of language and can also examine the extent to which
conversations between GHPs and their clients are influenced by cultural factors
(Hodges et al., 2008).

4.4.3.1. Genetic counselling consultations

Live genetic counselling sessions were audio-recorded and analysed, and the
sessions reflected the variety of cases presenting to all sections of our service. They
included familial cancer counselling, pre-symptomatic genetic testing and maternal
age counselling. In addition to examining the conversations that took place during
the counselling interactions, post-session questionnaires explored the interaction
from the perspective of both the genetic counsellor and the client.

4.4.3.2. Recruiting participants and recording

I approached the director of GSWA and obtained permission to conduct this stage
of the study. In April 2005, an expression of interest was circulated to the
 geneticists and genetic counsellors employed at GSWA. A memorandum (Appendix
J) was sent to 12 colleagues inviting them to participate in this exploratory phase of
my research. All five geneticists and five of the seven genetic counsellors replied.
Nine of the 10 respondents agreed to participate. Two colleagues did not respond.

The process for recruitment of GSWA counselling clients adhered to the Hospital
Ethics Committee’s stipulation that the researcher must not have any contact with
them. The approach was to be made by the genetic counsellors who recruited
participants from their client caseload and provided them with information about
the study. Once the client had given verbal consent, the counsellor informed the
researcher they had recruited a counselling client and when the clinic appointment was scheduled. Potential participants were then sent written information about the study and a consent form (Appendix K).

Of the nine colleagues who volunteered to participate in the study, eight recruited clients and recorded the eight consultations (Table 4.4). Sessions were conducted either by a clinical geneticist or genetic counsellor, with the exception of one case where both were present. The researcher had no contact with the clients prior to, and was not present during, the genetic counselling sessions. The GHP conducting the counselling session was given a hand-held digital recording device and operational instructions. In one session, the recording equipment malfunctioned resulting in a partial recording. After completion of the session, digital files were downloaded and stored on the department network drive prior to transcription using Dragon software™. This software was only available on a two computers in the department. Transcription could only be carried out on these two computer terminals after office hours and the digital data had to be stored on the network until then. Despite being stored on a network that was backed up daily, the digital sound files of four of the other sessions were lost from the department network as a result of a malfunction during an upgrade of the hospital computer operating system. These five sessions were excluded and the remaining three genetic counselling sessions were analysed. Table 4.4 summarises these sessions and highlights the three sessions that were analysed.
Table 4.4 Genetic counselling sessions and transcript analysis

<table>
<thead>
<tr>
<th>Session</th>
<th>Presenting issue</th>
<th>Client</th>
<th>Counselling by</th>
<th>Analysed</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Pre-pregnancy counselling for consanguinity</td>
<td>Couple (2nd cousins)</td>
<td>Male geneticist</td>
<td>Recording lost</td>
</tr>
<tr>
<td>2</td>
<td>Predictive testing for BRCA</td>
<td>Young woman</td>
<td>Male genetic counsellor</td>
<td>Recording lost</td>
</tr>
<tr>
<td>3</td>
<td>Breast cancer diagnostic testing</td>
<td>Married woman with children</td>
<td>Female geneticist</td>
<td>Recording lost</td>
</tr>
<tr>
<td>4</td>
<td>Predictive testing for HNPCC</td>
<td>Teenager</td>
<td>Female genetic counsellor</td>
<td>Recording lost. Partial transcript made</td>
</tr>
<tr>
<td>5</td>
<td>Predictive BRCA testing</td>
<td>Middle aged woman</td>
<td>Male geneticist</td>
<td>Full transcript</td>
</tr>
<tr>
<td>6</td>
<td>Maternal age</td>
<td>Married couple</td>
<td>Female genetic counsellor</td>
<td>Full transcript</td>
</tr>
<tr>
<td>7</td>
<td>CF carrier testing</td>
<td>Young married women</td>
<td>Female genetic counsellor</td>
<td>Full transcript</td>
</tr>
<tr>
<td>8</td>
<td>Pre-pregnancy counselling for Klinefelter syndrome</td>
<td>Young couple</td>
<td>Male geneticist and female genetic counsellor</td>
<td>Partial recording</td>
</tr>
</tbody>
</table>

4.4.3.3. Data analysis

The recorded sessions were transcribed verbatim by the researcher. Pseudonyms were introduced and any identifying details changed to preserve anonymity. First impressions of the structure, content and interactions of the sessions were made as the recordings were listened to a number of times. The transcripts were then read multiple times to appreciate the content of the sessions in terms of the general nature of the conversation, and to determine whether the counsellor verbalised the aims for the session. The analysis began by identifying any sequences in the data that demonstrated the negotiation of cultural factors in the dialogue and grouping related themes together. These were analysed to explore whether GHPs were aware of any cultural factors and if so, the techniques they use to work with them.
4.4.3.4. **Post-session questionnaires**

The post-session questionnaires contained a mixture of closed and open-ended questions relating to the perceived reasons for seeking genetic counselling and other issues, such as how time was spent during the session, understanding and satisfaction with the information presented impressions of the counsellor or client, perceptions of cultural background and barriers to communication.

On the day of the scheduled clinic appointment, I provided the recruiting GHP with two sealed envelopes containing the questionnaires that were to be completed after the consultation was recorded. One envelope contained the post-session questionnaire for the genetic counsellor or geneticist conducting the session to complete (Appendix L). The other contained the questionnaire that was to be given to the client at the end of the clinic appointment (Appendix M).

Client and counsellor responses to matching questions were analysed to determine whether perceptions of the two parties differed. Of particular interest were whether: (i) client expectations were met and (ii) the cultural background of client and counsellor affected outcomes. Whether individual style or attributes of the counsellor affected the general experience of participants was examined using a series of questions about client impressions of the GHP and session.

4.5. **Summary**

This chapter has described the three stage research plan, developed in response to the lack of knowledge about consanguinity in Australia and how genetic counsellors in Australasia work with couples in consanguineous relationships. The framework and data collection methods that underpinned the research were explored, along with the need for researcher reflexivity. Also described was the use of quantitative methods to complement the qualitative data. The methods used to generate the data sets for the three stages of research have been described in detail. The findings of each stage of the study are presented in Chapters Five, Six and Seven.
5. **Lived experiences of consanguineous couples**

The poor love she wore this beautiful ring on her right hand for as long as we ever knew her ... You know she had been left to look after her mother and he had been left to look after his mother and when they both finally passed over, they were married. I mean they would have been engaged probably for at least 30 years you know while they were waiting ... The family were just ‘Nope no way’. And they both stayed true to each other. They weren’t interested in anybody else. (Doreen)
5.1. Introduction

In this chapter I examine the lived experiences of individuals in consanguineous relationships in WA. Using questionnaires and interviews, their experiences and perceptions of genetic risk and genetic counselling, and community reactions to their relationship, are explored. The chapter also examines how their cultural framework affected the genetic counselling information that was received, interpreted and applied, and their perception of the support received from genetic counsellors, other health professionals and the wider Australian culture.

Previous research regarding the genetic implications of consanguinity and genetic counselling has focussed on cultures where the practice of consanguineous marriages is common. In WA, cousin couples tend to come from cultural backgrounds where consanguineous marriage is not commonly practised (Port et al., 2005). As a result, there is little or no information about Australian consanguineous couples’ experiences of genetic counselling or being in a consanguineous relationship, with respect to the attitudes of family and community. The effects on the couple and family of the social stigma attached to what Western society often views to be an incestuous relationship is poorly understood. Consanguineous couples often suffer because of societal prejudices when this is not an accepted marriage practice. There is also limited information on the attitudes of genetic counsellors and the wider Australian community towards consanguinity and the scope for genetic counsellors to support consanguineous couples. In Australia, genetic counsellors need to manage the challenge of ‘cultural diversity’ of consanguinity both as a practice associated with particular immigrant communities and more often from a wide range of backgrounds. The needs of these clients extend beyond accessing basic scientific information about their genetic risk and require counselling that is empathetic towards their social and family situations, and provides some tools for coping with these situations. In the absence of guiding literature on genetic counselling for these couples (other than the NSGC
guidelines), exploring the experiences of consanguineous couples and their genetic counselling experiences should better inform practice.

5.1.1. Aims

The aim of this phase of the research was to explore the gaps in knowledge about the lived experience of consanguineous couples, the issues they deal with in relation to their families, social interactions and genetic risk and their experiences with the genetic counselling interaction. Consanguineous couples who received genetic counselling were compared with couples who did not, to examine whether risk perception and the experiences of dealing with family and societal reactions were influenced by receiving genetic counselling. Issues explored that related to consanguineous relationships and experiences of genetic counselling included positive and negatives aspects of experiences with genetic counselling, risk perception, information received from genetic and other health professionals, and whether counsellors from a background in which consanguinity is socially unacceptable experience difficulties in giving objective and nondirective advice to consanguineous couples.

5.2. Methods

The methods are outlined in detail in Chapter Four. In summary, people in consanguineous relationships were recruited from a cohort that had attended GSWA for genetic counselling and from the wider community through a media release. Participants completed a postal questionnaire and were invited to participate further by completing a face to face interview. Transcripts of the interview data were analysed using thematic analysis.
5.3. Results

5.3.1. Response rate and sample characteristics

Questionnaires were sent to 262 individuals who had received genetic counselling at GSWA and 60 individuals who responded to the media release but had not had genetic counselling. Of the counselled individuals, 42 completed questionnaires were received (16% response rate), and of the non-counselled individuals 27 completed questionnaires were received (45% response rate). Overall, the response rate was 21%.

5.3.2. Questionnaires

5.3.2.1. Cohort demographics

Figure 5.1 shows the respondents were either first or second generation Australians (43%) or Europeans of British or Italian descent (40%). The largest group of respondents had no religious affiliation (28%), with the next largest groups being Catholic and Protestant (Figure 5.2).

![Figure 5.1 Reported ethnicity](image-url)
When participants were asked how and when their relationship with their cousin began, the majority said they met as adults (82%) and commonly compared their meeting to “falling in love with a stranger”. The most common type of consanguineous union was between first cousins (81%), 16% were second cousins and the remainder were either half first cousins or double first cousins. The majority of respondents had married this cousin (80%).

The majority of the couples had children from their consanguineous relationship (78%), with 15% reporting a genetic condition in one or more of their children and 63% reporting a pregnancy problem of some kind. The pregnancy problems described included miscarriages, stillbirth, neonatal death or TOP for reasons not stated. The genetic conditions and birth defects described included Zellweger syndrome, Cerebro-oculo-facio-skeletal syndrome (COFS), Meckel-Gruber syndrome, intellectual disability, medullary cystic disease, thanatophoric dysplasia, acquired hearing loss, and two children with undiagnosed conditions.

### 5.3.2.2. Consanguineous relationship

The next set of questions related to the reactions of others to the respondents’ consanguineous relationships. Many participants received little or no support from their families for their relationship (Figure 5.3), but the majority who had told
friends that they were in a consanguineous relationship found them either supportive or neutral (Figure 5.4).

![Figure 5.3 Support from family](image)

![Figure 5.4 Support from friends](image)

When asked if they would be agreeable to their children marrying a cousin, some said they would not encourage it even though they had not experienced difficulties in their own relationship. Others were against the idea and would discourage it.

### 5.3.2.3. Genetics and risk

The estimated risk for the general population in WA of having a baby with a birth defect is 4-5% (Bower et al., 2009) and if the risk for first cousins is double that, the correct estimate would be of the order of 8-10% (Bennett et al., 2002). Using a five point Likert scale, respondents were asked to rate their risk of having a baby with a birth defect compared to non-consanguineous couples (Figure 5.5). Many (66%) who had been to genetic counselling correctly indicated a higher risk; however,
three respondents in the group who had genetic counselling thought their risk was lower or much lower than other couples.

Figure 5.5  Risk perception of birth defects compared to other couples

Respondents were given a risk figure of 4% for any couple having a baby with a birth defect and then asked to nominate the risk for a couple who were first cousins. As shown in Figure 5.6, many respondents who had had genetic counselling accurately estimated the risk (8-10%); however, the majority nominated a lower risk figure, two couples nominated a risk lower than the population risk and four couples nominated a higher than accurate risk (greater than 10%).

Figure 5.6  Numerical risk of birth defects compared to other couples
The level of worry about genetic risk (risk perception) did not always match individual risk perception (Figure 5.7). For example, respondents who believed their risk of having a baby with a birth defect was greater than 20% were less worried than the person who perceived the risk to be less than 2.5%.

Figure 5.7  Risk perception

As shown in Figure 5.8, whether or not a person had received genetic counselling did not generally alter their level of worry, with the exception of the somewhat worried group, where the counselled group had a higher level of worry.

Figure 5.8  Level of worry
Reasons for worry

Respondents were asked an open-ended question about the reasons for their level of worry about their risk of having a baby with birth defects. Several commented that their worry was similar to any other prospective parents. Counselling respondents said that “like any other couple we were worried” and “most mothers during pregnancy feel some degree of concern regarding the health of their unborn child”. Others were worried due to society’s expectations about their relationship. Counselling participants said “I feel a certain amount of expectation from others that cousins will most likely have a genetic mutant” and that “being told many times that we may have children with defects”. Uncounselled participants gave similar responses such as “you have been told this is what happens in this type of relationship and that you shouldn’t get involved with a cousin” and that there was “stigma attached to relationship and having a child with birth defects”.

Even those that had received genetic counselling explained their level or worry based on inaccurate information such as “we were advised we had a 50/50 risk with every pregnancy” and “my mother was a doctor and she said what about the kids they are going to have health problems when they are born”.

Reasons for not worrying

Of equal interest were the reasons why some people did not worry about genetic risk and consanguinity. Some reasons that lessened concerns were related to being reassured after seeing a geneticist (“after genetic counselling and screening I felt the risk was little more than other couples”), a strong personal faith (“when I was pregnant I intuitively felt the baby was ok”) or knowing other related couples whose children have not had problems (“I know many couples who are relatives and had no problems” and “Knowing other cousin couples whose children are fine”).

Beliefs about the cause of birth defects

Participants were also asked about what they thought caused birth defects. A wide range of responses were given including difficulty in defining the causes
(“chemicals, pollution, drugs, alcohol, anything can happen”), genetic factors (“defective chromosomes and genes” and “deformed recessive genes”), and bad luck (“I believe in fate and luck of the draw”). While there was some degree of accuracy in the responses, the group who had been clients of GSWA tended to be more accurate and correctly attribute a combination of genetic and environmental factors. A number of uncounselled respondents indicated they were unsure of the cause of birth defects: “I don’t really know what causes most birth defects. It is obviously a gene isn’t it?” Some indicated it was “random natural processes”.

5.3.2.4. Genetic counselling

In response to the questions about genetic counselling, the respondents expressed some confusion about what genetic counselling actually was and whether they had undergone genetic counselling sessions. For example, some respondents (25%) recruited from outside GSWA referred to advice given by GPs and other medical practitioners as genetic counselling. One respondent had genetic counselling in another country. Another referred to participation in this research project as a form of genetic counselling. Conversely, three respondents from the GSWA group stated they had not had genetic counselling despite them being recruited via attendance at the service.

Of the 42 couples referred to GSWA, 66% (n=30) were referred by their doctor, while 26% (n=12) referred themselves. There were many reasons put forward as the motivation for seeking genetic counselling. The stated reasons were primarily related to genetics (seeking advice and factual information about risk and genetic testing and reassurance that it was alright to have children) or psychosocial issues (grief counselling; encouragement; informed choice or to satisfy the family or church). One couple attended because their family insisted on counselling prior to them sanctioning the relationship and another couple came at the suggestion of their bishop.
The majority of respondents (52%) who commented on their experience with genetic counselling were either neutral about their experience or found it to be helpful (n=36). Few found the experience unhelpful (n=3) and none very unhelpful. The helpful aspects are consistent with the goals of genetic counselling to provide information, empathise and empower in a non-judgmental environment. Stated helpful aspects were obtaining factual data and information, courage and confidence to tell their family about the relationship, gaining reassurance, normalising the relationship and putting the risk into perspective, empathy, kindness and respect for personal and religious beliefs, having someone listen and having a safe forum to discuss concerns. The stated unhelpful aspects were the lack of definite answers and a definitive gene test, not having an explanation for birth defects, the need to travel to the clinic, and leaving with guilt over the relationship.

To understand what the counselled respondents thought about cultural difference, they were asked whether their counsellor was from the same cultural background as themselves and how they felt about that. Over half (52%) thought their counsellor was from the same cultural background, a quarter (24%) thought the counsellor was from a different cultural background, and the remainder (24%) did not know whether or not they were different. When asked if they would prefer a counsellor from the same religious or ethnic background as themselves, half of the respondents were neutral (47%), and roughly equal numbers of respondents were divided between preferring (25%) or not preferring (28%) a counsellor from the same background as themselves. They were not asked about the preferred gender of the counsellor.

The next two questions investigated how genetic counselling affected the respondents’ feelings about their relationship with their partner and their feelings about having children. The majority of respondents (63%) said that genetic counselling had not changed how they felt about their relationship. Some respondents felt genetic counselling led to feeling better (23%) or much better (9%)
about their relationship. Two couples (5%) felt worse about their relationship after genetic counselling, with one respondent saying “it was confronting and made me realise our relationship was probably better off as just cousins”.

When asked about how they felt about having children after genetic counselling, (36%) indicated they felt the same as they did prior to counselling and 36% felt better. This may reflect the reassurance they obtained from having factual information about genetic risk. One respondent wrote “we felt more assured about having children”. Those who felt the genetic counselling had not impacted on their feelings about having children were possibly from the group that felt there was not a significantly increased risk for consanguineous couples and so had not felt much anxiety about having children prior to counselling. Three respondents (7%) felt worse about having children after counselling but did not indicate why.

When the participants were asked whether they knew genetic counselling was available, only 33% of the non-client group were aware that genetic counselling was available for them, compared to 70% of the GSWA clients. When asked whether they would have liked to have had genetic counselling at the beginning of their relationship, the majority (43%) said they would not, possibly because it may not have been relevant to them until pregnancy planning (Figure 5.9).

![Figure 5.9](image)

**Figure 5.9** Whether participants would have liked genetic counselling at the beginning of their relationship
5.3.3. Interviews

The interviews enabled 16 study participants to elaborate on the responses provided on the questionnaires and describe their lived experiences of being in a relationship with their cousin. Confidentiality was preserved with the use of pseudonyms and removal of any identifying information from the transcripts. Demographic information about the interview participants is shown in Table 5.1.

The median interview length was 45 minutes (range 24-64 minutes). The median length of the transcripts is 5871 words (range 3047-8305 words). The inclusion of brief clinical impressions of the participants’ demeanour during the interviews is intended to provide context for better understanding the data.
### Table 5.1 Characteristics of the interview participants

<table>
<thead>
<tr>
<th>Pseudonym</th>
<th>Age</th>
<th>Highest Level of Education</th>
<th>Ethnicity</th>
<th>Religion</th>
<th>Relationship length (yrs)</th>
<th>Genetic counselling</th>
<th>Year counselled</th>
<th>Years between counselling and interview</th>
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<tr>
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<td>Tertiary</td>
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<td>25</td>
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<td>Yes</td>
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<td>6</td>
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<tr>
<td>Ailsa</td>
<td>50s</td>
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<td>Jenny</td>
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<td>5</td>
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<td>Yes</td>
<td>2002</td>
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<td>Italian</td>
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</tr>
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<td>Anglican</td>
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<td>Doreen</td>
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<td>Australian</td>
<td>Anglican</td>
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<tr>
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<td>3.5</td>
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<td>N/A</td>
<td>N/A</td>
</tr>
</tbody>
</table>

#### 5.3.3.1. Clinical impressions of participants’ interview behaviours

The thesis author conducted all 16 interviews and noted that the participants were actively engaged in the process and were open and insightful with their responses. No observable differences in participant demeanour were evident between those interviews conducted at the participant’s home or at GSWA, between those that were interviewed as a couple or alone, or between older versus younger participants.
None of the questions caused participants to hesitate before answering or consistently required clarification or prompting. No participants declined to answer questions or indicated that they were withholding information. Several individuals became emotional and tearful when recalling past distressing experiences or events, but none expressed a wish to stop talking about these events. Many openly disclosed private matters such as family reactions, grief, loss, sexual relationships and children. Several participants stated that the opportunity to talk openly about their experiences was helpful, and for some their first opportunity to do so. Witnessing the depth of emotions, sadness, grief, anger and loss expressed by participants was sometimes difficult but it was also inspiring to see people’s determination and willingness to pursue their true love, sometimes at huge personal cost.

Some participants were very talkative about many and varied subjects whereas others answered the questions with brief responses and needed prompting to expand on their answers. Sometimes, participants referred to previous questions or answers later in the interview, indicating that they had continued to think about previous questions as the interview continued. Several participants expressed an interest in the research topic and noted the media articles that had caught their attention during the recruitment phase of the study. A few participants asked questions about the research data and indicated an interest in the research findings, although none asked specifically to obtain copies of the results of the data analysis.

5.3.3.2. **Themes identified in the interview data**

During the interviews, participants were asked questions about their experience of being in a consanguineous relationship and, if they had experienced genetic counselling, their recollection of that experience. The identified themes were grouped into four main areas, each of which contained sub themes. These are summarised in Table 5.2 and discussed in the sections that follow.
Table 5.2  Identified themes and sub-themes

<table>
<thead>
<tr>
<th>Main themes</th>
<th>Sub-themes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Family</td>
<td>Special bond and shared family</td>
</tr>
<tr>
<td></td>
<td>Family accepting vs. family opposed</td>
</tr>
<tr>
<td></td>
<td>Complex family relationships</td>
</tr>
<tr>
<td>Is it OK to be with your cousin?</td>
<td>Meeting as adults vs. growing up together</td>
</tr>
<tr>
<td></td>
<td>Resisting the relationship</td>
</tr>
<tr>
<td></td>
<td>“It’s just so wrong” vs. “we’re just two normal people”</td>
</tr>
<tr>
<td></td>
<td>Is cousin marriage legal?</td>
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<tr>
<td></td>
<td>Seeking approval</td>
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<tr>
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<td>The incest/inbreeding taboo</td>
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<tr>
<td></td>
<td>Healthy children as proof it is ok</td>
</tr>
<tr>
<td>Challenges</td>
<td>Deciding who, how and when to tell</td>
</tr>
<tr>
<td></td>
<td>Keeping secrets</td>
</tr>
<tr>
<td></td>
<td>Protecting the children</td>
</tr>
<tr>
<td></td>
<td>Feeling isolated</td>
</tr>
<tr>
<td>Genetics and genetic counselling</td>
<td>Genetic counselling helps</td>
</tr>
<tr>
<td></td>
<td>Risk perception and risk avoidance</td>
</tr>
<tr>
<td></td>
<td>Fear of having abnormal children</td>
</tr>
<tr>
<td></td>
<td>Understanding the genetics and risk</td>
</tr>
</tbody>
</table>

**Theme – family**

**Special bond and shared family**

Many participants reported an additional special bond with their partner or spouse over and above their love for each other. They felt that this may have been because they had a shared family, family background or similar upbringing.

I don’t know whether it’s because we’re related, or whether we’re just well suited, but he’s my soul mate, you know? That sounds stupid, but it’s we just understand each other and that doesn’t mean that we get along all the time, or we don’t have issues, but we come from similar I guess similar ideals. So I think that’s probably something to do with us being related. [Jenny, <30, counselled]
I think because we both have the same kind of mums so we were both brought up with the same outlook on life I suppose, because our parents, we come from the same family. [Melanie, <50, counselled]

This special bond was seen as an advantage that assisted in coping with additional stresses placed on the relationship. This was highlighted by Doreen, who felt that being cousins had given them an underlying closeness that had strengthened their relationship.

I guess we were lucky all the way through that he was a good man, a hell of a good man, and we had 28 years together before he died. There is always ups and downs but you know I think maybe the fact that we were cousins even got us through more so because of the fact that there was underlying other things. [Doreen, >50, uncounselled]

Family accepting vs. family opposed

Informing parents and relatives about the relationship was a stressful time for many of the participants. Some of the couples were worried about how their parents would react to the news of their relationship and exhibited a sense of fear and trepidation when they talked about breaking the news.

I should have said something but I didn’t because I was just so frightened of her reaction and you know losing Simon in the process it was just such a really hard situation to be in. [Natalie, <50, counselled]

Oh we kept it a secret for a couple of weeks or something and then ... he told his Mum and she said “Oh you are not serious”. And she just thought that we were really good friends you know. And he said “No I am serious” and Oh she cried but said “But there is not a lot I can do about it” ... I knew that my Mum and Dad would be a lot harder. [Melanie, <50, uncounselled]

Some participants, like Sophie, found that the anticipated fear of telling the family was worse than actually telling them.

I’ll never forget the night, I’ll never forget it. I had been coached and I was in tears now because I thought I was going to be, and it was really serious at the time, I
seriously thought that they were going to shun me from the family, and Doug, and it was going to be huge problems between our families and this was a really big, it doesn’t just affect Doug, you know, it’s huge. ... So she didn’t seem overly surprised and she certainly wasn’t angry at anything and so I always remember being, crying because I was worried and then crying because I was relieved. And then bloody crying again because I had to ah, speak to my bloody father, who, I’ll never forget his line, he said if the queen can do it so can you. [Sophie, <50, counselled]

**Family accepting**

Like Sophie’s parents, other parents were accepting and supportive. Some couples felt they were fortunate to not have any opposition from their family, whereas others had mixed reactions.

My Mum, she didn’t care, she was all for it, she didn’t worry. No that is what we wanted and that is what we got. They had to deal with it. [Bob, >50, uncounselled]

No we didn’t have any not talking to each other or anything. We, I went and lived with him [Bob] at his Mum’s place. The only thing she said was “don’t call me Aunty. You can start calling me Mum” <laughs>. [Linda, >50, uncounselled]

Some parents and relatives, who may have initially had some reservations about the relationship, later became more accepting and supportive. This greater acceptance was often after the passage of time, or the realisation that the relationship was going to continue despite their reservations and opposition.

My mother said that Stuart would always be a good provider and my father said “Well it’s your bed, you lie in it”. So I have for 25 years <laughs>. [Vicki, <50, counselled]

Simon wasn’t actually accepted in the family and I wasn’t accepted into his family until about a good two years after everyone had found out that it had happened. But now it is just very, very different. [Natalie, <50, counselled]

I think it would have been more of a problem on John’s side that he might have had to deal with it quite sensitively. I went home and my parents sort of knew really before I knew what was cooking. So in a sense that was to my benefit. I think he would have had to develop some strategies in order to tell his Mum and Dad and
for them to get used to it. ... but that was quite a problem for him and something that he had to deal with. [Mina, <50, uncounselled]

Another participant, Misha, had an initial negative reaction from her parents but they were more accepting after a temporary break in the relationship. Conversely, Misha’s partner’s parents were accepting all along, which she thought may have been because they had lived in a country where consanguineous marriage was an accepted practice.

When we got back together again I think my parents just thought well that’s her choice and so we did get married. Not that I was particularly into marriage but for Karl to be able to stay in Australia we had to be married to be able to get him a Visa. And so we got married quite quickly after I’d left the other person I was living with, probably about 6 months or so. And so it was fine and they were really happy and they celebrated with us. Karl’s parents were completely overjoyed because for them it’s not a big thing, it’s quite common. [Misha, <50, counselled]

Family opposed

When some families were informed about the consanguineous relationship, such as Vicki and Anne’s, the response was displeasure or disapproval of the relationship and choice of marriage partner.

The only real opposition I’ve had to face has been my parents, who did back away when I made my choice. They did support me through my marriage and the actual wedding. They kept all their reservations to themselves after they expressed it the once. And it hung like this huge, black cloud over our, my relationship with my parents for a long time. [Vicki, <50, counselled]

Grant’s Dad wasn’t very happy with it. I got introduced as Grant’s friend for about two years after we were married <laughs>. [Anne, <50, counselled]

One of my uncles was dead against it anyway because he just felt that it was wrong that it was incestuous and he was quite outspoken about it, not in a nasty way he was just one of those people that called a spade a spade and he just thought this was wrong. ... once he saw that well actually we were sticking with this and once we got engaged and got married he accepted it then. He just thought that it was
wrong and we shouldn’t be being allowed to do it. I think he thought my Mum and Dad and Duncan’s Mum and Dad should have not allowed it and they should have said “No you are not doing it” End of story. [Ailsa, >50 counselled]

When everyone found out we were together we did get some backlash from my Mum’s husband who is not related to me because she remarried when I was about 19. He breeds sheep and he just thinks that it is totally wrong that we are together. And he won’t let us stay at my Mum’s house together or anything like that. He wouldn’t come to the wedding. Most of our family were all happy to our faces but then my Mum told me that behind our backs people were saying it was a bit weird … We didn’t know whether to have a wedding or not cause right up until like two weeks before the wedding we heard that people were sort of, not against it, but they just didn’t think it was right. So we thought well why should we have a big wedding when most of the people don’t agree with it. [Nina, <50, counselled]

Other families had more immediate and unfavourable reactions in response to being told about the relationship. In some cases these reactions were strongly negative and occasionally violent.

We were seen out and about by other family members and they rang my parents up straight away “Did you know Simon and Natalie are doing this and this”. And when I got home all hell broke loose. [Natalie, <50, counselled]

After about three years dating we decided it was time to get married. I asked my Mum and she said “No”. So we ran away and my Dad then, on that day, my Dad knew that we were dating and he almost killed my husband. He was so angry. Really, really angry. First because he was my cousin, second because he didn’t want me to have a boyfriend. I was 17 years old and he was 22 … But everybody was so angry. My family with his family they stopped talking. [Bonnie, >50, uncounselled]

His Mum … she was kind of, didn’t want to know, if you know what I mean? And she went off her nut. There was a huge family argument. [Jenny, <30, counselled]

His Dad actually … he wanted proof that I was actually pregnant. So I thought we were going for a scan and he had actually taken me after hours without saying who to someone at <name> hospital for someone to do a secret abortion. I actually had to physically run out of the room before he could pin me down. So it was quite
frantic. Yeah it was horrible ... After that had happened I didn’t really trust anyone in the medical field <laughs>. [Lee, <50, uncounselled]

The participants displayed a strong need for support from the family and the impact of any family opposition to the relationship was significant, often resulting in consideration of ending the relationship. Sometimes couples delayed the relationship, often for considerable periods of time, in response to family opposition.

I think it was in the back of my mind, it definitely was, if I could chose that Grant wasn’t my cousin but be the same person that would have been much easier ... None of my family were around when we first met and his Mother had died. I think had his Mother been alive I think it might have been a bit different, we probably wouldn’t have ended up together. [Anne, <50, counselled]

I do think that if there had of been too many objections I probably wouldn’t have pursued it, I don’t know about him, but I think I am a bit of a chicken and I probably wouldn’t have pursued it if there had of have been a lot of objections from Mum or Dad or my sisters or my brother. [Noeleen, >50, uncounselled]

I just met one of them a couple of weeks ago and to them much the same thing happened as with us, ... She met him on holiday and so on and then because of the family opposition they just forgot about it. They just let it go for a few years. She married somebody else. It didn’t work out and they met again 20 years after and the spark was still there so they decided to get married. [Eric, >50 uncounselled]

Natalie and Simon bowed to family pressure and tried unsuccessfully to end their relationship. When they realised they could not be apart, Natalie moved out of home and was not in contact with her parents for many years.

We thought that we could forget about each other. I tried to date other guys but it just didn’t happen. Then we got back together again and then we got caught out again ... At that stage we had been intimate with each other and my parents were devastated that that had actually happened. Then I actually moved out of home. I picked up my bags and whatever I had, left my car and anything valuable that I had that I knew I couldn’t afford to pay for and I was out. [Natalie, <50, counselled]
In some cases the family pressured participants to end the relationship or rejected them outright. This resulted in significant emotional trauma and ongoing family conflict for the participants.

They did everything to try to break us up. Even when we were here. So it’s been hard, very hard. [Bonnie, >50, uncounselled]

When my Mother came over she was horrified, because she came over to [country] later and she was very negative and so she said “that’s it, you’ll have to come back home”. And so I came back to Australia and they worked very hard on splitting us up. And then when I finally did say “Okay, that’s it” because I had been away for eight months, my Mother made me go back to [country], a complete reverse face ... It was sort of like watching table tennis, which hand my ring was on – my left, my right, my left, my right – to try and work out what was going on. [Vicki, <50, counselled]

When we told his parents his Dad absolutely hit the roof and actually took his car off him. He was living out of home so he and packed up his stuff and took him home and just severed the relationship between us completely. He used to steal his Dad’s car and come down and visit me ... There was a lot of pressure and the entire family was split down the middle over the whole thing. It was a very painful and difficult time. [Lee, <50, uncounselled]

Participants had to cope with not only the reactions of parents and relatives but also their children. Bonnie had not revealed their consanguineous relationship to all of their children until she participated in this study. When she explained to her daughter why she was participating in this research, her daughter reacted negatively. Mona was also concerned about how her children would respond.

I have a very close relationship with my eldest daughter, so she knows. My other daughter she’s going to be 14 in January and she’s getting at that stage where she wants to know a lot of things and get very close to me. I had this appointment on the calendar and she says “What is this?” I said “I have an appointment at [hospital]” and she says “Why?” And I said “Because we’re participating in research for this, this and that”. And she says “What? That’s disgusting!” And I felt, oh, I
shouldn’t say anything then. My daughter’s reaction was bad yes. My daughter’s reaction was very upsetting. [Bonnie, >50, uncounselling]

Now that my children are older we think about it when they say “Ugh, ugh” and why is that or they compare themselves to cousins. And say “Oh I wouldn’t do that”. And I think we were a little bit blind to that. [Mona, >50, uncounselling]

**Complex family relationships**

For some consanguineous couples, being cousins created complex relationships within the family. Some participants struggled to negotiate these complex relationships. The challenges included describing the relationships, naming the relationships and managing family conflict.

It is very complicated because I am my own son’s second cousin and my father is his grandfather and his great uncle. So go figure. It really gets confusing. [Lee, <50, uncounselling]

I never called her Mum. Mine was Mum and she was Mother. I have got no idea what I ever called the old boy until he became a grandfather. And then I called him Pop. He was always Pop, Mother and Pop. Isn’t it funny that I can’t remember what I ever called him? ... I said if ever there was any silly niggles, you know your normal fights – you’re not going home I’m going home to Aunty Anne, you know all of a sudden it was Aunty Anne<laughs>. [Doreen, >50, uncounselling]

I didn’t have to get to know my in-laws. And my Mum and Dad immediately knew all about Doug and knew that he was the perfect person, and said “We’re so glad it’s Doug.” You know, plus I had, I had relationships with my sister and brother-in-law already, ready-made. That was also a down side. My mother-in-law has known me since I was a baby and treats me, I think accordingly, not with the same amount of respect as perhaps someone who had come into the family five years ago. She treated me almost like another daughter, which could be good but I don’t see it like that. I’d rather she didn’t ... so that’s a positive that is also negative sometimes. That they know everything ... what I was doing before Doug and I got into a relationship ... they know all my other bloody relationships and all the stupid things I’ve done, that mother-in-laws wouldn’t normally know, she knows. [Sophie, <50, counselling]
One of my sisters – who I don’t have a good relationship with anyway – she found it particularly difficult that Duncan and I had a relationship. She is younger than me, because she liked her big cousin from Australia too <laughs> and she was only 13. She actually was very jealous of the extra dimension of the relationship with me and Julie, because Julie was her Aunty and Julie was a really lovely lady and great fun and one of these people that you want to have a relationship with. So that has been, I think we didn’t have a good relationship anyway and there has been lots of jealousy there anyway. But I think it has been made worse because Duncan, she felt she had dibs on him as well. [Ailsa, >50, counselled]

While adjustment to the complex relationships created by the consanguineous union was required by some, many felt these complex relationships were positive. The benefits included already knowing the relatives, closer relationships within the family and having a shared history and upbringing that resulted in similar beliefs and values.

You fit into that extended family sort of. That circle that is sort of like the whole package. [Noeleen, >50, uncounselled]

I guess that’s good because you can sort of understand where those issues have come from. And I understand his Dad, you know, more than I would understand a Father-in-law … and I guess it’s negative because our Fathers aren’t speaking since our Nan died. So you know you do hear things from both sides about your Father … I have feelings about my Uncle that I don’t have about my Father-in-law, if that makes sense. I separate the two. [Jenny, <50, counselled]

We know each other like we know the family. Like we have similar histories and we share a maternal grandmother. [Anne, <50, counselled]

It’s nice because there is this shared history. Duncan and I our Grandfather and Grandmother are the same and we have got memories of them, and we are going to go back to <country> next year and take the kids back, and that is nice because that is the same there is a lot of the same places that we are taking them that is both of our family. [Ailsa, >50 counselled]
The use of humour and light-hearted banter, particularly around the topic of dealing with family and family relationships was common.

At weddings and such well actually it is very easy for us. We have only got half the amount of relations <laughs>, and half the cousins are all always on one table. [Margaret, >50, counselled]

I didn’t have to change my surname or any of my paperwork. That was probably the best thing. You go through as being a Miss and then you change to a Mrs and no-one questions you because they just think that you have kept your maiden name. [Natalie, <50, counselled]

It doesn’t cost so much does it when we have a party <laughs>. His cousins are my cousins so that automatically cuts out half <laughs>. [Linda, >50, uncounselled]

Christmas is dead easy <laughs>. When we lived in <country> we’d all just get together you know. So that is easy, we didn’t have to here and there. [Ailsa, >50, counselled]

I said to Daniel “When your kids have kids I’m going back to cousin. I am not going to be a grandmother” <laughs> I put my foot down there <laughs>. I can make my decision about which one I want to be. [Melanie, <50, uncounselled]

Theme – is it OK to be with your cousin?
Meeting as adults vs. growing up together

Participants differentiated between cousins that they had grown up with and cousins that they met as adults. The ‘acceptability’ of the relationship was perceived differently according to when the couples met and when the relationship started. Couples whose relationship started when they met as adults likened it to meeting an unrelated stranger and uncontrollably falling in love.

I think that it is better that me and Grant didn’t know each other ... I don’t think that there is anything wrong with it but I think it would be different if you had grown up with somebody, lived across the street from them and saw them every holiday and had a relationship with them for your full life. I think that is a little bit
different. But obviously then again there is nothing wrong with that. [Anne, <50, counselled]

Two strangers who just met. You know, I suppose the crux of it is, he was brought up in <country> and I was brought up here. I was only three when I came out here. He was a complete stranger when I met him, just two strangers meeting. I couldn’t look twice at any of my cousins here, but he was a complete stranger, I wasn’t brought up with him. I think it would have [been different] because he would have been my cousin and that cousin taboo would have been in there. But because he was a complete stranger it was totally different. [Vicki, <50, counselled]

I didn’t think of him as my cousin I mean I would never have looked at any of my other cousins. I had male cousins, but they were just my cousins who I had grown up with since I was little and I thought of them the way I think of my brother … but Duncan’s Mum wasn’t like my Aunty and he wasn’t like my cousin. [Ailsa, >50, counselled]

Overall, responses indicated that there was a sense that if you grew up together your relationship was closer and it was therefore closer to incest.

I don’t know that we would have ended up together … because I remember, you know, as a five year old, the last time I saw him and he was, well he’d have been 10 or 11 or something, and you know, he was teasing me the whole time … <laughs>. If it’s more sort of a brother-sister relationship I’m sure you wouldn’t look at someone the same way when you’re 20. [Jenny, <50, counselled]

I think it comes from not knowing each other. Whereas if you grow up together it is more like a brother and sister type thing [Nina, <50, counselled]

I mean in <country> where I grew up I had girls my age that were cousins and so on and we probably went to school together, played together whatever, even went dancing together and so on. And it was nothing more than that, not even the slightest thought of that. [Eric, >50, uncounseled]

Our children and my sister’s children they are all cousins and they all you know before they married other people, they go everywhere here and there together but there just didn’t seem to be any inclination that they were attracted to each other
... So I think probably if we hadn’t have met at that age, had we been younger probably it would have been different. [Noeleen, >50, uncounselled]

Resisting the relationship

Some couples felt comfortable with their feelings and trusted how they felt, but many were unsure about whether it was ‘right’ to allow the feelings for each other to develop. The participants felt a sense of the relationship being wrong, shameful, sinful or strange. Many tried to suppress their feelings and some initially resisted the relationship despite feeling attracted to their cousin.

I think he was worried probably ... It probably took him a long time to say anything to me. But we probably both knew anyway, for ages, without saying anything. [Misha, <50, counselled]

It didn’t feel like we were just cousins. There was something else we were very nervous about starting anything we just became really close friends. We had dated for about 12 months and I didn’t let Simon do anything intimate to me at all because I just didn’t know whether it was the right thing to do. [Natalie, <50, counselled]

My friends kept saying “When are you and Daniel going to get together?” and I used to say “Don’t be stupid, we can’t” I really honestly don’t know what happened but it did. [Melanie, <50, uncounselled]

“It’s just so wrong” vs. “We’re just two normal people”

Participants felt uncomfortable about having feelings for their cousin and were left with a sense that their relationship was wrong or something that they should avoid or not disclose to others. Some were unable to continue the relationship because of these negative feelings.

My background, my upbringing, I mean I am quite religious. I am Catholic so it is there not in the Bible as such but you are just brought up to think that you just don’t. You know it is just not the right thing. God dating your cousin, going to bed with your cousin that is just wrong you know. [Natalie, <50, counselled]
This particular weekend we’d gone to <country> for the weekend. That’s where we, well you know, really got together and really realised that this was what was going to happen. And it was really scary and really secretive ... I mean now clearly, we should never have worried. But who’s going to know that at the time? It felt like we were doing something really, really bad and really, really secret. And so nobody, nobody knew. [Sophie, <50, counselled]

He wore the shame that we had been together. He was given that abuse by his family “How could you be with your cousin” and all that sort of stuff ... He is still deeply ashamed that he was ever with me. He can’t look at me. You know I mean it hurts naturally. [Lee, <50, uncounselled]

Participants described the reactions of some people that they had disclosed their relationship to and how they had reacted as if they were doing something wrong by being with their cousin.

She said “I still can’t get past the disgust of you being with your cousin”. [Lee, <50, uncounselled]

I’ve only had that really bad reaction once and that was someone that I worked with ... She wasn’t you know revolted or anything but she just sort of said “You’re kidding? You can’t do that” [Jenny, <50, counselled]

There are a few people out there who kind of look at you as if, you know that is disgusting, or you know how could you do that sort of thing. When I went back to <country> the first time after Simon and I were officially out in society I was kind of looked down on quite a lot when I went over there ... I did get kind of looks as though people were looking down on me as if they were kind of fobbing me off because as far as they were concerned how could I have done something so disgusting. [Natalie, <50, counselled]

In contrast, other participants described themselves as being “normal” people and had a desire to be thought of as “normal” by others. They did not feel that there was anything wrong with their relationship and felt that there should be no need for them to justify their relationship to anyone.
We’re perfectly entitled to do this. We’re not abnormal. We’re not unusual. [Vicki, <50, counselled]

We’re people you see. We’re intelligent people ... There’s nothing strange about us. We’re quite normal! And we had normal relationships before this. [Sophie, <50, counselled]

Not only in the Bible in history as well...Just the examples there. Why couldn’t they complain to them? We’re just two normal people. [Bianca, >50, uncounselled]

Doreen thought that people’s responses to consanguineous relationships may depend on consanguineous couples’ own perception of their relationship.

I was never ashamed of the fact that we were cousins, you know I couldn’t see any reason not to hold my head high, whereas I do think a lot of people do, you know they sort of worry about what other people might think and what they might say and I think it is their own attitude when they try to tell people that does it. [Doreen, >50 uncounselled]

Is cousin marriage legal?

When participants talked about the legality of consanguineous marriage in Australia, varying levels of understanding were revealed. Some participants were aware that it is legal, some were less clear about the legality and others were oblivious to the laws in this area.

When we were getting married, and I don’t see us as having any different legal standing being two people, as any two people getting married. It never ever occurred to me that there was any legal standing that would get in the way actually. I thought first cousins were allowed to get married. So it never ever, never ever struck me as an issue with second cousins. [Vicki, <50, counselled]

I didn’t know exactly if it was legal in Australia. I sort of knew overseas because of royalty marrying blood lines and stuff like that but I didn’t know what it was like here in Australia. I had been on to that web site cousin couples as well and had a look at that but it is mainly American people. [Natalie, <50, counselled]
I think it is legal in both the eyes of the church and the law. [Doreen, >50, uncounseled]

Legal? I don’t know, are they? [Bonnie, >50, uncounseled]

Uncertainty about the legality of consanguineous marriage was a reason for family to object to the relationship in some cases.

I think it was my mother’s Victorian upbringing, I’ll be honest. My grandfather was extremely strict and a disciplinarian. Mum was brought up through the Salvation Army and, no, she truly, truly believed it was against the law for us to get married. ... my Mother thought it was illegal for cousins to marry, let alone second cousins, which is what we are. She totally believed that and because she believed that we became engaged with mum’s objection and with this idea in her head that you can’t do that, you’re not allowed to get married, it’s against the law. And I said no it’s not against the law, we’ve checked it out. [Vicki, <50, counseled]

Some participants sought clarification about the legal status of consanguineous unions prior to their marriage, because of a lack of knowledge about the law.

I don’t actually know about it in Australia. When we got married though in [US state] there was a friend of Grant’s whose mother was an attorney and he was a great guy, he knew from the outset that we were doing something and carrying on. We actually asked his Mum to look into it for us and she said “Yes it is ok, there is no law that says you can’t marry your cousin.” [Anne, <50, counseled]

I went into it a little bit. I found out a little bit about it from the births and marriages whatever, whether or not it was legal and I went into the church situation. [Margaret, >50, counseled]

Seeking approval

Many couples sought approval for their relationship not only from their family but others as well. They described obstacles that non-consanguineous couples would not normally have to face. Some couples had to get permission from their church to marry and others sought advice from doctors.
We went to the family doctor and asked what the complications would be or whatever and we also went to our parish priest to see how the church felt about it. We had to have a dispensation from the bishop. [Noeleen, >50, uncounseled]

We went and spoke to his doctor and to my doctor ... They both assured us that as far as they were concerned, and the minister also assured us, that there was no impediment to the marriage ... we spoke to the minister and sort of went through the front of the Bible and there was no reason there. It doesn’t say that you couldn’t marry your mother’s sisters offspring. [Doreen, >50, uncounseled]

We went to our GP and we discussed the fact that we wanted to get married. We asked him if there would be any negative impact on us having children because my mother’s main objection was that I would have deformed children ... We were told that we’d have a 60% risk factor of any inheritable disease. [Vicki, <50, counseled]

Once over these obstacles, disapproval of consanguineous relationships was expressed in many ways by family, friends and society and this lead to participants experiencing feelings of rejection and isolation.

People are going to sort of look sideways at you. I worked with a girl who I grew up with. I said I was going away and she said who with and I said my cousin and she said “Oh I hope that it doesn’t become incestuous” I remember her saying that very well. [Margaret, >50, counseled]

It was upsetting because this was my Aunt and Uncle and they were the only Aunt and Uncle I’d ever known and I’d really been fond of them as a kid even though ... I didn’t see them often. I would have loved to had a close relationship with them, so, yeah that was upsetting. [Jenny, <50, counseled]

I actually moved out of home ... I didn’t speak to my parents for about six months. [Natalie, <50, counseled]

When you are closely related to someone the opinion of them counts, your husband’s opinion counts or whatever the situation your child’s opinion counts you know. So I suppose if I was pregnant to someone who wasn’t my cousin and I didn’t really know his parents that well I wouldn’t give a flying fig what they thought of me you know it hurt, deeply emotionally hurt so and it took a long time to recover
from <cries> excuse me, and still hurts sometimes <cries> ... being isolated and alone. It really was difficult. [Lee, <50, uncounselled]

Incest/inbreeding taboo

Participants that felt a taboo about consanguineous relationships existed in Australia attributed this to the negativity around consanguineous relationships. The source of the taboo appeared to be related to the question of whether or not consanguineous relationships were incestuous.

They think it is like being with your sister and it’s so not like being with your sister. Or your brother. It’s like being with your Dad. You get that sort of reaction from people and it is not, it is not like that at all. You know the whole concept of Simon and I being cousins I don’t even relate to him as my cousin any more. We are husband and wife and that’s it. At the back of my mind it will always be there officially we are cousins. You know we’ve married and we have started our own family and we are getting on with it. So yeah but I do feel a lot of that comes from the father and daughter thing or the brother and sister thing. [Nina, <50, counselled]

It is quite interesting the reaction that you get, cause I have never been ashamed of it personally. I mean you shouldn’t be with a close relative but to me that means within your own family. So I don’t really see it as anywhere near incestuous or any of those horrible connotations that a lot of people do put on it. [Lee, <50, uncounselled]

I suppose it has a sort of a slight stigma. I am not quite sure why maybe from the point that I was discussing a while ago incest it is almost, it isn’t is it? Or is it? I don’t know. [Noeleen, >50, uncounselled]

It is not really that spoken of but you know when I worked [nursing] we would get quite a few kids. You know you would see a couple every year that you had something then it would come out that their parents were cousins. And just to see the remarks that people said about it. [Ailsa, >50, counselled]

Remarks such as those Ailsa heard in her workplace, and other insensitive comments and jokes, were difficult for participants to deal with. Some of these
comments were made directly to the participants and others heard them in general conversation. Vicki, who had been married to her cousin for 25 years, continued to be hurt by jokes that are bandied around when cousin marriage is mentioned.

She was going on and on about how cousins being married. I don’t know how it came up. Somehow it came up and she just said how it was incestuous and you know all sorts of bad things. It was really bad. The opportunity was there to have stopped her when she very first started and say “Well look” but we didn’t, we let her go on <laughs> and she did and then when she finished Duncan was a real sod he said “Oh that was interesting because we are cousins.” [Ailsa, >50, counselled]

You hear the jokes all the time, about, you know, the sort of Tasmanian type jokes. Yeah, well, they’re, you know, clearly not funny especially not now, for Doug and I. And it’s based on ignorance and funny for those people who don’t know, <pause> don’t know, so who are ignorant. [Sophie, <50, uncounseled]

Oh the jokes, yeah I still cringe. Cringe to this day when I hear jokes about cousins. I get quite offended, you know? I suppose it’s like Irish people getting offended about Irish jokes Oh the kissing cousins, the Elvis Presley film. I cringe when I think about that. [Vicki, <50, counselled]

Healthy children as proof it is ok

In response to the negative reactions and taboos associated with consanguineous relationships, participants spoke about the fear of having abnormal children and the relief they felt when they had normal children. Children with abnormalities were seen as evidence that their relationship was wrong, whereas, normal children were evidence that it was alright to be in a relationship with a relative. One participant, Natalie, wanted to educate others and saw participating in the research study as an opportunity to do this.

And that’s why I like to be involved in research or in letting people know that it ok and we are fine and we are normal. We are not abnormal and we haven’t had an abnormal child and that is proof. Kyle is proof that there is absolutely nothing wrong with it. [Natalie, <50 counselled]
I think my biggest fear about people’s attitudes is the children. Once I’ve had normal, healthy bouncing children, I don’t think it’s going to be a problem anymore ... Because they’ll be living proof that we’ve not done anything deliberately wrong to our offspring through being selfish and wanting a family. So I think that it’ll be much easier once I’ve given birth to at least one healthy child. [Sophie, <50, counselled]

I make people think about it and talk about it. Because when they say things I say look at my children. And I’ve got three boys, the eldest one did extremely well at school he was a state scholarship winner ... the next one is an outstanding sculptor and the youngest one is an engineer. [Margaret, >50, counselled]

In addition to the fear about having abnormal children, participants expressed fear of the reaction from others if this did occur. They felt that they would be blamed and any problem would be attributed to the consanguineous relationship even if it were not the cause.

If you get married you’ll have, Oh what did she call it? <pause> your children will be deformed. She had some word for it, I can’t remember it. And it always stuck in my mind. So when I had Kate and she had something wrong with her eyes I just fell to pieces. I absolutely fell to pieces. [Vicki, >50, counselled]

**Theme – challenges**

*Deciding who, how and when to tell*

Outside the family unit, participants faced difficulties when deciding how to respond to questions about their relationship and whether to disclose to friends and social contacts that they were cousins. The hesitation was almost always related to fear of how people might react and whether or not they would be judged in a negative way. Some participants had not experienced any difficulty being open to others about their relationship and others found a middle ground where it was possible to tell some people (those that they were closer to) but not others.

Grant interestingly enough has no qualms telling people. Not that he tells everybody but his good friends that he’s made after we got married. He’ll tell them whereas I am afraid of the way that people will react. [Anne, <50, counselled]
I haven’t kept it quiet from everybody. But I did find myself, as soon as I have said it, watching their face to see their reaction and immediately launching into an explanation and a justification. I found that that was important. My problem, not theirs. [Vicki, <50, counselled]

I think John is by nature less inclined to do that because he doesn’t see the need to. But if I know someone well and they talk a little bit about how did you two get to know each other and if I don’t have special need to then I just mention that we were related. Most people will say “Oh, oh really”. If I don’t need to, I don’t even always think about it maybe. But yeah some people think it makes a good story really <laughs>. Thankfully no one in my environment has actually made me feel bad about it and if they have thought that they have not indicated it to me. [Mona, <50, uncounselled]

Once participants had disclosed details of their relationship, the reactions they received were varied, ranging from negative responses that caused embarrassment or disrupted friendships to supportive reactions that normalised the situation.

I sort of wanted to tell our friends and I felt that it was only fair to them. And I think I told a couple of my girlfriends on the phone and one of them sort or reacted you know, a little bit negatively about it. But I think I didn’t talk to her, we didn’t talk to each other for quite a while but, we’re actually friends again now. I really thought that it would be worse than it is ... maybe I just have chosen my friends wisely, or the people that I’ve told wisely. But the reaction has been really good, really positive. And so many times I get told “Oh yeah, my Grandma did that, or my cousin married her cousin, or, my Mum and Dad were that” or you know? In fact, I would say 90% of the time, when you tell someone they will say they knew someone or they’ve heard of someone, or a relative did that. [Jenny, <50, counselled]

One person that I work with once he, I think he was a Sikh and it’s very bad in his religion, considered really bad. So when I told him his reaction was like “Oh no, my goodness, I can’t believe you’re married to your cousin”, but we just laughed and that was fine and I just said “Well it’s no problem for me” and you know that’s the only time anybody’s ever been negative and that was kind of in a friendly way so that didn’t really bother me. [Misha, <50, counselled]
It is funny the eyebrows always skip to the roof you know “Really”. I mean there is very few people that just take it in their stride. A lot of people they are “Really” and their eyebrows go and jaws drop. [Lee, <50, uncounseled]

Some participants did not feel comfortable enough to talk about their relationship with anyone. A very common question asked of any couple is How did you two meet? Some of the participants never felt able to answer that question truthfully owing to fear about how people would respond to them. Others, like Melanie, do not tell people because feel that they are no different to anyone else and there is no need for people to know.

I actually haven’t told anyone. My best friend from high school knew because obviously she was my best friend when I went travelling so she knew all about it. But that would be about it, all of my friends from university, through work none of them know. [Anne, <50, counselled]

They have to be very close friends and the subject has to come up. I never volunteer it. I find people have a watered down reaction [compared] to my mother’s. They stand back and say “But that’s not allowed”. And lots of jokes. Always jokes about cousins ... and inbreeding. I fear, my fear, my genuine fear is that they would reject us because they thought we were doing something we shouldn’t be doing. And it was me projecting that fear on them, not them reacting that way. [Vicki, <50, counselled]

If you don’t know people very well you are not sure what they would think and you are not sure how they would react to something. And if that would make them judge you without actually knowing you. You know they judge you in a negative way on the basis of the fact that you married your cousin rather than got to know you as a person. [Ailsa, >50, counselled]

They don’t really need to know. It doesn’t have anything to do with them. Daniel and I are just Daniel and I. There is nothing different about us apart from genes I suppose and that’s it. [Melanie, <50, uncounseled]
Keeping secrets

Participants expressed anxiety about revealing the nature of their relationship and expressed the need to keep this information a secret from family, friends and children. Many kept their romantic relationship a secret from their parents for some time because they feared the reaction. Natalie and Simon were together for four years before their relatives revealed to others they were a consanguineous couple. Bonnie also kept the relationship a secret for a long time. Some participants such as Sophie became accustomed to keeping a secret and had developed a well-rehearsed cover story. Sophie had “slipped” up once and let the truth be known but remains more comfortable with her “story”.

Then it got to a stage where we were actually seeing each other. We said “Ok lets take it a step further” and we actually started dating as such but we were actually doing it behind our parents backs because obviously we didn’t want them to find out. [Natalie, <50, counselled]

Before we got married, we dated about three and a half years, with one and a half was behind everybody’s back. [Bonnie, >50, uncounselled]

We still don’t tell our friends. Nobody knows, our friends still don’t know. And when we’re at family gatherings and family friends ask how we met we say, the standard response is that our parents were friends before we were born ... When you’ve been telling a lie that long it’s much easier. It’s habit. You know exactly what you can and can’t say without having to think about what you can and can’t say. [Sophie, <50, counselled]

I slipped, about a year ago, with one of our favourite friends, and I called Doug’s mother my aunty instead of my mother-in-law. If I had just gone “Oh what I mean is mother-in-law”, it would have been, it would have gone. But I didn’t and I went “Oh” and then I told him. And then I made him swear blindly he would never, never tell. And I wish I hadn’t told him and I wish he didn’t know, because I’m afraid of him telling someone else now, even though he probably wouldn’t. So, knowing now, after telling the truth once, how it felt afterwards, I’m glad I haven’t. [Sophie, <50, counselled]
The keeping of secrets had an ongoing impact on individuals and their relationships. Ailsa expressed a desire to be able to be more open and honest and felt that not doing so affected the relationship she had with others. Anne’s desire to keep her relationship secret was reflected in her concern about her desire to be an author in the future. Melanie secretly visited her mother because her relationship with her father ceased after she began the relationship with her cousin.

I do feel when people say to me “How did you and Duncan meet?” or “Where did you meet?” I feel like I have got to kind of take a bit of a breath and launch into my own, you know, prepared tale that I tell people. I don’t really like doing that. It would be nicer if you could just say “Oh you know whatever”. I think when you have close friends, it is kind of, it is not that you feel like you are being deceptive but it definitely is. It is not a weight that you carry, it is not something that ties you down but it would be nice to just be out in the open with it in a way ... If you can’t tell people the truth then that kind of taints the relationship from the very outset. [Ailsa, >50, counselled]

Well it is very strange because I always think – you know this is just me thinking in the middle of the night when I can’t sleep – but I would love to write a book. And then I think but what if I was to write a book that was really good and then they did some background research and they found out I was married to my cousin? I said to Grant I would have to write it under a different name – the things you think about <laughs>. [Anne, <50, counselled]

My Mum is cool. I don’t get to spend as much time with her because of Dad but he goes to work. When I first met Daniel, yeah he used to go to work early and Mum used to go to work about eight o’clock and I used to go there for breakfast. And then I would see Mum. But she goes early too so I maybe only get to see her maybe once a week or, I talk to her all the time. It makes it difficult when my Nanna and Pop come to visit because I can’t go there for dinner, or I can’t do this or I can’t do that you know. But I go there for breakfast every day and that gives me an hour or so with them and whatever. [Melanie, <50, uncounseled]

Protecting the children

Participants expressed concern for their children regarding how and when to explain their consanguineous relationship to them. They were concerned about
how the children would react, the impact of not telling them, when to tell them and the repercussions if the children then disclosed the relationship to others. Having experienced negative reactions themselves, they wished to protect their children from hurtful comments and jokes.

It’s not because I don’t want them to know. It’s not that I am embarrassed or ashamed of what we have done. But I still think though that you don’t want to be telling them at the age where they may go and tell their friends ... We have both said that we will tell them but I just don’t know whether we should start to get them used to the idea maybe early on. I don’t know ... it is just tricky to figure out when and how. We didn’t want to make a big deal about it for the kids but at the same time I suppose part of me was thinking well it is better they don’t know because kids tell people things and then I don’t want them to get any negative reactions back. [Anne, <50, counselled]

She knows the whole story. Everything. And I encourage her not to tell anyone because, not even, it’s irrelevant, they don’t need to know. I don’t want to put her in a position where she has to explain my actions. She shouldn’t have to defend what i’ve done. So, she has a right to know. And I said even when she gets a partner of her own, a husband, just don’t tell him. What he doesn’t know he will be more comfortable with and she won’t have to explain herself. And really, it’s nothing to do with anybody else. We’re a normal functioning family. When she was younger it didn’t matter because she didn’t understand the implications of her parents being cousins, but as she got to about 13, I explained it to her because I didn’t want her to go telling everybody and then being surprised or caught out as a result. So I tried to tell her it wasn’t a bad thing, but it wasn’t something that I wanted her to run around telling everybody because she would spend most of the time explaining why. [Vicki, <50, counselled]

I am just going to come out and tell him so that he doesn’t get caught out you know. If other children see something or hear something he will be able to say “So what? There is nothing wrong with me” you know justify himself. ... I am just going to be honest about it. I am not gonna hide it. I am just gonna when I think he is at an age where I think he is going to be able to understand ... I just don’t think lying to him or being dishonest to him or trying to keep it away from him until he is at an age where he really does understand like 17 or 18, he will go “Mum what the hell?”
and me having to explain it and him feeling betrayed all these years and not telling him. [Natalie, <50, counselled]

One of the interview questions asked the participants how they would feel if their children wanted to enter into a relationship with their cousin. In addition to the concern about the reactions of other people to consanguineous relationships, which reflected participants’ own experiences, there was concern about inbreeding as this would mean multiple generations of cousin marriage.

I would be afraid, because we are cousins and they would be cousins already and there would be more than one generation. That will be my concern but otherwise, nothing. [Bonnie, >50, uncounselled]

I just do hope that I give them enough social contact that they don’t have to fall back on getting acquainted closely with their cousin purely because they haven’t met anyone else. [Mona, >50, counselled]

My sister had three daughters the same age as our boys and they were brought up together. The first ones are seven months apart, the second are three months apart, the others are three weeks apart. He [Colin] used to say “I really hope they don’t get together because of our relationship”. You know we can’t say “Well you are cousins you can’t marry” but that makes them more related. ... You don’t want to give your children problems do you? [Margaret, >50, counselled]

Feeling isolated

Many of the participants reported being surprised when they first heard about the study because they had been unaware of any other couples in consanguineous relationships. Participants had often lived with a secret about their relationship and not discussed the issue with anyone, further compounding the feelings of isolation. There was a great sense of relief that they weren’t the only ones who married their cousin and many mentioned that they would like to meet others in the same situation. Lack of awareness about other consanguineous couples meant that even the participants held the common misconception that all cousin couples are from specific cultural backgrounds.
It would be great to meet and talk to some other people that were cousins because you don’t really get to share their experiences. Maybe that is a female thing where I am always saying I would love to share experiences but it is just interesting. But maybe you know is but maybe I just need to do some research on the Internet and have a look at you know. It is just interesting, I just have this stereotype that most cousins must be from a middle eastern or European background. It is interesting to hear that lots of cousins don’t. [Anne, <50, counselled]

I haven’t heard much about cousin marriage in Australia, ... I don’t think I’ve ever met anyone who is married to his cousin. It would be quite interesting to know more about other people. ... I mean it would be interesting to meet other people, or read about, not necessarily meet, other people’s experiences as well as I think. [Misha, <50, counselled]

I did see that thing on TV a few months ago about the cousin couples and they were saying about how many there are in WA. And I didn’t realise there were actually that many cousin couples. [Nina, <50, counselled]

There was a lady who lived opposite me, a Punjabi lady and she had 12 kids ... I found out her and her husband were cousins and then this one had married this cousin and then right throughout the family had all married family members and cousins and whatever else. And before I met this family I thought it was all a joke. I honestly thought I was a very rare breed of person. I really did for a long time. But then I live across the road from this vast family and they actually all are cousins, or some relation whether its first, second or third. And it was interesting and it made me feel a lot better <laughs> I thought I was all alone. [Lee, <50, counselled]

Challenges have built resilience

For some participants, their consanguineous relationship had been both a negative and a positive experience. Some even felt that the negative aspects of being in a relationship with their cousin were also positive, because they led to personal growth or other affirmative experiences. Their struggles and challenges had built resilience.

I just feel like everything has happened has happened for a reason and I just feel that it has just made me a better person because of it I am more understanding towards other people. I think it has been a great learning curve. It has helped me to
mature a lot. I was very young when I left home so I had to grow up really fast to be able to prove, I think that has been my main goal I wanted to prove to everyone that there is nothing wrong with me, there is nothing wrong with Simon, we can do this and we gonna prove everybody wrong you know. We can turn around and say that nothing can really tear us apart we have been through too much to for anything to tear us apart. So I think that really a positive thing. [Natalie, <50, counselled]

It was all about betrayal and rejection. But it made me stronger in the long term and that is a good thing <laughs>. [Lee, <50, uncounselled]

**Stereotypes and stigma**

Participants indicated that they felt society was against consanguineous marriage and that stereotypes and stigma persisted. Some participants were motivated to become involved with the research in the hope they could contribute to breaking down the stereotypes and stigma attached to consanguineous marriage.

It also interesting that the stereotypes that you have. Because you just have these connotations of uneducated people who just end up getting trapped into getting married to arranged marriages to their cousin. But obviously it’s not. [Anne, <50, counselled]

We’re surrounded by the jokes. It’s on TV, it’s in the movies, it’s, where the hell, I don’t know where it started from though. But then maybe it’s the same place where Irish people are stupid and Jewish people are tight with money. It’s stereotypes that are handy when you’re making jokes about people. [Sophie, <50, counselled]

I just hope that this goes some way towards breaking down the stigma that is there because it is rife. [Lee, <50, uncounselled]

Hopefully, somebody like you might be able to expel the myth and help people like us live without having to be secretive. Because it’s the ignorance of people that cause our secrecy. I just like the idea that someone was looking at it more. And maybe in the future everybody will be a bit more educated about it and it won’t be so weird. [Sophie, <50, counselled]
Theme – genetics and genetic counselling

The following themes relate to interview data from nine of the 16 participants that had been to genetic counselling. Some comments from the participants who did not have genetic counselling have been included for the purpose of comparison.

Genetic counselling helped the couples

As the questionnaire data revealed, consanguineous couples sought genetic counselling for many different, and often multiple, reasons. Interview participants reported seeking accurate information about the genetic risks. Some were searching for an explanation for the cause of a birth defect in their child and whether or not it was related to consanguinity. In Vicki’s case, her daughter’s birth defect was an autosomal dominant condition and thus not related to the consanguinity.

It was quite some time ago and I think because I walked away feeling happy with it, I don’t remember the details now. Which is really mad because I’m pregnant now. But I remember what I took away from it is that the risk, if we walked in as a normal couple the risk was very small. Very small, of having some kind of major problem. And we’d doubled it by being cousins. So it was still really small. And that’s what I remember and that’s all I wanted to know. [Sophie, <50, counselled]

Why did this happen? Why have I got this child with birth defects? Was it because of us? Clarify this. Was it because of us? Was it because of where Steve worked? Was it a hereditary thing that I wasn’t aware of in his family? That, you know, and we’ve compounded it. I needed to know. It wouldn’t have changed anything but I needed to know. [Vicki, <50, counselled]

Genetic counselling provided assistance to the couples making decisions about whether or not to marry and have children. As this is one of the goals of genetic counselling, this is not surprising. However, the interview data shows the impact that the information given in genetic counselling can have on people’s lives and decision making.
Because we were cousins we went and had genetic counselling. And so before we had kids we knew that we didn’t have any genetic disorders or any of the common genetic disorders to pass on in the first place, you know. We were more sure about, we ended up with a lower risk factor than two people off the street. And we knew that and that was really good. And I probably wouldn’t have done that if I’d just married someone, you know, that I just met. ... I think all close marriages, or people that are considering having kids from a close relationship they should be having genetic counselling. That’s right, to be able to make informed decisions. [Jenny, <50, counselled]

To have children and to make sure that, whether or not we could, that was all ... I think I thought that other people worried about our cousin relationship with the children thing. Having counselling would give me the ability to say “But we know that that’s not a problem”. And we did that. We told the family we’d have genetic counselling and at the time and before we got married so that they were aware that we were looking into it. [Sophie, <50 counselled]

Melanie described the value of genetic counselling. She and her partner were not aware that genetic counselling was available and made the decision not to have children. She went on to say that if they had more information, some advice about the risk and the risks put into perspective, their decision might have been different.

We just thought that you couldn’t have children because we were related and it is like a sin to marry and you would get abnormal children ... I would say probably about 60% that we would have thought about it more had we known that you know, like you said, the risk weren’t as bad as what people lead you to believe. [Melanie, <50, uncounselled]

Participants that had received genetic counselling were of the opinion that it should be recommended for consanguineous couples, because they felt it could provide reassurance and accurate risk assessment.

I think it is a good thing to put your mind at ease but I don’t think that you know it’s – <pause> I think it depends on the individual. [Anne, <50, counselled]

I actually know a girl who married her cousin as well. We used to work together ... she lost her first child and ... she actually went back to the genetic counsellor, she
went originally and she wanted to know why. She wanted an explanation because she thought it was because of the fact that her and her husband were cousins. And they kept reassuring her ... So she tried again and she has had two healthy girls. ... The genetic counselling is so informative and so helpful so extremely helpful so no problems even if I had to do it again or even advise anybody I’d be the first person to say “Go and get genetic counselling if you feel like you need it. Go and get it because it is so good”. [Natalie, <50, counselled]

I think there is definitely a role for it for the people that didn’t have that information and might think there wasn’t any problem. And they could have a known genetic thing in their family but not realise they could be at risk. I think it would probably be useful. [Ailsa, >50, counselled]

After having genetic counselling, participants felt reassured, supported and normalised, less isolated, and had a sense of regaining some control over their situation. The benefits came from learning that they were not alone, having information presented to them by health professionals and feeling like they were not being judged. For others, there was a sense of regaining control over their situation.

I think I was glad to have done it because I had that need to kind of, feeling of doing whatever you can do. You’re kind of powerless anyway. So if you’re in that situation then you kind of feel like you have to cover all the bases and make sure that you’re getting a very good ultrasonographer and getting genetic counselling and trying to cover as much as you can. ... Yeah, so in that sense that was useful and reassuring. [Misha, <50, counselled]

It was great. The doctor made us feel really comfortable and he was like God you are not the only people that come in here please don’t think that you are the only people and you are the most abnormal people in the world because that is just so not true. [Natalie, <50, counselled]

Fantastic experience. But then, you know the information we were given was reassuring, and I don’t just mean, we got good results, we didn’t have a problem, so that is a very good outcome. But I mean that when we went there and we found out there were so many other couples in WA like us that it wasn’t all a cultural thing. That there were other people just like us, not necessarily Italian or you know,
something like that, that were in this situation. We weren’t two-headed <laughs>. I suppose that was really comforting to find out. We walked out feeling empowered but also the knowledge of you know, these are the risk factors. It was just a really good experience. [Jenny, <50, counselled]

What we got from the counselling was everything we came for. We left realising that we weren’t weird, that there was a whole bunch of other people out there doing the same thing and thinking there was absolutely nothing wrong with it, that our risk of having children that were completely deformed was not much more than the regular population. Those were the things we came for and we took away with us. [Sophie, <50, counselled]

After receiving genetic counselling, participants were able to use accurate information to counter any objections or false claims from their relatives. In addition, the new knowledge gave them the tools to help deal with some of the negative reactions.

I think that my parents were very, very concerned about me getting pregnant because again of the whole thing down in society there has always been this thing of having abnormal children. I went for my genetic counselling. I sent them the letter we got from genetic counselling saying there shouldn’t be a problem there was a one in whatever chance and that reassured them a lot that everything was going to be OK. But even now like my Mum will say to my are you thinking of having another one and I say “Yeah I’ d like Kyle to have a brother or sister” and she goes “Oh do you really think you should be pushing fate” and I say “Mum it is not fate. There is nothing wrong. There is not fate”. [Natalie, <50, counselled]

To have the ammunition for anybody who would doubt our decision. We had medical facts that we could back up what we were doing. Because it is a worry, it’s still a worry you know that we’re having a baby. It’s still a worry I have to say, but it’s the worry comes from what people would say, what happens if we do? What happens if we do have ... you know, it isn’t our fault. [Sophie, <50, counselled]

Risk perception and risk avoidance

All genetic counselling clients perceive genetic risk differently and the participants in this study were no different. Some were not reassured by the risk information
given and perceived the risk of having children with a genetic abnormality to be high. Others were not concerned because they considered the risk to be low.

I think it’s very personal because it depends what you as a person consider to be a high risk. So whereas the person who was counselling me, who was lovely, thought that he was possibly reassuring, I found the risks quite high. So and that was the first thing other people would be told if they went and probably think it was very low and it wasn’t a risk that stopped us obviously, but it was certainly was there to carry through as a worry. [Misha <50, counselled]

I think I was never really that anxious about it to be honest with my first two pregnancies. When I was pregnant you know a lot of people go through the normal anxiety about something being wrong with the baby. I mean I was happy when we heard that were weren’t carriers of CF but I wasn’t really anxious about anything. I just felt “Oh they’ll be alright”. Which is possibly just a protective mechanism. [Ailsa, >50, counselled]

As soon as that baby is born you always ask “is it all right?” [Linda, >50, uncounselled]

The risk clearly played on the minds of participants and their families and many discussed the strategies they had considered to reduce or avoid the genetic risk.

If it comes to that and we can’t have kids, then we’ll do something, like we’ll get a donor egg, or something, you know. Or we’ll adopt or something. [Jenny, <50, counselled]

I suppose if Dan and I had done genetic counselling and we found out that there was going to be major issues with genetics I think that would have played on our minds quite a bit in the fact that it would have strained the relationship and may have ended it because we both wanted children. ... My grandmother suggested that we have one child that is my egg and another man’s sperm and one child that is Dan’s sperm and another woman’s egg so there is no chance of having a deformed child. [Nina, <50, counselled]
Fear of abnormal children

Fear of having abnormal children, both from the participants themselves and their relatives, was raised often throughout the interviews. The negative reactions from friends and family about the consanguineous relationship and risks of having children translated into heightened anxiety, to the point where many couples contemplated not having children.

When we planned to have our first baby we went for genetic counselling straight away. You know I think that you can’t help but feel more wary because obviously you are cousins and there is a greater chance whether it be just a small percent but still there is a greater chance ... so that was a problem. And I think also that kind of was why we put off having children for a long time because we just had to make sure in our minds that if something was wrong that there would be no oh well we shouldn’t have done that. [Anne, <50, counselled]

Oh, the worry about children. That does still, we’d like to have another child and so it does still whenever during the pregnancy I was quite worried about birth defects and just the increased risks. So that’s definitely an issue for me. It won’t go away. [Misha, <50, counselled]

Those are the sorts of things that we have to cope with now, if people know we’re having a baby. We’ll have to worry that we are deliberating imposing something on our baby, like a third head. You know? Do you know what I mean, the jokes? Being married is one thing, having a baby, some would say is irresponsible. [Sophie, <50, counselled]

The one thing that worries us with having kids, like Dan especially is very worried about having something wrong. But we don’t even know what could go wrong. So it is sort of one of these things we keep getting told that we are going to have deformed children but we don’t know what deformities or anything like that. And I don’t know anyone else that has married their cousin that has had kids so we can find out ... I told my doctors that I am married to my cousin and that we have had had genetic counselling and they say “Ok”, they don’t really approach the subject either so none has really even discussed anything with us. Because I didn’t see a genetic counsellor with Dan I didn’t ask and Dan is not the type of person to come out and ask either. [Nina, <50, uncounselled (partner was counselled)]
Family members, friends and others also feared that abnormal children would be a problem for the consanguineous couples. They often commented and let their opinions be known to the participants.

My Mum was happy in a way but not happy in another way because she was very concerned about the kids. But my Dad was angry, he was really angry. I was shaking when he came home. Well, he asked and my father say “Ok you can get married but your children will be always abnormal, all of your kids”. [Bonnie, >50, uncounselled]

You will have disfigured or disabled children, so that was her [mother-in-law] issue. And that’s what he was saying to her. That’s your issue, if you can prove that to me, you know, if you can prove me that’s something dangerous about what we’re doing then come and tell me and I’ll listen to you. But until you can come up with some facts keep your opinions to yourself. So that sort of, you know went on for quite some years. It’s much better now. [Jenny, <50, counselled]

The idea that kids are more prone to birth defects that seems to be very common. Like sometimes, in fact, that’s something that does follow on sometimes from us saying that we’re cousins. [Misha, <50, counselled]

Understanding the genetics and risk

The level of accuracy and understanding of the genetic risk varied amongst the participants.

I think it doesn’t matter if you’re cousins, if you’re related or not, you’ve always got a chance with something going wrong. But if you are cousins you have a greater chance to have some problems. [Bonnie, >50, uncounselled]

I think it is pretty much the same as everyone else, I think it is like 2% different for cousins couples is the statistics of it. And I just thought well 2% I think it is 94% chance of having a normal baby and for a normal couple it is 96% chance or something. So for 2% I don’t think it is that much and not so big a problem to risk not having one. [Natalie, <50, counselled]
They say well first cousins, but it’s second cousins that is more difficult, because first cousins they thought well don’t even think about which I think that’s wrong. Perfectly wrong. [Eric, >50, uncounselling]

Many people overestimated the genetic risks and experienced great anxiety as a result. After genetic counselling, they were informed more accurately about the risk and felt reassured.

I honestly thought that I could have a two headed baby with 16 toes and an IQ of zero. I really thought that that’s what could happen. [Lee, <50, uncounselling]

We thought it was huge. We thought that even though I’d seen, I knew of other couples that were married including the famous ones and I knew of babies that had been born of couples that didn’t have anything weird going on with them, anything obvious anyway. I still had a feeling they were just lucky. But when I left here I knew that they weren’t just lucky, that the chances were not greatly inflated, like you said, by being related. [Sophie, <50, counselled]

5.4. Discussion

5.4.1. Being in a consanguineous relationship in WA

Over half (57%) of the couples who presented to GSWA for genetic counselling between 1975 and 2001 identified with a Christian religion and most were from backgrounds where consanguineous marriage is not routinely practised (Port et al., 2005). The demographics of questionnaire respondents and interview participants in this study were similar to this.

The majority of respondents were in a first cousin marriage or relationship and this is the most popular form of close kin union worldwide (Bittles, 2008). In this sample of consanguineous couples, the 15% rate of birth defects is higher than would be expected but is possibly due to selection bias as the majority of participants were ascertained following their referral to a clinical genetics service.
Consanguineous couples indicated that they face a number of challenges regarding marriage and having children. These issues included opposition to their relationship and negative responses from family and friends. Many issues relating to family dynamics, the personalities involved, and opposition to the relationship due to other factors such as young age, family values and career progression were not specific to the consanguineous relationship.

5.4.2. Dealing with negative reactions to consanguineous relationships

The interview participants reported negative experiences related to their consanguineous relationship including their family’s reaction, suppression of their feelings because they felt like they were doing something wrong and keeping secrets from family and/or friends. Many friends and family of the participants interviewed expressed the view that consanguineous relationships are wrong, distasteful, dangerous or undesirable. However, positive experiences were also described. Some participants reported that their relationship might have been enhanced because they were related and had a closer relationship due to a shared history and background. The positive and negative aspects of being in a consanguineous relationship are summarised in Figure 5.10, with a greater number of negative social issues raised than positive ones.
The interview participants cited reasons for people’s negative reactions to their relationship that included fear of abnormal children, ignorance about actual genetic risks of consanguinity, the legal implications of cousin marriages, religious beliefs, fear of the family being judged by others and confusion between consanguinity, inbreeding and incest. People’s reactions were influenced by how information was presented and the participants’ (couples) own feelings about, and level of security with, their relationship. The strength of people’s reactions and their willingness to voice them openly when the participants revealed the consanguineous relationship was surprising.
Participants who began a relationship with a cousin they had grown up with (rather than those who met as adults) were more likely to experience negative reactions. Couples who grew up together were likened to siblings, whereas couples who met as adults were seen as strangers falling in love. A relationship with, or marriage to, a cousin that is more like a sibling may prompt a reaction related to the concept of incest. Similarly, thoughts of so-called inbreeding and the concomitant (often overestimated) high risk of birth abnormalities are mistakenly applied to first cousins. The origin of these beliefs is not clear but probably arises from a combination of factors including religious dogma and the ethics and morality of the individual, family or society (Bittles, 2008). Interestingly, many consanguineous couples disapproved of their children entering a consanguineous relationship. They suggested that this would represent another level of inbreeding which may be dangerous and that they wished to protect their children from having to face the negative issues they experienced. Future research could be directed towards whether people draw a line between acceptable and unacceptable relationships and at what point they decide that a relationship is incestuous.

Some of the participants had not married, as it went against their family’s wishes. Others had waited for many years until the deaths of relatives who opposed their relationship or until their first non-consanguineous ‘acceptable’ marriage had ended. Others had become estranged from their family or moved away from their family in order to be together. Isolation was a major problem for many couples. Even those who had reasonably accepting families felt they could not be open with friends and acquaintances for fear of rejection and ridicule. Many feared this would also be experienced by their children.

5.4.3. Genetics and risk

Some participants expressed fear of the genetic risk involved in having children and were concerned that a child with a genetic condition might make life more difficult. They feared being blamed, or receiving “I told you so” comments if they had
children with birth defects and that their children might have to deal with people’s negative reactions to their consanguineous relationship.

The risk perception amongst study participants could be related to their prior experience of having children and the univariate analysis performed did not take this into account. Some participants described their level of worry about birth defects as similar to other couples’, yet others had an increased level of worry because of their consanguineous relationship. Many felt reassured after they had received accurate information about the risk during genetic counselling but others did not have an accurate recall of the risk figures or had been given inaccurate information by medical professionals.

For some participants, their contact with GHPs may have been the first opportunity to access accurate information about genetic risk. Published data, along with data presented in this chapter, show wide variation in the risk figures given by genetic counsellors (Bennett et al., 2002). This highlights the importance of developing standardised information and the need for this information to be given to consanguineous couples in genetic counselling sessions.

5.4.4. The experience of genetic counselling

The participants found that genetic counselling was useful for determining their genetic risk and enabled them to correctly inform relatives who tended to overestimate this risk. Normalising the relationship and experiences also brought relief and reassurance, and hence the participants were prepared to recommend to genetic counselling to others. Some participants stated that an unhelpful aspect of genetic counselling related to the lack of definite answers or gene tests. This could suggest a failure of the genetic counsellor to recognise or address the expectations of the client prior to the consultation. Other unhelpful aspects of genetic counselling, as identified by participants, included the inconvenience involved in having to travel to the clinic.
Some people indicated they may not have continued the relationship or had children if the genetic counsellor had said the risk was too high. It was clear that the information given, and the way in which it was given, had a large impact on the lives of these consanguineous couples.

Questionnaire respondents had no particular preference regarding matching of cultural background with the counsellor and themselves, with many stating that cultural background did not matter as long as the counsellor was competent and interested in them. This is a common opinion amongst clients of counselling services (McAllister et al., 2008b; Meyer et al., 2013). The nature of individual cultural differences and perceptions of the communication process was not explored in the questionnaire. Future studies could examine how cultural differences influence the experience of genetic counselling.

For couples dealing with the genetic and social issues related to their consanguineous relationship, GHPs may be one of the only sources of support and accurate information. Some participants assumed that all their children were at risk of serious birth defects and they were relieved when the genetic counsellor explained that the risk was much lower than they imagined. Other participants were relieved to hear that their relationship was not unusual and that other cousin couples existed. They appreciated having someone to speak with who was not judging them as being strange or doing something wrong. These results suggest there is a potential for genetic counsellors to break down some of the social isolation these couples feel, to let them know that other couples enter into consanguineous relationships and that consanguinity is an accepted form of marriage in some cultures.

The stigma attached to consanguinity in our society may reduce access to genetic counselling (Read et al., 2012). To get the necessary information to consanguineous couples and to provide support, the option of genetic counselling needs to be made
available to these couples. Potential options for promoting genetic counselling may include educating GPs, priests or other health care professionals who might have contact with premarital and prenatal couples (Bishop et al., 2008). In addition, there is the potential to let people know about the availability of genetic counselling when obtaining permission to marry. In Australia couples are required to submit an intention to marry form which contains a question about whether or not the two parties are related.

The optimal time for contact with consanguineous couples might be when the relationship begins. This may allow genetic counsellors to mitigate concerns about whether it is wrong and to have some facts with which to inform doubting or fearful family members. Alternatively, genetic counselling may be optimal when consanguineous couples are considering having children. This latter option appeared to be the most desirable option for the couples in this cohort.

Of the nine interview participants that had genetic counselling, none described the experience in negative terms. While this may have been a true reflection of their experience, consideration also should be given to the possibility of ascertainment bias, or the participants perception that they should provide socially acceptable responses to a person representing the profession about which they are being questioned (Gu et al., 2011; Lee et al., 2009).

Genetic counselling was generally seen as a solution that can restore balance to the anxiety around having children, because it is a source of accurate information. It also has the potential to help consanguineous couples address the social stigmas and anxieties they experience, because it can assist in putting the relationship into perspective and empowering people to combat negative social responses.

5.4.5. Consanguinity as a form of cultural difference

Consanguinity is a presenting issue that illustrates some of the cultural factors that genetic counsellors and clients bring to the genetic counselling interaction. While
there is a high incidence of cousin marriages within particular migrant groups where consanguineous marriage is accepted and encouraged, in many Western societies, cousin marriage is often seen as a strange or even incestuous practice.

Consanguinity is uncommon in Australia and thus many genetic counsellors have little or no experience of counselling consanguineous couples. There is also a lack of literature to guide counsellors in managing consanguinity in the Australian context. The research results discussed in this chapter highlight some of the challenges facing the genetic counselling profession in the context of increasing cultural diversity and hybridity. While the issue of consanguinity is a relatively straightforward concept from a genetics perspective, many issues unrelated to the genetic risk arose in the questionnaires and interviews. These included how individuals, families and Western society respond to people who participate in activities that are deemed unacceptable.

5.5. Study limitations

It is important to acknowledge that the research discussed in this chapter has a recruitment bias. In addition, the response rate from the 262 individuals/couples in consanguineous relationships who had received genetic counselling at GSWA was relatively low (16%). It is possible that only those reasonably comfortable with their relationship agreed to participate in the study and it cannot be determined whether non-respondents differ in significant ways from respondents. The questionnaire participants were all sufficiently confident in their English language skills to complete a questionnaire written in English. Accessing a more diverse range of couples may have added depth to the investigation.

The questionnaire itself also had some limitations. The media release stated that genetic risk for first cousins was not significantly higher compared to other couples which could have biased their perception of risk. Some of the questionnaire items
such as present level of worry and reason for level of worry for those no longer intending to have children would be difficult to answer.

Despite the questionnaire items requiring a yes/no answer or a Likert scale response, a number of participants added written comments giving unsolicited responses. The following is one such example.

I don’t really agree with marrying or having sexual relations with your cousin. I think at a societal level our gene pool is better without this problem. However, if it’s your cousin you fall in love with and are compatible with then so be it. I am no longer in a sexual relationship with him. We are very firm and devoted friends, with a deep bond (probably a family bond). I am no longer in this relationship sexually as I felt it was inherently wrong. I wasn’t comfortable with people’s judgments.

The data obtained from 16 interviews cannot be seen as representative of all consanguineous couples. Rather, this work can be viewed as an exploration of the experiences and issues that commonly arise for people in a relationship with their cousin. Aside from the two retired couples interviewed, all the participants were female and the perspective of more males would have added to the data and potentially highlighted differences due to gender and gender role expectations. By conducting the interviews during office hours, the ability of males and working couples to participate was limited.

5.6. Summary

As far as I am aware, this is the first study of its kind exploring cousin couples’ experiences of being in a relationship with their cousin and whether they found genetic counselling useful. Because most of the couples were from Western countries and not from countries where cousin marriage was common, they felt stigma and isolation from their peers, many had not been totally supported by their relatives, some had been ostracised from the family, and relationships had been destroyed. Previous studies have tended to focus on the issues of biological
outcomes and genetic risks, or the cultural reasons for preference of consanguineous marriages. Hussain (1999) highlights the importance of research studies that provide an understanding of the socio-cultural context in which consanguineous marriages take place. The effect on the rest of society’s assumptions and ideas about the practice has not been explored.

It cannot be assumed that all consanguineous couples come from communities where this is a traditional type of marriage and that the couples’ family and peers are comfortable and accepting of their relationship. Comments made by some participants showed that they are unaware of the supportive role genetic counsellors can play. In some situations, the genetic counsellor is the only available sympathetic, non-judgmental person and thus is in a unique position to offer support and to share tools for managing the ramifications of societal responses and stigmas. Exploring each couple’s experiences and expectations of genetic counselling would allow their individual story and lived experience to be heard and individual objectives to be determined.

Many of the social issues facing consanguineous couples were substantial. This work demonstrates the need for awareness of the psychological impact in addition to the genetic risks, where the cultural context is diverse. Genetic counselling goals may be best met by adopting a more dialogical model that places greater emphasis on the social issues and encourages skilled transcultural genetic counsellors, who are not necessarily culturally similar to their client, to attend to each client as unique individuals, exploring their story and identifying the issues important to them.

The next chapter explores the opinions and experiences of Australasian genetic counsellors with respect to working with cultural difference and consanguinity. The chapter examines the cultural awareness they bring with them to the counselling interaction, how this influences communication, and their skills and ability to negotiate cultural differences.
6. Genetic counsellors’ views on consanguinity and cultural difference

While in genetic counselling (as in most health occupations) diversity is mostly regarded in terms of ethnicity, race and gender, it is important to recognize that everybody contributes to diversity because each one of us is different from the majority in at least one way. (Mittman et al., 2008, p.301)

I don’t think it is realistic to “know” the ins and outs of every culture and religion. I think the best that teaching programs can do is make us aware that differences do exist plus that we must view each client separately and learn about them as they come. (GC48)
6.1. Introduction

Chapter Five examined the perspective of WA couples in consanguineous relationships. The chapter focussed on their life experiences and their experiences of genetic counselling. In this chapter, the other side of the dialogical process is examined: the ideas and opinions about cultural difference and consanguinity that genetic counsellors bring to the interaction.

Just as clients are unique individuals, so too are genetic counsellors. While all counsellors are provided with similar training, their life experience, personality and biases inevitably influence the messages they convey to counsellees. This chapter examines how counsellors apply their training and life experiences to recognise and respond to differences between themselves and clients.

In a survey of heads of genetic counselling training programs and their graduates, Punales-Morejon & Rapp (1993) found that there were irreconcilable differences between the perceptions of what it is necessary to aim for, and possible to achieve, in training counsellors to serve an increasingly multicultural client population. Instructors felt they did not have the time or resources to teach subject matter relating to medical anthropology and cross-cultural counselling. Conversely, graduates felt they had not been equipped in their training to manage the cultural differences they encountered. Several years later Rapp (2000) interviewed genetic counsellors in the USA about the genetic counselling curricula and found that it paid scant attention to these cultural differences and cross-cultural counselling.

Despite the emerging models of genetic counselling that value person-centred counselling in developing cultural competence, there remains a tendency to group individuals together rather than recognise the uniqueness of individuals. There is a need to identify any barriers preventing genetic counsellors from implementing these more dialogical models of genetic counselling, so that practice guidelines and educational strategies can be developed to overcome them.
6.1.1. Aims
The chapter examines Australasian genetic counsellors’ views and experiences concerning cultural diversity. The impact of increasing diversity on practice, factors contributing to cultural difference, and how practice is modified in response to cultural diversity is explored. Genetic counsellors’ views on consanguinity and the associated genetic risks, how they work with these clients and whether consanguinity is perceived as a point of cultural difference were examined. Demographic data about genetic counsellors in Australasia were obtained to examine diversity within the genetic counselling workforce.

6.2. Methods
The methods used to generate the data in this chapter are described in detail in Chapter Four. In summary, a structured questionnaire explored Australasian genetic counsellors’ opinions of cultural diversity and consanguinity. A discussion group with a small sub-set of seven counsellors then explored these issues in more detail.

6.3. Results

6.3.1. Questionnaire

6.3.1.1. Demographics
Sixty one genetic counsellors responded to the email invitation to participate and gave their postal details. Questionnaires were mailed and 58 completed questionnaires were returned giving a response rate of 29% (58/200). The majority of respondents were female Caucasians and only two were male (7%). The majority of respondents were Australian-born (Figure 6.1). The nationality of participants is shown in Figure 6.2. The primary spoken language was English (Figure 6.3). The majority of counsellors had postgraduate qualifications (91%), 5.5% had other tertiary qualifications and 3.5% were hospital trained. The majority of respondents practised in a metropolitan unit (64%) with the location of practice reflecting the
distribution of counsellors’ numbers throughout Australasia. The greatest number practised in NSW (31%) followed by VIC (19%), WA (17%), QLD (16%), NZ (9%), SA (5%) and TAS (3%).

Figure 6.1 Country of birth

Figure 6.2 Nationality

Figure 6.3 Languages spoken
6.3.1.2. **Opinions of cultural difference**

Genetic counsellors were asked to state the advantages and disadvantages of a multicultural society. A definition of the term multicultural was not provided, in order to determine their understanding of the term. Multiculturalism as a concept was first raised in Australia in the 1960s to promote the integration of new immigrants, foster the notion of protecting the rights of ethnic minorities and is defined as the coexistence of culturally diverse groups (Meer et al., 2011). In their responses (Table 6.1), the counsellors described the advantages of multiculturalism in terms of inclusion and acceptance of the differences between discrete groups. Respondents were able to suggest multiple factors for each question and more disadvantages were raised than advantages. Four respondents stated that there were no disadvantages to multiculturalism.

Participants were then asked to list the four most important factors that contribute to cultural difference. The responses are shown in Table 6.2.

<table>
<thead>
<tr>
<th>Table 6.1</th>
<th>Genetic counsellors’ opinions of multiculturalism</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Advantages</strong></td>
<td><strong>n</strong></td>
</tr>
<tr>
<td>Acceptance of other’s culture</td>
<td>20</td>
</tr>
<tr>
<td>Diversity</td>
<td>17</td>
</tr>
<tr>
<td>Tolerance</td>
<td>15</td>
</tr>
<tr>
<td>Education</td>
<td>9</td>
</tr>
<tr>
<td>Enrichment</td>
<td>8</td>
</tr>
<tr>
<td>Awareness of diversity</td>
<td>4</td>
</tr>
<tr>
<td>Broader perspectives</td>
<td>3</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Table 6.2  Factors contributing to cultural difference

<table>
<thead>
<tr>
<th>Factor</th>
<th>n</th>
<th>Factor</th>
<th>N</th>
</tr>
</thead>
<tbody>
<tr>
<td>Religion</td>
<td>40</td>
<td>Gender</td>
<td>5</td>
</tr>
<tr>
<td>Language</td>
<td>35</td>
<td>Age</td>
<td>4</td>
</tr>
<tr>
<td>Values/Belief systems</td>
<td>30</td>
<td>Experiences</td>
<td>1</td>
</tr>
<tr>
<td>Family structure</td>
<td>23</td>
<td>Wealth</td>
<td>1</td>
</tr>
<tr>
<td>Appearance/dress</td>
<td>16</td>
<td>No response</td>
<td>6</td>
</tr>
<tr>
<td>Ethnicity/race</td>
<td>11</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

The genetic counsellors were also asked whether cultural difference affected the genetic counselling interaction. If they thought a culturally different background did have an impact on their counselling, they were asked to explain this further. All respondents said cultural difference did affect the counselling interaction. The stated ways that counsellors thought cultural difference impacted on the counselling interaction were: communication barriers, the need to use interpreters, the need to take more time, counsellors paying more attention to body language and language use, different interpretation of information, different thought processes and decision making styles in clients, different perceptions of genetics and risk amongst clients, individual versus family ownership of genetic information, and counsellors having a lack of knowledge of cultural norms and taboos.

One counsellor explained that to be culturally aware, you need information about the specific culture of the client. This counsellor commented that “preparation needs to be attended to. The genetic counsellor must know important aspects about the culture to counsel individual, couple, extended family” (GC44). Other comments indicated that some counsellors took a broad approach to cultural difference in their counselling interactions, commenting that “all interactions are between culturally different people” (GC56), and “it [cultural difference] may affect the interaction … but this should not be assumed” (GC27) and “different perspectives will influence any decisions and how information is used” (GC8).
Another counsellor highlighted the potential negative consequences of not taking cultural difference into account:

Decrease in effective communication, genetic counselling approach is different e.g. non-paternalistic/nondirective vs. paternalistic/directive. Alters the genetic counsellor’s way of conducting session with a decrease in effectiveness. [GC13]

The questionnaire then asked the genetic counsellors if they felt their training had equipped them to deal with cultural differences between themselves and their clients. This item involved a 4-point Likert scale from completely to not at all. No respondents thought their training completely equipped them to counsel culturally different clients, 12% of respondents indicated that their training was not at all adequate and the remaining 88% felt their training slightly equipped them or somewhat equipped to deal with cultural difference. Many counsellors commented that their training “did not allow for in-depth consideration of multicultural issues” (GC3) or overlooked “vital issues of multiculturalism” (GC20) and did not provide “training on cultural differences” (GC23). One counsellor commented that their training was “completely inadequate to train genetic counsellors in areas of cultural difference” (GC28).

Many stated they had learnt more from practical on-the-job experience and from their personal experiences: “the only way to really understand a culture is to live in it therefore a genetic counsellor will rarely have a full insight into a different culture” (GC1) and “basic information came from training but cultural specific came from individual families” (GC9). Some felt these lessons were possibly more valuable than courses. Some sought their own training. In their responses they also noted that there was a lack of relevant (Australian) literature on the topic. Others thought that “I don’t know that this training is possible, you need to ask each individual what their beliefs are … if possible” (GC16), but some acknowledged the value of a broad view of diversity and an individual approach in counselling. One counsellor who subscribed to this view explained that:
I don’t think it is realistic to “know” about the ins and outs of every culture/religion. I think the best that a teaching program can do is make us aware that differences do exist and that we must view each client separately and learn about them as they come. [GC48]

When asked whether there were any differences in manner, approach to their clients or reception by clients, between male and female genetic counsellors, 33% of respondents felt that there was a difference, 2% thought there was no difference, 24% were unsure and 40% did not know. Those that were unsure or did not know indicated they had not had the opportunity to draw comparisons with regard to gender differences in the approach to counselling, as they had not worked with any male genetic counsellors. Many felt that “there are differences between all genetic counsellors male and female” (GC27) and commented that the way counselling was delivered reflected individual counsellor differences such as “manner, approach etc. [which] are individual variables, not ones confined to gender groups” (GC47).

Some respondents indicated that clients may react differently to male genetic counsellors than they would to a female counsellor. These respondents commented that “perhaps in areas like prenatal diagnosis and hereditary breast cancer – women may prefer to discuss the topics with women” (GC22), that matching of gender between client and counsellor “may be helpful in male to male counselling sessions” (GC35), “a client may have different expectations of the genetic counsellor depending on their sex” (GC21), and in some “cultures that traditionally place women in traditional roles” (GC51) clients may find it difficult to seek help from female professionals.

Other responses indicated that the behaviour of male genetic counsellors may be different from female counsellors. A male respondent (GC19) observed that there was a “huge difference between the way I engage and empathised with patients compared to my female colleagues”. Other comments reflected gender stereotypes
such as “many clients (and doctors) feel males carry greater authority” (GC15), “geneticists who are males may be taken as having more authority in some cultures” (GC16), “males are not as comfortable discussing female issues and prenatal information, also they are generally not as perceptive” (GC50), and “possibly males are more directive” (GC56).

6.3.1.3. Cross-cultural genetic counselling practices

The questionnaire also examined genetic counsellors’ views regarding the importance of cultural awareness. Awareness of the cultural and social background of clients was highly valued, with 98% of respondents indicating on a 5-point Likert scale that they thought cultural awareness was important (53%) or essential (45%). The remaining 2% were undecided and none felt it was unimportant or not very important.

The reasons why respondents thought it was important to be aware of these issues included factors that influence genetic counselling practice, such as the provision of the “best possible service” (GC1), to enable counsellors to “tailor the session to the individual” (GC10), to obtain a “complete medical and social history” (GC32), to examine “ancestry and social environment” (GC34), to assist in “responding to the individual as opposed to the ‘culture’” (GC39), “to communicate more effectively and alters the information giving process and extraction of information” (GC41), and to provide the “genetic counsellor a framework from which to explore things” (GC40). Additional factors related to the way in which cultural awareness impacted on clients, including to understand why clients react “in a certain way and why they made a decision that way” (GC43), how “it affects rapport and can influence what they tell you and how they open up” (GC53), and the “impact or influence on their decisions” (GC54). One respondent commented that “we are so enveloped in our ‘Western culture’ that we can’t see that it influences everything we think and do and likewise for other cultures” (GC48).
Several respondents (n=6/58) raised system constraints to adopting a broader more dialogical perspective to diversity issues, such as not having “the luxury of knowing a lot about the background of our clients” (GC25), and it being “difficult to get an insight into a client’s culture in a limited time, although cultural and religious beliefs will impinge on their decision making” (GC21). Respondents cautioned about the need to “be careful about stereotyping” (GC52) individuals based on culture, were mindful to not “go in with a set hypothesis based on a client’s background” (GC6), and commented that “caution against assumptions and generalisation need to be heeded” (GC20). Another respondent agreed, saying that

My concern is that some people use cultural background as a generalisation. People are still individuals regardless of culture. [GC24]

The majority of respondents (n=52/58) indicated that during a genetic counselling session they always or frequently assessed a client’s prior knowledge about genetics (Table 6.3). They did this by asking questions primarily about the client’s educational experience. Most respondents (n=53/58) always or frequently modified their session to suit their client’s background knowledge of genetics, by varying the language used and complexity of information given. The methods they used to aid in the explanation of complex information to their clients included visual aids, analogies, the use of simple language, avoiding jargon, and giving fact sheets and written information.

Over half of the respondents indicated they commonly modified their session to suit their client’s cultural background (Table 6.3). These modifications included exploring the beliefs and experiences of their clients, and paying attention to appropriate dress, manners and social interactions. If using an interpreter, other modifications included allocating more time, visual aids, using simple language, omitting content and using less detail and the use of pictures. Up to 20% of
respondents sometimes or never individualised their practice according to client knowledge or cultural background.

Table 6.3  Modifications to practice to take into account cultural difference

<table>
<thead>
<tr>
<th>Assessed prior knowledge of genetics</th>
<th>Always %</th>
<th>Frequently %</th>
<th>Sometimes %</th>
<th>Never %</th>
<th>No response</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>60</td>
<td>29</td>
<td>9</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>Modified session to suit background knowledge</td>
<td>64</td>
<td>28</td>
<td>0</td>
<td>2</td>
<td>6</td>
</tr>
<tr>
<td>Modified session to suit cultural background</td>
<td>19</td>
<td>43</td>
<td>22</td>
<td>5</td>
<td>11</td>
</tr>
<tr>
<td>Modified the session if using an interpreter</td>
<td>36</td>
<td>26</td>
<td>22</td>
<td>5</td>
<td>11</td>
</tr>
</tbody>
</table>

6.3.1.4. Opinions on consanguinity

The median number of consanguineous clients seen per year by the study participants was two (range 0-20) with the highest number being seen by a genetic counsellor located in Metropolitan NSW. When asked if they thought the rate of consanguineous marriages was higher in any particular ethnic groups, 54% of the respondents thought that this was true. When asked to list these groups, respondents mixed geographical areas and religion in their answers: 35% specified Middle Eastern communities (Arabs and Lebanese), 5% specified Asians (Pakistan, India) and 14% specified Muslims.

A question about whether consanguineous relationships should be encouraged or discouraged was included in the questionnaire to ascertain the value counsellors placed on the life choices of clients. Almost all (96%) of the genetic counsellors stated they had no opinion on consanguineous relationships, saying individuals should have free choice about relationships they enter into. Only one respondent said consanguineous relationships should be encouraged and one said they should be discouraged. One respondent replied “it is not a counsellor’s place to make such
value judgments” (GC29) and another said that it was “not my role to comment on consanguinity” (GC17). Others suggested “Each to their own. Who am I to say who you can and can’t have relationships with” (GC16), that “surely it is up to the people in the relationship to decide” (GC22) and that “consanguinity is a personal choice and if an individual wishes to marry his or her cousin they should be allowed to do so” (GC23). Two respondents made reference to the preference for consanguineous marriages in some cultures, saying that “they should be respected as the choice of each couple or their culture” (GC8) and “I don’t think it is for me to discourage/change hundreds of years of cultural practice. Individuals who chose consanguineous partners should have freedom of choice” (GC20).

Participants were also asked to rank the four most important factors that affected their feelings about consanguinity. They were then given the option to state other factors contributing to their feelings. The rankings (based on the assumption that 1 is the highest and most important) are shown in Figure 6.4. Additional factors identified included domestic violence, stigma, arranged marriages (with coercion of one party), consanguinity over multiple generations, the counsellor’s own cultural perceptions that it is wrong, legislation, family attitudes and pressures.

![Figure 6.4 Genetic counsellors’ ranking of factors influencing opinions of consanguinity](image)

The next four questionnaire items related to the genetic counsellors’ knowledge, and perception of the risk of having a baby with a birth defect (Figures 6.5 and 6.6).
In both the general population risk and first cousin risk categories, a greater number of respondents underestimated the risk than overestimated it. When asked their impression of the risk on a 5-point Likert scale, the majority categorised both the general population risk (43%) and the consanguineous couples’ risk (45%) as low.

![Figure 6.5 Genetic counsellors’ estimations of genetic risk](image)

![Figure 6.6 Genetic counsellors’ perceptions of genetic risk](image)

Counsellors were asked what advice or information they would give to a consanguineous couple planning a pregnancy. The words advice and information were deliberately chosen in order to gauge genetic counsellors’ response to the inference of directiveness. The respondents’ comments were categorised into those that indicated they would give advice, information and other (Table 6.4). One respondent reacted strongly to the question stating “as with all clients I see, I
conduct myself in accordance with our ethical code of conduct and this is no different when dealing with different cultures and consanguineous couples” (GC42). Two stated clearly that they “would not give them advice about what to do, just give the facts” (GC21) and that “I wouldn’t give advice!!” (GC22).

The majority of respondents concentrated on taking family history details, giving information about risk and the available testing options. Only one genetic counsellor mentioned taking an “individualised approach for each couple” (GC22), although many referred to taking a careful family history and calculating the risk for each couple. Very few counsellors mentioned attending to emotional needs and issues relating to acceptance by friends and society, although one said they would “normalise the whole process” (GC13).

Respondents stated that the most important issues for consanguineous couples were acceptance by society (26%), opinions of family and friends (22%), genetic risk (29%), having access to correct risk information, and the impact of cultural and religious influences (23%). This is in concordance with the issues highlighted by consanguineous couples in Chapter Five.

Respondents were asked to rank from 1 to 4 the relationships that they considered to be incestuous. The majority ranked most highly first degree relationships such as parent-child (62%) and brother-sister (28%), with uncle-niece, aunt-nephew and grandparent-grandchild the next most highly ranked (10%). Five out of 58 respondents (9%) included first cousin relationships in their ranking of incestuous relationships.
Table 6.4 Genetic counsellors’ advice and information for consanguineous couples planning a pregnancy

<table>
<thead>
<tr>
<th>Categories</th>
<th>Quotes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Advice</td>
<td>See a genetic counsellor!!</td>
</tr>
<tr>
<td></td>
<td>Take Folic acid</td>
</tr>
<tr>
<td></td>
<td>Standard pre-pregnancy information</td>
</tr>
<tr>
<td></td>
<td>Pregnancy ultrasound at 18/20 weeks</td>
</tr>
<tr>
<td></td>
<td>Check family history</td>
</tr>
<tr>
<td></td>
<td>Depends on ethnicity</td>
</tr>
<tr>
<td>Information</td>
<td>Risk for autosomal recessive conditions is above population risk</td>
</tr>
<tr>
<td></td>
<td>Genetic testing for recessive condition is available</td>
</tr>
<tr>
<td></td>
<td>Prenatal genetic testing options</td>
</tr>
<tr>
<td></td>
<td>Risk in perspective (Greater chance of healthy children)</td>
</tr>
<tr>
<td>Other</td>
<td>Reassurance</td>
</tr>
<tr>
<td></td>
<td>Normalise the whole process</td>
</tr>
<tr>
<td></td>
<td>Clarify that there are many myths</td>
</tr>
<tr>
<td></td>
<td>Correct any misinformation given</td>
</tr>
</tbody>
</table>

6.3.2. Discussion group

6.3.2.1. Warm up exercises

As part of a warm up exercise, participants were asked to write down a brief description of themselves and their cultural background or alternately to give 10 answers to the question Who am I? Only two of the participants referred to socio-economic status (well-educated and middle class) when describing their own cultural background (Table 6.5) whereas this seemed to be important in descriptions of their clients’ cultural background in the questionnaire responses (Table 6.2).
Participants were asked to write down the first ideas that came into their minds in response to the term consanguinity and the responses are listed below in Table 6.6. Each participant noted at least one item but some listed multiple items.

Table 6.6  Genetic counsellors’ first thoughts about consanguinity

<table>
<thead>
<tr>
<th>First thoughts</th>
<th>No of respondents (n=)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Related marriage</td>
<td>1</td>
</tr>
<tr>
<td>Ashkenazi Jewish / first cousins</td>
<td>1</td>
</tr>
<tr>
<td>Religious / customary</td>
<td>2</td>
</tr>
<tr>
<td>Isolated / rural areas</td>
<td>1</td>
</tr>
<tr>
<td>Mixed blood (closely)</td>
<td>1</td>
</tr>
<tr>
<td>Cousin marriage</td>
<td>1</td>
</tr>
<tr>
<td>Increased genetic risk but less than most expect</td>
<td>1</td>
</tr>
</tbody>
</table>

6.3.2.2. Categories and themes identified from the discussion group

The questions put forward to guide and prompt discussion focussed on what limits genetic counsellors’ ability to manage cultural diversity, the resources needed to do this more effectively, and future research in this area. The discussion group transcript was reviewed using thematic analysis, enabling topics and responses to
be organised into categories and themes. Four categories and 10 themes were identified (Table 6.7).

Table 6.7  Categories and themes identified from the discussion group

<table>
<thead>
<tr>
<th>Category</th>
<th>Theme</th>
</tr>
</thead>
<tbody>
<tr>
<td>Definitions of cultural diversity</td>
<td>Points of cultural difference</td>
</tr>
<tr>
<td></td>
<td>Assumptions and stereotypes</td>
</tr>
<tr>
<td>Professional challenges in counselling clients from culturally different backgrounds</td>
<td>Patient autonomy</td>
</tr>
<tr>
<td></td>
<td>Paternalism in the medical model</td>
</tr>
<tr>
<td></td>
<td>Informed consent</td>
</tr>
<tr>
<td></td>
<td>Disclosure of personal information to clients – is it being directive?</td>
</tr>
<tr>
<td>Experiences of cultural difference in clinical practice</td>
<td>Challenges arising from cultural diversity</td>
</tr>
<tr>
<td></td>
<td>Lack of confidence in managing diversity</td>
</tr>
<tr>
<td>Education and training needs</td>
<td>Learning opportunities</td>
</tr>
<tr>
<td></td>
<td>Improved resources for managing cultural difference</td>
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**Definitions of cultural diversity**

The first discussion point focussed on conceptions of cultural difference. In particular, it examined the impact of counsellors’ own cultural background on interactions with clients.

**Points of cultural difference**

The participants described themselves and their cultural background using a broad range of descriptors, but when focussing on their experiences working with culturally different clients, they tended to refer to clients of a particular ethnic background or religion:

I found that to be able to work better with them, to find out more about the religion, and what it means to them …. but sometimes it can be challenging.
People’s religious beliefs, have a huge impact on how they incorporate the genetic information into their lives and how they deal with things. [GC2]

One participant described how socio-economic factors can alter the client’s agenda and necessitate a shift in the genetic counselling to a more client centred approach:

With the issue of different socio-economic levels, I think one of the things that I’ve leaned over my experience is that it’s important to identify what is important for them. Because we impose what we believe is important on them. [GC1]

Whether disability, or being a parent of a child with a disability, placed clients in a different cultural group was also raised. One participant, who was the mother of a disabled child, noted that prior to this point she had not considered herself as culturally different from others without disabled children:

I see parents of children with disability as being a specific culture of their own in many ways because of the distinctiveness it places on them within the community, the schooling systems and the health system ....I became a parent of a child with an intellectual disability and I think that put me into quite a different culture. Although it’s not something until I was sitting here listening to this that I actually have probably recognised. [GC3]

One participant worked in a community represented by multiple ethnic groups. This participant indicated that they felt socio-economic factors added a layer of complexity to the issue of ethnicity and race:

I think even though we can see them as a race in themselves, sometimes, like it does pay a role in the sense of like, education level and how much they know about scientific stuff and the awareness of disability so it’s not just cultural in the whole. Even though they may practice the same religion but every individual family works differently. [GC6]

Assumptions and stereotypes

Several participants talked about the dangers of categorising individuals into specific groups based on cultural identity or religion, and making assumptions
according to those categories. These participants recognised that categorisation can lead to stereotypical assumptions.

Quite often people will identify straight up I’m a Buddhist or I’m a Catholic or whatever. And the first reaction is that, you make some assumptions on the basis of that claim. And that’s a very dangerous thing because we all know that people can claim allegiance to religion but not actually believe in the letter of the law. [GC7]

**Professional challenges in counselling clients from culturally different backgrounds**

With regard to counselling culturally diverse clients, the main professional challenges identified by the participants were related to the dilemma of maintaining accepted principles of genetic counselling. These included the tensions between achieving ND while facilitating patient autonomy, paternalism, informed consent, and disclosure of personal information.

**Patient autonomy**

Participants indicated they found counselling challenging when clients were from culturally different backgrounds and the clients’ autonomous decision making values varied from their own values. More than one participant noted that they were internally conflicted when working with couples and families who placed less value on individual autonomy, particularly when women left decision making to their male partners. One prenatal genetic counsellor stated:

The real dilemma for me is when you’re seeing couples and it doesn’t necessarily need to be couples that come from certain cultural backgrounds, but you see couples together and the way that they function within their relationship is very much that the male tends to make the important decisions and the woman appears to be okay with that happening and that’s the way that they function as a couple. And because I’m coming off a background, you know, making autonomous decisions I sometimes wonder whether I’m actually causing harm by trying to encourage somebody to make their autonomous decision which is not actually the way they choose to live their life. And I only have a short time to do it and I don’t want to rock the boat. [GC4]
Another with a self-declared strong feminist background, indicated:

I immediately think this woman needs to be more empowered, and I think that’s one of the things I had to learn to do, was to back off and accept that part of feminism is allowing people to make choices about how they live their life in terms of dynamics of relationships, so you know, it’s just as bad for me to say, every woman needs to be empowered and make their own decisions as it is for someone to say no woman should be able to make her own decisions, I think a very big issue as well. [GC1]

Paternalism in the medical model

Participants raised the broad issue of the constraints imposed by working within the medical model, the complexity of genetic information, and the struggle to balance informed consent with the institutional barriers imposed by the medical model.

We’ve worked very hard in the past two decades to drop the paternalistic approach. You know, the interesting thing is I have my suspicions we will return to that, simply because the complexity of medicine will dictate that. We will no longer be able to discuss every nuance, every option, you know, the science behind the options as we progress. So I guess my suspicion is we will return to a paternalistic form of health care delivery. [GC1]

One participant expressed the idea that there may be a tendency for health professionals to make assumptions and apply their own opinions particularly in relation to consanguinity:

You know there’s this... I guess there’s this incorrect appreciation in the medical system that consanguinity is always bad. [GC1]

Informed consent

The challenge of obtaining informed consent was raised by several participants. The difficulty of defining, and determining if, informed consent was obtained and balancing this with the information requirements of the clients was highlighted as a particularly challenging professional issue:
Even though we do have those sort of medico-legal demands that they are informed, I think you have to at some point, say well, what is informed consent? I think the jury’s completely out on what that is and I think in realistic terms it’s not even actually attainable, so you actually have to say well what matters for you in this situation? What will be the information that you think you need to make your decision? And I agree with you, I think that’s hard. [GC1]

The participants were confident that genetic counsellors were able to recognise the benefits of tailoring information to the individual and their beliefs. They also noted the importance of determining the needs of the client upfront rather than imposing their own goals. Without taking into account the needs of the individual, informed consent may not always be obtained.

Sometimes you’ll have a couple who’ll nod and look and you know and you just know you’re going nowhere, so sometimes you can stop and say ‘look, I’m trying to give you an awful lot of information. What is it actually that you want to take away from this discussion?’ … sometimes their goals are so much more modest than ours. And I think in a lot of ways, it’s once again, it’s that autonomy issue. You have to respect that. [GC1]

*Disclosure of personal information to clients – is it being directive?*

One participant was concerned about the negative impact of personal disclosure to her clients, as this could result in projection of her own assumptions based on her personal experience of raising a child with a disability:

> It has made me be very conscious not to place my own experience in front of people. You know I’m very careful not to self-disclose unless it’s appropriate. I’m careful not to assume that everybody will feel the same way. [GC3]

In contrast, other participants felt that disclosure might be beneficial in some circumstances. Patient autonomy was also discussed with regard to maintaining ND. Participants noted that they sometimes felt conflicted when being nondirective and felt personal disclosure may benefit the client. One participant indicated that raising and discussing personal and professional experiences might be useful for clients:
We are giving them a great disservice by not giving them the benefit of our experience. And I don’t know why we’re so scared about doing that. I think we all would know if we were being coercive and I think we need to get rid of this idea of being either directive or nondirective and just think about being useful to our clients. And maybe be up-front about that with them and discuss that with them.

[GC4]

Experiences of cultural difference in clinical practice

In response to questions about difficult cases and issues related to cultural diversity, participants described many challenges in counselling clients from minority groups. There was also a lack of confidence with regard to managing diversity.

Challenges experienced by clients in minority groups

One participant suggested that clients from cultural minorities faced a number of unique challenges in relation to accessing genetic counselling and other services:

When you’re in a culture that is a different sort of culture it’s not a, ethnic culture, it’s a social culture and a culture that people are thrust into, and have to learn how to cope within that culture, and learn right the way through and deal with discrimination, deal with difficulties, have to fight for everything that comes their way, you know health, education, to employment. [GC3]

Another participant who worked in a very multi-ethnic society expressed the difficulties she experienced in trying to provide a service to individuals from several minority ethnic groups:

I think it is important that you know the cultural background of certain race or ethnicity but for me what I find challenging as a [ethnicity], what I find challenging working in a public setting as well, that I see [ethnicity, ethnicity and ethnicity] patients and parents and I do find that the problem of language barriers as well as different perceptions of disability and consanguinity. [GC6]

Lack of confidence in managing diversity

Several participants highlighted a lack of training and confidence in negotiating cultural diversity. The participants commented on their lack of training in relevant
areas (such as couple dynamics) and how cultural differences might impact on decision making and informed consent.

It’s [cultural difference] a very difficult thing to assess. Is this a mutually acceptable dynamic, or is this a case of male dominance, you know, over a female? And, you know, I think as genetic counsellors we’re not really trained to determine those differences but you need to be aware of them ... I think it’s a very big issue and I think it’s probably one that we don’t cover enough in genetic counselling training.

[GC1]

Another issue raised in the context of dealing with consanguinity was negotiating the impact of the norms and values of the society and the system within which you operate.

Working in a rural area a lot of the consanguinity that comes through is usually not an accepted cultural norm and it’s something that has occurred by chance in many cases. People find out the information after they’ve become close and someone in the family will say, ‘Oh did you know that you’re related?’ And they come with a great deal of shame to genetic counselling because of this and fear that they’re breaking the law. And I think that there is a not a cultural norm in the white Anglo-Saxon Australian community of marrying within your family and it is seen as in some ways distasteful. And the people that do come through with me, and even from their referral sources, it’s often like, a bit of a snigger and a giggle. And you know, and people make comments like ‘Oh, you know, there must be lots of work for you up in the back woods of you know wherever’ and ‘Oh, you know, that’s six finger country’ and that kind of thing comes across a lot. And so when people come for genetic counselling because of this they come, sort of, very quietly about it.

[GC3]

Participants also described themselves as cultural interpreters who used their skills to bridge the divide between the client and the perceived ‘norms’ of society:

You often feel that when you ask that question about consanguinity, there’s this immediate sort of partial wall that goes up and, you know, are you going to be judgmental about this? So I think I’ve worked fairly hard in my career to sort of say, it is okay, you know, people always think it’s worse than it is. ... I find that acknowledging that I think it’s okay and not in those sorts of words, but just in my
reactions I suppose and the way I deal with it, I find that really relaxes people and they’re much happier to talk about the consanguinity within the family and, you know, what it means to them in terms of the benefits or otherwise. So, that’s actually been quite good. [GC1]

**Education and training needs**

As the discussion reached a conclusion, participants’ attention turned to the future and they were asked to consider ways that the profession of genetic counselling could move forward in the endeavour to enhance skills in cultural diversity. In addition to the identified needs for cultural diversity training, participants mentioned the need for improved resources and innovative ways for genetic counsellors to learn about cultural diversity.

**Improved resources for managing cultural difference**

Participants expressed a need for the development of resources to enable better management of cultural difference.

I have heard a lot of the talks where we all seem to be doing the same thing, development of guidelines for using interpreters, translation aids and all those sorts of things and I just keep thinking why are we all trying to do the same thing in our own little units? You know, maybe there’s some way that we can, foster interstate and international collaborations and things like that. [GC5]

**Learning opportunities**

The participants indicated the discussion group was an opportunity to meet with colleagues, discuss professional issues and to exchange experiences to improve skill sets and confidence. Many viewed this as a learning opportunity and indicated discussion groups and participation in other research activities could also be used in future as a training tool.

I think this sort of forum is really useful ... if we could encourage more of these with better participation I think it’s a really good learning experience. [GC1]
Another participant suggested the development of exchange programs to encourage genetic counsellors to travel to other genetics units, both interstate and overseas. To this participant, these exchanges would be a useful means to broaden horizons of genetic counsellors, allowing them to observe how other units manage clinical practice and research.

6.3.2.3. Ideas for further research

The discussion group participants proposed some ideas for future research to address some of the challenges that they experienced in dealing with cultural diversity in clinical practice, including:

- Examining the benefits of exchange programs and interstate/international collaborations.
- Facilitating further focus group discussions to explore the question of whether there is a benefit of these for research and for training purposes.
- Encouraging the ASGC and Board of Censors to promote research participation as a learning experience, given that changes to requirements for certification in Australia now encompass reflective practice and research.
- Developing a forum for sharing information on research being undertaken.

6.4. Discussion

The questionnaire and discussion group results presented in this chapter yield insights into the extent to which genetic counsellors engage with diversity and how they implement this in practice. The results suggested a tendency to maintain a kind of professional detachment through nondirective information giving even when the counsellor identified differences that may be due to a different cultural perspective.
6.4.1. Demographic details of Australasian genetic counsellors

The demographic data obtained are comparable to the make-up of the profession in the United States (Lega et al., 2005). As little is known about the characteristics of Australasian genetic counsellors, these data provide a valuable insight and contribution to the literature.

6.4.2. Multiculturalism and cultural difference

At the time of the survey, genetic counsellors identified the stereotypical determinants of cultural difference in their clients with only one respondent mentioning broader dimensions such as socio-economic factors. Few counsellors identified the many other sources of cultural difference between themselves and their clients, and there was little or no mention of the influence of their own cultural determinants on interactions with clients. Participants in the discussion group did recognise that socio-economic factors contributed to their own cultural background. Identifying one’s own cultural determinants is the first step in recognising these differences in others.

More recently published research suggest that clients and genetic counsellors are increasingly aware of the need to think of cultural background in broader terms, to avoid stereotyping and to be aware of how their own cultural background can affect practice (Saleh et al., 2009). As a result of the inclusion of cultural competence in training programs, counsellors have an increased awareness of the impact of our own cultural background on our interaction with our clients. Saleh et al. (2009) conducted seven focus groups, involving 53 participants in two Australian states, to investigate the challenges facing genetic counsellors dealing with diversity. They found that participants viewed cultural diversity more broadly than simply country of origin, ethnicity and language, and included factors in their definitions such as geographical location, lifestyle factors, general literacy and health literacy levels.
Despite the increasing awareness of cultural diversity issues, genetic counsellors in this study continued to focus on areas such as language and information provision for clients who have English as a second language. Remembering that clients may lack basic reading ability is vital as only 50% of the Australian population (aged 15-74) have adequate literacy skills and 41% of adults have adequate health literacy skills (Australian Bureau of Statistics, 2008a).

Patient centredness is a concept that recognises the patient as a unique individual. Tailoring communication, access and information to the individual can enhance client satisfaction (Gale et al., 2010; Saha et al., 2008). Relational ethics also treats each client as an individual human being. Evans et al. (2004) have highlighted the principle of focusing on the relationship between counsellor and client, and recognising that engagement, dialogue and a sense of being present and respected as a whole person enhances information processing:

Valuable time is spent in ‘getting to know’ the client as a person with unique knowledge and understanding. Yet, to move beyond a level of particular information exchange between the counselor and client, it is necessary to find ‘a common ground’, for it is from this common ground that the relationship can progress to one that incorporates respect and trust. Clients interpret the information gained through counseling in the context of both the information provided to them and their beliefs, values and personal knowledge about genetics. The counselor in turn interprets and learns from clients and uses this additional or new knowledge to help to build understanding. (Evans et al., 2004 p.467)

6.4.2.1. Cultural diversity training

Participants considered that a co-ordinated approach to cultural competency training and provision of educational resources was a major priority. None of the participants in this study indicated that cultural awareness or competency were part of their ongoing training or professional development, rather cultural awareness had largely been developed through working with interpreters or community leaders, learning from colleagues and through personal life experiences.
Participants in the discussion group felt that the discussion highlighted to them the importance of diversity issues and was a learning experience.

As reviewed in Chapter Two, genetic counselling has progressed from an education process embedded in the medical model to a more inclusive dialogue between individuals, attending to psychosocial issues and incorporating more of Kessler’s counselling model. Weil (2001) has proposed the extension of Kessler’s model to promote an individualised approach that attends to the needs and circumstances of each client and better equips the genetic counsellor to encompass cultural diversity. ND as a model for genetic counselling was also reviewed in Chapter Two and the difficulty of maintaining this model was discussed. It was argued that there was a need for more dialogical approaches that engaged the background cultural factors of both client and counsellor and where a skilled counsellor assists clients address broader cultural issues even when their own values may be very different.

6.4.2.2. Gender diversity

Some of the comments from the participating counsellors relating to gender differences reflect stereotypical gender differences. For example the assertion that males are better for counselling males and females better placed to counsel regarding issues of pregnancy and TOP fails to consider the skills and attributes of each counsellor.

6.4.2.3. How cultural awareness and diversity influences practice

Many participants displayed a lack of flexibility in failing to tailor their genetic counselling sessions to suit individual clients. This may occur more frequently in inexperienced counsellors owing to a lack of awareness, or a lack of skill. A more flexible approach could be achieved by adopting a more client centred approach and identifying issues and information important to individual clients.
Menezes et al. (2010) explored the impact of Australian genetic counsellors’ own pregnancies on their work in the area of prenatal counselling. Applying the qualitative methods used in that study might facilitate training and skill development, as these techniques allow genetic counsellors to explore their own cultural identity. Further research on the genetic counsellors’ work with culturally diverse clients using the same model might also be informative.

6.4.3. Consanguinity – a matter of opinion

Australasian genetic counsellors’ views on consanguinity were explored to examine whether their opinions influenced the way in which they interact with clients. There is a generally negative view of consanguinity in our society. If this negative view was also held by genetic counsellors, it could potentially impact on the way they counsel to consanguineous clients. It is thus worth noting that nine per cent of the counsellors completing the questionnaire listed first cousin marriages in their ranking of respondents’ top four incestuous relationships.

Remaining impartial and non-judgmental may be difficult and unconscious actions, like altered tone or demeanour, might reflect Genetic counsellors’ opinions. It would be concerning if counsellors were unaware of their own beliefs and values, and how this affects their interaction with clients.

Genetic counsellors were asked to specify if consanguinity was more common in any particular ethnic group. It is not possible to determine if the responses given relate to a lack of knowledge about the distinction between ethnicity race and religion. The failure to attend to emotional aspects of consanguineous relationships could be because the respondents assumed that the questions referred to particular ethnic groups where consanguinity is common.
6.4.3.1. Advice and information for consanguineous couples

The tension surrounding ND was highlighted by respondents’ comments about whether or not consanguineous marriage should be encouraged, with many responding to the intentionally provocative question with comments such as “it is not my place to advise”. The nondirective pathway would be to ignore differences, adopt a neutral stance and focus on the more narrowly scientific issues. An alternative approach would be to encourage dialogue about the broader personal, family and cultural issues raised by consanguineous relationships and to engage in nondirective dialogue with the client about differences in value or world view questions, perhaps even reflecting the counsellor’s own values and experience.

6.4.3.2. Genetic counsellors’ views on consanguinity

A low number of genetic counsellors accurately identified the risk of birth defects for the general population and consanguineous couples, possibly reflecting the lack of accurate risk estimation figures in the literature (Bennett et al., 2002).

As members of a caring profession, genetic counsellors are generally assumed to be accepting, non-judgmental and nondirective health professionals. The thesis research discussed in this chapter aimed to identify how aware they are of any differences and how this might impact on their practice. Respondents may have been compelled to give the perceived socially acceptable answers to questions about their opinions on consanguinity, reflecting these professional expectations. A perceived lack of comfort and confidence in counsellors’ own responses was raised during the discussion group and proposed as a reason for the lack of participation in the questionnaire and discussion group. The questions about consanguinity were attempting to get to the crux of Western society’s ideas and why many find it a strange or even repulsive practice. My impression is that there is a fine line drawn between consanguinity and incest, and the aversion is related to the latter. It may have been more effective to use probing questions about genetic counsellors’ own
life and potential relationships with cousins, or about where they draw the line on consanguineous relationships.

6.4.4. Discussion group outcomes

The discussion group gave an insight into the difficulties genetic counsellors face in addressing cultural difference in their work and how they manage these difficulties. The small number of participants allowed the discussion to broaden into a more philosophical dialogue about the profession.

Despite the greater awareness of cultural diversity and an apparent desire to engage more with the issues, there appears to be a reluctance to move towards the more dialogical models of practice. Intellectually genetic counsellors may respect cultural difference but the data shows internal conflict and continued stereotypical definitions of culture. Tensions exist between ND and the ability to adopt a more dialogical model that would allow exploration of individual differences. On a positive note, there is affirmation of the need to engage positively with diversity.

Continued exploration of cultural engagement and genetic counsellor training is important, in particular continued education on broader definitions and the impact of cultural diversity on counsellors and their practice. Counsellors need to be confident in their ability to be adaptable, in their practice so that when they do meet cultural differences they explore the client’s unique experience and are able to respond in a way that best serves the interests of the client.

Participants in the discussion group considered the discussion more like professional development than participation in research. They suggested that further discussion groups would be valuable training and professional development tools. This view was also echoed by Australian genetic counsellors who participated in the (Saleh et al., 2009) study, who felt that the group discussion was a valuable tool for professional development and creating awareness of cultural issues. They
described the focus group discussion as one of the few opportunities they had had to engage with these issues.

6.4.5. **Study strengths and limitations**

This was the first Australasian study examining genetic counsellors’ perceptions of cultural diversity, particularly in relation to consanguinity. To the best of my knowledge there is one report of a focus group with Australian genetic counsellors about their perceptions of cultural difference (Saleh et al., 2009). The data obtained here provides valuable information about genetic counsellors’ demographics and perceptions of cultural difference and consanguinity. The education and training needs of a growing profession in increasingly complex times are also highlighted.

Because of the moderate response rate to the questionnaire, the opinions or experiences of respondents cannot be generalised. The utilisation of mailed questionnaires as a means of gathering data lends itself to a number of biases. As the respondents may represent a narrow view, there could be recruitment bias. Anonymously completing a questionnaire removes the opportunity to clarify questions and read body language, leaving respondents the opportunity to provide socially desirable responses (Gu et al., 2011). An interview format might have led to the collection of more comprehensive data and allow greater opportunity for detailed descriptions of genetic counsellors’ opinions and work practices.

Responses are dependent on the reader’s interpretations of the questions. A more extensive study may have resulted in a more consistent interpretation of the questions. As only demographic data about background such as profession, ethnicity religion and gender were requested, the questionnaire did not take into account each counsellor’s life experiences and other influential factors that they bring to sessions with clients. On reflection, a question exploring participants’ thoughts about their own cultural background and recording the terms they respond with may have given richer data. It is unlikely in this setting that
participants would mention sources of difference regarding private issues, such as sexual orientation.

Certain responses to the survey instrument prompted me to speculate about the impact of a genetic counsellor conducting research on genetic counsellors. Perhaps knowing that the questionnaire, albeit de-identified and confidential, came from a colleague affected the ability and comfort levels of participants to give ‘truthful answers’. Accessing thoughts and ideas that may be controversial or uncomfortable to express requires building up a level of trust and may be better achieved by someone from outside the profession. On the other hand, genetic counsellors may feel less threatened and open to criticism if working with a trusted colleague and therefore more likely to be open. Research has shown that being an outsider or different from the participant in some way allows the possibility of more in-depth examination or eliciting different responses, although there are some situations where being an outsider can hinder the research as participants can be reluctant to talk openly if they fear recrimination (Tinker et al., 2008).

The discussion group represented a very small sample of Australasian genetic counsellors and the purpose was to have an informal discussion, so the data obtained cannot be generalised. Formal focus group research would have necessitated different research methodology and data analysis.

6.4.6. Recommendations for increased engagement with diversity

The following recommendations are proposed as a means of increasing awareness of, and engagement with, diversity issues in genetic counselling practice. They can be implemented during training or professional development.

- Recognise that all clients are individuals and have different needs.
- Learn from the client by listening to their story.
- Assess each client’s concerns, taking time to explore their prior knowledge and beliefs about genetics, genetic conditions and genetic counselling.
- Avoid making cultural assumptions or categorising people into groups.
- Ask for reflective feedback to check clients’ understanding.
- Analyse one’s own cultural background and beliefs and their influence on practice.
- Focus counsellor training on developing reflective skills around the importance of personal views and cultural background in the counselling interaction.

6.4.7. Summary

The research described in this chapter brings new ideas and new applications of qualitative research methods to the profession. It offers an insight into the way Australasian genetic counsellors view cultural difference and consanguinity, and highlights some areas where greater training and discussion of these topics enable them to engage more effectively in increasingly diverse and complex times.

Diversity is a fact of life and indeed it makes our life rich and interesting. In genetic counsellor training and professional development, diversity needs to be normalised so that we can be comfortable with different ideas and practices, even if we hold alternative views about them. The major goal of genetic counselling is patient autonomy counsellors’ opinions must not impact on patients’ right to make autonomous and informed decisions. It is how we engage with different views and in our interaction with clients that is important.

We have progressed from the narrow definition of cultural difference and recognise the broader range of factors that contribute to diversity. Now there is a need to move on from defining people by their culture at all and deal with each client as a unique individual. It is important to explore the factors that influence a person’s thoughts and values, without defining or stereotyping them. The next challenge is how to relate to individual differences in an engaged and dialogical way.

In Chapter Seven I will explore if and how GHPs engage with cultural diversity in actual genetic counselling sessions, and the extent to which we are currently applying the models of genetic counselling that embrace ethnocultural diversity.
7. **Interactions between GHPs and clients**

The genetic counselor and patient participate in an educational exchange of genetic and biomedical information shaped by their unique psychosocial identities. The genetic counselor-patient relationship is the medium in which these activities occur. (McCarthy Veach *et al.*, 2007, p.726)
7.1. Introduction

Previous chapters have explored models of genetic counselling (Chapter Two), experiences of consanguineous clients with genetic counselling (Chapter Five) and how genetic counsellors deal with clients from culturally diverse backgrounds, particularly those in consanguineous relationships (Chapter Six). This chapter focuses on interactions between GHPs and their clients in clinical practice in WA. This preliminary exploration of clinical genetic counselling consultations examines how genetic counsellors and clients interact, the factors that influenced these interactions, and models of genetic counselling deployed.

7.1.1. Inside the ‘black box’ of genetic counselling

Genetic counselling was once regarded as a one-way communication process with counsellors imparting expert knowledge of genetics and medicine to less informed clients (Wolff et al., 1995). In 2002, Abrams and Kessler noted that the interaction between genetic counsellors and clients was more complex than this and much more than a one-way transmission of information:

Thought of as a process of communication, misses an essential point: genetic counseling is an interaction between human beings. It is more than a process, more than communication. It addresses the nitty-gritty of human dreams and wishes and hopes and the quotidian existence of joy and pain and – for better or worse – it’s based on a relationship between real thinking and feeling people. (Abrams et al., 2002, p.16)

Despite genetic counselling now being recognised as a complex interaction, until recently little research has been directed towards examining what happens in a genetic counselling session and how genetic counsellors communicate with their clients. Since early 2000 there have been a number of published qualitative studies of the interactions that take place during genetic counselling sessions and Meiser et al. (2008) reviewed 18 studies that used communication analysis of audio- or video-
taped genetic counselling sessions. These qualitative approaches have been found to be very useful when persons of differing cultures come together (Lewis, 2002).

As many qualitative studies of the communication process in genetic counselling are limited to the perspective of the counsellor, studies that take into account the two-way process of communication are needed. There is evidence to suggest that the changing goals of genetic counselling and the broader models of practice encompassing information provision, psychosocial and decision making support are not being fully implemented (Ellington et al., 2011). While communicating information is important, research should be directed towards understanding the extent to which the session is improved by addressing cultural and psychosocial issues. Meiser et al. (2008) has called for further studies exploring the impact of client socio-demographic status on providers’ communication behaviours.

In addition to a lack of knowledge about socio-demographic factors and genetic counselling, there is also little information about diversity issues and their impact on the client. An understanding of the perspective of the client is needed in order to have an understanding of their cultural needs and how those needs can be met. Michie and colleagues (1997) measured how directive the communication between counsellor and client was and found that counsellors were more directive with clients who they perceived as more concerned or of low socio-economic status. These counsellors adapted their counselling style but did so in response to their own perceptions of the clients’ concerns. A study of providers in familial cancer clinics also showed that counsellors change their communication technique based on clients’ clinical factors and socio-demographic variables, rather than distress or enquiring about patient’s expectations (Lobb et al., 2002). Further research would be useful in determining whether particular aspects of a client’s background would assist with identifying their counselling needs. The mismatch between assessments of understanding and needs could be extended to include cultural factors.
Eunpu (1997) states that despite the time constraints present in busy genetic clinics taking time to explore the clients’ narrative is critical. Each interaction should be approached as a previously unheard story, involving actively listening, assessing the background factors (including cultural issues) influencing that particular client, engaging with the client on a level determined by them, and not assuming what they should know or want to know (Punales-Morejon et al., 1992). Focusing on the client and their needs allows counsellors to spend more time exploring a client’s cultural background. The client also has something to give, be that information about themselves and their own thought process, the way they assimilate information, or details about their cultural identity (Sue et al., 1990; Wang, 2001).

In genetic counselling sessions, psychological and emotional issues affect the way a client can assimilate genetic information and make it personally relevant based on their values and beliefs (Ellington et al., 2011). If a genetic counsellor is focussed on education and information provision, they may be less likely to pay attention to emotional issues and cultural factors that might influence the client’s ability to understand and retain information. For example, failing to assess a client’s educational background, cultural beliefs about inheritance, and health literacy levels might result in the counsellor giving complex information about genetics to a person who is neither equipped, nor willing, to listen and understand. The assessment could be made either in advance of the session if possible or at the beginning of the session so that it can be tailored according to the client’s needs (Evans et al., 2004).

Whilst health literacy is a relatively well-studied area in the area of medical dialogue, it is a new topic of research for genetic counselling (Clement et al., 2009; Sudore et al., 2009). Health literacy is defined as the patient’s ability to use their literacy and numeracy skills to obtain and process health information, and is an important factor in a client’s ability to communicate and participate in the genetic counselling interaction (Lea et al., 2010). Roter and colleagues (2009) used the
Genetic Counseling Video Project to develop frameworks to assess the oral literacy demands of genetic counselling and discourse analysis to examine the relationship between a client’s health literacy and the efficacy of medical communication (Roter et al., 2009; Roter et al., 2008). They found that assessing the literacy levels of a client is a difficult skill for genetic counsellors and physicians to master.

Several studies have analysed relatively large numbers of recorded genetic counselling sessions to examine communication patterns and styles in the interaction between counsellors and clients (Babul-Hirji et al., 2010; Ellington et al., 2006; Fransen et al., 2006; Roter et al., 2008). The studies show that a broad model of practice that encompasses information, support and assistance with decision making is not being fully implemented in counselling sessions, with emphasis remaining on biomedical rather than psychosocial communication. Babul-Hirji et al. (2010, p.44) note that when a genetic counsellor’s “discourse emerges primarily as the voice of efficiency, education and scientific information”, the “human voice of the patient” is suppressed, yet when the ‘human voice’ of the counsellor comes across by attempts to build rapport, patients can participate in the dialogue and voice their feelings.

### 7.1.2. Aims

The purpose of this stage of the research was to conduct a preliminary, exploratory study of the process of genetic counselling by observing genetic counselling interactions and exploring how this counselling is experienced by both the GHP and the clients. Recorded and transcribed genetic counselling sessions and post-session questionnaires completed by both counsellor and client were designed to explore whether counsellor style, or cultural differences between client and counsellor, influenced the content of, and satisfaction with, the session.

This window into the complex activity of genetic counselling also provided an opportunity to explore the extent to which, in the context of a genetic counselling
session, GHPs are attuned to cultural difference. This study examined whether social and attitudinal issues were recognised in the sessions and, if so, whether the GHPs adapted their style in response. Of particular interest was whether in recognising and adapting to diversity, GHPs applied the more dialogical models of genetic counselling that have recently been developed. A close examination of the discursive strategies employed by counsellors could potentially identify the kinds of skills and resources that counsellors need in order to negotiate cultural factors.

### 7.2. Methods

The methods are outlined in detail in Chapter Four. In summary, live genetic counselling consultations were recorded and transcribed.Transcripts of the live genetic counselling sessions were reviewed, looking for sequences of the interaction important to the thematic focus of this study: negotiating cultural difference. After the completion of the clinic appointment, clients and GHPs completed a post-session questionnaire.

### 7.3. Results

#### 7.3.1. Description of participants

The GHPs in the recorded genetic counselling sessions were a male geneticist and two female genetic counsellors. They were born in Australia and aged in their late 20s to early 30s. The counsellees were a young professional woman seeking carrier testing, a couple of childbearing age wanting information about maternal age risks, and an elderly retired woman undergoing pre-symptomatic testing.

#### 7.3.2. Recorded genetic clinic sessions

The genetic counselling consultations covered a range of topics and demonstrated the general conversational style of these sessions. The audio-recordings of the clinic sessions were transcribed using standard transcription conventions (Table 4.3) and pseudonyms were introduced at this stage to ensure confidentiality.
Three genetic counselling sessions with full transcripts available are presented as a preliminary exploration of the process of genetic counselling. The length of sessions, people present in the consultation, the number of words spoken and the proportion spoken by the counsellor and client are shown in Table 7.1.

Table 7.1  Characteristics of recorded genetic counselling sessions

<table>
<thead>
<tr>
<th>Session</th>
<th>Session length (min)</th>
<th>Total words</th>
<th>Counsellor spoken words</th>
<th>Client spoken words</th>
<th>Persons present</th>
</tr>
</thead>
<tbody>
<tr>
<td>5</td>
<td>35</td>
<td>6110</td>
<td>4454 (73%)</td>
<td>1656 (27%)</td>
<td>2</td>
</tr>
<tr>
<td>6</td>
<td>61</td>
<td>7600</td>
<td>5830 (67%)</td>
<td>1770 (23%)</td>
<td>3</td>
</tr>
<tr>
<td>7</td>
<td>43</td>
<td>7630</td>
<td>5694 (75%)</td>
<td>1936 (25%)</td>
<td>2</td>
</tr>
</tbody>
</table>

7.3.2.1.  Negotiating cultural differences

The extracts presented here are taken from moments in the genetic counselling sessions that demonstrate attempts by the GHPs and clients to negotiate cultural differences and enhance communication.

Session Five

The client Joan was a 60 year old retired office worker requesting pre-symptomatic genetic testing for the BRCA1 mutation that has been identified in her family. At the time this consultation took place, the process for this type of testing comprised a series of three sessions, the first completed by a genetic counsellor, the second conducted by a clinical geneticist and the third session reserved for result delivery by both GHPs. The following extracts are from the second session of the process, which has the primary aim of ensuring the client has made an informed decision about proceeding with testing. The geneticist (DR1) is male and of a similar age to Joan’s children. At the completion of the second session, Joan signed a consent form and had blood drawn for genetic testing.

In extract 1 the geneticist begins the session with a statement (line 1) suggesting that they are mutually setting the agenda and ensuring that both he and Joan are
“on the same page”. Throughout the exchange Joan has limited opportunity to express her agenda and is frequently drawn back to the geneticist’s agenda (line 7).

Session 5 Extract 1

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>DR1</td>
<td>So just to make sure we are on the same page. You in your family there has been a BRCA1 mutation identified and as part of that you are here to find out, to talk about issues related to genetic testing. You have already had a session with the GC about that and pending those discussions perhaps then to go on to do undertake testing to see whether or not you have that mutation known to occur in your family.</td>
</tr>
<tr>
<td>2</td>
<td>Joan</td>
<td>Right, mmm.</td>
</tr>
<tr>
<td>3</td>
<td>DR1</td>
<td>The other bits of information I have got just from you just from reading your notes and speaking to [GC] just to make sure I am correct is that you haven’t had cancer yourself that again from reading [GC’s] notes irrespective of whether if you were to undertake testing irrespective of the results of that testing you are planning to have your ovaries removed. Is that right?</td>
</tr>
<tr>
<td>4</td>
<td>Joan</td>
<td>I am planning I have just had some tests a CA125 tests and they have come up it may well dependant on how well this test goes whether I may or may not [ Interruption]</td>
</tr>
<tr>
<td>5</td>
<td>DR1</td>
<td>Right so it is still not set in stone</td>
</tr>
<tr>
<td>6</td>
<td>Joan</td>
<td>It was set in stone until I got the result and then I thought well maybe leave it a bit longer but that’s something [ Interruption]</td>
</tr>
<tr>
<td>7</td>
<td>DR1</td>
<td>We can talk about the CA125 in particular OK? And looking at again looking at your family tree the earliest diagnosis of both breast and ovarian cancer were about 40 years of age in your family.</td>
</tr>
</tbody>
</table>

The geneticist then proceeds to rapport building and fact finding conversation. He asks Joan an indirect question (line 13) to explore her occupation and possibly education levels. In something as simple as rapport building conversation, there is the potential for misunderstanding. Joan doesn’t quite understand the point of the question, requiring him to reframe the question in a more direct fashion (line 15).

Session 5 Extract 2

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>13</td>
<td>DR1</td>
<td>And what do you do with your days?</td>
</tr>
<tr>
<td>14</td>
<td>Joan</td>
<td>Oh geez what do I do with my days? ... we are retired my husband and I we have been retired now and but we seem to fill it in family things generally just you know just generally fill the day in we just don’t sit down we are just on the move.</td>
</tr>
<tr>
<td>15</td>
<td>DR1</td>
<td>And what were you what was your occupation before you retired?</td>
</tr>
<tr>
<td>16</td>
<td>Joan</td>
<td>My occupation before I retired was administration I was just yeah</td>
</tr>
</tbody>
</table>
office work computers sat at the computer all day basically.

In Joan’s session there are many elements of dialogue that demonstrate the geneticist’s willingness to explore her concerns and address her questions. Despite this, there are several examples that indicate impatience with diversions and haste to re-focus the discussion. In Extract 3 Joan discusses her hormone replacement therapy in detail and expresses her concern about how the symptoms affected her life (line 22). DR1 moves back to taking a medical history (lines 25 and 29) and changes the subject (line 31).

**Session 5 Extract 3**

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>22</td>
<td>Joan</td>
<td>I was on HRT but I sort of stopped that and I actually didn’t go on that for years I because I was home. But when I went back into the workforce that’s when I needed to take them. But I am just going off them again now.</td>
</tr>
<tr>
<td>23</td>
<td>DR1</td>
<td>Right so</td>
</tr>
<tr>
<td>24</td>
<td>Joan</td>
<td>I’m in the process of [Interruption]</td>
</tr>
<tr>
<td>25</td>
<td>DR1</td>
<td>How long were you on HRT for?</td>
</tr>
<tr>
<td>26</td>
<td>Joan</td>
<td>Probably would have been about 5 years.</td>
</tr>
<tr>
<td>27</td>
<td>DR1</td>
<td>So from 55 to 60?</td>
</tr>
<tr>
<td>28</td>
<td>Joan</td>
<td>The doctor wasn’t overly keen on prescribing them to me.</td>
</tr>
<tr>
<td>29</td>
<td>DR1</td>
<td>That was really because you were having symptoms?</td>
</tr>
<tr>
<td>30</td>
<td>Joan</td>
<td>Yeah well yeah I had had them because of that and it improved my work. It was just so hard at work all the time to concentrate losing concentration and all that sort of stuff. So whereas when you are at home it doesn’t really matter.</td>
</tr>
<tr>
<td>31</td>
<td>DR1</td>
<td>Yeah. And aside from have you ever been in hospital or had any surgery?</td>
</tr>
</tbody>
</table>

In Extract 4 the geneticist explains the need to complete a consent form prior to the genetic testing proceeding (line 59). Joan explains that in her previous session she had sufficient explanation from the genetic counsellor about the genes involved (line 60). Despite this, and saying he won’t go into detail, DR1 gives a lengthy, albeit clear, explanation of tumour suppressor gene function (line 61). Repeating information can be beneficial in some situations but can lead to frustration if the client feels her statement was ignored. Her responses at lines 62, 64 and 66 suggest impatience and a possible lack of interest and engagement with the topic.
Session 5 Extract 4

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>59</td>
<td>DR1</td>
<td>So I imagine we’ll talk a lot about what [GC] talked about previously. But we might raise some new things along the way. And what we’ll do again there is a standard form that we go through for anyone whose having testing for genes associated with cancer. And we’ll go through this form together and we’ll fill it out as we go along and if at the end you don’t want to proceed we just rip it up and throw it away it will just save us going through the form twice [5]. Right so [cough] how’s your biology?</td>
</tr>
<tr>
<td>60</td>
<td>Joan</td>
<td>Up the creek [laugh]. No good at all. Actually GC was very good she explained the genes and that to me how they worked and so that is really great.</td>
</tr>
<tr>
<td>61</td>
<td>DR1</td>
<td>I won’t go into that too much again then other than saying we’ve all got genes in all our cells that are important in how our cells grow. And some of those genes are don’t grow genes or tumour suppressor genes of which the BRCA genes are some of those. So we all need cells that control genes that control cell growth so if I cut my skin for instance I need genes inside my skin cells to turn on and say grow skin cells grow. Similarly I need different genes to turn on in the same cells to then say stop growing. Otherwise the cell will just continue to grow and that’s what a cancer is. So then BRCA genes are really don’t grow genes or tumour suppressor genes and all of our genes come in pairs. So this is a person and we’ll say that this person has inherited a gene fault a mutation in one of those pairs of genes.</td>
</tr>
<tr>
<td>62</td>
<td>Joan</td>
<td>Mmmm yep.</td>
</tr>
<tr>
<td>63</td>
<td>DR1</td>
<td>But that’s ok because they’ve got a back up copy.</td>
</tr>
<tr>
<td>64</td>
<td>Joan</td>
<td>Yep yep yep</td>
</tr>
<tr>
<td>65</td>
<td>DR1</td>
<td>[4.0] Over time for various reasons you might get a change in the other gene but that’s actually not enough to stop it working [pause] might get another change still not enough to stop it working and so forth and so forth.</td>
</tr>
<tr>
<td>66</td>
<td>Joan</td>
<td>Yeah.</td>
</tr>
<tr>
<td>67</td>
<td>DR1</td>
<td>[3.0] Before a number of changes are accumulated and both copies don’t work but it’s at that point where you get two copies not working in the same cell that cancer can develop.</td>
</tr>
<tr>
<td>68</td>
<td>Joan</td>
<td>So that’s when the tumours develop.</td>
</tr>
</tbody>
</table>

Session Six

Session Six was attended by Paula and her husband Ron, and a female genetic counsellor (GC1). The couple presented for information about maternal age risks, as Paula wanted another pregnancy. They had previously had two children and two miscarriages. GC1 had spoken to Paula by phone prior to the clinic appointment but had not had contact with Ron.
At the beginning of the session, Ron appeared reticent to engage and somewhat hostile, questioning the need for their attendance at the clinic. GC1 began the session by setting the agenda and attempting to build rapport with Ron and then gave the couple some information about maternal age risks. The genetic counsellor assessed the couple’s level of knowledge and then adjusted the manner in which information was presented according to their level of understanding. Extract 1 begins about five minutes into the counselling session. After an exchange between the couple, GC1 focussed them back on their options (line 39), reiterated their options and tailors this to them individually (line 56), taking into account her understanding of their personal beliefs which she ascertained previously (line 42).

**Session 6 Extract 1**

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>36</td>
<td>Paula</td>
<td>Having the test you can do nothing other than abort the child is that right?</td>
</tr>
<tr>
<td>37</td>
<td>GC1</td>
<td>If you have the test?</td>
</tr>
<tr>
<td>38</td>
<td>Paula</td>
<td>Yeah.</td>
</tr>
<tr>
<td>39</td>
<td>GC1</td>
<td>If you have the test then you have got the option of either being prepared or interrupting the pregnancy whatever you choose to do. Have you discussed that between the two of you?</td>
</tr>
<tr>
<td>40</td>
<td>Paula</td>
<td>Yeah.</td>
</tr>
<tr>
<td>41</td>
<td>Ron</td>
<td>Yeah.</td>
</tr>
<tr>
<td>42</td>
<td>GC1</td>
<td>What would you do?</td>
</tr>
<tr>
<td>43</td>
<td>Ron</td>
<td>We have discussed it and I don’t know. I guess part of having today’s meeting or coming in today is about trying to find out some information so that we can make a decision before we start. I guess Paula’s view is and probably mine too is to some degree is once you start something knowing the risk factors you probably wouldn’t abort anyway. You make a decision and you decide to go ahead in that direction, so I think am I right in saying that?</td>
</tr>
<tr>
<td>44</td>
<td>Paula</td>
<td>[4] The problem thing is interesting, you have switched into management mode like you are having a meeting at work.</td>
</tr>
<tr>
<td>45</td>
<td>Ron</td>
<td>No I’m not. Verbal exchange</td>
</tr>
<tr>
<td>56</td>
<td>GC1</td>
<td>Yeah so you might decide that you don’t want to have a test or you might like to know just to be prepared.</td>
</tr>
</tbody>
</table>

Following on from her explanation about maternal age risks and chromosomes, GC1 explains the clinical features of Down syndrome and gives them the opportunity to ask questions (line 66). They return to the description of chromosomes and the
counsellor checks their level of understanding by asking about their background (line 68). Paula signals that she is hoping the information won’t be too complex (line 72) but GC1 does not explore this further and continues with her explanation.

Session 6 Extract 2

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>66</td>
<td>GC1</td>
<td>Yeah if you have any other questions about that is the main condition that we will be talking about today and...[pause]</td>
</tr>
<tr>
<td>67</td>
<td>Ron</td>
<td>You said something as well about [4.0] chromosomes was it?</td>
</tr>
<tr>
<td>68</td>
<td>GC1</td>
<td>Chromosomes yeah I will show you the little chromosomes, are either of you from a science background?</td>
</tr>
<tr>
<td>69</td>
<td>Paula</td>
<td>No.</td>
</tr>
<tr>
<td>70</td>
<td>Ron</td>
<td>No.</td>
</tr>
<tr>
<td>71</td>
<td>GC1</td>
<td>I just wanted to make sure talking to a scientist about this [laugh]. I’ll just give you a very brief outline of genetics and biology. Basically we are made up of billions of cells and what we are interested in is that little nucleus and the wiggly things are called the chromosome.</td>
</tr>
<tr>
<td>72</td>
<td>Paula</td>
<td>You are talking in little baby steps here.</td>
</tr>
<tr>
<td>73</td>
<td>GC1</td>
<td>[laughs] Excellent, they look a little bit like spaghetti.</td>
</tr>
</tbody>
</table>

The genetic counsellor then appeared to try to find some common ground between the couple as she attempted to help them reconcile their apparently competing goals. Ron felt the need to balance the risk of having a child with a chromosome abnormality and Paula’s desire to have another baby. In extract 3, GC1 asked them if they are “on the same wavelength” (line 392) and drew attention to the movement that occurred during the session by asking them if they felt closer to an understanding (line 394). What follows is a series of apparently disjointed exchanges until lines 417-426 when Ron and Paula are able to get their differing points of view across. Ron was able to identify the source of his fears, allowing the GC to address their prior experiences with pregnancy loss and how this understandably affects their risk perception (line 429). At the beginning of the session Ron was reluctant to engage in the discussion or give the counsellor any information about his thoughts and feelings, so GC1 worked hard to include him in the discussion in line 427, 429 and 431. It appears that she gained his trust, enabling
him to explain his feelings to Paula who, prior to hearing them here, appeared to have formed different opinions about what he wanted.

Session 6 Extract 3

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>392</td>
<td>GC1</td>
<td>And do you think you are kind of both on the same wavelength?</td>
</tr>
<tr>
<td>393</td>
<td>Paula</td>
<td>No I am on a totally different wavelength. But he has finally decided to join me, sort of.</td>
</tr>
<tr>
<td>394</td>
<td>GC1</td>
<td>Are you further apart at the end of this appointment, or closer?</td>
</tr>
<tr>
<td>395</td>
<td>Ron</td>
<td>Yes</td>
</tr>
<tr>
<td>396</td>
<td>GC1</td>
<td>Are you?</td>
</tr>
<tr>
<td>397</td>
<td>Paula</td>
<td>What? How do you mean?</td>
</tr>
<tr>
<td>398</td>
<td>GC1</td>
<td>In terms of</td>
</tr>
<tr>
<td>399</td>
<td>Paula</td>
<td>You what?</td>
</tr>
<tr>
<td>400</td>
<td>GC1</td>
<td>In what sense?</td>
</tr>
<tr>
<td>401</td>
<td>Paula</td>
<td>I don’t understand what you are talking about explain.</td>
</tr>
<tr>
<td>402</td>
<td>Ron</td>
<td>Further apart in our decision before we come in here [laugh]. No so oh look the chromosome one worries me.</td>
</tr>
<tr>
<td>403</td>
<td>Paula</td>
<td>Do you think Down syndrome was the only disability that could happen? You know there is other.</td>
</tr>
<tr>
<td>404</td>
<td>Ron</td>
<td>Oh look I knew there was others.</td>
</tr>
<tr>
<td>405</td>
<td>Paula</td>
<td>Interrupts</td>
</tr>
<tr>
<td>406</td>
<td>Ron</td>
<td>No, don’t try and shout us down it is wrong. I didn’t know it was that high a risk I knew there was other things possible that’s true I did but I didn’t know the risk factor was 1 in 39. In all fairness I didn’t so does it bring us further apart not really I did say yes it does but I was actually joking.</td>
</tr>
<tr>
<td>407</td>
<td>Paula</td>
<td>Oh ok.</td>
</tr>
<tr>
<td>408</td>
<td>GC1</td>
<td>Hmmm but you</td>
</tr>
<tr>
<td>409</td>
<td>Ron</td>
<td>Does it make me a bit more apprehensive yes it does</td>
</tr>
<tr>
<td>410</td>
<td>GC1</td>
<td>Yep</td>
</tr>
<tr>
<td>411</td>
<td>Ron</td>
<td>No I know we are not really any further apart but I need to go we need to go and think about it talk about it probably have a few of these discussions.</td>
</tr>
<tr>
<td>412</td>
<td>Paula</td>
<td>I want to know what you have done?</td>
</tr>
<tr>
<td>413</td>
<td>GC1</td>
<td>Yeah but you are still supportive but just more concerned. Either way I guess I have given you a lot of information and you probably think OK there’s Down syndrome and now I have pointed out all these other things but even regardless in any pregnancy you have got the risk of all these things it just when you get it all congested into one appointment it sounds more scary and</td>
</tr>
<tr>
<td>414</td>
<td>Paula</td>
<td>For me it hasn’t changed a thing just got another four weeks [laugh].</td>
</tr>
<tr>
<td>415</td>
<td>GC1</td>
<td>Wait a bit longer?</td>
</tr>
<tr>
<td>416</td>
<td>Paula</td>
<td>Yeah</td>
</tr>
<tr>
<td>417</td>
<td>Ron</td>
<td>Oh look you say that now but you may stroll, think these things through and you might think slightly differently doesn’t mean you won’t say yes or no doesn’t mean your passion to have another baby will change but might not change anything it might just [3.0] just has it</td>
</tr>
</tbody>
</table>
changed our position or not yes it has slightly only because it is slightly different it doesn’t mean it is no. We need to think about it that’s all.

418 Paula I’m gonna have a cry now.
419 Ron Oh.
420 GC1 There are some tissues there.
421 GC1 [Crying] you are the most frustrating man on the planet.
422 Paula Even just in coming here it is obviously a supportive thing.
423 Paula No you’re a bugger [laugh].
424 GC1 I mean are you in any way, I mean....
425 Paula You probably go home and go...
426 Ron I have had 14 years you have got to remember that I have said no for 14 years you have got to try and keep that in your head and then start saying that I am trying to do something I’m not. I said I’m not so against that idea I said that a year ago but now I am trying to just, I’m, it. It is ok for you to say yes, yes you want this but I have also got to try and make sure that the family stays together and keep the pieces together as much as you do so I would just like to know what is going on and I am not against you I am with you alright it might feel as though I am but I’m not.
427 GC1 If we could say right now if you could guarantee it was going to be a healthy baby you would be fully supportive of that?
428 Ron Yeah it is easy to say but what about the other way just at the moment?
429 GC1 Yeah but in the context of the history it must be scary to have all these options and things that might go wrong. And probably having had those miscarriages makes it even more difficult in both of your situation.
430 Ron It wasn’t any help having the miscarriages before.
431 GC1 But emotionally it obviously affected you as well. So you have that pain in your mind of losing and it is scary to contemplate putting yourself at risk of that happening again.
432 Paula It would be devastating but it doesn’t really come into the equation.
433 Ron Mmm.
434 GC1 You just desperately want to have a baby?
435 Paula If you don’t get pregnant then you’re not going to have one anyway are you so.
436 Ron Yeah.
437 GC1 Yeah. But it sounds like you just desperately want a chance.

The extracts show that the couple were conflicted about the decision making process and GC1 attempted to act as a mediator, enabling them to express their feelings and be heard by each other. Ron, in particular, moves a long way from his initial reticence to divulge any information (saying it was between him and his wife) to expressing so clearly where his reticence comes from (line 426). GC1 helped Ron
to communicate his views to Paula (line 417), in particular, that even though he was undecided about another pregnancy he understood the risks of a birth defect.

There are examples where GC1 acknowledges the difficulty of the decision the couple face (lines 413 and 428) and the problems that arise when both parties do not agree (lines 392, 394 and 429). The difficulty they appeared to have in expressing their views to each other and negotiating differing opinions may have been unique to this situation or a recurrent problem in their relationship and may have been more appropriately dealt with in ongoing couples therapy.

Although the interaction appeared difficult to manage at times, GC1 was able to inform the couple about their options and assist them with their decision about prenatal testing. She outlined the agenda at the start of the session, and searched for evidence of prior knowledge or experience from their peer group. She was able to navigate the differences in education levels and health literacy between her and the couple that left unexplored may have caused barriers between them.

**Session Seven**

Session Seven involved Jane, who presented for carrier testing after her newborn nephew was diagnosed with CF, and a female genetic counsellor (GC2). The genetic counsellor and client were Caucasian and similar in age and professional qualifications. Prior to Jane attending the clinic, she had telephone contact with the genetic counsellor and had an opportunity to consider some information about CF.

The first part of the session focussed on the facts and genetics relating to CF. There was a long exchange between counsellor and client that explored the probability of Jane and her partner having a child with CF (Extract 1). The counsellor used a variety of different ways to present probability and risk such as “the chances are slim” (lines 63) “it is possible” (line 65) “one in four” (line 69) and “75% chance” (line 77). The representations of risk ranged from numbers to proportions and words, often mirroring Jane’s language when she referred to chance (line 64) and being unlucky
This shows GC2’s flexibility in paying attention to the information needs of the Jane and following her lead.

*Session 7 Extract 1*

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>59</td>
<td>GC2</td>
<td>The deltaF508</td>
</tr>
<tr>
<td>60</td>
<td>Jane</td>
<td>So often you often have one mutation per family?</td>
</tr>
<tr>
<td>61</td>
<td>GC2</td>
<td>Yes the only other different mutation would come in from the other, would come in from the father’s side.</td>
</tr>
<tr>
<td>62</td>
<td>Jane</td>
<td>Ok so but what about in our family you couldn’t have something else?</td>
</tr>
<tr>
<td>63</td>
<td>GC2</td>
<td>The chances are very slim but it is possible the difficulty with genetics and medical is you never say never do you cause as soon as you say never it’ll happen [laughs] and you’ll get a different mutation do you know what I mean than your sister or something like that.</td>
</tr>
<tr>
<td>64</td>
<td>Jane</td>
<td>But it’s unlikely cause one of the kids would have shown up something cause you would have had two sets or something no?</td>
</tr>
<tr>
<td>65</td>
<td>GC2</td>
<td>Well it is possible that this side of the family here your dad’s side has got the deltaF508 and your mum’s might have an R117H mutation coming through because the reason why I say it is possible is that they say that of the general population walking around 1 in 25 people are carriers.</td>
</tr>
<tr>
<td>66</td>
<td>Jane</td>
<td>Ok that’s pretty..</td>
</tr>
<tr>
<td>67</td>
<td>GC2</td>
<td>Yes it is very common and that’s why we kind of do the testing and do the newborns. So technically it is possible that there is something coming through your mum’s but all your kids are a carrier or you’ve skipped being a carrier but the chances of your Dad being a carrier and your Mum being a carrier and not having an affected child is pretty good.</td>
</tr>
<tr>
<td>68</td>
<td>Jane</td>
<td>Yeah [Interruption]</td>
</tr>
<tr>
<td>69</td>
<td>GC2</td>
<td>But it is possible that both your Mum and Dad are a carrier and they haven’t had any children with cystic fibrosis because the chance is if you work it out if Mum and Dad are both carriers and it is the same for Cassie and her husband the chance of having an affected child is 1 in 4 with every pregnancy. So every pregnancy is a new event. So you can have all affected children but the chance is 1 in 4 every time and that’s just because, it comes down to this being a carrier (I’ll show you the picture) and that’s why we are testing you so in the egg you start off with your full complement. And here’s the sperm with the both copies of the number 7 chromosome then what happens that will split down and just put one into each because you only want to pass on half your genetic material.</td>
</tr>
<tr>
<td>12</td>
<td>Jane</td>
<td>Yep</td>
</tr>
<tr>
<td>13</td>
<td>GC2</td>
<td>And so for the sperm and let’s say this is the one with the mistake in it. The sperm has got a mistake in the CF gene so that one goes there and that one goes there and then it just pure mathematics</td>
</tr>
</tbody>
</table>
the chance that that is fertilised by that one.

14 Jane Yep
15 GC2 [2.0] That would be a baby with cystic fibrosis because both genes aren’t working that one will go with that one [3.0] only one’s not working so that would be a carrier and no signs or symptoms like Cassie she is well and all that sort of stuff. You wouldn’t think she had cystic fibrosis and same again you just repeat the process so you get a 1 in 4 chance, chance you can have a carrier or non-carrier, carrier is 50 and affected is 1 in 4 so even if you have two carriers who are definitely known carrier there is still only a 1 in 4 chance of having a bub with cystic fibrosis.

16 Jane Geez it’s pretty unlucky.
17 GC2 Yeah cause when you flip it around 1 in 4 it means 3 out of 4 you should you know what I mean like there’s.
76 Jane 75% chance [ Interruption].
77 GC2 75% yeah.

GC2 goes on to explain the options available to Jane and her husband if they were shown to be CF carriers. Because Jane has asked for information about PGD for her sister, GC2 explores Jane’s thoughts and beliefs about prenatal diagnosis and PGD in Extract 2. Jane is concerned about a clash between her beliefs and that of her family and the dilemma of having a TOP for a condition that a living family member has.

The discussion about these issues appears to be open and frank. GC2 has an easy conversational style and does not avoid these difficult and emotional topics. She acknowledges the difficulty of the situation and gives Jane the opportunity to discuss this further (line 129). She asks a direct question about whether Jane is feeling any pressure from her family (line 138).

Session 7 Extract 2

<table>
<thead>
<tr>
<th>Line</th>
<th>Speaker</th>
<th>Content</th>
</tr>
</thead>
<tbody>
<tr>
<td>127</td>
<td>GC2</td>
<td>Why you’d do PGD? Yep would that be you know as we sort of talk about all this sort of stuff what do you think about would you just sort of get pregnant and take the chance or definitely do the PGD?</td>
</tr>
<tr>
<td>128</td>
<td>Jane</td>
<td>Well my family is very religious so they are completely against any sort of abortion or anything like that but I am not [laugh]</td>
</tr>
<tr>
<td>129</td>
<td>GC2</td>
<td>Yeah OK it is difficult.</td>
</tr>
<tr>
<td>130</td>
<td>Jane</td>
<td>It would be frowned upon if I went and did that I think.</td>
</tr>
<tr>
<td>131</td>
<td>GC2</td>
<td>But PGD would be acceptable to the family?</td>
</tr>
<tr>
<td>132</td>
<td>Jane</td>
<td>Yes it would because it’s before you implant it you know what I mean? Yep I’m always in grey territory with that sort of thing anyway, my husband is like has no problem with abortion. I know his previous girlfriend has had one but I would be I would prefer not to even get</td>
</tr>
</tbody>
</table>
pregnant and have to face that because then maybe if I’m pregnant I wouldn’t want to get rid of it and look at Felix you know he is fine he is happy and everything and sure he may have a reduced life expectancy but his quality of life is quite good so faced with that I might not want to get an abortion so I’d rather not actually have to go through that so that’s definite.

133 GC2 It’s a very tricky situation that’s why I guess we’re here do you know what I mean so before you have even done your testing we work out how you are feeling what would you do and for some people they just don’t want to know because they don’t want to have to make a decision do you know? [Interruption]

134 Jane I know.

135 GC2 You know if you are a carrier and your husband’s a carrier what are you gonna do you know does that set up more anxiety rather than less anxiety do you know cause at the moment you are like no let’s just go for it and see.

136 Jane Yeah

137 GC2 So are you feeling pressure from your family or are you feeling exactly the same way as your family about the religious side of things and ending pregnancies? [2.0]

138 Jane [2.0] how do you mean?

139 GC2 Like as in some people say I feel pressure from my parents I couldn’t tell them what my decisions were and just do it without them knowing.

140 Jane Yeah.

141 GC2 Or would you do you know like feel exactly the same way as your family and you don’t want to actually end a pregnancy?

142 Jane No I wouldn’t have a problem with it. [Interruption]

143 GC2 So the two of you are on the same page.

144 Jane Yeah we are we are definitely.

GC2 appears to have developed a good rapport with Jane during her counselling session, during which there is a mixture of information giving and attention to emotional issues. GC2 displays a flexible style and often follows Jane’s lead into topics and issues of concern to her, thus allowing Jane to explore these further.

7.3.2.2. **Summary of sessions and clinical impressions**

After many hours listening to and transcribing the clinic sessions, it was apparent that both genetic counsellors and clinical geneticists spoke more often and for longer than their clients. Often this is necessary to explain complex information and options to clients. In the sessions presented here the clients occasionally interrupted and the genetic counsellors appeared to interrupt less and allowed clients time to ask questions and express their concerns more than the geneticist.
As described in a previous study (Meiser et al., 2008) both geneticist and counsellor tended to draw the conversation back to their own agenda and frequently reverted to the teaching model.

In this exploratory examination of a small sample of genetic counselling sessions, the dominant approach appeared to be a comfortable conversational style that allowed room to negotiate communication barriers. It is not possible to know whether the GHPs were mindful of differences in cultural background in these counselling sessions. However, it is apparent they were aware of differences in education and socio-economic levels and attempted to establish rapport so that clients were comfortable to discuss difficult issues with a relative stranger. Where cultural mismatches were recognised, attempts were made to address them through rapport building and ascertaining prior knowledge and concerns.

Building rapport through techniques such as social conversation and mirroring language illustrates some of the core skills required for genetic counselling. The ability to adapt to cultural difference is the extension of these skills. Examples of the assessment of cultural background being made in the recorded sessions included GC2 exploring religious beliefs about abortion, DR1 asking questions about occupation to obtain information about education levels and GC1 asking about science background to determine level of genetic understanding and health literacy. These questions are a basic assessment yet they require care to avoid misunderstanding or incorrect assumptions about knowledge levels.

The GHPs explained complex information and turned the educational components of the session into a two way interaction. For example, in Session Seven, GC2 explained chance, risk and statistics using simple language that mirrored terms her client used. In Session Five, DR1 used a simple analogy to explain the function of tumour suppressor genes. Both customised their sessions to suit the individual
needs of their clients. Repetition and varied ways of presenting information helps the listener to understand and remember the content (Bernhardt et al., 2000).

Interruptions to the flow of conversation were common with both client and GHPs interjecting while the other was speaking. This is common in general conversation. However, in the context of a genetic counselling session, interruptions by a GHP may leave the client feeling their concerns have not been heard or addressed. Joan appeared to become frustrated and withdrawn after stating she had enough information about genes yet the geneticist continued. He did not really engage with her about HRT or ovarian cancer screening and she may have wanted to spend more time exploring these topics. Interrupting the speaker can be a technique used to avoid topics that evoke emotional responses and are psychological in nature (Roter et al., 2008). It is important that GHPs are aware of any reticence on their part to explore these topics and the impact of avoiding them on client outcomes.

Clients do digress during sessions and maintaining focus could be seen as skilfully managing the limited amount of time available. However, when DR1 brought Joan back to his agenda in Session Five he addressed topics he thought were important rather than listening and exploring the issues that were important for the client. A more interactive approach would see GHPs giving clients more opportunity and assistance to articulate their concerns, then focusing on addressing these concerns and the issues underlying them.

Effective communication is due to both the skill of the counsellor and characteristics of the client. In Session Six extract 1, Ron appeared to be reluctant to engage with GC1. It is possible that Ron was defensive because didn’t understand what GC1 was saying or the reason for the consultation. Reluctance to engage with counselling can be due to many factors including anxiety, not understanding the purpose of genetic counselling, cultural differences and gender issues (Ulph et al., 2010).
7.3.3. Post-session questionnaires

This section presents the data collated from the post-session questionnaires given to both the client and GHP to complete. The data from seven pairs of questionnaires is presented. In sessions involving more than one client, the questionnaire was completed by one client only. If there was a geneticist and genetic counsellor present in the session, the questionnaire was completed by the geneticist. The data from questions addressed to both provider and client is discussed first, followed by the questions pertinent to either the client or GHP.

7.3.3.1. Agenda setting

GHPs and clients were asked to list the four most important issues they thought should be covered in the sessions. There was some agreement between the providers’ and clients’ listed responses (Table 7.2). However, while GHPs might assume they know what is important for clients, and there may be some overlap in the issues deemed important, in making assumptions they may neglect to ascertain what is actually important for the individual. This may arise if genetic services have protocols regarding the content of counselling sessions that do not allow for clients to identify the issues important to them. Some practitioners, particularly less experienced ones, may find comfort in following an agenda and neglect to deviate from it sufficiently to explore client concerns (Runyon et al., 2010).
7.3.3.2. **Awareness of cultural differences**

One of the main aims of this exploratory work was to explore whether clients and GHPs noticed cultural differences and if they did, how they interpreted them. The data in Chapter Five indicated that genetic counsellors tend to take a quite narrow view of cultural difference. In the post-session questionnaires the GHPs and clients identified country of origin as a source of cultural difference and also noted gender, education levels, employment, immigration, life experience, marital status and parenthood, suggesting a broader appreciation of cultural difference.

Participants in each genetic counselling session were asked if they felt they shared a similar background and whether or not they had things in common. While acknowledging it may not be possible to determine this after a single session, the question was designed to see if clients and GHPs made instant assessments about each other based on visible characteristics. The paired comments in response to this question (Table 7.3) indicate that perceived differences are not as important to the client as the GHP showing understanding, empathy and an ability to address their concerns. Few differences were identified and similarities were pointed out more often than differences. While all individuals are culturally different to some degree, this suggests that cultural differences can be overcome if GHPs engage with the client and address their concerns. Thus cultural similarity is not a requirement for successful counselling.
Table 7.3  Perceived elements of shared cultural background

<table>
<thead>
<tr>
<th>Session</th>
<th>Client response</th>
<th>Counsellor response</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>She spoke to me about her experiences doing her Masters so I could relate to my experiences with doing higher degree research</td>
<td>Almost identical background</td>
</tr>
<tr>
<td>2</td>
<td>The GC came across a caring human being with the best interests of her clients at heart</td>
<td>Similar socio-economic background no obvious differences, although we are in different stages of our life – in terms of family</td>
</tr>
<tr>
<td>3</td>
<td>I would doubt if there were any similarities [between our cultural backgrounds]; however, I feel that whoever you meet given time you will always find something in common. The fact that we were having that interview gives us something in common</td>
<td>From similar background</td>
</tr>
<tr>
<td>4</td>
<td>We possibly would have some things in common but we were not there for that purpose. It was too small a timeframe to establish a connection</td>
<td>From similar background</td>
</tr>
<tr>
<td>5</td>
<td>Similar background and country of origin</td>
<td>I felt that she and her husband were mirror images of myself, for the reasons they came to Australia and from the same upbringing</td>
</tr>
<tr>
<td>6</td>
<td>She was down to earth and I could communicate with her</td>
<td>Similar age, family came to Australia, married female</td>
</tr>
<tr>
<td>7</td>
<td>Not from similar background</td>
<td>From similar background, she was interested in biology</td>
</tr>
</tbody>
</table>

7.3.3.3.  In session communication

In the post-session questionnaire, participants were asked to mark on a pie chart the proportion of the session that they spent either talking or listening. The data was then compiled to show the proportion of time each party thought they spent talking (Figure 7.1). The data illustrates large discrepancies in assessments of how time was spent in a session. In Sessions 1-4 both parties thought they were talking for large amounts of the allocated time (positive discrepancy) and in Sessions 5-7 neither thought they were talking for large amounts of the allocated time (negative
discrepancy). In Session 7 the counsellor thought that they were talking for less than 5% of the session and the client 45%, which would leave 50% of the session with neither talking. Short silences are a normal occurrence but it would be highly unusual to have silence for half a session. This amount of silence is also not reflected in the session transcript.

The actual time allocations in a session could be accurately measured from the transcripts; however, this was not done as the purpose was to assess each party’s perception of how the time was spent. If the client felt that they spent a lot of time talking, they may feel more positive about whether or not their concerns were addressed. If the GHP underestimates the amount of time they spend listening to the client, this may leave the client with insufficient opportunity to express their concerns or feelings.

![Figure 7.1 Perceived time allocations in genetic counselling sessions](image)

Figure 7.1 Perceived time allocations in genetic counselling sessions

Participants were asked to mark on a pie chart the proportion of time spent talking about factual information and the time spent talking about feelings (psychosocial issues). Talking about feelings was further broken down into four categories: giving/being given factual information; eliciting/being asked about worries; being able/listening to talk about feelings; and exploring/talking about personal or family
experiences of genetic conditions. In Table 7.4 the results are displayed as a percentage of the sessions each participant thought was spent on each topic. Some interactions (Sessions 1, 4 and 7) show large discrepancies and others (Session 6) show close agreement in the estimations. The intention in this study was to explore participants’ perceptions of what occurred in the sessions. The topics discussed and time spent on each could have been measured accurately using tools such as the Manchester Observation Code (Liede et al., 2000).

Table 7.4 Perceived percentage of the session spent on each topic

<table>
<thead>
<tr>
<th>Time spent on topic</th>
<th>Participant</th>
<th>1</th>
<th>2</th>
<th>3</th>
<th>4</th>
<th>5</th>
<th>6</th>
<th>7</th>
</tr>
</thead>
<tbody>
<tr>
<td>Factual information</td>
<td>Client</td>
<td>45</td>
<td>25</td>
<td>35</td>
<td>32</td>
<td>25</td>
<td>75</td>
<td>13</td>
</tr>
<tr>
<td></td>
<td>GHP</td>
<td>20</td>
<td>47</td>
<td>75</td>
<td>91</td>
<td>36</td>
<td>87</td>
<td>66</td>
</tr>
<tr>
<td>GHP determining clients worries</td>
<td>Client</td>
<td>25</td>
<td>25</td>
<td>28</td>
<td>12</td>
<td>25</td>
<td>8</td>
<td>13</td>
</tr>
<tr>
<td></td>
<td>GHP</td>
<td>10</td>
<td>15</td>
<td>8</td>
<td>6</td>
<td>20</td>
<td>3</td>
<td>5</td>
</tr>
<tr>
<td>Client talking about feelings</td>
<td>Client</td>
<td>15</td>
<td>25</td>
<td>20</td>
<td>12</td>
<td>25</td>
<td>8</td>
<td>40</td>
</tr>
<tr>
<td></td>
<td>GHP</td>
<td>45</td>
<td>33</td>
<td>8</td>
<td>3</td>
<td>19</td>
<td>3</td>
<td>10</td>
</tr>
<tr>
<td>Personal experience</td>
<td>Client</td>
<td>15</td>
<td>25</td>
<td>17</td>
<td>45</td>
<td>25</td>
<td>8</td>
<td>34</td>
</tr>
<tr>
<td></td>
<td>GHP</td>
<td>25</td>
<td>5</td>
<td>8</td>
<td>0</td>
<td>25</td>
<td>6</td>
<td>10</td>
</tr>
</tbody>
</table>

7.3.4. Post-session evaluation of clients’ experiences

7.3.4.1. Importance of cultural difference

In response to questions asking whether or not the client thought their counsellor shared a similar background to them, the clients indicated in their questionnaire responses that any differences did not hinder the ability to have their issues addressed. This may reflect the skills of the counsellors in establishing rapport and overcoming any potential barriers to communication or may reflect a lack of any perceived differences. When asked if they would have preferred a counsellor who had a similar background to them, the majority of clients said they did not and
wrote responses explaining that the knowledge and empathy demonstrated by the counsellor was more important.

7.3.4.2. **Impact of GHPs’ personal style**

Clients were also asked in the post-session questionnaire about the qualities of their counsellors that made either a positive or negative impression on them. No comments about negative impressions were made, suggesting that either there were none, or that clients were too uncomfortable to respond in a negative way. The factors that left a positive impression with the clients are listed in Table 7.5.

7.3.4.3. **Clients’ experiences of genetic counselling**

The final question on the post-session questionnaire gave the opportunity for the clients to give feedback about their experience of genetic counselling. As shown in Table 7.6, their experience of genetic counselling was generally positive.

<table>
<thead>
<tr>
<th>Session</th>
<th>Positive impressions</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>She was smiling, kind and confident. She called me by my first name and allowed me time to say what was on my mind</td>
</tr>
<tr>
<td>2</td>
<td>She was very approachable and real. She had an incredible warmth in her manner but is also unobtrusive and not judgemental</td>
</tr>
<tr>
<td>3</td>
<td>The knowledge of the subject and the way it was conveyed</td>
</tr>
<tr>
<td>4</td>
<td>He gave the impression of much experience and understanding of genetics and our concerns and did his best to deal with them</td>
</tr>
<tr>
<td>5</td>
<td>Very caring and willing to let us talk</td>
</tr>
<tr>
<td>6</td>
<td>Very knowledgeable, her caring manner</td>
</tr>
<tr>
<td>7</td>
<td>She was really nice and understanding</td>
</tr>
</tbody>
</table>
Table 7.6  Clients’ experiences of genetic counselling

<table>
<thead>
<tr>
<th>Client</th>
<th>Quotes</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Being armed with knowledge and an understanding genetic counsellor I am far more confident about my future than I was a few months ago!</td>
</tr>
<tr>
<td>2</td>
<td>It was a very positive experience for me and I am very grateful to the GC for her advice, information and support</td>
</tr>
<tr>
<td>3</td>
<td>After my visits all the questions that were going through my head were answered and any areas that I didn’t understand were fully explained</td>
</tr>
<tr>
<td>4</td>
<td>I found this experience to be positive and was reassured that this was the correct step to take. Our discussion with the counsellor was excellent and we walked away confident of the advice and our understanding of our situation</td>
</tr>
<tr>
<td>5</td>
<td>Although the session has not brought me closer to a decision it did have a calming effect on me and made me realise there was no rush to making the decision</td>
</tr>
<tr>
<td>6</td>
<td>For me I had a lot to take in and not sure I was fully retaining the information. With everything going on I was on information overload</td>
</tr>
</tbody>
</table>

7.3.5. Post-session evaluation of GHP’ experiences

7.3.5.1. Cultural difference

When asked about their impressions of clients’ cultural backgrounds in the post-session questionnaire, most GHPs used common demographic, ethnic, religious and educational terms to describe their clients’ background. One genetic counsellor commented that although she had formed pre-conceived ideas about the client and her behaviour prior to the session, she tried to be open-minded and was surprised to discover they had more in common than she had anticipated.

7.3.5.2. Impact of personality factors on rapport

The GHPs were also asked about the qualities in their clients that made either a positive or negative impression on them. The question was similar to that posed to clients, but focussed on whether they felt they had established a good rapport (and if not, why not), and whether they formed any negative impressions of their clients.
The responses provided by the GHPs (Table 7.7) suggest that building rapport and engaging with clients is not always easy for these GHPs.

**Table 7.7  Qualities of clients that made impressions on genetic health providers**

<table>
<thead>
<tr>
<th>Session</th>
<th>Rapport developed</th>
<th>Negative impressions</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>With the woman, yes, but the husband didn’t seem to like me at the beginning (or didn’t want to be there), I had to earn his respect</td>
<td>She seemed a bit selfish in her views at times</td>
</tr>
<tr>
<td>3</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>4</td>
<td>Yes</td>
<td>No</td>
</tr>
<tr>
<td>5</td>
<td>Yes we had plenty of common ground for rapport building</td>
<td>She was stuck in her situation but I understood what a difficult and important decision she was making</td>
</tr>
<tr>
<td>6</td>
<td>Yes</td>
<td>She was a little concerned about the recording process and held back</td>
</tr>
<tr>
<td>7</td>
<td>She was quite responsive and open with me</td>
<td>She was wearing a baseball cap so her face was often obscured and she had her hands in her pockets</td>
</tr>
</tbody>
</table>

**7.3.5.3. The overall experience of genetic counselling**

The final question on the questionnaire invited GHPs to make any additional comments on the session and responses are shown in Table 7.8. The comments were generally positive, although two participants noted the impact of recording the session on those present. GC2 reported being overly aware that the session was being taped and felt stressed because the appointment wasn’t as straightforward as she had anticipated. She was also concerned the client’s voice was not loud enough to be recorded successfully. DR6 reported that her client asked questions after the recording finished and the session continued for a further twenty minutes. The client reported that she had some anxiety about the recording on her post-session questionnaire so this recorded session was not included in the analysis.
When recording live counselling sessions, it is not possible to remove the influence of the recording on the way the session was conducted or how the clients behaved in the session. I did not ask either group if they thought recording the session influenced the session but these comments indicate it might have done so for some participants.

Table 7.8  GHPs’ experiences of the sessions

<table>
<thead>
<tr>
<th>GHP</th>
<th>Quotes</th>
</tr>
</thead>
<tbody>
<tr>
<td>GC2</td>
<td>I think I was overly aware of the session being taped and stressed that the appointment wasn’t as straightforward as I had anticipated. I went in with my information giving agenda but I think the couple’s issue with the actual decision to have a baby was more important to focus on</td>
</tr>
<tr>
<td>GC5</td>
<td>I think it was a good session. I liked the fact the husband was there for his wife, I admired that</td>
</tr>
<tr>
<td>DR6</td>
<td>When I turned the recorder off she asked me about mastectomy and started discussing her feelings about this and the session continued for another 20 minutes</td>
</tr>
<tr>
<td>GC7</td>
<td>It went better than I had anticipated. I found her a pleasant young woman who was interested in what I had to say and some of the information. I found her relatively easy to engage</td>
</tr>
</tbody>
</table>

7.4. Discussion

This preliminary, exploratory study of live genetic counselling clinic consultations examined the content of sessions, communication style and whether or not counsellors and clients adjusted this in order to negotiate cultural differences. Individual sessions were examined and the post-session questionnaires were used to assess and compare the experiences and perspectives of both client and GHPs.

When asked whether they perceived a cultural difference between themselves and the other person present in the session, some clients and GHPs identified sources of cultural difference but neither seemed uncomfortable with them. The cultural differences that were identified were primarily based on physical appearance and gender. This is not unexpected, as more private cultural differences may not be
routinely identified as part of a counselling session. Clients were generally happy with their GHP and having a provider who attended to their questions or concerns was more important than whether or not they were culturally similar. When clients and GHPs perceived themselves to be similar or have similarities, this had the most impact on the session.

The GHPs did not give clients information about themselves but did check for points of difference that would impact on their interaction with their clients. The GHPs employed a range of strategies to effectively engage with their clients. Techniques such as matching language styles, enquiring about personal factors and interests, and relaying information in simple language with a personal focus were evident in the sessions. Conversation style and flow was also adapted to suit the client.

Examining the conversation structure in the sessions showed that turn taking is often limited, with both counsellors and clients interrupting the flow of conversation. The GHPs often spoke for lengthy periods, dominating the conversation and thus limiting the speaking time available to the client. Less dense communication with more speaking turns leads to greater interactivity and a more balanced, interactive and personalised format (Babul-Hirji et al., 2010; Roter et al., 2008). Clients may benefit more from counselling sessions if they have the opportunity to explore their feelings and concerns, regardless of the amount of time spent by the GHP giving factual information. While explaining complex information in a simple manner and facilitating a client’s understanding of a complex topic is important, exploring the thoughts, feelings and experiences of the client may be equally, or more, important (Ellington et al., 2011; Evans et al., 2004).

7.4.1. Research with colleagues as participants

Studies of actual genetic counselling sessions are important if we are to understand the interactions that occur between counsellor and client, and continue to improve practice. As Kessler explains, this aspect of the process of communication is often
neglected and the analysis and publishing of session transcripts is important in contributing to understanding the strengths and weaknesses of genetic counsellors, and in continuing to improve counselling and communication (Kessler, 1998).

Reflective practice has been incorporated into the requirements for certification by the ASGC. The aim is to encourage trainee counsellors to develop core competencies in the use of counselling skills, in addition to a client-centred approach, and the ability to reflect on the client–counsellor dynamics and self-awareness (Australasian Society of Genetic Counsellors, 2011). In this study, one colleague indicated she found it stressful having the session recorded, possibly reflecting concerns about scrutiny of her practice. The introduction of self-reflective practice to the certification process may mean that in the future this type of work may be viewed as a standard way to undertake professional development.

Undertaking the research with colleagues in my own department created a number of difficulties and imposed some limitations. Whilst understanding the importance of reflective practice, it was difficult to scrutinise and critique the work of my colleagues. It was also difficult to formulate a method of inviting colleagues, some of whom were also my friends, to participate in a manner that enabled them to make an informed and autonomous decision, free from the pressure of our professional and/or personal relationship. It was important that they did not feel coerced into participating. A written invitation to participate was chosen and this method allowed my colleagues to consider their willingness to participate in their own time and afforded them the option of declining in writing rather than having to negotiate the discomfort of declining in person. Two colleagues did not respond to the invitation to participate in the research and I did not approach them, or remind those that did not progress to recording sessions to avoid applying pressure or an obligation to participate.
The small number of sessions recorded was a consequence of the decision to undertake the work within my own unit, and the constraints placed upon recruitment by the hospital ethics committee. The hospital ethics committee approved the study with the proviso that I did not directly approach the clients. This meant that in addition to asking my colleagues to participate in the research, their input was also required for the recruitment of clients and the subsequent coordination of clinic appointments. This methodology placed an additional burden on the already limited time resources of my colleagues.

Even when the counsellors agreed to participate, it was difficult to remind them to recruit clients when they under intense workload pressure. In addition, there was no control over client recruitment, so counsellors may have been more likely to choose clients and sessions they felt comfortable with.

**7.4.2. Study strengths and limitations**

This exploratory work provides a rare window into the content of three genetic counselling sessions. The study shows that when the teaching model of genetic counselling dominates, counselling that addresses the psychosocial and cultural needs of the client is hindered.

A number of studies have examined the independent components of the counselling experience. Lippman-Hand *et al.* (1979b) analysed transcripts of 30 tape recorded genetic counselling sessions focussing on decision making about reproduction. Her study also included post-counselling interviews with counsellors. In another study, qualitative interviews were used to examine clients’ impressions of their video recorded counselling session (Evans *et al.*, 2004). The data obtained on their experiences of the interaction and how they perceived the importance of cultural difference, provided a window into the clients’ side of the interaction.

Later, Aalfs *et al.* (2007) attempted to establish the factors that enhance client satisfaction using pre- and post-session questionnaires for both client and
counsellor. In a study of familial cancer clinics, Lobb et al. (2005) also used pre- and post-consultation questionnaires to explore whether counsellors vary their communication style in response to client factors. Other than the current study, only one study has analysed transcripts of genetic counselling sessions and explored the counsellor and clients’ experiences of the interaction (Gale et al., 2010).

Many studies of actual genetic counselling sessions have been limited by small numbers of participants and this study was no exception. There are low numbers of practitioners at each genetics unit and the logistics of conducting cross-institutional observation studies significantly limits the opportunity to undertake large scale studies (Roter et al., 2006). The small numbers in this study meant that the sessions analysed represented only a small sample of the practitioners in this unit (seven of the 12 clinical staff) and the sessions did not capture the wide cross section and type of clients and conditions counselled in the department. The small number of possible participants meant that combining clinical geneticists and genetic counsellors in one study was a major limitation. Whilst these two types of GHPs do perform genetic counselling as such, they receive different types of training and have different roles to perform.

This exploration of the counselling session content is inevitably influenced by my position as a genetic counsellor and colleague of the health professionals that participated in the study. Attempting to remain objective and ensure reflexivity may not have been sufficient. An independent researcher may have added a different interpretation and perspective, and the addition of a data auditor may have overcome these limitations. The use of audio recordings rather than video recordings did not allow for the assessment of visual cues and non-verbal communication.
7.4.3. Clinical implications and future research

Despite an increased awareness of the importance of psychosocial issues, this study suggests genetic counselling may still be tethered to the teaching and medical models, possibly due to factors such as lack of confidence, service protocols and other institutional barriers. A deeper understanding of these factors might enlighten training and professional development and facilitate overcoming the barriers to applying more dialogical client-centred approaches to genetic counselling.

As a gap in our knowledge about the genetic counselling interaction from the perspective of the client exists, future research could be directed towards further evaluation of the needs of clients with respect to cultural issues and the extent to which clients have their needs met. Further work in this area is needed to address this side of the interaction, which is equally as important as the perspective of the counsellor. From the counsellors’ perspective, an idea of whether they are able to assess their client’s cultural needs and the ways in which they do this, along with the development of additional tools to help them assess cultural difference and health literacy, will be beneficial. Continued use of qualitative research methods in genetic counselling will further our understanding of the processes involved and continue to inform developments and improvements in practice.

7.5. Summary

Despite the challenges, this exploratory study of the communication between GHPs and clients provided unique findings. Examining the genetic counselling interaction from the perspective of both counsellor and client, the experiences of both were assessed through the analysis of session transcripts and post-session questionnaires completed by both parties.

The clients in this study thought that any perceived cultural differences seemed unimportant in comparison to the counsellor’s experience and demeanour,
including style, knowledge and empathy. The GHPs displayed techniques such as developing rapport, identifying issues and drawing out client’s concerns, recognising their uncertainty about a particular issue or fact and enabling clients to feel safe and heard. Genetic counsellors are trained in this area and because they are able to draw on their skills of reflection, education and counselling, they are in a position to overcome some barriers created by cultural difference. This is reflected in the sessions recorded in this study and supports the argument that genetic counsellors do not need to develop expertise in a particular area or culture, but rather be encouraged to develop their skills in viewing each client as a unique individual.

The transcripts of the counselling sessions showed a predominance of a unilateral monologue style consistent with the teaching model. There was, however, some evidence of an interactive style using conversational strategies and recognising cultural aspects. The predominance of the teaching model is perhaps partly due to the fact that some clients prefer this style and come to genetic counselling seeking information rather than psychosocial support. Counsellors should treat each client as unique and not assume they all present with psychosocial issues. The crucial step is in up-skilling counsellors so they can confidently assess prior knowledge, health literacy, information and support needs, without making prior assumptions about clients. The post-session questionnaires showed that this model of practice is desired by clients and leads to feelings of engagement with genetic counselling.
8. The challenges of dialogical engagement

Future challenges to the field of genetic counseling will not be in the mastery of new and complex clinical content; counselors have already demonstrated their abilities to excel in this very important aspect of practice. The challenges are the same as those faced by our medical colleagues – to listen more and speak less, to engage and empower clients, and to be emotionally present when they are needed. (Roter et al., 2006, p.219)
8.1. Inside genetic counselling

The practice of genetic counselling has evolved from existing within the medical model that is formulated around educating clients, into a much broader framework focussing on counselling rather than educating (McCarthy Veach et al., 2010). The development of counselling practice has been driven to some extent by modern technology, the increasing complexity of genetic information and clients’ ability to obtain information via the internet. Genetics, and life in general, is complex and becoming increasingly so. Clients expect more information and discussion, and in some cases are more psychologically aware and amenable to the discussion of feelings and emotions (Shiloh et al., 2006b). Rather than “doing what the doctor tells them”, they want to be involved in discussion of options and decision making. The challenge for the genetic counselling profession is to maintain the ability to adapt and develop models of practice that enable counsellors to engage with diversity in today’s complex world.

Calls for the inclusion of a psychosocial component in genetic counselling which includes ethnocultural issues moves the practice of genetic counselling further away from the paternalistic medical educational model towards a more inclusive and dialogical counselling model (Weil, 2003). The dialogical model conceptualises genetic counselling as a two-way interaction between counsellor and client, exploring issues relating to individuals’ and families’ experiences with inherited conditions (McCarthy Veach et al., 2007). This dialogical model can enhance the counselling process by allowing the counsellor to be more open and responsive to each individual client without excluding education of the client. Rather, this model allows information and education to be provided based on client needs rather than counsellor assumptions. The client should feel a greater ownership of the process of counselling and hence feel that their needs are being met.
A review of genetic counselling models in Chapter Two showed that more effective cross-cultural counselling could be provided by improving genetic counsellors’ resourcefulness in dealing with cultural diversity. Rather than training counsellors to work with specific ethnic groups (assuming homogeneity in such groups), adopting Weil’s (2003) bidirectional approach can move counselling forward from seeing clients as members of a discrete group stereotype, towards a true interaction where every client and interaction is seen as a new event. Approaching each client as an individual allows counselling sessions to focus on the client’s story, beliefs, emotions and dilemma. Assuming that no interaction between a counsellor and client is exactly the same, and by improving genetic counsellors’ attentiveness to the uniqueness of each client, the genetic counselling session should better meet the client’s needs.

A more dialogical, patient-centred approach to counselling and improved cross-cultural competency enables more effective engagement in counselling sessions because it focuses on the interaction between counsellor and client (Wiggins et al., 2013). The counsellor spends more time listening to the client’s story and concerns, communicates more effectively and is more likely to satisfy the client’s needs and expectations. This mode of practice is more aligned with the narrative approach where the counsellor learns about the client who has a ‘story’ to tell about their lived experience with a genetic condition that is influenced by their understanding, emotional response and their own unique cultural background (Glessner et al., 2012; Hodgson et al., 2013). The genetic counsellor is also a unique individual and how they respond to the client will depend on experiences in their personal and professional life and their own cultural background. They are equipped with skills and tools to listen, explain complex terms and act as an interpreter, advocate and ally for the client and their family. Resta (2002, p.20) confirms that “different counsellors will bring their own strengths, personality, and psychological baggage to counseling sessions.”
In contrast, genetic counselling following the educational model may stifle such dialogue, as the interaction is set up with the expert (the counsellor) imparting information to the listener/learner (the client). While a certain amount of information must be covered in order for informed decisions to be made, adequate attention must be given to the client’s agenda and concerns. This avoids the problems of mismatch between the client’s and counsellor’s expectations and may improve client satisfaction, shared decision making, and adaptation.

8.2. Summary of the tasks in this thesis

The overall approach of this thesis was to explore what is required to move towards a more dialogical model of genetic counselling. I used genetic counselling of consanguineous couples to examine diversity in a broader context than usually recognised, and to explore how genetic counsellors engage with cultural diversity in their practice. Throughout the thesis I have reflected on ways in which the genetic counselling profession has developed in response to increasing cultural complexity and burgeoning knowledge in the field of medical genetics.

Chapter Two examined the evolution of the genetic counselling profession. Various models of practice were evaluated and compared with current Australian practice. Although most of the literature is derived from the American experience, some parallels can be drawn. Current models of genetic counselling practice recognise the relevance of both genetic counsellors’ and their clients’ cultural background. However, there is a lack of information about how genetic counsellors perceive cultural difference and how they engage with diversity in their practice.

The review of the literature on consanguinity in Chapter Three revealed many studies addressing the genetic implications and socio-economic and cultural importance of consanguinity in communities where this is a common type of union. However, few studies were identified that addressed genetic counselling for cousin couples from backgrounds where cousin marriage is not traditionally practised,
including western cultures. The lack of research in this area suggests genetic counsellors need to reflect more critically on their assumptions about consanguineous marriage within a western secular culture.

As any genetic counselling interaction involves contributions from both the client and counsellor, the perspectives of both are important. In Chapter Five I first looked at the experiences of a group of genetic counselling clients, their presenting issue and how genetic counselling had impacted on their experiences. In Chapter Six, I looked at counselling process from the genetic counsellors’ perspective and the influence of the views that they bring with them to the interaction. Finally, in Chapter Seven I explored the genetic counsellor-client interaction by examining communication during three live genetic counselling sessions. This chapter synthesises these research findings, discusses them in relation to the dialogical model of genetic counselling and examines the implications of these findings for current practice, training of future practitioners and further research.

8.3. Research findings

8.3.1. Lived experiences of consanguineous couples

The questionnaire and interview data generated in this study has provided insight into the lived experience of consanguineous couples in Australia, and their experiences of genetic counselling. The difficulties faced by cousin couples, the impact of society’s closed views on cousin marriage and the effect these views have on couples was explored for the first time. The idea that cousin marriage is bad or dangerous is sometimes attributed to the perceived high risk of birth defects and society’s views of incest (Abrams et al., 2002; Bishop et al., 2008; Hamamy, 2012). These views are reflected in the persistence of negative comments, jokes about inbreeding, taboos and prejudice.
Recruiting consanguineous couples for the study was difficult. Most participants were concerned about confidentiality and a desire to avoid publicity, possibly because of their experience of society’s closed views of consanguinity. The media were helpful in identifying potential participants who had not been for genetic counselling, although they consistently requested that I ask these couples to volunteer for media interviews. This would have been unethical and therefore all requests were denied.

The cohort of consanguineous couples recruited for the study was not from certain ethnic groups or cultures where consanguinity is a culturally-based practice. Owing to the heterogeneity of the participants, their experiences cannot be considered representative of other consanguineous couples.

Many of these couples experienced strained family relationships and difficulty defining their relationship to mainstream society, resulting in concerns about the genetic risk becoming a secondary issue in some cases.

The qualitative data obtained from the questionnaires and interviews challenges the assumption that genetic risk is the cause for greatest concern in cousin couples. Genetic risk appears to be more of a concern for GHPs and wider society, and may underlie the closed views of consanguinity. Nevertheless, even though genetic risk was not the clients’ greatest concern, accurate information about genetic risk was reassuring for many. Others, with a family history of a genetic condition, were not so reassured and in some cases guilt and bad feelings about the relationship was amplified by diagnosis of a genetic condition. These results suggest that more attention needs to be focussed on the psychosocial issues for all couples in consanguineous unions, irrespective of whether they are from ethnic groups where this is common practice. Referral for psychotherapy may also be required for ongoing and more difficult problems. As part of this therapy, the consanguineous
relationship needs to be considered within the wider system of the whole family, including consideration of the complexity introduced by having shared relatives.

An anticipated outcome of the present study was that a better understanding of the lived experiences of WA consanguineous couples would be used to develop guidelines for genetic counselling and structuring of a service that addresses some of the issues faced by couples. An unexpected outcome was that many of the couples benefited from participating in the study. For some, it was the first time they became aware that there were other consanguineous couples and for others the interviews provided an opportunity to normalise their concerns and beliefs. This suggests there is potential to reduce the isolation felt by consanguineous couples and inform health professional and wider society about their circumstances in a more positive way.

The consanguineous couples participating in this study came from a variety of backgrounds and sought genetic counselling for a variety of reasons. In terms of explaining genetic information and risk, and offering psychosocial support, working with these clients differs little from working with non-consanguineous clients. However, the results of this study suggest that additional factors need to be taken into account, such as community misunderstanding of consanguineous relationships, differences in world views and values (in relation to marriage or human rights), and family dynamics.

8.3.2. Genetic counsellors and diversity

Chapter Six explored genetic counsellors’ ideas about cultural difference. The genetic counsellors in this study appeared to have narrow views about cultural difference that focused on the more obvious and visible characteristics such as race, ethnic background, language and religious affiliation. The counsellors also appeared to lack flexibility around session content and the allocation of time to explore a client’s cultural background, and little attention was given to the influence of their
own background in the interaction. There is evidence that the views of cultural
difference have broadened with the passage of time and there is increased
awareness of the importance of cultural competence. Further training and
professional development may be useful in assisting counsellors to feel comfortable
in identifying diversity issues.

The findings from the questionnaire data on genetic counsellors’ opinions about
consanguinity showed that in general they were open minded and nondirective in
their interaction with consanguineous couples. A small number of genetic
counsellors considered first cousin and uncle niece unions to be incestuous
relationships, reflecting the discomfort around consanguinity amongst people from
cultures where cousin marriage is not common. Whether this affected their ability
to provide unbiased support for their clients in practice was not measured.

The genetic counsellors in this study did not recognise consanguinity as a diversity
issue, reflecting the general view that consanguinity is a practice confined to
particular ethnic groups and religions (Teeuw et al., 2013). Discussions about
consanguinity were restricted to information giving about genetic risks and
screening options, with limited or no opportunity to explore the psychosocial
impact of being in a cousin couple relationship. This shows the dominance of the
education model in the profession and highlights the need to pay more attention to
the counselling model and seeing the client as an individual.

In terms of how the genetic counsellors managed cultural difference in their
counselling sessions, they appeared to modify how they delivered information and
the tools they used to deliver this information. For example they were aware of the
need to use interpreters where language differences were evident, they attempted
to assess the client’s prior knowledge before giving information, and they used tools
such as simple language and diagrams to aid their explanations. Many of these
approaches, however, focussed again on information giving. Other important issues
such as basic literacy, health literacy and maintaining a focus on the client’s agenda also need to be addressed within the counselling session.

8.3.3. Inside the ‘black box’

An increase in the number of qualitative studies of the genetic counselling process has recently provided a window into the ‘black box’ of genetic counselling. Chapter Seven of this study explored the complex dialogical interaction that occurs in genetic counselling sessions, providing further insight into the genetic counselling session, how counsellors interpret information and how their approach influences how the counselling session progresses.

The post-session questionnaires in this study addressed both client and counsellor perspectives of what occurs in a counselling session, specifically how cultural differences impact on the session. The analysis of genetic counselling session transcripts and the post-session questionnaires provides unique data because counsellors and clients were asked about their experiences of the session. Whether the needs of both parties were met, and if any perceived cultural differences impacted the interaction, were also addressed. Whilst several studies have utilised genetic counselling session transcripts, there is limited data on the content of the transcripts and the client(s) and counsellor impressions of those sessions.

Other studies have shown the continued dominance of the teaching model with GHPs dominating the conversation and drawing the client back to their own agenda. The influence of the teaching model was also evident in this limited exploratory study. The GHPs attempted to engage with clients by building rapport and addressing the clients’ concerns. They did not appear to engage with diversity issues such as difference in socio-economic status, gender and health literacy. Despite this, the clients felt that having common cultural background and diversity per se was not as important as the effect of the GHPs demeanour, how they interacted with
them, whether the GHPs were able to identify what was important to them and their family, and whether the GHP adhered to their own agenda.

The post-session questionnaires revealed that there was often a mismatch between the clients and GHP regarding what actually occurred in the sessions and how much time was spent on various activities. The clients placed little importance on cultural conformity or difference. In contrast, the GHPs commented that client demeanour and opinions affected their ability to engage and develop rapport. In the main, the GHPs’ approach to culturally diverse clients was to ignore the sources of difference and continue on with their own ‘routine’ information to be imparted. Only occasionally did they enquire about issues of importance to the client and in some cases modified their session and use of language. This may be because they were more comfortable with what they knew well and believed this was required to obtain informed consent for genetic testing and informed decision making. Some suggestions for overcoming the restrictions of this model of practice will be discussed in the next section.

8.3.4. Enhancing dialogical models: recognising the individual

Each genetic counselling interaction is unique, as both clients and counsellors bring their own belief, value and cultural systems, interwoven with every life experience, to a counselling session. In this complex and unique environment, counsellors need the skills to explain complex genetic information to clients who may have very different levels of general and health literacy. They also need to take into account psychological and or cultural factors (such as guilt, grief, anxiety and familial or cultural beliefs about patterns of inheritance or causes of genetic conditions) that might affect a client’s ability to assimilate this information. They need to think broadly about cultural difference and be adaptable in order to deal with each unique client and interaction encountered.
The findings from this study indicated that the dialogical model of counselling theoretically fits with clients’ desires for a counsellor who understands them, relates to them as an individual and tailors information and discussion of issues to their individual needs. Although most counsellors embraced the idea of diversity and dialogical counselling, there appeared to be a lag between the conceptual acceptance of the model and the ability to implement it in practice. This could be due to a number of reasons including, but not limited to, organisational constraints, level of experience, lack of skill or confidence with applying a more dialogical model, and inadequate training in implementing the model. Other possibilities include a lack of awareness of the importance of acknowledging diversity, a lack of understanding of what constitutes difference, and a failure to recognise how psychological and cultural factors influence the integration of genetic information.

Overall, the GHPs were good at establishing rapport and interacting with clients, matching communication styles and explaining complex information. It appears to be the lack of flexibility with which they perform these tasks and the limited flexibility they have in running their sessions that restrains them from operating in a more dialogical manner. The inclusion of psychosocial factors in counselling sessions was evident in some of the recorded interactions, but constraints on time, institutional or practical factors appeared to hinder maintenance of this focus. Counselling and communication techniques such as listening, reflection, assessing comprehension and using shorter speaking turns appeared to be underutilised.

The main work of genetic counselling is communication (about genetic information, assessment of ideas, thoughts emotions and feelings) to facilitate coping and decision making (Resta et al., 2006). The failure of GHPs to recognise the impact of psychological and cultural factors, as evident in this study, may threaten basic communication and may prevent them from carrying out their role effectively. Counselling cannot be truly dialogical unless clients have adequate levels of health literacy and are equipped to understand the information presented to them. Health
literacy embodies the goals of clear health communication and patient engagement in healthcare, requiring a diverse worldview and multidisciplinary perspective.

The advantage of a more dialogical model is that the relationship between client and counsellor is seen as a mutual process and engagement is reciprocal. This leads logically to viewing each interaction as a relationship between different individuals. I believe that a more dialogical model of practice is favourable and can be implemented into practice in a busy clinic. However, for effective implementation there needs to be further assistance for genetic counsellors to move beyond the constraints of the concept of ND and the medical model. The shift from being nondirective to being more engaged is a subtle one and in practice genetic counsellors need to be able to operate within some assumptions but be flexible enough to identify, and adjust to, the cues of cultural difference as they arise.

Being open and transparent about values and cultural frameworks is concordant with the view that it is not possible to be truly nondirective. In practice in the genetic counselling session, this requires genetic counsellors to be aware that both they and their clients come from particular positions, rather than neutral ones. Counsellors who are able to recognise these values and positions will be better placed to explicitly address differences as they are identified. Rather than ignoring the ‘elephant in the room’, by keeping up the pretence of ND and adopting the ‘safe’ position of focussing on information, client and counsellor differences can be acknowledged and addressed.

In this way, dealing with each situation and relating to clients as individuals with unique experiences will lead to the application of more dialogical models by genetic counsellors. By recognising and acknowledging the diversity in all clients and focussing on the core of the REM, the relationship between counsellor and client will improve. With this awareness, the counsellors may step beyond ND and identify factors which may be blocking a mutual understanding or open communication with
their client. In such a session, counsellors could openly acknowledge any differences that they themselves bring to the meeting and take time to explore (without making assumptions) what their clients might bring with them.

In an effort to extend the dialogical models reviewed in Chapter Two, I propose the enhancement of the REM with the addition of ‘INDIVIDUAL’ (Figure 8.1). This could be a useful tool to assist counsellors to conceptualise the contribution of all individual characteristics of both client and counsellor.

![Figure 8.1 REM adaptation](image)

8.3.5. Implications for cultural diversity training

Genetic counselling is a unique profession which has begun to examine its own processes. Further research and discussion provides an opportunity to examine,
critically analyse and suggest improvements in training, philosophy and processes. In addition to creating our own norms and ideas, we often draw from expertise in other professions. However, rather than simply using literature on transcultural training from nursing, psychology and counselling, we need to look more broadly at how engaging with diversity relates to our practice and how we can develop counsellors’ skills in managing the increasing complexity this brings.

This study has shown that counsellors feel they are not adequately trained to work effectively with cultural difference and the skills they develop are through their own motivation or ‘on-the-job’ experience. This suggests a need for a more coordinated approach to educating genetic counsellors, both the training of students and professional development for practising counsellors.

Some authors recommend training counsellors to specialise in counselling clients from particular cultures or religions (Wang, 2001). While this approach may have some merit for reducing access and language barriers for some clients, my view is that this is too narrow a focus, as it devalues people as individuals and limits the ability of every genetic counsellor to achieve true cultural competence. Categorising people is problematic, as it risks stereotyping and minimises the impact of complex interacting variables. Furthermore, given the relatively homogenous make-up of genetic counsellors in Australia, matching counsellor and client backgrounds would be difficult to achieve. In the situation where genetic counselling needs to be provided for conditions associated with particular ethnic or religious groups, it is important for counsellors to have some knowledge of the cultural norms in these communities. There are alternative ways to improve service provision and skills in engaging with diversity for all practitioners in these situations. The published information about cultural norms and beliefs of these different groups could be used as “handbooks” for genetic counsellors working with these clients (Wiggins et al., 2013). Other alternatives to improve service provision include increasing genetic counsellors’ awareness of the importance of transcultural dialogue, developing skills
in eliciting their client’s narrative accounts and a greater investment in applying models with more dialogical engagement. These skills can be applied by all genetic counsellors to all counselling interactions and represent true transcultural expertise (Wiggins et al., 2013).

### 8.4. Recommendations

A series of recommendations, informed by the findings from this thesis research, are proposed below as being worthy of further consideration.

#### 8.4.1. Training and professional development

- Include education and discussion about the dialogical models of practice and how they can be applied in practice in Master of Genetic Counselling courses.
- Increase exposure to diversity issues in Master of Genetic Counselling courses and accreditation processes.
- Facilitate continued development of skills that enable the counsellor to listen to the client’s ‘story’, address the issues that are significant for the client, and enhance their ability to adapt the language and style to meet the needs of each individual.
- Provide continuing education for genetic counsellors to improve their understanding of the impact of their own cultural background on their clients.
- Encourage exploration during professional supervision of how to recognise and respond to cultural difference and dissonance.

#### 8.4.2. Improved engagement with diversity issues

- Provide workshops or group discussions for genetic counsellors on diversity issues and the many different sources of diversity, to allow for in-depth discussions with colleagues.
- Develop tools and guidelines for exploring issues of cultural difference and obtaining background information about our clients. For example, Warren (2010) produced an
online toolkit for genetic counsellors with the aim of improving the delivery of culturally responsive, client-centred genetic counselling. It contains self-assessment quizzes and clinical tools to help counsellors assess client cultural and spiritual background.

- Continue to apply broader definitions of diversity to raise awareness that every client differs from the genetic counsellor in some way and that both clients and counsellors are unique.

- Raise awareness that diversity exists, is normal, and enriching diversity is important.

- Consider the terminology and language used when describing diversity and culture. Changes to terminology may avoid some negative stereotypical associations.

- Consider opportunities for mentoring that could result in both mentor and protégé learning about diversity together. Pairing genetic counsellors from minority groups with mentors from majority groups and vice versa may be useful for facilitating a shared understanding of the needs of each group.

- Provide group training models or workshops at conferences that encourage interaction between genetic counsellors and individuals from URMs, to enable counsellors to better understand the needs of clients from these minority groups.

8.4.3. Improved consanguinity counselling

- Increase awareness that consanguineous couples are not limited to certain ethnic groups.

- Facilitate education and awareness of consanguinity in society for health professionals.

- Broaden the training in the area of consanguinity to focus on a wider set of issues than genetic risk, particularly for couples from backgrounds where consanguinity is not an accepted practice.

- Reduce the isolation people in consanguineous relationships can feel by facilitating contact with other couples through websites and support groups such as the Association of Genetic Support of Australasia (AGSA).
- Continue to draw attention to psychological and socio-cultural factors (in addition to risk and genetics) during genetic counselling sessions.

- Provide counsellors with training on how to empower consanguineous couples with skills and tools to counter resistance and negative attitudes from family and friends. Develop a list of strategies to help cousin couples manage family challenges.

- Lobby the ASGC to develop a set of guidelines on genetic counselling for consanguineous couples (similar to the NSGC guidelines), but broaden the focus to include psychosocial issues.

8.4.4. Further research

A number of areas worthy of further research have been identified in this study. In particular, studies of why the education model continues to persist are needed as well as studies that explore ways to enable the profession to implement a more dialogical approach in counselling sessions.

The development of research methods or guidelines to assist genetic counsellors to explore their own cultural identity and possible prejudices would also be beneficial. At present, the value of doing this is recognised (Lewis, 2002), but there appears to be insufficient guidance, and few tools available, on how to explore this cultural identity and prejudice. In trying to develop a suitable tool, it may be useful to consider the scales used in other professions, such as the Multicultural Counseling Knowledge and Awareness scale. This scale has been successfully applied to assess the relationship between supervisors of genetic counsellors’ perceived levels of multicultural competence and their development as supervisors, along with their ability to evaluate their students’ multicultural skills (Lee et al., 2009).

The qualitative component of this case study on consanguineous couples was structured around a set of questions that allowed clients to tell their ‘story’ about their experiences with genetic counselling and being in a relationship with their cousin. Future research could explore the issues from a narrative approach eliciting
further personal stories and experiences. The terminology used around consanguinity or cousin marriage could also be explored, in particular whether alternative terms might contribute to breaking down the stigma associated with these relationships. Research in this area could draw on the opinions of clients, genetic counsellors and other health professionals.

Further work with consanguineous couples could also be directed towards examining the strategies that cousin couples use to counter societal stigma associated with their relationship. More research on the lived experience of the client using narrative approaches could also be undertaken. Asking cousin couples to tell their story in either video or written formats would give rich data and has the potential to elucidate factors to inform genetic counselling practice. Research could be used to help genetic counsellors explore what is important for consanguineous couples in the moment of genetic counselling. For example, how do consanguineous couples talk to their children about their relationship and what terminology do they use?

Underlying improved practice in genetic counselling is better training and further research could focus on improving the training of genetic counselling students and providing professional development opportunities for practising counsellors. A study could be undertaken to compare a pre-enrolment assessment of students and practising counsellors’ ideas on cultural difference. A student might respond more intuitively while an experienced genetic counsellor’s response might attempt to “present themselves in a socially desirable light” (Gu et al., 2011; Lee et al., 2009, p.297). Research could explore how genetic counsellors actually counsel consanguineous couples, whether they explore the psychosocial and cultural issues around consanguinity with clients, and effectively communicate the goals of the counselling session, and assess whether or not these goals have been met. Discussion and focus groups for genetic counsellors could be developed in each state, to compare and contrast the experiences of geographically diverse groups of
genetic counsellors, and to address the challenges that arise when working with culturally diverse clients.

8.5. Conclusion

Genetic counselling continues to use the education model as the primary practice model, despite growing awareness of the importance of psychosocial factors. While it is expected that there will be a time lag between acceptance of newer, more dialogical models of genetic counselling and their implementation, in this study I have identified a number of gaps in the practices of genetic counsellors in Australia that are preventing them from fully implementing these more dialogical models.

Transcultural genetic counselling has expanded our horizons and we are becoming increasingly aware of broader diversity issues. Whilst I agree that it is important to learn about the client’s cultural background and to be aware of the cultural differences, I feel this is too limited and risks the dangers of stereotyping. I disagree with the concept that to be an effective cross-cultural counsellor you need to be of a similar background to the client. The findings in Chapter Six show that clients prefer engagement with a counsellor, rather than simply sharing a similar background with their counsellor.

Developing genetic counsellors’ understanding of, and ability to engage with, the dialogical models of practice will empower them to explore each client’s story, learn about each unique individual and tailor their interactions accordingly. This study has contributed to knowledge on the process of genetic counselling and the impact of these processes on one of the most important parties in the interaction – the client. This study contributes to knowledge about consanguinity, the lived experience of couples in consanguineous relationships, genetic counsellors’ views of consanguinity and clients’ perceptions of genetic counselling.
Consanguinity is not usually considered to be a contributor to diversity, but is a practice that can be based strongly on cultural beliefs and norms, is not commonly practised in most western countries and has stigma associated with it. We need to reconsider our conception of consanguinity and cultural difference, particularly given that consanguineous couples are not always from cultures where it is a common practice. There is a need to look at the consanguineous relationship in relation to culture, family systems and wider society. This is the reason why it is a particular area of practice that can add to our knowledge and understanding of transcultural counselling.

Over the course of my candidature there has been a progression towards a transcultural model of genetic counselling but I am convinced further development is required. This thesis research can stimulate discussion about the future of the profession, course content for graduate students and continuing professional development for experienced counsellors. As an established profession, we need to have the confidence to move beyond the medical model and promote the importance of engaging with diversity if we are to deliver a client-centred service in an increasingly complex world.
9. References


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### Appendix A: Questionnaire – people in a consanguineous relationship

#### CONSANGUINITY QUESTIONNAIRE

**Code:**

**Project title:** Social, Psychological and Ethical Issues in Genetic Counselling of Consanguineous Couples in WA.

**INSTRUCTIONS:** Where choices are given please circle the one which best describes you.

### General Information

1. **Gender**
   - Male
   - Female

2. **Highest level of education completed**
   - Primary
   - Secondary
   - TAFE
   - Tertiary
   - Postgraduate
   - Other

3a. **Your Country of Birth:**

3b. **Mother’s country of Birth:**
    - Father’s country of birth:

4. **Date of Birth:**

5a. **Languages spoken:**

5b. **Language spoken at home:**

6. **Religious Affiliation**
   - Protestant
   - Catholic
   - Other Christian
   - Buddhist
   - Muslim
   - Jewish
   - No Religion
   - Other (Please specify)

7. **How are you and your partner/spouse related?**
   - 1st Cousins
   - 2nd Cousins
   - Uncle/Niece
   - Other (Please specify)

8. **Are you and your partner married?:**
   - Yes
   - No
   - If not married what is your relationship? (e.g.: de facto, boyfriend etc)

9. **How old were you when the relationship with your partner/spouse began?**

10. **How old was your partner/spouse when the relationship began?**

11. **How many children do you and your partner/spouse have?**

12. **How many children would you and your partner/spouse like to have?**

13. **How many pregnancies have you had with your partner/spouse?**

14. **Have you and your partner/spouse had any miscarriages?**
   - Yes
   - No
   - How many?

15. **Have you and your partner/spouse had any stillborn babies?**
   - Yes
   - No
   - How many?

16. **How important is having children to you?**
   - Very Important
   - Important
   - Not very important
   - Not important at all
   - What are the reasons?

### Consanguinity (Couples who are related)

17. **Does your immediate family know you are in a relationship with your relative?**
   - Yes
   - No

18. **Which of the following best describes your family’s reaction to your relationship?**
   - Strongly support
   - Support
   - No reaction
   - Oppose
   - Strongly oppose
   - Comments:

19. **Do people outside your family know you and your partner are related?**
   - Yes
   - No

20. **Which of the following best describes their reaction to your relationship?**
   - Strongly support
   - Support
   - No reaction
   - Oppose
   - Strongly oppose
   - Comments: 

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21. Describe how you met your relative and how your relationship began? ...........................................
...........................................................................................................................................................

22. Is there anyone else in your family whose partner/spouse is a relative? Yes No
If Yes, who: ........................................................................................................................................
How are they related to their partner/spouse? ........................................................................................

23. List four issues that are positive about relationships with relatives.
(1).................................................................................................................................................
(2).................................................................................................................................................
(3).................................................................................................................................................
(4).................................................................................................................................................

24. List four factors that are negative about relationships with relatives.
(1)....................................................................................................................................................
(2)....................................................................................................................................................
(3)....................................................................................................................................................
(4)....................................................................................................................................................

25. Should relationships with relatives be: Encouraged Neither Discouraged

26. Which of the following would best describe your reaction if your child wanted a relationship/marriage with a relative?
Strongly support Support No reaction Oppose Strongly oppose

Genetics

27. Have you any children with a genetic condition or birth defects?
No Go to Question 30 Yes Go to next question

28. What is the name of the condition/birth defect? .............................................................

29. What do you think is the cause of this condition/birth defect? .............................

30. Do you think that the risk of you and your partner/spouse having a baby with a birth defect is:
Much lower than other couples Lower than other couples The same as other couples Much higher than other couples

31. If the risk of any couple having a baby with a birth defect is 4%, what is the risk for a couple who are first cousins?
0-2½% 2½-5% 5-7½% 7½-10% 10-20% >20% Don’t know

32. Do you feel this risk is? High Moderate Low Don’t know

33a. How worried are you that you might have a baby with a birth defect?
Very worried Somewhat worried Not very worried Not worried at all

33b. What are the reasons you feel like this?
.......................................................................................................................................................... 

10. What do you believe are the causes of birth defects? ........................................................
..........................................................................................................................................................

Genetic Counselling

11. Have you and/or your partner/spouse ever had genetic counselling?
Yes Continue on next Q No Go to Question 46

12. Who referred you for genetic counselling?
13. Why did you go to genetic counselling?

14. What did you expect to get out of genetic counselling? (you can circle more than one)
   Information  Advice  Reassurance  Encouragement

15. Did the counsellor tell you what to do regarding marriage to your relative? Yes  No
   If yes what advice was given?

16. Did you find genetic counselling:
   Very unhelpful  Unhelpful  Neither  Helpful  Very helpful

17. What did you find most helpful about genetic counselling?

18. What was unhelpful about the genetic counselling?

43a. Was the counsellor from the same cultural background as you? Yes  No  Don’t know
43b. Would you have preferred a counsellor from the same religion or ethnic background as you? Definitely  Maybe  Not sure  Maybe not  Definitely not

44. After counselling how did you feel about your relationship with your relative?
   Much better  Better  The same  Worse  Much worse

45. After counselling how did you feel about having children?
   Much better  Better  The same  Worse  Much worse

46. Did you know genetic counselling was available? Yes  No
47. Would you have liked to have genetic counselling before beginning the relationship with your relative? Definitely  Probably  Possibly  No  Not sure

THIS PAGE WILL BE DETACHED FROM THE COMPLETED SURVEY IMMEDIATELY ON RECEIPT

Please return completed questionnaire to:

Helen Mountain
Associate Genetic Counsellor
Genetic Services of WA, King Edward Memorial Hospital, 374 Bagot Rd SUBIACO WA 6008

Thank you for taking the time to complete this questionnaire.
Feedback will be available to all participants.

YES  [ ]  NO  [ ]
I am willing to participate further by completing a face to face interview.
Name ____________________________
Date ______________________________
Telephone _________________________

I do not wish to be contacted to participate in any further research

YES  [ ]  NO  [ ]
I wish to be contacted by a genetic counsellor to arrange an appointment
Name ____________________________
Date ______________________________
Telephone _________________________

I do not wish to arrange an appointment for genetic counselling.
Appendix B: Press release – recruiting people in a consanguineous relationship

Relationships between cousins and other relatives are more common than most people think. They just don’t talk about it because people feel uncomfortable talking about a subject that is seen by some to be socially taboo.

Jokes about inbreeding perpetuated on the internet, prejudice and misinformed gossip often force Cousin Couples into a world of their own.

The reality is that lots of communities accept and encourage cousin marriage and “the chances that children of first cousins will be born with a genetic disease are NOT significantly higher than for unrelated couples”. (Journal of Genetic Counselling, April 2002).

For the first time a comprehensive study of Cousin Couples is being undertaken by a Perth PHD student from Murdoch University.

Helen Mountain is looking for volunteers to share their experiences in a supportive confidential environment. “I want to encourage open discussion about these relationships and promote a better understanding in the community”.

“Volunteers will be encouraged to share their experiences, beliefs and worries, in confidential questionnaires” she said.

“Once we have a better understanding of the issues involved we can improve genetic counselling and support services where necessary and direct health care resources where needed.”

Anyone who is in, or has had, a relationship with a relative is invited to contact Helen on 9340 1525. The study involves completing a short written survey and all information collected will be kept strictly confidential. The Murdoch University and King Edward Hospital Ethics committees have approved the study.
Appendix C: Letter of invitation to participate – opt-in

GENETIC SERVICES OF WESTERN AUSTRALIA

King Edward Memorial Hospital for Women
374 Bagot Road, SUBIACO WA 6008
Telephone: (08) 9340 1525
Facsimile: (08) 9340 1678

Princess Margaret Hospital for Children
Roberts Road, SUBIACO WA 6008
Telephone: (08) 9340 8828
Facsimile: (08) 9340 7058

DATE
NAME
ADDRESS

Dear

I am writing to ask if you would be willing to be contacted by a researcher based at King Edward Hospital for Women and Murdoch University, Ms Helen Mountain (Genetic Counsellor). Helen is undertaking a project for her PhD studies ‘Social, psychological and ethical issues in genetic counselling of consanguineous couples in WA’ and wishes to recruit suitable former patients from this hospital. Clients from this hospital are required for this study and from our records you would appear to be a potential participant for the study.

The purpose of the study is to gain a better understanding of relationships between relatives from a personal and societal point of view. Open discussion of experiences, beliefs and worries may assist us to provide better genetic counselling services. By giving a service that clients require health care resource allocation can be improved.

If you wish to hear more about this study, or be contacted further, could you please complete the tear-off slip at the bottom of this letter and return it in the envelope provided. If we hear from you within 6 weeks you will be contacted by the researcher, Helen to give you more information about the study. Should you wish to have further information about the study before making a decision as to whether or not you wish to participate, please telephone Helen on (08) 9340 1525 at Genetic Services King Edward Hospital.

The King Edward Memorial and Princess Margaret Hospitals Ethics Committee and Murdoch University Ethics Committee have approved the study, and the confidentiality of all participants is assured. The information gathered from the study may be published; however, names or any other identifying information will not be used. Whether or not you participate in this project, any future care you receive at this hospital will not be affected in any way.

Thank you for considering this request.

Yours sincerely

Assoc. Prof. Jack Goldblatt
Director
Genetic Service of WA
PLEASE TICK ONE BOX, THEN SIGN DATE AND RETURN THE SLIP IN THE ENVELOPE PROVIDED to:

Helen Mountain, Genetic Counsellor,
Genetic Services of WA,
King Edward Memorial Hospital for Women,
374 Bagot Road, Subiaco, WA 6008.

I wish to be contacted and to participate in the study ‘Social, psychological and ethical issues in genetic counselling of consanguineous couples in WA’.

☐

I would like further information on the study ‘Social, psychological and ethical issues in genetic counselling of consanguineous couples in WA’ so that I can decide whether or not to participate.

☐

Name: __________________________________________________________

Date: ________________________________

Signature: _____________________________________________________

Telephone Numbers: ____________________________________________
Appendix D: Letter of invitation to participate – opt-out

GENETIC SERVICES OF WESTERN AUSTRALIA
King Edward Memorial Hospital for Women
374 Bagot Road, SUBIACO WA 6008
Telephone: (08) 9340 1525
Facsimile: (08) 9340 1678

Princess Margaret Hospital for Children
Roberts Road, SUBIACO WA 6008
Telephone: (08) 9340 8928
Facsimile: (08) 9340 7058

DATE
NAME
ADDRESS

Dear

I am writing to ask if you would be willing to be contacted by a researcher based at King Edward Hospital for Women and Murdoch University, Ms Helen Mountain (Genetic Counsellor). Helen is undertaking a project for her PhD studies ‘Social, psychological and ethical issues in genetic counselling of consanguineous couples in WA’ and wishes to recruit suitable former patients from this hospital. Clients from this hospital are required for this study and from our records you would appear to be a potential participant for the study.

The purpose of the study is to gain a better understanding of relationships between relatives from a personal and societal point of view. Open discussion of experiences, beliefs and worries may assist us to provide better genetic counselling services. By giving a service that clients require health care resource allocation can be improved.

If you do not wish to hear more about this study, or be contacted further, could you please complete the tear-off slip at the bottom of this letter and return it in the envelope provided. If we do not hear from you within 3 weeks we will assume you are willing to be contacted and the researcher, Helen Mountain, will contact you shortly afterwards to give you more information about the study. Should you wish to have further information about the study before making a decision as to whether or not you wish to be contacted, please telephone Helen on (08) 9340 1525 at Genetic Services King Edward Hospital.

The King Edward Memorial and Princess Margaret Hospitals Ethics Committee and Murdoch University Ethics Committee have approved the study, and the confidentiality of all participants is assured. The information gathered from the study may be published; however, names or any other identifying information will not be used. Whether or not you participate in this project, any future care you receive at this hospital will not be affected in any way.

Thank you for considering this request.

Yours sincerely

Assoc. Prof. Jack Goldblatt
Director, Genetic Service of WA
PLEASE TICK ONE BOX, THEN SIGN DATE AND RETURN THE SLIP IN THE ENVELOPE PROVIDED to:

Helen Mountain, Genetic Counsellor,
Genetic Services of WA,
King Edward Memorial Hospital for Women,
374 Bagot Road, Subiaco, WA 6008.

I do not wish to be contacted and do not wish to participate in the study ‘Social, psychological and ethical issues in genetic counselling of consanguineous couples in WA’.

☐

I would like further information on the study ‘Social, psychological and ethical issues in genetic counselling of consanguineous couples in WA’ so that I can decide whether or not to participate.

☐

Name: ____________________________

Date: ____________________________

Signature: ____________________________

IF WE DO NOT HEAR FROM YOU WITHIN THREE (3) WEEKS WE WILL ASSUME YOU ARE WILLING TO PARTICIPATE IN THE STUDY.
Appendix E: Information and consent form – consanguinity

| School/Division Title: Institute of Science & Technology Policy, School of Social Science, Humanities & Education |
| Project title: Social, Psychological & Ethical Issues in Genetic Counselling of Consanguineous Couples in Western Australia |

Information for Participants.

I am a Murdoch University Ph.D. student (under the supervision of Dr. Ian Barns) and a Genetic Counsellor at Genetic Services of Western Australia (GSWA). I am investigating genetic counselling for couples who marry a relative (e.g. first cousin). The purpose of this study is to gain a better understanding of relationships between relatives from a personal and societal point of view. Open discussion of experiences, beliefs and worries may assist us to provide better genetic counselling services. By giving a service clients require health care resource allocation can be improved.

You can help with this study by agreeing to complete a survey. The time taken to complete the survey is expected to take no longer than 30 minutes. The survey contains written questions about your background and religion, family and experiences of genetic counselling, which may be seen as personal and private. After completing the survey you will be asked if you wish to continue your involvement with the study by completing an interview, conducted by Helen Mountain, where you will be asked more in-depth questions about the same issues. This interview will be recorded.

All information given will remain CONFIDENTIAL and your privacy will be respected at all times. No names or other identifying information will be used in any publication arising from the research. You can decide to withdraw from the study at any time. Participants that request feedback on the study will be sent a newsletter summarising the results obtained.

If you wish to participate in this study, please complete the attached form and return it together with your completed questionnaire in the envelope provided. If you have any questions about this project please contact either myself, Helen Mountain on 93401625 or my supervisor, Dr Ian Barns on 93602895. We are happy to discuss any concerns you may have on how the study is conducted. Alternatively you can contact Murdoch University’s Human Research Ethics Committee on 93606677.

Declaration:

I (the participant) have read the information above and any questions I have asked have been answered to my satisfaction. I agree to take part in this study; however, I know that I can change my mind and stop at any time. If I decide to participate in an interview I agree for this to be taped.

I understand that all information provided is treated as confidential and will not be released by the investigator unless required to do so by law.

I agree that research data gathered may be published provided my name or any information that might identify me is not used.

PARTICIPANT’S NAME: ______________________________________________________

SIGNATURE: ___________________________________ DATE: ______________

INVESTIGATOR: ___________________________________________________________

SIGNATURE: ___________________________________ DATE: ______________
Appendix F: Interviews schedule – people in a consanguineous relationship

Background information
1. Occupation – you and partner
2. Religious affiliation
3. Describe your cultural background
4. Relationship status (married engaged etc.)
5. Description of family (e.g. large small close etc.)
6. Children of the relationship
7. Health of the children
8. History of genetic disease in the family?

Questions about Cousin Marriage
9. Tell me about how the relationship with your cousin began?
10. Did you suppress your feelings for your cousin when you first met?
11. What influence did your family have on your decision to be with your cousin?
12. Have you discussed your relationship with your family?
13. Describe their response when they learnt of your relationship?
14. What was that like for you?
15. Have you discussed your relationship with your cousin with friends?
16. Is there anyone you would like to have discussed this with but have felt unable to?
17. Why do you avoid telling people about your relationship?
18. What is this like for you?
19. Have you explained your relationship to your children?
20. How do you feel about telling your children?
21. Can you tell me how you would feel about your child having a relationship with their cousin?
22. Why do you feel this way?
23. Do you feel there is a taboo against cousin marriage in Australia?
24. What impact have these taboos had on your life?
25. Do you believe cousin marriages are legal?
26. Do you feel there should be legislation about cousin marriage?
27. Do you know any other cousin couples?
28. What have the positive things about your relationship been?
29. What have the negative things about your relationship been?

Questions about genetic counselling
30. Have you had genetic counselling – if so when?
31. Tell me about your reasons for seeking genetic counselling
32. What was going to genetic counselling like for you?
33. Can you tell me about the genetic counsellor that you saw?
34. Do you think genetic counselling should be recommended for cousin couples?
35. How do you feel about your risk of having children with genetic conditions?

Feedback/Finish
36. Is there anything else you think is important in understanding cousin marriage that has not already been covered?
Appendix G: Questionnaire – genetic counsellors

GENETIC COUNSELLOR QUESTIONNAIRE

Project title: Social, Psychological and Ethical Issues in Genetic Counselling of Consanguineous Couples in Western Australia.

Throughout this questionnaire where choices are given circle the most appropriate response

Demographics

1. Gender: Male Female

2. Highest level of education completed:
   Secondary   TAFE   Tertiary Postgraduate   Other

3a. Country of Birth: ……………………

3b. Family’s nationality …………………………………..

4a. Languages spoken: ………………………………………………………………………………………..………..

4b. Primary language spoken at home: ………………………………………………………………………….

5. Religious Affiliation: Protestant Catholic Other Christian
   Buddhist Muslim Jewish No Religion Other (Specify)……

6. Professional Background: Science Nursing Social Work
   Psychology Other (Please specify) …………………………………………………………………………………

7a. In which state/territory do you practice? …………………………

7b. Is your practice: metropolitan outreach other (specify) ……………………………..

8. Do you regularly counsel any specific cultural/ethnic groups? No Yes
   If YES (specify)… ………………………………………………………………………………………………………

Cultural Difference

9. Is encouraging cultural difference within our society beneficial?
   Strongly agree Agree No opinion Disagree Strongly disagree
   Comments: ……………………………………………………………………………………………………………
   ……………………………………………………………………………………………………………………………
   ……………………………………………………………………………………………………………………………

10a. What do you perceive as possible advantages in having a multicultural society?
   …………………………………………………………………………………………………………………………………..
   …………………………………………………………………………………………………………………………………..
   …………………………………………………………………………………………………………………………………..

10b. What do you perceive as possible disadvantages in having a multicultural society?
   …………………………………………………………………………………………………………………………………..
   …………………………………………………………………………………………………………………………………..
   …………………………………………………………………………………………………………………………………..

11. What are the four most important factors that contribute to cultural difference?
    (1) ……………………………………………………………………………………………………………………………..
    (2) ……………………………………………………………………………………………………………………………..
    (3) ……………………………………………………………………………………………………………………………..
    (4) ……………………………………………………………………………………………………………………………..

12. Does cultural difference affect the genetic counselling interaction? Yes No
    If YES – What effect does it have? …………………………………………………………………………….
    …………………………………………………………………………………………………………………………………..
    …………………………………………………………………………………………………………………………………..

13. Has your genetic counselling training equipped you to deal with cultural differences between you and your clients? Completely Somewhat Slightly Not at all
Comments?

14. Do you think there are any differences (manner, approach, reception by clients etc) between male and female genetic counsellors? Yes No Unsure Don’t know
Comments

Genetic Counselling

15. How important is it to be aware of the cultural and social background of clients? Essential Important Undecided Not very important Unimportant
Comments

16. What techniques and “tools” do you use to explain complex genetic information to “non-experts”? Comments

17. How do you judge whether the client understands the concepts you are explaining?
Comments

18. To what extent has your training equipped you to explain complex genetic principles to the lay person? Completely Mostly Somewhat Not at all
Comments

Use the following descriptors to answer the following 4 questions
A = Always F = Frequently S = Sometimes N = Never

19. Do you assess your client’s prior knowledge about genetics and genetic testing? A F S N If so specify how?

20. Do you modify your counselling session to suit your clients’ background knowledge? A F S N If so specify how?

21. Do you modify your counselling session to suit your clients’ cultural background? A F S N If so specify how?

22. Do you modify your counselling session if using an interpreter? A F S N If so specify how?
Consanguinity

23. How many consanguineous couples have you counselled in the past year? ..............
24. How does this compare to previous years? More Less Same
25. If you DO NOT counsel consanguineous couples state why? ........................................

26. Should consanguineous relationships be: Strongly encouraged Encouraged
Neither Discouraged Strongly Discouraged
Comments:........................................................................................................................................

27. Rank from 1-4 how the importance of these factors may influence your feelings about consanguineous relationships.
Religion ..... Genetic risk .... Legislation .... Ethnic background ........
State any other factors that you think might influence your feelings ..................

28. Do you think consanguineous relationships are more common in particular ethnic groups? No Yes Please specify which groups ........................................

29. What is the general population risk of birth defects?
0-2½% 2½-5% 5-7½% 7½-10% 10-20% >20% Don’t know
30. What is your impression of this risk? Very High High Medium Low Very Low
31. What is the risk of birth defects for first cousins?
0-2½% 2½-5% 5-7½% 7½-10% 10-20% >20% Don’t know
32. What is your impression of this risk? Very High High Medium Low Very Low
33. What advice/information would you give to a consanguineous couple planning a pregnancy?.................................................................................................................................

34. What do you think are the 4 most important issues for relatives who have a relationship/marry?
(1) .........................................................................................................................................................
(2) .........................................................................................................................................................
(3) .........................................................................................................................................................
(4) .........................................................................................................................................................

35. Rank (from 1-4) the relationships that you regard as incestuous?
(1) .........................................................................................................................................................
(2) .........................................................................................................................................................
(3) .........................................................................................................................................................
(4) .........................................................................................................................................................

<table>
<thead>
<tr>
<th>Please return completed questionnaire to:</th>
<th>Your valuable time and assistance is appreciated.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Helen Mountain</td>
<td>This study forms part of Helen’s Ph.D. project</td>
</tr>
<tr>
<td>Associate Genetic Counsellor</td>
<td>at Murdoch University, Western Australia.</td>
</tr>
<tr>
<td>Genetic Services of WA</td>
<td></td>
</tr>
<tr>
<td>374 Bagot Rd SUBIACO WA 6008</td>
<td></td>
</tr>
</tbody>
</table>
Appendix H: Information and consent form – genetic counsellors

**Project title:** Social, Psychological and Ethical Issues in Genetic Counselling of Consanguineous Couples in Western Australia.

**Division Title:** Institute of Science and Technology Policy, Murdoch University.

Dear Colleague,

I am a Murdoch University Ph.D. student and Associate Genetic Counsellor at Genetic Services of Western Australia (GSWA). I am investigating genetic counselling for consanguineous couples under the supervision of Dr Ian Barns. The purpose of this study is to gain a better understanding of relationships between relatives from a personal and societal point of view. Open discussion of experiences, beliefs and concerns may assist us to provide better genetic counselling services. By giving a service clients require health care resource allocation can be improved and professional development undertaken.

You can assist me with this study by agreeing to complete a survey. The time taken to complete the survey will vary, but it is expected to take no longer than 30 minutes. The survey contains written questions about your background; personal opinions and genetic counselling practice. At a later date further work in the form of focus groups or a workshop may be carried out.

All information given will remain CONFIDENTIAL and your privacy will be respected at all times. No names or other identifying information will be used in any publication arising from the research. You can decide to withdraw from the study at any time. Feedback on the study can be provided in “Linkage” or at National conferences.

If you have any questions about this project please feel free to contact either, Helen Mountain on 08 9340 1525, email helen.mountain@health.wa.gov.au, my supervisor, Dr Ian Barns on 08 9360 2895. We are happy to discuss any concerns you have on how this study is being conducted, alternatively you can contact Murdoch University’s Committee on 08 9360 6677.

**Declaration:**

I (the participant) have read the information above and any questions I have asked have been answered to my satisfaction. I agree to take part in this study; however, I know that I can change my mind and stop at any time.

I understand that all information provided is treated as confidential and will not be released by the investigator unless required to do so by law.

I agree that research data gathered may be published provided my name or any information that might identify me is not used.

**PARTICIPANT’S NAME:** __________________________________________________________

**SIGNATURE:** ___________________________   **DATE:** ______________

**INVESTIGATOR:** __________________________________________________________

**SIGNATURE:** ___________________________   **DATE:** ______________
Appendix I: Discussion group program

Introduction (5 min)

Warm-up exercise (5 min)

Tell me about yourself and your cultural background
OR Give me 10 answers to the question: “Who am I?”

Brainstorming (5 min)

What is the first thing that comes to mind when you think about consanguinity?

Specific questions (30 min)

What are the limitations of genetic counselling with regard to managing cultural difference?
What resources do you need to manage cultural difference better?
Ideas for future research in the area

Personal experiences/ideas/issues that raise concern (15 min)
Appendix J: Memorandum – invitation to GSWA genetic counsellors and geneticists

GENETIC SERVICES OF WESTERN AUSTRALIA

King Edward Memorial Hospital for Women
374 Bagot Road, SUBIACO WA 6008
Telephone:  (08) 9340 1525
Facsimile:  (08) 9340 1678

Princess Margaret Hospital for Children
Roberts Road, SUBIACO WA 6008
Telephone:  (08) 9340 8828
Facsimile:  (08) 9340 7058

Memorandum

DATE: ..........................................................................................................................

TO: GSWA Colleagues ..................................................................................................

FROM: Helen Mountain .................................................................................................

SUBJECT: Further research for my PhD studies. ..........................................................

Dear Name,

I would like to determine levels of interest in participation in the next phase of my research project on the effect of cultural difference on the genetic counselling interaction, in order to determine whether it is worth my while commencing the arduous exercise of obtaining ethics approval!

My aim is to record and transcribe 6 to 10 actual genetic counselling sessions, and assess with the use of a pre- and post-session questionnaire. Using quantitative techniques I will look at issues such as client expectations and whether these are met, whether the cultural background of client (and counsellor) and counsellor style affect outcomes and the general experience of the participants. It goes without saying that all tapes and transcripts would be confidential and de-identified and participants would be included as co-authors on any resulting publications.

Please would you indicate below if you would be happy to have one/any sessions recorded and return this memo to me. Also any constructive comments or suggestions on this aspect of my research are gratefully accepted. If you have any questions or concerns about the process feel free to ask.

Thank you, Helen.

☐ YES. I would be willing to participate.

☐ NO. I do not wish to participate.

COMMENTS: ..............................................................................................................

.................................................................................................................................

.................................................................................................................................

.................................................................................................................................

.................................................................................................................................

.................................................................................................................................
Appendix K: Information and consent form – genetic counselling sessions

**School/Division Title:** Institute of Science and Technology Policy, School of Social Science, Humanities and Education

**Project title:** Social, Psychological and Ethical Issues in Genetic Counselling of Consanguineous Couples in Western Australia.

**Information for Participants.**

I am a Murdoch University PhD. student (under the supervision of Dr Ian Barns) and a Genetic Counsellor at Genetic Services of Western Australia (GSWA). I am investigating the effect of cultural differences on the interaction between clients and their genetic counsellor/geneticist. The purpose of this study is to gain a better understanding of how genetic counselling works and the effect of subtle or obvious differences between the two parties. By gaining a better appreciation of the process and what makes it a positive experience for clients and professionals we aim to deliver a better service and thus improved health care resource allocation.

You can help with this study by agreeing to have your genetic counselling session digitally recorded. The content of the session will be transcribed anonymously into written words and analysed using recognised data analysis techniques. You will also be asked to complete a short written questionnaire after the end of the session. The time taken to complete this will vary, but it is expected to take no longer than 20 minutes. The survey contains written questions about your experiences of genetic counselling. Your decision to participate (or not) will in no way affect your ongoing care at Genetic Services of WA or King Edward Hospital.

All information given will remain CONFIDENTIAL and your privacy will be respected at all times. No names or other identifying information will be used in any publication arising from the research. You can decide to withdraw from the study at any time.

If you are willing to participate in this study, please notify your genetic counsellor/geneticist prior to your appointment. You will also need to bring your completed consent form to your appointment. If you have any questions about this project please feel free to contact either myself, Helen Mountain on 93401525 or my supervisor, Dr Ian Barns on 93602895. We are happy to discuss any concerns you may have on how this study is being conducted, or alternatively you can contact Murdoch University’s Human Research Ethics Committee on 93606677.

******************************************************************************

**Declaration:**

I (the participant) have read the information above and any questions I have asked have been answered to my satisfaction. I agree to take part in this study; however, I know that I can change my mind and stop at any time.

I understand that all information provided is treated as confidential and will not be released by the investigator unless required to do so by law. I agree that research data gathered may be published provided my name or any information that might identify me is not used.

I consent to my genetic counselling session to be recorded and transcribed by Helen Mountain.

PARTICIPANT’S NAME: _______________________________________________________

SIGNATURE: ___________________ DATE: ___________________

INVESTIGATOR: ___________________________________________________________

SIGNATURE: ___________________ DATE: ___________________
Appendix L: Post-session questionnaire – genetic health professionals

POST-COUNSELLING SESSION QUESTIONNAIRE
Geneticist/ Genetic Counsellor

Client’s Initials: 

Your Name: 

1. What did you perceive as the client’s main reason for seeking genetic counselling? What were they most worried about?

________________________________________________________________________
________________________________________________________________________
________________________________________________________________________
________________________________________________________________________

2. What were the four most important issues that you wanted to cover in the session?

1) __________________________________________
2) __________________________________________
3) __________________________________________
4) __________________________________________

3. Divide the “pie charts” into sections to show how much of the session was spent on:

A: Giving factual information.
B: Eliciting what worried the client
C: The client talking about their feelings
D: Exploring their personal or family experience of genetic conditions.

A: You listening
B: You talking

4. List any other issues time was spent on during the session:

________________________________________________________________________
________________________________________________________________________
________________________________________________________________________
________________________________________________________________________

5. Mark the box that best describes how you felt after the session:
1) My understanding of the client’s situation was:

Excellent □ Satisfactory □ Average □ Unsatisfactory □ Poor □

Comments?: ______________________________________________________
__________________________________________________________________
__________________________________________________________________

2) Communication between the client and I was:

Excellent □ Satisfactory □ Average □ Unsatisfactory □ Poor □

Comments?: ______________________________________________________
__________________________________________________________________
__________________________________________________________________

3) I felt that client’s understanding of the information given was:

Excellent □ Satisfactory □ Average □ Unsatisfactory □ Poor □

Comments?: ______________________________________________________
__________________________________________________________________
__________________________________________________________________

4) At the end of the session my feeling of overall satisfaction was:

Excellent □ Satisfactory □ Average □ Unsatisfactory □ Poor □

Comments?: ______________________________________________________
__________________________________________________________________
__________________________________________________________________
__________________________________________________________________

6. Did you feel that you and the client developed a good rapport?
__________________________________________________________________
__________________________________________________________________
__________________________________________________________________

7. Was there anything about the client that made a negative impression on you?
__________________________________________________________________
__________________________________________________________________
__________________________________________________________________
__________________________________________________________________

8. Did you feel there was anything that hindered communication between you and the client?

YES □ NO □

If yes please specify:_________________________________________________
__________________________________________________________________
__________________________________________________________________
9. How would you describe your background?

____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

10. What impression did you have about the client’s background?

____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

11. Did you think they share a similar background to you (i.e.: did you feel you had things in common or not)?

YES ☐ NO ☐

Comments?: __________________________________________________________
____________________________________________________________________
____________________________________________________________________

12. Do you think the client would have preferred you to be from a similar background to them?

YES ☐ NO ☐

Comments?: __________________________________________________________
____________________________________________________________________
____________________________________________________________________

13. Any other thoughts or comments about the session?

____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

THANKYOU VERY MUCH FOR YOUR TIME

Please return the questionnaire in the envelope provided to Helen Mountain
Appendix M: Post-session questionnaire – clients

POST-COUNSELLING SESSION QUESTIONNAIRE
Genetic Counselling Client

Counsellor/Geneticist: ____________
Your Name: ____________

1. What was your main reason for seeking genetic counselling? What were you most worried about? _______________________________________________________
_____________________________________________________________________
_____________________________________________________________________
_____________________________________________________________________

2. What were the four most important issues that you wanted to cover in the session?
1) ________________________________________________________________
2) ________________________________________________________________
3) ________________________________________________________________
4) ________________________________________________________________

3. Divide the “pie charts” into sections to show how much of the session was spent on:

<table>
<thead>
<tr>
<th>A: Being given factual information.</th>
<th>B: Being asked about what worried you</th>
</tr>
</thead>
<tbody>
<tr>
<td>C: Being able to talk about your feelings</td>
<td>D: Talking about your personal or family experience of genetic conditions.</td>
</tr>
</tbody>
</table>

A: You listening
B: You talking

4. List any other things time was spent on during the session: ______________________
_________________________________________________________________________
_________________________________________________________________________

5. Mark the box that best describes how you felt after the session:

5.1 The counsellor’s understanding of my situation was:

Excellent □ Satisfactory □ Average □ Unsatisfactory □ Poor □

Comments ______________________________________________________________
_________________________________________________________________________
5.2 Communication between the counsellor and I was:

Excellent [ ] Satisfactory [ ] Average [ ] Unsatisfactory [ ] Poor [ ]

Comments ______________________________________________________
_________________________________________________________________

5.3 I felt that my understanding of the information given to me was:

Excellent [ ] Satisfactory [ ] Average [ ] Unsatisfactory [ ] Poor [ ]

Comments ______________________________________________________
_________________________________________________________________

5.4 At the end of the session my feeling of overall satisfaction was:

Excellent [ ] Satisfactory [ ] Average [ ] Unsatisfactory [ ] Poor [ ]

Comments ______________________________________________________
_________________________________________________________________

6. What was it about the counsellor that made a positive impression on you?
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

7. Was there anything about the counsellor that made a negative impression on you?
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

8. Did you feel there was anything that hindered communication between you and the counsellor?
   YES [ ] NO [ ]

   If Yes please specify: ________________________________
   ___________________________________________________
   ___________________________________________________

9. How would you describe your background?
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

10. What impression did you have about the counsellor’s background?
____________________________________________________________________
11. Did you think the counsellor shared a similar background to you (i.e.: did you feel you had things in common or not)?

YES □ NO □

Comments

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

12. Would you have preferred the counsellor to be from a similar background to you?

YES □ NO □

Comments

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

13. Any other thoughts or comments about your experience of genetic counselling?

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

_________________________________________________________________________

THANKYOU VERY MUCH FOR YOUR TIME

Please return the questionnaire in the envelope provided to Helen Mountain