Jumping Genes Drive Evolution

BY KEITH OLIVER AND WAYNE GREENE

Orthodox evolutionary theory does not tally with the fossil record, but a new school of thought points towards “jumping genes” as essential agents of periodic changes in the rate of evolution.

Current evolutionary thought is dominated by an assumption that biological lineages evolve through the slow and gradual accumulation of adaptive gene mutations. However, this does not match up with most of the fossil record. Instead, new species arise abruptly and periodically, and there are intermittent and often long periods when very little happens – a situation called evolutionary stasis.

Our evolutionary hypothesis, which we call “Transposon Thrust”, states that significant evolution cannot take place without the activity of “jumping genes”, which are more formally known as transposons or transposable elements. Discovered by Barbara McClintock in the 1950s, they are so-named because of their capacity to jump (or copy themselves) from one position to another in the DNA of an organism. In the 1980s, jumping genes, which are almost universally abundant in genomes, were written off as parasitic, junk or selfish DNA that we would be better off without.

However, ever-increasing evidence over the past decade has begun to turn this idea on its head, with many studies revealing that jumping genes can generate genetic changes of great variety and magnitude. As with other types of mutations, a proportion of the DNA changes caused by jumping genes will, by chance, be beneficial and be positively selected in evolution. Of course they can also cause harm, but jumping genes are only a minor source of known genetic disease, causing just over 0.5% of genetic diseases in humans. This short-term cost to a very small number of individuals is massively outweighed by the longer-term benefits to the evolution of the lineage. Because they promote adaptability, jumping genes are extremely useful, if not essential, genomic parasites. This is not to say that jumping genes are the only cause of evolution, but that they are hugely important and powerfully complement other processes such as:

• point mutations, where the wrong DNA bases are inserted at particular locations;
• horizontal transfer, where one organism transfers genes to another organism that is not its progeny; and
• polyploidy, where an organism ends up with more than the usual two copies of the genome.

Jumping genes can create useful genetic change, the raw material upon which natural selection acts, in two basic ways. First, they can operate in an active fashion, either by inserting into new locations of the genome to seed new genes or parts of genes, or by inadvertently copying and pasting existing genes or parts of genes from one location to another. Such activity tends to be transient since jumping genes become inactive as they succumb to the effects of random mutation over time.

Nevertheless, the mere presence of large numbers of inactive, but similar, jumping gene relics can secondarily cause genetic changes in a passive fashion. This is because they create a “hall of mirrors” – a plethora of virtually identical sites within the genome – which promotes major reorganisations of DNA by confusing the cellular machinery involved in its propagation. This can result in genes or parts of genes being either duplicated or lost altogether. The loss of genes is not always disadvantageous, but if it is then there will be selection against the affected individuals.

In their active mode, even small numbers of jumping genes will have a great impact on their host genome, and high activity is likely to reoccur with every new invasion of jumping genes into a lineage. New invasions can occur either by horizontal transfer, such as through viruses or bacteria, or by the natural origination of jumping gene activity from within a genome.

By contrast, to have significant passive effects on a genome, near-identical copies of jumping gene relics must be present in great numbers. This is the situation in humans and other primates, whose
genomes are comprised of jumping gene relics of two major varieties. These are the so-called *LINE-1* and *Alu* elements, which in the human genome are present in a whopping 500,000 and 1.1 million copies, respectively.

A central tenet of our Transposon Thrust hypothesis is that lineages that have active jumping genes, or large populations of the same type of jumping gene acting passively, are adaptable and spawn new species readily. Conversely, species whose genomes are deficient in jumping genes, or which possess a great mixture of different types of jumping gene relics, tend to undergo evolutionary stasis and may risk extinctions as they lack the capacity to adapt and diversify.

Transposon Thrust can provide answers to six key mysteries in evolutionary biology.

1. **Why do species appear suddenly in the fossil record?**

New species appear suddenly because jumping genes can cause major genetic changes in a lineage rather rapidly, rather than gradually. They do this by creating new genes, altering the control switches of existing genes or rearranging chromosomes. These large changes are thought to be the major means by which new species-specific traits evolve, and a significant number of them cannot be caused in any other way.

2. **What is the cause of punctuated equilibrium?**

Punctuated equilibrium is rapid evolution followed by slow evolution, or a stoppage in evolution, as is observed in the fossil record. This can be explained by the fact that jumping gene activity does not occur at a low and uniform rate over time. Instead, it sporadically occurs in sudden bursts resulting in rapid evolution, followed by decreasing activity and slowing evolution. These rapid bursts of evolution can happen when a new type of jumping gene is suddenly transferred into a lineage from some other lineage, or when a new type of jumping gene naturally emerges from within a genome. Jumping gene activity can also increase as a response to stress, temporarily increasing the rate of evolution. Successive waves of jumping gene activity thus account for alternating periods of rapid evolution and stasis, and can thereby reconcile evolutionary theory with palaeontology and the fossil record.

3. **Why are some lineages of organisms species-rich and others species-poor?**

Species-rich lineages, which among the mammals include rodents, bats and primates, have had successive bursts of jumping gene activity over evolutionary time, extending into recent times or to the present. Species-poor lineages, such as the primate cousins known as flying lemurs, have not had recent bursts of activity but probably had them in the very distant past. Such waves of activity may also help to explain why certain other groups of animals are particularly diverse, such as the songbirds, which account for 40% of all bird species, and the perciform (perch-like) fish, which account for 40% of all fish species, although there is insufficient data to verify this at present.

4. **Why do living fossil species change little over millions of years while other lineages evolve rapidly?**

Living fossils such as the lobe-finned coelacanth fish and the reptilian tuatara of New Zealand have remained virtually unchanged for 410 and 220 million years, respectively. As examples of evolutionary stasis, these fossil species appear to have had no new infiltrations of jumping genes, except in the very distant past. What little jumping gene relics they do possess are in low numbers and/or are very diverse, leaving little scope for passive effects either. As a result, they are effectively frozen in time. In contrast, most lineages of mammals have evolved rapidly following the extinction of the dinosaurs 65 million years ago.
5. Why do species have differing controls on jumping genes in reproductive cell DNA compared with ordinary somatic cell DNA?

Jumping gene activity in “somatic” body cells is heavily restricted by a number of mechanisms, including a chemical modification to jumping gene DNA called methylation. But these controls are temporarily relaxed in the DNA of reproductive cells (e.g. sperm, eggs and early embryos), which creates a window of opportunity to allow some jumping gene activity.

This difference between these two cell types can be explained by the fact that genetic changes caused by jumping gene activity in somatic cell DNA cannot benefit the lineage because they can’t be passed on to the next generation. Rather, they can be damaging to individuals if mutations lead to cancer, for example.

By contrast, jumping gene activity in reproductive cell DNA can create valuable genetic variation that can be inherited and that natural selection can work on. Thus, successful lineages from single-celled protozoa right through to mammals specifically permit jumping gene activity in reproductive cells for the potential benefit of future generations, and strictly minimise it in somatic cells where it is potentially harmful to the individual.

6. Why do almost all species only suppress jumping genes rather than eliminating them?

Although the types and total amount of jumping genes vary greatly between different groups of organisms, they often comprise a large, if not massive, fraction of the genome. Known mammalian genomes are at least one-third jumping gene DNA in origin, while plant genomes often have an even higher jumping gene DNA content of over two-thirds.

It has long been a puzzle as to why many species tolerate having so much of this so-called junk, parasitic or selfish DNA within their genomes. Our answer is that any species that eliminates its jumping genes cripples its evolutionary potential and greatly increases its chances of extinction, so it is not beneficial for it to do this. It is far better for a species to suppress jumping genes in somatic cells while allowing them some activity in reproductive cells in order to promote evolvability, at a cost to a small number of individuals in terms of inherited genetic disorders.

**CONCLUSION**

Compelling evidence now indicates that jumping genes have had a major role in evolution as irreplaceable sources of novel genetic changes. Far from being parasites or junk, jumping genes have made their host genomes flexible and dynamic so that the genomes themselves can promote their own evolution.

Their legacy is astounding, ranging from the creation (and sometimes destruction) of genes to the genome-wide seeding of gene control switches and wholesale rearrangement of chromosomes. Periodic bursts of jumping gene activity not only predict punctuated equilibrium as a general characteristic of evolution, but provide an explanation as to how some lineages are able to spectacularly diversify while others are liable to evolutionary stasis.

As more data becomes available in the future on jumping genes and their contribution to the genomes of a wide range of species, awareness of their pivotal role in evolution should also grow.

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