

'Jumping genes' repeatedly form new genes over evolution

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In the same way that Lego pieces can be arranged in new ways to build a variety of structures, genetic elements can be mixed and matched to create new genes, according to new research.

A long-proposed mechanism for creating [genes](#), called exon shuffling, works by shuffling functional blocks of DNA sequences into new genes

that express proteins.

A study, "Recurrent Evolution of Vertebrate Transcription Factors by Transposase Capture," published Feb. 19 in *Science*, investigates how genetic elements called transposons, or "jumping genes," are added into the mix during evolution to assemble new genes through exon shuffling.

Transposons, first discovered in the 1940s by Cornell alum and Nobel Prize-winner Barbara McClintock '23, M.A. '25, Ph.D. '27, are abundant components of genomes—they make up half of human DNA—and have the ability to hop and replicate selfishly in the [genome](#). Some transposons contain their own genes that code for enzymes called transposase proteins, which cut and paste genetic material from one chromosomal location to another.

The study, which focused on tetrapods (four-limbed vertebrates), is important because it shows that transposons represent an important force in the creation of [new genes](#) during evolution. The work also explains how genes critical for human development were born.

"We think it's very likely this mechanism may extend beyond vertebrates and could be more of a fundamental mechanism that occurs in non-vertebrates as well," said first author Rachel Cosby, Ph.D. '19, a postdoctoral researcher at the National Institutes of Health. Cosby is a former graduate student in the lab of senior author Cedric Feschotte, professor in the Department of Molecular Biology and Genetics in the College of Agriculture and Life Sciences.

"You are putting the bricks in in a different way and you construct a whole new thing," Feschotte said. "We are looking at the question of how genes are born. The originality is that we are looking at the role of transposons in creating proteins with novel function in evolution."

In the study, the researchers first mined existing databases for genomes of tetrapods, because genomes for more than 500 species have been fully sequenced. Cosby and colleagues searched for combinations of DNA sequences known to be characteristic of transposons fused to host sequences to find good candidates for study. They then chose genes that evolved relatively recently—within tens of millions of years ago—so they could trace the history of the gene's development through the vertebrate tree of life.

Though genes fused with these transposases are relatively rare, the researchers found them all over the vertebrate tree of life. The researchers identified more than 100 distinct genes fused with transposases born in the past 350 million years along different species lineages, including genes in birds, reptiles, frogs, bats and koalas, and a total of 44 genes born this way in the human genome.

Cosby and colleagues selected four recently evolved genes and performed a wide range of experiments in cell culture to understand their functions. They found the proteins derived from these genes are able to bind to specific DNA sequences and turn off gene expression. Such genes are known as transcription factors and act as master regulator genes for development and basic physiology. One such gene, PAX6, is well studied, plays a key role as a master regulator in the formation of eyes in all animals and is highly conserved throughout evolution.

"If you put a PAX6 gene from a mouse into a *Drosophila* [fruit fly], it works," Feschotte said. Though others have proposed before that PAX6 is derived from a transposase fusion, the researchers in this study further validated the hypothesis.

Cosby and colleagues isolated one of these recently evolved genes in bats, called KRABINER, and then used CRISPR gene-editing technology to delete it from the bat genome and see what genes were

affected, before adding it back in. The experiment revealed that when KRABINER was removed, hundreds of genes were dysregulated, and when they restored it, normal functioning returned. The protein expressed by the KRABINER gene bound to other related transposons in the bat genome, Cosby said.

"The experiment revealed that it controls a large network of other genes wired through the past dispersion of related transposons throughout the bat genome—creating not just a gene but what is known as a gene regulatory network," Feschotte said.

More information: Rachel L. Cosby et al. Recurrent evolution of vertebrate transcription factors by transposase capture, *Science* (2021). [DOI: 10.1126/science.abc6405](https://doi.org/10.1126/science.abc6405)

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