

How One Enzyme has Created a Third of the Human Genome

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Only about one percent of the human genome contains what we recognize as protein-coding genes: DNA sequences that are transcribed into RNA sequences and then translated into proteins. Much of the intervening space between genes consists of mobile DNA sequences, known as transposable elements, which have the ability to “copy and paste” themselves throughout the genome.

One type of transposable element are retrotransposons, so named because they are formed by converting RNA back into DNA through a reverse transcription process. Among the most prevalent of these, comprising nearly a third of the human genome, is a sequence called the Long Interspersed Element-1 (L1) retrotransposon. Like the Starbucks of the genome, an L1 appears on every corner—but how exactly it manages to replicate and spread so efficiently has long remained unclear.

Now, using advanced protein microscopy techniques, Damon Runyon Fellow Akanksha Thawani, PhD, and her colleagues at the University of California, Berkeley, have shed light on L1’s mechanism of proliferation, potentially offering a roadmap for the engineering of new gene therapies.

The team examined the protein encoded by the L1 sequence, an enzyme called ORF2. This enzyme, the researchers found, functions as “molecular scissors,” snipping the DNA strand (see illustration) in order to prime reverse transcription of itself back into DNA.

To get a more detailed picture of this mechanism of action, the team reenacted the process with lab-made target DNA and template RNA. By testing different scenarios, they identified the specific type of DNA site that ORF2 requires to make its cut.

Just as CRISPR therapies arose from the discovery of a naturally occurring genome editing system in bacteria, these insights into ORF2’s mechanism of genome editing may inform the development of new gene therapies for cancer. If we can harness the power of these “scissors” to make precise cuts and insertions, in other words, we could induce more desirable changes than another chain restaurant on the block.

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