

Alzheimer's could be triggered by 'genomic mosaicism'

Certain inherited genetic mutations lead to Alzheimer's disease (AD), but they are relatively rare. A [recent study](#) from my laboratory, however, shows that gene alterations that are not passed along by one's parents may also play a key role in triggering the disease. This happens as a result of a process that occurs in the cell nucleus, known as gene recombination (GR), which can make changes to the DNA "blueprint" in human neurons.

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We discovered that DNA sequences vary from cell to cell, meaning that our brains are a vast mosaic of distinct genomes, a phenomenon aptly referred to as "genomic mosaicism."

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Taking all this into account led us to conduct studies assessing mosaicism in AD brains. Our findings showed greater mosaicism—in particular, for increased [amyloid precursor protein] copy numbers. Most notably, segments of DNA in the APP gene were found to not only be amplified in some neurons. But certain APP segments increased in number more than others, hinting at GR.

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In principle, FDA-approved medications such as reverse-transcriptase inhibitors could be used today and may have special benefit for people in high-risk categories for whom no effective treatments currently exist. GR affecting different genes may underlie one or more of hundreds of other brain diseases and might also affect other cell types beyond the brain.

Read full, original post: [A New Idea about What Triggers Alzheimer's](#)