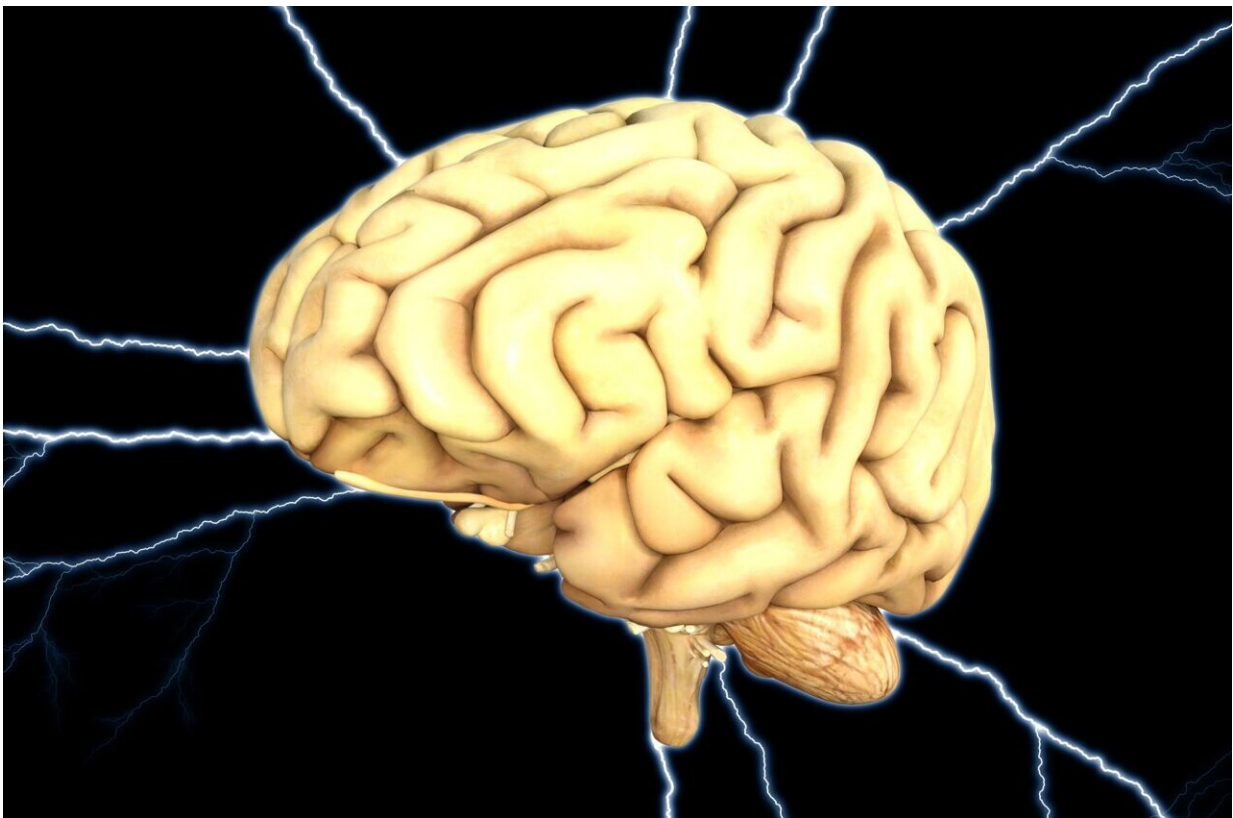


# Researchers introduce into human cells a genetic mutation that protects against Alzheimer's disease

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Credit: Pixabay/Pete Linforth.

Researchers from the Université Laval Faculty of Medicine and CHU de Québec–Université Laval Research Center have successfully edited the

genome of human cells grown in vitro to introduce a mutation providing protection against Alzheimer's disease. The details of this breakthrough were recently published in *The CRISPR Journal*.

"Some [genetic mutations](#) increase the risk of developing Alzheimer's disease, but there is a mutation that reduces this risk," says lead author Professor Jacques-P. Tremblay. "This is a rare mutation identified in 2012 in the Icelandic population. The mutation has no known disadvantage for those who carry it and reduces the risk of developing Alzheimer's disease. Using an improved version of the CRISPR gene editing tool, we have been able to edit the genome of human cells to insert this mutation."

The brains of those with Alzheimer's present amyloid plaques, which have a level of toxicity believed to cause neuron death. These plaques are formed when the [amyloid precursor protein](#) is cleaved by an enzyme called beta-secretase. "The Icelandic mutation makes it harder for this enzyme to cleave the amyloid precursor protein. As a result, the formation of amyloid plaques is reduced," explains Professor Tremblay.

In theory, introducing the Icelandic mutation into the genome of people at risk of developing Alzheimer's could prevent or slow the progression of the disease. "Unfortunately, we can't go back and repair the damage that caused neurons to die," says the researcher. "The treatment would therefore be particularly suitable for people from families affected by the hereditary form of the disease, which manifests itself in memory problems from the age of 35 to 40. If successful, it could also potentially be used to treat people with the most common form of Alzheimer's, which occurs after age 65, at the earliest signs of the disease."

"The challenge now is to find a way to edit the genome of millions of [brain cells](#)," says Professor Tremblay. "We are looking at different possibilities, including the use of non-infectious viruses, to deliver the

editing complex inside neurons. Now that the proof of concept has been established in [human cells](#) in vitro, we will test this approach in mice that express Alzheimer's disease. If the findings are conclusive, we hope to be able to conduct a [small-scale study](#) in people with [mutations](#) that cause the onset of Alzheimer's at age 35 to 40."

In addition to Jacques-P. Tremblay, the authors of the study published in *The CRISPR Journal* are Guillaume Tremblay, Joël Rousseau, and Cédric Mbakam.

**More information:** Guillaume Tremblay et al, Insertion of the Icelandic Mutation (A673T) by Prime Editing: A Potential Preventive Treatment for Familial and Sporadic Alzheimer's Disease, *The CRISPR Journal* (2022). [DOI: 10.1089/crispr.2021.0085](https://doi.org/10.1089/crispr.2021.0085)

Provided by Laval University

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