Was a gene mutation responsible for bigger human brains?

One of the major features that distinguish humans from other primates is the size of our brains, which underwent rapid evolution from about two to three million years ago in a group of our ancestors in Africa called the Australopithecines. During this period, the human brain grew almost three-fold to reach its current size.

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[Professor Pierre Vanderhaeghen's] lab discovered 35 hominid – present only in apes and humans – genes that were active in foetal brain tissue. They then became intrigued by three specific genes – all similar to NOTCH genes, an ancient gene family involved in sending messages between cells and that are present in all animals. They found that the three new genes, collectively named NOTCH 2NL, were created by a copy and paste error of an original NOTCH gene.

This error created entirely new proteins which likely helped our ancestors' cerebral cortex to balloon. This is the part of our brain responsible for our language, imagination and problem-solving abilities.

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As DNA in this area closely resembles another part of the genome where it was originally cut and pasted from millions of years ago, errors are more likely, said Prof. Vanderhaeghen. 'Patients who have (inherited) deletions in this area tend to be at risk of developing schizophrenia, whereas patients with duplications are more at risk of autistic spectrum disorder,' he said.

Read full, original post: Genetic error led humans to evolve bigger, but more vulnerable, brains