New gene variant linked to stroke

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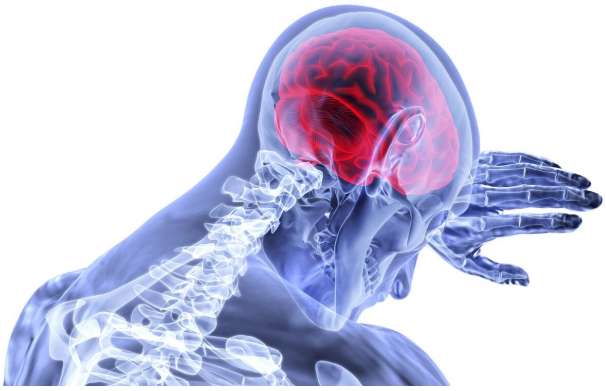

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Researchers at Lund University in Sweden believe they have identified a gene variant that can cause cerebral small vessel disease and stroke. The study is published in *Neurology Genetics*.

"The patients we have studied are from the same extended family, and several of them have been diagnosed with cerebral small vessel disease and suffered strokes. After tissue examination and using genetic sequencing methods, we found that they were carriers of a new gene variant that could be connected to their diagnoses," says Andreea Ilinca, researcher at Lund University and neurologist at Skåne University Hospital.

Stroke is either caused by a blood clot that leads to a lack of oxygen in the brain, or a hemorrhage in the brain. High blood pressure, high cholesterol levels, diabetes, atrial fibrillation and lifestyle factors such as smoking are known risk factors for stroke. However, an increasing amount of research is indicating that genetic factors also play a major role.

Therefore, the Lund researchers have studied an extended family, the majority of whom live in southern Sweden, where eight out of 15 people developed cerebral small vessel disease. The disease is characterized by ischemic stroke (cerebral infarction caused by blood clots) and cerebral hemorrhage, as well as mild cognitive impairment, autonomic nervous system dysfunctions and coordination difficulties.

When examining tissue from those that had experienced symptoms, the researchers could see microscopic changes in the blood vessels of the brain and in small skin vessels.

Using modern genetic analysis methods, they were also able to establish that they had found a new variant in the MAP3K6 gene, that they believe may be related to the disease. MAP3K6 is a gene that, among other things, affects the function of a protein that helps the brain's blood vessels to react correctly to damage, such as a low oxygen supply to the brain.

"By identifying genetic variants that are associated with disease in the vessels of the brain and early stroke, we can better understand what could prevent these harmful processes. Future studies that can give us more knowledge about the molecular disease mechanism can lead to new treatments", concludes Andreea Ilinca.

More information: Andreea Ilinca et al. MAP3K6 Mutations in a Neurovascular Disease Causing Stroke, Cognitive Impairment, and Tremor, *Neurology Genetics* (2021). DOI: 10.1212/NXG.0000000000000548

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