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# Genetic predisposition to rare form of stroke identified

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New research from a consortium led by Royal Holloway, University of London, has identified the first genetic link for Cerebral Venous Thrombosis (CVT): an uncommon form of stroke affecting mostly young individuals, and primarily women.

With stroke remaining the third commonest cause of death in the UK, these new findings offer the first evidence of genetic susceptibility to

CVT, providing valuable insights into this previously misunderstood disease.

CVT is a blood clot of a cerebral vein in the brain, accounting for less than 1% of all stroke cases worldwide. Unlike the more common stroke which clots arteries in the brain, CVT is caused by clots in the veins in the brain, ultimately leading to blood leaking into the brain and causing brain swelling. CVT can be potentially more fatal than the common form of stroke, especially in young patients, so preventing and predicting its onset has been a major goal.

Although numerous risk factors have previously been reported as contributors to adult CVT, its genetic basis has not been well understood. In this study, researchers were able to identify the first chromosomal region to strongly associate with a genetic susceptibility to CVT: the ABO gene—a gene that determines individual blood group status

In the new study, "Genome-wide association study identifies first locus associated with susceptibility to cerebral venous thrombosis," 882 Europeans diagnosed with CVT (and a control group of 1205 individuals of similar demographics without CVT) took part in the genome study.

By identifying an area of the human genome that strongly correlates with CVT, researchers found that this region more than doubles the likelihood of CVT: a risk greater than any previously identified genetic risk marker. Researchers were also able to demonstrate that those with an AB blood group faced a 5.6 times increased risk of CVT, compared with those from blood group O.

Professor Pankaj Sharma, Director of the Institute of Cardiovascular Research at Royal Holloway, University of London, said: "CVT is a form of stroke which affects young people, especially women. Our work, 10 years in the making, brings together a global international

collaboration that has identified a risk factor, as simple as someone's blood group, and presents a major step forward in predicting and understanding the mechanism of this disease.

"Needless to say, CVT has entered the public domain following its association with the AstraZeneca COVID-19 vaccine. Whether an individual's blood group status affects their risk of developing CVT following the vaccine remains to be seen."

**More information:** Gie Ken-Dror et al, Genome-Wide Association Study Identifies First Locus Associated with Susceptibility to Cerebral Venous Thrombosis, *Annals of Neurology* (2021). DOI: 10.1002/ana.26205

Provided by Royal Holloway, University of London

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