

Scientists find gene linked to schizophrenia

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An international study led by Université de Montréal scientists suggests that gene mutations may predispose some individuals to schizophrenia and provides new clues about the causes of this ambiguous disorder. Published today in the *Proceedings of the National Academy of Sciences*, the findings demonstrate that new mutations in the SHANK3 gene are found in schizophrenic patients.

"That these de novo or new mutations occur in schizophrenia is rather unexpected and may explain why the identification of the genes linked to this disease has been so difficult," says senior author Guy Rouleau, a Université de Montréal professor, director of the Sainte-Justine University Hospital Research Center and a scientist at the Research Centre of the Centre Hospitalier de l'Université de Montréal.

"Our findings show that a significant number of schizophrenia cases are the result of new genetic mutations in the SHANK3 gene. Where previous approaches have failed, our detailed analyses and rich patient database led us to this discovery. We are convinced that future studies will validate the SHANK3 gene as a marker for schizophrenia," continues Dr. Rouleau, who is Canada Research Chair in Genetics of the Nervous System.

Autism and schizophrenia link

"The SHANK3 gene has previously been linked to autism," adds lead author Julie Gauthier, a Université de Montréal researcher. "Not only does this suggest a molecular genetic link between these two



neurodevelopmental disorders, it suggests that SHANK3 may have a role in other brain disorders."

SHANK3 is protein involved in maintaining the physical structure of nerve cells. Mutations in this gene result in specific abnormalities in cell shapes. These deformations have been observed in some <u>schizophrenia</u> patients, providing further evidence of the importance of the SHANK3 gene in this disorder.

These findings were made possible through the collaboration of several institutions such as the Harvard Medical School, McGill University and the Université Paris Descartes; please read on for complete list of funders and partners.

Affecting approximately one percent of the population, schizophrenia is a chronic psychiatric disorder characterized by disturbances in thinking, behaviour and emotion. Symptoms include delusions, hallucinations and withdrawal from social activity.

More information: Proceedings of the National Academy of Sciences: www.pnas.org

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