

Autism and schizophrenia: Scientists measure gene mutation rate

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An international study led by University of Montreal scientists suggests family history may not be a good predictor of the presence of mutations predisposing to autism or schizophrenia.

The findings show how new or de novo gene mutations - alterations of the cell's DNA - play a role in these devastating conditions. Published in the <u>American Journal of Human Genetics</u>, this study has implications for disease prevalence and severity.

"This study emphasizes the importance of de novo mutations as <u>genetic</u> <u>factors</u> predisposing to <u>autism</u> and schizophrenia. We found an increased frequency of severe de novo mutations in critical brain <u>genes</u> in both of these diseases," says senior author and University of Montreal professor, Guy Rouleau.

"Harmful de novo mutations, as observed in this study, may in part explain the high global incidences of autism and schizophrenia," adds Dr. Rouleau, who is also director of the Sainte-Justine University Hospital Research Center and a scientist at the University of Montreal Hospital Research Centre.

Investigating human mutation rate:

The team analyzed 400 genes that are turned on in <u>nerve cells</u> from patients with autism or <u>schizophrenia</u> spectrum disorders. Their results



showed that there is an excess of de novo gene mutations associated with the two diseases.

Their study revealed that DNA taken directly from the patient's blood was superior to that taken from patient-derived cell lines. "The source of biological material is crucial for these types of experiments," says lead author Philip Awadalla, a University of Montreal pediatrics professor, scientist at the Sainte-Justine University Hospital Research Center and scientific director of the CARTaGENE project.

"In the process of confirming our findings, we were also able to provide one of the first direct estimates of the human mutation rate," continues Dr. Awadalla. "The number of mutations per generation is extremely small but on the order of what was previously indirectly inferred for human-chimpanzee comparisons. We also discovered that mutations can be introduced when cell lines are produced, which creates false-positive results. This artefact can significantly bias results and therefore great care needs to taken when analyzing these samples."

About de novo mutations:

Mutations are alterations of the cell's DNA that can occur because of errors in the DNA replication, which happen prior to cell division. Once DNA is changed, this mutation is passed down to a next generation. A mutation that is newly formed and therefore not inherited from either parent is called a de novo mutation.

More information: The paper, "Direct Measure of the de novo Mutation Rate in Autism and Schizophrenia cohorts," published in the *American Journal of Human Genetics*, was authored by Philip Awadalla, et al. <u>www.cell.com/AJHG</u>



Provided by University of Montreal

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