

Gene mutation linked to psychosis in Icelandic family

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A depiction of the double helical structure of DNA. Its four coding units (A, T, C, G) are color-coded in pink, orange, purple and yellow. Credit: NHGRI



(Medical Xpress)—A team of researchers from Iceland, Finland and Germany has found evidence of a gene mutation in a large family in Iceland that explains why so many of them suffer from psychosis. In their paper published in the journal *Nature Genetics*, the team outlines the process they used to find the mutated gene, how they used their findings to study cases in other families, and the ways their findings might impact research on psychiatric disorders going forward.

Psychosis is a broad term used to describe a condition in which a person experiences a disconnect between what their senses and the real world. Such people can experience hallucinations, have delusions and suffer from emotional extremes. Some ailments that cause psychosis are schizophrenia and bipolar disorders. Because such disorders are thought to have a genetic cause, researchers studying how psychosis comes about look at the genes of not only people experiencing psychosis, but other family members, as well. Research efforts have uncovered genetic abnormalities associated with psychosis, but thus far, no definitive mutations have been found that pinpoint its exact cause. In this new effort, the researchers report that they have found a gene mutation that appears to be at least partly responsible for several members of a single family suffering from several forms of the disorder.

The researchers report that six of the family members were diagnosed with schizophrenia, two with schizoaffective disorder and two with bipolar-related disorders. The team conducted a genotyping and imputation-based long-range phasing genetic analysis on family members and discovered that each of the 10 psychosis patients had evidence of mutations in the RBM12 gene. The researchers report that not all of the family members had the mutations, but most of those that did reported experiencing some degree of psychic disorder.

The research team ran their findings through the Genome Aggregation Database and discovered no other such mutations were listed. They also



searched medical records and found less than two dozen other people in Iceland with the mutations, all of whom were related to the family in which they were originally found. They conclude by suggesting that RBM12 appears to be important for normal brain function and suggest that further research on the gene might offer more insights into psychiatric disorders.

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