

Researchers uncover impact of mutations in the human genome on cognitive ability

19 December 2013, by Tom Langford

deCODE genetics reported today in the journal *Nature* that mutations associated with an increased risk of schizophrenia and autism also affect cognition in individuals without the disease or intellectual disability. The research suggests that cognitive abnormalities are fundamental in schizophrenia, yet outward signs of the disease only manifest in a subset of carriers of the mutations.

"This study provides one of the first footholds into biochemical understanding of humans' unique [cognitive abilities](#)," said study lead author Kari Stefansson, M.D., Dr. Med., CEO of deCODE genetics. "At the same time, these results raise important questions as to why certain mutations impact some individuals more dramatically than others."

Certain copy number variants (CNVs), one type of mutation, have been previously identified as strong risk factors for schizophrenia and autism, neuropsychiatric diseases which affect cognition. Researchers sought to understand what, if any, impact these CNVs may have on cognitive ability in disease-free carriers.

They administered neuropsychiatric tests to 1,268 individuals across four groups ([schizophrenia patients](#), control carriers of neuropsychiatric CNVs, control carriers of other CNVs and population controls), and found that carriers of CNVs that predispose to [neuropsychiatric disease](#) have cognitive abilities between people without the CNVs and people with schizophrenia.

"Our results suggest that neuropsychiatric CNVs can be used as an instrument for further study of the cognitive abnormalities that characterize schizophrenia, because whether or not an individual develops this disease is likely related to the expression of these genes," said Stefansson. "The findings also provide insight into which cognitive abilities put individuals at risk of

developing [schizophrenia](#) and demonstrate that control carriers provide an opportunity to study cognitive abnormalities without the confounding effects of psychosis or medication."

More information: "CNVs conferring risk of autism or schizophrenia affect cognition in controls." Hreinn Stefansson, Andreas Meyer-Lindenberg, Stacy Steinberg, et al. *Nature* (2013) [DOI: 10.1038/nature12818](https://doi.org/10.1038/nature12818)

Provided by deCode Genetics

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