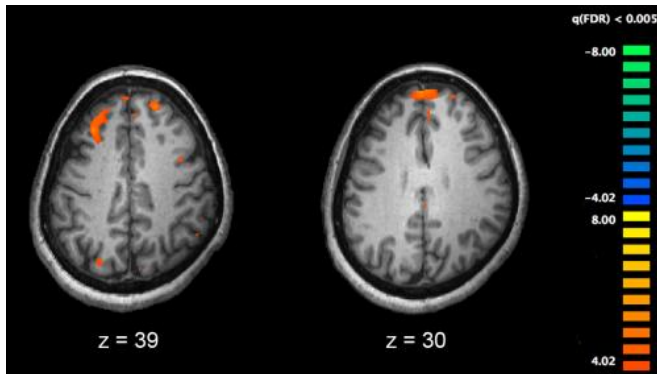


Study uses polygenic risk scores to determine schizophrenia risk in patients with chromosome deletion syndrome

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Functional magnetic resonance imaging (fMRI) and other brain imaging technologies allow for the study of differences in brain activity in people diagnosed with schizophrenia. The image shows two levels of the brain, with areas that were more active in healthy controls than in schizophrenia patients shown in orange, during an fMRI study of working memory. Credit: Kim J, Matthews NL, Park S./PLoS One.

A new study involving researchers from Children's Hospital of Philadelphia (CHOP) has uncovered genetic clues that may help identify which patients with chromosome 22q11.2 deletion syndrome (22q11.2DS) might develop schizophrenia. The findings, published in *Nature Medicine*, highlight the potential of polygenic scores, which account for the effects of numerous common genetic variants across the genome, to determine whether a patient is at high or low risk of developing the psychiatric condition.

22q11.2DS results from a missing segment of approximately 45 genes on one copy of chromosome 22 and is the most common genetic cause of [schizophrenia](#), occurring in nearly 30% of patients with the deletion. However, not all patients with the condition go on to develop schizophrenia,

which prompted the researchers to explore the potential contributions of other genetic variants across the genome.

Using [genetic data](#) from the International 22q11.2 Brain and Behavior Consortium (IBBC), a large NIH-funded consortium led by CHOP and the Perelman School of Medicine at the University of Pennsylvania, the researchers analyzed 965 patients with 22q11DS and evaluated genetic associations using polygenic risk scores for schizophrenia and cognitive ability. The research team found polygenic risk scores were key in assessing the probability of a given patient developing schizophrenia or cognitive decline.

"This work is an important next step in better understanding the causes of behavioral health differences associated with the chromosome 22q11.2 deletion," said co-author Donna McDonald-McGinn, MS, LCGC, Director of the 22q and You Center and, Associate Director of Clinical Genetics, Chief of the Section of Genetic Counseling at CHOP, Clinical Professor of Pediatrics at UPenn and Co-Principal Investigator of the IBBC.

"Although the funding for the IBBC ended in 2017, this important work continues through another NIH-funded program entitled the Genes to Mental Health Network (G2MH), where we have expanded our target populations to include other chromosomal deletions and duplications that have associated behavioral phenotypes, including the 22q11.2 deletion and duplication syndromes and the 16p11.2 deletion and duplication syndromes, among others."

More information: undefined undefined et al. Using common genetic variation to examine phenotypic expression and risk prediction in 22q11.2 deletion syndrome, *Nature Medicine* (2020). DOI: [10.1038/s41591-020-1103-1](https://doi.org/10.1038/s41591-020-1103-1)

Provided by Children's Hospital of Philadelphia

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