NEWS

Genetics: Deletions common in early schizophrenia syndrome

BY **JESSICA WRIGHT**

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Hot spot: One-third of the alterations found in children with childhood-onset schizophrenia are in the chromosomal region 22q11.2.

Chromosomal abnormalities may be more prevalent in individuals with a rare form of childhood-onset schizophrenia than in those with the classic presentation of the disorder, according to a study published 21 May in *Molecular Psychiatry*[1].

The researchers found an especially high prevalence of deletions in 22q11.2 — a chromosomal region associated with schizophrenia and autism — in children diagnosed with schizophrenia before 13 years of age.

These children show symptoms of schizophrenia, such as delusions and hallucinations, that do not emerge until adulthood in the more common form of the disorder.

The researchers presented **preliminary results** from this study in January at the **Salk Institute, Fondation IPSEN and *Nature* Symposium on Biological Complexity** in La Jolla, California.

Several studies published in the past few years have found that duplications and deletions of chromosomal regions, called **copy number variants** (CNVs), are **more prevalent in people with neuropsychiatric disorders** than in controls. Many of these CNVs have been seen in multiple disorders, suggesting the disorders have a shared genetic cause.

For example, about 30 percent of people with **a deletion in 22q11.2** have schizophrenia, and as much as 20 percent have autism. However, some researchers suggest that these children have **misdiagnosed childhood-onset schizophrenia** rather than autism.

In the new study, researchers scoured the genomes of 126 individuals with childhood-onset schizophrenia for 46 CNVs linked to schizophrenia, autism, intellectual disability or **epilepsy**, including 15q11.2, 16p12.1, 17q21.3 and **16p11.2**. They found that 15 of the individuals (12 percent) have one of these CNVs, and 4 (3 percent) have two CNVs. Of the 15 individuals with a disease-linked CNV, 5 have a deletion in 22q11.2.

The researchers also looked at the prevalence of these CNVs in two datasets of adults with classic schizophrenia. They found these CNVs in 5 percent of one group of 977 adults and 1.4 percent of another group of 649, smaller frequencies than in those with childhood-onset schizophrenia.

They then compared the prevalence of these CNVs in 69 of the children with schizophrenia and their full siblings. They found that 13 of the children with the disorder have a disease-linked CNV, compared with only 1 of their siblings.

References:

1: Ahn K. *et al. Mol. Psychiatry* Epub ahead of print (2013) **PubMed**