NEWS

Genetics: Chromosome 16 region linked to schizophrenia

BY **JESSICA WRIGHT**

19 MARCH 2013

A rare deletion on chromosome 16 is more common in individuals who have schizophrenia than in controls, according to a large genetics study published 16 January in *JAMA Psychiatry*[1]. This region is close to **16p11.2**, implicated in both autism and schizophrenia.

Studies suggest that such structural alterations to the genome can influence gene expression **in nearby regions**, potentially linking the two variations.

Several studies have identified duplications or deletions of DNA — known as **copy number variations** (CNVs) — that are **more common** in individuals with neuropsychiatric disorders than in controls. Most of these duplications or deletions **have links to multiple disorders**. For example, a particular CNV may increase the risk of schizophrenia, autism and general developmental delay.

Linked loci: A region 600 kilobases away from a classic autism-linked deletion may also be involved in neuropsychiatric disorders.

One of the most common CNVs found in individuals with autism is a duplication or deletion of an approximately **600-kilobase stretch in the 16p11.2** region. People with this CNV can have a range of neuropsychiatric conditions, including autism or schizophrenia, and are **often obese**.

The new study shows that a chromosomal region 600 kilobases away from 16p11.2 may be linked

to schizophrenia. Researchers found the deletion in 2 of 790 individuals with schizophrenia and none of 1,347 controls. They also found that of 662 children with schizophrenia who were tested along with their unaffected parents, 2 have the deletion, which they inherited from their mothers.

The researchers then confirmed this association in a separate group of 12,398 individuals with schizophrenia and 17,945 controls. In this group, nine people with schizophrenia and three controls have the deletion.

Overall, people with schizophrenia are about six times more likely to carry the deletion than are controls, the study found.

A 2011 study also found that this deletion is more common in a large **group of children with developmental disabilities** (including intellectual disability and autism) than in controls[2]. Another study has **linked deletions that encompass this region** to obesity[3]. As with other CNVs, the effects of the deletion are variable, the researchers note.

References:

1:Guha S. *et al. JAMA Psychiatry* **70**, 253-260 (2013) **PubMed**

2:Cooper G.M. *et al. Nat. Genet.* **43**, 838-846 (2011) **PubMed**

3: Bochukova E.G. *et al. Nature* **463**, 666-670 (2010) **PubMed**