MedicalXpress

X marks the spot -- TBL1X gene involved in autism spectrum disorder

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Autism Spectrum Disorder (ASD) affects about 1 in 100 children resulting in a range of problems in language, communication and understanding other people's emotional cues, all of which can lead to difficulties in social situations. Boys are three to four more times as likely to be affected as girls and consequently it has been suggested that the genes involved in this disorder may be linked to the X chromosome. New research published in BioMed Central's open access journal *Molecular Autism* used genome wide association study (GWAS) data to find a variation in the gene for transducin ß-like 1X-linked (TBL1X) which is associated with increased risk of ASD in boys.

A team of researchers across America combined three sets of genomic data incorporating over 3000 affected children and their family members or non-related case control individuals. The GWAS study compared single nucleotide polymorphisms (SNP) on the X chromosomes of the children with ASD to the control groups, and found differences within the genes for Duchenne muscular dystrophy (DMD), IL1RAPL2 (involved in inflammation), and in TBL1X. TBL1X is part of the Wnt-signaling pathway, which is in turn part of the complex mechanism controlling embryonic neurological development and the maintenance of brain function in adults.

Prof Eden Martin from the Hussman Institute for Human Genomics, who lead the multi-centre team explained, "The SNP in TBL1X is associated with an increase in risk for ASD of about 15%. This could reflect either an unidentified rare mutation (or mutations), which has

large impact, or a more common change with a more subtle effect, on the development of ASD. Further study of TBL1X will help us to pinpoint the DNA changes involved and help us to understand exactly how these changes and the Wnt-signaling pathway is involved in ASD."

Provided by BioMed Central