MedicalXpress

Gene screen to identify causes of autism

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A new screening method can be used to detect the chromosomal abnormalities most commonly associated with autism spectrum disorders. By screening for genetic defects associated with various kinds of cognitive impairment, the approach described in the open access journal *BMC Medical Genomics* will help clinicians identify the underlying causes of some patients' autism spectrum disorders (ASDs).

Joseph Buxbaum from Mount Sinai School of Medicine, New York, led a team of researchers tasked with evaluating the use of 'multiplex ligation-dependent probe amplification' (MLPA), a recently developed method of investigating DNA, in genetic counselling. He said, "MLPA is a relatively practical, inexpensive and fast tool for screening chromosome rearrangements in autism spectrum disorders".

ASDs have been increasingly associated with genetic abnormalities. At the same time, many children with ASDs also have some degree of cognitive impairment. In this study, the authors used MLPA on a group of 279 children with ASD, looking for abnormalities that are known to be associated with cognitive impairment. As Buxbaum describes, "By focussing on well-known genetic disorders, rather than assaying an individual's entire genome, MLPA allows for much more efficiency". As additional genetic abnormalities associated with ASDs are identified, additional probes can be used in future screens.

As well as demonstrating the effectiveness of MLPA as a screen for known genetic disorders, the authors also identified some new genetic changes that are likely to contribute to ASD, such as novel duplications

(extra copies of genetic material) in chromosomes 15 and 22, which may increase liability and/or exacerbate ASD symptoms.

Although there is no known cure for ASDs, early detection and commencement of special education and behavioural therapy can mitigate some of the negative symptoms.

Citation: Multiplex ligation-dependent probe amplification for genetic screening in autism spectrum disorders: Efficient identification of known microduplications and identification of a novel microduplication in ASMT, Guiqing Cai et al. *BMC Medical Genomics* (in press)

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