

Genetic test developed for those at-risk for Autism Spectrum Disorder: Accuracy to be studied

September 12 2012

(Medical Xpress)—A team of Australian researchers, led by University of Melbourne has developed a genetic test that is able to predict the risk of developing Autism Spectrum Disorder, ASD.

Lead researcher Professor Stan Skafidas, Director of the Centre for Neural Engineering at the University of Melbourne said the test could be used to assess the risk for developing the disorder. "This test could assist in the early detection of the condition in babies and children and help in the early management of those who become diagnosed," he said. "It would be particularly relevant for families who have a history of Autism or related conditions such as Asperger's Syndrome," he said.

Autism affects around one in 150 births and is characterized by abnormal [social interaction](#), impaired communication and repetitive behaviours. The test correctly predicted ASD with more than 70 per cent accuracy in people of central European descent. Ongoing validation tests are continuing including the development of accurate testing for other ethnic groups.

Clinical neuropsychologist, Dr Renee Testa from the University of Melbourne and Monash University, said the test would allow clinicians to provide early interventions that may reduce behavioural and [cognitive difficulties](#) that children and adults with ASD experience. "Early identification of risk means we can provide interventions to improve

overall functioning for those affected, including families," she said.

A [genetic cause](#) has been long sought with many genes implicated in the condition, but no single gene has been adequate for determining risk. Using US data from 3,346 individuals with ASD and 4,165 of their relatives from Autism [Genetic Resource](#) Exchange (AGRE) and Simons Foundation [Autism Research](#) Initiative (SFARI), the researchers identified 237 genetic markers (SNPs) in 146 genes and related [cellular pathways](#) that either contribute to or protect an individual from developing ASD.

Senior author Professor Christos Pantelis of the Melbourne Neuropsychiatry Centre at the University of Melbourne and Melbourne Health said the discovery of the combination of contributing and protective gene markers and their interaction had helped to develop a very promising predictive ASD test.

The test is based on measuring both genetic markers of risk and protection for ASD. The risk markers increase the score on the genetic test, while the protective markers decrease the score. The higher the overall score, the higher the individual risk.

"This has been a multidisciplinary team effort with expertise across fields providing new ways of investigating this complex condition," Professor Pantelis said.

The next step is to further assess the accuracy of the test by monitoring children who are not yet diagnosed over an extended study. The study has been published today in the journal *Molecular Psychiatry*.

Provided by University of Melbourne

Citation: Genetic test developed for those at-risk for Autism Spectrum Disorder: Accuracy to be studied (2012, September 12) retrieved 25 April 2024 from <https://medicalxpress.com/news/2012-09-genetic-at-risk-autism-spectrum-disorder.html>

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