Gene variation links to autistic-like traits

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Genetic information and the results from the AQ questionnaire were compared from nearly 1000 individuals from the Raine study (shown here some participants of the Raine Study)

Researchers have confirmed an association between a genetic mutation and a higher level of autistic-like traits in individuals.

The study investigated associations between autistic-like traits in a Western Australian population sample and five previously identified point mutations—affecting one or very few nucleotides— associated with Autism Spectrum Disorders (ASD).

ASD is the group of neuro-developmental conditions, including autism and Asperger syndrome, characterised by impairments in social interaction and communication, repetitive interests and behaviours.

A recent review estimated the median global prevalence of ASD at 62 cases per 10,000 children.

UWA Centre for Genetic Origins of Health and Disease expert Rachel Jones compared genetic information and the results from the Autism Spectrum Quotient (AQ) questionnaire of nearly 1000 individuals from the Raine Study—a WA Pregnancy Cohort born in 1989¬–1991.

"The AQ is a self-report questionnaire that provides a quantitative measure of autistic-like traits in the general population," Ms Jones says.

"Individuals are provided with 50 statements and asked to indicate on a four-point scale how well that statement applies to them."

Questions relate to social ability, attention to detail/patterns and the understanding of others.

Genetic association tests compared the results of the AQ test to the presence of five single nucleotide polymorphisms (SNPs) located on genes in the study group's DNA samples.

Secondary variables considered

"The analysis took into account covariates [such as male sex, mother's BMI, parent education and family income] that may also affect the AQ score and different models were tested to determine if they were of significance," Ms Jones says.

They found a positive association with one point mutation on a gene

known to be expressed in the brain.

"While these [remaining four] points might not be the actual cause of autistic-like traits, the causal variant may be located close by on the same chromosome."

Ms Jones says the study of autistic-like traits in the general population is a relatively new way of studying autism and may lead to understanding the causes of ASD.

"By measuring autistic-like traits in the population we are able to get a larger sample size and shed some light on what types of mutation are happening."

Identification of causal genes could assist in earlier diagnosis of ASD, leading to earlier implementation of intervention strategies to reduce the impact of ASD on families and the Australian healthcare system.

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