

New genetic study of Asperger syndrome, autistic traits and empathy

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Scientists from the University of Cambridge have identified 27 genes that are associated with either Asperger Syndrome (AS) and/or autistic traits and/or empathy. The research will be published tomorrow in the journal *Autism Research*. This is the first candidate gene study of its kind.

The research was led by Dr Bhismadev Chakrabarti and Professor Simon Baron-Cohen from the Autism Research Centre in Cambridge. 68 genes were chosen either because they were known to play a role in neural growth, social behaviour, or sex steroid hormones (e.g. testosterone and estrogen). The latter group of genes was included because AS occurs far more often in males than females, and because previous research from the Cambridge team has shown that foetal testosterone levels are associated with autistic traits and empathy in typically developing children.

The team carried out 2 experiments. First they looked at these genes in 349 adults in the general population, all of whom had filled in the Autism Spectrum Quotient (AQ) as a measure of autistic traits, and the Empathy Quotient (EQ) as a measure of empathy. Secondly, they looked at 174 adults with a formal diagnosis of AS, and compared them to controls.

The research found that single <u>nucleotide polymorphisms</u> (SNPs) in 27 out of the 68 genes were nominally associated with either AS and/or with autistic traits/empathy. 10 of these genes (such as CYP11B1) were



involved with sex steroid function, providing support for the role of this class of genes in autism and autistic traits. 8 of these genes (such as NTRK1) were involved in neural growth, providing further support to the idea that autism and autistic traits could result from aberrant patterns of connectivity in the developing brain. The other 9 genes (such as OXTR) were involved in social behaviour, shedding light on the biology of social and emotional sensitivity.

Dr Chakrabarti commented: "These 27 genes represent preliminary leads for understanding the genetic bases of AS and related traits, such as empathy, in the general population. All of these are good candidates for independent replication studies in both low and high functioning autism samples. 5 of the genes we found have been previously reported in autism, but the other 22 have never before been reported in association with AS, autistic traits or empathy. We now need to test models of how these genes interact and construct 'risk' models for the development of AS."

Professor Baron-Cohen added: "We chose to look at the genetics of AS because all other genetic studies have focused on classic autism, which can include learning difficulties and language delay. AS is a more 'pure' condition because these other factors are absent. These new results represent a significant advance over our previous work in showing that the sex steroid hormones (e.g. testosterone and oestrogen) influence social development and autistic traits. The new study also confirms earlier reports that other molecules (such as oxytocin) are important in understanding <u>autism</u>, autistic traits, and empathy."

Source: University of Cambridge (<u>news</u>: <u>web</u>)

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