

Study confirms a gene linked to Asperger Syndrome and empathy

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(Medical Xpress)—Scientists have confirmed that variations in a particular gene play a key role in the autism spectrum condition known as Asperger Syndrome. They have also found that variations in the same gene are also linked to differences in empathy levels in the general population.

A study to be published later this month in the journal *Molecular Autism* confirms previous research that people with Asperger Syndrome (AS) are more likely to carry specific variations in a particular gene. More strikingly, the study supports existing findings that the same gene is also linked to how much empathy typically shown by individuals in the general population.

The research was carried out by a team of researchers led by Professor Baron-Cohen at the Autism Research Centre at Cambridge University. Asperger Syndrome is an autism spectrum condition. The researchers looked for sequence variations (called single nucleotide polymorphisms or SNPs) in the gene known as GABRB3 in a total of 530 adults - 118 people diagnosed with AS and 412 people without a diagnosis.

The team found that certain SNPs in GABRB3 were significantly more common in people with AS.

They also discovered that additional genetic variations in the same gene were linked to scores on an empathy measure called the Empathy Quotient (EQ) in the general population.

AS is diagnosed when a person struggles with social relationships and communication, and shows unusually narrow interests and resistance to change, but has good intelligence and language skills. Most genetic studies of autistic spectrum conditions treat autism as if they are all very similar, whereas in reality there is considerable variation (e.g., in language level and intellectual ability).

Rather than studying people on the autistic condition spectrum, this new study looked only people with AS, as a well-defined subgroup of individuals within this range. The researchers examined the gene GABRB3 which regulates the functioning of a neurotransmitter called gamma-aminobutyric acid (GABA) and which contains a number of SNPs that vary across the population.

The volunteers were tested for 45 SNPs within this key gene. The team had previously found that SNPs in this gene were more common in adults with AS and also showed a relationship with empathy levels and tactile sensitivity (how sensitive people are to being touched) in the general population.

Testing a new sample of volunteers who had not taken part in previous studies, the researchers found that three of the SNPs were again more common in adults with AS, and two different SNPs in the same gene were again related to empathy levels in the [general population](#), confirming that the gene is involved in autism spectrum conditions.

Professor Baron-Cohen said: "We are excited that this study confirms that variation in GABRB3 is linked not just to AS but to individual differences in empathy in the population. Many candidate [genes](#) do not replicate across studies and across different

samples, but this genetic finding seems to be a solid result. Research now needs to focus on where this gene is expressed in the brain in autism, and how it interacts with other genetic and non-genetic factors that cause AS."

The team was co-led by Dr BhismaDev Chakrabarti from the Department of Psychology at Reading University. He commented: "Genes play an important role in autism and Asperger Syndrome. This new study adds to evidence that GABRB3 is a key gene underlying these conditions. This gene is involved in the functioning of a neurotransmitter that regulates excitation and inhibition of nerve cell activity so the research gives us vital additional information about how the brain may develop differently in people with Asperger Syndrome."

Varun Warriar, who carried out the study as part of his graduate research at Cambridge University, added: "The most important aspect of this research is that it points to common genetic variants in GABRB3 being involved in both AS and in empathy as a dimensional trait. Although GABRB3 is not the only gene to be involved in this condition and in empathy levels, we are confident that we have identified one of the key players. We are following this up by testing how much protein GABRB3 produces in the brain in autism, since a genetic finding of this kind becomes more explanatory when we can also measure its function."

More information: "Genetic variation in GABRB3 is associated with Asperger syndrome and multiple endophenotypes relevant to autism." Varun Warriar, Simon Baron-Cohen and BhismaDev Chakrabarti. *Molecular Autism* 2013, 4:48 [DOI: 10.1186/2040-2392-4-48](https://doi.org/10.1186/2040-2392-4-48)

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