research snapshot

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Can Microgene Variation Influence Autism Spectrum Disorder?

What is this research about?

Some mental health concerns are connected to issues in the nervous system. A subset of psychiatry has been developed to explore these concerns. This field is called neuropsychiatry. One area of this field explores how mental health concerns are linked to rare variations in genes.

Autism spectrum disorder (ASD) is a mental health concern that can be connected to gene variation. ASD that is caused by issues in the nervous system create a higher chance of other neurological issues later in life. Thus, understanding the cause of mental health concerns can assist in placing preventative health care in place where greater risk occurs.

This study explores a specific rare duplication in a section of more than 50 genes. An issue with these areas of genes has already been connected with intellectual disability. There is question of whether duplication of the same genes may also be connected in ASD.

What did the researchers do?

A case study was done on a female diagnosed with ASD. She also had concerns with anxiety and self-harming behavior, such as hair pulling.

What you need to know:

Autism Spectrum Disorder may be connected to issues in certain genes. The genes in question have been connected with intellectual disabilities and aggressive behavior. It is important to know if gene variation is a factor in ASD, as it can lead to other problems later in life.

A summary of the woman's medical history was outlined. Then DNA microarray analysis was used to identify any deletion or duplication in the sections of genes being explored. Quantitative Real-Time PCR (qRT-PCR) was performed to confirm the duplication. Finally, the woman's results were compared with both her parents and control groups.

What did the researchers find?

The analysis revealed only 1 significant copy number variation (CNV) in her genome. The CNV found was at Xp11.22-p11.23. The duplication was confirmed in the qRT-PCR. The duplication was not present in either parent. Control group results did not show signs of the duplication either.

How can you use this research?

The gene variation outlined may be connected with delays in a number of areas that will impact daily living. Although further research needs to be completed to confirm these results, medical staff may want to consider these concerns as a possible factor for a client's other issues.

About the Researchers

Dr. Stephen Scherer holds the GlaxoSmithKline-CIHR Pathfinder Chair in Genetics and Genomics at the University of Toronto and SickKids.

stephen.scherer@sickkids.ca

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Chung, B.H.Y., Drmic, I., Marshall, C.R., Grafodatskaya, D., Carter, M., Fernandez, B.A., Weksberg, R., Roberts, W. and Scherer, S.W., 2011. Phenotypic Spectrum Associated With Duplication of Xp11.22-p11.23 Includes Autism Spectrum Disorder. *European Journal of Medical Genetics, 54*(5), pp. e516-e520.

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Dup(X)(p11.22-p11.23), Chromosome X duplication, Intellectual disability, Autism Spectrum Disorder, Trichotillomania

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KT@neurodevnet.ca

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