

research snapshot

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Is SHANK1 Connected to Autism Spectrum Disorder in Men?

What is this research about?

Autism spectrum disorder (ASD) is a lifelong condition. Signs of ASD are problems with social situations and communication, and repetitive and restrictive behaviors. ASD is caused by a variety of gene and environmental factors. ASD is more commonly found in men than women. The reason for this disparity is not known.

The SHANK genes regulate how information moves throughout the brain by supporting the structure of the cell at the synapse. The SHANK 2 and 3 genes are connected to ASD. Errors in these two SHANK genes cause signal issues in the brain that contribute to ADS. This research explores if variations in the SHANK1 gene could be connected with ASD.

What did the researchers do?

1,954 gene samples were analyzed for variations in the SHANK1 gene. These samples were taken from 2 groups: people with ASD and people with intellectual disorders (ID). 1,614 of the samples were from people with ASD. 340 of the samples came from people with ID. The samples came from Canada and various countries in Europe. The samples came from unrelated people. 15,122 people made up the control group. The gene samples were analyzed with the use of

What you need to know:

This research shows that all SHANK genes are connected with ASD. Exploration of the SHANK1 gene and how it connects to the presentation of high functioning ASD in males is important. This research will help us understand how genes affect ASD and the connection between sex and ASD.

programs shown to provide reliable information on gene variations.

When variations in SHANK 1 were found, the rest of the family was assessed for ASD symptoms and SHANK1 gene variations. As well, background information about the family members was gathered when possible. These details are necessary, as this information helps to tell if gene variation in SHANK1 was inherited or not.

What did the researchers find?

The gene variations found in SHANK1 were deletions that also affected the neighboring genes. In one instance SHANK 1 deletions were found in multiple family members. In the other instance the deletions in SHANK1 were in only one member of the family, even though a sibling was also diagnosed with ASD. When

the two families were compared the variation in the SHANK1 gene was the only common factor. This finding supports the theory that SHANK1 could influence the presentation of ASD. Further support was found because there was a higher rate of SHANK1 variation in people with ASD than in the control group.

In the family where SHANK1 variations were found in 6 family members, ASD only presented in the 4 men with this deletion. The female members had anxiety, but this is believed to be an unrelated disorder. All males with the SHANK1 deletion were considered to have high functioning ASD. This is the first time that gene variations connected with ASD have only presented in one sex. This information will be important to understand the prominence of ASD in men.

How can you use this research?

Practitioners will learn that there is genetic evidence about a connection between a person's sex and the presentation of ASD. It is important to note that this connection has only been found for one type of variation in one gene.

Families will learn about how certain genes may put male family members at higher risk for ASD.

About the Researchers

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Citation

Sato, D., Lionel, A.C., Leblond, C.S., Prasad, A., Pinto, D., Walker, S., O'Connor, I., Russell, C., Drmic, I.E., Hamdan, F.F., Michaud, J.L., Endris, V., Roeth, R., Delorme, R., Huguet, G., Leboyer, M., Rastam, M., Gillberg, C., Lathrop, M., Stavropoulos, D.J., Anagnostou, E., Weksberg, R., Fombonne, E., Zwaigenbaum, L., Fernandez, B.A., Roberts, W., Rappold, G.A., Marshall, C.R., Bourgeron, T., Szatmari, P. and Scherer S.W., 2012. SHANK1 Deletions in Males With Autism Spectrum Disorder. *The American Journal of Human Genetics*, 90 (5), pp. 879–887.

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