research snapshot

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Is Autism Spectrum Disorder Connected to Deletions in the Neurexin 3 Gene?

What is this research about?

Current diagnostic tools for autism spectrum disorder (ASD) can be used as early as age 4. However, by age 4 an important window of intervention has passed. Thus, finding other ways to diagnose ASD will help take advantage of earlier interventions. Genetic testing is one of the diagnostic tools being considered. For genetic testing to be effective relevant gene variations must be known.

103 gene variations have been found in individuals diagnosed with ASD. Only 10% of ASD cases have been found to have clinically relevant gene variations. When finding what gene variations are relevant other nervous system issues must be considered. For example, intellectual disabilities (ID) and ASD have some gene variations that overlap. Thus, subjects should be tested for ID and ASD to ensure overlapping gene variations are accounted for.

The most common genetic test done when looking for gene variation is whole-exome sequencing (WES). This test is less expensive, but only considers part of the genetic information. More costly and exploring all genetic information is the whole-genome sequencing (WGS). Using WGS could help find more clinically relevant gene variations for ASD.

What you need to know:

Current knowledge of genetic risk factors for autism spectrum disorder only account for 10% of cases. To find other genetic risk factors for ASD a broader genetic analysis must occur. This paper presents a pilot project that begins to complete a broader analysis.

What did the researchers do?

32 people diagnosed with ASD were selected from the Autism Genetic Resource Exchange. Genetic material from these people and their families were analyzed. To be included there had to be one or more siblings without ASD and genetic material had to be available. Only 6 people with ASD had intellectual disabilities (ID), of the 30 ASD people tested for IDs.

What did the researchers find?

When a WGS was completed more gene mutations were found. This is likely a result of the extent WSG explores over WES. In the 32 cases studied, 16 were found to have an identified

genetic risk for ASD. The researchers suggest that using WSG instead of WES will help to decrease the time needed to diagnosis a person with ASD.

How can you use this research?

Policy makers will learn that costly tests can deliver better long term finances when earlier diagnosis occurs.

About the Researchers

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Citation

Vaags, A., Lionel, A.C., Sato, D., Goodenberger, M., Stein, Q., Curran, S., Ogilvie, C., Ahn, J.W., Drmic, I., Senman, L., Chrysler, C., Thompson, A., Russell, C., Prasad, A., Walker, S., Pinto, D., Marshall, C.R., Stavropoulos,D.J., Zwaigenbaum, L., Fernandez, B.A., Fombonne, E., Bolton, P.F., Collier, D.A., Hodge, J.C., Roberts, W., Szatmari, P., and Scherer, S.W., 2012. Rare Deletions at the Neurexin 3 Locus in Autism Spectrum Disorder. *The American Journal of Human Genetics, 90* (1), pp. 133-141.

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