

Newer genetic testing methods may provide benefit for children with suspected autism

1 September 2015



Quinn, an autistic boy, and the line of toys he made before falling asleep. Repeatedly stacking or lining up objects is a behavior commonly associated with autism. Credit: Wikipedia.

The use of two newer genetic testing technologies (chromosomal microarray analysis and whole-exome sequencing) among children with autism spectrum disorder may help identify genetic mutations potentially linked to the disorder, according to a study in the September 1 issue of *JAMA*. The study also found that children with certain physical anomalies were more likely to have genetic mutations, findings that may help identify children who could benefit most from genetic testing.

Autism spectrum disorder (ASD) represents a diverse group of neurodevelopmental conditions. The clinical presentation and outcome vary substantially. The use of genome-wide tests to provide [molecular diagnosis](#) for individuals with ASD requires more study, according to background information in the article.

Stephen W. Scherer, Ph.D., of the Hospital for Sick Children, Toronto, and colleagues performed chromosomal microarray analysis (CMA) and whole-exome sequencing (WES) in a group of 258 unrelated [children](#) with ASD to determine the molecular diagnostic yield (the percentage of subjects with a genetic alteration [mutation] that may contribute to the features of [autism spectrum disorder](#)) of these tests. All children underwent CMA; a random subset of 95 also underwent WES. All children underwent detailed clinical assessments for the presence of any major congenital abnormalities and minor physical anomalies and were stratified into 3 groups of increasing morphological severity (physical aberrations): essential, equivocal, and complex.

Of the 258 children, 24 (9.3 percent) received a molecular diagnosis from CMA and 8 of 95 (8.4 percent) from WES. The yields were statistically different between the morphological groups. Among the children who underwent both CMA and WES testing, the estimated proportion with an identifiable genetic cause was 15.8 percent. This included 2 children who received molecular diagnoses from both tests. The clinical yield for [genetic testing](#) was much higher (37.5 percent) in children with ASD who had more complex clinical presentations based on physical examination.

"In the current study, we have demonstrated differences related to morphological stratification of ASD [children] based on clinical examination. Our data suggest that medical evaluation of ASD children may help identify populations more likely to achieve a molecular diagnosis with genetic testing," the authors write.

"It seems likely that genetic testing of children with ASD will continue to increase. In a survey of parental interest in ASD genetic testing, 80 percent of parents indicated that they would want a sibling younger than 2 years tested to identify ASD-risk mutations even if the test could not confirm or rule

out the diagnosis. For some children with positive genetic test results, treatment plans targeting ASD-associated medical conditions can be offered."

The researchers conclude that if "replicated in additional populations, these findings may inform appropriate selection of molecular diagnostic testing for children affected by ASD."

"For ASD, as well as for other behaviorally defined disorders, the results reported by Tammimies et al provide clear guidance," writes Judith H. Miles, M.D., Ph.D., of University of Missouri Health Care, Columbia, in an accompanying editorial.

"Foremost, the data indicate that physicians responsible for children with ASD should arrange access to a genetic evaluation using techniques that have the best chance of determining an etiologic diagnosis. It is incontrovertible that precise diagnoses pave the way to better medical care, improved surveillance, better functional outcomes, and informed genetic counseling, often with the possibility of prenatal or preimplantation diagnosis."

More information: DOI:

[10.1001/jama.2015.10078](https://doi.org/10.1001/jama.2015.10078)

[DOI: 10.1001/jama.2015.9577](https://doi.org/10.1001/jama.2015.9577)

Provided by The JAMA Network Journals

APA citation: Newer genetic testing methods may provide benefit for children with suspected autism (2015, September 1) retrieved 27 September 2022 from

<https://medicalxpress.com/news/2015-09-genetic-methods-benefit-children-autism.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.