Benefits and drawbacks of using genomic discoveries for autism spectrum disorder

childhood disability LINK

Summary

Autism spectrum disorders (ASDs) are a group of lifelong developmental disorders that are characterized by problems with social interactions, communication, repetitive behaviors (e.g. hand flapping), and restricted interests. Many studies have found that the risk of developing ASDs is strongly influenced by a complex web of genetic and environmental factors. Even though individuals with ASDs show many of the same behaviors, the underlying causes for each case can vary significantly from one individual to another. Therefore, it is very difficult to develop genetic screening tests to reliably diagnose or predict the risk of passing on an ASD. As more information is being rapidly discovered on possible genetic links for ASD, the authors of this article urge caution in incorporating new findings into ASDs diagnostics before the results can be validated. This article is a summary of significant advances in evidence-based practices related to ASD diagnosis, characterization, and treatment. It includes a discussion on the scientific, ethical, policy, and communication aspects of translating new discoveries into clinical and diagnostic tools.

What families should know

Even though advances have been made in understanding some of the genetic factors influencing ASDs, for most forms of ASDs, genetic screenings cannot be used reliably for diagnosis or predicting inheritance. Still, some genetic tests (e.g. for SHANK2) can provide useful information about ASD risks to help start behavioral and/or medical interventions at an earlier age.

What practitioners should know

When communicating information about genetic influences and risks of ASDs to individuals with ASD and their families, be mindful to discuss not only what is known with some confidence but also any limitations in knowledge.

Reference

Scherer, S. W., & Dawson, G. (2011). Risk factors for autism: translating genomic discoveries into diagnostics. Human genetics, 1-26.

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