What do the results of these genetic tests mean?

In addition to aiding a provider in determining the appropriate treatment plan, genetic testing may help parents and families understand any genetic risks that may be present, and can therefore assist in planning discussions.

A positive test result means your child was found to have differences in his/her genetics compared to the genetics of children that do not have ASD. If your child's result is positive, talk with your health care provider about the test result and how it may be related to your child's ASD diagnosis. You may also want to discuss:

- Any changes to your child's medical management and treatment
- What these results might mean for future pregnancies or other family members

A negative test result means the testing performed did not find a genetic explanation that might contribute to your child's ASD diagnosis.

An uncertain result (commonly referred to as a variant of uncertain significance or a VUS) means that a genetic change was identified, but based on today's scientific knowledge, it is unclear if this change is actually having an effect. If your child's result is a VUS, talk with your health care provider about any recommended next steps.

Genetic results counseling for all whole exome tests are included when ordered through LabCorp.

These counseling sessions can be over the phone. You can schedule this counseling by going to https://www.integratedgenetics.com/genetic-counseling or call 1-855-422-2557 for more information.

Where can I find more information?

Autism Speaks
www.autismspeaks.org
1-888-AUTISM2 (1-888-288-4762)
familyservices@autismspeaks.org

National Institutes of Mental Health
www.nimh.nih.gov/health/topics/autism-spectrum-disorders-asd
1-866-615-6464
nimhinfo@nih.gov

References
1. When do children usually show symptoms of autism? | NICHD - Eunice Kennedy Shriver National Institute of Child Health and Human Development. Available at: https://www.nichd.nih.gov/health/topics/autism/conditioninfo/symptoms-appear. Accessed: October 7, 2019.
2. Schaefer GB, Mendelsohn NJ. Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions. Genet Med. 2013;15:399. doi: 10.1038/gim.2013.32.
3. Fragile X syndrome - Genetics Home Reference - NIH. Available at: https://ghr.nlm.nih.gov/condition/fragile-x-syndrome. Accessed: October 7, 2019.

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Genetic Testing for Autism Spectrum Disorder

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What Is Autism Spectrum Disorder (ASD)?

Autism Spectrum Disorder is a condition that arises during early childhood development that can affect communication and behavior. Signs and symptoms of ASD often occur when a child is developing and learning social skills, which is why it is a "developmental disorder". It is known as a "spectrum" disorder because there is a wide range of type and severity of symptoms that an individual may display.

What causes ASD?

As there are many different forms of ASD, there are also many possible causes. Doctors and scientists are still unsure of the exact causes of ASD, but it is known that those with ASD have brains that function differently. There is no evidence of vaccines causing ASD. Instead, ASD is likely a result of changes in brain development before birth while still in the womb. There are likely many different reasons ASD may develop in someone, including genetic causes.

How is ASD diagnosed?

ASD is diagnosed through a behavioral exam conducted by a licensed health care specialist. Autism can be diagnosed at any age with symptoms generally appearing during early childhood when a child should be developing and learning social skills. However, symptoms may appear even earlier as many children show signs of autism by 12 months to 18 months.[1]

Why is my child's healthcare provider ordering genetic testing?

Even though ASD is diagnosed through behavioral testing, many parents are still hoping to find an explanation for why their child has autism. Genetic testing may also:

- Provide information about what to expect in the future as a child grows and develops
- Lead to changes in the child's medical care and future treatment options, or participation in clinical trials
- Provide information that may help secure funding for services such as medical therapies or school programs
- Help families understand the chance that autism could also affect a future child
- End the search for a cause, eliminating the need for further genetic testing
- Empower and connect families with similar forms of ASD

Visit https://www.labcorp.com/asd for more information about the testing your provider may have ordered.

What types of genetic testing might be ordered?

There are a few primary tests that healthcare providers use to assess possible genetic contributions to ASD. These tests are:

- Fragile X testing
- Chromosome testing
- Whole exome sequencing (WES)

What is Fragile X testing?

As many as 5% of individuals with ASD have Fragile X syndrome.[2] Fragile X syndrome is caused by a changes in the FMR1 gene and is passed from mother to child. Fragile X syndrome may cause speech and language delays, as well as behavioral difficulties such as hyperactivity. Many people with fragile X have similar physical features such as large ears and a long face.[3]

What is chromosome testing?

Chromosomes are the structures inside the cells of our bodies that store DNA. Most people have 46 chromosomes, 23 inherited from their mother, and 23 inherited from their father. Chromosome disorders vary widely in severity, but many chromosome disorders are associated with ASD.[2]

Chromosome testing uses advanced technology to determine if your child has an extra or missing chromosome, if part of a chromosome is missing (called a deletion), or if an extra part of a chromosome is present (called a duplication). Some chromosome disorders are inherited from a parent, while others happened only in the child's DNA and did not come from the mother or father.

What is Whole Exome Testing?

Since there are many genetic conditions associated with autism, some doctors recommend whole exome sequencing (WES) to test for as many possible genetic conditions at the same time. WES examines every gene in a child's DNA looking for genetic changes, sometimes called variants, which may be contributing to ASD.

It is helpful to submit samples for the child and both biological parents for WES testing. Any genetic changes found in the child can then be compared to the parents' DNA. DNA changes seen only in the child are more likely to be a cause of the child's condition.