Ambry Genetics®
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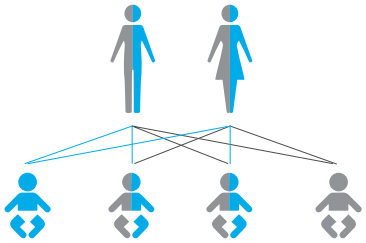
Understanding Your AutismNext Carrier Genetic Test Result

INFORMATION FOR PATIENTS WITH **ONE PATHOGENIC MUTATION** OR **VARIANT LIKELY PATHOGENIC**

Result	**CARRIER**	The testing done shows that you/your family member has one pathogenic mutation (a disease-causing change in the gene, like a spelling mistake) or variant that is likely disease-causing, in a gene that causes an autism spectrum disorder (ASD). This means you/your family member is a "carrier" for an ASD. It is unlikely that having only one mutation in this gene causes an ASD.
Gene	**DEFINITION**	Genes are instructions for how our bodies work and develop. Everyone has two copies of each gene, one from each parent. A mutation (change in the gene, like a spelling mistake) in both copies of certain recessive genes can cause ASDs. Carriers have one mutation in a copy of their gene. Carriers do not usually have the symptoms of an ASD, but can have children with an ASD.
Screening Options	**FAMILY MEMBERS**	If a carrier's partner is also a carrier of a mutation in the same gene, there is a 1 in 4 (25%) chance to have a child with a ASD in each pregnancy. Genetic testing for a carrier's partner may help both learn more about this risk. In addition, other adult family members may wish to be tested to see if they carry the mutation found in your family.
Next Steps	**DISCUSS**	It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers. They can be tested for the same mutation, if they choose to.
Reach Out	**RESOURCES**	• Ambry's Neurology Site for Families patients.ambrygen.com/neurology • Autism Speaks autismspeaks.org • Talk About Curing Autism tacanow.org • Autism Society autism-society.org • National Autism Association nationalautismassociation.org • National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca

How Recessive ASDs Are Inherited

For some ASDs, people who have a mutation in only one gene are "carriers" and do not usually have symptoms. If they have children with a partner who is a carrier for a mutation in the same gene, there is a 1 in 4 (25%) chance for them to have a child with an ASD in each pregnancy together. There is a 2 in 4 (50%) chance for them to have a child who is a carrier (usually without symptoms), and a 1 in 4 (25%) chance for them to have a child who does not have an ASD, and is not a carrier.



- ASD carrier (one gene mutation)
- Does not have ASD, and is not a carrier (no gene mutations)
- Has ASD (two gene mutations)

Please discuss this information with your healthcare provider. The field of genetics is continuously changing, so updates related to your genetic testing result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, avnd should not be considered or taken as medical advice.

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One Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1 866 262 7943 Fax +1 949 900 5501 ambrygen.com