Ambry Genetics®

Understanding Your Carrier Chromosomal Microarray Test Result

INFORMATION FOR PATIENTS WITH **WITH ONE PATHOGENIC OR LIKELY PATHOGENIC COPY NUMBER VARIATION INVOLVING AN AUTOSOMAL RECESSIVE GENE**

Chromosomes and Genes	Chromosomes are packages of DNA. They are made up of genes that provide instructions for how our bodies develop. Almost everyone has two copies of each chromosome, one from each parent. Variants (changes) in our genes or chromosomes can cause genetic conditions. These changes may be passed down in families or be brand new for the person who has a genetic condition. Even if there is no history of the specific condition in your family, it can still be caused by a change in a person's genes or chromosomes.
SNP Array	Single nucleotide polymorphism microarray (SNP Array) testing is a specific type of genetic test used to find changes in a person's chromosomes. These changes are called copy number variations (or CNVs). CNVs are changes that involve extra ("gain") or missing ("loss") of a person's chromosomes. SNP Array testing is not designed to find all types of genetic changes in a person's DNA, only CNVs.
Result	The SNP Array testing found a CNV that is either pathogenic (known to be disease-causing) or likely pathogenic (likely to be disease-causing) involving a gene that is known to cause a recessive condition. This means the person who was tested is a "carrier" for a genetic condition. The test result is unlikely to explain the genetic cause of the symptoms of the person who was tested.
Further Testing	Talk with your healthcare provider to learn more about whether you or your family member might have other genetic test results still pending or need additional genetic testing.
Family Members	Often, a CNV can be found in someone even if it was not passed down from a parent. In other families, the CNV can be inherited from earlier generations. Talk to your healthcare provider about what this test result means for the rest of your family. Your family members may also be carriers for the same genetic condition. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

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, and should not be considered or interpreted as medical advice.

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