Ambry Genetics®

Understanding Your Positive Chromosomal Microarray Test Result

INFORMATION FOR PATIENTS WITH **ONE OR MORE PATHOGENIC** OR **LIKELY PATHOGENIC COPY NUMBER VARIATIONS**

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). Either type of CNV should be treated as a positive result. The test result is likely to explain the genetic cause of the symptoms of the person who was tested.
Management Options	Management options vary by condition and other factors. Knowing the genetic cause of the symptoms may also help avoid other tests or procedures. Talk to your healthcare provider about which treatment options may be helpful.
Family Members	Many people with a genetic condition are the first person in their family to have it. 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 how the specific genetic condition may run in your family based on your test results and family history, and what this means for the rest of your family. 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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