Ambry Genetics®

Understanding Your Chromosomal Microarray Test Result

INFORMATION FOR PATIENTS WITH **ONE OR MORE REGIONS OF HOMOZYGOSITY**

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 did not find any CNVs that are known to be disease-causing. However, the testing did find one or more regions of homozygosity (ROH). Typically, the chromosome a person inherits from one parent is slightly different from the chromosome inherited from the other parent. ROHs are when sections of a person's chromosomes are identical on both copies. ROHs are not expected to be disease-causing on their own, but they do increase the chance that the person has a recessive genetic condition.
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.
Management Options	Management options vary by condition and other factors. Talk to your healthcare provider about which treatment options may be helpful.

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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