

A chromosomal microarray (CMA) is a test that can be done to look for specific kinds of chromosome changes called <u>microdeletions</u> and microduplications.

Much like a traditional chromosome analysis, CMA generally looks at all 23 pairs of chromosomes. While the chromosome analysis is looking for large changes (extra or missing pieces of chromosomes, or extra or missing whole chromosomes), the CMA testing can look for much smaller <u>missing</u> or <u>extra</u> pieces of chromosome material.

It can be difficult to understand how each test is similar and different, so let's look at it a different way. Imagine a bookshelf full of books. A traditional chromosome analysis would be like looking at the shelf, and figuring out if there are any extra or missing books, or if the books or in the wrong order. CMA testing would be like taking a book off of the shelf and looking through it to see if there are any missing or extra pages. Traditional chromosome analysis can find large differences, while CMA can take a more in-depth look.

CMA testing can be done for a number of reasons, including for children and adults who may have an underlying genetic condition, but it's not clear what that genetic condition is. Each genetic condition is related to specific genes. If we have an idea of what genetic condition someone might have, then we can offer them testing that is more targeted to what we think is most likely causing it. If someone has health issues that could have a genetic cause but are not specific (some examples may include <u>developmental</u> and <u>intellectual</u> delays, or autism spectrum disorder), CMA testing may be used to take a wider look at a patient's chromosomes to try to determine if an underlying genetic cause can be found.

CMA testing can also be done during pregnancy; click <u>here</u> to read more.

Because test results from CMA testing may not necessarily be straightforward, it is important to talk with healthcare provider who is familiar with CMA testing who can explain more about the risks, benefits, and limitations of the testing.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for pregnancyrelated questions.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.



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A chromosome analysis, or karyotype, is the traditional testing that can been done on cells from chorionic villus sampling (CVS) and amniocentesis procedures, or from a blood sample. A karyotype involves the lab examining the cells and determining the number of chromosomes. How the lab determines the number of chromosomes...

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With genetic testing, there are generally three different results that we can get: a positive, a negative, or a variant of uncertain significance (VUS). Positive A positive result means that a harmful change (called a pathogenic variant) was found in a gene that causes that gene to not work properly....

How to Decide

Whether or not to pursue any genetic testing is a very personal decision. In some cases, moving forward with genetic testing may help to provide an answer to a health question that is running in someone's family, or may help to provide a name to a medical condition that someone...