

# Chromosomal Microarray: Test Information for Families

## What is a chromosomal microarray?

A chromosomal microarray is a genetic test offered to children and adults who have developmental delay, multiple birth defects or autism spectrum disorders. It is sometimes referred to as “microarray” or “array CGH”.

Chromosomal microarrays have the ability to find small changes in genetic material (DNA). It is more detailed than a regular chromosome test (karyotype). It specifically looks for extra and/or missing pieces of DNA. Sometimes, extra and/or missing DNA happens by chance and sometimes it can be inherited. These DNA changes may affect a person’s health and development. It is estimated that 10-15% of individuals with developmental delay may have an abnormal chromosomal microarray result.

## What are the possible test results? What happens next?

1. The chromosomal microarray did not find any missing or extra pieces of DNA
  - This means that your/your child’s health or developmental concerns cannot be explained with this test.
  - This test does not look for all possible genetic changes. It does not rule out the possibility that your/your child’s concerns are genetic. Your Health Care Provider may or may not recommend additional genetic testing.
2. The chromosomal microarray found missing and/or extra pieces of DNA that are seen in the general population and are unlikely to explain your/your child’s health or developmental concerns
  - This means that your/your child’s health or developmental concerns cannot be explained with this test.
  - This test does not look for all possible genetic changes. It does not rule out the possibility that your/your child’s concerns are genetic. Your Health Care Provider may or may not recommend additional genetic testing.
3. The chromosomal microarray found missing and/or extra pieces of DNA that are likely an explanation for your/your child’s health or developmental concerns
  - This means that your/your child’s health or developmental concerns can be explained with this test.
  - A Geneticist may be able to provide you with additional information regarding your /your child’s genetic diagnosis.
  - Your Health Care Provider may offer additional blood work to parents to learn if the extra and/or missing pieces of DNA were inherited.
4. Uncertain Test Result: The chromosomal microarray found missing and/or extra pieces of DNA and it is unknown if this explains your/your child’s health or developmental concerns
  - This is referred to as a “Variant of Unknown Clinical Significance”.
  - Before your next appointment, both parents will be offered additional blood work to determine if the extra and/or missing pieces of DNA were inherited as this may help us understand your /your child’s test results.

\*\*There is a small chance that co-incidental findings not related to your/your child’s condition may be identified. These findings will be discussed with you if the results could have a significant impact on your/your child's health.