



Microarray Analysis Test

Your doctor has ordered a genetic test called a microarray (MY cro a ray) analysis. This test is to find out if your child has a medical condition caused by a missing or extra piece of chromosome material. This test is also known by several other names, such as chromosomal microarray, whole genome microarray, array comparative genomic hybridization (array CGH or aCGH) or SNP microarray.

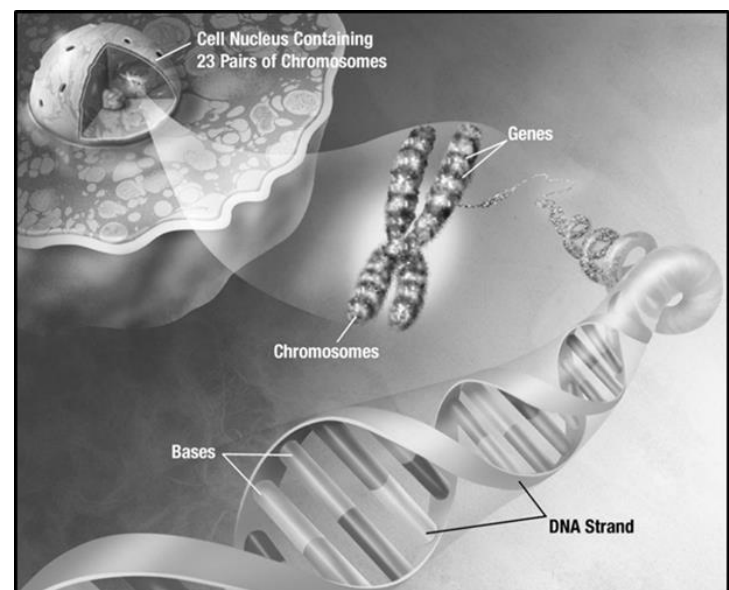
Chromosomes – what they are

The human body is made up of billions of building blocks called cells. Inside each cell are thousands of genes, made of DNA, that tell the body how to grow and develop. These genes are arranged one after another on structures called chromosomes. Each cell normally contains 46 chromosomes that are arranged in 23 pairs. The first 22 pairs are numbered 1 through 22 based on size. These chromosomes are the same in both males and females. The 23rd pair of chromosomes is called the “sex chromosomes” because they determine the person’s genetic gender. We get our chromosomes from our parents and pass them on to our children. One chromosome from each pair comes from the mother, while the other chromosome in the pair comes from the father. When the mother and father are not related to each other by blood, the chromosomes from the mother and father are usually different from each other. However, when the mother and father are blood relatives, many parts of the chromosomes are genetically identical.

What this test can tell me

This test can find out if a person has missing or extra chromosome material. Missing chromosome material is called a “chromosome deletion.” Extra chromosome material is called a “chromosome duplication.”

When a person has a missing or extra piece of chromosome material, it can cause health problems such as birth defects, seizures, delays in development, and autism. However, some missing or extra pieces of chromosome material cause no problems.



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What this test can tell me, continued

Missing or extra pieces of chromosome material can be passed down from a parent, or can happen as a new event in a child. Microarray analysis may also detect large parts of a chromosome that are genetically identical. Having genetically identical chromosome parts could mean that a person's parents are blood relatives or have a common ancestor. It may also mean that a person got both copies in the chromosome pair from a same parent rather than one copy from each parent. Having genetically identical chromosome part(s) can increase the chance of certain genetic disorders in that person.

How the test is done

Microarray analysis is usually done on a blood sample. In the laboratory, the DNA (chromosome material) is taken from the cells in the sample and compared to a normal DNA sample to see if there are any missing or extra pieces of chromosome material. The microarray analysis also compares the DNA of each chromosome pair to see if any large parts are genetically identical. It can take up to 4 weeks to get results.

Types of results from this test

- **Normal result:** no missing or extra pieces of chromosome material were found. No genetically identical large chromosome parts were found.
- **Likely pathogenic:** a missing or extra piece of genetic material was found. This is likely to cause health or learning problems. Your child's doctor will talk to you about how this result will affect your child's health.
- **Likely benign:** a missing or extra piece of chromosome material was found, but it is not likely to cause health or learning problems.
- **Variant of unknown significance (VUS or VOUS):** a missing or extra piece of chromosome material was found, but it is not clear if this causes any health or learning problems. Sometimes testing the parents may be recommended to see if this missing or extra piece came from a parent. This information can be helpful to find out if this may be the cause of your child's health or learning problems.
- **Regions of Homozygosity (ROH):** one or many chromosome parts were found to be genetically identical. This may mean that your child's biological parents are related by blood (share a common ancestor) or that your child got two copies of the same chromosome from the same parent, rather than one copy from each parent. Although this does not always cause health or learning problems, it may increase the chance of certain genetic disorders. The results may lead to changes in health care or a discussion about risks for future pregnancies. The results also may be helpful for other family members. Your child's doctor may recommend other genetic testing for your child or other family members depending on these results.

After the test

- After the blood draw, you may take your child home.
- These results can take up to 4 weeks to come back. After the test results come back, your child's doctor will talk with you about the results.

If you have any questions, please be sure to ask your child's doctor who ordered this test. If your doctor has questions about this test or the results, he or she can call Laboratory Client Services at 800-934-6575 and ask to speak with a Laboratory Genetic Counselor.

For directions to the nearest Laboratory Service Center, please call Laboratory Services at (800) 934-6575 or visit our website at NationwideChildrens.org/Lab.