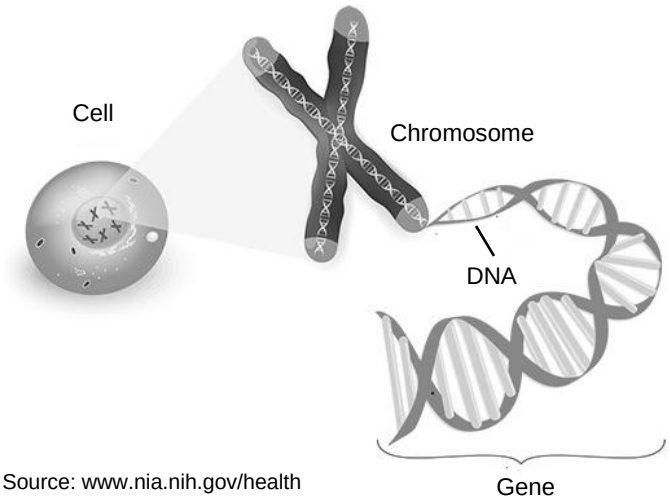
Microarray Analysis Test

Your health care provider has ordered a genetic test called a microarray (MY-cro-a-ray) analysis. The test is also known as chromosomal microarray, whole genome microarray, array comparative genomic hybridization (array CGH or aCGH) or SNP microarray. This test helps to find out if your child has a medical condition caused by:

- a small missing piece of chromosome, called a deletion
- a small extra piece of chromosome, called a duplication
- the entire chromosome pair being passed down from one parent
- large parts of multiple chromosomes being the same

This test does not check for every possible genetic disease or give information about a specific gene.



Source: www.nia.nih.gov/health

Picture 1 Chromosomes hold thousands of genes. Genes tell our body how to grow and work.

Chromosomes

The body is made up of billions of cells. Inside each cell are chromosomes. Chromosomes are structures that contain thousands of genes (Picture 1). Genes are made up of DNA. They tell the body how to grow and work. Genes also hold information about traits, such as our eye and hair color and blood type.

Each cell normally has 46 chromosomes that are arranged in 23 pairs. To form the pair, one chromosome comes from the mother and the other comes from the father.

In the first 22 pairs, female and male chromosomes should match up in size and shape. The last chromosome pair is called the sex chromosome because it tells what genetic gender a person is born with. A female has two copies of the X chromosome. A male has one copy of the X chromosome and one copy of the Y chromosome.

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How the test is done

A blood sample is preferred for microarray analysis. Sometimes spit (saliva) or a tissue sample (like skin) may be used. This test compares the patient's sample to a normal control sample to find very small missing or extra chromosome pieces that cannot be seen under a microscope. The test does not show structural changes in chromosomes. It can take up to 4 weeks to get the test results.

Why do the test

- The test's results may lead to:
  - finding the genetic cause for your child's medical condition.
  - changes in your child's health care.
  - learning the risk for your child to pass down a genetic change to their children.
  - knowing the risk for future pregnancies when both parents and child are tested.
- When a person has a missing or an extra piece of chromosome:
  - It can cause health problems, such as birth defects, seizures, delays in development, learning problems and autism.
  - Sometimes there are no problems.

  With a missing or extra piece of chromosome, the doctor may also want to test parents and other family members. This can help find out if the change was passed down from a birth parent or if it is something new that happened in the child by chance.
- When chromosome part(s) are alike or the entire chromosome pair is the same:
  - It can increase the chance of certain genetic disorders that cause health problems.
  - Sometimes there are no problems.

  Usually, chromosomes in a pair are different since one copy comes from each parent. If a large part of one chromosome pair is the same, it could mean that both chromosome copies came from the same parent. When many parts of the chromosomes are the same, it could mean that parents are related by blood in some way.

Types of test results

- **Normal:** There are no missing or extra pieces of chromosomes, and no large parts of the chromosomes are the same.
- **Likely pathogenic:** There is a missing or extra piece of chromosome that may cause health or learning problems.

- **Variant of unknown significance (VUS):** There is a missing or extra piece of chromosome, but it is not clear if it will cause any health or learning problems. Finding a VUS is common.
- **Regions of Homozygosity (ROH):** One or many chromosome parts is genetically the same.

Your health care provider will talk to you about the test results. Sometimes more genetic testing will be ordered.

If you have any questions, please ask the health care provider who ordered this test or your genetic counselor.

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