Chromosome Analysis Test

Your child’s doctor has ordered a genetic test called a chromosome (CRO mo soam) analysis. The test is done to find out if your child has a medical condition called a chromosome abnormality. This means a person has a different amount or structure of genetic material in each of his or her cells.

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.

Genes tell the body how to grow and develop. The genes are arranged one after another on structures called chromosomes.

Each cell usually contains 46 chromosomes, 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.

What this test can tell me

This test can tell if a person has normal-appearing chromosomes or a chromosome abnormality. A chromosome abnormality can be:

- An atypical number of chromosomes (a number greater or less than 46 chromosomes)
- Chromosome(s) with missing or extra material (genes)
- Chromosome(s) with the correct amount of material (genes) but in an atypical structure

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What this test can tell me, continued

When a person has an atypical number of chromosomes or missing or extra chromosome material (genes), it may cause health problems such as birth defects and delays in development.

When a person has one or more chromosome(s) with an atypical structure, but the correct amount of chromosome material (genes), that person can have a higher chance to have pregnancy losses or to have a child with missing or extra chromosome material.

Some chromosome abnormalities are passed down from a parent, while some happen to a person as a new event.

How the test is done

Chromosome analysis is usually done on a blood sample. In the laboratory, the cells from the sample are placed in several special chemicals. These chemicals help the cells grow in number by dividing. That allows the chromosomes to be seen under a microscope. Also, the chromosomes are colored with a dye to give them a striped or banded look. This helps specially trained genetic lab technicians tell the chromosomes apart under the microscope.

Next, a camera attached to a microscope is used to take a picture of all the chromosomes in one cell. The chromosomes are matched into pairs and organized in order of size using a computer imaging program. This arrangement of chromosomes is called a karyotype (CARE o type). (See Picture 2.) It may take up to 3 weeks for the cells to grow and be examined under a microscope.

A laboratory director with special training in chromosome testing examines the karyotype for correct number, size, shape, and band pattern of the chromosomes. A person may have a chromosome abnormality if there is an atypical number of chromosomes, if a chromosome(s) has extra or missing material, or if a chromosome(s) has an unusual structure. The results may lead to changes in health care or a discussion about risks for future pregnancies. The results may also be helpful for other family members.

Please note, chromosome analysis can only find changes that can be seen under a microscope. Depending on these results, your child’s doctor may recommend other genetic testing.
After the test

- After the blood draw, you may go home.
- These results can take up to 3 weeks to come back. After your child’s doctor gets the test results, the 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 the doctor has questions about this test or the results, he or she can call Laboratory 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.