Chromosome Analysis

Does this test have other names?

Genetic testing, karyotyping

What is this test?

This test looks for changes, or abnormalities, in the chromosomes that make up your body's DNA, or genetic road map.

Your chromosomes are found in the inner part of your cells, called the nucleus. They contain all the genes that have been passed down to you from your mother and father.

A person typically has 23 pairs of chromosomes in each cell (23 pairs = 46 total chromosomes). One of these pairs is called the sex chromosomes (X and Y). They determine if you will be male or female. If you are male, you have an XY pair. If you are female, you have an XX pair. The other 22 pairs are called autosomes, and they are the same in males and females.

Cells for chromosome analysis can come from a blood sample, from inside a bone (bone marrow sample), from a swab of cells taken from inside your mouth, or from a sample of your skin or hair. Cells can also be taken from the fluid that surrounds a baby inside a mother's uterus (amniocentesis). Or they can be taken from a small piece of the placenta (chorionic villus sampling).

Cells taken for chromosome analysis are sent to a lab. Here they are prepared and arranged in order from largest to smallest. By looking at your chromosomes under a microscope and taking pictures of them (karyotyping), lab specialists may be able to tell whether or not you have any extra or missing chromosomes or pieces of chromosomes.

Abnormalities in your chromosomes help healthcare providers diagnose many health conditions. In some cases, your chromosomes can help your healthcare provider predict a health problem before you even have symptoms. Chromosome studies done during a pregnancy may predict problems that a baby may be born with or develop later in life.

Why do I need this test?

You may need this test for a variety of reasons, from helping to diagnose disease to finding out whether you have any changes in your genes that may be passed on to your children. Here are some reasons to have this test:

- To help diagnose or plan treatment for a disease
- To find out your risk of developing a disease you may have inherited
- To find out whether you carry a genetic change that may be passed on to your children
- To find out whether your unborn child may have a genetic problem
- To diagnose a genetic problem in a newborn or young child
- To find out why you are having trouble getting pregnant
- To find out why you are having miscarriages, or losing a baby before birth

What other tests might I have along with this test?

Your healthcare provider may also order other types of genetic testing that look for certain genes within your chromosomes. For example, you may have a test called fluorescence in situ hybridization (FISH) analysis, which looks at specific parts of your chromosomes. You may also have a more detailed chromosome test called a chromosomal microarray. This type of test looks at the chromosomes very closely for small extra or missing pieces. Other testing may look at the spelling (sequencing) of one or more genes. You may also have a blood test to look for abnormal proteins that might be a sign certain genes aren't working the way they should.

What do my test results mean?

Test results may vary depending on your age, gender, health history, and other things. Your test results may be different depending on the lab used. They may not mean you have a problem. Ask your healthcare provider what your test results mean for you.

Normal chromosomes are reported as:

- 44 autosomes plus 2 X chromosomes for a woman (karyotype 46,XX)
- 44 autosomes plus 1 X and 1 Y chromosome for a man (karyotype 46,XY)

Your healthcare provider will get a report from the lab that explains any abnormalities found in your or your child's chromosome analysis. A karyotype picture may also be included in the report.

Your healthcare provider should arrange for you to talk with a certified genetic counselor who can help you understand the results of this test.

How is this test done?

This test needs a sample of your cells. Your healthcare provider can get the cells in many ways. These are some options:

- Taking a blood sample by putting a needle into a vein in your arm
- Taking a blood sample from a newborn by making a small prick on the baby's heel
- Taking a blood sample from the umbilical cord of the fetus
- Taking out a sample of cells from the spongy center of a bone, called the bone marrow. Bone marrow cells can give healthcare providers important information about blood cancers like leukemia and lymphoma.
- Taking a sample of amniotic fluid by putting a long, thin needle through the pregnant woman's skin and into the fluid that surrounds the growing fetus
- Taking a sample of the placenta during the first trimester of pregnancy (chorionic villus sampling)
- Taking a small piece of tissue. This is called taking a biopsy.
- Taking a swab of cells from inside your cheek

Does this test pose any risks?

Each method of taking a sample carries certain risks. Some have more risks than others. Ask your healthcare provider to discuss all the risks and benefits with you before your test.

What might affect my test results?

Depending on how your test is done, certain things may affect your results. Ask your healthcare provider to explain these possibilities to you.

How do I get ready for this test?

Some chromosome analysis tests, such as amniocentesis, bone marrow sampling, or a tissue biopsy, do need special preparation. Ask your healthcare provider how you should prepare for your test. Blood sampling or cheek swabs usually don't need any preparation.