

Scott & White: Understanding Cytogenetic Testing

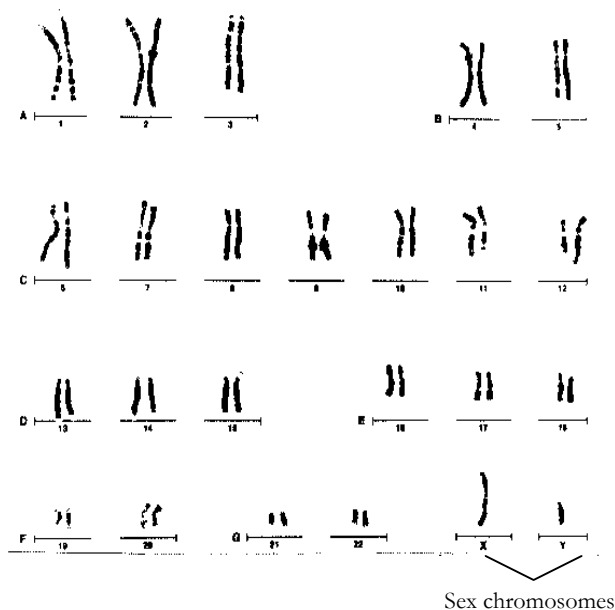
Do you or a family member fit into one of these categories?

- ▶ 3 or more miscarriages
- ▶ History of multiple congenital anomalies
- ▶ Family history of chromosome abnormality
- ▶ Diagnosis of Cancer (specifically leukemia, lymphoma, and breast cancer)

If so, your doctor may be considering ordering a chromosome test. This brochure is designed to give you some basic information on chromosomes and chromosome testing.

What are chromosomes?

- ▶ Chromosomes are the structures within cells that contain your body's DNA (genetic information). This genetic information tells your body how to grow and develop. When we look inside a cell, we expect to see 46 chromosomes in 22 pairs of autosomes and 1 pair of sex chromosomes. Below is a picture of a male's chromosomes (X and Y). A female would have two X chromosomes and no Y chromosome.



- ▶ Banding of the chromosomes is caused by chemicals and allows for the matching of chromosomes under the microscope. Each band contains 100-1000 genes, but changes in single genes cannot be seen under the microscope.

What does the term “cytogenetics” mean?

- ▶ Cytogenetics is a term for the field of science that studies the structure of chromosomes. The lab that performs chromosome tests is called the cytogenetics lab.

What kind of sample is needed to look at my chromosomes?

- ▶ Different kinds of samples are needed for different indications. Chromosome tests can be run on blood, bone marrow, amniotic fluid, and tissue.
- ▶ The indication for your testing dictates what kind of sample the doctor sends to the cytogenetics lab. Please ask your doctor what type of sample is required in your case.

Can you tell if I have a genetic disorder like cystic fibrosis or sickle cell disease by looking at my chromosomes?

- ▶ No, changes in single genes can only be tested for by a DNA test.

What is FISH testing?

- ▶ Your doctor may have ordered a special test called FISH testing. FISH stands for Fluorescent In Situ Hybridization. This testing can detect small changes in the chromosomes that can be missed sometimes by standard cytogenetic techniques.

Are there any special preparations for the chromosome test?

- ▶ No, you are not required to fast or take any special preparations for this test.

What happens if my chromosome test comes back abnormal?

- ▶ Your doctor will explain your test results to you or may refer you to a genetic counselor for a more in depth discussion. Chromosome test results can help doctors make a diagnosis, choose treatment options, and/or decide what further testing is needed.