



Chromosome Analysis: What it is & What it Tells You ©2002

What is a chromosome and how are they inherited?

In the nucleus (center) of a person's cells, deoxyribonucleic acid (DNA) is chemically bound to protein molecules and organized into 23 pairs of distinct, physically separate, microscopic units called chromosomes. Chromosomes are the structures in our cells which contain our genes. Genes code for traits and characteristics of a person such as their blood type and eye color.

Typically human cells have a total of 46 total chromosomes or 23 pairs. We inherit one member of each pair from our biological mother, the other member of each pair from our biological father. The first 22 pairs of chromosomes are called "autosomes" and the last pair are called the "sex chromosomes". Females have 2 X sex chromosomes, while males have one X and one Y sex chromosome.

Mature egg and sperm cells originate from precursor cells which contain 46 total chromosomes. When maturing, egg and sperm cells undergo a unique cell division process called "meiosis", where they divide in half. The result is a mature egg or sperm which contains 23 chromosomes each. In this way, each parent donates one half of their genetic material to a child. The gender of the child depends on which sex chromosome is inherited from the father. The mother can only give an X sex chromosome. If the father gives an X, the child is a girl, while if the father gives the Y, the child will be a boy.

What is a chromosome analysis?

Chromosome analysis is an analysis of the number and structure of all 46 chromosomes. The accuracy of a chromosome study is 99.9%. (Microdeletions or single gene abnormalities, which are not visible on a chromosome study, will not be detected.) Chromosomes may be studied from a blood sample, skin biopsy or other tissue. It is estimated that there are about 400 different types of chromosome abnormalities that have been described in humans. They fall into 2 categories: **numerical** and **structural**. **Numerical** chromosome abnormality simply means that a person has a total number of chromosomes different than 46; usually 47 or sometimes 45 chromosomes, in each cell of their body, respectively. Health problems and birth defects are usually present as a result of having the extra or missing genetic material.

The other type of chromosome abnormality, **structural**, may/may not result in obvious health problems. It depends on whether the structural problem results in a net gain or loss of chromosome material. If the chromosome material is simply in a rearranged fashion, yet there are still 46 total chromosomes, the person tends to have no clinical consequences from the rearrangement. However, this type of chromosome rearrangement can cause an increased chance for pregnancy losses or infants born with birth defects - because the rearrangement interferes with meiosis. The result after meiosis and fertilization is that there can be too much or too little genetic material in the zygote (fertilized egg). The pregnancy is "unbalanced" chromosomally, and may miscarry or result in the birth of a child with health problems. About 1 in 500 persons in the general population carry a rearrangement in their chromosome material. Persons with family or personal histories of multiple pregnancy losses, unexplained stillbirths, or early infant deaths, may be at a slightly greater chance to have a rearrangement in their chromosomes.

Will a chromosome analysis pick up all possible defects?

No. There are roughly 35,000 genes contained on the 46 chromosomes. **A chromosome study has nothing to do with looking at the individual genes.** For example, even when both parents have normal chromosomes and the baby has a normal chromosome study on an amniocentesis, there is still a 2-3% chance for the baby to have a birth defect.

How can we be studied to know which genes we carry that might give us a higher chance to have a child with a birth defect?

There is no such study at present. It is estimated that humans carry 5-50 deleterious recessive genes, which when passed on by both parents to their child simultaneously, result in a genetic condition, such as Cystic Fibrosis, Tay Sachs or Sickle Cell Anemia. Couples are screened for these types of genes based on their family history and ethnic background. Disease causing genes have different frequencies in different areas of the world, and this is why ancestry is important. A person who is Ashkenazi Jewish for example, has a 1 in 30 chance to carry the gene for Tay Sachs, while a person who is not Ashkenazi Jewish has a 1 in 300 chance to carry this gene.

The Human Genome Project has mapped the location of all of our genes on the chromosomes. The completion of this project, however, does not mean that there is testing for all disease causing genes. In fact, many tests currently available for single gene disorders are only informative when a relative has the disease in question (i.e. their DNA is studied and then compared to yours). With a negative family history for these types of problems, recommendations for carrier testing is based on ethnic background.

It is important to understand the difference between a chromosome study and a separate DNA or biochemical test for the presence/absence of a specific gene. Unfortunately at present, there is no genetic test that screens for everything.