

Genetics is a vast and increasingly complicated field of science. The information here will help to provide a basic background in general genetics concepts, and can be a resource as you come up with questions.

## We'll start with the basics: The birds and the bees and how we pass on our genes....



Our bodies are made up of billions of cells. We have skin cells, heart cells, brain cells, etc. These cells all have special functions in our bodies, but one thing they have in common is that each cell has a set of 46 chromosomes, or 23 pairs. The first 22 pairs of chromosomes are the same in men and women and the 23rd pair is different. Women have two 'X' chromosomes for their 23rd pair, and men have one 'X' chromosome and one 'Y' chromosome (see picture to the right of a set of male/female chromosomes, also called a karyotype).

Along the chromosomes are the individual instructions, or genes, that tell our bodies how to grow and function, including things such as hair color, eye color, and height. Since we have two copies of every chromosome, we also have two copies of every gene. All in all it is estimated that we each have about 20,000 pairs of genes, one set form our mother and one set from our father. Males only have one X chromosome though, so they only have one copy of all of the genes on the X chromosome, and one copy of the genes on the Y chromosome.

Genes are specific instructions which contain our genetic code, or DNA (deoxyribonucleic acid). DNA is made up of four similar chemicals, called bases: adenine (A), thymine (T), cytosine (C), and guanine (G). The specific order of these letters (bases) is what makes the

instructions (genes) work properly. If one or more of these letters is changed, deleted, or duplicated, and this change causes the gene to not work as usual, this is called a mutation. Mutations in our genes may lead to genetic conditions in ourselves or other family members.

In order for our bodies to grow and function as usual, it is important that we have the typical 46 chromosomes. It is also critical that we have only two of each. Many of the genetic conditions that are discussed in prenatal genetics are due to a baby having an extra (called a trisomy) or missing (called a monosomy) chromosome. An important thing to remember is that we have no control over how many chromosomes are packaged in our egg or sperm cells. If a baby has an extra or missing chromosome, there is nothing that was done to cause it and nothing that could have been done to prevent it.

Other types of genetic conditions involve changes in the instructions (genes) that are on the chromosomes. Some of the more common genetic conditions involving a single gene can be categorized into the three main ways that they can be passed down in a family: <u>autosomal recessive</u>, <u>autosomal dominant</u> and <u>X-linked</u>. These three terms are also referred to as 'patterns of inheritance', and understanding the basics behind these terms will help to understand how genetic conditions may run in a family.

Because each gene does a specific job in the body, if someone inherits a non-working gene(s), what job that gene(s) does in the body generally determines what the genetic condition will be. For example, if someone inherits a broken copy of a gene that is responsible for building the bone in your body, then that person may have a genetic disorder that causes problems with their bones, while someone who inherits a broken copy of a gene that helps to form your ears may have issues with their hearing.

Now that we've gone over the basics, click on the following links to learn more about the following areas:

- Inheritance Patterns
- Family History
- Why Genetic Counseling?
- <u>Genetic Testing</u>

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