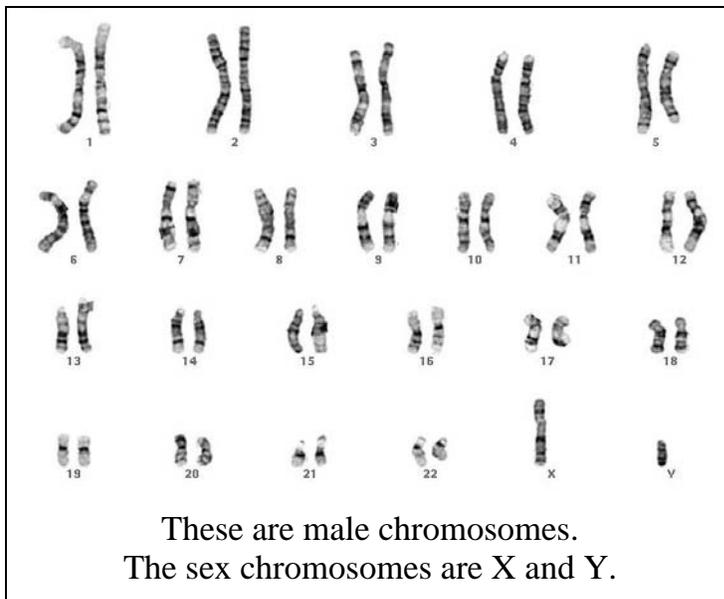


# Learning about chromosomes

## What are chromosomes?

Chromosomes are thread-like structures found in the cells of our bodies. They hold thousands of pieces of genetic material called genes. The genes contain DNA, which is the code for the “master plan” for the body. The DNA provides instructions for how the body will grow, develop and function.

There are 46 chromosomes in each cell that come in 23 matching pairs. One chromosome in each pair is inherited from the person’s mother and one from his or her father.

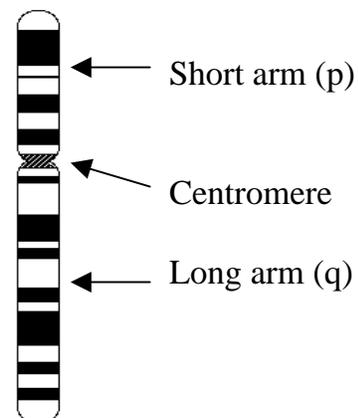


Twenty-two of the pairs are similar in men and women. They are numbered, 1 through 22, from largest to smallest.

The 23rd pair is the sex chromosomes and determines whether the person is a male or female.

Girls have two x chromosomes (XX) and boys have an x and y chromosome (XY).

Each chromosome has a band at the “waist” called the centromere. As the centromere is not exactly in the middle, it divides the chromosome into two parts or “arms” of different lengths. The short arm is called “p” and the long arm is called “q”.



A karyotype test is done to look at a person's chromosomes. The chromosomes from one cell are stained so that they can be seen under a microscope. Then a picture is taken of the chromosomes. Each chromosome is examined for its size, location of the centromere and pattern of light and dark bands. Then the picture is re-arranged to show the chromosomes in 23 matched pairs.

Most of a person's DNA is similar to others, but a small percent is different. This makes each person unique. For example, the differences in DNA give us our individual features, hair and skin colour. Sometimes, the differences result in an inherited health condition.

## **What happens if there is a problem with chromosomes?**

Sometimes a problem or error is found with a gene or chromosome. The error may explain why a person has certain characteristics or behaviours. An error may point to potential problems with health or development. The possible effects of the error depend on its exact location.

As each gene has a specific position on a chromosome, an error is given an "address". The position or address is made up of the number of the chromosome, the arm letter (p or q) and a number that relates to its position on the arm.

Examples of problems with chromosomes include:

- A small piece of genetic material may be missing from a chromosome. This is called a deletion. The deletion is identified by its address – which chromosome is involved and where the deletion starts and ends.
- A part or a whole chromosome may be duplicated. This means there are extra genetic instructions, which could result in a birth defect or health problem.

If there is a problem with your child's chromosomes, you will be able to meet with a developmental pediatrician and a genetic counselor. They will help you learn about the error or problem and discuss what this might mean for your child and your family.