

## Karyotype Activity

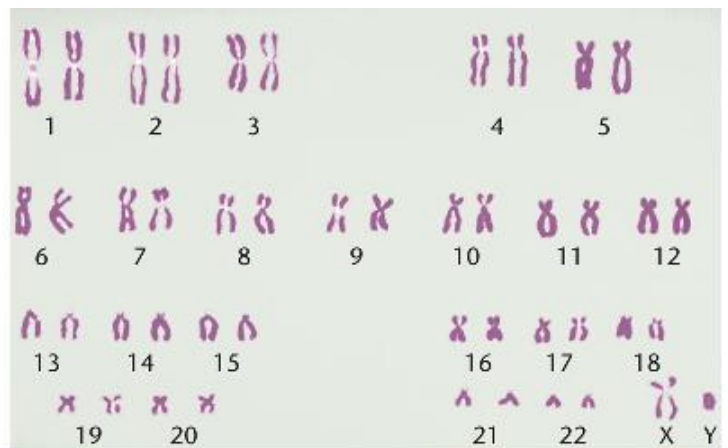
### Part 1: Normal Human Karyotype

1. Read "Characterizing Chromosomes" on pages 208 – 209 in *Insights*, then define:

a. *Karyotype*:

b. *Diploid*:

Below is a human **karyotype**. The image on the left is a picture of an actual cell's chromosomes under a microscope. To generate the image on the right, the chromosomes from the image on the left were arranged into homologous pairs and then ordered in decreasing size. Typically the sex chromosomes are placed after chromosome pair 22.



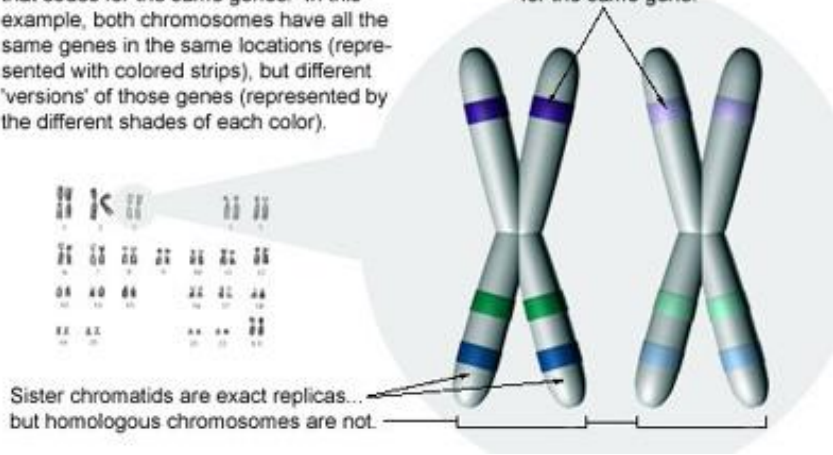
2. During which stage in the cell cycle was the first image taken? How do you know?
3. How many pairs of chromosomes are in the human genome? How many total chromosomes? Why do they come in pairs?
4. Is this karyotype from a male or female? Explain.
5. How might a karyotype be useful to a geneticist (someone who studies genetics)?

Part 2: Homologous Chromosomes

Figure B-11: Homologous Chromosomes

Homologous chromosomes contain DNA that codes for the same genes. In this example, both chromosomes have all the same genes in the same locations (represented with colored strips), but different 'versions' of those genes (represented by the different shades of each color).

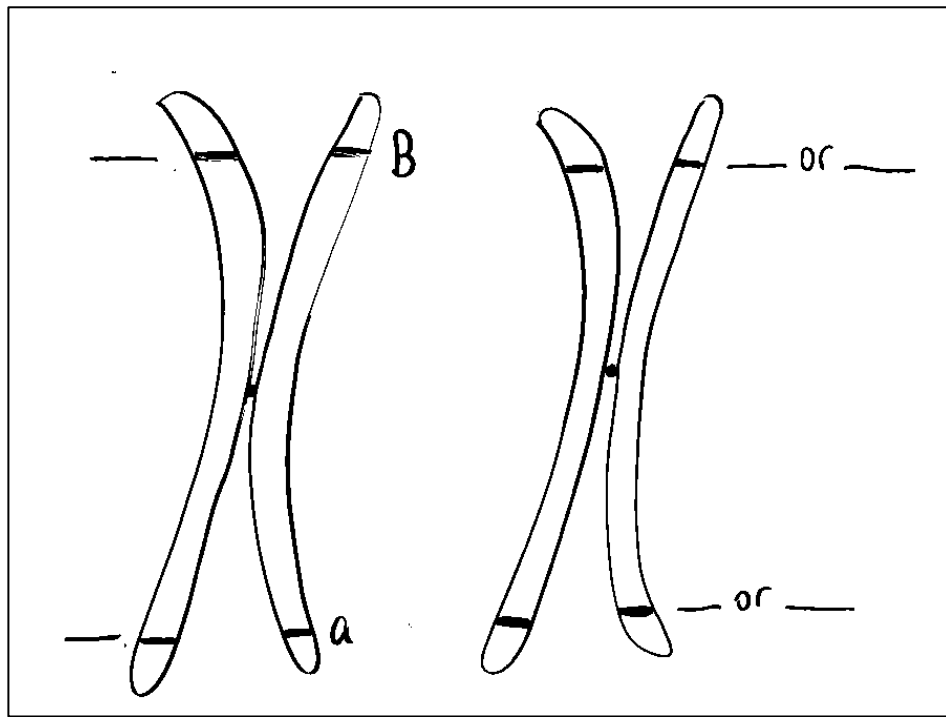
Homologous regions code for the same gene.



6. What is the same about a pair of homologous chromosomes? What can be different?

7. What do you think is meant by a "gene location"?

8. Complete the diagram below by filling in the blanks for the missing gene labels (alleles)



### Part 3: Chromosomal Disorders / Abnormal Karyotypes

**Chromosomal Disorders:** Humans have 46 chromosomes, arranged as 23 pairs, in each cell. Each chromosome of a pair is inherited from each of the parent.

1. An error may occur in the cell division either before or after the dad's sperm fertilized the mom's egg. The embryo might have an decreased or increased number of chromosomes. If an individual has three copies of a chromosome when there should only be two, it is called a **trisomy**. This means that one parent gave them an extra copy of a chromosome. When only one copy of a chromosome is present where there should be two, it is known as **monosomy**. This means that the child only received a copy of the chromosome from one of their parents instead of two.
2. Some genetic conditions also occur as a result of the **sex chromosomes** (the 23<sup>rd</sup> homologous chromosome pair) being more or less in number. A healthy male has one X and one Y chromosome (XY) while a healthy female has two X's (XX). Sometimes a female has only a single X chromosome (X) or a male can have an extra X chromosome (XXY).
3. In certain cases the number of chromosomes may be intact, but a part of a chromosome is missing. This is called a **deletion**, which will result in the loss of a single gene or a few genes. **Translocations** occur when parts of the chromosomes get transferred from one chromosome to the other. You can see this on a karyotype when one chromosome is too short and another is too long. Translocations do not cause a person to develop incorrectly, but they may cause later life complications like infertility or miscarriages.

A common way to diagnose an individual with a chromosomal disorder is to prepare a karyotype for them. Each pair of you will now get a karyotype of a fetus developing in a mother's womb. The fetus is suspected to have a chromosomal disorder. Compare this karyotype to the normal karyotype on page one in order to identify if the fetus has a chromosomal disorder, and if they do what might be causing it. I will give you a sheet titled "Information on Chromosome Disorders" that will help you learn about the fetus' disorder.

#### **Your Task:**

1. Ask your instructor for an abnormal karyotype as well as Information on Chromosome Disorders.
2. Read through all of the disorders then compare your karyotype to the descriptions to determine which chromosomal abnormality is depicted in your karyotype
3. Prepare a short presentation (about 2 min) to be given to the class along with a visual that addresses the following:
  - ✓ The chromosomal abnormality and the name of the syndrome
  - ✓ The cause of the chromosomal disorder
  - ✓ Symptoms as well as the frequency of the disorder and life expectancy if relevant