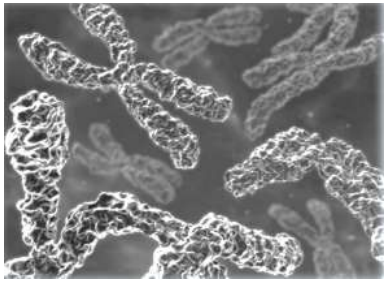
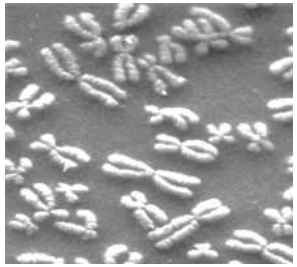
Karyotypes Lab

Directions: Read and highlight the background information.



Background

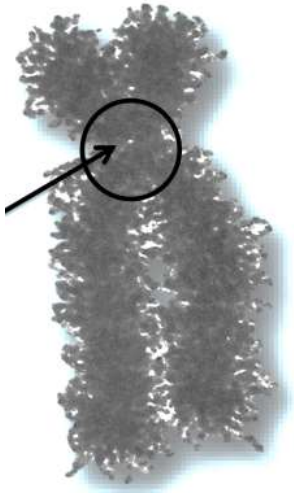
We can learn a lot by looking at chromosomes! They can tell us everything from the likelihood that an unborn baby will have a genetic disorder to whether a person will be male or female. Scientists often analyze chromosomes in prenatal testing and in diagnosing specific diseases.



To detect chromosomal disorders such as Down Syndrome, a scientist examines the chromosomes from a person's cell using a method called a **karyotype**. Simply, a karyotype is a picture of all the chromosomes in the cell. Karyotypes organize the genome (all the DNA in a person's cells) according to their number, size, shape, and other chromosome structures.

In this lab, you will take the role of a geneticist and cut out chromosomes from a picture taken of a patient's genome. Then, you will match up the chromosomes using size and banding patterns as guides. Once the chromosomes are matched, you will be able to detect both the sex of the individual and if that person has too many or not enough chromosomes. Remember that humans have a total of 46 chromosomes.

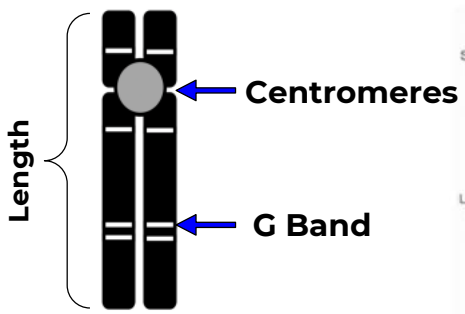
A **chromosome** is a single piece of DNA which contains many genes. Chromosomes are organized into the shape (shown to the right) at the time cells begin to divide. When we are organizing the chromosomes, one way to lineup and match the pair is to use the centromere found here:

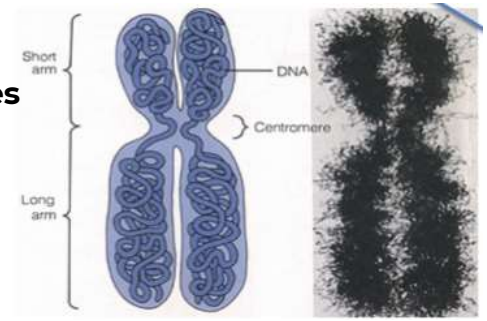


G BANDING

During the reproduction process of mitosis, the 23 different pairs of human chromosomes condense and are visible using a light microscope. To visualize the chromosomes to build a karyotype, technicians must block the cell division. When they do this, they also stain the condensed chromosomes with Giemsa dye. This dye stains particular regions of chromosomes that are dense in base pairs Adenine (A) and Thymine (T), producing a dark band.

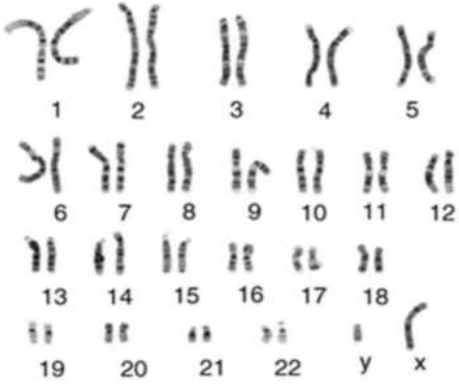
There is a common misconception about these bands. Most people think that these bands represent one particular gene. That is not the case. In fact the thinnest bands can





contain over a million base pairs and code for potentially hundreds of genes. Make sure you use this G Banding to help you organize similar chromosomes when developing your karyotype. Therefore, in summary, when putting together your karyotype make sure you compare each chromosome for the following;

1. Length
2. Placement of their centromeres
3. Location and sizes of the G-bands



This is an actual finished product of Karyotype. As you can see all sister chromatids are lined up according to their size and bands. Also notice the difference between the "y" and "x" chromatids. This karyotype represents a patient that is a male. Since this patient had all 23 chromosomes without any abnormalities, he does not have any genetic disorders.

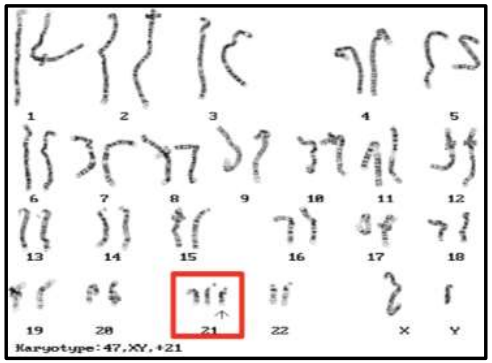
Example Human Chromosomal Disorders

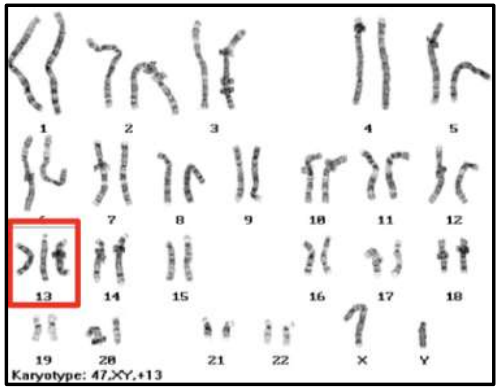
The karyotype to the left is an example of a "normal" karyotype. All 22 autosomal chromosomes (chromosomes #1 through 22) are paired up. There are none that have only 1 copy or have more than 2 copies. There are also 1 pair of sex chromosomtes (the X and Y chromosomes). Again, not just one and not more than 2 sex chromosomes.

This is not true for all people. About 0.2% of the population has a chromosomal abnormality. That equates to about 659,000 people in the United States. In humans, there are many chromosomal abnormalities that lead to a spontaneous abortion (commonly called a "miscairrage") but many more lead to a viable pregancy and offspring. Below are a few of some more common chromosomal abnormalities and their karyotypes.

DOWN SYNDROME (trisomy 21)
The result of an extra copy of chromosome 21. People with Down syndrome are 47, 21+. Down syndrome affects 1 in 700 children and alters the child's phenotype either moderately or severely:

- Characteristic facial features
- Short stature
- Heart defects
- Susceptibility to respiratory disease
- Shorter lifespan
- Often sexually underdeveloped and sterile
- Usually some degree of intellectual disability

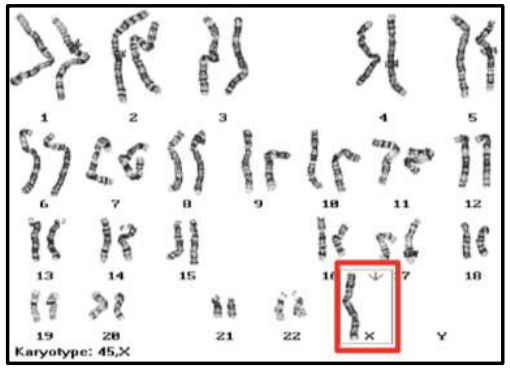




PATAU SYNDROME (trisomy 13)
Only five percent to 10 percent of children with this condition live past their first year. Individuals with trisomy 13 often have:

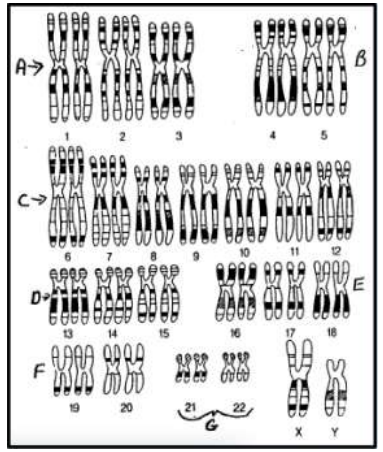
- Severe intellectual disability
- Physical abnormalities in many parts of the body. Heart defects, brain or spinal cord abnormalities
- Very small or poorly developed eyes
- Extra fingers and/or toes
- An opening in the lip (a cleft lip) with or without an opening in the roof of the mouth (a cleft palate)
- Weak muscle tone

Turner's Syndrome (Monosomy X)
Monosomy is the state of having a single copy of a chromosome pair instead of the usual two copies found in diploid cells. This can be partial if a portion of the second chromosome copy is present. Monosomy occurs in 1 in 5000 live births (98% of monosomy fetuses die before birth). The only viable monosomy in humans is Turner's when females have only 45 chromosomes! XO individuals are genetically female, however, they do not mature sexually during puberty and are sterile.



Pre-Lab Questions

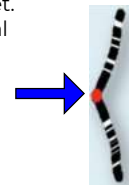
Directions: Use the completed karyotype below to answer the following questions.



1. What is the total number of chromosomes found in this patient's cell? _____
2. What are three different chromosome characteristics used to organize the karyotype?
   a. _____
   b. _____
   c. _____
3. Which pair is not given a number? Why?
   _____
   _____
4. What is the sex of this patient? _____
5. Are there any genetic disorders found in this patient? Explain. _____
   _____
   _____

Lab Procedure

1. On the last page of this packet you will see a pool of human chromosomes. Lab technicians have done the tough work already staining the chromosomes so a picture can be readily available to you. Detach this page from the packet.
2. Cut out each chromosome and use the "Example Human Chromosomal Abnormalities" to guide you.
3. Use the centromere (gray) in the middle of each chromosome to help line up matching chromosomes.



4. On your "Karyotype Panel" Line up each chromosome according to size and G bands. The larger chromosomes come first. Place your cut out chromosomes in order from large to small. Use the normal human chromosome karyotype as a reference. The 23rd pair (sex chromosomes) comes last. Paste those on the top. That will tell you what sex your individual is.
5. Once you have lined up all your chromosomes, glue or tape them down onto the sheet.
6. Once all your chromosomes are pasted on the sheet, determine your sex. If you have an X and Y chromosome you have a chromosomal male and XX, you have a chromosomal female.
7. Finally, count each chromosome and make sure you have a total of 46 chromosomes. If you have either "more" or "less", use the Example Human Chromosomal Disorders to determine what genetic disorder exists in your patient.