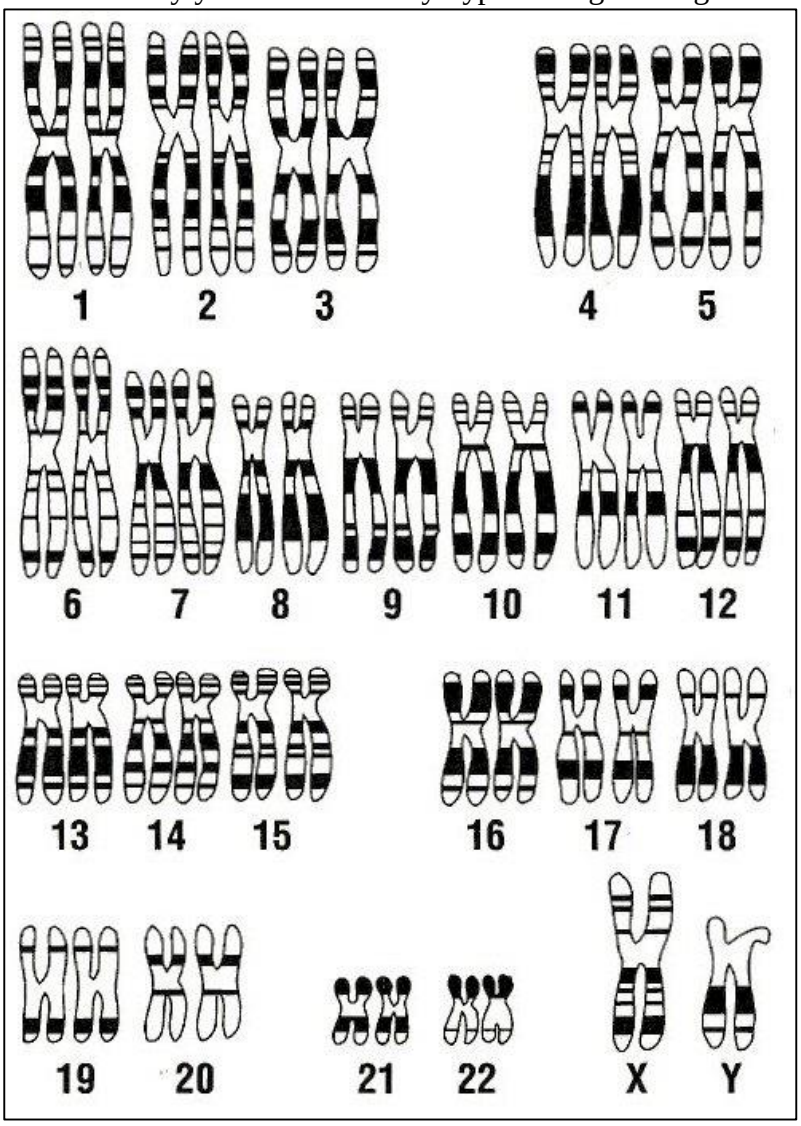
Human Karyotypes

Occasionally chromosomal material is lost or rearranged during the formation of gametes or during cell division of the early embryo. These changes, which usually result from nondisjunction or translocation, are usually so severe that the pregnancy ends in miscarriage. Although the frequency is nearly impossible to measure, some researchers estimate 1 in 156 live births have some kind of chromosomal abnormality. Amniocentesis and CVS can be used to collect fetal cells in order to study the chromosomes to determine if there is any kind of chromosomal abnormality.

Some of the abnormalities associated with chromosome structure and number can be detected by a procedure called a **karyotype**. To prepare a karyotype, fetal cells are harvested and cultured *in vitro*. Chromosomes are stained and a digital photograph is made. Using photo editing software, the images of the chromosomes are matched in homologous pairs and arranged in order by size, with the sex chromosomes last. The chromosomes must come from a mitotic cell so that the chromosomes are replicated, condensed, and visible under a microscope. Abnormalities in chromosome number, shape, or size can be detected this way. The technique can also be used to show prospective parents whether they have such abnormalities that could be passed on to their offspring.

In this activity you will use a karyotype to diagnose a genetic disorder.



Procedure

1. Obtain a set of chromosomes and record the number.
2. Carefully cut out one chromosome and then its homolog. If you don't have tape to fasten them to a piece of paper, set them aside so as not to lose them. Continue cutting out and pairing chromosomes to prepare the karyotype.

Questions

1. What criteria are used to arrange the chromosomes in pairs?
2. What is the sex of the individual in your karyotype?
3. On the basis of the information you can gather from your karyotype, provide a diagnosis. Give the number of the karyotype you used.
4. What stage during the cell cycle is best for producing a karyotype?
5. Would the diagnosis of Turner's syndrome in a single cell necessarily mean that every cell of the body would contain 45 chromosomes? Explain.
6. Explain how nondisjunction could result in Down syndrome.
7. People with Down syndrome are not always sterile. Imagine two people with

Down syndrome have a child. Is it possible for that child to be normal? Explain.
8. Would it ever be possible to produce a baby who has 48 chromosomes? Explain.
9. More males than females suffer from color blindness. Speculate as to why females who have Turner's syndrome have a similar incidence of color blindness as males.
10. Would a genetic counsellor be able to use a karyotype to identify a carrier of cystic fibrosis?