

Karyotypes

Pre-Lab Discussion

Several human genetic disorders are caused by additional, missing, or damaged chromosomes. One way of studying genetic disorders is to observe the chromosomes themselves. In order to do this, cells from a person are grown in a laboratory. After the cells have reproduced a few times, they are treated with a chemical that stops cell division at the metaphase stage. During metaphase, the chromosomes are at the best length for identification.

The cells are treated further, stained, and then placed on glass slides. The chromosomes are observed under the microscope, where they are counted, checked for abnormalities, and photographed. The photograph is then enlarged, and the chromosomes are individually cut out. The chromosomes are identified and arranged in *homologous* pairs. Homologous chromosomes are identical, or matching, chromosomes. The arrangement of homologous pairs is called a *karyotype*.

In this investigation, you will use a sketch of chromosomes to make a karyotype. You will also examine the karyotype to determine the presence of any genetic defects.

Problem

How can chromosomes be observed?

Materials (per student)

Scissors
Glue or transparent tape

Safety

Be careful when handling sharp instruments. Note all safety alert symbols next to the steps in the Procedure and review the meanings of each symbol by referring to the symbol guide on page 10.

Procedure

Part A. Analyzing a Karyotype

1. Observe the karyotype in Figure 1. Notice that the two sex chromosomes, pair number 23, do not look alike. They are different because this karyotype is of a male, and a male has an X and a Y chromosome.

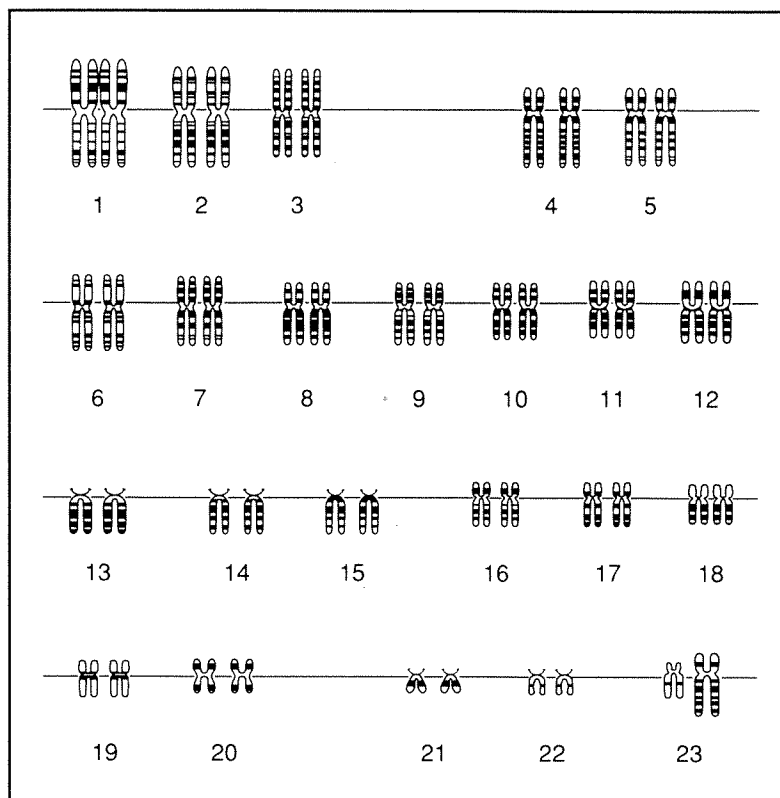


Figure 1

2. Identify the centromere in each pair of chromosomes. The centromere is the area where each chromosome narrows.

Part B. Using a Karyotype to Identify a Genetic Disorder

1. Study the human karyotype in Figure 2. Notice that 23 chromosomes are numbered 1 through 23.
2. To match the homologous chromosomes, look carefully at the unnumbered chromosomes. Note their overall size, the position of the centromere, and the pattern of the light and dark bands. Next to the unnumbered chromosome that is most similar to chromosome 1, write 1.
3. Repeat step 2 for chromosomes 2 through 23. **Note:** Many genetic disorders involve missing or extra chromosomes.

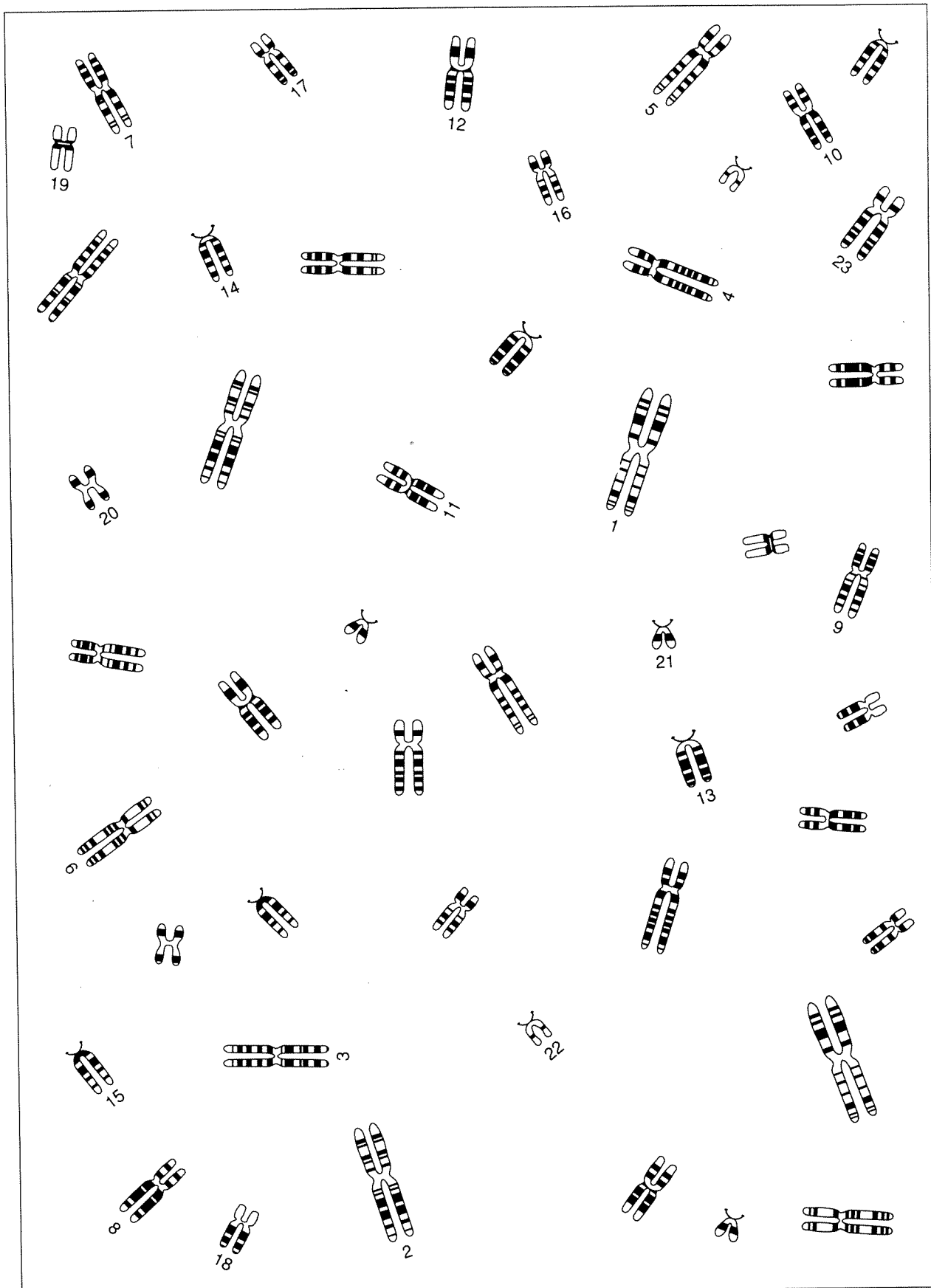
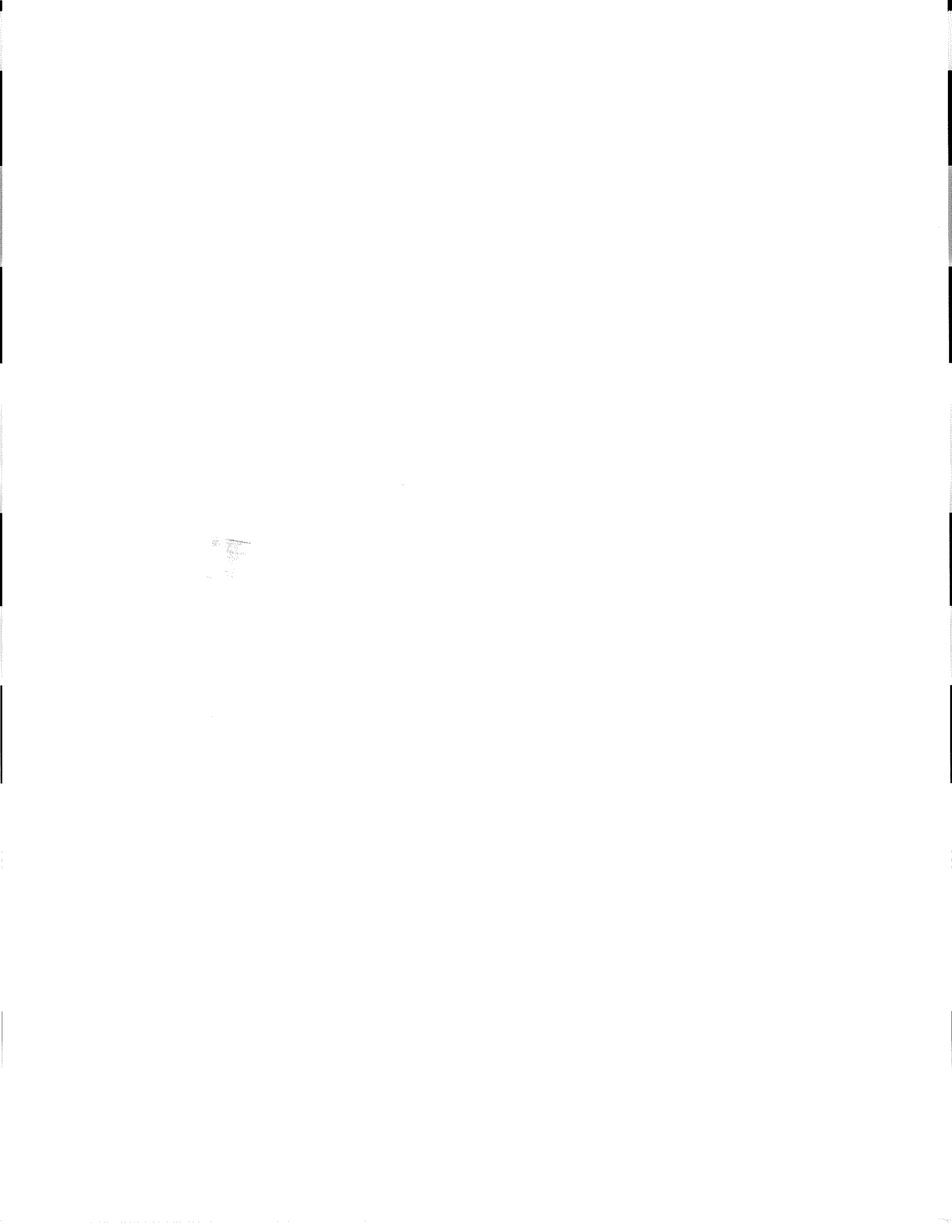


Figure 2



4. After all the chromosomes have been identified, use scissors to cut them out. Arrange the chromosomes in their appropriate place in Figure 3. Note the presence of any genetic defects.

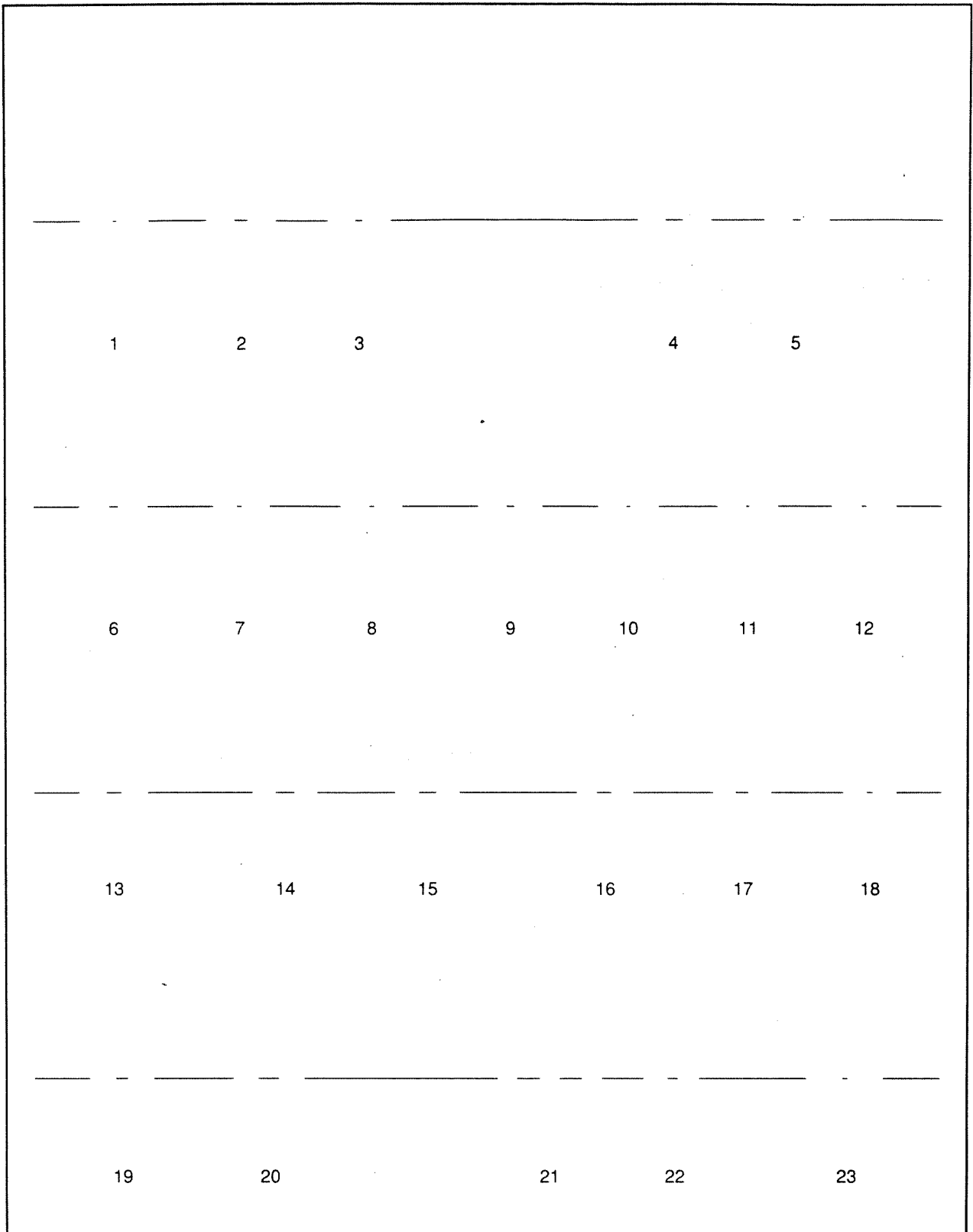
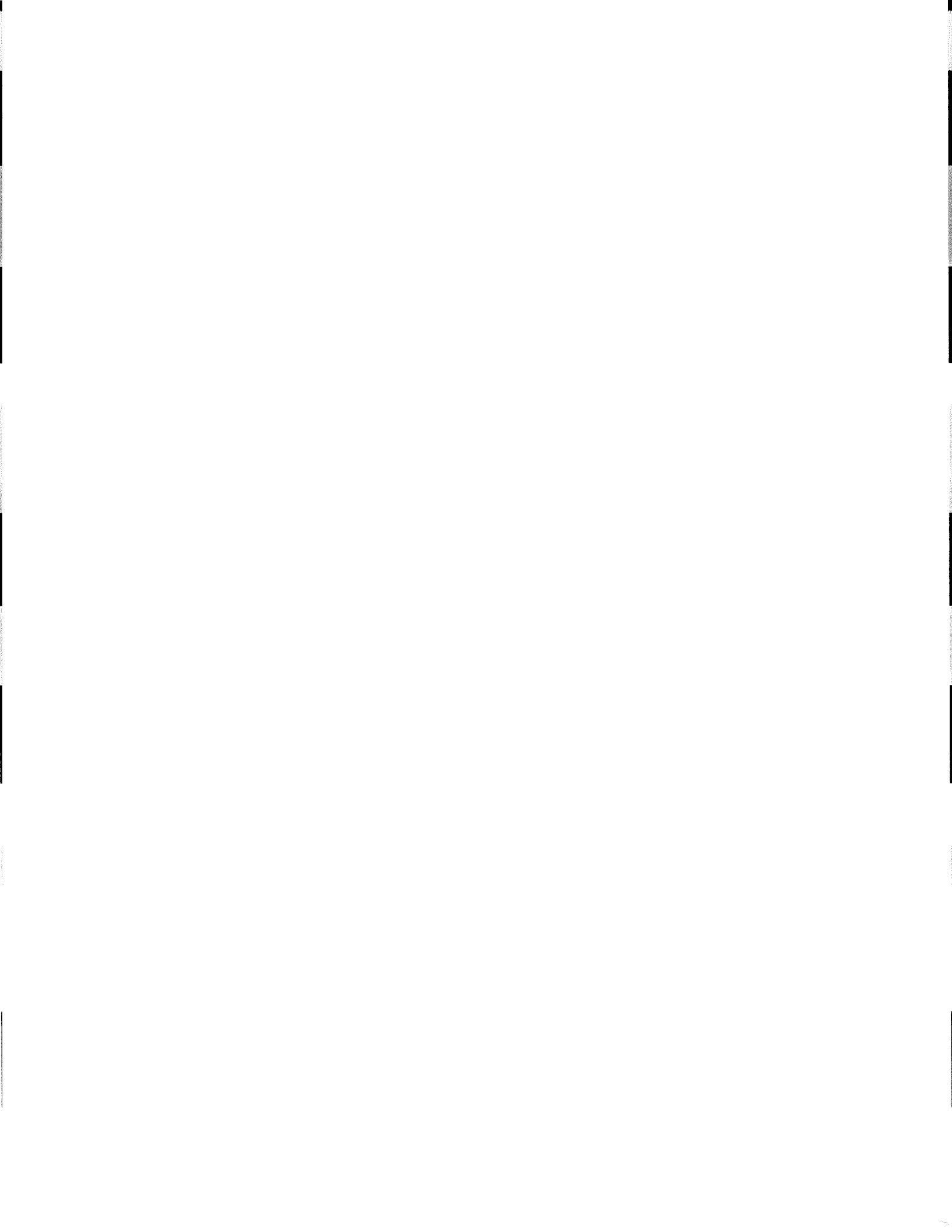


Figure 3



5. Observe the karyotypes in Figures 4 and 5. Note the presence of any genetic defects.

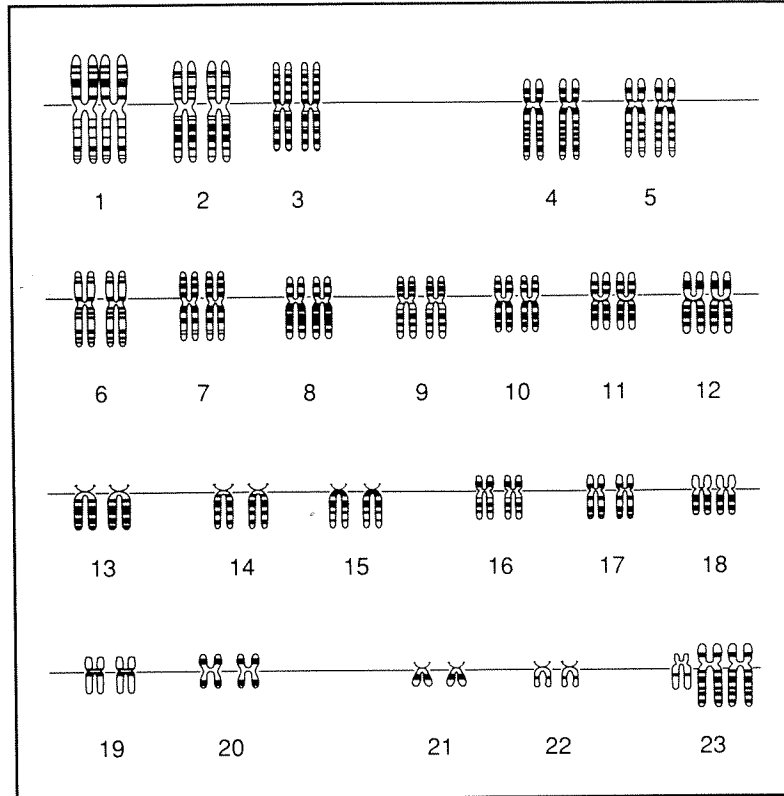


Figure 4

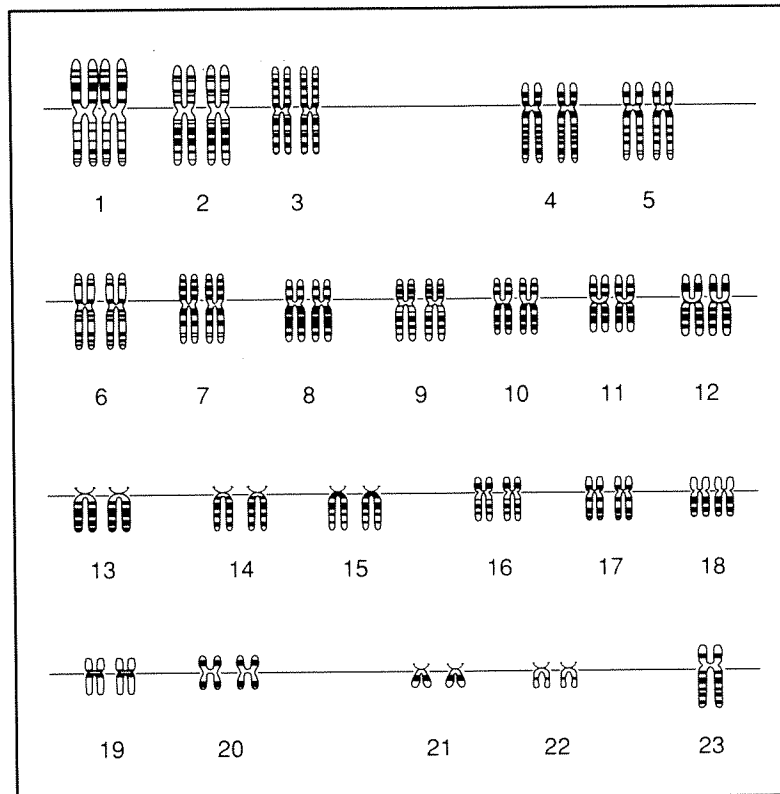


Figure 5

Observations

1. How many autosomes are present in your karyotype? _____
2. How many sex chromosomes are present in your karyotype? _____
3. Are there any abnormalities? If so, where? _____

Analysis and Conclusions

1. Is your karyotype that of a normal person or a person with a genetic disorder? If it is the latter, identify the disorder. _____

2. Is your karyotype that of a male or a female? Explain. _____

3. How does the karyotype in Figure 4 differ from the karyotype in Figure 1?

4. How does the karyotype in Figure 5 differ from the karyotype in Figure 1?

5. Do the karyotypes in Figures 4 and 5 exhibit any genetic disorder? If so, identify the disorder.

Critical Thinking and Application

1. What happens during meiosis that ultimately results in a defect characterized by the addition of chromosomes? By the deletion of chromosomes? _____

2. The human male determines the sex of his offspring. Explain this statement.

Going Further

Using reference material, research the following human genetic disorders: Patau syndrome and Edwards syndrome. For each disorder find the cause, type of mutation, and characteristics. Construct a karyotype of each.

