Name KEY Class _____ Date _____

Making Karyotypes

Introduction

Several human genetic disorders are caused by extra, missing, or damaged chromosomes. In order to study these disorders, cells from a person are grown with a chemical that stops cell division at the metaphase stage. During metaphase, a chromosome exists as two chromatids attached at the centromere.

The cells are stained to reveal banding patterns and 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 images of the chromosomes are individually cut out. The chromosomes are identified and arranged in homologous pairs. 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 chromosomal abnormalities.

Problem

How can chromosomes be observed?

Pre-Lab Discussion

Read the entire investigation. Then work with a partner to answer the following questions.

1. What clues to the presence of certain genetic disorders can be seen in a karyotype?
   If there are, more or less, than 2 pairs of chromosome
2. Why might a laboratory worker attempting to diagnose a genetic disorder prefer to work with photographs of chromosomes rather than the chromosomes themselves?
   Its easier to visualize
3. Why would it be much more difficult to construct a karyotype of unstained chromosomes?
   Harder to see the individual genes in bands
4. Which pair of chromosomes can contain two very different chromosomes and still be considered normal? Explain your answer.
   The 23rd pair = sex chromosomes
5. How do autosomes differ from sex chromosomes?
   They only carry gens for the body, not gender

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 meaning of each symbol by referring to Safety Symbols on page 8.

Procedures

Part A. Analyzing a Karyotype

1. Observe the normal human 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.

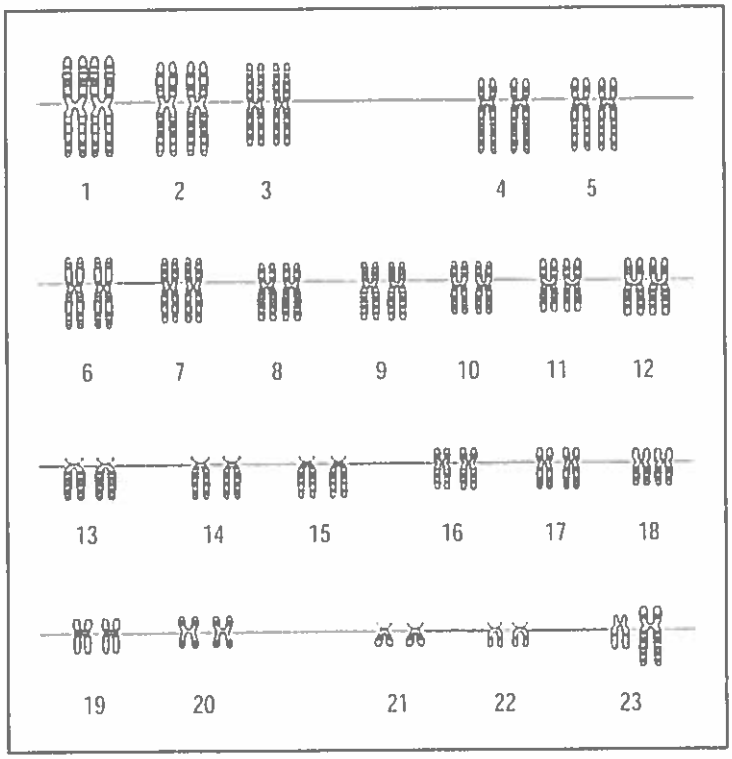

Figure 1

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 chromosomes in Figure 2 on page 125. 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.

5. Observe the karyotypes in Figures 4 and 5. Note the presence of any chromosomal abnormalities.

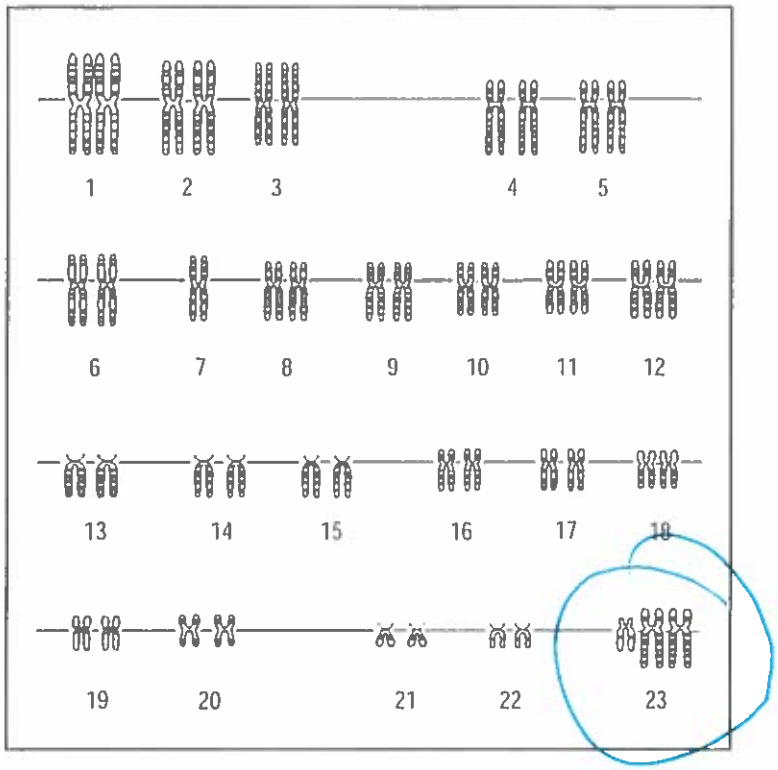

Figure 4

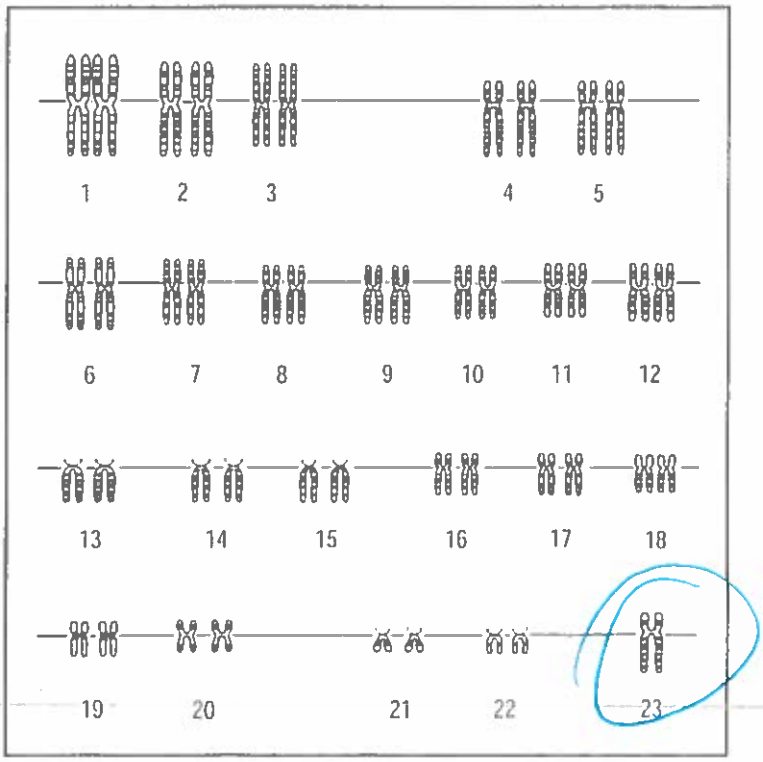

Figure 5

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6. Draw a data table in the space below in which to record your observations of the karyotypes shown in Figures 1, 3, 4, and 5. Record any evidence of chromosomal abnormalities present in each karyotype. Record the genetic defect, if you know it, associated with each type of chromosomal abnormality present.

Figure	Anything Abnormal?
1	Normal
3	Abnormal, 3 chromosomes on #21
4	Abnormal; 3 sex chromosomes
5	Abnormal; only one sex chromosome

Analysis and Conclusions

1. **Comparing and Contrasting** Of the four karyotypes that you observed, which was normal? Which showed evidence of an extra chromosome? An absent chromosome?

1= Normal, 3+4 = extra chromosomes
5= one absent chromosome

2. **Formulating Hypotheses** What chromosomal abnormality appears in the karyotype in Figure 4? Can you tell from which parent this abnormality originated? Explain your answer.

Has 3 sex chromosomes, one Y + 2 X's
originated from mother!

3. **Inferring** Are chromosomal abnormalities such as the ones shown confined only to certain parts of the body? Explain your answer.

NO - could be anywhere.

4. **Drawing Conclusions** Are genetic defects associated with abnormalities of autosomes or of sex chromosomes? Explain your answer.

Both - depends on the disorder.

5. **Posing Questions** Formulate a question that could be answered by observing chromosomes of different species of animals.

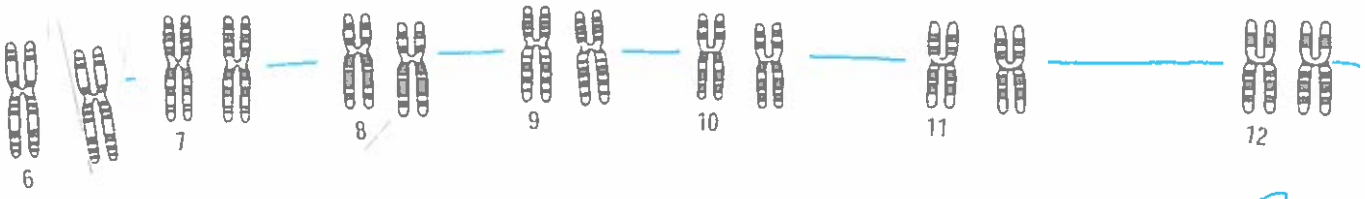
★ will be a diff. answer...

Going Further

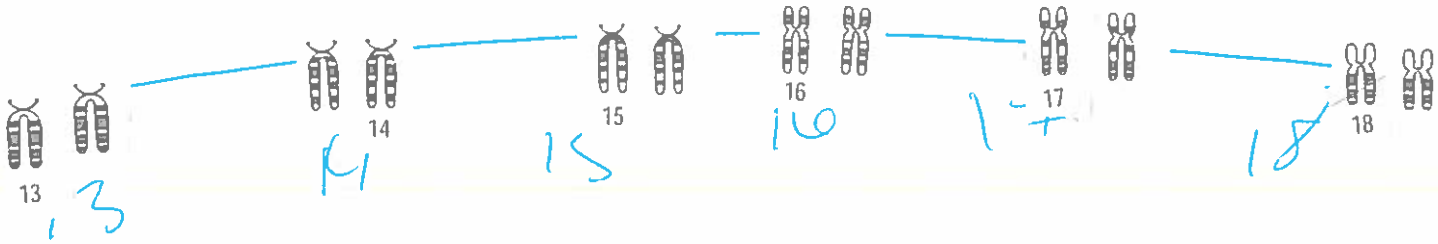
Using library materials or the Internet, research one type of deletion syndrome (a syndrome that results from loss of parts of chromosomes). Write a short paragraph describing the chromosomal abnormality involved and the characteristics of the disorder.

Figure 3

1 2 3 4 5



6 7 8 9 10 11 12



13 14 15 16 17 18

19 20 21 22 23