Name _____

Honors Biology Chromosome Karyotype Lab

A Chromosome Study

In this activity, you will create a karyotype from a page of mixed chromosomes. Karyotypes are created by matching homologous pairs and numbering them from largest to smallest. Abnormalities, such as extra or deleted chromosomes can then be diagnosed. Pictured chromosomes will be used for this model rather than real chromosomes, but the process is the same for real chromosomes extracted from cell or fetal samples.

Two karyotypes will be created, the first represents a normal human karyotype of a male or a female, the second represents and abnormal karyotype. You will then compare and diagnose the abnormality present in the patient of the second karyotype.

Normal Human Karyotype

Examine the page marked "normal". These chromosomes are actually enlarged photographs of what is seen through a microscope. Note that the sex chromosomes have been labeled for you as either X or Y chromosomes. They have been marked this way to indicate these are the sex chromosomes. Cut out each chromosome with scissors, to make it go faster, cut them out as squares rather than trying to cut around the margin of each chromosome.

Prepare a karyotype of these chromosomes. A karyotype is a pattern or picture of chromosomes from one cell **grouped into pairs** and **organized by size**.

--Pair up each of the chromosomes with its homologous pair, use the size and markings on the chromosomes to determine pairs. Temporarily put the two unshaded chromosomes aside.

--On a blank paper, arrange the chromosome pairs from largest to smallest and number them. You numbers should range from 1(largest) to 22(smallest). Put the sex chromosomes last, this is pair #23. Glue or tape the chromosomes to the paper in the correct order.

Sex chromosomes determine the sex of the individual. A female develops when the sex chromosomes match--XX . A male develops if the two sex chromosomes are unmatched--XY. (These chromosomes are unshaded on your karyotype)

1. How many total chromosomes are present in this karyotype? _____

2. How many chromosomes are present in each cell of this human? _____

3. Does your karyotype represent a male of a female? _____

4. Chromosomes that are NOT sex chromosomes are called autosomes. How many total autosomes are present in your normal karyotype? _____

Body cells are called somatic cells. Somatic cells include the skin, liver, muscle, stomach and other bodily cells. The karyotype you prepared is from a somatic human cell. The term **diploid chromosome number** refers to the number of chromosomes in a somatic cell.

The **diploid number** varies from species to species, however it does not differ from somatic cell to somatic cell within the same organism. To find your diploid number, simply count the number of chromosomes in your karyotype.

The diploid chromosome number is also called the **2n number**.

4. What is the diploid chromosome number for your karyotype? _____

5. What is the 2n chromosome number for your karyotype? ____

The HAPLOID CHROMOSOME NUMBER refers to the number of chromosomes in an organisms sex cells. Sperm in males, eggs in females. The **haploid number**, or **1n number** is always -half- that of the diploid number.

6. What would the haploid chromosome number for your karyotype be? ____

Abnormal Human Karyotype

--Examine the page marked "Abnormal". Look at the top corner, what Set do you have?____

--Prepare a second karyotype as you did the first. In this karyotype, you will discover an abnormality in the chromosome number. Finding incorrect chromosomes numbers in human somatic cells of an unborn baby alerts doctors to the fact that their child is abnormal and will be born with birth defects.

*If the unborn has an extra number 13 chromosome, it it born with Patau syndrome. An extra chromosome 18 results in Edward syndrome. An extra chromosome number 21 results in Down Syndrome. A missing sex chromosome results in an X0 offspring who has Turner's syndrome. An extra X chromosome results in Klinefelters syndrome (XXY).

7. How many chromosomes are present in the abnormal karyotype: _____

8. What is the diploid chromosome number for this karyotype: _____

9. Which chromosome pair is abnormal? _____

10. What syndrome does this unborn have? _____

11. What sex will the unborn child be? _____

Analysis:

12. Define the following terms:
    Somatic Cell

    Karyotype

    Diploid chromosome number

    Autosome

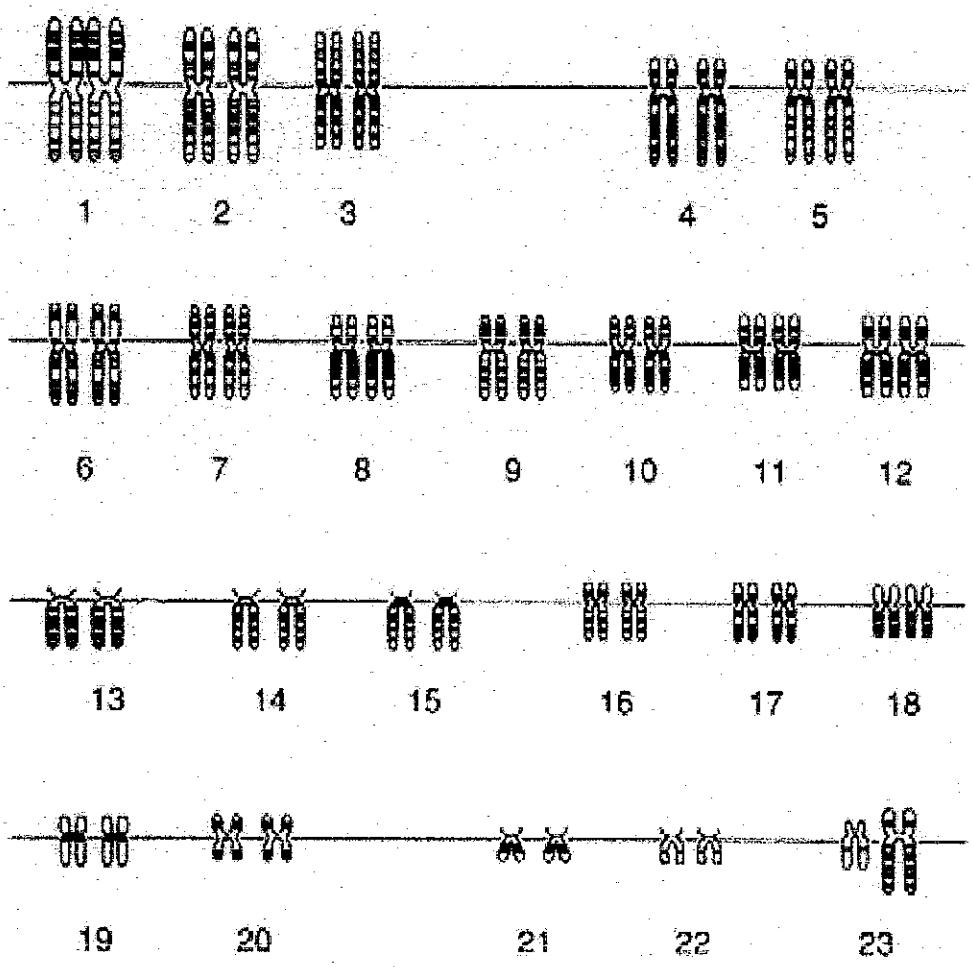
14. Describe two types of information that can be gained about a child before it is born through a karyotype.

TABLE 1 GENETIC DISORDERS CAUSED BY AN ABNORMAL CHROMOSOME NUMBER

Name of abnormality	Chromosome affected	Description of abnormality
Down syndrome, or Trisomy 21	#21	47 chromosomes; mental retardation with specific characteristic features; may have heart defects and respiratory problems
Edwards' syndrome, or Trisomy 18	#18	47 chromosomes; severe mental retardation; very characteristic malformations of the skull, pelvis, and feet, among others; die in early infancy
Patau syndrome, or Trisomy 13	#13	47 chromosomes; abnormal brain function that is very severe; many facial malformations; usually die in early infancy
Turner's syndrome	Single X in female (XO)	45 chromosomes; in females only; missing an X chromosome; do not develop secondary sex characteristics; are infertile
Klinefelter's syndrome	Extra X in male (XXY)	45 chromosomes; in males only; sterile, small testicles; otherwise normal appearance
XYY syndrome	Extra Y in male (XYY)	47 chromosomes; in males only; low mental ability; otherwise normal appearance
Triple X syndrome	Extra X in female (XXX)	47 chromosomes; sterility sometimes occurs; normal mental ability

4. Arrange the pairs according to their length. Begin with the largest chromosomes and move to the smallest.
5. Tape each pair of homologous chromosomes to a human karyotyping form. Place the centromeres on the lines provided. Place the longest chromosome at position 1, and the shortest at position 22. Place the two sex chromosomes at position 23.
6. The diagram you have made is a karyotype, as in **Figure 1.** Analyze your karyotype to determine the sex of the individual. Use the information in **Table 1** to guide your analysis.

Example of karyotype



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.

Normal Karyotype 1 or 2 (circle)

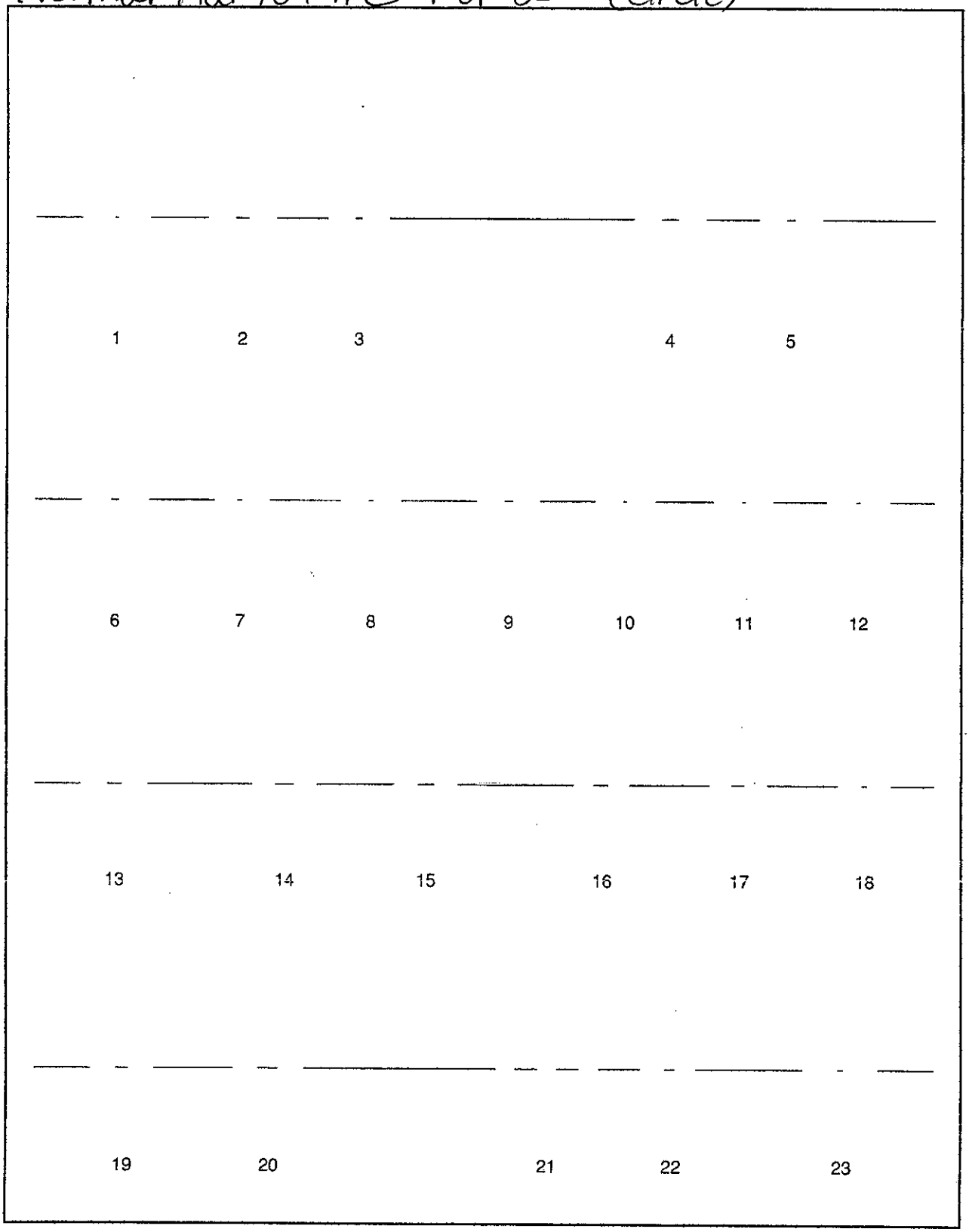


Figure 3

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.

Set ___ (identify letter)

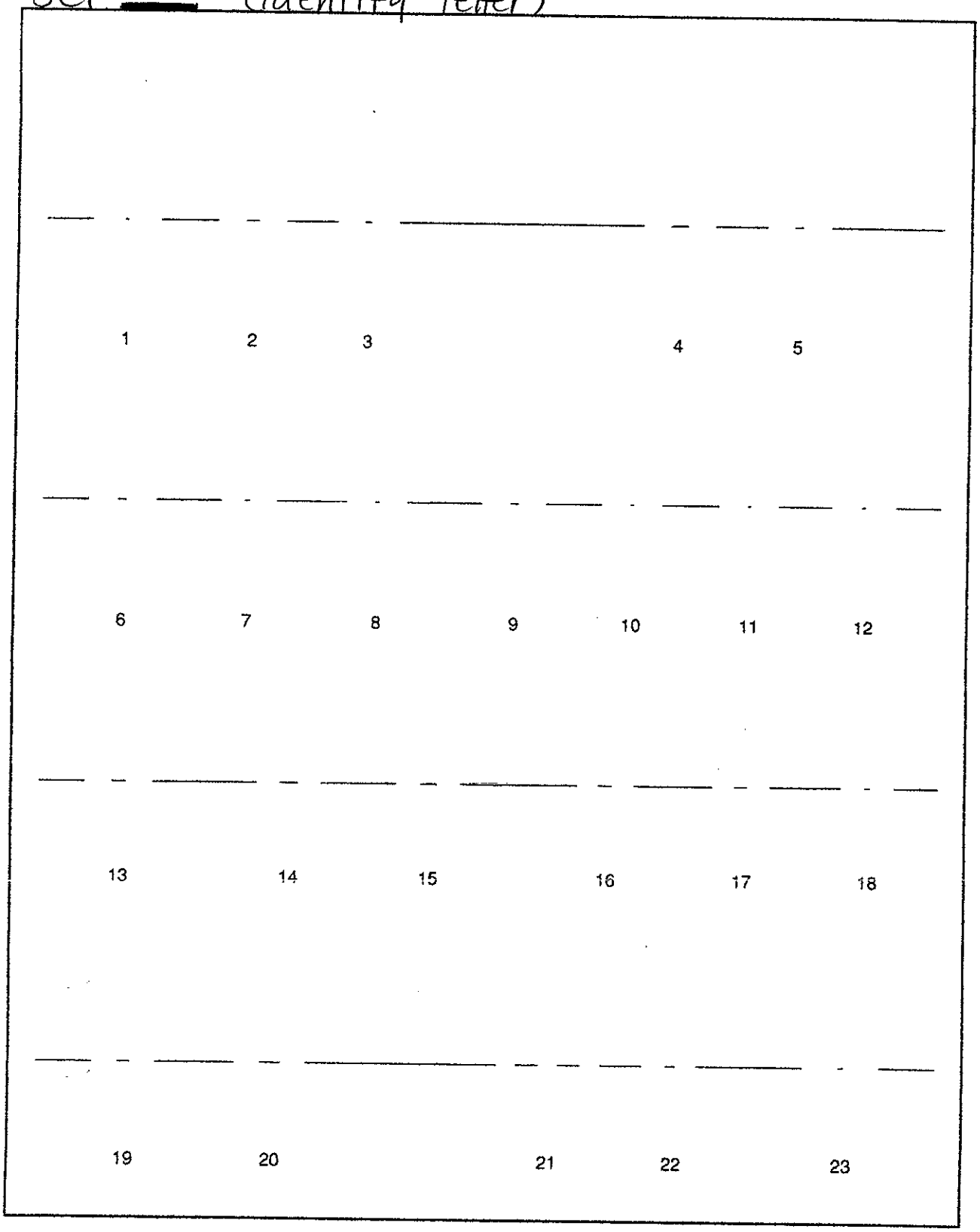


Figure 3

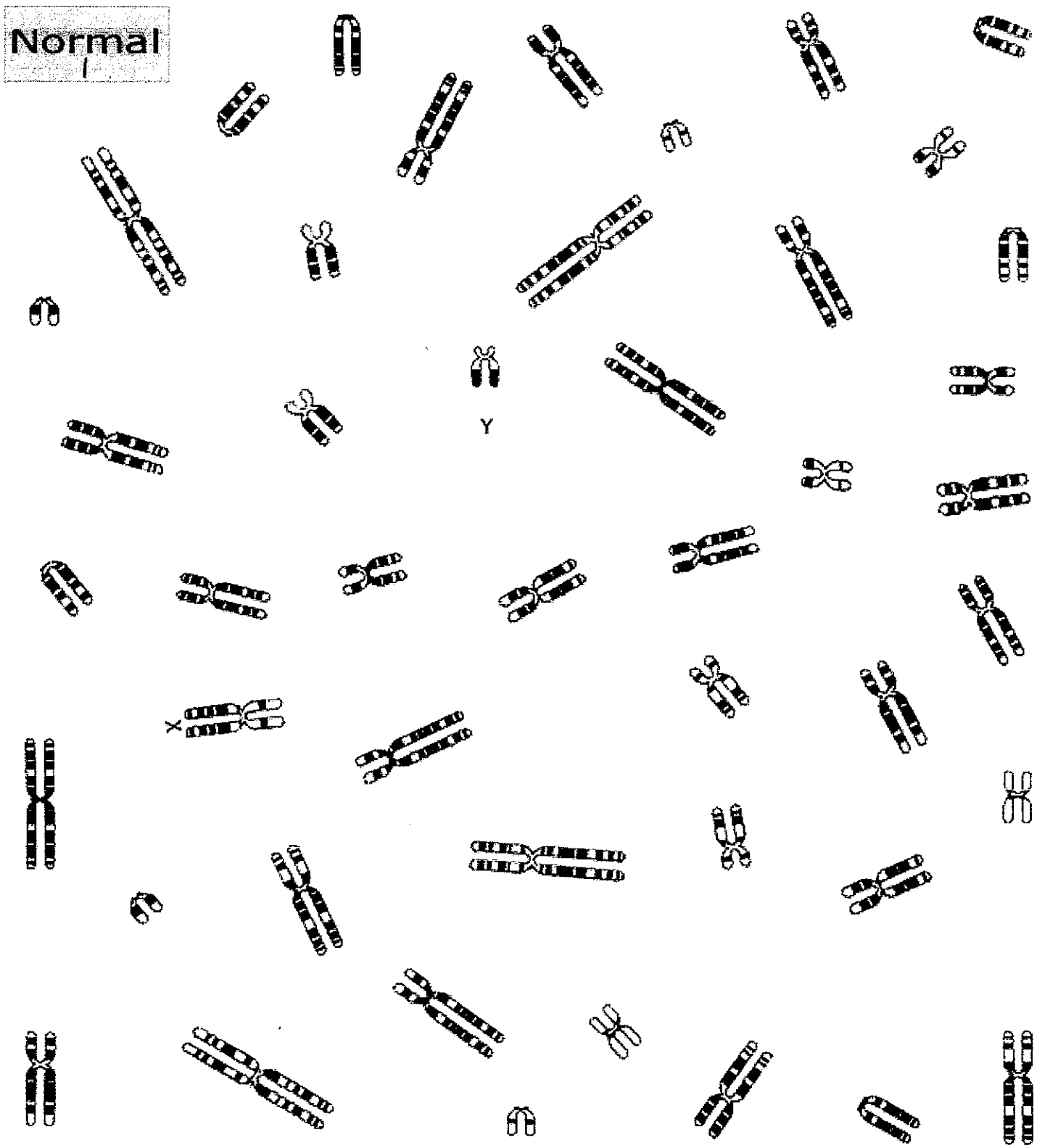
3. **Identifying Relationships** Assume that two students started with the same photomicrograph. One student concluded that the individual had Down syndrome. The other student concluded that the individual had Edwards' syndrome. Explain how this could happen.

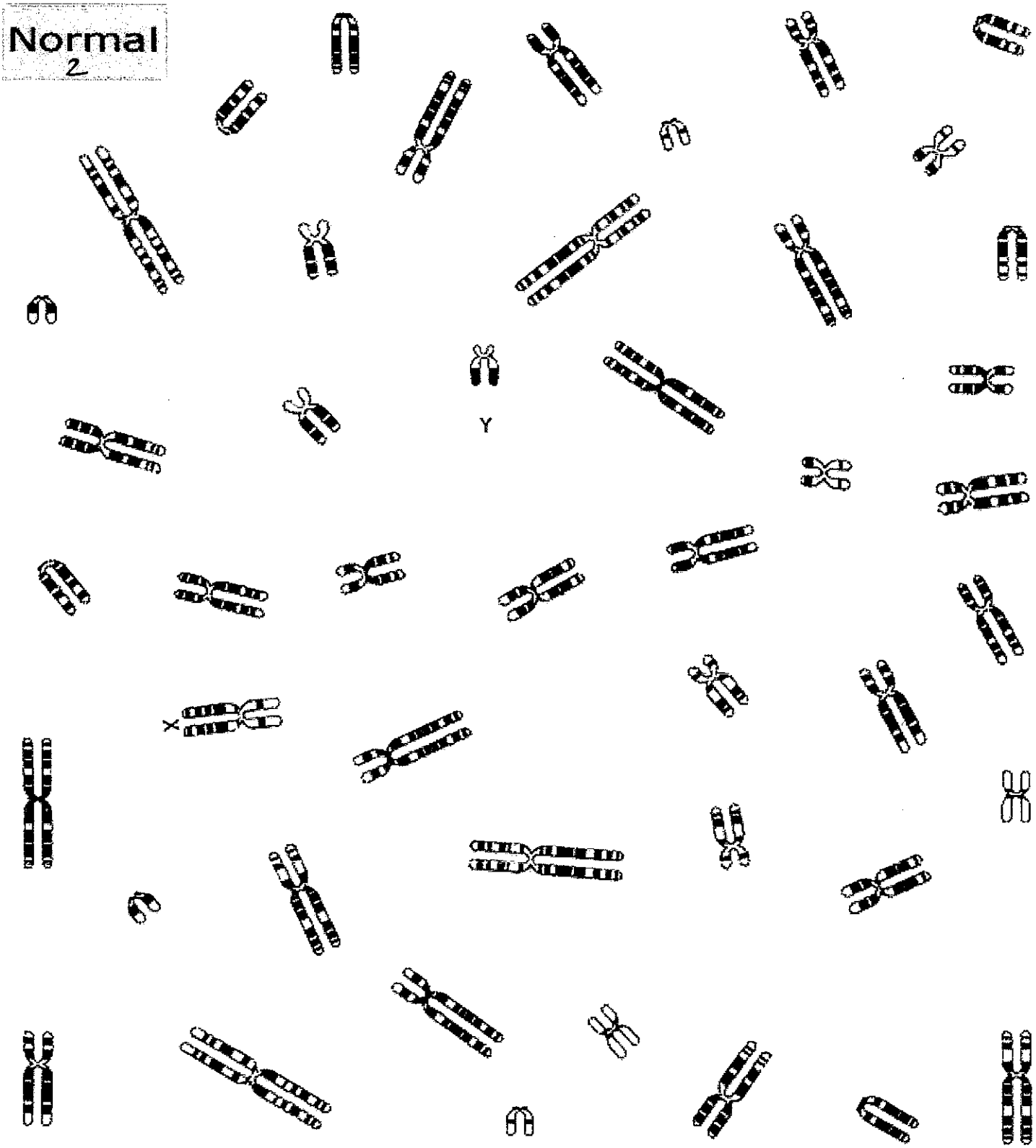
Conclusions

1. **Drawing Conclusions** How is sex determined in a person who has more than two sex chromosomes? Explain your answer.

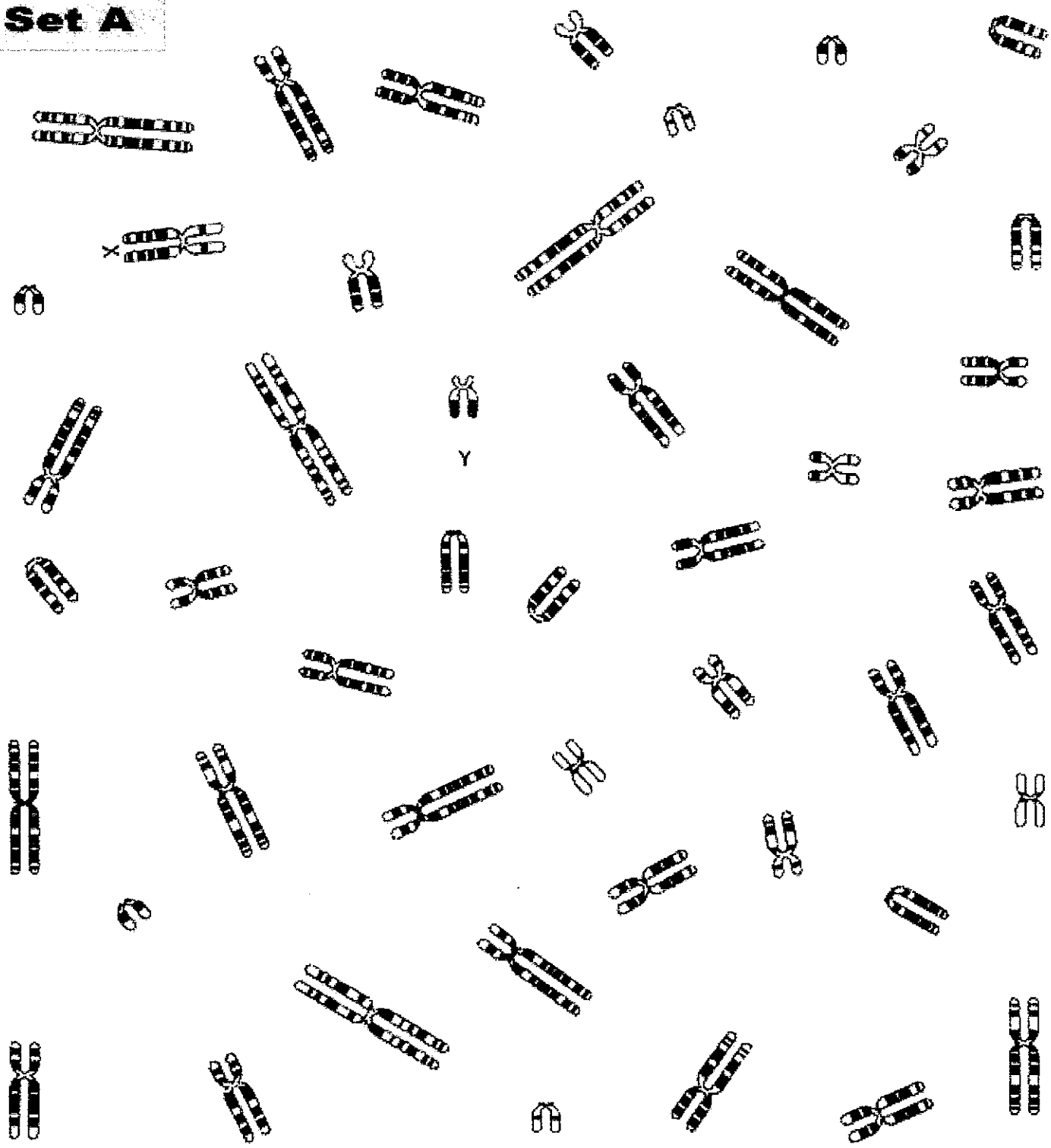
2. **Drawing Conclusions** In this lab, you examined karyotypes for the presence of abnormal chromosome numbers in both autosomes and sex chromosomes. Which condition seems to have a greater influence on a person's health: trisomy of an autosome or trisomy of a sex chromosome?

3. **Making Predictions** Assume that an individual has a deletion mutation in one of their chromosomes. What would the karyotype look like in this situation?

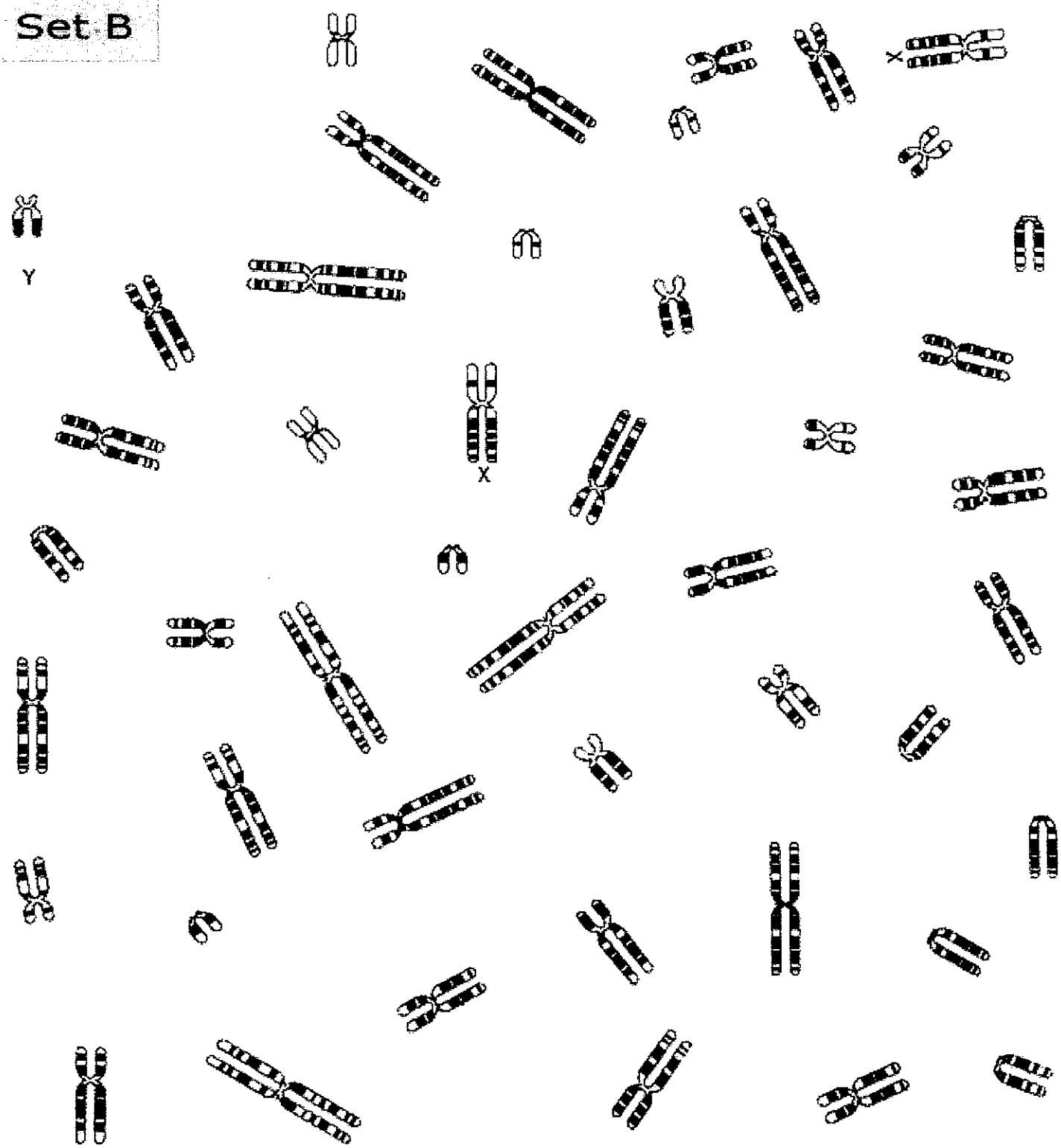

Normal
Y
X


Normal
2
Y
X

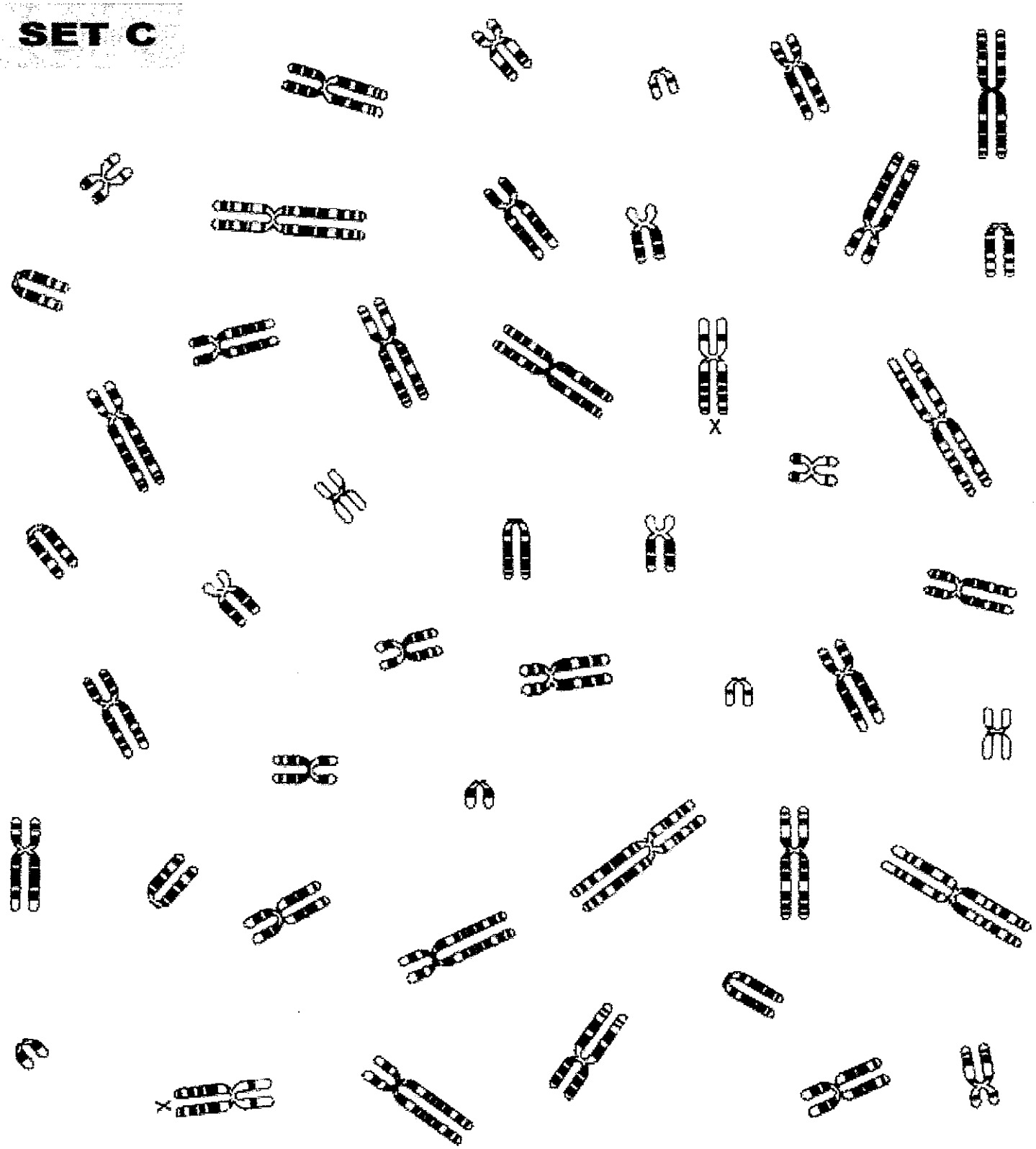
Set A



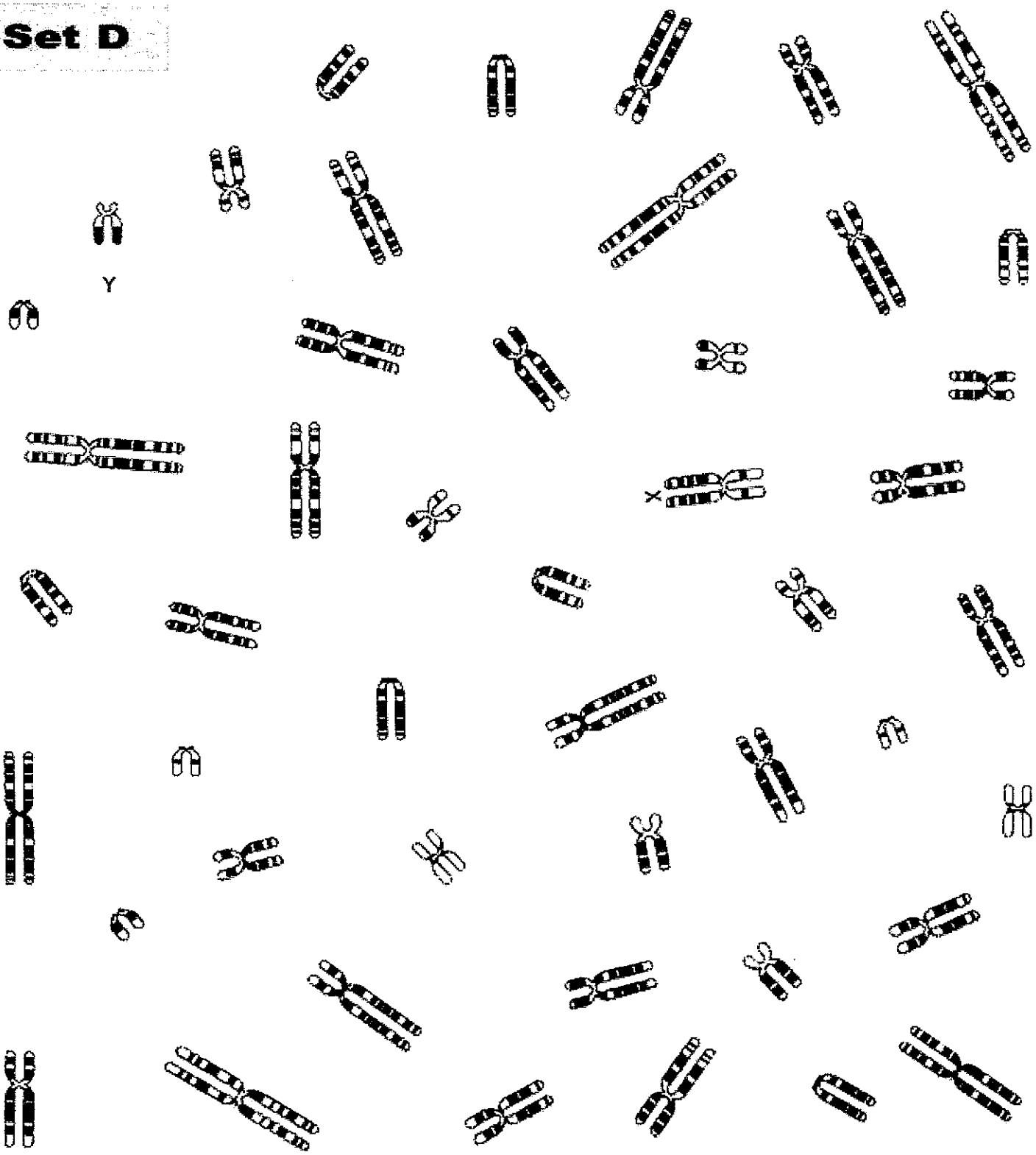
Set B



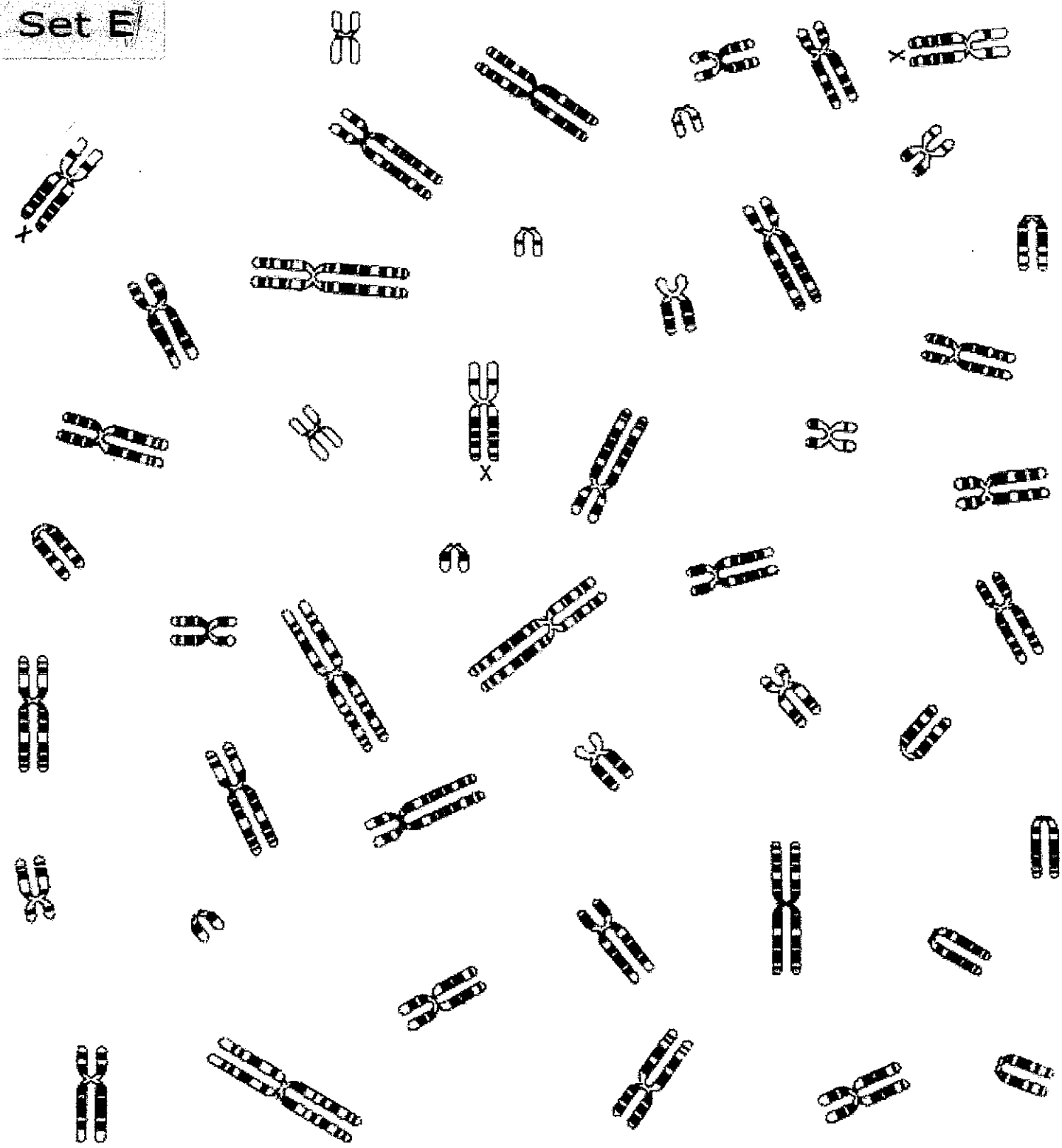
SET C



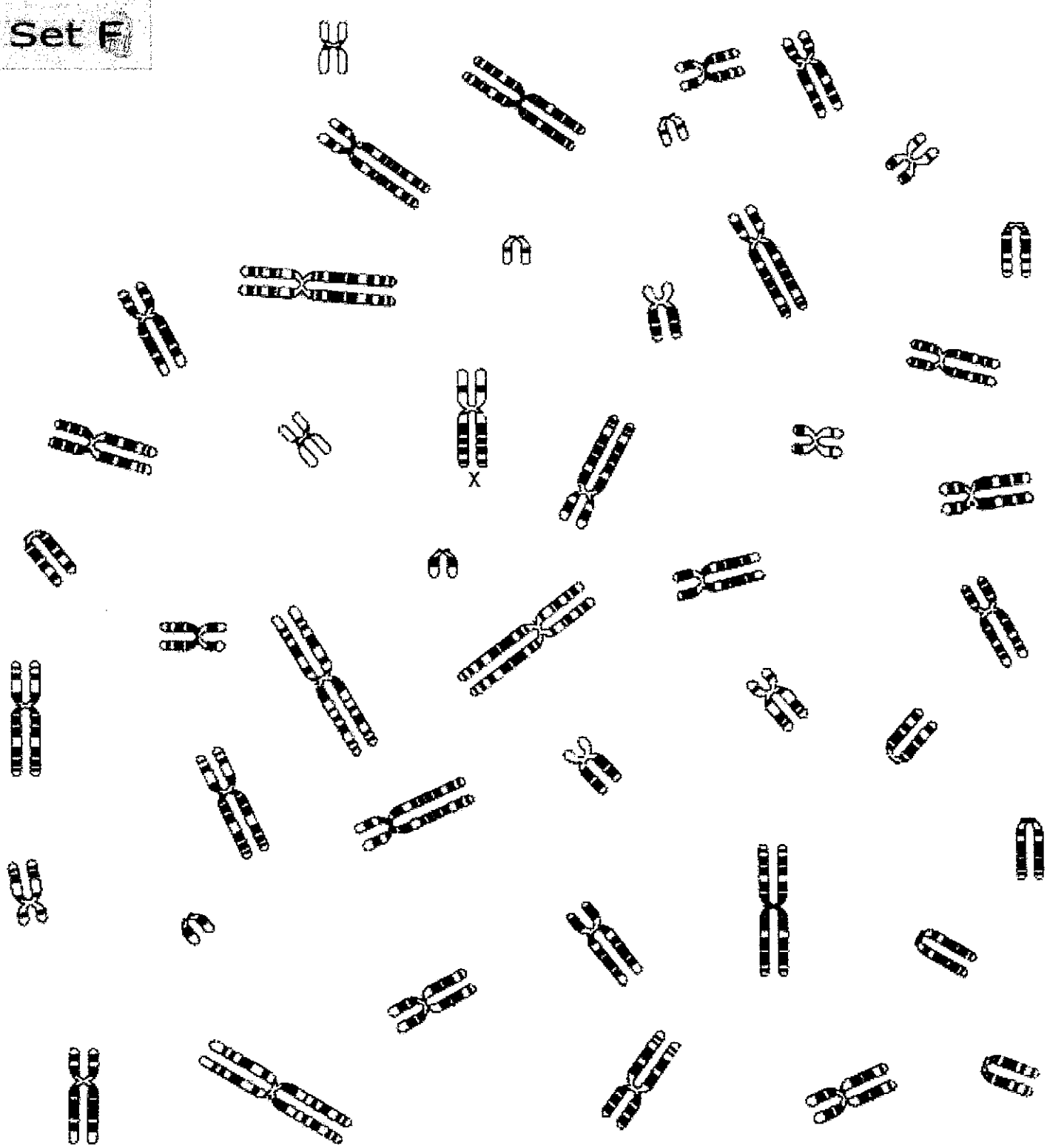
Set D



Set E



Set F



Set 6