

A Chromosome Study

26

An examination of the chromosomes of a cell under high magnification can give much information about an organism. If the cells are from an unborn human, its sex can be determined before it is born. It can also be determined if the unborn may have certain birth defects or problems caused by improper chromosome numbers in its cells.

The pages given to you by your teacher show chromosomes from a normal and abnormal unborn human. These are from body (somatic) cells and have been enlarged about 5000 times natural size.

In this investigation, you will

- (a) learn what a karyotype is.
- (b) prepare a karyotype of a normal human's chromosomes.
- (c) prepare a karyotype of an abnormal human's chromosomes.

Materials



- scissors
- tape
- 1 page of normal chromosomes
- 1 page of abnormal chromosomes
- 2 charts for mounting of chromosomes

The X-like appearance of all chromosomes is confusing to students (especially in light of sex chromosome terminology). The X appearance is due to the fact that karyotyping chromosomes are "caught" in metaphase. Therefore, each chromosome is actually paired with the original and copy still held together by a centromere.

Procedure

Part A. Normal Human Karyotype

● Examine the page marked "Normal Human Chromosomes" supplied by your teacher. These chromosomes are actually an enlarged drawing of what is seen through a microscope. The chromosomes have also been stained to show their "banded" appearance. Note that two chromosomes are unshaded. They have been marked this way to aid you in preparing the karyotype. Cut out each chromosome with scissors. **CAUTION:** *Always be careful with scissors.* To make the task easier and faster, leave margins of paper along each chromosome. Cut them out as rectangles or squares as shown here:



The numbering system on the charts helps to group those chromosomes of similar shape together and thus allows one to refer to the specific pairs by number.

- Prepare a karyotype of these chromosomes. A karyotype is a pattern of chromosomes from one cell grouped into pairs and then organized by size.
- Match all chromosomes into pairs. To help determine pairs, use the banding patterns seen on the chromosomes. Temporarily put the two "unshaded" chromosomes aside. Mount each chromosome pair onto the numbered chart provided by your teacher.

- Position the longest pair on the upper left-hand corner. Consider them as pair number 1. Tape them into place. The next longest pair should follow until all pairs are taped on the sheet in decreasing order of size.

● The two unshaded chromosomes left over should be mounted in the lower right-hand corner above the words "sex chromosomes." Sex chromosomes determine the sex of an organism. In humans, a female results if both sex chromosomes match. These chromosomes are called XX sex chromosomes. In males, sex chromosomes do not match. They are called XY. The Y sex chromosome is much smaller than the X chromosome.

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

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

Body cells are called somatic cells. Somatic cells are skin, liver, muscle, stomach, or kidney cells. The karyotype you just prepared is from a somatic human cell. The term "diploid chromosome number" or " $2n$ number" refers to the total number of chromosomes in any somatic cell of an organism. The diploid number varies from species to species. However, it does not differ from somatic cell to somatic cell of an organism.

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

4. What is the $2n$ chromosome number for your karyotype? _____

5. How many chromosomes would you expect to find in cells taken from the intestine of the person whose karyotype you just prepared?

6. Which sex chromosomes are present in the karyotype you prepared? _____

Analysis

1. Define
 - (a) somatic cell _____
 - (b) karyotype _____
 - (c) diploid or $2n$ chromosome number _____
 - (d) sex chromosomes _____
2. When karyotyping, what two major pieces of information can be gained about a child before it is born?

7. What is the sex of the person in the karyotype you prepared? _____

Part B. Abnormal Human Karyotype

Examine the page marked "Abnormal Human Chromosomes" supplied to you by your teacher. Again, the sex chromosomes are unshaded to make identification easier.

● Prepare a second karyotype as you did the first one. Use a new numbered chart to tape the chromosomes in place.

Finding the incorrect chromosome number in human somatic cells of an unborn alerts a doctor and parents to the fact that their unborn is abnormal and will be born with birth defects. If the unborn has an extra number 8 chromosome, it is born with trisomy 8 syndrome. If the unborn has an extra number 13 chromosome, it is born with Patau syndrome. An extra number 18 chromosome results in Edward syndrome. An extra number 21 syndrome results in Down syndrome. (A syndrome is a series of defects or problems.)

8. How many chromosomes are present in this karyotype? _____

9. What is the diploid chromosome number for this karyotype? _____

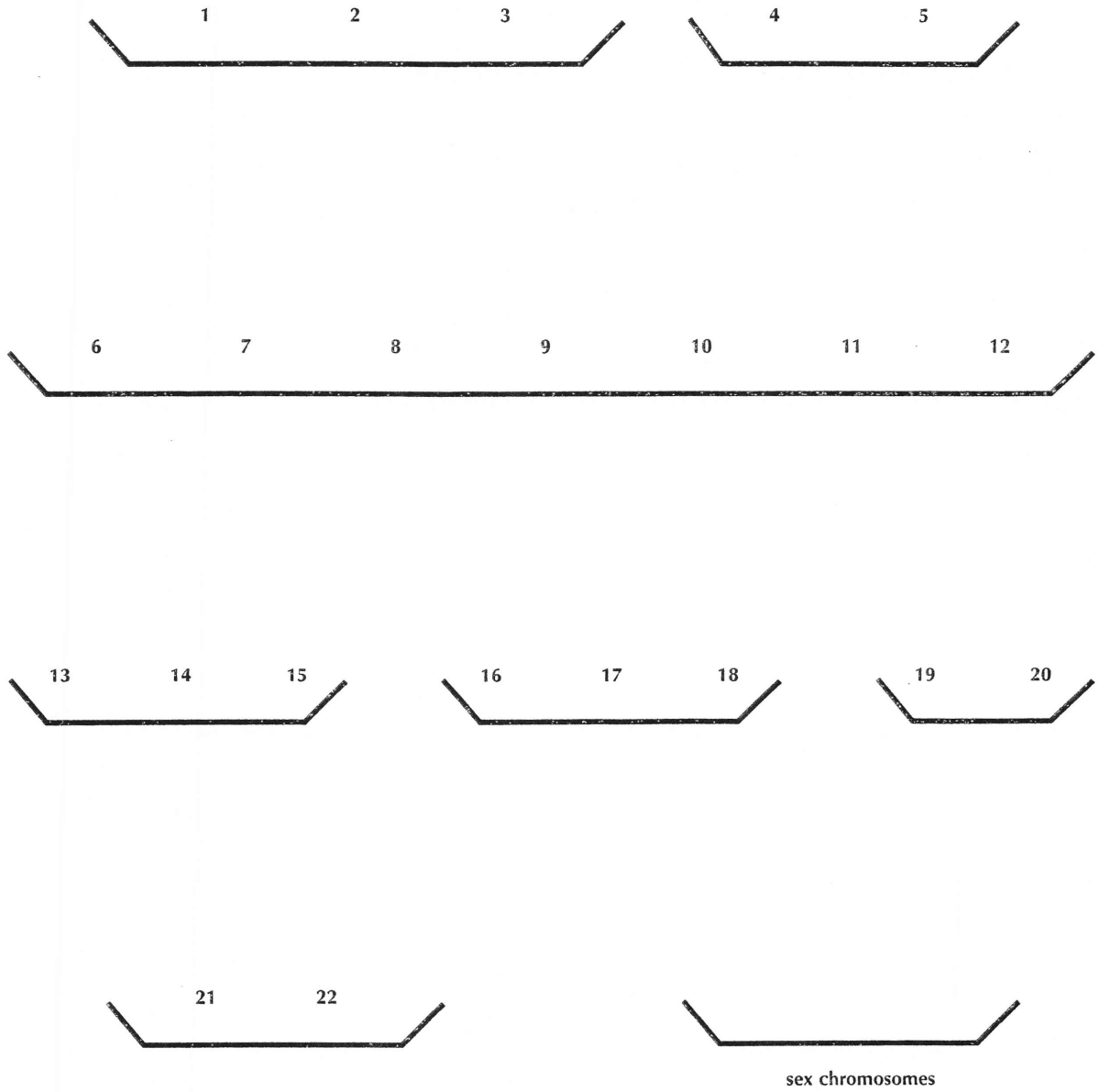
10. (a) Which chromosome pair is abnormal?

(b) How is it abnormal? _____

11. What syndrome does this unborn have? _____

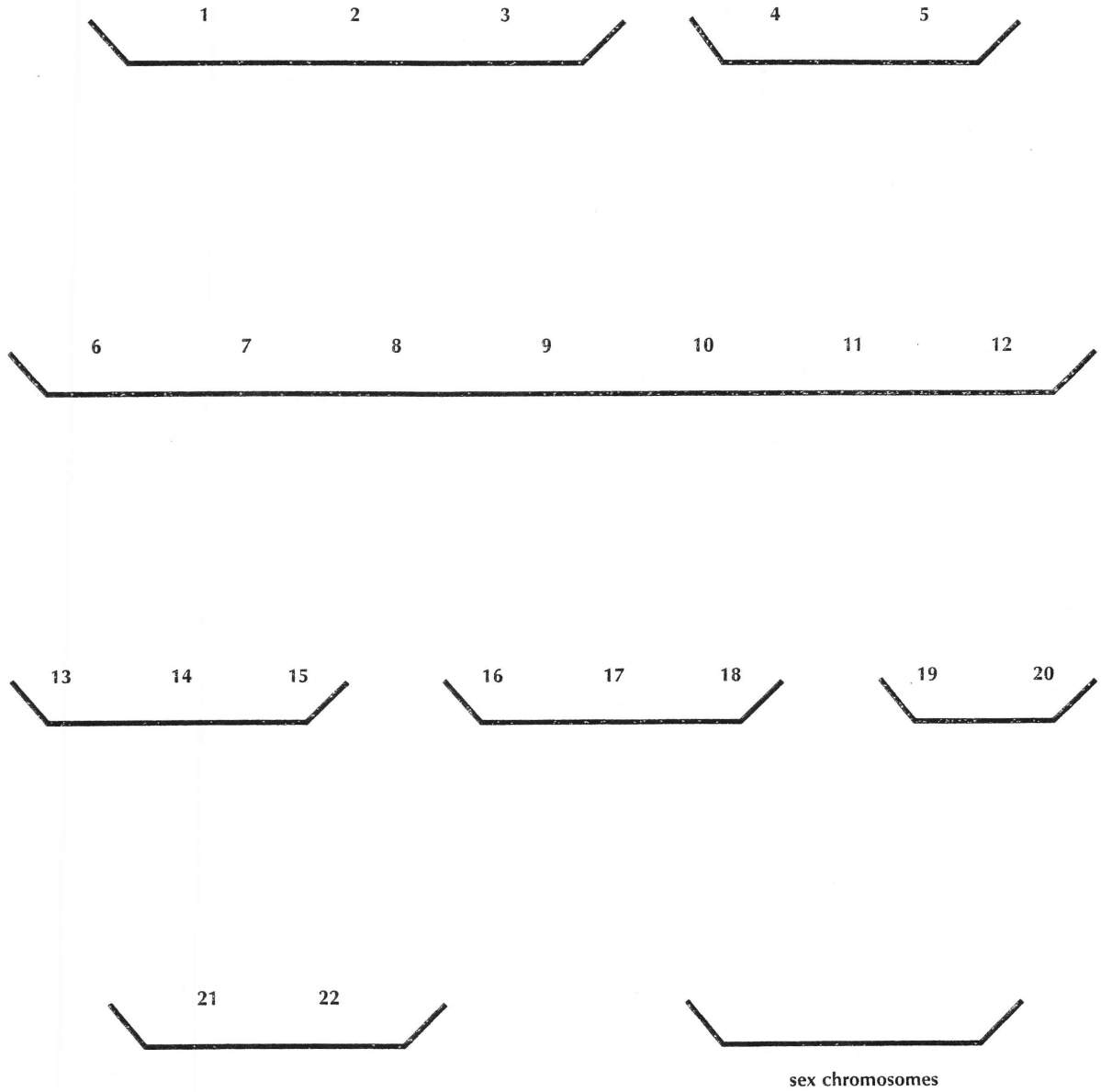
12. What sex will this unborn child be? _____

KARYOTYPE CHART FOR INVESTIGATION 26, "A CHROMOSOME STUDY"



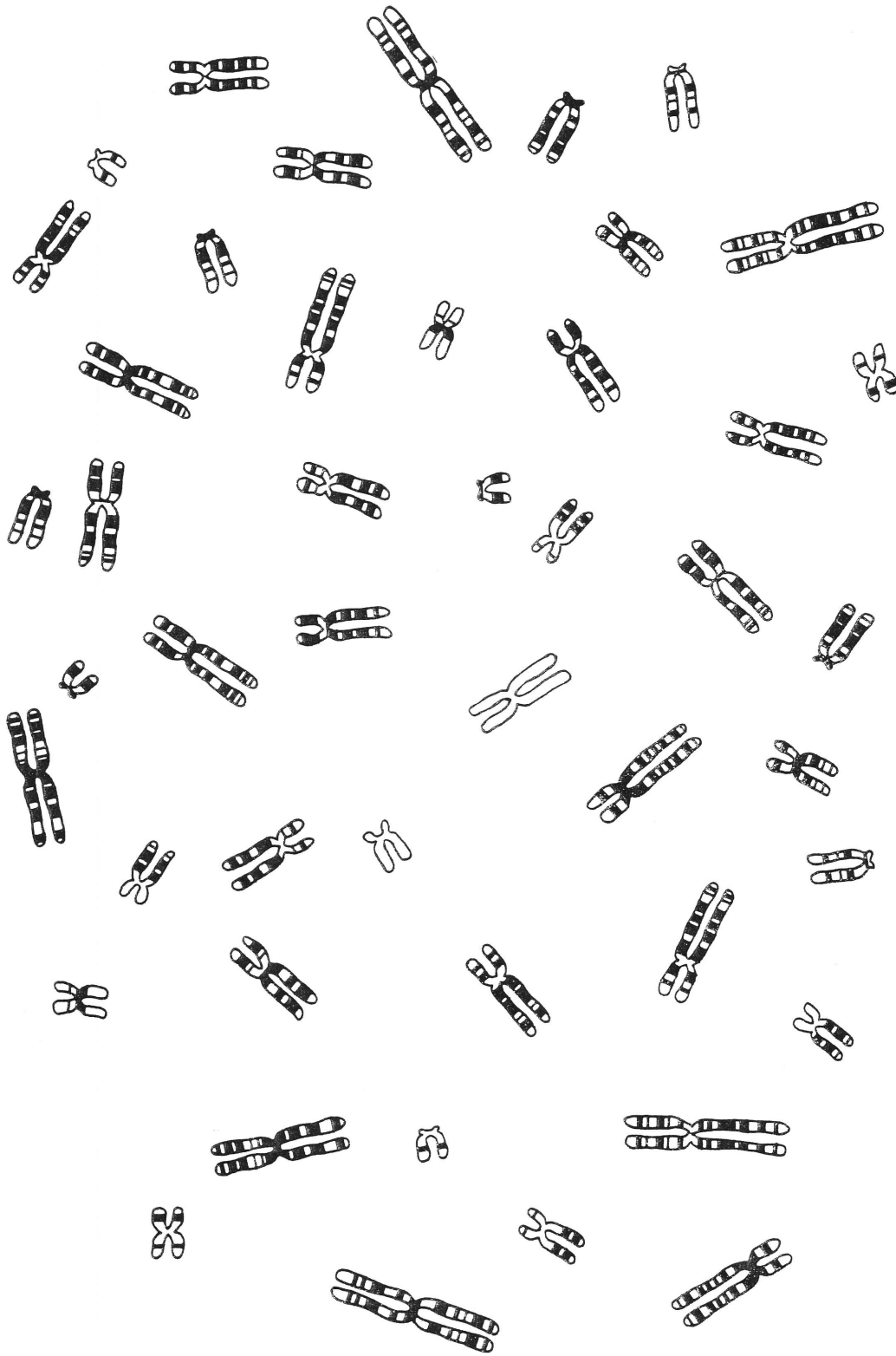
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KARYOTYPE CHART FOR INVESTIGATION 26, "A CHROMOSOME STUDY"



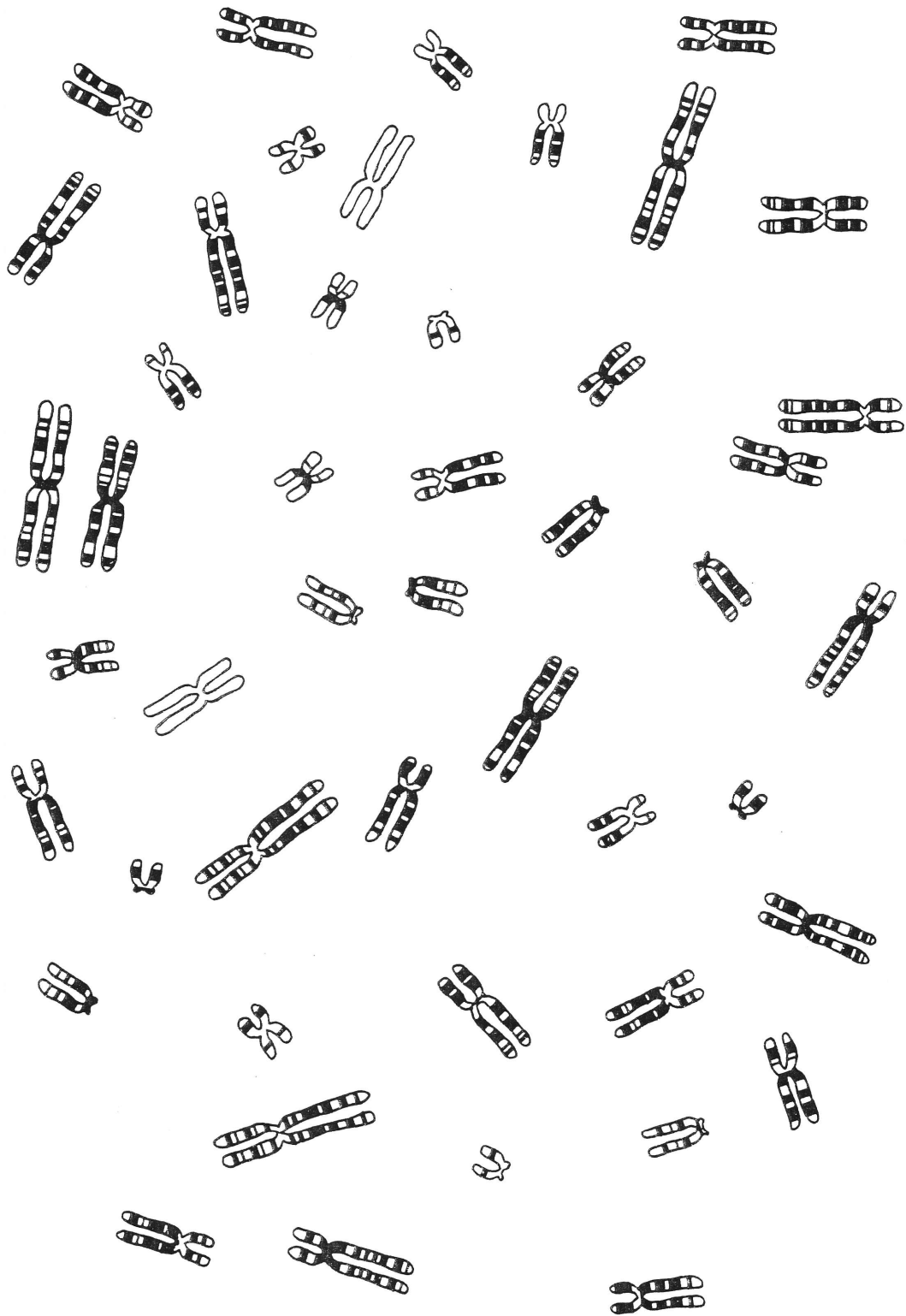
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NORMAL HUMAN CHROMOSOMES FOR INVESTIGATION 26, "A CHROMOSOME STUDY" (PART A)



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ABNORMAL HUMAN CHROMOSOMES FOR INVESTIGATION 26, "A CHROMOSOME STUDY" (PART B)



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