Human Karyotyping Activity

Background:

Occasionally chromosomal material is lost or rearranged during the formation of gametes or during cell division of the early embryo. Such changes, primarily the result of nondisjunction or translocation, are so severe that the pregnancy ends in miscarriage – or fertilization does not occur at all. It is estimated that one in 156 live births have some kind of chromosomal abnormality.

Some of the abnormalities associated with chromosome structure and number can be detected by a test called a karyotype. A karyotype can show prospective parents whether they have certain abnormalities that could be passed on to their offspring, or it may be used to learn the cause of a child's disability. Karyotypes can also reveal the gender of a fetus or test for certain defects through examination of cells from the uterine fluid – a procedure called amniocentesis – or through sampling of placental membranes. Over 400,000 karyotype analyses are performed each year in the U.S. and Canada.

To create karyotypes, chromosomes from a cell are stained and photographed. The photograph is enlarged and cut up into individual chromosomes. The homologous pairs are identified and arranged in order by size (with the exception of the sex chromosomes; these appear last). These tests are typically done on a sample of blood, although any body cell could be used. The cell must be undergoing mitosis – preferably in metaphase – so that the chromosomes are replicated, condensed, and visible under a microscope.

Purpose:

- To understand what a karyotype is and how it is performed
- To understand the reason for performing a karyotype, especially for those with a higher risk of genetic defect in their lineage.
- To determine what genetic defect is present in a chromosome sample.

Procedure:

1. You will be working in groups of four. As a group, you will complete four different karyotypes.
2. Create a data page in your lab notebook. This is where you will glue your homologous chromosomes.
3. Working with your chosen karyotype, start by cutting out the largest chromosome. Match this chromosome with its pair from the same page, and cut this one out as well. Glue BOTH chromosomes side by side on the "data" page. Remember, karyotypes arrange chromosomes from largest to smallest.
4. Continue this procedure until you have matched all chromosomes and glued each of them in the corresponding place on the data page.
5. In the event that you have an extra chromosome, DO NOT THROW IT OUT! It is the chromosome that causes your disorder and you must match it correctly.
6. Once your chromosomes are all cut out and included in the karyotypes, answer the questions in your lab notebook using complete sentences.

Data

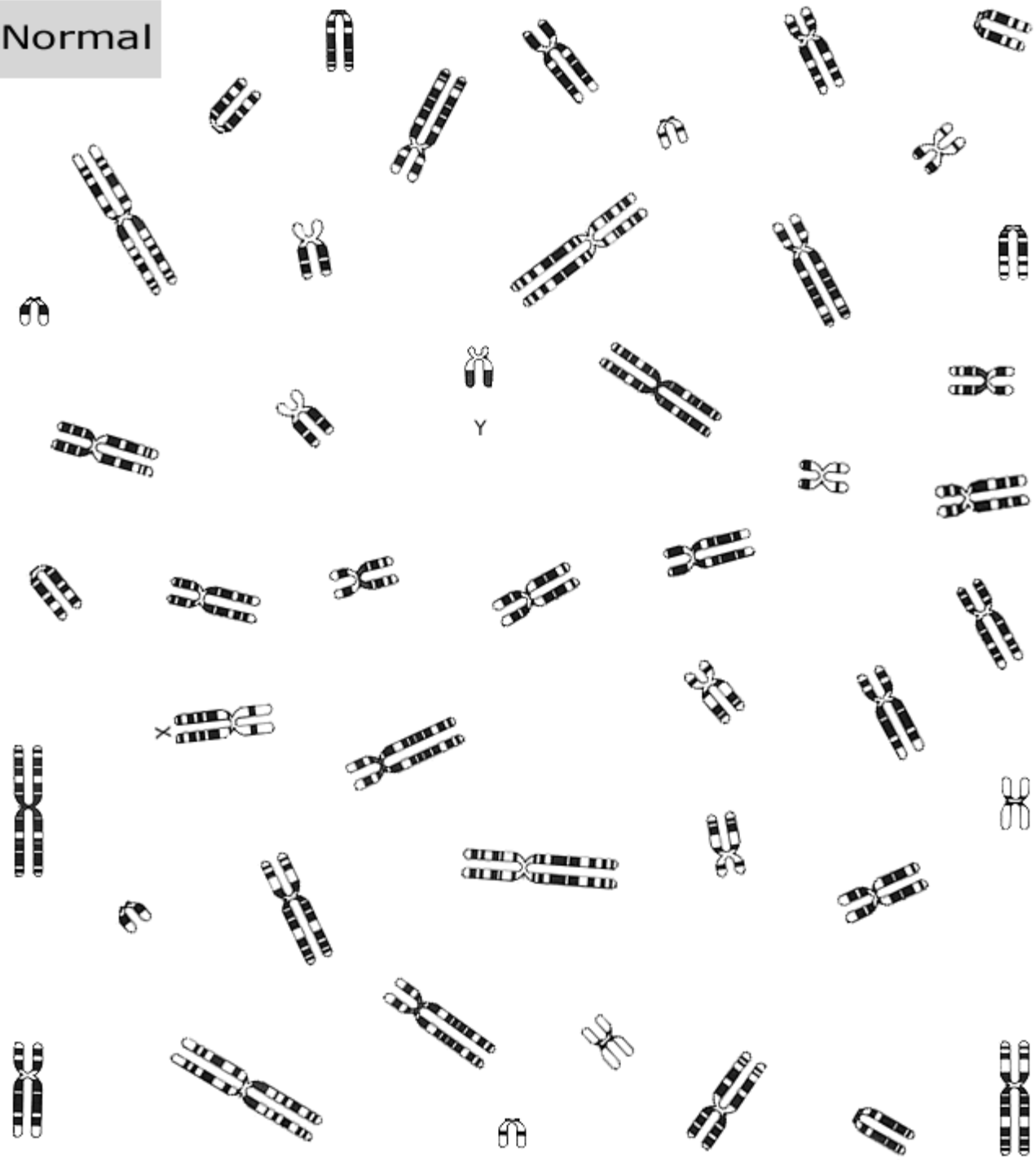
In your lab notebook create a data table where you will glue your chromosomes. You must have 23 boxes, since humans have 23 pairs of chromosomes. Number your boxes 1-22, then X Y

Reviewing Main Ideas Questions

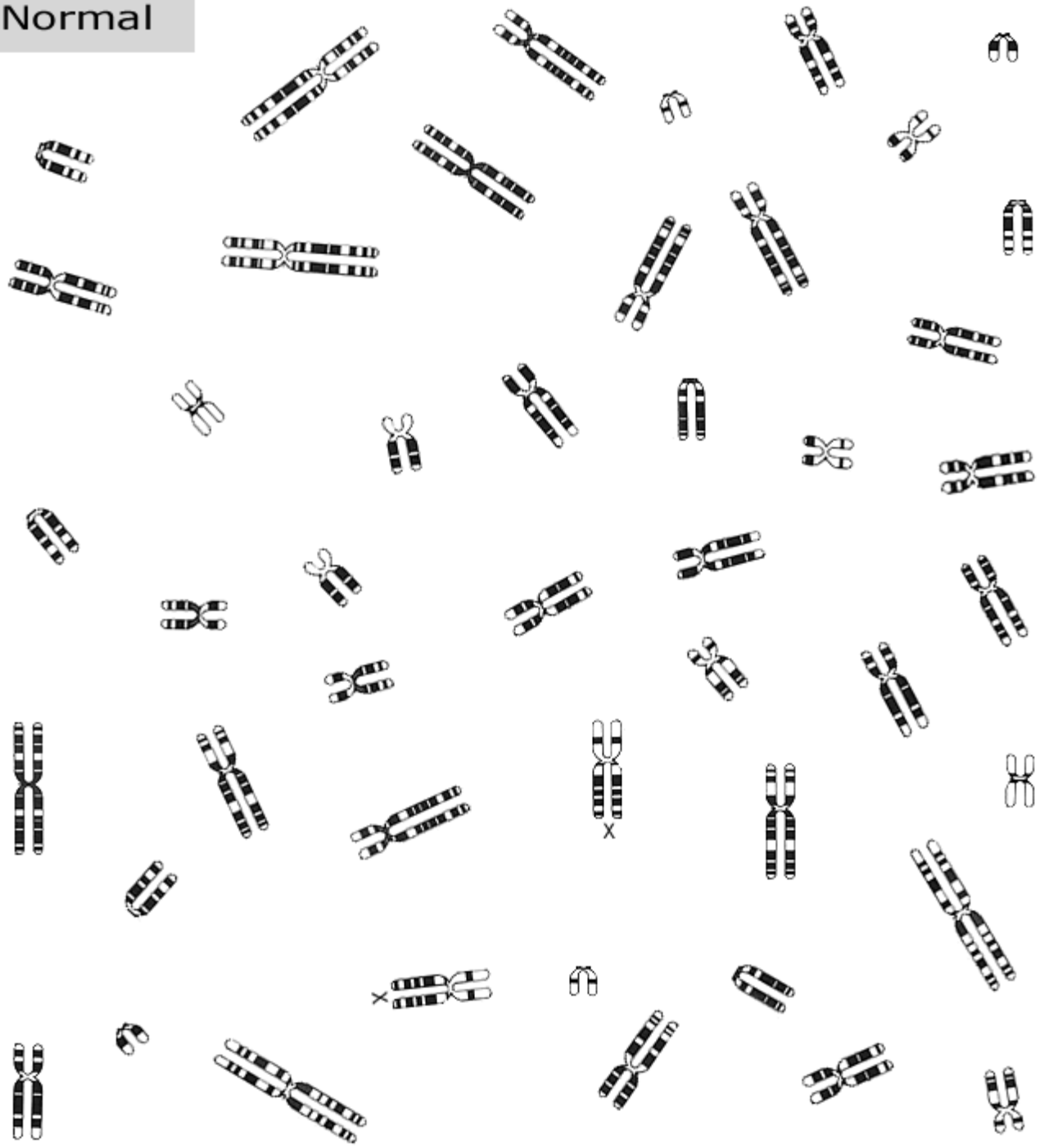
In your lab notebook answer the following questions in complete sentences:

1. Why are there two copies of each chromosome?
2. How many chromosomes does a human somatic cell have?
3. Which chromosome does not have a homologous pair in a male?
4. How can you tell if there is a disorder based on the karyotype?
5. Define *nondisjunction.*
6. What happens as a result of nondisjunction?
7. Define *translocation.*
8. Give two reasons why karyotypes are done.
9. Why must a cell be undergoing mitosis in order to create a karyotype?
10. Does it matter what type of cell is used? Why?

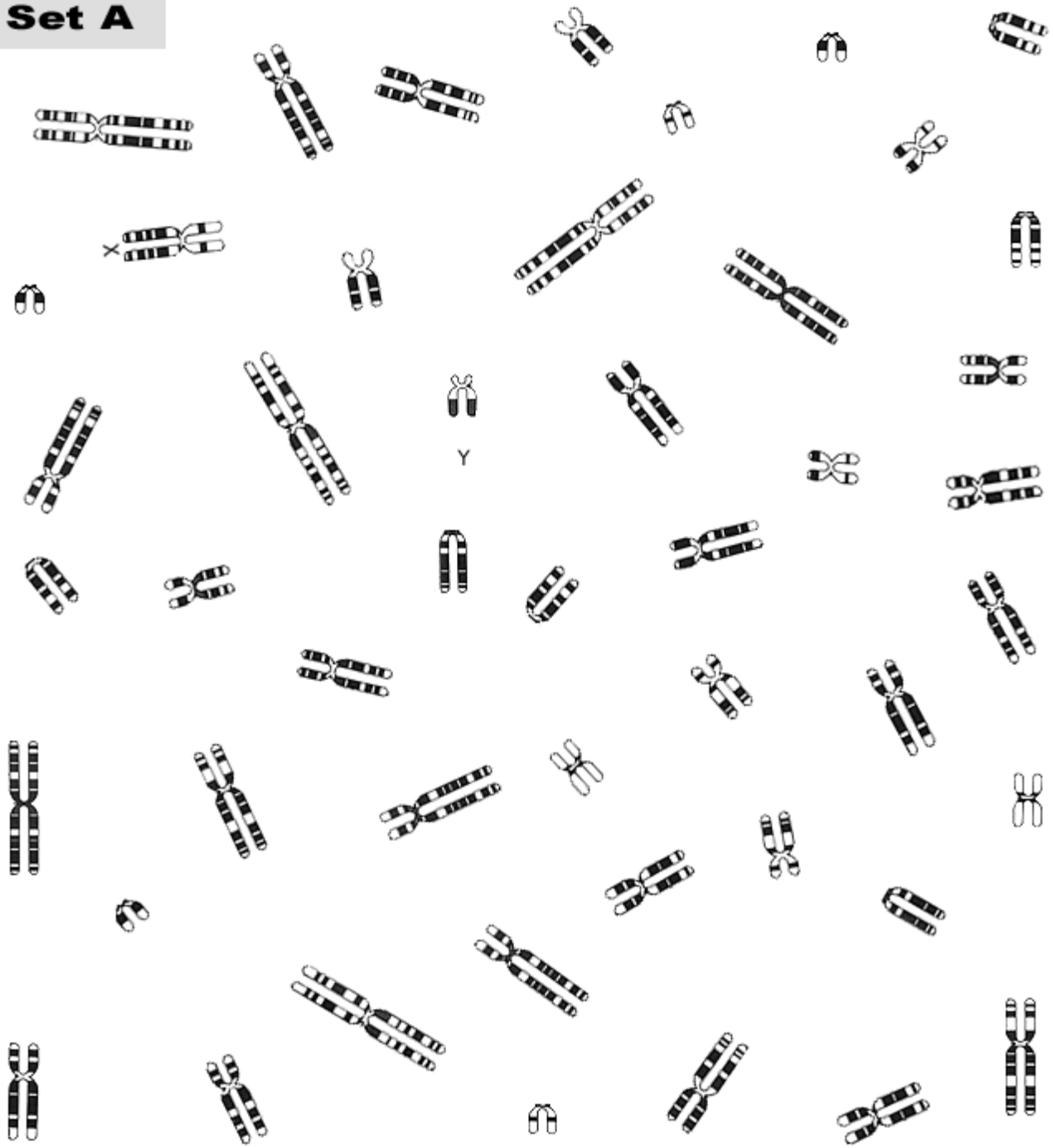
Normal



Normal



Set A



Set B