

NAME _____

Due Date of Lab: _____

Human Karyotyping Lab

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 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 a karyotype, 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.

The purpose of this laboratory experience is:

- understand what a karyotype is and how it is performed.
- 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.
- to investigate a variety of genetic disorders that commonly occur and are studied in biology classes.

Materials: The following materials are needed to perform this laboratory experience:

- Scissors
- tape
- ruler
- small envelope

Procedure: The following procedure is utilized to perform this laboratory experience:

1. Using the attached sheets, complete two different karyotypes: One normal male or One normal female, One from the different disorders of your choice out of the four.
2. Working slowly and carefully, using scissors cut out the chromosome on one page labeled "1" and find its EXACT match elsewhere on the page (it will not be numbered). Cut out this chromosome and tape BOTH chromosomes side by side on a "data page" that has the heading filled out.
3. Continue this procedure until you have matched all chromosomes and taped each of them in the corresponding place on the data page.
4. If you are caught short of time, use the coin envelope to store any chromosomes you may have clipped out and not matched. **DO NOT CUT OUT ALL CHROMOSOMES AND THEN ATTEMPT TO MATCH THEM!!! Cut out only one at a time or you will lose chromosomes.**
5. In the event that you have an extra chromosome, **DO NOT THROW IT OUT!** It is the chromosome that causes your mutation/disorder and you must match it correctly.
6. Once your chromosomes are all cut out and included in the karyotypes, answer the questions and complete the lab.

Human Karyotyping Activity Lab 14 Answers

L Cohen



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