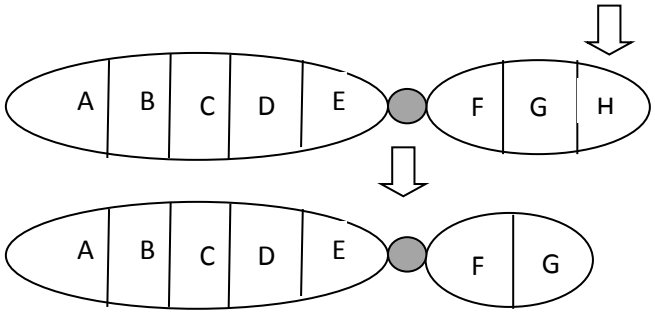
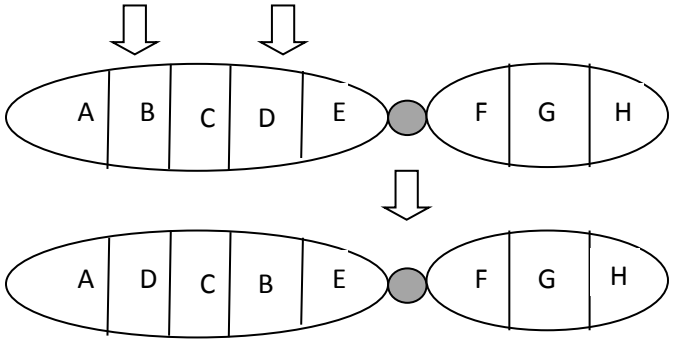
Karyotypes Detect Chromosome Mutations

Chromosomes may become altered during meiosis. These mutations involve large sections that involve many genes. Chromosome may have sections deleted, duplicated, inverted, and translocated. A deletion may occur at the end of a chromosome where a portion breaks off, resulting in a loss of genes.



The diagram shows a chromosome with sections of genes labeled A-H. The arrows indicate that area H has been deleted.

A break may also occur somewhere in the middle of a chromosome and get lost. If the chromosome breaks in the middle and then gets rejoined in reverse order, the chromosome has an inversion mutation. No genetic material is lost when the chromosome is inverted; however, the reverse order will impact transcription and protein production.

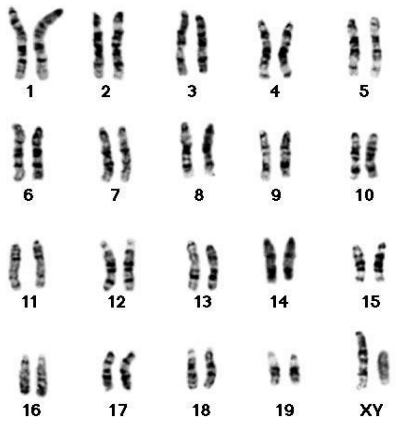


The diagram shows a chromosome with sections of genes labeled A-H. The arrows indicate that area BCD has been inverted.

Sometimes a section of a chromosome may break off during crossing over and attach to a nonhomologous chromosome. This type of mutation is called a translocation. An example of a translocation would be if a piece of chromosome number 3 breaks off and attaches to chromosome number 10. If a section of chromosome breaks off during crossing over and attaches to its homologous chromosome, a duplication occurs. Both of these types of mutations cause problems in gametes that form because some will have extra genes and some will be deficient.

Another type of mutation that may occur during meiosis is nondisjunction. Nondisjunction is the failure of chromosomes to separate properly in cell division. Normally spindle fibers pull homologous chromosomes apart during meiosis; however, if the chromosomes fail to separate and the spindle pulls both sets to one side, then some gametes will have an extra chromosome while the other will be missing an entire chromosome. Nondisjunction can lead to a trisomy condition or a monosomy condition. The most common trisomy condition occurs on chromosome 21, called Down Syndrome. A common monosomy syndrome occurs on the sex chromosome. Turner syndrome is a monosomy X condition in which the female has only one X chromosome.

Scientists use karyotypes to show chromosomal mutations. A karyotype is a picture of all the chromosomes in a cell. It is prepared by capturing cells in the metaphase stage of mitosis and then staining them with a chemical. The stain produces bands on the chromosomes. Each chromosome will show different patterns. Scientists arrange the chromosomes based on size from largest to smallest. The homologous chromosomes are then matched according to their size, shape, banding patterns, and centromere location. A normal human has 46 chromosomes. Of these 46 chromosomes, 44 are autosomes and 2 are sex chromosomes. So a normal male karyotype is written 46, XY, and a normal female karyotype is written 46, XX. Chromosomal disorders can be detected by analyzing the chromosomal pairs. Mutations such as deletions, additions, or nondisjunctions can be detected by carefully inspecting the pairs of chromosomes within a karyotype.



Name _____ Date _____Per_____
Biology

Chromosomal Mutations & Karyotype WebQuest

PreLab Questions

Answer the following questions while reading "Karyotypes Detect Chromosomal Mutations."

1. Draw a chromosomal mutation where a deletion has occurred.

2. What does a deletion mutation result in?

3. What is an inversion mutation?

4. Draw an inversion mutation.

5. What does an inversion mutation impact?

6. What is a translocation mutation?

7. When does a translocation mutation occur?

8. What problems are caused by translocation and duplication mutations?

9. What is nondisjunction?

10. What can nondisjunction lead to? Provide an example of each.

11. What is a karyotype and what is it used for?

12. How are homologous chromosomes matched?

13. How many chromosomes does a normal human have?

14. Write the karyotype for a normal male and a normal female.

WebQuest Procedure:

1. Enter the web address:

www.biology.arizona.edu/human_bio/activities/karyotyping/karyotyping.html

2. Read the introduction and answer the following questions:

    a. What do banding patterns on chromosomes represent?

    b. How are chromosomes analyzed within a karyotype?

3. Read the assignment. You will evaluate three patient histories, complete their karyotypes, and diagnose any missing or extra chromosomes.

4. Click on Patient Histories at the bottom of the page.
5. Scroll to the bottom of the page and click on Patient A.
6. Follow the instructions on the screen. Your mission for each patient is to match the chromosome given to the ones on the karyotype. If you are correct, the computer will give you another chromosome to match. If you are wrong, the computer will ask you if you want to "Try Again" at the bottom of the screen; Click this to retry. You will repeat these steps until all of the chromosomes are matched. A screen will then pop up, congratulate you on preparing the karyotype, and ask you to diagnosis the patient using two questions.
7. Complete the data table for each patient.

Patient	Patient's History	Completed Karyotype	Diagnosis
A			
B			
C			