

CLASS SET

How Can Karyotype Analysis Explain Genetic Disorders?

A karyotype is a picture which the chromosomes of a cell have been stained so that the banding pattern of the chromosomes appears. Cells in metaphase of cell division are stained to show distinct parts of the chromosomes. The cells are then photographed through the microscope, and the photograph is enlarged. The chromosomes are cut from the photograph and arranged in pairs according to size, arm length, centromere position, and banding patterns. Karyotypes have become of increasing importance to genetic counselors as disorders and diseases have been traced to specific visible abnormalities of the chromosomes.

OBJECTIVES:

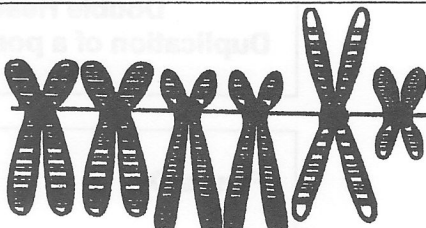
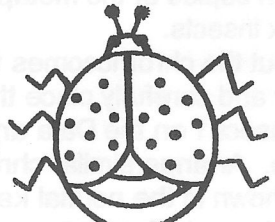
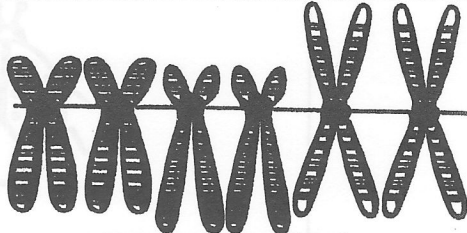
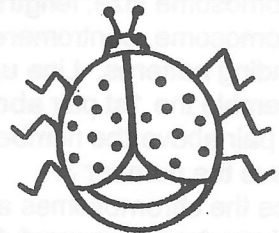
- * Construct a karyotype from the metaphase chromosomes of a fictitious insect.
- * Analyze prepared karyotypes for chromosome abnormalities.
- * Identify the genetic disorders of six fictitious insects by using the insects' karyotypes.

MATERIALS:

- * Photocopies of metaphase chromosomes from six insects
- * Scissors
- * Tape

PROCEDURE:

For this investigation, assume that a new species of insect has been discovered. This insect has three pairs of very large chromosomes. Researchers have been able to trace four genetic disorders to specific chromosomal abnormalities in this insect. Study the karyotypes and phenotypes of normal male and female insects as illustrated below.

 <p>Normal karyotype - male</p>	 <p>Normal phenotypic male</p>
 <p>Normal karyotype - female</p>	 <p>Normal phenotypic female</p>

Note that the normal male insect has a pair of sex chromosomes similar to those of the human male, one large and one small. In the same way, the female has a pair of sex chromosomes similar to those of the human female, both large. These sex chromosomes make up the third pair of chromosomes.

(Size Reduction Disorder)

This disorder appears when there is a *monosomy* (only one chromosome of the pair) of the sex chromosomes (3rd pair). A single large chromosome produces a small *female* insect, while a single small chromosome produces a small *male* insect. See fig. A.

(Clear Wing Disorder)

This disorder appears as a result of a *trisomy* (three chromosomes instead of two) of the 2nd pair of chromosomes. The extra chromosome produces sterile insects that also lack coloring in their wings. See fig. B.

(Double Head Disorder)

A duplication of a portion of one chromosome from the 1st pair produces an insect with a double head. The affected chromosome appears slightly larger than a normal chromosome for that pair. This disorder also produces banding on the wings and additional body segments. See fig. C.

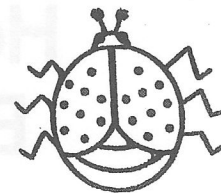
(Un-segmented Disorder)

The deletion of a short segment of the large sex chromosome (3rd pair) results in a loss of body segmentation and a reduction of body size. The affected chromosome is only very slightly smaller than normal so only great care in constructing the karyotype will reveal this condition. See fig. D.

Procedure:

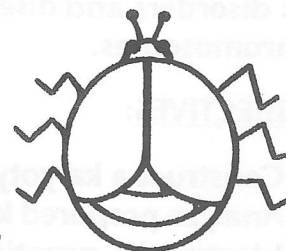
1. Obtain copies of the metaphase chromosomes of six insects.
2. Cut out the chromosomes for insect 1 from the copy and carefully place them along the line for insect 1 on the Data and Observations page. Arrange similar chromosomes together as shown in the normal karyotypes in Fig. 1. Match up similar chromosomes by comparing chromosome size, length of the arms of each chromosome, centromere position, and banding patterns. Line up chromosomes that resemble the 1st pair above the Number 1, the 2nd pair above the number 2, and the 3rd pair above the number 3.
3. Once the chromosomes are positioned, tape their centromeres carefully to the line. This represents the karyotype for one insect.
4. Repeat steps 2 & 3 for the rest of the insects.

Figure A



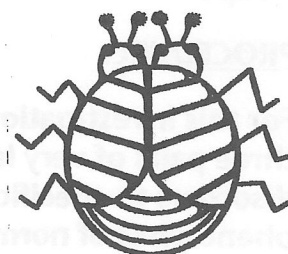
Size Reduction Disorder
Monosomy of the 3rd pair

Figure B



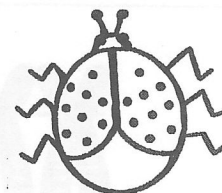
Clear Wing Disorder
Trisomy of the 2nd pair

Figure C



Double Head Disorder
Duplication of a portion of the 1st pair

Figure D



Un-segmented Disorder
Deletion of segment of 3rd pair

Name: _____ Date: _____ Per: _____

Name: _____

Insect Karyotype Lab

Insect 1

Insect 2

(1st pair) (2nd pair) (3rd pair)

(1st pair) (2nd pair) (3rd pair)

Insect 3

Insect 4

(1st pair) (2nd pair) (3rd pair)

(1st pair) (2nd pair) (3rd pair)

Insect 5

Insect 6

(1st pair) (2nd pair) (3rd pair)

(1st pair) (2nd pair) (3rd pair)

Analysis Questions:

1. Identify:

- 1) insect sex (male or female)
- 2) genetic disorder name (size reduction, clear wing, etc...) (if any)
- 3) chromosome error (monosomy, trisomy, etc...) (if any)
- 4) chromosome location (1st pair, 2nd pair, 3rd pair) (if applicable)

	Sex	Genetic Disorder Name	Chromosome Error	Chromosome Location
Insect 1				
Insect 2				
Insect 3				
Insect 4				
Insect 5				
Insect 6				

2. Which disorder that you found was the hardest to detect? _____

Why? _____

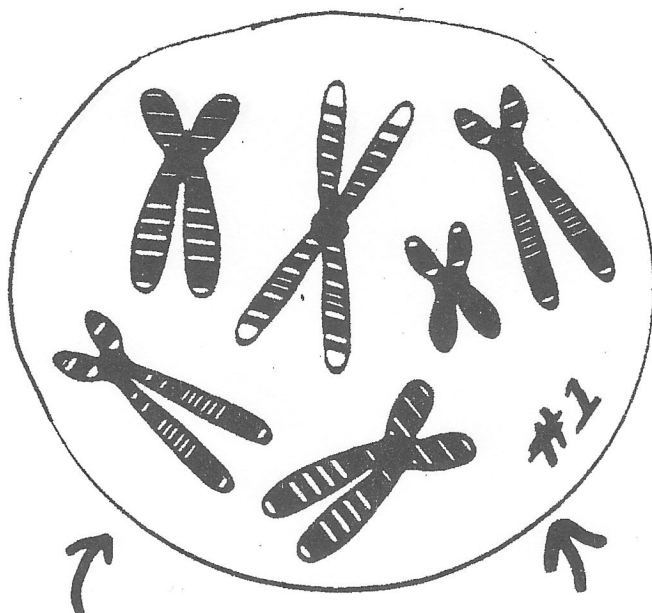
3. Which disorder that you found was the easiest to detect? _____

Why? _____

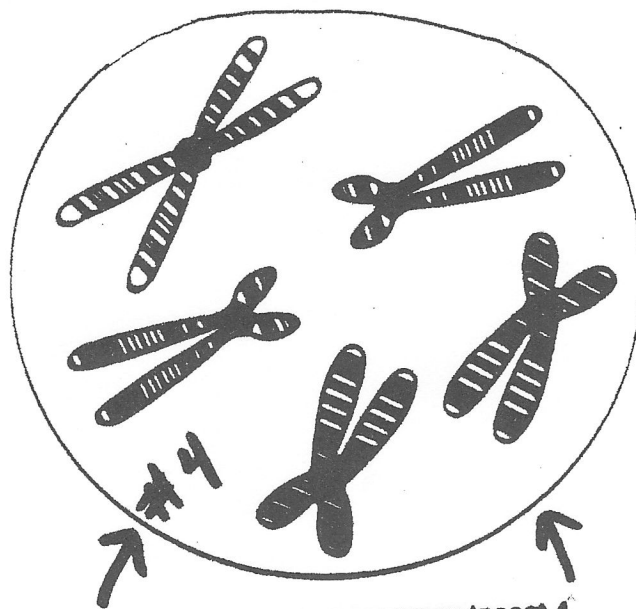
4. How can a mutation on one chromosome affect more than one thing on an organism?

Example: A duplication of one of the chromosomes of the 1st pair produces a double head and, at the same time, affects the wing pigmentation and body segmentation.

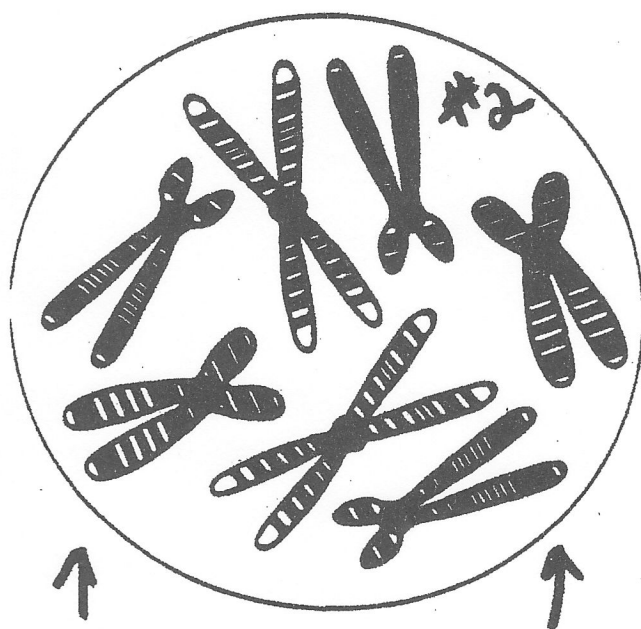
5. What kind of information would be required if karyotype analysis were to be used to detect the genetic of real organisms? _____



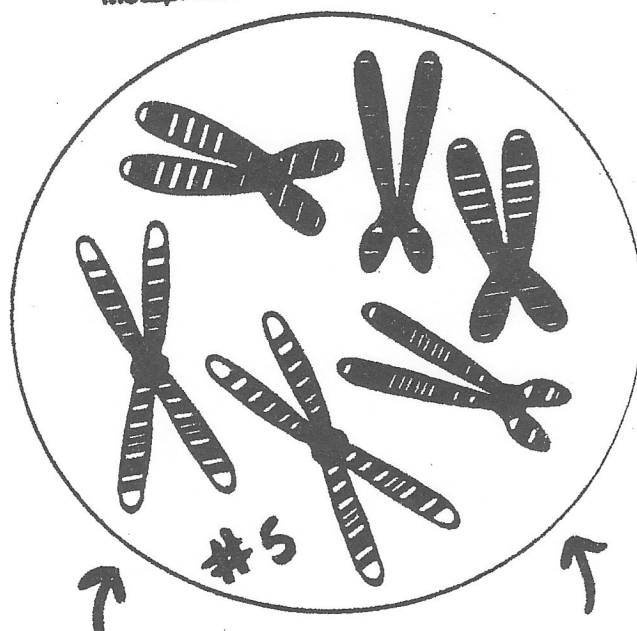
Metaphase chromosomes Insect 1



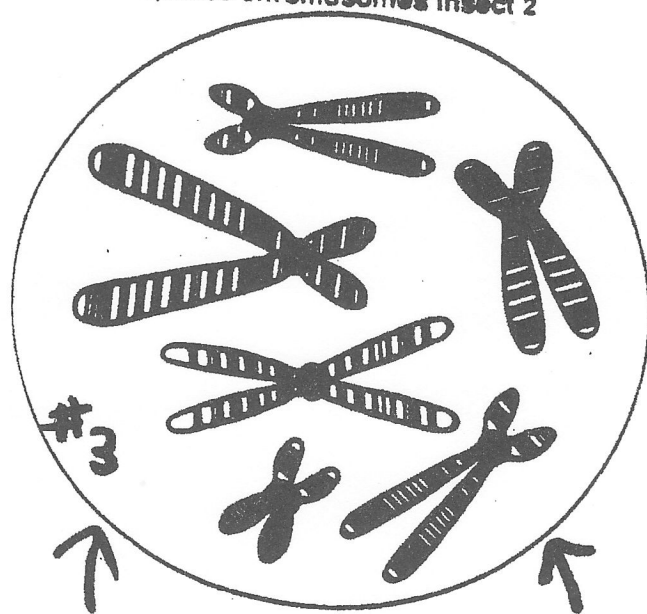
Metaphase chromosomes Insect 4



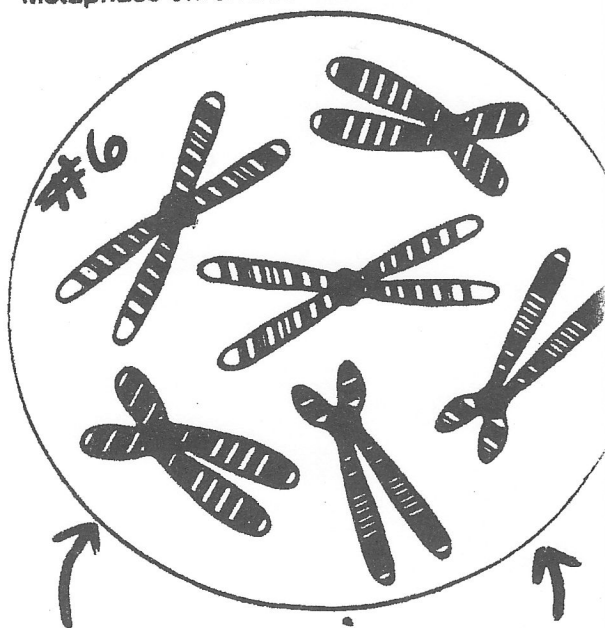
Metaphase chromosomes Insect 2



Metaphase chromosomes Insect 5



Metaphase chromosomes Insect 3



Metaphase chromosomes Insect 6