



How Can Karyotype Analysis Detect Genetic Disorders?

Lab 12-2

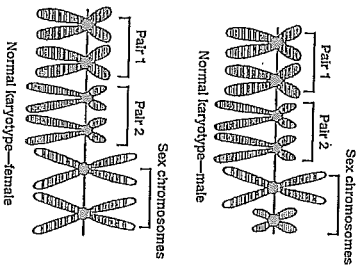
A karyotype is a picture in which the chromosomes of a cell have been stained so that the banding pattern of the chromosomes is visible. Cells in metaphase of cell division are stained to show distinct parts of the chromosomes. The cells are then photographed through the microscope, and the photograph 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 increasingly important to genetic counselors as disorders and diseases have been traced to specific visible abnormalities of the chromosomes.

OBJECTIVES

- Construct karyotypes from the metaphase chromosomes of six fictitious insects.
- Analyze the karyotypes for chromosome abnormalities.
- Identify the genetic disorders of the insects by using their karyotypes.
- Hypothesize how karyotype analysis can be used to detect genetic disorders.

PROCEDURE

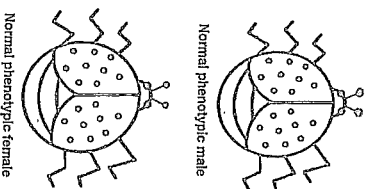
For this Investigation, assume that a new species of insect has been discovered. The insect has three pairs of very large chromosomes. Researchers have been able to trace four genetic disorders to specific

Figure 1


MATERIALS

- photocopies of metaphase chromosomes from six fictitious insects (2 pages)
- scissors
- glue
- goggles

chromosomal abnormalities in this insect. Study the karyotypes and phenotypes of normal male and female insects as illustrated in Figure 1.



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PROCEDURE CONTINUED

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 chromosome pair 3.

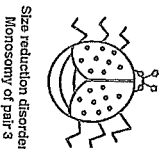
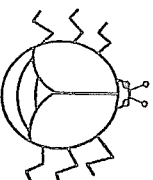
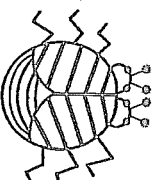
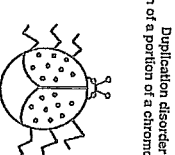
The disorder known as size reduction disorder appears when there is a monosomy of the sex-chromosome pair. A single large sex chromosome produces a small female insect. A single small sex chromosome produces a small male insect. This disorder is shown in Figure 2.

Clear wing disorder, as shown in Figure 3, appears to result from trisomy of the chromosomes of pair 2. The extra chromosome of the second pair produces sterile insects that lack coloring in their wings. Since sterility always results, the clear wing disorder is not passed on to progeny.

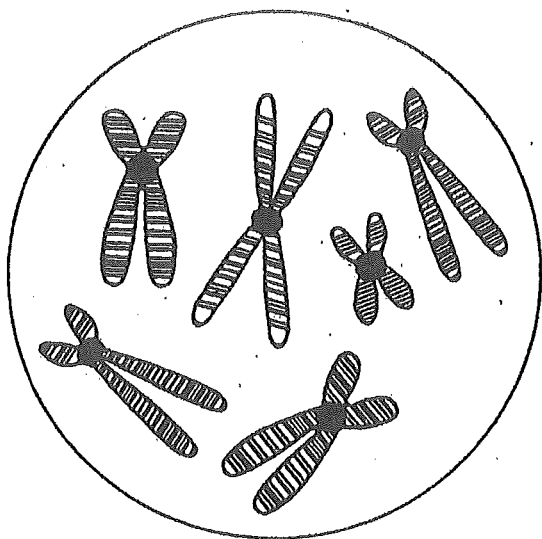
A duplication of a portion of a chromosome from pair 1 produces an insect with a double head. This duplication also produces banding on the wings and additional body segments. See Figure 4.

The deletion of a short segment of the large sex chromosome results in a loss of body segmentation and a reduction of body size. This disorder is shown in Figure 5.

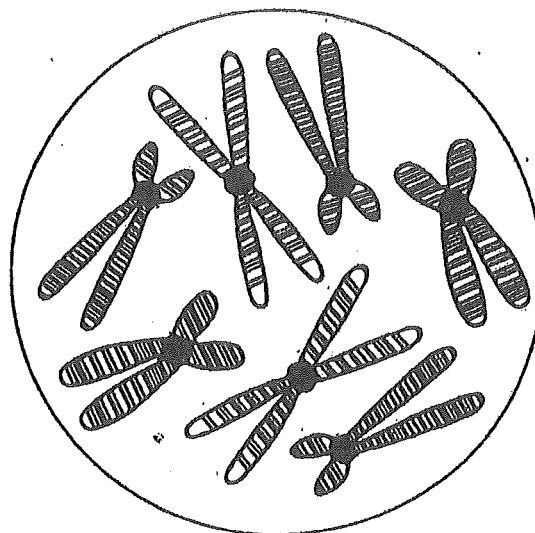
1. Obtain copies of the metaphase chromosomes of six insects from the teacher.
2. Write a hypothesis that describes how karyotype analysis can be used to detect the presence of a genetic disorder. Write your hypothesis in the space provided.
3. Cut out the chromosomes for insect 1 from the photocopy and place them along the line for insect 1 in Data and Observations. Arrange similar chromosomes together as shown in the normal karyotypes in Figure 1. Match up similar chromosomes by comparing chromosome size, length of the arms of each chromosome, centromere position, and banding patterns. Be sure to line up chromosomes that resemble the first pair of the normal karyotype above the number 1, those that resemble the second pair above the number 2, and those that resemble the third pair (sex chromosomes) above the number 3.

Figure 2

Figure 3

Figure 4

Figure 5


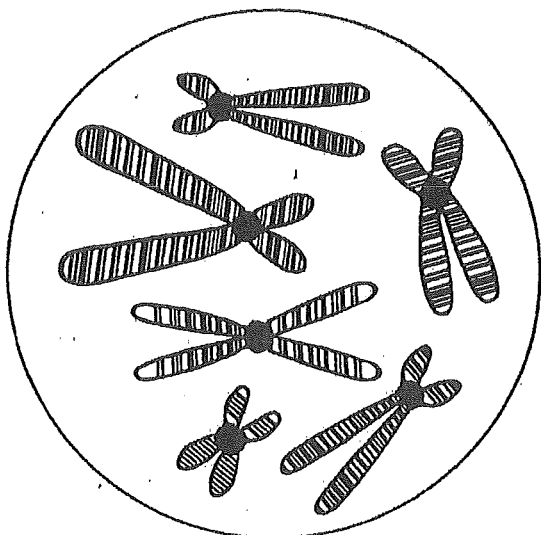
4. Once the chromosomes are positioned, glue their centromeres to the straight line. This represents the karyotype for one insect.
5. Repeat steps 3 and 4 for each of the other fictitious insects.



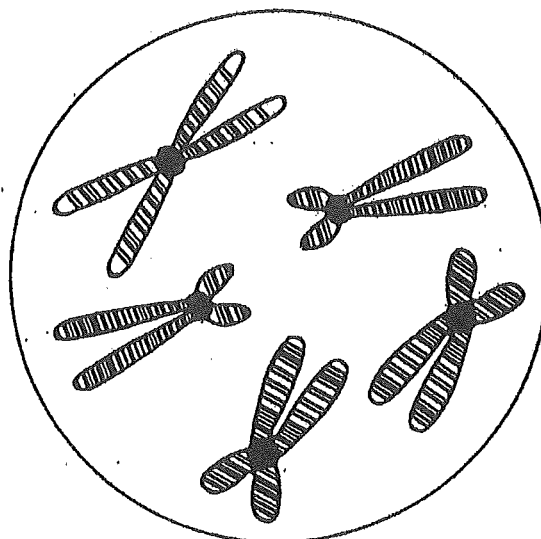
Metaphase chromosomes Insect 1



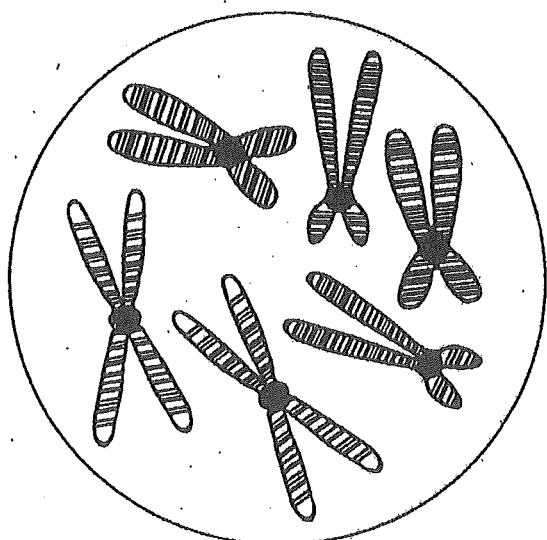
Metaphase chromosomes Insect 2



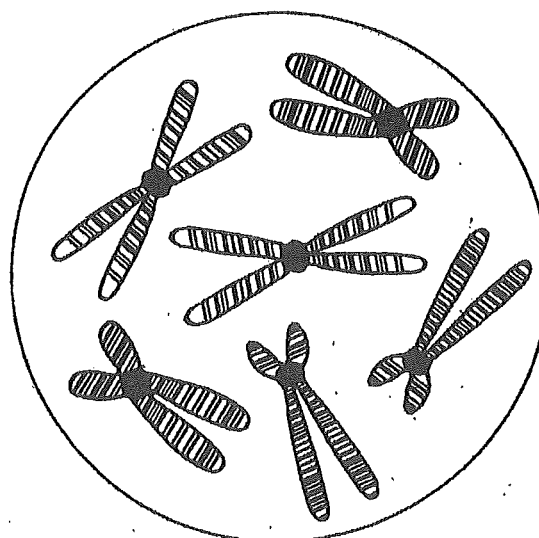
Metaphase chromosomes Insect 3



Metaphase chromosomes Insect 4



Metaphase chromosomes Insect 5



Metaphase chromosomes Insect 6

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- ## HYPOTHESIS

DATA AND OBSERVATIONS

Insect 2

1

Insect 4

1

Insect 6

1 **2** **3**

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ANALYSIS

1. Identify the sex, chromosome error, and genetic disorder for each of the fictitious insects.

	Sex	Chromosome error	Genetic disorder
Insect 1	_____	_____	_____
Insect 2	_____	_____	_____
Insect 3	_____	_____	_____
Insect 4	_____	_____	_____
Insect 5	_____	_____	_____
Insect 6	_____	_____	_____

2. Which type of chromosome abnormality is the most difficult to detect by means of a karyotype?

The easiest? _____

Why? _____

3. How can duplication of a chromosome of the first pair produce a double head and, at the same time, affect the wing pigmentation and body segmentation?

4. What kind of information is required in using karyotypes to detect genetic disorders in real organisms?

CHECKING YOUR HYPOTHESIS

Was your hypothesis supported by your observations? Why or why not?

FURTHER INVESTIGATIONS

- Books on human genetics will have discussions of chromosome abnormalities and karyotype analysis. Borrow a book from the teacher or the library and investigate some of the common abnormal karyotypes in humans.
- Obtain additional copies of metaphase chromosomes from the six fictitious insects. Alter the chromosomes and construct new karyotypes using the chromosomes. Imagine how the resulting insects might look. Draw pictures of the new fictitious insects.