How Can Karyotype Analysis Explain Genetic Disorders? 15–2

INVESTIGATION

LAB

A karyotype is a picture in 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.
- Hypothesize how karyotype analysis can be used to explain the presence of a genetic disorder.

MATERIALS

photocopies of metaphase chromosomes from six insects (2 pages)

scissors

rubber cement

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 in Figure 1.

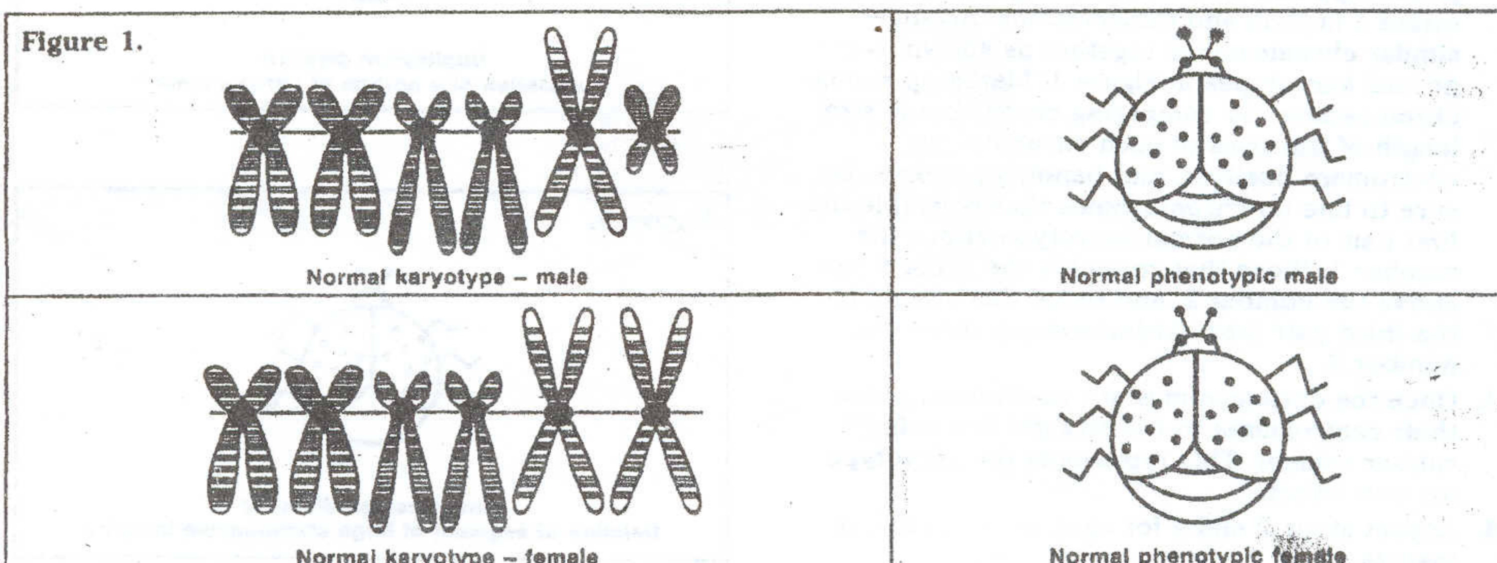


Figure 1. Normal karyotype – male; Normal phenotypic male; Normal karyotype – female; Normal phenotypic female

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.

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

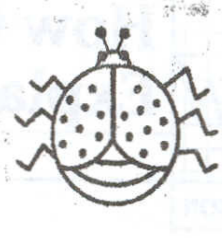
Clear wing disorder, as shown in Figure 3, appears to result from trisomy of the chromosomes of the second pair. The extra chromosome of the second pair produces sterile insects that lack coloring in their wings. Since sterility always results, the clear wing disorder is not passed on to progeny.

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.

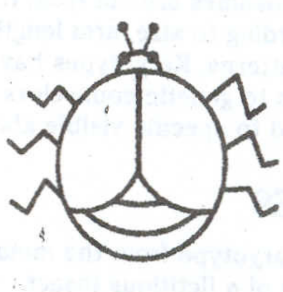
1. Obtain copies of the metaphase chromosomes of six insects from your teacher.
2. Write a **hypothesis** to describe how karyotype analysis can be used to explain the presence of a genetic disorder. Write your hypothesis in the space provided.
3. Cut out the chromosomes for insect 1 from the photocopy 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.
4. Once the chromosomes are positioned, paste their centromeres to the straight line using rubber cement. This represents the karyotype for one insect.
5. Repeat steps 3 and 4 for each of the fictitious insects.

Figure 2.



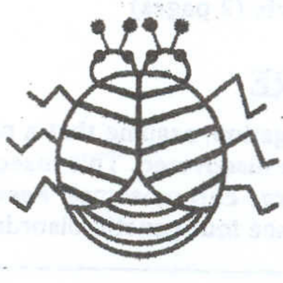
Size reduction disorder
Monosomy of chromosome 3

Figure 3.



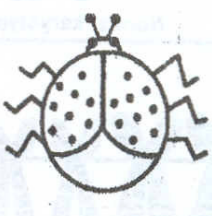
Clear wing disorder
Trisomy of chromosome 2

Figure 4.



Duplication disorder
Duplication of a portion of chromosome 1

Figure 5.



Unsegmented disorder
Deletion of segment of large chromosome in pair 3

6. Compare your karyotypes with the karyotypes of the normal insects and with the descriptions of the genetic disorders.
7. Complete the Analysis for this Investigation.

HYPOTHESIS

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DATA AND OBSERVATIONS

Insect 1

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1 2 3

Insect 2

____

1 2 3

Insect 3

____

1 2 3

Insect 4

____

1 2 3

Insect 5

____

1 2 3

Insect 6

____

1 2 3

ANALYSIS

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

	Sex	Genetic disorder	Chromosome error
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? ________________

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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? ________________________________

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

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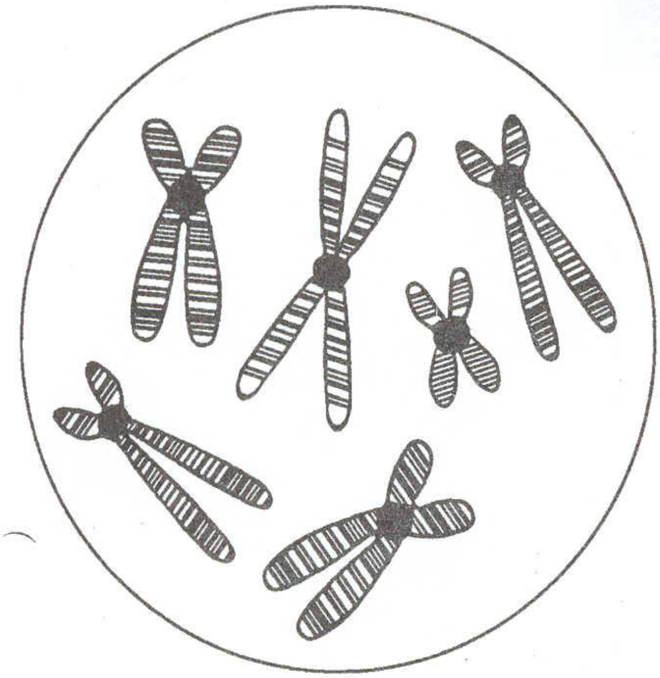
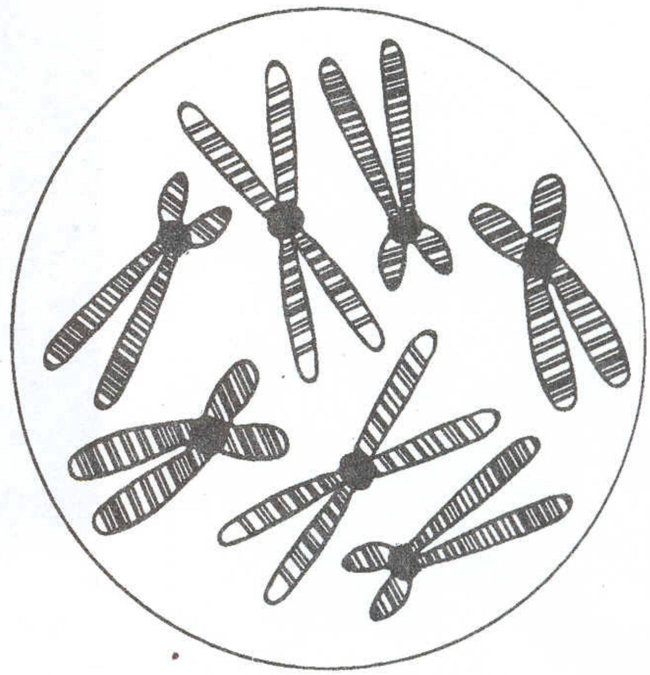
HOW TO USE THE STUDENT MODELS

Paper models for activities 15–2, 16–1, and 21–1 can be found in this teacher edition. In order for students to complete these activities, you must photocopy or mimeograph copies of the models for your students. Note that each team will need four copies of page T29 for Exploration 16–1, "DNA Sequencing."

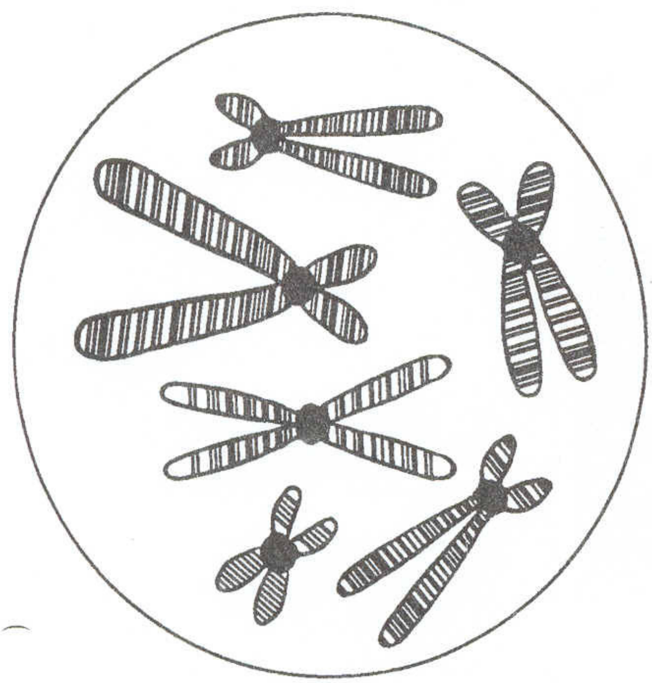
METAPHASE CHROMOSOMES FOR INVESTIGATION 15–2, "HOW CAN KARYOTYPE ANALYSIS EXPLAIN GENETIC DISORDERS?"



Metaphase chromosomes Insect 1

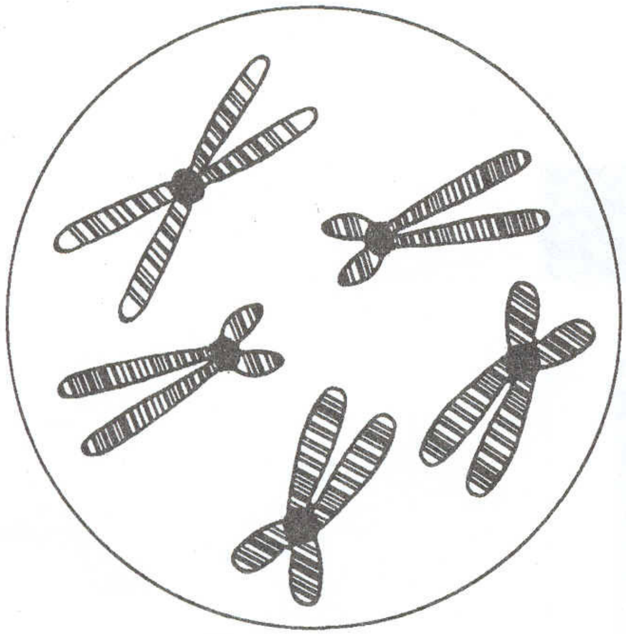


Metaphase chromosomes Insect 2

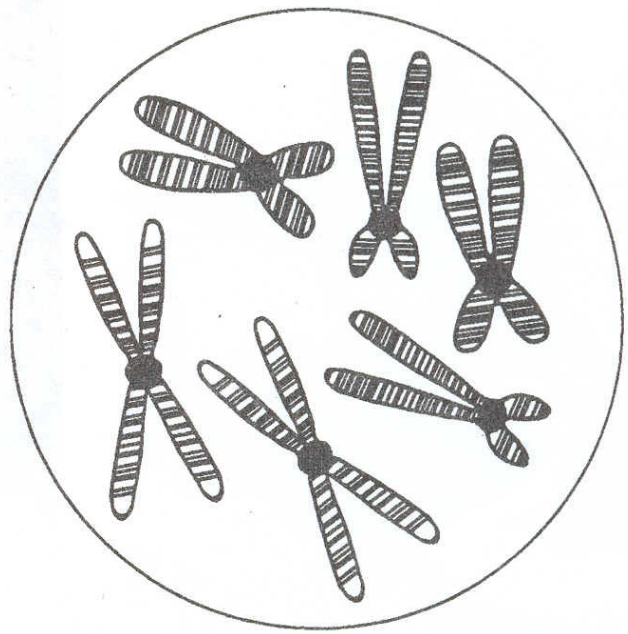


Metaphase chromosomes Insect 3

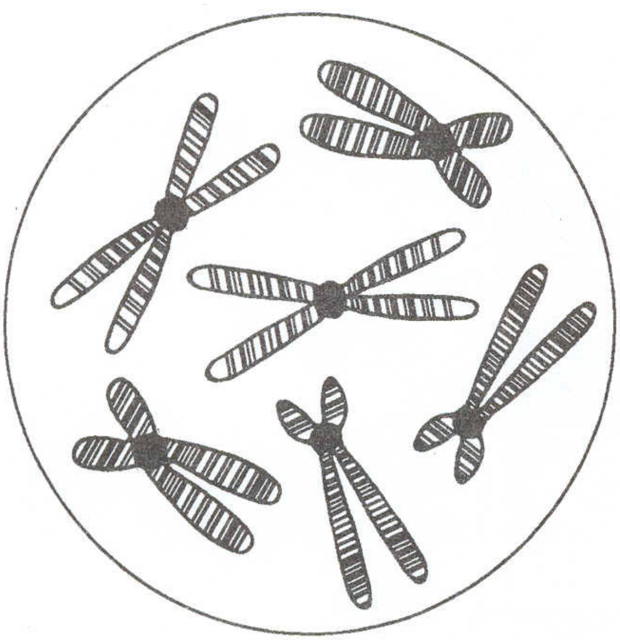
METAPHASE CHROMOSOMES FOR INVESTIGATION 15–2,
"HOW CAN KARYOTYPE ANALYSIS EXPLAIN GENETIC DISORDERS?"



Metaphase chromosomes Insect 4



Metaphase chromosomes Insect 5



Metaphase chromosomes Insect 6