

Karyotype Analysis

Background:

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 are stained, and then photographed through the microscope and the photograph is enlarged. The chromosomes are cut from the photograph and arranged in pairs according to size and centromere position. Karyotypes have become of increasing importance to genetic counselors as disorders and diseases have been traced to specific visible abnormalities of the chromosomes.

Purpose:

Researchers have been studying a population of rare insects, *Insectus highschoolus*, found only on the Inderkum High School campus. The researchers have observed various forms of the insects and have concluded that many of these forms are the result of genetic disorders.

One researcher found a sample of six partially digested individuals in the stomach of a seagull (that was found dead). Because these insects were partially digested, the appearance of the individuals could not be determined, although most of the bodies remained. Undigested cell samples of each individual were taken to determine the karyotypes.

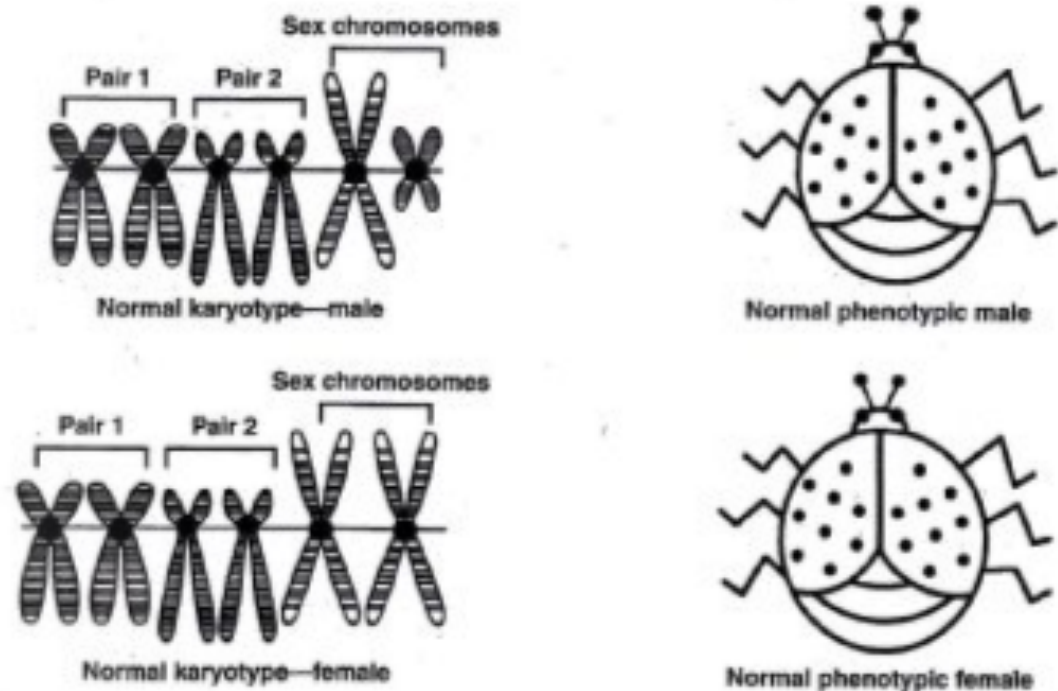
You will prepare the karyotypes of the insects and analyze them for chromosomal abnormalities. Furthermore, you will identify the disorders based on your analysis and try to figure out how a karyotype analysis can be used to explain the presence of a genetic disorder.

Procedures

1. Study the karyotypes and phenotypes (physical characteristics) of a normal male and female insect.

Note: the normal male and female insect's third pair of chromosomes are sex chromosomes. These chromosomes are similar to that of humans; the male has one small and large sex chromosome (XY) and the female has two large sex chromosomes (XX) (Figure 1).

Figure 1



2. Study the observed genetic disorders that the insects have:
 - a. **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 (Figure 2).
 - b. **Clear-wing disorder** 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 (Figure 3).
 - c. **Duplication disorder appears when** a portion of a chromosome in the first pair has been duplicated. This disorder results in two heads, a banding pattern on the wings, and additional body segments (Figure 4).
 - d. **Unsegmented disorder** appears when a small segment of the sex chromosome is deleted. The results are a loss of body segmentation and reduction of body size. (Figure 5)
3. Obtain copies of the metaphase chromosomes of six insects.
4. Cut out the chromosomes for insect 1 from the photocopy and place them along the line for insect 1 in Data and Observations.
5. Arrange similar chromosomes together as shown in the normal karyotypes in Figure 1.
6. Match up similar chromosomes by comparing chromosome size, length of the arms of each chromosome, centromere position and banding patterns.
7. Be sure to line up chromosomes that resemble the first pair of the normal karyotype above number 1, those that resemble the second pair above number 2 and those that resemble the third pair (sex chromosomes) above number 3.
8. Once the chromosomes are positioned, paste their centromeres to the straight line. This represents the karyotype for one insect.
9. Repeat the above steps for each of the remaining insects.
10. Compare your karyotypes with the karyotypes of the normal insects and with the descriptions of the genetic disorders.
11. Complete the Analysis Questions.

Figure 2



Size reduction disorder
Monosomy of pair 3

Figure 3



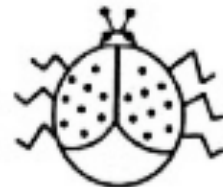
Clear wing disorder
Trisomy of pair 2

Figure 4



Duplication disorder
Duplication of a portion of a chromosome in pair 1

Figure 5



Unsegmented disorder
Deletion of segment of the large chromosome in pair 3

Data and Observations:

Insect 1

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 Questions:

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

| | Sex | Genetic Disorder | Chromosomal Error |
|-----------------|------------|-------------------------|--------------------------|
| Insect 1 | | | |
| Insect 2 | | | |
| Insect 3 | | | |
| Insect 4 | | | |
| Insect 5 | | | |
| Insect 6 | | | |

2. Which type of chromosome abnormality was the most difficult to detect through the karyotype? The easiest?

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

4. What kind of information would be required if a karyotype analysis were used to detect the genetic disorder of a real organism?

5. How can karyotype analysis be used to explain the presence of a genetic disorder?



Metaphase chromosomes Insect 1



Metaphase chromosomes Insect 4



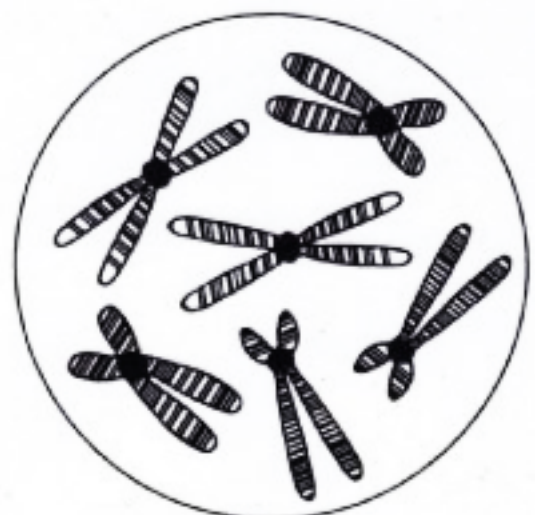
Metaphase chromosomes Insect 2



Metaphase chromosomes Insect 5



Metaphase chromosomes Insect 3



Metaphase chromosomes Insect 6