INVESTIGATION
1-A

GUIDED INQUIRY

Using Karyotypes to Diagnose Genetic Disorders

Skills and Strategies

- Planning and Conducting
- Processing and Analyzing Data
- Evaluating
- Communicating

Safety

- Handle scissors with care.

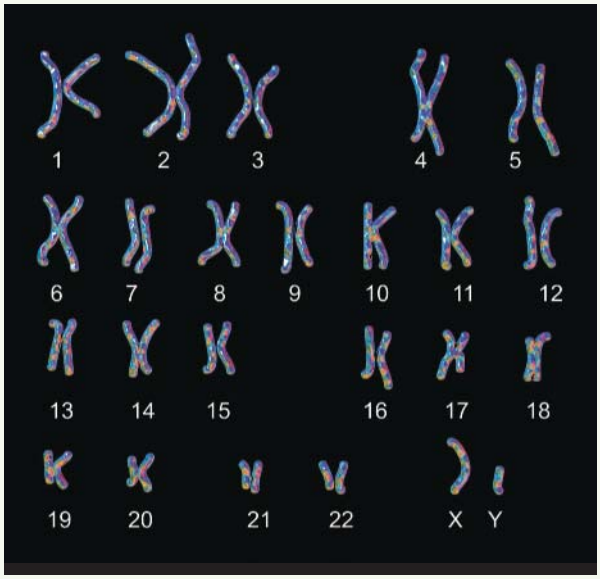
What You Need

- model chromosome image
- scissors
- tape or glue stick
- paper

In this activity, you will model and analyze a karyotype. A karyotype can be prepared before or after a baby is born to look for abnormalities in the number or structure of chromosomes within the cells. To prepare a karyotype, scientists take a tissue sample and then grow it in a culture dish in the lab. A chemical is added to the sample to stop cells during mitosis. Cells from this tissue are then placed on a microscope slide, stained, and treated with a chemical that makes the cells burst open. The stained chromosomes are then photographed through a microscope.

Question

What information can you infer from a karyotype?



To make a karyotype, the chromosomes are paired, placed in order of decreasing size, and numbered. **Identifying: Is this the karyotype of a genetic male or female? Explain.**

Procedure

1. Your teacher will supply you with a model image of a person's chromosomes.
   a) Cut out the chromosomes, and examine them closely to help you decide how to match them up in pairs. Remember that the sex chromosomes are not an identical pair. The X chromosome is larger than the Y chromosome.
   b) Compare your pairings with those of other groups. Confirm your decisions before moving on to the next step.

2. Tape or glue the chromosomes to a piece of paper. Arrange and number the pairs from largest to smallest. Leave a few centimetres of space between each pair. Place the sex chromosomes last. Number each pair of chromosomes from 1 to 22. Then label the sex chromosomes.
3. Examine your karyotype. Record the number of autosomes and the number of sex chromosomes.
4. Your teacher will give you an additional chromosome. Try to match it with one of the pairs in your original karyotype. Tape it in place next to the correct pair.
5. Examine your new karyotype. Note how it differs from your original.

Analyze and Interpret

1. Was your first karyotype from a somatic cell (body cell) or a gamete (sex cell)? How can you tell?
2. Based on your first karyotype, how confident are you that it represents someone who has no genetic abnormalities? Explain your reasoning.
3. Diagnose the disorder indicated by your karyotype in Procedure step 4. Give evidence for your answer. (Trisomy 13 = Patau syndrome; Trisomy 18 = Edward syndrome; Trisomy 21 = Down syndrome)
4. Cri-du-chat syndrome is a genetic disorder that results from a deletion of genes on the short arm of a chromosome 5. Make a sketch to show how the fifth chromosome pair might look in the karyotype of a child who has cri-du-chat syndrome.
5. Could you use a karyotype to detect a mutation in a single gene? Explain why or why not.

Conclude and Communicate

6. Did you find it difficult to match up the chromosome pairs? What features were most helpful when you were trying to identify homologous chromosomes?
7. You worked with a model image of chromosomes. Why might it be more challenging to interpret a photo taken through a microscope? What do you think is the likelihood of error when scientists interpret a karyotype?

Extend Further

8. Research and describe the cause and symptoms of Williams syndrome. Can it be detected using a karyotype? Why or why not?