**Karyotyping Activity**

**I. Introduction**

This exercise is a simulation of human karyotyping using digital images of chromosomes from actual human genetic studies. You will be arranging chromosomes into a completed karyotype, and interpreting your findings just as if you were working in a genetic analysis program at a hospital or clinic. Karyotype analyses are performed over 400,000 times per year in the U.S. and Canada. Imagine that you were performing these analyses for real people, and that your conclusions would drastically affect their lives.

During mitosis, the 23 pairs of human chromosomes condense and are visible with a light microscope. A karyotype analysis usually involves blocking cells in mitosis and staining the condensed chromosomes with a dye. The dye stains regions of chromosomes that are rich in the base pairs Adenine (A) and Thymine (T) producing a dark band. A common misconception is that bands represent single genes, but in fact the thinnest bands contain over a million base pairs and potentially hundreds of genes. For example, the size of one small band is about equal to the entire genetic information for one bacterium.

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The analysis involves comparing chromosomes for their length, the placement of centromeres (areas where the two chromatids are joined), and the location and sizes of bands. You will electronically complete the karyotype for three individuals and look for abnormalities that could explain the phenotype.

This exercise is designed as an introduction to genetic studies on humans. Karyotyping is one of many techniques that allow us to look for several thousand possible genetic diseases in humans. You will evaluate 3 patients' case histories, complete their karyotypes, and diagnose any missing or extra chromosomes. Then you'll conduct research on the internet to find web sites that cover some aspect of human genetics.

**II. Procedure**  
  
 1. Go to following website :

<http://www.biology.arizona.edu/human_bio/activities/karyotyping/karyotyping2.html>

2. Enter the following information about **Patient A**.

Patient Identity = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

How Chromosomes Obtained = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

3. Click on Patient Histories and Complete **Patient A’s** Karyotype.

4. Using the chromosome illustrated to the left, click on the number in the karyotype that

matches. Continue until all chromosome pairs are matched up.

5. Choose where the extra chromosome belongs.

6. What notation would you use to characterize **Patient A’s** karyotype?

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Circle One : Male Female

7. Use the Diagnosis Table and determine what diagnosis you would give Patient A?

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

8. Repeat steps 3-7 using Patient B and Patient C.

**Patient B**

Patient Identity = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

How Chromosomes Obtained = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

What notation would you use to characterize **Patient B’s** karyotype?

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Circle One : Male Female

Determine what diagnosis you would give **Patient B**?

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Patient C**

Patient Identity = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

How Chromosomes Obtained = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

What notation would you use to characterize **Patient C’s** karyotype?

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Circle One : Male Female

Determine what diagnosis you would give **Patient C**?

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9. Examine the following karyotypes. Identify the genetic disorders.

**Genetic Disorder = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

**Genetic Disorder = \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**