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| **Introduction** |

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| 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.Bio12AP **Karyotyping Activity** Name: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_Access the activity online at: <http://www.biology.arizona.edu/human_bio/activities/karyotyping/karyotyping.html>  |

**G Banding**

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| 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 Giemsa 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.http://www.biology.arizona.edu/human_bio/activities/karyotyping/graphics/chromsmear.gif |



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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 G-bands. You will electronically complete the karyotype for three individuals and look for abnormalities that could explain the phenotype. |

**Your assignment**

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| 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.** **On a separate sheet of paper you should turn in a total of 7 answers (2 for each patient, 1 for the internet search).** |

**Interpreting the karyotype**

Lab technicians compile karyotypes and then use a specific notation to characterize the karyotype. This notation includes the total number of chromosomes, the sex chromosomes, and any extra or missing autosomal chromosomes. For example, **47, XY, +18** indicates that the patient has 47 chromosomes, is a male, and has an extra autosomal chromosome 18. **46, XX** is a female with a normal number of chromosomes. **47, XXY** is a patient with an extra sex chromosome.

**Making a diagnosis**

The next step is to either diagnose or rule out a chromosomal abnormality. In a patient with a normal number of chromosomes, each pair will have only two chromosomes. Having an extra or missing chromosome usually renders a fetus inviable. In cases where the fetus makes it to term, there are unique clinical features depending on which chromosome is affected. Listed below are some syndromes caused by an abnormal number of chromosomes.

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| **Diagnosis** | **Chromosomal Abnormality** |
| Normal # of chromosomes | patient's problems are due to something other than an abnormal number of chromosomes. |
| Klinefelter's Syndrome | one or more extra sex chromosomes (i.e., XXY) |
| Down's Syndrome | Trisomy 21, extra chromosome 21 |
| Trisomy 13 Syndrome | extra chromosome 13 |

**Record your Answers:**

**Patient A**

A1. What notation would you use to characterize patient A’s karyotype? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

A 2. What diagnosis would you give patient A? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Patient B**

B1. What notation would you use to characterize patient B’s karyotype? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

B 2. What diagnosis would you give patient B? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**Patient C**

C1.. What notation would you use to characterize patient A’s karyotype? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

C2. What diagnosis would you give patient A? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**WebResearch Q:** Find a strong website on a topic related to chromosomal inheritance/ alterations. Summarize the info you find there in 3-5 sentences and list the web address here: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_