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Honors Biology Mr. Collea

**Karyotype Activity**

*(*[*http://www.biology.arizona.edu/human\_bio/activities/karyotyping/karyotyping.html*](http://www.biology.arizona.edu/human_bio/activities/karyotyping/karyotyping.html)*)*

**Background Information**

Go to Collea’s Corner to access this on-line activity. 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.

**G Banding**

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.

 **Chromosome Smear**



 **Identifying features**



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.

Karyotypes done on an unborn fetus requires an additional step known as an **amniocentesis**. Amniocentesis is a medical procedure used in prenatal diagnosis of chromosomal abnormalities and also used for sex determination. During this procedure a small amount of amniotic fluid, which contains fetal tissue and cells, is sampled from the amniotic sac surrounding a developing fetus, and the fetal DNA is examined for genetic abnormalities. The most common reason to have an "amnio" is to determine whether a baby has certain genetic disorders or a chromosomal abnormality, such as Down syndrome.

**Your assignment**

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 that covers some aspect of the genetic disease diagnosed in patients’ A and B

**Patient Histories:**

**Patient A**

Patient A is the nearly-full-term fetus of a forty year old female. Chromosomes were obtained from fetal epithelial cells acquired through amniocentesis.

**Patient B**

Patient B is a 28 year old male who is trying to identify a cause for his infertility. Chromosomes were obtained from nucleated cells in the patient's blood.

**Patient C**

Patient C died shortly after birth, with a multitude of anomalies, including polydactyly and a cleft lip. Chromosomes were obtained from a tissue sample.

Name \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Honors Biology Mr. Collea

**Karyotype Activity**

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**Summary Sheet**

**1. Interpreting the Karyotype**

 What notation would you use to characterize Patient A's karyotype? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**2. Making a Diagnosis**

 What diagnosis would you give patient A?

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**3. Research the Disease (***Be sure to use the Chromosomal Disorders link on Collea’s Corner for help with this section.)*

 **(a)** Briefly describe the disease?

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 **(b)** How do people get this disease?

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 **(c)** List some of the symptoms of this disease.

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 **(d)** How is this disease treated?

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 **(e)** List three interesting facts about this disease.

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**1. Interpreting the Karyotype**

 What notation would you use to characterize Patient B's karyotype? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**2. Making a Diagnosis**

 What diagnosis would you give patient B?

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**3. Research the Disease (***Be sure to use the Chromosomal Disorders link on Collea’s Corner for help with this section.)*

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 **(b)** How do people get this disease?

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 **(c)** List some of the symptoms of this disease.

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 **(d)** How is this disease treated?

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 **(e)** List two interesting facts about this disease. .

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