

Name: _____ Date: _____

Student Exploration: Human Karyotyping

Vocabulary: autosome, chromosomal disorder, chromosome, genome, karyotype, sex chromosome

Prior Knowledge Question (Do this BEFORE using the Gizmo.)

A **chromosome** is a rod-shaped structure made of coils of DNA. Most human cells have 23 pairs of chromosomes.

1. Why do you think humans have two sets of 23 chromosomes? (Hint: Where did each set come from?) _____

2. How do you think different people's chromosomes would compare? _____

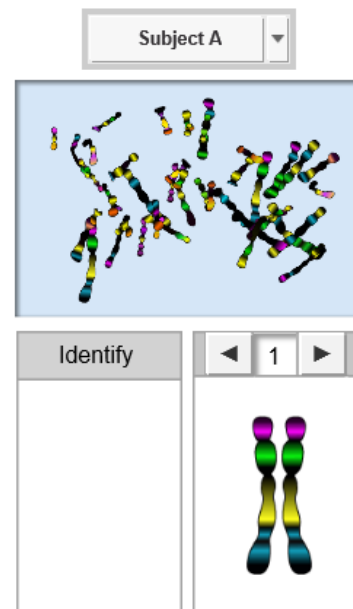
Gizmo Warm-up

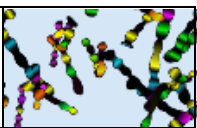
Scientists use **karyotypes** to study the chromosomes in a cell. A karyotype is a picture showing a cell's chromosomes grouped together in pairs.

In the *Human Karyotyping Gizmo™*, you will make karyotypes for five individuals. Take a look at the SIMULATION pane. Use the arrows to click through the numbered list of chromosomes at the bottom right of the pane.

1. How does the appearance of the chromosomes change as you move through the list? _____

2. Examine the chromosomes labeled **x** and **y**. How do these two chromosomes compare? _____



Activity A: Male and female karyotypes	<u>Get the Gizmo ready:</u> Click Reset.	
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Question: How are male karyotypes different from female karyotypes?

1. Compare: In the SIMULATION pane, make sure **Subject A** is selected. Click on and drag one of subject A's chromosomes to the area labeled **Identify**. Use the arrows to compare the chromosome you picked with chromosomes 1 through 22 and also with X and Y.

Which chromosome did you select? _____

2. Create: Drag the chromosome to the appropriate position on the KARYOTYPING pane. Then select another chromosome, identify it, and place it on the karyotype.

When you have identified and placed all of the chromosomes, click the **camera** (📷) to take a snapshot of the karyotype. Paste the snapshot into a document, and label it "Subject A."

3. Count: Chromosomes 1 through 22 are called **autosomes**. Examine the karyotype you have created. How many total autosomes do human cells have? _____

4. Draw conclusions: Look at chromosome pair 23. These chromosomes are known as **sex chromosomes** because they determine the sex of an individual. Females have two copies of the X chromosome. Males have one X chromosome and one Y chromosome.

Examine the karyotype. Is subject A a male or female? _____

How do you know? _____

Click the DIAGNOSIS tab to check your answer.

5. Analyze: Select **Subject B** from the SIMULATION pane. Complete subject B's karyotype. Take a snapshot of the completed karyotype, paste it into your document, and label it.

Examine the karyotype. Is Subject B a male or female? _____

How do you know? _____

Click the DIAGNOSIS tab to check your answer.

6. Think and discuss: On the SIMULATION pane, compare the X and Y chromosomes. Which chromosome do you think has more DNA? Explain. _____

Activity B: Chromosomal disorders	Get the Gizmo ready: Click Reset.	
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Question: How can you use a karyotype to diagnose a disease?

1. Compare: Select **Subject C** from the SIMULATION pane. Identify each of subject C's chromosomes, and place them on the KARYOTYPING pane. Once you have completed the karyotype, take a snapshot of it. Paste the snapshot into a document. Label it "Subject C."

How does subject C's karyotype differ from a normal karyotype?

2. Diagnose: A **chromosomal disorder** occurs when a person's cells do not have the correct number of chromosomes. The table below lists three common chromosomal disorders.

Disorder	Description	Subject	Symptoms
Down syndrome	Extra chromosome 21		
Klinefelter syndrome	Extra X in male (XXY)		
Turner syndrome	Single X in female (XO)		

Use the table to determine which disorder subject C has. Record your diagnosis in the third column of the table, and then click on the DIAGNOSIS tab to check your answer. Summarize the information on the DIAGNOSIS tab in the fourth column of the table.

3. Repeat: Complete the karyotypes for **Subject D** and **Subject E**. Determine which disorder each subject has, and use the information from the Gizmo's DIAGNOSIS tab to complete the table. Be sure to keep snapshots of both karyotypes.

(Activity B continued on next page)

Activity B (continued from previous page)

4. Generalize: Another chromosomal disorder, called Edward's syndrome, occurs when a person's cells have three copies of chromosome 18. People who have Edward's syndrome are severely mentally retarded and their skeletons are malformed. Most people with Edward's syndrome die in infancy.

Use the above information about Edward's syndrome and the descriptions of Down syndrome, Klinefelter syndrome, and Turner syndrome in the table on the previous page to compare these four different chromosomal disorders.

- A. Which type of chromosomal disorders seems to have the greatest affect on a person's health—disorders involving autosomes or sex chromosomes?

- B. Why do you think this might be the case? _____

5. Extend your thinking: Klinefelter syndrome only affects males, and Turner syndrome only affects females. Examine the karyotypes of the subjects you diagnosed with Klinefelter syndrome and Turner syndrome.

- A. How do you think sex is determined in a person with a chromosomal disorder involving the sex chromosomes? _____

- B. Individuals with a genetic disorder called trisomy X have three X chromosomes. (These individuals are normal and do not have any particular symptoms.)

What sex would a person with trisomy X be? _____

6. Summarize: The **genome** of an organism is its total genetic material. What aspects of the genome can and cannot be determined through karyotyping?

