



Name:

Thihini Nagendran

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Student Exploration: Human Karyotyping

Directions: Follow the instructions to go through the simulation. Respond to the questions and prompts in the orange boxes.

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?)

We have two sets of 23 chromosomes because we have one set from each parent.

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

I think different people would have different chromosomes.

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.

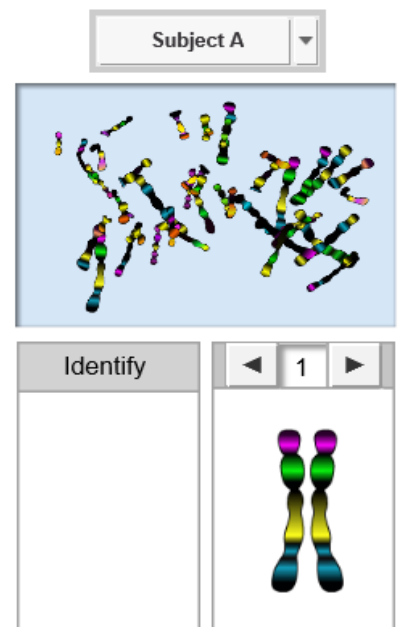
In the *Human Karyotyping* Gizmo, you will make karyotypes for five individuals. Take a look at the SIMULATION pane. Use the arrows to click through the numbered list of chromosomes at the bottom right of the pane.

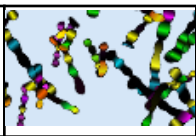
1. How does the appearance of the chromosomes change as you move through the list?

The appearance of the chromosomes change in size because they are getting smaller as you move through the list. The shape is also changing.

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

The x chromosome is much larger than the y chromosome.



Activity A: Male and female karyotypes	Get the Gizmo ready: <ul style="list-style-type: none"> Click Reset. 	
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Question: How are male karyotypes different from female karyotypes?

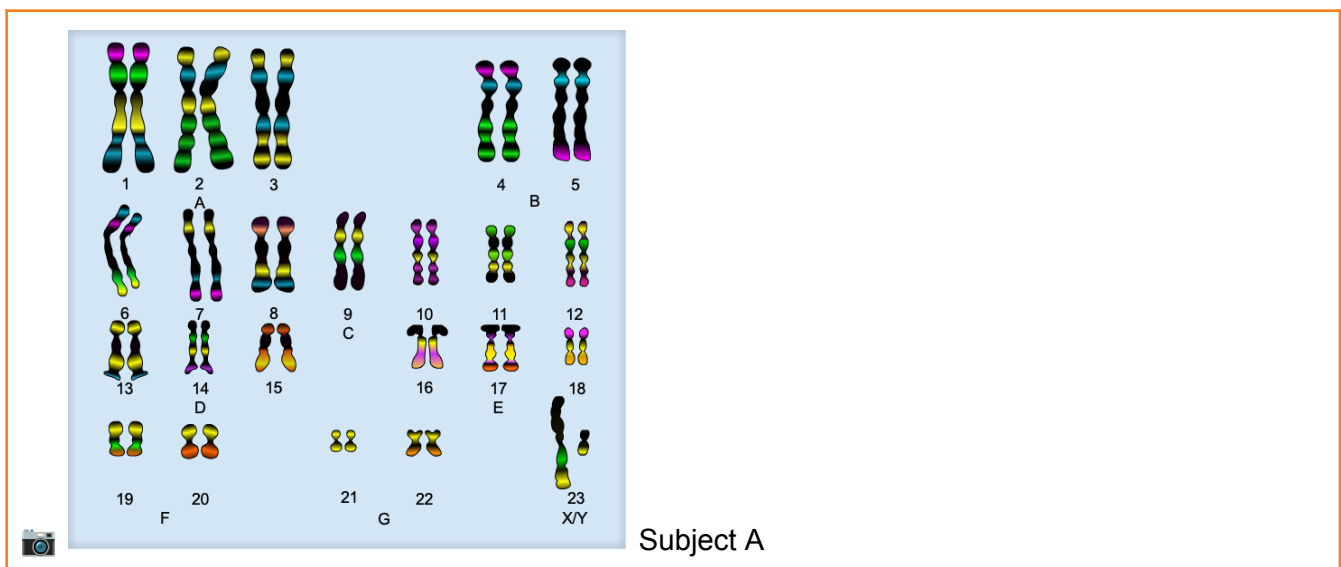
1. **Compare:** In the SIMULATION pane, make sure **Subject A** is selected. Click on and drag one of subject A's chromosomes to the area labeled **Identify**. Use the arrows to compare the chromosome you picked with chromosomes 1 through 22 and also with X and Y.

Which chromosome did you select?

I selected 8

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 below, 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?

22

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?

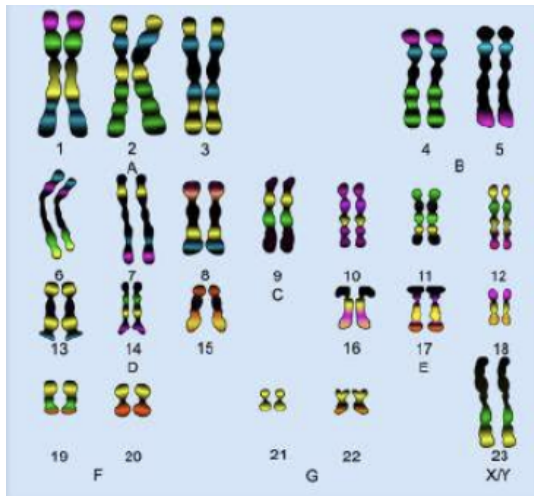
Male

How do you know?

Because the chromosome pair 23 has both X and Y chromosomes

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.



Subject B

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

Female


How do you know?

Because the chromosome pair 23 of subject B has two X chromosomes

Click the DIAGNOSIS tab to check your answer.

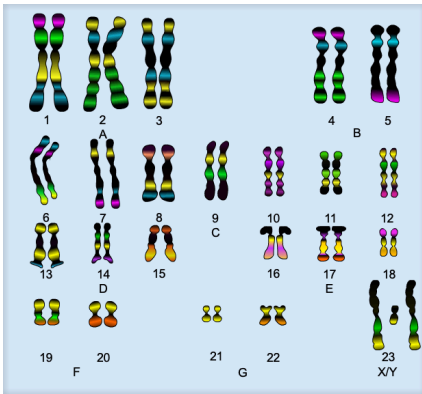
6. Think and discuss: On the SIMULATION pane, compare the X and Y chromosomes. Which chromosome do you think has more DNA? Explain.

I believe that the X chromosome has more DNA because it is larger in size

<p>Activity B:</p> <p>Chromosomal disorders</p>	<p><u>Get the Gizmo ready:</u></p> <ul style="list-style-type: none"> 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."



Subject C

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

The 23rd chromosome pair contains 3 chromosomes instead of the 2 that a normal karyotype has

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	E	Respiratory issues and small head. Vision and speech problems, hearing loss, and heart defects.
Klinefelter syndrome	Extra X in male (XXY)	C	Defect becomes apparent in puberty when secondary sexual characteristics fail to develop. Testicular changes occur that eventually result in infertility in the majority affected.
Turner syndrome	Single X in female (XO)	D	Incorrect development of ovaries and hormone production sex-hormones

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. 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 have severe intellectual disabilities 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?

Disorder involving autosomes because disorders like Edward's syndrome where there are three copies of chromosome 18 could cause people to die in infancy.

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

This is because genes in the autosomes can affect bodily functions and disrupting these genes and chromosomes could cause complications to vital organs or bodily functions.

4. 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?

Sex could be determined by the presence or absence of a Y chromosome because females only have X chromosomes while males have both X and Y.

- 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?

Female

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

Karyotyping can provide information about a person's sex and what chromosomal disorders they have but they cannot provide information on specific traits that are in a person's genes.