

# Karyotypes: Diagnosing Chromosome Disorders

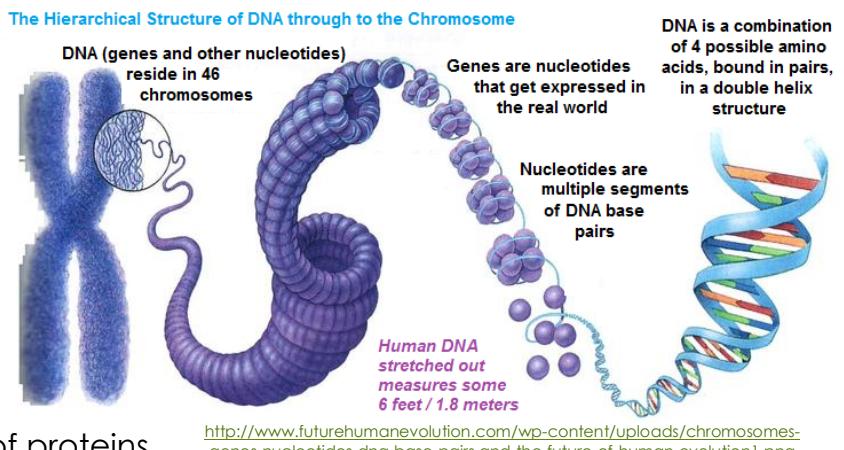
## HASPI Medical Biology Lab 17

### Background/Introduction



### DNA, Genes, and Chromosomes

All cells contain genetic information in the form of **deoxyribonucleic acid (DNA)** molecules. DNA contains the genetic instructions that form a species' characteristics. DNA is made up of four nucleotides: adenine, thymine, guanine, and cytosine. The arrangement, or order, of these nucleotides creates the directions for a specific protein. **Genes** are regions in the DNA that contain the instructions that code for the formation of proteins.



<http://www.futurehumanevolution.com/wp-content/uploads/chromosomes-genes-nucleotides-dna-base-pairs-and-the-future-of-human-evolution1.png>

Humans have more than 20,000 genes. All cells in an organism have the same genetic content, but the genes used (expressed) by the cell may be regulated in different ways. For example, the gene to create the actin and myosin proteins that are capable of contracting are expressed in muscle cells, but are dormant (not expressed) in skin cells. Not all DNA codes for a protein. Some segments of DNA are involved in regulatory or structural functions, and some have no as-yet known function.

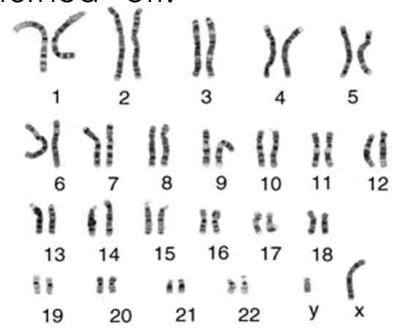
In each cell, DNA, and therefore genes, are wound into tight structures called **chromosomes**. Chromosomes are primarily formed during mitosis, or cell division, to prevent damage or loss of delicate DNA strands. Each chromosome consists of a single, very long DNA molecule. Humans have 23 pairs of chromosomes, 22 pairs of **autosomes**, and 1 pair of **sex chromosomes**. As their name implies, sex chromosomes contain the genes that primarily impact male and female characteristics.

### Gene Expression and Regulation

How does a cell know when to express a gene? Some genes are always "on," and are responsible for cell processes such as metabolism, repair, and protein synthesis. Other genes are regulated and have the ability to be turned "on" and "off." For example, the gene that codes for insulin is only turned "on," or expressed, by pancreatic cells when it receives a signal from the brain. The brain only sends the signal when it senses a high level of glucose in the blood. As soon as the glucose level in the blood drops, the brain sends a signal to the pancreatic cells to stop using the gene for insulin, and the gene is turned "off."

### Karyotypes

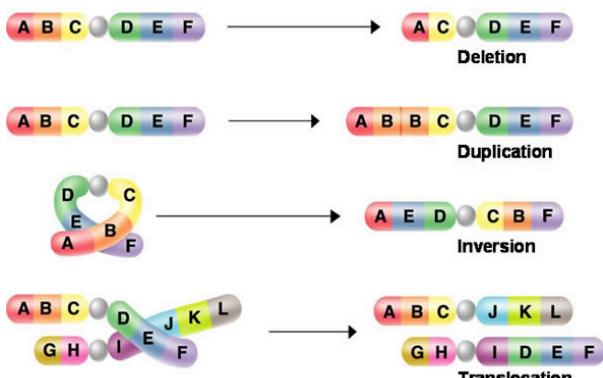
A genetic mutation that occurs when chromosomes do not separate correctly during cell division can be observed by looking at human DNA when it is in chromosome form. Observation can determine if there are any extra chromosomes, or missing or damaged chromosomes. This is done by removing the chromosomes from a cell, spreading them out, and making them visible since they are microscopic. A picture is then taken and a



<http://www.biotechnologyonline.gov.au/images/contentpages/karyotype.jpg>

genetic counselor orders the chromosomes into the 22 pairs of autosomes and 2 sex chromosomes. This ordering and pairing of chromosomes is called a **karyotype**. A normal karyotype is shown on the previous page. The dark or light areas on the chromosomes are areas where the DNA is more or less tightly coiled up. Over 500,000 karyotypes are performed in the U.S. every year, especially for genetic and reproductive medicine.

Collecting cells from an unborn fetus, or the amniotic fluid surrounding a fetus, for karyotyping is a common occurrence for expecting parents who wish to make sure no genetic abnormalities exist. The common process for obtaining these cells occurs through **amniocentesis**, in which a needle is inserted into the womb. There are risks of miscarriage or other damage to the fetus associated with the amniocentesis procedure, so it is usually not chosen to be performed unless there is a suspected possibility of genetic abnormalities.



[http://www.goldiesroom.org/Multimedia/Bio\\_Images/19%20Applied%20Genetics/05%20Chromosome%20Mutations.htm](http://www.goldiesroom.org/Multimedia/Bio_Images/19%20Applied%20Genetics/05%20Chromosome%20Mutations.htm).

## Chromosome Mutations

Chromosome mutations can occur when entire chromosomes are **duplicated** or **deleted**. They may also occur when sections of chromosomes are **duplicated**, **deleted**, **inverted**, **inserted** into a different chromosome, or **translocated** with another chromosome. The image to the left diagrams these occurrences. As you can see, chromosomal mutations change the size and/or organization of the light/dark bands on a chromosome, making it easy to see that a mutation has occurred when completing a karyotype.

## Review Questions – answer questions on a separate sheet of paper

- What are the four nucleotides that make up DNA?
- Within the human body, where is DNA located?
- What are genes? What do genes contain instructions to create?
- Do all genes code for proteins? Explain your answer.
- What are chromosomes?
- How many chromosomes do humans have?
- Where did you get your chromosomes?
- What is the difference between autosomes and sex chromosomes?
- How is gene expression regulated?
- What is a karyotype?
- What is the purpose of performing a karyotype?
- Hypothesize how the chromosomes of the parents would look compared to the chromosomes of their child.
- Make a table that describes and depicts (draw a picture) the different types of chromosome mutations.

Name(s):

Period:

Date:

## Part A. Karyotypes: Diagnosing Chromosome Disorders

### HASPI Medical Biology Lab 17a

#### Scenario



You work as part of the genetic counseling team at Genetic Horizons, Inc. Your team is responsible for creating and organizing karyotypes of expecting couples who want to make sure their child will not have a genetic mutation, and therefore a genetic disease. One of your teammates has already collected and made the fetus's chromosomes visible.

#### Materials

Patient Karyotype sheet

Scissors

Tape/glue

#### Procedure/Directions

Your lab team will be given tasks, or directions, to perform on the left. Record your questions, observations, or required response to each task on the right.

	Task	Response
1	Obtain a "Patient Karyotype" sheet. You can choose from three patients.	<b>Normal Human Karyotype</b> 
2	Write the Patient ID number on the "Patient Education" form and the "Karyotype Results" sheet on the next two pages.	
3	It is important for the parents to understand DNA, chromosomes, and the procedure. Complete the patient education information.	
4	Cut out and organize the chromosomes from the patient sheet into a karyotype on the "Karyotype Results" sheet. Use the normal human karyotype at right for comparison.	
5	Identify any chromosomal abnormalities that may exist. Use the "Genetic Disorders Guide" below to determine what disease might result, and complete the analysis.	

Name(s):

Period:

Date:

## Patient Medical Sheet

Patient ID: \_\_\_\_\_

### Patient Education

In order to educate your patients, please write an overview of the following so they can have a better understanding of how you produced the results. Remember, the patients are not scientists so try to explain what is happening in as simple terms as possible, and also define any science vocabulary you may use. Do not copy the Background information directly, but put these concepts into your own words. You should write a minimum of 3 complete sentences for each item below.

a. DNA and chromosomes are:

b. In terms of chromosomes, humans have:

c. Problems, called mutations, can sometimes occur in chromosomes. Three examples of mutations and the disorders they may cause are:

d. We will be performing an amniocentesis. The purpose, procedure, and risks are:

e. From the cells we collect, we will document your results in the form of a karyotype. A karyotype can be defined as, and will allow us to:

Name(s):

Period:

Date:

## Karyotype Results

Patient ID: \_\_\_\_\_

Tape/paste the patient chromosomes here. Highlight or circle any chromosomal abnormalities found in the patient's karyotype.

1

2

3

4

5

6

7

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9

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11

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14

15

16

17

18

19

20

21

22

XX or XY

Name(s):

Period:

Date:

## Genetic Disorders Guide\*

Genetic Disorder	Chromosome Abnormality	Effects
<b>Cri du Chat</b>	Deletion of upper portion of chromosome #5	Frequency is 1 in 20,000 births. The name is French, meaning "cry of the cat," and represents the sound that infants born with this disorder make. Individuals with this disorder have symptoms that include severe mental retardation, widely spread eyes, low chin set, smaller than average head size, small mouth, low birth weight, hyperactivity, scoliosis, and on occasion serious organ defects.
<b>Edward's Syndrome</b>	Extra chromosome #18	Frequency is 1 in 8,000 births. Individuals will have severe mental retardation, receding jaw, low set ears, clenched fists, and heart abnormalities. Mortality rate is very high with most individuals not surviving past 2 months of age.
<b>Klinefelter's Syndrome</b>	Additional X sex chromosome	Affects males only and frequency is 1 in 1,000 male births. Symptoms do not become evident until puberty when sex characteristics begin to develop. Males with this disorder will be taller than average, lanky, have incomplete testicle development (making them sterile), and develop female physical characteristics (breast development, lack of body hair, etc.).
<b>Turner's Syndrome</b>	Only one sex chromosome (normally X)	Affects females only and frequency is 1 in every 2,000 female births. Individuals with this disorder have symptoms that include short stature, joint swelling, low hairline, low set ears, wide chest, neck webbing, and ovary dysfunction (sterile). They may also suffer from congenital heart disease, hypothyroidism, diabetes, vision loss, and hearing loss.
<b>Down Syndrome</b>	Extra chromosome #21	Frequency is 1 in 700 births and becomes more common as the age of the mother increases. Individuals with this disorder have symptoms ranging from different levels of mental retardation, short stature, increased susceptibility to infections, heart defects, flat noses, down-sloping eyes, thick necks and tongues, a single long crease along the palm of the hand, underdeveloped ears, and simple fingerprints. As a result of the heart defects, infant mortality is high and individuals with Down syndrome rarely live beyond 40 years old.

\*Note: This is an incomplete list, and many more types of genetic disorders occur.

Name(s):

Period:

Date:

## Analysis & Interpretation

### Analysis Questions – answer questions on a separate sheet of paper

1. How many autosomes does the patient have?
2. How many sex chromosomes does the patient have?
3. What is the sex of this patient?
4. What chromosomal abnormalities were found? Use the “Genetic Disorders Guide” to determine what disorder will result.
5. What is the frequency of this disorder?
6. What are the physical features of people with this disorder?
7. What are the symptoms of this disorder?
8. What are the treatment options for this disorder?
9. What recommendations would you give to the parents in this situation?
10. Asking questions is an important part of the scientific process. Craft at least 3 questions that could be answered using karyotyping.

## Part B. Genetic Interactions in Human Traits

### HASPI Medical Biology Lab 17

#### Scenario



Human gene interactions can be very complex, especially in traits such as skin color. On the other hand, some genetic traits are fairly simple and are expressed through a dominant/recessive interaction. In this activity, you will perform your own survey of human genetic traits of your choice to determine whether specific phenotypes are dominant, recessive, or whether there are more complex interactions occurring.

#### Directions

- 1** Choose three of the human traits from Table 1. Each trait listed has a description of two alleles that contribute to the gene that controls the phenotypes for each trait.
- 2** The goal of this activity is to determine whether each trait is dominant/recessive, and if it is both, which phenotypes are dominant and which phenotypes are recessive. There may also be traits that display co-dominance, multiple alleles, or sex-linkage.
- 3** Create an anonymous survey that includes whether each individual surveyed is male or female, and which phenotypes are displayed for the three traits you have chosen.
- 4** Survey a minimum of 100 people.
- 5** Organize your results for each trait into a table, graph, or chart. Include the number of each phenotype and separate the results for males and females.
- 6** Answer the analysis questions based on your survey results.

**Table 1. Human Traits**

Widow's peak or straight hair line	Dimples or no dimples	Detached or attached earlobes
Widow's Peak Hairline	Dimples	Detached Earlobes
Straight Hairline	No Dimples	Attached Earlobes
Tongue roll or no tongue roll	Cleft chin or smooth chin	Freckles or no freckles
Can Roll Tongue	Cleft Chin	Freckles
Can't Roll Tongue	Smooth Chin	No Freckles
Curly hair or straight hair	Left-handed or right-handed	Colorblindness or normal vision
Naturally Curly Hair	Left-handed	Normal Vision
Naturally Straight Hair	Right-handed	Simulated Colorblind Vision

<http://learn.genetics.utah.edu/content/begin/traits/>

## Analysis & Interpretation

### Analysis Questions – answer questions on a separate sheet of paper

1. For each of the three human traits you chose, identify which phenotype is dominant and which is recessive. Explain your answer using your results.
2. Was there any difference between the male and female population for each trait? If yes, discuss the differences and if no, discuss why you think there was none.
3. Did any of your results not demonstrate a dominant/recessive interaction? Explain your answer.
4. Most scientists agree that an "increase in the number of tests performed, increases the validity of experimental results." How do you think this could be applied to this activity?
5. Sometimes scientific experiments create more questions than answers. Create one question that arose from your results and explain how you could answer that question.
6. Explain heredity and how traits are inherited.
7. Explain how the three human traits you chose relate to genes and chromosomes.
8. It is important to understand that some genes are expressed only during development and then "turn off" and are never used again during an individual's life cycle. Looking at Table 1, hypothesize which of these traits' genes were only active during development, and which traits' genes would still be active. Explain your answer for each trait.

### Connections & Applications

Your instructor may assign or allow you to choose any of the following activities. As per NGSS/CCSS, these extensions allow students to explore outside activities recommended by the standards.

1. **ASKING QUESTIONS:** Go to the following website:

<http://learn.genetics.utah.edu/content/begin/tour/>. This is an interactive animation that explains the relationship and function of DNA, genes, and chromosomes.

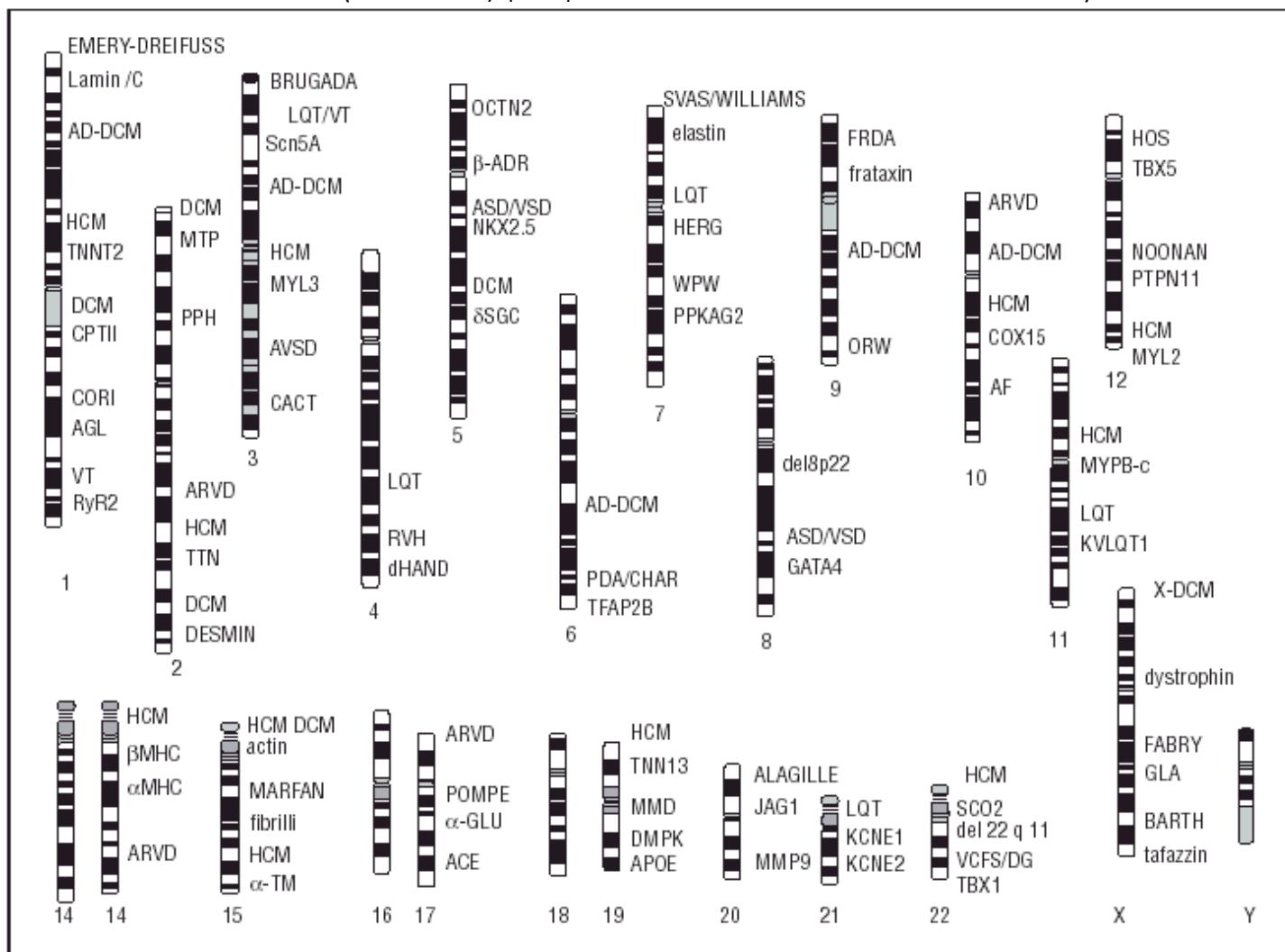
Complete the "Tour of the Basics." Create a minimum of three USEFUL questions and answers based on each of these topics:

- a. "What is DNA?"
- b. "What is a Gene?"
- c. "What is a Chromosome?"
- d. "What is a Protein?"
- e. "What is Heredity?"
- f. "What is a Trait?"

2. **CREATE A MODEL:** Using any materials that you have available, create a model of DNA, genes, and chromosomes. Your model must be labeled and show the differences AND relationship between DNA, genes, and chromosomes. Before creating your model, discuss your idea and the materials you will be using with your instructor to ensure they are safe and appropriate.

3. **GENES ON A CHROMOSOME:** There has been a great deal of energy in the scientific community placed into mapping the genes located on chromosomes, and consequently the location of mutations that can cause genetic diseases. The Human Genome Project was completed in April of 2003 and provided researchers worldwide with the ability to expand their genetic research. Much of this research is targeted on genetic diseases, and the possibility of producing cures for these currently non-curable disorders. The following diagram is a summary of genetic cardiovascular diseases. Every gene is identified with a gene code to make it easier for researchers to identify it. Choose and research 5 of the gene codes on the diagram below. Create a table that includes the following information for each disease:

- The gene code and the name of the disease it represents
- The chromosome (number) on which it is located
- A description of the disease
- Prevalence (how many people have or are born with this disease)



<http://www.revespcardiol.org/imatges/255/255v57n04/grande/255v57n04-13060505tab01.gif>

## Resources & References

- Genetic Science Learning Center. 2011. Using Karyotypes to Predict Genetic Disorders. Learn Genetics.