UNIVERSITY OF MICHIGAN SCHOOLS OF EDUCATION AND PUBLIC HEALTH

How SIMILAR or DIFFERENT are we?

Student Reader

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How SIMILAR or DIFFERENT are we?
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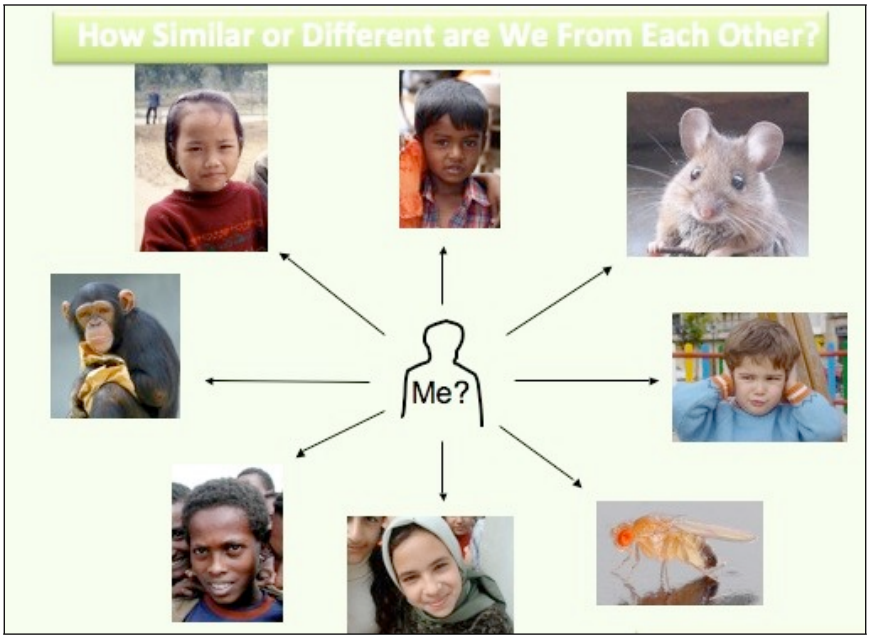
How SIMILAR or DIFFERENT Are We?



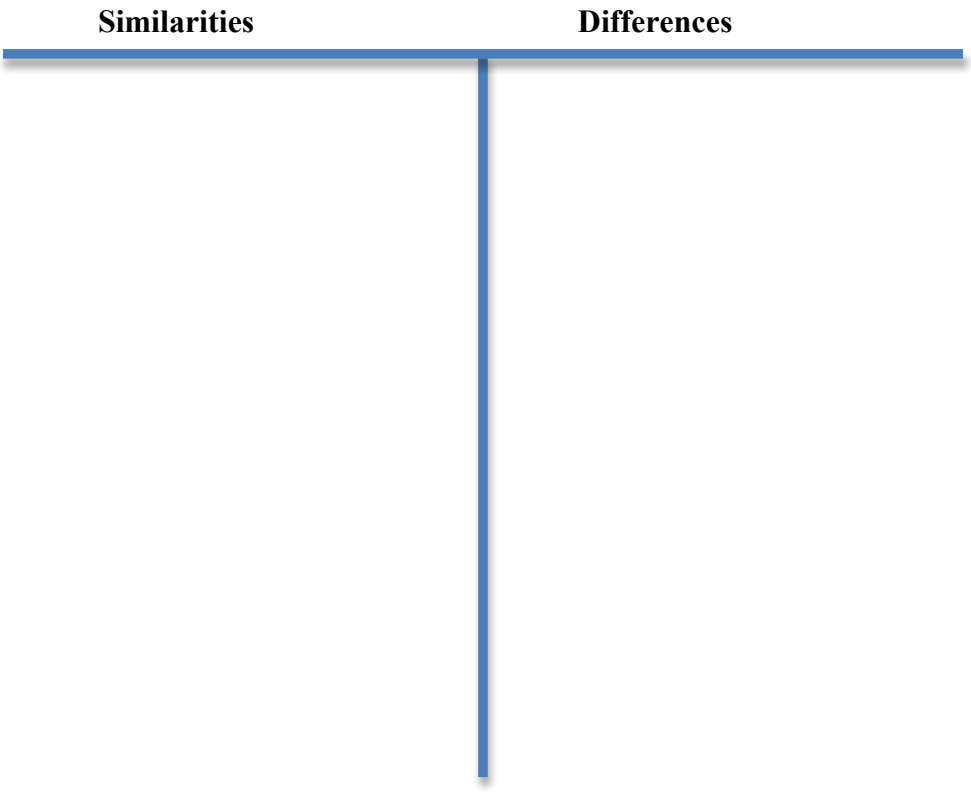
Activity 1.1

How Similar or Different Are We?

PURPOSE
In this activity you will begin to answer the question **How similar or different are we?** through careful observations of others. You will make comparisons between yourself and other people as well as other species in order to write scientific explanations to begin to answer the question.

1. Take a moment and look around at the people near you. On a scale from 1 to 10, 1 being completely different, 10 being identical, how similar do you think you are to the other members of your group? _____

2. Do you notice a difference in hair color? Height? Skin color? Eye color? Eyesight? What about health conditions, like Asthma? What about the internal characteristics, like heart and lungs? List the physical features that are similar or different.



Explanation 1- How similar am I to others?

Look back at your answer to number 1. Your answer to the question, **How similar am I to others?** was actually a claim. A **claim** is a statement that answers the original question. Scientists explain things by making claims based on evidence from the data they collect. You made observations about your classmates that can be used as data. If good data is not provided in the explanation, then there is no reason to believe that the claim is a good one.

Do you still agree with your claim based on the observations you made in your chart? Data from observations or an experiment become **evidence** when you use it to support a claim you have made. The better the evidence you have, the stronger your claim will be. If the evidence does not support the claim you may need to change your claim.

Scientific explanations have three parts **claim**, **evidence** and **reasoning**. **Reasoning** tells why the data count as evidence to support the claim. Reasoning includes the important scientific principles. These principles help to build an argument to convince people that the evidence given is strong enough to support the claim.

Anyone can make a claim, but a good scientific explanation includes the evidence and reasoning that show the claim to be true. In this unit, when you write an explanation, remember to always include a claim, two pieces of evidence, and reasoning in your paragraph. Do you have enough evidence to support your claim? Now, practice writing an explanation to answer the question "How biologically similar am I to others?"

- Together with your class, make a new **claim** based on the evidence from your chart.
- Identify the **evidence** that supports your claim from your observations.
- Give a **reason** why that evidence supports your claim based on scientific principles.

3. What is your claim? (Rewrite your statement from question #1 based on your observations)

4. What are two pieces of evidence to support your claim? (Use your chart to identify evidence)

5. What reasoning (scientific principles) can you use to show why your evidence supports your claim? (*Scientific principle could be related to biological features. See class chart.*)

Explanation 2- How similar or different are people around the world?
Consider the images of humans of different races and ethnic backgrounds. List the biological features that are similar or different.

Similarities	Differences

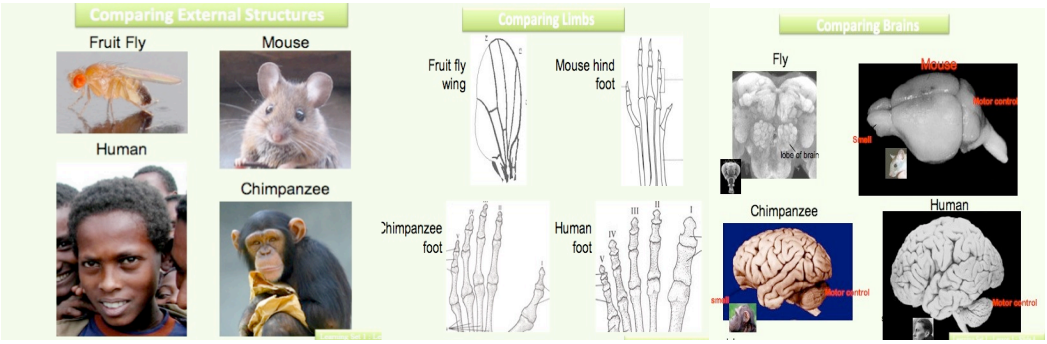
Look at the slides. How similar do you think humans from two different parts of the world are? Work with your group to write a scientific explanation using **claim, evidence and reasoning** to answer this question. Share your explanation with the class.

6. What is your claim? (Write your statement based on your observations)

7. What are 2 pieces of evidence to support your claim? (Use your chart to identify evidence)

8. What reasoning (scientific principles) can you use to show why your evidence supports your claim? (*Scientific principle could be related to biological features. See class chart.*)

Explanation 3- How similar or different are we from other species?



Look at the slides 2-4. Consider the images of humans and images of other species such as a fly, a mouse, and a chimpanzee. List the physical features that are similar or different in humans and other species.

Similarities	**Differences**

How similar do you think humans are from other species? Write your own scientific explanation using **claim, evidence and reasoning** to answer this question. Share your explanation with the class.

9. What is your claim? (Write your statement based on your observations)

10. What are 2 pieces of evidence to support your claim? (Use your chart to identify evidence)

11. What reasoning (scientific principles) can you use to show why your evidence supports your claim? (*Scientific principle could be related to biological features. See class chart.*)

Reading 1.1

Before You Read...

Definition of Biological: Any features relating to the biology of an organism. This includes the cells or organs that make up an organism or the biological molecules that are found in an organism. Some features of a person are not included when thinking of something biological, such as clothing.

Looking back at the charts that you made, you can see that humans share some ***biological*** similarities. Why do you think humans share so many biological features?

Apply

In class you had a discussion about some of the biological characteristics that make you similar and different from the other people in your class. The following reading talks about two girls whose mothers solved the mystery of why they had so many biological similarities.

Key words: DNA, fraternal twins.

DNA helps families find adopted kids' siblings

By LIZ DOUP
Knight Ridder Newspapers
http://www.dfw.com/mld/timesleader/living/14219929.htm?source=rss&channel=timesleader_living

FORT LAUDERDALE, Fla. — The eyes, the same. Hairline, the same. Nose, ditto.

When Eileen Surrey stared at the baby's picture on the Internet six years ago, she was stunned. "I thought I was looking at a picture of my child," says Surrey, of Boynton Beach, Fla.

Now, if a DNA test is reliable, Surrey was on to something. Her daughter, Renee, 8, has a sister — near Philadelphia. And their moms think the girls, adopted five months apart from the same Chinese orphanage, are fraternal twins.

DNA is genetic material inherited from parents

"I knew instantly there was a connection," Surrey says. So how do sisters — born halfway around the world — become separated and find each other again on American soil?

Their story starts in 1999 when Surrey answered a question posted on an adoption Web site by Andrea Ettingoff. Ettingoff wanted help with 16-month-old Annie's eating problem. After exchanging baby photos, Surrey and Ettingoff were struck by their daughters' physical similarities. Five years passed with e-mails and occasional face-to-face visits strengthening their convictions. These girls are related, they thought.

Ettingoff, clinical director for the Children's Crisis Treatment Center in Philadelphia, wanted more proof. She pushed for DNA testing. "I didn't need the DNA," says Surrey, reluctant to test at first. "They're sisters. I already knew in my heart."

But in December, when the girls were 7, the moms swabbed inside their daughters' cheeks for a DNA analysis. In January the results arrived: The girls are sisters, within 82.9 percent likelihood. Fraternal twins, the moms believe, because the girls are developing in similar ways. And both love the notion they have each other. "She's my sister," Renee says, tossing her long dark hair. "I wanted a sister. It's great!"

The popularity of DNA testing for adoptees has grown as their numbers have mushroomed. In the past decade, Chinese adoptions have more than tripled — nearly 8,000 last year. As a result, more parents are looking for a connection. Atlanta-based Kinsearch Registry is a non-profit DNA database started 18 months ago to help connect siblings separated by international adoptions.

1. The newspaper article says that the sisters recognized each other because they were developing in similar ways. What do you think that means?

2. In the article, the sisters were able to find out that they were related by using DNA. What do you think DNA is?

3. In the article it said that the DNA testing showed that there was an 82.9% likelihood that they were sisters. What do you think this means? How do you think DNA testing helped them figure it out?

4. How do you think the information in this article can help us answer the question: **How similar or different are we?**

Activity 1.2

Driving Question Board-- How Similar or Different are We?

PURPOSE
This unit starts with a driving question (DQ) **"How similar or different are we?"** Did the homework reading about DNA testing help you begin to answer this question? Think back to the observations that you made comparing yourself to your classmates, other people in the world and other species. Did these observations give you enough evidence to answer the question? What else do we need to know? What other questions do we need to ask in order to develop a scientific argument that answers the driving question?

The purpose of this activity is for you to generate questions related to the driving questions that are interesting to you. Your teacher will construct a driving question board (DQB) out of the questions you raise. The DQB is a place for your class to collect everything you learn in one place. Then you can see and remember everything you've done to help you answer the driving question.

Write the Driving Question below:

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1. This unit is about similarities and differences in biological features. Remember that biological features include biological parts both inside and outside of our body. Make a list of questions that you have about biological features. What questions could you ask that might help you figure out the role of DNA? What other questions do you need to answer in order to answer the driving question? Your teacher will use your questions to organize the rest of the unit. Write your questions in the space below.

2. Discuss your questions with your group and determine which questions you all agree are important to ask. Write those questions on sticky notes.
3. Group the questions together that are similar.

CONCLUSION
The DQB that you, your classmates, and your teacher create will help you keep track of your progress during the unit. You should ask yourself how each activity in the unit helps you understand how similar or different we are. Draw a copy of the class driving question board on the next page. As you learn new things in this unit you can add them to your board.

Driving Question Board

Activity 1.3

How similar or different is our skin?-Building a Model of Skin

PURPOSE

Scientists use models to explain how things happen or to help them understand a complex problem or system. They also use models to make predictions about how things behave. Have you ever seen a model of teeth at the dentist's office? Because it is hard to see inside our own mouth, the dentist uses the model to show us the shape and size of each tooth. They can also use the model to show how the teeth fit together when you chew your food. Computer simulations can help doctors learn how to do surgery or predict the outcome of new procedures. Models can also be drawings or even mathematical equations. Models are used to show the relationship between parts of things that are not easy to see.

In this lesson you will draw a model of the skin. Your model will help you answer the driving question "How similar or different are we?"

1. Observe your skin with a hand lens. Draw a detailed picture of what you see and label it.

Draw a detailed picture of your skin.

2. Compare your drawing with someone else at your table. Describe similarities and differences.

Similarities	Differences

3. Prepare a slide of the skin on the back of your hand by following these instructions:

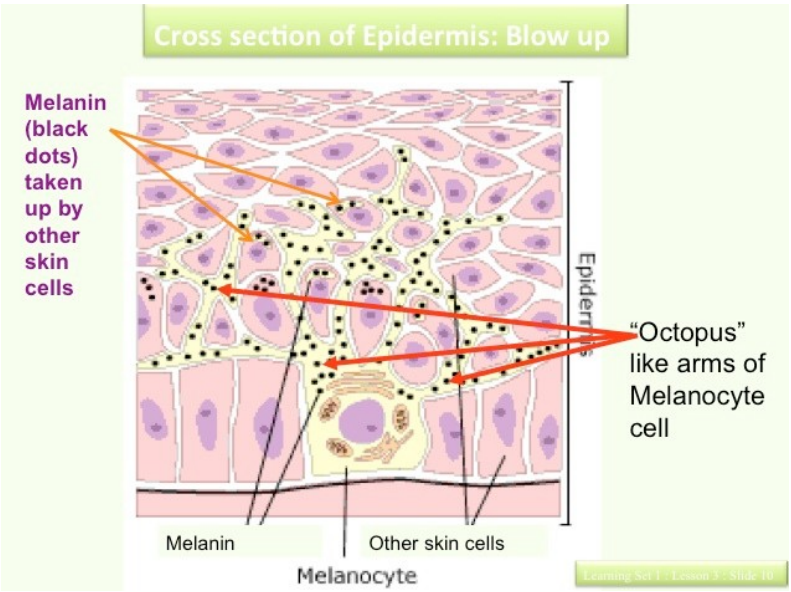
1. Cut a small square out of the center of a small card.
2. Cover the hole with a piece of clear tape
3. Lightly press the sticky side of the tape to the back of your hand.
4. Observe your slide, sticky side up, under a microscope
5. Draw observations of your skin and label any parts you can identify.

Drawing of skin as seen under a microscope.

4. Observe someone else's slide and their model of the skin. Describe similarities and differences between yours and theirs.

Similarities	Differences

Could you see many differences? How could you change your model of the skin to make it more useful in explaining differences in skin color? What needs to be added to your model?



5. Look at the model of skin in above slide. How does this slide help us explain differences in skin color? What useful information does it provide about our skin?

6. How similar or different are light and dark skin? Make a prediction by drawing a model of each and comparing them. Make sure your models include melanocytes and melanin and label them.

Dark Skin Model	Light Skin Model

7. Compare your model to the dark and light skin slide that your teacher shows and make any needed changes. List similarities and differences in light and dark skin.

Dark and Light Skin Comparison

Similarities	**Differences**

8. How similar or different is our skin? Write a scientific explanation to answer this question. Make sure that you make a **claim** related to the question. Use **evidence** from your model and the slides that you observed. Use information about the work that melanocytes do to write your **scientific reasoning**.

Claim: (Answers the question)

Evidence: (2 or more statements that support your claim from your observations of skin and skin cells)

Scientific Reasoning: (reasons why the evidence supports your claim based on scientific principles about melanocytes and melanin related to what they do).

Reading 1.3

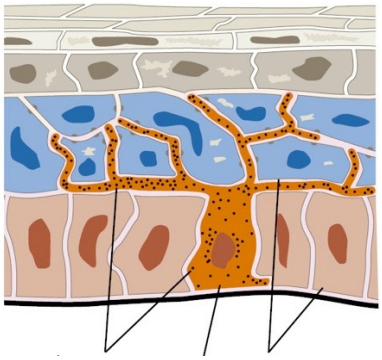
Before You Read...

Think about the key words below. How are they related? What do they have to do with skin color? Circle the words as you encounter them in the reading. Think about how they may help us to answer the Driving Question- "How similar or different are we?"
Key words: organs, epidermis, cells, organelles, melanocytes, melanin,

How Cells Affect Skin Color

How similar or different are we? When you look around in class, on the street, at the library or at the store, you might notice that people have different skin colors. When you look in the mirror, what do you see? You see eyes, a nose, a mouth, some skin, hair and all types of body parts. These are things that we all have in common. If you looked at your body at the microscopic level you would see that we all have about 10 trillion cells. Cells are the basic building blocks of all life, so in order to understand what makes people different and similar to one another, we have to understand the properties of cells and how they function. For example, our muscles are made of a unique type of cell, our livers are made up of a different unique type of cells. It is the difference between the types of cells that make each organ unique. In order to understand how the body works, you need to understand how cells work. All bodily functions and body developments, such as reproduction, fighting infections and skin color work at the cellular level.

In class you made models of the skin and observed slides of actual skin cells. We can learn more about skin color by looking at the cells that make up the skin. The skin is made up of specific kinds of cells that are packed into layers. The top layer of cells is called the epidermis. This layer of cells provides strength to our skin. Groups of cells with similar structure and function are called tissue. The epidermis is made mostly made of epithelial tissue.

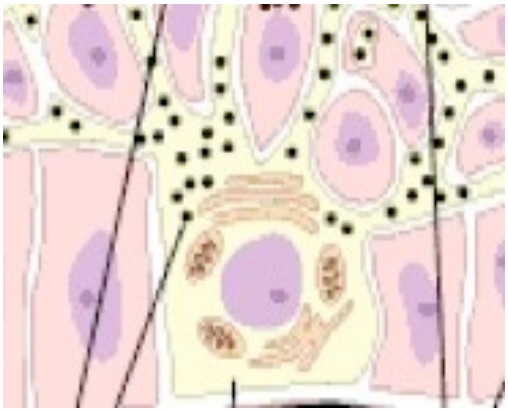


Melanin Melanocytes other skin cells

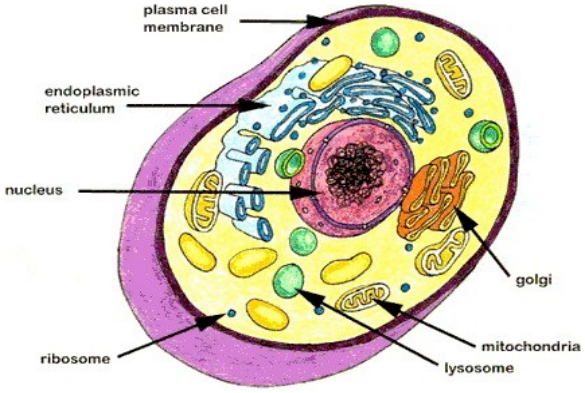
The epidermis has special cells that are involved in skin color. At the lower layer of the epidermis, there are cells called Melanocytes. Melanocytes are skin cells that produce molecules called melanin. Melanin is a substance in skin, which gives the skin its color. The other skin cells take up the melanin produced by the melanocytes.

An **organelle** is a structure in a cell that has a specific function.

Look at the images below. Within cells, there are many organelles. You might have heard of the mitochondria and the nucleus- these are some organelles of the cell.



melanocyte skin cell



animal cell

What are some similarities and differences between the melanocyte skin cell and the animal cell pictured above?

Similarities: _____

Differences: _____

Look closely at the diagrams of the skin: If melanocytes produce melanin so far below the top layer of the skin, how do you think melanin reaches the top layer of the skin to give people their skin color?

Apply

In the previous reading, you learned that skin color comes from melanin in the skin cells. Does your skin color ever change? Are you the same color in the summer as in the winter? What might cause your skin change? The following reading explains why skin color and skin composition differ between regions of the world. Think about how you might relate to this text.

Key words: melanin, pigment, melanocytes, melanin, ultraviolet radiation, DNA.

Skin Color Adaptation

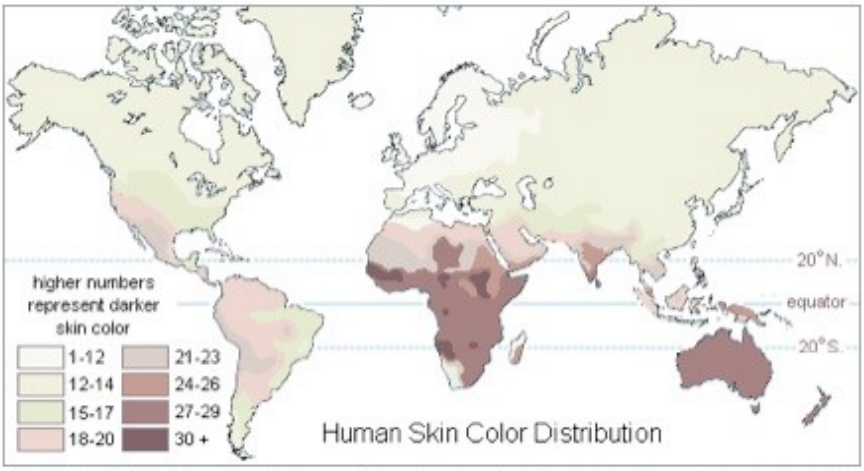
http://anthro.palomar.edu/adapt/adapt_4.htm (Text modified)

Some of the variation in human skin coloration (Sub-Saharan African, Indian, Southern European, and Northern European)

Non-forested tropical regions of the earth usually have the most intense ultraviolet radiation from the sun. People with darker skin in those regions have an advantage. Their skin has more melanin. Melanin acts as a protective biological shield against ultraviolet radiation, which is an invisible, high-energy part of sunlight. Melanin helps to prevent sunburn damage that could result in DNA changes and, subsequently, melanoma --a cancer of the skin. Melanoma is a serious threat to life. In the United States, approximately 54,000 people get this aggressive type of cancer every year and nearly 8,000 of them die from it. Those at highest risk are European Americans. They have a 10 times higher risk than African Americans. They also have less melanin in their skin cells.

Ultraviolet radiation reaching the earth usually increases in summer and decreases in winter. The skin's ability to tan in summertime is an adaptation to this seasonal change. Tanning is primarily an increase in the number and size of melanin granules due to the stimulation of ultraviolet radiation.

It would be harmful if melanin acted as a complete shield. A certain amount of ultraviolet radiation (UVB) must penetrate the outer skin layer in order for the body to produce vitamin D. However, too much ultraviolet radiation penetrating the skin may cause the break down of folic acid (one of the B vitamins) in the body, which can cause anemia , which is a deficiency in red blood cells. Because folic acid is needed for DNA replication in dividing cells, its absence can have an effect on many body processes. It may be that the ability to produce melanin was an advantage for our early human ancestors. It helped some of them to survive by preserving the body's folic acid supply in addition to reducing the chances of developing skin cancer.



The distribution of human skin color in about 1500 A.D.--darker skin colors are found mostly between 20 degrees north and south of the equator

People who live in far northern latitudes, where solar radiation is relatively weak most of the year, have an advantage if their skin has little shielding pigmentation. Most people have less melanin where ultraviolet radiation is weak. In such an environment, very dark skin is a disadvantage because it can prevent people from producing enough vitamin D, potentially resulting in rickets disease (abnormal bone formation resulting from inadequate calcium in bones) in children.

For more information on the evolutionary aspect of skin color go to:
www.clinuvel.com/dermatology/melanin

1. What are the advantages and disadvantages to having dark skin color?

2. What are the advantages and disadvantages to having light skin color?

3. How can the environment affect how similar or different we are?

Driving Question

As of now, you have learned that humans are very similar to each other, but also have many differences. An example of a difference is skin color. Some people have darker skin while some have lighter skin. The reading says that different skin colors come from different amounts of melanin (a type of molecule) found in skin cells. Even though people have many differences, there are common biological features that all humans share, like lungs and a heart. All people need to use their lungs to breathe, and use their heart to transport oxygen throughout the body. We all have melanocytes. The difference is that in some people the melanocytes produce more melanin than in others.

The Driving Question asks, "How similar or different are we?" Look back at your explanation to the question "How similar or different am I to others?" in lesson one. What do you think? Has your thinking changed? How would you answer the Driving Question given all of the information you know about skin color? Make a claim. Give evidence and scientific reasoning.

Your **claim** should answer the question "How similar or different are we?"

Your **evidence** should be from the pictures you saw showing the biological structures in skin and the maps showing the distribution of skin color.

Your **reasoning** should be scientific principles related to the function of the biological features of the skin.

After you write your explanation, circle your claim, underline your evidence and put a check by your scientific reasoning.

Activity 1.4

Macro & Molecular Chart

Copy the final macro-molecular chart you generated as a class in the space below.

Macro **Larger** **size** **Smaller** **Molecular**		

Check for understanding:
Look back to the reading "DNA helps families find adopted kids' siblings". Skim through this reading to find any specific biological structures that are not already in our macro-molecular chart. Identify where you would put the structure(s) in our chart, and explain why. If you don't think the specific structure(s) you find match up with any of our general biological structures, explain why, and suggest a general structure we might be able to add to our chart.

Activity 1.5

Atoms & Molecules

1) What do you already know about **molecules**? Write and/or draw *anything* you think you already know about what a molecule is, what a molecule looks like, examples of molecules, etc.

2) What do you already know about **atoms**? Write and/or draw *anything* you think you already know about what an atom is, what an atom looks like, examples of atoms, etc.

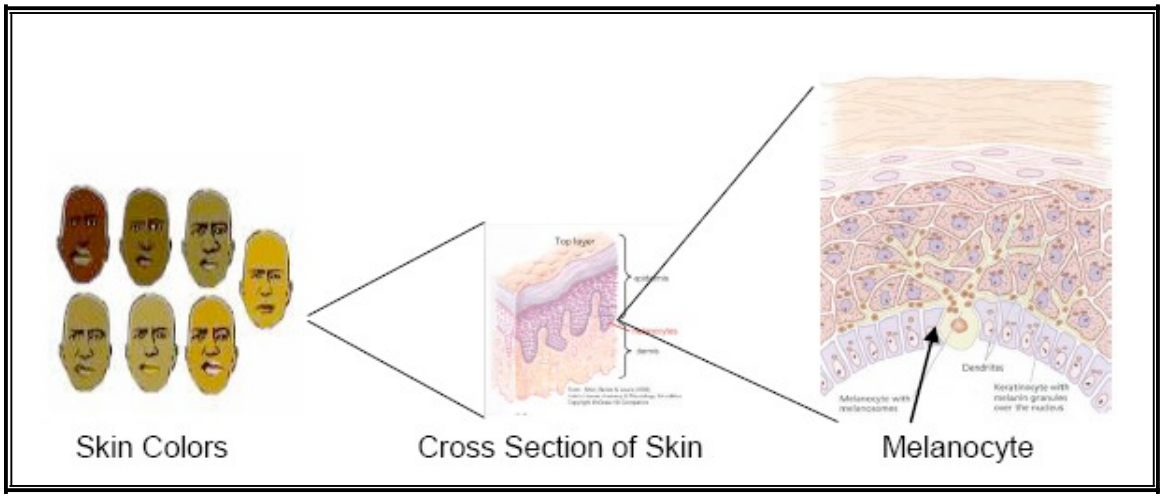
3) Write the names and symbols of some common elements below.

Check for understanding:

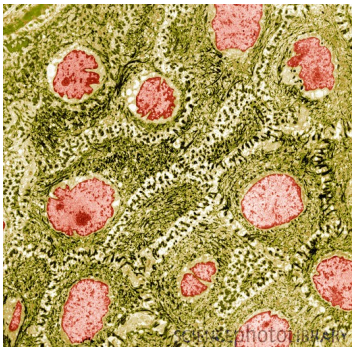
1) Describe how atoms and molecules are related to each other.

2) Look back to the macro-molecular chart we created in the previous lesson. Describe how atoms and molecules are related to the other general structures listed.

Learning Set 2:
What Happens Inside Our Cells to Make Us SIMILAR or DIFFERENT?



Activity 2.1
What happens inside skin cells?



Skin cells

Overview of our skin cells
From: http://kidshealth.org/kid/htbw/skin.html

Look down at your hands for a minute. Even though you can't see anything happening, your epidermis is hard at work. At the bottom of the epidermis, new skin cells are forming.

When the cells are ready, they start moving toward the top of your epidermis. This trip takes about 2 weeks to a month. As newer cells continue to move up, older cells near the top die and rise to the surface of your skin. What you see on your hands (and everywhere else on your body) are really dead skin cells.

These old cells are tough and strong, just right for covering your body and protecting it. But they only stick around for a little while. Soon, they'll flake off. Though you can't see it happening, every minute of the day we lose about 30,000 to 40,000 dead skin cells off the surface of our skin.

So just in the time it took you to read this far, you've probably lost about 40,000 cells. That's almost 9 pounds (4 kilograms) of cells every year! But don't think your skin might wear out someday. Your epidermis is always making new skin cells that rise to the top to replace the old ones. Most of the cells in your epidermis (95%) work to make new skin cells.

And what about the other 5%? They make a substance called **melanin** (say**: mel**-uh-nun). Melanin gives skin its color. The darker your skin is, the more melanin you have. When you go out into the sun, these cells make extra melanin to protect you from getting burned by the sun's ultraviolet, or UV, rays.

The next layer down is the **dermis** (say: **dur**-mis). You can't see your dermis because it's hidden under your epidermis. The dermis contains nerve endings, blood vessels, oil glands, and sweat glands. It also contains **collagen** and **elastin**, which are tough and stretchy types of **proteins**.

The third and bottom layer of the skin is called the **subcutaneous** (say: sub-kyoo-**tay**-nee-us) **layer.** It is made mostly of fat and helps your body stay warm and absorb shocks, like if you bang into something or fall down. The subcutaneous layer also helps hold your skin to all the tissues underneath it.

Genetically engineered mice
Adapted from: http://www.physorg.com/news171221180.html

Scientists have worked with mice that have been genetically engineered without the **proteins** TCF3 and TCF4, which reside in the nucleus of skin cells. They found that without TCF3 and TCF4, all of the layers of the mice's skin still develop properly, but they cannot be maintained. The skin cells lose their capacity to self-renew and replace skin cells that have died. The scientists showed that the epidermis cannot be maintained long-term without these two **proteins**.

WikiAnswers about skin
From http://wiki.answers.com

The skin protects the body by warding off organisms and substances that cannot penetrate it's outer layers. **Keratin**, the **protein** that forms the main component of the dead, surface face cells makes the skin waterproof and tough.

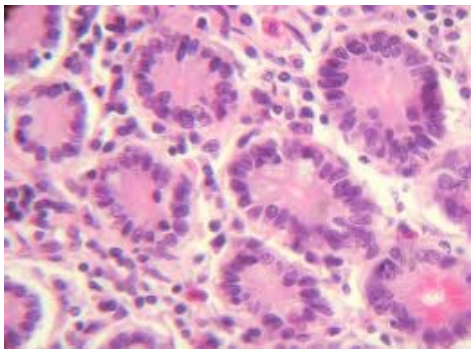
Proteins and the health of our skin
From: http://www.judymcfarland.com/skin.shtml

Elastic skin is a sign that a person has ample **collagen**, the strong cement-like **protein** that is found both inside and outside cells to help them retain their structure. Collagen is a structural **protein** and it is responsible for giving the cells that make up our skin their strength and elasticity. In fact, collagen comprises 30 percent of the total body **protein**. Its strong white fibers, stronger than steel wire of the same size, and yellow elastic networks, called **elastin** (another type of **protein**), form the connective tissue that holds our body together. Collagen strengthens the skin, blood vessels, bones, and teeth. It is the intracellular cement that holds together the cells in various organs and tissues. Collagen is one of the most valuable **proteins** in the human body. A person who has been sick, or who has been on an extremely low-protein diet, very often sees the muscles in his or her arms and legs begin to sag, which is a sign that they have probably lost collagen.

It is important to understand the value of **protein** in our diet. **Proteins** are necessary for tissue repair and for the construction of new tissue. Every cell needs **protein** to maintain its life. **Protein** is also the primary substance used to "replace" worn-out or dead cells.

Student Reader
Activity 2.1

What happens inside intestine cells?



Intestine cells

Overview of our intestine cells

Adapted from: http://www.blurtit.com/q299461.html

The intestines are associated mainly with the digestion and absorption of food. In human beings, intestines are comprised of the small intestine and the large intestine. The main function of intestinal cells is to secrete digestive juices. The cells in the small intestine help in a major part of digestion. This is the place where food products are absorbed. Cells in the large intestine help with water absorption and excretion (removal) of solid waste substance.

Intestine cells and digestion

Adapted from Wikipedia

The small intestine is where most chemical digestion takes place. Most of the digestive **proteins** that act in the small intestine cells are secreted by (released by) the pancreas and enter the small intestine via the pancreas. These **proteins** enter the small intestine in response to a hormone (hormones are another kind of **protein**), which is produced in the cells of the small intestine in response to the presence of nutrients from the digestion of food. The hormone secretin also causes a chemical to be released into the small intestine from the pancreas in order to neutralize the potentially harmful acid coming from the stomach.

Specific cells found in the intestines

Adapted from : http://wwwmgs.bionet.nsc.ru/mgs/gnw/trrd/thesaurus/Di/small.html

Within our intestines there are many different kinds of cells. Some of the kinds of intestine cells are described below.
Enetrocytes transport substances from lumen (inner part) of the intestine to the circulatory system. These cells also synthesize (make) the **proteins** needed for digestion and absorption.

Paneth cells contain antibacterial **proteins**, and zinc, which play a role in regulating the normal bacterial flora of the small intestine.

Enteroendocrine cells release numerous hormones and hormone-like secretions.

How nutrients are absorbed into intestine cells
From: http://www.helium.com/items/820147-structure-and-function-of-the-small-intestine

Cells lining the inner surface of the small intestine are specially adapted for the absorption of nutrients and chemicals, which you have eaten or drank.

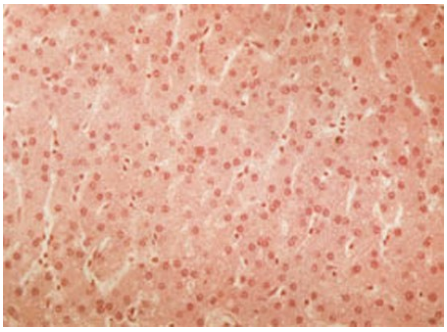
There are three primary methods by which nutrients are absorbed into intestine cells. The first of these mechanisms is known as simple diffusion. Simple diffusion requires no energy input, and takes place when there is a high concentration on one side of the small intestine cells relative to a lower concentration on the other side. Nutrients flow from areas of high concentration to areas of lower concentration.

The second method of nutrient absorption of the small intestine is known as active transport. Active transport requires energy input and allows the transport of nutrients from an area of low concentration to an area of relatively higher concentration. There are typically **proteins** involved which act as carriers for the nutrients which are being absorbed. These carrier **proteins** are then transported across the surface of the small intestine cells.

The last method of nutrient absorption in the small intestine is known as facilitated diffusion. In facilitated diffusion there are carrier **proteins**, however no energy input is required to move the nutrients.

Student Reader
Activity 2.1

What happens inside liver cells?



Liver cells

Hepatocytes

From http://www.ehow.com/about_5106552_functions-liver-cell.html

It's no secret that liver cells are socialites that know how to throw a party. They like to have a number of supporting cells around them at all times. Hepatocytes (also called parenchymal cells) are the head honchos. These popular cells make up 70 to 80 percent of the liver's mass and are involved in synthesizing protein, cholesterol, bile salts, fibrinogen, phospholipids and glycoproteins. In other words, hepatocytes ensure that our blood coagulates so we don't bleed to death, that cell communication is tip-top and that we are able to carry fats in the bloodstream. Other functions of the hepatocytes include the transformation of carbohydrates, protein storage, start of the formation and secretion of bile (which is needed for the digestion of fats in the small intestive), and detoxification and excretion (release) of substances. Thanks to these main cells, we are able to fight off disease, produce waste, transport materials throughout the body and process everything from drugs and insecticides to steroids and pollutants.

Liver cell functions

Adapted from http://www.detox-guide.com/liver-function.html

The liver cells extract its required nutrients, oxygen, toxins, and wastes from the blood. The toxins and wastes are either stored within the liver or neutralized. Let us simply breakdown the various liver cell functions performed each day:

- Produces a yellowish-green substance called bile. It is stored within the gallbladder and then used when required for the absorption (digestion) of fats.
- Regulates our carbohydrate metabolism to control our blood sugar level by storing glucose and then releasing glucose back into our system when needed for energy.
- Produces **proteins** to transport substances such as fats, hormones, and drugs).
- It stores vitamins A, D, K, B12, and folate as well as synthesize blood clotting factors.
- It helps create and break down hormones.

- Detoxifies and transforms internal toxins and environmental toxins so that they can be excreted (released).
- Liver cells, known as Kupffer cells, function to filter, ingest, and breakdown the toxic materials like dead cells, microorganisms, chemicals, drugs, and debris found in the blood passing through the liver.

Liver disease
From http://www.nlm.nih.gov/medlineplus/ency/article/002441.htm

Proteins normally help the body repair tissue. They also prevent fatty buildup and damage to the liver cells. In people with severely damaged livers, **proteins** are not properly processed. Waste products may build up and affect the brain.

Looking at liver function blood work to detect hepatitis C disease
Adapted from http://www.dummies.com/how-to/content/looking-at-liver-function-bloodwork.html

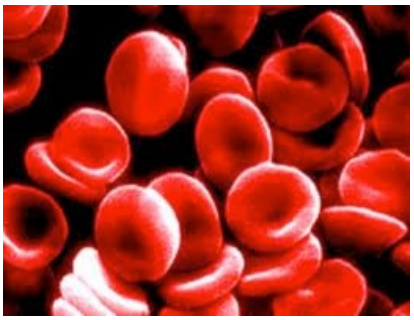
When you go for a physical, your healthcare practitioner frequently orders blood tests. Many folks first find out they have hepatitis C after undergoing a routine blood test and finding that one or more of the tests is abnormal. Because p**roteins** are extremely important to the function of the different liver cells that make up your liver organ, blood tests show the levels of different **proteins**, normally found in your liver, which have entered your blood. There is no one test that indicates hepatitis C disease. Tests for the effects of hepatitis C or other diseases that affect the function of your liver include:

Liver protein tests: These tests measure current liver cell injury by the amount of **proteins** that are "leaked" out of damaged or dying liver cells.

Liver function tests: These tests look at levels of proteins made by the liver. If your liver damage is such that your liver function is impaired and cannot function properly, levels of these **proteins** will be low.

Student Reader
Activity 2.1

What happens inside red blood cells?



Red blood cells

The functions of red blood cells

Adapted from http://www.helium.com/items/763669-the-functions-of-red-blood-cells

Red blood cells, known also as RBCs, have several important roles to play in our bodies. The primary function of red blood cells is to carry oxygen from the lungs to the tissues around your body. As a secondary function, they are also a key player in getting waste carbon dioxide from your tissues to your lungs, where it can be breathed out. When red blood cells stop functioning properly, you can rest assured that many things are going to go wrong in your body.

In order to properly understand the function of a red blood cell, you have to understand something about the structure. A typical RBC is about 6-8 micrometers in diameter, about the same as the width of a spider web strand. An RBC is biconcave in shape. Think of it like a miniature donut, only the hole in the center doesn't poke all the way through. This small shape and physical structure allows the RBC to squish in to the small capillaries where your blood vessels are the smallest. Without this ability to flex, they would easily get stuck and cause obstructions in your circulation.

The oxygen carried in your red blood cells is stored in a special **protein** known as hemoglobin. There are several different types of hemoglobin and the exact structure of this important **protein** is quite complicated, so will not be discussed here. It is simply enough to know that this **protein** is important in allowing our bodies to absorb the oxygen we breathe in. Without it, or without a properly functioning red blood cell, we would suffer serious health consequences.

Factors such as body chemistry and temperature are vital to hemoglobin function. Your red blood cells need to grab on to oxygen in the lungs and let go of it in the tissues. Subtle changes in the body's chemistry and temperature of your blood (along with some other effects) allow the hemoglobin molecules to catch and release oxygen at the proper times.

The second important function of red blood cells is the ability of red blood cells to carry carbon dioxide. Carbon dioxide is considered a waste product in every cell in your body. You need some way of getting rid of it all the time, or you will die rather quickly. Red blood cells serve as the vehicle to rid your body of this waste.

The process by which your red blood cells transport carbon dioxide is different than oxygen transport. RBCs contain another **protein** called carbonic anhydrase. As the carbon dioxide enters the RBC, this **protein**, with the help of some water, converts it into another chemical called bicarbonate. This bicarbonate is later removed either by your lungs or your kidneys. Some carbon dioxide is dissolved in your blood directly and a small amount is actually carried on the hemoglobin molecules, but the vast majority is converted to bicarbonate.

Because red blood cells are so important to your body, when they don't work properly it often leads to disease.

Activity 2.1
Cell Activities Table

Cell Type	Cell Activities
Skin Cells	
Intestine Cells	
Liver Cells	
Red Blood Cells	

Follow-up: In your new small group, briefly look back at your readings about the different types of cells. What does each type of cell appear to have in common? In other words, there is something that is necessary for and important to the functioning of all different types of cells in our bodies. What is it?

Optional Activity 2.1

Viewing Cell Videos---Part I

PURPOSE
In order to understand how we come to have some of our differences, such as different skin colors, we need to start thinking about what is really going on inside cells. You will be watching some videos that show examples of some activities of cells.

Directions: Briefly describe what is being **DONE** in each video. Focus on the action taking place and use verbs in your description.

Video	Description of action
1	
2	
3	
4	
5	
6	

Optional Activity 2.1

Viewing Cell Videos---Part II

Directions

We will look again at the cell videos, but this time you are going to identify the specific change that is happening for each cellular activity. As you identify the change taking place, write this in your activities chart below next to the corresponding cellular activity.

Video	Cellular Activity	Change
1. Cytoplasmic Streaming		
2. Mitosis		
3. Phagocytosis		
4. Neutrophil Chasing Bacterium		
5. Paramecium Exocytosis		
6. Bacteria Reproducing		
7. Melanocyte		

Activity 2.2

Where are proteins in our bodies?

1) Where do you think proteins are found in our bodies?

2) Where are proteins found in a chicken's body?

Purpose: In this activity you will perform a test on different parts of the chicken to determine if they have proteins or not. You will use a solution called **Biuret** solution. If there are no proteins present it will stay blue. If there are proteins present, it will turn purple or violet.

Materials Needed: (make sure your group has all the necessary materials before beginning)

- bags with chicken parts, labeled
- 15 mL Biuret solution for detecting the presence of protein
- 15 mL mixing solution
- plastic pipets (1 for mixing solution, 1 for Biuret solution, 1 for each type of chicken)

Procedure: Read through the entire procedure before beginning!

Part I---Making Predictions:

- Predict which chicken parts will have proteins in them. Record your predictions in the table below.

Part II---Observing the Control:

- Add one pipet full of mixing solution to each of the bags, including the bag with no chicken parts.
- Add one pipet full of Biuret solution to the bag with **only the mixing solution** (no chicken parts).

- Record the results of adding the Biuret solution to the bag without chicken parts in the table.

Part III---Testing the Chicken:

- Each bag with chicken parts should now have mixing solution in it as well. Without touching anything inside the bag, gently mash the chicken pieces so they are thoroughly exposed to the mixing solution.
- Add one pipet full of Biuret solution to each bag of chicken parts.
- Record your results in the table.

3) What is the purpose of putting Biuret solution in the bag without chicken parts?

Where are proteins in chicken parts?

Part being tested	Prediction – Is protein present?	Result – Is protein present?
Mixing solution without any chicken		

4) Using complete sentences, write a scientific explanation that answers the question: **Where are proteins found in chickens?** Make sure you include all the parts of a scientific explanation.

Claim: Answer to the question.

Evidence: Use data from your chart.

Scientific Reasoning: Use what you learned (scientific principles) about proteins.

5) **Infer:** Where are proteins found in human beings? Using the results of this activity make an inference about where you think proteins are found in our bodies.

6) **Make Connections:** How does this activity relate to you and your body? *You may want to revisit this question after reading "Proteins--what exactly are they for?"*

Reading 2.2

Proteins: What Exactly Are They For?

Think back to the videos watched yesterday and the corresponding chart we made. Do you think there are any similarities between these activities? Why?

The following reading gives a description of proteins, where we get them and how they are used inside the body.

Key words: proteins, building blocks.

When you think of proteins, what exactly goes through your mind? Many people think of proteins in regard to nutrition. On television, you might see commercials or infomercials that advertise protein shakes or pills. You may also see television shows that stress the importance of a balanced diet consisting of protein in meat and beans. Since proteins are an important and essential part of your diet, it is important to understand why there is such an emphasis on including protein in your diet.

It is very important that we eat enough proteins. Proteins are the building blocks of the body. Proteins are useful for growth, tissue repair and replacement inside the body. For example, bones and muscles need a steady supply of proteins in order stay healthy.

Think about the different foods that you eat on a daily basis. Do you eat meat or fish? If you do, these are excellent sources of protein. However, meat is not the only way to incorporate protein into your diet. If you ate cereal this morning with milk, or had some vegetables at lunchtime, then you had protein today.

Proteins are molecules that are required for the structure, function, and regulation of the body's cells, tissues, and organs; and each protein has unique functions.

What do proteins do once they enter the body? When your body eats food that contains proteins, your body breaks down the food and rebuilds the protein so that it may be useful for *your* body. When the proteins are reassembled, different cells use them for different activities. It is important to note that your body does not take in the proteins and use them as they are. Your body always breaks them down and rebuilds them for the body to use. That way they can be used for specific jobs around the body, such as breaking down food and other molecules, building molecules that are used in the cell, transporting molecules around the body, and cellular division.

1. What do proteins do and why are they important for bodily functions?

2. What are some functions of proteins inside the body?

3. What are some ways to make sure that you get protein in your body?

4. List 4 foods from your diet that provide protein.

Activity 2.3A

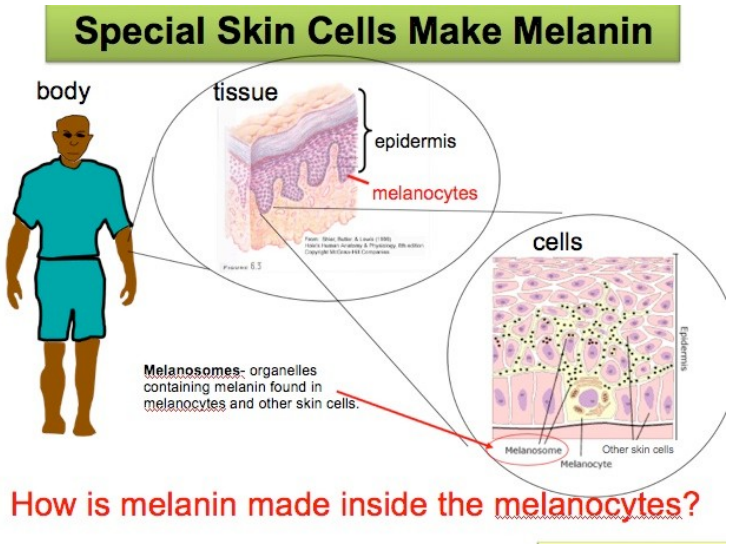
Modeling Proteins in Skin Cells

Purpose

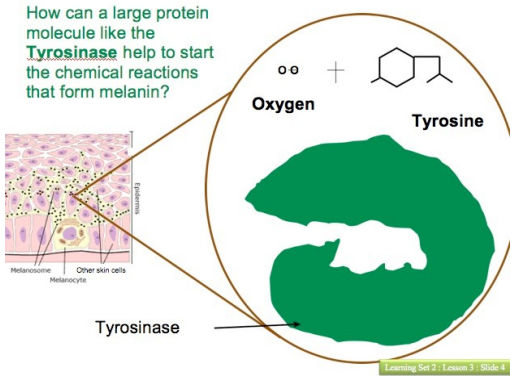
At the beginning of this unit we asked, "How similar or different are we?" We found out that in order to really identify our similarities and differences we have to look closer at what is going on in the cells that make up our bodies. In this lesson you will explore how our skin cells are able to make the melanin that causes differences in our skin color.

In the last two lessons you found out that protein is a very important molecule in our body. You tested a chicken and found protein in many body parts including the skin. What else do we need to know about protein in order to determine if it is involved in the process of making melanin in the skin? Write two questions below that you think would be important to know.

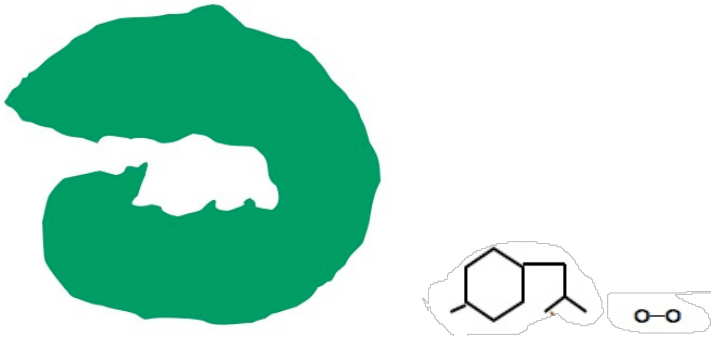
1.What else do you think we need to know about proteins? What questions do you have about proteins?



2. We have already learned that melanin is made inside the organelles of skin cells called melanocytes. When we look inside of those organelles called melanosomes, we find protein molecules called tyrosinase. We also find two other molecules, oxygen and tyrosine. They need to react together in order to begin the process of making melanin. Some proteins can help start chemical reactions without being changed themselves. Look closely at the shape of the inside of the tyrosinase protein and the shape of the oxygen and the tyrosine. How could tyrosinase help these two substances react?



3. Complete the picture below by drawing the tyrosine and oxygen molecules inside of the tyrosinase to show the new molecule that is being formed. Describe how the new molecule is formed below the picture.



4. What else do we need to know about tyrosinase in order to understand why we have different skin color? What questions do you have about tyrosinase. Write 2 questions below.

Add these questions to your Driving Question Board.

Activity 2.3B

Observing Tyrosinase

Have you ever seen apple or banana slices that start to turn brown? This process is called enzymatic browning. As the fruit is cut the plant cells are exposed to oxygen. Plant cells contain the enzyme tyrosinase just like our skin. When exposed to oxygen the tyrosinase helps start the chemical reactions that produce the pigment melanin in plants. This is why you see brown spots on your fruit when you cut it. Although you may not like sinking your teeth into the mushy brown spots, enzymatic browning does serve a purpose. It protects fruit from insects, bacteria, and fungi.

Question: Does tyrosinase always make the same amount of melanin?

Hypothesis:

_____

_____

Materials
fruit- bananas or apples
Lemon juice
hand lens

Procedure (Steps 1-3 have already been done by your teacher)

- Prepare 2-6 small pieces of fruit from the same apple or banana.
- Dip half of the fruit in lemon juice and leave the other half untreated.
- Let the fruit sit out overnight.
- Observe both the treated and untreated fruit and record your observations.

Observations

Similarities in fruit	Differences in fruit

Why do you think the fruit in the lemon juice looks different from the fruit not treated with lemon?

Conclusion:

1. **Claim**: Answer the question "Does tyrosinase always make the same amount of melanin?"
2. **Evidence:** Use your observations comparing the fruit to tell why you think tyrosinase does or does not make the same amount of melanin.
3. **Scientific reasoning**: Use what you know about how tyrosinase helps make melanin to explain why your observations support your claim.

Claim

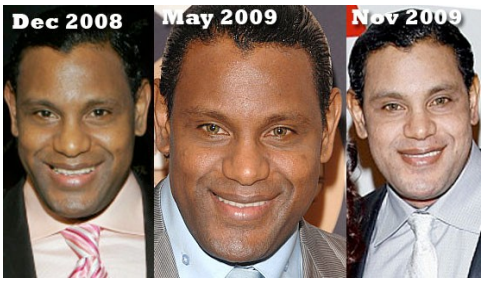
Evidence

Scientific Reasoning

.

Reading 2.3

What Happened to Sammy Sosa?



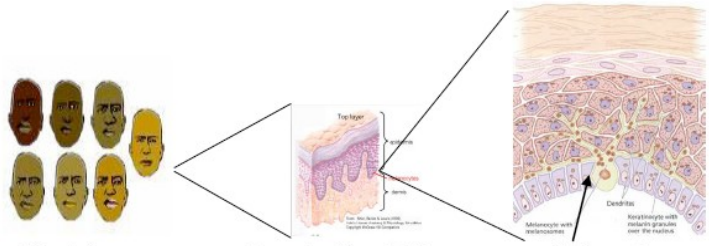
/en.wikinoticia.com/entertainment/gossip/22592-sammy-sosa-and-skin-whitening

Baseball legend Sammy Sosa created headlines when he arrived at the Latin Grammys in 2009 looking very different. Sammy Sosa's skin had changed color. He went from brown to almost white skin tone is a matter of months. Was he sick? According to Sosa he was using a skin cream to soften his skin and it also lightened his skin color.

How could this happen? Many lightening creams advertise that they work by interfering with the skin's ability to create melanin. We know from other lessons that people with less melanin in their skin cells have lighter skin. In order to understand how these creams work we need to know how melanin is formed in skin cells.

Special Skin Cells Make Pigment

Think back to the chicken investigation. We learned that proteins are a type of molecule found in the body. We also learned that many parts of the body contain proteins including the skin. How does this investigation help us explain the differences in skin color that we see? Could proteins be involved in this process? To find the answers to these new questions we need to look at what is happening inside of the cells.



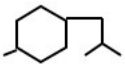
We have already learned that although people can range from very dark to very light skin color, the parts of their skin are the same. We can see this by looking at a cross section of what a person's skin tissue might look like. All skin is made of different layers (epidermis and dermis). Within the skin tissue, there are cells called melanocytes. Look closely at the image on the far right. The small shaded dots are organelles inside of the melanocytes that produce melanin molecules. Melanin is a type of pigment that gives skin its color. The more melanin produced the darker the skin color will be. How is melanin produced? Are proteins involved in this process? In order to explain why different people have different amount of melanin we need to know the answer to these questions.

How is melanin produced inside skin cells?

Have you ever seen an assembly line in a factory where cars are produced? In an assembly line, machines work, one by one, to put the car together. In the assembly line, each machine is doing something different and has a specific role in putting together the car.

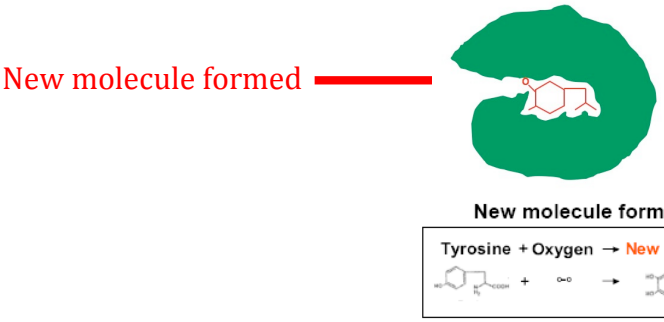
There is a similar process that takes place inside the body, more specifically, inside the cells. In order for melanin to be produced, there is an assembly line of protein molecules that add and change parts of other molecules. Protein molecules are responsible for helping chemical reactions in the body. This chain of chemical reactions produces melanin. **Tyrosinase** is the protein responsible for the first chemical reaction needed to produce melanin. If Tyrosinase does not act, then melanin cannot be made.

Oxygen



Tyrosine– small molecule needed build melanin

Tyrosinase– large molecule made of protein



In the above diagram, tyrosinase acts on tyrosine (a small molecule involved in the production of melanin). tyrosinase acts like a lock. It has a specific shape that allows the oxygen and the tyrosine molecules to fit together inside of it like a key. This is the first step in the chemical reactions that produce melanin. At the end of the chemical equation, melanin is produced.

- Why is the shape of tyrosinase so important to the production of melanin?

- If tyrosinase is unable to make melanin, what might happen to skin color?

Now that we know how the skin cells make melanin, how did skin cream change Sammy Sosa? Many of the skin whitening creams on the market today, claim to change the ability of skin cells to make melanin.

Melanin Vanish →

Another cream called Covermark Luminous for Ethnic/Dark & Heavily Pigmented Skin" explains in their advertisements how their product works:

The innovation of Covermark's all-NEW Luminous range is a 5 step de-pigmentation action in ALL stages of the melanin formation:

Actions inside the Melanocyte cell:

Step 1. Prevent tyrosinase synthesis. (Nanopeptide-1, an innovative biomimicking peptide)
Step 2. Sustain tyrosinase in its inactive form (Diacetyl Boldine, a modified natural active)
Step 3. Inhibit formation of matured melanosomes (Sucrose Laurate and Pea Extract)
Step 4. Diminish tyrosinase activity (Alpha-Arbutin, Vitamin C, herbal extracts rich in flavonoids and phenylflavone derivatives)
Step 5. Slow down the transfer of melanosomes to the upper layers of the epidermis
www.skinlight.co.uk/product_465_Luminous+Supreme+Whitening+Cream.html

Look at the advertisements above.

3. Sammy Sosa said that he used skin-lightening creams. How do you think Sammy Sosa's skin changed from dark to light?

4. Think back to the fruit investigation from the last lesson. Is what happened to Sammy Sosa's skin similar to what happened to the fruit? Why or Why not?

Making Connections to our Driving Question

How similar or different are we?

In learning set one we began to answer the question "How similar or different is our skin?" We found out that everyone has melanocytes in their skin that produce melanin. We also found out that people with darker skin have more melanin than people with lighter skin. Can the things that we learned in the last lesson help us add more to our answer to this question?

Turn to back to Learning Set 1 page 17-18 in your reader. Think about your answers to the question "How similar or different is our skin?"

- Is melanin made the same way in light and dark skin?
- Does light and dark skin contain the same protein molecules?
- What might cause the difference in the amount of melanin?

5. Write a new scientific explanation to answer this question. Add new sentences to reflect any new information that you feel is important to answering the question.

Claim: Your answer to the question How similar or different is our skin?

Evidence: Data and information from the apple and chicken labs and readings to support your claim.

Scientific Reasoning: Use scientific principles to explain why your claim is accurate.

Reading 2.4A
Don't Pass the Milk, Please

Lactose Intolerance KWL Chart

What do you Know?	What do you Want to Know?	What have you Learned?

The following case will provide an introduction to some of the concepts you will be learning in this lesson. Circle the key words as you come to them in the reading. Try to use the case to understand what the Key Words mean and how they are used in the body. Add their meaning to the **L** in your KWL chart. As you read this case, think about how the people in the case differed from each other.

Key words: Lactose, lactase, Lactose, Intolerant, digest, protein.

Don't Pass the Milk, Please

It was a warm summer morning and Jason was late for his basketball game at the corner park. He quickly threw on some clothes and ran down the stairs from his bedroom.

As he zoomed past the dining room, his mom called after him, "Jason, you can't leave this house without some breakfast! Come on back here and have a seat with your sister."

"But mom", Jason complained, "I have a basketball game and I'm really late."

His mom answered, "It doesn't matter. Have a quick breakfast then. You need to put something in your stomach."

"O.K, fine. I'll just have a bowl of cereal… is that O.K. with you, Mom?" Jason asked.

"Don't get smart with me. Chelsea, pass your brother the box of cereal and milk", said Mom

Chelsea gave Jason the box of cereal. Jason quickly filled his bowl with cereal, added milk and sped through his meal. Before long, Jason was out of the door and on his way to the basketball game. As he ran he noticed his stomach felt a little funny. This was not the first time his stomach felt a little upset in the morning. Usually he ignored it and it went away so he kept on running.

When Jason arrived at the game, his friends immediately gave him a hard time.

“Jason! Where have you been? We are down a man because of you! We have to make up two baskets in order to tie with the other team!” cried Marvin, one of his friends.

“Well… my mom made me stay and eat breakfast.”

Instantly, all his friends began to laugh. Just as Jason started to join in, he suddenly grabbed his stomach and doubled over in pain.

“Hey man, are you ok?” asked Marvin.
The pain became worse and worse, until Jason finally asked to be taken home. Something was very wrong.

As Jason and his friends entered the house, his mother ran towards them. “What’s wrong with him? Jason, are you alright?” she asked.

Jason replied, “I think I need a doctor, Mom. I don’t feel so good!”

At the doctor’s office, Jason went through several tests. The doctor asked Jason if this was the first time that he felt this way. Jason admitted that he had been feeling sick in the mornings for a while but today was the worst. A few weeks later, after all of the tests were analyzed, Dr. Sandoval sat Jason and his mom down and began to explain why Jason felt sick.

“You have what is called lactose intolerance” said Dr. Sandoval.

“Lactose intolerance? What is that? Is it serious?” replied Jason.

Dr. Sandoval tried to calm Jason down. “No, it is not serious. You are a completely normal young man. Lactose is a sugar found in milk and intolerance means that you have an extreme sensitivity to it. Therefore, lactose intolerance means that you cannot digest milk, but it doesn’t mean that you can’t do all of the normal things that other people do. Many people all over the world have this condition.”

Jason was confused. He remembered that his sister ate the same exact breakfast that he ate, but did not complain of pain. Jason asked the doctor, “Why didn’t my sister get sick? She drank the same milk that I drank.”

Enzyme: Protein molecules responsible for helping chemical reactions in the body.

“Well” replied Dr. Sandoval, “although you and your sister are similar in many ways, you are different because her body can produce a particular protein. This protein is called lactase. Proteins have many functions, and one function is to help in digestion of

food. Lactase is a protein that breaks down lactose. Lactose is a type of sugar found in milk.

Jason, you, on the other hand, cannot digest lactose. You do not have enough lactase protein. The small intestine needs lactase protein to break down lactose. If lactose is not digested, it can cause gas and stomach cramps."

Justine asked Dr. Sandoval, "What can we do to prevent this from happening again?"

"Jason can stay away from food and drink products that contain milk or he can look for milk alternatives", replied Dr. Sandoval. "While your sister is drinking milk with her cereal next time, you will have to find something else to eat."

Jason was relieved to find out that he was fine. He was even happier to know that he did not have to drink milk anymore.

In "Don't Pass the Milk Please", Jason became ill when he had milk in his cereal.

Use the Key words from this case to write sentences below to describe what you "**Learned"** about Lactose intolerance from the reading. Share your ideas to complete the class KWL chart.

Activity 2.4
Investigating the Enzyme Protein Lactase

Purpose (Day 1)

In this activity you will determine whether the medicine used to treat lactose intolerance has lactase in it. Lactose free milk is made from cow's milk. You will also determine whether it contains lactase. You will use test strips that are able to detect the presence of glucose. When you put the strips in any liquid they will turn a certain color to indicate how much glucose is present. Using the chart you will be able to determine how much, if any, glucose is in milk before and after you have added lactose intolerance medicine to it.

Question 1: How does the medicine used to treat lactose intolerance work?
Hypothesis: (What do you think will happen. Explain why you think so)

Question 2: How is lactose free milk different from regular milk?
Hypothesis: (What do you think will happen. Explain why you think so)

Materials

- Crushed 1/2 lactose intolerance pill
- 5-10 mLs milk
- 5-10 mLs lactose free milk
- 3 glucose test strips

Procedure
Control: No Medicine

1. Make sure that you have all of your materials.
2. Using the chart on the next page, predict what you think the color of the strip will be after dipping it in the milk.
3. Put one of the glucose test strips in the cup of milk for 1 second and then wait 30 seconds. (Have a classmate time you for 30 seconds after taking the glucose strip out of the cup of milk.)
4. After 30 seconds, compare the color to the chart on the bottle. What color is your strip?
5. Record the results in the chart below.

Experiment 1: Lactose intolerance pill in milk.

1. Add the crushed lactose intolerance pill to the cup of milk. (It is important not to add the pill before this point.)
2. Swirl the pill gently into the milk.
3. Use the heat of your hand to help warm the milk by holding the cup in your hands. DO NOT PUT YOUR HANDS OR FINGERS IN THE MILK.
4. Have a classmate time you while holding the milk for 5 minutes.
5. Using the chart on the next page, predict what you think the color of the strip will be after dipping it in the milk.
6. After heating the milk with the lactase in your hand for minutes, use a fresh glucose test strip to test for the presence of glucose. Put one of the glucose test strips in milk for 1 second.
7. Wait 30 seconds before comparing the color to the chart on the bottle.
8. What color is the strip now? Record the results in the chart below.

Experiment 2: Lactose Free milk.

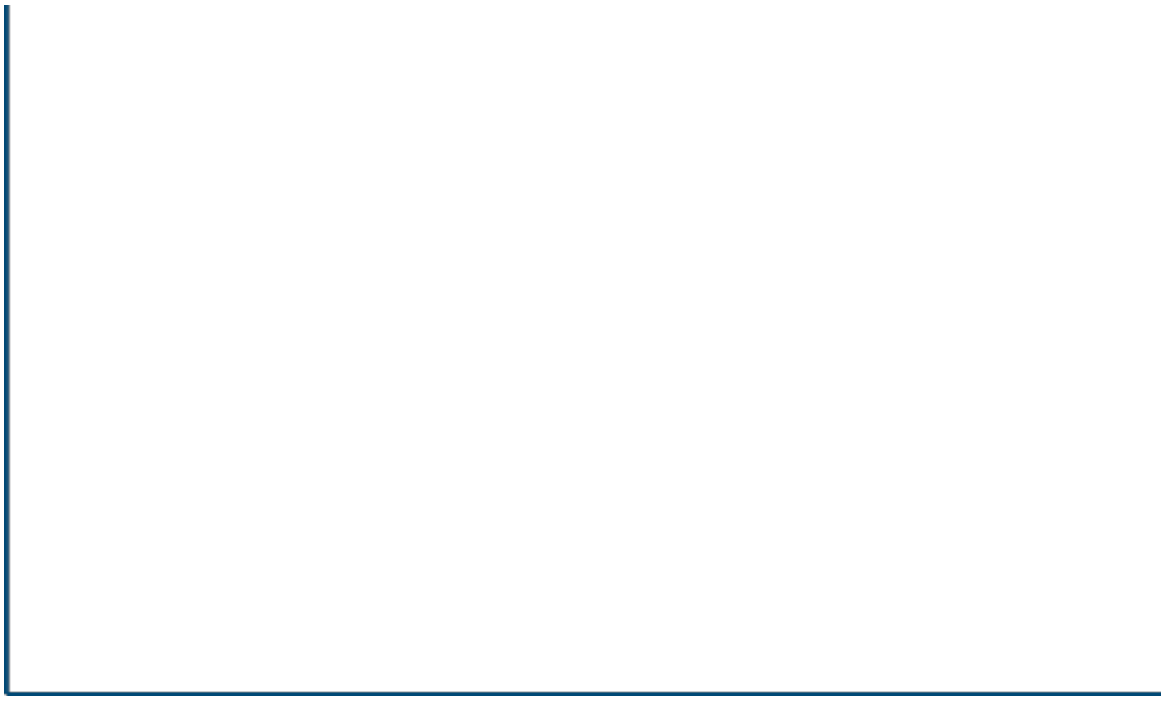
1. People with lactose intolerance can drink lactose free milk. Lactose free milk is made from cow's milk. Look at the results from the last test. How do you think they might make lactose free milk? Using the chart on the next page, predict what you think the color of the strip will be after dipping it in the lactose free milk.
2. Put one of the glucose test strips in the cup of milk for 1 second and then wait 30 seconds. (Have a classmate time you for 30 seconds after taking the glucose strip out of the cup of milk.)
3. After 30 seconds, compare the color to the chart on the bottle. What color is your strip?
4. Record the results in the chart below.

Observations: Charts, Tables, Graphs

Milk	**Predict the Color of Strip**	**Color of Strip?**	**Glucose (mg/dl)**
Without pill in regular milk			
5 minutes after adding pill to regular milk			
Without pill in lactose free milk			

Graph Your Results (Day 2)

Glucose mg/dl



Milk without pills Milk with pills Lactose free milk

Analysis of Data

1. What are the variables in the experiment?

- **Controlled**_____
- **Independent** _____
- **Dependent** _____

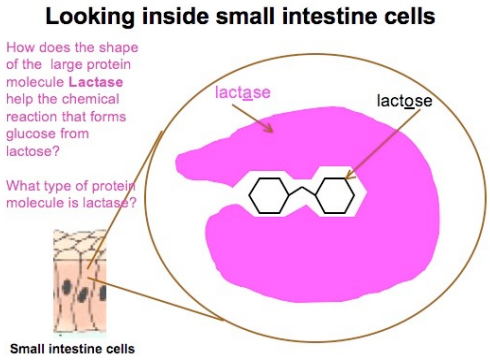
2. Are there any variables that could have affected the reliability of the data?

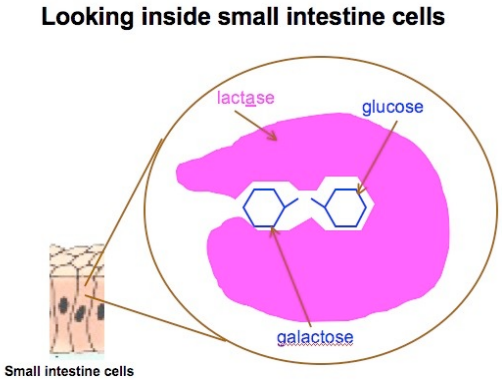
3. Why do you think the glucose test strip changed color after dipping it in the solution containing the pill?

4. Why are we testing milk for the presence of glucose before and after adding the pill?

5. What evidence do you have that the medicine used to treat lactose intolerance contains lactase?

6. What evidence do you have that lactase is already in the lactose free milk?





Conclusion-- Write a scientific explanation to answer the questions "How does the medicine used to treat lactose intolerance work? How is lactose free milk different from regular milk?" Write a separate explanation for each question. Include a **claim** and **evidence** from your experiment. Think about the slides that described how lactase breaks down lactose in the cells and the information recorded under "L" in your "KWL" from the "Don't Pass the Milk, Please" story. Include this information in your **scientific reasoning** for each explanation.

How does the medicine used to treat lactose intolerance work?
Claim:

Evidence:

Reasoning:

How is lactose free milk different from regular milk?
Claim:

Evidence:

Reasoning:

Think back to the story "Don't Pass the Milk, Please". Jason could not digest milk. After doing this activity, how do you think the results of your experiment might help Jason?

Reading 2.4B

New Spoof of Milk Mustache Ad Spotlights Lactose Intolerance

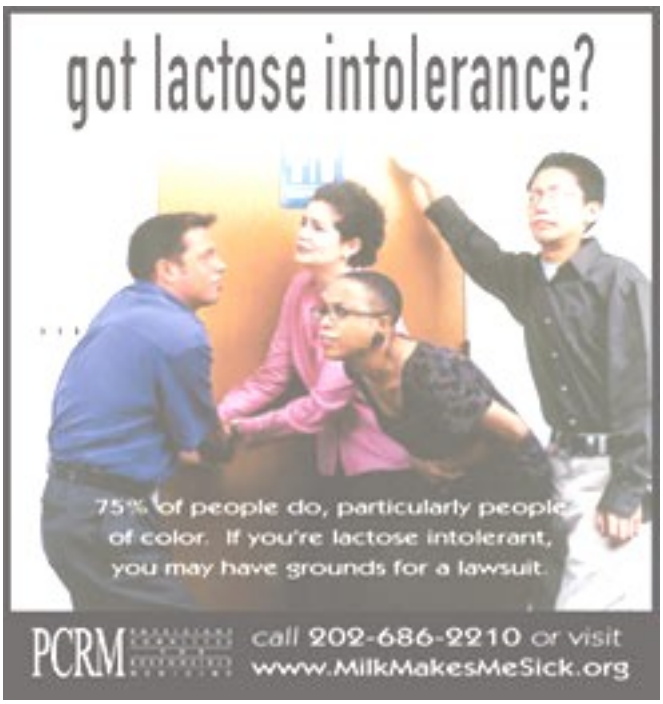
This article is about a group that would like to sue the dairy industry. They think that people with lactose intolerance are being treated unfairly. Answer the questions below before you read the article. Read the article and then decide if you have changed your mind about any of your answers.

Anticipation – Reaction Guide

	Before Reading	***After Reading***	
	What do you think?	***What does the author think?***	***What do you think now?***
Are people with lactose intolerance really sick? Why or why not?			
Could environmental differences (related to where people live) affect the ability of some groups of adults to digest lactose?			
Do you think we need to drink milk?			
Should dairy products have warning labels on them? Why or Why not?			

New Spoof of Milk Mustache Ad Spotlights Lactose Intolerance

Adapted from :http://www.pcrm.org/newsletter/jun05/milk_ad.html



Physicians' Committee for Responsible Medicine (PMRC) is launching a new campaign this month to draw attention to lactose intolerance and to find plaintiffs for an upcoming lawsuit against the dairy industry. A take-off on the milk mustache images, PCRM's ad reaches out to people with lactose intolerance in the Washington, D.C., area. The ad asks "Got Lactose Intolerance?" and explains that those who do may have grounds for a lawsuit.

About 75 percent of people worldwide are lactose intolerant, meaning they lose the ability to digest the milk sugar lactose after infancy. Symptoms can include nausea, cramps, bloating, gas, diarrhea, and other gastrointestinal distress.

This shift in enzyme activity is natural. According to the American Academy of Family Physicians, 60–80 percent of African Americans, 50–80 percent of Hispanic Americans, and at least 90 percent of Asian Americans are lactose intolerant. In 1988, the *American Journal of Clinical Nutrition* reported, "It rapidly became apparent that this pattern was the genetic norm, and that lactase activity was sustained only in a majority of adults whose origins were in Northern European or some Mediterranean populations." Almost 1,000 years ago dairy farming began in Northern Europe. It may have been an advantage for people in dairy farming areas of the world to continue to produce lactase in order to digest milk as an adult. It may have helped early Northern Europeans living in a dairy-farming environment to survive by providing a food source for adults as well as children.

Despite the prevalence of this condition, the dairy industry spends hundreds of millions of dollars each year trying to convince the American public that everyone needs milk. Many people are unaware of why they become sick after consuming dairy products, and may force themselves to consume them believing dairy products are necessary.

PCRM's public interest lawyers are planning a lawsuit against dairy producers on behalf of D.C. residents who are lactose intolerant. We will ask the court to mandate that milk products carry special labels warning about lactose intolerance.

Reading 2.5

Talking About Ethical Issues

Ethics provides a way for people to work through a problem that does not have a clear or right answer and determine what actions might be best for resolving that problem. When using ethics to determine the best action, you should consider all the information available including:

- What facts you know about the problem.
- What you don't know about the problem.
- Who is concerned about the problem (stakeholders)?
- What are concerns of the stakeholders?
- What possible solutions exist?

An example of an ethical problem is, —If killing is wrong… can you justify the death penalty? Can you kill in self-defense?

Can you think of other ethical problems?

Why do you think it is important to use ethics to help solve problems?

Often people will disagree when they use ethics to try to find the best actions to solve a problem. The following rules will help you work together when disagreements happen:

- Recognize and understand that there are multiple perspectives in the discussion. A discussion is not a competition with a winner and a loser.
- Be civil, polite, and courteous to other participants in the discussion.
- Communicate your ideas using relevant supporting evidence from what you have learned in class.
- Demonstrate appropriate use of vocabulary and scientific content. Make your scientific statements factual and complete. Connect your ideas to other scientific concepts and experiences that you have had.
- Show that you have an understanding of:
    - The stakeholders
    - The values and issues of the stakeholders
    - Alternative decisions that may be made

Are there other rules that you or your class think should be on this list? If so, list them below.

_____

_____

Activity 2.5A:<u>How Should People with Lactose Intolerance Be Treated?</u>

You have learned a lot about the protein lactase and how it works to break down lactose. You have also learned that people who do not make lactase are lactose intolerant. Currently, people who are lactose intolerant can buy special dairy products that already have lactase. However, dairy products with lactase are not available in all supermarkets and they are more expensive than milk, cheese, or ice cream without lactase. Some people who are lactose intolerant believe this is unfair.

One solution is to require that all dairy products have lactase added to them before they are sold.

Consider the following information as you use ethics to determine if this is the best action to solve this problem:

- Milk without lactase costs $2.50 a gallon.
- Milk with lactase costs $5.00 a gallon
- The cost of ice cream, cheese, yogurt, and many other products is dependent on the price of milk.
- 11% of the people in the United States are lactose intolerant.

Your class will work together using ethics to try to solve the problem of how to treat people with lactose intolerance fairly.

What do you think is the ethical problem?

_____
_____

What are relevant facts? (Consider what you read above and what you have learned about lactose intolerance.)

_____
_____
_____
_____
_____
_____

What are questions that are still unknown?

_____
_____
_____

Consider the following action:

To be fair to people that are lactose intolerant, all dairy products sold in the United States will have lactase added to them.

As your class uses ethics to decide if this is the best action, what stakeholders should you consider?

_____
_____
_____

What stakeholder did your teacher assign you?

_____

What do you think are the concerns of your stakeholder?

_____
_____
_____
_____
_____
_____
_____

How does your stakeholder feel about adding lactase to all dairy products? My stakeholder (circle one):

Completely agrees Agrees Disagrees Completely disagrees

Explain why your stakeholder feels this way:

_____
_____
_____
_____
_____

Who are the stakeholders?	What are the concerns of the stakeholders?

What are the possible solutions?

What solution do you think is the best solution? Why?

Activity 2.5B
Should decisions in our society be made based on how similar or different we are?

Many decisions in our society are already being made based on how similar or different we are. The fate of people accused of a crime, paternity cases, and treatments for diseases are all being decided through genetic testing. In this unit we have already looked at ethics related to the treatment of people with lactose intolerance. Genetic testing is a fairly new technology that has just recently become available and affordable. It is being considered for use in many new ways. Who will decide if these uses are ethical?

You will!

One of the main activities you will do in this unit is to research ethical dilemmas related to a specific disease or trait that you find interesting. You will use the ethical decision making framework to guide your research. You may present your findings as a poster, PowerPoint, brochure, commercial or talk show. However you choose to present your findings you must present an argument to support your position. Your presentation must include a **claim, evidence and reasoning** for the actions that you recommend.

On the next pages, you will find a collection of traits and diseases. Choose one of them to research. Each trait has a little bit of information about it and websites for further research. You can also use information found in your student reader.

When your project is completed, you will share it with your classmates. Try to make your project interesting to others around you. If you are a good artist, use your art abilities to make a colorful poster or brochure. In addition, make sure that you include enough information so that other people reading your poster or brochure will be able to understand more about the disease or trait. Rather than cutting and pasting information you find on the Internet directly to your poster or brochure, restate the information in your own words. Finally, have fun with it!

Genomics Projects

Each group should pick a topic:

- Skin color
- Lactose intolerance
- FH disease (high cholesterol and heart disease)
- Sickle cell anemia
- Other possible topics from list provided

Your group should focus on ethical issues brought up in the unit such as the use of skin bleaching products, paying more for lactose free products, advertising practices related to being lactose intolerant, and DNA testing for FH disease or sickle cell anemia. Issues can be presented in one of the following ways.

1. Poster or Powerpoint- Your group could do a poster/powerpoint presentation on an ethical issue related your topic.

2. Pamphlet- Your group could design a pamphlet from the perspective of a stakeholder in an ethical issue related to one of the above topics. You would present the pamphlet and the issues as if you were that stakeholder.

3. Demonstrate a lab related to your topic- browning apple lab- skin color, protein lab, lactase lab- lactose intolerance, DNA extraction lab.

4. Performance- Your group could write and perform a skit, poem or rap related to one of the above topics.

5. Art project- Your group could create an artwork, sculpture, painting related to your topic. You must also include a short report or poster providing information about the topic and art work.

Presentation Proposal

Group Names: _____

_____

Date: _____

Part I: Identify a trait or disease that your group is interested in researching from the list on pages. Pick a first and second choice.

1st choice_____

2nd choice _____

Why did you pick this topic? Why do you think this question is important?

_____

_____

_____

_____

_____

_____

Part II: What type of presentation would you like prepare? Choose from the list provided.

1st choice

_____

2nd choice

_____

Why did you pick this type of presentation?

_____

_____

_____

_____

_____

Guidance Questions-- TRAIT OR DISEASE

Trait or Disease Choice_____

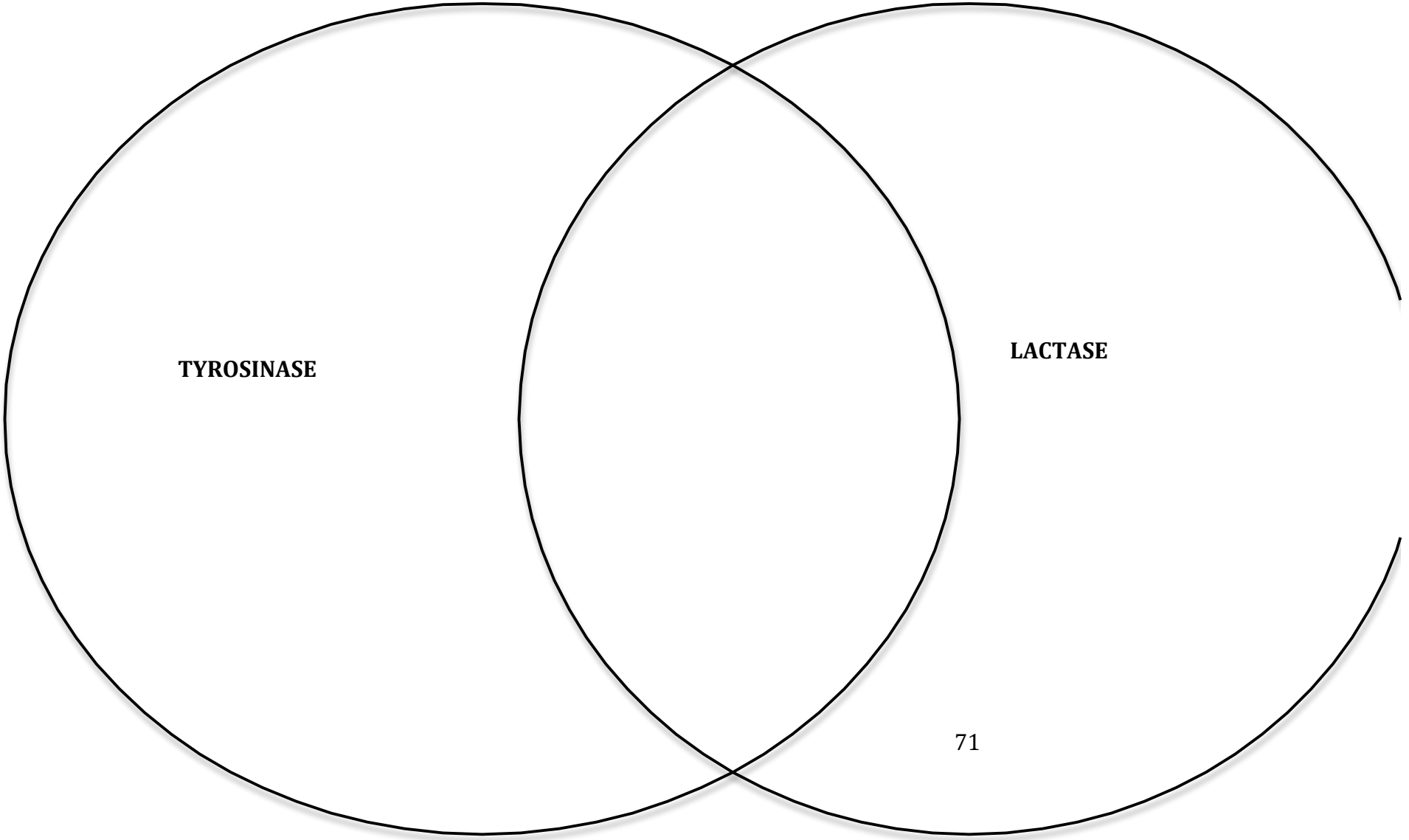
Using the provided websites or other websites you have found, answer the following questions:

- What effect does this trait have on the whole body?

- What effect does this trait have on the cells?

- What effect does this trait have on the proteins?

Compare tyrosinase and lactase by completing the Venn Diagram below



Reading 2.6 – Protein Shape is Dependent on Amino Acids

Before Reading

Try to come up with an answer for each question below BEFORE you read the text. Then look for confirmation of your predictions AS YOU READ.

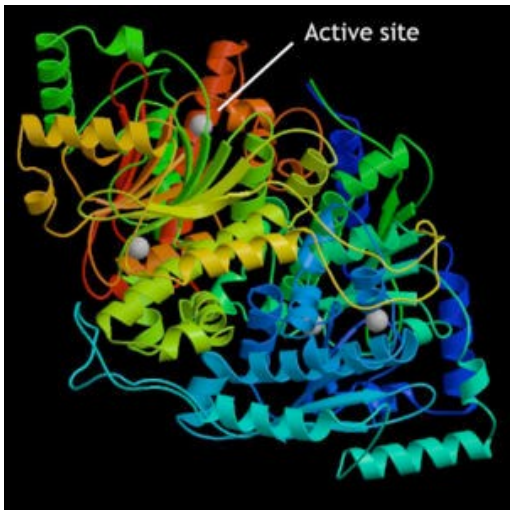
	Prediction	Answer
What type of shape do proteins have?		
What property of proteins largely determines the protein's function?		
What are the individual parts of a protein called?		
What determines the shape of proteins?		
How many different amino acids can make up a protein?		

What are proteins made of?

Every protein has a specific job to do in the body. But, what makes each protein capable of fulfilling its duties?

Each protein has a specific 3-dimensional shape because each protein has a specific job. In fact, the structure of the protein greatly determines its function. For example, the human body produces proteins to help stop bleeding. If there is a change to the shape of these proteins, the proteins are not able to stop bleeding effectively. This condition is called hemophilia, and people with this disorder experience uncontrollable bleeding even after

the most minor of injuries or even spontaneously just because their blood clotting protein has a slightly different shape. The unique 3-dimensional shape of proteins even makes it difficult to switch two proteins and expect them to work the same. For example, you cannot use tyrosinase to break down milk, nor can you use lactase to produce melanin because each enzyme is shaped uniquely for its job.



http://en.wikipedia.org/wiki/Enzyme#Enzyme_structure_and_mechanism

Take a look at the figure above. This is a 3-dimensional model of a protein. Notice how the protein bends, folds and twists around itself. The shape of a protein results from the different amino acids and how they interact. For example, the reason people with hemophilia experience a change in their blood clotting protein function is because there is a change in the amino acid sequence for that particular protein causing the protein to change shape.

Amino Acids: The Building Blocks of Proteins

Amino acids are the building blocks that make up a protein. Each amino acid is a molecule with a specific structure. There are 20 amino acids that link together to make up all proteins.

Within an amino acid, there is a **variable group,** represented as **R.** The variable group is the unique part of the amino acid that gives the amino acids certain properties. For example, some amino acids like to be surrounded by water while other amino acids do not.

After Reading:

1. Why can't different proteins perform the same function?

_____

_____

_____

2. How do proteins depend on amino acids for their shape?

_____

_____

Activity Sheet 2.6 – Modeling Proteins

Purpose: Scientists frequently use models for many different purposes. Today you will explore different types of models for proteins and amino acids. Since you might see proteins or amino acids represented by different models, it is important to understand each model's benefits and drawbacks.

Brainstorming

Why do you think scientist use models? For instance, you may have seen a model of an eyeball when you last visited the eye doctor.

_____

_____

_____

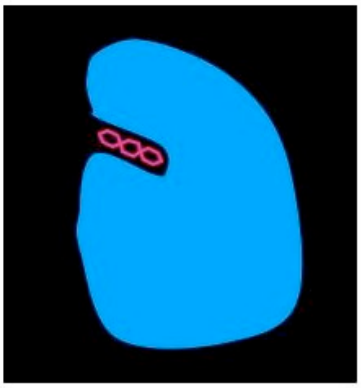
Using the ideas that your class came up with, do you think a model of proteins would be helpful? Why?

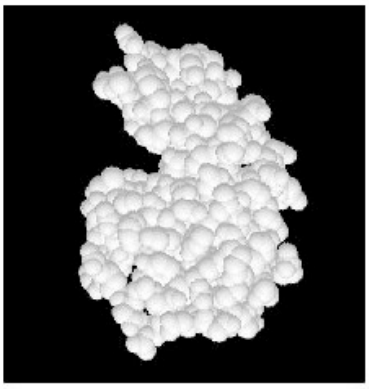
_____

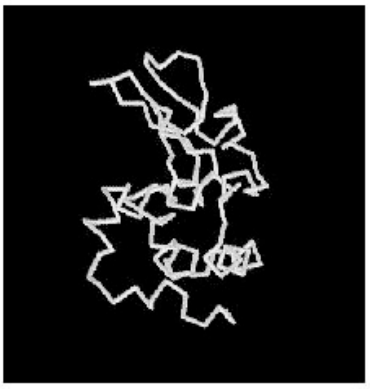
_____

_____

Look at the three images below. Each image is a different type of model for the same protein. Brainstorm what you think are the advantages of each model and write those predictions in the space provided below each image.







3 different models of the same protein

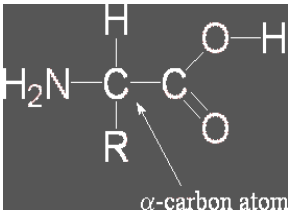
1. _____
_____
_____
_____

2. _____
_____
_____
_____

3. _____
_____
_____
_____

Reading 2.6B – Bonding of Amino Acids

Adapted from - http://www.scienceclarified.com/Al-As/Amino-Acid.html



Using the above picture, what do you think the line between each letter means?

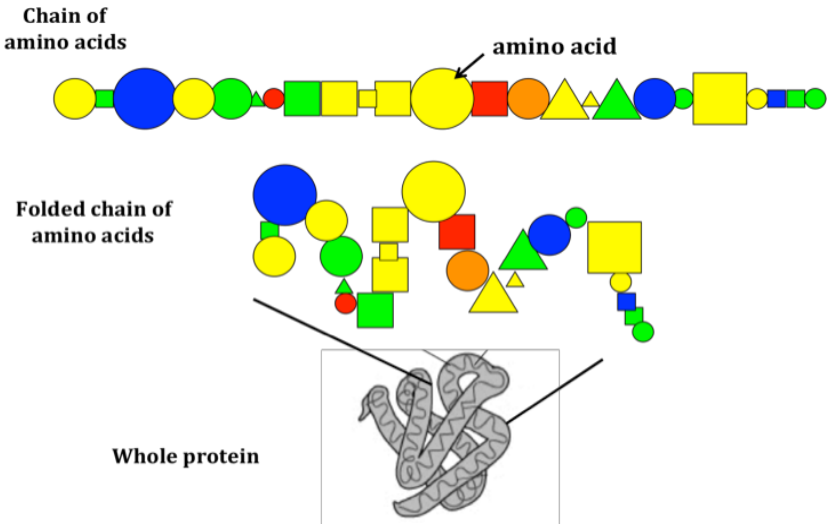
_____

_____

Amino acids are simple organic compounds made of carbon, hydrogen, oxygen, nitrogen, and, in a few cases, sulfur. Amino acids bond together to form protein molecules, the basic building blocks of all living things. Amino acids can vary widely. Only about 20 amino acids are common in humans and animals, with 2 additional ones present in a few animal species. There are over 100 lesser known amino acids found in other living organisms, particularly plants.

The first few amino acids were discovered in the early 1800s. Although scientists determined that amino acids were unique compounds, they were unsure of their exact significance. Scientists did not understand their importance in the formation of proteins—chemical compounds responsible for the structure and function of all cells—until the first part of the twentieth century.

An important characteristic of amino acids is their ability to join together in chains. The chains may contain as few as 2 or as many as 3,000 amino acid units. Amino acids become proteins when 50 or more are joined together in a chain.



The millions of different proteins in living things are formed by the bonding of only 20 amino acids. Like the 26 letters of the alphabet that join together to form different words, the 20 amino acids join together in different combinations and sequences to form a large variety of proteins. Even though about 10 or fewer letters form most words, 50 to more than 3,000 amino acids form proteins. Because each amino acid can be used many times along the chain and because there are no restrictions on the length of the chain, the number of possible combinations for the formation of protein is enormous.

The order of amino acids in the chain, however, is extremely important. Just as not all combinations of letters make sense, not all combinations of amino acids make functioning proteins. Some amino acid combinations can cause serious problems. For example, in reading 2.6, you read about how a change in the amino acid combination of a blood clotting protein caused uncontrollable bleeding spontaneously or after minor injuries.

After Reading Questions:

What are proteins made of? Explain why all of the pieces of a protein are not the same.

_____

_____

_____

_____

How are 20 different amino acids able to join together to make millions of different proteins?

_____

_____

_____

_____

If you were to describe a protein to a friend how would you describe it?

_____

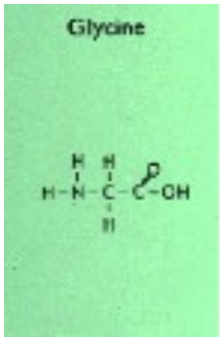
_____

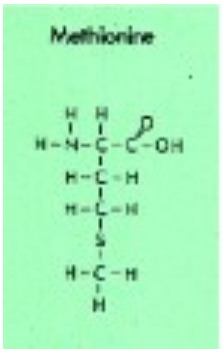
_____

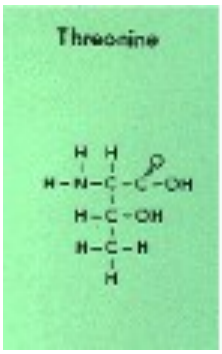
_____

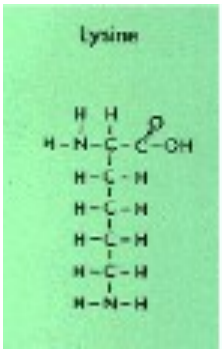
The Building Blocks of Proteins

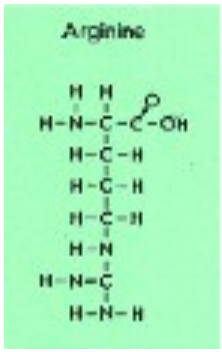
Purpose: Below you will see the chemical structure for 11 different amino acids. Part of each amino acid's structure is the same in all amino acids. What makes amino acids and ultimately proteins unique is there variable region. Today you will be identifying how similar and different these amino acids are and exploring the different ways that amino acids can be modeled.

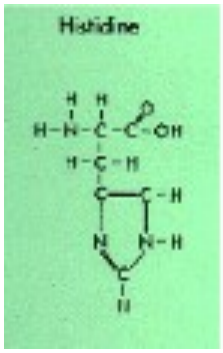


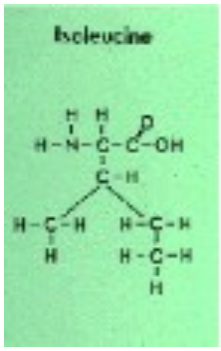


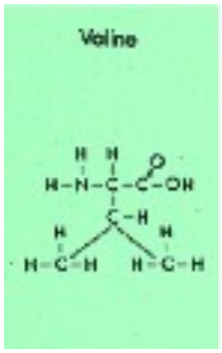


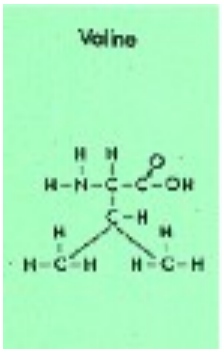


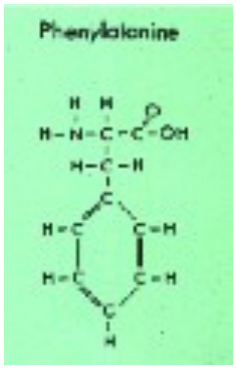


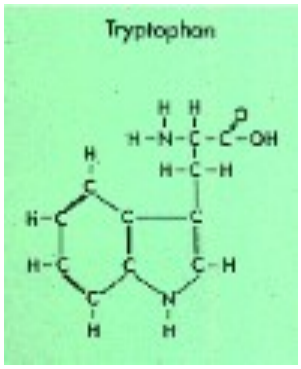








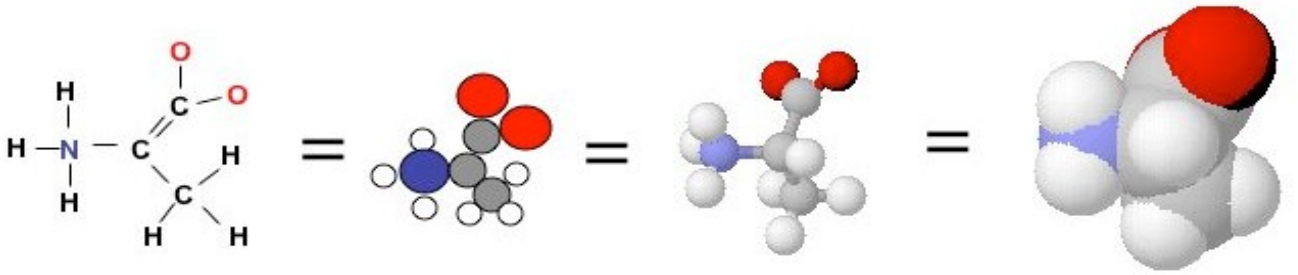




Write or draw what you see the above amino acid structures have in common in the space below.

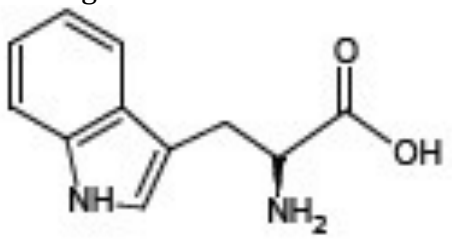
Amino Acid Models

Look at the four images below. Each image is a different type of model for the **same** amino acid. What do you think are the advantages of each model? Write down your ideas in the space provided below each image.



1. _____ 2. _____ 3. _____ 4. _____

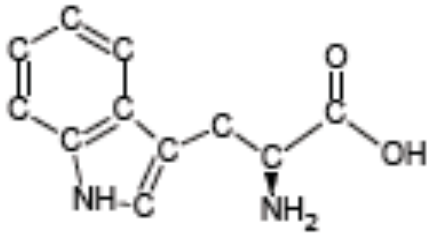
Below is another chemical structure model of a different amino acid. What are the advantages of both chemical structures?



What do you think are the pros and cons of this model?

_____

_____



What do you think are the pros and cons of this model?

_____

_____

Activity Sheet 2.7A - Building Proteins

Purpose: Proteins are essentially a long chain of amino acids that fold on top of themselves because of the properties of amino acids. Protein structure is important for protein function, so it is important to understand what influences protein structure. Today you will build a model of proteins to understand how amino acids influence protein structure.

Rules Amino Acids Follow When Folding:
- Positive and negative charged amino acids attract each other
- Amino acids with the same kind of charge repel each other
- Hydrophobic amino acids repel both water and other hydrophilic amino acids
- Hydrophilic amino acids attract water
- Hydrophobic amino acids attract each other

Thumb Tack Color Key	
Red	positive charge (+)
Yellow	negative charge (-)
Green	hydrophobic
Blue	hydrophilic

Below draw as best you can the first sequence of amino acids you folded.

After trading 2 red thumb tacks for 2 green thumb tacks and refolding the toober, draw below how your amino acid sequence has changed.

After trading 2 yellow thumb tacks for 2 blue thumb tacks and refolding the toober, draw below how your folded amino acid sequence has changed.

Describe how your initial amino acid sequence changed into the final toober you folded.

Activity Sheet 2.7B -- Building Proteins

Purpose: Today you will extend what you learned about how amino acid properties affect protein shape to a new case—that of the lactase protein and consider its function in influencing lactose tolerance by building a model of part of the lactase protein.

Introducing Activity

What is the function of lactase?

_____

_____

What are two functions of proteins like lactase?

1. _____

   _____

2. _____

   _____

Lactase Toober Model

Below is the amino acid sequence for the active site (or mouth) of the lactase protein/enzyme. Make a model of the amino acid sequence DIPIY ITENGVGLTN using the below amino acid key. You will also need to use the thumbtack color key from activity sheet 2.8A to determine what colors to use and to help you fold the toober.

DIPIY ITENGVGLTN (AA 1266-1280)

A = alanine -- Hydrophobic	C = cysteine -- Hydrophilic	D = aspartic acid -- Negative
E = glutamic acid -- Negative	F= phenylalanine-- Hydrophobic	G = glycine -- Hydrophilic
H = histidine -- Positive	I = isoleucine -- Hydrophobic	K = lysine -- Positive
L = leucine -- Hydrophobic	M = methionine -- Hydrophobic	N = asparagine -- Hydrophilic
P = proline -- Hydrophobic	Q = glutamine -- Hydrophilic	R – arginine -- Positive
S = serine -- Hydrophilic	T = threonine -- Hydrophilic	V = valine -- Hydrophobic
W = tryptophan -- Hydrophobic	Y = tyrosine -- Hydrophilic	

Positive = Red	Negative = Yellow	Hydrophobic = Green	Hydrophilic = Blue

Drawing the Lactase Active Site (Mouth)

After you make your model of lactase using the above amino acid sequence, draw a picture of the "lactase mouth" section of your model.

Change the color of one of your thumbtacks in the lactase active site (mouth). Did the shape of the active site change? If so, how might this affect this lactase's ability to breakdown lactose

What might happen to the person who has this abnormal protein?

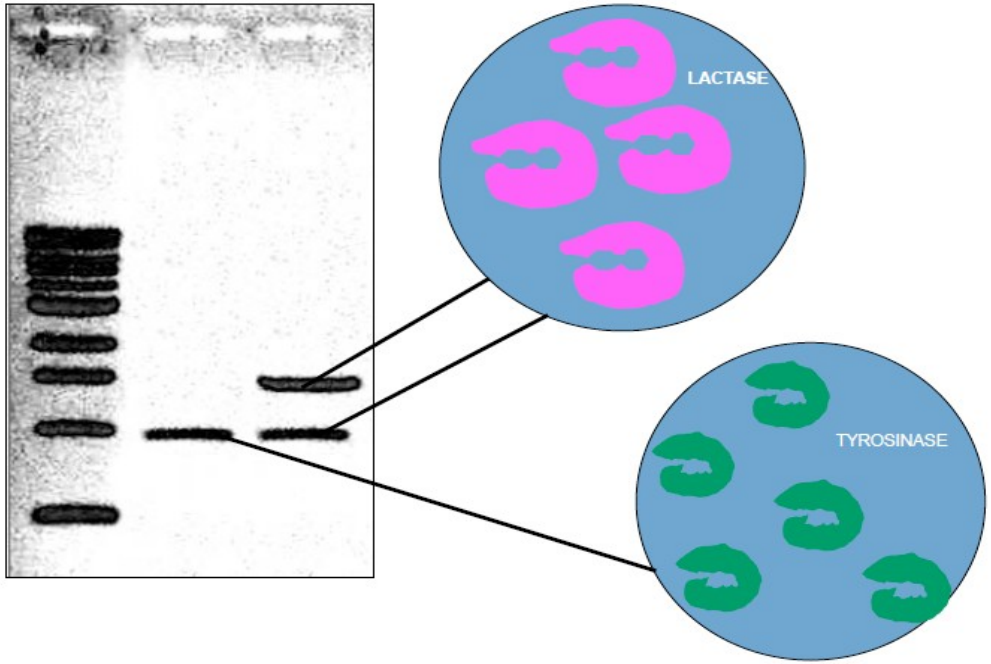
Check for Understanding

1. Based on what you learned from these activities with protein models, what general can you say about the amino acid sequence in a protein and its relationship to the protein's function?

2. If the protein structure is changed, could the function of the protein be affected?

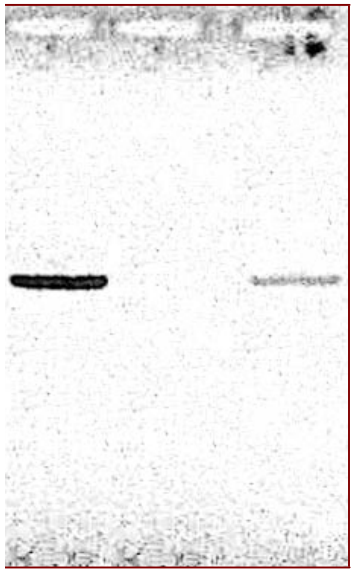
Activity Sheet 2.8

Gel Electrophoresis



Why do you think the column with lactase has two darkened rectangles while the column with tryosinase only has one darkened rectangle?

Who has lactose intolerance?



You are going to analyze some gel electrophoresis data. There are three samples on the gel to the left. The first sample came from a person who does not have lactose intolerance. The other two samples came from people who don't know whether or not they are lactose tolerance. Diagnose person 2 and person 3 and write a scientific explanation indicating whether or not you think the person should drink milk.

Person #2: _____

_____

_____

_____

_____

_____

_____

Person #3: _____

_____

_____

_____

_____

_____

Trait Descriptions and Websites

Androgen Insensitivity Syndrome

AIS, also known as Androgen Resistance Syndrome, results from mutations in the gene that codes for a protein that detects the presence of the hormone androgen. When the protein is missing, it causes sexual differentiation disorders. In some cases a person with AIS might have a female body, but be missing a uterus, fallopian tubes or ovaries. In another type, the person may have a male body but reduced sperm production and fertility.
http://en.wikipedia.org/wiki/Androgen_insensitivity_syndrome
http://www.rch.org.au/publications/CAIS.pdf
http://www.aissg.org/21_OVERVIEW.HTM
http://www.geneclinics.org/profiles/androgen/details.html

Cystic fibrosis

Cystic Fibrosis (CF) is a condition in which there is an overproduction of mucus in the lungs, often leading to lung infections, scarring, and eventually death. CF is one of the most common inherited diseases.

http://en.wikipedia.org/wiki/Cystic_fibrosis
http://www.marchofdimes.com/pnhec/4439_1213.asp
http://www3.nbnet.nb.ca/normap/CF.htm
http://www.cftrust.org.uk/aboutcf/publications/booklets/

Diabetes

Diabetes is a disorder that is characterized by hyperglycemia (high blood sugar). There are three types: type 1, which is due to the autoimmune destruction of insulin producing cells; type 2, which is due to insulin-resistance; and gestational diabetes, which is like type 2 but occurs during pregnancy. Although genes play a role in diabetes, scientists have found that the environment has a role in causing diabetes as well.

http://en.wikipedia.org/wiki/Diabetes
http://www.diabetes.org/about-diabetes.jsp
http://darwin.nmsu.edu/~molbio/diabetes/disease.html
http://diabetes.webmd.com/default.htm

Familial Hypercholesterolaemia

People with familial hypercholesterolaemia (FH) have a lack of the protein that remove cholesterol from the blood. This is caused by a change in the gene that codes for this protein. Symptoms include the early development of hardening of the arteries and heart disease.

www.netdoctor.co.uk/diseases/facts/familialhypercholesterolaemia.htm

www.cdc.gov/features/HeartDisease/

www.wvdhhr.org/bph/oehp/hp/cardio/fh.htm

www.medped.org/MEDPED-What-is-FH.html

Hemophilia

Hemophilia is a bleeding disorder caused by changes in the genes that code for proteins responsible for blood clotting. This disorder can cause problems with bleeding from prolonged bleeding after an injury or surgery to spontaneous bleeding into joints, muscles or the brain. Mutations in these genes can cause a reduction in the production of the proteins needed for blood clotting or can cause abnormal proteins to be produced.

www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001564/
www.nhlbi.nih.gov/health/dci/Diseases/hemophilia/hemophilia_what.html
www.kidshealth.org/parent/medical/heart/hemophilia.html
www.emedicinehealth.com/hemophilia/article_em.htm

Lactose Intolerance

People with Lactose intolerance are unable to digest a type of sugar found in milk. They lack the protein necessary to digest this sugar, which can cause bloating, gas and abdominal cramps.

www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001321/
www.kidshealth.org/kid/health_problems/allergiesimmune/lactose.html
www.nichd.nih.gov/health/topics/lactose_intolerance.cfm

Parkinson‘s Disease

Parkinson‘s Disease is a brain disorder that can cause tremors, stiffness, difficulty with balance, and slowness of movement. It occurs when 80% of the cells that produce dopamine (which is vital in keeping body/muscle movements coordinated and smooth) are damaged. Second to Alzheimer‘s, it is the most common neurodegenerative disease.

http://www.parkinson.org http://en.wikipedia.org/wiki/Parkinson%27s#Genetic
http://www.pdf.org/AboutPD/
http://www.nlm.nih.gov/medlineplus/tutorials/parkinsonsdisease/htm/index.htm
http://www.apdaparkinson.org/user/AboutParkinson.asp

PKU

Phenylketonuria is a genetic condition resulting from a defective gene that normally codes for the protein that converts the amino acid phenylalanine to tyrosine. This often results in severe mental retardation, seizures, and hyperactivity, among other symptoms.

http://en.wikipedia.org/wiki/Phenylketonuria
http://www.ygyh.org/pku/whatisit.htm
http://www.medschool.lsuhsc.edu/genetics_center/louisiana/article_PKU.htm
http://www.peds.umn.edu/pku/Basics.html

Polycystic kidney disease

Polycystic kidney disease is a genetic disease in which the kidneys develop multiple cysts. It can also damage other organs such as the liver, pancreas, heart and brain. The disease often leads to the complete failure of the kidneys.

http://en.wikipedia.org/wiki/Polycystic_kidney_disease
http://www.ygyh.org/pkd/whatisit.htm
http://www.kidney.org/atoz/pdf/polycystic.pdf
http://www.pkdcure.org

Sickle Cell Disease

This disease is in which the body produces sickle shaped red blood cells. Symptoms consist of painful episodes that affect the bones in the back, the chest, and the long bones. It is caused by a mutation in the gene that codes for the protein that carries the oxygen in red blood cells. This change in the protein causes the red blood cell to take on the crescent shape.
www.genome.gov/10001219
www.ncbi.nlm.nih.gov/pubmedhealth/PMH0001554/
www.nhlbi.nih.gov/health/dci/Diseases/Sca/SCA_WhatIs.html
www.kidshealth.org/teen/diseases_conditions/blood/sickle_cell_anemia.html

Tay-Sachs Disease

Tay-Sachs disease usually is seen in infants between 3 and 6 months old. It occurs because a protein related to the brain and spinal cord function is not being produced. This protein breaks down toxic substances.Tay-Sachs can affect motor skills, hearing, vision, and intellectual ability.

www.ghr.nlm.nih.gov/condition/tay-sachs-disease
www.kidshealth.org/parent/medical/genetic/tay_sachs.html
www.nlm.nih.gov/medlineplus/taysachsdisease.html
www.ygyh.org/tay/whatisit.htm

Thalassemia

Thalassemia is a genetic disorder in which the patient has red blood cells that are easily damaged and therefore die quickly. The disease can be treated by regular blood infusions.

http://en.wikipedia.org/wiki/Thalassemia
http://www.thalassemia.com
http://sickle.bwh.harvard.edu/menu_thal.html
http://www.noah-health.org/en/blood/thalassemia/

Werner syndrome

Werner Syndrome is a genetic disorder in which the patient starts aging during puberty. Following puberty, they age rapidly, so that by the time they reach age 40, they often appear as though they are several decades older.

http://en.wikipedia.org/wiki/Werner_syndrome

http://children.webmd.com/Werner-Syndrome
http://www.madisonsfoundation.org/content/3/1/display.asp?did=426
http://ghr.nlm.nih.gov/condition=wernersyndrome

Xeroderma pigmentosum

Xeroderma pigmentosum, or XP, is a genetic disorder is which the patient is particularly sensitive to UV light. The sensitivity to light leads to skin cancer at a young age. Patients with XP often find it necessary to avoid sunlight.

http://en.wikipedia.org/wiki/Xeroderma_pigmentosum
http://www.xps.org/student_tips.htm
http://www.rare-disorders.com/xeroderma-pigmentosum.html
http://www.webmd.com/skin-problems-and-treatments/Xeroderma-Pigmentosum-10968

Learning Set 3:

How Does Our DNA Make Us SIMILAR or DIFFERENT?

Reading 3.1 – The Medical Case: Familial Hypercholesterolemia

Before Reading

Try to come up with an answer for each question below BEFORE you read the text. Then look for confirmation of your predictions AS YOU READ.

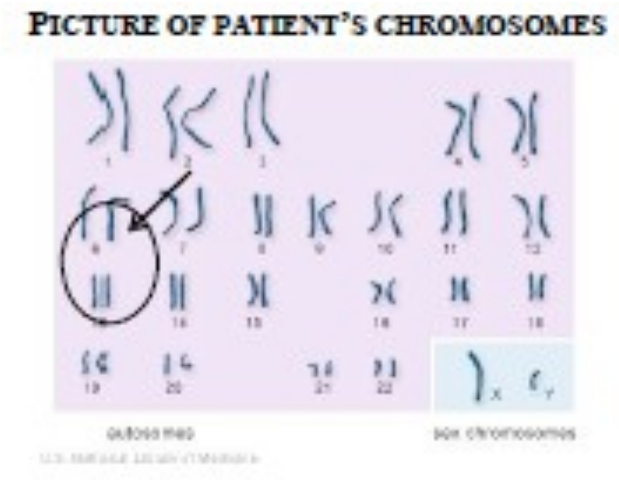
	Prediction	Answer
What are chromosomes made of?		
Could a problem with a chromosome cause a change in how a protein acts?		
How are chromosomes, DNA and proteins related to each other?		
Do you think obesity can be caused by a problem with a person's DNA? Why?		

The Medical Case: Familial Hypercholesterolemia

Rachel was excited to start her first pre-med hospital program at her undergraduate university. She always dreamt of becoming a doctor, but since she was only 18 and inexperienced in medicine, she knew that she had a challenge ahead of her.

Doctor Lewis was going to be Rachel's advising doctor. Dr. Lewis invited Rachel into his office and handed Rachel a folder. Inside the folder were descriptions and pictures of several patients. There was one description that particularly stood out to Rachel. Before she read the description, she looked at pictures of different sections of the patient's body.

These pictures showed fatty deposits that looked like bulges on the arms, knees, elbows and toes of the patient like the picture to the right. She tried to think of what this patient was suffering from. As she continued to read, she noticed a picture that showed the patients chromosomes. One of his chromosomes, chromosome 19, had an arrow pointing at one spot on the chromosome. This arrow indicated something was different at this spot on chromosome 19. Rachel remembered that chromosomes are made up of tightly coiled DNA.



Rachel concluded that the patient was having a problem with his DNA. But Rachel still could not understand how having problems with DNA could lead to such a severe illness. She continued to read and look at pictures in the folder. She shuffled through the papers. Then, she found a paper that talked about LDL receptor proteins. It said that problems with this protein could cause a disease called **Familial Hypercholesterolemia.** Rachel stopped to think.

Rachel asked Dr. Lewis, "Doctor, LDL receptors are proteins right?"

Dr. Lewis replied, "Yes they are Rachel. Who's case are you looking at?"

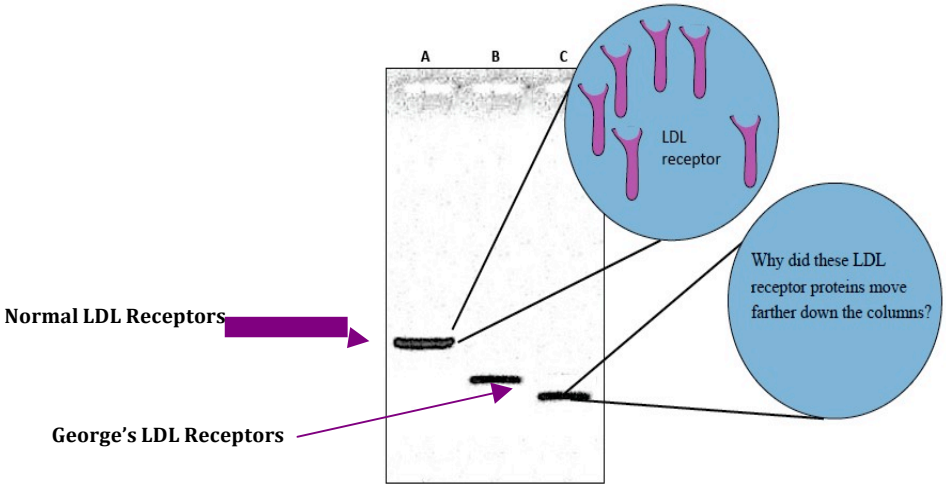
Rachel answered, "This patient, George, has bulges on his toes, and problems with his DNA. Do you think something could also be wrong with the LDL receptor proteins in his body?"

Dr. Lewis took the file from Rachel and began to look at it. He saw a report with Gel Electrophoresis data in George's file.

Activity 3.1

Analyzing Gel Electrophoresis Data: LDL Receptor Protein

Help Rachel and Dr. Lewis with their diagnosis. Analyze the data below to determine if Rachel is correct in thinking that there might be something wrong with the patient's proteins.



1. What do you think the above gel electrophoresis data tell you about the 3 different LDL receptor proteins added to the gel?

_____

_____

_____

2. Why do you think the proteins in columns B and C moved down the columns more than the protein put in column A?

_____

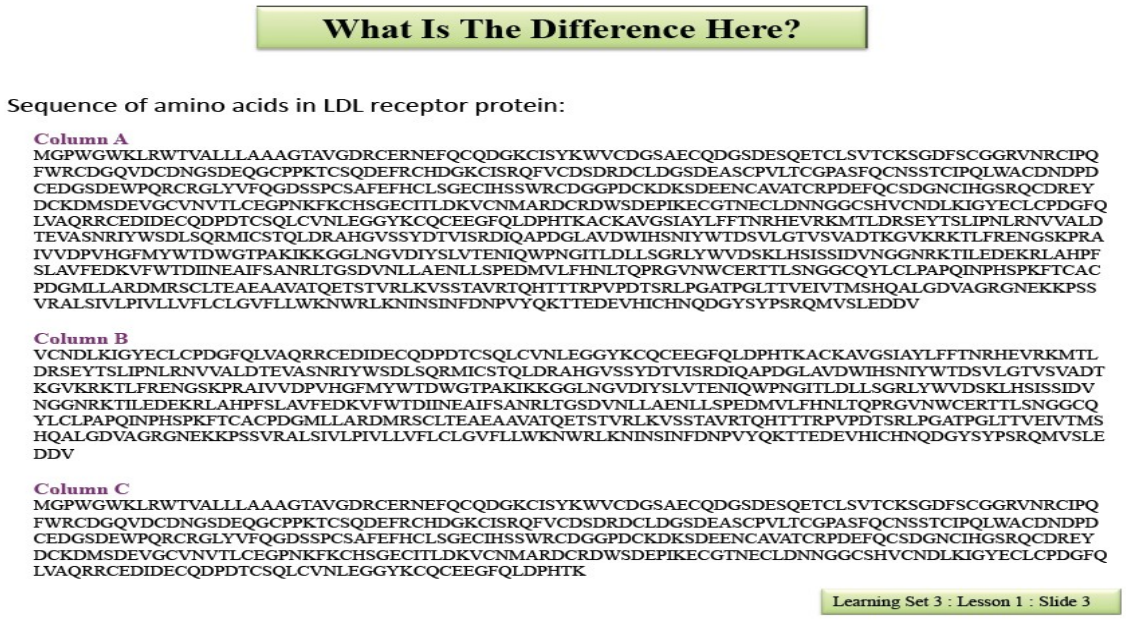
_____

_____

3. Why do you think column C has George's protein?

_____

_____



4. Write a scientific explanation why George's LDL receptor protein moved the farthest down the gel column.

_____

_____

_____

_____

_____

5. Write a scientific explanation telling if different amino acid sequences can result in the same protein?

_____

_____

_____

_____

_____

6. What led Rachel to realize that the patient George had a problem with his DNA?

_____

_____

_____

The Medical Case: Familial Hypercholesterolemia – part 2

Dr. Lewis looked closely at the medical file. He said, "Oh yes. George is very young. He had a heart attack when he was only 25. We have been treating him for familial hypercholesterolemia ever since. I wish we knew about it earlier. We might have been able to treat him sooner. Let's go talk to the patient himself. I think it will make more sense to you then."

Dr. Lewis and Rachel walked into George's hospital room. Rachel stopped at the door when she saw the patient.

Rachel exclaimed to Dr. Lewis, "George is obese. This is a good indication that he cannot remove cholesterol from his blood. Since the LDL receptors are unable to remove cholesterol from the blood, George's body has more fat deposits than usual. But what does that say about his DNA?"

Dr. Lewis and Rachel returned to the office.

Dr. Lewis asked Rachel, "What did you think of George? Did you notice that his obesity is a result of his illness?"

Rachel thought hard, and then replied, "Yes, George cannot make LDL receptors in his body. So he will have more fat deposits, and that's why he is obese."

Doctor Lewis helped Rachel by asking, "And if the gene that is responsible for making LDL receptors is defective, how does that affect his health condition?"

Rachel paused for a moment, and then answered, "DNA is made up of genes and genes holds instructions for proteins. And since the LDL receptor is a protein, a chromosome must contain the information that codes for it. A problem with the protein or part of the chromosome must be what causes familial hypercholesterolemia."

"Very good", replied Doctor Lewis, "When we look at patients, it is important to remember that some of their medical conditions are due to DNA disorders. In this case, George has a problem with part of his chromosome 19, which caused a problem with his proteins, and that is why he is ill."

At the end of the day, Rachel went home. She could not stop thinking about the patient with familial hypercholesterolemia.

After Reading Questions

In "The Medical Case: Familial Hypercholesterolemia", Rachel struggled to understand her patient's medical condition. Answer these questions that helped Rachel diagnose her patient.

1. From the reading, what do you think are some of the physical signs of familial hypercholesterolemia?

_____

_____

_____

2. Do you think these differences in amino acid sequence might affect the LDL receptor's ability to function properly? Was Rachel correct in linking George's symptoms to the LDL receptor's in his body? What is your evidence?

_____

_____

_____

_____

3. Using the case you just read, explain how DNA and proteins are related?

_____

_____

_____

_____

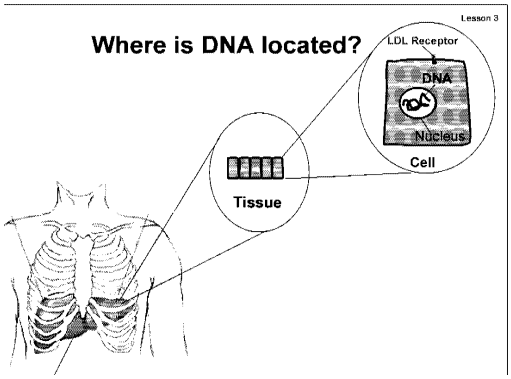
4. Using the LDL receptor protein gel electrophoresis data, the amino acid sequences for the three LDL receptor proteins that were placed in the gel and the information from this reading, write a scientific explanation stating why George has familial hypercholesterolemia.

Activity Sheet 3.2a

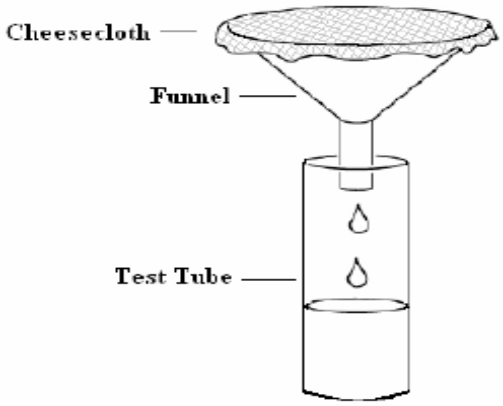
Purpose: Today you will extract DNA from a strawberry and explore how and where DNA is stored in living things.

Do all cells contain DNA? What is your reasoning?

_____
_____
_____
_____
_____



Materials (per group)
1 - Ziploc Freezer bag
1 – strawberry
10 mL - DNA extraction buffer in a small plastic cup
Small funnel fitted with cheesecloth
50 mL test tube
Popsicle stick
20 mL - Cold ethanol

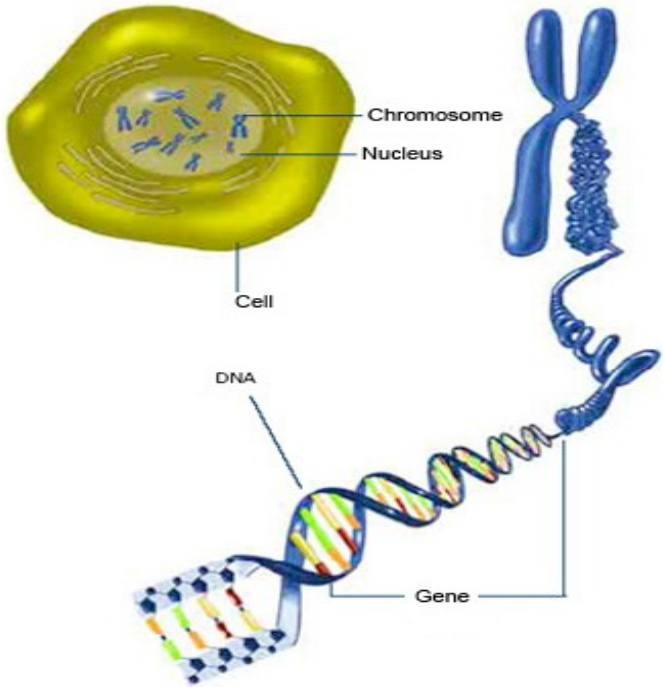


Procedure:

1. Place one strawberry in the Ziploc bag.
2. Smash the strawberry using your fist/fingers for 2 minutes. (Be careful not to break the bag!)
3. Add 10 mL of extraction buffer to the bag.
4. Kneed/smash the strawberry in the bag again for 1 minute.
5. Assemble your filtration apparatus as shown above.
6. Pour the strawberry slurry into the filtration apparatus and let all of the slurry completely filter through into your test tube.
7. Slowly pour cold ethanol into the tube.
8. Observe what is happening inside the test tube. What do you think the string-like material in your test tube is?
9. Dip the popsicle stick into the tube where the strawberry extract and ethanol layers come into contact with each other. You should see thin string-like material. Try to gently twirl one of the strings around your popsicle stick, so that you can gently pull it out of the test tube. DNA is extremely fragile, so it is extremely important for you to be gentle when trying to remove the DNA string-like material from your test tube.

Describe what the DNA that you extracted from the strawberry looked like.

Do you have any ideas why you can see DNA without a microscope, but you cannot see a cell (which is where DNA is found) without the aid of a microscope?



What does the above picture tell you about DNA, chromosomes and cells?

Reading 3.2– Genes and Gregor Mendel

While reading, write down any questions you have about how genes, DNA and proteins relate to each other in the chart on the next page. Then, you and your group will discuss how your questions relate to the driving question and add this to your chart.

Genes and Gregor Mendel

In the past, learning about genes meant learning about Gregor Mendel, one of the first scientists to study genes. He studied genes by observing plants and the characteristics that were passed on from parent to offspring. Gregor Mendel might have been surprised to learn that most genes contain nothing more than instructions for assembling proteins. He might have asked what proteins could possibly have to do with the color of a flower, the shape of a leaf, a human blood type, or the sex of a newborn baby. The answer is that proteins have everything to do with these things.

Many proteins can regulate chemical reactions. For example, a gene that codes for a protein to produce melanin can control skin color. Another gene produces a protein specialized for the production of white blood cells to fight off diseases.

The gene combinations that might result when two individuals have offspring can be predicted. The genes of the offspring can be compared to determine the genetic variations that will result. Those gene combinations determine the kinds of proteins that are produced, and those proteins determine the characteristics that make people similar and different from each other.

In this unit, you will learn how genes code for proteins in cells, and how those proteins contribute to the similarities and differences you may encounter in people. In lesson 1, you read that genes are actually small pieces of DNA. In the next activity, you will build a model of DNA. This model is a representation of what DNA looks like in your cells. Pay close attention to the different bases, how they pair with each other and the shape of the DNA molecule.

Question	**How does this relate to the DQ?**

Activity Sheet 3.3A

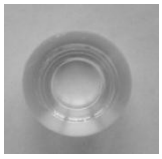
Purpose: Today you are going to build a model of DNA, so you can explore the molecules that make up DNA.

Assembling a DNA model

Make sure you have contains the following materials:

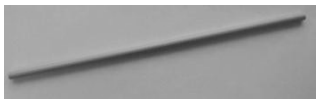
6 T (orange)
6 A (blue)
6 G (green)
6 C (yellow)

1 base
(light brown)

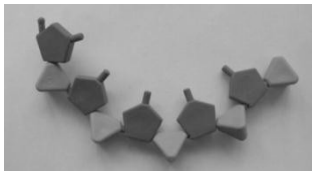


12 spacers
(transparent)

1 cap (white)

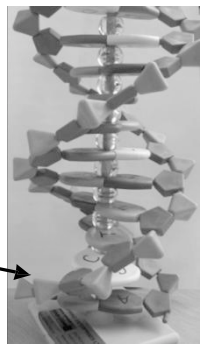


1 rod (grey)
of alternating deoxyribose (red)
and phosphate (purple)



2 side chains composed

1. Assemble the model **stand** by pushing the **rod** firmly into the hole in the **base**.
2. Join the DNA bases (the pieces labeled A, T, C, and G). Not all of the pieces will fit together.
3. Slide the paired DNA bases onto the rod in whatever order you choose, with a spacer in between each pair.
4. Attach the side chain to the paired DNA bases, by inserting the deoxyriboses into the holes on the outside of the DNA bases.
5. The model is finished when the second chain has been attached and the double-helix completed. Put the white cap on the top to keep the DNA bases from falling off.



Do you notice any patterns about how the DNA bases attach to each other? What is the sequence of the DNA bases in your model?

Reading 3.3 – Structure of DNA

In lesson1 you discussed genes. In order to understand how genes work, it is important to understand where genes come from. Genes are found in **DNA- deoxyribose nucleic acid**. While reading, think back to the DNA model that you built in class.

DNA is a long molecule made up of units called nucleotides. Nucleotides are made up of 3 parts: a sugar called deoxyribose, a phosphate group and a chemical base. There are 4 kinds of bases in DNA. These bases are adenine, guanine, cytosine and thymine. The backbone of the DNA molecule is made up of the sugars and the phosphate groups. The interior of the DNA molecule is made up of the bases. The bases pair in a very specific way and these pairs are referred to as base pairs. Adenine pairs with Thymine and Cytosine pairs with Guanine; for every Adenine base, there is one Thymine base and for every Cytosine base, there is one Guanine base. The four different bases could be strung together in many different ways, making it possible to have many combinations of bases.

Eventually, scientists found that DNA was shaped into something called a "double helix". Two strands of DNA lined up next to each other, then twisted into what looks like a spiral staircase or a twisted ladder. In this model, the backbone is on the outside, and the bases are in the middle. The discovery of the double helix structure of DNA is to science what the Mona Lisa is to painting. It‘s been called the single biggest discovery of all time. But it wasn‘t just stumbled upon- it was a race. Specifically, it was a race between two teams of young scientists working in Britain and California. Rosalind Franklin and Maurice Wilkins were trying to identify the structure by studying X-ray diffractions of the DNA molecule. But Jim Watson and Francis Crick studied a little bit of everything. A few have gone so far as to accuse Watson and stealing Franklin’s x-ray work.
In any case, Watson and Crick’s inquisitive working style ultimately allowed them to determine the DNA structure first, in 1953 – an achievement that led to their Nobel Prize in 1962. Meanwhile, Franklin passed away in 1958 from cancer.
Think back to what you learned about genes. You read and discussed that genes were instructions for making proteins. DNA is the important part of genes. DNA gives the letter code for which amino acids are to be made. When the DNA bases are in a certain order, they have a specific meaning as an amino acid sequence. Amino acid sequences come from the genes, and the genes are made up of DNA bases that give specific instructions.

What combinations do DNA bases make? (How do they pair?)

_____
_____

How are DNA base pairs similar to an amino acid sequence?

_____
_____
_____
_____

Activity Sheet 3.3B

Building the DNA Structure for the LDL Receptor Protein

Purpose: Yesterday you explored the structure of a DNA model with a random sequence. But to be able to find out if DNA holds instructions for building proteins, we need to build the DNA structure for a real protein to see what it tells us. Today you are going to build a model for a portion of a specific piece of DNA that codes for the LDL receptor protein.

To answer the questions below, you may want to refer back to Reading 3.2b or look at the DNA model you made yesterday.

What are the 4 chemical bases found in DNA? _____

_____

How do these chemical bases pair up?

_____

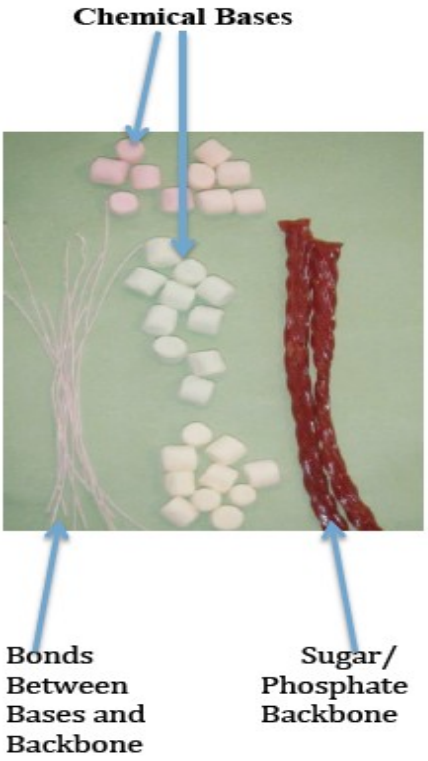
_____

Below is one of the DNA strands of the double-helix for the LDL receptor protein. In the next column, fill in the chemical base that pairs with the given chemical bases. These two strands together create the double-helix of a piece of DNA. You will use these two strands to build your DNA structure today.

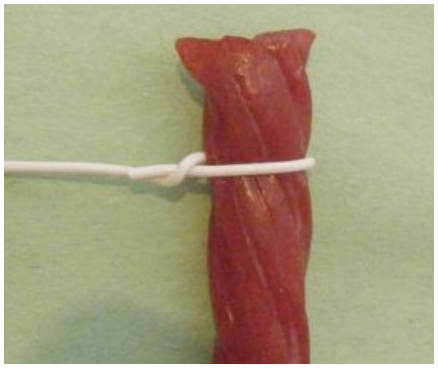
Given DNA Strand	Matching DNA Strand	RNA
T		
G		
G		
C		
G		
C		
T		
G		
T		
G		
DNA Base Pairs		

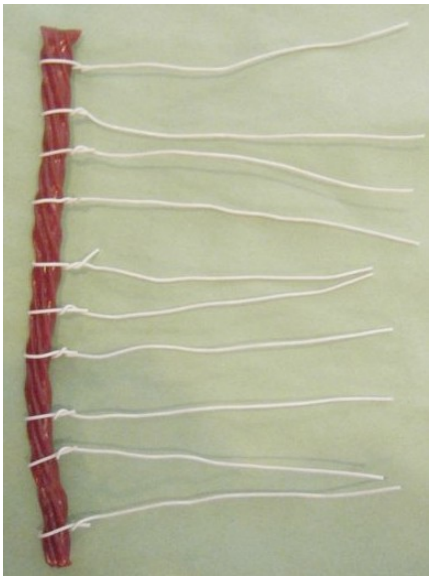
Materials:
5 Orange Marshmallows
10 Green Marshmallows
10 Yellow Marshmallows
5 Pink Marshmallows
2 pieces of Licorice
10 - 15 cm pieces of bell wire per group

* You will have a couple extra marshmallows of each color just in case you lose one. It is ok if you have 2 or 3 of each color leftover when you are finished.



1. Attach your wire to one of your pieces of licorice. See pictures below.

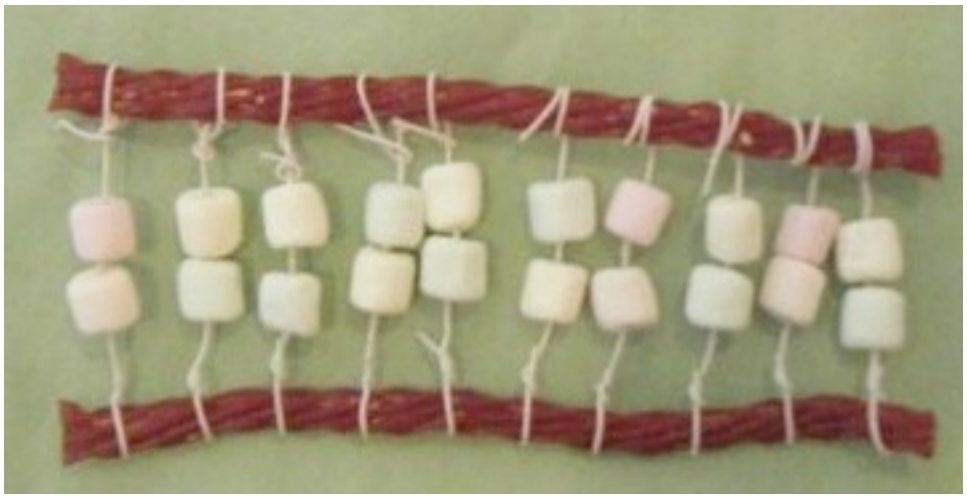




2. Using the LDL receptor protein DNA strand that you worked on above and the Marshmallow Color Key found below, start attaching the base pair marshmallows to your wire. See picture below.

Marshmallow Color	Chemical Base
Orange	*Thymine (T)*
Green	*Guanine (G)*
Yellow	*Cytosine (C)*
Pink	*Adenine (A)*

3. Attach the other piece of licorice so your model looks like a ladder like in the picture below.



4. Carefully, twist your DNA model so that it looks like the double-helix model you made yesterday.

How does today's model of the DNA structure compare to yesterday's?

_____
_____
_____

What are the pros and cons of building models of DNA?

_____
_____

Activity 3.4a

Review:
We learned in a previous lesson that there is a relationship between proteins and genes. Describe the relationship between proteins and genes.

_____

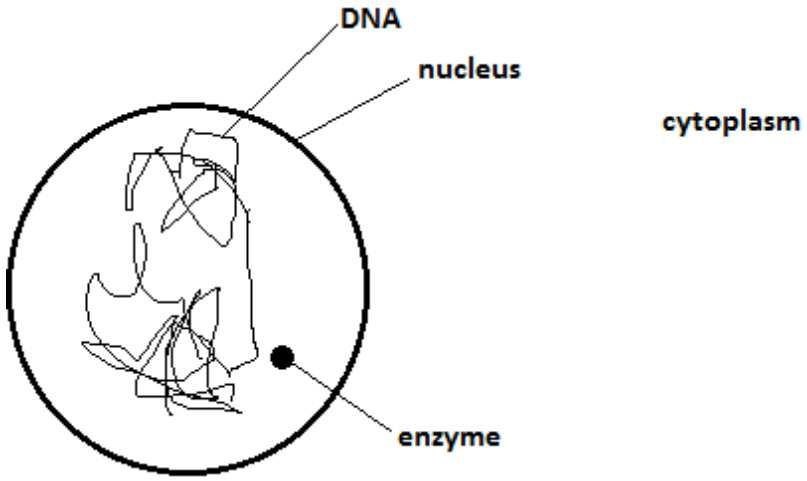
_____

_____

From DNA to Proteins: A Quick Overview
In the last lesson you learned that a gene is a segment or a piece of DNA. You also learned that each gene codes, or contains instructions for, the making of a protein. There are many steps involved in getting the codes, or instructions, from our DNA to make the proteins our bodies need to perform the many activities we learned about. In this activity you will first get a brief overview of the process of getting the instructions from our DNA to make proteins.

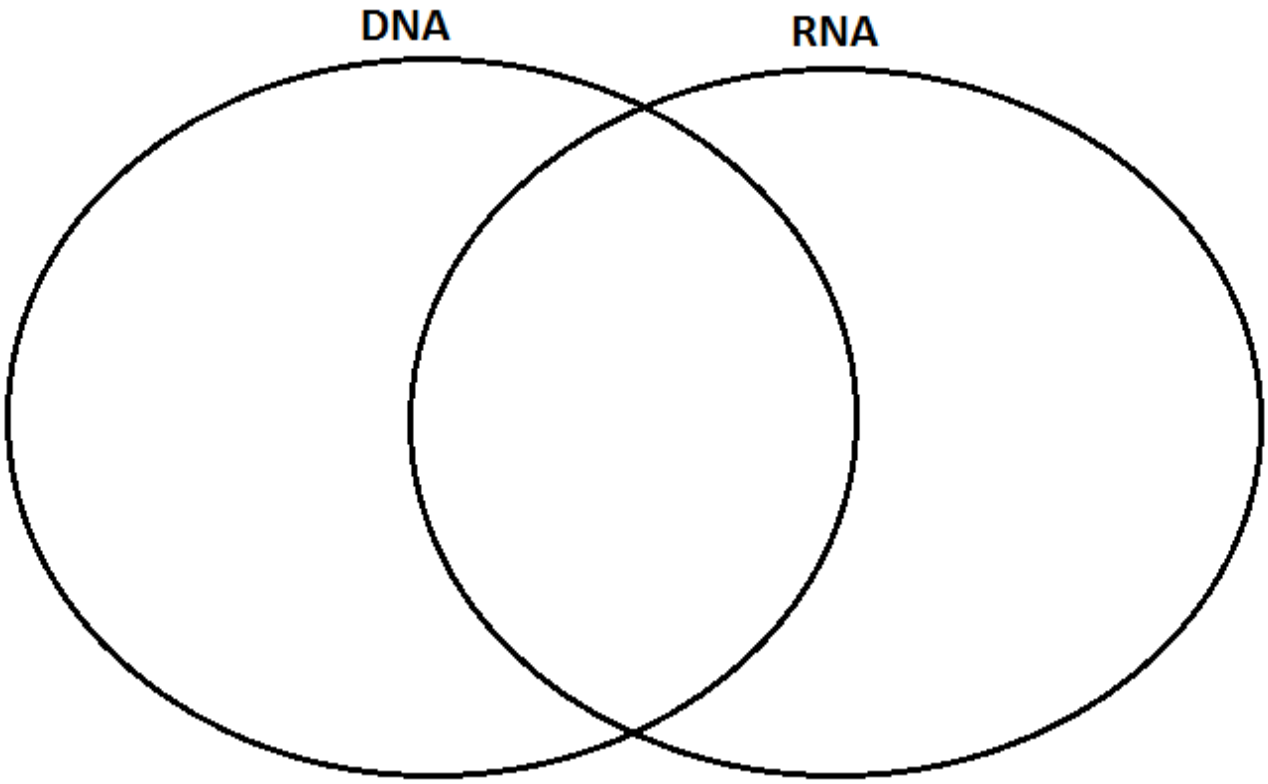
You will watch an animation. Listen carefully to your teacher as you watch the animation and draw out a "timeline," or the basic steps of the process in the boxes below. Don't forget to label each thing you draw, and write a brief statement about what your picture shows. The first one has been done for you.

The first step happens in the nucleus of the cell.



From DNA to Proteins: An overview, cont.

Venn Diagram comparing DNA and RNA



Check for Understanding:

1) Using the pictures you drew and what we discussed as a class, describe the process of transcription in your own words. Be sure to include specific cellular vocabulary and include the location where transcription takes place.

2) Describe the process of translation in your own words. Be sure to include specific cellular vocabulary and include the location where translation takes place.

3) What does it mean to say that DNA contains the instructions for making proteins?

Activity 3.4b

Review:

1) What did we learn the letters A, T, G, and C stand for?

---

2) What do these letters represent?

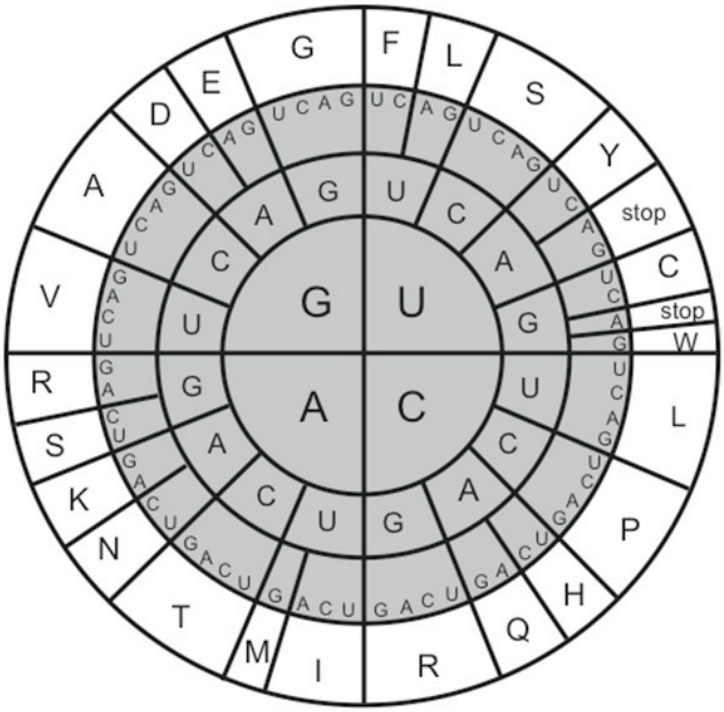
The specifics of transcription and translation

In this activity you will work with your teacher to learn and take notes on the specific processes involved in making proteins: transcription and translation.

As your teacher models how a gene is transcribed and translated you should write a brief statement to describe what happens in each step.

Transcription STEP 1:
STEP 2:
STEP 3:
Translation STEP 4:
STEP 5:

Amino Acid Chart



AMINO ACIDS		
A = alanine	C = cysteine	D = aspartic acid
E = glutamic acid	F = phenylalanine	G = glycine
H = histidine	I = isoleucine	K = lysine
L = leucine	M = methionine	N = asparagine
P = protine	Q = glutamine	R - arginine
S = serine	T = threonine	V = valine
W = tryptophan	Y = tyrosine	

Check for understanding:
Explain why the sequence (the order) of the DNA bases that make up a gene is important for making a protein.

Activity 3.4c

Modeling the making of the LDL Receptor protein

What is the LDL Receptor?

_____

_____

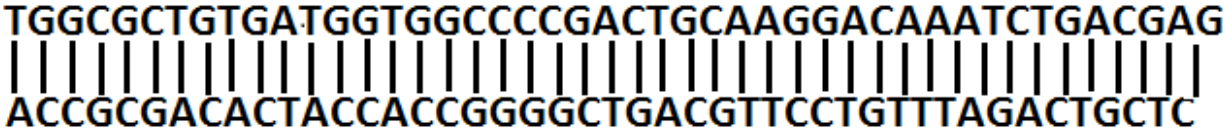
Directions:

Using your marshmallow models from Lesson 2 you and your group will model for yourselves the transcription and translation processes to "make" the LDL Receptor protein. Your desk will represent a "cell." The white sheet of paper your teacher gave you will represent the "nucleus." You will act as the enzyme and then as the ribosome.

Materials

1 piece of licorice
9 marshmallows
9 pieces of wire (7 cm)
6 gumdrops
marshmallow DNA model
White paper to represent the nucleus of the cell

Two-stranded DNA sequence for part of the LDL Receptor Gene



1) Which piece of the DNA will you transcribe? Look at your model Circle the gene segment you will transcribe from the part of the LDL receptor gene above.

RNA color code: **U- White, A- Pink, G- Green, C- Yellow**

2) Transcribe the gene segment. Draw a picture of your model. Write the mRNA sequence:

3) Break the mRNA up into codons and write the codons below:

4) Using the amino acid chart, translate the codons into amino acids. Make a key to identify the color gumdrops you used for your amino acids. Draw your model and write the amino acid sequence:

Check for understanding:

1) Where does transcription (DNA to RNA) take place?

_____

2) Where does translation (RNA to protein) take place?

_____

3) In your own words, describe how our genes are used as instructions to make proteins.

_____

_____

_____

_____

_____

_____

_____

4) Using what you've learned in this lesson about the relationship between DNA and proteins, write a scientific explanation to answer the question: How similar or different are we?

_____

_____

_____

_____

_____

Reading 3.4

Read and answer the questions that follow.

TAIPEI TIMES

Published on TaipeiTimes
http://www.taipeitimes.com/News/world/archives/2005/12/27/2003286244 (Edited)

Fish research helps uncover genetics of human skin color

DPA , WASHINGTON
Tuesday, Dec 27, 2005, Page 7

Scientists in the US have discovered what they believe is the gene that helps to determine whether a human has dark or light colored skin. In an article published in the journal *Science*, a team from Pennsylvania State University said two variations of the same gene strongly influence skin pigmentation.

The researchers reported that according to their findings 99 percent of the population of Europe has one version of the gene SLC24A5. In Africa, between 93 and 100 percent of the population have the other type. According to the main author, Keith Cheng, the discovery reveals important insights into the evolution of skin color in humans.

In an accompanying article also published in *Science*, molecular biologist Richard Sturm described the discovery as "absolutely original and pioneering". Dark pigmentation protects skin from harmful ultraviolet rays emitted by the sun. Ultraviolet light destroys vitamin B (folic acid) in the body which can lead to a number of serious health problems and in some cases to birth defects. But in climates with relatively little sunshine, dark pigmentation also prevents important chemical processes that take place in the skin such as vitamin D production. So as humans migrated to colder climates in the north it became important for skin to evolve to adapt to changing conditions.

Cheng and his colleagues identified the color gene for the first time in a rare breed of zebrafish. They examined the genetic code of a variety of zebrafish that had a golden pigmentation as well as lighter colored stripes than the common version. They concluded a gene that influences the melansomes -- granules that make the pigment melanin -- in the skin caused the difference between the two types of fish. The equivalent gene in humans is called SLC24A5. It causes melansomes to either grow in size and clump together causing dark skin, or to shrink, and the space between the particles to increase causing light colored skin.

Reading 3.4

Follow-up Questions

1. Why do people in Africa have different skin color than people in Europe?

2. How did scientists find that a gene affects skin color? (What did they look at?)

3. We learned that Tyrosinase was the protein responsible for the production of melanin. How could a gene for skin color have an effect on the protein Tyrosinase?

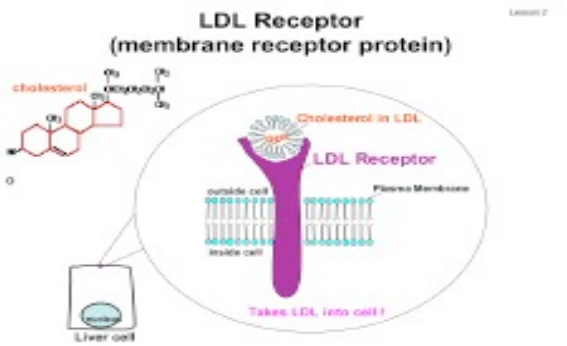
4. How does the information in this reading and the processes of transcription and translation help us answer the question "How similar or different is our skin?"

Reading 3.5a: Genomics Medical Journal

Genomics Medical Journal

LDL Receptor Proteins and Familial Hypercholesterolemia

www.ncbi.nlm.nih.gov/pubmed/9484998



Name of protein: LDL receptor

Type of protein: Membrane receptor protein

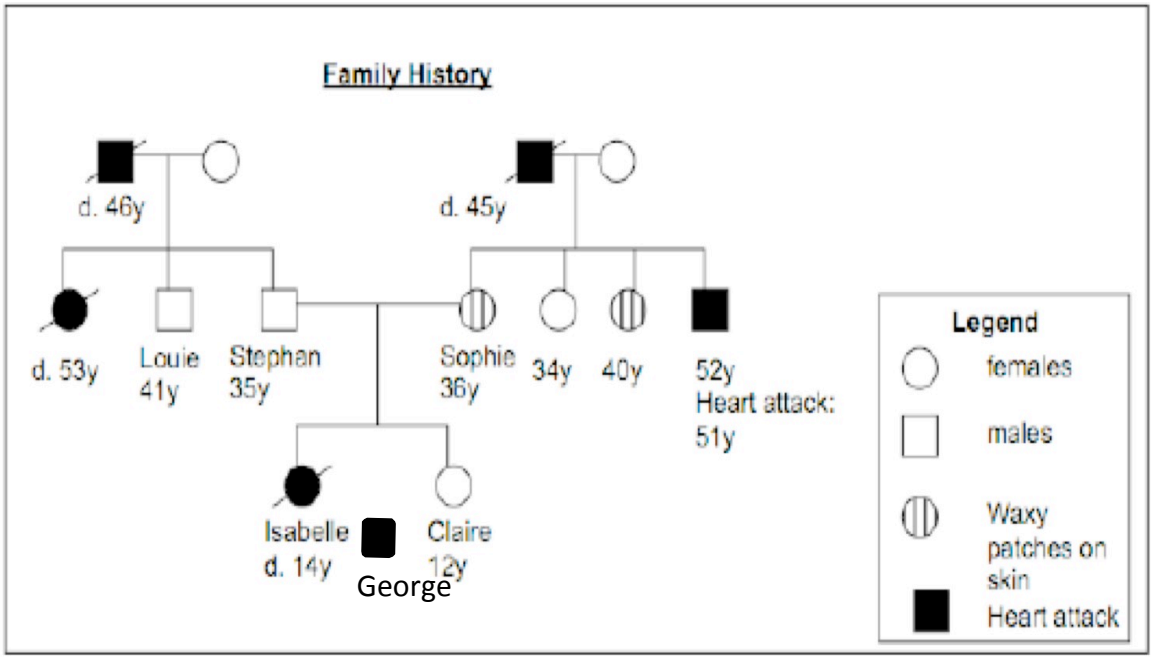
Cell type found in: Abundant in liver cells

The LDL Receptor is an example of a membrane receptor protein. When molecules bind to receptor proteins, a response is triggered on the inside of the cell. The LDL Receptor protein binds to a special particle called LDL, which contains cholesterol and brings cholesterol into the cell. The LDL Receptor protein is found in many cell types, but is abundant in the liver where cholesterol is taken up to be used for other things in the body such as building cell membranes or making hormones.

If the LDL Receptor protein could not function, then LDL and its associated cholesterol would not get into cells so that the cholesterol could be used. This causes an increase of cholesterol in the blood, which can build up in the arteries and cause heart attacks.

Environmental factors such as diet and lifestyle can also affect the cholesterol levels in patients that have problems with their LDL Receptor proteins. A study done in 1998 found that Chinese families living in Canada had higher levels of cholesterol in their blood than family members living in China. They both had problems with their LDL receptor proteins. The only difference that was found was in their lifestyle and diet. The Canadian Chinese patients had cholesterol levels similar to other western FH patients. They also had diets much higher in fat than their family members in China.

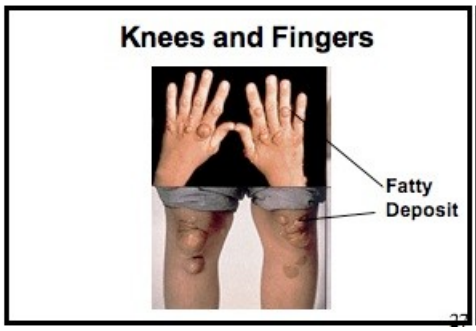
George's Family History
Rachael needed more evidence to explain what was wrong with George.
She interviewed George and was able to collect this information about his family.

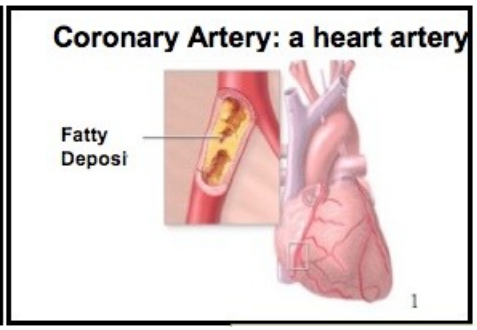


She also reviewed his symptoms. Here is the evidence she collected.

Reviewing George's Symptoms
Familial Hypercholesterolemia (FH)

- **Very high cholesterol in blood**
- **Waxy patches on the skin**
- **Chest pain and heart attacks at a young age**
- **Build up of fatty deposits on under skin and in arteries**





Think about George's symptoms and family history as you answer these questions.

1. What is the LDL Receptor protein doing in the body?

2. How is the shape of this protein important for its function? Do you think Georges LDL Receptor protein is the same shape as other people's LDL receptors? Give evidence for your answer.

Predict:

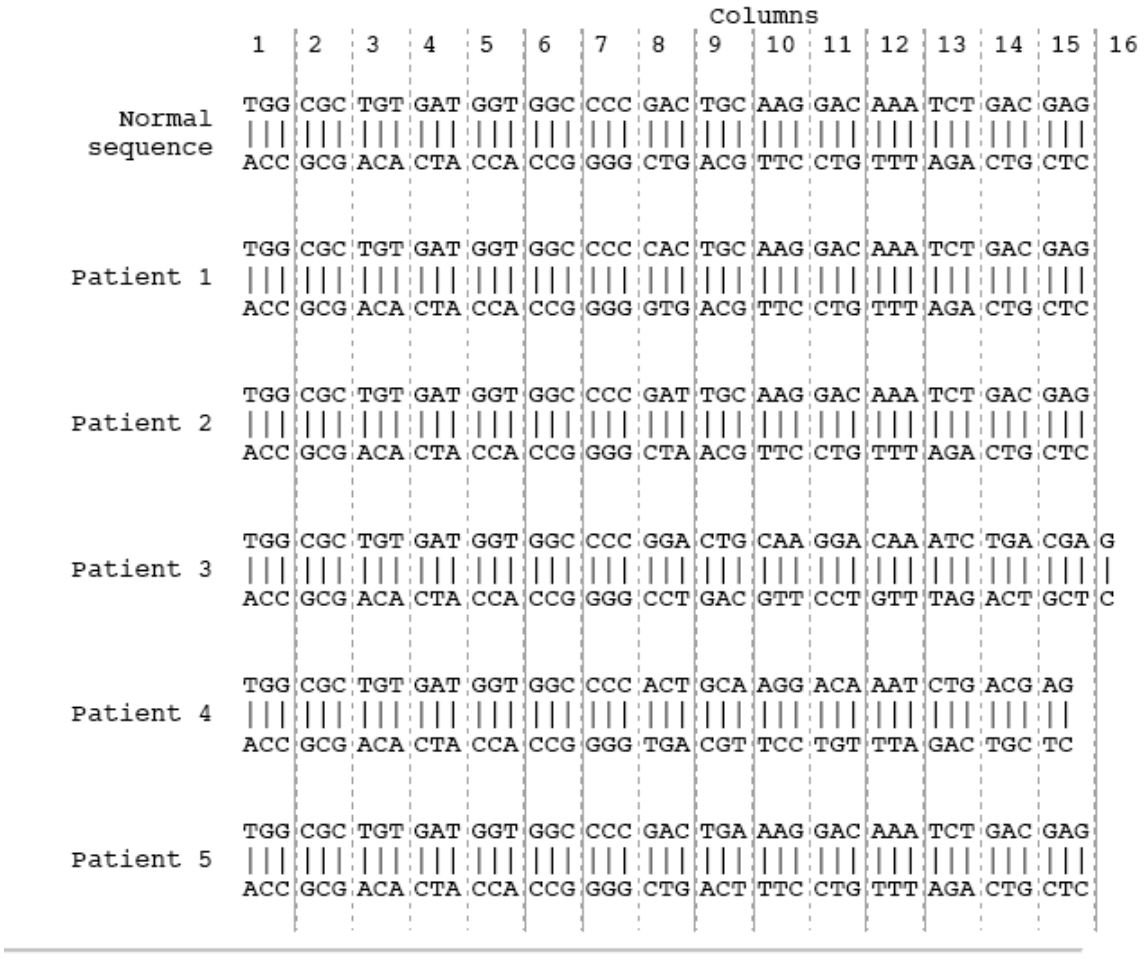
3. What would happen to the cells and tissues in the body if the LDL Receptor Protein did not work?

4. What would happen to the whole person if LDL Receptor Protein did not work?

5. What evidence does the article give that supports the claim that things in George's environment (such as diet) can affect the work done by his LDL Receptor Protein?

A Closer Look

You have already seen the family history of our patient George. Four different patients from George's family with high cholesterol want to know whether they have Familial Hypercholesterolemia or another disease. After they saw the doctor, their DNA sequences for the LDL receptor gene were sent to you. Patient 4 in the report is George and the other patients are his family members. It is your job as a laboratory technician to determine whether these patients have FH (familial hypercholesterolemia) disease.



Compare your patient to the normal sequence. Answer the following questions about the differences in the DNA sequence above. What kind of change is found in each DNA sequence? Also indicate which column (location) you found the change.

Laboratory Results for Familial Hypercholesterolemia

Complete this lab report after each team of technicians has presented their findings.

Patient	**DNA Sequence Difference**	**Location**	**FH Positive or negative**
1			
2			
3			
4- George			
5			

1. Convert your patient's DNA sequences to RNA sequences (Transcription), then determine the amino acid sequences (Translation).

   Findings for Patient #_____

   Location of DNA sequence change: _____

   DNA sequence:

   RNA sequence:

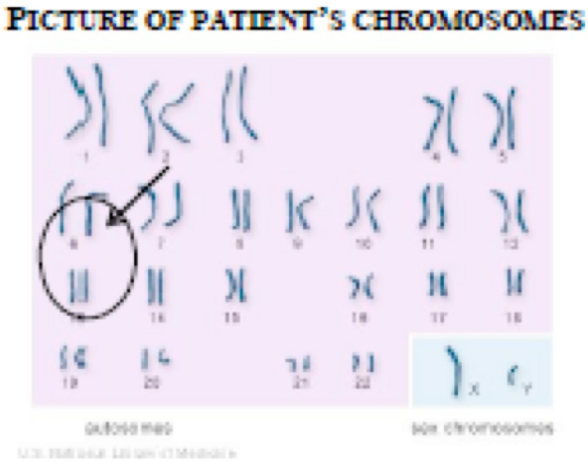
   Amino Acid sequence:

2. **What effect did the change in the DNA sequence have on the amino acid sequence?** Write a scientific explanation explaining how the amino acids sequence changed. **If the amino acid sequence did not change, write an explanation for why not.** (Hint: What is your **claim**? What are 2 pieces of **evidence** to support your claim? How can you tie your evidence to scientific principles in your **reasoning**?)

3. Based on your analysis of the DNA samples and the resulting amino acid sequence; **How do you think the change in DNA might affect the function of the LDL Receptor protein?** In complete sentences, write a scientific explanation of the affect on the protein function. (Hint: What is your **claim**? What are **2 pieces of evidence** to support your claim? How can you tie your evidence to scientific principles in your **reasoning?**)

4. Do you think that the person is likely to have FH? Why? What you would tell the patient.

Reading 3.5b: Mutations: Are they good or bad?



Remember George's Medical report showing a problem with his chromosome number 19? When there are changes in the DNA sequence, it can lead to a change in protein structure. When there is a change in protein shape, the function of the protein also changes.

When there is a DNA change from the normal sequence variations, this is called a mutation. **DNA mutations** or **variations** can occur for several reasons. There can be environmental reasons, such as UV light and nuclear radiation, and there can be mistakes that are made in the cell when DNA replicates. These kinds of changes cause errors in the DNA sequence. Since genes are made up of DNA, genes will also be affected by mutations. Many of these gene changes account for slight differences between people such as hair and eye color.

Some genetic mutations can actually have positive effects on traits. For example, some people that carry a gene for sickle cell also have immunity to malaria as a result. On the other hand, some changes in DNA might not have an effect on protein activity, and therefore never show as a different trait. But, some gene variations may result in disease or an increased risk for disease. Therefore, some mutations may have positive effects; some have no effects, while others may have negative effects. Although all differences in DNA are the result of mutations, scientists *only* refer to a change as a mutation when it is not part of the normal variations between people.

1. What is a DNA mutation and what are some reasons that mutations occur?

2. What are some good affects that genetic variations can have? Provide examples that you know about.

3. What are some bad effects that genetic mutations can have? Provide examples that you know about.

Types of Mutations

http://www.geneticheath.com/G101_Changes_in_DNA.shtml (Edited)

Point Mutation

A point mutation is a simple change in one base (A, T, C, or G) of the gene sequence. This is equivalent to changing one letter in a sentence, such as this example, where we change the 'c' in cat to an 'h':

Original
The fat cat ate the rat.
Point Mutation
The fat **h**at ate the rat.

As you can see, a simple letter change in a sentence changes the entire meaning of the sentence. Imagine what it can do to DNA and proteins.

Frame-shift mutation

In a frame-shift mutation, one or more bases are inserted or deleted, the equivalent of adding or removing letters in a sentence. This type of mutation can make the DNA meaningless and often results in a shortened protein. An example of a frame-shift mutation using our sample sentence is when the 't' from cat is removed and all the letters shift to the left:

Original
The fat cat ate the rat.
Frame Shift
The fat caa tet her at.

Deletion

Mutations that result in missing DNA are called deletions. These can be small, such as the removal of just one DNA base or longer deletions that affect a large number of genes on the chromosome. Deletions can also cause frame-shift mutations. In this example, the deletion eliminated the word cat.

Original
The fat cat ate the rat.
Deletion
The fat ate the rat.

Insertion

Mutations that result in the addition of extra DNA are called insertions. Insertions can also cause frame-shift mutations, and generally result in a nonfunctional protein.

Original
The fat cat ate the rat.
Insertion
The fat cat **xlw** ate the rat.

Notice that the insertion of letters made the sentence difficult to read. Imagine what could happen to genes if random DNA bases were inserted into a gene. Would the gene still have the same function?

Go back to your lab results on page 123 and identify the type of mutation for your patient. Add that information to your chart of their DNA.

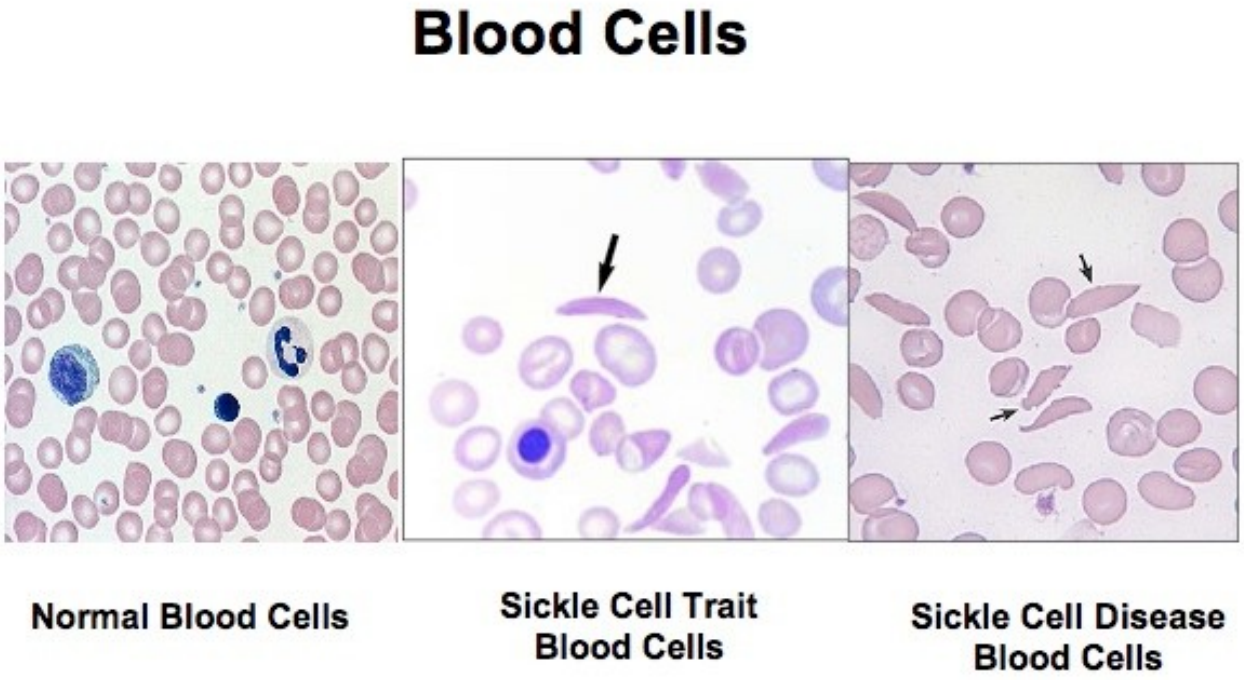
Check Your Understanding

1. Use what you learned in this lesson to write a scientific explanation for the following question; **How can two people have different versions of the same protein?**

2. Use what you have learned in the last lessons to write a scientific explanation that explains; **How similar or different is George from others?**

Learning Set 4:

Why do Some People Have Diseases Others Do Not?



Dale Lloyd II, No. 39. (The Rice Football Webletter)

Reading 4.1

Sudden Death and Sickle Cell Trait: How Knowing Your Genes Can Save Your Life

February 21st, 2011

Modified from article at: http://blog.pathway.com/sudden-death-and-sickle-cell-trait-how-knowing-your-genes-can-save-your-life/#more-1338

Dale Lloyd II, No. 39. (The Rice Football Webletter)

On a mild September afternoon in 2006, 19-year-old cornerback Dale Lloyd II stepped onto the practice field for a conditioning workout with the Rice University football team. After running 16 consecutive sprints of 100 yards each, he collapsed. He died the next day from acute exertional rhabdomyolysis (ER) associated with a genetic condition called sickle cell trait. The sickle cell trait can cause red blood cells to change from round to sickle-shaped, create "logjam" in blood vessels and kill. The National Athletic Trainers Association links "sickling" to the deaths of nine athletes in the past eleven years, including five college football players.

Lloyd did not know that he had the sickle cell trait, yet if he had known, some simple precautions could have saved his life. In order to keep other young athletes from suffering the same fate, Lloyd's parents filed a wrongful death lawsuit in September 2008 against Rice University and the National Collegiate Athletic Association (NCAA).

When the lawsuit was settled in June 2009, the NCAA agreed to require that, beginning in the 2010-2011 academic year, all Division I athletes undergo testing for sickle cell trait (PMID 20825310). It is estimated that this will ultimately affect more than 160,000 athletes.

This rare risk of sudden death for those with sickle cell trait is a vivid example of how a person's genes and lifestyle or environment play important roles in determining the risk of disease.

1. The article said that Dale had a "genetic condition" called sickle cell trait. What do you think that means?

2. Why do you think Dale reacted so differently to the same exercise that the other students experienced? What systems, organs or tissues in his body might be affected because he has the sickle cell trait?

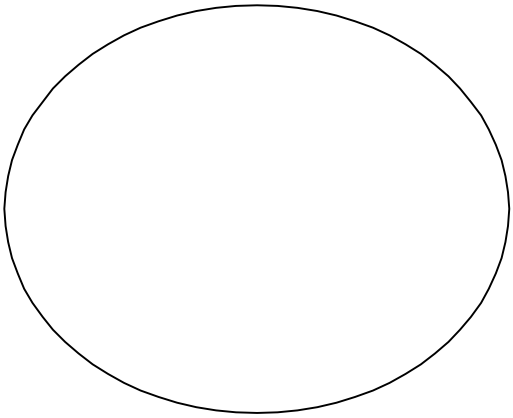
3. In the last lesson we looked at many types of evidence to determine if someone had Familial Hypercholesterolemia. What type of evidence do you think might tell us if a person had Sickle Cell disease or trait?

4. Should college athletes be forced to test for sickle cell? Do you think it will cause some athletes to be treated unfairly by schools, coaches and others? Could it affect scholarships that are offered? Is it worth the risk if it saves lives?

A Closer Look

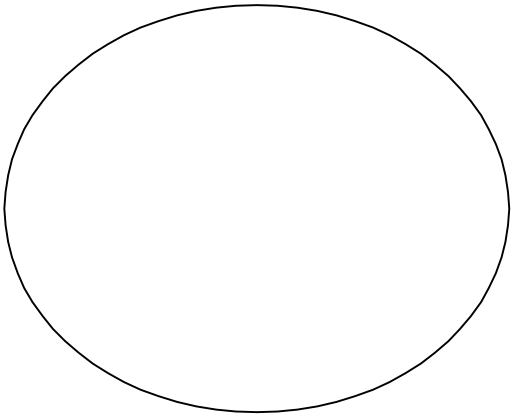
In class you looked at red blood cells under a microscope. While looking at these blood cells, you might have noticed that the shape of the red blood cells from a person with Sickle Cell disease was shaped differently from the person with healthy red blood cells. What were the differences you saw? Draw the red blood cell from the person with Sickle Cell disease and the person with healthy red blood cells, then write a brief description about what you observed regarding the shape of the red blood cells.

Sickle Cell disease red blood cells



Description

Healthy red blood cells



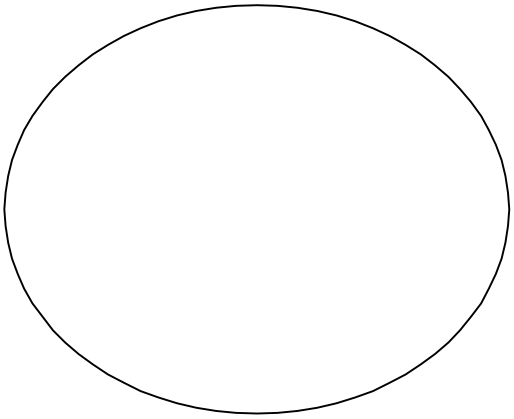
Description

How can people have different red blood cells?·· Analyzing hemoglobin

Look back at the article, "Sudden Death and Sickle Cell Trait: How Knowing Your Genes Can Save Your Life". Look back at your drawings of normal and sickle cell blood samples:

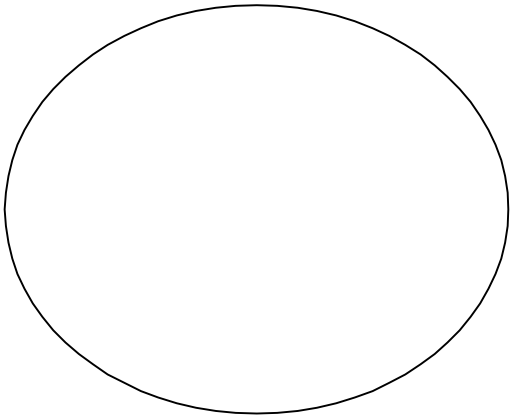
- Predict what the young man in the article's blood might look like. What evidence do you have to support your ideas?

Sickle Cell Trait
red blood cells
Prediction



Evidence

Sickle Cell Trait
red blood cells
Observation



Description

Similarities

Differences

What might have caused the differences in the blood cells of people with and with out Sickle Cell trait or Sickle Cell anemia?

- Why do you thinks so? Give evidence from what you already know about what causes differences in the activities of cells in the body.

Claim

Evidence from Video

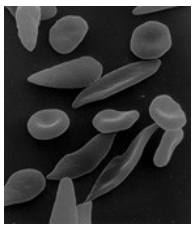
Reading 4.1b
Sickle Cell Disease: The Crooked Red Blood Cells

Red Blood Cells Supply Oxygen

What comes to mind when you read the words “red blood cells”? Do you think of arteries and veins? Do you think of blood flowing through your body? Do you think of oxygen? If you do, that is good because they are all related to red blood cells. In a healthy person, blood can flow through the veins because that person’s red blood cells are soft and can squeeze through small veins in the body, making the movement of oxygen possible. However, if something were to happen to the shape of the red blood cells that made it more difficult for them to pass through the veins, people would have a hard time getting oxygen to be delivered throughout the body.

Why is it called Sickle Cell Disease?

The composition of hemoglobin is the same in all people. The genes that code for hemoglobin are identical throughout the world. Occasionally, however, one of the genes is altered or mutated by any of a variety of "accidents" that can occur in nature. These alterations in the genes (called "mutations") are very rare. Most mutations in hemoglobin produce no problem. Occasionally, however, the alteration in the protein changes aspects of its shape and function. The change in shape creates the red blood cells to go from round to what is called “sickled”. This shape looks like a crescent moon or a banana. Since the red blood cells are no longer round, their ability to move through veins becomes stifled. This leads to organ failure, severe pain and or bone problems.



Crescent moon

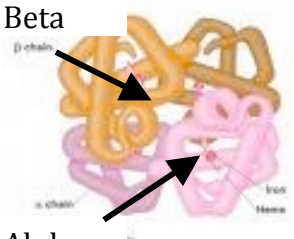
Can I Run a Marathon With Sickle Cell Disease?

The affect of Sickle Cell Disease on Oxygen Intake

Imagine trying to run a race while someone plugged your nose. You might imagine feeling suffocated or pain in your chest. This is because oxygen is not being transported around your body. In order for our bodies to do even the simplest things in life, oxygen is required.

Oxygen can be transported through the body by a protein in the blood called hemoglobin. Hemoglobin is the main substance of the red blood cell. It helps red blood cells carry oxygen from the air in our lungs to all parts of the body. As you can imagine, hemoglobin is very important for breathing.

Beta



Alpha

Hemoglobin is made from two similar types of proteins that "stick together". Both proteins must be present for the hemoglobin to pick up and release oxygen normally. One type of protein is called alpha, and the other is called beta. People with sickle cell conditions make a different form of hemoglobin; the beta component is mutated. As a result, red blood cells with this disease do not live as long as normal

red blood cells. They become stiff, distorted in shape and have difficulty passing through the body's small blood vessels. When sickle-shaped cells block small blood vessels, less blood can reach different parts of the body. Tissues that do not receive a normal blood flow eventually become damaged. This is what causes the complications of sickle cell disease.

Sickle cell disease would prevent someone from being able to run a marathon because they would not be able to transport enough oxygen throughout their body. The shape of their blood cells makes it difficult for them to travel through the small blood vessels.

What do you think?

1. What are some other activities that you think someone with sickle cell disease could not do? What are some activities that they could do?

2. Why do you think most athletes don't know that they have this condition?

3. Do you think the trait or the disease might affect athletes more than other people? Why?

4. Why do you think Sickle Cell is called a "genetic condition". (How are problems with proteins related to genes?)

Activity 4.2a - Analyzing Hemoglobin

1. Below is the DNA sequence for part of a normal hemoglobin protein: write the corresponding RNA and protein sequences. Then use your toobers to determine the shape of the hemoglobin protein.

DNA

TAC CAC GTG GAC TGA GGA CTC CTC TTC AGA CGG CAA TGA CGG GAC

RNA

_____

Protein

_____

Prediction

How will a mutation affect the protein structure and function?

_____

_____

2. Draw the shape of your assembled normal subunit of hemoglobin:

3. *Below is the DNA sequence for part of a hemoglobin protein in a sickle cell. How is it different from the normal hemoglobin? Write the corresponding RNA and protein sequences. Use your toobers to determine the shape of the hemoglobin protein.*

   DNA

   TAC CAC GTG GAC TGA GGA CAC CTC TTC AGA CGG CAA TGA CGG GAC

   RNA

   _____

   Protein

   _____

4. Using the sequence for Sickle Cell disease that you were given, draw the shape of your assembled Sickle Cell subunit of hemoglobin.

5. How similar or different are people with and without sickle cell disease?

	Similarities	Differences
DNA		
Amino acids		
Protein		
Cells		

Activity 4.3a (Optional)

How are gene mutations passed from parent to child?

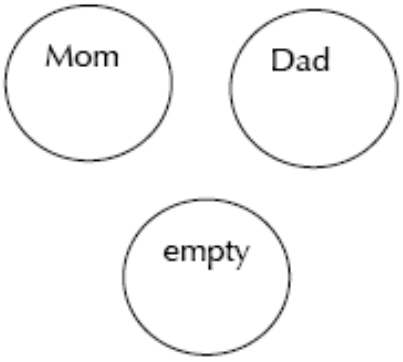
Adapted from "Pasta Genetics-Mendelian Inheritance of Fanconi Anemia and HLA type", Biological Sciences Initiative, *University of Colorado at Boulder,* www.colorado.edu/Outreach/BSI

Introduction

In this activity you will use colored pasta to represent human chromosomes carrying the hemoglobin beta gene. You will be given the pasta representing the parents and then be asked to create several possible embryos and determine whether any of them could have sickle cell anemia.

Setup

You will receive two dishes of pasta, one dish represents the mother, the other represents the father. Arrange the dishes as shown below.



Embryo

The spiral shaped pasta represents chromosome 11 that carries the hemoglobin beta genes.

Red represents the normal gene (allele) Hb^A
Green represents the mutant gene (allele) Hb^S
Each parent has one of each

Question

Will any of the embryos created have sickle cell anemia?

Hypothesis

_____

_____

Question

What percentage of the embryos created will carry the trait for sickle cell?

Hypothesis

_____

_____

Question
What percentage of the embryos will have normal hemoglobin?
Hypothesis

_____

_____

Procedure
Remember that one copy of chromosome 11 is placed into each egg and sperm (gametes). The egg and sperm (gametes) then unite to form a fertilized egg with two copies of chromosome 11, one from each parent. You will now use the parent's simulated DNA to generate simulated embryos.

1. Locate the dish representing the mom. Place your hand here. Close your eyes.
2. Without looking, pick up one spiral shaped pasta and put it in the dish representing the embryo.
3. Then locate the dish representing the dad. Without looking, pick out one spiral shaped pasta and put it in the dish representing the embryo.

Results
Note the colors of the pasta in the table below. Then note the hemoglobin type and Sickle Cell Anemia status of your embryo in the table. Replace the noodles in their original dishes, close your eyes, and repeat the process 2 more times.

Embryo	Colors of spiral pasta	Hemoglobin Type (Genotype)	Sickle Cell Anemia Status (Phenotype)
1			
2			
3			

Questions
How many of the embryos you created have normal hemoglobin?

How many of the embryos you created have Sickle Cell Anemia?

How many of the embryos you created carry the Sickle Cell Trait?

Out of the whole class
How many embryos were generated?

In the classroom, what fraction of embryos created have at least one mutant gene for beta hemoglobin?

What fraction of embryos created have Sickle Cell Anemia?

What fraction of embryos created have Sickle Cell Trait?

What if one of the parents had two normal hemoglobin beta genes and one parent has two mutant genes? Do you think that the outcome for the embryos would be different? Why or Why not?

Reading 4.3

How Can Sickle Cell Disease Be Passed On?

Connecting the Punnett Square and Genetics

How did Dale Lloyd get the sickle cell trait? Did anyone else in his family have the trait or the disease? Scientists have been studying questions like this about genetics since the 1800's. The following article tells how Mendel used plants to discover how traits are passed from one generation to another.

Gregor Mendel

Gregor Mendel was called the "father of genetics" because he studied how pea plants passed their genes to their offspring (descendents). Mendel looked at how plants obtained different characteristics. One day as Mendel was walking through a monastery, he noticed an interesting looking plant. He picked the plant and replanted it next to a similar plant. He wanted to see what their offspring would look like. He was curious about whether the parent plants would pass their genes to the offspring.

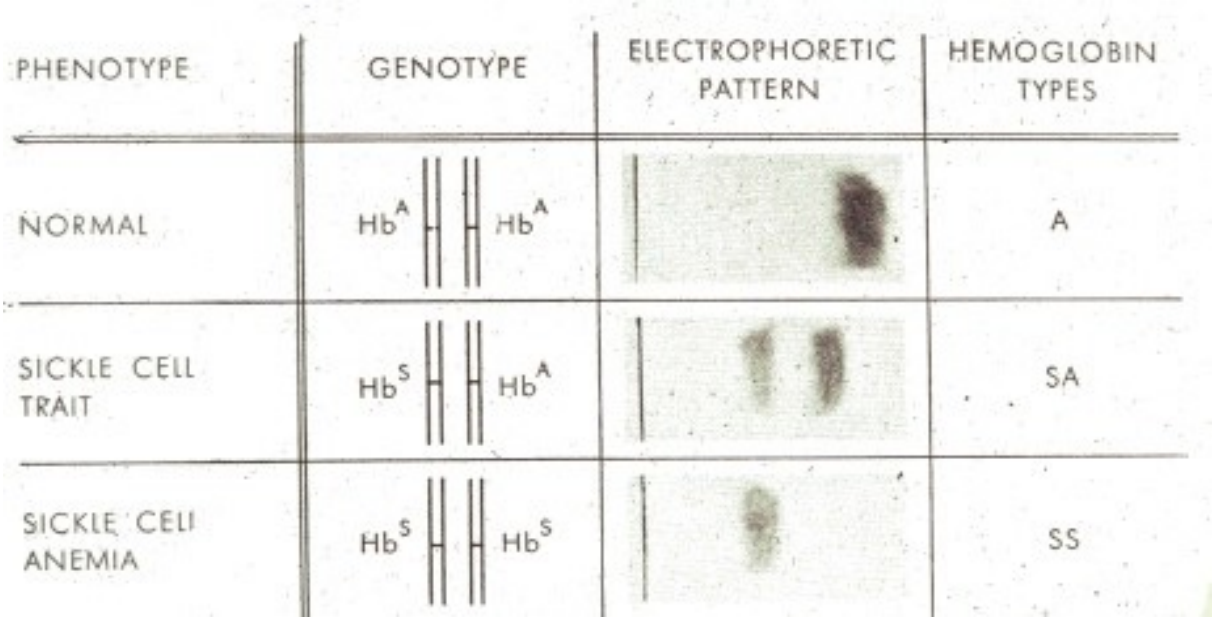
In the future generation, he found that the offspring plants had retained some of the parental traits. Traits are characteristics inherited from parents. Each offspring carried one gene from each parent. Mendel studied many generations of plants trying to discover patterns that would explain how and why certain traits were passed from parent to offspring. These tests encouraged more work to be done on heredity. Heredity refers to the idea that genes can be passed from parents to offspring. It was Mendel's work that allowed the study of genetics to emerge.

The study of genetics involves studying gene combinations that might result from breeding any two individuals. These combinations can be determined by drawing a diagram known as a Punnett Square. Punnett Squares are modeling tools that can be used to predict and compare the genetic variations that will result from a genetic cross. In the Punnett Square, you explore different gene combinations and the kinds of biological characteristics that come from them.

In the 1900's, people began to realize that there was an important part missing in Mendel's work-…genes and DNA are related. From this point, scientists began to learn that genes are made up of DNA, and that through genes proteins are built. Further research revealed that mutations in genes could also be passed to offspring, and could cause a change in the offspring's protein shape. These changes in protein shape can lead to diseases, such as Sickle Cell disease.

In the previous reading, you read about Sickle Cell disease. Sickle Cell disease is called a genetic disease. When scientists say that something is genetic, it means

that it can be passed from one generation to the next. A genetic trait is something that your mother or father can pass to their children through their genes.



The chart above shows the genotype and phenotype of people with the sickle cell trait and the disease. An organism's ***genotype*** is the set of genes that it carries. An organism's ***phenotype*** is all of its observable characteristics. What evidence do we have that a person with the sickle cell trait is producing both types of hemoglobin?

_____

_____

If a person with the sickle cell trait is making both types of hemoglobin then what does that tell you about their genes?

_____

_____

If a child has sickle cell anemia, could either of their parents have a mutated gene? Give evidence for your answer.

_____

_____

Activity 4.3b

How Do Children Receive Their Parent's Genes?

The following activity will provide a basic example of how parents can pass their genes to their offspring.

Scenario 1: Suppose a father has a gene for Sickle Cell and a mother does not. If *SS* means that a person has sickle cell disease, determine which children will have the disease.

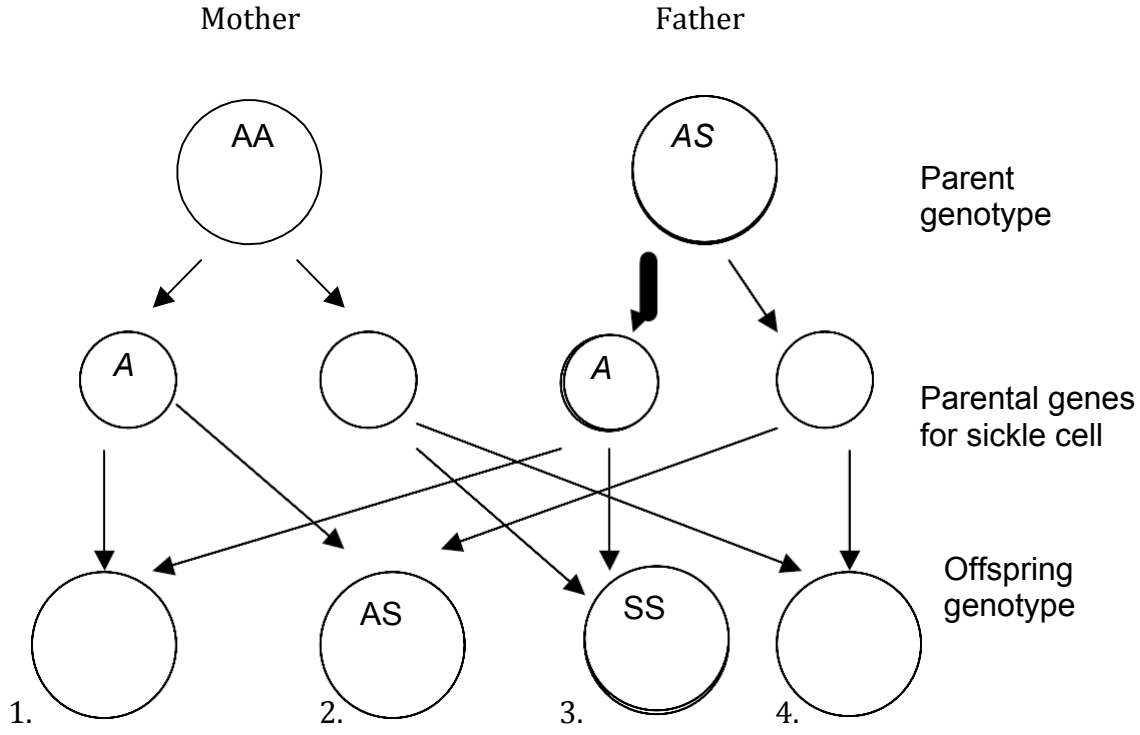
Fill in the circles with the appropriate genes. Some of the circles have been filled in for you. Remember that the offspring will have 2 genes because each offspring receives one gene from each parent.

A = Normal hemoglobin gene
S = Mutated hemoglobin gene

AA = No- Sickle Cell Anemia
SA = No- Sickle Cell Anemia/ Sickle Cell Trait
SS = Yes- Sickle Cell Anemia

Mother's genes – AA

Father's genes - AS



Scenario 2: Suppose neither the father or mother have sickle cell anemia but both have the trait. If *AS* means that a person has sickle cell trait, determine which children will have the disease.

Use the Punnett Square to help determine the gene combinations. A **Punnett square** is a chart, which predicts all possible gene combinations in a cross of parents (whose genes are known). Punnett squares are named for an English geneticist, **Reginald Punnett**. He discovered some of the basic principles of genetics. The outside squares represent the genes of the parents. The inside squares show the genes of all possible offspring. Fill in the middle squares in order to determine the probability of each type of offspring.

Draw a Punnett Square below with both parents having an "A" and "S" gene.

A = Normal hemoglobin gene
S = Mutated hemoglobin gene

AA = No- Sickle Cell Anemia
AS = No- Sickle Cell Anemia/ Yes Sickle Cell trait
SS = Yes- Sickle Cell Anemia

After filling in the circles and the Punnett Square, answer the following questions.

1. How many of the offspring have Sickle Cell disease? Remember, a person will have Sickle Cell disease if they carry BOTH mutated hemoglobin genes (SS). (Circle one)

   a. 3/4 b. 1/2 c. 1/4 d. 0

2. How can a child get Sickle Cell disease from the parents?

3. Why is Sickle Cell disease called a genetic disease? In other words, explain what happens at the genetic level when someone has sickle cell.

4. Using the chart below and complete sentences, write a molecular explanation for how Sickle Cell disease arises from a mutation in the hemoglobin gene. You will need to remember the video you watched about the young girl with Sickle Cell disease. (Your explanation should include a description of the disease at the trait, tissue, cellular, protein, and DNA levels.)

Explanation Chart

Use these guidelines to help you construct a molecular explanation.	Write a claim and two pieces of evidence to answer the questions in the spaces below.
Trait/Symptom Level: Are there physical problems related to sickle cell disease that doctors and patients can see or describe by examining their body and behaviors? (Such as fatty deposits, coughing, or weakness.)	
Tissue/Organ Level: Are there problems related to sickle cell disease that happen inside the body that affect the organs and/or tissues (such as the heart, circulatory system or brain)?	
Cell Level: Are there problems that happen to the cells or in the cells caused by the disease?	

Activity 4.4 (Optional)

Natural Selection: How can environmental factors such as malaria affect the frequency of organisms with gene mutations within a population?

Adapted from "Allele Frequencies and Sickle Cell Anemia Lab", the GENETICS Project and the Genetics Education Partnership University of Washington, Dept. of Molecular Biotechnology, http://genetics-education-partnership.mbt.washington.edu/Download/file.html

Introduction: Gene frequency refers to how often a gene mutation occurs in a population. Gene frequencies can change in a population over time, depending on the **'selective forces'** shaping that population. Predation, food availability, and disease are all examples of selective forces. **Evolution occurs when gene frequencies change in a population!**

In this activity, red and white beans are used to represent the normal and mutant beta hemoglobin gene. The RED beans represent gametes (sperm or egg cells) carrying the normal beta hemoglobin gene (A), and the WHITE beans represent gametes (sperm or egg cells) carrying the mutant beta hemoglobin gene (S). All of the beans together represent the entire gene pool. A **gene pool** is the genetic make-up of a specific population, and is the combination of all the genes for traits members of the population exhibit. This gene pool exists in a region of Africa that is infested with malaria. You are simulating the effects of a high frequency of malaria on the gene frequencies of a population.

Materials:
75 red beans, 25 white beans, 5 containers (e.g. paper cups)

Hypothesis/Prediction:
What do you think will happen to the frequencies of the A and S alleles as a result of the presence of malaria? (Will the frequency of A increase or decrease? What about S?) Formulate a hypothesis and corresponding prediction. Be sure to explain your reasoning.

_____

_____

_____

Procedure:

1. Together with your lab partner, obtain five containers and label them as follows:
   1) AA 2) AS 3) SS 4) Non-surviving alleles 5) Gene Pool
2. Place the 75 red and 25 white beans in the Gene Pool container and mix the beans up.
3. Simulate fertilization by PICKING OUT two 'alleles' (beans) WITHOUT LOOKING.
4. For every two beans that are chosen from the gene pool, another person will FLIP A COIN to determine whether that individual is infected with malaria.
5. Using the table below, the coin flipper tells the bean picker in which containers to put the beans.

Genotype	Phenotype	Malaria (heads coin toss)	Not infected (Tails in coin toss)
AA (red-red)	No sickle cell disease. Malaria susceptibility.	Place in Non-surviving	Survive: place in AA
AS (red-white)	No sickle cell anemia, some symptoms when exposed to low oxygen. Malaria resistance.	Survive: place in AS	Survive: place in AS
SS (white-white)	Sickle cell disease.	Place in Non-surviving	Survive for a brief time: place in SS

Repeat steps 3–5 until all the beans in the Gene Pool are used up.

1. At the end of the round, COUNT the number of individual red beans (A alleles) and white beans (S alleles) in the containers labeled AA and AS. These individuals survive to reproduce. RECORD those numbers in the F1 TOTAL SURVIVING ALLELES table. Put them in the gene pool afterwards.
2. Because SS individuals do not survive to reproduce, move all beans from the SS alleles container into the Non-surviving alleles container.

STOP AFTER ONE GENERATION. CHECK WITH YOUR TEACHER BEFORE GOING ON!

3. Repeat the procedure for the F2 generation. Record your results in the F2 TOTAL SURVIVING GENES table

Results

F1 TOTAL SURVIVING GENES: (very important to record)

Number of A (RED) genes surviving (Count out of AA and AS containers)	
Number of S (WHITE) genes surviving (Count out of AS container)	

Put the survivors in the gene pool and create the next generation.

F2 TOTAL SURVIVING GENES: (very important to record)

Number of A (RED) genes surviving (Count out of AA and AS containers)	
Number of S (WHITE) genes surviving (Count out of AS container)	

Class Results

On the class chart, record your number of A alleles surviving for the next generation and number of S alleles surviving from both the F1 TOTAL SURVIVING GENES and F2 TOTAL SURVIVING GENES tables. Then record the class totals below and calculate the frequencies using the formula below.

Using the formulas below, calculate the % gene frequency for each gene in each generation:

$$\frac{\text{Total A}}{\text{Total A+S}} \times 100 = \text{\% Gene A} \qquad \frac{\text{Total S}}{\text{Total A+S}} \times 100 = \text{\% Gene S}$$

Class Results Table

	Parents		F1		F2	
	A	S	A	S	A	S
Class Total						
Allele Frequency						

Discussion Questions

1. What do the red and white beans represent in this simulation? What does the coin represent?

2. Natural selection is a process, in the theory of evolution, where organisms that are best suited to their environment survive and pass on their genetic characteristics in increasing number to succeeding generations while organisms that are less adapted tend to be eliminated. What are the “selective forces” in the environment in this simulation (the forces changing the gene frequencies)?

3. What was the general trend you observed for Allele A over the three generations (did it increase or decrease)? What was the general trend for Gene S over time? Was your hypothesis supported?

4. Do you anticipate that the trends in question 3 will continue for many generations? Why or why not?

5. Since few people with sickle cell anemia (SS) are likely to survive to have children of their own, why hasn’t the mutant gene (S) been eliminated? (Hint: what is the benefit of keeping it in the population?)

6. Why is the frequency of the sickle cell gene so much lower in the United States than in Africa?

Reading 4.4

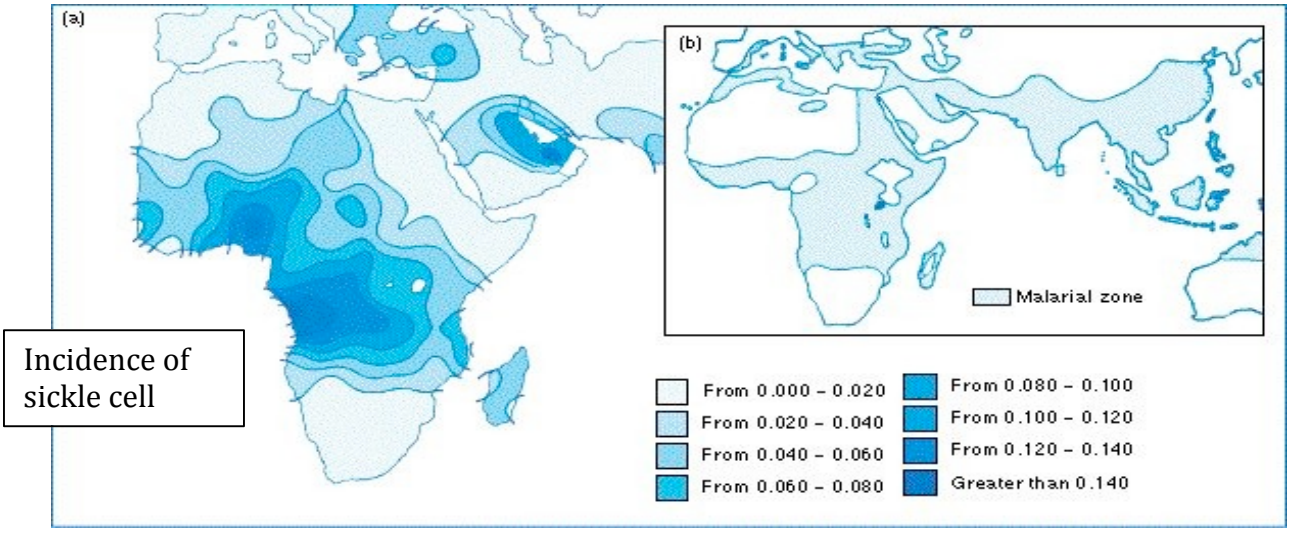
Are All Mutations Bad? The Case of the Sickle Cell Mutation

A Mutation Story:

A gene known as HbS (the gene for sickle cell) was the center of a medical and evolutionary detective story that began in the middle 1940s in Africa. Doctors noticed that patients who had sickle cell anemia [a form of sickle cell disease], a serious hereditary blood disease, were more likely to survive malaria, a disease that kills some 1.2 million people every year.

What was puzzling was why sickle cell anemia was so prevalent in some African populations. How could a "bad" gene -- the mutation that causes the sometimes-lethal sickle cell disease -- also be beneficial? On the other hand, if it didn't provide some survival advantage, why had the sickle gene persisted in such a high frequency in the populations that had it?

The sickle cell mutation is an error in the DNA code of the gene that tells the body how to make a form of hemoglobin, the oxygen-carrying molecule in our blood. Every person has two copies of the hemoglobin gene. Usually, both genes make a normal hemoglobin protein. When someone inherits two mutant copies of the hemoglobin gene, the abnormal form of the hemoglobin protein causes the red blood cells to lose oxygen and warp into a sickle shape during periods of high activity. These sickled cells become stuck in small blood vessels, causing a "crisis" of pain, fever, swelling, and tissue damage that can lead to death. This is sickle cell anemia.



Incidence of sickle cell

Researchers found that the sickle cell gene is especially prevalent in areas of Africa hard-hit by malaria. As much as 40 percent of the population in some regions, carry at least one hemoglobin gene for sickle cell. It turns out that, in these areas, the sickle cell gene carriers have been naturally selected, because the trait gives them some resistance to malaria. Their red blood cells, containing some abnormal hemoglobin,

tend to sickle when they are infected by the malaria parasite. Those infected cells flow through the spleen, which picks them out because of their sickle shape -- and the parasite is eliminated along with them.

Scientists believe the sickle cell gene appeared and disappeared in the population several times, but became permanently established after a particularly vicious form of malaria jumped from animals to humans in Asia, the Middle East, and Africa.

In areas where the sickle cell gene is common, the immunity associated with the Sickle Cell gene has become a selective advantage. Unfortunately, it is also a disadvantage because the chances of being born with sickle cell anemia are relatively high.

1. The article above talks about some of the advantages and disadvantages of having the sickle cell gene. What is an advantage to having the sickle cell gene?

2. The article also shows that not all mutations are bad. Explain why you agree or disagree with the statement: “All mutations are a disadvantage to a person.” Give evidence for your answer.

Challenge Question:
What difference might the advent of new technology to diagnose and treat sickle cell disease and trait make on the frequencies of the A and S alleles in the population?

Returning to the Driving Question

Write a scientific explanation to answer the following question:

How do environmental conditions, such as "selective pressures" caused by malaria, affect how similar or different we are?

*Make a claim; give evidence and scientific reasoning for your answer.

Activity Sheet 4.5

Fill this data table in as you analyze the data found below.

	Lactose Intolerance (Yes or No)	DNA Mutation (Yes or No) If yes, what is the mutation?	Protein Present (Yes or No)	RNA (Yes or No)	The DNA Sequence Found Near the Lactase Protein Gene – If there is a difference, what is it?	Genes On or Off?
Jason						
Mom						
Chelsea						
Maya						

Predictions:

Why do you think Jason might be lactose intolerant?

Why do you think the rest of Jason's family does not have lactose intolerance?

DNA Sequence Analysis of Lactase Protein Gene

Mom:	no mutations in lactase genes
Jason:	no mutations in lactase genes
Chelsea:	no mutations in lactase genes
Maya:	no mutations in lactase genes

1. Do these results help explain why Jason is different from his sisters? Why or why not?

Gel Electrophoresis: Lactase Protein Samples from Jason's Family

Lactose intolerance:

1) No (Mom)
2) Yes (Jason)
3) No (Chelsea)
4) No (Maya)

Why does Jason have no protein?
Why Mom and Chelsea have less protein?

2. Do all the family members have the same amount of lactase protein? Explain.

_____

_____

_____

3. Do you think that you need a lot of protein to break down lactose? Explain why you think so? (Hint: How much protein do Mom and Chelsea have? Can they break down lactose?)

_____

_____

_____

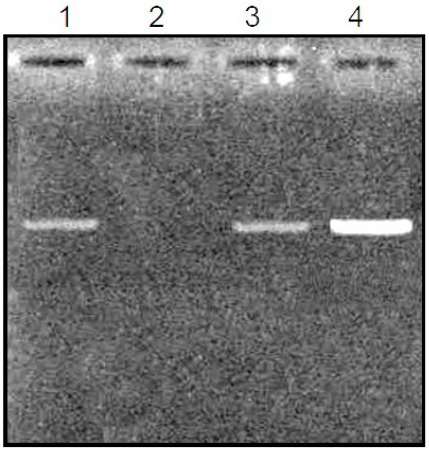
4. Using the RNA and lactase protein gel electrophoresis data, why do you think Jason does not have any lactase protein?

_____

_____

_____

RNA Analysis of Lactase Protein Gene



Lactose intolerance:

1) No (Mom)
2) Yes (Jason)
3) No (Chelsea)
4) No (Maya)

5. The image above is an electrophoresis gel that shows the RNA results of Jason and his family. What do you notice about the gel?

6. Why are some of the lines brighter than other lines?

7. Why do you think there is no line for Jason?

8. Is transcription of the lactase gene happening in Jason's small intestine cells? Why or why not?

DNA Sequence of the Area Near the Lactase Gene

To help understand why Jason is lactose intolerant, the DNA lab looked at the DNA near the lactase gene. It could be possible that this part of the DNA could affect the function of the lactase gene. Below is the DNA sequence for each family member. There are two sets because the lab sequenced near both copies of the lactase gene.

Circle any differences you notice in the base pairs.

Mom:	ATTTGC |||||| TAAACG ATCTGC |||||| TAGACG	Jason:	ATCTGC |||||| TAGACG ATCTGC |||||| TAGACG
Chelsea:	ATCTGC |||||| TAGACG ATTTGC |||||| TAAACG	Maya:	ATTTGC |||||| TAAACG ATTTGC |||||| TAAACG

9. What do you notice about the gene sequences near the gene?

_____

_____

_____

10. How is the DNA sequence near the lactase genes in Jason different from the other members of his family?

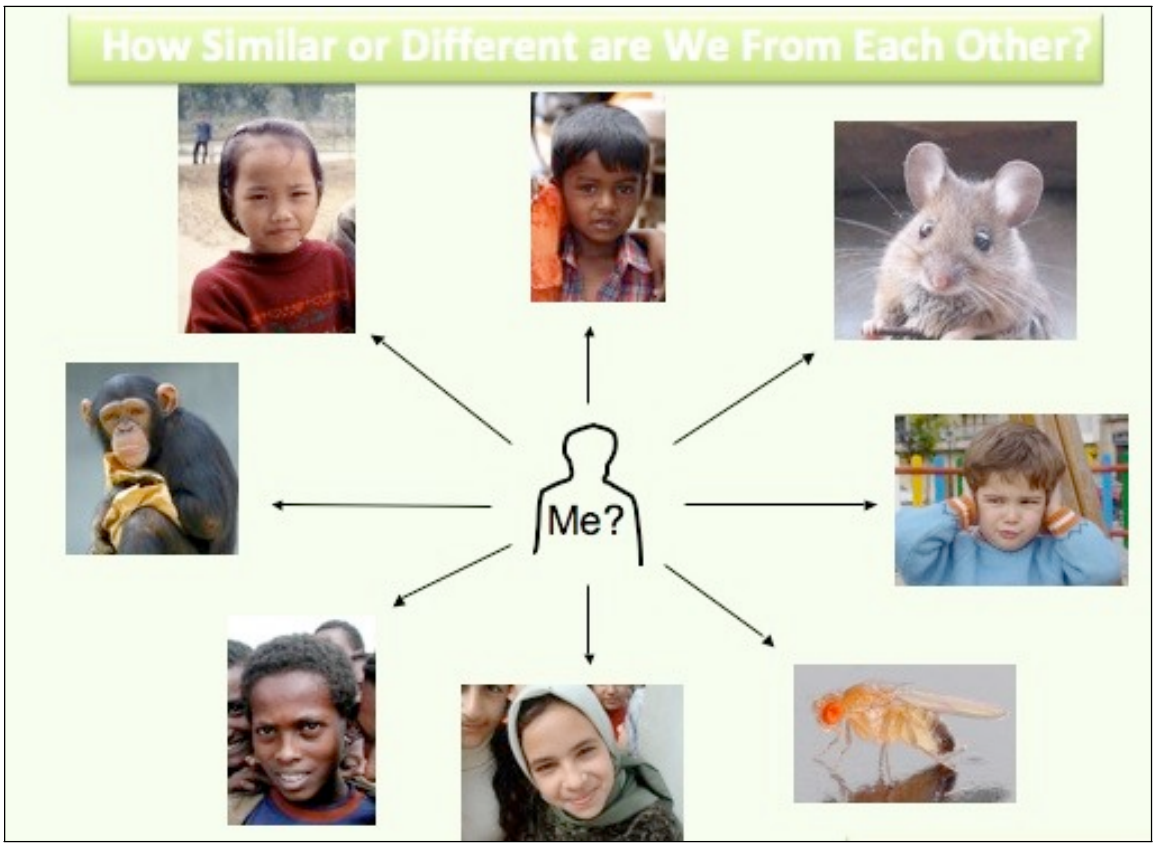
_____

_____

_____

11. Using the above data, write a scientific explanation answering the question: Why does Jason have lactose intolerance?

Learning Set 5
Are We More SIMILAR or DIFFERENT?



Activity 5.1a

Introduction to the Human Genome Project

Review:

1) Throughout this unit we have spent time examining different ethical dilemmas. What is an ethical dilemma?

2) What are some examples of ethical dilemmas? (Identify two we studied in this unit AND one of your own.)

Read the following story and answer the questions that follow.

Priya Should Find Out If She Inherited a Fatal Disease (or should she?)

http://www.ornl.gov/sci/techresources/Human_Genome/publicat/genechoice/2_priya.html

Priya has just lost her mother to an illness called Huntington's disease. It was hard for Priya to watch her mother die. First her mother had strange mood changes. Then her arms and legs began twitching. Soon she couldn't talk or control her movements. In the end, she was totally bedridden and could barely get food down without choking.

Priya knows that Huntington's disease usually strikes people in middle age. It is always fatal, and there is no treatment. She also knows that since the disease is inherited, she has a strong chance of getting it herself. Priya just learned about a test she can take. The test will tell if she carries the gene for Huntington's disease. She is tempted to take the test. She thinks that if she could find out once and for all whether she will get the disease, she could plan for her future. On the other hand, she wonders if it is better not knowing. At least then Priya would still have some hope.

1) If you were Priya, what would you do? Would you want to find out whether you have the disease or not? Why?

2) Based upon all that you have learned in the unit, how do you think it is possible for Priya to take a test that can tell her if she carries the gene for Huntington's disease?

The Human Genome Project

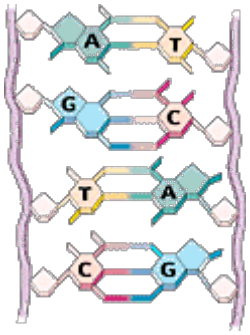
http://www.ornl.gov/sci/techresources/Human_Genome/publicat/genechoice/2_priya.html

In Learning Sets 2 and 3 we learned that genes contain the instructions for making proteins and for carrying out all the activities that goes on inside our cells. The complete set of genes for a human being is called the human **genome**. It may help to think of the human genome as a book of instructions, with each gene a single instruction. You have a copy of this instruction book inside nearly every one of the trillions of cells in your body. The book is written in a language that only your cells can read—DNA!

We will have to learn this language if we want to learn all the secrets of the genes. So this is exactly what scientists from around the world have decided to do. They have set out to learn the language of the genes. This international effort is called the **Human Genome Project**. The United States is spending 3 billion dollars over 15 years on this project. Other countries also are investing large amounts on research. It is a huge undertaking that involves researchers in biology, physics, engineering, computer science, and many other fields.

The task they have taken on is challenging and exciting, but difficult. We learned that genes are made of **DNA**, which contains chemical ingredients called **bases**. There are only four bases, but they repeat one after the other in an ever-changing order throughout the genes. Think of the four bases as letters of the alphabet, combining together in some strange language to spell out each gene's instruction.

With just four letters, the alphabet of this mysterious language is very short. However, the words written in this language are not short at all. A single gene has thousands of bases. Some genes have millions of bases. So each gene is like a single word with thousands or millions of letters to it. In addition, it is hard to figure out where each gene begins and ends, because the spaces in between also are filled with long strings of letters.

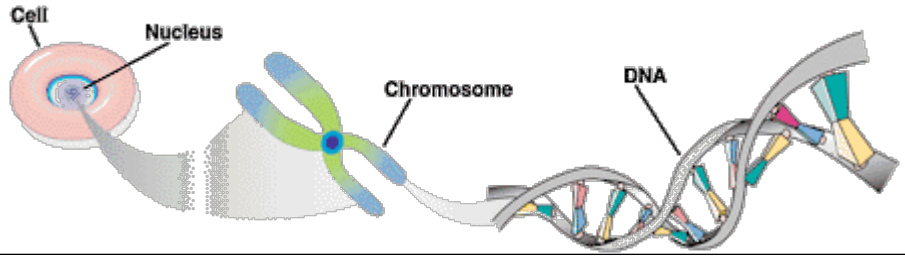


This close-up shows the four bases that make up the genetic code. The bases are thymine (T), adenine (A), guanine (G), and cytosine (C).

The immediate goal of the Human Genome Project is to put together a copy of the human instruction book, letter by letter. Having this copy to read will make it easier to decipher the language of the genes. But making the copy is very difficult. First, scientists have to get inside a cell. Then they have to get inside the **nucleus** of the cell to the DNA. The DNA is curled into tight coils, so they have to uncurl it.

Then they have to look at the DNA to see which of the four bases comes first, which second, which third, and so on. Then they have to write this down.

It may sound simple, but it isn't. This is a job that involves unthinkably small objects and incredibly large numbers. It would seem to be an impossible task. Yet, because of advances in computers, microscopes, chemical analysis, and other tools of science, it is a job that is practically done!



3) What is the human genome?

4) What is the purpose of the Human Genome Project?

5) Why is this project important to humanity?

6) What would *you* hope scientists could learn from this project?

Checking In

How would knowing the human genome help answer the Driving Question?

Activity 5.1b

Comparing Genomes

Make a prediction before the activity:

1) Write a scientific explanation to answer the question: How similar are two humans' DNA sequences? (Hint: Your claim can be in the form of a percentage)
   Accept all reasonable scientific explanations. Samples have been provided to guide you.

2) Write a scientific explanation to answer the question: How similar is a human DNA sequence to a chimpanzee DNA sequence? (Hint: Your claim can be in the form of a percentage)

Directions during the activity:

1) Your teacher will give your group a packet of DNA sequences for two humans or a human and a chimp. It is recommended you split up the sheets to each member of your group to make the comparison process more efficient.
2) Count the number of differences between the two DNA sequences for an entire page. (You should record this number somewhere so you do not forget it.)

3) Each line contains 60 DNA bases. Determine the total number of bases for each page.
4) Determine the total # of identical bases for each page:

Total # of identical bases = Total # of bases – Total # of different bases

5) Determine the total # of identical bases for all pages.
6) Determine the total # of bases for all pages.
7) Determine the percent of identical bases:

$$\% \text{ of identical bases} = \frac{\text{total \# of identical bases (from 5)}}{\text{total \# of bases (from 6)}} \times 100\%$$

Record your data in the table below:

	Total # bases	**Total # identical bases**	**% identical DNA**
Two humans			
Human & chimp			

Follow-up:

1) How similar are two humans' DNA sequences?

2) What does this mean about these two people?

3) Do you think you would find similar results if you took any two people from any location in the world? Explain what you mean.

4) How similar are humans and chimpanzees?

5) What does this mean about chimps and humans?

6) Explain what degree of similarity you would expect to find if you compared humans to other species of animals, such as alligators, or flies. Explain.

7) Revise your scientific explanation comparing two humans' DNA based on what you learned in this activity. Make sure you include: **Claim, Evidence, and Reasoning**

Why is it important for scientists to "map" the genomes of humans and other organisms.

Revisit the Driving Question:
Based on what you learned in this lesson, write a scientific explanation answering the driving question. Make sure you include: **Claim, Evidence, and Reasoning**

Lesson 2: How similar are we in how the environment affects our genes?

Think back to what you learned about Jason's lactose intolerance and the human genome.

1) What did you find was the cause of Jason's lactose intolerance? Provide evidence from the previous lesson to support your answer.

_____

_____

_____

2) What was the "switch" that enabled Maya, Chelsea, and Mom to make lactase, but not allow Jason to make lactase? Where is this "switch" located? Be as specific as possible.

_____

_____

_____

3) Do you think that Jason's genome more similar or different from the rest of his family?

_____

_____

_____

Activity 5.2a

In this activity you will work with your group to create a macro-molecular chart for a specific phenomenon. In the process of doing this you also want to focus on how the environment interacts with the gene for your phenomenon.

Directions:

1) Use the readings related to your phenomenon to answer the following questions. The readings for each of the phenomena are:

Skin Color: LS1, How cells affect skin color LS1, Skin color adaptation LS2, What happened to Sammy Sosa? LS3, Fish research helps uncover genetics of human skin color
Lactose Intolerance: LS2, Don't Pass the Milk Please LS2, New Spoof of Milk Mustache Ad Spotlights Lactose Intolerance
Familial hypercholesterolemia:

LS3, The Medical Case: Familial Hypercholesterolemia LS3, The Medical Case: Familial Hypercholesterolemia, part 2 LS3, Genomics Medical Journal
Sickle cell: LS4, Sudden Death and Sickle Cell Trait: How Knowing Your Genes Can Save Your Life LS4, Sickle Cell Disease: The crooked red blood cells LS4, Are All Mutations Bad? A Mutation Story

2) Use the readings identified above to help you identify the following characteristics of your phenomenon from the macro world to the molecular world. Enter the characteristics into the Genomics Unit Phenomenon table.
    a. The phenomenon
    b. Possible biological physical traits displayed by individuals experiencing this phenomenon
    c. Organ involved
    d. Type of cells involved
    e. Protein
    f. Gene expression: What does the gene tell the protein to do? And, what happens if the gene has a mutation or the protein is malfunctioning in some way?
    g. Gene-environment interaction
3) Complete your section of the table and be prepared to share out your findings with the rest of your class.
4) As your classmates share out the characteristics of their phenomenon, enter the details into the table.

Genomics Unit Phenomena Table 1

Phenomenon	**Skin color**	**Lactose intolerance**
Possible Traits		
Organ		
Cell type		
Protein		
Gene expression		
Environment interaction		

Genomics Unit Phenomena Table 2

Phenomenon	**Familial hypercholesterolemia**	**Sickle cell**
Possible Traits		
Organ		
Cell type		
Protein		
Gene expression		
Environment interaction		

Check For Understanding:
Write a scientific explanation: How can the environment influence our genes?

Claim: How can the environment influence our genes?

_____

_____

_____

Evidence: Use evidence from the table you just completed.

_____

_____

_____

Reasoning: Explain how your evidence supports your claim with science from the readings if necessary.

_____

_____

_____

Activity 5.2b

In this activity you and your group will be creating a concept map which includes details for your specific phenomenon. You want to synthesize and review all the content we learned about the phenomenon.

What is a concept map? What is the purpose of a concept map? What are some characteristics of concept maps?

_____

_____

_____

Directions:

1) Begin by looking back through your student reader for information about your phenomenon. Identify key ideas (words or short phrases) about the phenomenon. Refer to the macro-molecular diagram to guide you. Be sure to include the key ideas from the previous activity. For example, a key word for lactose intolerance might be "lactase."
2) Write these words/short phrases on Post-its (one word/phrase per Post-it).
3) Work as a group to move the words around the chart paper, trying different placements of words until your group comes to consensus on a general structure for your concept map.
4) Using the marker, begin connecting the words/phrases to each other. For every connecting line you draw you must also have connecting words to show the relationship.
5) Add in extra information as you see necessary. Extra information could include evidence from readings and activities we've done in class.
6) Tape down the Post-its so they do not fall off your chart paper when it is hung up.
7) Copy your group's final concept map in the space below.

Copy your group's final concept map here (you may turn the paper to the side also):

Appendix: Ethical Dilemmas

Ethical Discussion

Should Individuals Who Have "Drug Addiction Genes" Be Treated Differently?
From the Scholastic website article, The Role of Genes and Drug Addiction
http://teacher.scholastic.com/scholasticnews/indepth/headsup/intro/index.asp?article=genes

A major finding about the genetics of drug addiction was reported in 2004 by investigators at Duke University Medical Center. The researchers were able to identify a specific protein—PSD-95—that had a relationship to drug addiction. Mice that had low amounts of PSD-95 were much more sensitive to cocaine (than mice with normal amounts of PSD-95).

The researchers concluded that mice with normal amounts of PSD-95 were less likely to become addicted to cocaine (than mice with low amounts of PSD-95). According to Marc G. Caron, Ph.D., an investigator who was part of the research team, PSD-95 "likely plays a role in addiction to other drugs—including nicotine, alcohol, morphine, and heroin.‖

Consider the following scenario:

The case of Jacob puts the dangers of drugs in focus. Jacob began using OxyContin (a prescription drug for pain) at 18. Eventually Jacob moved from OxyContin to heroin. –If I'd never touched OxyContin, I wouldn't have done heroin,‖ he claims. Before long, Jacob was selling OxyContin pills to help support his habit. (Selling prescription drugs makes you a drug dealer and subject to criminal prosecution.) One day Jacob didn't have money to buy heroin. Desperate, Jacob robbed the local liquor store. Fortunately, no one was hurt but Jacob was arrested. Now he faces criminal charges from robbery to drug dealing. The judge has ordered Jacob to go to rehab to help him with his addiction. Jacob's lawyer read about a —Drug Gene‖ and has asked Jacob to get tested to see if he is genetically sensitive to drug addiction, meaning if he has the —Drug Gene‖ his body would produce low levels of the PSD-95 protein.
You are asked by the judge in Jacob's case to act as an adviser. Your job is to discuss the topic of individuals that have these —Drug Genes‖ and to make recommendations about how Jacob's case should be handled once his genetic test comes back. Consider the following:

A) It is estimated that genes contribute about 60% of a persons' vulnerability to drugs, an individual's environment contributes the other 40%. That means drug gene sensitive people are not –doomed‖ if they have this –drug gene‖ but they might want to be aware of the gene and its potential dangers.
B) Testing every drug addict for the —Drug Gene‖ will cost millions of dollars
C) Addiction adds to crime rates and violence.
D) If an individual tests positive for the —Drug Gene‖, what is their responsibility as an individual? Should they go to jail? Should they be forced to get help? Should they be let go because it's a –genetic–problem?

Use the ethical decision framework that follows to make a decision about what course of action the judge should take with Jacob if he is found to have the ―Drug Gene.‖ Should the results of Jacob‘s ―Drug Gene‖ test affect what happens to Jacob? Should Jacob go to jail? Rehab? Counseling? Other?

Your class will now work together using ethics to try to make a recommendation to the judge about what action(s) he/she should take.

Ethical Decision-Making Framework

What do you think is the ethical problem?

What are relevant facts? (Consider what you read above and what you have learned about the ―Drug Gene‖.)

What are questions that are still unknown?

Here are some potential actions the judge could take.

- Send Jacob to jail
- Send Jacob to rehab

List three more actions the judge could take:

What stakeholders should you consider?

What stakeholder did your teacher assign you? _____

What do you think are the concerns of your stakeholder?

What action does your stakeholder think the judge should take?

Explain why your stakeholder has chosen that action:

Now as a whole class, fill out this chart with all the stakeholders and their actions.

Who are the stakeholders?	**What action is this stakeholder recommending?**

What action(s) do you recommend the judge should take? Why?

Resources
Scholastic, The Role of Genes in Drug Addiction
http://teacher.scholastic.com/scholasticnews/indepth/headsup/intro/index.asp?article=genes

Ethical Discussion

Should Employees Undergo Genetic Testing to See if They Are Sensitive to Certain Chemicals They Might be Exposed to on the Job?

Beryllium (**Be**) is found naturally in food and water, but high **Be** exposure is mostly caused by working with **Be** in factories where **Be** dust can be easily inhaled. Beryllium is used in such products as cell phones, aircraft engine parts and nuclear workers use them to make triggers for nuclear bombs.

Some people who are exposed to **Be** develop a disease called Chronic Beryllium Disease (CBD). CBD is a disease that is characterized by an accumulation of T cells in the lungs. T cells help the body fight infection. When there are too many T cells in the lungs they damage the lungs and cause shortness of breath and dry coughing.

Changes in a gene called, HLA DP (**h**uman **l**eukocyte **a**ntigen) causes people to be sensitive to exposure to **Be**. HLA DP has the instructions to make a protein that helps T cells do their job. When scientists looked at how the change in the HLA DP gene would affect the protein made, they discovered that people who are sensitive to **Be** have a different sequence of amino acids. Only a small portion (1-15%) of the population is **Be** sensitive.

What do you think is happening to the protein to cause some people to be sensitive to Be?

Read the following article about contract workers in the California based Livermore Lab.

Beryllium Exposure Warning Arrives Too Late for Contract Workers at Livermore Lab
http://www.chronicberylliumdisease.com/news/nw_020808_contract_workers_livermore_lab_ptr.htm (edited)

February 8, 2008 — At Lawrence Livermore Labs in California, as many as 178 GSE Construction workers may have been exposed to the known carcinogen (cancer causing) beryllium, a toxic metal that can cause lung cancer and chronic beryllium disease. Results from a routine beryllium test revealed dangerously high levels of beryllium as early as July 2007. GSE contract workers may have been unknowingly exposed to beryllium and placed at risk of developing beryllium related diseases (CBD).

Normally, when exposures to **Be** becomes known, the policy is to test workers for beryllium sensitivity, to make sure the workers are not at risk of developing beryllium related diseases and to prevent further beryllium exposure.

Since the Livermore lab does not plan to get rid of the beryllium because it will cost millions of dollars, they have chosen to test all of the exposed workers to see if they have the gene for **Be** sensitivity. If the test comes back positive, how should the workers be treated?

They have asked you to advise them on how to treat all their workers fairly. Should the **Be** sensitive workers be fired? Should the **Be** sensitive workers get extra health insurance? Should **Be** sensitive workers be forced to wear protective gear? Should the company they work for help them find a different job?

Use the ethical decision framework that follows to make a decision about what course of action the Livermore lab should take.

Ethical Decision-Making Framework

What do you think is the ethical problem?

What are relevant facts? (Consider what you read above and what you have learned about beryllium sensitivity.)

What are questions that are still unknown?

Here are some potential actions the Livermore lab could take.

- Offer extra health care to the people who are sensitive
- Fire the workers who are sensitive
- Treat all workers the same (no one is fired and all workers get the same amount of health care and the same protective gear)

List three more actions the Livermore lab could take:

What stakeholders should you consider?

What stakeholder did your teacher assign you? _____

What do you think are the concerns of your stakeholder?

What action does your stakeholder think the Livermore lab should take?

Explain why your stakeholder has chosen that action:

Now as a whole class, fill out this chart with all the stakeholders and their actions.

Who are the stakeholders?	**What action is this stakeholder recommending?**

What action(s) do you recommend the Livermore labs take? Why?

Resources

Canadian Center for Occupational Health and Safety, Beryllium Disease
http://www.ccohs.ca/oshanswers/diseases/beryllium.html

****Beryllium Network, Beryllium Exposure Warning Arrives Too Late for Contract Workers at Livermore Lab
http://www.chronicberylliumdisease.com/news/nw_020808_contract_workers_livermore_lab.htm

CDC Public Health Genomics, HLA-DPB1 and Chronic Beryllium Disease
http://www.cdc.gov/genomics/hugenet/reviews/beryllium.htm