1.1. The Mystery Study Guide

1.1.a. What causes death?

The failure of one system can cause failure of next→ ending in lack of brain activity

1.1.b. What clues may be found at a scene of a mysterious death that may help to determine the cause of death?

Vomit, blood, fingerprints, DNA, saliva, bite marks, bullets, poison, etc

1.1.c. If someone was interested in a career with responsibility to determine the cause of death, what careers should he or she consider and investigate?

Forensic Pathologist (Medical Examiner)	Medical doctor that primarily does autopsies and determines cause of death
Toxicologist	PhD (usually) who tests body fluids (blood, vitreous humor, urine) for presence of toxins & medications to help determine cause of death
Coroner	Elected official that works with police & helps decide whether to have autopsy & whether a crime has been committed

1.1.d. What are examples of human body **systems**? 1.1.e. What **organs** make up the different body systems?

Urinary *"place for pee"*	**Nervous** *"full of nerves"*	**Endocrine** *"secrete within"*	**Digestive**	**Respiratory**	**Cardio.** *"heart & small vessels"*	Immune
Kidneys Ureters Bladder Urethra	Brain Spinal Cord Peripheral Nerves Sense organs (taste buds, ears, eyes, etc)	Pancreas **Thymus** Thyroid Pituitary Pineal Gland Adrenal Glands *("toward kidneys")* Ovaries/Testes	Teeth/Tongue Salivary Glands **Pharynx** Esophagus *("eater within)* Stomach Small Intestine Large Intestine (Colon) Rectum Liver Gall Bladder	Nasal Cavity **Pharynx** Larynx Trachea *("air tube")* Bronchus Bronchiole *("little bronchus")* Alveoli*** *("hollow")* ****then back the opposite direction*	Atria *("entrance halls for blood")* Ventricles *("bellies" that pump out blood)* Veins Venules *(little veins)* Arteries Arterioles *(little arteries)* Capillaries *("hairs"→place arteries turn to veins)* Erythrocytes *("red cells")*	Bone Marrow **Thymus** Spleen Lymph Nodes *("water knots")* Tonsils Leukocytes *("white cells")* **Appendix**

1.1.f. What are examples of interactions between body systems?

Urinary	Filters waste out of blood, removing cellular waste from all systems
Nervous	Tells other systems what to do via electrical signals (i.e. signal to poop)
Endocrine	Secretes hormones that signal other systems to do things (i.e. hunger)
Digestive	Absorbs nutrients (small intestine) to feed all other systems
Respiratory	Brings in oxygen needs by all cells and removes carbon dioxide waste
Cardiovascular	Transportation system→brings nutrients, hormones, O_2 to all systems, carries waste away
Immune	Protects us by preventing, trapping and killing pathogens *("disease starters")*
Skeletal	Provides structural support, protects soft organs (i.e. heart) & makes blood cells

1.1.g. What might be the consequence of malfunctions in any of the body systems?

Urinary	Waste will build up, killing person (kidney failure) unless they have dialysis
Nervous	Miscommunication causes problems like paralysis, Parkinson's, epilepsy, etc
Endocrine	People can experience gigantism, thyroid disorders, clotting disorders (hemophilia), etc
Digestive	Celiac disease, Crone's disease, etc can interfere with absorption of nutrients
Respiratory	Cystic fibrosis or infections can cause fluid build-up, person can drown
Cardiovascular	Vision loss or limb loss if blood doesn't circulate to those areas (like in diabetes)
Immune	Autoimmune disorders if it's overactive, inability to fight disease if it's underactive
Skeletal	Can't fight disease if not making WBCs, can't circulate oxygen if not making RBCs

1.1.h. What is a system?

Parts that work together to do a job (i.e. a SCHOOL system, a BODY system, a COMPUTER system☺) In the case of human body systems, similar **cells** make up **tissues** and the tissues form organs, which work together.

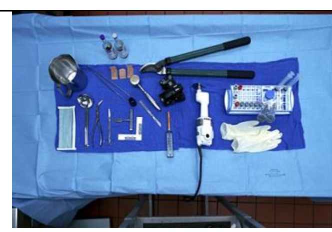
1.1 i. Are all sources of information accurate and reliable?

Sources are listed as **citations** in a **bibliography**. **Primary sources** are firsthand accounts (Darwin's Origin of Species & **secondary sources** reference primary sources (modern biology books). We should always cite all sources used to avoid **plagiarism**. Students use sources, but **outline** and summarize and rewrite the information to show their own understanding when writing answers to conclusion questions.

1.1.j. How can you tell if information on the Internet is accurate and reliable?

the ONION® AMERICA'S FINEST NEWS SOURCE™	Anything with answers in the name are NOT (i.e. Yahooanswers, wikianswers, etc). If anyone can post there, it's NOT reliable. Government sites (.gov) and educational pages (.edu) are usually MOST reliable. Information is usually reliable if the SAME answer can be found on MULTIPLE sites (that's why it's good to have **documentation** of at least 2 sources).

1.1.k. What is an autopsy and how can it be used to determine the cause of death?

A medical examiner opens up the body cavities, weighs and examines organs, extracts fluids for a toxicologist to analyze. **Autopsies** *("self eyes")* let medical examiners see things with their own eyes. It's also called a post-mortem *("after death")* and used to determine cause of death. Autopsies on people who have been murdered fall into the category of **forensic** *("crime")* **science**.	

1.1.l. Why is confidentiality of patient information important? 1.1.l. Who should keep patient information confidential?

Everyone deserves privacy to prevent embarrassment and possible damage to relationships or loss of job. **Biomedical scientists** (EMTs, Medical Examiners, any kind of doctor, nurses, pharmacists, etc,) can be fired or sued or even lose their license for violating HIPAA.

1.1.m. Is there ever a time when patient confidentiality should be broken?

Patient confidentiality can be broken for a patient who is under 18 (parents have rights to their info) or someone who signed a release form (for a spouse, etc, to have access) or in cases of suspected abuse

2.2 Heart Anatomy Study Guide by Hisrich

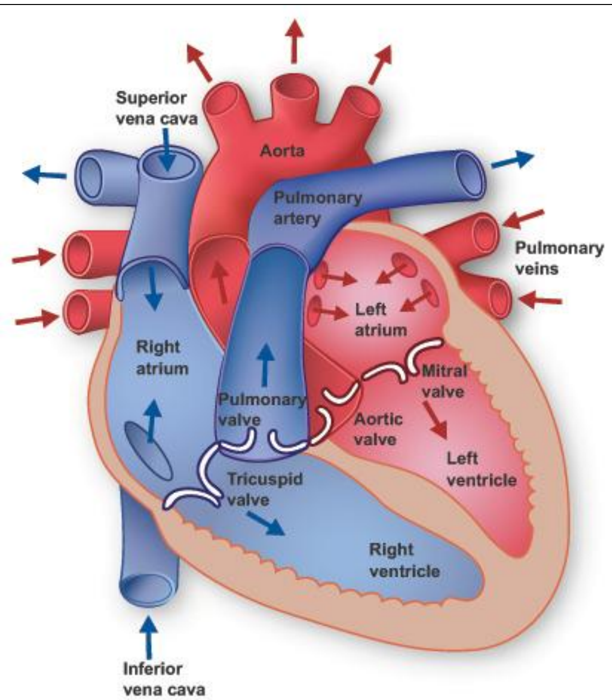
2.2.a. Why is the heart considered a pump?

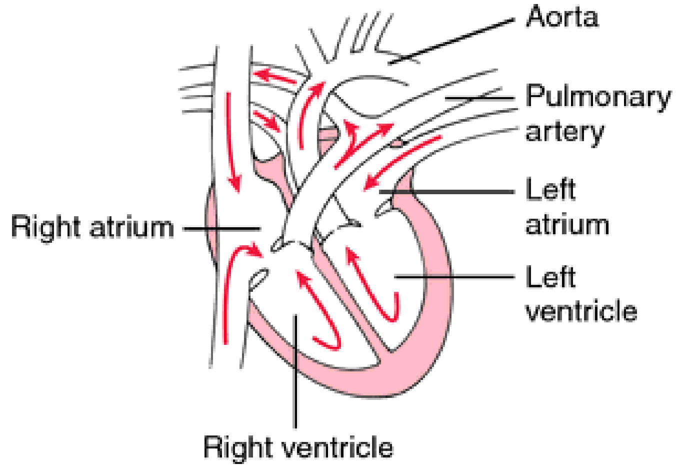
Pumps move fluids using pressure	The heart is a pump because it moves a fluid (blood) using pressure (contractions of ventricles). The heart powers the whole **cardiovascular system**.

2.2.b. What are the structures that make up the human heart?

Category	Characteristics	Includes
chambers	open, like rooms—hold blood	right and left **atrium** and right and left ventricles
valves	flaps, like doors—let blood move one way	**tricuspid**, **bicuspid (mitral)**, **aortic** & pulmonary
veins	bring blood to heart—tubes, like halls	**superior & inferior vena cavas** & pulmonary veins
arteries	carry blood from heart—tubes, like halls	pulmonary arteries, **aorta**

2.2.c. How are these structures organized?



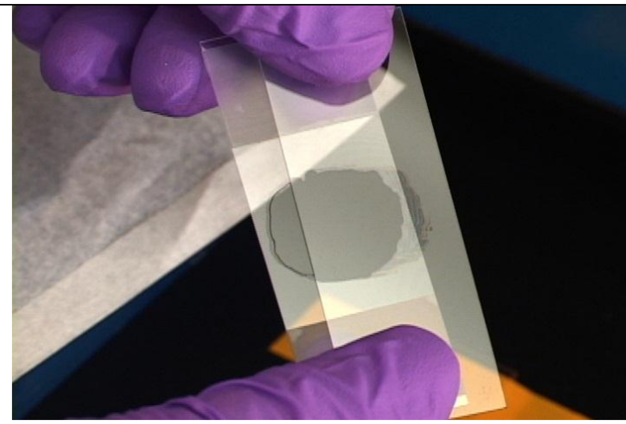


2.2.d. What is the pathway blood takes as it passes through the heart?

right atrium→**tricuspid valve**→right ventricle→pulmonary **valve**→pulmonary arteries→lungs for oxygen→pulmonary veins→**left atrium**→**bicuspid (mitral) valve**→left ventricle→**aortic valve**→**aorta**→arteries all over body→arterioles→capillaries (to drop off oxygen, nutrient & hormones & pick up waste & CO_2)→venules→veins→**vena cavas**→right atrium

2.2.e. What is meant by the term ***tissue***?

A **tissue** is a group of **cells** that work together to do something. Tissues make up organs, such as blood vessels and the heart. People who study tissues & make slides from them are called **histologists** *("one who studies tissues")*

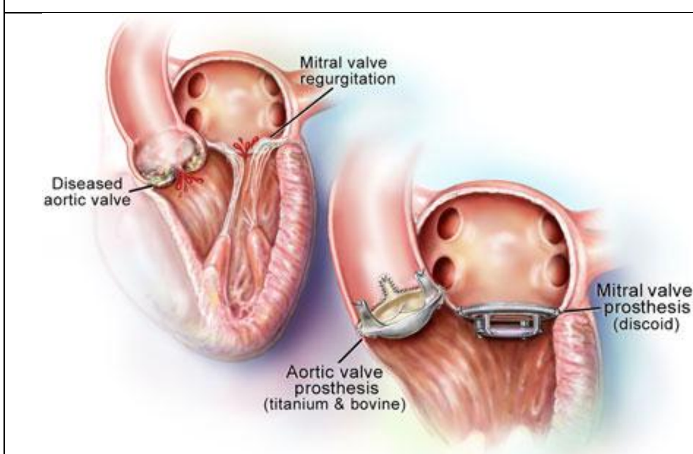


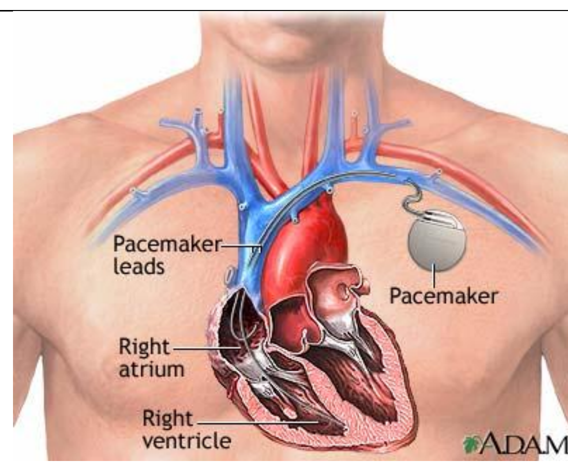
2.2.f. What are the different types of cardiac **tissue** and how do they differ?

Tissue Name	Literal Meaning	What it Does
myocardium	*"muscle heart"*	makes up thick muscle layer
endocardium	*"inside heart"*	forms inner surface of chambers
pericardium	*"on heart"*	sac that surrounds the heart

2.2.g. How do principles of engineering apply to heart structure and function?

The heart is a pump, just like artificial pumps created by engineers.	Biomedical Engineers create things like artificial heart valves & pacemakers to help treat heart problems.





2.3 The Heart at Work Study Guide by Hisrich

2.3.a. In what ways can technology be used to collect and analyze cardiovascular data?

Thing measured	Tool used	Used how
Blood pressure	**Sphygmomanometer** *"pulse measurer"*	Determines systolic and diastolic arterial pressure
Heart rate	Timer	Used to find beats during 10 seconds (then multiply by 6 for bpm)
Electricity within heart	**EKG**	Electrodes on skin pick up current and show graphically

2.3.e. What is an EKG? 2.3.f. How can an EKG be used in the diagnosis and treatment of heart disease?

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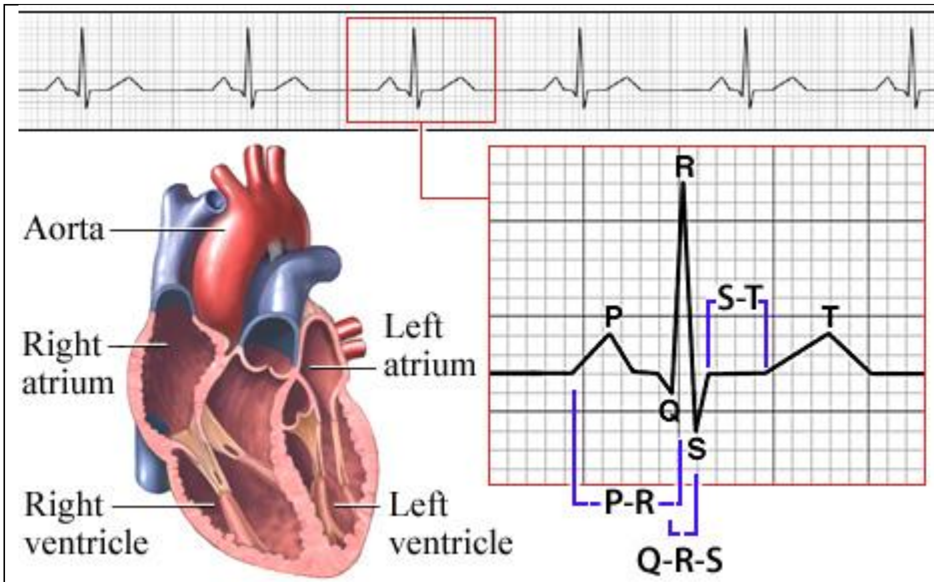

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<td>

EKGs, or **electrocardiographs** *("electricity heart pictures")* measure the heart's electrical activity and display it in the form of a picture:

- P wave→signal passes from **SA node** (sinoatrial node) to AV node (atrioventricular node), moving across atria
- QRS interval→signal passes from AV node through Purkinje fibers & the ventricles contract
- T wave→the ventricles repolarize & the heart is relaxed

EKGs are examined for missing, extra or malformed waves.
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EKGs are taken when heart problems are suspected and can be used in **cardiology** *("the study of the heart")* to diagnose heart attacks, lack of blood flow to the heart, arrhythmia *("no rhythm")*, lack of forcefulness of heart muscle, muscle parts that are too thick or heart parts that are too big, birth defects of the heart, heart valve diseases.
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</table>

2.3.b. What factors can influence heart rate?

<table>
<tr><td>Lower heart rate</td><td>Raise heart rate</td></tr>
<tr>
<td>

Short-term method

- Sleeping or relaxing
- Hydrating yourself

Long term method

- Exercise
- Reducing stress
- Eating fruits, vegetables, nuts, beans & fish
</td>
<td>

Short-term method

- Exercising or other rapid movements
- Being scared or very stressed briefly
- Drinking caffeine or alcohol

Long term method

- Being out of shape
- Increasing stress
</td>
</tr>
</table>

***Factors that influence heart rate can be determined by creating a **hypothesis** (testable prediction) and then testing it out, like we did in class.

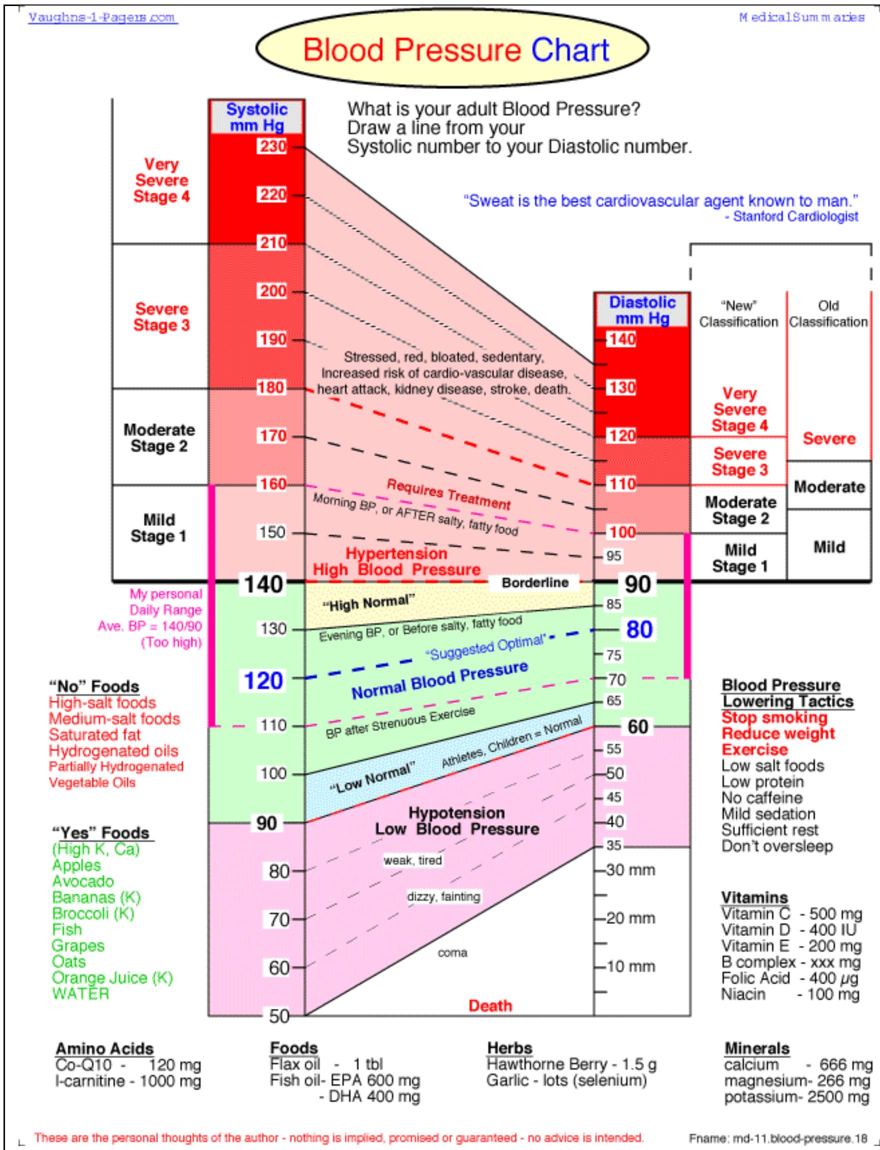
2.3.c. What is the relationship between **blood pressure** and cardiovascular function?

A normal **blood pressure** reading usually indicates a healthy heart, with higher readings indicating that the heart is stressed. It measures the **pressure** on vessel walls from the movement of blood particles.

- Top number→**Systolic**→pressure in arteries as the ventricles contract & the chambers emptying (always higher)
- Bottom number→**Diastolic**→pressure in arteries when ventricles are relaxed & the chambers are filling with blood (always lower)

Blood Pressure Category	Systolic mm Hg (upper #)		Diastolic mm Hg (lower #)
Normal	less than 120	and	less than 80
Prehypertension	120 – 139	or	80 – 89
High Blood Pressure (Hypertension) Stage 1	140 – 159	or	90 – 99
High Blood Pressure (Hypertension) Stage 2	160 or higher	or	100 or higher
Hypertensive Crisis (Emergency care needed)	Higher than 180	or	Higher than 110

2.3.d. What factors can influence blood pressure?



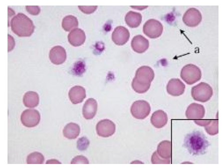
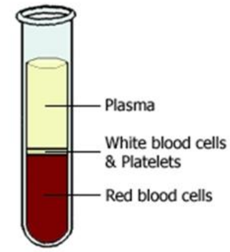
According to the Mayo Clinic, the top 10 ways to reduce your blood pressure or prevent hypertension *("extra tension")* **are to**:

1. **Lose extra pounds and watch your waistline**
2. **Exercise regularly**
3. **Eat a healthy diet**
4. **Reduce sodium in your diet**
5. **Limit the amount of alcohol you drink**
6. **Avoid tobacco products and secondhand smoke**
7. **Cut back on caffeine**
8. **Reduce your stress**
9. **Monitor your blood pressure at home and make regular doctor's appointments**
10. **Get support from family and friends**

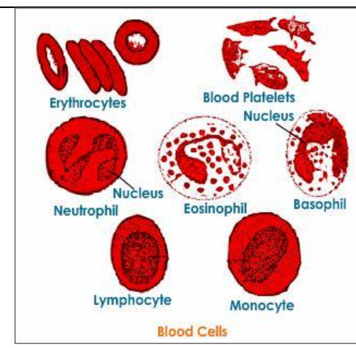
http://www.mayoclinic.com/health/high-blood-pressure/HI00027

2.4 Blood: The River of Life

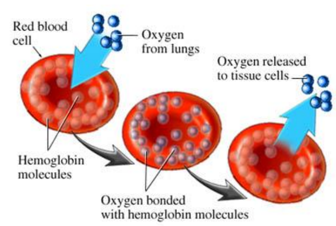
2.4.a What is the general composition of human blood?

Erythrocytes ("red cells")	Leukocytes ("white cells")	Thrombocytes ("clotting cells")	Plasma ("former")	
	 		 Liquid that carries all cells, as well as hormones, nutrients, salts, etc	
No nucleus, made in bone marrow by stem cells				

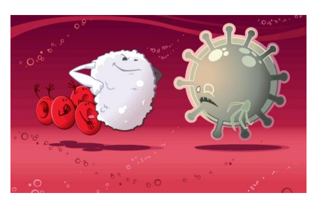
2.4.b. Why is blood classified as a tissue?

Tissues are groups of cells that perform a similar function and have a common origin. Blood transports materials throughout the body and is all made in the bone marrow. The study of tissues is **Histology** ("tissue study") and the person who makes slides is a **Histology Technician**. The removal of tissues from the body is called a **Biopsy**.	

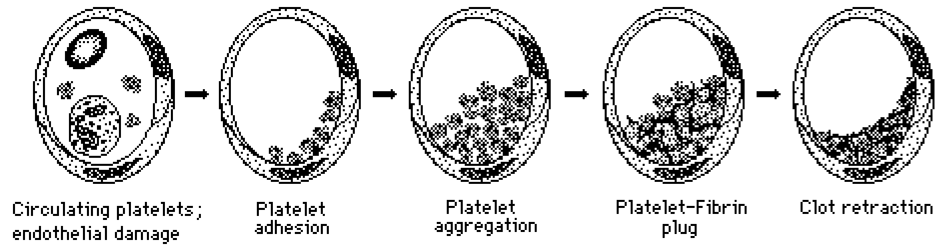
2.4.c What are the characteristics and function of red blood cells?

7-8 µm wide Flat for increased S.A. Carry oxygen, using **hemoglobin** ("blood balls") protein Survive ~ 4 months	

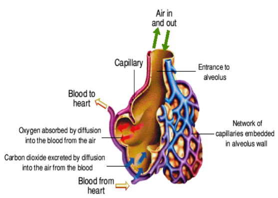
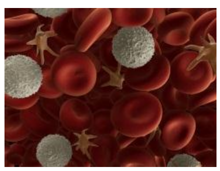
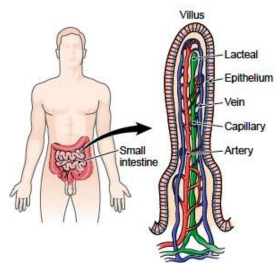
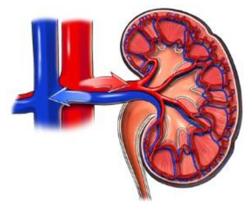
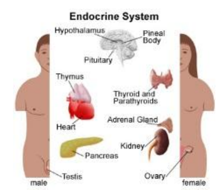
2.4.d. What are the characteristics and functions of white blood cells?

12 -25 µm wide Fight diseases Part of immune system A high count can indicate infection	

2.4.e. What are the characteristics and function of platelets?

Flat and sticky Help blood clot Impaired by Aspirin	

2.4.f. In what ways does blood directly relate to other human body tissues and systems?

Respiratory System	Immune System	Digestive System	Urinary System	Endocrine
				
Picks up oxygen and drops off carbon dioxide at the alveoli	Circulates white blood cells and antibodies to fight disease	Picks up nutrients from the small intestine and carries them to all other body tissues	Carries cellular waste from all the body's tissues to the kidneys, where it is filtered out and removed in the urine.	Picks up hormones from all the endocrine organs and carries them to all other tissues

2.4.g. Why are most cells so small?

<table>
<tr>
<td>Cells must be small enough that materials coming IN can absorb all the way IN
1. Oxygen
2. Nutrients
3. Water
and materials going OUT
4. Carbon dioxide
5. Cellular waste
can escape

If cells get TOO BIG they'll either starve to death or drown in their own waste</td>
<td>Cells need to MAXIMIZE their surface area in relation to their volume (red blood cells do this by being somewhat flat)

In our experiment, the agar cubes represented cells and the sodium hydroxide represented nutrients, oxygen and water. The small cells were penetrated more quickly and effectively than the large cells.</td>
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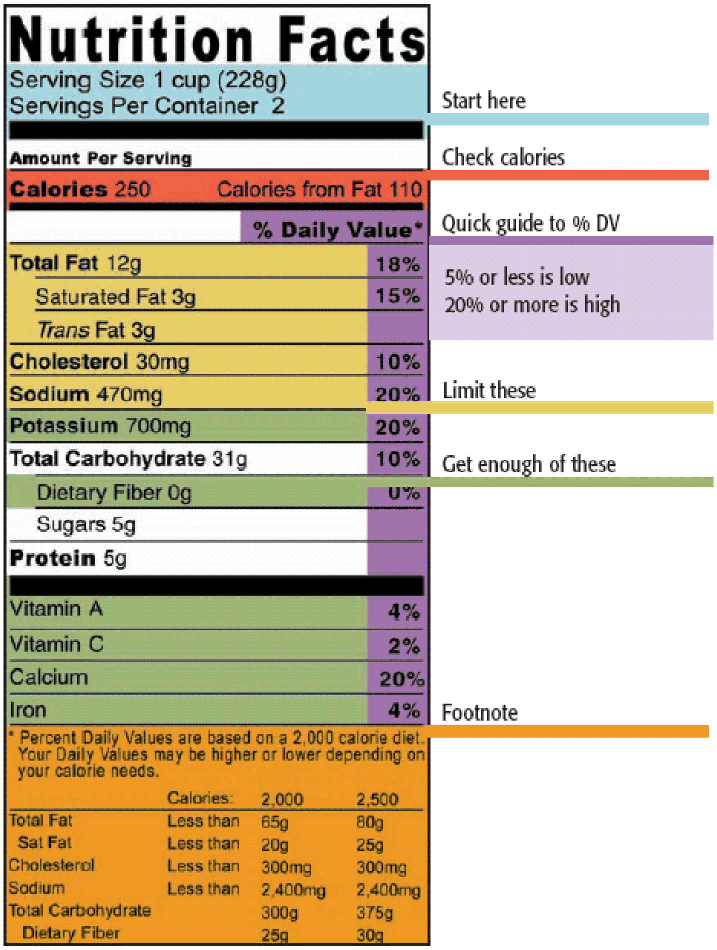
3.1 What's in our Food? Study Guide by Hisrich

3.1.a. What are the nutrients identified on food labels?



Vitamins and minerals—make sure to get enough, many can be obtained from fruits/vegetables and whole grains.

Serving Size—all values below it are for one serving

Calories—total energy

Total Fat—includes all fats, long term energy source & component of cells, supports brain (should be <30% of diet)

- Saturated fat—bad for heart, should be avoided
- Trans fat— worst kind, should be avoided
- Unsaturated fat—best kind, good for heart, calculate by subtracting other fats from total fat

Cholesterol—not necessary, liver makes it, limit intake

Sodium—table salt, limit to keep blood pressure healthy, but get some

Total Carbs— includes all carbs, quick energy source, whole grains are best (should be bulk of diet)

- Dietary fiber—from plants, not digestible, helps digestive system (helps you poop)
- Sugars— not necessary for health, limit them
- Starch—better than sugars, calculate by subtracting fiber and sugars from total carbs

Protein—needed to build muscle and repair cells, should be about 10% of total calories

3.1.b. How is the amount of energy in a food determined?

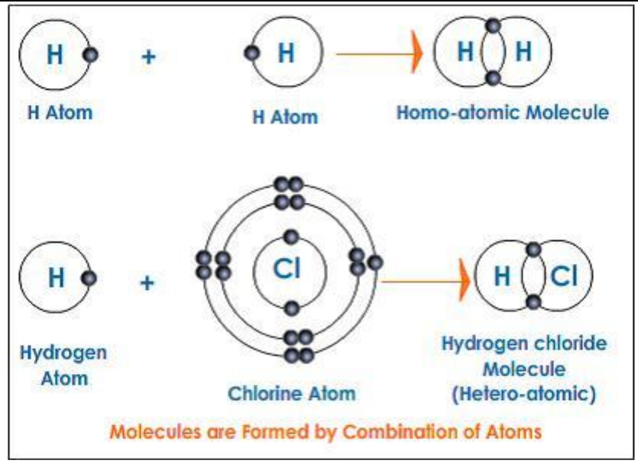
The energy in food is determined via calorimetry ("energy measurement"), in which a food item is burned to break the **chemical bonds** in the **compounds** that make up the food and the energy from the bonds is turned into heat/light & the heat is captured in water. The change in mass of the food and change in temperature of the water can be used to determine the Cal/g (energy per unit mass) of the food material. Food energy is measured in **calories** *("heat measurement"),* with one **calorie** being the amount of heat energy that will raise 1 g of water 1ºC.

3.1.e. What is the role of a **chemical bond** in energy transfers?

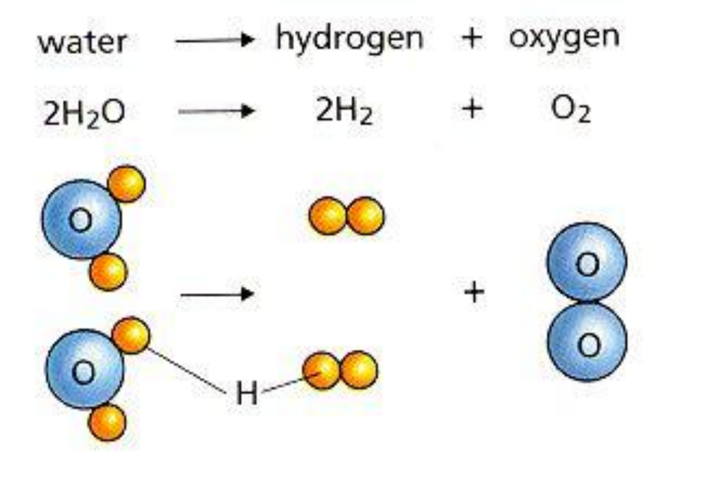
Energy is released when **chemical bonds** are broken in **chemical reactions**. For instance, the body gets energy from breaking down food **molecules** during digestion. Energy is also given off if the **molecules** are broken apart by burning the foods.

3.1.c. What is the basic structure of all matter?

Food is a type of matter because it has mass and takes up space. All matter is made up of **atoms** (there are about 100 different kind of **atoms** and different kinds of **atoms** are called "**elements**") & the **atoms** form bonds to make **compounds**. A **compound** in which the atoms share electrons is formed by a **covalent bond**. Most of the **nutrients** that living things take in and are made of are bound by **covalent bonds**. There's another kind of bond, called an **ionic bond**, in which the atoms are ions (have opposite charges from losing or gaining electrons) and are attracted because of their opposite charges. Materials like salts have ionic bonds & are formed by ions. Matter is usually a mixture of different **molecules** (different types of **compounds**).



3.1.d. What is a **chemical reaction**?



A **chemical reaction** is when a **molecule** forms from **atoms** coming together or when the bonds between the **atoms** are broken. Whenever that happens, there are signs that it has happened. One or more of the following will happen:

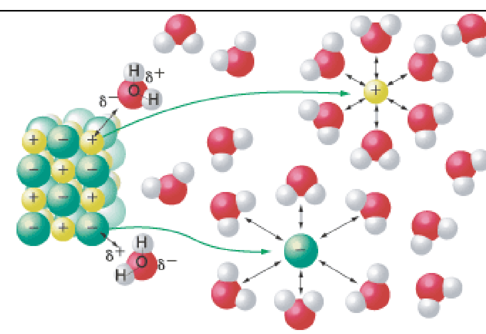
1. Energy change (it will glow and/or heat will be absorbed or released)
2. Color change (a new color will show up)
3. Odor change (a smell will be given off)
4. Precipitate (a solid will form from 2 liquids)
5. Gas produced (a gas will be given off)

3.1.f. What is the relationship between nutrients, food, **chemical reactions**, and energy?

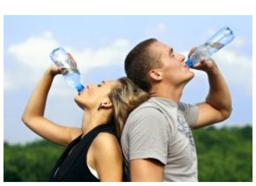
Food is made of nutrients & the nutrients are made of **molecules**. The **molecules** can be broken down through **chemical reactions**, giving off energy.

3.1.g. Why is water balance such an important factor in maintaining **homeostasis**?

Water helps maintain **homeostasis** ("staying same"), keeping fluid levels constant in the body. **Covalent bonds** hold the **atoms** WITHIN a water **molecule** together & the **polarity** of water (slight positive charge at one end and slight negative charge at the other) make water **molecules** attract to each other and attract the **ions** in salts, making water the universal **solvent**, dissolving more **solutes** than any other liquid & forming many different kinds of **solutions**. Salts are very **hydrophilic** ("water loving") & dissolve easily in water.



3.1.h. Are sports drinks a valuable tool in maintaining water balance?

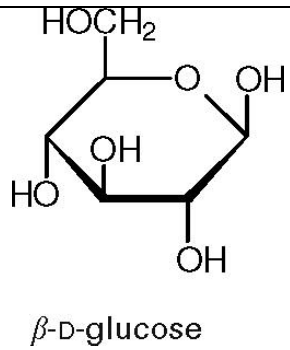


Sports drinks are really only useful for hardcore athletes, exercising for multiple hours a day. For most people, water is the best way to remain hydrated. Hardcore athletes may need the carbs from the sugars in sports drinks to maintain their energy levels and may need the electrolytes (salts) to keep their muscles working. But for most of us, we don't need the extra sugar & salts & should simply drink water.

3.2 Macromolecules Study Guide by Hisrich

3.2.a. What are the main structural components of carbohydrates, proteins and lipids?

Carbohydrates, **proteins** and **lipids** are all **macromolecules** ("big molecules") because they are very large and consist of lots of atoms. Nucleic acids (DNA & RNA) are also **macromolecules**.

<table>
<tr><th></th><th>Carbohydrates</th><th>Proteins</th><th>Lipids</th></tr>
<tr><td rowspan="2">Building Blocks</td><td>Monosaccharides
("single sugars")</td><td>Amino acids</td><td>Hydrophilic heads & hydrophobic tails (fatty acid chains)</td></tr>
<tr><td>


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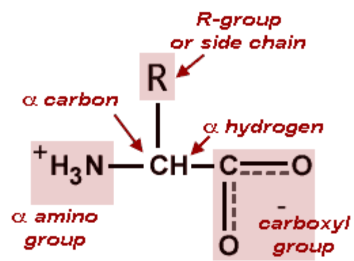

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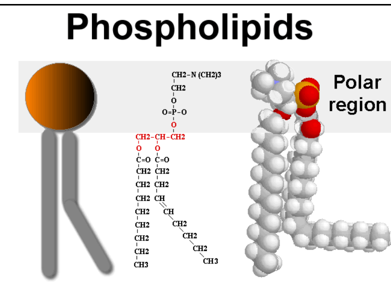
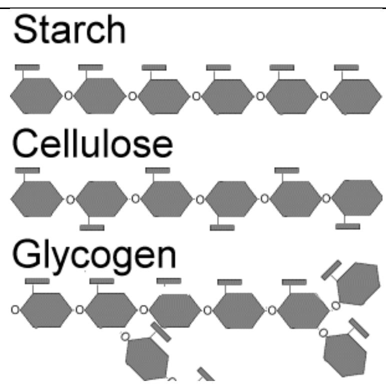


</td></tr>
<tr><td>Macromolecule</td><td>


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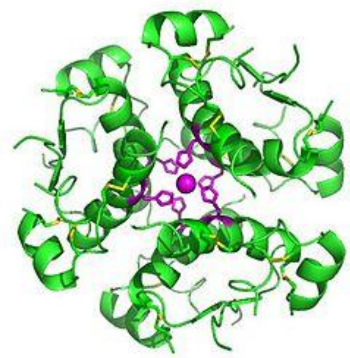
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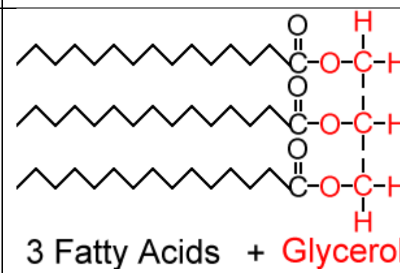

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Carbohydrates & **proteins** are **polymers** & have many parts. **Polymers** are made of **monomers** (simple building blocks). The **monomers** that make up carbohydrates are **monosaccharides** & the **monomers** that make up proteins are **amino acids**. Complex **carbohydrates** (starch and fiber) are considered **polysaccharides** *("many sugars")* because they have 3 or more **monosaccharides** that make them up.

3.2.c. What types of foods supply carbohydrates, proteins and lipids?

Carbohydrates	**Proteins**	**Lipids**
		
grains, fruits, veggies, dairy, sweets	meat, beans, nuts, eggs, dairy	meat, oils, nuts, veggies, dairy

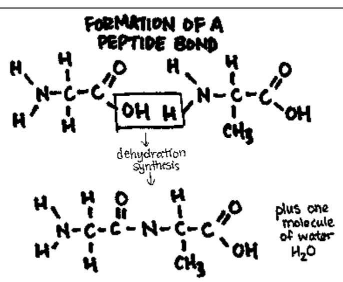
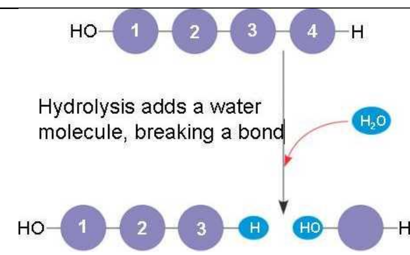
3.2.b. How do **carbohydrates**, **proteins** and **lipids** differ in structure and function? 3.2.g. How can **macromolecules** be detected in foods?

	General Characteristics	Primary functions	Detection via **chemical indicators**
Carbohydrates	Made up of carbon rings • **Monosaccharides** (one ring) ***fructose, **glucose** & galactose • **Disaccharides** (2 rings) ***lactose & sucrose • **Polysaccharides** (3+ rings) ***also called complex carbs ***starch, fiber & cellulose	Quick energy (complex carbs give longer term energy & unused carbs are converted to and stored as fat for long term storage)	**Monosaccharides** detected with Benedict's solution—*turns murky when heated when present* Starch (a **polysaccharide**) detected with iodine—*turns purple when present*
Proteins	Twisty, complicated, folded, like tangled yarn	Build body tissues, send chemical signals (as hormones), & heal/repair	Detected with Buiret solution—*turns purple when present*
Lipids	Have a "head" that is hydrophilic *("loves water")* and one or more fatty acid chain "tails" that are hydrophobic *("hate water")*	Store energy long term, allow nerves to function, cushion organs	Detected by rubbing against paper—*leaves greasy smear when present*

3.2.h. What are some of the limitations of **chemical indicators**?

If food has much color, it can cover up the color changes. For instance, a dark soda like Coke wouldn't get the kind of clear results that a clear liquid like Sprite would. Chocolate graham crackers would be hard to interpret a starch test on. Also, many indicators can tell what TYPE of molecule (i.e. Benedict's can tell simple sugars), but not narrow it down past that (was it glucose, fructose or galactose?).

3.2.d. What is **dehydration synthesis**? 3.2.e. What is **hydrolysis**? 3.2.f. How do **dehydration synthesis** and **hydrolysis** relate to food?

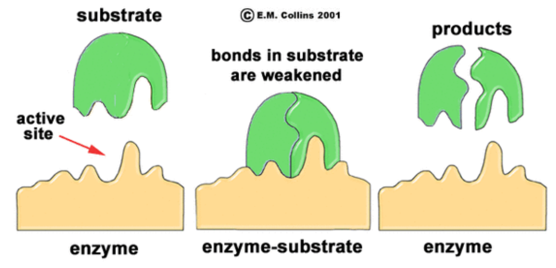
Dehydration synthesis *("remove water & come together")*	Comparison	**Hydrolysis** *("split with water")*
 This is the method by which **macromolecules** are built up in plants & animals—it's how things like complex carbohydrates & proteins form. As 2 molecules join to form one, they give off a water molecule. It's how things GROW. This requires **ATP** (energy).	They are exact opposites—infinitely reversible reactions	 This is the way **macromolecules** are broken down during digestion. Each water molecule can break one bond, breaking **polymers** down into **monomers**—for example polysaccharides (like starch) break down into simple sugars & proteins break down into amino acids. This results in the creation of **ATP** (energy).

****Note: **Electrolyte** was listed as a key term, but I cannot figure out how it fits in with ANY essential question! It's basically a fancy word for salts though & they're needed for metabolism and muscle movement (in small quantities).*

3.3 Molecules Working Together Study Guide by Hisrich

3.3. a. What is an **enzyme**?

Enzymes are types of proteins that initiative or speed up chemical reactions (known as **catalysts**), either breaking down or putting together molecules. Examples of important **enzymes** within the body are lactase, amylase, maltase, sucrase (names typically end in ase). **Enzymes** are taken in through food. **Enzymes** are not destroyed in reactions, so they can be used over and over again, much like a key. Every **enzyme** works best at a particular pH.



3.3. b. What is the general role of **enzymes** in the human body? 3.3. f. What are examples of **enzymes** found in the digestive system?

Carboydrases break down carbohydrates and include:

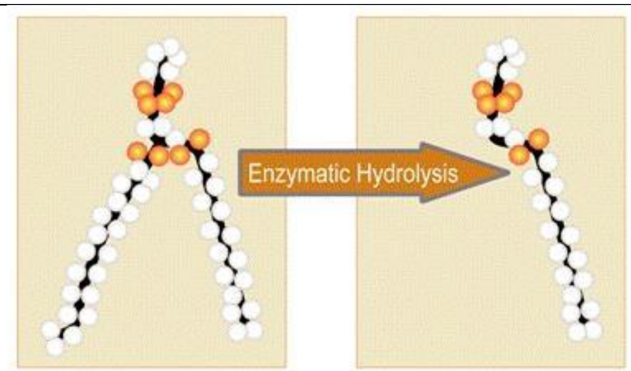
- amylase (breaks down starch)
- sucrase (breaks down sucrose)
- lactase (breaks down lactose)

Proteinases or proteases break down proteins, include:

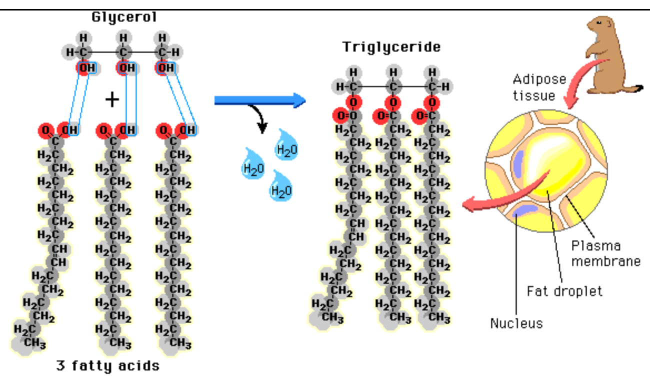
- pepsin (works in stomach)
- trypsin (active in small intestine)

Lipases break down lipids & includes:

- human pancreatic lipase (breaks down dietary fats)

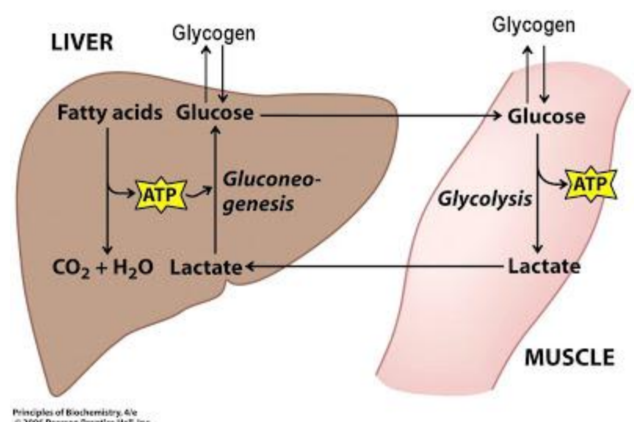


Some **enzymes** break down molecules (called **substrates**), usually during digestion. They facilitate the process of hydrolysis ("cutting with water").



Other **enzymes** bring molecules together to form larger molecules (see above). That's useful for building up tissues (and it's how we grow and how plants grow).

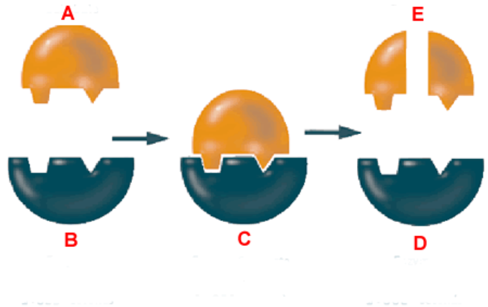
Glycogen synthase is an **enzyme** that builds glucose molecules into glycogen for storage in the liver. This is an example of dehydration synthesis.



3.3. d. Why are **enzymes** important to human health?

Enzymes allow us to break down molecules (digesting food or destroying pathogens) & also to grow and build body tissues and store energy. Humans could not SURVIVE at all without enzymes. Even missing a single enzyme can cause problems and potentially cause death.

3.3. c. How are **enzymes** able to function with such specificity?

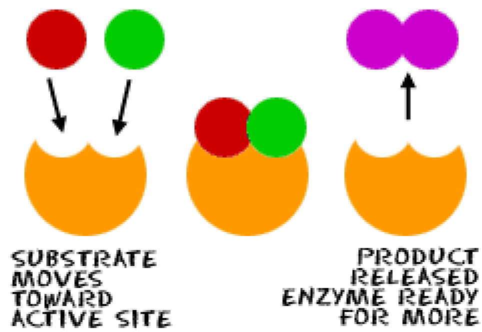


Each **enzyme** is specific to a particular **substrate** (the molecule that it acts upon) and they fit together like a lock and key.

The **enzyme** is on the bottom and substrate above.

← **substrate** breaking down

substrate coming together →



3.3. e. What might happen if an **enzyme** was missing or didn't work properly?

Enzyme deficiencies cause diseases and can even lead to death and interfere with **homeostasis**. Below are examples.	
Lactose intolerance	Lack of lactase **enzyme** makes it impossible for a person to digest lactose (milk sugars) & causes abdominal discomfort
Gluten intolerance	Lack of **enzymes** such as DPP-IV makes it impossible to digest the gluten from wheat & causes malabsorption (can cause death if not addressed)
Gaucher's disease	Lack of the glucocerebrosidase **enzyme** inhibits the breakdown of fatty acid glucosylceramide, causing it to build up. It can cause enlarged organs, liver malfunction, skeletal disorders & bone lesions, neurological problems, and can lead to death if not treated.

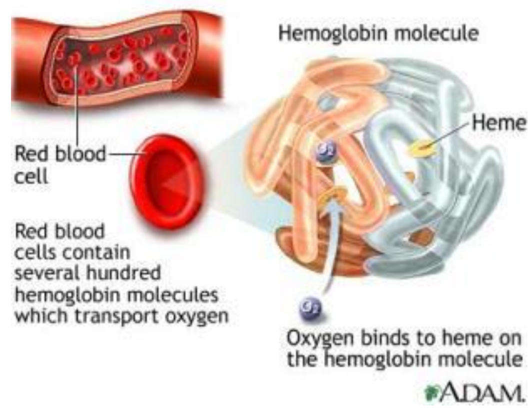
Most **enzyme** deficiencies can be treated with **enzyme** supplements (if they exist).

Lactose intolerance	Gluten intolerance	Animal product **enzyme** supplements for vegetarians
		

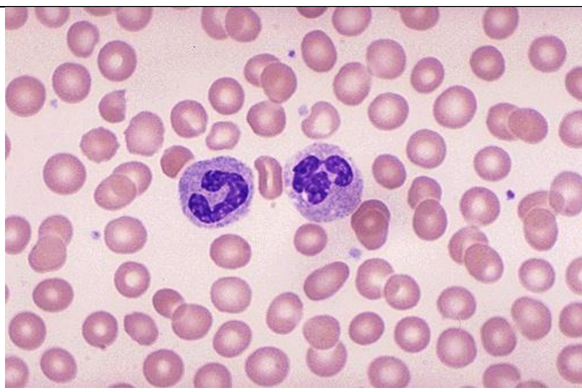
4.1 What is Sickle Cell? Study Guide by Hisrich

4.1.a. How do cells get the oxygen they need for energy production?

Erythrocytes (red blood cells) contain a **protein** called **hemoglobin** ("round blood")—hundreds of molecules of it, actually. **Hemoglobin** binds to oxygen, picking it up from the alveoli and dropping it off in capillary beds throughout the bodies tissues. **Hemoglobin** is also the **protein** that picks up the carbon dioxide waste produced by all cells and brings it back to the alveoli so that the respiratory system can remove it from the body.



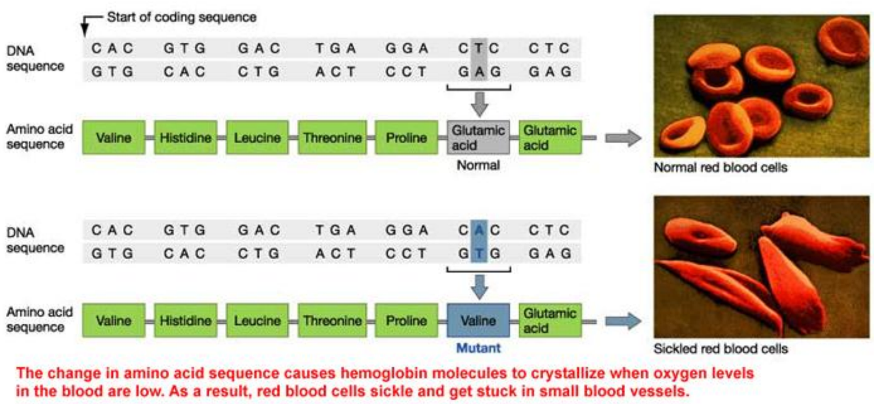
4.1.b. What do normal red blood cells look like when placed under a microscope?



Normal red blood cells are round, but sort of flat in the middle (to increase surface area). The slide shown left is from a person with normal red blood cells. Most of the cells shown are erythrocytes. The large ones in the center are leukocytes (white blood cells) and the specks are thrombocytes (platelets).

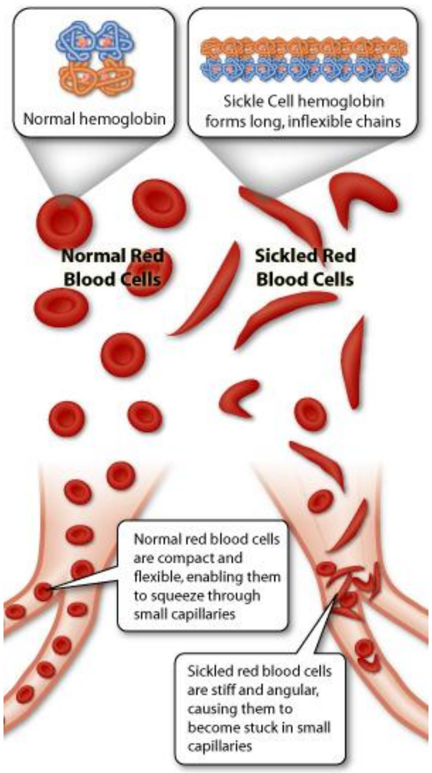
4.1.c. Why do some people have differently shaped red blood cells?

People with **sickle cell anemia** have red blood cells with an abnormal shape. They are called "sickle" because they are shaped like the cutting tool called a sickle.



Sickle cell anemia *("no blood")* is a recessive genetic **trait** that must be inherited from both parents. It's called anemia because the blood lacks normal **hemoglobin**. **Hemoglobin** contains two parts—**alpha globin** and **beta globin**. In a person with **sickle cell anemia**, there is a mutation in the **beta globin** portion, resulting in the substitution of ONE incorrect **amino acid**, causing the entire **protein** to fold incorrectly.

4.1.d. What effect does the altered shape of the red blood cell have on the health of the individual?

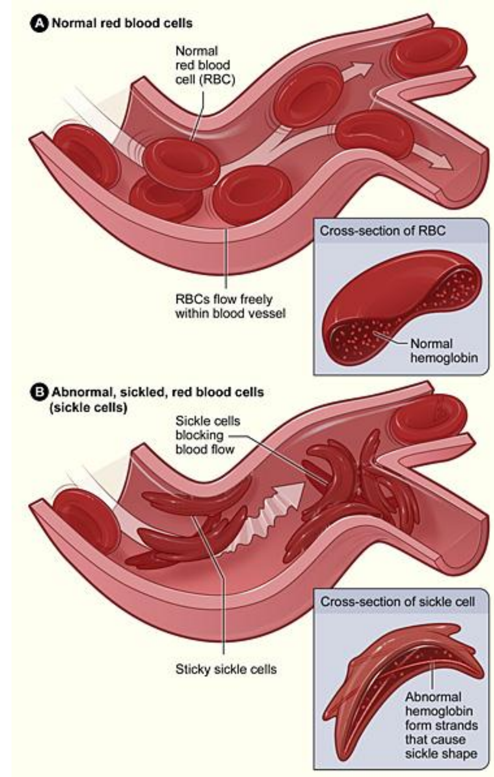


The **hemoglobin protein's** abnormal shape causes the red blood cells to have an abnormal shape. The can still carry oxygen, but they tend to get stuck in capillaries and make it difficult for blood to circulate to all the body's tissues.

The primary **symptom** of **sickle cell anemia** is pain, which is caused by lack of oxygen/nutrients to the body tissues.

Complications of **sickle cell anemia** include swelling of hands and feet, enlargement of the spleen, increased infections, acute chest syndrome (like pneumonia), eye problems & more.

The **prognosis** for a person with **sickle cell** is that there is no cure. Medications can treat the symptoms. Bone marrow and stem cell transplants can also reduce the effects.

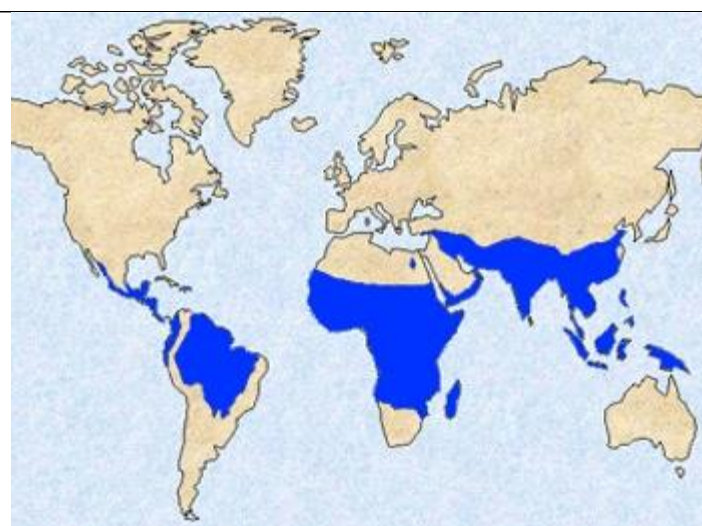


4.1.e. What is the difference between someone having the **sickle cell trait** and having **sickle cell anemia**?

Sickle Cell Trait	Both	**Sickle Cell Anemia**
Normal **hemoglobin**	Red blood cells have **hemoglobin**	Abnormal **hemoglobin**
Normal red blood cells	Have red blood cells	Sickle shaped red blood cells
Inherited one copy of the mutation	Inherited the gene from parent(s)	Inherited 2 copies of the mutation
No ill effects	Have protection from malaria	Many health complications
	Occurs mostly in people descended from those in the tropics	

4.1.f. Where in the world does **sickle cell** disease occur most often?

Sickle cell anemia occurs most in the tropics, in places like South America & Central Africa. It occurs there because the mutation protects from malaria, and malaria is a big killer in the tropics (temperatures allow mosquitos to thrive). There is no survival benefit to the trait in more Northern climates, so the disease wouldn't persist here. However, due to immigration **sickle cell** disease is a problem even in places like the United States. It is a problem in people whose ancestors come from the affected regions (African Americans, Indian Americans, Middle Eastern immigrants, South American immigrants, etc). **Sickle cell** is virtually non-existent in people of European descent.



4.2 What Causes Sickle Cell? Study Guide by Hisrich

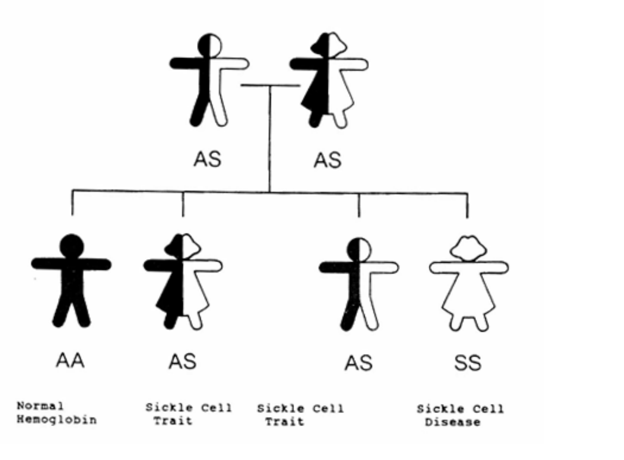
4.2.a. How does someone get sickle cell disease?

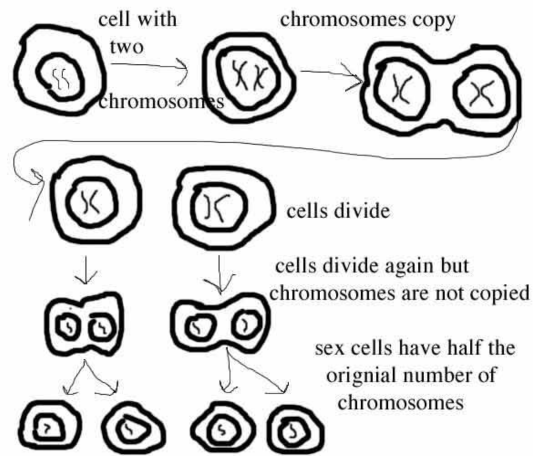
They must inherit the **recessive trait** (**mutation**) from BOTH parents.

4.2.b. Can sickle cell disease spread from one person to another the same way as a cold or the flu?

No. It's **hereditary** (passed from parents), NOT **contagious** like an infectious disease.

4.2.c. How are diseases inherited from parents? 4.2.i. Why does sickle cell disease run in families, yet is not present in every generation?



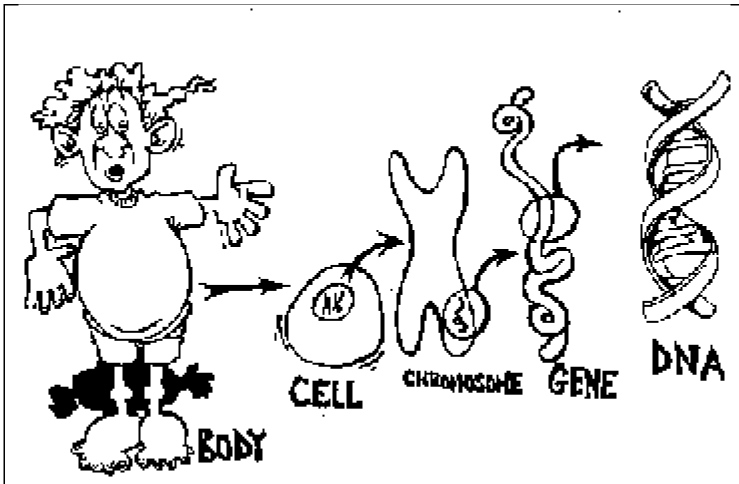


All of our **genetic material** comes from our parents—half from mom and half from dad. Sex cells (eggs and sperm) are produced through meiosis & always contain HALF of the parents' genetic material. We each have 2 copies of every **gene**, one from each parent. When we have kids, we randomly pass them ONE of our two **genes** and our partner does the same. In the case of a recessive genetic disease like sickle cell anemia, a person must inherit the **mutation** from her mother AND father to have the disease. If they only inherit the mutation from ONE parent, the **dominant trait** (which is NORMAL) will be expressed and the person will have sickle cell trait (not sickle cell disease). That's why diseases like sickle cell can skip generations.

4.2.d. What are examples of human diseases that are inherited?

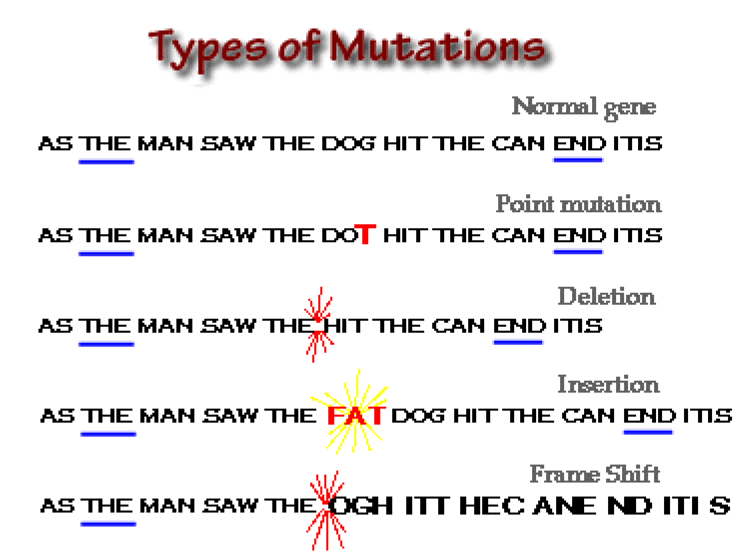
Other genetic disorders include cystic fibrosis, Huntington's disease, triple X syndrome, Duchenne muscular dystrophy, Down's Syndrome, Cry of the Cat, color blindness, hemophilia, Tay Sachs, Turner Syndrome, Polycystic kidney disease and many more.

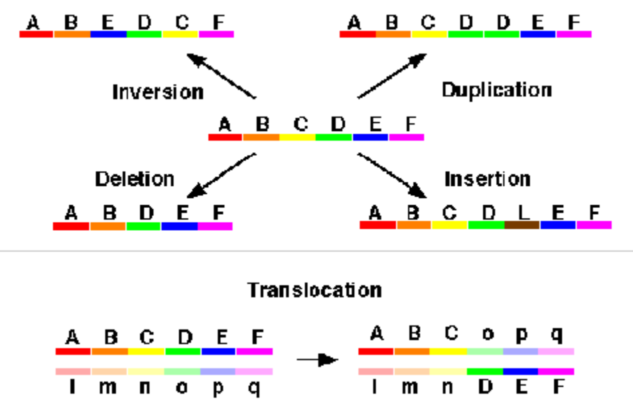
4.2.e. What is a **chromosome**? 4.2.g. How many **chromosomes** do humans normally have?



Chromosomes are made up of many genes that are all connected together in a long strand of DNA. Humans normally have 46 **chromosomes** (23 from mother and 23 from father), though it's possible to survive with 45 IF the missing **chromosome** is the **sex chromosome** X and the person is a female (Turner Syndrome). Otherwise the loss of a **chromosome** causes death. People can also survive with an extra **chromosome** (depending on the chromosome). Examples are Triple X Syndrome, Down Syndrome (Trisomy 21), Klinefelter's Syndrome (XXY) or Superman Syndrome (XYY). If an **autosomal** (non-sex) **chromosome** is missing, it is ALWAYS deadly & if there is no **sex chromosome** X (and the person has only a Y) it is ALWAYS deadly.

4.2.f. What is a **mutation**?

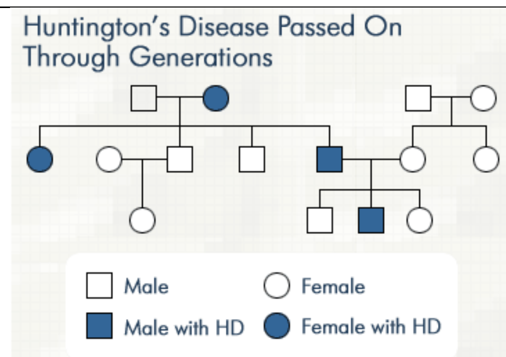




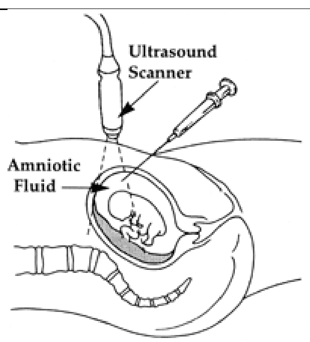
Mutations are mistakes in the DNA code. They can be caused by exposure to UV light, exposure to radioactive material, exposure to x-rays, aging, or just bad luck.

4.2.h. How are **pedigrees** used to track diseases?

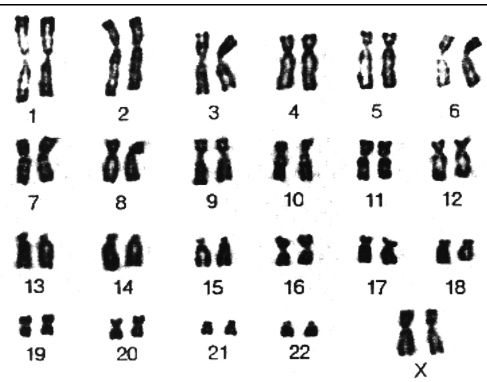
Pedigrees are used by genetic counselors to trace the history of a disease through families, determine the genotypes of people & therefore determine their risk of the disease (and their risk of passing on disorders). Pedigrees can be used to figure out the genotype and phenotype of an unknown ancestor, the genotypes of known, living family members & potential inheritance.



4.2.j. How can doctors and genetic counselors calculate the probability of a child inheriting a disease?



Amniocentesis



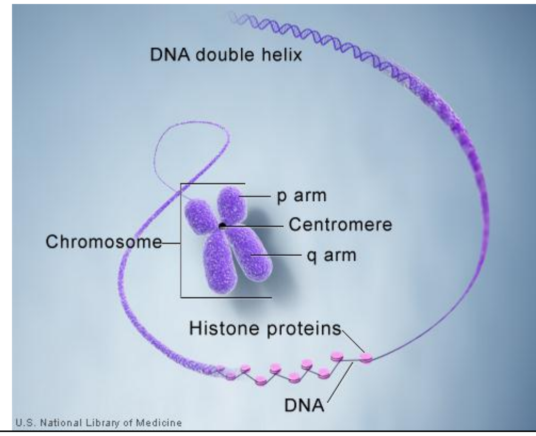
This is a karyotype for a female with a normal **chromosome** count.

Karyotypes show an individual's chromosomes. They are used to diagnose **chromosomal** abnormalities (i.e. Triple X, Turner Syndrome, Klinefelter's Syndrome, Down's Syndrome, Cry of the Cat & Superman Syndrome). They can be used on an adult who suspects he has a **chromosomal** abnormality, but that is unusual. Typically they are done on a fetus at about 20 weeks gestation. The obstetrician must perform amniocentesis to extract genetic material. A **karyotype** of the genetic material is then created. It shows the 22 pairs (hopefully) of **autosomal chromosomes** and the single pair (hopefully) of **sex chromosomes**. If there is an abnormal **chromosome** number, parents must decide whether to carry the child to term or whether to abort the fetus. There is no cure (at least yet) for **chromosomal** abnormalities.

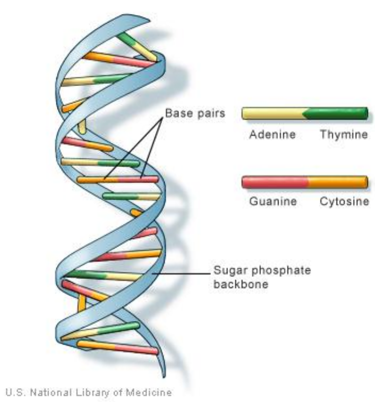
4.3 How Do Chromosomes Carry Information?
Study Guide by Hisrich

4.3.a. What are chromosomes made of? 4.3.c. What is the relationship between chromosomes, DNA, & **genes**?

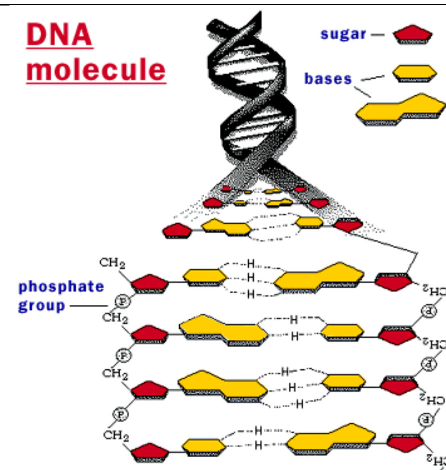
Chromosomes are tightly would packages of DNA that each contain multiple **genes** (from about 20 to more than 100 each). In order to package itself as tightly as possible, the DNA winds itself around histone proteins. However, DNA is a different kind of molecule than a protein. Proteins are made of 20 different kinds of amino acids, whereas DNA is made of the 4 **nucleotides adenine**, **guanine**, **cytosine**, and **thyamine**.



4.3.b. What is DNA?



DNA stands for deoxyribonucleic acid and is the 4th kind of macromolecule (in addition to proteins, carbohydrates & lipids. It is found in the nucleus of the cells of living organisms, from strawberries to grass to flies to humans. It has the structure of a double **helix**, with two complimentary strands held together by **hydrogen bonds**. **Adenine** always pairs with **thymine** & **cytosine** always pairs with **guanine**. The name comes from the fact that the sugar attached to each **nucleotide** is deoxyribose & the building blocks of DNA are nucleic acids. DNA was first isolated in 1869, but wasn't found to be the molecule of heredity until 1952. Since DNA is too small to see with a microscope, **models** can be used to help show the structure.

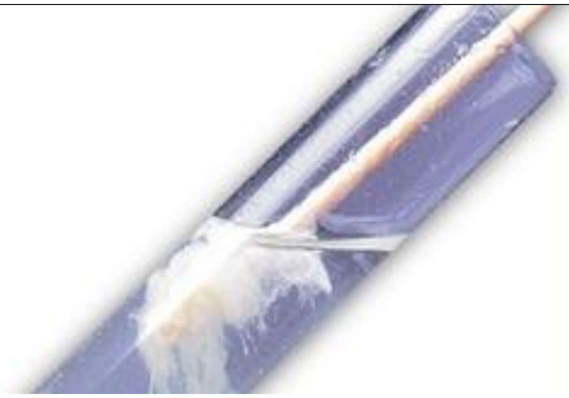


4.3.d. Does every cell in an organism have the same DNA?

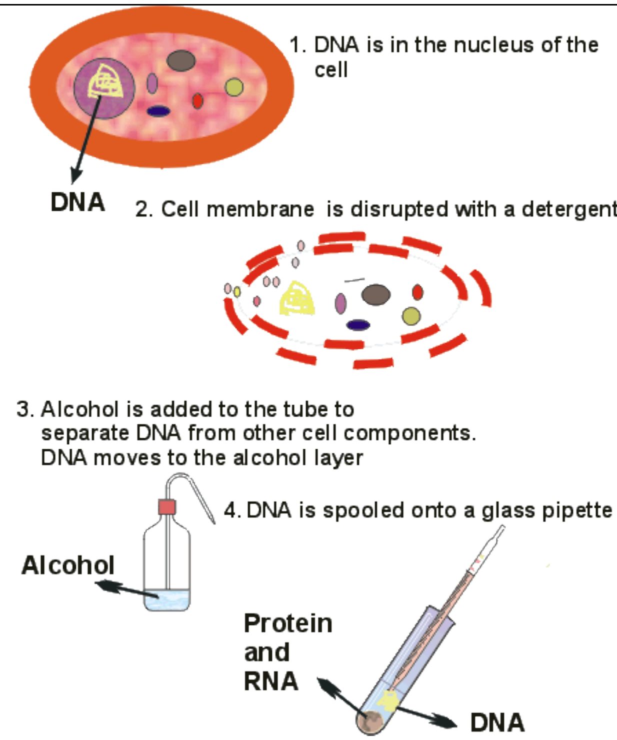
Old Answer	New Answer
Yes. Every cell with a nucleus, from those in the skin to those in muscles to those in an eyeball, contain all of the chromosomes of the individual and all the DNA. The reason the tissues are different is because different genes are "turned on" in different cells.	Maybe not. Here's a quote from a 2009 study, "AAA is one of the rare vascular diseases where tissue samples are removed as part of patient therapy. When they compared them, the researchers discovered major differences between BAK genes in blood cells and tissue cells coming from the same individuals, with the suspected disease "trigger" residing only in the tissue." **http://www.sciencedaily.com/releases/2009/07/090715131449.htm**

4.3.e. How do scientists isolate DNA in order to study it?

1. Break open the cells (**lysis**) to get the DNA out of the nucleus—this step requires the use of a **buffer** to maintain pH (DNA is pH sensitive)
2. Removing membrane lipids using detergent
3. Removing proteins by adding the enzyme protease (optional)
4. Precipitating out the DNA, using ice cold alcohol

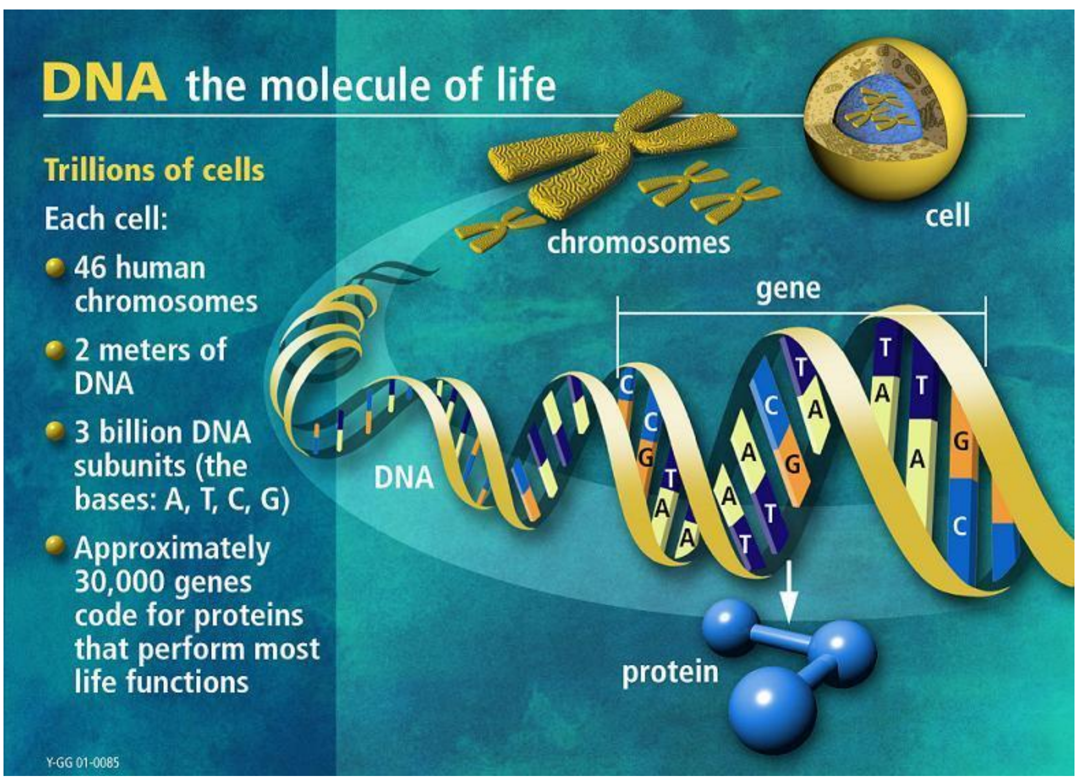


The DNA forms a **supernatant**—floating on the surface. It can be removed using an instrument like a toothpick.



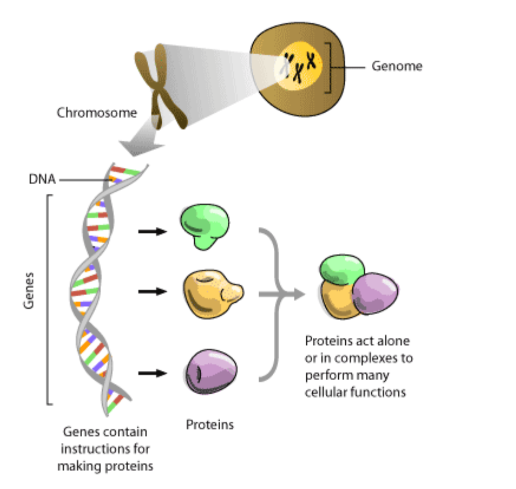
4.3.f. How much DNA is in a single human cell?

A human genome is 46 chromosomes, with a total of 3 billion base pairs. Each base pair is 0.00000000034 meters long. 3×10^9 base pairs * 3.4×10^{-10} meters/bp = 1 m of DNA. Other estimates are up to 3 meters.



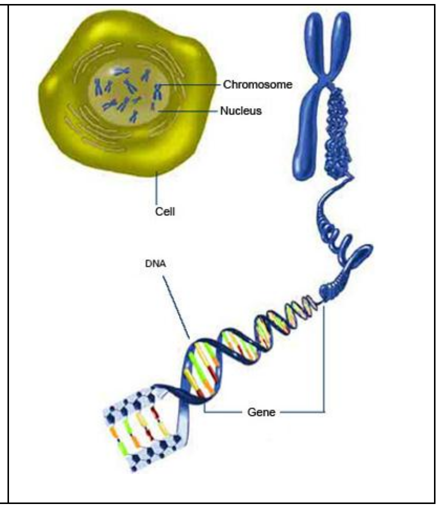
4.4 What is the DNA Code? Study Guide by Hisrich

4.4.a. What is a gene? 4.4.b. What is the DNA code? 4.4.c. What is the connection between genes and proteins?

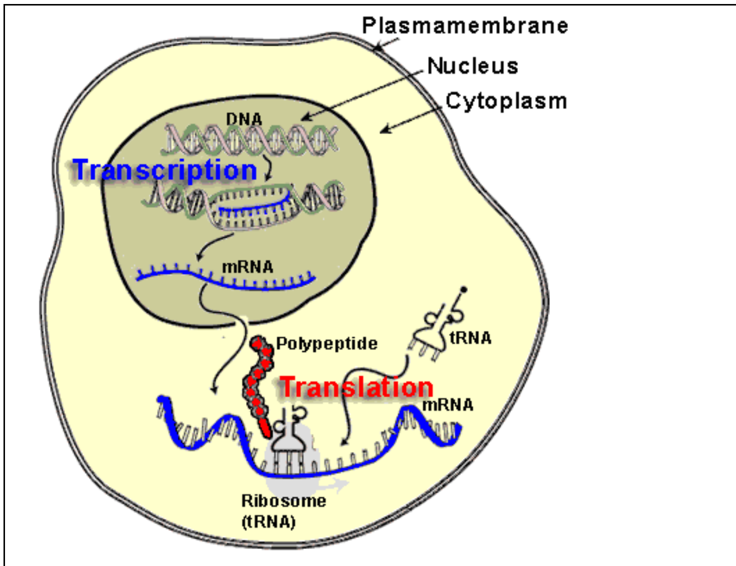


A gene is a segment (piece) of a chromosome & is made of DNA. Genes range in size from about 1,000 base pairs to over 1 million base pairs (pairs of **nucleotides**) in length. Each gene codes for the production of one protein & each protein determines one trait. Our genes determine our heredity (traits like hair color, height, and even personality).

Every 3 **nucleotides** is called a codon and codes for a different **amino acid**. A string of **amino acids** makes a protein.



4.4.d. How are proteins produced in a cell?



TRANSCRIPTION

First RNA polymerase transcribes the DNA in the nucleus of a cell. Transcription factors unwind the DNA and allow the **ribonucleic acid** (**RNA**) polymerase to transcribe one strand of the DNA into a single strand of **mRNA** (**messenger RNA**).

TRANSLATION

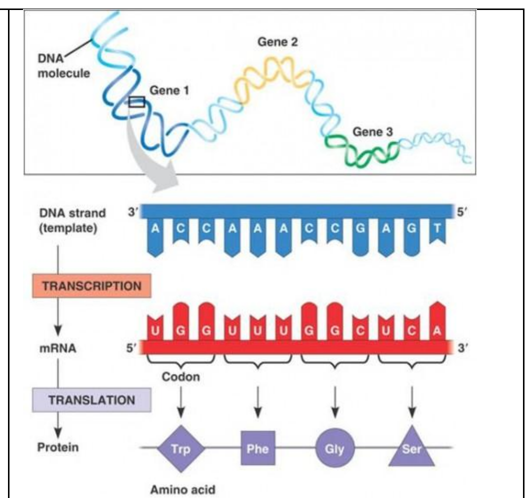
mRNA then travels through the nuclear membrane to go to the cytoplasm. The **ribosomes** in the cytoplasm translate the **mRNA**, using **tRNA** (**transfer RNA**). Each **tRNA** molecule brings one **amino acid** to the **mRNA** until a long string of **amino acids** forms, creating the **primary structure** of the protein.

4.4.e. How does the sequence of **nucleotides** in DNA determine the sequence of **amino acids** in a protein?

Second base

First base	U	C	A	G	Third base
U	UUU, UUC – Phenyl-alanine F UUA, UUG – Leucine L	UCU, UCC, UCA, UCG – Serine S	UAU, UAC – Tyrosine Y UAA Stop codon UAG Stop codon	UGU, UGC – Cysteine C UGA Stop codon UGG Tryptophan W	U C A G
C	CUU, CUC, CUA, CUG – Leucine L	CCU, CCC, CCA, CCG – Proline P	CAU, CAC – Histidine H CAA, CAG – Glutamine Q	CGU, CGC, CGA, CGG – Arginine R	U C A G
A	AUU, AUC, AUA – Isoleucine I AUG Methionine start codon M	ACU, ACC, ACA, ACG – Threonine T	AAU, AAC – Asparagine N AAA, AAG – Lysine K	AGU, AGC – Serine S AGA, AGG – Arginine R	U C A G
G	GUU, GUC, GUA, GUG – Valine V	GCU, GCC, GCA, GCG – Alanine A	GAU, GAC – Aspartic acid D GAA, GAG – Glutamic acid E	GGU, GGC, GGA, GGG – Glycine G	U C A G

The **nucleotides** in the DNA are transcribed into a complimentary (opposite) string of **mRNA**. The only difference is that RNA contains uracil in place of thyamine. During **translation**, **tRNA** attaches the correct **amino acid** for each **codon** (group of 3 **nucleotides**). Each chain of **amino acids** formed is a protein.



4.4.f. What determines the shape of a protein?

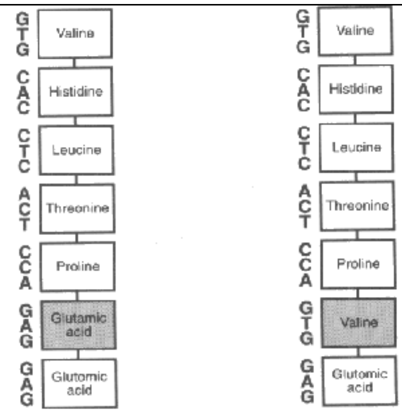
Protein shape is determined by the **primary structure** (sequence) of **amino acids**. Some **amino acids** are **hydrophilic** *("water loving")* & some are **hydrophobic** *("water hating")*. That leads to forces of attraction and repulsion that cause the protein to fold into complex shapes.

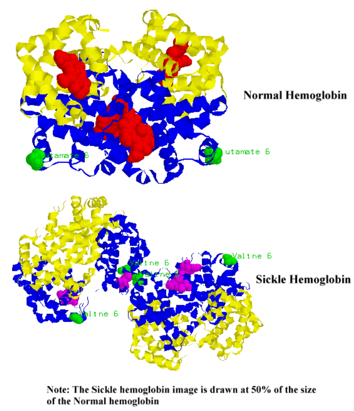
4.4.g. Is the shape of a protein affected by its surrounding environment?

Proteins fold differently depending on whether they are in an aqueous solution (mostly water), such as blood or whether they are in a lipid. When they are exposed to water, the fold so that the **hydrophilic** bits are on the outside and the **hydrophobic** bits on the inside, away from the water. When they are in a lipid, they do the exact opposite.

4.4.h. If the DNA code is changed, does the shape of a protein change? 4.4.i. Can changing just one nucleotide in a gene change the shape of a protein?

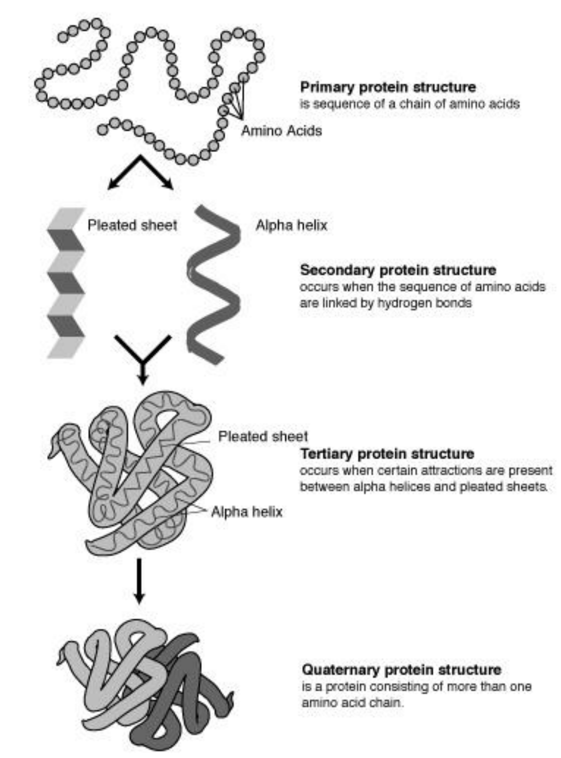
It depends. Most of the time, if the DNA code changes (called a **mutation**), the **amino acid** chain changes and that CAN cause a change in the shape of the protein. However, there are multiple codons that code for the SAME **amino acid**. For instance, if CCC becomes CCA, the **amino acid** coded for is still proline. Also, if a **hydrophobic amino acid** is replaced with another **hydrophobic amino acid**, they will both usually lead to the same folding. However, if a **hydrophilic amino acid** is replaced with a **hydrophobic** one, the protein shape will almost definitely change. That's the case with sickle cell hemoglobin.





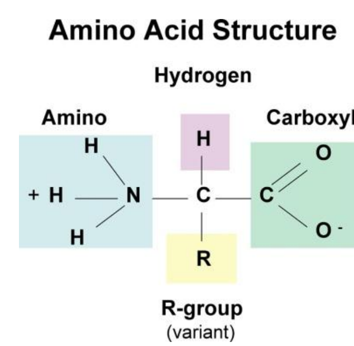
Only the **exon** portions of DNA code for proteins. The **introns** (also called "junk DNA") do not appear to code for proteins. Therefore if there is a mutation within an **intron**, the shape of the protein will not be altered.

4.4.j. Is it possible to design proteins that have specific characteristics? 4.4.k. How are proteins designed?



Yes. That's what Genetic Engineers do. They create sequences of DNA that will result in particular sequences of **amino acids** to form the protein they want. The **primary structure** is the proper sequence of amino acids. The **secondary structure** creates the protein's backbone. The **tertiary structure** involves the way the side chains interact due to **hydrophobic/hydrophilic** interactions, hydrogen bonds, disulfide bridges & ionic bonds. Only some proteins have **quaternary structure**, formed by the way the polypeptides in a complex protein interact.

More about **amino acid** structure: Every **amino acid** has at least 3 parts—1) The **amino group** (nitrogen bonded to 2 hydrogens), 2) the **carboxyl group** (carbon double bonded to oxygen and possibly to a **hydroxyl group**) and 3) the R-group (the part that is different in different **amino acids**. **Hydrophilic amino acids** have for their R-group a **hydroxyl group** (a hydrogen joined to an oxygen by a polar covalent bond), giving them a charge and making them water soluble. Because of the **hydroxyl group**, they are called alcohols. **Hydrophobic amino acids** lack the **hydroxyl group** & are non-polar (uncharged), making them insoluble in water.



4.5 Mistakes Happen Study Guide by Hisrich

4.5.a. Can a person with 45 or 48 chromosomes survive?

Humans normally have 46 chromosomes (23 from mother and 23 from father), though it's possible to survive with 45 IF the missing chromosome is the sex chromosome X and the person is a female (Turner Syndrome). A missing autosomal chromosome OR missing X sex chromosome is ALWAYS deadly.

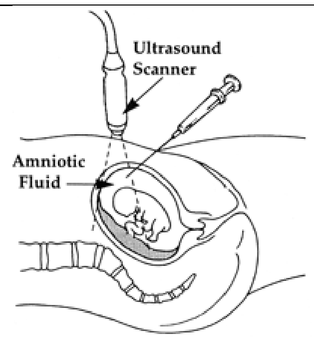
People can also survive with an extra chromosome (depending on the chromosome). It's typically only possible if the extra is a sex chromosome (the exception is Down's Syndrome).

Disorders with extra chromosomes (47)				Disorder with missing chromosome (45)
Trisomy 21 3 of chromosome 21 Down's Syndrome	XXX 3 sex chromosomes Triple X	XXY 3 sex chromosomes Klinefelter's Syndrome	XYY 3 sex chromosomes Superman Syndrome	XO 1 sex chromosome Turner Syndrome

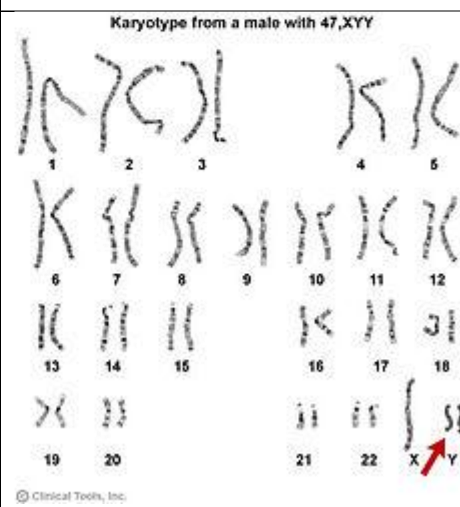
4.5.b. What happens if someone has more or fewer than 46 chromosomes?

Fewer	More
Dies before mother gives birth or within a year after (exception is Turner Syndrome, which results in retardation and infertility).	Dies before mother gives birth (exceptions shown in table above). If the person lives (s)he often experiences mental retardation and physical abnormalities (including infertility).

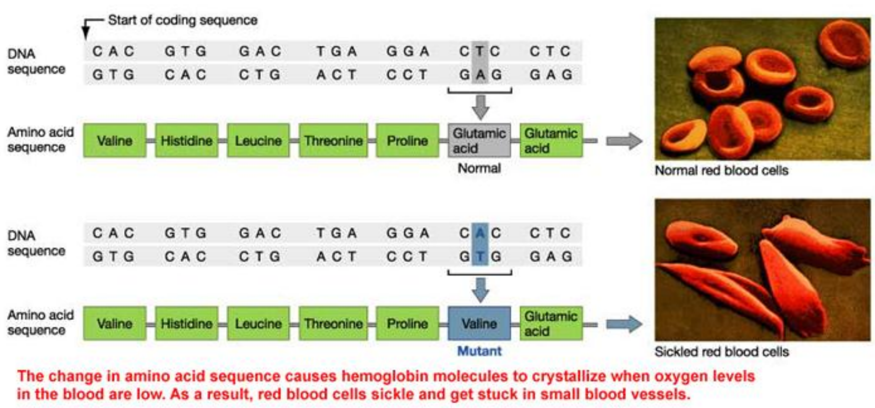
4.5.c. How can doctors detect if a patient has an abnormal number of chromosomes?



Karyotypes show an individual's chromosomes. They are used to diagnose chromosomal abnormalities (see table in 4.5.a.). They can be used on an adult who suspects he has a chromosomal abnormality, but that is unusual. Typically they are done on a fetus at about 20 weeks gestation. The obstetrician must perform amniocentesis to extract genetic material. A **karyotype** of the genetic material is then created. It shows the 22 pairs (hopefully) of autosomal chromosomes and the single pair (hopefully) of sex chromosomes. If there is an abnormal chromosome number, parents must decide whether to carry the child to term or whether to abort the fetus. There is no cure (at least yet) for chromosomal abnormalities.

Down's Syndrome	Turner Syndrome	Superman Syndrome
		

4.5.d. Can changing a single nucleotide in a gene cause a disease?

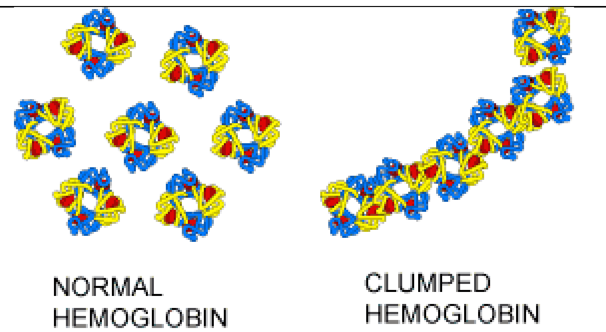


Yes (remember sickle cell?). These **mutations** are called single nucleotide polymorphisms ("many forms") are the most common type of **mutation** that occurs.

Since sickle cell disease is **recessive trait** (as are MOST **mutations**), an individual must receive a mutated allele from EACH parent to be affected. Otherwise, the normal **dominant trait** will be expressed (called sickle cell trait).

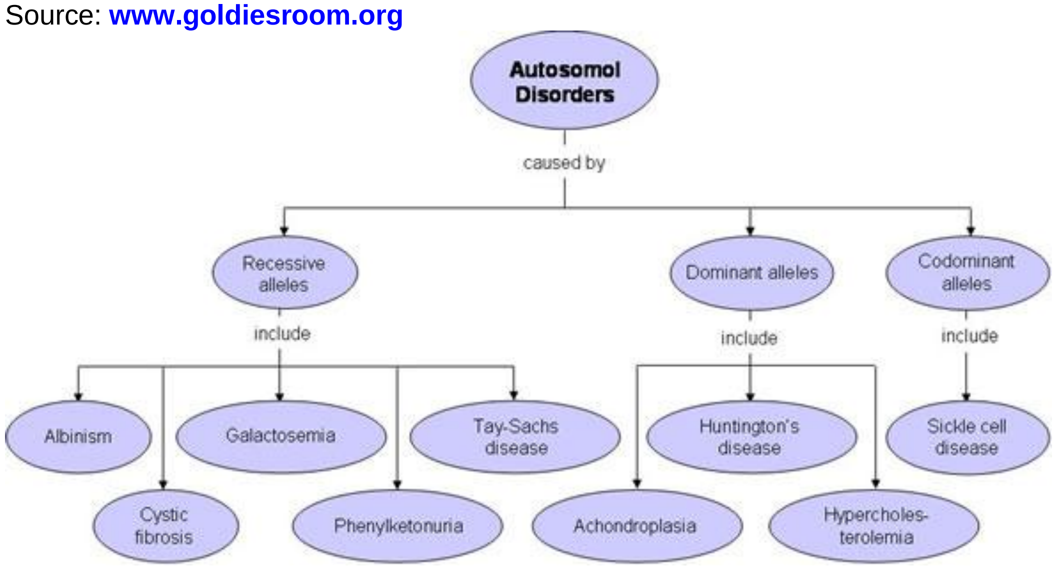
4.5.e. How is sickle cell hemoglobin different from normal hemoglobin?

Sickle cell hemoglobin simply has a single amino acid in the beta globin section that is different from that in normal hemoglobin. Instead of glutamic acid, a person with mutated hemoglobin has valine. Normal hemoglobin molecules don't attract each other, but mutated hemoglobin molecules do, causing them to clump (or **polymerize**) into long chains, pushing the blood cells into an elongated sickle shape. Valine is non-polar (and therefore **hydrophobic**) whereas glutamic acid is negatively charged, making it **hydrophilic**. The substitution of a **hydrophobic** amino acid for a **hydrophilic** one causes the protein to fold differently & behave differently.



4.5.f. What DNA mutations are directly linked to inherited diseases?

Source: **www.goldiesroom.org**



Autosomal mutations (shown in graphic left) are found in chromosome pairs 1-22 (not the sex chromosomes). The most common one are shown in the table left. Recessive diseases require a **mutated** allele to be inherited from each parent, whereas dominant disorders like dwarfism (achondroplasia) require only one. Sickle cell disease is sometimes considered co-dominant because in low oxygen environments, the cells of a person with sickle cell TRAIT may become sickle, though under normal conditions they do not.

Sex-linked **mutations** (carried on X-chromosome) include color-blindness, hemophilia, Duchenne muscular dystrophy, vitamin D resistant rickets, fragile X syndrome, Congenital aqueductal stenosis (hydrocephalus) & more.

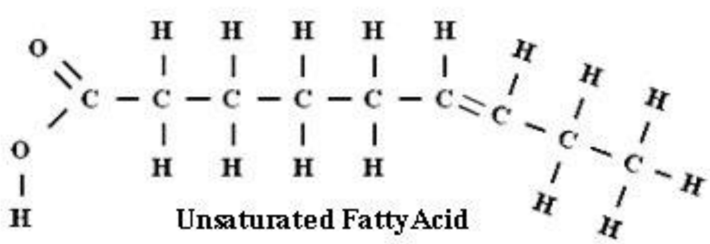
5.1 Cholesterol Study Guide by Hisrich

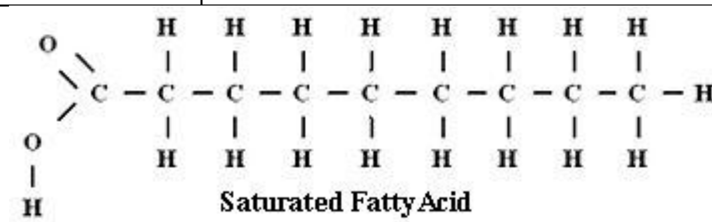
5.1.a. Are all fats the same?

All fats have hydrophilic heads and one or more hydrophobic tails (**fatty acid** chains). They are primarily carbon chains, coated in hydrogen atoms, and have some oxygen atoms in the head. However, there are different kinds of fats.

5.1.b. What is the difference between **saturated** and **unsaturated fats**?

Unsaturated Fats (or **Fatty Acids**)	Both	**Saturated Fats** (or **Fatty Acids**)
Healthier From fish, plant oils, seeds and nuts Usually liquids 1+ double bonds between the C atoms (less H atoms attached) Necessary for health Can be **monounsaturated** (one double bond) or **polyunsaturated** (multiple double bonds)	Not polymers *("many parts")* (no repeating building block) **Triglycerides** are the most common form consumed by humans Intake should be limited	Unhealthy Mostly from animal sources Usually solids All single bonds between carbon atoms (maximum # of H atoms attached) Not necessary for health (ideal amount = zero)


Unsaturated Fatty Acid


Saturated Fatty Acid

5.1.c. Why **unsaturated fats** are considered healthier than **saturated** ones?

Unsaturated Fats (or **Fatty Acids**)	**Saturated Fats** (or **Fatty Acids**)
Fewer calories	More calories
Lower **LDL** ("bad cholesterol") levels	Raise **LDL** ("bad cholesterol") levels
Lessen risk of heart disease	Raise risk of heart disease
Liquids, don't link up and clog arteries	Solids that stick to each other in the bloodstream, creates plaque that clogs arteries
Omega-3 fatty acids are a type of **polyunsaturated fat**. They are essential **fatty acids** and the body cannot make them.	Trans fats are a man-made **saturated fat** and are the worst kind for the heart's health
	

5.1.d. What is **cholesterol**?

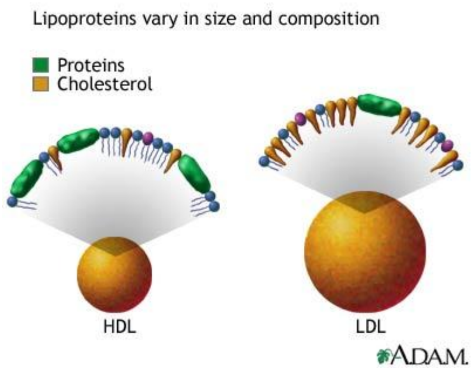
Cholesterol is a lipid made in the liver of animals. It helps form cell membranes & is found in all tissues, but especially nervous and fat tissue. It protects the skin and helps nerve cells function. It also helps detoxify the blood.

5.1.e. Why are so many foods advertised as non-fat and **cholesterol**-free?

Humans do not need to consume **cholesterol** to be healthy. The human liver makes it. Most humans take in too much **cholesterol** from their food, putting the health of their hearts at risk.

5.1.f. What are **LDL** and **HDL**?

LDL	Both	**HDL**
Low Density Lipoprotein Carry **cholesterol** through blood to all tissues—if there's too much it just stays in the blood Raises risk of heart disease Leads to blood vessel blockages—white blood cells try to digest LDL & convert it to a toxic form. White blood cells create inflammation & that draws more cells & plaque	Part lipid, part protein Carry **cholesterol**	**High Density Lipoprotein** Pick up **cholesterol** in the bloodstream and take it to the liver for removal from the body Lowers risk of heart disease Reduces blood vessel blockages



5.1.g. Why are there so many drugs available to lower **cholesterol** or **LDL**?

5.1.h. How are **LDL**, **HDL**, and **cholesterol** related to heart disease?

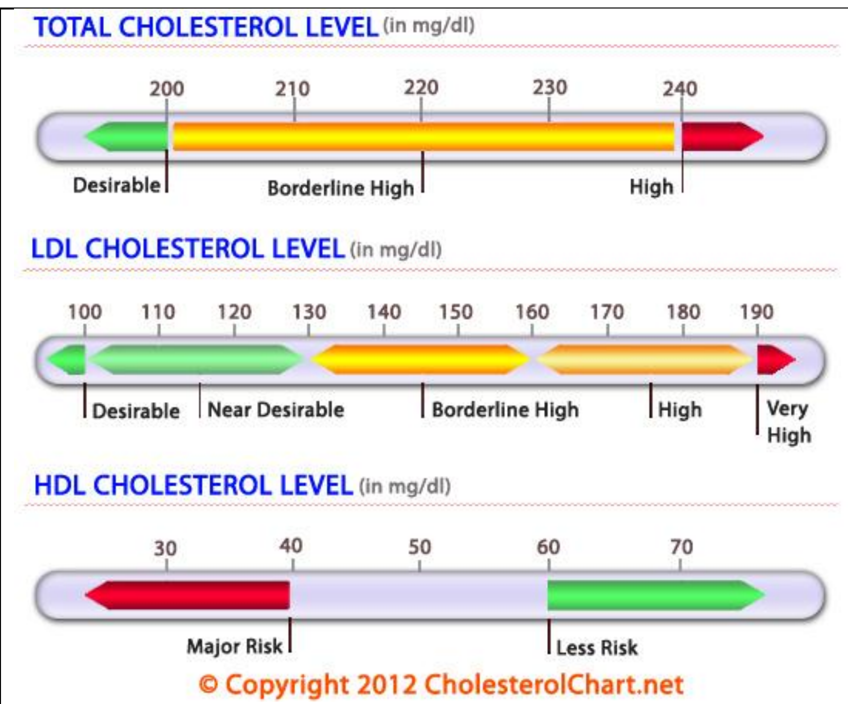
Heart disease is the #1 killer of Americans, killing over ½ million Americans per year.

HDL, **LDL** & total **cholesterol** levels are highly correlated with risk of heart disease and heart attack. Keeping levels healthy is a great way to protect the cardiovascular system.

Here are facts from the CDC*:

- 71 million American adults (33.5%) have high LDL, or "bad," cholesterol
- People with high total cholesterol have approximately **twice the risk** of heart disease as people with optimal levels

*http://www.cdc.gov/cholesterol/facts.htm



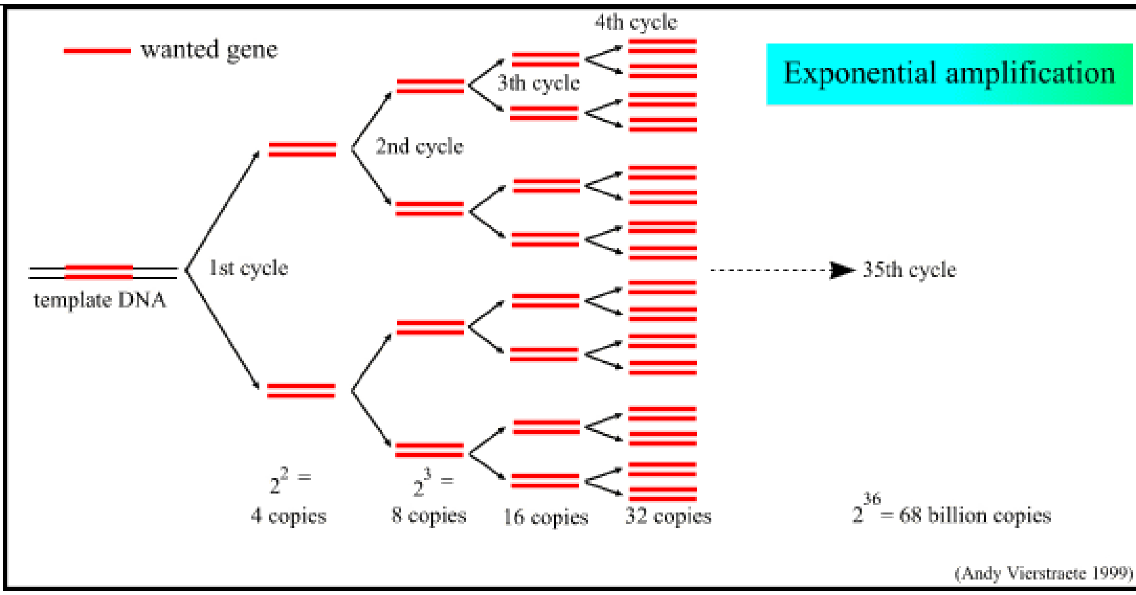
5.2. Molecular Biological Techniques Study Guide

5.2.a. How do crime scene investigators get enough DNA evidence from a single drop of blood?

The put the DNA through an **amplification** process called “**PCR**.”

5.2.b. What is **PCR**?

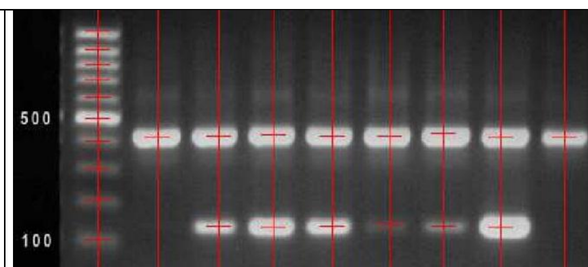
PCR stands for **polymerase chain reaction** and is a way to take a small amount of DNA and **amplify** it to create a much larger sample size (that can then be analyzed). **PCR** amplifies the DNA **exponentially**.



5.2.c. How is DNA analyzed without sequencing it?

DNA can be analyzed through gel **electrophoresis**. **Electrophoresis** allows the comparison of an unknown piece of DNA to a known gene. The more the pieces of DNA match up, the more similar the DNA sequences. DNA from a person with a disease (like **familial hypercholesterolemia**) can be compared to someone who wants to know whether (s)he has that disorder.

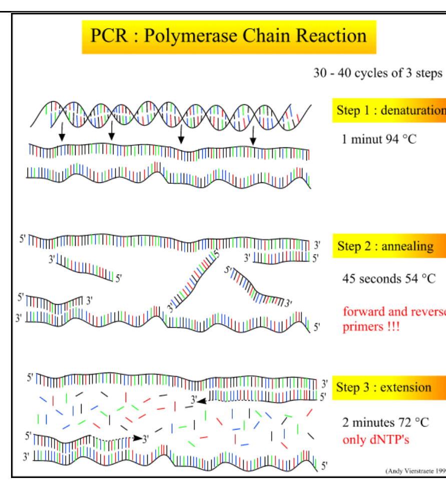
Ways in which our genes vary are called **polymorphisms** (“many forms”) and are the ways in which one person’s DNA is compared to another’s. Electrophoresis shows whether 2 people have the SAME form of a particular gene or a DIFFERENT form.



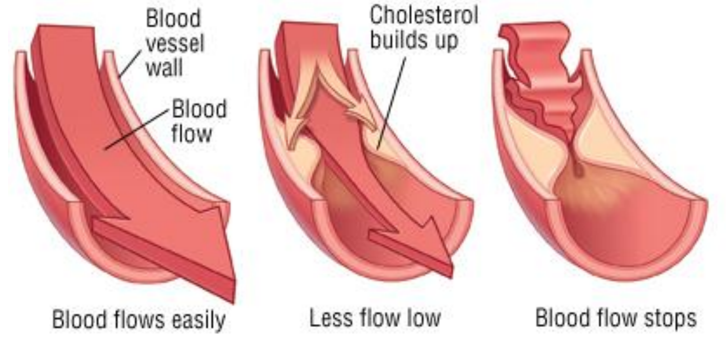
5.2.d. What does **PCR** do and how does it work?

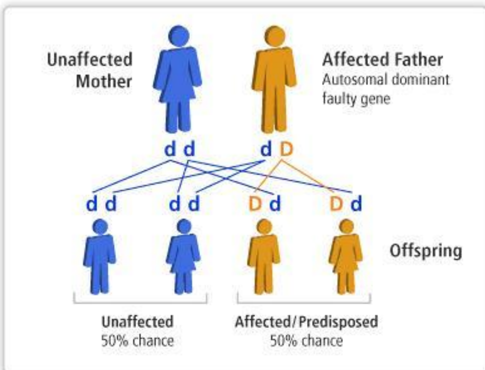
PCR is a way to amplify DNA to make a much larger sample. Here are the steps:

1) Denature the DNA to separate the strands
2) Left and right primers pair to complementary sequences
3) Taq **polymerase** (an enzyme that catalyzes formation of DNA) is attaches to the priming sites and extend (synthesize) a new DNA strand
4) Steps repeat until there are billions of copies



5.2.e. Can genetic diseases or disorders be diagnosed using a small blood or saliva sample from a patient?





Yes. The DNA can be **amplified** and then run through **electrophoresis**.

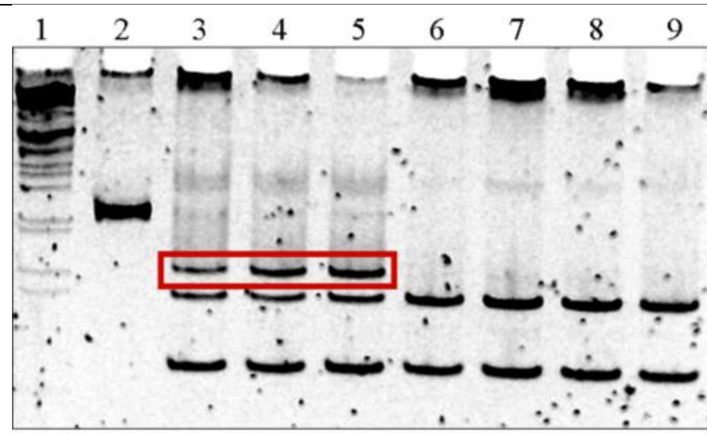
Familial Hypercholesterolemia *("high cholesterol in the blood*") is a dominant autosomal genetic disorder is the result of a **mutation** in DNA that is passed from parents to their offspring. The disease typically occurs when a person inherits a dominant **allele** from one parent, giving him a heterozygous *("full of different things joined together")* genotype (Hh). On very RARE occasions, the person has TWO affected parents and inherits the mutation from BOTH of them, giving him a **homozygous** *("full of same things joined together")* dominant genotype (HH). Either will result in **familial hypercholesterolemia**, but a homozygous dominant genotype makes the condition far worse.

The **phenotype** *("showing type")* of a person with familial hypercholesterolemia is that LDL cholesterol (generally called "bad cholesterol" builds up in the bloodstream, leading to very high cholesterol levels in the blood and putting the person at high risk for a heart attack.

5.2.f. Why are DNA tests on television programs and movies shown as patterns of stripes or bands on film or in gels? 5.2.g. What is gel **electrophoresis** and how are the results interpreted?

The bands shown on TV are gel **electrophoresis** results. Gel **electrophoresis** is used to compare unknown DNA to known DNA. The steps are:

1) **Amplify** the DNA sample
2) Use **restriction endonuclease** to cut the DNA into pieces
3) Make an **agarose** gel (source is seaweed) that the DNA can travel across in a **linear** (straight) line
4) Load the DNA samples into the wells in the **agarose** gel and put the wells in the negative end of the **electrophoresis** apparatus
5) Turn on the **electrophoresis** apparatus and let it run about 30 minutes—DNA will travel toward the + electrode because of its – charge
6) Stain the **agarose** gel to get the DNA to appear
7) Compare the lanes of DNA



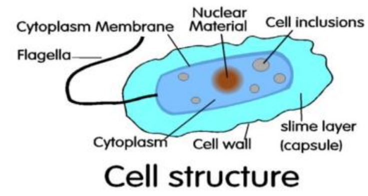
Gene screening diagnostic test for premature menopause

To interpret results, geneticists look at which RFLPs (lines) match between lanes. Where lines match, the DNA strand is the same length and that means the DNA is the same. Where lines DO NOT match up, there's a difference in the DNA strand. For example, in the gel above the RFLPs in lanes 3-5 match, showing similar DNA and the RFLPs in lanes 6-9 match, showing similar DNA. The most disimilar DNA is in lane 2 & appears to be unrelated to the rest. Lane 1 is the KNOWN reference DNA, used to determine the number of base pairs in each RFLP.

6.1 Bacteria Study Guide by Hisrich

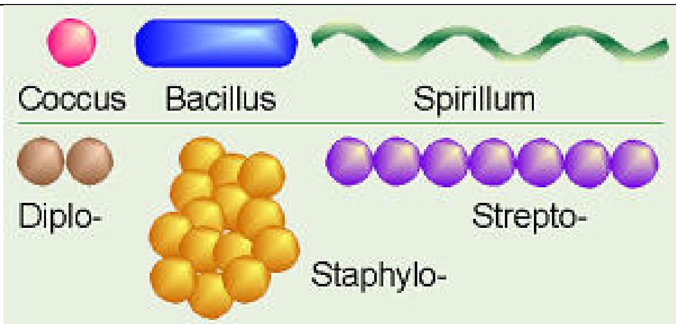
6.1.a. What are bacteria?

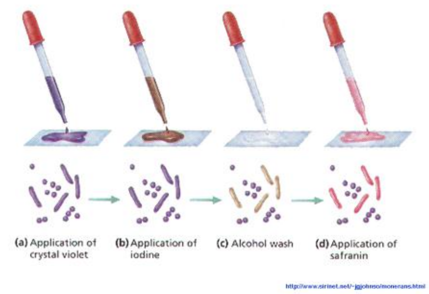
Bacteria are single-celled living organisms that are found all around us (and in us). They don't have organelles or a nucleus, the way animal cells do.

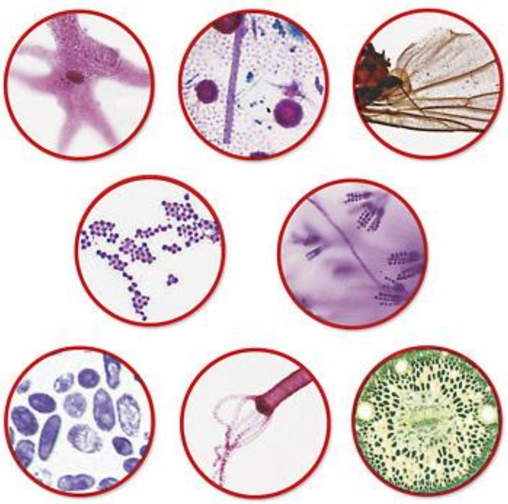


6.1.b. How do bacteria differ from one another? 6.1.f. How do scientists and doctors tell one bacteria from another?

Bacteria can be sorted into categories based on morphology ("study of shape") or **gram staining** results (red or dark blue/purple). Bacteria come in three different shapes—**bacillus** ("rods"), spirilla ("spirals") & **cocci** ("round or berry-shaped") and either exist as single cells, in pairs (diplo-), in clumps (staphylo-) or in strings (strepto-).







These are some examples of what various bacteria look like under a microscope.

<u>Scientists look for 3 main things to tell bacteria apart</u>:

1) Color—red or blue/purple

(determined by Gram staining)

2) Shape—round, rod-shaped or spiral

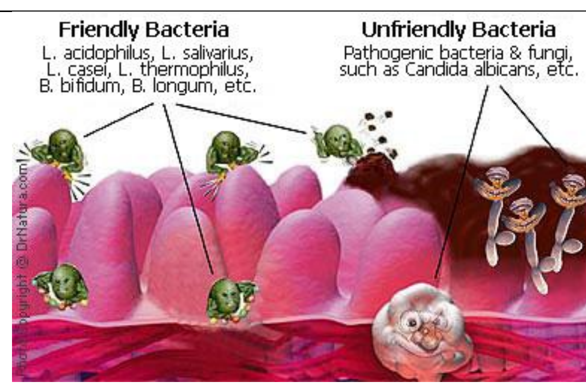
3) Colony type—single, paired, clumped or strings

6.1.c. Do all bacteria cause disease?

About 99% of bacteria fall into the categories of benign (non-harmful) or even beneficial. Less than 1% of all bacteria varieties are pathogenic ("disease-causing").

An example of beneficial bacteria are lactobacillus (found in dairy products—help with digestion). In fact, bacteria are so important to digestion that there are about 100 trillion of them in our guts (more bacteria cells than there are human cells in our WHOLE body!)

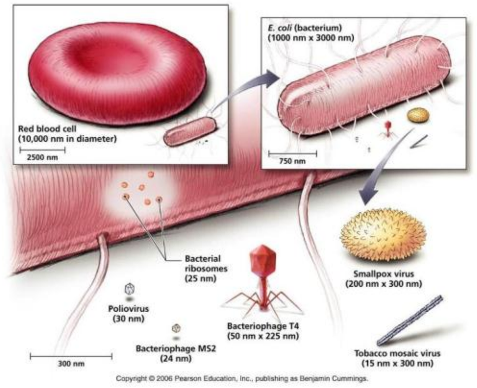
We would DIE without bacteria & in fact can become ill if they are killed off (for example, by antibiotics).



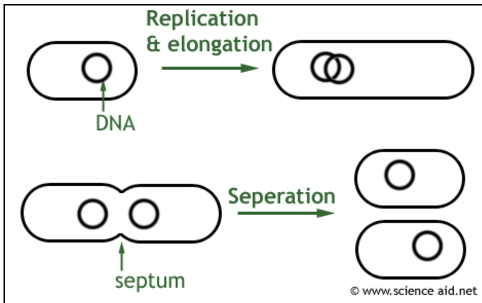
6.1.d. What is the size of bacteria compared to the size of human cell?

Bacteria cells are much smaller than human cells. There are about 70 trillion HUMAN cells in our bodies, but even MORE BACTERIAL cells (more than 100 trillion), so they are quite small.

The diagram shown to the right shows that a typical bacillus bacteria is about ¼ the size of a red blood cell. And viruses are much smaller still.



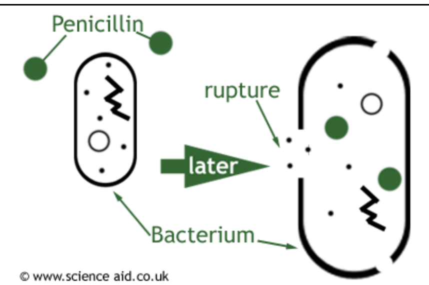
6.1.e. If bacteria are living cells, how do they reproduce?



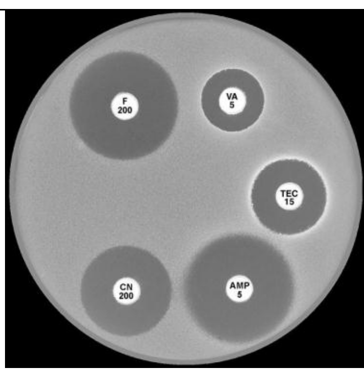
Bacteria typically reproduce asexually *("without sex")*, where one cell doubles its DNA and then splits into 2. This process is also called binary fission *("one breaking" into 2).* The drawback of binary fission is that it doesn't create any genetic diversity at all (the way sex does). Therefore, in order to create genetic diversity, one of 3 things can happen: 1) Conjugation: one bacterium gives another bacteria some of its genetic material by squirting it through a protein tube 2) Transformation: a bacterium takes up genetic material from the environment (often from dead cells) 3) Transduction: viruses called bacteriophages *("bacteria eaters")* insert their own genomes into the bacterium & the bacteria then replicate the viruses.

6.1.g. How are bacterial infections treated? 6.1.h. Can the same treatment be used for all bacterial infections?

Bacterial infections are treated with **antibiotics** *("against life")* unless they are **antibiotic-resistant** or there are no antibiotics available to treat them. **Antibiotics** are chemicals that attack/kill particular bacteria and are usually derived from other bacteria (who use them as a defense) or from organisms like mold (that's where penicillin was discovered. Some **antibiotics** are considered "broad spectrum" and can be used against many different bacteria (i.e. penicillin). Others are "narrow spectrum" and are effective against only a couple bacteria. An example is Azithromycin, used to treat gonorrhea.

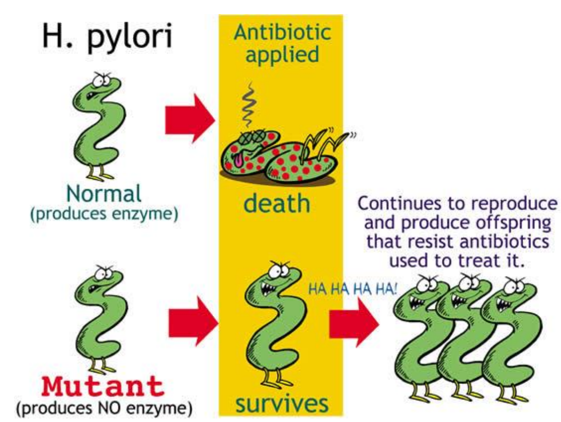


6.1.i. What is ***antibiotic resistance***, and why is it a major health problem today?



Antibiotic effectiveness can be measured by growing bacteria in a petri dish and then applying **antibiotic** disks and measuring the **zone of inhibition** (area bacteria cannot grow). The larger the **zone of inhibition**, the more effective the **antibiotic** against the bacteria.

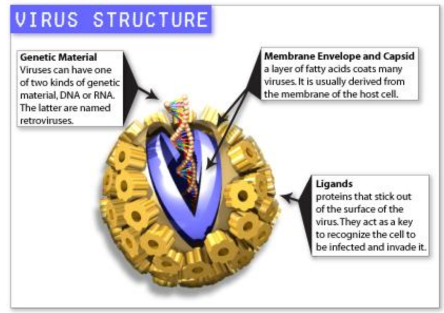
Antibiotic resistance is when bacteria develop immunity to the **antibiotic** that would usually attack them. It is due to the overuse and misuse of **antibiotics**. A few major causes exist: 1) Prescription of unnecessary **antibiotics** (say for a viral infection) 2) Failure to finish a dose of **antibiotics** 3) The use of **antibiotics** in livestock (the majority of **antibiotics** used in the U.S. go to farm animals). Now some diseases have developed resistance (ex: tuberculosis) and can no longer be treated with antibiotics.



6.2 Viruses Study Guide by Hisrich

6.2. a. What is a **virus**?

A **virus** is an EXTREMELY small structure that causes disease. Sometimes they are considered microorganisms, but technically they are NOT alive, since they cannot reproduce outside of living cells. They are very simple, being made up only of genetic material (either DNA or RNA) and a protein coating called a **capsid**, which is surrounded by a lipid **envelope**. A single virus particle is called a **virion**.

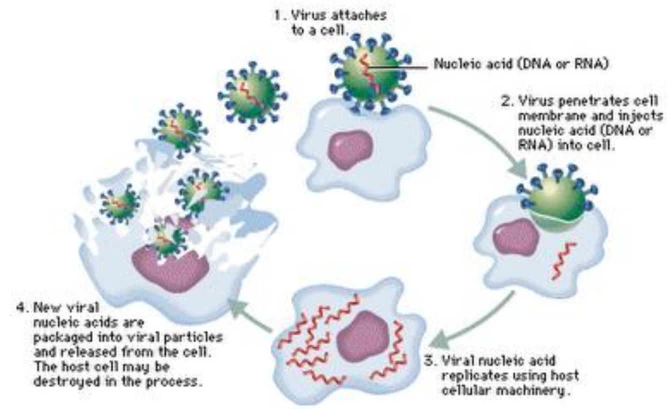


6.2. b. How are **viruses** different from bacteria?

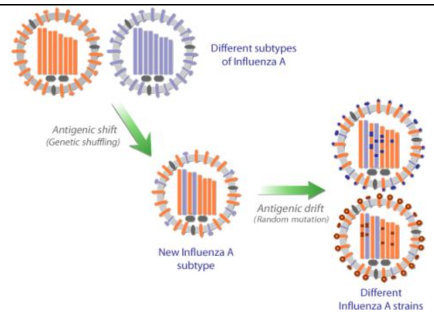
Bacteria	Both	Viruses
Definitely alive	XXX	Often considered non-living
Bigger than viruses	Smaller than human cells	Smaller than bacteria
Can be prepped on slides and viewed by light microscope	Invisible to the naked eye Can be seen using technology	Viewable only with an **electron microscope**
Gram staining used to distinguish between gram + and gram -	XXX	Gram staining not used for viruses
Shaped like rods, spheres or spirals	Can be rod-shaped	Shaped like rods, polyhedrons or more complex geometric shapes
Usually reproduce asexually	No sexual reproduction	Can only replicate in living **host cells**
Many form colonies	XXX	Do not form colonies
Some harmful, but 99% are beneficial (many even NEEDED for life)	Can be harmful	All harmful
Contain DNA	Contain genetic material, but no nucleus Genetic material evolves over time, causing diseases to change	Can have DNA or RNA
No drugs to prevent infection	Infectious (can be spread from person to person via body fluids, etc)	*Prevented* with vaccines (many)
Treated with antibiotics (many)		Few drugs to treat (rest and fluids only, in SOME cases, **antivirals** can be used)
Can become "resistant" to antibiotics	Humans can fight them Hand washing is effective prevention, acts by breaking down outside of organism through **lysis**	Don't become "resistant" to vaccines
Becoming harder to fight (due to antibiotic resistance)		Can be eradicated (aka smallpox); therefore easier to fight in a way
Vulnerable to viruses (bacteriophages attack them)	XXX	Not vulnerable to bacteria
Being infected does NOT protect you from future infections		Being infected can protect you from future infections

6.2. c. How do **viruses** reproduce and cause disease?

To reproduce, **viruses** must be inside a **host cell**. Because viruses are technically non-living, their form of reproduction is often called "replication." An analogy is that what's on paper cannot be copied without a copy machine. In this analogy, the paper represents the **virus's** genetic material and the copy machine the **host cell**.

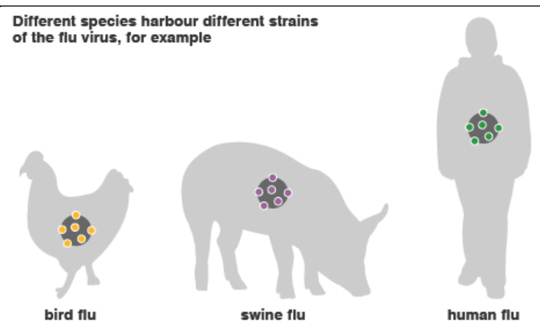


6.2. d. Why is every cold we get a new disease?

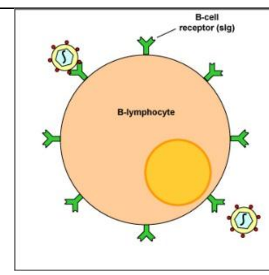


Every cold (and every **VIRUS**, for that matter) that we get is a new disease because once we've gotten a virus we develop antibodies to it and are no longer vulnerable to the exact **virus** again. However, over time **viruses** mutate and the genetic material changes. Once it has changed enough that our antibodies don't recognize it, we are vulnerable once more and can get the **virus**. That's why a flu vaccine has to be carefully matched to the strains likely to infect humans or it won't effectively prevent the flu.

6.2. e. Why do humans, cats, dogs, and birds all get the different viral infections?

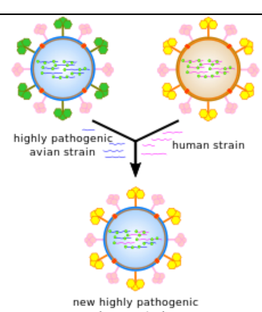


Different species get different viral infections because each virus is specially adapted to attack a certain species' cells and can't effectively attack the cells of other species because of the differences in structure. For instance, bacteriophages are specially adapted to replicate within bacteria cells. Small pox could only replicate within humans, but the closely related cow pox could infect cows and also humans.



The **virus** has to fit into the cell's receptors in order to replicate within the cell.

6.2. f. Why do epidemics of diseases caused by newly discovered viruses still occur?



Again, over time **viruses** mutate and the genetic material changes. That can make us vulnerable to the changed **virus**. Also, **viruses** can sometimes adapt to jump from one species to another (aka swine flu). Sometimes a strain that affects one species and a strain that affects another will combine material to form a new, more deadly strain (see picture to left).