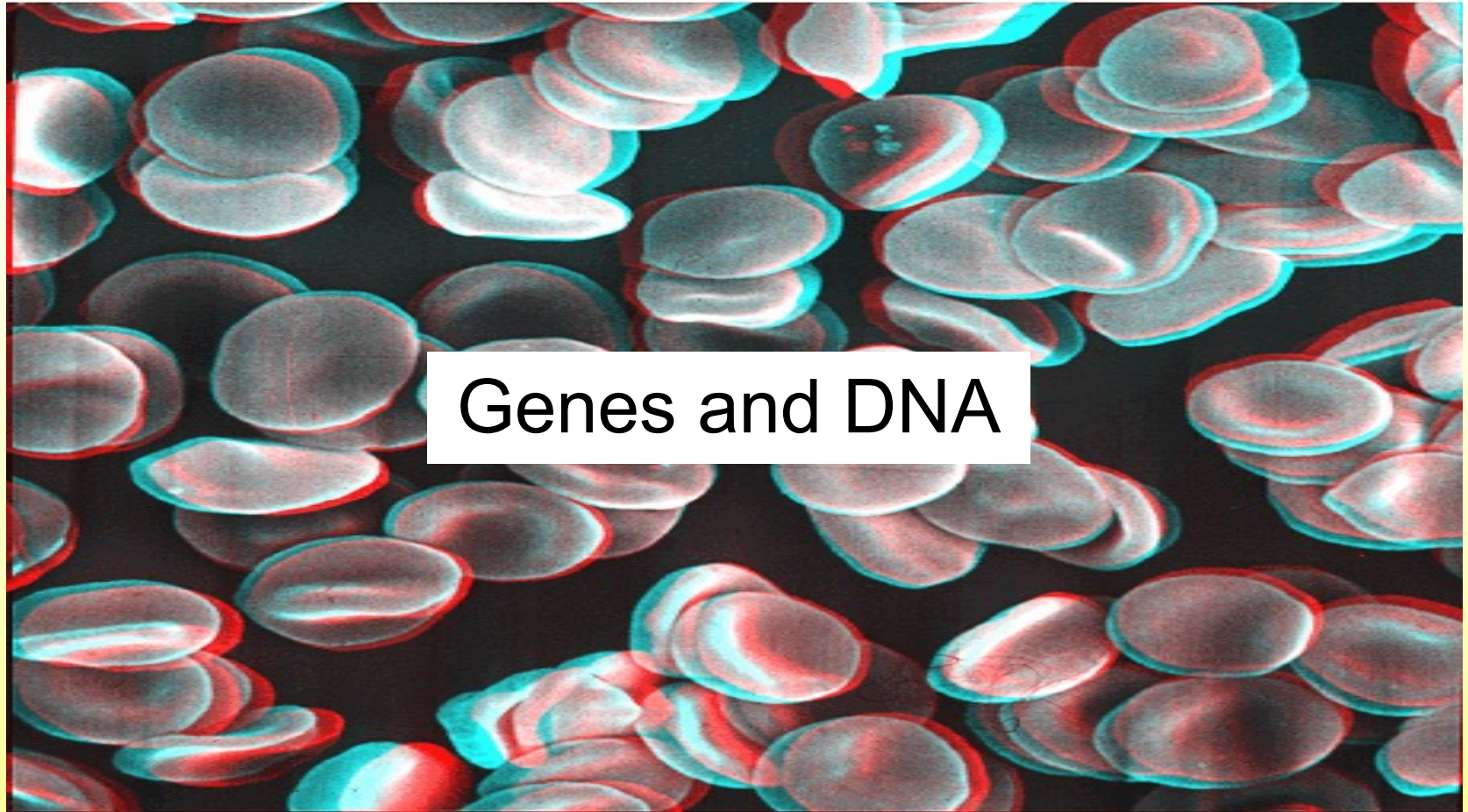


The Pieces Inside of You that Make You Who You Are



Genes and DNA

Who are you?

This question can be answered many ways...

- Personality traits

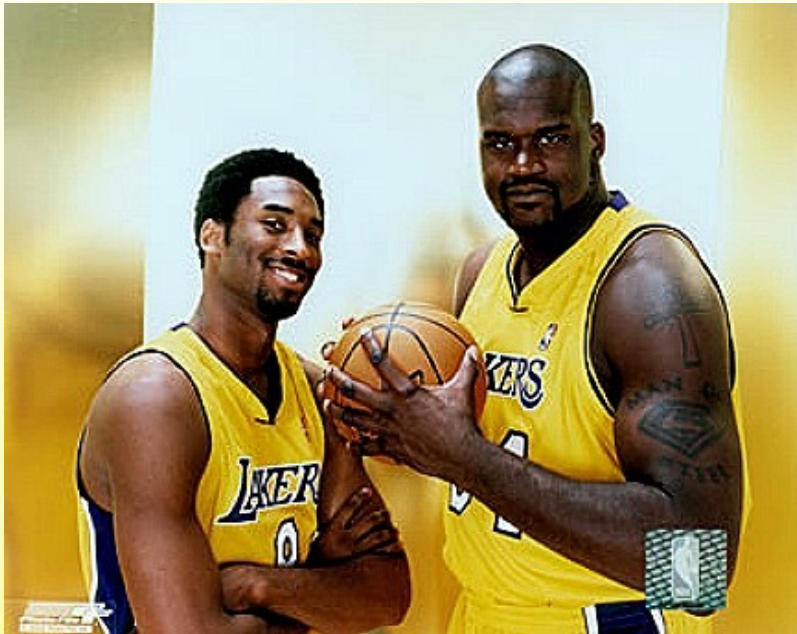
- Kindness
- Vegetarian or Carnivore
- Athlete

***Traits you can control



Who are you?

This question can be answered many ways...



Kobe Bryant and Shaquille O'Neal

- Physical Traits
 - Height
 - Eye color
 - Hair color and texture
 - Foot size
 - Bone strength
- Others???
- Age
- Sex

***Traits you can't control

What determines your physical characteristics?

- GENES

- Genes are molecules in every cell that act like recipes.
- They tell your cells how to behave by coding for proteins.
- Genes determine how we look, move and grow .
- You get your genes from your parents.



What are your genes?

- DNA (Deoxyribonucleic Acid)
 - DNA holds your specific code for every part of your body. It is the collection of recipe books.
 - A gene is made of a long strand of DNA.
 - There are about 30,000 genes in your DNA.



DNA

How Does DNA work?

Like different kinds of desserts, one recipe (gene) makes a cake while the other makes pudding. Both recipes make desserts, but the desserts are different. Some genes are more closely related than others like chocolate cake and white cake compared with a jolly rancher.

Ingredients:
Sugar, Flavoring



Ingredients:
Sugar, Flour, Eggs,
Baking soda, Vanilla

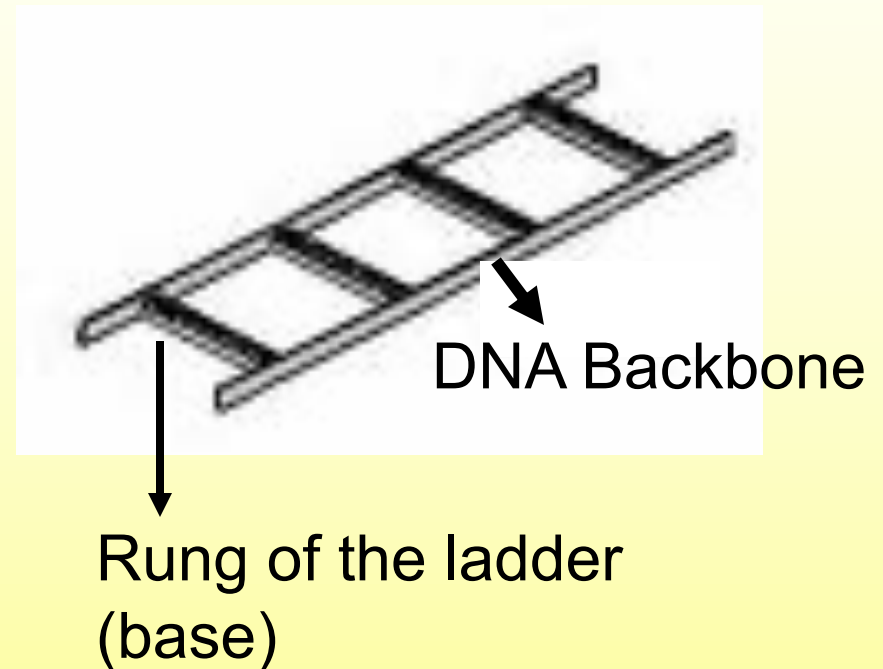


Ingredients:
Sugar, Flour, Eggs,
Baking soda, Chocolate



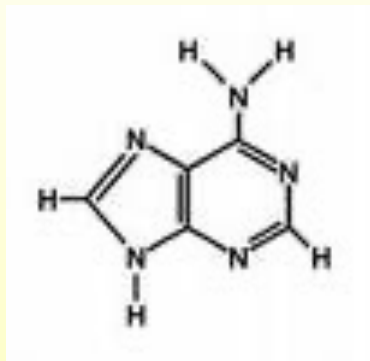
What is DNA made of?

- Nucleotides
 - A nucleotide is composed of two parts:
 - DNA Backbone
 - made of phosphorous, oxygen, carbon, and hydrogen
 - Rungs of the Ladder: Bases
 - Made of oxygen, carbon, hydrogen, and nitrogen

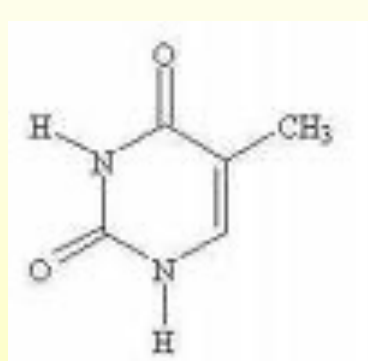


Bases are Important!

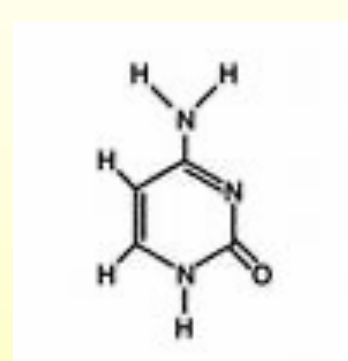
- There are four bases:



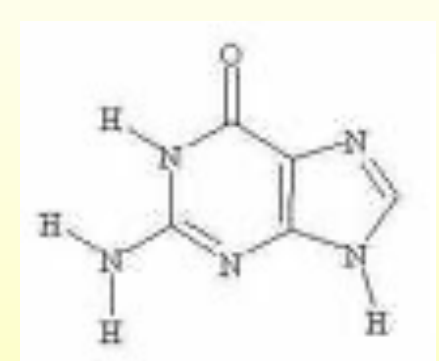
Adenine
A



Thymine
T



Cytosine
C

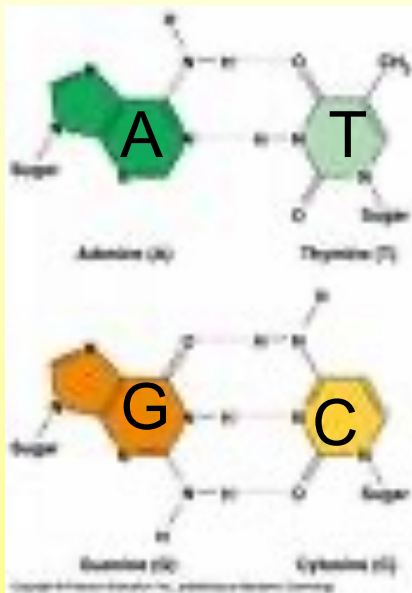


Guanine
G

- The order of these bases along a strand of DNA codes for your genes.

DNA is Fashionable

- Just like a model wearing a suit jacket with suit pants, or a t-shirt with blue genes,
A only pairs with T and
G only pairs with C.

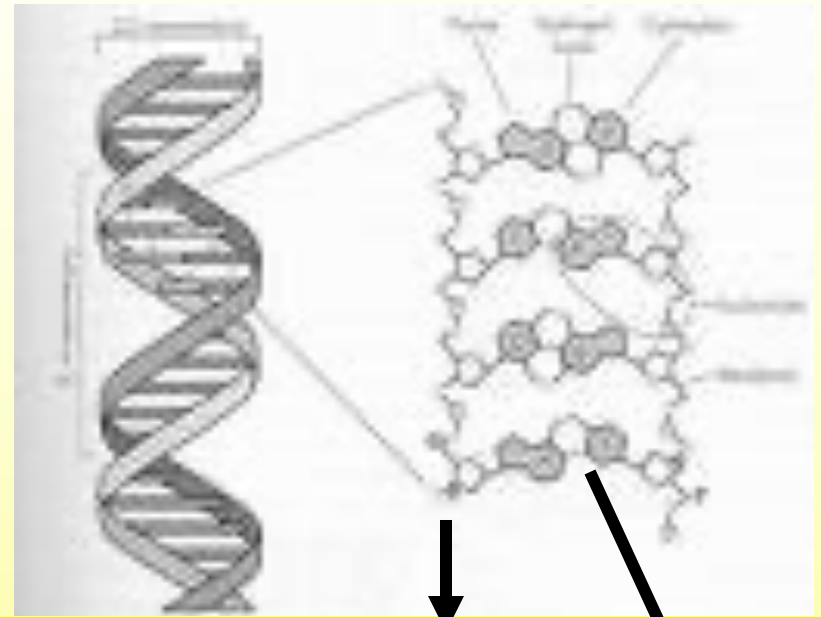


Remembering Key:
“AT”
G and C look alike



DNA Structure

In these two rows, the bases interact to make a twisted ladder shape, called a double helix (the bases make up the rungs on the ladder).



DNA
double
helix

DNA
backbone

Bases

What do Nucleotides do?

- Three nucleotides in a row code for one amino acid.
- A long string of amino acids makes a protein.

A strand of: CTG ACT CCT GAG GAG AAC TCT

Codes for: Leu Thr Pro Glu Glu Lys Ser

Which is the beginning of the protein _____

Nucleotide → Amino Acid → Protein



Proteins allow the body to perform and function

How do you know which nucleotides code for which amino acids?

1st position (5' end) ↓	2nd position				3rd position (3' end) ↓
	T	C	A	G	
T	Phe Phe Leu Leu	Ser Ser Ser Ser	Tyr Tyr STOP STOP	Cys Cys STOP Trp	T C A G
C	Leu Leu Leu Leu	Pro Pro Pro Pro	His His Gln Gln	Arg Arg Arg Arg	T C A G
A	Ile Ile Ile Met	Thr Thr Thr Thr	Asn Asn Lys Lys	Ser Ser Arg Arg	T C A G
G	Val Val Val Val	Ala Ala Ala Ala	Asp Asp Glu Glu	Gly Gly Gly Gly	T C A G

What do proteins do?

- Proteins make you!
 - Your hair is a protein

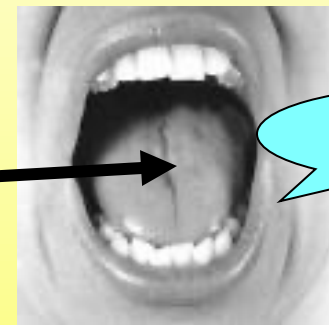
- The protein Keratin

- The sequence of the amino acids of this protein determine it's structure and give you straight, curly, or frizzy hair.



- Your food is digested by proteins

- The protein salivary amylase is in your saliva and begins digestion of carbohydrates like bread and sugar in your mouth.



Mmm!

Evidence for DNA

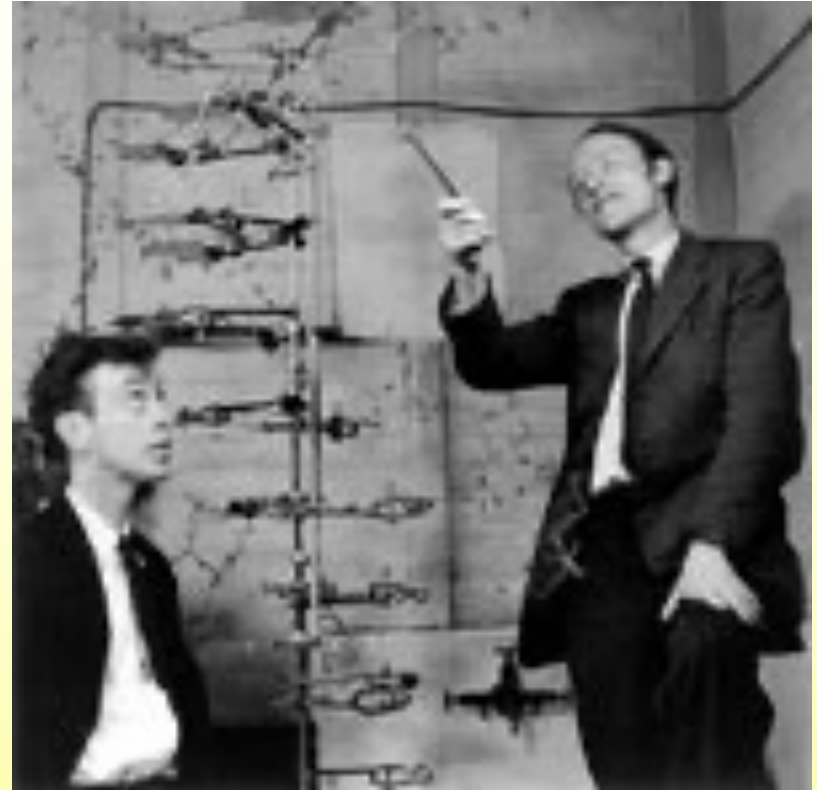
- Freidrich Miescher discovered the substance of DNA in 1868, while researching the nucleus of fish sperm. He did not know it's purpose.
- Scientists predicted that DNA held the information of inheritance, but they weren't sure how.
- Using X-ray diffraction, Rosalind Franklin discovered the structure of DNA as a double helix in 1951. She was not noted for this discovery until her death in 1958.



Rosalind Franklin

DNA

- In 1953, Frances Crick and James Watson modeled the chemical structure of DNA.
- The order of four molecules codes for every part and kind of life.
- Life is incredibly diverse!



Life can show “such unity at the molecular level and yet such spectacular diversity at the level of whole organisms.”

How Genes and DNA are Passed On



How Do Cells Divide?

- Mitosis!
 - All cells replicate and divide through mitosis. This is how we grow and replace aging cells.
 - This involves all parts of the cell.
- DNA is the collection of recipes that codes for life; it is in every cell.
- When a cell replicates and divides, the DNA replicates and divides too.



How does DNA replicate?

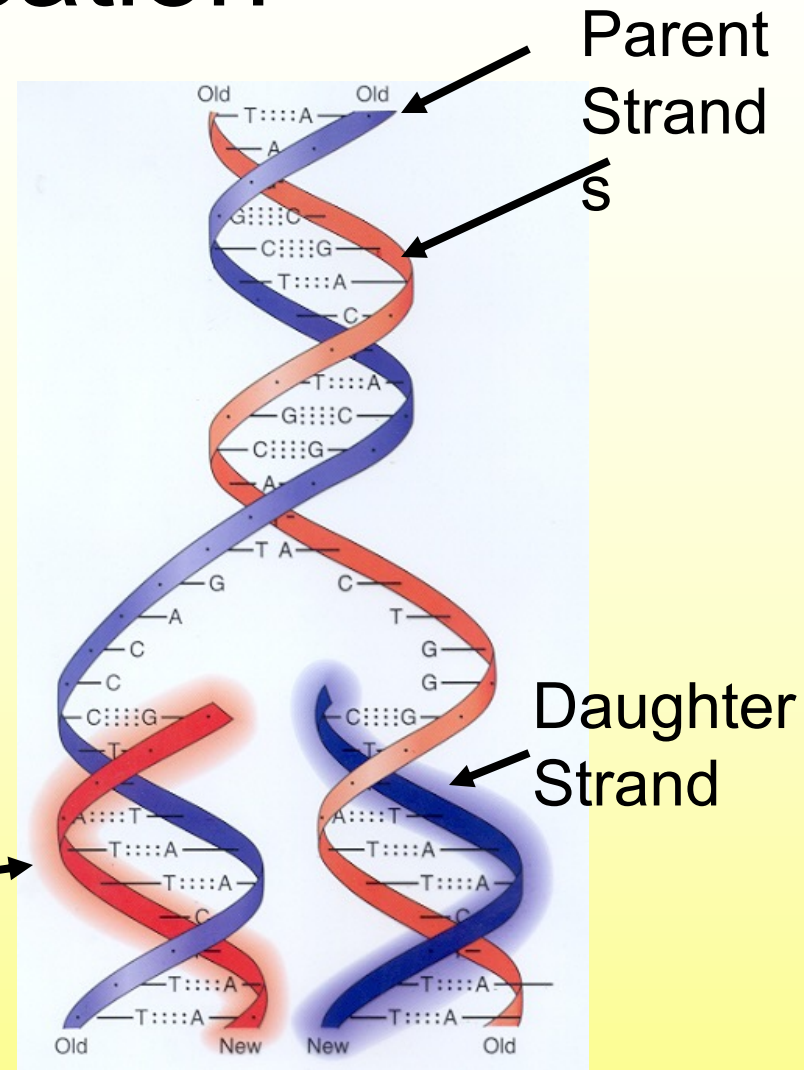
- The two strands of DNA must be separated and copied.
- One protein separates the two strands of DNA.
- Another protein brings the correct new base to pair with the existing base, thus using it as a template.
 - A with T, and G with C (Just like when making amino acids)



DNA Replication

- In this way, two DNA strands are formed from one.
 - Each new DNA double helix has one old strand (the parent strand) and one new strand (the daughter strand).

Daughter Strand



Your DNA

- All of the DNA in a cell of a human is called the “human genome.”
- The human genome has over 3.2 billion base pairs.
 - If you were to string out one cell’s DNA, it would be 6 feet long.
 - How can 6 feet of DNA fit into the nucleus of the cell, in every cell of your body???

DNA Condenses

- DNA tightly wraps around a spool just like kite string. This is a chromosome.
- Condensing a strand of DNA is like taking a very long string, sewing it into a shirt and balling up the shirt into a very tight wad. The string is the DNA.



DNA



Condensed DNA

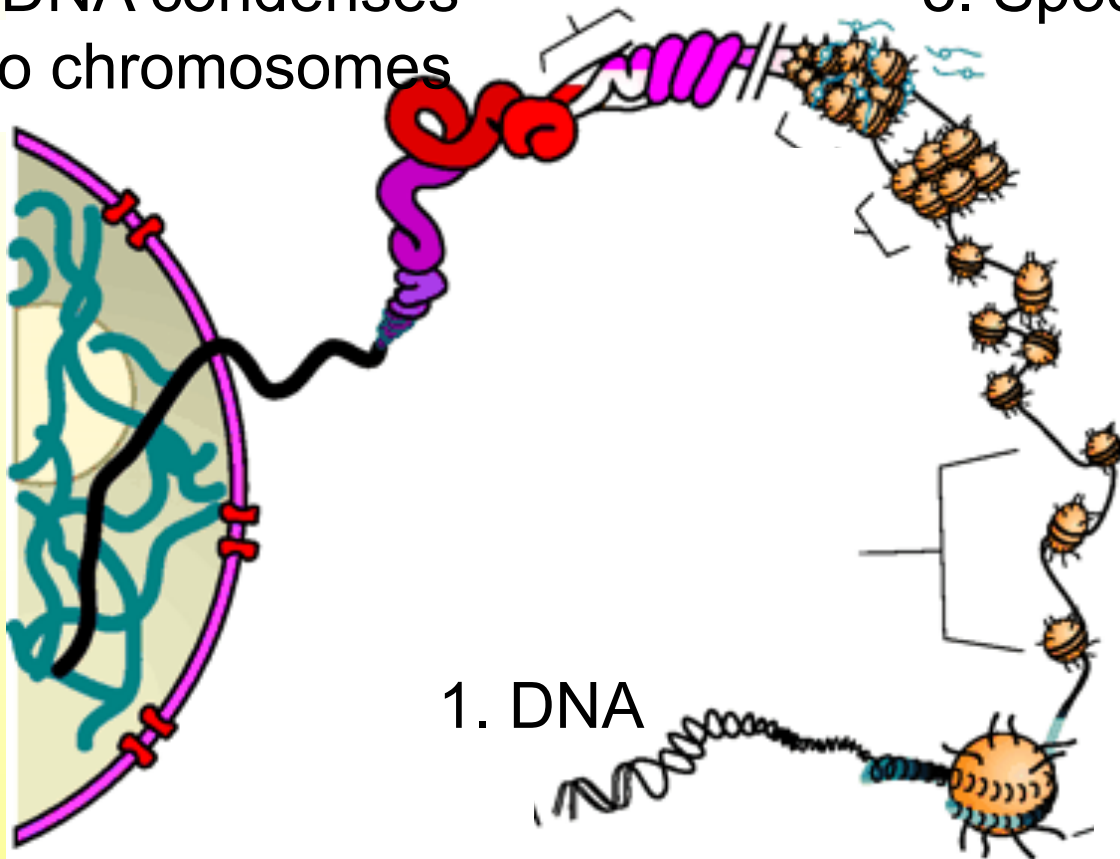


Chromosome

How DNA Condenses

4. DNA condenses into chromosomes

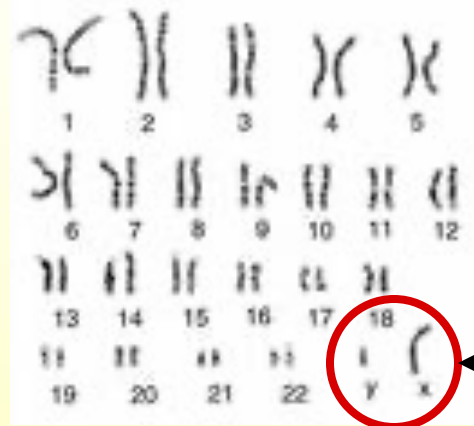
3. Spooled DNA



2. DNA wraps around proteins

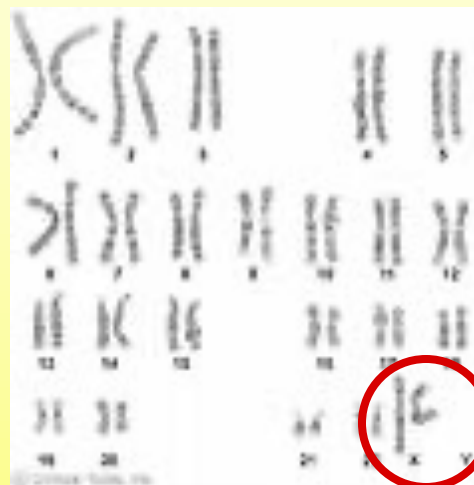
All of the Chromosomes can be seen on a Karyotype

- Karyotypes are made using the amniotic fluid from a pregnant female.
- Karyotypes show
 - the number of chromosomes
 - the sex of the individual
 - large errors in chromosome structure



Typical male karyotype

XY chromosomes

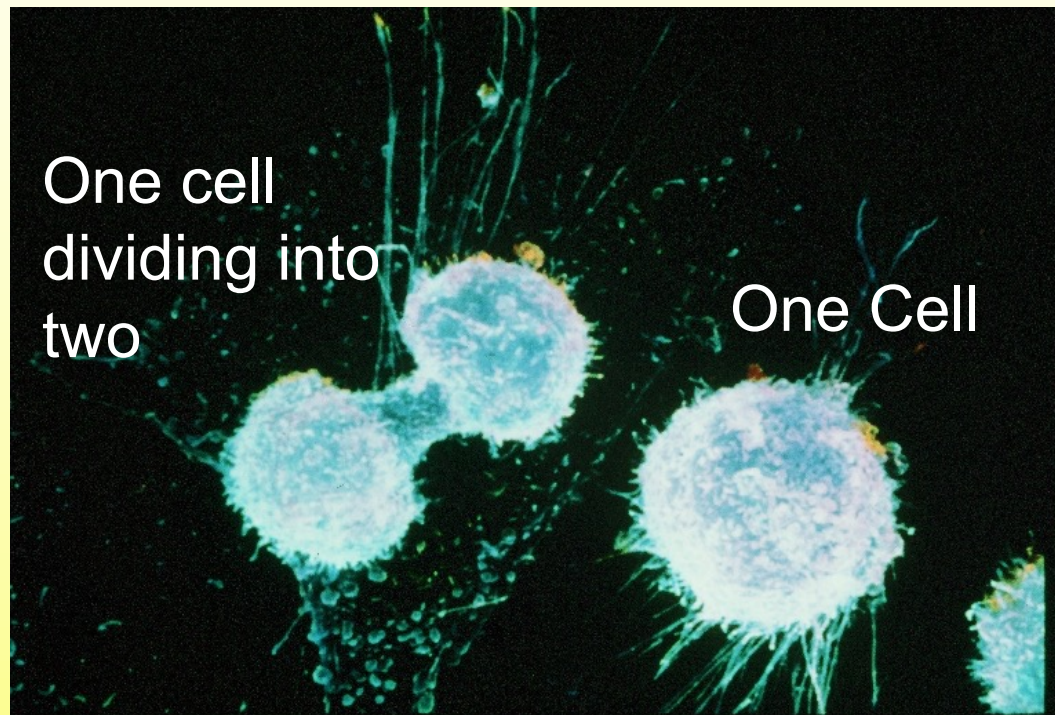


Typical female karyotype

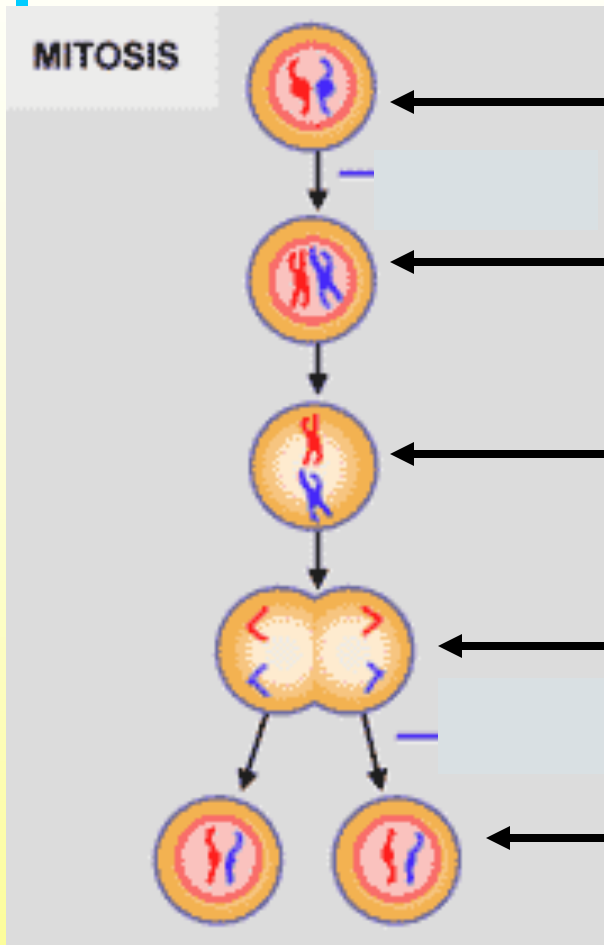
Two X chromosome

Once DNA replicates, how do cells replicate and divide?

- The body grows by one cell dividing into two, and those two dividing into four.
- Skin cells live for many days, blood cells which carry oxygen live for months, nerve cells last a lifetime.



Cells Divide by the Process of Mitosis



Cell with a single copy of DNA

DNA replicates to form chromosomes (two copies of DNA)

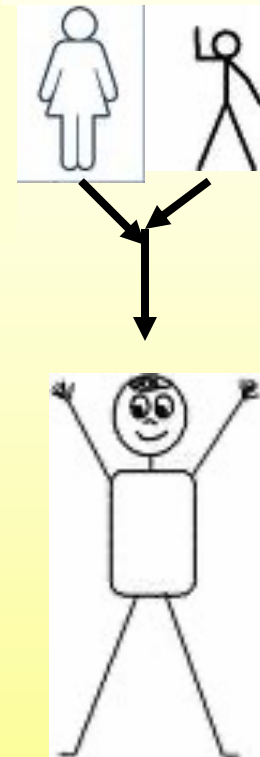
Chromosomes line up in the middle of the cell

Chromosomes are split. Half of each chromosome travels to either end of the cell

The cell divides to form two new cells with their own DNA

How do parents pass on genes to their children?

- Traits are passed on from both parents.
- You have some traits from your mom and some from your dad.
- Because traits are coded for by DNA, you have some of your mom's DNA and some of your dad's DNA. But your DNA is unique to you; no one else has exactly the same DNA as you (unless you are a twin).



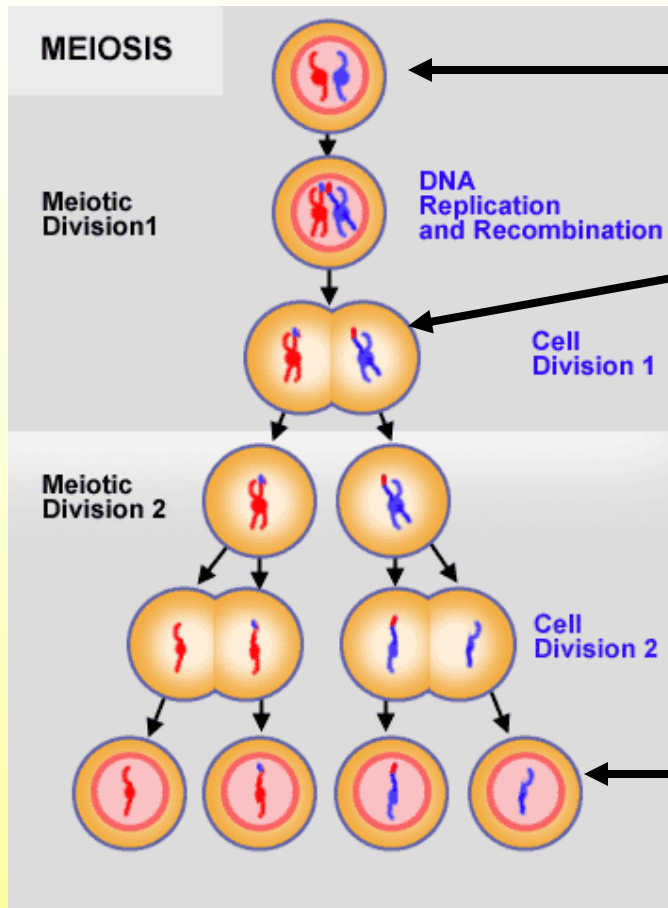
Do the Math

- Humans have 46 chromosomes in each cell.
- If you get 46 chromosomes from each parent you will have 92 chromosomes total, but humans can only have 46.

How do we get 46 chromosomes by getting DNA from two people?

- In the process of meiosis, a cell with 46 chromosomes replicates and divides, making cells with 23 chromosomes in each cell.
- Cells with only 23 chromosomes are called sex cells. They are only in certain parts of the body. Both males and females have sex cells.

Meiosis



Original cell with 46 chromosomes

Some DNA can detach from one chromosome and switch with DNA from the chromosome that holds similar genes. This is called crossing over.

After two cell divisions, four cells with 23 chromosomes each are formed.

How do sex cells form new individuals?

- Sex cells in females are eggs.
- Sex cells in males are sperm.
- One sex cell from the mother (an egg) merges with one sex cell from the father (a sperm) to form a gamete. This cell continues to divide, until it forms a complete organism.
- The individual grows through mitosis.



A Growing Human

Mitosis VS. Meiosis

- Mitosis

- One division
- Forms 2 cells
- Newly formed cells have 46 chromosomes
- Forms normal cells
- Results in growth

- Meiosis

- Two divisions
- Forms 4 cells
- Newly formed cells have 23 chromosomes
- Forms sex cells
- Allows for procreation.

It is biologically beneficial to be different from our parents

- Theory:
 - A child must be similar enough to its' parents to survive in a similar environment, but have the chance to be different enough to survive in a changing environment.

It is a benefit to be genetically different from our parents, in case we must survive in an environment that is different from the one they live in. With different genes, we may be better prepared.



People in drastically different environments have slightly different genes.



How are our traits a combination of our parents traits?

- Traits are not mixed when they are passed on.
 - For example, if your mom has blue eyes and your dad has yellow eyes, you will not have green eyes. You will either have blue or yellow eyes.
- Some traits are determined by dominance.
 - If you have two genes in you that code for the same trait, one trait will win over the other. The trait that “wins” is the dominant gene. The trait that does not get expressed is the recessive gene.
- Some traits, like height, are determined by genes from both parents. There is not one dominant gene for these traits.

Dominant Traits:

-attached or unattached earlobes

-widow's peak hairline

Combination Traits:

-height

-hair color

Dominant Traits

Each person has two genes for every trait.

- Dominant physical characteristics will show up unless both genes code for the recessive trait.

Dominant trait: Unattached earlobes, “U”

Recessive trait: Attached earlobes, “A”

If your genes code: You will have:

UU → Unattached

UA → Unattached

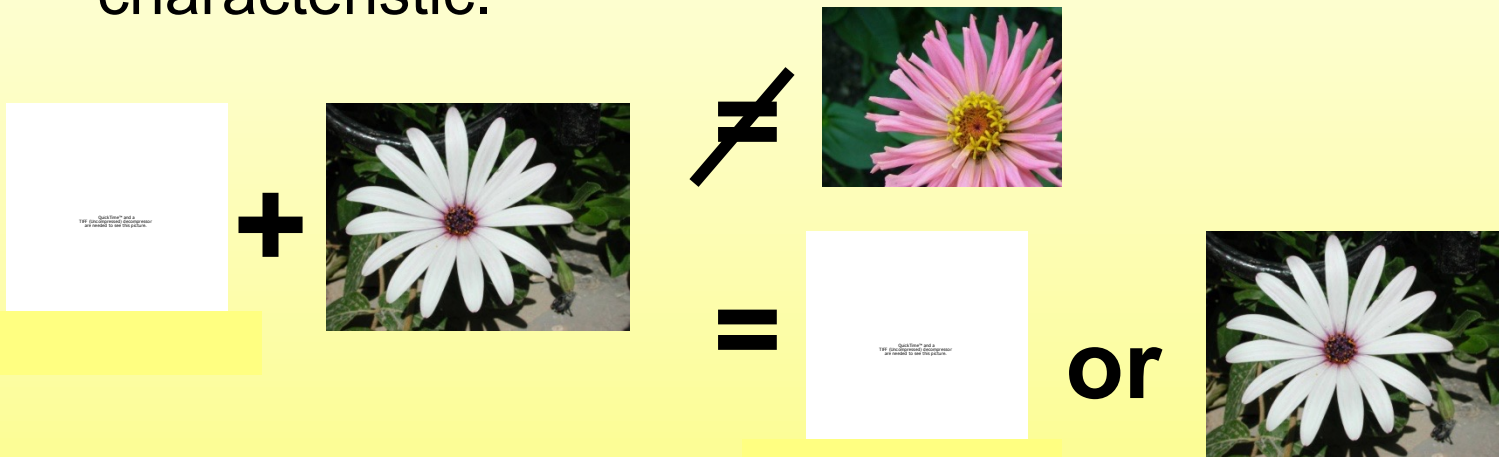
AA → Attached



Attached Unattached
“A” “U”

Evidence for Dominance

- In the late 1800's, Gregor Mendel, a monk, was studying pea plants.
- He noticed that when he bred two plants with different traits, their offspring did not show a mix of characteristics, but instead showed either characteristic.



Predicting Physical Characteristics

- When a scientist knows the characteristics of the parents, he or she can predict the characteristics of the offspring using a Punnett-Square
- Genes of both parents are labeled on the outside of the big square.
- Each combination of the parent's genes in the small squares is a possible combination for the offspring.

Genes of Parent 1:		A	B
Genes of Parent 2:	A	AA	AB
	B	AB	BB

Offspring could be:
AA, AB, AB, or BB

Probability (P)

- By knowing which genes the parents have, you can figure out the probability those parent's have of having a child with a certain trait.

If the parents are both dominant (DD):

	D	D
D	DD	DD
D	DD	DD

4 out of 4 children will have the dominant trait.

$$P = 4/4$$

If the parents are both dominant but also carry the recessive gene (DR) R = recessive

	D	R
D	DD	DR
R	DR	RR

3 out of 4 children will have the dominant trait.

$$P = 3/4$$

P = Probability
It's a fraction!

Try It

Dominant Trait: Straight Thumb (S)

Recessive Trait: Hitchhiker's Thumb (H)



S



H

If Parent 1 has Hitchhiker's Thumb (HH)

And Parent 2 does not (SH)...

Parent 1:

H

H

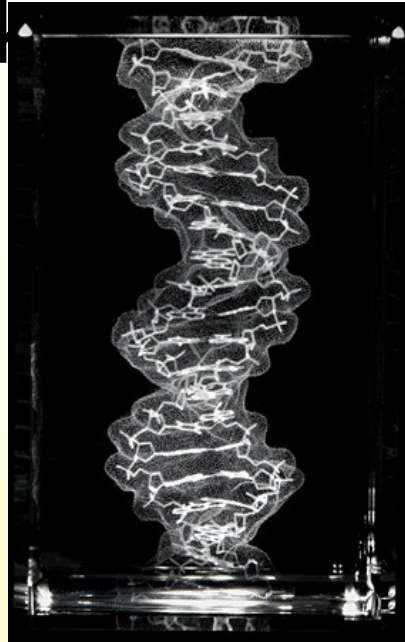
Parent 2:	S	SH	HH
	H	HH	HH

What probability is there that a child will have hitchhiker's thumb (the recessive trait)?

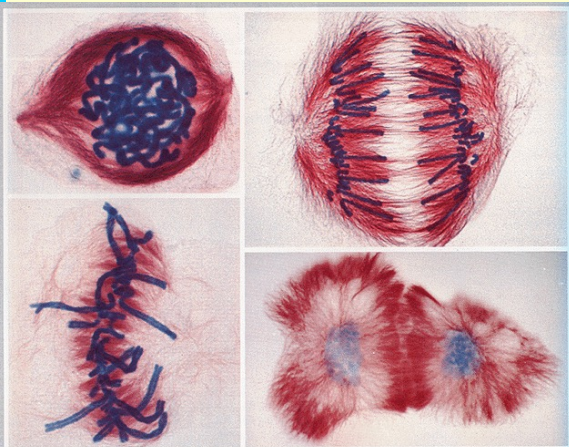
3 out of 4 = $\frac{3}{4}$

*Dominant traits are not always the most common.

This is how the
body works



And this is what we
can do...



Plant cells in various stages of mitosis: (a) prophase; (b) metaphase; (c) anaphase; (d) telophase (all magnified about 2,700 times).

Genetic Disorders

Disorder, Cause, and Result



Genes Make You...

But what happens if your body doesn't work exactly as it is supposed to?

- Genetic Disorders
 - Genetic Disorders result when there is a change in your genes that changes the way your body functions.
 - Sometimes the change can be so large that your body cannot function.
- Changes can occur at any stage in DNA replication, mitosis, or meiosis.

Mutations

- All genetic disorders are caused by a mutation
- Mutation: A change in the genetic base-code for a protein.
- A mutation can occur at almost any stage in development
 - DNA replication ,mitosis, meiosis, chromosome separation.
- Environmental factors can lead to mutations as well.
- Mutations can be beneficial, harmful, or neutral.

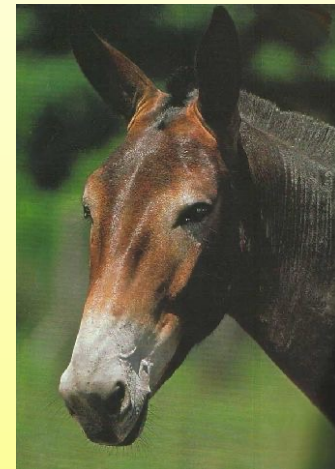


Horse

+



Donkey



Mule

Types of Mutations



Beneficial Mutations: Dark and light skinned people.

Dark skin: + resistant to sunburn

- generate less Vitamin D

*Increased survival in sunny environments, like the equator.

Light skin: + generate more Vitamin D

- prone to sunburn

*Increased survival in less sunny environments.

Harmful: Mutations in genes coding for proteins that control growth; this results in un-controlled cell growth.

***This decreases a person's chance of survival.

Neither: Attached earlobes

***This does not increase or decrease a person's chance of survival.

Common Genetic Disorders

Disorder

- Sickle-Cell Anemia
- Down Syndrome
- Lactose Intolerance
- Colorblindness

Mutation

- Change in one base pair
- Chromosomes do not separate evenly in meiosis
- Gene does not produce particular protein that digests sugars in milk
- Multiple genes that allow us to see color are not coded for (on X chromosome)

Common Genetic Disorders

Disorder

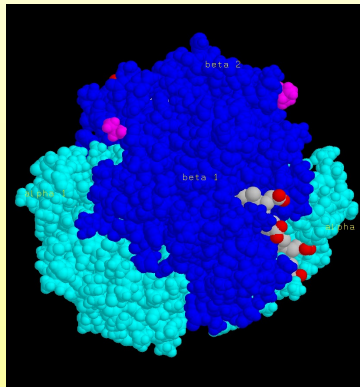
- Muscular Dystrophy
- Alzheimer Disease
- Cancer

Mutation

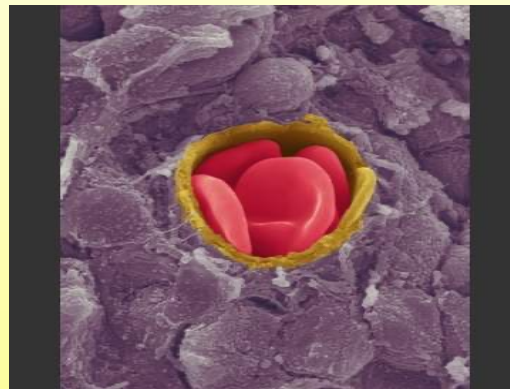
- Two recessive genes (passed from parents or develops over time)
- Multiple genes and environmental effects; gene coding for protein that interferes with nerve shape is over produced
- Multiple genes and environmental effects; changes in genes that code for growth

Small Changes: Sickle Cell Anemia

- Individuals with Sickle Cell Anemia have oddly shaped red blood cells.
 - The shape of the mutated protein allows the cell to bind to itself, changing the cells shape.
 - The shape of the red blood cell doesn't allow it to flow easily through the body, disabling the individual.



Typical protein in red blood cells



Red blood cell with typical protein flowing through body

*Red blood cells with mutated proteins would get stuck in such small openings and cause clogs.

Small Changes: Sickle Cell Anemia

- Sickle Cell Anemia occurs in individuals that have one single change in the order of their base pairs.
 - Adenine changes to Thymine
 - This causes the coded amino acid to change from Glu to Val.
 - This changes the structure of the protein.

Nucleotide: GTG ← GAG

Amino Acid: Val ← Glu

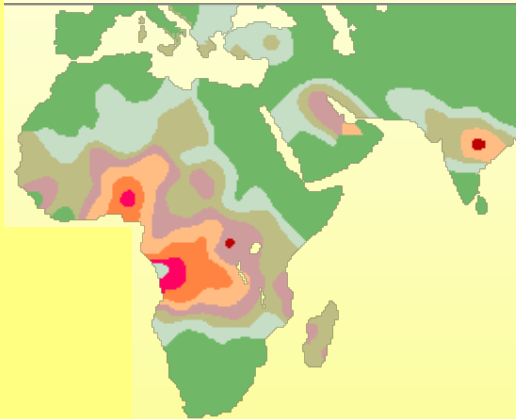


Blood cell
with
Sickle Cell
protein

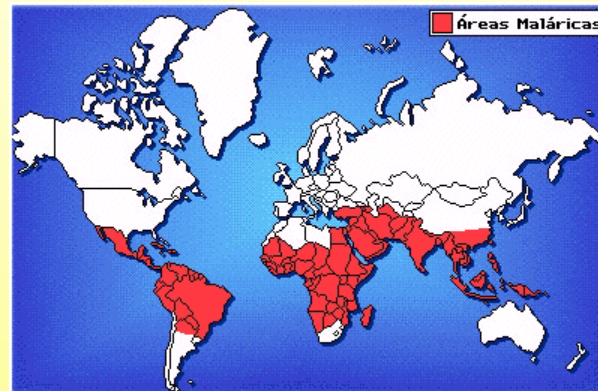
Blood cell with
normal protein

Health Note

- Sickle-Cell Anemia is a recessive disorder; the individual must have both recessive genes to have the disorder.
- People with one dominant and one recessive gene for the protein that causes Sickle-Cell Anemia are resistant to Malaria, a deadly disease spread by mosquitoes.



Sickle-Cell Anemia common in red, orange, peach, and purple areas



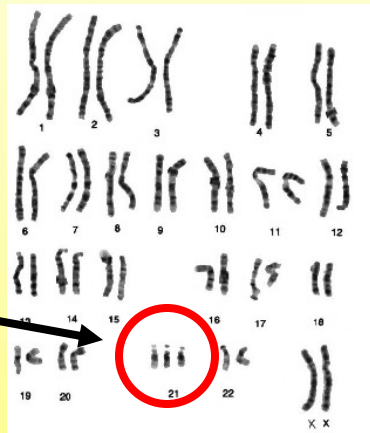
Malaria is common in regions in red

Sickle-Cell Anemia is common in parts of the world that are heavily impacted by Malaria

Large Changes: Down Syndrome

- Down Syndrome is one of the few genetic disorders where an individual can survive with an extra chromosome.
- People with Down Syndrome tend to have large foreheads and slight mental retardation.
- The extra chromosome can be seen in a karyotype.

Three copies of chromosome 21 instead of two.

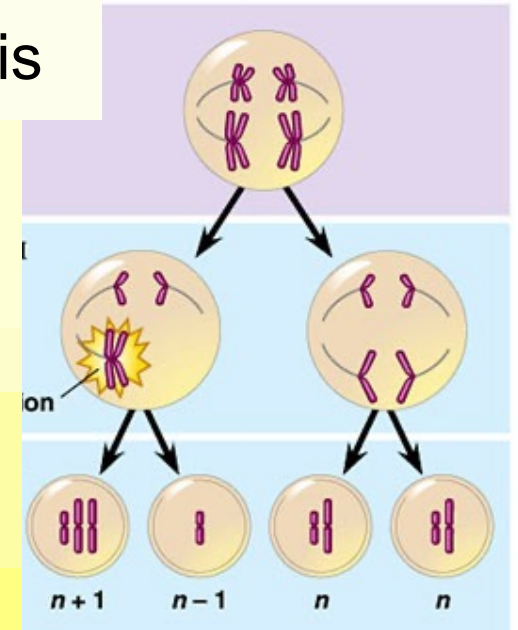


An individual with Down Syndrome

How Down Syndrome Occurs

- In meiosis, chromosomes do not split into different cells evenly:
One cell gets 3 chromosomes, while one cell gets one.
- The individual with three chromosomes 21 will most likely survive, but will have Down Syndrome.
- The individual with one chromosome 21 will not survive.

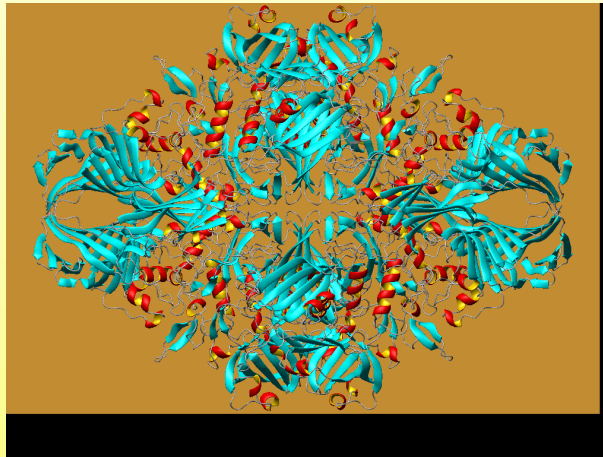
Meiosis



↑
Individual with Down Syndrome gets this cell from one parent.

Lactose Intolerance

- Individuals with lactose intolerance cannot digest the sugar in dairy products (lactose).
- They do not produce the protein (lactase) that breaks down lactose; this is due to four mutations within the gene that makes lactase.
- When lactose intolerant people drink milk or other dairy products, undigested lactose builds up in their stomach, making a great environment for bacteria. The bacteria thrive and reproduce very quickly. This creates bloating and sickness.



Protein: Lactase
*Not produced in
individuals with
lactose intolerance

Mutations in Lactase Gene

Two of the four mutations are “point” mutations.

- Point mutation: One base pair in the DNA sequence in the gene has been changed. This affects only one amino acid.

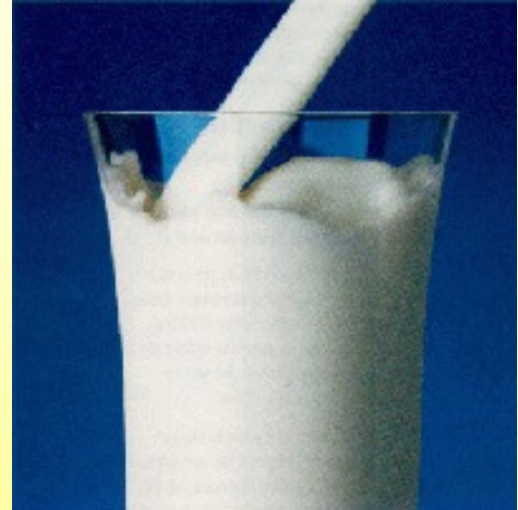
Original DNA: AGC CAT AGG

Amino acids: Ser His Arg

Mutated DNA: AGC CAA AGG

Amino acids: Ser Glu Arg

*His changes to Glu



Mutations in Lactase Gene

Two of the four mutations are “frame shift” mutations.

- Frame shift mutation: One base has been inserted into the gene; the rest of the bases are moved over one. This affects many amino acids.

Original DNA: ATT CGT TAC GAA ACG

Amino acids: Iso Arg Tyr Glu Thr

Mutated DNA: ATT CCG TTA CGA AAA CG

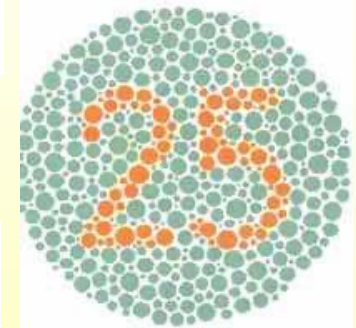
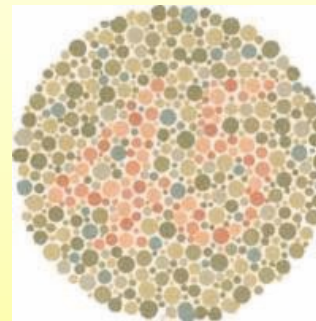
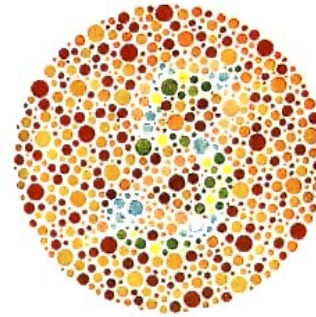
Amino acids: Iso Pro Leu Arg Lys

*C inserted. Remaining bases shifted over one and amino acids changed.



Colorblindness

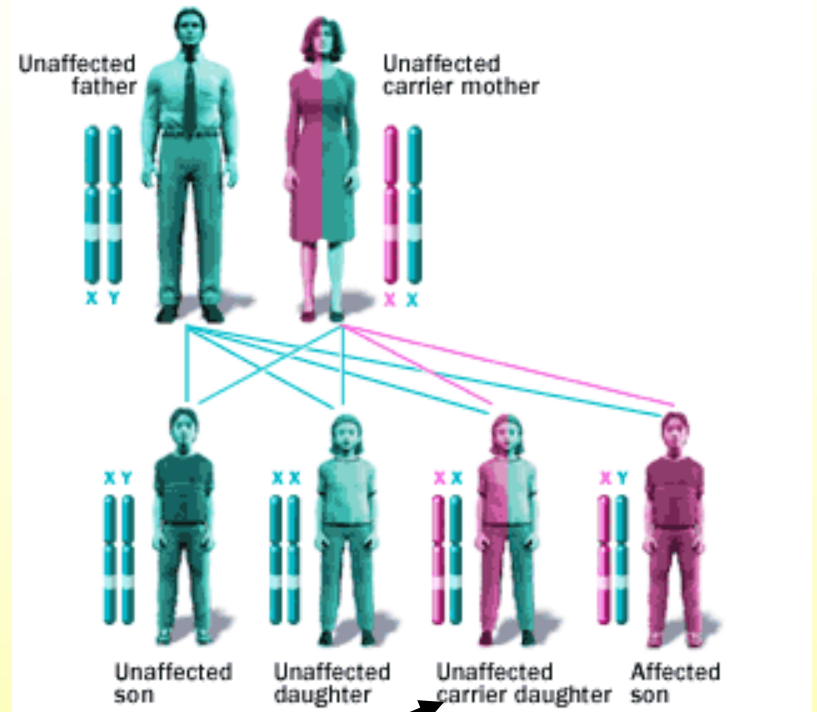
- Caused by multiple mutations on X chromosome.
- Males have one X chromosome and one Y chromosome.
- Females have two X chromosomes.
- When genes on one chromosome do not code correctly, the body naturally goes to the other chromosome to see if it holds a more usable genetic code.
- Males do not have this advantage.
- If the one X chromosome that a male has mutated genes coding for color receptors, the male will be color blind.
- This is called “X-linked Inheritance”



These circles are used to test colorblindness. If you can't see the number in each circle, you are colorblind

X-Linked Inheritance

- A male has one Y chromosome and one X chromosome
- Because his father is the only parent that carries the Y chromosome, the son must inherit his Y chromosome from his father and the X chromosome from his mother.
- A male inherits the colorblindness gene from his mother.
- Females can be colorblind if the mother holds the mutated gene and the father is colorblind.
- Other X-Linked traits:
 - Muscular dystrophy
 - Hemophilia

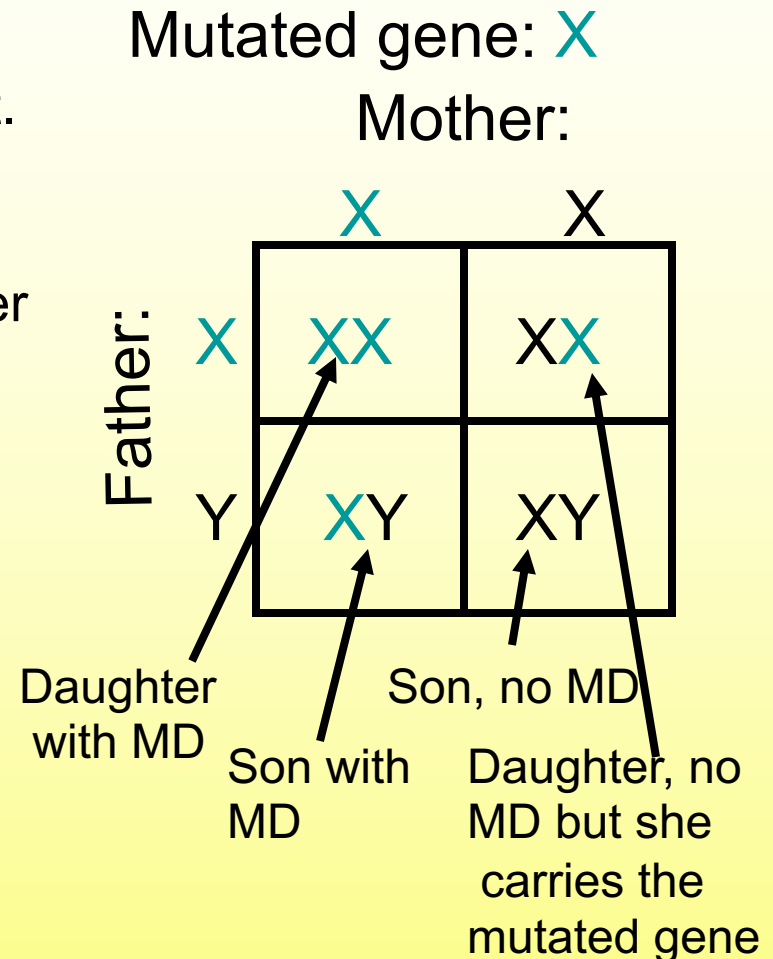


Daughter with ability to pass on colorblind gene

Colorblind son

Muscular Dystrophy (MD)

- Individuals with muscular dystrophy have muscles that shrink over time.
- Muscular dystrophy is an X-Linked trait.
- Males have muscular dystrophy if it is passed from his mother.
- Females have muscular dystrophy if her father has it and her mother carries the gene for it.
- It is a recessive condition in females
 - To be affected by the mutation, a female must have both mutated genes.
- 1 out of 10,000 people have MD



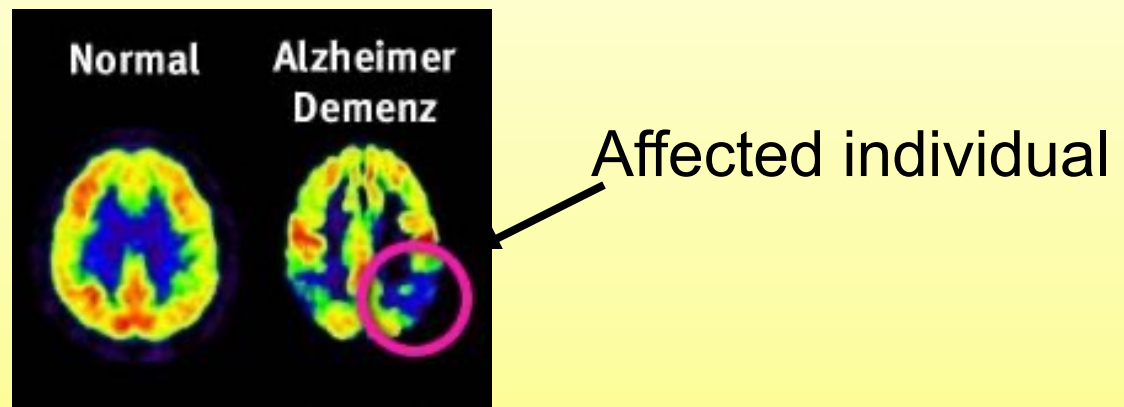
Affects of Muscular Dystrophy

- A very small change in the genome results in a very noticeable physical change...
- The rest of the body still thrives!



Alzheimer Disease

- Individuals with Alzheimer disease lose the ability to create and keep memories.
- This genetic disease is thought to be due to a combination of environmental and genetic effects.
- Scientists do not know which genes affect Alzheimer Disease.
- They hypothesize that a certain protein that affects the structure of nerves is over produced. This destroys nerves in the brain that are responsible for storing memories.



MRI of the Brain

Cancer

- Like Alzheimer disease, cancer is caused by both genetic mutations and environmental effects.
 - Environmental effects, such as excess sun exposure, are thought to be a partial cause of genetic mutations.
- There are many types of cancer, but they all involve un-controlled cell growth.



Lance Armstrong,
7 time winner of the
Tour de France,
Testicular cancer



Nancy Reagan,
Former First Lady,
Breast cancer



Bob Marley,
reggae musician,
Died of skin cancer
in 1981

Cancer

- Mutations occur in three types of genes to cause cancer (un-controlled cell growth):
 - 1. Genes that promote normal cell growth are mutated and function at a higher level.
 - 2. Genes that stop cell growth are mutated and do not function.
 - 3. Genes which code for proteins that repair DNA mutations are mutated.
 - Genes that code for proteins that check for mutations in DNA act as a second protection against mutation.
 - Without this check system, cancer is extremely deadly.

If mutations are harmful, why are there mutations?

- Mutations occur in individuals by chance.
- If the environment is quickly changing, a mutation may benefit an individual to survive better in the new environment.
- While some mutations are harmful, organisms need mutations to adapt to a changing environment.



Animals in this environment, like fish, have adapted to the water with fins and gills.

It would take a species that lived in a lake many mutations to be able to survive in a desert.

Animals in this environment, like lizards, have adapted to the dryness with legs and being cold-blooded.



Even with harmful mutations,
we persist

