Science 10 Unit 1

GENETICS

1.1 DNA Structure and Function

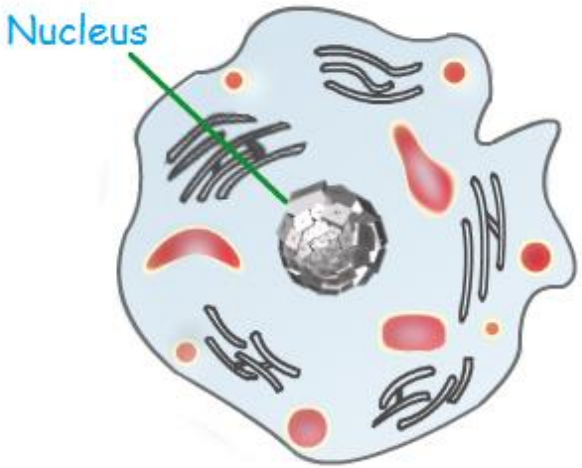
Part I- The Nucleus: Control Centre of the Cell

- Every cell in your body has a specific **JOB-** but how do they become specialized?

  E.g. hair cells vs. skin cells vs. retina in the eye

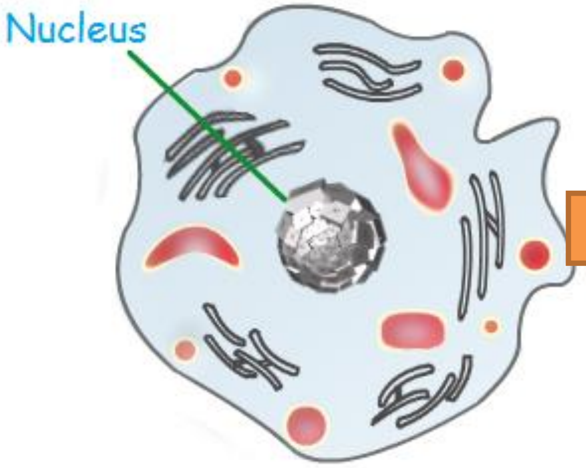
The **NUCLEUS** in the cell contains the master set of instructions that tells the cell:

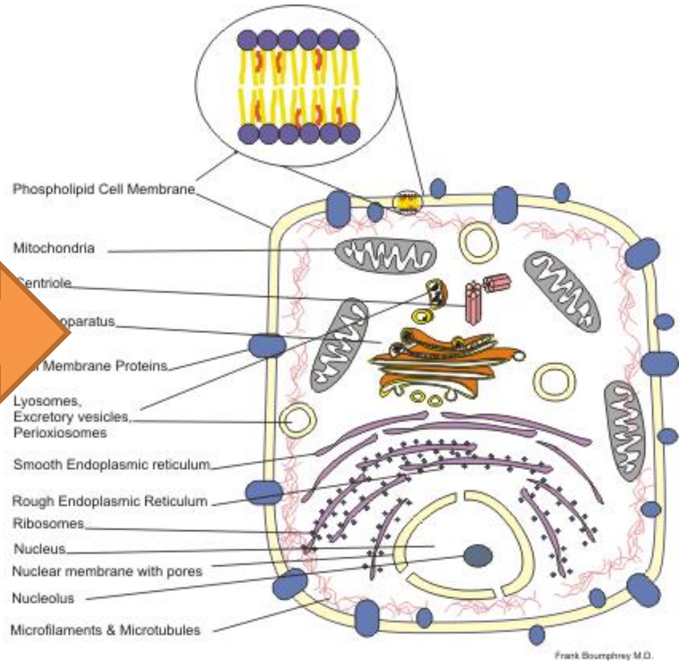
- what it will **BECOME**
- how it will function
- when it will **REPRODUCE** and **GROW**
- when it will die



1.1 DNA Structure and Function

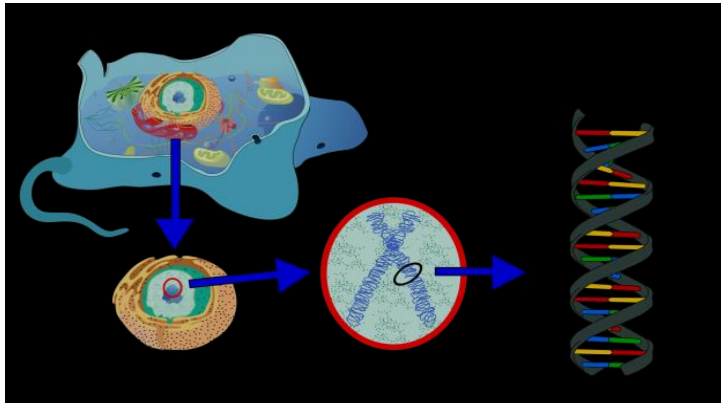
- *But how does the nucleus do this? How does it send messages to the rest of the cell?*





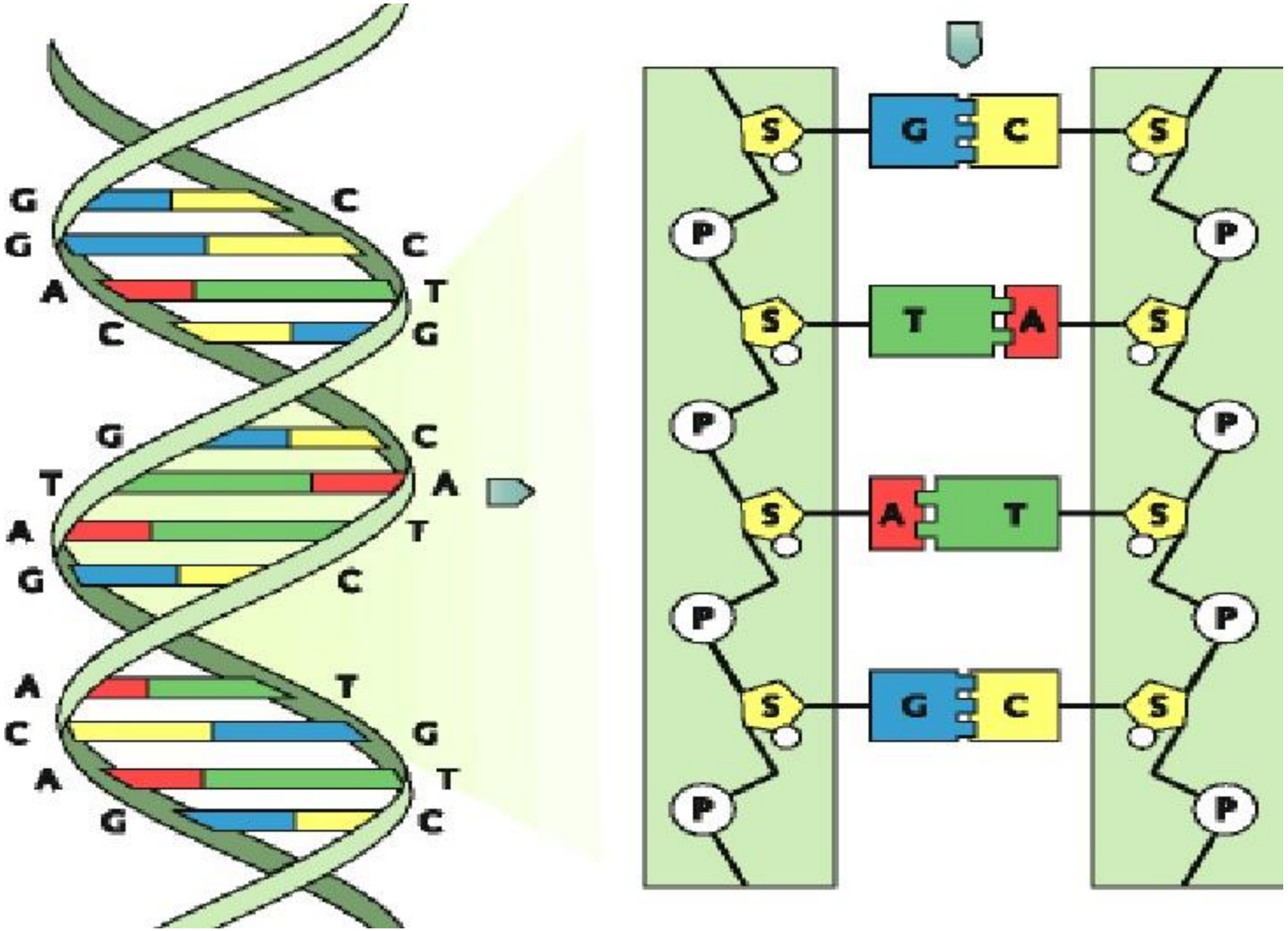
1.1 DNA Structure and Function

The nucleus contains <u>DNA</u>, which carries the master set of <u>INSTRUCTIONS</u> for cell function.



- DNA (**<u>DEOXYRIBONUCLEIC ACID</u>)** is a double stranded helix that looks like a twisted ladder
- the sides of DNA are made of **<u>SUGAR</u>** and **<u>PHOSPHATES</u>**
- the steps of DNA are made up of 4 **<u>BASES:</u>**

Adenine	Cytosine
Guanine	Thymine



G C
G C
A T
C G
G C
T A
A T
G C
A T
C G
A T
G C
S G C S
P P
S T A S
P P
S A T S
P P
S G C S
P P

1.1 DNA Structure and Function

- Where are the bases located?
- Name the 4 bases
- Where are the sugars and phosphates located?
- What is this shape called?

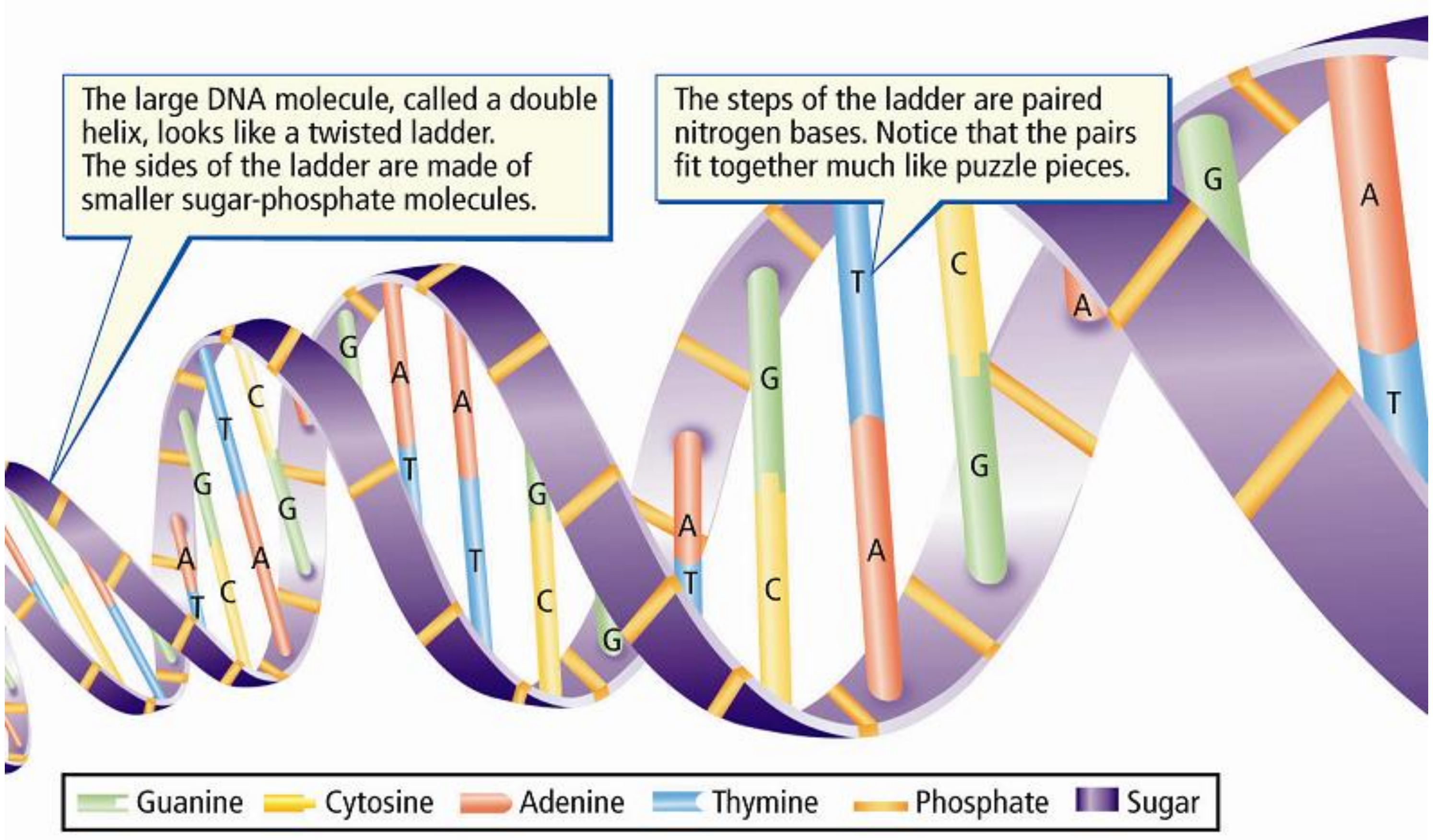


Figure 4.5 The structure of the DNA molecule

1.1 DNA Structure and Function

Pairing Rules

- adenine (A) always pairs with **THYMINE** (T)

  → Apple Tree

- cytosine (C) always pairs with **GUANINE** (G)

  → Car Garage

1.1 DNA Structure and Function

2. DNA has many functions:

A. DNA contains **INSTRUCTIONS FOR ALL CELL FUNCTIONS** and, therefore, DNA indirectly controls all of the functioning of all living things.

B. DNA **DETEMINES THE HEREDITARY TRAITS** of an individual

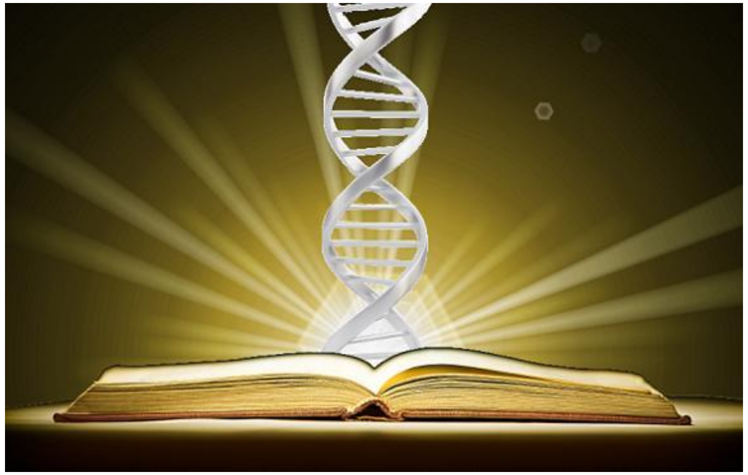
C. DNA **EVOLVES** (changes through mutations and recombination). This allows for new characteristics & abilities to appear which may help an individual to survive & reproduce.

D. Self replication: DNA has the ability to **MAKE COPIES OF ITSELF.**

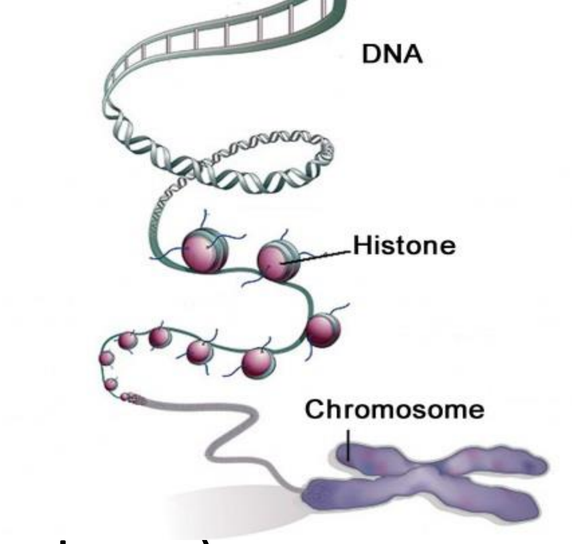
1.1 DNA Structure and Function

3. **The arrangement of bases in DNA directs all cell activity**

- the bases are like letters that carry a message (**CODE**)
- the code gives **INSTRUCTIONS** for a specific task.



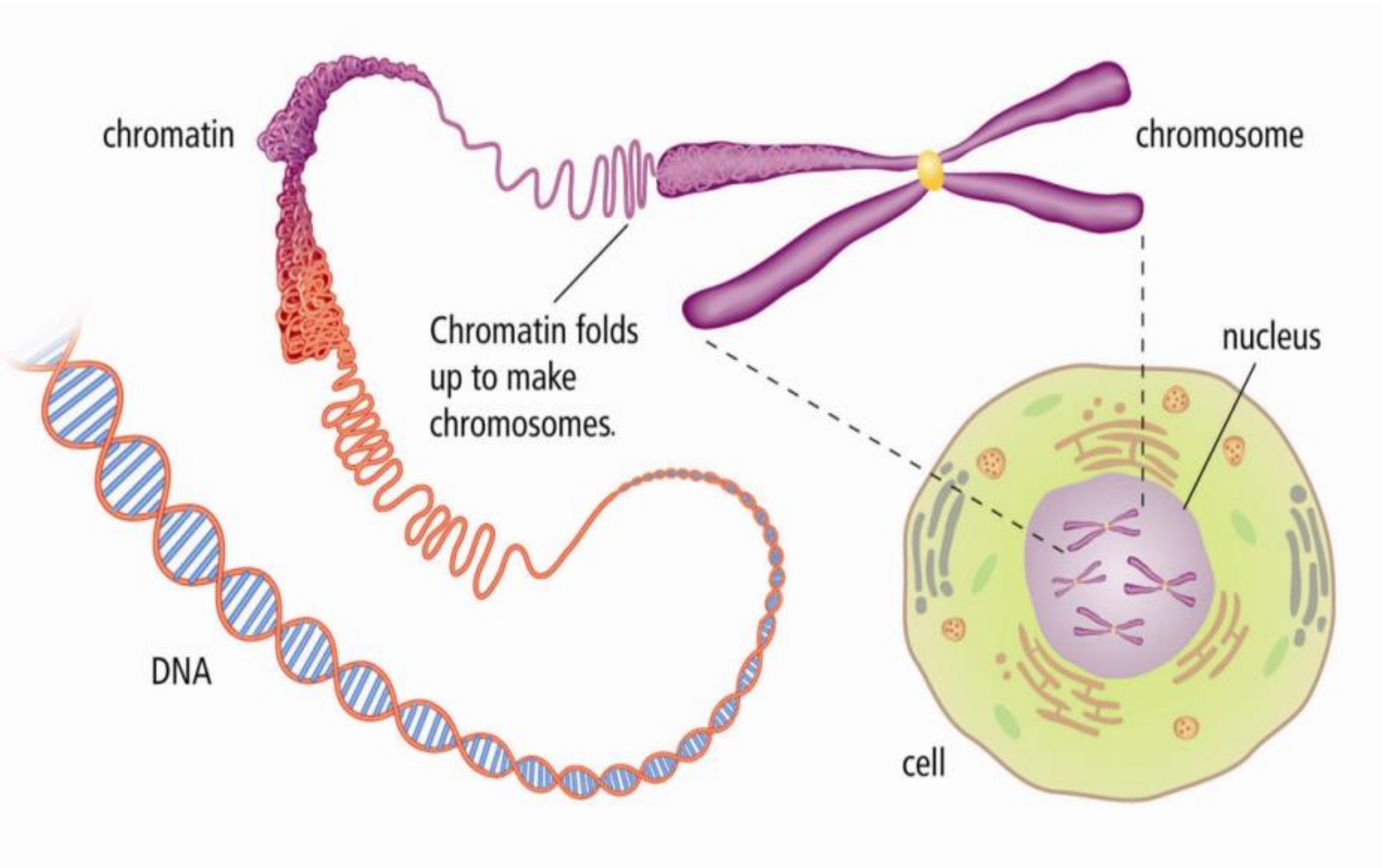
1.1 DNA Structure and Function



4. **DNA is stored in the form of chromatin**

- chromatin is made up of DNA and **PROTIENS** called histones (very dense)
- when a cell is growing, parts of the **CHROMATIN** unwind so that the targeted section of DNA can be read to make messages that control the rest of the cell.
- when cells reproduce, the entire chromatin coils up and makes an X-shaped structure called a **CHROMOSOME**

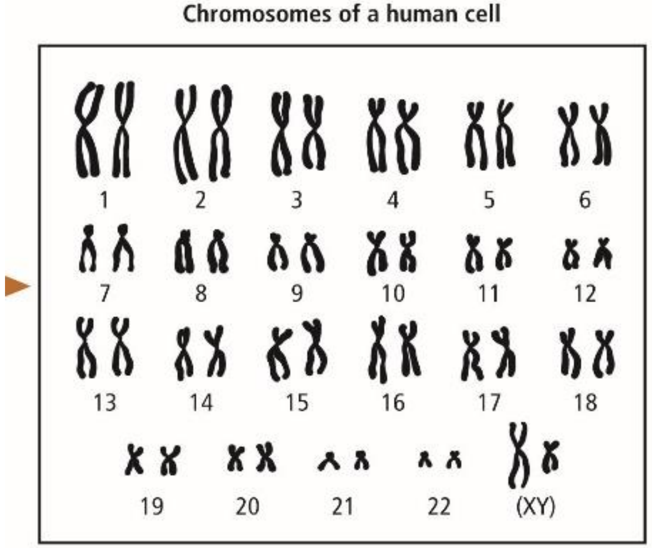
Some terms you should know….

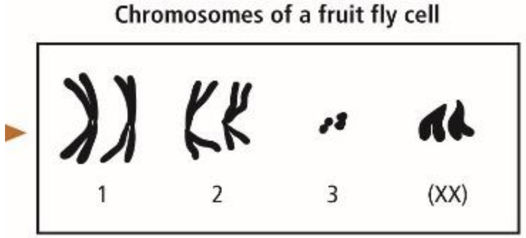


1.1 DNA Structure and Function

5. **Every organism has a characteristic number of chromosomes**

- chromosomes are always in **PAIRS** in the nucleus
- humans have 46 chromosomes (=**23** pairs)
- in males, the 23rd pair of chromosomes are **"XY"**
- in females, the 23rd pair of chromosomes are **"XX"**
- cows have 60 chromosomes; corn has 20 chromosomes





1.1 DNA Structure and Function

6. Genes are found on chromosomes

- A **<u>GENE</u>** is a small segments of DNA found at specific places on a chromosome that code for a protein
- genes can vary in length from 100s to 1000s of **<u>BASES</u>**
- the arrangement of bases will decide what kind of protein is produced

e.g. ACCATAGG → make protein “A”

AGGCGTTA → make protein “B”

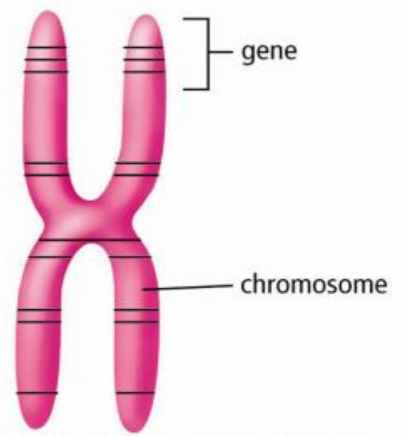
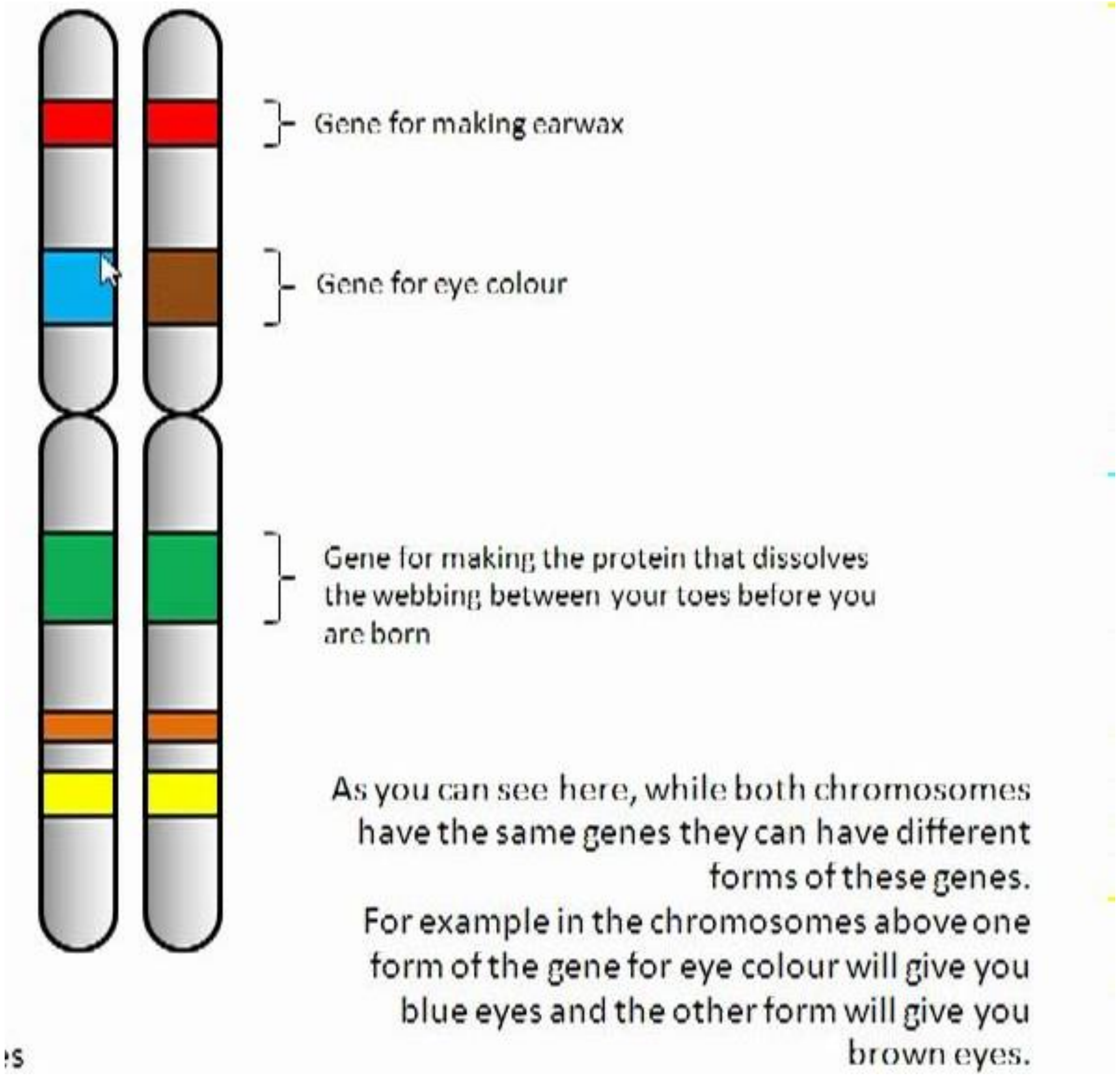


Figure 4.9 Genes are located on chromosomes and contain the information to produce a protein.

1.1 DNA Structure and Function

- each chromosome carries 1000s of **GENES**
- your body uses 90 000 to 100 000 different **PROTEINS**



1.2 Protein Synthesis

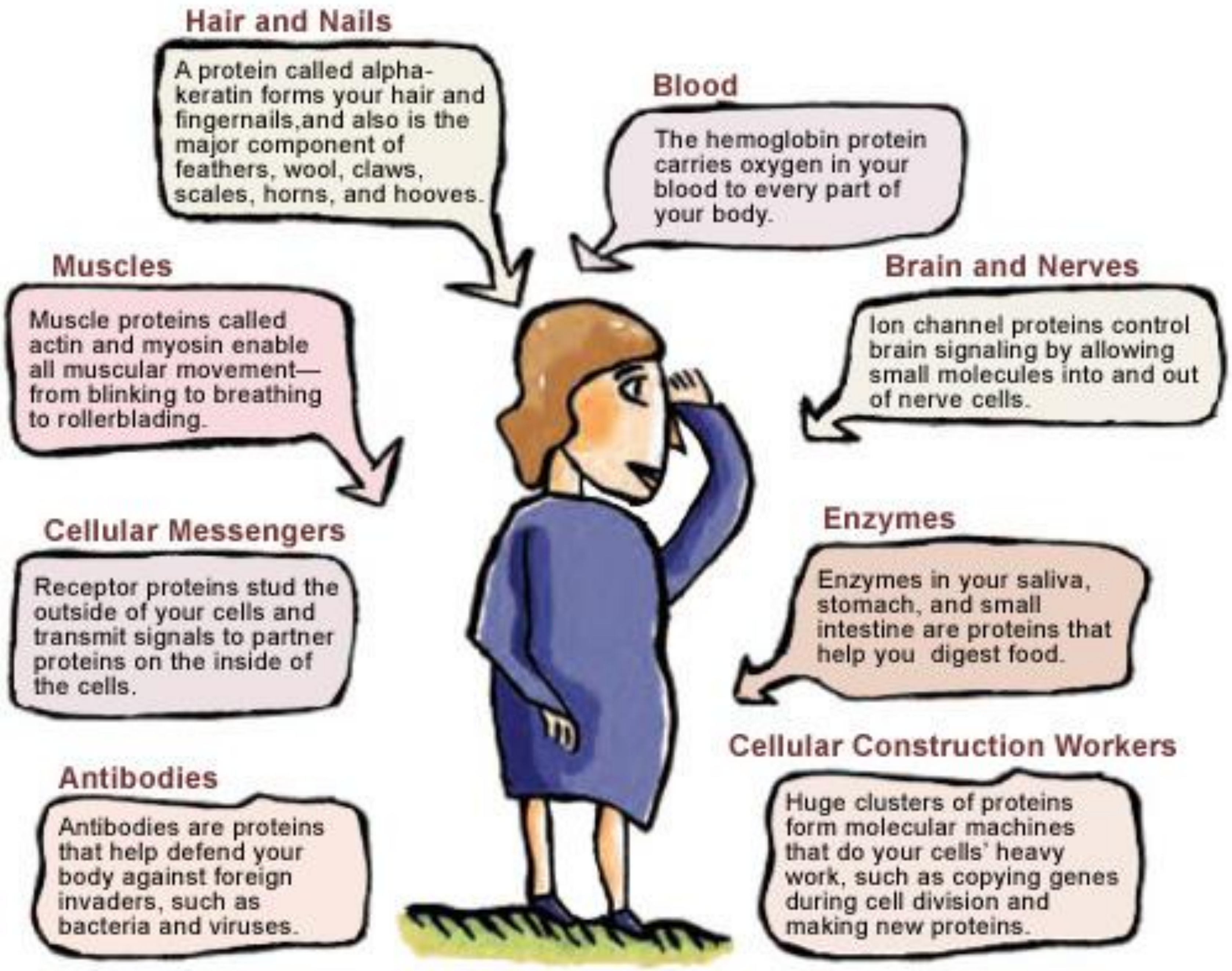
Part I- The Role of Proteins

Why are proteins so important anyways?

- Humans share most of the same protein families with **WORMS**, flies, and plants
- Hair grows by forming new cells at the base of the root. As they move upward through the skin they are cut off from their nutrient supply and start to form a hard protein called **KERATIN.** As this occurs, the hair cells die. The dead cells and keratin form the shaft of the hair.

1.2 Protein Synthesis

- Fingernails grow about **THREE TO FOUR** times as quickly as toenails
- Each hair grows about **1/4 INCH**/month and grows for up to **6** years.
- The most expressive muscles are the facial muscles. We need **17** muscles to smile and **43** muscles to frown.
- The most numerous are the skeletal muscles. When we walk for instance, we use **200** muscles.


Hair and Nails
A protein called alpha-keratin forms your hair and fingernails,and also is the major component of feathers, wool, claws, scales, horns, and hooves.
Blood
The hemoglobin protein carries oxygen in your blood to every part of your body.
Muscles
Muscle proteins called actin and myosin enable all muscular movement—from blinking to breathing to rollerblading.
Brain and Nerves
Ion channel proteins control brain signaling by allowing small molecules into and out of nerve cells.
Cellular Messengers
Receptor proteins stud the outside of your cells and transmit signals to partner proteins on the inside of the cells.
Enzymes
Enzymes in your saliva, stomach, and small intestine are proteins that help you digest food.
Antibodies
Antibodies are proteins that help defend your body against foreign invaders, such as bacteria and viruses.
Cellular Construction Workers
Huge clusters of proteins form molecular machines that do your cells' heavy work, such as copying genes during cell division and making new proteins.

1.2 Protein Synthesis

Proteins Have 2 Main Functions

1. Structural: proteins help make up all structures in living things

Examples: a) **MUSCLE PROTEINS**

b) **HAIR, NAILS, BONES**

c) **BLOOD VESSELS, LIGAMENTS**

1.2 Protein Synthesis

2. Functional: other proteins help us to keep our bodies functioning properly and to digest our food.

Examples:

a) **ENZYMES TO SPEED UP CHEMICAL REACTIONS**

b) **HAEOMOGLOBIN IN RED BLOOD CELLS**

c) **ANTIBODIES USED IN THE IMMUNE SYSTEM**

d) **TRANSPORT PROTEINS FOUND IN CELL MEMBRANES**

1.2 Protein Synthesis

- Only certain genes are **ACTIVATED** in a cell
- Depending which genes are active, different proteins are produced, and this causes cells to have different functions.

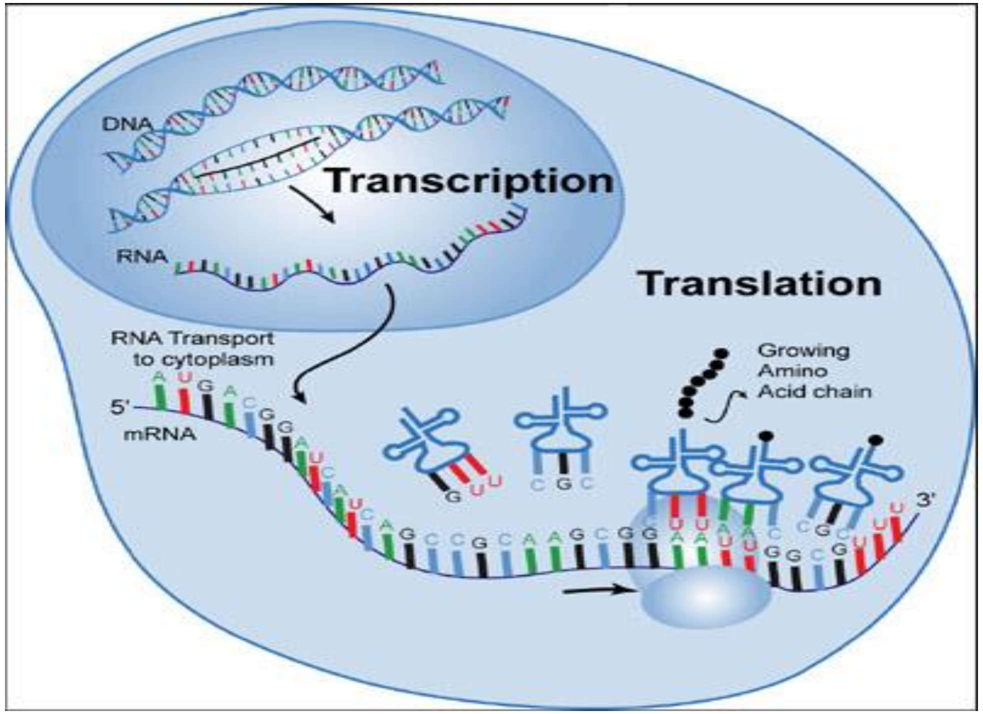
  E.x. You do not have skin pigment genes being used by your stomach cells

- **ONE GENE → ONE TYPE OF PROTEIN → ONE FUNCTION**

1.2 Protein Synthesis

Part II- Protein Synthesis

- The making of proteins can be broken down into two steps:
    1. **TRANSCRIPTION**
    2. **TRANSLATION**



1.2 Protein Synthesis

Act One: **TRANSCRIPTION** (*Trans* = across, *cription* = to write)

- The coded message of a gene on DNA has **INSTRUCTIONS** on how to make **particular PROTEINS** that our bodies need.

- The instructions from a gene are copied from DNA to **MESSANGER RIBONUCLEIC ACID (MRNA)** in the nucleus.

DNA	mRNA
A	U
T	A
C	G
G	C

1.2 Protein Synthesis

- RNA has slightly different bases: A, G, C and **URACIL** instead of thymine
- Once complete, the mRNA moves through the **NUCLEAR PORES** and into the cytoplasm where the proteins are made.
- The process of making mRNA is called **TRANSCRIPTION**

1.2 Protein Synthesis

Watch the General Process

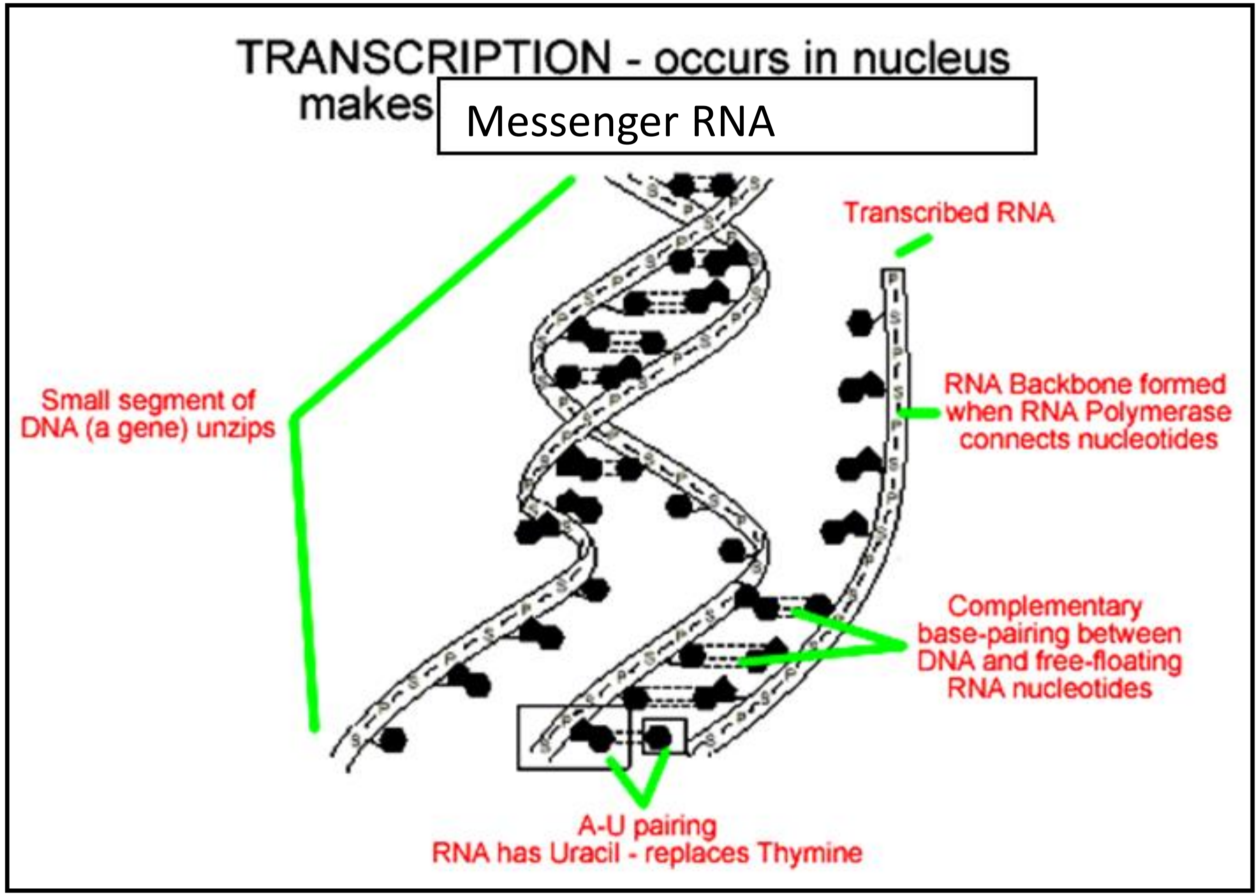
- **Step 1**: **UNZIPPING** of the DNA from a double strand into two single strands

- **Step 2**: **RIBOSOMAL BASES** pair up with the bases on one of the DNA strands to form mRNA

- **Step 3**: **ENZYMES** help form the RNA **BACK BONE** and checks for **ERRORS**

- **Step 4**: The **MRNA** is released, leaves the nucleus, & the **DNA ZIPS BACK UP**

1.2 Protein Synthesis



1.2 Protein Synthesis

Act Two: **TRANSLATION**

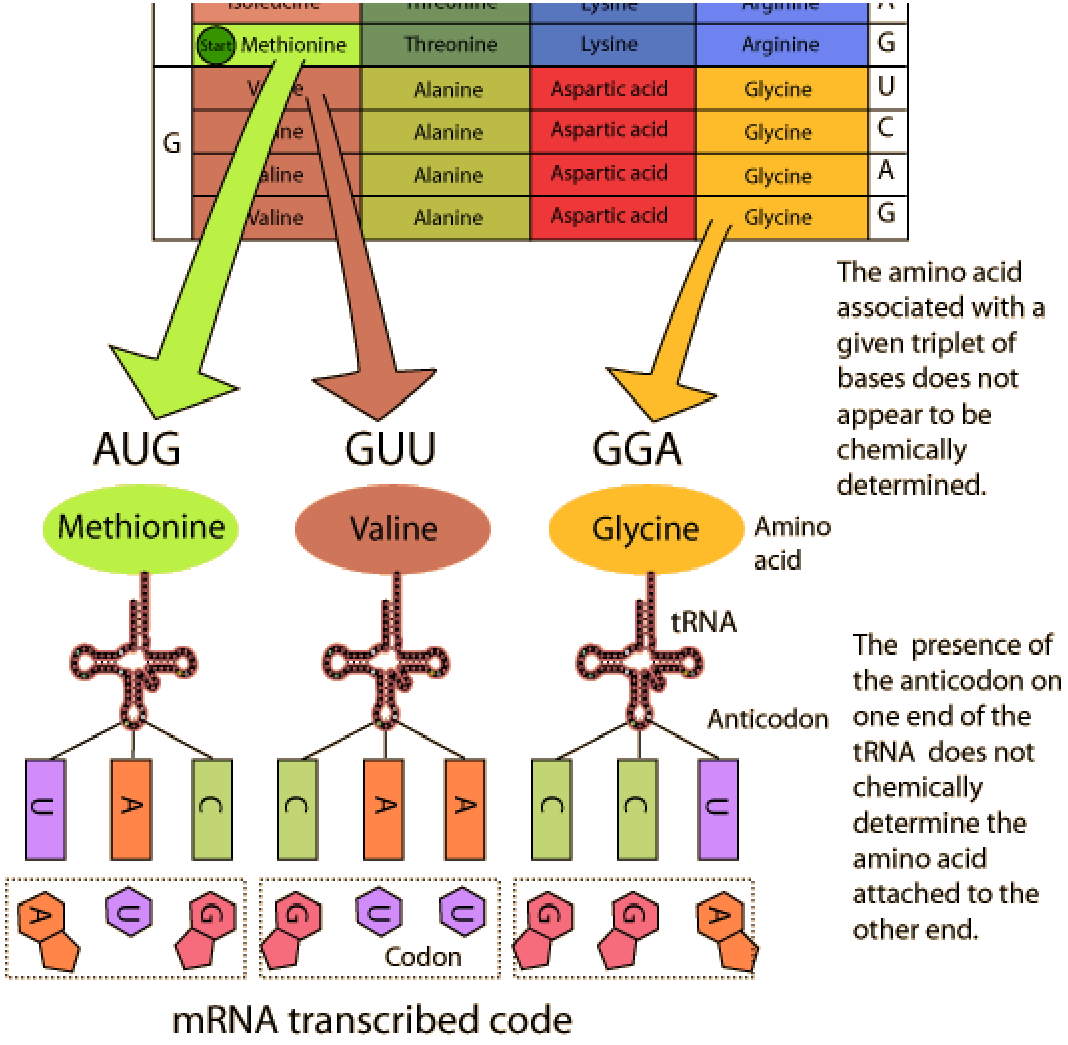
- The mRNA moves into the cytoplasm and pairs up with a **RIBOSOME**. It is here that the mRNA will be "read" to make a protein.

- The mRNA code is made up of groups of **TRIPLET_CODES** known as **CODONS.** Each codon codes for a specific **AMINO ACID**.

Eg. AGC = Serine   Eg. UGC = Cysteine

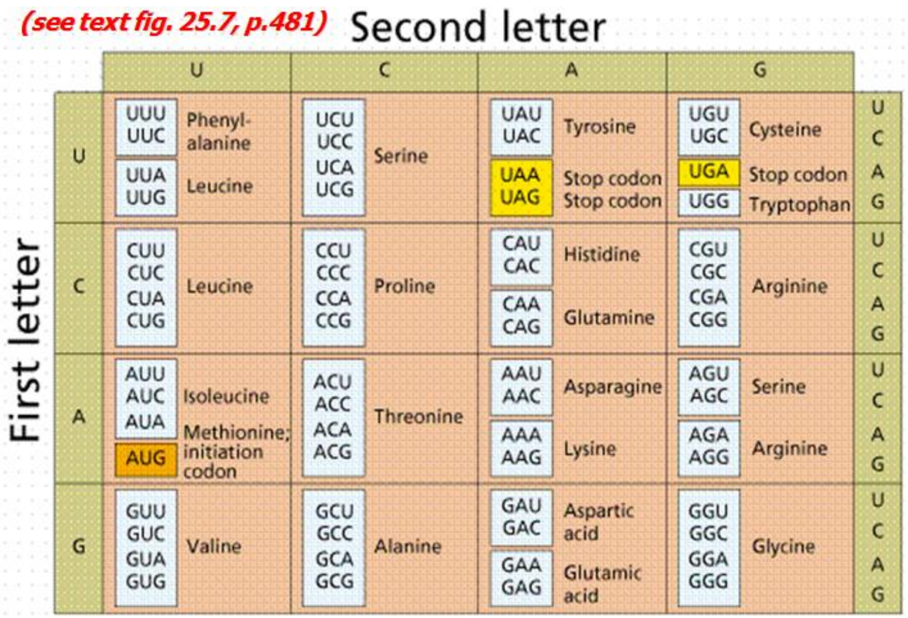
1.2 Protein Synthesis

- It takes **3_** bases to make one codon. Why? Because there are about **20** amino acids and we only have 3 **BASES** in the alphabet.
  - With a _SINGLE_ nucleotide, there are only **4** possible codes (4^1).
  - For _DOUBLE_ nucleotides, there are only **16** possible codes (4^2).
  - However, for _TRIPLET_ nucleotides there are **64** possible codes (4^3)



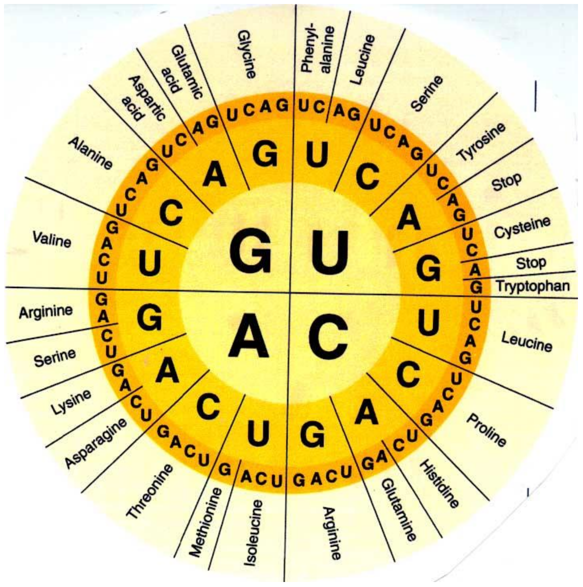
1.2 Protein Synthesis

- The amino acid for each codon is given in the tables below:



(see text fig. 25.7, p.481)

First letter	Second letter: U		Second letter: C		Second letter: A		Second letter: G		Third letter
U	UUU UUC	Phenyl-alanine	UCU UCC UCA UCG	Serine	UAU UAC	Tyrosine	UGU UGC	Cysteine	U C
	UUA UUG	Leucine			UAA UAG	Stop codon Stop codon	UGA	Stop codon	A
							UGG	Tryptophan	G
C	CUU CUC CUA CUG	Leucine	CCU CCC CCA CCG	Proline	CAU CAC	Histidine	CGU CGC CGA CGG	Arginine	U C
					CAA CAG	Glutamine			A G
A	AUU AUC AUA	Isoleucine	ACU ACC ACA ACG	Threonine	AAU AAC	Asparagine	AGU AGC	Serine	U C
	AUG	Methionine; initiation codon			AAA AAG	Lysine	AGA AGG	Arginine	A G
G	GUU GUC GUA GUG	Valine	GCU GCC GCA GCG	Alanine	GAU GAC	Aspartic acid	GGU GGC GGA GGG	Glycine	U C
					GAA GAG	Glutamic acid			A G

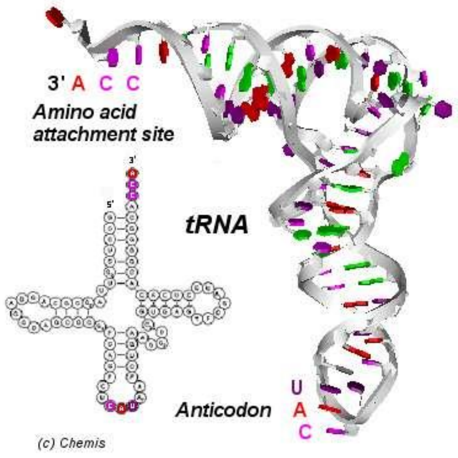


- Now you try. What is the amino acid for each of the following codons?

CAU : _______________ AUG : _________________ CAU : ______________

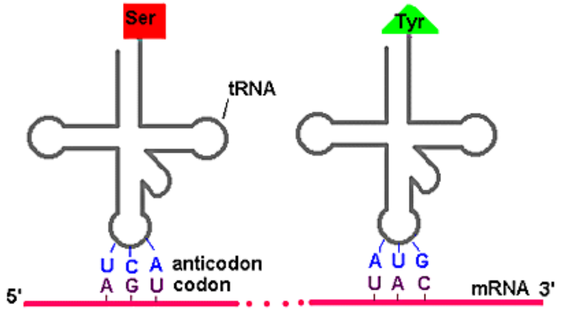
UGA : _____________ GCC : ___________________ AAA : ______________

<u>1.2 Protein Synthesis</u>



- During translation, the written code (codons) on mRNA is '**<u>TRANSLATED</u>**' into a specific amino acid sequence by **<u>TRANSFER RIBONUCLUEIC ACID</u>** (tRNA) in the cytoplasm.

- A tRNA molecule is a small piece of RNA that has **an <u>AMINO ACID</u>** attached to it.

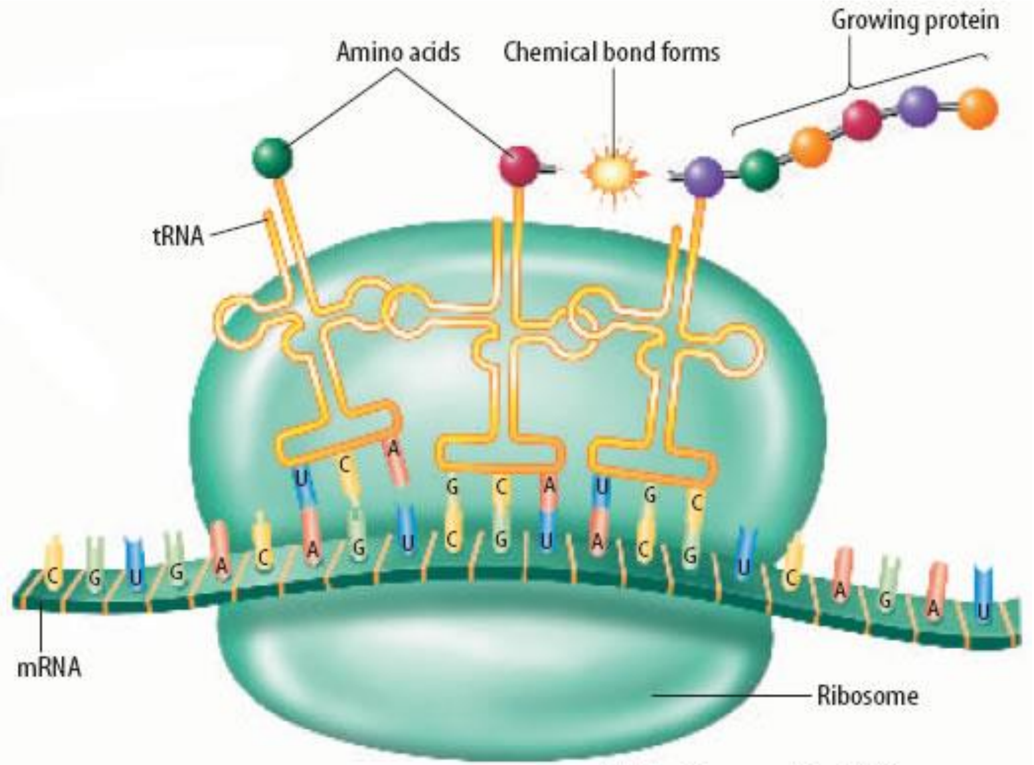


- The tRNA also has a special sequence of 3 bases known an **<u>ANTICODON</u>**.

- There is at least one type of tRNA for each of the 20 amino acids.

1.2 Protein Synthesis

- As the correct **AMINO ACIDS** are brought to the ribosome by the **tRNAs**, they are joined together to form the **PROTEIN CHAIN** that the original DNA coded for.
- Eventually, the finished protein chain moves to the **GOLGI BODY**, where it is packaged and released to do its "job".



At the ribosome, the RNA's message is translated into a specific protein.

SUMMARY OF PROTEIN SYNTHESIS:

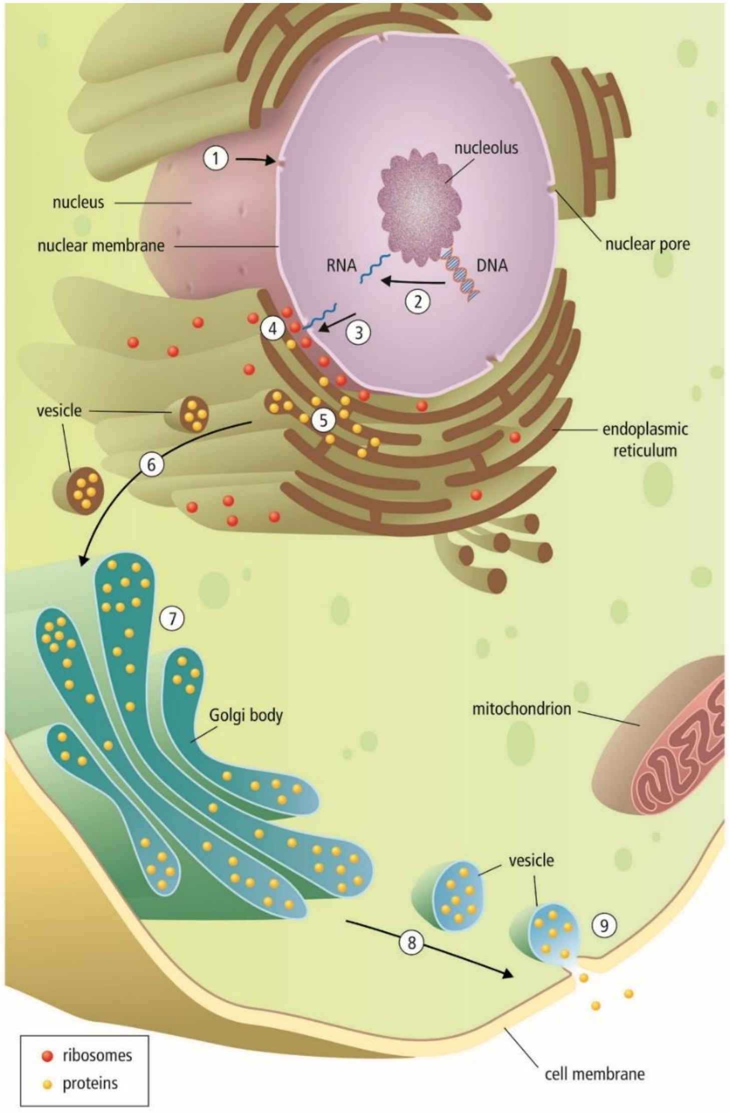


Figure 4.11 The production of a protein in an animal cell

Describe what happens at each step:

1. ________________________________
____
____

2. ________________________________
____
____

3. ________________________________
____
____

4. ________________________________
____
____

5. ________________________________
____
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6. ________________________________
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7. ________________________________
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8. ________________________________
____
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9. ________________________________
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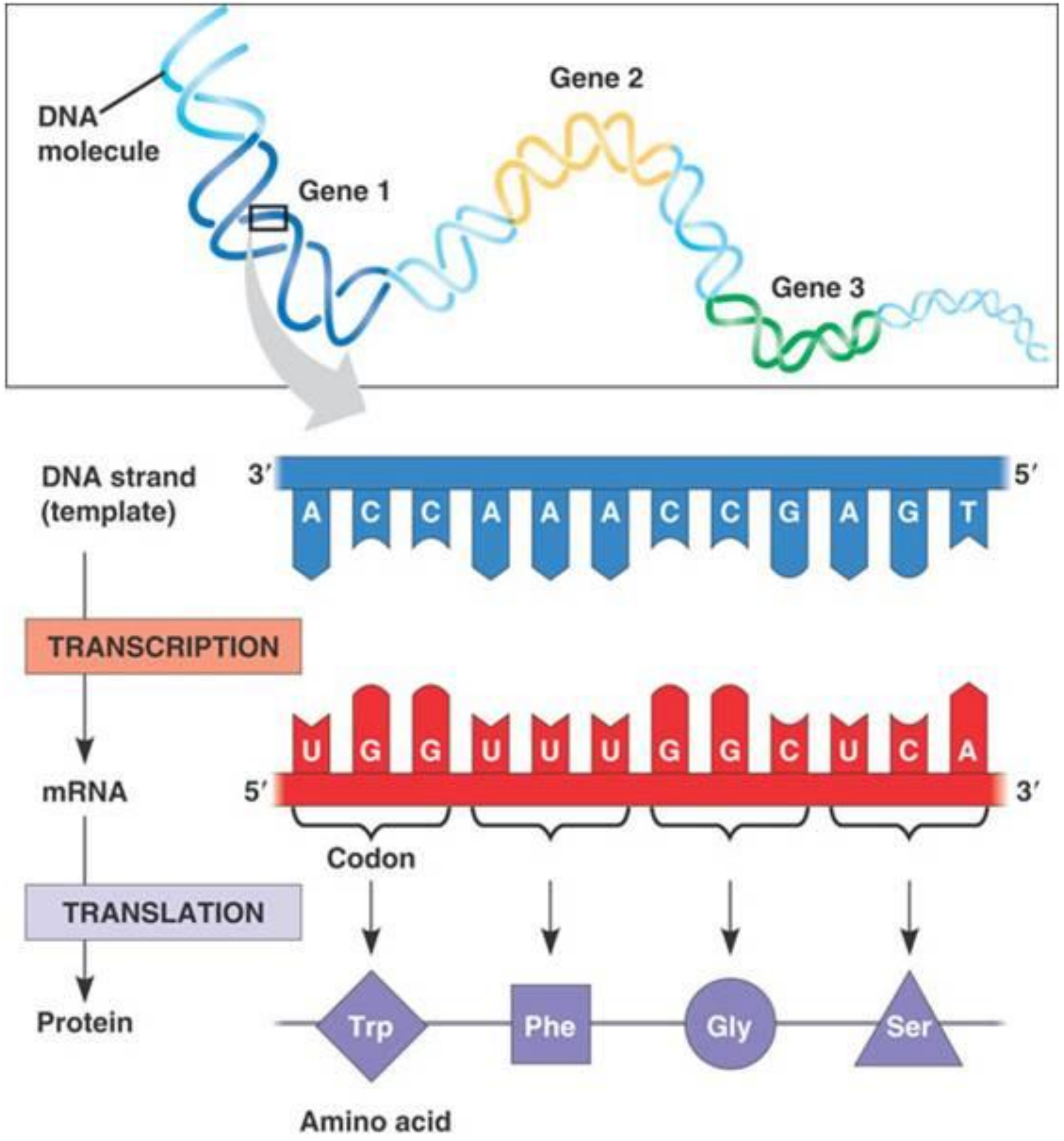


Worksheet:

DNA AND PROTEIN SYNTHESIS REVIEW

Section 4.2: Mutations

- Real or Fake?




2.1 Mutations

A BAD NIGHT AT THE THEATRE

Question: What if something goes wrong during ***translation***?

Answer: **IF TRANSLATION IS ALTERED, THE PROTIEN PRODUCT WILL BE DIFFERENT**

PROTEIN FUNCITON DEPENDS ON SHAPE! IF THE SHAPE CHANGES, IT'S FUNCTION CHANGES TOO!

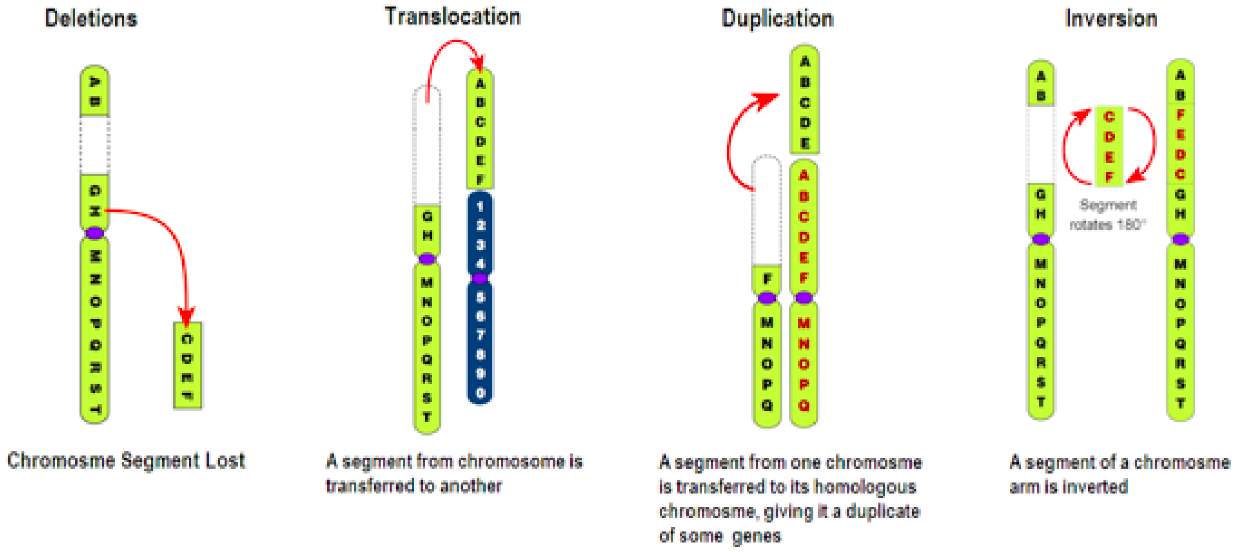
- Mutations can **CHANGE** in the base sequence of DNA
- When the **BASES** ('letters') **change**, the **INSTRUCTIONS** used to make the protein also change.
- Different **AMINO ACIDS** will be used, resulting in a different protein. This may alter protein shape, and also change or "break" protein function.

<u>2.1 Mutations</u>

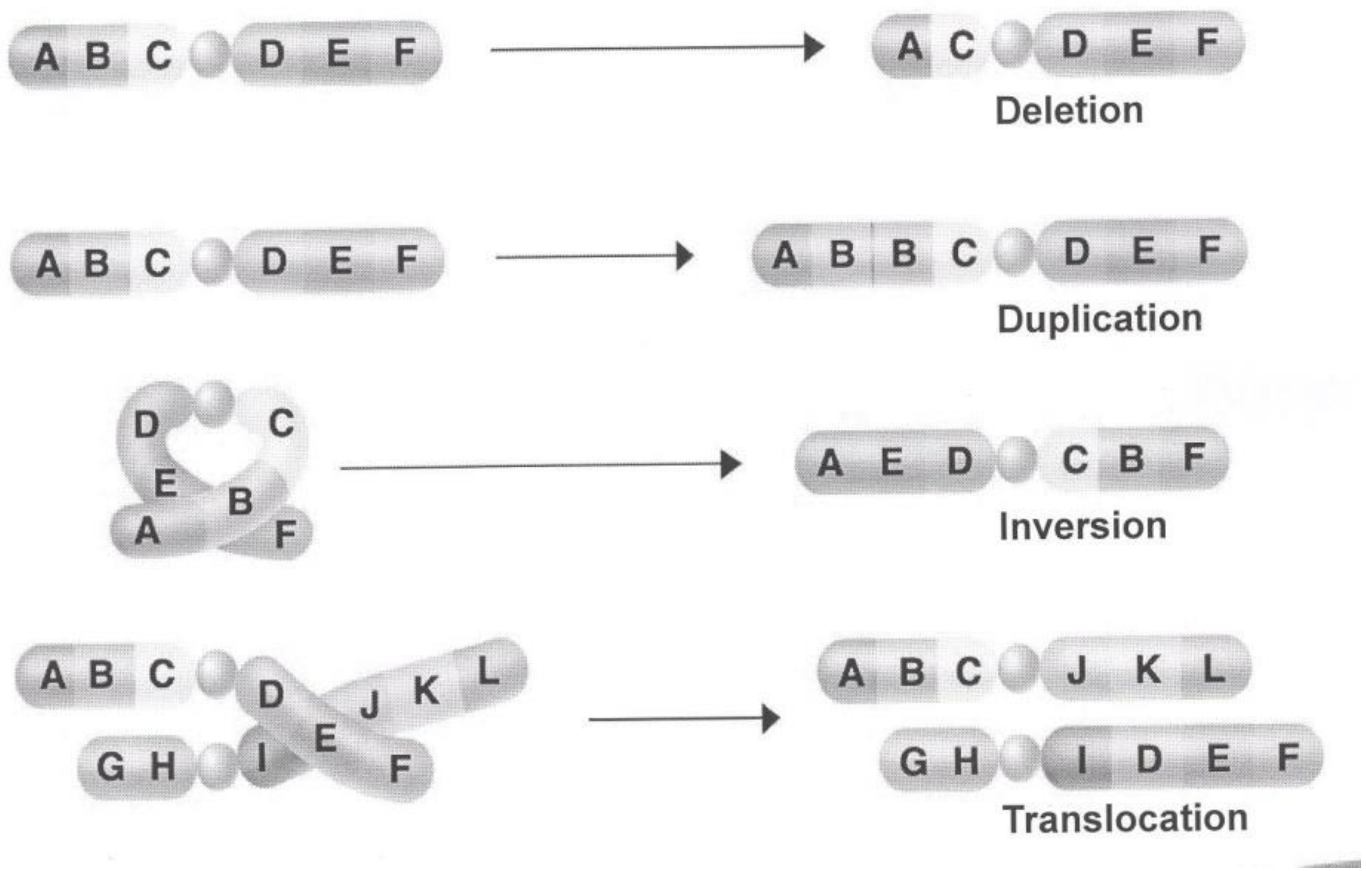
There are 2 types of MUTATION:

1. <u>CHROMOSOMAL</u> mutations: a mutation of **all or part** of a chromosome.

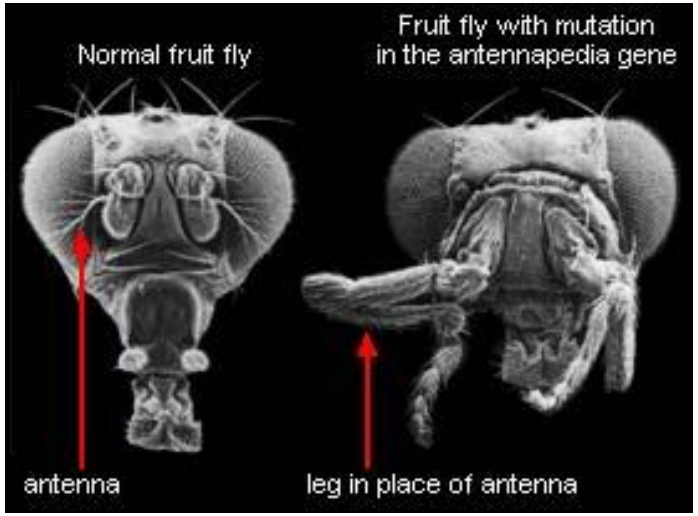
- This usually involves **<u>LARGE SEGMENTS OF DNA</u>**, and therefore, **<u>CAN HAVE GREATER IMPACT THAN SMALLER GENE MUTATIONS</u>** .
- There are four types of chromosomal mutations that involve part of the chromosome:



Chromosomal Mutations…



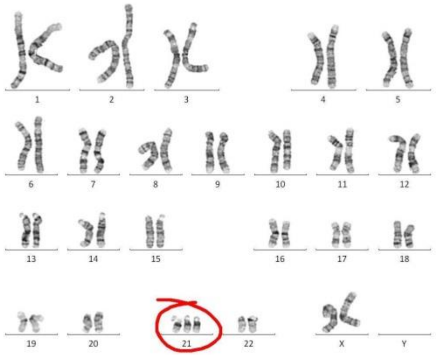
2.1 Mutations



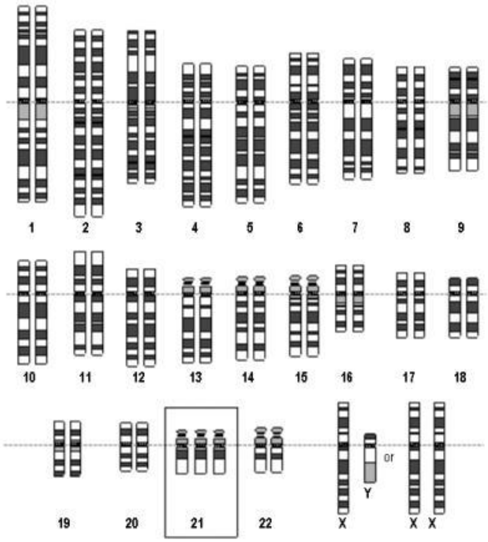
- E.g. Fruit fly with legs in place of antennae. This mutation involves moving a controller gene telling the fruit fly where to put the legs

- E.g. Down's syndrome. This is a genetic disorder where there is an extra chromosome #21. The extra chromosome resulted from improper cell division.

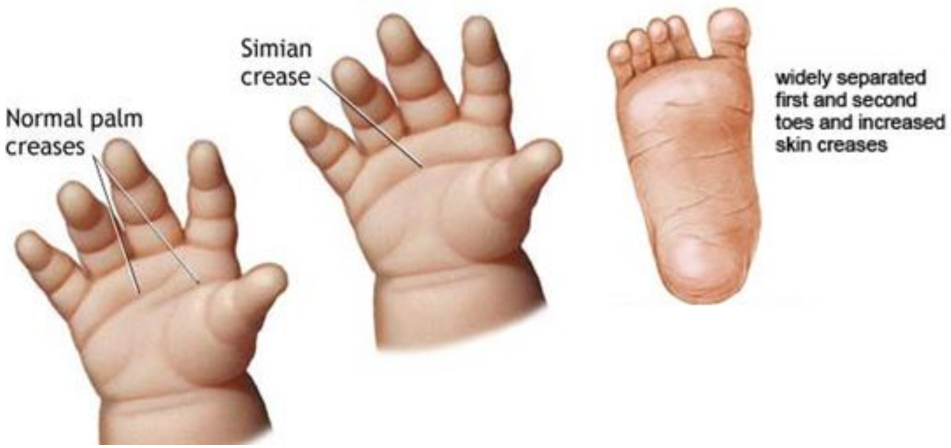
Down syndrome karyotype

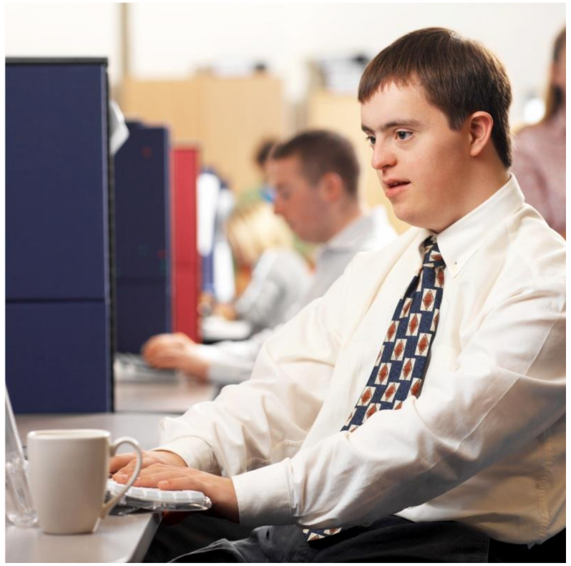


- Examples:
  Down's syndrome – (Trisomy 21) **47** chromosomes, extra chromosome at pair **#21**



flattened nose and face, upward slanting eyes,

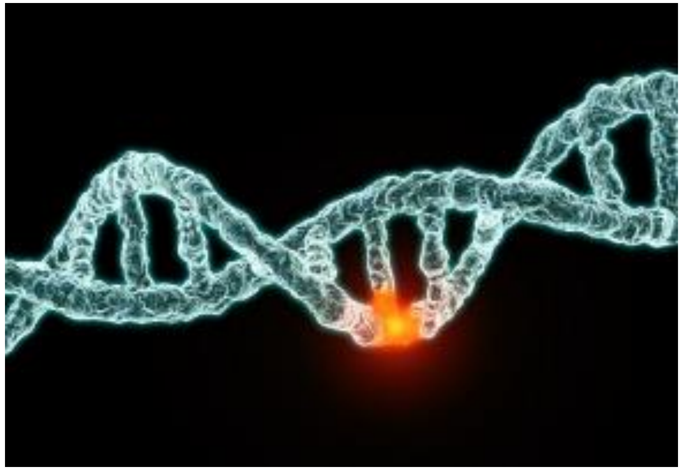




2.1 Mutations

2. **GENE mutations**: a mutation that occurs within a gene at some **point** along a chromosome. This mutation is only a change of one or a few 'letters' (bases).

- It usually only affects **ONE GENE,** and therefore, **CAN HAVE LESS IMPACT THAN LARGER CHROMOSOMAL MUTATIONS**.



2.1 Mutations

- Recall the sequence of protein synthesis:

DNA	→	**RNA**	→	**protein**
ACTGCT….		UGACGA….		leu- ade-cyt…
(in nucleus)		(to cytoplasm)		(at ribosome)

- If the sequence of DNA changes by mutation, then the RNA sequence copied from the DNA will be different, and this will code for a different amino acid, which results in a different protein. This is called a **GENE MUTATION**

- E.g. the coat of the Spirit Bear is **WHITE** because the gene that codes for **FUR COLOUR** (dark brown) is mutated and no longer works

2.1 Mutations

- there are three main ways that gene mutations can occur:
- DELETION AND ADDITION MOST HARMFUL → FRAMESHIFT

Substitution	**e.g. replace "C" with "A"**	**ACTGCA → AATGCA**
Deletion	**e.g. lose a base all together**	**ACTGCA → ACGCA**
Addition	**e.g. add another "A"**	**ACTGCA → ACTAGCA**

2.1 Mutations

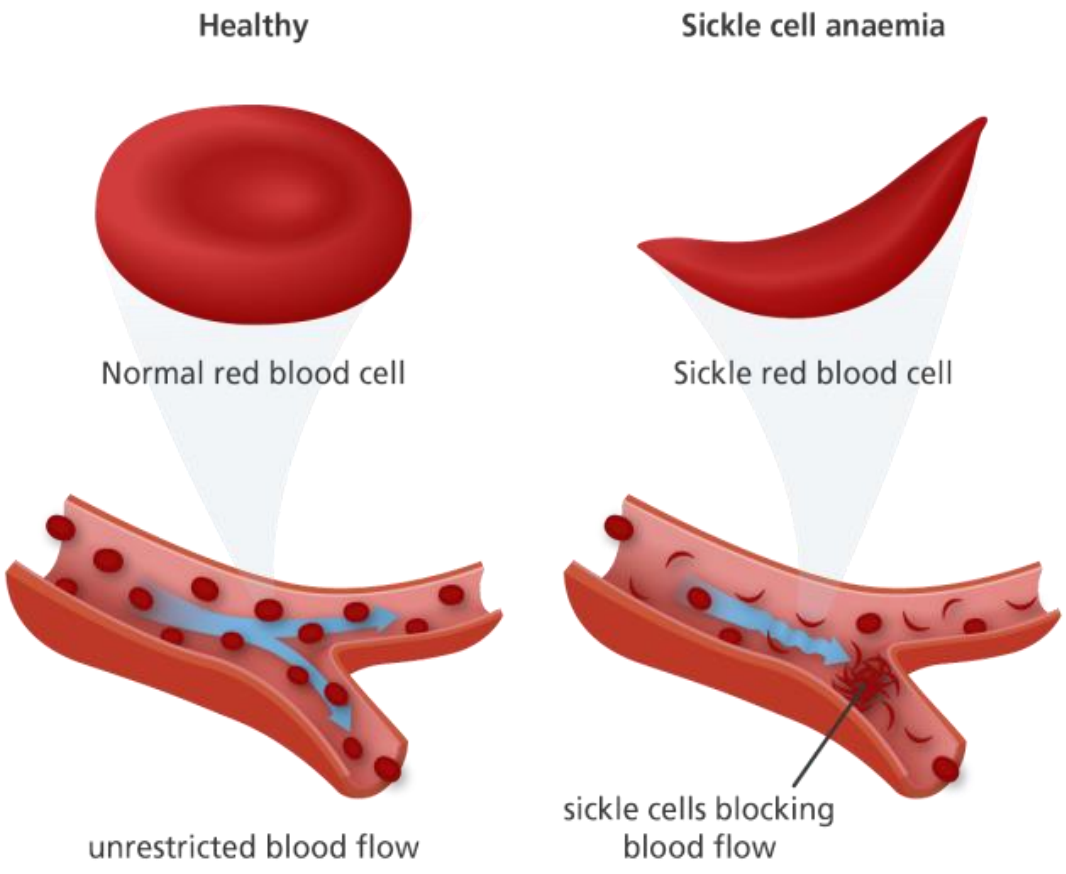
I. Types of Gene Mutations

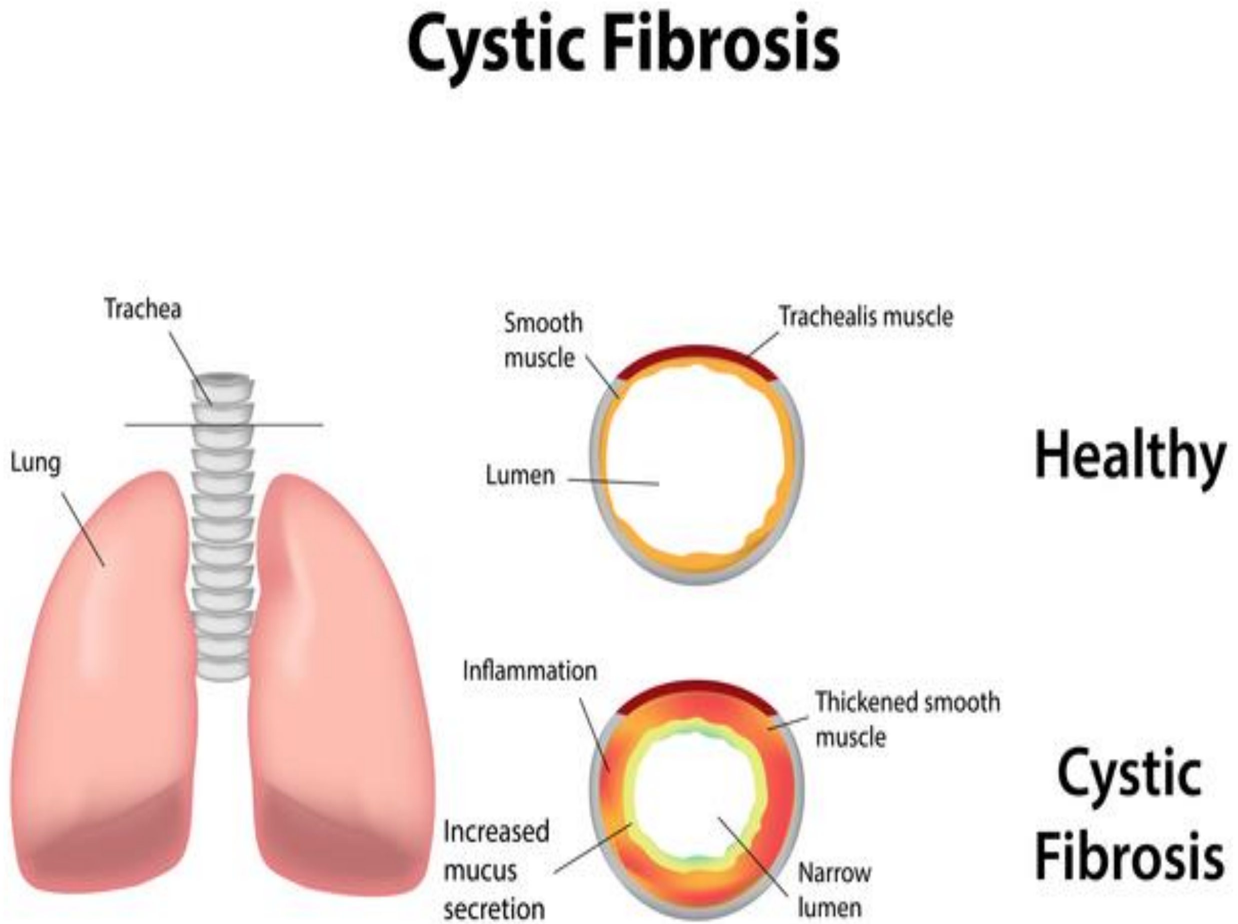
- mutations are often a bad thing because the protein does not work and your body needs it to function. These are known as **NEGATIVE MUTATIONS** and they **DECREASE** survival rates.

- e.g. mutated gene → **SICKLE-CELL ANEMIA** (misshapen red blood cells that don't carry O_2).

- e.g. more than 1300 different mutations in one gene → **CYSTIC FIBROSIS** (build up of mucus in the lungs)

Sickle Cell Anemia and Cystic Fibrosis

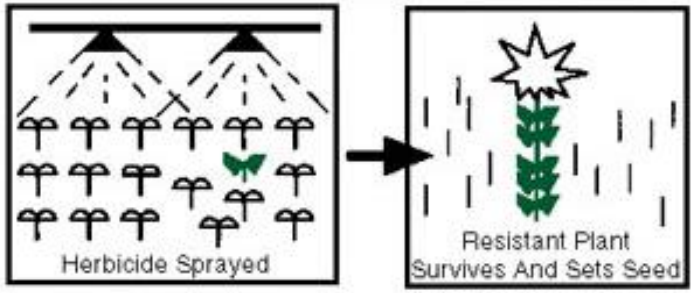




2.1 Mutations

- sometimes mutations can be a good thing (e.g. the mutated protein works better than the "normal" protein or has a new beneficial function). These are known as **POSITIVE MUTATIONS** and they **INCREASE** survival rate.

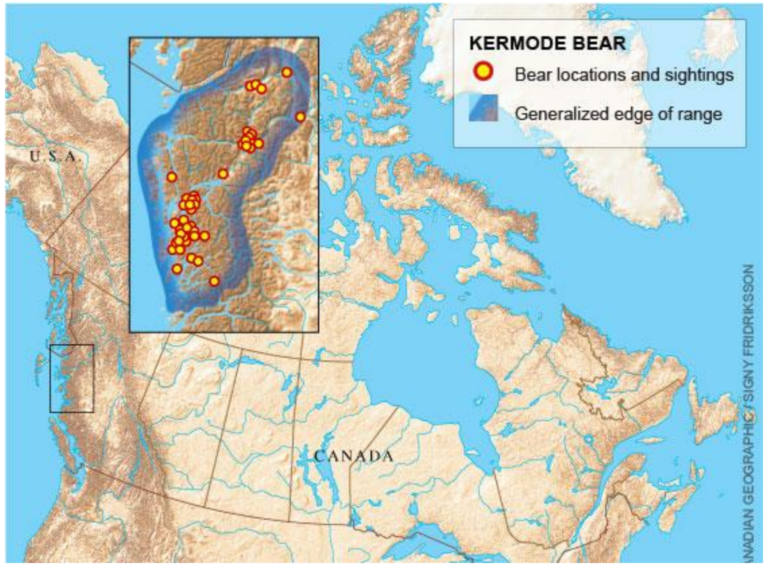
- e.g. mutated gene → proteins that make them **RESISTANT TO HIV INFECTION**. These people cannot get AIDS.
- e.g. some plants have mutated genes that codes for proteins that make them **RESISTANT TO CHEMICAL HERBICIDES**.



2.1 Mutations

- sometimes mutations have no effect because they occur in a part of the DNA that is not used for protein synthesis or the mutated protein is not required for survival. These are known as **NEUTRAL MUTATIONS** and have they have **NO EFFECT** on the organism.

- e.g. mutation in the gene that codes for fur colour (Spirit Bear). Although white, the survival rate of the organism is not changed.



2.1 Mutations

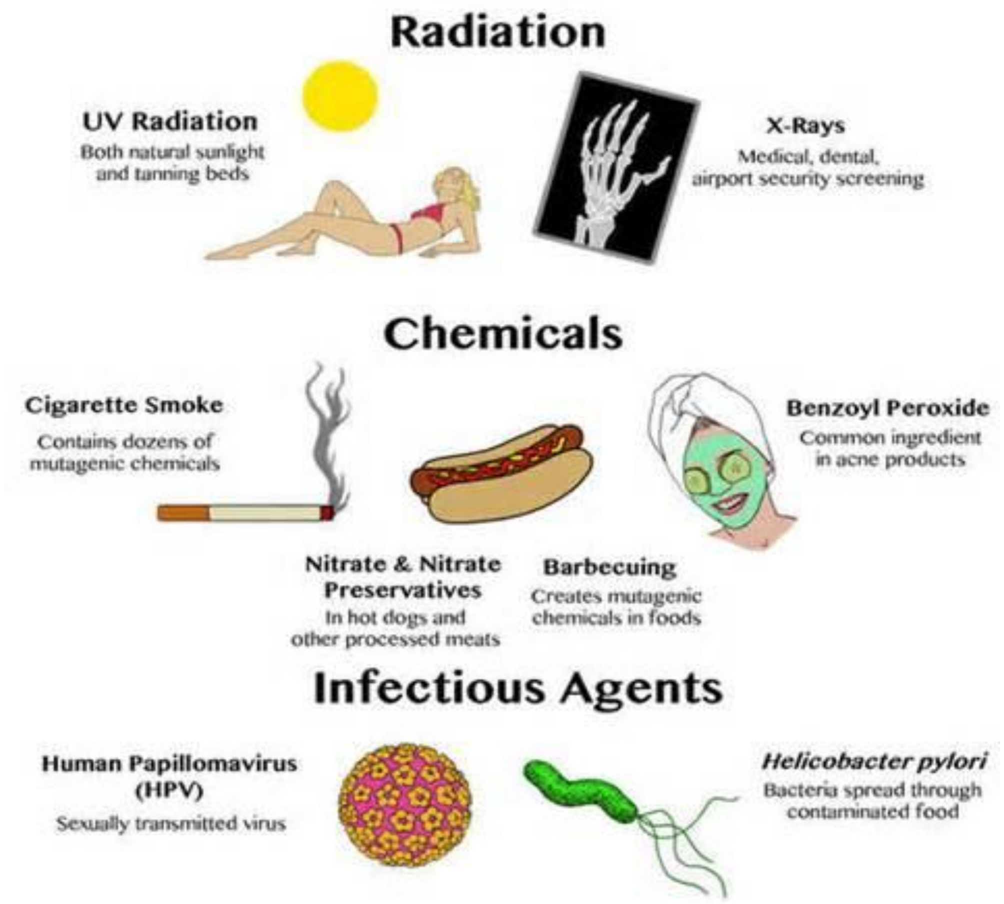
II. Causes of Mutations

- Mutations can occur naturally during DNA replication and cell division. The human body has systems in place that will often fix errors in DNA replication, or destroy cells that are working abnormally due to mutations.

- There are also factors in the environment that may increase chances for mutations to occur.

2.1 Mutations

- **MUTAGENS**= substances or factors (e.g. virus, cigarette smoke, X-rays) that can cause mutations in DNA



A CARCINOGEN IS A CANCER CAUSING MUTAGEN