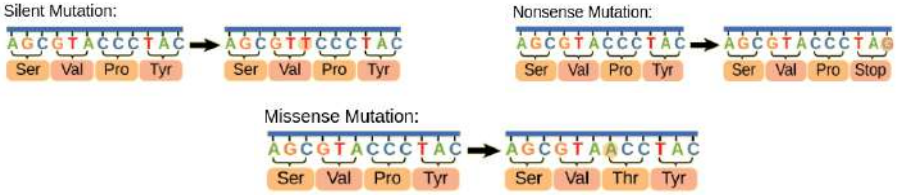
Mutations

What is a mutation?

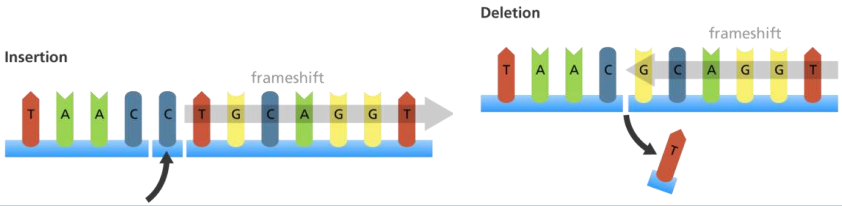
- A mutation is a change in the **DNA** sequence that can affect how a **gene** works or is shown.

Types of Mutations:

- **Point Mutations:**
  - **Missense Mutation**: This causes a different **amino acid** to be incorporated into the protein, potentially changing its **function**.
  - **Nonsense Mutation**: This changes an amino acid codon into a **stop** codon, leading to a **shortened** protein that may not work properly.
  - **Silent Mutation**: This changes a nucleotide but does not change the **amino acid** due to the redundancy of the **genetic code**, usually having no effect on the protein.



- **Frameshift Mutations**: These occur when **nucleotides** are added or deleted from the DNA sequence, changing the **reading frame** of the genetic code.
  - **Insertion**: Adding one or more nucleotides shifts the reading frame and changes the **amino acid** sequence.
  - **Deletion**: Removing one or more nucleotides shifts the reading frame and also changes the **amino acid** sequence.

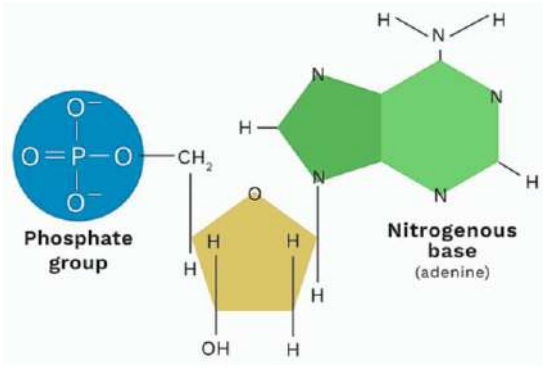


DNA, Protein Synthesis, & Gene Expression

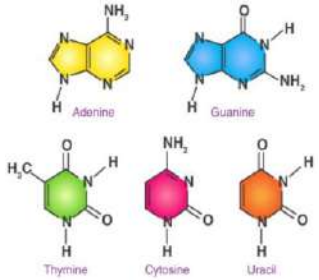
Characteristics of DNA

A. Structure

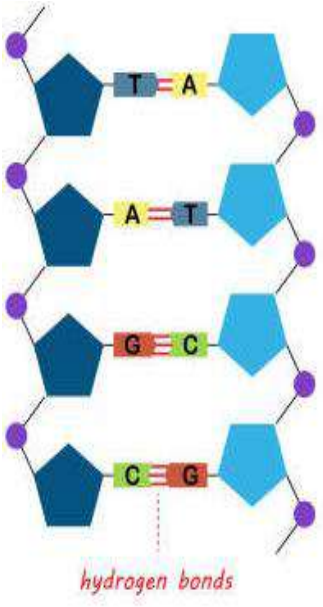
- DNA is a **double helix** structure made of **two** strands of building blocks called **nucleotides**.
- Each building block, or nucleotide, has three parts: a **phosphate** group, a **sugar** (called deoxyribose), and one of four **bases**: **adenine**, **thymine**, **cytosine**, and **guanine**.



Deoxy/ribose sugar

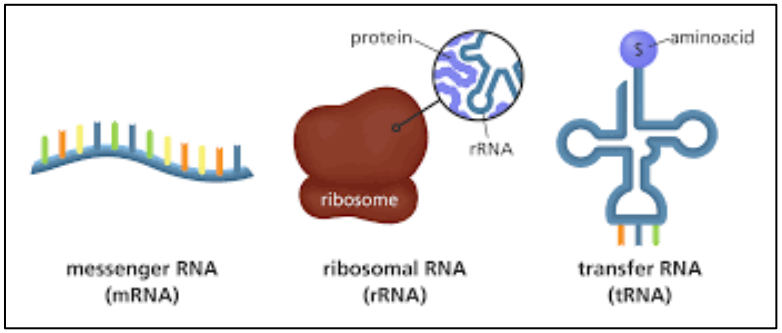


- The base pairing rules tell us that **adenine** pairs with **thymine**, and **cytosine** pairs with **guanine**.



B. Function

- The main job of DNA is to store **genetic** information that tells living things how to **develop** and function.
- DNA gives instructions for making **proteins**, which are important for **functioning** in the body.

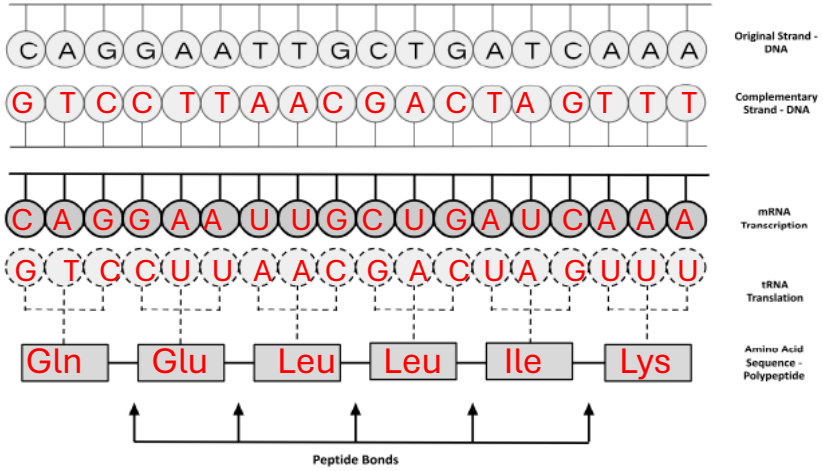


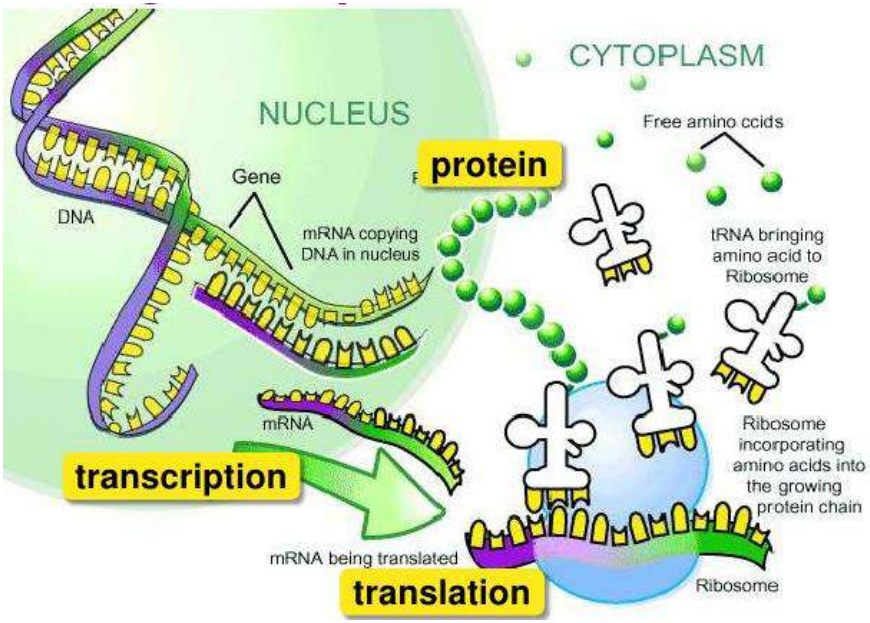
D. Codons and Anticodons:

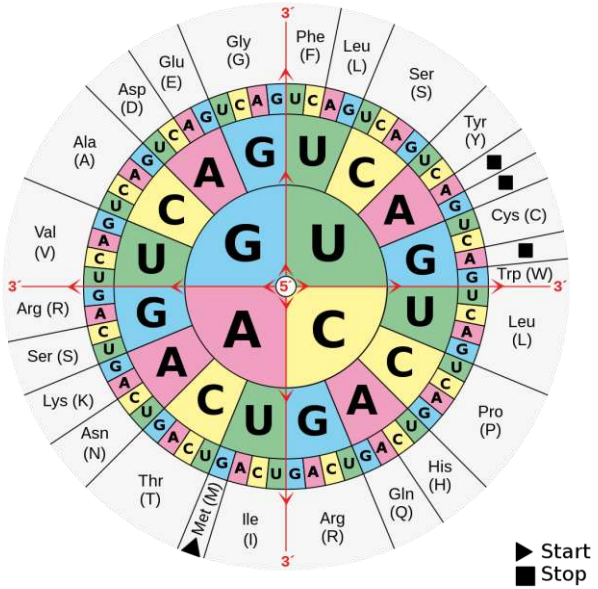
- **Codons**: These are three-letter codes on mRNA that tell what **amino acid** to make (e.g., **AUG** for methionine).
- **Anticodons**: These are three-letter codes on tRNA that match the mRNA codons, making sure the right **amino acid** is added.

E. Steps of Translation:

- **Initiation**: The ribosome starts at the **start** codon (**AUG**) on the mRNA.
- **Elongation**: tRNA brings the correct **amino acids** to the ribosome, matching **codons** to the mRNA codons. The ribosome connects the **amino acids** to form a chain.
- **Termination**: When the ribosome reaches a special **stop** codon (**UAA**, **UGA**, or **UAG**), the translation ends, and the **protein** is released.







DNA Replication

A. Purpose and When

- DNA replication makes sure each new cell gets a **exact** copy of the DNA during S-phase of **cell** division.

B. Steps of DNA Replication

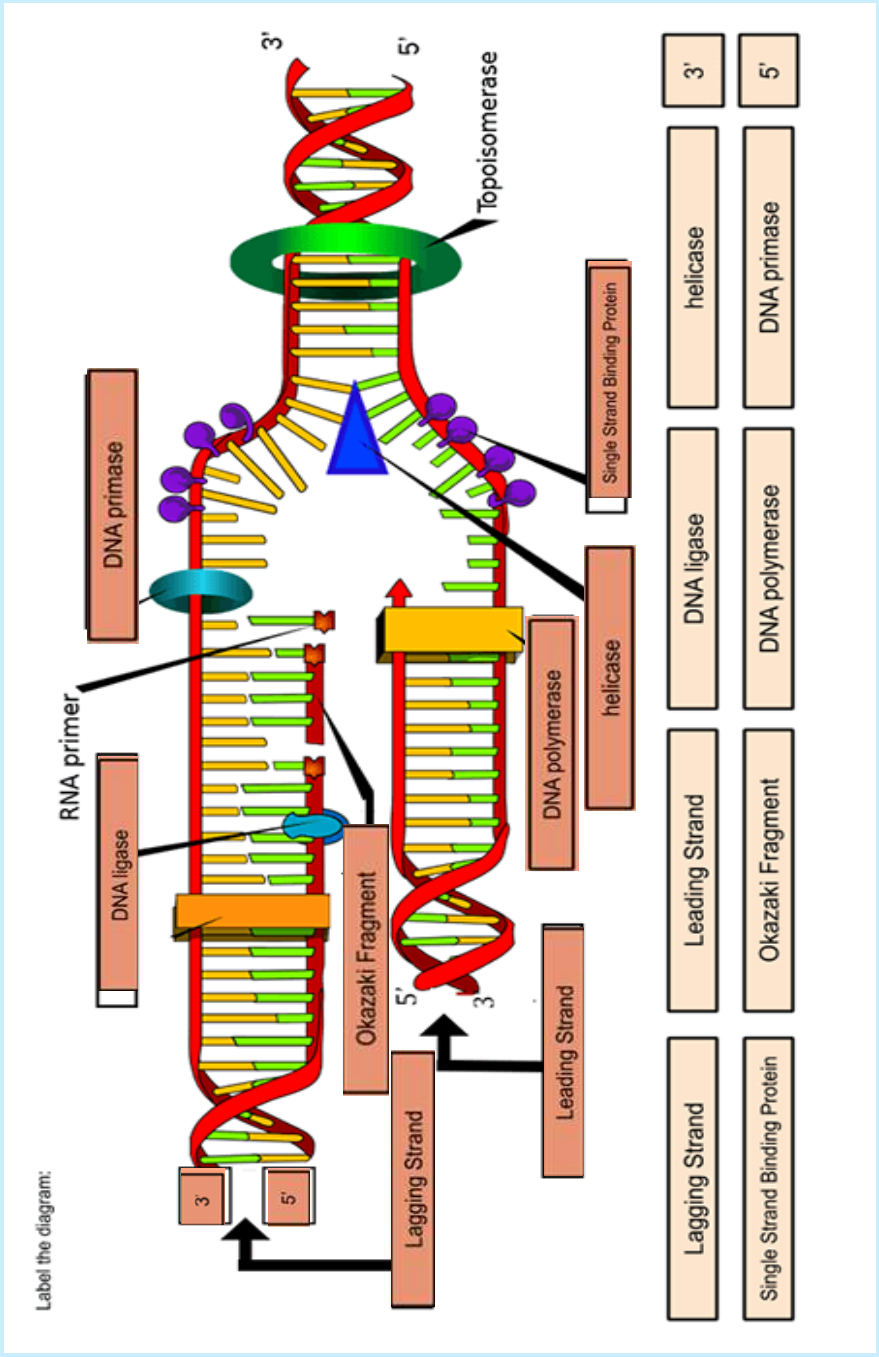
- Unwinding the DNA:
  - An enzyme called **DNA helicase** unwinds the double helix and creates a place called the **replication fork** where copying happens.
- Complementary Base Pairing:
  - The enzyme **DNA polymerase** adds new nucleotides to match the template strand.
- Leading and Lagging Strands:
  - The **leading** strand is made in one long piece in the direction of the **replication** fork.
  - The **lagging** strand is made in small pieces called **Okazaki** fragments, which are connected by another enzyme called **ligase**.
- Formation of Two New DNA Molecules:
  - Each new DNA molecule has one **original** strand and one **new** strand. This is called **semiconservative** replication.

D. Directionality:

- DNA polymerase can only add nucleotides in the **5' to 3'** direction, so the leading strand is continuous while the lagging strand has **discontinuous** pieces.

E. Practice DNA Sequence:

5' - **A T C G G C T A** - 3'



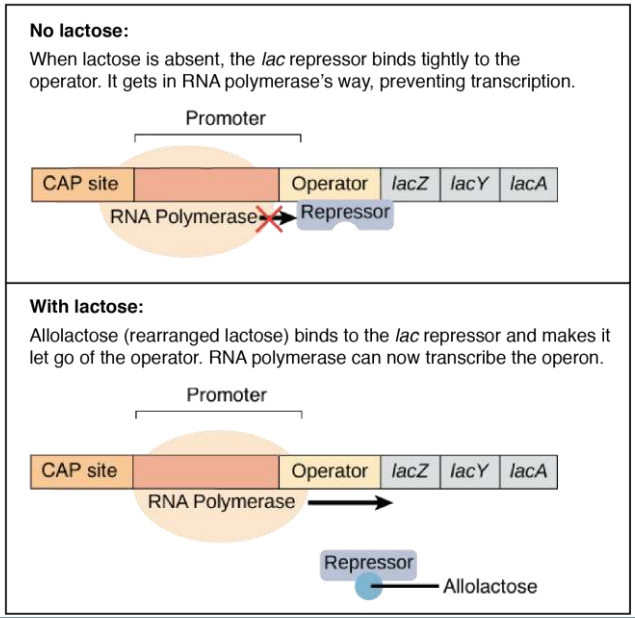
Gene Regulation

A. What is Gene Regulation?

- Gene regulation is how cells control when and how much **proteins** are made from a **gene**.

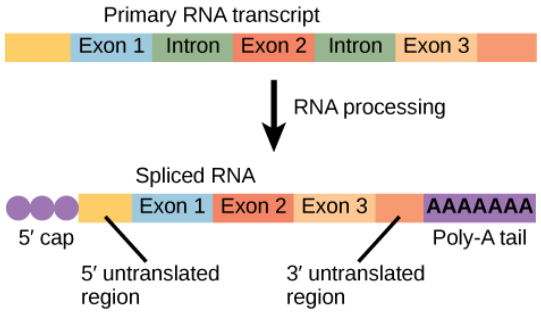
B. Steps of Gene Regulation:

- **Transcriptional control**: special proteins bind to DNA to decide if genes are **on** or **off**.
- **RNA processing control**: this is about editing the **mRNA** to remove extra parts and keep the important ones.
- **Translational control**: this controls how much **protein** is made from **mRNA**.
- **Post-translational control**: this includes changes made to **proteins** after they are made, like adding special **chemical groups**.



mRNA Processing:

- **Splicing**: Non-coding parts (**introns**) are removed, and coding parts (**exons**) are joined together.
- **5' cap and poly-A tail**: A **5' cap** and a **poly-A tail** are added to protect the mRNA and help it leave the nucleus.



Translation

A. Purpose

- Translation changes the **mRNA** sequence into a **protein** by linking **amino acids** together in the order from the mRNA **codons**.

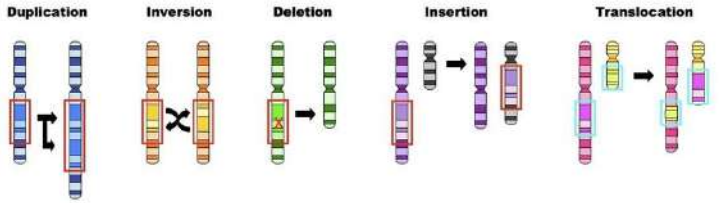
B. Location:

- Translation happens in the **ribosome** at the **cytoplasm**.

C. Types of RNA involved:

- **mRNA (messenger RNA)**: carries instructions from the **nucleus** to the **ribosome**.
- **tRNA (transfer RNA)**: brings the correct **amino acids** to the ribosome, matching them to the mRNA **codons** using **anticodons**.
- **rRNA (ribosomal RNA)**: makes up part of the ribosome and helps in joining the **amino acids** to make **proteins**.

- **Chromosomal mutations**: These involve changes in the structure or number of **chromosomes**.
  - **Duplication**: A segment of DNA is copied, leading to extra genetic material.
  - **Deletion**: A segment of DNA is lost, resulting in missing genetic information.
  - **Inversion**: A segment of DNA is flipped to the opposite direction.
  - **Translocation**: A segment of DNA is moved to a different location, either within the same chromosome or to a different one.



C. Causes of Mutations:

- **Spontaneous mutations**: These happen naturally due to errors in DNA **replication** or repair.
- **Induced mutations**: These are caused by external factors such as **radiation**, chemicals, or **viruses**.

D. Effects of Mutations:

- **Beneficial mutations**: These can give an advantage in survival or reproduction, such as increased **resistance** to diseases.
- **Harmful mutations**: These can lead to genetic disorders or increase the risk of diseases.
- **Neutral mutations**: These do not have any significant effect on the organism.

Epigenetics

A. What is Epigenetics?

- Epigenetics is the study of how environmental factors can change gene expression without changing the DNA sequence itself.
- These changes can sometimes be passed down to future generations.

B. Examples of Epigenetic Factors

- **Diet and Nutrition:**
  A mother's diet during pregnancy can alter gene expression in her baby, affecting traits like metabolism or risk of obesity.
- **Stress and Mental Health:**
  Chronic stress can "turn off" genes that help regulate mood, potentially leading to mental health issues like anxiety or depression.
- **Exercise:**
  Regular exercise can activate genes that improve muscle function and overall health.
- **Smoking:**
  Smoking can cause epigenetic changes that lead to health problems like lung cancer. These changes can remain even after someone quits smoking.

Transcription

A. Purpose & Location

- The purpose of transcription is to copy a gene's **DNA** sequence into **messenger** RNA (mRNA), which carries information from the **nucleus** to the **ribosome**.

B. Steps of Transcription:

- **Initiation**: The enzyme RNA polymerase attaches to the **promoter** part of the gene and unwinds the **DNA**.
- **Elongation**: RNA polymerase adds RNA building blocks that match the DNA strand, following the rules that **adenine** pairs with **uracil** and **cytosine** pairs with **guanine**.
- **Termination**: Transcription ends when RNA polymerase reaches a special **termination** sequence and releases the new **RNA** strand.

DNA: 5' - T A G G C C C G A - 3'

MRNA: 3' - [ ] - 5'