



# Medical Analysis Department

## Faculty of Science



## Lec.1

# GENE STRUCTURE & ORGANIZATION

Human Genetics

Lec. I

2<sup>nd</sup>/ Grade – Spring Semester 2021-2022

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# دیکتیو

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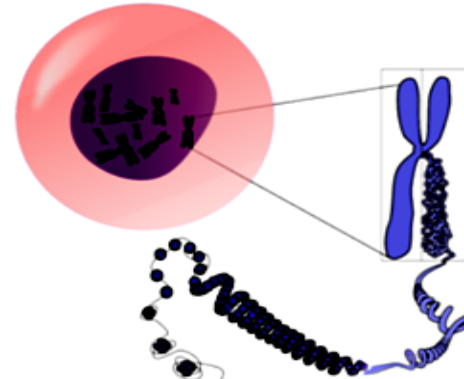
# Questions

- 1. What is the difference between Genome & Gene?**
- 2. Mention the control genes? The role of each?**
- 3. Absence of G & C in TATA box?**
- 4. Define stop codons & types?**

# What is Gene?

A segment of a DNA molecule containing the code for the amino acid sequence of a polypeptide chain and the regulatory sequences necessary for expression.

1. Your genome is located within the nucleus (purple) of your cells.



2. Sets of genes are packaged into chromosomes. Humans have 23 pairs.

5. DNA is made of paired molecules called nucleic acids (A,T,G,C).



3. Genes are comprised of DNA.

4. DNA naturally forms a helix shape.

# What are Gene types ?

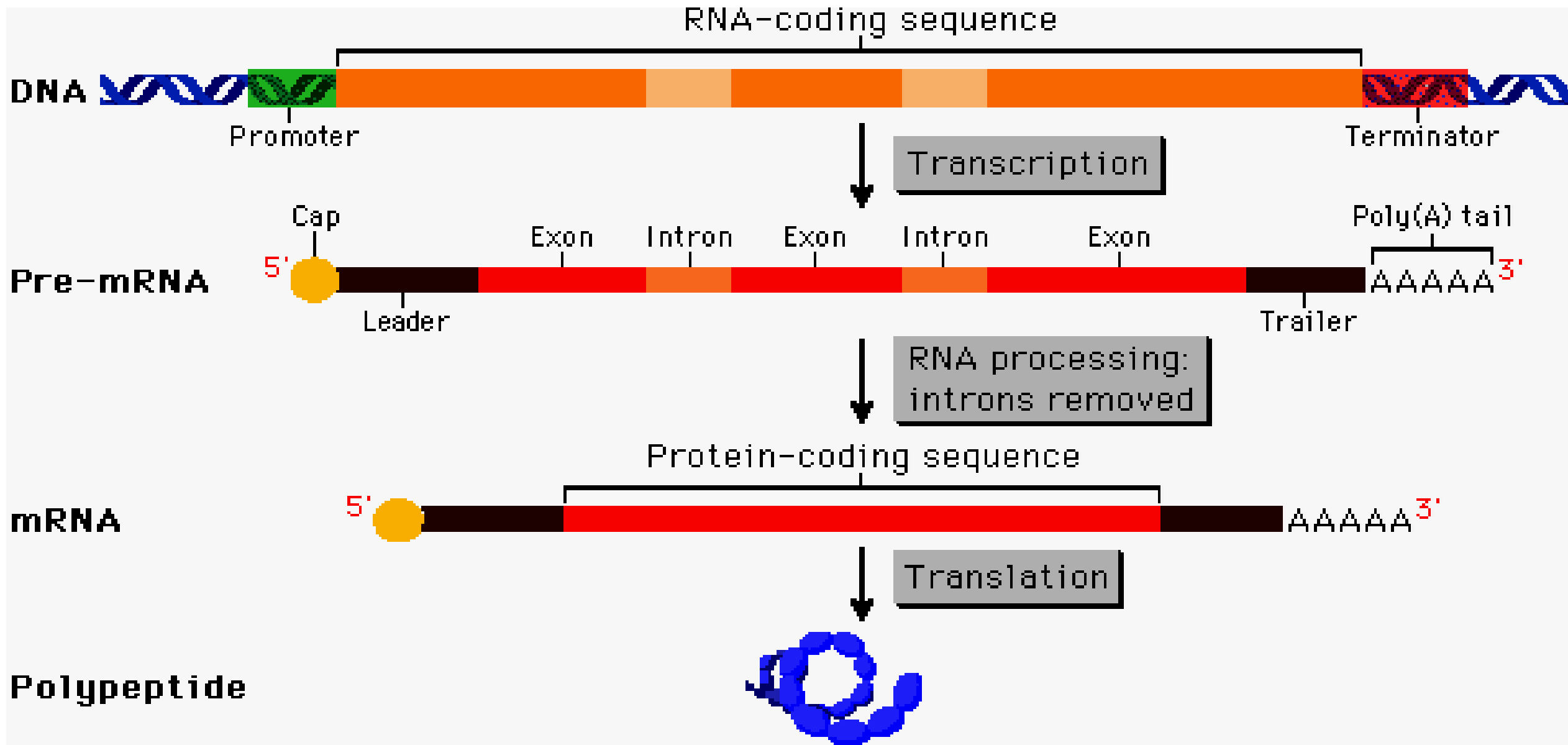
## 1. Control Genes

Genes that are ubiquitously (uniformity) expressed at stable levels in different biological situations, have been used to standardize quantitative expression

## 2. Structural Genes

Are nucleotide sequences that encode for the various protein products produced by the cell.

# What is Gene Structure?



# What are control Genes ?

## 1. Promoter

- DNA sequences that define where transcription of a gene by RNA polymerase begins.
- Located at the 5' end of the gene.
- Composed of a **core promoter** sequence and a **proximal promoter** sequence.

- **Core promoter** marks the start site for transcription **by** binding RNA polymerase and other proteins necessary for copying DNA to RNA.
- **Proximal promoter** region binds transcription factors that modify the affinity of the core promoter for RNA polymerase.

• **Promoter sequences** define the direction of transcription and indicate which DNA strand will be transcribed; this strand is known as the **sense strand**.



- Promoter sequence called the **TATA box**
- Located 25 to 35 base pairs upstream of the transcription start site.
- Transcription factors bind to the TATA box and initiate the formation of the RNA polymerase transcription complex, which promotes transcription.
- What is TATA box?

- A DNA sequence that indicates where a genetic sequence can be read and decoded and transcription begins.
- The TATA box is named for its conserved DNA sequence, which is most commonly TATAAA.
- The TATA box is able to define the direction of transcription and also indicates the DNA strand to be read.
- Proteins called transcription factors can bind to the TATA box and recruit an enzyme called RNA polymerase, which synthesizes RNA from DNA.

# What are Enhancers and Silencers ?

- **Enhancers** are short motifs that contain binding sites for transcription factors that can act at a distance (often several kb) from a gene to stimulate transcription.
- **Silencers** suppress gene expression during differentiation and progression through the cell cycle and/or confine (lock) it within specific chromatin boundaries (and thus are also called '**insulators**')  
silencers lie at the opposite functional extreme lie silencers.

# Promoter Mutation ?

**Depending on the location and the nature of the genetic defect, a mutation in the promoter region of a gene may disrupt the normal processes of gene activation by disturbing the ordered recruitment of TFs at the promoter. As a result a promoter mutation can decrease or increase the level of mRNA and thus protein.**

# Control Genes ?

- **Terminator (Stop Codon)**

A stop codon is actually a nucleotide triplet within messenger RNA which signals termination of the translation process. The nonsense codon actually terminates the process of protein formation. This is done by stopping the addition of any more amino acids in the protein chain, thus, the chances of making mistakes during protein formation are lowered considerably.

Codon		Name
DNA	RNA	
TAG	UAG	"amber"
TAA	UAA	"ochre"
TGA	UGA	"opal" (or "umber")

# Presence of mutation in Stop Codon?

In genetics, a point-nonsense mutation is a point mutation in a sequence of DNA that results in a premature stop codon, or a point-nonsense codon in the transcribed mRNA, and in a truncated, incomplete, and usually nonfunctional protein product.

| THANK YOU