The gene & protein synthesis

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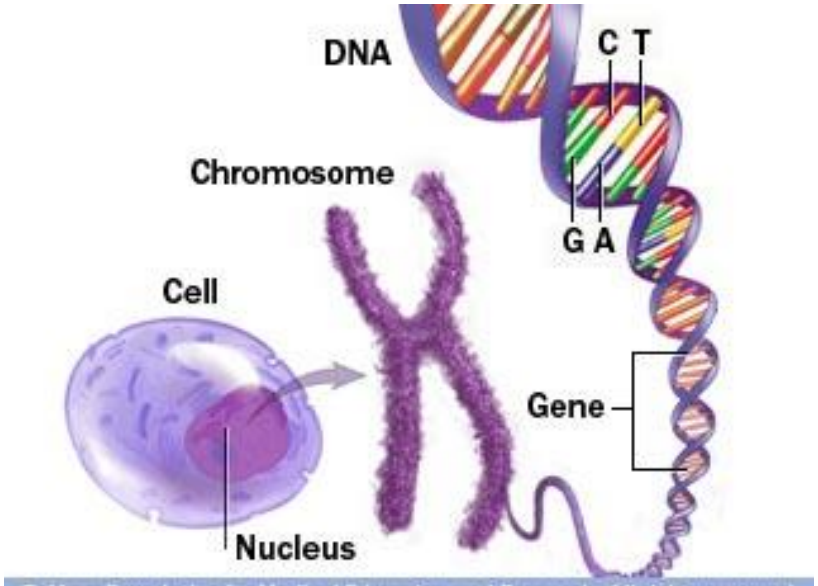
GENES

Genes are a set of instructions that determine what the organism is like, its appearance, how it survives, and how it behaves in its environment.

There are 30,000 genes in each cell



examples of genetic diseases

Neurology
Muscular dystrophy
Spinocerebellar ataxia
Hereditary neuropathy
Dystonia
Early onset Alzheimer's disease
Familial multiple sclerosis
Familial amyotrophic lateral sclerosis
Neurofibromatosis

Nephrology
Autosomal dominant polycystic kidney disease
Hereditary nephritis

Disorders of renal physiology

Hematology
Hemoglobinopathies
Hereditary disorders of hemostasis
Hereditary hypercoagulability

Pulmonary disorders
Adult-onset cystic fibrosis
Alpha-1-antitrypsin deficiency

Cardiac disorders
Conduction abnormalities
Cardiomyopathy

Infectious disease
Immune deficiencies

Metabolic disorders
Hemochromatosis
Lipid disorders
Homocysteine

Gastroenterology
Osler-Weber-Rendu disease
Polyposis

Oncology
BRCA1/2
Familial adenomatous polyposis and hereditary nonpolyposis colon cancer
Familial prostate cancer
Multiple endocrine neoplasia
Hippel-Lindau disease
Li-Fraumeni syndrome

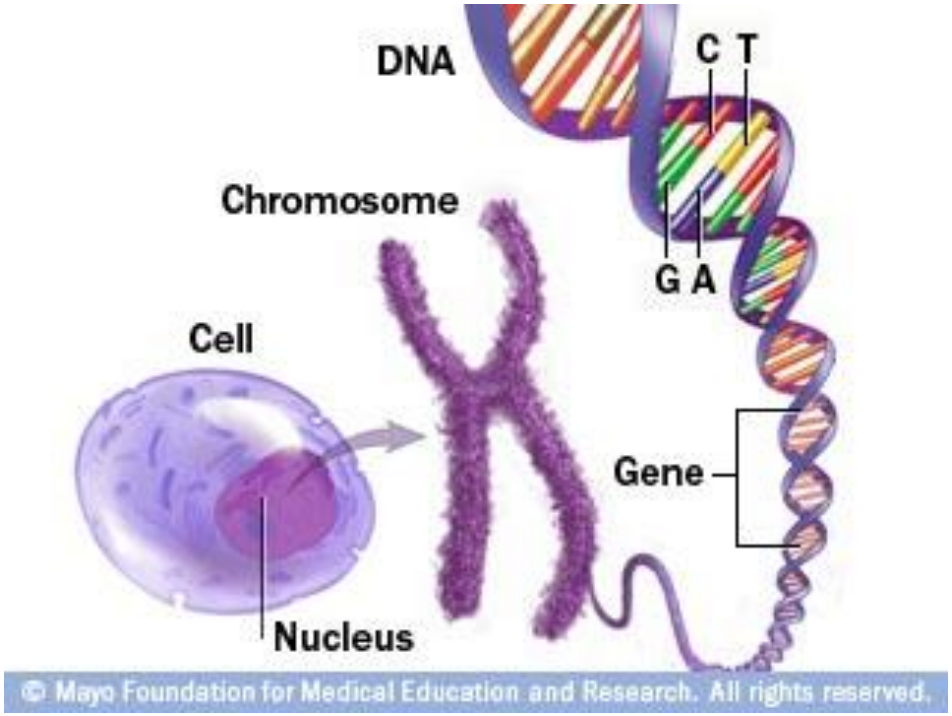
Musculoskeletal disorders
Inherited disorders of connective tissue — Marfan's, Ehlers-Danlos, osteogenesis imperfecta

Dermatology
Icthyosis
Bullous disorders

*This is a far from complete list.

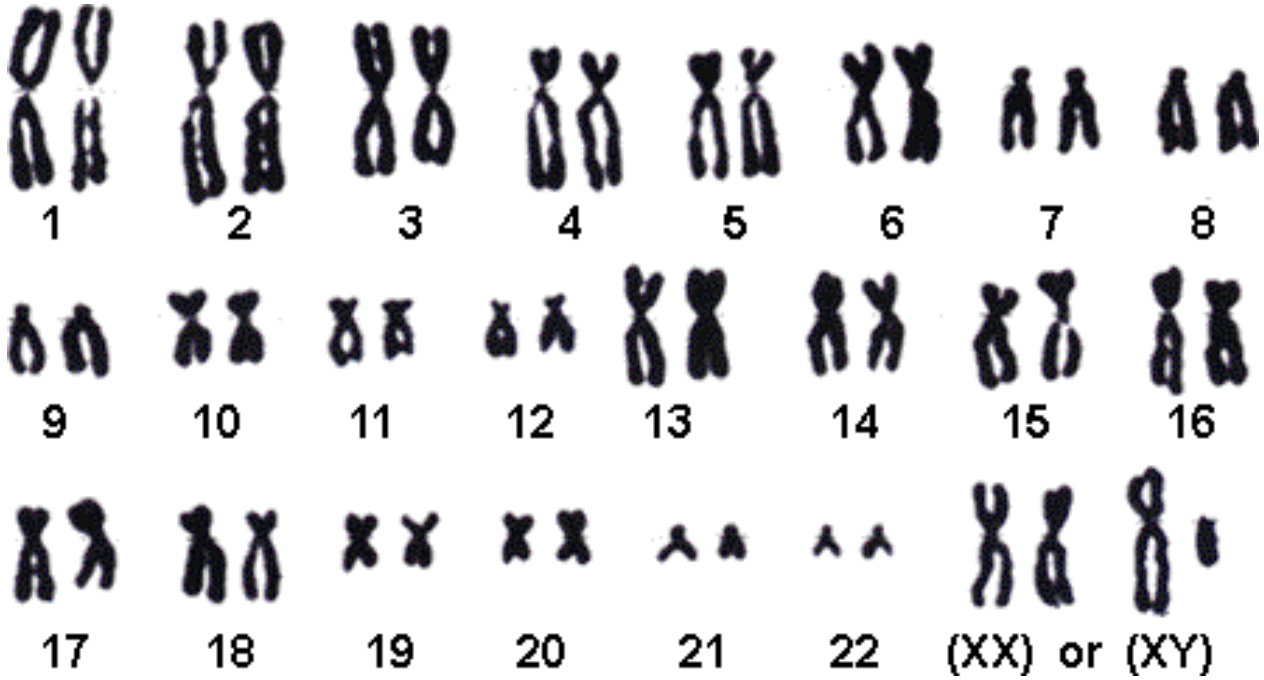
What is genetic material?

- There are chromosomes in the nucleus of each cell
- Each **chromosome** is made up of DNA tightly coiled many times around proteins called histones that support its structure.
- DNA is actually 3 METRES long.



Chromosomes

- Humans = 46 chromosomes



The Barr body

- a small, densely staining structure in the cell nuclei of females, consisting of a condensed, inactive X chromosome. It is regarded as diagnostic of genetic femaleness

DNA

- Deoxyribonucleic acid (DNA)
- a self-replicating material present in nearly all living organisms as the main constituent of chromosomes.
- It is the carrier of genetic information.

Building blocks of DNA

- Phosphoric acid (phosphate group)
- Sugar called Deoxyribose
- 4 nitrogenous bases:

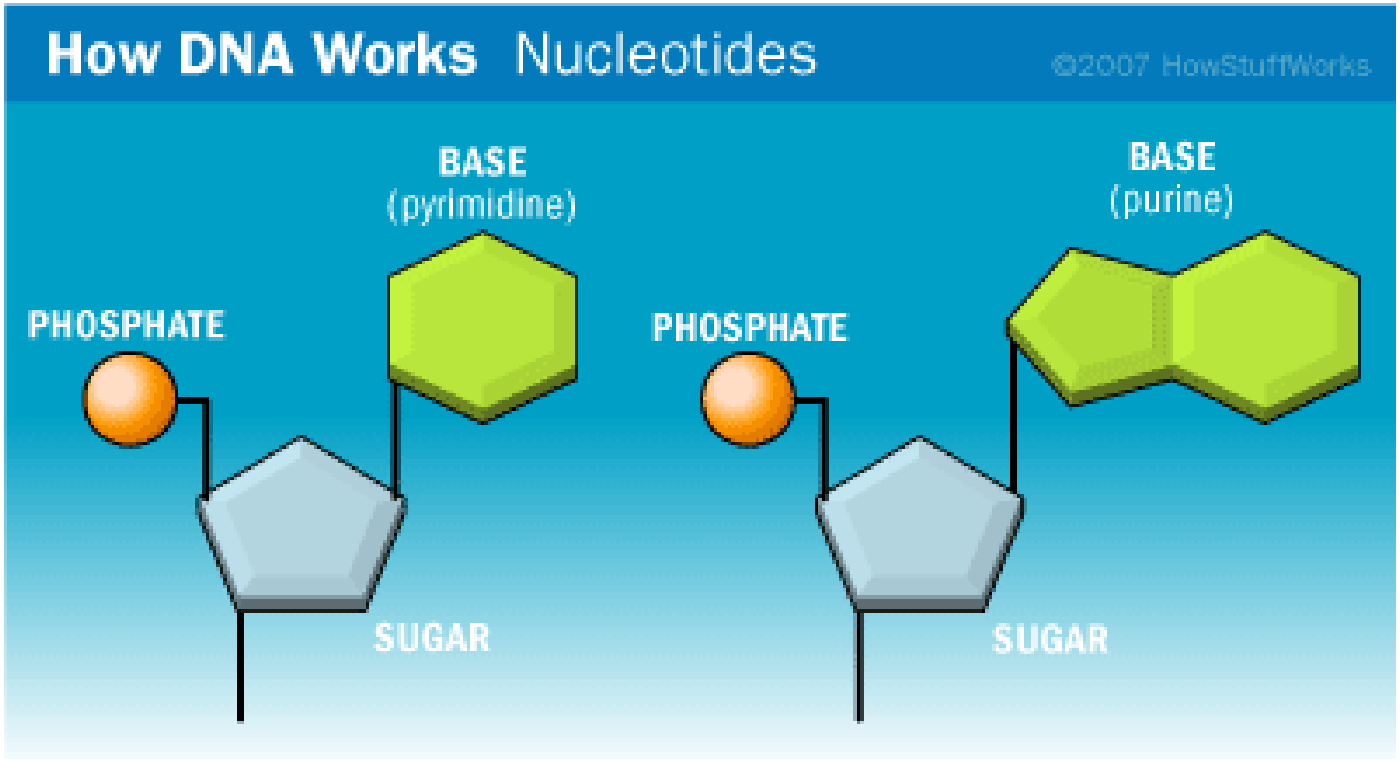
2 purines:

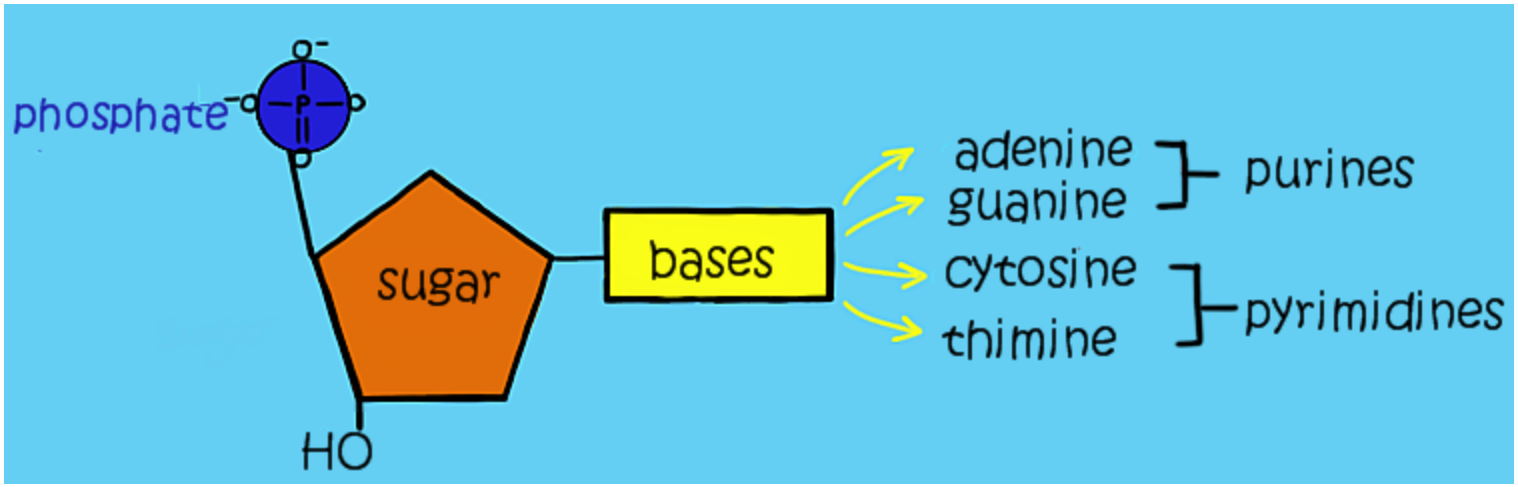
ADENINE , GUANINE

2 pyrimidines,

THYMINE, CYTOSINE

- The phosphoric acid and deoxyribose form the two helical strands that are the backbone of the DNA molecule and the nitrogen bases lie between the two strands and connect them.


How DNA Works Nucleotides
©2007 HowStuffWorks
BASE
(pyrimidine)
PHOSPHATE
SUGAR
BASE
(purine)
PHOSPHATE
SUGAR


phosphate
sugar
bases
HO
adenine
guanine
purines
cytosine
thimine
pyrimidines

DNA STRUCTURE

- Loose hydrogen bonds between purine and pyrimidine bases (holding the two DNA strands together)
- Purine base bonds with pyrimidine base
- Adenine bonds with Thymine
- Guanine bonds with Cytosine

HOW DNA CONTROLS CELL FUNCTION

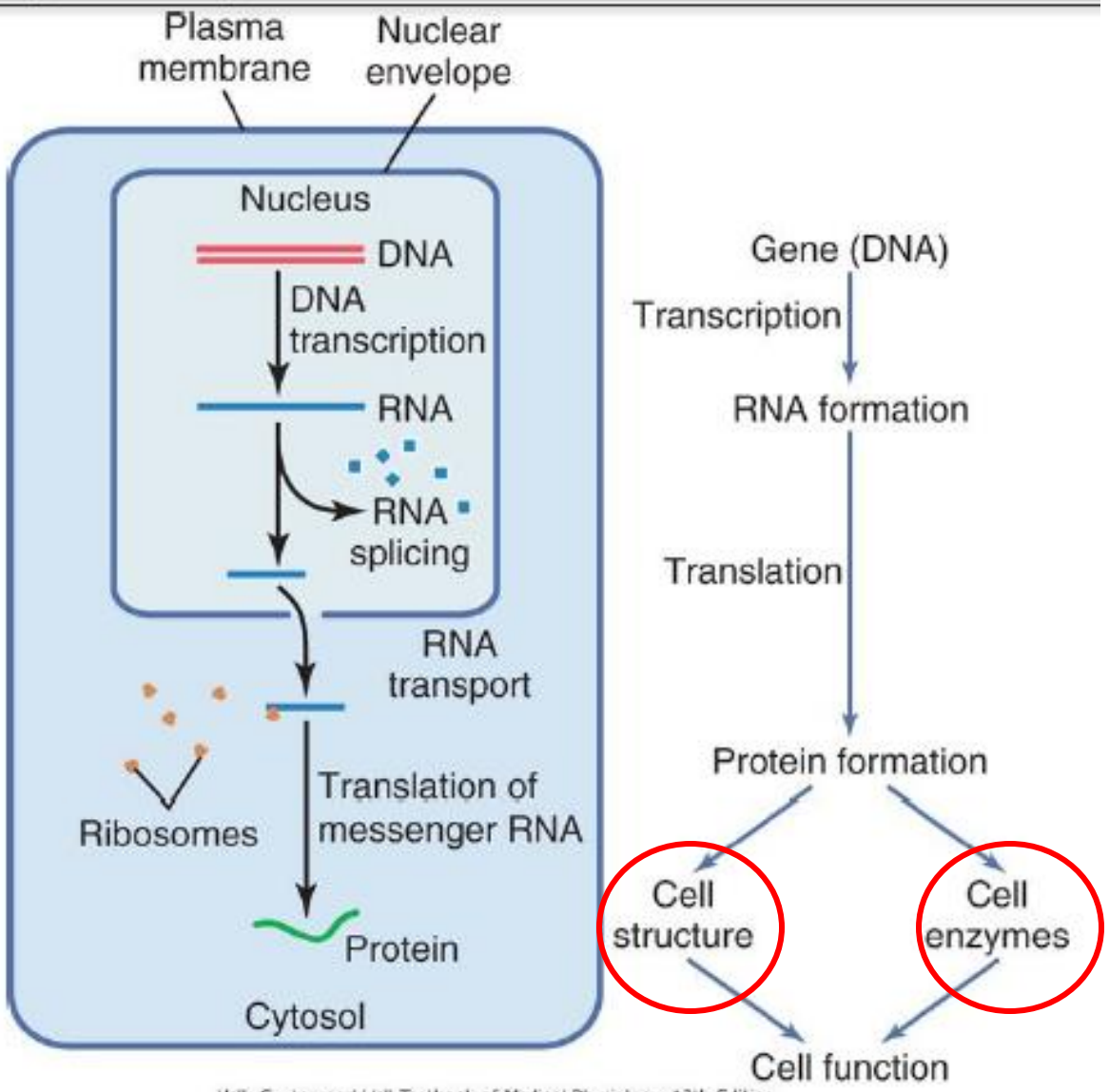


Figure 3-1 General schema by which the genes control cell function.

THE DNA & PROTEIN SYNTHESIS

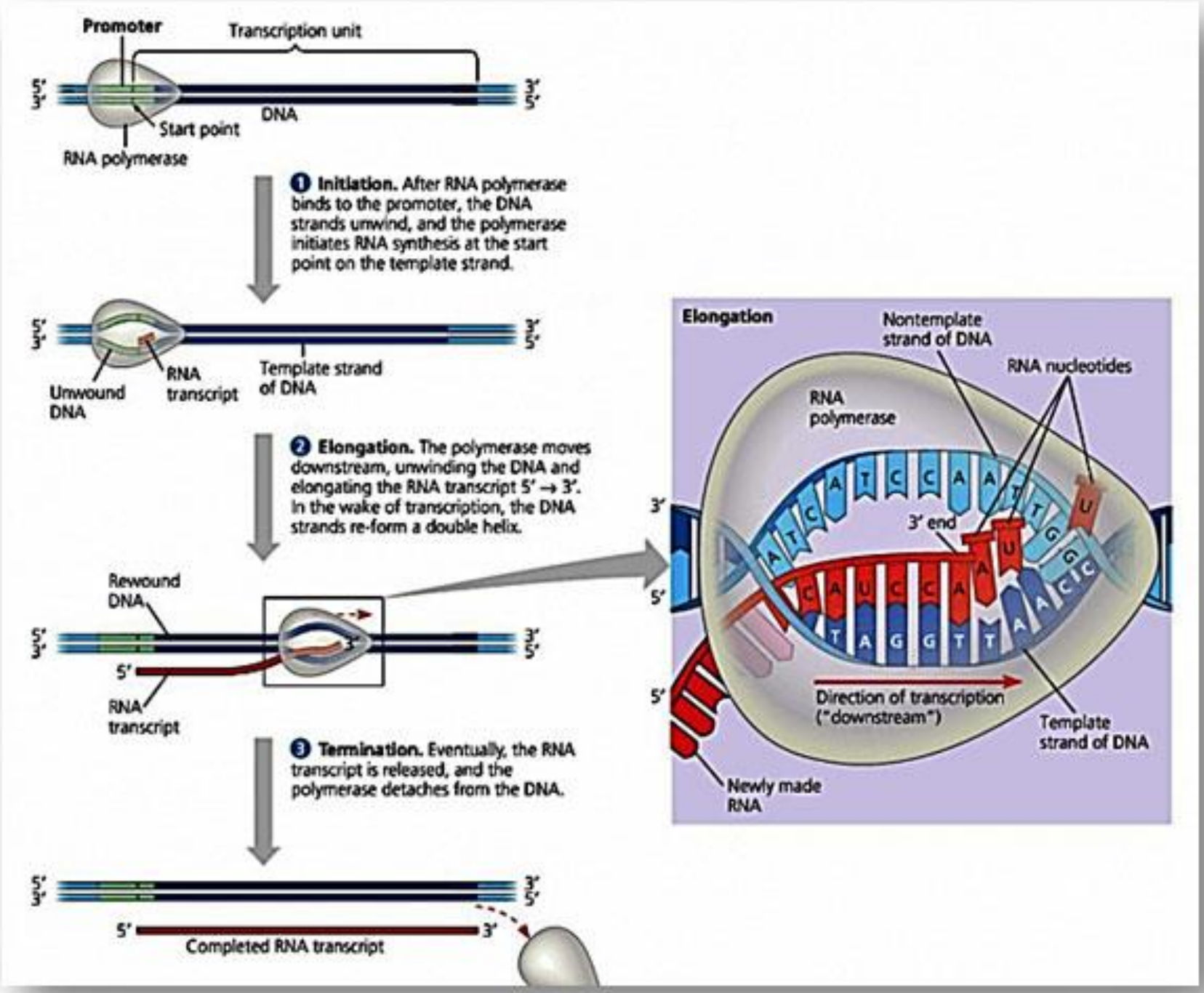
Stages of protein synthesis

Stage 1: TRANSCRIPTION

- DNA is located in the nucleus of the cell, yet most cell functions occur in the cytoplasm
- How can DNA control the chemical reactions of the cytoplasm?
- Intermediary nucleic acid → RNA
- Its formation is controlled by the DNA in the nucleus.
- DNA code is transferred to RNA by **TRANSCRIPTION**

Transcription = RNA SYNTHESIS

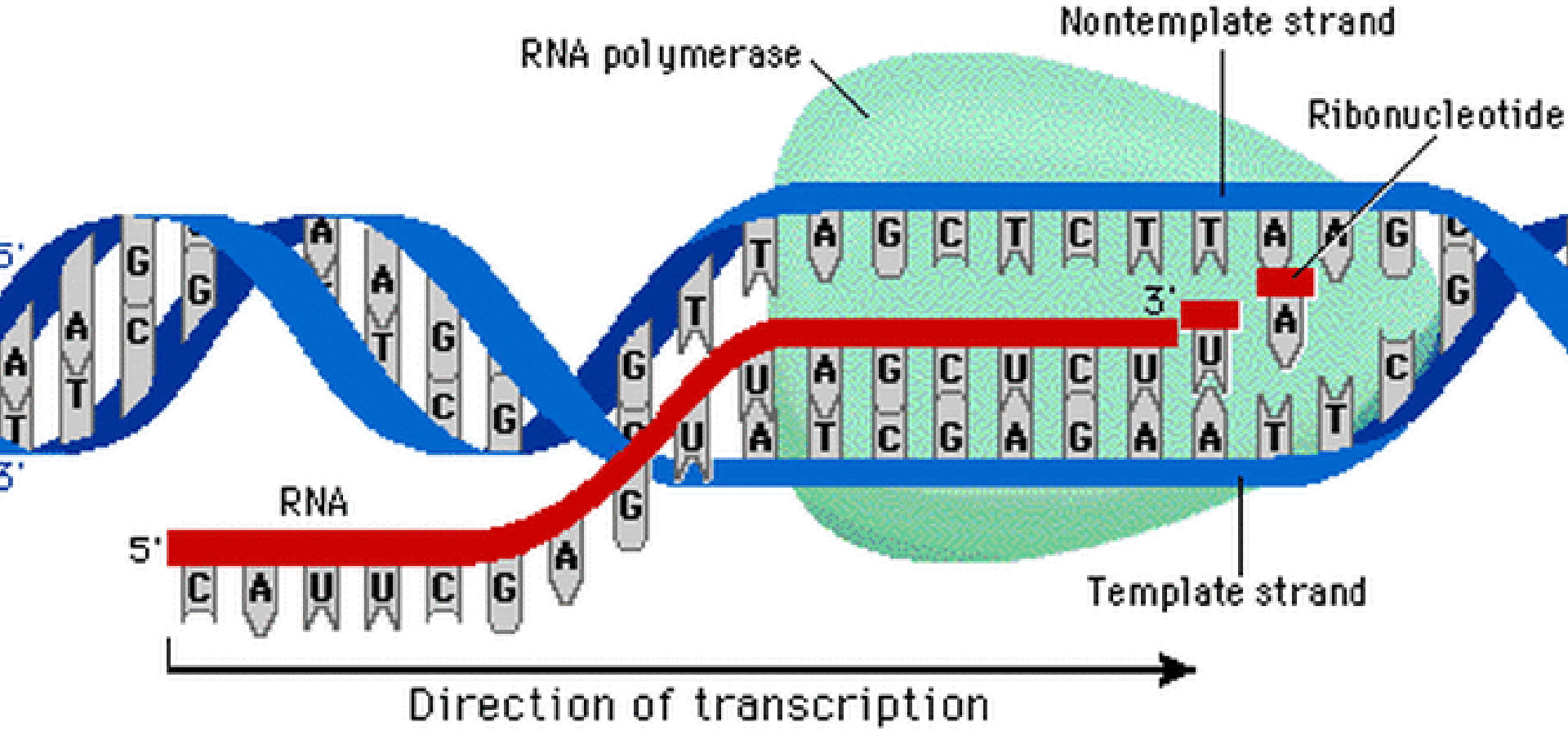
- During synthesis of RNA, the two strands of DNA separate temporarily;
- One of these strands is used as a template for synthesis of RNA molecule (template strand)
- Code triplets in the DNA cause the formation of complementary code triplets in the RNA (codons)
- Codons will in turn control the sequence of amino acids in a protein (TRANSLATION)


Promoter
Transcription unit
5′
3′
DNA
3′
5′
Start point
RNA polymerase
1 Initiation. After RNA polymerase binds to the promoter, the DNA strands unwind, and the polymerase initiates RNA synthesis at the start point on the template strand.
Unwound DNA
RNA transcript
Template strand of DNA
2 Elongation. The polymerase moves downstream, unwinding the DNA and elongating the RNA transcript 5′ → 3′. In the wake of transcription, the DNA strands re-form a double helix.
Rewound DNA
RNA transcript
3 Termination. Eventually, the RNA transcript is released, and the polymerase detaches from the DNA.
Completed RNA transcript
Elongation
Nontemplate strand of DNA
RNA nucleotides
RNA polymerase
3′ end
Direction of transcription ("downstream")
Template strand of DNA
Newly made RNA

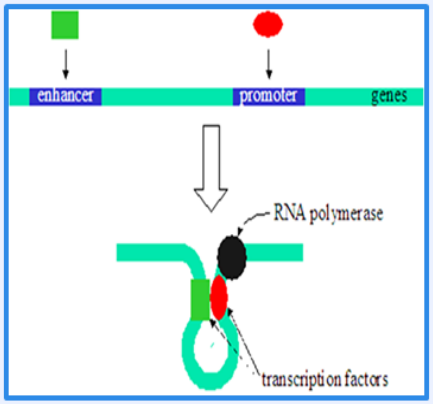
Transcription Enzymes

RNA polymerase: The enzyme that controls transcription and is characterized by:

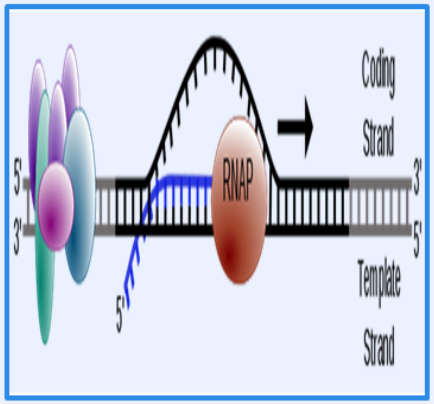
- It unwinds double helical DNA to produce a single-stranded DNA template,
- It selects the correct ribonucleotide and catalyzes the formation of a phosphodiester bond,
- It detects termination signals where transcript ends.


Nontemplate strand
RNA polymerase
Ribonucleotide
3'
RNA
5'
Template strand
Direction of transcription

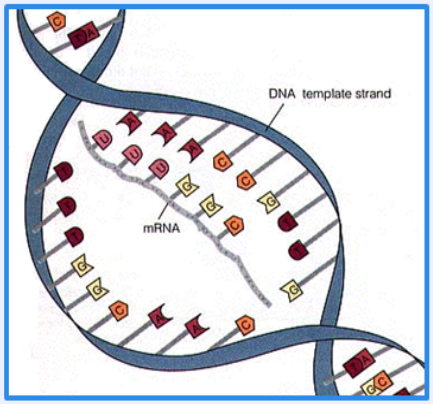
Transcription is divided into 3 phases:



Initiation



Elongation



Termination

Initiation

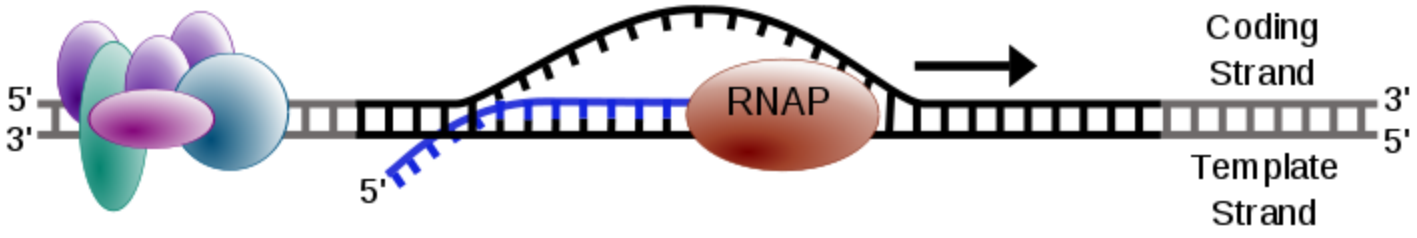
- The RNA polymerase binding causes the unwinding of the DNA double helix which expose at least 12 bases on the template.

- This is followed by initiation of RNA synthesis at this starting point.

Elongation

- **RNA polymerase** directs the sequential binding of ribonucleotides to the growing RNA chain in the 5' - 3' direction.

- Each ribonucleotide is inserted into the growing RNA strand following the rules of base pairing. This process is repeated until the desired RNA length is synthesized……..



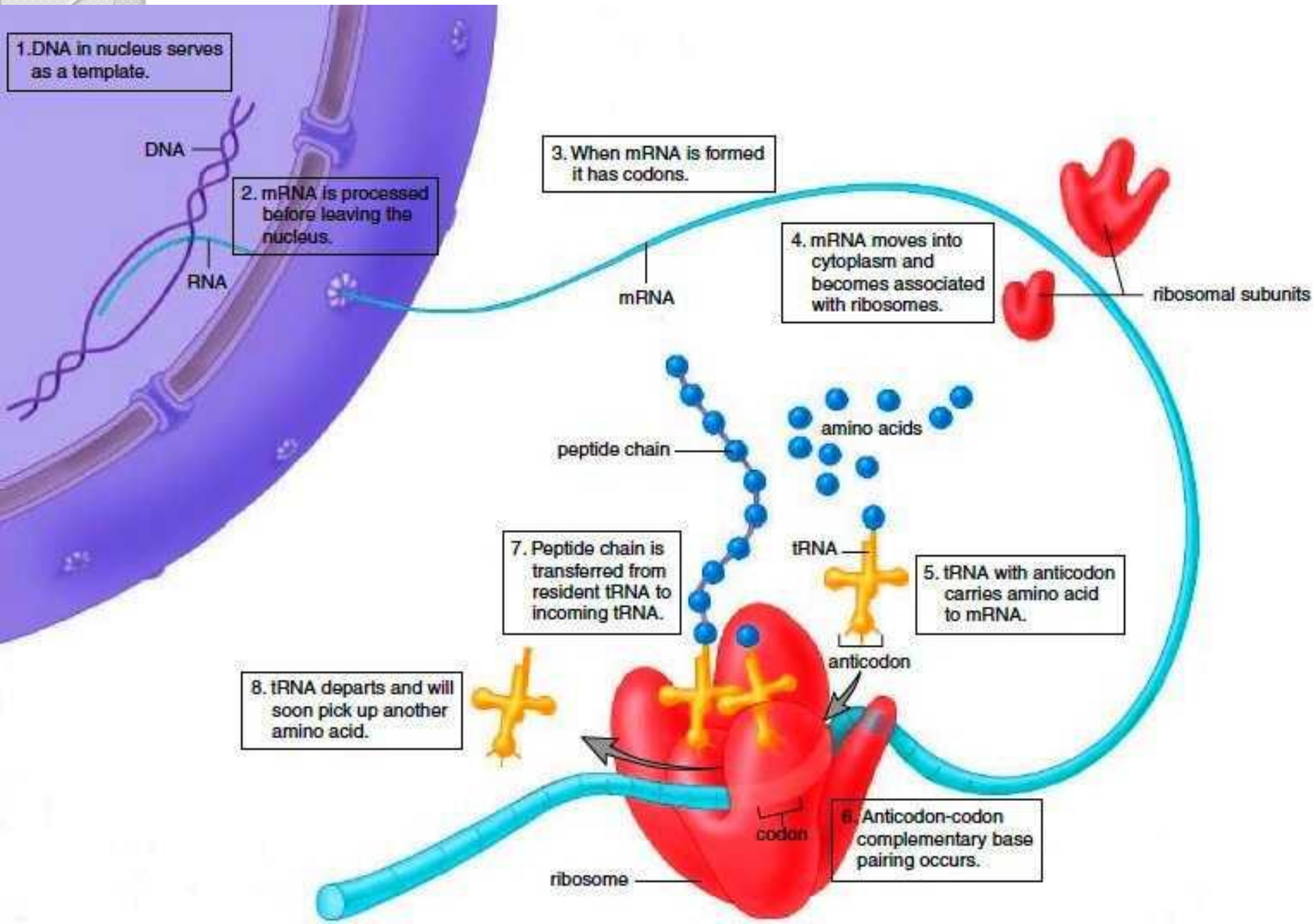
Termination

- Terminators at the end of genes; signal termination. These work in conjunction with RNA polymerase to loosen the association between RNA product and DNA template. The result is that the RNA dissociate from RNA polymerase and DNA and stops transcription.

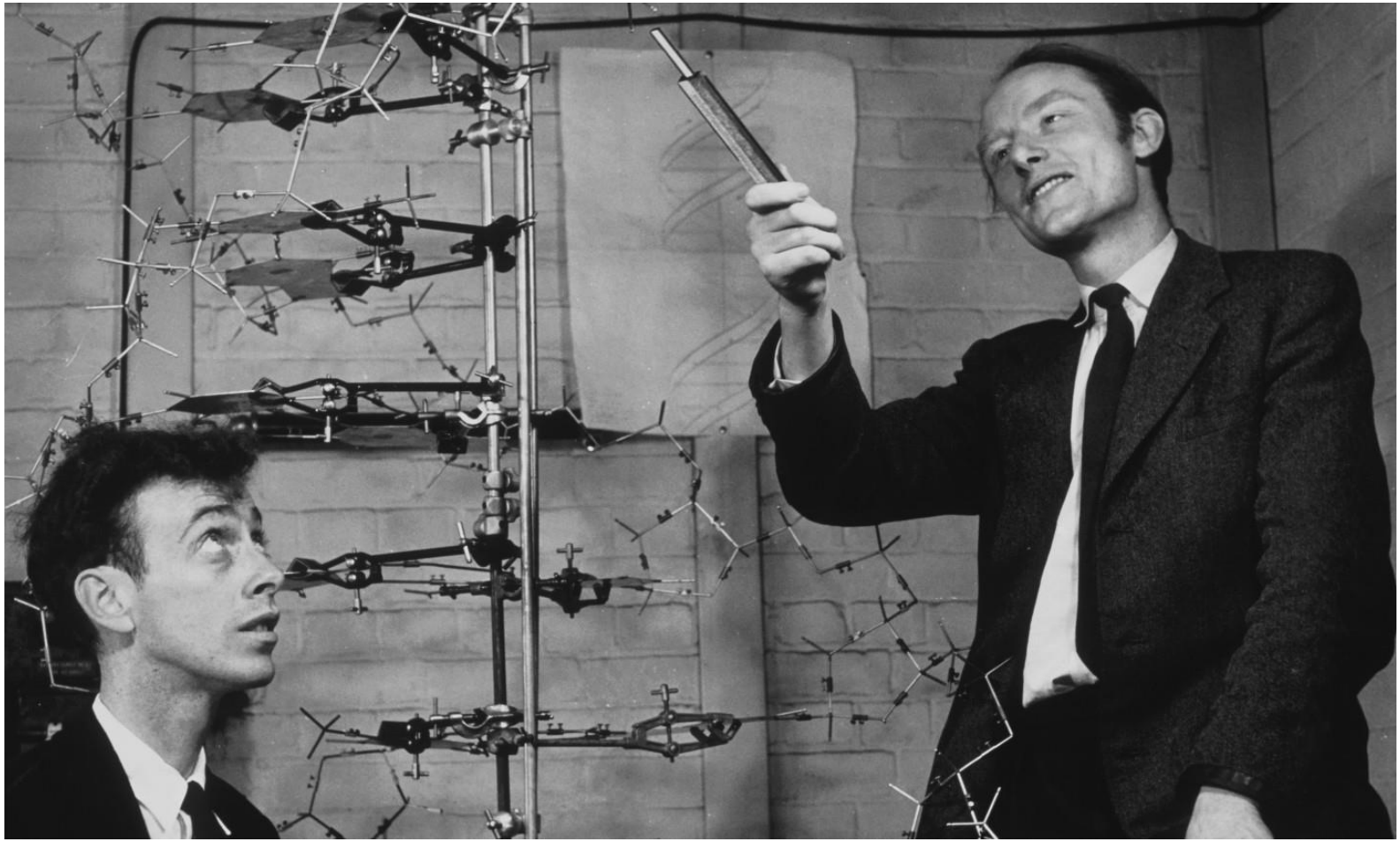
- The product is **immature RNA or pre mRNA** (Primary transcript).

Immature mRNA

- These immature mRNAs are processed (**Post transcription processing**) in the nucleus to give mature mRNAs that are transported to the cytoplasm where to participate in protein synthesis.


1. DNA in nucleus serves as a template.
DNA
2. mRNA is processed before leaving the nucleus.
RNA
3. When mRNA is formed it has codons.
mRNA
4. mRNA moves into cytoplasm and becomes associated with ribosomes.
ribosomal subunits
amino acids
peptide chain
tRNA
5. tRNA with anticodon carries amino acid to mRNA.
anticodon
7. Peptide chain is transferred from resident tRNA to incoming tRNA.
8. tRNA departs and will soon pick up another amino acid.
codon
6. Anticodon-codon complementary base pairing occurs.
ribosome

THANK YOU (Watson and Crick)



ANY QUESTIONS??

Video of protein synthesis