«Biomedical foundations of special pedagogy and psychology:
the basics of human genetics»

BASES OF MOLECULAR GENETICS

Genetics is a fundamental science that studies the laws of heredity and variability that are universal for all living organisms in unity, since heredity is conservative in nature, and variability generates not only the diversity of living nature as a whole, but also provides intraspecific diversity.

Genetics is a fundamental science that studies the processes of the continuity of life at the molecular, cellular, organismic and population levels.

In accordance with research methods, genetics is divided into:

- Biochemical
- Physiological
- Molecular
- Population.

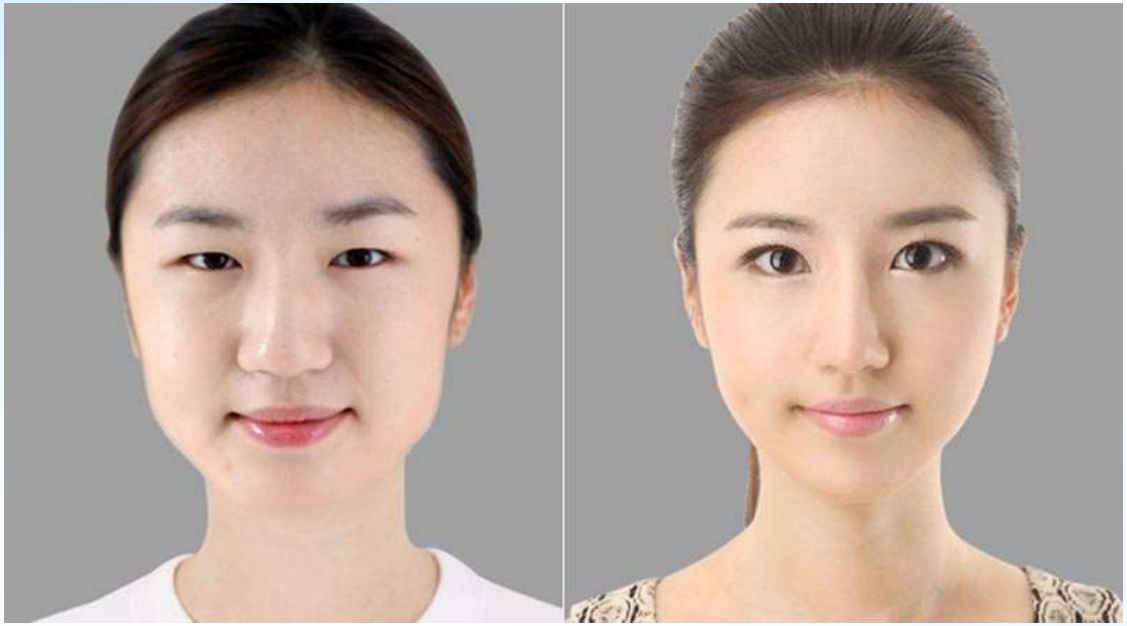
In relation to the object of research distinguish between genetics:

- Microorganisms
- Plants
- Animals
- Human.

Heredity - the property of living organisms to maintain similar characteristics from generation to generation, to ensure functional continuity, as well as a certain pattern of individual development (ontogenesis) in certain environmental conditions.

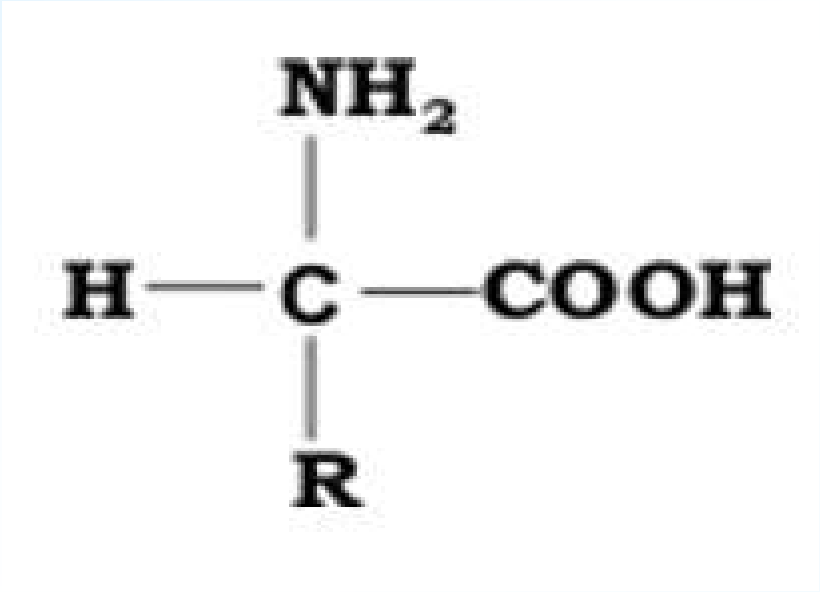
Variability is the property of living organisms to lose existing or acquire new traits that distinguish them from their parental forms, as well as the ability of living organisms to respond to environmental factors by morphophysiological changes.

Hereditary (genotypic) - due to the occurrence of mutations and their combinations when crossing;
Non-hereditary (modification) - caused by external conditions and is not rigidly fixed in the genotype;



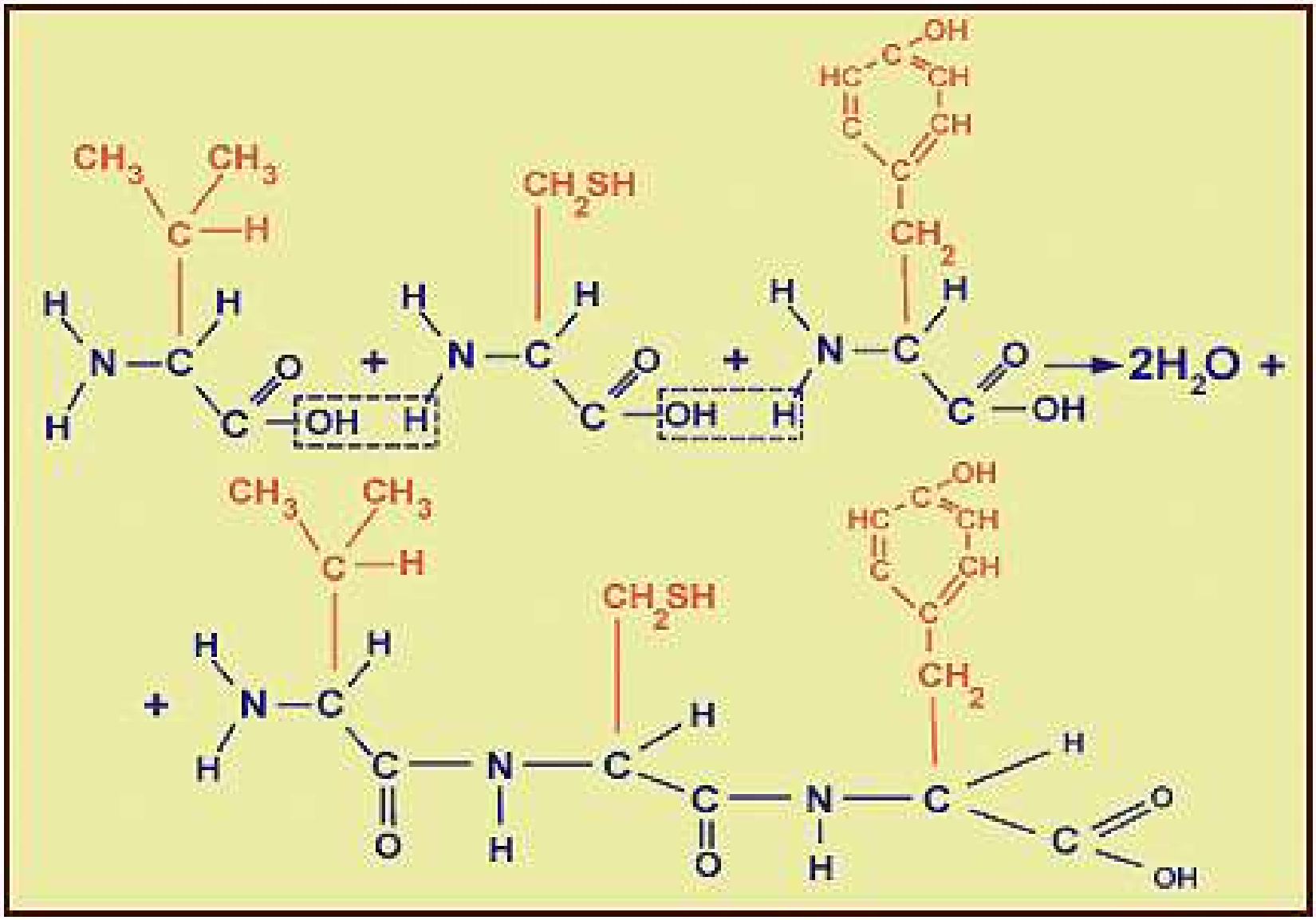
Ontogenetic - changes in the individual development of the body or with the differentiation of cells;
Geographical - the formation of features in organisms of one species under the influence of spatial and geographical factors (forms, races, subspecies, etc.)
Qualitative - a fundamental change in properties and structures;
Quantitative - a change in the magnitude or severity of an indicator

Proteins are large polymer molecules built from monomeric amino acid units. There are **twenty** different types of amino acids in proteins. All protein amino acids (with the exception of proline) are characterized by a common structure, the essential elements of which are:

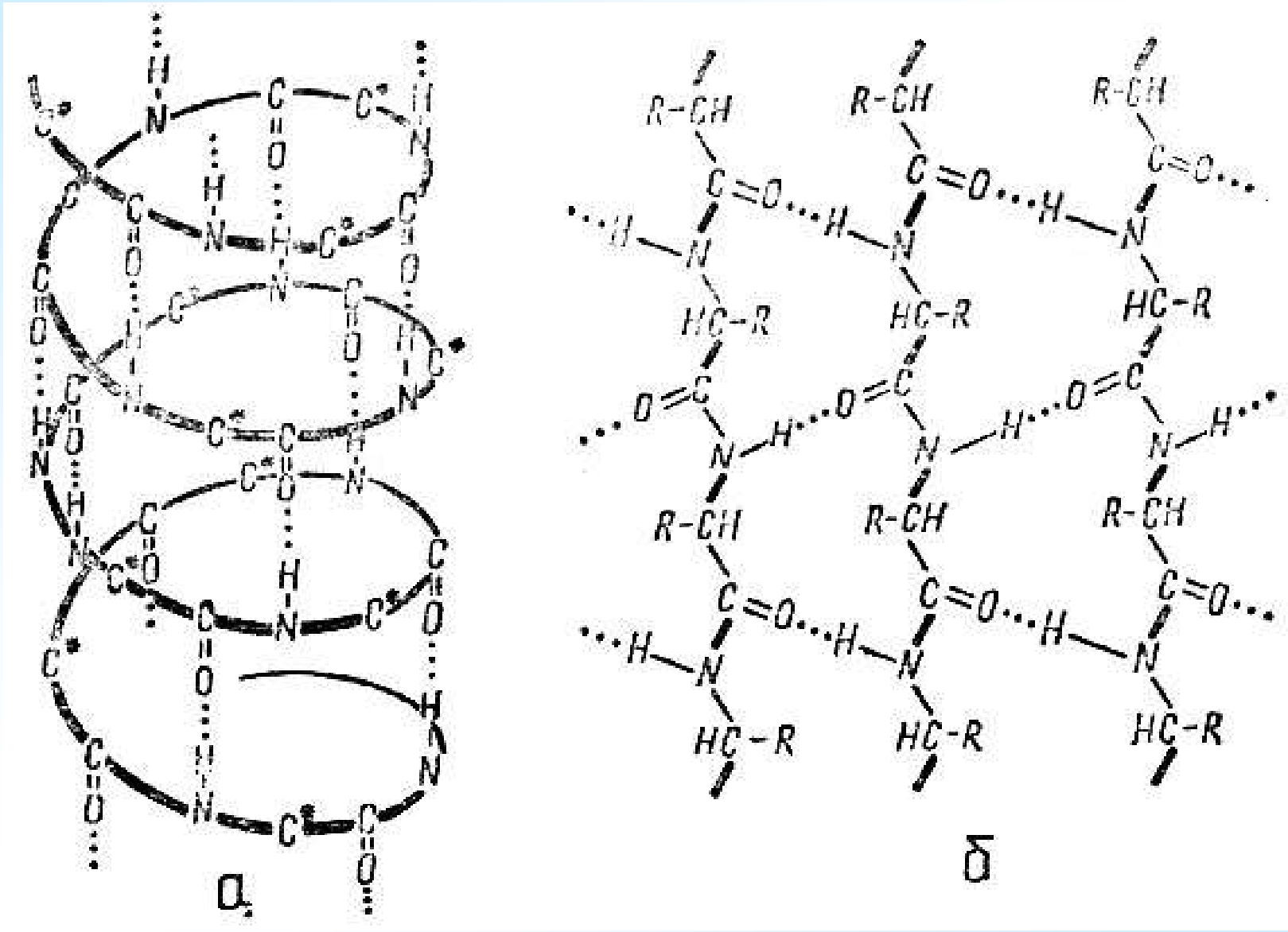


NH_2 – amino group;
COOH – carboxyl group;
H – hydrogen atom;
radical R – side group.

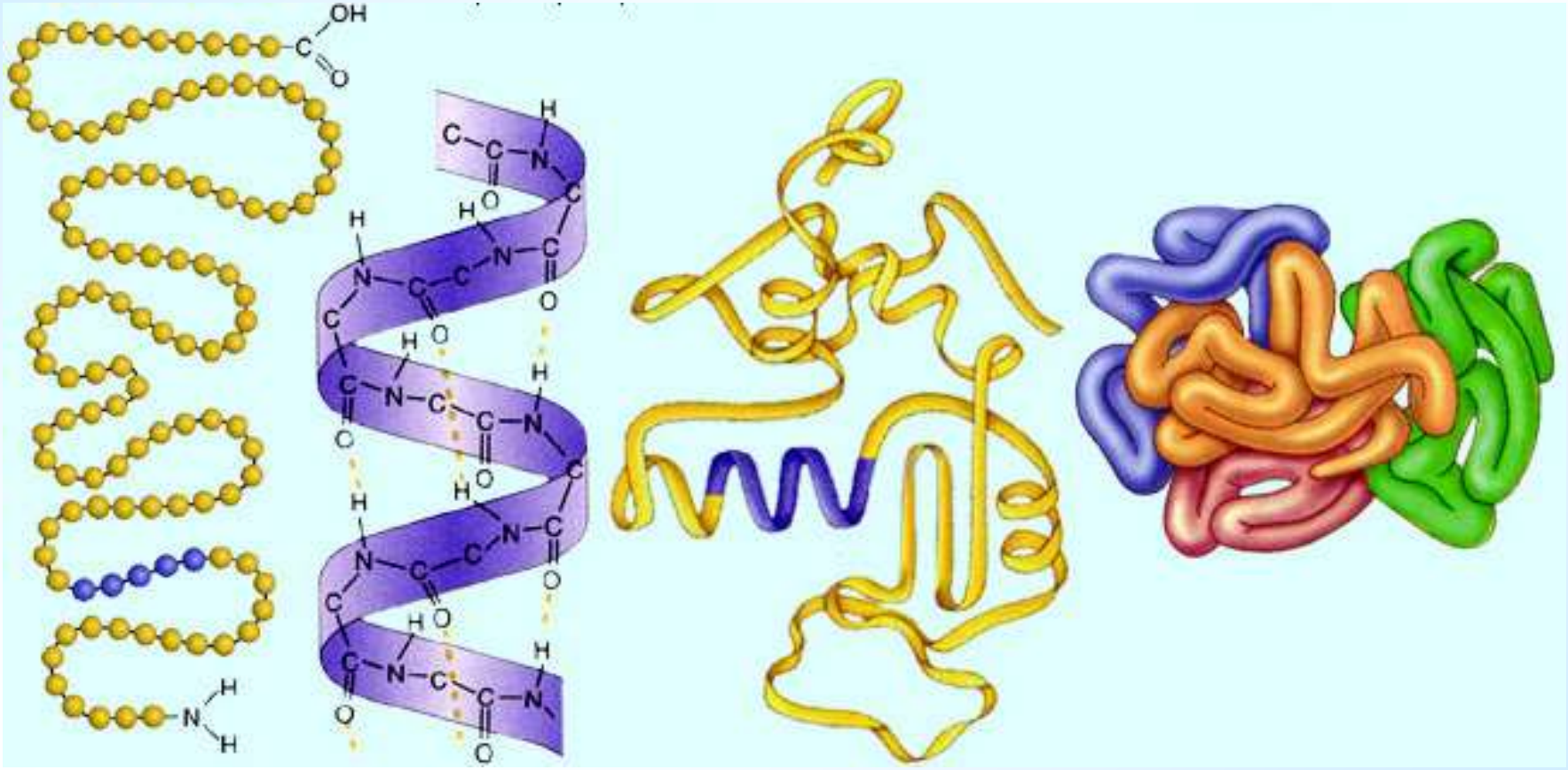
PRIMARY PROTEIN STRUCTURE



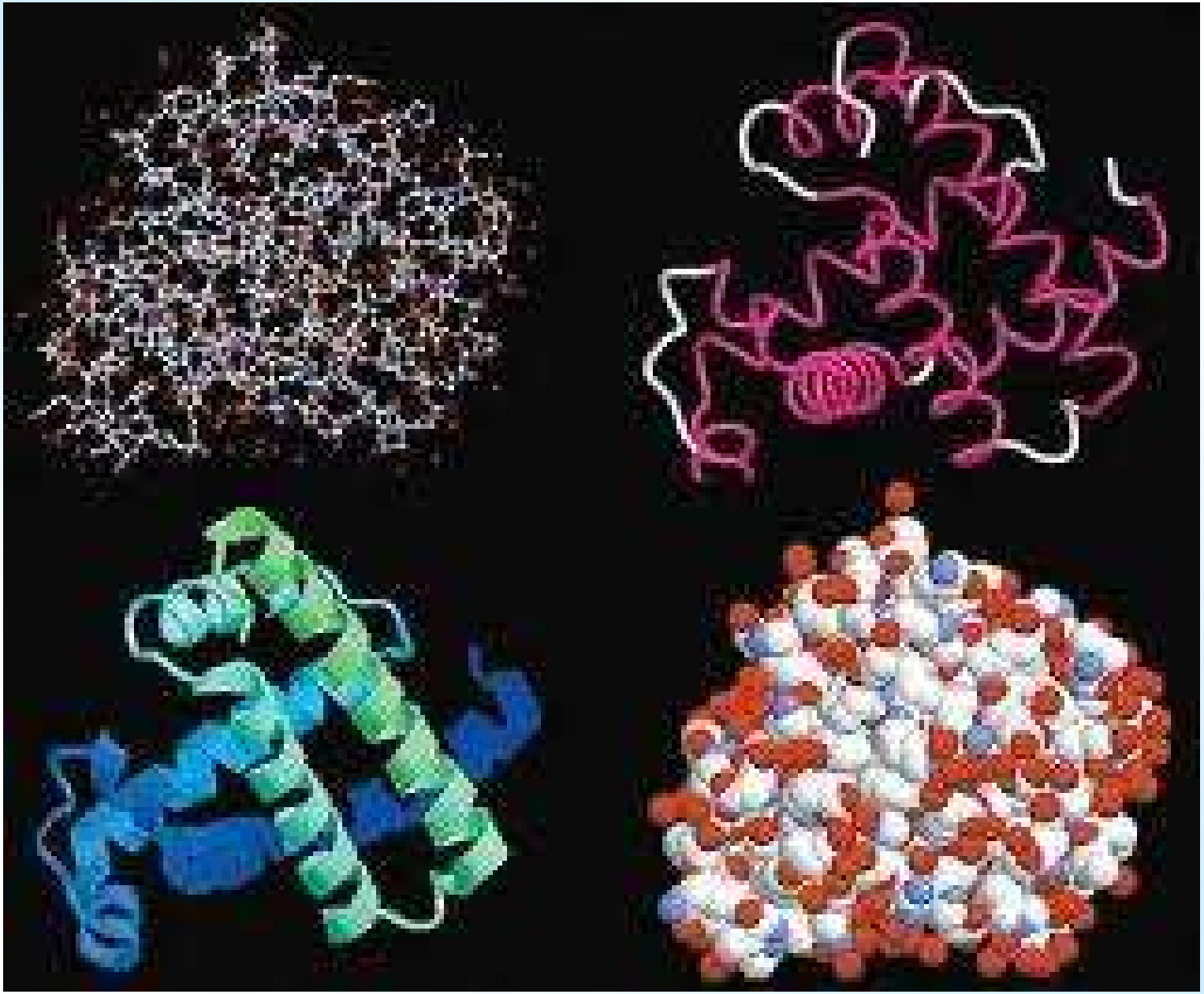
PRIMARY PROTEIN STRUCTURE: SPATIAL MODEL



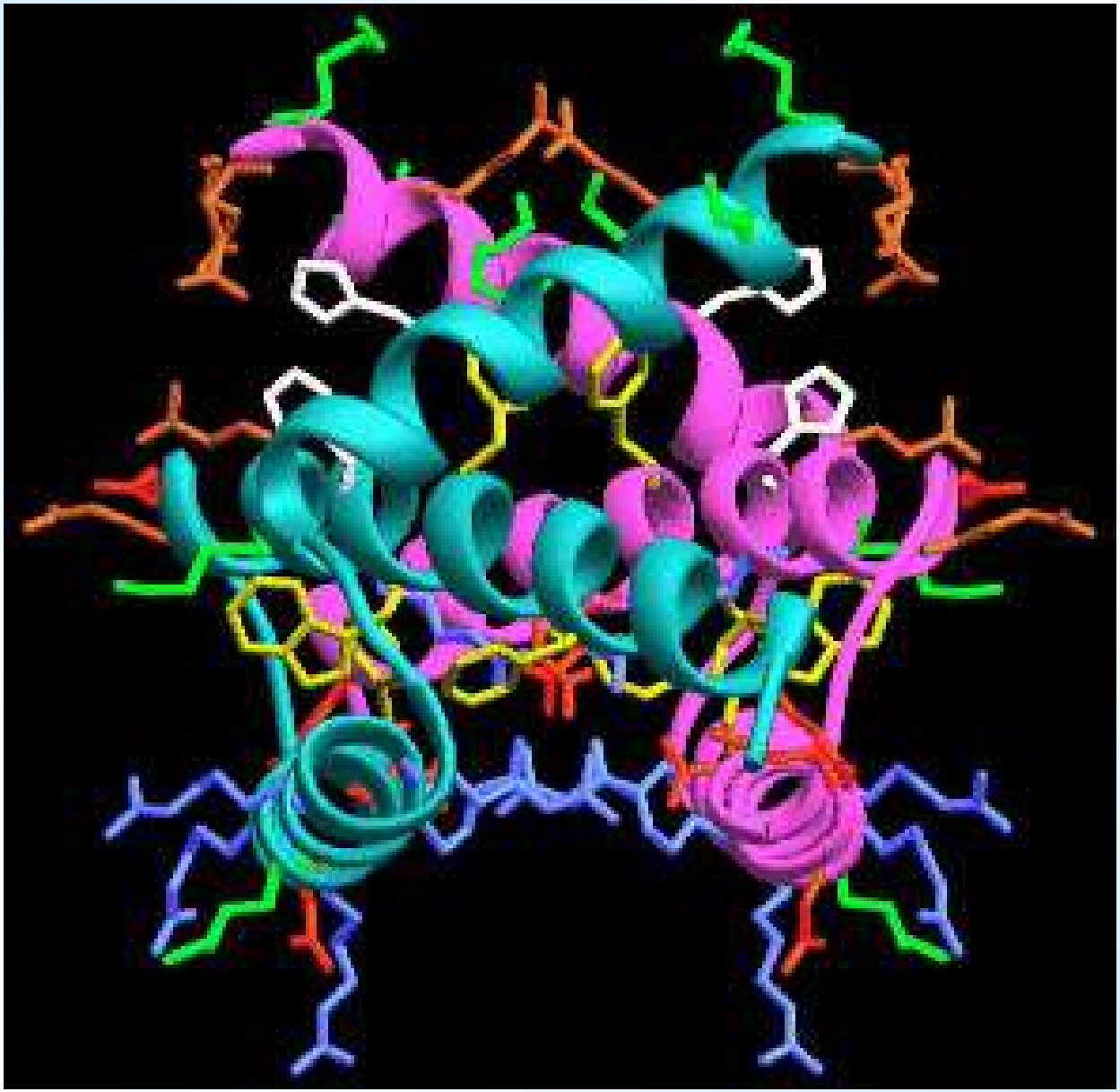
PRIMARY, SECONDARY, TERTIARY, QUATERNARY PROTEIN STRUCTURE



Various proteins under a laser microscope



A mixture of different proteins under a laser microscope



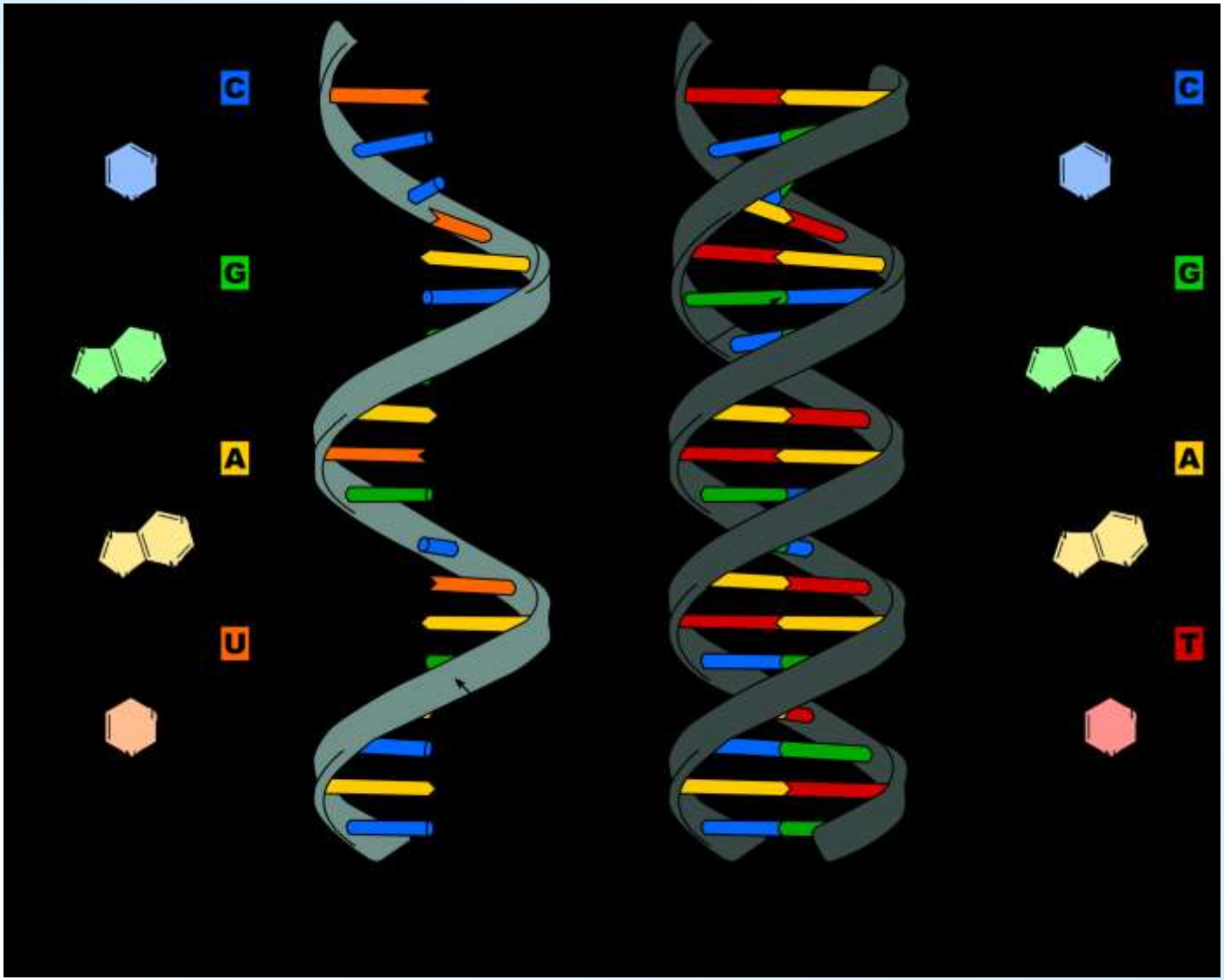
Amino Acid Classification Table (Latest)

Sl. No.	Name	Three letter code	Single letter code	Molecular Weight	pI	Essential/ Non-essential	No. of codons	Remarks
Nonpolar, aliphatic R-group								
1	Glycine	Gly	G	75	5.97	Nonessential	4	Smallest amino acid, Optically inactive
2	Alanine	Ala	A	89	6.01	Nonessential	4	
3	Proline	Pro	P	115	6.48	Nonessential*	4	Imino acid
4	Valine	Val	V	117	5.97	Essential	4	
5	Leucine	Leu	L	131	5.98	Essential	6	
6	Isoleucine	Ile	I	131	6.02	Essential	3	
7	Methionine	Met	M	149	5.74	Essential	1	Sulfur containing
Aromatic R-Group								
8	Phenylalanine	Phe	F	165	5.48	Essential	2	
9	Tyrosine	Tyr	Y	181	5.66	Nonessential*	2	
10	Tryptophan	Trp	W	204	5.89	Essential	1	Least occurring amino acid in proteins
Polar, uncharged R-group								
11	Serine	Ser	S	105	5.68	Nonessential*	6	
12	Threonine	Thr	T	119	5.87	Essential	4	
13	Cysteine	Cys	C	121	5.07	Nonessential*	2	Sulfur containing
14	Asparagine	Asn	N	132	5.41	Nonessential	2	
15	Glutamine	Gln	Q	146	5.65	Nonessential*	2	
Positively charged R-Group (Basic amino acids)								
16	Lysine	Lys	K	146	9.74	Essential	2	
17	Arginine	Arg	R	174	10.76	Nonessential*	6	Highest pI
18	Histidine	His	H	155	7.59	Essential	2	pI near physiological pH
Negatively charged R-Group (Acidic amino acids)								
19	Aspartate	Asp	D	133	2.77	Nonessential	2	
20	Glutamate	Glu	E	147	3.22	Nonessential	2	
Proteinogenic non-standard amino acids (coded by 'amber' stop codon – UAG)								
21	Selenocysteine	Sec	U	168	5.47	Nonessential*	1	Selenium containing, 21st amino acid
22	Pyrrolysine	Pyl	O	255	-	Nonessential*	1	Largest amino acid, 22nd amino acid, present in methanogenic archaea

* *Conditionally Essential*

DNA structure and function

DNA - deoxyribonucleic acid - a biological macromolecule, a carrier of genetic information in all eukaryotic cells.



The relationship between the number of different bases in DNA was established by **Chargaff in 1949** and played an important role in the construction of the double helix. Chargaff found that in DNA of very different origins, the amount of adenine is equal to the amount of thymine, and the amount of guanine is equal to the amount of cytosine. This pattern was called "Chargaff's rule" and is expressed as follows:

Chargaff

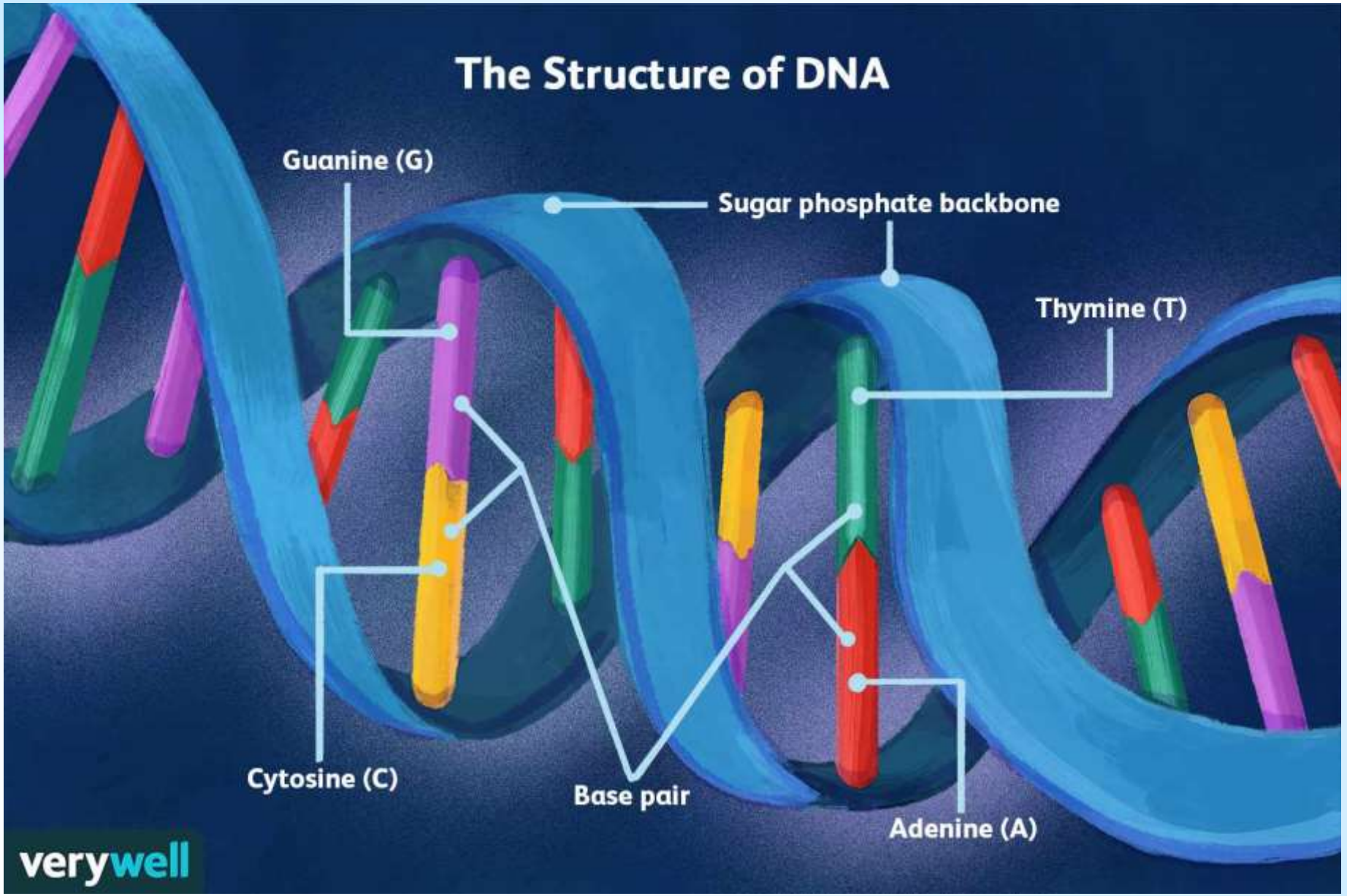
- **Chargaff's rule**: C = G and T = A

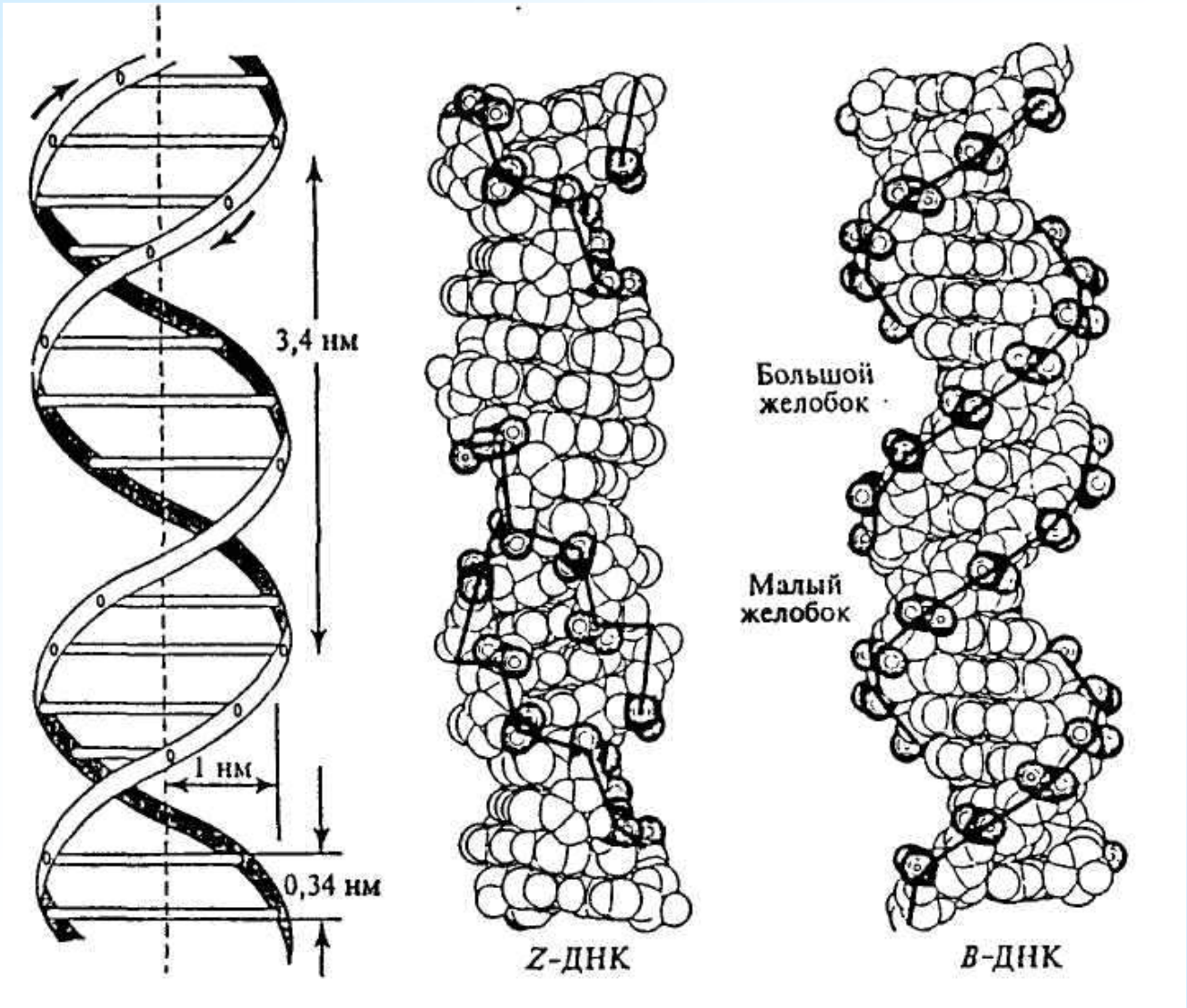
Chargaff's Data

	Base Composition (Mole Percent)			
Organism	A	T	G	C
Escherichia coli	26.0	23.9	24.9	25.2
Yeast	31.3	32.9	18.7	17.1
Herring	27.8	27.5	22.2	22.6
Rat	28.6	28.4	21.4	21.5
Human	30.9	29.4	19.9	19.8

- The percentages of guanine [G] and cytosine [C] bases are almost equal in any sample of DNA.

- The percentages of adenine [A] and thymine [T] bases are almost equal in any sample of DNA.


The Structure of DNA
Guanine (G)
Sugar phosphate backbone
Thymine (T)
Cytosine (C)
Base pair
Adenine (A)
verywell

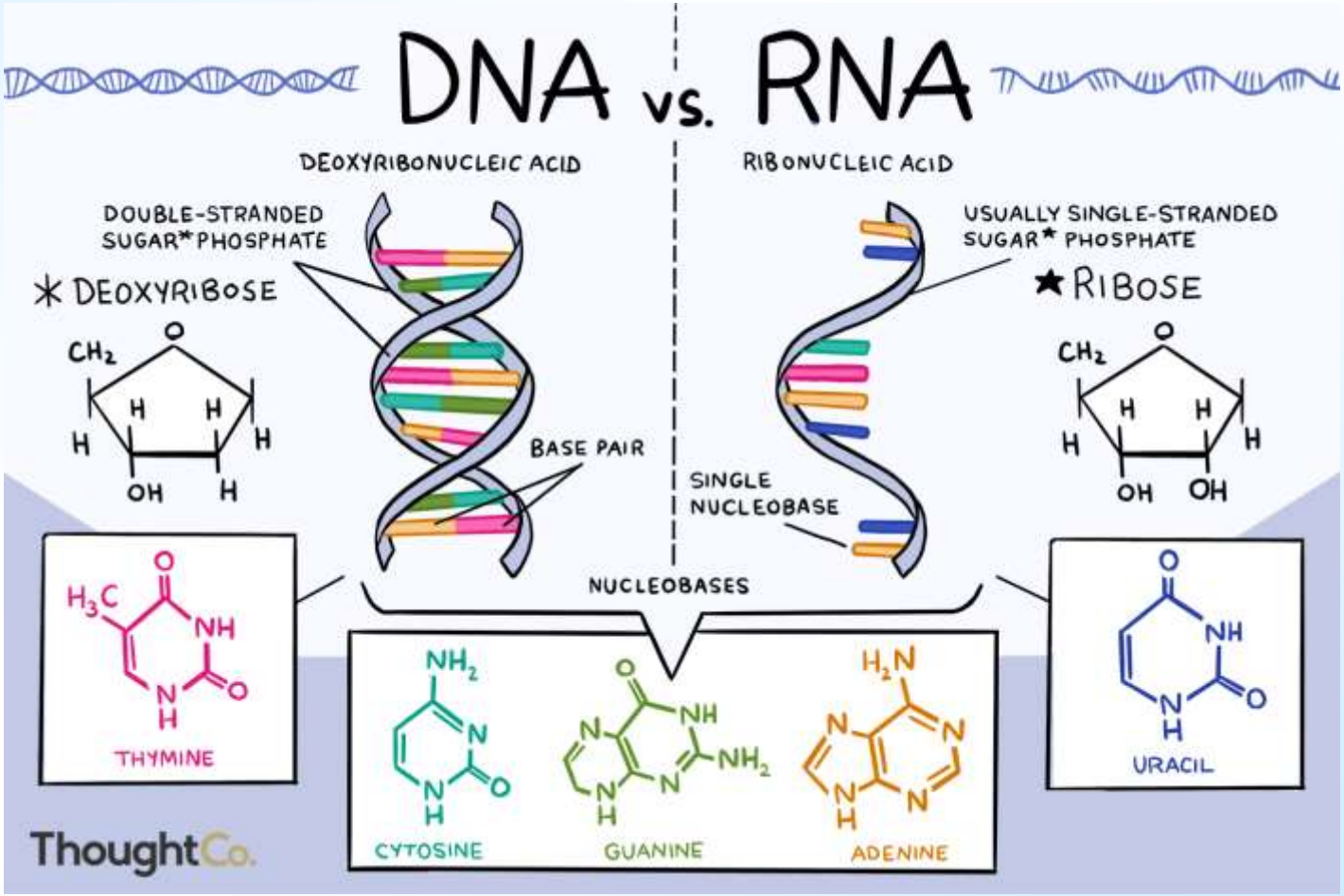


Model of the secondary and spatial structure of DNA

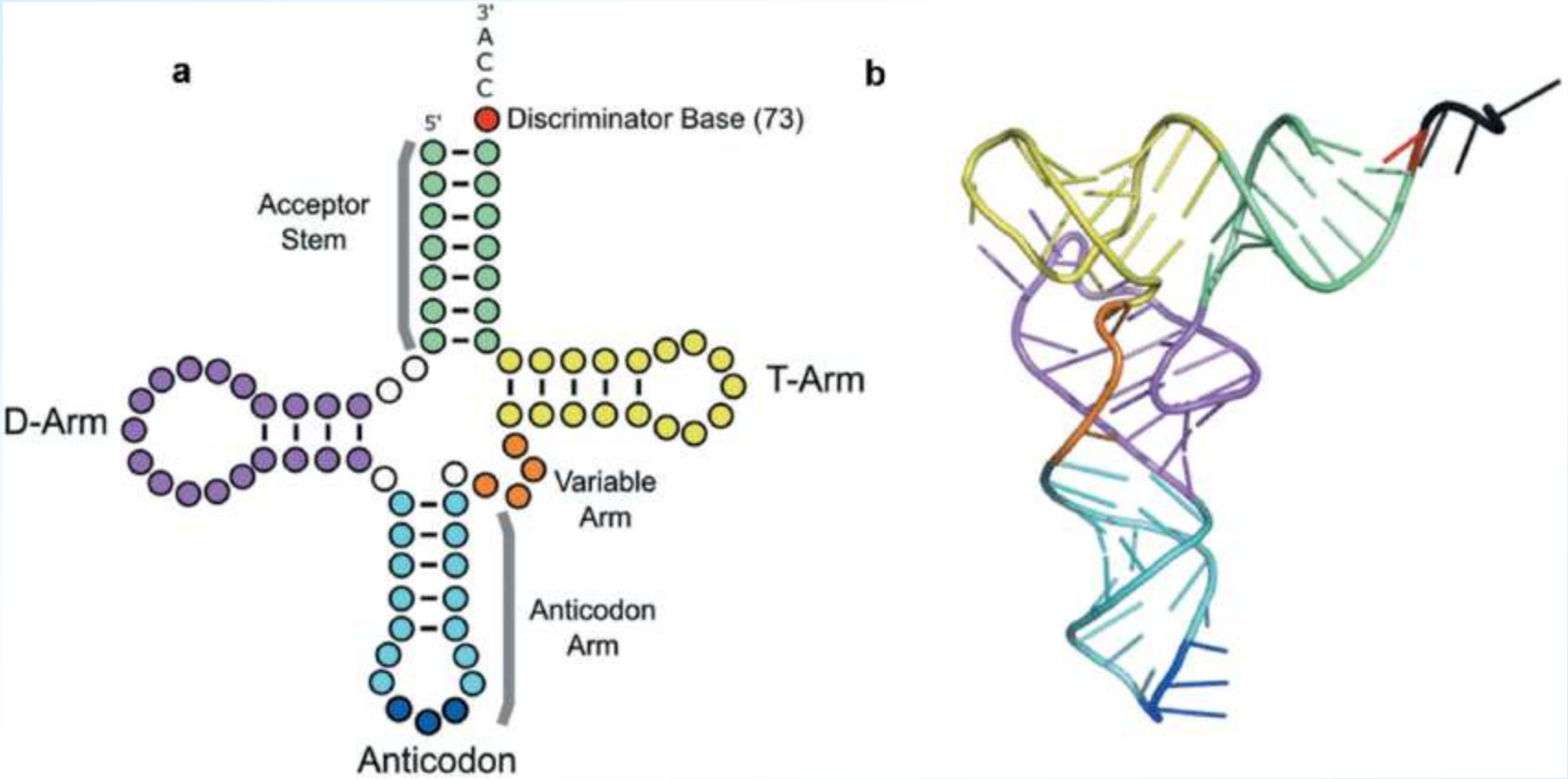
RNA structure and function

RNA - ribonucleic acid, has much in common with the structure of DNA, but differs in a number of features:

- RNA carbohydrate, to which purine or pyrimidine bases and phosphate groups are attached, is **ribose**;
- RNA, as well as DNA, contains nitrogenous bases adenine, guanine and cytosine. But instead of thymine, RNA contains **uracil**;
- unlike double-stranded DNA, RNA is a single-stranded molecule.



RNA is of different types: informational or matrix (mRNA), transport (tRNA), ribosomal (rRNA), the nucleus of eukaryotic cells contains a heterogeneous nuclear (hnRNA)

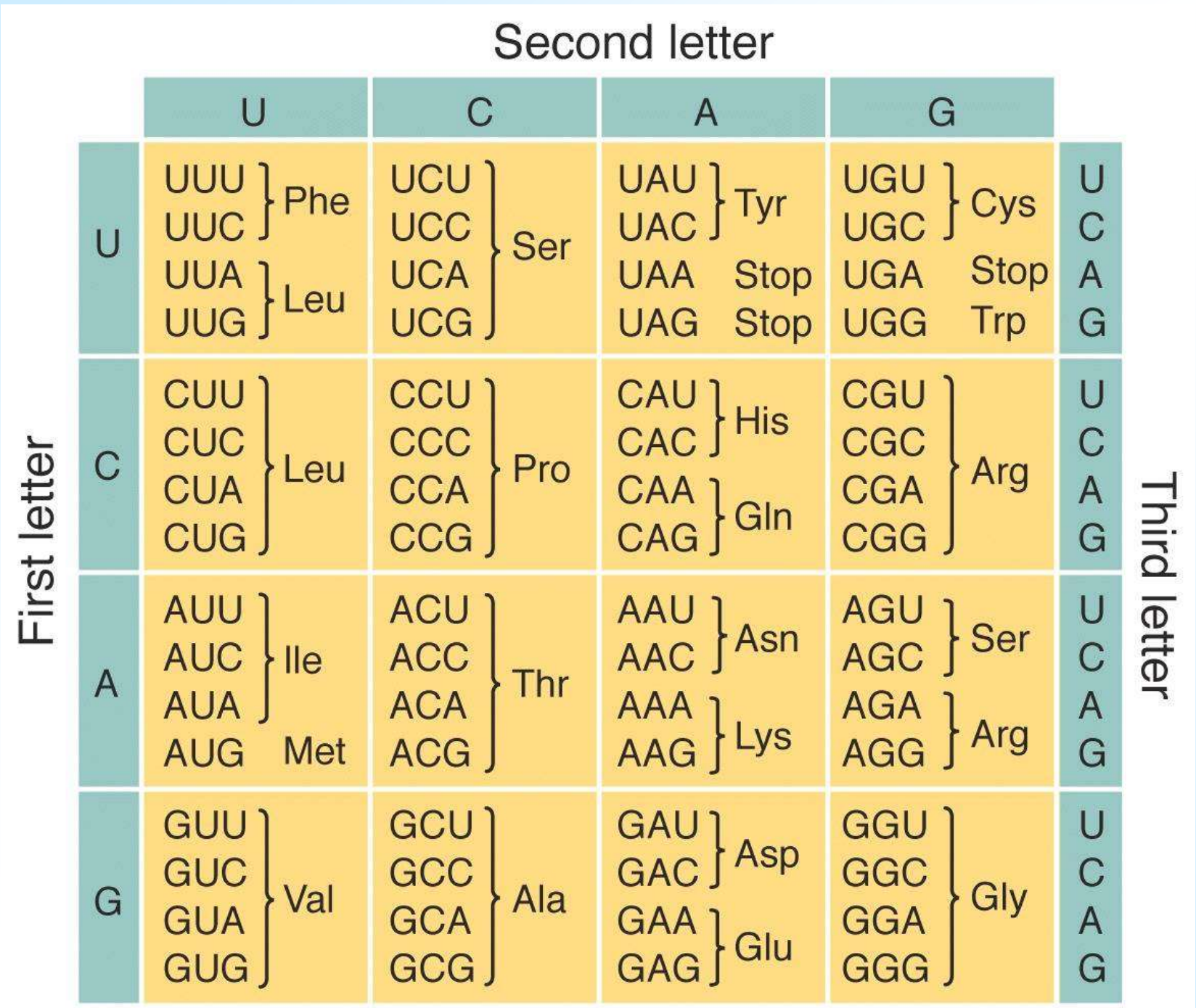


The genetic code

The genetic code is a unified system for recording hereditary information in nucleic acid molecules in the form of a sequence of nucleotides.

Properties of the genetic code:

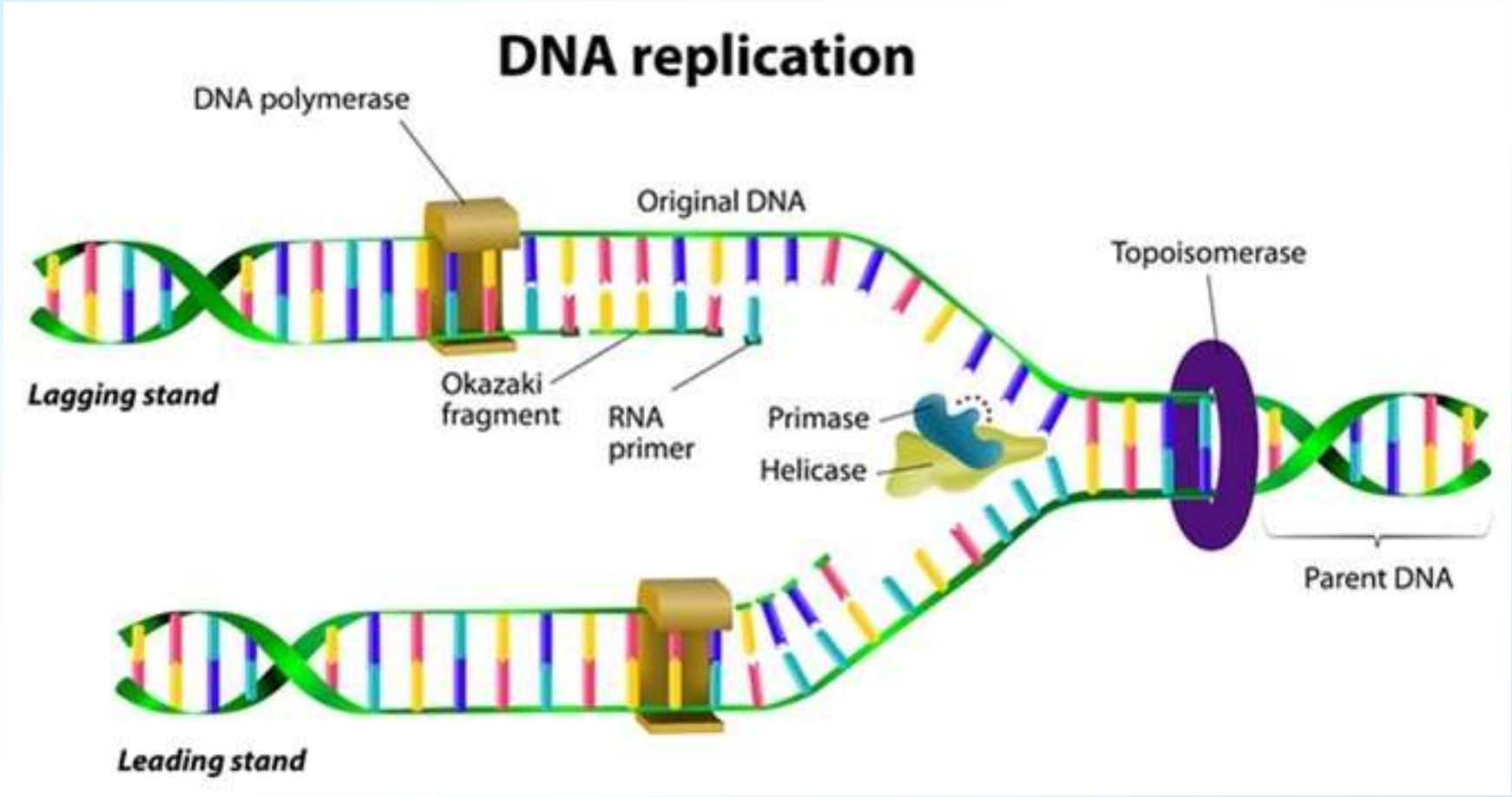
- The genetic code is triplet. Triplet (codon) - a sequence of three nucleotides that encodes one amino acid;
- The degeneracy of the genetic code is due to the fact that one amino acid can be encoded by several triplets (20 amino acids, and 64 triplets),
- Unambiguity - each given codon corresponds to one and only one definite amino acid.
- The code does not overlap, i.e. in the ABCDEFGH base sequence, the first three bases, ABC, encode amino acid 1, DEF - amino acid 2, and so on. There are no commas in the code, i.e. there are no signs separating one codon from another.
- The genetic code is universal, i.e. all information in nuclear genes for all organisms with different levels of organization (for example, butterfly, chamomile, cancer, frog, boa constrictor, eagle, man) is encoded in the same way.


Second letter
U
C
A
G
First letter
Third letter
U
UUU Phe
UUC
UUA Leu
UUG
UCU Ser
UCC
UCA
UCG
UAU Tyr
UAC
UAA Stop
UAG Stop
UGU Cys
UGC
UGA Stop
UGG Trp
C
CUU Leu
CUC
CUA
CUG
CCU Pro
CCC
CCA
CCG
CAU His
CAC
CAA Gln
CAG
CGU Arg
CGC
CGA
CGG
A
AUU Ile
AUC
AUA
AUG Met
ACU Thr
ACC
ACA
ACG
AAU Asn
AAC
AAA Lys
AAG
AGU Ser
AGC
AGA Arg
AGG
G
GUU Val
GUC
GUA
GUG
GCU Ala
GCC
GCA
GCG
GAU Asp
GAC
GAA Glu
GAG
GGU Gly
GGC
GGA
GGG

MATRIX PROCESSES IN THE CELL

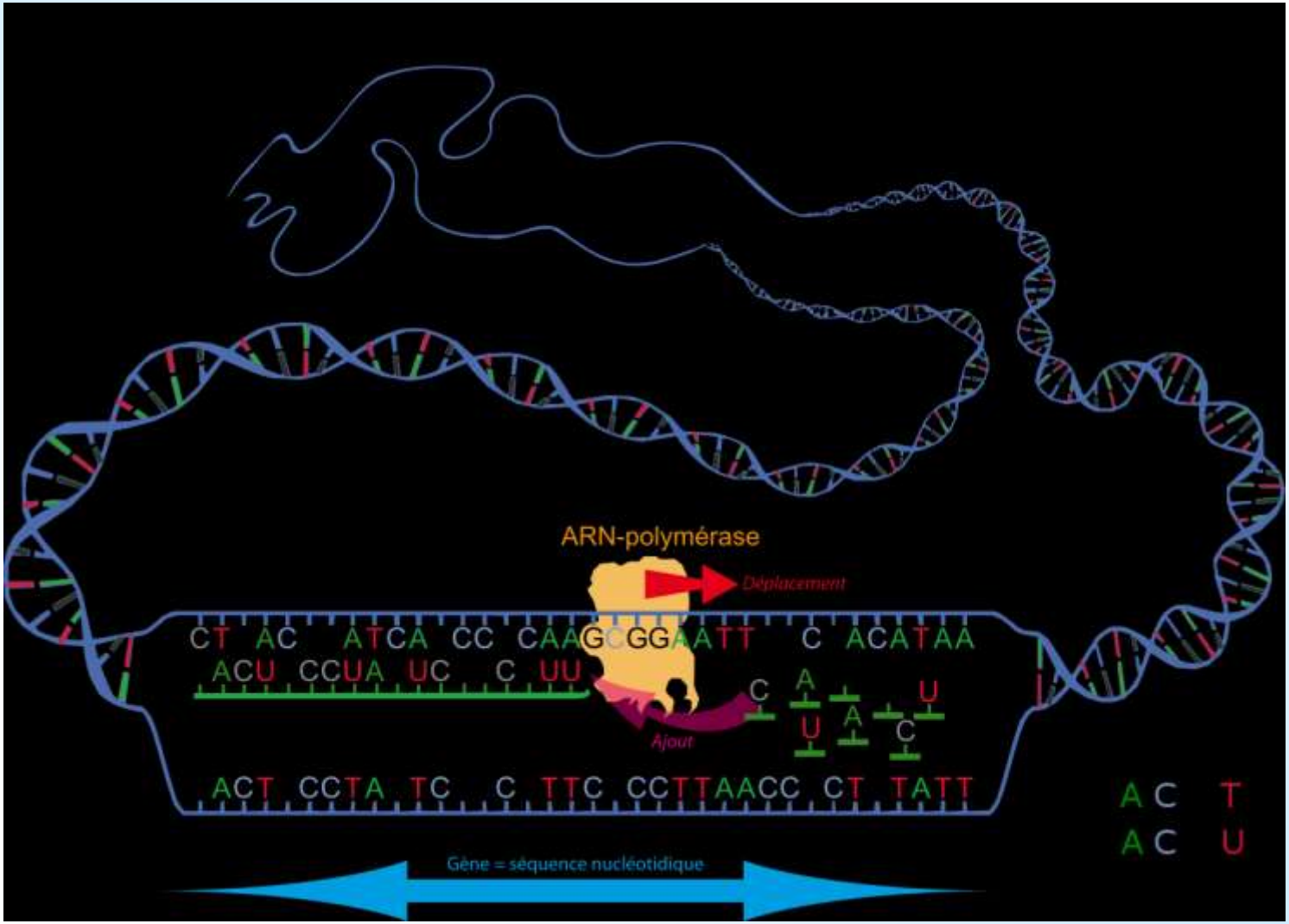
Replication

Replication or reduplication (doubling) of DNA is called its synthesis.



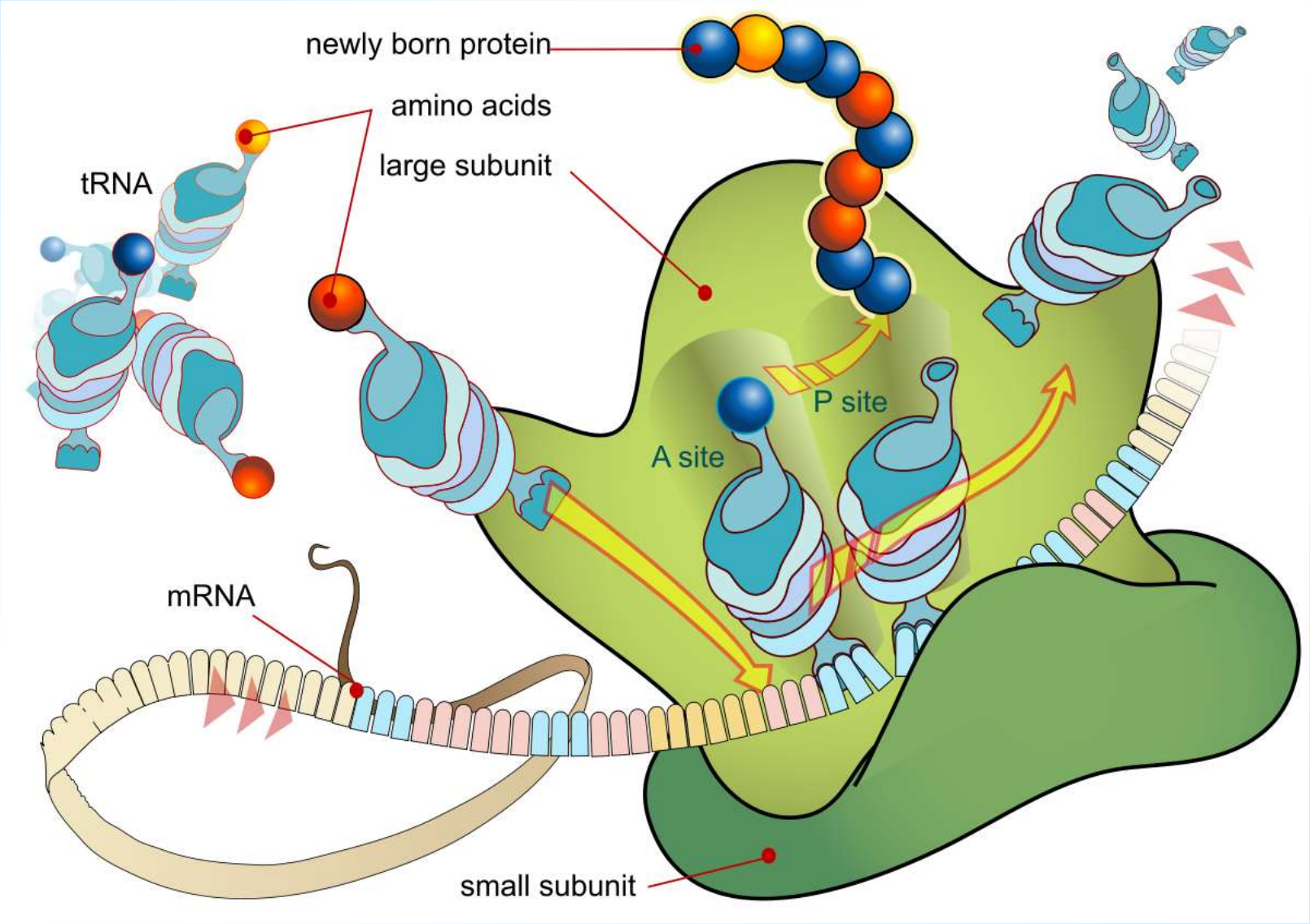
Protein biosynthesis. Transcription

Transcription (rewriting) is the synthesis of mRNA (the primary product of the gene) on the DNA template, which is carried out in the nucleus on a sense DNA strand in a despiralized state.

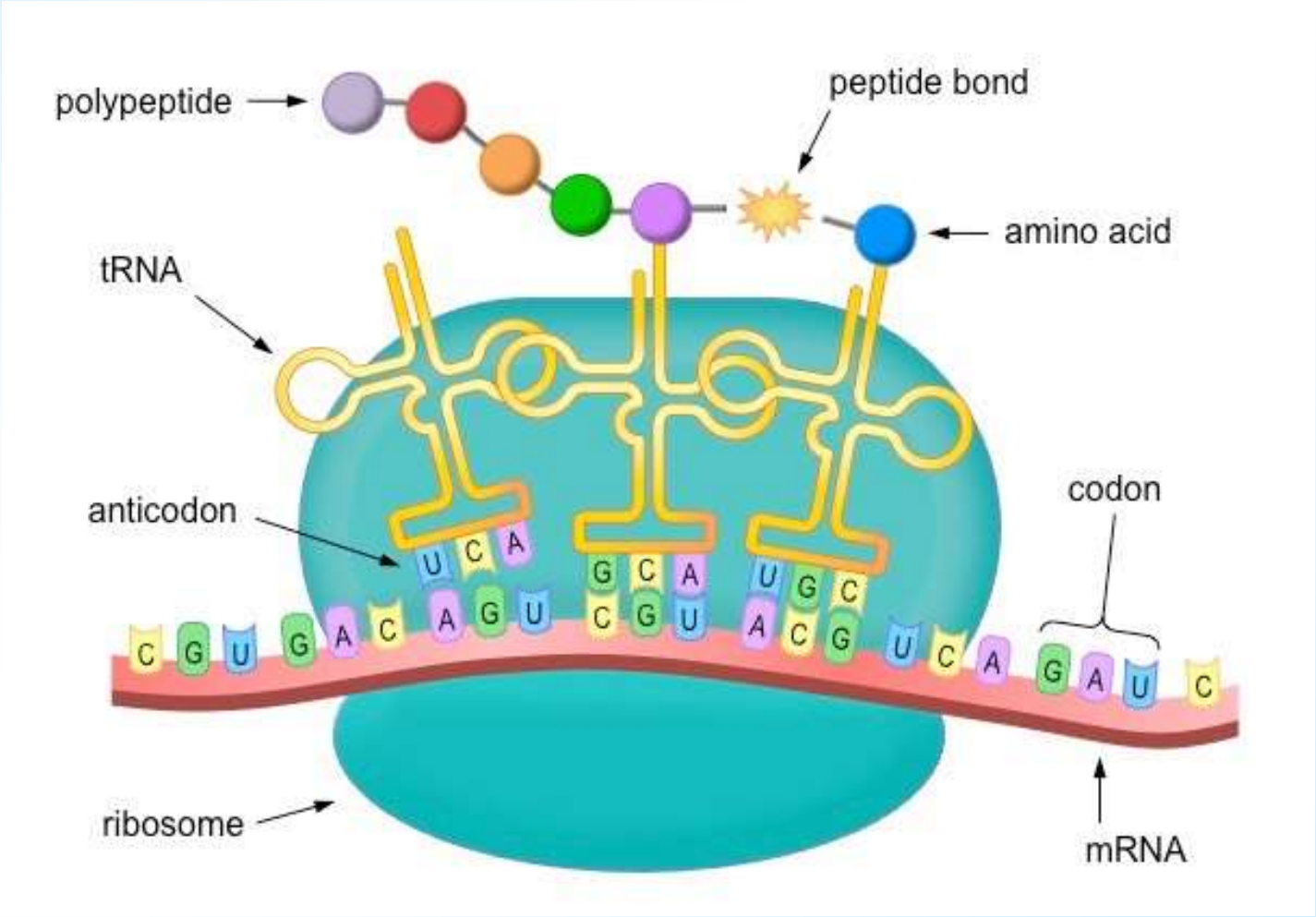


Protein biosynthesis. Translation

Translation (translation) is the process of translating mRNA genetic information into a polypeptide structure.



Mature messenger RNA enters the cytoplasm, where the translation process takes place - decoding of mRNA into the amino acid sequence of the protein. The decoding process is carried out in the direction from 5 '→ 3' and takes place in the ribosomes. The complex of mRNA and ribosomes is called a polysome. Like transcription, the translation mechanism consists of three stages: initiation, elongation and termination



PROTEIN BIOSYNTHESIS