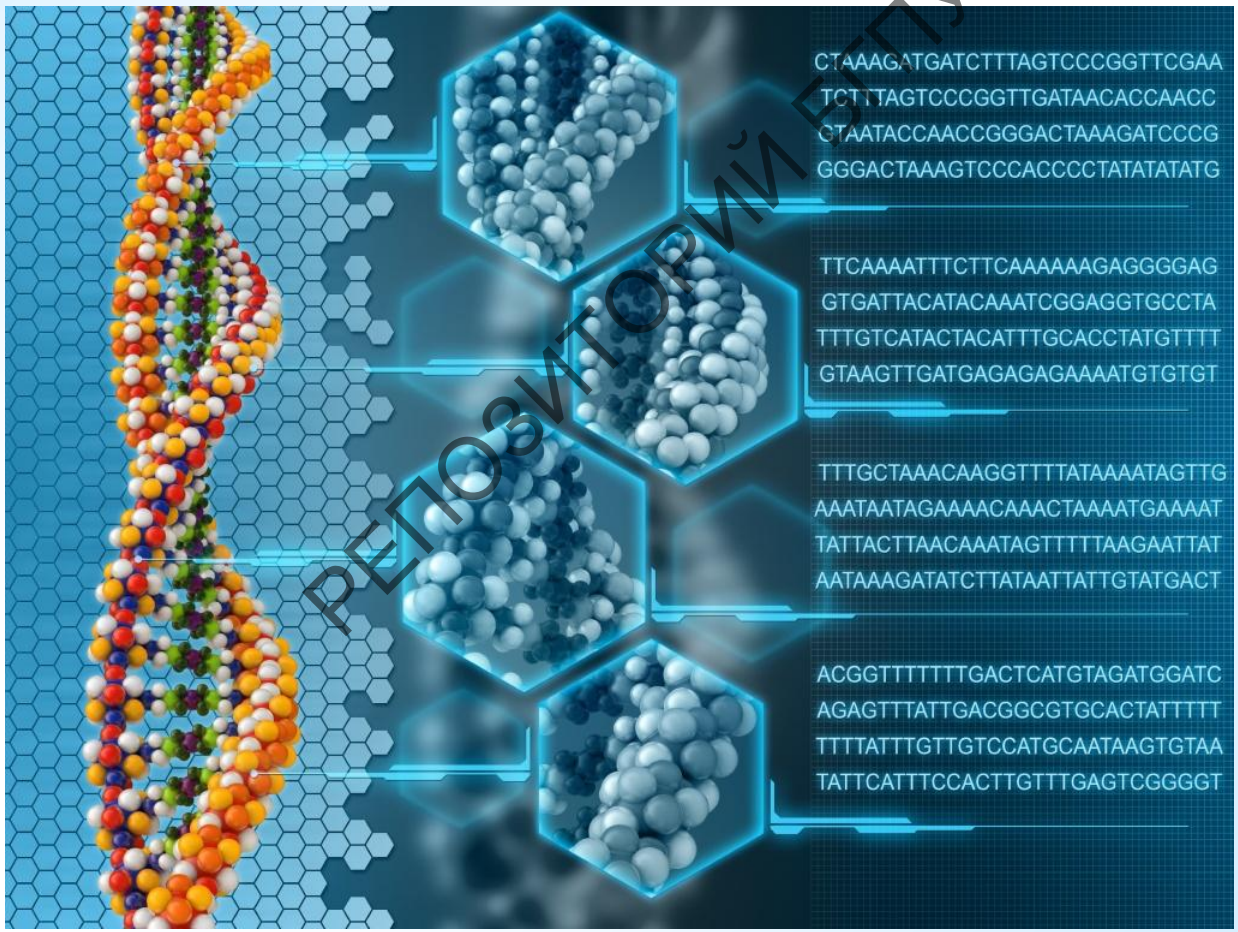
«Biomedical foundations of special pedagogy and psychology: the basics of human genetics»

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.

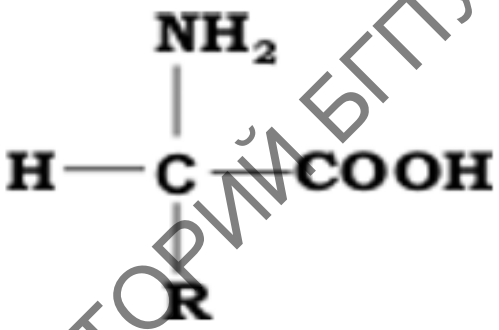
Heredity - the property of living organisms to maintain similar characteristics from generation to generation, to ensure functional continuity, as well as a certain pattern of individual development (ontogenesis) in certain environmental conditions.

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;

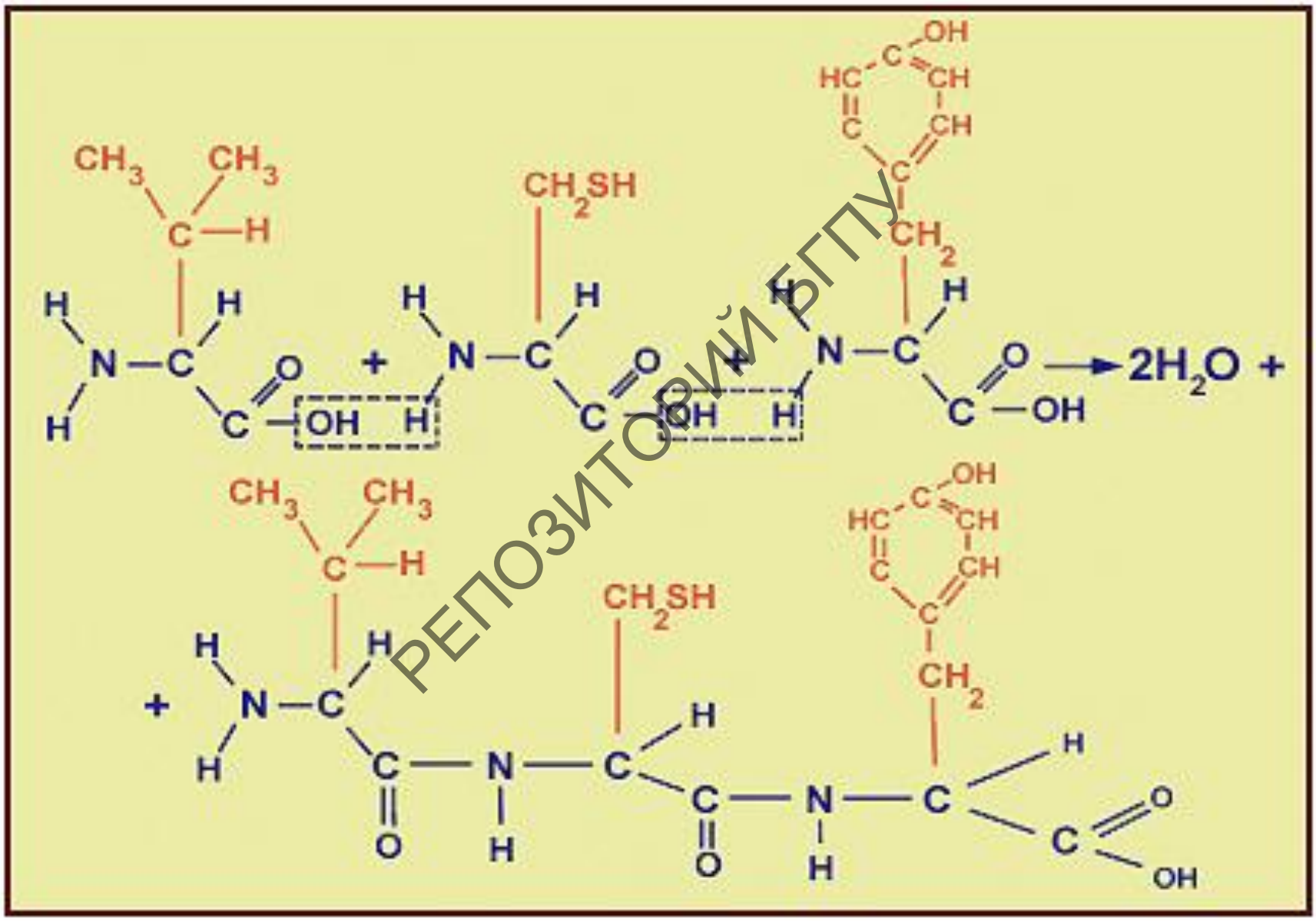
Ontogenetic - changes in the individual development of the body or with the differentiation of cells;
Geographical - the formation of features in organisms of one species under the influence of spatial and geographical factors (forms, races, subspecies, etc.)
Qualitative - a fundamental change in properties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

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

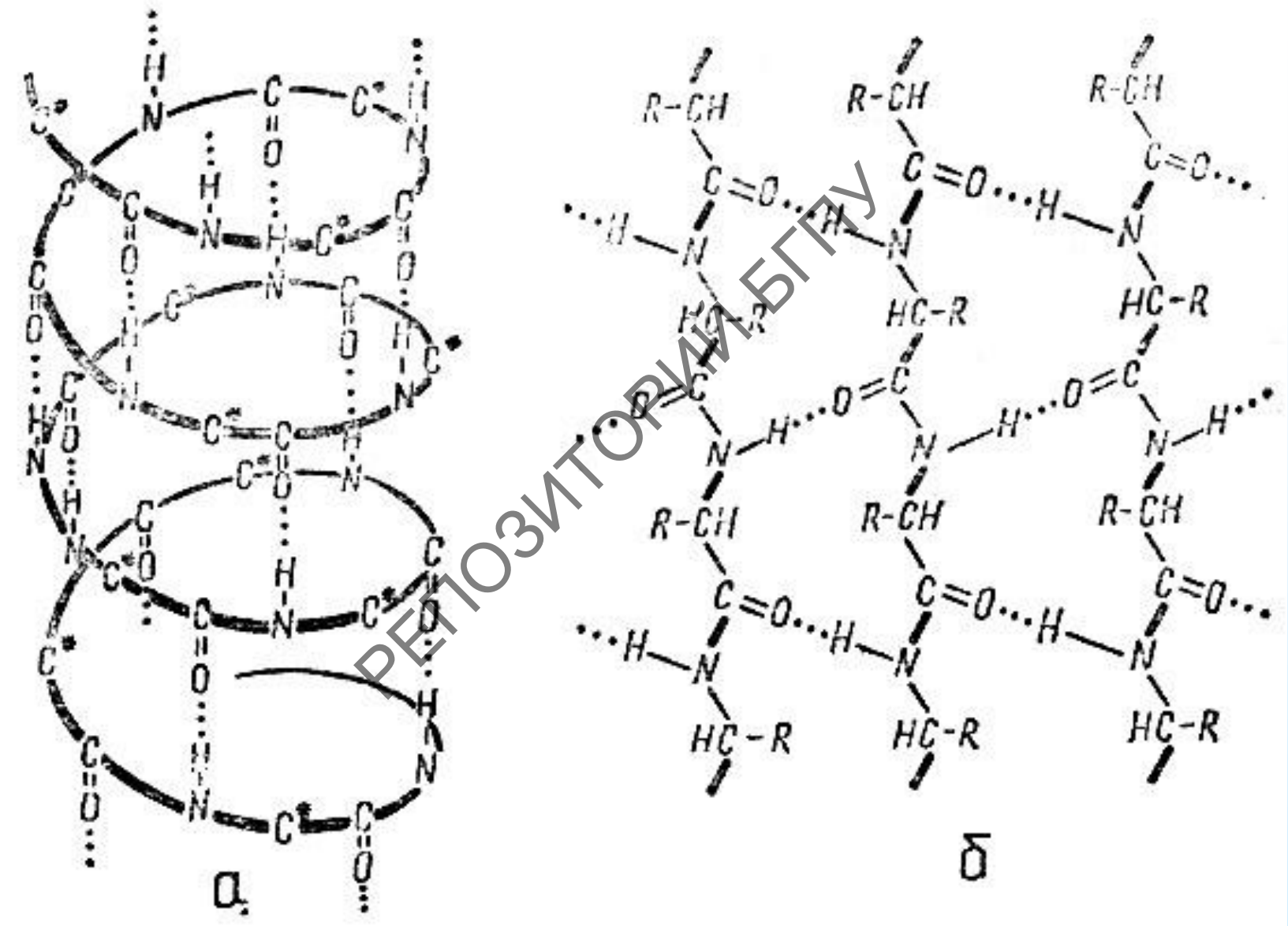


NH_2 – amino group;
COOH – carboxyl group;
H – hydrogen atom;
radical R – side group.

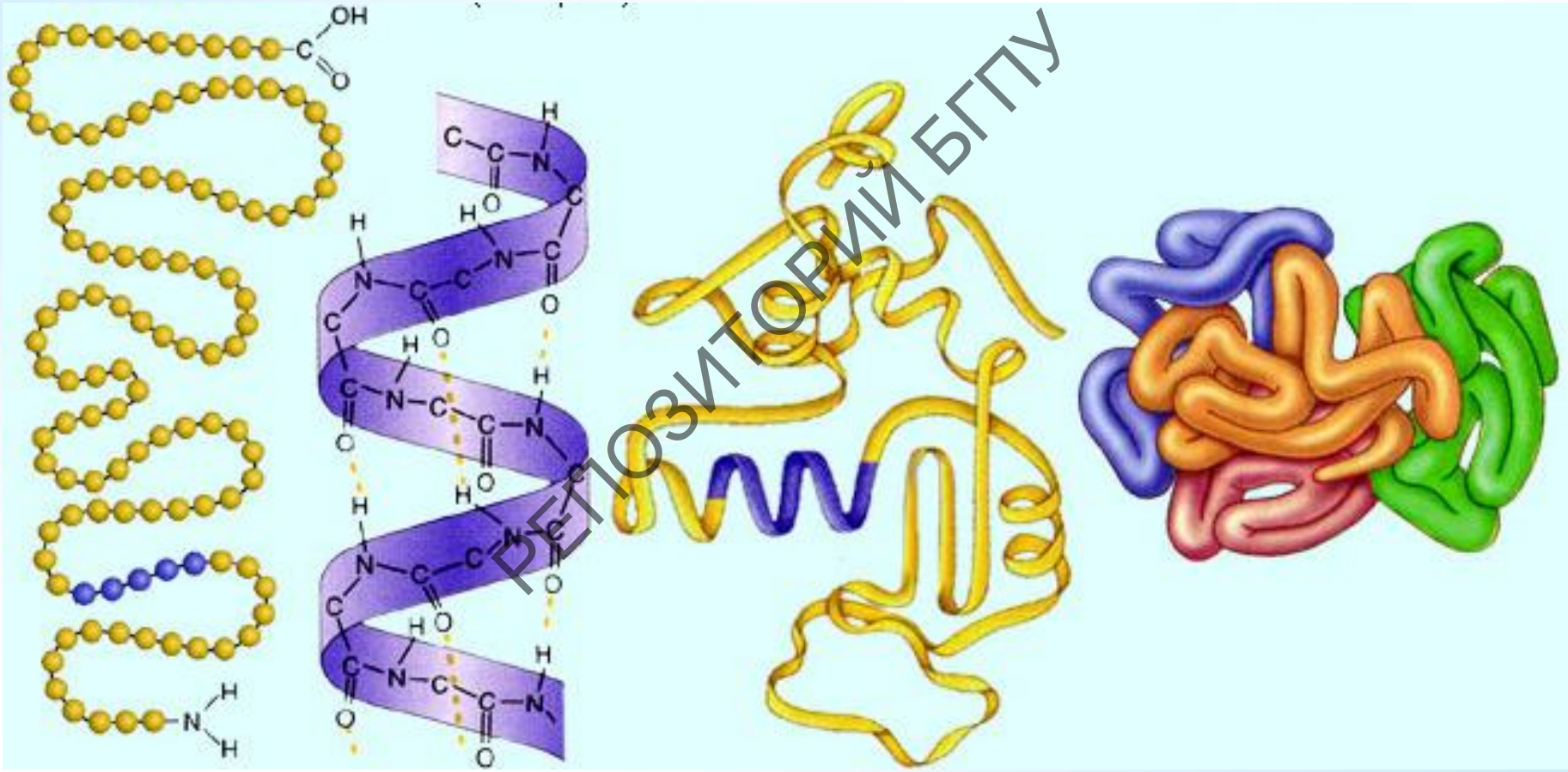
Primary (linear) protein structure



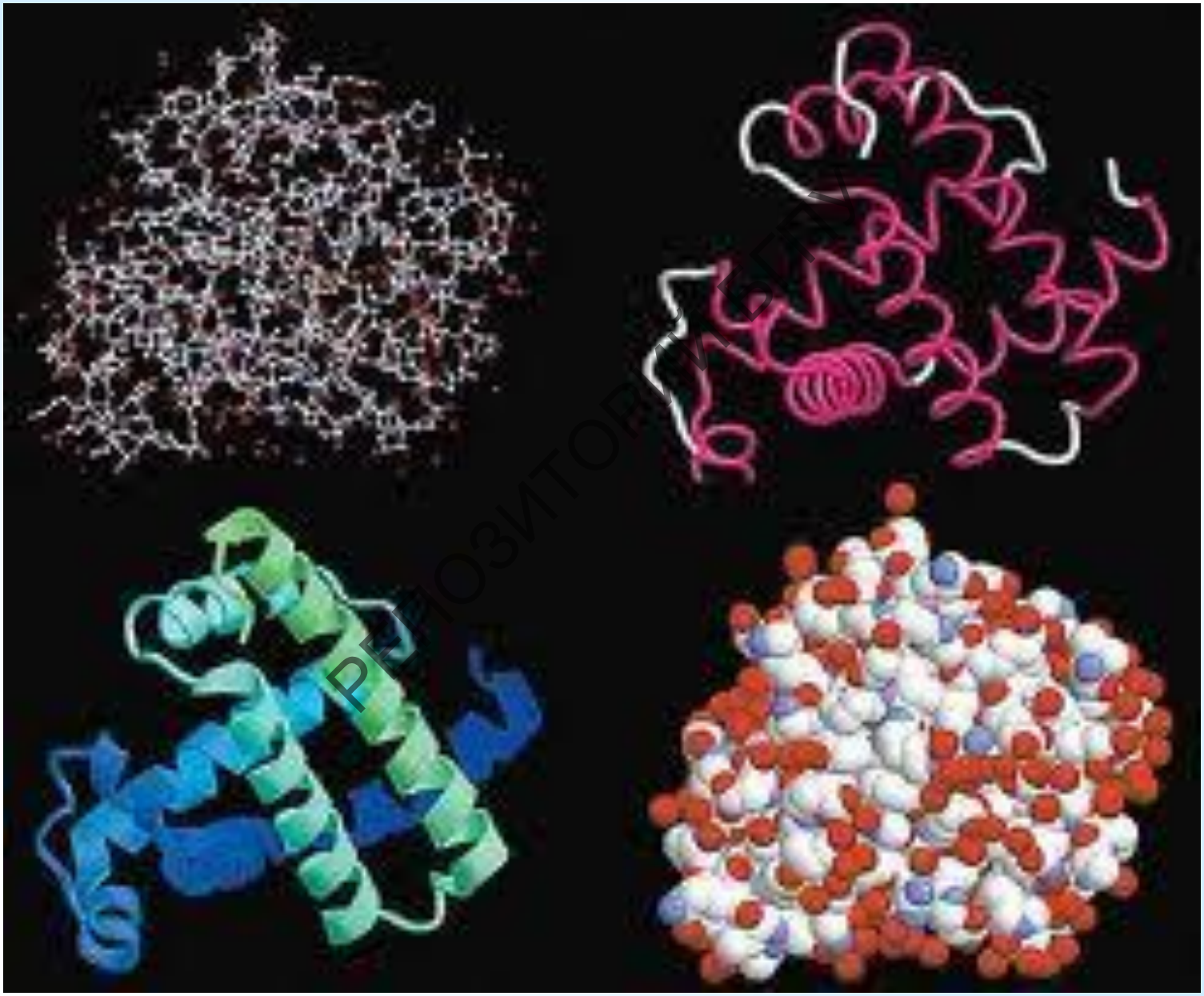
Secondary (helical) protein structure



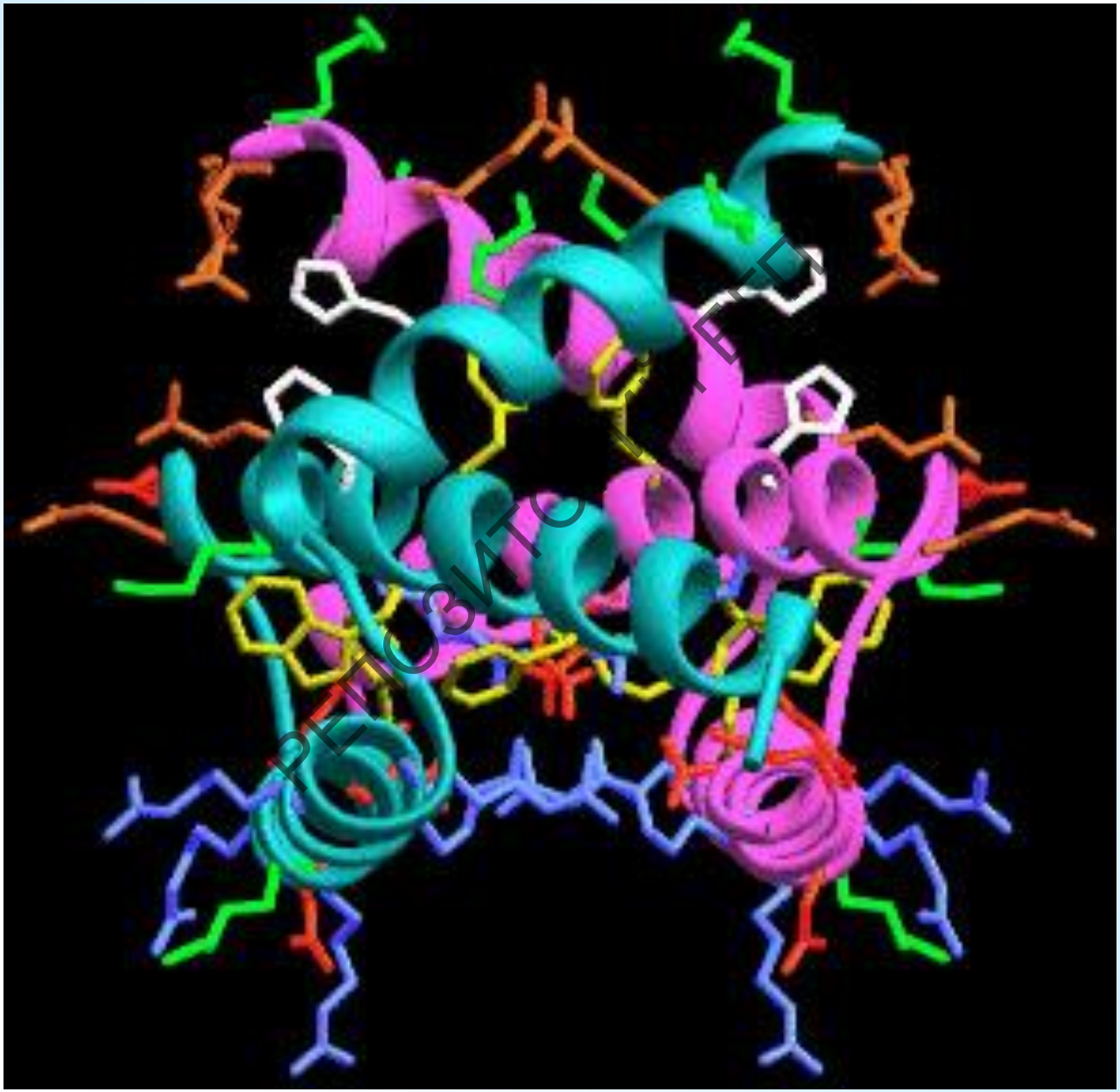
Tertiary (globular) protein structure,



Quaternary protein structure

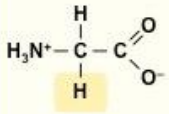


Quaternary protein structure

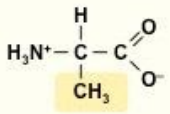


Classification of amino acids by the nature of the side groups

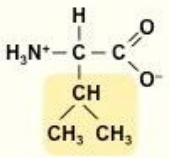
NON-POLAR



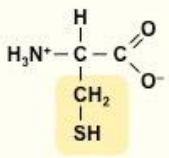
Glycine
(Gly / G)



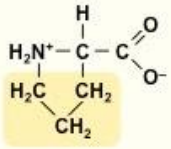
Alanine
(Ala / A)



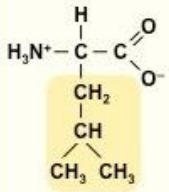
Valine
(Val / V)



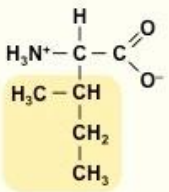
Cysteine
(Cys / C)



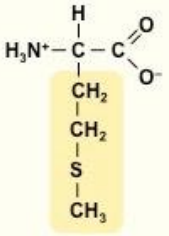
Proline
(Pro / P)



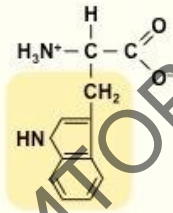
Leucine
(Leu / L)



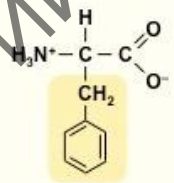
Isoleucine
(Ile / I)



Methionine
(Met / M)

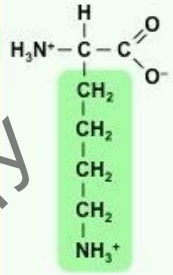


Tryptophan
(Trp / W)

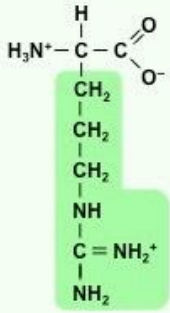


Phenylalanine
(Phe / F)

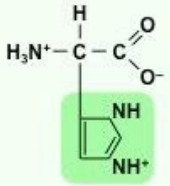
+ CHARGE



Lysine
(Lys / K)

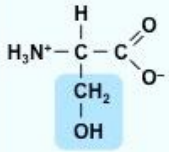


Arginine
(Arg / R)

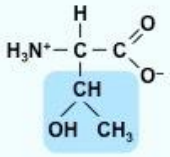


Histidine
(His / H)

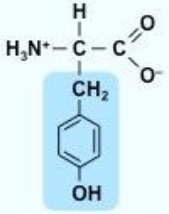
POLAR



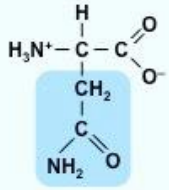
Serine
(Ser / S)



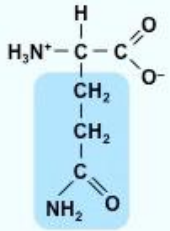
Threonine
(Thr / T)



Tyrosine
(Tyr / Y)

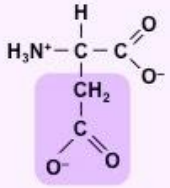


Asparagine
(Asn / N)

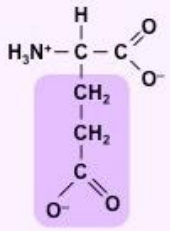


Glutamine
(Gln / Q)

– CHARGE



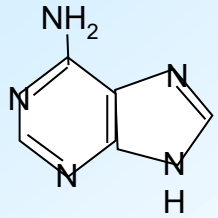
Aspartic Acid
(Asp / D)



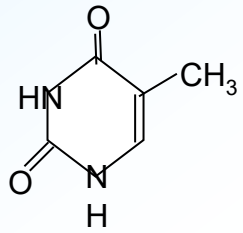
Glutamic Acid
(Glu / E)

DNA structure and function

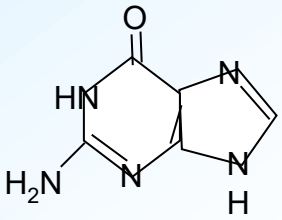
DNA - deoxyribonucleic acid - a biological macromolecule, a carrier of genetic information in all eukaryotic cells.



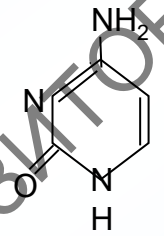
Adenine



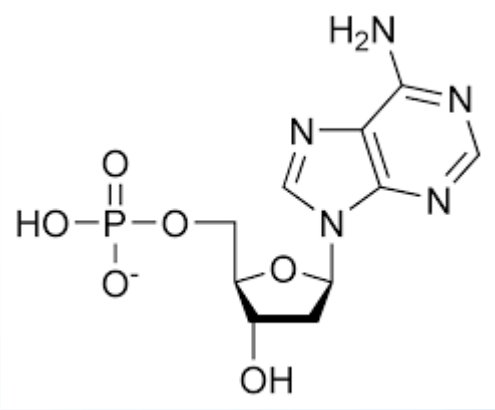
Timin



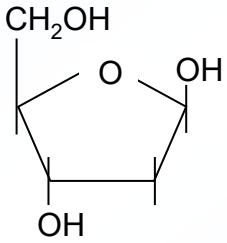
Guanine



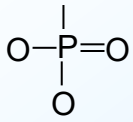
Cytosine



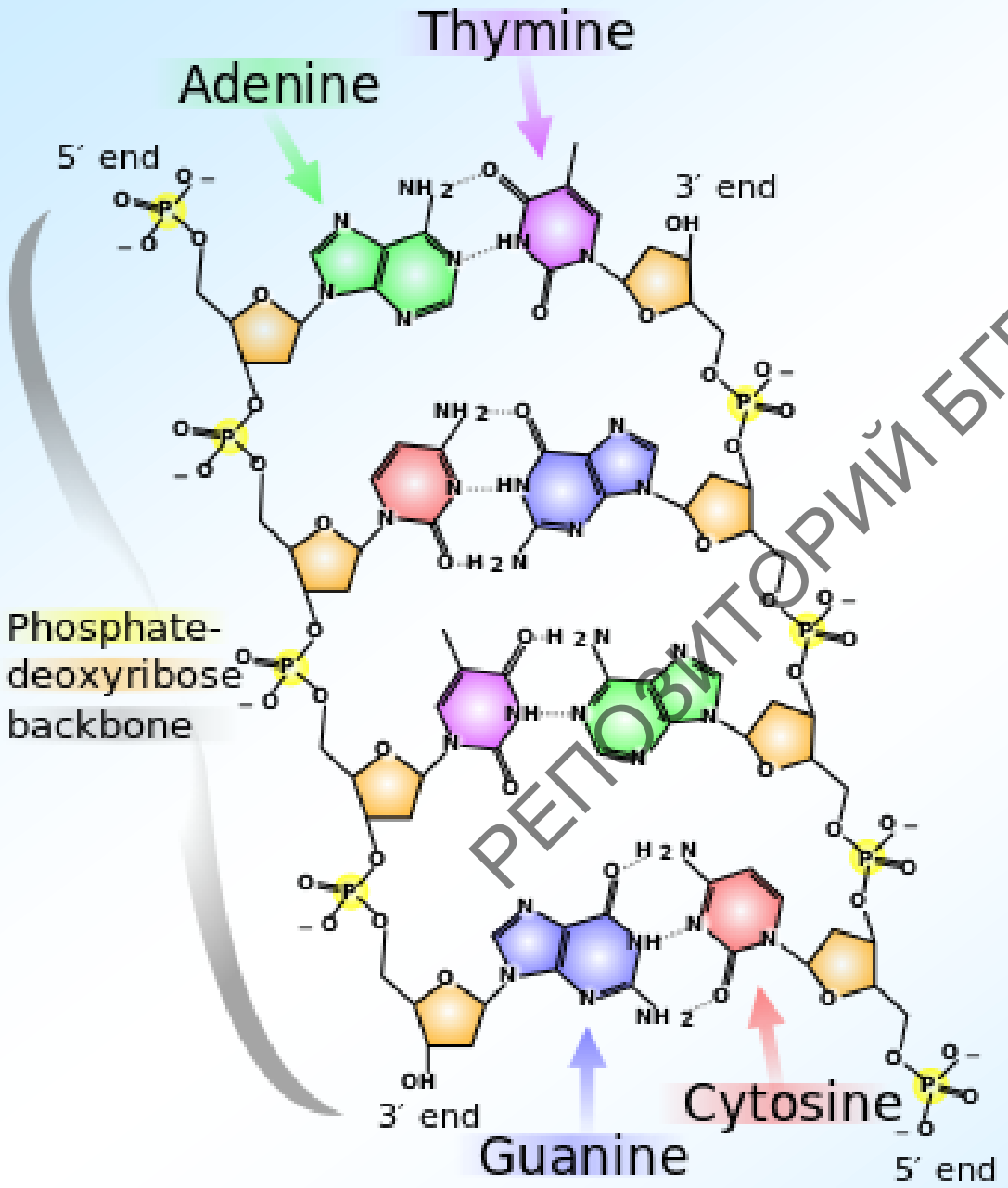
DNA nucleotide



Deoxyribose



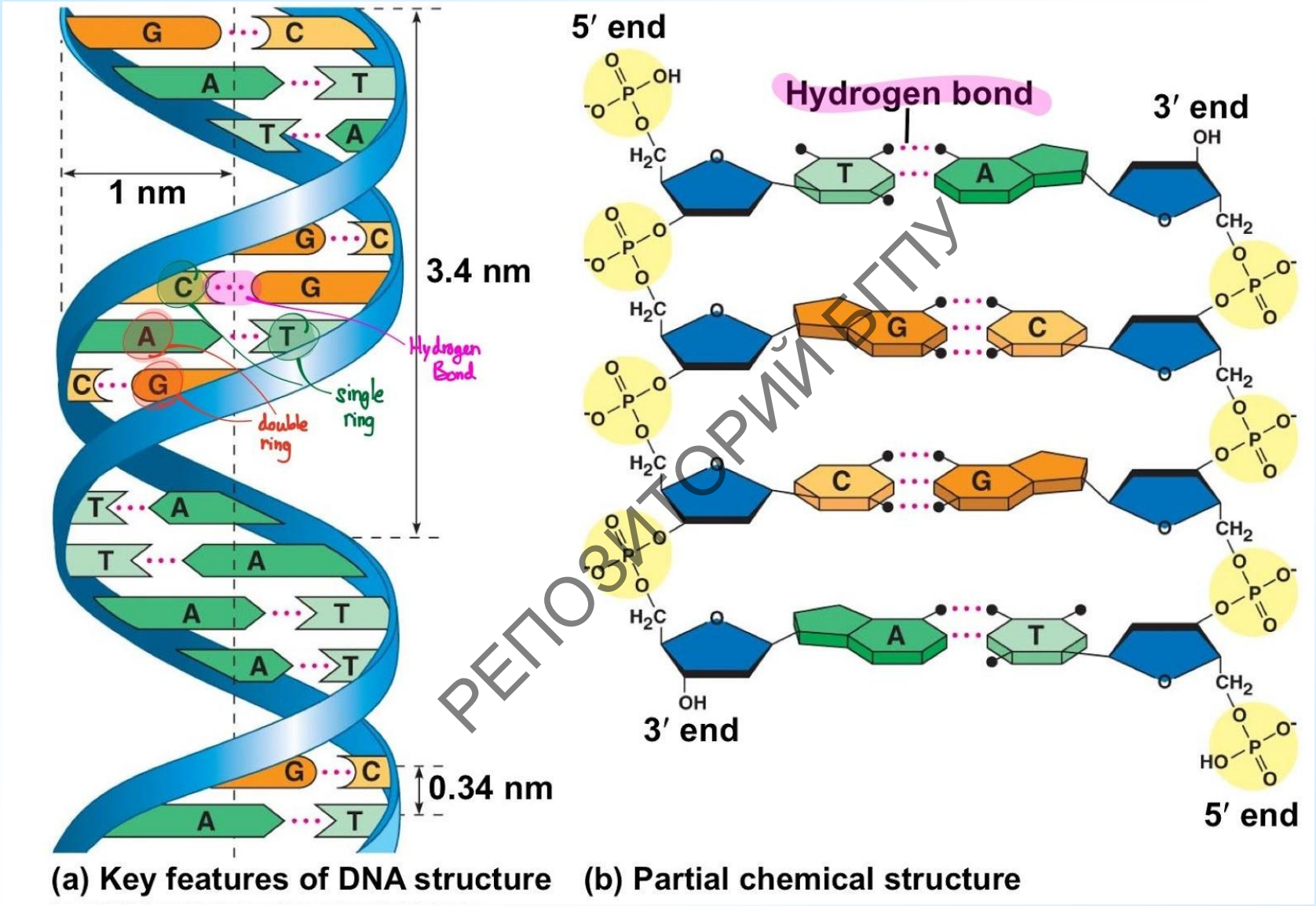
Phosphoric Acid Residue



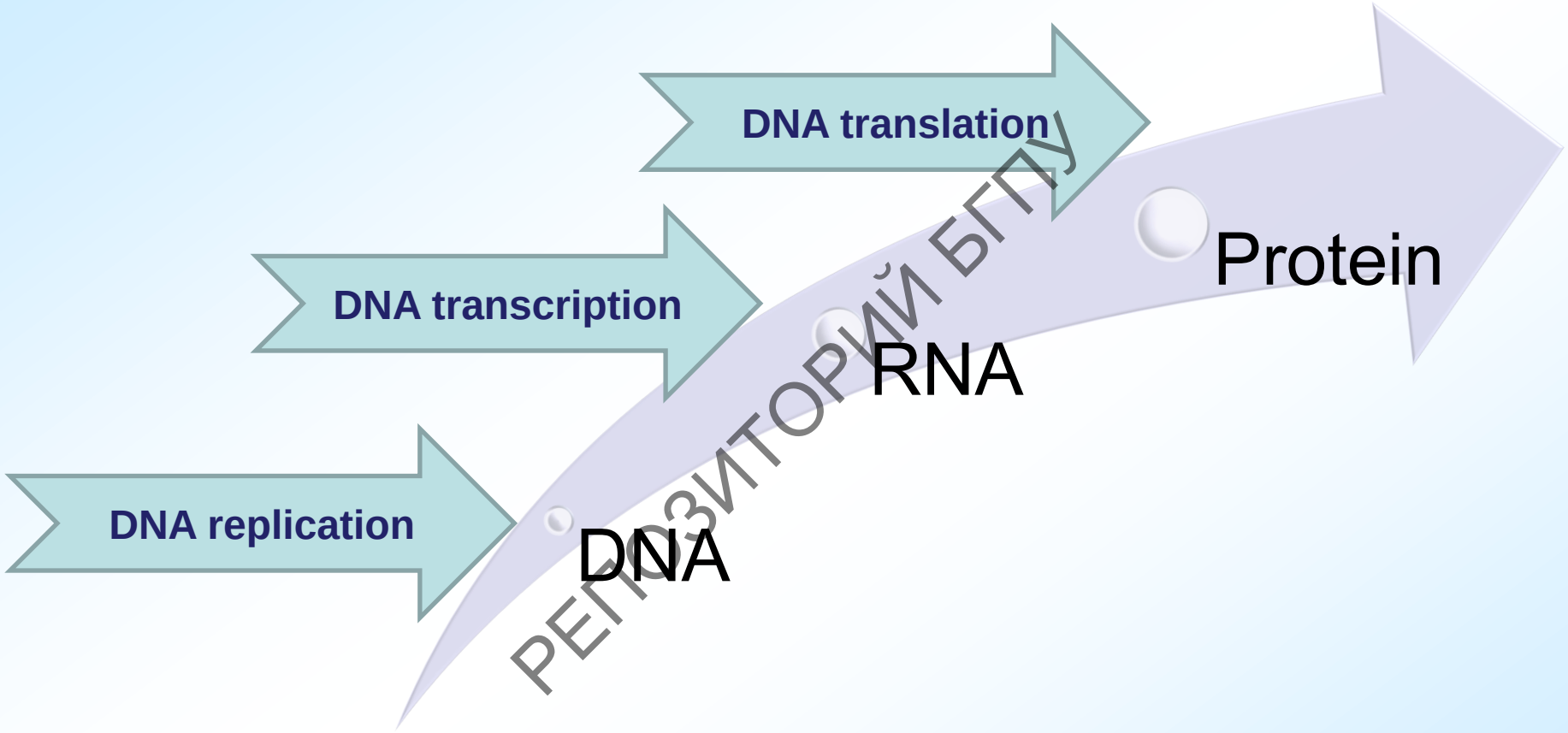
The relationship between the number of different bases in DNA was established by Charguff in 1949 and played an important role in the construction of the double helix. Charguff found that the amount of adenine in DNA of various origins is equal to the amount of thymine, and the amount of guanine is equal to the amount of cytosine. This pattern was called the “Chargaff rule” and is expressed as follows: A = T; G = C или

$$\frac{A+G}{C+T}=1$$

Model of the structure of DNA



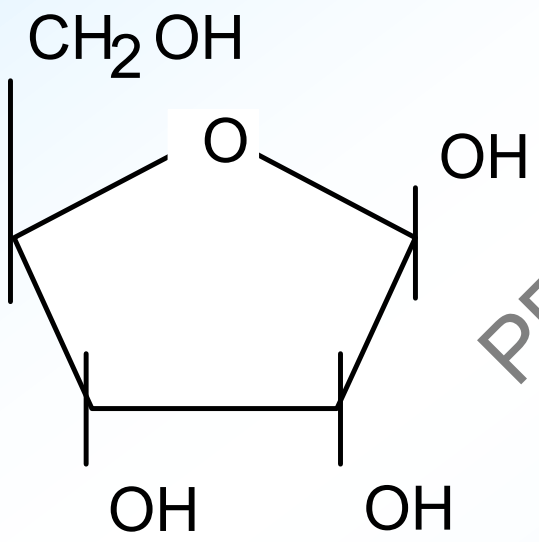
Scheme of the relationship of DNA, RNA and protein



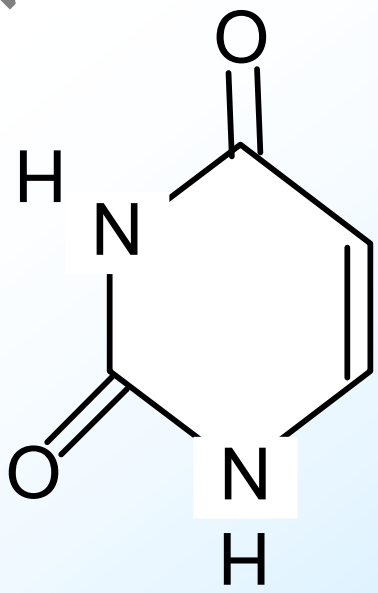
The structure and functions of RNA

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

- ✓ RNA, like 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.
- ✓ the RNA carbohydrate to which purine or pyrimidine bases and phosphate groups are attached is ribose;



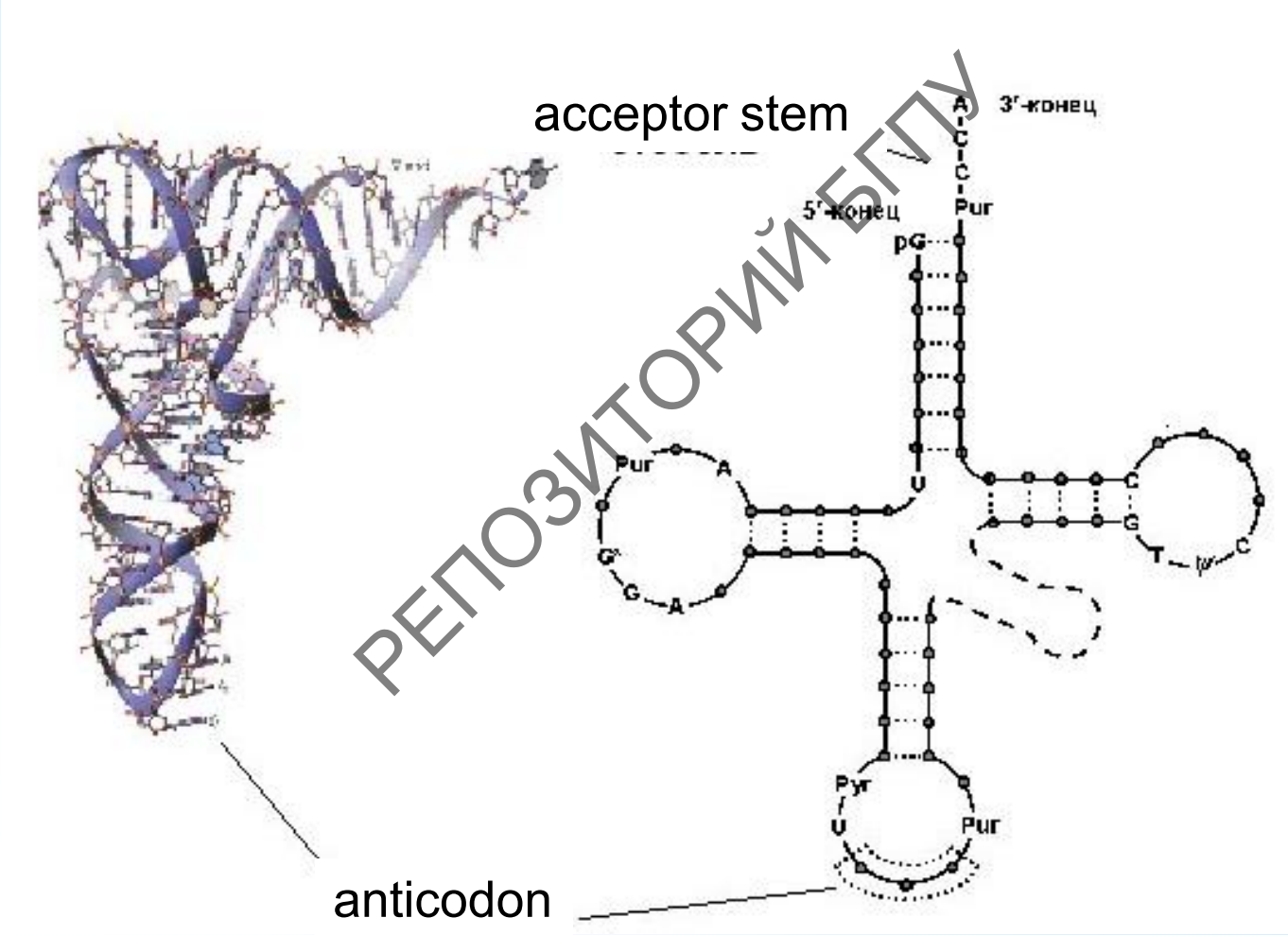
Ribose



Uracil

There are different types of **RNA**:

- ✓ informational or template (mRNA),
- ✓ transport (tRNA),
- ✓ ribosomal (rRNA),
- ✓ heterogeneous nuclear (hRNA)



Genetic code

The genetic code is a single system for recording hereditary information in nucleic acid molecules in the form of a nucleotide sequence

Genetic Code Properties :

The genetic code is triplet. Triplet (codon) - a sequence of three nucleotides encoding one amino acid;

The degeneracy of the genetic code is due to the fact that one amino acid can be encoded by several triplets (amino acids 20, and triplets –64),

Unambiguity - each given codon corresponds to one and only one specific amino acid.

The code does not overlap, i.e. in the base sequence ABCDEFGIJKLMN 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, human) is encoded identically.

Genetic Code Table

1st letter	Second Letter: U		Second Letter: C		Second Letter: A		Second Letter: G		3rd letter
U	UUU	Phe	UCU	Ser	UAU	Tyr	UGU	Cys	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu	UCA		UAA	Stop	UGA	Stop	A
	UUG		UCG		UAG	Stop	UGG	Trp	G
C	CUU	Leu	CCU	Pro	CAU	His	CGU	Arg	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Gln	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Ile	ACU	Thr	AAU	Asn	AGU	Ser	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lys	AGA	Arg	A
	AUG	Met	ACG		AAG		AGG		G
G	GUU	Val	GCU	Ala	GAU	Asp	GGU	Gly	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glu	GGA		A
	GUG		GCG		GAG		GGG		G

Genetic Code Table

UUU, UUC — phenylalanine UUA, UUG — leucine	UCU, UCC, UCA, UCG — serine	UAU, UAC — tyrosine UAA, UAG — stop	UGU, UGC — cysteine UGA — stop UGG — tryptophan
CUU, CUC, CUA, CUG — leucine	CCU, CCC, CCA, CCG — proline	CAU, CAC — histidine CAA, CAG — glutamine	CGU, CGC, CGA, CGG — arginine
AUU, AUC, AUA — isoleucine AUG — methionine	ACU, ACC, ACA, ACG — threonine	AAU, AAC — asparagine AAA, AAG — lysine	AGU, AGC — serine AGA, AGG — arginine
GUU, GUC, GUA, GUG — valine	GCU, GCC, GCA, GCG — alanine	GAU, GAC — aspartic acid GAA, GAG — glutamic acid	GGU, GGC, GGA, GGG — glycine

Genetic Code Table

SECOND POSITION

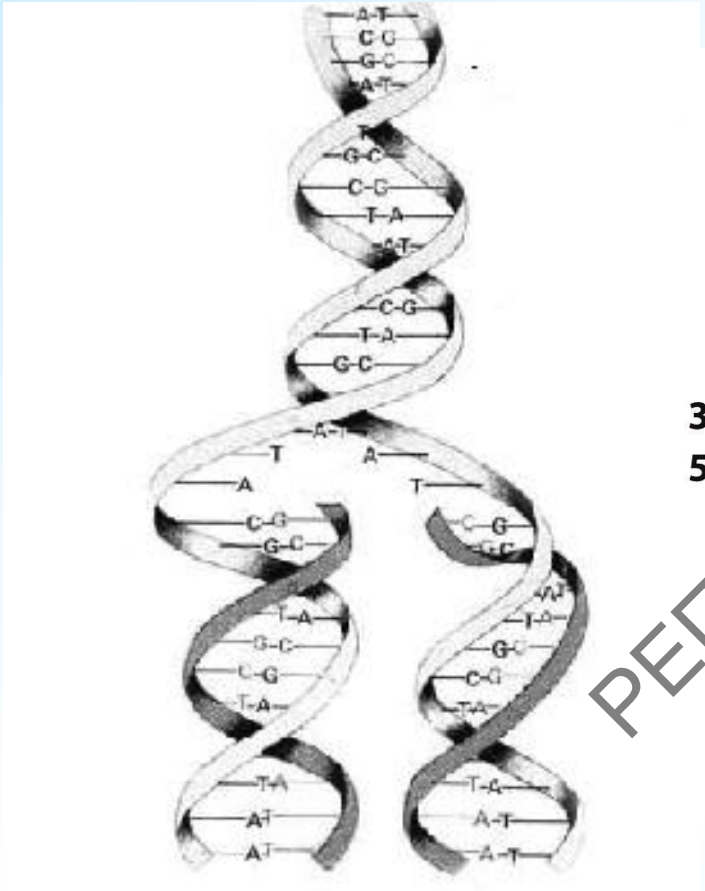
FIRST POSITION	U	C	A	G	THIRD POSITION
U	phenyl-alanine	serine	tyrosine	cysteine	U
					C
	leucine		stop	stop	A
			stop	tryptophan	G
C	leucine	proline	histidine	arginine	U
					C
			glutamine		A
					G
A	isoleucine	threonine	asparagine	serine	U
					C
	* methionine		lysine	arginine	A
					G
G	valine	alanine	aspartic acid	glycine	U
					C
			glutamic acid		A
					G

* and start

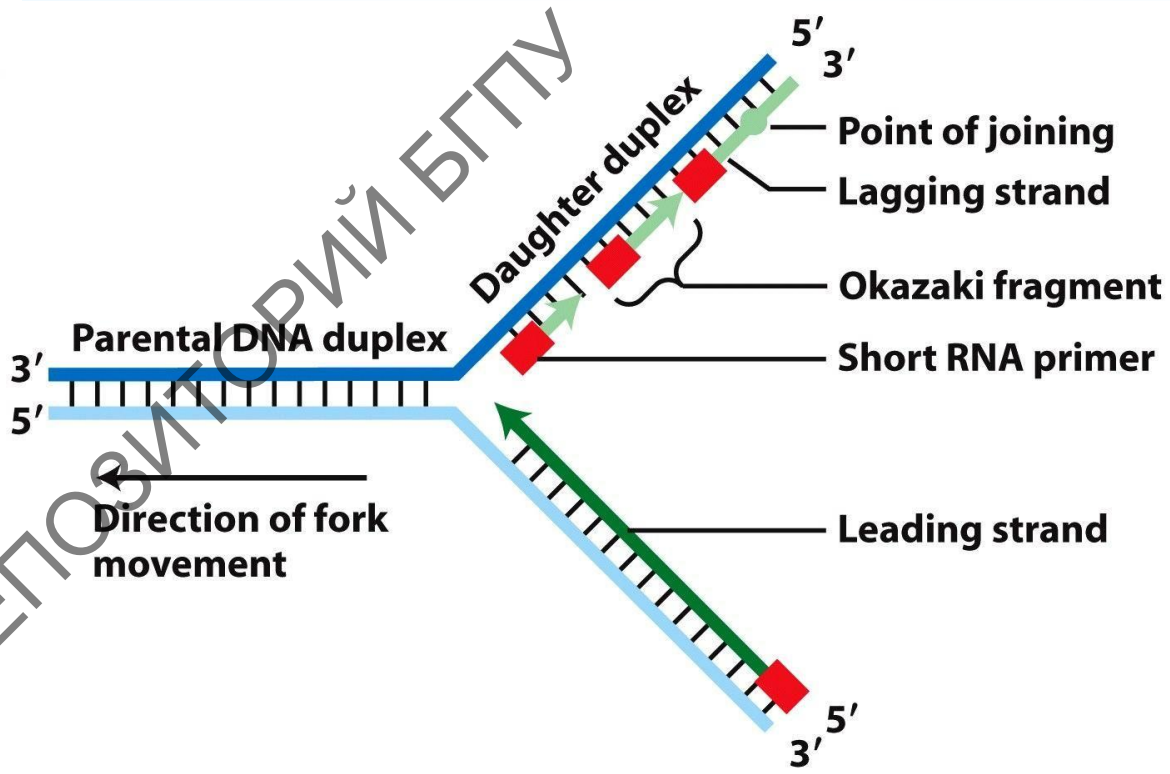
Matrix processes in the cell

DNA replication

DNA replication or reduplication (doubling) is called its synthesis.



DNA replication scheme



Replicative fork structure diagram

Protein biosynthesis. Transcription

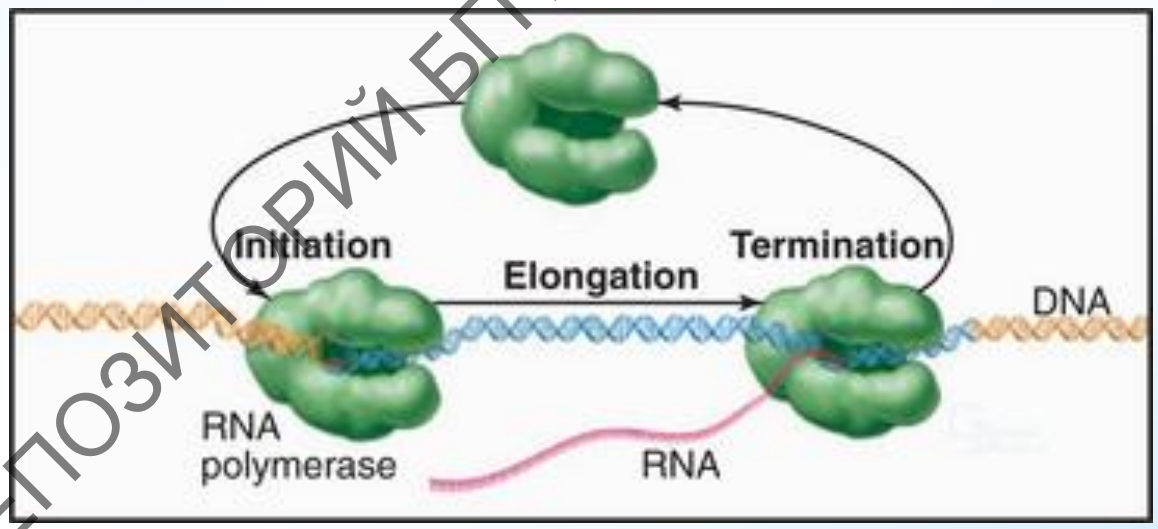
Transcription (rewriting) is the synthesis on the DNA matrix of mRNA (the primary product of a gene), carried out in the nucleus on a sense strand of DNA in a despiralized state.

In the process of transcription, three stages can be distinguished.

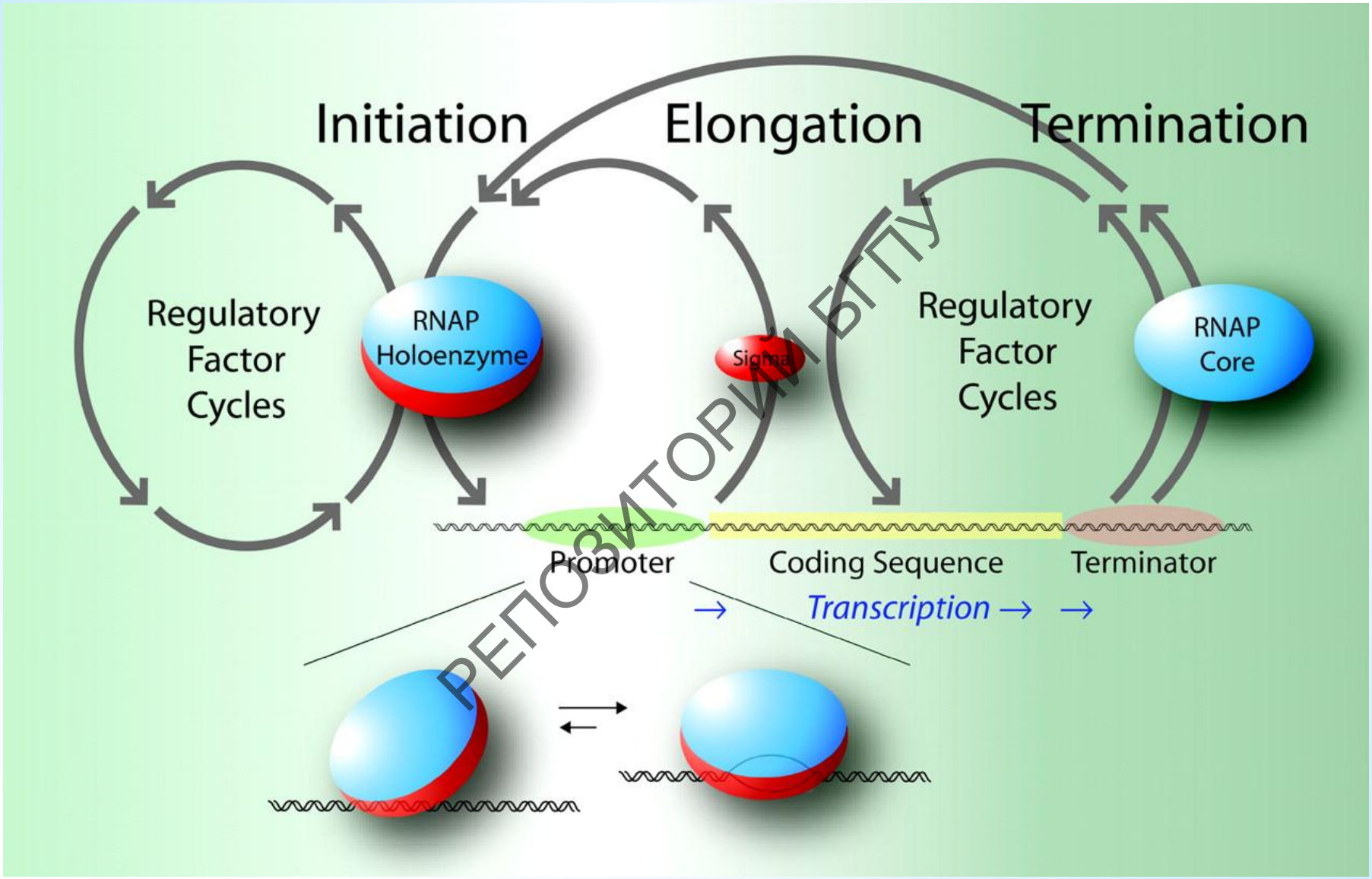
1 - **initiation** of transcription the beginning of the synthesis of RNA strands,
the first bond is formed between nucleotides.

2 - **elongation** - extension of the thread, its elongation,

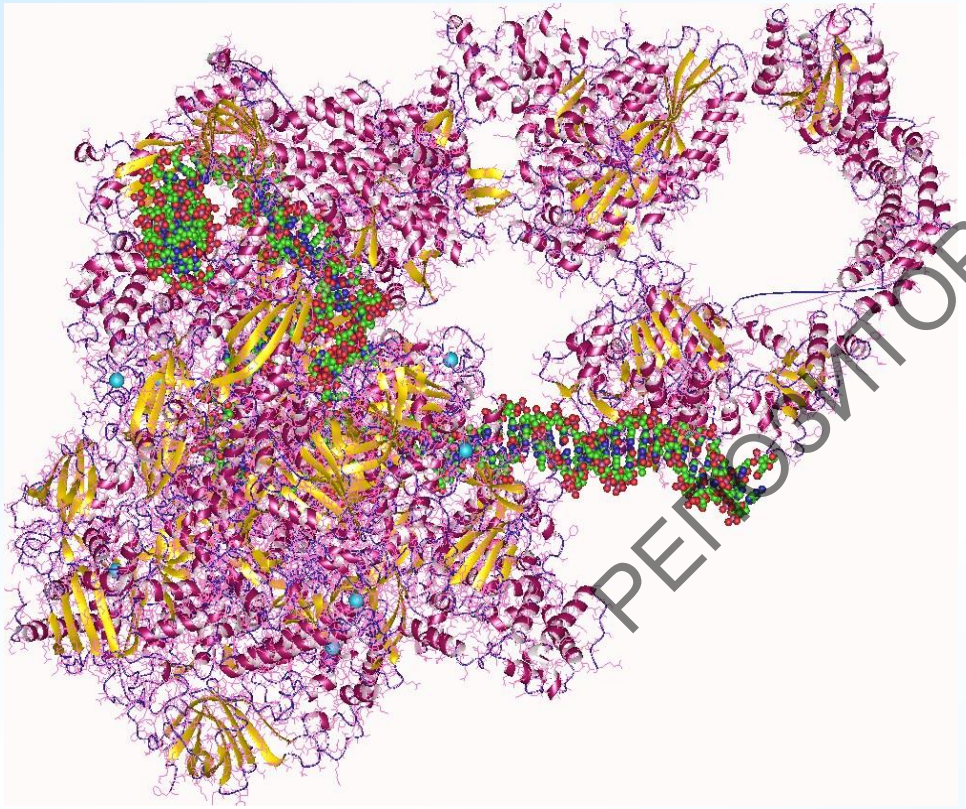
3 - **termination** - the synthesis is completed, there is a release of the synthesized RNA.

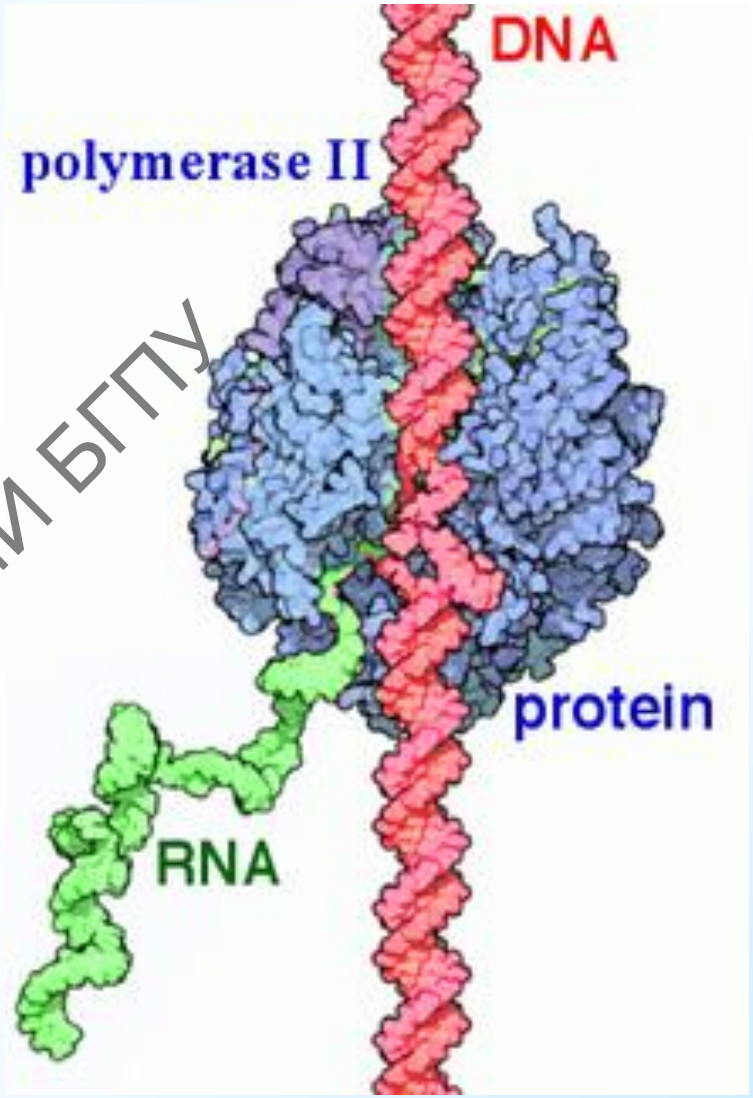


The general scheme of the transcription cycle



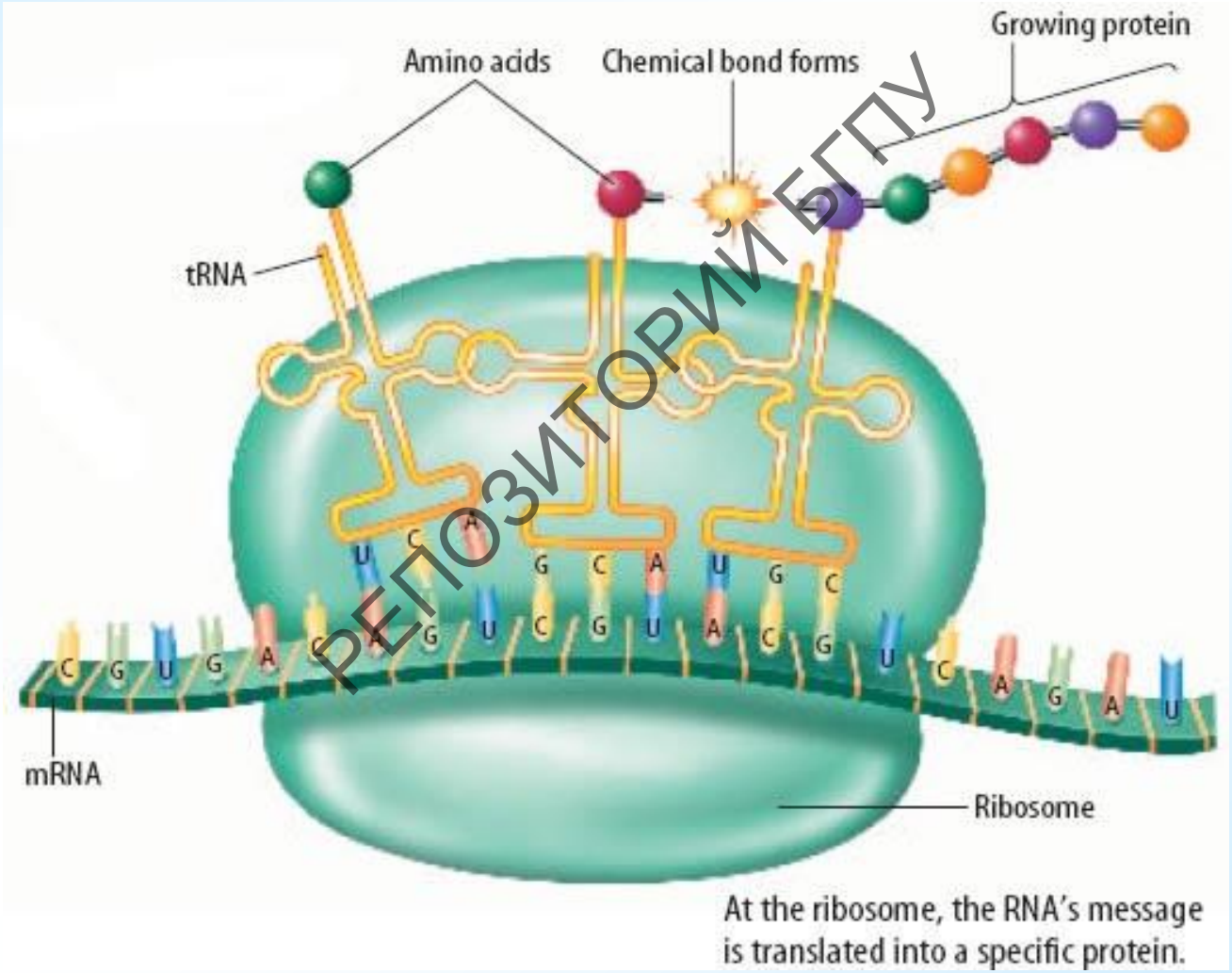
RNA polymerase





TRANSLATION (second stage of protein synthesis)

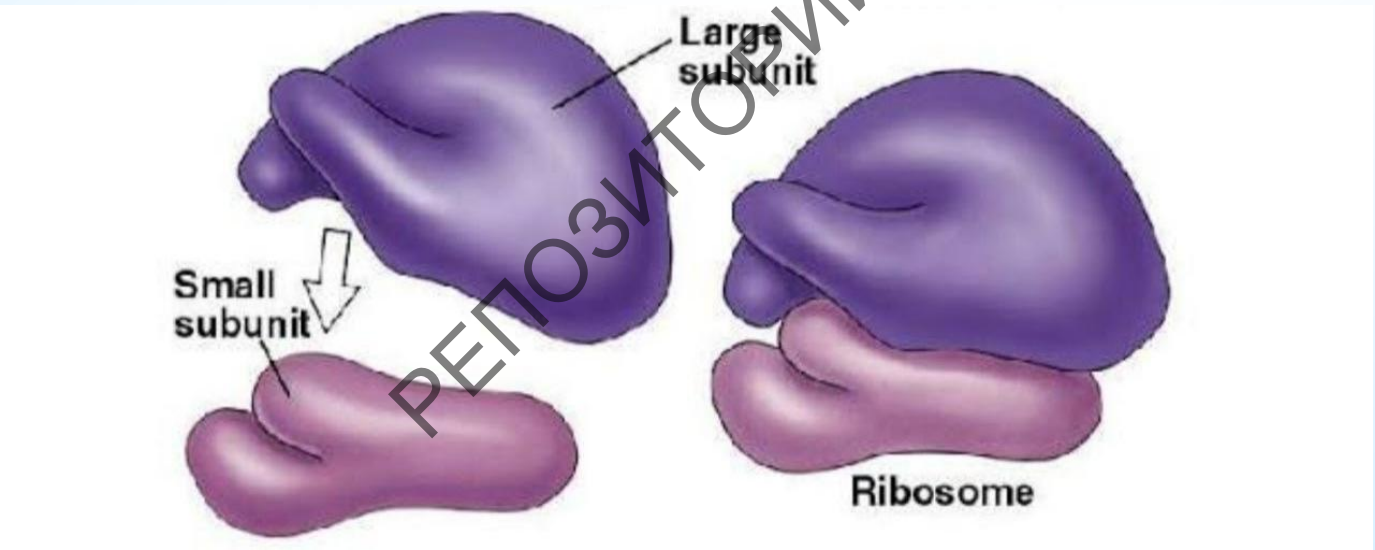
Translation is the process of translating mRNA genetic information into the structure of a polypeptide.



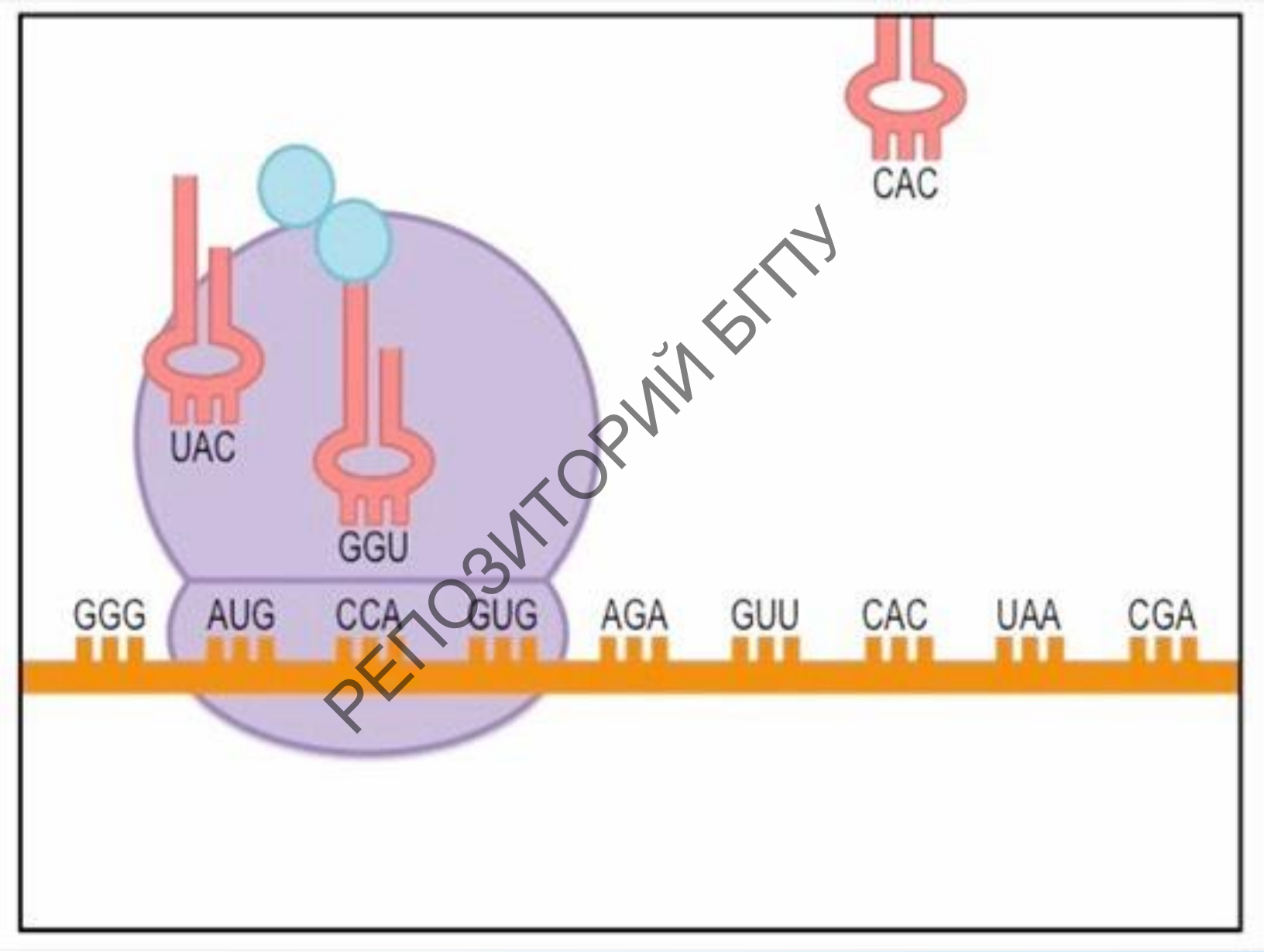
At the ribosome, the RNA's message is translated into a specific protein.

TRANSLATION (second stage of protein synthesis)

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



TRANSLATION (second stage of protein synthesis)



TRANSLATION (second stage of protein synthesis)