#### 1. General information on the course

Full course name	Modern Problems of Molecular Biology
Full official name of a higher education institution	Sumy State University
Full name of a structural unit	Academic and Research Medical Institute. Кафедра фізіології і патофізіології з курсом медичної біології
Author(s)	Obukhova Olha Anatoliivna
Cycle/higher education level	The Second Level Of Higher Education, National Qualifications Framework Of Ukraine – The 7th Level, QF-LLL – The 7th Level, FQ-EHEA – The Second Cycle
Semester	18 weeks during the 3rd semester or 18 weeks during the 4th semester
Workload	After approval of the standard. The volume of the discipline is 5 credits. ECTS, 150 hours, of which 36 hours. is contact work with the teacher (36 hours of practical classes)
Language(s)	English

### 2. Place in the study programme

Relation to curriculum	Elective course available for study programme "Medicine"
Prerequisites	Basic knowledge of general biology, organic chemistry, medical biology
Additional requirements	There are no specific requirements
Restrictions	There are no specific restrictions

### 3. Aims of the course

The main purpose of the course is to master the student to interpret the biological nature and mechanisms of human diseases that arise from molecular and genetic changes in the genotype and anthropogenic changes in the environment

### 4. Contents

### Module 1. Molecular basis of heredity

Topic 1 Subject and tasks of molecular biology

The main stages of development of molecular biology and molecular genetics, their relationship with classical genetics. Practical significance of molecular biology. The most important modern achievements of biotechnology, prospects for their use in clinical medicine. The concept of molecular medicine.

Topic 2 Macromolecules as objects of molecular biology

Chemical composition of DNA and its macromolecular organization. Types of DNA helices. Molecular mechanisms of DNA recombination, replication, and repair. The concept of nucleases and polymerases.

Topic 3 Proteins and their role in ensuring biological specificity

Amino acids and their properties. Formation of a polypeptide chain. Primary, secondary, tertiary and quaternary protein structures. Conformation is the basis of protein properties. Folding. The concept of prion diseases. Classification of proteins according to their biological functions. Carrier proteins, signaling, protective, structural, receptor, regulatory, enzymes. The concept of proteomics.

Topic 4 Molecular mechanisms of DNA replication, recombination and repair

Chemical composition of RNA. Atypical RNA bases. Macromolecular organization of RNA. RNA types and their biological functions. General characteristics of the replication process. Events occurring in the replication fork. Telomere replication, telomerase. The value of underreplication of finite chromosome fragments in aging mechanisms. Replication error correction systems. Correction properties of DNA polymerases. Mechanisms for repairing damaged DNA. Direct and excision repair. Post replicationMM-type and recombinant type repair. SOS repair. The concept of DNA repairsdiseases. Molecular mechanisms of general genetic recombination. Break-Connection and Copy-Chase Hypotheses. Site-specific recombination. Gene conversion.

Topic 5 Molecular organization of genes

Pro-and eukaryotic gene structure. Classification of genes according to their functions. Structural genes. The concept of "housekeeping" genes and terminal differentiation genes. Types of pro-and eukaryotic regulatory genes.

Topic 6 Gene expression and its regulation

The concept of gene expression. The current state of central dogma of molecular genetics. Properties of the genetic code. Stages of protein biosynthesis. Enzymatic mechanisms and stages of transcription. Processing of primary transcripts. Alternative processing, RNA editing. Activation of amino acids. Molecular organization of ribosomes. Initiation, elongation and termination of polypeptide chain synthesis. Post-translational modification of proteins.

Topic 7 Organization of genomes of non-cellular and cellular organisms.

Organization of the genome of viruses. The concept of lysogenic and lytic cycles of viruses. Features of the genome and life cycle of retroviruses. The bacterial genome. Plasmids.

Topic 8 Organization of the human genome

Modern ideas about the human genome. Unique, moderately and highly repetitive DNA. Genes encoding polypeptides, RNA. Multigene families. Gene superfamily and their products. Pseudogens. Transposons. Scattered and tandem repeats. Mini-and microsatellite DNA. Extra-nuclear heredity. Mitochondrial genome.

Module 2. Molecular basis of hereditary diseases

Topic 9 Molecular mechanisms of gene mutations

Molecular mechanisms of gene mutations. Classification of gene mutations. The concept of monogenic hereditary diseases

Topic 10 Molecular mechanisms of chromosomal and genome mutations

Molecular and cytological mechanisms of chromosomal mutations. Modern methods of studying human karyotype: differential coloring, FISH-method, etc. Classification of mutations by causes. Mutagenic factors, methods for determining the mutagenic activity of substances. Antimutagenesis. Generative and somatic mutations

Topic 11 Regulation of the cell cycle. Apoptosis

The mitotic cycle and its regulation. The role of cyclins and cyclin-dependent kinases. Principles of mitogenic signal transmission. The role of growth factors, integrins and cadherins. Mitotic cycle checkpoints. Apoptosis.

Topic 12 Molecular basis of oncogenetics

Genetic mechanisms of carcinogenesis. General characteristics of genes involved in carcinogenesis: viral oncogenes, proto-oncogenes, tumor suppressor genes, mutator genes. Carcinogenic factors.

#### Module 3. Modern issues of genetic technologies

Topic 13 Nucleic acid studies

Methods of nucleic acid research. Methods of DNA extraction from plant and animal tissues and its purification. Enzymes used for genetic engineering studies. Restrictase. DNA probes. DNA electrophoresis.

Topic 14 Methods of DNA diagnostics

Identification of DNA and RNA fragments by hybridization methods. Southern-, Northern-, Western blotting. Cloning of nucleic acid fragments in vitro. Polymerase chain reaction. DNA sequencing. Impressions for DNA diagnostics. Direct and indirect methods. DNA chips. Molecular-genetic research methods in forensic medicine

Topic 15 Methods of genetic engineering

The concept of genetic engineering. Recombinant DNA, principles of their construction. Cloning of nucleic acid fragments in vivo. Definition of vector concept in biology. Vectors: plasmids, bacteriophages, cosmids, artificial chromosomes. Methods for finding specific recombinant DNA. Genomic DNA libraries, cDNA libraries.

Topic 16 Transgenic organisms. Gene therapy

The principle of construction of transgenic organisms. Transgenic bacteria. The main areas of application in the economy and medicine. Recombinant drugs. Transgenic plants. Main directions of use of transgenic plants. Transgenic animals as disease models and bioreactors. Environmental safety issues. Principles of gene therapy. Ex vivo and in vivo gene therapy. Viral and non-viral vectors in gene therapy. Perspectives and limitations of gene therapy. Gene vaccines. Gene therapy in oncology.

Topic 17 Cloning of organisms and cells.

The concept of cloning. Natural and artificial clones. History of cloning of organisms. Biological and ethical problems of cloning. Therapeutic cloning and its prospects in medicine

Topic 18 Final lesson on the subject "Modern problems of molecular biology"

### 5. Intended learning outcomes of the course

After successful study of the course, the student will be able to:

LO1	Be able to identify and identify the leading clinical symptoms and molecular genetic syndromes, using preliminary history of the patient, patient examination data, knowledge of the person, his organs and systems, to establish the most probable syndrome preliminary clinical diagnosis of the disease
LO2	To evaluate information about the general condition of the patient, evaluate the condition of organs and systems of the body, based on the results of molecular genetic laboratory studies to evaluate information about the diagnosis.
LO3	Be able to find the necessary information in the professional literature and databases of other sources, analyze, evaluate and apply this information. Be able to apply modern digital technologies, specialized software.

## 7. Soft Skills

## 8. Teaching and learning activities

## Topic 1. Subject and tasks of molecular biology

pr.tr.1 "The subject and tasks of molecular biology" (full-time course)

Basic stages of development of molecular biology and molecular genetics, their interrelation with classical genetics. The practical importance of molecular biology. The most important modern achievements of biotechnology, prospects for their use in clinical medicine. The concept of molecular medicine.

### Topic 2. Macromolecules as objects of molecular biology

pr.tr.2 "Macromolecules as objects of study of molecular biology" (full-time course)

The chemical composition of DNA and its macromolecular organization. Types of DNA helices. Molecular mechanisms of recombination, replication, and DNA repair. The concept of nuclease and polymerase.

### Topic 3. Proteins and their role in ensuring biological specificity

pr.tr.3 "Proteins and their role in providing biological specificity" (full-time course)

Amino acids and their properties. Formation of the polypeptide chain. Primary, secondary, tertiary and quaternary protein structures. Conformation is the basis of protein properties. Folding. The concept of prion diseases. Classification of proteins according to their biological functions. Carrier proteins, signaling, protective, structural, receptor, regulatory, enzymes. The concept of proteomics.

### Topic 4. Molecular mechanisms of DNA replication, recombination and repair

pr.tr.4 "Molecular mechanisms of DNA replication, recombination and repair" (full-time course)

Chemical composition of RNA. Atypical RNA bases. Macromolecular organization of RNA. RNA types and their biological functions. General characteristics of the replication process. Events occurring in the replication fork. Telomere replication, telomerase. The value of underreplication of finite chromosome fragments in aging mechanisms. Replication error correction systems. Correction properties of DNA polymerases. Mechanisms for repairing damaged DNA. Direct and excision repair. Post replicationMM-type and recombinant type repair. SOS repair. The concept of DNA repairsdiseases. Molecular mechanisms of general genetic recombination. Break-Connection and Copy-Chase Hypotheses. Site-specific recombination. Gene conversion.

### **Topic 5. Molecular organization of genes**

pr.tr.5 "Molecular organization of genes" (full-time course)

Pro-and eukaryotic gene structure. Classification of genes according to their functions. Structural genes. The concept of "housekeeping" genes and terminal differentiation genes. Types of pro-and eukaryotic regulatory genes

#### Topic 6. Gene expression and its regulation

pr.tr.6 "Gene expression and its regulation" (full-time course)

The concept of gene expression. The current state of central dogma of molecular genetics. Properties of the genetic code. Stages of protein biosynthesis. Enzymatic mechanisms and stages of transcription. Processing of primary transcripts. Alternative processing, RNA editing. Activation of amino acids. Molecular organization of ribosomes. Initiation, elongation and termination of polypeptide chain synthesis. Post-translational modification of proteins

### Topic 7. Organization of genomes of non-cellular and cellular organisms.

pr.tr.7 "Organization of genomes of non-cellular and cellular organisms" (full-time course)

Organization of the genome of viruses. The concept of lysogenic and lytic cycles of viruses. Features of the genome and life cycle of retroviruses. The bacterial genome. Plasmids

#### **Topic 8. Organization of the human genome**

pr.tr.8 "Organization of the human genome" (full-time course)

Modern ideas about the human genome. Unique, moderately and highly repetitive DNA. Genes encoding polypeptides, RNA. Multigene families. Gene superfamily and their products. Pseudogens. Transposons. Scattered and tandem repeats. Mini-and microsatellite DNA. Extra-nuclear heredity. Mitochondrial genome.

### **Topic 9. Molecular mechanisms of gene mutations**

pr.tr.9 "Molecular mechanisms of gene mutations" (full-time course)

Molecular mechanisms of gene mutations. Classification of gene mutations. The concept of monogenic hereditary diseases.

#### Topic 10. Molecular mechanisms of chromosomal and genome mutations

pr.tr.10 "Molecular mechanisms of chromosomal and genome mutations" (full-time course)

Molecular and cytological mechanisms of chromosomal mutations. Modern methods of studying human karyotype: differential coloring, FISH-method, etc. Classification of mutations by causes. Mutagenic factors, methods for determining the mutagenic activity of substances. Antimutagenesis. Generative and somatic mutations

### Topic 11. Regulation of the cell cycle. Apoptosis

pr.tr.11 "Regulation of the cell cycle. Apoptosis" (full-time course)

The mitotic cycle and its regulation. The role of cyclins and cyclin-dependent kinases. Principles of mitogenic signal transmission. The role of growth factors, integrins and cadherins. Mitotic cycle checkpoints. Apoptosis.

#### **Topic 12. Molecular basis of oncogenetics**

pr.tr.12 "Molecular basis of oncogenetics" (full-time course)

Genetic mechanisms of carcinogenesis. General characteristics of genes involved in carcinogenesis: viral oncogenes, proto-oncogenes, tumor suppressor genes, mutator genes. Carcinogenic factors.

#### **Topic 13. Nucleic acid studies**

pr.tr.13 "Nucleic acid studies" (full-time course)

Methods of nucleic acid research. Methods of DNA extraction from plant and animal tissues and its purification. Enzymes used for genetic engineering studies. Restrictase. DNA probes. DNA electrophoresis.

### **Topic 14. Methods of DNA diagnostics**

pr.tr.15 "Methods of DNA diagnostics" (full-time course)

Identification of DNA and RNA fragments by hybridization methods. Southern-, Northern-, Western blotting. Cloning of nucleic acid fragments in vitro. Polymerase chain reaction. DNA sequencing. Impressions for DNA diagnostics. Direct and indirect methods. DNA chips. Molecular-genetic research methods in forensic medicine

### **Topic 15. Methods of genetic engineering**

pr.tr.15 "Methods of genetic engineering" (full-time course)

The concept of genetic engineering. Recombinant DNA, principles of their construction. Cloning of nucleic acid fragments in vivo. Definition of vector concept in biology. Vectors: plasmids, bacteriophages, cosmids, artificial chromosomes. Methods for finding specific recombinant DNA. Genomic DNA libraries, cDNA libraries

#### **Topic 16. Transgenic organisms. Gene therapy**

pr.tr.16 "Transgenic organisms. Gene therapy" (full-time course)

The principle of construction of transgenic organisms. Transgenic bacteria. The main areas of application in the economy and medicine. Recombinant drugs. Transgenic plants. Main directions of use of transgenic plants. Transgenic animals as disease models and bioreactors. Environmental safety issues. Principles of gene therapy. Ex vivo and in vivo gene therapy. Viral and non-viral vectors in gene therapy. Perspectives and limitations of gene therapy. Gene vaccines. Gene therapy in oncology

## Topic 17. Cloning of organisms and cells.

pr.tr.17 "Cloning of organisms and cells" (full-time course)

The concept of cloning. Natural and artificial clones. History of cloning of organisms. Biological and ethical problems of cloning. Therapeutic cloning and its prospects in medicine.

# Topic 18. Final lesson on the subject "Modern problems of molecular biology"

pr.tr.18 "Final lesson on the subject "Modern problems of molecular biology"" (full-time course) abstract defense and computer testing

# 9. Teaching methods

9.1 Teaching methods

Course involves learning through:

TM1	Practical classes
TM2	Case-based learning (CBL).
TM3	Team-based learning (TBL).
TM4	Research-based learning (RBL)
TM5	Educational discussion / debate

Practical classes in the discipline include consideration of information on the diagnosis of molecular genetic pathologies, using knowledge of the genotype and karyotype of man, based on the results of molecular genetic laboratory research. Students are given the opportunity to identify and record a genetic-molecular-genetic symptom or syndrome, using preliminary history and genetic-molecular examination of the patient, adhering to the relevant ethical and legal norms. Be able to establish the most probable or syndromic diagnosis of the disease. Assign laboratory and / or instrumental examination of the patient. Carry out differential diagnosis. Establish a preliminary and genetic diagnosis.

The discipline provides students with the following soft skills: GC 2. Ability to learn and master modern knowledge, GC 3. Ability to apply knowledge in practical situations, GC 4. Knowledge and understanding of the subject area and understanding of professional activities, GC 10. Ability to use information and communication technologies , GC 11. Ability to search, process and analyze information from various sources.

## 9.2 Learning activities

LA1	Preparation for the practical lessons
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LA2	E-learning in systems (MIX.sumdu.edu.ua, Google Meet, Zoom)
LA3	Preparation and presentation of the report
LA4	Individual research project (preparation of multimedia presentations)
LA5	Watching movies
LA6	Essay writing
LA7	Self-learning
LA8	Preparation for current and final control

# 10. Methods and criteria for assessment

## 10.1. Assessment criteria

Definition	National scale	Rating scale
Outstanding performance without errors	5 (Excellent)	$170 \le RD \le 200$
Above the average standard but with minor errors	4 (Good)	$140 \le RD < 169$
Fair but with significant shortcomings	3 (Satisfactory)	$120 \le RD < 139$
Fail – some more work required before the credit can be awarded	2 (Fail)	$0 \le \text{RD} < 119$

# 10.2 Formative assessment

	Description	Deadline, weeks	Feedback
FA1 Computer testing			
FA2 Protection of presentations and reports			
FA3 Interviews and oral comments of the teacher on his results			
FA4 Independent performance of situational exercises by students in practical classes and their discussion.			

FA5		
Teacher's		
instructions in		
the process of		
performing		
practical tasks		

# 10.3 Summative assessment

	Description	Deadline, weeks	Feedback
SA1 Scores for current educational activity			
SA2 Final control: differentiated test (according to the regulations)			

Form of assessment:

	Points	Minimum points	Можливість перескладання з метою підвищення оцінки
3 semester		200 scores	5
SA1. Scores for current educational activity		120	
	120	Не передбачено	No
SA2. Final control: differentiated test (according to the regulations)		80	
Report (preparation, presentation, defense)	30	Не передбачено	No
Computer testing KROK-1	50	Не передбачено	No
4 semester	200 scores		
SA1. Scores for current educational activity	120		
	120	Не передбачено	No
SA2. Final control: differentiated test (according to the regulations)	80		
Report (preparation, presentation, defense)	30	Не передбачено	No
Computer testing KROK-1	50	Не передбачено	No

In special situations, work during the semester can be done remotely. Computer testing

## 11. Learning resources

# 11.1 Material and technical support

MTS1	Computers, computer systems and networks
MTS2	Library funds
MTS3	Multimedia, video and audio, projection equipment (video cameras, projectors, screens, smart boards, etc.)

# 11.2 Information and methodical support

Essential Reading	
1	Medical Biology:textbook /S.Ya. Paryzhak, Z.D. Vorobets - Lviv: Qvart, 2020 436 p.
2	Step 1. Lecture notes 2021 Biochemistry and Medical genetics. NewYork. Kaplan, Inc 2021 - 403 c.
3	Molecular Cell Biology 9E (International Edition) & Achieve for Molecular Cell Biology 9E (2-Term Access; International Edition) H; BERK A. LODISH Published by Macmillan Learning, New York, 2021
Supplemental Reading	
1	Molecular biology of the cell. 6th ed. / B. Alberts, A. Johnson, J. Lewis et al. — N. Y.: Garland Science, 2014. — 1464 p
2	Molecular Cell Biology. 8th ed. / H. Lodish, A. Berk, Kaiser C.A. et al. — N. Y.: W.H. Freeman & Co. Ltd, 2016. — 1280 p.
3	Kaplan Medical's USMLE STEP 1. Biochemistry and Medical Genetics. Lecture notes 2018 432 p
4	The associason of vitamin D receptor gene (VDR)polymorphisms with high blood pressure in stroke patients of ukrainian population / Obukhova O.A., Ataman A.V., Zavadska M.M., Piven S.M., Levchenko Z.M.// Wiadomosci lekarskie (Warsaw, Poland : 1960). – 2020. – Vol. 73 (11). – P. 2349–2353.
5	Ataman A.V., Harbuzova V.Y., Obukhova O.A., Dubovyk Y.I. Analysis of Ectonucleotide Pyrophosphatase/Phosphodiesterase 1 Gene K121Q Polymorphism Association with Some Risk Factors of Atherosclerosis in Patients with Acute Coronary Syndrome // Cytology and Genetics. – 2018. – Vol. 52 (2). – P. 127–131.
Web-based and electronic resources	
1	OMIM (Online Mendelian Inheritance in Man) – An Online Catalog of Human Genes and Genetic Disorders http://omim.org/
2	https://pubmed.ncbi.nlm.nih.gov/
3	https://www.ncbi.nlm.nih.gov/
4	https://ocw.sumdu.edu.ua/content/994