Khanty-Mansiysk Autonomous Okrug-Ugra
«Surgut State University»

Approved by
Deputy Rector for Academic Affairs

_____E.V. Konovalova

_13_ June 2024г., Record No 5

Human genetics

Syllabus

Department **Morphology and physiology**

Curriculum s310501-ЛечДелоИн-24-1.plx
Specialty 31.05.01 General Medicine

Qualification **General Practitioner**

Form of education **Full-time**

Total (in credits) **2**

Total academic hours 72
including:
Classes 56
Self-study 16

Control:
2nd term-credit

Course outline in terms

Academic year (Term)	2 (1.2)		Total	
Weeks	21 3/6			
Types of classes	Cur	Syl	Cur	Syl
Lectures	16	16	16	16
Practical	40	40	40	40
Classes total	56	56	56	56
Contact training	56	56	56	56
Self-study	16	16	16	16
Total	72	72	72	72

The Syllabus is compiled by:
PhD in Biological Sciences, Associate Professor, Soltys T. V., lecture Maksimova Anna Sergeevna

The Syllabus
Human genetics

Developed in accordance with Federal State Educational Standard:
Federal State Educational Standard of higher education in the specialty 31.05.01 General medicine (Order of the Ministry of Education and Science of the Russian Federation on August, 12, 2020. №988)

Based on the Curriculum:
31.05.01 GENERAL MEDICINE
Specialization: General Medicine
Approved by the Academic Council of Surgut State University, «13» June 2024 Record No 5.

The Syllabus was approved by the department
Morphology and physiology

Head of Department, Doctor of Medicine, Professor Stolyarov V.V.

1. COURSE OBJECTIVES	
1.1	Purpose: To form students ' understanding of the molecular and cellular foundations of genetics, its physiological and ontogenetic aspects, the doctrine of heredity and variability in families and populations, evolutionary and ecological genetics.
1.2	To know in the field of human genetics.
1.3	To solve genetic problems.
1.4	To have skills of cytogenetic, biochemical and prenatal diagnostics of hereditary diseases, carrying out preventive measures-identification of high-risk groups among the population of their prevention and gene therapy

2. COURSE OVERVIEW	
Course code (in curriculum)	Б1.О.04
2.1	**Assumed background:**
2.1.1	Biology (school course)
2.2	**Post-requisite courses and practice:**
2.2.1	Neurology, medical genetics, neurosurgery
2.2.2	Propaedeutics of internal diseases
2.2.3	Obstetrics

3. COMPETENCES UPON COMPLETION OF THE COURSE (MODULE)

GPC-4.2: Knows instrumental and morphological criteria for diagnosing diseases and conducting clinical interpretation of the results of instrumental examination methods

GPC-4.3: Able to apply knowledge and skills in conducting a diagnostic search for diseases using medical equipment (products) to establish a diagnosis

GPC-5.2: Knows the structure of the cell, the phases of its division, the theoretical principles of genetics and the biology of insects and helminths, their role in the etiology of human diseases

GPC-7.2: Able to prescribe modern treatment regimens based on approved current Clinical Recommendations

PC-1.1: Demonstrates knowledge in etiology, pathogenesis, diagnostic criteria (clinical - subjective, physical, laboratory, instrumental, identifies the patient's common pathological conditions, symptoms, disease syndromes and diagnoses nosological forms according to the International Statistical Classification of Diseases and Related Health Problems, X - XI revisions

PC-9.1: Analyzes medical information

PC-9.2: Provides evidence-based health information

PC-10.1: Participates in applying new methods and techniques aimed at protecting the public health

By the end of the course students must

3.1	**know:**
3.1.1	Rules of safety and work in medical and biological laboratories; the laws of genetics, its significance for medicine, the laws of heredity and variability in individual development as a basis for understanding the pathogenesis and etiology of hereditary and multifactorial human diseases; the main pathological conditions, syndromes and symptoms of genetic diseases, as well as methods of their treatment; new methods and techniques in the field of human genetics.
3.2	**be able to:**
3.2.1	Use new methods and techniques aimed at protecting the health of citizens; solve typical tasks on the following topics: organization of hereditary material; regulation of gene expression; cytoplasmic heredity; patterns of inheritance; genetics of populations; analyze pedigrees, make a forecast of the development of a hereditary disease in proband and his relatives; assess morpho-functional, physiological conditions and pathological processes in genetic diseases, to identify families and groups of people with an increased risk of developing a particular disease with a hereditary predisposition; describe the morphological changes of the studied macroscopic, microscopic preparations and electronograms; provide publicly available medical information.

4. STRUCTURE AND CONTENTS OF THE COURSE (MODULE)

Class Code	Topics /Class type	Term / Academic	Academic hours	Competences	Literature	Notes
	Module 1. Patterns of inheritance					
1.1	History and significance of human genetics /Lecture/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	

1.2	Structure and functions of genetic material /Lecture/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.3	Replication of DNA and chromosomes /Lecture/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.4	DNA repair. Molecular mechanisms of genetic recombination /Lecture/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.5	Regulation of gene activity /Lecture/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.6	Mendelism (hybridological method, the laws of inheritance of G. Mendel's traits, the discreteness of heredity) /Practice/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.7	The chromosomal theory of heredity /Practice/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.8	Structure and functions of genetic material /Practice/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.9	Multilevel organization of the genome /Practice/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.10	Interaction of allelic and non-allelic genes /Practice/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.11	Gene coupling and crossing-over. Genetics of sex /Practice/	2	2	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
1.12	Preparation for an oral survey, tests. Solving genetic problems /Self-control/	2	4	GPC-5.2 PC-10.1	L1.1 L2.1 L3.1	
	Module 2. Variability					
2.1	Genotype and phenotype /Lecture/	2	2	GPC-4.3 GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
2.2	Molecular mechanisms of mutagenesis /Lecture/	2	2	GPC-4.3 GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
2.3	Variation of the manifestation of hereditary traits in the individual development of organisms. Modification and reaction rate /Practice/	2	2	GPC-4.3 GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
2.4	Theoretical foundations of mutational variability /Practice/	2	2	GPC-4.3 GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
2.5	Spontaneous mutation process. Mutagenesis inhibitors. Detection of mutagens /Practice/	2	2	GPC-4.3 GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
2.6	Non-chromosomal inheritance /Practice/	2	2	GPC-4.3 GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
2.7	Preparation for an oral survey, tests. Solving genetic problems. Writing research papers /Self-control/	2	4	GPC-4.3 GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
	Module 3. The genetic basis of evolution					
3.1	Genetics and ontogenesis /Practice/	2	2	GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	

3.2	Genes controlling embryonic induction /Practice/	2	2	GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
3.3	The genetic foundations of evolution. Population genetics /Practice/	2	2	GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
3.4	Synthetic theory of evolution. Genetics and problems of evolutionary theory. Evolution of the human genome /Practice/	2	2	GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
3.5	Preparation for an oral survey, tests. Solving genetic problems. Writing research papers /Self-control/	2	4	GPC-5.2 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
	Module 4. Hereditary diseases					
4.1	Heredity and human pathology /Lecture/	2	2	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.2	Chromosomal abnormalities and associated syndromes /Practice/	2	2	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.3	Monogenic human diseases /Practice/	2	2	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.4	Diseases with non-traditional types of inheritance (diseases inherited linked to sex, mitochondrial) /Practice/	2	2	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.5	Diseases with non-traditional types of inheritance (genomic imprinting(DGI), expansion of trinucleotide repeats(DETR), prion diseases (PD)) /Practice/	2	1	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.6	General characteristics and classification of hereditary metabolic diseases(amino acids (aminoacidopathy), carbohydrates) /Practice/	2	2	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.7	Hereditary metabolic diseases (lipids, erythron, lysosomal and peroxisomal) /Practice/	2	1	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.8	Genetics of widespread diseases. Prevention of hereditary pathology. Treatment of hereditary diseases /Practice/	2	2	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	
4.9	Preparation for an oral survey, tests. Writing research papers /Self-control/	2	4	GPC-4.2 GPC-4.3 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	

4.10	Control work / Control./	2	0	GPC-4.2 GPC-4.3 GPC-5.2 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	protection of abstracts
4.11	Interview on credit questions / Credit/	2	0	GPC-4.2 GPC-4.3 GPC-5.2 GPC-7.2 PC-1.1 PC-9.1 PC-9.2 PC-10.1	L1.1 L2.1 L3.1	interview on theoretical issues, solving genetic problems

5. ASSESSMENT TOOLS

5.1. Assessment tools

Presented by a single document

5.2. Assessment tools for diagnostic assessment

Presented by a single document

6. COURSE (MODULE) RESOURCES

6.1. Recommended Literature

6.1.1. Core

	Authors	Title	Publish, year	Quantity
L1.1	Nussbaum R. L., McInnes R. R., Willard H. F.	Thompson & Thompson Genetics in Medicine	Amsterdam: Elsevier, cop. 2016	3

6.1.2. Additional literature

	Authors	Title	Publish, year	Quantity
L2.1	Shevchenko V. A., Topornina N. A., Stvolinskaja N. S.	Human genetics: a textbook for students of higher educational institutions education	M.: Vlados, 2004	30

6.1.3. Methodical literature

	Authors	Title	Publish, year	Quantity
L3.1	Zhivogljad R. N., Soltys T. V.	Morphological characteristics of congenital deformities: teaching aid	Surgut: publishing house SurSU, 2007, electronic resource	1

6.2. Internet resources

E1	http://iprbookshop.ru/
E2	http://e.lanbook.com/
E3	http://www.znanium.com
E4	http://www.studmedlib.ru/

6.3.1 Software

6.3.1.1	Application Software Package Microsoft Offise

6.3.2 Information Referral systems

6.3.2.1	Information and legal portal Garant.ru http://www.garant.ru
6.3.2.2	Legal reference system Consultant Plus http://www.consultant.ru

7. MATERIAL AND TECHNICAL SUPPORT OF DISCIPLINE (MODULE)

7.1	The classroom for practical classes, group and individual consultations, current and intermediate control, for independent work is equipped with a blackboard, a portable projector, a computer, an interactive whiteboard, microscopes, racks with visual aids, standard furniture for the teacher (table, chair), standard furniture for students (educational tables and chairs for 18 seats), visual aids, micro-preparations.