

MED 2011- Medical Genetics I

Course Name	Code	Semester	Type of course	Theory (hours)	Group Work (hours)	ECTS
Medical Genetics I	MED 2011	III	Mandatory	8	18	2
Faculty, the educational program and education level	Faculty of Medicine, one-cycle Educational Program "Medicine"					
Author (s)	<p>Sophiko Tskvitinidze – Assistant Professor, Dr. of Biology E.MAIL: tskvitinidze.s@gmail.com TEL: +995 593 744 133 /+ 995 599 122 633 Consultation days and time: individually</p>					
Educational course format	<p>Total: 60 hours Contact hours: 30 h 1. Lecture – 8 h 2. Group work/Seminars – 18 h 3. Midterms – 2 h 4. Final exam -2 h Independent work – 30 h</p>					
Prerequisites	MED 1001 - Molecular basis of cell					
The purpose (s) of tutorial course/modules	<p>The course aims to teach the main principles in Genetics, Structure of Genetic material (DNA, RNA Protein) and inheritance characteristics; Principles of Mendelian and nonmendelian inheritance. Genetic basis of diseases based on chromosome structural and number changes, which will allow the student to access the deepness of heredity and hereditary disorders and use that knowledge to give proper consultation in their medical activity.</p>					
Teaching and learning strategy	<p>Lectures - Monologue, explanation, demonstration (video, Power-point); Group working/Seminars-The student performs the task, which is thematically derived from the course, but additionally requires independent working, searching of literary materials from the Internet and processing of information. Student's verbal activity and knowledge deepness will be assessed; Abstract preparation process will be connected with the method of work with literature, periodic issues and other additional sources working out. Written form of abstract should be supported with slideshow. Group discussion/Problem based learning – Students will divide in the groups and discuss around topic, express their opinions. The aim of this activity is development of the listening, debate and critical thinking, analyzing and reconciliation skills.</p>					

Assessment criteria

Maximum score- 100:

Midterm assessment -60 scores, that includes:

- Attendance -10 scores
- Activity in group – 10 scores
- Abstract preparation and presentation – 10 scores
- Discussion – 10 scores
- **Midterm Exam – 20 scores**

Group Work are Assessment Based on the Following Criteria (maximum 10 scores)

10 scores - Student has been able to present complete and thorough knowledge of the subject, a substantial amount of detailed and relevant information. Demonstrate considerable depth of understanding of the studied main and additional literature. Bring forward a balanced view of the main arguments on the issues.

9 scores - Student has been able to bring forward a consistent number of deductions on most of the topics tackled. make very good comments on the different perspectives on most of the issues. Demonstrates knowledge of the main readers.

8 scores - Student has been able to bring forward a consistent knowledge, Has properly developed terminology. Demonstrates knowledge of the main readers.

7 points - Student has been able to present some factual information sufficiently linked with the topic. demonstrate a good understanding of the topics selected. make a good attempt to bring forward a balanced view of some arguments on the issues. Terminology is partially developed.

6 scores - Student has been able to make some good comments on the different perspectives on some of the issues. Make poor deductions on most of the topics tackled. analyse some causes and results of human interactivity related to the issues.

5 scores - Student has been able to demonstrate inconsistent comments on the different perspectives on some of the issues. Terminology is partially developed. Present mediocre level of knowledge. Make poor deductions.

4 scores - Student demonstrates general overview of the topics. Terminology is not developed. Information sufficiently linked with the topic. Demonstrate irrelevant understanding of the literature.

3 scores – Student demonstrates general/superficial and inconsistent knowledge of the subject. No sufficient knowledge of the literature.

2 scores - Student demonstrates general comments, no knowledge of the terminology, no consistency.

1 scores – Student demonstrates insufficient answer, not terminology awareness, chronologic manner of the answer, mostly wrong, no knowledge of literature.

0 score : Student demonstrates not even elementary knowledge of the topics.

Abstract Preparation and presentation - Grading criteria – Maximum 10 scores

- 1.Actuality of the problem – 1 sc;
- 2.Accurate planning – 1 sc;
- 3.Review of the literature on the issue -1sc;
- 4.Research methods relevance with the research goals – 1sc;
- 5.Logical argumentation and correlation with the main theme – 1sc;
- 6.Reference accuracy and correlation with the main source - 1sc;
- 7.Culture of writing – 1sc;

	<p>8. Language and style accuracy - 1sc; 9. Visual and technical side of the material – 1sc; 10. Culture of the discussion and listening to the opponent- 1 sc.</p> <p>Discussion – grading criteria (maximum 10 scores)</p> <ul style="list-style-type: none"> • Critical thinking- 2 sc; • Culture of debates - 2 sc; • Argumentativeness - 2 sc; • Time management - 2 sc; • Academic and visual side of the presented material - 2 sc. <p>Midterm Exam - the written test – (40 questions 0.5 score each). Minimal score of midterm assessment (for final exam) – is 11; to take in account that student will receive the maximum score at the final exam.</p> <p>Final Exam -40 Is held in the written test form (test consists of 80 questions, each question is rated as 0.5 scores). Students have to score equal or more than 70% from final exam maximum score (40X70/100=28 maximum 28 scores from the overall 40) to pass the final examination. Credit will be given to the student if he has collected at minimum 51 scores out of 100. The students' assessment has to be done in the following way: Positive rate:</p> <ul style="list-style-type: none"> • (A) Excellent- 91 or more scores; • (B) Very Good- 81-90 scores; • (C) Good- 71-80 scores ; • (D) Satisfactory- 61-70 scores; • (E) Enough- 51-60 scores; <p>Negative rate:</p> <ul style="list-style-type: none"> • (FX) Failure - 41-50 scores, which means that a student needs to work more and an independent and considerable further work is required to pass the exam once again to be re-awarded; • (F) Fail - 40 scores or less, which means that the student's diligence is not sufficient and student has to learn the subject all over again. <p>The student can pass the additional exam during the same semester. The time interval between the final and the additional exams should be not less than 10 days.</p>
The basic literature	<ol style="list-style-type: none"> 1. David L. Rimoin's, Reed E.Pyritz, Bruce R.Korf. Essential Medical Genetics. Elsevier Saunders, 2013; 2. Jocelyn E.Krebs, Elliott S.Goldstein, Stephen T.Kilpatrick Lewi's Genes XI, Jones and Barlett Learning, 2014.
The auxiliary literature	

The tutorial/training course content

№	Subjects	Lecture (hour) 8	Work in group (hour) 22
1	Genetics and Genomics in Medicine, challenges and promising future.	1	2
2	NDA, Proteins and Chromosome; Human genome; Human chromosome learning, staining and identification methods.	1	3
3	Genetic code; Gene structure and organization; gene realization in central dogma – DNA – RNA – Protein; Changes in gene expression regulation and its connection with medicine.	1	2
4	Clinical cytogenetic; importance of mitosis and meiosis in medicine. Chromosome number disorders: Heteroploidy (triploidy, tetraploidy) and aneuploidy (monosomy, trisomy). Autosomic and sechromosomal anomalies. Review of diseases caused by chromosome number changes.	2	4
5	Chromosome structural aberrations and review of diseases caused by aberrations.		2
	Midterm	1	2
6	Mendelian genetics. Genotype, phenotype. Alleles. Monogenic trial: Autosomal dominant and autosomal recessive inheritance.	1	3
7	Nonmendelian inheritance. Linked gene inheritance. X-linked and Holandric inheritance.	1	2
	Final Exam		2

Learning Outcomes

Criteria	Competences
Knowledge and understanding	Student will have deep and essential knowledge of genetic basis of inheritance; the characteristics inherits by offspring; realizes the inheritance way of recessive and dominant genes/signs and able to predispose feasible results. Understand the basis behind each signs of human body as well in normal as in pathology.
Applying knowledge	Will be able to: <ul style="list-style-type: none"> • define heredity characteristics based of molecular structural level; • find a right method and original ways for clearance of disease inheritance specificity; • genealogic maps designing

Making Judgment	Will be able to: <ul style="list-style-type: none">• estimate genetic pathologies risk factor and make the appropriate conclusions• use genetic anamnesis to define inheritance characteristic of various diseases;• account the preventive measures in population
Communication Skills	Will be able to collaborate with the colleagues and this field specialists as verbally, as well as by writing forms