

MED 2012 –Medical Genetics II

Course Name	Code	Semester	Type of course	Theory (hours)	Group Work (hours)	ECTS
Medical Genetics II	MED 2012	IV	Mandatory	25	46	5
Faculty, the educational program and education level	Medical Faculty, one-cycle Educational Program “Medicine”					
Author (s)	<p>Sophiko Tskvitinidze, Assistant Professor, Doctor of Biology E.MAIL: tskvitinidze.s@gmail.com TEL: +995 593 744 133 /+ 995 599 122 633 Consultation days and time: individually, according to the agreement</p>					
Educational course format	Lecture, Work in Group					
Educational course format	<p>Total: 150 hours Contact hours: 75 h, among them</p> <ol style="list-style-type: none"> Lecture – 25 h Group work – 46 h Midterms – 2 h Final exam -2 h <p>Independent work – 75 h</p>					
Prerequisites	MED 2011					
The purpose (s) of tutorial course/modules	<p>The aim of the course is to give the students essential knowledge in medical genetics, which helps them to understand vital processes being in human body and reach the reason of pathological processes and pathology in inheritance.</p> <p>Course will help students in future working activities, to indicate modern methodology for diagnostics and treatment of genetic pathologies, as well they will be able to explore not only patient, but whole family and family history to define heredity or predisposition characteristic of pathology and ability to give genetic consultations.</p>					
Teaching and learning strategy	<p>Lectures - Monologue, explanation, demonstration (video, Power-point);</p> <p>Work in group -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;</p> <p>Abstract – for abstract preparation student will use a method of working on the books, besides that he/she will read publications additionally, search for additional material to write the abstract and present it as a slide show</p> <p>Discussion – students will be divided into groups in order to discuss the specific issue. The aim of</p>					

this form of the activity is to develop the culture of listening to the opponent, critical thinking, analyzing, and on the basis summing up opinions and conclusion skills.

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 scores- 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;

	<p>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; 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) – total 20 scores. 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 style="text-align: center;">Final Exam -40</p> <p>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>
<p>The basic literature</p>	<ol style="list-style-type: none"> 1. David L. Rimoin's, Reed E.Pyeritz, 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. 3. Robert L. Nussbaum; Roderick R.McInnes; Huntington F Willard. Thompson & Thompson Genetics in Medicine. Elsevier Saunders. VII ed, 2007.

The auxiliary literature	<ul style="list-style-type: none"> Jocelyn E.Krebs, Elliott S.Goldstein, Stephen T.Kilpatrick Lewi's Genes XI, Jones and Barlett Learning, 2014
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The tutorial/learning course content

Nº	Subjects	Lecture (hour) 20	Work in group (hour) 50
1	Review of human genetic research methods; Genealogy- examples of genealogic maps, twin method, etc).	2	2
2	Nucleic acid analysis methods (NDA isolation, PCR, electrophoresis), protein analysis as a result of gene expression.	2	4
3	Mutations. Genome, chromosome and gene mutations. Review of mutation based genetic diseases.	2	4
4	Mitochondrial genome. Maternal inheritance. Mitochondrial genome mutations and diseases.	2	3
5	Genetic polymorphism. Antigens; Genetic markers. Blood group genetics and importance for transfusion medicine.	2	4
6	Genetic variability in individuals and in populations. Gene polymorphism. Ethnic specificity of genetic disease.	2	3
	Midterm Exam		2
7	Genetic predisposition, parental role in genetically predisposed disease development (cancer, diabetes etc) and disease prevention	2	4
8	Epigenetic.	2	2
9	Terratogenesis and mutagenesis.	2	3
10	Gene carting and identification. Disease causing gene identification.	2	3
11	Genetic enginery and medicine.	2	3
12	Genetic diagnostics. Genetic testing. genetic consultations and population screening	1	4
13	Treatment and supervision of genetic diseases. Considerable issues in treatment. Treatment strategies. Gene therapy. Perspectives.	1	4
14	ELS issues in clinical genetics.	1	3
	Final Exam		2

Learning Outcomes

Criteria	Competences
Knowledge and understanding	<p>Student will know:</p> <ul style="list-style-type: none"> - The basis of genetic diseases; - Possible diagnostic and treatment ways; - The essence of maternal inheritance; - Ethno specificity of some diseases is depends of the distribution of different alleles.\; - the role of epigenetic factors, teratogenes and mutagenesis in disease development. - the capabilities of gene enginery for medicine gene therapy perspectives and will have sufficient knowledge to give consultations in ethic frames.
Applying knowledge	<p>Student will able to:</p> <ul style="list-style-type: none"> - estimate the situation and make the independent clinical consultation within the ethic norms ; - define disease rate and inherit characteristics in different populations.
Communication Skills	<p>Student will able to:</p> <p>Working in group, listening, questions asking will make student develop free communications skills, which combines the relations with patients and their families as with auditorium.</p>
Learning ability	<p>Student will be able to plan and manage his own learning process, deep his knowledge by means of using information technologies.</p>