

**MED 3002 –Genetic Diseases**

Course Name	Code	Semester	Type of course	Theory (hours)	Work in Group (hours)	ECTS
Genetic Diseases	MED 3002	V	Mandatory	25	46	5
Faculty, the educational program and education level	Faculty of Medicine, one-cycle Educational Program “Medicine”					
Author (s)	<b>Sophiko Tskvitinidze</b> ,– Assistant Professor, Dr. of Biology E.MAIL: <a href="mailto:tskvitinidze.s@gmail.com">tskvitinidze.s@gmail.com</a> TEL: +995 593 744 133 /+ 995 599 122 633 Consultation days and time- individually					
Educational course format	Lecture, work in group					
Educational course Loading	<b>Total:</b> 150 acad. hours <b>Contact hours:</b> 75 h, that includes: <ol style="list-style-type: none"> <li>1. Lecture – 25 h</li> <li>2. Team work – 46 h</li> <li>3. Midterms – 2 h</li> <li>4. Final exam -2 h</li> </ol> <b>Independent work</b> – 75 h					
Prerequisites	MED 2011-2012 Medical Genetics I, II					
The purpose (s) of tutorial course/modules	The course aims to introduce common and rare genetic diseases, explain their molecular-genetic and patho-physiological basis, which enable students to distinguish various genetic diseases and obtain insight of current and promising solutions based on research and treatment ways and enable to seek the right ways to solve problem.					
Teaching and learning strategy	<b>Lectures</b> - Monologue, explanation, demonstration (video, Power-point); <b>Group working</b> -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 will be assessed; <b>Group discussion/Problem based learning</b> – Students will divide in the groups and discuss around topic, express their opinions. The aim is the listening, skills of debate and critical thinking, analyzing, reconciliation. <b>Abstract preparation and presentation</b> - Students choose material from provided problematic topics or independent way, process sufficient material, will work with the books and presents in auditorium for estimation. <b>Discussion</b> - students will be divided into two groups to discuss specific issues with the purpose to					

	<p>improve the following skills: listening, critical thinking, analysis, debate and exchange of point of views on the basis of conclusions skills development.</p> <p><b>Consultation</b> – individual support work (weekly) with students through advices recommendations, answer the questions to achieve a good results of study</p>
<p><b>Assessment criteria</b></p>	<p><b>Maximum score- 100</b>, that includes:</p> <ol style="list-style-type: none"> <li><b>1. Midterm assessment -60 scores:</b> <ul style="list-style-type: none"> <li>• Attendance -10 scores;</li> <li>• Activity – 10 scores:</li> <li>• Discussion – 10 scores;</li> <li>• Abstract preparation and presentation -10 score</li> <li>• <b>Midterm Exam – 20 scores</b></li> </ul> </li> </ol> <p>The final score for group work activity is calculated by the arithmetic average.</p> <p><b>Group Work are Assessed Based on the Following Criteria</b> (maximum 10 point)</p> <p><b>10 scores-</b> 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.</p> <p><b>9 scores</b> - 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.</p> <p><b>8 scores</b> - Student has been able to bring forward a consistent knowledge, Has properly developed terminology. Demonstrates knowledge of the main readers.</p> <p><b>7 scores</b> - 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.</p> <p><b>6 scores</b> - 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.</p> <p><b>5 scores</b> - 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.</p> <p><b>4 scores</b> - Student demonstrates general overview of the topics. Terminology is not developed. Information sufficiently linked with the topic. Demonstrate irrelevant understanding of the literature.</p> <p><b>3 points</b> – Student demonstrates general/superficial and inconsistent knowledge of the subject. No sufficient knowledge of the literature.</p> <p><b>2 scores</b> - Student demonstrates general comments, no knowledge of the terminology, no consistency.</p> <p><b>1 score</b> – Student demonstrates insufficient answer, not terminology awareness, chronologic manner of the answer, mostly wrong, no knowledge of literature.</p> <p><b>0 score:</b> Student demonstrates not even elementary knowledge of the topics.</p> <p>Abstract preparation and presentation’s criteria (10 scores - max.):</p>

1. Actuality of appointed problem – 1 score;
2. Academic content - 1 score;
3. Literature data's observation in the frame of subject -1score;
4. Correspondence between the research methods and research purpose -1 scores;
5. Coherence of argumentation- 1 score;
6. Correctness of conclusion and the connection with the main text - 1 score;
7. The presented matter's visual and technical aspects - 1 score;
8. Debating and listening culture - 1 score;
9. Accuracy and reliability of indicated references and literature sources – 1 score;
10. Proper language and speaking style – 1 score.

**Discussion assessment criteria (10 scores- max.)**

1. Argumentativeness of represented factual material -2 scores;
2. Complexity of represented factual material -2 scores;
3. Discussion activity – 2 scores;
4. Debating and listening culture - 2 scores;
5. Keeping within the time-limit – 2 scores.

**Midterm Exam – 20 scores**

(Written test -40 questions, 0,5 score for 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.

**Final Exam -40**

Is held in the written test form (test consists of 80 questions, each question is rated as 0,5 score).

The final exam would accounted as passed in case of maximum 70% or more ( $40 \times 70 / 100 = 28$  scores).

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:**

- (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; (FX) Failure - 41-50 scores,

**Negative rate:**

(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.

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.

<b>The basic literature</b>	<ol style="list-style-type: none"> <li>1. David L. Rimoin's, Reed E.Pyeritz, Bruce R.Korf. <b>Essential Medical Genetics</b>. Elsevier Saunders, 2013;</li> <li>2. Jocelyn E.Krebs, Elliott S.Goldstein, Stephen T.Kilpatrick <b>Lewi's Genes XI</b>, Jones and Barlett Learning, 2014.</li> <li>3. Robert L. Nussbaum; Roderick R.McInnes; Huntington F Willard. <b>Thompson &amp; Thompson Genetics in Medicine</b>. Elsevier Saunders. VII ed, 2007.</li> </ol>
<b>The auxiliary literature</b>	<ol style="list-style-type: none"> <li>1. B. ALBERTS, A. JONSON, J. LEWIS, M. RAFF, K. ROBERTS, P. WALTER. MOLECULAR BIOLOGY OF THE CELL. FIFTH EDITION, 2008</li> <li>2. CYTOLOGY/CYTOGENETICS, HUMANA PRESS TOTOWA NEW JERSEY, SEC. EDDIT., 2005.</li> </ol>

### The tutorial/training course content

№	Subjects	Lecture	Work in group
		(hour)	(hour)
		25	50
1	Genetic diseases and medicine, problems and challenges. Dysmorphology. Genetic testing and applications in medicine. Genome sequencing method.	2	4
2	Human karyotype. Chromosomes. Congenital Anomalies and Prenatal diagnostics. Techniques for chromosome analysis.	2	4
3	Aneuploidy. Maternal age influence the origin of aneuploidy in humans. Patho-physiology and genetics of various aneuploidic diseases (trisomy 21(down syndrome), trisomy 18 (Edwards syndrome), trisomy 13 (patau syndrome), Klinefelter syndrome (47,XXY); Monosomy - Turner syndrome (45,X)); Evaluation of dysmorphic child.	3	4
4	Abnormalities of chromosome structure. Translocations and other rearrangements: Inversion and copy number variants 15q11-q13 duplications (Huntington's disease), ring chromosomes; Deletions - 22q11.2 deletion syndrome (Velocardiofacial/DiGeorge syndrome), Williams syndrome (7q11deletion syndrome). Aberrations have changed over the past few years.	3	6
5	Single-gene disorders. Mendelian inheritance and monogenic traits – Autosomal dominant (myotonic dystrophy) and autosomal recessive diseases (Albinism, Alkaptonuria etc); Genealogic analysis of genetic diseases.	3	4
6	Sex-linked dominant and sex-linked recessive genetic diseases; Hemophilia, genetic deafness etc. Mutations, rare genetic diseases (Rett syndrome (RTT)	2	4
	<b>Midterm Exam</b>		2
7	Non-Mendelian inheritance; Imprinting - Uniparental Disomy (UPD), mosaicism, mitochondrial disorders. Complex genetic disorders. Multi-factorial diseases.	2	4

8	Cancer genetics.	2	4
9	Teratogens: Maternal Medications, Maternal Drug Exposure, Maternal Infections, Maternal illness. Influence of environment and environmental pollution to human genetic structure.	2	4
10	Available Treatment of genetic disorders. Personalized medicine. Family based research. Treatment strategy. Promising gene therapy.	2	4
11	Newborn screening. Genetic consultations. ELSI implications.	2	4
	<b>Final Exam</b>		2

### Learning Outcomes

Criteria	Competences
Knowledge and understanding	<p>After completing the current course student will have a deep knowledge of the following issues :</p> <ul style="list-style-type: none"> <li>• Molecular basis of genetic diseases ;</li> <li>• Rare and common genetic diseases ;</li> <li>• Risk assessment and consultation;</li> <li>• Problem-solving ways;</li> </ul>
Applying knowledge	<p>Student will be able to:</p> <ul style="list-style-type: none"> <li>• realize and define an inheritance characteristic of genetic diseases;</li> <li>• evaluate the risks of revealing some genetic pathology in generations and define prevention strategies for at-risk families.</li> </ul>
Communication Skills	<p>Student will be able to provide consulting and prevention measures for some of genetic diseases within the ethic frames in a right way.</p>
Life-long learning ability	<p>Will be able to acquire, access and update knowledge independently, accept new strategies and technologies for better future.</p>
Value	<p>Will be able to:</p> <ul style="list-style-type: none"> <li>• treat patients without emotion in spite of any expression of the disease;</li> <li>• express tolerance towards patients and the community to promote the establishment of a similar attitude.</li> </ul>