

# MOLECULAR AND CHROMOSOMAL BASIS OF GENETIC DISEASES

1	Course Title:	MOLECULAR AND CHROMOSOMAL BASIS OF GENETIC DISEASES	
2	Course Code:	TTB5007	
3	Type of Course:	Optional	
4	Level of Course:	Second Cycle	
5	Year of Study:	1	
6	Semester:	1	
7	ECTS Credits Allocated:	7.00	
8	Theoretical (hour/week):	3.00	
9	Practice (hour/week):	0.00	
10	Laboratory (hour/week):	0	
11	Prerequisites:	None	
12	Language:	Turkish	
13	Mode of Delivery:	Face to face	
14	Course Coordinator:	Prof. Dr. ÜNAL EGELİ	
15	Course Lecturers:	Prof. Dr. Ünal EGELİ Prof. Dr. Gülşah ÇEÇENER Dr. Öğr. Üyesi Işıl Ezgi ERYILMAZ	
16	Contact information of the Course Coordinator:	Prof. Dr. Ünal EGELİ egeli@uludag.edu.tr Tıbbi Biyoloji Anabilim Dalı 0224 295 41 51	
17	Website:		
18	Objective of the Course:	Understanding the molecular basis of genetic diseases	
19	Contribution of the Course to Professional Development:	Anticipating genetic diseases, contributing to genetic counseling	
20	Learning Outcomes:		
		1	Understanding the molecular and chromosomal bases of hereditary types of diseases
		2	Understanding the changes occurring at molecular and chromosomal levels and associating them with related diseases
		3	To have knowledge about prenatal diagnosis and genetic counseling
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21	Course Content:		
		<b>Course Content:</b>	
Week	Theoretical	Practice	
1	Molecular genetic basis of inherited diseases		
2	Genotypic and phenotypic features of hereditary diseases		

<b>3</b>	Genetic Counseling	
<b>4</b>	Forms of inheritance	
<b>5</b>	Examples of hereditary diseases at the gene level	
<b>6</b>	Structure of human chromosomes	
<b>7</b>	Cytogenetic bases of chromosomal diseases	
<b>8</b>	Structural chromosome anomalies and related diseases	
<b>9</b>	Numerical chromosome anomalies and related diseases	
<b>10</b>	Diseases resulting from sex chromosome anomalies	
<b>11</b>	Cytogenetic diagnostic methods of chromosomal diseases	
<b>12</b>	Prenatal diagnosis of chromosomal diseases	
<b>13</b>	Prenatal diagnosis of molecular diseases	
<b>14</b>	Prenatal diagnosis of molecular diseases	

Assessment			Number	Duration (hour)	Total Work Load (hour)
Activites					
Theoretical Quiz	0	0.00	14	3.00	42.00
Practicals/Labs			0	0.00	0.00
Self study and preperation			14	6.00	84.00
Final Exam	1	100.00			
Homeworks			3	16.00	48.00
Projects			0	0.00	0.00
Contribution of Term (Year) Learning Activities to		0.00			
Field Studies			0	0.00	0.00
Midterm exams			0	0.00	0.00
Contribution of Final Exam to Success Grade		100.00			
Others			4	10.00	40.00
Final Exams			1	2.00	2.00
Measurement and Evaluation Techniques Used in the			1		
Semester final exam					
Total Work Load					216.00
Total ECTS//3W					7.20
ECTS Credit of the Course					7.00

<b>Contribution Level:</b>	<b>1 very low</b>	<b>2 low</b>	<b>3 Medium</b>	<b>4 High</b>	<b>5 Very High</b>
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