



## AYDIN ADNAN MENDERES UNIVERSITY COURSE INFORMATION FORM

Course Title		Hereditary Diseases							
Course Code		TL071		Course Level		Short Cycle (Associate's Degree)			
ECTS Credit	2	Workload	54 (Hours)	Theory	2	Practice	0	Laboratory	0
Objectives of the Course		To learn the causes, symptoms, diagnosis, treatment of inherited diseases and have knowledge about prevention.							
Course Content		The terms of genetic and heredity, mutation and its types, mutagens, chromosomal, monogenic and poligenic defects, polyploidy, aneuploidy, trisomi types of somatic chromosomes, sex chromosome aneuploidies, diseases caused by mutation in genes, sex-linked inherited diseases, clinical laboratory evaluation and screening tests, prevention							
Work Placement		N/A							
Planned Learning Activities and Teaching Methods				Explanation (Presentation), Discussion, Case Study, Problem Solving					
Name of Lecturer(s)		Ins. Adem KESKİN, Ins. Tuğçe OKTAV							

### Assessment Methods and Criteria

Method	Quantity	Percentage (%)
Midterm Examination	1	40
Final Examination	1	70

### Recommended or Required Reading

1	Klug, W.S. and Cummings, M.R. 2002. Genetik Kavramlar. Palme yayınevi, Ankara, 816 s.
2	Başaran, A. 2010. Tıbbi Biyoloji, Ders kitabı. Pelikan yayıncılık, Ankara. 584 s.
3	Bozcuk, A.N. 2000. Genetik. Palme yayıncılık, Ankara, 320 s
4	Campbell, N.A. and Reece, J.B. 2008. Biyoloji. Palme yayıncılık, Ankara, 1247 s.

Week	Weekly Detailed Course Contents	
1	Theoretical	The terms of genetic and heredity
2	Theoretical	Chromosomes
3	Theoretical	Mutation
4	Theoretical	Chromosomal, monogenic and poligenic defects
5	Theoretical	Changes in chromosome number: Polyploidy, aneuploidy
6	Theoretical	Trisomy types of somatic chromosomes: Patau syndrome, Edward syndrome, Down syndrome
7	Theoretical	Sex chromosome aneuploidies: Turner syndrome, Klinefelter syndrome, Jacobs syndrome
8	Intermediate Exam	Midterm
9	Theoretical	Autosomal Dominant Diseases: Huntington's disease, Marfan Syndrome
10	Theoretical	Autosomal Recessive Diseases: Phenylketonuria, Tay-Sachs Disease
11	Theoretical	Autosomal Recessive Diseases: SMA (Spinal Muscular Atrophy)
12	Theoretical	Sex-linked inherited diseases
13	Theoretical	Diseases Related to X Chromosome
14	Theoretical	Diseases Related to Y Chromosome
15	Theoretical	Diseases Related to X and Y Chromosomes

### Workload Calculation

Activity	Quantity	Preparation	Duration	Total Workload
Lecture - Theory	14	0	2	28
Individual Work	12	1	0.5	18
Midterm Examination	1	3	1	4
Final Examination	1	3	1	4
Total Workload (Hours)				54
[Total Workload (Hours) / 25*] = ECTS				2

\*25 hour workload is accepted as 1 ECTS



**Learning Outcomes**

1	To learn the terms belong to genetic and heredity.
2	To learn mutation and mutagens.
3	To learn chromosomal inherited diseases .
4	To learn inherited diseases related to gene mutation.
5	To learn diagnosis, treatment process of inherited diseases and prevention .

**Programme Outcomes (Anesthesia )**

1	To be able to recall basic knowledge about human anatomy
2	To be able to recall the knowledge about Ataturk's principles and the history of Turkish Revolution
3	To be able to recall the knowledge about ethical and moral values
4	To be able to recall the knowledge of Turkish grammar and be able to use it
5	To be able to communicate effectively with patient, their family, and own team
6	To be able to control, use, and maintain the anesthesia machines
7	To be able to recall the information about anesthesia application in the system diseases
8	To be able to recall the issues that needed to be considered in follow-up of patients in intensive care.
9	To be able to make the patients' care in intensive care
10	To be able to apply the cardiopulmonary resuscitation.
11	To be able to apply the drug, liquid and blood to the patient.
12	To be able to apply nasogastric tube to the patient and to aspirate.
13	To be able to assist the implementation of general anesthesia to patient.
14	To be able to recall the drugs used in general and regional anesthesia and learn to use them safely.
15	PO15. Can help during the maintenance, ending and post anaesthesia process.
16	Can help the practices of anesthesia and sedation outside the operation room.
17	Can communicate at the basic level of a foreign language and use this language in his job.
18	Be able to communicate at a basic level in a foreign language and be able to use this language in professional fields
19	To have the appropriate knowledge of basic sciences at the level of interest, to use specific medical terms and terminology of field

**Contribution of Learning Outcomes to Programme Outcomes** 1:Very Low, 2:Low, 3:Medium, 4:High, 5:Very High

	L1	L2	L3	L4	L5
P7	3	3	3	3	3
P11	3	3	3	3	3

