



## 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	Trisomi 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: Phenolketonuria, Tay-Sacs 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** (*Medical Documentation and Secretarial Practices* )

1	Write and pronounce medical terms correctly.
2	Collect data of medical statistics and report the results periodically after analyzing them scientifically.
3	Learn basic structure of human body and important system diseases.
4	Know principles and rules of medical secretaryship. Protect patients' rights and privacy.
5	Use Turkish and body language in a correct and effective way.
6	Run internal and external correspondence of the foundation, keep the files of the documents after classification, organize them and archive in line with filing techniques.
7	Establish verbal and written communication inside the foundation and out of foundation.
8	Make the coding procedure of diseases and health problems according to existing international classification systems.
9	Run the counseling services for patients and their relatives.
10	Solve the problems that are encountered in work life quickly and effectively.
11	Use the necessary equipment for professional practices such as computer and office devices effectively.
12	Improve professional knowledge and skills continuously.
13	Executes any patient registration-documentation processes
14	Makes archiving operations
15	Prepares medical documents
16	Knows Turkish history and Atatürk's revolutions.
17	Adapt to team work in application areas.
18	Knows and defines diseases.
19	Have general information about the world and biological formations.
20	English speaking writer

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

	L1	L2	L3	L4	L5
P18	4	4	4	4	4

