



## AYDIN ADNAN MENDERES UNIVERSITY COURSE INFORMATION FORM

Course Title		Hereditary Diseases							
Course Code		TL071		Couse Level		Short Cycle (Associate's Degree)			
ECTS Credit	2	Workload	54 ( <i>Hours</i> )	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 (Physiotherapy)**

1	To be able to recall the information of research methods and statistics so as to follow the developments, monitor and interpret scientific literature
2	To have the appropriate knowledge of basic sciences at the level of interest, to use specific medical terms and terminology of physical therapy
3	To be able to recall knowledge of the general structure and properties of musculoskeletal system and the joints and to evaluate the story of musculoskeletal diseases.
4	To be able to comprehend the methods of measurement of the range of motion of joints and to measure it.
5	To be able implement a general evaluation of posture analysis and gait analysis.
6	To be able to recall the knowledge about general characteristics of musculoskeletal diseases, osteoporosis, osteoarthritis, rheumatoid arthritis, ankylosing spondylitis, especially rheumatic diseases, mechanical low back and neck pain, disc herniation, soft tissue disorders and to apply appropriate physiotherapy.
7	To be able to recall the knowledge and gain skills about the devices and the agents of heater used in physical therapy, indications and contraindications of using, and the necessary information about how to apply on patients.
8	To be able to recall the knowledge of the electromagnetic field.
9	To be able to recall what Elektroakapunktur, Laser, Biofeedback, cervical and lumbar traction systems, pneumatic compression therapy are, and how to apply them, which one is applicable to patients.
10	To be able to recall what manipulation-mobilization is and which patients are suitable for this application.
11	To be able to recall what massage and hydrotherapy treatments are and which patients are suitable for these applications.
12	To be able to gain the professional and ethical awareness, apply gained knowledge and skills in real life situations and transfer gained knowledge to individuals around her/his environment, and improve behavior of life-long learning.
13	To gain knowledge about methods of diagnosis, protection and treatment of diseases
14	To be able to recall the knowledge and gain skills about physical therapy and rehabilitation methods to be applied to neurological disorders.
15	To be able to recall the knowledge and gain skills about physical therapy and rehabilitation methods to be applied to cardiopulmonary disorders.
16	To be able to recall the knowledge and gain skills about physical therapy and rehabilitation methods to be applied to pediatric patients.
17	To be able to gain knowledge about the effects of fitness and exercise on metabolism and responses of body systems to them.
18	To have knowledge about rehabilitation services
19	To become individuals who can do interdisciplinary team work, with a sense of social responsibility and entrepreneur.
20	To be able to recall the knowledge about Atatürk's Principles and the History of Turkish Revolution.
21	To be able to gain the knowledge and ability to become contemporary individuals who can use Turkish language grammar well and know a foreign language knowledge necessary to follow the developments in the profession.

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

	L1	L2	L3	L4	L5
P2	3	3	3	3	3
P14	3	3	3	3	3

