



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 (Dialysis)

1	To be able to comprehend the duties and responsibility of dialysis technicians. To be able to work in a team with members of other health professions.
2	To be able to acquire a general knowledge of human anatomy, physiology and biochemistry
3	To be able to gain knowledge of blood-borne infectious diseases, especially infectious diseases such as hepatitis and universal prevention methods
4	To be able to have knowledge of blood-borne infectious diseases, especially infectious diseases such as hepatitis and universal prevention methods
5	To be able to recognize hemodialysis machine, and have knowledge and skills will be used it during operation of dialysis
6	To be able to have the knowledge of application on peritoneal dialysis and skills be able to train patient on this.
7	To be able to acquire dialysate characteristics, have necessary skills on preparation and application
8	To be able to gain the knowledge and skills on the basic principles of water treatment, application methods, and control of purified water as a level of practitioner
9	To be able to comprehend the principles of patient care, complications during dialysis operation what patients may be encounter and perform necessary knowledge and skills to take necessary measures to protect patient from these complications.
10	To be able to gain knowledge and equipment related to educating on problems that the long-term dialysis patients may have.
11	To be able to understand periodic examinations during the follow up dialysis patients and recognize pathologies in the early period, and have the knowledge and skills to take necessary precautions in time
12	To be able to have the knowledge of the dialysis patients, physiological, social and psychological problems, and perform necessary support skills on these issues for the patient
13	In general to be able to comprehend the knowledge of, drugs, dosage, side effects, and toxic effects, routes of administration of drugs and drug use in patients with chronic renal failure
14	To be able to acquire a high level knowledge of fluid and electrolyte problems with general issues nephrology, acid-base balance disorder, nephrology and urology kidney disease, chronic and acute renal failure.
15	To be able to comprehend the methods of diagnosis and treatment of diseases of the system, and have knowledge of fighting and protecting from especially problems that can be seen in dialysis patients as level of practitioner and getting patient compliance.
16	To be able to have knowledge of statistics and research methods as a level of following the developments, monitoring and interpreting scientific publications.
17	To be able to gain the knowledge of foreign language as a level of communicating and following developments.
18	To be able to be willing to self-improvement as an individual committed to the principles and reforms of Atatürk and keeping on the some of the rules of social life, customs and traditions, depending on the interests of the country on their own interests as a member of society,

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	5	5	5	5	5
P15	5	5	5	5	5
P16	3	3	3	3	3

