



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-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 (Environmental Health)

1	They have the appropriate level of knowledge about the basic sciences which has an interaction with the environment and the environment itself.
2	They have gained the basic concepts, skills and qualifications in the Environmental health theoretical and practical lessons. And then they can establish the connections that are necessary to protect the environment and people's health in the light of these competencies.
3	They can use the approaches and the information of basic and applied research in different disciplines. They can follow the innovations and developments in their field, and have self-development competency with the terms of the day.
4	They know and apply the analysis methods used in the evaluation of environmental factors (drinking water, waste water treatment, air pollution, meteorological data, land values, food control, radiation measurement, etc.).
5	They have a professional and ethical consciousness, and have the ability to recognize the environmental problems and also can formulate a solution to these problems. They apply the gained knowledges and skills faced in real life situations, transfers the knowledge to individuals around, and wins the life-long learning behavior.
6	They are able to use their professional knowledge in their lives and behave sensitively toward the local and global environmental problems and effectively uses to the legislation and management tools the necessary for the solution.
7	Gained the ability to adapt the changing in a positive way themselves, to understand the core values and cultures of the society which are living. Sensitive to the universal and the social values, interests of the country, have adopted the concept of sustainable development, environmentally conscious, productive, behaves aware of the ethical and professional responsibility.
8	Provides a healthy interact of individual, society and the environment and take responsibility in the necessary situations for the continuity.
9	They gain the ecologically-based solving skills the problems and the delays that may arise in interaction with each other of living and nonliving environment. Interests of local and national, and Ecological and historical values of our country, and contribute to the protection and the development of them.
10	Exhibits the appropriate behaviours for the protection and the development of plants, animals, and inanimate environment, and the especially human health.
11	Knows the value of energy for life, recognizes the types of energy, and have conscious of the importance, using and dissemination of renewable energy sources.
12	Knows the properties of information and communication technologies, and uses them in the process efficiently and professionally.
13	They aware of the democracy, rule of law, human rights, the national and universal cultural characteristics, and sensitive towards to the nature, society and people.
14	Knows the importance of Atatürk's principles and reforms, make them a way of life.
15	Uses effectively the Turkish in speaking and writing.
16	Has at least one foreign language ability to be able to follow the knowledge in their profession and to communicate with colleagues.
17	To have the appropriate knowledge of medical 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
P8	4	4	4	4	4
P10	4	4	4	4	4

