COURSE OUTLINE "MOLECULAR BASIS OF GENETIC DISEASES"

1. GENERAL

SCHOOL	HEALTH SCIENCES			
DEPARTMENT	MOLECULAR BIOLOGY AND GENETICS			
LEVEL OF STUDIES	ISCED LEVEL 6			
COURSE CODE	**MBG620**	**SEMESTER**	6^{th} and 8^{th}	
COURSE TITLE	MOLECULAR BASIS OF GENETIC DISEASES			
TEACHING ACTIVITIES *If the ECTS Credits are distributed in distinct parts of the course e.g. lectures, labs etc. If the ECTS Credits are awarded to the whole course, then please indicate the teaching hours per week and the corresponding ECTS Credits.*			**HOURS/WEEK**	**ECTS CREDITS**
			2	3
COURSE TYPE *Background, General Knowledge, Scientific Area, Skill Development*	SCIENTIFIC AREA			
PREREQUISITES:	NO			
TEACHING & EXAMINATION LANGUAGE:	GREEK ENGLISH FOR ERASMUS STUDENTS			
COURSE OFFERED TO ERASMUS STUDENTS:	YES			
COURSE URL:	https://eclass.duth.gr/courses/418347/			

2. LEARNING OUTCOMES

Learning Outcomes

Please describe the learning outcomes of the course: Knowledge, skills and abilities acquired after the successful completion of the course.

The main learning outcome of the course is to present to the students a unified picture of the ways genomic variants that predispose to or are associated with genetic diseases affect molecular pathways and cellular mechanisms by disrupting the cellular, tissue, organ, system, or body homeostasis.

The course will approach the issue of genotype-phenotype correlation in the context of diseases with a strong genetic basis. In other words, the course aims to connect the deranged molecular mechanisms that characterize a genetic disorder and its phenotypic consequences. The scope of the course is expected to be reached via the detailed presentation and analysis of specific genetic disorders that follow patterns of Mendelian inheritance, as well as multifactorial diseases that are believed to have a prominent genetic component. Both disease categories will serve as representative examples of large disease groups, such as enzymopathies, neurodegenerative, neurodevelopmental, musculoskeletal, mitochondrial, as well as multifactorial diseases.

Upon successful completion of the course, students are expected to:

- Be able to correlate the genetic causes/basis of an inherited disease (pattern of inheritance, type of genetic variation/mutation, etc) with (a) particular biochemical/molecular pathway(s) or mechanism(s) involved,
- Understand how the presence of genetic variation(s) that cause disruption or derangement of the normal function of a biochemical/molecular pathway can affect one or more crucial cellular functions,
- Comprehend the concept of genotype-phenotype correlation at the level of tissue, organ, system, and organism,
- Realize the multidisciplinarity that characterizes the efforts to unravel the pathogenetic mechanisms that underlie the inherited diseases, as well as the efforts to manage and/or treat these diseases with state-of-the-art and emerging novel molecular approaches.

General Skills

Name the desirable general skills upon successful completion of the module

Search, analysis and synthesis of data and information,
ICT Use
Adaptation to new situations
Decision making
Autonomous work
Teamwork
Working in an international environment
Working in an interdisciplinary environment
Production of new research ideas
Project design and management
Equity and Inclusion
Respect for the natural environment
Sustainability
Demonstration of social, professional and moral responsibility and sensitivity to gender issues
Critical thinking
Promoting free, creative and inductive reasoning

- Analysis and synthesis of data and information
- Autonomous- and team-work
- Equity and Inclusion
- Production of new research ideas
- Development of critical thinking
- Promoting free, creative and inductive reasoning

3. COURSE CONTENT

1. Introduction to the patterns of inheritance of human traits and diseases
2. Types of human genome variation – Laboratory techniques for analyzing human genome variation
3. The molecular basis of genetic diseases I (enzymopathies, metabolic disorders)
4. The molecular basis of genetic diseases II (receptors, membrane proteins, other structural/tissue-specific proteins)
5. The molecular basis of genetic diseases III (nervous system: neurodegenerative, neurodevelopmental diseases)
6. The molecular basis of genetic diseases IV (mitochondrial diseases, genetic imprinting diseases)
7. The genetic component of multifactorial diseases in human (basic concepts)
8. The molecular basis of genetic diseases V (multifactorial diseases: cardiometabolic, autoimmune, others)
9. The molecular basis of genetic diseases VI [multifactorial diseases: neurodevelopmental, psychiatric – Neuroimaging genetics]
10. Animal models that contributed to the unraveling of the pathophysiology of human genetic diseases
11. State-of-the-art and emerging molecular therapies for genetic diseases (examples)
12. Oral presentation of key scientific publications from students (I) – Focus on articles that are worth being presented because they depict the interdisciplinary investigation and methodology that led to the unraveling of the molecular/cellular basis of a genetic disease
13. Oral presentation of key scientific publications from students (II), as above

4. LEARNING & TEACHING METHODS - EVALUATION

TEACHING METHOD *Face to face, Distance learning, etc.*	Face to face
USE OF INFORMATION & COMMUNICATIONS TECHNOLOGY (ICT) *Use of ICT in Teaching, in Laboratory Education, in Communication with students*	Use of ICT in teaching Use of ICT in communication with students
TEACHING ORGANIZATION *The ways and methods of teaching are described in detail.* *Lectures, Seminars, Laboratory Exercise, Field Exercise, Bibliographic research & analysis, Tutoring, Internship (Placement), Clinical Exercise, Art Workshop, Interactive learning,*	

Activity	***Workload/semester***
Lectures	26

Study visits, Study / creation, project, creation, project. Etc. *The supervised and unsupervised workload per activity is indicated here, so that total workload per semester complies to ECTS standards.*	Independent study, bibliographic research & analysis	44
	Oral presentation (project)	20
	Course Total	**90**

STUDENT EVALUATION *Description of the evaluation process* *Assessment Language, Assessment Methods, Formative or Concluding, Multiple Choice Test, Short Answer Questions, Essay Development Questions, Problem Solving, Written Assignment, Essay / Report, Oral Exam, Presentation in audience, Laboratory Report, Clinical examination of a patient, Artistic interpretation, Other/Others* *Please indicate all relevant information about the course assessment and how students are informed*	**Student evaluation languages** Greek, English **Method (Formative or Concluding)** Concluding **Student evaluation methods** Written exam with multiple choice test (50%) Written Exam with Short Answer Questions (30%) Written Exam with Essay Development Questions (10%) Presentation in audience (10%)

5. SUGGESTED BIBLIOGRAPHY

1) Concepts of Genetics, Authors: Klug, Cummings, Spencer, Palladino

2) Thompson & Thompson – Genetics in Medicine, Authors: Nussbaum, McInnes, Willard

3) Lecture slides and course notes available to students via DUTH e-class