

## COURSE OUTLINE “HUMAN GENETICS”

### 1. GENERAL

<b>SCHOOL</b>	HEALTH SCIENCES		
<b>DEPARTMENT</b>	MOLECULAR BIOLOGY AND GENETICS		
<b>LEVEL OF STUDIES</b>	ISCED LEVEL 6		
<b>COURSE CODE</b>	<b>MBG401</b>	<b>SEMESTER</b>	7 <sup>th</sup>
<b>COURSE TITLE</b>	HUMAN GENETICS		
<b>TEACHING ACTIVITIES</b> <i>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.</i>		<b>HOURS/WEEK</b>	<b>ECTS CREDITS</b>
		4	6
<b>COURSE TYPE</b> <i>Background, General Knowledge, Scientific Area, Skill Development</i>	BACKGROUND		
<b>PREREQUISITES:</b>	NO (but it is desirable that the students have successfully completed previous courses in the subject of Genetics)		
<b>TEACHING &amp; EXAMINATION LANGUAGE:</b>	GREEK ENGLISH FOR ERASMUS STUDENTS		
<b>COURSE OFFERED TO ERASMUS STUDENTS:</b>	YES		
<b>COURSE URL:</b>	<a href="https://eclass.duth.gr/courses/ALEX01146/">https://eclass.duth.gr/courses/ALEX01146/</a>		

### 2. LEARNING OUTCOMES

<p><b>Learning Outcomes</b> <i>Please describe the learning outcomes of the course: Knowledge, skills and abilities acquired after the successful completion of the course.</i></p> <p>In the undergraduate curriculum of the Department of Molecular Biology and Genetics, the course “Human Genetics” is the last of the taught 5 courses in the field of Genetics. For completing and fully benefiting from this course, it is necessary that students have developed a deep understanding of Genetics concepts and phenomena, as can be gained from “Genetics I” and “Genetics II”, “Genomics” and “Population Genetics”.</p> <p>This course aims to present the principles of Genetics as they apply to Humans. Very large part of the course concerns the role of Genetics in human health and the interactions between basic scientific knowledge and clinical application. The genetic, molecular and chromosomal basis of inherited diseases, cancer, multifactorial diseases with a genetic component, as well as other pathological conditions are presented extensively. Teaching includes a presentation of the basic principles, but also a very extensive use of examples. The course covers the disciplines of genetic diagnostics, genetic counselling, genetic ethics, laboratory genetics and the emerging field of genetic medicine and molecular medicine.</p> <p>Upon successfully completing this Human Genetics course, the students should be able to:</p> <ol style="list-style-type: none"> <li>1. comprehend the important role of Human Genetics in health &amp; disease and its interaction with medicine</li> <li>2. understand the rules and exceptions to the phenomena of heredity in humans and apply this knowledge to the analysis of pedigrees (family trees)</li> <li>3. understand the molecular, chromosomal, protein, cellular basis of genetic disease</li> <li>4. know about developmental disorders and their genetic etiology</li> <li>5. collaborate with laboratory scientists and doctors of various specialties, as a team dedicated to the diagnosis and treatment of genetic diseases</li> <li>6. provide genetic counseling and contribute to the management of actual cases of genetic disease</li> </ol>
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7. understand the principles of bioethics in relation to Human Genetics and how these are applied in research and clinical practice for the treatment of genetic diseases
8. know the principles and tools of human genetics and genomics in order to plan and carry out research for the identification, mapping and cloning of human disease genes
9. present the basic approaches to management and treatment of genetic diseases, as well as the research directions for the development of pharmaceutical and modern advanced biotechnological treatments

#### **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*

Search, analysis and synthesis of data and information

Adaptation to new situations

Decision making

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### **3. COURSE CONTENT**

The course content (syllabus) includes the following topics:

Introduction to Human Genetics – the role of Genetics in Health

Mendelian inheritance in humans

Haemoglobinopathies and thalassaemias

Molecular, biochemical, and cellular basis of human genetic disease

Pharmacogenetics

Cytogenetics – principles, methods, concepts

Cytogenetics – disorders involving autosomal chromosomes

Cytogenetics – disorders involving sex chromosomes

Human Population Genetics

Complex (multi-factorial) inheritance and disease

Clinical genetics and genetic counselling

Prenatal diagnosis

Mapping genes involved in human genetic disease

Positional cloning

Human Genome Project

Ethics in Human Genetics – dilemmas in research and clinical practice

Gene therapy for genetic disease

### **4. LEARNING & TEACHING METHODS - EVALUATION**

#### **TEACHING METHOD**

*Face to face, Distance learning, etc.*

Face to face

<p align="center"><b>USE OF INFORMATION &amp; COMMUNICATIONS TECHNOLOGY (ICT)</b></p> <p align="center"><i>Use of ICT in Teaching, in Laboratory Education, in Communication with students</i></p>	<p>Use of ICT in Teaching, in communicating with students and in practical training.</p> <p>Use of the eClass platform for communication, for organising student essays and homework, for sharing lecture notes, literature.</p> <p>Use of MS Teams for video calls</p>															
<p align="center"><b>TEACHING ORGANIZATION</b></p> <p><i>The ways and methods of teaching are described in detail.</i></p> <p><i>Lectures, Seminars, Laboratory Exercise, Field Exercise, Bibliographic research &amp; analysis, Tutoring, Internship (Placement), Clinical Exercise, Art Workshop, Interactive learning, Study visits, Study / creation, project, creation, project. Etc.</i></p> <p><i>The supervised and unsupervised workload per activity is indicated here, so that total workload per semester complies to ECTS standards.</i></p>	<table border="1"> <thead> <tr> <th align="center"><b>Activity</b></th> <th align="center"><b>Workload/semester</b></th> </tr> </thead> <tbody> <tr> <td>Lectures</td> <td align="center">130</td> </tr> <tr> <td>Other Interactive teaching</td> <td align="center">10</td> </tr> <tr> <td>Essay preparation and presentation</td> <td align="center">10</td> </tr> <tr> <td>Bibliographic research &amp; analysis</td> <td align="center">20</td> </tr> <tr> <td>Seminars</td> <td align="center">10</td> </tr> <tr> <td><b>Course Total</b></td> <td align="center"><b>180</b></td> </tr> </tbody> </table>		<b>Activity</b>	<b>Workload/semester</b>	Lectures	130	Other Interactive teaching	10	Essay preparation and presentation	10	Bibliographic research & analysis	20	Seminars	10	<b>Course Total</b>	<b>180</b>
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<p align="center"><b>STUDENT EVALUATION</b></p> <p><i>Description of the evaluation process</i></p> <p><i>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</i></p> <p><i>Please indicate all relevant information about the course assessment and how students are informed</i></p>	<p>Examinations take place typically in Greek, but can be in English if there is an international audience (e.g. Erasmus+ students).</p> <p>Concluding evaluation in the form of a written examination</p> <p>The examination may contain a mixture of multiple choice questions, true or false questions, short answer questions, longer (essay) questions, problems solving, etc. Example of a typical distribution:</p> <p>Student evaluation methods (%)</p> <p>Written exam with multiple choice test (20)</p> <p>Written Exam with Short Answer Questions (30)</p> <p>Written Exam with Essay Development Questions (30)</p> <p>Written Exam with Problem Solving (20)</p> <p>Students are informed about the course requirements, evaluation, assessment, and examination style during the first week; this info is repeated at the last lecture.</p> <p>All relevant information can be found on eClass.</p>															

## 5. SUGGESTED BIBLIOGRAPHY

The following bibliography was used in the planning and preparation of this course:

- Σύγχρονη Κλινική Ιατρική — Andrew Read, Dian Donnai — εκδ. Πασχαλίδης
- Αρχές Ιατρικής Γενετικής — Gelehrter, Collins, Ginsburg — εκδ. Π.Χ. Πασχαλίδης
- Γενετική Ανθρώπου — Κ. Τριανταφυλλίδης και Αναστ. Κουβάτση — εκδ. Π.Χ. Πασχαλίδης
- Στοιχεία Εφαρμοσμένης Γενετικής του Ανθρώπου — Δ. Κ. Μουρελάτος — Β΄ έκδοση, εκδ. University Studio Press
- Γενετική – Βασικές Αρχές – Snustad & Simmons – εκδ. Τζιόλα
- Βασικές Αρχές Γενετικής – Klug, Cummings, Spencer, Palladino – Ακαδημαϊκές εκδ.
- iGenetics - μια Μεντελική Προσέγγιση – P.J. Russel – Ακαδημαϊκές εκδ.
- Κλασική και Μοριακή Γενετική — Κων. Τριανταφυλλίδης — εκδ. Κυριακίδη
- Γενετική τόμος Α΄ — Μ. Γ. Λουκάς — εκδ. Σταμούλης

- Εισαγωγή στη Γενετική – Στ. Αλαχιώτης – εκδ. Ελληνικά Γράμματα
- Ανασυνδυασμένο DNA — Watson, Myers, Caudy, Witkowski— Ακαδημαϊκές εκδ.
- Genes VIII — Benjamin Lewin — Ακαδημαϊκές εκδ.
- DNA I – Το Ανθρώπινο Γονιδίωμα — Carina Dennis & Richard Gallagher — εκδ. Πασχαλίδης
- DNA II – 50 Χρόνια DNA — Julie Clayton & Carina Dennis — εκδ. Πασχαλίδης
- New Clinical Genetics — Read, Donnai — εκδ. Scion (2007)
- Introduction to Genetic Analysis — 8th edition — Griffiths, Wessler, Lewontin, Gelbart, Suzuki, Miller — εκδ. Freeman
- Concepts of Genetics — 7th edition — William S. Klug & Michael R. Cummings — εκδ. Prentice Hall
- Analysis of Genes and Genomes — R. J. Reece — εκδ. Wiley
- Molecular Biology of the Gene — Watson, Hopkins, Roberts, Steitz, Weiner — εκδ. Benjamin/Cummings
- Principles of Genetics — Gardner, Simmons, Snustad — εκδ. Wiley
- Recombinant DNA — Watson, Gilman, Witkowski, Zoller — εκδ. Scientific American Books
- Genetic Mapping of Disease Genes — Pawlowitzki, Edwards, Thompson — εκδ. Academic Press
- Genome Mapping - A Practical Approach — Paul Dear — εκδ. IRL Press
- Genome — Matt Ridley — εκδ. Harper Perennial
- Nature via Nurture — Matt Ridley — εκδ. Harper Collins Publishers
- A Passion for DNA — James D. Watson — εκδ. Oxford
- DNA – the secret of life — James D. Watson — εκδ. Knopf
- The Common Thread — John Sulston & Georgina Ferry — εκδ. Bantam Press

Recommended textbooks through the “Eudoxus” platform (textbooks available in Greek)  
 Ιατρική Γενετική Thompson & Thompson — Nussbaum, McInnes, Willard — εκδ. Π. Χ. Πασχαλίδης  
 2011 — κωδικός στον "Εύδοξο" 13256587

Μοριακή Γενετική του Ανθρώπου — Γεώργιος Δεδούσης — εκδ. Utopia 2021 — κωδικός στον  
 "Εύδοξο" 102075373

Μοριακή Γενετική του Ανθρώπου — Tom Strachan και Andrew P. Read — εκδόσεις Broken Hill  
 Publishers Ltd 2024 — κωδικός βιβλίου στον Εύδοξο: 112691149