

## COURSE OUTLINE “LABORATORY 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>MBG523</b>	<b>SEMESTER</b>	7 <sup>th</sup>
<b>COURSE TITLE</b>	LABORATORY 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>	
	2	3	
<b>COURSE TYPE</b> <i>Background, General Knowledge, Scientific Area, Skill Development</i>	SCIENTIFIC AREA		
<b>PREREQUISITES:</b>	NO		
<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/418343/">https://eclass.duth.gr/courses/418343/</a>		

### 2. LEARNING OUTCOMES

<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>The scope of the course is to provide students with the theoretical background in the field of laboratory genetics and to present applications of this discipline that are relevant throughout human life (in preimplantation testing, prenatally, postnatally, during childhood/adolescence/adult life), also in view of the newly established specialty of “Laboratory Genetics”.</p> <p>The course will approach basic concepts for the theoretical training of a future laboratory geneticist, including the following subjects: a) popular methodologies and techniques applied in genetic diagnosis and testing, b) the spectrum and the purpose of the various genetic tests currently available, c) the interpretation of results and the means to mine relative information from databases and data repositories, d) good practices of communicating test results to individuals, e) genetic counseling approaches preceding and following genetic testing, f) the current status and common practices in the field of genetic diagnostics in our country and abroad.</p> <p>Upon successful completion of the course, students are expected to:</p> <ul style="list-style-type: none"> <li>• Be able to connect a genetic syndrome or inherited disorder (based on the type of chromosomal aberration or molecular genetic variation/mutation) with one or more appropriate laboratory techniques applied in genetic diagnostics, as well as the analytical potential and limitations of each technique,</li> <li>• Know the different categories of genetic tests that are available or suitable in each case, which can be performed by a trained laboratory geneticist, as well as the benefits and limitations of each testing category,</li> <li>• Have become familiar with the genetics nomenclature of human genome variation, as well as the genetic/genomic databases, repositories and computational tools available for mining relevant information,</li> <li>• Have a comprehension of the basic principles of genetic counseling, including taking a family history, constructing a pedigree, developing specific skills when communicating with the individuals/patients/family members involved,</li> </ul>

- Know the current status and the requirements/conditions of acquiring the specialty of “Laboratory Genetics” in Greece.

### 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
- Autonomous- and team-work
- Decision making
- Equity and Inclusion
- Production of new research ideas
- Demonstration of social, professional and moral responsibility and sensitivity to gender issues
- Critical thinking
- Promoting free, creative and inductive reasoning

### 3. COURSE CONTENT

1. Popular laboratory techniques applied in genetic diagnosis and testing (karyotyping, molecular genetics/genomics, molecular karyotyping)
2. Principles of genetics nomenclature (human genome variation nomenclature) – Examples from cytogenetics and molecular genetics
3. Genetic tests (sensitivity, specificity, precision, CE-IVD, SOP, ISO, etc)
4. Categories of genetic tests (I) – Prenatal, preimplantational, presymptomatic, diagnostic (when and how) (invasive and non-invasive sampling approaches)
5. Categories of genetic tests (II) – Forensics, Pharmacogenetics, Nutrigenetics – D-t-C testing
6. Categories of genetic tests (III) – Cancer (familial and sporadic)
7. Genetic/Genomic databases and genetic tests/accredited diagnostic laboratories databases // Literature repositories // Online demonstration of the use of genomic databases and computational tools (interactive)
8. Reporting genetic tests results // Case studies
9. Genetic counseling (taking a family history, drawing genetic pedigrees, calculating genetic predisposition, testing minors, issues of consent, bioethics, personal data protection-GDPR)
10. Clinical genetics (Medical Genetics)/Referral for genetic tests – Case reports and discussion with a guest clinical geneticist (interactive)
11. A diagnostics lab in Greece (I) – Case reports (eg cytogenetics) & discussion with a guest laboratory geneticist (interactive)
12. A diagnostics lab in Greece (II) – Case reports (eg molecular genetics) & discussion with a guest laboratory geneticist (interactive)
13. Laboratory Genetics Specialty – Presentations from students

### 4. LEARNING & TEACHING METHODS - EVALUATION

<b>TEACHING METHOD</b> <i>Face to face, Distance learning, etc.</i>	Face to face
<b>USE OF INFORMATION &amp; COMMUNICATIONS TECHNOLOGY (ICT)</b> <i>Use of ICT in Teaching, in Laboratory Education, in Communication with students</i>	Use of ICT in teaching Use of ICT in communication with students

<p><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>	<b>Activity</b>	<b>Workload/semester</b>
	Lectures	26
	Independent study, Bibliographic research & analysis	55
	Seminars	6
	Oral Presentation	3
	<b>Course Total</b>	<b>90</b>
<p><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><b>Student evaluation languages</b> Greek, English</p> <p><b>Method (Formative or Concluding)</b> Summative</p> <p><b>Student evaluation methods</b> Written exam with multiple choice test (50%) Presentation in audience (40%) Written Exam with Short Answer Questions (10%)</p>	

## 5. SUGGESTED BIBLIOGRAPHY

1. **Genetic tests: Understanding genetic testing and its result.** Authors: Georgitsi, Katafigiotis, Kostoulas, Lambropoulos, Sakaloglou, Syrrou, 1<sup>st</sup> edition, University Studio Press, Thessaloniki, 2023. ISBN 978-960-12-2630-9 (Greek edition only)
2. **New Clinical Genetics.** Authors: Read & Donnai, 3<sup>rd</sup> edition, SCION Publishing LTD, Banbury, 2015. ISBN 978-1-907904-67-7
3. **Thompson & Thompson Genetics in Medicine.** Authors: Nussbaum, McInnes, Willard, 8<sup>th</sup> edition, Elsevier, 2015. ISBN 978-143-770-696-3
4. **Lecture slides and course notes** available to students via the DUTH e-class platform.