



ΕΛΛΗΝΙΚΗ ΔΗΜΟΚΡΑΤΙΑ

HELLENIC REPUBLIC

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ΕΘΝΙΚΗ ΑΡΧΗ ΑΝΩΤΑΤΑΤΗΣ ΕΚΠΑΙΔΕΥΣΗΣ

HELLENIC AUTHORITY FOR HIGHER EDUCATION

## University of West Attica

School of Health and Care Sciences

Department of Biomedical Sciences and Midwifery

### Postgraduate Studies

**“Applications of Biomedical Technology in Infertility - Male and Female Factor”**

Course Outline

**GENETICS OF INFERTILITY**



ATHENS 2023



## COURSE OUTLINE

### (1) GENERAL

|  |   |                                    |         |
|--|---|------------------------------------|---------|
| SCHOOL   |   | School of Health and Care Sciences |         |
| ACADEMIC UNIT  |   | Biomedical Sciences and Midwifery  |         |
| LEVEL OF STUDIES   |   | Postgraduate Studies               |         |
| COURSE CODE  | MY1.1   | SEMESTER                           | First   |
| COURSE TITLE   |   | Genetics of infertility            |         |
| INDEPENDENT TEACHING ACTIVITIES<br><i>if credits are awarded for separate components of the course, e.g. lectures, laboratory exercises, etc. If the credits are awarded for the whole of the course, give the weekly teaching hours and the total credits</i> |   | WEEKLY TEACHING HOURS              | CREDITS |
| Review of existing literature  |   | 2                                  | 8       |
| Experimental procedure   |   |                                    |         |
| Add rows if necessary. The organisation of teaching and the teaching methods used are described in detail at (d).  |   |                                    |         |
| COURSE TYPE<br><i>general background, special background, specialised general knowledge, skills development</i>  | Skills development  |                                    |         |
| PREREQUISITE COURSES:  |   |                                    |         |
| LANGUAGE OF INSTRUCTION and EXAMINATIONS:  | Greek   |                                    |         |
| IS THE COURSE OFFERED TO ERASMUS STUDENTS  | Yes   |                                    |         |
| COURSE WEBSITE (URL)   | <a href="https://eclass.uniwa.gr/">https://eclass.uniwa.gr/</a> |                                    |         |

### (2) LEARNING OUTCOMES

|  |
|--|
| <p>Learning outcomes</p> <p><i>The course learning outcomes, specific knowledge, skills and competences of an appropriate level, which the students will acquire with the successful completion of the course are described.</i></p> <p>Consult Appendix A</p> <ul style="list-style-type: none"> <li>• Description of the level of learning outcomes for each qualifications cycle, according to the Qualifications Framework of the European Higher Education Area</li> <li>• Descriptors for Levels 6, 7 &amp; 8 of the European Qualifications Framework for Lifelong Learning and Appendix B</li> <li>• Guidelines for writing Learning Outcomes</li> </ul>   |
| <p>The purpose of the course is to teach the students, at a theoretical and practical level, methods that facilitate the understanding of the modern approach to the genetic investigation of infertility in humans.</p> <p>After successfully completing the course, students will:</p> <ul style="list-style-type: none"> <li>• be aware of the genetic factors in the justification of infertility in humans</li> <li>• be able to know basic principles that govern the Genetics of reproduction and in particular the genetic causes of infertility in humans, as well as how to investigate them.</li> <li>• have proven knowledge and understanding that supports the existence of genetic causes that account for a large proportion of "unexplained" infertility in humans. At the same time, they will acquire the background to understand the methods used by modern science to diagnose genetic causes of infertility.</li> <li>• Be able to use their knowledge and skills to guide and counsel people with unexplained infertility towards the possible investigation of genetic causes.</li> </ul> |

### General Competences

*Taking into consideration the general competences that the degree-holder must acquire (as these appear in the Diploma Supplement and appear below), at which of the following does the course aim?*

*Search for, analysis and synthesis of data and information, with the use of the necessary technology*  
*Adapting to new situations*  
*Decision-making*  
*Working independently*  
*Team work*  
*Working in an international environment*  
*Working in an interdisciplinary environment*  
*Production of new research ideas*

*Project planning and management*  
*Respect for difference and multiculturalism*  
*Respect for the natural environment*  
*Showing social, professional and ethical responsibility and sensitivity to gender issues*  
*Criticism and self-criticism*  
*Production of free, creative and inductive thinking*  
*.....*  
*Others...*  
*.....*

- Search, analysis and synthesis of data and information, using the necessary technologies
- Adaptation to new situations
- Project planning
- Autonomous work
- Teamwork

### (3) SYLLABUS

1. Principles of Human Genetics (1)
2. Principles of Human Genetics (2)
3. Genetics of Reproduction (1)
4. Genetics of Reproduction (2)
5. Chromosomal abnormalities
6. Gene variants (mutations)
7. Prenatal screening and diagnostic options
8. Fertility and Genetics
9. Genetic factors of Infertility
10. Assisted Reproduction and Genetics
11. Pre-conception diagnosis and selection
12. Preimplantation diagnosis and selection
13. The science of Genetic Counseling

#### Laboratory/Tutorial Exercises

1. Isolation of genetic material from biological materials
2. Laboratory diagnosis of hemoglobinopathies
3. Laboratory diagnosis of thrombophilia
4. Laboratory diagnosis of cystic fibrosis
5. Laboratory diagnosis of Beta thalassemia

#### Educational Software

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#### (4) TEACHING and LEARNING METHODS - EVALUATION

|   |  |                          |
|---|--|--------------------------|
| <p><b>DELIVERY</b><br/><i>Face-to-face, Distance learning, etc.</i></p>   | <p>In the classroom and in the Lab face to face.</p>   |                          |
| <p><b>USE OF INFORMATION AND COMMUNICATIONS TECHNOLOGY</b><br/><i>Use of ICT in teaching, laboratory education, communication with students</i></p>   | <ul style="list-style-type: none"> <li>➤ CytoScape, GenMAPP, SeqMap, FASTA, DNADot</li> <li>➤ Learning process support through the electronic platform e-class, Microsoft Teams, Skype Business</li> </ul>   |                          |
| <p><b>TEACHING METHODS</b><br/><i>The manner and methods of teaching are described in detail.</i><br/><i>Lectures, seminars, laboratory practice, fieldwork, study and analysis of bibliography, tutorials, placements, clinical practice, art workshop, interactive teaching, educational visits, project, essay writing, artistic creativity, etc.</i></p> <p><i>The student's study hours for each learning activity are given as well as the hours of non-directed study according to the principles of the ECTS</i></p>                            | <b>Activity</b>  | <b>Semester workload</b> |
|   | Lectures   | 42                       |
|   | Laboratory/Tutorial Exercises  | 20                       |
|   | Interactive Teaching   | 13                       |
|   | Study and Analysis of the Literature   | 26                       |
|   | Preparation of Study   | 26                       |
|   | Writing Assignment   | 26                       |
|   | Independent Study  | 47                       |
|   | <b>Course total</b>  | <b>200</b>               |
| <p><b>STUDENT PERFORMANCE EVALUATION</b><br/><i>Description of the evaluation procedure</i></p> <p><i>Language of evaluation, methods of evaluation, summative or conclusive, multiple choice questionnaires, short-answer questions, open-ended questions, problem solving, written work, essay/report, oral examination, public presentation, laboratory work, clinical examination of patient, art interpretation, other</i></p> <p><i>Specifically-defined evaluation criteria are given, and if and where they are accessible to students.</i></p> | <ol style="list-style-type: none"> <li>1. Written final exam (60%) which includes: <ul style="list-style-type: none"> <li>• Multiple choice questions</li> <li>• Short Answer Questions</li> <li>• Problem solving</li> </ul> </li> <li>2. Presentation of Individual or Group Work (40%)</li> </ol> |                          |

## (5) ATTACHED BIBLIOGRAPHY

### Suggested bibliography:

1. Toshinobu Miyamoto et.al. *Reprod Med Biol.* 2017;16:81–88, Human male infertility and its genetic causes.
2. Cariati et al. *J Transl Med* (2019) 17:267. The evolving role of genetic tests in reproductive medicine.
3. Elizabeth A. Normand et.al. *Fertility and Sterility®* Vol. 109, No. 2, February 2018, Exome and genome sequencing in reproductive medicine
4. Fahad Alsohime et. al. *N Engl J Med* 382;3 [nejm.org](http://nejm.org) January 16, 2020, JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency
5. Daniela Lorenzi et.al. *JBRA Assisted Reproduction* 2020;24(2):104-114, First custom next-generation sequencing infertility panel in Latin America: design and first results  
Gheldof A, et al. *J Med Genet* 2019;56:271–282, Genetic diagnosis of subfertility: the impact of meiosis and maternal effects

- Related academic journals: