**COURSE OUTLINE**

1. **GENERAL**

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| **SCHOOL** | School of Health Sciences | | | | |
| **ACADEMIC UNIT** | Faculty of Medicine | | | | |
| **LEVEL OF STUDIES** | Undergraduate | | | | |
| **COURSE CODE** | **ΙΑΕ704** | **SEMESTER** | | **4th** | |
| **COURSE TITLE** | **MEDICAL GENETICS IN CLINICAL PRACTICE, PREVENTION AND PRENATAL GENETICS** | | | | |
| **INDEPENDENT TEACHING ACTIVITIES** *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* | | | **WEEKLY TEACHING HOURS** | | **CREDITS** |
|  | | | 2 | | 2 |
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| *Add rows if necessary. The organisation of teaching and the teaching methods used are described in detail at (4).* | | |  | |  |
| **COURSE TYPE**  *general background,  special background, specialised general knowledge, skills development* | Special background  Specialised general knowledge | | | | |
| **PREREQUISITE COURSES:** |  | | | | |
| **LANGUAGE OF INSTRUCTION and EXAMINATIONS:** | Greek | | | | |
| **IS THE COURSE OFFERED TO ERASMUS STUDENTS** |  | | | | |
| **COURSE WEBSITE (URL)** | https://ecourse.uoi.gr/course/view.php?id=185 | | | | |

1. **LEARNING OUTCOMES**

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| **Learning outcomes** | |
| *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.*  *Consult Appendix A*   * *Description of the level of learning outcomes for each qualifications cycle, according to the Qualifications Framework of the European Higher Education Area* * *Descriptors for Levels 6, 7 & 8 of the European Qualifications Framework for Lifelong Learning and Appendix B* * *Guidelines for writing Learning Outcomes* | |
| The course introduces the student in the expertise of human genetics in clinical practice and in approaches for the genetic investigation in human reproduction. The course content introduces the student in understanding clinical problems which involve inherited and congenital factors and how to diagnose and prevent the birth of children with genetic conditions.  In addition, an outline of laboratory diagnostic methods and their rapid expansion through the years is provided, which introduces the student in latest diagnostic tools and methods.  Finally, the combination of clinical and laboratory practice will give an overview for the management of genetic conditions in the current practice of clinical genetics.  At the end of the course the student will be able to:   * Understand the basic and critical characteristics of genetic conditions * create a family pedigree and understand the mode of inheritance of a genetic condition and recommend special tests * recognize and appreciate the need for gamilial genetic evaluation * be able to distinguish key characters in a real case or case study of a genetic disease and appreciate their role in diagnosis * recognize and understand the different categories of genetic tests such as: prenatal testing, pre-implantation diagnosis (of Monogeneic or Chromosomal Diseases) * Understand laboratory tools and techniques for the identification of individuals who are carriers of genetic conditions * Identify major features for several genetic conditions and evaluate their role in the diagnosis * Use the testing methods for the identification of carriers of a genetic condition and individuals with high risk for genetic disorders * propose ways of diagnostic approach to a genetic disease that they will face in their clinical practice (e.g. in cases of frequent inherited diseases (e.g. beta-thalassemia), in couples of reproductive age or during pregnancy or when undergoing methods assisted, in families with children with congenital abnormalities of mental retardation and neurodevelopmental disorder * Analyze and calculate the major genetic characteristics of a genetic condition and associate them with the effective testing method * understand the role of genetic counseling in the prevention of genetic diseases * be able to navigate and search Medical Genetics databases (OMIM, DECIPHER, NCBI, Human Phenotype Ontology, ENCODE, exac, PharmGKB) * Collaborate with his/her colleague for the presentation and the management of a case report | |
| **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…*  *…….* |
| * Working independently * Team work * Clinical and laboratory thinking * Development of diagnostic practice for the management of cases with genetic diseases * Search for, analysis and synthesis of data and information, with the use of the necessary technology * Showing social, professional and ethical responsibility and sensitivity to gender issues | |

1. **SYLLABUS**

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| Most common monogenic disorders in clinical practice.  Genetic family history / family pedigree - symbols  Common polygenic and complex disorders, patterns of inheritance and clinical manifestation.  Chromosomal abnormalities in clinical practice.  Clinical and genetic history-clinical aspects of genetic counseling.  Clinical disorders of sex development and differentiation.  Clinical genetics and investigation of pregnancy loss.  Clinical genetics and assisted reproduction.  Prenatal diagnosis  Biochemical markers of fetal abnormalities in maternal blood and ultrasound evaluation of congenital abnormalities.  Non-invasive prenatal diagnosis-clinical application.  Clinical genetic databases in genetic counseling. |

1. **TEACHING and LEARNING METHODS - EVALUATION**

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| **DELIVERY** *Face-to-face, Distance learning, etc.* | Lectures in the auditorium/hall  face to face |
| **USE OF INFORMATION AND COMMUNICATIONS TECHNOLOGY** *Use of ICT in teaching, laboratory education, communication with students* | - Use of ICT in teaching, PowerPoint presentations   * Use of literature search engines HEALLINK, PUBMED, SCOPUS, Medline, GOOGLE SCHOLAR, and Databases – genetic information analysis tools * Use of the e-course platform for communication with students, and for posting lectures, scientific articles, instructions, useful links, evaluation questionnaires, information on attending conferences and seminars related to the course, etc. |
| **TEACHING METHODS**  *The manner and methods of teaching are described in detail.*  *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.*  *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* | |  |  | | --- | --- | | ***Activity*** | ***Workload of each students group*** | | Lectures | 26 | | Tutorial exercises | 10 | | Study preparation and literature analysis | 20 | | Independent study | 20 | | total | ***76*** | |
| **STUDENT PERFORMANCE EVALUATION**  *Description of the evaluation procedure*  *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*  *Specifically-defined evaluation criteria are given, and if and where they are accessible to students.* | Written final exam with multiple-choice, short-answer questions, which assesses the student's understanding of the knowledge offered and critical thinking  Rating scale: 1-10  Upgradable grade: 5 |

1. **ATTACHED BIBLIOGRAPHY**

*Teaching - study material*

Medical Genetics. Thompson & Thompson, R.L. Nussbaum, R.R. McInnes, H.F. Willard, Broken Hill Publishers Ltd

*- Related academic journals:*

Genetics in Medicine

European Journal of Human Genetics

Am Journal of Human Genetics

Clinical Genetics

Plos Genetics

Nature Genetics