**COURSE OUTLINE**

1. **GENERAL**

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| **SCHOOL** | Health Sciences |
| **ACADEMIC UNIT** | Biological Applications and Technology |
| **LEVEL OF STUDIES** | Undergraduate |
| **COURSE CODE** | **BEE805** | **SEMESTER** | **6th , 8 th** |
| **COURSE TITLE** | INTRODUCTION TO (MEDICAL) 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** |
|  | 3 | 3 |
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| *Add rows if necessary. The organisation of teaching and the teaching methods used are described in detail at (d).* |  |  |
| **COURSE TYPE***general background, special background, specialised general knowledge, skills development* | Special backgroundSkills development |
| **PREREQUISITE COURSES:** | Basic Genetics ΒΕΥ403Molecular Genetics ΒΕΥ601 |
| **LANGUAGE OF INSTRUCTION and EXAMINATIONS:** | Greek |
| **IS THE COURSE OFFERED TO ERASMUS STUDENTS** | YES (in English and French) |
| **COURSE WEBSITE (URL)** | http://ecourse.uoi.gr/course/view.php?id=343 |

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*
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| By the end of this course students should be able to:* take a family history and construct a pedigree
* interpret a pedigree
* understand a scientific publication
* know where to find up-to-date and reliable information, ( web sites, Databases, e.g. PubMed, OMIM, Orphanet e.t.c)
* understand the main modes of Mendelian and non- Mendelian inheritance
* understand the clinical implications and potential impact on diagnosis of genetic phenomena: penetrance, expressivity, new mutations, mosaicism, variation in expression, heterogeneity
* understand the advantages and limitations of cytogenetic, molecular cytogenetic and molecular techniques . Case presentations are included to illustrate the genetic approaches and serve as paradigms.
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| **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…**…….* |
| * Analysis and synthesis of data and informationwith *the* use *of the* necessary technology*.*
* Individual work
* Team work
* Promotion of the free, creative and inductive thinking, through the development of critical skills, as well as the understanding of the principles and the interactions of the various genetic phenomena.
* Understanding of ethics, social responsibility and diversity and demonstration of social, professional and ethical responsibility and sensitivity to diversity issues.
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1. **SYLLABUS**

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| **Pedigrees**- Take a family history and construct a pedigree-Modes of Mendelian and non-Mendelian inheritance-Penetrance, expressivity, mosaicism and their clinical implications **Cytogenetics**-Mitosis, meiosis, karyotype. -Normal and abnormal karyotypes - Numerical and structural chromosome abnormalities -Sex determination and abnormalities.**Mendelian disorders**. -DNA, genes, mutations.-Genomes- Cytogenetic and molecular diagnostic techniques.**-**Mendelian disorders and their mode of inheritance (dominant, recessive, X-linked dominant and recessive, Y-linked)-gene frequency, founder effect-Genotype-Phenotype-loss of function, gain of function, happloinsufficiency, dominant negative effects, dosage sensitivity etc**Epigenetics**- epigenetic regulation, genomic imprinting, CpG islands, uniparental disomy , - DNA methylation, **Linkage analysis**- Meiosis and recombination, introduction and principles of linkage analysis, DNA markers- **Multifactorial inheritance** -Principles of multifactorial inheritance-polygenic, multifactorial traits (genes, environment)-multifactorial vs single-gene inheritance-congenital malformations-Predisposition and predisposing factors -Genetic and environmental modifiers of single gene disorders **Genetic tests**-Diagnostic, prognostic, screening tests -Techniques and approaches **Cancer Genetics**-Oncogenes, tumor suppressor genes , genomic instability -Cancer cytogenetics-Sporadic, familial cases. -Predisposition**Genetic disorders**- Genetic counselling- Prenatal diagnosis-Genetic approaches to the treatment of diseases-Personalized genetic medicine-Ethical issues in the application of genetic information**Web tools** |

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

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| **DELIVERY***Face-to-face, Distance learning, etc.* | Students work in small groups in the classroom and they are actively involved in the process. Learning is achieved with individual work and teamwork as well and constant interaction between students and teacher. |
| **USE OF INFORMATION AND COMMUNICATIONS TECHNOLOGY** *Use of ICT in teaching, laboratory education, communication with students* | e-course, videos |
| **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* |

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| ***Activity*** | ***Semester workload*** |
| Lectures | 26 |
| Individual written tasks  | 18 |
| Collaborative tasks | 6 |
| Study | 20 |
| **Course Total** | **70** |

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| **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.* | Ι.Continuous evaluation through compulsory work given weekly (80%)ΙΙ. Final written and oral evaluation (20%) |

1. **ATTACHED BIBLIOGRAPHY**

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| *- Suggested bibliography:**- Related academic journals:*1. Hartwell LH, Hood L, Goldberg ML, Reynolds AE, Silver LM.Γενετική Εκδόσεις UTOPIA 2014.
2. Read A, Donnai D. Σύγχρονη Κλινική Γενετική . Εκδόσεις Πασχαλίδη 2010.
3. Scientific Journals
4. Databases
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