**SYLLABUS**

1. **Programme Details**

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| **1.1.** | **GRIGORE T. POPA UNIVERSITY OF MEDICINE AND PHARMACY IASI** |
| **1.2.**  | **FACULTY : MEDICINE / DEPARTMENT: MEDICINE OF THE MOTHER AND CHILD** |
| **1.3.** | **DISCIPLINE: GENETICS** |
| **1.4.**  | **FIELD of STUDY: MEDICINE** |
| **1.5.** | **STUDY CYCLE: BACHELOR**  |
| **1.6.** | **PROGRAMME of STUDY: English**  |
| 1. **Discipline Details**
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| **2.1.** | **Name of the Discipline: GENETICS** |
| **2.2.** | **Teaching staff in charge with lectures: Prof. dr. Cristina Rusu** |
| **2.3.** | **Teaching staff in charge with seminar activities: Şef lucr. dr. Monica Pânzaru** |
| **2.4. Year**  | **II** | **2.5. Semester** | **I/II** | **2.6. Type of evaluation**  | E1/E2 | **2.7. Discipline regimen**  | **mandatory** |

1. **Overall Time Estimates (hours/semester of didactic activity)**

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| * 1. **Number of hours per week**
 | 6 | **Of which: 3.2. lectures** | 2 | * 1. **seminar/ laboratory**
 | 4 |
| * 1. **Total hours in the curriculum**
 | 84 | **Of which: 3.5. lectures** | 28 | **3.6. seminar/ laboratory** | 56 |
| **Distribution of time**  |  |  |  |  | Hours |
| **Study time using coursebook materials, bibliography and notes**  | **30** |
| **Further study time in the libray, online and in the field** | **5** |
| **Preparation time for seminars / laboratories, homework, reports, portfolios and essays** | **25** |
| **Tutoring** |  |
| **Examinations** | 6 |
| **Other activities** |  |
| **3.7. Total hours of individual study** |  | 66 |
| **3.8. Total hours / semester** |  | 150 |
| **3.9. Number of credits**  |  | 6 |

1. **Prerequisites (where applicable)**

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| **4.1. curriculum** | Basic notions of biochemistry and cell biology |
| **4.2. competences** | No case |

1. **Conditions (where applicable)**

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| **5.1. for lecture delivery** | videoprojector |
| **5.2. for seminar / laboratory delivery** | No case |

1. **Specific Competences Acquired**

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| **Professional Competences (knowledge and skills)** | * Human karyotype: realization and interpretation
* Sex chromatin: realization and interpretation
* Recognizing the most frequent chromosomal genetic syndromes
* Genetic counseling in chromosomal genetic diseases
* Paternity analysis
* Realization of maternal-fetal and familial anamnesis and drawing the family pedigree
* Recognizing the pattern of inheritance of monogenic hereditary diseases
* Genetic counseling in monogenic diseases
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| **Transversal Competences (roles, personal and professional development)** | * Teamwork skills
* The ability to communicate with patients
* Compliance with bioethical principles
* Open to self-improvement
* IT skills
* decision-making and problem solving skills
* the ability to critically evaluate informations
* the ability to deal with complex medical situations and collaborate with colleagues in other specialties
* Effective use of information sources and communication resources and training assistance, both in Romanian and in an international language
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1. **Obiectives of the Discipline (related to the acquired competences)**

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| **7.1. General Obiective** | Skills necessary to apply knowledge of current practice in medical genetics |
| **7.2. Specific Obiectives**  | Indications and interpretations of genetic test, diagnosis of genetic disorders, principles of genetic counseling, methods and indication for prenatal diagnosis |

1. **Contents**

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| **8.1. Lecture** | **Teaching methods**  | **Comments** |
| **I. HUMAN GENETICS AND IT’S ROLE IN MEDICINE**  **A. Human Genetics’ content.** **B. Human, heredity and environment.** | **ppt** | 1 hour |
| **II.DNA STRUCTURE AND CELLULAR ORGANISATION**  **A. DNA – molecular substrate of heredity.** **B. DNA structure.** **C. Nuclear genome** **D. Mitochondrial DNA** | **ppt** | 3 hours |
| **III. GENE STRUCTURE, LOCALIZATION AND IDENTIFICATION**  **A. Classical theory on gene structure.** **B. Actual theory on gene structure** **C. Gene isolation, cloning and analysis. (recombinant DNA technology)** **D. Gene localization –** principles of gene mapping | **ppt** | 2 hours |
| **IV. GENETIC INFORMATION EXPRESSION (GENE FUNCTION)**  **A. Classical theory on gene function** **B. Actual theory on gene function.** | **ppt** | 2 hours |
| **V. INHERITANCE OF GENETIC INFORMATION**  **A. DNA replication.** **B. Inheritance of monogenic traits** **C. Polygenic/ multifactorial inheritance.**  | **ppt** | 2 hours |
| **VI. GENETIC VARIABILITY**  **A. Definition. Sources.** **B. Gene mutations.** | **ppt** | 2 hours |
| **VII. GENERAL DATA CONCERNING GENETIC DISORDERS**  **A. Heredity- environment interaction as cause of human disorders** **B. Genetic disorders****C. Chromosomal disorders*.*** **D. Monogenic disorders.** **E. Molecular disorders** **F. Multifactorial disorders** **G. Developmental Genetics. Birth defects (congenital anomalies).**   **H. Inter-sexual states (ambiguous genitalia).** **I. Cancer Genetics** **J. Prevention of genetic disorders** **K. Treatment of genetic disorders** | **ppt** | 16 hours |
| **Bibliography**Elements of medical genetics; Autor: Cristina Rusu; Editura: Performantica – Institutul National de Inventica, Iasi, 2014 |
| **8.2. Seminar / Laboratory** | **Teaching methods**  | **Comments** |
| ***WEEK I*** Genetic apparatus of the cell. Cell cycle. Nucleus during cell division. | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK II*** Mitosis. Meiosis. Division and recombination errors  | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK III*** Sexual chromatin | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK IV*** Human chromosomes**:** methods to obtain chromosomes, identification. | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK V*** Chromosomal abnormalities: classification, mechanisms, consequences | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK VI*** Chromosomal syndromes. Indications for human chromosomes study | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK VII*** Determinism of normal hereditary traits. Monogenic traits. Blood groups | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK VIII*** Polygenic traits. Practical value of the study of normal hereditary traits. | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK IX*** Genetic disorder. Genetic counselling. Family history | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK X*** Monogenic disorders: characteristics, ways of transmission (autosomal dominant / recessive disorders and X- linked disorders). | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK XI*** Multifactorial disorders:characteristics, empirical risks. Hardy-Weinberg law | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK XII*** Genetic counselling: aims and circumstances | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK XIII*** Genetic counselling: genetic risk categories | Oral presentation, ppt, exercises | 4 hours |
| ***WEEK XIV*** Molecular diagnosis**.** Prenatal diagnosis. Neonatal screening | Oral presentation, ppt, exercises | 4 hours |
| **Bibliography**Medical genetics - practical lessons; Autor: Cristina Rusu, Monica Panzaru; Editura: Performantica – Institutul National de Inventica, Iasi, 2014 |

1. **Correlations between the contents of the discipline and the expectations of the epistemic community, of profesional associations and of employers in the field**

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| Curricula and knowledge offered to students are adapted to facilitate the integration of medical genetics issues into the larger concept of molecular medicine. Based on the medical genetics knowledge, students are able to understand the importance of medical genetics specialty in the core curriculum of medical specialties, the need of genetic tests in human pathology and the importance of prevention to limit the harmful effects of mutations. Curricula and knowledge are consistent with what is done in other universities in the country and abroad. Standardization of curricula and adaptation to new knowledge are elements discussed in periodic meetings of Romanian Society of Medical Genetics. |  |

1. **Evaluation**

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| **Type of activity**  | **10.1. Evaluation criteria:**  | **10.2. Methods of evaluation** | **10.3. Percentage of final grade** |
| **10.4. Lecture** | Grade for multiple choice test | standardized multiple choice test | 50% |
| **10.5. Seminar / Laboratory** | Average grade of ongoing examinations | ongoing evaluation | 10% |
| Grade for practical examination | practical exam | 40% |
| **Minimum standard of performance: at least grade 5 to pass the discipline** |

**Date: 1.10.2019 Signiture of Didactic Co-ordinator**

**Prof.dr. Eusebiu Vlad Gorduza**

 **Signiture of Department Director Conf.dr. Mihaela Grigore**