**OPTIONAL DISCIPLINES - OFFER**

**UNIVERSITY YEAR 2020-2021**

|  |  |
| --- | --- |
| ***Item-uri*** | ***Cerinţe*** |
| **Lecture title** | Elements of clinical genetics |
| **Objectives** | Know indications for genetic tests and basic notions concerning genetic counselling, prenatal screening and diagnosis, prevention and therapy; Recognize inheritance pattern |
| **Aim group** | General Medicine 3rd year English series |
| **Participants** | Minimum 30; maximum 70 |
| **Topics** | 1. Genetic disorders – classification, genetic factors involved, diagnostic tests; indications and limits of usual genetic tests ((sex chromatin, karyotype, FISH, molecular tests);
2. Genetic examination: principles, family history, pedigriu drawing and interpretation;
3. Genetic counselling: principles, steps, risk categories;
4. Prenatal diagnosis: principles, methods used for prenatal screening (biochemical, ultrasound), methods used for prenatal diagnosis (amniocentesis, chorionic villus sample, chordocentesis), major indications of prenatal screening and diagnosis;
5. Neonatal and presimptomatic screening;
6. Principles of prevention and therapy in Medical Genetics
 |
| **Bibliography** | 1. Elements of Medical Genetics – author: Cristina Rusu;2. Medical Genetics – Practical Lessons – author: Cristina Rusu |
| **Competences (abilities acquired)** | Recognize major categories of genetic disorders and genetic factors involved in every category;Know indications and limits of usual genetic tests;Able to take a corect family history and draw the pedigriu as well as its’interpretation both for genetic disorders and disorders with genetic predisposition;Use general principles of prevention and therapy for different genetic disorders |
| **Teaching methods and lecture notes** | PowerPoint presentations and lecture notes (no images with copyright), posted in E-learning platform. |
| **Responsable** | Lecturer Monica Panzaru, MD PhD |
| **Associated lecturer** | Prof. Cristina Rusu, MD PhD |
| **Keywords** | Clinical Genetics; Indications genetic tests; Prenatal screening; Recurrence risk; Genetic counselling |