**Study programme: REPRODUCTIVE BIOLOGY**

**Course title:** Medical Genetics

**Teacher:** Mihajla Djan

**Course status:** elective

**ECTS:** 5

**Requirements: -**

**Course objectives**

Course objective is to introduce to the students genetic basis of hereditary diseases in humans and available methods of diagnostics.

**Learning outcomes**

After successfully finished pre-exam and exam obligations student is able to:
- differ and describe modes of chromosome abnormalities, monogenic and multifactorial diseases;
- identify mechanisms of gene expression as basis of the human hereditary diseases;
- understand principles of medical diagnostics of hereditary diseases with different complexity and ethiology;
- desribe methodology of basic techniques of molecular genetics in medical diagnostics;
- use data bases and scientific literature in the field of medical genetics.

**Syllabus**

*Theoretical instruction*

Introduction to medical genetics. Chromosome anomalies. Changes in chromosomal number and structure as cause of infertility. Clinical cytogenetics. Autosomal monogenic diseases. Diagnostics of monogenoc diseases. Sex-linked monogenic diseases. Heritable methabolism disorders. Genetic basis of multifactorial and complex diseases. Medical diagnosis of hereditary diseases. Reccurence index. Calculation of probability. Data base search in the field of medical genetics. Gene therapy. Ethical issues in medical genetics.

*Practical instructions:*

Pedigree analysis. Clinical cytogenetics. FISH. PCR diagnosis of hereditary monogenic disease. Genotyping of monogenic disease. Association analysis genotype-phenotype. Association analysis SNP – phenotype. Bioinformatics methods in medical diagnosis. Calculation of probability for hereditary diseases. Data base search in the field of medical genetics Using of tools in data bases for estimation of genetic basis of hereditary diseases.

**Literature**

1. Turnpenny P., Ellard S., Emery's elements of medical genetics, 12th Ed., Elsevier, Churchill Livingstone, 2005.

| **Weekly teaching load** | **Lectures:** 2 | **Practical lectures:** 2+0+0 |
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**Teaching methods**

Lectures, Problem solving, Computer lectures

**Evaluation of knowledge (maximum score 100)**

| **Pre-exam obligations** | points | **Final exam** | points |
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| Student engagement in lectures | | Final test | |
| Student engagement in practical lectures | | Oral exam | 60 |
| seminars | 40 | | |