| Faculty of Medicine<br>University of Niš | Study program:<br>**INTEGRATED ACADEMIC STUDIES OF MEDICINE**<br>*ACCREDITATION 2018* | |
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**Course title: Medical genetics**

**Course head:** Prof. dr Marija Vukelić-Nikolić

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| **Course status:** | Elective |
| **Semester**: VI | **Year of study**: III |
| **ECTS: 4** | **Course code**: M-III-20.b |

**Course purpose:**

Students should acquire the knowledge of:

- Principles of disease inheritance
- Diagnosis of herteditary diseases
- Patient history in the diagnosis of hereditary diseases
- Basic principles of physical examination if a congenital disease is suspected
- Types of genetic tests
- Types of chromosomal aberrations, etiology, pathogenesis, clinical picture, diagnosis and treatment of chromosomopathies;
- Types of mutations, etiology, pathogenesis, clinical picture, diagnosis and treatment of monogenic diseases;
- Etiology, pathogenesis, clinical picture, diagnosis and treatment of multifactorial diseases;
- Hereditary factors in the onset and development of cancer;
- Genetic counselling and principles of ethics in medical genetics

**Course outcome (knowledge, skills, attitudes):**

A student should be qualified to:

- Recognize the type and mode of disease inheritance;
- Select appropriate diagnostic method;
- Interpret molecular-genetic and cytogenetic laboratory results;
- Assess/calculate the risk of disease recurrence;
- Inform the patient in accordance with the basic principles of genetic counselling;
- Search for and use genetic information which could be used in routine practice;
- Suggest appropriate measures in the prevention and treatment of particular diseases.

**Number of classes of active teaching: 45**

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| **Lectures: 30** | **Practice: 11** | **OFT: 4** |

**Course content**

*Theoretical teaching*

DNA technology: structure and DNA processes. Mutations. Recombinations.
Recombinant DNA technology. Use of the methods of genetic engineering. Methods of study of disease mechanisms. Diagnostic molecular tests. Treatment of genetic diseases. Chromosomopathies: Chromosome structure. Karyotype. Cell cycle and division. Gametogenesis. Types of chromosome aberrations. Consequences of chromosome aberrations. Aneuploidy. Polyploidy. Syndromes associated with deletions and microdeletions. Syndromes of chromosomal instability. Methods of chromosomal analysis. Prenatal diagnosis of chromosomopathies.
Monogenic diseases: Principles of inheritance. Autosome-dominant and autosome-recessive inheritance. Sex-linked inheritance. Atypical Mendelian inheritance (anticipation and mitochondrial inheritance). Monogenic diseases. Diseases of trinucleotid repetitions. Methods in the diagnosis of monogenic diseases. Biochemical genetics. Pharmacogenetics.
Multifactorial diseases: Oligogenic and polygenic inheritance. Multifactorial inheritance. Multifactorial diseases.
Oncogenetics: Oncogenes. Tumor suppressor genes. DNA repair mechanisms. Cancerogenesis. Familial cancer syndromes.
Genetic counseling. Ethics in medical genetics: Detection of carriers and presymptomatic diagnosis.

Prenatal diagnosis of genetic diseases. Risk assessment. Population screening and community genetics. Ethics in medical genetics.

*Practical teaching*

Molecular-genetic diagnostic methods. Cytogenetic diagnostic methods. Modes and types of inheritance. Recurrence risk caculations. Hereditary diseases in pediatrics. Prenatal diagnosis.

*Seminars:*

Monogenic diseases. Cancer genetics

| **3. OFT** | |
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| 1. | Seminar I: Monogenic diseases (problem-based learning) |
| 2. | Seminar II: Cancer genetics (problem-based learning) |

**Recommended literature:**

1. Turnpenny PD, Ellard S. Emerijevi osnovi medicinske genetike. Data status; 2011
2. Young DI. Medical genetics. Oxford University press; 2005.
3. Gelehrter TD, Collins FS, Ginsburg D. Principles of medical genetics. Lippincott Williams & Wilkins;1998.
4. Nussbaum RL, McInnes RR, Willard HF. Thompson & Thompson Genetics in medicine. Elsevier Health Sciences; 2015.
5. Lecture handouts.

Dopunska literatura:

1. Strachan T, Read A. Human Molecular Genetics 4. Garland Science/Taylor & Francis Group; 2011.

**Teaching methods:**

- Interactive theoretical and practical teaching
- Consultations
- Seminar papers

**Required previously passed exams:**

- Molecular and human genetics
- Biochemistry

**Grade (max. 100 points)**

**Pre-exam obligations**

- Attendance and activity at lectures: 0 – 10 points
- Activity at practice classes: 0 – 20 points
- Seminar papers: 0 – 20 points

**Final exam**

- Written exam/Oral exam: 0 – 50 points