UNIVERSITY OF NOVI SAD
FACULTY OF MEDICINE

Study program: Integrated Academic Studies in Medicine

Course title: Biochemistry and Genetics of Inherited Metabolic Diseases

Teacher: Karmen M. Stankov, Jasmina N. Katanić

Course status: elective

ECTS Credits: 3

Condition: Medical Biochemistry

Course aim

The main objective of the course is to enable the medical students to acquire the knowledge and skills about the biochemical and genetic basis of inherited metabolic diseases, as well as other inherited metabolic aberrations, with possibilities of diagnostics, prevention and therapy. This elective subject will enable students to learn about the functional and structural aberrations of chromosomes and genes mutations in inherited diseases, to learn about biochemical and molecular mechanisms of inherited diseases pathogenesis, models of inheritance and up-to-date methods of diagnostics, prevention and therapy of inherited diseases

Expected outcome of the course:

Medical students will acquire the comprehensive knowledge about the biochemical and genetic basis of inherited diseases. Skills that will be developed comprise the adequate approach to diagnostics in patients with inherited diseases, with case reports. The specificities of laboratory tests, up-to-date methods of molecular diagnostics of inherited diseases, and therapy possibilities with emphasis on prevention, including methods of pre-natal and post-natal diagnostics.

Course description

Theoretical education

1. Biochemical and genetic basis of inherited metabolic diseases
2. Classification of inherited metabolic diseases
3. Chromosomal basis of inheritance
4. Molecular basis of chromosomal aberrations and gene mutations in inherited diseases
5. Molecular basis of monogenic, polygenic and multifactorial inheritance
6. Biochemical mechanisms in inherited metabolic diseases pathogenesis
7. Molecular basis of diagnostics, prevention and therapy of inherited diseases.
8. Methods of prenatal and postnatal diagnosis of inherited diseases
9. Possibilities of inherited diseases therapy
10. Pharmacogenomics - genomic variation in response to therapy

Practical education

1. Methodology of inherited diseases diagnostics
2. Methods of biochemical laboratory diagnostics of inherited metabolic diseases
3. Numerical and structural chromosomal aberrations
4. Gene mutations detection
5. Polymerase chain reaction
6. Methods of laboratory diagnostics of gene mutations
7. Fluorescent in situ hybridization
8. Methods of prenatal diagnostics
9. Therapeutic possibilities of inherited metabolic diseases
10. Methods of pharmacogenetic analyses in drug metabolism

Literature

1. Robert L. Nussbaum, MD, Roderick R. McInnes. Thompson & Thompson's Genetics in Medicine. 7th Edition. ISBN: 978-1-4160-3080-5. 2007.
2. Strachan T, Read A. Human Molecular Genetics, 3rd ed. Garland Science/Taylor&Francis Group, 2003. ISBN:9780815341499.

Number of active classes	**Theoretical classes:** 30	**Practical classes:** 15

Teaching methods

Plenary lectures, problem sessions, independent presentations carried out by students.

Student activity assessment (maximally 100 points)			
Pre-exam activities	**points**	**Final exam**	**points**
Lectures	8	Written exam	15
Practices	12	Oral exam	40
Colloquium			
Essay	25		