UNIVERSITY OF NOVI SAD
FACULTY OF MEDICINE

Study program: Doctoral Academic Studies in Biomedical Sciences

Name of the subject: MOLECULAR BASIS OF INHERITED DISEASES

Teacher(s): Karmen M. Stankov

Status of the subject: elective

Number of ECПБ points: 20

Condition: -

Goal of the subject

The main objective of the course is to enable the PhD students to acquire the knowledge and skills about the molecular basis of inherited diseases, as well as other inherited conditions, with possibilities of diagnostics, prevention and therapy. The course 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..

Outcome of the subject

PhD students will acquire the comprehensive knowledge about the methodology and application of molecular methods applied in analysis 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.

Content of the subject

Theoretical lectures

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

Practical lectures

1. Methodology of inherited diseases diagnostics; 2. Methods of molecular diagnostics of inherited 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 diseases; 9. Molecular markers analysis.

Recommended literature

1. Thompson & Thompson Genetics in Medicine, 8th Edition from Robert Nussbaum, Roderick McInnes, Huntington Willard. 2015.
2. Marks' Basic Medical Biochemistry. 5th edition, by Michael A. Lieberman and Allan Marks, 2017.
3. Fukao T, Nakamura K. Advances in inborn errors of metabolism. J Hum Genet. 2019;64(2):65.
4. Stankov K, Benc D, Draskovic D. Genetic and epigenetic factors in etiology of diabetes mellitus type 1. Pediatrics. 2013; 132(6):1112-22.
5. Davison J. Gene Editing: A View Through the Prism of Inherited Metabolic Disorders. New Bioeth. 2018;24(1):2-8.

Number of active classes	**Theory:** 60	**Practice:** 45

Methods of delivering lectures. Theoretical and practical classes.

Evaluation of knowledge (maximum number of points 100)

activity during theoretical lectures: 20
practical lectures: 30
seminars: 10
written exam: 20
oral exam: 20