DESCRIPTION OF COURSE UNIT FOR DOCTORAL STUDIES AT VILNIUS UNIVERSITY

Scientific Area/eas, Field/ds of Science	Medical and Health Sciences (M 000): Medicine (M 001); Public Health (M 004) Natural Sciences (N 000): Biology (N 010)			
Faculty, Institute, Department/Clinic	Faculty of Medicine Institute of Biomedical Sciences Department of Human and Medical Genetics			
Course unit title (ECTS credits, hours)	**Medical Genetics** 10 credits (270 hours)			
Study method	**Lectures**	**Seminars**	**Consultations**	**Self-study**
Number of ECTS credits	-	-	1	9
Method of the assessment (in 10 point system)	Written and oral exam. Three open-ended questions.			

PURPOSE OF THE COURSE UNIT

To acquire knowledge about the general principles of genetic counseling, to know the laws of probability, to perform risk calculation in cases of various modes of inheritance. To deepen knowledge in the fields of medical genetics, diagnostic dysmorphology, syndromology, cancer genetics, prenatal diagnostics and clinical teratology. To understand the etiology, pathogenesis, clinical signs, treatment possibilities of inherited metabolic diseases. To understand the results of biochemical genetic tests.

THE MAIN TOPICS OF COURSE UNIT

Principles of genetic counselling and risk calculation. Genealogical analysis; personal history; phenotype, elements of morphology; physical measurements; differential diagnosis using clinical databases; genetic testing for developmental and intellectual disabilities; the laws of probability; risk calculation for different modes of inheritance; Bayes' theorem; Mendel's laws on inheritance; Hardy-Weinberg equilibrium; consanguineous marriage; balanced chromosome rearrangements; polygenic, multifactorial, mitochondrial inheritance; uniparental disomy, imprinting.
Chromosomal disorders and genetic syndromes. Autosomal tirsomies; sex chromosome abnormalities; deletions and duplications of the autosomes, microdeletion syndromes; uniparental disomy and imprinting disorders; the most common genetic syndromes characterized by: very small stature, moderate short stature, early overgrowth, senile-like appearance, unusual brain and/or neuromuscular findings with associated anomalies, facial and/or limbs defects, osteochondrodysplasias and osteochondrodysplasia with osteopetrosis, craniosynostosis, other skeletal dysplasias, connective tissue disorders, hamartoses, ectodermal dysplasias; most common sequences and associations.
Inherited metabolic diseases. Classification, pathogenesis, clinical manifestations. Newborn screening. Biochemical genetic testing. The inherited causes and differential diagnosis of hypoglycemia, metabolic acidosis, lactic acidosis, hyperammonaemia, hepatic dysfunction. Management and treatment of patients with inherited metabolic diseases, possibilities of prenatal diagnosis.
Medical genetics. Cardiovascular disorders (congenital heart disease, inherited cardiomyopathies, rhythm and conduction disorders, etc.); renal disorders (congenital disorders of the urinary tract, cystic, nephrotic, renal tubular disorders); respiratory disorders (cystic fibrosis, asthma, hereditary pulmonary emphysema, interstitial and restrictive pulmonary disorders); gastrointestinal disorders

(congenital anomalies of gastrointestinal tract and hepatobiliary duct system, inflammatory bowel disease, disorders of bile pigment metabolism); hematologic disorders (hemoglobinopathies, thalassemias, anaemias, hemophilias, other disorders of hemostasis, leukemias, lymphomas); immunologic disorders (immunodeficiency, complement defects, disorders of leukocyte function, systemic lupus erythematosus, rheumatoid disease, amyloidosis and other protein deposition diseases); endocrinologic disorders (genetic disorders of the pituitary gland, thyroid, parathyroid, adrenal gland, diabetes mellitus); anomalies of central nervous system, mental, behavioral and neurological disorders (neural tube defects, other anomalies of the central nervous system, intellectual disability, autism spectrum disorders, Alzheimer disease, epilepsy, hereditary ataxias, hereditary spastic paraplegia, autonomic and sensory disorders, multiple sclerosis); neuromuscular disorders (congenital muscular dystrophies, hereditary motor and sensory neuropathies, congenital myopathies, spinal muscular atrophies, motor neuron disease, myasthenias, myotonic dystrophies, nondystrophic myotonias); ophthalmologic disorders (color vision defects, optic atrophy and congenital blindness, glaucoma, defects of cornea, anomalies of lens, hereditary retinal and choroidal degenerations, strabismus); genetics of deafness; craniofacial anomalies; genodermatoses; connective tissue disorders (fibrillinopathies, Ehlers-Danlos syndrome, etc.); skeletal disorders (disorders predisposing to bone fragility, chondrodysplasias, disorders of bone mineralization and structure, the dysostoses, arthrogryposes, skeletal deformities, etc.).

Cancer genetics: monogenic hereditary cancer (Li-Fraumeni syndrome, familial breast cancer, retinoblastoma, familial adenomatous polyposis, Lynch syndrome, familial melanoma, Cowden syndrome, hereditary non-polyposis colorectal cancer); monogenic predisposition to cancer (Von Hippel-Lindau syndrome, tuberous sclerosis, basal cell nevus syndrome, MEN1 and MEN2 syndrome, neurofibromatosis, Peutz-Jeghers, Denys-Drash syndrome); cancers of multifactorial origin.

Prenatal diagnostics (PD). Indications for prenatal genetic counseling. Innovative PD methods and procedures. Prenatal genetic screening. Possibilities of UG in the detection of genetic and multifactorial diseases of the fetus. Preimplantation genetic diagnostics. PD of multifactorial diseases. Fetal therapy and intrauterine correction of congenital anomalies. Genetic diseases/conditions of a woman that may complicate pregnancy and/or affect fetal development. Legal and bioethical aspects of prenatal diagnostics.

Clinical teratology. Possible teratogenic effects, basic principles of teratology; teratogenesis and congenital anomalies.

RECOMMENDED LITERATURE SOURCES

1. Harper P.S. Practical Genetic Counselling. 8th Edition. Hodder Arnold, 2019.
2. Young I. D. Introduction to Risk Calculation in Genetic Counseling. 3rd Edition. Oxford University press, 2006.
3. Firth H.V, Hurst J.A. Oxford Desk Reference: Clinical Genetics and genomics. Oxford University Press, 2017.
4. Kenneth Lyons Jones, Marilyn Crandall Jones, Miguel del Campo. Smith's Recognizable Patterns of Human Malformation. 8th Edition. Elsevier, 2021.
5. Carey J.C, Cassidy S. B, Battaglia A, Viskochil D. Management of Genetic Syndromes, 4th Edition. WileyBlackwell, 2021.
6. Utkus A, Cimbalistienė L. Sindromologija II dalis. Vilniaus universitetas, 2014.
7. Rimoin D, Pyeritz R, Korf B. Emery and Rimoin's Principles and Practice of Medical Genetics, 6th Edition. Academic Press, 2013.
8. Songailienė J. Biocheminė laboratorinė paveldimųjų medžiagų apykaitos ligų diagnostika. Vilniaus universitetas, 2013.

9. Hoffmann G.F, Zschocke J, Nyhan W.L. Inherited Metabolic Diseases. A Clinical Approach. 2nd Edition. Springer, 2017.
10. Milunsky A, Milunsky J.M. Genetic Disorders and the Fetus: Diagnosis, Prevention, and Treatment. Hoboken, NJ : Wiley-Blackwell, 2021.
11. Rascon J.ir kt. Vaikų onkologija. Vilniaus universiteto leidykla, 2020.
12. Hodgson SV ir kt. A Practical Guide to Human Cancer Genetics. 4th Edition. Springer, 2007.
13. http://www.omim.org/
14. http://www.orpha.net/
15. https://www.ncbi.nlm.nih.gov/books/NBK1116/

CONSULTING LECTURERS

1. Coordinating lecturer: Eglė Preikšaitienė (Prof. Dr.).
2. Algirdas Utkus (Prof. Dr. HP).
3. Aušra Matulevičienė (Assoc. Prof. Dr.).

APPROVED:

By Council of Doctoral School of Medicine and Health Sciences at Vilnius University: 29th of September 2022

Chairperson of the Board: Prof. Janina Tutkuvienė