

**Program of Study** : General Medicine  
**Course** : Medical Genetics  
**Abbreviation** : **LGE/VA012**  
**Schedule** : 25 hours of seminars  
5 hours of exercises  
**Course Distribution** : 4<sup>th</sup> year  
**Number of Credits** : 2  
**Course Form** : Seminars, exercises

### **Seminars:**

**Teachers:** prof. MUDr. Martin Procházka, Ph.D.  
prof. RNDr. David Friedecký, Ph.D.  
MUDr. Václava Curtisová, MSC.  
doc. MUDr. Jana Volejníková, Ph.D.  
doc. MUDr. Peter Rohoň, Ph.D.  
MUDr. Júlia Štellmachová  
MUDr. Lucia Punová  
MUDr. Veronika Woitková  
MUDr. Radek Stražil  
RNDr. Pavlína Čapková, Ph.D.  
Mgr. Mária Janíková, Ph.D.  
Mgr. Dita Vrbická, Ph.D.  
Mgr. Zuzana Spurná, Ph.D.  
Mgr. Kristýna Kolaříková  
Mgr. Petr Vrtěl

**Study:** Block (from 8am to 13pm)  
**Room:** Lecture room (basement)  
**Block date:** 05. 12. – 09. 12. 2022  
30. 01. – 03. 02. 2023  
27. 02. – 03. 03. 2023  
08. 05. – 12. 05. 2023

		Subject	No. of Less.	Teacher
1	M o n d a y	Introduction, case report assignment	2	Volejníková
		The role of genetics in medicine		Volejníková/Curtisová
		Ethics in medical genetics	2	Volejníková
2	T u e s d a y	Single-gene disorders: definition, classification, diagnostics – the most common and clinically relevant examples, prognosis	2	Volejníková
		Molecular genetics – diagnosis	1	Spurná/Janíková
		Prenatal diagnostics of genetic diseases	1	Punová/Procházka/Woitková
3	W e d n e s d a y	Inborn errors of metabolism: definition, classification, diagnostics – examples, prognosis	2	Friedecký
		Oncogenetics	1	Vrtěl P./Janíková
		Epigenetics, dynamics of epigenome	1	Janíková/Spurná
4	T h u r s d a y	DNA analysis: methods, clinical applications	2	Janíková/Spurná
		Polygenic and multifactorial inheritance: definition, diagnostics, examples, clinical impact	3	Curtisová/Procházka
		Mitochondrial inheritance		
5	F r i d a y	Disorders of sexual differentiation (DSD): classification, diagnostics, clinical management, ethics	2	Woitková/Stražil
		Teratogenic agents in pregnancy	1	Stražil/Woitková/Punová
		Chromosomal disorders: classification, examples	1	Čapková
		Pre-implantation genetic diagnostics (PGD)	1	Vrbická
5	F r i d a y	Genetics in hematology	1	Rohoň/Kolaříková
		Neurogenetics	1	Kolaříková/Rohoň
		Genetic risk calculation	1	Kolaříková/Vrtěl
		Case presentations	2	Štellmachová

## Exercises:

**Teachers :** prof. MUDr. Martin Procházka, Ph.D.  
MUDr. Václava Curtisová, MSc.  
MUDr. Jana Volejníková, Ph.D.  
MUDr. Júlia Štellmachová  
MUDr. Lucia Punová  
MUDr. Veronika Woitková  
Mgr. Zuzana Spurná  
Mgr. Mária Janíková, Ph.D.  
RNDr. Pavlína Čapková, Ph.D,

**Study :** Block

**Block Date :** 05. 12. – 09. 12. 2022  
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	<b>Block Day</b>	<b>Subject</b>	<b>No. of Less.</b>
1	<b>Monday</b>	<b>Genetic counseling I. – example, practical training</b>	<b>1</b>
2	<b>Tuesday</b>	<b>Genetic counseling II. – pedigree construction</b>	<b>1</b>
3	<b>Wednesday</b>	<b>DNA methods – practical demonstration</b>	<b>1</b>
4	<b>Thursday</b>	<b>Cytogenetics – practical demonstration</b>	<b>1</b>
5	<b>Friday</b>	<b>Case presentations, final evaluation of the course</b>	<b>1</b>

White coats and slippers for practical training.

**Completed by:** Examination

The examination in medical genetics will take the form of a written test at the Institute of Medical Genetics. In the case of two unsuccessful attempts, an oral examination follows in the presence of a three-member commission (see questions below).

**Requirements:** Students are accepted in the absence of classes with a maximum of 10 %. The attendance of seminars and all practical trainings and submission of the seminar work are required to get credits. However, one day of absence - for example in cases of acute illness or appointment at GP - is allowed providing the written apology is sent by the student (e-mail address – [julia.stellmachova@fnol.cz](mailto:julia.stellmachova@fnol.cz); [vladimira.drgova@fnol.cz](mailto:vladimira.drgova@fnol.cz)). In case of long-term illness or other serious cases students can register in a different course than their previous option if personal permission of the head or deputy of the department is given.

## **Literature:**

1. **Chapters of Medical Genetics for general medicine students, in press 2022**
2. H.M.Kingston: ABC of Clinical Genetics, third edition 2002
3. Dorian J. Pritchard & Bruce R. Korf: Medical Genetics at a Glance, 2003
4. Peter Turnpenny, Sian Ellard: Emery's Element sof Medical Genetics, 2012
5. Thompson and Thompson: Genetics in medicine. W.B. Sauders Company, 2016

## **Medical Genetics – questions for exam**

1. The Central Dogma of Molecular Biology, Expression of Genetic Information
2. Basic Types of Mendelian Inheritance, Mendel's Laws and Their Applications in Clinical Genetics
3. Autosomal Dominant Inheritance Criteria. Clinical Examples of Autosomal Dominant Hereditary Disorders
4. Autosomal Recessive Inheritance Criteria. Clinical Examples of Autosomal Recessive Hereditary Disorders
5. Criteria for Gonosomal Recessive and Gonosomal Dominant Inheritance. Clinical Examples of X-Linked Diseases
6. Non –Mendelian Inheritance - Genomic Imprinting, Somatic Germinal Mosaicism, Examples
7. Non –Mendelian Inheritance - Mitochondrial Inheritance - Criteria and Clinical Manifestations of Mitochondrial Hereditary Diseases
8. Polygenic Inheritance, Multifactorial Diseases, Examples
9. Basic Characteristics and Structure of Human Chromosomes
10. Cell Division (Mitosis, Meiosis)
11. Numerical and Structural Aberrations of Autosomes - Types, Mechanism of Origin, Clinical Examples
12. Aberration of Gonosomes - Types, Mechanism of Origin, Clinical Examples
13. Indications for Postnatal and Prenatal Cytogenetic Examination
14. Methods of Cytogenetic and Molecular-Cytogenetic Analysis: Karyotyping, FISH, Array-CGH, MLPA (Principles of Methods, Utilization)
15. Preimplantation Genetic Examinations - Preimplantation Screening and Diagnostics, Indication for Examination, Methods of Preimplantation Diagnostics
16. Somatosexual Abnormalities - Sexual Differentiation Disorders, Examples
17. Inherited Metabolic Disorders - Inheritance, Classification, Diagnostics, Newborn Screening in The Czech Republic
18. Genetic Background of Cancer
19. Basic Characteristic of Tumor, Examples of Clinical Utilisation of Genetic Features of Tumors
20. Genetics of Hematooncological Diseases
21. Characteristics and Examples of Hereditary Tumor Syndromes
22. Examples of Genetic Syndromes with a High Risk of Tumor Developing
23. Neurogenetics - Characteristics, Examples of Diseases

24. Examples of Genetic Diseases in Ophthalmology
25. Screening Examinations in Pregnancy - Combined Screening of the First Trimester, Triple Test, New Screening Methods (Non-Invasive Prenatal Screening of Circulating Fetal DNA in Maternal Plasma etc.)
26. Prenatal Diagnostics - Non-Invasive Prenatal Diagnostics (Ultrasound, First And Second Trimester Screening, Magnetic Resonance, Non-Invasive Prenatal Examination of Free Circulating Fetal DNA in Maternal Plasma), Invasive Prenatal Diagnostics (Chorionic Villus, Amniocentesis, Cordocentesis, Fetoscopy)
27. Teratogenic Factors in Pregnancy - Biological, Chemical and Physical Teratogens, Effects of Teratogens on The Fetus - Examples
28. Human Genome: Definition, Structure, Variability and Dynamics
29. Definitions of Human Gene, Mutations, Variants, Gene and Variants Nomenclature - Examples
30. Evaluation of Gene Variants (Assessment of Pathogenicity) - Evaluation of Variants Association with Disease, Basic Criteria for Variant Assessment, Use of Reference Sequences and Clinical Variant Databases
31. Genetics of Internal Diseases - Inheritance, Examples of Disease
32. Introduction to Population Genetics, Allelic and Genotypic Frequencies and Calculation of The Risk of Genetic Disease Transmission to The Next Generation
33. DNA structure, Basic Properties of DNA - Direct and Indirect DNA Diagnostics
34. Basic Molecular Methods of DNA Diagnostics
35. Immunogenetics - HLA System (Examination by Molecular Genetic Methods, Association of HLA With Diseases, Histocompatibility in Transplantation), Genetics of Immunopathological Conditions, Immunogenetics of Infections and Immunopharmacogenetics
36. Pharmacogenetics - Clinical Impacts of Polymorphisms in Genes Encoding Drugs
37. Basic Epigenetic Mechanisms and Examples of Diseases
38. Ethics in medical genetics