JAGIELLONIAN UNIVERSITY
MEDICAL COLLEGE

# Clinical Genetics

Educational subject description sheet

## Basic information

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|---|---|
| **Department**<br>Faculty of Medicine | **Didactic cycle**<br>2016/17 |
| **Field of study**<br>Medical Program | **Realization year**<br>2019/20 |
| **Study level**<br>long-cycle master's degree program | **Lecture languages**<br>English |
| **Study form**<br>full-time | **Block**<br>obligatory for passing in the course of studies |
| **Education profile**<br>general academic | **Mandatory**<br>obligatory |
| **Disciplines**<br>Medical science | **Examination**<br>graded credit |
| **Subject related to scientific research**<br>Yes | **Standard group**<br>E. Clinical non-procedural medical disciplines |

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|---|---|
| **Subject coordinator** | Mirosław Bik-Multanowski |
| **Lecturer** | Mirosław Bik-Multanowski, Anna Madetko-Talowska, Katarzyna Szewczyk, Artur Dobosz |

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| **Periods**<br>Semester 7, Semester 8 | **Examination**<br>graded credit<br><br>**Activities and hours** lecture: 30 | **Number of ECTS points**<br>2.0 |

## Goals

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|---|---|
| C1 | Providing up-to-date knowledge regarding the clinical picture of the most common hereditary diseases (including monogenic diseases, chromosomal aberrations, metabolic diseases and cancers). |
| C2 | Providing current knowledge on conducting genetic counseling. |
| C3 | Providing up-to-date knowledge about prenatal diagnosis and the role of genetics in reproductive failures. |
| C4 | Providing current knowledge on primary, secondary and tertiary prevention of hereditary diseases. |
| C5 | Acquiring knowledge and practical foundations on the subject of modern genetic diagnostics using molecular biology techniques and cytogenetic methods. Rules for the interpretation of genetic testing results in the context of genetic counseling. |

## Subject's learning outcomes

| Code | Outcomes in terms of | Effects | Examination methods |
|---|---|---|---|
| **Knowledge – Student knows and understands:** | | | |
| W1 | development, structure and functions of the human body in normal and pathological conditions | O.W1 | test |
| W2 | symptoms and course of diseases | O.W2 | test |
| W3 | methods of diagnostic and therapeutic procedures appropriate for specific disease states | O.W3 | test |
| W4 | environmental and epidemiological determinants of the most frequent diseases | E.W1 | test |
| W5 | basic methods of fetal diagnostics and therapy | E.W5 | test |
| W6 | environmental and epidemiological determinants of the most frequent human neoplastic diseases | E.W23 | test |
| W7 | basics of early detection of neoplastic diseases and principles of screening in oncology | E.W24 | test |
| W8 | the causes, symptoms, principles of diagnosis and therapeutic management of the most common hereditary diseases | E.W37 | test |
| W9 | the types of biological materials to be used for laboratory diagnosis and the rules for the collection of test material | E.W39 | test |
| W10 | theoretical and practical basics of laboratory diagnostics | E.W40 | test |
| **Skills – Student can:** | | | |
| U1 | identify medical problems and prioritize medical management | O.U1 | test |
| U2 | plan the diagnostic procedure and interpret its results | O.U3 | test |
| U3 | plan own learning activities and constantly learn in order to update own knowledge | O.U5 | test |
| U4 | communicate with the patient and his family in an atmosphere of trust, taking into account the needs of the patient | O.U7 | test |

| | | | |
|---|---|---|---|
| U5 | communicate and share knowledge with colleagues in a team | O.U8 | test |
| U6 | critically evaluate the results of scientific research and adequately justify the position | O.U9 | test |
| U7 | carry out a medical history with an adult patient | E.U1 | test |
| U8 | carry out a medical interview with the child and his or her family | E.U2 | test |
| U9 | carry out a physical examination of a child of all ages | E.U4 | test |
| U10 | perform differential diagnosis of the most common diseases of adults and children | E.U12 | test |
| U11 | plan diagnostic, therapeutic and prophylactic procedures | E.U16 | test |
| U12 | interpret the results of laboratory tests and identify the causes of abnormalities | E.U24 | test |
| U13 | maintain patient's medical records | E.U38 | test |
| **Social competences – Student is ready to:** | | | |
| K1 | to establish and maintain deep and respectful contact with patients and to show understanding for differences in world views and cultures | O.K1 | test |
| K2 | to be guided by the well-being of a patient | O.K2 | test |
| K3 | respect medical confidentiality and patients' rights | O.K3 | test |
| K4 | take actions towards the patient on the basis of ethical norms and principles, with an awareness of the social determinants and limitations of the disease | O.K4 | test |
| K5 | use objective sources of information | O.K7 | test |
| K6 | formulate conclusions from own measurements or observations | O.K8 | test |

## Calculation of ECTS points

| **Activity form** | **Activity hours*** |
|---|---|
| lecture | 30 |
| preparation for classes | 10 |
| preparation for classes | 20 |
| preparation of multimedia presentation | 6 |
| | |
| **Student workload** | **Hours**<br>60 |
| **Workload involving teacher** | **Hours**<br>30 |

* hour means 45 minutes

## Study content

| No. | Course content | Subject's learning outcomes | Activities |
|---|---|---|---|
| 1. | Basic concepts of genetics, the value of deciphering of the human genome in medical practice. Mutagenesis and teratogens. Phenocopies and genocopies. The basics of dysmorphology. | W1, W4, U1 | lecture |
| 2. | Construction and analysis of a pedigree in medical genetics. Theoretical and empirical risks in genetics. | W2, W8, U1, U10, U7, U8, U9, K1, K2, K3 | lecture |
| 3. | Mendelian and non-Mendelian inheritance; Mitochondrial diseases, complex diseases. | W1, W2, W4, W8, U1, U10, U7, U8, U9 | lecture |
| 4. | Chromosomal aberrations. Phenotype-genotype correlation. | W1, W2, W4, W8, U1, U4, U7, U8, U9 | lecture |
| 5. | Genetic basis of cancer. Diagnostics, genetic counseling and prevention in hereditary cancers. | W1, W2, W6, W7, W9, U1, U11, U3, U4 | lecture |
| 6. | Principles of genetic counseling. Psychological, ethical and social issues in genetic counseling. Patients' support groups. | W2, W3, U1, U11, U13, U4, U5, U6, U7, U8, U9, K1, K2, K3, K4, K6 | lecture |
| 7. | Diagnosing genetically determined diseases based on the analysis of dysmorphic features. Diagnostic strategy in chromosomal aberrations. | W2, W8, W9, U1, U4, U5, U7, U8, U9, K1, K2, K3 | lecture |
| 8. | Genetic diagnosis and counseling in monogenic and in complex diseases. | W2, W8, W9, U1, U4, U5, U7, U8, U9, K1, K2, K3 | lecture |
| 9. | Newborn baby with suspected genetic disease. | W2, W8, W9, U1, U10, U2, U3, U4, U5, U9, K2, K5, K6 | lecture |
| 10. | Disorders of sex determination. Diagnostic strategy and genetic counseling. | W2, W8, U1, U10, U4, U7, U8, U9, K1, K2, K3, K6 | lecture |
| 11. | Genetic aspects of infertility. Preimplantation diagnosis and prenatal testing; indications, methods, interpretation of results. | W2, W4, W5, W8, U1, U11, U2, U4, U7, K1, K2, K3, K5, K6 | lecture |
| 12. | Genetic factors in the etiology of intellectual disability, autism and common neurologic and psychiatric conditions. Behavioral genetics. | W2, W8, U1, U10, U13, U2, U4, U7, U8, U9, K1, K2, K3, K4, K5, K6 | lecture |
| 13. | Genetic aspects of selected diseases of the circulatory, digestive, respiratory, urinary and hematopoietic systems. | W2, W3, W4, U3, K5, K6 | lecture |
| 14. | Inborn errors of metabolisms. | W2, W8, U1, U11, U12, U13, U2, U3, U4, U5, U7, U8, U9, K1, K2, K3, K4, K5, K6 | lecture |
| 15. | Modern molecular and cytogenetic tests. Interpretation of the results of genetic testing in a clinical context. | W10, W2, W3, W5, W8, W9, U1, U11, U12, U13, U2, U6, K5, K6 | lecture |

## Course advanced

**Teaching methods:**

case study, classes / practicals, demonstration, discussion, case study method, presentation, group work, seminar, lecture,

lecture with multimedia presentation, practical classes

| Activities | Examination methods | Credit conditions |
|---|---|---|
| lecture | test | at least 18/30 points (written test) |

**Additional info**

Grading score:
18-20 points - 3.0, 21-22 points - 3.5, 23-24 points - 4.0, 25-26 points - 4.5, 27-30 points - 5.0

## Entry requirements

not applicable

## Literature

**Obligatory**

1. Firth HV, Hurst JA: "Oxford Desk Reference Clinical Genetics & Genomics" 2nd edition, Oxford University Press 2012

**Optional**

1. Tobias ES, Connor M, Ferguson-Smith M: "Essentials Medical Genetics", 6th edition, Wiley-Blackwell

## Standard effects

| Code | Content |
|---|---|
| E.U1 | carry out a medical history with an adult patient |
| E.U2 | carry out a medical interview with the child and his or her family |
| E.U4 | carry out a physical examination of a child of all ages |
| E.U12 | perform differential diagnosis of the most common diseases of adults and children |
| E.U16 | plan diagnostic, therapeutic and prophylactic procedures |
| E.U24 | interpret the results of laboratory tests and identify the causes of abnormalities |
| E.U38 | maintain patient's medical records |
| E.W1 | environmental and epidemiological determinants of the most frequent diseases |
| E.W5 | basic methods of fetal diagnostics and therapy |
| E.W23 | environmental and epidemiological determinants of the most frequent human neoplastic diseases |
| E.W24 | basics of early detection of neoplastic diseases and principles of screening in oncology |
| E.W37 | the causes, symptoms, principles of diagnosis and therapeutic management of the most common hereditary diseases |
| E.W39 | the types of biological materials to be used for laboratory diagnosis and the rules for the collection of test material |
| E.W40 | theoretical and practical basics of laboratory diagnostics |
| O.K1 | to establish and maintain deep and respectful contact with patients and to show understanding for differences in world views and cultures |
| O.K2 | to be guided by the well-being of a patient |
| O.K3 | respect medical confidentiality and patients' rights |
| O.K4 | take actions towards the patient on the basis of ethical norms and principles, with an awareness of the social determinants and limitations of the disease |
| O.K7 | use objective sources of information |
| O.K8 | formulate conclusions from own measurements or observations |
| O.U1 | identify medical problems and prioritize medical management |
| O.U3 | plan the diagnostic procedure and interpret its results |
| O.U5 | plan own learning activities and constantly learn in order to update own knowledge |
| O.U7 | communicate with the patient and his family in an atmosphere of trust, taking into account the needs of the patient |
| O.U8 | communicate and share knowledge with colleagues in a team |
| O.U9 | critically evaluate the results of scientific research and adequately justify the position |
| O.W1 | development, structure and functions of the human body in normal and pathological conditions |
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