Pomeranian Medical University in Szczecin

# SYLLABUS of the MODULE(SUBJECT)

## General Information

| Code | | | Title | ***Clinical Genetics*** |
|---|---|---|---|---|
| Module type | | | | *Obligatory* |
| Faculty | | | | *Faculty of Medicine* |
| Field of study | | | | *medicine* |
| Major | | | | *Not applicable* |
| Level of study | | | | *II level/long-cycle (S2J)* |
| Mode of study | | | | *intramural* |
| Year of study | | | | *IV* |
| Semester | | | | *block* |
| ECTS points | | | | *4,5* |
| Types of training | | | | *seminars/classes 55 h* (25h seminars; 30 h classes) |
| Tutor responsible for the module | | | | *Prof. dr hab. n. med. Jan Lubiński* |
| Tutors conducting the subject | | | | Prof. dr hab. n. med. Jan Lubiński (jan.lubinski@pum.edu.pl)<br>Prof. dr hab. n. med. Cezary Cybulski (cezary.cybulski@pum.edu.pl)<br>Prof. dr hab. n. med. Jacek Gronwald, (jacek.gronwald@pum.edu.pl)<br>Prof. dr hab. n. med. Tadeusz Dębniak (tadeusz.debniak@pum.edu.pl)<br><br>dr n.med. Katarzyna Gołębiewska (katarzyna.golebiewska@pum.edu.pl)<br><br>dr n.med. Katarzyna Białkowska (katarzyna.bialkowska@pum.edu.pl)<br>dr n.med. Wojciech Kluźniak (wojciech.kluzniak@pum.edu.pl)<br>dr n.med. Helena Rudnicka (helena.rudnicka@pum.edu.pl)<br><br>Mgr Jakub Deptuła (jakub.deptula@pum.edu.pl)<br><br>dr n.med. Elżbieta Złowocka-Perłowska (elzbieta.zlowocka.perlowska@pum.edu.pl) |
| WWW | | | | www.genetyka.com |
| Language | | | | English |

## Detailed information

| | | |
|---|---|---|
| Module objectives | | It is to provide the basic knowledge in the field of modern clinical genetics with respect to any branch of medicine. Knowledge should include the rules of inheritance and diagnosis of hereditary diseases, the identification of genes responsible for the formation of genetic diseases, mechanisms of regulation of gene expression; mechanisms of DNA repair. |
| **Prerequisite /essential requirements/** | Knowledge | Knowledge of the: basic concepts of genetics, chromosome structure and description of proper human karyotype . |
| | Skills | To be able to solve problems with use of the Mendel laws. Knowledge of the basic concepts of genetics, chromosome structure and description of proper human karyotype |
| | Competences | The habit of self-study, group work |

**Description of the learning outcomes for the subject /module**

| Number of learning outcome | Student, who has passed the (subject) Knows /is able to /can: | SYMBOL (referring the standards) EKK | Method of verification of learning outcomes |
|---|---|---|---|
| KL2JPW01 | knows the basic concepts of genetics | K_C.W1 | Single choice test. Exam |
| KL2JPW02 | describes phenomena of coupling and cooperation of genes | K_C.W2 | Single choice test. Exam |
| KL2JPW03 | correctly describes human karyotype and different stages of sex determination | K_C.W3 | Single choice test. Exam |
| KL2JPW04 | describes structure of chromosomes and molecular base of mutagenesis | K_C.W4 | Single choice test. Exam |
| KL2JPW05 | knows the principles of inheritance of a variety of traits, inheritance of quantitative traits, independent inheritance of traits and inheritanceofextra-nucleargenetic information | K_C.W5 | Single choice test. Exam |
| KL2JPW06 | knows genetic conditions of human blood groups and Rh complex serological incompatibility | K_C.W6 | Single choice test. Exam |
| KL2JPW07 | describes aberration of autosomes and heterosomes causingdiseases, incl. oncogenesis | K_C.W7 | Single choice test. Exam |
| KL2JPW08 | knows factors affecting primary and secondary genetic equilibrium of population | K_C.W8 | Single choice test. Exam |
| KL2JPW09 | knows principles for diagnostics of gene and chromosome mutations responsible for hereditary diseases, incl. neoplasms | K_C.W9 | Single choice test. Exam |
| KL2JPW10 | defines advantages and threats related to GMO's found in ecosystem | K_C.W10 | Single choice test. Exam |
| KL2JPW11 | knows genetic mechanism for contracting drug-resistance by micro-organisms and malignant cells | K_C.W11 | Single choice test. Exam |
| KL2JPW12 | defines clinical course of specific and non- | K_C.W27 | Single choice test. |

| | | | |
|---|---|---|---|
| | specific inflammation and describes the processes of regeneration of tissues and organs | | Exam |
| KL2JPW13 | knows indications for genetic examination in order to individualize pharmacotherapy | K_C.W40 | Single choice test. Exam |
| KL2JPW14 | knowskeydirectionsoftherapy development, in particular the potential of cellular therapy, gene therapy and target therapy in certain diseases | K_C.W41 | Single choice test. Exam |
| KL2JPW15 | knows the basic principles of ethics in genetics | | Single choice test. Exam |
| KL2JPW16 | has knowledge of fetal development and developmental defects | | Single choice test. Exam |
| KL2JPW17 | He knows the symbols of pedigree description | | Single choice test. Exam |
| KL2JPU01 | analyses genetic crosses and genealogy of human traits and diseases and evaluates risk of the birth of chromosome aberration affected children | K_C.U1 | Single choice test. Exam |
| KL2JPU02 | identifies indications during prenatal examination | K_C. U2 | Single choice test. Exam |
| KL2JPU03 | makes decision on cytological and molecular examination | K_C.U3 | Single choice test. Exam |
| KL2JPU04 | performs morphometric measurements, analyzes morphograms and notes karyotypes of diseases | K_C.U4 | Single choice test. Exam |
| KL2JPU05 | assesses risk of the occurrence of disease in progeny on the basis of family predispositions and influence of environmental factors | K_C.U5 | Single choice test. Exam |
| KL2JPU06 | Uses a code of ethical conduct in the field of genetics | | Single choice test. Exam |
| KL2JPU07 | Can read the correct DNA bases sequence | | Single choice test. Exam |
| KL2JPU08 | Knows how to conduct interview with the patient | | Single choice test. Exam |
| KL2JPU09 | can make and analyze pedigree on the basis of interview with the patient | | Credit |
| KL2JPK01 | accepts the need for standards of conduct | K_K01 | Credit |
| KL2JPK02 | recognizes concept and need for responsibility for property he/she has been entrusted with | K_K02 | Credit |
| KL2JPK03 | Demonstrates the awareness for self-education, understands the need for continuing professional education, can inspire and organize learning processes in others | K_K03 | Credit |
| KL2JPK04 | co-operates with team members; can co-operate within a group and take different roles | K_K04 | Credit |
| KL2JPK05 | adheres to proper examiner/examinee relationship while performing functional tests and observations | K_K05 | Credit |

**Matrix presenting the learning outcomes of the subject/module in relation to the form of classes**

| Number of learning outcome | Student, who has passed the (subject) Knows /is able to /can: | Types of training | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| | | Lecture | Seminar | Laboratory classes | Projectwork | Clinical classes | Classes | Practical classes | Other |
| KL2JPW01 | knows the basic concepts of genetics | | X | | | X | | | |
| KL2JPW02 | describes phenomena of coupling and cooperation of genes | | X | | | X | | | |
| KL2JPW03 | correctly describes human karyotype and different stages of sex determination | | X | | | X | | | |
| KL2JPW04 | describes structure of chromosomes and molecular base of mutagenesis | | X | | | X | | | |
| KL2JPW05 | knows the principles of inheritance of a variety of traits, inheritance of quantitative traits, independent inheritance of traits and inheritance of extra-nuclear genetic information | | X | | | X | | | |
| KL2JPW06 | knows genetic conditions of human blood groups and Rh complex serological incompatibility | | X | | | X | | | |
| KL2JPW07 | describes aberration of autosomes and heterosomes causing diseases, incl. oncogenesis | | X | | | X | | | |
| KL2JPW08 | knows factors affecting primary and secondary genetic equilibrium of population | | X | | | X | | | |
| KL2JPW09 | knows principles for diagnostics of gene and chromosome mutations responsible for hereditary diseases, incl. neoplasms | | X | | | X | | | |
| KL2JPW10 | defines advantages and threats related to GMO's found in ecosystem | | X | | | X | | | |
| KL2JPW11 | knows genetic mechanism for contracting drug-resistance by micro-organisms and malignant cells | | X | | | X | | | |
| KL2JPW12 | defines clinical course of specific and non-specific inflammation and describes the processes of regeneration of tissues and organs | | X | | | X | | | |
| KL2JPW13 | knows indications for genetic examination in order to individualize pharmacotherapy | | X | | | X | | | |
| KL2JPW14 | knows key directions of therapy development, in particular the potential of cellular therapy, gene therapy and target therapy in certain diseases | | X | | | X | | | |
| KL2JPW15 | knows the basic principles of ethics in genetics | | X | | | X | | | |
| KL2JPW16 | has knowledge of fetal development and developmental defects | | X | | | X | | | |
| KL2JPW17 | He knows the symbols of pedigree description | | X | | | X | | | |
| KL2JPU01 | analyses genetic crosses and genealogy of human traits and | | X | | | X | | | |

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|---|---|---|---|---|---|---|---|---|---|
| | diseases and evaluates risk of the birth of chromosome aberration affected children | | | | | | | | |
| KL2JPU02 | identifies indications during prenatal examination | | X | | | X | | | |
| KL2JPU03 | makes decision on cytological and molecular examination | | X | | | X | | | |
| KL2JPU04 | performs morphometric measurements, analyzes morphograms and notes karyotypes of diseases | | X | | | X | | | |
| KL2JPU05 | assesses risk of the occurrence of disease in progeny on the basis of family predispositions and influence of environmental factors | | X | | | X | | | |
| KL2JPU06 | Uses a code of ethical conduct in the field of genetics | | X | | | X | | | |
| KL2JPU07 | Can read the correct DNA bases sequence | | X | | | X | | | |
| KL2JPU08 | Knows how to conduct interview with the patient | | X | | | X | | | |
| KL2JPU09 | can make and analyze pedigree on the basis of interview with the patient | | X | | | X | | | |
| KL2JPK01 | accepts the need for standards of conduct | | X | | | X | | | |
| KL2JPK02 | recognizes concept and need for responsibility for property he/she has been entrusted with | | X | | | X | | | |
| KL2JPK03 | Demonstrates the awareness for self-education, understands the need for continuing professional education, can inspire and organize learning processes in others | | X | | | X | | | |
| KL2JPK04 | co-operates with team members; can co-operate within a group and take different roles | | X | | | X | | | |
| KL2JPK05 | adheres to proper examiner/examinee relationship while performing functional tests and observations | | X | | | X | | | |

| **Module (subject) contents** | | |
|---|---|---|
| **Symbol of teaching programme** | **Content of teaching programme** | **References to learning outcomes** |
| TK01 | Indications for genetic testing. Principles of cytogenetic studies. Chromosome construction, karyotype, chromosome aberrations | KL2JPW01; KL2JPW02; KL2JPW03; KL2JPW04; KL2JPW05; KL2JPW06; KL2JPW07; KL2JPW08; KL2JPU01; KL2JPU02; KL2JPU03; KL2JPU04; KL2JPU05; KL2JPU06; KL2JPU07; KL2JPU08; KL2JPU09; KL2JPK05 |
| TK02 | Genetics of disorders of sex differentiation and determination and failures in reproduction. | KL2JPW03; KL2JPW04; KL2JPW05; KL2JPK01 |
| TK03 | Molecular techniques for detecting mutations | KL2JPU01; KL2JPW04; KL2JPW05; KL2JPU06; KL2JPU07; KL2JPU08 |
| TK04 | Hereditary cancers. Principles of inheritance and diagnosis of hereditary diseases; Mitochondrial diseases | KL2JPW05; KL2JPW07; KL2JPU08; KL2JPU09; KL2JPW14; KL2JPW17; KL2JPK02 |
| TK05 | Pedigree clinical criteria, indications for DNA tests. | KL2JPW05; KL2JPW09; KL2JPW13; KL2JPU09 ; KL2JPU08; KL2JPU07; KL2JPK05 |

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|---|---|---|
| TK06 | Hereditary cancer:<br>- breasts<br>- colon<br>-prostate<br>Von Hippel-Lindau syndrom<br>- neurofibromatosis type NF1, NF2 | KL2JPW05; KL2JPW09; KL2JPW11; KL2JPW 12; KL2JPW13; KL2JPW14; KL2JPU01; KL2JPU09; KL2JPK02; KL2JPK04 |
| TK07 | Von Willebrand syndrom.<br>Homologous recombination and gene therapy | KL2JPW09; KL2JPW14; KL2JPU09; KL2JPU08; KL2JPU07; KL2JPU06; KL2JPK02; KL2JPK04 |
| TK08 | Monogenic diseases<br>-autosomal recessive.<br>-autosomal dominant.<br>Chromosomal X-linked monogenic diseases.<br>Multigene diseases.<br>Chromosomal diseases. | KL2JPW02; KL2JPW05; KL2JPW07; KL2JPW09; KL2JPW14; KL2JPU01; KL2JPU08; KL2JPU09; KL2JPK02 |
| TK09 | Malformations. Prenatal Diagnosis. Causes. Concepts. | KL2JPW09; KL2JPW13; KL2JPW16; KL2JPW17; KL2JPU07; KL2JPU08; KL2JPU09; KL2JPK04 |
| TK10 | Ethics in genetics, legal aspects of genetic examinations. | KL2JPW15; KL2JPU06; KL2JPK03 |

**References and educational resources**


1. Molecular Cell Biology/Harvey Lodish W.H.Freeman;7 ED. cop. 2012 r ISBN 9781429234139

2. Essential medical genetics/ Edward S. Tobias, Michael Connor, Malcolm Ferguson-Smith. 6th ed Chichester;Wiley-Blackwell.cop.2011 ISBN 9781405169745

**3.** . Medical genetics / Lynn B. Jorde, John C. Carey, Michael J. Bamshad. 5th ed. Philadelphia : Elsevier, cop. 2016; ISBN 978-0-323-18835-7

4. Genetics in Medicine/Thompson & Thompson. Robert L.Nussbaum, Roderick R. McInnes, Huntington F.Willard. 8th ed. Philadelphia : Elsevier, cop. 2016; ISBN 978-1-4377-0696-3


**Student's workload (balance sheet of ECTS points)**

| Form of student's activity (in-class participation; activeness, produce a report, etc.) | Workload [h] | | |
|---|---|---|---|
| | Tutor | Student | Average |
| activities that require direct participation of tutors | | 55 | |
| Preparation to the classes | | | |
| Reading of the indicated/specified literature | | | |
| Report writing/project making | | | |
| Time spent to prepare for the exam | | | |
| Other | | | |
| | | | |
| Student's workload in total | | | |
| **ECTS points for the subject** | **4.5** | | |

**Remarks at the end**

Methods of assessment, for example:
E – exam- problem resolving S
– verifying of practical skills R
– report
D – discussion P
– presentation
Others-