Appendix 5
to Resolution No. 15630
of Senate of Wroclaw Medical University
of 30 March 2016

Syllabus for the academic year 2016/2017

Description of the course

Module/Course	CLINICAL GENETICS	Group of detailed education results	
		Group code: C	Group name: **preclinical**
Faculty	English Division		
Major	Medicine		
Specialties	----------		
Level of studies	Uniform magister studies X* 1st degree studies ☐ 2nd degree studies ☐ 3rd degree studies ☐ postgraduate studies ☐		
Form of studies	X full-time ☐ part-time		
Year of studies	III	Semester	X Winter ☐ Summer
Type of course	X obligatory ☐ limited choice ☐ free choice / elective		
Course	☐ major X basic		
Language of instruction	☐ Polish X English ☐ other		

* mark ☐ with an X

Number of hours

Form of education

Unit teaching the course	Lectures (L)	Seminars (SE)	Auditorium classes (AC)	Major Classes – not clinical (MC)	Clinical Classes (CC)	Laboratory Classes (LC)	Classes in Simulated Conditions (CSC)	Practical Classes with Patient (PCP)	Specialist Classes – magister studies (SCM)	Foreign language Course (FLC)	Physical Education obligatory (PE)	Vocational Practice (VP)	Self-Study (Student's own work)	E-learning (EL)
Winter Semester														
	20			50										
Summer Semester														

TOTAL per year:													
	20			50									

Educational objectives (max. 6 items)

C1. Understand molecular mechanism of human inheritance. Be familiar with the aetiology, symptomatology and management of human genetic disorders.

C2. Know dysmorphologic nomenclature and understand principles of genetic testing methods, their applications and limitations and interpretation of results.

C3. Assessment of the indications for genetic testing in prenatal and posnatal clinical setting.

C4. Take relevant history, construct pedigrees, perform clinical examination and offer genetic counselling.

C5. Identify the legal, ethical and social implications of genetic testing, including predictive testing, carrier testing and prenatal diagnosis.

C6. Make diagnosis of genetic conditions/syndromes and perform genetic counselling.

Education result matrix for module/course in relation to verification methods of the intended education result and the type of class

Number of course education result	Number of major education result	Student who completes the module/course knows/is able to	Methods of verification of intended education results (forming and summarising)	Form of didactic class ***enter the abbreviation*
W 01	CW1	understands basic concepts of genetics	test, oral response, colloquium, written examination	L MC
W02	CW2	describes genetic linkage and gene-gene interactions	test, oral response, colloquium, written examination	L MC
W03	CW3	describes a normal human karyotype and different types of sex determination	test, oral response, colloquium, written examination	L MC
W04	CW4	is familiar with chromosome and molecular basis of mutagenesis	test, oral response, colloquium, written examination	L MC
W05	CW5	is familiar with inheritance of a variety of traits, quantitative traits, mendelian inheritance and non-nuclear inheritance	test, oral response, colloquium, written examination	L MC
W06	CW7	describes autosome and	test, oral response,	L

		heterosome aberrations in the context of their pathogenicity including oncogenesis	colloquium, written examination	MC
W07	CW8	is familiar with primary and secondary factors influencing genetic equilibrium of the population	test, oral response, colloquium, written examination	L MC
W08	CW9	is familiar with basic diagnostic methods of chromosome aberrations and gene mutations responsible for hereditary and acquired disorders, including tumours	test, oral response, colloquium, written examination	L MC
U 01	CU1	analysis transmission of traits and pedigrees with traits and disorders as well as assesses risk of having offspring affected by chromosomal aberrations	test, oral response, colloquium, written examination	MC
U02	CU2	identifies indications for prenatal diagnosis	test, oral response, colloquium, written examination	MC
U03	CU3	makes decisions about cytogenetic and molecular testing	test, oral response, colloquium, written examination	MC
U04	CU4	makes morphologic measurements, analysis morphograms and describes karyotypes	test, oral response, colloquium, written examination	MC
U05	CU5	assesses risk of affected offspring by analysing familial predispositions and environmental factors	test, oral response, colloquium, written examination	MC
K 01	CK1	acknowledges the necessity of studying throughout lifetime, inspires	test, oral response, colloquium, written examination	MC
K02	CK2	is able to cooperate and work as a part of a group, taking on different roles	test, oral response, colloquium, written examination	MC
K03	CK3	is able to prioritize tasks set by themselves or others	test, oral response, colloquium, written examination	MC
K04	CK4	correctly identifies and settles work related dilemmas	test, oral response, colloquium, written examination	MC

** L - lecture; SE - seminar; AC – auditorium classes; MC – major classes (non-clinical); CC – clinical classes; LC – laboratory

classes; SCM – specialist classes (magister studies); CSC – classes in simulated conditions; FLC – foreign language course; PCP practical classes with patient; PE – physical education (obligatory); VP – vocational practice; SS – self-study, EL – E-learning .

Please mark on scale 1-5 how the above effects place your classes in the following categories: communication of knowledge, skills or forming attitudes:
Knowledge: +++
Skills: +++
Social competences: +

Student's amount of work (balance of ECTS points)	
Student's workload (class participation, activity, preparation, etc.)	Student Workload (h)
1. Contact hours:	70
2. Student's own work (self-study):	109
Total student's workload	179
ECTS points for module/course	6,5
Comments	

Content of classes (please enter topic words of specific classes divided into their didactic form and remember how it is translated to intended educational effects)

Lectures

1. Introduction to genetic aspects of sporadic, familial and hereditary cancers. Genetic basis of carcinogenesis.
2. Major groups of genes involved in carcinogenesis. High (oncogenes, tumour suppressor and mutator genes), moderate and low penetrance genes.
3.Autosomal dominant cancer susceptibility syndromes. Clinical characteristics, clinical and pedigree criteria for diagnosis, diagnostic methods, genetic counselling. Part I: retinoblastoma, Li-Fraumeni syndrome, Wilms syndrome.
4. Autosomal dominant cancer susceptibility syndromes. Clinical characteristics, clinical and pedigree criteria for diagnosis, diagnostic methods, genetic counselling. Part II: familial adenomatous polyposis, hereditary non-polyposis colorectal cancer.
5. Autosomal dominant cancer susceptibility syndromes. Clinical characteristics, clinical and pedigree criteria for diagnosis, diagnostic methods, genetic counselling. Part III: neurofibromatosis type I and II, hereditary melanoma, MEN1 and MEN2.
6. Autosomal recessive chromosomal instability syndromes.
7. The mechanism of metastasis. Clinical implications.
8. Molecular basis of mutagenesis, carcinogenesis and teratogenesis. Similarities and contrasts.
9. Personalised medicine as a new paradigm of patient management in the XXI century. Part I: significance of personalised medicine in oncology: in diagnosis, prognosis and treatment of breast, ovarian and colorectal cancers.
10. Personalised medicine as a new paradigm of patient management in the XXI century. Part II: significance of personalised medicine in diagnosis, prognosis and treatment of gastric cancer, brain tumours and malignant melanoma.
11. Personalised medicine in cardiology as a new paradigm of patient management in the XXI century.
12. Personalised medicine in endocrinology as a new paradigm of patient management in the XXi century; examples: diabetes and cystic fibrosis treatment.

13. Homeotic genes. Teratogy.
14. Genetic aspects of dementias.
15. Genetic and clinical aspects of mitochondrial disorders.

Seminars

Practical classes

1. Introduction to clinical genetics. Clinical genetics as a medical specialty. Types of genetic disorders. Genetic counselling. Definition. Principles and practice. Family history and construction of pedigrees. Diagnostic information. Calculation of recurrence risk. Communication. Breaking bad news.

2. Congenital anomalies. Aetiology, epidemiology, classification [malformation, dysplasia, dysruption, deformation; sequence, field defect, association, syndrome]. Isolated vs multiple anomalies. Teratogenic exposure. Common birth defects: heart anomalies; cleft lip and/or palate; club foot; congenital hip dislocation; neural tube defects.

3. Dysmorphology. Developmental delay/Intellectual disability. Definition. Nomenclature. Examination checklist. Syndrome diagnosis. "Facial gestalt". Psychomotor development: gross motor skills, social development, language. Intellectual disability.

4. Numerical aberrations of autosomal chromosomes. Basic concepts: polyploidy (triploidy, tetraploidy), aneuploidy, trisomy, monosomy, nondysjunction, mosaicism (examples: Pallister-Kilian syndrome and hypomelanosis of Ito), chimerism. Down syndrome. Patau syndrome. Edwards syndrome. Triploidy [karyotype and phenotype, natural history, medical issues and clinical management, recurrence risk, prenatal diagnosis, genetic counselling].

5. Aberrations of sex chromosomes. Sex chromosome aneuploidy: Turner syndrome, Klinefelter syndrome, trisomy X syndrome, 47,XYY syndrome [karyotype and phenotype, natural history, medical issues and clinical management, recurrence risk, prenatal diagnosis, genetic counselling]. Hypergonadotropic hypogonadism.

6. Structural chromosome aberrations. Basic concepts: deletion, inversion, insertion, isochromosome, duplication, balanced and unbalanced translocations; microaberrations, genomic imprinting. Syndromes: Wolf-Hirschhorn, cri du chat, Prader-Willi, Miller-Dieker, Angelman, DiGeorge, Williams, Beckwith-Wiedemann, Silver-Russell, Smith-Magenis [karyotype and phenotype, natural history, medical issues and clinical management, recurrence risk, prenatal diagnosis, genetic counselling].

7. Autosomal dominant inheritance. Basic concepts: characteristics of autosomal dominant inheritance, new mutations, expression, penetrance, recurrence risk, pleiotropy, somatic and germ-line mosaicism. Non-mendelian inheritance. Dynamic mutations. Anticipation. Disorders:, Marfan syndrome, *osteogenesis imperfecta*, neurofibromatosis type 1 and 2, familial hypercholesterolemia, Huntington's disease, ADPKD, skeletal dysplasias (achondroplasia , thanatophoric dysplasia, campomelic dysplasia), [gene, phenotype, natural history, medical issues and clinical management, recurrence risk, prenatal diagnosis, genetic counselling]. Special issues: presymptomatic testing.

8. Autosomal recessive inheritance. Basic concepts: characteristics of autosomal recessive inheritance, recurrence risk, heterogeneity (allelic and non- allelic), role of consanguinity, carrier status, founder effect. Disorders: cystic fibrosis, phenylketonuria, albinism, alkaptonuria, sickle-cell anaemia, spinal muscular atrophy, hemochromatosis, Wilson's disease, mukopolisacharydosis (I, II, III, VI), Smith-Lemli-Opitz syndrome [gene, phenotype, natural history, medical issues and clinical management, recurrence risk, prenatal diagnosis, genetic counselling].

9. X-linked inheritance. Basic concepts: characteristics of X-linked inheritance, recurrence risk, chromosome X inactivation, obligatory and potential carriers. Disorders: haemophilia (type A and B), fragile X syndrome (and *FMR1*-related disorders), hypophosphataemia, Dychenne's and Becker's muscular dystrophy, Rett syndrome, daltonism [gene, phenotype, natural history, medical issues and clinical management, recurrence risk, prenatal diagnosis, genetic counselling].

10. Sex determination. Disorders of sex development. Role of X and Y chromosomes in sex determination and differentiation. Dysmorphology of external genitalia. Disorders of sex development – revised nomenclature [androgene insensitivity syndromes, congenital adrenal hyperplasia, complete and mixed gonadal dysgenesis].

11. Prenatal diagnosis. Reproductive genetic counselling. Non-invasive methods of prenatal diagnosis (biochemistry, ultrasound markers of aneuploidy and monogenic disorders, free fetal DNA testing). Invasive methods of prenatal diagnosis (chorionic villus sampling, amniocentesis, cordocentesis). Indications for invasive prenatal diagnosis. Chromosome abnormalities: fertility problems and pregnancy loss. Preimplantation genetic diagnosis. Legal and ethical aspects.

12. Cancer predisposition syndromes. Sporadic, familial and "hereditary" cancers. Genetic testing for cancer susceptibility. Indications for molecular testing. Ethical and legal aspects. Genetic counselling and clinical management for mutation carriers. Syndromes: hereditary breast/ovarian cancer. Familial adenomatous polyposis. Hereditary non-polyposis colorectal cancer. Li-Fraumeni syndrome. Von Hippel-Lindau syndrome. Multiple endocrine neoplasia. Retinoblastoma.

13. Dysmorphology in practice.

14. Monogenic disorders in practice.

15. Cancer predisposition syndromes in practice.

16. Short stature: diagnostic algorithm and differential diagnosis.

17. Introduction._Structure and function of genes and chromosomes. Origin of genetic variation. Diagnosing genetic disorders - overview of laboratory techniques (cytogenetics and molecular biology).

18. Clinical cytogenetics._Human chromosomes (numerical and structural aberrations; polimorphisms). Sampling, transport and storing of biological material for cytogenetic analysis. Human karyotype. Methods of chromosome analysis (culture, staining). Indications for performing chromosome analysis (prenatal and postnatal).

19. Molecular cytogenetics. Fluorescence *in situ* hybridization (FISH). Probes. Using FISH as a diagnostic

method – examples. Comparative genomic hybridization (CGH and arrayCGH). Multiplex ligation-dependent probe amplification (MLPA).

20. International System of Cytogenetic Nomenclature.

21. Epigenetics. Epigenetic regulation of gene expression. Role of epigenome dysregulation in genetic syndromes (AS/PWS, SRS/BWS). Diagnosing imprinting defects.

22. Molecular methods in clinical genetics. Methods of DNA analysis (PCR and variations, sequencing, SNaPshot, NGS). Testing for known mutations. The founder effect. Common mutations and hot-spots. Mutation screening.

23. Oncogenetics. Laboratory investigations of cancer. Methods of detecting chromosome instability. Testing for cancer predisposition syndromes.

Other

Basic literature (list according to importance, no more than 3 items)

1. Medical Genetics (fourth edition) – LB Jorde, JC Carrey, MJ Bamshad
2. Essential Medical Genetics – M Connor, M Ferguson-Smith
3. Molecular Diagnosis of Genetic Diseases – R Elles

Additional literature and other materials (no more than 3 items)

1. Practical Genetic Counselling – PS Harper
2. A Practical Guide to Human Cancer Genetics – SV Hodgson, WD Foulkes, C Eng, ER Maher
3. Oxford Desk Reference Clinical genetics – HV Firth, JA Hurst

Didactic resources requirements (e.g. laboratory, multimedia projector, other...)
Multimedia projector, laptops, blackboard or whiteboard, chalk or markers

Preliminary conditions (minimum requirements to be met by the student before starting the module/course)
Knowledge of the genetic and molecular basis of disorders and inheritance.

Conditions to receive credit for the course (specify the form and conditions of receiving credit for classes included in the module/course, admission terms to final theoretical or practical examination, its form and requirements to be med by the student to pass it and criteria for specific grades)
Form of receiving credit: MQC tests, oral responses, short tests, case-based analysis
Conditions for receiving credit: gaining credit for two MCQ tests or the final tests, presence in no less than 90% of classes.

Grade:	**Criteria** (only for courses/modules ending with an examination)
Very Good (5.0)	>93% correct answers on the MCQ test
Good Plus	85-92% correct answers on the MCQ test

(4.5)	
Good (4.0)	77-84% correct answers on the MCQ test
Satisfactory Plus (3.5)	69-76% correct answers on the MCQ test
Satisfactory (3.0)	62-68% correct answers on the MCQ test

Name and address of module/course teaching unit, contact: telephone and e-mail address

DEPARTMENT OF GENETICS, WROCLAW MEDICAL UNIVERSITY, MARCINKOWSKIEGO 1, 50-368, WROCŁAW; contact person: Karolina Pesz, tel: 71 784 13 26; email: karolina.pesz@umed.wroc.pl

Coordinator / Person responsible for module/course, contact: telephone and e-mail address

Professor Maria M. Sąsiadek, tel. 71 784 12 55; email: maria.sasiadek@umed.wroc.pl

List of persons conducting specific classes: full name, degree/scientific or professional title, discipline, performed profession, form of classes.

Maria Sąsiadek – professor, Head of Department, clinical genetics, genetic consultant , lectures

Błażej Misiak – MD, PhD, clinical genetics, genetic consultant, classes

Karolina Pesz – MD, PhD, clinical genetics, genetic consultant, classes

Aleksandra Jakubiak – MD, PhD student, clinical genetics, genetic consultant, classes

Izabela Łaczmańska - MSc, PhD, medical genetics, diagnostician, classes

Paweł Karpiński – MSc, PhD, medical genetics, researcher, classes

Justyna Gil - MSc, PhD, medical genetics, diagnostician, classes

Date of Syllabus development

27.06.2016

Syllabus developed by

Karolina Pesz

Signature of Head of teaching unit

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Prodziekan ds. Studiów
w Języku Angielskim
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