Course description

Part 1

General information about the course	
1. Major of study: obstetric	**2. Study level:** unified MSc **3. Form of study:** intramural
4. Year: I	**5. Semester:** I, II
6. Course name: Embryology and Genetics	
7. Course status: required	
8. Course contents and assigned learning outcomes - Providing knowledge about spermatogenesis, spermiogenesis, oogenesis, insemination, fertilization, early stages of the entire human body development and development of specific systems and organs - Providing knowledge about placenta structure and functions - Providing knowledge about nucleotide functions in the cells, primary and secondary DNA and RNA structures, structure of chromosomes and chromatin; molecular basis of the mutagenesis - Providing knowledge about inheritance of the different numbers of traits, inheritance of the quantitative traits, independent inheritance and X- linked, inheritance of the extranuclear genetic information - Providing knowledge about genetical determined diseases in the context of cancer prevention, prenatal diagnostics and genetical counselling. - Providing knowledge about genetic determining of the human blood groups and serological conflict in the Rh system. - Development of skills of using dysmorphological diagnostics in genetical and immunological examination - Development of skills of disease risk estimation using inheritance principles and impact of environmental factors - Raising awareness about the application of the principles of general ethics and professional contact with the people affected by diseases and their parents Learning outcomes / reference to learning outcomes indicated in the standards For knowledge – student knows and understands: A.W11. A.W12. A.W13. A.W14. A.W15. A.W16. For skills student can do: A.U4. A.U5. For social competencies student is ready to:	
9. Number of hours for the course	**45**
10. Number of ECTS points for the course	**3**

11. Methods of verification and evaluation of learning outcomes		
Learning outcomes	Methods of verification	Methods of evaluation*
Knowledge	choice test	*
Skills	choice test	*
Competencies	choice test	*

* The following evaluation system has been assumed:

Very good (5,0) – the assumed learning outcomes have been achieved and significantly exceed the required level
Better than good (4,5) – the assumed learning outcomes have been achieved and slightly exceed the required level
Good (4,0) – the assumed learning outcomes have been achieved at the required level
Better than satisfactory (3,5) – the assumed learning outcomes have been achieved at the average required level

Satisfactory (3,0) – the assumed learning outcomes have been achieved at the minimum required level
Unsatisfactory (2,0) – the assumed learning outcomes have not been achieved

Course description

Part 2

Other useful information about the course	
12. Name of Department, mailing address, e-mail: Department of Biochemistry and Medical Genetics, 40-752 Katowice, Medyków Street 18 phone: +48 322528432; 4th floor, room 437 biochigen.sum.edu.pl	
13. Name of the course coordinator: Prof. Paweł Niemiec, PhD.	
14. Prerequisites for knowledge, skills and other competencies: Basic knowledge of embryology and human genetics at the high school level.	

15. Number of students in groups	In accordance with the Senate Resolution
16. Study materials	Notice board, Department of Biochemistry and Medical Genetics www.biochigen.sum.edu.pl
17. Location of classes	Department of Biochemistry and Medical Genetics, workroom no 10, building C1, Medyków 18
18. Location and time for contact hours	Department of Biochemistry and Medical Genetics, room 437, building C2, Medyków 18

19. Learning outcomes

Number of the course learning outcome	Course learning outcomes	Reference to learning outcomes indicated in the standards
C_K01	Student describes spermatogenesis, spermiogenesis, oogenesis, insemination, fertilization, early stages of human body development, specified systems and organs	A.W11.
C_K02	Characterize development, structure and functions of placenta	A.W12.
C_K03	Explains genetics of human blood groups system and causes of serological conflict in the Rh system.	A.W13.
C_K04	Explains utility of knowledge about genetic- based diseases in cancer prevention and prenatal diagnostics	A.W14.
C_K05	Links chromosomal aberrations with specified disease entities	A.W14.
C_K06	Notice necessity of genetic counselling	A.W14.
C_K07	Describes function of nucleotides in human cells	A.W14.
C_K08	Have knowledge about primary and secondary DNA structure, mention bonds stabilizing these structures	A.W14.
C_K09	Describes structure od chromatin	A.W14.
C_K010	Describes structure of chromosomes and molecular basics of mutagenesis	A.W14.
C_K011	Characterizes main inheritance laws of different number of traits, inheritance quantitative traits, independent inheritance of traits and extranuclear genetic information	A.W15.
C_S01	Differs the most common malformations and chromosomal diseases using dysmorphology diagnostics	A.W16., A.U4.
C_S02	Assigns techniques used in genetic and immunological examination for	A.U5.

	proper diagnostic applications	
C_S03	Estimates disease risk using inheritance laws and impact of environmental factors.	A.U4.

20. Forms and topics of classes	**Number of hours**
21.1. Lectures	**40**
Structure and function of genetic material - basic genetic terms; structure of DNA, RNA, chromatin, gene, genome; the mitochondrial genome; cell cycle and replication - basic assumptions; genetic code and expression of genetic information - basic assumptions.	3
Variability and inheritance. Noninherited variability- modificational, fluctuative. Inherited variability: recombination, mutation. Molecular basis of mutagenesis - formation of single-gene and chromosomal (structural and numerical) mutations. Spontaneous and induced mutations. Mutagenic factors - physical, chemical, biological. Repair of mutations and DNA damage.	3
Basic principles of inheritance - single gene inheritance. Features of autosomal dominant and recessive inheritance. Features of sex-linked dominant and recessive inheritance. Incomplete dominance, codomination, multiple alleles. Examples of autosomal dominant inherited diseases (achondroplasia, myotonic dystrophy, Marfan's syndrome, Huntington's disease, osteogenesis imperfecta) and recessive (cystic fibrosis, sickle cell anemia, monogenic metabolic blocks - tyrosinemia, phenylketonuria, alkaptonuria, albinism). Examples of sex-linked diseases, recessive (Duchenne and Becker muscular dystrophy) and dominant (hypophosphatemic rickets types I and II, fragile X chromosome syndrome).	3
Basic principles of inheritance - multi-gene inheritance. The interaction of many genes in the development of one trait of a cumulative, complementary and epistatic character. Interactions between genetic and environmental factors in shaping the phenotype. Opportunity, odds ratio, risk, synergy. Examples of multigene and multifactorial diseases: ischemic heart disease, diabetes mellitus type I and II, hypertension, mental diseases, autoimmune diseases and neurodegenerative diseases.	3
Genetics and cancer prevention. The basis of neoplastic diseases: proto-oncogenes, suppressor genes, repair factors. Models of tumor formation: two-stroke and multi-stroke. The most common hereditary neoplasms: breast and ovarian cancer (mutations of BRCA1 and BRCA2 genes), colorectal cancer (mutations of MLH1, APC genes). TP53 gene and Li-Fraumeni syndrome. DNA repair deficits - Xeroderma pigmentosum. Personalized medicine in the treatment of cancer.	3
Principles of genetic counseling - part 1. Conditions determining the validity of genetic counseling. Elements of genetic counseling. Cytogenetic methods and molecular biology techniques used in the diagnosis of genetic diseases.	3
Principles of genetic counseling - part 2. Prenatal diagnosis - non-invasive methods (USG, Doppler examination) and invasive methods (chorionic villus sampling, amniocentesis, cordocentesis, fetoscopy). Genetic preimplantation diagnostics. Gene therapy.	2
The formation of reproductive cells, characterization of spermatogenesis, spermiogenesis, oogenesis with hormonal regulation. Female sexual cycle- ovary and menstrual, morphological changes, changes after ovulation completed with pregnancy. Insemination, capacitation - mechanisms counteracting polyspermia, cortical reaction. Fertilization- stages, multiple pregnancy, in vitro fertilization.	3
Early stage of prenatal development- characteristics of preembryonic stage: cleavage, morula, blastocyst, implantation into the endometrium, differentiation of the embryonic node - epiblast, hypoblast, syncytiotrophoblast formation. The formation of the three germ layers, the division and fate of the mesoderm, the formation of somites, their differentiation and fate. Placenta- structure and function. Fetal membranes and other extraembryonic structures- formation and functions	3
Organogenesis- development of primary organs. Formation of the central nervous system, neurulation, role of the neural tube in the development of the embryo, the development of the sense organs. Formation of cardiovascular, musculoskeletal, respiratory, digestive, urogenital systems, limb formation. Characteristics of the period: embryonic and fetal prenatal development of humans.	3
Characterization of differentiation and morphogenesis. Migration, fusion, apoptosis- role in morphogenetic degradation. Characteristics of contact inhibition. Characteristics of embryonic induction.	3
Regulation of prenatal development at the molecular level. The most important signalling pathways related to the development of the organism. Genes involved in the control of embryonic development. Participation of homeotic genes - the role of homeobox genes in the formation of an organism pattern.	2
Disorders of the prenatal development process - part 1. Environmental threats of embryonic development - teratogens. Factors influencing the teratogen effect, critical periods of sensitivity. Effects of chemical teratogenic factors: alcohol, smoking, hormones, medicines, drugs. Effects of infectious teratogenic factors: viruses (rubella, cytomegalovirus, herpes, chickenpox, herpes zoster, influenza, HIV), protozoa (*Toxoplasma gondi*), bacteria (Pale spirochete). Effects of the physical teratogenic factors: ionizing	3

radiation, noise, temperature. Effects of maternal teratogenic factors: diabetes mellitus, phenylketonuria.	
Disorders of the prenatal development process - part 2. Congenital disorders of various etiologies: classification, Polish Register of Congenital Defects. Pathogenetic mechanism of congenital disorders: disruptions, deformations, malformations, displasia. Characteristics of small congenital disorders, examples, dysmorphologic features. Examples of large congenital disorders, taking into account their different localization: CNS defects, heart defects, kidney defects, limb defects. Etiology of large congenital disorders. Examples of monogenic large congenital disorders: achondroplasia, brittle bone syndrome, Marfan syndrome, Duchenne muscular dystrophy. Examples of large congenital disorders of chromosomal etiology (Down syndrome, Turner syndrome, Williams syndrome). Primary and secondary prevention of congenital disorders.	3
22.2. Seminars	
23.3. Labs	**5**
Estimation of genetics diseases risk. Monogenic disease inheritance (autosomal dominant, recessive; X- linked dominant, recessive) - lineage analysis, mono- and multiple gene crosses. Populational risk. Impact of environmental factors in determining monogenic diseases (phenylketonuria, hyperhomocysteinemia). Non-nuclear inheritance - genetic crosses. Genetic conditions of blood groups and causes of serological conflict in the Rh system - genetic crosses.	
Dysmorphological diagnostics of genetical diseases. Structural and numerical chromosomal aberrations. Dysmorphic features in the most common chromosomal syndromes (Down, Klinefelter, Turner, Edwards, Patau syndrome), microdeletion syndromes (Cri du chat, Prader-Willi, Angelman, Williams, Wolf-Hirschhorn syndrome) and monogenic diseases (Marfan syndrome, Achondroplasia, Fragile X syndrome).	

24. Readings

1. Alberts B et al. Molecular biology of the cell. New York : Garland Science, 2008.

2. Jorde, Lynn B. Medical Genetics. Philadelphia : Mosby Elsevier, 2010.

3. Bartel H. Embriologia. Podręcznik dla studentów. Wydanie IV. PZWL. Warszawa 2005. (Embriology. Textbook for the students. 4th edition. PZWL. Warszawa 2005.

4. Kurpisz Maciej (red.). Molekularne podstawy rozrodczości człowieka i innych ssaków. Wydanie I. Poznań 2002. (Molecular basis of reproduction of humans and other mammals. 1st edition. Poznań 2002).

5. Jura Czesław, Klag Jerzy (red.). Podstawy embriologii zwierząt i człowieka. Tom 1 i 2. Wydanie I. Wydawnictwo Naukowe PWN. Warszawa 2005. (Fundamentals of animal and human embryology. Volume 1st and 2nd. 1st edition.)

6. Friedman J. Genetics. Baltimore : Williams and Wilkins, 1992.

7. Sadler Thomas W. Langman's essential medical embryology. Lippincott Williams & Wilkins. Philadephia 2005.

25. Detail evaluation criteria

In accordance with the recommendations of the inspection bodies
Completion of the course – student has achieved the assumed learning outcomes
Detail criteria for completion and evaluation of the course are specified in the course regulations