Annex No. 1a

Course description
Part 1

General information about the course	
1. Major of study: Obstetrics **2. Study profile**: general academic	**3. Study level:** the first degree study **4. Form of study**: intramural **5. Cycle:** 2026 - 2029
6. Year: I	**7. Semestr:** I
8. Course name: Genetics	
9. Course status: obligatory	

10. Course contents.

To provide knowledge about the genetic basis of human blood groups and serological conflict in the Rh system.
To equip graduates with knowledge of chromosome structure and the molecular basis of mutagenesis.
To equip graduates with knowledge regarding the principles of inheritance of multiple traits, quantitative trait inheritance, independent inheritance, sex-linked inheritance, as well as extranuclear genetic inheritance.
To provide knowledge about genetically determined diseases and their significance in diagnostics, including prenatal diagnostics.
To equip graduates with knowledge of modern techniques used in genetic research.
To provide knowledge about the processes of spermatogenesis, spermiogenesis, and oogenesis, as well as insemination and fertilization.
To equip graduates with knowledge of human embryonic development, as well as the structure and function of fetal membranes and the placenta, and the stages of development of individual organs.
To provide knowledge about the genetic and immunological causes of infertility.
To develop the ability to assess the risk of a disease manifesting based on principles of inheritance and the influence of environmental factors.
To develop the ability to apply knowledge of the genetic determinants of diseases in disease prevention and prenatal diagnostics.
To develop awareness of the importance of acting in the best interest of the patient, respecting the dignity and autonomy of persons under care, demonstrating understanding of differences in worldview and culture, and showing empathy in relationships with the patient, her family, or caregiver within the professional competences of a midwife.

Assigned learning outcomes:

In terms of knowledge, the student knows and understands: A.W13, A.W14, A.W15, A.W16, A.W17, A.W18, A.W19, A.W20.
In terms of skills, the student is able to: A.U5, A.U6.
In terms of social competences, the student: is aware of the importance of acting in the best interest of the patient, respecting the dignity and autonomy of persons under care, demonstrating understanding of differences in worldview and culture, and showing empathy in the relationship with the patient, her family, or caregiver within the professional competences of a midwife.

11. Number of hours for the course (contact/ communication hours/ self-study)	**42/-/20**	**12. Number of ECTS points for the course**	**2**
13. Form of passing the course: grade credit			

14. Methods of verification and evaluation of learning outcomes		
Learning outcomes	Methods of verification	Methods of evaluation
Knowledge	One- choice test	Obtaining at least 70% correct answers

Skills	One- choice test	Obtaining at least 70% correct answers
Competencies	Observation	

***** The following evaluation system has been assumed:

Very good (5.0) – the intended learning outcomes have been achieved and significantly exceed the required level.

Above good (4.5) – the intended learning outcomes have been achieved and clearly exceed the required level.

Good (4.0) – the intended learning outcomes have been achieved and exceed the required level to some extent.

Fairly good (3.5) – the intended learning outcomes have been achieved at a level slightly above the required standard.

Satisfactory (3.0) – the intended learning outcomes have been achieved at the required level.

Fail (2.0) – the intended learning outcomes have not been achieved.

Course description
Part 2

Other useful information about the course	
15. Name of Department, mailing address, e-mail: Department of Biochemistry and Medical Genetics, 40-752 Katowice Medyków str. 18, phone 32 208 88 64, biogen@sum.edu.pl	
16. Name of the course coordinator: PhD Paweł Niemiec prof. SUM	
17. Prerequisites for knowledge, skills and other competencies: Basic knowledge about genetics and embryology.	
18. Number of students in groups:	In accordance with the Ordinance of the Rector
19. Study materials	http://biochigen.sum.edu.pl, notice board of Department of Biochemistry and Medical Genetics
20. Location of classes	Department of Biochemistry and Medical Genetics, workroom no 10, building C1, Medyków 18, Lecture room of School of Health Sciences
21. Location and time for contact hours	http://biochigen.sum.edu.pl

22. Learning outcomes

Number of the course learning outcome	Course learning outcomes	Reference to the learning outcomes specified in (underline as appropriate): the educational standards / approved by the Senate of the Medical University of Silesia
P_W1	Describes the processes of spermatogenesis, spermiogenesis, oogenesis, fertilization, and conception.	A.W13.
P_W2	Describes the stages of human embryonic development, the structure and function of fetal membranes and the placenta, as well as the stages of development of individual organs.	A.W14.
P_W3	Explains the genetic determinants of human blood groups and the causes of Rh serological conflict.	A.W15.
P_W4	Presents the structure of chromosomes and the molecular basis of mutagenesis.	A.W16.
P_W5	Characterizes the principles of inheritance of multiple traits, quantitative traits, independent assortment, and the inheritance of extranuclear genetic information.	A.W17.
P_W6	Explains issues related to genetically determined diseases and their importance in diagnostics, including prenatal diagnosis.	A.W18.
P_W7	Explains genetic and immunological causes of infertility.	A.W19.
P_W8	Describes modern techniques used in genetic research.	A.W.20

P_U1	Assesses the risk of disease occurrence based on the principles of inheritance and the influence of environmental factors.	A.U5.
P_U2	Applies knowledge of genetic disorders in disease prevention and prenatal diagnosis.	A.U6.
P_K01	Is aware of the importance of acting in the best interest of the patient, respecting the dignity and autonomy of individuals under care, showing understanding of different worldviews and cultural differences, and demonstrating empathy in relationships with the patient, her family, or caregiver within the professional competence of a midwife.	Point 1.1 of the general learning outcomes

23. Teaching methods: Lectures, multimedia methods, problem-based learning, demonstration, video-based learning, interactive exercises, case study, group discussion.

24. Forms and topics of classes	**Number of hours (contact)**
24.1. Lectures	**22**
1. Structure and function of genetic material – basic genetic concepts; structure of DNA, RNA, chromatin, chromosome, gene, and genome; mitochondrial genome; the cell cycle and replication – basic principles; the genetic code and gene expression – basic principles.	**5**
2. Basic principles of inheritance – monogenic inheritance. Examples of autosomal dominant diseases (achondroplasia, myotonic dystrophy, Marfan syndrome, Huntington's disease, osteogenesis imperfecta) and autosomal recessive diseases (cystic fibrosis, sickle cell anemia, single-gene metabolic disorders – tyrosinemia, phenylketonuria, alkaptonuria, albinism). Examples of X-linked diseases, recessive (Duchenne and Becker muscular dystrophy) and dominant (hypophosphatemic rickets types I and II, fragile X syndrome).	**3**
3. Basic principles of inheritance – polygenic inheritance. The interaction of multiple genes in the development of a single trait, with cumulative, complementary, and epistatic effects. Interactions between genetic and environmental factors in shaping the phenotype. Chance, odds ratio, risk, synergy. Examples of polygenic and multifactorial diseases: ischemic heart disease, type I and type II diabetes, hypertension, mental disorders, autoimmune diseases, and neurodegenerative diseases.	**3**
4. Formation of germ cells; characteristics of the processes of spermatogenesis, spermiogenesis, and oogenesis. Hormonal regulation of gametogenesis. The female reproductive cycle – ovarian and menstrual cycles, morphological changes, post-ovulatory changes ending in pregnancy. Sperm deposition and capacitation, mechanisms preventing polyspermy, cortical reaction. Fertilization. Multiple pregnancy. Infertility and sterility – genetic and immunological causes. Assisted reproductive techniques. The placenta – structure and functions. Fetal membranes and other extraembryonic structures – formation and functions.	**5**
5. Early period of prenatal development – cleavage, morula, blastocyst, implantation into the uterine mucosa, differentiation of the embryonic node – epiblast and hypoblast, formation of the syncytiotrophoblast. Formation of the three germ layers, division and fate of the mesoderm, formation of somites, their differentiation and developmental fate.	**3**
6. Characteristics of processes occurring during differentiation and morphogenesis. Cell migration, fusion, and apoptosis – their significance in morphogenetic remodeling. Characteristics of contact inhibition. Features of the process of embryonic induction. Regulation of prenatal development at the molecular level. The most important signaling pathways involved in organismal development. Developmental regulatory genes. The role of	**3**

homeotic genes – the function of homeobox genes in establishing the body plan of the organism.	
24.2. Seminars	**10**
1. Molecular basis of mutagenesis – the formation of single-gene mutations and chromosomal mutations (structural and numerical). Spontaneous and induced mutations. Mutagenic factors – physical, chemical, and biological.	**3**
2. Principles of genetic counseling. Conditions determining the justification for genetic counseling. Elements of a genetic consultation. Cytogenetic methods and molecular biology techniques used in the diagnosis of genetic diseases. Prenatal diagnostics – non-invasive methods (ultrasound, Doppler examination) and invasive methods (chorionic villus sampling, amniocentesis, cordocentesis, fetoscopy). Preimplantation genetic diagnosis. Gene therapy.	**2**
3. Disorders of prenatal development – Part 2. Congenital defects of various etiology: classification, Polish Registry of Congenital Malformations. Pathogenetic mechanisms of congenital defects: disruptions, deformations, malformations, dysplasias. Examples of major congenital defects according to localization: central nervous system defects, heart defects, genitourinary system defects, digestive system defects, limb defects. Etiology of major developmental defects. Examples of major congenital defects of monogenic etiology: achondroplasia, osteogenesis imperfecta (brittle bone syndrome), Marfan syndrome, Duchenne muscular dystrophy. Examples of major congenital defects of chromosomal etiology: Down syndrome, Turner syndrome, Williams syndrome. Primary and secondary prevention of congenital defects.	**2**
4. Disorders of prenatal development – Part 2. Environmental threats – teratogens. Factors influencing the effect of a teratogen, critical periods of susceptibility. Characteristics of the effects of chemical teratogens: alcohol, cigarette smoking, hormones, medications, drugs, infectious teratogens: viruses (rubella, cytomegalovirus, herpes simplex, varicella, shingles, influenza, HIV), protozoa (Toxoplasma gondii), bacteria (Treponema pallidum), physical teratogens: ionizing radiation, noise, temperature, and maternal teratogenic factors: diabetes, phenylketonuria.	**3**
24.3. Classess	**10**
1. Estimation of the risk of genetic diseases – Inheritance of monogenic disorders (autosomal recessive, autosomal dominant, X-linked recessive, X-linked dominant) – pedigree analysis, mono- and multi-gene crosses. Population risk. Contribution of environmental factors to the development of monogenic diseases (phenylketonuria). Extranuclear (non-nuclear) inheritance – cross analysis. Genetic determinants of blood groups and causes of serological conflict in the Rh system – cross analysis.	**3**
2. Dysmorphological diagnosis of genetically determined diseases. Structural and numerical chromosomal aberrations. Dysmorphic features in the most common chromosomal syndromes (Down syndrome, Klinefelter syndrome, Turner syndrome, Edwards syndrome, Patau syndrome), microdeletion syndromes (Cri-du-chat syndrome, Prader–Willi syndrome, Angelman syndrome, Williams syndrome, Wolf–Hirschhorn syndrome), and monogenic diseases (Marfan syndrome, achondroplasia, Fragile X syndrome).	**2**
3. Individual stages of the embryonic period. Organogenesis – development of primary organs. Formation of the central nervous system, the process of neurulation, the role of the neural tube in embryonic development, development of the sensory organs. Formation of the following systems: cardiovascular, musculoskeletal, respiratory, digestive, and genitourinary systems; formation of the limbs.	**3**
4. Characteristics of the processes occurring in individual weeks of embryonic and fetal life.	**2**

24.6. Self-study	**20**
25. Total ECTS credits:	**2**

ACTIVITY	NUMBER OF HOURS / STUDENT WORKLOAD
Scheduled contact hours with the teacher	**42**
Preparation for different types of classes	
Participation in consultations	
Time for project preparation/documentation and self-directed learning	
Self-study	**20**
Preparation for the examination	
Participation in the examination	
Practical training	
Clinical practice	
TOTAL NUMBER OF HOURS	**62**
TOTAL NUMBER OF ECTS CREDITS	**2**

26. References:

1. Larsen's human embryology. Gary C Schoenwolf Autor; Steven B Bleyl Autor; Philip R Brauer Autor; Philippa H Francis-West Autor; Elsevier (Amsterdam). 2021
2. Netter's atlas of human embryology. Larry R Cochard Autor; Angelique N Dueñas Autor; Frank Henry Netter (1906-1991). Elsevier (Amsterdam). 2025

27. 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