Annex No. 1a

Course description
Part 1

General information about the course	
1. Major of study: Nursing **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. Providing knowledge about the function of the genome, transcriptome and human proteome. Providing knowledge about the basic concepts of gene expression regulation, including epigenetic regulation. Providing knowledge about the structure of chromosomes and the molecular basis of mutagenesis. Providing knowledge of the principles of inheriting different numbers of traits, inheriting quantitative traits, independent inheritance of traits, and inheritance of non-nuclear genetic information. Developing skills to estimate the risk of disclosure of a given disease based on the principles of inheritance and the impact of environmental factors. Developing skills to use knowledge about genetically determined diseases in cancer prevention and prenatal diagnosis. **Assigned learning outcomes:** For knowledge – student knows and understands: C.W11, C.W12, C.W13, C.W14, C.W15. For skills student can do: C.U5, C.U6	

11. Number of hours for the course (contact/ communication hours/ self-study)	**30/-/15**	**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_W01	Knows the genetic determinants of human blood groups and Rh incompatibility (Rh serological conflict).	A.W11.
P_W02	Understands the issues related to genetically determined diseases.	A.W12.
P_W03	Knows the structure of chromosomes and the molecular basis of mutagenesis.	A.W13.
P_W04	Describes the principles of inheritance of multiple traits, the inheritance of quantitative traits, independent assortment of traits, and the inheritance of extranuclear genetic information.	A.W14.
P_W05	Describes modern genetic research techniques.	A.W15.
P_U01	Is able to estimate the risk of the occurrence of a given disease based on the principles of inheritance and the influence of environmental factors.	A.U5.
P_U02	Is able to use the determinants of genetic diseases in disease prevention.	A.U6.
23. Teaching methods: Lectures, multimedia methods, problem-based learning, interactive exercises, genetic crosses, group discussion.		
24. Forms and topics of classes		**Number of hours (contact)**

24.1. Lectures	**20**
1. Structure and function of genetic material - basic genetic concepts; 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**
2. Variation and heredity - Hereditary variability: recombination and mutational. Molecular basis of mutagenesis - formation of single-gene and chromosomal mutations. Spontaneous and induced mutations. Mutagenic factors - physical, chemical, biological. Repair of mutations and DNA damage.	**3**
3. Mechanisms of epigenetic inheritance - The main mechanisms of epigenetic control of gene expression - DNA methylation, histone acetylation, RNAi (miRNA, dsRNA). Disturbance of the epigenetic profile and diseases. Factors causing epigenetic changes. Parental imprinting. Characteristics of X chromosome inactivation - the role of the XIST gene and its methylation.	**3**
4. Basic principles of inheritance - single gene inheritance. Features of autosomal dominant and recessive inheritance. Features of X-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 X-linked diseases, recessive (Duchenne and Becker muscular dystrophy) and dominant (hypophosphatemic rickets types I and II, fragile X chromosome syndrome).	**3**
5. Basic principles of inheritance - multi-gene inheritance. The interaction of many genes in conditioning one trait of a cumulative, complementary and epistatic nature. Interactions between genetic and environmental factors in determining phenotype. Opportunity, odds ratio, risk, synergy. Examples of multigenic and multifactorial diseases: coronary artery disease, diabetes mellitus type I and II, arterial hypertension, mental, autoimmune and neurodegenerative diseases.	**3**
6. Principles of genetic counseling - Conditions determining the validity of genetic counseling. Elements of genetic counseling. Cytogenetic methods and molecular biology techniques used in the diagnosis of genetic diseases. Prenatal diagnosis - non-invasive methods (USG, Doppler examination) and invasive methods (chorionic villus sampling, amniocentesis, cordocentesis, fetoscopy). Genetic preimplantation diagnostics. Gene therapy.	**3**
7. Personalized medicine. Traditional approach and personalized medicine. Factors influencing the effectiveness of traditional therapies. Molecular stratification of patients. Pharmacogenetics and pharmacogenomics. Examples of the use of personalized medicine in oncology, cardiology and other fields of medicine.	**2**
24.2. Seminars	
24.3. Classess	**10**
1. Estimating the risk of genetic diseases. Inheritance of monogenic diseases (autosomal recessive, autosomal dominant, X-linked recessive, X-linked dominant) - pedigree analysis, single-gene crosses.	**2**
2. Non-nuclear inheritance. Environmental factors. Population risk. The role of environmental factors in conditioning single-gene diseases (phenylketonuria, hyperhomocysteinemia). Genetic determinants of blood groups and causes of serological conflict in the Rh system - crossbreeds.	**2**
3. Multi-gene and multi-factor inheritance. Gene interaction: cumulative genes, complement, epistasis. Several-gene crosses.	**2**
4. Dysmorphological diagnosis of genetic diseases. Structural and numerical chromosomal aberrations. Dysmorphic features in the most common	**2**

chromosomal syndromes (Down, Klinefelter, Turner, Edwards, Patau syndrome), microdeletion syndromes (Cr idu chat, Prader-Willi, Angelman, Williams, Wolf-Hirschhorn syndrome).	
5. The use of genetics in medicine.	**2**
24.6. Self-study	**15**
25. Total ECTS credits:	

ACTIVITY	**NUMBER OF HOURS / STUDENT WORKLOAD**
Scheduled contact hours with the teacher	**30**
Preparation for different types of classes	
Participation in consultations	
Time for project preparation/documentation and self-directed learning	
Self-study	**15**
Preparation for the examination	
Participation in the examination	
Practical training	
Clinical practice	
TOTAL NUMBER OF HOURS	**45**
TOTAL NUMBER OF ECTS CREDITS	**2**

26. References:

1. Alberts B et al. Molecular biology of the cell. New York: Garland Science, 2008.
2. Jorde LB et al. Medical Genetics. Elsevier, 2015.
3. Epstein RJ. Human Molecular Biology. Cambridge: Cambridge University Press, 2003.
4. Connor M., Ferguson-Smith M. Essential Medical Genetics. Wiley-Blackwell, 1997.

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