Course description

Part 1

General information about the course	
1. Major of study: Physiotherapy	**2. Study level:** unified MSc **3. Form of study:** intramural
4. Year: 2022-2023	**5. Semester:** I
6. Course name: Genetics	
7. Course status: required	
8. Course contents and assigned learning outcomes Objectives of the course: 1. To give the knowledge of the genetic determinants of disease development in the human population. 2. To give the with knowledge of genetic and phenotype-related determinants of motor skills. Learning outcomes / reference to learning outcomes indicated in the standards For knowledge – student knows and understands: A.W20, A.W21	
9. Number of hours for the course	16
10. Number of ECTS points for the course	1

11. Methods of verification and evaluation of learning outcomes

Learning outcomes	Methods of verification	Methods of evaluation*
Knowledge	Grade credit – MCQ Report	*
Skills		*
Competencies		*

* 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
Unstatisfactory (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, ul. Medyków 18, 40-752 Katowice, tel. 32 252 84 32	
13. Name of the course coordinator: dr hab. n. med. Paweł Niemiec, prof. SUM	
14. Prerequisites for knowledge, skills and other competencies: Knowledge of the basics of inheritance.	

15. Number of students in groups	In accordance with the Senate Resolution
16. Study materials	E-learning SUM platform https://eduportal.sum.edu.pl/ Website of the Department of Biochemistry and Medical Genetics http://biochigen.sum.edu.pl
17. Location of classes	Department of Biochemistry and Medical Genetics, ul. Medyków 18, 40-752 Katowice, building C1, room 10
18. Location and time for contact hours	On the website of the Department of Biochemistry and Medical Genetics http://biochigen.sum.edu.pl

19. Learning outcomes

Number of the course learning outcome	Course learning outcomes	Reference to learning outcomes indicated in the standards
C_K01	To give the knowledge of the genetic determinants of disease development in the human population.	A. W20
C_K02	To give the with knowledge of genetic and phenotype-related determinants of motor skills.	A. W21

20. Forms and topics of classes	**Number of hours**
21.1. Lectures	**6**
1. Basics of multi-gene inheritance. Multi-gene inheritance - cooperation of genes in determining one cumulative trait, complementary and epistasis. Interactions between genetic and environmental factors in determining the phenotype. Odds ratio, risk, synergy. Introduction to the genetics of ischemic heart disease. Monogenic forms of cardiovascular diseases (exemplified by familial hypercholesterolaemia). Polymorphisms of genes encoding key proteins involved in atherosclerosis (exemplified by genes involved in the regulation of lipid metabolism and blood pressure).	3
2. Multigenic and multifactorial diseases. Fundamentals of genetics of type I and II diabetes, hypertension, mental, autoimmune and neurodegenerative diseases. Monogenic forms of diseases. Mutations in candidate genes, polymorphisms of genes influencing susceptibility.	3
22.2. Seminars	

23.3. Labs	**10**
1. Gene diseases: Mutational variability - gene mutations. Spontaneous and induced mutations, mutagens. Examples of monogenic genetic diseases and features of autosomal dominant inheritance (achondroplasia, myotonic dystrophy, Marfan syndrome, Huntington's disease, osteogenesis imperfecta) and recessive (monogenic metabolic blocks - tyrosinemia, phenylketonuria, alkaptonuria, albinism) in humans. Examples of diseases and features of sex-linked, recessive (Duchenne and Becker muscular dystrophy) and dominant (hypophosphatemic rickets types I and II, fragile X syndrome) inheritance in humans. Risk assessment of monogenic diseases.	2
2. Chromosomal diseases: Chromosomal mutations (structural and numerical). Disease syndromes caused by autosomal structural aberrations, microdeletions, translocations. Numerical chromosomal mutations, aneuploids. Autosomal trisomes: chromosome 13 (Patau's syndrome), chromosome 18 (Edwards' syndrome), chromosome 21 (Down's syndrome), chromosome 22. Trisomes of sex chromosomes: XXY, XXX, XYY. X chromosome monosomy (Turner syndrome). Dysmorphological diagnosis.	2
3. Genetic counseling. Goals and rules of genetic counseling. Application of cytogenetic methods in the diagnosis of chromosomal aberrations. Prenatal diagnosis. Invasive and non-invasive prenatal testing methods and their use in the diagnosis of genetic diseases and birth defects. Genetic preimplantation diagnostics.	2
4. Genetic determinants of motor skills: Heritability of individual components of human motor fitness. Genetic determinants of differentiation of individual types of muscle fibers. Mutations and polymorphisms of genes influencing motor performance (*IGF-1, MSTN, ACTN3, EPOR, VDR, ACE*). Genetic doping.	2
5. The use of genetics in medicine. Personalized medicine. Gene therapy. Epigenetic inheritance in different pathologies.	2
24.4. Self-education	9
25. Readings	
1. Jorde, Lynn B. Medical Genetics. Philadelphia : Mosby Elsevier, 2010. 2. Friedman J. Genetics. Baltimore : Williams and Wilkins, 1992.	
26. 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	