

Human Genetics

HEALTH SCIENCES			
FACULTY	HEALTH SCIENCES		
DEPARTMENT	NURSING		
LEVEL OF EDUCATION	UNDERGRADUATE		
LESSON CODE	0805.5.009.0	SEMESTER OF STUDIES	5 th
COURSE TITLE	HUMAN GENETICS		
SELF-ENDED TEACHING ACTIVITIES		HOURS OF TEACHING / WEEK	CREDIT UNITS
Theory		2	
Coaching school			
Laboratory			
Clinical exercise			
Total		2	3
COURSE TYPE:	ELECTIVE COMPULSORY		
PREREQUISITE COURSES:	NO		
LANGUAGE OF TEACHING and EXAMINATIONS:	Greek		
THE COURSE IS OFFERED TO ERASMUS STUDENTS:	NO		
WEBSITE COURSE	https://eclass.hmu.gr/courses/NURS229/		
Learning results			
<p>The course aims to introduce the fundamentals of human genetics.</p> <p>A big part of the course concerns on the role of genetics on human health and the interactions between basic scientific knowledge and clinical application. The genetic, molecular and chromosomal basis of hereditary diseases, cancer and other diseases with a genetic component, as well as other pathological conditions are comprehensively presented. The module covers the disciplines of genetic diagnostics, genetic counseling and the emerging field of genetic medicine.</p> <p>After completing the course, students will be able to:</p> <ul style="list-style-type: none"> • know the organization of human genome and the molecular etiology of various genetic diseases • understand the concept of genetic material sequencing, its applications and resulted ethical issues • learn terminology related to molecular the molecular evolution of genes, population structure and natural selection • describe new methodologies of human genetic material analysis, as well as clinical aspects originated by genetic diseases, • understand new pathways on diagnosis, prognosis and recommended therapy of genetic diseases [Gene Therapy] 			

General Skills	
Search, analysis and synthesis of data and information using the appropriate technology; autonomous working skills; collaborative working skills; provision of independent and critical thinking	
Course content - Theory outline	
1 st week	Introduction to human genetics – The role of genetics in health: Human genome and chromosome
2 nd week	Biochemical and molecular basis of a genetic disease: DNA structure. Macro and micro-disorders. Mutations. Point mutations and determination.
3 rd week	Mendelian inheritance in human: Study of the inheritance, Mendel’s laws. Basic Mendelian inheritance patterns – non-Mendelian inheritance patterns. Monogenic disorders. Disorders due to multifactor inheritance
4 th week	Blood diseases: Thalassaimias, Rhesous (anti-D, IgG).
5 th week	Cytogenetics: Chromosomes – Caryotype, Abnormalities of autosomal chromosomes. Abnormalities of sex chromosomes. Numerical abnormalities syndromes (Down, Edwards, Patau). Structural chromosomes abnormalities syndromes (Cri-du-Chat syndrome).
6 th week	Genetic syndromes detection methods
7 th week	Cancer genetics in human: Family cancer syndromes, cancer related syndromes (retinoblastoma, breast and ovarian hereditary cancer, Hereditary Colon Polyposis, Li-Fraumeni syndrome, Von Hippel-Lindau syndrome, κλ) – Cancer cytogenetics.
8 th week	Population genetics: The study of the human evolution history (taxonomy, phylogeny based on morphological and molecular data, molecular clock). Molecular level evolution (rate of sequencing evolution, transposable elements, genes and proteins evolution, horizontal gene delivery. Diversity. Population structure. Natural selection. Regulations.
9 th week	Population genetics (continued)
10 th week	Clinical genetics and genetic counseling: Genetics clinical applications. Genetic counseling – clinical case studies of genetic diseases and handling. Prenatal screening.
11 th week	Genome sequencing: Human genome analysis program (Human Genome Project)/ applications. Ethics in human genetics. Location based cloning. Genes mapping genes involved in genetic diseases.
12 th week	Gene therapy: Future therapy. Functions and applications against cancer, cystic fibrosis etc. Stem cells/Ethical Issues.
13 th week	Genetic modification: The method CRISPR-Cas9 and its function. Structural and functional analysis of Cas9 technique of the CRISPR system. Experimental and non-experimental applications to date. Ethical issues towards the human genome modification.
TEACHING and LEARNING METHODS - EVALUATION	
TEACHING METHOD	Traditional lectures using power-point software Case studies
USE OF INFORMATION AND COMMUNICATION TECHNOLOGIES	Slides show. Video view. Use of the e-class electronic platform to store presentations in digital format for easy access by students. Communication with students on issues related to the educational process through the same platform

TEACHING ORGANIZATION	Activity Lectures (total 13x2) Total course (13x2) = 26	Semester workload 90
STUDENT EVALUATION	100% from a written final exam	
RECOMMENDED BIBLIOGRAPHY (into Greek language)		
<ul style="list-style-type: none"> ● Thomson and Thomson, Ιατρική Γενετική, Εκδόσεις Πασχαλίδης 2011. ● Krebs J. Lewin's, Γονίδια X, εκδόσεις Broken Hill 2012. ● Brown T.A., Γονιδιώματα- σύγχρονες ερευνητικές προσεγγίσεις, Εκδόσεις Broken Hill 2010. 		