UNIVERSITÀ DI PAVIA

Anno Accademico 2020/2021

GENETICS AND HUMAN BIOLOGY

Enrollment year	2019/2020
Academic year	2020/2021
Regulations	DM270
Academic discipline	BIO/18 (GENETICS)
Department	DEPARTMENT OF EARTH AND ENVIRONMENTAL SCIENCES
Course	NATURAL SCIENCES AND TECHNOLOGIES
Curriculum	PERCORSO COMUNE
Year of study	2°
Period	1st semester (28/09/2020 - 23/12/2020)
ECTS	9
Lesson hours	72 lesson hours
Language	Italian
Activity type	WRITTEN AND ORAL TEST
Teacher	SEMINO ORNELLA (titolare) - 9 ECTS
Prerequisites	To deal with the content of the course, basic knowledge of chemistry and mathematics and notions of biology of the animal and plant cell are necessary.
Learning outcomes	Aim of this course is to achieve an adequate level of knowledge of: (1) the mode of transmission of hereditary characters at cellular, individual and population level. This will provide the student with the ability to evaluate the genetic variability of living organisms (in line with the 2030 Agenda for Sustainable Development, in particular with Goal 15 - Protect, restore and promote sustainable use of terrestrial ecosystems / halt biodiversity loss; (2) the structural and functional characteristics of the genetic material; (3) gene expression in prokaryotic and eukaryotic organisms. (4) molecular and phenotypic evolution; (5) human evolution based on DNA data.
Course contents	Mitosis and meiosis. Mendelian genetics. The chromosome theory of

	heredity. Analysis of pedigrees. Sex-linked inheritance. Extrachromosomal inheritance. Sex determination. Recombination and chromosome mapping. Physical mapping. Karyotype. Genomic mutations, chromosome mutations (variation in number and structure). Polyploidy. Monosomy and trisomy in humans. Somatic and germinal mosaicism. The genetic material. Structure and function of the chromosome. Replication and transcription of DNA. Translation. Genetic code. Gene mutations. Population Genetics. The Hardy-Weinberg (H-W) principle. Genetic structure of populations. Evaluation of the genetic variability, consequences of mutation, genetic drift, migration, assortative union and natural selection. Founder effect and bottleneck. Genetic markers and their use for the reconstruction of human evolution. The emergence of evolutionary theory. Genetic variation (in phenotypes, in protein structure, in nucleotide sequences) in natural populations. Molecular evolution. Molecular phylogenies. Phylogenetic trees. Phylogeography. Rates of molecular evolution. Molecular clock. Molecular evolution and phenotypic evolution. Speciation. Evolution of the human species on the basis of recent data at the DNA level. Topics object of exercise sections: - Mitosis and meiosis. - Mendel's laws. - Sex linked transmission. - Analysis of pedigrees. - Association, genetic maps. - Molecular Genetics: transcription, translation and genetic code. - Population genetics, Hardy-Weinberg equilibrium. - Reconstruction of molecular phylogenetic trees (mitochondrial DNA -mtDNA- and Male Region of Y chromosome -MSY-).
Teaching methods	This course consists of lectures integrated by seminars and by exercise sessions that will allow students to practice on topics of formal, population and molecular genetics.
Reccomended or required readings	- P.J. Russell – Benjamin Cummings - iGenetics. A Molecular Approach. - D.P. Snustad e M.J. Simmons - John Wiley & Sons Inc- Principles of Genetics.
Assessment methods	There will be a single final exam for the course (there are no intermediate exams) and it consists of two parts. The first is a written text with exercises covering formal, population and molecular genetics. Students who pass the written text will sustain an oral exam over the entire program of the course. The oral exam is usually offered a few days (2-4) after the written text.
Further information	The course has a dedicated web site on the e-learning portal of the University of Pavia, Kiro, that students can access using their login credentials. It is worth to mention that some of the topics in this course, especially those of “population genetics”, are in line with the 2030 Agenda for Sustainable Development, in particular with Goal 15 - Protect, restore and promote sustainable use of terrestrial ecosystems / halt biodiversity loss.

Sustainable development goals - Agenda 2030	$lbl_legenda_sviluppo_sostenibile