

Main course information	
Academic subject	Human Genetics
Degree course	master's degree in Biomedical Sciences
Classe di laurea	LM6
ECTS credits (CFU)	6
Compulsory attendance	yes
Teaching language	Italian
Accademic Year	2019/2020

Docente responsabile	
Name & SURNAME	Nicoletta Archidiacono
email	nicoletta.archidiacono@uniba.it
Tel.	0805442482
Tutorial time/day	every day from 2pm to 4pm

Course details	Study area	SSD code	Type of class
	genetics	BIO/I8	Lessons and laboratory

Teaching schedule	Year	Semester
	first	first

Modalità erogazione	CFU/ECTS	Lessons (hours)	CFU/ECTS lab	Lab hours	CFU/ECTS tutorial/workshop	Tutorial/workshop hours	CFU/ECTS field trip	Field trip Hours
		5,5	44	0,5	6			

Time management	Total hours	Teaching hours	Self-study hours
	150	50	100

Academic Calendar	First lesson	Final lesson
	30/09/2019	17/01/2020

Syllabus	
Course entry requirements	knowledge of genetics, biochemistry, physiology and molecular biology
Expected learning outcomes (according to Dublin Descriptors) (it is recommended that they are congruent with the learning outcomes contained in A4a, A4b, A4c tables of the SUA-CdS)	
<i>Knowledge and understanding</i>	Understanding the structure and functioning of the human genome, the origin of genetic pathology at the molecular level to understand the patterns of hereditary transmission and the normal and pathological phenotypic manifestation.
<i>Applying knowledge and understanding</i>	Application of a wide range of methodologies for genetic research
<i>Making informed judgements and choices</i>	Acquisition of autonomy in areas related to the evaluation and interpretation of experimental data and in setting the strategies for applying molecular techniques to the study of human genetics
<i>Communicating knowledge and understanding</i>	Ability to explain key concepts and methodologies in human genetics using correct terminology.
<i>Capacities to continue learning</i>	Ability to use databases and read relevant literature critically and independently.

Syllabus	
Course content	Review of key concepts in human genetics. Pedigree Inheritance of complex characters

	<p>Hardy Wienberg law  Polymorphisms  Organization and structure of the genome  Mutations  Haemoglobinopathies  X linked inheritance and chromosome X inactivation  Imprinting  Cancer genetics  Mitochondrial disorders and origin of mitochondria  Genomic disorders  Genome evolution  Laboratory: pathological and normal human karyotype</p>
Course books/Bibliography	Tom Strachan-Andrew Read: Human Molecular Genetics
Notes	
Teaching methods	Lectures with the use of PowerPoint and computer use to set up normal and pathological human karyotypes
Assessment methods (indicate at least the type written, oral, other)	Oral interview
Evaluation criteria (Explain for each expected learning outcome what a student has to know, or is able to do, and how many levels of achievement there are)	Students will be assessed on the acquisition of notions as well as the ability to reason and make connections with other disciplines such as physiology, biochemistry and molecular biology. The details of the other disciplines are not required, but students should show understanding of what aspects of these disciplines have a bearing on human genetics. The knowledge of only the notions is not evaluated beyond an average evaluation (24 - 26/30)
Further information	