Main course information	
Academic subject	Diagnostic research in Molecular Genetics
Degree course	Master's degree in Biomedical Sciences
Classe di laurea	LM6
ECTS credits (CFU)	4
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/18	Lectures

Teaching schedule	Year	Semester
	second	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
	4	32						

Time management	Total hours	Teaching hours	Self-study hours
	100	32	68

Academic Calendar	First lesson	Final lesson
	30/09/19	17/01/20

Syllabus	
Course entry requirements	Courses of the first year of the master degree
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)	
Knowledge and understanding	Understand why and how to choose a diagnostic test in the diagnosis of genetic diseases and how and why a genetic disease screening is performed.
Applying knowledge and understanding	Choose a diagnostic test in cases of diseases involving hereditary material. Understand the genetic approach and family management. Link the diagnosis to the knowledge of the pathology and of the overall picture of the single family under examination. Examples of diagnostics of some particularly significant pathologies will provide useful information for these purposes.
Making informed judgements and choices	Acquisition of autonomy in areas related to the evaluation and interpretation of experimental data necessary to provide answers to those seeking genetic counseling
Communicating knowledge and understanding	Acquisition of the correct terminology to understand how to set up a diagnosis in genetic diseases.
Capacities to continue learning	Acquisition of the ability to understand the molecular origin of genetic diseases.

Sylabus	
Course content	Review of notions of molecular human genetics Overview of the main diagnostic tecniques.

	Prenatal diagnosis, broad outlines of forensic genetics and gene therapy. Examples of genetic diseases illustrated as models to understand strategies to be used in dignostics
Course books/Bibliography	Tom Strachan-Andrew Read: Human Molecular Genetics
Notes	Journal articles: integrate the texts used for General Genetics and Molecular Biology if necessary. PowerPoint of the lessons and handouts provided by the teacher are available as support.
Teaching methods	lectures with the use of PowerPoint
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	In addition to ascertaining the acquisition of notions, the interview will asses the ability to explain why a genetic test is requested, and who requests, why a screening is prescribed, twhen the search for pathological mutations is necessary and what it is for. More than the technical details we insist on the strategies. Knowledge of only the concepts and techniques is not evaluated beyond an average evaluation (24 - 26/30).
Further information	