General Information	**MASTER DEGREE IN BIOTECHNOLOGIES**
Title of the subject	Medical Genetics
Degree Course (class)	Medical Biotechnologies and Molecular Medicine (LM-9)
ECTS credits	6
Compulsory attendance	yes
Language	Italian
Academic year	2020-21

Subject Teacher	
Name and Surname	Giuseppe Merla
email address	g.merla@operapadrepio.it
Place and time of reception	Upon request

ECTS credits details	Discipline sector (SSD)	Area
	MED/03	characterizing

Study plan schedule	Year of study plan		Semester	
	1		2	

Time management	Lessons	Laboratory	Exercises	Total
CFU	5	1		6
Total hours	125	25		150
In-class study hours	40	12		52
Out-of-class study hours	85	13		98

Syllabus	
Prerequisites / Requirements	

Expected learning outcomes (according to Dublin descriptors)	
Knowledge and understanding	The students will get the main notions of Medical and Human Genetics
Applying knowledge	At the end of the course, the students will be able to put into practice the acquired knowledges and apply the methods and tools learned during the course. In order to use these skills in their professional career.
Making informed judgments and choices	The student, actively participating to the course by interacting with other classmates, on general issues and specific topics of Medical Genetics will be stimulated to express their own judgment and ideas, ensuring autonomy of judgment on the topics discussed.
Communicating knowledge	The active participation to the lessons, supported by the Laboratory exercitation and discussion of clinical cases, will allow the student to acquire mastery of the specialized scientific terminology of the

	subject. This will allow the development of their communication skills, which are useful for the proper conduct of the oral exam.
Capacities to continue learning	The learn ability will be stimulated through the continuous involvement of the students during the lessons on the specific topics of the program and on the new discoveries, newspaper articles concerning Human and Medical Genetics that will come out.

Study Program	
Content	• Introduction to Medical Genetics: organization of the human genome • Variability of the human genome: mutations and polymorphisms • Mendelian genetics: phenotypic variability, penetrance, expressiveness, pleiotropism, phenocopies. Method of transmission and identification of disease genes • Epigenetic modifications: Inactivation of the X chromosome, imprinting diseases, chromatin alteration diseases: the Kabuki Syndrome • Non-Mendelian genetics: the genetics of diseases mitochondrial diseases, dynamic mutation diseases, expansion of repeated units, Mosaicism and Imprinting • Genetic Biobanks and Rare Genetic Diseases • Chromosomal pathology: alterations in number and structure of chromosomes and their consequences on the phenotype • Classical and molecular cytogenetics: identification techniques e diagnosis of genetic diseases • Genetic counseling and genetic testing • Prenatal Diagnosis and NIPT • The genetics of tumors: the genetic basis of cancer • Presentation and Discussion of Selected Clinical Cases
Bibliography and textbooks	Thompson &Thompson "Genetica in Medicina", Edizione Italiana B. Dallapiccola e G. Novelli. Genetica Medica essenziale. Ed. CIC Edizioni internazionali T. Strachan and A. Read. Genetica molecolare umana. Ed. Zanichelli
Notes to textbooks	
Teaching methods	Lectures
Assessment methods (oral, written, ongoing assessment)	oral
Evaluation criteria (describe criteria for each of the above expected outcomes)	Learning verification will be carried out through an oral exam. The evaluation will be expressed in 30/30. The purpose of the test will not be solely to ascertain the level of knowledge and skills acquired by the student. The student must also demonstrate that he has acquired a critical and methodological approach to the problems that medical genetics poses.
Further information	