UNIVERSITÀ DI PAVIA

Anno Accademico 2021/2022

MEDICAL GENETICS

Enrollment year	2021/2022
Academic year	2021/2022
Regulations	DM270
Academic discipline	MED/03 (MEDICAL GENETICS)
Department	DEPARTMENT OF MOLECULAR MEDICINE
Course	BIOMEDICAL LABORATORY TECHNIQUES
Curriculum	PERCORSO COMUNE
Year of study	1°
Period	(04/10/2021 - 21/01/2022)
ECTS	2
Lesson hours	16 lesson hours
Language	Italian
Activity type	WRITTEN TEST
Teacher	MINELLI ANTONELLA (titolare) - 2 ECTS
Prerequisites	Good knowledge of Biology and Genetics in their basic contents, regarding the chemical and physical nature of the genetic material and its expression. The course program provides for a constant reminder of its.
Learning outcomes	To know well the use of fundamental genetic terms and the concepts associated with them. To know the role of Medical Genetics in its application to clinical practice. To know the classification of genetic diseases, their main aspects also in relation to genetic tests. Understand how Mendelian traits are transmitted through family trees, and, based on assigned genotypes, predict their recurrence risk. Tok know the structure of the human genome and its variations it and the possible links with phenotypes.
Course contents	Introduction to the course: the vocabulary of genetics. Genes, genomes and genetic variability. Genetic diseases and their classification.

	Methods of studying the transmission of Mendelian traits: genealogical trees Examples of monogenic diseases. Inheritance models: from the analysis of family trees to the assignment of genotypes to the calculation of the recurrence risk. Architecture of the human genome. Human genetic variations. Molecular basis of diseases.
Teaching methods	Lectures
Reccomended or required readings	Clementi M. Elementi di Genetica Medica, EdiSES Editore
Assessment methods	Written/oral exam
Further information	
Sustainable development goals - Agenda 2030	$lbl_legenda_sviluppo_sostenibile