



HUMAN MOLECULAR GENETICS	
Enrollment year	2021/2022
Academic year	2021/2022
Regulations	DM270
Academic discipline	BIO/18 (GENETICS)
Department	DEPARTMENT OF BIOLOGY AND BIOTECHNOLOGY "LAZZARO SPALLANZANI"
Course	EXPERIMENTAL AND APPLIED BIOLOGY
Curriculum	Scienze biomediche molecolari
Year of study	1°
Period	1st semester (01/10/2021 - 14/01/2022)
ECTS	9
Lesson hours	72 lesson hours
Language	Italian
Activity type	ORAL TEST
Teacher	PELLEGATA NATALIA SIMONA (titolare) - 6 ECTS RANZANI GUGLIELMINA - 3 ECTS
Prerequisites	Basic knowledge of genetics and molecular biology is essential: mechanisms of cell division, segregation of mendelian traits, recombination, genetic vs. physical mapping, mendelian inheritance in pedigrees, DNA replication, transcription, translation, gene mutations, variability, Hardy–Weinberg equilibrium.
Learning outcomes	The student is expected to acquire a good knowledge of: the structure and organisation of the human genome; the genetic and molecular bases of inherited diseases and of complex diseases; the carcinogenic process associated with both inherited and sporadic tumours; the new tools to investigate human genetic variability.
Course contents	The human genome (structure, organization, function): unique and repeated sequences, genes and gene families, "non-coding DNA" and

	<p>RNAs that regulate gene expression. Large-scale projects on the human genome. The genetic variability: protein and DNA polymorphisms. DNA polymorphisms as a tool in forensic genetics and in medical genetic research. Inheritance patterns for monogenic disorders; identification of the disease-gene by linkage analysis. The cystic fibrosis and the CFTR gene. The human hemoglobins. Structure, organization and evolution of globin genes. Globin genes' mutations: the molecular basis of hemoglobinopathies and thalassemias. The cancer as a genetic disease: oncogenes and tumor-suppressor genes, genome instability. Hereditary cancer: the genetic model of retinoblastoma. Sporadic and hereditary colorectal cancers: cancer predisposition and cancer progression. The epigenetics and the regulation of gene expression; epigenetics and cancer. The DNA repair systems; inherited diseases associated with NER system defects. Trinucleotide repair disorders; the Huntington's chorea. Complex diseases: the genetic component and the "GWAS". Pharmacogenetics and pharmacogenomics: examples in oncology. Next-generation sequencing: applications.</p>
Teaching methods	The course consists of lessons where teacher-student interactions are welcomed.
Reccomended or required readings	HUMAN MOLECULAR GENETICS: Tom Strachan & Andrew Read (John Wiley & Sons Ed.)
Assessment methods	ORAL EXAM
Further information	ORAL EXAM
Sustainable development goals - Agenda 2030	\$lbl_legenda_sviluppo_sostenibile