Main course information	
Academic subject	Molecular Biology of Human Pathologies
Degree course	Master's Degree on Biosanitary Sciences
Classe di laurea	LM-6
ECTS credits (CFU)	8
Compulsory attendance	Yes
Teaching language	Italian
Accademic Year	2019/2020

Docente responsabile	
Name & SURNAME	Guglielmina Alessandra CHIMIENTI
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Tutorial time/day	12-13 am, tuesday

Course details	Study area	SSD code	Type of class
	Molecular biology	Bio-11	Lecture/workshop

Teaching schedule	Year	Semester
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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
	8	60	0,5	6				

Time management	Total hours	Teaching hours	Self-study hours
	200	66	134

Academic Calendar	First lesson	Final lesson
	October 2019	January 2020

Syllabus	
Course entry requirements	Knowledge in the BDS SSD sectors obtained during the three-year degree in class L-13 or similar
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	Knowing the information contained in the DNA and its ways of use and understanding how alterations of biomolecular mechanisms may be associated with the onset or risk of developing a disease.
Applying knowledge and understanding	Knowing the genome and how the information it contains are used in the eukaryotes, starting from the simplest ones as the theoretical and experimental models to get to understand and integrate omics sciences such as genomics, transcriptomics, epigenomics, for the comprehension of more complex organisms, with particular attention to humans, in the physiological and pathological conditions.
Making informed judgements and choices	Understanding biomolecular mechanisms, in order to correctly and autonomously perform and analyze data obtained through molecular diagnostics.
Communicating knowledge and understanding	Acquisition of the vocabulary and terminology related to molecular diagnostics in order to understand any further information through specific bibliography.
Capacities to continue learning	Ability to assess the role of genetic information in the onset or risk of developing a disease.

Sylabus

Course content	Omic sciences: genomics, transcriptomics, proteomics, metabolomics. **Genomics**. Genome size and number of genes, paradox of C value. Eukaryotic genomes: single copy sequences, medium and highly repeated sequences. Base composition, CpG islands, isocore. Characteristics of the eukaryotic gene. High yield study methods (high throuput) for the study of allelic variants. **NGS and metagenomics**. Sequencing platforms: 454 and pyrosequencing, Illumina, Solid, Ion torrent. Thirth generation platforms: SMRT (single molecule real-time sequencing), Oxford nnopore. Metagenomics: the intestinal microbiota. **Pharmacogenomics.** From pharmacogenetics to pharmacogenomics. Pharmacokinetics (what an organism does to a drug) and pharmacodynamics (what a drug does to an organism). Genes that influence drug response: enzymes of xenobiotic metabolism and polymorphisms. Personalization of care, an example: neuroleptic malignant syndrome and dopamine D2 receptor. **Epigenetics.** Epigenetic mechanisms: histone modification and DNA methylation. The sixth base of the genome: 5-hydroxymethyl cytosine. Histonic code, bromo- and chromo-domains. Chromatin structure and gene silencing. Imprinting and associated pathologies: Prader Willi syndrome, Angelman syndrome, Rett syndrome. X-chromosome inactivation. DNA methylation and aging and human diseases. Epigenetics and environment. Study methods of epigenetic phenomena: study of DNA methylation status, MLPA and variants, bisulfite method and variants. **Transcriptomics.** Transcriptional regulation of gene expression in eukaryotes: characteristics of eukaryotic promoters. Sensitivity to DNase I, LCR, related pathologies. Chromatin structure, chromatin remodeling complexes, protein factors and transcription. Chromosomal territories and transcription factories, nuclear matrix. **Techniques for the study of gene expression.** S1 mapping, primer extension, RNase protection. Reporter genes. Gene array technology: microarray, how to do it, how to use it, what it is used for. Examples of the study of differentiated expression, expression chip in the study of human diseases. SNP chip. **q-PCR.** Study of the kinetics of the PCR reaction, reaction efficiency. Chemicals for real time: sybr green, linear probes and probes with secondary structure, q-RT-PCR: absolute (generation of the standard curve) and relative quantization of transcription (deltaCt and delta-delta Ct method). **Post-transcriptional gene regulation**. mRNA maturation, export, stability and degradation. Regulatory RNAs: miRNA, siRNA, lncRNAs. Applications of RNA interference. miRNA in cancer: role as oncogenes and tumor suppressors, interactions with p53, sponge RNA. CRISPR-Cas9 technology. **Techniques for the study of DNA-protein interactions.** EMSA, DNA footprinting, immunoprecipitation. Chromatin immunoprecipitation assay. Fusion proteins: expression synthesis, purification and applications: pull-down and double hybrid assays. FRET. **Mitochondria.** origin and similarity with bacteria. Mitochondrial activity. Size, number, location, duplication, fusion and fission. Relevance of mitochondrial dysfunction to Parkinson's disease. mtDNA in yeast and plants, editing and splicing. mtDNA in metazoans, structure, organization, gene content. human mtDNA. Muller ratchet principle and strategies to slow it down. Replication, transcription, peculiarity of the genetic code. **Mitochondria, population genetics and evolution.** Maternal inheritance. Genetic variability of human mtDNA, haplotypes and haplogroups. Mitochondrial inheritance and mitochondrial pathologies: Maternal transmission of mtDNA; homoplasmia, heteroplasmy, threshold effect. Types of mtDNA defects, point mutations, rearrangements, common deletion, depletion. Characteristics of mitochondrial diseases, study methods, mitochondrial disorders. **Animal viruses.** general characteristics, envelope, infection mechanisms, effects of infection, classification. DNA virus: SV40, hepadnavirus; RNA + virus: poliovirus, RNA-virus: flu. **Workshops.** Expression of the Green Fluorescent Protein in transformed bacteria: a reporter gene expressed in a regulated manner. Chromatographic purification of the recombinant protein.

Course books/Bibliography	BIOLOGIA MOLECOLARE, Amaldi, Benedetti, Pesole, Plevani, CEA FONDAMENTI DI BIOLOGIA MOLECOLARE, Allison, Zanichelli MOLECOLARE del GENE, Watson, Zanichelli Il GENE, Lewin, Zanichelli
Notes	Power point lessons are available as support.
Teaching methods	Frontal lessons with use of audiovisual supports.
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 the acquisition of individual and necessary notions, the ability to integrate these notions in order to obtain a complete view of the genetic information and its use in a system as complex as man is evaluated. Through a written report produced by the student, the student's ability to grasp practical exercises as a real "experiment" to be included in his own cultural background is assessed in order to deepen his knowledge of the scientific method of investigation.
Further information	