UNIVERSITÀ DI PAVIA

Anno Accademico 2019/2020

MEDICAL GENETICS

Enrollment year	2019/2020
Academic year	2019/2020
Regulations	DM270
Academic discipline	MED/03 (MEDICAL GENETICS)
Department	DEPARTMENT OF MOLECULAR MEDICINE
Course	MEDICAL AND PHARMACEUTICAL TECHNOLOGIES
Curriculum	PERCORSO COMUNE
Year of study	1°
Period	1st semester (14/10/2019 - 17/01/2020)
ECTS	5
Lesson hours	40 lesson hours
Language	Italian
Activity type	WRITTEN TEST
Teacher	CICCONE ROBERTO - 2 ECTS ROSSI ELENA - 3 ECTS
Prerequisites	No
Learning outcomes	The aim of the course is to provide theoretical and conceptual bases of human and medical genetics as well as the related diagnostic-clinical aspects. The student must achieve the theoretical knowledge and laboratory practice that underlies chromosomal, monogenic and polygenic diseases; he must learn the methodologies of molecular genetics and cytogenetics aimed at the diagnosis of genetic diseases, and the instrumental technologies which allow the molecular analysis and the study of genes and genome.
Course contents	Mutations: classification criteria for mutations. Genomic mutations and chromosomal mutations: how a chromosomal preparation is obtained after villocentesis, amniocentesis or venous blood sampling.

	Non-invasive prenatal diagnosis. NIPT Epidemiology of chromosomal abnormalities. Down syndrome: which studies to identify candidate genes and genes responsible for specific phenotypic characteristics; trisomy 21 from translocation; microsatellites to determine parental origin; microsatellites and minisatellites and their use. Patau syndrome, Edwards syndrome. Mice model mice: transgenic mice and knock-out mice Numerical abnormalities of sex chromosomes. X-chromosome inactivation and preferential inactivation in specific structural rearrangements of the X chromosome. Structure chromosomal abnormalities: translocations. Phenotype in apparently balanced translocations. Complex translocations with examples. Structure chromosomal abnormalities: deletions and duplications. Microdeletion syndromes: Velocardiofacial syndrome, Williams syndrome and Prader-Willi syndrome (hints). Why genomic disorders. Indications for chromosome/genetic analysis in the neonatal period, during childhood, puberty and reproductive age. Disorders of sexual development. Male and female infertility. Gene mutations, pathogenetic mechanisms in dominant mutations: Alagille syndrome, Marfan syndrome, Osteogenesis imperfecta and Achondroplasia. Examples of canalopathies: Long QT syndrome and cystic fibrosis. Gene therapy in cystic fibrosis. Diagnostic approaches in genomic, chromosomal and gene mutations MicroRNAs in physiological and pathological processes
Teaching methods	Frontal lessons
Reccomended or required readings	- Genetica umana e medica - Giovanni Neri, Maurizio Genuardi, Seconda Edizione - Masson - 2010 - Genetica & Genomica - Tom Strachan Judit Goodship Patrick Chinnery - Zanichelli - 2015
Assessment methods	Written test
Further information	Reception: by appointment, write to roberto.ciccone@unipv.it and / or to rossie@unipv.it
Sustainable development goals - Agenda 2030	$lbl_legenda_sviluppo_sostenibile