

Anno Accademico 2019/2020

| MEDICAL GENETICS | | |
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| 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. | |

| atudies to identify condidate games and games represential for energies | |
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| phenotypic characteristics; trisomy 21 from translocation; microsatellite to determine parental origin; microsatellites and minisatellites and thei use. Patau syndrome. Edwards syndrome | es r |
| 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 (bints). 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 | |
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| Assessment methods Written test | |
| Further information Reception: by appointment, write to roberto.ciccone@unipv.it and / or rossie@unipv.it | to |
| Sustainable development | |
| goals - Agenda 2030 \$Ibl legenda sviluppo sostenibile | |