COURSE (TITLE): MEDICAL GENETICS

LECTURER:

YEAR and SEMESTER: II, I

CREDITS (CFU): 6

SECTOR (SDS): MED03

ACADEMIC YEAR:

ASSESSMENT:

LOCATION: Department of Environmental, Biological and Pharmaceutical Science and Technologies, Via Vivaldi 43 Caserta

COURSE OBJECTIVES/OUTCOMES:

At the end of the course, the student will have acquired a basic preparation that will allow him to understand the principles that govern the transmission of Mendelian diseases, the technology and interpretation of genetic tests and their use in prenatal and postnatal counseling. It will also have the appropriate knowledge to recognize the main genetic diseases and complex syndromes and the basic principle of medical ontology.

SYLLABUS (overview)

Basic preparation to understand the complex laboratory approach to manage different categories of medical conditions with genetic causes

SYLLABUS (Detailed description):

1. Genetic tests.

The analysis of the karyotype and the bandages, FISH. arrayCGH. PCR, sequencing, real-time PCR, MLPA. Linkage analysis, microsatellites, CNV and SNPs. Next generation sequencing (NGS), WGS, exome, targeted NGS. Noninvasive Prenatal Testing (NIPT)

2. Chromosomal pathology

Genetic counseling: reproductive risk dependent and independent of the partner. Aneuploidy in abortions and risk of recurrence. Triploidy from a paternal or maternal double, tetraploidy. Autosomal (21, 18, 13, 16) and sex chromosomes trisomies (Kilnefelter). Mosaicism. Monosomies, Turner syndrome

3. Sub-chromosomal pathology

Chromosomal deletions, paracentric and pericentric inversions. Unbalanced and balanced translocations, Robertsonian, chromosomal markers. Sub-microscopic deletions and duplications (Williams, diGeorge, Cri du Chat, Smith Magenis, Wolf Hirschhorn)

4. Mendelian monoallelic disorders with *de novo* mutations

(ex: craniosynostosis, achondroplasia, rare syndromes, etc.).

5. Mendelian diseases with autosomal dominant transmission

(eg neurofibromatosis, S. Marfan, polycystic kidney, osteogenesis imperfecta).

6. Mendelian diseases related to the X chromosome

(muscular dystrophy of Duchenne and Becker, hemophilia, Rett syndrome).

7. Mendelian diseases with autosomal recessive transmission

(cystic fibrosis, alpha and beta thalassemia, spinal muscular atrophy, hemochromatosis,

glycogen storage disorders)
7. Dynamic mutations
Dynamic mutations in non-coding regions (X-fragile, myotonic dystrophy). Dynamic mutations in coding regions (Huntington disease, spino-cerebellar ataxias)
9. Mutations in chromosomal regions with imprinting
(Prader-Willi, Beckwith Wiedemann, Angelman, Silver-Russel).
10. Mitochondrial DNA mutations
(MERFF, MELAS, LHON, KS, S. Leigh)
11. Multi-factorial characters and GWAs studies

TEXTBOOKS:
Thompson and Thompson

ADDITIONAL READING:
Strachan Human Molecular Genetics