

**SUBJECT: GENETICS DIAGNOSIS**

Coordinator	Virginia Nunes
Teachers	Virginia Nunes, Manel Esteller, Josep Oriola, Ignacio Blanco, Victor Moreno, Manel Gene, Anna Soler (invited prof.), Nuria Sala (invited prof.), Marga Nadal (invited prof.) y Concepció Lázaro (invited prof.)

**JUSTIFICATION:**

The last advances in the isolation and characterization of genes have promoted enormously the possibility of the genetic diagnosis for many diseases and for very diverse situations. There is nowadays a great quantity of technologies that can be in use for detecting routine mutations in genes of interest, as well as alternative technologies of indirect diagnosis. A master in Biomedicine has to contemplate, obviously, a section dedicated to the genetic diagnosis and to the prevention of hereditary diseases. In this subject we try to explain the importance and possibility of realizing a genetic diagnosis, in the context of the knowledge and technology currently applicable to the Human Genetics

**OBJECTIVES:**

The principal aim of this subject is to provide the knowledge necessary for the comprehension of the molecular bases of the genetic diagnosis and for the correct accomplishment of the type of diagnosis most suitable to every particular situation. Likewise, it also includes a detailed explanation of the diverse experimental technologies that can be in use in the different types of diagnosis and use someone of these technologies in laboratory practices.

**CONTENTS AND AGENDA:****Contents:**

Genetics Diagnosis concept. Direct and indirect diagnosis. Genetic Diagnosis applied to monogenic and complex disease. Genetic variability among individuals. Technologies used in different types of diagnosis.

**Agenda:**

**Topic 1.- General consideration on the course:** Hereditary diseases. Genetics variation among individuals: pathogenic mutation, polymorphism. Types of mutations in the human diseases

**Topic 2.- Chromosomal analysis:** Human chromosomes. Chromosomal anomalies. Technique used for the diagnosis of chromosomal alterations

**Topic 3.- Study of monogenic diseases direct and indirect methods:** Polymorphic markers. Linkage analysis. Co-segregation analysis. Examples

**Topic 4.- Methods for mutation detection:** DNA based test methods: Sequence analysis, Mutation scanning: CSCE; DHPLC, PCR-SSCP, PCR-DGGE; PTT. Targeted mutation analysis:

Melting curve analysis, OLA,MLPA.....

**Topic 5.- New cytogenetics strategies:** FISH, Fiber FISH, TMA and FISH, CGH, CGH microarray, M-FISH, FISH M-tel, Nuclear architecture: FISH-2D, FISH-3D

**Topic 6.- Population screening and risk calculations:** Genotype relative risk. High penetrance genes, low penetrance genes. Bayes's theorem. Multiple risk factors. Predictive models

**Topic 7.- Genetic Diagnosis of Hereditary Multiple endocrine neoplasias:** Multiple endocrine neoplasia Type 1 (MEN1). Multiple endocrine neoplasia Type 2 (MEN2). Von Hippel-Lyndau Syndrome (VHL). Familial Parganglioma Syndrome

**Topic 8.- Hereditary cancer. From bench to the bed side:** Cancer as a health problem: incidence, mortality and morbidity, prevention, etiology. Susceptibility. Biological bases of cancer susceptibility and predisposition. Breast cancer. Genetic predisposition testing. Genetic Counselling.

**Topic 9.- Forensic Genetics:** Concepts used in Forensic Genetics. Methods used for Forensic Genetics Studies. Paternity testing. Genetic markers. Likelihood ratio.

**Topic 10.-Genetic analysis of complex diseases:** Main concepts, Measurements of genetic influence. GWA. Haplotypes, TagSNPs, HapMap project. Selection of study types. Exemples

#### **METHODOLOGY AND SUBJECT ORGANIZATION:**

The subject has 2,5 ECTS, that are equivalent to a global work on the part of the pupil of approximately 65-75 h.

#### **Education presencial:**

**Theoretical classes:** A total of 20 hours among theory and seminars, of approximately of two hours each one, given by teachers and invited teachers in the topic. During these classes and seminars also there will treat each other practical cases of different diseases.

**Practical Classes:** Ten hours of practices. The student will have the possibility to do experiments in the PBEC laboratories. They will perform sequencing, DNA metilation analysis, Rett syndrome analysis. They will also be familiarised with genomic platforms as a tool to be used in genetic diagnosis.

#### **Non presencial work:**

##### **Task to developing:**

To search and to read bibliography adapted to assimilate the concepts given in the theoretical classes

To solve problems related with diagnostic cases, for different diseases

##### **Pupil's own study:**

Every hour of class presencial will need an additional task on the part of the pupil of between 1,5 and 2 hours

## **EVALUATION**

### **Criteria and evaluation procedure:**

The pupil will prepare written work on a genetic disease explaining the genetic characteristics of it and which are the genetic techniques used for its diagnosis. The pupil will also write an example of family with this disease, he will draw the pedigree of the family and he will put a case of genetic counselling for the disease

The pupil will present a work related with the practical part of the course.

## **BIBLIOGRAPHY**

Strachan T and Read A P. Human Molecular Genetics . Taylor & Francis ed. (2009). ISBN: 0815341490-97

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