GENETICA UMANA (MG000013)

1. language

Italian.

2. course contents

Coordinator: Prof.ss MARCELLA ZOLLINO

Year Course: 2

Semester: I

UFC: 4

Modules and lecturers

- Genetica Umana MG0018 (MED/03) 3 CFU:

	Lezioni (ore)
Prof. Marcella Zollino (I Canale)	28
Prof. Eugenio Sangiorgi (I Canale)	9
Prof. Francesco Danilo Tiziano (II Canale)	28
Prof. Elisabetta Tabolacci (II Canale)	9

- Genetica Medica Attivita' Professionalizzante MG000011 (MED/03) 1 CFU:

	Tirocinio (ore)
Prof. Marcella Zollino (I Canale)	25
Prof. Eugenio Sangiorgi (I Canale)	25
Prof. Francesco Danilo Tiziano (II Canale)	25
Prof. Elisabetta Tabolacci (II Canale)	25

3. bibliography

Genetica Umana e Medica. Neri, Genuardi. Quinta Edizione. EDRA S.p.A.

Alternatively, other free choise Medical Genetics books, in which the topics of the course are covered in an exhaustive manner.

4. learning objectives

The course is structured to provide the student with the essential elements to understand the genetic basis of human diseases, by connecting genomic data with clinical signs. In particular, it aims to clarify the models of inheritance and pathogenesis, whether monofactorial or multifactorial; to indicate the appropriateness of the genetic tests, the methods of their correct interpretation and the significance of the genetic results both for the assessment of the recurrence risk in families (and the appropriate method for familial screening), and for targeted therapeutic approaches. The course aims to provide the student with the most modern scientific knowledge regarding techniques for the genetic diagnosis, the study of disease mechanisms, and genetic engineering

methods for research and therapy.

Knowledge will be disseminated through lectures accompanied by clinical examples.

The ability to understand will be assessed during the lectures with questions posed to students and open discussion of the answers.

The autonomy of judgment and communication skills will be evaluated through the proposal of family pedigrees and the presentation of exemplary clinical situations; students will be asked to discuss about them. During the lectures, the interaction will take place with a limited number of students, chosen randomly.

During ongoing tests, each individual student will be assessed on the ability to understand, the ability to apply the concepts in clinical practice, the autonomy of judgment and the ability to identify criticisms.

5. PREREQUISITES

Basic school training and knowledge of basic scientific elements in the fields of genetics, chemistry and mathematics are required. It is advisable to have taken the Biology examination, that is scheduled in the first year of the Degree in Medicine and Surgery, due to the connection of the covered topics.

6. teaching methods

Teaching methods include both lectures explaining the scientific bases, and classroom simulations of real situations in clinical practice. Students are asked about the framing of the problem and the possible approaches for its solution. During these interactive situations, ability to communicate with the patient and to interact with colleagues will be assessed; whether the topics transmitted in the lectures have been assimilated, creating critical awareness and independent judgment will be checked as well The teacher-student interaction will also be used by the teacher to understand the gaps in the student's ability to understand and the methods that the teacher can adopt to fill them.

Exercises on the topics covered during the lectures will be provided in itinere.

7. other informations

The teachers will be available to receive students, on the days and in the spaces that will be indicated during the course.

8. methods for verifying learning and for evaluation

The final examination will be the tool for voting, expressed out of thirty.

It will consist of a written test of 31 multiple choice questions, followed by an oral test. You can access the oral test if you have passed the written test with a minimum of 18. There is no penalty for incorrect answers.

The oral test can modify the result of the written test up to a maximum of 6 points, which can be used both in a positive and in a negative sense. The questions will be different for the two channels, but will focus on the same topics, including those dealt with in the lectures and during the professionalizing internship.

In particular, the questions of the written test will be formulated in order to verify learning and ability

to apply theoretical knowledge to clinical situations. The questions will be in part based on the description of a real clinical problem, or of a specific pedigree; the student's autonomy of judgment can be deduced from the type of response. The oral examination will have the specific purpose of verifying the student's communication skills, his ability of establishing links between the topics covered in different modules of the program and his ability of highlighting limits and criticisms.

The maximum final evaluation of 30 with honors can be achieved if the student has achieved a minimum mark of 25 in the written test. This possibility exists if the topics of the program are studied extensively on textbooks, as the lectures are not exhaustive of the topic on which they are articulated.

9. program

Module 1

- *Organization of the human genome*
- *Concept of: allele; locus; heterozygosity; compound heterozygosity; homozygosity; penetrance; expressiveness; AR, AD and X-linked mendelian inheritance*
- *Genomic imprinting*
- *Deseases caused by imprinting disturbance: molecular mechanisms and genetic tests.*
- *Prader-Willi syndrome and Angelman syndrome. Beckwith-Wiedemann syndrome and Silver Russell syndrome*
- *Mitochondrial inheritance*
- *Multifactorial diseases*

Module 2

- *Intragenic variants*
- *Dynamic mutations. Fragile X syndrome, Steinert myotonic dystrophy; Huntington's disease*
- *Human chromosomes. Cytogenetic analysis. Balanced and unbalanced chromosomal rearrangements.*
- *Molecular cytogenetics analysis: array-CGH/SNP array; FISH; MLPA; Real time PCR.*
- *Chromosomal syndromes. Trisomy 21, trisomy 13, trisomy 18. Turner syndrome. Klinefelter syndrome. Mosaic trisomy 8. Pallister-Killian syndrome.*
- *Array-CGH/SNP array: classification of copy number variants (CNV); causes of pathogenicity of benign variants*
- *Haploinsufficiency syndromes: molecular mechanisms*
- *Genomic disorders*
- *Dominat negative and gain-of-function effects*
- *Chromatinophaties*
- *Chanelopathies*

Module 3

- *Population genetics: Hardy-Weinberg equilibrium*
- *HW equilibrium and modifying factors*
- *Linkage and linkage disequilibrium*
- *Genetic basis of cancer*
- *Hereditary tumors: molecular basis; example of retinoblastoma and breast-ovarian cancer.*
- *Constitutional conditions predisposing to cancer: molecular bases*
- *Chromosome fragility diseases and overgrowth syndromes*

Module 4

- *Genetic and phenotypic heterogeneity.*
- *Hemoglobinopathies*
- *Haploinsufficiency and dominant-negative effect*
- *Classical and new generation sequencing (NGS) techniques. Classification and interpretation of genomic variants*
- *cDNA, transcriptome; methyloma*
- *Recombinant DNA*
- *Application of genetic engineering in medicine.*
- *Introduction to the therapy of genetic diseases*
- *Gene therapy news*