

Introduction to human and medical genetics

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Part 1 – Basic concepts of human genetics

1. Introduction to human genetics
2. Mendelian genetics
3. Penetrance and expression
4. Chromosomal number abnormalities
5. Chromosomal structural abnormalities
6. Basic concepts of mitochondrial genetics
7. Basic concepts of dynamic mutations
8. Basic concepts of multifactorial disorders
9. De novo variants and mosaicism

This section comprises 12 lectures lasting between 60 and 90 minutes each

Objective:

To learn the basis of human and medical genetics; to gain knowledge on mendelian, chromosomal, mitochondrial and multifactorial disorders, with practical examples from real cases.

Part 2 – Basic concepts of medical genetics

1. Indications to genetic testing
2. Choice of genetic testing
3. Interpretation of genetic testing
4. Testing asymptomatic subjects
5. Prenatal and preimplantation diagnosis
6. Variant classification and interpretation
7. Overview of major genetic databases
8. The non-coding DNA (with Dr. Elisa Giorgio)
9. Therapies in genetics (with Dr. Edoardo Errichiello)
10. Reproduction genetics (with Dr. Fabio Sirchia)

This section comprises 12 lectures lasting between 60 and 90 minutes each

Objectives:

1) to recognize a genetically determined disorder; 2) to correctly evaluate the model of inheritance; 3) to know the principal indications to genetic testing, be able to select the most appropriate genetic tests and to correctly interpret positive, negative and ambiguous results; 4) to know the basics of genetic counseling; 5) to be able to manage the reproductive problems of a family, including prenatal diagnosis.