

## SYLLABUS DEL CORSO

### Genetics

2223-6-H4102D039-H4102D173M

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#### Aims

- Acquire knowledge regarding the major diagnostic genetic techniques.
- Gain knowledge about the role of prenatal genetic counselling and prenatal screening.
- Acquire skills in recognizing the most common facial dysmorphism and most common genetic pediatric conditions.
- Gain knowledge about the role of genetics in the diagnosis and treatment of pediatric cancers.

#### Contents

The course will provide the students with basic concepts of the main genetic investigation techniques used both in the context of prenatal diagnosis and to investigate a suspected genetic condition during childhood. The students will gain knowledge regarding the most frequent genetic condition that can affect the fetus and the child. The course will address the importance of prenatal genetic counselling, the role of dysmorphism in the diagnostic approach of genetic disorders, the importance of the multidisciplinary approach in the follow-up of genetic syndromes. Finally, the course will provide knowledge on the genetics of pediatric tumors.

#### Detailed program

- Genetic diagnostic techniques: Karyotype, FISH, CGH-array, the role of Next Generation Sequencing
- Prenatal diagnosis: the importance of genetic counselling; the role of prenatal screening tests. Hints on pre-implantation diagnosis.
- Dysmorphism in newborns and children; the clinical presentation and the diagnostic approach to the most common genetic syndromes
- The role of a multidisciplinary approach in the management of genetic disorders

- The role of genetic and environmental factors in pediatric cancers; focus on clinical features of genetic cancer syndromes

## **Prerequisites**

Fundamentals of molecular genetics and biology

## **Teaching form**

Frontal lectures  
Problem based learning  
Simulation activities; experience in the laboratory

## **Textbook and teaching resource**

## **Semester**

## **Assessment method**

## **Office hours**

## **Sustainable Development Goals**

GOOD HEALTH AND WELL-BEING

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