
COURSE (TITLE): Innovative methods and models for the study of genetic diseases

LECTURER:

YEAR and SEMESTER: First / Second

CREDITS (CFU): 3

SECTOR (SDS): BIO/13

ACADEMIC YEAR:

ASSESSMENT: Oral Examination

LOCATION: Institute of Genetics and Biophysics (IGB), National Research Council of Italy (CNR),

Via Pietro Castellino 111, 80131 Naples

COURSE OBJECTIVES/OUTCOMES:

This programme will allow the acquisition of methodological and technological knowledge and the use of cell and animal models for the study of genetic diseases.

SYLLABUS (overview)

- Human diseases. Disease-Gene identification in the pre-genomic era. The Human Genome Project. Next-generation sequencing.
- The adaptive immune response. Regulation of transcription in eukaryotic cells
- Molecular basis of neoplastic transformation. The hallmarks of cancer. Genetic and epigenetic mechanisms implicated in human tumorigenesis.

SYLLABUS (Detailed description):

Patterns of inheritance of genetic diseases: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y-linked. Identification of disease genes in the pre-genomic era: genetic markers, linkage analysis, physical maps, candidate genes, mutation analysis, types of mutations. The human genome project, the single nucleotide polymorphisms (SNPs), the 1000 genomes project. Next generation sequencing technologies, exome sequencing vs whole exome sequencing, clinical sequencing and ethical implications. Application examples of NGS approaches: identification of genes responsible for rare skeletal diseases.

Genetics of the adaptive immune response. The HLA and antigen presentation. Generation of clonal diversity in the immune system: the TCR and BCR. Current approaches in vaccine design. Mouse models.

Regulation of transcription in eukaryotic cells. Promoters and enhancers, CpG islands. DNA methylation and histone modifications, chromosomal architecture. X-chromosome inactivation. Chromosomal territories. The 3D genome.

Hallmarks of Cancer.

Oncogenes, tumor suppressors and signal transduction.

Dysregulation of cell cycle control and apoptosis in neoplastic cells.

Epigenetic mechanisms mediated by chromatin modifications and non-coding RNAs.

Animal models of human cancer.

Cancer Stem Cells and tumor microenvironment.

Invasion and metastasis mechanisms.

Impact of genomic and transcriptomic analyses on tumors classification and prognosis, along with novel personalized therapies.

TEXTBOOKS:

Notes from lectures

ADDITIONAL READING:

Suggested during the course.
