UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Cytogenetics and Cytogenomics in Human Genetic Disorders

2627-1-F0602Q126

Aims

The course aims to provide students with specific knowledge and skills in the field of human pathologies caused by genome architecture.
1.Knowledge and understanding: the student will be able to know the mechanisms underlying the formation of the main chromosomal anomalies, to understand the principles of genotype-phenotype correlation and the problems to be faced in genetic counseling. The student will also be introduced to basic concepts of genome evolution. 2. Applying knowledge and understanding: The student will be able to recognize the main chromosomal anomalies, discuss their phenotypic implications and make hypotheses on possible pathogenetic mechanisms. 3. Making judgements: The student will be able to Discuss clinical cases with pathological karyotypes; read and discuss original works critically. 4. Communication skills: the student will be able to know how to use the language of the discipline to discuss critically the topics covered. 5. Learning skills: the student will have the necessary skills to deal independently with further specialized studies in medical genetics.

Contents

To provide knowledge of the genome architecture and evolution, as well as chromatin organization and its dynamics; the chromosome structure, functions, changes during the cell cycle and under the influence of environmental factors. To provide understand of the significance of changes in chromosomal structure for the development and diseases of the individual.

Detailed program

Part 1: introduction to cytogenetic techniques. 1. the architecture of chromatin; behavior of chromosomes in mitosis and meiosis. 2. the study of the karyotype: from the collection of the sample to the banding; definition of the normal

human karyotype and ISCN nomenclature. 3. Main standard techniques of chromosome differential staining. 4. Molecular cytogenetic techniques: FISH, CGH, arrayCGH, SNP array, Chromosome conformation capture techniques. Part 2: chromosomal pathologies 1. number abnormalities: formation mechanisms and associated phenotypes (triploidies, tetraploids, mixoploidies). 2. chromosomal aneuploidies: formation mechanisms and associated phenotypes. 3. fragile sites and chromosomal instability diseases. 4. .UPD and genomic imprinting. 5. Structure anomalies: formation mechanisms and associated phenotypes. 6. Genomic Disorders: NAHR: Non-Allelic Homologous Recombination. Part 3: applications of the karyotype study in medical diagnostics and research. 1. the indications to study the karyotype. 2. chromosomal abnormalities in the fetus, spontaneous abortions, infertility and infertility. 3. cytogenetics in tumors. 4. 3D chromatin structure. Methods of analysis of 3D chromatin. Hi-C method. Topologically associating domains (TAD); Hierarchical structure of chromatin. Part 4: Genome evolution. 1. Basic mechanisms and comparative genomics. 2. Conservation indices and their application. 3. Transposable elements in the human genome: origin, biomedical relevance, domestication and contribution to genomic plasticity. 4. Restrictors of transposable elements: mechanisms and evolution.

Prerequisites

Genetics, Molecular Biology

Teaching form

18 lessons of 2 hours in the erogative mode in presence: exposition of basic concepts.

3 lessons of 2 hours conducted in interactive mode in presence: student involvement and discussion of clinical cases

Textbook and teaching resource

Scientific articles and reviews provided by the teacher during lessons. The slides of classoroom lessons will be available on the elearning Platform.
Text:
-Human and Medical Genetics - 4th edition or later; Authors: Neri and Genuardi - Edra Publishing
.Genetica e genomica nelle scienze mediche-2 edizione; Strachan-edizioni Zanichelli

Semester

Second semester

Assessment method

There are no in itinere tests.

Oral exam: Interview on the topics of the lesson

Office hours

On appointment; mail to: angela.bentivegna@unimib.it; manuela.sironi@unimib.it

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION | GENDER EQUALITY