UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Cytogenetics

2324-1-F0601Q073

Aims

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. 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

Even today the study of the karyotype is the most requested among the genetic laboratory investigations. The aims of the course is to to approach the study of human chromosomes, starting from their structure and morphology up to the pathogenetic mechanisms underlying chromosomal anomalies. The new frontiers of cytogenomics and the new investigation techniques on the spatial organization of chromatin and its role in gene expression will also be discussed.

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.

Prerequisites

Genetics, Molecular Biology

Teaching form

Frontal lessons

Textbook and teaching resource

Scientific articles provided by the teacher during lessons. The slides of classoroom lessons will be available on the elearning Platform.

Semester

Second semester

Assessment method

oral exam.
Interview on topics covered in class and on in-depth topics not covered in class but belonging to the program

Office hours

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

Sustainable Development Goals

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