UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Diagnostic Technologies For The Biomedical Laboratory

2122-3-I0302D035

Aims

MOLECULAR GENETICS: models of mendelian disorders and effect of mutation on cellular function. Approaches used to study Chromosomal syndromes, microdelection syndromes and imprinting disorders. Karyotype analysis. Genetic variation: mutation and polymorphism. Tools of molecular karyotype analysis. Bioinformatics.

BIOTECHNOLOGICAL DIAGNOSTICS: Molecular diagnostics of Thrombophilia, Hereditary hemochromatosis, cardiovascular risk, and cancer. Epigenetic modifications. Pharmacogenetics and pharmacogenomics. Tools for gene expression analysis (Calibrated RT-PCR, Real Time RT-PCR, Microarrays). Molecular diagnostics in solid tumors and haematological malignancies

TECHNICAL SCIENCES: Classification and description of the main features of qualitative and quantitative analytical methods. Internal and external reference standards, calibration curves, data analysis, sources of errors. Principles of chromatography. HPLC and GC. Derivatives for chromatography. Mass spectrometry.

THERAPEUTIC DRUG MONITORING: Explain the basic principles of therapeutic drug monitoring, monitoring of toxic substances and drugs and related methods

Contents

The primary goal of the course is to provide tools for the understanding of the laboratory techniques to approach hereditary diseases. Provide knowledge of human molecular genetics and cytogenetics. Provide knowledge of

clinical molecular diagnostic, of chromatography and of spectroscopy for qualitative and quantitative analysis, including knowledge of Therapeutic drug monitoring.

Detailed program

Human Mendelian diseases: transmission and phenotypic complexity. The genes and their role in genetic diseases. Human diseases deriving from uniparental dysomy and related techniques. Chromosomic syndromes and microdeletions. Karyotype and its analysis. FISH, CGH, Fiber FISH and molecular analysis. New genetic techniques. Genomics and Proteomics.

Molecular diagnostics of cardiovascular risk and thrombophilia (FV, FII, MTHFR, CBS, APO-E, gene mutations), hemochromatosis molecular diagnosis (HFE, TFR2, FPN1, etc), molecular diagnosis of cancer. Genetic and phenotypic screening.

Pharmacogenetics e pharmacogenomics.

Genetic expression studies (calibrated RT-PCR, Real Time RT-PCR, Microarrays).

Classification of analytical methods. Qualitative and quantitative analysis and their terminology and methods.

Sample preparation: extraction and purification by chromatographic techniques. Chromatography and its principles: efficiency of a chromatographic column, , resolution, selectivity)

Reference products: internal and external calibrators. Gas-chromatography/HPLC: derivatization reactions. Instrumental characteristics and components: of GC and HPLC: injectors, columns, detectors. Mass spectrometry: instruments, principles, components: ionization processes (EI-CI), analyser (magnetic, quadrupolar, ion trap), detectors (EM-PM). EI spectra interpretation. Examples of practical applications.

General principles of therapeutic drug monitoring: definition and rational basis of the Therapeutic Drug Monitoring (TDM); main analytical methods of drug monitoring (Particular parts of mass spectrometry: ESI and APCI ionization processes; Orbitrap and linear trap mass analysers; tandem mass spectrometry and its different modality of use). Rationalization of the TDM of the most frequently monitored drugs, toxins and drugs.

Prerequisites

Having passed the compulsory courses of the degree

Teaching form

Lectures and exercises

Textbook and teaching resource

The Teachers will provide educational materials

Semester

First semester

Assessment method

Written test: multiple choice test and open questions

Oral Test: discussion of written test

Office hours