



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Tecnologie Diagnostiche di Laboratorio Biomedico

2021-3-I0302D035

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#### Aims

**MOLECULAR GENETICS:** models of mendelian disorders and effect of mutation on cellular function. Approaches used to study Chromosomal syndromes, microdeletion 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. Chromosomal 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**

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