SYLLABUS DEL CORSO

Citogenetica

2122-1-H4101D004-H4101D014M

Aims

The course will provide the essential theoretical knowledge of biology and genetics, also focusing on the possible future application in the medical field. The subjects of the course will provide the necessary knowledge to understand the vital processes, both on the cellular and molecular level, as well as the laws of heredity and the processes involved in the generation of phenotypic diversity. The acquired knowledge will contribute to a better understanding of the processes involved in normal and pathological situations.

Contents

Structure and function of the most important cellular macromolecules; DNA duplication and repair mechanisms; transcription and RNA processing; translation and protein sorting; molecular and cellular mechanisms responsible for gene expression and its regulation, analyzing epigenetic mechanisms, transcriptional and post-transcriptional regulation; signal transduction pathways; molecular and cellular mechanisms which control the cell cycle, cellular growth and differentiation as well as cell-to-cell interactions; basic concepts of heredity and the transmission patterns of inherited traits; mechanisms which can generate phenotypic variants in men; methodologies used in genetic analysis; most important biotechnological applications in medicine (gene-based and cell-based therapy).

Detailed program

GENERAL BIOLOGY

– Classification of living organisms – Structure od prokaryotic and eukaryotic cells – Viruses, classification, lytic and lisogen cycle.

CELL AND MOLECULAR BIOLOGY.

Chemical composition and molecular organization of the cell – water, carbohydrates, lipids, proteins and nucleic acids. Identification of the chemical compound carrying the genetic information – Molecular basis of inheritance – DNA replication. Telomerases – Mechanisms of DNA repair. Correlation with human diseases, aging and cancer. - RNA, structure and function – Transcription and RNA maturation – The genetic code, and its biological implication (redundancy, frameshift) – Translation – Protein sorting – Gene expression regulation in prokaryotes and eukaryotes – Molecular genetic tools (restriction enzymes, vectors, Southern blotting, PCR, sequencing, microarrays). Molecular cloning.

– Structure and function of the cytoskeleton – Cell adhesion mechanisms – Endocytosis and Exocytosis – Cell-to-cell communication in complex organisms – Signal transduction and the role of protein kinases – Cell cycle and its regulatory mechanisms. _ Mytosis and Meiosis – Apoptosis – Cell differentiation processes: embrional and adult stem cells.

GENETICS

– Human reproduction – Genetic variability – Inheritance – Genes: genotype and phenotype – Diploidy and reproduction. Honologous chromosomes, alleles and loci, homozygosity and heterozygosity – Mendel's laws – Alleles: wild-type, mutated and multiple ones, dominance and recessivity – Mendel's law's implementation: epistasis, penetrance and expressivity – Sex chromosomes. Sex determination – How to build and analyze a family tree – Chromosome X inactivation. Its implication in the phenotipic manifestations of genetic diseases – Test cross and inheritance of genes localized on different chromosomes – Crossing over and its genetic consequences – Recombination frequencies calculation and genetic maps – Principles and consequences of mytochondial inheritance and genomic imprinting – Examples of monogenic inheritance: blood groups (AB0, Rh), color blindness – Multigenic inheritance and quantitative genetics – Characters showing a treshold effect – Multifactorial disorders – Population genetics and Hardy-Weinberg equilibrium.

CYTOGENETICS

– Methods for chromosome analysis – Normal human caryotype – Chromosomic and genomic mutations and their effect on meiosis and phenotype – Deletions, inversions, duplications, translocations and non-disjunctions – Down's, Turner's and Klinefelter's syndrome – Chromosomal mutations and leukemia: Philadelphia chromosome and Burkitt's lymphoma – Germinal and somatic mutations, mosaicism.

MOLECULAR GENETICS

Relationship between DNA content and organism complexity – DNA assembly in the nucleus of eukaryotic cells – Structural differences between prokaryotic and eukaryotic genes – Genome organization in prokaryotic and eukaryotic cells. Characteristics of human genome – Gene mutations: development mechanisms – Mutation consequences on gene products – Examples of autosomic domint and recessive mutations, as well as X-linked ones – Mytochondrial gene mutations – Genomic instability - DNA plymorphisms and their use as genetic markers – - Elements of developmental biology – Immunogenetics. Generation of antibody diversity - Cancer genetics. Oncogenes and tumor suppressor genes (Rb1, WT1 and p53) – Stategies for the diagnosis of genetic diseases (direct and indirect) – The human genome project: future implications – Gene therapy: general concepts and applications.

Prerequisites

Basic sciences (chemistry, physics)

Teaching form

Lectures

Group discussion
In the Covid-19 emergency period, the frontal lessons will take place remotely in videoconference

Textbook and teaching resource

PPT presentation and registration of lessons

G. De Leo, E. Ginelli, S. Fasano. BIOLOGIA E GENETICA, EdiSES, 2013

other textbooks:

E.Ginelli, M.Malcovati. MOLECOLE, CELLULE E ORGANISMI, EdiSES, 2016

H. Lodish, A. Berk, S.L. Zipursky, P. Matsudaira, D. Baltimore, J. Darnell. MOLECULAR CELL BIOLOGY, FREEMAN, 2016.

Strachan. GENETICA MOLECOLARE UMANA, Zanichelli, 2014

P.J. Russell. GENETICA UN APPROCCIO MOLECOLARE. PEARSON, 2014.

Semester

Second semester

Assessment method

Written exam with multiple choices quiz: total of 20 questions, if > 15 correct answers the student will have access to the oral exam.

Oral examination on the subjects taught during the lectures

and on further subjects (part of the program) not exhaustively discussed in class

Office hours

By appointment