UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

SYLLABUS DEL CORSO

Genetics I

1819-1-H4102D002-H4102D008M

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 better understand the processes involved in normal and pathological situations.

Contents

molecular and cellular mechanisms responsible for gene expression and its regulation, analyzing epigenetic mechanisms, basic concepts of heredity and the transmission patterns of inherited traits; mechanisms which can generate phenotypic variants in men.

Detailed program

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 - The human genome project: future implicationGENETICS – 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 - The human genome project: future implication

Prerequisites

Basic sciences (chemistry, physics)

Teaching form

Lectures

Group discussion

Textbook and teaching resource

Thompson & Thompson Genetics in Medicine, 8e, 2015; iGenetics: Pearson New International Edition: A Molecular Approach. Pearson, 2014

Semester

Second semester

Assessment method

Written exam with multiple choices quiz (30% of the final mark)

Oral examination on the subjects taught during the lectures

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

Office hours

by appointment