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**COURSE (TITLE): Human Genetics**

**LECTURER:**

**YEAR and SEMESTER: 2nd year, 1<sup>st</sup> semester**

**CREDITS (CFU): 6**

**SECTOR (SDS): BIO/18-Genetica**

**ACADEMIC YEAR:**

**ASSESSMENT: questionnaire + oral exam**

**LOCATION: Department of Environmental, Biological and Pharmaceutical Science and Technologies, Via Vivaldi 43 Caserta**

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**COURSE OBJECTIVES/OUTCOMES:**

The aim of this course is to teach the principles of transmission, modification and expression of heritable traits in human beings and populations, through the study of the methods of Mendelian Genetics and Molecular Genetics and the use of modern bioinformatic tools.

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**SYLLABUS (overview)**

The human genome. Gene structure and function. Human genetic diversity: mutation and polymorphism. Cytogenetic and genome analysis. The chromosomal and genomic basis of disease. Inheritance patterns of single-gene traits. Complex inheritance of multifactorial disorders. Principles of population genetics. Identification of the genetic basis for human disease. Molecular databases and their use through bioinformatic tools.

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**SYLLABUS (Detailed description):**

1. The human genome. Gene structure and function. Molecular mechanisms of gene expression and regulation. Epigenetic modifications and their relevance for gene expression. Genomic imprinting and X-chromosome inactivation.
  2. Human genetic diversity. Mutations and polymorphisms. Type of mutations and their consequences.
  3. Cytogenetic and genome analysis. Human chromosomes. Structural and numerical chromosome anomalies. Mechanisms underlying chromosomal anomalies. Uniparental disomy. Microdeletions and microduplications. Chromosomal bases of sex determination.
  4. Inheritance patterns of single-gene traits. Genotype and phenotype. Pedigrees. Autosomal mendelian inheritance. X-linked inheritance.
  5. Non-mendelian single-gene inheritance. Mosaicism. Genomic imprinting defects. Dynamic mutations. The mitochondrial genome and maternal inheritance.
  6. Inheritance of multifactorial traits. Quantitative and qualitative traits. Distinguish between the effects of genotype and environment. Genetic variations in populations. The law of Hardy-Weinberg. Effects of mutation, natural selection, migration, genetic drift, stratification, assortative mating and inbreeding. Ethnic differences in allele frequency.
  7. Gene mapping. Linkage analysis and association studies. Linkage disequilibrium. Positional cloning. GWAS.
  8. DNA and protein databases. Genome browsers. Human mutation databases. Basic tools for
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sequence alignment and analysis.

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**TEXTBOOKS:**

Thompson & Thompson. Genetics in Medicine. 8th Edition. Elsevier

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**ADDITIONAL READING:**

Scientific articles provided by the lecturer during the course

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