COURSE DETAILS
"INTEGRATED COURSE OF HUMAN AND MEDICAL GENETICS"
SSD MED/03; BIO/13*

DEGREE PROGRAMME: MEDICINE AND SURGERY (P11)
COORDINATOR: PROF. NUNZIA PASTORE
ACADEMIC YEAR 2024-2025

GENERAL INFORMATION – TEACHER REFERENCES

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GENERAL INFORMATION ABOUT THE COURSE

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TEACHING LANGUAGE: ENGLISH
CHANNEL (IF APPLICABLE): 1
YEAR OF THE DEGREE PROGRAMME: I
SEMESTER: II
CFU: 7

REQUIRED PRELIMINARY COURSES (IF MENTIONED IN THE COURSE STRUCTURE "REGOLAMENTO")

Molecular and cellular Biology, Biochemistry

PREREQUISITES (IF APPLICABLE)

Previous knowledge on Molecular and Cellular Biology, Histology, Human Embryology and Biochemistry is required for a proper understanding of the topics discussed in this Integrated Course.

LEARNING GOALS

The course focalizes on the study of genetics applied to cells, individuals, families and general population. The learning goal of the course will be to provide students with the most updated information on the contribution of genetics on physiopathology in humans.

EXPECTED LEARNING OUTCOMES (DUBLIN DESCRIPTORS)

Students will be provided with the basic knowledge to enable them to understand the mechanisms underlying monogenic and complex genetic diseases. At the end of the course the student should be capable to a) recognize a genetic disease; b) recommend the most appropriate tests to establish the diagnosis; c) suggest the most appropriate therapeutic approach or direct the patient to the appropriate specialist; d) coordinate and manage the different medical specialists needed for the management of genetic disorders with multisystemic involvement; e) prevent the complication and advice the families on risk factors and reproductive risks.

Knowledge and understanding

This integrated course focuses on the understanding of the basic mechanisms underlying human genetic conditions (cancer included). The course provides students with knowledge and basic methodological tools needed to analyze monogenic and complex genetic disorders. The concomitant professional training activities with the description of clinical cases will help the student to bring the basic knowledge into clinical practice improving medical reasoning.

Applying knowledge and understanding

We aim to train medical doctors who can:

- *Recognize transmission of hereditary normal and pathological characteristics, determine the models of transmission and assess the probability of occurrence in the offspring;*
  Understand the importance of genetic diseases for individuals and their families;
- *Be aware that advances in molecular genetics can have an immediate impact on life and well-being of patients and their families;*
- *Recognize the impact of new knowledge in medical genetics on professional responsibilities of physicians towards patients;*
- *Know how to give patients all available choices to deal in the best possible way with genetic disorders;*
- *Appreciate the need for a doctor to continuously integrate the knowledge of basic sciences with clinical practice in the patient's care;*
- *Be aware of the ethical issues regarding medical genetics;*
- *Be aware of the need for genetic counselling and supportive care for patients undergoing genetic testing.*

COURSE CONTENT/SYLLABUS

Medical Genetics MED03.

Mendelian transmission of hereditary characteristics. Alleles and loci. Dominant and recessive disorders. Homozygosity and heterozygosity. The analysis of family trees. Examples of the most common autosomal dominant, recessive and X-linked genetic diseases. Factors affecting the expression of genes (modifier genes, imprinting, etc.). Penetrance and variable expressivity. The mitochondrial transmission. Anticipation and imprinting in genetic diseases. Calculation of the genetic risk for Mendelian traits. Mutations: different typology and effect of mutations. Haploinsufficiency. International nomenclature of mutations. DNA polymorphism. The molecular mechanisms of genetic recombination. GWAS studies. The multifactorial nature of normal and pathological characters in individuals. Quantitative genetics. Epistasis. The calculation of the empirical risk for multifactorial traits. Polygenic inheritance and multifactorial diseases. Basis for developmental genetics. Epigenetic inactivation of the X chromosome. Basics of Cancers genetics. Familial syndromic tumors.
Loss of heterozygosity. The importance of miRNA in cancer genetics. Hardy-Weinberg equilibrium. Population genetics: selection, drift, founder effect and heterozygote advantage. The impact of Darwinism in medicine. The impact of the human genome project in human and medical genetics. Genomic medicine. Disease gene identification. Functional

genomics. NGS approaches in human and medical genetics. Diagnostic tools in medical genetics. Pharmacogenetics. Therapeutic approaches to genetic diseases. Animal models in human and medical genetics. Genetic counselling, prenatal diagnosis.

Applied Biology BIO13.

Meiosis (Gametogenesis in males and females) – Chromosomes structure. Karyotype – Techniques for standard and molecular cytogenetic analyses - CGH array. Quantitative chromosomal disorders – Euploidy and aneuploidy – Trisomy disorders in humans. Trisomy 21, 18, 13, aneuploidies of sexual chromosomes, Turner and Klinefelter. Microdeletions syndromes: Wolf-Hirschhorn, Cri-du-chat, Williams, Di George. Qualitative Chromosomal disorders – Balanced rearrangements: Inversions, reciprocal translocations - Unbalanced rearrangements: deletions, unbalanced translocations, isochromosomes, ring chromosomes. Techniques to detect chromosomal abnormalities.

TEACHING ACTIVITIES			
Week	**Day / Hour**	**Lessons/Seminars**	**Teacher**
3°W 24-28 March 2025	**Fri** **15.00-17.00**	Medical Genetics. Introduction to Medical genetics. The concept of precision medicine. Mendel laws and their implications in medical genetics. Mutation mechanisms and international nomenclature of mutations.	Pinelli
4°W 31 March- 4 April 2025	**Fri** **15.00-17.00**	Medical Genetics. How to build a pedigree. Autosomal dominant inheritance. Acondroplasia. Osteogenesi imperfetta, CMT1, diseases due to mutations in FGFR3. Autosomal recessive diseases. CF and haemoglobinopathies.	Pinelli
5°W 7-11 April 2025	**Tue** **13.00-15.00**	Medical Genetics. X-linked inheritance (dominant and recessive). The examples of Duchenne/Becker and hemophilia. X inactivation and genetic diseases	Lauritano
	Thurs **15.00-17.00**	Medical Genetics. Exceptions to mendelian inheritance (incomplete penetrance and variable expressivity, allelic and locus heterogeneity). Waardenburg, Marfan, ADPKD, Neurofibromatosis, Retinitis pigmentosa.	Lauritano
	Fri **15.00-17.00**	Medical Genetics. Diseases due to trinucleotide expansions: Huntington disease, X-fragile, Myotonic dystrophy, Friedreich ataxia, Diseases due to polyalanine expansion. Genomic imprinting and associated diseases (Angelman-Prader-Willi, Beckwith-Wiedemann).	Lauritano
6°W 14–16/23-24 April 2025	**Mon** **13.00-15.00**	Medical Genetics. Techniques in molecular genetics (Southern blotting, PCR, electrophoresis, restriction enzyme analysis, DNA chip, sequence analysis) - Next generation sequencing.	Rosato
	Tue **15.00-17.00**	Medical Genetics. Population Genetics. General principles and tools (Hardy Weinberg). Linkage disequilibrium.	Pinelli
	Wed **13.00-15.00**	Medical Genetics. Mitochondrial disorders (LHON, MLS, Leigh). Multifactorial diseases. Behavioral genetics. The autistic spectrum (ASD) Attention deficit hyperactivity disorder (ADHD). Schizophrenia.	Pinelli
	Fri **15.00-17.00**	Applied Biology. Meiosis, gametogenesis	Izzo
7°W 28-30 April 2 May 2025	**Tue** **9.30-11.30**	Applied Biology. Chromosomes structure. Techniques for standard and molecular cytogenetic analyses	Izzo
	Fri **15.00-17.00**	Applied Biology. Karyotype – Techniques for standard and molecular cytogenetic analyses. Quantitative chromosomal disorders – Euploidy and aneuploidy.	Izzo
8°W 5-9	**Tue** **15.00-17.00**	Applied Biology. Trisomy disorders in humans. Trisomy 21, 18, 13	Izzo

May 2025	**Thurs 15.00-17.00**	Applied Biology. Aneuploidies of sexual chromosomes, Turner and Klinefelter. Microdeletions syndromes: Wolf-Hirschhorn, Cri-du-chat, Williams, Di George	Izzo
	Fri 15.00-17.00	Applied Biology. ADI	Izzo
9°W 12-16 May 2025	**Tue 15.00-17.00**	Applied Biology. Qualitative Chromosomal disorders	Izzo
	Thurs 15.00-17.00	Applied Biology. Balanced rearrangements: Inversions, reciprocal translocations	Izzo
	Fri 15.00-17.00	Applied Biology. Unbalanced rearrangements: deletions, unbalanced translocations, isochromosomes, ring chromosomes	Izzo
10°W 19-23 May 2025	**Tue 13.00-15.00**	Applied Biology. Techniques to detect chromosomal abnormalities.	Izzo
	Thu 15.00-17.0	Medical genetics. Pharmacogenomics and Pharmacogenetics.	Pastore
	Fri 13.00-17.00	Medical genetics. Genetic tests. Diagnostic, predictive and carrier testing - Risk assessment Bioethical issues, Genetics counseling, Prenatal diagnosis **ADI.** Risk assessment and genetic counseling	Rosato /Pinelli
11°W 26-30 May 2025	**Mon 13.00-15.00**	Medical genetics. Cell cycle. Oncogenes and oncosoppressors. Adult and pediatric tumors: clinical phenotypes and molecular mechanisms - Hereditary tumors.	Calcagni
	Tue 15.00-17.00	Medical genetics. miRNA in cancer and genetic diseases.	Rosato
	Thurs 15.00-17.00	Medical genetics. Animal models in human and medical genetics. Gene expression control. Epigenetics. Transcriptional modulation and their influence in medical genetics.	Pastore
	Fri 13.00-17.00	Medical genetics. Autophagy and lysosomal disorders. **ADI** Interpretazione di pedigree	Calcagni
12°W 2-6 June 2025	**Tue 15.00-17.00**	Medical genetics. The human genome project. Structure and function of the human genome (including repeated sequences structure and functions Sine, Alu etc). Functional genomics. Available Databases and their use in medical genetics. OMICS in medical genetics (transcriptomics, proteomics, metabolomics)	Pastore
	Thurs 15.00-17.00	Medical genetics. Classification of Human malformations. Developmental biology. Signal transduction pathways critical for human development. Examples of developmental disorders.	Calcagni
	Fri 15.00-17.00	Medical genetics. Therapeutic approaches	Pastore

READINGS/BIBLIOGRAPHY (last available edition)

- *Nussbaum, McInnes, Willard, Thompson & Thompson Genetics in Medicine, Elsevier*
- *Tom Strachan, Andrew Read. Human Molecular Genetics, Garland Sciences*
- *Tom Strachan, Goodship, Chinnery. Genetics and Genomics in Medicine, Garland Sciences.*

Other supporting didactic material available at the website of Course Coordinator.

TEACHING METHODS

Teachers will use for this course lectures, practical exercises on various activities (e.g., calculation of the risk factors, interpretation of pedigrees, interpretation of chromosomal abnormalities), description of actual clinical cases observed at the Medical Genetic Service.

Online material can be found at https://www.ncbi.nlm.nih.gov/books/NBK1116/.

EXAMINATION/EVALUATION CRITERIA

*For **integrated courses**, this field should encompass all modules, with indication of the relative weight of each module on the final mark. For integrated courses, this field should be coordinated by the reference teacher for the course.*

a) Exam type:

*For **integrated courses**, there should be one exam.*

Exam type	
written and oral	
only written	
only oral	X
project discussion	
other	

In case of a written exam, questions refer to: (*)	**Multiple choice answers**	
	Open answers	
	Numerical exercises	

(*) multiple options are possible

b) Evaluation pattern:

The oral exam consists in questions on the topics illustrated from the Medical Genetics and Applied Biology modules indicated in the syllabus. The students will be questioned also on the practical activities performed during the course