



UNIVERSITÀ DEGLI STUDI DI PALERMO

DEPARTMENT	Scienze e Tecnologie Biologiche, Chimiche e Farmaceutiche		
ACADEMIC YEAR	2022/2023		
BACHELOR'S DEGREE (BSC)	BIOTECHNOLOGIES		
INTEGRATED COURSE	GENETICS - INTEGRATED COURSE		
CODE	03577		
MODULES	Yes		
NUMBER OF MODULES	2		
SCIENTIFIC SECTOR(S)	BIO/13, BIO/18		
HEAD PROFESSOR(S)	CORONA DAVIDE	Professore Associato	Univ. di PALERMO
OTHER PROFESSOR(S)	CORONA DAVIDE	Professore Associato	Univ. di PALERMO
	CONIGLIARO ALICE	Professore Associato	Univ. di PALERMO
CREDITS	12		
PROPAEDEUTICAL SUBJECTS			
MUTUALIZATION			
YEAR	2		
TERM (SEMESTER)	2° semester		
ATTENDANCE	Mandatory		
EVALUATION	Out of 30		
TEACHER OFFICE HOURS	CONIGLIARO ALICE Monday 15:00 18:00 Sezione di Biologia e Genetica del Dipartimento BiND in via Divisi 83, oppure attraverso la piattaforma TEAMS. A causa di possibili impegni istituzionali o riunioni di lavoro potrebbe non essere possibile ricevere gli studenti nel giorno e alle ore indicate, pertanto sarebbe preferibile fissare un appuntamento tramite e-mail. CORONA DAVIDE Monday 09:00 12:00 Dipartimento STEBICEF Viale delle Scienze - Edificio 16 Tuesday 09:00 12:00 Sede del Consorzio Universitario, corso Vittorio Emanuele, 92, 93100 Caltanissetta		

PREREQUISITES	Basic knowledge of Cell Biology, Biochemistry and Molecular Biology
LEARNING OUTCOMES	Comprehension and ability to understand: Acquisition of integrated cultural skills in formal and molecular genetics; acquisition of advanced scientific knowledge about biochemical, molecular, functional and evolutionary aspects of genes and genomes. Understanding the molecular mechanisms of cancer and of basic laboratory techniques for the analysis of nucleic acids and their fields of application in research and diagnostics. Capacity to apply Comprehension and understanding: Acquisition of in-depth methodological, technological and instrumental knowledge,, with reference to typical genetic survey instrumental methodologies; techniques of acquisition and analysis of data; statistical tools and support information. Making judgments: Acquisition of conscious independent judgment in evaluating and interpreting data and processing of specialized scientific literature. Communicative ability: Acquisition of adequate skills and tools for communication with reference to the ability to present experimental and bibliographic data and the transmission and dissemination of information on the molecular genetics of topical themes. Learning ability: Acquisition of adequate capacity for the development and deepening of competencies, with reference to the consultation of databases of DNA sequences, structure and organization of genes, etc .; learning of molecular genetic technologies and innovative functional genomics; the use of advanced cognitive tools for the continuous updating of knowledge.
ASSESSMENT METHODS	Evaluation in progress , written test and final oral examination, scoring evaluation in thirtieths. The tests that contribute to student assessment are: two written tests (one to be held in the middle "test in progress" and the other at the end of the course) and an oral presentation, using power point, in the classroom at the end of course on an topic agreed with the teacher. The written tests, each lasting 60 minutes, consist of twenty-six multiple choice questions (1 point will be assigned for every correct answer) and two open questions (up to three points for every open question). To pass each of the two quiz tests a minimum of 18 points are required. The final exam grade, out of thirty, will be averaged by the grades obtained in the two written tests and in the oral presentation. The student who gives up or fails at least one of two written tests must undergo an oral examination on the entire program and must conduct the power point presentation. The tests described above are proposed to assess whether the student has acquired knowledge and understanding of the topics, has attained ability to interpret and to judge independently and has gained adequate analytical and communication skills. The evaluation criteria for the oral exam will be: - Excellent: 30 - 30 cum laude Outcome: excellent knowledge of the topics, excellent language properties, good analytical ability, the student is able to apply knowledge to solve the proposed problems - Very good: 26-29 Outcome: good knowledge of the topics, full ownership of language, good analytical ability, the student is able to apply knowledge to solve the proposed problems - Good: 24-25 Outcome: good knowledge of the topics, good language properties, limited ability to independently apply knowledge to solve the problems proposed - Satisfactory: 21-23 Outcome: the candidate does not have full mastery of the main topics but possesses the knowledge, satisfactory language properties, low ability to independently apply the acquired knowledge for the resolution of problems - Sufficient: 18-20 Outcome: basic knowledge of the main topics and of the technical language, little or no ability to autonomously apply the acquired knowledge - Insufficient Outcome: the candidate does not have an acceptable knowledge of the topics treated during the course
TEACHING METHODS	Lectures, classroom exercises, laboratory exercises

MODULE
GENERAL AND MOLECULAR GENETICS

Prof. DAVIDE CORONA

SUGGESTED BIBLIOGRAPHY

Snustad e Simmons – Principi di Genetica, Ed. Edises
Strakan e Reed – Genetica Molecolare Umana – Ed. UTET
Peter J. Russel - iGENETICS A Molecular Approach - Ed Pearson

AMBIT	50078-Discipline biotecnologiche comuni
INDIVIDUAL STUDY (Hrs)	98
COURSE ACTIVITY (Hrs)	52

EDUCATIONAL OBJECTIVES OF THE MODULE

The General and Molecular Genetics module aims at providing the student with basic knowledge on the inheritance of genetic traits and on the molecular mechanisms responsible for transmitting the genetic information in prokaryotic and eukaryotic organisms.

SYLLABUS

Hrs	Frontal teaching
12	Principles of genetic transmission . Segregation of alleles and independent assortment . multiple , dominant alleles . Atypical Mendelian ratios and variability of gene expression . Inheritance associated with sex . Mendelian genetics in humans : family trees , association maps . physical basis of association : crossing-over and recombination . frequency of recombination and gene order . genetic maps , cytogenetic maps and physical maps . The Genome projects , organization of databases , and supporting tools .
6	Genetics of Bacteria : transformation , conjugation and transduction . The selective and non-selective systems . Bacteriophages : titration. The various stages of the infection cycle : lytic cycle and lysogenic . Transposable elements .
10	The flow of genetic information , and organization of the genome : Complementation , cistron and gene concept . General aspects of replication . Protein synthesis and the genetic code . Structure of prokaryotic and eukaryotic chromosomes . Structure and organization of the eukaryotic genome . Multigene families : origin and evolution. Evolutionary Genetic elements . Extranuclear inheritance : structure and expression of the mitochondrial DNA and chloroplast .
6	The regulation of gene expression : The basics of transcriptional regulation in prokaryotes , concept of operon , genetic evidence of allostery . mechanisms of regulation of transcription in eukaryotes : transcriptional regulation , post- transcriptional regulation mechanisms .
6	Origin of genetic variability: Gene mutations , spontaneous , induced , action of mutagenic agents . Molecular basis of gene mutations and repair mechanisms . chromosomal mutations : structural changes , alterations in the number . mutation ratio : phenotype , gene mutations and altered proteins .
Hrs	Workshops
12	Basic Drosophila handling, sexing, virgining, hybrid cross, X chromosome linked inheritance

MODULE APPLIED MOLECULAR GENETICS

Prof.ssa ALICE CONIGLIARO

SUGGESTED BIBLIOGRAPHY

L'essenziale di biologia molecolare della cellula. Autori: Alberts, Bray ed altri; Casa Editrice: Zanichelli - 2020 ISBN: 9788808520241

- La Cellula: un approccio molecolare. Cooper Geoffrey. Piccin Editore, 2011. ISBN: 8829921335
- Genetica molecolare umana, Tom Strachan Andrew Read, Seconda edizione Zanichelli
- Reviews ed articoli forniti dal docente.

Essential Cell Biology. Alberts B and Others. Fifth Edition 2018. WW Norton & Co. ISBN9780393680393

The Cell: A molecular Approach. Cooper Geoffrey. Edition 2019. OUP USA. ISBN 1605358630

- Reviews on specific topic provided by the teacher

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INDIVIDUAL STUDY (Hrs)	98
COURSE ACTIVITY (Hrs)	52

EDUCATIONAL OBJECTIVES OF THE MODULE

The student at the end of the module must have gained cultural and technical basis in molecular pathology and molecular diagnostic methods that allow:

- To know the different molecular tests through which it is possible to identify the mutations occurring in the genome;
- To interpret the processes that, through the activity of genes, lead to cancer development;
- The analysis to assess the predisposition to cancer;
- The experimental models and the experimental activity on cells or their components to understand the specific phenotypes related to the development and progression of cancer.

SYLLABUS

Hrs	Frontal teaching
16	Strategies and techniques for the identification and analysis of mutations: allelic Series and polymorphisms, analysis of restriction polymorphisms (mini- and microsatellites) . Diagnostic Use of polymorphisms. Techniques for the "screening" or the detection of specific mutations and related examples in the biomedical field: RFLP and sickle cell anemia; ASO, dot blot and reverse dot blot (Thalassemia); ARMS-PCR and OLA (Cistic Fibrosis); Southern blot and PCR (muscular dystrophy); Protein truncation test (Becher dystrophy); Hetero duplex analysis: SSCP; DGGE; DHPLC expansion of triplets (Fragile X syndrome and Huntington's chorea); Mitochondrial diseases and heteroplasmy; Genomic Imprinting and RFLP (methylation specific PCR) and Prader Willi Syndrome, Angelmann Syndrome. Gene sequencing (Sanger and Next Generation Sequencing); Prenatal diagnosis
12	Classification of tumors; Concept of malignancy; Initiation and promotion; mutagenesis; Tumor Clonality , 'Viral Mutagenesis; DNA and RNA viruses ; Experiments of Bishop and Varmus and their meaning; Weinberg experiments and their meaning Oncogenes and protooncogenes
12	Classification of Oncogenes; Mutational mechanisms of tumor suppressor genes and Oncogenes; The Retinoblastoma gene: Identification and Function; Cell Cycle; The structure and function of P53 gene ; The structure and function of MDM2 gene; The telomerase; The Mutators Genes ; Physiological and pathological angiogenesis; The Metastatic Cascade
Hrs	Workshops
12	mRNA isolation from cultured cells; Real Time PCR, TaqMan probes, analysis of sequence variations using RealTime PCR. Technology for genotyping. Use of different markers for genotyping.