



# UNIVERSITÀ DEGLI STUDI DI PALERMO

<b>DEPARTMENT</b>	Biomedicina, Neuroscienze e Diagnostica avanzata		
<b>ACADEMIC YEAR</b>	2024/2025		
<b>BACHELOR'S DEGREE (BSC)</b>	NEUROPHYSIOPATHOLOGY TECHNIQUES		
<b>INTEGRATED COURSE</b>	BIOLOGICAL AND MOLECULAR BASES OF LIFE		
<b>CODE</b>	22325		
<b>MODULES</b>	Yes		
<b>NUMBER OF MODULES</b>	3		
<b>SCIENTIFIC SECTOR(S)</b>	BIO/13, MED/03, BIO/10		
<b>HEAD PROFESSOR(S)</b>	CARLISI DANIELA	Professore Associato	Univ. di PALERMO
<b>OTHER PROFESSOR(S)</b>	CARLISI DANIELA	Professore Associato	Univ. di PALERMO
	CONIGLIARO ALICE	Professore Associato	Univ. di PALERMO
	PICCIONE MARIA	Professore Associato	Univ. di PALERMO
<b>CREDITS</b>	6		
<b>PROPAEDEUTICAL SUBJECTS</b>			
<b>MUTUALIZATION</b>			
<b>YEAR</b>	1		
<b>TERM (SEMESTER)</b>	1° semester		
<b>ATTENDANCE</b>	Mandatory		
<b>EVALUATION</b>	Out of 30		
<b>TEACHER OFFICE HOURS</b>	<p><b>CARLISI DANIELA</b> Monday 08:30 16:30 Sono disponibile per il ricevimento ogni giorno previo appuntamento, presso la Sezione di biochimica del Policlinico. Accanto la biblioteca di Medicina. Contatto: daniela.carlisi@unipa.it</p> <p><b>CONIGLIARO ALICE</b> 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.</p> <p><b>PICCIONE MARIA</b> Friday 13:00 14:00 UOC Genetica Medica Ospedale Cervello via Trabucco 180 Palermo</p>		

<p><b>PREREQUISITES</b></p>	<p>Students must have a background of chemistry in order to understand the interaction of molecules within the cells, biology and biochemistry. functional alphabetic competence; - mathematical competence and competence in science, technology and engineering; - digital competence; - personal, social competence and ability to learn to learn.</p>
<p><b>LEARNING OUTCOMES</b></p>	<p>-Knowledge and understanding: Acquisition of the specific language of the disciplines of Biology, Biochemistry and medical genetics. Know the basis of the cellular organization of living organisms; know and understand basic biological processes such as growth, cell division, sexual reproduction and embryonic development; the mechanisms of replication of the genetic material and its onset variability; the mechanisms of gene expression, the genetic basis of heredity and the methods of transmission of hereditary characteristics. know and understand the characteristics of biological molecules and the underlying principles of their chemical-physical behavior, to know the structure and properties of organic compounds, learn about molecules of biological interest and some cellular metabolic processes; ---Ability to apply knowledge and understanding: Ability to recognize and independently apply the knowledge of basic biological and biochemical processes of cells and organisms; the laws that regulate the transmission of hereditary characteristics. ---Judgment independence: Be able to evaluate and integrate knowledge independently acquired in biology, genetics and biochemistry in the study of organisms and in detail of man; the implications that alterations of biological processes, biochemical and cellular, have on human pathologies; ---Communication skills: Ability to present simply and communicate clearly, i main processes of biology, genetics and biochemistry. ---Learning ability: Ability to update knowledge in the biomedical field through consultation of the scientific bibliography specific to the sector of expertise; Ability to learn and follow appropriately, using knowledge acquired, the subsequent courses of your study curriculum; Capacity' to delve deeper into the topics also by attending courses or seminars specific courses or 1st level Masters</p>
<p><b>ASSESSMENT METHODS</b></p>	<p>Students will take oral exams and will be minimum proposed three questions on all parts of each module program . The final examination aims to evaluate: a) knowledge and understanding of principal topics. To verify whether the student has reached interpretative competence. b) analytical and synthetic capabilities; the students must show their ability to apply knowledge in the professional context and to solve complex problems. c) the ability of the student to interact with the examiner, with his presentation and argumentative skills, d) if he has acquired the specific terminology of the basic disciplines. The grades are on a scale from 18 to 30 for each presentation; the final grade will be the average of the result of the two modules. It will be assigned as the following evaluation grid: A – A+= Excellent Excellent knowledge of teaching contents; students should show high analytical and synthetic capabilities and should be able to apply their knowledge to solve highly complex problems. Ability to express ideas clearly and effectively. Ability to listen. B =Very good Very good knowledge of the teaching contents and excellent language control; students should show analytical and synthetic skills and be able to apply their knowledge to solve problems of medium and, in some cases, even higher complexity. C= Good Good knowledge of teaching contents and good language control; the students should be able to apply their knowledge to solve problems of medium complexity D=Satisfactory Average knowledge of the teaching contents, in some cases limited to the main topic; acceptable ability to use the specific discipline language and independently apply the acquired knowledge. E= Sufficient Minimum teaching content knowledge, often limited to the main topic; modest ability to use the subject specific language and independently apply the acquired knowledge. F= Fail Lack of an acceptable knowledge of the main teaching content knowledge; very</p>

	little or no ability to use the specific subject language and apply independently the acquired knowledge. Compensatory tools and dispensatory measures will be guaranteed by the Disability and Neurodiversity Center - University of Palermo (Ce.N.Dis.) to students with disabilities and neurodiversity, based on specific needs and in implementation of current legislation
<b>TEACHING METHODS</b>	lectures will be offered by the teacher using power point slides

**MODULE  
APPLIED BIOLOGY**

*Prof.ssa ALICE CONIGLIARO*

**SUGGESTED BIBLIOGRAPHY**

- P.Bonauo, C.Crisafulli, R.D'Angelo, M.Francolini, S.Grimaudo, C.Rinaldi, P.Riva, M.G.Romanelli. Editore: EdiSES. ISBN9788833190389

-Chiara Donati, Massimo Stefani, Nicolò Taddei " Biologia e Genetica " Zanichelli, 2017 ISBN: 978-88-08-52052-4

<b>AMBIT</b>	10338-Scienze biomediche
<b>INDIVIDUAL STUDY (Hrs)</b>	30
<b>COURSE ACTIVITY (Hrs)</b>	20

**EDUCATIONAL OBJECTIVES OF THE MODULE**

Understand the cellular organization that underlies living organisms.  
- To know the basic biological processes of molecular biology.  
- Understanding the processes of gene expression.  
- Understanding the mechanisms of cell division and sexual reproduction

**SYLLABUS**

<b>Hrs</b>	<b>Frontal teaching</b>
2	Introduction to the course: illustration of the objectives of the course, the methods of exams, and the teaching material. Living organism concept. The cell: structural and functional organization. Differences between eukaryotic and prokaryotic cells.
4	Biological membranes structure and functions. Membrane transport. Cellular receptors.
2	Nucleic acids: structure and function; DNA as genetic material; Main classes of RNA.
2	Gene expression: Transcription and Maturation of primary transcripts in eukaryotes.
4	Genetic code; ribosome structure and protein synthesis: initiation, elongation and termination phases of translation.
2	DNA duplication process in eukaryotes.
2	Chromatin and Chromosomes; Cell cycle and Mitosis.
2	Meiosis and gametogenesis in humans. Sexual reproduction.

## MODULE BIOCHEMISTRY

*Prof.ssa DANIELA CARLISI*

### SUGGESTED BIBLIOGRAPHY

"Le basi della biochimica" di Emine E. Abali, Susan D. Cline, David S. Franklin, Susan Viselli terza edizione italiana condotta sulla ottava edizione americana a cura di Niccolò Taddei; ed Zanichelli; ISBN: 9788808299826. 2023

"Chimica e Biochimica" di Bertoldi M, Colombo D, Magni F, Marin O, Palestini P; ed EdiSES. 2015; ISBN 9788879598781

<b>AMBIT</b>	10338-Scienze biomediche
<b>INDIVIDUAL STUDY (Hrs)</b>	30
<b>COURSE ACTIVITY (Hrs)</b>	20

### EDUCATIONAL OBJECTIVES OF THE MODULE

The objective of the biochemistry module is to provide students with the knowledge necessary to understand the main biochemical processes that allow cell life and the functioning of the organism as a whole.

The student must know the structure and function of the main biological macromolecules; Understand the main metabolic processes; understand the mechanisms that regulate and integrate the biochemical processes and connect them with some disease.

## SYLLABUS

Hrs	Frontal teaching
2	General concepts of general and inorganic chemistry
1	Aminoacids. Protein structure and different levels of molecular organization. Oxygen-binding chromoproteins. Myoglobin (Mb). Hemoglobin (Hb).
2	Enzyme: General concepts. Mechanism of enzymatic catalysis. The active site. Enzyme specificity. Isoenzymes. Constitutive and inducible enzymes. Enzyme kinetics. Coenzymes and prosthetic groups.
1	Fatty acids and Lipids: biological characteristics of lipids; physical properties.
2	Structure and composition of biological membranes. Membrane transport. Osmosis. Simple and facilitated diffusion. Glucose transporters (GLUTs). Primary and secondary active transport. Sodium-glucose symporter. Sodium/potassium –dependentATPase.
2	Membrane receptors and signal transduction mechanisms.
2	Introduction to metabolism: catabolic and anabolic pathway. Role of ATP and reducing power in the connection between catabolism and anabolism.
1	Carbohydrates: Definition and description of carbohydrates; generality and biological role.
5	Glucidic metabolism: Glycolysis, pentose phosphate pathway Glycogenolysis, glycogen synthesis and their regulation. Fermentations and LDH. The metabolic fate of pyruvate. The Krebs cycle. The mitochondrial respiratory chain and oxidative phosphorylation and ATP synthesis. Gluconeogenesis.
2	Metabolism of lipids. Digestion and absorption of fatty acids. Beta-oxidation regulation and energy yield. Adjustment. Formation, properties and role of ketone bodies. The synthesis of fatty acids and its regulation. Lipoproteins and atherosclerosis.

**MODULE  
MEDICAL GENETICS**

*Prof.ssa MARIA PICCIONE*

**SUGGESTED BIBLIOGRAPHY**

Giovanni Neri e Maurizio Genuardi  
Genetica umana e medica  
Editore: Masson-Edra 2014

<b>AMBIT</b>	10338-Scienze biomediche
<b>INDIVIDUAL STUDY (Hrs)</b>	30
<b>COURSE ACTIVITY (Hrs)</b>	20

**EDUCATIONAL OBJECTIVES OF THE MODULE**

**LEARNING OUTCOMES**

**Knowledge and ability to understand**

By the end of the monographic course in MEDICAL GENETICS, students should have acquired adequate knowledge (to the extent of their competence) and the ability to understand topics concerning: the main genetic, genomic, chromosomal and methylation pattern alteration diseases, diagnosis techniques, the care of patients suffering from genetic disease

**Ability to apply knowledge and understanding**

By the end of the course, students must have attained the ability to translate the acquired knowledge into professional practice. In particular, they must be able to:

have personal, social and learning to learn skills

Use the corpus of theoretical knowledge from other disciplines useful in recognising the health and care needs of children with genetic and/or chromosomal disorders

acquire personal, social and learning to learn skills

integrate theoretical and practical knowledge with other social and anthropological sciences in order to identify the needs of adult and/or child patients with genetic disease, counselling and support/support also for the parental couple

inform the adult patient and/or child and parents about the diagnostic process, natural history and treatment of genetic and/or chromosomal disorders

**Autonomy of judgement**

Students must achieve critical and judgemental skills. To this end, they must have learnt the methodological procedure of argumentation, which ensures the acquisition of these skills.

The teaching methods and tools for the achievement of this descriptor are: group work for the discussion of themes and issues related to the study programmes; active and participative lectures by students, making use of dialogue, debate, guided reading of international literature.

**Communication skills**

To be able to express in all forms of oral interview the information acquired and to translate it into clinical practice; to demonstrate effective communication skills with users and in relations with other professionals; to demonstrate communication skills with children and their parents; to provide with awareness and responsibility the professional service that must always be respectful of the rights of the person assisted and of the ethical and deontological values of reference; to present the work elaborated during the training period.

**Learning capacity**

- To develop the ability to continue to study independently in order to constantly update one's knowledge by collecting, organising and interpreting information from the various paper and computer sources. Ability to pursue studies fully, using the knowledge acquired in the course;

- To develop actions for comprehensive care in the case of disabling or inauspicious pathologies.

- To develop personal, social and learning to learn competence

**COURSE LEARNING OBJECTIVES**

Basic knowledge of the main genetic, genomic, chromosomal and / or methylation pattern alteration syndromes.

Knowledge of the natural history and follow-up programs of the syndromes studied for a global management and a person-centered habilitation / rehabilitation intervention.

**EVALUATION METHOD**

Oral test (oral interview aimed at verifying both acquired knowledge and exposition)

Compensatory tools and dispensatory measures will be guaranteed by the Disability and Neurodiversity Center - University of Palermo (Ce.N.Dis.) to students with disabilities and neurodiversity, based on specific needs and in implementation of current legislation

**SYLLABUS**

<b>Hrs</b>	<b>Frontal teaching</b>
2	When to suspect a genetic syndrome Clinical-diagnostic algorithm Laboratory techniques for the diagnosis of genetic and/or chromosomal syndromes
8	Main chromosome syndromes (Down syndrome, Turner syndrome, Klinefelter syndrome, trisomy 13, trisomy 18, mosaic trisomy 8, 5p- syndrome, 4p- syndrome, genomic rearrangement syndromes, supernumerary chromosome markers etc.)
8	Main genetic syndromes (fragile X syndrome, overgrowth syndromes, genomic imprinting anomaly syndromes, etc.)
2	Main congenital metabolic diseases