



## TEACHING SCHEDULE

<b>Master's degree</b>	<b>Biology</b>
<b>Teaching course:</b>	<b>Clinical and Applied Biochemistry</b>
<b>Credits:</b>	<b>10</b>
<b>Year</b>	<b>I</b>
<b>Semester:</b>	<b>II</b>
<b>Professor:</b>	<b>Alberto Zullo</b>
<b>Teaching support:</b>	<b>none</b>
<b>Student reception time:</b>	<b>Tuesday 10:30 - 12:30 AM</b>
<b>Address:</b>	<b>Via Port'Arsa, 11</b>

### COURSE PRESENTATION:

The course deals with the following topics:

- Biochemical mechanisms involved in the pathophysiology of metabolic diseases: diabetes, obesity, hypertension.
- Isolation and analysis of nucleic acids and proteins.
- Biochemical bases of congenital myopathy and cardiomyopathy: canalopathies.
- Clinical Biochemistry: basic and advanced diagnostic procedures - analytical techniques used in the diagnostic procedures - use, acquisition and interpretation of diagnostic test results.

The course, through the study of the biochemical characteristics of biological molecules, the biochemical mechanisms involved in the pathophysiology of diseases, and the analytical techniques for biological macromolecules, allows the student to acquire theoretical bases to solve diagnostic problems with laboratory methods.

### EDUCATIONAL TARGETS

Knowledge and understanding:

Knowledge of the biochemical mechanisms involved in the pathophysiology of diabetes, obesity, and hypertension. Understanding of the biochemical bases of congenital *cores* miopathies (Central Core Disease) and cardiomyopathy (long QT). Understanding the chemical-physical processes underlying the isolation and analysis of nucleic acids and proteins. Understanding basic and advanced diagnostic procedures. Understanding the analytical techniques used in diagnostic procedures. Understanding the use, acquisition and interpretation of diagnostic test results. Ability to link diagnostic issues with diagnostic procedures and biological molecules.

Practical application of knowledge and understanding:

The knowledge of the biochemical characteristics of biological molecules, the biochemical mechanisms involved in the pathophysiology of diseases, and the analytical techniques of biological macromolecules provides the theoretical bases for solving diagnostic problems with laboratory methods.

Autonomy of judgment:

During the course, students are encouraged to deepen their own learning and to apply the theoretical knowledge acquired to solve specific questions raised during training time.

Communicative skills:

The course also aims to stimulate the students' ability to communicate the acquired knowledge using specific and appropriate languages.

Ability to learn on their own:

Students are encouraged to deepen and update their knowledge by independently consulting specialized books and scientific articles.

## **REQUESTED PREREQUISITES**

Basic concepts of organic chemistry and general chemistry;

In-depth knowledge of general biochemistry;

Knowledge of basic concepts of mathematics and physics

## LESSON ATTENDANCE

Topics discussed in the course are complex and specialized. Therefore, a learning session enriched with insights, examples, critical analysis, and discussions with the professor, help students with learning and improves their competence.

## COURSE CONTENT

Biochemical mechanisms involved in the physiopathology of metabolic diseases: definition, epidemiology, molecular bases, diagnostic tests for diabetes, obesity and hypertension. Canalopathies: definition; molecular bases; electrochemical equilibrium in the cell; study of the biophysical properties of ion channels using the patch clamp technique. Malignant Hyperthermia and Central core disease: definition, epidemiology, pathophysiology, molecular bases, diagnostic tests. Hereditary arrhythmic syndromes: definition, epidemiology, pathophysiology, molecular bases, diagnostic tests for long QT syndrome. Laboratory safety guidance: risk assessment, prevention and precaution. Diagnostic procedures: privacy, biological samples, sampling mode. Analytical techniques and instruments used in diagnostic procedures: centrifugation and fractionation techniques. Electrophoresis of proteins and nucleic acids. Principles and applications of chromatography, HPLC, dHPLC, Western blotting, spectrophotometry, fluorimetry, chemiluminescence. Nucleic acid extraction procedures. Principles and applications of FISH, CGH, PCR, MLPA, QF-PCR and sequencing techniques. Cell culture procedures. Fluorimetric and immunofluorescence techniques: cytofluorimeter and fluorescence microscope. Acquisition, use and interpretation of biochemical data in diagnostic tests. Basics of diagnostic tests: variability, reference values, true value, estimate, error, precision, accuracy, quality control, interpretation of an analytical results, false positive and false negative, discriminating value (ROC curve), sensitivity, specificity, predictability, prevalence, incidence. Elements of Statistics.

## TEACHING METHODS

Frontal lessons, case-reports and workshops

## REFERENCE BOOKS

- Cardiopatie congenite dell'adulto. R. Calabrò, L. Daliento, B. Sarubbi. Piccin, 2008.
- Ion Channels of Excitable Membranes (3rd Edition). B. Hille. Sinauer Associates Inc, 2001.
- Patch Clamping: An Introductory Guide to Patch Clamp Electrophysiology. A. Molleman. John Wiley and Sons, LTD, 2003.
- Biochimica e biologia molecolare: Principi e tecniche. Wilson Keith-Walker John. Raffaello Cortina Editore, 2006.

- Medicina di laboratorio e diagnostica genetica. L. Sacchetti, P. Cavalcanti, G. Fortunato. Idelson-Gnocchi, 2007.
- Malignant hyperthermia: a review. Rosenberg H, Pollock N, Schiemann A, Bulger T, Stowell K. Orphanet J Rare Dis. 2015 Aug 4;10:93. doi: 10.1186/s13023-015-0310-1. Review.
- Core myopathies and malignant hyperthermia susceptibility: a review. Brislin RP, Theroux MC. Paediatr Anaesth. 2013 Sep;23(9):834-41. doi: 10.1111/pan.12175. Epub 2013 Apr 25.
- Core myopathies. Jungbluth H, Sewry CA, Muntoni F. Semin Pediatr Neurol. 2011 Dec;18(4):239-49. doi: 10.1016/j.spen.2011.10.005.
- DNA sequencing technologies: 2006-2016. Mardis ER. Nat Protoc. 2017 Feb;12(2):213-218. doi: 10.1038/nprot.2016.182. Epub 2017 Jan 5.
- Fetal Medicine: Basic Science and Clinical Practice 2nd edition. Edited by Charles H. Rodeck and Martin J. Whittle. London: Churchill Livingstone, 2008.
- Widmann - Interpretazione clinica degli esami di laboratorio 11/ed . Ronald A. Sacher, Richard A. McPherson. McGraw-Hill, 2001.
- Materiale didattico distribuito durante il corso.

## EXAM

Student assessment is based on oral interview. Evaluation criteria: relevance of the answers to the questions; content quality; ability to link different course topics; ability to give examples; use of technical language; overall expressive capacity of the student.

## EXAM SCHEDULE

<http://www.dstunisannio.it/index.php/biologia-31/appelli-31>

## EXAM BOOKING

<https://servizistudenti.unisannio.it/pls/self/gissweb.home>

## SYLLABUS

### SYLLABUS MODEL

Topic	Number of hours	Reference text	Type of lesson
Diabetes	10	Study material provided during the course	Frontal
Obesity	10	Study material provided during the course	Frontal

Hyperthension	10	Study material provided during the course	Frontal
Malignant Hyperthermia and Congenital myopathies	3	<p>Malignant hyperthermia: a review. Rosenberg H, Pollock N, Schiemann A, Bulger T, Stowell K. Orphanet J Rare Dis. 2015 Aug 4;10:93. doi: 10.1186/s13023-015-0310-1. Review.</p> <p>Core myopathies and malignant hyperthermia susceptibility: a review. Brislin RP, Theroux MC. Paediatr Anaesth. 2013 Sep;23(9):834-41. doi: 10.1111/pan.12175. Epub 2013 Apr 25.</p> <p>Core myopathies. Jungbluth H, Sewry CA, Muntoni F. Semin Pediatr Neurol. 2011 Dec;18(4):239-49. doi: 10.1016/j.spen.2011.10.005.</p>	Frontal
Long-QT	3	Cardiopatie congenite dell'adulto. R. Calabrò, L. Daliento, B. Sarubbi. Piccin, 2008	Frontal
Patch clamp	3	<p>Ion Channels of Excitable Membranes (3rd Edition). B. Hille. Sinauer Associates Inc, 2001.</p> <p>Patch Clamping: An Introductory Guide to Patch Clamp Electrophysiology. A. Molleman. John Wiley and Sons, LTD, 2003.</p>	Frontal and workshop
Centrifugation e fractionation	4	Biochimica e biologia molecolare: Principi e tecniche. Wilson Keith-	Frontal

techniques		Walker John. Raffaello Cortina Editore, 2006.	
Electrophoretic techniques	4	Biochimica e biologia molecorare: Principi e tecniche. Wilson Keith-Walker John. Raffaello Cortina Editore, 2006.	Frontal
Chromatographic techniques	4	Biochimica e biologia molecorare: Principi e tecniche. Wilson Keith-Walker John. Raffaello Cortina Editore, 2006.	Frontal
Fluorimetric, spectrophotometric, and immunofluorescence techniques	6	Biochimica e biologia molecorare: Principi e tecniche. Wilson Keith-Walker John. Raffaello Cortina Editore, 2006.	Frontal
Procedures for nucleic acid and protein extraction and analysis	4	Materiale didattico distribuito durante il corso	Frontal
FISH e CGH-Array	3	Fetal Medicine: Basic Science and Clinical Practice 2nd edition. Edited by Charles H. Rodeck and Martin J. Whittle. London: Churchill Livingstone, 2008.	Frontal
PCR	3	Fetal Medicine: Basic Science and Clinical Practice 2nd edition. Edited by Charles H. Rodeck and Martin J. Whittle. London: Churchill Livingstone, 2008  Materiale didattico distribuito durante il corso	Frontal
MLPA	3	Fetal Medicine: Basic Science and Clinical Practice 2nd edition. Edited by Charles H. Rodeck and Martin J. Whittle. London: Churchill Livingstone, 2008	Frontal
QF-PCR	3	Fetal Medicine: Basic Science and	Frontal

		Clinical Practice 2nd edition. Edited by Charles H. Rodeck and Martin J. Whittle. London: Churchill Livingstone, 2008	
DNA sequencing technologies	3	DNA sequencing technologies: 2006-2016. Mardis ER. Nat Protoc. 2017 Feb;12(2):213-218. doi: 10.1038/nprot.2016.182. Epub 2017 Jan 5.	Frontal
Clinical interpretation of laboratory tests	4	Medicina di laboratorio e diagnostica genetica. L. Sacchetti, P. Cavalcanti, G. Fortunato. Idelson-Gnocchi, 2007.  Widmann - Interpretazione clinica degli esami di laboratorio 11/ed . Ronald A. Sacher, Richard A. McPherson. McGraw- Hill, 2001.	Frontal