



UNIVERSITÀ DEGLI STUDI DI PALERMO

DEPARTMENT	Biomedicina, Neuroscienze e Diagnostica avanzata		
ACADEMIC YEAR	2017/2018		
BACHELOR'S DEGREE (BSC)	BIOMEDICAL LABORATORY TECHNIQUES		
INTEGRATED COURSE	CLINICAL BIOCHEMISTRY AND CLINICAL MOLECULAR BIOLOGY - INTEGRATED COURSE		
CODE	01557		
MODULES	Yes		
NUMBER OF MODULES	3		
SCIENTIFIC SECTOR(S)	BIO/12, MED/46		
HEAD PROFESSOR(S)	BAZAN VIVIANA	Professore Associato	Univ. di PALERMO
OTHER PROFESSOR(S)	BELLIA CHIARA	Professore Associato	Univ. di PALERMO
	AGNELLO LUISA	Professore Associato	Univ. di PALERMO
	BAZAN VIVIANA	Professore Associato	Univ. di PALERMO
CREDITS	12		
PROPAEDEUTICAL SUBJECTS			
MUTUALIZATION			
YEAR	1		
TERM (SEMESTER)	2° semester		
ATTENDANCE	Mandatory		
EVALUATION	Out of 30		
TEACHER OFFICE HOURS	AGNELLO LUISA Monday 10:00 12:00 Sezione di Biochimica Clinica, Medicina Molecolare Clinica e Medicina di Laboratorio, Dipartimento BIND, Via del Vespro 129 Wednesday 10:00 12:00 Sezione di Biochimica Clinica, Medicina Molecolare Clinica e Medicina di Laboratorio, Dipartimento BIND, Via del Vespro 129 BAZAN VIVIANA Tuesday 12:00 13:30 Dipartimento di Oncologia, piano terra BELLIA CHIARA Monday 12:00 14:00 Sezione di Biochimica Clinica, Medicina Molecolare Clinica e Medicina di Laboratorio, Dipartimento di Biomedicina, Neuroscienze e Diagnostica Avanzata (BiND), Via del Vespro 129 Tuesday 12:00 14:00 Sezione di Biochimica Clinica, Medicina Molecolare Clinica e Medicina di Laboratorio, Dipartimento di Biomedicina, Neuroscienze e Diagnostica Avanzata (BiND), Via del Vespro 129		

PREREQUISITES	As with all the teachings of the first year of the course of study is required the possession of knowledge and skills defined annually by the ministerial decree concerning the tests for admission to degree courses planned at national level. Basic knowledge of the areas of physics, chemistry and biology and basic laboratory still represent essential points for the use of the training provided by the Integrated course of Clinical Biochemistry
LEARNING OUTCOMES	Targets of the Biochemistry and Clinical Molecular Biology Integrated course are to acquire the basic knowledge to critically evaluate the biochemical data at the in relation to human disease. In particular: • knowledge of the main laboratory tests and the cellular, molecular and pathophysiological basis which are prerequisites and foundation. • critical interpretation of laboratory tests results with regard to analytical and biological variation; reliability of diagnostic laboratory tests, examples of correct forms of laboratory reports. • basic concepts on the most relevant methodologies used in Clinical Biochemistry and their limitations. • appropriate use of laboratory tests in screening, staging and treatment of the disease. • correct interpretation of laboratory tests results and their critical correlation with molecular and cellular events induced by the disease. Adoption of guidelines and flowchart based on "Evidence Based Medicine". Be able to relate to colleagues and health operating in laboratory to understand and synthesize relevant information about all the problems, understanding their content and devising and agreeing on how to study. Implement self-protection measures in the collection and handling of biological samples.
ASSESSMENT METHODS	The learning assessment consists in an oral examination. The oral examination consists in a conversation, in order to check the competences and the knowledge required by the course. The questions will tend to test the learning achieved by assessing a) the acquisition of knowledge; b) the processing capacity, c) the possession of adequate presentation capacity. The sufficiency threshold will be reached if the student shows knowledge and understanding of the issues at least in broad outline, and has sufficient application skills; he must also have presentation and argumentative skills allowing the transmission of his knowledge to the examiner. Below this threshold, the examination will be insufficient. The more the candidate will be able to interact with the examiner with his argumentative and presentation skills, and the more his knowledge and application capabilities will go into detail on the subjects under evaluation, the more the judgement will be positive. The evaluation is expressed using a 30-point scale. ECTS grades: A – A+ Excellent (30-30 cum laude) - Grade descriptors : 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. ECTS grade : B Very good (27-29) - Grade descriptors: 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. ECTS grade: C Good (24-26)- Grade descriptors: Good knowledge of teaching contents and good language control; the students should be able to apply their knowledge to solve problems of medium complexity ECTS grade: D Satisfactory (21-23)- Grade descriptors: 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. ECTS grade: E Sufficient (18-20) - Grade descriptors: Minimum teaching content knowledge, often limited to the main topic; modest ability to use the subject specific language and independently apply the acquired knowledge. ECTS grade: F Fail (1-17) - Grade descriptors: Lack of an acceptable knowledge of the main teaching content knowledge; very little or no ability to use the specific subject language and apply independently the acquired knowledge. Exam failed. A sufficient grade in each module is necessary to pass the final exam. If the mark is insufficient in any of the modules, the student cannot pass the final examination.
TEACHING METHODS	The didactic activity takes place through lectures.

MODULE
MEDICINE AND LABORATORY TECHNICAL SCIENCES

Prof.ssa CHIARA BELLIA

SUGGESTED BIBLIOGRAPHY

G. Federici, P. Cipriani, C. Cortese, A. Fusco, P. Ialongo e C. Milani: Medicina di Laboratorio. 3a Edizione, McGraw-Hill, 2009.

L. Spandrio. Principi e Tecniche di Chimica Clinica. Piccin, 2000

Stoppini M, Bellotti V. Biochimica Applicata. Edises.

De Marco C, Cini C. Principi di metodologia biochimica. Piccin.

AMBIT	10341-Scienze e tecniche di laboratorio biomedico
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INDIVIDUAL STUDY (Hrs)	45
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COURSE ACTIVITY (Hrs)	30
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EDUCATIONAL OBJECTIVES OF THE MODULE

Main targets of the course are: critical interpretation of medical laboratory tests in relation to analytical and biological variation; evaluation of the clinical performances of laboratory tests; how to report laboratory tests correctly; fully description of the techniques used in medical laboratories.

SYLLABUS

Hrs	Frontal teaching
2	Intro to Laboratory Medicine: handling and storage of biological samples; pre-analytical issues. Biological and analytical variation, accuracy and precision, analytical and clinical sensibility and specificity, laboratory errors, reference values, quality controls.
2	Separating techniques: dialysis, ultra-filtration, gel-filtration, analytical and preparative centrifugation, types of centrifuge, ultracentrifugation and main applications in clinical biochemistry.
2	UV-visible spectroscopy: electromagnetic waves and spectrum, electronic transitions, absorption and emission; main components of a spectrophotometer, Lambert and Beer's equation, calibration line, direct analysis, analysis with absorption increase or decrease, kinetic analysis. Main application fo UV-visible spectrometry: serum creatinine, glucose, total cholesterol, LDL and HDL cholesterol, triglycerides, direct and indirect bilirubinemia, AST, ALT.
1	Fluorometry: basic concepts, emission phenomena, main components of a fluorometer, quantitative analysis, main application in clinical biochemistry.
2	Electrophoresis: definition, basic concepts, main components of an electrophoresis systems. Protein electrophoresis: SDS-PAGE, serum protein electrophoresis. Capillary electrophoresis: main characteristics, components, electroendosmotic flow.
2	Chromatography: basic concepts; partition coefficients; theoretical plates end column efficiency; isocratic and gradient elution; characteristics of a chromatographic peak. Paper and thin layer chromatography; adsorption, partition, ionic exchange and molecular exclusion chromatography. Main detectors. Qualitative and quantitative analysis.
2	Isolation of nucleic acid: phenol-clorophorm purification, salting-out, chromatographic isolation. Electrophoresis of nucleic acid.
2	Amplification of nucleic acids: basic concepts; clinical applications; genotyping; RT-PCR; real-time PCR: chemistry, quantification of nucleic acids in biological samples, allelic discrimination. Restriction enzymes, RFLP and SNPs. Hybridization of nucleic acids: reverse dot blot. Application of capillary electrophoresis in molecular biology: Sanger sequencing and identification of mutations. Clinical molecular biology: hereditary thrombophilia, farmacogenetics of oral anticoagulant.

MODULE CLINICAL BIOCHEMISTRY AND CLINICAL MOLECULAR BIOLOGY

Prof.ssa LUISA AGNELLO

SUGGESTED BIBLIOGRAPHY

L. Sacchetti, P. Cavalcanti, G. Fortunato, L. Pastore, F. Rossano, D. Salvatore e F. Scopacasa: Medicina di Laboratorio e Diagnostica Genetica. Idelson-Gnocchi Editori, 2007.
G. Federici, P. Cipriani, C. Cortese, A. Fusco, P. Ialongo e C. Milani: Medicina di Laboratorio. 3a Edizione, McGraw-Hill, 2009.
I. Antonozzi, E. Gulletta. Medicina di Laboratorio Logica & Patologia Clinica. Piccin , 2012
L. Spandrio: Biochimica Clinica Speciale. Piccin Editore, 2006.
W.J. Marshall e S.K. Bangert: Biochimica in Medicina Clinica. Mc Graw-Hill, 1997.
B. Barbiroli, F. Filadoro, C. Franzini, L. Sacchetti, e F. Salvatore: Medicina di Laboratorio. UTET, 1996.

AMBIT	10338-Scienze biomediche
INDIVIDUAL STUDY (Hrs)	90
COURSE ACTIVITY (Hrs)	60

EDUCATIONAL OBJECTIVES OF THE MODULE

Acquisition of basic and applied concepts of clinical biochemistry in order to evaluate methods and clinical applications of laboratory tests critically. Specific aim of the course is to be able to identify the correct tests in relation to clinical conditions of the patients and to be able to understand them in relation to the diagnosis.

Particularly, the students must:

- know the main laboratory tests and the biological, molecular and pathological basis for their use in medicine;
 - use laboratory tests in screening, diagnosis, prognosis and therapy of diseases correctly;
 - understand laboratory tests results and connect them to the molecular and cellular aspect of the pathologic state
- Knowledge of the molecular mechanisms underlying the main clinical syndromes arising from genetic alteration.

SYLLABUS

Hrs	Frontal teaching
2	Laboratory tests: definition, types, requests.
4	Pre-analytical phase: preparation of the patient, collection of biological samples, processing and identification of biological samples. General laboratory techniques with descriptions of the principles. Post-analytical phase: data collection, calculation, automatic processing. Analytical variation, analytical error, quality control systems. Intra-individual and inter-individual biological variation, reference values, nomenclature and reporting of laboratory tests. Clinical sensitivity and specificity, predictive laboratory tests. Methodological approaches in clinical biochemistry. The main analytical techniques, automation in clinical biochemistry.
2	Biotechnologies and Clinical Biochemistry
2	Enzymes: tissue and organ profiles. Acid and alcalin phosphatase, aldolase, aminotrasferase, LDH, gammaGT, cholinesterase, amylase, lipase, protease, G-6-PD, piruvate kinase, lysozyme.
4	Metabolism of glucose: Diabetes: biochemistry of DMT1 and DMT2; biochemistry of diabetes complications; clinical biochemistry diagnostics (serum glucose, urine glucose, insulin, C-peptide, glucagon, cortisol, GH, glicated proteins and their significance, OGTT, ketone bodies in serum and urine, lactic acid, piruvic acid, dyslipidemia in diabetic patients, urine test in diabetic patients. Hypoglicemia: clinical biochemistry
2	Metabolism of lipids: Fatty acids, cholesterol, HDL-cholesterol, LDL-cholesterol, tryglicerides, phospholipids. Lipoprotein. Dyslipidemia. Hypercholesterolemia. Atherosclerosis and cardiovascular diseases.
1	Myocardial infarction: risk factors, biochemical modification in the necrotic area; clinical enzymes, new markers of myocardial infarction.
4	Metabolism of proteins: Plasma proteins. Electrophoresis of plasma proteins: interpretation of results. Clinical biochemistry of kidney disease. Hyperammonemia. Clinical biochemistry of liver failure. Urea metabolism: clinical biochemistry of enzymatic deficiency
3	Clinical Biochemistry of jaundice. Clinical biochemistry of exocrine pancreas. Clinical biochemistry of healthy and pathologic renal function. Clinical Biochemistry of heme metabolism.
2	Endocrine system: Clinical biochemistry of hypothalamus-pituitary axis: GH and growth, prolactin and amenorrhea, GnRH, LH, FSH and reproduction, ACTH, TSH e functional tests. Clinical biochemistry of endocrine pancreas: insulin and glucagon, somatostatin. Clinical biochemistry of gastrointestinal hormones. Clinical biochemistry of adrenal cortex hyperplasia and hypofunction.
2	Clinical biochemistry of hyperthyroidism, thyreotoxicosis, hypothyroidism. Clinical biochemistry of testis, ovary and sex differentiation.
2	Clinical biochemistry of rickets. Clinical biochemistry of osteoporosis and osteomalacia. Calcium homeostasis. Hormone receptors and their importance in clinical biochemistry.

MODULE TECHNICAL CLINICAL MOLECULAR MEDICINE SCIENCE

Prof.ssa VIVIANA BAZAN

SUGGESTED BIBLIOGRAPHY

-Robbins: Le basi patologiche delle malattie, Vol. I. Ed. Piccin
 -The Molecular Basis of Cancer - J. Mendelsohn, P. Howley, M. Israel, J. Gray, C. Thompson - Saunders, Elsevier
 -Genome Analysis: Current Procedures and Applications – Poptsova, Caister Academic
 -Next-generation Sequencing: Current Technologies and Applications – Xu, Caister Academic

Il docente si riserva di estrapolare i contenuti dai numerosi testi di riferimento in lingua inglese relativi alla Molecular Pathology. Pubblicazioni scientifiche su riviste internazionali, dispense e testi delle slides verranno fornite agli studenti durante il corso.

AMBIT	10341-Scienze e tecniche di laboratorio biomedico
INDIVIDUAL STUDY (Hrs)	45
COURSE ACTIVITY (Hrs)	30

EDUCATIONAL OBJECTIVES OF THE MODULE

The course will explore structure, properties and functions of nucleic acids, in particular mutations and their consequences on the function of proteins in relation to human diseases, the methods for the identification of deficit genes, the clinical aspects of Molecular Biology in relation to neoplastic disease.

Course objectives:

- to provide the basic knowledge on the development of benign and malignant neoplasms
- to identify the main family-hereditary syndromes
- to provide students with the knowledge of modern diagnostic techniques both molecular and cellular for identifying a genetic disease
- to acquire the principles for the proper performance of molecular pathology techniques on histological and cytological samples
- to explore the area of molecular pathology techniques to integrate the anatomic pathological diagnosis.

SYLLABUS

Hrs	Frontal teaching
15	Neoplastic diseases - definition, classification and epidemiology - Biology of tumor growth: benign and malignant tumors - molecular basis of cancer - essential alterations for malignant transformation - The cell cycle - oncogenes, tumor suppressor genes and signal transduction - Instability of gene expression and mechanisms of DNA repair - Epigenetics and cancer - Angiogenesis, invasiveness and metastasis - Immunity of tumor
6	Genetic predisposition - autosomal dominant disease (retinoblastoma, FAP, Li Fraumeni, MEN-1 and MEN-2, HNPCC) - familial syndromes (HBOC, Lynch I and II, familial melanoma)
21	Pathology and molecular diagnostics - "Cancer genomics": - structural aberrations - Copy Number Variation - somatic and germline mutations - "Transcriptome Analysis": - Analysis of microarray - microRNA and cancer Techniques for molecular diagnosis: - Flow Cytometry - Proteomics - Techniques DNA extraction and RNA - PCR and its applications - direct automated sequencing and pyrosequencing - the Real-Time PCR and its applications - the future of sequencing: Next Generation sequencing - the era of digital PCR
3	Bases of personalized medicine - What are the "target therapies" - Definition of pharmacogenetics - Therapies against cancer - primary and acquired resistance - Monoclonal antibodies in cancer therapy - immunotherapy in the treatment of tumors
12	Molecular pathogenesis of the major cancers. Hematological malignancies myeloid and lymphoid neoplasms. From the clinic to the laboratory: molecular diagnosis and evaluation of minimal residual disease in CML, targeted treatment of CML, Lung cancer, EGFR gene mutation analysis, analysis of ALK rearrangements and ROS1, amplification analysis of c-MET, lung cancer targeted treatment, neoplasms of the gastrointestinal tract, small intestine and colorectal tumors, gastrointestinal stromal tumors (GIST), Hepatocellular carcinoma: molecular prognostic and predictive factors, mutational analysis of the KRAS gene, NRAS, BRAF, PI3K, c-KIT, PDGFR, targeted treatment of mCRC and GIST, melanoma, mutational analysis of the BRAF gene, and GNA11 GNAQ, targeted treatment in melanoma
3	animal models and cell culture for the study of the molecular pathogenesis of tumors - Long non-coding RNAs and Cancer - Hepatocellular carcinoma: molecular prognostic and predictive factors - The liquid biopsy in the management of cancer patient
Hrs	Practice
5	Preparation of the biological sample - Extraction of nucleic acids (DNA, RNA) from blood and tissue embedded in paraffin - Electrophoresis on agarose gel
5	Setting up a PCR reaction - Establishment of a sequencing reaction - Capillary Electrophoresis (direct automated sequencer) - electropherogram analysis