



UNIVERSITÀ DEGLI STUDI DI PALERMO

DEPARTMENT	Biomedicina, Neuroscienze e Diagnostica avanzata		
ACADEMIC YEAR	2016/2017		
MASTER'S DEGREE (MSC)	MEDICAL BIOTECHNOLOGIES AND MOLECULAR MEDICINE		
INTEGRATED COURSE	MEDICAL AND FORENSIC GENETICS, MOLECULAR EPIDEMIOLOGY AND BIOINFORMATICS - INTEGRATED COURSE		
CODE	17134		
MODULES	Yes		
NUMBER OF MODULES	3		
SCIENTIFIC SECTOR(S)	ING-INF/05, BIO/13, MED/42		
HEAD PROFESSOR(S)	SEIDITA GREGORIO	Ricercatore	Univ. di PALERMO
OTHER PROFESSOR(S)	TRAMUTO FABIO	Professore Associato	Univ. di PALERMO
	VITABILE SALVATORE	Professore Ordinario	Univ. di PALERMO
	SEIDITA GREGORIO	Ricercatore	Univ. di PALERMO
CREDITS	12		
PROPAEDEUTICAL SUBJECTS			
MUTUALIZATION			
YEAR	1		
TERM (SEMESTER)	1° semester		
ATTENDANCE	Mandatory		
EVALUATION	Out of 30		
TEACHER OFFICE HOURS	<p>SEIDITA GREGORIO</p> <p>Tuesday 14:30 16:30 Sezione di Biologia e Genetica via divisi, 83 (Dipartimento di Biopatologia e Biotecnologie Mediche)</p> <p>Wednesday 11:00 13:00 Sezione di Biologia e Genetica via divisi, 83 (Dipartimento di BiND)</p> <p>TRAMUTO FABIO</p> <p>Monday 14:00 16:00 Dipartimento di Promozione della Salute, Materno-Infantile, Medicina Interna e Specialistica di Eccellenza "G. D'Alessandro" AOUP "P. Giaccone" Via del Vespro, 133 Piano terra</p> <p>Wednesday 14:00 16:00 Dipartimento di Promozione della Salute, Materno-Infantile, Medicina Interna e Specialistica di Eccellenza "G. D'Alessandro" AOUP "P. Giaccone" Via del Vespro, 133 Piano terra</p> <p>Friday 14:00 16:00 Dipartimento di Promozione della Salute, Materno-Infantile, Medicina Interna e Specialistica di Eccellenza "G. D'Alessandro" AOUP "P. Giaccone" Via del Vespro, 133 Piano terra</p> <p>VITABILE SALVATORE</p> <p>Monday 16:30 18:30 Piattaforma Microsoft Teams, Dipartimento di Biomedicina, Neuroscienze e Diagnostica avanzata, Plesso di Radiologia – 1° piano, Stanza n. 108.</p>		

PREREQUISITES	The student should have the basic knowledge of biology and genetics. It must also know the basic fundamentals of biostatistics and show ability to run the most important operating systems (i.e. iOS, Microsoft Windows).
LEARNING OUTCOMES	<p>Knowledge and understanding At the end of the course students will be able to develop, analyze and solve problems related personal identification, detection, or the exclusion of family relationships using appropriate techniques of molecular biology; analyze and apply the methods available to study the genetic variability and its relationship with human diseases; experimental use genetic information in a statistical sense and develop comparison skills with international databases; evaluation capacity of a biological evidences. Understanding of the ethical and legal problems associated with the personal identification procedures; At the end of the course students will be expected to have acquired the needed skills for critically knowing and using informatics tools and programs for genomic and proteomic data analysis. The above results will be achieved through the knowledge of both biological database organization models and search strategies, and pairwise and multiple sequence alignments.</p> <p>Graduates will demonstrate knowledge of the molecular epidemiology applications in different health situations, knowing how to interpret and evaluate the results. Recognize the nature of biomedical data in order to be able to properly apply the most appropriate methods for analyzing statistics, and describe and properly detect the presence or absence of relationships between variables. To know and critically use computer tools applied to the analysis of experimental genetic information and epidemiological items. Develop the ability to properly interpret the report obtained after comparison of experimental data with international databases. Organization and access to biological databases. Techniques of alignment, simple and multiple, of gene and protein sequences. Knowing some of the most common data analysis automated techniques, developed from an analysis of appropriate biological principles.</p> <p>Applying knowledge and understanding Students will spend the knowledge acquired during the course in Human Molecular Genetics directly into work (in research or analysis laboratories, public or private). They will know how to apply the new techniques for the investigation of genetic variability, considering also bioethical implications this analysis raise. Students should be able to access and use the most common on-line biological databases as well as to use the most common on-line pairwise and multiple sequence alignments tools and programs.</p> <p>Graduates must also demonstrate:</p> <ul style="list-style-type: none"> • competence in the design and organization of epidemiological studies appropriate to the various health needs; • competence in the application and interpretation of measures of statistical description, of association and of the main descriptive graphics in molecular epidemiology; • Knowledge of laboratory techniques and computer aimed at assessing the genetic characteristics and potential epidemiological pattern. Use and search in online biological databases. • Use and do research of gene and protein sequences. • Use simple data analysis tools. <p>Making judgments Be able to independently meet the professional issues related to knowledge of the course. Students will be able to evaluate in a rational and autonomous basic knowledge provided by the course and will be able to address issues of Human Molecular Genetics through a scientific approach. Autonomy, the application and interpretation of the measures of the main descriptive statistics and graphs in molecular epidemiology. Students will be expected to have acquired autonomous and critic judgment skills for evaluating and analysing significance and limits of the programs and tools produced results.</p> <p>Communicative skills Ability to communicate and disseminate the knowledge acquired during the course in their professional responsibilities. Students will gain a communicative methodology of scientific / experimental in the survey genetics and genetic-forensic and data analysis. They will know how to manage the genetic investigations with particular decision-making autonomy to guarantee confidentiality. Gain ability to evaluate the results. Students should be able to clearly report on the topics dealt with during the course in its own professional domain. Following a strong scientific approach,</p>

	<p>the use of correct technical and computer science based-terms is also required.</p> <p>Learning skills Application of knowledge of the course in order to improve the overall approach to their health care professional. Ability to update by consulting their own scientific publications of the Molecular Genetics sector with particular attention to the Human and network resources. Mastery of bio-IT methods for the purpose of the organization, construction and access to databases, in particular those genomic. This course aims at developing students' awareness of the acquired competences for self-directed learning of new bioinformatics contents and methods. The development of students' awareness for scientific publications and for new tool and program position papers will be a further target.</p>
ASSESSMENT METHODS	<p>The Learning evaluation is done by oral examination. The outcome of the evaluation is expressed by a vote ranging from 1-30. The oral exam consists of a conversation usually lasting about 20-30 minutes in order to check the disciplinary knowledge of the curriculum contents. In the assessment of learning it is taking into account the participation and evaluation obtained of test carried out during the course.</p> <p>The following table shows the learning evaluation scheme with the ECTS grades, the equivalent rating of thirty and their significance.</p> <p>A – A+ / Excellent 30/30 cum laude 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.</p> <p>B / Very good 27-29/30 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.</p> <p>C / Good 24- 26/30 Good knowledge of teaching contents and good language control; the students should be able to apply their knowledge to solve problems of medium complexity</p> <p>D / Satisfactory 21-23/30 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.</p> <p>E / Sufficient 18-20 Minimum teaching content knowledge, often limited to the main topic; modest ability to use the subject specific language and independently apply the acquired knowledge.</p> <p>F / Fail 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.</p> <p>A preliminary and optional examination is carried out during the lessons. This is a test with 6 open questions to be performed in 90 minutes. The exam questions will focus on the lessons contents until then.</p>
TEACHING METHODS	<p>Lessons in the classroom; practical lessons and exercises in classroom and bioinformatic classroom.</p>

MODULE MOLECULAR EPIDEMIOLOGY

Prof. FABIO TRAMUTO

SUGGESTED BIBLIOGRAPHY

Rothman Kenneth J, Epidemiologia, Idelson-Gnocchi.
 Salemi M, The Phylogenetic Handbook ..., Cambridge University Press
 Hall Barry G, Phylogenetic Trees Made Easy ..., Sinauer Associates Inc

AMBIT	20885-Attività formative affini o integrative
INDIVIDUAL STUDY (Hrs)	42
COURSE ACTIVITY (Hrs)	33

EDUCATIONAL OBJECTIVES OF THE MODULE

The purposes of this module are:
 a) to outline the fundamentals of epidemiological data analysis and interpretation.
 b) to discuss the theoretical aspects of technics in molecular epidemiology.

SYLLABUS

Hrs	Frontal teaching
4	Elements of biostatistics Epidemiological studies: cross-sectional studies, case-control studies, cohort studies
2	Epidemiological data sources and sampling strategies. Epidemiological bias and confounding factors.
2	Molecular methods applied to epidemiology. Molecular epidemiology of infectious and chronic diseases
4	Nucleic acids searches in international databases. Multiple alignment: algorithm
4	Fundamentals of molecular evolution and models. Phylogenetics: trees and algorithms. Genetic recombination analysis
Hrs	Practice
4	Databases for genetic information
4	Softwares for the analysis of genetic sequences
4	Softwares for the analysis of genetic evolution
5	Softwares for inferring phylogenies (evolutionary trees)

MODULE MEDICAL AND FORENSIC GENETICS

Prof. GREGORIO SEIDITA

SUGGESTED BIBLIOGRAPHY

- Strachan & Read "Genetica Molecolare Umana . Garland Sciences 4 Edizione.
- Butler J.M. "Fundamental of Forensic DNA typing". Academic Press (2010).
- Dispense fornite dal docente durante il corso in file pdf (articoli e metodologie).

AMBIT	50644-Discipline biotecnologiche comuni
INDIVIDUAL STUDY (Hrs)	75
COURSE ACTIVITY (Hrs)	75

EDUCATIONAL OBJECTIVES OF THE MODULE

Medical and Forensic Genetics module aims to provide students with:

- the theoretical basis for understanding problems related to the analysis of genetic variation. It provides guidelines for applying molecular genetics techniques;
- Detailed knowledge of the main techniques of molecular biology and molecular genetics applied in the biomedical field;
- Analysis of the main polymorphisms of forensic interest and the organization of a forensic genetics laboratory.

SYLLABUS

Hrs	Frontal teaching
4	Genome structure and function. Genetic variability and measurement of variability. Organization of the Human Genome.
4	Gene and chromosomal mutations. Main techniques for the study of human genetic variations.
4	The main Genetic tests on the individual and their meaning. The distribution of genes in families and populations, mode of transmission of characters, Hardy Weinberg Equilibrium.
4	Counseling in medical genetics. Biological samples of forensic interest, evidence sampling techniques, evidences conservation and characterization. Extraction of DNA from biological samples of forensic interest. Quantization and DNA amplification.
4	Human STR loci: Biology and Genetics. STR forensic features. Capillary electrophoresis. Quality control and validation of methods procedures. The DNA database. Missing persons and mass disaster.
4	DNA Low Copy Number samples. Y chromosome STR markers. Analysis of Mitochondrial genome. Analysis of chromosome markers X. Genetic aspects of the forensic interest DNA polymorphisms (mutation rate; Hardy Weinberg theorem and its applications, Investigation and personal identification, Paternity Investigation).
Hrs	Practice
6	Pubmed and MyNCBI. Databases of public domain DNA. Management software and analysis of nucleotide and amino acid sequences. Analysis of the main information of a biological sequence (BLAST, CLUSTAL, ...)
6	Software for the study and design of synthetic DNA sequences.
6	Genetic counseling. Family trees and Risk-of-recurrence calculation. Tutorials on the Hardy Weinberg equation.
3	Technical investigation of genetic variation
6	The judicial inspection for sampling biological evidences. Predictive and related technical tests. DNA extraction from biological traces in Forensic Genetics and Molecular Diagnostics.
6	DNA extracted genomic characterization: quantity and quality (Real time PCR). Amplification of genomic DNA.
6	The Italian DNA Data Bank. Procedures for forensic genetics laboratories accreditation. Biostatistic in forensic genetics. The paternity investigations. Routines and exceptions.
6	Random Match Probability. limits and boundaries of Forensic Genetics.
6	Genetic characterization technologies for Low Copy Number DNA samples (low template DNA).

MODULE
BIOINFORMATICS APPLIED TO GENOMICS AND PROTEOMICS

Prof. SALVATORE VITABILE

SUGGESTED BIBLIOGRAPHY

Introduzione alla Bioinformatica - Valle Giorgio, Helmer Citterich Manuela, Attimonelli Marcella, Pesole Graziano - Zanichelli - ISBN 8808-07967-8

Introduzione alla Bioinformatica - Lesk Arthur M. - Mc Graw Hill - ISBN 88-386-6190-1

AMBIT	20885-Attività formative affini o integrative
INDIVIDUAL STUDY (Hrs)	42
COURSE ACTIVITY (Hrs)	33

EDUCATIONAL OBJECTIVES OF THE MODULE

In this course, students will learn fundamental concepts and methods in bioinformatics, a field at the intersection of biology and computing. It surveys a wide range of topics including biological databases organization, structure, and search strategies; pairwise and multiple sequence alignment principles; the most common on-line alignment search tools; and microarrays image analysis. The final part of the course will be devoted to the introduction of one of the most used biological inspired model: the Artificial Neural Networks.

SYLLABUS

Hrs	Frontal teaching
4	Course introduction; Information representation and coding; Files and images; Databases.
2	Biological databases organization and structure.
2	Pairwise sequence alignment.
2	BLAST: Basic Local Alignment Search Tool
2	Multiple sequence alignment; Clustal Omega.
2	Microarray image analysis.
2	An introduction to Artificial Neural Networks.
Hrs	Practice
6	Search strategies in biological databases.
6	Sequence alignment.
5	Neural Networks.