UNIVERSITÀ DEGLI STUDI DI PALERMO

DEPARTMENT	Scienze e Tecnologie Biologiche, Chimiche e Farmaceutiche		
ACADEMIC YEAR	2022/2023		
MASTER'S DEGREE (MSC)	MOLECULAR AND HEALTH BIOLOGY		
INTEGRATED COURSE	GENETIC AND CYTOGENETIC METHODOLOGIES		
CODE	21261		
MODULES	Yes		
NUMBER OF MODULES	2		
SCIENTIFIC SECTOR(S)	BIO/18		
HEAD PROFESSOR(S)	BARRA VIVIANA	Ricercatore a tempo determinato	Univ. di PALERMO
OTHER PROFESSOR(S)	BARRA VIVIANA	Ricercatore a tempo determinato	Univ. di PALERMO
CREDITS	6		
PROPAEDEUTICAL SUBJECTS			
MUTUALIZATION			
YEAR	1		
TERM (SEMESTER)	2° semester		
ATTENDANCE	Mandatory		
EVALUATION	Out of 30		
TEACHER OFFICE HOURS	**BARRA VIVIANA** Tuesday 10:00 11:00 Studio Pt 84, Dip STEBICEF, viale delle Scienze, Ed. 16, piano -1 o Microsoft Teams		

DOCENTE: Prof.ssa VIVIANA BARRA

PREREQUISITES	Basic knowledge of Genetics.
LEARNING OUTCOMES	Knowledge and understanding: acquisition of terminologies and methodological elements at the base of molecular genetic approaches for the subsequent understanding of cellular pathways under genetic-epigenetic control. Ability to apply knowledge and understanding: to be able to understand the "rationale" of experiments using genetics, cytogenetics and epigenetics approaches. Ability to collect, interpret and process, scientific data derived from the study of scientific papers that use their own genetics, cytogenetic and epigenetic methods. Problem solving abilities. Making judgments: to be able to integrate knowledge of the experimental data and synthesis presented in scientific papers. Communication skills: to be able to work in a team, ability to present scientific arguments and their conclusion orally and in writing to specialist and non-specialist audiences. Learning skills: ability to learn autonomously the technical and methodological approach in molecular genetic research by making use of their knowledge or of scientific sources.
ASSESSMENT METHODS	The evaluation will be made by a final oral exam. The final will be comprehensive and will cover material from the entire course. The assessment will take into account the level of knowledge of the topics treated during the course and skills reasoning demonstrated during the examination. In detail: Insufficient- the student does not possess the basic knowledge of Genetics' topics. 18-21- limited knowledge of basic subjects associated with fragmentary and incomplete exposure. 22-25- mastery of only basic issues associated with discrete scientific language abilities. 26-29- more than good grasp of the topics covered in the course, full of scientific language 30-30 cum laude- excellent mastery and ability to present the arguments of both modules (1&2), demonstrating excellent reasoning skills, good mastery of scientific language.
TEACHING METHODS	Class lectures on all topics of the course.

MODULE
GENETIC METHODOLOGIES

Prof.ssa VIVIANA BARRA

SUGGESTED BIBLIOGRAPHY	
Il materiale didattico necessario sarà fornito agli studenti sotto forma di presentazioni Power point basate su recenti pubblicazioni scientifiche in lingua inglese, riguardanti i sistemi di sorveglianza cellulari dell'aneuploidia e relativi modelli, insieme agli articoli originali in lingua inglese (formato pdf)	
AMBIT	20879-Attività formative affini o integrative
INDIVIDUAL STUDY (Hrs)	51
COURSE ACTIVITY (Hrs)	24
EDUCATIONAL OBJECTIVES OF THE MODULE	
This module is aimed to provide the knowledge of scientific methods/tools underlying the research in cancer molecular genetics. The course will consist of Landmark publications in a variety of model organism systems dealing with the molecular dissection of the spindle assembly checkpoint to understand the rationale and the conclusions. In addition the use of genomic screens, siRNA approaches and molecular cytogenetics tools will be addressed.	

SYLLABUS

Hrs	Frontal teaching
6	Evidences of aneuploidy as an oncogenic or as a tumor suppressor factor. Aneuploidy tolerance. proteotoxic stress as a consequence of aneuploidy. Models
6	Molecular dissection of the checkpoints working in mitosis. Defects of genes of the spindle assembly checkpoint (SAC) and aneuploidy.
10	Methods to study mutations of SAC genes and chromosomal instability. Alterations of centromeric proteins (CENPs) and chromosomal instability. The kinetochore assembly alterations and chromosomal instability. Models to explain aneuploidy occurrence. relationship between CIN and aneuploidy. Tetraploidy as a mean to generate aneuploid cells: models.
2	CRISPR: gene and transcript editing

MODULE
CYTOGENETIC AND EPIGENETIC METHODOLOGIES

Prof.ssa VIVIANA BARRA

SUGGESTED BIBLIOGRAPHY

Tom Strachan, Andrew Read. Genetica Molecolare Umana Ed. Zanichelli, 2021 ISBN: 9788808520302
Il materiale didattico necessario sarà fornito agli studenti durante il corso (presentazioni proiettate a lezione). Le lezioni sono basate su recenti pubblicazioni scientifiche in lingua inglese nell'ambito della citogenetica e dell'epigenetica di malattie umane tra cui il cancro che saranno fornite dal docente.

AMBIT	20879-Attività formative affini o integrative
INDIVIDUAL STUDY (Hrs)	51
COURSE ACTIVITY (Hrs)	24

EDUCATIONAL OBJECTIVES OF THE MODULE

This module will provide the theoretical knowledge of chromosomal structure and of the epigenetics mechanisms, the methodological bases that allow to study human diseases including cancer at the cytogenetic and epigenetic level. The module will prepare students for the analytical and critical reading of the scientific literature.

SYLLABUS

Hrs	Frontal teaching
3	Structure and organization of chromosomes. Euchromatin/heterochromatin, repetitive DNA and transposons, centromere, telomere.
3	Basics of epigenetics at the molecular level (DNA methylation, post-translational modifications of histones and the histone code, chromatin remodelling, writers, readers and erasers). Histone variants: CENP-A and its modifications.
4	Classical and molecular cytogenetic techniques (karyotype, FISH, Multicolor-FISH, fiber-FISH, mBAND). Analysis of pathological alterations. Fanconi Anemia, common fragile sites and chromosome breaks. Karyotypic instability and chromosome translocations in tumors. Centromeric Epialleles.
4	Genomic technologies for cytogenetics. CGH-array and SNP-array. Next Generation Sequencing techniques (NGS): Ion Torrent and Illumina (from the preparation of the library to the sequencing methods). Analysis of pathological alterations. MECP2 in neurological disorders. Circulating tumor cell sequencing.
8	Methods for the analysis of DNA methylation: COBRA, MSP, MeDIP, methylation array, whole genome bisulfite sequencing. Epigenetic alterations in the aetiology and progression of human diseases. ICF syndrome and DNA methylation alterations at centromere. DNA methylation loss and cancer initiation/progression. Chimeric transcripts induced by hypomethylation of LINE-1 in tumors.
2	Genome and epigenome editing. Hints on TALEN system. CRISPR applied to site-specific DNA methylation/demethylation.