# UNIVERSITÀ DEGLI STUDI DI PALERMO

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| **DEPARTMENT** | Scienze e Tecnologie Biologiche, Chimiche e Farmaceutiche | | |
| **ACADEMIC YEAR** | 2016/2017 | | |
| **MASTER'S DEGREE (MSC)** | MOLECULAR AND HEALTH BIOLOGY | | |
| **INTEGRATED COURSE** | MOLECULAR GENETICS AND GENETIC AND CYTOGENETIC METHODOLOGIES - INTEGRATED COURSE | | |
| **CODE** | 16480 | | |
| **MODULES** | Yes | | |
| **NUMBER OF MODULES** | 2 | | |
| **SCIENTIFIC SECTOR(S)** | BIO/18 | | |
| **HEAD PROFESSOR(S)** | DI LEONARDO ALDO | Professore Associato | Univ. di PALERMO |
| **OTHER PROFESSOR(S)** | LENTINI LAURA<br>DI LEONARDO ALDO | Professore Associato<br>Professore Associato | Univ. di PALERMO<br>Univ. di PALERMO |
| **CREDITS** | 12 | | |
| **PROPAEDEUTICAL SUBJECTS** | | | |
| **MUTUALIZATION** | | | |
| **YEAR** | 1 | | |
| **TERM (SEMESTER)** | 2° semester | | |
| **ATTENDANCE** | Mandatory | | |
| **EVALUATION** | Out of 30 | | |
| **TEACHER OFFICE HOURS** | **DI LEONARDO ALDO**<br>Friday 14:30 16:00 Studio docente, dipartimento STEBICEF, viale delle Scienze, Ed.16 piano -1<br>**LENTINI LAURA**<br>Thursday 15:00 17:00 Sede del Consorzio Universitario, corso Vittorio Emanuele, 92, 93100 Caltanissetta<br>Friday 11:00 13:00 Studio docente e Aula Microsoft Teams Dip. STEBICEF, viale delle Scienze, Ed.16 , piano -1. | | |

**DOCENTE:** Prof. ALDO DI LEONARDO

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| **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 control.<br><br>Ability to apply knowledge and understanding: to be able to understand the "rationale" of molecular genetics experiments inherent biological problems. Ability to collect, interpret and process, scientific data derived from the study of scientific papers that use their own methods of molecular genetics. Problem solving abilities.<br><br>Making judgments: the ability to integrate knowledge of the experimental data and synthesis presented in scientific papers. To formulate hypothesis with incomplete data.<br><br>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<br><br>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 and intermediate quiz. 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.<br><br>In detail:<br>Insufficient- the student does not possess the basic knowledge of Genetics' topics.<br>18-21- limited knowledge of basic subjects associated with fragmentary and incomplete exposure.<br>22-25- mastery of only basic issues associated with discrete scientific language abilities.<br>26-29- more than good grasp of the topics covered in the course, full of scientific language<br>30-30 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** | Lectures. |

## MODULE
## GENETIC AND CYTOGENETIC METHODOLOGIES

*Prof. ALDO DI LEONARDO*

**SUGGESTED BIBLIOGRAPHY**

During the course students will be provided with PowerPoint presentation regarding the topics treated and with PDF files of the correlate scientific articles.

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| **AMBIT** | 20879-Attività formative affini o integrative |
| **INDIVIDUAL STUDY (Hrs)** | 102 |
| **COURSE ACTIVITY (Hrs)** | 48 |

**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 |
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| 12 | Evidences of aneuploidy as an oncogenic or as a tumor suppressor factor. Aneuploidy tolerance. proteotoxic stress as a consequence of aneuploidy. Models |
| 16 | Molecular dissection of the checkpoints working in mitosis. Defects of genes of the spindle assembly checkpoint (SAC) and aneuploidy. |
| 16 | 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. epigenetics and aneuploidy |
| 4 | Molecular Cytogenetics techniques : FISH, Chromosome painting, multicolor FISH, SKY, Comparative Genomic Hybridization; CGH array. |

**MODULE**
**MOLECULAR GENETICS**

*Prof.ssa LAURA LENTINI*

| **SUGGESTED BIBLIOGRAPHY** | |
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| Tom Strachan& Andrew Read: Genetica molecolare umana, Zanchelli 2012.<br>Durante il corso vengono forniti agli studenti i necessari strumenti per lo studio della disciplina; in particolare vengono fornite dal docente le presentazioni in Power Point, files multimediali e articoli scientifici pdf. | |
| **AMBIT** | 50507-Discipline del settore biomolecolare |
| **INDIVIDUAL STUDY (Hrs)** | 102 |
| **COURSE ACTIVITY (Hrs)** | 48 |
| **EDUCATIONAL OBJECTIVES OF THE MODULE** | |
| The modules will focus on recent advances in Genomics and cancer molecular genetics. Specifically they will be addressed genetic factors with respect to altered genes and cellular pathways that control correct cell proliferation. Reading material is taken from the primary literature. | |

## SYLLABUS

| Hrs | Frontal teaching |
|---|---|
| 4 | Analysis of the structure of genes, chromosomes and genomes. Gene function in the post-genomic Era. |
| 10 | -Genetics in the post-genomic Era: Structural, comparative and functional genomics (transcriptome and proteome). -Two channels microarray and gene-chips for genomic analysis. Interpretation of microarray data. CGH array. -Organization of the human genome: gene structure, genomic polymorphisms: SNPs, RFLP, VNTR, STRs. Haplotype. -Pharmacogenomics. |
| 4 | -The cell cycle regulation and analysis of the cell cycle checkpoints. Role of pRB and TP53 tumor suppressors in genomic instability. Viral oncoproteins of DNA viruses (E6-E7; EIA-E1B; LargeTag) their cellular targets and checkpoints. |
| 2 | -The ATM gene, CHK1 CHK2 effectors and their function in the cell cycle checkpoints. |
| 4 | Role of cyclin-dependent kinases inhibitors (CDKIs) p21 and p16. The ARF locus. Relationship between ARF and MDM2. The pathway: E2F1, p14 / 19ARF. |
| 10 | -Genetics and genomics of cancer: The origin of cancer: mutations in oncogenes (activation) / tumor suppressor genes. -Carcinogenesis and tumor progression models (retinoblastoma, colorectal, head / neck). Cancer stem cells. |
| 4 | -Hippo pathway:LATS1 / 2 genes, the role of the Hippo protein pathway in response to anti-mitotic drugs (coactivator TAZ) (experiments). |
| 8 | Human genetic diseases models: cystic fibrosis, muscular dystrophy, Huntington's chorea. Gene therapy |
| 2 | Gene Therapy |