



UNIVERSITÀ DEGLI STUDI DI PALERMO

DEPARTMENT	Biomedicina, Neuroscienze e Diagnostica avanzata		
ACADEMIC YEAR	2019/2020		
MASTER'S DEGREE (MSC)	MEDICINE AND SURGERY		
INTEGRATED COURSE	BIOLOGY AND GENETICS - INTEGRATED COURSE		
CODE	01617		
MODULES	Yes		
NUMBER OF MODULES	2		
SCIENTIFIC SECTOR(S)	BIO/13		
HEAD PROFESSOR(S)	CONIGLIARO ALICE Professore Associato Univ. di PALERMO GRIMAUDDO STEFANIA Professore Associato Univ. di PALERMO ALESSANDRO Professore Ordinario Univ. di PALERMO RICCARDO		
OTHER PROFESSOR(S)	FONTANA SIMONA Professore Associato Univ. di PALERMO DI BELLA MARIA Ricercatore Univ. di PALERMO ANTONIETTA CONIGLIARO ALICE Professore Associato Univ. di PALERMO GRIMAUDDO STEFANIA Professore Associato Univ. di PALERMO ALESSANDRO Professore Ordinario Univ. di PALERMO RICCARDO PIPITONE ROSARIA Professore Associato Univ. di PALERMO MARIA		
CREDITS	10		
PROPAEDEUTICAL SUBJECTS			
MUTUALIZATION			
YEAR	1		
TERM (SEMESTER)	2° semester		
ATTENDANCE	Mandatory		
EVALUATION	Out of 30		
TEACHER OFFICE HOURS	ALESSANDRO RICCARDO Monday 15:00 18:00 Via Divisi 83 Friday 15:00 18:00 Via Divisi 83 CONIGLIARO ALICE Monday 15:00 18:00 Sezione di Biologia e Genetica del Dipartimento BiND in via Divisi 83, oppure attraverso la piattaforma TEAMS. A causa di possibili impegni istituzionali o riunioni di lavoro potrebbe non essere possibile ricevere gli studenti nel giorno e alle ore indicate, pertanto sarebbe preferibile fissare un appuntamento tramite e-mail. DI BELLA MARIA ANTONIETTA Tuesday 16:00 18:00 Dip. Biomedicina, Neuroscienze e Diagnostica avanzata - Sezione di Biologia e Genetica Via Divisi, 83 90133 Palermo FONTANA SIMONA Thursday 15:30 16:30 Dipartimento di Biomedicina, Neuroscienze e Diagnostica avanzata, Sezione di Biologia e Genetica - Via Divisi, 83. A causa di possibili altri impegni istituzionali o riunioni di lavoro potrebbe non essere possibile ricevere gli studenti nel giorno e alle ore indicate. Per questo è preferibile comunque fissare un appuntamento via e-mail. GRIMAUDDO STEFANIA Monday 10:00 12:00 Studio docente Piazza delle Cliniche, 2, piano -1. Dipartimento PROMISE previo appuntamento MAIL		

	GRIMAUDO STEFANIA		
	Wednesday	10:00 12:00	Studio docente Piazza delle Cliniche,2, piano -1. Dipartimento PROMISE previo appuntamento MAIL
	PIPITONE ROSARIA MARIA		
	Wednesday	14:30 16:30	Laboratorio di Patologia Molecolare sito al II piano del Dipartimento Promise, piazza delle cliniche n. 2. Previo appuntamento tramite il seguente indirizzo e-mail: rosariamaria.pipitone@unipa.it.
	Thursday	15:00 17:00	Laboratorio di Patologia Molecolare sito al II piano del Dipartimento Promise, piazza delle cliniche n. 2. Previo appuntamento tramite il seguente indirizzo e-mail: rosariamaria.pipitone@unipa.it.
	Friday	12:00 14:00	Laboratorio di Patologia Molecolare sito al II piano del Dipartimento Promise, piazza delle cliniche n. 2. Previo appuntamento tramite il seguente indirizzo e-mail: rosariamaria.pipitone@unipa.it.

PREREQUISITES	The student must have basic knowledge of chemistry in order to be able to understand the mode of interaction of molecules within the cell. The chemical properties of water; the different types of chemical bonds as well as the main classes of biological macromolecules (sugars, lipids, proteins and nucleic acids) are topic to be known.
LEARNING OUTCOMES	<p>Knowledge and understanding</p> <ul style="list-style-type: none"> - Acquisition of the specific language of Biology and Genetics topics; - to Know and understand the basic biological processes of living organisms and the way in which hereditary characteristics are transmitted to the offsprings; - to know and understand the genetic basis of the most common human diseases or those diseases with an high incidence in the territory. <p>Applying knowledge and understanding</p> <p>Ability to distinguish, organize and implement, in a independent way:</p> <ul style="list-style-type: none"> - the knowledge of basic biological processes of cells and organisms; - The laws which regulate the transmission of hereditary characteristics in living species; - The main methods of cell biology and molecular genetics. <p>Making judgments</p> <p>Being able to evaluate and integrate, in an autonomous manner:</p> <ul style="list-style-type: none"> - The acquired knowledge in biology and genetics in the study of organisms and in particular man; -the consequences of the alterations of biological processes on human diseases. <p>Communication skills</p> <p>Ability to communicate and explain, in a simple way, even to a non-expert public, the biological and genetic processes.</p> <p>Learning ability</p> <p>Ability to properly use the scientific literature for a continuous update of knowledge in the biomedical field.</p> <p>Ability to learn and follow appropriately, using the knowledge acquired in the course, the subsequent teaching courses of the curriculum for the final degree in Medicine and Surgery.</p>
ASSESSMENT METHODS	<p>There will be two tests: an ongoing evaluation constituted by 6 open questions and an oral examination at the end of the course.</p> <p>Objective of the ongoing evaluation of the duration of 90 minutes and to verify the possession of disciplinary skills and knowledge gained after conducting more than a third of the program. The questions tend to verify a) the knowledge gained, and b) the ability of elaborative and synthesis skills. As for the assessment of knowledge, it will be required the ability to contextualize the topic within a specific cellular process illustrating the properties and characteristics of the molecule and / or biological activity.</p> <p>As for the verification of the elaborative abilities, it will be evaluated the ability to extrapolate the minimum details of the process in a clear and concise manner and the understanding of their implications for the topic. Each question will receive a score from 0 to IV with IV the highest score and 0 the lowest score. The evaluation scheme is the following:</p> <p>0 - insufficient knowledge of the contents required by the specific question or the student does not answer;</p> <p>I 18-20 vote; minimum basic knowledge of the subject requested and poor elaborative capacity;</p> <p>II vote 21-24; just enough knowledge of the subject, and limited language abilities;</p> <p>III 25-27 vote good knowledge of the issues and good language abilities; the student is able to correlate the different topics which has studied</p> <p>IV 28-30 vote more than good acquisition of the course content and excellent language abilities and synthesis abilities</p> <p>The oral test consists of a conversation usually lasting 20-30 minutes in order to check the knowledge of the topics of the course related to the remaining two thirds of the program. The final Evaluation will also consider the assessment reached in the ongoing evaluation.</p> <p>Final vote will be expressed according the following scheme:</p> <p>Final vote will be expressed according the following scheme:</p> <p>30-30 e Lode: A-A+ Excellent</p> <p>27-29: B Very good</p> <p>24-26: C Good</p> <p>21-23: D Satisfactory</p> <p>18-20: E Sufficient</p> <p>1-17: F Fail</p>

	The final evaluation is calculated as the average of the scores given in the two tests
TEACHING METHODS	Lessons in the classroom
DOCENTE: Prof.ssa ALICE CONIGLIARO- Sede HYPATIA	
PREREQUISITES	The student must have basic knowledge of chemistry in order to be able to understand the mode of interaction of molecules within the cell. The chemical properties of water; the different types of chemical bonds as well as the main classes of biological macromolecules (sugars, lipids, proteins and nucleic acids) are topic to be known.
LEARNING OUTCOMES	<p>Knowledge and understanding</p> <ul style="list-style-type: none"> - Acquisition of the specific language of Biology and Genetics topics; - to Know and understand the basic biological processes of living organisms and the way in which hereditary characteristics are transmitted to the offsprings; - to know and understand the genetic basis of the most common human diseases or those diseases with an high incidence in the territory. <p>Applying knowledge and understanding</p> <p>Ability to distinguish, organize and implement, in a independent way:</p> <ul style="list-style-type: none"> - the knowledge of basic biological processes of cells and organisms; - The laws which regulate the transmission of hereditary characteristics in living species; - The main methods of cell biology and molecular genetics. <p>Making judgments</p> <p>Being able to evaluate and integrate, in an autonomous manner:</p> <ul style="list-style-type: none"> - The acquired knowledge in biology and genetics in the study of organisms and in particular man; -the consequences of the alterations of biological processes on human diseases. <p>Communication skills</p> <p>Ability to communicate and explain, in a simple way, even to a non-expert public, the biological and genetic processes.</p> <p>Learning ability</p> <p>Ability to properly use the scientific literature for a continuous update of knowledge in the biomedical field.</p> <p>Ability to learn and follow appropriately, using the knowledge acquired in the course, the subsequent teaching courses of the curriculum for the final degree in Medicine and Surgery.</p>
ASSESSMENT METHODS	<p>There will be two tests: an ongoing evaluation constituted by 6 open questions and an oral examination at the end of the course.</p> <p>Objective of the ongoing evaluation of the duration of 90 minutes and to verify the possession of disciplinary skills and knowledge gained after conducting more than a third of the program. The questions tend to verify a) the knowledge gained, and b) the ability of elaborative and synthesis skills. As for the assessment of knowledge, it will be required the ability to contextualize the topic within a specific cellular process illustrating the properties and characteristics of the molecule and / or biological activity.</p> <p>As for the verification of the elaborative abilities, it will be evaluated the ability to extrapolate the minimum details of the process in a clear and concise manner and the understanding of their implications for the topic. Each question will receive a score from 0 to IV with IV the highest score and 0 the lowest score.</p> <p>The evaluation scheme is the following:</p> <p>0 - insufficient knowledge of the contents required by the specific question or the student does not answer;</p> <p>I 18-20 vote; minimum basic knowledge of the subject requested and poor elaborative capacity;</p> <p>II vote 21-24; just enough knowledge of the subject, and limited language abilities;</p> <p>III 25-27 vote good knowledge of the issues and good language abilities; the student is able to correlate the different topics which has studied</p> <p>IV 28-30 vote more than good acquisition of the course content and excellent language abilities and synthesis abilities</p> <p>The oral test consists of a conversation usually lasting 20-30 minutes in order to check the knowledge of the topics of the course related to the remaining two thirds of the program. The final Evaluation will also consider the assessment reached in the ongoing evaluation.</p> <p>Final vote will be expressed according the following scheme: Final vote will be expressed according the following scheme: 30-30 e Lode: A-A+ Excellent 27-29: B Very good 24-26: C Good 21-23: D Satisfactory 18-20: E Sufficient 1-17: F Fail</p> <p>The final evaluation is calculated as the average of the scores given in the two tests</p>
TEACHING METHODS	Lessons in the classroom

PREREQUISITES	The student must have basic knowledge of chemistry in order to be able to understand the mode of interaction of molecules within the cell. The chemical properties of water; the different types of chemical bonds as well as the main classes of biological macromolecules (sugars, lipids, proteins and nucleic acids) are topic to be known.
LEARNING OUTCOMES	<p>Knowledge and understanding</p> <ul style="list-style-type: none">- Acquisition of the specific language of Biology and Genetics topics;- to Know and understand the basic biological processes of living organisms and the way in which hereditary characteristics are transmitted to the offsprings;- to know and understand the genetic basis of the most common human diseases or those diseases with an high incidence in the territory. <p>Applying knowledge and understanding</p> <p>Ability to distinguish, organize and implement, in a independent way:</p> <ul style="list-style-type: none">- the knowledge of basic biological processes of cells and organisms;- The laws which regulate the transmission of hereditary characteristics in living species;- The main methods of cell biology and molecular genetics. <p>Making judgments</p> <p>Being able to evaluate and integrate, in an autonomous manner:</p> <ul style="list-style-type: none">- The acquired knowledge in biology and genetics in the study of organisms and in particular man;-the consequences of the alterations of biological processes on human diseases. <p>Communication skills</p> <p>Ability to communicate and explain, in a simple way, even to a non-expert public, the biological and genetic processes.</p> <p>Learning ability</p> <p>Ability to properly use the scientific literature for a continuous update of knowledge in the biomedical field.</p> <p>Ability to learn and follow appropriately, using the knowledge acquired in the course, the subsequent teaching courses of the curriculum for the final degree in Medicine and Surgery.</p>
ASSESSMENT METHODS	<p>There will be two tests: an ongoing evaluation constituted by 6 open questions and an oral examination.</p> <p>Objective of the ongoing evaluation of the duration of 90 minutes and to verify the possession of disciplinary skills and knowledge gained after conducting more than a third of the program. The questions tend to verify a) the knowledge gained, and b) the ability of elaborative and synthesis skills. As for the assessment of knowledge, it will be required the ability to contextualize the topic within a specific cellular process illustrating the properties and characteristics of the molecule and / or biological activity.</p> <p>As for the verification of the elaborative abilities, it will be evaluated the ability to extrapolate the minimum details of the process in a clear and concise manner and the understanding of their implications for the topic. Each question will receive a score from 0 to IV with IV the highest score and 0 the lowest score. The evaluation scheme is the following:</p> <p>0 - insufficient knowledge of the contents required by the specific question or the student does not answer;</p> <p>I 18-20 vote; minimum basic knowledge of the subject requested and poor elaborative capacity;</p> <p>II vote 21-24; just enough knowledge of the subject, and limited language abilities;</p> <p>III 25-27 vote good knowledge of the issues and good language abilities; the student is able to correlate the different topics which has studied</p> <p>IV 28-30 vote more than good acquisition of the course content and excellent language abilities and synthesis abilities</p> <p>The oral test consists of a conversation usually lasting 20-30 minutes in order to check the knowledge of the topics of the course related to the remaining two thirds of the program. The final Evaluation will also consider the assessment reached in the ongoing evaluation.</p> <p>Final vote will be expressed according the following scheme:</p> <p>30-30 e Lode: A-A+ Excellent</p> <p>27-29: B Very good</p> <p>24-26: C Good</p> <p>21-23: D Satisfactory</p> <p>18-20: E Sufficient</p> <p>1-17: F Fail</p> <p>The final evaluation is calculated as the average of the scores given in the two tests</p>
TEACHING METHODS	Lessons in the classroom.

MODULE BIOLOGY

Prof. RICCARDO ALESSANDRO - Sede IPPOCRATE, - Sede IPPOCRATE

SUGGESTED BIBLIOGRAPHY

"BIOLOGIA E GENETICA" G. De Leo, E. Ginelli, S. Fasano, Ed. Edises, 2019

"BIOLOGIA MOLECOLARE della CELLULA" B. Alberts et al., Ed. Zanichelli, Quinta edizione 2011

"BIOLOGIA MOLECOLARE della CELLULA" H. Lodish et al., Ed. Zanichelli, 2009

"PRINCIPI DI GENETICA"- D. P. Snustad, M. J. Simmons, Ed. Edises, V edizione.

LE SCIENZE (Ed. ital. Scientific American), NATURE, SCIENCE, BIOLOGICAL REVIEWS, Articoli vari

AMBIT	50400-Discipline generali per la formazione del medico
INDIVIDUAL STUDY (Hrs)	90
COURSE ACTIVITY (Hrs)	60

EDUCATIONAL OBJECTIVES OF THE MODULE

Identify the main biological structures, the organization and functioning of a eukaryotic cell; Distinguish viruses, prokaryotic and eukaryotic cells; Analyze and compare the flow of the genetic information in the virus, in haploid organisms and in diploid organisms. Interpret the mechanisms of gene expression in prokaryotes and eukaryotes; Interpret the mechanisms of cellular interactions, gene regulation, cell differentiation and "planning" of development. Interpret and use scientific methodology and recognize different biomedical technologies

SYLLABUS

Hrs	Frontal teaching
4	Scientific Method The cellular Theory; Classification of organisms; Notes on Evolution of species; The Prokaryotic and Eukaryotic Cell; The main classes of biological macromolecules
2	Cytomembranes, organization and functional examples.
6	The genetic material, experimental identification and genetic value of nucleic acids. Composition and structure of nucleic acids; DNA replication.
1	Genome viruses, prokaryotes and eukaryotes.
5	cell cycle, mitosis. Meiosis Gametogenesis. Notes on the reproduction of living organisms.
2	Cell Death: necrosis, apoptosis and autophagy. Molecular Mechanisms and their alterations in pathology
4	Organization of DNA in eukaryotic chromosomes, structural hierarchy of the chromatin material: Structure of genes.
7	Messenger RNA, ribosomal RNA and transfer RNA: structures and functions.
3	Genetic code
6	"Transcription" in prokaryotes and eukaryotes cells; nucleolar organizer and ribosome biogenesis; Processing and Differential splicing of RNA.
5	Protein synthesis in prokaryotes and eukaryotes cells
4	Regulation of gene expression in prokaryotes
4	Regulation of gene expression in eukaryotes; Main transcriptional regulation mechanisms during the process of differentiation; Role of non coding RNA (microRNA and Long non coding RNA)
4	Nuclear totipotency and Cloning; Homeotic genes. Stem cells and their use in regenerative medicine and other therapeutical application
3	Eukaryotic genome, Chromosomes; nuclear and mitochondrial DNA; Genes and amount 'of DNA, repeated sequences; Gene families;

MODULE BIOLOGY

Prof.ssa ALICE CONIGLIARO - Sede HYPATIA, - Sede HYPATIA

SUGGESTED BIBLIOGRAPHY

"BIOLOGIA E GENETICA" G. De Leo, E. Ginelli, S. Fasano, Ed. EdiSES, 2019
 "BIOLOGIA MOLECOLARE della CELLULA" B. Alberts et al., Ed. Zanichelli, Quinta edizione 2011
 "BIOLOGIA MOLECOLARE della CELLULA" H. Lodish et al., Ed. Zanichelli, 2009
 "PRINCIPI DI GENETICA"- D. P. Snustad, M. J. Simmons, Ed. Edises, V edizione.
 LE SCIENZE (Ed. ital. Scientific American), NATURE, SCIENCE, BIOLOGICAL REVIEWS, Articoli vari

AMBIT	50400-Discipline generali per la formazione del medico
--------------	--

INDIVIDUAL STUDY (Hrs)	90
-------------------------------	----

COURSE ACTIVITY (Hrs)	60
------------------------------	----

EDUCATIONAL OBJECTIVES OF THE MODULE

Identify the main biological structures, the organization and functioning of a eukaryotic cell;
 Distinguish viruses, prokaryotic and eukaryotic cells;
 Analyze and compare the flow of the genetic information in the virus, in haploid organisms and in diploid organisms.
 Interpret the mechanisms of gene expression in prokaryotes and eukaryotes;
 Interpret the mechanisms of cellular interactions, gene regulation, cell differentiation and "planning" of development.
 Interpret and use scientific methodology and recognize different biomedical technologies

SYLLABUS

Hrs	Frontal teaching
4	Scientific Method The cellular Theory; Classification of organisms; Notes on Evolution of species; The Prokaryotic and Eukaryotic Cell; The main classes of biological macromolecules
2	Cytomembranes, organization and functional examples.
6	The genetic material, experimental identification and genetic value of nucleic acids. Composition and structure of nucleic acids; DNA replication.
1	Genome viruses, prokaryotes and eukaryotes.
5	Cell Cycle, Mitosis. Meiosis Gametogenesis. Notes on reproduction of living organisms.
2	Cell death: necrosis, apoptosis and autophagy - Molecular mechanisms and their alterations in pathologies
4	Organization of DNA in eukaryotic chromosomes, structural hierarchy of the chromatin material: Structure of genes.
7	Messenger RNA, ribosomal RNA, transfer RNA: structures and functions.
3	The genetic code
6	"Transcription" in prokaryotes and eukaryotes cells; nucleolar organizer and ribosome biogenesis; The maturation of mRNA; Processing and Differential splicing of RNA.
5	"Translation" in prokaryotes and eukaryotes cells
4	Regulation of gene expression in prokaryotes
4	Regulation of gene expression in eukaryotes; Main transcriptional regulation mechanisms during the process of differentiation; Role of non-coding RNA (microRNAs and Long non-coding RNAs)
4	Cellular differentiation, nuclear totipotency and Cloning; Homeotic genes. Stem cells and their use in regenerative medicine and other therapeutic applications
3	Eukaryotic genome, Chromosomes; nuclear and mitochondrial DNA; Genes and amount 'of DNA, repeated sequences; Gene families;

MODULE GENETICS

Prof.ssa MARIA ANTONIETTA DI BELLA - Sede CHIRONE, - Sede CHIRONE

SUGGESTED BIBLIOGRAPHY

"BIOLOGIA E GENETICA" G. De Leo, E. Ginelli, S. Fasano, Ed. Edises, 2019

"PRINCIPI DI GENETICA"- D. P. Snustad, M. J. Simmons, Ed. Edises, V edizione.

LE SCIENZE (Ed. ital. Scientific American), NATURE, SCIENCE, BIOLOGICAL REVIEWS, Articoli vari

AMBIT	50400-Discipline generali per la formazione del medico
--------------	--

INDIVIDUAL STUDY (Hrs)	60
-------------------------------	----

COURSE ACTIVITY (Hrs)	40
------------------------------	----

EDUCATIONAL OBJECTIVES OF THE MODULE

To analyze and to compare the mechanisms responsible of transmission and variations of the genetic information in the viruses, in haploid and diploid organisms.

To identify the patterns of inheritance of genetic traits from organism to organism and to calculate the probability for each scenario.

To compare the different modes of inheritance of genetic characters .

To identify the modes of inheritance of human traits and disorders and to distinguish the effects that occur on the phenotype during development, and by a combination gene-environment.

To understand the practical clinical applications of genetic testing and to select the approaches of analyzing mutations and studying the molecular basis of genetic diseases.

SYLLABUS

Hrs	Frontal teaching
10	Genetic variations and Mutations. Principles of genetic variations; Crossing-over and meiosis as source of recombination. Mutations in somatic and germ line cells; Point mutations and DNA polymorphism. Some mechanisms source of mutations and DNA repair. DNA damage and correlations with cancer, aging and disease development. Chromosome abnormalities, Aneuploidies and their origins.
4	Principles of DNA technologies; Restriction Endonucleases; Vector molecules; DNA cloning; Polymerase Chain Reaction; DNA sequencing; Human genome project: benefits and main disadvantages. Notes on genome editing technologies; Omics sciences: Genomics, Epigenomics, Transcriptomics, Proteomics, and metabolomics.
6	Classical principles of genetics, human genetics; Genotype and Phenotype; Diploidy and sexual reproduction; Mendel's laws; Dominant and recessive phenotypes; Codominant phenotypes; Linkage of alleles
10	Examples of human genetic traits; Link between genotype and phenotype. Inborn errors of metabolism; Locus heterogeneity, allelic heterogeneity; Matrilineal hereditary; Genetic and Hereditary diseases; Complex and multifactorial characters, environmental factors; Comparison between monogenic and polygenic traits; Multiple alleles and Epistasis; Variable expression of phenotypes and penetrance; Genomic Imprinting and epigenetic mechanisms; Principles of population genetics
5	Sex determination in animals and humans; X-Linked recessive inheritance in Drosophila and in humans; X-chromosome inactivation and gene dosage; Y-linked inheritance
5	Standard cytogenetic karyotyping; Chromosome banding methods and chromosome FISH; International Human chromosome nomenclature; Pedigrees: graphical representation and analysis; Counseling and risk assessment; prenatal diagnosis; clinical application of genetic testing to identify molecular basis of diseases; forensic genetics;

MODULE GENETICS

Prof.ssa ROSARIA MARIA PIPITONE - Sede HYPATIA, - Sede HYPATIA

SUGGESTED BIBLIOGRAPHY

"BIOLOGIA E GENETICA" G. De Leo, E. Ginelli, S. Fasano, Ed. Edises, 2019
 "BIOLOGIA MOLECOLARE della CELLULA" B. Alberts et al., Ed. Zanichelli, Quinta edizione 2011
 "BIOLOGIA MOLECOLARE della CELLULA" H. Lodish et al., Ed. Zanichelli, 2009
 "PRINCIPI DI GENETICA"- D. P. Snustad, M. J. Simmons, Ed. Edises, V edizione.
 LE SCIENZE (Ed. ital. Scientific American), NATURE, SCIENCE, BIOLOGICAL REVIEWS, Articoli vari

AMBIT	50400-Discipline generali per la formazione del medico
--------------	--

INDIVIDUAL STUDY (Hrs)	60
-------------------------------	----

COURSE ACTIVITY (Hrs)	40
------------------------------	----

EDUCATIONAL OBJECTIVES OF THE MODULE

Analyze and compare the manifestations of continuity and variability of genetic information in viruses, in organisms haploid and in diploid ones. Identify the mode of transmission of hereditary characters and evaluate the probability of them appearance in the progeny; Compare the transmission modes of hereditary characters. Understanding the Mechanisms of inheritance also in the human species; To identify in man the inheritance of normal and mutated characters and to distinguish them the expression during the development and life of the individual, in the relationship between genotype and environment; Select the approaches methods for the study of hereditary diseases Using genetic analysis methods in medical practice Interpreting i results of cytogenetic analysis and molecular investigations of the human genome for the purposes of genetic counseling and applications in medicine.

SYLLABUS

Hrs	Frontal teaching
10	Variability and Mutation, Molecular Mechanisms and Biological Relevance. Crossingover and meiosis as a genetic "mixer". Somatic and germline mutations; Mutations and polymorphisms of DNA; Some mechanisms of onset of mutations and DNA repair: correlations with human pathologies, with cell aging and cancer; Chromosomal, genomic and mutations their origin.
4	Gene analysis methods, restriction endonucleases, gene vectors; DNA cloning; Chain Polymeric Reaction; The Genome project: opportunities and risks; Notes on genomic editing; Omic sciences: Genomics, Epigenomics, transcriptomics, proteomics and metabolomics.
6	Formal genetics and human genetics, Genotype and phenotype. Diploidy and sexuality. Mendel and his experiments. Intermediate inheritance, Evaluation of the Mendelian concept of "dominance" and "recessivity"; The linkage, the associated characters.
10	Examples of hereditary traits in the human species: transmission and genotype-phenotype relationship. Comparison between mono- and polygenic characters; Codominant phenotypes; Enzymatic deficiencies hereditary and genetic heterogeneity at multiple loci; Allelic heterogeneity; Cytoplasmic inheritance; Distinction between genetic and hereditary diseases. Inheritance of quantitative and multi-factorial characters; Multiple alleles; Epistasis. Variability of gene expression; Expressiveness and Penetrance, incidence of environmental factors; Imprinting and epigenetic inheritance; Basics of population genetics.
5	Genetic basis of sex. The determination of sex in animal species and in humans; Inheritance associated with sex: diagenic in Drosophila and in the human species; gene dosage and Lyonization; Heredity olandrica human species.
5	566/5000 Human karyotype: Banding of chromosomes; In situ hybridization techniques (FISH); International terminology for the representation of the karyotype. Formal graphic representation of genotypes. Study of family trees. Genetic counseling and prenatal diagnosis: the probability in human genetics e in the analysis of family trees for genetic prognosis. Applications of Genetics in Medicine: molecular technologies for diagnosis; use of polymorphisms associated with mutations for the molecular diagnosis of genetic pathologies; forensic examples.

MODULE GENETICS

Prof.ssa SIMONA FONTANA - Sede IPPOCRATE, - Sede IPPOCRATE

SUGGESTED BIBLIOGRAPHY

"BIOLOGIA E GENETICA" G. De Leo, E. Ginelli, S. Fasano, Ed. EdiSES, 2013
 "EREDITA': PRINCIPI E PROBLEMATICHE DELLA GENETICA UMANA" M.R. Cummings, Ed. EdiSES, Terza edizione 2014
 LE SCIENZE (Ed. ital. Scientific American), NATURE, SCIENCE, BIOLOGICAL REVIEWS, Articoli vari

AMBIT	50400-Discipline generali per la formazione del medico
INDIVIDUAL STUDY (Hrs)	60
COURSE ACTIVITY (Hrs)	40

EDUCATIONAL OBJECTIVES OF THE MODULE

Identify the main biological structures, the organization and functioning of a eukaryotic cell;
 Distinguish viruses, prokaryotic and eukaryotic cells;
 Analyze and compare the flow of the genetic information in the virus, in haploid organisms and in diploid organisms.
 Interpret the mechanisms of gene expression in prokaryotes and eukaryotes;
 Interpret the mechanisms of cellular interactions, gene regulation, cell differentiation and "planning" of development.
 Interpret and use scientific methodology and recognize different biomedical technologies

SYLLABUS

Hrs	Frontal teaching
10	Variability and mutation, molecular mechanisms and biological relevance, meiosis as a genetic mixer. Somatic and germinal mutations. DNA polymorphisms and mutations. Mechanisms of mutations onset, DNA repair, relation with human pathologies, cell aging and cancer. Chromosome and genomic mutations, cause of occurrence.
4	Methods of gene analysis, restriction endonucleases, vectors for cloning and expression, DNA cloning, polymerase chain reaction, Genome Project: opportunities and risks; overview on genome editing; -Omics sciences: genomics, transcriptomics, proteomics and metabolomics;
6	Formal and human genetics, genotype and phenotype, diploidy and sex, Mendel and his experiments, intermediate heredity, recessive and dominant, linkage
10	Examples of genetic transmission in humans. genotype-phenotype relationship. Analysis of mono and polygenic traits. hereditary enzymatic deficiencies, co-dominant phenotype, multi-loci heterogeneity, allelic heterogeneity, cytoplasmatic heterogeneity, quantitative traits, and multifactorial traits, multiple alleles, epistasis, variability in genetic expression, expressivity, penetrance, role of environment and its incidence, background of population genetics, Imprinting and epigenetic heredity.
5	Genetic basis of sex, sex determination in animal species, X- and traits in Drosophila and in humans, gene dosage and Lyonization, Y-linked traits in humans
5	Human karyotype, chromosome banding, Methods of in situ hybridization (FISH), international representation of karyotype, genotype representation, genealogical trees, genetic counseling and prenatal diagnosis, probability in human genetics, applications of genetics in medicine, molecular methods for diagnosis of genetic diseases, forensic examples

MODULE BIOLOGY

Prof.ssa STEFANIA GRIMAUDO - Sede CHIRONE, - Sede CHIRONE

SUGGESTED BIBLIOGRAPHY

"BIOLOGIA E GENETICA" G. De Leo, E. Ginelli, S. Fasano, Ed. Edises, 2019

"BIOLOGIA MOLECOLARE della CELLULA" B. Alberts et al., Ed. Zanichelli, Quinta edizione 2011

"BIOLOGIA MOLECOLARE della CELLULA" H. Lodish et al., Ed. Zanichelli, 2009

"PRINCIPI DI GENETICA"- D. P. Snustad, M. J. Simmons, Ed. Edises, V edizione.

LE SCIENZE (Ed. ital. Scientific American), NATURE, SCIENCE, BIOLOGICAL REVIEWS, Articoli vari

AMBIT	50400-Discipline generali per la formazione del medico
INDIVIDUAL STUDY (Hrs)	90
COURSE ACTIVITY (Hrs)	60

EDUCATIONAL OBJECTIVES OF THE MODULE

Identify the main biological structures, the organization and functioning of a eukaryotic cell;
Distinguish viruses, prokaryotic and eukaryotic cells;
Analyze and compare the flow of the genetic information in the virus, in haploid organisms and in diploid organisms.
Interpret the mechanisms of gene expression in prokaryotes and eukaryotes;
Interpret the mechanisms of cellular interactions, gene regulation, cell differentiation and "planning" of development.
Interpret and use scientific methodology and recognize different biomedical technologies

SYLLABUS

Hrs	Frontal teaching
4	Scientific Method The cellular Theory; Classification of organisms; Main aspects of evolution; Prokaryotic and eukaryotic cells; main biological macromolecules.
2	Citomembrane, organization and functional examples.
6	The genetic material, experimental identification and genetic value of nucleic acids. Composition and structure of nucleic acids; DNA replication.
1	Genome viruses, prokaryotes and eukaryotes.
5	Cell cycle, Mitosis. Meiosis and Gametogenesis. Notes on reproduction of living organisms.
2	Cell Death: necrosis, apoptosis and autophagy. Molecular mechanisms and pathological alterations.
4	Organization of DNA in eukaryotic chromosomes, structural hierarchy of the chromatin material: Structure of genes.
7	Messenger RNA, ribosomal RNA and transfer RNA: structures and functions.
3	Genetic code
6	"Transcription" in prokaryotes and eukaryotes cells; nucleolar organizer and ribosome biogenesis; Processing and Differential splicing of RNA.
5	Protein synthesis in prokaryotes and eukaryotes cells
4	Regulation of gene expression in prokaryotes.
4	Regulation of gene expression in eukaryotes. Main transcriptional regulation mechanisms during the process of differentiation. Role of non coding RNA (micro RNA and Long Non Coding RNA).
4	Cell differentiation. Nuclear totipotency and Cloning; homeotic genes. Stem cells use in regenerative medicine and other therapeutics applications.
3	Eukaryotic genome, Chromosomes; nuclear and mitochondrial DNA; Genes and amount of DNA, repeated sequences, gene families.