Course Title	**Human Genetics in the 21st century**				
Course Code	ABS313				
Course Type	Program specific Elective				
Level	BSc (Level 1)				
Year / Semester	3rd year /5th semester				
Teacher's Name	Dr Kyproulla Christodoulou				
ECTS	6	Lectures / week	3+2*	Laboratories / week	-
Course Objectives	Education in the cutting edge field of human genetics through lectures and laboratory exercises. Understanding the perspective on providing diagnostic services to individuals and families with diseases that have a genetic background or genetic vulnerability. Familiarity with basic laboratory methods used to investigate and diagnose hereditary diseases and disorders.				
Learning Outcomes	At the completion of the course the student will be able to: • Recognize the types of monogenic inheritance and polygenic inheritance. • Understand the concepts of genomic imprinting, acceleration and genetic vulnerability. • Develop skills to record the family tree and determine the type of inheritance in a family. • Understand the four main pillars of human genetics – cytogenetics, biochemical genetics, medical genetics and cancer genetics • Recognize and record karyotypes. • • Understand and record haplotypes. • • Understand the basic methods of genetic testing. • • Understand the results of genetic tests and their significance for the individual, as well as genetic risk assessment. • • Know the basic principles of genetic counseling. • • Understand statistical genetics methods such as linkage analysis and genome-wide association. • • Know the importance of personalized medicine and the gene therapies that have been developed to date.				
Prerequisites	-	Required	-		
Course Content	Theory: • Introduction to the human genome – organization, structure and gene expression • Models of monogenic inheritance • Polygenic inheritance • Genomic imprinting • Anticipation				

	• Heredity, genetic susceptibility and multifactorial diseases • Pedigrees / Family trees • Cytogenetics • Genomics • Medical genetics • Biochemical genetics • Cancer genetics • Genetic testing methods • Genetic risk assessment • Genetic counselling • Gene therapy Workshops: • Pedigree construction • Determining the mode of inheritance in family trees • Genotype and haplotype analysis • Karyotyping • Genetic linkage analysis • Genome-wide association studies
Teaching Methodology	The teaching of the course includes lectures to help students understand the theoretical background, and laboratory exercises in order to get a better comprehension of the main concepts of Human Genetics. Methods such as discussion, questions/answers, and pros/cons, are used to enhance student's participation. PowerPoint and image-rich material and short animations are used to better understand the content of Human Genetics.
Bibliography	Textbooks: • Human Genetics: the basics, by Ricky Lewis, Routledge • ISBN 1259700933, 9781259700934. Human Genetics: concepts and applications, by Ricky Lewis, Published: McGraw Hill Education • ISBN 1605353132, 9781605353135. A primer of Human genetics, by Greg Gibson, Published; Sinauer. References: • A list of articles will be provided for further reading.
Assessment	Course Work 40% • Mid-term Test 20% • Workshops/exercises/case studies 20% Final Exam 60% For student evaluation, the overall grade is determined by a written midterm exam (20%), a laboratory grade (20%) and a written final exam (60%). The mid-term exam is carried out between the 6th and 8th week and it mainly includes short answer- questions and problem- solving questions and examines specific modules of the course. As far as the workshops/exercises/case studies grade is concerned, students also have to prepare a report for each workshop topic and the

	report is evaluated with discussion, questions/answers related to the field of Human genetics. The final exam of the course is carried out during the 14th-16th week of each semester and includes short answer questions, decision questions, and problem-solving questions regarding all course modules. The final assessment of the students is formative and summative and is assured to comply with the subject's expected learning outcomes and the quality of the course.
Language	Greek, English