

Name of the module: Genetics 2nd year Medicine

Number of module

BGU Credits:

ECTS credits: number of credits in the
European Credit Transfer System

Academic year: 2nd year medicine

Semester: 2nd

Hours of instruction: by the schedule

Lectures 35 hours

PBL: 4 hours

Location of instruction:

Specific classroom numbers are indicated in the schedule.

Language of instruction: Lectures will be given in Hebrew.

Cycle: B.Med.Sc/Medical Sciences

Position: Obligatory module intended for 2nd year medical students, as part of their preclinical teaching.

Field of Education: Genetics.

Responsible department: Institution of Human Genetics, Soroka University Medical Center.

General prerequisites: Students should complete successfully the following modules (given in prior modules): Basic pathology, Molecular Biology, Medical Biochemistry, Medical Physiology .

Grading scale: Score more than 65 in combination of MQT (100%)

Course Description:

Aims of the module: The goal of the genetics module is to introduce and teach basic principles and practice in modern genetics.

Objectives of the module: Objectives are to establish basic understanding of the fundamentals of human genetics. Provide a modern view on modern genetics its importance and implication in medical practice.

Learning outcomes of the module: On successful completion of the course, the student should be able to:

1. Describe all inheritance modes.
2. Analyze pedigrees from variety of inheritance modes .
3. Outline the mitosis and meiosis processes including the DNA and chromosomal content.
4. Describe a Karyotype. Outline main chromosomal abnormalities and its clinical significance.
5. Assess risk in genetics. Including recurrence risk calculation and bayesian analysis
6. Describe Mendelian concepts of inheritance, some representing mendelian disorders and its implication in medical practice.
7. Describe Non-Mendelian concepts of inheritance, some representing non-mendelian disorders (imprinting, mitochondrial etc) and its implication in medical practice
8. Describe polygenic and multifactorial inheritance, some representing disorders and its implication in medical practice.
9. Describe basic concepts in molecular genetics (central dogma, DNA and RNA processing, protein biology). Mutation types , methods used in clinical diagnosis (Sanger sequencing, MLPA, RLFP, Western blot, PCR, CMA, SNP array, NGS)
10. Describe basic concepts of population genetics, allele frequencies, including Hardy–Weinberg
11. Describe genetics screening tests and strategies including common diseases as Fragile X , Cystic Fibrosis, Spinal Muscular atrophy etc
12. Describe genetic screens for aneuploidy during pregnancy, Chorionic Villous Sampling , Amniocentesis, Karyotype implication, perinatal counseling, Non invasive prenatal diagnosis, Preimplantation Genetic Diagnosis/Screening
13. Describe congenital malformation, the various types and developmental processes and factors involved creating malformation .
14. Be familiar with the option of treatment in genetic diseases. Will be familiar with concepts of personalized medicine and pharmacogenetics.
15. Be familiar with concepts of oncogenetics, oncogenetic syndromes and DNA damage repair processes and implication in practice.
16. Discuss ethical issues in genetic counseling and testing

Attendance regulation: Attendance to the oral lectures is not obligatory. Participation in the PBLs, Clinical discussions is obligatory.

Teaching arrangement and method of instruction: Instruction in the module is based on frontal oral lectures, clinical discussions, PBLs.

Lecturer: Dr Arie Koifman

Contact details:

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Module evaluation: at the end of the semester the students will evaluate the module, in order to draw conclusions, and for the university's internal needs

Confirmation:

Last update:

Assessment:

Students will be assessed in the module by Combination of a MCQ exam (passing with a score of 65 or higher) which will be 80% of the final score and an assessment of in class discussion or assignments , which will be 20% of the final score. Passing each part is obligatory.

Work and assignments: Students are required to take active part in the PBLs meetings, and present a ten minute presentation to the whole class – summarizing their PBL.

Time required for individual work: in addition to attendance in class, the students are expected to do their assignment and individual work: students are required to study and review the lectures at home. Roughly 1 hour per an hour lecture. PBL learning and preparation will take 4hr.

Module Content\ schedule and outlines: See attached

Required reading: Students are expected to read the relevant textbook chapters as will be assigned in the individual study units

Additional literature: Module textbook is :” Medical Genetics 4e by Lynn B. Jorde PhD, John C. Carey MD MPH and Michael J. Bamshad MD (2009)

*All learning material will be available to the students on the module's website (high-learn)/ library/ electronic documents available to BGU students

