

General Information

Human Molecular Genetics - 136088

2020/2021 Winter Semester
Wednesday, 8:30-11:30
To be notified

Teaching Staff:

Instructor: Sara Selig
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04-8295277/81

Office Hours: By appointment on Zoom

TA's: No TAs

Prerequisites: 134020, 134082, 134119 (you can consult with the lecturer if you don't have all three)

Credits: 3

Study hours per week: Lecture - 3

Course Goals and Description

The course aims to introduce a wide spectrum of topics related to human genetics with emphasis on human molecular genetics. In addition to basic topics, the course will include the newest developments in the field of human genetics and will be updated on a yearly basis in order to present the current status of this quickly emerging field to the students. The course is based on prerequisite courses such as basic genetics, and molecular biology in order to enable discussion of advanced topics in genetics. The main goal of the course is to provide the students with wide knowledge in human genetics and the ability to read and comprehend published work in the field. The course includes three hours of lecture per week. Students are requested to review the topics taught in the previous weeks since many lectures are heavily based on topics covered earlier in the course. In addition, a published paper in Human Genetics is provided on the course site at the beginning of the semester. The students are required to read this manuscript on their own. Several questions in the final exam will be related to this manuscript.

Learning Outcomes

The student will be familiar with many topics related to the field of human genetics. These will include basic principles in human genetics, different modes of inheritance, methods in human molecular genetics and human cytogenetics, different mechanism of pathology of human genetics diseases, human genetics projects such as the human genome project, 1000 genome

project, and more, cancer genetics, methods for genetic testing both preimplantation, during pregnancy and after birth and approaches for gene therapy. Topics will include the newest developments in the field. Ethical issues related to human genetics will be discussed. The students will be expected to integrate knowledge from different topics in order to understand many aspects of a certain genetic disease.

On successful completion of this module, students should be able to:

- Demonstrate an understanding of the various types of gene mutations and explain the mechanisms by which they lead to human disease
- Analyze a pedigree and determine the mode of inheritance
- Assess the relevance of the structure of the human genome and its different components to the mechanisms of different human genetics disorders
- Analyze the results of basic genetic testing carried out with polymorphic markers
- Evaluate methods and techniques described in current literature in human genetics
- Determine which types of genetic testing can be carried out for different genetic disorders
- Describe the newest technologies in the field of genetics such as next generation sequencing, and genome editing technologies
- Navigate relevant websites related to human genetics
- Explain how modern molecular techniques can be used for treatment of human cancer and genetic disorders
- Analyze and interpret genetic data and critically appraise scientific papers
- Be mindful to ethical issues related to human genetics

Course Content/Topics

- Introduction to human genetics (Mendelian inheritance)
- The genome organization
- Mutations
- Microsatellite-expansion diseases
- Inborn errors in metabolism
- Cloning of genes involved in Mendelian disorders
- Human genome, HapMap and 1000 genome projects
- Cancer genetics
- Non-Mendelian genetics: Mosaicism, imprinting, allelic exclusion, mitochondrial inheritance
- Cytogenetics: methods in classical cytogenetics, molecular cytogenetics, cytogenetic disorders.
- Sex chromosomes, sex determination
- Multifactorial inheritance
- Genetic testing: methods, pre-implantation diagnosis, prenatal diagnosis.
- Gene therapy including genomic editing.

Assignments and Grading Procedures

A published paper in Human Genetics will be provided on the course site several weeks after the start of the semester. The students are required to read this paper on their own. Several questions in the final exam will be related to this manuscript.

Exam 100%

Course Schedule (Topics, assignments, Exams)

| Topic | Lecture Topic/Sub-topic | Textbook Reference : T& T, 7th ed | Textbook Reference : S&R, 3rd ed | Key Dates (Assignment/Exam) |
|--------------|------------------------------------------------------------------|---------------------------------------------------------|--------------------------------------------------------|------------------------------------|
| 1 | Introduction to single-gene inheritance, The genome organization | Thompson and Thompson, Ch. 7 | Strachan and Read Ch. 4, 9 | |
| 2 | Mutations and polymorphisms | Thompson and Thompson, Ch. 9 | Strachan and Read Ch. 11 | |
| 3 | Principles of molecular disease and microsatellite diseases | Thompson and Thompson, Ch. 11, 7 | Strachan and Read Ch. 16, 11 | |
| 4 | Inborn errors in metabolism | Thompson and Thompson, Ch. 12 | | |
| 5 | Gene mapping and disease gene identification | Thompson and Thompson, Ch. 10 | Strachan and Read Ch. 13, 14 | |
| 6 | Human genome project and following projects | | Strachan and Read Ch. 8 | |
| 7 | Cancer genetics | Thompson and Thompson, Ch. 16 | Strachan and Read Ch. 17 | |
| 8 | Non-Mendelian inheritance | Thompson and Thompson, Ch. 7 | Strachan and Read Ch. 16 | |
| 9 | Cytogenetics and molecular cytogenetics | Thompson and Thompson, Ch. 5 | | |
| 10 | Clinical cytogenetics and sex determination | Thompson and Thompson, Ch. 6 | | |
| 11 | Multifactorial inheritance | Thompson and Thompson, Ch. 8, 10, 17 | Strachan and Read Ch. 15 | |
| 12 | The treatment of human disease | Thompson and Thompson, Ch. 13 | Strachan and Read Ch. 21 | |
| 13 | Genetic testing | Thompson and Thompson, Ch. 15 | Strachan and Read Ch. 18 | |

Course Requirements & Course Policies

While attendance is not obligatory, students are highly encouraged to attend lectures. This year the course will be given in hybrid teaching, therefore all given lectures will be given at the campus according to the schedule, but will also be shown on ZOOM and recorded. **The exam will include in addition to the presentations that appear on the course site, also material that was discussed during the lectures and does not appear on the presentations.** Students are expected to be familiar with material taught in previous lectures, since the course is built gradually on accumulating knowledge and lectures integrate material from previous topics. Two textbooks are recommended for further reading and clarifications of material taught in class. The relevant chapters for each topic appear in the syllabus. An exam at the end of the semester will determine 100% of the grade of the course. The exam will be composed of questions with multiple-choice answers that cover all the topics taught during the semester. Approximately three questions will be relevant to the manuscript in human genetics, uploaded to the course site during the semester. The students are required to read this paper on their own. These questions will assess the ability of the student to read and understand on their own a manuscript in human genetics.

In case a student wants to meet with the lecturer by Zoom, an appointment can be arranged in advance. If possible, a day or two prior to the exam, a review class on Zoom will be scheduled. Students are strongly encouraged to participate during lectures and ask questions related to unclear topics. All efforts will be done so that this will be possible also during the hybrid teaching.

In the case that exams will not be carried out at the campus due to the COVID-19 situation, the multiple choice exam described above will be given online. In both cases, the exam determines 100% of the final grade.

Accommodation for Students with special needs

Students who will be on reserve duty during one of the exam dates, will be able to be examined at an alternative date.

Text book(s) and/or other materials

Course site: <http://moodle.technion.ac.il/course/view.php?id=2576>

Textbooks:

Genetics in Medicine, Thompson and Thompson, 7th edition, 2007

Human Molecular Genetics, Strachan and Read, 3rd edition, 2004

The lectures are based also on published manuscripts that will be quoted in the lecture presentations.

Another useful site:

Online inheritance in Man (OMIM): <http://www.omim.org/>

Academic Integrity

During examinations, you must do your own work. Any collaborative behavior during the examinations will result in failure of the exam and may lead to failure of the course and university disciplinary action.