Sackler Faculty of Medicine
Tel Aviv University

MSc. in Medical Sciences with Specialization in Genetic Counseling

Reasons for applying

Genetic counseling is an essential component of providing information to patients about diagnosis of genetic diseases. Genetic counseling is the application of all the genetic information for clinical genetics. As genomic information is advancing rapidly and has immediate clinical significance, there is a shortage of consultants in this field and has raised the need to train more genetic counselors in Israel. At the *Department of Human Molecular Genetics and Biochemistry, Sackler School of Medicine*, these advanced methods are incorporated in research, and enable our program to train genetic counselors and expose them to the most innovative information. The goal is to provide genetic institutes with the best-trained conselors.

Teachers and coordinators of the program

The program is based on the faculty members teaching at the Graduate School of Medicine, the members of the clinical staff in hospitals and genetic institutes and on genetic counselors.

Curriculum

The program rationale, goals and objectives:

Genetic counseling is the practical application of all theoretical areas of human genetics. This is a rapidly developing field, expected to develop even further. A genetic counselor provides services in aspects of genetic diseases, prenatal diagnosis, identification of carriers of genetic diseases, genetic elements in malignant diseases, and more. In view of the rapid advances in diagnostics of genetic diseases, this area has most important implications for individuals and their families. The profession of a genetic counseling involves thorough knowledge of human genetics, molecular technologies of diagnosis and treatment, a deep understanding of clinical and molecular aspects of hereditary diseases and

orientation in psychological aspects associated with treating people. The M.Sc. program includes theoretical knowledge and practical training at genetic institutes affiliated to Tel Aviv University.

Today there is increasing demand for genetic counseling worldwide, and there is a shortage of genetic counselors in Israel, in particular.

In Israel, currently, there are two programs of genetic counseling at two institutions: M.Sc. at the School of Medicine of the Hebrew University and at the Technion. At both institutions, this is a non-thesis degree, with six students graduating once every two years in each institute.

Currently, the requirements of the Ministry of Health for a work certification in genetic counseling includes a Master's degree in Human Genetics or Genetic Counseling, followed by an internship of two years at a recognized genetic institute in a hospital, and finally, a Certification Exam of the Ministry of Health. Genetic institutes prefer graduates of the dedicated genetic counseling program for an internship. The programs currently available are not sufficient to meet the needs for genetic counseling, and therefore we believe there is an urgent need for a new program, which will be part of the distinguished Sackler School of Medicine at Tel Aviv University.

<u>Target population, expected number of students, complementary studies</u>
B.Sc. graduates of the Faculties of Life Sciences and Medicine are candidates for the Genetic Counseling program. Graduates with a B.Sc. degree in biology and medical sciences will not require complementary studies. A graduate of another faculty, e.g., School of Health Professions, will be evaluated on his/her merits, and usually, will have to pass complementary courses before starting his/her studies in Genetic Counseling.
In addition, the program will consider the possibility of accepting Psychology graduates with appropriate complementary courses.

The number of students planned is 12 every two years.

Acceptance conditions

The minimum GPA for candidates in their undergraduate degree is 85. In addition, the candidates have to fill in a questionnaire and undergo an interview.

Details of the program without a thesis

The *MSc. in Medical Sciences with Specialization in Genetic Counseling* is a non-thesis M.Sc. program. Acceptance to any of the other M.Sc. programs of the Sackler School of Medicine or the Department of Human Molecular Genetics and Biochemistry does not constitute automatic acceptance to the genetic counseling program.

Requirements

M.Sc. in Medical sciences with Specialization in Genetic Counseling, includes:

• *Participation in courses* - a total of 45 academic points.

Required courses given at the Graduate School of Medicine - 18 points, required designated courses - 23 points, an elective course to be chosen from the Graduate School of Medicine courses – 4 points.

• *Practical training* in genetic counseling will take place in the genetic institutes of affiliated hospitals (5 hospitals). The training will include observation of genetic counseling in genetics institutes, with personal supervision by a genetic counselor. Students will be exposed to various counseling areas. They will be divided into groups of two/three students, each group trained in a different genetic institute and each student will be supervised by a genetic counselor. During the second semester of the first year, the training will take place once a week. In the second year, the training will be given twice a week (full days). There will be a rotation between the institutes, i.e., each student will rotate in all genetic institutes affiliated to the Sackler School of Medicine. Thus, the students will be exposed to different methods practiced in different hospitals and to different types of genetic counselors. They will sit with genetic counselors and medical geneticists during different counseling sessions. During the practical training, the students will submit structured reports of the cases observed in the clinic.

• The *final project* will be chosen from the cases the students are exposed to in their training and will be submitted in writing to their personal supervisor from the genetics institute.

• The *final grade* of the degree will be calculated as follows: 70% GPA in courses, 20% final project grade, 10% evaluation of the practical training given by the responsible genetic counselors (clinical instructors).

Required courses of the program

Selected from the faculty courses – 18 academic points

1. Developmental biology and embryology. 3 wh - 0104.2506.01
2. Anatomy. 4 wh - 0104.1401.01
3. Biostatistics. 4 wh 0103.0010.01.
4. Scientific Writing. 1.5 wh 0103.7000.01.
5. Ethics in Medicine. 0.5 wh 0103.6000.01.
6. Decision Making in Medicine. 2 wh - 0103.0032.01.
7. Ovarian Physiology. 2 wh - 0114.6565.01
9. Genetics in the Post-Genomic Era. 1 wh - 0114.6545.01.

Required new courses opened for the specific program, given at the hospitals, concentrated to one day a week:

1. Cytogenetic and Molecular Laboratory Methods. 4 wh: lectures, exercises and laboratory.
2. Clinical Genetics. 6 wh: lectures and student seminars.
3. Imaging Techniques and Fetal medicine. 4 wh. Lectures.
4. Aspects of Genetic Counseling. 4 wh: lectures, workshops, simulations, seminars.

A new required course opened for the program, and also open for other students of the Graduate School, will take place at the university:

5. Genetic Screening programs. 2 wh. Lectures and seminars.

New molecular methods – practical lab course of one week – 3 wh

Total hours of designated required courses 23 wh.

First semester in first year
1. Embryology.
2. Anatomy.
4. Biostatistics

Second semester in first year
1. Anatomy
2. Genetic Screening programs.
3. Ovarian Physiology.
4. Clinical Genetics
5. Methods of Imaging and Fetal Medicine
6. Genetic Counseling
7. Cytogenetic and Molecular Laboratory Methods
8. One day a week: Observations of genetic counseling in genetic institutes

First semester in second year
1. Decision Making in Medicine
2. Clinical Genetics
3. Imaging Techniques and Fetal Medicine
4. Genetic Counseling
5. Cytogenetic and Molecular Laboratory Methods
6. Two days a week: Observations of genetic counseling in genetic institutes

Second semester in second year
1. Clinical Genetics
2. Scientific Writing
3. Ethics in Medical Research.
4. Genetics in the Post-Genomic Era
5. Two days a week: Observations of genetic counseling in genetic institutes

For questions, please contact

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Prof. Karen Avraham (karena@post.tau.ac.il)

Prof. Lina Basel (basel@post.tau.ac.il)

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Syllabus of the course: Genetic Counseling

The aim of the course is to give an insight of genetic counseling to the students. We will talk about counseling strategies, approach to problematic cases, ethical problems in genetic counseling, The genetic law in Israel, etc. The students will use simulations and discussion on cases in order to practice different methods of counseling.

1. Genetic Counseling - history and nature.
2. Pedigree - how to draw and interpret it.
3. Genetic counseling skills
4. Philosophy of genetic counseling and understanding the counselees.
5. Risk calculation (e.g. Baysian analysis, Hardy Weinberg law).
7. Psychological research in genetic counseling.
8. Coping with uncertainty and risk perception - an explanation of the problems by example cases.
9. Different ways to present results and risks. Display cases, simulations.
10. Genetic and law in Israel, introduction to ethics and genetics.
11. Ethics of clinical research in Israel.
12. Genetic information given to family members – genetics and law.
13. Simulated cases and analysis.
14. Simulated cases and analysis.
15. Seminars in selected subjects.
16. A patient with CMT will meet the students and present the side of being affected with a genetic disorder.
17. Termination of pregnancy - rules and examples.
18. Termination of Pregnancy - dealing with grief and loss after the termination of pregnancy.

19. The way to give "bad news" – including examples and practice (role playing games).

20. Risk management – genetics and the law.

21. Pre-symptomatic diagnosis - lectures and seminars.

22. Protocols of work in different cases - "rolling cases"

23. Advanced genomic testing (CMA / Exome) – “Is a lot of information good"? (role-playing games).

24. Rolling cases

25. Simulated cases and filming process.

26. Analysis of simulation cases.

27. Summary and discussion of cases raised by the students.

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Syllabus of the course: Genetic Screenings

The aim of the course is to give students access to genetic screenings. Why do we need screenings in the general population for diagnosing genetic diseases? Topics include: significance for diagnostics of common genetic diseases, who are they designed for, and how do we choose diseases for screening.

1. Introduction and goals of genetic screening.
2. Types of screenings.
3. Preconceptional molecular screening.
4. Screening tests during pregnancy
5. Ataxia Telangiectasia is one of the diseases in the molecular screening. The students will meet with people from the AT family association.
6. Neonatal screening in Israel.
7. A meeting with families from the SMA family association.
8. Screening in the orthodox families – "Dor Yesharim".
9. Pre-symptomatic screening tests.
10. Direct to consumer screenings (23 and Me, for example)
11. Familial Dysautonomia – a meeting with the FD Family Association.
12. Screening tests in common neoplasia (ovary, breast and colon cancer)
13. Seminars
14. Seminars

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Syllabus of Course: Cytogenetic and Molecular Laboratory Methods for Clinical Genetic Diagnostics

The aim of the course is to give the students an approach to basic material in cytogenetic and molecular genetics with emphasis on laboratory methods that are applied today in clinical cases. Emphasis will be given on the theory and understanding of the lab methods and understanding of the results. The course will include new molecular methods and application of bioinformatic analysis in the genetic world.

1. Cytogenetics - chromosome structure, Karyotype, mitosis, meiosis.

2. Cytogenetics - cytogenetic analysis methods / karyotype + FISH, staining. Exercise of karyotypes.

3. Cytogenetics - common chromosomal syndromes (translocations, inversions, chromosomal aberrations, sex chromosomal disorders and inactivation).

4. Cytogenetics – visiting a cytogenetic laboratory and practice.

5. Introduction to molecular genetics.

6. DNA / RNA / protein

7. Genetic mapping (the beginning) - for example: linkage analysis, The Human Genome Project, HAP MAP, GWAS

9. Types of mutations and methods for analysis of different mutation types.

10. Types of mutations and methods for analysis of different mutation types.

11. Biochemical methods of analysis.

12. Methods for analysis of triplet repeat expansion + exercise

13. CMA – understanding the methods and practical utilization.

14. Using CMA for genetic analysis – exercise.

15. Using bioinformatics databases to analyze mutations.

16. NGS – understanding the technology.

17. Practicing the use of databases - computer class

18. Whole genome / whole exome sequencing - theory and practice

19. Whole genome / whole exome sequencing - theory and practice

20. Unusual ultrasonic findings in pregnancy – the use of sequencing and exomes – examples and practice.

21. Evaluation of postnatal cases – the way to approach findings in exomes – examples and practice.

22. Guidelines for cytogenetic and molecular laboratories and quality control in genetic laboratories

23. Single cell level diagnostics - PGD

24. Single cell level diagnostics – PGS + PGD

25. Ethical and philosophical aspects of PGD.

26-28 More bioinformatics practice.

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Clinical Genetics Course Syllabus:

The aim of the course is to give the students an approach to clinical and medical genetics with emphasis on inheritance modes and common syndromes. These subjects are important milestone in everyday work of every genetic counselor.

1. Clinical medicine in the genomic era and principles of the clinical approach for a suspected disease or genetic syndrome.
2. Autosomal recessive and autosomal dominant inheritance - principles of consultation and common diseases (lecture and seminars of students on common diseases).
3. X-linked recessive and dominant modes of inheritance - principles of consultation and common syndromes (lecture and seminars of students on common syndromes).
4. Mitochondrial inheritance - principles of consultation and common syndromes (lecture and seminars of students on common syndromes).
5. Multifactorial inheritance - principles of consultation and syndromes prevalence (lecture and seminars of students on common syndromes).
6. Trinucleotide repeats - principles of consultation and common syndromes prevalence (lecture and seminars of students on common syndromes).
7. Fragile X syndrome.
8. Epigenetics – principles.
9. Microdeletions principles of consultation.
10. Seminars on common syndromes with microdeletions.
11. Biochemical screenings and nuchal translucency as a tool for disease diagnosis in the fetus.
12. Invasive procedures (chorionic villus, amniocentesis, cordocentesis) and non-invasive prenatal testing (NIPT).

13. Dysmorphology, anomalies, malformations, sequence- definitions and clinical approach in counseling.

14. Fetal clinical genetics - Introduction

15. Search engines and clinical databases.

16. Practice with the search engines.

17. Infertility, recurrent miscarriage

18. Regulation and genetic counseling un abnormal biochemical screening and soft signs – what is accepted in Israel.

19. Summary of prenatal genetic counseling - presentation of rolling prenatal clinical cases and using search engines and databases.

20. Genetic evaluation in mental retardation.

21. Genetics of autism.

22. Cystic Fibrosis.

23. Introduction for oncogenetics.

24. Breast and ovarian cancer and gastrointestinal tumors.

25. Malignancy of childhood - bone marrow failure syndromes, neuroblastoma, and Wilm's.

26. "Tumor tailored medicine" - biological treatments.

27. Hearing loss

28. Seminars of students on important syndromes

29. Seminars of students on important syndromes

30. Seminars of students on important syndromes

31. Ataxia and movement disorders (including Huntington's, SCA, SPG, dystonia and recessive disease)

32. Metabolic diseases - storage diseases (lysosomal storage, glycogen storage, mucopolysaccharidoses)

33. Metabolic Diseases - peroxysomal disorders, adrenoleukodysrophy

34. Neurocutaneus syndromes

35. Ciliopathies.

36. Hypotonia - monogenic diseases and syndromes of CNS

37. Hypotonia - muscle nerve diseases (SMA, DMD, Myotonic dystrophy)

38. Nephrotic syndrome, Allport syndrome, nephronophthisis

39. Common multifactorial diseases- celiac disease / IBD / diabetes as a model.

40. Skin diseases

41. Connective tissue diseases – clinic and common syndromes. Marfan syndrome, Loeys-Dietz, Ehlers-Danlos, Osteogenesis Imperfecta, Beals.

42. Eye diseases - anterior eye chamber, posterior eye chamber and the optic nerve

43. Cardiomyopathy and conduction disorders

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Course Syllabus: Imaging Methods and Fetal Medicine

The aim of the course is to give to the students an approach to different imaging methods (ultrasound, echo, MRI) and the way each enable us to diagnose different congenital malformations. Emphasis will be put on the multidisciplinary approach to diagnosis and how does it give us the best way of analyzing the case and giving genetic counseling. In addition, lectures in topics of fetal medicine like: fetal treatment during pregnancy, twin pregnancies, etc. These topics are important in everyday work of a genetic counselor.

1. Introductory lecture: Methods for imaging the fetus
2. Ultrasound during pregnancy monitoring.
3. Nuchal translucency – the meaning and the genetic evaluation.
4. The ultrasonographic evaluation of "soft signs".
5. The genetic evaluation of "soft signs".
6. Congenital heart malformations – the imaging point of view
7. Congenital heart malformations – the genetic evaluation
8. The diagnosis of anomalies in the brain – US and MRI
9. The diagnosis of malformations in the Neural Tube spectrum
10. The genetic evaluation of Central Nervous system.
11. Skeletal dysplasias – The imaging point of view.
12. Skeletal dysplasias – The genetic evaluation.
13. The genetic evaluation of polyhydramnios and olygohydramnios.
14. Sex determination malformations – the imaging point of view.
15. Sex determination malformations – the genetic evaluation
16. TORCH and imaging findings.

17. Limb malformation – the imaging point of view
18. Limb malformation – the genetic evaluation.
19. Congenital malformation of gastrointestinal and abdominal systems.
20. The diagnosis of fetal neoplasia in ultrasounds.
21. The US diagnosis of congenital malformation in the chest cavity.
22. The diagnosis of congenital malformation of the face – the imaging point of view.
23. The diagnosis of congenital malformations of the face – the genetic evaluation.
24. The genetic evaluation of fetal growth disorders – IUGR and LGA
25. The diagnosis of genitourinary and renal malformations – the imaging point of view.
26. The diagnosis of genitourinary and renal malformations – the genetic evaluation.
27. Multiple pregnancies.
28. Invasive procedures and in utero treatments.
29. Fetal aspects of maternal disorders.

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Genetics in the Post-Genome Era

(the course will be delivered in English)

Prof. Karen B. Avraham, Instructor

Introduction to genomics

Discovery of human disease genes

High-throughput sequencing

Bioinformatics for genomics

Transcriptomics

Non-coding RNAs

Human genetic variation

Genome editing

Comparative genomics

Transgenic mouse models for human disease

Personalized/Precision medicine

Direct-to-consumer genetic testing

Pharmacogenomics

Gene therapy