Medical genetics
residency programme

1. General information

Duration of study:	4 years
Name in English:	Medical genetics
General supervisor:	Aavo-Valdur Mikelsaar, Professor of Human Biology and Genetics, University of Tartu, Ravila 19, 50411 Tartu, tel. 374 210, fax 374 212

2. Schedule of cycles and studies

Studies in medical genetics will be undertaken under the direction of the Chair of Human Biology and Genetics in the Department of General and Molecular Pathology of the University of Tartu.

During the first two years, the resident will make a thorough study of the different methods of examining patients which are necessary to diagnose genetic diseases and obtain the information required to provide genetic counselling.

Studies will be undertaken by examining actual patients. There will be a deliberately representative range of patients to be examined (from different hospitals) to enable the resident to gain greater experience of solving genetic problems by examining different forms of disease.

First year of study

Supervisor: Senior lecturer Ruth Mikelsaar.

The resident will work at the Chair of Human Biology and Genetics in the Department of General and Molecular Pathology of the University of Tartu, using the residential centre for young disabled people in Karula in the county of Viljandimaa as a base for examining patients. The resident will be expected to:

- personally carry out clinical examinations of patients with genetic defects and syndromes; acquire the ability to describe the anomalous characteristics of patients and to draw up written medical histories, examination results, diagnoses, differential diagnoses and conclusions; the number of patients examined during the first year is expected to be at least 15;
- acquire the ability to use specific programs, e.g. POSSUM, when diagnosing genetic diseases and syndromes and birth defects, and also the ability to use the large databases of specialist literature in the field of genetics (OMIM, PubMed);
- acquire the ability to prepare chromosome preparations and analyse aberrations in order to make correct diagnoses;
- attend genetics lectures and seminars given for doctoral and master's students (1 credit);
- give at least one presentation at a seminar or conference.

Second year of study

Supervisor: Assistant Gunnar Tasa.
The resident will work at the Chair of Human Biology and Genetics in the Department of General and Molecular Pathology of the University of Tartu, and he/she will now be expected to:

- examine patients using molecular-genetic examination methods, with the aim of learning the methodology to determine mutations causing genetic diseases and acquiring both the ability to use differential diagnostic analysis and an understanding of the significant genetic heterogeneity of such diseases;
- acquire the ability to use DNA separation, sequencing and restriction analysis methods, PCR and electrophoresis methods and the methods for analysing DNA cloning and microsatellites;
- participate in the molecular-genetic research work being undertaken at the Chair and give at least one presentation at a seminar or conference;
- attend the 'Molecular medicine' course aimed at doctoral students (3 credits);
- familiarise him/herself with the molecular-genetic research opportunities at the United Laboratories of Tartu University Hospital (the molecular diagnostics laboratory and the immunological analysis laboratory).

Third year of study

The resident will work at the following clinics or departments of Tartu University Hospital (for two to three months at each):
In the Women's Clinic he/she will take part in clinical and genetic examinations of new-born babies, familiarise him/herself with the use of ultrasound diagnostics to identify fetal defects and with the use of amniocentesis, and learn the method of cultivating amnion cells to analyse fetal chromosomes. The resident will examine patients with problems relating to fertility and sexual differentiation;

In the Children's Clinic he/she will be involved in examining children with genetic diseases and defects and in counselling their families on the defects and diseases;

In the Medical Genetics Centre of the United Laboratories of Tartu University Hospital he/she will be involved in providing genetic counselling to patients referred there for consultation.

During the year the resident will be expected to personally examine or participate actively in the examination of at least 40 patients.

Fourth year of study

The resident will work in the clinics at Tartu University Hospital or in other similar hospitals and departments in Estonia (Tallinn). The work will be organised in cycles of two to three months. The precise schedule and period of work in the different clinics and the appointment of a supervisor will be agreed in advance with the clinic concerned.

The Internal Medicine Clinic, the Neurology Clinic, the Dermatology Clinic or the Eye Clinic (or another department or hospital with a similar profile in line with the

particular wishes of the resident, in the field of psychiatry, urology-andrology, oncology, orthopaedics, etc.).
During the year the resident will be expected to personally examine or participate actively in the examination of at least 40 patients.

In both the third and fourth years of study the resident will remain in regular contact with the Chair of Human Biology and Genetics in order to attend relevant seminars and conferences and also to be able to use the opportunities available at the Chair for assisting the resident in the genetic examinations of the patients for whom he/she is caring in the different clinics.

3. Individual curriculum

It is expected that the resident will have to spend the first two years at the Chair in order to learn the methods of genetic examination and develop skills in genetic analysis. Without this, when working in different hospitals or in departments with different profiles he/she would be unable to identify genetic problems, not to mention not having the ability to plan and/or carry out genetic examinations and to provide counselling for patients and/or their families and also colleagues on genetics-related issues.
With regard to study in the third and fourth years, the time to be spent in the different clinics and the order in which they come can be planned relatively freely. This will be agreed with the heads of the establishments concerned.

This residential programme in medical genetics will prepare a specialist who will be able to perform medical-genetics work with a wide range of patients and forms of disease and to participate as a specialist in national genetics programmes (such as the gene bank), where there is a real need for trained medical geneticists.

One option when drawing up individual curricula would be to supplement the provision of in-depth training on genetics for doctors in other fields, who would in this way be able to add an additional speciality (neurologist-geneticist, paediatrician-geneticist, etc.), and to students having completed their doctoral studies. There are as yet no specific rules for drawing up such curricula.

4. Content of training

4.1. List of the main specialist skills acquired during the residency

– skills in genetic counselling and the relevant ethical principles;
– methods of family screening, the ability to collect and analyse data in a targeted manner and to draw conclusions, and the ability to determine the type of heritability of genetic diseases and to calculate the risk;
– the methodology for the genetic examination of patients, and genetic syndrome diagnostics;
– human chromosome examination methods, karyotype symbols and aberration diagnostics;
– the principle methods in the molecular diagnostics of diseases, DNA separation, PCR, electrophoresis, restriction analysis, blotting, etc.
– an understanding and knowledge of the treatment of genetic diseases and the prospects for gene treatment.

4.2. Subjects covered in the theoretical and practical training

The theoretical bases of human genetics:
– molecular genetics and gene action;
– cytogenetics in cell cultures, the normal structure of chromosomes with differential colouring, anomalies in the structure and number of chromosomes, the reasons for this and the consequences;

– tumour and molecular cytogenetics;
– the most significant metabolism diseases, their genetic causes and action, clinical pictures, biochemical bases, biochemical diagnostics;
– the effect of exogenous toxins before (mutagenesis) and during (teratogenesis) pregnancy;
– mathematical treatment of genetic heritability in populations (population genetics) and families (linkage analysis), with the criteria for genetic screening.

Identification of genetic risks:
– risk calculations in the case of monogenic diseases, on the basis of family-tree data;
– principles of empirical risk calculation in the case of multifactorial diseases;
– risk of repetition in the case of chromosome aberrations;
– risks relating to exogenous toxins before and during pregnancy;
– risk calculations on the basis of molecular-genetic markers.

Basics of genetic counselling, including predictive DNA diagnostics taking account of the psychological and ethical aspects.
Principles of treating diseases of genetic origin.
Legal bases for genetic counselling and diagnostics; data protection, biological reliability, radiation protection and organisation of work.

5. Amount of experience to be acquired as a doctor

This is expressed in terms of genetic counselling having been provided independently in the required number of cases and for diseases of genetic origin in all fields of medicine. Altogether there must be at least 100 documented cases involving at least 30 different genetic problems or diseases: these will include the risk of repetition, the prognosis and a description of the nature of the disease for the person in need which will, where necessary, be drawn up in writing for that person and the doctor treating him or her.

6. Independent work

The resident's independent work will consist of clinically examining patients and using cytogenetic and molecular-genetic methods to explain the genetic causes of diseases. Work with specialist literature, the drawing up and presentation of abstracts and the presentation of scientific papers at seminars and conferences will also be considered as independent work.
Primary literature:
Korf Bruce R.: Human Genetics. A problem-based approach. Blackwell Science, 1996
Strachan T., A.P.Read: Human Molecular Genetics. BIOS, 1999
Mueller R.F., I.D.Young: Emery's elements of Medical Genetics. Churchill Livingstone, 1995
Gelehrter T.D., F.S.Collins, D.Ginsburg: Principles of Medical Genetics. Williams&Wilkins, 2nd. ed., 1997

7. Amount of theoretical study and the ways in which this is carried out

As far as the speciality is concerned, the resident will be required to attend the series of lectures organised for doctoral and master's students on human genetics (1 credit) and the human molecular medicine course (3 credits). The resident will also be expected to attend genetics-related lectures given at both the Faculty of Medicine and the Faculty of Biology and Geography in Tartu by outside lecturers and our experts, and to attend other courses.

8. Preliminary assessments

Preliminary assessments will take place in the presence of the relevant supervisor(s) and the general supervisor. The resident will present a written report of the work he/she has done during the year.
The first year will be deemed to have been passed successfully if the resident has personally carried out clinical and cytogenetic examinations of at least 15 patients and is able to submit medical histories in the appropriate form. He/she must be able to prepare chromosome preparations, analyse karyotypes and make cytogenetic diagnoses. He/she must be able to use computer programs (POSSUM, etc.) as tools for diagnosing genetic diseases and computer databases for obtaining additional information on those diseases. He/she must have given at least one significant presentation at a seminar or conference.

The second year will be deemed to have been passed successfully if the resident is able to present written reports showing that he/she is familiar with and able to use the following molecular-genetic examination methods: the different DNA isolation methods, DNA amplification by the polymerase chain reaction (PCR), agarose and polyacrylamide gel electrophoresis methods, restriction analysis, microsatellite analysis, DNA sequencing, the main techniques in cloning DNA, and molecular-genetic identification. The proof of being able to use these methods will be reports on examinations undertaken to solve specific problems (at least 10). The resident must have given at least one significant presentation at a seminar or conference during the year.

The third and fourth years will be deemed to have been passed successfully if the resident has personally carried out and/or been actively involved in genetic examinations and consultations with at least 40 patients and is able to submit medical histories in the appropriate form. In addition, the resident must present proof that he/she has given at least one significant presentation at a seminar or conference during the year and has participated in theoretical studies in which a speciality can be acquired (e.g. courses on clinical immunology, pharmacology, psychology, etc.).

9. Final exam of the residency

The final exam will be oral but will include a written element. In order to take the exam, all four years must have been passed successfully. The resident must bring to the exam all the examination records on the patients he/she has examined during the residency.