MEDICAL GENETICS

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Objectives
In order to be able to practise as a medical geneticist a specialist must have the following knowledge and skills:

- A knowledge of the structure of the human genotype and the effect of genetic mutations as a cause of disease.
- A knowledge of modes of inheritance and the ability to carry out the genealogical research needed for assessing the heritability of a disorder and on that basis estimate the heritability of the disorder in a family.
- Mastery of the most important cytogenetic and molecular genetic methods and the ability to apply them in the diagnosis of hereditary disorders.
- A knowledge of the most common hereditary disorders in different branches of medicine and the ability to diagnose rare hereditary disorders and to organise diagnostic tests for them.
- A knowledge of pre-natal diagnostic techniques and practices in Finland.
- An ability to provide genetic counselling, i.e. to give patients an explanation of the main characteristics of a disorder, the mode of inheritance, the genetic background and risk of recurrence so that the patients themselves can make the best use of this information.
- An ability to understand questions relating to the hereditability of a disorder from the point of view of the patient and his or her immediate and extended family and to support patients suffering from hereditary disorders.
- A knowledge of legislation in this field and the fundamental ethical issues and principles of genetic medicine and an ability to apply these principles in their work.
- Familiarity with the current state of research in medical genetics and an ability to provide expertise in matters relating to rare and hereditary disorders in various specialisms.
- An ability in their own work environment to arrange further training for other staff members in this domain and manage genetic information on the local population.

STRUCTURE OF THE TRAINING COURSE
The course lasts a total of five years.

Health centre placement (9 months)

Specialised training (4 years 3 months)

The medical genetics training includes clinical patient care in HUSLAB's Medical Genetics Department and in the Department of Foetal Research at the HUS (Hospital District of Helsinki and Uusimaa) Women's Clinic, as well as a laboratory placement related to medical genetics in the molecular genetics and cytogenetics laboratory serving the HUSLAB Medical Genetics Department or the molecular pathology laboratory of HUSLAB's Pathology Department.
The course is divided up as follows:
- At least 30 months of clinical patient care in the Department of Medical Genetics,
  of which 4-6 months in pre-natal diagnostics.
- A total of 8-12 months of laboratory diagnostics related to the field of medical genetics,
  of which at least 4 months in molecular genetic diagnostics and
  at least 4 months in cytogenetic diagnostics.
- A 9-month elective placement
  of which it is recommended that 6 months be spent on a hospital placement in one of the following fields: paediatrics, paediatric neurology, neurology, internal medicine or obstetrics and gynaecology.

There are four training positions in the medical genetics programme: two full-time hospital doctor positions serving HUSLAB's Medical Genetics Department and two part-time posts in medical genetics at Helsinki University, one for a PhD student and one for a postdoctoral researcher with the possibility of a part-time position in the HUS. A placement in these part-time positions will be recognised as a clinical or laboratory training placement, depending on the personal training programme. A clinical placement in part-time position will be recognised as specialised medical training equivalent to no more than one year's work as a full-time hospital doctor. Three months of the clinical patient care requirement may be filled by working in the genetics clinic of the

Väestöliitto (Family Federation).
Academic work in Finland or abroad, for example writing a thesis whose subject and methods are closely related to medical genetics, may count for up to 6 months of the elective part of the specialist medical training, with the consent of the training coordinator. The trainee should discuss what training is acceptable with the specialised medical training coordinator.

On-the-job training
Practical work is supplemented by regular structured on-the-job training opportunities and meetings with patients, intended to provide hands-on training experience or to develop a better understanding of the specialism. On-the-job training is organised for 2-4 hours per week.

Teaching hospitals and instructors: http://www.med.helsinki.fi/erikoislaakari

Theoretical training course (80 hours)
Because of the rapid development of genetic research, the theoretical training is a particularly important part of medical genetics. The theoretical training must comprise as many wide-ranging advanced courses in genetics as possible (rather than one-off lectures and talks), attendance at a school of medical genetics (e.g. European School of Medical Genetics) or participation in congresses abroad at least once during the training period (e.g. the American Society of Human Genetics and the European Society of Human Genetics).

Face-to-face management training (30 credits)

The management portfolio provides a training framework, containing a record of the trainees' personal objectives, their own observations drawn from the workplace and an analysis based on them, comments on meetings with supervisors, feedback received and study assignments and results under the training programme. Administrative training that forms part of the postgraduate training in primary health care will count towards this training. The training also includes preparation of a written presentation, a practical exercise and best practice.

The course also includes three two-day face-to-face teaching sessions common to all specialisms and at least two face-to-face specialised teaching days (or 4 afternoons), making a total of 10 credits, the preparation of self-development assignments and a written assignment. The training can be completed over a period of 2-6 years.

10 face-to-face training days = 10 credits	***Development assignments 10 credits***	***Written assignments 5 credits***	***Portfolio 5 credits***

For more information see: www.med.helsinki.fi/erikoislaakari/johtamiskoulutus and http://blogs.helsinki.fi/lahijohtajakoulutus.

Monitoring the learning process and assessment tools
In the assessment process we try to follow principles approved by the faculty. An annual assessment is carried out during the training period. Both the number of people specialising in medical genetics and the number of teachers in this field are small, which makes it easier to develop good personal contacts between teacher and trainee specialists. The fact that the majority of the training takes place in the same department also contributes to this. The department also tries to find a personal tutor for doctors undergoing specialist training.

National examination
Books (latest edition)
1. Rimoin D.L., Connor J.M., Pyeritz, R.E. & Korf B.R. (Eds.): Emery and Rimoin´s principles and practice of medical genetics, Vol. 1–3 (Churchill Livingstone)
2. Harper P.: Practical genetic counselling (Butterworth-Heinemann)
3. Gardner R.J.M & Sutherland G.R.: Chromosome abnormalities and genetic counselling (Oxford Univ. Press)
4. Strachan T. & Read A.P.: Human molecular genetics (BIOS Scientific Publ.)
5. Hodgson S.V., Foulkes W.D., Eng C. & Maher E.R.: A practical guide to human cancer genetics (Cambridge Univ Press)

Journals (journals dating from the three years preceding the examination)
1. American Journal of Human Genetics
2. Journal of Medical Genetics
3. Nature Genetics
4. European Journal of Human Genetics

Relevant aspects of medical law.