Specialised subject: Medical Genetics

Course content (major subject)

A) Knowledge	
1	General principles of medical biology
1.1	Cell biology, with special emphasis on human cell biology
1.2	Molecular biology, with special emphasis on the structure and function of human genes
1.3	Reproductive and developmental biology in humans
2	General principles of medical genetics
2.1	Fundamentals of human genetics, with special emphasis on formal genetics, population genetics and on clinical genetics and syndromology
2.2	Pre-natal and post-natal cytogenetics, tumour cytogenetics with cell cultivation, differential display of chromosomes with banding techniques and fluorescence in situ hybridisation. Analysis and interpretation of karyotypes. Causes and effects of numerical and structural inherited and acquired chromosome anomalies
2.3	The most important genetic disorders and (where known) their molecular basis and molecular genetic means of detecting them
2.4	The most important metabolic diseases, their genetic causes and effects, their symptoms, biochemical principles and biochemical means of detecting them
2.5	Effect of external noxious factors before (mutagenesis) and during (teratogenesis) pregnancy, and in particular gametogenesis, embryonic and foetal development, including any possible effects of external noxious factors
2.6	Research into mutagenesis with a special emphasis on the causes of mutation and how they are tested for
2.7	Evolutionary theory with a special emphasis on evolution in humans
2.8	Immunogenetics
3	Ecology and ecotoxicology with an emphasis on ecogenetics and pharmacogenetics
4	Genetic statistics, epidemiology and population genetics; principles of quantitative genetics
5	Legislation relevant to practising physicians, and in particular legislation concerning social welfare and healthcare, including knowledge of the institutional system - the Austrian healthcare and social insurance system - basic legal principles relating to documentation and medical malpractice-

	cooperation with other healthcare professions
6	Basic principles of multidisciplinary coordination and cooperation, and in particular guidance on social services, institutions and possibilities for rehabilitation

B) Skills and experience	
* indicative number of analyses, diagnoses, decisions, counselling sessions performed and for which documented proof can be provided	
1	Giving a medical genetic diagnosis of genetic disorders, inherited anomalies and anomaly complexes, including a written, detailed and critical interpretation of the differential diagnosis. Documented proof from the genetic counselling service or institutional advisory service of at least 20 different diagnoses 100*
2	Establishing and processing cell cultures. Conducting chromosome analyses by means of all the relevant techniques, including differential chromosome staining and molecular cytogenetic techniques, karyotype analysis and interpretation Post-natal: 200* Pre-natal: 100* (These cases should include 40 chromosomal aberrations, 10 of which should be structural aberrations) The chromosome analyses also involve identifying and assessing risks of chromosomal disorders as well as providing a detailed epicritic assessment and differential diagnosis for the attendant physicians
3	Giving molecular genetic diagnoses of genetic disorders involving different types of heredity either by detecting gene mutations directly or by means of indirect genotyping for at least three loci with different degrees of detection method difficulty. This also involves providing a detailed epicritic assessment and differential diagnosis for the attendant physicians as well as specialist reports from counselling sessions with 150* patients, with at least 20* cases involving family history
4	Calculating the risk of monogenic disorders on the basis of pedigree data in families with autosomal dominant and recessive inheritance as well as X-linked recessive inheritance, and in families with multifactor genetic disorders as well as on the basis of coupled marker inheritance.
5	Providing genetic counselling and advice
5.1	Basic principles of genetic counselling, including ethical and psychological aspects, and in particular the concepts involved in genetic counselling and the technique of interviewing; this includes theory/practice seminars
5.2	Providing genetic counselling and advice in the field of genetic disorders in general, as well as those caused by noxious teratogenic agents, for at least 30* different disorders, in each case taking into account the relevant differential diagnoses and creating a family medical history over three generations, as well

	providing as a detailed written epicritic assessment for the attendant physicians and report of the counselling session for the person seeking advice. Counselling sessions may be held in conjunction with diagnosis and risk calculation activities (i.e. for the same patients/families). 200*
6	Conducting quality assurance and documentation activities specifically tailored to the discipline in question
7	Providing written summaries, documentation and assessments of how disorders progress and the resulting prognoses (ability to issue certificates, reports, etc.)

Appendix 21

Specialised subject: Medical Genetics

A. Definition of subject area

The specialised subject of Medical Genetics covers the diagnosis of genetic disorders, the determination of the risk of occurrence of disorders, the counselling of patients and their families on genetic issues, as well as pure research in the specialised subject area and applied research, particularly through the application of cytogenetic, biochemical and molecular genetic techniques and through the application of knowledge about the course and patterns of biological functions in humans, the aetiology and pathogenesis of genetic disorders, general human genetics, cytogenetics, molecular genetics, dysmorphology, clinical genetics including syndromology, population genetics and genetic epidemiology.

B. Minimum duration of training and training subjects

1. Major subject: four years

2. Mandatory minor subjects:
2.1. Internal Medicine: six months

3. Optional minor subjects:
3.1. Optional minor subjects where restrictions apply:
One year in one or more of the following specialised subjects, where each subject selected must be followed for a period of at least three months:
Ophthalmology and Optometry, Blood Group Serology and Transfusion Medicine, Gynaecology and Obstetrics, Otolaryngology, Dermatology and Venereology, Internal Medicine, Paediatric Surgery, Paediatrics, Paediatric Psychiatry, Neurology, Orthopaedics and Orthopaedic Surgery, Plastic, Cosmetic and Reconstructive Surgery, Psychiatry, Urology
3.2. Optional minor subjects where no restrictions apply:
Six months in one or more elective specialised subjects, where each subject selected must be followed for a period of at least three months