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Training Requirements for the Specialty of Medical Genetics

European Standards of Postgraduate Medical Specialist Training

Preamble

The UEMS is a non-governmental organization representing national associations of medical specialists at the European level. With a current membership of 39 national associations and operating through 43 Specialist Sections and European Boards, the UEMS is committed to promote the free movement of medical specialists across Europe while ensuring the highest level of training which will pave the way to the improvement of quality of care for the benefit of all European citizens. The UEMS areas of expertise notably encompass Continuing Medical Education, Post Graduate Training and Quality Assurance. It is the UEMS' conviction that the quality of medical care and expertise is directly linked to the quality of training provided to the medical professionals. Therefore the UEMS committed itself to contribute to the improvement of medical training at the European level through the development of European Standards in the different medical disciplines. No matter where doctors are trained, they should have at least the same core competencies.

In 1994, the UEMS adopted its Charter on Post Graduate Training aiming at providing the recommendations at the European level for good medical training. Made up of six chapters, this Charter set the basis for the European approach in the field of Post Graduate Training. With five chapters being common to all specialties, this Charter provided a sixth chapter, known as "Chapter 6", that each Specialist Section was to complete according to the specific needs of their discipline. More than a decade after the introduction of this Charter, the UEMS Specialist Sections and European Boards have continued working on developing these European Standards in Medical training that reflects modern medical practice and current scientific findings. In doing so, the UEMS Specialist Sections and European Boards did not aim to supersede the National Authorities' competence in defining the content of postgraduate training in their own State but rather to complement these and ensure that high quality training is provided across Europe.

At the European level, the legal mechanism ensuring the free movement of doctors through the recognition of their qualifications was established back in the 1970s by the European Union. Sectorial Directives were adopted and one Directive addressed specifically the issue of medical training at the European level. However, in 2005, the European Commission proposed to the European Parliament and Council to have a unique legal framework for the recognition of the Professional Qualifications to facilitate and improve the mobility of all workers throughout Europe. This Directive 2005/36/EC established the mechanism of automatic mutual recognition of qualifications for medical doctors according to training requirements within all Member States; this is based on the length of training in the Specialty and the title of qualification. Given the long-standing experience of UEMS Specialist Sections and European Boards on the one hand and the European legal framework enabling Medical

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Specialists and Trainees to move from one country to another on the other hand, the UEMS is uniquely in position to provide specialty-based recommendations. The UEMS values professional competence as "the habitual and judicious use of communication, knowledge, technical skills, clinical reasoning, emotions, values, and reflection in daily practice for the benefit of the individual and community being served"1. While professional activity is regulated by national law in EU Member States, it is the UEMS understanding that it has to comply with international treaties and UN declarations on Human Rights as well as the WMA International Code of Medical Ethics.

This document derives from the previous Chapter 6 of the Training Charter and provides definitions of specialist competencies and procedures as well as how to document and assess them. For the sake of transparency and coherence, it has been renamed as "Training Requirements for the Specialty of X". This document aims to provide the basic Training Requirements for each specialty and should be regularly updated by UEMS Specialist Sections and European Boards to reflect scientific and medical progress. The three-part structure of this documents reflects the UEMS approach to have a coherent pragmatic document not only for medical specialists but also for decision-makers at the National and European level interested in knowing more about medical specialist training.

Clinical Genetics is the medical specialty concerned with the medical elements of Genetic Services provided to individuals and families (and sometimes populations) with, or at risk of, diseases, syndromes and conditions which have, or may have, a genetic basis. This includes the provision of diagnostic and genetic counselling services, information about the condition and its implications, including management and follow-up, prognosis, screening, prevention and reproductive options, and therapeutic possibilities. This is based on thorough clinical assessment, family (pedigree) medical information, conventional laboratory investigations and imaging, and specialized genetic tests and their interpretation. Other components of Genetic Services include laboratory genetics (cytogenetics, molecular genetics and genomics, biochemical genetics), specialized genetic counselling, the provision of a knowledge and skills resource for all other medical disciplines, teaching and research. The core activities of Genetic Services can be defined as: 'integrated clinical and laboratory services, provided for those with, or concerned about, a disorder with a significant genetic component (both inherited and sporadic). Due to the sharing of genes and DNA variation among family members, the whole family, not only the affected individual, represents the core patient in Clinical/Medical/Human Genetics.

Clinical Genetics is a rapidly developing medical specialty due to scientific advances in DNA sequencing technologies. The future of European Clinical Genetics will depend on the equitable expansion of the specialty, the quality of training offered to trainees, and the resources to integrate advances in genomics into clinical care. This document relates to individuals with medical qualifications who seek to train in the specialty of Clinical Genetics. It is recognized that there may be areas of overlap with training programs for other genetic professionals (laboratory geneticists and counsellors) and that there may be opportunities for periods of joint training during specialization.

Rare and undiagnosed diseases constitute an important area of expertise for Clinical Geneticists as the large majority of these diseases have a genetic origin. Currently, it is estimated that for every 1,000 people, one new patient has a diagnosis of a rare disease per annum. Considering a caseload of about 100 rare disease patients / year for each geneticist, 1 geneticist per 100,000 population is required – and this relates only to the manpower needs for *rare* diseases. In view of the growing involvement of clinical geneticists with common cancers (ovary, breast, colorectal), inherited cardiac conditions, fetal medicine teams (prenatal genetics) and assisted reproduction/preimplantation genetic diagnosis, the estimated manpower needs are 1 clinical geneticist for every 50,000 population

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Aims of Clinical and Medical Genetics (further essential parts of the ETR are described in the supplementary "Description of the specialty" and "Syllabus"):

- 1. To provide a service whose goal is to assess, investigate, and diagnose diseases and medical conditions that are genetic and/or hereditary, or *may be* genetic and/or hereditary.
- 2. To provide a service that provides specialist information about genetic and/or hereditary disease, including recommendations for screening where appropriate.
- 3. To provide a service that investigates and offers counselling in relation to reproductive options and prenatal genetics.
- 4. The prevention of genetic and/or hereditary disease, and serious disability, according to the choice made by those at risk of having affected offspring, based on full information and expert counselling.
- 5. To contribute to the management of patients and families affected by genetic disease, in collaboration with other medical specialists, including treatment.
- 6. To be advocates, where necessary, of those affected by genetic and/or hereditary diseases and conditions, most of which are rare disorders.
- 7. To conduct and contribute to clinical and genomic research to enhance knowledge of the causation and natural history of genetic and/or hereditary diseases and conditions.
- 8. To teach and instruct medical undergraduates and postgraduates in clinical genetics and genomics, in order to raise the knowledge base across all medical specialties.
- 9. To provide a knowledge and skills resource to all medical specialties, including through multidisciplinary meetings.
- 10.To contribute to the public understanding of genetics and genomics, and their role in health and disease.

I. TRAINING REQUIREMENTS FOR TRAINEES

1. <u>Content of training and learning outcome</u>

Clinical Genetics is a field of Medicine concerned with the investigation, diagnosis, treatment, prevention, and research into inherited and somatic disorders caused by variation in the human genome. The scope of patient care activities includes the recognition of discrepancies leading to abnormalities of the genome and the developing pathological processes, the early identification of individuals and families at risk, the identification of the genetic defect and the preventive care of affected family members, and prevention of intellectual and physical disability in those born with genetic disorders, in addition to the rehabilitation of such patients.

This specialty training is aimed at giving doctors qualifications in the field of Clinical Genetics to enable them to treat patients with genetic diseases and their families in the light of current and expanding knowledge on the subject, with particular emphasis on understanding the molecular and cellular pathogenic mechanisms of such diseases, and their diagnosis and treatment. Clinical Geneticists must also be able to carry out screening for the early identification of individuals and families with a high risk of contracting common diseases with a major social impact (malformations in general, familial cancers, inborn errors of metabolism, etc.).

Theoretical knowledge:

- Fundamental knowledge of hereditary diseases;
- Molecular biology and organization of genes (including transcription, translation and relevant regulatory mechanisms, mutagenesis and repair mechanisms, mechanisms of gene editing); variation of the human genome and post-translation changes of gene products; relation with other analytical results and their clinical correlation;
- Recognition of indications for individual genetic consultation;
- Genetic diagnostic tests with conventional cytogenetic, molecular cytogenetic and molecular genetics;
- Reasons for medical genetic testing; effects of genetic defects; effects of structural and numeric chromosome aberrations (including malignant illnesses); possibilities, methods and risks of prenatal diagnosis;
- The concepts of dominance, recessivity, sex linkage, penetrance, sex limitation, pathogenicity of mutations, phenocopies, and variable expressivity; polygenic disorders;
- Mutational mechanisms: somatic, constitutional and germline; gender differences;
- ISCN (International System for Human Cytogenetic) nomenclature; HGVS (Human Genome Variation Society) nomenclature; genetic database standards; quality assurance; internal and external quality control;
- Direct gene diagnostics/analysis for differential diagnosis; direct gene diagnostics/analysis for determination of carrier status; direct gene diagnostics/analysis for pre-natal and preimplantation diagnosis/analysis; pre-symptomatic gene diagnostics/analysis; Gene Banks;
- Analysis of the following abnormalities and recognition of their diagnostic, prognostic and/or therapeutic interest: chromosome number abnormalities; mosaicism; structure abnormalities in equilibrium and disequilibrium; chromosomal micro-rearrangement; identification of a chromosomal marker; identification of chromosomal variants; chromosome fragility and chromosome breakage syndromes.
- Analysis of the following pathological types and recognition of their diagnostic, prognostic and/or therapeutic interest: monogenic disorders (autosomal dominant, autosomal recessive, X-linked recessive, X-linked dominant).
- Oligo- and polygenic disorders; mitochondrial disorders, imprinting disorders, anticipation disorders

Competencies required of the trainee

Clinical Geneticists need a wide range of clinical skills, as genetic disorders can affect people of all ages and involve all body systems. Communication skills are particularly important in explaining complex concepts and genetic test results to families, in order to enable them to make informed decisions and choose appropriate course(s) of action.

Clinical Geneticists have an important role in public education and public debate about ethical, social, and other diverse issues that arise from new developments in the clinical application of genetic/genomic knowledge. Theoretical genetics, medical knowledge and specialist-level clinical skills, genetic counselling and communication skills, laboratory skills, and ancillary competences, are specifically described in the Syllabus (attached).

The specialty of Clinical Genetics requires assessed and documented episodes for 'Practical Skills'. The numbers in parentheses are minimum numbers of tests/ interventions/ approaches which the

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candidates have to perform in order to be eligible for exam. This means that the candidates have to have technical skills to make them eligible to prepare and sign diagnostic and laboratory reports. They have to be able to perform these tests. They have to have the capability to supervise technical staff's work as well as counsellors. They have to have the capability supervise the counsellor in taking care for patients.

Cytogenetics

- 1. Sampling and transport; preparation, application and carry through cell cultures
- 2. Cultivation of peripheral blood (lymphocytes) (10)
- 3. Preparation of chromosomes (10) according to standard methods and using synchronization techniques
- 4. Staining of chromosomes for banding (the acquisition of the principal types of chromosomal bands); knowledge of the resolution level of the chromosomal bands
- 5. Karyotype; knowledge of the different analyzable materials and the culture requirements; validity criteria of chromosomal analysis
- 6. Microscope analysis of metaphase chromosomes
- 7. Karyotyping using digital analyzing software (30)
- 8. Describing karyotypes (20), verification of numeric and structural chromosomal aberrations
- 9. Diagnosing mosaicism (5)
- 10. Identification of chromosomal micro-rearrangement (5)
- 11. Identification of a chromosomal marker (3)
- 12. Chromosome fragility and chromosome breakage syndromes (5)
- 13. Indication of genetic tests in infertility, congenital malformations and oncohaematological diseases (40)
- 14. Diagnosing with FISH method, evaluation and interpretation (50), centromeric and telomeric probes, specific probe of a locus on metaphase or interphase
- 15. The indication and evaluation of multicolor-FISH method (10)
- 16. The regulation of biobanking, the method of archiving samples
- 17. The regulation of data management
- 18. CGH, validity criteria of molecular cytogenetics assays/analysis

Molecular Genetics

- 1. Sampling, specimen-transport and specimen-treatment
- 2. Extraction and preparation of DNA and RNA (genomic DNA, RNA, RNApolyA+) (DNA isolation from peripheral blood, tissue (fibroblast, saliva, etc.), chorionic villus sample, amniotic fluid)
- 3. Storage of nucleic acids
- 4. Cloning of nucleic acids
- 5. MLPA (20)
- 6. Sanger-sequencing (50)
- 7. Massive parallel sequencing (next generation sequencing) (10) associated bioinformatics, including international databases
- 8. CGH (30) (array technology)
- 9. PCR (10)
- 10. Reverse PCR

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- 11. PCR RFLP (20)
- 12. Real-time quantitative PCR (20)
- 13. Fluorescence PCR (40)
- 14. Molecular cytogenetics (FISH)
- 15. Different methods of study/analysis of DNA polymorphisms (SNP, microsatellites)
- 16. Fragment analysis (10)
- 17. Linkage analysis
- 18. Southern and Northern blotting
- 19. Labelling of probes
- 20. Different methods of gene expression study at RNA level
- 21. Validity criteria of molecular genetics analysis
- 22. The interpretation of mutations/pathological variations using international classification and using adjunctive data from e.g. other fields of pathology and laboratory medicine
- 23. Verification of mutation
- 24. Genome-wide association studies (GWAS)
- 25. The interpretation of multiple mutations/pathological variations and SNPs in one individual in the diagnosis, assessment of risk and therapeutic options for patients
- 26. Epigenetics
- 27. Pharmacogenetics
- 28. Biochemical genetics and metabolic disease
- 29. Biobanking

Clinical and Counselling Skills

- 1. Construct and analyze a detailed family pedigree (100)
- 2. Take detailed medical history of affected individuals (100)
- 3. Take detailed medical history of family members and have ability to confirm clinical diagnoses (30)
- 4. Perform clinical examination of all body systems with recognition and interpretation of abnormal clinical signs (30)
- 5. Ability to perform sample collection such as venipuncture and skin biopsy (30)
- 6. Ability to provide a definitive diagnosis or list of differential diagnoses for unidentified genetic disorders (20)
- 7. Ability to provide detailed phenotypic descriptions (20)
- 8. Indication for and appropriate use of genetic tests and evaluate results: biochemical, cytogenetic and molecular (50)
- 9. Expertise in interpretation of genotype/phenotype data (20)
- Ability to clearly explain complex genetic concepts and results to individuals and families (20)
- 11. Manage the interpretation and implications of genetic test results for the wider family (20)
- 12. Advise on natural history, variability and prognosis of genetic disorders (20)
- 13. Input into general patient management, including medical surveillance, treatment, rehabilitation, educational and social care (10)
- 14. Recognition of the most common genetic disorders (20)
- 15. Investigation and counselling for infertility (20)
- 16. Genetic assessment, diagnosis and counselling for hereditary cancers (20)
- 17. Genetic assessment, diagnosis and counselling for learning disability and dysmorphic

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syndromes in children, including use of syndrome databases or atlases for diagnosis (20)

- 18. Genetic assessment, diagnosis and counselling for neurogenetic disorders (20)
- 19. Genetic assessment, diagnosis and counselling for genetic sensory disorders (20)
- 20. Genetic assessment, diagnosis and counselling for genetic disorders affecting all other body systems, including, but not limited to , inherited renal, dermatological, connective tissue and skeletal disorders (20)
- 21. Diagnosis and analysis of genetic factors implicated in common disorders with polygenic ethiology (10)
- 22. Counselling for prenatal diagnosis (30)
- 23. Fetal ultrasound examination together with gynecologist, indications, interpretation of abnormal findings and counselling (30)
- 24. Indications and use of chorionic villus sampling and placental biopsy (20)
- 25. Indications and use of amniocentesis (20)
- 26. Chromosome abnormalities and their detection by karyotyping, FISH and Q-PCR (10)
- 27. Fetal diagnosis by biochemical methods (10)
- 28. Fetal diagnosis by direct gene testing (10)
- 29. Analysis of prenatal array CGH analysis (10)
- 30. Counselling and organization of non-invasive prenatal testing for fetal genetic disorders performed on fetal cell-free DNA (5)
- 31. Counselling and organization of preimplantation genetic diagnosis (5)
- 32. Counselling and management of genetic testing for carrier detection (10)
- 33. Counselling, management and follow-up for predictive genetic testing (10)
- 34. Organization of medical screening tests for high risk individuals (10)
- 35. Participation and coordination of multidisciplinary team meetings with other medical specialties, such as oncology, immunology, endocrinology, radiology and biochemistry
- 36. Application of pharmacogenetics and personalized medicine
- 37. Application and participation in newborn and population based screening tests for genetic disorders
- 38. Application of bioinformatics in diagnosis of sequencing results (genome, exome and gene panels)
- 39. Expertise with clinical, cytogenetic and molecular genetic databases
- 40. Knowledge of legal issues relating to genetics
- 41. Understanding and discussion of ethical implications related to genetic disease and its management
- 42. Ability to maintain appropriate databases
- 43. Ethics, patient rights and protection of privacy relating to clinical practice and research
- 44. Knowledge of research protocols and procedures
- 45. Ability to take informed consent, including consent for testing children, adults lacking capacity and individuals participating in research
- 46. Participation in clinical trials
- 47. Contribution to teaching, training, research and service development

Genomic Medicine

- 1. The practice and use of massive parallel sequencing (Next Generation Sequencing, -NGS) and associated bioinformatics, including international databases
- 2. Gene dosage techniques based on NGS

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- 3. The advantages and disadvantages of single gene vs. gene panels vs. whole exome vs. whole genome analysis
- 4. The interpretation of multiple mutations/pathological variants and SNPs in one individual in the diagnosis, assessment of risk and therapeutic options for patients
- 5. Pharmacogenomics
- 6. Epigenomics
- 7. Biochemical genetics and metabolic disease
- 8. The role of genomic testing for somatic mutations/pathological variants in tumors, and how this informs treatment

2. Organization of training

a. Schedule of training

The optimal Clinical Genetics training is 5 years consisting of 1 year of common trunk and 4 years training in Clinical Genetics Centre in an accredited program. The training of 4 years is also accepted. However, those countries that have a 4 year-course must arrange a preliminary general training, covering medicine and pediatrics if possible, before, and separate from, the 4 year specialist training. The key purpose of this is the acquisition of core clinical skills. Depending on national regulations, the training may start immediately after completion of medical school, or be preceded by an internship. Trainees must maintain an accurate logbook of their training and rotations.

Optimal training would be:

- 1 years common medical trunk training including some of the following: general practice, pediatrics (including pediatrics neurology ward), obstetrics and gynecology, neurology and psychiatry, internal medicine, and rehabilitation.
- 4 years specialty orientated practice: including cytogenetic training (8 months), molecular genetics training (4 months), oncology (4 months), and practical genetic counselling (8 months).
- if the candidate already had a board exam, the training in only 4 years, repetition of the 1 year common trunk is not necessary.
- if the candidate had PhD qualification of a field that includes genetic (molecular genetic) laboratory training, the duration of the 4 years training is reduced to 3 years.

b. Curriculum of training

The general aim of the training program is to enable the clinical geneticist to work effectively as a consultant. The trainee must demonstrate the ability to record and convey patient details of history, examination and investigation findings to senior staff. The trainee must communicate effectively with patients and relatives, and be able to pass on both technical information in a way that it can be received with understanding, and distressing information in a sensitive and caring manner.

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c. Assessment and evaluation

Countries will use assessment strategies appropriate to their needs. In due course there will be a move to a common approach to determining whether an individual is suitable to be recognized as a 'European medical specialist with additional clinical genetics competence'. Thus, there will need to be an assessment of knowledge, through a form of written examination, possibly online. This examination would use scenarios from an agreed list of core clinical conditions and test knowledge in the areas of relevant science and clinical practice (diagnosis, investigation, interpretation, prevention and treatment). This assessment may take the form a 'best of five' (multiple choice) format, but has yet to be decided.

d. Governance

The governance of an individual's training program will be the responsibility of the Program or Course Director and the institution(s) in which the training program is being delivered. A trainer (who will have satisfied the requirements laid out below, Section II) will be responsible to the Program Director for delivering the required training in their area of practice.

II. TRAINING REQUIREMENTS FOR TRAINERS

1. <u>Process for recognition as trainer</u>

a. Requested qualification and experience

Trainers should be certified clinical geneticists and must be recognized by the national authority. Trainers should provide evidence of academic activities (clinical and/or basic research, publications in peer reviewed journals and participations in clinical genetic scientific meetings) and professional experience. They should possess the necessary administrative, communicative, teaching and clinical skills and commitment to conduct the program. Trainers and Training Program Directors must be in active clinical practice and engaged in training in the training center. Training Program Director must be a certified specialist for a minimum of 5 years. He/she organizes the activities of the educational program in all institution that participate in the program.

b. Core competencies for trainers

- 1. Familiar with all aspects of Clinical Genetics.
- 2. Experienced in teaching and in supporting learners.
- 3. Trained in the principles and practice of medical education.
- 4. Act as a lecturer to a peer-audience on a regular basis, attend national meetings and able to demonstrate appropriate participation in continuing professional development.
- 5. Able to recognize trainers whose professional behavior is unsatisfactory and initiate corrective and supportive measures as needed.

2. <u>Quality management for trainers</u>

Trainers and Program Directors will have their job description agreed with their employer which will allow them sufficient time for support of trainees. Feedback from trainees is necessary for optimal training.

The educational work of trainers and Program Directors will be appraised no less than an annual basis within their Institution as local circumstances determines.

III. TRAINING REQUIREMENTS FOR TRAINING INSTITUTIONS

1. <u>Process for recognition as training center</u>

a. Requirement on staff and clinical activities

A training center is a place, or number of places, where trainees are able to develop their clinical genetics competences. Thus, training may take place in a single institution, or in a network of institutions working together, to provide training in the full spectrum of clinical conditions and skills detailed in the curriculum. A training institution must have national accreditation, in agreement with UEMS standards, and should possess an adequate infrastructure and offer qualitative and quantitative clinical exposure.

Each participating institution in a network must be individually recognized as a provider of a defined section of the curriculum. Training centers must have a sufficient throughput of patients, an appropriate case-mix to meet training objectives, and be adequately resourced with teaching staff. The training must expose the trainee to a broad range of clinical experience.

The training of a trainee will be led and managed by a specialist. This specialist will be active in the practice, with personal responsibility for the management of patients with a wide range of genetic conditions. Within a training center there should be a team of specialists, each with subspecialty expertise and able to supervise and train a trainee. Allied specialties must be present to a sufficient extent to provide the trainee with the opportunity of developing his/her skills in a multidisciplinary approach to patient care. There is no specific trainee/trainer ratio required, but there should be a minimum of two teachers in a training center, and it is likely that non-medical healthcare professionals will also be engaged.

The trainee should be involved in the diagnosis and management process of new patients (outpatients and in-patients), as well as their follow up. A trainee must demonstrate increasing personal responsibility for the global care of patients with genetic conditions. There should be written general guidelines within the training institution concerning patient care and patient information (including informed consent), referrals, medical records, documentation, on-call and back-up schedules, attendance at conferences and educational/training courses.

The staff of a training center should engage collaboratively in regular reviews and audit of the center's clinical activity and performance. There should be regular multi-disciplinary meetings to determine optimal care for patients, involving both medical and other healthcare professionals.

There will be clinical engagement beyond the Center with other clinical groups such as Rehabilitation Medicine, Orthopedics, Pediatrics, Surgery, Obstetrics and Gynecology, Dermatology, Ophthalmology, etc.

Specialist staff appointed to a training center will have completed all training requirements themselves and will have been trained also in teaching and mentoring trainee staff, staff as well as in working in a multidisciplinary team with lab and genetic counsellors.

b. Requirement on equipment, accommodation

A training center should have sufficient equipment and support to enable the clinical practice that would be expected of a training center and thus provide the necessary educational opportunities for trainees.

The trainee must have adequate time and opportunities for practical and theoretical study and have access to adequate professional literature.

Computing and Information Technology and library resources must be available. All trainees must engage in clinical audit and have the opportunity to engage in research.

2. Quality Management within Training Institutions

Participation of the training institution in a certified quality management program with an external auditing process on a regular basis is consistent with good governance. Criteria of quality management at specialty training institutions include the following:

Accreditation

Training institutions need to be accredited with competent National Medical Boards.

Additional accreditation on a supra-national level, such as that provided by the European Society of Human Genetics (ESHG), is strongly recommended.

A training institution must have an internal system of medical audit or quality assurance. Quality assurance must be an integral part of the training program of all training institutions/networks. A national register of approved institutions/networks should be available.

Internal regulations: There should be written general guidelines within the training institution concerning patient care and patient information (including informed consent), referrals, medical records, documentation, leave (annual, study), maternity/paternity, residents 'working schedules, attendance to conferences and to educational activities. These should be available to staff and trainees.

Clinical governance

Employee-structure at training institutions needs to be designed in a way to accommodate for specialty training. Workload has to be managed with a priority on training.

Manpower planning

Training institutions should appoint a coordinator responsible for the composition, implementation and supervision of a specialty training program. Roles of trainer and trainee need to be clearly defined. Allotted time of at least one day per workweek should be implemented for specialty training interaction.

Manpower planning is under jurisdiction of each member state according to their needs for Clinical Genetics specialists.

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Regular report

Annual reports on various aspects of an institution's specialty training program should be made publically available.

External audit

Training institutions should appoint a coordinator who is also responsible for compliance of the training program with current guidelines, directives or regulations of competent medical boards, as well as the local medical school.

Transparency of training programs

Based on national and regional guidelines, UEMS strongly encourages training institutions to formulate defined training programs and make them publicly available (e.g. on their website). It would be expected that a training center would publish details of the training provision available with details of the clinical service it provides and the trainers. Such information would include the training programs, the nature of the clinical or laboratory experiences in which a trainee would be engaged, and the support and interaction with the trainer and Program Director. There would be a named individual whom a prospective trainee might contact and discuss the program.

Framework of approval

As part of training programs it should also be made clear how and by whom key achievements of training will be ascertained leading to a higher level of clinical responsibility and new assignments.

To assist a European medical specialist with additional Clinical Genetics competence moving from one EU country to another it would be expected that they have satisfactorily completed a training program. After the examination in Clinical Genetics they may be able to demonstrate that he/she has the required knowledge, clinical and laboratory skills and competences, as well as having demonstrated appropriate professional behaviors. Such accomplishments would be verified both by relevant documents and by the testimony of trainers and other staff who have worked with the trainee.

Feedback from trainers and trainees

Feedback about program quality from both trainers and trainees must be systematically sought, analyzed and acted upon. Trainers and trainees should be actively involved in using its results for program improvement and development.

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