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Description of "Rare and Undiagnosed Diseases" as a Medical Competency in EU: Aims and objectives in competency training

Competency Profile

Care of rare and undiagnosed disease patients is a multidisciplinary medical competency concerned with the provision of medical services to individuals, families and groups of affected individuals who have, or are at risk of having, conditions that are differentiated from common diseases primarily by their low incidence or prevalence. Such care includes diagnostic and counselling services that provide information about each condition and its implications, including management, prognosis, screening, prevention and reproductive options, as well as therapeutic possibilities. Information provided is based on clinical assessment, individual or family medical information, conventional laboratory investigations, imaging, and specialized genetic tests that can require complex interpretation. Besides conventional laboratory genetics (cytogenetics, molecular genetics, biochemical genetics), novel components of the services include specialized genetic and genomic approaches such as next generation sequencing and array technologies. The genetic studies include integrated clinical and laboratory services in rare disease management involving any disorder with a significant genetic component, whether inherited or sporadic.

Dedicated institutions already exist in several EU recognized medical specialties, with full medical career training systems. However, while there may be institutions or units that specialize in one or a few types of rare disease, institutions devoted exclusively to rare and undiagnosed diseases are scarce or lacking. As internal medicine, neurology, medical genetics and paediatric units often receive rare disease cases, these units could be re-organized into spokes of hub-and-spoke networks for rare diseases, which could improve care and provide career training specifically focused on rare diseases. Such full-range training is lacking at the moment. The existing educational pathways should be modified to provide education on rare diseases with a multidisciplinary perspective. They should be flexible enough to accommodate

educational needs that cover a spectrum of knowledge bases, including those that address all rare diseases and those that target only a limited number of rare conditions. These educational pathways could be provided in collaboration with the ERNs, and by the European university system. They should include courses on rare diseases, based on a broadly agreed-upon syllabus, and clinical fellowships on selected rare diseases. There should be an examination, with a certification of competence. All this could give rise to an integrated system that will produce experts on rare and undiagnosed diseases, who will ultimately become professionals with dedicated careers at the centres of hub-and-spoke networks. Due to the differences in European health care systems across member states, there may be different national emphases on the various elements of this training programme. We propose below a set of minimum criteria that will be recognized in programmes throughout the EU but

This document relates to training programs which is for qualified individuals intending to acquire UEMS CESMA certification and training in the competency of rare diseases. The individual obtains his/her certification (European Board Qualification – EBQ) through an Assessment run by the respective Board. Any such Assessment by the Board, itself needs a formal Visitation and Appraisal by UEMS-CESMA, every 5 years. A positive result from the Appraisal leads to Certification of the particular Competency Assessment. It recognizes that there may be areas of overlap with training programmes for other genetic professionals, especially in internal medicine, paediatrics, medical genetics, and neurology and that there may be opportunities for joint training for periods of the course.

One challenge for medical education in rare diseases, which differentiates it from common diseases (including genetic diseases), is that the rare disease discipline lacks reinforcement of recently acquired information due to the small patient numbers. When a physician attends an educational event on a common disease, there will likely be encounters with patients having that disease soon and often. The same does not apply when a disease is rare. The framework of any educational initiative in rare diseases must take into account this challenge.

Entry criteria

These may vary from country to country but would generally include a specified period of general medical training that includes an adult +/- paediatric +/- prenatal medicine "internship", prior to entering competency training in rare disease units. Some countries may have a minimum period of training to be undertaken before specialisation. Essentially, the duration of the training is 4 years, with a year of common trunk.

Educational goals

A concise description is provided below, with more details available in the Syllabus. This knowledge base lists the hallmarks of a trained medical doctor ready for qualification for the umbrella exam (certification) in rare diseases.

Knowledge and Skills

Fundamentals of rare disease genetics

- Cellular and molecular mechanisms that underpin human inheritance
- Chromosome structure and function, mitosis and meiosis, and the origin of aneuploidy and other imbalances
- The structure of DNA and RNA, replication, transcription and translation.
- Genetic epidemiology and biostatistics
- Risk assessment
- Population genetics, the principles of screening, and basic mathematical genetics
- Bioinformatics and basics of sequencing technology/testing
- Epigenetics
- Pharmacogenetics / pharmacogenomics
- Principles of acquired genetic disorders

Clinical understanding

- Common and unusual patterns of inheritance
- Taking a detailed medical and family history, pedigree construction
- Ability to perform genetic risk assessment, including the use of Bayes' Theorem to incorporate conditional risk information
- Ability to undertake risk assessment
- Diagnosis, investigation and management of individuals and their families with rare inherited/genetic diseases
- Therapeutic aspects and emerging innovative therapies in rare diseases
- Paediatric genetics including training in dysmorphology (knowledge of common dysmorphic syndromes, their aetiology and the use of dysmorphology databases)
- Adult rare disorders, including knowledge of late onset diseases and conditions with a significant genetic component presenting in adult life
- Prenatal rare diseases paradigms, fetal dysmorphology, and knowledge of the effects of common teratogens on fetal development
- Screening programmes
- Competent clinical examination of both paediatric and adult patients, especially in relation to dysmorphic signs and features, and neurological examination and interpretation
- Gene therapy, its current and future applications, and other strategies for the treatment of genetic disease.
- Common diseases with a rare component/variant

- Multifactorial/polygenic disorders in rare disease fields
- Sub-specialty areas, including:
  - Inherited metabolic disorders
  - Neurogenetic diseases
  - Neuromuscular rare diseases
  - Cardiovascular genetics
  - Reproductive genetics
  - Other subspecialties of specific interest to the trainee, e.g., connective tissue disorders, immunology, etc.

Technical skills:

Laboratory skills

- Thorough knowledge of principles of classic laboratory techniques used in genetic diagnostic testing
- Thorough knowledge of new laboratory techniques used in genetic diagnostic testing, including SNP and CGH arrays, whole genome sequencing and exome sequencing
- Understanding the interpretation of results from cytogenetic, molecular genetic, biochemical genetic and genomic analyses (array, exome and whole genome analyses)
- Knowledge about preanalytical handling of samples and logistics
- Awareness of quality issues in genetic testing
- Knowledge of international nomenclature systems used in genetic reporting
- The time spent and the practical expertise gained in laboratory work may vary among countries, but it should be sufficient to ensure highly specialized knowledge.

Biobanking

- Understand principles of biobanking
- Awareness of ELSI issues

IT

- Use of information technology including online resources and databases related to human genetics
- Rare diseases codification and ontologies
- Awareness and use of online data sharing resources

Non-technical skills:

Genetic counselling and communication skills

- Training in genetic counselling for all types of genetic diseases and genetics-related situations encountered in practice. This includes pre- and post-testing counselling in relation to reproductive options, including predictive genetic testing. Where applicable, training in co-counselling with other professionals, with specialists in other fields of medicine

- Understanding and handling of emotional reactions and personal and family crises in relation to the impact of genetic disease and the genetic diagnostic process
- Understanding ethical, legal and social issues, and the importance of consent and confidentiality
- Development of good communication skills with patients and families, colleagues in medical care centres and other specialists and healthcare professionals
- Understand ethical, legal and social issues in relation to genetic and genomic medicine
- Issues relating to patient confidentiality, consent and disclosure of results.

Management

- Knowledge of national laws relating to genetic services and practice, general healthcare policy, goals and priorities
- Understanding the organization and management of genetic services
- Opportunities to participate in departmental/service activities related to organizational planning, financial management, and monitoring and maintaining quality standards
- Development of multidisciplinary team operations and leadership skills

Teaching

- Develop teaching skills by participating in the education and training of various categories of staff
- Involvement with patient groups and patient/family education

Maintaining good medical practice

- Understand and practice medical professionalism, honesty, integrity, an aspiration to excellence, fairness, and avoidance of discrimination
- Develop a commitment to lifelong learning through continuing professional development and attend relevant courses and conferences.
- Participate in audit and clinical governance
- Adhere to accepted consent and confidentiality procedures
- Timely management of medical documentation and communication with patients, families, and professionals

Supplementary education and training

- Subcompetency training: Some trainees will elect to develop expertise in a subcompetency area such as cancer genetics, dysmorphology, neurogenetics, etc. This may also vary from country to country.
- Knowledge and understanding of the principles of evidence-based medicine
- Involvement and initiatives in courses, programmes and social issues related to rare diseases
- Knowledge of patient registries, patient support organisations

Technicals

- A written agreed curriculum for the training period should be set up as a contract between the trainee and the supervisor if not otherwise determined by national regulations, this must be within the frame of UEMS ETR scope of harmonization and standardization.
- Trainees should maintain a Training Logbook including details of clinical and laboratory experience, all educational activities, research, and publications
- A mechanism should be in place for continuous assessment of trainees against agreed quality standards; some countries will have a nationally prescribed system for assessment and certification
- Specialist examination may be compulsory in some countries

Research

- Medical genomics has a rapidly changing knowledge base and during competency training participation in research should be encouraged. Some trainees may wish to participate in scientific projects and research leading to a higher academic degree. On completion of training, some academic clinical/medical geneticists will continue to lead research programmes whilst many others will collaborate with laboratory-based colleagues within a broader team.
- Understand the principles of research methodology including clinical trials

Time frame for specialist training

- The training period should involve a minimum of 4 years full time work, with the option of one additional year spent in another competency before, after, or as a part of the specialist training. Part time work would extend the training period.
- In the longer training period (5 years), up to one year could be in another speciality relevant to rare diseases.
- The time spent in laboratory work may vary among countries according to national curricula.
- A period of research resulting in a PhD/other higher exam may, if appropriate, replace training for a variable period of time according to national guidelines. However, in absence of national guidelines, it is recommended that this time period should not be longer than 1/3 of the total training period.