



# UNION EUROPÉENNE DES MÉDECINS SPÉCIALISTES EUROPEAN UNION OF MEDICAL SPECIALISTS

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RUE DE L'INDUSTRIE, 24  
BE- 1040 BRUSSELS  
[www.uems.eu](http://www.uems.eu)

T +32 2 649 51 64  
F +32 2 640 37 30  
[info@uems.eu](mailto:info@uems.eu)

UEMS 2020/12

## Training Requirements for the Competency of Rare and Undiagnosed Diseases

### *European Standards of Postgraduate Medical 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 that 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 to provide 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 Chapter, the UEMS Specialist Sections and European Boards have continued working on developing these European Standards in Medical training that reflect 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

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doctors according to training requirements within all Member States; this is based on the length of training in the Competency and the title of qualification. Given the longstanding experience of UEMS Specialist Sections and European Boards on the one hand and the European legal framework enabling Medical Specialists and Trainees to move from one country to another on the other hand, the UEMS is uniquely positioned to provide competency-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”. 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 Chapter 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 Competency of Rare and Undiagnosed Diseases”. This document aims to provide the basic Training Requirements for each competency and should be regularly updated by UEMS Specialist Sections, Multidisciplinary Joint Committees, or European Boards to reflect scientific and medical progress. The three-part structure of this document 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 levels interested in knowing more about medical specialist training. It is important to note that at the moment there is no such medical speciality recognized in the Annexe V. Therefore we use the term of “competency” instead of medical speciality, even if, hopefully, it will become an independent medical professional soon.

A “Rare Disorder” (rare disease, orphan disease) is defined according to the European standards as one having a prevalence of not more than five affected persons per 10.000. In different parts of the world, where consanguinity is accepted, different rare diseases can become frequent. Rare diseases can be of genetic origin, multifactorially determined or caused by environmental factors. Rare diseases can be part of different specialities ranging from genetics, through infections to different type of cancers. To date, 5.000-8.000 rare diseases are known, affecting 6-8% of the world population; 80% are of genetic origin while 20% are multifactorial. More than 50% affect children and 30% of them die before the age of 5. The number of rare diseases is increasing partly because of the intense development of genetic testing modalities and the new therapeutic modalities achievable. Rare diseases affect around 30 million EU citizens; they are recognised as a global public health priority and an exemplar domain for precision public health. A special challenge is the undiagnosed group of rare diseases.

With the formation of the 24 thematic European Reference Networks (ERN) in March 2017, the need for Rare Disease Specialists became evident. ERN’s are virtual networks involving healthcare providers across Europe. The aim of ERN’s is to harmonize diagnostic strategies and perhaps in the future therapeutic approaches regarding rare diseases across the European Union. The need for rare disease specialists involves all speciality groups and Rare Disease Centers from all countries should have a minimum of 10 rare disease specialists. Whereas not all rare diseases are genetic conditions, the genetic departments are expected to feature prominently.

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## **Aims and goals of Rare and Undiagnosed Disease Speciality training and assesment (Further essential parts of the ETR are described in the supplementary “Description of the comptenecy” and “Syllabus”):**

1. To construct a tool for training and qualification/certification system for service specialists whose goal is to assess, investigate, and diagnose diseases and medical conditions that are rare, having a prevalence of less than 1 in 2.000.
2. To create a system that provides specialist knowledge-based training information about rare and undiagnosed diseases, including recommendations for screening where appropriate.
3. To provide a service that offers education/knowledge base of counselling in relation to reproductive options and prenatal genetics in rare diseases.
4. The primary prevention of rare diseases of multifactorial or nongenetic origin, based on the knowledge and mitigation of risk factors related to medical treatments, maternal health, infections, diet, lifestyles, living environment and workplace
5. The primary and secondary prevention of rare hereditary diseases, according to: i) the choice made by those at risk of having affected offspring, based on full information and expert counselling; ii) effective programmes of newborn screening allowing early measures to prevent the onset of diseases
6. To contribute to the management of patients and families affected by rare diseases, in collaboration with other medical specialists, including treatment.
7. Knowledge on European Reference Networks (ERN), CPMS system essentials.
8. To be advocates, where necessary, for those affected by rare diseases. EURORDIS knowledge base.
9. To conduct and contribute to clinical and genomic research to enhance knowledge of the causation and natural history of rare diseases and conditions.
10. To teach and instruct medical undergraduates and postgraduates in rare and undiagnosed diseases, in order to raise the knowledge base across all medical specialties.
11. To provide a knowledge and skills resource to all medical specialties, including through multidisciplinary meetings.
12. Orphanet and orphacode knowledge base.
13. To provide a service in collaboration with clinical specialists, researchers and geneticists for diagnosis and description of a novel disorder.
14. The European Certificate in Rare and Undiagnosed Diseases is intended to be the main knowledge-based assessment tool for training and assessment across Europe, with the ultimate aim of establishing world class-leading standards in that competency throughout all countries.
15. The rare cancers are recognised under different specialties (Rare adult solid cancers, pediatric oncology, etc), some basics are still included in this competency field as well.
16. Special attention and care should be taken for expertise in the undiagnosed fields and disease management. Management of undiagnosed cases, recontacting, data sharing, participation in the existing and emerging networks.
17. Basics of communcable diseases, and selected parts of the rare diseases (including tropical), basics in bacteriology, virology, parasitology. Migration and its communicable diseases consequences.
18. Toxicology, applied pharmacotoxicology, teratology basics.
19. To facilitate connections between individuals affected with the same rare disorder or those with as yet undiagnosed diseases.
20. To contribute to the public awareness for rare diseases.

## **I. TRAINING REQUIREMENTS FOR TRAINEES**

## **1. Content of training and learning outcome**

The Rare and Undiagnosed Disease Competency is a field of medicine concerned with the investigation, diagnosis, treatment, prevention, and research into rare and undiagnosed diseases. The scope of patient care activities includes the recognition of these diseases, the early identification of individuals and families at risk, the identification of the possible underlying (genetic) defect and the preventive care of affected family members and identification of environmental, infectious, toxicological and/or diet/lifestyle-related risk factors to prevent diseases in population.

This competency training is aimed at giving doctors qualifications in the field of “Rare and Undiagnosed Diseases” to enable them to manage the treatment of patients with rare diseases and their families in 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. Rare disease specialists must also be able to coordinate the follow-up of patients affected by rare diseases.

### **Elements of knowledge base (see details in Description of Competency file and in Syllabus)**

- *Basic theoretical genetics / Basic science*
- *Clinical/Medical knowledge and specialist-level skills*
- *Genetic counselling and communication skills*
- *Laboratory skills*
- *Maintaining Good Medical Practice*
- *IT skills*
- *Ethics and law*
- *Biobanking and registries*
- *Management training*
- *Teaching*
- *Quality assurance*
- *Research*

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## Competencies required to gain by the trainee

- sufficient knowledge and experience to manage a complex rare disease.
- shared knowledge of experts by having several specialists under one roof.
- able to undertake research in the particular rare disease and improve not only the care of one individual, but of other patients with the same condition.
- holistic vision of the patients.
- good communication skills.
- developing patient registries.
- coordinating patient routes and follow-up.

Knowledge, Technical and Non-technical skills are discussed under different headings in a formalized and more elaborate Syllabus and again in the Curriculum.

## 2. Organization of training

### a. Schedule of training

The optimal Rare Disease Competency (speciality) training is 5 years consisting of 1 year of common trunk and 3 years training in a Rare Disease Center in an accredited program and/or center and an optional extra year spent in another competency before, after, or as a part of the specialist training. The training of 3 years is also accepted, if the candidate has additional year(s) in research related to rare diseases. However, those countries that have a 3 year-course must arrange a preliminary general training, covering medicine and pediatrics if possible, before, and separate from, the 4 year specialist training. The specialist training is defined here as training in institutions involved in rare disease care. This includes training in the units with profiles of following medical specialties with rare disease outpatient clinic and/or ward: Allergology, Anaesthesiology, Cardiology, Cardiothoracic Surgery, Dermatology and Venereology, Emergency Medicine, Endocrinology, Gastroenterology, Geriatrics, Gynaecology and Obstetrics, Infectious Diseases, Internal Medicine, Laboratory Medicine / Medical Biopathology, Medical Genetics, Medical Microbiology, Medical Oncology, Nephrology, Neurology, Neurosurgery, Occupational Medicine, Ophthalmology, Oro-Maxillo-Facial Surgery, Orthopaedics, Traumatology, Otorhinolaryngology, Paediatrics, Pneumology, Psychiatry, Public Health Medicine, Radiology, Radiation Oncology and Radiotherapy, Rheumatology, Surgery, Urology. 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 as a sub-speciality qualification of 2 years built on Clinical/Medical/Human Genetics speciality. 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), internal medicine, emergency unit, and genetics.
- 3 years different competency oriented practice including the previously mentioned specialities.
- An option of one additional year spent in another competency before, after, or as a part of the specialist training. Since this is an option (not compulsory), it is technically not a requirement. Perhaps the 5<sup>th</sup> year could be rendered as an integral and required part of the Training programme. This is optional for those who have some other type of medical board qualification.

### **b. Curriculum of training**

The general aim of the training program is to enable the Rare Disease Specialist to work effectively as a consultant. The trainee must demonstrate the ability to record and convey patient details of history, examination and investigation 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. During the course of the training program classical methodologies will be applied. These include but are not limited to: lectures, seminars, bed side teaching, case reports, case scenario discussions, journal clubs, e-learning, webinars, computer assisted, self-instruction modules, problem-based learning, team-based learning, simulation etc.

### **c. Assessment and evaluation**

The European Certificate in Rare and Undiagnosed Diseases (ECRUD) is intended to be the main knowledge-based assessment tool for training and assessment across Europe and ultimately for the entire continent's experts, with the aim of establishing world class-leading standards in that competency throughout all countries.

Countries will use assessment strategies appropriate to their needs, provided that they introduce their own training and assessment systems. 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 rare and undiagnosed diseases competence'. Thus, there will need to be an assessment of knowledge, through a form of written examination. 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. Oral exam can be part of the process as well. At the beginning it will be likely 100-150 MCQ but later on with the strict control of the CESMA further testing options will be considered.

Assessments will be formalized and will become obligatory over time. They will consist of Formative Assessments and a Summative Assessment, specifically, a competence-based logbook and an Exit Examination, respectively. For Formative Assessments an option would be formal documentation of trainee's development and progress after review of evidence collected. Summative assessments takes place after a specified training period with the purpose of deciding whether the trainee has reached a standard to proceed to the next level of training or to be awarded a certificate of Completion of Training.

Assessment of progress of education and training must include continuous assessment which tests whether the trainee has acquired the appropriate knowledge, skills, attitudes and professional qualities. This must include formal annual evaluations and final evaluations. The annual evaluation must formalize the assessment of a trainee's competence to promote the trainee's improvement. Final completion of a training program should be dependent upon review of the trainee's portfolio as well as success in the final examination. The Training program director must provide an overall judgment

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about the trainee's competence and fitness to practice as an independent specialist in Rare and Undiagnosed Diseases. We propose the following Assessment Protocol:

## First Part (Theoretical)

- 100-150 Questions Multiple Choice
- 10 Gross description & Reducing Tech and modalities

## Second Part (Practical) based on virtual Pathology report

- 5 whole exome and 5 whole genome report
- 5 array interpretation
- 6 imaging (3 CT, 3 MRI result interpretation)

Pass rate will employ the Angoff method, which calculates a cut-off mark based on the performance of candidates in relation to a defined standard (absolute) as opposed to how they perform in relation to their peers (relative). It involves a judgement being made on exam items (test-centered) as opposed to exam candidates (examinee-centered).

## d. Grades of Competence:

1. Knowledge
  - 1.1. knows of
  - 1.2. knows basic concepts
  - 1.3. knows generally
  - 1.4. knows specifically and broadly
2. Clinical Skills
  - 2.1. Has observed – the trainee acts as an 'Assistant'. From complete novice through to being a competent assistant. At end of level 1 the trainee:
    - 2.2. Has adequate knowledge of the steps through direct observation.
    - 2.3. Demonstrates that he/she can handle the apparatus relevant to the procedure appropriately and safely.
    - 2.4. Can perform some parts of the procedure with reasonable fluency
    - 2.5. Can do with assistance - a trainee is able to carry out the procedure 'Directly Supervised'. From being able to carry out parts of the procedure under direct supervision, through to being able to complete the whole procedure under lesser degrees of direct supervision (e.g. trainer immediately available). At the end of level 2 the trainee
    - 2.6. Knows all the steps - and the reasons that lie behind the methodology.
    - 2.7. Can carry out a straightforward procedure fluently from start to finish
    - 2.8. Knows and demonstrates when to call for assistance/advice from the supervisor (knows personal limitations).
    - 2.9. Can do the whole procedure but may need assistance – a trainee is able to do the procedure 'indirectly supervised'. From being able to carry out the whole procedure under direct supervision (trainer immediately available) through to being able to carry out the whole procedure without direct supervision i.e. trainer available but not in direct contact with the trainee. At the end of level 3 the trainee
  - 2.10. Can adapt to well-known variations in the procedure encountered, without direct input from



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the trainer.

- 2.11. Recognizes and makes a correct assessment of common problems that are encountered.
  - 2.12. Is able to deal with most of the common problems.
  - 2.13. Knows and demonstrates when he/she needs help.
  - 2.14. Requires advice rather than help that requires the trainer to intervene
  - 2.15. Competent to do without assistance, including complications. The trainee can deal with the majority of procedures, problems and complications, but may need occasional help or advice.
  - 2.16. Can be **trusted** to carry out the procedure, independently, without assistance or need for advice. This concept would constitute one Entrustable Professional Activity (EPA). An EPA is ‘a critical part of professional work that can be identified as a unit to be entrusted to a trainee once sufficient competence has been reached’. This would indicate whether one could *trust* the individual to perform the job and not whether he is just competent to do it. At the end of level 5 the trainee:
    - 2.17. Can deal with straightforward and difficult cases to a satisfactory level and without the requirement for external input to the level at which one would expect a consultant to function.
    - 2.18. Is capable of instructing and supervising trainees.
3. Technical Skills
    - 3.1. Has observed.
    - 3.2. Can do with assistance.
    - 3.3. Can do whole but may need assistance.
    - 3.4. Competent to do without assistance, including complications, but may need advice or help.
    - 3.5. **Can be trusted to carry out the procedure, independently, without assistance or need for advice (EPA).** EPAs have been explained previously.

The above detailed classification of Competence Levels could be useful during the process of formative training, when it comes to deciding when an applicant is eligible to sit an eventual Specialist Exit examination, it is the evaluation of the EPAs which is essential. In this sense, the Eligibility Assessment Process is really the first part of the Examination and that explains the suggestion that the ‘5<sup>th</sup> level of Technical Skills competence’ should be included in a standardized Logbook Template for all trainees

## e. List<sup>1</sup> of comprehensive Entrustable Professional Activities (EPAs)<sup>2</sup>

- Evaluate and manage a new medical condition in an ambulatory patient and coordinate care between healthcare providers across multiple care settings
- Manage the care of patients with rare cancers across multiple care settings
- Manage the care of patients with complex medical conditions, and/or comorbidities, across multiple

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<sup>1</sup> Adopted with revisions from Karen. E. Hauer, Jeffrey Kohlwes, Patricia Cornett, Harry Hollander, Olle ten Cate, Sumant R. Ranji, Krishan Soni, William Iobst, and Patricia O’Sullivan (2013) Identifying Entrustable Professional Activities in Internal Medicine Training. Journal of Graduate Medical Education: March 2013, Vol. 5, No. 1, pp. 54-59 and the Alliance for Academic Internal Medicine. Internal Medicine End of training EPAs, 2012

<sup>2</sup> Definition: An EPA is ‘a critical part of professional work that can be identified as a **unit** to be **entrusted** to a trainee once sufficient competence has been reached’. An EPA goes a level higher than the traditional 4+ level of competence which is the ‘independence competency’. The key factor is **Entrustment**. The trainee is not only capable of tackling the particular procedures or units independently, but he can be **trusted** to do this by his tutors.



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care settings

- Manage transition of care for adult patients transferring to another care setting
- Manage transition of care for young patients transferring from pediatric to adult services
- Provide medical consultation to nonmedical specialties
- Lead a family meeting to discuss serious news (bad news, end of life care) with a patient and/or family and other health providers
- Obtain initial history, perform physical examination, and formulate a management plan for a new ambulatory patient in continuing care
- Manage the care of patients with chronic conditions across multiple care settings
- Access medical information to provide evidence-based care
- Facilitate the understanding of patients, their families, and members of the multidisciplinary team
- Recognize and diagnose common nonmedical conditions (i.e., surgical, neurological, dermatologic, psychiatric etc.) and refer appropriately to other specialty care
- Diagnose and manage patients with complex conditions needing other specialty care (inpatient or outpatient)
- Organize and maintain information and knowledge through medical practice to improve personal development when delivering care and educating others (journal club, etc.)
- Recognize when palliative care is needed and liaise with palliative care specialists
- Counsel patients appropriately
- Advocate for individual patients by representing them, supporting them and working for them
- Improve patient safety
- Provide age appropriate screening and preventative care
- Identify and address any need for quality improvement in a clinical setting
- Improve the quality and safety of healthcare at both individual and systems levels
- Provide telephone management for an ambulatory rare disease patient
- Provide care to nonnative speakers in an inpatient or outpatient setting through the use of appropriate translation services
- Develop and implement a management plan based on review of outcome data for ambulatory patient population
- Provide inpatient and outpatient care for patients with difficulty in accessing appropriate healthcare; advocate for individual patients where needed
- Participate in an in-hospital cardiopulmonary resuscitation
- Perform common procedures in internal medicine (lumbar puncture, thoracocentesis, central line insertion, joint aspiration)
- Undertake a research project (e.g., a degree or diploma, quality improvement, educational opportunity, other)
- Develop the practice of lifelong learning
- Demonstrate professional behavior at all time

## **f. Logbook Recommendation:**

**Purpose:** The purpose of the logbook is to document that the applicant has had direct and meaningful involvement in the rare disease evaluation, counseling and management of patients and/or families, and has received appropriate clinical supervision.

The EPA is a Unit and units can be counted. The certified Logbook with a category for EPA included is the key. Because the emphasis and attitudes regarding the spectrum of competences and education within any Medical Specialty vary significantly in the individual states, one cannot expect applicants to have attained EPA competency in each and every item listed in the Syllabus/Curriculum. In other words, one cannot expect Eligible Candidates to have attained must have attained 100% of the possible EPA Units in the Syllabus / Curriculum. The Eligibility Committee applies the correct degree of flexibility allowing for equivalence of some procedures. To give an example, the percentage of items in the

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Syllabus to be expected of an applicant attaining the EPA grade of competence, for the EBSQ General Surgery, is presently set at 65%. This is an arbitrary figure which was reached by evaluating the previous year's candidates' data but will obviously vary with each particular Assessment and possibly from year to year. Another important legal point is that each Examination Board has to establish this threshold when the Exam Webpage goes online.

Requirements: Logbook of the 55 cases must be completed in accordance with the instructions provided in this summary, and anticipates ongoing review of cases between the trainee and their program director, the applicant should assure that all requirements have been fulfilled before submitting the final logbook for review.

## Case Selection:

All cases must be obtained through accredited residency and/or training program.

Supervision for case encounters in genetics clinics must be provided by faculty who are certified

All 55 cases must be obtained during the inclusive dates of the applicant's training. No more than 2 cases may be obtained in any one day.

Each logbook entry must document a face-to-face interaction between the applicant and an individual patient and/or family.

A given patient or family may appear only once in an applicant's logbook, regardless of the number of encounters with that patient or family.

## Description of Logbook Headings/Columns:

Entry Number: The logbook spreadsheet allows a trainee to enter an unlimited number of cases while in training. For the final logbook that may be requested for audit, you must select 100 cases to submit that fulfill all of the defined requirements. The applicant must be able to identify each case by its entry number if questions arise about a logbook entry

Date: The date in month/day/year [MM/DD/YYYY] format identifies when the patient was seen

Patient Age Category: For each case, the patient's age must be defined as Infant (5 cases), Child and Adolescent (20 cases), or Adult (25 cases) or Undiagnosed of any age (5 cases). Age refers to age of the patient on the date of the clinic visit.

## Diagnosis:

No more than 5 cases may have the same specific diagnosis. Variations in genotype or phenotype of a specific diagnosis, such as age of onset or particular mutation, are not considered sufficient to count as separate diagnoses. It is the age at onset and not the age of diagnosis or the age at which the trainee saw the patient that should be taken into account in satisfying this requirement.

For each case, enter the diagnosis using the guidelines below:

Enter the diagnosis using the OMIM name or an ORPHACODE alternative title. All cases representing the same condition should be entered using the same diagnosis name.

Do not use abbreviations unless an OMIM/ORPHACODE alternative title.

Primary diagnosis must be listed first.

Use the most specific diagnosis for each case when known.

Log only those cases for which the diagnostic evaluation is complete. For example, "5p deletion syndrome" not "Rule out chromosome anomaly." If making a specific diagnosis was the reason for the referral, for example, is this Marfan syndrome?, use "Marfan syndrome" if the diagnostic evaluation is complete and this is the diagnosis or "Marfan syndrome, excluded" if the diagnostic evaluation is

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complete and this diagnosis was excluded but a more specific diagnosis could not be made. If a more specific diagnosis could be made, such as Shprintzen-Goldberg syndrome, use the more specific diagnosis.

If more than one patient or family with the same genetic category, age category, diagnosis, visit date, trainee role(s), and supervisor are recorded, clearly indicate that entries are not duplicated records or members of the same family, as follows: Neurofibromatosis, patient or family 1; Neurofibromatosis, patient or family 2.

Continuous medical education (CME) and continuous professional development (CPD) to keep updated with developments in diagnosis and management of rare conditions as well as of global professional skills is an obligation of the accredited expert. Type, duration, content and monitoring of CME/CPD activity need to be established and will fall under the authority of boards that should consider the general recommendations of the UEMS. The UEMS provides European Accreditation of CME (EACCME) for international events according to defined quality standards. It is recommended that trainees in the rare disease field are introduced to CME/CPD during their postgraduate training period.

The ECRUD examination will be a joint development of the UEMS Multidisciplinary Joint Committee (UEMS-MJC RUD) and the sections, MJC-s, National Medical Associations, and of the European scientific societies, world networks, like the Undiagnosed Disease Network International (UDNI) intended to join in this effort. The examination is overseen and supervised by the Examination Steering Committee. It will be open to candidates who are trainees or fully trained experts from any nation. The ECRUD will definitely be an excellence exam, and will be valid for practice only in countries where it is ratified as an official certificate for this purpose by national regulatory bodies or organisations.

The combination of the formative and the summative assessment modalities will be used for assessing the status of the competences acquired. Formative in-training assessment will be incorporated throughout the training period and should include evaluation tools based on mini-clinical evaluation exercise, direct observation of clinical encounters, skills and procedures, classic SWOT analysis (Strengths, Weaknesses, Opportunities, Threats) of procedures performed. Knowledge should be assessed with multiple choice questions (MCQ) during the training period. The final summative assessment is performed at predefined time points of the training period. – Like early during training e.g. after the first year, compulsory appraisal of the trainee is recommended in order to identify residents unfit for training, the CESMA will provide their help. After 3 years of training, "part I" of the exam for the European Certification in Rare and Undiagnosed Diseases (ECRUD) may be completed. - Towards the end of the training, national diploma or part II of ECRUD may be completed. Those colleagues, who do have their training completed, can proceed with the ECRUD directly. The details will be determined by the Examination Steering Committee, under strong supervision of the CESMA. The UEMS CESMA endorses the ECRUD exams as a label of excellence for specialist practice, however, not as a working licence. ECRUD examination covers relevant basic sciences and clinical topics appropriate for a specialist. An increasing number of European countries will hopefully officially adopt the ECRUD as their national examination. The existence of a supra-national examination in the rare and undiagnosed diseases provides an incentive for the development and improvement of departmental, university, national and European training programs. ECRUD examination achieves a uniformly high standard of knowledge throughout Europe as judged by an independent Board of Examiners. The UEMS Council of European Specialist Medical Assessments (CESMA) have defined

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recommendations on the development and organisation of assessment, selection and training of assessors.

Due to the current covid-19 virus outbreak pandemic, our lives and activities have been changed and are changing right now. We are facing a new situation on a global scale and we are reorganizing our work, many events have been canceled, postponed or re-organized in online format in order to keep everybody safe and collaborate all together to overcome this crisis. This also affects examinations worldwide. Taking this into consideration it is important for the future that we develop a system for doing the exam online and possible parts of the training as well, all according to UEMS CESMA guidelines.

Detailed description of: examination structure, manpower involved in construction and delivery, logistics, information provided to candidates and to examiners, conflicts of interest, procedure available for candidates to appeal, demographics of examiners and of candidates, language(s) in which the examination is conducted, proof of compliance with European legislation (e.g. GDPR).

## **g. Governance**

Governance of each 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. Naturally everything will be conducted under supervision of CESMA, EACCME and NASCE.

## **II. TRAINING REQUIREMENTS FOR TRAINERS**

### **1. Process for recognition as trainer**

#### **a. Requested qualification and experience**

Trainers/examiners should be certified rare disease specialists and must be recognized by a European or 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. They organize the activities of the educational program in all institutions that participate in the program.

#### **b. Core competencies for trainers**

1. Familiar with major and influential aspects of rare diseases.
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 be able to demonstrate appropriate participation in continuing professional development.

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5. Able to recognize trainers whose professional behavior is unsatisfactory and initiate corrective and supportive measures as needed.

## **2. Quality management for trainers**

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 determine.

## **III. TRAINING REQUIREMENTS FOR TRAINING INSTITUTIONS**

### **1. Process for recognition as training center**

#### **a. Requirements for staff and clinical activities**

A training center is a place, or number of places, where trainees are able to develop/acquire their competencies in rare diseases. 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 rare diseases. 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 to develop 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 (out-patients and in-patients), as well as their follow up. A trainee must demonstrate increasing personal responsibility for the global care of patients with rare disorders. 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

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Medicine, Orthopedics, Pediatrics, Surgery, Obstetrics and Gynecology, Dermatology, Ophthalmology, Psychiatry 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, and working in a multidisciplinary team with lab and genetic counsellors.

## **b. Requirements for 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 audits 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. Naturally everything will be conducted in accordance to CESMA, EACCME and NASCE guidelines. Criteria of quality management at competency 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 an European body, 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 registry 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 at conferences and 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 competency training. Workload has to be managed with a priority on training. The governance of the training program is primarily the responsibility of the Program Director and the institution(s) in which the training program tasks are being delivered. A trainer will be responsible to the Program Director for delivering the required training in their area of practice and competency. Training requirements for trainers, and a Process for recognition as a trainer are expected. Trainers are expected to have achieved the appropriate nationally recognized and certified qualification to allow them to practice as a specialist/consultant.

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## **Manpower planning**

Training institutions should appoint a coordinator responsible for the composition, implementation and supervision of a competency 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 competency training interaction.

Manpower planning is under the jurisdiction of each member state according to their needs for rare disease specialists.

## **Regular report**

Annual reports on various aspects of an institution's competency training program should be made publicly 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 is 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 to discuss the program.

## **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.