European Union Committee of Experts on Rare Diseases

EUCERD RECOMMENDATIONS

QUALITY CRITERIA FOR CENTRES OF EXPERTISE FOR RARE DISEASES IN MEMBER STATES

24 OCTOBER 2011

INTRODUCTION

1. THE EUROPEAN CONTEXT

Centres of expertise (CE) and European Reference Networks (ERN) in the field of rare diseases (RD) are mentioned in the HLG Report of November 2005, the Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of Regions on Rare Diseases: Europe's challenges (11.11.08) and the Council Recommendation on an action in the field of rare diseases (08.06.09), as well as in the Europlan Recommendations for the Development of National Plans and Strategies for Rare Diseases, and in Articles 12 and 13 of the Cross-Border Healthcare Directive (09.03.2011).

An analysis of the current situation in Europe regarding CEs and ERNs was developed in the framework of the European Commission's Rare Diseases Task Force (RDTF) from 2005 to 2009 and the European Union Committee of Experts on Rare Diseases (EUCERD) from 2010 to 2011. Several reports were published as a result of workshops where stakeholders had the opportunity to express their views.

The current recommendations are directly derived from the following documents:

- **Directive (EC 2011/24/EU) of the European Parliament and of the Council on the application of patients' rights in cross-border health care**
  http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2011:088:0045:0065:EN:PDF

- **Commission Communication, Rare Diseases Europe's Challenge**
  http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf

- **Council Recommendation (2009/C 151/02) of 8 June on an action in the field of rare diseases**
  http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF

- **Work of the High Level Group on Health Services and Medical Care during 2005**
  http://ec.europa.eu/health/archive/ph_overview/co_operation/mobility/docs/highlevel_2005_013_en.pdf

- **RDTF Report: Overview of Current Centres of Reference on rare diseases in the EU (September 2005)**
  http://www.eucerd.eu/upload/file/Publication/RDTFECR2005.pdf

- **RDTF Report: Centres of Reference for Rare Diseases in Europe – State-of-the-art in 2006 and Recommendations of the Rare Diseases Task Force (September 2006)**
  http://www.eucerd.eu/upload/file/Publication/RDTFECR2006.pdf

- **RDTF Report: European Reference Networks in the field of Rare Diseases: State of the art and Future Directions (July 2008)**
  http://www.eucerd.eu/upload/file/Publication/RDTFERN2008.pdf

- **EUCERD Workshop Report: Centres of expertise and European Reference Networks for Rare Diseases (8-9/12/2010)**
  http://www.eucerd.eu/upload/file/WorkshopReport/EUCERDWorkshopReportCECERN.pdf

- **EUCERD Workshop Report: National centres of expertise for rare diseases and networking between centres of expertise for rare diseases (21-22/03/2011)**
  http://www.eucerd.eu/upload/file/EUCERDReport220311.pdf

- **EUCERD Report: Preliminary analysis of the experiences and outcomes of ERNs for rare diseases (May 2011)**
  http://www.eucerd.eu/upload/file/Reports/ERN2011Analysis.pdf

- **EUROPLAN: Recommendations for the Development of National Plans and Strategies for Rare Diseases**
  http://www.europlanproject.eu/public/contenuti/files/Guidance_Doc_EUROPLAN_20100601_final.pdf

The specific excerpts of the above texts which have been carefully considered are given in Annex 1.

2. METHODOLOGY FOR THE ELABORATION BY THE EUCERD OF THE EUCERD RECOMMENDATIONS ON QUALITY CRITERIA FOR CENTRES OF EXPERTISE FOR RARE DISEASES IN MEMBER STATES

At the EUCERD workshop on National Centres of Expertise (CE) and European Reference Networks (ERN) for rare diseases (RD), held on 21-22 March 2011, it was proposed that the discussions would be used to fine tune the recommendations and concepts defined by the High Level Group on Health Care and Medical Services (HLG) and by the Rare Disease Task Force (RDTF).

Draft recommendations were elaborated by the EUCERD Scientific Secretariat from the documents published previously on national centres of expertise for rare diseases and ERNs which are cited in section 1. The draft recommendations were sent to all EUCERD members for their comments. Following this, a workshop was organised on 8 September 2011 with members of the EUCERD, including Member State (MS) representatives, to discuss the wording of the recommendations. The revised draft recommendations were then submitted to all EUCERD members ahead of the 3rd EUCERD meeting on 24-25 October 2011, at which these recommendations were adopted by the Committee.

These recommendations are intended to help MS in their reflections or policy developments concerning national plans and strategies for rare diseases when addressing the issue of organisation of healthcare pathways at national and European level. They may be of help to the Committee on Cross-Border Healthcare in the context of their reflection on European Reference Networks.

Please note that the order of the recommendations in this document is not a prioritisation.

RECOMMENDATIONS

Mission and scope of centres of expertise (CEs) for rare diseases (RD) in Member States (MS)

1. CEs tackle diseases or conditions requiring specific care due to the difficulty in establishing a diagnosis, to prevent complications and/or to set up treatments.
2. CEs are expert structures for the management and care of RD patients in a defined catchment area, preferably national, and at international level if necessary.
3. The combined scope of all CEs within a MS covers all RD patients' needs, even if they cannot provide a full range of services with the same level of expertise for each RD.
4. CEs bring together, or coordinate, within the specialised healthcare sector multidisciplinary competences/skills, including paramedical skills and social services, in order to serve the specific medical, rehabilitation and palliative needs of rare diseases patients.
5. CEs contribute to building healthcare pathways from primary care.
6. CEs have links with specialised laboratories and other facilities.
7. CEs collaborate with patient organisations to bring in the patients' perspective.
8. CEs contribute to the elaboration of good practice guidelines and to their dissemination.
9. CEs provide education and training to healthcare professionals from all disciplines, including paramedical specialists and non-healthcare professionals (such as school teachers, personal/homecare facilitators) whenever possible.
10. CEs contribute to and provide accessible information adapted to the specific needs of patients and their families, of health and social professionals, in collaboration with patient organisations and with Orphanet.
11. CEs respond to the needs of patients from different cultures and ethnic groups (i.e. have cultural sensitivity).

12. According to national/international ethical and legal frameworks, CEs should ensure respect of non-discrimination and non-stigmatisation of RD patients across Europe, within their sphere of competencies.
13. CEs contribute to research, to improve the understanding of the disease and to optimise diagnosis, care and treatment, including the clinical evaluation of long-term effects of new treatments.
14. The scope of diseases covered by each CE, or by a CE at national level, will vary depending on the size of the country and the structure of the national health care system.
15. CEs liaise with other CEs at National and European level when relevant.
16. A national directory of formally designated CEs is compiled and made publicly available, including on the Orphanet portal.

Criteria for designation of CEs for RD in MS

17. Capacity to produce and adhere to good practice guidelines for diagnosis and care.
18. Quality management in place to assure quality of care, including National and European legal provisions, and participation in internal and external quality schemes when applicable.
19. Capacity to propose quality of care indicators in their area and implement outcome measures including patient satisfaction.
20. High level of expertise and experience documented, for instance, by the annual volume of referrals and second opinions, and through peer-reviewed publications, grants, positions, teaching and training activities.
21. Appropriate capacity to manage RD patients and provide expert advice.
22. Contribution to state-of-the-art research.
23. Capacity to participate in data collection for clinical research and public health purposes.
24. Capacity to participate in clinical trials, if applicable.
25. Demonstration of a multi-disciplinary approach, when appropriate, integrating medical, paramedical, psychological and social needs (e.g. RD board).

26. Organisation of collaborations to assure the continuity of care between childhood, adolescence and adulthood, if relevant.
27. Organisation of collaborations to assure the continuity of care between all stages of the disease.
28. Links and collaboration with other CE at national, European and international level.
29. Links and collaboration with patient organisations where they exist.
30. Appropriate arrangements for referrals within individual Member States and from/to other EU countries if applicable.
31. Appropriate arrangements to improve the delivery of care and especially to shorten the time taken to reach a diagnosis.
32. Consideration of E-Health solutions (e.g. shared case management systems, expert systems for tele-expertise and shared repository of cases).

Process for designating and evaluating CEs for RD in MS

33. MS take action concerning the establishment and designation and evaluation of CEs and facilitate access to these centres.
34. MS establish a procedure to define and approve designation criteria and a transparent designation and evaluation process.
35. The designation criteria defined by MS are adapted to the characteristics of the disease or group of diseases covered by the CE.
36. CEs may not fulfill some of the designation criteria defined by the MS as long as the absence of fulfillment of those criteria does not impact on the quality of care and as long as CEs have a strategy in place to attain designation criteria in a defined time period.
37. The designation process at MS level ensures that the designated CEs have the capacity, and the resources to fulfill the obligations of designation.
38. The designation of a CE is valid for a defined period of time.
39. CE are re-evaluated on a regular basis through a process incorporated into the designation process at MS level.
40. The designating authority at MS level may decide to withdraw the designation of a centre of expertise if one or more of the conditions that formed the basis for

designation is no longer satisfied, or if there is no longer a need to maintain the national service.

The European dimension of CEs

41. MS with established CEs share their experience and quality indicators with other MS and coordinate their efforts to identify CEs for all RD patients at EU level.
42. Networking of CEs is a key element of their contribution to patient diagnosis and care, to ensure that expertise travels rather than patients themselves when appropriate; exchange of data, biological samples, radiological images, other diagnostic materials, and e-tools for tele-expertise are promoted.
43. Cross-border healthcare is organised, where appropriate, with designated CEs in neighbouring or other countries, where patients or biological samples can be referred to.
44. Member States should provide adequate information to professionals, citizens and patients organisations concerning the possibilities and conditions of access to health care at national and international levels in the field of rare diseases.
45. Designated CEs at MS level are the key elements of the future ERNs.

ANNEX I : EXTRACTS OF RELEVANT EUROPEAN TEXTS

Directive (EC 2011/24/EU) of the European Parliament and of the Council on the application of patients' rights in cross-border health care

(…)

(54) The Commission should support the continued development of European reference networks between healthcare providers and centres of expertise in the Member States. European reference networks can improve the access to diagnosis and the provision of high-quality healthcare to all patients who have conditions requiring a particular concentration of resources or expertise, and could also be focal points for medical training and research, information dissemination and evaluation, especially for rare diseases. This Directive should therefore give incentives to Member States to reinforce the continued development of European reference networks. European reference networks are based on the voluntary participation of their members, but the Commission should develop criteria and conditions that the networks should be required to fulfill in order to receive support from the Commission.

(…)

Article 12
European reference networks

1. The Commission shall support Member States in the development of European reference networks between healthcare providers and centres of expertise in the Member States, in particular in the area of rare diseases. The networks shall be based on voluntary participation by its members, which shall participate and contribute to the networks' activities in accordance with the legislation of the Member State where the members are established and shall at all times be open to new healthcare providers which might wish to join them, provided that such healthcare providers fulfill all the required conditions and criteria referred to in paragraph 4.
2. European reference networks shall have at least three of the following objectives:
   a) to help realise the potential of European cooperation regarding highly specialised healthcare for patients and for healthcare systems by exploiting innovations in medical science and health technologies;
   b) to contribute to the pooling of knowledge regarding sickness prevention;
   c) to facilitate improvements in diagnosis and the delivery of high-quality, accessible and cost-effective healthcare for all patients with a medical condition requiring a particular concentration of expertise in medical domains where expertise is rare;
   d) to maximise the cost-effective use of resources by concentrating them where appropriate;

e) to reinforce research, epidemiological surveillance like registries and provide training for health professionals;
f) to facilitate mobility of expertise, virtually or physically, and to develop, share and spread information, knowledge and best practice and to foster developments of the diagnosis and treatment of rare diseases, within and outside the networks;
g) to encourage the development of quality and safety benchmarks and to help develop and spread best practice within and outside the network;
h) to help Member States with an insufficient number of patients with a particular medical condition or lacking technology or expertise to provide highly specialised services of high quality.

3. Member States are encouraged to facilitate the development of the European reference networks:

a) by connecting appropriate healthcare providers and centres of expertise throughout their national territory and ensuring the dissemination of information towards appropriate healthcare providers and centres of expertise throughout their national territory;
b) by fostering the participation of healthcare providers and centres of expertise in the European reference networks.

4. For the purposes of paragraph 1, the Commission shall:

a) adopt a list of specific criteria and conditions that the European reference networks must fulfil and the conditions and criteria required from healthcare providers wishing to join the European reference network. These criteria and conditions shall ensure, inter alia, that European reference networks:
    i. have knowledge and expertise to diagnose, follow-up and manage patients with evidence of good outcomes, as far as applicable;
    ii. follow a multi-disciplinary approach;
    iii. offer a high level of expertise and have the capacity to produce good practice guidelines and to implement outcome measures and quality control;
    iv. make a contribution to research;
    v. organise teaching and training activities; and
    vi. collaborate closely with other centres of expertise and networks at national and international level;
b) develop and publish criteria for establishing and evaluating European reference networks;
c) facilitate the exchange of information and expertise in relation to the establishment of European reference networks and their evaluation.

5. The Commission shall adopt the measures referred to in paragraph 4(a) by means of delegated acts in accordance with Article 17 and subject to the conditions of Articles 18 and

19. The measures referred to in points (b) and (c) of paragraph 4 shall be adopted in accordance with the regulatory procedure referred to in Article 16(2).

6. Measures adopted pursuant to this Article shall not harmonise any laws or regulations of the Member States and shall fully respect the responsibilities of the Member States for the organisation and delivery of health services and medical care.

Article 13

Rare diseases

The Commission shall support Member States in cooperating in the development of diagnosis and treatment capacity in particular by aiming to:

a) make health professionals aware of the tools available to them at Union level to assist them in the correct diagnosis of rare diseases, in particular the Orphanet database, and the European reference networks;
b) make patients, health professionals and those bodies responsible for the funding of healthcare aware of the possibilities offered by Regulation (EC) No 883/2004 for referral of patients with rare diseases to other Member States even for diagnosis and treatments which are not available in the Member State of affiliation.

Council Recommendation (2009/C 151/02) of 8 June 2009 on an action in the field of rare diseases

(…)

(13) In July 2004, a Commission High-Level Group on Health Services and Medical Care was established to bring together experts from all Member States to work on practical aspects of collaboration between national health systems in the EU. One of this High-Level Group's working groups is focusing on European Reference Networks (ERNs) for rare diseases. Some criteria and principles for ERNs have been developed, including their role in tackling rare diseases. ERNs could also serve as research and knowledge centres, treating patients from other Member States and ensuring the availability of subsequent treatment facilities where necessary.

(14) The Community added value of ERNs is particularly high for rare diseases by reason of the rarity of these conditions, which implies both a limited number of patients and a scarcity of expertise within a single country. Gathering expertise at European level is therefore paramount in order to ensure equal access to accurate information, appropriate and timely diagnosis and high quality care for rare disease patients.

(15) In December 2006 an expert group of the European Union Rare Diseases Task Force issued a report 'Contribution to policy shaping: for a European collaboration on health services and medical care in the field of rare diseases' to the High-Level Group on Health Services and Medical Care. The expert group report outlines, inter alia, the importance of identifying centres of expertise and the roles that such centres should fulfil. It is also agreed that, in

principle and where possible, expertise should travel rather than patients themselves. Some measures called for in the report are included in this recommendation.

(16) Cooperation and knowledge sharing between centres of expertise has proven to be a very efficient approach to dealing with rare diseases in Europe.

(17) The centres of expertise could follow a multidisciplinary approach to care, in order to address the complex and diverse conditions implied by rare diseases.

(…)

IV. CENTRES OF EXPERTISE AND EUROPEAN REFERENCE NETWORKS FOR RARE DISEASES

11. Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.
12. Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.
13. Organise healthcare pathways for patients suffering from rare diseases through the establishment of cooperation with relevant experts and exchange of professionals and expertise within the country or from abroad when necessary.
14. Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.
15. Include, in their plans or strategies, the necessary conditions for the diffusion and mobility of expertise and knowledge in order to facilitate the treatment of patients in their proximity.
16. Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.

V. GATHERING THE EXPERTISE ON RARE DISEASES AT EUROPEAN LEVEL

17. Gather national expertise on rare diseases and support the pooling of that expertise with European counterparts in order to support:

a) the sharing of best practices on diagnostic tools and medical care as well as education and social care in the field of rare diseases;
b) adequate education and training for all health professionals to make them aware of the existence of these diseases and of resources available for their care;
c) the development of medical training in fields relevant to the diagnosis and management of rare diseases, such as genetics, immunology, neurology, oncology or paediatrics;
d) the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences;
e) the sharing of Member States' assessment reports on the therapeutic or clinical added value of orphan drugs at Community level where the relevant knowledge and expertise is gathered, in order to minimise delays in access to orphan drugs for rare disease patients.

The correct form when quoting this document is*:*

EUCERD Recommendations on Quality Criteria for Centres of Expertise for Rare Diseases in Member States, 24 October 2011
http://www.EUCERD.eu/upload/file/EUCERDRecommendationCE.pdf

Photography : P. Latron/Inserm