EUROPLAN
European Project for Rare Diseases National Plans Development
Coordinated by the Italian National Centre for Rare Diseases
Italian National Institute of Health
2012-2015

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EURORDIS
Rare Diseases Europe

EUCERD Joint Action / Work Package 4
EUROPLAN National Conference

Workshop Theme 4
Centres of Expertise and
European Reference Networks for Rare Diseases

EUCERD
Joint Action

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Relevant extracts from the

COUNCIL RECOMMENDATION
on an action in the field of rare diseases
(2009/C 151/02)

8 June 2009

COUNCIL RECOMMENDATIONS

(The Council of the EU) “HEREBY RECOMMENDS that Member States:

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

- Identify appropriate centres of expertise throughout their national territory by the end of 2013, and consider supporting their creation.

- Foster the participation of centres of expertise in European reference networks respecting the national competences and rules with regard to their authorisation or recognition.

- 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.

COUNCIL RECOMMENDATIONS

- Support the use of information and communication technologies such as telemedicine where it is necessary to ensure distant access to the specific healthcare needed.

- 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.

- Encourage centres of expertise to be based on a multidisciplinary approach to care when addressing rare diseases.

COUNCIL RECOMMENDATIONS

V. GATHERING THE EXPERTISE ON RARE DISEASES AT EUROPEAN LEVEL

- 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;

COUNCIL RECOMMENDATIONS

- (d) the development of European guidelines on diagnostic tests or population screening, while respecting national decisions and competences;

- (e) the sharing 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.

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EUCERD RECOMMENDATIONS ON CORE INDICATORS FOR RD NATIONAL PLANS / STRATEGIES

EUCERD Core Indicators, full version:
http://www.eucerd.eu/wp-content/uploads/2013/06/EUCERD_Recommendations_Indicators_adopted.pdf

Core Indicators

N°5. Existence of a national policy for establishing Centres of Expertise (CE) on RD

Short definition =
Existence of a strategy to identify and designate RD CEs at national/regional level

N°6. Number of national and regional Centres of Expertise adhering to the national policy

N° of CEs complying with national policy and n° CEs in line with EUCERD Recommendations on Quality Criteria for CEs that help MS designate their CEs

Core Indicators

N°7. Participation of national or regional Centres of Expertise in European Reference Networks (ERNs)

Indicator counts the CEs integrated in ERNs, differentiating between full and associated members. PS: more relevant in a few years when ERNs will be more established

N°10. Existence of a national policy on rare disease clinical practice guidelines development and implementation

Whether a national policy exists on developing, adapting and implementing clinical practice guidelines with the aim to ensure equal access to care for all RD patients

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GUIDELINES FOR DISCUSSION

Workshop Theme 4
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1. Designation and evaluation of CE

MAPPING OF CEs

- Starting from the recognition that expertise on RDs exist in all countries, what is the level of knowledge of the existing expertise in your country?
- Is there a mapping of structures providing expertise on rare diseases?
- Have their different roles and competences been acknowledged?

1. Designation and evaluation of CE

DESIGNATION CRITERIA for CEs

- Are designation criteria for CEs being defined? If not, is there a procedure in place to define and approve such designation criteria?
- Are the designation criteria such to adapt to the characteristics of the disease or group of diseases covered by each CE?
- What sort of quality management is ensured within CEs throughout the national territory?
- Please compare the designation criteria adopted in your country with the EUCERD Recommendations on Quality Criteria for CEs. **What recommended criteria are missing? Which ones could be incorporated?**

1. Designation and evaluation of CE

DESIGNATION PROCESS OF CEs

- At what stage of development is the process of designation of CEs in your country?
- What designation process is in place or will be put in place to designate CEs?
- Considering the mapping of existing resources, how to better rationalise the existing resources instead of creating new ones and new centres, while ensuring compliance with agreed quality criteria and standards?
- Are patient organisations involved in the designation of CEs?
- Is there a national directory of CEs? Is it publicly accessible?

1. Designation and evaluation of CE

EVALUATION OF CEs

- Are CEs evaluated on a regular basis?
- Is this evaluation process incorporated into the designation process at national level?
- Which “quality of care” indicators are adopted and what outcome measures are considered? Do they include “patient satisfaction” as a minimum?
- What actors are involved in the evaluation process? Specifically, are patients and patient organisations involved in the evaluation process of CEs?

1. Designation and evaluation of CE

INFORMATION ON AVAILABLE EXPERTISE AND CEs

- How expertise available in your country and CEs are made known and accessible to patients?
- Is there a publicly available national directory of formally designated CEs, including on the Orphanet portal?
- What other sources of information are used apart from Orphanet?

2. Scope and functioning of CEs

DEFINITION

- Analyse the (explicit or implicit) **definition of CEs** in your country and compare with the EUCERD Recommendation on Quality Criteria for CEs:

  "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. 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. Scope and functioning of CEs

SCOPE

- What **scope of CEs** in your country?
- Does the combined scope of all CEs within your country cover all RD patients' needs?
- How to ensure that all patients living with a RD have access to the appropriate CE in your country or abroad?
- Especially in smaller countries, where the existing expertise cannot possibly cover all RDs, do CEs rely on networks of experts?
- What distribution of competences does exist between the national and the regional level?

2. Scope and functioning of CEs

INVOLVEMENT OF PATIENTS & PATIENT ORGANISATIONS

- What type of collaboration with patients and their organisations is established in CEs?
- How to ensure that patient representatives are closely involved in the management and decision-making processes of CEs in a systematic manner?
- Are patients involved in those areas where they can be specific added value e.g. social counselling?
- How do individual patients are heard and involved (e.g. when a patient organisation does not exist for the specific disease(s) covered in the scope of the CE) ?

2. Scope and functioning of CEs

HIGH LEVEL OF EXPERTISE AND MOBILITY OF EXPERTISE

- How is the level of expertise available in CEs measured and accounted for?
- How do CEs share expertise amongst themselves and through networks of expertise in the country and abroad?
- What solutions are provided in order to support the mobility of expertise:

  a) amongst CEs;

  b) CEs and diagnostic laboratories;

  c) providers of care at local level?

2. Scope and functioning of CEs

HIGH LEVEL OF EXPERTISE AND MOBILITY OF EXPERTISE

- What is the role of e-health solutions?
- What concrete solutions do exist and/or could be put in place? e.g. shared case management systems, expert systems for tele-expertise, shared repository of cases, etc.
- What solutions are or could be prescribed “to bring highly specialised expertise on rare diseases to ordinary clinics and practices, such as a second opinion from a centre of excellence”? (*EUROPLAN Recommendations*)

3. Multidisciplinarity, healthcare pathways & continuity of care

- Are CEs in your country based on a multidisciplinary approach?

  Are they capable of bringing together, or coordinating, “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”? (*EUCERD Recommendations*)

- Do CEs have links with specialised laboratories and other facilities?

3. Multidisciplinarity, healthcare pathways & continuity of care

- What opportunities are provided for "**education and training** for healthcare professionals from all disciplines, including paramedical specialists and non-healthcare professionals (such as school teachers, personal/homecare facilitators)" ?
- How do CEs contribute to build "healthcare pathways" from primary care?
- How to develop a system based on the adoption of healthcare pathways for the provision of care?
- How do CEs link to local structures providing day-to day care?

3. Multidisciplinarity, healthcare pathways & continuity of care

- How to link medical expertise of specialised CEs to local medical, paramedical and social care?
- What solutions are provided for to support the mobility of expertise from CEs to local care providers, so as to allow the **treatment of patients in their proximity**?
- Specifically, what e-heath solutions could support the task?
- What mechanisms can be found to integrate paramedical and rehabilitation interventions (provided that they are prescribed by the CE) into the reimbursement schemes?

3. Multidisciplinarity, healthcare pathways & continuity of care

- Do CEs take into account the **ageing** of patients?
- Do they envisage “collaborations to assure the continuity of care between childhood, adolescence and adulthood, if relevant?”
- Is it envisaged that CEs organise “collaborations to assure the continuity of care between **all stages of the disease**”? How are these collaborations ensured in practical terms?

4. Access to information

- How do CEs “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”?

- In particular, what role do patient associations have in the provision of accessible information tailored to the needs of different users?

5. How to integrate research on RDs and provision of care

- What is the role of CEs in research?
- Do CEs contribute “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”?
- Do they contribute to state of the art research on relevant RDs? Do they have the capacity to participate in clinical trials?
- Is there a list of RD registry run by CEs accessible to the public?
- Do CEs that run a rare disease registry systematically involve the patient groups that are concerned by the disease being studied in the registry?

6. Good practice guidelines

- How do CEs “contribute to the elaboration of good practice guidelines and to their dissemination”?
- How are patients and their representatives involved in their development?
- How do CEs coordinate among themselves or network with other similar bodies to develop good practice guidelines?
- How could experts and CEs better contribute to the development of international/European good practice guidelines?

6. Good practice guidelines

- What measures do exist to adopt and/or adapt guidelines developed in other countries or by other international bodies where they don't exist for certain specific diseases, so to optimise efforts and resources?

- Do CEs recognise and adhere to existing good practice guidelines for the RDs they deal with?

7. Diagnostic and genetic testing

DIAGNOSTIC LABORATORIES

- How to compile an inventory of medical laboratories providing testing for RD?
- Is there an accreditation process for such laboratories based on quality criteria?
- How to ensure support and networking of such laboratories in order to have dedicated infrastructures and resources for the biological component of RDs?
- How to link them to CEs in a structured way?
- How to ensure partnership with laboratories outside the country when not available at national level?

7. Diagnostic and genetic testing

TRAVELLING OF DIAGNOSTIC MATERIAL & TELE-EXPERTISE

- What arrangements do exist to enable the travelling of biological samples, radiological images as well as other diagnostic material?
- How to organise DNA and sample exchanges at the national and European level?
- What sort of reimbursement agreements and policies do support these exchanges?

7. Diagnostic and genetic testing

GENETIC COUNSELLING

- What measures are in place to ensure families are directed towards the most appropriate diagnostic testing and rare disease centres of expertise?
- Is genetic counselling an integral part of genetic testing, made easily accessible and provided before and after genetic testing in CEs?
- Is it provided by adequately trained healthcare professionals?
- How to best integrate the EuroGenTest recommendations on genetic counselling into national practices?

8. Screening policies

NATIONAL LEGAL FRAMEWORK

- What population screening programmes – especially newborn screening (NBS) - do exist in your country?
- What measures are in place / should be adopted to ensure that existing NBS programmes are as comprehensive as possible?
- What measures could be put in place to evaluate and improve their performance and their actual coverage of population?
- What policies are envisaged to monitor changes in the population which can justify the provision of targeted screening practices?
- What legal basis does support NBS practices? If mandatory, are they accompanied by transparent and clear information to parents?

8. Screening policies

NATIONAL LEGAL FRAMEWORK

- "The national legal basis might furthermore regulate consistently the following issues:
  - the storage and the delayed use of samples and the associated consent;
  - the identification of eligible benefits;
  - the communication of results to parents and/or patients, including unintended findings;
  - the collection and communication of data for the assessment of the programme and for improving the knowledge on disease and treatment;
  - ensure quality control and quality assurance;
  - sustain funding".

(*from the Executive Report to the EC on NBS in the EU)*

8. Screening policies

DECISION MAKING PROCESS

- How to ensure that the **decision-making process** is associated to the national technology assessment process?

- What arrangements could be envisaged to involve patients and patient groups in the decision-making process for including additional NBS practices?

8. Screening policies

COLLABORATION AT EU LEVEL

- How to enable **collaboration on screening policies at EU level -** notably to deal with the assessment of elements that are common to all countries and are better dealt with at the EU level (e.g. efficacy of treatments, reliability of screening tests) without impairing the national competence?

- Does your country participate to existing committees ensuring collaborative efforts in this field e.g. the EUnetHTA?

- How to facilitate sharing documents and experiences for the benefit of countries that do not have the material available as yet?

8. Screening policies

AWARENESS AND TRAINING

- What **training courses** could be provided for all stage of NBS, and in particular on the communication to parents of the diagnostic suspicion and of positive confirmed diagnoses?

- What specific support and funding could be provided to patient groups with regard to patient empowerment after diagnosis?

  (e.g. by providing guidelines regulating the involvement of professions in the treatment of patients with disorders they screen for).

8. Screening policies

AWARENESS AND TRAINING

- In addition to NBS policies – that concerns only a limited number of RDs - what measures could be put in place to enable doctors and in particular neonatologists to investigate unusual symptoms in newborns?

- What specific training programmes could be developed by CEs for this purpose?

9. European and international collaboration – Cross-border healthcare and ERNs

PATIENTS' RIGHTS TO CROSS BORDER HEALTHCARE

- What national measures need to be adapted/ changed to comply with the Cross Border Health Care (CBHC) Directive and ensure equality of access and treatment of patients from all over the EU?

- How is the country defining the **list of treatments for which prior authorisation** is required according to the CBHC Directive?

- How to avoid too narrow definitions that could hamper the referrals of RD patients to healthcare providers abroad?

9. European and international collaboration – Cross-border healthcare and ERNs

PATIENTS' RIGHTS TO CROSS BORDER HEALTHCARE

- When a **CE sends a patient abroad for a second opinion**, is the authorisation automatically delivered?

  Although not compulsory under the CBHC rules, this is essential, as the decision of a recognised expert cannot be put into question.

- Is your country setting up **National Contact Points** as per the CBHC Directive?

  Are they equipped with information on which type of care is available in other EU countries, costs, rights and practical aspects on cross border care that will be received, in order to enable patients to make informed choices?

9. European and international collaboration – Cross-border healthcare and ERNs

PATIENTS' RIGHTS TO CROSS BORDER HEALTHCARE

- Are other measures envisaged to address the specific information needs of healthcare professionals, patient organisations and citizens in general?
- Are **travel and accommodation costs** reimbursed to patients travelling abroad under the EU rules?
  Member States have the option to reimburse or not these costs (Art.7.4 of CBHC Directive)
- Are **e-Health** measures implemented to access patient's written or electronic medical records?
- What solutions are put in place to ensure compatibility between health IT systems and that data is fully readable?

9. European and international collaboration – Cross-border healthcare and ERNs

EUROPEAN REFERENCE NETWORKS (ERNs)

- As a preparatory ground for the participation in ERNs, what specific measures do already exist that foster the connection of CEs and healthcare providers throughout the country and the sharing of information amongst them?

- Is there a framework in place to promote and support the common process for designation of national CEs and healthcare providers that may be affiliated in the future RD ERNs?

9. European and international collaboration – Cross-border healthcare and ERNs

EUROPEAN REFERENCE NETWORKS (ERNs)

- What measures do support in national centres the “core components” of a RD ERN as per the EUCERD Recommendations on ERNs? Notably:
  - disease registries,
  - quality assurance mechanisms for laboratory testing,
  - training and education tools,
  - information flow for good practice guidelines and best practices of diagnosis and care amongst MS,
  - telemedicine, cross border referral mechanisms, etc…

Reference: EUCERD Recommendations on RD ERNs

9. European and international collaboration – Cross-border healthcare and ERNs

EUROPEAN REFERENCE NETWORKS (ERNs)

- Where there may be no or limited number of CEs:

  Is there already a reflection process on measures to support the inclusion of healthcare providers in future ERN?

- What national measures do need to be put in place in order to support the establishment of RD ERNs under the terms of the Cross Border Healthcare Directive and to embed them in the national healthcare system?

- Specifically, what criteria does a national centre need to meet to become part of an ERN?

10. Sustainability of CEs

- What mechanisms do ensure that CEs are established and operate at national level in line with a sustainable plan?
- How is the long-term sustainability of CEs accounted for?
- What mechanisms do exist to verify the long-term sustainability of CEs at the moment of designation? And at the moment of evaluation?
- How are activities performed by the CEs but not strictly related to patient treatment funded (e.g. clinical research, production of guidelines for diagnosis and care; in-depth clinical and biological investigations; coordination of international networks, etc.)?

10. Sustainability of CEs

- How to make best use of **Structural Funds** in the forthcoming period 2014-2020?
- Is there scope for investments in rare disease CEs in the national strategic reference frameworks for Structural Funds?
- Are there in the country specific Operational Programmes for Health where projects for RDs infrastructures and human resources could be included?

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PROPOSALS

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