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Rare Diseases Europe

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

Moving forward: Key common issues that need to be taken into account in National Plans

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EURORDIS-EUROPLAN
Advisors Committee

CONSENSUS HAS ALREADY BEING BUILT ON KEY COMMON ISSUES

A solid policy and legal basis

- Communication of the Commission « Rare diseases, Europe's challenge » (2008)
- Council Recommendation on rare diseases (2009)
- Directive on the application of patients' rights in cross-border healthcare (2011) - establishing ***European reference networks***
- **EUCERD Recommendations:**
  - on **Quality Criteria of Centres of Expertise**, 2011
  - on **European Reference Networks**, 2013
  - on the information flow on **Clinical Added Value of Orphan Medicinal Products** (**CAVOMP**), 2012

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CONSENSUS IS CURRENTLY BEING BUILT ON SOME OTHER KEY COMMON ISSUES

- **Coding & Classification of all rare diseases**

- **EUCERD Recommendations on RD patient registries & Data collection** :
  - EMA /EUCERD workshops
  - EURORDIS CORD NORD Joint Declaration
  - EpiRare project, PARENT project…
- **Information to patients, families and professionals:**
  - Network of national rare disease information helplines with a free number – towards a unique number in EU
- **Research:**
  - IRDiRC and E-RARE project
- **Core Indicators for National Plans**

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NEW CHALLENGES MUST BE INTEGRATED

- **Recognition of rare diseases** and **integration in mainstream health and social services** in a time of severe economic constraints

- **Diagnosis for all**, including **very rare** and **still undiagnosed** diseases

- **Good practice guidelines** for diagnostic and care for all diseases or groups of diseases when relevant

The EUCERD : A place to build consensus, exchange best practices and foster pogress

- A European Committee of Experts on Rare Diseases

  with representatives from all Member States and other stakeholders: patient representatives, researchers, clinicians, industry

- Prepares new Recommendations on:
  - **Patient Rare Diseases Registries** (**ongoing**)
  - **Indicators for Monitoring RD National Plans** (**ongoing**)
  - New Born Screening (tbc)
  - Quality Testing (tbc)
  - Good Practice Guidelines for Diagnostic & Care… (tbc)

The EUCERD : A place to build consensus, exchange best practices and foster pogress

KEY COMMON ISSUES :

1. Centres of Expertise
2. European Reference Networks
3. Improved Access to Orphan Medicinal Products
4. Registries and data collection
5. Research: international initiatives & national issues
6. Coding and Classification
7. Access to diagnosis for all

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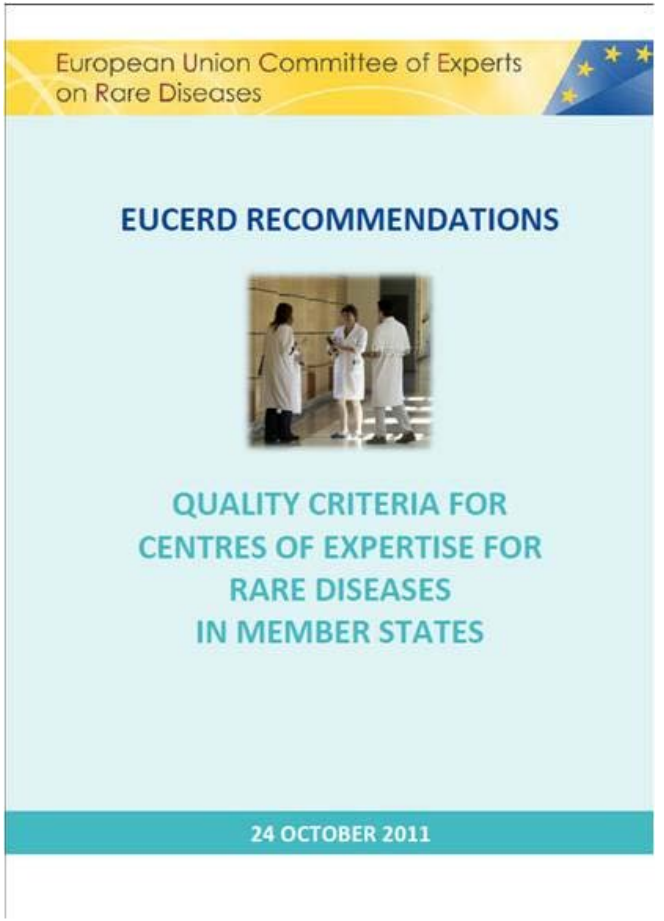
1. Identification and support to Centre of Expertise: Make the best use of the EUCERD quality criteria

HIGHLIGHTS

- Coordinate **multidisciplinary skills**, including **paramedical** & **social services**
- Contribute to building **health care pathways** from primary care
- Collaborate with **patients' organisations**
- Centres **designated by Member States**
- Organise collaborations for the **continuity of care** from childhood to adulthood and at all stages of the disease and, if necessary, organise referrals to other countries, produce guidelines



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1. Identification and support to Centres of Expertise: Be realistic!

Consider a step wise approach :

- Identification of experts in your country, supported to coordinate multidisciplinary skills with some budget allocation
- Candidate centres encouraged to define their current actions, their goals and their strategy to attain designation criteria
- **Patient organisations** actively involved at all levels: identification, collaboration in their activities, internal and external evaluation

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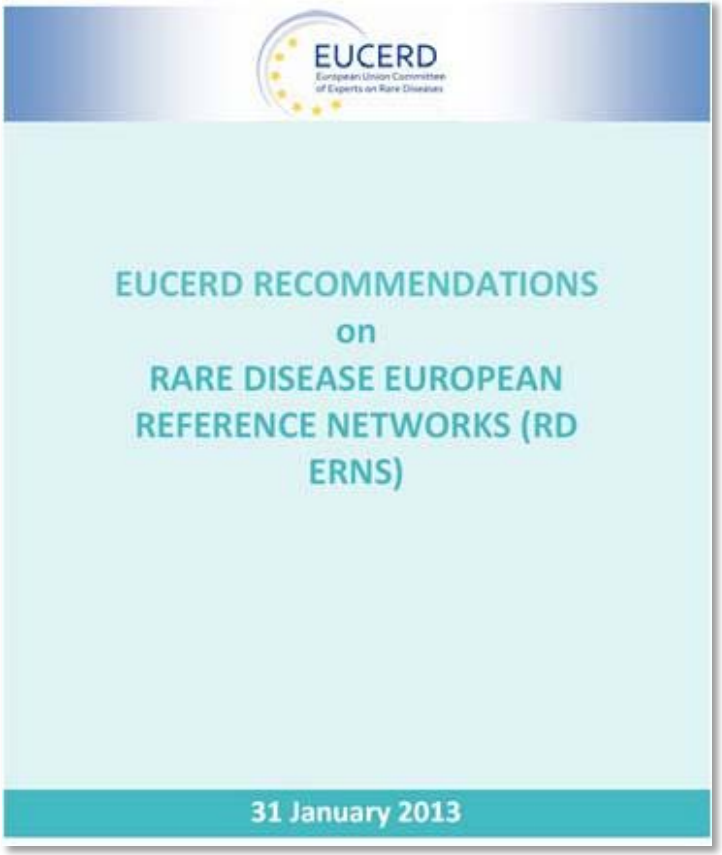
1. Identification and support to Centres of Expertise: Be realistic!

- Collaboration with other centres and experts at national, European and international level being essential
- Health authorities should implement mechanisms to measure performance and progress
- **KEEP in MIND**: The combined scope of centres should cover all patients needs at national level in the long term
- The availability of a National directory accessible to patients will help identifying and advocating for unmet needs

2. European Reference Networks (ERNs) for RDs: the EUCERD Recommendations

HIGHLIGHTS

- RD ERNs will link Centres of Expertise and specialised healthcare providers, social care providers, patient groups, diagnosis labs, research groups …
- Flexible framework for healthcare pathways to patients
- Will facilitate mobility of expertise + cross border healthcare for RD



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2. European Reference Networks (ERNs): our vision

The EUCERD Recommendations will be only implemented with:

- Identification and funding of CEs and specialised health and social care providers in all countries (funding being a EU MS competence)
- A **stepwise strategy for designation**, so **all RD patients** will have access to appropriate ERN in a defined period of time
- Based around the concept of medical specialties, diagnostic and therapeutic areas should be identified to have **approximately 20 to 30 ERNs, covering a wide range of RDs**
- **Long-term adequate funding** of the EU to support coordination and networking activities
- **Shared platforms and tools**: Registries, guidelines, training/ education, communications, telemedicine, quality assurance mechanisms for laboratory testing, indicators of performance

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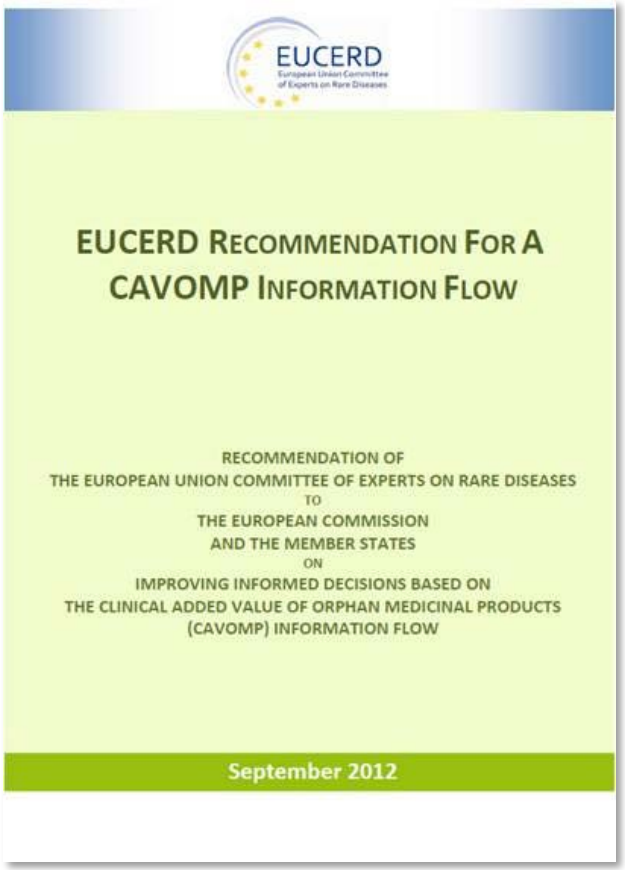
3. Improve Access to Orphan Medicinal Products: The EUCERD Recommendations on CAVOMP

The **CAVOMP :**

A process for exchanging knowledge between EU MS

respecting their responsibilities and the current regulatory processes

building on EMA and HTA agencies' (EUnetHTA) collaboration



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3. Improve Access to Orphan Medicinal Products: The EUCERD Recommendations on CAVOMP

HIGHLIGHTS

- **AIM of the CAVOMP**: **common European assessment** of the **Clinical Added Value** of an OMP
- **Relevant stakeholders involved at different and defined stages of OMPs development**: sponsor, patients, treating physicians, EMA, EuNetHTA, Centres of Expertise / ERNs
  - A **single report** based on existing assessments by relevant experts from Member States will be made at time of the Marketing Authorisation
  - **Helps National Authorities to fix OMPs' price & reimbursement** based on the common assessment report

4. Rare diseases patient registration and data collection : EUCERD recommendation

DRAFT TO BE ADOPTED on June 6

- **RD patient registries and data collections need to be internationally interoperable**

  The procedures to collect and exchange data need to be **harmonised and consistent**, to allow pooling of data, to reach sufficient statistically significant numbers for clinical research and public health purposes

- Use **international standards & nomencla**ture to code the diagnosis: either the OMIM code or the Orpha codes, alongside with current system: ICD and SNOMED-CT

- **Adopt a minimum common data set across rare disease**s, in collaboration with global initiatives

- **Adopt appropriate core data sets for disease-specific registries -** In the future, disease-specific registries could fall under the remit of RD ERNs

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4. Rare diseases patient registration and data collection : EUCERD recommendations

DRAFT TO BE ADOPTED on June 6

- **All sources of data should be considered as sources of information**, to speed up the acquisition of knowledge and the development of clinical research (including data DIRECTLY reported by patients)
- **Collected data should be used for public health and research purposes**: policy development, monitoring of care provision and therapeutic interventions, including off-label use of approved drugs and existing medications, multi-centre clinical studies
- **Multi-stakeholders model for registry governance**
- **Consent process in line with legal requirements at EU and International levels**
- **Sustainability**

5. Research : European and international initiatives and national issues

- EU MS to join **E-RARE**: Support collaborative research **at European level**
- Participate in **IRDiRC**: **International collaboration** to deliver 200 new therapies for RDs and means o diagnose most RDs by **2020**
- Encourage research on the burden of diseases on daily life, access to diagnosis, medical and social care, and evaluation of medical and social services at national level
- Promote public-private partnership

6. Coding and Classification

- **WHO working group chaired by the EU (Orphanet)**
- **Increased use of Orphacode**
- Support to **Orphanet for indexing the functional consequences of rare diseases with the Orphanet Disability Thesaurus**, based on the International Classification of Functioning, Disability and Health of WHO : contribution from 33 countries for 781 diseases
- To participate: ***disability.orphanet@inserm.fr***
- **and production of fact sheets on diseases and their consequences on daily life.**

7. Access to diagnosis for all, including very rare and still undiagnosed diseases

- **Extend the number of diagnosis reimbursed by the health care system** to all genetic and other diagnosis currently available at home or abroad
- **Making the best use of the Directive on cross border health care** to organise and reimburse the referral of DNA, biological samples or patients abroad if diagnosis of a disease is not available at home
- **Refer patients and families without diagnosis or with unclear diagnosis**, in particular in the case of mental disabilities, autistic spectrum disorders or recurrent psychiatric illnesses to research networks, in order to speed up their access to genome sequencing

OUR COMMON GOALS

To build with all stakeholders a comprehensive and long term strategy to address the daily needs of all rare disease patients everywhere in Europe of health and social services

To build a world model for research and provision of health and social services

A COMMON DEADLINE

“The Council of the European Union…hereby recommends that Member States

elaborate and adopt a **plan** or **strategy** as soon as possible, preferably **by the end of 2013** at the latest, aimed at guiding and structuring relevant actions in the field of rare diseases within the framework of their health and social systems”

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

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WHERE DO WE STAND ?

- **All 27 MS active**
- **Varying scope of National Plans**
- **Varying stage of development:**

1. Decision to elaborate a plan
2. Drafting group or stakeholder meetings
3. Plan submitted to national authority
4. Public consultation by national authority
5. National plan adopted
6. National plan implemented

LIKELY SCENARIO at Jan 1st, 2014

- **Possibly all or almost all 27 MS will have a plan adopted….**
- **…but :**
  - **not all areas covered** ***and/or***
  - **most actions without funding allocations** ***and/or***
  - **many policy measures difficult to implement** ***and/or***
  - **some disease areas left uncovered**

So we need to think to the next phase of National Plans !

NATIONAL PLANS, phase 2
IDENTIFYING BUILDING BLOCKS

- **The EU framework is established and in place (phase 1)**
- **From now we need to decide together the essential areas where concrete actions should be expected in all MS (phase 2)**

Starting from the
KEY COMMON ISSUES
that must constitute
the BUILDING BLOCKS
of the RD National Plans

Thank you!