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 2
DEFINITION, CODIFICATION, INFORMATION & TRAINING

EUCERD
Joint Action

Co-funded by the European Commission

Executive Agency for Health and Consumers

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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 RECOMMENDATION

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

II. ADEQUATE DEFINITION, CODIFICATION AND INVENTORYING OF RARE DISEASES

- Use for the purposes of Community-level policy work a common definition of rare disease as a disease affecting no more than 5 per 10 000 persons.

- Aim to ensure that rare diseases are adequately coded and traceable in all health information systems, encouraging an adequate recognition of the disease in the national healthcare and reimbursement systems based on the ICD while respecting national procedures.

COUNCIL RECOMMENDATION

- Contribute actively to the development of the EU easily accessible and dynamic inventory of rare diseases based on the Orphanet network and other existing networks as referred to in the Commission Communication on rare diseases.

- Consider supporting at all appropriate levels, including the Community level, on the one hand, specific disease information networks and, on the other hand, for epidemiological purposes, registries and databases, whilst being aware of an independent governance."

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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°4. Adoption of the EU RD definition

Short Definition =
Definition as laid down in OD Regulation EC 141/2000 & Cross Border Health Care Directive 2011/24/EU:
"no more than 5 patients per 10 000 persons"

N°8. NP/NS support to the development of/participation in a comprehensive national and/or regional RD information system

Existence of a nation-wide, comprehensive RD specific information system.
The participation in Orphanet Joint Action is also included.

Core Indicators

N°9. Existence of Help lines for RD

Refers to Help Lines for professionals only, for patients only and for both. Supported by private, public funding, or both.

N°11. Type of classification/coding used by the health care system

Type of coding system(s) used in view of better RD management and ultimately harmonising RD nomenclature

N°12. Existence of a national policy on registries or data collection on RD

Public support for their development and sustainability

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GUIDELINES FOR DISCUSSION

Workshop Theme 2
DEFINITION, CODIFICATION, INFORMATION & TRAINING

1. Definition of Rare Disease (RD)

The EU Regulation on Orphan Drugs 141/2000/EC defines RD as a disease "affecting no more than 5 per 10 000 persons".

- Is this definition adopted in your country?
- What is the status of this definition?

If the EU definition is not in use:

- How are the characteristics of RD defined?
- Assess readiness to adopt the EU definition & possible obstacles to its use in the country.

1. Definition of Rare Disease (RD)

- What mechanisms could be put in place to include diseases which don't strictly fall within the definition in National Plan for RD?

- How to ensure that **exceptions** to the definition are redressed or otherwise explained?

→ *some RDs are at the "frontier" of the definition:*

ex: thalassemia is not rare in some Mediterranean countries, but it is rare in some other European countries

2. Codification, Traceability of RD

- Which are the most used classification systems in your country: ICD9, ICD10, SNOMED, ICDO for rare cancers, MIM, ORPHA code…?
- For which purposes are they used (e.g. surveillance, reimbursement, provision of social support, etc)?
- Is your country part of Orphanet? if not, would you like it to be? Why and why not?

2. Codification, Traceability of RD

- What needs to be done in your country for a swift adoption of the WHO-led system, the ICD-11, when ready in 2015?
- What strategy could be put in place to ensure that all RD are coded and traceable in the health information and social systems?
- What initiatives are promoted at national level for the integrated use of administrative, demographic and health care data sources to improve the management of RD?

2. Codification, Traceability of RD

- What procedure, based on the ICD system, could be put in place to facilitate the smooth recognition of a disease as **‘rare disease’** in the national healthcare and social systems and thus allow people living with RD to access to both healthcare and social services?

- Is the ORPHA code already used in the hospitals and for data collection?

3. Registries and Databases

- Is there a policy for RD data collection and RD patient registration laid down in the RD National Plan/ Strategy?

- Are official lists of RD compiled in your country? For what purpose(s)?

- Is there an official governmental RD registry? And/or specific RD databases e.g. held by Centres of Expertise?

3. Registries and Databases

- What legal framework or mechanisms oversee the interactions among delocalised registries and central registries or databases, when the former are supposed to feed into a central registry or database?

- What mechanisms are / should be put in place to avoid overlaps?

- What measures do ensure the **interoperability** of different RD registries and the harmonisation of procedures to collect data and thus facilitate pooling of data for research/ public health purposes?

3. Registries and Databases

- How to stimulate the harmonisation of procedures and technical tools, in particular the development of minimum data sets, for both registries and biorepositories?
- Are RD patient registries using international standards and nomenclature to code the RD diagnosis, even if tentatively?
- What rules do ensure that **quality standards** of registries are consistently high?
- What system could ensure that **data directly reported by patients** are included along with data reported by clinicians?

3. Registries and Databases

- Assess and discuss the **participation and involvement of patients** in RD patient registries (in e.g. establishing registries, defining content, defining ethical issues, access to data; creating partnerships with healthcare professionals or industry; recruitment, etc).

- What measures could promote the involvement of patients as well as other stakeholders in the design, analysis and governance of RD registries?

- Is the NP / NS on RD also facilitating **access and sharing of data** to control how data is shared and published in the public domain?

3. Registries and Databases

- What other measures do ensure that RD patient registries adhere to good practice guidelines, including European and international ones?
- How to ensure, through appropriate funding mechanisms, the **long-term sustainability** of registries and databases?
- Do these registries and programmes receive government support?
- What space is there for private public partnerships to fund these infrastructures?

3. Registries and Databases

- Do RD patient registries usually envisage exit strategies in their work plans? What provisions are necessary to make sure that this occurs on a regular basis?

Discussion are ongoing on the **creation of a European Platform for Rare Disease Registration**, supported by the EC and aimed to provide common services and tools for the existing (and future) RD registries in the European Union.

→ What contribution could you country provide?

→ How a European Platform may help optimise national resources devoted to rare disease registration?

3. Registries and Databases

Additional questions from Workshop 3 on Research

- What initiatives & incentives are / should be in place to bring clinicians to actively participate in the data collection?

- How to motivate the sharing and open access to pre-competitive resources such as databases, biobanks or knowledge bases?

3. Registries and Databases

- How to engage in **international initiatives*** for the harmonisation and interoperability of RD registries, thus promoting the creation and functioning of registries with larger geographical scope?

  * Such as those promoted by the IRDiRC – International Rare Diseases Research Consortium

- Explore the feasibility of a common central resource or platform for creating or reconfiguring registries and describing the content of existing registries and databases.

4. Information on available care for RDs

- What are the existing information sources in your country? Are they of good quality? Please consider whether exist:
  - a national official website for RD;
  - help line(s), known to the public;
  - initiatives of Centres of Expertise and/or Patient Organisations or programmes to stimulate the development of information and educational material for patients or specific publics (teachers, social workers, etc.);
  - initiatives to raise awareness on RD, e.g. Rare Disease Day.
- Are ORPHANET, EURORDIS and other resources provided at EU level used in your country ?

4. Information on available care for RDs

- How are **specific disease** (or group of diseases) **information networks** organised?
- How is information exchanged amongst stakeholders and what level of involvement do they have?
- How are their European/international activities promoted and supported?
- How are they financially supported? Do they receive public funding?
- Discuss **mapping out information resources** on RD available at national level, and possibilities to introduce a system for **information quality assessment**.

5. Help Lines

- What kind of **help lines** (all diseases) exist in your country to assist RD patients and healthcare professionals?
- How to develop or consolidate existing patient-run help line services for RD?
- How to improve the service offered? How to improve their visibility, especially for patients?
- How are help lines financed? By private initiative or patient associations? Is there any government funding?
- How to ensure Help Lines' long-term sustainability?

5. Help Lines

- What national measures can be promoted to establish the **116 European number for RD Help Lines**?

  → With the “116 number”, the European Commission seeks to identify services of social value in Europe that could benefit from single European free phone numbers starting with 116.

NB: an application has been made by EURORDIS to the European Commission for a 116 number for rare disease help lines, but it needs to be supported by national administrations with specific measures.

6. Training healthcare professionals to recognise & code RDs

- What level of awareness and knowledge do healthcare professionals have of the RD classification and codification? What can be done to improve it?

- Due to the changes in the coding of RDs, which will appear in ICD11, what specific actions can be envisaged to prepare and train healthcare professionals in view of the introduction of ICD11?

7. Training healthcare professionals

- How are healthcare professionals trained in your country?
- **Discuss the various possibilities of trainings:**
  - Introduction of an **ad hoc educational package on RD in the curriculum of the medical degree course**
  - Development of training modules aiming to provide comprehensive care to RDs patients
  - Training on specific protocols for paediatricians to recognise relevant symptoms of RDs

7. Training healthcare professionals

- Specialist training on RDs for MDs in fields relevant to diagnosis, including post-graduate trainings
- Training opportunities for paramedical specialists and professional figures, such as e.g. people assisting patients with severe disabilities
- Measures and incentives for medical training of young doctors and scientists in the field of RD

7. Training healthcare professionals

- What measures can be taken to ensure adequate **lifelong training** of MDs and other healthcare professionals, including paramedical specialists?
- Discuss supporting measures and tools such as:
  - virtual universities
  - online training programmes or channels…
- Specifically, how could such IT infrastructures and programmes be supported?

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PROPOSALS

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