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

Logo NA
(conf. organiser)

EURORDIS
Rare Diseases Europe

EUCERD Joint Action / Work Package 4
EUROPLAN National Conference

Workshop Theme 3
RESEARCH FOR RARE DISEASES

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

- 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.

COUNCIL RECOMMENDATION

III. RESEARCH ON RARE DISEASES

- Identify ongoing research and research resources in the national and Community frameworks in order to establish the state of the art, assess the research landscape in the area of rare diseases, and improve the coordination of Community, national and regional programmes for rare diseases research.

- Identify needs and priorities for basic, clinical, translational and social research in the field of rare diseases and modes of fostering them, and promote interdisciplinary cooperative approaches to be complementarily addressed through national and Community programmes.

COUNCIL RECOMMENDATION

- Foster the participation of national researchers in research projects on rare diseases funded at all appropriate levels, including the Community level.

- Include in their plans or strategies provisions aimed at fostering research in the field of rare diseases.

- Facilitate, together with the Commission, the development of research cooperation with third countries active in research on rare diseases and more generally with regard to the exchange of information and the sharing of expertise.”

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

Developing, adapting and implementing clinical practice guidelines with the aim to ensure equal access to care for all RD patients

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

Ultimately, harmonising RD nomenclature worldwide will improve RD management

Core Indicators

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

Public support for their development and sustainability

N°13. Existence of RD research programmes and/or projects in the country

Describe the status of RD research in the country

N°14. Participation in European and international research initiatives

Ex: E-Rare, IRDiRC or other EU / international programmes

Core Indicators

N° 20. Specific public funds allocated for RD research

Policy decision(s) to allocate a portion of the National research budget specifically to RD research

N°21. Public funds specifically allocated for RD research actions/projects per year since the plan started

Total amount of public funds (in EUR) allocated to RD research projects or programmes

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GUIDELINES FOR DISCUSSION

Workshop Theme 3
RESEARCH FOR RARE DISEASES

1. Mapping of existing research resources, infrastructures and programmes for RDs

- Evaluation of RD research resources and infrastructures across different disciplines and sources of funds, both public and private.

- What is the scope of patient-driven research, i.e. research initiated and/or financed by patients and their associations?

- Is there a list or inventory of teams working at national level on RD research? Is it regularly updated?

  Are research projects on rare diseases identifiable and traceable within broader national research programmes?

2. Dedicated RD research programmes and governance of RD research funds

- Does a specific national RD research programme with dedicated funds exist? Is there a scope for such programme?
- How are funds allocated?
- What governance model does exist for handling RD research and related funds?

2. Dedicated RD research programmes and governance of RD research funds

- Is the creation of a “RD research centre”, such as the French “Fondation Maladies Rares”, embedded in the healthcare/research systems a viable option, acting as a one-stop shop for RD research projects?

  Please consider possible centralised activities such as:

  - centralised database of research projects on RD;
  - identification of priority and needs in the area of RD research;
  - centralisation of funding sources for RD research projects;
  - continuous funding schemes for RD research projects;
  - incubator for SMEs;
  - promotion of public-private partnerships with industry.

3. Sustainability of research programmes on RD

- How to ensure, through appropriate funding mechanisms, structural and long-term sustainability of research projects and research infrastructures in the field of RDs?

- Are there specific programmes to fund RD research, from basic and translational research?

- Do these programmes enable long term research by providing the assurance of long-term sustainability?

- Do national measures allow the reporting of research funded at the national level on RD?

3. Sustainability of research programmes on RD

- How are research programmes assessed?

- What mechanisms, in particular, do allow for the continuity and reiteration of successful initiatives and projects?

- What specific solutions should be devised in respect of **public health and social research** to optimise the provision of patient care and services for patients beyond healthcare?

- How are research infrastructures supported at national level? Has the possibility of recurring to EU Structural Funds for infrastructural projects been explored?

3. Sustainability of research programmes on RD

- Consider whether a combination of private and public funding is possible. How to engage private actors to fund RD research? What **public private partnership (PPP)** models are proposed? What type of research initiatives are better supported by PPP e.g. patients’ registries?

- Are there programmes to involve other private actors, different from the pharmaceutical industry, as for instance bank foundations?

4. Needs and priorities for RD research

- Is an assessment of needs and priorities for basic, clinical and translational research, as well as priorities for social research, been carried out in your country?

- How to best prioritise research needs in the country?

  Not everybody should do everything. How to make active choices aiming to provide good funding to good projects instead that little funding to many projects and not to duplicate efforts?

4. Needs and priorities for RD research

- How to make sure that **translational research** and the development of RD therapeutic solutions are ensured a prominent place in national prioritisation?

- How to improve awareness on the need for **research into quality of life, living conditions and social research on RDs** in general?

- How to ensure that funds are devoted to this type of research?

5. Participation of researchers and patient organisations in RD research projects

- What measures need to be adapted to foster **multi-centre studies** (both national and translational)?

- What **national networks** are necessary to support in order to promote RD research especially clinical and translational research?

- How to make the link between basic and translational research and **Centres of Expertise (CEs)**?

5. Participation of researchers and patient organisations in RD research projects

- What solutions could be devised in Centres of Expertise to “allow:

  1) researchers to fully integrate within clinical services; and

  2) clinicians to devote time to research without compromising care” (from the Final Report EUROPLAN I Conferences)?

- What mechanisms need to be put in place to facilitate the set-up of **clinical trials for small populations** run by academics in centres of expertise?

- How to best promote **interdisciplinary approaches** to research?

5. Participation of researchers and patient organisations in RD research projects

- What specific programmes target the recruitment of **young scientists** on RD research?
  - Are specific PhD programs on RDs proposed to students?
  - Are young researchers encouraged to enter the RD field via visible incentives to be foreseen?
- How to ‘institutionalise’ the participation of patients in research, especially in Centres of Expertise?
- How to strengthen the exchanges among **patient organisations**?
- How to promote the direct contact between researchers and patients (e.g. open labs day dedicated to patients, patients’ org.)?

5. Participation of researchers and patient organisations in RD research projects

- What other **collaborative modes** are supported at national level?

- Are different model of research collaboration supported ? For instance “charity networking” or cooperation among non-public non-industry players, such as charities and patient groups?

- How to support these actors, especially in the translational part of research where they usually lack means to translate excellent initial research into therapies?

6. RD research infrastructures and registries

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

- What rules do ensure that **quality standards** of registries are consistently high?

- 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?

6. RD research infrastructures and registries

- How to stimulate the harmonisation of procedures and technical tools, in particular the development of minimum data sets, for both registries and biorepositories?

- How to engage with **international initiatives** such as those promoted by the IRDiRC in favour of harmonisation and interoperability of RD registries and thus promoting the creation and functioning of registries with larger geographical scope?

- What initiatives and incentives are or should be in place to encourage researchers and clinicians to actively participate in the collection of data?

6. RD research infrastructures and registries

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

- What system could ensure that **data directly reported by patients** are included along with data reported by clinicians?

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

6. RD research infrastructures and registries

- How to motivate the sharing and open access to pre-competitive resources such as databases, biobanks or knowledge bases for the sake of maximising the scarce knowledge existing?

- What mechanisms do ensure the long-term sustainability of RD patient registries and other RD research infrastructures in your country? In particular:
  - 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?
  - How do different stakeholders share the financial responsibilities for the long-term sustainability of research infrastructure?
  - Are public-private partnerships considered as an option and if yes, how do they work and how are they regulated?

6. RD research infrastructures and registries

- Explore the feasibility of a common central resource or platform for creating or reconfiguring registries

- Discussions 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?

7. EU & international collaboration on RD research

- How to foster and support the participation of national researchers and laboratories, patients and patients organisations in EU-wide projects?
- Does your country plan to join **E-RARE** (for those who are not still partners) as the key programme supporting collaborative RD research at European level?
- Does your national research agency support the International Research consortium for Rare Disease Research (**IRDiRC)?**

7. EU & international collaboration on RD research

- What support is provided for collaborative research on RD through **European Reference Networks (ERNs)**?

- What national legislative measures do need to be put in place in order to support the development of ERNs as a catalyser of supranational collaborative RD research?

PROPOSALS

Workshop Theme 3
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