



EPIRARE

European Platform for Rare Disease Registries

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Luxembourg, October 26, 2011



EPIRARE

Project funded by the EU Commission (DG SANCO)

Starting date: **April 2011**

Duration: **30 months** (April 2011 – October 2013)

Coordinator : **National Centre for Rare Diseases, Istituto Superiore di Sanità – Rome, Italy**

General objective:

to build consensus and synergies to address regulatory, ethical, legal and technical issues associated with the registration of RD patients and to elaborate possible policy scenarios.



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IMPORTANCE OF REGISTRIES

Registries are scientific tools which can provide important information

- Natural history of the disease
- Epidemiological research
- Clinical research (patient recruitment for clinical trials)
- Disease surveillance
- Treatment evaluation (efficacy)
- Treatment monitoring (safety)
- Genotype-phenotype correlation
- Social planning
- Healthcare Services planning
-



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Rationale

- There are many registries (more than 500) on rare diseases with different aims with different coverage (regional, national, international) on a specific rare disease or on groups of them
- [The EU Council Recommendation on Rare Diseases \(adopted in June 2009\)](#) recommends **the implementation of registers and databases** for epidemiological purposes (point 15).
Therefore, many new initiatives are expected in the coming years.
BUT:
- There are **several difficulties** for EU data collection and exchange deriving from rules on data protection, quality issues, ownership of data.

“Today, even though many rare disease registries exist, the fact that they all operate in different ways, hinders the effective systematic data collection and sharing necessary for a positive effect on the health of patients”
(www.eurordis.org)



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EPIRARE: objectives

1. To analyse the present situation by conducting a survey targeting existing registries and to identify good practices

with reference to data collection; quality assurance; IT measures; legal issues; financial sustainability.

2. To build consensus and synergies to address regulatory, ethical, legal and technical issues associated with the registration of RD patients

3. To elaborate possible scenarios for EU policies on rare disease patient registries

4. To build consensus on scope and a governance model, representing the interests of relevant stakeholders, and to ensure the long-term sustainability of the EU platform for rare disease registries.



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A JOINT EFFORT

WP1 - Coordination of the project

WP Leader: **Domenica Taruscio**

National Centre for Rare Diseases, Istituto Superiore di Sanità, Rome (Italy)

WP2 - Dissemination of the project

WP Leaders: **Domenica Taruscio, Fabiola Gnessi**

National Centre for Rare Diseases, Istituto Superiore di Sanità (Italy)

WP3 - Evaluation of the project

WP Leaders: **Rumen Stefanov** Information Centre for Rare Diseases and Orphan Drugs, BAPES (Bulgaria); **Franco Cavallo** - University of Turin (Italy)

WP4 - Legal basis

WP Leader: **Tobias Schulte** - University of Maastricht (Netherlands)



Brussels, October 23, 2011



A JOINT EFFORT

WP5 - Policy scenarios on scope, aims, governance and long-term sustainability

WP Leader: Yann Le Cam - EURORDIS (France)

WP6 - Common data set and disease-specific data collection

WP Leader: Fabrizio Bianchi - National Council of Research Pisa (Italy)

WP7 - Data quality, validation and data sources integration

WP Leader: Manuel Posada de la Paz - Instituto de Salud Carlos III (Spain)

WP8 - Current needs of existing registries on rare diseases

WP Leader: Sabina Gainotti - National Centre for Rare Diseases, Istituto Superiore di Sanità (Italy)

Steering Committee, Advisory Board



Brussels, October 23, 2011



EPIRARE Partners

23 partners

14 countries

3 continents



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username: password:



In Europe a disease is considered rare when it affects no more than 5 individuals among 10,000 persons



A three-year project co-funded by the European Commission within the framework of the EU program of *Community Action* in the field Public Health

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[Deliverables & Meetings](#): [First Epirare Meeting](#)

Rome first Epirare meeting

11 - 12 July
2011
ISS

EPIRARE (European Platform for Rare Disease Registries) is a three-year project co-founded by the European Commission within the EU Program of Community Action in the field of Public Health.

The meeting will be held on **July, 11-12 2011 in Rome (Italy)**
Istituto Superiore di Sanità – Viale Regina Elena, 299

- Agenda
- Participants
- Accomodations
- Proceedings
- Meeting Secretariat
- How to get to the meeting site

Download
Epirare flyer



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Expectations from stakeholders - I

- Patients:

« a common reference framework, addressing scope, governance and long-term sustainability at the EU level,

will avoid wasteful fragmentation and duplication of time and resources,

and facilitate the setting up of more patient registries, especially for the rarest and most fragmented diseases throughout Europe» www.eurordis.org

“Patients do not want to see resources wasted to create a new register each time a new product is authorised for the same disease, or different research groups try to answer similar scientific questions”. (Eurordis)

- Industry:

- Reduce fragmentation/duplication of efforts
- Assessment of orphan drugs: safety, efficacy and cost-effectiveness



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Expectations from stakeholders - II

- **Researchers:**

- Guidelines and SOPs for specific data collection
- Identification of best practice models
- Sustainability
- “Freedom” of research

- **Public health institutions/authorities:**

- Comparability of data from different databases
- Connections with other data sources (prescriptions; disability support payments; death certificates; hospital discharge)
- Criteria for quality data
- Codification: with a view to ICD 11
- HTA
-



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The flyer

EPIRARE
European Platform for Rare Disease Registries

Background
The adoption of the EU Council Recommendation on rare diseases (2009/872/CE), which recommends support of registers and databases for epidemiological purposes, is expected to result in a burst of initiatives for rare diseases registration.

22 partners in 13 countries in 3 continents

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The project is co-funded by the European Commission within the framework of the EU program of Community Action in the field of Public Health. The project activities will develop from 2011 to 2013.

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Thanks for your attention
www.epirare.eu



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State of the Art

About this survey

Dear Colleague,

The European Commission has identified rare diseases registers as one of the priorities of the EU policy framework for rare diseases in Europe. According to the Commission Communication COM (2008) 679/2 on Rare diseases, registers and databases are key instruments to increase knowledge on rare diseases and develop clinical research.

Also, the recently launched Initiative International Rare Disease Research Consortium (IRDiRC) has identified registers, databases and biobanks as one of the priority means to foster transatlantic cooperation among EU, USA, Canada, China, Japan and other countries.

On 2010 the European Commission funded the EPIRARE Project (www.epirare.eu), in order to build consensus and synergies to address regulatory, ethical and technical issues associated with the registration of rare diseases patients and to elaborate possible policy scenarios.

Through this survey the EPIRARE project intends to explore the functioning, resources, problems, needs and expectations of existing registers on the European territory.

The final aim is to develop tools and services in support of existing registers and to favour the creation of new ones where needed. The survey is directed to both active and expired RD registers.

Your participation to the survey is of paramount importance as we believe that through joint efforts great advances can be contributed to the rare disease field.

Your participation to this survey will ensure your prompt access to the preliminary results and you will be further consulted on the preparation of support services and tools for RD Registers.

Any specific information on your register or database will be kept confidential.

Thank you in advance for your cooperation!

The EPIRARE coordinating team

- Survey on current activities and needs of RD registers
- 535 registers (sources: Orphanet, Treat NMD, Eurordis, DGSanco, DG Research, Hqip, etc.)
- Data collection will be launched this week, it will end on November 20°



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Explored areas

- General characteristics of RD registers
- Aims and scope (public health, research, other)
- Kind of data and register's sources
- Quality of data
- Ethical and legal issues
- Governance
- Access to data and sharing of data
- Security
- Sustainability
- Expectations and needs
- Expired registers



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