Development of the European Platform on Rare Diseases Registration

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The European Conference on Rare Diseases & Orphan Products
Berlin, 8-10 May 2014
Building the Platform:

- EU PLATFORM
- National registries
- Regional registries
- Local registries
- Hospital registries

Collaborations and interactions:

- IRDiRC
- EUROCAT
- RD-CONNECT
- ORPHANET JA
- EUCERD JA
- GRDR
- EPIRARE
- PARENT

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Development and Maintenance of the European Platform on Rare Diseases Registration

Why?
Rare Diseases Patient Registries (1/4)

- No single institution / no single country has sufficient numbers of patients to conduct clinical and translational research

- Geographic dispersion of rare diseases patients – major impediment to patient recruitment into trials

EU Platform to maximise output and use of data

to help reaching the critical mass of data for
- research/trials: clinical, epidemiological, etc.
- metaanalysis and comparisons across Member States and rare diseases
European Added-Value (2/4)

Based on the specificities of rare diseases:

- limited number of patients
- limited knowledge and expertise
- fragmentation of data sources across the Member States (600+ registries)

In addition

- section for ultra rare diseases
- section for non-diagnosed patients
European Commission’s Strategy in the Field of Rare Diseases (3/4)

1. To improve recognition and visibility on rare diseases
2. To support policies on rare diseases in the Member States
3. To develop European cooperation, coordination and regulation for rare diseases
Rare Diseases Patient Registries (4/4)

Key instruments for:

- increasing knowledge on RD
- supporting
  - clinical/translational research
  - epidemiological research
  - other studies
- contributing to EU policies:
  - health services planning - good clinical practices
    - therapy development and monitoring
    - research on care protocols
  - social services planning
Where?

At the European Commission's Joint Research Centre (JRC)

*The European Commission’s in-house science service*
The JRC in the European Commission

President
José Manuel Barroso

28 Commission Members

Commissioner
Máire Geoghegan-Quinn
Research, Innovation & Science

Director-General
Vladimír Šucha
Joint Research Centre

DG Research & Innovation (RTD)

www.jrc.ec.europa.eu
The JRC

- Established 1957
- 7 institutes in 5 countries
- 2822 permanent and temporary staff in 2012
- 1443 scientific publications in 2012
- Budget: €380 million annually, plus €69 million earned income

www.jrc.ec.europa.eu
The Mission of the Joint Research Centre

- To provide **scientific and technical support** for the conception, development, implementation and monitoring of **EU policies**

- Close to the policy-making process, it **serves the common interest** of the Member States, while being **independent of private or national special interests**

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IHCP Policy Support Areas

- Genetically Modified Organisms
- Nanotechnology
- Public Health
- Food and Consumer Products
- Chemical Assessment and Alternatives to Animal Testing

- Healthcare Quality
- Nutrition
- Health Information
- Behavioural Sciences
- Medical Devices
How?

Based on

**EUCERD Recommendations on rare disease patient registration and data collection**

1/3 Internationally interoperable, harmonised and consistent, to allow pooling of data

- international standards and nomenclature to code dg;
- adoption of a minimum common data set across RD (extendable with disease-specific data);

Complete data remain hosted by and property of individual registries.

shared tool (database and web interface)
Based on the output of EU-funded projects:

**EUCERD-JA**

- **Working Group on Registries**

  - proposed a **Common Data Set:**
    - Minimum data set / Basic data set /... / Expanded data set
    - based on contributions from MS, EU-funded projects (EUCERD-JA, EPIRARE, PARENT-JA), integrated with international initiatives (IRDiRC)

  ➡ **EU Platform: guidelines for sharing of data**
EUCERD Recommendations on rare disease patient registration and data collection

2/3 All sources of data should be considered to speed up the acquisition of knowledge and development of research
- registries: national, regional, local, hospital, research, etc.
- eHealth data
- patients entered data

3/3 Collected data utilised for public health and research purposes
EU Platform – Actions:

- **Pilot study**
  - use of the proposed common data set
  - **open call** for registries to participate

- **Setting up of the Advisory Board**
  - **open call** for interest – experts, all types of stakeholders
Goals of the Platform

- To serve as aggregation point for all RDs, across all EU MSs
  - open to other European Countries & Internationally
  - long term storage of the same data
  - quality processes, harmonization, standardization
  - link to the existing RD information networks

- To establish itself as a centre of RD-knowledge generation and to integrate the RD data into the wider context of health information, stratified medicine, biobanks, European Reference Networks

- To be a reference for EU / Member States policy makers

- To assure coherence with international RD initiatives involving registries activities (IRDiRC, RD-Connect, GRDR, etc.)
Objectives of the Platform

1/4 Promotion of the interoperability of existing standalone registries via

- dissemination of agreed Common Data Set expandable by disease

- guidelines for sharing of data

- provision of shared IT tools for data collection and analysis
Objectives of the Platform

2/4 Support for new registries by providing

- **IT tools** (data collection, data transmission)
- **guidelines** (data quality, data protection, ethical issues, etc.)
- **standards** (terminology, outcome measures, etc.)
- **training** (running registries, data quality, data protection, ethical issues, etc.)

1), 2): services provided to the registries, no cost for registries
Objectives of the Platform

3/4 Acting as a hub and providing access to all data collections in the field of rare diseases
- source of information on all existing sources of data collections so as to maximise collaboration and use of data

4/4 Involvement of all stakeholders
health professionals, researchers, patients, industry, policy makers, etc.
Thank You for Your Attention

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