# EUCERD recommendations on Registries and Data Collections for Rare Diseases

**Ségolène Aymé**
**INSERM US14, Paris, France**

**EURORDIS Membership Meeting**
**Dubrovnik, 31 May 2013**



EUCERD Joint Action

# Definitions

- Registry: *"a file of documents containing uniform information about individual persons, collected in a **systematic** and **comprehensive** way, in order to serve a pre-determined scientific, clinical or policy **purpose**"*
  - *Population-based registries*
  - *Non-population based registries*



EUCERD Joint Action

# Types of registries

- **Disease** registry: *data about all individuals with a defined disease for research purpose*
- **Clinical** registry: *data about all individuals with a defined disease attending a clinic, for clinical purpose*
- **Product** registry: *data about all individuals treated with a defined product for research and regulatory purpose*
- **Patient** registry: *data about individuals with a defined disease for recontacting purpose*

# The Commission Communication and the Council Recommendation 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.

# 569 RD Registries as strategic tools

**Number of disease registries per country**

| | |
|---|---|
| France | 135 |
| Germany | 98 |
| United Kingdom | 65 |
| Italy | 51 |
| Spain | 38 |
| The Netherlands | 21 |
| Belgium | 20 |
| Sweden | 18 |
| Austria | 15 |
| Portugal | 15 |
| Ireland | 12 |
| Poland | 9 |
| United States | 8 |
| Switzerland | 7 |
| Bulgaria | 6 |
| Finland | 5 |
| Norway | 5 |
| Czech Republic | 4 |
| Denmark | 4 |
| Serbia | 4 |
| Turkey | 4 |
| Hungary | 3 |

**Orphanet Report Series on Orphanet front page**



EUCERD Joint Action

# Challenges

- Fragmentation
- Lack of interoperability
- Difficult cross-border collaboration due to regulatory requirements
- Lack of sustainability (research funding)

# SANCO Annual work plan 2013

- **4.2.4.4. Support to rare diseases registries and networks in view of their sustainability**
- The aim of this action is to set up a sustainable platform to coordinate and maintain registries and networks on rare diseases. Registries and networks are key instruments in increasing knowledge of rare diseases and in developing clinical research. They are the only way to pool data in order to achieve a sufficient sample size for epidemiological research and/or clinical research. This action will build on activities and experiences developed through initiatives funded by the EU health programmes and research and innovation programmes.
- [Project grant/Administrative agreement with the Joint Research Centre in Ispra, Italy] **EUR 2 000 000**

# EUCERD contributions so far

- « Patient Registries in the field of rare diseases » - 2008,2011
- « Disease registries in Europe » - 2011
- « Health indicators of rare diseases » - 2011
  - « Conceptual framework and development of indicators from existing sources », 2010
  - « Conceptual framework for monitoring quality of care », 2011
- Core Recommendations on rare disease patient registration and data collection - June 2013
- Policy Scenarios for a European Platform - June 2013



EUCERD Joint Action

# EUCERD Core Recommendations on rare disease patient registration and data collection

## TO BE PRESENTED FOR ADOPTION AT THE NEXT EUCERD MEETING ON 5-6 JUNE 2013



EUCERD Joint Action

# Background to the Recommendations

- RDTF: *Patient Registries in the Field of Rare Diseases, Apr 2009, updated Jun 2011*
- EMA/ EUCERD: *Towards a Public-Private Partnership for Registries in the Field of Rare Diseases, Workshop Report, London, 4 Oct 2011*
- EURORDIS/ CORD/ NORD: *Joint Declaration of 10 Key Principles for Rare Disease Patient Registries, Nov 2012*
- EUCERD Joint Action: *Workshop Report on Rare Disease Registration, Luxembourg, 13 Nov 2012, and drafting group and breakout session discussions (29- 30th January 2013)*
- Joint EBE-EuropaBio Task Force on Rare Diseases and Orphan Medicines: *Position Paper for Rare Diseases and Orphan Drugs Registries and Databases*
- EPIRARE Rare Disease Registry survey
- ENCePP E-Register of Studies Guide



EUCERD Joint Action

# Recommendation 1 from the EUCERD

- RD registries and data collections need to be internationally **interoperable** as much as possible and the procedures to collect and exchange data need to be **harmonised** and consistent, to **allow pooling** of data when it is necessary to reach sufficient statistically significant numbers for clinical research and public health purposes



EUCERD Joint Action

# Recommendations 2 & 3 from the EUCERD

- **All sources of data** should be considered as sources of information for RD registries and data collections, to **speed up** the acquisition of **knowledge** and the development of clinical research.

- Collected data should be utilised for **public health** and **research purposes**.



EUCERD Joint Action

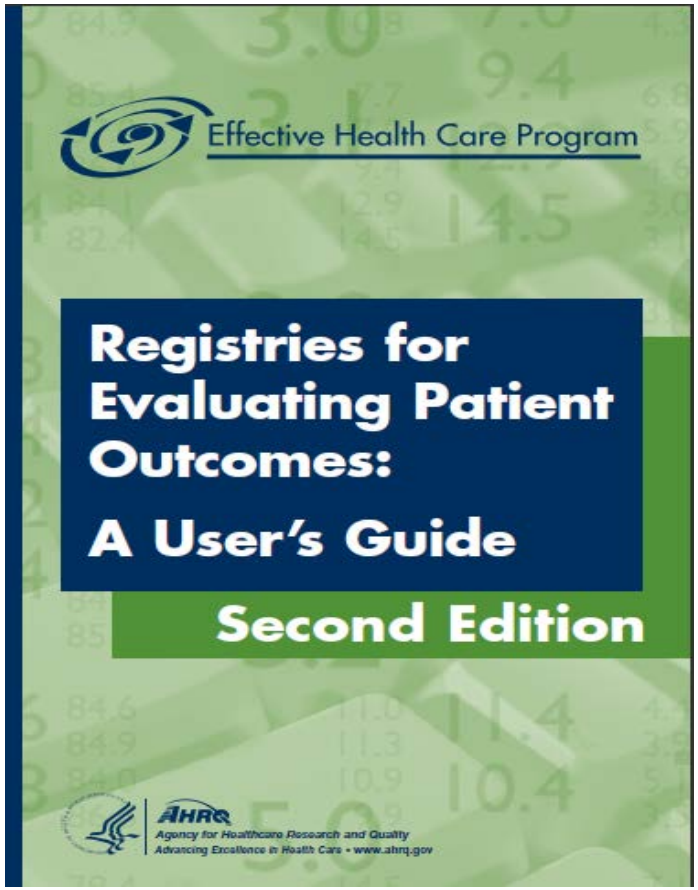
# Recommendation 4 from the EUCERD

- Patient registries and data collections should adhere to good practice guidelines in the field



EUCERD Joint Action

# Everything is in this US report




Gliklich RE, Dreyer NA, eds.

Registries for Evaluating Patient Outcomes: A User's Guide

Prepared by Outcome DEcIDE Center

AHRQ Publ. No. 07-EHC001-1. Rockville, MD: Agency for Healthcare Research and Quality, April 2007

2nd edition, September, 2010

- 55 contributors from industry, academia, health plans, physician societies and gov't
- 49 invited peer reviewers and public comment, including OCR, OHRP, IOM
- 38 case studies from many countries illustrate challenges and solutions

**http://effectivehealthcare.ahrq.gov**



EUCERD Joint Action

# Recommendations 5 & 6 from the EUCERD

- **Existing and future patient registries and data collections should be adaptable to serve regulatory purposes, where required**

- **Patient registries and data collections should be sustainable for the foreseeable timespan of the registries' utility**



EUCERD Joint Action

# Possible Policy Scenarios for the EU Registry Platform

**TO BE PRESENTED FOR DISCUSSION AT THE NEXT EUCERD MEETING ON 5-6 JUNE 2013**



EUCERD Joint Action

# A European Platform
# What for ? How ?



EUCERD
Joint Action

# Act as Incentive to collect and use good quality data(1)

- By minimising costs as much as possible
  - Shared tool (database and web interface)
  - Shared expert team
  - Common communication
- By supporting data producers
  - Training on regulatory obligations/data protection/ ethics
  - Training on standards: terminology, outcome measures, validated scales
  - Advising on how to establish new registry



EUCERD Joint Action

# Act as Incentive to collect and use good quality data(2)

- Act as incentive to collect and use good quality data
  - By improving quality
    - Respect of quality standards
    - Monitoring of data
  - By maximising output
    - Critical mass / incentive to collaborate
    - Standard exploitations / agregated data
    - Access to data by external groups
  - By securing long-term storage / repository of data

# Services to be implemented to make the European Platform a convincing/attractive tool



EUCERD Joint Action

# Service n°1 : Federation of national platforms and national disease registries

- Several countries have started to build a national database of rare diseases
- Many disease specific rgisters exist in several MS
- Start cross talk / work towards a harmonisation of initiatives to build a federation of national registers
- Propose to other countries to build their own national data collection
- Guide and support their efforts
- **Plus:** respects subsidiarity; EU platform design easy
- **Minus:** does not help individual disease registries

# Service n°2 : Platform to support registration

- Main goal: Ease data collection to increase knowledge on as many RD as possible, especially the ones not yet documented
- Instrument: Propose a technological platform to easilybuild disease registers
  - at no cost
  - or very low cost: minimal contribution by type of service
  - Common minimal datasets across RD
  - Common minimal datasets per disease/group of diseases

# Service N°3 : Platform to direct to sources of data

- Main goal: offer a unique entry point to access data on RD
- Document all existing collections (partnership with Orphanet)
- ? Assess quality and  provide an « official designation »
- Document what is collected, procedures and rules for accessing
- **Plus:** useful to data users  and lower cost
- **Minus:** does not help data producers

# Service N°4:Platform of services to registries

- Main goal: Ease the process of establishment of local registries by providing guidance, training, support

- Difficult to implement as requires a lot of experience and legitimacy
- Organise consensus on minimal datasets by disease/group of disease

- **Plus:** Real service to beginners
- **Minus:** does not help so much existing registers

# A European Platform
# What is it not for ?



EUCERD
Joint Action

# What the Platform cannot/will not do

- Will not replace the primary sources
  - Except may be for very rare diseases if there is an option for establishing primary data collection
- Will not very much decrease the cost of data collection and exploitation at primary sources
  - Most of the cost is personnel costs to chase data / patients and quality control, and to raise funding and collaborations
- Will not solve in anyway the problem of the sustainability of the data collection, only of the data storage of a (probably) minimal dataset.



EUCERD Joint Action

# Key factors for success

- Select clear achievable goals with immediate positive impact for a set of stakeholders
- Establish a long term plan with clear steps
- Establish a authoritative team which is crucial for platform's credibility
- Keep a bottom up approach at all stages
  - Definition/priorisation of goals and services by all data producers and data users
  - Definition/priorisation of goals according to utility
  - Monitor uptake of services / satisfaction of end users
- Do not raise too many/much expectation(s) among the primary source registries so as not to disappoint them

# Conclusion

- New opportunities to collect and access data to clinical research and public health purpose
- Providing that the inititatives have a real European added-value
- Necessity to impact on the design of the project and on its governance in the future
- Worry on funding at national level in most MS
- Project of the EC to build a European Research Infrastructure Network for Rare Diseases (ERIC)