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Current initiatives on Rare Disease Patient Registers at international level

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Summary

- **Definition and main objectives of Patient Registers**
- **Overview of the main initiatives in EU and USA**
- **Summary of EUROPLAN recommendations**

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The word

Register: UK English

Registry: USA English

: a written record containing regular entries of items or details

Patient Register main objectives

- Natural history of the disease
- Epidemiological research
- Clinical research (patient recruitment for clinical trials)
- Mutation database
- Genotype-phenotype correlation
- Disease surveillance
- Treatment evaluation (efficacy)
- Treatment monitoring (safety)
- Social Services planning
- Healthcare Services planning
- **…last but not least: instrumental in creating a patients' community**

Definition of “patient register”

- “…*an organized system that uses observational study methods to collect uniform data (clinical and other) to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves one or more predetermined scientific, clinical, or policy purposes*”. (AHRQ)

- “…*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*”. (WHO)

Evolution of the Rare Disease (RD) environment

- Very few researchers and clinicians were interested in careers investigating rare diseases
- Only a handful of companies were interested in investing in orphan drug development
- Limited funding opportunities
- Few patients groups were established.

- More researchers interested as funding has improved dramatically and rare diseases are "untapped" opportunities.
- Significant increase in interest from larger pharma-companies and biotech sector.
- Scientific advances such as gene tests, advanced therapies on the horizon.
- Rare diseases are increasingly recognised as models for common diseases.
- Increased public funding opportunities and public investments in RD infrastructures
- An empowered RD patient community

Growing interest on RD patient registers in EU

- COM (2008) 679 Communication from the Commission to the European Parliament “Rare Diseases: Europe’s challenges”.

- Council Recommendation of 8 June 2009 on an action in the field of rare diseases.

- EUCERD (Rare Disease Task Force)
  - ✓ Workshop on Patient Registries and Databases. March 13, 2008. www.eucerd.eu
  - ✓ EUCERD-EMA Workshop: Towards a public-private partnership for registries in the field of rare diseases. London , October 4th, 2011
  - ✓ EUCERD will elaborate recommendations on Rare Diseases Registers by November 2012

- Recommendations for the Development of National Plans for Rare Diseases – EUROPLAN www.europlanproject.eu

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EU main initiatives in the field of rare disease registers

Orphanet report on RD patient registers

569 Registries as strategic tools

Number of patient registries per country

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

Orphanet Report Series on Orphanet front page

orphanet

www.orpha.net

Challenges

- Scarcity of cases and complexity of diseases impose a large geographical coverage, trans-national
- Resources are limited, funding is limited in time
- Waste of resources
  - in developing tools for each registry
  - in duplicating efforts
- Waste of data: Clinicians are not epidemiologists
- Waste of expertise
- Waste of opportunities
- Increase of distrust among patients
- Demotivation among health professionals
- Drug registries vs disease registries

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The EPIRARE project: European Platform for Rare Disease Registries

- A DG SANCO 30-months project (www.epirare.eu), starting date April 2011
- Coordinated by the Istituto Superiore di Sanità (ISS) (Italian Ministry of Health)
- 10 Associated Partners (among which EURORDIS)

  23+ Collaborating partners
- Total budget of 1.1M € (60% of which from the European Commission)

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The EPIRARE project (I): Building Consensus and Synergies for the EU Registration of Rare Disease Patients

Main Objectives:

- To build consensus and synergies to address the legal, regulatory, ethical and technical issues associated with the registration of RD patients in Europe.
- To explore the definition of a minimum data set common to all RD
- To elaborate possible policy scenarios for EU policies on RD patient registers
- To propose the framework of a future EU register platform (legal basis, governance framework, option for sustainability, ect…).

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The EPIRARE project

- **Policy scenarios on scope, aims, governance and long-term sustainability** Defines the scope and a governance model for registries, **representing the interests of relevant stakeholders** (national and sub national public health authorities, researchers, drug industry, patients' associations and centres collecting data) in strategic decisions. ***WP Leader* - EURORDIS**

- **Common data set and disease-specific data collection**: Defines a common data set, which is independent of the registered rare disease, and **of other disease-specific modules**, in aims. order to provide information consistent with the agreed scope of the registries and useful for public health actions and for other.

- **Data quality, validation and data sources integration**: definition of the criteria for quality assessment of data, data sources and procedures in the registries.

- **Current needs of existing registries on rare diseases: a survey** will verify the current state of RD registries to identify their expectations and needs and will define the services and contents which help in performing activities of interest of the platform users.

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DG SANCO 2012 CALL FOR PROPOSALS

2012 CALL FOR PROPOSALS FOR PROJECTS

SECOND PROGRAMME OF COMMUNITY ACTION IN THE FIELD OF HEALTH (call closed March 10th 2012; proposals under review)

3.2.4.3. Support for European rare diseases information networks

The objective of this action is **to support the setting up of new rare disease registers or rare disease information networks**.

- This action will also contribute to **the reflection on criteria for designating ERN**
- Priority areas for this action are**: rare tumours, rare anaemias, cerebral palsies, neuromuscular diseases, cystic fibrosis, rare neurological disorders and rare syndromes associated with autism** (other diseases were also considered).
- This action seeks to co-finance at least five networks

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EU Joint Action: PARENT project

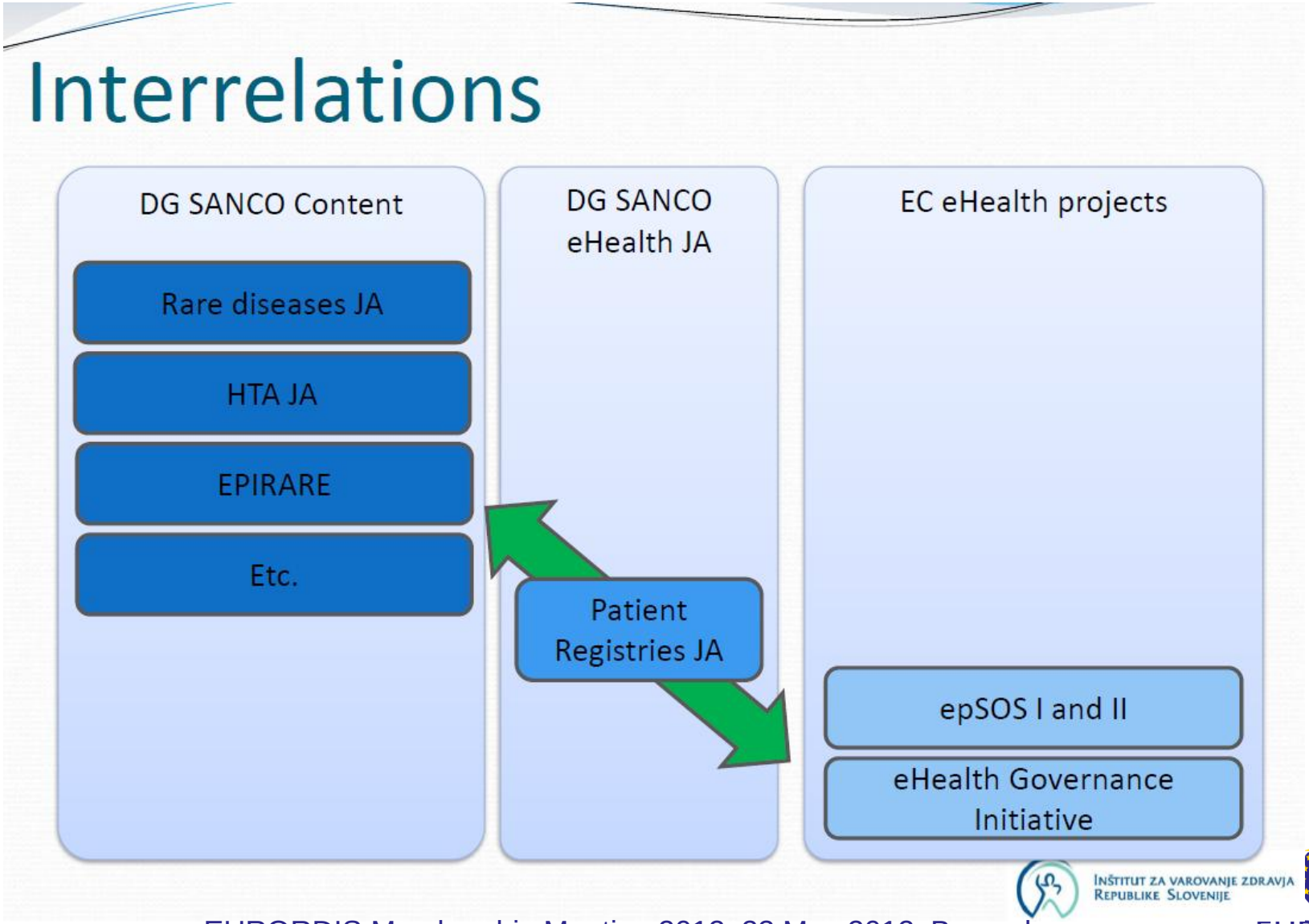
- **PARENT:** cross border PAtient REgistries iNiTiative

3.3.1.4. Cross-border e-Health instruments as supporting tools for medical information and research.

- Timing: 1th Sept 2012 – 28th Feb 2015, total duration: 30 months
- Budget: 3.4 Mio € (11 Associated partners , 8 Collaborating partners)

- ***Aim***: to rationalise and harmonise the **development and governance** of patient registries, thus enabling analyses of secondary data for public health and research purposes.
- ***Goal*:** support MS in developing **comparable and coherent patient registries** in fields where this need has been identified (e.g. chronic diseases, **rare diseases,** medical technology)
- ***Goal***: support MS states in the provision of **objective, reliable, timely, transparent, comparable and transferable information** on the relative efficacy and effectiveness of health technologies.

Coordination with other EU initiatives



INŠTITUT ZA VAROVANJE ZDRAVJA REPUBLIKE SLOVENIJE

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RD-CONNECT: An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research

DG-Research, 6 years duration, 12 M Euros, 25 partners.

The RD-Connect platform should provide:

- large-scale data management
- access to federated databases/patient registries
- biobank catalogues
- harmonised -omics profiles
- cutting-edge bioinformatics tools for data analysis.

All **patient data types** will be linked via the generation of a **unique identifier** ("RD-ID") developed jointly with the US NIH.

The RD-Connect platform will be one of the **primary enablers** of progress in **IRDiRC-funded research** and will facilitate **gene discovery, diagnosis and therapy development.**

USA main initiatives in the field of rare disease registers

NIH-Office of Rare Diseases Research (ORDR)

Global Rare Diseases Patient Registry and Data Repository (GRDR)

- **NIH-ORDR** initiative in collaboration with **a private service provider, Children Hospital of Philadelphia** and **WebMD**

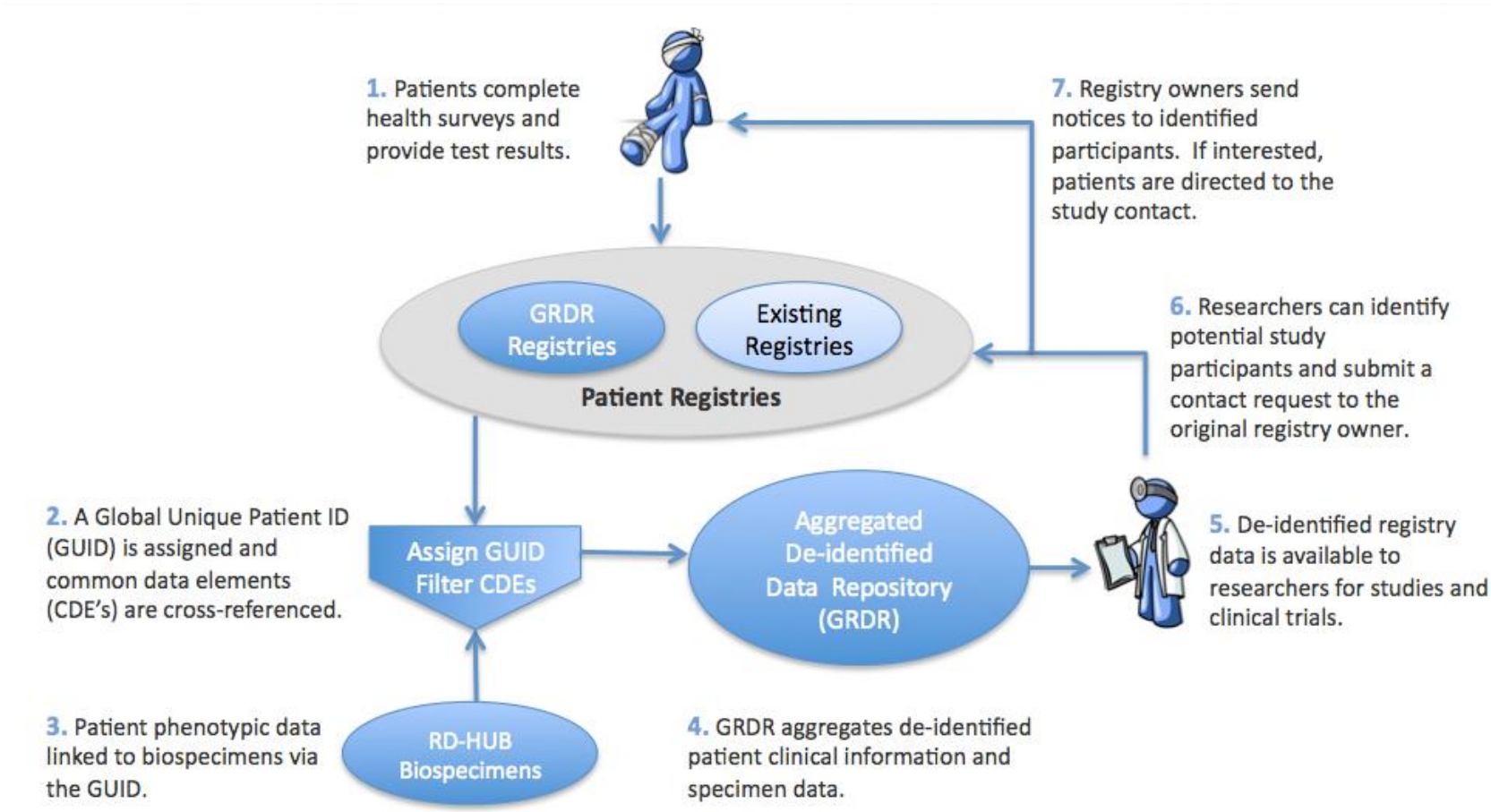
- **Pilot program** to establish a **Global Rare Diseases Patient Registry and Data Repository (GRDR)** to collect de-identified patient data for clinical research.

- **12 patient groups, each without a registry**, will be selected to assist in testing the implementation of the ORDR Common Data Elements (CDEs) in the newly developed registry infrastructure**.**

- **12 established patient registries** will be selected to integrate their de-identified data into the GRDR to evaluate the data mapping and data import/export processes.

Overview of the Global Rare Disease Registry and Data Repository

http://www.grdr.info/



Centers for Disease Control and Prevention (CDC)

Call: A Pilot Surveillance System for High Impact/Low Prevalence Congenital and Inherited Conditions

- Call for **cooperative agreement applications** to design and test the feasibility of a **surveillance system** for congenital and inherited conditions that have **low prevalence, but high health and economic impact** in the U.S. Population.
- **Goal**: to describe **the burden of disease and disability** from these conditions and **inform and evaluate policies and interventions** aimed at improving health services and health outcomes among people affected by these conditions and their families. Conditions selected: spina bifida, muscular dystrophy, and fragile X syndrome.

Agency for Healthcare Research and Quality AHRQ

Registry of Patient Registries (RoPR)

In September 2010 AHRQ awarded Outcome, a Quintiles Company, a task order to design and develop a Registry of Patient Registries (RoPR) database

Main Goals:

- To provids a **searchable database** of existing patient registries in the United States
- To facilitate the use of **common data fields and definitions** in similar health conditions (to improve opportunities for sharing, comparing, and linkage)
- To provide **a public repository of searchable summary results** (including results from registries that have not yet been published in the peer-reviewed literature)
- To offer **a search tool to locate existing data** that researchers can request for use in new studies
- To serve as a **recruitment tool for researchers and patients** interested in participating in clinical trials

Registry of Patient Registries (RoPR)

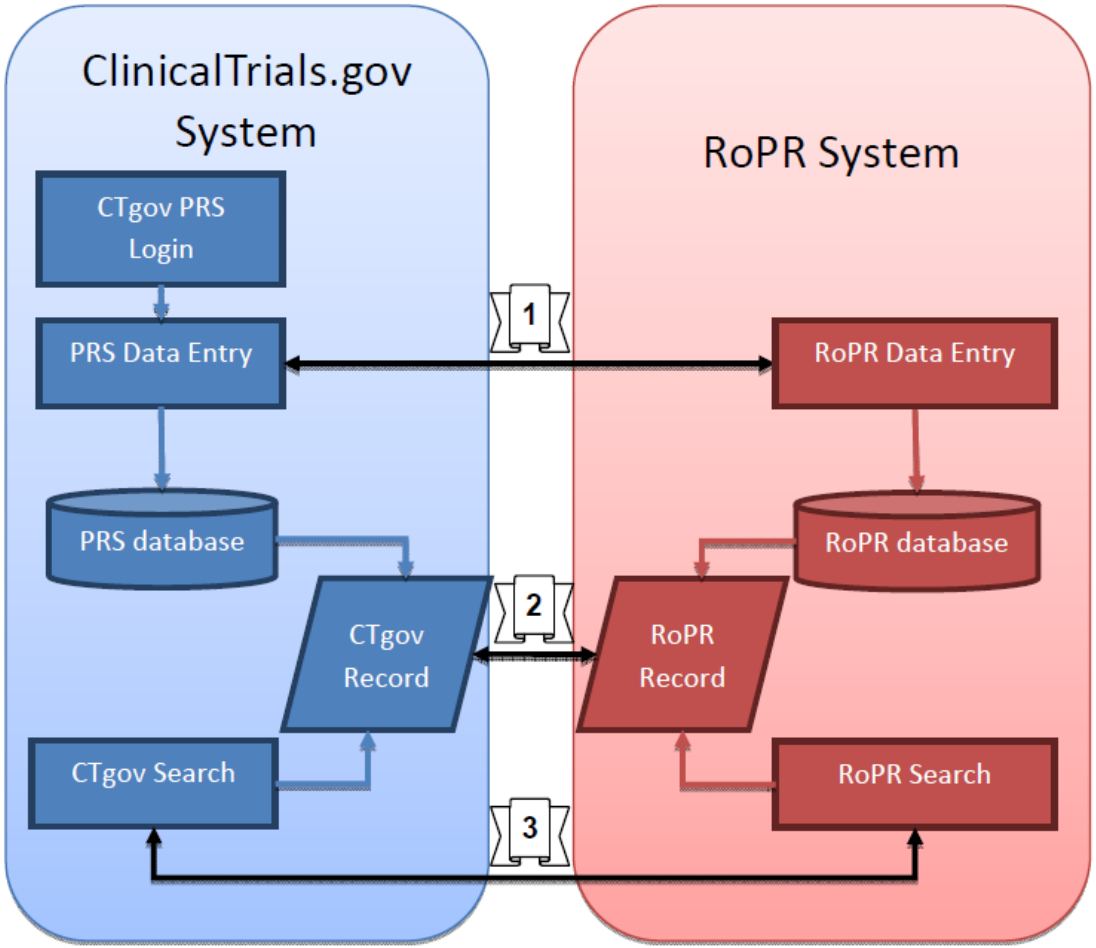


Figure 1 RoPR Integration with ClinicalTrials.gov

Summary of the EUROPLAN recommendations on RD Registers

EUROPLAN reccomandations on RD Registers (I)

- Disease-specific registries or registries for groups of rare diseases are an effective way to assess health care needs as well as to generate research in several areas, including epidemiology

- Often, they are the only existing source of scientific/clinical and epidemiologic information on rare diseases.

- They were considered to be a primary objective and a basic requirement to develop RD research.

- Appropriate measures to ensure the sustainability of registries, the quality of their data and the involvement of Centres of Expertise should be identified and put in place.

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EUROPLAN reccomandations on RD Registers (II)

- Initiatives at EU and international level would be most appropriate to ensure the widest population basis for the registration of rare disease cases.

- Synergies among different interests, e.g. research, public health information needs, orphan drugs effectiveness assessment, health care planning and health costs management, should be sought and registration activities should be promoted with the establishment of a platform providing common tools and services.

- The reasoned definition of common tools and services as well as of a common core set of variables to be collected, depending on the scope of the registries, may be of help to national policy makers to include appropriate provisions in their national plans/strategies to promote comparability and exchange of information among registries and Countries.

Summary of the EUROPLAN reccomandations on RD Registers (III)

- Initiatives are promoted at national level for the integrated use of administrative, demographic and health care data sources to improve the management of rare diseases

- International, national and regional registries for specific rare diseases or groups of rare diseases are promoted and supported for research and public health purposes, including those held by academic researchers.

- Collection and sharing of data from any valid sources, including Centres of Expertise, and their availability for public health purposes is promoted by public health authorities, in compliance with national laws.

- Participation of existing national registries in European/International registries is fostered.

- Instruments are identified for combining EU and national funding for registries.

The « Ten Golden Rules » I

1) Rare Disease Patient Registries are a major priority in the field.

2) Rare Disease Patient Registries should be global in geographic scope whenever possible.

3) Rare Disease Patient Registries should be defined around a disease, not solely a therapeutic intervention.

4) A Minimum Common Data Set should be consistently used in all Rare Disease Patient Registries.

5) Interoperability and harmonization between Rare Disease Patient Registries should be consistently pursued.

6) Rare Disease Patient Registries should be linked with biological specimens in biobanks.

The « Ten Golden Rules » II

7) Public-private partnerships should be encouraged to help ensure sustainability.

8) Data introduced into Rare Disease Patient Registries should be directly reported by Health Care Professionals as well as by patients to improve the completeness and robustness of data collection.

9) Patients and their representatives should be equally involved with other stakeholders in the governance of Rare Disease Patient Registries.

10) Rare Disease Patient Registries may serve to build the capacity and empower rare disease patients and their representatives in the development, governance, maintenance and use.

Wish for the future

…hopefully efforts will be successful,

for the benefit of RD patients AND with their active participation!

Thank you!