



EPIRARE Common Data Elements

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EPIRARE

“Building Consensus and Synergies for the EU
Registration of Rare Disease Patients”

Specific Objectives

- To identify the **needs** of the EU registries and databases on RD
- To identify **key issues to prepare a legal basis**
- To agree on the Register and Platform **Scope, Governance and long-term sustainability**
- To agree on a **Common data set, disease-specific data collection and data validation**
- To identify **tools and other facilities** supporting the operation of the platform users

The evidence base

Ad hoc funding, momentum and commitment (EPIRARE, CNMR)

- Survey of registries conditions and needs
- Survey of collected variables, definitions and formats
- Survey of patients expectations
- Consultation/survey of industry expectations
- Consultation/Information of national policy-makers
- Personal consultation with “payer” associations
- Review of policy actions and funding initiatives
- Liaison with US ORDR and RD-Connect (IRDiRC)
- Participation in EU initiatives and RD Committees

Main issues found in registries

Data quality control procedures
Coding and reference catalogues
Governance
Communication
Networking and data sharing

Needs of identified stakeholders' groups

- Registry holders
- Patients
- National HA and Payers
- Pharmaceutical Industry
- EMA and EC

=> *EPIRARE Deliverable 5*

Scope of the indicators and variables considered

- **Epidemiological information**
- **Information for RD policy planning and monitoring;**
- **Monitoring RD-dedicated health services and integration in the NHS;**
- **Health technology assessment (appropriateness of OD and other treatments)**
- **OD and other treatment**
- **Patient recruitment**
- **Patient care benchmarking**

The organization of the EPIRARE Database

DATA DOMAINS

1) Case characterization essentials

Minimum Set CDE:

EU GUID

(necessary elements:
Name, surname, date of birth, city of birth, country of birth, unique identifier)

- Consent
- Diagnosis
- City (and country) of residence
- Treatment centre ID (& city & country)
- Patient willingness x CT and donations

mandatory

2) Determinants and health service data

Genetic variants

Living conditions

Drugs and treatments

Health services and procedures

Costs

3) Outcomes

Age at death

Link to clinical data sources

Clinical data

DOMAIN SCOPES

Case notification completeness

Risk factors, health service monitoring

Natural history, HTA, patient recruitment

Selection of variables consistent with the intended aims and indicators

Domain 1) Case characterization essentials

- EU Global Unique Identifier (EU GUID): *name, surname, sex, date and place of birth, national ID*
- Patient sex
- Patient date of birth
- Patient place (+Country) of birth
- Diagnosis
- Patient place (+Country) of residence**
- ID Treatment Centre (+place+Country)**
- Current and past participation in clinical trials**
- Patient willingness to be contacted to participate in a clinical trial**
- Patient willingness to be contacted about donating biomaterial**
- Patient consent**
- Patient contact preferences**

Domain 2) Determinants and services

A sample selection of data

- other cases in the family
- healthy carriers in the family
- case parents are consanguineous
- Genetic features of the patient

- **Current orphan drug treatment**
- **Hospitalizations (number)**
- **Transplantations (date and biomaterial transplanted)**

Domain 3) Outcomes

- **Patient vital status (and date of death)**
- **Education level**
- **Occupational status**
- **Patient HRQoL index score**
- **Comorbidity**
- **Remarkable or unusual symptoms**

Recommended readings

Gliklich RE, Leavy MB, Levy D, Karl J, Champion DM, Taylor T. Registry of Patient Registries (RoPR) Policies and Procedures. Effective Health Care Program Research Report No. 41. 3rd edition (2014).
effectivehealthcare.ahrq.gov/reports/final.cfm.

Manuel Posada de la Paz· Stephen C. Groft (Eds.)
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EPIRARE deliverables (www.epirare.eu)



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Thank you for your attention!

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