The European Commission’s science and knowledge service
Joint Research Centre

European Platform on Rare Diseases Registration

Simona Martin
Agnieszka Kinsner-Ovaskainen
Monica Lanzoni
Andri Papadopoulou

Directorate F, Unit F.1

3rd European Reference Networks Conference
Vilnius, 9 March 2017
Development and Maintenance of the European Platform on Rare Diseases Registration
Development and Maintenance of the European Platform on Rare Diseases Registration

- limited number of patients
- scattered knowledge and expertise (diagnosis, treatment)
- fragmentation of data sources (>
600 registries)

High European added value
EU Platform on Rare Diseases Registration

Knowledge generation centre / unified source of information for RD

Providers of information

Users of information
EU Platform on Rare Diseases Registration

Knowledge generation centre / unified source of information for RD

- Registries
  - national
  - regional
  - local
  - hospital
  - patients'...

- European surveillance networks

Users of information
EU Platform on Rare Diseases Registration

Knowledge generation centre / unified source of information for RD

- Registries
  - national
  - regional
  - local
  - hospital
  - patients'
  ...

- European surveillance networks

- Healthcare providers

- Patients

- Researchers

- Industry

- Policy makers
EU Platform on RD Registration – Functions:

INTEROPERABILITY

SUSTAINABILITY
EU Platform on RD Registration - Functions:

INTEROPERABILITY – DATA SHARING

Inventory of RD registries
EU Platform on RD Registration - Functions:

INTEROPERABILITY – DATA SHARING

Inventory of RD registries

Central Metadata Repository
EU Platform on RD Registration- Functions:

INTEROPERABILITY – DATA SHARING

Inventory of RD registries

Central Metadata Repository

Web hub: access to RD data collections
EU Platform on RD Registration - Functions:

INTEROPERABILITY – DATA SHARING

Inventory of RD registries

Web hub: access to RD data collections

Central Metadata Repository

Standards

Tools
**EU Platform on RD Registration - Functions:**

**INTEROPERABILITY – DATA SHARING**

- **Inventory of RD registries**
- **Central Metadata Repository**
- **Web hub:** access to RD data collections
- **ERNs**
  - Standards
  - Tools
    - existing registries/ERNs
    - new registries/ERNs
2nd INTEROPERABILITY WORKSHOP
Luxembourg, 30 November 2017

Participants:
- ERNs Board of MS
- ERNs coordinators
- RD experts
- EU RD-related projects
- RD-Action
- SANTE C1, B3, B4, A4, JRC F1
- Registration of patient data for care / treatment purposes
- Virtual consultations
- Creation of new registries
- Interoperability of existing registries

- Interoperability for the 600+ European registries including ERN registries
- Support for creation of new registries including ERN registries
- Standards and tools for RD registries including ERN registries
FIRST INTEROPERABILITY WORKSHOP
7 – 8 April 2016, Luxembourg

Mapping interoperability needs of stakeholders for data collection and data sharing

- Registries
- National authorities
- Patients
- Regulators
Mapping interoperability needs of stakeholders

**Compatibility of procedures** for improving use of RD registry data / information exchange

1. Standards
   - Common data elements
   - Terminology
   - Data quality
   - Guidelines for creation of new registries

2. Linkage of RD registries with biobank data

3. Specific advice on legal, privacy aspects, etc.
First Interoperability Workshop

**Identification of:** ➢ the **specific outputs** of registry-related projects, appropriate to be incorporated in the Platform as interoperability tools.

**Registry-related projects**

- RD-JA
- Orphanet
- EPIRARE
- PARENT
- OSSE
- EUCERD-JA
- RD-CONNECT
- IRDiRC
- EXPAND

Recommendations of EC Expert Group RD
EU Platform on Rare Diseases Registration

INTEROPERABILITY

SUSTAINABILITY

Data Repository

JRC-EUROCAT Central Registry

JRC-SCPE Central Registry

Central Database

Central Database
EUROCAT Registries:

- **Active** 45 (25 countries)
- **Total** 69 (28 countries)

- Full: 33
- Associate: 6
- Affiliate: 6
- Past: 14
- Applicant: 10
SCPE Registries:

- Active: 24 (20 countries)
- Total: 33 (24 countries)

- Active: 24
- Non-active: 7
- Applicant: 2
Transfer to the JRC

Meetings JRC-EUROCAT Management Committee
JRC-SCPE Management Committee


JRC-EUROCAT Collaboration Agreement
JRC-SCPE Collaboration Agreement

SIGNATURE

Collaboration Agreement entered into force

Annual Registry Leader’s Meeting

Annual Plenary meeting
Data Transfer

2014

Declaration from registries

2015

Data transfer from individual registries

Rebuilding of Central Database

Data submission (regular)

Agreement

JRC-registries

IT Support team

Disclaimer: The contents of this presentation are the views of the author and do not necessarily represent an official position of the European Commission
o collection of data from the registries
o data management including data checking, standardisation, quality assessment, validation, statistical analysis for monitoring of clusters and trends
o documentation and implementation of ensure data security procedures
o provision of output of analysis for further dissemination
o communication with the registries and feedback on data-related issues and results of the monitoring
Transfer to the JRC

Meetings

JRC-EUROCAT Collaboration Agreement
JRC-SCPE Collaboration Agreement

SIGNATURE

JRC Legal advice Unit
IP Rights Office
Procurement Service

Data Protection
Information Security
Web Registration
Data acquisition at JRC-EUROCAT central registry level

- Data reception
  - JRC secured data portal
- Data Check
- Data Import
  - ECD software
- Registry file
  - @Central Registry
- Validation Verification
- Feed-back Reporting
- EUROCAT Central Database
  - ECD software
- Data Analysis
- Data Release
Data Security: the JRC-EUROCAT Information System

External to JRC
- Membership request from non-EUROCAT member Registry
- Data request for research purpose

Internal to JRC
- EUROCAT Central Registry (CR)
  - Managing and evaluating of application form
  - Managing and evaluation of data request form
  - Evaluation of the quality of the data sent by the applicant Registry
  - Selection of data from CR and research follow up the applicant Registry

- JRC-EUROCAT Central Database (CD)
  - Data validation with ECD software developed and maintained by Biomedical
  - Data analysis with ECD software developed and maintained by Biomedical

- EUROCAT WEBSITE [Hosted by Biomedical]
  - Dissemination of the results of analysis performed at the CR/communication with Registries

Data transmission via JRC-WEB portal with password
Data Protection: procedure for processing data at the JRC-EUROCAT Central Registry

Notification to the Commission’s Data Protection Officer of data processing on JRC EUROCAT Central Database.
- DP: Data Protection
The DP notification validation is managed online with DPO-2 System by
- Data Controller: PHPS HOU
- DPC: DP Coordinator (JRC-Ispra)
- DPO: Commission’s DP Officer
- EDPS: European DP Supervisor
Register of the DPO: http://ec.europa.eu/dataprotectionofficer/dpo_register_en.htm

Draft Notification submitted by Data Controller

Notification validation approved by Data Protection Coordinator

Notification validation approved by Data Protection Officer

Prior-check approved by European Data Protection Supervisor

Notification accepted by Data Protection Officer

Publication on the Register of the DPO
Data Request: procedure for obtaining data from the JRC-EUROCAT Central Registry

1. Lead Investigator
   Sends a study outline for initial assessment, and then the data request form and study protocol for detailed assessment

2. Management Committee
   Approves with specifics
   ➔ Data Request Form

3. Requested registries
   Give permission
   ➔ Registry Participation Form

4. Lead Investigator
   Agrees and signs
   ➔ Data Request Declaration

5. Management Committee
   Approves
   ➔ Data Release Approval
Website registration

**Roles** in the recording project:
- Site Owner (SO)
- Web Master (WM)
- Head of Unit (HOU)

- The Global Controllers:
  - Communication Unit/DG (COM)
  - Communication Correspondent (COCOM)
  - Local Informatics Security Officer (LISO)
  - Intellectual Property Rights Unit (IPR)
  - Data Protection Coordinator (DPC)
  - Information and Communication Technologies Unit (ICT)

**Steps** of the recording project:
- Definition and Development
- Approval and Validation
- Allowance and Compliance
- Activation and Production
- Revalidation and Deactivation

1. The new site is **relevant** (COM)

2. URL **not conflicting** with other JRC activities (COCOM)

3. Website complies with **JRC communication objectives** (COCOM)

4. Website complies with **Information Providers Guide** (COM) → Visual identity

5. Website complies with JRC **IT Security policy** (LISO) → Security Plan

6. Website complies with **Copyright rules** (IPR) → IP rights

7. Website complies with **Data Protection rules** (DPC) → DP Notification

8. All required technical parameters are known, Website is activated (ICT)

→ Website recorded on JRC Website Register
JRC-EUROCAT Central DATABASE evolution

Rebuilding Central Database

1st Data Collection

2nd Data Collection

3rd Data Collection

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<th>Cases</th>
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<tr>
<th>Date</th>
<th>Regs.</th>
<th>Cases</th>
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</tbody>
</table>

Var% cases: 10%
JRC -.europcat. Central Registry

European surveillance of congenital anomalies

Standardised procedures for data processing

Data release for European studies
Interoperability between registries

Level 6: requires that **conceptual models are documented** based on engineering methods enabling their interpretation and evaluation by other engineers.

Level 5: the system operates on data over time, state of that system will change, this affects data interchange. **Systems able to comprehend the changes occurred over time**.

Level 4: interoperating systems are aware of the methods used by each system. **Use of the data/context of its application is understood by the participating systems**.

Level 3: meaning of data is shared; **content of the information exchanged unambiguously defined**.

Level 2: common structure to exchange information; i.e., **common data format**.

Level 1: **communication protocol** between participating systems.

Level 0: stand-alone systems. No Interoperability.


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EUROCAT network – internal interoperability (1)

**Level 1: communication protocol**  
(Technical interoperability)

**Timing (twice a year) and rules for data transmission** (JRC-EUROCAT data portal) are defined.

**Level 2: common data format**  
(Syntactic interoperability)

.csv file with predefined named fields (variables)

For the formal correctness of data, two softwares were developed:

1) **EDMP**: software for management of data at registry level. Contains a routine to export data in predefined format; must be used also if the registry collects and manages the data in a different IT system, in order to assure that the data can be imported in

2) **ECD**: software for the management of the data at the Central Registry.

Detection of syntactic errors (data checks) are implemented in both softwares.

**Level 3: content of information exchange unambiguously defined**  
(Semantic interoperability)

Definition of variables, Codification systems; Instructions in EUROCAT Guide 1.4. International references codification systems are used for malformations/drugs/OMIM/Karyotypes.
EUROCAT network – internal interoperability (2)

Level 4: Use of data/context of its application is understood by the participating systems. (Pragmatic interoperability)

Different types of aggregation/analysis performed at the Central Registry defined/described/interpreted in EUROCAT Guide 1.4.; agreed use of data implemented in ECD/EDMP; results shared with and approved by registries before dissemination on the network's website.

Level 5: systems comprehend changes occurred over time. (Dynamic interoperability)

Changes in the data stored in the Central database (ECD) are traced. Modification in codification systems are implemented in ECD/EDMP.

Level 6: conceptual models are documented.

All the procedures of the network, algorithms and analysis applied, data request/sharing rules with third parties are defined in controlled documentation.
EUROCAT network – external interoperability

- data collected by the network is **codified** in an unambiguous way; when possible, the single variables are codified using validated international classification systems.

- data sent to the Central Registry is **pseudo-anonymized** using an **unique ID** for every case defined at local level (registries).

- possibility to **link data** with other databases (EUROCAT-derived projects); linked to spatial environmental databases through the place of residence of the case.
Thank You for Your Attention

www.jrc.ec.europa.eu