



The European Commission's science and knowledge service

Joint Research Centre



**Directorate-General
Joint Research Centre**

**Directorate-General
Health and Food Safety**

European Platform on Rare Diseases Registration

Knowledge generation centre for rare diseases

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Why an EU Platform on RD Registration?

➤ **To cope with the extreme fragmentation of data sources across EU Member States**

Many RD registries exist, but - the lack of interoperability severely limits the registries' potential
- no standardised data collection for most RDs



➤ **Benefits:**

Reach the critical number of patients for

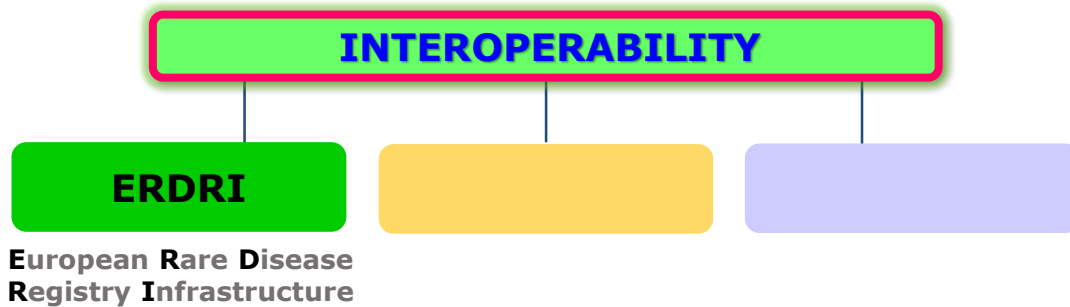
- studies (epidemiological, clinical, translational, pharmacological, etc.)
- research



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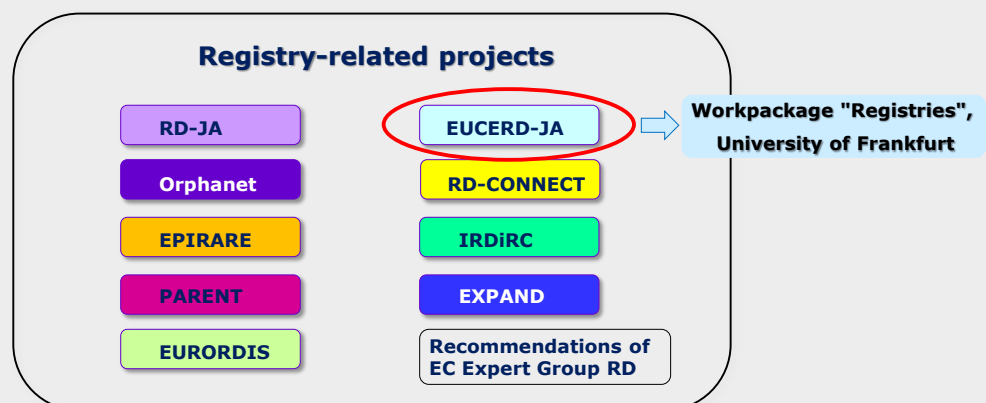


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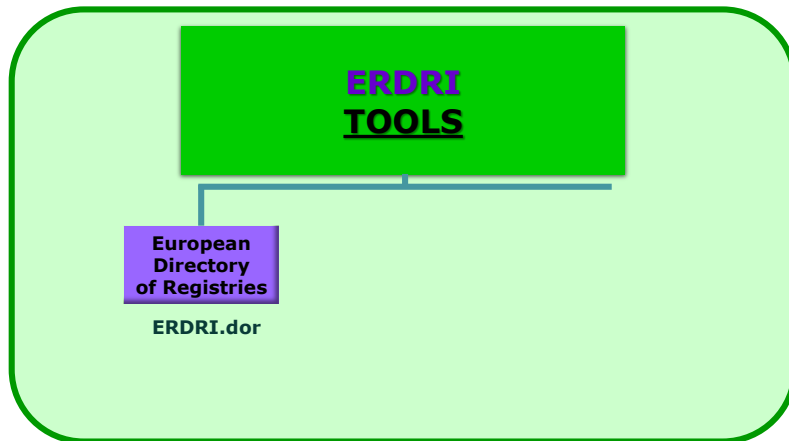
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FIRST INTEROPERABILITY WORKSHOP



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INTEROPERABILITY



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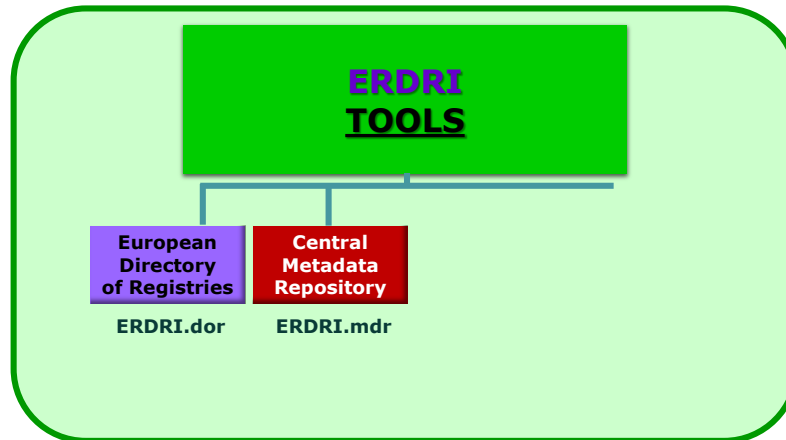
ERDRI.dor – The European Directory of RD registries

- List of participating RD registries
- Descriptive information
 - specific rare disease addressed
 - scope
 - operating institution
 - contact information, etc.)
 -
 - = characteristics of registries (27)

**CORE COMPONENT OF
ERDRI / EU RD PLATFORM**



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European RD Registry Infrastructure (ERDRI)

ERDRI.mdr – The Central Metadata Repository

Semantic interoperability

Central Metadata Repository ERDRI.mdr



=

collection of metadata on all data elements collected by participating registries

- Designation
- Definition
- Measurement unit + range

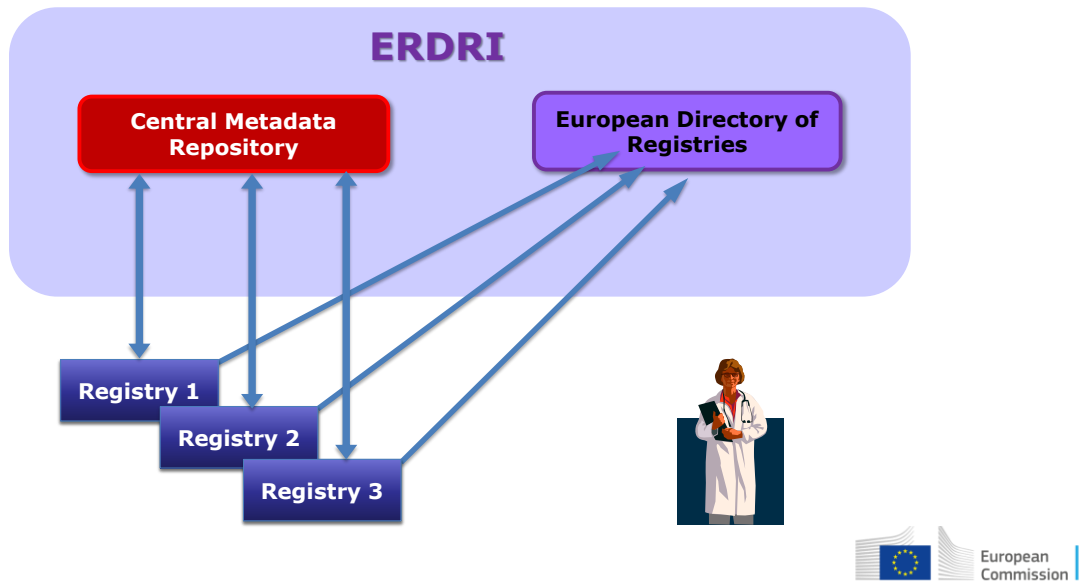
Consequence: use of common definitions for data elements

➔ terms are understood in the same way by all data providers + data users

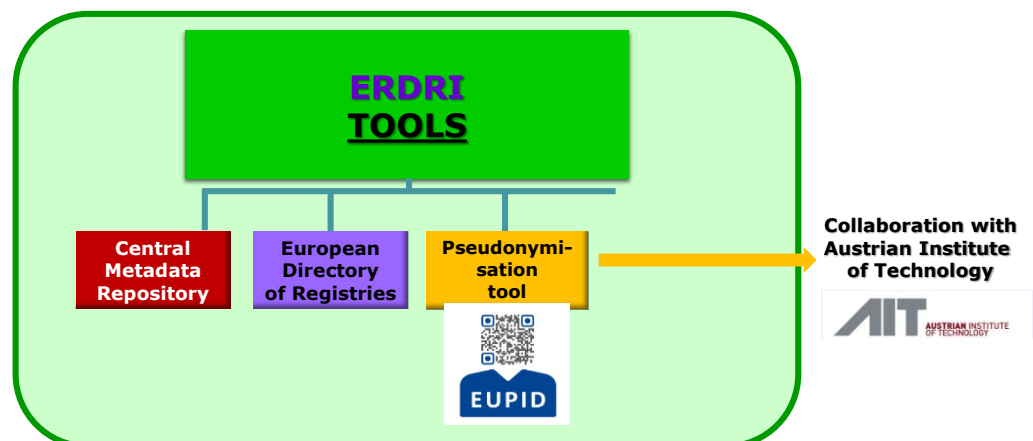
ERDRI.mdr = **thesaurus** of terms used by participating registries
common format



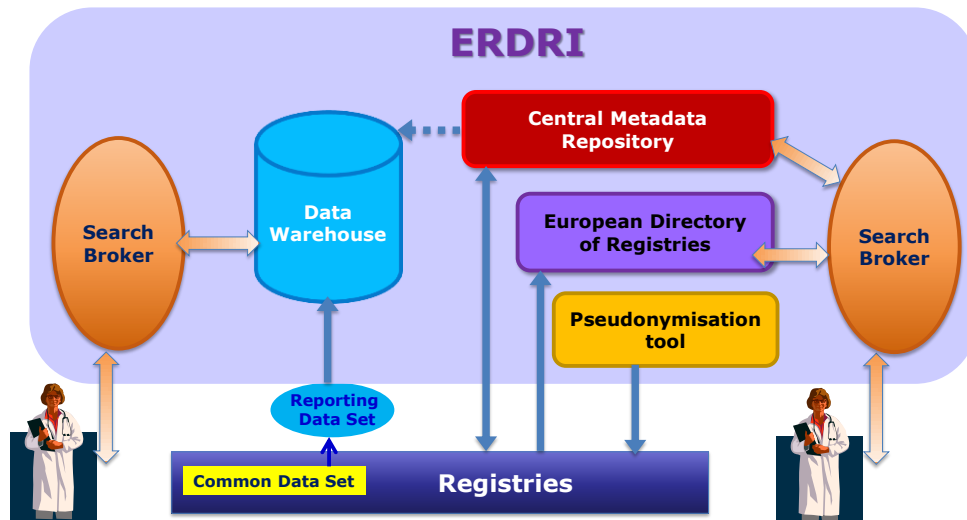
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ERDRI – European Rare Disease Registry Infrastructure

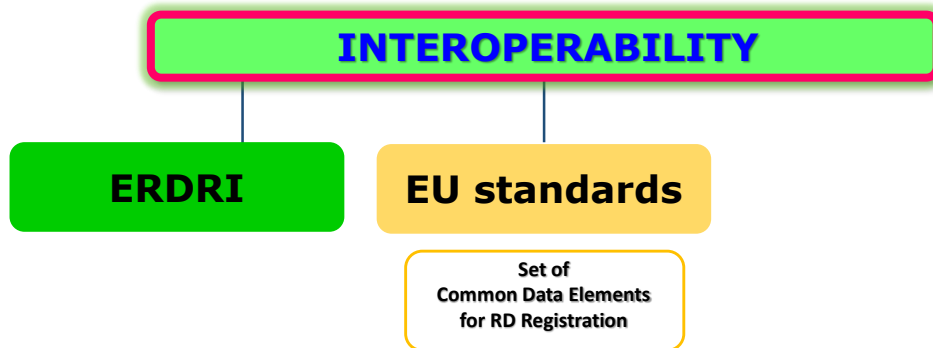
➤ PROVIDES the infrastructure and tools

❖ To make registries' data
SEARCHABLE
FINDABLE
interoperable
reusable

❖ If accessible – it depends on the data owner!



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- ❑ JRC ERDRI Training Workshop - MDR, DoR
 - session 1 – February 2018
 - session 2 – March 2018
- ❑ JRC ERDRI Training Workshop – pseudonymisation
 - March 2018



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