Collaborative research for rare diseases in the EU : the role of Orphanet in its implementation

Ana Rath
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CONFERENCE - Rare diseases and the European reference networks: how to guarantee European solidarity for patients?
Bilbao, 11 October 2023

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Co-funded by the Health Programme of the European Union

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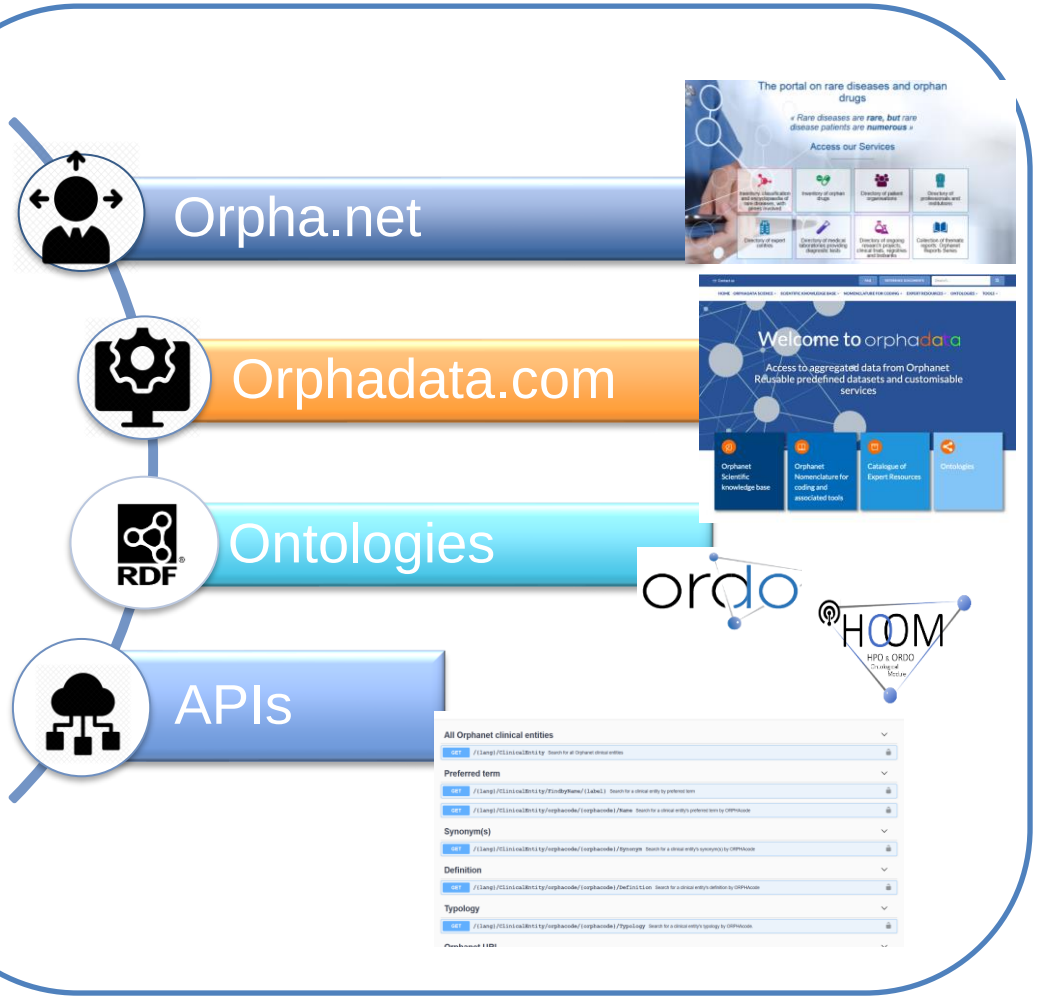
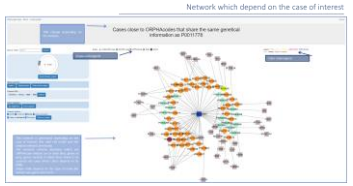
KNOW THE RARE FOR BETTER CARE

Orphanet data are used to provide services

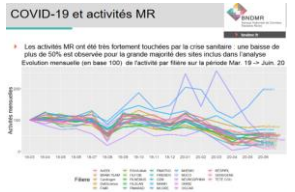


- Access to datasets

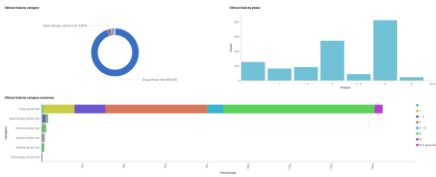
- Applications (diagnosis, …)



- Studies (health economics, epidemiology…)



- Data analysis tools



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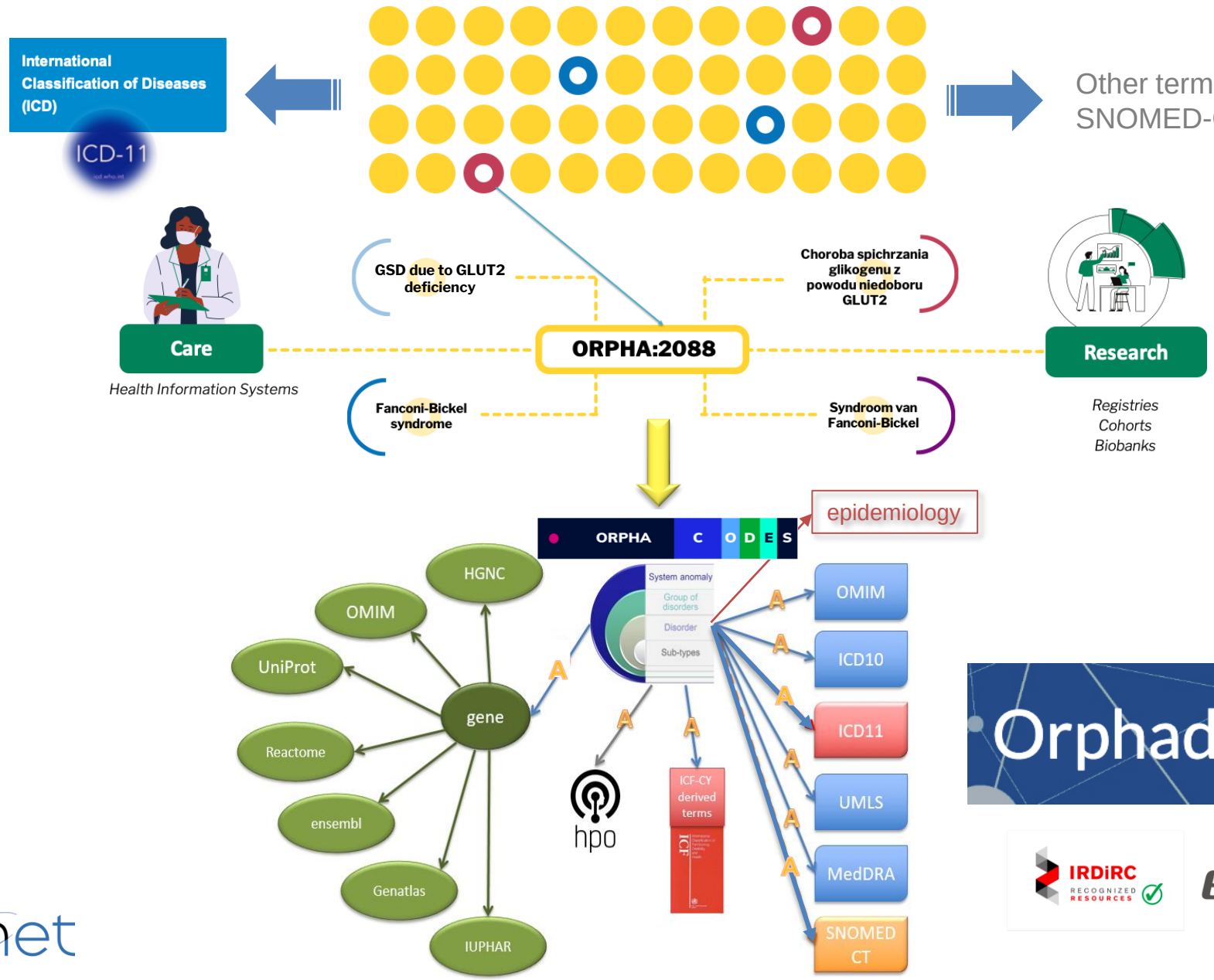
Orphadata Science: building interoperability for RD



Orphadata Science

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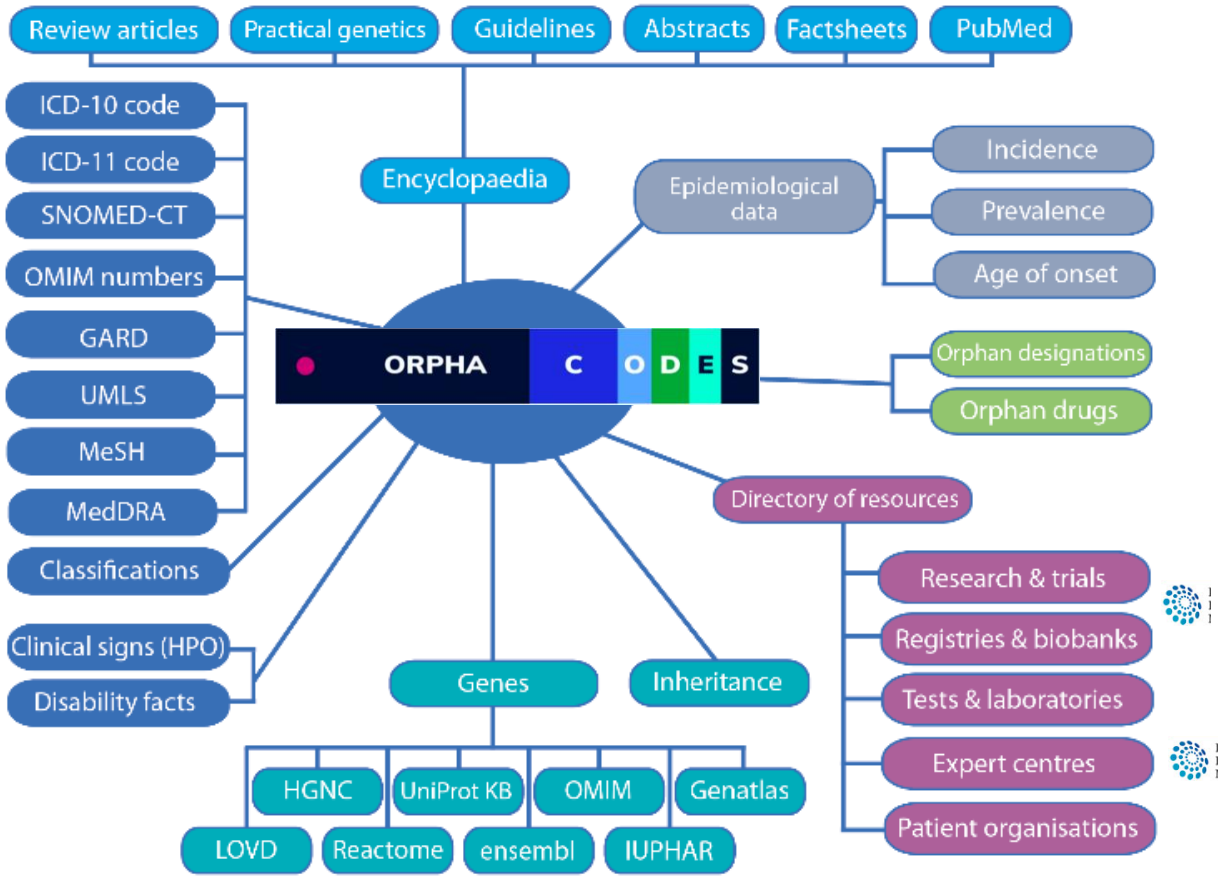
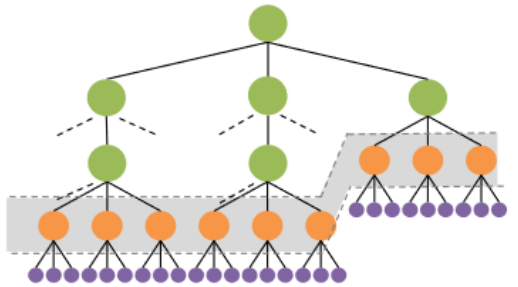
IRDiRC RECOGNIZED RESOURCES

elixir Core Data Resource

GBC

Systematic, standardized data collection allows for data exploitation





Group
Disorder
Subtype

"Classification level"

ENG FR SP DE IT NL PG PL RU CHN TUR

Comprehensive, standardized, evidence-based, interoperable, versioned, computable and free (CC-BY 4.0)

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Orphanet in the RD research ecosystem

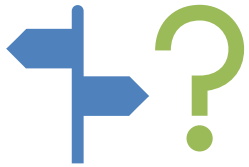
- To give visibility to RD (and ERN) research
  - Structured catalogue of projects, trials, registries, biobanks, infrastrcutures, orphan designations and drugs …
  - Orphanet Report Series
  - ERN clinical research platform
- Provide computable reference data
  - Orphadata: data and tools
- Tools for researchers, data sources and funders, within EU funded projects
  - Phenotypic-based variant prioritisation
  - PCOMs repository
  - Funded research analysis platform
  - Semantic mapping services

RD research ecosystem:
From a heterogeneous, non-interoperable, scattered resources landscape …

Registries/ biobanks catalogues

Cell lines Animal models

Knowledge bases

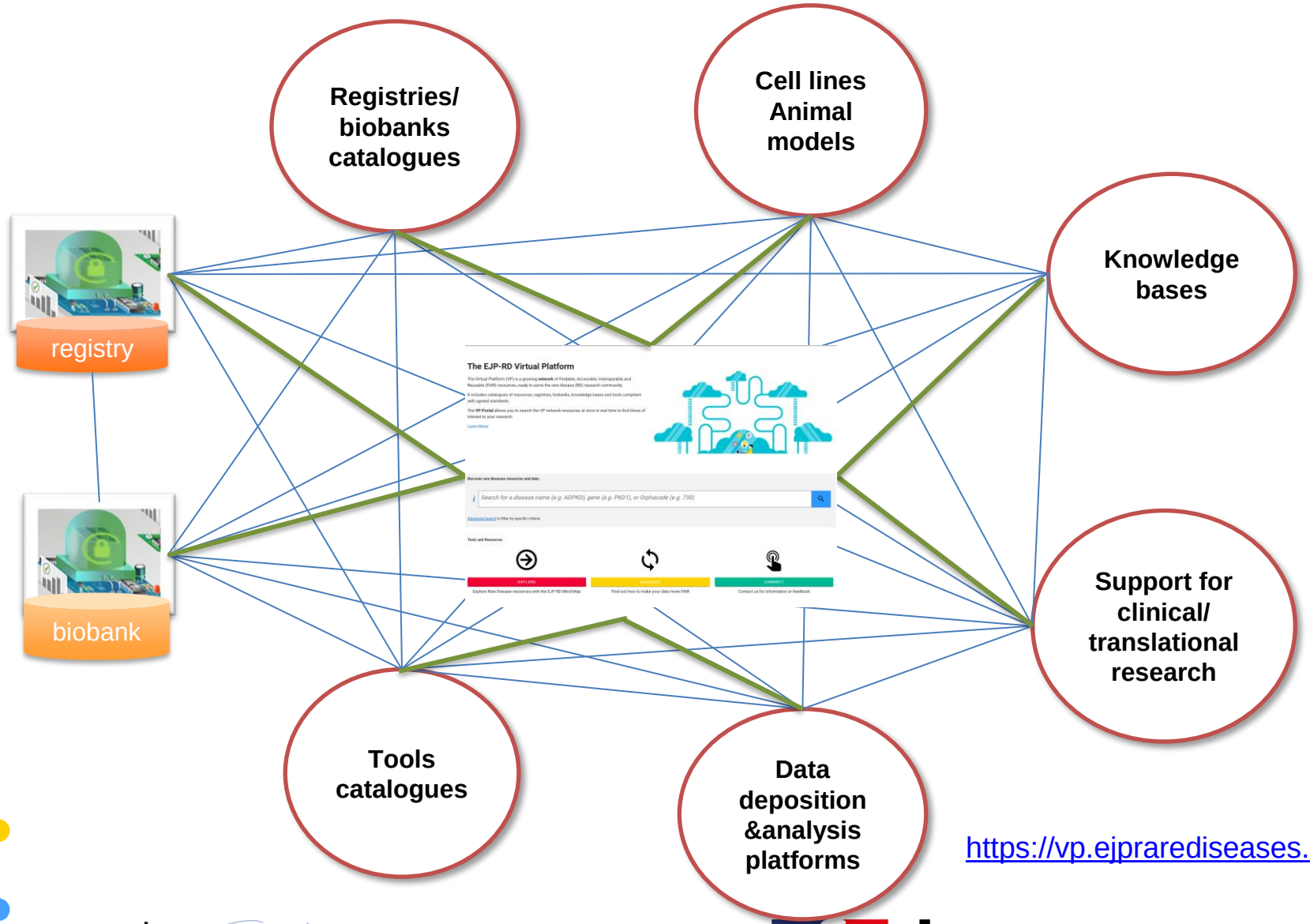


Support for clinical/ translational research

Tools catalogues

Data deposition & analysis platforms

...to the RD Virtual Platform: a network of federated resources



Decreasing fragmentation

Increasing interoperability

Increasing RD-readiness

through

Harmonisation

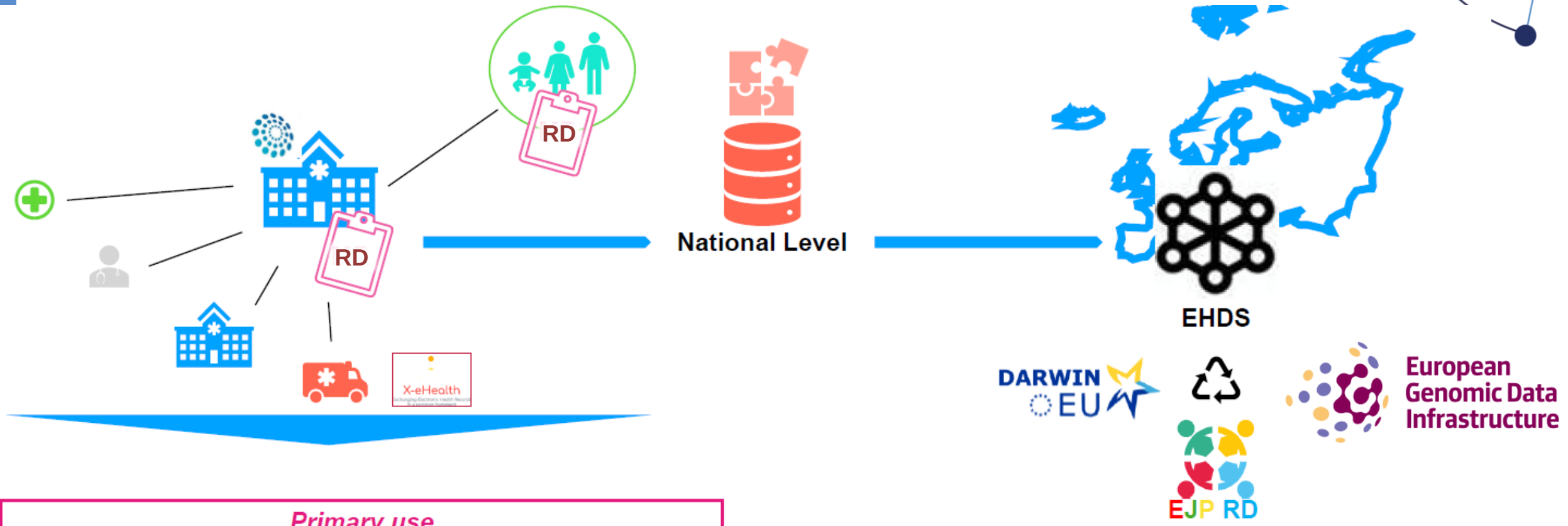
Standardisation

in a flexible way:

Common methodologies
Multipe technical solutions

https://vp.ejprarediseases.org

The future: RD in the data ecosystem



Primary use

- Better knowledge, best practices
- Continuity of care
- Better disability evaluation and compensation
- Adequate cross-border and primary care

Secondary use

- Research
- Evidence-base decision-making

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KNOW THE RARE

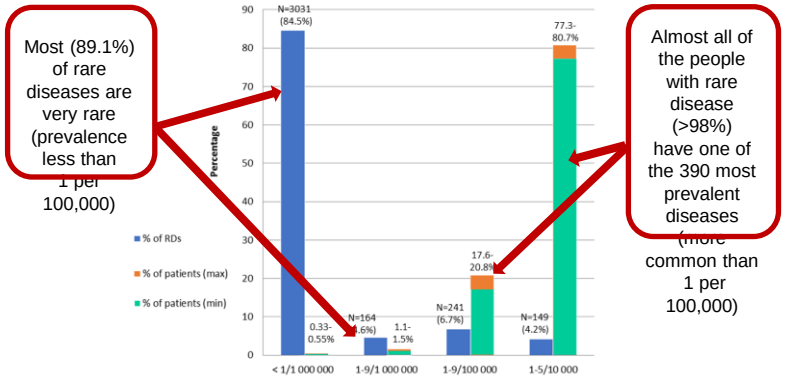
FOR BETTER CARE

THANK YOU
FOR YOUR ATTENTION

Orphadata science facilitates new knowledge

Global epidemiology

Most (89.1%) of rare diseases are very rare (prevalence less than 1 per 100,000)



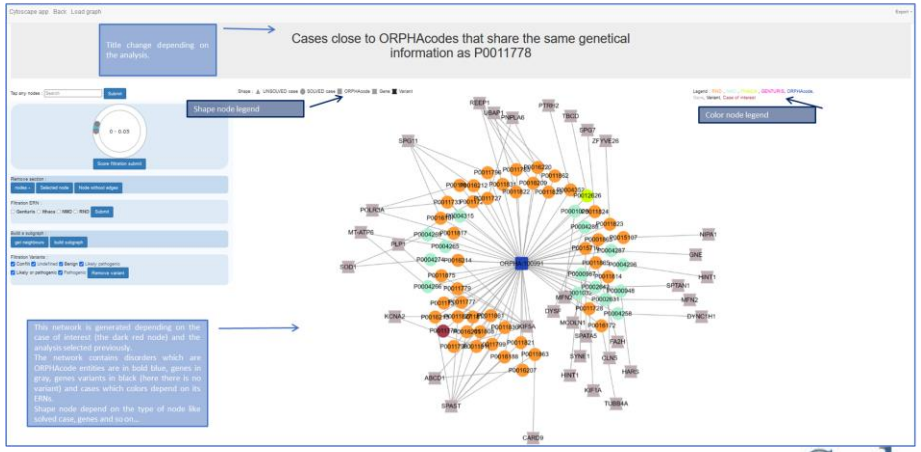
Almost all of the people with rare disease (>98%) have one of the 390 most prevalent diseases (more common than 1 per 100,000)

3.5 - 5.9% of the population (263 - 446 Million people) worldwide

https://www.nature.com/articles/s41431-019-0508-0

OrphaScape: Phenotype-based pipeline for variant prioritisation

Network which depend on the case of interest



SolveRD
Solving the Unsolved Rare Diseases

Semantic mapping services

orphanet_hierarchies_service Public
Service dedicated to support implementation and usage of Orphanet's classifications
0 Apache-2.0 0 0 0 Updated last week

Orphanet-GenesDisease-Mapper-API Public
Orphanet Genes Diseases Mapper API
Java 0 Apache-2.0 0 0 0 Updated last week

Orphanet-Endpoint-API Public
Orphanet mapping web services source code
Java 0 MIT 0 0 0 Updated on Jan 5, 2022

Orphanet-Mapping-API Public
This project focusses on the Orphanet mapping API.
Java 0 Apache-2.0 0 0 0 Updated on Jan 5, 2022

https://github.com/ejp-rd-vp

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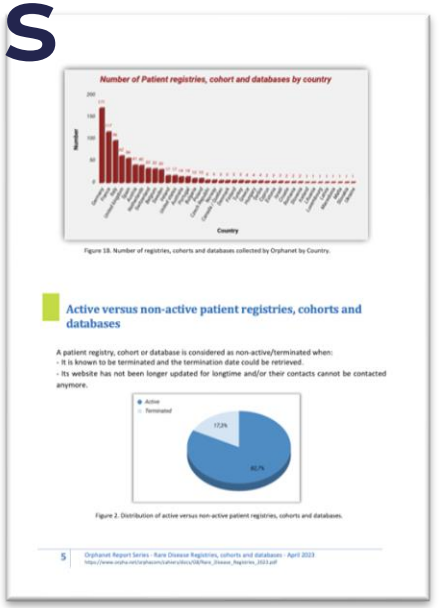
Orphanet Report Series

[Rare Disease Registries](#) April 2023

[Research Infrastructures useful to Rare Diseases](#) April 2023

Orphan Drugs

[Medicinal products for rare diseases in Europe](#) February 2023



https://www.orpha.net/consor/www/cgi-bin/Education_Home.php?lng=EN

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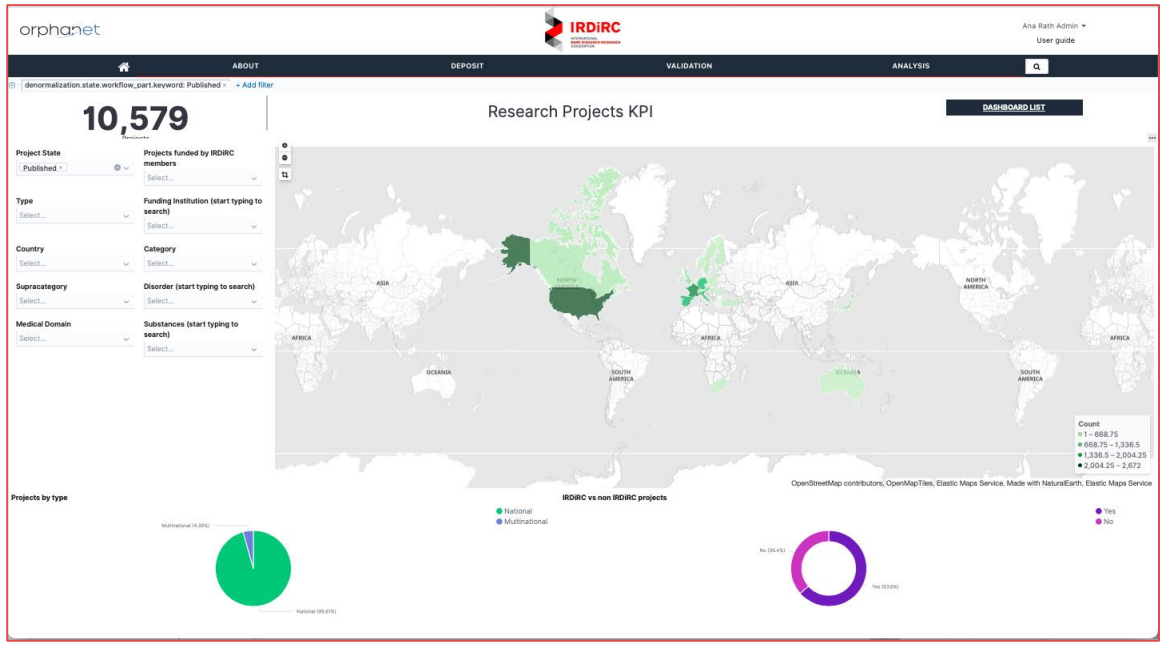
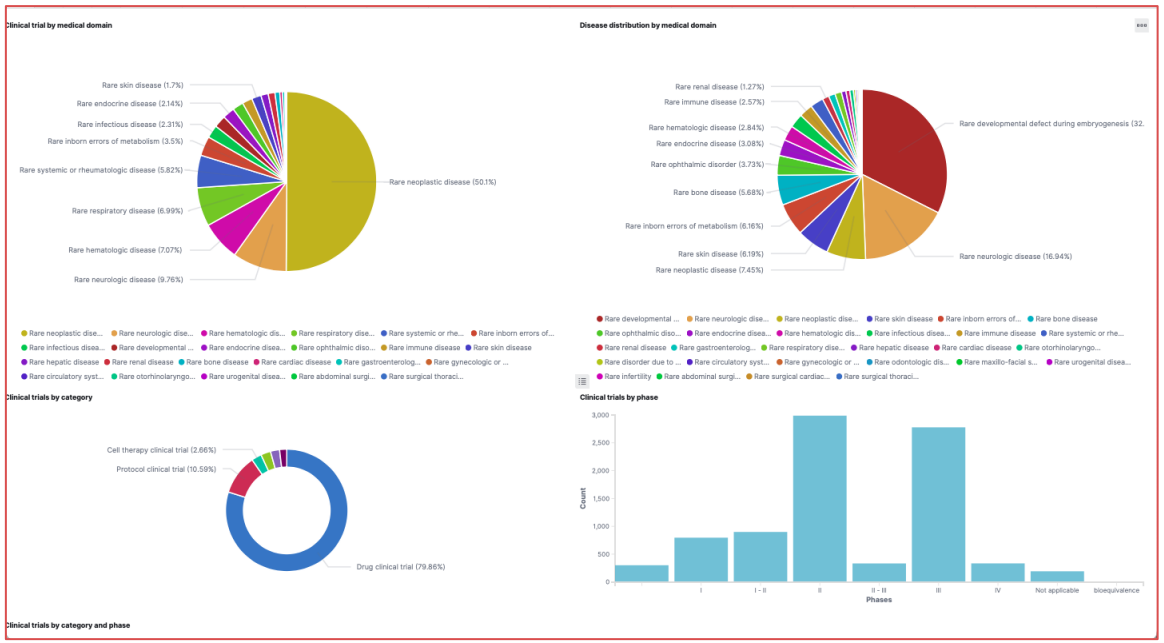
Visualisation & analysis of funded research

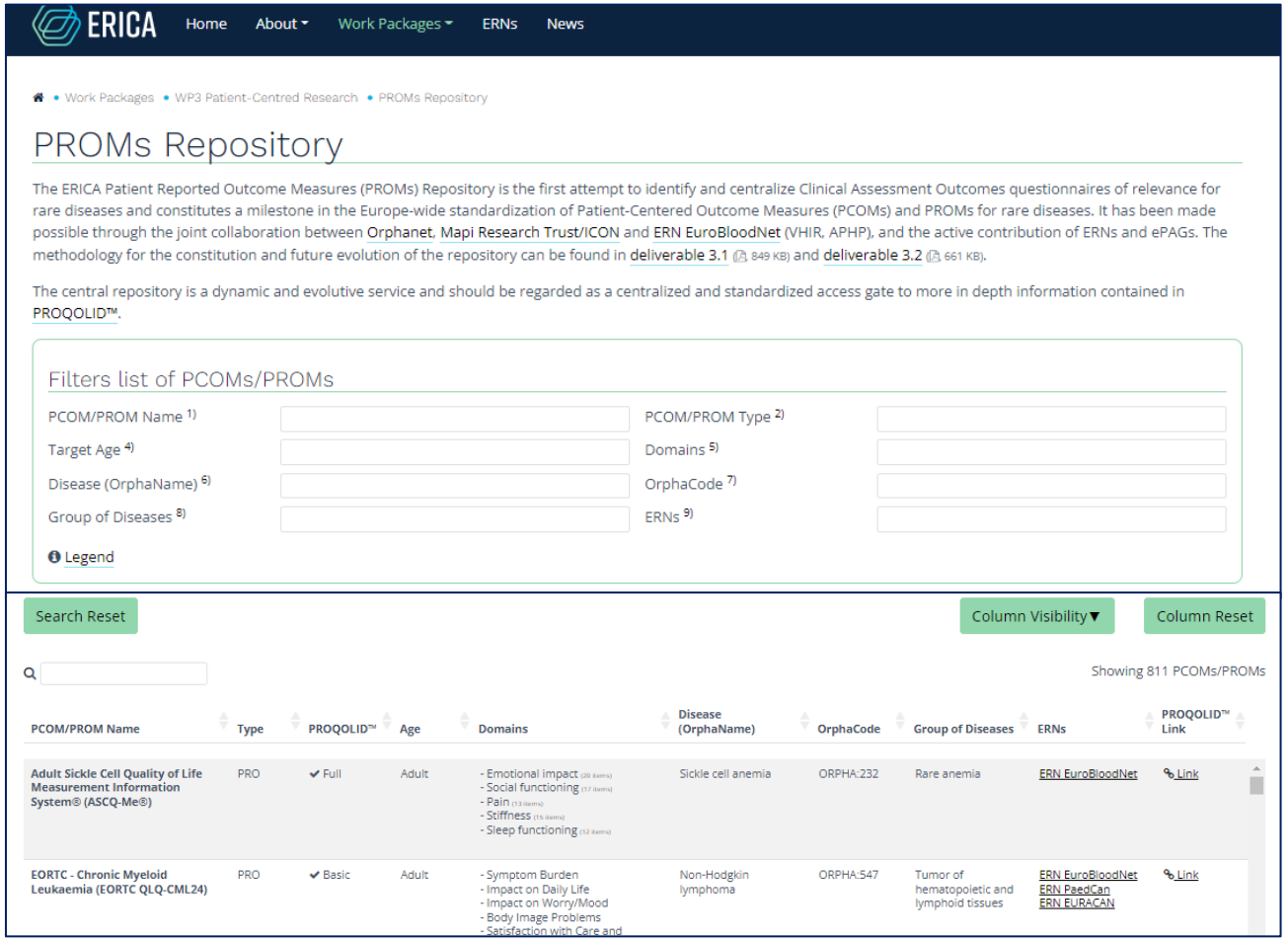




https://rare-research.orphanet.org

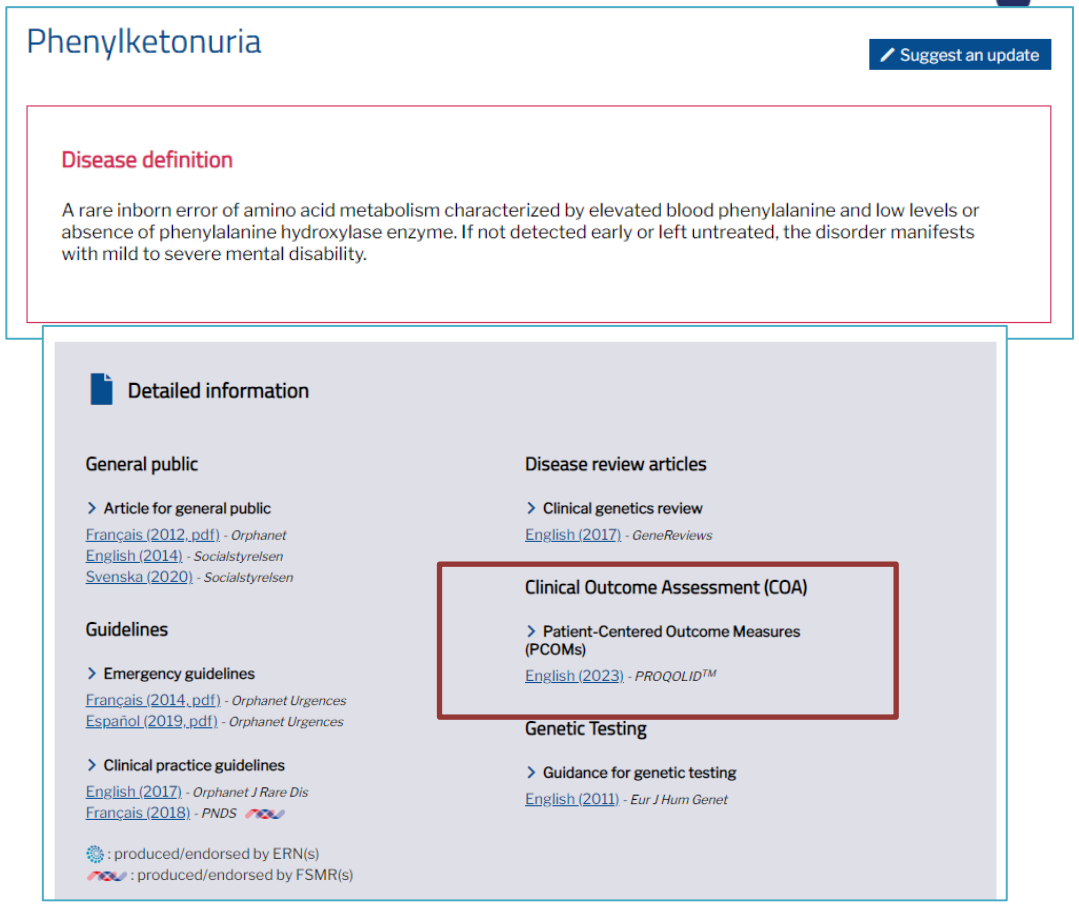
IRDiRC
INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM

EUROPEAN JOINT PROGRAMME RARE DISEASES



ERICA PROMs repository

https://erica-rd.eu/work-packages/patient-centred-research/proms-repository/



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