

ERN Registries: State of Play



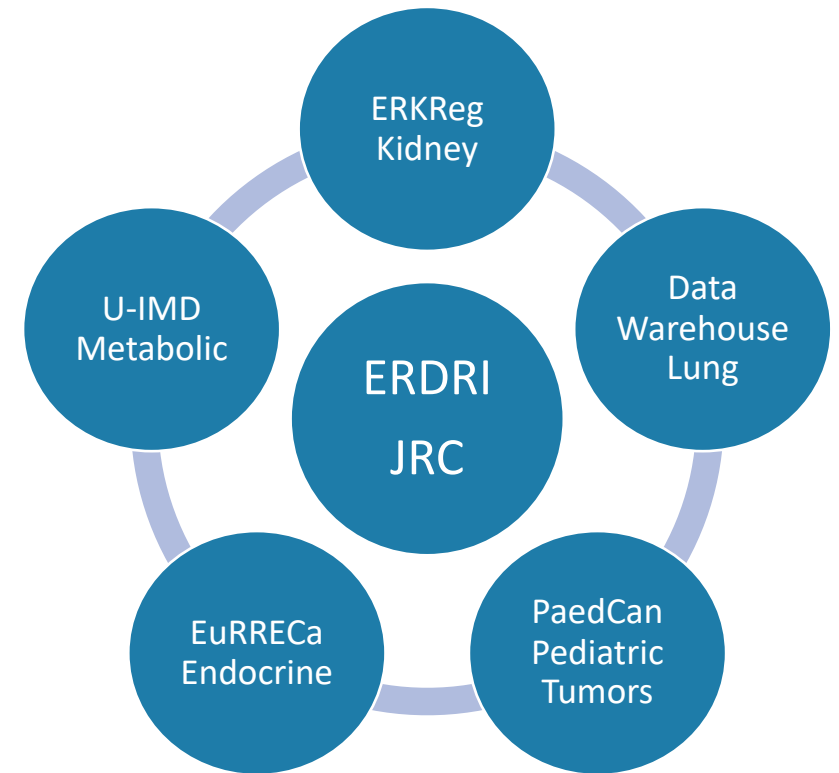
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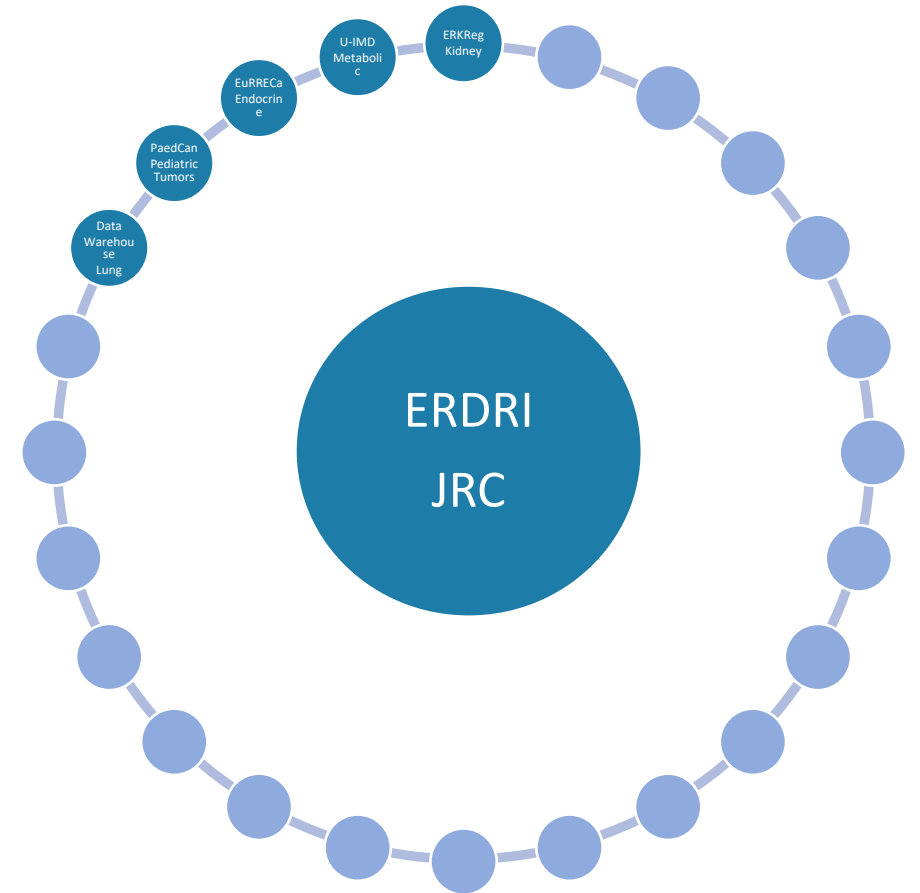
ERN-Wide Patient Registries

- 30 million patients with rare diseases in Europe
- **1.5-2 million patients treated in ERN centers**
- **Unique opportunity to collect information on**
 - disease demographics
 - patient cohorts available for clinical research
 - natural disease history / outcomes
 - efficacy and safety of disease management (,real world evidence‘)
- EU Health Programme 2018:
 - 5 ERNs funded to develop core registries

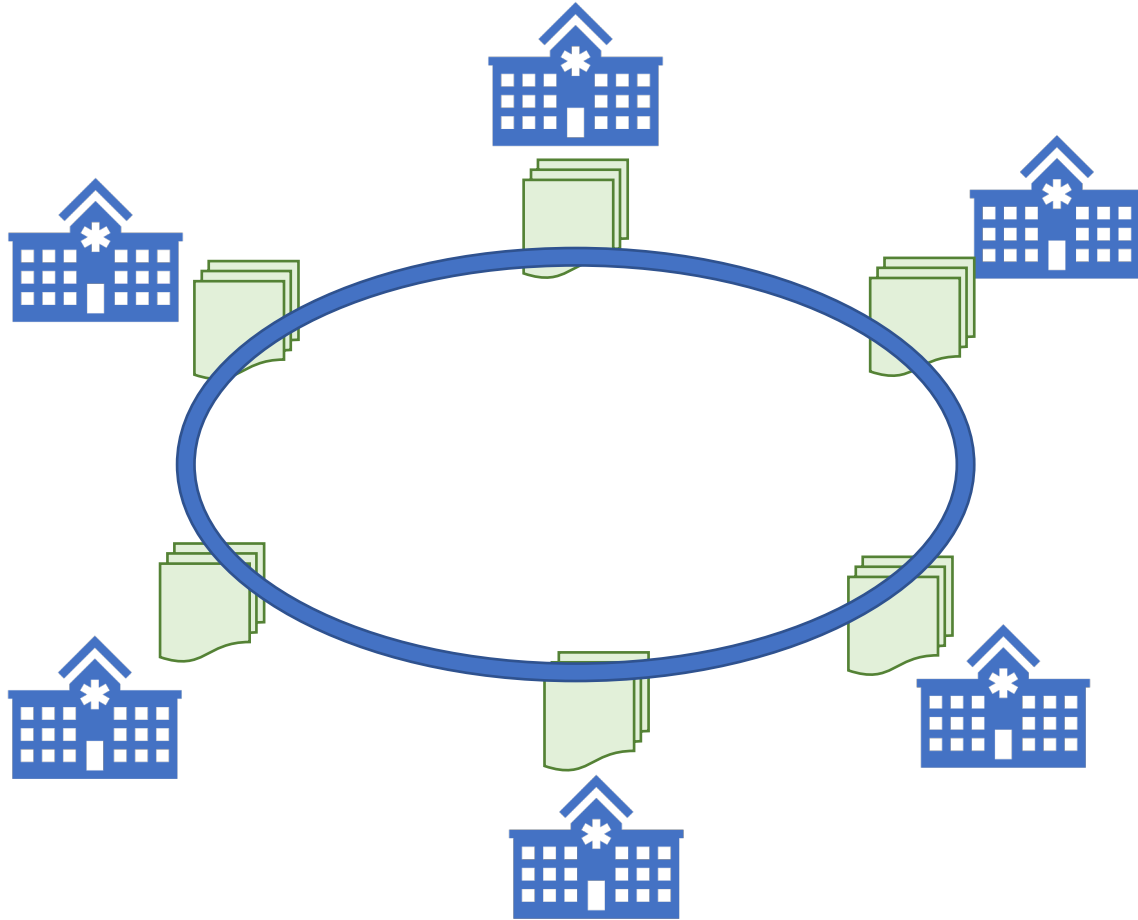


ERN-Wide Patient Registries

- Health Programme 2019:
Funding for registries of 19 remaining ERNs
- EU funding for designing, programming and roll-out of registries
- EJP-RD 2019-2024, ERICA 2021-2025:
Structured support for development of **FAIR** ERN registries



Federated Data Collection



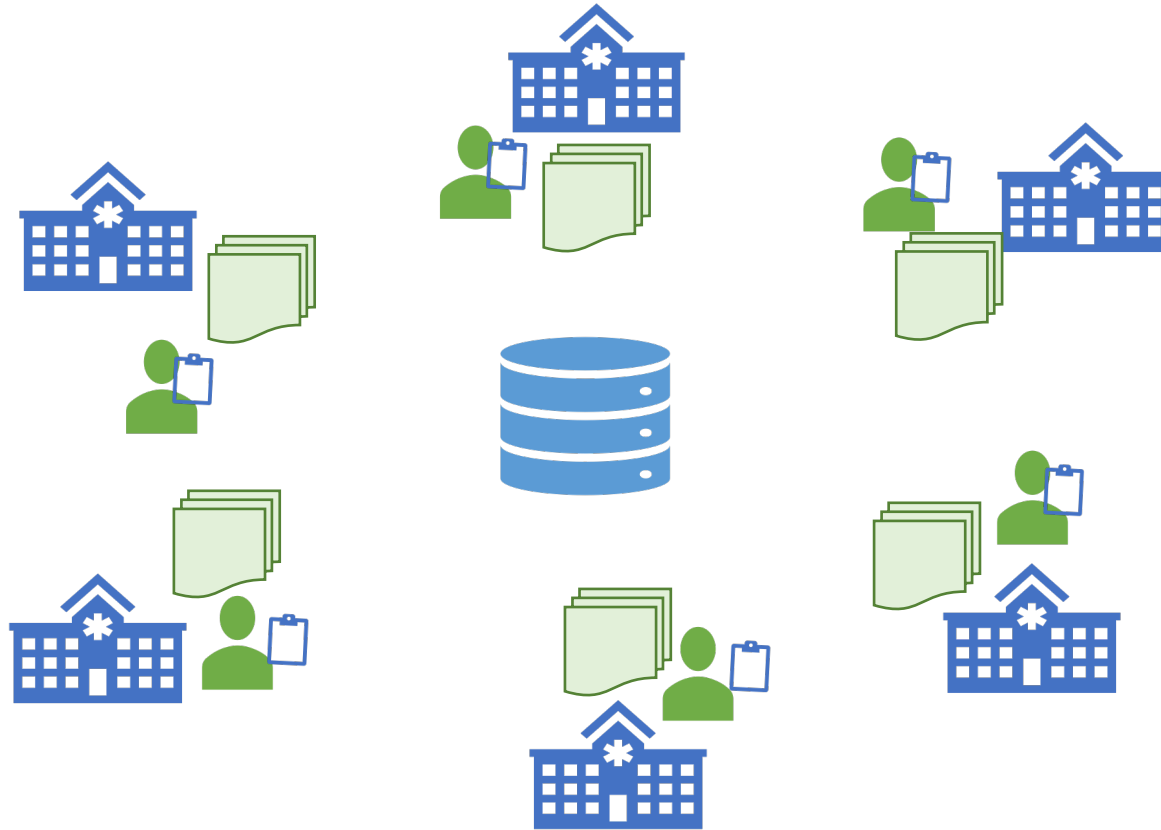
Advantage:

Data privacy preserved by design

But:

- Burden for hospital IT teams to set up eHR extracts
- Limited data sets available: unstructured data issue
- Limited statistical analysis options

Centralized Data Collection



Advantage:

Efficient data processing,
flexible data analysis

But:

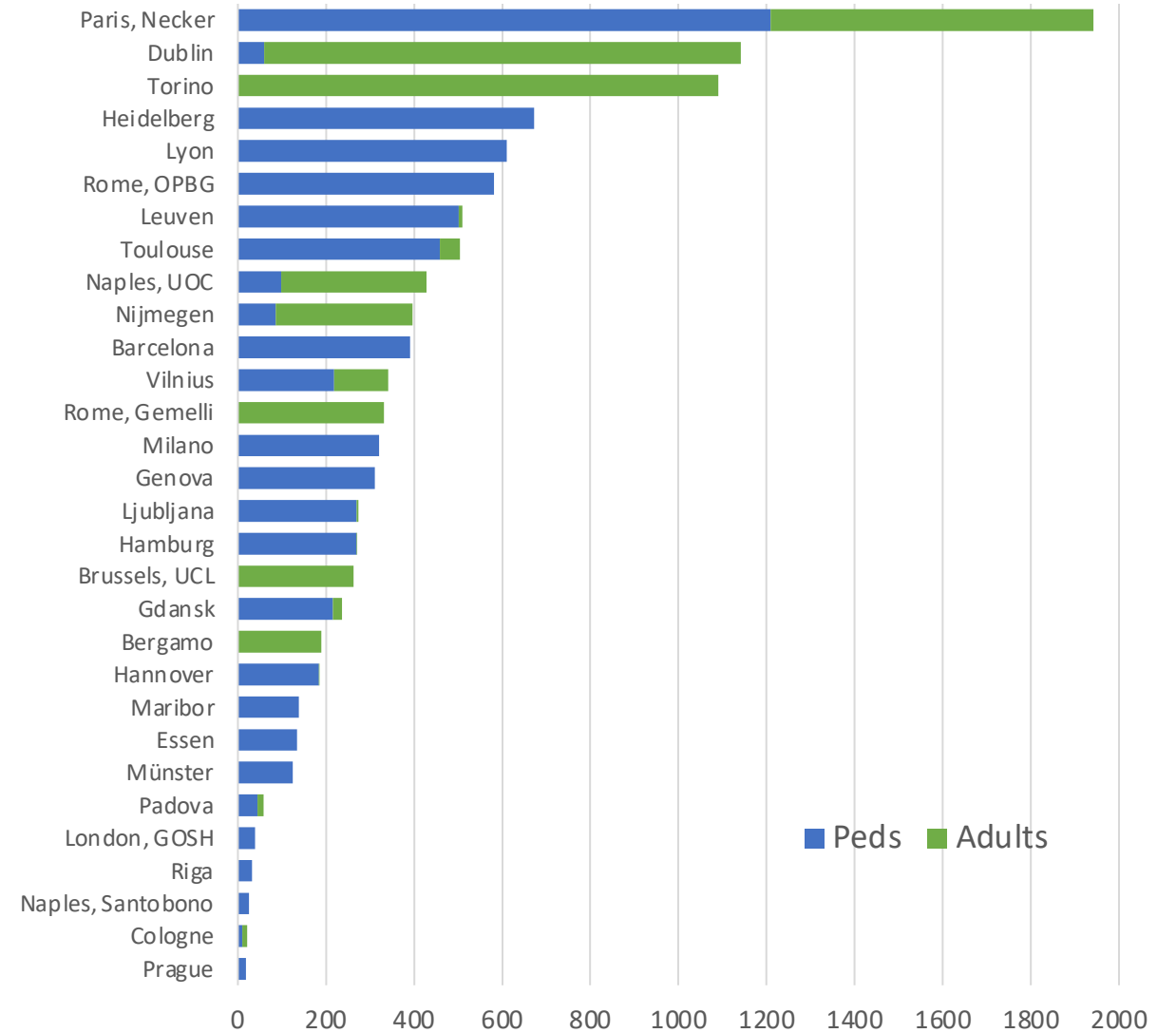
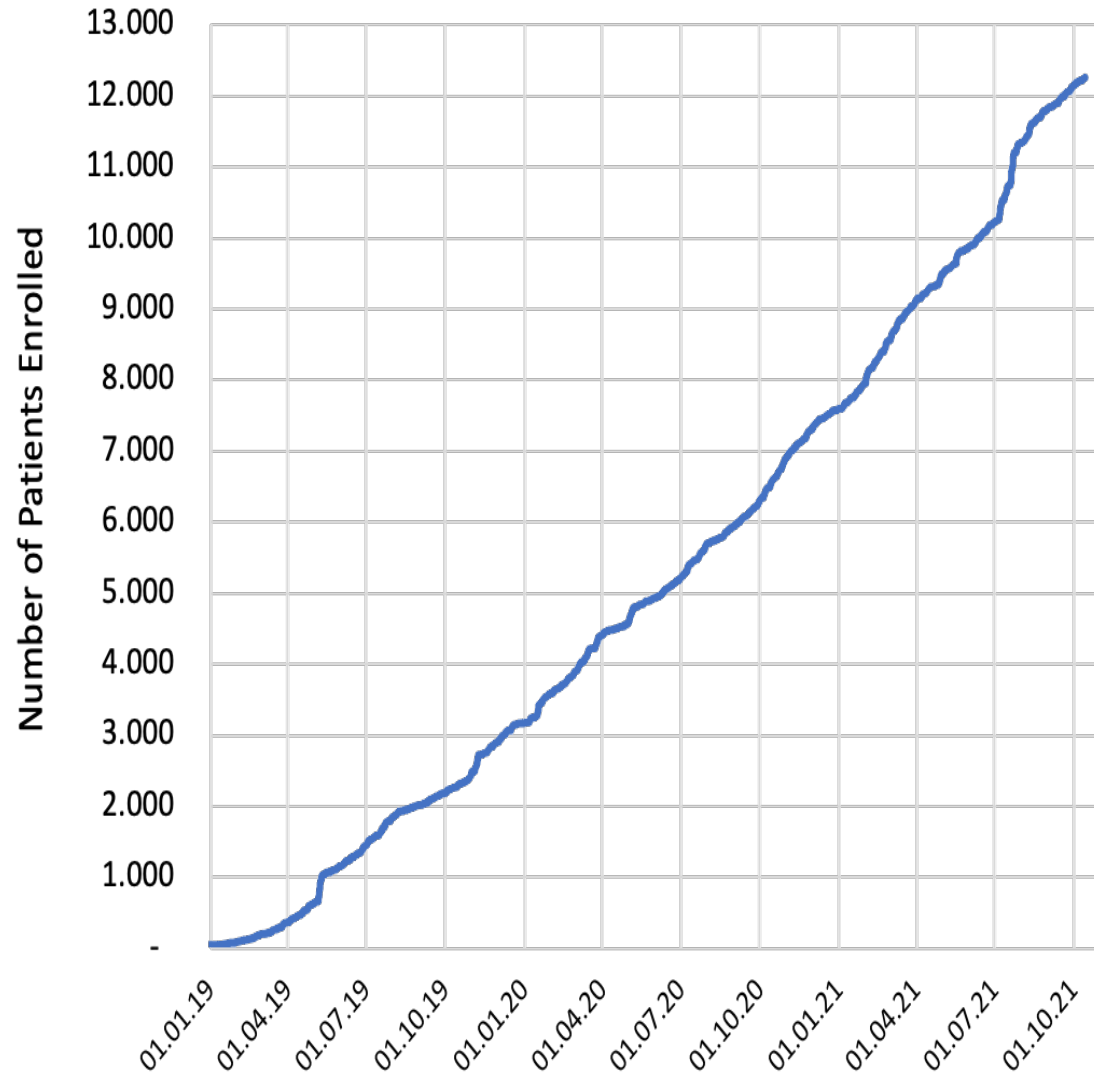
- GDPR requirements must be met
- Burden of data extraction and transfer

Automated EHR data extraction
currently not possible, due to

- partially unstructured documentation
- fragmented, non-interoperable IT systems

-> Manual data capture and transfer

European Rare Kidney Disease Registry: Patient Enrolment



Monitoring and Benchmarking of ERN Center Performance

	Pediatric patients	
	Your center	All ERKNet centers
% patients with hereditary glomerulopathies with genetic screening	83.6 % (61/73)	77.6 % (422/544)
% patients with multidrug resistant NS with comprehensive genetic screening	76.9 % (10/13)	59.1 % (26/44)
% normotensive patients (CKD1-3)	85.5 % (212/248)	76.2 % (1268/1663)
% patients with office blood pressure in target range	88.4 % (198/224)	79.6 % (1138/1430)
% patients with immunological glomerulopathies in clinical remission	0.0 % (0/0)	39.6 % (36/91)
% patients with persistent proteinuria who receive RAS antagonist therapy	100.0 % (1/1)	61.2 % (52/85)
% patients with hereditary and multidrug resistant NS on conservative treatment who are prescribed RAS antagonist therapy	63.2 % (12/19)	54.5 % (66/121)

WP2

**Data Collection,
Integration and Sharing**

WG Data collection
WG Legal aspects
WG Interoperability
WG Secondary data use

Standardized, GDPR compliant,
machine readable
Informed Consent Form
in all EU languages

Standardized Collaboration /
Data Transfer Agreements

Joint use of common data elements,
interoperability standards

Harmonized Data Access Policy

Pillar 2

Virtual Platform

FAIRification Stewards Team
Ethics & Regulatory Advisory Board



ERDRI.dor

RDConnect
orphanet

BBMRI-ERIC
EuroBioBank

Cell lines
Animal
models

hPSCreg

Semantic
standards

ordo
hpo
nextprot

Support for
clinical/
translational
research

ECRIN
EUROPEAN CLINICAL RESEARCH
INFRASTRUCTURE NETWORK

RaDiCo
Rare Disease Cohorts

EU-OPENSREEN
Chemical keys for life's locks

eatris

European infrastructure
for translational medicine

ChemBioNet
Network for Academic
Chemical Biology Research

EJP-RD
VP
Query &
Analysis

Registries/
biobanks
catalogs

Tools

elixir
bio.tools

Data
deposition
& analysis
platforms

RDConnect
GENOME-PHENOME ANALYSIS PLATFORM

EUROPEAN
GENOME-PHENOME
ARCHIVE

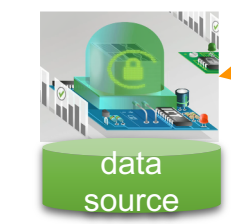
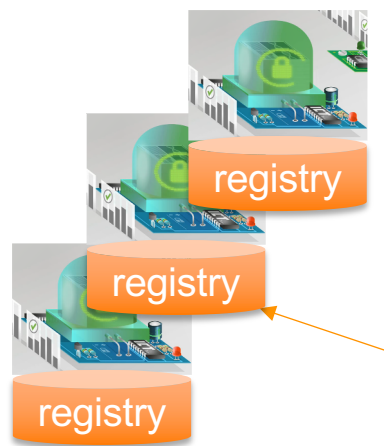
DECIPHER
GRCh37

Access &
privacy
control

ADA-M

EUPID

elixir
AAI



EJPRD X-omics

EU Vision: The European Health Data Space

Use case

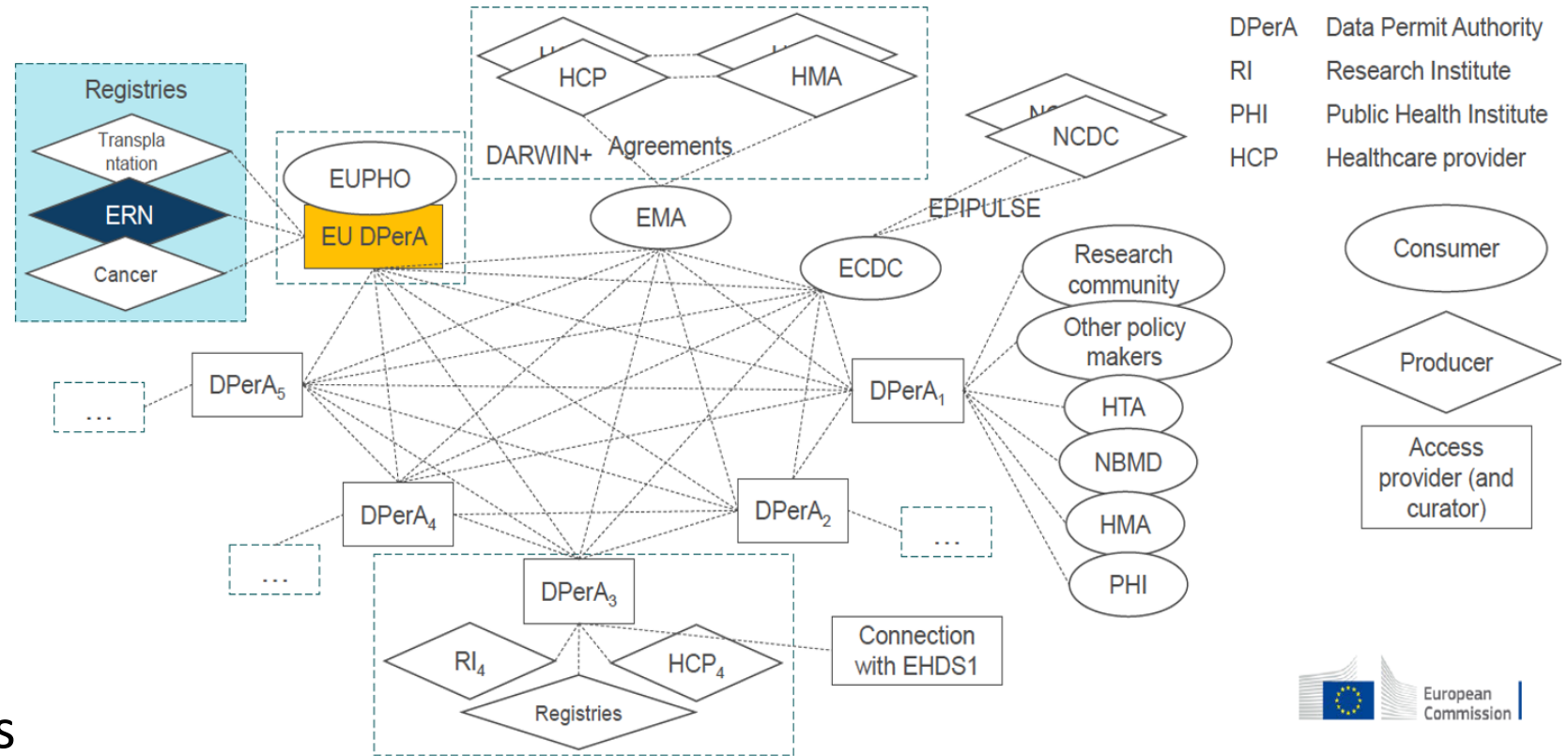
ERN Registries

Unprecedented potential to make RD data available to stakeholders

Aspects to consider:

GDPR – data privacy, dynamic informed consent

„Return on investment“ of PIs



Aspects of ERN Registry Data Sharing

What contents to share...

- Common Data Elements
- Disease specific data elements
- Phenotype/Intervention/Outcome data

how detailed...

- Counts (N patients with feature)
- Aggregated data (tables with descriptive stats)
- Anonymized patient level data
- Pseudonymized patient level data

when...

- Immediately upon collection ?
- Grace period (X years after collection) ?
- After publication?

... with whom?

Researchers inside ERN
Researchers outside ERN
Industry
National health authorities
EMA
HTA/Payers
Patient organizations