SESSION 1: EU ACTION PLAN FOR RARE DISEASES

PRIORITY ACTION 5: Comprehensive rare disease infrastructure cluster (CoRDIC)

EUROPEAN PRIORITY FOR RARE DISEASES

HLM4RARE WG5: Create, in each Member State, at least one comprehensive rare disease infrastructure cluster (CoRDIC) for research, innovation and care

CHALLENGES

- Rare disease research, innovation, diagnostics, care and clinical trial capacity remain dispersed across Member States and health systems.
- Patients often lack clear access points to navigate diagnostic, care and research pathways.
- Biobanks, registries, genomic initiatives, AI infrastructures and ERN centres remain insufficiently interconnected and interoperable.

ERN-EUROBLOODNET RESPONSE

Cross-border health

Best Practices

KEY INITIATIVES

JARDIN

EU ACTION PLAN FOR RARE DISEASES

- WG1 Governance & Action Plan
- WG2 Workforce Development
- WG3 Diagnostics & Early treatment
- WG4 Clinical Trials
- WG5 National Infrastructure
- WG6 Real-World Evidence
- WG7 Equitable access to therapies
- WG8 Funding & Sustainability

WORKING GROUP 5 (WG5) ASPIRATION

Creation of at least 1 Comprehensive Rare-Disease Infrastructure Clusters (CoRDIC) in each EU Member State

- CoRDICs should embrace and bundle the needed resources for research, innovation and care for rare disease patients: translational research facilities, advanced diagnostic laboratories, the necessary human resources to facilitate early access to diagnosis and state-of-the-art care. Infrastructure for clinical trials, with capacity to run Phase I to IV trials should be included.

- CoRDICS should be conceived as lighthouse nodes with an easily identifiable access point for rare disease patients to facilitate their diagnostic and care pathways as needed.

CORDIC: NATIONAL INTEGRATION

Clinical management	Advanced diagnostics	Digital infrastructure	Research & data leadership	Knowledge centres
• Multidisciplinary care units • Integrated paediatric + adult rare disease programmes • National referral centre status • Cross-border patient referral pathways • Highly Specialised Clinical Services	• Next-generation sequencing (NGS) platforms • Whole genome sequencing pipelines • Clinical bioinformatics teams • Molecular pathology laboratories • Variant interpretation boards	• ERN-trained clinicians using CPMS • digital imaging and data exchange • tele-consultation infrastructure • GDPR-compliant data governance • virtual multidisciplinary panels • cross-border case discussions • remote diagnosis and treatment planning	• clinical trial units • biobanks • translational medicine institutes • biomarker research laboratories • rare disease registries • structured patient registries • longitudinal outcome tracking • epidemiological data platforms • integration with European data infrastructures	• specialist training programmes • ERN fellowships • webinars and guideline development • professional education

ERNs roadmap

ERNs creation | **ERN implementation** | **ERNs integration** | **ERNs impact**

2017 — European Reference Networks — **2023** — European Reference Networks — **2027** — **2032**

ERNs MISSION

2019 call for membership to the existing European Reference Networks (ERNs)

Professionalization of ERNs

Statement of the ERN Board of Member States on Integration of the European Reference Networks to the healthcare systems of Member States

ERNs Actions Delivering

National Healthcare Systems

Joint **A**ction on **R**are **D**iseases and ERN **IN**tegration into National Healthcare Systems

To integrate the European Reference Networks (ERNs) into national healthcare systems

- **WP1 – Coordination & Management**
- **WP2 – JA Dissemination & ERN Dissemination**
- **WP3 – Evaluation**
- **WP4 – Sustainability & National Plan Capacity**
- **WP5 – National Governance and Quality Assurance**
- **WP6 – National Care Pathways and ERN Referral Systems**
- **WP7 – National Reference Networks & Undiagnosed Disease Programmes**
- **WP8 – Data Management**
- **WP9 – National Support Options for ERN-HCP**

- Duration:
  - 36 month (01.02.2024 - 31.01.2027; all other participants)
- Participants:
  - 58 partner institutions
  - 29 Member States
    - 28 Competent authorities (CA)
    - 29 Affiliated Entities (AE)
    - 1 Associated partner

Current situation in EU Member States and Norway

- One focus of the JARDIN project: **establishment and improvement of National Reference Networks** (NRNs), suited to integrate ERNs activities.
- As part of WP7 of the JARDIN project, an analysis was carried out to obtain an overview of the current situation of the NRNs and similar national structures in all MS and Norway.

Table 2: Categorisation of country by the responses of both stakeholders

Existence of NRNs (Category A*)	Existence of similar national structures (Category B**)	No national structure (Category C***)
Bulgaria (PO)[a]	Belgium (NPS)	Austria
Croatia (PO)	Cyprus	Lithuania
Czechia (PO)	Hungary	Luxembourg
Denmark[b]	Latvia (PO)	Malta
Estonia (PO)	Norway	Poland
Finland	Slovenia	Portugal
France		Sweden
Germany		
Greece		
Ireland (NPS)		
Italy		
Netherlands (NPS)		
Romania (NPS)		
Slovakia		
Spain (PO)		

PO: Patient organization
NPS: national policy stakeholder
Source: *JARDIN D7.1 State-of-play report, national reference networks, 12th February 2025*

ERN | EuroBloodNet

12th June 2026 - EHA Congress

Integration of the ERN into the National care and research system: Belgium

Béatrice GULBIS, M.D., PhD

Head Rare Disease Function

Co-coordinator ERN-EuroBloodNet

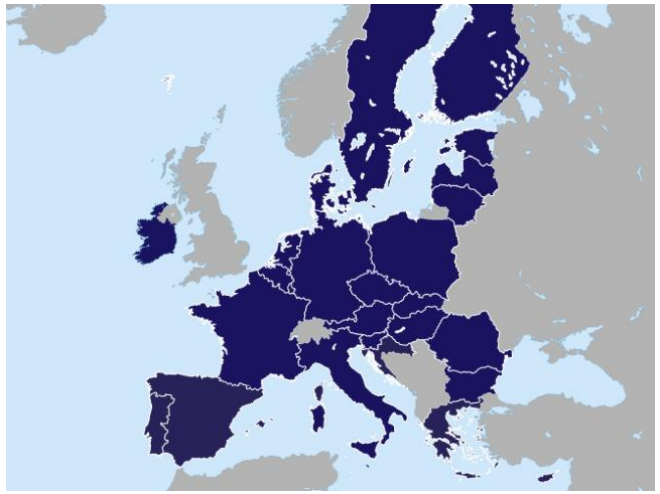
Rare diseases

Funded by the European Union

Contributing to the new Belgian Rare Disease Plan 2026-2030: Perspectives to share - Ecosystem

European Commission

Board of Member States



ERNs GOVERNANCE

ERN Coordinators Working Group
Project Managers Working Groups
ERN Thematic Working Groups

(Managers)

Local
National
EU
International

Expertise centres
Health care units

Patients RD

Contributing to the new Belgian Rare Disease Plan 2026-2030: Perspectives to share – European label - examples

MADE IN EU

EURORDIS
RARE DISEASES EUROPE

Belgian Rare Disease plan outside rare cancers

2013

First RD plan

2026-2030

24 ERNs
implementation, FU

2017-2022 (2019) & 2023-2027

European Reference Networks

Co-funded by the European Union

JARDIN

INTERNAL NEWSLETTER #4
JANUARY 2026

RARE DISEASE PLAN

European Union

Since 2017

European Commission/ DG Santé

24 ERNS working group

European Reference Network
for rare or low prevalence complex diseases

Expertise Centres

EURORDIS RARE DISEASES EUROPE

JARDIN

Belgium

Since2018

Belgian Health Authorities Federal/Regional

8 Rare Disease Functions

CHU de Liège · H.U.B · Institut des Maladies Rares · IPG · UZA · UZ Brussel · UZ Gent · UZ Leuven

7 Academic hospitals + 1
8 Human genetic centres

Expertise centres

RaDiOrg RARE DISEASES BELGIUM

Funded by the European Union

Contributing to the new Belgian Rare Disease Plan 2026-2030:
Did you say "Administrative burden"?

		Endo-ERN	EpiCARE	ERKNet	ERN CRANIO	ERN GENTURIS	ERN-BOND	ERN-EYE	ERNICA	ERN-LUNG	ERN-PaedCan	ERN-RND	ERN-Skin	EURACAN	EuroBlood Net	eUROGEN	EURO-NMD	GUARD-HEART	ITHACA	MetabERN	RARE-LIVER	ReCONNET	RITA	TransplantChild	VASCERN
Leuven University Hospital (UZ Leuven)	22	1	1	1	1	1	0	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0	1
Ghent University Hospital (UZ Gent)	21	1	0	1	1	1	1	1	1	1	1	1	1	1	0	1	1	0	1	1	1	1	1	1	1
Antwerp University Hospital (UZ Antwerpen)	15	1	0	0	1	0	1	1	0	1	0	1	0	1	1	1	1	1	1	1	1	0	0	0	1
University Hospital Brussels (UZ Brussel)	5	1	0	0	0	1	0	0	0	1	0	0	0	0	0	0	0	1	0	1	0	0	0	0	0
AZ Sint-Maarten (Mechelen)	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1
Cliniques universitaires Saint-Luc (UCLouvain)	15	1	1	1	0	0	0	0	1	1	1	0	0	1	1	0	1	0	1	1	1	1	0	1	1
H.U.B - University Hospital Erasme	7	1	0	0	0	0	0	0	0	1	0	1	1	0	1	0	1	0	1	0	0	0	0	0	0
H.U.B - Jules Bordet Institute	2	0	0	0	0	0	0	0	0	0	0	0	0	1	1	0	0	0	0	0	0	0	0	0	0
H.U.B - Hôpital Universitaire des Enfants Reine Fabiola (HUDERF)	1	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	In collaboration with UZ Brussel	0	0	0	0	0
Liège University Hospital (Centre Hospitalier Universitaire de Liège)	5	1	0	0	0	1	0	0	0	0	0	0	0	1	1	0	0	0	0	1	0	0	0	0	0
TOTAL	94	7	2	3	3	4	2	3	3	6	4	4	3	6	6	3	5	3	5	6	4	3	2	2	5

Table 2. Number of Belgian centres recognized as full members in the 24 existing ERNs, by hospital (as of December 2023)

Approval of application as full ERN member

- Same material as the one for EC/DG Santé

Cartography of expertise (new RD Belgian plan)

- Largely inspired by ERN material

Contributing to the new Belgian Rare Disease Plan 2026-2030
Where can I find the expertise?

ERN	Full Members
BOND	43
CRANIO	35
Endo-ERN	92
EpiCARE	38
ERKNet	64
ERN-EYE	51
ERNICA	39
ERN-LUNG	78
ERN-RND	63
ERN-SKIN	52
EURACAN	90
EuroBloodNet	89
eUROGEN	51
EURO-NMD	73
GENTURIS	44
GUARD-HEART	44
ITHACA	65
MetabERN	85
PaedCAN	79
RARE-LIVER	52
ReCONNET	54
RITA	61
TRANSPLANTCHILD	33
VASCERN	40
TOTAL	**1 415**

		Endo-ERN	EpiCARE	ERKNet	ERN CRANIO	ERN GENTURIS	ERN-BOND	ERN-EYE	ERNICA	ERN-LUNG	ERN-PaedCan	ERN-RND	ERN-Skin	EURACAN	EuroBlood Net	eUROGEN	EURO-NMD	GUARD-HEART	ITHACA	MetabERN	RARE-LIVER	ReCONNET	RITA	Transplan tChild	VASCERN
Leuven University Hospital (UZ Leuven)	22	1	1	1	1	1	0	1	1	1	1	1	1	1	1	1	1	1	1	1	1	1	0	1
Ghent University Hospital (UZ Gent)	21	1	0	1	1	1	1	1	1	1	1	1	1	1	0	1	1	0	1	1	1	1	1	1	1
Antwerp University Hospital (UZ Antwerpen)	15	1	0	0	1	0	1	1	0	1	0	1	0	1	1	1	1	1	1	1	1	0	0	0	1
University Hospital Brussels (UZ Brussel)	5	1	0	0	0	1	0	0	0	1	0	0	0	0	0	0	0	1	0	1	0	0	0	0	0
AZ Sint-Maarten (Mechelen)	1	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	1
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H.U.B - Jules Bordet Institute	2	0	0	0	0	0	0	0	0	0	0	0	0	1	1	0	0	0	0	0	0	0	0	0	0
H.U.B - Hôpital Universitaire des Enfants Reine Fabiola (HUDERF)	1	0	0	0	0	0	0	0	0	0	1	0	0	0	0	0	0	0	0	In collaboration with UZ Brussel	0	0	0	0	0
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TOTAL	94	7	2	3	3	4	2	3	3	6	4	4	3	6	6	3	5	3	5	6	4	3	2	2	5

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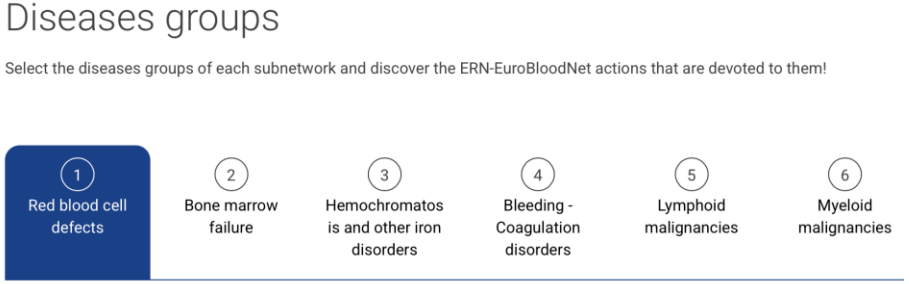
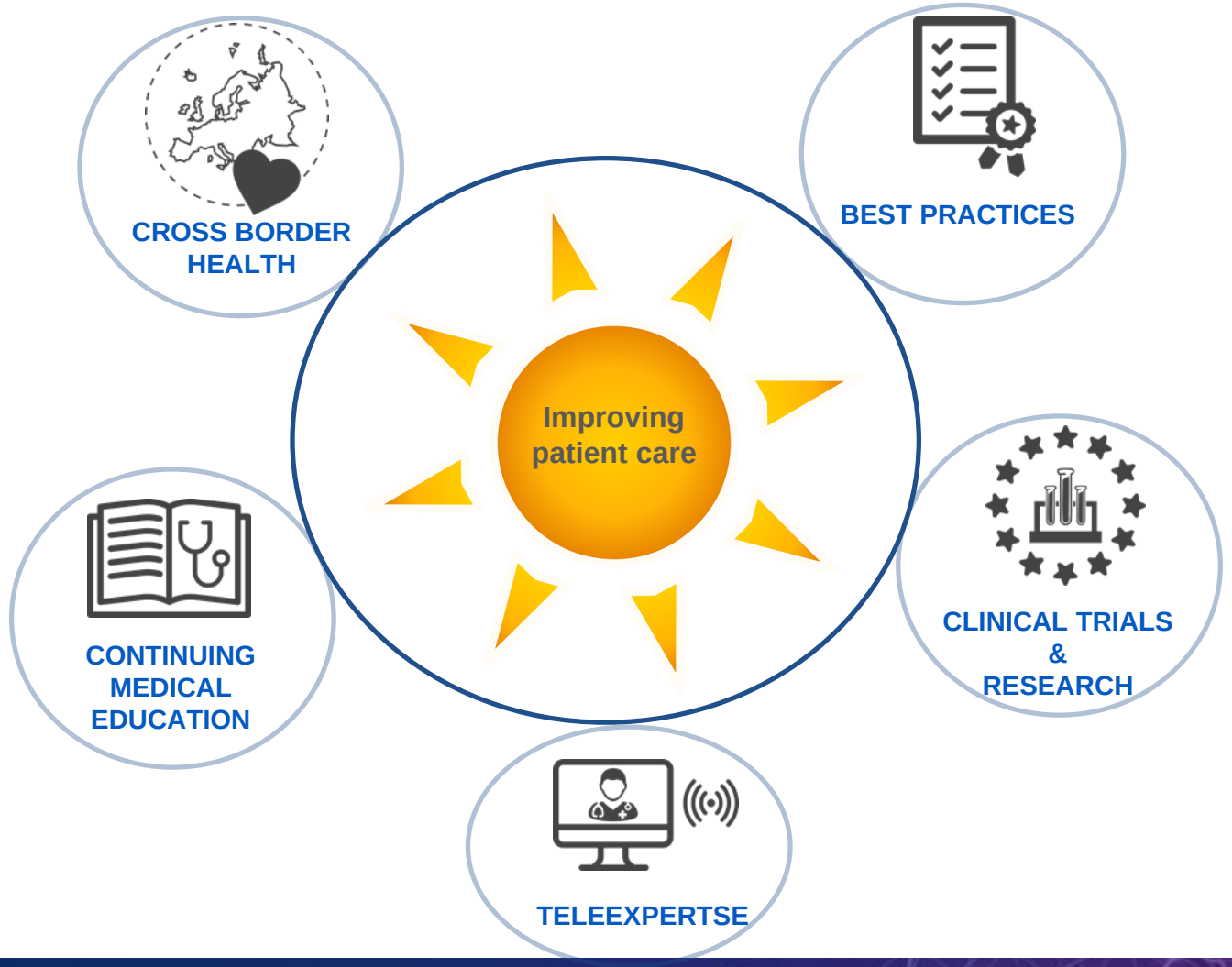


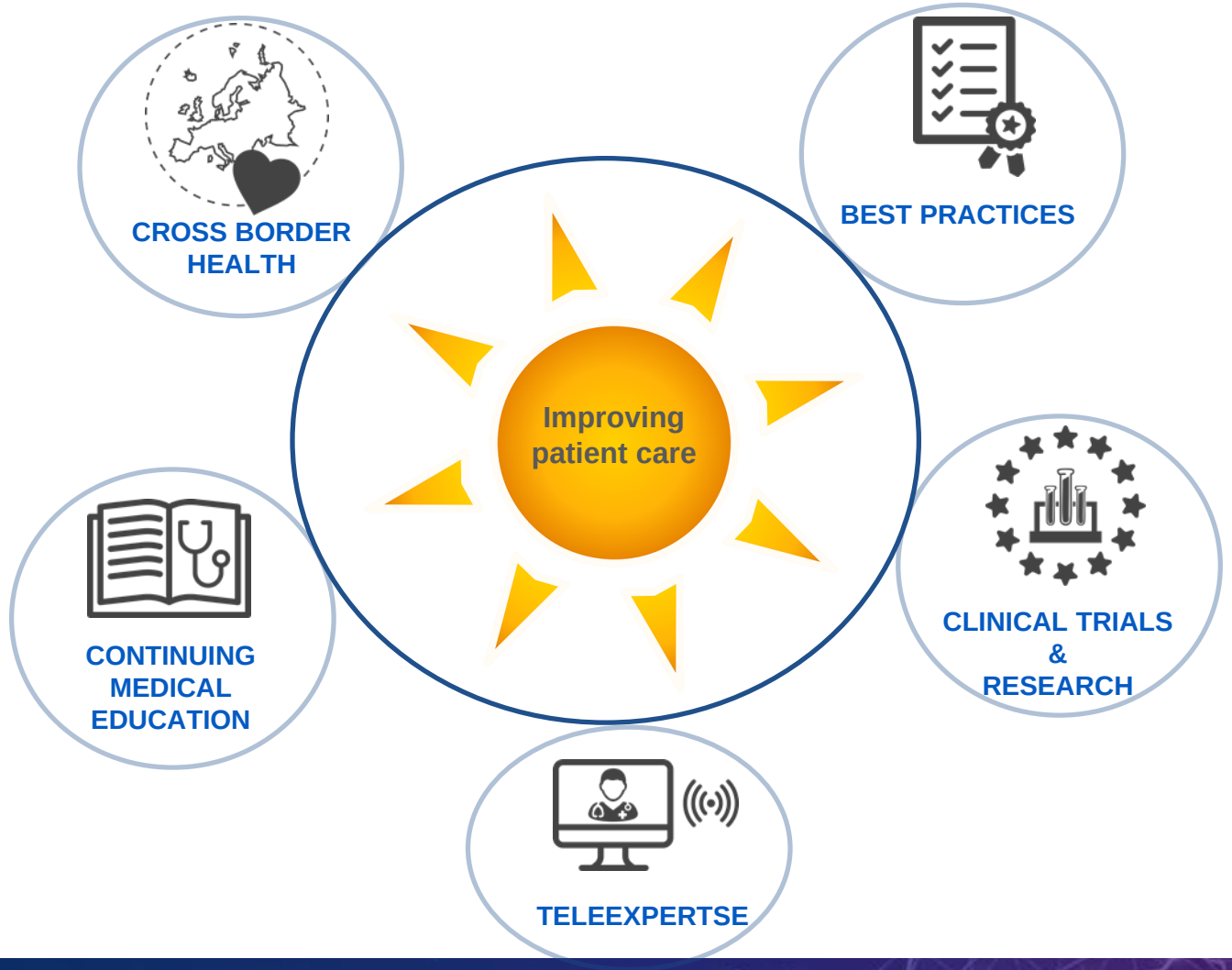
Contributing to the new Belgian Rare Disease Plan 2026-2030:
How can I ask for an expert opinion about a complex, rare case? AND avoid administrative burden…



Belgian action plan
From national tool to EU tool

CPMS Care 2.0

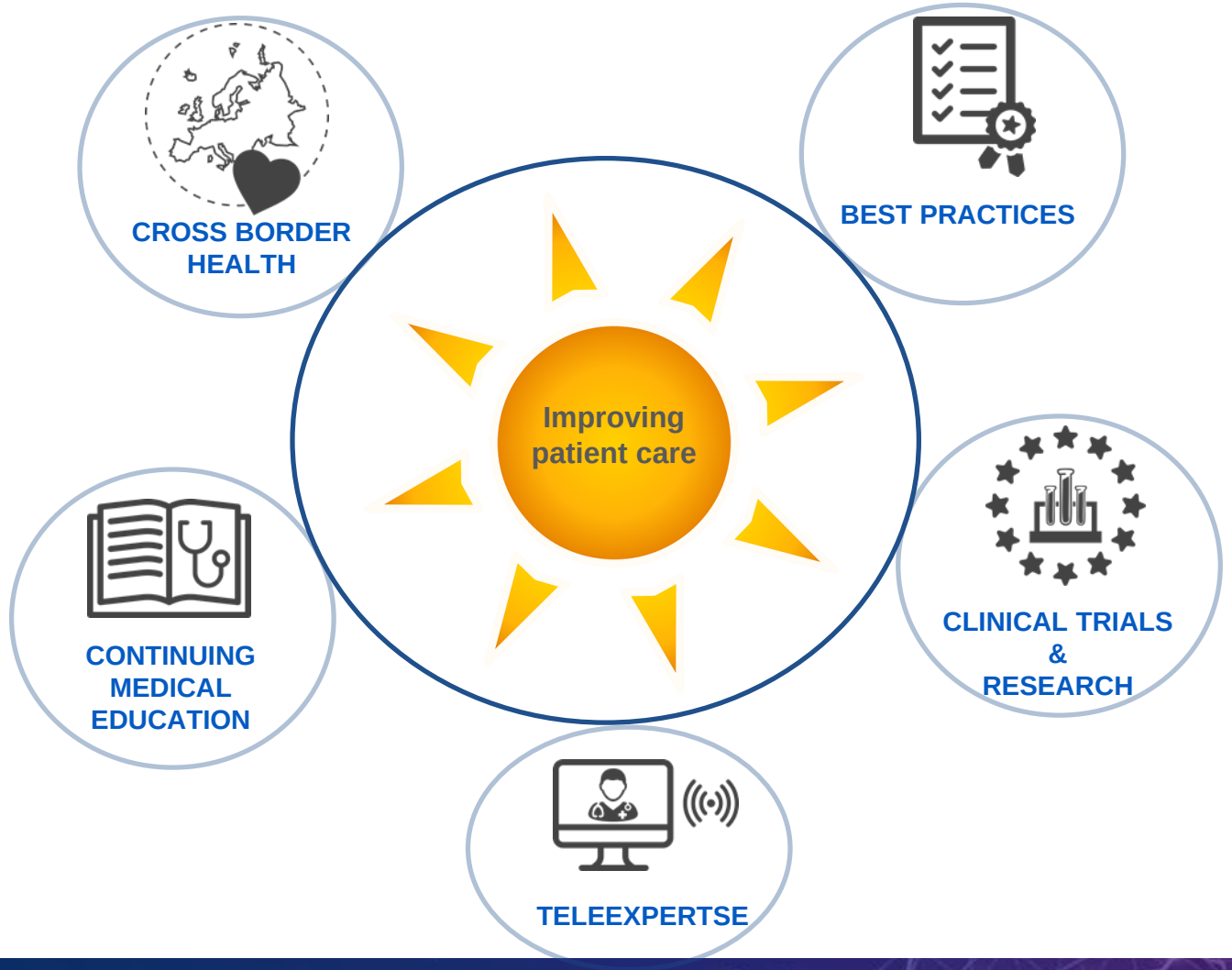
Contributing to the new Belgian Rare Disease Plan 2026-2030:
How can I participate into registries ?
AND avoid administrative burden…



Belgian central rare disease registry

- 16 common data elements (EU)
- ORPHA codes (but SNOMED in medical records)

Contributing to the new Belgian Rare Disease Plan 2026-2030: *Do we forget several aspects?*



To speed up diagnosis from the first line of care

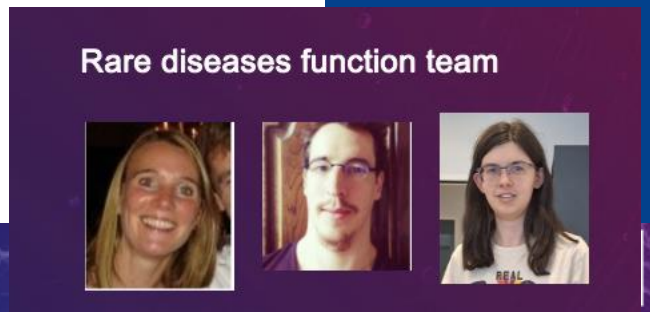
Common, national and EU, perspectives

Observations

- We are still a young community
- The new Belgian national plan on RD shares EC strategy on rare diseases
- Fragmentation across all levels represents a major barrier to the appropriate care of patients

Perspectives

- Accelerate visibility of expertise
- Adoption of dedicated, common tools
  - ORPHA codes
  - CPMS 2.0
  - …
- Include all level of care
  - General practitioners
  - …



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ERN | EuroBloodNet

12th June 2026 - EHA Congress

Integration of the ERN into the National care and research system: Italy

Raffaella Colombatti
Azienda Ospedale – Università di Padova, Italy

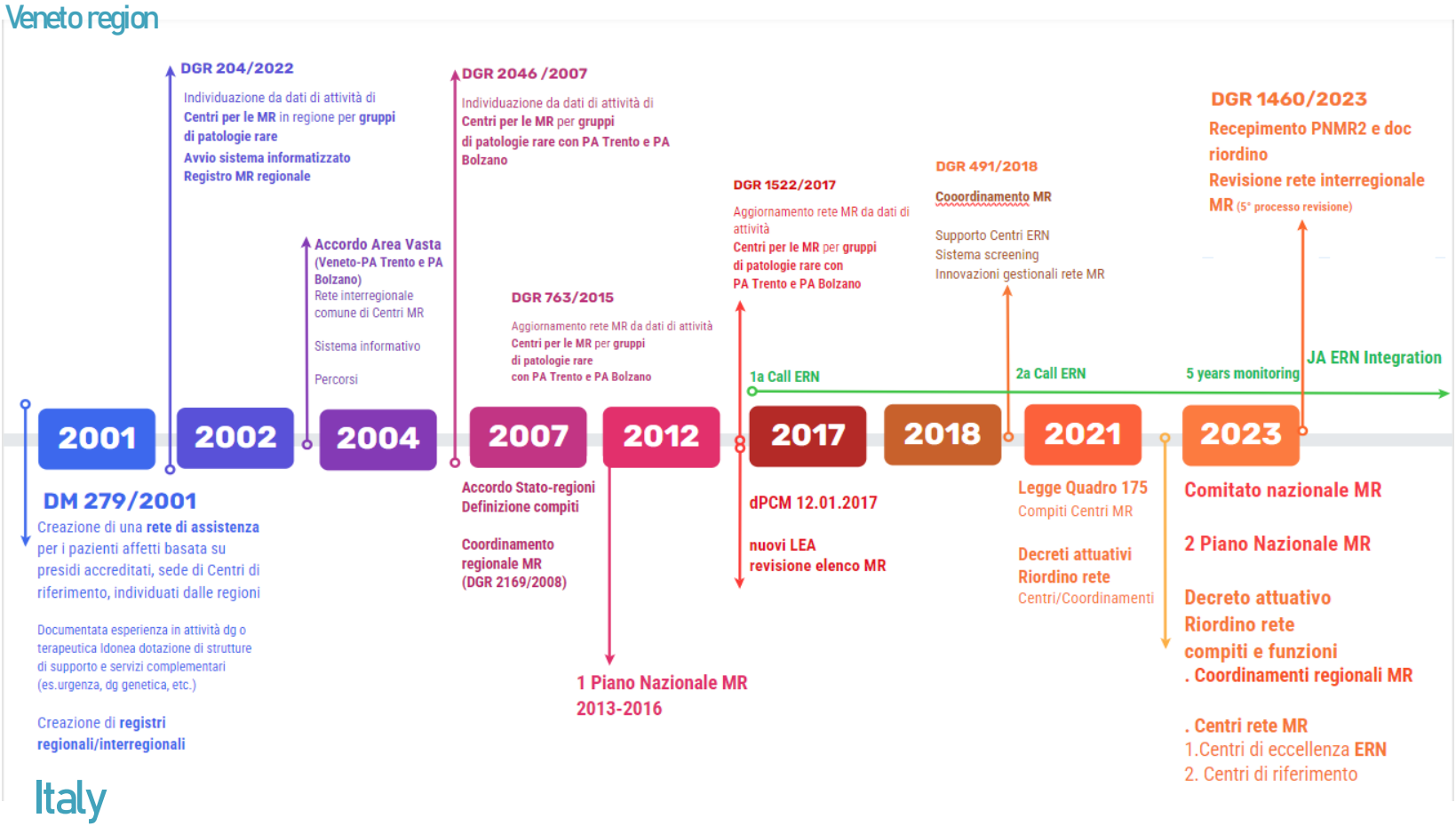
Funded by the European Union

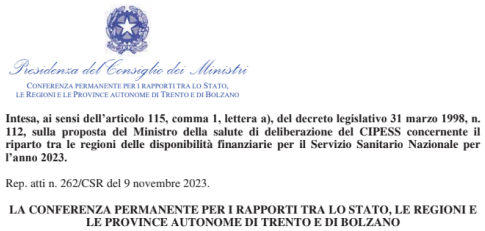
Background –Italy Rare Disease (RD) scenario

The NHS is decentralized.
The central government is responsible for defining health policy strategies, the national benefits package and the per capita budget (in collaboration with regions)

Regional level of governance: in charge of health service provision (monitored)

Laws & State-regions agreements
[adapted from *Italy- Health Systems review, 2022*]





PIANO NAZIONALE
MALATTIE RARE
2023-2026

Financing To Regions according data provision

HCPs data

↓

Regional Coordinating Centres (RHA)

↓

National level (MoH)

Data evaluated by the National RD Committee

Requirements for provision

1) Formal implementation of the National Rare Disease Plan (PNMR) and the Reorganization Document.

2) Formal approval of the Centers of Excellence (ERN), Reference and Coordination that perform the tasks and functions established by the Reorganization Document of the National Rare Disease Network.

3) Final summary report, accompanied by data, of the activities carried out by the Centers of Excellence (ERN), Reference and Coordination belonging to the National Rare Disease Network

RD Reference Centre name	RD Group (national List) 16 groups i.e. Neurological diseases	N new cases diagnosed per group year 2023	N new cases diagnosed per group year 2024	N RD patients resident per group at 31.12.2023	N RD patients NON-resident in the region per group at 31.12.2023	N therapeutic plans per RD group year 2023	N therapeutic plans per RD group year 2024

Each region sent the requested data to the MoH and received the 1st part of the NRD Plan2 financing

Data evaluation is ongoing at the national level for the 2nd tranche transfer to regions

Allocation at the regional level has occurred based on the N of pts dg and follow-up + ERN activities

Background –Italy RD scenario

Since 2001 national legislative framework

- RD List (ICD logic-not a nosological one)
- RD care network
- Registries

Interregional/regional RD population-based Registries
Registration IT-based linked to benefits for patients (high coverage)

Established data Flow (HCPs---Regional level---National level)
Core Dataset annually sent to RNMR-ISS (National Institute of Health)

2 National RD Plan - State-regions agreement on the RD national network
RD Ref Centres
RD HCP full-members ERNs
Coordinating Centre (regional)

At the local level (regional-interregional) the information supports the patients' care pathways
RD centres (hospital hubs)-local health services (pharm services-health districts-homecare)

2 National RD Plan: RD coding through ORPHAcodes
Already implemented in some areas
the National RD Plan envisages expanded ORPHAcode use
(i.e. starting with selected pilot groups of RDs)

Monitored population: 5.9 million inhabitants

Network model

1. RD Ref Centres
2. RD HCP full-members of 23 ERNs
3. Coordinating Centre (regional)

IT shared information system

Collecting data on residents and non-resident RD pts

Data collection at hospital and territorial level

Linked to benefits provision

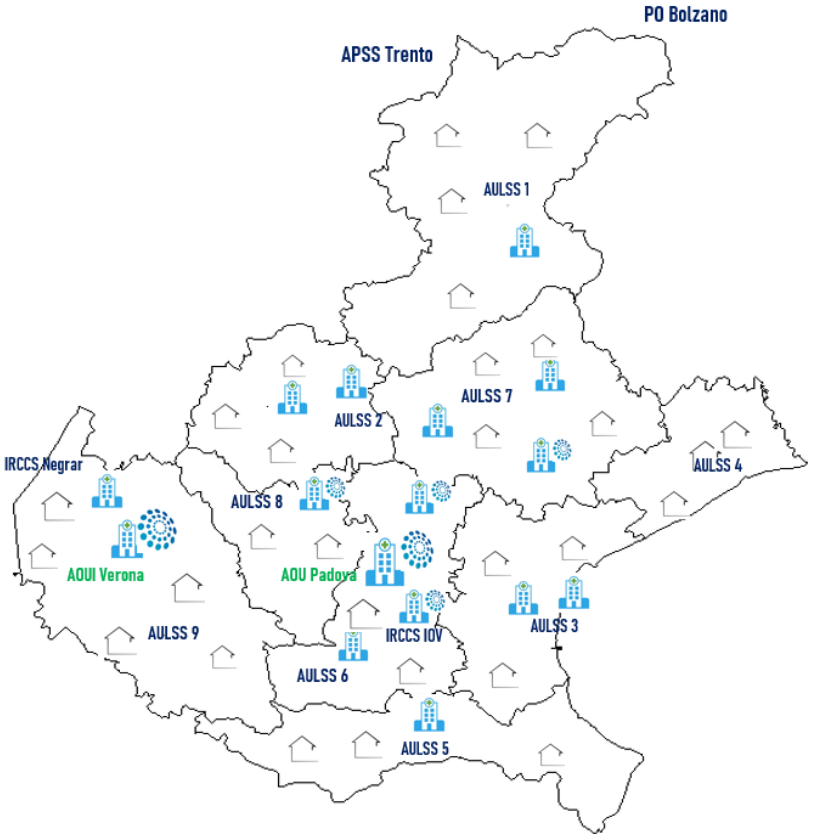
Registries ongoing since 2002

Monitored disease entities: | ICD | Omim | ORPHAcodes in use since 2007 in some regional RD Registries

Interoperability with the regional AdminHealthDB

Exhaustive population coverage (RD Centres primary source of data + local health services for RD patients followed outside the monitored area)

Since 2020 (RD Italian list + Orphanet-ERN)



ERNs in the National Health Care System- RD scenario

- ERN officially recognized in the Italian RD Legislative framework
  - -Law 175 speaks about ERN
  - -RD reorder document – 3 levels of centers: Ecxcellence (ERN); hub and spoke

- National Rare Disease Plan (PNMR) 2

- New National Rare disease Committee created, building the new PNMR3

- ERN in the National Committee on RD which includes all relevant stakeholders (Regions, AIFA, Eurordis, National allinace..)
  - coordinated by the MoH
  - ERN in the Decree and National Table for Thalassemias and Hemoglobinopathies with MoH, Regions, Experts and patients' associations

ORPHAcodes use for the coding of rare diseases: comparison of the accuracy and cross country comparability

ORPHACODES

This study tested the use of ORPHAcodes, a codification system for rare diseases, in five European countries/regions across different data sources from January 2019 to September 2021. ORPHAcodes were found to be a versatile resource for coding RDs which assure easiness of use and inter-country comparability across population and hospital databases.

3,133 ORPHAcodes collected

- Czech Republic
- Malta
- Romania
- Spain
- Veneto Region-Italy

53.6%

of ORPHAcodes described very low prevalence diseases (<1 case per million)

83.4%

of ORPHAcodes described disease entities more precisely than corresponding ICD-10 codes

- 31.6% rare developmental defects during embryogenesis
- 17.6% rare neurological diseases
- 9.2% inborn errors of metabolism

82.2% of ORPHAcodes were the disease/subtype of the disease aggregation level of the Orphanet classification

Group
Disorder
Subtype

RESEARCH Open Access

ORPHAcodes use for the coding of rare diseases: comparison of the accuracy and cross country comparability

Monica Mazzucato[1*], Laura Visonà Dalla Pozza[1], Paola Facchin[1], Cèline Angin[2], Francis Agius[3], Clara Cavero-Carbonell[4], Virginia Corrochano[5], Katerina Hanusova[6], Kurt Kirch[7], Deborah Lambert[8], Caterina Lucano[9], Sylvie Maiella[9], Monica Panzaru[10], Cristina Rusu[10], Stefanie Weber[7], Oscar Zurriaga[4], Miroslav Zvolsky[6] and Ana Rath[9*]

Angin *et al. Orphanet Journal of Rare Diseases* (2024) 19:28
https://doi.org/10.1186/s13023-024-03030-2

Orphanet Journal of Rare Diseases

RESEARCH Open Access

Coding undiagnosed rare disease patients in health information systems: recommendations from the RD-CODE project

Céline Angin[1*], Monica Mazzucato[2], Stefanie Weber[3], Kurt Kirch[3], Waed Abdel Khalek[4], Houda Ali[4], Sylvie Maiella[4], Annie Olry[4], Anne-Sophie Jannot[1,5] and Ana Rath[4]

Funded by the European Union

Network of Hospitals in 26SM
24 ERN Virtual consultation, research and training

ERNs

JARDIN

Joint Action JARDIN

ERN integration into national health care systems

Veneto region (AE) representing Interregional RD Board

The Italian Rare Diseases Network at the intersection of the regional, national and European dimension: a descriptive and regression analysis (submitted)

Snapshot of the Italian RD network following the reorganization required by the second National Plan for Rare Diseases (PNMR2).
Data was analysed from 275 hospitals and 18 regional networks, multivariate logistic regression models to identify the structural and organizational predictors of participation in RD networks.

WP5 Governance

coordinated by (IRCSS IOR (NCA) & AOU Pisa
Task 5.1

THANK YOU!

European Reference Network | Hematological Diseases (ERN EuroBloodNet)

Funded by the European Union