EuRREB Quality Manager
Dr. Ana Priego Zurita

Leiden University Medical Center

‘5 years of European Registries of Rare Endocrine and Bone Conditions’

EuRREB
European Registries for Rare
Endocrine and Bone conditions

Five Years of the European Registries for Rare Endocrine and Bone Conditions

Ana Priego Zurita

Quality Manager EuRREB
Leiden University Medical Center

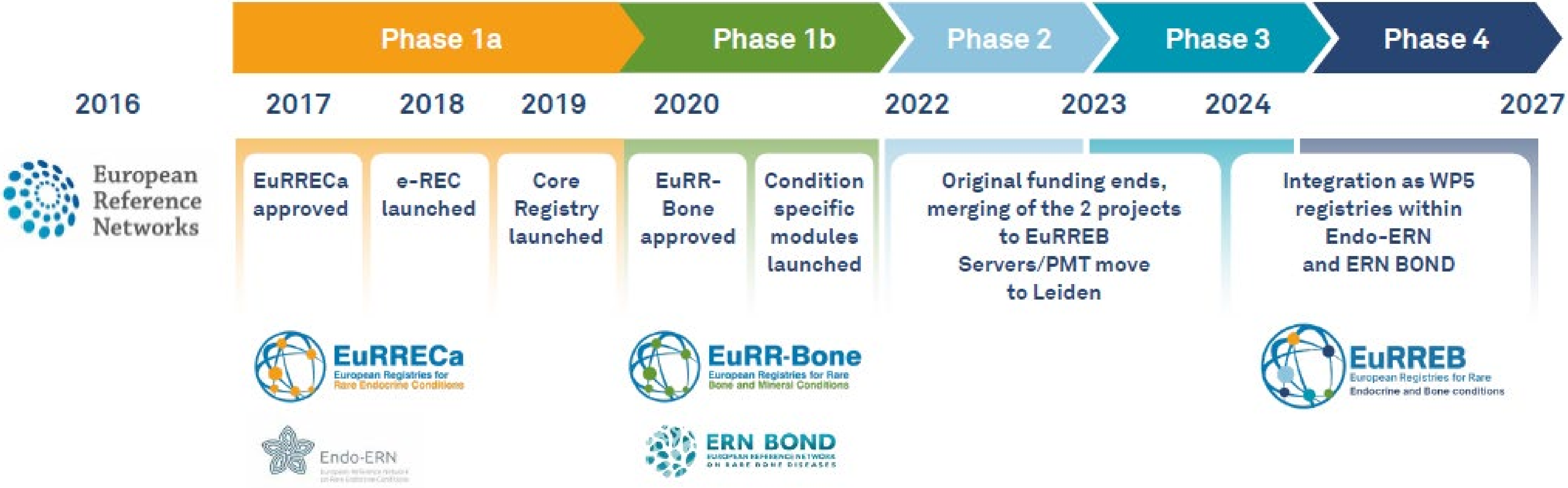
EuRREB
European Registries for Rare
Endocrine and Bone conditions

Funded by
the European Union

Why Registries Matter?

- Low prevalence of rare endocrine and bone conditions lead to fragmented knowledge
- Registries allow collection of standardised clinical information on rare conditions
- Could bring the patient voice to service planners
- Registries connect clinicians, researchers, and patients
- Enhance visibility of specialist centres, condition and other organisations

The Evolution of EuRRECa and EuRR-Bone to EuRREB



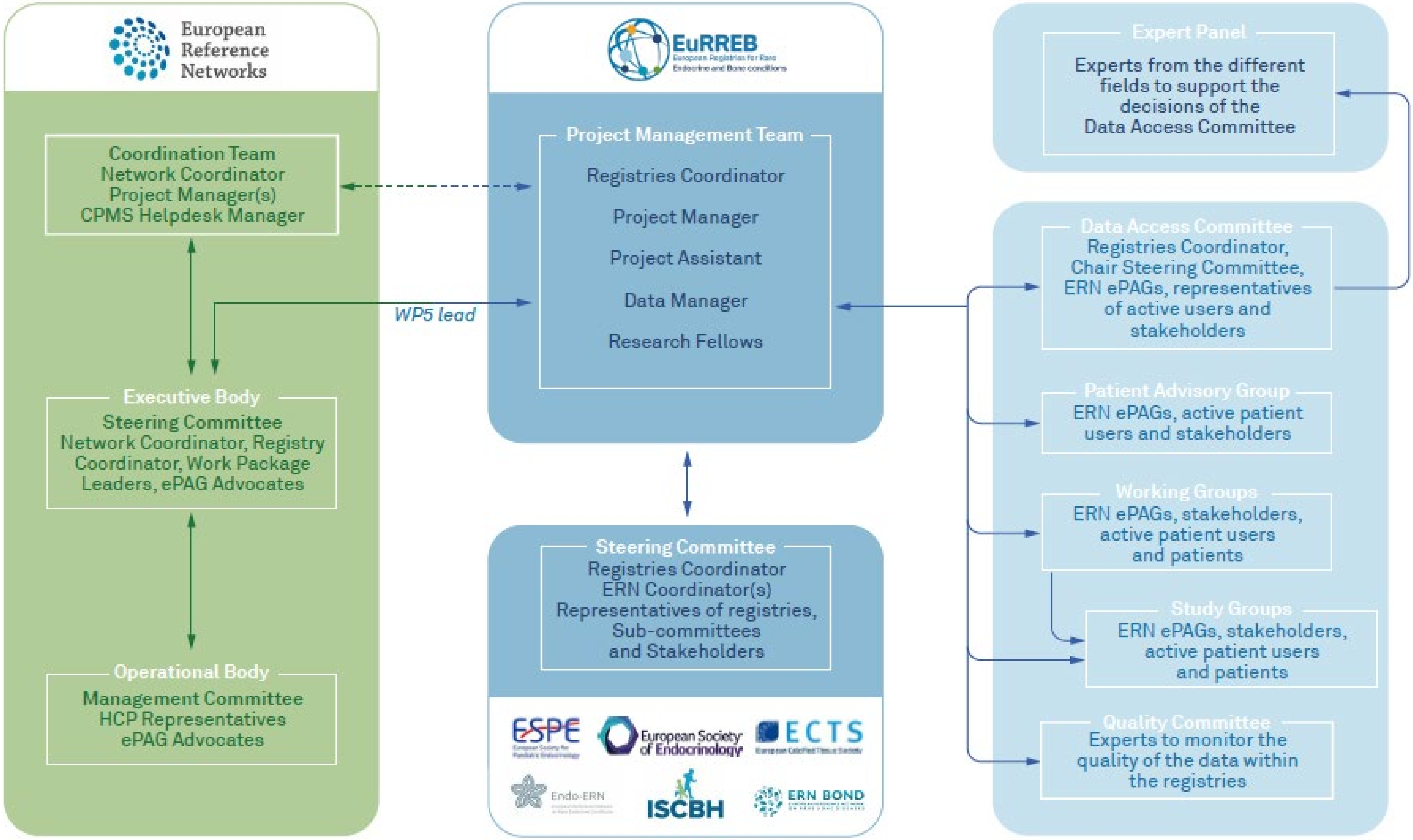
- 2017: EuRRECa approved
- 2018: e-REC launched
- 2019: Core Registry launched
- 2020: EuRR-Bone approved
- Condition specific modules launched
- 2022: Original funding ends, merging of the 2 projects to EuRREB Servers/PMT move to Leiden
- 2024: Integration as WP5 registries within Endo-ERN and ERN BOND

European Registries for Rare Endocrine conditions (EuRRECa)

European Registries for Rare Bone conditions (EuRR-Bone)

European Registries for Rare Endocrine and Bone conditions

EuRREB Registries Governance



European Reference Networks

Coordination Team
Network Coordinator
Project Manager(s)
CPMS Helpdesk Manager

Executive Body
Steering Committee
Network Coordinator, Registry Coordinator, Work Package Leaders, ePAG Advocates

Operational Body
Management Committee
HCP Representatives
ePAG Advocates

WP5 lead

EuRREB
European Registries for Rare Endocrine and Bone conditions

Project Management Team
Registries Coordinator
Project Manager
Project Assistant
Data Manager
Research Fellows

Steering Committee
Registries Coordinator
ERN Coordinator(s)
Representatives of registries,
Sub-committees
and Stakeholders

ESPE
European Society of Endocrinology
ECTS
Endo-ERN
ISCBH
ERN BOND

Expert Panel
Experts from the different fields to support the decisions of the Data Access Committee

Data Access Committee
Registries Coordinator,
Chair Steering Committee,
ERN ePAGs, representatives of active users and stakeholders

Patient Advisory Group
ERN ePAGs, active patient users and stakeholders

Working Groups
ERN ePAGs, stakeholders, active patient users and patients

Study Groups
ERN ePAGs, stakeholders, active patient users and patients

Quality Committee
Experts to monitor the quality of the data within the registries

The Registries in a Nutshell

Patient encounter with any rare endocrine or bone related condition

First Encounter

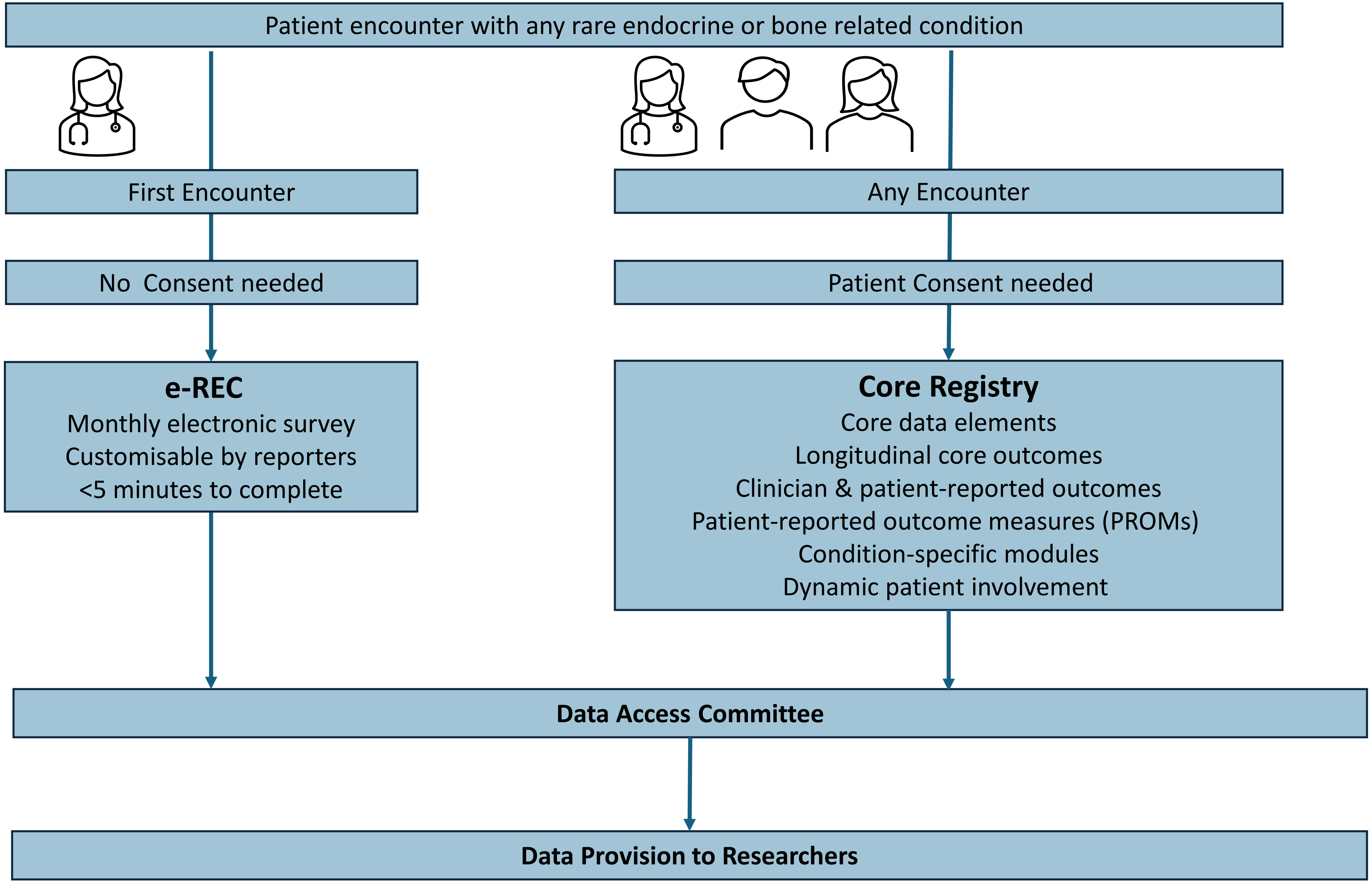
No Consent needed

e-REC
Monthly electronic survey
Customisable by reporters
<5 minutes to complete

Data Access Committee

Data Provision to Researchers

The Registries in a Nutshell



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Core Registry

Core data elements

Longitudinal core outcomes

Clinician & patient-reported outcomes

Patient-reported outcome measures (PROMs)

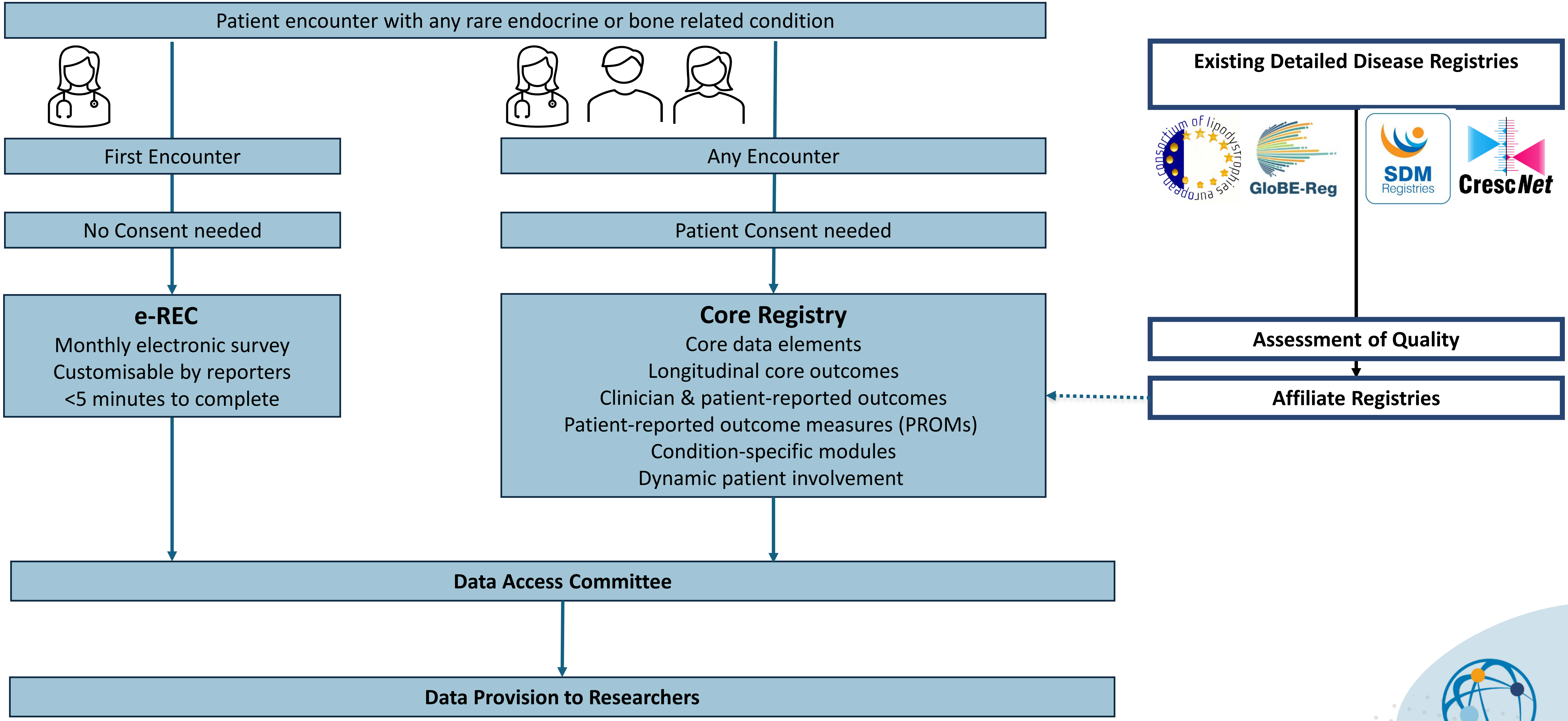
Condition-specific modules

Dynamic patient involvement

Data Access Committee

Data Provision to Researchers

The Registries in a Nutshell



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Existing Detailed Disease Registries

european consortium of lipodystrophies

GloBE-Reg

SDM Registries

CrescNet

Assessment of Quality

Affiliate Registries

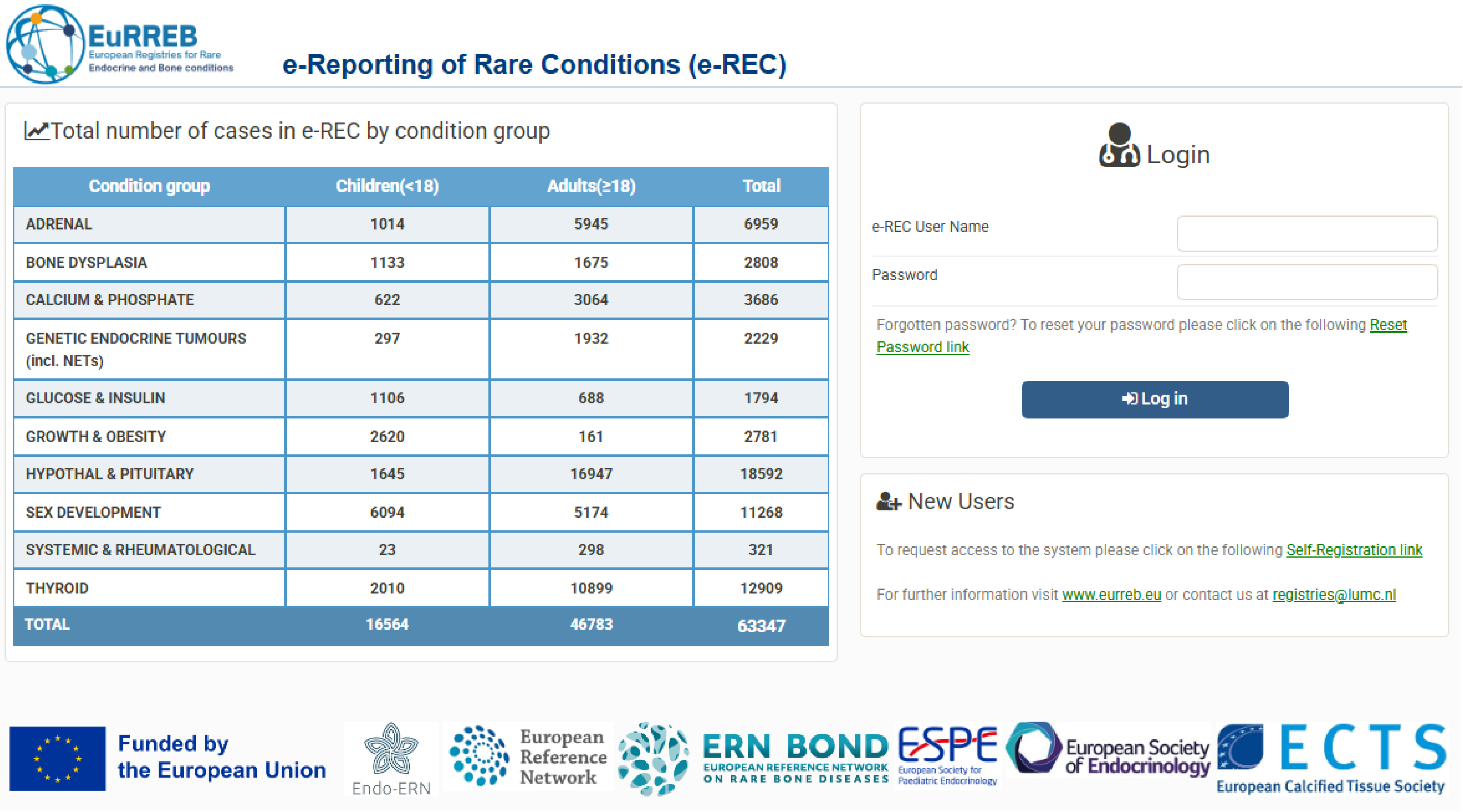
Data Access Committee

Data Provision to Researchers

e-REC

EuRREB
European Registries for Rare
Endocrine and Bone conditions

Live Case Count



EuRREB European Registries for Rare Endocrine and Bone conditions

e-Reporting of Rare Conditions (e-REC)

Total number of cases in e-REC by condition group

Condition group	Children(<18)	Adults(≥18)	Total
ADRENAL	1014	5945	6959
BONE DYSPLASIA	1133	1675	2808
CALCIUM & PHOSPHATE	622	3064	3686
GENETIC ENDOCRINE TUMOURS (incl. NETs)	297	1932	2229
GLUCOSE & INSULIN	1106	688	1794
GROWTH & OBESITY	2620	161	2781
HYPOTHAL & PITUITARY	1645	16947	18592
SEX DEVELOPMENT	6094	5174	11268
SYSTEMIC & RHEUMATOLOGICAL	23	298	321
THYROID	2010	10899	12909
TOTAL	16564	46783	63347

Login

e-REC User Name

Password

Forgotten password? To reset your password please click on the following Reset Password link

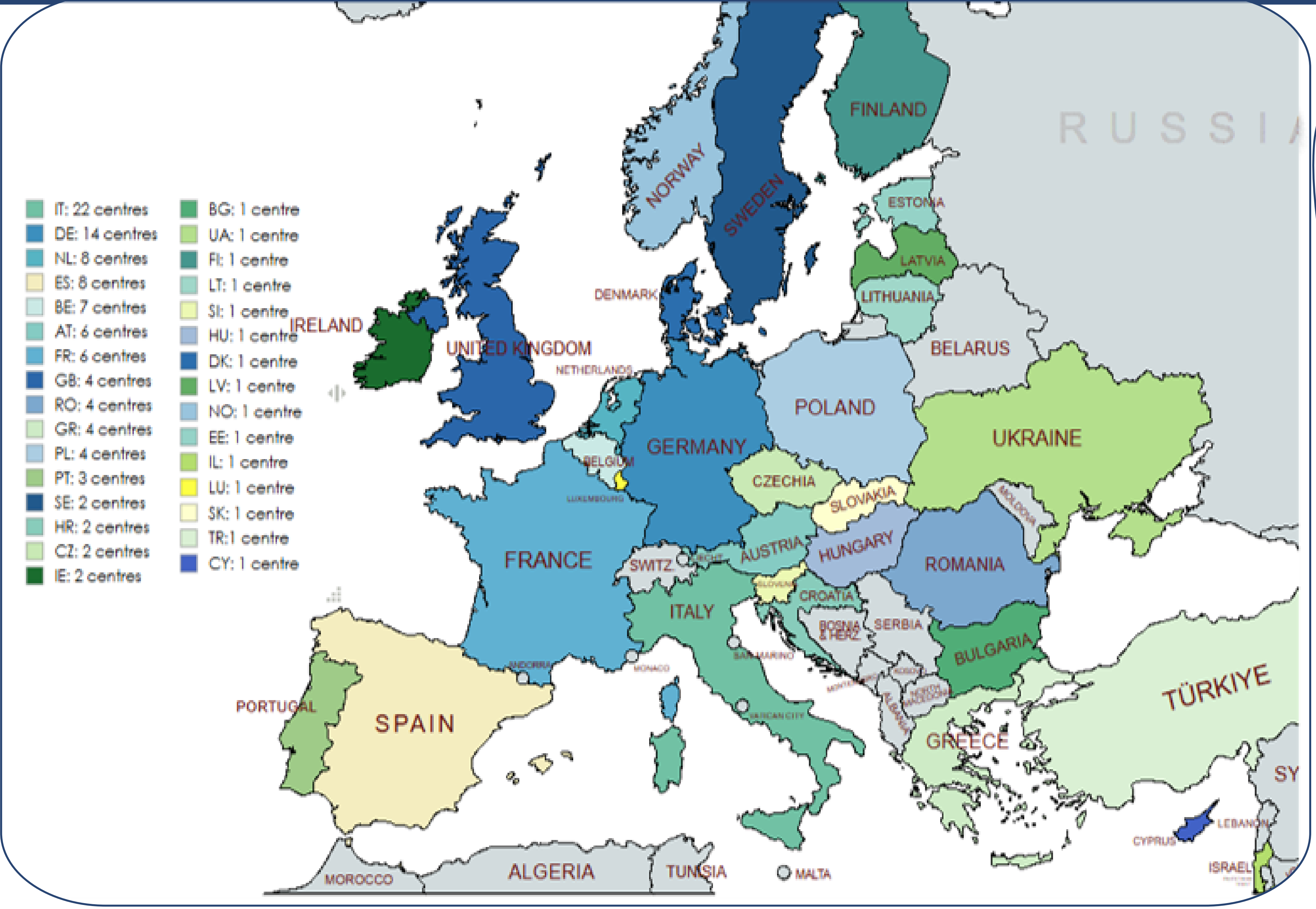
New Users

To request access to the system please click on the following Self-Registration link

For further information visit www.eurreb.eu or contact us at registries@lumc.nl

Funded by the European Union

Centres Reporting in e-REC



July 2018 – June 2025

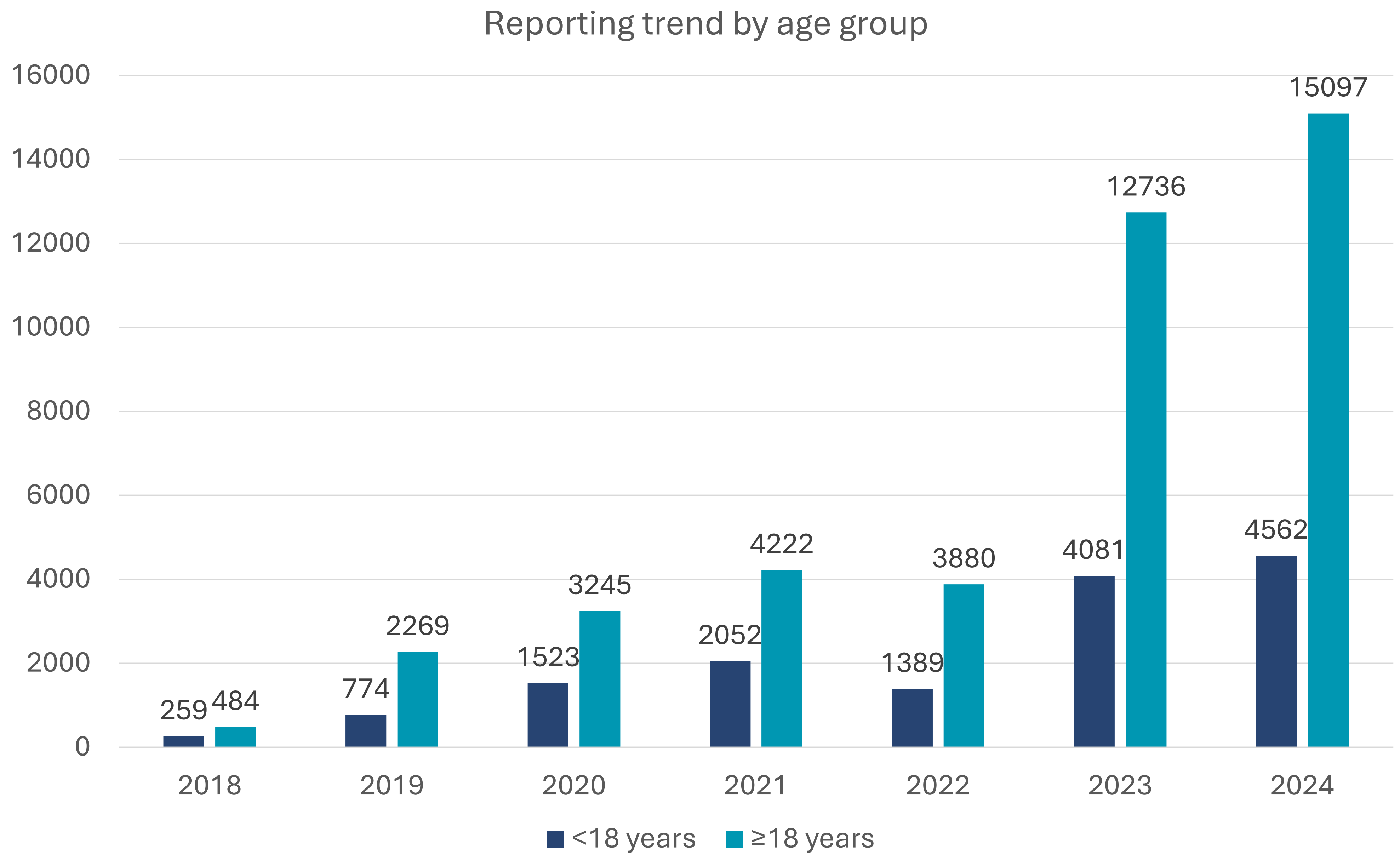
Active reporters:
116 centres from 32 countries
(5 non-EU)

HCPs ERN affiliation
Endo-ERN only - **65**
Endo-ERN and ERN-BOND - **29**
ERN-BOND only - **3**
Not affiliated to either – **19**

Number of patients in e-REC:
61683 cases
(45498 in adults, 16185 in children)

Full report at eurreb.eu

Reporting in e-REC



Full report at eurreb.eu

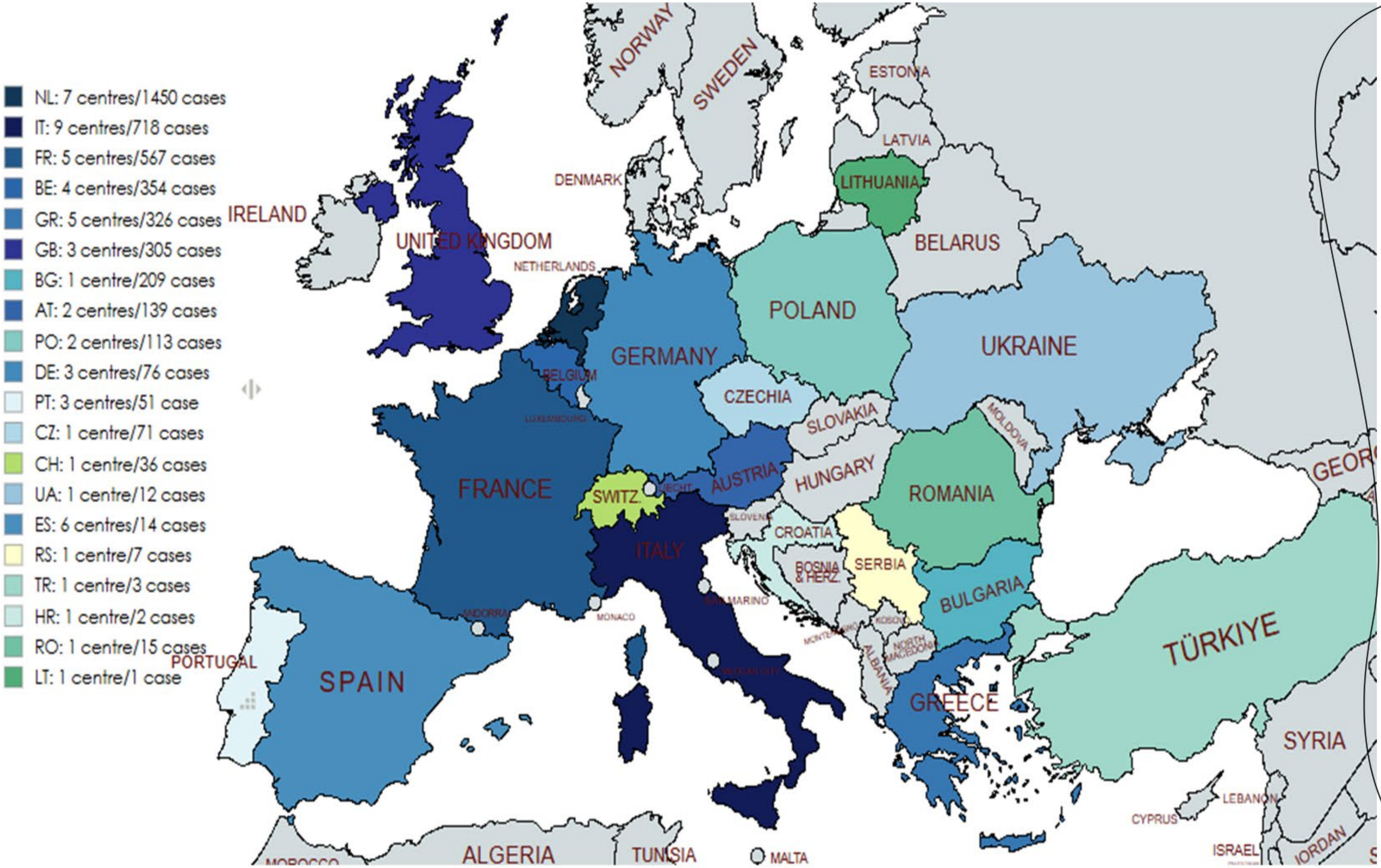
The Core Registry

EuRREB
European Registries for Rare
Endocrine and Bone conditions

Levels of Participation

	0 Consent	1 Common Data Elements	2 Generic outcomes	3 Diagnosis-specific outcomes
 **Clinician**		*Clinician enters basic data called the Common Data Elements: Date of birth, country of residemce, information on the diagnosis, etc*	*Clinician completes generic questionnaires on behalf of the patient*	*Clinician fills out condition-specific modules with clinician-specific content*
 **Patient**	*Informed consent from the patient*		*Patient completes questionnaires to assess, for example, quality of life or how pain or fatigue may impact their lives*	*Patient fills out condition-specific modules with patient-specific content*

Centres Reporting in the Core Registry



Oct 2019 – June 2025

60 centres from 22 countries (6 non-EU)

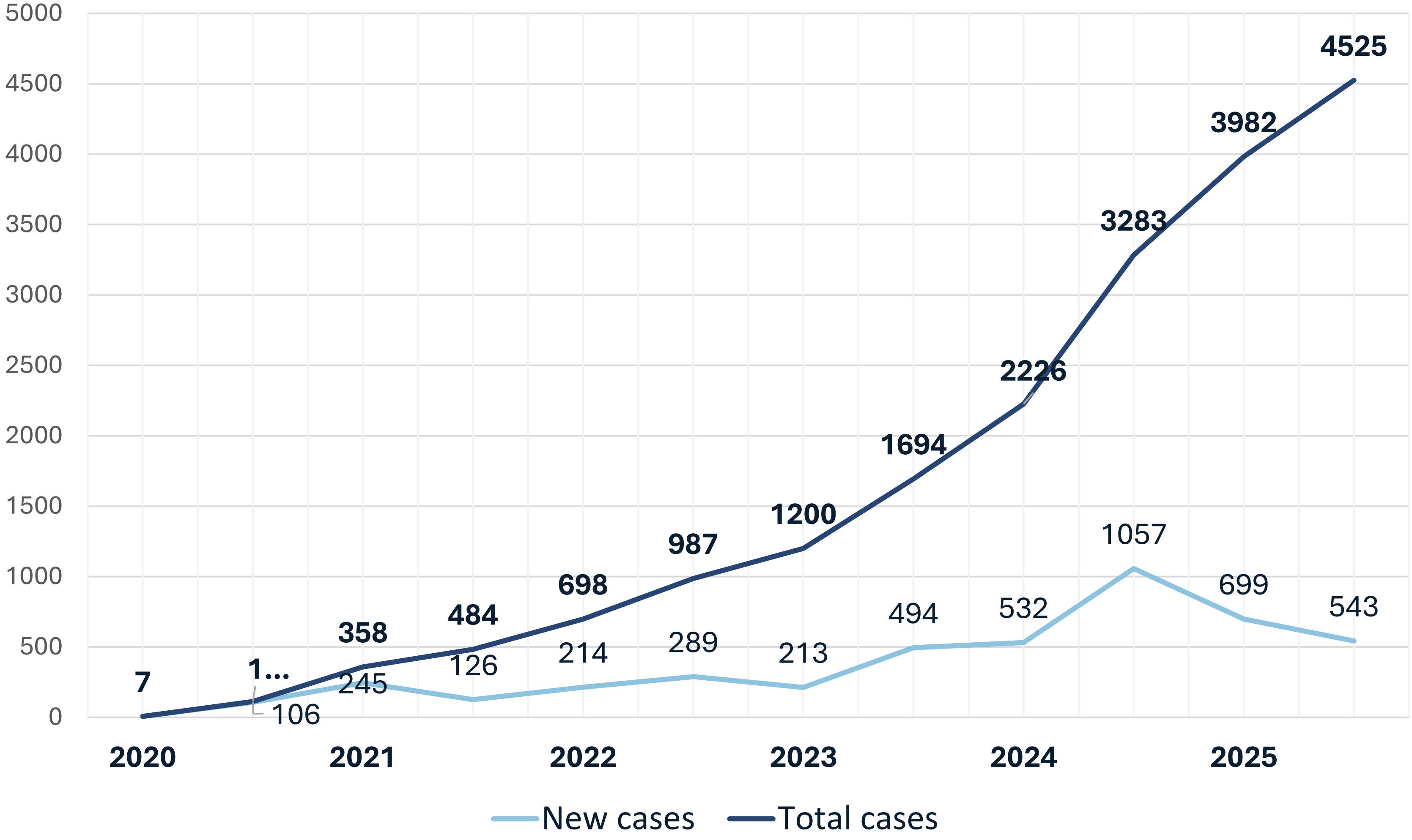
<u>HCPs ERN affiliation</u>
Endo-ERN only 19
Endo-ERN and ERN BOND 17
ERN BOND only 4
Not affiliated to either 20

Number of patients in the Core registry: 4480

Full report at eurreb.eu

Reporting Trends

Patient Records in the Core Registry



Where Are We

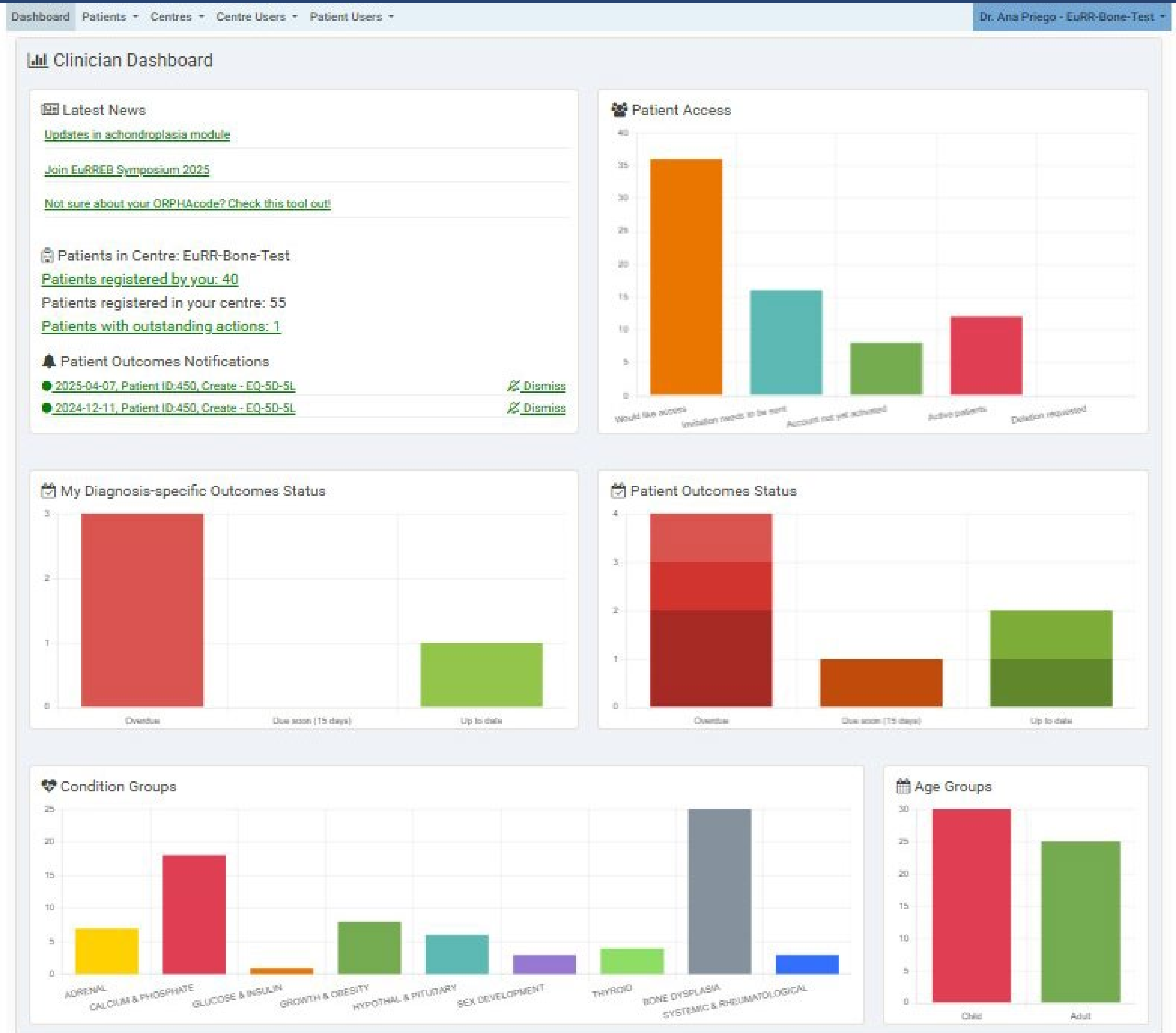
EuRREB
European Registries for Rare
Endocrine and Bone conditions

Achievements for Clinicians

- Harmonised, high-quality data collection tools
- Access to working and study groups
- Access to condition-specific modules
- Network
- Authorship

The Core Registry Dashboard

- Latest news
- Overview of patients in the centre
- Acces to patients with outstanding actions
- Notifications and access to outcomes completed by patients



Clinician Dashboard

Latest News

Updates in achondroplasia module

Join EuRREB Symposium 2025

Not sure about your ORPHAcode? Check this tool out!

Patients in Centre: EuRR-Bone-Test

Patients registered by you: 40

Patients registered in your centre: 55

Patients with outstanding actions: 1

Patient Outcomes Notifications

● 2025-04-07, Patient ID:450, Create - EQ-5D-5L Dismiss

● 2024-12-11, Patient ID:450, Create - EQ-5D-5L Dismiss

Patient Access

My Diagnosis-specific Outcomes Status

Patient Outcomes Status

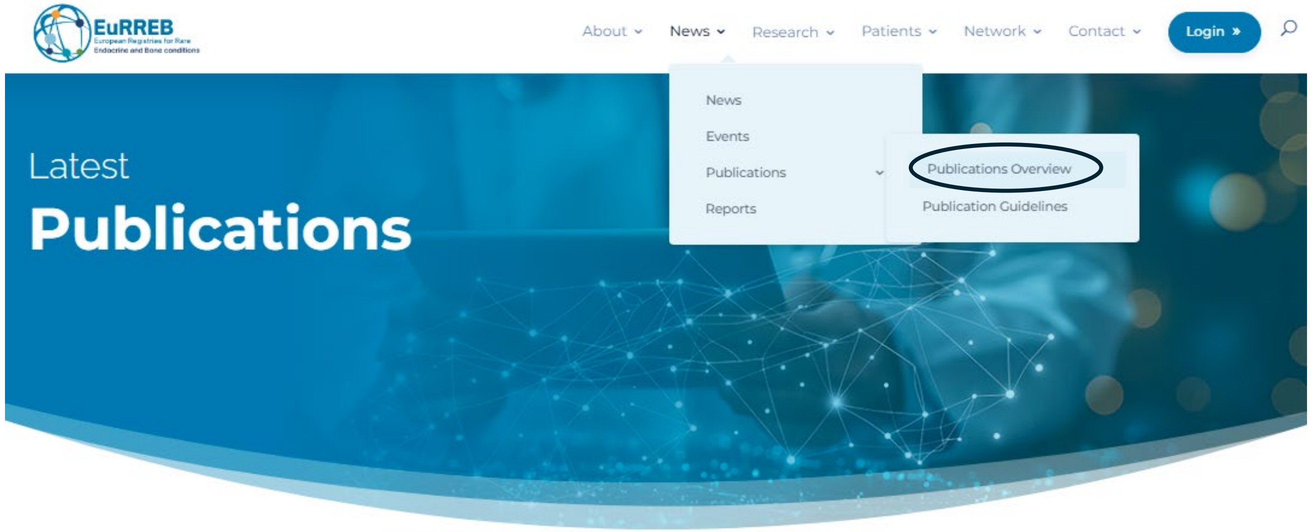
Condition Groups

Age Groups

- Patient access status
- Clinician and patient outcomes status
- Distribution by MTG and age group

Achievements for Researchers

- **Data request submitted 36 / data requests approved 35**



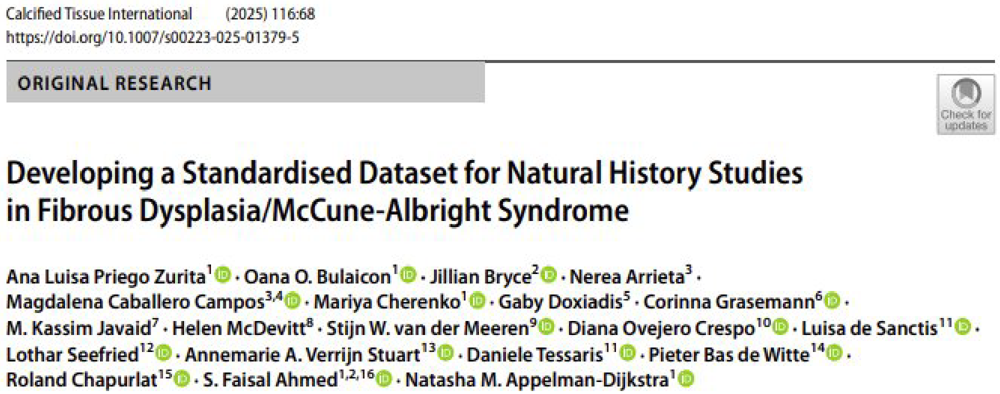
Appelman-Dijkstra *et al.*
Orphanet Journal of Rare Diseases (2025) 20:331
https://doi.org/10.1186/s13023-025-03862-6

Orphanet Journal of
Rare Diseases

RESEARCH **Open Access**

Developing a standard dataset in the European registries for rare endocrine and bone conditions—a Melorheostosis dataset

Natasha M. Appelman-Dijkstra[1*], Mariya Cherenko[1], Gavin P. R. Clunie[2], Thomas Funck-Brentano[3], Corinna Grasemann[4], Adalbert Raimann[5], Willem F. Lems[6] and Martine Cohen-Solal[3]



Calcified Tissue International (2025) 116:68
https://doi.org/10.1007/s00223-025-01379-5

ORIGINAL RESEARCH

Developing a Standardised Dataset for Natural History Studies in Fibrous Dysplasia/McCune-Albright Syndrome

Ana Luisa Priego Zurita[1] · Oana O. Bulaicon[1] · Jillian Bryce[2] · Nerea Arrieta[3] · Magdalena Caballero Campos[3,4] · Mariya Cherenko[1] · Gaby Doxiadis[5] · Corinna Grasemann[6] · M. Kassim Javaid[7] · Helen McDevitt[8] · Stijn W. van der Meeren[9] · Diana Ovejero Crespo[10] · Luisa de Sanctis[11] · Lothar Seefried[12] · Annemarie A. Verrijn Stuart[13] · Daniele Tessaris[11] · Pieter Bas de Witte[14] · Roland Chapurlat[15] · S. Faisal Ahmed[1,2,16] · Natasha M. Appelman-Dijkstra[1]

Condition-Specific Modules and Active Studies

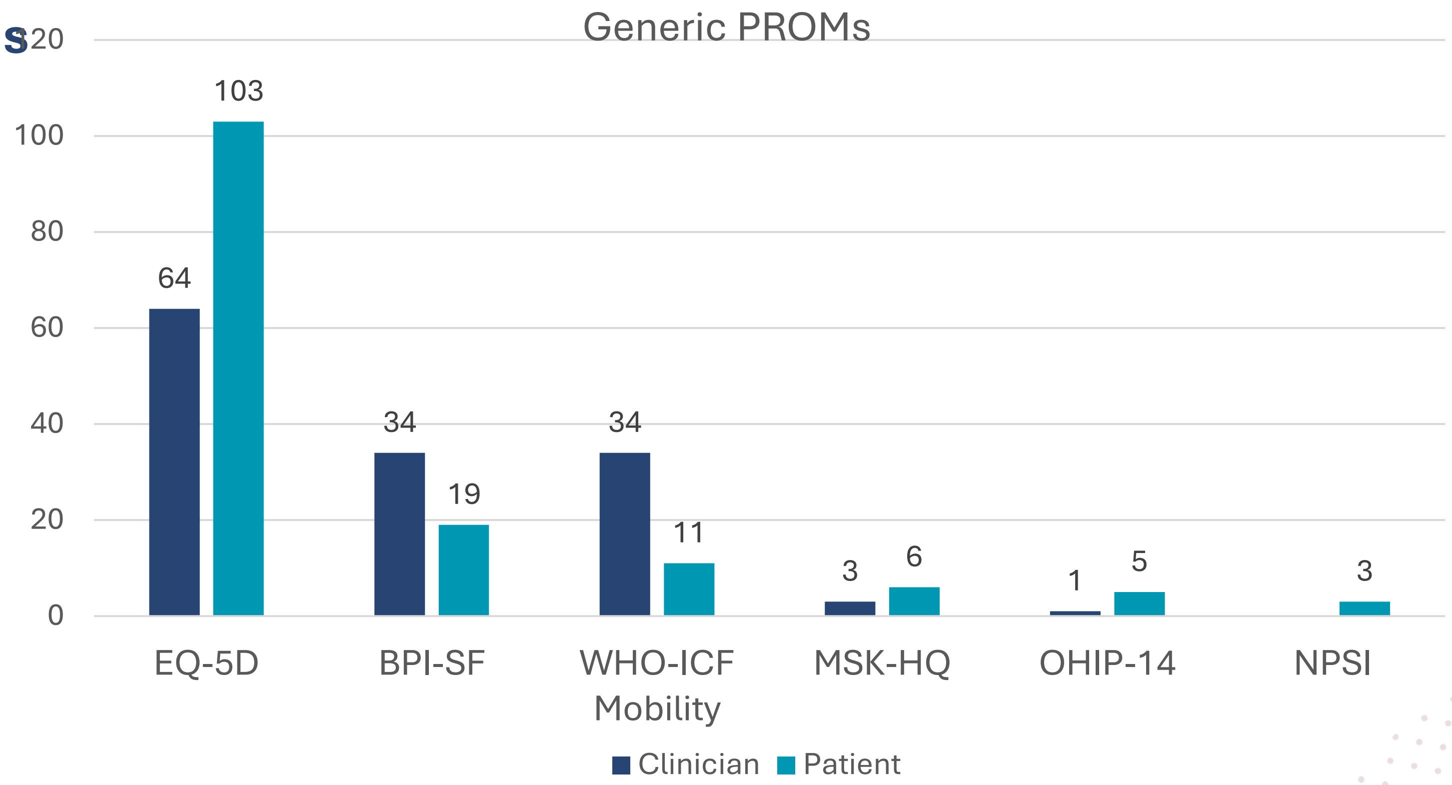
CSM	MTG	Year	N patients in CR	Outcomes completed/ N patients	N active countries	N active centers	Study group leads	Active study
iPPSD/PHP	MTG2	2021	36	12/13	3	4	Agnès Linglart/Diana-Alexandra Ertl	Natural History of iPPSD/PHP
Pituitary adenoma	MTG6	2021	1081	2019/1240	6	9	Alberto Pereira	1. Epidemiological Surveillance of Pituitary Tumours within Endo-ERN 2. The Clinical Usefulness of the PANOMEN 3 Grade Score 3. Cell-lineage specific differences in presentation and outcomes of non-functioning pituitary adenomas – a multicentre study in patients seen at Endo-ERN reference centres
Achondroplasia	BD	2022	193	133/114	4	4	Klaus Mohnike/ Ines Alves	Natural History of Achondroplasia
FD/MAS	BD	2022	713	1433/543	6	7	Natasha Appelman-Dijkstra	Natural History of FD/MAS
Osteogenesis imperfecta	BD	2022	185	76/66	6	6	Wolfgang Hoegler	Under development
Parathyroid carcinoma	MTG2	2022	14	15/13	3	3	Maria Luisa Brandi	Natural History of Parathyroid Carcinoma
Rare hypophosphataemia	MTG2	2023	92	66/38	5	5	Agnès Linglart/Diana-Alexandra Ertl	Under development
Melorheostosis	BD	2023	20	19/11	2	2	Martine Cohen-Solal	Natural History of Melorheostosis
Rare obesity	MTG5	2023	106	11/11	2	2	Erica van den Akker	Natural History of Rare Obesity
Gender Incongruence	MTG7	2023	328	326/320	4	5	Martine Cools/Silvia Ciancia	Current status of healthcare for transgender adolescents across Europe
Paediatric differentiated thyroid carcinoma	MTG8	2024	107	96	8	12	Hanneke van Santen/ Sarah Clement	Natural History of Paediatric DTC
STUDY: Transition outcomes in pituitary patients	MTG6	2025	98	Closed survey			Savi Shishkov	Transition outcomes in patients with pituitary conditions during transition age

Condition-Specific Modules under Development

CSM	MTG	Year	N patients in CR	Outcomes completed	Study group leads	Active study
Langerhans Cell Histiocytosis	Sys	2025	14	2	Polyzois Makras	Natural History of endocrine and bone complications of LCH
Chronic Nonbacterial Osteitis (CNO)	Sys	2025	7	4	Elizabeth Winter	Under development
Pregnancy and Rare Endocrine and Bone conditions	all	2025	0	0	Tim Korevaar and Natasha Appelman-Dijkstra	Under development
Multiple Endocrine Neoplasia type 1 (MEN-1)	MTG4	2025	3	0	Frederic Castinetti, Gerlof Valk, Maria Luisa Brandi	Under development
Hypoparathyroidism	MTG2	2025	21		Maria Yavropolou, Corinna Grasemann, Eva Kassi	Under development

Achievements for Patients

- **Access to Core Registry to view their data and complete outcomes**
- **Patient representatives – governance, working and study groups**
- **Implementation of PROMs**



Challenges and Lessons Learned

Ethics & GDPR: delays and harmonisation challenges

Limited resources at centres for data entry

Importance of ongoing engagement with contributors and patients

Looking Ahead

- **Consolidate already existing modules and promote research with the collected data**
- **Develop new condition-specific modules and expand PROMs**
- **Incorporate Key Performance Indicators into EuRREB**
- **Continue to work toward FAIRification / overarching projects**
    - **ERDERA**
- **Integration with European Health Data Space**

Call to Action

- Keep contributing data
    - Voucher opportunities
    - Student grants

- Join working and study groups

- Use EuRREB data for research

Ana Priego Zurita
Quality Manager

Tess de Rooij
Project Manager

Mariya Cherenko
Data Manager

Natasha Appelman-Dijkstra
Lead

Jacqueline van der Blom
Project Assistant

Faisal Ahmed
Co- Lead

Endo-ERN
European Reference Network
on Rare Endocrine Conditions

ERN BOND
EUROPEAN REFERENCE NETWORK
ON RARE BONE DISEASES

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LUMC

Ways to contact us:

- 🌐 eurreb.eu
- ✉ registries@lumc.nl
- drop-in sessions via Zoom
- in European Registries for Rare Endocrine and Bone Conditions

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Questions?

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