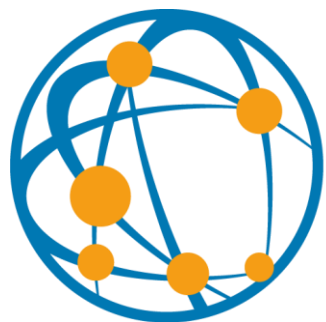


# EUROPEAN REGISTRIES FOR RARE ENDOCRINE AND BONE CONDITIONS: ELECTRONIC TOOLS FOR MAPPING AND STUDYING RARE DISEASES

Cherenko M, Priego Zurita AL, de Rooij TM, Ahmed SF, Appelman-Dijkstra NM



**EuRRECa**  
European Registries for  
Rare Endocrine Conditions



**EuRR-Bone**  
European Registries for Rare  
Bone and Mineral Conditions



Endo-ERN  
European Reference Network  
on Rare Endocrine Conditions

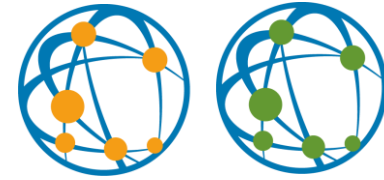


**ERN BOND**  
EUROPEAN REFERENCE NETWORK  
ON RARE BONE DISEASES



Funded by  
the European Union

# The Evolution of EuRRECa and EuRR-Bone



2016

2017

2018

2019

2020

2022

2023

2024

Endo-ERN  
ERN BOND  
approved

EuRRECa  
approved

e-REC

Core  
Registry

EuRR-  
Bone  
uses  
EuRRECa  
platform

Condition  
specific  
modules  
launched

Original  
EuRRECa  
project  
funding  
ends

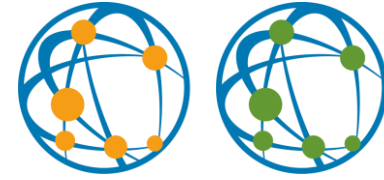
Original  
EuRR-Bone  
project  
funding  
ends

Supported  
by Endo-  
ERN & ERN  
BOND



Transfer of  
Registries  
from Glasgow  
to Leiden

Project  
Management Team  
and ICT team  
transferred to  
Leiden



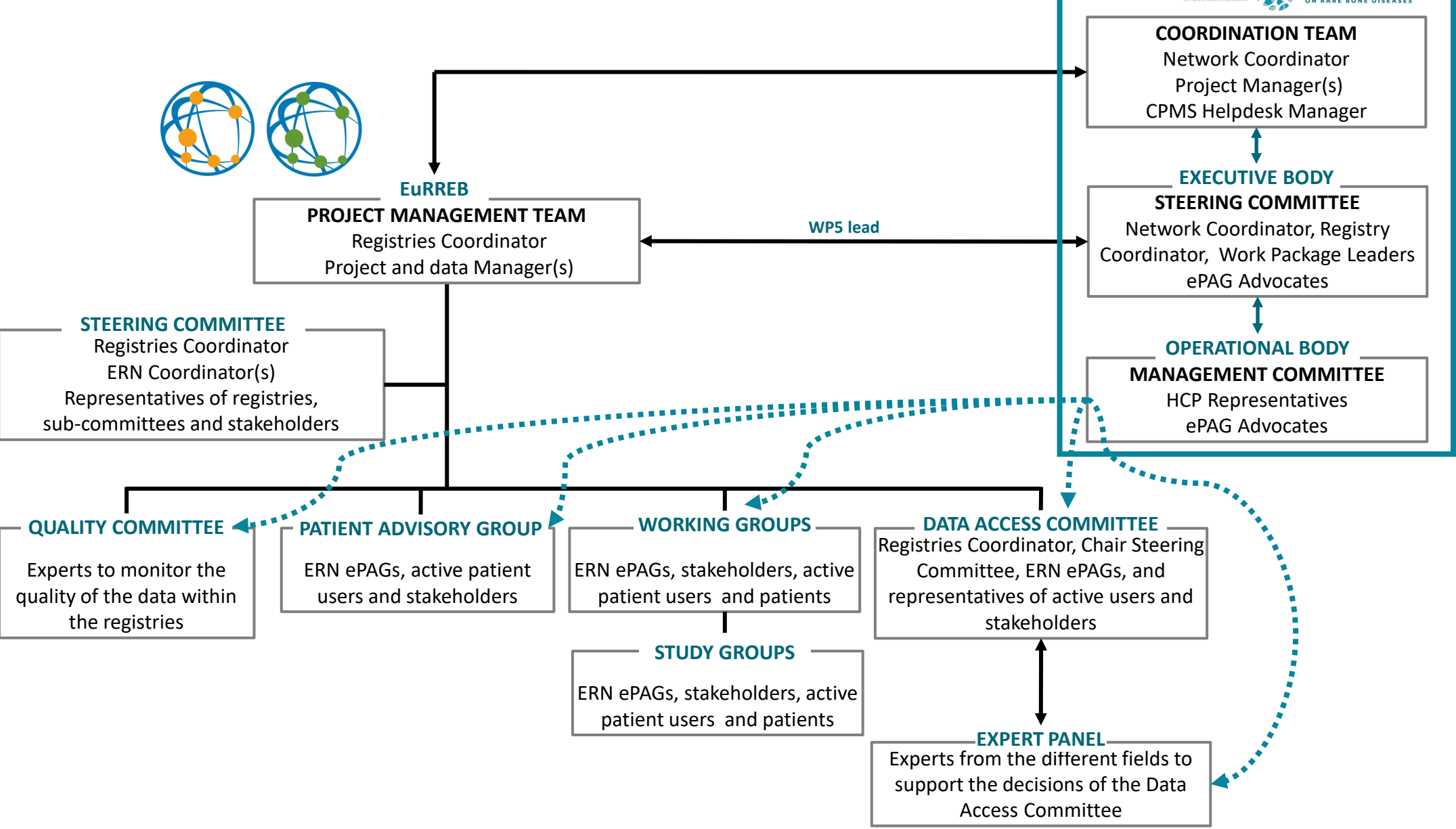
## Welcome

EuRREB – the European Registries for Rare Endocrine and Bone conditions consists of [EuRRECa](#) and [EuRR-Bone](#). The registries aim to maximize the opportunity for all patients, healthcare professionals, and researchers to participate and use high quality, patient-centred registries for rare endocrine and bone/mineral conditions. We work closely together with [Endo-ERN](#) and [ERN BOND](#) and are supported by patients, researchers, clinicians, [scientific societies](#), [patient societies](#) to build this database.

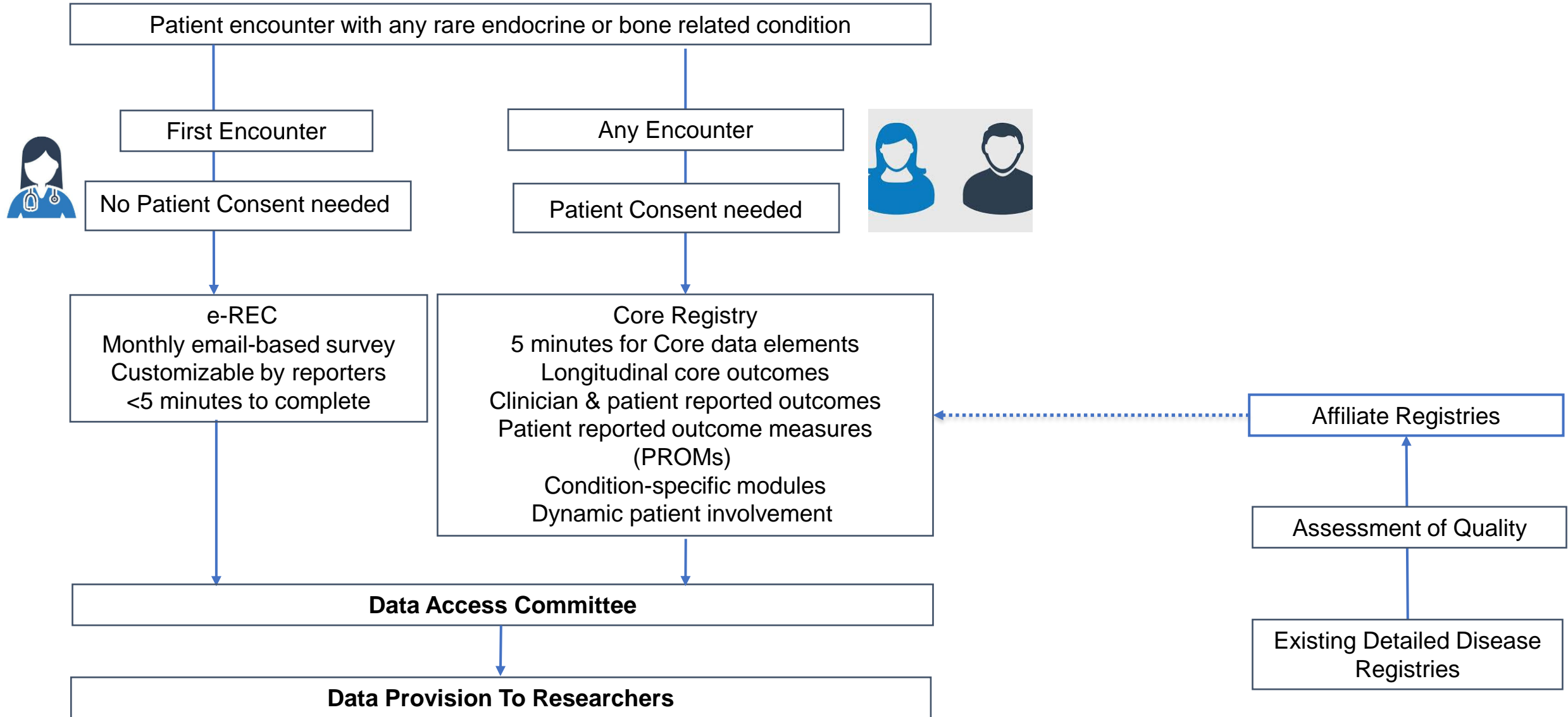
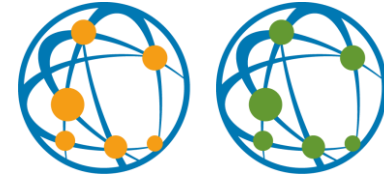
## Upcoming Events

- MAY 10** **Joint Congress of ESPE and ESE 2025**  
May 10<sup>th</sup> - May 13<sup>th</sup>, 2025
- SEP 11** **ERN BOND General Assembly**  
September 11<sup>th</sup>, 2025
- SEP 12** **EuRREB Symposium 2025**  
September 12<sup>th</sup>, 2025

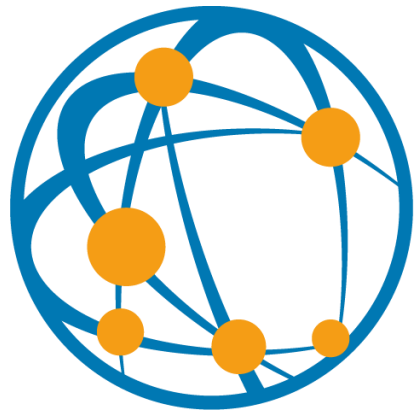
# EuRREB Registries Governance Year 2024



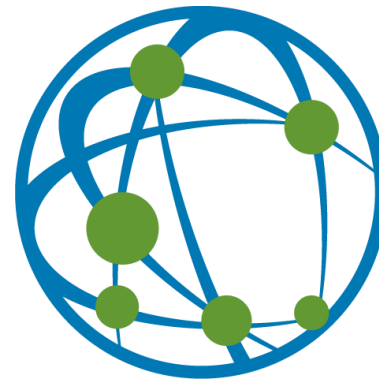
# The Registries in a Nutshell



e-REC



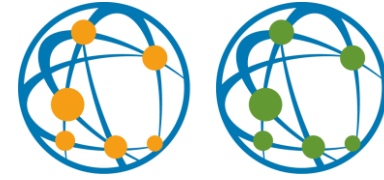
**EuRRECa**  
European Registries for  
Rare Endocrine Conditions



**EuRR-Bone**  
European Registries for Rare  
Bone and Mineral Conditions



# e-REC: Live Case Count



## e-Reporting of Rare Conditions (e-REC)



Returns Reporting Setup Centre Reporters Centre Users Return Periods Reporting Centres Users Pending Approvals Reference Data ▾

📈 Total number of cases in e-REC by condition group

Condition group	Children(<18)	Adults(≥ 18)	Total
ADRENAL	778	3905	4683
BONE DYSPLASIA	768	1019	1787
CALCIUM & PHOSPHATE	454	2074	2528
GENETIC ENDOCRINE TUMOURS (incl. NETs)	232	1359	1591
GLUCOSE & INSULIN	862	440	1302
GROWTH & OBESITY	1156	120	1276
HYPOTHAL & PITUITARY	1204	11952	13156
SEX DEVELOPMENT	5058	3738	8796
SYSTEMIC & RHEUMATOLOGICAL	0	1	1
THYROID	1368	7100	8468
<b>TOTAL</b>	<b>11880</b>	<b>31708</b>	<b>43588</b>

👤 Login

e-REC User Name

Password

Forgotten password? To reset your password please click on the following [Reset Password link](#)

➔ Log in

👤+ New Users

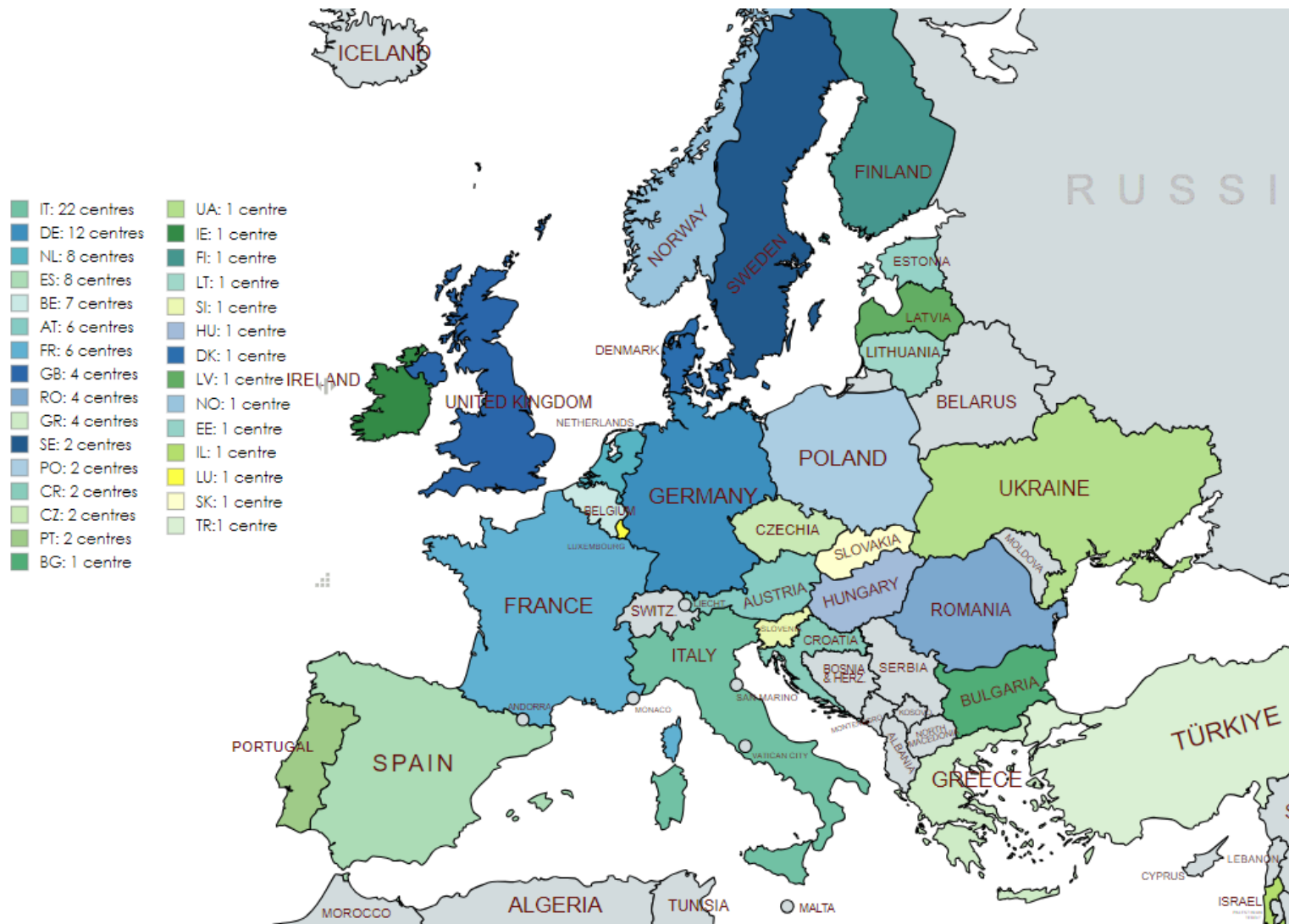
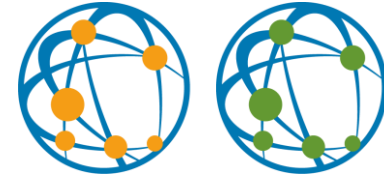
To request access to the system please click on the following [Self-Registration link](#)

For further information visit [www.eurreb.eu](http://www.eurreb.eu) or contact us at [registries@lumc.nl](mailto:registries@lumc.nl)



This website is part of the project '777215 / EuRECa' and '946831 / Eurr-Bone' which have received funding from the European Union's Health Programme (2014-2020). The content of this website cannot be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains. Project: 101156495 (Endo-ERN), Project: 101157116 (ERN BOND); EU4H-2023-ERN2-IBA-01.

# Centres Reporting in e-REC



July 2018 – October 2024

## Active reporters

107 centres from 31 countries (5 non-EU)

## HCPs ERN affiliation

Endo-ERN only - 56

Endo-ERN and ERN BOND - 29

ERN BOND only - 3

Not affiliated to either – 19

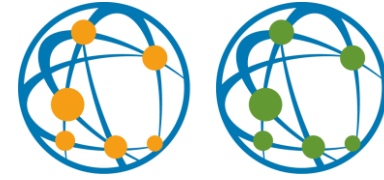
## Number of patients in e-Rec:

45476 cases

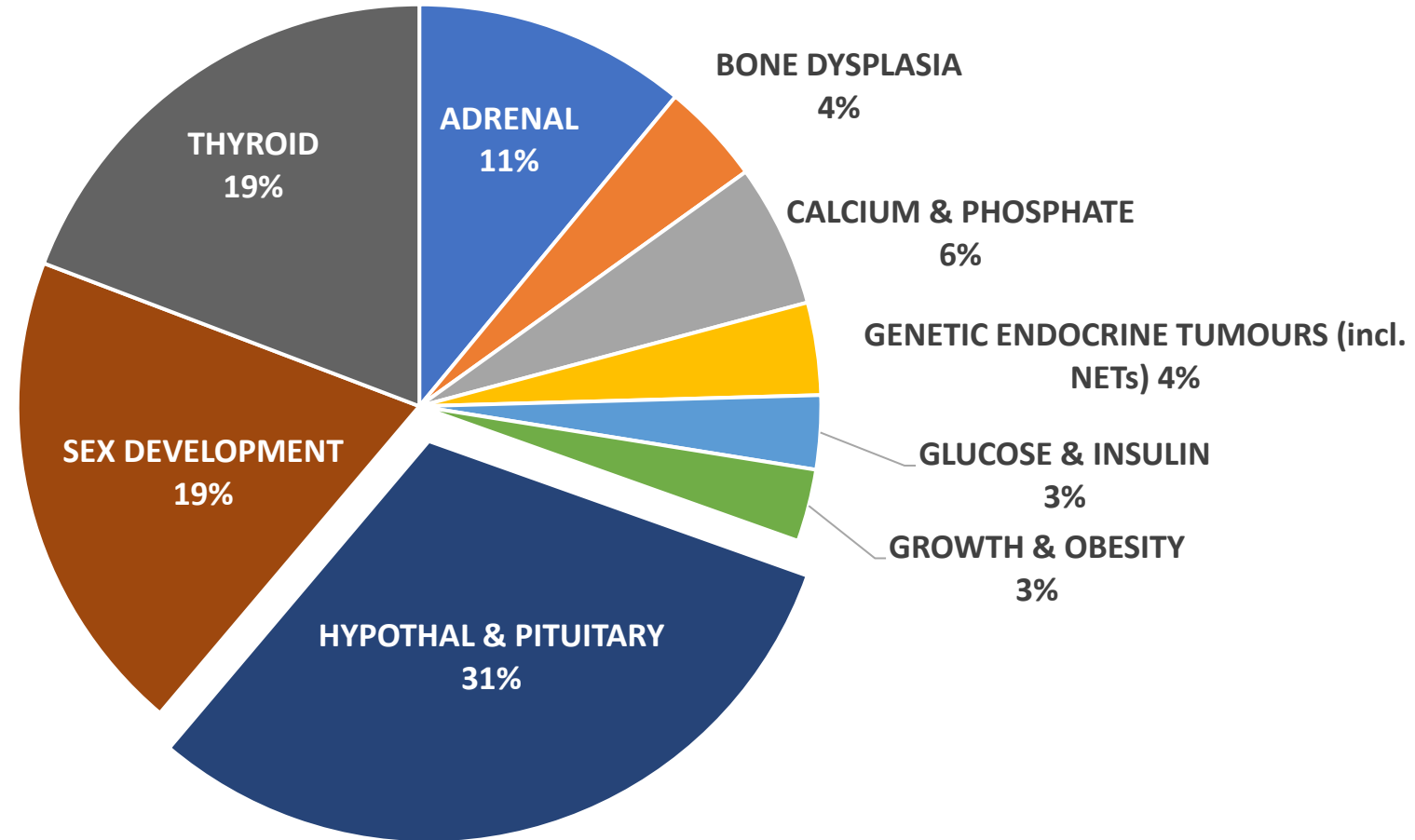
(33364 in adults, 12112 in children)

Full annual report at [eureb.eu](http://eureb.eu)

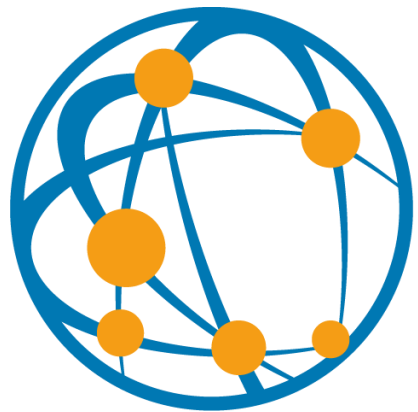




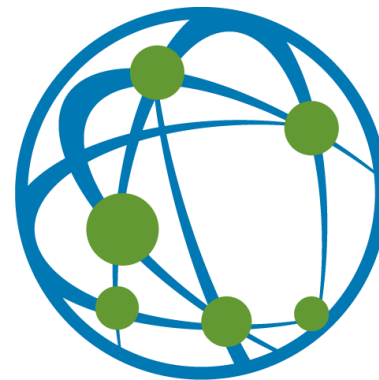
Cases (n=45476)



# Core Registry

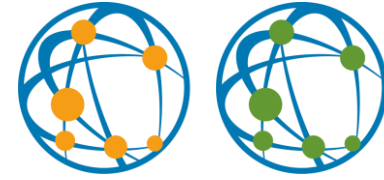


**EuRRECa**  
European Registries for  
Rare Endocrine Conditions



**EuRR-Bone**  
European Registries for Rare  
Bone and Mineral Conditions

# Levels of Participation in the Core Registry

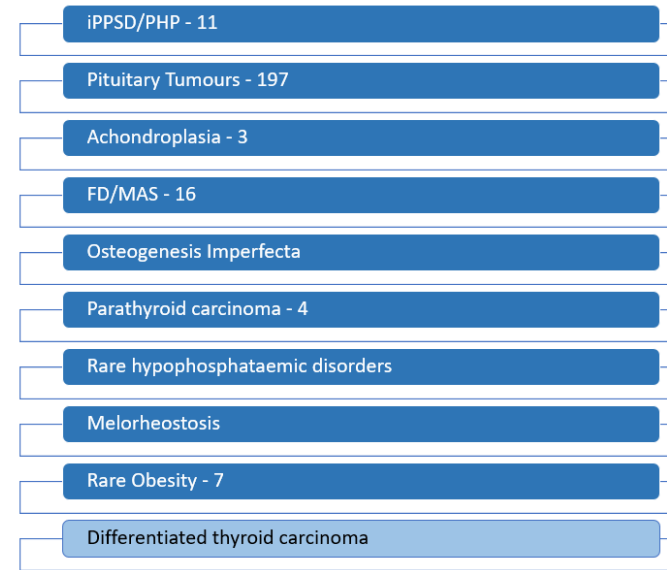
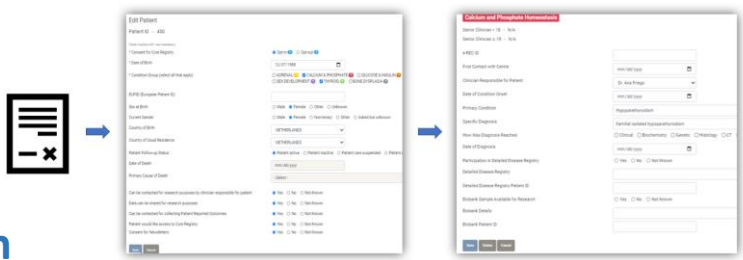


## 1 Common Data Elements

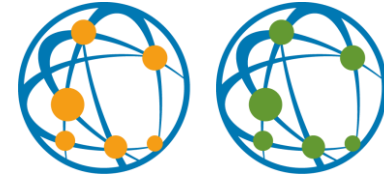
## 2 Generic outcomes

## 3 Diagnosis-specific outcomes

Clinician  
Patient



# Common Data Elements



[Edit Patient](#) ▶ Patient ID - 4165

[Edit To Do](#)

[Is Test Case](#) ?

Fields marked with \* are mandatory

\* Consent for Core Registry  Opt-in ?  Opt-out ?

\* Centre

\* Date of Birth ?

\* Condition Group (select all that apply)

ADRENAL ?  CALCIUM & PHOSPHATE ?  GLUCOSE & INSULIN ?  THYROID ?  BONE DYSPLASIA ?

Pseudonymised ID (e.g. SPIDER)

\* Sex at Birth  Male  Female  Other  Unknown

Current Gender  Male  Female  Non-binary  Other  Asked but unknown

\* Country of Birth

\* Country of Usual Residence

\* Patient Follow-up Status  Patient active  Patient inactive  Patient care suspended  Patient care

Date of Death

Primary Cause of Death

\* Can be contacted for research purposes by clinician responsible for patient  Yes  No  Not Known

\* Data can be shared for research purposes  Yes  No  Not Known

Can be contacted for collecting Patient Reported Outcomes  Yes  No  Not Known

\* Patient would like access to Core Registry  Yes  No  Not Known

Consent for Newsletters  Yes  No  Not Known

[Save and continue](#)

[Edit Patient Condition](#) ▶ Patient ID - 4165 Date of Birth - 1961-01-01

[Edit To Do](#)

**Calcium and Phosphate**

Senior Clinician < 18 - N/A  
Senior Clinician ≥ 18 - N/A

e-REC ID ?

\* First Contact with Centre ?

\* Clinician Responsible for Patient ?

\* Date of First Clinical Manifestations ?

\* Primary Condition

\* Specific Diagnosis

The full hierarchy of Conditions and Specific Diagnoses can be found in the [Core Registry Conditions Dictionary](#)

How Was Diagnosis Reached ?  Clinical  Biochemistry  Genetic  Histology  CT  MRI

\* Date of Diagnosis ?

Participation in another Detailed Disease Registry  Yes  No  Not Known

Name of Detailed Disease Registry

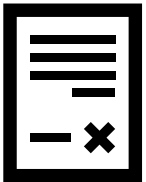
Detailed Disease Registry Patient ID

\* Biobank Sample Available for Research  Yes  No  Not Known

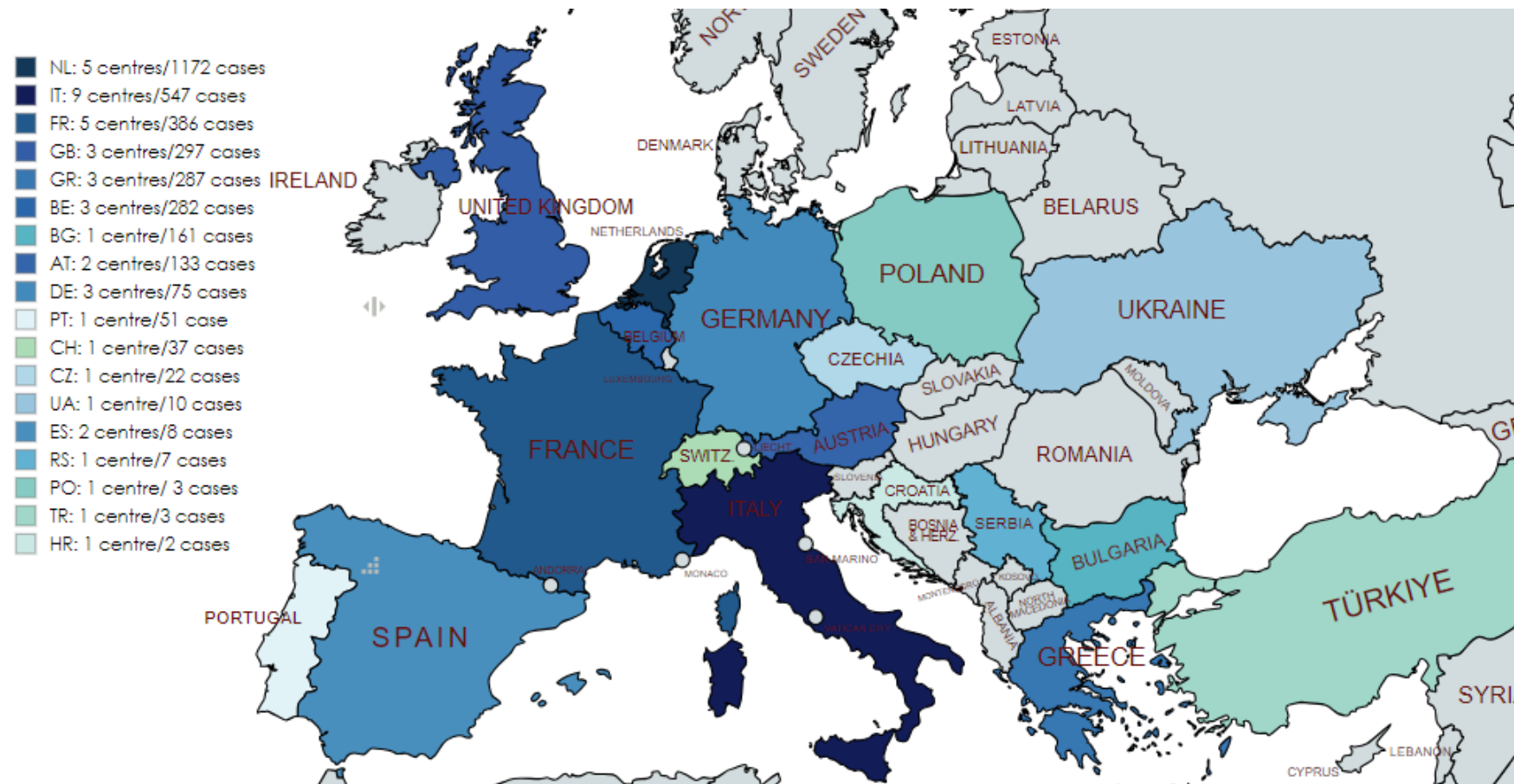
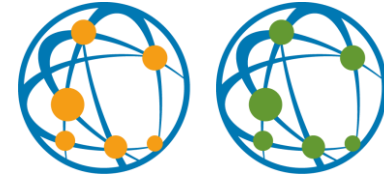
Biobank Details

Biobank Patient ID

[Save](#)



# Centres Reporting in the Core Registry



**From Oct 2019 till Oct 2024:  
48 centres from 20 countries  
(5 non-EU)**

*HCPs ERN affiliation:*

Endo-ERN only **14**

Endo-ERN and ERN BOND **14**

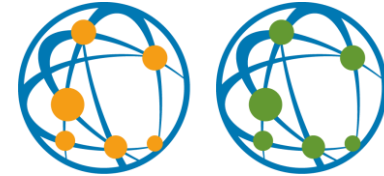
ERN BOND only **4**

Not affiliated to either **16**

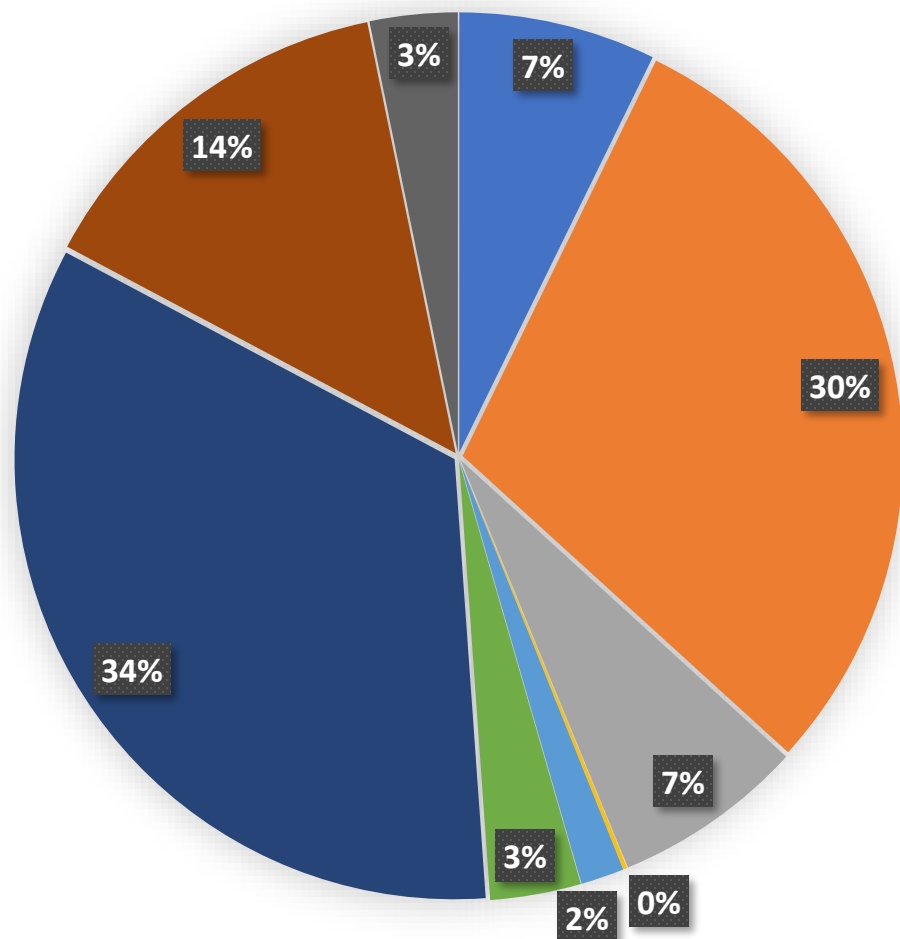
**3507 cases**

Full annual report at  
[eureb.eu](http://eureb.eu)

# Centres Reporting in the Core Registry



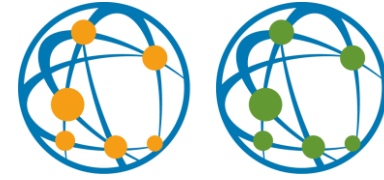
Condition groups (n=3507 cases)



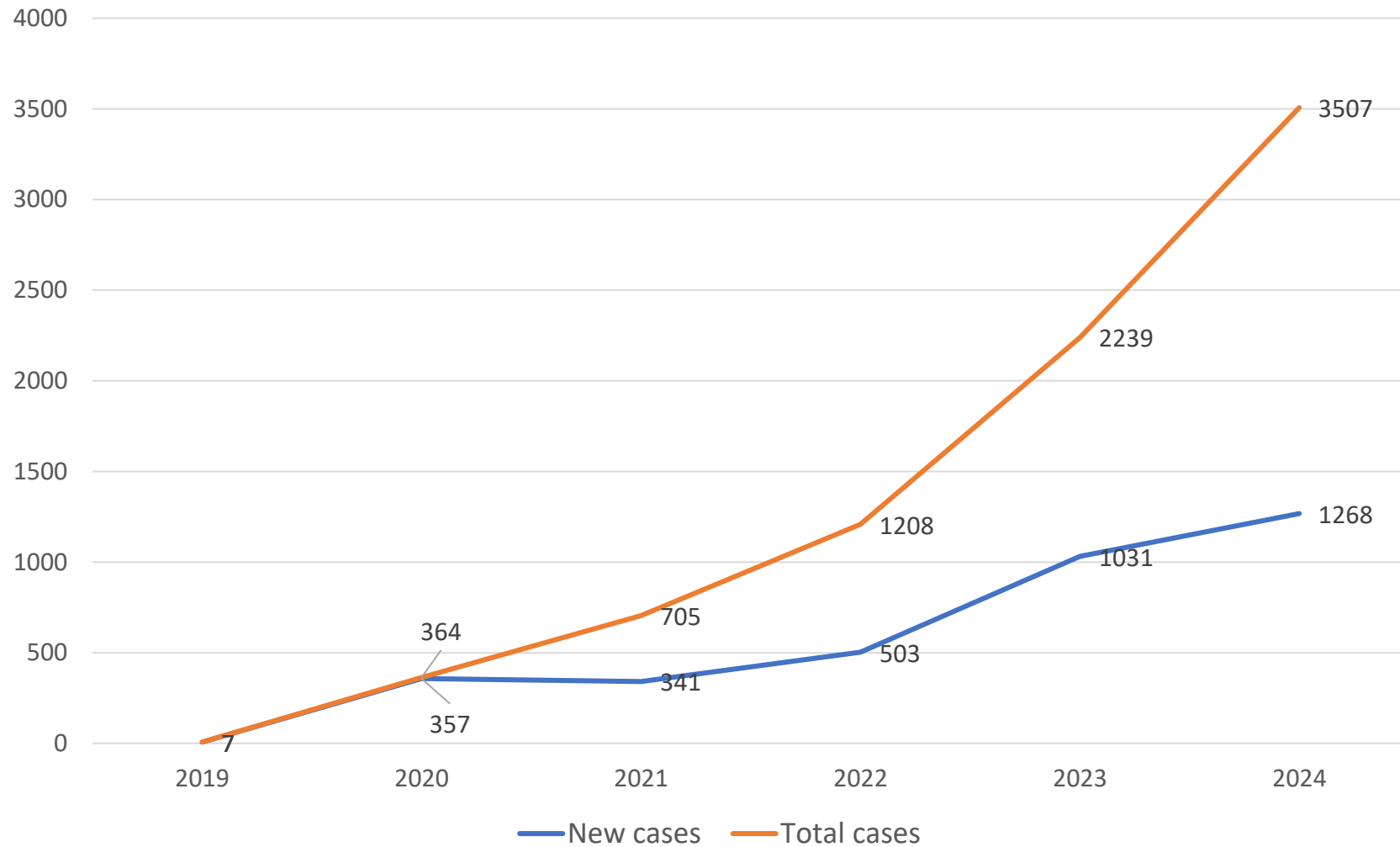
- Adrenal
- Bone Dysplasia
- Calcium and Phosphate
- Genetic Disorders of Glucose and Insulin Homeostasis
- Genetic Endocrine Tumour Syndromes
- Growth and Genetic Obesity
- Pituitary
- Sex Development and Maturation



# Reporting in the Core Registry

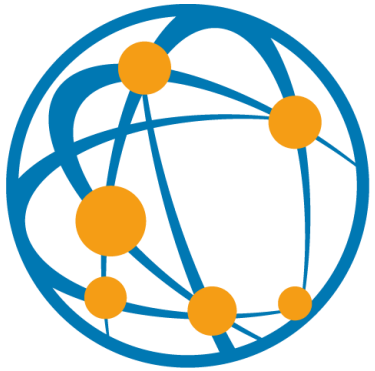


## Time trends

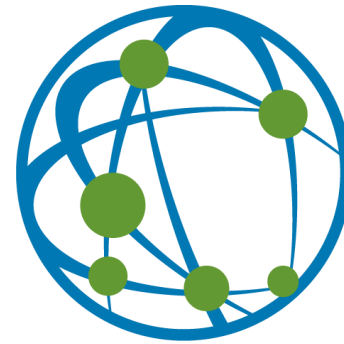


Full annual report at [eurreb.eu](http://eurreb.eu)

# Condition-Specific Outcomes within EuRREB

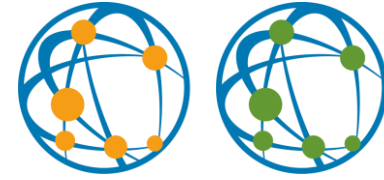


**EuRRECa**  
European Registries for  
Rare Endocrine Conditions



**EuRR-Bone**  
European Registries for Rare  
Bone and Mineral Conditions

# Developing the Condition-Specific Module



Working group



## Consensus

- Secondary survey
- Meetings
- Final dataset

## Plan

- From the dataset to the platform
- Developments

## Build

- Beta version

## Test

- Feedback
- Final changes

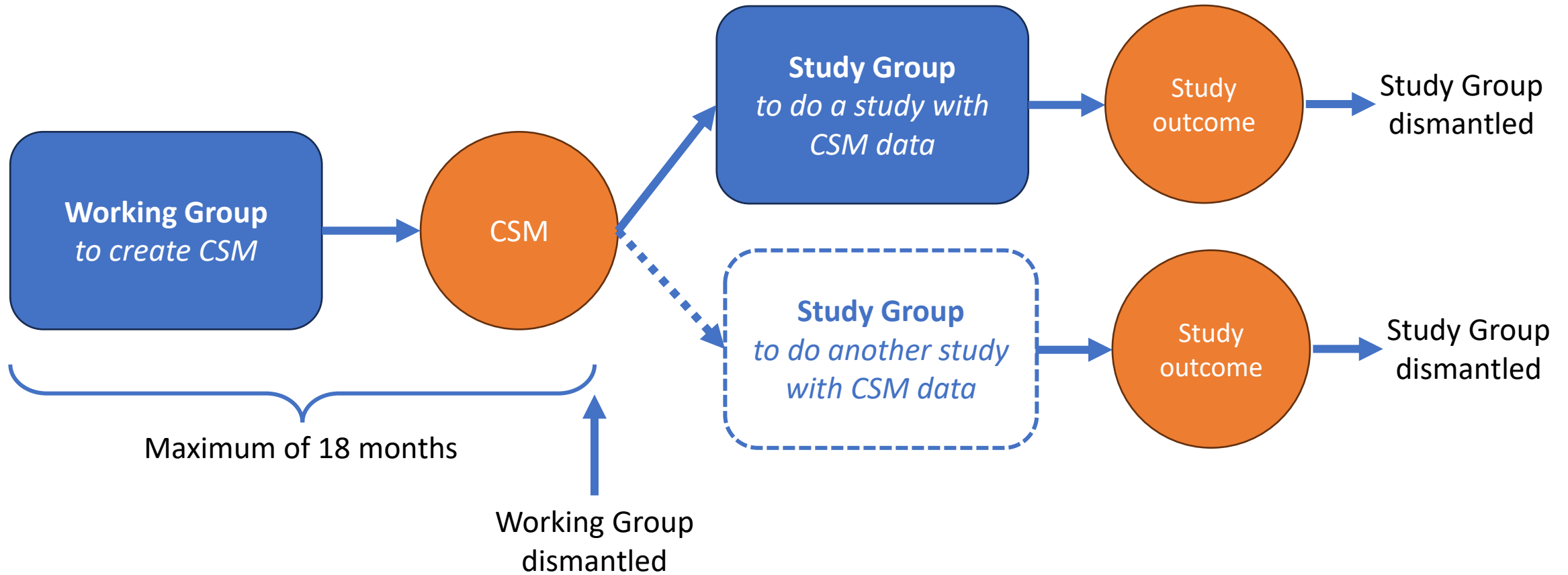
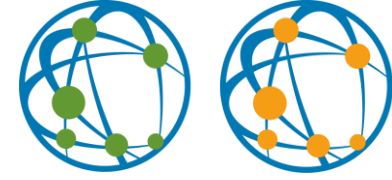
## Live version

- Available for users

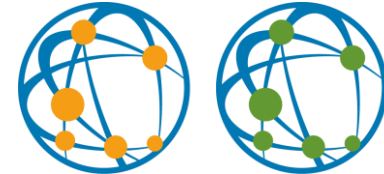
Patients

Clinicians

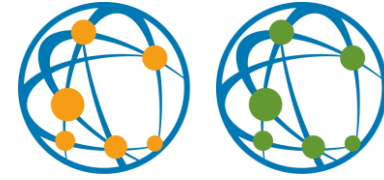
# Study Groups and Working Groups



# Condition-Specific Modules



N	CSM	MTG	Year	N patients in CR	Outcomes completed	Study group lead
1	iPPSD/PHP	MTG2	2021	32	11	Agnès Linglart/Diana-Alexandra Ertl
2	Pituitary Adenoma	MTG6	2021	739	1114	Alberto Pereira
3	Achondroplasia	BD	2022	174	125	Klaus Mohnike/ Ines Alves
4	FD/MAS	BD	2022	642	1406	Natasha Appelman-Dijkstra
5	Osteogenesis Imperfecta	BD	2022	107	26	Wolfgang Hoegler
6	Parathyroid Carcinoma	MTG2	2022	14	15	Maria Luisa Brandi
7	Rare Hypophosphataemia	MTG2	2023	84	61	Agnès Linglart/Diana-Alexandra Ertl
8	Melorheostosis	BD	2023	20	18	Luca Sangiorgi/Martine Cohen-Solal
9	Rare Obesity	MTG5	2023	68	10	Erica van den Akker
10	Gender Incongruence	MTG7	2023	152	159	Martine Cools/Silvia Ciancia
11	Pediatric Differentiated Thyroid Carcinoma	MTG8	2024	66	30	Hanneke van Santen/ Sarah Clement
12	Langerhans Cell Histiocytosis	Sys	2024/2025	0	0	Polyzois Makras
13	Chronic Nonbacterial Osteomyelitis	Sys	2024/2025	0	0	Elizabeth Winter
14	ROHHAD(NET)	MTG5	2024/2025	0	0	Mehul Dattani



## Patient Details

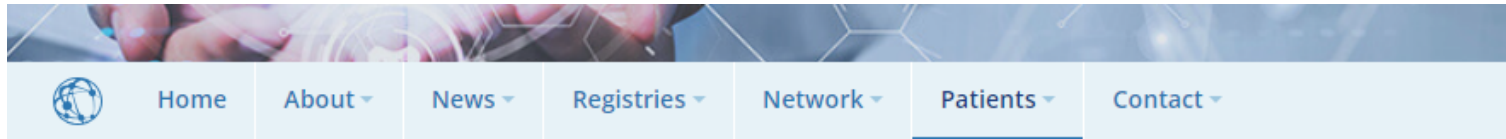
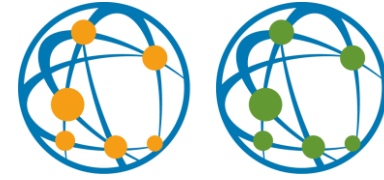
This is a copy of the data held in the Core Registry relating to your case. If you want to request changes or corrections to this data please contact the System Administrator: [registries@lumc.nl](mailto:registries@lumc.nl)

\* Some of your requested questionnaires are now due, please fill them in by following the highlighted tabs.

Patient Info	CALCIUM & PHOSPHATE	* Outcomes - CALCIUM & PHOSPHATE	SEX DEVELOPMENT	BONE DYSPLASIA	Outcomes - BONE DYSPLASIA	Centre	Patient Reported Outcomes - EQ-5D	* Patient Reported Outcomes - Generic	Settings
Patient Registry ID		4004							
Consent Type for Registry		Opt-in							
Date added to Registry		2022-06-21							
Date of Birth		1998-12-08							
Pseudonymised ID (e.g. SPIDER)									
Sex at Birth		Female							
Current Gender		Female							
Country of Birth		NETHERLANDS							
Country of Usual Residence		NETHERLANDS							
Patient Follow-up Status		Patient active							

[Edit Account Details](#)[Change Password](#)[Request Deletion of Data](#)





🏠 / Patients / Tools and Outcomes

## PROMs – Patient Reported Outcome Measures

Patient Reported Outcome Measures (PROMs) are like special surveys that help doctors understand how patients feel about their health and daily life. Since there are many different types of PROMs, it can be hard to pick the right one. At EuRREB, our team of experts and patients work together to decide which quiz is best. We also consider their cost and whether they are available in different languages.

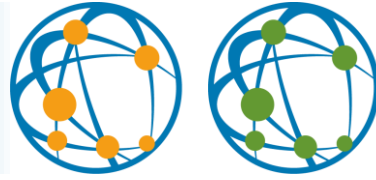
We welcome discussions about how to choose the right PROM with those interested.

Here are some of the surveys we use in our platform:

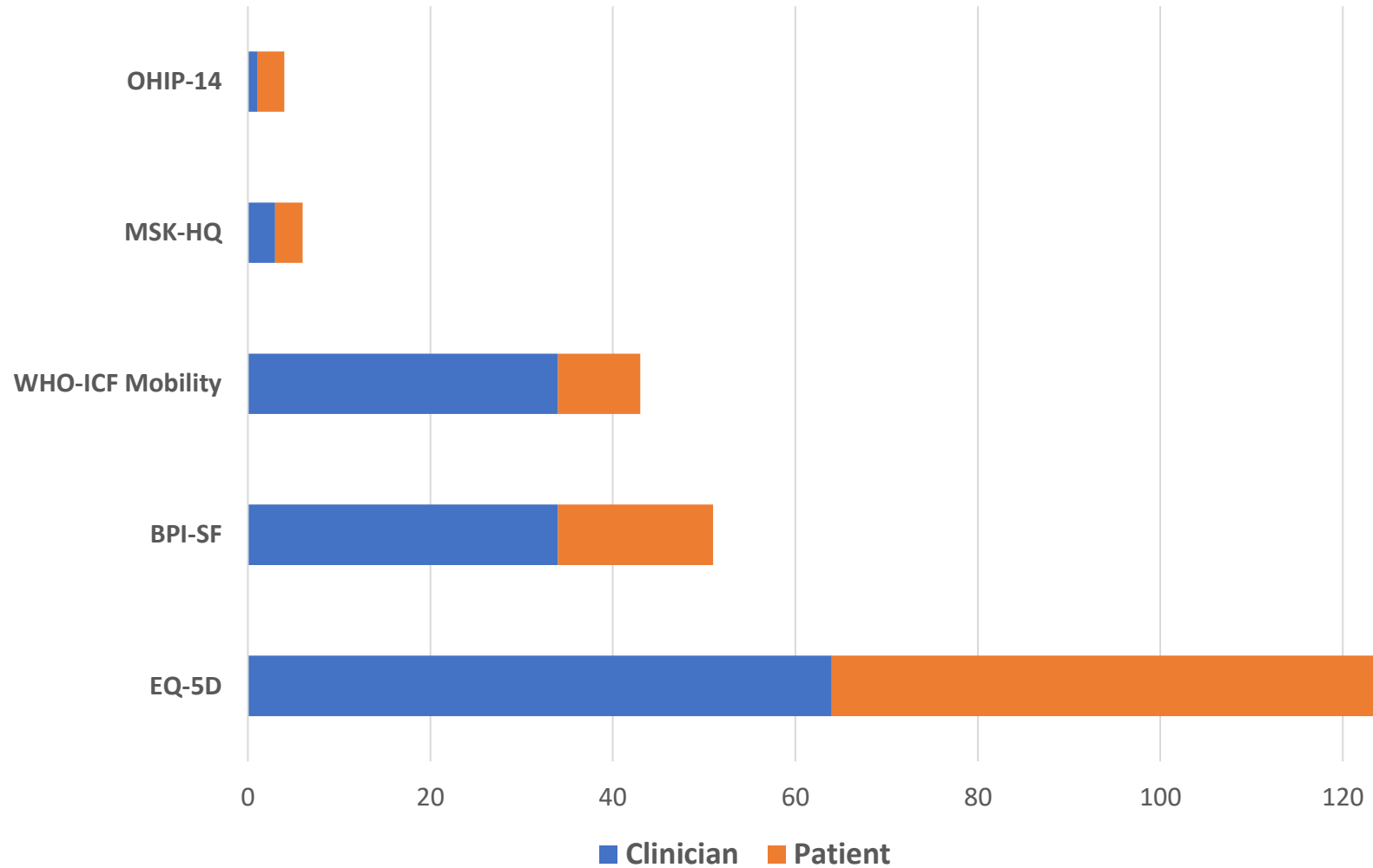
- **EQ-5D** – This survey helps people talk about their overall health and well-being.
- **BPI** – This survey is used to understand how much pain someone feels.
- **WHO-ICF Mobility** – This survey helps describe how well someone can move and do daily activities.
- **MSK-HQ** – This survey helps people with muscle or joint problems like arthritis, talk about their symptoms.
- **NPSI** – This survey helps figure out different types of pain that come from nerve problems.
- **OHIP14** – This survey allows people to share how mouth and teeth problems affect their lives.
- **PHPQoL** – This survey helps people with calcium-related issues tell how they feel.

If you want to know more about our PROMs you can always ask your doctor or contact us at [registries@lumc.nl](mailto:registries@lumc.nl).

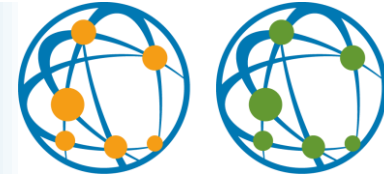
# Patient-Reported Outcome Measures (PROMs)



## Generic PROMs



# For More Information



<https://www.eurreb.eu/>



## Online Drop-in sessions

- Every second Friday 14:00-15:00 CEST
- Every fourth Wednesday 16:00-17:00 CEST

Find the link on our homepage!



Join our next platform Drop-In session  
Friday 10 May 2024 on 14:00 – 15:00 CEST

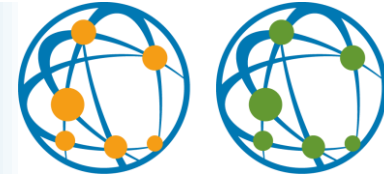


[registries@lumc.nl](mailto:registries@lumc.nl)



[European Registries for Rare Endocrine and Bone Conditions](#)

# Acknowledgements



## LUMC project management team

Prof. Natasha M. Appelman-Dijkstra, coordinator EuRR-Bone  
Prof S. Faisal Ahmed, coordinator EuRRECa  
Tess de Rooij, project manager  
Jacqueline van der Blom de Gunst, project assistant  
Dr. Ana Priego Zurita, quality manager  
Dr. Mariya Cherenko, data manager



**EuRR-Bone**  
European Registries for Rare  
Bone and Mineral Conditions



**EuRRECa**  
European Registries for  
Rare Endocrine Conditions



Endo-ERN  
European Reference Network  
on Rare Endocrine Conditions



ERN BOND  
EUROPEAN REFERENCE NETWORK  
ON RARE BONE DISEASES



European  
Reference  
Networks



Funded by  
the European Union



University  
of Glasgow



Leiden University  
Medical Center

[www.eurreb.eu](http://www.eurreb.eu)

[registries@lumc.nl](mailto:registries@lumc.nl)

