

# European Rare Endocrine and Bone Conditions (WP5)

Prof. Natasha Appelman-Dijkstra - Leiden University Medical Center,



# European Registries for Rare Endocrine and Bone conditions

Prof. Natasha Appelman-Dijkstra on behalf of WP5



**EuRREB**

European Registries for Rare Endocrine and Bone conditions

Endo-ERN GA 2025



# The Evolution of EuRRECa and EuRR-Bone to EuRREB



2016



2017

**EuRRECa approved**



**Endo-ERN**  
European Reference Network  
on Rare Endocrine Conditions

**European Registries for Rare Endocrine conditions (EuRRECa)**

2018

**e-REC launched**

2019

**Core Registry launched**

2020

**EuRR-Bone approved**



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

**European Registries for Rare Bone conditions (EuRR-Bone)**

2022

**Condition specific modules launched**

2023

**Original funding ends, merging of the 2 projects to EuRREB  
Servers/PMT move to Leiden**

2024

**Integration as WP5 registries within Endo-ERN & ERN BOND**

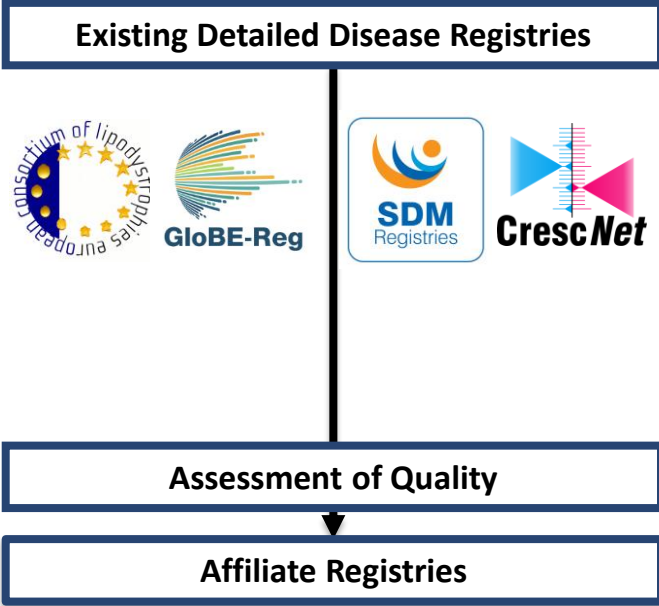
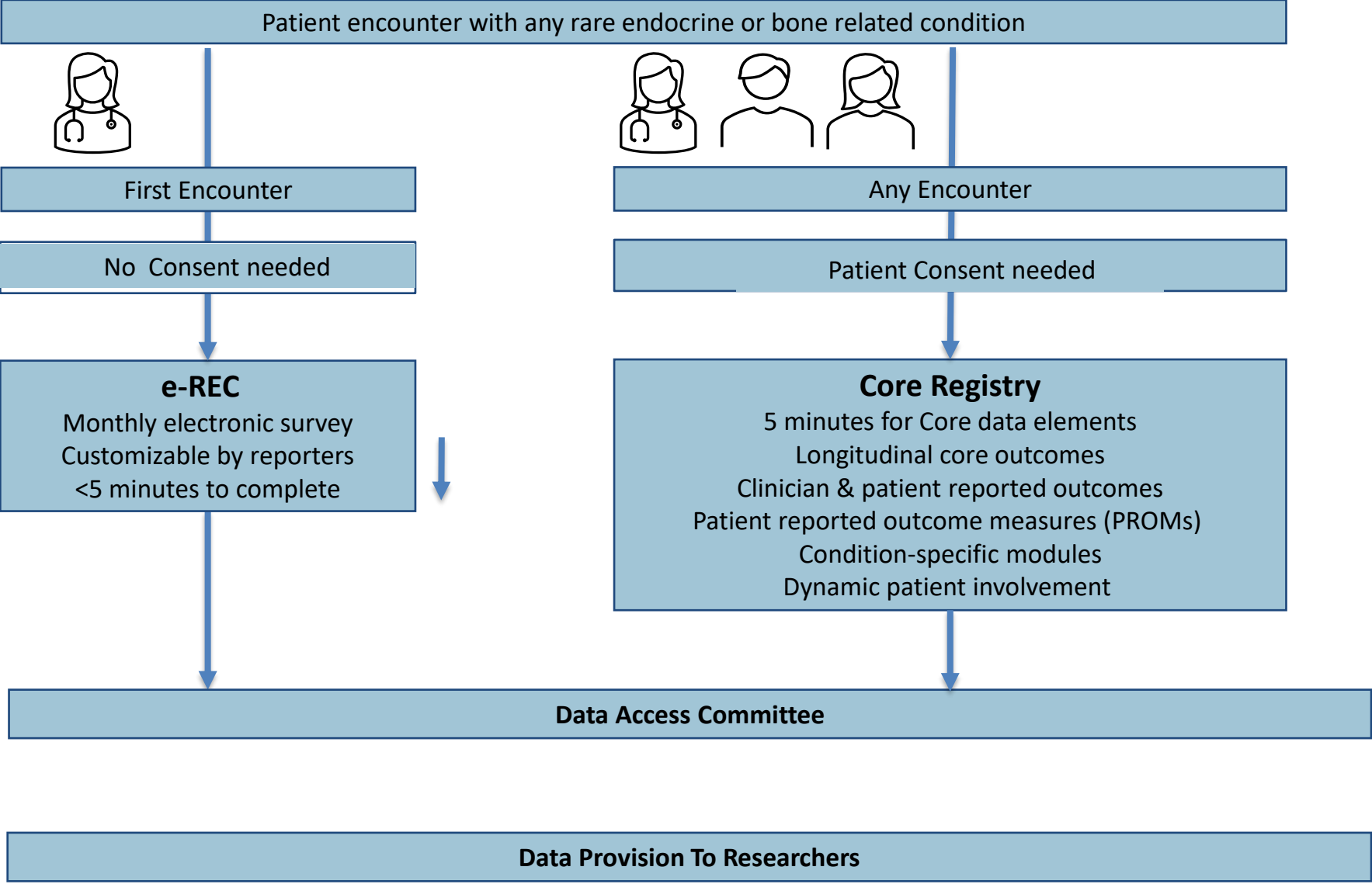


**European Registries for Rare Endocrine and Bone conditions**

2027



# The Registries in a Nutshell



# e-REC



# Live Case Count



## 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	957	5399	6356
BONE DYSPLASIA	1072	1401	2473
CALCIUM & PHOSPHATE	592	2679	3271
GENETIC ENDOCRINE TUMOURS (incl. NETs)	278	1735	2013
GLUCOSE & INSULIN	1068	621	1689
GROWTH & OBESITY	2437	153	2590
HYPOTHAL & PITUITARY	1547	15502	17049
SEX DEVELOPMENT	5818	4762	10580
SYSTEMIC & RHEUMATOLOGICAL	7	42	49
THYROID	1764	9263	11027
<b>TOTAL</b>	<b>15540</b>	<b>41557</b>	<b>57097</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)



Funded by  
the European Union



Endo-ERN



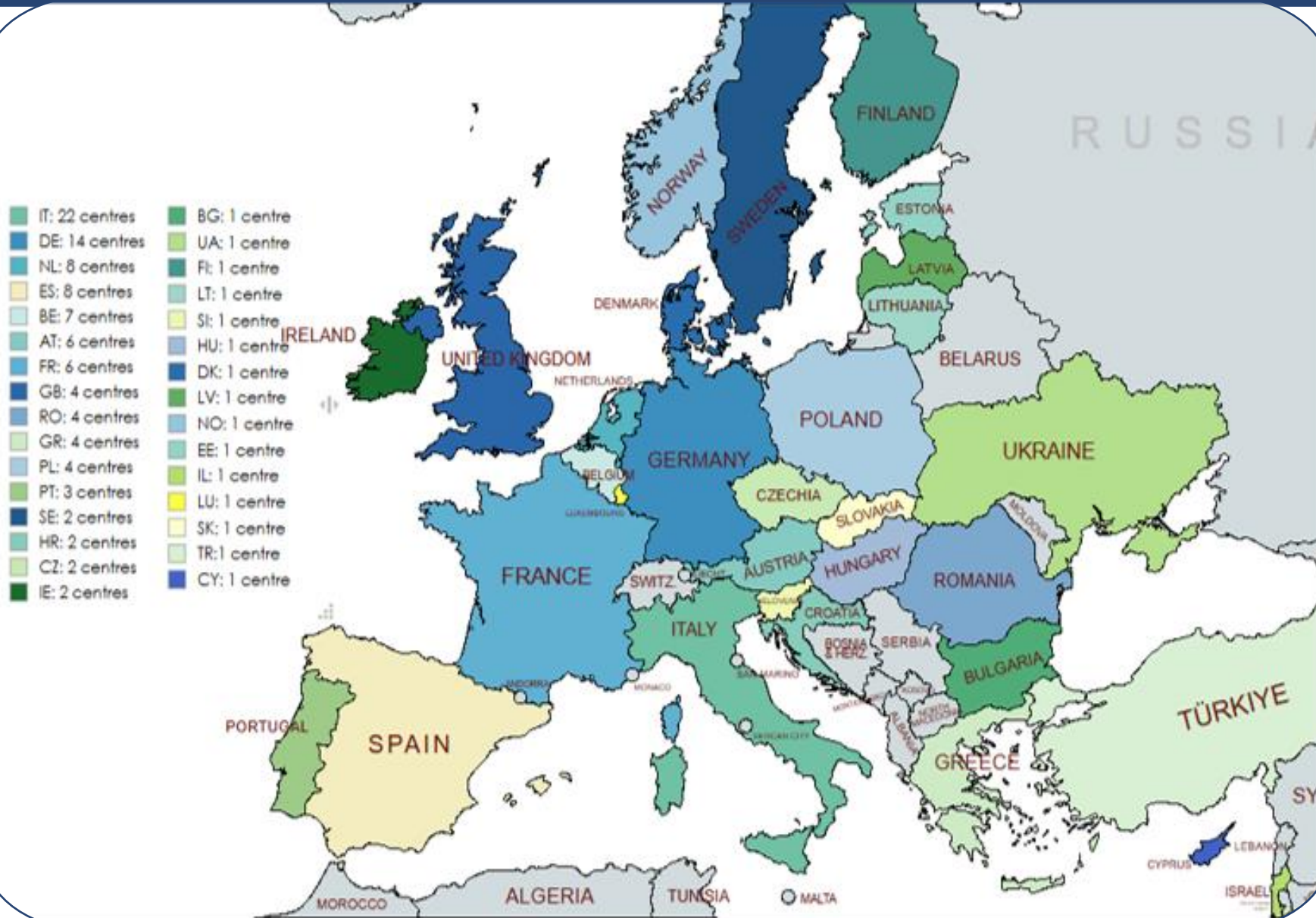
European  
Reference  
Network



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# Centres Reporting in e-REC



July 2018 – December 2024

## Active reporters:

113 centres from 32 countries  
(5 non-EU)

## HCPs ERN affiliation

Endo-ERN only - 62

Endo-ERN and ERN-BOND - 29

ERN-BOND only - 3

Not affiliated to either – 19

## Number of patients in e-Rec:

56573 cases

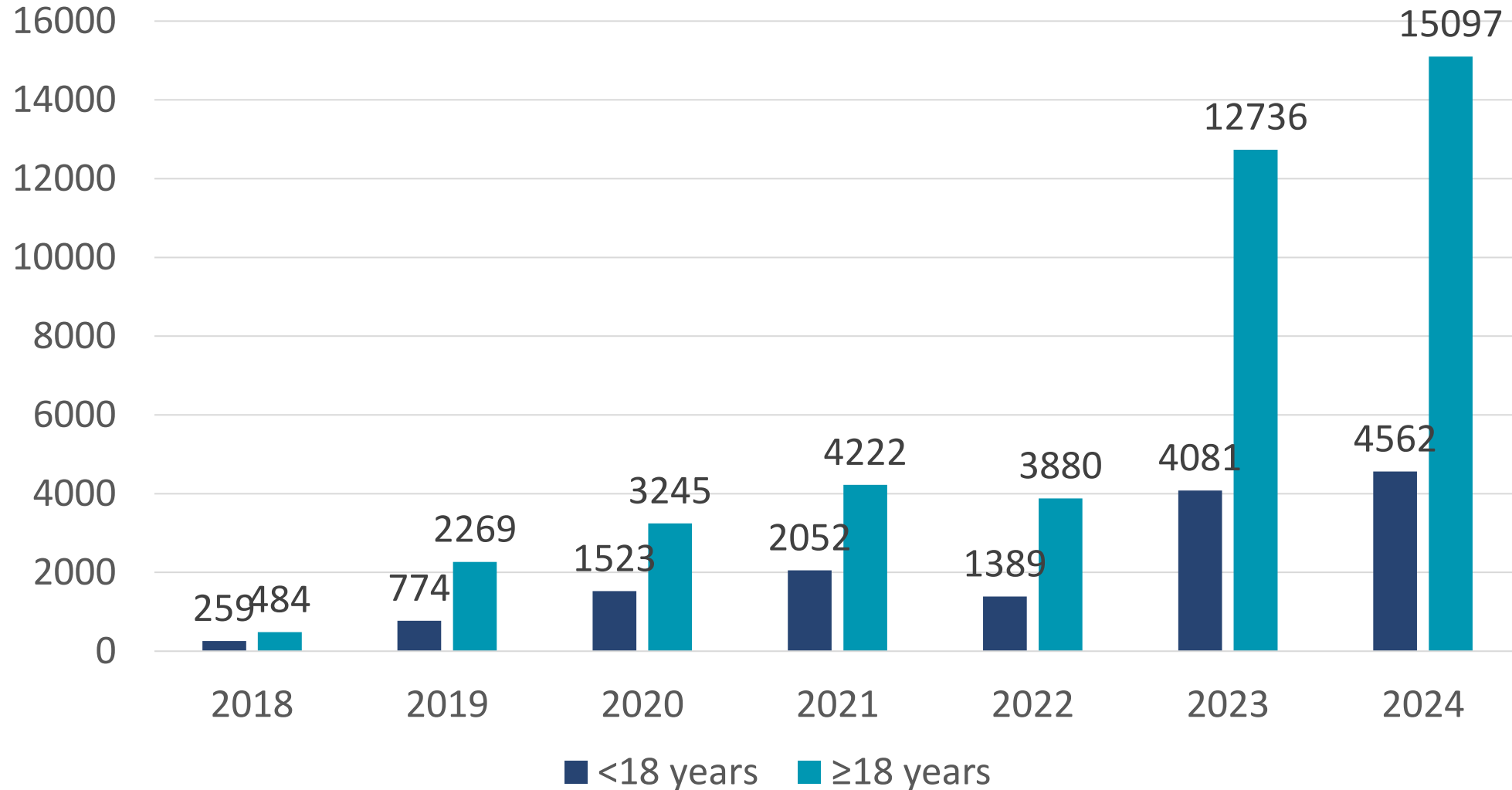
(41933 in adults, 14640 in children)

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



# Reporting in e-REC

Time trends in reporting  
(results according to age group)



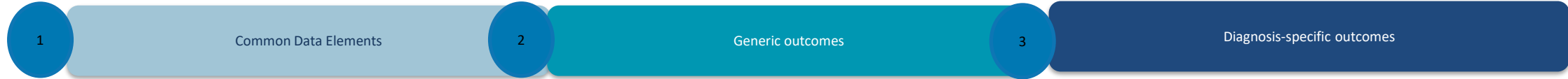


# The Core Registry



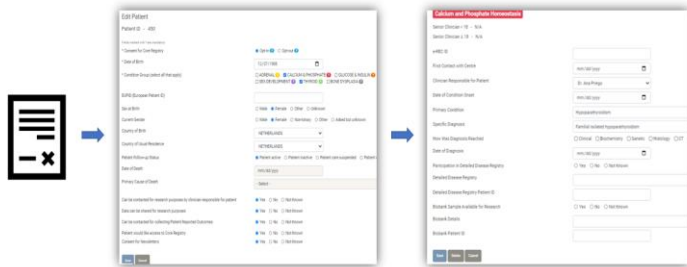
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# Levels of Participation



Clinician

Patient



**EQ-5D**  
10 languages

**WHO-ICF**  
English

**BPI-SF**  
10 languages

**OHIP**  
4 languages

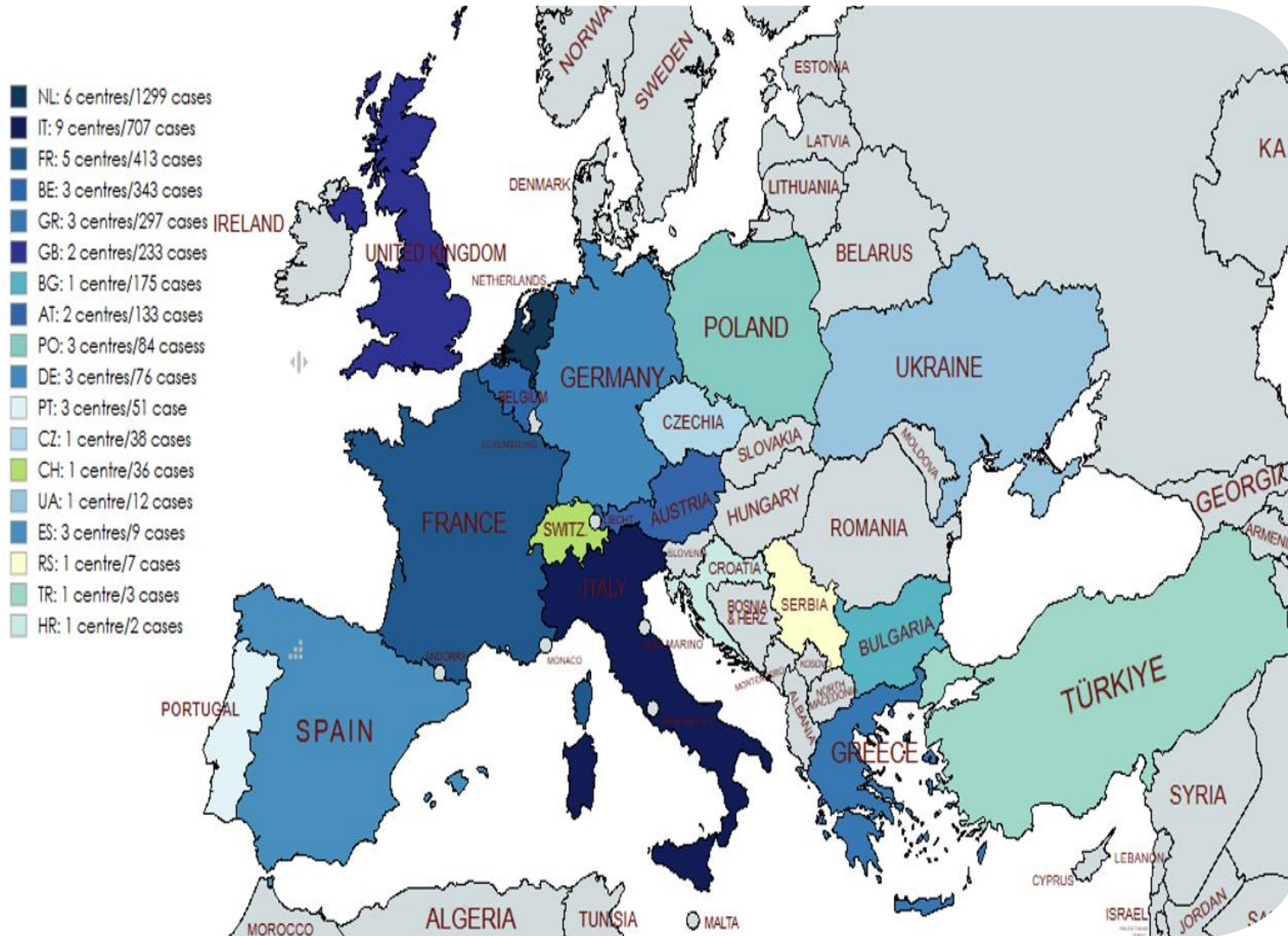
**MSK-HQ**  
6 languages

**NPSI**  
8 languages

1	iPPSD/PHP
2	Pituitary adenoma
3	Achondroplasia
4	FD/MAS
5	Osteogenesis imperfecta
6	Parathyroid carcinoma
7	Rare hypophosphataemia
8	Melorheostosis
9	Rare obesity
10	Gender Incongruence
11	Paediatric differentiated thyroid carcinoma
12	Langerhans Cell Histiocytosis
13	Chronic Nonbacterial Osteitis (CNO)



# Centres Reporting in the Core Registry



Oct 2019 – December 2024

51 centres from 20 countries

**HCPs ERN affiliation**

Endo-ERN only 16

Endo-ERN and ERN BOND 15

ERN BOND only 4

Not affiliated to either 16

**Number of patients in the Core registry:**

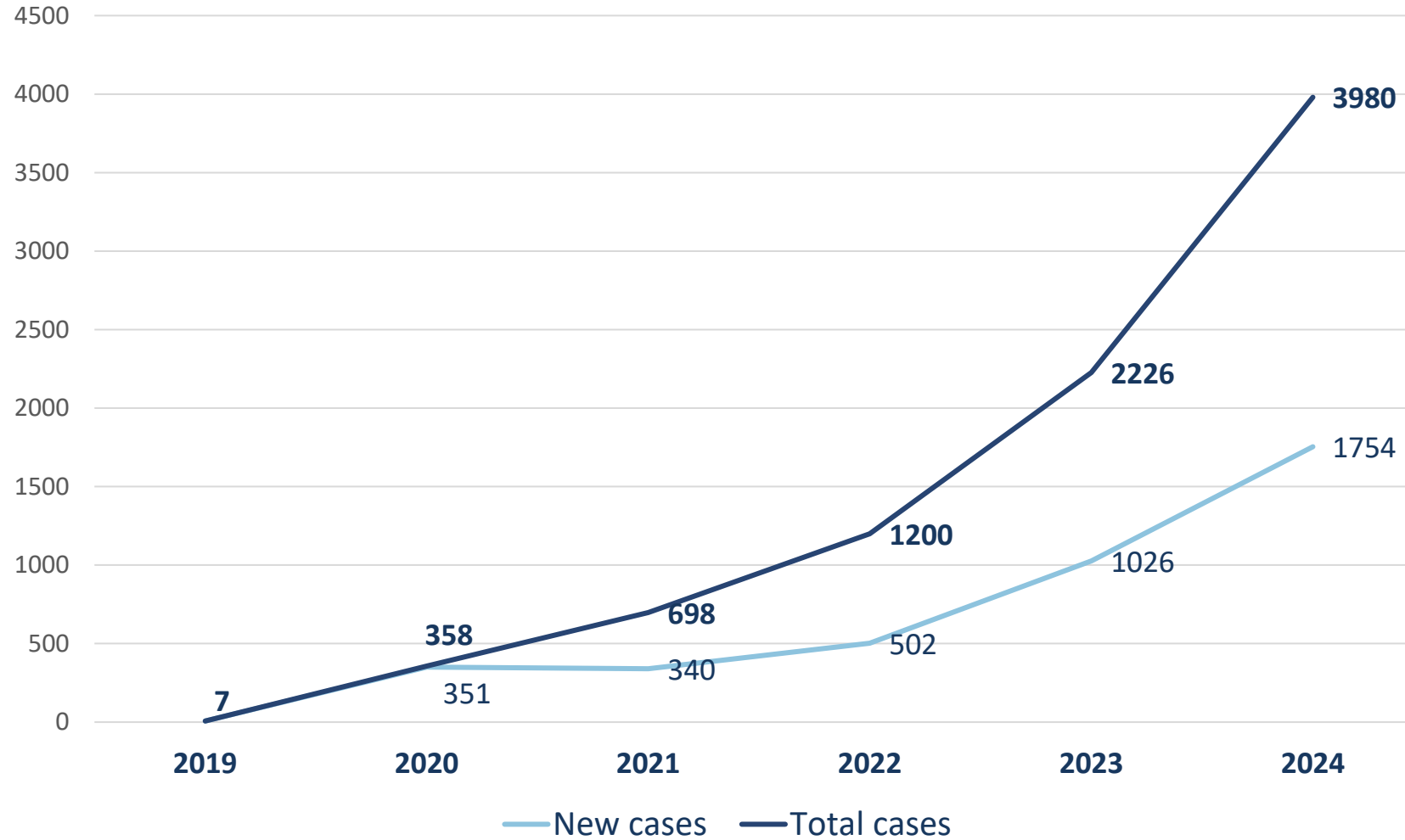
3980 cases

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



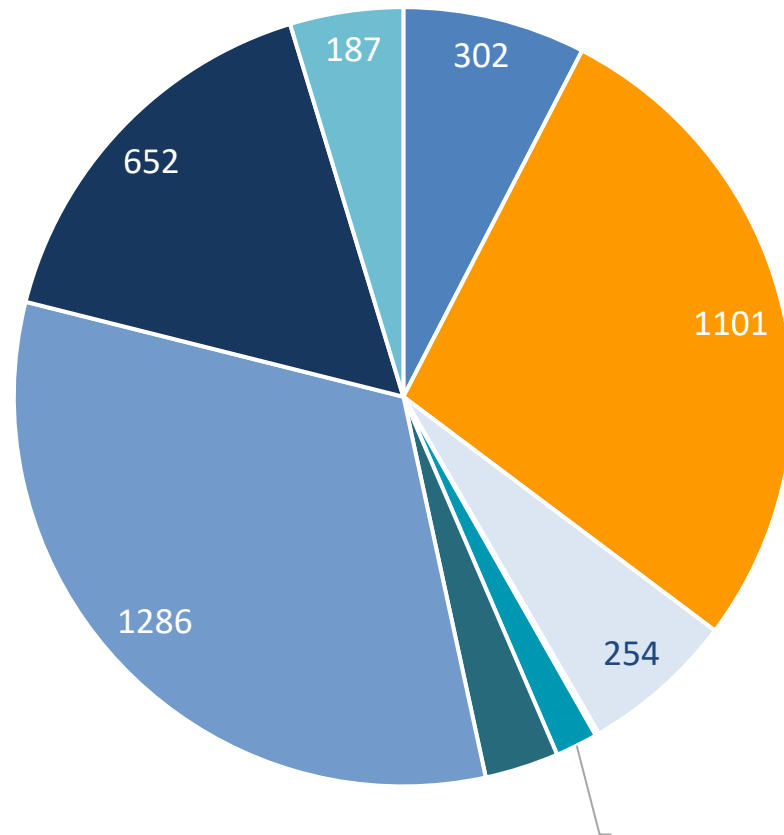
# Reporting Trends in the Core Registry

## Patient Records in the Core Registry



# Cases Distribution in the Core Registry by MTG

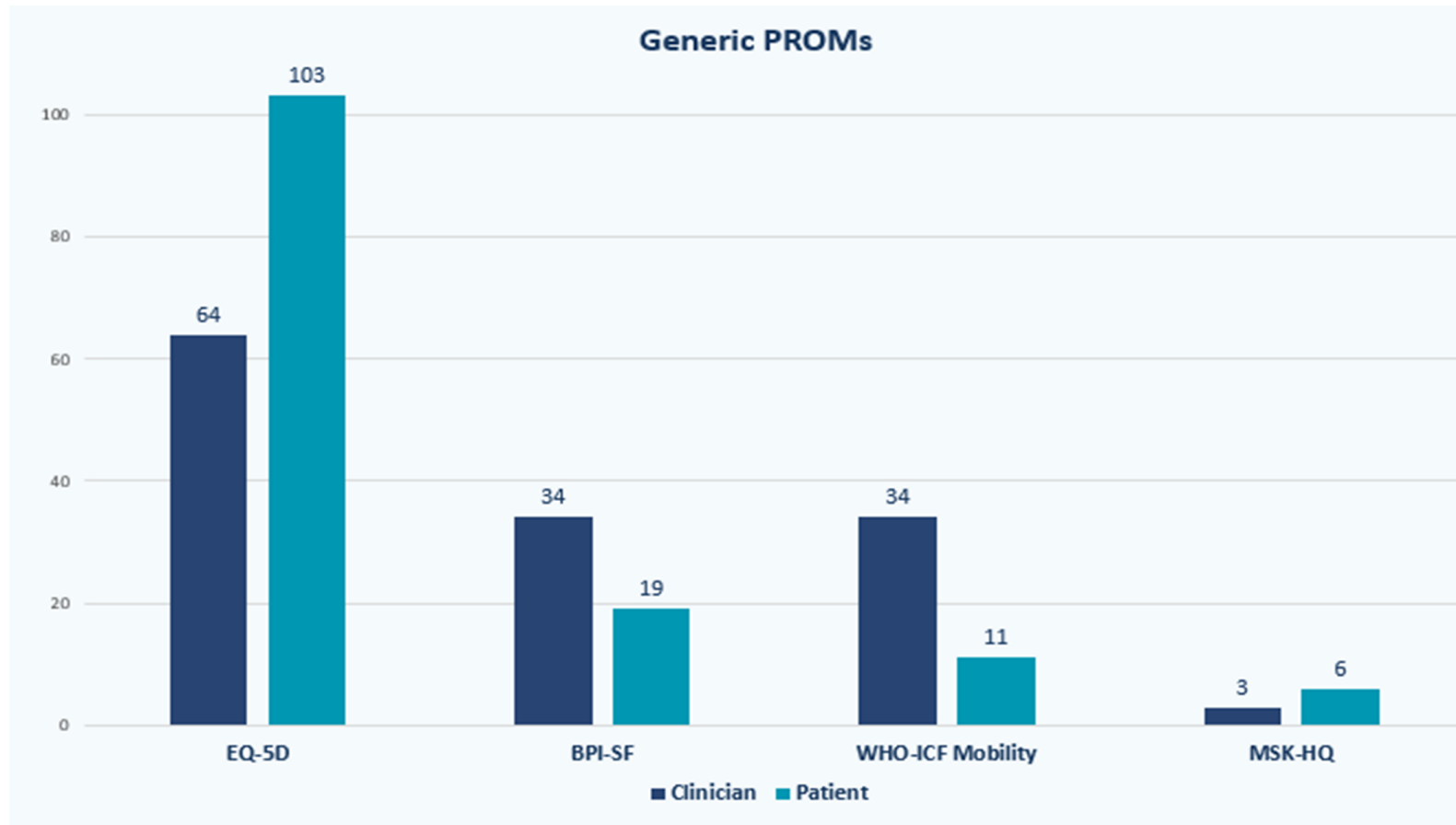
Total Number of Cases by MTG  
(n=3980)



- Adrenal
- Bone Dysplasia
- Calcium and Phosphate
- Genetic Disorders of Glucose and Insulin Homeostasis



# Patient-Reported Outcome Measures (PROMs)



Lately OHIP-14 and NPSI were added to the platform.



# The Condition-Specific Modules



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# Condition-Specific Modules and Active Studies

CSM	MTG	Year	N patients in CR	Outcomes completed	Study group leads	Active study
iPPSD/PHP	MTG2	2021	32	11	Agnès Linglart/Diana-Alexandra Ertl	Natural History of iPPSD/PHP
Pituitary adenoma	MTG6	2021	739	1114	Alberto Pereira	<ol style="list-style-type: none"> <li>1. Epidemiological Surveillance of Pituitary Tumours within Endo-ERN</li> <li>2. The Clinical Usefulness of the PANOMEN 3 Grade Score</li> <li>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</li> </ol>
Achondroplasia	BD	2022	174	125	Klaus Mohnike/ Ines Alves	Natural History of Achondroplasia
FD/MAS	BD	2022	660	1422	Natasha Appelman-Dijkstra	Natural History of FD/MAS
Osteogenesis imperfecta	BD	2022	107	26	Wolfgang Hoegler	Under development
Parathyroid carcinoma	MTG2	2022	14	15	Maria Luisa Brandi	Natural History of Parathyroid Carcinoma
Rare hypophosphataemia	MTG2	2023	84	61	Agnès Linglart/Diana-Alexandra Ertl	Under development
Melorheostosis	BD	2023	20	18	Martine Cohen-Solal	Natural History of Melorheostosis
Rare obesity	MTG5	2023	68	10	Erica van den Akker	Natural History of Rare Obesity
Gender Incongruence	MTG7	2023	152	159	Martine Cools/Silvia Ciancia	Current status of healthcare for transgender adolescents across Europe
Paediatric differentiated thyroid carcinoma	MTG8	2024	66	30	Hanneke van Santen/ Sarah Clement	Under development





# Condition-Specific Modules under Development

CSM	MTG	Year	N patients in CR	Outcomes completed	Study group leads	Active study
Langerhans Cell Histiocytosis	Sys	2025	0	0	Polyzois Makras	Under development
Chronic Nonbacterial Osteitis (CNO)	Sys	2025	0	0	Elizabeth Winter	Under development
Pregnancy and Rare Endocrine and Bone conditions	all	2025	0	0	Tim Korevaar and Natasha Appelman-Dijkstra	Under development



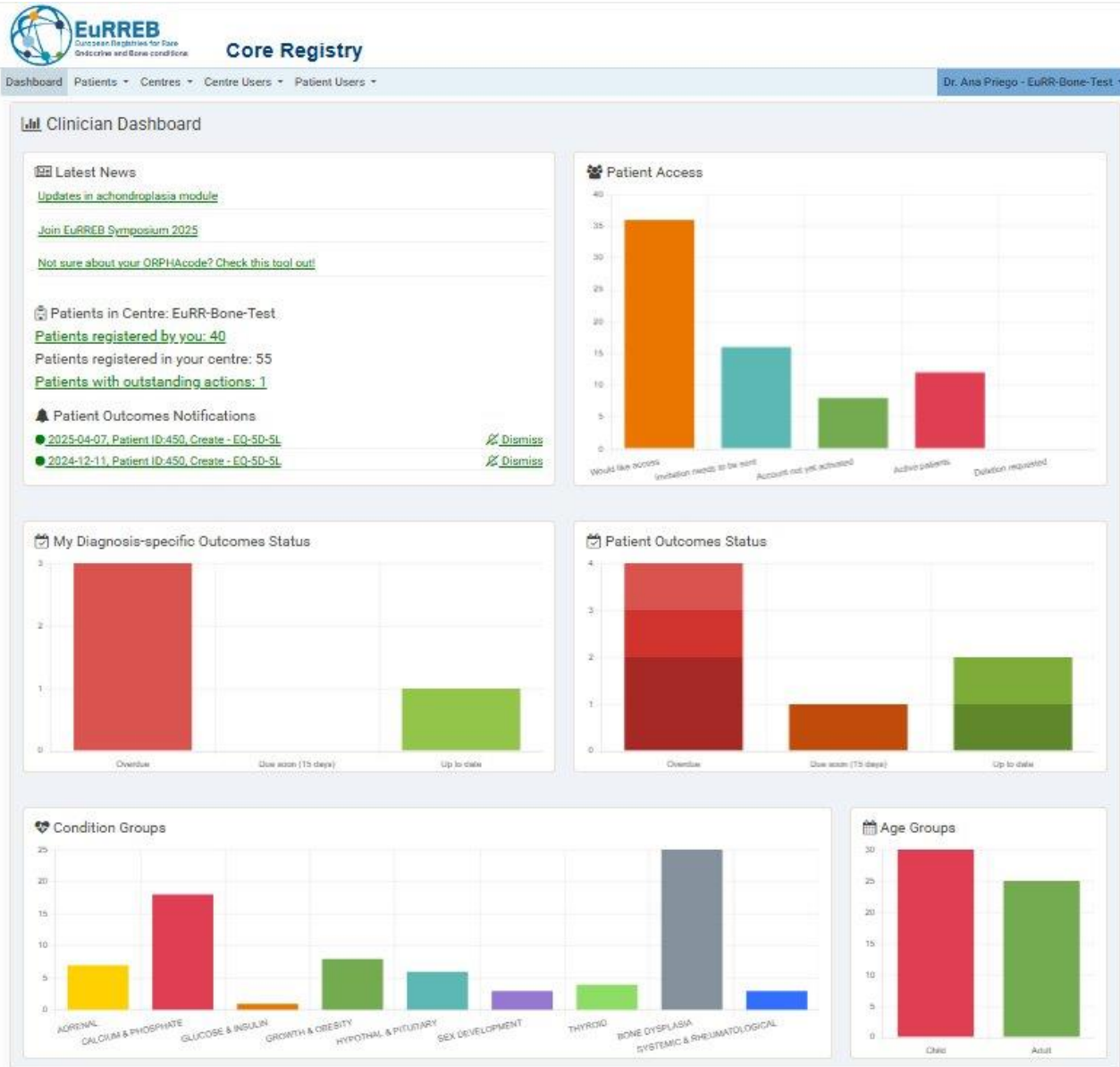
# The Platform Updates



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# The Core Registry Dashboard



Different features of the dashboard:

- the access to all patients registered in your centre
- the latest news
- the distribution of cases by MTG and age groups
- patient access status
- patient and clinician outcomes status



# The Reminders for Responsible Clinicians



## Core Registry

Dashboard Patients Centres Users Centre Users Patient Users Reference Data Emails Reporting

Mariya Cherenko - Leiden University Medical Center

### Diagnosis-specific Outcome Details

Patient ID - 4004 Date of Birth - 1998-12-08 Condition Group - Bone Dysplasia

[Supplemental material to complete this module](#)

[Read more about this](#)

Questionnaire: Achondroplasia\_V2.2 - adult

Created Date: 2025-04-30

Completed By: Mariya Cherenko

Reminders for Responsible Clinician - Mariya Cherenko ?

Add Reminder

Name	Reminder Date	Details	Created On	Created By	Actions
No reminders found					

Date of Birth	1998-12-08	e-REC ID	Primary Condition	Primary bone dysplasia with micromelia
Current Gender	Female	Clinician Responsible for Patient	Specific Diagnosis	Achondroplasia

Patient history Anthropometry Monitoring Medical therapy Surgery and other therapies Clinical outcome

#### Add Reminder

1 month 2 months 3 months 6 months 1 year 2 years 3 years 4 years 5 years 10 years

\* Name

\* Reminder Date

Details

Cancel Save



# The EuRREB Participating Centre Voucher



# Two Types of Vouchers

## Centre Set-Up Voucher

- €500 per centre to support starting data entry into the Core Registry
- Once a year, 3 exclusively for members of Endo-ERN
- 50% of amount when centre received ethics approval
- 50% of amount when centre has reported 100 cases in Core Registry

**Deadline: 1<sup>st</sup> of October 2025**  
You will receive an e-mail from Endo-ERN when applications open

## Advanced Centre Voucher

- €1500 per active centre to support collecting high quality data for a specific EuRREB study
- Once every 6 months, depending on study launches (<https://eurreb.eu/core-registry/active-studies/>)
- Open to all EuRREB centres that are already reporting cases in the Core Registry
- Two deliverables are expected:
  - 1) Uploading data of all relevant registered patients in the Core Registry into the CSM of interest
  - 2) Entering 80% of e-REC reported patients in the CSM of interest for 2 years



# The KPIs



# Key Performance Indicators

## Implement in the Core Registry at two levels:

- General: applicable to all the conditions covered and collected as part of the Common Data Elements.

*e.g. Do you assess quality of life in this patient?*

- Within Condition-specific module: developed together with working and study groups within EuRREB.

## Implement in e-REC (Monthly surveys):

- How many of your newly diagnosed patients are on ...
- How many ... surgeries have been performed

## examples

- MTG 2 – e.g., number of parathyroid surgeries in your center per year
- MTG 6 e.g., number of pituitary surgeries per year





# Thank you!

## Ways to contact us:



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



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



drop-in sessions via Zoom



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