European Registries for Rare Endocrine and Bone conditions

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On behalf of Work Package 5

Endo-ERN General Assembly
Athens, Greece - April 21, 2026

EuRREB
European Registries for Rare
Endocrine and Bone conditions

Funded by
the European Union

EuRREB Registries Team Changes

e-REC

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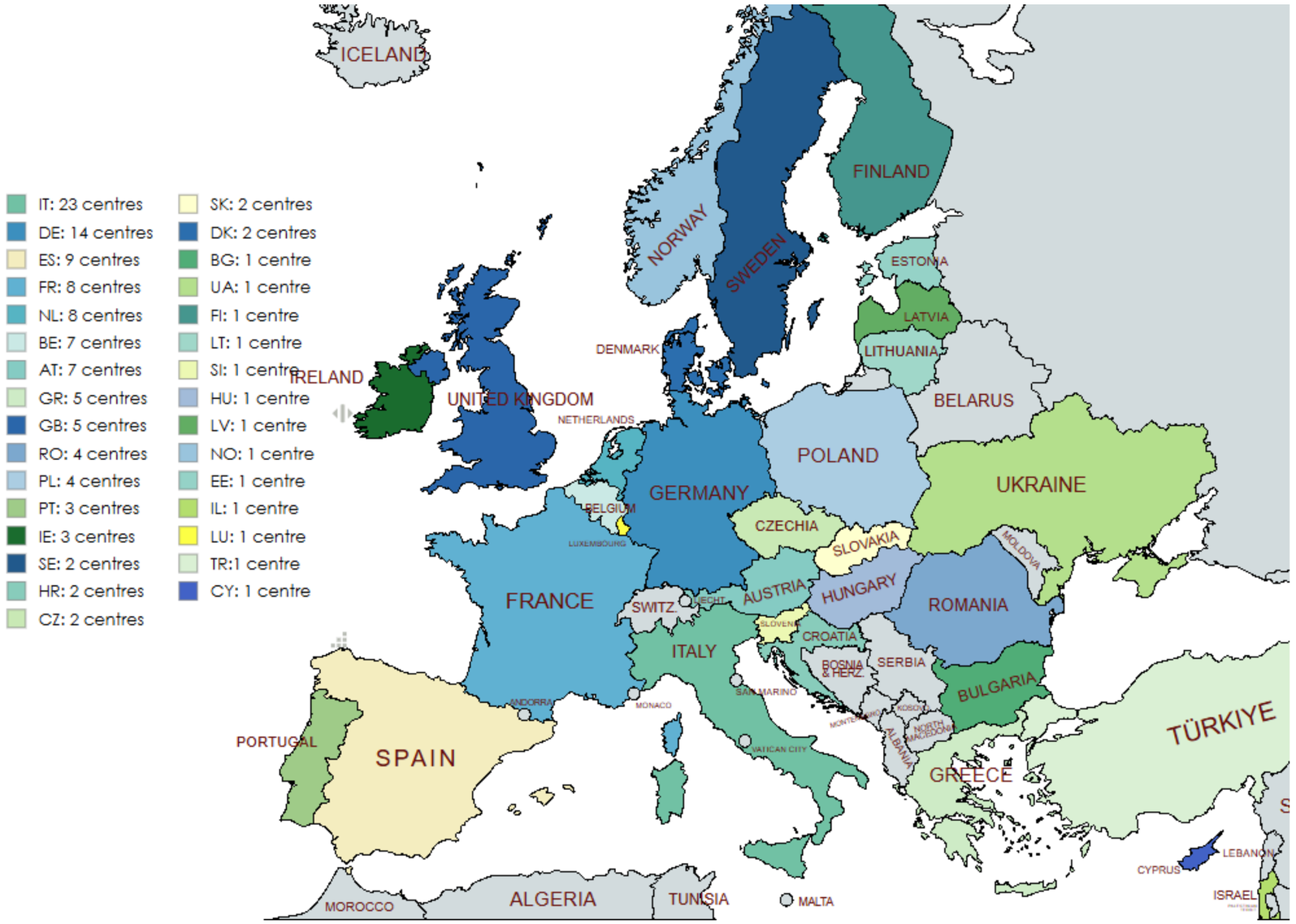
Endocrine and Bone conditions

Centres Reporting in e-REC



July 2018 – December 2025

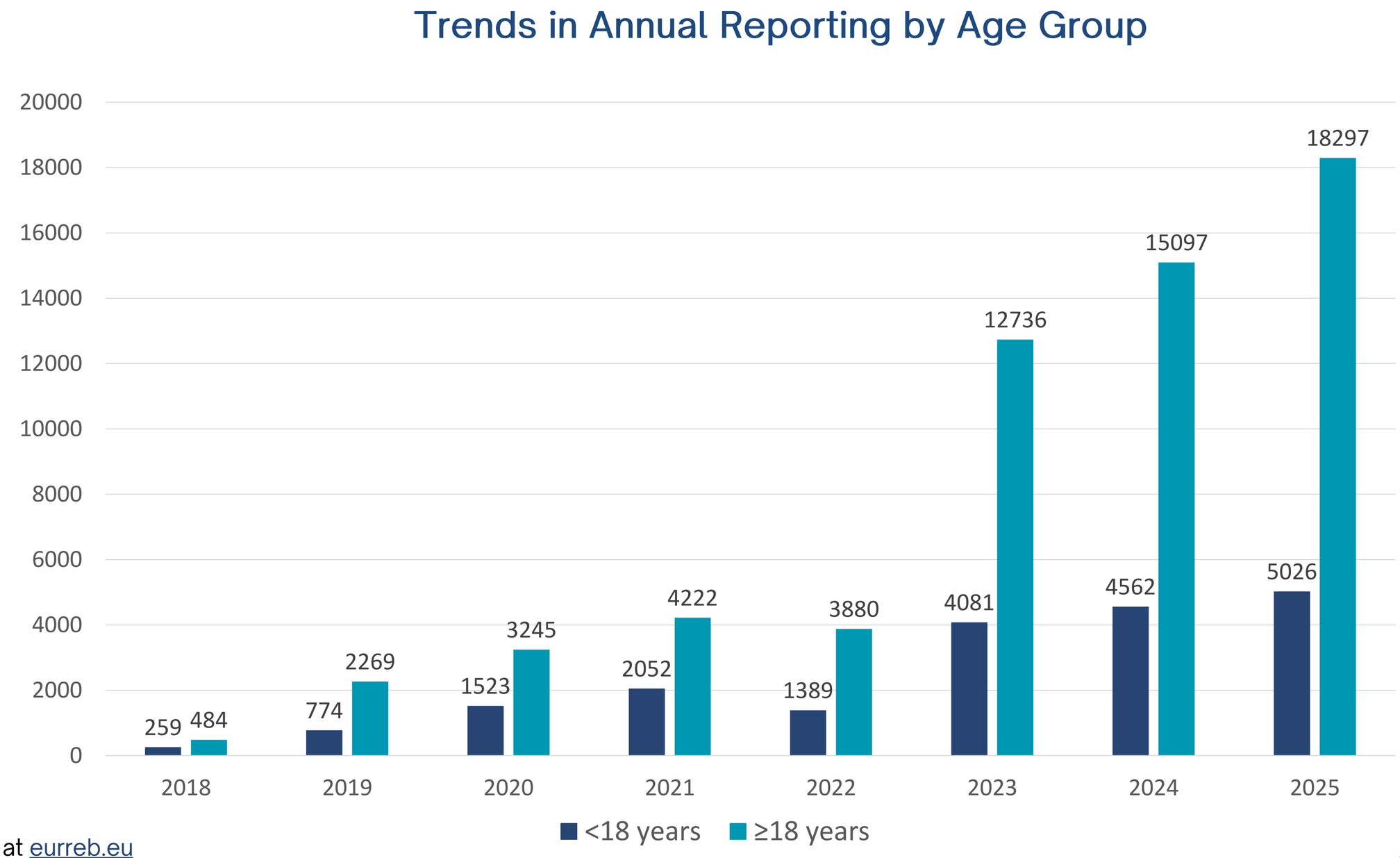
Active reporters:
122 centres from 32 countries
(5 non-EU)

<u>HCPs ERN affiliation</u>
Endo-ERN only - **67**
Endo-ERN and ERN-BOND - **30**
ERN-BOND only - **5**
Not affiliated to either – **20**

<u>Number of patients in e-REC:</u>
79896 cases
- 60230 in adults
- 19666 in children

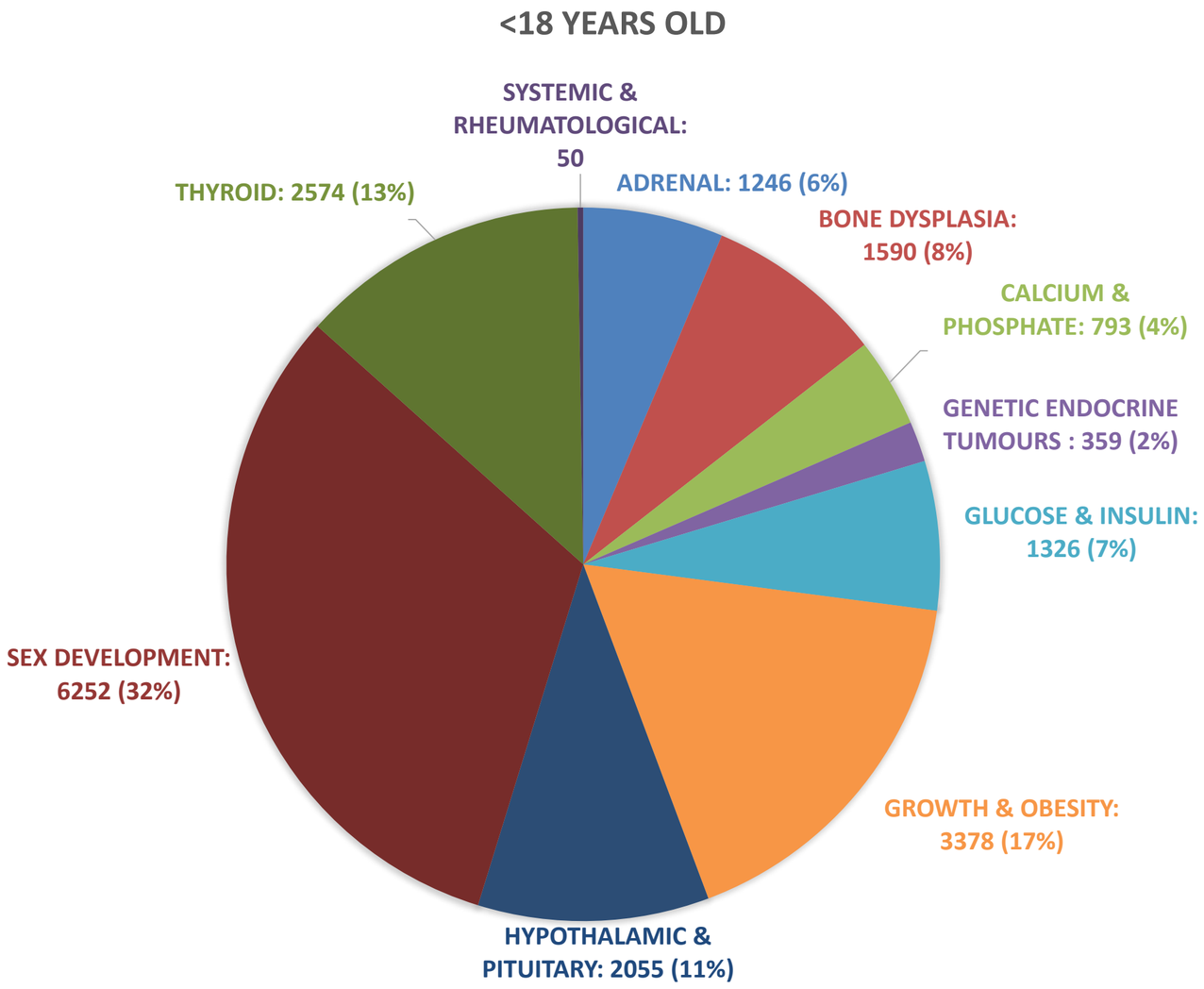
Full report at eurreb.eu

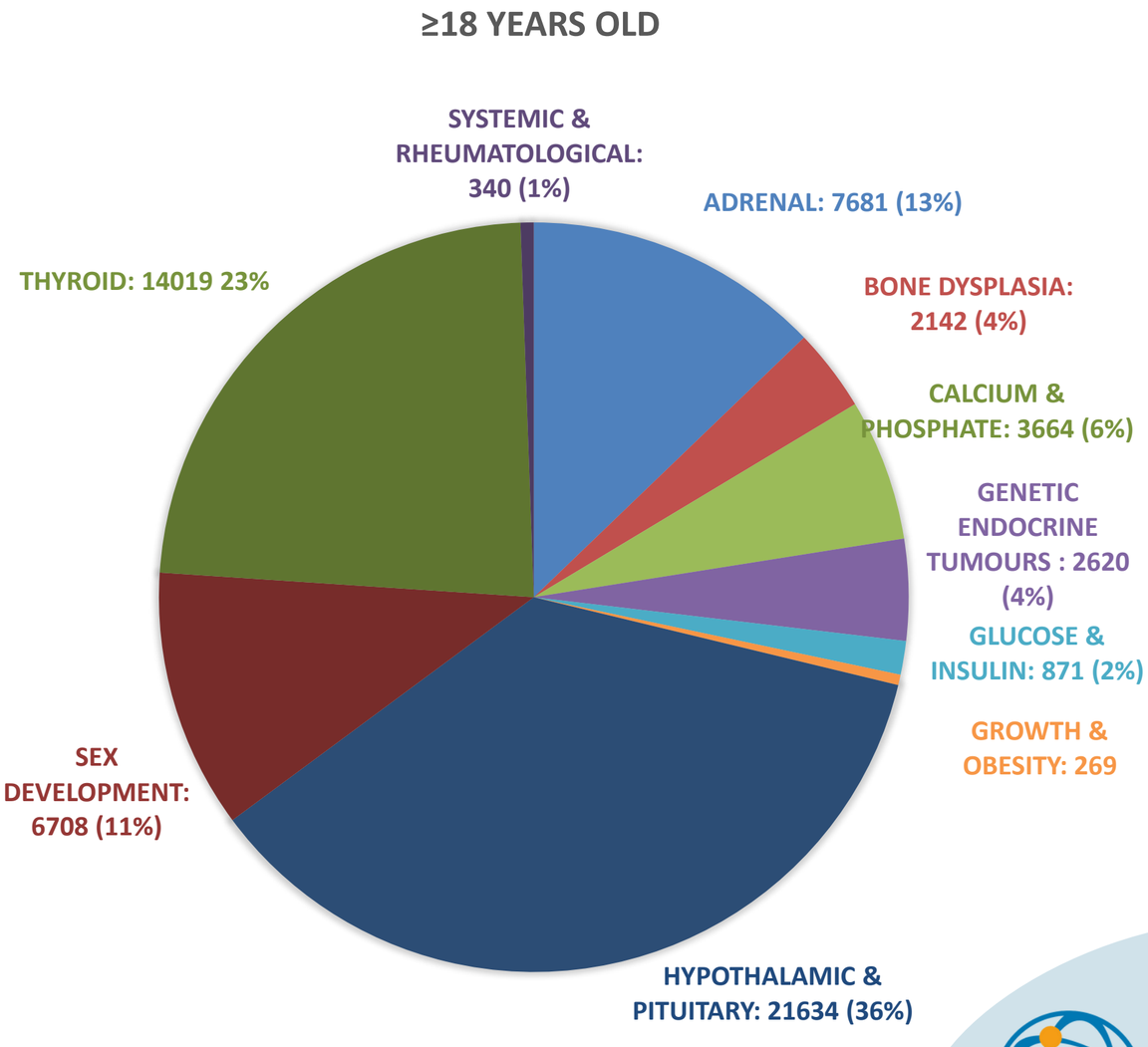
Reporting in e-REC



Full report at eurreb.eu

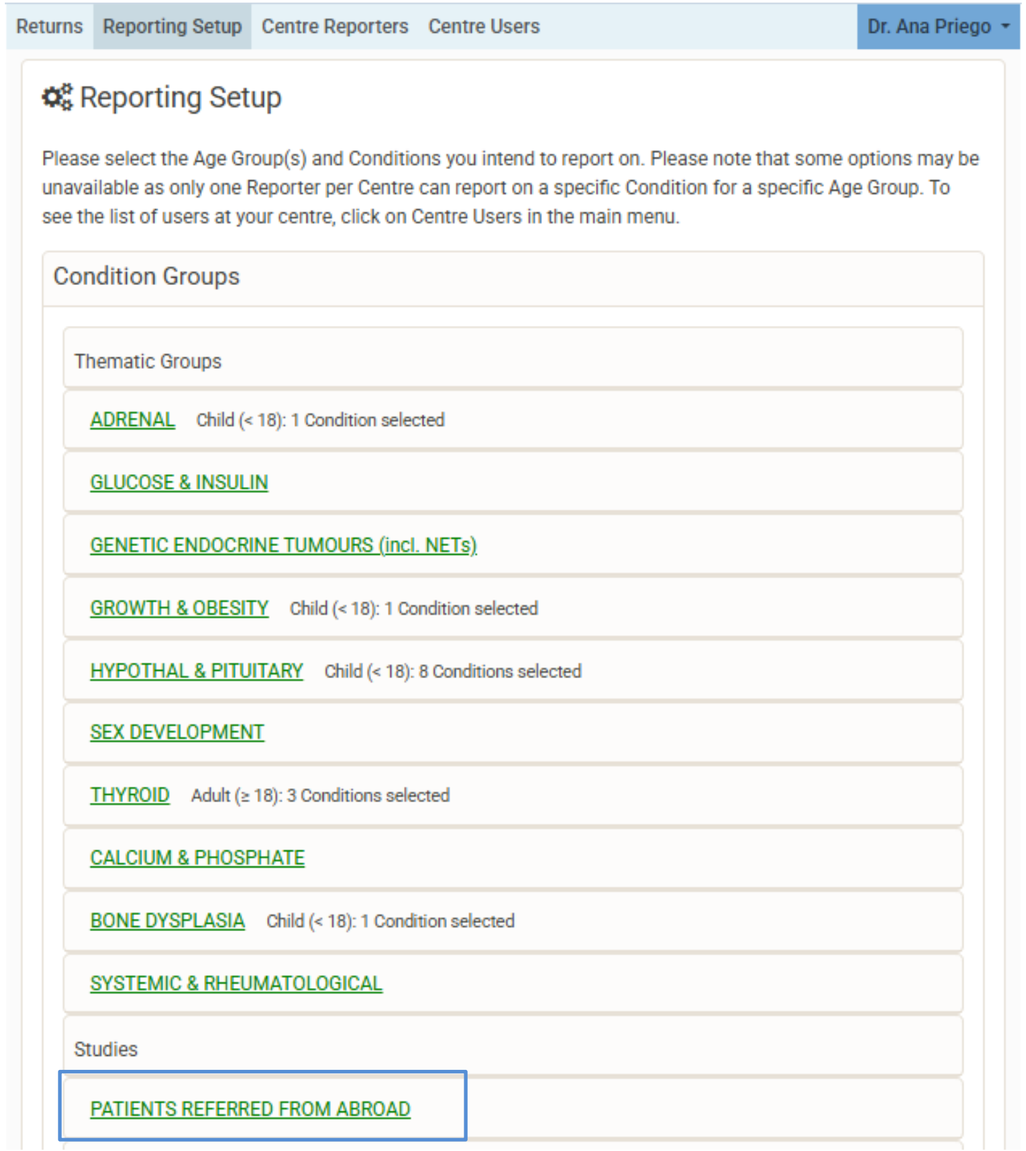
Cases Distribution in e-REC by MTG and Age Group

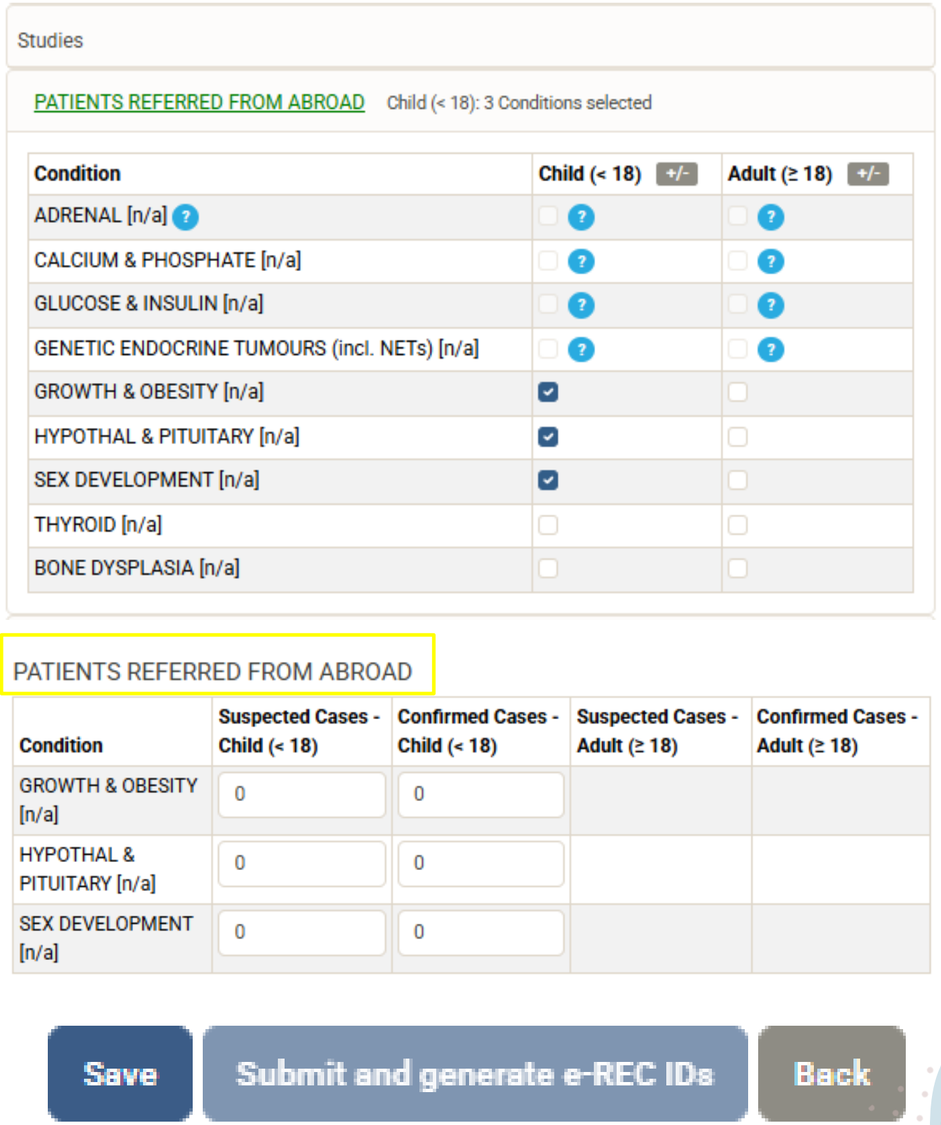




Full report at eurreb.eu

New in e-REC – Patients Referred from Abroad





Platform Developments – Yearly Centre Summary

Yearly Centre Summary

Scope: Centre: EuRR-Bone-Test

Show entire centre

Download as Excel

Cases by MTG and Age Group | Cases by Year(Suspected + Confirmed, all age groups)

MTG	Condition	ORPHAcode	Adults Suspected	Adults Confirmed	Children Suspected	Children Confirmed	Total
ADRENAL			**0**	**0**	**1**	**0**	**1**
	Cortisol producing adenomas	ORPHA443287	0	0	1	0	1
GLUCOSE & INSULIN			**0**	**0**	**1**	**1**	**2**
	Rare diabetes	ORPHA101952	0	0	1	0	1
	Hyperinsulinism	ORPHA276525	0	0	0	1	1
	Insulin resistance syndrome	ORPHA181368	0	0	0	0	0
HYPOTHAL & PITUITARY			**0**	**0**	**1**	**0**	**1**
	Pituitary Adenoma	ORPHA99408	0	0	1	0	1
	Acromegaly	ORPHA963	0	0	0	0	0
	Cushing disease	ORPHA96253	0	0	0	0	0
	Prolactinoma	ORPHA2965	0	0	0	0	0
	Non-functioning pituitary adenoma	ORPHA91349	0	0	0	0	0
	Acquired Hypopituitarism	ORPHA95502	0	0	0	0	0
	Craniopharyngioma	ORPHA54595	0	0	0	0	0
THYROID			**0**	**0**	**1**	**4**	**5**
	Thyroid hormone signaling disorders	ORPHA183631	0	0	0	1	1
	Congenital Hypothyroidism	ORPHA442	0	0	0	2	2
	Congenital Hyperthyroidism	ORPHA424	0	0	1	0	1
	Non-metastatic thyroid carcinoma	ORPHA100088	0	0	0	1	1
CALCIUM & PHOSPHATE			**0**	**0**	**0**	**0**	**0**
	Hypocalcaemic vitamin D dependent rickets	ORPHA289157	0	0	0	0	0
	Autosomal recessive hypophosphataemic rickets	ORPHA289176	0	0	0	0	0

Yearly Centre Summary

Scope: Centre: EuRR-Bone-Test

Show entire centre

Download as Excel

Cases by MTG and Age Group | Cases by Year(Suspected + Confirmed, all age groups)

MTG	Condition	ORPHAcode	2023	2024	2025	Total
ADRENAL			**0**	**0**	**1**	**1**
	Cortisol producing adenomas	ORPHA443287	0	0	1	1
GLUCOSE & INSULIN			**0**	**2**	**0**	**2**
	Rare diabetes	ORPHA101952	0	1	0	1
	Hyperinsulinism	ORPHA276525	0	1	0	1
	Insulin resistance syndrome	ORPHA181368	0	0	0	0
HYPOTHAL & PITUITARY			**1**	**0**	**0**	**1**
	Pituitary Adenoma	ORPHA99408	1	0	0	1
	Acromegaly	ORPHA963	0	0	0	0
	Cushing disease	ORPHA96253	0	0	0	0
	Prolactinoma	ORPHA2965	0	0	0	0
	Non-functioning pituitary adenoma	ORPHA91349	0	0	0	0
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THYROID			**0**	**5**	**0**	**5**
	Thyroid hormone signaling disorders	ORPHA183631	0	1	0	1
	Congenital Hypothyroidism	ORPHA442	0	2	0	2
	Congenital Hyperthyroidism	ORPHA424	0	1	0	1
	Non-metastatic thyroid carcinoma	ORPHA100088	0	1	0	1
CALCIUM & PHOSPHATE			**0**	**0**	**0**	**0**
	Hypocalcaemic vitamin D dependent rickets	ORPHA289157	0	0	0	0
	Autosomal recessive hypophosphataemic rickets	ORPHA289176	0	0	0	0

The Core Registry

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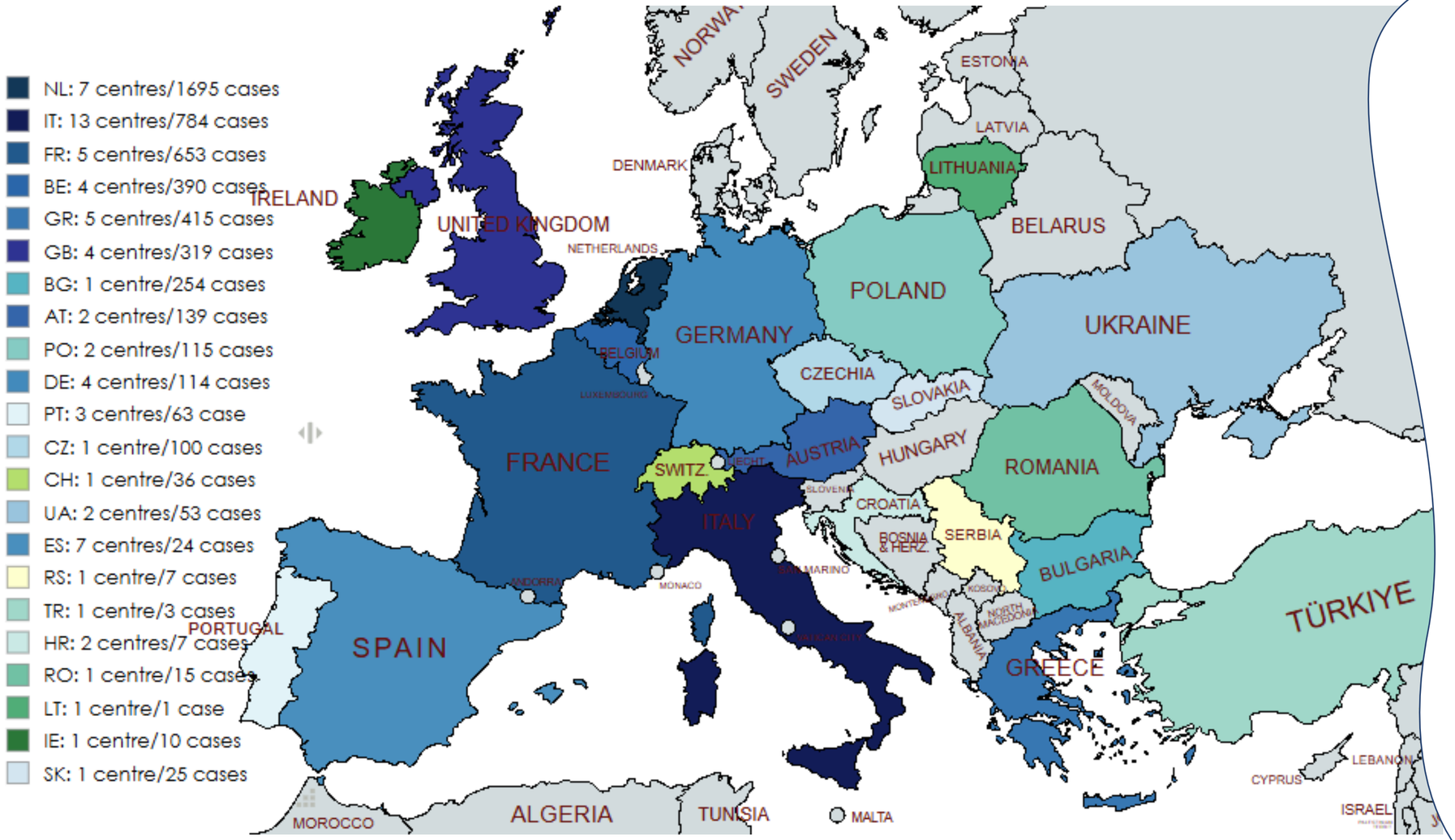
The Three Components of the Core Registry: Core Data, Generic Outcomes, and Disease Modules

	0 Consent	1 Core Data Fields	2 Generic outcomes	3 Diagnosis-specific outcomes
 Clinician		*Clinican enters basic data, including a set of Common Data Elemetns: Date of birth, country of residence, 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*

Geographical Coverage in the Core Registry



Oct 2019 – December 2025

70 centres from 22 countries

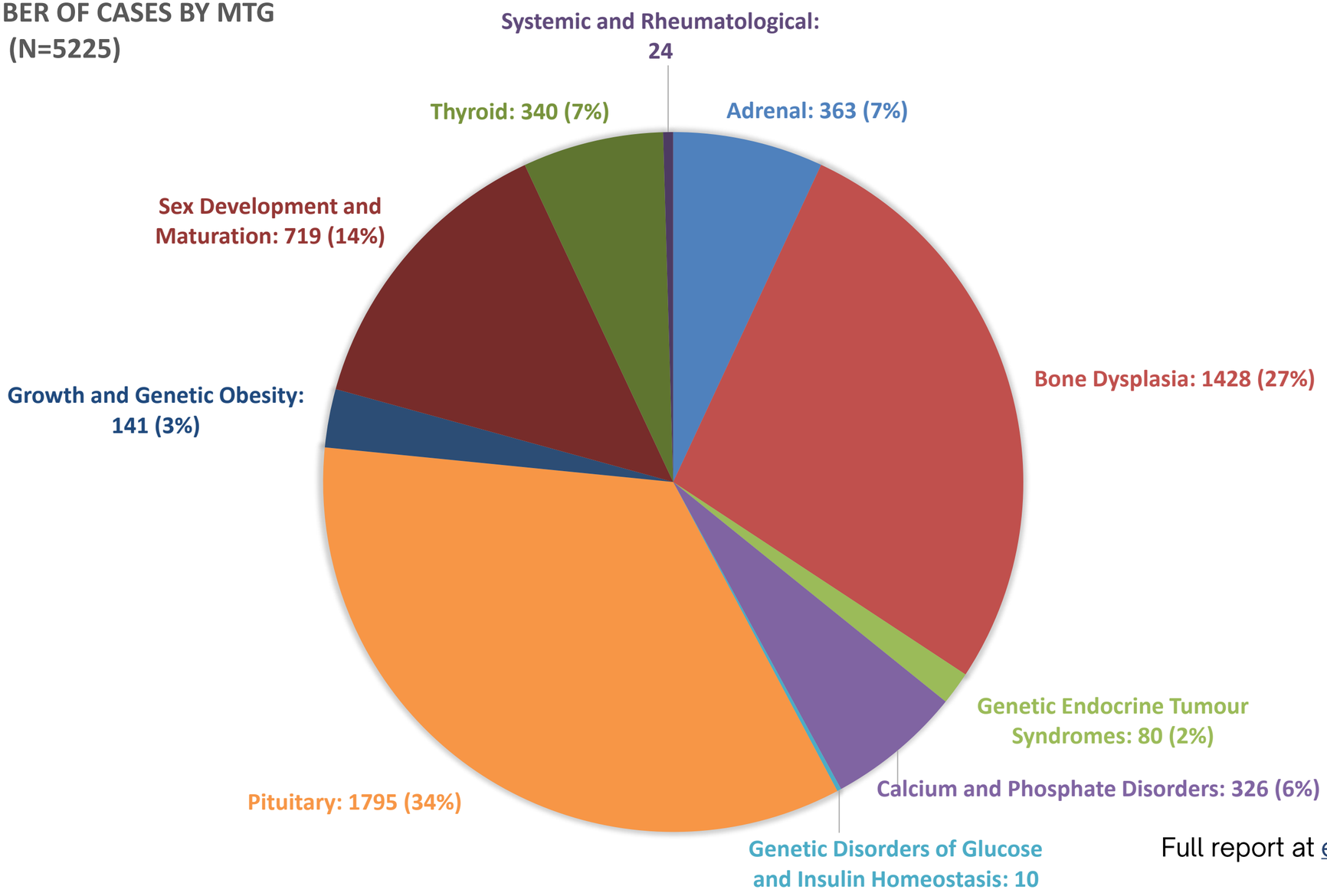
<u>HCPs ERN affiliation</u>
Endo-ERN only **25**
Endo-ERN and ERN BOND **19**
ERN BOND only **5**
Not affiliated to either **21**

<u>Number of patients in the Core Registry:</u>
5225 cases

Full report at eurreb.eu

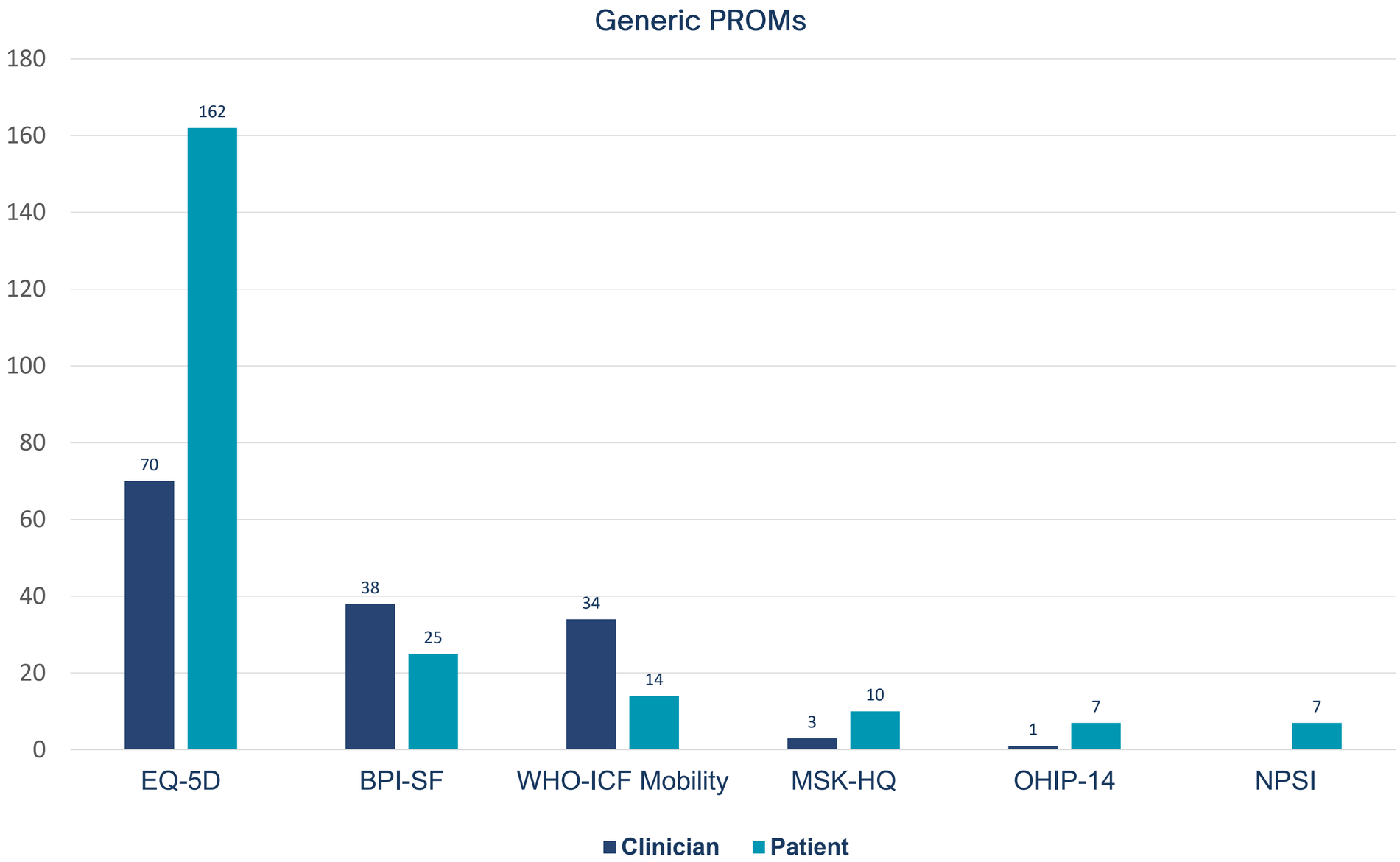
Cases Distribution in the Core Registry by MTG

TOTAL NUMBER OF CASES BY MTG (N=5225)



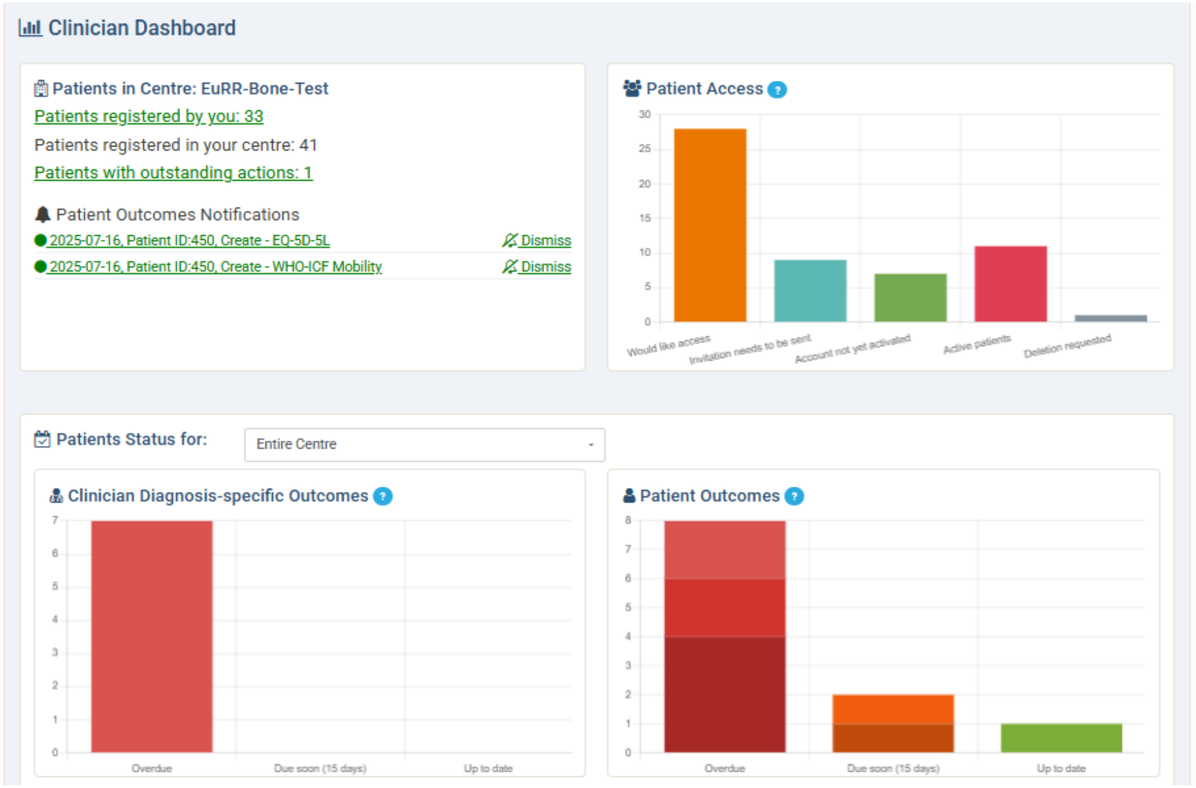
Full report at eurreb.eu

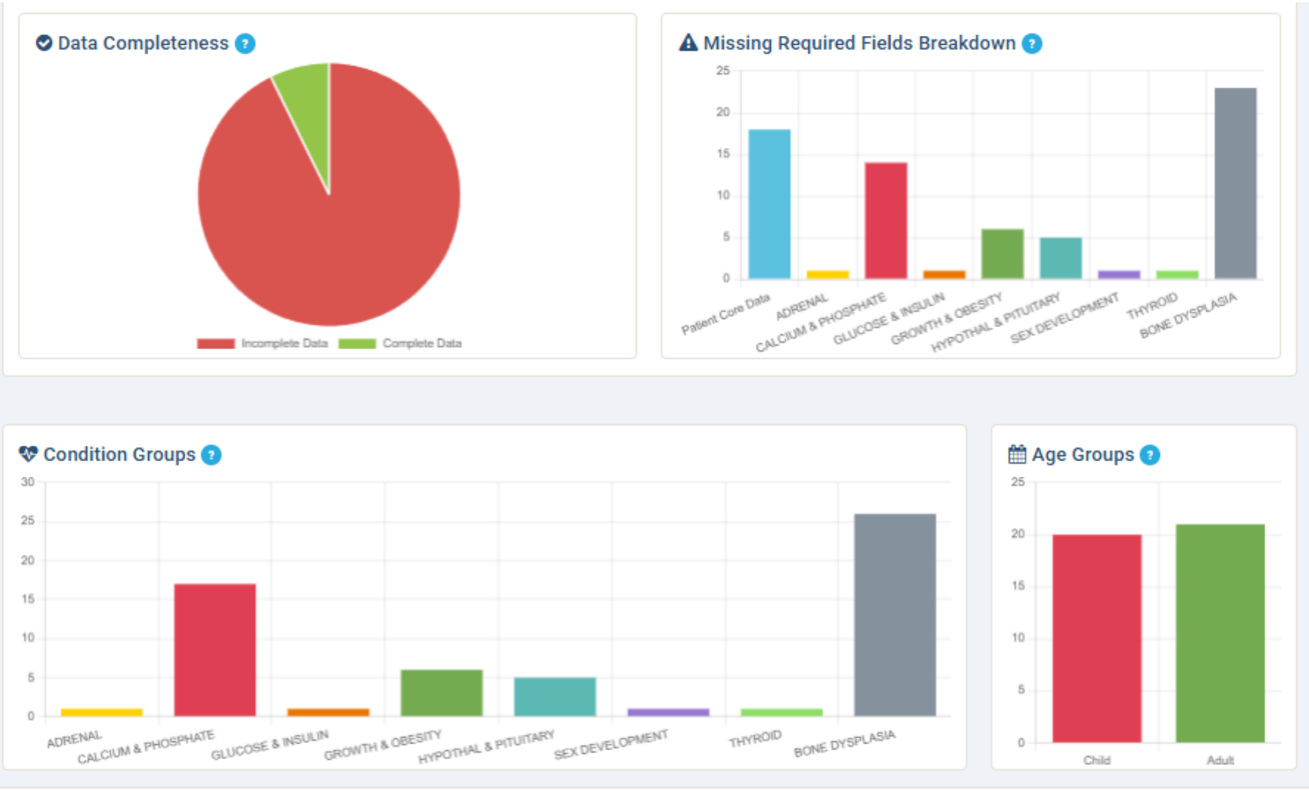
Patient-Reported Outcome Measures (PROMs)



Platform Developments – Data Completeness

New





The Condition-Specific Modules

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Condition-Specific Modules and Active Studies

CSM	MTG	Year	Countries	Centres	Study group leads	Active study
iPPSD/PHP	MTG2	2021	3	4	Agnès Linglart/Diana-Alexandra Ertl	Natural History of iPPSD/PHP 2021-2026
Pituitary Tumour	MTG6	2021	10	17	Alberto Pereira (1-3) Nienke Biermasz (4) Gerald Raverot (5)	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 4. Prolactinoma 5. The Clinical Usefulness of the Trouillas Classification
Achondroplasia	MTG 5	2022	4	4	Klaus Mohnike/ Ines Alves Carmen Vleggeert-Lankamp	Spinal deformities in Achondroplasia
FD/MAS		2022	12	16	Natasha Appelman-Dijkstra	Natural History of FD/MAS
Osteogenesis Imperfecta		2022	11	16	Natasha Appelman-Dijkstra (1) Wolfgang Högler (2,3)	1.Natural History of adults with OI 2. Natural History of children with OI 3. Pain in OI
Parathyroid Carcinoma	MTG2	2022	3	3	Maria Luisa Brandi	Natural History of Parathyroid Carcinoma
Rare Hypophosphataemia	MTG2	2023	6	6	Agnès Linglart/Diana-Alexandra Ertl	2022-2026
Melorheostosis		2023	2	2	Martine Cohen-Solal	Natural History of Melorheostosis
Rare Obesity	MTG5	2023	2	2	Erica van den Akker	2022-2026

Condition-Specific Modules Active and under Development

CSM	MTG	Year	Countries	Centres	Study group leads	Active study
Gender Incongruence	MTG7	2023	4	5	Martine Cools/Silvia Ciancia	Current status of healthcare for transgender adolescents across Europe
Paediatric Differentiated Thyroid Carcinoma	MTG8	2024	11	19	Hanneke van Santen/ Sarah Clement	Natural history of paediatric differentiated thyroid carcinoma
Langerhans Cell Histiocytosis	Sys	2025	2	2	Polyzois Makras	Natural History of endocrine and bone complications of LCH
Chronic Nonbacterial Osteitis (CNO)		2025	3	3	Elizabeth Winter	Under development
Women's Reproductive Health - Pregnancy and Rare Endocrine and Bone Conditions **LAUNCHED April 15, 2026**		2026	-	-	Tim Korevaar (1) and Natasha Appelman-Dijkstra/Tenna Toft/Claudia Finis (2)	Outcomes of pregnancy in rare endocrine (1) bone and mineral (2) conditions
Multiple Endocrine Neoplasia type 1 (MEN-1) **LAUNCHED March 2026**	MTG4	2025	-	-	Frederic Castinetti, Gerlof Valk, Maria Luisa Brandi	Natural History of MEN-1
Hypoparathyroidism **LAUNCHED April 2, 2026**	MTG2	2026	-	-	Are currently formed	Under development

Study Group Policy Update

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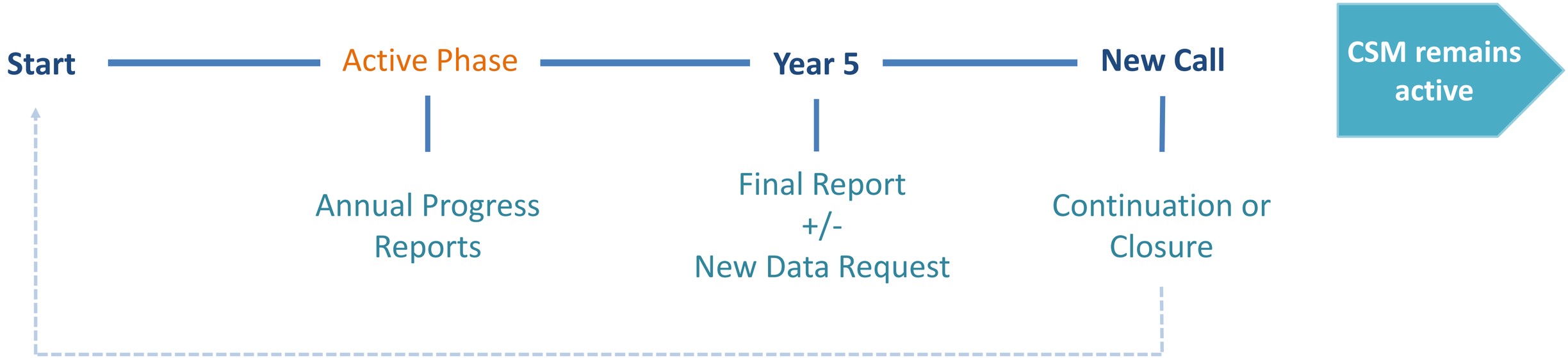
Lifecycle and Renewal of CSM-Based Studies



The EuRREB Participating Centre Voucher

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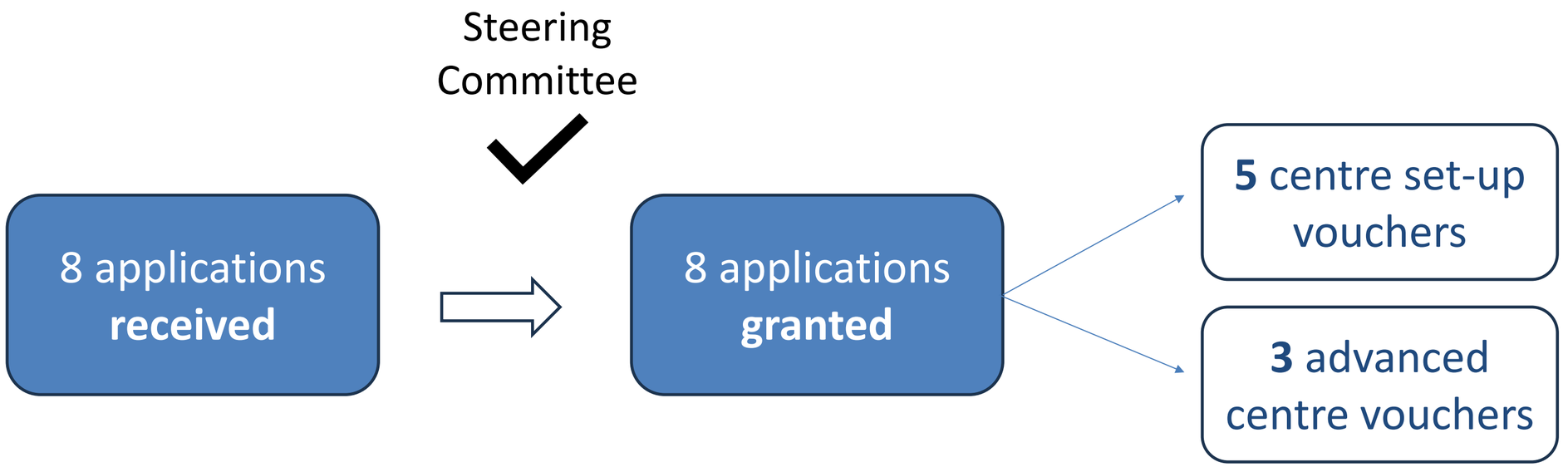
Results First Call – October 2025



<u>Second call</u>
Status: closed - submission deadline April 19, 2026

<u>Specific calls</u>
Hypoparathyroidism module has sponsoring for vouchers
Status: call in preparation; expected deadline 1 June 2026

Publications 2025

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2025

1. **Feasibility of transition research in pituitary disease using patient registries: a EuRREB secondary survey** 2025 | Savi Shishkov, Violeta Iotova, Iris Pelsma, Ana Luisa Priego Zurita, Nienke Biermasz, On Behalf Of The Endo-ern Pituitary Transition Of Care Study Group, And Faisal Ahmed

2. **Gender incongruence module** 2025 | Silvia Ciancia, Mariya Cherenko, Daniel Klink, Kanetee Busiah, Aneta Gawlik-starzyk, Wiktoria Kempinska, Hedi L. Claahsen-van Der Grinten, S. Faisal Ahmed, Sabine E. Hannema, And Martine Cools

3. **Developing a standard dataset in the European registries for rare endocrine and bone conditions – a melorheostosis dataset** 2025 | Natasha M. Appelman-Dijkstra, Mariya Cherenko, Gavin P. R. Clunie, Thomas Funck-brentano, Corinna Grasemann, Adalbert Raimann, Willem F. Lems & Martine Cohen-Solal.

4. **Developing a standardised dataset for natural history studies in fibrous dysplasia/McCune-Albright syndrome** 2025 | Ana Luisa Priego Zurita, Oana O. Bulaicon, Jillian Bryce, Nerea Arrieta, Magdalena Caballero Campos, Mariya Cherenko, Gaby Doxiadis, Corinna Grasemann, M. Kassim Javaid, Helen McDevitt, Stijn W. Van Der Meeren, Diana Ovejero Crespo, Luisa De Sanctis, Lothar Seefried, Annemarie A. Verrijn Stuart, Daniele Tessaris, Pieter Bas De Witte, Roland Chapurlat, S. Faisal Ahmed & Natasha M. Appelman-Dijkstra

Call for Participation

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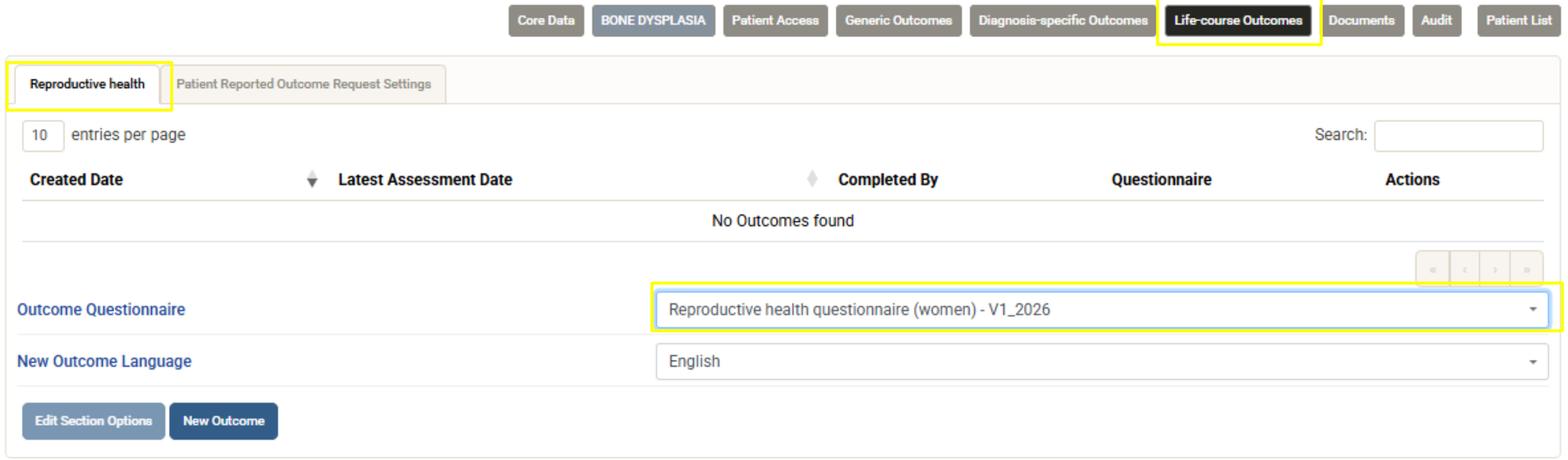
Women’s Reproductive Health Module

Contribute to the Pregnancy Module



Interested in developing a condition-specific study?

Get in touch!

Thank you!

Ways to contact us:

eurreb.eu

registries@lumc.nl

drop-in sessions via Zoom

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