



**EuRREB**

European Registries for Rare  
Endocrine and Bone conditions

# **Core Registry Annual Activity Report**

March 2025

## Introduction

The Core Registry (<https://eurreb.eu/registries/core-registry/>) was launched in June 2019 as part of the EuRRECa project, and since April 2020, it has also supported the EuRR-Bone project. Since March 2025, EuRRECa and EuRR-Bone have merged into a single project: the European Registries for Rare Endocrine and Bone conditions (EuRREB).

The platform includes patient access and reporting of patient and clinician-reported generic or condition-specific outcomes. Reporting by patients can occur in their preferred language. The Registry has ethics approval, and information sheets, and consent forms are available in multiple languages on its website. (<https://eurreb.eu/registries/ethics-approval/>).

## Aim of this Report

To describe the activity of the Core Registry between June 2019 and January 2025, with a particular focus on developments since our [last annual report](#).

## Results

Since January 2024, an additional **21** centres have registered, bringing the **total number to 135 registered centres** (with 323 registered users). Of the 135 centres, 51 (38%) have entered data in the Core Registry and are classified as 'Active Centres'.

These centres are located in 20 different countries, 18 are based in Europe (four of them are non-EU members), othertwo are Malaysia and Egypt. The highest number of participating centres per country is registered in Italy (nine centres), followed by the Netherlands (six centres) and France (five centres). The total amount of participating centres in the Core Registry and entered cases per country are shown on the map in figure 1.

Of these 51 centres, 15 are a combination of Endo-ERN and ERN BOND centres, 16 are exclusive Endo-ERN centres, four exclusive ERN BOND, and 16 are not related to ERNs (European research networks).

By January 1<sup>st</sup> 2025, 3980 records had been registered. The number of patient records increased by more than 70% compared to 2023 (Fig. 2). The biggest number of cases were entered in the Netherlands (1299 cases), followed by Italy (707 cases) and France (413 cases). Twenty-five centres (49%) have entered more than 10 cases and 3914 cases in total (98%), the median in reporting is seven cases per centre (range 1-767). The distribution of cases within condition groups differs a lot: ranging from six cases in 'Genetic Disorders of Glucose & Insulin Homeostasis' to 1286 cases in 'Hypothalamic & Pituitary Conditions'. "Bone Dysplasia" group is the second leading in number of cases MTG (Fig. 3). In 2024, the 'Hypothalamic & Pituitary Conditions', 'Bone Dysplasia' and 'Sex Development' MTGs showed an impressive increase in numbers in comparison to 2023.

The Core Registry gives the possibility to the patients to have their own account. So far 1298 from 3980 patients (33%) would like to have access to the platform. In total 345 (27%) patients with the interest to have an access, have activated their account on the Core Registry platform.

Patient-reported outcome measures (PROMs) have been filled in on the Core Registry platform by both clinicians and patients (Fig 4). The most frequently used are EQ-5D for the assessment of quality of life, Brief Pain Inventory Short Form (BPI-SF) and WHO-ICF mobility questionnaire. Several other questionnaires (in multiple languages) are also available on the platform such as MSK-HQ (Musculoskeletal Health Questionnaire), OHIP-14 (Oral health impact profile), PHQoL (Quality-of-Life Questionnaire for Patients with Primary Hyperparathyroidism) and since 2024 – NPSI (Neuropathic Pain Symptom Inventory).



Fig. 1 – The number of centres and cases per country

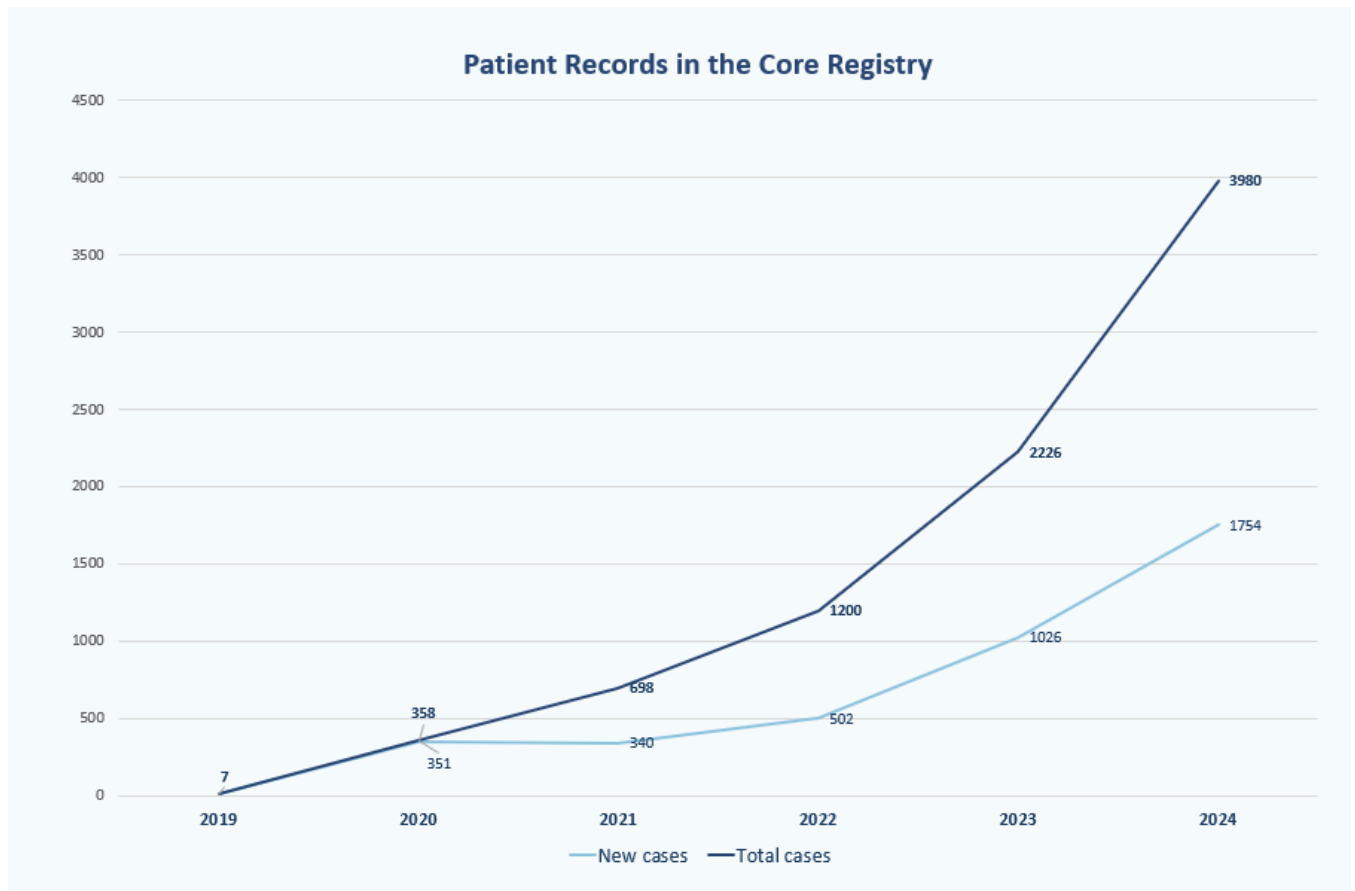


Fig. 2 – The number of patient records in the registry over time

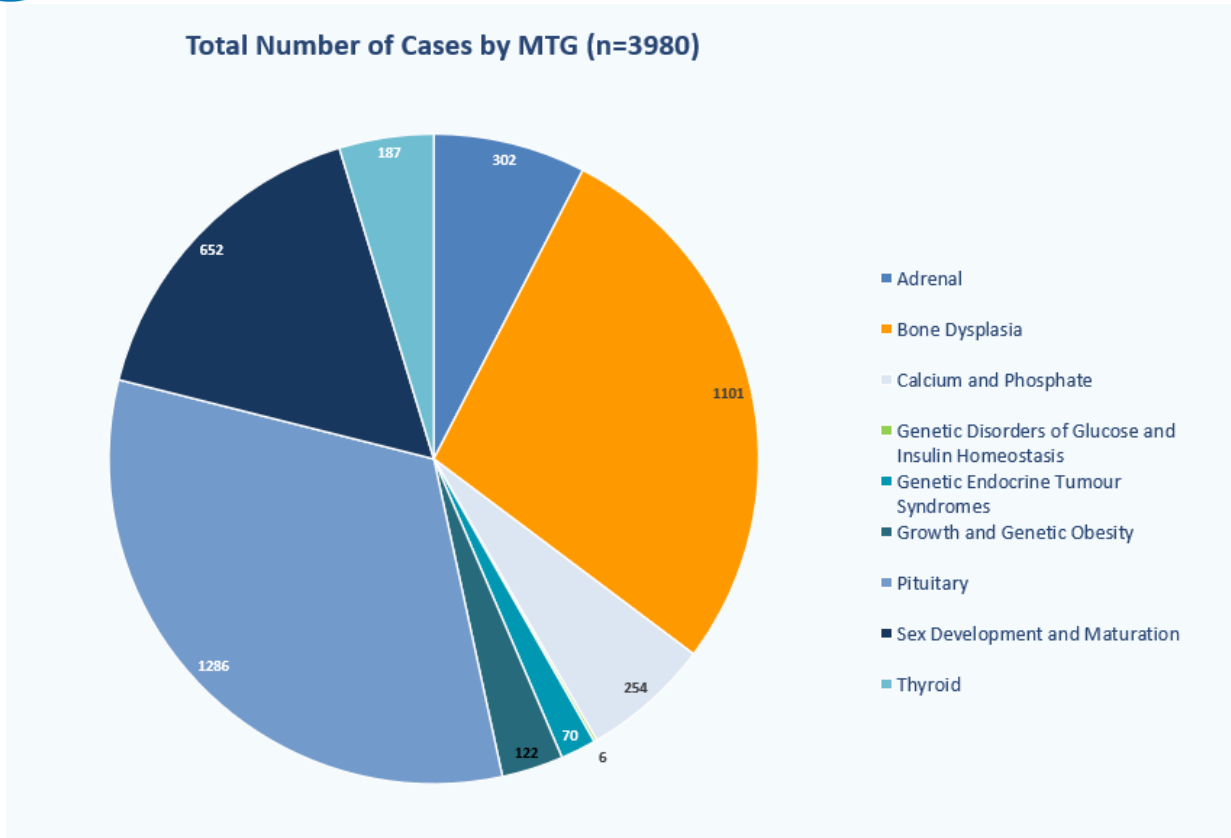


Fig. 3 - The proportion of patients that have been registered with the broad group of conditions or main thematic groups (MTGs) within Endo-ERN and ERN BOND

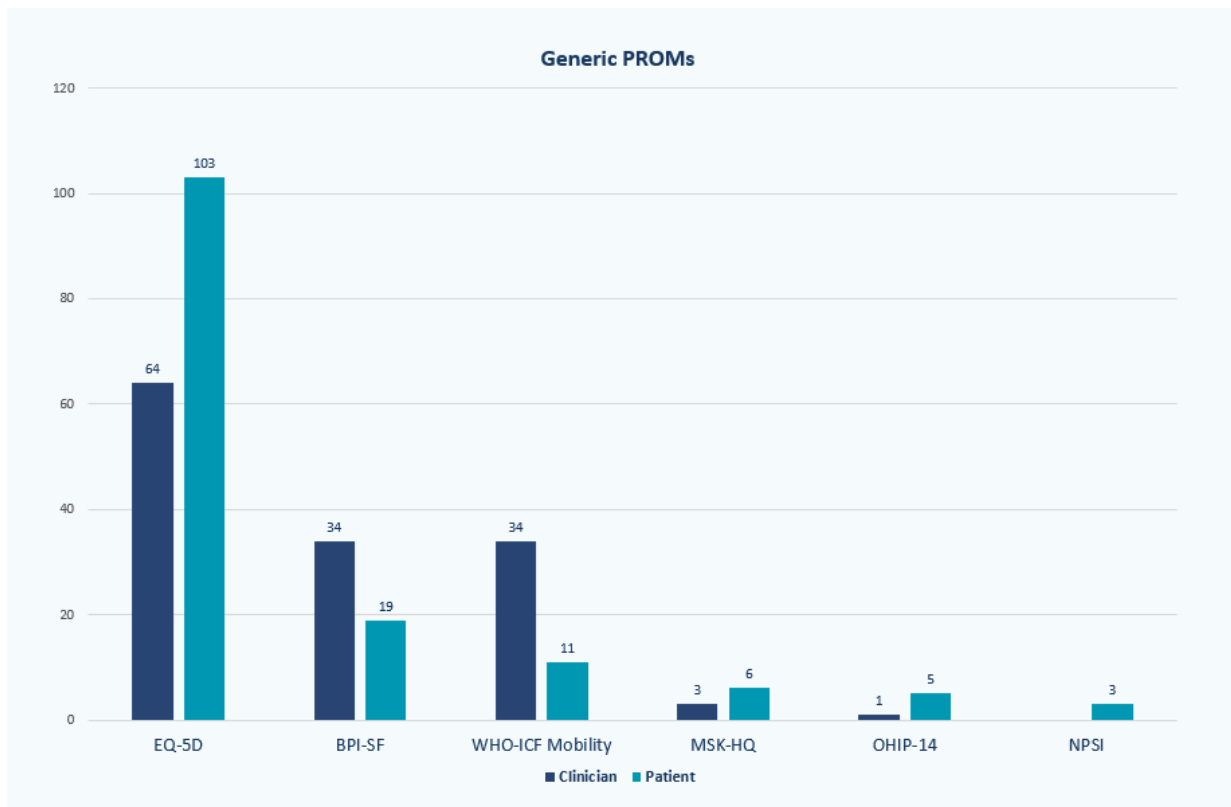


Fig. 4 - Patient-reported outcome measures (PROMs), filled in by clinician or patient

From 2021 to the end of 2024, 11 condition-specific modules have been launched on the Core Registry platform, covering six of the nine MTGs, including four within the Bone Dysplasia group. By January 2025, a total of 3264 outcomes had been recorded, representing an increase of more than 160 percent in 2024 compared to 2023.

At present, the most active modules are the Pituitary Tumour, Fibrous Dysplasia/McCune-Albright Syndrome (FD/MAS), and Gender Incongruence modules (Fig. 5). In 2024, a new condition group, Systemic and Rheumatological, was introduced to the platform. This group will include two new modules, currently in development, focusing on endocrine and bone complications of Langerhans Cell Histiocytosis and Chronic Nonbacterial Osteitis.

The registry continues to expand, with an increasing number of centres, users, and patients. A wide range of conditions across all ten main thematic groups are now represented, with patients registered from 51 active centres, 35 of which are within the related ERNs and 16 outside them. The Core Registry platform also provides a means for patients to complete Patient-Reported Outcomes (PROs), further enhancing the value of the data collected. To date, eleven condition-specific outcome measures have been developed within six MTGs, supporting improved understanding and management of these rare conditions.

N	CSM	MTG	Year	N patients in CR	Outcomes completed
1	iPPSD/PHP	MTG2	2021	35	13
2	Pituitary adenoma	MTG6	2021	804	1160
3	Achondroplasia	BD	2022	178	125
4	FD/MAS	BD	2022	653	1417
5	Osteogenesis imperfecta	BD	2022	145	74
6	Parathyroid carcinoma	MTG2	2022	14	15
7	Rare hypophosphataemia	MTG2	2023	85	65
8	Melorheostosis	BD	2023	20	18
9	Rare obesity	MTG5	2023	77	11
10	Gender Incongruence	MTG7	2023	313	311
11	Pediatric differentiated thyroid carcinoma	MTG8	2024	137	55
12	Langerhans Cell Histiocytosis	Sys	2024/2025	0	0
13	Chronic Nonbacterial Osteitis	Sys	2024/2025	0	0

*Fig 5. Condition-specific modules developed within the platform, year of release, total number of patients with this specific condition in the Core registry and number of completed outcomes per module. For one patient more than one outcome could be created.*

*(iPPSD/PHP – pseudohypoparathyroidism, FD/MAS - Fibrous Dysplasia/McCune-Albright Syndrome)*

### Recommendations for 2025

- The dissemination of results within Endo-ERN, ERN BOND, and beyond remains a priority, with efforts to encourage participation from additional interested centres.
- The Registries project management team will explore the possibilities to facilitate data entry by participating centres
- Data cleaning and module updates are planned for 2025.

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17<sup>th</sup> March, 2025



*Funded by the European Union. Views and opinions expressed are however those of the author(s) only and do not necessarily reflect those of the European Union or HaDEA. Neither the European Union nor the granting authority can be held responsible for them. Project: 101156495 (Endo-ERN), Project: 101157116 (ERN BOND); EU4H-2023-ERN2-IBA-01.*