EuRREB
European Registries for Rare Endocrine and Bone conditions

e-REC
Annual Activity Report

March 2026

registries@lumc.nl

eurreb.eu

The 2025 annual report on the activity within the e-Reporting of Rare Endocrine Conditions (e-REC) platform within the European Registries for Rare Endocrine and Bone conditions (EuRREB)

By: Mariya Cherenko, Jacqueline van der Blom-de Gunst, Ana Priego Zurita, Natasha Appelman-Dijkstra

Publication date: Thursday, March 5, 2026

Introduction

e-REC (e-Reporting of Rare Endocrine Conditions) is an electronic reporting platform that captures activity and allows for a better understanding of the occurrence of a wide range of rare endocrine and bone conditions. For clinical networks such as Endo-ERN and ERN BOND, the e-REC platform allows continuous reporting of core indicators of activity and enables these clinical networks to objectively map the conditions and related activity. However, the platform is open to all centres that look after people with such conditions and not just limited to reference centres within ERNs.

Participants can adapt the reporting setup and report any newly encountered cases of any of the condition groups that have been included in Endo-ERN and ERN BOND.

From July 2018 through December 2024, EuRREB's e-REC platform expanded substantially in both reach and volume, growing from 66 centres in 22 countries with 20,097 cumulative new cases by December 2022 to 101 centres in 30 countries with 36,914 cases by December 2023, and further to 113 centres in 32 countries with 56,573 cases by December 2024 (41,933 adult; 14,640 paediatric). This scale-up coincided with the launch of EuRR-Bone in 2020 and the mandated participation under the ERN continuous monitoring programmes of Endo-ERN in 2023 and ERN BOND in 2024. The mandatory engagement associated with the continuous monitoring programme substantially increased interaction with the platform, resulting in participation from 92% of Endo-ERN centres and 68% of ERN BOND centres.

e-REC reporting is ORPHAcode-based and supported by ongoing curation of EuRREB condition dictionaries, enabling harmonised surveillance across MTGs and centres(EuRREB). The platform has also been used for targeted initiatives, including mapping COVID-19 infection in patients with rare endocrine conditions (in collaboration with the ESE Rare Disease Committee), and has evolved to capture greater diagnostic granularity. For instance, by 2024/2025 reporting, the platform included more detailed pituitary adenoma diagnostic categories, enabling more granular capture of pituitary case mix (e.g., non-functioning adenoma, prolactinoma, acromegaly, Cushing disease as specific adenoma diagnoses). In 2024, e-REC expanded thematic coverage with the introduction of MTG10 (Systemic & Rheumatological), demonstrating the platform's capacity to incorporate new domains aligning collaborations with other ERNs like ERN-EuroBloodNET for the endocrine and bone complications of Langerhans cell histiocytosis or with the European Calcified Tissue Society for Chronic Non-bacterial Osteitis. Previous annual reports can be downloaded from our website.

This annual report has been developed to describe the current activity in e-REC starting July 2018 to the most recent reporting year ending December 2025 inclusive.

Results

In 2025, a total of 122 centres from 32 countries had reported 23,341 new cases of rare endocrine or bone conditions in e-REC making the total of cases within the platform 79,896. Since January 2025, 9 new centres have joined the platform. Among those 32 countries, 29 are located in Europe. Of the 122 centres, 67 are members of Endo-ERN, 30 are members of both Endo-ERN and ERN BOND, five centres are ERN BOND members, and 20 centres are not related to either Endo-ERN or ERN BOND. Since a centre can report in Endocrine conditions (both paediatric and adult) as well as Bone conditions (both paediatric and adult), a centre can have more than one reporter, albeit one reporter or a maximum of two reporters (e.g., a bone-reporter and an endocrine-reporter) is preferred to overcome double counting. At the moment the total number of e-REC reporters is currently 383.
The total amount of participating in e-REC centres in different countries is shown on the map in figure 1, with Italy having the largest number of centres, n=23, followed by Germany, n=14, and Spain, n=9.

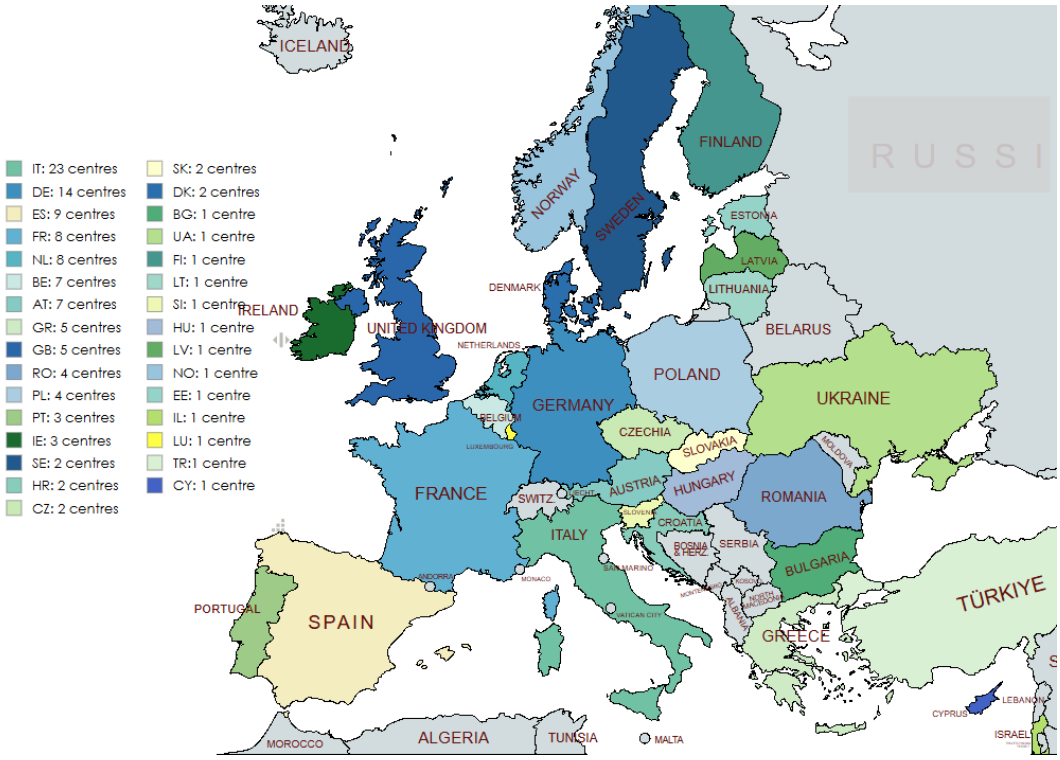

Fig. 1 – Number of centres in each country that are active (i.e. have submitted one or more returns between July 2018 and December 2025)

Currently, 99 centres are reporting new cases in the paediatric age group and 98 in the adult age group.
Reporters can report in 10 different Main Thematic Groups (MTGs) subdivided in children (<18 years old) and adults (≥18 years old). The distribution of cases within MTGs differs between children and adults and this remains stable over the years. Conditions within the 'Sex Development', 'Growth & Obesity' and 'Thyroid' condition groups are most commonly reported in children (figure 3), whereas in adults the most commonly reported conditions are within the 'Hypothalamic and Pituitary', 'Thyroid' (70%) and 'Adrenal' condition groups (figure 4). In 2025, the 'Hypothalamic and Pituitary', 'Bone Dysplasia' and 'Thyroid' MTGs showed an impressive increase in numbers in children's group in comparison to 2024 (46%, 70% and 43% increase respectively). In adults a peak was noticed in the 'Bone Dysplasia', 'Thyroid' and 'Adrenal' MTGs (46%, 30% and 42% increase respectively), and there is still an increasing number of cases in the 'Hypothalamic and Pituitary' and 'Sex Development' condition groups.

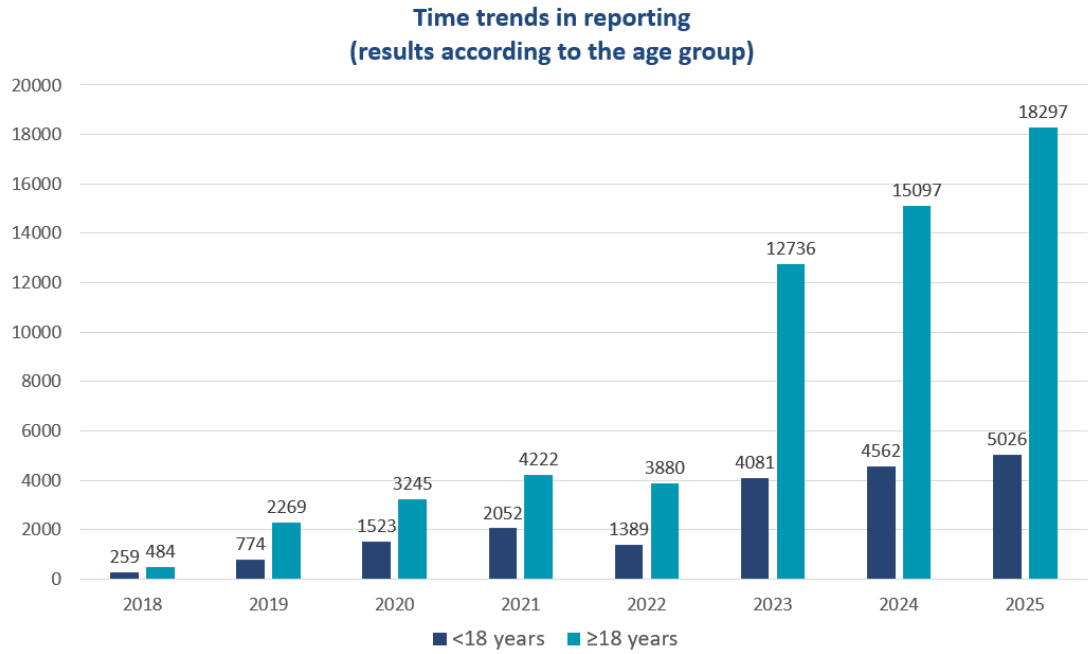

Fig. 2 – The trends in cases reported between July 2018 and December 2025

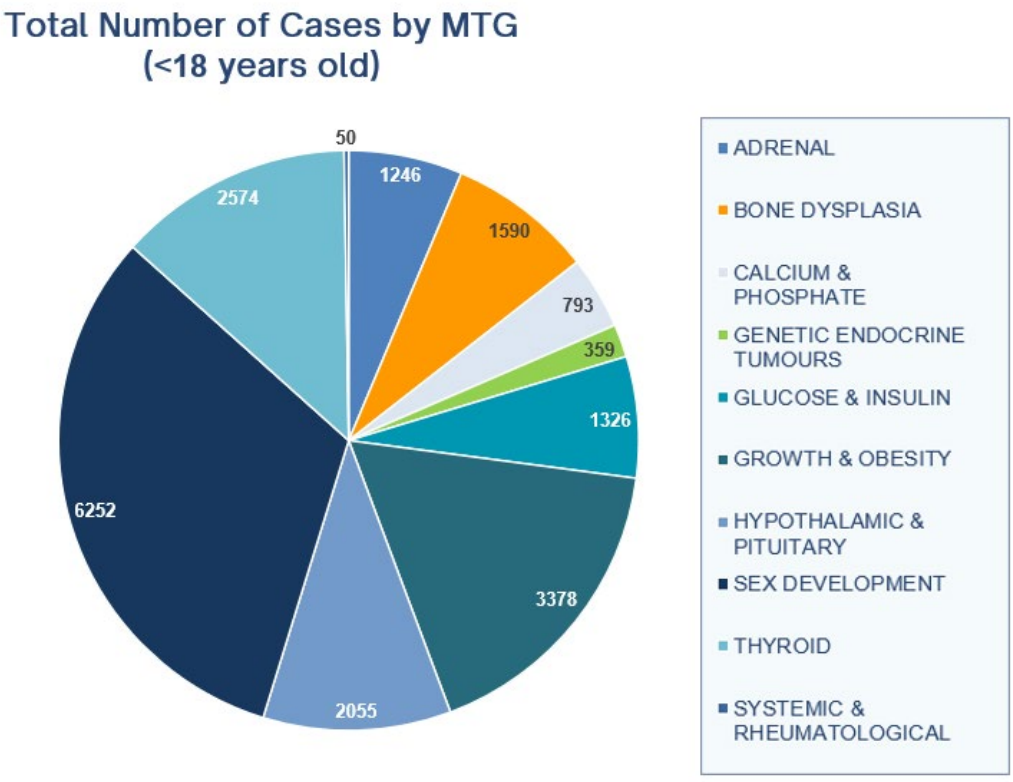

Fig. 3 – Distribution of cases between different MTGs (Main Thematic Groups) in children

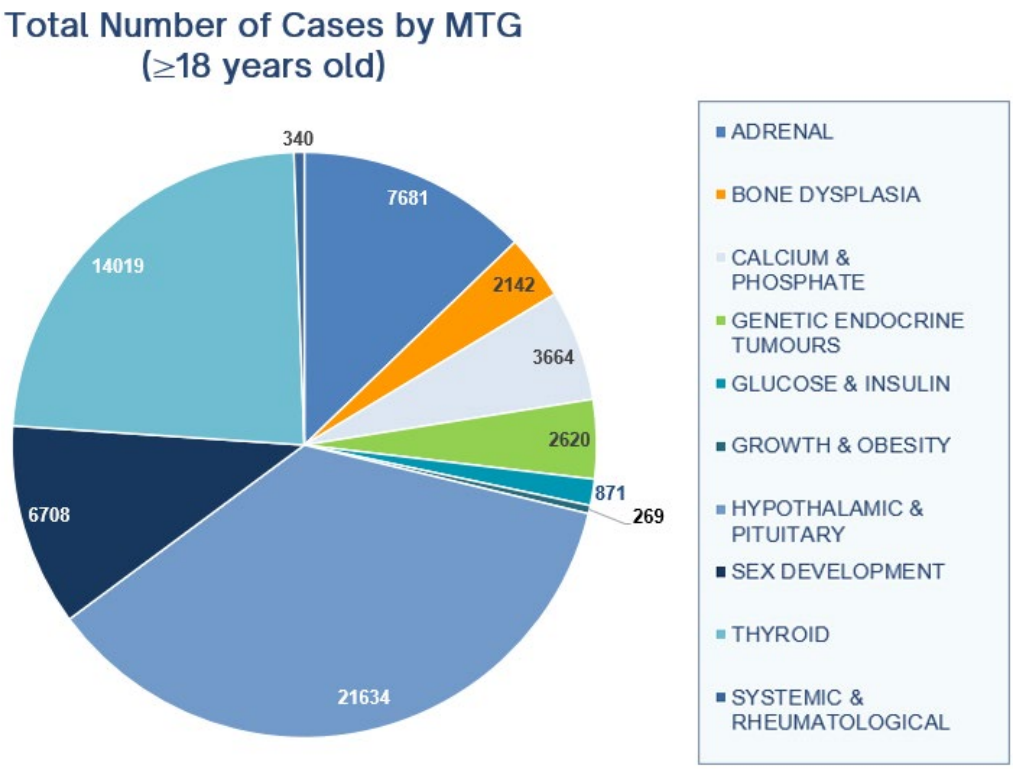

Fig. 4 – Distribution of cases between different MTGs (Main Thematic Groups) in adults.

Results by Main Thematic Group

Outcomes of Main Thematic Group 1: Adrenal

In total, 8,927 cases were reported by 76 centres from 26 countries (2,833 new cases in 2025). Among these, 1,246 were paediatric cases and 7,681 were adult cases.

In children the most reported condition was congenital adrenal hyperplasia (n=762, 61%), followed by primary adrenal insufficiency (n=327, 26%), adrenal Cushing syndrome (n=70, 6%), sporadic pheochromocytoma/ paraganglioma (n=47, 4%), adrenocortical carcinomas (n=27, 2%) and familial hyperaldosteronism (n=13, 1%). The median of cases reported by centre was 14 (IQR=16), with a maximum of 187 cases reported by one of the centres.

In adults the most reported cases were sporadic pheochromocytoma/paraganglioma (n=2,399, 31%), followed by adrenal Cushing syndrome (n=1,559, 20%), primary adrenal insufficiency (n=1,415, 18%), congenital adrenal hyperplasia (n=970, 13%), adrenocortical carcinomas (n=764, 10%) and familial hyperaldosteronism (n=574, 8%). The median of cases reported by centre was 78 (IQR=116), with a maximum of 1,103 cases reported by one of the centres since 2019.

Outcomes of Main Thematic Group 2: Calcium & Phosphate

In total, 4,457 cases were reported by 76 centres from 29 countries (1,177 new cases in 2025), among them 793 paediatric cases and 3,664 adult cases.

In children the most reported condition was X-linked hypophosphatemia (n=212, 27%), followed by IPPSD/ Pseudohypoparathyroidism (n=163, 21%), hypoparathyroidism (n=149, 19%), hyperparathyroidism including parathyroid cancer (n=57, 7%), hypophosphataemic rickets (n=52, 7%), familial hypocalciuric hypercalcaemia

(n=52, 7%), and other conditions. The median of cases reported by centre was 10 (IQR=14), with a maximum of 110 cases reported by one of the centres.

In adults the most reported diagnoses were hyperparathyroidism including parathyroid cancer (n=1,955, 53%), followed by hypoparathyroidism (n=884, 24%), PTH independent hypercalcaemia (n=188, 5%), X-linked hypophosphatemia (n=180, 5%) and other conditions. The median number of cases reported by centre was 43 (IQR=91), with a maximum of 331 cases reported by one of the centres.

Outcomes of Main Thematic Group 3: Genetic Disorders of Glucose & Insulin Homeostasis

In total, 2,197 cases were reported by 55 centres from 27 countries (512 new cases in 2025), including 1,326 paediatric cases and 871 adult cases.

In children the most reported condition was rare diabetes (n=685, 52%), followed by hyperinsulinism (n=528, 40%) and insulin resistance syndrome (47, 4%). Also 41 cases (3%) of genetic and 17 cases (1%) of acquired lipodystrophy were reported. The median number of cases reported by centre was 18 (IQR=33), with a maximum of 183 cases reported by one of the centres.

In adults the most reported cases were in rare diabetes group (n=475, 54%), followed by insulin resistance syndrome (n=129, 15%) and hyperinsulinism (n=52, 6%). Also 180 cases (21%) of genetic and 35 cases (4%) of acquired lipodystrophy were reported. The median number of cases reported by centre was 20 (IQR=42), with a maximum of 104 cases reported by one of the centres.

Outcomes of Main Thematic Group 4: Genetic Endocrine Tumour Syndromes

In total, 2,979 cases were reported by 68 centres from 23 countries (811 new cases in 2025), including 359 paediatric cases and 2,620 adult cases.

In children the most reported condition was MEN Type 2 (n=137, 38%), followed by MEN Type 1 (n=112, 31%), Von Hippel Lindau syndrome (n=46, 13%), hereditary pheochromocytoma-paraganglioma (n=33, 9%), Carney complex (n=15, 4%) and other NETs (n=16, 5%). The median number of cases reported by centre was 7 (IQR=6), with a maximum of 61 cases reported by one of the centres.

In adults the most reported condition was hereditary pheochromocytoma-paraganglioma (n=999, 38%), followed by MEN Type 1 (n=557, 21%), MEN Type 2 (n=394, 15%), Von Hippel Lindau syndrome (n=212, 8%) and Carney complex (n=33, 1%). Other NETs were reported in 425 cases (16%). The median of cases reported by centre was 31 (IQR=46), with a maximum of 257 cases reported by one of the centres.

Outcomes of Main Thematic Group 5: Growth & Genetic Obesity Syndromes

In total, 3,647 cases were reported by 62 centres from 24 countries (1,285 new cases in 2025), including 3,378 paediatric cases and 269 adult cases.

In children the most reported condition was rare genetic obesity (n=850, 25%), followed by Prader Willi syndrome and Prader Willi-like syndrome (n=537, 16%), Noonan syndrome (n=527, 16%), Silver-Russell syndrome (n=512, 15%), overgrowth syndrome (n=472, 14%), Beckwith-Wiedemann syndrome (n=411, 12%), GH resistance syndromes (n=67, 2%). Till now 2 cases of ROHHAD syndrome were reported. The median number of cases reported by centre was 20 (IQR=26), with a maximum of 601 cases reported by one of the centres.

In adults the most reported condition was also Prader Willi syndrome and Prader Willi-like syndrome (n=127, 47%). Other conditions were rarely reported in adults: rare genetic obesity (n=51, 19%), overgrowth syndrome (n=27, 10%), Silver-Russell syndrome (n=24, 9%), Beckwith-Wiedemann syndrome (n=20, 7%), and Noonan

syndrome (n=19, 7%). The median of cases reported by centre was 6 (IQR=12), with a maximum of 50 cases reported by one of the centres.

Outcomes of Main Thematic Group 6: Hypothalamic & Pituitary Conditions

In total, 23,689 cases were reported by 79 centres from 29 countries (6388 new cases in 2025), including 2,055 paediatric cases and 21,634 adult cases.

In children the most reported condition was congenital hypopituitarism (n=1,033, 50%), followed by acquired hypopituitarism (n=629, 31%), pituitary adenoma (n=321, 16%) and craniopharyngioma (n=72, 3%). The median number of cases reported by centre was 21 (IQR=34), with a maximum of 277 cases reported by one of the centres.

In adults the most reported condition was pituitary adenoma (n=17,135, 79%), followed by acquired hypopituitarism (n=3,602, 17%), craniopharyngioma (n=464, 2%) and congenital hypopituitarism (n=433, 2%).
Since 2023, specific diagnoses of pituitary adenomas have been implemented in the e-REC registry. This led to more detailed reporting – 3,268 new cases of non-functioning pituitary adenoma (15%), 2,500 new cases of prolactinoma (12%), 1,261 new cases of acromegaly (6%), and 1,032 new cases of Cushing disease (5%) were registered. The median number of cases reported by centre was 271 (IQR=364), with a maximum of 1,731 cases reported by one of the centres.

Outcomes of Main Thematic Group 7: Sex Development & Maturation

In total, 12,960 cases were reported by 65 centres from 26 countries (3,151 new cases in 2025), among them 6,252 paediatric cases and 6,708 adult cases.
In children the most reported condition was gender incongruence (n=2,744, 44%), followed by XY DSD (n=1,301, 21%), chromosomal DSD (n=1251, 20%), XX DSD (n=628, 10%), isolated congenital normosmic hypogonadotropic hypogonadism (n=196, 3%), isolated congenital anosmic hypogonadotropic hypogonadism (n=132, 2%). The median of cases reported by centre was 32 (IQR=68), with a maximum of 2,497 cases reported by one of the centres.

In adults the most reported condition also was gender incongruence (n=4,487, 67%), followed by chromosomal DSD (n=921, 14%), XX DSD (n=430, 6%), XY DSD (n=298, 4%), isolated congenital normosmic hypogonadotropic hypogonadism (n=353, 5%), isolated congenital anosmic hypogonadotropic hypogonadism (n=219, 3%). The median number of cases reported by centre was 55 (IQR=243), with a maximum of 1,367 cases reported by one of the centres.

Outcomes of Main Thematic Group 8: Thyroid

In total, 16,593 cases were reported by 73 centres from 27 countries (5,499 new cases in 2025), including 2,574 paediatric cases and 14,019 adult cases.
In children the most reported condition was congenital hypothyroidism (n=1,847, 72%), followed by non-metastatic thyroid carcinoma (n=384, 15%), thyroid hormone signalling disorders (n=157, 6%), congenital hyperthyroidism (n=160, 6%). In summer 2025 specific diagnosis of Paediatric-onset Graves disease has been implemented in the e-REC registry, since that time 26 new cases (1%) were reported in this category.
The median number of cases reported by centre was 20 (IQR=52), with a maximum of 192 cases reported by one of the centres.
In adults the most reported condition was non-metastatic thyroid carcinoma (n=13,613, 97%), followed by thyroid hormone signalling disorders (n=266, 2%), congenital hypothyroidism (n=112, 0%), congenital hyperthyroidism (n=28, 0%). The median number of cases reported by centre was 150 (IQR=242), with a maximum of 2,441 cases reported by one of the centres.

Outcomes of Main Thematic Group 9: Bone Dysplasia

In total, 3,732 cases were reported by 61 centres from 24 countries (1,317 new cases in 2025), including 1,590 paediatric cases and 2,142 adult cases.

In children the most reported condition was osteogenesis imperfecta (n=368, 23%), followed by achondroplasia (n=215, 14%), fibrous dysplasia/McCune-Albright syndrome (n=152, 10%), multiple osteochondroma (n=61, 4%), dysostosis (n=56, 4%), dyschondrosteosis/Leri-Weill (n=44, 3%), short stature due to ACAN mutation (n=30, 2%) and spondyloepiphyseal dysplasia congenita/SEDC (n=29, 2%). Other groups of conditions reported frequently in this MTG are - rare bone disease related to a common gene or pathway defect (n=191, 12%), primary bone dysplasia (PBD) with disorganized development of skeletal components (n=58, 4%), PBD with defective bone mineralisation (n=56, 4%), PBD with reduced bone density (n=55, 4%), PBD with increased bone density (n=23, 1%). The median number of cases reported by centre was 13 (IQR=38), with a maximum of 295 cases reported by one of the centres.

In adults the most reported condition was fibrous dysplasia/McCune-Albright syndrome (n=780, 37%), followed by osteogenesis imperfecta (n=489, 23%), multiple osteochondroma (n=74, 4%) and osteonecrosis (n=59, 3%). Other groups of conditions reported frequently in this MTG are - PBD with defective bone mineralisation (n=290, 14%), PBD with reduced bone density (n=169, 8%), osteopetrosis and related disorders (n=49, 2%). The median number of cases reported by centre was 10 (IQR=38), with a maximum of 450 cases reported by one of the centres.

Outcomes of Main Thematic Group 10: Systemic & Rheumatological

In the summer of 2024, this new MTG was added to platform to serve the needs of the reporters dealing with systemic and rheumatological conditions causing endocrine and bone complications. This MTG comprises now four conditions: Langerhans and non-Langerhans cell histiocytosis (LCH and non-LCH), chronic nonbacterial osteitis (CNO), isolated sternocostoclavicular hyperostosis (SCCH) and SAPHO syndrome.
In total, 390 cases were reported by seven centres from seven countries (350 new cases in 2025), including 50 paediatric cases and 340 adult cases.
LCH was reported in 240 adult patients and eight children, non-LCH was reported in 22 adult patients only, CNO was reported in 47 adults and 42 children, SCCH – in 24 adults, SAPHO syndrome in seven adults. The median number of cases reported by centre was 10 (IQR=52), with a maximum of 250 cases reported by one of the centres.

Conclusions

There is wide variability in the number of cases encountered for different conditions amongst different centres.
The condition group with the largest number of reporters is MTG6 (Hypothalamus & Pituitary) which also has the highest number of reported cases.
The number of centres that have registered to use the platform continues to increase as well as the number of centres that are actively reporting cases. The continuous monitoring programmes of Endo-ERN and ERN BOND have been important drivers of this increase during the last years. For Endo-ERN, 92% of HCPs is using e-REC, meaning that the platform is adherent to its key performance indicator (KPI) for this ERN. For ERN BOND however the KPI is not met, since 68% instead of the required 75% of HCPs are using the platform, the coming year we will continue to focus on this to make sure that this KPI is met in 2027. In 2026 changes will be made to the platform to facilitate the new KPI for ERNs which is monitoring foreign patients especially patients from Ukraine.
In 2025 no articles with e-REC data/use of the e-REC platform were published.

Publications related to e-REC

Title	Electronic reporting of rare endocrine conditions within a clinical network: results from the EuRRECa project
Date	October 30th, 2023
Issue name	Endocrine Connections
Issue number	v12.12
DOI	10.1530/ec-23-0434
Authors	Ali SR, Bryce J, Priego-Zurita AL, Cherenko M, Smythe C, de Rooij TM, Cools M, Danne T, Katugampola H, Dekkers OM, Hiort O, Linglart A, Netchine I, Nordenstrom A, Attila P, Persani L, Reisch N, Smyth A, Sumnik Z, Taruscio D, Visser WE, Pereira AM, Appelman-Dijkstra NM & Ahmed SF

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.