

**European Registry for Rare Bone and Mineral Conditions (EuRR-Bone):  
Results from the Platform for e-Reporting of Rare Conditions (e-REC).**

**Introduction**

**e-REC** is an electronic reporting system which captures activity and allows for a better understanding of the occurrence of the rare conditions covered within networks such as **ERN BOND** and **Endo-ERN**. This is a joint platform originally set up by **EuRRECa** and has been specifically adapted for **EuRR-Bone**.

The platform is open to all centres that look after people with rare bone and mineral conditions. It allows continuous reporting of core indicators of activity and enables clinical networks to objectively map the conditions and related activities.

**Aim**

This report has been developed to describe the current activity in e-REC during the period of April 2020 to December 2022.

**Methods**

The e-REC platform issues invitations to clinicians who have participated in e-REC from April 2020 to December 2022 to ask them to complete a monthly return. Participants can create a reporting set up and report any newly encountered cases of any of the conditions that have been included in Endo-ERN, ERN-BOND and COVID-19 infections.

**Results**

Country	Centers	Country	Centers
 Austria	2	 Israel	1
 Belgium	6	 Italy	11
 Croatia	1	 Latvia	1
 Czechia	1	 Lithuania	1
 Estonia	1	 Netherlands	3
 Finland	1	 Romania	1
 France	1	 Spain	3
 Germany	8	 Sweden	1
 Hungary	1	 UK	4

*Table 1 – Number of centres in each country that are active (i.e. have submitted 1 or more returns between April 2020 and December 2022).*

By December 2022, a total of 48 centres from 18 countries had reported on e-REC. Of these, 13 are in Endo-ERN only, 6 in ERN Bond only, 17 are joint Endo-ERN and ERN-BOND affiliated centres and 12 are neither.

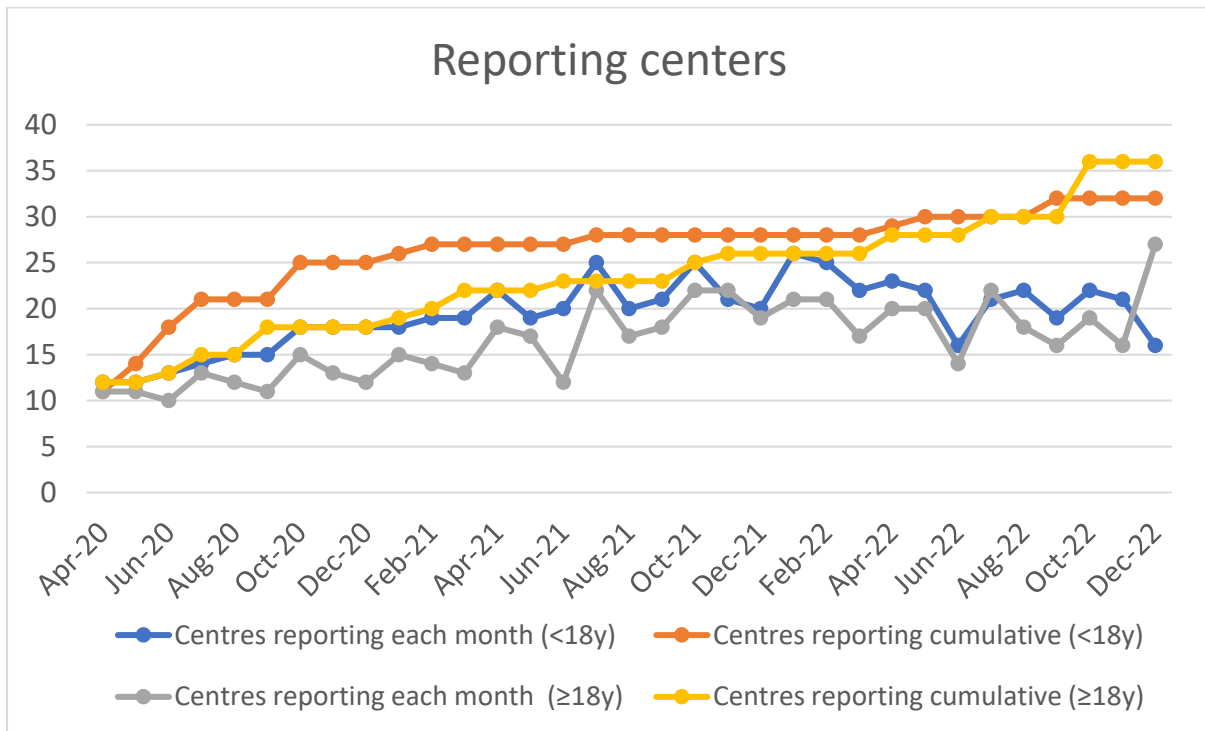


Fig.1 – Change in the number of active reporting centres between April 2020 and December 2022.

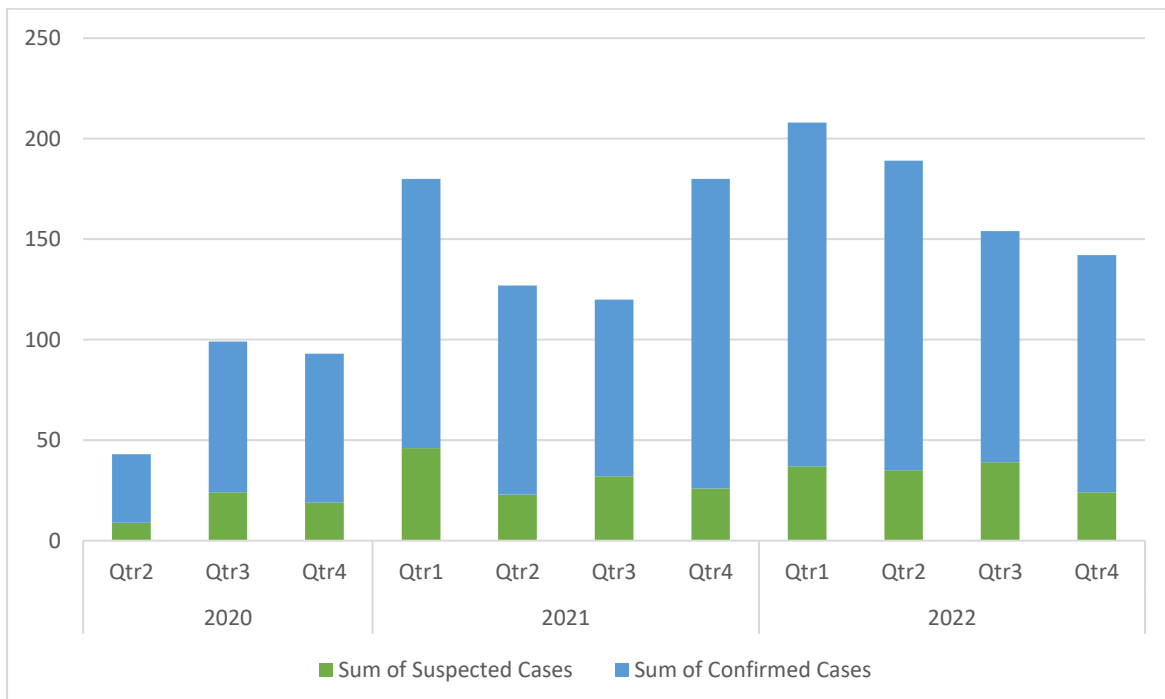


Fig.2 – Change in the number of suspected and confirmed cases in bone dysplasia and Calcium and phosphate between April 2020 and December 2022.

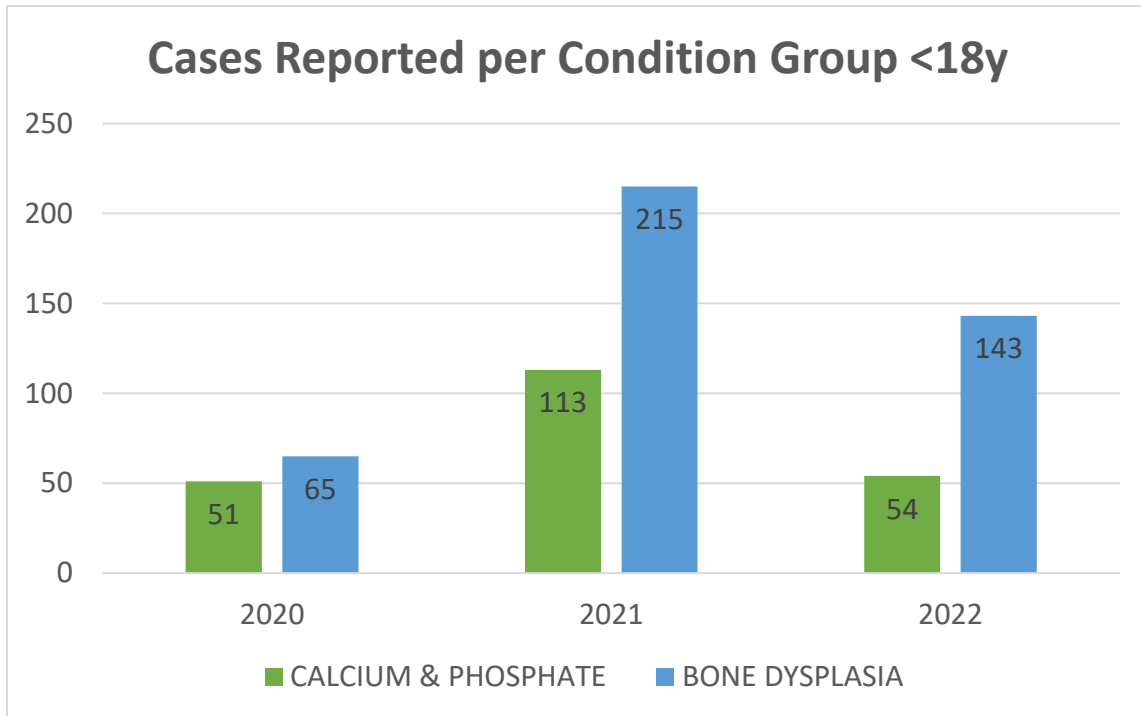


Fig.3 - Cases reported per group of conditions <18y.

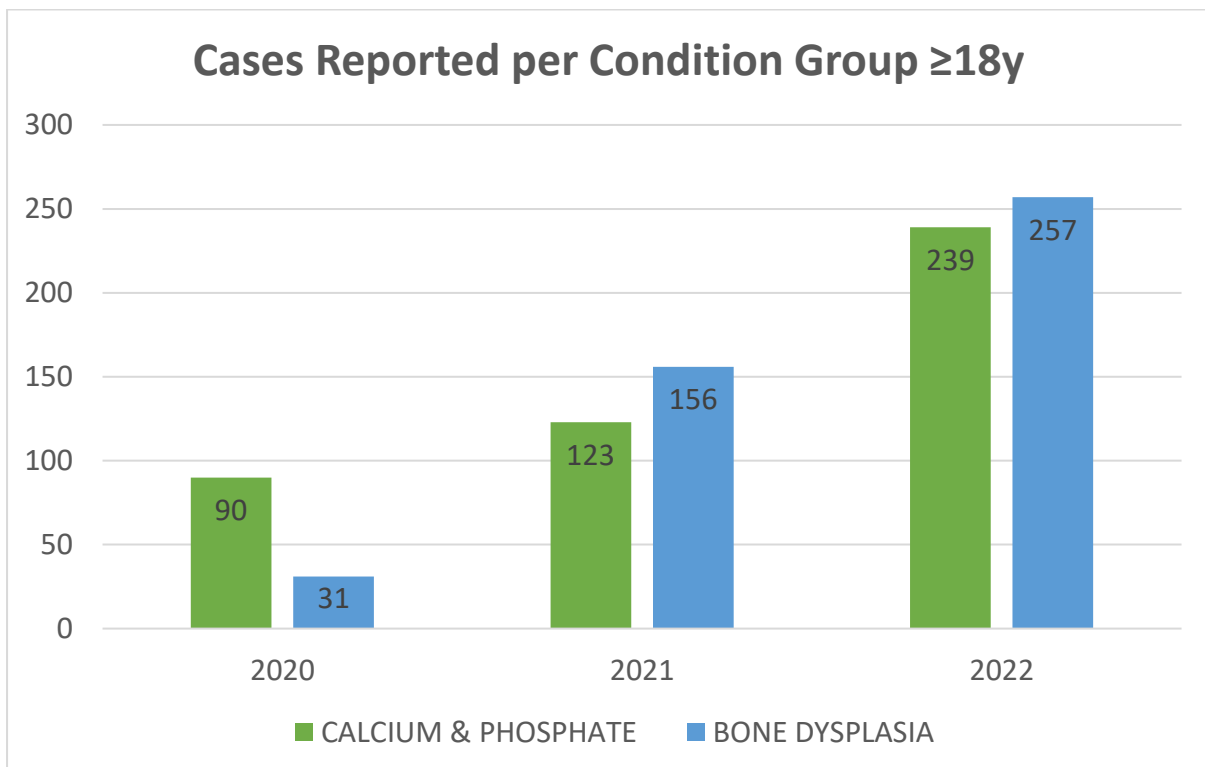


Fig.4 - Cases reported per group of conditions ≥18y.

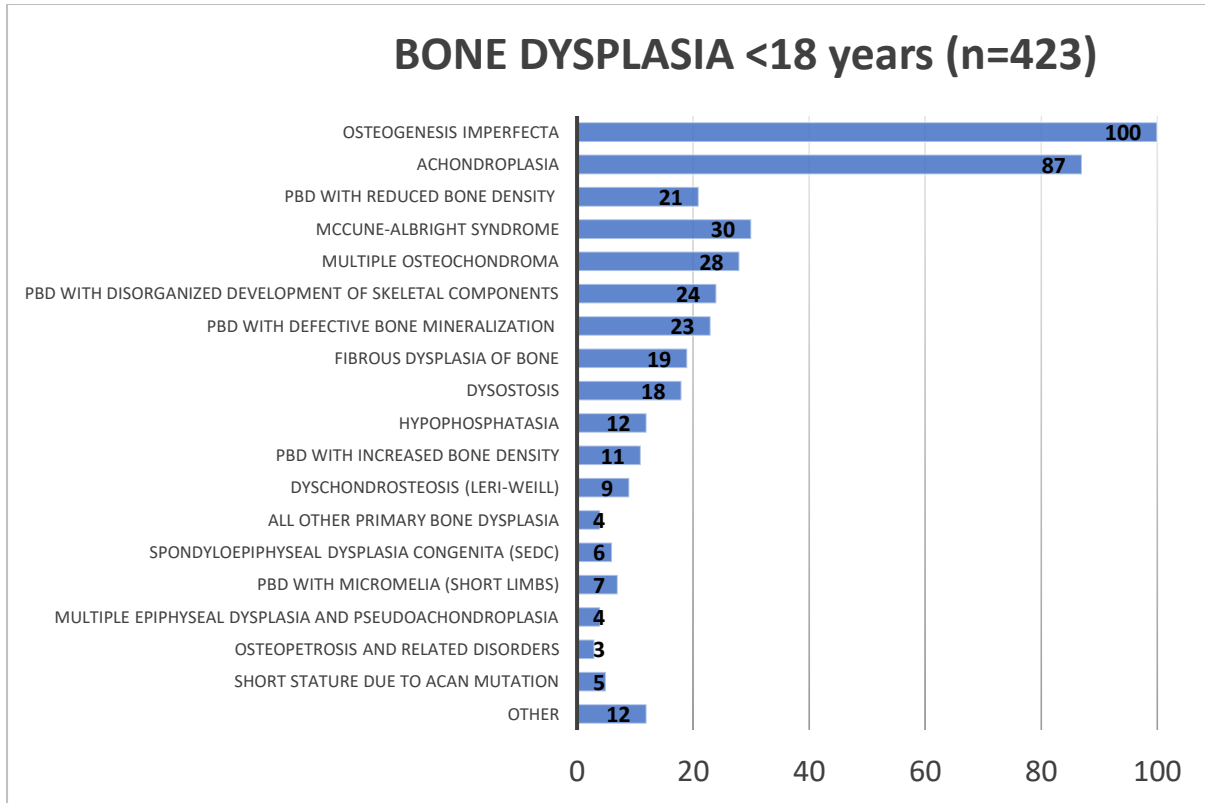


Fig. 5 - Specific diagnosis reported from April 2020 to December 2022 in <18y.

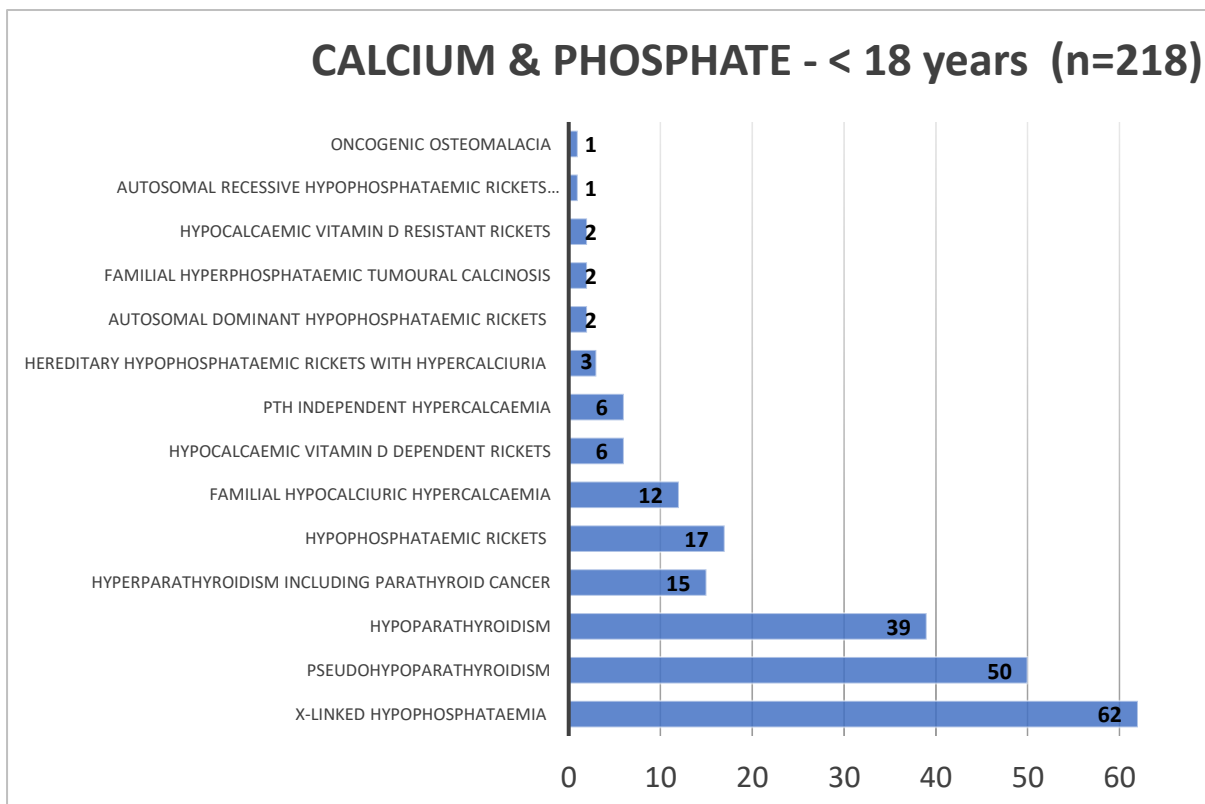


Fig.6 - Specific diagnosis reported from April 2020 to December 2022 in <18y.

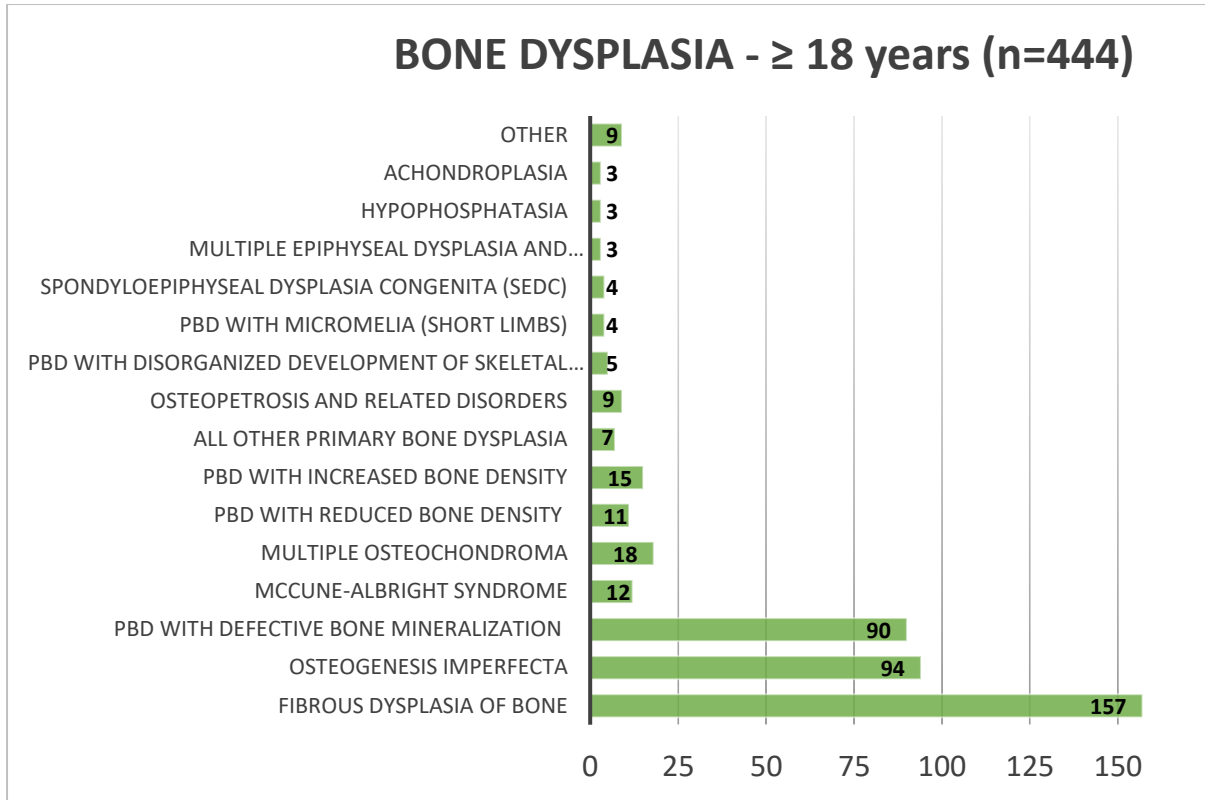


Fig. 7 - Specific diagnosis reported from April 2020 to December 2022 in  $\geq 18y$ .

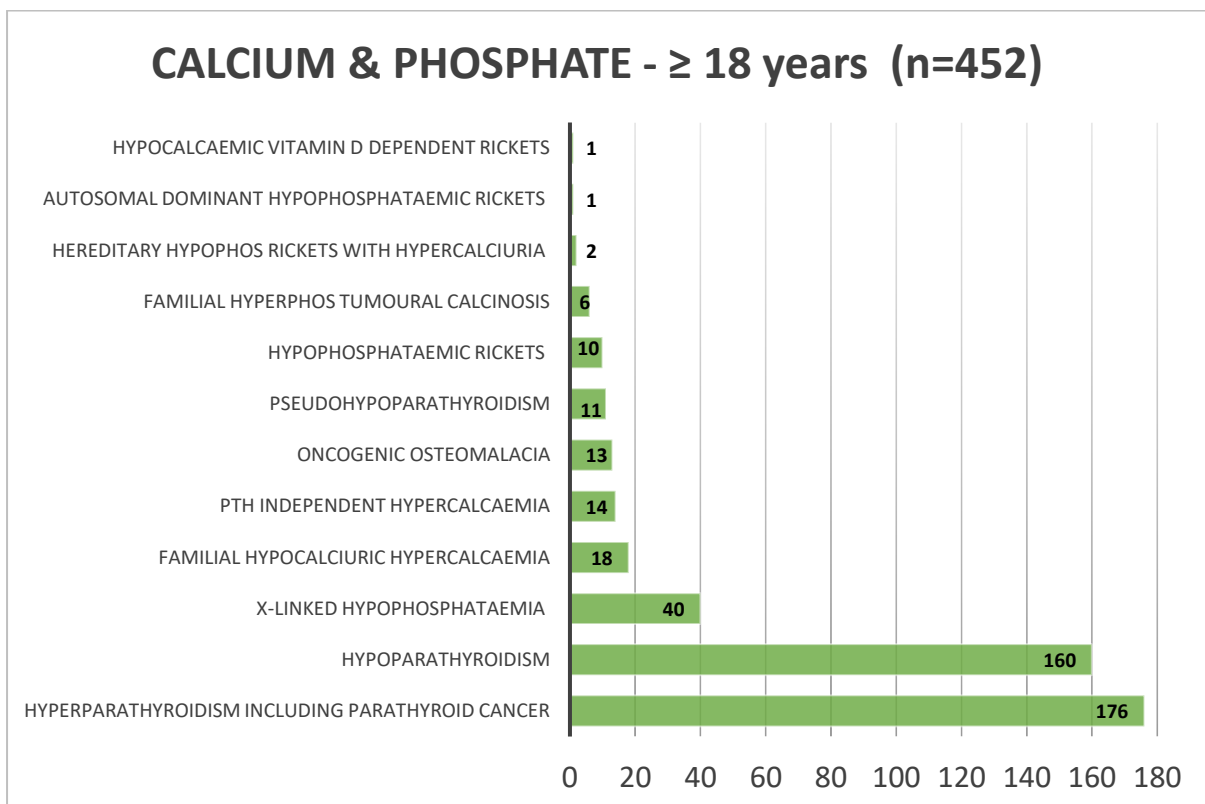


Fig.8 - Specific diagnosis reported from April 2020 to December 2022 in  $\geq 18y$ .

## **Findings**

e-REC is a simple platform that can be used to capture new encounters with patients with bone and mineral conditions.

Reporting centres have slowly increased since the EuRR-Bone project was launched in April 2020.

A total of 1537 cases were reported from April 2020 to December 2022. Paediatric cases of osteogenesis imperfecta and achondroplasia were being the most commonly reported in the bone dysplasia group. Among calcium and phosphate conditions - X-linked hypophosphataemia, pseudohypoparathyroidism and hypoparathyroidism were leading.

In the adult group, calcium and phosphate conditions are the most reported with hypoparathyroidism, hyperparathyroidism including parathyroid cancer and X-linked hypophosphataemia. In the bone dysplasia group the most reported cases were within fibrous dysplasia of bone, osteogenesis imperfecta and primary bone dysplasia with defective bone mineralization.

There is wide variability in the number of cases encountered for different conditions amongst different centres.

## **Recommendations**

The use of the platform should continue to be disseminated widely. Members of ERNs should be encouraged to utilize the platform for the yearly continuous monitoring.

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