Characteristics of patients with Osteogenesis Imperfecta: first results of the Core registry of the European Registries for Rare Endocrine and Bone Conditions

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Introduction

The European Registries for Rare Endocrine and Bone Conditions (EuRREB) were created in collaboration with the European Reference Network on Rare Endocrine Conditions (Endo-ERN) and the European Reference Network on Rare Bone Diseases (ERN BOND) to support the needs of healthcare providers, patients and researchers by providing high-quality registries. One of the conditions in the focus of EuRREB is Osteogenesis Imperfecta (OI, ORPHA 666). OI subsumes a group of rare, genetic bone disorders caused by variants in COL1A1, COL1A2 or collagen-associated genes. To date, information on treatment data is scarce. In 2022 the condition-specific module on OI was created in the Core Registry, collecting a specific dataset proposed by the group of experts in this field.

Aim

To describe the patient population and data entered in the EuRREB between October 2019 and April 2026.

Methods

Clinical contributors are invited to register new and existing cases of rare bone conditions mapped according to the Orphanet. While e-REC collects only the number of new cases seen monthly in the reporting centre, the Core Registry collects a set of clinician-reported common data elements as well as longitudinal patient and clinician-reported outcomes, generic and condition-specific.

Results

1049 individuals newly diagnosed with OI were recorded in e-REC (234 suspected and 815 confirmed cases); 446 children (43%) and 603 adults (57%) originating from 18 countries and evaluated in 45 expert centers.

Till now, more detailed information was collected in the Core Registry from 352 patients (267 adults (76%) and 85 children (24%)), 197 females (56%) from 11 countries (16 centres) (Fig. 1).

Fig. 1 – Reporting countries and centres in the Core Registry

The median age at diagnosis was 6.7 (IQR 0-27.8) years. Median time to diagnosis was 0.1 (IQR 0-9.7) years. Molecular genetic testing was performed in 65 children (76%) and 175 of the adults (66%). OI types distribution is shown in Fig.2.

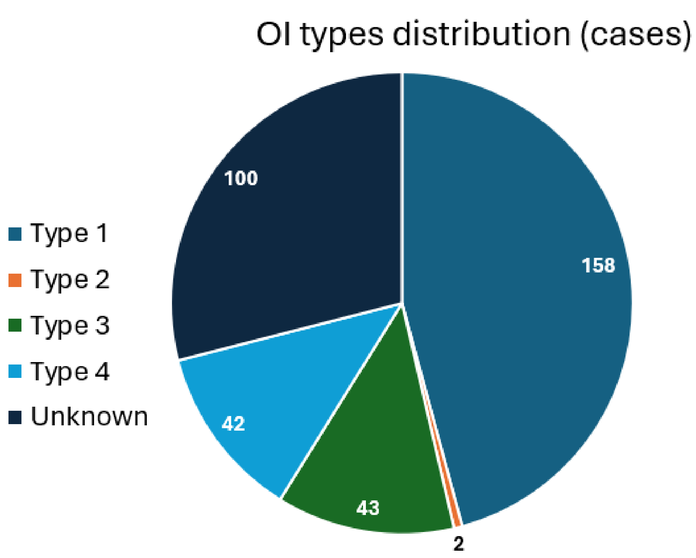

Fig. 2 – Distribution of OI cases per type

The Core OI module was completed 167 times (from 1 to 15 times per patient) for 88 (25%) patients (70 adults and 18 children).

Out of these, fractures were reported in 10/18 children(56%) and 32/70 adults(46%).

Nitrogen containing bisphosphonates were used in 10/18 children (56%) and 40/70 (57%) adults. 9/70 (13%) adults reported other medication like etidronate, teriparatide, or romosozumab at some timepoint. 6/18 (30%) children and 28/70 (40%) adults did not use any kind of bone active drugs.

DEXA was performed in 57/70 (81%) adults and in 9/18 (50%) of the children.

Surgeries were performed for fractures and deformities in 9/18 (50%) children and 21/70 (30%) adults.

Conclusion

These results contribute to a better understanding of OI via collection of real-world data and can support significant improvements in diagnosing, screening and therapeutic decision-making in the outpatient clinic. Furthermore, the EuRREB module represents a remarkable outreach effort across European countries, bringing together underrepresented regions in OI research and highlighting its wide-reaching continental scope.

How to access the OI module in EuRREB?

Survey on Treatment Initiation in Adult OI

Endo-ERN European Reference Network on Rare Endocrine Conditions
ERN BOND EUROPEAN REFERENCE NETWORK ON RARE BONE DISEASES
Funded by the European Union
Leiden University Medical Center
European Society of Endocrinology
ESPE European Society for Paediatric Endocrinology
ECTS European Calcified Tissue Society