

The European Registry for Rare Bone and Mineral Conditions (EuRR-Bone): First Year Experience of the Use of an e-Reporting Tool



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Introduction

EuRR-Bone was founded in April 2020 and is the affiliated registry of the European Reference Network for Rare Bone Diseases (**ERN BOND**). Bone and mineral conditions are seen by a variety of specialists, therefore there is a close connection with the European Registries for Rare Endocrine Conditions (**EuRECa**) and its affiliated European Reference Network for Rare Endocrine Conditions (**Endo-ERN**). EuRR-Bone is open to all centres willing to participate, regardless their affiliation to ERNs.

Methods

e-REC (e-Reporting of Rare Conditions) is an electronic reporting platform that does not collect personal identifiable information. After creating an account and customizing a reporting set-up, invitations are issued on a monthly basis and only the number of new cases encountered is requested. Data are available to all collaborators following approval by the joint Data Access Committee of EuRECa and EuRR-Bone.

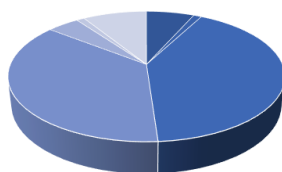
Results

35 centres are actively reporting in e-REC. of these 23 are Endo-ERN members, 12 are joint Edo-ERN and ERN BOND and 12 belong to neither.

By December 2020, 35 centres were reporting Mineral conditions and 20 centres were reporting Bone Dysplasia conditions.

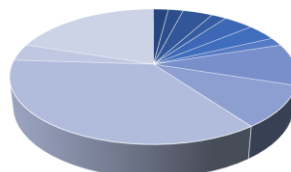
From April to December 2020 234 cases were registered.

Mineral Conditions Adults (n=90)



- Familial hypocalcaemic hypercalcaemia (6)
- Hereditary hypophosphataemic rickets with hypercalcaemia (1)
- Hyperparathyroidism including parathyroid cancer (37)
- Hypoparathyroidism (33)
- Oncogenic osteomalacia (4)
- PTH independent hypercalcaemia (1)
- X-linked hypophosphatemia (8)

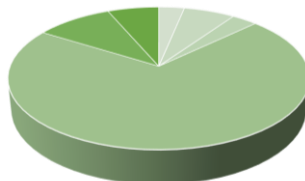
Mineral Conditions Children (n=50)



- AR hypophosphataemic rickets (1)
- Fam. hypocalcaemic hypercalcaemia (2)
- Hyperparathyroidism incl. parathyroid cancer (2)
- Hypocalcaemic vit. D resistant rickets (1)
- Hypophosphataemic rickets (5)
- Pseudohypoparathyroidism (18)
- X-linked hypophosphatemia (10)
- AD hypophosphataemic rickets (1)
- Fam. hyperphosphataemic tumoral calcinosis (1)
- Hypocalcaemic vit. D dependent rickets (2)
- Hypoparathyroidism (5)
- Oncogenic osteomalacia
- PTH independent hypercalcaemia (2)

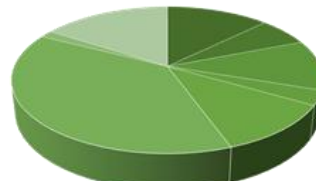
Fig. 1, 2 - Mineral conditions reported between April and December 2020.

Bone Dysplasia Conditions Adults (n=31)



- Achondroplasia (1)
- Bone dysplasia with disorganized development of skeletal components (2)
- Bone dysplasia with increased bone density (1)
- Fibrous Dysplasia of the Bone (22)
- McCune-Albright syndrome (3)
- Osteogenesis Imperfecta (2)

Bone Dysplasia Conditions Children (n=63)



- Achondroplasia (8)
- BD with disorganized development of skeletal components (4)
- Fibrous dysplasia of the bone (7)
- Hypophosphatasia (2)
- McCune-Albright syndrome (7)
- Osteogenesis imperfecta (24)
- Primary bone dysplasia with micromelia (1)
- Primary bone dysplasia with reduced bone density (10)

Fig. 3, 4 - Bone dysplasia conditions reported between April and December 2020.

Country	Centers	Country	Centers
	Austria 1		Italy 7
	Belgium 2		Latvia 1
	Croatia 1		Lithuania 1
	Czech Republic 1		Netherlands 3
	Estonia 1		Romania 1
	France 1		Spain 2
	Germany 6		Sweden 1
	Hungary 1		United Kingdom 4
	Israel 1		

Table 1 – Number of active centres per country.

Take Home Message

EuRR-Bone is a new open access registry for rare bone and mineral conditions. **e-REC** is a promising tool enabling networks to objectively map conditions and related activity, providing a better understanding of the occurrence of the rare bone and mineral conditions.

Endorsed by



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