EuRR-Bone Newsletter November 2022

Welcome to the new EuRR-Bone newsletter!

EuRR-Bone in brief

The European Registries for Rare Bone and Mineral Conditions - EuRR-Bone are funded by the European Union's Health Programme. EuRR-Bone, working closely with ERN BOND and Endo-ERN, aims to create a high-quality, patient-centred pan-European registry for rare bone and mineral conditions in collaboration with the EuRRECa project (European Registries for Rare Endocrine Conditions). EuRR-Bone, with the support of the scientific societies and patient organizations, will be of service to all patients, health care professionals and researchers involved in the care of people with rare bone and mineral conditions. EuRR-Bone is also strictly connected with patients and their caregivers, to collect data that really matter.

Welcome to EuRR-Bone's fourth newsletter! Founded in April 2020 EuRR-Bone is almost 3 years active now.

Website & Twitter

Get regular updates about EuRR-Bone activities at the project's website **https://eurr-bone.com/** The website provide an overview of the EuRR-Bone registries, including practical information and useful documents.

For more updates, follow EuRR-Bone twitter profile **@EuRRBone**.

The website has been visited by more than 4.000 different visitors worldwide more than 14.000 times!

Highlight e-REC

e-REC is an electronic reporting system which captures new patient encounters, allowing 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, specifically adapted for EuRR-Bone. The platform is open to all centres that look after people with rare bone and mineral conditions.

By June 2022, a total of 46 centres from 18 countries had reported on e-REC. The e-REC bone dysplasia and mineral list of conditions is continuously updated!

The reporting system is capturing data on variants of unknown significance (VUS) detected in bone dysplasia and Denosumab related rebound effects.

EuRR-Bone Website visit Countryview

AGE CATEGORY	MINERAL CONDITIONS	BONE DYSPLASIA	OVERALL
Paediatric patients	186	340	526
Adults	410	370	780
Overall (pediatric&adults)	596	710	1.306

Transfer of server

EuRR-Bone's data server was transferred to the Leiden University Medical Centre in Leiden, the Netherlands. The new ethics approval documentation is available at the website.

The Core Registry

The Core Registry allows patient and health care professionals participation. It collects a core data set for a wide range of rare bone and mineral conditions. In addition, condition-specific modules are available for achondroplasia, osteogenesis imperfecta, FD/MAS and more recently, rare hypophosphataemic disorders. A module collecting data on parathyroid carcinoma will be launched soon. These modules collect patient and clinician reported outcomes.

New developments

- **New navigation buttons – to make it more user friendly and easier to guide you through the registry**
- **Outstanding tasks (Edit To Do button) – a tool to allow a reporter to make notes**
- **The Brief Pain Inventory – Short Form (BPI-SF) is now available on the patient and the clinician platform**

Registry Drop-in sessions

A series of sessions, scheduled on every second Friday of the month at 2pm (CET) and the fourth Wednesday in every month at 4pm CET via ZOOM, have been arranged to demonstrate the registries (e-REC and Core Registry). You don't need to register, just drop in. Click on the following link to join: https://us02web.zoom.us/j/86149844975
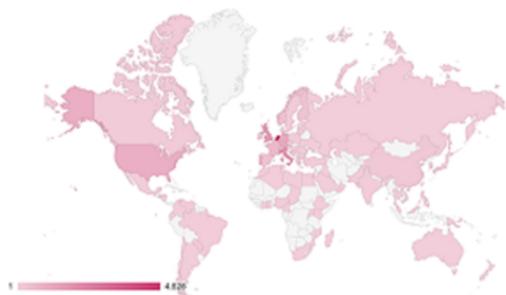
In addition, some demonstration videos are available @ https://eurreca.net/e-rec-demonstration-videos/

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EuRR-Bone Meetings

EuRR-Bone PGB meeting - **The Project Governing Board (PGB) meeting took place online on Thursday 13 October 2022**

EuRR-Bone and EuRRECa steering committee meeting **– October 28th, 2022**

Conferences & Meetings

EuRR-Bone has been present at various conferences and meetings:

▶ **The 14th <u>International Conference on Osteogenesis Imperfecta</u> was held between 30 August and 2 September 2022 in Sheffield, UK. The registries exhibition booth received many visits of experts interested in the project. Find the abstracts <u>here</u>**

▶ **The European Society for Paediatric Endocrinology held its 60th annual meeting on 15-17 September 2022 in Rome. EuRR-Bone and EuRRECa were present with a joint exhibition booth. There, many came to visit the booth and discovered the use and objectives of the registries. Research performed with the data collected in the registries was presented as well by Ana Priego**

▶ **ERICA – ERN Data Management Strategy Multistakeholder Workshop in Heidelberg. EuRR-Bone and EuRRECa were represented in this event as well as ePAG representatives.**
This workshop included discussions aimed at helping ERNs in setting up, maintaining and improving their registries. Face to face interaction facilitated learning about and from other registries. Learn more <u>here</u>. To get access to the workshop presentations please contact the <u>ERICA office</u>

Upcoming Events

🌐 **The <u>Dutch Society for Calcium and Bone Metabolism</u> will hold its annual meeting on 10-11 November 2022 in Zeist, The Netherlands. Research performed with the registries data will be presented by Ana Priego**

🌐 **OIFE virtual Investigator Meeting will take place on 18 November 2022 and the registries will present current activity and data regarding Osteogenesis Imperfecta**

🌐 **EuRR-Bone Final Meeting will be the 13th of February**

🌐 **ERN BOND General Assembly – will take place in Bologna, on 4 and 5 May 2023**

🌐 **1st Joint EuRRECa and EuRR-Bone meeting – 3 April 2023. Click <u>here</u> to register**

🌐 **Prof. Faisal Ahmed inaugural lecture - 3 April 2023**

Bone2Gene: Artificial Intelligence for the diagnosis of genetic skeletal diseases

There are currently more than 400 known different rare bone diseases with very heterogeneous phenotypic features that make their accurate diagnosis a very challenging task. The Bone2Gene project aims at training an artificial intelligence that would be able to detect and recognize various and subtle dysmorphisms and patterns caused by different bone diseases. To accomplish this, a large number of training data is required. To learn more about this project and ways to contribute contact Dr. B. Javanmardi (e-mail: bjav@uni-bonn.de) or watch this presentation https://www.youtube.com/watch?v=Mypx_cCoQkU