



## MELORHEOSTOSIS MODULE

This disease specific module is a joint project of the Rare Bone Disease action group of the European Calcified Tissue Society (ECTS), the European Reference Network for Rare Bone Disorders (ERN BOND) and the European Registries for Rare Bone and Mineral Conditions (EuRR-Bone).

### ECTS-ERN BOND Melorheostosis study group:

- **Prof Martine Cohen-Solal** - Co-chair ECTS RBDAG- rheumatologist - [Inserm U1132 Bioscar](#) and universit  Paris-Cit  [Centre Viggo Petersen H pital Lariboisi re](#), Paris, France
- **dr Luca Sangiorgi** - ERN BOND coordinator, Director of the Department: Rare Skeletal Diseases, [Rizzoli Orthopaedic Institute](#), Italy
- **Prof Bo Abrahamsen** – ECTS board member -endocrinologist – University of Southern Denmark and [Holb k Hospital](#), Denmark
- **dr Natasha Appelman-Dijkstra** – endocrinologist, EuRR-Bone coordinator and WP5 lead ERN BOND, [Leiden University Medical Center](#), The Netherlands
- **dr Mariya Cherenko** – EuRR-Bone/EuRRECa research fellow, [Leiden University Medical Center](#), The Netherlands
- **dr Gavin Clunie** – rheumatologist - [Nuffield Health Ipswich Hospital](#) - Cambridge, United Kingdom
- **dr Corinna Grasemann** - endocrinologist - Consultant Pediatric Endocrinologist at [Children’s Hospital Bochum](#), Bochum Germany
- **Prof W.F. Lems** – ECTS board member- rheumatologist – [Amsterdam UMC](#), The Netherlands
- **dr Ana Luisa Priego Zurita** – EuRR-Bone/EuRRECa research fellow, [Leiden University Medical Center](#), The Netherlands
- **Prof Heide Siggelkow** - ECTS board member -endocrinologist – [MVZ ENDOKRINOLOGIKUM](#) G ttingen, Germany
- **Prof Carola Zillikens** – endocrinologist - [Erasmus Medical Center](#) Rotterdam, The Netherlands

### About the Registries:

EuRR-Bone started in April 2020 and works closely with the European Registries for Rare Endocrine Conditions project ([EURRECa](#)) and the European Reference Networks for Rare Bone (ERN BOND) and Rare Endocrine conditions (ENDO-ERN) and includes 2 registries platform:

- An e-reporting program ([e-REC](#)) that captures new clinical encounters
- A centralized [Core](#) registry containing [Disease-specific modules](#) ( already available for the bone and mineral conditions Achondroplasia, Osteogenesis imperfecta, Fibrous Dysplasia/McCune Albright syndrome, Rare Hypophosphatemia, Inactivating PTH/PTHrP signalling disorder/IPPSD (pseudohypoparathyroidism) and Parathyroid carcinoma

The registries were founded to support the needs of ERN BOND but are open to all experts. In other words, non-ERN centers can participate at an equal level.

Experts can participate in 2 levels:

1. Using the e-REC platform which is an easy to use monthly counting system and simply registers new cases of rare conditions. This does not require informed consent from the patient, access to e-REC can be requested [here](#)
2. Using the Core registry and its disease specific modules. This option does require informed consent. The [EuRR-Bone website](#) shows examples from approved protocols and informed consent forms, if needed a data management plan or other IRB documents are available upon request. [Data access policy](#), [Data sharing agreements](#) can be found here as well.

For both platforms you will need to set up an account, the Core Registry requires a 2 factor identification using a OTP like google authenticator. To start using the Core you need to have an access to the Core registry. You can request it [here](#).

In the following pages we will provide a quick instruction on how to get access to the Core registry and start the module.

**Short introduction on the Melorheostosis module** (see also the inserted **step-by-step tutorial** below)

This module was prepared by the ECTS in collaboration with ERN BOND and EuRR-Bone, According to the ORPHAnet coding Melorheostosis falls under primary bone dysplasia with increased bone density (ORPHA93444) and to avoid large dropdown lists, the first registration within the Core registry is under this orphacode, after this the more detailed code can be chosen :Melorheostosis (ORPHA2485), Melorheostosis with osteopoikilosis (ORPHA1879), Buschke-Ollendorff syndrome (ORPHA1306) or Isolated osteopoikilosis (ORPHA166119). After entering the detailed diagnosis the Melorheostosis module appears automatically and is ready to use.

The modules should be updated every 12 months when patients are under active control. If not updates should be provided every 3 years. Of course additional data can be entered on demand. The Module requests a small amount of clinical data, information on pain (using BPI) and QoL (using MSK-HQ) these are available in different languages for the patient and for the clinician if needed as well. Next to the disease specific outcomes we collect **EQ5D and mobility** information on all patients with bone and mineral conditions, so please fill these out as well (or ask the patient to fill them out if they would like to participate).

PROMS can be requested automatically to the patient by the system and the clinician receives information when filled in.

#### **Tools used in the module :**

[MSK-HQ](#) - is a short questionnaire that allows people with musculoskeletal conditions to report their symptoms and quality of life in a standardized way

#### **Brief Pain Inventory**

#### **EQ5D**

We hope that you will use the module and of course contributors will be included in any future output according to our [publication policy](#).

Please scroll down for the detailed **step-by-step tutorial**

If you have questions please contact us on: [EuRR\\_Bone@lumc.nl](mailto:EuRR_Bone@lumc.nl) or join our regularly [drop-in session](#) via zoom.

On behalf of the Study Group

Dr. Mariya Cherenko,

Dr. Natasha Appelman-Dijkstra

# How to use the disease specific module in Core registry

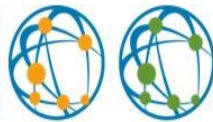


**EuRRECa**  
European Registries for  
Rare Endocrine Conditions



**EuRR-Bone**  
European Registries for Rare  
Bone and Mineral Conditions

## Login in Core registry



Go to <https://eurr-bone.com/registries/core/>

HOME STRUCTURE ▾ REGISTRY ▾ PATIENTS NEWS ▾

### CORE

The Core Registry collects a core data set for a wide range of rare bone and mineral conditions. The data collected will be used to improve clinical care as well as research and oversight to data access is provided by the Data Access Committee. The Core Registry will also advise participants on other suitable studies and registries. As the Core Registry has been functional within EuRRECa since 2019 it has been approved by the National Ethics Service and Information Governance authorities in the UK. This is the reason EuRR-Bone could embark the Core registry quickly and the registry is open for rare bone and mineral conditions.

[Request Access to the Core Registry](#)

[Login to the Core Registry](#)

[Graphic Walkthrough of the Core Registry](#)

If you have no  
account yet, you  
can request the  
access [here](#)

**EuRRECa** **EuRR-Bone** e-Reporting of Rare Conditions (e-REC) and Core Registry

### New User Registration Request

Fields marked with \* are mandatory

\* Application Role Requested ☐ e-REC Reporter ☐ Core Registry Clinical Contributor

Title

\* Surname

\* Given Name

Middle Names

\* Email (and User Name)

\* Password

\* Confirm Password

Telephone

I am the Centre Lead for this Centre ☐

Filter Centres by Country (optional)

\* Centre

# Login in Core registry



Go to <https://eur-bone.com/registries/core/>  
and click on "Login to the Core Registry"  
if you already have an account

HOME STRUCTURE REGISTRY PATIENTS NEWS

## CORE

The Core Registry collects a core data set for a wide range of rare bone and mineral conditions. The data collected will be used to improve clinical care as well as research and oversight to data access is provided by the Data Access Committee. The Core Registry will also advise participants on other suitable studies and registries. As the Core Registry has been functional within EuRRECa since 2019 it has been approved by the National Ethics Service and Information Governance authorities in the UK. This is the reason EuRR-Bone could embark the Core registry quickly and the registry is open for rare bone and mineral conditions.

[Request Access to the Core Registry](#)

[Login to the Core Registry](#)

[Graphic Walkthrough of the Core Registry](#)

## Login

User Name

a.l.priego\_zurita@umc.nl

Password

\*\*\*\*\*

6 digit PIN from your mobile device

[Click on the following link for further information about OTP authentication](#)

Log in

You will need to download OTP  
authenticator

# Add a new patient



Core Registry

Dashboard

Patients

Centres

Centre Users

## Clinician Dashboard

Patients in Centre: EuRR-Bone-Test

Patients registered by you: 2

Patients registered in your centre: 27

Patients with outstanding actions: 0

Search  
List  
Add  
Deleted Patients

Click here

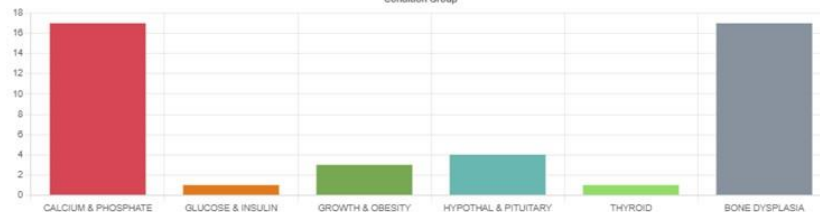
Downloads

Group

Child

Adult

## Condition Group



# Fill in the fields



**Add Patient**

Fields marked with \* are mandatory

**Mandatory fields** →

- \* Consent for Core Registry
- \* Date of Birth
- \* Condition Group (select all that apply)

**If applicable** →

- EUPID (European Patient ID)
- Sex at Birth
- Current Gender
- Country of Birth
- Country of Usual Residence
- Patient Follow-up Status
- Date of Death
- Primary Cause of Death

**SAVE** →

**Choose "Bone dysplasia"** →

Opt-in ☒ Opt-out ☐

ADRENAL ☐ CALCIUM & PHOSPHATE ☐ GLUCOSE & INSULIN ☐  
☒ BONE DYSPLASIA ☐

Male ☐ Female ☐ Other ☒ Unknown

Male ☐ Female ☐ Non-binary ☐ Other ☐ Asked but unknown

NETHERLANDS

NETHERLANDS

Patient active ☐ Patient inactive ☐ Patient care suspended ☐

mm/dd/yyyy

- Select -

Yes ☐ No ☐ Not Known ☐

Yes ☐ No ☐ Not Known ☐

Yes ☐ No ☐ Not Known ☐

Yes ☐ No ☐ Not Known ☐

Yes ☐ No ☐ Not Known ☐

Save Cancel

# Go to condition group section



**EuRECa** **EuRR-Bone** **Core Registry** **Dashboard** **Patients** **Centres** **Centre Users** **Mariya Cherenko - EuRR-Bone-Test**

**Edit Patient**

Patient ID - 4275 [Edit To Do](#)

Fields marked with \* are mandatory

\* Consent for Core Registry

\* Date of Birth

\* Condition Group (select all that apply)

EUPID (European Patient ID)

Sex at Birth

Current Gender

Country of Birth

Country of Usual Residence

Patient Follow-up Status

Date of Death

Primary Cause of Death

Can be contacted for research purposes by clinician responsible for patient

Data can be shared for research purposes

Can be contacted for collecting Patient Reported Outcomes

Opt-in ☒ Opt-out ☐

ADRENAL ☐ CALCIUM & PHOSPHATE ☐ GLUCOSE & INSULIN ☐  
GENETIC ENDOCRINE TUMOURS ☐ GROWTH & OBESITY ☐ HYPOTHAL & PITUITARY ☐  
SEX DEVELOPMENT ☐ THYROID ☐ ☒ BONE DYSPLASIA ☐

test Cher

Male ☐ Female ☐ Other ☒ Unknown

Male ☐ Female ☐ Non-binary ☐ Other ☐ Asked but unknown

NETHERLANDS

NETHERLANDS

Patient active ☐ Patient inactive ☐ Patient care suspended ☐ Patient care terminated ☐ Deceased ☒ Unknown

mm/dd/yyyy

- Select -

Yes ☐ No ☐ Not Known ☐

Yes ☐ No ☐ Not Known ☐

Yes ☐ No ☐ Not Known ☐

**BONE DYSPLASIA** **Genetic Outcomes** **Audio** **Patient List**

Click here to fill in the bone dysplasia tab



# Fill in primary condition and specific diagnosis



**EuRRECa** European Registry for Rare Endocrine Conditions | **EuRR-Bone** European Registry for Rare Bone and Mineral Conditions | **Core Registry** | Dashboard | Patients | Centres | Centre Users | Mariya Cherenko - EuRR-Bone-Test

**Edit Patient Condition** | Core Data | **CALCIUM & PHOSPHATE** | Patient Access | Generic Outcomes | **Diagnosis-specific Outcomes** | Audit | Patient List

Patient ID - 551 | Date of Birth - [ ] | **Edit To Do**

**Bone Dysplasia**

Senior Clinician < 18 - N/A  
Senior Clinician ≥ 18 - N/A

If applicable → e-REC ID [ ]

First Contact with Centre [mm/dd/yyyy] [ ]

Clinician Responsible for Patient [Dr. Ana Priego] [v]

Date of Condition Onset [mm/dd/yyyy] [ ]

Primary Condition [Primary bone dysplasia with increased bone density] [v] ← **Select the Primary condition - "PBD with increased bone density", for Specific diagnosis – choose from the list**

Specific Diagnosis [Melorheostosis with osteopoikilosis] [v]

The full hierarchy of Conditions and Specific Diagnoses can be found in the Core Registry Conditions Dictionary

How Was Diagnosis Reached  
☐ Clinical ☐ Biochemistry ☐ Genetic ☐ Histology ☐ CT ☐ MRI ☐ PET ☐ USS ☐ X-Ray  
☐ Other ☐ Not Known

Date of Diagnosis [mm/dd/yyyy] [ ]

Participation in Detailed Disease Registry  
☐ Yes ☐ No ☐ Not Known

Detailed Disease Registry [ ]

Detailed Disease Registry Patient ID [ ]

**Save** **Cancel**

# Go to diagnosis-specific outcome



**EuRRECa** European Registry for Rare Endocrine Conditions | **EuRR-Bone** European Registry for Rare Bone and Mineral Conditions | **Core Registry** | Dashboard | Patients | Centres | Users | Centre Users | Patient Users | Reference

**Edit Patient Condition** | Core Data | Patient Access | Generic Outcomes | **Diagnosis-specific Outcomes** | Audit | Patient List

Patient ID - 4270 | Date of Birth - [ ] | **Edit To Do**

**Bone Dysplasia**

Senior Clinician < 18 - N/A  
Senior Clinician ≥ 18 - N/A

e-REC ID [ ]

First Contact with Centre [04/12/2022] [ ]

Clinician Responsible for Patient [Natasha Appelman-Dijkstra] [v]

Date of Condition Onset [01/01/2020] [ ]

Primary Condition [Primary bone dysplasia with increased bone density] [v]

Specific Diagnosis [Melorheostosis] [v]

The full hierarchy of Conditions and Specific Diagnoses can be found in the Core Registry Conditions Dictionary

**Click here to fill in the disease-specific module**

# Fill in the new outcome



Core Registry

Dashboard Patients Centres Users Centre Users Patient Users Reference Data

## Diagnosis-specific Outcomes

Patient ID - 4270 Date of Birth -

Core Data

BONE DYSPLASIA

Patient Access

Generic Outcomes

Audit

Patient List

Bone Dysplasia

Supplemental material to complete this module

No Outcomes found

New Outcome

Click here to fill in the new outcome for disease-specific module

# Fill in the data into the module and save



## Add Diagnosis-specific Outcome

Patient ID - Date of Birth - Condition Group - Bone Dysplasia

Questionnaire: Melorheostosis, Buschke-Ollendorff syndrome, Isolated osteopoikilosis

New Outcome

Date of Birth		e-REC ID		Primary Condition	
Current Gender	Male	Clinician Responsible for Patient	Natasha Appelman-Dijkstra	Primary bone dysplasia with increased bone density	
				Specific Diagnosis	Melorheostosis
				Date of Diagnosis	2022-04-12

General Clinical features Genetic analysis Fractures Surgery Medication MSK-HQ BPI

Family ID ?

Proband ☐ Not Known ☐ Yes ☐ No

Family history ☐ Negative ☐ Positive - Maternal ☐ Positive - Paternal ☐ Positive - Unknown ☐ Unknown

Save Back



# Fill in the generic outcomes



**EuRRECa** European Registries for Rare Endocrine Conditions **EuRR-Bone** European Registries for Rare Bone and Mineral Conditions **Core Registry** Dashboard Patients Centres Users Centre Users Patient Users Reference Data

Diagnosis-specific Outcomes **Core Data** **BONE DYSPLASIA** **Patient Access** Generic Outcomes Audit Patient List

Patient ID - 4270 Date of Birth - [REDACTED]

Bone Dysplasia

Supplemental material to complete this module

No Outcomes found

[New Outcome](#)

Give an access to your patient (in case of consent)

# Fill in the generic outcomes



**EuRRECa** European Registries for Rare Endocrine Conditions **EuRR-Bone** European Registries for Rare Bone and Mineral Conditions **Core Registry** Dashboard Patients Centres Users Centre Users Patient Users Reference Data Mariya Cherenko - Leiden University

Generic Outcomes **Core Data** **BONE DYSPLASIA** **Patient Access** **Diagnosis-specific Outcomes** Audit Patient List

Patient ID - 450 Date of Birth - [REDACTED]

EQ-5D BPI-SF PROMIS Profile WHO ICF Patient Reported Outcome Request Settings

Show 10 entries Search:

Created Date	Completed By	Outcome	Mobility	Self Care	Activity	Pain	Anxiety	EQ VAS	Actions
2022-11-02	Dr. Ana Priego	EQ-5D-5L	2	2	2	2	1	71	
2021-03-24	Dr. Ana Priego	EQ-5D-5L		1	1	2	2	74	
2021-03-24	Dr. Ana Priego	EQ-5D-5L		2	2	1	1	80	

Current Patient EQ-5D Request Settings: EQ-5D-5L every 3 months, starting on 2022-06-30

[Patient EQ-5D Request Settings](#) [Create EQ-5D](#)

Click here to fill in the new generic outcome for your patient: mobility, EQ-5D or other

# Choose the frequency for PROMs



Core Registry

Dashboard Patients Centres Users Centre Users Patient Users Reference Data Mariya Cherenko - Le

Generic Outcomes

Core Data BONE DYSPLASIA Patient Access Diagnosis-specific Outcomes Audit

Patient ID - 450 Date of Birth -

EQ-5D BPI-SF PROMIS Profile WHO ICF Patient Reported Outcome Request Settings

Create or edit requests for the Patient to complete generic questionnaires

Show 10 entries

Generic Outcome Questionnaire	Request Frequency (months)	Request Start Date	Actions
WHO-ICF Mobility	9	2022-11-01	<a href="#">Edit</a> <a href="#">Delete</a>

[Add Outcome Request](#)

Previous 1

Click here to choose the frequency of email requests to the Patient to self-complete a Patient Generic Outcome Questionnaire

# Choose the frequency for PROMs



Core Registry Dashboard Patients Centres

Generic Patient Reported Outcome Request Settings

These settings control the frequency of email requests to the Patient to self-complete a Patient Generic Outcome Questionnaire

Outcome Questionnaire: WHO-ICF Mobility

Request Frequency: 6 months

Date of Next Request: mm/dd/yyyy

[Save](#) [Cancel](#)

Click here to choose the frequency of email requests to the Patient to self-complete a Patient Generic Outcome Questionnaire

## Contact us!



Please don't hesitate to contact us by e-mail  
[EuRR\\_bone@lumc.nl](mailto:EuRR_bone@lumc.nl) in case of any questions.

Or join our regularly drop-in sessions <https://eur-bone.com/>

Next sessions:  
13.01.2023 at 14.00  
10.02  
10.03  
25.01.2023 at 16.00  
22.02  
22.03

