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:

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- **Prof Bo Abrahamsen** – ECTS board member -endocrinologist – University of Southern Denmark and Holbæk Hospital, Denmark
- **dr Natasha Appelman-Dijkstra** – endocrinologist, EuRR-Bone coordinator and WP5 lead ERN BOND, Leiden University Medical Center, The Netherlands
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- **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 Hypophasphatemia, 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 used 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 or join our regularly drop-in session via zoom.

On behalf of the Study Group

Dr. Mariya Cherenko,

Dr. Natasha Appelman-Dijkstra

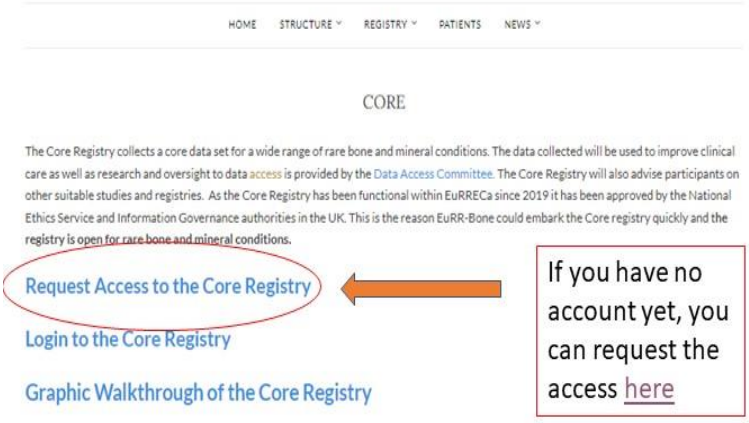
How to use the disease specific module in Core registry

EuRRECa
European Registries for Rare Endocrine Conditions

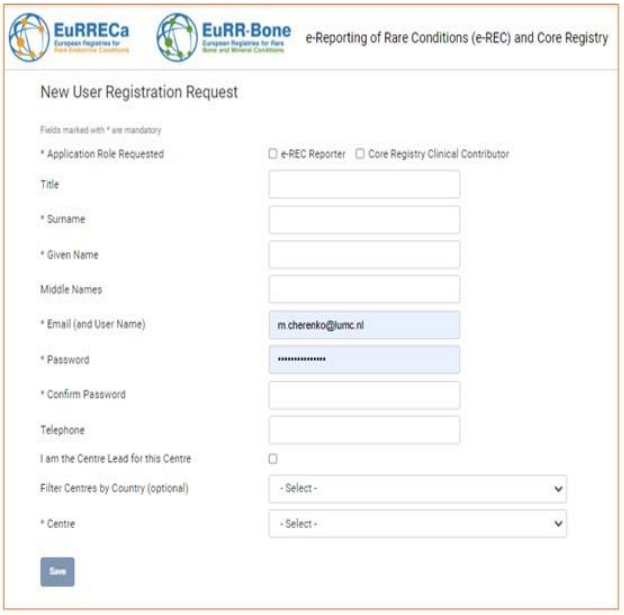
EuRR-Bone
European Registries for Rare Bone and Mineral Conditions

Login in Core registry

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

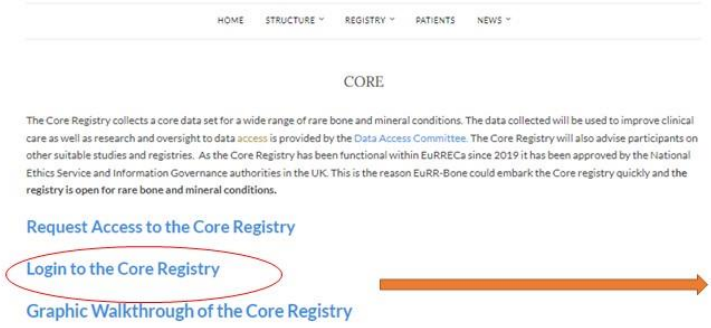


If you have no account yet, you can request the access here



Login in Core registry

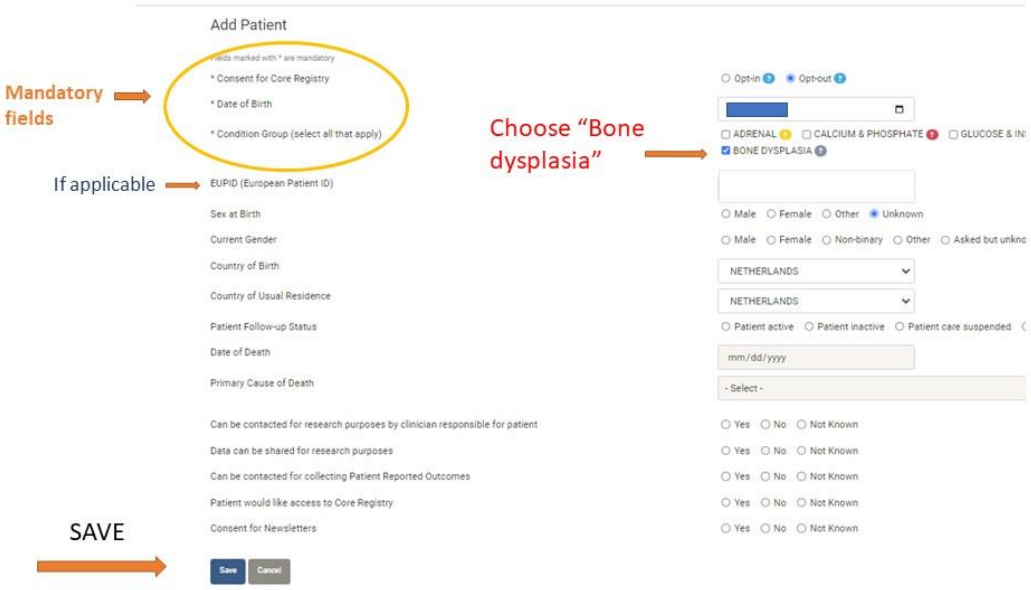
Go to https://eurr-bone.com/registries/core/
and click on "Login to the Core Registry''
if you already have an account



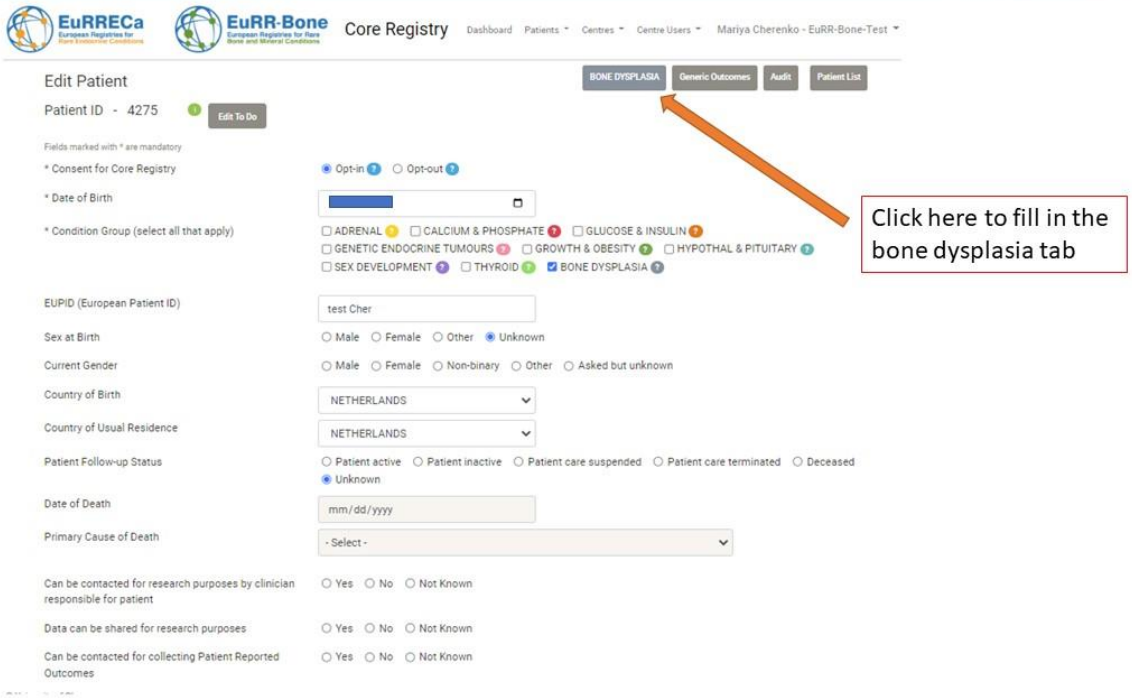
You will need to download OTP authenticator

Add a new patient

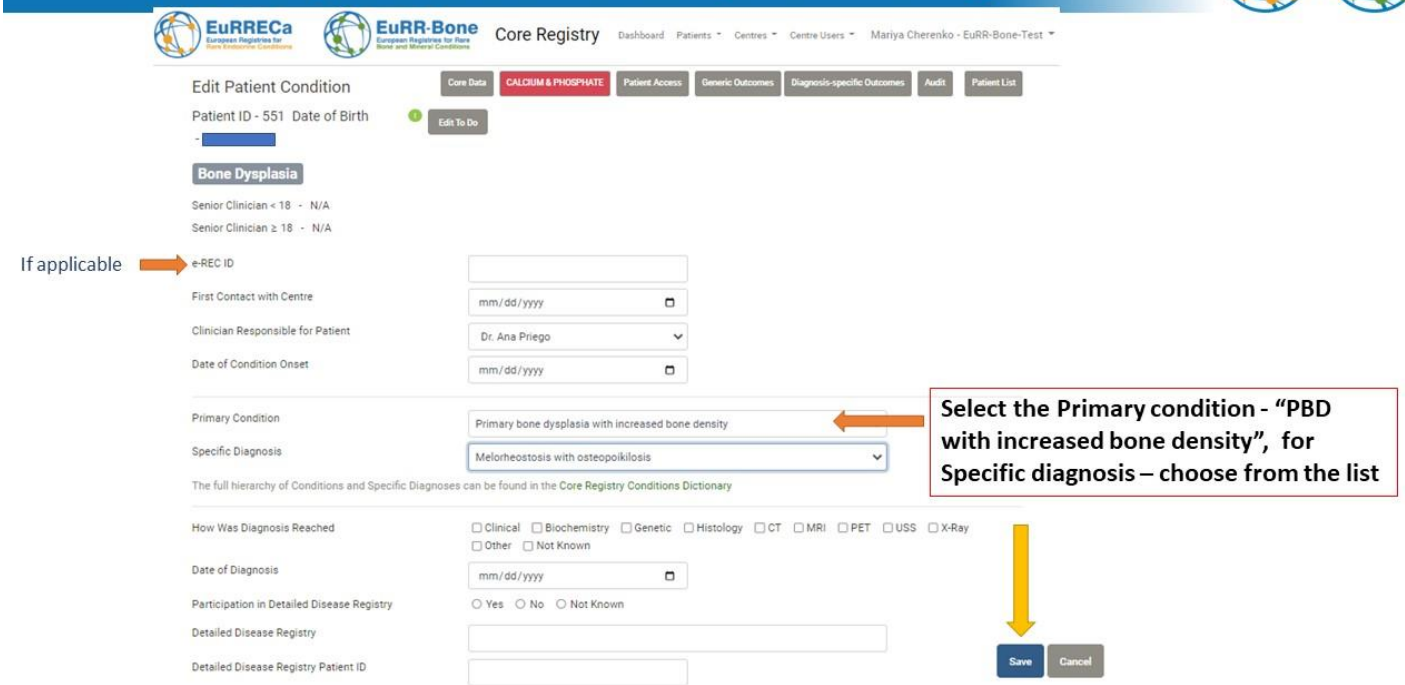
Fill in the fields



Go to condition group section

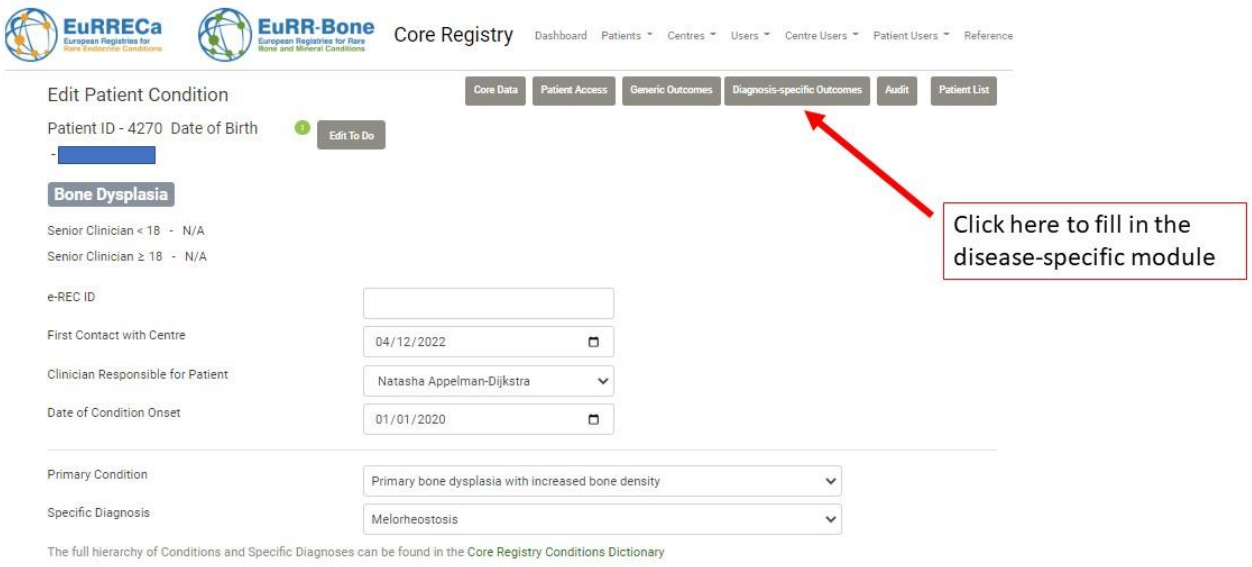


Fill in primary condition and specific diagnosis



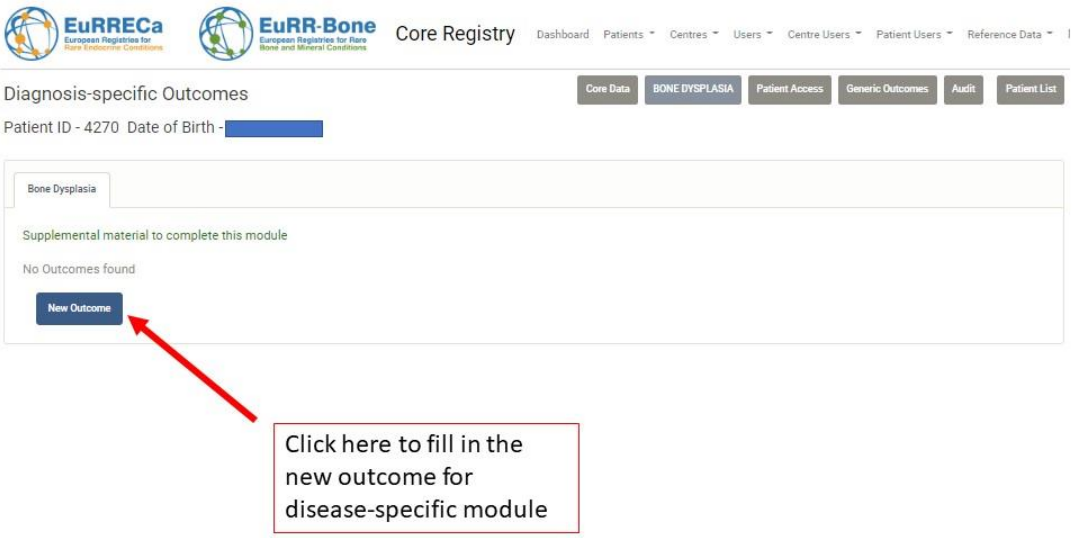
Select the Primary condition - "PBD with increased bone density", for Specific diagnosis – choose from the list

Go to diagnosis-specific outcome

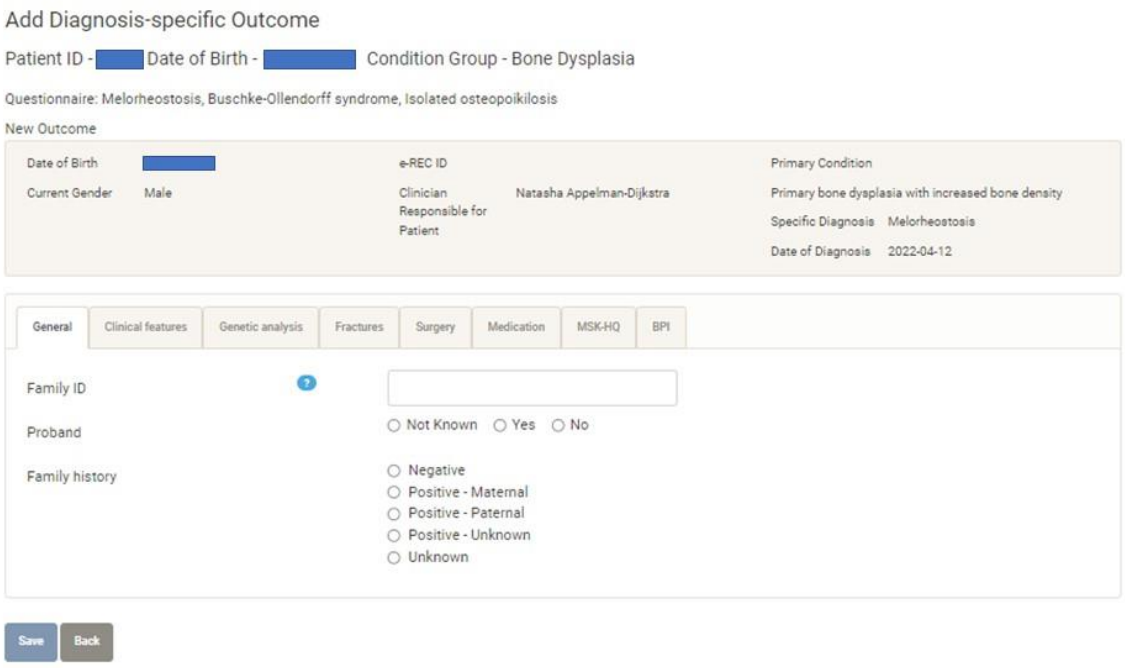


Click here to fill in the disease-specific module

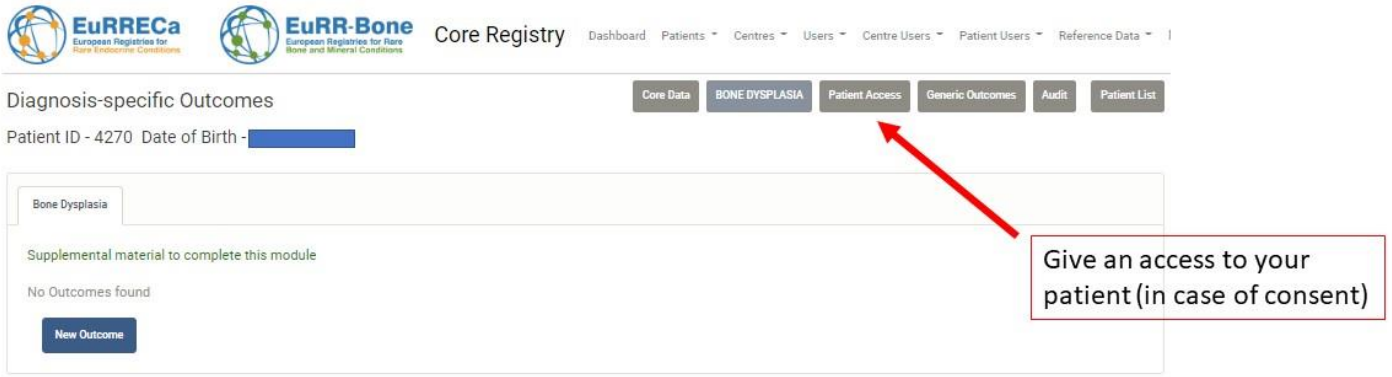
Fill in the new outcome



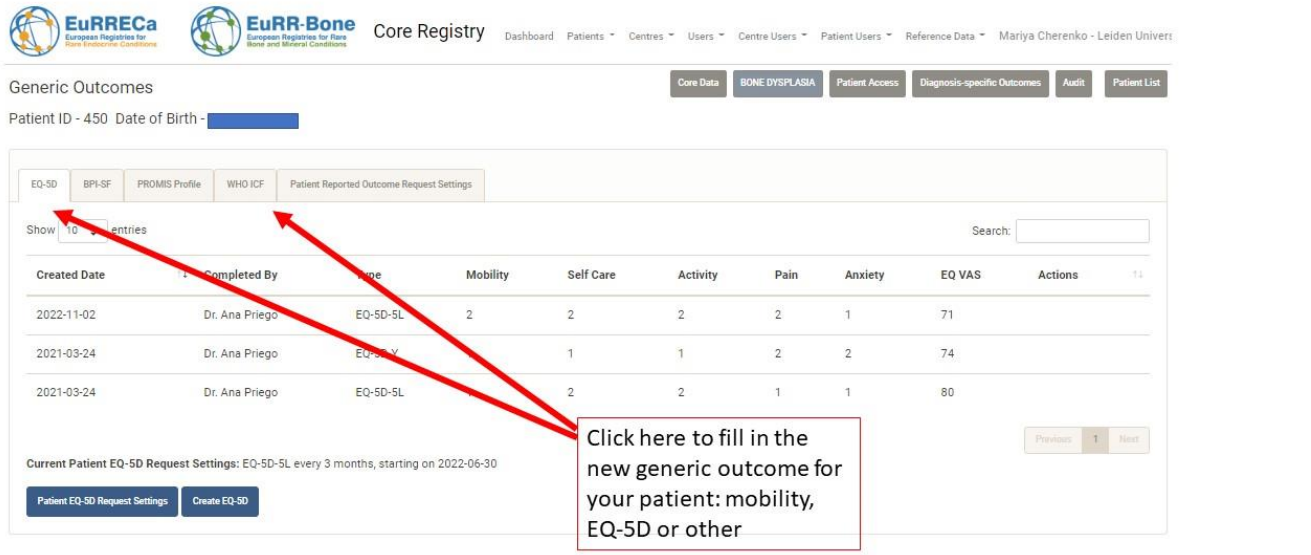
Fill in the data into the module and save



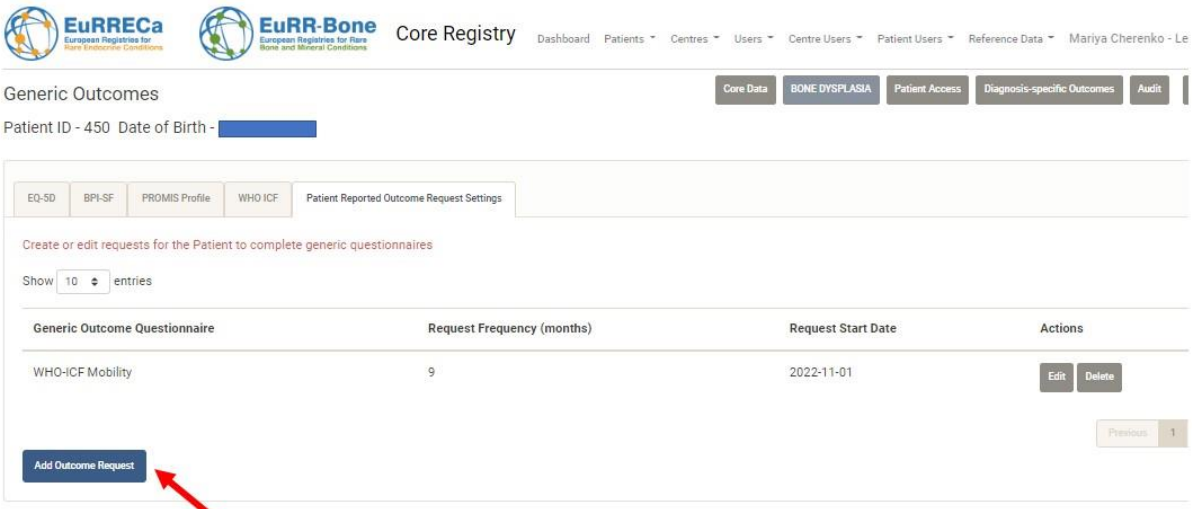
Fill in the generic outcomes



Fill in the generic outcomes

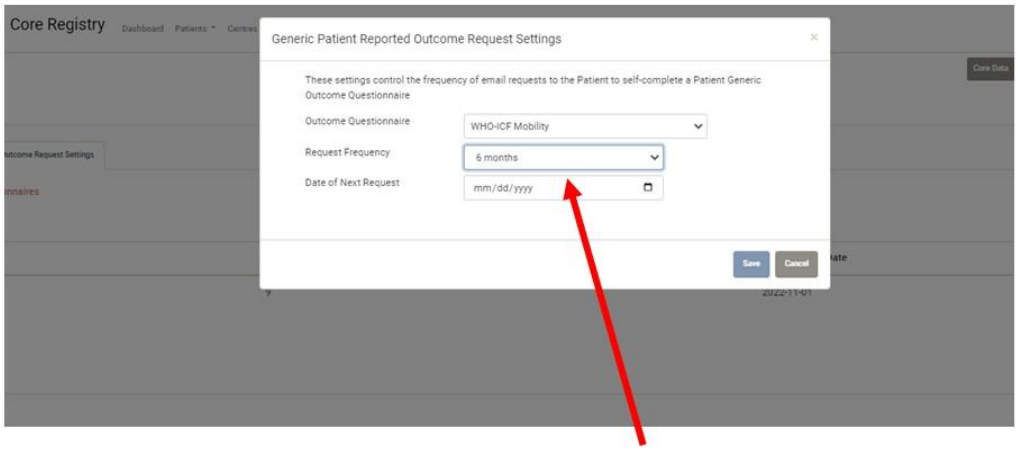


Choose the frequency for PROMs



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



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 in case of any questions.

Or join our regularly drop-in sessions https://eurr-bone.com/

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