Using the Pseudohypoparathyroidism Conditions module in the Core registry

EuRRECa
European Registries for
Rare Endocrine Conditions

EuRR-Bone
European Registries for Rare
Bone and Mineral Conditions

Pseudohypoparathyroidism conditions module

This condition-specific module is open for the following diagnoses

- Pseudohypoparathyroidism (iPPSD) **ORPHA97593**
- Pseudopseudohypoparathyroidism (iPPDS2) **ORPHA79445**
- Pseudohypoparathyroidism type 1A (IPPSD2) **ORPHA79443**
- Pseudohypoparathyroidism type 1B (IPPSD3) **ORPHA94089**
- Pseudohypoparathyroidism type 1C (IPPSD2) **ORPHA79444**
- Pseudohypoparathyroidism type 2 **ORPHA94090**
- Pseudohypoparathyroidism with Albright hereditary osteodystrophy **ORPHA457059**
- Pseudohypoparathyroidism without Albright hereditary osteodystrophy **ORPHA457062**
- Acrodystostosis Type 1 (iPPDS4) **ORPHA950**
- Acrodystostosis Type 2 (iPPDS5) **ORPHA950**
- Blomstrand chondrodysplasia (iPPSD1) **ORPHA50945**
- Eiken disease (iPPSD1) **ORPHA79106**
- Hypertension and brachydactyly Syndrome (iPPSD6) **ORPHA1276**
- Pseudohypoparathyroidism, all known genes excluded (iPPSDx) **ORPHA97593**

In this guide, we will show you how to use the module step by step

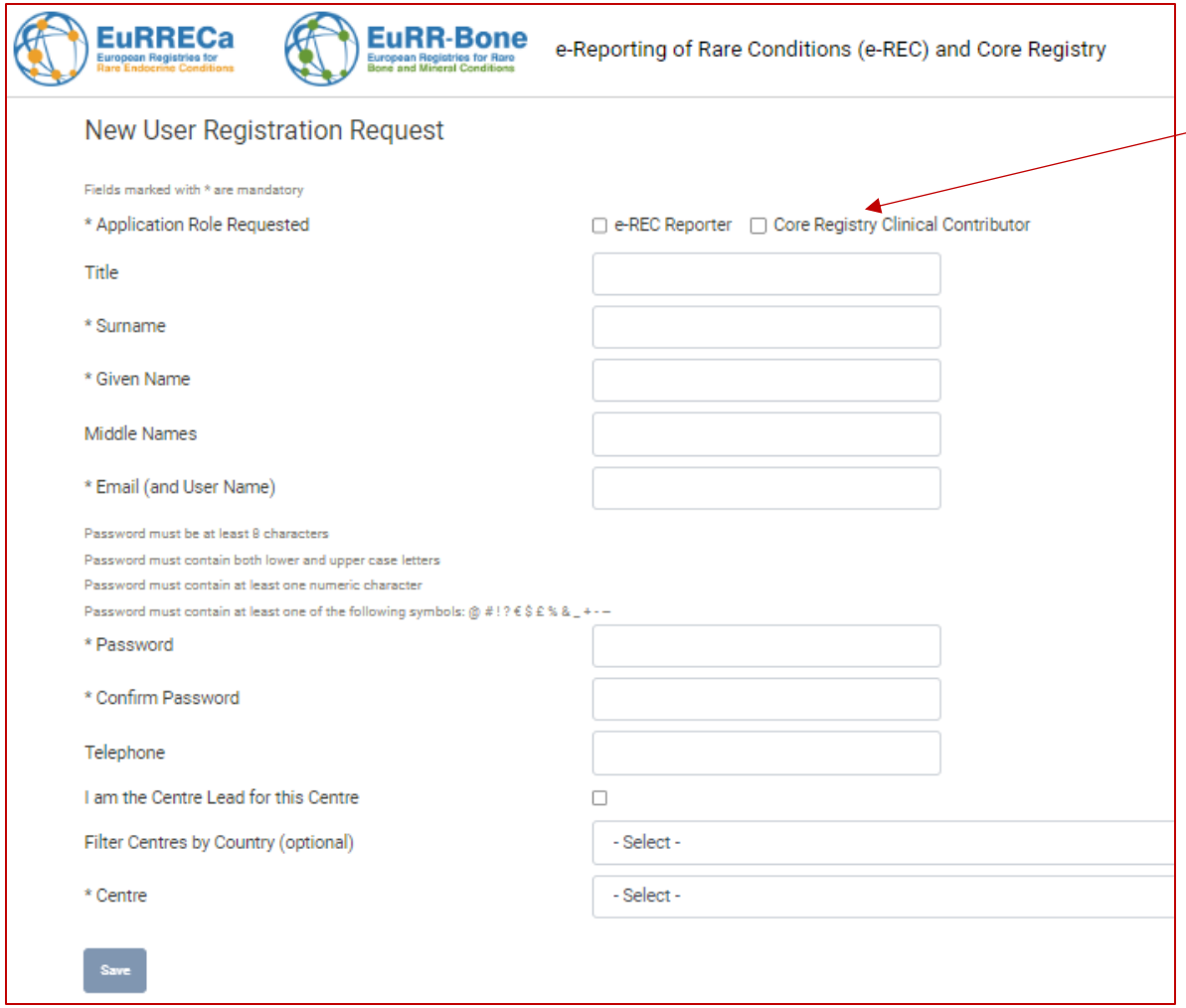
Register in the Core registry

Go to https://eurreb.eu/

If you have no account yet, you can request the access here or choose "information" from the "access" button

Register in the Core registry

Follow the self-registration link:



You need an access to Core registry to use the module

Fill in the mandatory information.
Your request will be approved as soon as possible.

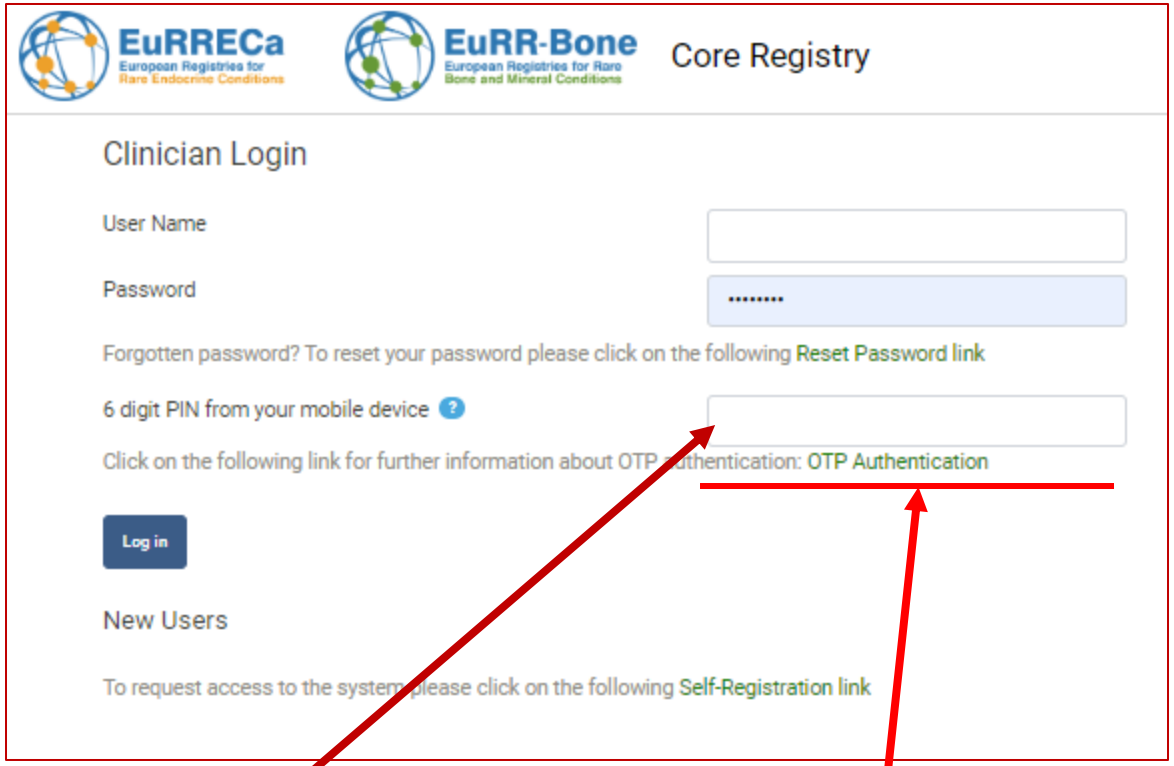
Login to the Core registry

Go to https://eurreb.eu/

Login to Core registry using this button and your credentials

Login to the Core registry

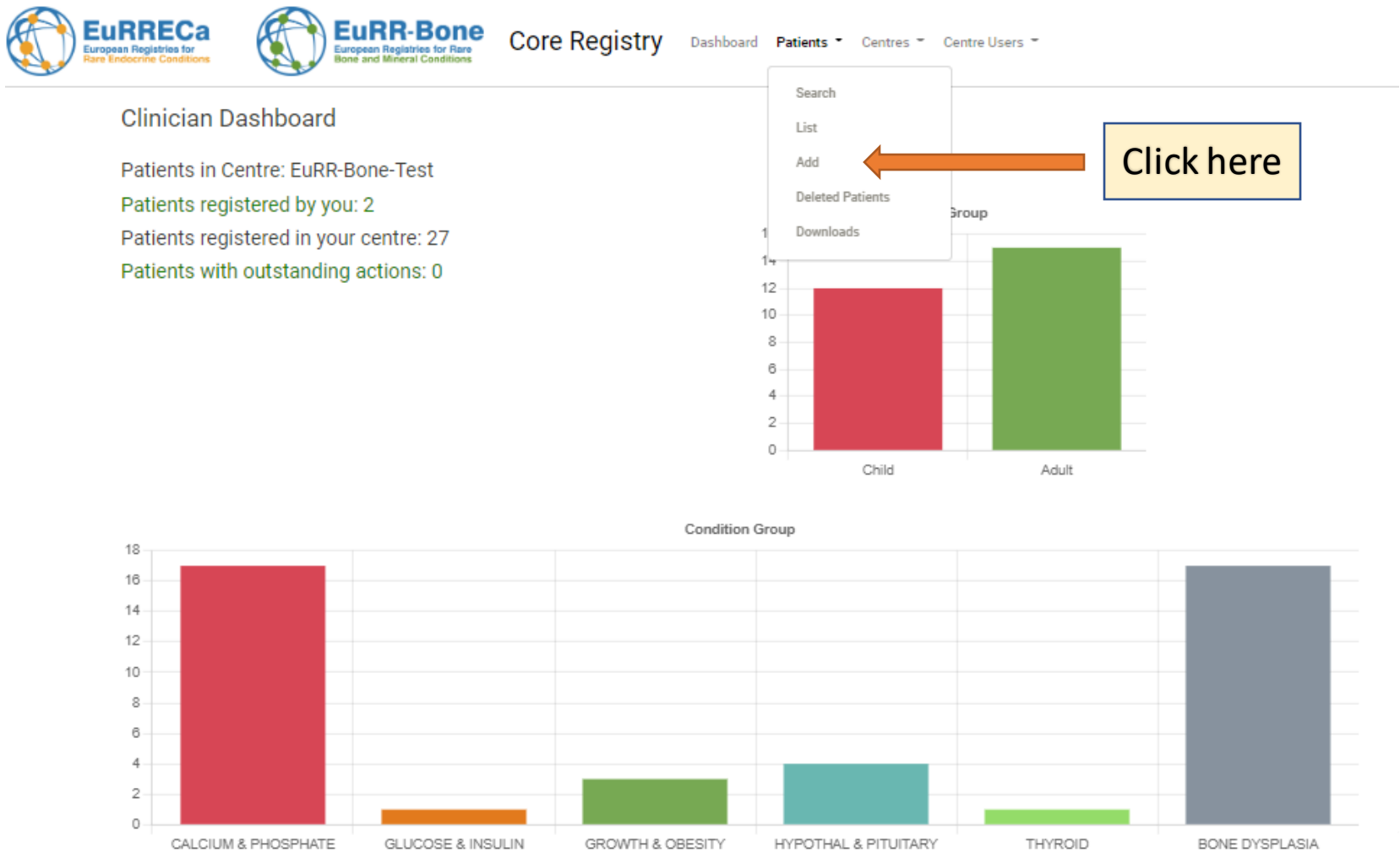
Choose clinician login and then provide your credentials:



Provide your credentials and 6 digit PIN, if needed, reset your password

You will need to download OTP authenticator

Add a new patient



Fill in the Common Data Elements

Mandatory fields

Select **"Calcium & Phosphate"**

Add Patient

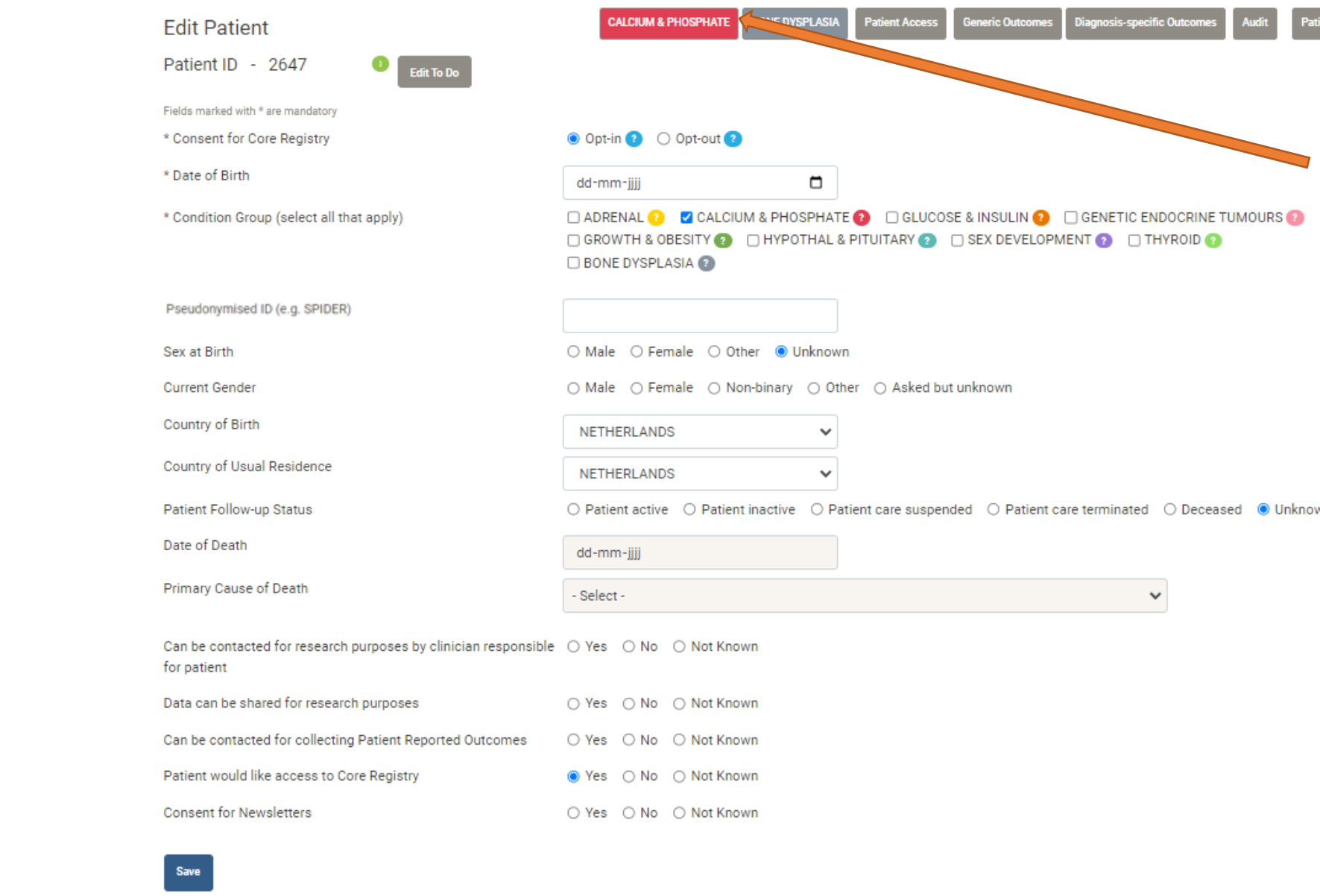
Fields marked with * are mandatory

* Consent for Core Registry	○ Opt-in ○ Opt-out
* Date of Birth	dd-mm-jjjj
* Condition Group (select all that apply)	☐ ADRENAL ☑ CALCIUM & PHOSPHATE ☐ GLUCOSE & INSULIN ☐ GENETIC ENDOCRINE TUMOURS ☐ GROWTH & OBESITY ☐ HYPOTHAL & PITUITARY ☐ SEX DEVELOPMENT ☐ THYROID ☐ BONE DYSPLASIA
Pseudonymised ID (e.g. SPIDER)	
Sex at Birth	○ Male ○ Female ○ Other ◉ Unknown
Current Gender	○ Male ○ Female ○ Non-binary ○ Other ○ Asked but unknown
Country of Birth	NETHERLANDS
Country of Usual Residence	NETHERLANDS
Patient Follow-up Status	○ Patient active ○ Patient inactive ○ Patient care suspended ○ Patient care terminated ○ Deceased ◉ Unknown
Date of Death	dd-mm-jjjj
Primary Cause of Death	- Select -
Can be contacted for research purposes by clinician responsible for patient	○ Yes ○ No ○ Not Known
Data can be shared for research purposes	○ Yes ○ No ○ Not Known
Can be contacted for collecting Patient Reported Outcomes	○ Yes ○ No ○ Not Known
Patient would like access to Core Registry	○ Yes ○ No ○ Not Known
Consent for Newsletters	○ Yes ○ No ○ Not Known

SAVE

Save Cancel

Go to the Patient Condition section



A navigation menu will appear, click on **"Calcium & Phosphate"**

Edit the Patient Condition section

Edit Patient Condition

Core Data | Generic Outcomes | Documents | Audit | Patient List

Patient ID · Date of Birth

01

Edit To Do

Calcium and Phosphate

Senior Clinician < 18 - N/A

Senior Clinician ≥ 18 - N/A

e-REC ID

First Contact with Centre dd-mm-jjjj

Clinician Responsible for Patient Not Assigned

Date of First Clinical Manifestations dd-mm-jjjj

Select a Clinician Responsible for the patient

Primary Condition Pseudohypoparathyroidism

Specific Diagnosis Pseudohypoparathyroidism (iPPSD)

- **Select the "Pseudohypoparathyroidism" as Primary Condition**
- **Select a Specific Diagnosis from the list**

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 dd-mm-jjjj

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

Save | Cancel

Save

Access the Disease-specific Module

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

Core Data | Generic Outcomes | Diagnosis-specific Outcomes | Documents | Audit | Patient List

Edit Patient Condition

Patient ID - 01 Date of Birth

Edit To Do

Calcium and Phosphate

Senior Clinician < 18 - N/A

Senior Clinician ≥ 18 - N/A

Click here to fill in the disease-specific module

e-REC ID

First Contact with Centre: dd-mm-jjjj

Clinician Responsible for Patient

Date of First Clinical Manifestations: dd-mm-jjjj

Primary Condition: Pseudohypoparathyroidism

Specific Diagnosis: Pseudohypoparathyroidism (iPPSD)

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

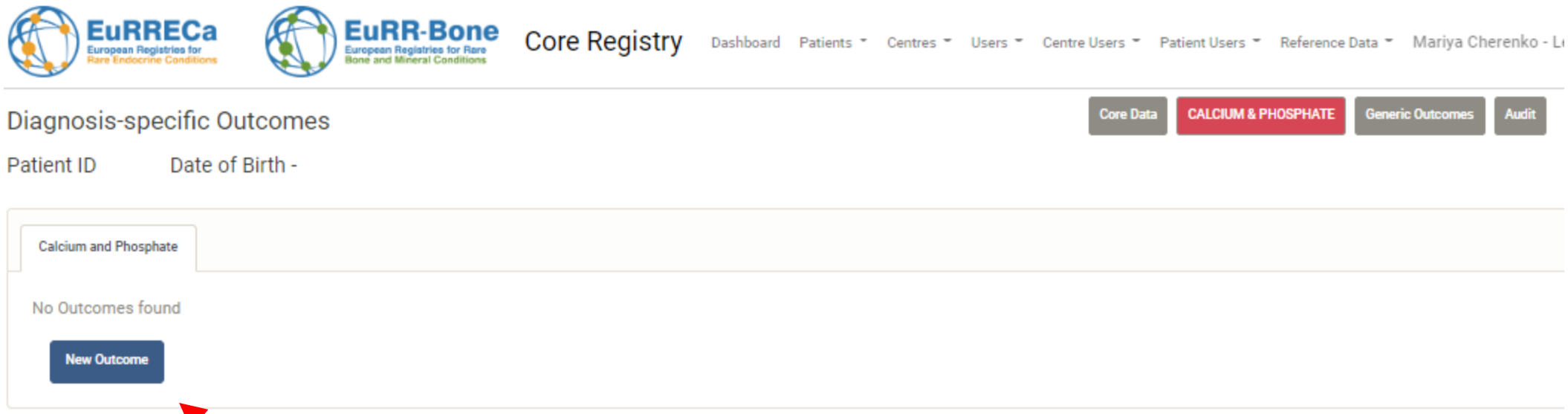
How Was Diagnosis Reached: ☐ Clinical ☐ Biochemistry ☐ Genetic ☐ Histology ☐ CT ☐ MRI ☐ PET ☐ USS ☐ X-Ray ☐ Other ☐ Not Known

Date of Diagnosis: dd-mm-jjjj

Participation in Detailed Disease Registry: ○ Yes ○ No ○ Not Known

Save Cancel

Fill in a Disease-specific outcome



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

Fill in the data into the module and save

Patient ID - Date of Birth Condition Group - Calcium and Phosphate

Questionnaire: iPPSD outcome questionnaire v3 - iPPSD outcome questionnaire v3

New Outcome

Date of Birth	e-REC ID	Primary Condition	Pseudohypoparathyroidism
Current Gender	Clinician Responsible for Patient	Specific Diagnosis	Pseudohypoparathyroidism (iPPSD)
		Date of Diagnosis	

Perinatal features | Genetic findings | Endocrine Status | Auxology parameters | Clinical findings | Clinical musculoskeletal features | Radiological musculoskeletal features | Other Radiological findings | Therapy | Surgical interventions

Assessment Date dd-mm-jjjj

Age at Assessment Date

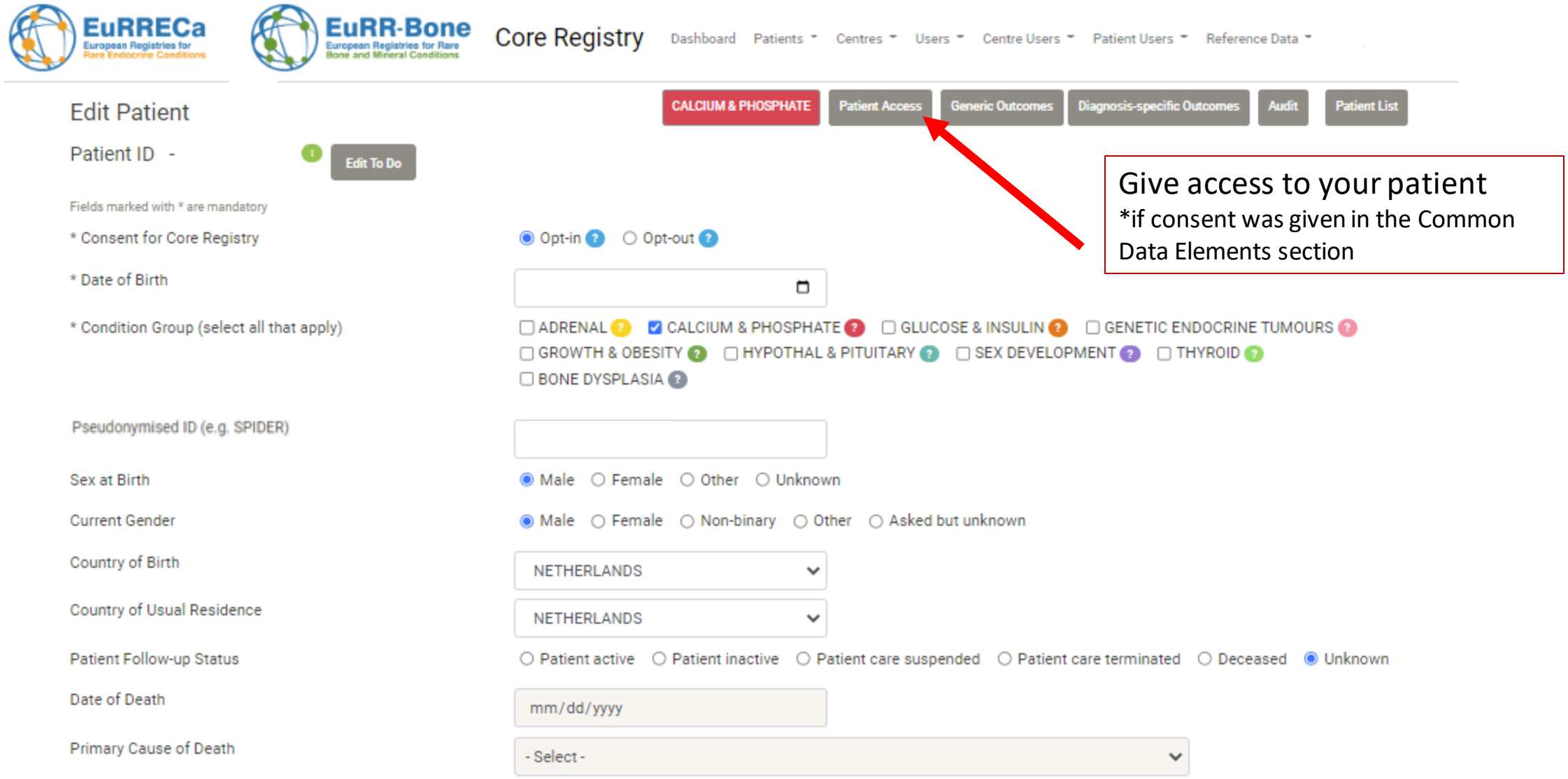
Gene variant (cDNA)

Gene variant (protein)

Gene

- [ ] PTH1R
- [ ] GNAS, maternal allele
- [ ] GNAS, paternal allele
- [ ] GNAS, parental allele not known
- [ ] abnormal methylation at least at GNAS A/B DMR
- [ ] PRKAR1A
- [ ] PDE4D
- [ ] PDE3D
- [ ] Other
- [ ] No genetic analysis
- [ ] Unknown

Give the Patient access to the Core Registry



Give access to your patient
*if consent was given in the Common Data Elements section

Contact us!

Please do not hesitate to contact us by e-mail at
registries@lumc.nl in case of any questions

We invite you to join our regularly drop-in sessions https://eurreb.eu/