



The European  
Rare Kidney Disease Registry

## Newsletter April 2022

### About ERKReg

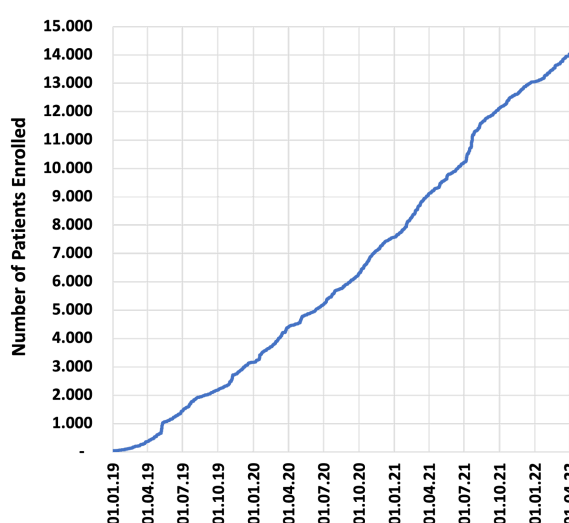
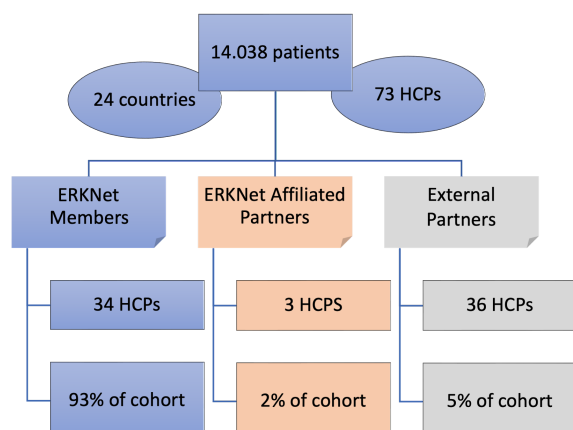
The European Rare Kidney Disease Registry "ERKReg" is **the largest and only patient registry for all rare kidney diseases**.

ERKReg collects basic demographic data as well as disease-specific information on treatment and disease progression and indicators of quality of care.

### Aims of ERKReg

The ERKReg Registry is intended to facilitate the **identification of patient cohorts** for clinical trials. The monitoring of performance indicators should contribute to the **harmonisation and optimisation of the diagnosis and treatment** of rare kidney diseases in the long term. In addition, the collected data can be used to answer **research-specific questions**.

### Data inclusion by 1st April 2022



## New data items and features

For patients with proven causative gene mutation

### Harmonization of genetic data

Genetic data entry will now follow the HGVS (Human Genome Variation Society) nomenclature.

It is mandatory to indicate the DNA sequence change (c.), whereas the amino-acid change (p.) and the DNA reference sequence number (NM\_) are optional. Centre queries will be sent out to correct data that were previously entered in the wrong format.

Genetic diagnostics for primary renal diagnosis

Date of screening (leave field empty if unknown) 10/10/2000 (dd/mm/yyyy)

Was a causative gene abnormality identified? Yes

Receipt date of genetic results 11/10/2000 (dd/mm/yyyy)

Methods

☒ Individual gene testing

☐ NGS gene panel screening

☐ Exome sequencing

Inheritance	Affected gene	Zygosity
AD	PKD1	Heterozygous

Mutation (e.g. NM\_000123.4, c.123A>G, p.Arg123His)

Type of variant (c.): >, del, dup, inv, ins, delins

c. 123A>G Mandatory field

p. 7 Optional field

NM\_ Optional field

Further comments on genetics

For patients with Cystinosis

### Collecting medication history

For patients with Cystinosis (Orphacode: 213), treatment with Cystagon or Procysbi can be recorded in the medication database. Upon saving a visit update, a notification reminds you for completing the data.

500-0021 P M-11/2013 Metabolic nephropathy CKD4 Next visit due: 14/05/2020

Basic data Add visit Termination

Previous visits: 14/05/2019

Medications Extracorp. Therapies

Medication history

ongoing medication

terminated medication

Medication	Start date	Single dose	Frequency	Dose/kg/day	Stop date
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Click on a medication to edit or [Add medication]

Note: If you cannot find the medication on the list, please contact tanja.wlodkowski@med.uni-heidelberg.de.

Medication \*NEW\*

Medication Cysteamine (Cystagon)

Start date (dd/mm/yyyy)

Stop date (dd/mm/yyyy)

Leave empty if medication is ongoing

For patients with Fabry disease

### Collecting medication history and NEW key performance indicator (KPI)

For adult patients with Fabry disease (Orphacode: 324), treatment with Replagal or Fabrazyme can be recorded in the medication database. Upon saving a visit update, a notification reminds you for completing the data. In addition, a new key performance indicator has been added: % adult male Fabry disease patients receiving enzyme replacement therapy.

500-0023 P M-11/2013 Metabolic nephropathy CKD4 Next visit due: 23/03/2023

Basic data Add visit Termination

Previous visits: 23/03/2022

Medications Extracorp. Therapies

Medication history

ongoing medication

terminated medication

Medication	Start date	Single dose	Frequency	Dose/kg/day	Stop date
------------	------------	-------------	-----------	-------------	-----------

Click on a medication to edit or [Add medication]

Note: If you cannot find the medication on the list, please contact tanja.wlodkowski@med.uni-heidelberg.de.

Medication \*NEW\*

Medication Agalsidase alpha (Replagal)

Start date (dd/mm/yyyy)

Stop date (dd/mm/yyyy)

Leave empty if medication is ongoing

Important: If you spot any technical issue, please let us know!

## **Include your patients to the sub-registries**

We encourage all ERKReg sites to additionally add their patients to the disease-specific sub-registries!

- ERKReg also serves as a platform for **disease-specific subregistries**. The participation in certain sub-registries is optional. Upon approval an extended case report form is collected for the respective patients.
- Three sub-registries are active:
  - ERKNet/ESPN dRTA sub-registry
  - ERKNet/ESPN pediatric SLE sub-registry
  - Eurocys registry for cystinuria
- Sub-registries currently in programming stage:
  - Sub-registry for Bartter syndrome patients
  - Sub-registry for MPGN patients

Find more information about the sub-registries



### **SAVE THE DATE – Next ERKReg User Day | 8<sup>th</sup> June 2022**

We are planning to organize the **Third ERKReg User Day**. It will be held virtually on 8 June 2022 from 2:00–4:30 PM (CET). This meeting will be held for those **local team members who are in charge of entering patients** into the ERKReg database. We will present information on new registry features and encourage the exchange of user experience.

Please register for the meeting at: <https://forms.gle/znS1PLAg4rS3VApbA>

## Contact

The ERKReg registry is open to all interested nephrology services around the globe.

If you are interested in participating the ERKReg registry, please get in touch with the ERKReg project management:

ERKReg access and training /legal  
documentations/informed  
consent forms

IT support

Tanja Wlodkowski:

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Clémence le Cornec:

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Visit the ERKReg website

Read the ERKReg  
publication

Find useful documents



**Virtual participation  
click to register**



## ERKNet – The European Rare Kidney Diseases Reference Network

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