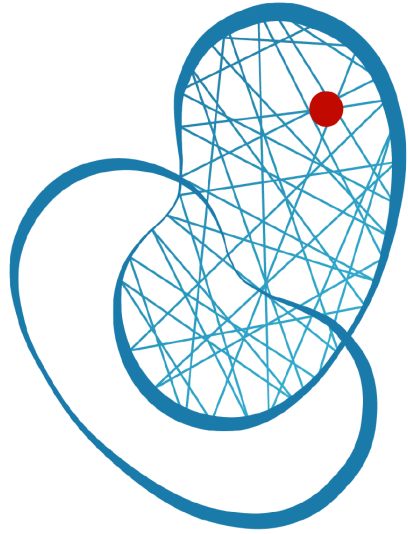




ERKNet

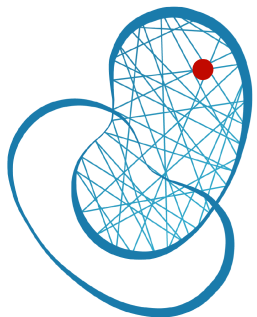
The European
Rare Kidney Disease
Reference Network

ERKNet: Rare kidney diseases
in a European framework

Franz Schaefer
Heidelberg University



Funded
by the European Union



ERKNet

The European
Rare Kidney Disease
Reference Network

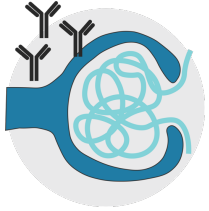
	Total
Full members	63
Affiliated Partners	9
Pediatric units	52
Adult units	43
Pediatric patients	33,214
Adult patients	34,777
Total patients	67,991
EU countries covered	24



Funded by
the European Union



Thematic Working Groups



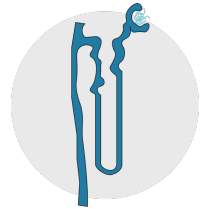
Immune Glomerulopathies

Co-Chairs: Dieter Haffner, Jürgen Floege,
Claire Dossier



Hereditary Glomerulopathies

Co-Chairs: Beata Lipska-Zietkiewicz,
Kalman Tory



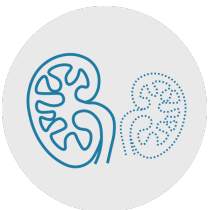
Tubulopathies

Co-Chairs: Tom Nijenhuis,
Gema Ariceta, Martin Konrad



AD structural diseases

Co-Chairs: Olivier Devuyst, Djalila Mekahli,
Jan Halbritter



CAKUT & Ciliopathies

Co-Chairs: Albertien van Eerde,
Ana Teixeira, Stefan Kohl



Pediatric Transplantation

Co-Chairs: Julien Hogan
Burkhard Tönshoff



TMA

Co-Chairs: Nicole van de Kar,
Savino Sciascia



Pediatric Dialysis

Co-Chairs: Claus Schmitt, Jerome
Harambat, Justine Bacchetta



Metabolic Nephropathies

Co-Chairs: Aude Servais, Elena Levtchenko,
Japp Groothoff



ERN Services to Rare Disease Community

Knowledge **Application**

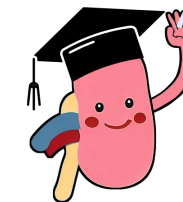
- Clinical Practice Guideline Development
- CPMS Online Consultation System

Knowledge **Dissemination**

- OrphaNet disease descriptions
- Disease information
- Webinars and eLearning
-> RD Curriculum
- Clinical exchange programme

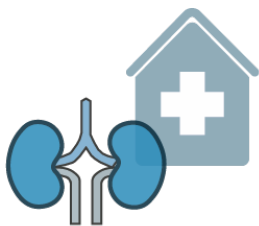
Knowledge **Generation**

- Online surveys
- Patient registries
- Research exchange programme
- Clinical trials
- Virtual data sharing and networking



Clinical experience

2 years
in the field of
rare kidney diseases



Webinars

3 years
every 2 weeks



including
Webinar-related
exams

54 topics
pediatric &
adult diseases

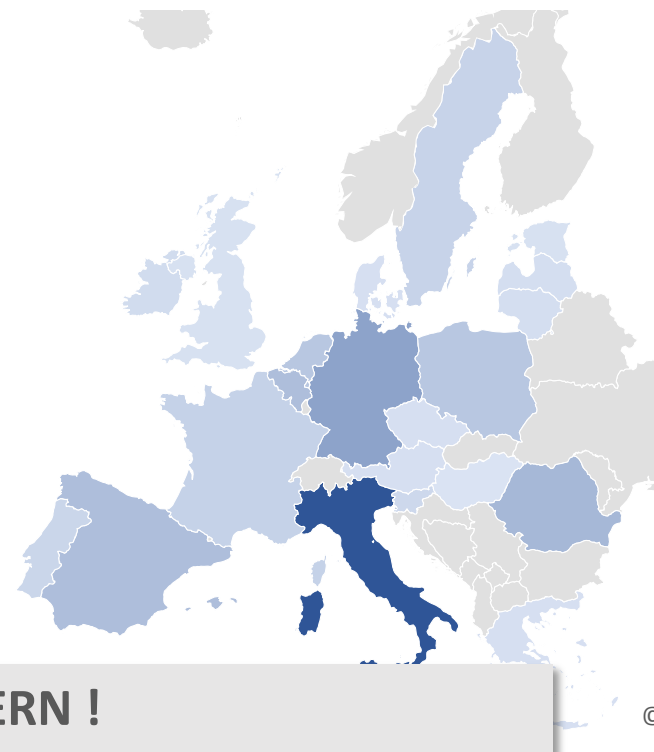
Requirements:
Attendance to 80% of ERKnet Webinars
≥ 75% correct answers in the exams

eLearning cases



topic related
cases
basic &
complex tests

Requirements:
Processing of 80% of all cases
≥ 75% correct answers



Italy	117
Germany	53
Romania	36
Belgium	31
Spain	31
Poland	24
Netherlands	24
France	15
Sweden	14
Portugal	12
Malta	11
Slovenia	10
Ireland	7
Lithuania	5
Latvia	5
Austria	4
Greece	4
Czech Rep.	4
Denmark	4
UK	3
Estonia	3
Hungary	1
Total	418

First Rare Disease Curriculum launched by an ERN !

- Class 1 from 2021 – 2023: 231 students from 59 ERKNet centres
- Class 2 from 2022 – 2024: 57 students from 26 ERKNet centres
- Class 3 from 2023 – 2025: 59 students from 22 ERKNet centres
- Class 4 from 2024 – 2027: 73 students from 32 ERKNet centres

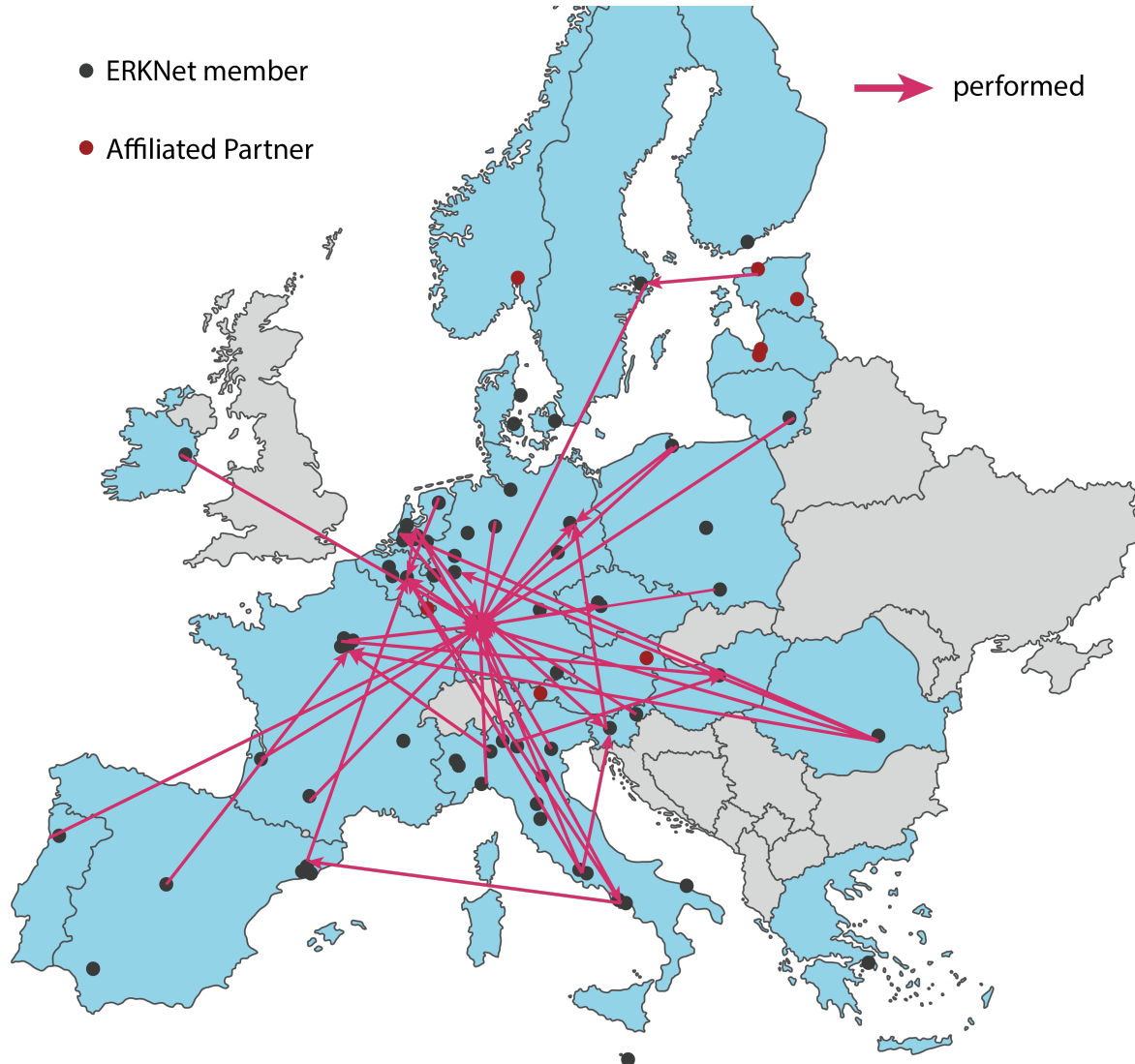


Clinical Exchange Programme



European
Reference
Networks

Exchange Programme
2021-2022



2021-2023: 63 exchanges, 91 person weeks

- 33 young clinicians in training
- 24 senior experts
- 6 project managers
- **30 ERKNet HCPs from 17 countries involved**

ERNet Guideline Development

Published

Congenital nephrotic syndrome	EJHG 2020, Nat Rev Nephrol 2021
Cystinuria	Kidney Int 2021
Bartter syndrome	Nat Rev Nephrol 2021
Distal renal tubular acidosis	NDT 2021
FSGS/SRNS post-transplant recurrence	Ped Transpl 2021
Nephrogenetic diagnostics	NDT 2021
Kidney dysplasia	NDT 2022
Lower urinary tract obstruction	Nat Rev Urol 2022
ADPKD management	NDT 2022
Primary hyperoxaluria	Nat Rev Nephrol 2023
Lipid apheresis in children	Atherosclerosis 2024
Bardet-Biedl Syndrome	EJHG 2024
Tuberous Sclerosis Complex	Nat Rev Nephrol 2024
Methylmalonic acidemia	Kidney Int Rep 2024
Nephrogenic diabetes insipidus	NDT in press

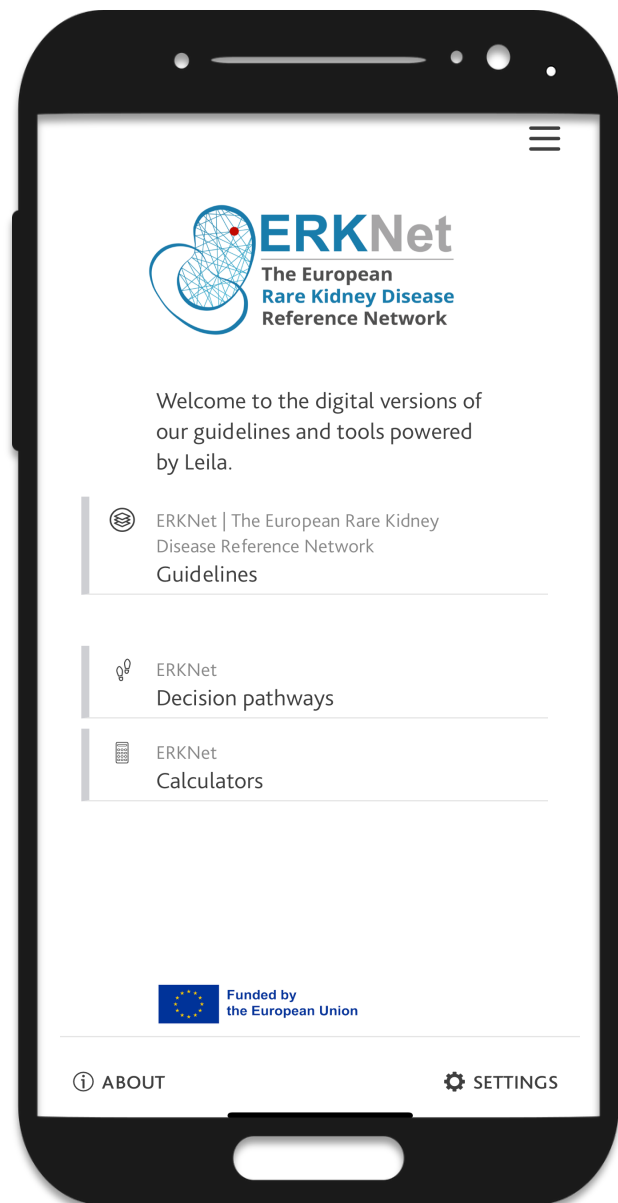
In development

Dent Disease	NDT under revision
Alport	NDT under revision
CV evaluation & management in ped CKD	Paper in preparation
ARPKD	Paper in preparation
ACEi/ARB in ped CKD	Paper in preparation
Monoclonal gammopathies of renal significance	Paper in preparation
Williams Beuren Syndrome	Paper in preparation
Lowe Syndrome	Paper in preparation
KRT initiation: strategic choices, modalities, timing, shared decision	Paper in preparation
Megaureter	Literature screening
Renal hypomagnesemia	Literature screening
Nephrocalcinosis	Literature screening
Postnatal PUV	in planning
VUR	in planning

Guideline Mobile App



ERKNet
Leila



Guidelines:

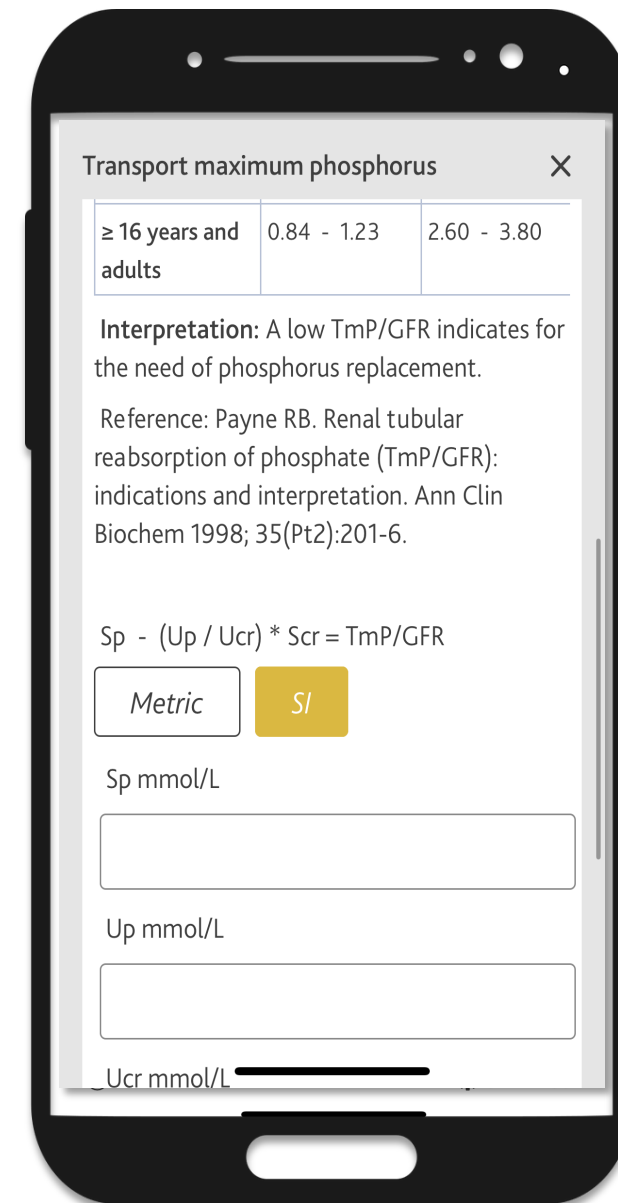
- ERKNet guidelines
- ERKNet endorsed guidelines
- Short cut to recommendations
- Full text
- Search function

Decision pathways:

- 7 algorithms integrated to date
- More to be extracted/developed

Calculators:

- Tubular function tests
- Acid-base disorders
- Urinary solute ratios
- eGFR



for Physicians

for Patients

What is... ▾ ERKNet ePAGs ▾ Patient Organisations Your Doctor Your Kidney Disease ▾ Media ▾ News Events ▾

Patient Journeys



Find your Doctor

For each rare kidney disease renowned ERKNet experts are listed with contact information.



Find your Centre

The patient website shows an interactive map to find an ERKNet expert centre in your country.



Find your Disease

The patient website provides information documents (brochures, websites, patient communities) on many rare kidney diseases in most European languages.



Find Patient Journeys

Experience reports from patients with rare kidney diseases to guide other patients through the different steps of the diagnosis and disease management.



Find your Patient Community

The patient website provides links to different patient organisations and communities in different countries to connect between patients.



Find more information here

Visit <https://patients.erknet.org> or scan the QR code



ERKNet Disease Information Brochures



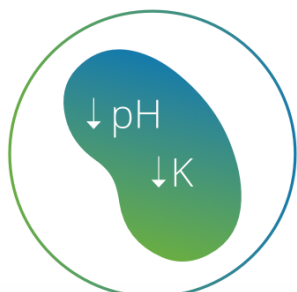
www.patients.erknet.org

[ERKNet ePAGs](#)[Patient Organisations](#)[Your Doctor](#)[Learn more about](#)[Your Kidney Disease](#)[Media](#)

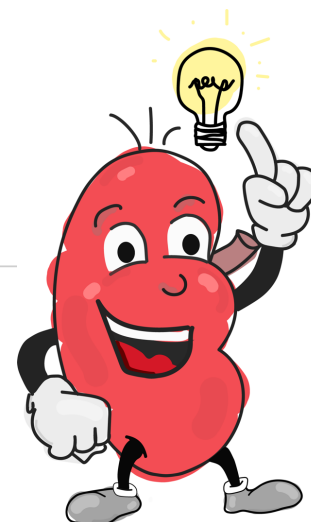
Alport Syndrome

[Read more](#)

ANCA-associated Vasculitis (AAV)

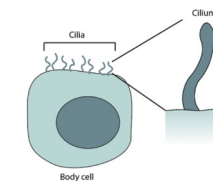
[Read more](#)

Autosomal Recessive Distal Renal Tubular Acidosis (dRTA)

[Read more](#)

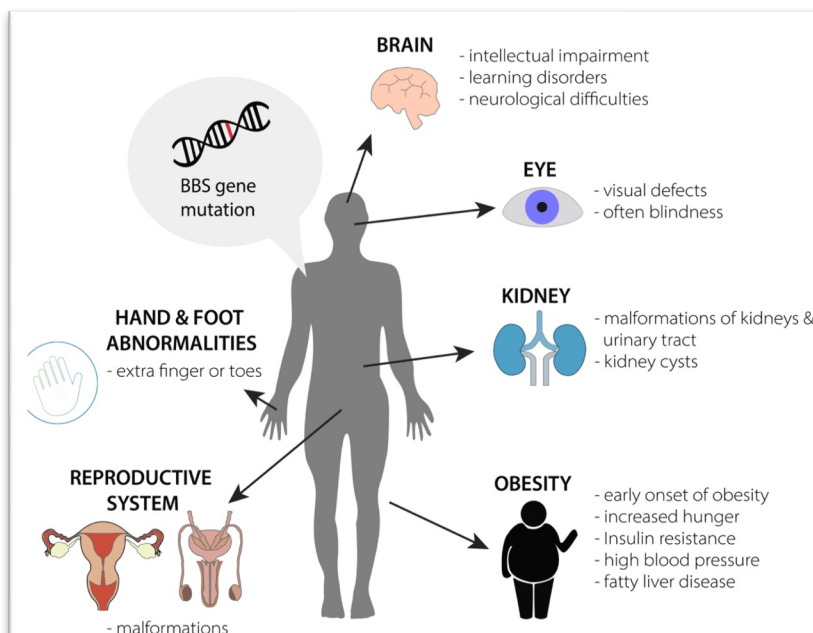
DISEASE DEFINITION

Bardet-Biedl syndrome (BBS) is a rare disease affecting several organs, including the kidneys. BBS is caused by an abnormally functioning cell component called a cilium (or cilia, plural) which is present on many cell types from different organs. Cilia are long, thin, hair-like projections that enable the cell to receive signals from outside and inside the cell. Bardet-Biedl syndrome is therefore categorized as a 'ciliopathy' (to find out more about cilia and their function please see pages 4 and 7).



Ciliopathies

Disease group which is related to abnormal cilia function.

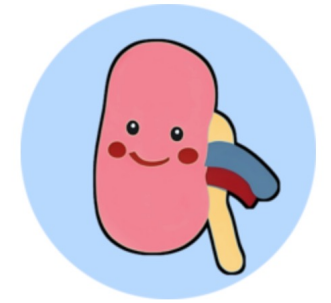


Multi-Level Patient Participation

- **Active patient participation** in Working Groups, Task Forces, ExCom, Network Board, Registry Board
- **Patient Disease Brochures:**
Patients co-write and review documents
- **Webinars:** Patient's voice
- **Patient Journeys:** 12 documents produced by ePAGs
- **Guideline Projects:** Obligatory patient involvement
- **Patient website:**
Specific contents, news and events for patients
- Regular ePAG **Feedback Sessions**
- **Patient Experience Survey**
- **Patient Initiated Research Projects**

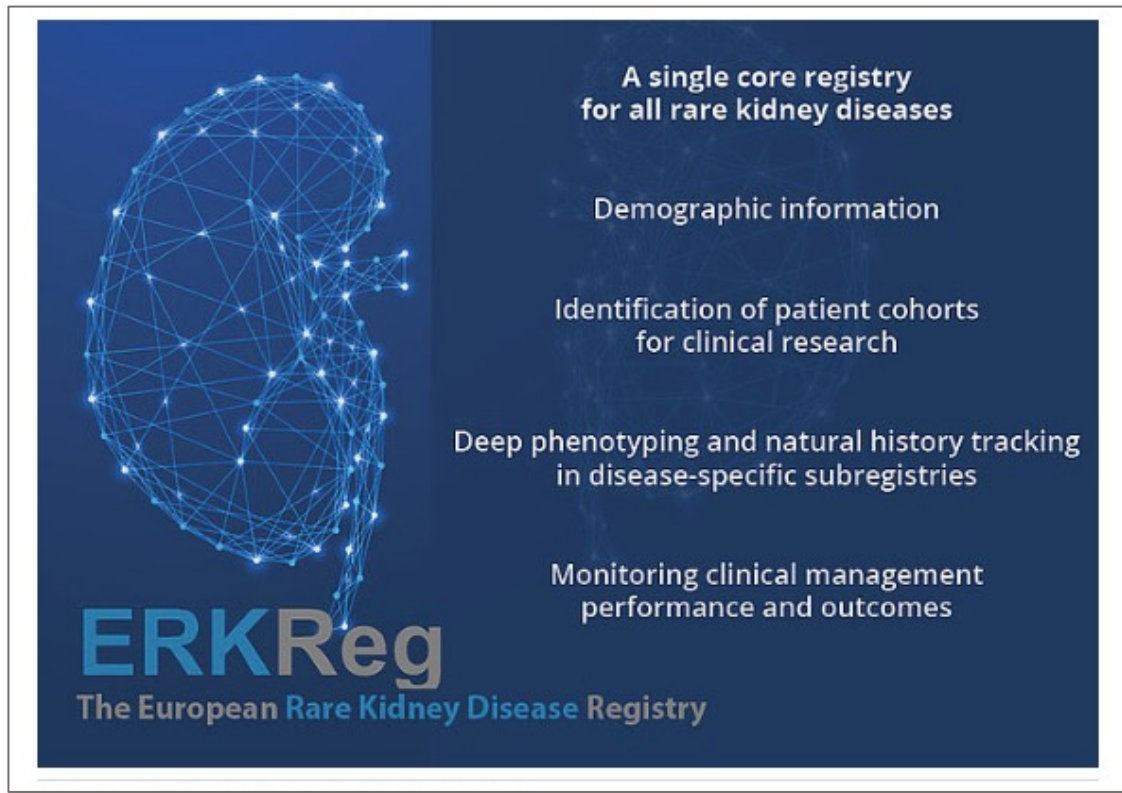


'ERKy-GPT'

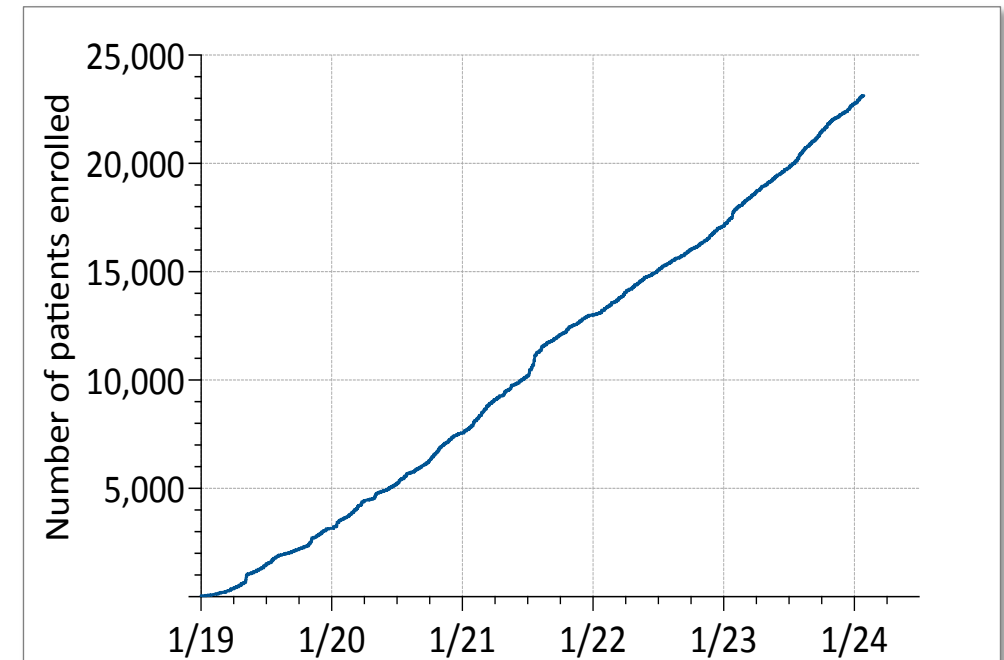
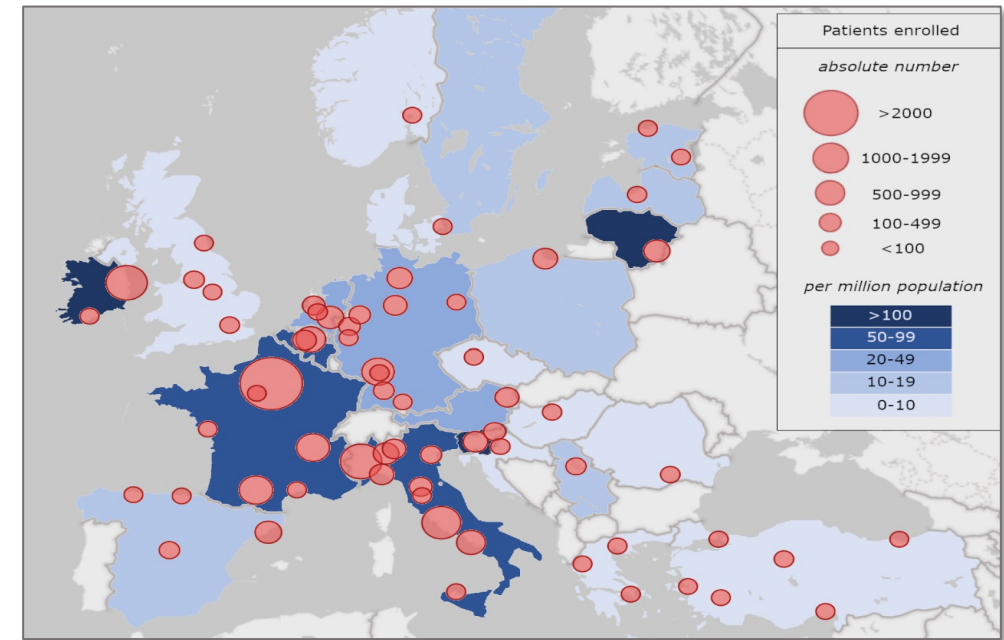


Hi, I am Erky-GPT!

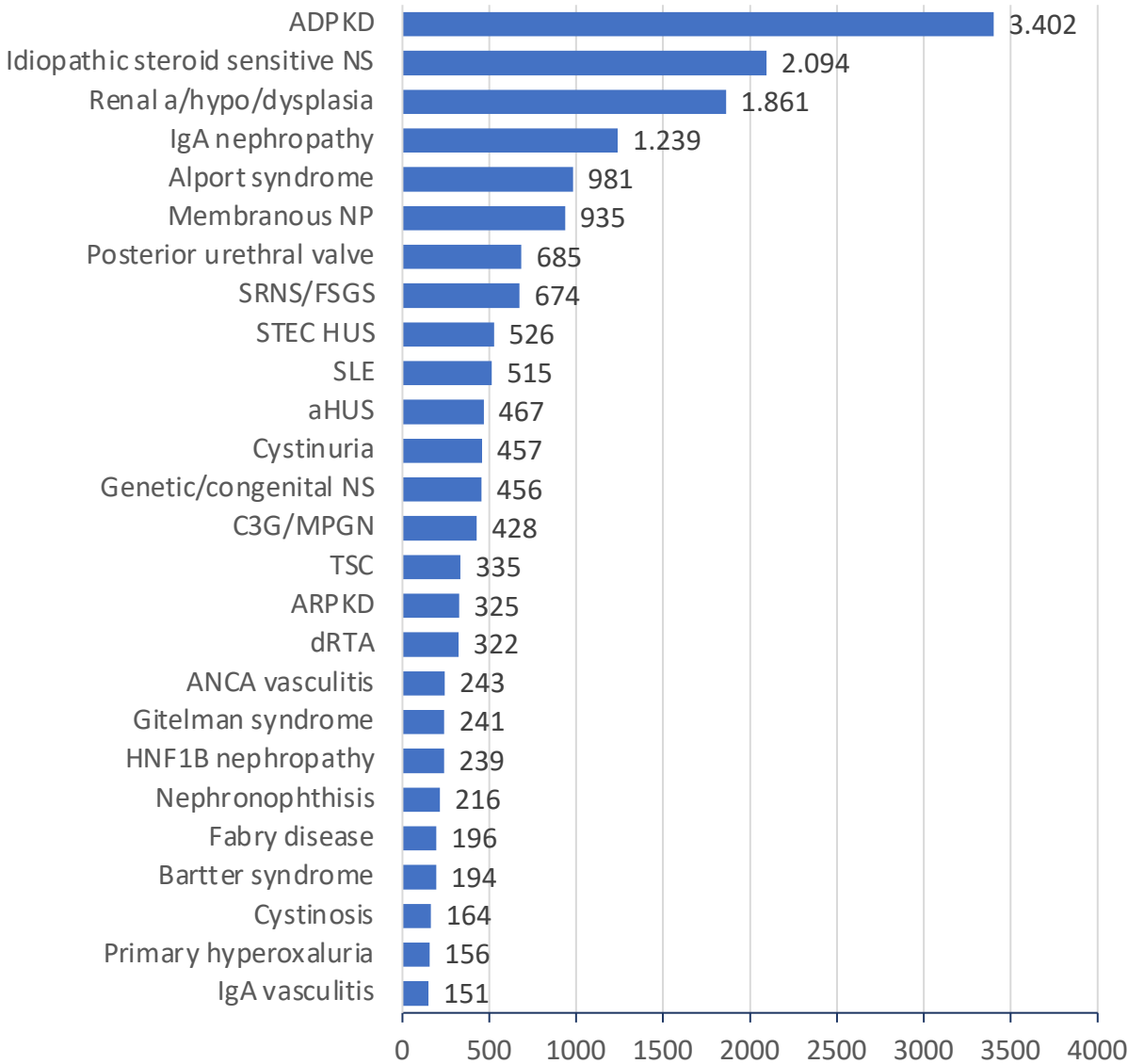
- Semantic AI tool for quick online consultations
 - Knowledge base: Ped Neph textbook, Clinical Practice Guidelines, patient brochures...
 - Beta testing by patients and healthcare professionals:
Answers too scientific and not sufficiently patient-friendly
- Expanded inclusion of patient-related peer-reviewed documents



- Centralized online registry
- 26,000 patients enrolled since 1/2019 at 80 specialized units in 24 countries
- 100 new patients added per week
- Annual follow-up achieved in 75%



Patient Cohorts for Clinical Research



Cohort Finder by diagnosis / disease group ▾

Diagnosis / disease group Nephrotic syndrome without extrarenal manifestations
(select diagnosis below)

[Any]

-

Glomerulopathies

-

Nephrotic syndrome without extrarenal manifestations

-

Nephrotic syndrome, idiopathic

Idiopathic steroid-sensitive nephrotic syndrome

Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance

-

Idiopathic steroid-resistant nephrotic syndrome

Idiopathic multidrug-resistant nephrotic syndrome

Idiopathic steroid-resistant nephrotic syndrome with sensitivity to second-line immunosuppressive therapy

-

Nephrotic syndrome, genetic

Genetic steroid-resistant nephrotic syndrome (SRNS)

Congenital nephrotic syndrome, Finnish type

Congenital nephrotic syndrome, no genetic cause specified

+

Glomerulopathy as part of a syndromic disorder

IgA nephropathy

Membranous glomerulonephritis

+

Collagenopathies (inc. Alport syndrome)

+

Primary membranoproliferative glomerulonephritis (MPGN)

+

Glomerulopathy as part of a systemic disorder

Idiopathic non-lupus full-house nephropathy

+

Tubulopathies

+

Metabolic nephropathies

+

Thrombotic microangiopathies

+

Renal or urinary tract malformations

+

Hereditary cystic renal diseases (ciliopathies)

+

Rare causes of hypertension

Current age

from years to years

Current treatment

Current CKD stage

All centers

Total: 1624 (seen in 39 ERKReg centers)

Active: 1413 (seen in 38 ERKReg centers)

Your center

Total: 116

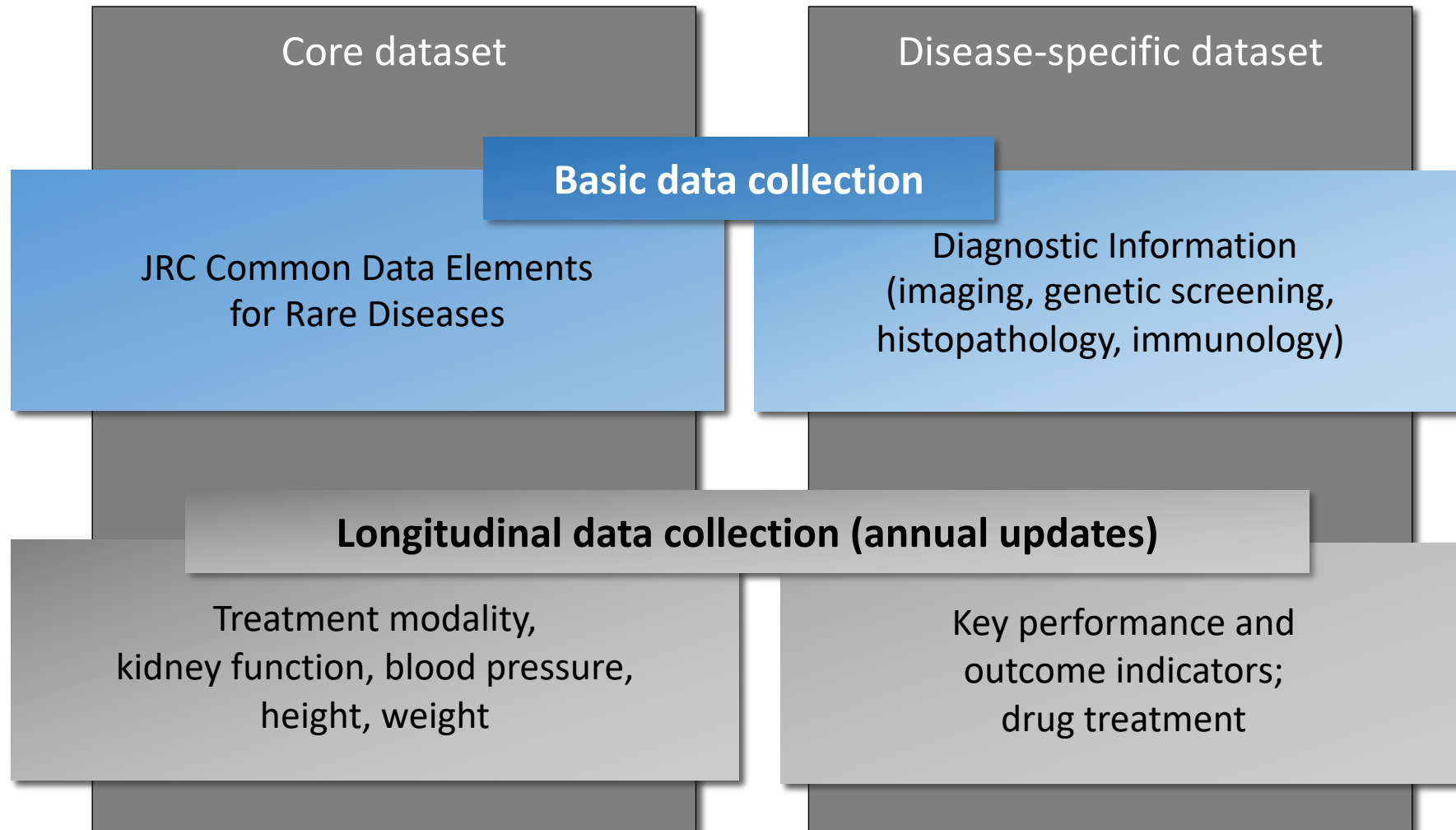
Active: 103

Data Capture



ERKReg

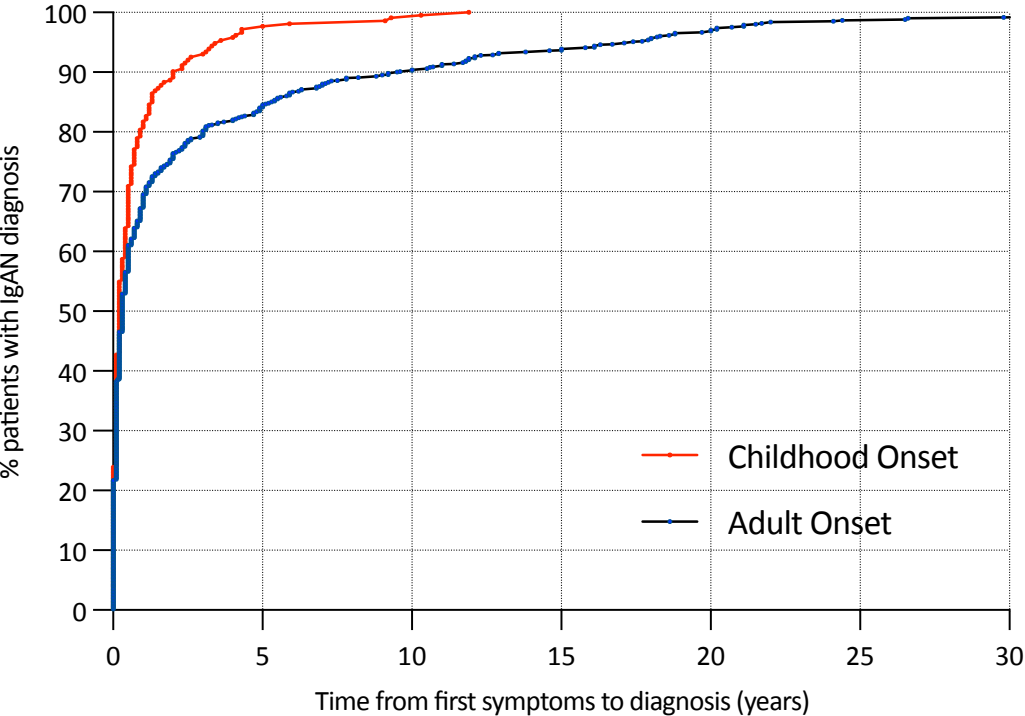
The European Rare Kidney Disease Registry



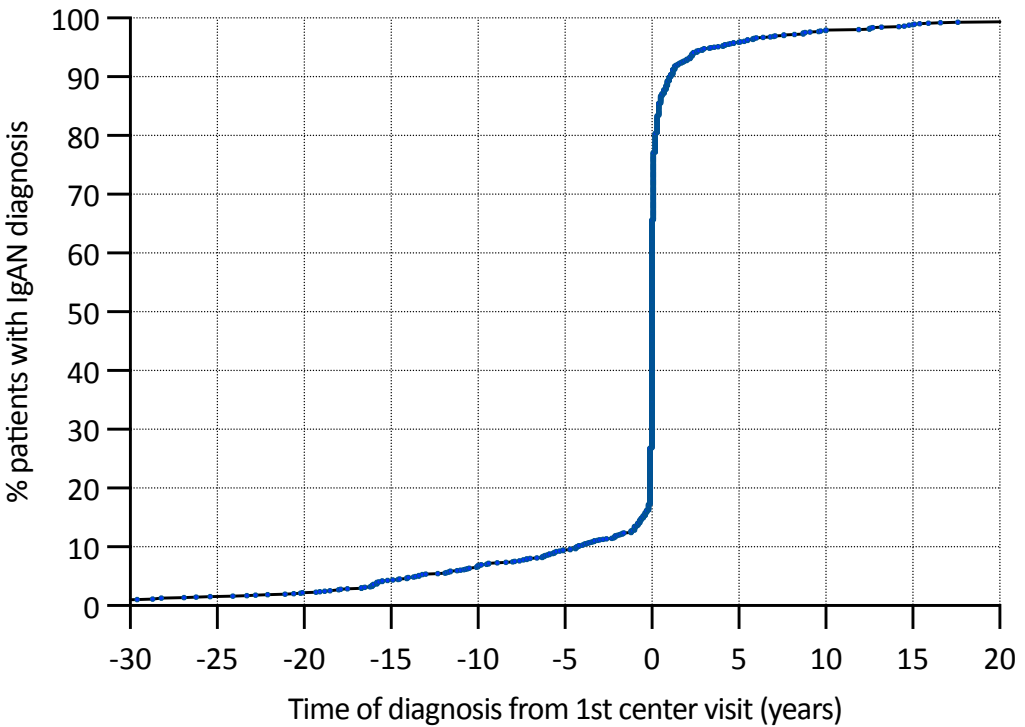
Demographic Information

IgA Nephropathy

Time from First Symptoms
to Diagnosis



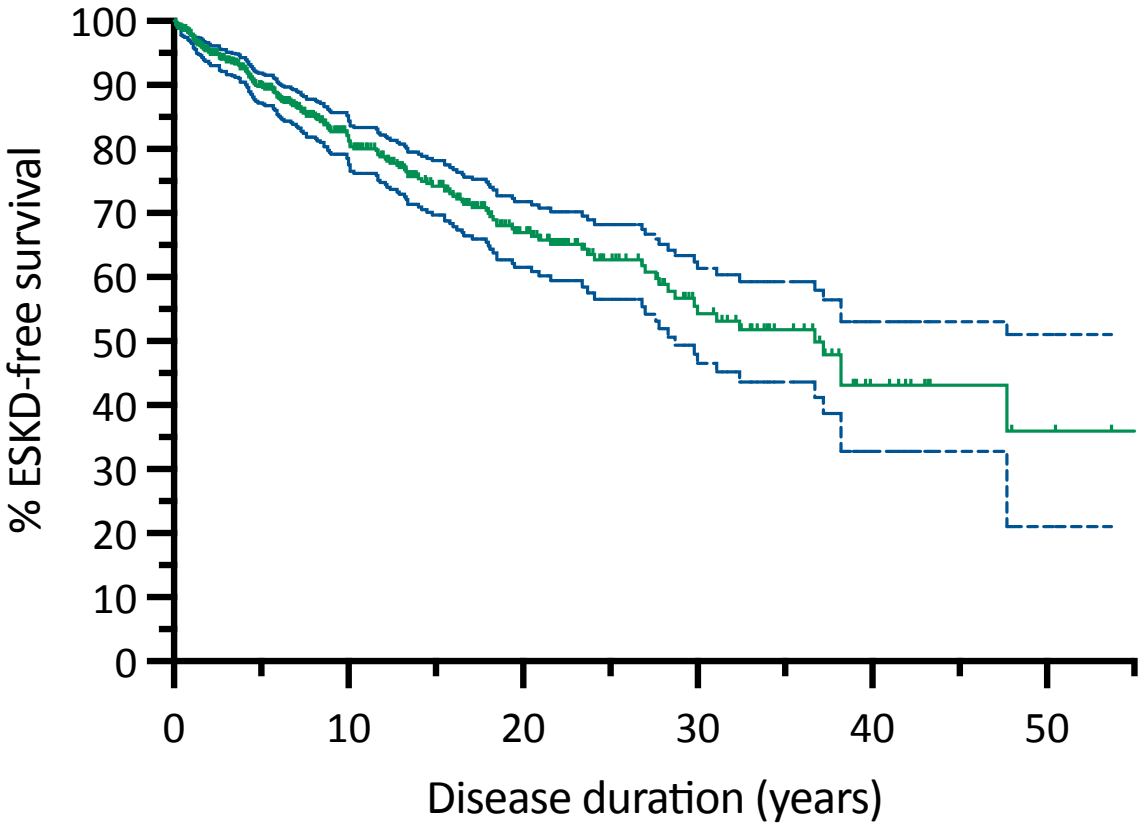
Time of Diagnosis Relative
to Referral to Reference Center



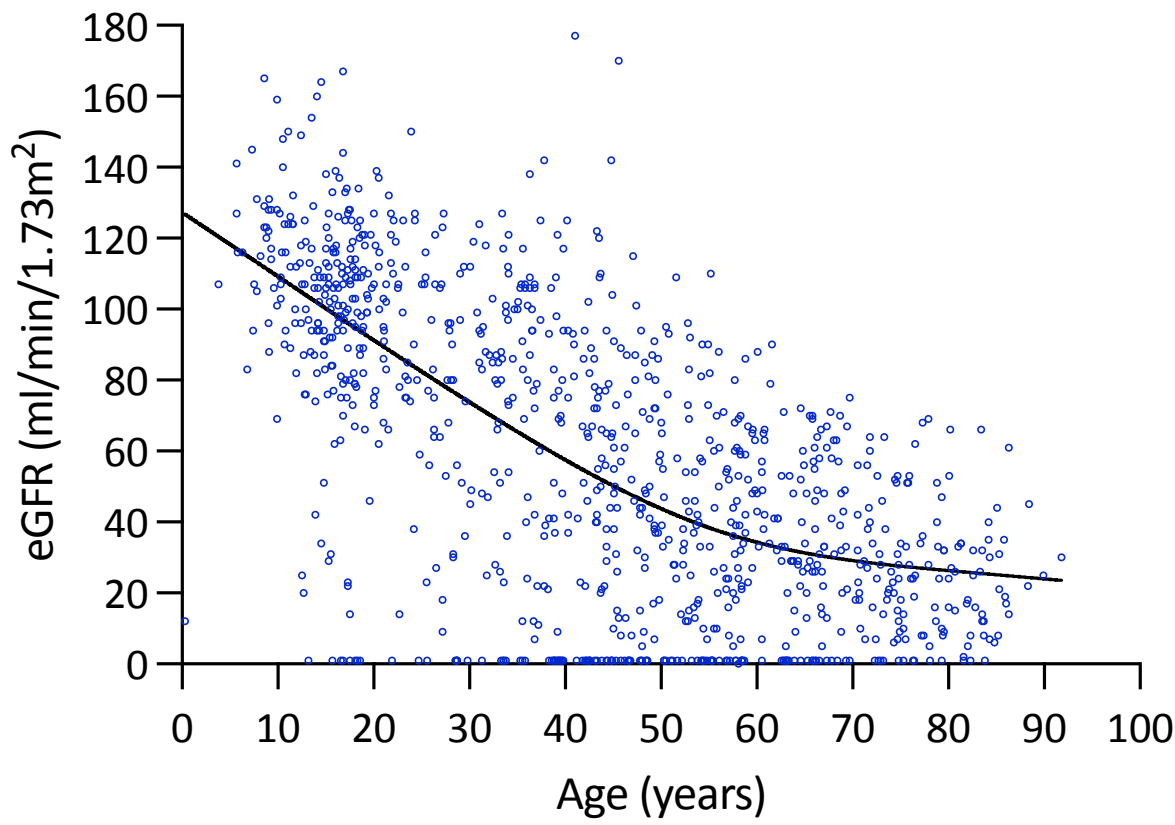
Disease Outcome Information

IgA Nephropathy

Kidney Survival



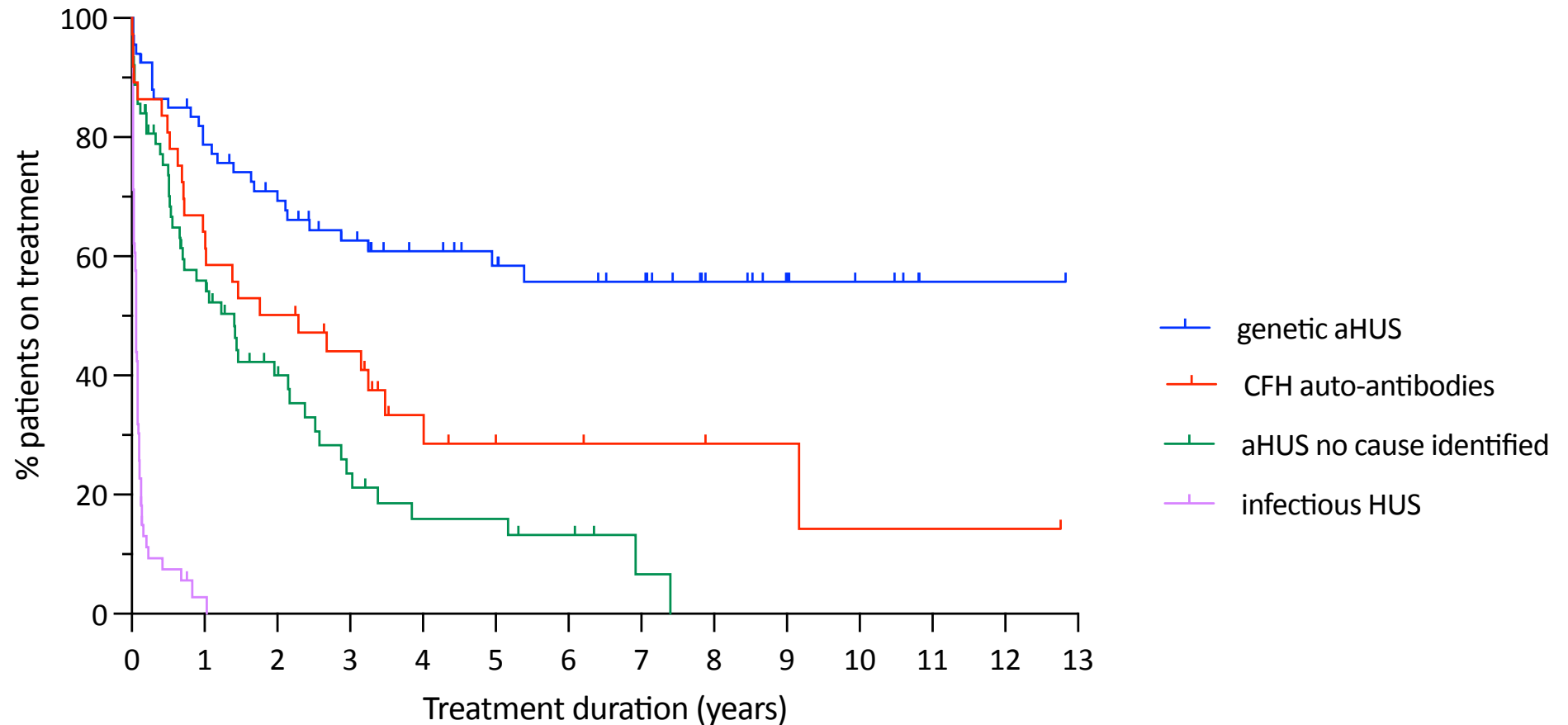
eGFR



Standard of Care Information

Hemolytic uremic syndrome

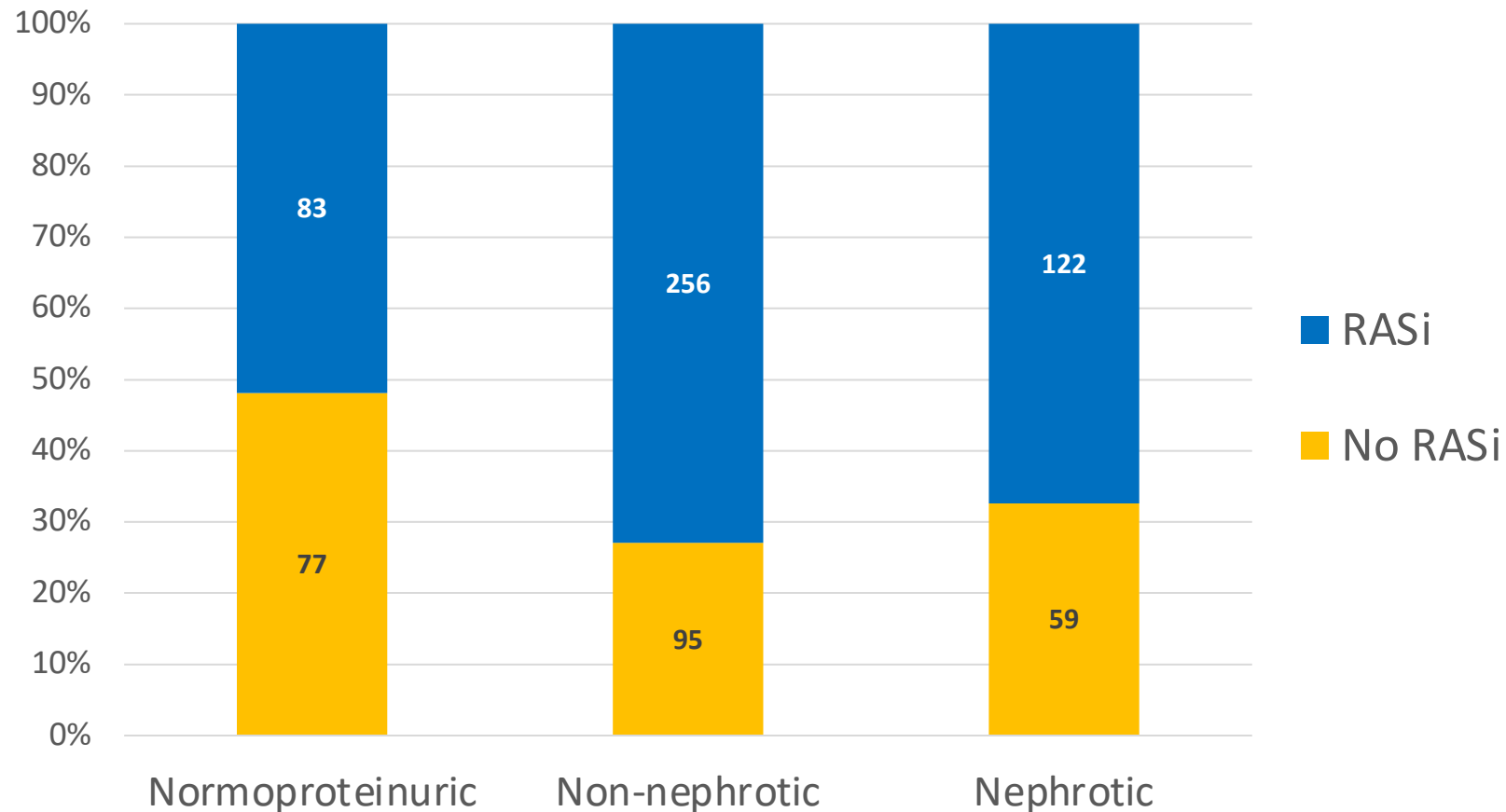
Time to discontinuation of C5 inhibitor therapy



Information on Guideline Adherence

RAS Inhibitor Treatment by Proteinuria in IgA Nephropathy

Last observation, 700 patients with CKD stage 1-4



Continuous Key Performance Indicator Monitoring

Your center

All ERKNet centers

KPI Monitoring

General

Glomerulopathies

Tubulopathies & metabolic
nephropathies

Thrombotic microangiopathies

Structural kidney disorders

CAKUT, Ciliopathies & OUP

Pediatric CKD

Pediatric Diet

CKD 3-5

% children (<16y) with height > 3rd percentile

81.1 %
(43/53)

78.5 %
(522/665)

% children (1-16y) with height < 3rd percentile on
growth hormone therapy

22.2 %
(2/9)

18.0 %
(22/122)

% children with BMI < 5th percentile

12.2 %
(9/74)

9.8 %
(79/809)

% children with BMI > 85th percentile

25.7 %
(19/74)

22.4 %
(181/809)

Children < 3 years with BMI < 5th percentile receiving
enteral feeds

50.0 %
(2/4)

23.8 %
(5/21)

% patients with office systolic BP < 75th percentile

28.2 %
(20/71)

32.9 %
(250/760)

% patients with office systolic BP < 50th percentile

8.5 %
(6/71)

11.7 %
(89/760)

% patients with hemoglobin > 11 g/dl

85.1 %
(63/74)

74.0 %
(599/809)

Serum phosphorus in normal range for age

91.9 %
(68/74)

81.7 %
(661/809)

% patients with serum bicarbonate > 20 mmol/L

90.5 %
(67/74)

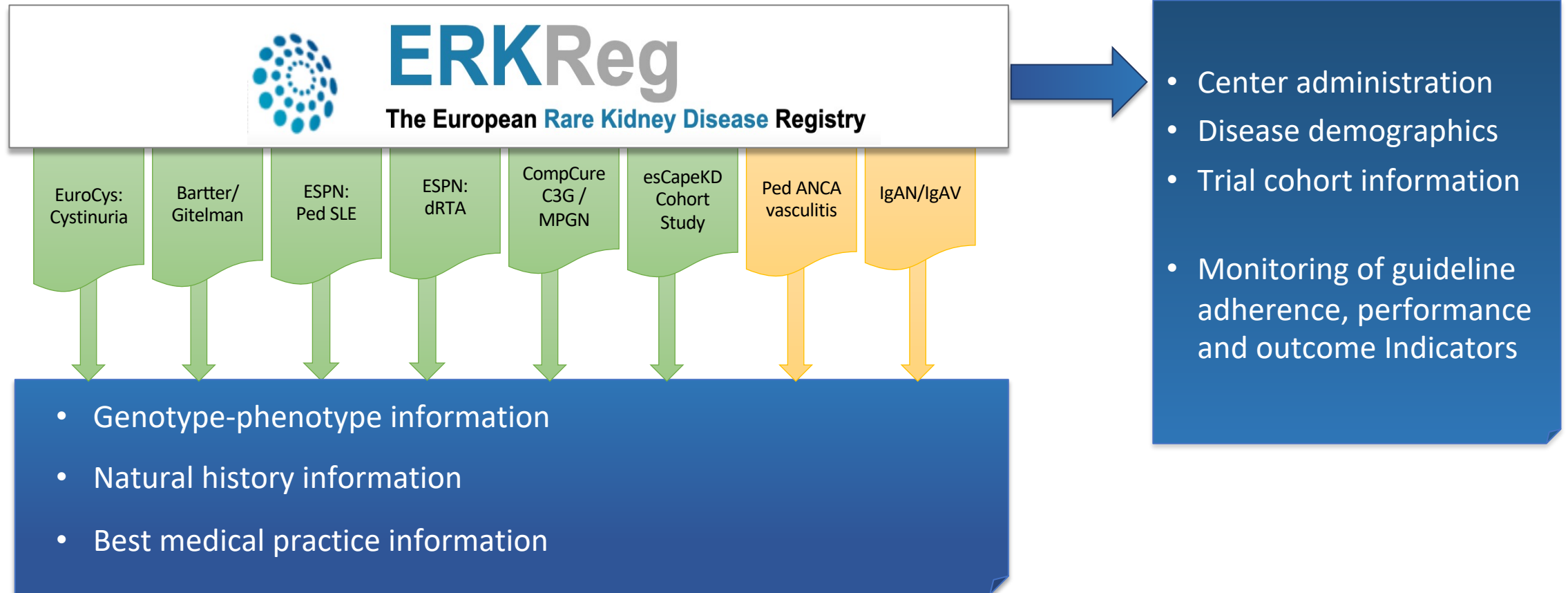
84.2 %
(681/809)



Interactions with Commercial Researchers

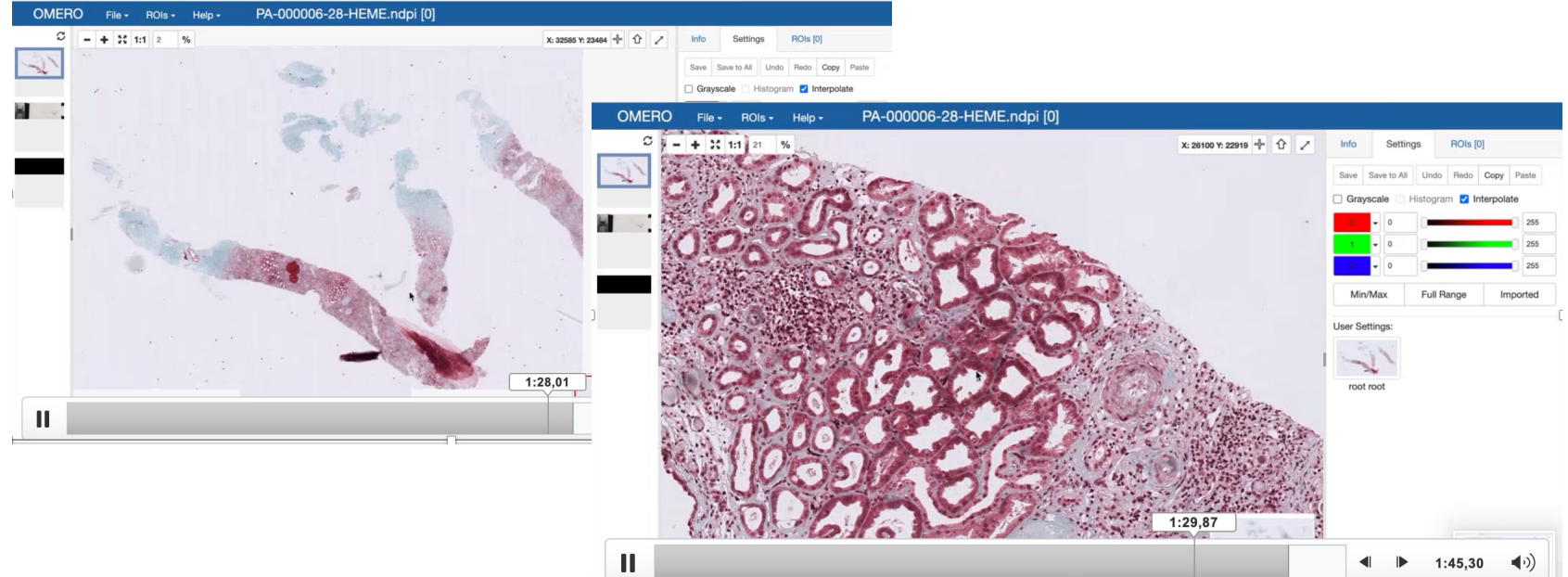
- **Brokerage function:** Central office identifies patients, informs HCPs and puts them in contact with company
 - Company A: Alport Trial – analysis completed
 - Company B: C3G trial – analysis completed
 - Company B: IgA Nephropathy trial – envisioned
 - Company B : atypical HUS trial – unfeasible
 - Company B : SLE nephritis – envisioned
 - Company C: Idiopathic NS – envisioned
- **Request for data access:** Provision of aggregated demographic and clinical data
 - Company C: aHUS – Report completed
 - Company B: C3G/IC-MPGN – Analysis ongoing
 - Company D: AMKD and ADPKD analysis – Analysis and survey ongoing
 - Company E: Cystinuria subregistry – in negotiation
 - Company F: Genetic podocytopathies – in negotiation
- **Development of sub-registries** on ERKReg platform for prospective data (natural history / SoC studies)
 - Compcure: C3G/MPGN cohort study – ongoing
 - Company G: Stone subregistry – ongoing, contracting in process
 - Companies H/I: Pediatric chronic kidney disease cohort study – in negotiation
 - Companies J/B: IgA Nephropathy – in negotiation
 - Company F: Genetic podocytopathies – envisioned

Modular Extensions: Disease Specific Sub-Registries



ERKNet Digital Histopathology Archive

- In collaboration with nephropathologists serving ERKNet centers
- Platform for upload of digitalized slides established
- Aims: Large case collections for classification studies
Empowerment of innovative AI approaches



Capture of Genetic Information



Genetic diagnostics for primary renal diagnosis

Date of screening

(leave field empty if unknown)

12/04/2023 (dd/mm/yyyy)

Was a causative gene abnormality identified?

Yes

Receipt date of genetic results

11/05/2023 (dd/mm/yyyy)

Methods

- ☐ Individual gene testing
- ☒ NGS gene panel screening
- ☐ Exome sequencing
- ☐ CNV-array / MLPA

Inheritance	Affected gene	Zygosity	Mutation (e.g. c.123A>G, p.Arg123His, NM_000123.4) Type of variant (c.) : >, del, dup, inv, ins, delins	
AR	NPHP1	Homozygous	c.	?_110881368_110962545del
			p.	?
			NM_	000272.4

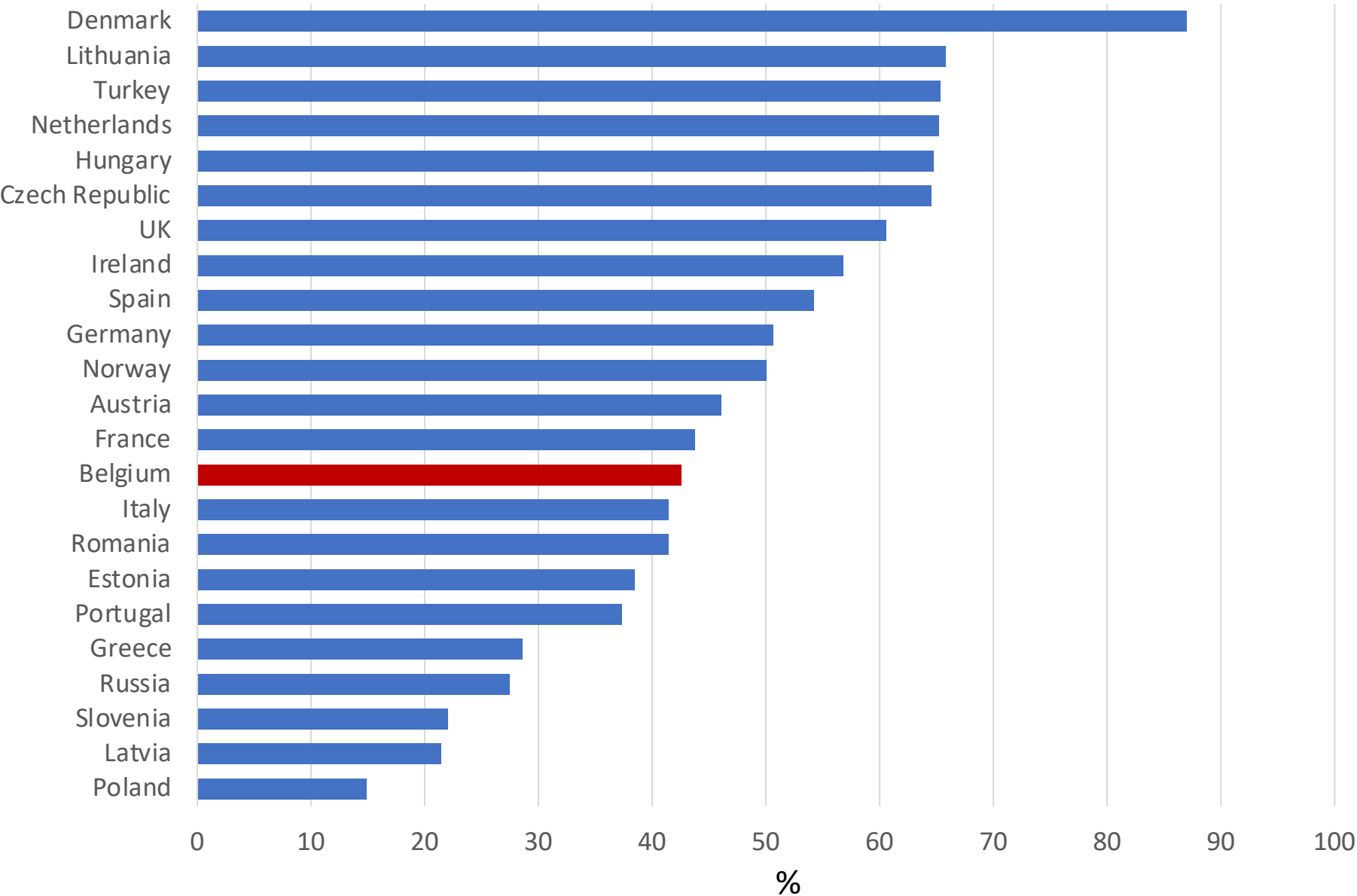
Mandatory
Optional
Optional

Further comments on genetics

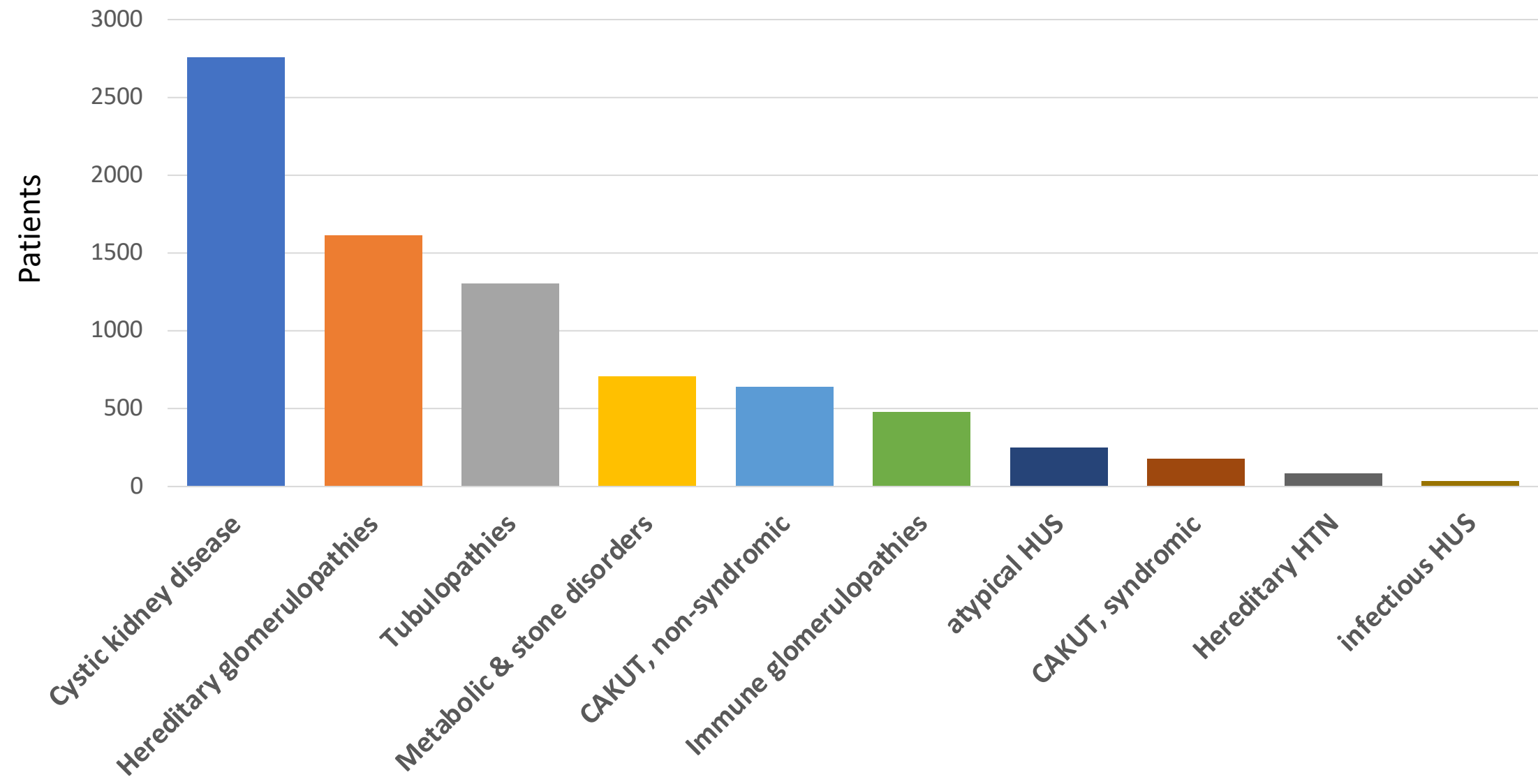
Genetic Testing Rate by Country



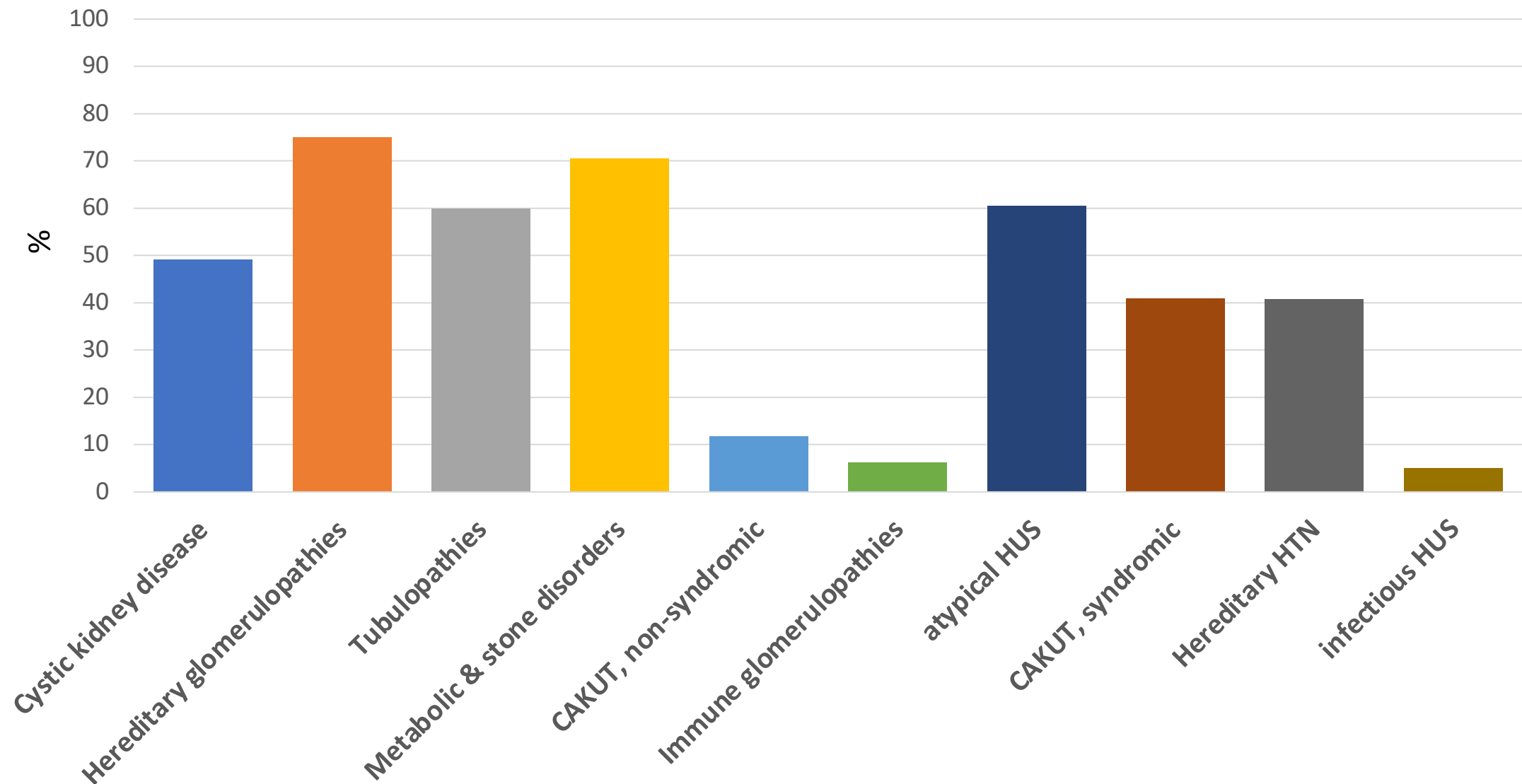
16,319 cases
with hereditary/
congenital disorders



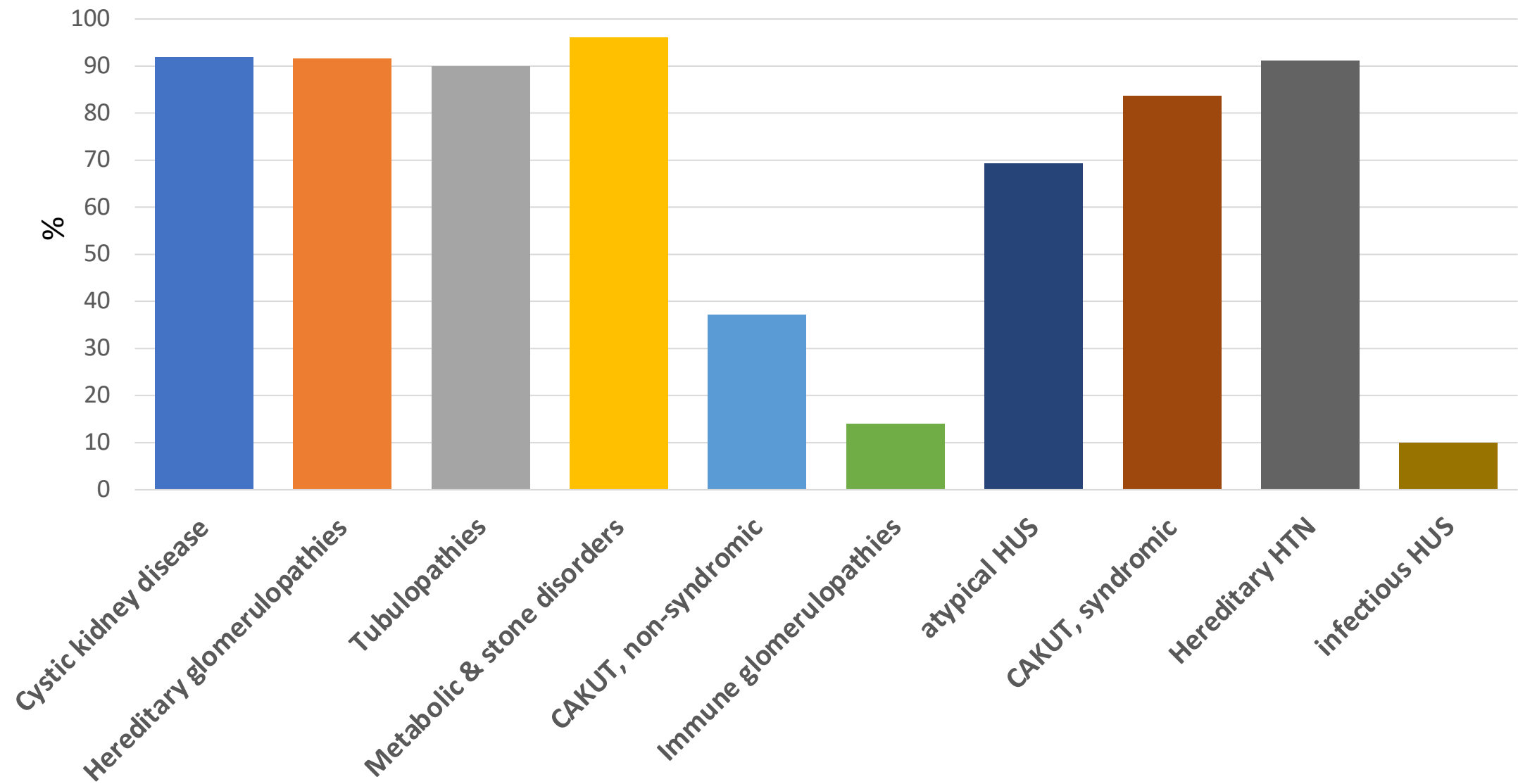
Genetic Testing by Disease Group



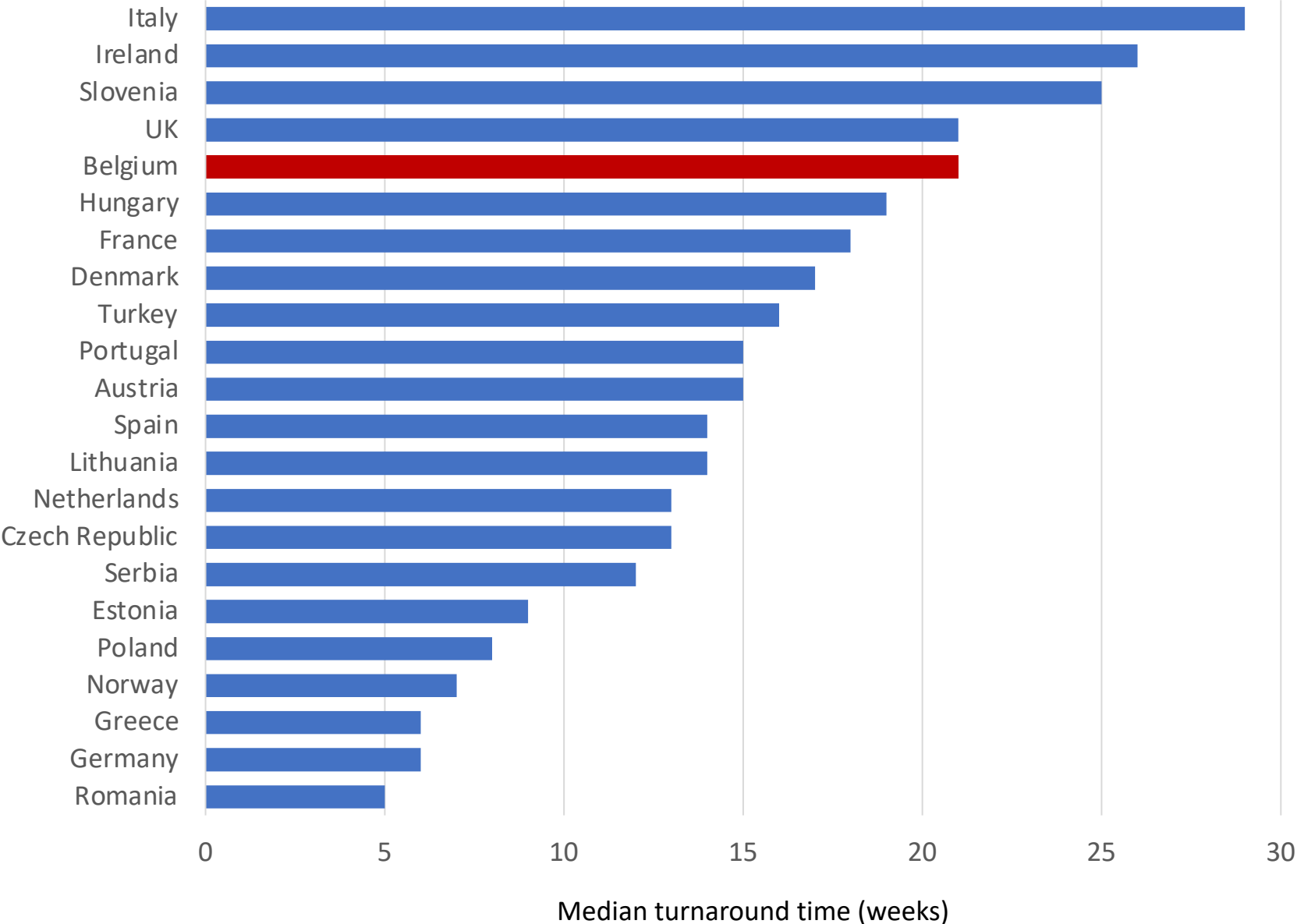
Testing Rate by Disease Group



Positive Testing Rate by Disease Group



Genetic Testing: Laboratory Turnaround Time

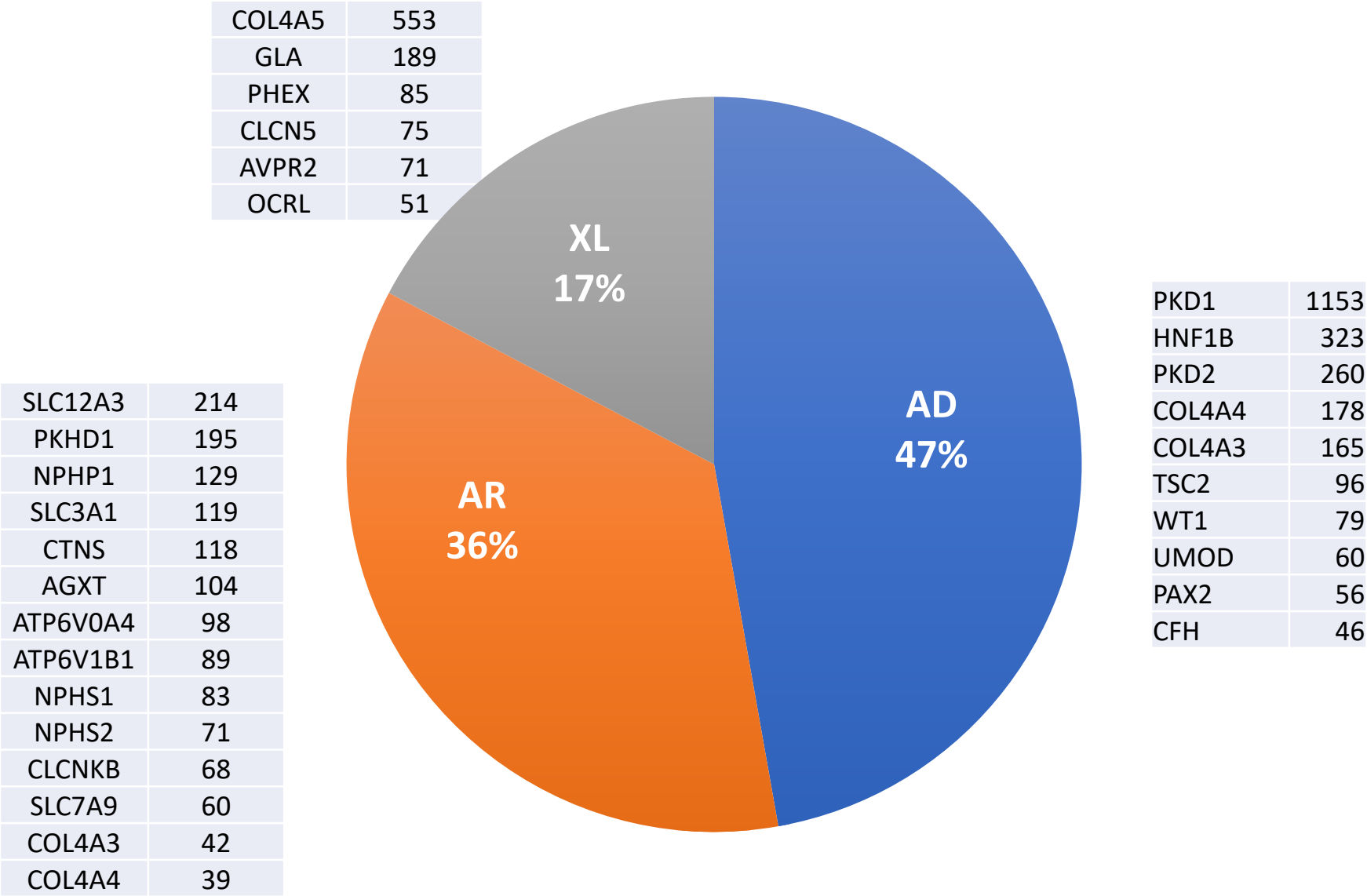


Most Common Causative Genes



ERKReg

The European Rare Kidney Disease Registry



New: ERKNet Research Exchange Programme

First 5 funded projects 2024/25:

Observational study on epidemiological data in STEC-HUS patients reported in ERKReg

N.vandeKar, Nijmegen & ERKReg Coordination Center

Presentation and early outcome in juvenile Lupus nephritis (ERKReg + subregistry data)

M.Vivarelli, Rome & ERKReg Coordination Center

Characterization of clinical courses of patients with kidney hypoplasia/dysplasia

S.Kohl & M. Liebau, Cologne & ERKReg Coordination Center

Genetic variant Interpretation and validation of ERKReg data

J. van der Velde & A. van Eerde, Groningen/Utrecht & ERKReg Coordination Center

Genotypic and phenotypic characterization of patients carrying SLC4A4 variants causing isolated or syndromic proximal RTA

F. Petzold, Leipzig

Thank you!

