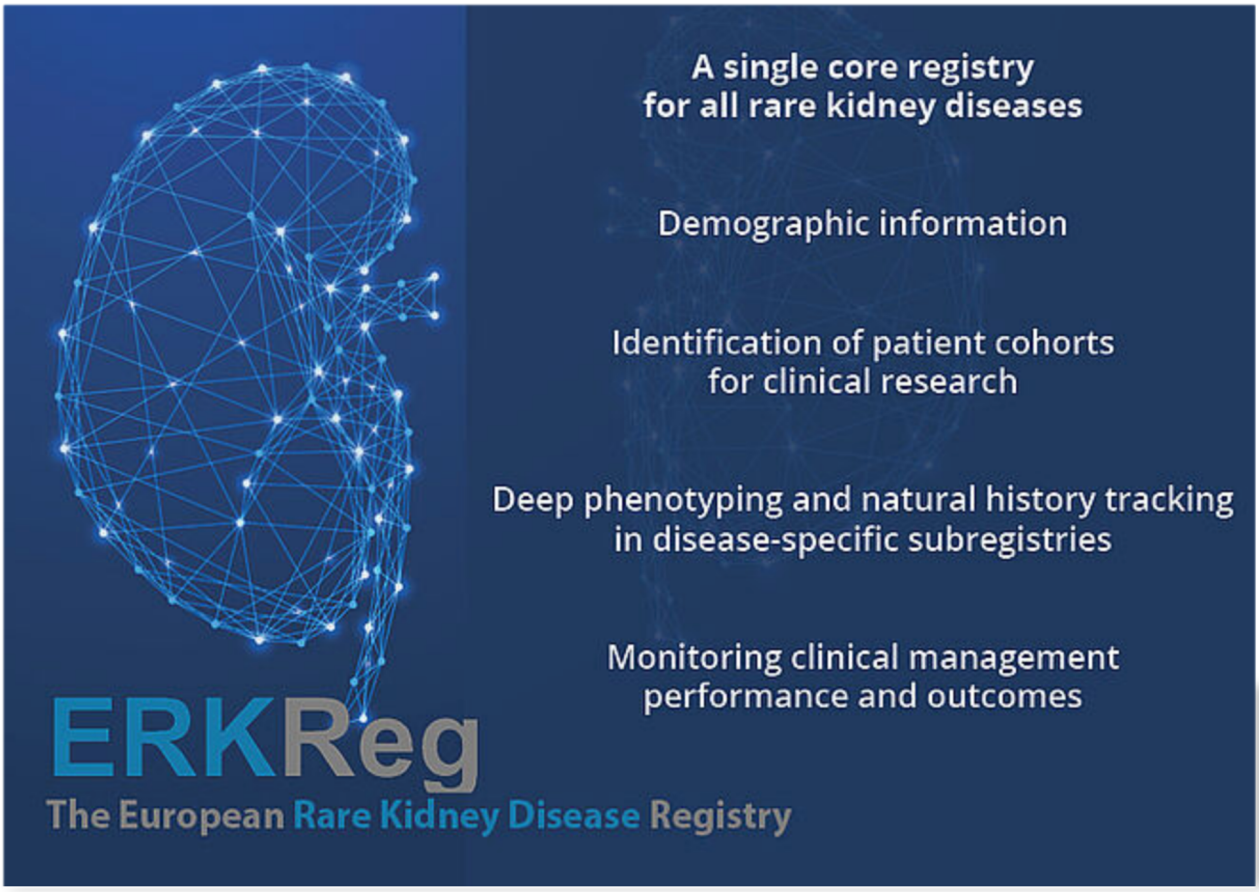
ERKReg The European Rare Kidney Disease Registry

4th Annual Registry Report

(Includes data up to 31st of December 2023)



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INTRODUCTION

We present the fourth Annual ERKReg Registry Report, which provides information on disease demographics, diagnostic assessments, and the quality of treatment performance in adults and children with rare kidney conditions through 31st December 2023.

In the fourth year of the registry, participation increased to 100 nephrology centres. The vast majority of patients (94.4%) were entered by 53 Healthcare Providers (HCPs) which are members of ERKNet, 1.6% were entered by 5 Affiliated ERKNet Partners, and 4.0% of the recorded patients have been entered by 42 non-ERKNet centres. The total number of patients entered until 31st December 2023 was 22,687. Currently, approximately 30% of all patients with rare kidney diseases that are in the care of our HCP members are enrolled in the Registry.

The registry underwent further technical refinements and detailed data quality management. Online plausibility checks ensure the entry of biologically and technically plausible values. Once per year, a more refined data quality check on more complex, logical errors is performed by our statistician. Queries based on the data consistency checks as well as update requests are sent out and answered by the participating sites.

The registry provides a performance and outcome monitoring service to the participating units. A set of key performance and outcome indicators (KPI) are continuously analyzed, and centre results are made available to the contributing sites both online in real-time and in annual written KPI reports.

ERKReg also serves as a platform to integrate disease-specific subregistries. To date, five subregistries have been implemented: the dRTA registry, the pediatric lupus nephritis registry, the Eurocys registry for cystinuria, the Italian registry for Alport, the subregistry for Childhood-onset SLE, and the subregistry for Bartter/Gitelman.

Visit us: www.registry.erknet.org

Contact us if you consider your participation in the registry: contact@erknet.org

Find the first ERKReg publication: https://pubmed.ncbi.nlm.nih.gov/34078418/

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Overarching objectives of the registry

- To inform how many patients with rare kidney diseases are treated across Europe
- To find and inform patients rapidly when novel therapeutic opportunities arise
- To help optimizing and monitor the quality of patient care
- To provide a platform for comprehensive sub-registries of rare kidney diseases for which more detailed knowledge is needed

Data collection

The registry prospectively records two datasets:

1. Common data set including JRC Common Data Elements (CDE), ensuring interoperability with other RD registries and essential Information relevant to all patients with rare kidney diseases.
2. Disease-specific dataset (please see section below on KPI monitoring) including basic demographic information and disease-specific key performance and outcome indicators, for which statistics and benchmarking features are provided.

Information on current treatment modalities and patient status is updated annually.

Continuous monitoring of center performance

- 62 disease-specific key performance and outcome indicators (KPIs) for rare kidney diseases were selected by ERKNet experts in a Delphi consensus finding process and subsequently implemented into the Registry Database.
- A statistics and benchmarking module provides feedback of relevant performance information to the centers. This statistic is continuously available to the centers via the Registry Website.
- The KPI monitoring functionality of the registry allows the participating European Reference Centers to review and compare their diagnostic and therapeutic performance as well as patient outcomes against the consortium average.

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RESULTS

Cumulative patient enrolment

By 31st December 2022, 17,079 patients with rare kidney diseases (6,425 by adult units (37,6%), 10,654 by pediatric units (62,4%) had been enrolled to ERKReg at 44 ERKNet Member centres (15,989 patients), 5 Affiliated Partner centres (318 patients), and 40 other external (including non-European) centres (772 patients).
In 2023, 5608 patients have been enrolled (2,503 by adult units (44,6%), 3104 by pediatric units (55,4%). By 31th December 2023 a total of 22,687 patient records were included (8928 by adult units (39,4 %), 13,758 by pediatric units (60,6 %), 53 ERKNet Member centres (21,419 patients), 5 Affiliated Partner centres (368 patients), and 42 other external centres (900 patients).

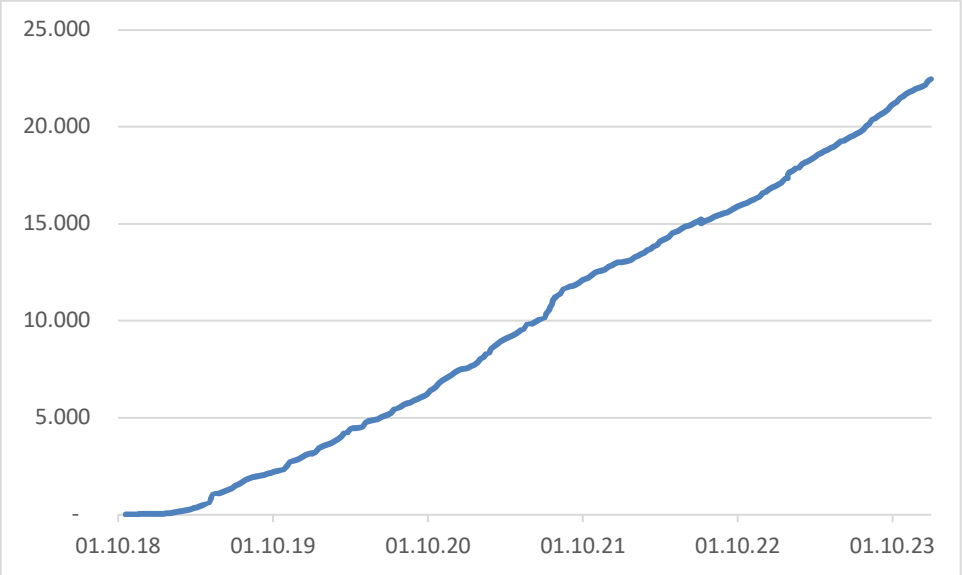

Figure 1. Cumulative enrolment curve

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100

Geographic distribution

Table 1. Number of patients enrolled per healthcare provider

Number of patients enrolled by 31st December 2023. Centres listed with at least 10 patients enrolled.

Healthcare Provider	Adult nephrology unit	Pediatric nephrology unit	Total
Paris, Necker-Enfants Malades University Hospital	1,044	1801	2,845
Turin, University of Torino-Ospedale HUB Torino Nord	1,317		1,317
Dublin, Beaumont Hospital	1,114		1,114
Leuven, University Hospitals Leuven	376	666	1,042
Lyon, Hôpital Femme Mère Enfant & Université de Lyon		1,034	1,034
Toulouse, Toulouse, University Hospital	347	663	1,010
Heidelberg, University Hospital		891	891
Rome, Bambino Gesù Children's Hospital		774	774
Genoa, Istituto Giannina Gaslini		683	683
Nijmegen, Radboud UMC	461	192	653
Vilnius, Vilnius University Hospital Santaros Klinikos	272	339	611
Naples, UOC Nephrology and Dialysis – Università della Campania	503	102	605
Madrid, Hospital Universitario 12 de Octubre	526	57	583
Barcelona, Hospital Universitari Vall d'Hebron		501	501
Milan, IRCCS Ca' Granda Ospedale Maggiore Policlinico		479	479
Bucharest, Fundeni Clinical Institute	281	198	479
Rome, Gemelli Hospital	451		451
Bergamo, Azienda Ospedaliera Papa Giovanni XXIII	434		434
Hamburg, University Medical Center Hamburg-Eppendorf		419	419
Gdansk, University Clinical Centre Gdansk	85	328	413
Ljubljana, University Medical Centre	45	334	379
Florence, Meyer Children's Hospital		376	
Amsterdam, Amsterdam, Academic Medical Center (AMC)	59	306	365
Brussels, Cliniques universitaires Saint-Luc - UCL	349		349
Padua, Azienda Ospedaliera di Padova	22	325	347
Hannover, Hannover Medical School	3	324	327
Turin, AOU Città della Salute e della Scienza di Torino		298	298
Vienna, Department of Pediatrics; Vienna General Hospital		289	289

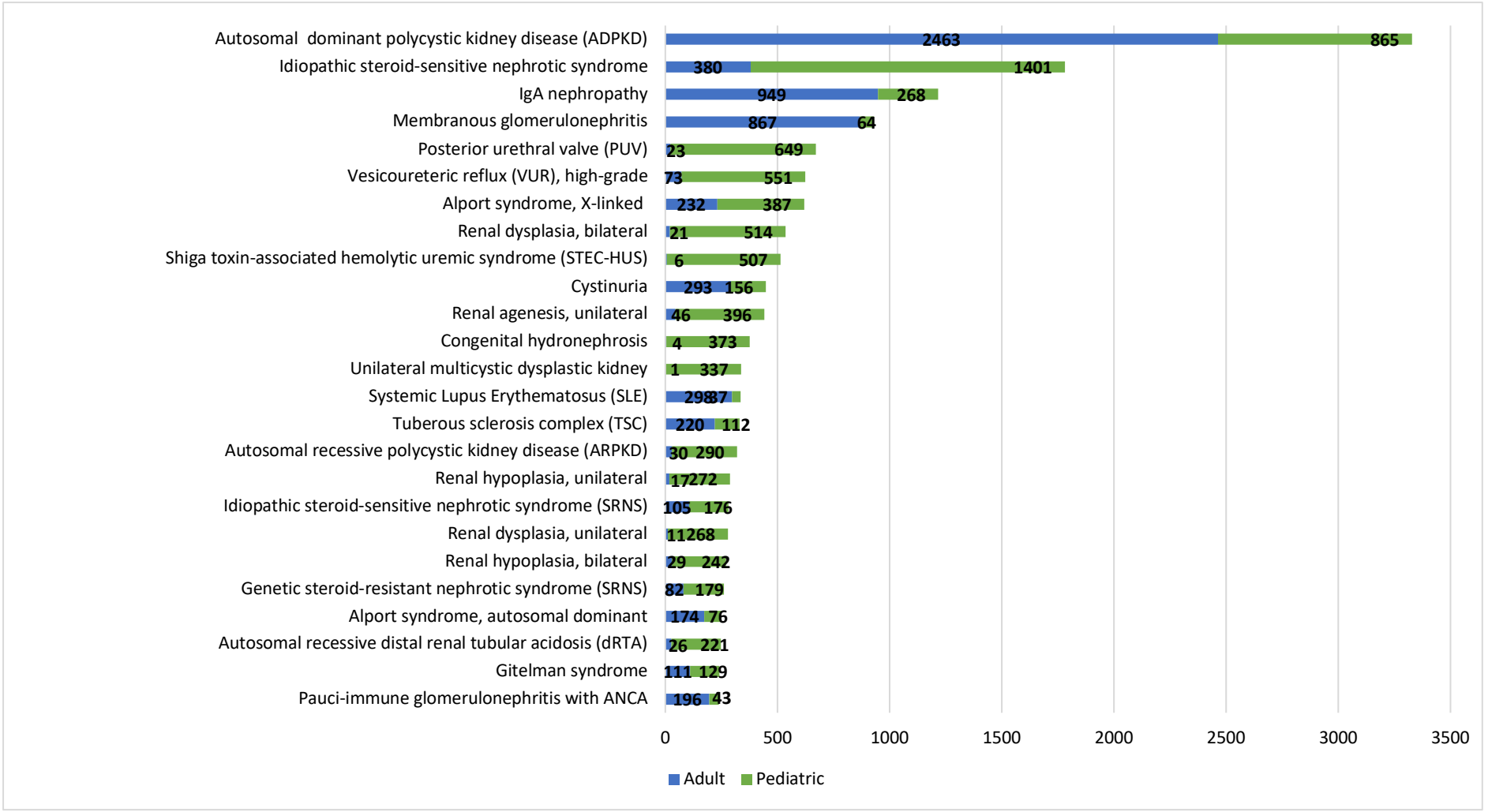
Dublin, Children`s Health Ireland (CHI) at Temple Street		281	281
Brescia, ASST Spedali Civili Brescia	265		265
Montpellier, University Hospital		258	258
Bordeaux, University Hospital		227	227
Athens, Laiko Generall Hospital of Athens	206		206
Essen, University Children`s Hospital		204	204
Münster, University Hospital		181	181
Tübingen, University Hospital		156	156
Maribor, University Medical Center		154	154
Bologna, AOU Policlinico S. Orsola Bologna	150		150
Barcelona, Hospital de la Santa Creu I Sant Pau and Fundacio Puigvert	143		143
Belgrade, University Children's Hospital		109	109
Manchester, Royal Manchester Childrens Hospital		109	109
Naples, A.O. Santobono-Pausilipon		100	100
Groningen, University Medical Center Groningen	100		100
Lodz, Instytut "Centrum Zdrowia Matki Polki"		79	79
Bari, AOU Consoziale policlinico di Bari – Opsedale Pediatrico Giovanni XXIII	75		75
Utrecht, University Medical Center	51	19	70
Cologne, University Hospital	27	37	64
Leipzig, University Hospital	57		57
London, Great Ormond Street Hospital		49	49
Ghent, UZ Ghent		40	40
Ankara, Hacettepe University Faculty of Medicine		36	36
Paris, Hopital La Pitié Salpetrière Paris	36		36
Porto, Centro Hospitalar Universitário do Porto	2	33	35
Riga, University Children Hospital		35	35
Tallinn, North Estonian Medical Center	30		30

Table 2. Number of patients per disease group

Number of patients enrolled by 31st December 2023. The classification adult/pediatric refers to current patient age.

Disease Group	Adult	Pediatric	Total
Immune Glomerulopathies	3,694	2,908	6,602
Congenital Malformations & Ciliopathies	455	3,921	4,376
Autosomal Dominant Structural Kidney Disorders	2,812	1,085	3,897
Tubulopathies	644	1,251	1,895
Heriditary Glomerulopathies	669	1,119	1,788
Obstructive Nephropathies	47	1,371	1,418
Thrombotic Microangiopathies	145	799	944
Metabolic and Stone Diseases	305	534	839
Rare Causes of Hypertension	10	169	179
Undefined	147	602	749

Table 3. Number of patients per diagnosis (total and per unit) – Most 25 common diagnoses (>100 patients enrolled)



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Kidney Function

Table 4. CKD stage distribution per disease group

Patients enrolled by 31st December 2023. The classification adult/pediatric refers to the patient age at enrolment.

CKD 1-5 = Chronic Kidney Disease stages, Pediatric patients (<18 years)

	CKD 1	CKD 2	CKD 3	CKD 4	CKD5	Total
Immune Glomerulopathies	**2,846**	**1,199**	**1,172**	**472**	**317**	**6,006**
Adult	607	854	1,090	452	270	3,273
Pediatric	2,239	345	82	20	47	2,733
Congenital Malformations & Ciliopathies	**1,678**	**1,253**	**750**	**228**	**243**	**4,152**
Adult	72	91	130	36	43	372
Pediatric	1,606	1,162	620	192	200	3,780
Autosomal Dominant Structural Kidney Disorders	**1,334**	**779**	**788**	**292**	**222**	**3,415**
Adult	497	629	755	288	215	2,384
Pediatric	837	150	33	4	7	1,031
Tubulopathies	**1,088**	**414**	**159**	**29**	**21**	**1,711**
Adult	218	176	103	16	17	530
Pediatric	870	238	56	13	4	1,181
Heriditary Glomerulopathies	**861**	**332**	**245**	**63**	**97**	**1,598**
Adult	174	140	145	45	39	543
Pediatric	687	192	100	18	58	1,055
Obstructive Nephropathies	**587**	**412**	**215**	**59**	**65**	**1,338**
Adult	4	12	7	4	2	29
Pediatric	583	400	208	55	63	1,309
Thrombotic Microangiopathies	**468**	**224**	**113**	**29**	**59**	**893**
Adult	14	32	39	15	21	121
Pediatric	454	192	74	14	38	772
Metabolic and Stone Diseases	**351**	**206**	**135**	**41**	**40**	**773**

Adult	92	72	57	18	7	246
Pediatric	259	134	78	23	22	527
Rare Causes of Hypertension	**143**	**20**	**6**	**2**	**-**	**171**
Adult	4	1	1	1	-	7
Pediatric	139	19	5	1	5	164

Figure 2. CKD stage 5 Treatment

1,227 patients in CKD stage 5 have been enrolled by 31st December 2023. The classification adult/pediatric refers to the patient age at enrolment.

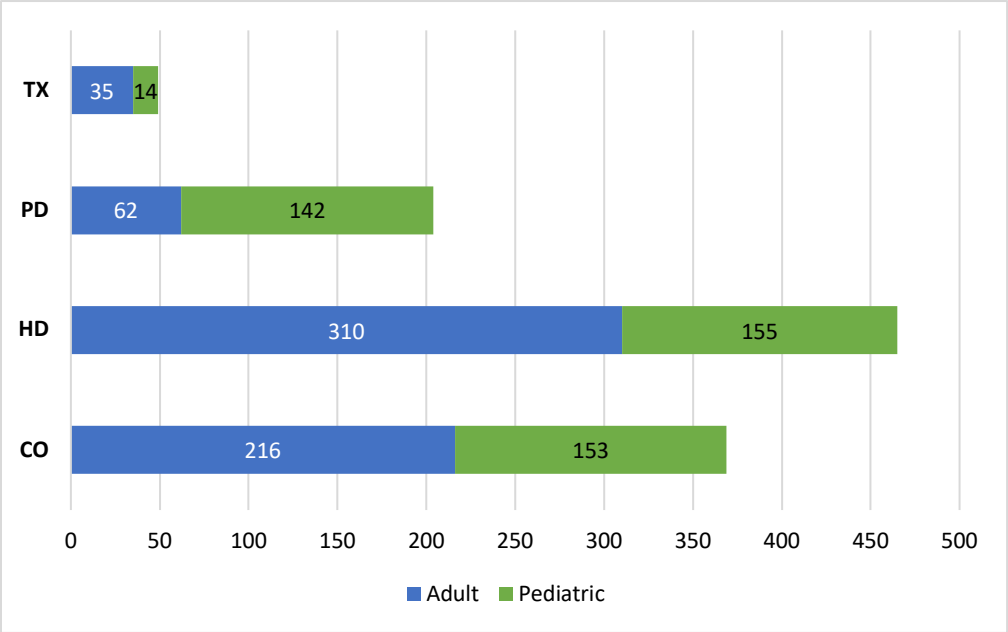

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Table 5. Age at diagnosis per disease group.

Disease Group	Median (IQR) years
Obstructive Nephropathies	0.0 (0.0-0.3)
Congenital Malformations & Ciliopathies	0.3 (0.0-5.1)
Rare Causes of Hypertension	3.3 (0.8-8.7)
Thrombotic Microangiopathies	3.3 (1.4-8.3)
Metabolic and Stone Diseases	4.7 (1.0-16.1)
Tubulopathies	6.4 (0.9-15.8)
Heriditary Glomerulopathies	9.0 (3.3-27.7)
Immune Glomerulopathies	20.0 (6.6-52.2)
Autosomal Dominant Structural Kidney Disorders	23.8 (9.4-39.7)

Table 6. Time from first symptoms to diagnosis.

Patients enrolled by 31st December 2023. The classification adult/pediatric refers to the patient age at diagnosis.

	Median (months)	IQR (months)
Thrombotic Microangiopathies	**0.8**	**0.5-1.1**
Adult	1.4	0.7-4.2
Pediatric	0.7	0.4-1.0
Obstructive Nephropathies	**1.2**	**0.5.-3.7**
Adult	0.7	0.1-2.8
Pediatric	1.2	0.5-3.7
Immune Glomerulopathies	**1.7**	**0.8-6.8**
Adult	3.3	1.2-12.6
Pediatric	1.1	0.5-3.3
Congenital Malformations & Ciliopathies	**1.8**	**0.6-6.9**
Adult	8.9	0.9-168.4
Pediatric	1.7	0.6-6.2
Autosomal Dominant Structural Kidney Disorders	**3.2**	**0.3-38.5**

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Adult	1.0	0.0-58.6
Pediatric	5.4	0.8-26.4
Tubulopathies	**4.2**	**1.0-23.9**
Adult	12.2	0.8-82.5
Pediatric	3.5	1.0-14.1
Metabolic Nephropathies	**7.4**	**1.0-24.5**
Adult	13.2	2.3-78.8
Pediatric	6.0	1.0-18.6
Heriditary Glomerulopathies	**11.1**	**1.8-49.6**
Adult	48.7	7.3-183.8
Pediatric	7.2	1.4-29.9