ERKReg

The European Rare Kidney Disease Registry

Project No. **777304**

ERKReg
The European Rare Kidney Disease Registry

Work package 04:
Deliverable Report D4.1

Registry Annual Report
(Data included until 31.12.2019)

Start date of project: 2018-01-01 Duration: 36 months

Organisation name of lead contractor for this deliverable:

Partner 01: UKL-HD

This project has received funding from the European Union´s Health Programme (2014-2020)		
Dissemination Level		
PU	Public	**X**
PP	Restricted to other programme participants (including the Commission Services)	
RE	Restricted to a group specified by the consortium (including the Commission Services)	
CO	Confidential, only for members of the consortium (including the Commission Services)	

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This report is part of the project '777304 / ERK-REG', which has received funding from the EU Health Programme (2014-2020).

INTRODUCTION

We present the first Annual ERKReg Registry Report, which provides information on disease demographics and the quality of treatment performance in children and adults with rare kidney conditions.

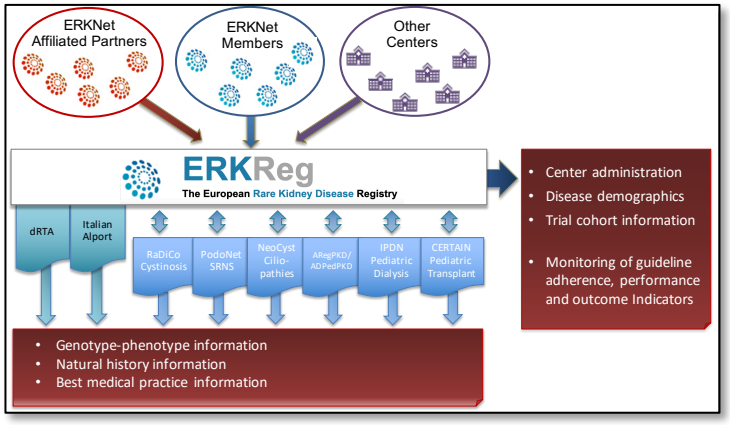
In January 2017, ERKNet received a 3-year grant from the EU Health Programme for Rare Disease Registries. The first year of the project was mainly devoted to establishing the Registry by selecting the variables to be captured, programming the database and the electronic case report forms, coping with the site-specific legal requirements, introducing the registry to the participating sites, training the local users and organizing efficient workflows at the site level.

In October 2018, the European Rare Kidney Disease Registry (ERKReg) was officially launched and the first ERKNet member units started data entry in the first half of 2019.

The modular structure of the Registry allows to integrate additional, more detailed disease-specific sub-registries. To date, the International distal renal tubular acidosis (dRTA) Registry and the Italian Alport Registry are accessible through the ERKReg registry.

In addition to serving the network's intrinsic needs, the registry contributes to the efforts to collate a large rare disease patient database on the European level. To that end, non-ERKNet centers are welcome to participate in the Registry.

A website for online data entry has been set up under www.registry.erknet.org. Please contact the ERKNet Central Office at contact@erknet.org if you consider your participation in the registry.



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Objectives

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

Data collection

- The registry records in a prospective way two datasets:
    1. Common data set including JRC minimum data set, ensuring interoperability withother RD Registries and essential Information relevant to all patients with rare kidney diseases.
    2. Disease-specific dataset (please see section below on KPI monitoring)
       basic demographic information and disease-specific key performance and outcome indicators, for which statistic and benchmarking features are provided.
- Information on current treatment modalities and patient status is collected by annual updates.

Continuous monitorong of center performance

- A set of 42 disease-specific key performance and outcome indicators (KPIs) for rare kidney diseases were selected by ERKNet members in an iterative Delphi consensus finding process and subsequently implemented into the Registry Database.
- A statistics and benchmarking module provides continuously feedback relevant performance information to the centers. This statistics is continuously available to the centers via the Registry Website.
- This accomplishment permits the participating European Reference Centers to review and compare their diagnostic and therapeutic performance as well as patient outcomes against the ERKNet average.

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Contributing Centers

Within the first 12 months since the launch of the registry, 71% of the ERKNet sites have started enrolling patients. A total of 3189 patients were enrolled by December 31, 2019.

Active ERKNet healthcare providers:	27/38 (71%)
Active Affiliated Partners:	3/11 (27%)
Active external centers:	7

ERKNet Reference Expert Centers
Barcelona, Hospital Universitari Vall d'Hebron
Bergamo, Azienda Ospedaliera Papa Giovanni XXIII
Cologne, University Hospital
Essen, University Children`s Hospital
Gdansk, University Clinical Centre Gdansk
Genova, Istituto Giannina Gaslini
Hamburg, University Medical Center Hamburg-Eppendorf
Hannover, Hannover Medical School
Heidelberg, University Hospital
Leuven, University Hospitals Leuven
London, Great Ormond Street Hospital
Lyon, Hôpital Femme Mère Enfant & Université de Lyon
Milano, IRCCS Ca' Granda Ospedale Maggiore Policlinico
Münster, University Hospital
Naples, A.O. Santobono-Pausilipon
Naples, UOC Nephrology and Dialysis - Università della Campania
Newcastle, Royal Victoria Infirmary
Nijmegen, Radboud UMC
Padova, Azienda Ospedaliera di Padova
Paris, Necker-Enfants Malades University Hospital
Prague, University Hospital Motol
Rome, Gemelli Hospital
Rome, Rome, Bambino Gesù Children's Hospital
Siena, University Hospital
Torino, San Giovanni Bosco Hospital and University Of Torino
Toulouse, Toulouse, University Hospital
Vilnius, Vilnius University Hospital Santaros Klinikos

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Affiliated Partners
Ljubljana, University Medical Centre
Maribor, University Medical Centre
Riga, University Children Hospital

External Centers
Ankara, Hacettepe University Faculty of Medicine
Barakaldo, Ped.Nephrology, Cruces University Hospital
Berlin, Charité - Universitätsmedizin Berlin
Moscow, Pirogov Russian Natl. Research Med. University
Oviedo, Asturias, Oviedo University Hospital
Palermo, ISMETT
Teheran, Iran University of Medical Sciences

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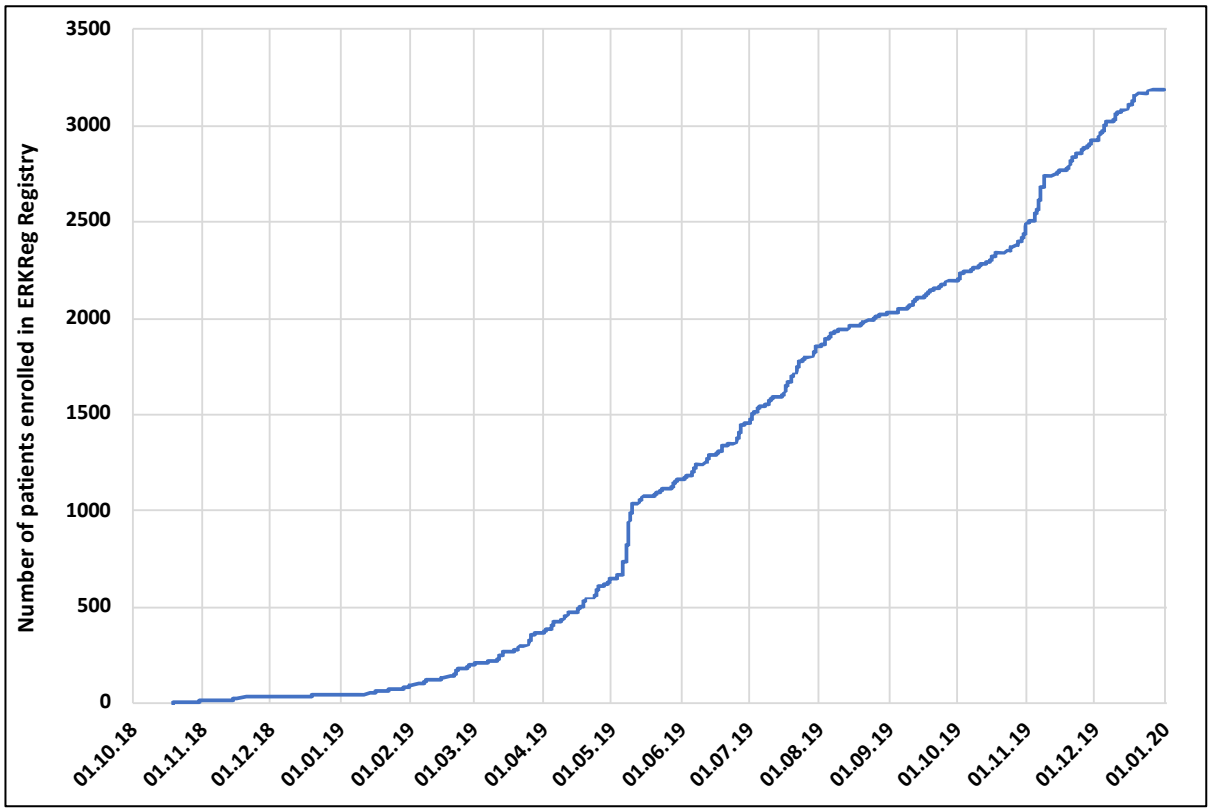
RESULTS

Cumulative patient enrollment

By 31st December 2019, 3189 patients with rare kidney diseases (30% adults, 70% children) had been enrolled to ERKReg at 27 ERKNet Member centers, 3 Affiliated Partner centers, and 7 other external (incl. non-European) centers.



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Number of patients per healthcare provider

Number of patients included in the Registry on 31st December 2019

Healthcare Provider	Pediatric	Adult	Total
Paris, Necker-Enfants Malades University Hospital	190	288	478
Torino, San Giovanni Bosco Hospital and University of Torino		372	372
Heidelberg, University Hospital	351		351
Milano, IRCCS Ca' Granda Ospedale Maggiore Policlinico	263		263
Barcelona, Hospital Universitari Vall d'Hebron	241		241
Rome, Rome, Bambino Gesù Children's Hospital	220		220
Naples, UOC Nephrology and Dialysis - Università della Campania	48	167	215
Hamburg, University Medical Center Hamburg-Eppendorf	145		145
Vilnius, Vilnius University Hospital Santaros Klinikos	102	40	142
Gdansk, University Clinical Centre Gdansk	113	2	115
Hannover, Hannover Medical School	86		86
Leuven, University Hospitals Leuven	67	5	72
Genova, Istituto Giannina Gaslini	71		71
Bergamo, Azienda Ospedaliera Papa Giovanni XXIII		62	62
Maribor, University Medical Centre	59		59
Essen, University Children`s Hospital	47		47
Padova, Azienda Ospedaliera di Padova	44	2	46
Münster, University Hospital	37		37
London, Great Ormond Street Hospital	32		32
Rome, Gemelli Hospital		23	23
Ljubljana, University Medical Centre	17		17
Ankara, Hacettepe University Faculty of Medicine	13		13
Naples, A.O. Santobono-Pausilipon	13		13
Cologne, University Hospital	9	1	10
Moscow, Pirogov Russian Natl. Research Med. University	8		8
Oviedo, Asturias, Oviedo University Hospital	8		8
Teheran, Iran University of Medical Sciences	7		7
Prague, University Hospital Motol	6		6
Riga, University Children Hospital	5		5
Toulouse, Toulouse, University Hospital	5		5
Lyon, Hôpital Femme Mère Enfant & Université de Lyon	4		4
Barakaldo, Ped.Nephrology, Cruces University Hospital	3		3
Berlin, Charité - Universitätsmedizin Berlin		3	3
Newcastle, Royal Victoria Infirmary	1		1
Nijmegen, Radboud UMC	1		1
Palermo, ISMETT	1		1
Siena, University Hospital	1		1

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Number of patients per country

Number of patients included in the Registry on 31st December 2019

Country	Pediatric	Adult	Total
Italy	661	626	1287
Germany	675	4	679
France	199	288	487
Spain	252		252
Lithuania	102	40	142
Poland	113	2	115
Slovenia	76		76
Belgium	67	5	72
United Kingdom	33		33
Turkey	13		13
Russia	8		8
Iran	7		7
Czech Republic	6		6
Estonia	5		5
The Netherlands	1		1

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Number of patients per disease group (total and per country)

Number of patients included in the Registry on 31st December 2019

Disease Group	Total	Pediatrics	Adults
Glomerulopathies	1234	707	527
CAKUT	655	622	33
Ciliopathies	572	311	261
Tubulopathies	320	257	63
Thrombotic Microangiopathies	163	146	17
Metabolic Nephropathies	152	107	45
Rare Causes of Hypertension	17	14	3

Disease Group	IT	DE	FR	ES	LT	PL	SLO	BE	UK	TR	RUS	IRN	CZ	EST	NL	Total
Glomerulopathies	611	220	206	64	54	51	16	11						1		1234
CAKUT	163	223	74	64	19	44	44	20	2					2		655
Ciliopathies	274	84	101	41	23	20	9	18						2		572
Tubulopathies	107	60	39	39	8		2	12	24	13	8	7			1	320
Thrombotic Microangiopathies	80	40	14	16	3		2	1	1				6			163
Metabolic Nephropathies	39	31	33	22	12		1	9	5							152
Rare Causes of Hypertension	1	4	4	4	1		1	1	1							17

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Number of patients per diagnosis (total and per country) – 30 most common diagnoses

Number of patients included in the Registry on 31st December 2019

Diagnosis	Total	IT	DE	FR	ES	PL	LT	SLO	BE	UK	TR	RUS	IRN	CZ	EST	NL
ADPKD	338	173	22	79	12	17	9	7	18						1	
Idiopathic SSNS	253	65	72	44	8	32	22	5	4						1	
IgA nephropathy	159	96	13	41	3		5	1								
Renal dysplasia, bilateral	114	29	63	2	13	2		2	3							
Membranous glomerulonephritis	112	99	3	9		1										
Posterior urethral valve	108	35	27	14	15	14		1	2							
Vesicoureteric reflux, high-grade	91	34	8	11	12	2	6	14	4							
Shiga toxin-associated hemolytic uremic syndrome (STEC-HUS)	86	52	25	2	5			2								
Alport syndrome, X-linked	86	31	13	27	7	1	3	4								
ARPKD	86	35	29	2	13	2	2	2							1	
Autosomal recessive distal renal tubular acidosis	61	6	2	6	13				1	9	13	7	4			
Idiopathic SRNS with sensitivity to second-line immunouppressive therapy	56	34	10	2	6	1	1	1	1							
Infantile nephropathic cystinosis	50	16	10	13	6				5							
Genetic steroid-resistant nephrotic syndrome	50	6	25	6	7	3	2		1							
Bartter syndrome	44	18	14	3	5				1	3						
Renal hypoplasia, bilateral	42	14	12	9		2	4								1	

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Congenital hydronephrosis	41	4	18			1	6	10	2								
Atypical hemolytic uremic syndrome with complement gene abnormality	40	17	6	5	4		2		1					5			
C3 glomerulonephritis	39	15	9	3	10			1	1								
Unilateral multicystic dysplastic kidney	37	7	12	2	3		1	8	4								
Bardet-Biedl syndrome	37	27	6	2	2												
Renal agenesis, unilateral	36	19	11	2			1	2	1								
Pauci-immune glomerulonephritis with ANCA	35	27		4	3	1											
Idiopathic multidrug-resistant nephrotic syndrome	35	14	6	10	3	2											
Nephrogenic diabetes insipidus	34	16	3	3	5				2	4							1
Juvenile nephronophthisis	32	21	6	3			2										
Bilateral multicystic dysplastic kidney	32	1	22	3	5				1								
Mixed connective tissue disease	32	32															
Idiopathic SSNS with secondary steroid resistance	31	19	5	1	1	2	3										
Pediatric systemic lupus erythematosus	31	20	3	6	2												
Gitelman Syndrome	30	17	7	1	1					4							

Abbreviations:
IT – Italy
DE – Germany
FR – France
ES – Spain
LT – Lithuania
PL – Poland
CZ – Czech Republic
EST – Estonia
SLO – Slovenia
BE – Belgium
UK – United Kingdom
TR – Turkey
RUS – Russia
IRN – Iran
NL – The Netherlands

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Age at disease manifestation / Age at diagnosis per disease group

Patients enrolled by 31st December 2019

Disease Group	Age at disease manifestation (years)		Age at diagnosis (years)	
	N	Median (IQR)	N	Median (IQR)
CAKUT	118	3.2 (0.3-9.7)	332	1.2 (0.2-7.9)
Metabolic nephropathies	63	1.2 (0.5-5.0)	145	2.5 (0.9-9.9)
Tubulopathies	144	5.4 (1.4-12.5)	300	3.8 (0.9-11.6)
Thrombotic microangiopathies	121	4.6 (1.7-8.6)	149	4.0 (1.6-8.4)
Rare causes of hypertension	10	1.9 (1.0-4.2)	17	5.0 (2.1-11.2)
Ciliopathies	261	14.8 (3.5-32.2)	542	13.3 (3.3-33.1)
Glomerulopathies	725	16.2 (6.3-49.4)	1167	13.3 (4.7-40.8)

N= number of patients, IQR=interquartile range

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CKD stage distribution per disease group

Patients enrolled by 31st December 2019

Disease Group	CKD 1	CKD 2	CKD 3	CKD 4	CKD5	Total
CAKUT	**630**	**212**	**196**	**71**	**68**	**1177**
Adults	120	111	151	59	37	478
Pediatrics	510	101	45	12	31	699
Glomerulopathies	**184**	**192**	**160**	**53**	**55**	**644**
Adults	3	7	14	5	2	31
Pediatrics	181	185	146	48	53	613
Ciliopathies	**191**	**147**	**118**	**46**	**49**	**551**
Adults	40	65	81	32	26	244
Pediatrics	151	82	37	14	23	307
Tubulopathies	**189**	**66**	**35**	**5**	**4**	**299**
Adults	19	20	13	3	2	57
Pediatrics	170	46	22	2	2	242
Thrombotic Microangiopathies	**78**	**34**	**24**	**10**	**11**	**157**
Adults	2	2	5	5	3	17
Pediatrics	76	32	19	5	8	140
Metabolic Nephropathies	**58**	**38**	**29**	**14**	**8**	**147**
Adults	14	10	12	6	2	44
Pediatrics	44	28	17	8	6	103
Rare Causes of Hypertension	**12**	**3**	**1**			**16**
Adults	1	1				2
Pediatrics	11	2	1			14

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

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Number of patients per diagnosis

Codes marked with an asterisk are internal ERKNet Code in the absence of an official Orphacode.

Glomerulopathies (OC 93548)			
Orphacode	**Diagnosis name**	**1st Diagnosis**	**2nd Diagnosis**
69061	Idiopathic steroid-sensitive nephrotic syndrome	253	2
9999982*	IgA nephropathy	159	3
97560	Membranous glomerulonephritis	112	
88917	Alport syndrome, X-linked	86	
567552	Idiopathic steroid-resistant nephrotic syndrome with sensitivity to second-line immunouppressive therapy	56	1
656	Genetic steroid-resistant nephrotic syndrome	50	3
329931	C3 glomerulonephritis	39	1
567550	Idiopathic multidrug-resistant nephrotic syndrome	35	1
97563	Pauci-immune glomerulonephritis with ANCA	35	
809	Mixed connective tissue disease	32	
567546	Idiopathic steroid-sensitive nephrotic syndrome with secondary steroid resistance	31	
93552	Pediatric systemic lupus erythematosus	31	
9999975*	Immune complex associated membranoproliferative glomerulonephritis, not otherwise specified	29	
9999981*	Lupus like glomerulopathy ('full house' nephropathy)	26	3
900	Shiga toxin-associated hemolytic uremic syndrome (STEC-HUS)	21	
88918	Alport syndrome, autosomal dominant	20	
536	Systemic Lupus Erythematosus	18	
761	Immunoglobulin A vasculitis (Henoch Schönlein nephritis)	17	1
839	Congenital nephrotic syndrome, Finnish type	16	
329903	Immunoglobulin-mediated mebranoproliferative glomerulonephritis	15	
220	Denys-Drash syndrome	13	1
727	Microscopic polyangiitis	13	
97564	Pauci-immune glomerulonephritis without ANCA	13	
9999977*	Collagenopathy, not further specified	12	
85445	AA amyloidosis	9	
93571	Dense deposit disease	9	
88919	Alport syndrome, autosomal recessive	8	1

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97556	Congenital nephrotic syndrome, no genetic cause specified	8	1
342	Familial Mediterranean fever	7	
85443	AL amyloidosis	7	
91138	Cryoglobulinemic vasculitis	6	
183	Eosinophilic granulomatosis with polyangiitis	5	
449395	IgG4-related kidney disease	5	
9999965*	Alport syndrome, digenic	5	
255249	Leigh syndrome with nephrotic syndrome	4	
9999976*	Microscopic (including familial) hematuria	4	1
182050	MYH9-related disease	3	
1830	Schimke immuno-osseous dysplasia	3	
9999972*	Nephrotic syndrome, syndromic, not otherwise specified	3	
280406	Nephrotic syndrome, familial, steroid-resistant, with sensorineural deafness	2	
347	Frasier syndrome	2	1
375	Anti-glomerular basement membrane disease	2	
85450	Hereditary amyloidosis with primary renal involement	2	
9999970*	C3 glomerulopathy secondary to MGRS	2	
2065	Galloway-Mowat syndrome	1	
314701	Primary systemic amyloidosis	1	
767	Polyarteritis nodosa	1	
797	Sarcoidosis	1	
84090	Fibronectin glomerulopathy	1	
9999971*	Proliferative glomerulonephritis with monoclonal immune deposits (PGNMID)	1	

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CAKUT (OC 93545)			
Orphacode	**Diagnosis name**	**1st Diagnosis**	**2nd Diagnosis**
93173	Renal dysplasia, bilateral	114	21
93110	Posterior urethral valve	108	27
9999969*	Vesicoureteric reflux, high-grade	91	25
97362	Renal hypoplasia, bilateral	42	10
2190	Congenital hydronephrosis	41	12
97363	Unilateral multicystic dysplastic kidney	37	11
93100	Renal agenesis, unilateral	36	14
97364	Bilateral multicystic dysplastic kidney	32	8
93172	Renal dysplasia, unilateral	22	14
9999968*	Neurogenic bladder, congenital or acquired	22	
97361	Renal hypoplasia, unilateral	18	9
238646	Congenital primary megaureter, obstructed form	11	4
2970	Prune belly syndrome	9	3
887	VACTERL/VATER association	7	
9999985*	Ureteropelvic junction obstruction (bilateral or in solitary kidney)	6	2
107	BOR (branchio-oto-renal) syndrome	5	1
289365	Vesicoureteric reflux, familial	5	1
93930	Bladder exstrophy	5	
238650	Congenital primary megaureter, refluxing form	4	
238654	Congenital primary megaureter, nonrefluxing and unobstructed form	4	
93111	RCAD (Renal cysts and diabetes) syndrome	4	
105	Atresia of urethra	3	1
1475	Renal coloboma syndrome	3	
567	Di George syndrome (22q11.2 deletion)	3	
97368	Drug-related renal tubular dysgenesis	3	
238637	Megacystis-megaureter syndrome	2	
2774	Multicentric carpo-tarsal osteolysis with or without nephropathy	2	
3027	Caudal regression sequence	2	1
52	Alagille syndrome	2	1
857	Townes-Brocks syndrome	2	
97369	Renal tubular dysgenesis of genetic origin	2	
110	Bardet-Biedl syndrome	1	
2237	HDR (Hypoparathyroidism-deafness-renal disease) syndrome	1	
2260	Oligomeganephronia	1	
2704	Ochoa syndrome	1	
435372	Anterior urethral valve	1	
881	Turner syndrome	1	
97367	Renal tubular dysgenesis due to twin-twin transfusion	1	
9999986*	Congenital primary megaureter, refluxing and obstructed	1	1

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Ciliopathies (OC 93587)			
Orphacode	**Diagnosis name**	**1st Diagnosis**	**2nd Diagnosis**
730	Autosomal dominant polycystic kidney disease	338	2
731	Autosomal recessive polycystic kidney disease	86	
110	Bardet-Biedl syndrome	37	
93592	Juvenile nephronophthisis	32	1
805	Tuberous sclerosis complex	22	
93591	Infantile nephronophthisis	17	1
93111	RCAD (Renal cysts and diabetes) syndrome	13	
220497	Joubert syndrome with renal defect	5	
2318	Joubert syndrome with oculorenal defect	5	
474	Jeune syndrome (ciliopathies)	5	
140969	Saldino-Mainzer (conorenal) syndrome	3	
64	Alström Syndrome	3	1
84081	Senior-Boichis syndrome	2	
88949	MUC1-related autosomal dominant tubulointerstitial kidney disease	2	
217330	Renin-related familial juvenile hyperuricemic nephropathy type 2	1	
88950	UMOD-related autosomal dominant tubulointerstitial kidney disease	1	

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Tubulopathies (OC 93603)			
Orphacode	**Diagnosis name**	**1st Diagnosis**	**2nd Diagnosis**
402041	Autosomal recessive distal renal tubular acidosis	61	
112	Bartter syndrome	44	1
223	Nephrogenic diabetes insipidus	34	
358	Gitelman syndrome	30	
214	Cystinuria	27	1
89936	X-linked hypophosphatemia	27	
2197	Hypercalciuria, idiopathic	16	
93608	Autosomal dominant distal renal tubular acidosis	13	
93612	Cystinuria type A	11	
3337	Fanconi syndrome, primary	10	
31043	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement	8	
91500	Tubulointerstitial nephritis and uveitis syndrome	8	
157215	Hereditary hypophosphatemic rickets with hypercalciuria	3	
89937	Autosomal dominant hypophosphatemic rickets	3	
93613	Cystinuria type B	3	
9999967*	Fanconi syndrome, induced by other drug	3	
171876	Generalized pseudohypoaldosteronism type 1	2	
2196	Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement (2196.0)	2	
244305	Hypophosphatemia, dominant, with nephrolithiasis or osteoporosis	2	
289176	Autosomal recessive hypophosphatemic rickets	2	
289390	Sjogren Syndrome	2	
30924	Primary hypomagnesemia with secondary hypocalcemia	2	
69076	Familial renal glucosuria	2	
199343	EAST syndrome	1	
240863	Fanconi syndrome, Cisplatin induced	1	
2697	Arthrogryposis-renal dysfunction-cholestasis syndrome	1	
34527	Familial primary hypomagnesemia with normocalciuria and normocalcemia	1	1
93610	Distal renal tubular acidosis with hemolytic anemia	1	

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Metabolic Nephropathies (OC 93593)			
Orphacode	**Diagnosis name**	**1st Diagnosis**	**2nd Diagnosis**
411629	Infantile nephropathic cystinosis	50	2
93598	Primary hyperoxaluria type 1	18	
324	Fabry disease	16	1
534	Lowe syndrome	15	
93622	Dent disease type 1 (CLCN5-related)	13	
300547	Autosomal recessive infantile hypercalcemia	9	
411634	Juvenile nephropathic cystinosis	7	
28	Methylmalonic acidemia, Vitamin B12-responsive	6	1
27	Methylmalonic acidemia, Vitamin B12-unresponsive	5	
470	Lysinuric protein intolerance	3	1
93623	Dent disease type 2 (OCRL-related)	3	
206428	Hypoxanthine-guanine phosphoribosyltransferase deficiency	2	
223713	Tubulopathy due to mitochondrial oxidative phosphorylation disorder	2	
1031	Amelogenesis imperfecta-nephrocalcinosis syndrome	1	
2088	Glycogen storage disease due to GLUT2 deficiency	1	
364	Glycogen storage disease due to glucose-6-phosphatase deficiency	1	

Thrombotic Microangiopathies (OC 93573)			
Orphacode	**Diagnosis name**	**1st Diagnosis**	**2nd Diagnosis**
90038	Shiga toxin-associated hemolytic uremic syndrome (STEC-HUS)	86	
544472	Atypical hemolytic uremic syndrome with complement gene abnormality	40	3
9999987*	Atypical hemolytic uremic syndrome, not further specified	21	
93581	Atypical hemolytic uremic syndrome with anti-factor H antibodies	7	
79282	Atypical hemolytic uremic syndrome due to Methylcobalamin deficiency, type CblC	3	
544493	Streptoccous pneumoniae-associated hemolytic uremic syndrome	2	
93583	Congenital thrombotic thrombocytopenic purpura	2	
93585	Acquired thrombotic thrombocytopenic purpura	2	

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Rare Causes of Hypertension (OC 93618)			
Orphacode	**Diagnosis name**	**1st Diagnosis**	**2nd Diagnosis**
97598	Renal artery stenosis, congenital	10	
526	Liddle Syndrome	2	
636	Neurofibromatosis type 1	2	
904	Williams syndrome	2	1
757	Pseudohypoaldosteronism type 2	1	

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KEY PERFORMANCE INDICATOR MONITORING

General	Pediatrics	Adults
Time (months) from 1st symptom to diagnosis: Median (IQR)	1.5 (0.6 - 7.4)	3.2 (0.7 - 25.4)
Time (months) from referral to diagnosis: Median (IQR)	0.2 (0.0 - 3.3)	0.2 (0.0 - 0.9)
% hereditary disease patients with any genetic screening	60.1 %	25.3 %
% hereditary disease patients with NGS screening (panel, WES)	30.1 %	13.9 %

Glomerulopathies	Pediatrics	Adults
% patients with hereditary glomerulopathies with genetic screening	67.0 %	40.4 %
% patients with multidrug resistant NS with comprehensive genetic screening	5.0 %	6.2 %
% normotensive patients (CKD1-3)	74.4 %	37.6 %
% patients with office blood pressure in target range	75.2 %	81.4 %
% patients with immunological glomerulopathies in clinical remission	32.0 %	15.0 %
% patients with persistent proteinuria who receive RAS antagonist therapy	62.0 %	80.3 %
% patients with hereditary and multidrug resistant NS prescribed RAS antagonist therapy	62.5 %	50.0 %
% adult patients with statin therapy	---	45.1 %
% adult patients with LDL cholesterol <100 mg/dl	---	56.6 %
% children with steroid sensitive idiopathic nephrotic syndrome who are obese	10.8 %	---
% children with steroid sensitive idiopathic nephrotic syndrome with height < 3rd percentile	6.2 %	---

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Tubulopathies / Metabolic Nephropathies	Pediatrics	Adults
% patients with hereditary nephropathy with genetic confirmation	59.5 %	40.0 %
% children (<16y) with normal length/height SDS	74.0 %	---
% patients with renal tubular acidosis maintaining normal serum bicarbonate	52.9 %	45.5 %
% cystinosis patients in whom at least one cystine blood level has been obtained during the past 12 months	85.2 %	100.0 %
% cystinuria patients free of new stones in past 12 months	80.0 %	50.0 %
% patients with stone forming diseases free of stones in past 12 months	80% %	51.4 %
% Fabry disease patients with at least one proteinuria measurement in past 12 months	50.0 %	85.7 %

Thrombotic Microangiopathies	Pediatrics	Adults
% aHUS patients with genetic/autoantibody screening (NGS, CFH autoantibodies)	40.6 %	35.3 %

Autosomal Dominant Structural Kidney Disorders	Pediatrics	Adults
% ADPKD patients with genetic screening	48.7 %	11.3 %
% ADPKD patients with at least one total kidney volume measurement by MRI	3.3 %	13.4 %
% normotensive ADPKD patients (CKD1-4) by office BP	81.7 %	29.2 %
% ADPKD patients screened for intracranial aneurysm when family history is positive	0.0 %	---
% TSC patients treated with mTOR inhibitors for indication AML>3 cm	0.0 %	---
% TSC patients treated with mTOR inhibitors for neurological indication (SEGA/Epilepsy)	0.0 %	---
% VHL patients with children or planning a pregnancy offered prenatal genetic counseling	0.0 %	---
% VHL patients with regular audiology assessment (every 2 years, starting at age 5)	0.0 %	---

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CAKUT/Ciliopathies/Obstructive uropathies	Pediatrics	Adults
% familial cystic disease patients with genetic screening	59.9 %	16.0 %
% patients with PUV detected by prenatal ultrasound	67.3 %	33.3 %
Mean (SD) of febrile UTIs in past 12 months in OUP / VUR patients	0.3 (0.9)	0.9 (1.2)

Pediatric Chronic Kidney Disease stage 3-5	
% children (<16y) with height > 3rd percentile	72.2 %
% children (1-16y) with height < 3rd percentile on growth hormone therapy	28.6 %
% children with BMI < 5th percentile	11.8 %
Children < 3 years with BMI < 5th percentile receiving enteral feeds	35.3 %
% patients with office systolic BP < 75th percentile	29.0 %
% patients with office systolic BP < 50th percentile	10.6 %
% patients with hemoglobin > 11 g/dl	72.8 %
Serum phosphorus in normal range for age	86.8 %
% patients with serum bicarbonate > 20 mmol/L	87.8 %

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Pediatric Dialysis	
% children (<16y) with height > 3rd percentile	48.8 %
% children (1-16y) with height < 3rd percentile on growth hormone therapy	33.3 %
% children with BMI < 5th percentile	10.4 %
Children < 3 years with BMI < 5th percentile receiving enteral feeds	66.7 %
Serum phosphorus in normal range for age	59.1 %
% patients with hemoglobin > 11 g/dl	47.0 %
% patients with serum bicarbonate > 20 mmol/L	86.1 %
PD peritonitis rate	0.4 episodes per year

Pediatric Transplantation	
% children with pre-emptive transplantation	30.3 %
% children (<16y) with height > 3rd percentile	74.1%
% children (1-16y) with height < 3rd percentile on growth hormone therapy	13.0%
% normotensive children (by office BP)	61.9%
% patients with hemoglobin > 10 g/dl	82.1%
% patients without severe metabolic acidosis	79.2%
% patients with biopsy proven rejection	2.0%

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