Supplementary Material: Targeted Exome Sequencing Provided Comprehensive Genetic Diagnosis of Congenital Anomalies of the Kidney and Urinary Tract

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S. Table 1. The 60 genes included in the gene panel design.

Gene	MIM gene ID	Inheritance	Renal phenotype	Reference
ACE	106180	AR	Renal tubular dysgenesis	[1]
ACVR2B	602730	Unknown	Renal agenesis and renal hypoplasia in mice	[2]
AGT	106150	AR	Renal tubular dysgenesis	[1]
AGTR1	106165	AR	Renal tubular dysgenesis	[1,3]
AGTR2	300034	AR	UPJO	[4]
BICC1	614295	AD	Cystic renal dysplasia	[5]
BMP4	112262	AD	Renal hypodysplasia	[6]
BMP7	112267	AD	Renal dysplasia	[7,8]
CDC5L	602868	AD	Multicystic renal dysplasia	[8,9]
CHD1L	613039	AD	Renal hypodysplasia, VUR, and UPJO	[8,10]
DLX5	600028	AR	Urethral malformation in mice	[11]
DLX6	600030	Unknown	Urethral malformation in mice	[11]
DSTYK	612666	AD	Renal hypodysplasia and UPJO	[12]
EMX2	600035	Unknown	Urinary tract anomalies in mice compound heterozygous for Pax2 and Emx2	[13]
EYA1	601653	AD	Branchio-oto-renal syndrome, multicystic dysplastic kidney, and renal hypoplasia	[14,15]
FGF20	605558	AR	Renal agenesis	[16]
FOXC1	601090	AD	Renal hypodysplasia	[17]
FRAS1	607830	AR	Fraser syndrome	[18,19]
FREM2	608945	AR	Fraser syndrome	[19,20]
GATA3	131320	AD	Hypoparathyroidism, sensorineural deafness, and renal disease (HDR) syndrome	[21]
GDF11	603936	Unknown	Candidate gene	[20]
GDNF	600837	AD	Renal agenesis	[22,23]
GFRA1	601496	AD	Renal agenesis	[23]
GREM1	603054	AR	Renal agenesis	[19]
HNF1B	189907	AD	Renal hypodysplasia, MCDK, and renal cyst	[24]
HOXC11	605559	Unknown	Renal agenesis and renal hypodysplasia in mice	[25]
HOXA11	142958	Unknown	Renal agenesis and renal hypodysplasia in mice	[25]
HOXD11	142986	Unknown	Renal agenesis and renal hypodysplasia in mice	[25]
KAL1	300836	XLR	Renal agenesis and Kallman syndrome	[26]
MMP-1	120353	AR	Obstructive uropathy	[27]
MMP-3	185250	Unknown	Obstructive uropathy	[27]
MMP-8	120355	Unknown	Obstructive uropathy	[27]
MUC1	158340	AD	Medullary cystic kidney disease type 1	[28]
NOTCH2	600275	AD	Alagille syndrome and Hajdu-Cheney syndrome with renal anomalies	[29,30]
PAX2	167409	AD	Renal coloboma syndrome and renal hypodysplasia	[31]
PKD1	601313	AD	Autosomal dominant polycystic kidney disease	[32]
PKD2	173910	AD	Autosomal dominant polycystic kidney disease	[32]

PKHD1	606702	AR	Autosomal recessive polycystic kidney disease	[33]
REN	179820	AR	Renal tubular dysgenesis	[1]
RET	164761	AD	Renal agenesis	[22]
ROBO2	602431	AD	VUR	[34,35]
SALL1	602218	AD	Renal hypodysplasia	[15]
SIX1	601205	AD	Branchio-oto-renal syndrome	[36]
SIX2	604994	AD	Renal hypodysplasia	[6]
SIX4	606342	Unknown	Candidate gene	[20]
SIX5	600963	AD	Branchio-oto-renal syndrome	[37]
SLIT2	603746	AD	Renal agenesis and MCDK	[38]
SOX17	610928	AD	VUR and UPJO	[39]
SPRY1	602465	Unknown	Candidate gene	[20]
TBX18	604613	AD	Urinary tract anomlies	[40]
TFAP2A	107580	AD	Branchio-oto-renal syndrome	[41]
TNXB	600985	AD	VUR	[42]
TRAP1	606219	AR	VUR, renal agenesis, and VACTERL association	[43]
TSC2	191092	AD	Cystic kidney disease in mice	[44]
UMOD	191845	AD	Medullary cystic kidney disease type 2 and hyperuricemic nephropathy	[45]
UPK3A	611559	AD	Renal adysplasia	[46]
USF2	600390	AD	Renal hypodysplasia	[47]
WNT4	603490	AD	Renal hypodysplasia	[48]
WT1	607102	AD	Diffuse mesangial sclerosis	[49]
XPNPEP3	613553	AR	Renal cysts and dysplasia	[50]

MIM, mendelian inheritance in man; AR, autosomal recessive; UPJO, ureteropelvic junction obstruction; AD, autosomal dominant; VUR,vesicoureteral reflux; MCDK, multicystic dysplastic kidney; XLR, X-linked recessive; VACTERL, vertebral defects, anal atresia, cardiac defects, trachea-esophageal fistula, renal anoamalies, and limb abnormalities.

S. Table 2. Single nucleotide variant prioritization and filtering workflow.

Step		Process	Variants (average)
1		Joint call variants	828,905
	1-1	Variants in coding region	18,398
	1-2	Not synonymous variants	12,078
	1-3	Rare variants (minor allele frequency < 1%) in 1000G, ExAC, and KRGDB	3,698
2		Sum of variants in each sample	5,285 (56.2)
	2-1	Variants in target 60 genes	620 (6.6)
	2-2	Qualified variants (Depth ≥ 5, VAF > 0.3, GQ ≥ 20)	358 (3.8)
3		Variant classification	
		Pathogenic	6
		Likely pathogenic	2
		Variants of uncertain significance	26
		Benign	324

1000G, 1000 Genomes Project; ExAC, Exome Aggregation Consortium; KRGDB, Korean Reference Genome Database; VAF, variant allele frequency; GQ, genotype quality.

S. Table 3. Variants of uncertain significance.

Patient ID	Gene Transcript number	Nucleotide	Amino acid	Segregation	Frequency		MutationTaster	Renal phenotype	Extra-renal phenotype	Renal function	Sanger seqeuncing
					KRGDB	ExAC					
3	*RET* NM_020975.6	c.1921G>A	p.Ala641Thr	NA	0.0014	0.00004	DC	Bilateral RHD, horseshoe kidney, and left VUR	Oligohydramnios	ESRD at 10.4 yr	Yes
8	*TNXB* NM_019105.8	c.9749C>T	p.Thr3250Met	NA	0	0.00006	DC	Left renal agenesis, right renal cysts, and right VUR	Testicular cystic mass and epidydimal cysts	CKD	No
8	*ROBO2* NM_002942.5	c.3553T>C	p.Trp1185Arg	NA	0	0	NA	Left renal agenesis, right renal cysts, and right VUR	Testicular cystic mass and epidydimal cysts	CKD	No
13	*TNXB* NM_019105.8	c.2090T>C	p.Leu697Pro	NA	0.0018	0.00006	DC	Right MCDK and left VUR	Imperforate anus, duodenal obstruction due to annular pancreas, and choledochal cyst	Normal at 9.8 yr	No
14	*RET* NM_020975.6	c.1618A>G	p.Arg540Gly	NA	0.0009	0.00004	DC	Bilateral RHD	Epilepsy, attention deficit hyperactivity disorder, and post-transplant diabetes mellitus	ESRD at 17.2 yr	No
16	*TNXB* NM_019105.8	c.1364G>A	p.Gly455Asp	NA	0.0009	0.00001	DC	Bilateral RHD	Birth asphyxia, nephrocalcinosis, hearing loss, periventricular leukomalasia, and cardiomyopathy	CKD	No
17	*RET* NM_020975.6	c.874G>A	p.Val292Met	Mother	0.0018	0.0006	DC	Right RHD	Hearing loss	CKD	Yes

20	*NOTCH2* NM_024408.4	c.5684G>A	p.Arg1895His	NA	0.0009	0.0001	DC	Left renal agenesis	Micropenis	CKD	Yes
22	*ROBO2* NM_002942.5	c.1435C>T	p.Arg479Trp	NA	0	0.00003	DC	Left RHD, right renal cysts, and bilateral VUR	Oligohydramnios, post-transplant diabetes mellitus	ESRD at 6.6 yr	No
28	*TNXB* NM_019105.8	c.2030A>G	p.Asp677Gly	NA	0.0045	0.0025	DC	Bilateral RHD and bilateral VUR	Preterm and oligohydroamnios	CKD	No
28	*NOTCH2* NM_024408.4	c.5557G>C	p.Asp1853His	NA	0	0.00002	DC	Bilateral RHD and bilateral VUR	Preterm and oligohydroamnios	CKD	No
30	*CHD1L* NM_004284.6	c.1841G>A	p.Arg614Gln	NA	0.0077	0.0001	DC	Bilateral RHD and renal cysts	Atrial septal defect, short stature, hearing loss, and hypothyroidim	ESRD at 2.9 yr	No
31	*CHD1L* NM_004284.6	c.2345T>C	p.Leu782Ser	NA	0.0036	0.00005	DC	Right renal agenesis and left hydronephrosis	Developmental delay, retroperitoneal pesudocyst, and hearing loss	CKD	No
41	*HNF1B* NM_000458.4	c.439C>G	p.Gln147Glu	NA	0	0	DC	Bilateral RHD and renal cysts	Patent ductus arteriosus, ureter stone, pancreas hypoplasia, common bile-duct dilatation, and hepatic cyst	CKD	Yes
55	*ROBO2* NM_002942.5	c.3585G>T	p.Gln1195His	NA	0	0.00001	NA	Bilateral VUR	No	CKD	Yes
58	*SLIT2* NM_004787.4	c.674G>A	p.Arg225His	NA	0.0005	0.0001	DC	Right MCDK and bilateral VUR	Parietal lobe epilepsy, multiple neuropathic pain, hypomagnesemia, and hyperuricemia	ESRD at 21.1 yr	No

63	*HNF1B* NM_000458.4	c.313G>A	p.Glu105Lys	NA	0.0014	0.0001	DC	Bilateral RHD and left VUR	No	ESRD at 20.1 yr	Yes
66	*SLIT2* NM_004787.4	c.1046C>T	p.Ser349Phe	NA	0.0005	0	DC	Bilateral VUR	No	CKD	No
73	*SIX2* NM_016932.5	c.707C>T	p.Pro236Leu	NA	0.0005	0.0002	DC	Bilateral RHD and bilateral VUR	Post-transplant diabetes mellitus	ESRD at 13.6 yr	No
73	*SLIT2* NM_004787.4	c.4488G>T	p.Arg1496Ser	NA	0.0014	0.00002	DC	Bilateral RHD and bilateral VUR	Post-transplant diabetes mellitus	ESRD at 13.6 yr	No
76	*TNXB* NM_019105.8	c.2030A>G	p.Asp677Gly	NA	0.0045	0.0025	DC	Right RHD and bilateral VUR	No	CKD	No
76	*NOTCH2* NM_024408.4	c.5557G>C	p.Asp1853His	NA	0	0.0002	DC	Right RHD and bilateral VUR	No	CKD	No
79	*DSTYK* NM_015375.3	c.1718T>C	p.Ile573Thr	NA	0	0	DC	Bilateral VUR	Complex febrile seizure and inguinal hernia	CKD	No
81	*ACE* NM_000789.4	c.2186G>A c.2803C>T	p.Arg729Gln p.Pro935Ser	NA	0.0009 0.0005	0.00003 0.00002	DC DC	Bilateral VUR	No	ESRD at 15.5 yr	Yes
86	*CHD1L* NM_004284.6	c.968A>T	p.Asp323Val	NA	0.0027	0.0001	DC	Right renal agenesis and left UVJO	Hematometra with upper vaginal obstruction	CKD	No

KRGDB, Korean Reference Genome Database; ExAC, Exome Aggregation Consortium; NA, not available; DC, disease causing; RHD, renal hypodysplasia; ESRD, end stage renal disease; CKD, chronic kidney disease; MCDK, multicystic dysplatic kidney; VUR, vesicoureteral refluex; UVJO, ureterovesical junction obstruction.

S. Table 4. Clinical presentation of study participants.

Patient ID	Sex	Age at enrollment (yr)	Right kidney	Left kidney	Extra-renal phenotype	Renal outcome	Parental sampling
1	F	21.1	Renal agenesis	Renal cortical cysts	No	ESRD at 20.8 yr	Father and mother
2	M	18.5	RHD	RHD	No	CKD stage 3 at 21.0 yr	ND
3	M	20.6	RHD	RHD and VUR	Oligohydroamnios	ESRD at 10.4 yr	NA

4	M	21.8	RHD	RHD	Preterm, oilgohydramnios, developmental delay, microcephaly, spastic quadriplesia, inguinal hernia, and post-transplant diabetes mellitus	ESRD at 6.8 yr	ND
5	F	19.4	RHD and UPJO	RHD and UPJO	No	ESRD at 18.7 yr	ND
6	F	19.3	No	Renal agenesis	No	Normal at 21.7 yr	ND
7	M	18.4	RHD	RHD	Morning glory discs, dilated cardiomyopathy, and hypothyrodism	ESRD at 10.4 yr	NA
8	M	17.0	Renal cortical cysts and VUR	Renal agenesis	Testicular cystic mass and epidydimal cysts	CKD stage 3 at 19.5 yr	NA
9	M	17.0	RHD	RHD and renal cortical cysts	No	ESRD at 16.9 yr	ND
10	M	17.3	No	RHD and VUR	Thyroid colloid cysts	CKD stage 2 at 20.1 yr	ND
11	M	20.0	RHD and renal cortical cysts	RHD and renal cortical cysts	Oligohydraminos	ESRD at 16.9 yr	ND
12	M	15.3	RHD	RHD	No	CKD stage 3 at 18.0 yr	ND
13	M	8.5	MCDK	VUR	Imperforate anus, duodenal obstruction due to annular pancreas, and choledochal cyst	Normal at 9.8 yr	NA
14	M	19.1	RHD	RHD	Epilepsy, attention deficit hyperactivity disorder, and post-transplant diabetes mellitus	ESRD at 17.2 yr	NA
15	F	14.5	RHD	RHD	Pre-auricular pit and hearing loss	ESRD at 5.4 yr	ND
16	M	16.3	RHD	RHD	Birth asphyxia, nephrocalcinosis, hearing loss, periventricular leukomalasia, and cardiomyopathy	CKD stage 5 at 17.3 yr	NA
17	F	17.6	RHD	No	Hearing loss	CKD stage 2 at 19.8 yr	Father and mother
18	M	0.8	RHD and VUR	RHD	Oligohydramnios	CKD stage 3 at 3.5 yr	NA
19	M	20.1	RHD	RHD	No	CKD stage 3 at 22.8 yr	NA
20	M	16.0	No	Renal agenesis	Micropenis	CKD stage 5 at 16.0 yr	NA
21	M	9.5	RHD	RHD	No	CKD stage 2 at 12.3 yr	NA

22	M	8.3	Renal cortical cysts and VUR	RHD and VUR	Oligohydramnios, post-transplant diabetes mellitus	ESRD at 6.6 yr	NA
23	M	9.1	RHD	RHD	Preterm, oilgohydramnios, and atrial septal defect	ESRD at 6.0 yr	NA
24	M	7.8	Renal agenesis	Duplicated ureter and hydronephrosis	Preterm, oilgohydramnios, tetralogy of Fallot, imperforate anus, anorectal malformation, patent urachus, strabismus, and inguinal hernia	CKD stage 2 at 10.2 yr	NA
25	F	13.7	RHD and renal cortical cysts	RHD and renal cortical cysts	Preterm and oligohydroamnios	ESRD at 7.4 yr	NA
26	M	6.1	RHD	RHD	Preterm and oligohydroamnios	CKD stage 3 at 8.9 yr	NA
28	M	5.9	RHD and VUR	RHD and VUR	Preterm and oligohydroamnios	CKD stage 3 at 8.7 yr	NA
29	M	5.3	RHD and renal cortical cysts	RHD and renal cortical cysts	Preterm, oliogohydroamnios, inguinal hernia, and developmental delay	ESRD at 1 mo	Father and mother
30	F	4.8	RHD and renal cortical cysts	RHD and renal cortical cysts	Atrial septal defect, short stature, hearing loss, and hypothyroidim	ESRD at 2.9 yr	NA
31	M	5.5	Renal agenesis	Hydronephrosis	Developmental delay, retroperitoneal pseudocyst, and hearing loss	CKD stage 3 at 8.3 yr	NA
32	M	8.3	RHD and VUR	VUR	No	CKD stage 2 at 11.2 yr	ND
33	M	15.9	RHD	Renal agenesis and VUR	No	CKD stage 5 at 18.7 yr	ND
34	M	12.2	RHD and renal cortical cysts	RHD and renal cortical cysts	Preterm, imperforate anus, hypospadias, and choledocal cyst	CKD 3 at 13.6 yr	ND
35	M	10.9	RHD	RHD	Preterm, oligohydroamios, branchial cleft cyst, lymphoma, developmental delay, autism, and inguinal hernia	ESRD at 12.1 yr	NA
36	M	4.6	RHD and renal cortical cysts	RHD and renal cortical cysts	Oligohydramnios, hereditary multiple exostoses and congenital ptosis	CKD stage 3 at 7.5 yr	ND
37	M	12.8	RHD and renal cortical cysts	RHD and renal cortical cysts	Preterm, oliogohydroamnios, attention deficit hyperactivity disorder	ESRD at 12.2 yr	ND
38	M	13.8	RHD and renal cortical cysts	RHD, renal cortical cysts, and VUR	Preterm, mental retardation, attention deficit hyperactivity disorder, hearing loss, and intermittent exotropia	CKD stage 3 at 16.6 yr	ND
39	F	22.1	RHD and renal cortical cysts	RHD and renal cortical cysts	No	ESRD at 13.4 yr	ND

40	M	11.2	RHD and renal cortical cysts	RHD and renal cortical cysts	Nail dysplasia	ESRD at 5.2 yr	ND
41	M	18.1	RHD and renal cortical cysts	RHD and renal cortical cysts	Patent ductus arteriosus, ureter stone, pancreas hypoplasia, common bile-duct dilatation, and hepatic cyst	CKD stage 5 at 20.3 yr	NA
42	M	9.8	RHD and renal cortical cysts	RHD and renal cortical cysts	Preterm, jeunal atresia, cerebral palsy (periventricular leukomalacia)	CKD stage 3 at 12.8 yr	ND
43	M	15.4	RHD and renal cortical cysts	RHD and renal cortical cysts	Hyperuricemia	CKD stage 3 at 15.8 yr	ND
44	M	24.5	RHD	Renal cortical cysts	Hyperuricemia	CKD stage 2 at 27 yr	ND
45	F	10.2	RHD and renal cortical cysts	RHD and renal cortical cysts	Preterm, oligohydraminios, hypothyroidism and Fanconi syndrome	CKD stage 5 at 13.2 yr	ND
46	M	23.2	RHD and renal cortical cysts	RHD	Thrombocytopenia	ESRD at 18.5 yr	ND
48	F	28.0	RHD	RHD	Hypothyroidism	ESRD at 27.9 yr	ND
49	M	1.6	VUR	RHD and VUR	Preterm and hypospadias	Normal at 4.4 yr	ND
50	M	26.1	Mid-ureteral obstruction	MCDK	Post-transplant diabetes mellitus	ESRD at 17.6 yr	ND
51	M	11.9	No	MCDK and ureterocele	Preterm, atrial septal defect, bilateral undescended testis, and omphalocele	Normal at 11.9 yr	ND
52	F	12.2	RHD and renal cortical cysts	MCDK	Preterm and oliogohydroamnios,	ESRD at 11 mo	ND
53	M	21.3	RHD	RHD and VUR	No	ESRD at 7.2 yr	ND
54	M	20.0	VUR	VUR	No	ESRD at 20.0 yr	ND
55	M	19.9	VUR	VUR	No	CKD stage 3 at 22.4 yr	NA
56	M	18.2	VUR	VUR	Intermittent exotropia	CKD stage 3 at 21.1 yr	ND
57	M	17.6	VUR	VUR	No	ESRD at 16.7 yr	ND
58	M	22.8	MCDK and VUR	VUR	Parietal lobe epilepsy, multiple neuropathic pain, hypomagnesemia, and hyperuricemia	ESRD at 21.1 yr	NA

59	M	16.8	VUR	VUR	No	CKD stage 5 at 19.8 yr	ND
60	M	17.3	RHD and VUR	RHD and VUR	No	ESRD at 17.0 yr	ND
61	M	19.3	VUR	VUR	Optic disc coloboma and strabismus	CKD stage 5 at 22.4 yr	ND
62	M	22.3	VUR	VUR	No	CKD stage 3 at 23.6 yr	ND
63	M	23.2	RHD	RHD and VUR	No	ESRD at 20.1 yr	NA
64	M	14.2	VUR	RHD and VUR	No	ESRD at 5.6 yr	ND
65	M	25.2	VUR	VUR	No	ESRD at 16.9 yr	ND
66	M	18.9	VUR	VUR	No	CKD stage 3 at 21.3 yr	NA
67	M	12.0	RHD and VUR	VUR	Atrial septal defect and epiblepharon	CKD stage 4 at 14.9 yr	ND
68	M	12.1	RHD and VUR	RHD and VUR	Oligohydraminos	ESRD at 8.9 yr	ND
69	M	21.6	RHD	RHD	No	ESRD at 20.0 yr	ND
70	M	21.2	RHD and VUR	VUR	Intellectual and developmental delay, nocturnal enuresis, and insomnia	CKD stage 4 at 21.2 yr	ND
71	M	9.0	VUR	VUR	Preterm	CKD stage 2 at 11.8 yr	ND
72	F	22.9	VUR	VUR	bilateral superior vena cava	ESRD at 21.6 yr	ND
73	M	12.9	RHD and VUR	RHD and VUR	Post-transplant diabetes mellitus	ESRD at 13.6 yr	NA
74	M	8.6	RHD	VUR	Preterm, umbilical hernia, inguinal hernia, and cryptorchidism	CKD stage 2 at 10.6 yr	ND
75	M	7.9	VUR	VUR	Preterm	CKD stage 4 at 10.8 yr	ND
76	M	19.7	RHD and VUR	VUR	No	CKD stage 2 at 19.7 yr	NA

77	M	11.9	RHD	RHD and VUR	No	CKD stage 4 at 11.9 yr	ND
78	M	4.9	RHD and VUR	VUR	No	ESRD at 4 mo	ND
79	M	15.5	VUR	VUR	Complex febrile seizure and inguinal hernia	CKD stage 5 at 18.2 yr	NA
80	M	9.3	RHD, renal cortical cysts, and VUR	RHD, renal cortical cysts, and VUR	Oligohydraminos, developmental delay and sensory neural hearing loss	ESRD at 2 mo	ND
81	M	15.8	VUR	VUR	No	ESRD at 15.5 yr	NA
82	M	15.5	RHD and renal cortical cysts	RHD and renal cortical cysts	Diabetes mellitus	Normal at 18.1 yr	ND
83	M	16.9	PUV	PUV	Hearing loss	CKD stage 3 at 19.8 yr	ND
84	M	5.3	PUV and RHD	PUV and RHD	Preterm and oliogohydroamnios	CKD stage 4 at 8.2 yr	ND
85	M	4.3	PUV and RHD	PUV and RHD	Preterm, oliogohydroamnios, hypothyrodism, inguinal hernia, hypertrophic cardiomyopathy, auricular benign cyst, and developmental delay	ESRD at 5 mo	ND
86	F	20.6	Renal agenesis	UVJO	Hematometra with upper vaginal obstruction	CKD stage 2 at 23.6 yr	NA
87	M	13.8	UPJO	UPJO	No	ESRD at 13.4 yr	ND
88	F	3.1	VUR	Renal agenesis	No	CKD stage 2 at 5.8 yr	ND
89	M	14.7	Hydronephrosis	MCDK	No	CKD stage 2 at 5.1 yr	ND
90	F	14.4	UPJO	MCDK	Partial bicornuate uterus or partial septated uterus	CKD stage 2 at 15.4 yr	ND
91	F	10.7	MCDK	Hydronephrosis	Preterm, oliogohydroamnios, imperforated anus, vaginal anomaly, and ambiguous genitalia	CKD stage 2 at 13.1 yr	ND
92	M	10.4	RHD and renal cortical cysts	MCDK	Oligohydraminos, choledochal cyst, and hypomagnesemia	CKD stage 2 at 13.0 yr	Father and mother
93	M	3.5	Renal cortical cysts	MCDK	Preterm and portal vein thrombosis	Normal at 3.5 yr	ND

94	M	15.6	RHD and renal cortical cysts	RHD and renal cortical cysts	Developmental delay, Atrial septal defect, cleft palate, cerebral palsy, congenital patellar dislocation, congenital blepharoptosis, and skeletal dysplasia	CKD sate 2 at 18.1 yr	ND
95	M	14.5	RHD and renal cortical cysts	RHD and renal cortical cysts	Optic disc anomaly, hemolytic anemia, and attention deficit hyperactivity disorder	ESRD at 3.2 yr	Father and mother
96	M	10.9	VUR and ureterocele	RHD	No	CKD stage 3 at 13.9 yr	ND

Yr, years; F, female; M, male; ESRD, end stage renal disease; RHD, renal hypodysplasia; CKD, chronic kidney disease; ND, not done; NA, not available; VUR, vesicoureteral reflux; UPJO, ureteropelvic junction obstruction; MCDK, multicystic dysplastic kidney; mo, monthrs; UVJO, ureterovesical junction obstruction; PUV, posterior urethral valve.

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