Supplemental Table 1. Panels (utilized in this study) and the genes included in each of them are summarized below.

Nephrotic Syndrome (NS)/Focal Segmental Glomerulosclerosis (FSGS) Panel	ACTN4, ANKFY1, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CDK20, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, DLC1, EMP2, FAT1, GAPVD1, GON7, INF2, ITGA3, ITGB4, ITSN1, ITSN2, KANK1, KANK2, KANK4, KAT2B, KIRREL1, LAGE3, LAMA5, LAMB2, LMX1B, MAFB, MAGI2, MYH9, MYO1E, NEU1, NFKB2, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP205, NUP93, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TNS2, TP53RK, TPRKB, TRIM8, TRPC6, TTC21B, WDR4, WDR73, WT1, XPO5, YRDC
Alport syndrome Panel	COL4A3, COL4A4, COL4A5, COL4A6
Autosomal Dominant Polycystic Kidney Disease (ADPKD) panel	DNAJB11, GANAB, HNF1B, PKD1, PKD2
Recessive Polycystic Kidney Disease (ARPKD) panel	DZIP1L, PKHD1
Hereditary cystic kidney disease panel	ANKS6, CEP164, CEP290, CEP83, COL4A1, CRB2, DCDC2, DICER1, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, RPGRIP1L, SDCCAG8, SEC61A1, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423
Nephrotic Syndrome	NPHS1, NPHS2, WT1, PLCE1, LAMB2
Autosomal Dominant and Recessive Polycystic Kidney Disease (ADPKD and ARPKD) Panel	DNAJB11, DZIP1L, GANAB, HNF1B, PKD1, PKD2, PKHD1
Distal Renal Tubular Acidosis Panel	ATP6V0A4, ATP6V1B1, CA2, SLC4A1
Atypical Hemolytic Uremic syndrome (s-HUS) panel	C3, CFB, CFH, CFHR1, CFHR3, CFHR5, CFI, DGKE, MCP, THBD

Tuberous Sclerosis Complex Panel	TSC1, TSC2
Microscopic Hematuria (custom panel)	ACTN4, ADCY10, APOL1, C1QA, C1QB, C1QC, C3, CASR, CD2AP, CFH, CFI, CLCN5, CLDN14, CLDN16, CLDN19, COL4A3, COL4A4, COL4A5, COL4A6, CYP24A1, FN1, INF2, MUC1, MYH9, MYO1E, NPHS2, OCRL, SEC61A1, SLC34A1, SPRY2, VHL
Neurohypophyseal Diabetes Insipidus and Nephrogenic Diabetes Insipidus Panel	AQP2, AVP, AVPR2
Custom Glycosuria panel	SLC5A2, SLC5A2
Nephrolithiasis and Nephrocalcinosis Panel	ADCY10, AGXT, APRT, ATP6V0A4, ATP6V1B1, CA2, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, FAM20A, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, OCRL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, VDR, XDH
Nephrolithiasis Panel	ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, CA2, CASR, CLCN5, CLDN16, CLDN19, CYP24A1, FAM20A, GPHN, GRHPR, HOGA1, HPRT1, KCNJ1, MOCOS, MOCS1, OCRL, PREPL, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC4A1, SLC7A9,SLC9A3R1, UMOD, VDR, XDH
Alagille syndrome	ABCB11, ABCB4, ABCC2, ABCG5, ABCG8, ACOX2, AKR1C4, AKR1D1, ALDOB, AMACR, ATP8B1, BAAT, CC2D2A, CFTR, CLDN1, CYP27A1, CYP7A1 CYP7B1, DCDC2, DGUOK, DHCR7, EHHADH, FAH, GNAS, GPBAR1, HNF1B, HSD17B4, HSD3B7, INVS, JAG1, KMT2D, LIPA, MKS1, MPV17, MYO5B, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PKD1L1, PKHD1, POLG, SCP2, SERPINA1, SLC10A1, SLC10A2, SLC25A13, SLC27A5, SLC51A, SLC51B, SMPD1, TALDO1,TJP2, TMEM216, TRMU, UGT1A1, UTP4, VIPAS39, VPS33B
Bartter syndrome panel	BSND, CASR, CLCNKA, CLCNKB, GNAII, KCNJ1, MAGED2, SLC12A1, SLC12A3
Wilms tumor	WT1
Periodic Paralysis Panel	CACNA1S, KCNJ1, RYR1, SCN4A

Total blue print panel : https://www.bcm.edu/research/medical-genetics-labs/test_detail.cfm?testcode=1390

Supplemental Table 2. "Other" indications for referral are summarized in this table

Name of disease	Number
Hypertension	2
CKD	2
Rhabdomyolysis	2
Diamond-Blackfan anemia	1
Nephronophthisis	3
Bilateral Wilms Tumor	1
Renal Tubular Acidosis	1
Bartter syndrome	2
Diabetes insipidus	2
Hypokalemic paralysis	1
Glycosuria	1
Hyperuricemia and nephropathy	1
Townes brock syndrome	1
Nephrocalcinosis	5
Macrocephaly and low muscle tone	1
Alagille syndrome	1
Gitelman syndrome	1
a-HUS	3
Micropenis	1
Mitochondrial	1
Bilateral renal angiomyolipoma	1
Hypophosphatemia	1
VACTERL	1
Hyperoxaluria	1
Hypomagnesemia	2
Hypocalcemia	1
Autism and renal artery stenosis	1
Joubert	2
	43

Supplemental Table 3. Detection rates of different tests among indications for referral

Cystic Kidney disease total of 49

	Total number	Positive	VUS	Negative
Panel	19	15	2	2
CMA	3	0	0	3
CMA/ES	13	4	7	2
ES	3	3	0	0
Not done	11 (3 asymptomatic patients with family history of ADPKD, 4 insurance denial, 1 patient not available, 3 patients not interested to pursue genetic testing)			

CAKUT total of 41

	Total Number	Positive	VUS	Negative
Panel	0	0	0	0
CMA	9	5	0	4
CMA/ES	22	8*	12	2
ES	1	1	0	0
Total Blueprint	1	0	1	0
Not done	8 (7 not interested, 1 insurance denial)			

*includes three partially diagnosed cases

Hematuria Total number of 38

	Total number	Positive	VUS	Negative
Panel	15	10	4	1
CMA	2	0	1	1
Panel and ES	1	1	0	0
CMA/ES	12	7	2	3
Panel and CMA and ES	1	1	0	0
ES	1	1	0	0
Not done	6 (3 not interested to pursue genetic testing, 3 insurance denial)			

Proteinuria total number of 21

	Total number	Positive	VUS	Negative
Panel	10	7	2	1
CMA	0	0	0	0
Panel and CMA	1	0	1	0
CMA/ES	8	5	2	1
CMA and Panel and Total Blueprint panel	1	0	1	0
Not done	1 (patient was not interested to pursue genetic testing)			

Other indications total number of 43

	Total number	Positive	VUS	Negative
Panel	11	5	3	3
CMA	4	2	0	2
CMA/ES	14	8	6	0
Panel and CMA, and ES	1	0	1	0
Total BluePrint	3	1	1	1
CMA and Total Blueprint	1	1	0	0
Panel and CMA and Total Blueprint	1	1	0	0
Not done	8 (4 not interested to pursue genetic testing, 4 due to insurance)			

Supplemental Table 4. Impact on management in patients with partial diagnosis

Patient Number	L1	L2	L3	L4	L5	Type of genetic testing (1,2,3,4,5)	Partial diagnosis	Gene/locus	SNV	Phenotype
RGC-0022	-	-	+	-	+	2,4	Hearing Loss	*GJB2*	NM_004004.5; c.35delG, (p.G12VfsX2) and c.416G>A, (p.S139N)	Proteinuria, PUV, bilateral hearing loss
RGC-0023	-	-	+	-	+	2,4	Cataract	*CHMP4B*	NM_176812.4; c.508_510delGAA, (p.E170del) (Het)	Cataract and proteinuria
RGC-0036	-	-	+	-	+	2,4	Sickle cell anemia	*HBB*	NM_000518.4; c.20A>T, (p.E7V)(Hom)	Sickle cell anemia and proteinuria
RGC-0106	-	-	-	+	+	2,4	DD	*NAA15*	NM_057175.3; c.439C>T (p.Q147X) (Het)	Syndromic CAKUT
RGC-0133	-	-	+	-	+	2,4	DD	*CHMP1A*	NM_002768.4; c.88 C>T (p.Q30X) (Hom)	Hyperoxaluria, DD, non-verbal, wheelchair bound

L1, impact on medical or surgical treatment; L2, change of medical diagnosis; 3, providing diagnostic certainty; 4, subsequent evaluation for other body system involvement; 5, cascade family member testing; CNV, copy number variant; DD, developmental delay; PUV, posterior urethral valve; SNV, single nucleotide variant. Type of testing; 1, panel; 2, CMA; 3, proband ES; 4, trio ES; 5, Total blueprint panel.

Supplement Table 5. Demographics, phenotype, genetic variants' information, clinician's comments and recommendations for patients without diagnostic result who were found to have variants of uncertain significance (VUS)

Patient ID	Gender	Age	Phenotype	Testing (1,2,3,4, 5)	Gene	Variant	gnomAD	CADD score	Clinician's comment	Recommendation
RGC-0007	M	4	Cystic kidney	3	*PKD1*	NM_001009944; c.971G>T,(p.R324L)(Het) (inherited from father)	0	13.07		Paternal kidney US recommended
RGC-0007	M	4	Cystic kidney	3	*TRPC6*	NM_004621; c.2116G>A, p.V706I (Het) (inherited from mother)	2/250230	28	Recessive disorder and only one heterozygous variant	
RGC-0007	M	4	Cystic kidney	3	*LAMB2*	NM_002292; c.5233G>A, (p.A1745T)(Het) (inherited from mother)	0	24.5	Recessive disorder and only one heterozygous variant	
RGC-0007	M	4	Cystic kidney	3	*MYO1E*	NM_004998; c.1615C>A, (p.L539M)(Het) (inherited from father)	0	16.21	Recessive disorder and only one heterozygous variant	
RGC-0007	M	4	Cystic kidney	3	*ITGA8*	NM_003638; c.1492A>G (p.M498V)(Het)(inherited from mother)	1/251076	0.876	Inherited from the affected mother but variant predicted benign and phenotype does not match with dysplastic kidney in the mother	
RGC-0011	M	2	Cystic kidney	3	*PKD1*	NM_001009944; c.8119G>A (p.V2707M) (Het) (inherited from father)	0	9.007		Paternal kidney US recommended
RGC-0011	M	2	Cystic kidney	3	*PKD1*	NM_001009944; c.4151C>T (p.T1384I) (Het) (inherited from father)	0	24.5		Paternal kidney ultrasound recommended
RGC-0011	M	2	Cystic kidney	3	*PKD1*	NM_001009944; c.3239C>A (p.P1080H) (Het) (inherited from mother)	2/214486	23.5		Maternal kidney US recommended

RGC-0012	F	16.8	Hematuria	2	*TRPC6* and *YAP1*	arr(hg19) 11q22.1 (101,450,649-102,064,511)x3	NA	NA	*TRPC6* might be disrupted therefore this VUS might have clinical consequences (clinical significance of a duplication of these or any genes in this region is not currently known. This region in its entirety is not known to vary in copy number in normal population	Annual UA and follow up with nephrology
RGC-0016	M	17.1	Kidney stone	3	*TRPC6*	NM_004621; c.1678G>A (p.A560T) (Het) (inherited from mother)	0	17.98	VUS inherited from unaffected mother	Follow up in 2 years for reanalysis of tES
RGC-0016	M	17.1	Kidney stone	3	*SLC4A4*	NM_001098484; c.149G>C (p.G50A)(Het)(inherited from mother)	390/249120	20.6	Recessive disorder and only one heterozygous variant	
RGC-0025	M	18	Proteinuria	3	*COL4A5*	NM_ 033380.1; c.2180C>G (p.P727R) (inherited from mother)	0	27.3	Phenotype does not match Alport and mother does not have microscopic hematuria	Follow up recommended
RGC-0027	F	16	CAKUT	3	*IL17RD*	NM_017563.3 ;c.8C>G (p.P3R) (Het)(inherited from father)	o	26.1	Phenotype does not fit with Kallmann syndrome but VUS could contribute to kidney anomaly	Testing of family members/paternal kidney ultrasound recommended
RGC-0040	F	3.4	CAKUT	3	*CHD7*	NM_017780; c.8378C>G (p.A2793G)(Het)(inherited from father)	4/244694	24	Phenotype does not fit	
RGC-0040	F	3.4	CAKUT	3	*PKD2*	NM_000297; c.2420G>A (p.R807Q)(Het)(inherited from father)	754/251136	26.1	Phenotype does not fit	
RGC-0040	F	3.4	CAKUT	3	*PKD1*	NM_001009944; c.6749C>T	0	23.5	Phenotype does not fit	

						(p.T2250M)(Het)(inherited from mother)				
RGC-0040	F	3.4	CAKUT	3	*MSR1*	arr 8p22(16032346-16073395)x1	NA	NA	Likely benign	
RGC-0040	F	3.4	CAKUT	3	*MSR1*	arr 8p22(15965430-16021863)x3	NA	NA	Likely benign, Inherited from mother	
RGC-0048	F	9.8	CAKUT	3	*LRP2*	NM_004525; c.2456G>A (p.R819H)(Het)(inherited from father)	27/251146	25.1	Patient does not have full phenotype of Donnai-Barrow Syndrome	Brain MRI was normal, urine beta 2 microglubin is high, RBP recommended
RGC-0048	F	9.8	CAKUT	3	*LRP2*	NM_004525; c.403G>A (p.D135N)(Het)(inherited from mother)	0	24		
RGC-0048	F	9.8	CAKUT	3	*PKD1*	NM_001009944; c.10325C>T (p.A3442V)(Het) (inherited from father)	1/244174	12.83	Phenotype does not fit	Paternal kidney US recommended
RGC-0048	F	9.8	CAKUT	3	*PKD1*	NM_001009944; c.5530G>C (p.G1844R)(Het)(inherited from father),	0	21.3	Phenotype does not fit	Paternal kidney US recommended
RGC-0048	F	9.8	CAKUT	3	*SLC7A9*	NM_014270; c.814G>A (p.V272M) (Het) (inherited from father)	0	13.93	Recessive disorder and only one heterozygous variant	
RGC-0048	F	9.8	CAKUT	3	*FRAS1*	NM_025074; c.6584A>G (p.E2195G)(Het)(inherited from father)	238/280120	22.4	Two *FRAS1* variants on the same chromosome	
RGC-0048	F	9.8	CAKUT	3	*FRAS1*	NM_025074;c.9553G>A (p.G3185R)(Het)(inherited from father)	197/279136	24.8		
RGC-0048	F	9.8	CAKUT	3	*NPHP1*	NM_000272; c.830G>A (p.R277Q)(Het) (inherited from mother)	173/282608	2.723	Recessive disorder and only one heterozygous variant	
RGC-0059	F	12	CAKUT	3	*CC2D2A*	NM_001080522; c.2597A>G, (p.N866S)(Het)(inherited from mother)	45/280162	16.74	Recessive disorder and only one heterozygous variant	

RGC-0059	F	12	CAKUT	3	*PKHD1*	NM_138694; c. 5750A>G, (p. Q1917R)(Het)(inherited from father)	0	32	Recessive disorder and only one heterozygous variant	
RGC-0059	F	12	CAKUT	3	*ITGA8*	NM_003638; c. 1336G>A, (p.V446I)(Het)(inherited from mother)	126/282842	2.758	Recessive disorder and only one heterozygous variant	
RGC-0064	M	12	CAKUT	3	*NID1*	NM_002508.2; c.3680dupC (p.G1228RfsX9)(Het)(inherited from father)	0	6.445	VUS seems likely pathogenic, affected sibling positive	Brain MRI, testing siblings
RGC-0064	M	12	CAKUT	3	*NID1*	NM_002508.2; c.1297C>T (p.R433X)(Het)(inherited from mother)	0	12.17	VUS seems likely pathogenic, affected sibling positive	Brain MRI, testing siblings
RGC-0065	F	1	Other (Nephromegaly and Nephrocalcinosis)	3	*CRB2*	NM_173689.5; c.1298C>T (p.P433L)(Het)	124/282294	15.97	Recessive disorder and only one heterozygous variant	
RGC-0065	F	1	Other (Nephromegaly and Nephrocalcinosis)	3	*LIG4*	NM_002312.3; c.686A>G (p.H229R)(Het)	178/281618	23.3	Recessive disorder and only one heterozygous variant	
RGC-0065	F	1	Other(Nephromegaly and Nephrocalcinosis)	3	*PSAT1*	NM_058179.2; c.94T>C (p.Y32H)(Het)	5/280748	25.2	Recessive disorder and only one heterozygous variant	
RGC-0065	F	1	Other(Nephromegaly and Nephrocalcinosis)	3	*COL4A5*	NM_000495.4; c.4450T>C (p.Y1484H)(Het)	0	25.4	Unrelated to patient's phenotype	
RGC-0065	F	1	Other(Nephromegaly and Nephrocalcinosis)	3	*SH3YL1*	Arr 2p25.3 (66097-239712)x1	NA	NA	Non disease-associated regions	No parental follow-up recommended
RGC-0065	F	1	Other(Nephromegaly and Nephrocalcinosis)	3	*NUP62CL*	Arr Xp22.3 (106384817-106398144)x1	NA	NA	Non disease-associated regions	No parental follow-up recommended
RGC-0069	M	2.3	Proteinuria	1,2	*CD2AP*	NM_012120.2; c. 1286_1288dup (p.E429dup)(Het)	126/250158	3.562	In-frame duplication	tES recommended
RGC-0094	F	3.5	CAKUT	3	*COL4A5*	NM_000495.4; c.2600T>C (p.I867T)(Het)	2/182030	16.49	Unrelated to patient's phenotype	Reanalysis of ES

RGC-0095	M	9	Cystic kidney	1	*PKD1*	NM001009944.2; c.8498C>A (p.P2833H) (Het)	0	6.179	Conserved codon, seems likely pathogenic	Testing affected mother
RGC-0096	M	10.7	Cystic Kidney	3	*PKD1*	NM_001009944.2; c.10810 G>A (p.E3604K) (Het)(inherited from father)	0	22.9	Seems likely pathogenic based on PKDB and Clinvar	Paternal kidney US recommended
RGC-0096	M	10.7	Cystic Kidney	3	Portion of *PLEKHA7*, *ABCC8* and 6 other genes	arr[GRCh37] 11p15.1(16957123_17439236)x3(inherited from father)	NA	NA		
RGC-0096	M	10.7	Cystic Kidney	3	Portion of *VCX3A*, entire *HDHD1*, *STS*, *VCX*, and *PNPLA4*	arr[GRCh37] Xp22.31(6453036_8131810)x2 (inherited from mother)	NA	NA	Patient affected by Arthrogryposis and *VCX3A* is disrupted	Testing of siblings
RGC-0099	M	1.5	CAKUT	3		arr[GRCh37] (X)x1~2,(Y)x1 (Mosaic gain)(155186Kb) associated with mosaic Klinefelter syndrome	NA	NA	20-30% mosaicism	Follow up
RGC-0103	M	0.01	CAKUT	3	*FAT1*	NM_005245.3; c.7014C>A (p.S2338R)(Het)(inherited from mother)	7/280606	4.937		Follow up
RGC-0103	M	0.01	CAKUT	3	*FAT1*	NM_005245.3; c.9320G>T (p.C3107F)(Het)(inherited from mother)	5/249052	24.9		
RGC-0103	M	0.01	CAKUT	3	*PPEF1*	arr [GRCh19] Xp22.13 (18821936-18822035)x0	NA	NA	No disease association	
RGC-0104	F	14.8	Proteinuria	3	*MYO1E*	NM_004998.3; c.2627C>G (p.T876R)(Het)(inherited from mother)	400/282482	20.8	Recessive disorder and only one heterozygous variant	
RGC-0104	F	14.8	Proteinuria	3	*PLCE1*	NM_016341.3; c.2032A>G (p.M678V)(Het)(inherited from father)	280/280870	23.6	Recessive disorder and only one heterozygous variant	
RGC-0107	F	5.3	CAKUT	3	*BICC1*	NM_001080512.2; c.707A>G	8/282402	3.304	Strong evidence at gene level	Paternal kidney US recommended

						(p.N236S)(Het)(inherited from father)				
RGC-0109	M	15.6	Other (Hyperuricemic and nephropathy)	5	*WT1*	NM_024426.4; c.358G>A (p.G120S)(Het)	0	22.4	No phenotype overlap	
RGC-0109	M	15.6	Other	5	*WDR19*	NM_025132.3; c.*7C>T (Het)	54/275970	0.436	Recessive disorder and only one heterozygous variant	
RGC-0109	M	15.6	Other	5	*PKHD1*	NM_138694.3; c.2744C>T (p.A915V)(Het)	0	26.3	No phenotype overlap (kidneys not enlarged)	
RGC-0109	M	15.6	Other	5	*PKHD1*	NM_138694.3; c.7675G>C (p.V2559L) (Het)(inherited from mother)	280/282300	9.628	Predicted benign	
RGC-0111	F	0.01	CAKUT	3	*COL4A4*	NM_000092.4; c.3394C>G (p.P1132A)(Het)(inherited from father)	1/248216	25.4	No phenotype overlap	
RGC-0111	F	0.01	CAKUT	3	*LAMB2*	NM_002292.3; c.4224+19G>C(Het) (inherited from father	3031/279460	4.844	Recessive disorder and only one heterozygous variant	
RGC-0122	M	4	Hematuria	3	*COL4A3*	NM_000091.4; c.1483C>T (p.H495Y)(Het)(inherited from father)	202/280910	0.689	Kidney biopsy showed TBM	Testing other siblings
RGC-0122	M	4	Hematuria	3	*FN1*	NM_212482.1; c.3626C>T (p.T1209I)(Het)(inherited from mother)	2/251442	24.4	Biopsy does not fit	
RGC-0125	F	14	Other(a-HUS)	1,3	*THBD*	NM_000361; c.1456G>T (p.D486Y)(Het)	2115/276574	1.136		Reanalyze ES
RGC-0125	F	14	Other(a-HUS)	1,3	*DGKE*	NM_003647; c.303G>C (p.K101N)(Het)	39/282288	22.9	Recessive disorder and only one heterozygous variant	
RGC-0125	F	14	Other(a-HUS)	1,3	*GANAB*	NM_198335.2; c.1652A>C (p.N551T)(Het)(inherited from father)	0	22.3	No family history on paternal side, and positive family history from maternal side	

RGC-0125	F	14	Other(a-HUS)	1,3	*ALMS1*	NM_015120.4; c.9463A>T (p.T3155S)(Het)(inherited from father)	65/249098	24.7	Recessive disorder and only one heterozygous variant	
RGC-0125	F	14	Other(a-HUS)	1,3	*TRIOBP*	NM_0010391412; c.6632A>T (p.Q2211L)(Het)(inherited from father)	40/280310	28.2	Recessive disorder and only one heterozygous variant	
RGC-0125	F	14	Other(a-HUS)	1,3	*GRHPR*	NM_012203.1; c.374G>A (p.R125Q)(Het)(inherited from father)	78/282832	29.7	Recessive disorder and only one heterozygous variant	
RGC-0125	F	14	Other(a-HUS)	1,3	*DGKE*	NM_003647.2; c.303G>C (p.K101N)(Het)(inherited from father)	39/282288	23.2	Recessive disorder and only one heterozygous variant	
RGC-0126	M	17	Proteinuria	1,2,5	*CFH*	NM_000186.3; c.2270A>C (p.N757T)(Het)	0	0.112	No phenotype overlap	
RGC-0126	M	17	Proteinuria	1,2,5	*CD2AP*	NM_012120.2; c.164A>C (p.K55T)(Het)	63/282798	30	Fits with biopsy report and family history	
RGC-0126	M	17	Proteinuria	1,2,5	*NPHS2*	NM_014625; c.725C>T (p.A242V)(Het)	1962/281850	25.3	Recessive disorder and only one heterozygous variant	
RGC-0126	M	17	Proteinuria	1,2,5	*LAMB2*	NM_004646; c.2740G>A (p.G914A)(Het)	0	27.7	Although patient has two variants in *LAMB2*, Family history of proteinuria in this patient suggest AD mode of inheritance	Parental testing for KFM in *LAMB2*
RGC-0126	M	17	Proteinuria	1,2,5	*LAMB2*	NM_004646; c.1193C>T (p.T398I)(Het)	694/282716	7.968		Parental testing for KFM in *LAMB2*
RGC-0130	M	20	Cystic kidney	3	*SEC61A1*	NM_013336.3; c.554 C>G (p.T185S)(Het)(de novo)	0	28.4	Patient's phenotype has overlap with reported phenotype associate with this gene	
RGC-0136	F	7	CAKUT	3	*MT-RNR2*	m.2872C>T(Homoplasmic)(inherited from mother)	0	NA	Mother also homoplasmic suggesting that this	

									variant is more likely to be benign	
RGC-0138	F	16	CAKUT	5	*GJB3*	NM_024009.2; c.223C>T (p.R75C)(Het)(inherited from mother),	42/282762	29.7	Mother does not have hearing loss	
RGC-0138	F	16	CAKUT	5	*WFS1*	NM_006005.3; c.527T>C (p.V176A)(Het)(inherited from mother)	3/250700	22.4	Mother does not have hearing loss	
RGC-0138	F	16	CAKUT	5	*GATA3*	NM_0010022951; c.826C>T (p.R276W)(Het)	0	32	Patient's phenotype has overlap with reported phenotype associate with this gene, patient has hypoparathyroidism	KFM testing of other family members
RGC-0138	F	16	CAKUT	5	*NPHS1*	NM_004646.3; c.7C>A (p.L3M)(Het)	8/185830	4.560	Recessive disorder and only one heterozygous variant	
RGC-0139	M	15	Cystic kidney	3	*PRKD1*	NM_002742.2; c.1947T>G (p.F649L)(Het)(inherited from mother)	2/250352	11.83	Mother reported to have heart disease	
RGC-0139	M	15	Cystic kidney	3	*PKD1*	NM_001009944.2 ; c.7061A>C (p.Q2354P)(Het)(inherited from father)	0	27.1	Likely the cause of ADPKD, there is history of ADPKD in father	Testing of siblings for specific variant in *PKD1*
RGC-0139	M	15	Cystic kidney	3	*PKD1*	NM_001009944.2 ; c.6097G>A (p.A2033T)(Het)(inherited from father)	11/275498	23.2	there is history of ADPKD in father	Testing of siblings for specific variant in *PKD1*
RGC-0140	F	8	CAKUT	3	*KIAA1109*	NM_015312.3; c.822-3T>C (Het)(inherited from father)	2/247734	6.412	Recessive disorder and only one heterozygous variant	
RGC-0140	F	8	CAKUT	3	*COL4A4*	NM_000092.4 ; c.2985C>T (p.P995=)(Het)(inherited from father)	19/280864	0.112		
RGC-0140	F	8	CAKUT	3	*FANCC*	NM_000136.2; c.998T>C (p.L333P)(Het)(inherited from father)	2/249760	22.4	Recessive disorder and only one heterozygous variant	

RGC-0140	F	8	CAKUT	3	*AHI1*	NM_017651.4; c.1621G>T (p.D541Y)(Het)(inherited from mother)	0	24.2	Recessive disorder and only one heterozygous variant	
RGC-0140	F	8	CAKUT	3	*TMTC3*	NM_181783.3; c.10A>G (p.I4V)(Het)(inherited from father)	13/276640	12.90	Recessive disorder and only one heterozygous variant	
RGC-0140	F	8	CAKUT	3	*CFH*	NM_000186.3; c.506A>G (p.H169R)(Het)(inherited from father)	3/251074	0.014	No phenotype overlap	
RGC-0141	F	0.9	Other (Hypocalcemia)	3	*TRPC6*	NM_004621.5; c.101T>C (p.M34T) (Het)(inherited from father)	2/199782	24.5	Father does not have kidney disease	
RGC-0141	F	0.9	Other (Hypocalcemia)	3	*SLC12A1*	NM_000338.2; c.2282G>A (p.R761Q)(Het)(inherited from mother)	26/282292	22.7	Recessive disorder and only one heterozygous variant	
RGC-0141	F	0.9	Other (Hypocalcemia)	3	*INVS*	NM_014425.3; c.2822A>G (p.H941R)(Het)(inherited from mother)	1/251378	6.868	Recessive disorder and only one heterozygous variant	
RGC-0141	F	0.9	Other (Hypocalcemia)	3	*ITGA8*	NM_003638.1; c.1156T>C (p.F386L)(Het)(inherited from mother)	14/282834	22.4	Recessive disorder and only one heterozygous variant	
RGC-0141	F	0.9	Other (Hypocalcemia)	3	*APOL1*	NM_003661.3; c.334C>T (p.R112C)(Het)(de novo)	4/251190	11.68	Variant discussed with experts and seems benign	
RGC-0142	F	0.5	Other (a-HUS)	1,4	*CFH*	NM_000186.3; c.3357C>G (p.D1119E)(Het)(inherited from mother)	3/282870	11.94	Patient has homozygous *CFHR3-CFHR1* deletion	Follow up
RGC-0142	F	0.5	Other (a-HUS)	1,4	*ITGA8*	NM_003638.2; c.840T>C(p.S280=)(Het)(inherited from father)	215/282194	7.427	Recessive disorder and only one heterozygous variant	
RGC-0148	F	2	Hematuria	1	*ACTN4*	NM_004924.5; c.751C>T (p.R251W)(Het)(inherited from father)	3/143328	32		Paternal kidney evaluation

RGC-0149	M	3	Hematuria	1	*COL4A4*	NM_000092.4; c.1442G>T (p.G481V)(Het)(inherited from mother)	1/143110	26	Segregation study suggests this variant causes hematuria in this family	
RGC-0150	M	1.6	Other (DI)	1	*AVPR2*	NM_000054.4; c.910+5G>T (intronic)(Hem)	0	9.197	Segregation study suggests this variant causes DI in this family	
RGC-0153	F	9	Other (Glycosuria)	1	*SLC5A2*	NM_00304.3; c.1665+4A>T (Het)(inherited from father)	0.003%	19.57		Determine father's phenotype
RGC-0158	M	8	Hematuria	1	*COL4A3*	NM_000091.4; c.4445C>T (p.A1482V)(Het)	223/143266	22.5	May describe phenotype	
RGC-0158	M	8	Hematuria	1	*NPHS1*	NM_004646.3; c.2614G>A (p.V872I)(Het)	3/143174	18.54	Pt also has nephrotic syndrome and also has a pathogenic variant in *NPHS2*	
RGC-0169	M	2	Cystic kidney	1	*PKD1*	NM_001009944.2; c.7146C>G (p.S2382R)(Het)	0	23.3		
RGC-0169	M	2	Cystic kidney	1	*NPHP1*	NM_000272.3; duplication of whole gene		NA	Reported in autism but there was not concern about DD or ASD in this patient	
RGC-0170	F	5	Proteinuria	1	*CUBN*	NM_001081.3; c.2677A>G (p.T893A)(Het)	146/143296	4.896	Phase of the two VUSs are unknown	Parents did not provide samples
RGC-0170	F	5	Proteinuria	1	*CUBN*	NM_001081.3;c.5285T>G, (p.V1762G)(Het)	12/282422	5.578	Phase of the two VUSs are unknown	Follow up
RGC-0170	F	5	Proteinuria	1	*NPHS1*	NM_004646.3; c.710T>C (p.L237P)(Het)	4/251428	31	Recessive disorder and only one heterozygous variant	
RGC-0170	F	5	Proteinuria	1	*PAX2*	M_003990.4; c.809G>A (p.R270H)(Het)	7/251366	29.2		Ophthalmology evaluation
RGC-0170	F	5	Proteinuria	1	*PLCE1*	NM_016341.3; c.642A>T (p.G214G)(Het)	293/280000	10.49	Recessive disorder and only one	

									heterozygous variant	
RGC-0170	F	5	Proteinuria	1	*SMARCAL1*	M_014140.3; c.1196C>T (p.T399M)(Het)	353/282896	0.899	Recessive disorder and only one heterozygous variant	
RGC-0170	F	5	Proteinuria	1	*WDR73*	NM_032856.3; c.481G>T (p.V161F)(Het)	5/248808	11.65	Recessive disorder and only one heterozygous variant	
RGC-0174	M	14	Other (Alagille syndrome)	1	*JAG1*	NM_000214.2; c.776G>T (p.G259V)(Het)(VUS)	0	26.6		tES recommended
RGC-0180	F	5	Proteinuria	1	*INF2*	NM_022489.3; c.2851C>T (p.R951W)(Het)	1/154628	23.6	Father had trace of protein in dipstick	Testing of siblings for *INF2*
RGC-0184	M	4	Proteinuria	3	*EVC2*	arr[GRCh37] 4p16.2(5616917_5699833)x3	NA	NA		Parental testing recommended
RGC-0187	F	4	Cystic kidney	3	*PUF60*	NM_078480.2 ; c.1292C>T (p.P43L)(Het)(inherited from father)	0	22.5	Father does not have kidney disease	
RGC-0187	F	4	Cystic kidney	3	*GLIS2*	NM_032575.2; c.1244C>T (p.P415L) (Het)(inherited from mother)	21/176552	26.8	Recessive disorder and only one heterozygous variant	
RGC-0187	F	4	Cystic kidney	3	*ARHGDIA*	NM_001301242.1; c.544A>G (p.T182A)(Hom)(both parents are carrier)	1/249280	6.032	Seems disease causing	Testing of siblings recommended
RGC-0187	F	4	Cystic kidney	3	*FLNA*	NM_001456.3; c.1399C>T (p.R467C)(Het)(inherited from mother)	1/177746	23.6	Mother unaffected	

DI, Diabetes insipidus; Es, Exome sequencing; PKDB, Autosomal Dominant Polycystic Kidney Disease Mutation Database; RBP, Retinol-binding Protein; TBM, Thin basement membrane; UA, Urine analysis; US, Ultrasound. Type of the testing;1, panel; 2, CMA; 3, proband ES; 4, trio ES; 5, Total Blueprint panel.