

## Supplementary Tables

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**Supplementary Table 1: List of genes sequenced using custom Roche NimbleGenSeqCap or a Roche NimbleGenHeatSeq panel**

Gene	OMIM
<i>ACE</i>	Renal tubular dysgenesis
<i>ACTN4</i>	Glomerulosclerosis, focal segmental, 1
<i>ADAMTS13</i>	Thrombotic thrombocytopenic purpura, familial
<i>ADCK4</i>	Nephrotic syndrome, type 9
<i>AGT</i>	Renal tubular dysgenesis
<i>AGTR1</i>	Renal tubular dysgenesis
<i>AGXT</i>	Hyperoxaluria, primary, type 1
<i>AHI1</i>	Joubert syndrome 3
<i>ANKS6</i>	Nephronophthisis 16
<i>ANLN</i>	Focal segmental glomerulosclerosis 8
<i>APOA1</i>	Amyloidosis, 3 or more types
<i>APOA2</i>	
<i>APOE</i>	Lipoprotein glomerulopathy
<i>APOL1</i>	End-stage renal disease, nondiabetic, susceptibility to glomerulosclerosis, focal segmental, 4, susceptibility to
<i>APRT</i>	Adenine phosphoribosyltransferase deficiency
<i>AQP2</i>	Diabetes insipidus, nephrogenic
<i>ARHGAP24</i>	None listed – for discussion of possible association with focal segmental glomerulosclerosis
<i>ARHGDI1</i>	Nephrotic syndrome, type 8
<i>ARL13B</i>	Joubert syndrome 8
<i>ARL6</i>	Bardet-Biedl syndrome 3
<i>ATP6V0A4</i>	Renal tubular acidosis, distal, autosomal recessive
<i>ATP6V1B1</i>	Renal tubular acidosis with progressive deafness
<i>ATP7B</i>	Wilson disease
<i>AVPR2</i>	Diabetes insipidus, nephrogenic Nephrogenic syndrome of inappropriate antidiuresis
<i>B2M</i>	Amyloidosis, familial visceral
<i>B9D1</i>	Meckel syndrome 9 Joubert syndrome 27
<i>B9D2</i>	Meckel syndrome 10 Joubert syndrome 34
<i>BBS1</i>	Bardet-Biedl syndrome 1
<i>BBS10</i>	Bardet-Biedl syndrome 10
<i>BBS12</i>	Bardet-Biedl syndrome 12
<i>BBS2</i>	Bardet-Biedl syndrome 2
<i>BBS4</i>	Bardet-Biedl syndrome 4
<i>BBS5</i>	Bardet-Biedl syndrome 5
<i>BBS7</i>	Bardet-Biedl syndrome 7
<i>BBS9</i>	Bardet-Biedl syndrome 9
<i>BICC1</i>	{Renal dysplasia, cystic, susceptibility to}
<i>BMP4</i>	Microphthalmia, syndromic 6

<i>BSND</i>	Bartter syndrome type 4a Sensorineural deafness with mild renal dysfunction
<i>C1QA</i>	C1q deficiency
<i>C1QB</i>	C1q deficiency
<i>C1QC</i>	C1q deficiency
<i>C3</i>	C3 deficiency {Hemolytic uremic syndrome, atypical, susceptibility to, 5}
<i>C5orf42</i>	Joubert syndrome 17
<i>CA2</i>	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis
<i>CASR</i>	Hypocalcaemia, autosomal dominant with Bartter syndrome Hypocalcuric hypercalcaemia, type I
<i>CC2D2A</i>	COACH syndrome Joubert syndrome Meckel syndrome
<i>CCND1</i>	{von Hippel-Lindau, syndrome, modifier of}
<i>CD151</i>	Nephropathy with pretibial epidermolysis bullosa and deafness
<i>CD2AP</i>	Glomerulosclerosis, focal segmental, 3
<i>CD46</i>	{Hemolytic uremic syndrome, atypical, susceptibility to, 2}
<i>CEP164</i>	Nephronophthisis 15
<i>CEP290</i>	Bardet-Biedl syndrome, Joubert syndrome, Meckel syndrome, Senior-Loken syndrome
<i>CEP41</i>	Joubert syndrome 15
<i>CEP83</i>	Nephronophthisis 18
<i>CFB</i>	{Hemolytic uremic syndrome, atypical, susceptibility to, 4}
<i>CFD</i>	Complement Factor D deficiency
<i>CFH</i>	Complement factor H deficiency and {Hemolytic uremic syndrome, atypical, susceptibility to, 1}
<i>CFHR1</i>	{Hemolytic uremic syndrome, atypical, susceptibility to}
<i>CFHR2</i>	
<i>CFHR3</i>	{Hemolytic uremic syndrome, atypical, susceptibility to}
<i>CFHR5</i>	Nephropathy due to CFHR5 deficiency
<i>CFI</i>	Complement factor I deficiency {Hemolytic uremic syndrome, atypical, susceptibility to, 3}
<i>CHD1L</i>	None listed
<i>CHRM3</i>	Prune belly syndrome
<i>CLCN5</i>	Dent disease Nephrolithiasis, type I Proteinuria, low molecular weight, with hypercalcuric nephrocalcinosis
<i>CLCNKA</i>	Bartter syndrome, type 4b, digenic
<i>CLCNKB</i>	Bartter syndrome, type 3 Bartter syndrome, type 4B, digenic
<i>CLDN16</i>	Hypomagnesemia 3, renal
<i>CLDN19</i>	Hypomagnesemia 5, renal, with ocular involvement
<i>CNNM2</i>	Hypomagnesemia 6, renal Hypomagnesemia, seizures and mental retardation
<i>COL4A1</i>	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps
<i>COL4A3</i>	Alport syndrome 2, autosomal recessive

	Alport Syndrome 3, autosomal dominant Haematuria, familial benign
<i>COL4A4</i>	Alport syndrome, autosomal recessive Haematuria, familial benign
<i>COL4A5</i>	Alport syndrome 1, X-linked
<i>COL4A6</i>	Deafness, X-linked 6
<i>COQ2</i>	Coenzyme Q10 deficiency, primary, 1
<i>COQ6</i>	Coenzyme Q10 deficiency, primary, 6
<i>CRB2</i>	Focal segmental glomerulosclerosis 9 Ventriculomegaly with cystic kidney disease
<i>CSPP1</i>	Joubert syndrome 21
<i>CTNS</i>	Cystinosis, nephropathic Cystinosis, late-onset juvenile or adolescent nephropathic Cystinosis, atypical nephropathic
<i>CUL3</i>	Pseudohypoaldosteronism, type IIE
<i>DCDC2</i>	Nephronophthisis 19
<i>DGKE</i>	Nephrotic syndrome, type 7
<i>DSTYK</i>	Congenital anomalies of kidney and urinary tract
<i>DZIP1L</i>	Polycystic kidney disease 5
<i>EHHADH</i>	Fanconi renotubular syndrome 3
<i>EMP2</i>	Nephrotic syndrome, type 10
<i>ETFA</i>	Glutaric acidemia IIA
<i>ETFB</i>	Glutaric acidemia IIB
<i>ETFDH</i>	Glutaric acidemia IIC
<i>EYA1</i>	Branchiootorenal syndrome 1, with or without cataracts
<i>FAH</i>	Tyrosinemia, type I
<i>FAN1</i>	Interstitial nephritis, karyomegalic
<i>FGF20</i>	Renal hypodysplasia/aplasia 2
<i>FGF23</i>	Hypophosphatemic rickets, autosomal dominant Tumoral calcinosis, hyperphosphatemic, familial 2
<i>FN1</i>	Glomerulopathy with fibronectin deposits 2
<i>FRAS1</i>	Fraser syndrome 1
<i>FREM1</i>	Manitoba oculotrichoanal syndrome
<i>FREM2</i>	Fraser syndrome 2
<i>FXD2</i>	Hypomagnesemia 2, renal
<i>GANAB</i>	Polycystic kidney disease 3
<i>GATA3</i>	Hypoparathyroidism, sensorineural deafness, and renal dysplasia
<i>GLA</i>	Fabry disease
<i>GLI3</i>	Pallister-Hall syndrome
<i>GLIS2</i>	Nephronophthisis 7
<i>GPC3</i>	Simpson-Golabi-Behmel syndrome, type 1 Wilms tumour, somatic
<i>GRHPR</i>	Hyperoxaluria, primary, type II
<i>GRIP1</i>	Fraser syndrome 3
<i>GSN</i>	Amyloidosis, Finnish type
<i>HNF1B</i>	Autosomal Dominant Tubulointerstitial Disease- <i>HNF1B</i>
<i>HNF4A</i>	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young
<i>HOGA1</i>	Hyperoxaluria, primary, type III

<i>HPSE2</i>	Urofacial syndrome 1
<i>HSD11B2</i>	Apparent mineralocorticoid excess
<i>IFNG</i>	
<i>IFT122</i>	Cranioectodermal dysplasia 1
<i>IFT140</i>	Short-rib thoracic dysplasia 9 with or without polydactyly
<i>IFT43</i>	Cranioectodermal dysplasia 3
<i>INF2</i>	Glomerulosclerosis, focal segmental, 5
<i>INPP5E</i>	Joubert syndrome 1
<i>INVS</i>	Nephronophthisis 2, infantile
<i>IQCB1</i>	Senior-Loken syndrome 5
<i>ITGA3</i>	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital
<i>ITGA8</i>	Renal hypodysplasia/aplasia 1
<i>JAG1</i>	Alagille Syndrome
<i>KAL1</i>	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)
<i>KCNJ1</i>	Bartter syndrome, type 2
<i>KIF7</i>	Joubert syndrome 12
<i>KLHL3</i>	Pseudohypoaldosteronism, type IID
<i>LAMB2</i>	Nephrotic syndrome, type 5, with or without ocular abnormalities
<i>LCAT</i>	Norum disease
<i>LMX1B</i>	Nail-patella syndrome
<i>LRIG2</i>	Urofacial syndrome 2
<i>LYZ</i>	Amyloidosis, renal
<i>MAGI2</i>	Nephrotic syndrome 15
<i>MAPKBP1</i>	Nephronophthisis 20
<i>MASP1</i>	3MC syndrome
<i>MEFV</i>	Familial Mediterranean fever
<i>MKKS</i>	Bardet-Biedl syndrome 6
<i>MKS1</i>	Bardet-Biedl syndrome 13 Joubert syndrome 28 Meckel syndrome 1
<i>MUC1</i>	Autosomal Dominant Tubulointerstitial Disease- <i>MUC1</i>
<i>MYH9</i>	Epstein syndrome and Fechtner syndrome
<i>MYO1E</i>	Glomerulosclerosis, focal segmental, 6
<i>NEK8</i>	Nephronophthisis 9 Renal-hepatic-pancreatic dysplasia 2
<i>NOTCH2</i>	Alagille syndrome 2 Hajdu-Cheney syndrome
<i>NPHP1</i>	Joubert syndrome 4 Nephronophthisis 1 Senior-Loken syndrome
<i>NPHP3</i>	Meckel syndrome 7 Nephronophthisis 3 Renal-hepatic-pancreatic dysplasia 1
<i>NPHP4</i>	Nephronophthisis 4 Senior-Loken syndrome 4
<i>NPHS1</i>	Nephrotic syndrome, type 1
<i>NPHS2</i>	Nephrotic syndrome, type 2

<i>NR3C2</i>	Pseudohypoaldosteronism type I, autosomal dominant
<i>NUP107</i>	Nephrotic syndrome, type 11
<i>NUP205</i>	Nephrotic syndrome, type 13
<i>NUP93</i>	Nephrotic syndrome, type 12
<i>OCRL</i>	Dent disease 2 Lowe syndrome
<i>OFD1</i>	Joubert syndrome 10
<i>PAX2</i>	Papillonephrosis Glomerulosclerosis, focal segmental, 7
<i>PDSS2</i>	Coenzyme Q10 deficiency, primary, 3
<i>PKD1</i>	Polycystic kidney disease 1
<i>PKD2</i>	Polycystic kidney disease 2
<i>PKHD1</i>	Polycystic kidney disease 4, with or without hepatic disease
<i>PKHD1L1</i>	None listed but suggested as a PKD locus
<i>PLCE1</i>	Nephrotic syndrome, type 3
<i>PRPS1</i>	Phosphoribosylpyrophosphate synthetase superactivity
<i>PTPRO</i>	Nephrotic syndrome, type 6
<i>REN</i>	Renal tubular dysgenesis Hyperuricemic nephropathy, familial juvenile 2
<i>RET</i>	Congenital renal defects
<i>ROBO2</i>	Vesicoureteral reflux 2
<i>RPGRIP1L</i>	COACH syndrome Joubert syndrome 7 Meckel syndrome 5
<i>RRM2B</i>	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)
<i>SALL1</i>	Townes-Brocks syndrome 1 Townes-Brocks branchiotorenal-like syndrome
<i>SARS2</i>	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis
<i>SCARB2</i>	Epilepsy, progressive myoclonic 4, with or without renal failure
<i>SCNN1B</i>	Liddle Syndrome Pseudohypoaldosteronism
<i>SCNN1G</i>	Liddle Syndrome Pseudohypoaldosteronism
<i>SDCCAG8</i>	Bardet-Biedl syndrome 16 Senior-Loken syndrome 7
<i>SEC61A1</i>	Hyperuricemic Nephropathy, familial juvenile, 4
<i>SGPL1</i>	Nephrotic syndrome 14
<i>SIX1</i>	Branchiootic syndrome 3
<i>SIX5</i>	Branchiootorenal syndrome 2
<i>SLC12A1</i>	Bartter syndrome, type 1
<i>SLC12A3</i>	Gitelman syndrome
<i>SLC1A1</i>	Dicarboxylic aminoaciduria
<i>SLC22A12</i>	Hypouricemia, renal
<i>SLC2A2</i>	Fanconi-Bickel syndrome
<i>SLC2A9</i>	Hypouricemia, renal, 2
<i>SLC34A1</i>	Nephrolithiasis/osteoporosis, hypophosphatemic, 1 Fanconi renal tubular syndrome 2
<i>SLC3A1</i>	Cystinuria
<i>SLC4A1</i>	Renal tubular acidosis

<i>SLC4A4</i>	Renal tubular acidosis, proximal, with ocular abnormalities
<i>SLC5A2</i>	Renal glucosuria
<i>SLC7A7</i>	Lysinuric protein intolerance
<i>SLC7A9</i>	Cystinuria
<i>SLC9A3R1</i>	Nephrolithiasis/osteoporosis, hypophosphatemic, 2
<i>SMARCAL1</i>	Schimke immunosseous dysplasia
<i>SOX17</i>	Vesicoureteral reflux 3
<i>TBX18</i>	Congenital anomalies of kidney and urinary tract 2
<i>TCTN1</i>	Joubert syndrome 13
<i>TCTN2</i>	Joubert syndrome 24 Meckel Syndrome 8
<i>TCTN3</i>	Joubert syndrome 18
<i>THBD</i>	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}
<i>TMEM138</i>	Joubert syndrome 16
<i>TMEM216</i>	Joubert syndrome 2 Meckel syndrome 2
<i>TMEM231</i>	Joubert syndrome 20 Meckel syndrome 11
<i>TMEM237</i>	Joubert syndrome 14
<i>TMEM67</i>	COACH syndrome Joubert syndrome Meckel syndrome Nephronophthisis 11 Bardet Biedl Syndrome 14, modifier of
<i>TNXB</i>	Vesicoureteral reflux 8
<i>TRIM32</i>	Bardet-Biedl syndrome 11
<i>TRPC6</i>	Glomerulosclerosis, focal segmental, 2
<i>TSC1</i>	Tuberous sclerosis-1
<i>TSC2</i>	Tuberous Sclerosis-2
<i>TTC21B</i>	Nephronophthisis 12
<i>TTC8</i>	Bardet-Biedl syndrome 8
<i>UMOD</i>	Autosomal Dominant Tubulointerstitial Disease- <i>UMOD</i>
<i>VHL</i>	von Hippel-Lindau syndrome
<i>VIPAS39</i>	Arthrogryposis, renal dysfunction, and cholestasis 2
<i>VPS33B</i>	Arthrogryposis, renal dysfunction, and cholestasis 1
<i>WDPCP</i>	?Bardet-Biedl syndrome 15
<i>WDR19</i>	Nephronophthisis 13 Senior-Loken syndrome
<i>WDR35</i>	Cranioectodermal dysplasia 2 Short-rib thoracic dysplasia 7 with or without polydactyly
<i>WDR73</i>	Galloway-Mowat syndrome
<i>WNK4</i>	Pseudohypoaldosteronism, type IIB
<i>WNT4</i>	SERKAL Syndrome
<i>WT1</i>	Nephrotic syndrome type 4 Wilms tumour Denys-Drash syndrome Frasier syndrome
<i>XPNPEP3</i>	Nephronophthisis-like nephropathy 1
<i>ZMYND10</i>	Ciliary dyskinesia, primary, 22



ZNF423	Joubert syndrome 19 Nephronophthisis 14

**Supplementary Table 2: Original and revised pathological diagnosis in patients who underwent percutaneous renal biopsy**

**DDD**, Dense Deposit Disease; **EM**, Electronmicroscopy; **FSGS**, Focal segmental glomerulosclerosis, **GN**, Glomerulonephritis; **IF**, immunofluorescence; **IgA**, Immunglobin A; **LM**, light microscopy; **MPGN**, Membranoproliferative Glomerulonephritis **TI**, tubulointerstitial; **TIKD**, Tubulointersitital kidney disease; **TMA**, Thrombotic Microangiopathy

Family Number	Number	Original Diagnostic Category	Original pathological Diagnosis	Family History	Number of Glomeruli on LM	Percentage Fibrosis (%)	Reviewed	Available for Review			Change in Diagnosis	New Diagnostic Category	New Pathological Diagnosis
								LM	IF	EM			
<b>Patients with a Molecular Diagnosis</b>													
1	1A	TIKD	Familial TIKD	Yes	3	20	Yes	Yes	No	No	No	TIKD	Familial TIKD
2	2A	TIKD	TIKD/ Gouty Nephropathy	Yes	18	50	Yes	Yes	Yes	No	No	TIKD	TIKD/Gouty Nephropathy
2	2B	TIKD	TI Fibrosis	Yes	7	20	Yes	Yes	Yes	Yes	No	TIKD	TI Fibrosis
2	2C	TIKD	TI Fibrosis	Yes	60	60	Yes	Yes	Yes	Yes	No	TIKD	TI Fibrosis
2	2D	Glomerulonephritis	MPGN/DDD	Yes	5	20	Yes	Yes	Yes	No	No	Glomerulonephritis	MPGN
3	3A	TIKD	Familial TIKD	Yes	12	80	Yes	Yes	Yes	Yes	No	TIKD	Familial TIKD
3	3B	TIKD	Familial TIKD	Yes	16	70	Yes	Yes	Yes	Yes	No	TIKD	Familial TIKD
3	3C	TIKD	Active TI nephritis	Yes	38	75	Yes	Yes	Yes	Yes	No	TIKD	Active TI nephritis
3	3D	TIKD	Familial TIKD	Yes	2	75	Yes	Yes	Yes	Yes	No	TIKD	Familial TIKD
3	3E	TIKD	Familial TIKD	Yes	12	75	Yes	Yes	Yes	Yes	No	TIKD	Familial TIKD
3	3F	TIKD	TI Fibrosis	Yes	18	10	Yes	Yes	Yes	Yes	No	TIKD	TI Fibrosis
4	4A	TIKD	Early fibrosis	Yes	2	10	Yes	Yes	Yes	Yes	No	TIKD	Early TI fibrosis
4	4B	TMA	TMA	Yes	11	20	Yes	Yes	Yes	Yes	No	TMA	TMA
5	5A	TIKD	TI Inflammation	Yes	0	?	No	No	No	No	No	TIKD	TI Inflammation
6	6A	TIKD	Early nephronophthisis	Yes	19	<10	Yes	Yes	Yes	Yes	No	TIKD	Early nephronophthisis
6	6B	TIKD	Juvenile nephronophthisis	Yes	14	60-70	Yes	Yes	Yes	Yes	Yes	TMA	TMA & TIKD
7	7A	Glomerulonephritis	Proliferative GN	Yes	20	10	Yes	Yes	Yes	No	No	Glomerulonephritis	Proliferative Glomerulonephritis
8	8A	Glomerulonephritis	IgA Nephropathy	Yes	22	30	Yes	Yes	Yes	Yes	No	Glomerulonephritis	IgA Nephropathy
8	8B	Non-Specific	Arteriosclerosis with fibrosis	Yes	3	70	Yes	Yes	Yes	Yes	No	Non-specific	Arteriosclerosis with fibrosis
9	9A	Glomerulonephritis	Focal Proliferative GN	Yes	14	25	Yes	Yes	Yes	Yes	No	Glomerulonephritis	Focal Proliferative GN

11	11A	FSGS/Alport	FSGS	Yes	24	80	Yes	Yes	Yes	Yes	No	FSGS/Alport	FSGS
12	12A	FSGS/Alport	Alport Syndrome	Yes	1	10	Yes	Yes	Yes	Yes	No	FSGS/Alport	Alport Syndrome
13	13A	FSGS/Alport	Alport Syndrome	Yes	4	10	Yes	Yes	Yes	Yes	No	FSGS/Alport	Alport Syndrome
14	14A	FSGS/Alport	FSGS/ADPKD	No	6	>50	Yes	Yes	Yes	Yes	No	FSGS/Alport	FSGS
15	15A	TIKD	TIKD	Yes	166	50	Yes	Yes	Yes	Yes	No	TIKD	TIKD
15	15B	TMA	Chronic TMA/FSGS	Yes	24	75	Yes	Yes	Yes	Yes	No	TMA	Chronic TMA/FSGS
15	15C	TMA	Chronic TMA	Yes	12	30	Yes	Yes	Yes	Yes	No	TMA	Chronic TMA
16	16A	FSGS/Alport	Alport Syndrome	Yes	2	10	Yes	Yes	Yes	Yes	No	FSGS/Alport	Alport Syndrome
17	17A	FSGS/Alport	Alport Syndrome	Yes	6	30-55	Yes	Yes	Yes	Yes	No	FSGS/Alport	Alport Syndrome
18	18A	Glomerulonephritis	Fibrillary GN	Yes	29	40	Yes	Yes	Yes	Yes	Yes	TMA	TMA & TBMD
18	18B	Glomerulonephritis	Fibrillary GN	Yes	12	50	Yes	Yes	Yes	Yes	Yes	TMA	TMA & TBMD
18	18C	Glomerulonephritis	Fibrillary GN	Yes	16	20	Yes	Yes	Yes	Yes	Yes	TMA	TMA & TBMD
18	18D	Glomerulonephritis	Fibrillary GN	Yes	3	60	Yes	Yes	Yes	Yes	Yes	TMA	TMA & TBMD
19	19A	TIKD	TIKD	Yes	10	20	Yes	Yes	Yes	Yes	Yes	TMA	TMA & TBMD
20	20A	TMA	Acute TMA	Yes	12	15	Yes	Yes	Yes	Yes	No	TMA	Acute TMA
20	20B	Non-Specific	Oligomegonephronia	Yes	7	75	Yes	Yes	Yes	Yes	No	Non-specific	Oligomegonephronia
21	21A	Non-Specific	Within normal limits	Yes	36	0	Yes	Yes	Yes	Yes	No	Non-specific	Within normal limits
21	21B	Non-Specific	Mesangial Proliferation	Yes	10	60-70	yes	Yes	Yes	Yes	No	Non-specific	Mesangial Proliferation
22	22A	Non-Specific	Arteriosclerosis	Yes	53	5	No	No	No	No	No	Non-specific	Arteriosclerosis
23	23A	Non-Specific	Severe fibrosis	No	0	>70	No	No	No	No	No	Non-specific	Severe fibrosis

**Patients without a Molecular Diagnosis**

1	1A	TIKD	Familial TIKD	Yes	3	20	Yes	Yes	No	No	No	TIKD	Familial TIKD
10	10A	Glomerulonephritis	IgA Nephropathy	Yes	30	20-25	Yes	Yes	Yes	Yes	No	Glomerulonephritis	IgA Nephropathy
10	10B	Glomerulonephritis	IgA Nephropathy	Yes	11	60	Yes	Yes	Yes	Yes	No	Glomerulonephritis	IgA Nephropathy
10	10C	TMA	Chronic TMA	Yes	17	60	Yes	Yes	Yes	Yes	No	TMA	Chronic TMA
24	24A	TIKD	Chronic Allergic Interstitial Nephritis	Yes	20	80	Yes	Yes	Yes	Yes	No	TIKD	Chronic Interstitial Nephritis - allergic type
25	25A	TIKD	Acute interstitial nephritis	Yes	6	75	Yes	Yes	Yes	Yes	no	TIKD	Acute interstitial nephritis

26	26A	Non-specific	Non-specific mild changes	Yes	7	<10	No	No	No	No	No	Non-specific	Non-specific mild changes
26	26b	TIKD	Nephronophtthisis	Yes	9	50	Yes	Yes	Yes	Yes	No	TIKD	Nephonophtthisis
26	26C	TMA	Chronic TMA	Yes	4	15	Yes	Yes	Yes	Yes	No	TMA	Chronic TMA
27	27A	TIKD	Chronic Interstitial Nephritis	Yes	31	<10	Yes	Yes	Yes	No	No	TIKD	TIKD
28	28A	Non-specific	Arteriosclerosis	Yes	57	75	Yes	Yes	Yes	Yes	Yes	TMA	Chronic TMA
28	28B	Glomerulonephritis	IgA Nephropathy	Yes	13	75	Yes	Yes	Yes	No	No	Glomerulonephritis	IgA Nephropathy
28	28C	Non-specific	Hypertensive Nephrosclerosis	Yes	5	50	Yes	Yes	Yes	no	No	Non-specific	Hypertensive Nephrosclerosis
29	29A	Glomerulonephritis	IgA Nephropathy	Yes	26	40	Yes	Yes	Yes	No	No	Glomerulonephritis	IgA Nephropathy
29	29b	Glomerulonephritis	IgA Nephropathy	Yes	41	50	Yes	Yes	Yes	No	No	Glomerulonephritis	IgA Nephropathy
30	30A	Glomerulonephritis	IgA Nephropathy	Yes	11	20	Yes	Yes	Yes	Yes	No	Glomerulonephritis	IgA Nephropathy
31	31A	Glomerulonephritis	IgA Nephropathy	Yes	12	15	Yes	Yes	Yes	Yes	No	Glomerulonephritis	IgA Nephropathy
32	32A	Glomerulonephritis	IgA Nephropathy	Yes	6	<10	Yes	Yes	Yes	No	No	Glomerulonephritis	IgA Nephropathy
32	32B	Glomerulonephritis	IgA Nephropathy	Yes	20	75	Yes	Yes	Yes	Yes	No	Glomerulonephritis	IgA Nephropathy
33	33B	Glomerulonephritis	Crescentic GN	No	18	<19	Yes	Yes	Yes	No	No	Glomerulonephritis	Childhood GN
34	34A	Glomerulonephritis	MPGN	Yes	11	<10	Yes	Yes	Yes	Yes	Yes	TMA	Chronic TMA
34	34B	FSGS/Alport	Alport Syndrome	Yes	18	70	Yes	Yes	Yes	Yes	No	FSGS/Alport	Alport Syndrome
34	34C	TMA	Chronic TMA	Yes	16	20	Yes	Yes	Yes	Yes	No	TMA	Chronic TMA
35	35A	Glomerulonephritis	Congenital Nephrotic Syndrome	No	20	30	Yes	Yes	Yes	Yes	Yes	FSGS/Alport	FSGS
36	36A	Glomerulonephritis	Mesangial GN/Minimal Change variant	No	17	0	Yes	Yes	Yes	Yes	No	Glomerulonephritis	Mesangial GN/Minimal Change Variant
37	37A	FSGS/Alport	Alport Syndrome	Yes	-	-	Yes	Yes	Yes	Yes	No	FSGS/Alport	Alport Syndrome
38	38A	FSGS/Alport	FSGS	Yes	18	35	Yes	Yes	Yes	Yes	No	FSGS/Alport	FSGS
39	39A	FSGS/Alport	FSGS	No	19	30	Yes	Yes	Yes	Yes	No	FSGS/Alport	FSGS
40	40A	TMA	Chronic TMA	Yes	16	60	Yes	Yes	Yes	Yes	No	TMA	Chronic TMA
41	41A	TMA	Acute or organising TMA	no	22	75	Yes	Yes	Yes	Yes	No	TMA	Acute or organising TMA
42	42A	Non-specific	Hypertensive Nephrosclerosis	Yes	16	60	Yes	Yes	Yes	Yes	no	Non-specific	Hypertensive Nephrosclerosis
43	43A	Non-specific	Arteriosclerosis	Yes	13	80	Yes	Yes	Yes	Yes	No	Non-specific	Arteriosclerosis
44	44A	Non-specific	Arteriosclerosis	Yes	24	70	Yes	Yes	Yes	Yes	No	Non-specific	Arteriosclerosis
45	45A	TMA	Chronic TMA	Yes	5	30	Yes	Yes	No	Yes	No	TMA	Chronic TMA

50	47A	Non-specific	Glomerulosclerosis	Yes	24	50	no	no	No	No	No	Non-specific	Glomerulosclerosis
51	48A	Non-specific	Prominent Glomerulosclerosis	Yes	53	30	No	No	No	No	No	Non-Specific	Prominent Glomerulosclerosis

**Supplementary Table 3: ACMG Evidence in those in those in whom a pathogenic variant was detected, including presence or absence of ACMG pathogenic criteria weighted as very strong (PVS1), strong (PS1-4), moderate (PM1-6), or supporting (PP1-5)**

**AD**, Autosomal dominant; **AR**, Autosomal recessive; **c.**, coding DNA; **CHR**, Chromosome number; **D.C.**, Disease causing **Del**, deleterious; **F**, female; **het**, heterozygous; **hom**, homozygous, **M**, male; **N**, no; **p**, protein; **path**, pathogenic **Y**, yes

\***PPT**, Polyphen 2 (<http://genetics.bwh.harvard.edu/pph2>)

#**SIFT**, Sorting Intolerant from tolerant (<http://sift.jcvi.org/>)

~**MT**, Mutation taster (<http://mutationtaster.org>)

ID	Institute at Which Diagnosis Was Made	Gene	Sex	CHR	Zygosity	Inheritance	Genetic Disease (OMIM Phenotype MIM No.)	c and p change	RefSeq ID	PP2* SIFT# MT~	PVS	PS1	PS2	PS3	PS4	PM1	PM2	PM3	PM4	PM5	PM6	PP1	PP2	PP3	PP4	pp5	variant class
2A	RCSI	UMOD	M	16	het	AD	Medullary Cystic Kidney Disease Type II (603860)	c.G767G>A p.Cys256Tyr	Nc_000016.10	-	N	N	N	N	N	N	Y	N	N	N	N	Y	Y	Y	Y	N	Likely path.
2B	Wake Forest	UMOD	F	16	het	AD	Medullary Cystic Kidney Disease Type II (603860)	c.G767G>A p.Cys256Tyr	Nc_000016.10	-	N	N	N	N	N	N	Y	N	N	N	N	Y	Y	Y	Y	N	Likely path.
2C	Wake Forest	UMOD	M	16	het	AD	Medullary Cystic Kidney Disease Type II (603860)	c.G767G>A p.Cys256Tyr	Nc_000016.10	-	N	N	N	N	N	N	Y	N	N	N	N	Y	Y	Y	Y	N	Likely path.
2D	Wake Forest	UMOD	M	16	het	AD	Medullary Cystic Kidney Disease Type II (603860)	c.G767G>A p.Cys256Tyr	Nc_000016.10	-	N	N	N	N	N	N	Y	N	N	N	N	Y	Y	Y	Y	N	Likely path.
3A	Wake Forest	MUC1	F	1	het	AD	Medullary Cystic Kidney Disease Type I (174000)	c. ins(3n+1) in VNTR p. MUC1fs	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	Y	N	N	N	N	Path.
3B	Wake Forest	MUC1	F	1	het	AD	Medullary Cystic Kidney Disease Type I (174000)	c. ins(3n+1) in VNTR p. MUC1fs	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	Y	N	N	N	N	Path.
3C	Wake Forest	MUC1	F	1	het	AD	Medullary Cystic Kidney Disease Type I (174000)	c. ins(3n+1) in VNTR p. MUC1fs	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	Y	N	N	N	N	Path.
3D	Wake Forest	MUC1	M	1	het	AD	Medullary Cystic Kidney Disease Type I (174000)	c. ins(3n+1) in VNTR p. MUC1fs	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	Y	N	N	N	N	Path.

3E	Wake Forest	MUC1	M	1	het	AD	Medullary Cystic Kidney Disease Type I (174000)	c. ins(3n+1) in VNTR p. MUC1fs	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	Y	N	N	N	N	Path.
3F	Wake Forest	MUC1	F	1	het	AD	Medullary Cystic Kidney Disease Type I (174000)	c. ins(3n+1) in VNTR	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	Y	N	N	N	N	Path.
4A	Boston Children's Hospital	HNF1B	F	17	het	AD	Renal Cysts and Diabetes Syndrome (137920)	c.544+3_544 +6del /	NM_000458.2	:	Y	N	N	N	N	N	Y	N	N	N	N	N	N	N	N	N	Likely path.
4B	Boston Children's Hospital	HNF1B	M	17	het	AD	Renal Cysts and Diabetes Syndrome (137920)	c.544+3_544 +6del /	NM_000458.2	-	Y	N	N	N	N	N	Y	N	N	N	N	N	N	N	N	N	Likely path.
5A	Boston Children's Hospital	NPHP1	F	2	hom	AR	Nephron- ophthisis 1, juvenile (256100)	c.555_556insA p.Pro186Hisfs* 2	NM_000272.3	:	Y	N	N	N	N	N	Y	N	N	N	N	Y	N	N	N	N	Path.
6A	Boston Children's Hospital	IFT140	M	16	hom	AR	Mainzer-Saldino Syndrome (266920)	c.634G>A p.Gly212Arg	NM_014714.3	1.0 Del. D.C	N	Y	N	N	N	N	Y	N	N	N	Y	N	Y	Y	N	Path.	
6B	Boston Children's Hospital	IFT140	F	16	hom	AR	Mainzer-Saldino Syndrome (266920)	c.634G>A p.Gly212Arg	NM_014714.3	1.0 Del. D.C	N	Y	N	N	N	N	Y	N	N	N	Y	N	Y	Y		Path.	
7A	Wake Forest	MUC1	F	16	het	AD	Medullary cystic kidney disease type I (174000)	c. ins(3n+1) in VNTR p. MUC1fs	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	Y	N	N	N	N	Path.
8A	RCSI	COL4A 5	M	X	hemi	X linke d	Alport Syndrome 1, X linked (301050)	c.2959_2976 del p.987_992del	NM_000495	-	Y	N	N	N	N	N	Y	N	Y	N	N	N	N	N	Y	Y	Likely path.
8B	RCSI	COL4A 5	M	X	hemi	X linke d	Alport Syndrome 1, X linked (301050)	c.2959_2976 del p.987_992del	NM_000495	-	Y	N	N	N	N	N	Y	N	Y	N	N	N	N	N	Y	Y	Likely path.
9A	RCSI	COL4A 5	M	X	hemi	X linke d	Alport Syndrome 1, X linked (301050)	c.3427 G>A p.Gly1143Ser	NM_000495	1.0 Del. D.C.	N	N	N	N	N	Y	Y	N	N	Y	N	N	Y	Y	N	N	Likely path.
11 A	Boston Children's Hospital	COL4A 5	M	X	hemi	X linke d	Alport Syndrome 1, X linked (301050)	c.2605G>A p.Gly869Arg	NM_033380.2	1.0 Del. D.C.	N	N	N	Y	N	N	Y	Y	N	N	N	Y	Y	Y	Y	N	Path.
12 A	Boston Children's Hospital	COL4A 5	F	X	hemi	X linke d	Alport Syndrome 1, X linked (301050)	c.2396G>A p.Gly799Asp	NM_033380.2	1.0 Del. D.C.	N	N	N	N	N	N	Y	Y	N	N	N	Y	Y	Y	N	Likely path.	
13 A	Boston Children's Hospital	COL4A 5	M	X	hemi	X linke d	Alport Syndrome 1, X linked (301050)	c.1423+1G>T	NM_033380.2	-	Y	N	N	N	N	N	Y	Y	N	N	N	N	N	N	Y	N	Path.
14 A		FANCI	M	15	hom	AR	Fanconi Anaemia,	c.217A>T p.Ile73Phe	NM_0011133 78.1	0.81 Del. D.C.	N	N	N	N	N	N	Y	N	N	N	Y	N	Y	Y	N	Likely Path.	

	Boston Children's Hospital						complementation group I (609053)																					
15 A	Wake Forest	UMOD	F	16	het	AD	Medullary Cystic Kidney Disease, Type II (603860)	c.317G>A p.Cys106Tyr	Nc_000016.10	-	N	N	N	N	N	N	Y	N	N	N	N	Y	Y	Y	Y	N	Likely Path.	
15 B	Wake Forest	UMOD	M	16	het	AD	Medullary Cystic Kidney Disease, Type II (603860)	c.317G>A p.Cys106Tyr	Nc_000016.10	-	N	N	N	N	N	N	Y	N	N	N	N	Y	Y	Y	Y	N	Likely Path.	
15 C	Wake Forest	UMOD	M	16	het	AD	Medullary Cystic Kidney Disease, Type II (603860)	c.317G>A p.Cys106Tyr	Nc_000016.10	-	N	N	N	N	N	N	Y	N	N	N	N	Y	Y	Y	Y	N	Likely path.	
16 A	RCSI	COL4A5	M	X	hemi	AD	Alport Syndrome 1, X linked (301050)	c.1762G>A p.Gly588Ser	NM_000495	1.0 Del D.C.	N	N	N	N	N	N	Y	Y	N	N	N	N	Y	Y	Y	N	Likely path.	
17 A	RCSI	COL4A5	M	X	hemi	X linked	Alport Syndrome 1, X linked (301050)	c.3310G>T p.Gly1104Cys	NM_000495	1.0 Del D.C.	N	N	N	N	N	N	Y	Y	N	N	N	N	N	Y	Y	N	Likely path.	
18 A	RCSI	INF2	M	14	het	AD	Glomerulosclerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
18 B	RCSI	INF2	F	14	het	AD	Glomerulosclerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
18 C	RCSI	INF2	M	14	het	AD	Glomerulosclerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
18 D	RCSI	INF2	M	14	het	AD	Glomerulosclerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
19 A	Wake Forest	MUC1	M	16	het	AD	Medullary Cystic Kidney Disease, type I (174000)	c. ins(3n+1) in VNTR p. MUC1fs	-	-	Y	Y	N	Y	N	N	N	N	N	N	N	N	N	N	N	N	Path.	
20 A	RCSI	HNF1B	F	17	het	AD	Renal Cysts and Diabetes Syndrome (137920)	c.1255_1256del p.Ala419fs	NM_001304286	-	Y	N	N	N	N	N	Y	N	N	N	N	Y	N	N	Y	N	Path.	
20 B	RCSI	HNF1B	M	17	het	AD	Renal Cysts and Diabetes Syndrome (137920)	c.1255_1256del p.Ala419fs	NM_001304286	-	Y	N	N	N	N	N	Y	N	N	N	N	Y	N	N	Y	N	Path.	
21 A	Boston Children's Hospital	C3	M	19	het	AD	Susceptibility to atypical haemolytic syndrome (612925)	c.4534C>T p.Arg1512Cys	NM_000064.2	0.65 Del. D.C.	N	N	N	N	N	N	N	N	N	Y	N	Y	Y	Y	Y	N	Likely path.	

21 B	Boston Children's Hospital	<i>INF2</i>	F	14	het	AD	Glomerulos- clerosis focal segmental, 5 (613237)	c.353T>A p.Ile118Asn	NM_022489.3	1.0 Del. D.C.	N	N	N	N	N	N	Y	N	N	Y	N	Y	N	Y	N	N	Likely path.
22 A	Boston Children's Hospital	<i>WNK4</i>	F	17	het	AD	Pseudo- hypoaldoster- onism - hypertensive CKD (614491)	c.506C>T p.Pro169Leu	NM_032387.4	0.69 Del. D.C.	N	Y	N	N	N	N	Y	N	N	N	N	y	N	Y	Y	N	Path.
23 A	Boston Children's Hospital	<i>SLC3A1</i>	M	2	het	AD	Cystinuria (220100)	c.1799G>A p.Gly600Glu	NM_000341.3	1.0 Del. D.C.	N	Y	N	N	N	N	N	N	N	N	N	Y	Y	Y	N	Likely path	



