Supplementary Tables

List of Supplementary Tables

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

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

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

	Alport Syndrome 3, autosomal dominant Haematuria, familial benign
COL4A4	Alport syndrome, autosomal recessive Haematuria, familial benign
COL4A5	Alport syndrome 1, X-linked
COL4A6	Deafness, X-linked 6
COQ2	Coenzyme Q10 deficiency, primary, 1
COQ6	Coenzyme Q10 deficiency, primary, 6
CRB2	Focal segmental glomerulosclerosis 9 Ventriculomegaly with cystic kidney disease
CSPP1	Joubert syndrome 21
CTNS	Cystinosis, nephropathic Cystinosis, late-onset juvenile or adolescent nephropathic Cystinosis, atypical nephropathic
CUL3	Pseudohypoaldosteronism, type IIE
DCDC2	Nephronopthisis 19
DGKE	Nephrotic syndrome, type 7
DSTYK	Congenital anomalies of kidney and urinary tract
DZIP1L	Polycystic kidney disease 5
EHHADH	Fanconi renotubular syndrome 3
EMP2	Nephrotic syndrome, type 10
ETFA	Glutaric acidemia IIA
ETFB	Glutaric acidemia IIB
ETFDH	Glutaric acidemia IIC
EYA1	Branchiootorenal syndrome 1, with or without cataracts
FAH	Tyrosinemia, type I
FAN1	Interstitial nephritis, karyomegalic
FGF20	Renal hypodysplasia/aplasia 2
FGF23	Hypophosphatemic rickets, autosomal dominant Tumoral calcinosis, hyperphosphatemic, familial 2
FN1	Glomerulopathy with fibronectin deposits 2
FRAS1	Fraser syndrome 1
FREM1	Manitoba oculotrichoanal syndrome
FREM2	Fraser syndrome 2
FXYD2	Hypomagnesemia 2, renal
GANAB	Polycystic kidney disease 3
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia
GLA	Fabry disease
GLI3	Pallister-Hall syndrome
GLIS2	Nephronophthisis 7
GPC3	Simpson-Golabi-Behmel syndrome, type 1 Wilms tumour, somatic
GRHPR	Hyperoxaluria, primary, type II
GRIP1	Fraser syndrome 3
GSN	Amyloidosis, Finnish type
HNF1B	Autosomal Dominant Tubulointerstitial Disease- *HNF1B*
HNF4A	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young
HOGA1	Hyperoxaluria, primary, type III

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

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

SLC4A4	Renal tubular acidosis, proximal, with ocular abnormalities
SLC5A2	Renal glucosuria
SLC7A7	Lysinuric protein intolerance
SLC7A9	Cystinuria
SLC9A3R1	Nephrolithiasis/osteoporosis, hypophosphatemic, 2
SMARCAL1	Schimke immunoosseous dysplasia
SOX17	Vesicoureteral reflux 3
TBX18	Congenital anomalies of kidney and urinary tract 2
TCTN1	Joubert syndrome 13
TCTN2	Joubert syndrome 24 Meckel Syndrome 8
TCTN3	Joubert syndrome 18
THBD	{Hemolytic uremic syndrome, atypical, susceptibility to, 6}
TMEM138	Joubert syndrome 16
TMEM216	Joubert syndrome 2 Meckel syndrome 2
TMEM231	Joubert syndrome 20 Meckel syndorome 11
TMEM237	Joubert syndrome 14
TMEM67	COACH syndrome Joubert syndrome Meckel syndrome Nephronophthisis 11 Bardet Biedl Syndrome 14, modifier of
TNXB	Vesicoureteral reflux 8
TRIM32	Bardet-Biedl syndrome 11
TRPC6	Glomerulosclerosis, focal segmental, 2
TSC1	Tuberous sclerosis-1
TSC2	Tuberous Sclerosis-2
TTC21B	Nephronophthisis 12
TTC8	Bardet-Biedl syndrome 8
UMOD	Autosomal Dominant Tubulointerstitial Disease- *UMOD*
VHL	von Hippel-Lindau syndrome
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2
VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1
WDPCP	?Bardet-Biedl syndrome 15
WDR19	Nephronophthisis 13 Senior-Loken syndrome
WDR35	Cranioectodermal dysplasia 2 Short-rib thoracic dysplasia 7 with or without polydactyly
WDR73	Galloway-Mowat syndrome
WNK4	Pseudohypoaldosteronism, type IIB
WNT4	SERKAL Syndrome
WT1	Nephrotic syndrome type 4 Wilms tumour Denys-Drash syndrome Frasier syndrome
XPNPEP3	Nephronophthisis-like nephropathy 1
ZMYND10	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,** Immuonglobin A**; LM,** light microscopy; **MPGN,** Membranoproliferative Glomerulonephritis **TI,** tubulointerstitial; **TIKD,** Tubulointersititал 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			
Patients with a Molecular Diagnosis													
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 nephonopthisis
6	6B	TIKD	Juvenile nephronopthisis	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	Nephronophthisis	Yes	9	50	Yes	Yes	Yes	Yes	No	TIKD	Nephonopthisis
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	Inhe-itance	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-opthisis 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	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	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	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	N	Y	Y	N	Likely Path.

	Boston Children's Hospital						complementation group I (609053)																					
15A	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.
15B	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.
15C	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.
16A	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.
17A	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.
18A	RCSI	*INF2*	M	14	het	AD	Glomerulo-sclerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
18B	RCSI	*INF2*	F	14	het	AD	Glomerulo-sclerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
18C	RCSI	*INF2*	M	14	het	AD	Glomerulos-clerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
18D	RCSI	*INF2*	M	14	het	AD	Glomerulo-sclerosis focal segmental, 5 (613237)	c.640C>T p.Arg214Cys	NM_001031714	1.0 Del D.C.	N	N	N	N	N	Y	N	N	N	Y	N	N	N	Y	Y	N	Likely path.
19A	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	N	Path.
20A	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.
20B	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.
21A	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	*INF2*	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	*WNK4*	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	*SLC3A1*	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	N	Y	Y	Y	N	Likely path