Supplement Table 1

Patient	Gender	Nucleotide change	Effect on protein	Reference	Score (Mutation Taster, Polyphen-2, SIFT)	Age at onset (CNS/months)	Response to CsA (time to CR/PR (months))	Kidney biopsy	Time to ESRD (months)	Renal outcome
NPHS1										
E2	m	c.515_517delCCA (H)	p.Thr172del	[1]		CNS	0	Interstitial sclerosis	70	RTx
E4	m	c.614_621delCACCCCGGinsTT (H)	p.Thr205_Arg207delinsIle	[1]		CNS	0	Not done		CKD
E5	f	c.2386G>A (h), c.2928-3C>G (h)	p.Gly796Arg, splice mutation	[2],[2]		CNS	0	FSGS	22	RTx
E6[e]	m	c.2618_2620delTCAinsCC (H), c.2552C>T (H)	p.Phe873Serfs*32, p.Ala851Val	[3],[4]		CNS	N	Finnish type	40	RTx
E7[e]	f	c.2618_2620delTCAinsCC (H), c.2552C>T (H)	p.Phe873Serfs*32, p.Ala851Val	[3],[4]		CNS	0	Not done		CKD
E9	f	c.1699T>A (H)	p.Cys567Ser	[2]		CNS	0	DMS	38	RTx
E10	m	c.3108A>G (H)	p.Pro1036Pro/splice mutation[a]	[5]		CNS	0	Not done		CKD
E11	m	c.1019C>A (H)	p.Pro340His	[6]		CNS	0	Not done		CKD
E12[e]	f	c.2618_2620delTCAinsCC (H), c.2552C>T (H)	p.Phe873Serfs*32, p.Ala851Val	[3],[4]		CNS	0	Not done	94	RTx
E17[e]	f	c.2618_2620delTCAinsCC (H), c.2552C>T (H)	p.Phe873Serfs*32, p.Ala851Val	[3],[4]		CNS	N	Not done	37	RTx
E19[e]	f	c.2618_2620delTCAinsCC (H), c.2552C>T (H)	p.Phe873Serfs*32, p.Ala851Val	[3] [4]		CNS	0	MCN	23	RTx
E20[e]	m	c.2618_2620delTCAinsCC (H), c.2552C>T (H)	p.Phe873Serfs*32, p.Ala851Val	[3],[4]		CNS	0	Not done	23	RTx
E21	m	c.563A>T (H), c.3286G>T (H)	p.Asn188Ile, p.Gly1096Cys	[7],[2]		CNS	0	Not done	62	RTx
H15	m	c.1248dupA (h), c.2606_2607dupCC (h)	p.Cys417Metfs*2, p.Asn870Profs*36	[8],[1]		CNS	N	Finnish type		normal GFR
H19	f	c.866G>A (H)	p.Trp289*	[6]		CNS	0	FSGS	0	RTx
H21	f	c.2540_2543delCTAA (h), c.2800C>T (h)	p.Thr847Argfs*57, p.Gln934*	[9],[10]		CNS	0	FSGS	34	RTx
H24[f]	m	c.2815+2T>A (H)	Splice mutation	[4]		CNS	0	Not done	No data	RTx
H25[g]	f	c.1379G>A (H)	p.Arg460Gln	[3]		CNS	0	MCN	No data	RTx
H26[g]	m	c.1379G>A (H)	p.Arg460Gln	[3]		CNS	0	Not done	No data	RTx
H28[f]	m	c.2815+2T>A (H)	Splice mutation	[4]		CNS	0	Not done	31	RTx
H42	f	c.3250dupG (H)	p.Val1084Glyfs*12	[11]		CNS	0	Not done	No data	RTx
HH16	m	c.928G>A (h), c.2816-3T>G (h)	p.Asp310Asn, splice mutation	[12],[13]		CNS	CR (2 months)	MCN		normal GFR

K18	f	c.866G>A (H)	p.Trp289*	[6]		CNS	0	Not done	30	RTx
K19	m	c.1738T>C (H)	p.Trp580Arg	Novel	Dc, pd, d	CNS	No data	Not done	No data	HD
K20	m	c.515_517delCCA (H)	p.Thr172del	[1]		CNS	No data	Not done	No data	HD
K21	m	c.515_517delCCA (H)	p.Thr172del	[1]		CNS	0	Finnish type	8	HD
K22[h]	m	c.3250dupG (H)	p.Val1084Glyfs*12	[11]		CNS	0	Finnish type	15	RTx
K23[h]	f	c.3250dupG (H)	p.Val1084Glyfs*12	[11]		CNS	0	Finnish type	24	RTx
K24	m	c.1868G>T (H)	p.Cys623Phe	[1]		CNS	0	Finnish type	12	RTx
MS1	f	c.2816-4_2822del11 (H)	Splice mutation	[2]		CNS	0	Finnish type	72	RTx
MS2	f	c.614_621delCACCCCGGinsTT (H)	p.Thr205_Arg207delinsIle	[1]		CNS	N	Not done	24	RTx
MS9	f	c.515_517delCCA (H)	p.Thr172del	[2]		CNS	0	DMS	25	RTx
MS10	f	c.1713delC (H)	p.Ser571Argfs*51	[2]		CNS	0	Finnish type	7	RTx
MS28	m	c.658T>G (h), c.2746G>T (h)	p.Ser220Ala, p.Ala916Ser	[14],[15]		168	N	FSGS	1	RTx
MS37	m	c.320C>A (h), c.1868G>T (h)	p.Ala107Glu, p.Cys623Phe	[2]		9	0	MCN	134	RTx
NPHS2										
E14	m	c.413G>A (H)	p.Arg138Gln	[16]		CNS	N	MCN	82	RTx
E15	f	c.871C>T (h), c.686G>A (h)	p.Arg291Trp, p.Arg229Gln	[16],[17]		CNS	0	MCN	255	RTx
E18	m	c.412C>T (h), c.413G>A (h)	p.Arg138*, p.Arg138Gln	[16] ,[16]		CNS	0	Not done	192	RTx
E22	f	c.503G>A (H)	p.Arg168His	[18]		31	0	IgA nephropathy	52	RTx
E28	m	c.413G>A (h), c.419delG (h)	p.Arg138Gln, p.Gly140Aspfs*41	[16],[16]		42	0	MC		normal GFR
E30	m	c.413G>A (h), c.868G>A (h)	p.Arg138Gln, p.Val290Met	[16],[19]		158	PR (6 months)	MC		CKD
E36	m	c.686G>A (h), c.890C>T (h)	p.Ala297Val, p.Arg229Gln	[17],[17]		141	0	FSGS		normal GFR
E38	m	c.467dupT (H)	p.Leu156Phefs*11	[20]		8	N	FSGS	104	RTx
E42	m	c.725C>T (H)	p.Ala242Val	[20]		40	N	FSGS		CKD
E51	m	c.413G>A (H)	p.Arg138Gln	[16]		10	0	MC	103	RTx
E56	f	c.413G>A (H)	p.Arg138Gln	[16]		9	0	Not done	65	RTx
H4	f	c.686G>A (h), c.1032delT (h)	p.Phe344Leufs*4, p.Arg229Gln	[20],[17]		12	No data	MCN		No data
H9	f	c.413G>A (H)	p.Arg138Gln	[16]		5	0	DMS	42	RTx
H20	m	c.413G>A (h), c.686G>A (h)	p.Arg138Gln, p.Arg229Gln[b]	[16],[17]		45	N	Mesangio-proliferative GN	58	RTx

H27	f	c.413G>A (H)	p.Arg138Gln	[16]		30	0	FSGS	77	RTx
I1	f	c.855_856delAA (H)	p.Arg286Thrfs*17	[16]		111	N	FSGS	12	RTx
HD3	f	c.413G>A (h), c.451+2T>A	p.Arg138Gln, splice mutation	[16],[22]		42	N	MCN	48	RTx
HD4	m	c.413G>A (h), c.503G>A (h)	p.Arg138Gln, p.Arg168His	[16],[18]		96	N	FSGS	38	RTx
HD5	m	c.873+2T>A (h), c.794+7A>G (h)	Splice mutation, splice mutation	[22]		156	N	FSGS	73	RTx
HD6	m	c.538G>A (H)	p.Val180Met	[16]		198	N	FSGS	42	RTx
HD7	f	c.686G>A (h), c.929A>C (h)	p.Glu310Ala, p.Arg229Gln	[23],[17]		51	N	FSGS		normal GFR
HD8	m	c.413G>A (h), c.873+2T>A (h)	p.Arg138Gln, splice mutation	[17],[22]		42	N	FSGS	57	RTx
HH4	f	c.413G>A (H)	p.Arg138Gln	[17]		65	PR	FSGS	25	RTx
HH27	f	c.413G>A (H)	p.Arg138Gln	[17]		CNS	N	FSGS	47	RTx
HH28	f	c.413G>A (H)	p.Arg138Gln	[17]		7	N	FSGS	18	RTx
K1	m	c.413G>A (h), c.467dupT (h)	p.Arg138Gln, p.Leu156Phefs*11	[17],[20]		15	0	FSGS	69	RTx
K2	f	c.413G>A (H)	p.Arg138Gln	[16]		74	N	FSGS	80	RTx
K3	f	c.413G>A (h), c.868G>A (h)	p.Arg138Gln, p.Val290Met	[16],[19]		72	No data	FSGS		normal GFR
K4	f	c.353C>T (H)	p.Pro118Leu	[18]		30	N	FSGS	15	RTx
K5	f	c.353C>T (H)	p.Pro118Leu	[18]		CNS	0	Not done	0	PD
K6	m	c.873+1G>T (H)	Splice mutation	Novel	Dc, na, na	48	N	FSGS		normal GFR
K7	m	c.413G>A (H)	p.Arg138Gln	[16]		45	0	FSGS	78	RTx
K8	m	c.871C>T (h), c.686G>A (h)	p.Arg229Gln, p.Val290Met	[17],[19]		241	0	MCN		normal GFR
M6	f	c.1150T>C (H)	p.384Glnext*7	[24]		96	N	FSGS		CKD
MS3[i]	f	c.413G>A (H)	p.Arg138Gln	[16]		CNS	0	FSGS	86	RTx
MS4[i]	f	c.413G>A (H)	p.Arg138Gln	[16]		CNS	N	FSGS	40	RTx
MS8	m	c.413G>A (H)	p.Arg138Gln	[16]		CNS	0	DMS	83	RTx
MS14	f	c.413G>A (h), c.868G>A (h)	p.Arg138Gln, p.Val290Met	[16],[19]		132	0	FSGS		normal GFR
MS17	m	c.413G>A (H)	p.Arg138Gln	[16]		5	0	Not done		CKD
MS19	m	c.467dupT (H)	p.Leu156Phefs*11	[20]		24	PR	FSGS	46	RTx
MS20	f	c.868G>A (H)	p.Val290Met	[19]		11	0	MCN		normal GFR
MS36	f	c.983A>G (h), c.686G>A (h)	p.Gln328Arg, p.Arg229Gln	[22]		106	0	FSGS		normal GFR
MS42	f	c.G686A (h), c.929A>T (h)	p.Glu310Val, p.Arg229Gln	[17],[17]		48	N	FSGS	102	RTx

WT1

E1	f	c.1384C>T (h)	p.Arg462Trp	[25]		CNS	0	Interstitial nephritis	8	RTx
E3	m	c.1384C>T (h)	p.Arg462Trp	[25]		CNS	0	DMS	15	PD
E8	m	c.1390G>A (h)	p.Asp464Asn	[25]		CNS	0	DMS	17	RTx
E13	m	c.1390G>A (h)	p.Asp464Asn	[25]		CNS	0	DMS	4	RTx
E16	m	c.1385G>A (h)	p.Arg462Gln	[26]		CNS	0	FSGS	4	RTx
E25	m	c.1384C>T (h)	p.Arg462Trp	[25]		7	0	DMS	0	RTx
E35	f	c.1384C>T (h)	p.Arg462Trp	[25]		50	0	FSGS	2	RTx
E47	m	c.1384C>T (h)	p.Arg462Trp	[25]		60	0	DMS	0	RTx
E48	f	c.1384C>T (h)	p.Arg462Trp	[25]		7	0	FSGS	0	RTx
H5	m	c.1384C>T (h)	p.Arg462Trp	[25]		16	0	FSGS		normal GFR
H18	f	c.1384C>T (h)	p.Arg462Trp	[25]		10	0	DMS	0	PD
H29	f	c.1394A>C (h)	p.His465Pro	[27]		17	0	Not done	46	RTx
H40	f	c.1288C>T (h)	p.Arg430*	[28]		147	0	FSGS	50	HD
H44	f	c.1432+5G>A (h)	Splice mutation	[29]		60	0	Not done[c]		normal GFR
HH31	m	c.1357T>C (h)	p.Cys453Arg	[30]		CNS	0	DMS	32	RTx
HH32	f	c.1390G>A (h)	p.Asp464Asn	[25]		24	0	DMS	No data	RTx
HH33	f	c.1339G>T (h)	p.Gly447Cys	[31]		CNS	0	DMS	3	RTx
HH34	f	c.1432+5G>A (h)	Splice mutation	[29]		108	N	MCN		CKD
HH35	f	c.1432+5G>A (h)	Splice mutation	[29]		120	0	MCN		normal GFR
K9	f	c.1385G>C (h)	p.Arg462Pro	[29]		6	0	DMS	1	RTx
K10	f	c.1391>G (h)	p.Asp464Gly	[25]		11	0	DMS	1	RTx
K11	f	c.1301G>A (h)	p.Arg434His	[25]		CNS	0	DMS	0	RTx
K12	m	c.1390G>C (h)	p.Asp464His	[32]		CNS	0	DMS		normal GFR
K13	f	c.1384C>T (h)	p.Arg462Trp	[25]		CNS	0	DMS	22	PD
K14	f	c.1372C>G (h)	p.Arg458Gly	Novel	Dc, pd, d	CNS	No data	DMS		normal GFR
K15	f	c.1432+5G>A (h)	Splice mutation	[29]		CNS	N	FSGS		normal GFR
K16	m	c.1339+2T>C (h)	Splice mutation	Novel	Dc, na, na	CNS	0	Not done		normal GFR
K17	f	c.1432+4C>T (h)	Splice mutation	[33]		48	0	FSGS		normal GFR
MS5	f	c.1366T>C (h)	p.Cys456Arg	[27]		CNS	0	DMS	21	RTx

MS16	f	c.1432+4C>T (h)	Splice mutation	[33]		48	0	FSGS	171	RTx
MS21	f	c.1394A>C (h)	p.His465Pro	[27]		15	PR (1.5 months)	FSGS	97	RTx
MS27	f	c.1432+4C>T (h)	Splice mutation	[33]		61	PR (8 months)	FSGS	133	RTx
MS29	f	c.1384C>T (h)	p.Arg462Trp	[25]		6	0	DMS	1	RTx
TRPC6										
E40	m	c.395T>C (h)	p.Met132Thr	[34]		104	N	FSGS	12	RTx
E45	m	c.253_264dupGCATACATGTTT (h)	p.Ala85_Phe88dup	Novel	Dc, na, na	97	N	FSGS	64	RTx
E50	m	c.2270G>A (h)	p.Gly757Asp	[2]		12	0	FSGS		CKD
H30	m	c.523C>T (h)	p.Arg175Trp	Novel	Dc, pd, d	14	N	FSGS	4	RTx
H45	m	c.2656G>T (h)	p.Glu886*	Novel	Dc, na, na	96	0	Not done	26	HD
INF2										
E33	f	c.787T>G (h)	p.Ser263Ala	Novel	Pm, b, t	15	N	FSGS	22	RTx
E55	m	c.658G>A (h)	p.Glu220Lys	[35]		156	0	FSGS		normal GFR
HD2	m	c.1448C>T (h)	p.Ser483Phe	Novel	Pm, b, t	48	N	FSGS		normal GFR
LAMB2										
MS6[j]	m	c.961T>C (h), c.4140C>A (h) and c.4177C>T (h)	p.Cys321Arg, p.Asn1380Lys and p.Lys1393Phe	[36], [36] and [36]		CNS	0	Not done	7	RTx
MS7[j]	m	c.961T>C (h), c.4140C>A (h) and c.4177C>T (h)	p.Cys321Arg, p.Asn1380Lys and p.Lys1393Phe	[36], [36] and [36]		CNS	0	Not done	32	RTx
MS32	m	c.2725dupA (h)	p.Ile909Asnfs*23	Novel	Dc, na, na	CNS	0	Not done	6	PD
PLCE1										
H22	f	c.4465T>C 8 (h), c.6377_6378delAA (h)	p.Cys1489Arg, p.Lys2126Argfs*17	Novel; novel	Dc, pd, d; Dc, na, na	31	0	DMS	2	RTx
K25	f	c. 4483C>T (H)	p.Arg1495*	Novel	Dc, na, na	CNS	No data	DMS	5	PD
ACTN4										
HH25	f	c.2020C>T (h)	p.Arg674Cys	Novel	Dc, pd, d	21	CR (2.5 months)	MCN		normal GFR

ARHGDIA									
K34	m	c.358C>T (H)	p.Arg120*	[37]	CNS	0	DMS	1	PD
LMX1B									
K26[k]	f	c.737G>A (h)	p.Arg246Gln	[38]	18	N	FSGS		normal GFR
K27[k]	m	c.737G>A (h)	p.Arg246Gln	[38]	108	N	FSGS	276	RTx
Novel[d]									
HH3	f	Novel mutation	Novel mutation		24	N	FSGS	6	RTx
MS11[l]	f	Novel mutation	Novel mutation		101	0	Not done		normal GFR
MS12[l]	f	Novel mutation	Novel mutation		31	0	FSGS	No data	RTx

Abbreviations

m=male; f=female; dc=disease causing; pd=probably damaging; d=damaging; na=not applicable; pm=polymorphism; b=benign; t=tolerated; 0=no CsA treatment; Y=patient responded to CsA treatment; N=patient did not respond to CsA treatment; CR=complete remission; PR=partial remission; MCN=minimal change nephropathy; DMS= diffuse mesangial sclerosis; FSGS =focal segmental glomerulosclerosis; GFR=glomerular filtration rate; CKD=chronic kidney disease; PD=peritoneal dialysis; HD=hemodialysis; RTX=renal transplantation

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Footnotes

[a]c.3108A nucleotide constitutes part of the GT 5' donor splice site recognition motif essential for the excision of IVS2; [b]the pathogenicity of p.Arg229Gln in this case is unclear as it is discussed to depend on the trans-associated mutation [21]; [c]nephroblastoma was identified in this patient; [d]these gene mutations have not been published so far.[e-l] familial cases indicated by footnotes.