

Supplementary data

Supplementary Table 1: Genes targeted by next-generation sequencing analysis

No.	Gene	Disease	No.	Gene	Disease
1	SLC12A1	Type 1 Bartter	61	MUC1	MCKD, ADTKD
2	KCNJ1	Type 2 Bartter	62	SEC61A1	Autosomal-dominant tubulo-interstitial and glomerulocystic kidney disease with anemia
3	CLCNKB	Type 3 Bartter	63	REN	ADTKD
4	BSND	Type 4a Bartter	64	EYA1	BOR
5	CLCNKA	Type 4b Bartter	65	SIX2	BOR
6	SLC12A3	Gitelman	66	CD2AP	FSGS
7	CASR	Hypocalcemia	67	NPHS1	FSGS/CNS
8	MAGED2	Type 5 Bartter	68	NPHS2	FSGS/CNS
9	CLDN10	Salt-losing tubulopathy	69	PLCE1 (NPHS3)	FSGS/CNS
10	CFTR	Cystic fibrosis	70	SMARCAL1	FSGS
11	CLCN5	Dent-1	71	LAMB2	FSGS
12	OCRL	Dent-2	72	SCARB2	FSGS
13	SLC26A3	Congenital chloride diarrhea	73	COQ2	FSGS
14	KCNJ10	EAST syndrome	74	COQ6	FSGS
15	CLDN16	Hypomagnesemia	75	ITGA3	FSGS
16	CLDN19	Hypomagnesemia	76	ITGB4	FSGS
17	FXYD2	Hypomagnesemia	77	GLEPP1 (PTPRO)	FSGS
18	EGF	Hypomagnesemia	78	MYO1E	FSGS
19	TRPM6	Hypomagnesemia	79	ARHGDIA	FSGS
20	KCNA1	Hypomagnesemia	80	ADCK4	FSGS
21	CNNM2	Hypomagnesemia	81	TTC21B	FSGS
22	HNF1B	Hypomagnesemia	82	NUP93	FSGS
23	PCBD1	Hypomagnesemia	83	NUP107	FSGS

24	CFH	aHUS	84	NUP205	FSGS
25	CFI	aHUS	85	NUP85	FSGS
26	MCP (CD46)	aHUS	86	NUP133	FSGS
27	C3	aHUS	87	NUP160	FSGS
28	CFB	aHUS	88	CRB2	FSGS
29	DGKE	aHUS FSGS	89	CUBN	FSGS
30	THBD	aHUS	90	EMP2	FSGS
31	CFHR1	aHUS	91	FAT1	FSGS
32	ADAMTS13	TTP	92	KANK1	FSGS
33	FN1	Fibronectin	93	KANK2	FSGS
34	SLC4A1	Distal RTA	94	KANK4	FSGS
35	ATP6V0A4	Distal RTA	95	PDSS2	FSGS
36	ATP6V1B1	Distal RTA	96	PTPRO	FSGS
37	SLC4A4	Proximal RTA	97	XPO5	FSGS
38	CA2	Mixed RTA	98	ACTN4	FSGS (AD)
39	EHHADH	Fanconi syndrome (OMIM #615605)	99	ANLN	FSGS (AD)
40	SLC34A1	Fanconi syndrome (OMIM #613388)	100	ARHGAP24	FSGS (AD)
41	SLC2A2	Fanconi-Bickel	101	INF2	FSGS (AD)
42	BCS1L	Fanconi syndrome	102	LMX1B	FSGS (AD)
43	CTNS	Cystinemia	103	MYH9	MYH9 nephropathy (AD)
44	NR3C2	PHA type 1	104	PAX2	FSGS (AD)
45	SCNN1A	PHA type 1	105	TRPC6	FSGS (AD)
46	SCNN1B	PHA type 1	106	WT1	FSGS/CNS
47	SCNN1G	PHA type 1	107	WDR73	FSGS, Galloway-Mowat
48	KLHL3	PHA type 1	108	MAGI2	Nephrotic syndrome
49	CUL3	PHA type 1	109	AVIL	Nephrotic syndrome
50	WNK1	PHA type 2	110	TNS2	Nephrotic syndrome
51	WNK4	PHA type 2	111	DLC1	Nephrotic

					syndrome
52	AQP2	Nephrogenic diabetes insipidus, type II	112	CDK20	Nephrotic syndrome
53	AVPR2	Nephrogenic diabetes insipidus, type I	113	ITSN1	Nephrotic syndrome
54	AVP	Central diabetes insipidus	114	ITSN2	Nephrotic syndrome
55	COL4A3	Alport syndrome	115	KIRREL1	Nephrotic syndrome
56	COL4A4	Alport syndrome	116	SGPL1	Congenital nephrotic syndrome
57	COL4A5	Alport syndrome	117	LMNA	FSGS(AD)
58	COL4A1	Hematuria, CAKUT	118	LAMA5	Nephrotic syndrome
59	GLA	Fabry disease	119	GAPVD1	Nephrotic syndrome
60	UMOD	MCKD, ADTKD	120	ANKFY1	SRNS
			121	GON7	Galloway-Mowat
			122	LAGE3	Galloway-Mowat
			123	OSGEP	Galloway-Mowat
			124	TPRKB	Galloway-Mowat
			125	TP53RK	Galloway-Mowat
			126	NUP133	Galloway-Mowat
			127	WDR4	Galloway-Mowat
			128	WDR73	Galloway-Mowat

Supplementary Table 2: The primers for PCR

		5'	3'
<i>LAMA5</i>	F	CCTGTAACTGCTACGGCCAC	
	R	GGTAGCAGCTTGGGAAGCCC	

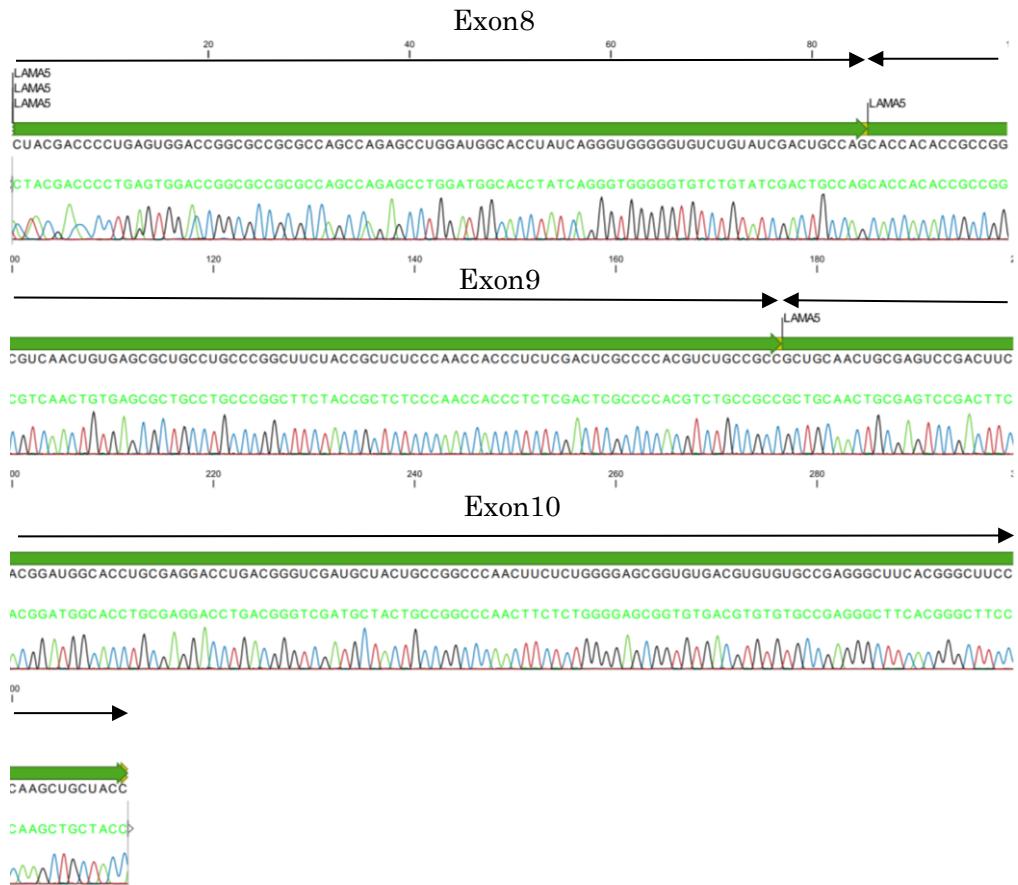
Supplementary Table 3: The primer sets for extension PCR

		5'	3'
Set1	F	tagcgttaaacttaagcttaccATGGAGACAGACACACTCCTGC	
	R	CCGATGGTGGTGATGATGGTATGATGGTATGCGGCTGGGCCGCG TCACCAGTGGAAC	
Set2	F	CCGCATACCATCATCACCATCACACCACATGGGAGGAGGCAGG CGCGGGCTTCAG	
	R	GTTGTACTCCAGGCCGCGGGACAGCGGTGGACTCTGC	

Supplementary Figure 1

Direct sequencing for transcript analysis

① The single band shown in the control (Co) and the upper band shown in patient (Pt) corresponded to exon 8, exon 9 and 10; the splicing pattern was normal.



② The lower band shown in Pt corresponded to exon 8 and exon 10; exon 9 skipping occurred.

