

Supplementary Table S1. Fifty-seven genes included in the targeted exome sequencing

Gene	OMIM number		Inheritance	Phenotypes	Reference
	Gene	Phenotype			
<i>ACTN4</i>	604638	603278	AD	FSGS1 , late onset SRNS	[1]
<i>ALG1</i>	605907	608540	AR	Congenital disorder of glycosylation, type Ik	[2]
<i>ANLN</i>	616027	616032	AD	FSGS8	[3]
<i>APOL1</i>	603743	612551	AR?	FSGS4 , susceptibility to	[4]
<i>ARHGAP24</i>	610586		AD	Adolescent-onset FSGS	[5]
<i>ARHGDIA</i>	601925	615244	AR	NPHS8 , Early onset-SRNS	[6]
<i>CAPN12</i>	608839		AR	FSGS	[7]
<i>CD151</i>	602243	609057	AR	Nephropathy with pretibial epidermolysis bullosa and deafness	[8]
<i>CD2AP</i>	604241	607832	AR/AD	FSGS3	[9]
<i>CFH</i>	134370	609814	AD/AR	Complement factor H deficiency with membranoproliferative glomerulonephritis type II	[10]
<i>COL4A3</i>	120070	203780	AR	Alport syndrome 2, autosomal recessive	[11]
		104200	AD	Alport syndrome 3, autosomal dominant	
		141200	AD	Benign familial hematuria,	
<i>COL4A4</i>	120131	203780	AR	Alport syndrome 2, autosomal recessive	[11]
		141200	AD	Benign familial hematuria,	
<i>COL4A5</i>	303630	301050	XL	Alport syndrome 1, X-linked	[12]
<i>COQ2</i>	609825	607426	AR	Primary coenzyme Q10 deficiency 1 with nephropathy	[13]
<i>COQ6</i>	614647	614650	AR	Primary coenzyme Q10 deficiency 6 with FSGS	[14]
<i>COQ8B</i>	615567	615573	AR	NPHS9	[15]
<i>CRB2</i>	609720	616220	AR	FSGS9	[16]
<i>CUBN</i>	602997	261100	AR	Megaloblastic anemia-1, Finnish type	[17]
<i>DGKE</i>	601440	615008	AR	NPHS7 ; aHUS7, susceptibility to	[18]
<i>E2F3</i>	600427		?	FSGS	[19]
<i>EMP2</i>	602334	615861	AR	NPHS10	[20]
<i>FN1</i>	135600	601894	AD	Glomerulopathy with fibronectin deposits 2	[21]
<i>FOXP3</i>	300292	304790	XL	IPEX syndrome	[22]
<i>GPC5</i>	602446		Risk allele?	FSGS	[23]
<i>INF2</i>	610982	613237	AD	FSGS5	[24]
<i>ITGA3</i>	605025	614748	AR	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	[25]
<i>ITGB4</i>	147557	226650	AR	Congenital FSGS with epidermolysis bullosa	[26]

<i>KANK1</i>	607704		AR	Nephrotic syndrome	[27]
<i>KANK2</i>	614610	617783	AR	NPHS16	[27]
<i>KANK4</i>	614612		AR	Nephrotic syndrome	[27]
<i>LAMA5</i>	601033		AR	Nephrotic syndrome	[28]
<i>LAMB2</i>	150325	609049	AR	Pierson syndrome	[29]
		614199		NPHS5 with or without ocular abnormalities	
<i>LMNA</i>	150330	151660	AD	Familial partial Lipodystrophy with FSGS	[30]
<i>LMX1B</i>	602575	161200	AD	Nail-patella syndrome	[31]
<i>MEFV</i>	608107	249100	AR	Familial Mediterranean fever with nephrotic syndrome due to AA amyloidosis	[32]
<i>NEIL1</i>	608844		AR	SRNS	[33]
<i>MMACHC</i>	609831	277400	AR	Nephrotic syndrome and thrombotic microangiopathy caused by cobalamin C deficiency	[34]
<i>MT-TL1</i>	590050	540000	Mt	MELAS syndrome with FSGS	[35]
<i>MT-TY</i>	590100		Mt	FSGS and dilated cardiomyopathy	[36]
<i>MYH9</i>	160775	155100	AD	Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss (Epstein syndrome, Fechtner syndrome)	[37]
<i>MYO1E</i>	601479	614131	AR	FSGS6	[38]
<i>NPHS1</i>	602716	256300	AR	NPHS1 , congenital nephrotic syndrome, Finnish type	[39]
<i>NPHS2</i>	604766	600995	AR	NPHS2	[40]
<i>NUP107</i>	607617	616730	AR	NPHS11	[41]
		618348		Galloway-Mowat syndrome 7	
<i>NXF5</i>	300319		XR	FSGS with co-segregating heart block disorder	[42]
<i>PAX2</i>	167409	616002	AD	FSGS7	[43]
		120330		Papillorenal syndrome	
<i>PDSS2</i>	610564	614652	AR	Primary coenzyme Q10 deficiency 3 with SRNS	[44]
<i>PLCE1</i>	608414	610725	AR	NPHS3	[45]
<i>PODXL</i>	602632		AD	FSGS	[46]
			AR	Congenital nephrotic syndrome with omphalocele	
<i>PTPRO</i>	600579	614196	AR	NPHS6	[47]
<i>SCARB2</i>	602257	254900	AR	Progressive myoclonic epilepsy 4, with or without renal failure	[48]
<i>SMARCAL1</i>	606622	242900	AR	Shimke immune-osseous dysplasia with SRNS	[49]
<i>SYNPO</i>	608155		AD	FSGS	[50]

<i>TRPC6</i>	603652	603965	AD	FSGS2	[51]
<i>TTC21B</i>	612014		AR	FSGS	[52]
<i>WDR73</i>	616144	251300	AR	Galloway-Mowat syndrome 1	[53]
<i>WT1</i>	607102	194080	AD	Denys-Drash syndrome	[54, 55]
		136680		Frasier syndrome	
		256370		NPHS4	

OMIM, Online Mendelian Inheritance in Man; AD, autosomal dominant; AR, autosomal recessive; XL, X-linked; Mt, mitochondrial; SRNS, steroid-resistant nephrotic syndrome; NPHS (nephrotic syndrome) and FSGS (focal segmental glomerulosclerosis) in bold indicate disease titles in OMIM.

Supplementary Table S2. Genotypes and phenotypes of patients with disease-causing mutations

Gene	Patient ID	Mutations	Age at Onset (Years)	Sex ^b	Family History	Mode of Onset	Kidney Biopsy	Renal Outcome	Time to ESRD (Years)
WT1 (REFSEQ: NM_024426.5)									
SRNS-20		c.1400G > A, p.R467Q	At birth	M	N	NS	ND	ESRD	0.1
SRNS-42		c.1136deIT, p.V379Dfs ^a	6.5	M	N	PU	FSGS	ESRD	2.7
SRNS-126		c.1231C > T, p.H411Y ^a	1.5	F	N	NS	FSGS	ESRD	7.3
SRNS-151		c.1315C > T, p.R439C	At birth	M	N	NS	ND	ESRD	0.0
SRNS-156		c.760C > A, p.P254T ^a	3.3	M	N	NS	FSGS	ESRD	6.1
SRNS-186		c.1363C > T, p.P455S	8.0	M	Y	NS	FSGS	Normal eGFR	NA
SRNS-222		c.1316G > A, p.R439H	At birth	M	N	NS	FSGS	ESRD	0.4
SRNS-223		c.1316G > A, p.R439H	At birth	F	N	NS	DMS	ESRD	0.0
SRNS-224		c.1316G > A, p.R439H	At birth	F	N	NS	FSGS	Death	NA
SRNS-225		c.1316G > A, p.R439H	At birth	F	N	NS	ND	ESRD	0.0
SRNS-226		c.1316G > C, p.R439P	At birth	M	NA	PU	ND	ESRD	1.8
SRNS-227		c.1315C > T, p.R439C	At birth	F	N	NS	ND	ESRD	0.0
SRNS-228		c.1324C > A, p.Q442K	1.0	F	N	ESRD	ND	ESRD	0.0
SRNS-229		c.1372T > A, p.C458S	At birth	M	N	NS	DMS	ESRD	1.1
SRNS-230		c.1399C > T, p.R467W	At birth	M	N	PU	DMS	ESRD	1.8
SRNS-231		c.1405G > T, p.D469Y	At birth	F	NA	NS	DMS	ESRD	0.0
SRNS-232		c.785 – 1G > C in intron 2 ^a	NA	M	NA	NA	ND	NA	NA
SRNS-233		c.1447 + 4C > T in intron 9	6.6	F	N	NS	FSGS	CKD	NA
SRNS-234		c.1447 + 4C > T in intron 9	2.6	F	N	NS	FSGS	Normal eGFR	NA
SRNS-235		c.1447 + 4C > T in intron 9	3.5	M	N	ESRD	ND	ESRD	0.0
SRNS-236		c.1447 + 4C > T in intron 9	6.8	M	N	NS	MesPGN	ESRD	15.4
SRNS-237		c.1447 + 4C > T in intron 9	At birth	F	N	CKD	ND	ESRD	0.7
SRNS-238		c.1447 + 5G > A in intron 9	At birth	M	N	PU	FSGS	ESRD	19.7
SRNS-239		c.1447 + 5G > A in intron 9	5.0	F	N	NS	FSGS	ESRD	4.2
SRNS-240		c.1447 + 5G > A in intron 9	6.8	F	N	PU	FSGS	ESRD	12.9
SRNS-241		c.1447 + 5G > A in intron 9	11.4	M	N	CKD	FSGS	ESRD	0.3
SRNS-242		c.1447 + 5G > A in intron 9	12.3	M	N	PU	FSGS	Normal eGFR	NA
SRNS-243		c.1419_1430del12, p.H474_T477del ^a	At birth	M	N	ESRD	ND	ESRD	0.0
SRNS-244		c.1381T > C, p.C461R	2.2	F	N	ESRD	MesPGN	ESRD	0.0
SRNS-245		c.1297T > C, p.C433R	At birth	F	N	ESRD	ND	ESRD	0.0

COQ6 (REFSEQ: NM_182476.2)

SRNS-61	c.686A > C, p.Q229P c.782C > T, p.P261L	1.1	M	N	PU	FSGS	Normal eGFR	NA
SRNS-103	c.124G > T, p.G42C ^a c. 782C > T, p.P261L	At birth	F	N	NS	FSGS	ESRD	0.4
SRNS-203	c.484C > T, p.R162* c.782C > T, p.P261L	9.1	M	N	PU	FSGS	ESRD	0.8
SRNS-251	c.189_191del3, p.K64del c.782C > T, p.P261L	3.9	M	N	NS	FSGS	ESRD	2.2
SRNS-252	c.189_191del3, p.K64del c.686A > C, p.Q229P	2.0	F	N	NS	FSGS	ESRD	1.1
SRNS-253	c.189_191del3, p.K64del c.782C > T, p.P261L	3.9	F	N	NS	FSGS	ESRD	0.1
SRNS-254	c.189_191del3, p.K64del c.782C > T, p.P261L	2.7	F	N	NS	FSGS	ESRD	1.9
SRNS-255	c.189_191del3, p.K64del c.782C > T, p.P261L	1.2	F	Y	NS	FSGS	ESRD	0.1
SRNS-256	c.189_191del3, p.K64del c.782C > T, p.P261L	3.1	M	N	NS	FSGS	ESRD	0.4
SRNS-257	c.686A > C, p.Q229P c.782C > T, p.P261L	At birth	M	N	NS	FSGS	ESRD	1.7
SRNS-258	c.189_191del3, p.K64del c.782C > T, p.P261L	1.1	M	N	NS	FSGS	ESRD	0.2
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NPHS1 (REFSEQ: NM_004646.3)								
SRNS-85	c.2156_2163del8, p.L719Pfs*4 c.2464G > A, p.V822M	At birth	F	Y	NS	MesPGN	Death	0.0
SRNS-206	c.2156_2163del8, p.L719Pfs*4 c.3250dupG, p.V1084Gfs*12	At birth	M	N	NS	MesPGN	ESRD	2.5
SRNS-207	c.2442C > G, p.Y814* c.1379G > A, p.R460Q	At birth	F	N	NS	ND	ESRD	3.0
SRNS-208	c.188A > G, p.Q63R c.1885G > T, p.E629*	At birth	M	N	NS	FSGS	ESRD	1.6
SRNS-209	c.3027C > G, p.Y1009* c.3478C > T, p.R1160*	At birth	F	N	NS	ND	ESRD	3.2
SRNS-210	c.2765C > A, p.A922D c.3287 – 11G > A in intron 24	At birth	M	N	NS	FSGS	CKD	NA
SRNS-211	c.2156_2163del8, p.L719Pfs*4	At birth	M	N	NS	ND	ESRD	4.7

SRNS-212	c.3478C > T, p.R1160* c.58 + 2T > C in intron 1 ^a c.1338delT, p.I466Mfs*16 ^a	At birth	F	Y	NS	MesPGN	ESRD	1.5
SRNS-213	c.526 + 1G > A in intron 4 c.1632_1634del3, p.545del	At birth	M	N	NS	ND	Normal eGFR	NA
SRNS-214	c.3213dupG, p.L1072Afs*24 ^a c.3478C > T, p.R1160*	At birth	M	N	NS	ND	Death	0.0
SRNS-215	c.139delG, p.A47Pfs*81 (homozygote)	At birth	M	N	NS	MesPGN	ESRD	1.8
<hr/> <i>NUP107</i> (REFSEQ: NM_020401.3)								
SRNS-71	c.934delT, p.Y312Tfs ^a c.2492A > C, p.D831A	4.8	M	N	PU	FSGS	ESRD	8.7
SRNS-259	c.2071C > T, p.Q691* c.2492A > C, p.D831A	4.3	M	Y	NS	FSGS	ESRD	4.2
SRNS-260	c.627_663dup37, p.L225Ffs*15 ^a c.2492A > C, p.D831A	3.8	F	N	PU	FSGS	ESRD	3.0
SRNS-261	c.1079_1083del5, p.E360Gfs*6 c.2492A > C, p.D831A	3.4	M	Y	NS	FSGS	ESRD	2.0
SRNS-262	c.1079_1083de5l, p.E360Gfs*6 c.2492A > C, p.D831A	2.4	M	N	PU	FSGS	ESRD	2.7
SRNS-263	c.1079_1083del5, p.E360Gfs*6 c.2492A > C, p.D831A	3.8	M	N	ESRD	ND	ESRD	0.0
SRNS-264	c.469G > T, p.D157Y c.2492A > C, p.D831A	10.9	F	Y	CKD	FSGS	ESRD	2.1
SRNS-265	c.1079_1083del5, p.E360Gfs*6 c.2492A > C, p.D831A	4.0	F	N	ESRD	ND	ESRD	0.0
SRNS-266	c.2492A > C, p.D831A c.1735 - 3T > G in intron 20	4.1	M	Y	PU	FSGS	ESRD	7.4
<hr/> <i>COQ8B</i> (REFSEQ: NM_024876.3)								
SRNS-25	c.759C > A, p.N253K (homozygote)	1.1	F	NA	NS	FSGS	ESRD	1.5
SRNS-35	c.737G > A, p.S246N c.532C > T, p.R178W	6.7	M	N	PU	FSGS	CKD	NA
SRNS-93	c.737G > A, p.S246N c.1548C > A, p.Y516 ^a	9.9	F	N	PU	FSGS	Normal eGFR	NA
SRNS-246	c.449G > A, p.R150Q c.759C > A, p.N253K	5.1	M	Y	PU	FSGS	ESRD	5.1
SRNS-247	c.737G > A, p.S246N	10.8	F	N	PU	FSGS	ESRD	2.0

SRNS-248	c.759C > A, p.N253K c.737G > A, p.S246N (homozygote)	9.2	F	N	PU	FSGS	ESRD	3.0
SRNS-249	c.737G > A, p.S246N c.1468C > T, p.R490C	6.9	F	N	PU	FSGS	ESRD	3.9
SRNS-250	c.737G > A, p.S246N (homozygote)	13.0	F	N	PU	FSGS	Normal eGFR	NA
<i>MYH9</i> (REFSEQ: NM_002473.5)								
SRNS-205	c.3494G > T, p.R1165L	16.8	F	Y	PU	ND	ESRD	17.5
SRNS-273	c. 2152C > T, p.R718W	1.3	M	N	NS	MesPGN	ESRD	5.3
SRNS-274	c.287C > T, p.S96L	20.0	M	N	PU	FSGS	ESRD	0.7
SRNS-275	c.287C > T, p.S96L	12.1	F	N	PU	ND	ESRD	8.3
SRNS-276	c.2104C > T, p.R702C	8.7	F	NA	PU	MesPGN	ESRD	7.8
SRNS-277	c.287C > T, p.S96L	12.4	M	NA	PU	MesPGN	ESRD	10.5
<i>INF2</i> (REFSEQ: NM_022489.3)								
SRNS-63	c.233T > G, p.L78R ^a	11.0	M	N	PU	FSGS	ESRD	6.7
SRNS-69	c.658G > A, p.E220K	11.1	F	N	PU	FSGS	ESRD	4.0
SRNS-268	c.658G > A, p.E220K	7.4	M	Y	PU	FSGS	ESRD	5.8
SRNS-269	c.658G > A, p.E220K	11.7	M	N	NS	FSGS	ESRD	5.5
SRNS-270	c.230T > C, p.L77P	9.2	F	N	NS	FSGS	ESRD	3.4
SRNS-271	c.529C > T, p.R177C	12.6	F	Y	PU	FSGS	Normal eGFR	NA
<i>PAX2</i> (REFSEQ: NM_003987.4)								
SRNS-26	c.76dupG, p.V26Gfs*28	5.3	M	N	PU	FSGS	ESRD	10.2
SRNS-31	c.563A > G, p.N188S ^a	3.4	M	N	NS	ND	Normal eGFR	NA
SRNS-32	c.222_225dup4, p.G76Dfs ^a	13.4	M	Y	PU	FSGS	CKD	NA
SRNS-95	c.74G > A, p.G25E ^a	7.2	F	N	PU	FSGS	ESRD	7.3
SRNS-191	c.419G > A, p.R140Q	7.8	M	N	PU	FSGS	Normal eGFR	NA
<i>NPHS2</i> (REFSEQ: NM_014625.3)								
SRNS-27	c.503G > A, p.R168H c.467dupT, p.L156Ffs*11	1.3	F	NA	NS	ND	ESRD	3.6
SRNS-47	c.412C > T, p.R138*	2.1	M	N	NS	FSGS	ESRD	4.9
SRNS-136	c.503G > A, p.R168H c.502C > T, p.R168C c.851C > T, p.A284V	6.9	M	N	PU	FSGS	CKD	NA
SRNS-216	c.358T > C, p.S120P c.503G > A, p.R168H	At birth	M	N	NS	FSGS	ESRD	8.4
<i>COL4A5</i> (REFSEQ: NM_000495.4)								
SRNS-49	c.834 + 1G > A in intron 14	10.0	F	Y	PU	FSGS	CKD	NA

SRNS-81	c.956G > A, p.G319D	10.1	M	Y	PU	FSGS	ESRD	10.1							
SRNS-87	c.4946delT, p.L1649Rfs*4 ^a	12.9	M	Y	PU	FSGS	ESRD	6.7							
SRNS-120	c.1165 + 1G > A in intron 19	3.8	M	N	NS	FSGS	ESRD	6.7							
SRNS-134	c.4082T > A, p.L1361* ^a	14.0	M	Y	PU	FSGS	ESRD	8.4							
SRNs-190	c.4532G > A, p.R1511H	12.8	M	N	PU	FSGS	CKD	NA							
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COL4A4 (REFSEQ: NM_000092.4)															
SRNS-53	c.1111delG, p.D371Tfs ^a	0.8	F	Y	PU	MesPGN	ESRD	18.9							
	c.1323_1340del18, p.P444_L449del														
	c.2630G > A, p.R877Q	3.6	M	N	NS	ND	Death	2.3							
	c.1046G > A, p.R349Q ^a	2.5	F	N	NS	FSGS	Normal eGFR	NA							
	c.2630G > A, p.R877Q	14.3	F	N	PU	FSGS	Normal eGFR	NA							
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MAFB (REFSEQ: NM_005461.4)															
SRNS-204	c.194G > T, p.S65I	9.8	M	Y	PU	ND	Normal eGFR	NA							
	c.183C > A, p.S61R	12.5	F	N	PU	FSGS	CKD	NA							
	c.211C > G, p.P71A	4.4	M	N	PU	FSGS	ESRD	0.6							
	c.212C > T, p.P71L	1.2	M	N	PU	ND	Normal eGFR	NA							
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LAMB2 (REFSEQ: NM_002292.3)															
SRNS-217	c.1503_1504delAT, p.C502*	0.7	F	N	NS	FSGS	ESRD	10.8							
	c.4267delT, p.C1423Vfs*29														
SRNS-218	c.2283-2286del4, p.S762Rfs*29	At birth	F	N	NS	FSGS	CKD	NA							
	c.536C > T, p.S179F														
SRNS-219	c.474delT, p.A159Pfs*33 ^a	At birth	F	N	NS	ND	ESRD	0.1							
	c.1328_1329del2, p.H443Rfs*11 ^a														
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WDR19 (REFSEQ: NM_025132.3)															
SRNS-289	c.3533G > A, p.R1178Q	9.6	M	N	PU	FSGS	ESRD	1.4							
	c.3703G > A, p.E1235K														
SRNS-290	c.3533G > A, p.R1178Q	6.2	F	Y	PU	MesPGN	ESRD	3.0							
	c.3703G > A, p.E1235K														
SRNS-291	c.1853T > C, p.L618P	At birth	M	N	CKD	ND	ESRD	0.3							
	c.3533G > A, p.R1178Q														
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SMARCAL1 (REFSEQ: NM_014140.3)															
SRNS-144	c.1682G > A, p.R561H	6.0	M	N	NS	FSGS	ESRD	3.4							
	c.1851 + 1G > T in intron 9 ^a														
SRNS-287	c.1411dupA, p.I471Nfs ^a	5.5	F	N	NS	FSGS	ESRD	1.5							
	c.1484A > C, p.Q495P ^a														
SRNS-288	c.1484A > C, p.Q495P ^a	3.5	M	N	PU	FSGS	ESRD	2.1							

		c.1851 + 1G > T in intron 9 ^a						
MT-TL1 (REFSEQ: NC_012920)								
SRNS-284	mtDNA3243A > G	18.9	F	Y	PU	DMS	CKD	NA
SRNS-285	mtDNA3243A > G	11.8	F	Y	PU	FSGS	ESRD	6.0
SRNS-286	mtDNA3243A > G	9.8	F	N	PU	FSGS	ESRD	5.3
FOXP3 (REFSEQ: NM_014009.3)								
SRNS-283	c.736 - 2A > G in intron 7 ^a	3.4	M	N	NS	MNP	Normal eGFR	NA
ACTN4 (REFSEQ: NM_004924.5)								
SRNS-267	c.785C > T, p.S262F	3.5	M	Y	NS	FSGS	ESRD	1.2
LMX1B (REFSEQ: NM_002316.3)								
SRNS-279	c. 668G > A, p.R223Q	2.1	F	N	NS	FSGS	ESRD	1.86
ANLN (REFSEQ: NM_018685.4)								
SRNS-65	c.2305A > T, p.L769* ^a	7.7	M	N	PU	FSGS	Normal eGFR	NA
TRPC6 (REFSEQ: NM_004621.5)								
SRNS-37	c.523C > G, p.R175G ^a	8.5	F	N	PU	FSGS	ESRD	2.3
COL4A3 (REFSEQ: NM_000091.4)								
SRNS-199	c.4793T > G, p.L1598R	0.5	F	N	NS	DMS	ESRD	0.9
TP53RK (REFSEQ: NM_033550.3)								
SRNS-221	c.194A > T, p.K65M (homozygote)	At birth	F	NA	NA	ND	Death	0.0
DGKE (REFSEQ: NM_003647.2)								
SRNS-272	c.501C > G, p.C167W c.610dupA, p.T204Nfs*4	0.5	M	N	PU	FSGS	CKD	NA
LCAT (REFSEQ: NM_000229.1)								
SRNS-278	c.794_801del8, p.E265Afs*18 c.931delT, p.F311Lfs*99 ^a	9.6	M	Y	PU	FSGS	Normal eGFR	NA
COQ2 (REFSEQ: NM_015697.7)								
SRNS-168	c.392A > G, p.D131G ^a c.518G > A, p.R173H ^a	At birth	F	N	NS	FSGS	ESRD	0.3
PODXL (REFSEQ: NM_005397.3)								
SRNS-220	c.3G > T, p.M1? c.926G > A, p.W309*	At birth	M	Y	NS	ND	ESRD	0.0

^aNovel mutations

^bSex of patients with WT1 mutations and sex reversal followed by their karyotypes

NA, not available; ND, not done; NS, nephrotic syndrome; PU, proteinuria; CKD, chronic kidney disease; ESRD, end-stage renal disease; eGFR, estimated glomerular filtration rate; FSGS, focal segmental glomerulosclerosis; DMS, diffuse mesangial sclerosis; MesPGN, mesangial proliferative glomerulonephritis; MNP, membranous nephropathy; M, male; F, female; Y, yes; N, no

Supplementary Table S3. Variants of unknown significance detected by targeted exome sequencing.

Patient ID	Gene	Mutation
SRNS-6	<i>MYH9</i>	c.2440C>T, p.R814W
SRNS-8	<i>CUBN</i>	c.1811C>T, p.P604L; c.6490C>T, p.P2164S
SRNS-18	<i>ACTN4</i>	c.283C>T, p.R95W
SRNS-33	<i>ARHGAP24</i>	c.1865G>A, p.R622H
SRNS-48	<i>CAPN12</i>	c.281G>A, p.R94H; c.862G>A, p.V288M
SRNS-50	<i>MYH9</i>	c.2440C>T, p.R814W
SRNS-94	<i>LMX1B</i>	c.20C>G, p.P7R
SRNS-99	<i>CAPN12</i>	c.281G>A, p.R94H; c.862G>A, p.V288M
SRNS-105	<i>PTPRO</i>	c.1279G>C, p.E427Q; p.3530C>T, p.S1177F
SRNS-108	<i>PAX2</i>	c.1127A>C, p.Q376P
SRNS-116	<i>CD151</i>	c.173T>G, p.L58R; c.685G>A, p.G229S
SRNS-117	<i>KANK1</i>	c.971A>G, p.Y324C; c.1282G>A, p.G428R
SRNS-129	<i>ACTN4</i>	c.449C>T, p.T150M
SRNS-143	<i>LAMB2</i>	c.1327C>T, p.H443Y; c. 2945G>A, p.R982Q
SRNS-146	<i>COL4A3</i>	c.1637C>T, p.P546L; c.3394C>T, p.P1132S
SRNS-153	<i>ARHGAP24</i>	c.365G>A, p.R122Q
SRNS-154	<i>MYH9</i>	c.5188C>T, p.R1730C
SRNS-163	<i>TTC21B</i>	c.2569G>A, p.A857T; c.2866A>G, p.M956V
SRNS-169	<i>COL4A4</i>	c.2045A>G, p.D682G
SRNS-171	<i>TRPC6</i>	c.2066A>C, p.K689T
SRNS-178	<i>PAX2</i>	c.1127A>C, p.Q376P
SRNS-180	<i>ACTN4</i>	c.1279G>A, p.A427T
SRNS-183	<i>COL4A3</i>	c.3476G>A, p.R1159H

Supplementary Table S4. Comparison between patients with autosomal dominant (AD) mutations and patients with autosomal recessive (AR) mutations

	AD mutation (+) patients (n=59)	AR mutation (+) patients (n=58)	Ratio of AD mutations to AR mutations	P value
Age at onset				
Congenital	11 (18.6%)	16 (27.6%)	11/27 (40.7%)	
Infantile	5 (8.5%)	6 (10.3%)	5/11 (45.5%)	
1 – 6 years	16 (27.1%)	22 (38.0%)	16/38 (42.1%)	
7 – 12 years	16 (27.1%)	12 (20.7%)	16/28 (57.1%)	
≥13 years	10 (16.9%)	1 (1.7%)	10/11 (90.9%)	0.005 ^b
Data unavailable	1 (1.7%)	1 (1.7%)		
Sex ratio ^a (male:female)	35:24	30:28		
Family history (+)	7 (11.9%)	11 (19.0%)	7/18 (38.9%)	
Mode of onset				
Nephrotic syndrome	26 (44.1%)	33 (56.9%)	26/59 (44.1%)	0.161 ^c
Proteinuria	25 (42.4%)	20 (34.5%)	25/45 (55.6%)	
CKD/ESRD	7 (11.9%)	4 (6.9%)	7/11 (63.6%)	
Data unavailable	1 (1.7%)	1 (1.7%)		
Steroid responsiveness				
Responder	0	0	0	
Nonresponder	33 (55.9%)	31 (53.4%)	33/64 (51.6%)	
Initial nonresponder	32	31	32/63 (50.8%)	
Late nonresponder	1	0	1/1	
No treatment	25 (42.4%)	26 (44.8%)	25/51 (49.0%)	
Data unavailable	1 (1.7%)	1 (1.7%)		
Renal biopsy				
FSGS	32 (54.2%)	38 (65.5%)	32/70 (45.7%)	
Minimal change	0	0	0	
Others	10 (16.9%)	8 (13.8%)	10/18 (55.5%)	
Not done	16 (27.1%)	11 (19.0%)	16/27 (59.3%)	
Data unavailable	1 (1.7%)	1 (1.7%)		
Renal function at the last FU				
Normal eGFR	11 (18.6%)	5 (8.6%)	11/16 (68.8%)	0.114 ^d
CKD stage 2–4	3 (5.1%)	5 (8.6%)	3/8 (37.5%)	
ESRD	44 (74.6%)	47 (81.0%)	44/91 (48.4%)	
Data unavailable	1 (1.7%)	1 (1.7%)		
Duration (years) from onset to ESRD (n=91)	4.0±5.0	2.8±3.4	3.6 ± 4.3	0.184

^aSex of patients with WT1 mutations and sex reversal followed by their karyotypes.

^bDisease onset ≥13 years group (10/11, 90.9%) versus other onset age groups (48/104, 46.2%)

^cNephrotic syndrome group (26/59, 44.1%) versus other mode of onset groups (32/56, 57.1%)

^dNormal eGFR group (11/16, 68.8%) versus other eGFR groups (47/99, 47.5%)

FSGS, focal segmental glomerulosclerosis; eGFR, estimated glomerular filtration rate; CKD, chronic kidney disease; ESRD, end-stage renal disease; FU follow-up.