Supplemental Material

Supplemental Figure 1: Renal biopsy findings in individuals with SRNS in whom the causative mutation was identified in 21 SRNS-causing genes (27 examined) in 615 affected individuals from 526 families.

Supplemental Figure 2. Distribution for age of onset of NS in 102 individuals with homozygous mutations in *NPHS2*.

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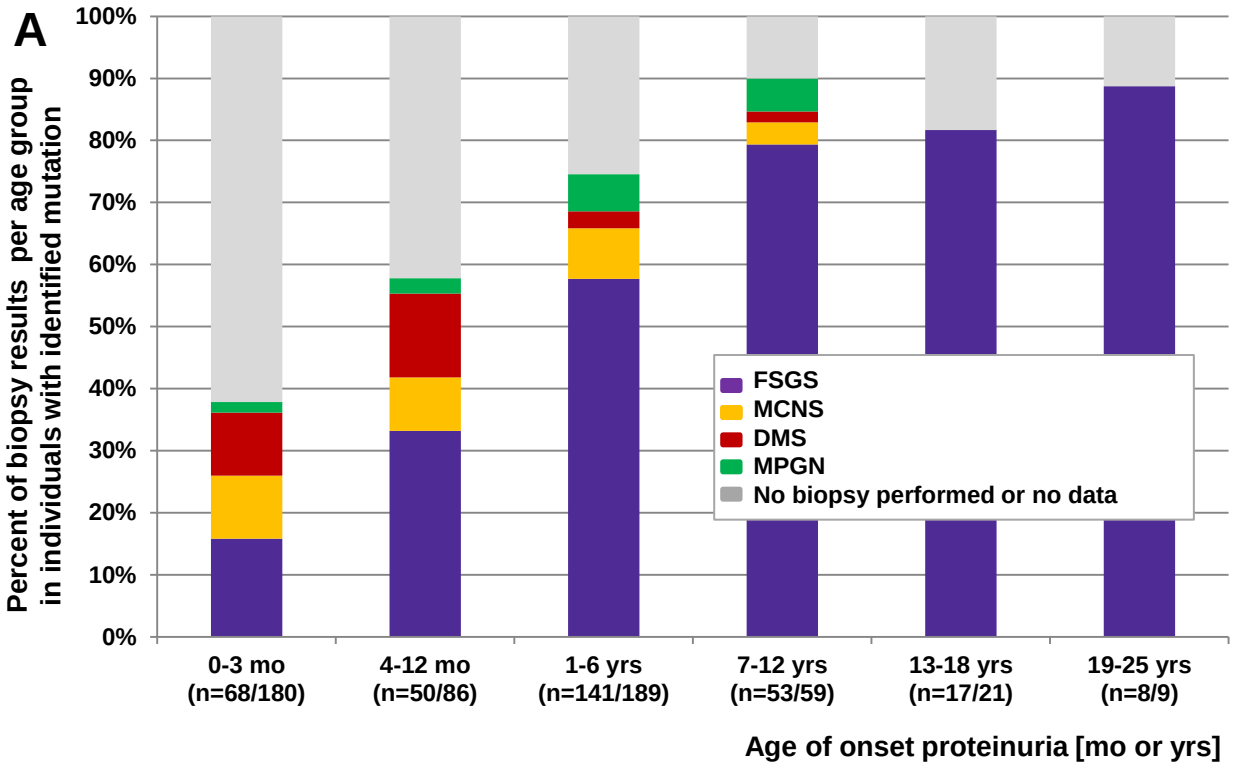
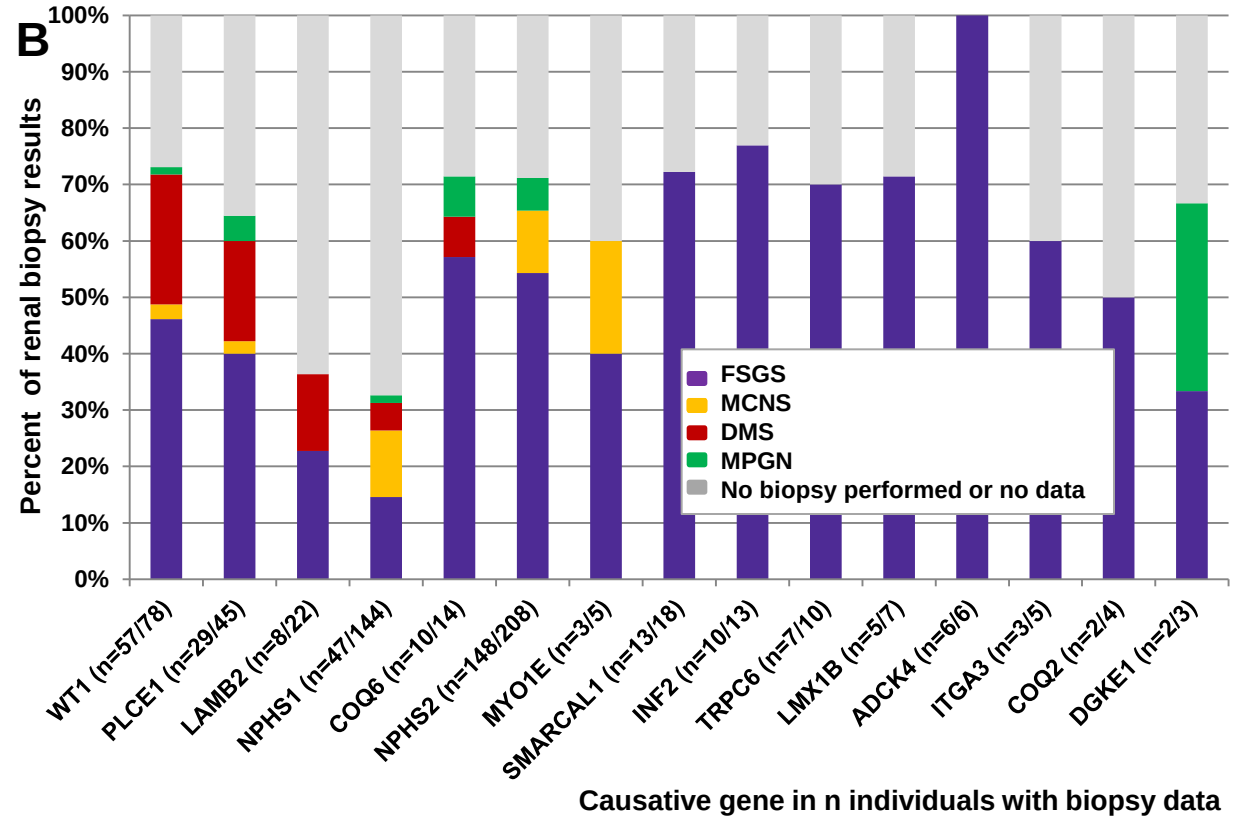
Supplemental Table 4. Genotypes and phenotypes of mutations in *WT1* detected by Sanger sequencing and high-throughput sequencing in 35 out of 1,783 families with SRNS.

Supplemental Table 5. Genotypes and phenotypes of mutations in *TRPC6, ARHGAP24 and INF2* detected by Sanger sequencing and high- throughput sequencing in 17 out of 1,783 families with SRNS.

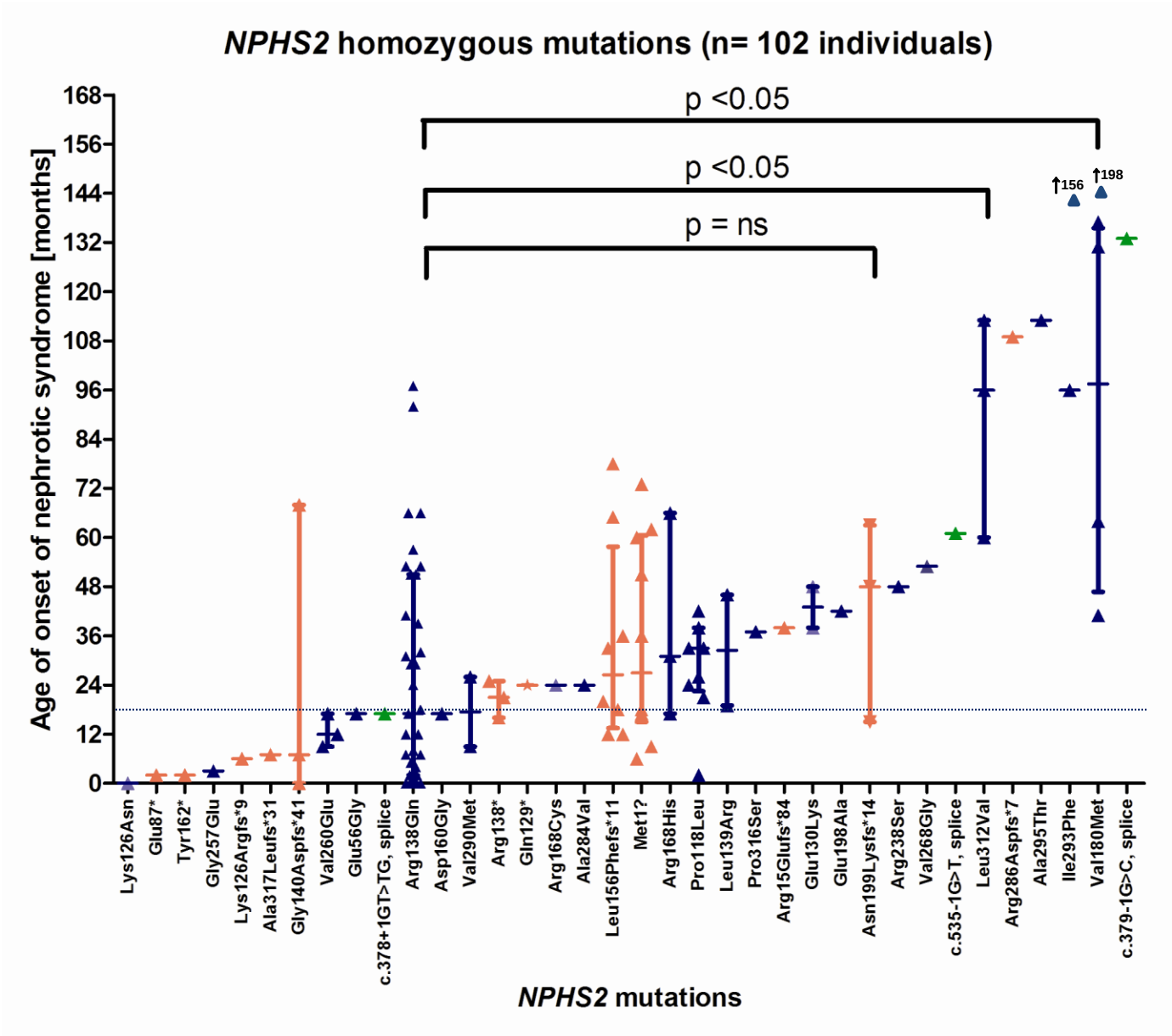




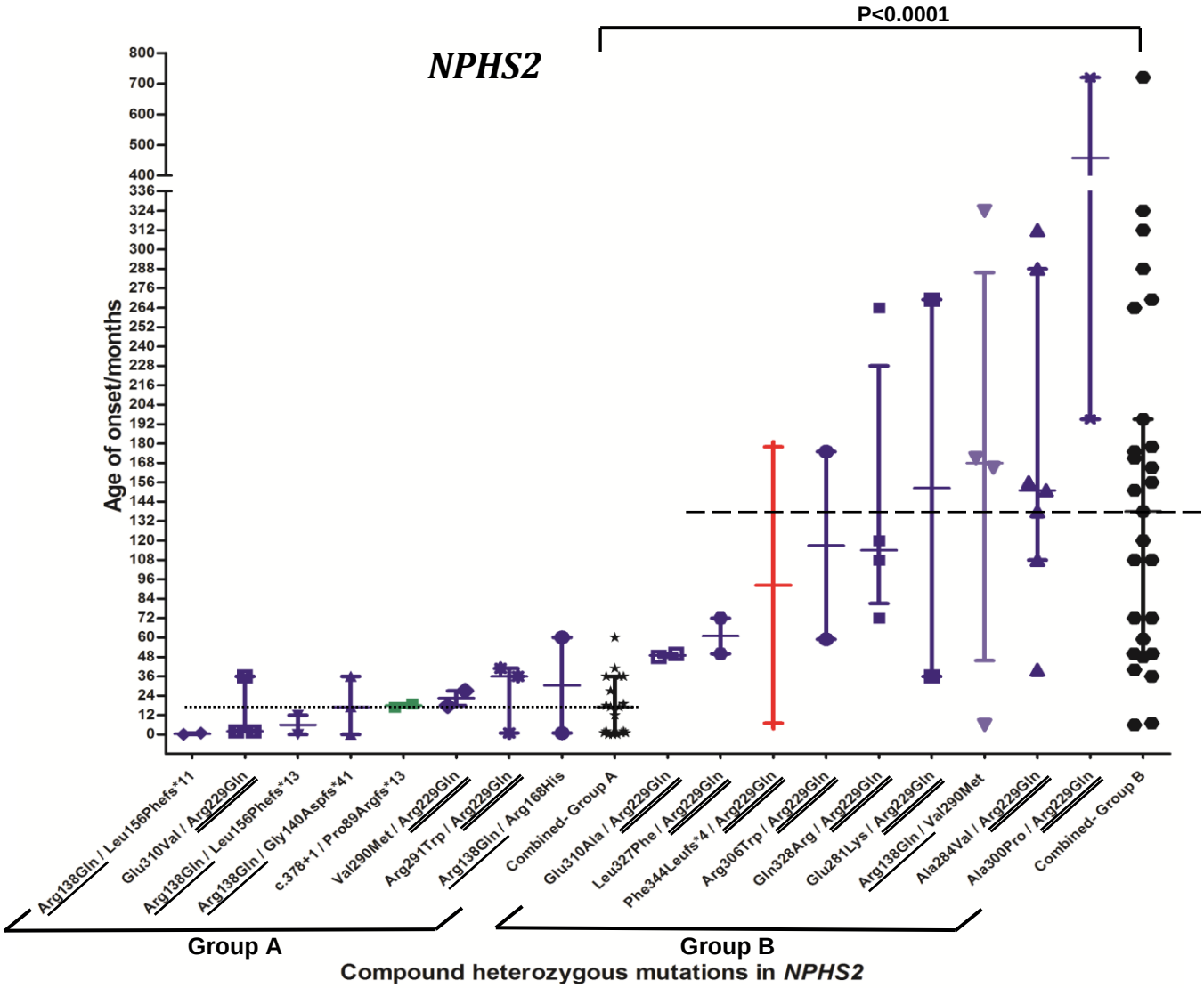
Supplemental Figure 1

Supplemental Figure 1: Renal biopsy findings in individuals with SRNS in whom the causative mutation was identified in 21 SRNS-causing genes (27 examined) in 615 affected individuals from 526 families.

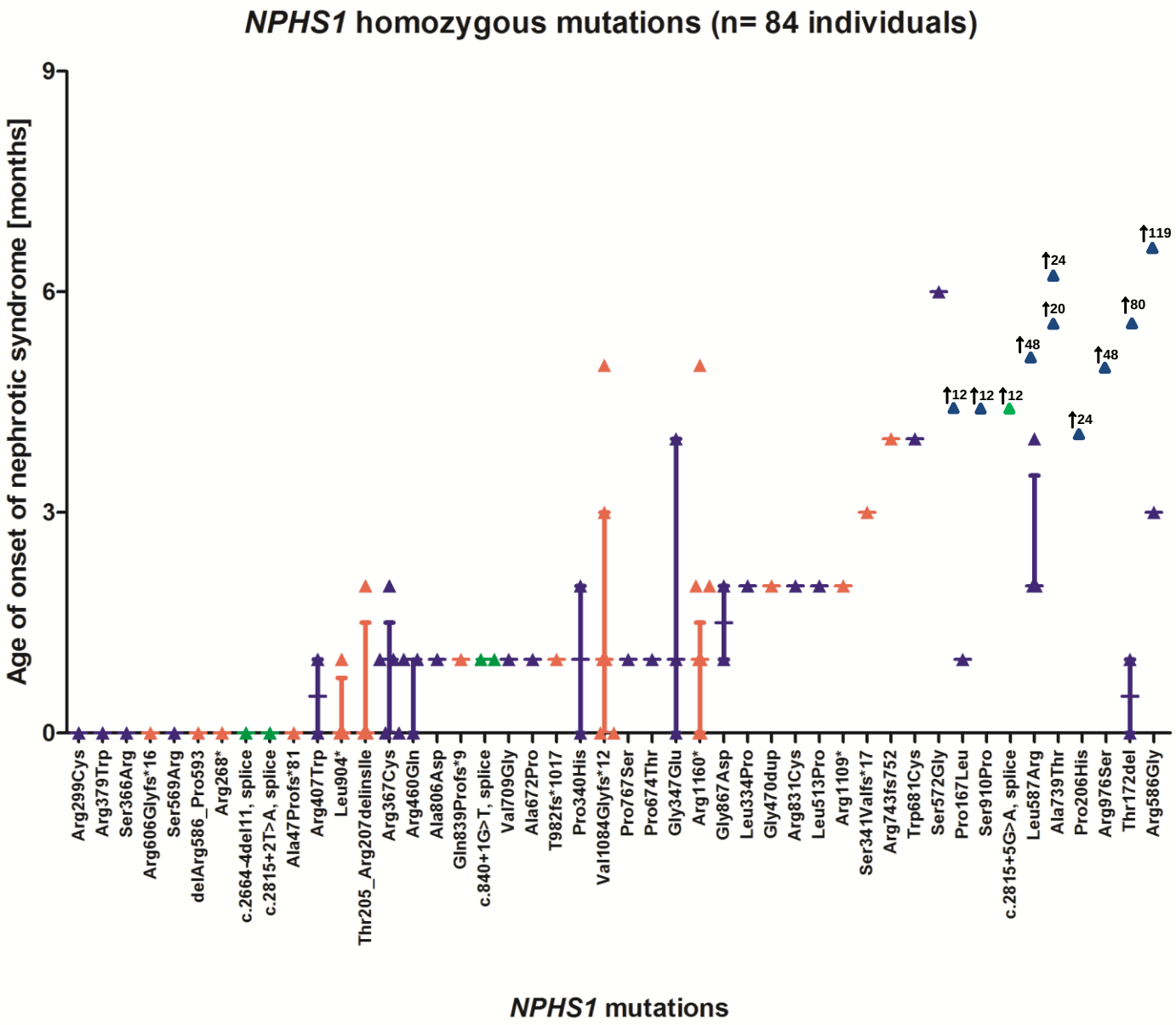
(A) In 337 of the displayed 544 individuals a renal biopsy was performed. Renal biopsy findings are represented by age groups. Note that renal biopsy was less frequently performed in infants and that the developmental phenotype of DMS is more frequent in infants. Not shown are 14 patients with an age of onset later than 25 years, 30 individuals, in whom no age of onset of proteinuria was available, and 27 patients with inconclusive biopsy results or Wilms tumor. **(B)** Renal biopsy findings by gene in 350 of 582 individuals with identified mutation. 20 patients with an inconclusive biopsy result and 7 patients with Wilms tumor are not included, as well as 6 individuals with disease-causing mutations in the genes *ARHGDIA, ARHGAP24, CUBN, CFH* and *ITGB4,* in whom two or less individuals with biopsy result were available for each gene.



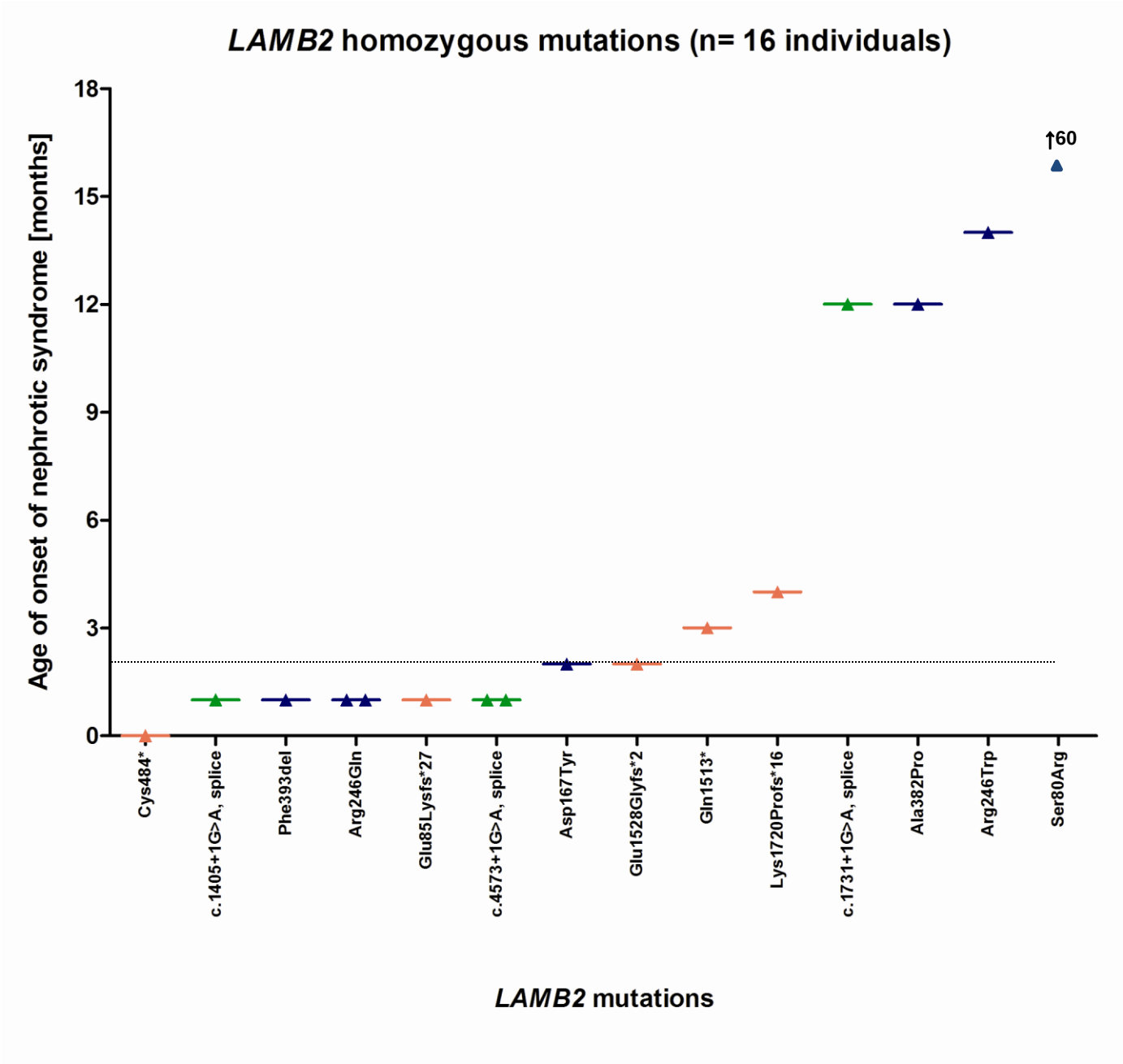
Supplemental Figure 2. Distribution for age of onset of NS in 102 individuals with homozygous mutations in *NPHS2*. Homozygous *NPHS2* mutations are shown for 36 different alleles. The x-axis indicates the mutations sorted by median age of onset. The y-axis indicates the age of onset of SRNS. The median age of onset was significantly different between the European founder mutation Arg138Gln (17 months) *versus* Leu312Val (96 months, p<0.05), or Arg138Gln (17 months) *versus* Val180Met (97.5 months, p<0.05). Symbols are colored red, green, or blue if the allele is truncating, splice or missense, respectively. Numbers with arrows represent two outliers for age of onset. Blue dotted line reflects the median age of onset for the European founder mutation Arg138Gln. p values are from two-tailed Student's t-test; mo= months.



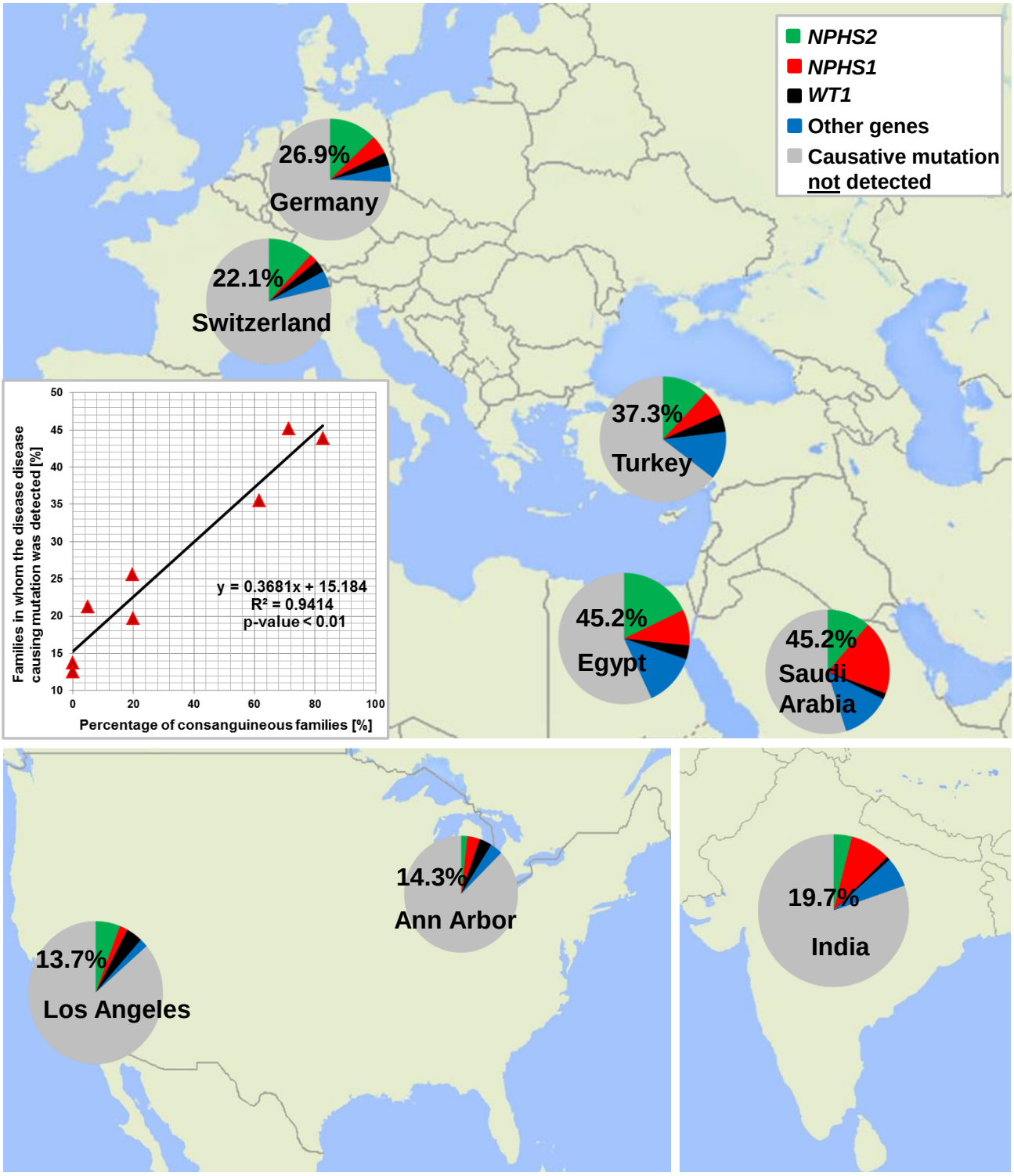
Supplemental Figure 3. Distribution for age of onset of NS in individuals with compound heterozygous mutations in *NPHS2*. Fourty-six individuals had compound heterozygous *NPHS2* mutations represented by 17 different alleles. Since the severity of phenotype (age of onset) of individuals with compound heterozygous mutations is decided by the "milder" mutation, the 'X-axis' descriptor shows first the allele thought to cause later onset (before "/") and the second the allele thought to cause earlier onset (after "/") as derived from data with homozygous mutations **(Appendix Figure 2).** The mutations were sorted from left to right by increasing median age of onset. For early onset (<4 yrs) the data was grouped as "Combined-Group A". For late onset (>4 yrs) the data was grouped as "Combined-Group B". For group A (n = 19), median age of onset was 17 months (dotted line) and for group B (n = 27), median was 138 months (dashed line). Age of onset was statistically significant between the two groups ($p < 0.0001$). P values are from two-tailed student's t-test. Symbols are colored red, green, or blue if the severity-deciding allele is truncating, splice, or missense, respectively. The European founder mutation Arg138Gln is underlined, it occurs more frequently (four of five times) in the early onset group A. The allele Arg229Gln is double underlined. It occurs more frequently (eight of 11 times) in the group with later onset (group B). Arg229Gln occurs with second alleles that were recently described to be compatible with causing SRNS in combination with Arg229Gln.



Supplemental Figure 4. Distribution for age of onset of SRNS in 82 individuals with homozygous mutations in *NPHS1*. Homozygous *NPHS1* mutations are shown for 46 different alleles. The x-axis indicates the mutations sorted by median age of onset. The y-axis indicates the age of onset of SRNS. Symbols are colored red, green, or blue if the allele is truncating, splice, delins or missense, respectively. Numbers with arrows represent eight outliers for age of onset. Note that for 76% (35/46) of homozygous *NPHS1* mutations there was congenital nephrotic syndrome, i.e. age of onset <3 month; mo= months.

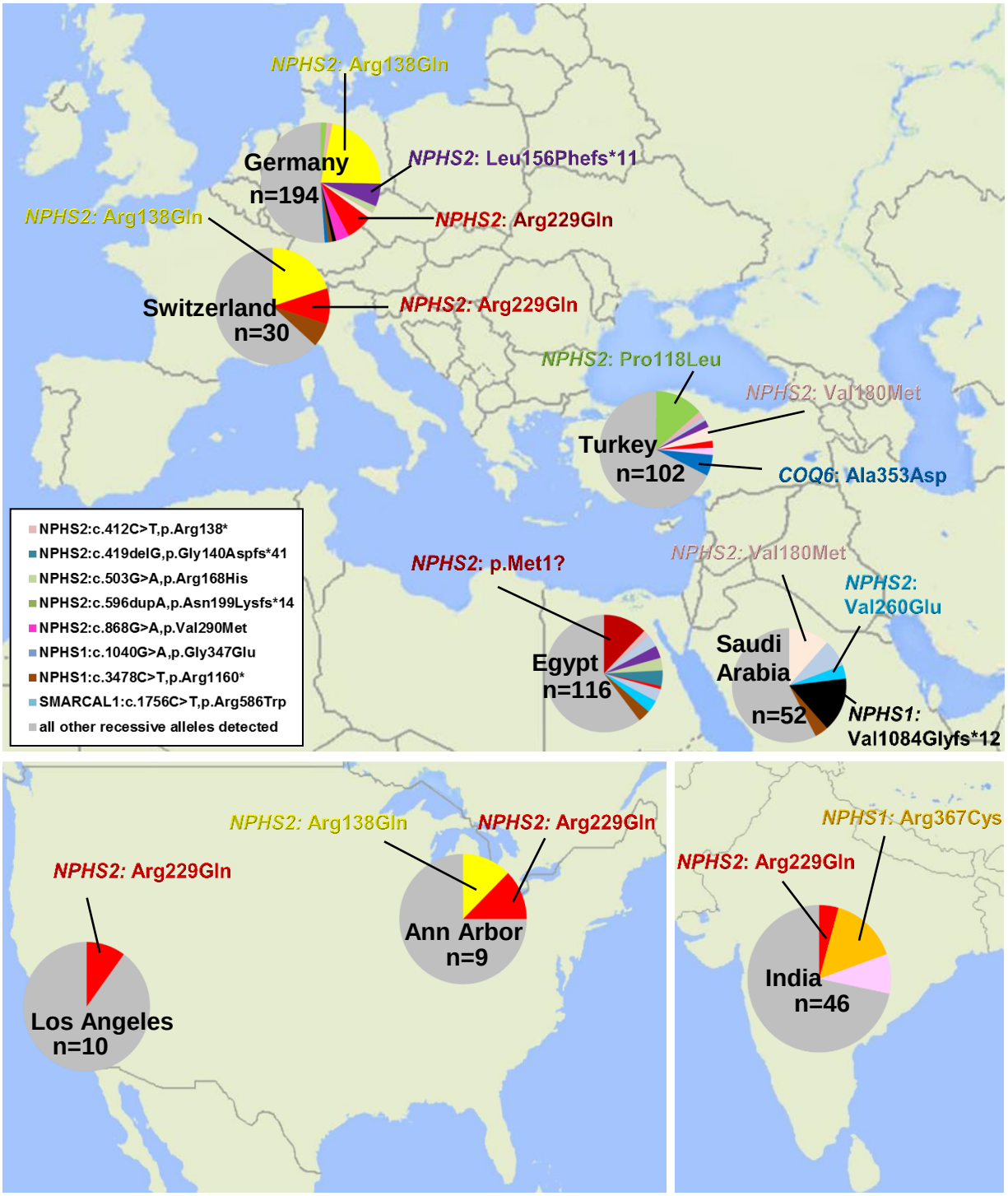


Supplemental Figure 5. Distribution for age of onset of SRNS in 16 individuals with homozygous mutations in *LAMB2*. Homozygous *LAMB2* mutations are shown for 14 different alleles. The x-axis indicates the mutations sorted by the median age of onset. The y-axis indicates the age of onset of SRNS. Symbols are colored red, green, or blue if the allele is truncating, splice, or missense, respectively. The number with arrows represents one outlier (Ser80Arg, age of onset 60 months). Note that all individuals with homozygous N-terminal truncating mutations and the one delins mutation (Cys484*, Phe393del, Glu85Lysfs*27) manifest before two month of life (dashed horizontal line), whereas all three individuals with homozygous C-terminal truncating mutations (Glu1528Glyfs*2, Gln1513*, and Arg1719Argfs*19) manifest after ≥ two months of life; mo= months.



Supplemental Figure 6. Percentage of genetic findings in SRNS families from the 8 largest contributing centers. We obtained samples from 1,783 SRNS families worldwide and detected the disease-causing mutation in 526 families (29.5%). For 8 centers we detected the disease causing mutations in the following fractions: (families, in whom we detected the causative mutation/total families examined from this center): Saudi-Arabia (45.2%, 28/62), Egypt (45.2%, 66/146), Turkey (37.3%, 62/169), Germany (26.9%, 123/457), Switzerland (22.1 %, 21/94), India (19.7%, 25/127,), Ann Arbor (14.3%, 8/56), and Los Angeles (13.7%, 7/51).

Inset: The detection rate of the disease-causing mutations strongly correlates with the rate of consanguinity between the different centers (R^2=0.9414).



Supplemental Figure 7. Most frequent 18 SRNS-causing alleles in 126 families from the 8 largest contributing centers. In the 8 largest centers the causative mutation was detected in 559 families for a recessive gene. Pie chart segment represent percent frequency for alleles per center if allele was detected ≥ 5 times in the 8 centers (n=126 families and 224 alleles). We detected the different founders alleles with the following frequencies (allele detected per center/total alleles in this center): The European founder *NPHS2*: Arg138Gln in Germany (22.2%, 43/194), Switzerland (20%, 6/30) and Ann Arbor (22.2 %, 2/9); the Turkish founder *NPHS2*: Pro118Leu in Turkey (13.7%,14/102), Germany (1.5%, 3/194); the Egyptian founders *NPHS2*: Met1? in Egypt (12%,14/116) and *NPHS2*: Asn199Lysfs*14 in Egypt (4.3%, 5/116); the North African founder *NPHS2*: Val180Met in Saudi Arabia (11.5%, 6/52) and the Indian founder *NPHS1*: Arg367Cys in India (15.2 %,14/116). The most prominent allele are called out in the map, for the less prominent alleles see color legend.

Supplemental Table 1: Characteristics of an international cohort of 1,783 families with 2,016 affected individuals.

	Gender (n= 2,016 individuals)				Parental Consanguinity (n= 1,783 families)				Age of onset (n= 2,016 individuals) [months]
	male	female	No Data	Total	Yes	No	No Data	Total	Median (range)
All individuals	1,067 (52.9%)	932 (46.2%)	17 (0.8%)	2,016	372 (20.9%)	1,308 (73.4%)	103 (5.7%)	1,783	41 (0- 756)
No detection of the disease causing-gene	776 (55.0%)	623 (44.2%)	12 (0.8%)	1,411	188 (15.0%)	981 (78.0%)	88 (7.0%)	1,257	53 (0- 756)
Causative gene detected	301 (48.9%)	309 (50.3%)	5 (0.8%)	615	184 (35.0%)	327 (62.2%)	15 (2.9%)	526	18 (0- 720)
Dominant	49 (41.2%)	70 (58.8%)	0 (0%)	119	7 (6.5%)	98 (91.6%)	2 (1.9%)	107	38 (0- 444)
Recessive	251 (50.7%)	239 (48.3%)	5 (1.0%)	495	177 (42.2%)	229 (54.6%)	13 (3.2%)	419	12 (0- 720)

Supplemental Table 2. Twenty six SRNS-causing genes that were investigated using microfluidic multiplex PCR and NGS.

Gene	Protein	Accession #	Chro mo-some	Total Exon count	Coding exon count	Amplicon count	Amino acid conservation to species	First publication
Autosomal recessive								
ADCK4	AarF domain containing kinase 4	NM_024876.3	19	15	14	14	*S. cerevisiae*	[1]
ARHGDIA	Rho GDP dissociation inhibitor (GDI) alpha	NM_001185078.1	17	6	5	6	*C. elegans*	[2]
CD2AP	CD2-associated protein	NM_012120.2	6	18	18	18	*C. elegans*	[3]
CFH	Complement factor H	NM_000186.3	1	22	22	25	*D. rerio*	[4]
COQ2	Coenzyme Q2 4-hydroxybenzoate polyprenyltransferase	NM_015697.7	4	7	7	9	*C. elegans*	[5]
COQ6	Coenzyme Q6 monooxygenase	NM_182476.2	14	12	12	12	*C. elegans*	[6]
CUBN	Cubilin (intrinsic factor-cobalamin receptor)	NM_001081.3	10	67	67	73	*C. elegans*	[7]
DGKE	Diacylglycerol kinase, epsilon	NM_003647.2	17	12	11	17	*D. rerio*	[8]
ITGA3	Integrin, alpha 3 (antigen CD49C, alpha 3 subunit of VLA-3 receptor)	NM_005501.2	17	25	25	26	*D. rerio*	[9]
ITGB4	Integrin, beta 4	NM_000213.3	17	40	39	47	*C. elegans*	[10]
LAMB2	Laminin, β2	NM_002292.3	3	32	32	35	*C. elegans*	[11]
MEFV	Pyrin	NM_000243.2	16	10	10	17	*M. musculus*	[12]
MYO1E	Homo sapiens myosin IE (MYO1E)	NM_004998.3	15	28	28	28	*C. elegans*	[13]
NEIL1	Nei endonuclease VIII-like 1	NM_001256552.1	15	10	10	10	*D. rerio*	[14]
NPHS1	Nephrin	NM_004646.3	19	29	29	28	*C. elegans*	[15]
NPHS2	Podocin	NM_014625.2	1	8	8	10	*C. elegans*	[16]
PDSS2	Prenyl (decaprenyl) diphosphate synthase, subunit 2	NM_020381.3	6	8	8	10	*D. melanogaster*	[17]
PLCE1	Phospholipase C, epsilon 1	NM_016341.3	10	33	31	46	*C. elegans*	[18]
PTPRO	Protein tyrosine phosphatase, receptor type, O	NM_030667.2	12	27	26	28	*D. rerio*	[19]
SCARB2	Scavenger receptor class B, member 2	NM_005506.3	4	12	12	12	*C. elegans*	[20]
SMARCAL1	SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a-like 1	NM_014140.3	2	18	16	20	*C. elegans*	[21]
Autosomal dominant								
ACTN4	Actinin, alpha 4	NM_004924.4	19	21	21	21	*C. elegans*	[22]
ARHGAP24	Rho GTPase activating protein 24	NM_001025616.2	4	10	9	19	*D. rerio*	[23]
INF2	Inverted formin, FH2 and WH2 domain containing	NM_022489.3	14	23	21	28	*D. rerio*	[24]
LMX1B	Homo sapiens LIM homeobox transcription factor 1, beta (LMX1B)	NM_001174147.1	9	8	8	9	*C. elegans*	[25]
TRPC6	Transient receptor potential cation channel, subfamily C, member 6	NM_004621.5	11	13	13	20	*D. rerio*	[26]
WT1	Wilms tumor 1	NM_024426.4	11	10	10	13	*S. cerevisiae*	[27]

Supplemental Table 3. Genotypes and phenotypes of mutations in 14 recessive SRNS-causing genes (*NPHS2, NPHS1, PLCE1, SMARCAL1, COQ2, COQ6, DGKE, MYO1E, LAMB2, CUBN, ITGA3, ITGB4, CFH* and *PDSS2)* detected by Sanger sequencing and high-throughput sequencing in 280 out of 1,783 families with SRNS.[#]

Family-Individual	Nucleotide change	Amino Acid change	Exon (Zygosity, Segregation)	Poly-phen	SIFT	Muta-tion-taster	Amino acid conservation to species	EVS allele frequencies in EA	Gen-der	Ethnic origin	Parental Consan-guinity	Age of onset	Biopsy	Therapy and response	Extrarenal Manifesta-tions	Refer-ence	Me-thod
NPHS2																	
A3231-22	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	GG=0/GA=0/AA=4300	F	African-Arabic	Y	5 yrs 2 mo	MPGN	SR	ND	novel	SSe
A1248	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	ND	FSGS (4 yrs 9 mo)	SR, CP-NR, CsA-NR	ND	novel	SSe
A3531-21	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	4 yrs 3 mo	FSGS	SR	ND	novel	SSe
A5099-28	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	5 yrs	FSGS	SR, CP-NR	SS	novel	SSe
A4653	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	-		Arabic	N					novel	SSe
-22									F			1 yr 6 mo	MPGN (1 yr 6 mo), FSGS (2 yrs 1 mo)	SR, CP-NR, CsA-NR, MMF-NR	ND		
-38									F			9 mo	Mesangioproliferative GN	SR, CP-NR	ND		
A1759-22	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	3 yrs	ND	Steroid-PR	ND	novel	SSe
A4340	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	-		Arabic	Y					novel	SSe
-21									F			1 yr 5 mo	FSGS (2 yrs)	SR	ND		
-22									M			6 mo	ND	ND			
A4384-21	c.1A>T	p.Met1?	1 (hom)	NA	NA	NA	NA	-	F	Arabic	N	1 yr 5 mo	MesP	SR	ND	novel	SSe
A3649-21	c.42delG	p.Arg15Glufs*84	1 (hom)	NA	NA	NA	NA	-	M	Indian	Y	3 yrs 2 mo	FSGS	SR	ND	novel	SSe
A2303-21	c.59C>T	p.Pro20Leu	1 (hom)	0.012	del	DC	*H. sapiens*	AA=0/AG=14/GG=3694	F	Turkish	Y	ND	MPGN (5 yrs 5 mo)	SR	SS	[16]	SSe
A4382-22	c.104_126dupGCCGCGGGCGCCAGGAGGCTGGG	p.Pro43Alafs*64	1 (het)	NA	NA	NA	NA	-	M	Arabic	Y	1 yr 9 mo	MCNS (2 yrs 1 mo)	SR	ND	novel	SSe
	c.596dupA	p.Asn199Lysfs*14	5 (het)	NA	NA	NA	NA	-								novel	SSe
A5196-21	c.167A>G	p.Glu56Gly	1 (hom)	0.016	del	DC	*M. musculus*	-	F	African-Arabic	N	1 yr 5 mo	FSGS	SR, CsA-NR, Tac-NR	ND	novel	SSe
A1867	c.259G>T	p.Glu87*	1 (hom)	NA	NA	NA	NA	-	F	Turkish	ND	ND	ND	ND	ND	[28]	SSe
A941-21	c.259G>T	p.Glu87*	1 (het)	NA	NA	NA	NA	-	F	Slavic	N	2 yrs 1 mo	MCNS (2 yrs 2 mo)	SR, CsA-PR	UTA	[28]	SSe
	c.379-2A>C	Splice	2 (het)	NA	NA	NA	NA	-								novel	SSe
1312	c.264_265delGC	p.Pro89Argfs*13	1 (het)	NA	NA	NA	NA	-	F	ND	N	1 yr 7 mo	FSGS (1 yr 8 mo)	SR, CsA-ND	PAS	novel	SSe
	c.378+1G>A	Splice	2 (het)	NA	NA	NA	NA	-								novel	SSe
A2079	c.264_265delGC	p.Pro89Argfs*13	1 (het)	NA	NA	NA	NA	-	F	European	N	1 yr 5 mo	FSGS (1 yr 5 mo)	SR, CsA-R	ND	novel	SSe
	c.378+1GT>TG	Splice	2 (het)	NA	NA	NA	NA	-								novel	SSe
A1840	c.353C>T	p.Pro118Leu	2 (hom)	0.925	del	DC	*C. elegans*	-	F	Turkish	Y	ND	FSGS (3 yrs)	SR, CsA-NR	ND	[29]	SSe
A2926-21	c.353C>T	p.Pro118Leu	2 (hom)	0.925	del	DC	*C. elegans*	-	F	Turkish	Y	3 yrs 2 mo	FSGS (3 yrs 6 mo)	SR, CsA-NR	ND	[29]	SSe
A3312-21	c.353C>T	p.Pro118Leu	2 (hom)	0.925	del	DC	*C. elegans*	-	F	Turkish	Y	2 yrs 2 mo	ND	SR, CP-NR	ND	[29]	SSe
A3318-31	c.353C>T	p.Pro118Leu	2 (hom)	0.925	del	DC	*C. elegans*	-	M	Turkish	Y	2 mo	ND	ND	ND	[29]	SSe
A167-21	c.353C>T	p.Pro118Leu	2 (hom)	0.925	del	DC	*C. elegans*	-	F	ND	Y	2 yrs 9 mo	MCNS, FSGS, AN (2 yrs 5 mo)	SR, CsA-PR, ESRD 3 yrs 9 mo	ND	[29]	SSe
A3320	c.353C>T	p.Pro118Leu	2 (hom)	0.925	del	DC	*C. elegans*	-		Turkish	Y					[29]	SSe
-21									F			2 yrs 9 mo	FSGS (2 yrs 10 mo)	SR	ND		
-22									M			1 yr 9 mo	FSGS (1 yr 10 mo)	ND	ND		
A2359-21	c.377delA	p.Lys126Argfs*9	2 (hom)	NA	NA	NA	NA	-	M	Asian	Y	6 mo	ND	SR	ND	novel	SSe
A2161	c.378+1GT>TG	splice	2 (hom)	NA	NA	NA	NA	-	M	Indian	Y	1 yr 5 mo	MCNS (1 yr 5 mo)	SR, CsA-NR	ND	novel	SSe
A1832	c.379-1G>C	splice	2 (hom)	NA	NA	NA	NA	-	M	Turkish	N	11 yr 1 mo	FSGS (11 yr 1 mo)	SR, CP-NR, CsA-NR	ND	novel	SSe
A2269-23	c.385C>T	p.Gln129*	3 (hom)	NA	NA	NA	NA	-	F	Saudi Arabia	Y	2 yrs	ND	SR	ND	[30]	SSe
A3916-22	c.385C>T	p.Gln129*	3 (het)	NA	NA	NA	NA	-	M	Hispanic	N	2 yrs	ND	SR	UTA	[30]	SSe
	c.739-1G>T	Splice	5 (het)	NA	NA	NA	NA	-								novel	SSe

A4284-21	c.388G>A	p.Glu130Lys	3 (hom)	1	del	DC	*C. elegans*	-	F	Arabic	Y	3 yrs 2 mo	MPGN	ND	ND	novel	Flui
A4462-22	c.388G>A	p.Glu130Lys	3 (hom)	1	del	DC	*C. elegans*	-	F	Arabic	Y	ND	MPGN (4 yrs)	SR, CP-NR, MMF	ND	novel	SSe
A679	c.397delA	p.Arg133Glufs*2	3 (het)	NA	NA	NA	NA	-		ND	N					novel	Flui
	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292								[16]	
-21												ND	ND	ND	ND		
-22												ND	ND	ND	ND		
A287-21	c.397-398delAG	p.Arg133Serfs*33	3 (het)	NA	NA	NA	NA	-	M	Russian	N	1 yr	MCNS (1 yr)	SR	N	novel	SSe
	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292								[16]	SSe
A1961	c.397delA	p.Arg133Glufs*2	3 (het)	NA	NA	NA	NA	-	M	White	N	3 yrs 2 mo	FSGS (3 yrs 2 mo)	SR	ND	[31]	SSe
	c.855_856delAA	p.Arg286Thrfs*17	7 (het)	NA	NA	NA	NA	-								[16]	SSe
A354-21	c.412C>T	p.Arg138*	3 (hom)	NA	NA	NA	NA	-	M	ND	N	ND	MCNS (4 yrs 8 mo)	SR, CsA-CR	AS, CF	[16]	SSe
A139-21	c.412C>T	p.Arg138*	3 (hom)	NA	NA	NA	NA	-	M	ND	N	2 yrs 1 mo	FSGS (2 yrs 4 mo)	SR, CP-NR	ND	[16]	SSe
A4461-26	c.412C>T	p.Arg138*	3 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	ND	FSGS (2 yrs)	SR, CP-NR	ND	[16]	SSe
A3116-21	c.412C>T	p.Arg138*	3 (het)	NA	NA	NA	NA	-		Asian	N	11 yr 6 mo	FSGS (12 yrs 5 mo)	CsA	ND	[16]	SSe
	c.538G>A	p.Val180Met	5 (het)	0.577	del	DC	*D. rerio*	TT=0/TC=1/CC=4299								[16]	SSe
236	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		European	N				ND	[16]	SSe
-21									F			2 yrs 5 mo	FSGS (2 yrs 9 mo)	SR			
-22									M			2 mo	FSGS	SR, ESRD 9 yrs 3 mo			
348	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		European	N					[16]	SSe
-21									M			3 yrs 3 mo	FSGS (5 yrs 11 mo)	SR	SS		
-22									M			2 yrs	ND	ND	SS		
													FSGS				
398	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		European	N					[16]	SSe
-21									F			2 yrs 7 mo	FSGS (3 yrs)	SR, CsA-R	ID		
-23									F			8 mo	FSGS (3 yrs)	SR, CsA-R, ESRD 4 yrs	ND		
460	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		European	N					[16]	SSe
-21									M			ND	MCNS (5 yrs 5 mo) MCNS (2 yrs 7 mo)	SR, CP-NR	GR		
-22									F			2 yrs 5 mo		ESRD 3 yrs	SS		
A646	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	European	N	4 yrs 9 mo	ND	SR	ND	[16]	SSe
747	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		European	N					[16]	SSe
-21									M			3 yrs 5 mo	FSGS (5 yrs 6 mo)	SR, CsA-R	ND		
-22									M			2 yrs 8 mo	FSGS (3 yrs 6 mo)	SR, Plasma exchange, ESRD 3 yrs 10 mo	ND		
975	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		ND	N					[16]	SSe
-21									M			4 yrs 3 mo	FSGS	SR, CP-NR	ND		
-22									M			1 yr 6 mo	FSGS	ND	recurrent pancreatitis		
1305	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	ND	N	1 yr	FSGS	SR	ND	[16]	SSe
A126	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	ND	N	7 mo	FSGS (7 mo)	SR	SS, VUR	[16]	SSe
A5193-21	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	Hispanic	N	2 yrs 5 mo	FSGS (4 yrs 5 mo)	SR, CsA-NR	SS	[16]	SSe
A825-21	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	Slavic	N	2 mo	FSGS (2 mo)	SR	ND	[16]	SSe
A1686	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	European	N	4 yrs 5 mo	FSGS	SR	ND	[16]	SSe
A1730	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		German	N					[16]	SSe
-21									F			CNS	MCNS (3 mo)	SR	ND		
-22									F			CNS	ND	ESRD 3 yrs	ND		
A2821-21	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	European	N	ND	ND	ND	SS	[16]	SSe
A3025-21	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	White	N	8 yrs 1 mo	FSGS (8 yrs 6 mo)	SR	ND	[16]	SSe
A1992	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	European	N	ND	FSGS (7 yrs 8 mo)	ND	ND	[16]	Flui
A4002-21	c.413G>A	p.Arg138Gln	3 (hom)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	White	N	4 yrs 5 mo	FSGS	SR, CsA-PR	ND	[16]	SSe
515	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		European	N					[16]	SSe
	c.419delG	p.Gly140Aspfs*41	3 (het)	NA	NA	NA	NA	-								[16]	SSe
-21									M			ND	ND	SR	ID		
-22									M			birth	ND	ND	None		
A266-21	c.413G>A	p.Arg138Gln	3 (het)	0.999	Del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	ND	N	1 yr 5 mo	ND	SR, CsA-PR	MC, ID, SS,	[16]	SSe
	c.419delG	p.Gly140Aspfs*41	3 (het)	NA	NA	NA	NA	-					ND		FD	[16]	SSe

355	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		European	N					[16]	SSe
	c.467dupT	p.Leu156Phefs*11	4 (het)	NA	NA	NA	NA	-								[32]	SSe
-21									M			ND	FSGS (4 mo and 3 years 4 mo)	SR, CsA-NR	ND		
-22									F			1 mo	ND	ESRD 6 yrs 7 mo	ND		
A2231-21	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	White	N	5 yrs 1 mo	ND	ND	ND	[16]	Flui
	c.467delT	p.Leu156Cysfs*25	4 (het)	NA	NA	NA	NA	-								[29]	Flui
942	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		ND	N					[16]	SSe
	c.503G>A	p.Arg168His	4 (het)	0.999	del	DC	*C. elegans*	-								[29]	SSe
-21									M			1 mo	FSGS	ACE-I	hypospadias		
-22									M			1 mo	ND	ACE-I	ND		
A52	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292								[16]	SSe
	c.538G>A	p.Val180Met	5 (het)	0.577	del	DC	*D. rerio*	TT=0/TC=1/CC=4299								[16]	SSe
-21									F	ND	N	9 yrs 4 mo	FSGS	SR, CsA-PR	ND		
-22									F	ND	N	ND	ND	ND	ND		
A1990	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	White	N	8 mo	MCNS (8 mo)	SR, CsA-NR	ND	[16]	SSe
	c.643C>T	p.Gln215*	5 (het)	NA	NA	NA	NA	-								[31]	SSe
1261	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	ND	ND	3 yrs 7 mo	FSGS with MesP (3 yrs 10 mo)	SR, CsA-NR	ND	[16]	SSe
	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979								[33]	SSe
A858	c.413G>A	p.Arg138Gln	3 (het)	0.999	Del	DC	*C. elegans*	TT=0/TC=8/CC=4292		White	N					[16]	SSe
	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979								[33]	SSe
-24									M			ND	ND	ND	ND		
-25									M			ND	ND	ND	ND		
-26									M			ND	ND	ND	ND		
A1112	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	European	N	3 yrs 6 mo	FSGS	SR, CsA-PR	ND	[16]	SSe
	c.790G>T	p.Glu264*	6 (het)	NA	NA	NA	NA	-								[34]	SSe
A2084	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292		ND	ND					[16]	SSe
	c.856delA	p.Arg286Aspfs*7	7 (het)	0.999	NA	NA	NA	-								novel	SSe
-21									ND			ND	ND	ND	ND		
-22									ND			1 yr 1 mo	FSGS (1 yr 1 mo)	SR	ND		
A2432-21	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	European	N	ND	ND	ND	GR	[16]	SSe
	c.868G>A	p.Val290Met	7 (het)	0.989	del	DC	*D. rerio*	TT=0/TC=1/CC=4299								[35]	SSe
A268-21	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	ND	N	13 yrs 9 mo	MPGN (14 yrs 2 mo), MCNS (17 yrs 3 mo)	SR, CsA	ND	[16]	SSe
	c.868G>A	p.Val290Met	7 (het)	0.989	del	DC	*D. rerio*	TT=0/TC=1/CC=4299								[35]	SSe
A4426-21	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	European	N	ND	MCNS (27 years), FSGS (30 years)	ND	ND	[16]	SSe
	c.868G>A	p.Val290Met	7 (het)	0.989	del	DC	*D.rerio*	TT=0/TC=1/CC=4299								[35]	SSe
A49	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	M	ND	N	14 yrs 3 mo	FSGS (3 yrs 6 mo, 13 yrs mo)	SR, CsA- NR, CP-NR	ND	[16]	SSe
	c.868G>A	p.Val290Met	7 (het)	0.989	del	DC	*D.rerio*	TT=0/TC=1/CC=4299								[35]	SSe
A2547-21	c.413G>A	p.Arg138Gln	3 (het)	0.999	del	DC	*C. elegans*	TT=0/TC=8/CC=4292	F	European	N	5 yrs 5 mo	MCNS (5 yrs 5 mo)	SR, CsA	ND	[16]	SSe
	c.1150T>C	p.*384Glnext*7	8 (het)	NA	NA	NA	NA	-								novel	SSe
A3679	c.416T>G	p.Leu139Arg	3 (hom)	0.999	del	DC	*C. elegans*	-		Hispanic	N					[36]	SSe
-22									M			3 yrs 10 mo	FSGS (3 yrs 10 mo)	SR, MMF-NR, CsA-PR	ND		
-23									M			1 yr 9 mo	ND	ND	ND		
A3133-21	c.419delG	p.Gly140Aspfs*41	3 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	5 yrs 8 mo	FSGS (6 yrs)	ND	ND	[16]	Flui
A4603-21	c.419delG	p.Gly140Aspfs*41	3 (hom)	NA	NA	NA	NA	-	F	Arabic-african	N	7 months	MPGN	SR, CP-NR	ND	[16]	Flui
A1645	c.419delG	p.Gly140Aspfs*41	3 (het)	NA	NA	NA	NA	-	M	Turkish	N	1 yr	DMS (1 yr) and FSGS (7 yrs 4 mo)	SR, CP-NR	ND	[16]	SSe
	c.506T>C	p.Leu169Pro	4 (het)	0.991	del	DC	*D. rerio*	-								[32]	SSe
A2057-21	c.451+3A>T	Splice	3 (het)	NA	NA	NA	NA	-	F	European	N	5 yrs 3 mo	ND	ND	ND	[37]	SSe
	c.868G>A	p.Val290Met	7 (het)	0.989	del	DC	*D. rerio*	TT=0/TC=1/CC=4299								[35]	SSe
1334	c.467dupT	p.Leu156Phefs*11	4 (hom)	NA	NA	NA	NA	-	M	ND	Y	2 yrs 9 mo	FSGS	SR, CsA-NR	ND	[32]	SSe
A28	c.467dupT	p.Leu156Phefs*11	4 (hom)	NA	NA	NA	NA	-		Turkish	Y					[32]	SSe
-21									F			5 yrs 5 mo	FSGS (5 yrs 5 mo) MCNS (1yrs 3 mo)	SR, CP -NR, CsA-NR	SS		
-22									M			1 yr 1 mo		SR, CP-NR	ND		
A3577-21	c.467dupT	p.Leu156Phefs*11	4 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	6 yrs 6 mo	FSGS	SR	ND	[32]	SSe
A4624-21	c.467dupT	p.Leu156Phefs*11	4 (hom)	NA	NA	NA	NA	-	F	Arabic-african	Y	1 yr 8 mo	MPGN (1 yr 8 mo)	SR, CP-NR, CsA-NR	ND	[32]	SSe
A818-21	c.467dupT	p.Leu156Phefs*11	4 (hom)	NA	NA	NA	NA	-	M	Turkish	Y	3 yrs	FSGS (3 yrs 9 mo)	SR, CsA, CP-NR	ND	[32]	SSe
A270-21	c.467dupT	p.Leu156Phefs*11	4 (het)	NA	NA	NA	NA	-	F	ND	N	2 yrs 7 mo	MCNS (2 yrs 7 mo), FSGS (3 yrs 4 mo)	SR, CP, CsA-PR	SS	[32]	SSe
	c.948delT	p.Ala317Leufs*31	8 (het)	NA	NA	NA	NA	-								[38]	SSe

A2278-21	c.467dupT	p.Leu156Phefs*11	4 (het)	NA	NA	NA	NA	-	F	Arabic	ND	1 yr	FSGS (3 yrs 7 mo, 4 yrs 3 mo)	SR, CsA-NR, MMF	bilateral VUR	[32]	SSe
	c.779T>A	p.Val260Glu	6 (het)	0.991	del	DC	*C.elegans*	-								[29]	SSe
A4193-23	c.479A>G	p.Asp160Gly	4 (hom)	1	del	DC	*D. melanogaster*	-	M	Arabic	Y	1 yr 5 mo	ND	ND	ND	[16]	SSe
A1934	c.486C>A	p.Tyr162*	4 (hom)	NA	NA	NA	NA	-	M	Hispanic	N	2 mo	FSGS	SR	LVH	novel	SSe
A5175-21	c.502C>T	p.Arg168Cys	4 (hom)	1	del	DC	*C. elegans*	-	M	Yemeni	Y	2 yrs	ND	SR	ND	[29]	SSe
A4008-23	c.503G>A	p.Arg168His	4 (hom)	0.999	del	DC	*C. elegans*	-	M	Arabic	Y	1 yr 5 mo	cFSGS (2 yrs 2 mo)	SR, CP-NR	ND	[29]	SSe
A1897	c.503G>A	p.Arg168His	4 (hom)	0.999	del	DC	*C. elegans*	-	F	Turkish	Y	2 yrs 7 mo	FSGS (2 yrs 7 mo)	SR	ND	[29]	SSe
A4327-23	c.503G>A	p.Arg168His	4 (hom)	0.999	del	DC	*C. elegans*	-	F	Arabic	Y	5 yrs 6 mo	FSGS (3 yrs 2 mo)	SR, CP-NR, CsA-NR	Umbilical hernia	[29]	SSe
A2564-21	c.538G>A	p.Val180Met	5 (hom)	0.577	del	DC	*D. rerio*	TT=0/TC=1/CC=4299	ND	ND	ND	ND	ND	ND	ND	[16]	SSe
A3379-21	c.538G>A	p.Val180Met	5 (hom)	0.577	del	DC	*D. rerio*	TT=0/TC=1/CC=4299	F	Arabic	Y	10 yrs 11 mo	FSGS	SR, CP, CsA-NR	ND	[16]	SSe
A3574-21	c.538G>A	p.Val180Met	5 (hom)	0.577	del	DC	*D. rerio*	TT=0/TC=1/CC=4299	M	Arabic	Y	5 yrs 4 mo	IgM nephropathy (5 yrs 4 mo)	SR, CP-PR, CsA-PR, MMF	ND	[16]	SSe
A1071-21	c.538G>A	p.Val180Met	5 (hom)	0.577	del	DC	*D. rerio*	TT=0/TC=1/CC=4299	M	Turkish	Y	11 yr 5 mo	FSGS (11 yr 5 mo)	SR, SP-NR, CsA-NR	ND	[16]	SSe
A3035-21	c.538G>A	p.Val180Met	5 (hom)	0.577	del	DC	*D. rerio*	TT=0/TC=1/CC=4299	F	Arabic	Y	3 yrs 5 mo	FSGS	SR, RTX-NR, MMF-NR	ND	[16]	SSe
A147-21	c.593A>C	p.Glu198Ala	5 (hom)	0.808	del	DC	*D. rerio*	-	M	Turkish	Y	3 yrs 6 mo	FSGS (8 yrs 5 mo)	SR, CP, CsA-NR	ND	novel	SSe
A4653-31	c.596dupA	p.Asn199Lysfs*14	5 (hom)	NA	NA	NA	NA	No	F	Arabic	Y	1 yr 3 mo	FSGS	SR, CP-NR	ND	novel	SSe
A4341-23	c.596dupA	p.Asn199Lysfs*14	5 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	4 yrs	FSGS (6 yrs 2 mo)	SR, CP-NR	Asthma	novel	SSe
A4383-21	c.596dupA	p.Asn199Lysfs*14	5 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	5 yrs 3 mo	ND	SR, CP-PR	ND	novel	SSe
A3792-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	European	N	8 yrs	FSGS, MPGN (8 yrs)	SR	HA	[33]	SSe
	c.826_833dupCACTCACT	p.Ala279Thrfs*17	7 (het)	NA	NA	NA	NA	-								novel	SSe
A3447	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979		White	Y					[33]	SSe
	c.841G>A	p.Glu281Lys	7 (het)	0.991	del	DC	*C. elegans*	-								novel	SSe
-21									F			3 yrs	ND	ND	ND		
-22									F			22 yrs 5 mo	FSGS (22 yrs 6 mo)	SR	ND		
370	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979		European	N					[33]	SSe
	c.851C>T	p.Ala284Val	7 (het)	0.998	del	DC	*C. elegans*	-								[35]	SSe
-33									F			9 yrs	FSGS (15 yrs 6 mo)	SR	ND		
-31									M			24 yrs	FSGS (24 yrs)	SR, CsA-NR, CP-NR	ND		
489	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979		European	N					[33]	SSe
	c.851C>T	p.Ala284Val	7 (het)	0.998	del	DC	*C. elegans*	-								[35]	
-24									M			11 yr 6 mo	MesP (13 yrs 8 mo)	SR, CP-NR	ND		SSe
-27									F			3 yrs 4 mo	FSGS (3 yrs 8 mo)	SR, CP-NR, CsA-NR	ND		
A2200-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	Hispanic	N	13 yrs	FSGS	SR	ND	[33]	SSe
	c.851C>T	p.Ala284Val	7 (het)	0.998	del	DC	*C. elegans*	-								[35]	SSe
A4934-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	White	N	26 yrs	FSGS (27 yrs)	SR, CsA-PR	ND	[33]	SSeS
	c.851C>T	p.Ala284Val	7 (het)	0.998	del	DC	*C. elegans*	-								[35]	Se
A5243-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	M	African-Arabic	Y	12 yrs 7 mo	ND	SR	ND	[33]	SSe
	c.851C>T	p.Ala284Val	7 (het)	0.998	del	DC	*C. elegans*	-								[35]	SSe
A2242-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	European	N	13 yrs	MCNS (13 yrs), FSGS (14 yrs)	SR, MMF-NR, Tac-NR, CsA-NR	ND	[33]	SSe
	c.862G>A	p.Ala288Thr	7 (het)	0.999	del	DC	*C. elegans*	-							ND	[33]	SSe
A1698	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	Central Slavic	N	2 yrs 3 mo	ND	ND	ND	[33]	SSe
	c.868G>A	p.Val290Met	7 (het)	0.989	del	DC	*D. rerio*	TT=0/TC=1/CC=4299								[35]	SSe
A1634	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	European	N	1 mo	MCNS (2 yrs 10 mo)	ND	ND	[33]	SSe
	c.871C>T	p.Arg291Trp	7 (het)	0.978	del	DC	*G. gallus*	-							ND	[16]	SSe
A3356-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	M	White	N	1 yr 6 mo	MPGN (1 yr 6 mo)	SR, CP, CsA-R	ID	[33]	SSe
	c.868G>A	p.Val290Met	7 (het)	0.989	del	DC	*D.rerio*	TT=0/TC=1/CC=4299									SSe
A4956-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	M	Indian	N	4 yrs 4 mo	FSGS (6 yrs 5 mo)	SR, Tac, RTX, CsA-NR	ND	[33]	SSe
	c.890C>T	p.Ala297Val	8 (het)	0.627	del	PMP	*H. sapiens*	-								[33]	SSe
585	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979		European	N					[33]	SSe
	c.898G>C	p.Ala300Pro	8 (het)	0.953	del	DC	*G. gallus*	-								novel	SSe
-21									F			16 yrs 3 mo	FSGS (16 yrs 3 mo)	SR	ND		
-13									F			60 yrs	FSGS (60 yrs)	SR	ND		

A667	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979		European	N					[33]	SSe
	c.916A>T	p.Arg306Trp	8 (het)	0.983	del	DC	*C. elegans*	-								novel	SSe
-21									F			14 yrs 7 mo	FSGS	Steroids, CsA	ND		
-22									M			ND	ND	ND	ND		
A5280-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	M	European	N	30 yrs	FSGS (30 yrs)	SR, CsA-NR	ND	[33]	SSe
	c.928G>A	p.Glu310Lys	8 (het)	0.936	del	DC	*D. rerio*	-								[39]	SSe
454	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979		European	N					[33]	SSe
	c.929A>T	p.Glu310Val	8 (het)	0.96	del	DC	*D. rerio*	-								[33]	SSe
-21									M			ND	ND	ND	ND		
-22									M			2 mo	FSGS (9 yrs 6 mo)	SR, CsA	ND		
1285	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	ND	N	4 yrs 2 mo	FSGS	SR, CsA-NR	ND	[33]	SSe
	c.929A>C	p.Glu310Ala	8 (het)	0.837	del	DC	*D. rerio*	-								novel	SSe
A3279-31	c.686G>A	p.Arg229Gln	5 (het)	0.313	Del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	German	N	ND	ND	SR	ND	[33]	SSe
	c.929A>T	p.Glu310Val	8 (het)	0.96	del	DC	*D. rerio*	-								[33]	SSe
A1495	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	Indian	N	1 yr 8 mo	MCNS (1 yr 8 mo)	SR, CP-PR	ND	[33]	SSe
	c.965G>C	p.Arg322Pro	8 (het)	1	del	DC	*C. elegans*	-								[40]	SSe
A907-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	Del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	M	Central	N	13 yrs 4 mo	FSGS (13 yrs 4	SR, CsA-NR	ND	[33]	SSe
	c.961delC	p.Leu321Phefs*27	8 (het)	NA	NA	NA	NA	-		slavic			mo)			novel	SSe
A239-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	ND	N	4 yrs 2 mo	MCNS with MesP	SR, CsA-PR	ND	[33]	SSe
	c.979C>T	p.Leu327Phe	8 (het)	0.997	del	DC	*C. elegans*	-					(16 yrs 2 mo)			[33]	SSe
A277-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	ND	N	6 yrs	MCNS (19 yrs 9	SR, CsA-PR	ND	[33]	SSe
	c.979C>T	p.Leu327Phe	8 (het)	0.997	del	DC	*C. elegans*	-					mo)			[33]	SSe
A4326	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979		White	N					[33]	SSe
	c.983A>G	p.Gln328Arg	8 (het)	0.108	tol	DC	*M. musculus*	-								[38]	SSe
-21									F			9 yrs	ND	ACE-I	ND		
-22									F			6 yrs	FSGS	ACE-I	ND		
A4678-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	European	N	ND	FSGS (31 yr)	MMF-NR, CsA-NR	ND	[33]	SSe
	c.983A>G	p.Gln328Arg	8 (het)	0.108	tol	DC	*M. musculus*	-								[38]	SSe
1301	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	ND	N	10 yrs	MCNS (24 yrs 1	Steroids, ACE-I	ND	[33]	SSe
	c.983A>G	p.Gln328Arg	8 (het)	0.108	tol	DC	*M. musculus*	-					mo)			[38]	SSe
A3169-22	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	White	N	14 yrs 10 mo	FSGS (14 yrs 10)	SR	ND	[33]	SSe
	c.1032delT	p.Phe344Leufs*4	8 (het)	NA	NA	NA	NA	NA								[41]	SSe
A900-21	c.686G>A	p.Arg229Gln	5 (het)	0.313	del	DC	*X. tropicalis*	TT=2/TC=319/CC=3979	F	European	N	7 mo	MCNS (1 yr 2 mo)	SR, CP-NR	ND	[33]	SSe
	c.1032delT	p.Phe344Leufs*4	8 (het)	NA	NA	NA	NA	-								[41]	SSe
A3453-21	c.714G>T	p.Arg238Ser	5 (hom)	0.925	del	DC	*X. tropicalis*	-	M	Arabic	Y	4 yrs	ND	SR, Tac-NR	ND	[29]	SSe
A3120-21	c.779T>A	p.Val260Glu	6 (hom)	0.991	del	DC	*C. elegans*	-	F	Arabic	Y	9 mo	FSGS (1 yr 4 mo)	SR	ND	[29]	SSe
A2337-21	c.779T>A	p.Val260Glu	6 (hom)	0.991	del	DC	*C. elegans*	-	F	Arabic	N	ND	MPGN, secondary	ND	ND	[29]	SSe
													FSGS (2 yrs 1 mo)				
A3183-21	c.779T>A	p.Val260Glu	6 (hom)	0.991	del	DC	*C. elegans*	-	F	Arabic	Y	1 yr	FSGS (1 yr)	SR	ND	[29]	SSe
A4329-23	c.779T>A	p.Val260Glu	6 (hom)	0.991	del	DC	C.elegans	-	F	Arabic	N	1 yr 5 mo	MPGN (2 yrs)	SR	ND	[29]	SSe
1336	c.868G>A	p.Val290Met	7 (hom)	0.989	del	DC	*D. rerio*	TT=0/TC=1/CC=4299	F	ND	Y	9 mo	MCNS (12 yrs)	SR	ND	[35]	SSe
A1616	c.868G>A	p.Val290Met	7 (hom)	0.998	del	DC	*D. rerio*	TT=0/TC=1/CC=4299	F	White	N	ND	ND	ND	ND	[35]	Flui
A4705	c.877A>T	p.Ile293Phe	8 (hom)	0.999	del	DC	*C. elegans*	-		Hispanic	Y					novel	SSe
-21									F			13 yrs	ND	SR, CP-NR	ND		
-22									F			8 yrs	FSGS (ND)	CP-NR	ND		
A1430	c.883G>A	p.Ala295Thr	8 (hom)	1	del	DC	*C. elegans*	-	M	Turkish	Y	9 yrs 5 mo	FSGS (9 yrs 5 mo)	ND	ND	novel	SSe
A2239-21	c.926C>T	p.Ala309Val	8 (hom)	0.742	del	DC	*C. elegans*	-	M	Turkish	N	ND	FSGS (ND)	ND	ND	novel	Flui
A3541	c.934C>G	p.Leu312Val	8 (hom)	0.677	del	DC	*X. tropicalis*	-		Arabic	Y					[31]	SSe
-22									F			9 yrs 5 mo	MesP (9 yrs 6 mo)	ND	SS		
-23									M			8 yrs	MesP (5 yrs 5 mo)	ND	SS		
-24									F			5 yrs	ND	ND	SS		
A3828-21	c.946C>T	p.Pro316Ser	8 (hom)	0.909	del	DC	*C. elegans*	-	F	Indian	Y	3 yrs 1 mo	FSGS (4 yrs 4 mo)	SR	ND	novel	SSe
1058	c.1150T>C	p.*384Glnext*7	8 (hom)	NA	NA	NA	NA	-		Turkish	Y					novel	SSe
-41									M			ND	ND	ND	ND		
-42									F			ND	ND	SR	ND		
NPHS1																	
A1803-21	c.139delG	p.Ala47Profs*81	2 (hom)	NA	NA	NA	NA	-	M	White	N	birth	MCNS (1 mo)	CsA-NR	ND	novel	Flui
A1893-21	c.139delG	p.Ala47Profs*81	2 (het)	NA	NA	NA	NA	-	F	Hispanic	N	birth	ND	ND	ND	novel	SSe
	c.3595-2A>G	splice	29 (het)	NA	NA	NA	NA	-								novel	
A657-21	c.313G>A	p.Asp105Asn	3 (het)	1	del	DC	*D. melanogaster*	-	F	African-	N	1 mo	FSGS (1 mo)	CsA	ND	[42]	Flui
	c.1913A>G	p.Tyr638Cys	14 (het)	0.999	del	DC	*D. rerio*	-		American						novel	

A2559-21	c.313G>A c.2928G>T	p.Asp105Asn p.Arg976Ser	3 (het) 22 (het)	1 0.873	del del	DC DC	*D.melanogaster* *D.melanogaster*	- AA=0/AC=2/CC=4298	M	African-American	N	3 yrs 8 mo	MCNS (3 yrs 8 mo)	ND	ND	[42] [43]	Flui
A1480-21	c.319G>A c.3244G>A	p.Ala107Thr p.Gly1082Arg	3 (het) 24 (het)	0.929 0.994	del del	DC PMP	*D.melanogaster X. tropicalis*	TT=0/TC=0/CC=2203 TT=0/TC=0/CC=2203	M	European	N	5 yrs 8 mo	ND	ND	ND	[43] novel	Flui
A1641-21	c.398-1G>A c.3478C>T	splice p.Arg1160*	3 (het) 27 (het)	NA NA	NA NA	NA NA	NA NA	-	M	European	N	1 mo	ND	ND	ND	novel [44]	SSe
A915-21	c.468C>G c.2928G>T	p.Tyr156* p.Arg976Ser	4 (het) 22 (het)	NA 0.995	NA del	NA DC	NA *D. rerio*	- AA=0/AC=2/CC=4298	M	European	N	5 yrs	MCNS (5 yrs)	ND	ND	[44] [43]	Flui
A4960-21	c.500C>T	p.Pro167Leu	4 (hom)	0.988	del	DC	*C. elegans*	-	M	Indian	Y	1 yr	ND	ND	ND	[45]	SSe
A3850	c.513delGTG c.1316A>G	p.Ile171_Thr172delins p.Tyr439Cys	4 (het) 11 (het)	NA 1	NA del	NA DC	NA *D. melanogaster*	- -		Egyptian	Y					novel novel	Flui
-21									F			2 mo	MPGN (3 yrs)	ND	ND		
-22									F			2 mo	ND	ND	ND		
A3583-22	c.515-517delCCA	p.Thr172del	4 (hom)	NA	NA	NA	M. musculus	-	M	Arabic	Y	2 mo	CNS (2 mo)	ESRD 5 yrs after onset	Subaortic stenosis	[46]	Flui
A3490-21	c.532C>T c.2928G>T	p.Gln178* p.Arg976Ser	5 (het) 22 (het)	NA 0.995	NA del	NA DC	NA *D. melanogaster*	- AA=0/AC=2/CC=4298	F	White	N	10 yrs 5 mo	FSGS (10 yrs 5 mo)	CsA	ND	[44] [43]	Flui
A3628-21	c.563A>T c.791C>G	p.Asn188Ile p.Pro264Arg	5 (het) 7 (het)	0.573 0.675	tol tol	PMP PMP	*X. tropicalis* *M. musculus*	AA=0/AT=84/TT=4216 CC=1/CG=133/GG=4166	M	Indian	N	1 yr 4 mo	MCNS (3 yrs 3 mo)	ND	ND	[47] [47]	Flui
A669-21	c.614_621delinsTTe	p.Thr205_Arg207delinsIle	6 (hom)	NA	NA	NA	NA	-	F	Arabic	N	birth	ND	ND	ND	novel	SSe
A1543-21	c.614_621delinsTTe	p.Thr205_Arg207delinsIle	6 (het)	NA	NA	NA	NA	-	M	White	N	1 mo	ND	ND	ND	[48]	SSe
	c.1307_1308dupAC	p.Val437fs*2	10 (het)	NA	NA	NA	NA									novel	
A1943-21	c.614_621delinsTTe	p.Thr205_Arg207delinsIle	6 (het)	NA	NA	NA	NA	-	M	European	N	1 mo	Finnish type (1 mo)	ND	ND	novel	SSe
	c.1715A>G	p.Ser572Gly	13 (het)	1	del	DC	*D. melanogaster*									novel	
A5242-22	c.617C>A	p.Pro206His	6 (hom)	1	del	DC	*X. tropicalis*	-	F	Arab	Y	2 yr	ND	ND	HA	novel	Flui
A3652-21	c.766C>T c.1234G>A	p.Arg256Trp p.Gly412Ser	7 (het) 10 (het)	1 0.999	del del	PMP DC	*D. melanogaster* *D. rerio*	- TT=0/TC=1/CC=4299	M	Indian	Y	5 yrs 2 mo	MCNS (11 yrs 7 mo)	ND	ND	[48] novel	Flui
A2070-21	c.791C>G	p.Pro264Arg	7 (hom)	0.675	tol	PMP	*D. melanogaster*	-	M	Turkey	N	ND	ND	ND	ND	[47]	Flui
A1619-21	c.791C>G c.1223G>A	p.Pro264Arg p.Arg408Gln	7 (het, p) 10 (het, m)	0.675 1	tol del	PMP DC	*D. melanogaster* *D. rerio*	- -	M	African American	N	11 yr 7 mo	FSGS (11 yrs 7 mo)	ND	ND	[47] [46]	Flui
A1739-21	c.791C>G c.3151A>G	p.Pro264Arg p.Thr1051Ala	7 (het) 23 (het)	0.675 0.016	tol tol	PMP PMP	*D. melanogaster* *D. rerio*	- -	M	Arab	Y	10 yr	FSGS (10 yrs)	ND	ND	[47] novel	Flui
A5144-23	c.802C>T	p.Arg268*	7 (hom)	NA	NA	NA	NA	-	F	Arab	Y	4 days	ND	ND	ND	[49]	Flui
A3380-21	c.928G>A c.2816-3T>G	p.Asp310Asn splice	8 (het) 21 (het)	0.99 NA	del NA	DC NA	*D. melanogaster* NA	- -	M	Asian	ND	1 mo	ND	ND	ND	[50] novel	Flui
A4400-22	c.1020delT	p.Ser341Valfs*17	9 (hom)	NA	NA	NA	NA	-	M	Egyptian	N	3 mo	ND	ND	HA	novel	Flui
A4339-26	c.1040G>A	p.Gly347Glu	9 (hom)	1	del	DC	*D. melanogaster*	-	M	Arab	Y	1 mo	ND	ND	ND	[45]	Flui
A4378-21	c.1040G>A	p.Gly347Glu	9 (hom)	1	del	DC	*D. melanogaster*	-	F	Egyptian	N	4 mo	ND	ND	Intestinal obstruction	novel	Flui
A2094	c.1089C>G c.2928G>T	p.Ser363Arg p.Arg976Ser	9 (het) 24 (het)	0.922 0.995	del del	PMP DC	*D. melanogaster* *D. melanogaster*	- AA=0/AC=2/CC=4298		European	N					novel [43]	Flui SSe
-22									M			3 mo	ND	CP-NR	ND		
-21									M			1 yr	FSGS (ND)	SR, CP-NR	mitralis valve prolapse		
A3543-22	c.1096A>C	p.Ser366Arg	9 (hom)	1	del	DC	*D. rerio*	-	M	Serbian	N	4 days	ND	ND	ND	[46]	Flui
A2030-21	c.1096A>C c.3478C>T	p.Ser366Arg p.Arg1160*	9 (het) 27 (het)	0.997 NA	del NA	DC NA	*D. melanogaster* NA	- -	M	Montenegro	N	1 yr 8 mo	ND	ND	ND	[46] [46]	SSe
A2617-21	c.1099C>T	p.Arg367Cys	9 (Hom)	1	del	DC	*M. musculus*	-	F	European	Y	1 yr	Finnish type (1 yr)	ND	ND	novel	SSe
A2062-21	c.1099C>T c.2625G>A	p.Arg367Cys p.Trp875*	9 (het) 19 (het)	1 NA	del NA	DC NA	*M. musculus* NA	- -	M	Chinese	N	1 mo	ND	ND	Hypothyroid-ism	[47] [44]	Flui
A4444-21	c.1099C>T c.3286+2T>C	p.Arg367Cys splice	9 (het) 24 (het)	1 NA	del NA	DC NA	*M. musculus* NA	- -	M	Indian	N	3 mo	ND	ND	deafness, MC, FD	novel [39]	Flui
A5128-22	c.1219C>T	p.Arg407Trp	10 (hom)	0.938	del	PMP	*D. rerio*	-	F	Indian	Y	1 mo	ND	ND	ND	[45]	SSe
A5068-21	c.1219C>T c.2422delC	p.Arg407Trp p.Leu808Trpfs*39	10 (het, m) 16 (het, p)	0.938 NA	del NA	PMP NA	*D. rerio* NA	-	M	Indian	N	1 mo	ND	ND	ND	[45]	SSe

A3439-21	c.1223G>A c.1703T>C	p.Arg408Gln p.Val568Ala	10 (het) 13 (het)	1	del tol	DC DC	*D. rerio* *X. tropicalis*	-		Slavisch	N	1 yr	MCNS (1 yr)	ND	ND	[46] novel	Flui
A1116	c.1234G>T c.3250dupG	p.Gly412Cys p.Val1084Glyfs*12	10 (het) 24 (het)	1 NA	del NA	DC NA	*D. rerio* NA	- -		White	ND					novel novel	SSe
-22									M			1 mo	ND	ND	ND		
-25									M			1 mo	ND	ND	ND		
A1028-21	c.1248dupA c.2606_2607dupCC	p.Cys417Metfs*2 p.Asn870Profs*36	10 (het) 19 (het)	NA NA	NA NA	NA NA	NA NA	-		ND	N	1 mo	ND	ND	ND	novel	Flui
A813-21	c.1339G>A c.1802G>C	p.Glu447Lys p.Gly601Ala	11 (het) 14 (het)	0.996 1	del del	DC DC	*X. tropicalis* *D. melanogaster*	- -	F	Pacific Islander	N	14 yrs 8 mo	FSGS (14 yrs 8 mo)	ESRD	ND	[44] [46]	Flui
A3250-21	c.1339G>A c.1802G>C	p.Glu447Lys p.Gly601Ala	11 (het) 14 (het)	0.996 1	del del	DC DC	*X. tropicalis* *D. melanogaster*	- -	F	Chinese	N	3 yrs	MPGN	ND	ID	[44] [46]	Flui
A1831-21	c.1379G>A c.2769_2776delCACGCCC	p.Arg460Gln p.Asn924Trpfs*16	11 (het) 20 (het)	0.999 NA	tol NA	PMP NA	*M. musculus* NA	-	F	White	N	1 mo	ND	ND	ND	[44] [48]	SSe
A1475-21	c.1404_1405insGGC	p.Gly470dup	11 (hom)	NA	NA	NA	NA	-	M	Arabic	N	ND	FSGS	ND	ND	novel	SSe
A5098-25	c.1538T>C	p.Leu513Pro	12 (hom)	0.873	del	DC	*D. rerio*	-	M	Arabic	Y	2 mo	ND	ND	ND	[51]	SSe
A1193-21	c.1555C>T c.2596C>T	p.Pro519Ser p.Arg866*	12 (het) 19 (het)	1 NA	del NA	DC NA	*D. rerio* NA	-	M	European	N	4 mo	ND	ND	ND	[48] [48]	SSe
A1433-21	c.1707C>G	p.Ser569Arg	13 (hom)	1	del	PMP	*D. melanogaster*	-	M	Arabic	Y	birth	DMS (1 mo)	ND	ID	[52, 53]	SSe
A2554-21	c.1715A>G	p.Ser572Gly	13 (hom)	1	del	DC	*D. melanogaster*	-	F	Turkish	ND	6 mo	MCNS (1 yr 9 mo)	ND	FD, hepatomegaly	novel	Flui
A1801-21	c.1716C>G c.3478C>T	p.Ser572Arg p.Arg1160*	13 (het) 27 (het)	0.999 NA	del NA	DC NA	*D. melanogaster* NA	-	F	Maltese	N	1 mo	DMS (1yr)	ND	ND	[53] [46]	SSe
A3819-21	c.1738T>G c.3211G>T	p.Trp580Gly p.Gly1071Trp	13 (het) 24 (het)	0.973 0.975	del del	DC DC	*C. elegans* *M. musculus*	- -	F	European	N	1 mo	ND	NTX, nephrectomy (4 yrs)	ND	[54] novel	Flui
A4282-21	c.1756A>G	p.Arg586Gly	13 (hom)	0.002	del	PMP	*M. musculus*	-	F	Somalian	Y	9 yrs11 mo	FSGS (10 yr)	CsA-PR	ND	[45]	Flui
A5191-21	c.1881C>A c.2014G>A	p.Ser627Arg p.Ala672Thr	14 (het) 15 (het)	1 1	del del	DC DC	*D. rerio* *D. melanogaster*	- -	M	Turkish	N	4 mo	ND	ND	ND	novel [39]	Flui
A1981-21	c.2019C>A c.3478C>T	p.Asn673Lys p.Arg1160*	15 (het) 27 (het)	0.999 NA	del NA	DC NA	*D. melanogaster* NA	-	F	European	N	birth	ND	ND	ND	[45] [46]	SSe
A3432-24	c.2020C>A	p.Pro674Thr	15 (hom)	1	del	DC	*D. rerio*	-	M	Arab	Y	1 mo	ND	ND	ND	novel	Flui
A2087	c.2215G>A	p.Ala739Thr	17 (hom)	1	tol	DC	*M. musculus*	-		Asian	N		ND			[44]	Flui
-21									F			1 yr 8 mo		CsA-PR	ND		
-22									F			ND		CsA-PR			
A683-21	c.2216C>T c.3478C>T	p.Ala739Val p.Arg1160*	17 (het) 27 (het)	0.997 NA	tol NA	DC NA	*M. musculus* NA	- -	M	Turkish	N	2 mo	mesangial sclerosis	ND	Deafness, ID, PAS	[44] [46]	Flui
A2250-21	c.2227C>T c.3482-2A>G	p.Arg743Cys splice	17 (het) 27 (het)	0.997 NA	del NA	DC NA	*D. rerio* NA	-	M	European	N	2 mo	ND	ND	ND	[46]	SSe
A5159-21	c.2299C>T	p.Pro767Ser	17 (hom)	1	del	DC	*D. melanogaster*	-	F	Turkish	Y	1 mo	ND	ND	ND	novel	SSe
A5020-21	c.2386G>A c.2815+3A>G	p.Gly796Arg splice	18 (het) 20 (het)	1 NA	del NA	DC NA	*X. tropicalis* NA	- -	F	Malay	N	2 mo	ND	ND	MC	[55] novel	SSe
A5060-22	c.2514_2515dupCC	p.Gln839Profs*9	19 (hom)	NA	NA	NA	NA	-	M	Arab	Y	1 mo	ND	ND	ND	novel	Flui
A1680-21	c.2542A>C c.2548del10	p.Lys848Gln p.A850fs*873	19 (het) 19 (het)	0.999 NA	del NA	DC NA	*C. elegans* NA	-	F	Turkish	N	birth	ND	ND	ND	novel novel	SSe
A931-21	c.2606_2607dupCC	p.Asn870Profs*36	19 (het)	NA	NA	NA	NA	-	F	White	N	1 mo	ND	ND	FD	[46] [45]	SSe SSe
	c.2928G>T	p.Arg976Ser	22 (het)	0.873	del	DC	*D. melanogaster*										
A1500-21	c.2728T>C	p.Ser910Pro	20 (hom)	0.959	del	DC	*D. rerio*	-		African-American	N	1 yr	MCNS (1 yr)	ND	ND	[45]	Flui
A2330-21	c.2815+5G>A	splice	20 (hom)	NA	NA	NA	NA	-	M	Turkish	Y	1 yr	ND	ND	ND	[45]	SSe
A1177-21	c.3250dupG	p.Val1084Glyfs*12	24 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	4 days	ND	ND	ND	novel	SSe
A1550-21	c.3250dupG	p.Val1084Glyfs*12	24 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	1 mo	ND	ND	ND	novel	SSe
A2538-22	c.3250dupG	p.Val1084Glyfs*12	24 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	1 mo	ND	ND	ND	novel	SSe
A3075-24	c.3250dupG	p.Val1084Glyfs*12	24 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	3 mo	ND	ND	VSD	novel	SSe
A2553-29	c.3325C>T	p.Arg1109*	26 (hom)	NA	NA	NA	NA	-	M	Turkish	Y	1 mo	ND	ND	ND	[15]	SSe
A2210	c.3478C>T	p.Arg1160*	27 (hom)	NA	NA	NA	NA	-		Libanese	Y					[46]	SSe
-21									F			1 mo	ND	CsA, ACE-I	ND		
-23									F			6 days	ND	ACE-I	ND		

A3708-21	c.3478C>T	p.Arg1160*	27 (hom)	NA	NA	NA	NA	-	M	Sudanese	Y	2 mo	FSGS (2 yr)	ND	ND	[46]	Flui
A3509-21	c.3478C>T	p.Arg1160*	27 (hom)	NA	NA	NA	NA	-	F	Asian	Y	1 mo	ND	ND	ND	[46]	Flui
A3594-21	c.3478C>T	p.Arg1160*	27 (hom)	NA	NA	NA	NA	-	F	Arab	Y	At birth	ND	ND	ND	[46]	Flui
A4427-23	c.3478C>T	p.Arg1160*	27 (hom)	NA	NA	NA	NA	-	F	Yemenite	N	At birth	ND	ND	ND	[46]	SSe
COQ2																	
A758-21	c.683A>G	p.Asn228Ser	3 (hom)	0.918	tol	DC	*C. elegans*	-	M	White	ND	ND	ND	ND	ND	[5]	Flui
A1727-21	c.518G>A c.683A>G	p.Arg173His p.Asn228Ser	3 (het) 3 (het)	1 0.918	del tol	DC DC	*C. elegans* *C. elegans*	- -	M	European	N	2 yrs 6 mo	FSGS	CsA	ND	novel [5]	Flui
A103	c.683A>G c.856C>T	p.Asn228Ser p.Leu286Phe	3 (het) 5 (het)	0.918 0.997	tol del	DC DC	*C. elegans* *D. melanogaster*	- -	F	European	N	1 yr 3 mo	FSGS	SR, CsA-PR	ND	[5] novel	Flui Flui
A4285-24	c.890T>C	p.Tyr297Cys	5 (hom)	0.997	Del	DC	*C.elegans*	-	M	Arabic	Y	5 mo	ND	ND	MC	novel	Flui
PLCE1																	
A4622-21	Deletion of Ex 2-20	-	Exon 2 - 20 (hom)	NA	NA	NA	NA	-	F	ND	Y	ND	FSGS (8 mo)	SR, CP-NR	ND	novel	CNV
A1802	c.961C>T	p.Arg321*	2 (hom)	NA	NA	NA	NA	-	F	Indian	Y	1 mo	MCNS (2 mo)	ND	Recurrent pancreatitis	[5b]	SSe
A1678-21	c.2576_2577insT	p.Gln859Hisfs*31	9 (hom)	NA	NA	NA	NA	-	M	Turkish	Y	7 yrs 4 mo	DMS	ND	ND	novel	Flui
A1393-21	c.3058C>T	p.Gln1020*	9 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	8 mo	FSGS (8 mo)	PD (9 mo)	ND	[30]	SSe
A3424-21	c.3058C>T	p.Gln1020*	9 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	2 mo	ND	ND	ND	[28]	Flui
A3233-27	c.3169C>T	p.Arg1057*	10 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	2 yrs 5 mo	ND	SR, CP-NR	ND	novel	Flui
A3617-25	c.3379_3380delAA	p.Asn1127*	11 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	7 mo	cFSGS (8 mo)	ND	ND	novel	Flui
A3317-45	c.3736C>T	p.Arg1246*	14 (hom)	NA	NA	NA	NA	AA=0/AG=1/GG=4189	F	Turkish	Y	1 yr 5 mo	ND	SR	ND	[57]	Flui
F722-21	c.3736C>T	p.Arg1246*	14 (hom)	NA	NA	NA	NA	AA=0/AG=1/GG=4189	F	White	N	6 mo	FSGS (6 mo)	SR	ASD	[57]	Flui
A4997-22	c.4451C>T	p.Ser1484Leu	19 (hom)	1	del	DC	*S. cerevisiae*	-	M	Turkish	Y	5 yrs	FSGS	Steroids-PR	Prader Willi syndrome, ID	[18]	Flui
A3921-22	c.4506+2T>C	splice	Intron19 (hom)	NA	NA	NA	NA	-	F	ND	Y	6 mo	FSGS (6 mo)	ND	ND	novel	Flui
A3510-21	c.4600A>G	p.Lys1534Glu	20 (hom)	0.998	del	DC	*S. cerevisiae*	-	F	Turkish	Y	1 yr 5 mo	ND	ND	ND	novel	Flui
A59-21	c.4887delA	p.Ala1630Glnfs*40	21 (hom)	NA	NA	NA	NA			Turkish	Y	7 mo	FSGS (1 yr)	ND	ND	novel	Flui
A3217-21 -22	c.4978_4981delCAGA	p.Gln1660Leufs*9	23 (hom)	NA	NA	NA	NA	-	Arabic F M		Y	ND 1 yr	ND ND	ESRD (4 yrs) ESRD (2 yrs)	ND Bilat. clubfoot	novel	SSe
A1450-23	c.5357G>T	p.Arg1786Ile	25 (hom)	1	del	DC	*S. cerevisiae*	-	F	Arabic	Y	9 mo	ND	ND	ND	novel	SSe
A3869-24	c.5521A>G	p.Lys1841Glu	26 (hom) m, p	1	del	DC	*S. cerevisiae*	-	M	Arabic	Y	7 mo	FSGS (9 mo)	ND	ND	novel	Flui
A3024-22	c.5630C>T	p.Ser1877Phe	27 (hom)	1	del	DC	*S. cerevisiae*	-	M	Turkish	Y	1 yr 5 mo	ND	ND	ND	novel	Flui
A4286-22	c.5906G>A	p.Arg1969Gln	28 (hom)	0.723	tol	DC	*M. musculus*	-	F	Arabic	Y	2 yrs 3 mo	FSGS	ESRD (2 yrs 6 mo)	ND	novel	Flui
A1420-21 -23	c.5951_5953delACA	p.Asn1984del	28 (hom)	NA	NA	NA	NA	-	Arabic F F		Y	ND	FSGS (2 yrs 7 mo) FSGS (4 yrs)	SR, CP-NR SR, CP-NR	Cataract ND	novel	Flui
A5018-21	c.6004-1G>A	splice	Intron 28 (hom)	NA	NA	NA	NA	-	M	Arabic	N	5 mo	ND	ACE-I	ND	novel	Flui
A2267	c.6342+3A>G	splice	Intron 30 (hom)	NA	NA	NA	NA	-	M	Pakistani	Y	5 mo	FSGS	ND	Club foot	novel	Flui
COQ6																	
A3600-21 -22	c.1058C>A	p.Ala353Asp	9 (hom)	0.999	del	DC	*S. cerevisiae*	-	M M	White	Y	4 yrs ND	FSGS (4 yrs) ND	CsA ND	ND ND	[6]	Flui
A5261-21	c.1058C>A	p.Ala353Asp	9 (hom)	0.999	del	DC	*S. cerevisiae*	-	M	Turkish	N	3 yrs 2 mo	FSGS (3 yrs 10 mo)	CsA, CP	ND	[6]	Flui
A938-21	c.1154A>C c.1235A>G	p.Asp385Ala p.Tyr412Cys	11 (het) 12 (het)	0.999 1	del del	DC DC	*S. cerevisiae* *S. cerevisiae*	- GG=0/GA=1/AA=4299	M	European	N	4 yrs 6 mo	MPGN (4 yrs 6 mo)	CsA	ND	novel novel	Flui
DGKE																	

A4431	c.610delA	p.Thr204Glnfs*6	3 (hom, p, m)	NA	NA	NA	NA	-		Turkish	Y				[8]	Flui
-21									F			17 yrs	FSGS	ESRD (22 yrs)	ND	
-22									F			8 yrs	MPGN (16 yr)	ND	ND	
A1419-21	c.1632T>G	p.Tyr544*	12 (hom)	NA	NA	NA	NA	-	F	Arabic	Y	1 yr 7 mo	ND	ND	ND	novel	
SMARCAL1																
A3146-21	c.49C>T	p.Arg17*	4 (het)	NA	NA	NA	NA	-	F	European	N	9 yrs	FSGS (10 yrs)		ID	[21]	Flui
	c.836T>C	p.Phe279Ser	5 (het)	0.985	tol	DC	*C. intestinalis*	-								[58]
A3877-21	c.68G>T	p.Arg23Leu	4 (het)	1	del	DC	*C. elegans*	-	M	Indian	N	6 yrs	MCNS (10 yrs), FSGS (ND)	CP-PR	deafness	novel	Flui
	c.1129G>C	p.Glu377Gln	7 (het)	0.425	tol	PMP	*D. rerio*	CC=1/CG=76/GG=4223								[59]
F979-21	c.836T>C	p.Phe279Ser	4 (het, m)	0.995	NA	NA	*C. intestinalis*	-	F	Arabic-Asian	N	4 yrs	FSGS+MesP (5 yrs 5 mo)	SR, CP-NR, CsA-PR, dialysis at 8 yrs		[58]	Flu
	c.1930C>T	pArg644Trp	12 (het, p)	1	NA	NA	*C. elegans*									[21]
A968-21	c.1129G>C	p.Glu377Gln	7 (hom)	0.425	tol	PMP	*D. rerio*	CC=1/CG=76/GG=4223	F	African	N	10 yrs	FSGS (10 yrs)	SR, CP	ND	[59]	Flui
A925-21	c.1736C>A	p.Ser579*	12 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	3 yrs	FSGS (4 yrs)	deceased (7 yrs)	SIDD, NHL	[21]	Flui
A4162-21	c.1736C>A	p.Ser579*	12 (hom)	NA	NA	NA	NA	-	F	European	N	4 yrs	FSGS (4 yrs)	SR, CsA	VSD, PDA, celiac disease, HU	[21]	Flui
F310	c.1756C>T	p.Arg586Trp	12 (hom)	1	del	DC	*C. elegans*	-		Italian						[21]	SSe
-21									M		Y	12 yrs	ND	ESRD (22 yrs), NTX (22 yrs)	SED		
-22									M			8 yrs	ND	ESRD (12 yrs), deceased (12 yrs) ND	SED		
-23									M			11 yr	ND		SED		
A1683-21	c.1756C>T	p.Arg586Trp	12 (hom ,m,p)	1	del	DC	*C. elegans*	-	M	White	Y	7 yrs	FSGS (7yrs)	SR, CP-NR	BLC, macular pigment anomaly	[21]	Flui
A3623-21	c.1756C>T	p.Arg586Trp	12 (hom)	1	del	DC	*C. elegans*	-	F	Indian	N	7 yrs	FSGS (8 yrs)	ND	SS	[21]	Flui
A3700-21	c.1756C>T	p.Arg586Trp	12 (hom)	1	del	DC	*C. elegans*	-	M	Indian	N	10 yrs	FSGS (10 yrs)	ND	ND	[21]	Flui
A3090-21	c.1859G>A	p.Trp620*	13 (het)	NA	NA	NA	NA	-	M	European	N	4 yrs	FSGS (4 yrs)	ND	Café au Lait maculas, SS	novel	Flui
	c.2542G>T	p.Glu848*	18 (het)	NA	NA	NA	NA	-								[21]
A3922-22	c.1860G>A	p.Trp620*	13 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	8 yrs	FSGS (8 yrs)	SR, CP-PR	ND	novel	Flui
A4708-21	c.1928T>C	p.Leu643Pro	13 (het)	0.999	del	DC	*D. melanogaster*	-	M	White	N	10 yrs	cFSGS (10 yrs)	RTX	Bilateral hip subluxation	novel	Flui
	c.2542G>T	p.Glu848*	18 (het)	NA	NA	NA	NA									[21]
A4380-21	c.2187A>C	p.Leu729Phe	15 (hom)	0.999	del	DC	*C. intestinalis*	-	M	African-Arabic	ND	5 yrs	ND	SR	URA, SS, FD, SED, HA	novel	Flui
A5017-29	c.2207delT	p.Val736Glyfs*76	15 (hom)	NA	NA	NA	NA	-	M	Yemeni	Y	6 yrs	ND	SR	GR	novel	Flui
A3222-22	c.2459G>T	p.Arg820Leu	16 (het)	1	Del	DC	C. elegans	-	F	European	N	1 yrs 11 mo	FSGS 2 yrs 10 mo	ND	SS, FD	[21]	Flui
	c.2542G>T	p.Glu848*	17 (het)	0.085	NA	NA	X. tropicalis	-								[21]
MYO1E																
A3661-21	c.554A>G	p.Asp185Gly	7 (het)	0.866	tol	DC	*C. intestinalis*	CC=0/CT=15/TT=4275	F	Burmese	N	3 yrs	ND	ND	ND	novel	Flui
	c.2464T>A	p.Leu822Met	22 (het)	0.075	tol	DC	*D. rerio*	-								novel
A3163-21	c.1162G>C	p.Asp388His	11 (hom)	1	del	DC	*C. intestinalis*	-	F	Turkish	N	7 yrs	FSGS (10 yrs)	SR, CP-NR, MMF-NR	ND	novel	Flui
A3191-21	c.1567C>T	p.Arg523Trp	15 (het)	1	del	DC	*C. elegans*	-	M	Australian	ND	6 mo	ND	ND	ND	novel	Flui
	c.3094_3097delACAA	p.The103Profs*73	27 (het)	NA	NA	NA	NA									novel
F878	c.1807_1808dupGT	p.Lys604*	18 (hom)	NA	NA	NA	NA	-	F	Syrian	Y	1 yr 8 mo	FSGS	SR, CsA-NR	ND	novel	Flui
A3556-21	c.1978C>T	p.Gln660*	19 (hom)	NA	NA	NA	NA	-	M	India	Y	2 mo	MCNS (3 yrs)	SR	ND	novel	Flui
LAMB2																
A2356-23	c.736C>T	p.Arg246Trp	7 (hom)	1	del	DC	*C.elegans*	-	M	Asian	Y	14 mo	Finnish type	ND	blindness, HU	[11]	Flui
A2073-21	c.737G>A	p.Arg246Gln	7 (het, p)	1	del	DC	*C. elegans*	-	M	African	N	2 mo	DMS (2 mo)	ND	ND	[11]	Flui
	c.1477delT	p.Cys493Alafs*4	11 (het, m)	NA	NA	NA	NA	-		American						novel
A5019-23	c.928T>C	p.Cys310Arg	8 (het, p)	1	del	DC	*C. elegans*	-	M	Malaysian	N	5 mo	FSGS (5 mo)	ND	ND	novel	Flui
	c.4198_4199delCT	p.Leu1400Glufs*7	26 (het, m)	NA	NA	NA	NA	-								novel
A4683-21	c.1144G>C	p.Ala382Pro	9 (hom)	0.81	tol	DC	*M. musculus*	-	F	Egyptian	Y	1 yr	No Bx		ID, Nystagmus	novel	Flui

A5143-24	c.1178_1180delTCT	p.Phe393del	9 (hom)	NA	NA	NA	NA	-	F	Aravic	Y	1 mo	FSGS (8 mo)	ESRD (1 yr)	Nystagmus, congenital cataract	novel	Flui
A1613-21	c.1405+1G>A	splice	10 (hom)	NA	NA	NA	NA	-	M	German	N	1 mo	dilated tubules with microcysts	ND	HU	[60]	Flui
A3905-21	c.1405+2insA c.5240del	splice p.Asp1747Alafs*74	10 (het, p) 31 (het, m)	NA	NA	NA	NA	- -	F	Indian	N	3 days	DMS (on autopsy)	deceased (2 mo)	ND	novel novel	Flui
A2373-21	c.1452T>A	p.Cys484*	11 (hom, m, p)	NA	NA	NA	NA	-	M	ND	ND	CNS	ND	ND	Pierson Syndrome	novel	SSe
A5284-21	c.1731+1G>A	splice	13 (hom)	NA	NA	NA	NA	-	F	Asian	Y	1 yr	ND	SR	MC, ID, SS, FD	novel	Flui
A5136-24	c.3100_3101delAG c.4734dupA	p.Ser1034Leufs*20 p.Asp1579Argfs*20	21 (het) 28 (het)	NA NA	NA NA	NA NA	NA NA	- -	M	ND	N	2 mo	ND	ND	ND	novel novel	Flui Flui
A2263-23	c.4537C>T	p.Gln1513*	27 (hom)	NA	NA	NA	NA	-	M	Arabic	Y	3 mo	ND	ND	MC, micro-coria, microph-thalmia	novel	Flui
A4381-21	c.4582insG	p.Glu1528Glyfs*2	28 (hom)	NA	NA	NA	NA	-	M	Egyptian	Y	2 mo	ND	deceased	FD	novel	Flui
A4981-21	c.5158_5162delAAGGC	p.Lys1720Profs*16	31 (hom)	NA	NA	NA	NA	-	M	Turkish	Y	4 mo	ND	ND	FD, microcoria, glaucoma	novel	Flui
CUBN																	
A807-21	c.348+2T>C	splice	3 (het, m)	NA	NA	NA	NA	GG=0/GA=5/AA=4295	M	European	N	11 yr 11 mo	FSGS (11 yr 11 mo)	SRNS, CsA	Asthma	novel	Flui
	c.5069C>T	p.Ala1690Val	34 (het, p	0.994	tol	DC	*D. rerio*	AA=0/AG=20/GG=4280								novel	Flui
A994-21	c.5518G>A	p.Gly1840Ser	37 (hom)	0.430	del	PMP	*D. melanogaster*	-		Hispanic	N	6 yr	FSGS (6 yr)	SRNS	ND	novel	Flui
A3316-21	c.4036C>T c.4610C>T	p.Gln1346* p.Ser1537Phe	28 (het, p) 31 (het, m)	NA 0.994	NA del	NA DC	NA *D. rerio*	- -	F	Turkish	N	ND	ND	ND	ND	novel novel	Flui Flui
A4007-21	c.5209+1G>C c.6928_6934delGAGGTTA	Splice p.Glu2310Cysfs*3	35 (het, m) Exon 45 (het, p)	NA NA	NA NA	NA NA	NA NA	- -	M	European	N	4 yrs 10 mo	ND	ND	ND	novel [61]	Flui Flui
ITGB4																	
A1697-21	c.469+5G c. 2990C>A	splice p.Pro997His	5 (het) 26 (het)	NA 0.975	NA del	NA DC	NA *D.rerio*		F	African-German	N	9 yrs 9 mo	FSGS (9 yrs 9 mo)	SR, CsA	ND	novel	Flui
PDSS2																	
A3853-22	c.1145C>T	p.Ser382Leu	8 (hom)	1	del	DC	*D. rerio*	AA:AA=0/AG=0/GG=2203,	M	African-arabic	Y	1 yr 11 mo	ND	ND	Cerebral palsy, ID	[1/]	Flui
A3264-21	c.1151C>A	p.Ala384Asp	8 (hom)	1	del	DC	*M. musculus*	-	M	Arabic	N	birth	ND	ND	ND	novel	Flui
CFH																	
A3655-21	c.1064A>G	p.Tyr355Cys	8 (hom)	1	del	PMP	*X. tropicalis*	-	F	Indian	N	9 yrs 4 mo	Diffuse proliferative GN (10 yrs)	Steroids-PR	ND	[62]	Flui
ITGA3																	
A3338-21	c.428G>A	p.Arg143His	4 (hom)	0.882	del	DC	*D. melanogaster*	-	M	Arabic	Y	4 yrs	ND	ND	ichthyosis, ectodermal dysplasia, Mb. Hirsch-sprung, TS, kidney dysplasia	novel	Flui
A1605	c.2593delG	p.Asp865Thrfs*38	21 (hom)	N/A	N/A	N/A	N/A	-	M	Turkey	Y	2 mo	ND	ND	Tachydyspnoe	novel	Flui

AA=African american. ACE-I=ACE-inhibitors. AN=Alport nephropathy. ASD=atrial septal defect. BLC=bilateral cryptorchidism. cFSGS=collapsing focal segmental glomerulosclerosis. CNS=congenital nephrotic syndrome. CNV=Copy Number Variant. CP=cyclophosphamide. CsA=cyclosporine A. DC=disease causing. DDS=Denys-Drash-syndrome. del=deleterious. DMS=diffuse mesangial sclerosis. EA=European American. ESRD=end-stage renal disease. F=female. FD=facial dysmorphism. Flui=Fluidigm. FSGS=focal segmental glomerulosclerosis. GBM=glomerular basement membrane. GN=glomerulonephritis. GR=growth retardation. HA=heart anomalies. HD=hemodialysis. het=heterozygous. hom=homozygous. HU=hematuria. ID=Intellectual disability. m=maternal. M=male. MC= Microcephalus.

MCNS=minimal change nephrotic syndrome. MesP=mesangial proliferation. MMF=mycophenolat mofetil. mo=month. MPGN=membranoproliferative glomerulonephritis. N=no. NA=not applicable. ND=no data or not done. NHL=non-Hodgkins lymphoma. NR=No response. p=paternal. NTX=kidney transplantation. PAS=pulmonary artery stenosis. PD=peritoneal dialysis. PDA=persistent ductus arteriosus. PMP=polymorphism. PR=partial response. RTX=rituximab. SED=spondylo-epiphyseal dysplasia. SIDD=Schimke immuno-osseous dysplasia. SR=steroid-resistant nephrotic syndrome. SS=short stature. SSe=Sanger sequencing. Tac=tacrolimus. tol=tolerated. TS=tracheal stenosis. URA=unilateral renal agenesis. UTA=urinary tract anomalies. VSD=ventricular septal defect. VUR=vesico-ureteral reflux. WT=Wilms tumor. Y=yes. yrs=years. "-"=no entry.

#Not shown in this table are families with identified disease causing mutations that were published previously by our group: 61 families with *NPHS1* mutations,[41, 45, 48, 63-65] 42 families with *NPHS2* mutations,[38, 41, 63, 65, 66] 50 families with *WT1* mutations,[41, 67-69] 16 families with mutations in *PLCE1*,[18, 65, 70] seven families with *LAMB2* mutations,[41, 65, 71-73] five families with *COQ6* mutations,[6] one family with *TRPC6* mutation,[74] three families with *ITGA3* mutations,[9] one family with *CUBN* mutation,[7] three families with *ADCK4* mutations,[1] one family with *ARHGDIA* mutation.[75]

Supplemental Table 4. Genotypes and phenotypes of mutations in *WT1* detected by Sanger sequencing and high-throughput sequencing in 35 out of 1,783 families with SRNS.[#]

Family-Individual	Nucleotide change	Amino acid change	Legacy change	Exon (Zygosity, Segregation)	SIFT	Amino acid conservation to species	EVS allele frequencies in EA	Gender	Ethnic origin	Parental consanguinity	Age of onset	Biopsy (at age)	Therapy and response	Extrarenal Manifestations	Reference	Method
WT1																
A5218-21	c.872+1G>A	splice	IVS4+1G>A	4 (het)	NA	NA	-	M	Arabic	Y	6 mo	bilateral WT	ACE-I, chemotherapy, tumorectomy	ND	novel	SSe
A3167-22	c.1001+6T>G	splice	IVS5+1T>G	5 (het)	NA	NA	-	M	Egyptian	Y	3 yrs	ND	SR	ND	novel	SSe
A1987-21	c.1178G>A	p.Cys376Tyr	p.Cys308Tyr	7 (het)	NA	*S. cerevisiae*	-	F	White	N	29 yrs	ND	ND	ND	novel	Flui
A4602-21	c.1283G>A	p.Cys428Tyr	p.Cys360Tyr	8 (het)	del	*S. cerevisiae*	-	F	White	N	2 mo	ND		ND	[76]	SSe
A3808-21	c.1300C>T	p.Arg434Cys	p.Arg366Cys	8 (het)	del	*S. cerevisiae*	-	M	White	N	1 mo	DMS (1 mo)	SR	UTM	[27]	SSe
A2624-21	c.1301G>A	p.Arg434His	p.Arg366His	8 (het)	del	*S. cerevisiae*	-	M	Asian	N	prenatal	ND	deceased in utero at 31 weeks of gestation	Ambiguous genitalia	[77]	SSe
A3194-21	c.1301G>A	p.Arg434His	p.Arg366His	8 (het)	del	*S. cerevisiae*	-	F	Turkish	N	2 mo	ND	ND	MC	[77]	SSe
A3940-21	c.1301G>A	p.Arg434His	p.Arg366His	8 (het)	del	*S. cerevisiae*	-	F	Pakistani	N	1 mo	ND	ND	ID	[77]	SSe
A3947-21	c.1301G>A	p.Arg434His	p.Arg366His	8 (het)	del	*S. cerevisiae*	-	M	Turkish	N	7 days	ND	Albumin	UTM	[77]	SSe
A4335-21	c.1301G>A	p.Arg434His	p.Arg366His	8 (het)	del	*S. cerevisiae*	-	F	White	N	<30 days	DMS (1 mo), MCNS (2 yrs)	ND	UTM	[77]	SSe
A5212-21	c.1337C>G	p.Thr446Arg	p.Thr378Arg	8 (het)	del	*S. cerevisiae*	-	F	European	N	5 yrs	FSGS (5 yrs)	SR, CsA	ND	[69]	SSe
A2370-21	c.1339+5G>A	splice	IVS8+5G>A	8 (het)	NA	NA	-	M	Arabic	Y	5 mo	ND	ND	ND	[78]	Flui
A2364-21	c.1357T>C	p.Cys453Arg	p.Cys385Arg	9 (het)	del	*S. cerevisiae*	-	M	European	N	1 yr	DMS (1 yr)	ND	BLC	[79]	SSe
A4160-21	c.1366T>C	p.Cys456Arg	p.Cys388Arg	9 (het)	del	*S. cerevisiae*	-	M	European	N	3 mo	ND	ND	UTM, UTM	[38]	SSe
A3545-21	c.1369C>T	p.Glu457*	p.Glu389*	9 (het)	NA	NA	-	F	White	N	7 mo	DMS (1 yr)	ND	ND	[80]	SSe
A3015-21	c.1373G>A	p.Arg458Gln	p.Arg390Gln	9 (het)	del	*D. rerio*	-	F	Hispanic	ND	10 yrs	FSGS (ND)	ND	ND	novel	SSe
A943-21	c.1384C>T	p.Arg462Trp	p.Arg394Trp	9 (het)	del	*S. cerevisiae*	-	M	German	N	2 yrs	Sclerotic GN	ND	DDS, right WT, BLC	[77]	SSe
A3526-21	c.1384C>T	p.Arg462Trp	p.Arg394Trp	9 (het)	del	*S. cerevisiae*	-	M	Hispanic	N	7 yrs	WT right	Chemotherapy	ND	[77]	SSe
A3715-21	c.1384C>T	p.Arg462Trp	p.Arg394Trp	9 (het)	del	*S. cerevisiae*	-	M	Arabic	Y	6 mo	ND	ND	FD, UTM	[77]	SSe
A5022-26	c.1384C>T	p.Arg462Trp	p.Arg394Trp	9 (het)	del	*S. cerevisiae*	-	M	Arabic	N	3 yrs	DMS (3 yrs)	ND	UTM	[77]	SSe
A2270-21	c.1384C>T	p.Arg462Trp	p.Arg394Trp	9 (het)	del	*S. cerevisiae*	-	F	White/Spanish	N	birth	DMS	ND	ND	[77]	Flui
A2072-21	c.1385G>A	p.Arg462Gln	p.Arg394Gln	9 (het)	del	*S. cerevisiae*	-	M	African American	N	1 week	ND	ND	Hypospadias, BLC	[69]	SSe
A4925-21	c.1390G>A	p.Asp464Asn	p.Asp396Asn	9 (het)	del	*S. cerevisiae*	-	F	White	N	1 mo	DMS (1 mo)	ND	ND	[77]	SSe
A1726-21	c.1393C>T	p.His465Tyr	p.His397Tyr	9 (het)	del	*C. elegans*	-	M	Hungarian	N	9 mo	MCNS (1 yr)	ND	BLC	novel	Flui
A3161-21	c.1432+4C>T	splice	IVS9+4C>T	9 (het)	NA	NA	-	F	Turkish	N	9 yrs	FSGS (9 yrs)	SR, CsA-NR	ND	[81, 82]	SSe
A3671-21	c.1432+4C>T	splice	IVS9+4C>T	9 (het)	NA	NA	-	F	Indian	N	5 yrs	FSGS (7 yrs)	SR	ND	[81, 82]	SSe
A3911-21	c.1432+4C>T	splice	IVS9+4C>T	9 (het)	NA	NA	-	F	White	N	3 yrs	Thin GBM	SR	UTM	[81, 82]	SSe
A1684-21	c.1432+4C>T	splice	IVS9+4C>T	9 (het)	NA	NA	-	F	Hispanic	N	2 yrs 5 mo	FSGS (3 yrs)	SR, CP-NR, Tac	ND	[81, 82]	Flui
A2963-21	c.1432+5G>A	splice	IVS+5G>A	9 (het)	NA	NA	-	M	Hispanic	ND	34 yrs	FSGS	ND	ND	[79]	SSe
A1271-21	c.1432+5G>A	splice	IVS+5G>A	9 (het)	NA	NA	-	M	ND	N	7 mo	DMS	ACE-I	FD, UTM, HA	[79]	SSe
A3707-21	c.1432+5G>A	splice	IVS+5G>A	9 (het)	NA	NA	-	F	Hispanic	N	5 yrs	FSGS (9 yrs)	SR	UTM, delayed bone age	[79]	SSe
A3770-21	c.1432+5G>A	splice	IVS+5G>A	9 (het)	NA	NA	-	F	Arabic	N	1 yr	DMS (3 yrs)	Steroids	ND	[79]	SSe
A3868-22	c.1432+5G>A	splice	IVS+5G>A	9 (het)	NA	NA	-	F	Arabic	N	3 yrs	FSGS+MPGN (3 yrs)	SR, CP-NR	ND	[79]	SSe
A5139-21	c.1432+5G>A	splice	IVS+5G>A	9 (het)	NA	NA	-	F	Russian	N	ND	ND	SR, CsA	UTM, VUR	[79]	SSe
A5141-21	c.1432+5G>A	splice	IVS+5G>A	9 (het)	NA	NA	-	F	White	N	4 yrs	FSGS (4 yrs)	SR, CP, CsA, Tac	ND	[79]	SSe

AA=African american. ACE-I=ACE-inhibitors. AN=Alport nephropathy. ASD=atrial septal defect. BLC=bilateral cryptorchidism. cFSGS=collapsing focal segmental glomerulosclerosis. CNS=congenital nephrotic syndrome. CNV=Copy Number Variant. CP=cyclophosphamide. CsA=cyclosporine A. DC=disease causing. DDS=Denys-Drash-syndrome. del=deleterious. DMS=diffuse mesangial sclerosis. EA=European American. ESRD=end-stage renal disease. F=female. FD=facial dysmorphism. Flui=Fluidigm. FSGS=focal segmental glomerulosclerosis. GBM=glomerular basement membrane. GN=glomerulonephritis. GR=growth retardation. HA=heart anomalies. HD=hemodialysis. het=heterozygous. hom=homozygous. HU=hematuria. ID=Intellectual disability. m=maternal. M=male. MC= Microcephalus. MCNS=minimal change nephrotic syndrome. MesP=mesangial proliferation. MMF=mycophenolat mofetil. mo=month. MPGN=membranoproliferative glomerulonephritis. N=no. NA=not applicable. ND=no data or not done. NHL=non-Hodgkins lymphoma. NR=No response.

p=paternal. NTX=kidney transplantation. PAS=pulmonary artery stenosis. PD=peritoneal dialysis. PDA=persistent ductus arteriosus. PMP=polymorphism. PR=partial response. RTX=rituximab. SED=spondylo-epiphyseal dysplasia. SIDD=Schimke immuno-osseous dysplasia. SR=steroid-resistant nephrotic syndrome. SS=short stature. SSe=Sanger sequencing. Tac=tacrolimus. tol=tolerated. TS=tracheal stenosis. URA=unilateral renal agenesis. UTA=urinary tract anomalies. VSD=ventricular septal defect. VUR=vesico-ureteral reflux. WT=Wilms tumor. Y=yes. yrs=years. "-"=no entry.

#Not shown in this table are families with identified disease causing mutations that were published previously by our group: 61 families with *NPHS1* mutations,[41, 45, 48, 63-65] 42 families with *NPHS2* mutations,[38, 41, 63, 65, 66] 50 families with *WT1* mutations,[41, 67-69] 16 families with mutations in *PLCE1,*[18, 65, 70] seven families with *LAMB2* mutations,[41, 65, 71-73] five families with *COQ6* mutations,[6] one family with *TRPC6* mutation,[74] three families with *ITGA3* mutations,[9] one family with *CUBN* mutation,[7] three families with *ADCK4* mutations,[1] one family with *ARHGDIA* mutation.[75]

Supplemental Table 5. Genotypes and phenotypes of mutations in *TRPC6, ARHGAP24, ~~and~~ INF2* and *LMX1B* detected by Sanger sequencing and high-throughput sequencing in 22 out of 1,783 families with SRNS.[#]

Family-Individual	Nucleotide change	Amino Acid change	Exon (Zygosity, Segregation)	Poly-phen	SIFT	Mutation-taster	Amino acid conservation to species	EVS allele frequencies in EA	Gender	Ethnic origin	Parental Consanguinity	Age of onset	Biopsy	Therapy and response	Extrarenal Manifestations	Reference	Method
TRPC6																	
F505-41	c.395T>C	p.Met132Thr	2 (het)	0.801	del	DC	*D. rerio*	-	M	European	N	8 yrs 6 mo	MCNS (8 yrs) with FSGS	ND	Hypospadias	[74]	SSe
A1657	c.428A>G	p.Asn143Ser	2 (het)	0.998	del	DC	*D. rerio*	EA:CC=0/CT=0/TT=4299 AA:CC=0/CT=3/TT=8598	F	African American	N	8 yrs	FSGS (8yrs)	ND	ND	[83]	Flu
A5246-21	c.1195C>T	p.Arg399*	4 (het)	NA	NA	NA	NA	-	M	European	N	35 yrs	FSGS (35 yrs)	ND	ND	novel	Flui
A3310-21	c.2536-2539delAATA	p.Asn846Valfs*12	11 (het)	NA	NA	NA	NA	-	M	Turkish	N	2 yrs 7 mo	FSGS (2 yrs 9 mo)	ND	ND	novel	Flui
F1174-21	c.2605C>T	p.Gln869*	12 (het)	NA	NA	NA	M. musculus	-	M	European	N	13 yrs 1 mo	ND	SR transplant at 15 yrs	Factor V leiden	novel	Flui
A2401-31	c.2656G>T	p.Glu886*	13 (het)	NA	NA	NA	NA	-	F	European	N	ND	ND	ND	ND	novel	Flui
A664-21	c.2683C>T	p.Arg895Cys	13 (het)	0.905	del	DC	*D. rerio*	-	F	Jewish	N	3 yrs 2mo	ND	ND	ND	[83]	Flui
A4717-21	c.2684G>T	p.Arg895Leu	13 (het)	0.905	del	DC	*D. rerio*	-	M	Indian	M	1 yr 6 mo	cFSGS (1 yr 8 mo)	ND	ND	[84]	Flui
ARHGAP24																	
A4000	c.120G>A	p.Trp40*	2 (het)	NA	NA	NA	NA	-		White	N					novel	Flui
-21									F			32 yrs	FSGS	HU	ND		
-22									F			ND	ND	ND	ND		
INF2																	
A1949-21	c.317G>C	p.Arg106Pro	2 (het)	1	del	DC	*D. rerio*	-	M	African American	N	6 yrs	FSGS (6 yrs)	SR	ND	[85]	Flui
A1927	c.451T>C	p.Cys151Arg	3 (het)	0.987	del	DC	*X. tropicalis*	-		American	N					[86]	Flui
-11									M			ND	ND	ND	Charcot-Marie Tooth		
-21									M			11 yr	FSGS (11 yr)	SR			
A4722-21	c.530G>A	p.Arg177His	4 (het)	0.999	del	DC	*D. rerio*	-	M	European	N	27 yrs	FSGS (27 yrs)	SR	ND	[85]	Flui
A3029-21	c.550G>A	p.Glu184Lys	4 (het, p)	0.999	del	DC	*D. rerio*	-	M	European	N	ND	FSGS (15 yrs)	SR, CP	ND	[24]	Flui
A5091	c.550G>A	p.Glu184Lys	4 (het)	0.999	del	DC	*D. rerio*	-		Indian	N					[24]	Flui
-23									M			27 yrs	FSGS (27 yrs)	SR, RTX	ND		
-24									M			30 yrs	FSGS (30 yrs)	SR, ACE-I, RTX	ND		
A828	c.641G>A	p.Arg214His	4 (het)	0.999	del	DC	*D. rerio*	-		Serbian	N					[24]	Flui
-32									M			18 yrs	FSGS (29 yrs)	SR, ACE-I	ND		
-33									M			ND	ND	ND	ND		
-41									M			ND	ND	ND	ND		
A3184-21	c.652C>T	p.Arg218Trp	4 (het, m)	1	del	DC	*D. rerio*	-	M	European	N	12 yrs	FSGS (13 yrs)	SR, CP, CsA	ND	[24]	Flui
A4017-21	c.653G>A	p.Arg218Gln	4 (het, m)	1	del	DC	*D. rerio*	-	M	European	Y	22 yrs	FSGS (22 yrs)	SR	HTN	[24]	Flui
A5050 -21	G.658G>A	p.Glu220Lys	4 (het, p)	0.999	del	DC	*D. rerio*	-	M	Bulgarian	N	17 yrs	FSGS (17 yrs)	SR, CsA	ND	[24]	Flui
LMX1B																	
A54-21	c.737G>A	p.Arg246Gln	4 (het)	1	del	DC	C. elegans	-	F	German	N	6 yrs	ND	SR	familial NS	[87]	Flui
A200-21	c.737G>A	p.Arg246Gln	4 (het)	1	del	DC	C. elegans	-	F	Turkish	Y	8 yrs	FSGS (8 yrs)	SR, ESRD and HD (9 yrs)	familial NS	[87]	Flui
A2175-21	c.737G>A	p.Arg246Gln	4 (het)	1	del	DC	C. elegans	-	M	Swiss	N	4 yrs	FSGS (4 yrs)	SR, CsA-PR, ACE-I	Factor-XII deficiency	[87]	Flui
A3180-21	c.737G>A	p.Arg246Gln	4 (het)	1	del	DC	C. elegans	-	F	German	N	18 yrs	FSGS (43 yrs)	HD (43 yrs), NTX (47 yrs)	ND	[87]	Flui
-22									M			32 yrs	FSGS (33 yrs)	HD (44 yrs)	ND		
-31									F			ND	FSGS (ND)	ACE-I	ND		
-32									F			ND	ND	ACE-I	ND		

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