SUPPLEMENTARY METHODS

DNA extraction: DNA was extracted from EDTA blood using QIAamp® DNA Blood Mini Kit (Qiagen, Germantown, MD), as per manufacturer instructions. The quality and the quantity of DNA extracted was assessed using NanoDrop™ spectrophotometer (Thermo Scientific, Wilmington, DE) and 1% gel electrophoresis before next generation sequencing.

Whole exome sequencing (WES): DNA libraries were prepared using 100 ng of DNA, physically sheared on an ultrasonicator (Covaris), followed by ligation of adapter sequences on to fragmented DNA to generate indexed libraries, and exome enrichment using TruSeq Exome kit (Illumina, San Diego, CA), as per manufacturer protocol. Enriched libraries were quantified by Qubit fluorometer (ThermoFisher) and their size distribution measured using Bioanalyzer (Agilent). Three of 34 samples underwent cluster generation using Illumina Cbot followed by paired end sequencing (2x100 bp) using flowcell v3 on Illumina HiSeq2000 platform. The remainder were sequenced (2x150 bp) on Illumina NovaSeq platform using S2/S4 flowcell.

Processing of sequenced reads: Paired-end sequenced reads were processed using the Dynamic Read Analysis for GENomics Bio-IT (DRAGEN, Illumina) platform. The reads were demultiplexed and then mapped and aligned to the reference genome (GRCh37; hg19) using the seed generation algorithm followed by Smith-Waterman algorithm. This was followed by variant calling using Haplotype Caller (Dragen), merging of individual variant call files (VCF) using VCFtools (*vcftools.sourceforge.net*) and annotation of merged VCFs using ANNOVAR (*annovar.openbioinformatics.org*).

Variant prioritization: Based on literature search, 89 genes were considered relevant for genetic testing in nephrotic syndrome [i,ii,iii,iv,v]. The list of genes, along with the mean coverage of exonic regions, is provided in **Suppl. Table S1**. Variants in these genes were considered potentially disease causing if they fulfilled one of the following criteria: *(i) rare and deleterious,* with rarity defined as minor allele frequency (MAF) of less than 0.1% in the population databases of 1000 Genomes Project [vi], Exome Aggregator Consortium (Exac) [vii] and Genome Aggregation Database (gnomAD) [viii]; and deleteriousness predicted by assertion of pathogenicity on at least two computational tools, including Polymorphism Phenotyping v2 (PolyPhen2; *http://genetics.bwh.harvard.edu/pph2/*), Sorting Intolerant from Tolerant (SIFT; *https://sift.bii.a-star.edu.sg/*), Mutation taster v2 (*http://www.mutationtaster.org/ChrPos.html*), Combined Annotation Dependent Depletion (CADD; *https://cadd.gs.washington.edu/*) and Genomic Evolutionary Rate Profiling score

(GERP_RS;*http://varianttools.sourceforge.net/Annotation/dbNSFP*), Eigen (*https://omictools. com/eigen-tool*), and where relevant, Human Splicing Finder v3.1 (*http://umd.be/HSF3/*); *(ii) novel and deleterious*, with novelty defined by absence in the two population databases as well as in the Database of Single Nucleotide Polymorphism (DbsNP; *https://www.ncbi.nlm.nih.gov/snp/*) and ClinVar (*https://www.ncbi.nlm.nih.gov/clinvar/*); or *(iii) reported,* in causative association with disease (congenital or steroid resistant nephrotic syndrome) in ClinVar (*https://www.ncbi.nlm.nih.gov/clinvar*) or Human Genome Mutation Database (HGMD; *http://www.hgmd.cf.ac.uk*), particularly if reported as 'pathogenic' or 'likely pathogenic' in ClinVar.

Variants shortlisted based on above criteria were excluded if any of the following conditions were fulfilled: *(i) high (≥0.1%) MAF in south Asian population* of ExAC; *(ii) failing to have causative phenotype*, such as a variant called in heterozygous state in a gene following autosomal recessive pattern of inheritance [ix]; or *(iii) low depth:* variant with read depth of <10x. Prioritised variants were classified from benign to pathogenic using the web-based clinical INTERpretation of VARiants (wINTERVAR; *http://wintervar.wglab.org/*), with or without modifications to follow the criteria outlined by the 2015 guidelines of the American College of Medical Genetics and Genomics (ACMG) [x].

Variant validation: Variants considered causative of disease were validated by Sanger sequencing using the ABI3730 genetic analyzer (Applied Biosystems). Sanger sequencing on parents' samples was used to confirm allele segregation for compound heterozygous variations.

Haplotype analysis of p.Gly867Asp mutant allele: The NGS data was used to obtain all single nucleotide polymorphisms flanking the mutation (±500 kbp). Variants with MAF ≥0.05% and significant difference in frequency (P<0.05) in patients with and without the mutation (p.Gly867Asp) were selected for haplotype analysis by Phase v2.1 (*http://stephenslab.uchicago.edu/phase/download.html*) to obtain haplotypes segregating with Gly867 and Asp867 associated alleles.

Supplementary Table SI Panel of 89 genes examined for association with congenital nephrotic syndrome along with coverage

Gene	*Disease*	*Inheritance*	*Transcript*	*Mean Coverage*
ACTN4	Glomerulosclerosis, focal segmental, 1	AD	NM_004924.5	109.40
ALG1	Congenital disorder of glycosylation, type Ik	AR	NM_019109.4	126.06
ALMS1	Alstrom syndrome	AR	NM_015120.4	104.55
ANKS6	Nephronophthisis 16	AR	NM_173551.4	46.96
ANLN	Focal segmental glomerulosclerosis 8	AD	NM_001284301.2	61.67
APOL1	End-stage renal disease, nondiabetic, susceptibility to Glomerulosclerosis, focal segmental, 4, susceptibility to	-	NM_145343.2	51.61
ARHGAP24	-	-	NM_001025616.2	111.73
ARHGDIA	Nephrotic syndrome, type 8	AR	NM_001301242.1	44.86
AVIL	-	-	NM_006576.3	65.40
CD151	Nephropathy with pretibial epidermolysis bullosa and deafness	-	NM_001039490.1	49.87
CD2AP	Glomerulosclerosis, focal segmental, 3	-	NM_012120.2	124.88
CFH	Complement factor H deficiency (hemolytic uremic syndrome, atypical, susceptibility to, 1)	AR|AD	NM_000186.3	134.46
CLCN5	Nephrolithiasis, type I: Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis	XLR|XLR	NM_001127898.3	77.52
COL4A1	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps	AD	NM_001845.5	126.02
COL4A3	Alport syndrome 2, autosomal recessive; Alport syndrome 3, autosomal dominant; hematuria, benign familial	AR|AD|AD	NM_000091.4	131.02
COL4A4	Alport syndrome 2, autosomal recessive| hematuria, familial benign	AR|AD	NM_000092.4	118.25
COL4A5	Alport syndrome 1, X-linked	XLD	NM_000495.4	90.51
COQ2	Coenzyme Q10 deficiency, primary, 1	AR	NM_015697.7	81.81
COQ6	Coenzyme Q10 deficiency, primary, 6	AR	NM_182476.2	149.02
COQ7	Coenzyme Q10 deficiency, primary, 8	AR	NM_016138.4	53.76
COQ8B	Nephrotic syndrome, type 9	AR	NM_001142555.2	52.54
COQ9	Coenzyme Q10 deficiency, primary, 5	AR	NM_020312.3	130.77
CRB2	Focal segmental glomerulosclerosis 9; ventriculomegaly with cystic kidney disease	AR|AR	NM_173689.6	41.61
CUBN	Finnish type	AR	NM_001081.3	136.24
CYP11B2	Hypoaldosteronism, congenital, due to CMO I deficiency; hypoaldosteronism, congenital, due to CMO II deficiency	AR|AR	NM_000498.3	121.62
DGKE	{Hemolytic uremic syndrome, atypical, susceptibility to, 7}| Nephrotic syndrome, type 7	AR|AR	NM_003647.2	58.76

E2F3	-	-	NM_001949.4	61.62
EMP2	Nephrotic syndrome, type 10	AR	NM_001424.5	45.32
EXT1	Chondrosarcoma| Exostoses, multiple, type 1	AR|AD	NM_000127.2	114.96
FAT1	-	-	NM_005245.3	75.17
FN1	Glomerulopathy with fibronectin deposits 2	AD	NM_001306129.1	127.17
G6PC	Glycogen storage disease	AR	NM_000151.3	126.16
GATA3	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	AD	NM_001002295.1	94.05
GFND1	Glomerulopathy with fibronectin deposits 1	AD	MIM:137950	-
GLA	Fabry disease| Fabry disease, cardiac variant	X-linked	NM_000169.2	92.44
IGAN1	{IgA nephropathy, susceptibility to, 1}	?AD	MIM:161950	-
IGAN2	{IgA nephropathy, susceptibility to, 2}	?AD	MIM:613944	-
INF2	Glomerulosclerosis, focal segmental, 5	AD	NM_001031714.3	63.42
ITGA3	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital	AR	NM_002204.3	52.16
ITGB4	Epidermolysis bullosa of hands and feet| Epidermolysis bullosa, junctional, non-Herlitz type| Epidermolysis bullosa, junctional, with pyloric atresia	AD|AR|AR	NM_001256876.1	101.91
KANK1	Cerebral palsy, spastic quadriplegic, 2	-	NM_001136191.2	70.34
KANK2	Nephrotic syndrome, type 16	AR	NM_001320269.1	45.10
KANK4	-	-	NM_006014.4	72.41
LAGE3	Galloway-Mowat syndrome 2, X-linked	XLR	NM_002292.3	24.05
LAMB2	-	-	NM_170708.3	126.50
LMNA	Cardiomyopathy, dilated, 1A| Charcot-Marie-Tooth disease, type 2B1| Emery-Dreifuss muscular dystrophy 2, autosomal dominant| Emery-Dreifuss muscular dystrophy 3, autosomal recessive| Heart-hand syndrome, Slovenian type| Hutchinson-Gilford progeria| Lipodystrophy, familial partial, type 2| Malouf syndrome| Mandibuloacral dysplasia| Muscular dystrophy, congenital| Restrictive dermopathy, lethal	AD|AR|AD|AR|AD| AR,AD|AD| AD|AR|AD|AR	NM_001282626.1	108.23
LMX1B	Nail-patella syndrome	AD	NM_001174146.1	56.96
LRP2	Donnai-Barrow syndrome	AR	NM_004525.2	130.34
MAFB	Multicentric carpotarsal osteolysis syndrome	AD	NM_005461.4	49.48
MAGI2	Nephrotic syndrome, type 15	AR	NM_001301128.1	66.23
MED28	-	-	NM_025205.4	79.14
MEFV	Familial Mediterranean fever | Familial Mediterranean fever	AD|AR	NM_000243.2	93.60
MT-TL1	-	-		-
MUC1	Medullary cystic kidney disease 1	AD	NM_002456.5	62.84

MYH9	Deafness, autosomal dominant 17| Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	AD|AD	NM_002473.5	112.51
MYO1E	Glomerulosclerosis, focal segmental, 6	AR	NM_004998.3	125.65
NEIL1	-	-	NM_001256552.1	62.27
NEU1	Sialidosis, type I| Sialidosis, type II	AR|AR	NM_000434.3	119.58
NPHP4	Nephronophthisis 4| Senior-Loken syndrome 4	AR|AR	NM_015102.4	52.17
NPHS1	Nephrotic syndrome, type 1	AR	NM_004646.3	112.08
NPHS2	Nephrotic syndrome, type 2	AR	NM_001297575.1	86.29
NUP107	Galloway-Mowat syndrome 7; nephrotic syndrome, type 11; ?ovarian dysgenesis 6	AR	NM_020401.3	60.98
NUP205	?Nephrotic syndrome, type 13	-	NM_015135.2	71.10
NUP93	Nephrotic syndrome, type 12	AR	NM_014669.4	56.54
NXF5	-	-	NM_032946.2	83.26
OCRL	Dent disease 2| Lowe syndrome	XLR; |XLR	NM_001587.3	94.16
OSGEP	Galloway-Mowat syndrome 3	AR	NM_017807.3	62.15
PAX2	Glomerulosclerosis, focal segmental, 7; papillorenal syndrome	AD; AD	NM_001304569.1	106.34
PDSS2	Coenzyme Q10 deficiency, primary, 3	AR	NM_020381.3	119.97
PLCE1	Nephrotic syndrome, type 3	AR	NM_016341.3	124.63
PMM2	Congenital disorder of glycosylation, type Ia	AR	NM_000303.2	133.97
PODXL	-	-	NM_001018111.2	61.63
PTPRO	Nephrotic syndrome, type 6	AR	NM_030668.2	137.09
SCARB2	Epilepsy, progressive myoclonic 4, with or without renal failure	AR	NM_001204255.1	123.97
SGPL1	Nephrotic syndrome, type 14	AR	NM_003901.3	66.27
SMARCAL1	Schimkeimmunoosseous dysplasia	AR	NM_001127207.1	122.11
SPRY2	IgA nephropathy, susceptibility to, 3	AD	NM_001318536.1	99.38
SYNPO	-	-	NM_001166208.1	87.84
TP53RK	Galloway-Mowat syndrome 4	AR	NM_033550.3	62.52
TPRKB	Galloway-Mowat syndrome 5	AR	NM_001330386.1	64.71
TRPC6	Glomerulosclerosis, focal segmental, 2	AD	NM_004621.5	96.58
TTC21B	Nephronophthisis 12	AR,AD	NM_024753.4	142.42
TUBAL3	-	-	NM_001171864.1	61.15
VIPAS39	Arthrogryposis, renal dysfunction, and cholestasis 2	AR	NM_001193314.1	125.19

VPS33B	Arthrogryposis, renal dysfunction, and cholestasis 1	AR	NM_001289148.1	137.52
WDR73	Galloway-Mowat syndrome 1	AR	NM_032856.3	73.33
WT1	Denys-Drash syndrome, Frasier syndrome, nephrotic syndrome, type 4	AD,somaticmutation	NM_000378.4	103.30
XPO5	-	-	NM_020750.2	57.90
ZMPSTE24	Mandibuloacral dysplasia with type B lipodystrophy; restrictive dermopathy; lethal	AR	NM_005857.4	118.18

Supplementary Table SII Clinical and demographic characteristics of included patients

ID	*Sex*	*Religion*	*State of origin*	*Age at onset, days*	*Consanguinity*	*Family history*	*Extra-renal features*	*Low birth weight*	*Prematurity*	*Seizures*	*Weight SDS*	*Length SDS*	*eGFR, ml/min per 1.73 m²*	*Serum albumin, g/dl*	*Total cholesterol, mg/dl*
4^1	Boy	Hindu	Haryana	5	0	0	Developmental delay	0	1	0	-1.05	-3.68	86.67	2.2	492
8	Boy	Hindu	Uttar Pradesh	30	0	0	0	1	1	1	-1.56	-4.46	60.00	1.5	171
18	Boy	Muslim	Uttar Pradesh	15	1	0	Clubbing	1	1	1	0.2	NA	60.00	0.6	201
28	Boy	Hindu	Punjab	20	0	0	Atrial septal defect	0	0	0	NA	NA	65.00	3.1	NA
30	Girl	Hindu	Delhi	NA	0	0	0	0	0	0	NA	NA	NA	NA	NA
40^1	Boy	Hindu	Delhi	5	0	1	0	1	0	1	NA	NA	22.60	1	NA
46	Boy	Hindu	NK		0	0	0	0	0	0	NA	NA	NA	NA	NA
47^1	Boy	Hindu	Rajasthan	30	0	0	0	0	0	0	-2.48	-4.63	24.00	2.3	283
51	Boy	Hindu	Delhi	90	0	1	Oculocutaneous albinism, developmental delay, microcephaly, hepatomegaly	0	0	1	NA	NA	4.24	2.1	NA
52	Girl	Hindu	Delhi	NA	0	0	0	0	0	0	NA	NA	NA	NA	NA
59^3	Girl	Hindu	Nepal	45	0	1	Atrial septal defect	1	1	0	-1.88	-4.06	21.85	1.1	391
80	Boy	Hindu	Telangana	30	0	0	0	1	1	0	-3.94	NA	NA	1.2	188
85	Girl	Hindu	Madhya Pradesh	15	0	1	0	0	0	0	-3.29	-11.73	49.00	1.3	349
146^1	Girl	Hindu	Delhi	4	0	0	0	1	1	1	-4.02	-3.54	46.40	1.2	233
150^2	Boy	Hindu	Punjab	60	0	0	0	1	1	1	-3.14	-2.16	36.00	1.1	247
154	Girl	Muslim	Uttar Pradesh	4	1	1	Hiatus hernia, microcephaly, developmental delay, hypotonia	1	0	0	-2.2	-1.3	64.00	0.9	406
157	Boy	Hindu	Uttar Pradesh	15	0	1	Aqueductal stenosis,	0	0	1	-4.61	-5.12	58.00	1.3	295

							obstructive hydrocephalus								
162^[1]	Boy	Muslim	Delhi	30	1	0	0	1	1	0	0.6	NA	96.00	1.3	428
163	Girl	3	Punjab	30	0	0	0	0	0	0	-4.55	-3.92	77.33	1.4	234
165^[3]	Girl	Hindu	Bihar	45	0	0	0	0	0	0	-4.22	-2.85	32.00	0.9	276
168	Girl	Muslim	Uttar Pradesh	20	1	1	Café au lait spot; hirsutism	1	0	0	-2.73	-1.71	104.00	1.7	243
169^[2]	Girl	Muslim	Uttar Pradesh	26	1	0	0	1	0	0	-3.61	-4.07	94.00	0.6	402
173^[1,2]	Girl	Hindu	Uttar Pradesh	60	0	0	0	1	1	1	-4.14	-5.17	17.00	0.7	172
180	Girl	Hindu	Uttar Pradesh	85	0	0	0	0	1	0	-4.32	-6.39	200.00	0.8	250
181	Boy	Muslim	Uttar Pradesh	12	0	0	Dysmorphic facies	0	1	0	0.8	NA	36.00	1.2	342
196^[1]	Boy	Muslim	Uttar Pradesh	15	1	0	0	1	0	1	-4.14	-4.52	28.29	0.9	474
201^[1]	Girl	Muslim	Delhi	20	1	1	0	0	0	0	-2.23	-3.37	196.00	0.56	237
217	Boy	Muslim	Rajasthan	2	1	1	0	NA	NA		-2.69	-0.23	23.66	1.44	309
228^[1]	Boy	Hindu	Uttar Pradesh	15	0	0	0	NA	NA	1	-4.91	-0.5	201.00	1.1	239
235^[1]	Girl	Muslim	Uttar Pradesh	45	0	0	0	1	0	0	-5.83	-5.17	123.90	1.4	200
240	Boy	Hindu	Puducherry	70	1	1	Dysplastic ears	NA	NA	0	-0.12	-0.59	134.23	0.9	271
266	Boy	Muslim	Delhi	15	1	0	0	NA	NA	NA	NA	NA	NA	1.23	296
267^[3]	Girl	Hindu	Bihar	15	0	1	Atrial septal defect	0	0	0	NA	NA	NA	1.2	NA
X1	Girl	Muslim	Punjab	15	1	0	Microcoria,microcornea	NA	NA	NA	NA	NA	NA	NA	NA

eGFR estimated glomerular filtration rate; NA not available

^Indicates patients that shared the following variations: [1]c.2600G>A; [2]c.3478C>T and [3]c.C1099C>T

Supplementary Table SIII Quality metrics based on raw (FASTQ) and mapped (BAM) reads

Sample ID	*Total mapped reads*	*Percentage of total mapped reads over reference genome*	*Mean region coverage depth*
4	36903956	73.80%	48.2
8	36883757	74.50%	48.9
18	39101579	76.00%	52.8
28	39925438	97.86%	57.45
30	33396553	97.6%	43.96
40	36402136	97.77%	49.82
46	46559034	97.63%	57.5
47	48347870	97.67%	51.46
51	41882937	98.14%	58.09
52	54820473	97.81%	59.33
59	43196032	97.71%	58.23
80	46494263	97.72%	57
85	41809738	97.7%	42.99
146	35916462	97.55%	42.14
150	48251461	97.55%	49.67
154	37484353	98.19%	50.85
157	47974741	97.63%	51.34
162	50014242	97.72%	54.71
163	33651571	97.79%	44.01
165	48225036	97.75%	61.04
168	47661449	98.01%	51.14
169	35009940	97.71%	41.01
173	33637511	97.71%	34.08
180	28249162	97.89%	37.79
181	32296239	97.56%	34.09
196	30935558	97.36%	35.64
201	33626554	98.26%	59.42
217	74654485	98.17%	115.89
228	55633896	98.08%	90.73
235	66570469	98.09%	97.98
240	35311365	97.81%	53.55
266	40554026	97.94%	66.39
267	48404929	98.04%	82.57
X1	53300045	99.17%	90.9

Supplementary Table SIV Lists of prioritized variants for individual patients. Variants in bold were considered relevant

Patient ID	*Gene*	*Chromosome: Position*	*Exon*	*Consequence (Base-pair and amino acid)*	*Zygosity*	*Change*	*Frequency in 1KG; Exac; Exac_SAS; gnomAD*	*Polyphen*	*CADD-Phred*	*Eigen_raw*	*GERP++_RS*
4	***NPHS1***	**19:36317523**	**29**	**c.3619delG:p.E1207Kfs*29**	**Heterozygous**	**fs*del**	**.; .; .; $4.0x10^{-6}$**	**.**	**.**	**.**	**.**
	NPHS1	**19:36333089**	**19**	**c.G2600A:p.G867D**	**Heterozygous**	**NS**	**.; 0.00002173; 0.0001; 0.00001634**	**D**	**28.7**	**0.74**	**3.88**
	ITGB4	17:73753376	39	c.G5314A:p.E1772K	Heterozygous	NS	0.0002; 0.000059; 0; 0.000072	B	23	-0.305	3.03
8	***NPHS1***	**19: 36335078_36335079delGT**	**16**	**c.2138_2139delAC: p.D713Gfs*12**	**Heterozygous**	**fs*del**	**.; .; .; .**	**.**	**.**	**.**	**.**
	NPHS1	**19:36341342**	**5**	**c.C532T: p.Q178X**	**Heterozygous**	**Tr***	**.; .; .; 0.00001193**	**.**	**35**	**0.545**	**3.26**
	COQ8B	19:41198902	14	c.T1250C: p.L417P	Heterozygous	NS	.; .; .; .	D	25.6	0.831	5.37
18	***NPHS1***	**19:36322049**	**27**	**c.3388-1G>A**	**Homozygous**	**Sp**	**.; 0.0000083; 0; $3.98x10^{-6}$**	**.**	**22.2**	**0.771**	**4.1**
	NPHS1	19:36333098	19	c.G2591A: p.R864H	Homozygous	NS	0.0002; 0.0001; $7.2x10^{-5}$; 0.0001	D	34	0.501	4.93
	FN1	2:216242961	35	c.G5647C: p.V1883L	Heterozygous	NS	.; .; .; .	B	22.9	-0.123	4.6
28	***NPHS2***	**1:179530456**	**3**	**c.G419A: p.G140E**	**Homozygous**	**NS**	**.; $8.24 x10^{-6}$; $6.1 x10^{-5}$; .**	**D**	**32**	**0.983**	**5.82**
	MUC1	1:155162020	2	c.C113G: p.S38W	Heterozygous	NS	0.0004; 0.0002; 0.0013; 0.0002	D	22	-0.814	-3.48
30	No variants prioritized										
40	***NPHS1***	**19:36333089**	**19**	**c.G2600A: p.G867D**	**Homozygous**	**NS**	**.; 0.000022; 0.0001; 0.000016**	**D**	**28.7**	**0.74**	**3.88**
	ALMS1	2:73799812	16	c.A10805G: p.N3602S	Heterozygous	NS	.; 0.0001; 0.001; 0.0001	D	24.5	0.46	4.44
	ARHGDIA	17:79826497	7	c.G758A: p.R253H	Heterozygous	NS	.; .; .; $7.50 x10^{-6}$	.	.	.	.
46	***PLCE1***	**10:96058156**	**24**	**c.C5188T: p.Q1730X**	**Homozygous**	**Tr***	**.; .; .; .**	**.**	**41**	**0.964**	**5.6**
	ALMS1	2:73653592	6	c.G1249T: p.G417W	Heterozygous	NS	.; .; .; .	D	26	0.325	3.8

	INF2	14:105181131	21	c.G3632T: p.R1211L	Heterozygous	NS	.; 0.0002; 0.0011; 0.000098	P	22.8	-0.822	-2.53
47	***NPHS1***	**19:36333089**	**19**	**c.G2600A: p.G867D**	**Homozygous**	**NS**	**.; 0.000022; 0.0001; 0.00001634**	**D**	**28.7**	**0.74**	**3.88**
	COL4A3	2:228173699	49	c.G4547A: p.R1516Q	Heterozygous	NS	.; 0.000075; 0.0003; 0.000072	D	28.4	1.083	5.97
	CRB2	9:126128285	3	c.508_509del: p.C170fs	Heterozygous	fs*del	.; .; .; .	.	.	.	.
51	*COL4A4*	2:227924195	28	c.C2309T: p.P770L	Heterozygous	NS	.; 0.0000663; 0; 0.00003606	D	25.1	0.717	5.99
	NUP93	16:56864492	10	c.G980A: p.R327H	Heterozygous	NS	.; 0.0000165; 0.0001; 0.00002388	D	34	0.949	5.11
52	*ARHGAP24*	4:86921681	10	c.G2053A: p.D685N	Heterozygous	NS	0.0008; 0.0009; 0.0064; 0.0009	P	29.3	0.295	5.56
	SCARB2	4:77116941	2	c.A194G: p.Y65C	Heterozygous	NS	0.0006; 0.0003; 0.0021; 0.0003	D	25.3	0.53	4.35
59	***NPHS1***	**19:36339610**	**9**	**c.C1099T: p.R367C**	**Homozygous**	**NS**	**.; 0.00003308; 0.0001; 0.00003978**	**D**	**28.7**	**0.528**	**4.37**
	ARHGAP24	4:86921681	10	c.G2053A: p.D685N	Heterozygous	NS	0.0008; 0.0009; 0.0064; 0.0009	P	29.3	0.295	5.56
80	***NPHS1***	**19:36336350**	**14**	**c.A1850G: p.H617R**	**Heterozygous**	**NS**	**.; 0.0000084; 0; $7.98x10^{-6}$**	**D**	**22.8**	**0.43**	**4.56**
	NPHS1	**19:36340176**	**7**	**c.C802T: p.R268X**	**Heterozygous**	**Tr***	**.; 0.000034; $6.1x10^{-5}$; 0.00002407**	**.**	**35**	**0.166**	**2.65**
	COQ2	4:84188827	6	c.G1013A: p.G338E	Heterozygous	NS	.; .; .; 0.00001133	P	23.6	0.125	4.77
	CUBN	10:16918949	57	c.A9053C: p.Y3018S	Heterozygous	NS	.; 0.0001; 6.1 $x10^{-5}$; 0.0001	D	23.2	0.24	3.39
	TTC21B	2:166805994	3	c.C172T: p.R58X	Heterozygous	Tr*	.; 0.000058; 0.0002; 0.000040	.	36	0.751	4.52
85	***NPHS1***	**19:36339251**	**10**	**c.C1219T: p.R407W**	**Homozygous**	**NS**	**.; $8.2x10^{-6}$; 0; 0.000020**	**D**	**27**	**0.207**	**3.29**
	ANKS6	9:101558508	1	c.G266A: p.G89D	Homozygous	NS	.; .; .; 0	B	27.5	0.069	3.36
	KANK2	19:11304445	4	c.G311C: p.G104A	Heterozygous	NS	0.0004; 0.0005; 0.0037; 0.0004	D	13.59	-0.083	4.38
146	***NPHS1***	**19:36333089**	**19**	**c.G2600A: p.G867D**	**Homozygous**	**NS**	**.; 0.000022; 0.0001; 0.000016**	**D**	**28.7**	**0.74**	**3.88**
	KANK1	9:710861	7	c.T95C: p.F32S	Heterozygous	NS	0.0006; 0.0004; 0.0023; 0.0003	P	12.73	0.092	3.58

150	***NPHS1***	**19:36321958**	**27**	**c.C3478T: p.R1160X**	**Homozygous**	**Tr***	**.; 0.000066; 0.0002; 0.00009943**	**.**	**37**	**0.251**	**3.74**
154	***OSGEP***	**14:20920566**	**2**	**c.A157T: p.I53F**	**Homozygous**	**NS**	**.; $2.47x10^{-5}$; 0.000182; $1.22x10^{-5}$**	**B**	**15.82**	**-0.4**	**0.619**
	ANLN	7:36459872	11	c.G1964A: p.R655Q	Heterozygous	NS	.; 0.000016; 0.0001; $7.96 x10^{-6}$	P	29.3	0.455	4.95
	MAGI2	7:77649189	22	c.G3811A: p.A1271T	Heterozygous	NS	0.0004; 0.0002; 0.0012; 0.0002	P	28.3	-0.075	4.59
157	***NPHS1***	**19:36333370**	**18**	**c.C2417A: p.A806D**	**Homozygous**	**NS**	**.; $8.24 x10^{-6}$; 0; $7.95 x10^{-6}$**	**D**	**23.7**	**0.374**	**4.46**
	INF2	14:105181022	21	c.G3523A: p.D1175N	Heterozygous	NS	.; 0.000017; 0; $8.15 x10^{-5}$	D	19.12	0.097	4.73
	NPHP4	1:5937173	20	c.C2797T:p.R933W	Heterozygous	NS	.; 0.0000416; 0; 0.00002938	P	16.48	-1.152	-9.61
162	***NPHS1***	**19:36333089**	**19**	**c.G2600A:p.G867D**	**Homozygous**	**NS**	**.; 0.000022; 0.0001; 0.000016**	**D**	**28.7**	**0.74**	**3.88**
	ALMS1	2:73747129	11	c.C9764G:p.S3255C	Heterozygous	NS	0.001; 0.0006; 0.0044; 0.0005	D	23.4	-0.174	3.76
	COL4A1	13:110817289	46	c.G4070C:p.G1357A	Heterozygous	NS	.; .; .; .	D	26	0.667	4.3
	ITGB4	17:73739874	26	c.C3043T:p.R1015C	Heterozygous	NS	.; 0.0001; 0.0007; 0.0001	D	28.9	0.186	2.94
	KANK1	9:742265	14	c.C3757T:p.L1253F	Heterozygous	NS	.; .; .; $3.98 x10^{-6}$	D	26.3	0.472	4.28
163	***NPHS1***	**19:36335272**	**15**	**c.C2020T:p.P674S**	**Homozygous**	**NS**	**.; .; .; .**	**D**	**27**	**0.728**	**3.67**
	KANK1	9:710853	7	c.87delC: p.D29fs	Heterozygous	fs*del	.; $8.4x10^{-6}$; $6.8 x10^{-5}$; $3.99 x10^{-6}$	.	.	.	.
	VPS33B	15:91561079	2	c.C133G: p.L45V	Heterozygous	NS	0.0002; 0.000058; 0.0004; 0.000043	D	25.7	0.668	5.45
165	***NPHS1***	**19:36339610**	**9**	**c.C1099T: p.R367C**	**Heterozygous**	**NS**	**.; 0.00003308; 0.0001; 0.00003978**	**D**	**28.7**	**0.528**	**4.37**
	NPHS1	**19:36341349**	**5**	**c.527-2A>G**	**Heterozygous**	**Sp**	**.; .; .; .**	**.**	**22.6**	**0.788**	**4.09**
	ALMS1	2:73680160	8	c.C6503T: p.S2168L	Heterozygous	NS	0.0002; 0.000075; 0.0005; 0.000060	D	25.2	0.094	3.48
168	***NPHS1***	**19:36330221**	**22**	**c.C3027G: p.Y1009X**	**Homozygous**	**Tr***	**.; $8.24 x10^{-6}$; $6.1 x10^{-5}$; $7.95 x10^{-6}$**	**.**	**38**	**0.513**	**1.06**
	ALMS1	2:737997787	17	c.A10771C: p.T3591P	Heterozygous	NS	0.0008; 0.0003; 0.0025; 0.0003	D	18.17	-0.413	-1.18
	ARHGAP24	4:86921681	10	c.G2053A: p.D685N	Heterozygous	NS	0.0008; 0.0009; 0.0064; 0.0009	P	29.3	0.295	5.56

	CRB2	chr9:126135651 Exon-10	10	c.2841delG: p.P947fs	Heterozygous	fs*del	.; .; .; .	.	.	.	.
169	***NPHS1***	**19:36321958**	**27**	**c.C3478T: p.R1160X**	**Homozygous**	**Tr***	**.; 0.000066; 0.0002; 0.000099**	**.**	**37**	**0.251**	**3.74**
	CD151	11:837277	6	c.A379C: p.K127Q	Heterozygous	NS	0.0002; 0.0004; 0.0031; 0.0004	B	15.64	-0.087	4.26
	TTC21B	2:166747104	24	c.C3148T: p.R1050W	Heterozygous	NS	.; .; .; 3**.99** x10^{-6}	D	34	0.891	4.76
173	***NPHS1***	**19:36321958**	**27**	**c.C3478T: p.R1160X**	**Heterozygous**	**Tr***	**.; 0.000066; 0.0002; 0.000099**	**.**	**37**	**0.251**	**3.74**
	NPHS1	**19:36333089**	**19**	**c.G2600A: p.G867D**	Heterozygous	NS	.; 0.000022; 0.0001; 0.000016	D	28.7	0.74	3.88
	ITGA3	17:48153013	12	c.C1588T: p.R530C	Heterozygous	NS	.; 0.00002481; 6.1x10^{-5}; 0.00002389	D	27.8	0.244	5.54
	SYNPO	5:150036540	3	c.G2603A: p.G868E	Heterozygous	NS	0.0002; 0.0004; 0.0019; 0.0001	D	17.24	0.444	4.91
180	***NPHS1***	**19:36337056**	**12**	**c.C1481A: p.S494X**	**Heterozygous**	**Tr***	**.; .; .; .**	**.**	**39**	**0.581**	**4.15**
	NPHS1	**19:36340541-36340548**	**6**	**c.614_621delinsTT: p.T205_A207delinsI**	**Heterozygous**	**fs*del**	.; **0.00001653**; **0.0001; 0.0000199**	**.**	**.**	**.**	**.**
	NPHP4	1:5937221	20	c.2749delG:p.E917fs	Heterozygous	fs*del	.; .; .; .	.	.	.	.
181	***NPHS1***	**19:36332715**	**20**	**c.T2717C:p.I906T**	**Homozygous**	**NS**	**.; .; .; .**	**D**	**26.8**	**0.821**	**4.78**
	FN1	2:216271099	19	c.C2848T:p.H950Y	Heterozygous	NS	.; 8.24x10^{-6}; 6.1x10^{-5}; 0.00003183	P	24.6	0.271	5.14
196	***NPHS1***	**19:36333089**	**19**	**c.G2600A:p.G867D**	**Homozygous**	**NS**	**.; 0.00002173; 0.0001; 0.00001634**	**D**	**28.7**	**0.74**	**3.88**
	KANK2	19:11304341	4	c.G415T:p.A139S	Heterozygous	NS	.; .; .; .	D	22.6	0.161	4.38
	NPHP4	1:5947516	18	c.G2315A:p.R772H	Heterozygous	NS	.; 0.00001685; 0; 8.06x10^{-6}	D	33	0.754	5.46
201	***NPHS1***	**19:36333089**	19	**c.G2600A:p.G867D**	**Homozygous**	NS	**.; 0.00002173; 0.0001; 0.00001634**	**D**	**28.7**	**0.74**	**3.88**
	EXT1	8:119122904	1	c.G382T:p.A128S	Heterozygous	NS	.; 0.00001648; 0.0001; 0.0000159	B	12.59	-0.582	5.47
	LRP2	2:170068592	37	c.C6166T:p.R2056W	Heterozygous	NS	0.0004; 0.0001; 0.001; 0.0001	D	34	0.815	5.88
217	*ARHGDIA*	17:79826519	7	c.T736G:p.C246G	Heterozygous	NS	.; .; .; .	.	10.02	1.931	0.929
	FAT1	4:187527277	17	c.G10297A:p.V3433I	Heterozygous	NS	0.0006; 0.0002; 0.0014; 0.0002	B	19.37	-0.266	5.56
	ITGB4	17:73727328	10	c.G1094A:p.R365Q	Heterozygous	NS	.; 0.0001; 0.0006; 0.0001	P	24.3	-0.129	4.12

228	***NPHS1***	**19:36333089**	19	**c.G2600A:p.G867D**	**Heterozygous**	**NS**	**.; 0.00002173; 0.00001634**	**0.0001;**	**D**	**28.7**	**0.74**	**3.88**
	NPHS1	**19:36340541-36340548**	6	**c.614_621delinsTT; p.T205_A207delinsI**	**Heterozygous**	**fs*del**	**.; 0.00001653; 0.0000199**	**0.0001;**	**.**	**.**	**.**	**.**
	COQ6	14:74427966	9	c.G907A:p.A303T	Heterozygous	NS	0.0002; 0.0003; 0.0003	0.0023;	B	24.1	0.071	5.33
	COQ9	16:57490845	5	c.A524G:p.K175R	Heterozygous	NS	0.0002; 0.0001; 0.0001	0.0009;	P	23.6	0.464	5.68
	ITGB4	17:73745039	27	c.C3229T:p.R1077C	Heterozygous	NS	.; 0.00002598; 0.00003209	0;	D	32	0.584	4.93
235	***NPHS1***	**19:36333089**	19	**c.G2600A:p.G867D**	**Homozygous**	**NS**	**.; 0.00002173; 0.00001634**	**0.0001;**	**D**	**28.7**	**0.74**	**3.88**
	LAMB2	3:49162269	21	c.A2974G:p.I992V	Heterozygous	NS	0.0004; 0.0005; 0.0005	0.0038;	D	23.9	0.562	4.36
240	*WT1*	11:32450063	2	c.T749A:p.M250K	Heterozygous	NS	0.0002; 0.0003; 0.0002	0.0019;	B	24.4	0.09	5.62
266	No prioritized variations											
267	***NPHS1***	**19:36339610**	9	**c.C1099T: p.R367C**	**Heterozygous**	**NS**	**.; 0.000033; 0.0000398**	**0.0001;**	**D**	**28.7**	**0.528**	**4.37**
	NPHS1	**19:36341889**	4	**c.C500T: p.P167L**	**Heterozygous**	**NS**	**.; $8.3x10^{-6}$; $6.1x10^{-5}$; $3.9x10^{-6}$**		**D**	**27.6**	**0.555**	**5.99**
	FAT1	4:187540958	10	c.C6782T: p.T2261M	Heterozygous	NS	0.001; 0.0009; 0.0008	0.0033;	D	24.6	0.624	5.05
X1	***LAMB2***	3:49168499		**c.C799T; p.R267Ter**	**Homozygous**	**Tr**	**-, 0, 0, -**		**D**	**36**	**0.85**	**4.76**

B benign; D deleterious; del deletion; fs frameshift; NS non synonymous; P pathogenic; Tr truncating

Supplementary Table SV List of 44 single nucleotide polymorphisms (SNP), flanking p.Gly867Asp, selected for haplotype analysis

Haplotype region	*Chromosomal Coordinates*	*SNP ID*	*Reference allele A*	*Alternate; allele B*	*P-value**
5'-H2	chr19:35850672	rs142125121	T	C	0.0268
	chr19:35863180	rs201159994	A	G	0.0005
	chr19:35863226	rs150552589	G	T	2.2498E-08
5'-H1	chr19:35898796	rs112270905	T	TC	7.0269E-11
	chr19:35898899	rs16970294	A	G	7.0269E-11
	chr19:35899037	rs113510419	T	C	7.0269E-11
	chr19:35899068	rs142160831	GTGA	G	7.0269E-11
	chr19:35991373	rs2293695	C	T	0.025
	chr19:35998362	rs4254439	T	G	0.0459
	chr19:36004106	rs4806163	A	G	1.5754E-06
	chr19:36004171	rs12460932	C	A	0.0459
	chr19:36017928	rs10775583	G	C	0.0005
	chr19:36018272	rs12461911	C	T	0.0001
	chr19:36033460	rs2239945	C	T	0.0001
5'-H	chr19:36048741	rs2230181	G	T	0.0152
	chr19:36157740	rs61741212	C	T	0.0332
	chr19:36168914	rs2285421	T	C	2.0518E-05
	chr19:36218478	rs11670414	C	T	0.0047
	chr19:36224705	rs231591	A	G	5.3275E-08
	chr19:36233470	rs3746278	G	A	0.0113
	chr19:36234489	rs28656784	T	C	0.0332
	chr19:36235431	rs3761087	A	G	0.0256
	chr19:36236909	rs10402601	G	C	0.0256
	chr19:36246418	rs11549030	C	G	0.0429
	chr19:36269915	rs231230	T	C	0.0081
	chr19:36270052	rs231231	A	G	0.0081
	chr19:36273534	rs2291067	G	A	0.0035
	chr19:36275074	rs62112163	G	A	0.0035
	chr19:36278470	rs231235	C	G	0.0081

	chr19:36321910	rs731934	G	A	0.001
	chr19:36322270	rs2071327	C	T	0.0007
	chr19:36322509	rs466452	G	A	1.0815E-05
Mutation	chr19:36333089	G867D	C	T	3.53E-16
3'-H1	chr19:36351935	rs35854130	G	T	0.00006686
	chr19:36549684	rs61742664	G	A	0.015
3'-H2	chr19:36574063	rs45567532	C	T	7.6486E-08
	chr19:36577579	rs4806263	C	T	7.6486E-08
	chr19:36577742	rs77938609	G	A	0.015
	chr19:36583651	rs61494900	G	A	7.6486E-08
	chr19:36590329	rs2285745	T	C	0.0015
	chr19:36594063	rs17851502	C	T	7.6486E-08
	chr19:36595436	rs1008328	A	C	0.0044
	chr19:36603703	rs2072605	T	A	0.0005
	chr19:36674305	rs4805162	A	G	0.0066
	chr19:36727365	rs2070132	G	A	0.0013

Supplementary Table SVI Haplotype analysis of single nucleotide polymorphism markers flanking the mutation, indicating segregation of a core haplotype along with the Gly867Asp variant

No.	*~12 kbp region*	*~135 kbp region*	*Core-Haplotype (~500 kbp region)*			*~153 kbp region*	*Allele Count*
	rs142125121-rs150552589	*rs112270905-rs2239945*	*rs2230181-rs466452*		*rs35854130-rs61742664*	*rs45567532-rs2070132*	
	5'-H2	*5'-H1*	*5'-H*	***G867D***	*3'-H1*	*3'-H2*	
1	ABB	BBBBAAAABBB	AAAAAAAAAAAAAAAAAB	**B**	BA	BBABBBBBAB	13
2	ABB	BBBBAAAABBB	AAAAAAAAAAAAAAAAAB	**B**	BA	AAAABABBAB	1
3	AAA	BBBBAAAABBB	AAAAAAAAAAAAAAAAAB	**B**	BA	BBABBBBBAB	1
4	AAA	AAAABABABAA	AAAAAAAAAAAAAAAAAB	**B**	BA	AAAAAABABA	2
5	BAA	AAAAABAABBB	AAAAAAAAAAAAAAAAAB	A	BA	BBABBBBBAB	1
6	AAA	AAAABABABBB	AAAAAAAAAAAAAAAAAB	A	AA	AAAAAABAAA	1
7	BAA	AAAAAAAABBB	AAAAAAAAAAAAAAAAAB	A	AB	AABAAABAAB	1
	Other haplotypes						46

**Allele A refers to the major allele and B refers to the minor allele. In cases of Gly867Asp, B is a mutant allele. The grey shaded area refers to the mutant allele associated core haplotype*

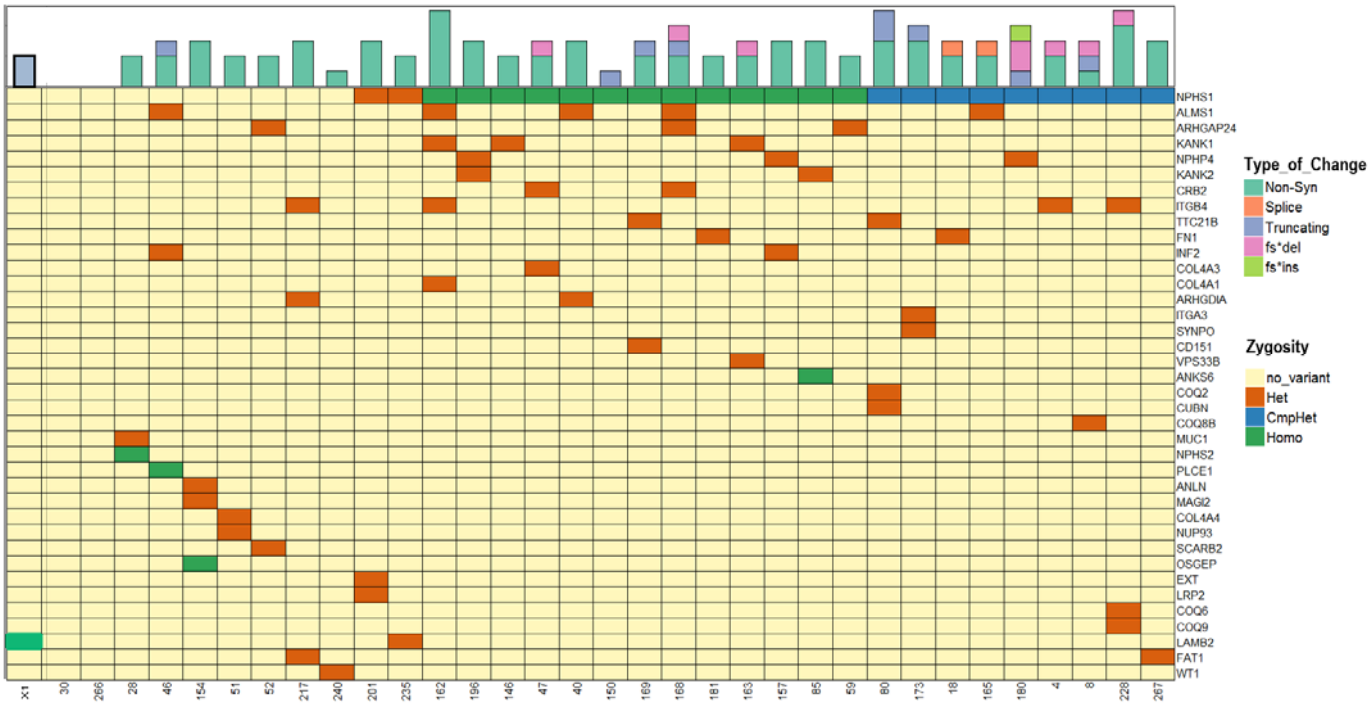
Supplementary SVII Studies examining the genetic basis of congenital nephrotic syndrome in 10 or more patients

Author, year	*N*	*Method of sequencing (number of genes)*	*Ethnicity*	*Etiology, %*						
				NPHS1	*NPHS2*	*WT1*	*PLCE1*	*LAMB2*	*Others*	*Unknown*
Koziell, 2002 [xi]	41	Sanger (2)	British, Maltese, Turkish, Asian	73	10	NT	NT	NT	NT	15
Sako, 2005 [xii]	13	Sanger (4)	Japanese	15	8	0	NT	NT	0 for *ACTN4*	77
Machuca, 2010 [xiii]	117	Sanger (8)	West Europe; Turkey; North Africa	61	15	2	2	2	0	19
Schoeb, 2010 [xiv]	67	Sanger (1)	Worldwide	58	Exc	Exc	NT	NT	NT	42
Buscher, 2010 [xv]	62	Sanger, panel (10)	German	53	13	23	2	5	2 (*ARGHDIA*)	3
Santin, 2011 [xvi]	15	Sanger (8)	Spanish	80	7	13	0	NT	NT	0
Lee, 2011 [xvii]	15	Not stated	Korean	40	7	40	0	7	0	7
Mbarek, 2011 [xviii]	12*	Linkage, Sanger (6)	Tunisian	60	0	0	0	40	0 for *CD2AP*	0
Cil, 2015 [xix]	80	Sanger (4)	Turkish; Middle East; East Europe	46	16	6	NT	4	NT	28
Sadowski, 2015 [xx]	235	Next-generation	Worldwide	40	11	9	2	6	3	31
Trautman, 2015 [xxi]	98	Sanger or next-generation	Europe, Middle East, Latin America	NA	NA	NA	NA	NA	NA	34
Sen, 2017 [xxii]	31	Next-generation	Worldwide	39	6	3	0	10	0	42
Wang, 2017 [xxiii]	12	Next-generation	Chinese	50	0	8	0	8	8 (*ADCK4*)	25
Li, 2018 [xxiv]	12	Sanger or next-generation	Chinese	67	0	8	0	0	8 (*COQ6*)	17
Sharief, 2019 [xxv]	11	Not stated	Arab, Asian, African	64	0	9	0	27	0	0
Nishi, 2019 [xxvi]	36	Not stated	Japanese	42	3	22	0	8	3 (*CRB2*)	22
Dufek, 2019 [xxvii]	69	Not stated	European	80	1	13	1	3	1 (*SGPL1*)	14
Berody, 2019 [xxviii]	55	Sanger (5)	European	65	9	7	2	0	0	16
Sinha, 2019 [xxix]	15	Next generation (27) or Sanger (<4)	Indian	53	0	7	7	0	0	33
Nagano, 2020 [xxx]	13	Targeted next generation	Japanese	31		15		31	8 (*LAMA5*)	15
Present study	34	Next-generation	Indian	74	4	0	4	4	4 (*OSGEP*)	11

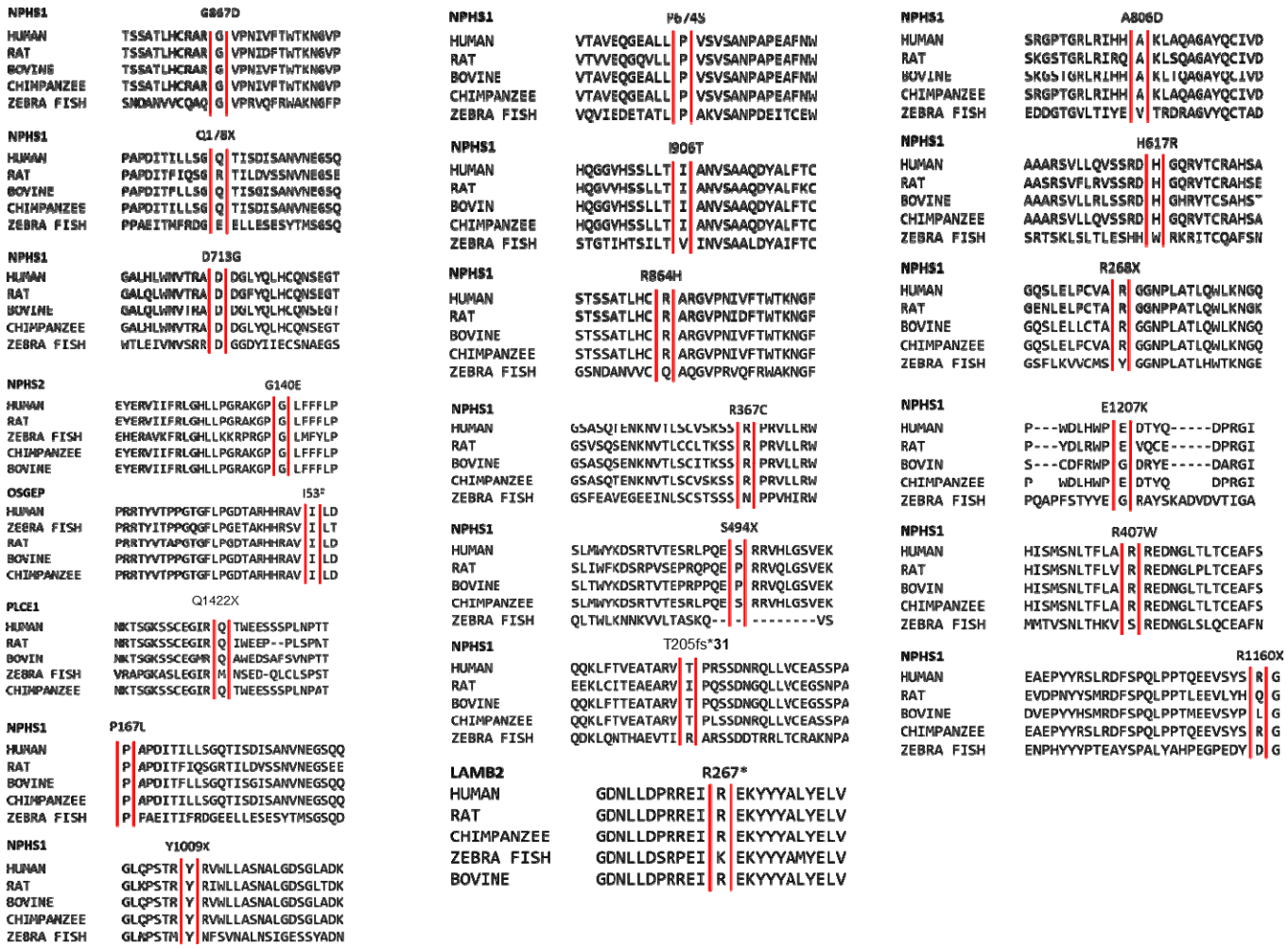
Exc excluded; NA not available; NT not tested
Only latest and largest paper for each group was included, unless overlap of patients between studies appeared unlikely
**Refers to 12 patients from 5 families*

Supplementary Table SVIII Frequency of haplotype markers of 5'-H1/5'H/3'-H1/3'-H2 region including Gly867Asp variant, (as indicated in Supplementary Table S6) from the1000 Genome population dataset

Sub-populations	*South Asian*	*European*	*East Asian*	*American*	*African*
Number of disease core haplotype carriers	2	20	1	7	1
Number of subjects	489	503	504	347	661
Frequency, %	0.20	1.98	0.09	1.008	0.07



Supplementary Figure S1 Heatmap representing prioritized variants per sample. Each column represents a patient while rows indicate genes. Individual cells are colored according to zygosity of variant while the type of change is indicated at the top of each column.



Supplementary Figure S2 Images indicating degree of conservation across species for variants to which pathogenicity was attributed

SUPPLEMENTARY REFERENCES

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