

## Supplementary Table 1.

### List of the genes present in the smallest region of overlap in patients with constitutional distal chromosome 6p duplication presenting with glomerulopathy.

The entries are based on information collected at OMIM (<http://www.ncbi.nlm.nih.gov/omim/>), NCBI GENE (<http://www.ncbi.nlm.nih.gov/gene/>) and Nexus Copy Number Software ver 7.5 (Biodiscovery, Hawthorne, CA) databases.

Chromosomal coordinates are given according to the UCSC genome annotation database for the Mar. 2006 GenBank freeze assembled by NCBI (hg18, Build 36.1).

Gene	Name	OMIM ID	Chromosomal coordinates	Function of the encoded protein.
<b>Protein-coding genes with RefSeq.</b>				
<b>FOXQ1</b>	forkhead box Q1	*612788	1257674-1259993	Transcription factor involved in embryonic development, cell cycle regulation, tissue-specific gene expression, cell signaling, and tumorigenesis
<b>FOXF2</b>	forkhead box F2	*603250	1335067-1340831	Transcription factor involved in embryonic development (limbs, gastrointestinal track, genitalia, epithelial to mesenchymal transition, extracellular matrix organization)
<b>FOXC1</b>	forkhead box C1	*601090 #602482 #601631	1555679-1559128	Transcription factor involved in embryonic development (ocular, cardiovascular and neuronal in particular) tumorigenesis and epithelial-mesenchymal transition. <b>Genotype -phenotype:</b> Axenfeld-Rieger syndrome, type 3; Iridogonidys-genesis, type 1
<b>GMDS</b>	GDP-mannose 4,6-dehydratase	*602884	1569033-2190867	EC 4.2.1.47. Catalyzes the conversion of GDP-mannose to GDP-4-keto-6-deoxymannose, the first step in the synthesis of GDP-fucose from GDP-mannose, using NADP+ as a cofactor.
<b>MYLK4</b>	myosin light chain kinase family, member 4	n/a	2608861-2696153	Protein phosphorylation (Serine/Threonine protein kinase)
<b>WRNIP1</b>	Werner helicase interacting protein 1	*608196	2710664-2730978	Replication factor that interacts with WRN, the protein that is defective in Werner syndrome. Accumulates at sites of DNA damage by interacting with polyubiquitinated proteins and also binds to DNA polymerase delta.
<b>SERPINB1</b>	serpin peptidase inhibitor, clade B (ovalbumin), member 1	*130135	2777564-2787282	Monocyte/neutrophil elastase inhibitor, a member of the serpin (serine proteinase inhibitor) family of proteinase inhibitors
<b>SERPINB6</b>	serpin peptidase inhibitor, clade B (ovalbumin), member 6	*173321 #613453	2893391-2917398	Member of the serpin (serine proteinase inhibitor) family, originally discovered as a placental thrombin inhibitor. Plays an important role in the inner ear in the protection against leakage of lysosomal content during stress. <b>Genotype-phenotype:</b> Deafness, autosomal recessive 91
<b>SERPINB9</b>	serpin peptidase inhibitor, clade B (ovalbumin), member 9	*601799	2832498-2848545	Member of the serpin (serine proteinase inhibitor) family. Endogenous inhibitor of caspase-1.
<b>NQO2</b>	NAD(P)H dehydrogenase, quinone 2	*160998 #114480	2945048-2965109	EC 1.10.99.2. NAD(P)H:quinone acceptor oxidoreductase-2, a member of the thioredoxin family of enzymes. Catalyzes the 2-electron reduction of various quinones, redox dyes, and the vitamin K menadione <b>Genotype-phenotype:</b> susceptibility to breast cancer
<b>RIPK1</b>	receptor (TNFRSF)-interacting serine-threonine kinase 1	*603453	3022056-3060420	A key signaling molecule in the programmed necrosis pathway, which plays important roles in development, tissue damage response, and antiviral immunity.
<b>BPHL</b>	biphenyl hydrolase-like	*603156	3063924-3098431	A hydrolytic enzyme that contains a serine residue in their active site.
<b>TUBB2A</b>	tubulin, beta class IIa	*615101 # 615763	3098900-3102782	Structural element of microtubules. Plays important role in mitosis, intracellular transport, neuron morphology, and ciliary and flagellar motility. Highest expression in brain, followed by gastrointestinal track and kidney. <b>Genotype-phenotype:</b> Cortical dysplasia, complex, with other brain malformations 5
<b>TUBB2B</b>	tubulin, beta class IIb	*612850 #610031	3169493-3172967	Structural element of microtubules. Plays important role in mitosis, intracellular transport, neuron morphology, and ciliary and flagellar motility. Expression predominantly in central and peripheral nervous system during neuronal migration and differentiation. <b>Genotype-phenotype:</b> Polymicrogyria, symmetric or asymmetric
<b>PSMG4</b>	proteasome (prosome, macropain) assembly chaperone 4	n/a	3204160-3213299	Chaperone protein which promotes assembly of the 20S proteasome

Gene	Name	OMIM ID	Chromosomal coordinates	Function of the encoded protein.
<b>SLC22A23</b>	solute carrier family 22, member 23	*611697	3214205-3401792	Member of a large family of transmembrane proteins that function as uniporters, symporters, and antiporters to transport organic ions across cell membranes
<b>PXDC1</b>	PX domain-containing protein 1	n/a	3667834-3697245	A protein with one phox-homology domain of unknown function.
<b>FAM50B</b>	family with sequence similarity 50, member B	*614686	3794630-3796550	A functional retrotransposon on chromosome 6 derived from the FAM50A gene on X chromosome. Its expression is regulated by the mechanism of parental (paternal) imprinting. The function of the encoded protein remains unknown, however its expression is observed during spermatogenesis and tumorigenesis.
<b>PRPF4B</b>	pre-mRNA processing factor 4B	*602338	3966567-4010216	The protein encoded is involved in pre-mRNA splicing and in signal transduction. Regarded as a CDK-like kinase (Clk) with homology to mitogen-activated protein kinases (MAPKs).
<b>FAM217A</b>	family with sequence similarity 217, member A	n/a	4013591-4024456	Encodes a protein of unknown function. Is most abundantly expressed in tissues rich in highly ciliated cells, such as olfactory sensory neurons, and is predicted to be important to cilia.
<b>ECI2</b>	peroxisomal D3,D2 enoyl-CoA isomerase	*608024	4060925-4080830	The protein encoded is a key mitochondrial enzyme involved in beta-oxidation of unsaturated fatty acids.
<b>CDYL</b>	chromodomain protein, Y-like	*603778	4651391-4900777	Autosomal homolog of chromodomain Y. Similarly, it possess a chromo-domain, a motif implicated in chromatin binding and gene suppression, and a catalytic domain believed to be involved in histone acetylation.
<b>RPP40</b>	ribonuclease P/MRP 40kDa subunit	*606117	4940278-4949296	An element of complex forming Ribonuclease P (RNase P) that removes the 5-prime leader sequences from precursor tRNA molecules.
<b>PPP1R3G</b>	protein phosphatase 1, regulatory subunit 3G	n/a	5030718-5032454	A glycogen-targeting subunit that in combination with protein phosphatase 1 controls glycogenesis in different organs.
<b>LYRM4</b>	LYR motif containing 4	*613311 #615595	5053651-5206182	A mitochondrial protein, member of the ISCU/NFS1/LYRM4 complex responsible for the biogenesis of Fe-S (iron- sulphur) clusters. It interacts directly with the complex activator frataxin. <b>Genotype-phenotype:</b> Combined oxidative phosphorylation deficiency 19; susceptibility to cognitive defect in schizophrenia
<b>FARS2</b>	mitochondrial phenylalanyl-tRNA synthetase 2	*611592 #614946	5206582-5716815	A member of the class II aminoacyl-tRNA synthetases that catalyze attachment of phenylalanine to its cognate tRNA.
<b>NRN1</b>	neuritin 1	*607409	5943230-5952837	A glycosylphosphatidylinositol (GPI)-anchored neuronal protein that functions extracellularly to promote neurite outgrowth and branching. It is a glutamate and neurotrophin receptor target gene.
<b>F13A1</b>	coagulation factor XIII, A1 subunit	*134570 #613255 #608446 #188050	6089309-6265923	EC 2.3.2.13. A subunit of factor XIII, the last zymogen to become activated in the blood coagulation cascade. <b>Genotype-phenotype:</b> factor XIII deficiency (bleeding disorder); protection against myocardial infarction, protection against venous thrombosis)
<b>LY86</b>	lymphocyte antigen 86	*605241	6533932-6600215	A leucine rich repeat (LRR)-bearing molecule that transmits an activation signal leading to massive B-cell proliferation as well as resistance against apoptosis.
<b>Non-coding mRNA molecules</b>				
<b>MIR6720</b>	microRNA 6720	n/a	1335547-1335645	no data
<b>MIR4645</b>	microRNA 4645	n/a	2799263-2799340	no data
<b>MIR3691</b>	microRNA 3691	n/a	5093465-5093555	no data
<b>LINC01011</b>	long intergenic non-protein coding RNA 1011	n/a	2933199-2936404	no data
<b>GMDS-AS1</b>	GMDS antisense RNA 1	n/a	2190985-2358824	no data
<b>LY86-AS1</b>	LY86 antisense RNA 1	n/a	6291696-6568058	no data

Gene	Name	OMIM ID	Chromosomal coordinates	Function of the encoded protein.
<b>Open reading frames</b>				
<b>C6orf195</b>	chromosome 6 open reading frame 195	n/a	2567970-2580297	Uncharacterized protein
<b>C6orf201</b>	chromosome 6 open reading frame 201	n/a	4024438-4075998	Uncharacterized protein
<b>SERPINB9P1</b>	serpin peptidase inhibitor, clade B member 9, pseudogene 1	n/a	2799889-2821743	pseudogene
<b>HTATSF1P2</b>	HIV-1 Tat specific factor 1 pseudogene 2	n/a	2965388-2970004	pseudogene