

Supplementary Table 1. Sequence of primers used for amplification of SPECC1L coding exons.

Primer name	Sequence (5' to 3')
SPECC1L-E3-For1	TCTAAAGCGCTTCCTCCAG
SPECC1L-E3-Rev1	AGTGTTCCCAGCACCAGAAG
SPECC1L-E4-For1	GAAGTTCTGTTTCTGTAGGACAG
SPECC1L-E4-Rev1	TGGATGAAATGATATGATGTCCAG
SPECC1L-E5_1-For1	ACAGACTCTAAGGCAAGATTATTG
SPECC1L-E5_1-Rev1	TCAGGGACTGATAGCCAAACAG
SPECC1L-E5_2-For1	AACAGAATACTGCCATCCGTG
SPECC1L-E5_2-Rev1	TGGTGACTGCGTTCTTTGAG
SPECC1L-E5_3-For1	GTCAGCAGAGCGATAAGTTGG
SPECC1L-E5_3-Rev1	CCTCTCACAGGCAGGAAAAC
SPECC1L-E6-For1	CTCCCCTTGAAACTGTGTG
SPECC1L-E6-Rev1	TCGGTTTTGAACTCCCAAAG
SPECC1L-E7-For1	ATACTGTGGAAGGAAGTGATTTAG
SPECC1L-E7-Rev1	GGAATATTTGCACTATACTTAACAG
SPECC1L-E8-For1	AGAAATAGATACTAGCAATTCTCTG
SPECC1L-E8-Rev1	CTCTGAAAGATCCACATAGATGG
SPECC1L-E9-For3	CTGATATAAAAATTTTCAGTATCATTTTC
SPECC1L-E9-Rev5	CAGTAAAGAATACCAAATCAGACC
SPECC1L-E10-For1	TCAGGCGAGTCCTTAAGTGG
SPECC1L-E10-Rev1	TCTAGATACATCCCTGCTGAGG
SPECC1L-E11-For1	TGTGTTACAACCTTACTATAGCAG
SPECC1L-E11-Rev1	ACTCTCTATTACCCATTATGACAG
SPECC1L-E12-For1	GTTGAAGTTCCTGGATTGG
SPECC1L-E12-Rev1	GGTGAGAGGACAGCTTGAGG
SPECC1L-E13-For1	ACTGCACCTGGCCTCACAG
SPECC1L-E13-Rev1	CACAGTGCTCAAATGCCATC
SPECC1L-E14-For1	CAGTTAGCAAAGAATTCTGACAAC
SPECC1L-E14-Rev1	GTGACACGTAGTAAGCTACGAC
SPECC1L-E15-For1	CAGGTAGCCCTTGGACACAG
SPECC1L-E15-Rev1	GATCCCAGAAGACCCAGATG
SPECC1L-E16-For1	AGATGCAGAAGCTCAGGCTC
SPECC1L-E16-Rev1	CTTTTACCAGGCAGCCACAC
SPECC1L-E17-For1	AGCAGAGTCTTGGATGATTTTCAG
SPECC1L-E17-Rev1	AGCCTTCATCAACCCTAGTGAG
SPECC1L-E9-For2	GCCAAGATGGTCTCGATCTCC
SPECC1L-E9-Rev2	AGGCTTCTGTCTAAGACTAACAC
SPECC1L-E9-For2	GCCAAGATGGTCTCGATCTCC
SPECC1L-E9-Rev3	CTCAAACCTTCTATTAAATATCACC

Supplementary Table 2. Rare Variants Fitting Autosomal Dominant Model Detected in Trio.

Gene	Genomic location (RefSeq)	MAF in 1000 Genomes Project	MAF in ESP6500SI	dbSNP	SIFT	Polyphen2	cDNA alteration	Protein alteration	GERP+ +	Other occurrence
FAN1	15:31222827 (NM_014967.4)	-	-	-	0	1	2869G>T	G957C	5.11	-
VAC14	16:70731102 (NM_018052.3)	-	-	-	0	0.992	1895C>T	T632M	5.05	-
SPECC1L	22:24718137 (NM_015330.4)	-	-	-	0	0.995	1189A>C	T397P	5.01	-
C9orf93	9:15920397 (NM_173550.2)	-	-	-	0	0.999	3730C>T	R1244C	4.12	-
PCSK5	9:78506117 (NM_001190482.1)	-	-	-	0	0.969	20G>A	C7Y	3.43	-
RGS22	8:101018334 (NM_015668.3)	-	-	-	0	0.956	2365G>A	E789K	2.52	-
AQP1	7:30951801 (NM_198098.2)	-	-	-	0	0.988	277C>T	R93C	2.47	-
TREH	11:118532621 (NM_007180.2)	-	-	-	0	-	502C>T	R168C	-	-
SP7	12:53722988 (NM_001173467.1)	-	-	-	0	-	238A>C	S80R	-	-
ZFHX2	14:23995258 (NM_033400.2)	-	-	-	0.05	-	3893G>A	G1298D	-	-
TLE2	19:3006609 (NM_001144761.1)	-	-	-	0	-	1351G>C	D451H	-	-
MFSD11	17:74763505 (NM_001242536.1)	-	-	-	-	-	565_567delCTT	L189-	-	-
SYT14	1:210187111 (NM_001146261.2)	-	-	-	-	-	331_336delAGTACA	ST111-	-	-
SMCHD1	18:2732423 (NM_015295.2)	0.0005	-	rs113434340	-	-	3209T>C	I1070T	-	-
KIDINS220	2:8925950 (NM_020738.2)	0.0005	-	rs144492511	0	0.983	2150C>T	S717L	5.2	-
CLGN	4:141331044 (NM_001130675.1)	-	0.000154	-	0.03	0.972	224T>C	V75A	5.71	-
CCDC40	17:78061565 (NM_001243342.1)	-	0.000079	-	0.02	1	2609G>A	R870H	3.33	-
TUBAL3	10:5436423 (NM_024803.2)	-	0.000077	-	0.02	-	398C>T	A133V	-	-
KCNAB2	1:6158552 (NM_001199860.1)	-	-	-	0	0.958	1022C>T	P341L	5.15	1
MTRF1L	6:153323630 (NM_001114184.1)	-	-	-	0	0.971	191T>C	L64S	4.55	1
S100A10	1:151955753 (NM_002966.2)	-	0.00246	rs142923959	0	0.985	180C>G	D60E	4.06	3
ZNF77	19:2934625 (NM_021217.2)	0.0005	0.002307	rs141883841	0	0.999	500G>A	C167Y	2.31	3
GOLGB1	3:121388135 (NM_001256488.1)	0.0014	0.001461	rs114155768	0.03	0.993	9005G>A	R3002H	3.23	3
KSR1	17:25924412 (NM_014238.1)	-	0.00016	-	0.04	-	1007C>T	T336M	-	5
SLCO4A1	20:61296353 (NM_016354.3)	-	-	-	-	-	1190_1191delICT	L398fsX551	-	6
DISP2	15:40659437 (NM_033510.1)	-	0.003767	rs140120816	0	0.997	1124A>T	Y375F	4.75	7
DOCK8	9:420579 (NM_001190458.1)	0.0018	0.002614	rs116920018	0	0.999	3719A>G	Y1240C	5.68	7