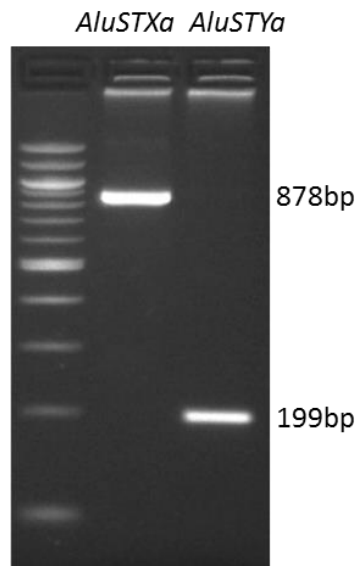
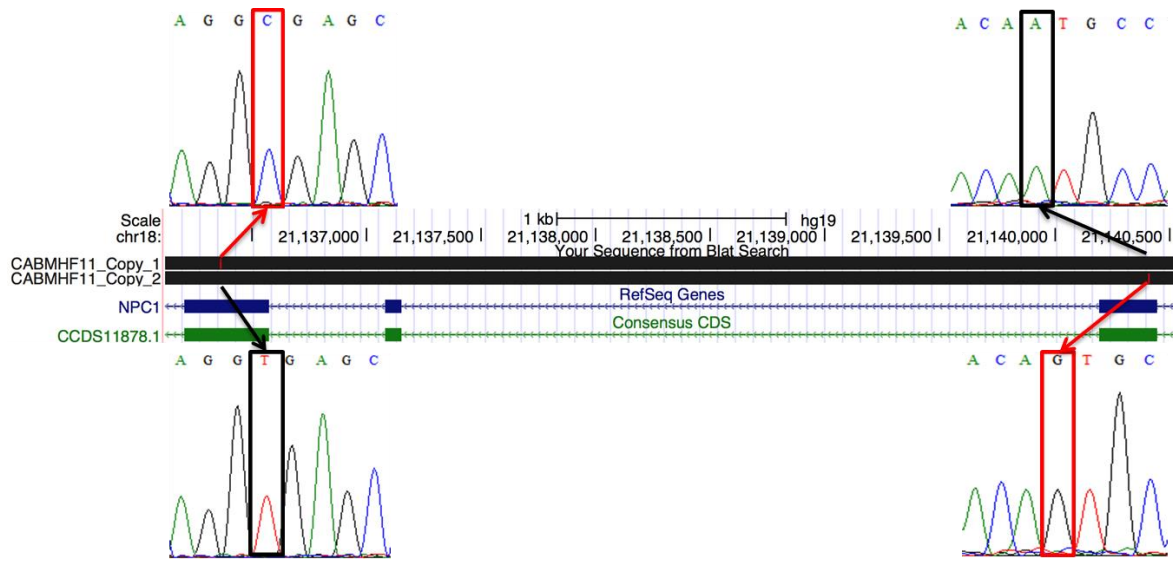


**Supplemental Figure S1. Electropherograms for variants in genes encoding GLA and SMPD1 in case 95RD414.** (a) Variant “X,100653420,C,A,95RD414” on gene *GLA* was heterozygous with signals of both reference allele (G) and alternative allele (T) on the forward strand. Sequencing of the reverse strand confirmed the heterozygous signals of both reference allele (C) and alternative allele (A). (b) Variant “11,6413175,C,A,95RD414” on the gene *SMPD1* was homozygous with the predominant signal of mutant allele (A) on the forward strand and predominant signal of mutant allele (T) on the reverse strand.



**Supplemental Figure S2. Gender determination for 95RD414.** The banding patterns indicate the patient is a female.



**Supplemental Figure S3. Compound Heterogeneity of variants in the gene encoding NPC1 in case CABMHF1.** Two copies of gene *NPC1* in case CABMHF11 are shown as the thick solid black track. Two mutants on different copies are marked in red bar. Red arrows lead to the electropherogram of the mutant allele (circled by red box) and its flanking sequence, while black arrows lead to the mutant-corresponding reference allele (circled by black box) and its flanking sequence. The RefSeq gene annotation for *NPC1* is shown in blue and the CCDS coding sequence annotation is shown in green. Exonic regions are shown as solid boxes, while non-exonic regions are shown as thin lines, with arrows indicating the direction of the gene.

**Table S1. LSD candidate genes.**

A1BG	AEBP1	AP2B1	ARFRP1	ASAP1	ATP6V0E1	BLOC1S5	CAV2	CFH
A2M	AFF4	AP2M1	ARGLU1	ASAP2	ATP6V0E2	BLOC1S6	CAV3	CFHR4
AAGAB	AFM	AP2S1	ARHGAP12	ASAP3	ATP6V1A	BLVRB	CBL	CFTR
AAK1	AFMID	AP3B1	ARHGAP27	ASGR1	ATP6V1B1	BMP1	CBLB	CHAF1A
AATF	AGA	AP3B2	ARHGAP33	ASGR2	ATP6V1B2	BNIP1	CBLC	CHCHD10
ABCA1	AGAP1	AP3D1	ARHGDI1A	ASIC4	ATP6V1C1	BORCS5	CBR3	CHI3L1
ABCA2	AGAP2	AP3M1	ARL1	ASNA1	ATP6V1C2	BORCS6	CC2D1B	CHIA
ABCA3	AGFG1	AP3M2	ARL10	ASNS	ATP6V1D	BPTF	CCDC109B	CHID1
ABCA5	AGPAT5	AP3S1	ARL11	ASPH	ATP6V1E1	BRI3	CCDC122	CHIT1
ABCA6	AGTRAP	AP3S2	ARL13A	ASPN	ATP6V1E2	BSG	CCDC22	CHM
ABCA7	AHCY	AP4B1	ARL13B	ASXL2	ATP6V1F	BST1	CCDC53	CHML
ABCB6	AHCYL2	AP4E1	ARL14	ATF4	ATP6V1G1	BST2	CCDC93	CHMP1A
ABCB9	AHNAK	AP4M1	ARL14EP	ATG10	ATP6V1G2	BTD	CCNT1	CHMP1B
ABCC10	AHSG	AP4S1	ARL14EPL	ATG12	ATP6V1G3	C10orf54	CCR5	CHMP2A
ABCC2	AKR1A1	AP5B1	ARL15	ATG13	ATP6V1H	C12orf49	CCZ1	CHMP2B
ABCD4	AKR1B10	AP5M1	ARL16	ATG14	ATP7B	C12orf66	CCZ1B	CHMP3
ABHD17B	AKR1C1	AP5S1	ARL17A	ATG16L1	ATP8A1	C16orf62	CD163	CHMP4A
ABHD5	AKT1S1	AP5Z1	ARL17B	ATG16L2	ATP8B1	C18orf8	CD164	CHMP4B
ABL1	ALDH18A1	APC2	ARL2	ATG2A	ATP9A	C19orf24	CD177	CHMP4C
ABL2	ALDH3A2	APCS	ARL2BP	ATG2B	ATP9B	C19orf47	CD180	CHMP5
ACAD11	ALDOA	APEH	ARL3	ATG3	ATPIF1	C1GALT1	CD1B	CHMP6
ACAN	ALG1	APEX1	ARL4A	ATG4A	ATRAID	C1GALT1C1	CD1D	CHPT1
ACAP1	ALPI	APH1A	ARL4C	ATG4B	ATRN	C1orf43	CD1E	CHST15
ACAP2	ALPP	APLP2	ARL4D	ATG4C	ATXN2	C1QB	CD207	CISD2
ACAP3	ALS2	APOA1	ARL5A	ATG4D	AXL	C1QTNF5	CD22	CLCN1
ACE	AMBRA1	APOA5	ARL5B	ATG5	AZU1	C1R	CD2AP	CLCN2
ACOT9	AMDHD2	APOB	ARL5C	ATG7	B4GALT5	C1S	CD302	CLCN3
ACP1	AMOT	APOC3	ARL6	ATG9A	BACE2	C2orf72	CD36	CLCN4
ACP2	AMPH	APOC4	ARL6IP1	ATG9B	BAIAP2L1	C3	CD37	CLCN5
ACP5	AMY2B	APOD	ARL6IP4	ATHL1	BAIAP2L2	C3orf58	CD4	CLCN6
ACPP	ANG	APOE	ARL6IP5	ATP11A	BAK1	C4B	CD59	CLCN7
ACSF2	ANGPT1	APOF	ARL6IP6	ATP11C	BAX	C5	CD5L	CLCNKA

ACSF3	ANKFY1	APP	ARL8A	ATP13A2	BCAP31	C5AR1	CD63	CLCNKB
ACSS1	ANKRD12	APPL1	ARL8B	ATP2A1	BCAS1	C6orf136	CD68	CLEC11A
ADA	ANO1	AQP8	ARL9	ATP5A1	BCAT2	C6orf58	CD74	CLEC4F
ADAM28	ANPEP	ARAP1	ARMCX1	ATP5C1	BCHE	C7orf55-LUC7L2	CDC42	CLEC4G
ADAM8	ANXA5	ARAP2	ARMCX3	ATP5F1	BCL10	C9	CDH1	CLIC1
ADAMDEC1	ANXA7	ARAP3	ARMCX6	ATP5I	BCR	C9orf116	CDH13	CLINT1
ADAMTSL2	AOAH	ARF1	ARMS2	ATP5J2	BDH2	C9orf72	CDH3	CLN3
ADAP1	AP1AR	ARF3	ARPC5	ATP5O	BECN1	C9orf91	CDK4	CLN5
ADAR	AP1B1	ARF4	ARRB1	ATP6AP1	BGN	CA2	CDK7	CLN6
ADCY5	AP1G1	ARF5	ARRB2	ATP6AP1L	BHLHE40	CA6	CDS2	CLN8
ADD1	AP1G2	ARF6	ARSA	ATP6AP2	BHMT	CACFD1	CEACAM1	CLTA
ADIPOQ	AP1M1	ARFGAP1	ARSB	ATP6V0A1	BIN1	CALR	CEACAM3	CLTB
ADRA2A	AP1M2	ARFGAP2	ARSD	ATP6V0A2	BIRC6	CALU	CELA1	CLTC
ADRB1	AP1S1	ARFGAP3	ARSG	ATP6V0A4	BLK	CAMP	CELSR3	CLTCL1
ADRB2	AP1S2	ARFGEF1	ARSK	ATP6V0B	BLOC1S1	CANT1	CES1	CLU
ADRB3	AP1S3	ARFGEF2	ART5	ATP6V0C	BLOC1S2	CARNS1	CES2	CMA1
ADRBK1	AP2A1	ARFIP1	ASAH1	ATP6V0D1	BLOC1S3	CAT	CETN2	CMPK2
ADRBK2	AP2A2	ARFIP2	ASAH2	ATP6V0D2	BLOC1S4	CAV1	CFB	CNP
COCH	CSF1R	DAG1	DRAM1	EPG5	FCGRT	GABARAPL2	GNAT2	HAAO
COL11A1	CSMD1	DAGLB	DRAM2	EPHX1	FCN1	GALC	GNB1	HABP2
COL11A2	CSN3	DAP	DSC3	EPN1	FCN2	GALNS	GNB2	HADHA
COL18A1	CSNK2A1	DAPK1	DSCR3	EPN2	FDX1L	GALNT10	GNB3	HAPLN1
COL1A1	CSNK2B	DAPK2	DST	EPN3	FETUB	GALNT2	GNB4	HAPLN4
COL1A2	CSRP1	DAPK3	DTNBP1	EPRS	FGD1	GALNT5	GNE	HCK
COL2A1	CSRP3	DCD	DUSP1	EPS15	FGD2	GALNT6	GNPDA1	HCLS1
COL3A1	CST3	DCN	DUSP10	EPS15L1	FGD4	GAS6	GNPTAB	HDAC4
COL5A1	CST4	DCTN1	DUSP27	EPX	FGFR2	GATA2	GNPTG	HDAC5
COL5A2	CST6	DCTN4	DUSP3	ERAP1	FGFR3	GATAD2B	GNS	HDAC6
COL6A1	CST7	DDR2	DVL2	ERAP2	FGFR4	GATM	GOLGA4	HEPH
COL7A1	CSTA	DDX3X	DYM	ERBB2	FGG	GBA	GOLGA5	HERC1
COL8A1	CSTB	DECR2	DYNC1H1	ERBB3	FGL1	GBAS	GOLGB1	HEXA
COLEC11	CTBS	DEFA5	DYRK1B	ERBB4	FHL1	GBE1	GOLIM4	HEXB
COMMD1	CTDSP1	DEPDC1	ECE1	ERLIN1	FIG4	GBF1	GOLM1	HFE

COMMD10	CTDSPL	DEPDC5	ECM1	ERP27	FIS1	GDAP2	GPD2	HGS
COMMD2	CTNS	DEPTOR	EDEM2	ERP29	FKBP10	GDF3	GPLD1	HGSNAT
COMMD3	CTSA	DGAT2L6	EDNRB	ERRFI1	FKBP14	GEM	GPNMB	HHIPL1
COMMD4	CTSB	DHDDS	EEA1	ESD	FKBP15	GET4	GPR137	HIF1A
COMMD5	CTSC	DHPS	EEF1A1	ETFDH	FKBP7	GGA1	GPR137B	HIP1
COMMD6	CTSD	DHRS2	EEF2	F10	FLCN	GGA2	GPR155	HIP1R
COMMD7	CTSE	DHX35	EFHD2	F12	FLNC	GGA3	GPX1	HIST1H1D
COMMD8	CTSF	DIP2B	EGF	F2R	FLOT1	GGH	GPX5	HK1
COMMD9	CTSG	DIRC2	EGFR	FAAH	FLOT2	GHR	GPX6	HK2
COMP	CTSH	DKK3	EHD1	FAF1	FLT1	GIF	GRHPR	HLA-A
COMTD1	CTSK	DLL1	EHD2	FAHD1	FMN2	GIGYF2	GRK1	HLA-B
COPS7A	CTSO	DLX2	EHD3	FAM118B	FMOD	GIMAP1	GRK4	HLA-C
COQ4	CTSS	DMBT1	EHD4	FAM132B	FMR1	GIMAP5	GRK5	HLA-DMA
COQ6	CTSV	DMXL2	EHHADH	FAM134A	FN1	GIT1	GRK6	HLA-DMB
COX4I2	CTSW	DNAJB12	EI24	FAM20A	FNBP1	GIT2	GRK7	HLA-DOA
CP	CTSZ	DNAJC11	EIF3A	FAM20C	FNIP2	GKN1	GRN	HLA-DOB
CPA2	CTTN	DNAJC13	EIF3B	FAM214B	FOLR1	GLA	GRSF1	HLA-DQA1
CPA3	CUBN	DNAJC15	EIF3K	FAM21C	FOXO1	GLB1	GSAP	HLA-DRA
CPA5	CUTA	DNAJC16	EIF4B	FAM3A	FOXO1	GLB1L	GSDMA	HLA-DRB5
CPB2	CUX1	DNAJC5	EIF4E	FAM3C	FOXO3	GLCE	GSN	HLA-E
CPD	CXCL12	DNAJC6	EIF4G1	FAM45A	FOXO4	GLG1	GSTM1	HLA-F
CPM	CXCR1	DNAJC8	EIF5A2	FAM50A	FOXO6	GLMP	GSTO1	HLA-G
CPN1	CXCR2	DNASE1	ELANE	FAN1	FOXRED2	GLS	GSTT1	HMOX1
CPNE2	CXCR4	DNASE1L1	ENDOD1	FAP	FRRS1	GLT8D1	GTF2A1	HNF4A
CPNE5	CYB561A3	DNASE1L3	ENPEP	FASN	FSD1L	GLTPD2	GTF2H1	HNRNPA1
CPQ	CYB5A	DNASE2	ENPP1	FASTKD3	FTH1	GM2A	GUF1	HNRNPA2B1
CPS1	CYB5R1	DNM1	ENPP2	FBLN1	FTL	GMNN	GUK1	HNRNPA3
CPVL	CYBB	DNM1L	ENPP3	FBLN2	FUCA1	GMPR	GUSB	HNRNPD
CR1	CYP2C18	DNM2	ENPP4	FBN1	FUCA2	GMPR2	GXYLT1	HNRNPH2
CR2	CYP2C9	DNM3	ENPP5	FBXL2	FUNDC1	GNA11	GYS2	HOXB7
CREG1	CYP2D6	DOC2A	ENTPD4	FBXO6	FUNDC2	GNA13	GZMA	HOXC13
CRIM1	CYP4A22	DOLK	ENTPD6	FBXO7	FUS	GNA14	GZMB	HP
CRIP2	CYTH2	DPP4	ENTPD8	FCER1G	FYCO1	GNAI1	GZMK	HPS1

CRP	D2HGDH	DPP7	EPB41L5	FCGBP	GAA	GNAI3	GZMM	HPS3
CSAD	DAB2	DPY19L2	EPDR1	FCGR3A	GABARAP	GNAQ	H2AFZ	HPS4
HPS5	INHBB	KIAA1161	LNPEP	MASP1	MRPL27	NAGLU	NSMCE2	PARK7
HPS6	INHBE	KIAA1324	LONP1	MAX	MRPL30	NAGPA	NT5DC2	PARP14
HPSE	INPP1	KIAA1715	LPCAT2	MBD5	MRPS15	NAPA	NT5E	PARVG
HPX	INPP4A	KIAA1919	LPIN2	MCAM	MRPS18C	NAPG	NTHL1	PBLD
HRAS	INPP4B	KIF16B	LPL	MCM2	MRPS2	NAPSA	NTRK1	PBXIP1
HRG	INPP5A	KIT	LPO	MCOLN1	MRPS25	NARF	NUDCD3	PCDHB14
HS2ST1	INPP5B	KLF15	LRBA	MCOLN2	MRRF	NBR1	NUDT13	PCDHGA2
HSDL1	INPP5D	KLHL12	LRG1	MCOLN3	MRS2	NCEH1	NUMB	PCOLCE
HSP90AA1	INPP5E	KLHL9	LRIG1	MDM2	MS4A1	NCF2	NUMBL	PCSK5
HSPA13	INPP5F	KLK1	LRP1	ME2	MSR1	NCF4	NXPE2	PCSK9
HSPA1A	INPP5J	KNG1	LRP1B	MED26	MT3	NCK2	OAS2	PCYOX1
HSPA1B	INPP5K	KRT18	LRP2	MED28	MTCH1	NCSTN	OCRL	PDCD6IP
HSPA1L	INPPL1	KRT8	LRP3	MEGF9	MTDH	NDFIP1	OGDH	PDE6C
HSPA2	INSIG1	KXD1	LRP5L	MET	MTHFD1L	NDFIP2	OGDHL	PDGFB
HSPA5	IQSEC1	LACTB2	LRP6	MFGE8	MTHFD2	NDRG1	OLFML1	PDGFRA
HSPA6	IQSEC2	LAMP1	LRP8	MFSD1	MTHFR	NDST1	OLFML2B	PDGFRB
HSPA8	IQSEC3	LAMP2	LRPAP1	MFSD11	MTM1	NDUFA9	OLR1	PDIA2
HSPBAP1	IRGM	LAMP3	LRRK2	MFSD12	MTMR1	NECAP1	OMD	PK3
HTR2B	ITCH	LAMTOR1	LSS	MFSD5	MTMR10	NECAP2	OMG	PDLIM7
HTRA1	ITFG1	LAMTOR2	LUC7L3	MFSD7	MTMR11	NEDD4	OPLAH	PDP2
HYAL1	ITFG2	LAMTOR3	LUM	MFSD8	MTMR12	NEDD4L	OPTN	PEA15
HYAL2	ITGA8	LAMTOR4	LXN	MGAM	MTMR14	NEFH	ORAI1	PEAR1
HYAL3	ITGAL	LAMTOR5	LY6E	MGARP	MTMR2	NEFL	OSBP	PEBP1
IAH1	ITGB2	LAPTM4A	LY86	MGAT1	MTMR3	NES	OSBP2	PEBP4
IDS	ITGB5	LAPTM4B	LY96	MGAT3	MTMR4	NEU1	OSBPL10	PER3
IDUA	ITIH1	LAPTM5	LYPD8	MGAT4B	MTMR6	NEU3	OSBPL11	PES1
IFI30	ITIH3	LATS2	LYPLA2	MGP	MTMR7	NEU4	OSBPL1A	PEX11G
IFITM1	ITIH4	LBP	LYST	MGST1	MTMR8	NEURL2	OSBPL2	PEX3
IFNGR1	ITM2B	LCAT	LYZ	MICALCL	MTMR9	NGFRAP1	OSBPL3	PFKFB2
IFT27	ITM2C	LCN2	LZTS2	MICU2	MTOR	NGLY1	OSBPL5	PGBD5
IGF1R	ITSN1	LCN8	M6PR	MINPP1	MTSS1	NHLRC3	OSBPL6	PGLYRP2

IGF2R	ITSN2	LDHC	MAFF	MIOS	MVB12A	NHSL2	OSBPL7	PGRMC1
IGFALS	JAK1	LDLR	MAN2A1	MITF	MVB12B	NIPA2	OSBPL8	PHB
IGFBP1	JARID2	LDLRAP1	MAN2A2	MLPH	MVP	NIPSNAP1	OSBPL9	PHB2
IGFBP3	JCHAIN	LEPROT	MAN2B1	MLST8	MYADM	NIT1	OSGEP	PHF20L1
IGFBP5	KANSL1L	LEPROTL1	MAN2B2	MLX	MYBPC2	NKX2-1	OSTM1	PHKG2
IGFBP6	KCNAB2	LGALS3BP	MAN2C1	MMP14	MYH1	NMT2	OXCT1	PI4K2A
IGFBP7	KCNE1	LGALS9B	MANBA	MMP2	MYH11	NOA1	P2RX4	PI4K2B
IHH	KCNE2	LGMN	MAP1A	MMP8	MYH2	NOL8	P4HB	PI4KA
IL16	KCNK6	LIFR	MAP1LC3A	MMP9	MYLPF	NOP14	PA2G4	PI4KB
IL2RA	KCTD12	LIN37	MAP1LC3B	MMS19	MYO1C	NOS3	PABPC4L	PIAS4
IL2RB	KCTD7	LIN52	MAP1LC3B2	MON1A	MYO3A	NPC1	PACS2	PICALM
IL2RG	KDELC1	LIPA	MAP1LC3C	MON1B	MYO5A	NPC2	PACSIN3	PIGR
IL4I1	KDELC2	LIPC	MAP3K13	MPEG1	MYO5B	NPRL2	PAH	PIK3C2A
IL4R	KDR	LIPT1	MAPK1	MPO	MYO6	NPTN	PAM	PIK3C2B
IL6R	KEAP1	LITAF	MAPK3	MRAS	MYO7A	NRBF2	PARD3	PIK3C2G
IL6ST	KERA	LMAN1	MAPK8IP3	MRC1	NAAA	NRCAM	PARD6A	PIK3C3
ILF3	KIAA0196	LMAN2	MAPT	MREG	NAALADL2	NRXN1	PARD6B	PIK3CA
ILVBL	KIAA0513	LMBRD1	MARCH2	MRFAP1	NAGA	NSD1	PARD6G	PIK3CB
IMPAD1	KIAA1033	LMTK2	MARCO	MROH6	NAGK	NSF	PARK2	PIK3CD
PIK3CG	PLSCR3	PSEN2	RAB32	RAC1	RNF167	SCAMP3	SIGMAR1	SLC38A2
PIK3R1	PLTP	PSENN	RAB33A	RAD23A	RNF185	SCARB1	SIL1	SLC38A7
PIK3R2	PM20D2	PSTPIP1	RAB33B	RAD9A	RNF41	SCARB2	SIRT1	SLC38A9
PIKFYVE	PMPCA	PTBP1	RAB34	RAMP2	RNH1	SCARF1	SIRT2	SLC39A1
PIM2	PNLIPRP1	PTEN	RAB35	RAMP3	RPH3A	SCO2	SKAP2	SLC39A3
PINK1	POFUT1	PTGDS	RAB36	RANGRF	RPL13	SCP2	SLC11A1	SLC39A8
PIP	POFUT2	PTH1R	RAB37	RAP2B	RPL23	SCPEP1	SLC11A2	SLC40A1
PIP4K2A	POGLUT1	PTP4A3	RAB38	RARRES1	RPL3	SDC2	SLC12A3	SLC44A2
PIP4K2B	POLQ	PTPDC1	RAB39A	RASEF	RPL5	SDCBP	SLC12A4	SLC46A1
PIP4K2C	POLR3C	PTPRB	RAB39B	RASGRF2	RPL6	SDF4	SLC12A6	SLC46A3
PIP5K1A	PON1	PTPRF	RAB3A	RASGRP3	RPL7	SDHC	SLC12A9	SLC48A1
PIP5K1B	PON2	PTPRZ1	RAB3B	RB1CC1	RPL7A	SEBOX	SLC15A3	SLC4A2
PIP5K1C	POR	PTTG1IP	RAB3C	RBL2	RPL8	SEC14L3	SLC15A4	SLC6A17
PIPSL	POSTN	PUSL1	RAB3D	RBM15	RPLP1	SEC14L4	SLC16A6	SLC7A5

PITPNA	PPA1	PYCR2	RAB3GAP2	RBM19	RPN1	SEH1L	SLC17A5	SLC8B1
PITPNB	PPARGC1A	PYGM	RAB3IL1	RBM39	RPN2	SEMA3A	SLC20A1	SLCO1B3
PITPNC1	PPCDC	PZP	RAB3IP	RBP1	RPS17	SEMA3G	SLC22A8	SLCO2B1
PITPNM1	PPCS	QSOX1	RAB40A	RBSN	RPS23	SEMA4B	SLC22A9	SLPI
PITPNM3	PPP1R18	RAB10	RAB40AL	RC3H2	RPS25	SERINC1	SLC25A13	SMAP1
PKLR	PPP1R3B	RAB11A	RAB40B	RCBTB1	RPS26	SERINC2	SLC25A16	SMAP2
PKM	PPP2R3C	RAB11B	RAB40C	RCBTB2	RPS27	SERPINA1	SLC25A24	SMCR8
PKN1	PPT1	RAB11FIP1	RAB41	RDH14	RPS28	SERPINA11	SLC25A26	SMN1
PLA2G15	PPT2	RAB11FIP2	RAB42	RDH16	RPS3	SERPINA3	SLC25A28	SMNDC1
PLA2G2A	PQLC2	RAB11FIP3	RAB43	RECQL5	RPS3A	SERPINA4	SLC25A3	SMPD1
PLA2G6	PRCP	RAB11FIP4	RAB44	REG4	RPS4X	SERPINA5	SLC25A30	SMPDL3A
PLA2G7	PRDX1	RAB11FIP5	RAB4A	RELB	RPS4Y2	SERPINA6	SLC25A32	SMPDL3B
PLAC8	PRDX6	RAB12	RAB4B	RELN	RPS6	SERPINB11	SLC25A33	SMURF1
PLAU	PRELID1	RAB13	RAB5A	REN	RPS6KA3	SERPINB8	SLC25A35	SMURF2
PLBD1	PRF1	RAB14	RAB5B	RET	RPTN	SERPINE1	SLC25A36	SNAP25
PLBD2	PRG2	RAB15	RAB5C	RFESD	RPTOR	SERPINE2	SLC25A37	SNAP29
PLCB1	PRKAA1	RAB17	RAB6A	RGS19	RPUSD4	SERPINF1	SLC25A38	SNAP91
PLCG1	PRKAB1	RAB18	RAB6B	RHEBL1	RRAGA	SERPINF2	SLC25A4	SNAPIN
PLCG2	PRKAG1	RAB19	RAB6C	RHOB	RRAGB	SERPING1	SLC25A51	SNCA
PLCL2	PRKAG2	RAB1A	RAB7A	RHOG	RRAGC	SERPINI2	SLC26A11	SNF8
PLD1	PRKCI	RAB1B	RAB8A	RILP	RRAGD	SETX	SLC29A3	SNRPA
PLD2	PRKCZ	RAB20	RAB8B	RILPL1	RRAS	SFTPD	SLC2A13	SNRPC
PLD3	PRL	RAB21	RAB9A	RILPL2	RRAS2	SFXN3	SLC2A6	SNX1
PLD4	PRLR	RAB22A	RAB9B	RIMS1	RSAD2	SGIP1	SLC2A8	SNX10
PLEKHA8	PRMT3	RAB23	RABAC1	RIMS2	RUBCN	SGOL1	SLC30A2	SNX11
PLEKHM1	PROS1	RAB24	RABEP1	RIN1	RUFY1	SGSH	SLC30A4	SNX12
PLEKHO2	PRPH	RAB25	RABEP2	RIN2	RUFY3	SH3GL1	SLC31A1	SNX13
PLIN2	PRSS1	RAB26	RABEPK	RIN3	RUNX2	SH3GL2	SLC31A2	SNX14
PLOD1	PRSS16	RAB27A	RABGAP1	RNASE2	RYR2	SH3GL3	SLC33A1	SNX15
PLOD2	PRTN3	RAB27B	RABGAP1L	RNASE3	S100A6	SH3GLB1	SLC34A1	SNX16
PLOD3	PSAP	RAB28	RABGEF1	RNASE4	SACM1L	SH3GLB2	SLC35A4	SNX17
PLP1	PSD	RAB29	RABIF	RNASE6	SAE1	SH3KBP1	SLC35A5	SNX18
PLP2	PSD2	RAB2A	RABL2A	RNASEK	SAMD10	SH3TC2	SLC35F6	SNX19



PLS3	PSD3	RAB2B	RABL2B	RNASET2	SBF1	SHMT1	SLC36A1	SNX2
PLSCR1	PSD4	RAB30	RABL3	RNF13	SBF2	SIAE	SLC37A2	SNX20
PLSCR2	PSEN1	RAB31	RABL6	RNF146	SCAMP2	SIDT2	SLC37A3	SNX21
SNX22	ST3GAL1	SULT1C2	THBS4	TNFAIP3	UBE2Q1	VPS35	ZNF512	
SNX24	ST3GAL3	SULT2A1	THNSL1	TNNC1	UBE4B	VPS36	ZNF57	
SNX25	ST6GAL1	SUMF1	TIMM13	TNPO2	UBL3	VPS37A	ZNF706	
SNX27	ST6GALNAC2	SUMF2	TIMP2	TNR	UBQLN2	VPS37B	ZNF789	
SNX29	ST8SIA2	SUPT16H	TKTL1	TNXB	UCP2	VPS37C	ZNF84	
SNX3	ST8SIA3	SUPT20H	TLDC1	TOLLIP	UGDH	VPS37D	ZNRF1	
SNX30	STAB1	SUPV3L1	TLR2	TOM1	UGP2	VPS39	ZNRF2	
SNX31	STAB2	SUV39H1	TLR3	TOMM40	UGT2B10	VPS41		
SNX32	STAM	SVIL	TLR7	TOP3B	UHRF2	VPS45		
SNX33	STAM2	SYNGR1	TLR9	TOR3A	ULK1	VPS4A		
SNX4	STAMBP	SYNGR2	TM7SF3	TP53INP1	ULK2	VPS4B		
SNX5	STARD10	SYNJ1	TM9SF1	TPCN1	ULK3	VPS8		
SNX6	STARD3	SYNJ2	TM9SF2	TPCN2	UNC93B1	VSIG4		
SNX7	STARD3NL	SYNRG	TM9SF4	TPP1	UPP1	VTA1		
SNX8	STEAP3	SYP	TMBIM1	TPP2	UQCRC1	VTI1B		
SNX9	STK10	SYPL1	TMC6	TPRA1	UQCRC2	VTN		
SOCS5	STK11	SYPL2	TMED10	TPRG1L	UROC1	VWA5A		
SOD1	STK11IP	SYT1	TMED7	TPSAB1	UROD	VWF		
SOD3	STK17B	SYT10	TMED9	TPST1	USP10	WASL		
SORD	STOM	SYTL2	TMEM104	TRAF6	USP16	WBP2		
SORL1	STON1	SZT2	TMEM106A	TRAPPC2L	USP2	WDR11		
SOST	STON2	TACC2	TMEM106B	TRIM13	USP32	WDR24		
SPACA3	STS	TAF6L	TMEM127	TRIM14	USP8	WDR26		
SPARC	STX10	TAGLN2	TMEM135	TRIM23	UTP18	WDR41		
SPARCL1	STX11	TAGLN3	TMEM141	TRIM25	UVRAG	WDR45		
SPATA18	STX12	TARDBP	TMEM144	TRIM63	VAC14	WDR45B		
SPATS2L	STX16	TBC1D1	TMEM175	TRIM7	VAMP2	WDR48		
SPG11	STX17	TBK1	TMEM176B	TRIP6	VAMP3	WDR59		
SPG20	STX18	TCEAL1	TMEM179B	TRPM7	VAMP4	WDR6		
SPG21	STX19	TCEAL8	TMEM192	TSG101	VAMP7	WDR81		

SPHK2	STX1A	TCIRG1	TMEM2	TSPAN1	VAMP8	WDR91		
SPINT2	STX1B	TCN2	TMEM205	TSPAN3	VAPB	WIPI1		
SPNS1	STX2	TDG	TMEM237	TSPAN6	VASN	WIPI2		
SPOCD1	STX3	TECPR1	TMEM256	TSPAN7	VAT1	WWP1		
SPON1	STX4	TECPR2	TMEM50A	TSP0	VCP	WWP2		
SPP2	STX5	TEX10	TMEM50B	TTC9B	VDAC1	XIRP2		
SPPL2A	STX6	TF	TMEM55A	TTR	VEPH1	XPNPEP2		
SPTBN1	STX7	TFAP2A	TMEM55B	TTYH2	VMA21	XRCC6BP1		
SQRDL	STX8	TFE3	TMEM59	TTYH3	VMP1	YTHDF1		
SQSTM1	STXBP1	TFEB	TMEM63A	TUFM	VNN1	ZBTB8OS		
SRA1	STXBP2	TFEC	TMEM63B	TXNRD1	VNN3	ZC3H11A		
SRC	STXBP3	TFR2	TMEM65	TYR	VPS11	ZC3H18		
SRGAP1	STXBP4	TFRC	TMEM74	TYRP1	VPS13C	ZCCHC8		
SRGN	STXBP5	TG	TMEM8A	U2AF1	VPS16	ZDHHC3		
SRL	STXBP5L	TGFBR2	TMEM9	U2AF2	VPS18	ZFYVE16		
SRPX	STXBP6	TGFBR3	TMEM92	UBA52	VPS25	ZFYVE26		
SRRD	SUB1	TGFBRAP1	TMOD2	UBAP2	VPS26A	ZFYVE9		
SSR1	SULF2	TGM2	TMPRSS11B	UBAP2L	VPS28	ZG16		
SSR3	SULT1A1	TGOLN2	TMPRSS7	UBB	VPS29	ZNF292		
ST14	SULT1B1	THAP8	TMTC4	UBE2B	VPS33A	ZNF507		

**Table S2. Sequencing primers for Sanger validation**

Case	Gene ID	Chr	Position	Forward Primer	Reverse Primer	length (bp)	length (bp)*
00RD098	<i>GLB1</i>	3	33099692	TTCCCTGCTCTTTTTCACTCACAG	CTGCAATTTCTGTTACTACAAACACC	235	
00RD098	<i>KRIT1</i>	7	91870373	TACCTAGGGAACACTACTTCACA	AAACTGTAAGAACCAGGCCCC	595	
82RD265	<i>SLC31A1</i>	9	116021039	<b>CAAGCAGTCTGACCAAAAGGT</b>	CAGGCATGGAATTGTAGCGAA	385	<b>2096</b>
82RD265	<i>SLC31A1</i>	9	116022721	AAGTACCCATGAGTTGCCAGA	<b>CTTCAACAACCTCCCACTGCA</b>	382	
95RD414	<i>GLA</i>	X	100653420	ATGGCTGCTCCTTTATTCATGT	AAACCAAGAAAGTGTGGTTGCT	397	
95RD414	<i>SMPD1</i>	11	6413175	TGACTGTGCAGACCCACTGT	TGCTTTCATGGTTACCCACA	308	
CABMHF1	<i>NPC1</i>	18	21136367	TGATTCCTGCCATGAGATAGCAACT	<b>CCCATCTAGCAGTAGTCAACATGTA</b>	556	<b>4415</b>
CABMHF1	<i>NPC1</i>	18	21140411	<b>GTATTCAGTGGGCTTTTCTTTGAGT</b>	CATGGAGGTATTTGTTTCTTGTCTA	457	
CABMHF1	<i>SMPD1</i>	11	6415259	CACCATCCCTGTTGTCCCATGGAGT	CACAGGGCTCCGAGGGTGGGT	713	
CABMHF3	<i>TBCK</i>	4	107183332	GAGGAGTATACTGGGACGCCA	TAGGTTAGTTGCACTCACCACC	719	

\* Bolded primers were used to amplify large fragments that contain both variants