

Table S1. Genes in the targeted resequencing panel.

AARS2	CTSA	FIG4	IFIH1	PIGT	SDHAF1
ABCC1	CTSD	FOLR1	ISCA2	PITX2	SERPINC1
ABCC6	CYP27A1	FOXC1	ITM2B	PLP1	SERPINE1
ABCD1	CYP2U1	FUCA1	JAM3	POLG	SLC16A2
ACP5	DARS	GALC	KARS	POLH	SLC17A5
ACTA2	DARS2	GAN	KIF5A	POLR1C	SLC2A10
ACVRL1	DDB2	GBA	L2HGDH	POLR3A	SLC46A1
ADA2	DHFR	GBE1	LAMB1	POLR3B	SNORD118
ADAR	EARS2	GCDH	LARS2	POLR3K	SOX10
AIMP1	EGR2	GFAP	LMNB1	PRNP	SPG11
AIMP2	EIF2B1	GJA1	MAG	PROC	SUCLG1
ALDH3A2	EIF2B2	GJB1	MAN2B1	PROS1	SUMF1
APLP1	EIF2B3	GJC2	MANBA	PROZ	SUOX
APLP2	EIF2B4	GLA	MAPT	PSAP	SURF1
APOE	EIF2B5	GLB1	MARS2	PSEN1	TARS2
APP	ENG	GRN	MCOLN1	PSEN2	TGFBR2
ARSA	EPRS	GSN	MLC1	PSENNEN	TMEM106B
ASPA	ERCC1	GTF2E2	MMACHC	PYCR2	TREM2
AUH	ERCC2	GTF2H5	MPLKIP	RARS	TREX1
CA2	ERCC3	HARS2	MPV17	RARS2	TTR
CBS	ERCC4	HBB	MRM2	REEP1	TUBB4A
CLCN2	ERCC5	HEPACAM	MRPL38	RNASEH2A	TYMP
CNTNAP1	ERCC6	HEXA	MTCL1	RNASEH2B	TYROBP
COA8	ERCC8	HEXB	MTHFD1	RNASEH2C	VARS2
COL3A1	F2	HIKESHI	MTHFR	RNASET2	VHL
COL4A1	F5	HMGCL	NF1	RNF113A	VPS11
COL4A2	FA2H	HSPD1	NFU1	RPIA	WARS2
COLGALT1	FAM126A	HTRA1	NKX6-2	RRM2B	XPA
CSF1R	FAM126A	IARS2	NOTCH3	SAMHD1	XPC
CST3	FARS2	IBA57	NUBPL	SARS2	
CTC1	FBN1	IDUA	PEX1	SCP2	