Supplementary Table 2. Other variants identified in the exome sequencing data in the proband.

						Max. MAF gnomAD (v4.1)		All-of-Us							ACMG Criteria/ Classification	In linked interval?	
Gene	Transcript	cDNA (c.)	Protein Change (p.)	Zyg	Read Count (Variant:WT)	AF	AF Population	MAF	SIFT	PolyPhen-2	FATHMM	Mutation Taster	REVEL	CADD			Exclusion criteria
BCS1L	NM_001371449.1	c.670C>T	p.(Arg224Cys)	Het	520 (226:294)	3.1e-5	European (non-Finnish)	8.0e-6	0.00[1]	1.0[1]	-2.61[1]	0.99[1]	0.778[1]	32[1]	PM2_P, PP3_M/ VUS	No	Not in linked interval; Gene is associated with recessive disorders
DIS3L2	NM_152383.5	c.2074A>C	p.(Asn692His)	Het	119 (63:56)	2.2e-5	European (non-Finnish)	Not present	0.03[1]	0.994[1]	0.87[3]	0.33[3]	0.433[2]	28.6[1]	PM2_P/VUS	No	Not in linked interval; Gene is associated with a recessive disorder
GLYR1	NM_032569.4	c.1141C>T	p.(Arg381Cys)	Het	67 (29:38)	3.4e-6	European (non-Finnish)	4.0e-6	0.00[1]	1.0[1]	-0.24[2]	0.80[1]	0.67[1]	32.0[1]	PM2_P, PP3_P/ VUS	No	Not in linked interval; IMPC mouse with normal hearing
ITIH2	NM_002216.3	c.1223T>C	p.(Leu408Ser)	Het	58 (29:29)	2.2e-5	East Asian	4.0e-6	0.03[1]	1.0[1]	-1.49[2]	0.39[3]	0.71[1]	25.4[1]	PM2_P, PP3_P/ VUS	No	Not in linked interval
PLCB2	NM_004573.3	c.431G>T	p.(Arg144Leu)	Het	201 (115:86)	1.7e-4	Admixed American	2.0e-6	0.02[1]	0.954[1]	0.93[3]	0.59[3]	0.30[2]	26.8[1]	PM2_P/VUS	No	Not in linked interval
SLC10A2	NM_000452.3	c.724_725insC	p.(Phe242Serfs*5)	Het	44 (19:25)	0	--	0	--	--	--	--	--	--	PM2_P/VUS	No	Not in linked interval; Gene is associated with a recessive disorder
SMARCA4	NM_001387283.1	c.3355C>T	p.(Arg1119Cys)	Hom	725 (725:0)	8.5e-7	European (non-Finnish)	0	0.00[1]	1.0[1]	-1.07[2]	0.99[1]	0.89[1]	31[1]	PM2_P, PP3_M/ VUS	No	Not in linked interval; Gene is associated with a dominant disorder, phenotype mismatch
STON1	NM_006873.4	c.2167G>A	p.(Gly723Arg)	Het	51 (18:33)	0	--	-0	0.01[1]	1.0[1]	2.94[3]	0.19[3]	0.30[2]	31[1]	PM2_P/VUS	No	Not in linked interval;
*SUPV3L1**	NM_003171.5	c.1925G>A	p.(Ser642Asn)	Het	48 (29:19)	5.0e-5	Admixed American	2.2e-5	0.0[1]	1.0[1]	1.03[3]	0.93[1]	0.47[2]	35[1]	PM2_P	No	Not in linked interval;
TRPV2	NM_016113.5	c.1837C>T	p.(Gln613*)	Het	192 (106:86)	1.7e-5	Admixed American	1.2e-5	--	--	--	--	--	37[1]	PM2_P	No	Not in linked interval; Gene associated with perception to pain caused by heat
UBXN2B	NM_001077619.2	c.274G>A	p.(Ala92Thr)	Het	43 (18:25)	1.7e-5	Admixed American	6.0e-6	0.00[1]	1.0[1]	0.22[3]	0.54[3]	0.55[1]	27.6[1]	PM2_P/VUS	No	Not in linked interval; IMPC mouse with normal hearing
USP3	NM_006537.4	c.11C>G	p.(Pro4Arg)	Het	167 (94:73)	0	--	0	0.00[1]	0.45[3]	-1.81[1]	0.26[3]	0.63[1]	23.8[2]	PM2_P, PP3_P/ VUS	No	Not in linked interval; Unknown function

Pathogenicity is represented as [1]deleterious, [2]neutral, or [3]benign prediction, whereas "--" represents variant not scored.

Abbreviations: IMPC, International Mouse Phenotyping Consortium; VUS, variant of uncertain significance; Zyg, zygosity

*Predicted to cause aberrant splicing