Supplementary Table 1. *In silico* analysis of all detected genetic variants

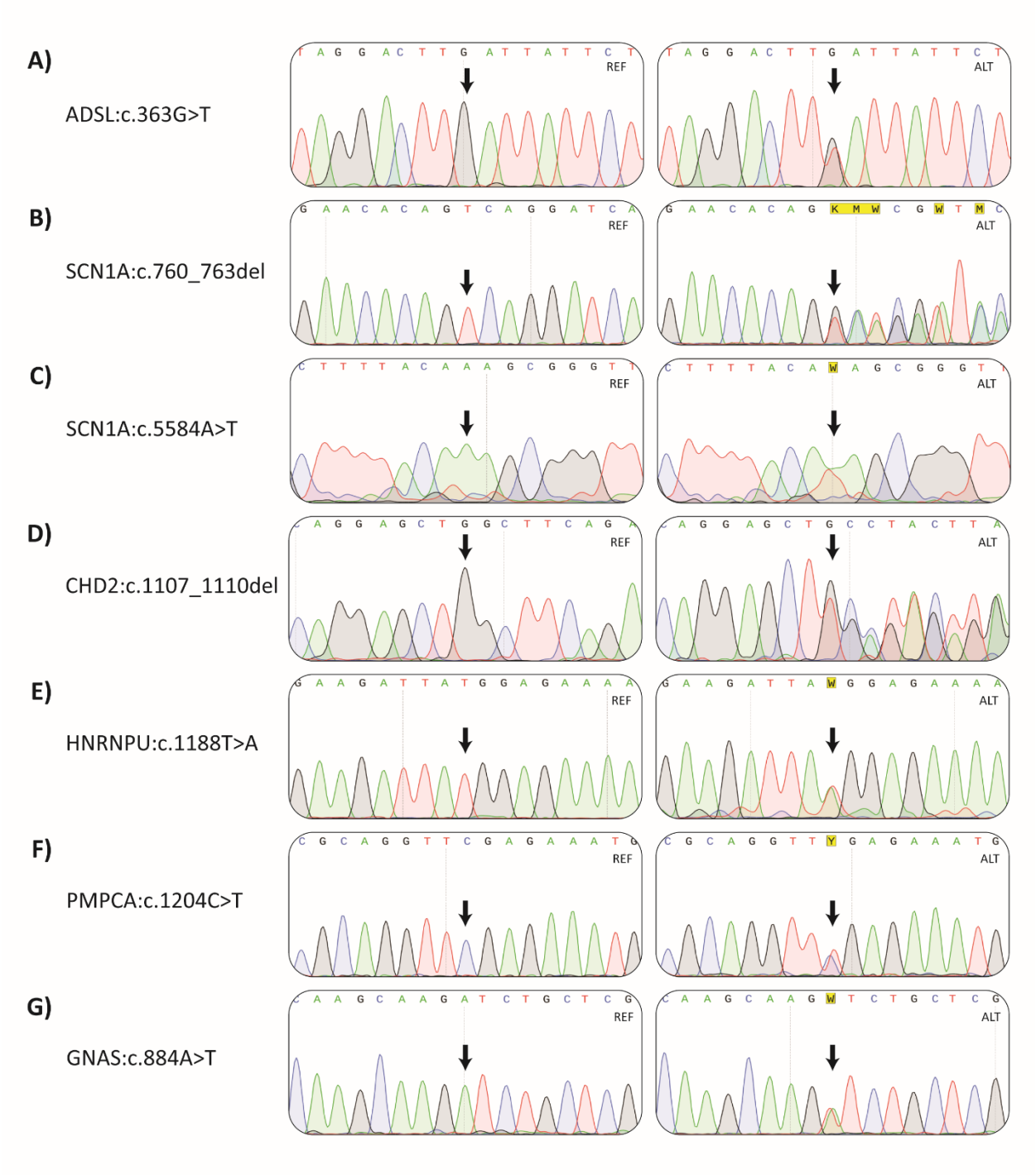
Gene	Variant	REVEL	BayesDel addAF	BayesDel noAF	MetaRNN	PROVEAN	CADD	SIFT	PolyPhen-2	MutPred2
ALDH7A1	c.1566-1G>T	N/A	Pathogenic Strong 0.5342	Pathogenic Strong 0.66	N/A	N/A	Pathogenic Strong 33	N/A	N/A	N/A
	c.328C>T (p.Arg110Ter)	N/A	Pathogenic Strong 0.4874	Pathogenic Strong 0.66	N/A	N/A	Pathogenic Strong 37	N/A	N/A	N/A
TSEN54	c.919G>T (p.Ala307Ser)	Benign Moderate 0.342	Benign Moderate -0.1606	Uncertain -0.0108	Benign Strong 0.04387	Benign Moderate -0.76	Pathogenic Supporting 27.5	Uncertain 0.024	Probably Damaging 0.997	0.159
KCNQ2	c.997C>T (p.Arg333Trp)	Pathogenic Moderate 0.937	Pathogenic Strong 0.5294	Pathogenic Strong 0.5227	Pathogenic Strong 0.9824	Pathogenic Moderate -7.16	Pathogenic Supporting 28.2	Pathogenic Supporting 0.001	Probably Damaging 1.000	0.766
PNPO	c.674G>A (p.Arg225His)	Pathogenic Strong 0.981	Pathogenic Moderate 0.3984	Pathogenic Moderate 0.5161	Pathogenic Strong 0.9865	N/A	Pathogenic Moderate 32	N/A	Probably Damaging 1.000	0.872
ADSL	c.340T>C (p.Tyr114His)	Pathogenic Strong 0.97	Pathogenic Strong 0.4357	Pathogenic Strong 0.5797	Pathogenic Moderate 0.9297	Pathogenic Supporting -4.53	Pathogenic Supporting 27.7	Pathogenic Supporting 0	Probably Damaging 1.000	0.741
	c.363G>T (p.Leu121Phe)	Pathogenic Moderate 0.857	Pathogenic Strong 0.4294	Pathogenic Moderate 0.3789	Pathogenic Moderate 0.8753	Uncertain -3.89	Uncertain 24.8999	Pathogenic Supporting 0.001	Probably Damaging 0.999	0.656
SCN1A	c.760_763del (p.Thr254Cysfs Ter4)	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
SCN1A	c.5584A>T (p.Lys1862Ter)	N/A	Pathogenic Very Strong 0.625	Pathogenic Strong 0.66	N/A	N/A	Pathogenic Strong 40	N/A	N/A	N/A
SCN1A	c.5308T>C (p.Tyr1770His)	Pathogenic Strong 0.977	Pathogenic Strong 0.5641	Pathogenic Strong 0.5725	Pathogenic Strong 0.9414	Pathogenic Supporting -4.6	Pathogenic Supporting 27.8999	Pathogenic Supporting 0	Probably Damaging 1.000	0.913
SCN1A	c.3067dup (p.Ile1023Asnfs Ter7)	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
SCN2A	c.424A>G (p.Asn142Asp)	Pathogenic Moderate 0.926	Pathogenic Moderate 0.3009	Pathogenic Supporting 0.1945	Pathogenic Moderate 0.8971	Pathogenic Supporting -4.49	Pathogenic Supporting 26.5	Pathogenic Supporting 0	Probably Damaging 0.998	0.930

Gene	Variant	REVEL	BayesDel addAF	BayesDel noAF	MetaRNN	PROVEAN	CADD	SIFT	PolyPhen-2	MutPred2
MMAA	c.593_596delCTGA (p.Thr198SerfsTer6)	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
GRIN1	c.797A>T (p.Tyr266Phe)	Uncertain 0.649	Pathogenic Supporting 0.2077	Uncertain 0.06053	Pathogenic Supporting 0.7677	Uncertain -2.52	Uncertain 23.5	Benign Moderate 0.259	Possibly Damaging 0.462	0.641
CACNA1E	c.1054G>A (p.Gly352Arg)	Pathogenic Moderate 0.913	Pathogenic Strong 0.4788	Pathogenic Moderate 0.4499	Pathogenic Moderate 0.9086	Pathogenic Supporting -6.09	Pathogenic Moderate 32	Uncertain 0.006	Probably Damaging 1.000	0.907
CACNA1A	c.5440G>A (p.Val1814Ile)	Pathogenic Moderate 0.808	Pathogenic Moderate 0.2608	Pathogenic Supporting 0.1369	Pathogenic Supporting 0.8064	Benign Moderate -0.99	Uncertain 24.2999	Benign Supporting 0.075	Probably Damaging 1.000	0.739
CHD2	c.1107_1110delGGCT (p.Ala370GlnfsTer3)	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
AMT	c.452_466del (p.Lys151_Leu155del)	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
SLC13A5	c.1207_1217dup (p.Pro407ArgfsTer12)	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A	N/A
DCX	c.305G>C (p.Arg102Pro)	Pathogenic Strong 0.978	Pathogenic Very Strong 0.6954	Pathogenic Strong 0.7611	Pathogenic Strong 0.9394	Pathogenic Moderate -7	Pathogenic Supporting 26.2	Pathogenic Supporting 0.001	Probably Damaging 1.000	0.971
CSNK2B	c.35G>A (p.Trp12Ter)	N/A	Pathogenic Strong 0.618	Pathogenic Strong 0.6499	N/A	N/A	Pathogenic Strong 41	N/A	N/A	N/A
KCNQ2	c.602G>A (p.Arg201His)	Pathogenic Strong 0.968	Pathogenic Strong 0.5786	Pathogenic Strong 0.5934	Pathogenic Strong 0.9867	Pathogenic Supporting -4.43	Pathogenic Supporting2 8.7999	Pathogenic Supporting 0	Probably Damaging 1.000	0.807
SCN1A	c.1625G>A (p.Arg542Gln)	Benign Moderate 0.307	Benign Moderate -0.2344	Benign Supporting -0.115	Benign Strong 0.009378	Uncertain -2.92	Benign Moderate 20.1	Benign Supporting 0.048	Benign 0.416	0.222
SCN1A	c.1239C>A (p.Tyr413Ter)	N/A	Pathogenic Very Strong 0.625	Pathogenic Strong 0.66	N/A	N/A	Pathogenic Strong 36	N/A	N/A	N/A

Gene	Variant	REVEL	BayesDel addAF	BayesDel noAF	MetaRNN	PROVEAN	CADD	SIFT	PolyPhen-2	MutPred2
HNRNPU	c.1188T>A (p.Tyr396Ter)	N/A	Pathogenic Strong 0.618	Pathogenic Strong 0.6499	N/A	N/A	Pathogenic Strong 36	N/A	N/A	N/A
RHOBTB 2	c.1532G>A (p.Arg511Gln)	Uncertain 0.474	Pathogenic Supporting 0.1833	Uncertain 0.02552	Pathogenic Supporting 0.7836	Benign Supporting -2.05	Pathogenic Supporting 28.1	Benign Supporting 0.094	Probably Damaging 1.000	0.743
PMPCA	c.667C>T (p.Arg223Cys)	Benign Supporting 0.352	Uncertain 0.1455	Uncertain 0.05404	Pathogenic Supporting 0.7814	Pathogenic Supporting -5.28	Pathogenic Moderate 32	Pathogenic Supporting 0	Probably Damaging 1.000	0.899
	c.1204C>T (p.Arg402Ter)	N/A	Pathogenic Strong 0.5599	Pathogenic Strong 0.5665	N/A	N/A	Pathogenic Strong 44	N/A	N/A	N/A
HSD17B1 0	c.680T>G (p.Leu227Arg)	Pathogenic Supportin 0.717	Pathogenic Supporting 0.2147	Uncertain 0.07061	Pathogenic Strong 0.9519	Pathogenic Supporting -5.51	Uncertain 25.1	Pathogenic Supporting 0	Possibly Damaging 0.824	0.860
ASH1L	c.6440C>T (p.Ala2147Val)	Benign Supporting 0.455	Uncertain 0.1137	Benign Supporting -0.0744	Pathogenic Supporting0. 7662	Benign Moderate -0.99	Pathogenic Supporting 28.5	Uncertain 0.003	Possibly Damaging 0.939	0.766
GNAS	c.884A>T (p.Asp295Val)	Pathogenic Strong 0.99	Pathogenic Strong 0.5843	Pathogenic Strong 0.6015	Pathogenic Strong 0.9902	Pathogenic Moderate -8.75	Pathogenic Moderate 32	Pathogenic Supporting 0	Probably Damaging 1.000	0.916

Cases not marked with color were analyzed using CES and cases colored in gray were analyzed using WES.

Abbreviation: N/A – nonavailable



Supplementary Figure 1. Electropherograms of new variants in *ADSL*, *SCN1A*, *CHD2*, *HNRNPU*, *PMPCA* and *GNAS* genes.