

**Supplementary Table 1. Polymorphic SNPs genotyped at Sequenom.**

Assay_Name	Gene
rs1227070	<i>CDH23</i>
rs1867998	<i>CDH23</i>
rs4999379	<i>CDH23</i>
rs723274	<i>CDH23</i>
rs1055518	<i>CDH23</i>
rs4746085	<i>CDH23</i>
rs7910896	<i>CDH23</i>
rs10942605	<i>GPR98</i>
rs1700510	<i>GPR98</i>
rs2010355	<i>GPR98</i>
rs2438351	<i>GPR98</i>
rs3098356	<i>GPR98</i>
rs4944143	<i>MYO7A</i>
rs6592711	<i>MYO7A</i>
rs12793189	<i>MYO7A</i>
rs2156568	<i>MYO7A</i>
rs10825347	<i>PCDH15</i>
rs1930164	<i>PCDH15</i>
rs2583026	<i>PCDH15</i>
rs4570494	<i>PCDH15</i>
rs6481065	<i>PCDH15</i>
rs7095441	<i>PCDH15</i>
rs10763126	<i>PCDH15</i>
rs10825195	<i>PCDH15</i>
rs17728345	<i>PCDH15</i>
rs1937392	<i>PCDH15</i>
rs7095317	<i>PCDH15</i>
rs9416371	<i>PCDH15</i>
rs9828914	<i>SLC4A7</i>
rs1494420	<i>SLC4A7</i>
rs2072227	<i>USH1C</i>
rs2240488	<i>USH1C</i>
rs2883581	<i>USH1C</i>
rs12033676	<i>USH2A</i>
rs2168924	<i>USH2A</i>
rs2168924	<i>USH2A</i>
rs12635944	<i>USH3A</i>
rs6801898	<i>USH3A</i>
rs718841	<i>WHRN</i>
rs942520	<i>WHRN</i>

rs10759697	<i>WHRN</i>
rs10982231	<i>WHRN</i>
rs4979379	<i>WHRN</i>
rs7034891	<i>WHRN</i>
rs7036125	<i>WHRN</i>

**Supplementary Table 2. Novel UV2-UV4 and pathogenic variants.**

Gene name	DNA change	Protein change	MAF (%) in controls <sup>a</sup>	Ortholog Conservation <sup>d</sup>	1000 Genomes <sup>f</sup>	Pathogenicity
<i>CDH23</i>	c.2177-2A>G	p.?	0 [0/96 CEPH]	NA	No	Pathogenic
<i>CDH23</i>	c.6254_6254-3delCAGGinsT	p.?	0 [0/96 CEPH]	NA	No	Pathogenic
<i>CDH23</i>	c.6712+1G>A	p.?	0	NA	No	Pathogenic
<i>CDH23</i>	c.7305dup	p.Leu2436ThrfsX3	0 [0/96 CEPH]	NA	No	Pathogenic
<i>CDH23</i>	c.7362G>A*	p.Thr2454Thr	0	NA	No	UV3
<i>CDH23</i>	c.9122T>C	p.Leu3041Pro	0	16 / 17 (94.12%)	No	UV4
<i>MYO7A</i>	c.1138G>A	p.Glu380Lys	0 [0/93 CEPH]	21 / 23 (91.30%)	No	UV2
<i>MYO7A</i>	c.1258A>T	p.Lys420X	0 [0/96 CEPHs]	NA	No	Pathogenic
<i>MYO7A</i>	c.1798-3C>G	p.?	0 [0/96 CEPH]	NA	No	UV2
<i>MYO7A</i>	c.223G>C	p.Asp75His	0	24 / 24 (100%)	No	UV4
<i>MYO7A</i>	c.3108+1G>A	p.?	0	NA	No	Pathogenic
<i>MYO7A</i>	c.338_348dup	p.Glu117SerfsX33	0 [0/96 CEPH]	NA	No	Pathogenic
<i>MYO7A</i>	c.4131dup	p.Gly1378TrpfsX6	0 [0/96 CEPH]	NA	No	Pathogenic
<i>MYO7A</i>	c.4293G>A	p.Trp1431X	0	NA	No	Pathogenic
<i>MYO7A</i>	c.4838delA	p.Asp1613ValfsX32	0 [0/96 CEPH]	NA	No	Pathogenic
<i>MYO7A</i>	c.-48A>G (IVS1-2A>G)	p.?	0	NA	No	Pathogenic
<i>MYO7A</i>	c.5824G>T	p.Gly1942X	0	NA	No	Pathogenic
<i>MYO7A</i>	c.6377delC	p.Pro2126Leuf sX5	0 [0/96 CEPH]	NA	No	Pathogenic
<i>MYO7A</i>	c.6577C>T	p.Leu2193Phe	0	23 / 23 (100.00%)	No	UV4
<i>MYO7A</i>	c.722G>C	p.Arg241Pro	0	23 / 23 (100.00%)	No	UV4
<i>PCDH15</i>	c.2823delG	p.Gly942ValfsX22	0 [0/96 CEPH]	NA	No	Pathogenic
<i>PCDH15</i>	c.3501+2T>C	p.?	0	NA	No	Pathogenic
<i>PCDH15</i>	Large deletion: Exon 9-18	p.?	NA	NA	No	Pathogenic
<i>PCDH15</i>	Large deletion: Exon 10	p.?	NA	NA	No	Pathogenic
<i>USH1C</i>	c.1016G>A	p.Arg339Gln <sup>b</sup>	0	17 / 21 (80.95%)	No	UV2
<i>USH1C</i>	c.2227-1G>T <sup>c</sup>	p.?	0	NA	No	Pathogenic

<i>USH1C</i>	c.446_448delA GG	p.Glu149del	NA	NA	No	Pathogenic
<i>USH2A</i>	c.1019A>T	p.His340Leu	0	17 / 17 (100.00%)	No	UV3
<i>USH2A</i>	c.6049G>T*	p.Gly2017Cys	0	15/17 (88.24%)	No	UV2
<i>USH2A</i>	c.10724G>A	p.Cys3575Tyr	0	17 / 17 (100.00%)	No	UV4
<i>USH2A</i>	c.11047+1G>A	p.?	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.11065C>T	p.Arg3689X	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.11390-1G>C	p.?	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.11872_11873 delAC	p.Gln3959Asn fsX53	0	NA	No	Pathogenic
<i>USH2A</i>	c.12295-3T>A	p.?	0 [0/96 CEPH]	NA	No	UV2
<i>USH2A</i>	c.12457G>A	p.Ala4153Thr	0.12	13 / 17 (76.47%) <sup>e</sup>	No	UV4
<i>USH2A</i>	c.13130C>A	p.Ser4377X	0	NA	No	Pathogenic
<i>USH2A</i>	c.13621C>T	p.Gln4541X	0	NA	No	Pathogenic
<i>USH2A</i>	c.14139G>A	p.Trp4713X	0	NA	No	Pathogenic
<i>USH2A</i>	c.14285A>G	p.Asn4762Ser	0	17 / 17 (100.00%)	No	UV3
<i>USH2A</i>	c.14403C>G	p.Tyr4801X	0	NA	No	Pathogenic
<i>USH2A</i>	c.14446A>T	p.Lys4816X	0	NA	No	Pathogenic
<i>USH2A</i>	c.14911C>T	p.Arg4971X	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.1859G>T	p.Cys620Phe	0	17 / 17 (100.00%)	No	Pathogenic
<i>USH2A</i>	c.2236C>G	p.Pro746Ala	0	17 / 17 (100.00%)	No	UV4
<i>USH2A</i>	c.2610C>A	p.Cys870X	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.2942_2943ins T	p.Cys982Leufs X2	0	NA	No	Pathogenic
<i>USH2A</i>	c.2994_3007del	p.Cys999Leufs X9	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.3407G>A	p.Ser1136Asn	0	17 / 17 (100.00%)	No	UV4
<i>USH2A</i>	c.3518C>A	p.Ser1173X	0	NA	No	Pathogenic
<i>USH2A</i>	c.4133T>C	p.Leu1378Pro	0	15 / 17 (88.24%)	No	UV4
<i>USH2A</i>	c.4354dup	p.Cys1452Leu fsX25	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.4732C>T	p.Arg1578Cys	0	15 / 17 (88.24%)	No	UV4
<i>USH2A</i>	c.4761delG	p.Ser1588Hisf sX5	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.4821G>A	p.Trp1607X	0.12	NA	No	Pathogenic

<i>USH2A</i>	c.5252 G>T	p.Gly1751Val	0	14 / 17 (82.35%)	No	UV2
<i>USH2A</i>	c.5329 C>T	p.Arg1777Trp	0	6 / 17 (35.29%)	No	UV3
<i>USH2A</i>	c.5603T>G	p.Phe1868Cys	0 [0/96 CEPH]	11 / 17 (64.71%)	No	UV2
<i>USH2A</i>	c.5614_5620del GCTGTCTG	p.Ala1872Leuf sX58	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.5834_5835ins TC	p.Arg1946Leu fsX22	0	NA	No	Pathogenic
<i>USH2A</i>	c.5898_5899del AA	p.Asn1967Trp fsX5	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.651+1G>A	p.?	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.6854A>G	p.Asn2285Ser	0 [0/96 CEPH]	16 / 17 (94.12%)	No	UV2
<i>USH2A</i>	c.6928A>C	p.Thr2310Pro	0 [0/96 CEPH]	17 / 17 (100.00%)	Yes (1/1258)	UV2
<i>USH2A</i>	c.769G>A	p.Gly257Arg	0 [0/96 CEPH]	16 / 17 (94.12%)	No	UV2
<i>USH2A</i>	c.8231G>A	p.Trp2744X	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.8261delT	p.Ile2754Asnf sX15	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	c.842C>A	p.Thr281Lys	0 [0/93 CEPH]	15 / 17 (88.24%)	No	UV2
<i>USH2A</i>	c.9371+1G>C	p.?	0.12	NA	No	Pathogenic
<i>USH2A</i>	c.9459C>A	p.Cys3153X	NA	NA	No	Pathogenic
<i>USH2A</i>	c.9842G>T	p.Cys3281Phe	0 [0/96 CEPHs]	17 / 17 (100.00%)	No	UV2
<i>USH2A</i>	c.9912dup	p.Glu3305Arg fsX41	0	NA	No	Pathogenic
<i>USH2A</i>	Large deletion: Exon 47	p.?	0 [0/96 CEPH]	NA	No	Pathogenic
<i>USH2A</i>	Large deletion: Exon 50-55	p.?	0 [0/96 CEPH]	NA	No	Pathogenic
<i>GPR98</i>	c.10016G>A	p.Ser3339Asn	0	17 / 19 (89.47%)	No	UV3
<i>GPR98</i>	c.10736_37delC C	p.Ala3579Valf sX6	0 [0/96 CEPH]	NA	No	Pathogenic
<i>GPR98</i>	c.13433G>T*	p.Ser4478Ile	0	19 / 20 (95.00%)	No	UV3
<i>GPR98</i>	c.14404C>T	p.Arg4802X	0	NA	No	Pathogenic
<i>GPR98</i>	c.15144delC	p.Ser5048Argf sX29	0 [0/96 CEPH]	NA	No	Pathogenic
<i>GPR98</i>	c.2398C>T	p.Arg800X	0.11	NA	No	Pathogenic
<i>GPR98</i>	c.4123G>C	p.Asp1375His	0	14 / 20 (70.00%)	No	UV2
<i>GPR98</i>	c.6307G>T	p.Glu2103X	0	NA	No	Pathogenic
<i>GPR98</i>	c.6856C>T	p.Arg2286X	NA	NA	No	Pathogenic

<i>GPR98</i>	c.6962_63delT G	p.Val2321AlafsX4	NA	NA	No	Pathogenic
<i>GPR98</i>	c.9623+1G>A	p.?	0	NA	No	Pathogenic
<i>GPR98</i>	c.9974T>C	p.Ile3325Thr	0 [0/96 CEPHs]	17 / 19 (89.47%)	No	UV2
<i>GPR98</i>	Large deletion: Exon 83	p.?	0 [0/96 CEPHs]	NA	No	Pathogenic

### Legend to Supplementary Table 2.

\*Last nucleotide of an exon. Variant is predicted to significantly lower efficiency of adjacent splice site as predicted by *in silico* analysis (Human Splicing Finder, <http://www.umd.be/HSF/> and Splice Site Prediction by Neural Network, [www.fruitfly.org/seq\\_tools/splice.html](http://www.fruitfly.org/seq_tools/splice.html)).

NA-Not Applicable

<sup>a</sup> Minimum Allele Frequency in 876 control chromosomes unless stated otherwise

<sup>b</sup> Identified in NCUS proband with RP atypical for Usher syndrome

<sup>c</sup> Identified in NCUS proband who was diagnosed with sector RP and hearing loss.[22]

<sup>d</sup> Alignments were made using Usher Syndrome Missense Analysis tool (USMA). Full alignments are available via a link from LSDB for Usher syndrome. Unless stated otherwise, the mutated residue was not found in ortholog species

<sup>e</sup> Mutated residue was found in *Ciona intestinalis* and *Ciona savignyi* (family of sea squirts)

<sup>f</sup> Accessed on July 13th 2011 (<http://browser.1000genomes.org/index.htm>)

**Supplementary Table 3. Novel UV1 and Neutral variants.**

Gene name	DNA change	Protein change	MAF (%) in controls <sup>a</sup>	Pathogenicity
<i>CDH23</i>	c.10036G>C	p.Glu3346Gln	0.11	UV1
<i>CDH23</i>	c.129C>T <sup>+</sup>	p.Ser43Ser	NA	UV1
<i>CDH23</i>	c.1307G>A	p.Ser436Asn	0.34	Neutral
<i>CDH23</i>	c.1369C>T	p.Arg457Trp	0.23	UV1
<i>CDH23</i>	c.1595C>T	p.Thr532Met	0	UV1
<i>CDH23</i>	c.173A>G	p.Gln58Arg	NA	UV1
<i>CDH23</i>	c.1752+6G>A	p.?	NA	UV1
<i>CDH23</i>	c.1919C>T	p.Thr640Met	0	UV1
<i>CDH23</i>	c.198G>A	p.Val66Val	NA	UV1
<i>CDH23</i>	c.204C>T	p.Gly68Gly	NA	UV1
<i>CDH23</i>	c.2235C>T <sup>+</sup>	p.Ile745Ile	0	UV1
<i>CDH23</i>	c.2289+20G>C	p.?	NA	UV1
<i>CDH23</i>	c.2970C>T	p.Asp990Asp	0.69	Neutral
<i>CDH23</i>	c.3337G>C	p.Glu1113Gln	0	UV1
<i>CDH23</i>	c.3664G>A	p.Ala1222Thr	NA	UV1
<i>CDH23</i>	c.3801C>T	p.Thr1267Thr	0.11	UV1
<i>CDH23</i>	c.3845A>G	p.Asn1282Ser	0.93	Neutral
<i>CDH23</i>	c.3895G>A	p.Val1299Ile	0	UV1
<i>CDH23</i>	c.4231G>A	p.Glu1411Lys	0	UV1
<i>CDH23</i>	c.4287C>T	p.Pro1429Pro	0	UV1
<i>CDH23</i>	c.442G>A [ENST00000224721]	p.Gly148Arg [ENST00000224721]	NA	UV1
<i>CDH23</i>	c.444+3T>C [ENST00000224721]	p.?	NA	UV1
<i>CDH23</i>	c.4786C>T	p.Arg1596Cys	0	UV1
<i>CDH23</i>	c.4846-19G>A	p.?	NA	UV1
<i>CDH23</i>	c.4890C>T <sup>+</sup>	p.Asn1630Asn	0	UV1
<i>CDH23</i>	c.4947G>A	p.Thr1649Thr	0	UV1
<i>CDH23</i>	c.5067+15G>A	p.?	NA	UV1
<i>CDH23</i>	c.551G>A	p.Arg184His	0	UV1
<i>CDH23</i>	c.5541C>T	p.Asn1847Asn	0.12	UV1
<i>CDH23</i>	c.5660C>T	p.Thr1887Ile	0.34	Neutral
<i>CDH23</i>	c.6197G>A	p.Arg2066Gln	0.36	Neutral
<i>CDH23</i>	c.6648C>T <sup>+</sup>	p.Ala2216Ala	0	UV1
<i>CDH23</i>	c.67+19G>A	p.?	NA	UV1
<i>CDH23</i>	c.7722C>T	p.Tyr2574Tyr	0.34	Neutral

<i>CDH23</i>	c.8722G>A*	p.Gly2908Arg	0	UV1
<i>CDH23</i>	c.9014C>T	Ala3005Val	0	UV1
<i>CDH23</i>	c.9015G>A	Ala3005Ala	0.11	UV1
<i>CDH23</i>	c.9238G>A	Ala3080Thr	0.23	UV1
<i>CDH23</i>	c.9319+11G>A	p.?	NA	UV1
<i>CDH23</i>	c.9973C>G	Arg3325Gly	0	UV1
<i>MYO7A</i>	c.1081-10G>C	p.?	NA	UV1
<i>MYO7A</i>	c.1407G>A	Val469Val	NA	UV1
<i>MYO7A</i>	c.133-7C>T	p.?	NA	UV1
<i>MYO7A</i>	c.1543A>C	Lys515Gln	0.12	UV1
<i>MYO7A</i>	c.1554+8G>A	p.?	NA	UV1
<i>MYO7A</i>	c.1817G>A	p.Arg606His	0	UV1
<i>MYO7A</i>	c.1969C>T	p.Arg657Trp	0	UV1
<i>MYO7A</i>	c.2057G>A	p.Arg686His	0.11	UV1
<i>MYO7A</i>	c.2120G>A	p.Arg707His	0	UV1
<i>MYO7A</i>	c.2617C>T	p.Arg873Trp	0.11	UV1
<i>MYO7A</i>	c.2886G>C	p.Gln962His	0	UV1
<i>MYO7A</i>	c.3453G>A	p.Leu1151Leu	NA	UV1
<i>MYO7A</i>	c.359 G>A	p.Arg120His	0	UV1
<i>MYO7A</i>	c.4023C>T	p.Pro1341Pro	0	UV1
<i>MYO7A</i>	c.4074C>T	p.Ser1358Ser	0.47	Neutral
<i>MYO7A</i>	c.4161C>A	p.Asp1387Glu	0	UV1
<i>MYO7A</i>	c.4461C>T	p.Asn1487Asn	0.23	UV1
<i>MYO7A</i>	c.4505 A>G	p.Asp1502Gly	0	UV1
<i>MYO7A</i>	c.4619 C>T	p.Ala1540Val	0	UV1
<i>MYO7A</i>	c.4845C>A	p.Pro1615Pro	NA	UV1
<i>MYO7A</i>	c.4950C>T	p.Asn1650Asn	0	UV1
<i>MYO7A</i>	c.495G>A	p.Thr165Thr	0.12	UV1
<i>MYO7A</i>	c.5122C>A	p.Arg1708Ser	0	UV1
<i>MYO7A</i>	c.5216G>A	p.Arg1739Gln	NA	UV1
<i>MYO7A</i>	c.5245C>T	p.Arg1749Trp	0.12	UV1
<i>MYO7A</i>	c.54G>C	p.Gln18His	0.12	UV1
<i>MYO7A</i>	c.5619G>A	p.Arg1873Arg	NA	UV1
<i>MYO7A</i>	c.569T>G	p.Leu190Trp	0 [0/96 CEPH]	UV1
<i>MYO7A</i>	c.5866G>A	p.Val1956Ile	0.24	UV1
<i>MYO7A</i>	c.6051+16C>G	p.?	NA	UV1
<i>MYO7A</i>	c.6509C>T	p.Thr2170Ile	NA	UV1
<i>MYO7A</i>	c.6626G>A	p.Arg2209Gln	0	UV1
<i>MYO7A</i>	c.6640G>A	p.Gly2214Ser	0.11	UV1
<i>MYO7A</i>	c.973A>T	p.Ile325Phe	0	UV1



<i>MYO7A</i>	c.974T>G	p.Ile325Ser	0	UV1
<i>MYO7A</i>	c.5598C>A	p.Leu1866Leu	0.23	UV1
<i>PCDH15</i>	c.2625G>C	p.Ser875Ser	NA	UV1
<i>PCDH15</i>	c.319-20A>T	p.?	NA	UV1
<i>PCDH15</i>	c.3502-8C>T	p.?	NA	UV1
<i>PCDH15</i>	c.3795A>G	p.Glu1265Asp	0	UV1
<i>PCDH15</i>	c.3983+12T>C	p.?	NA	UV1
<i>PCDH15</i>	c.4974A>C	p.Ser1658Ser	0.11	UV1
<i>PCDH15</i>	c.5247_5249de ITCC	p.Pro1752del	0.57	Neutral
<i>PCDH15</i>	c.5263C>T	p.Pro1755Ser	NA	UV1
<i>PCDH15</i>	c.5358_5359in sCCTCTT	p.Ile1786_Pro17 87insProLeu	NA	UV1
<i>PCDH15</i>	c.5359C>T	p.Pro1787Ser	NA	UV1
<i>PCDH15</i>	c.5398G>A	p.Val1800Ile	NA	UV1
<i>PCDH15</i>	c.5550C>A	p.Thr1850Thr	NA	UV1
<i>PCDH15</i>	c.5707A>G	p.Ile1903Val	NA	UV1
<i>USH1C</i>	c.1086-13G>T	p.?	NA	UV1
<i>USH1C</i>	c.1430G>A	p.Arg477Gln	0	UV1
<i>USH1C</i>	c.2340C>T	p.Val780Val	NA	UV1
<i>USH1C</i>	c.324T>C	p.Phe108Phe	0	UV1
<i>USH1G</i>	c.1152C>T	p.Asp384Asp	0	UV1
<i>USH1G</i>	c.388A>G	p.Lys130Glu	0	UV1
<i>USH1G</i>	c.501C>G	p.Arg167Arg	0.12	UV1
<i>USH1G</i>	c.566G>A	p.Arg189Gln	0.12	UV1
<i>USH1G</i>	c.678C>A	p.Gly226Gly	0	UV1
<i>USH1G</i>	c.705G>A	p.Glu235Glu	0.12	UV1
<i>USH1G</i>	c.83C>T	p.Pro28Leu	NA	UV1
<i>GPR98</i>	c.10260C>T <sup>+</sup>	p.Phe3420Phe	0	UV1
<i>GPR98</i>	c.10796+9A> G	p.?	NA	UV1
<i>GPR98</i>	c.10873C>G	p.Leu3625Val	NA	UV1
<i>GPR98</i>	c.10927A>G	p.Thr3643Ala	0.11	UV1
<i>GPR98</i>	c.11599G>A	p.Glu3867Lys	NA	UV1
<i>GPR98</i>	c.12121- 16A>G	p.?	NA	UV1
<i>GPR98</i>	c.13232-7A>G	p.?	NA	UV1
<i>GPR98</i>	c.13358A>G	p.His4453Arg	0	UV1
<i>GPR98</i>	c.13919G>A	p.Gly4640Glu	0	UV1
<i>GPR98</i>	c.14309G>A	p.Arg4770His	0.92	Neutral
<i>GPR98</i>	c.14856C>T	p.His4952His	0	UV1
<i>GPR98</i>	c.15105C>T <sup>+</sup>	p.Ser5035Ser	NA	UV1
<i>GPR98</i>	c.15301G>A <sup>+</sup>	p.Gly5101Arg	0.36	Neutral
<i>GPR98</i>	c.15343C>T	p.Leu5115Leu	0	UV1

<i>GPR98</i>	c.16164A>G	p.Arg5388Arg	1.54	Neutral
<i>GPR98</i>	c.16312A>G	p.Thr5438Ala	0.48	Neutral
<i>GPR98</i>	c.18026G>A	p.Arg6009Gln	0	UV1
<i>GPR98</i>	c.18040T>C	p.Phe6014Leu	0	UV1
<i>GPR98</i>	c.18273A>G	p.Ala6091Ala	0.23	UV1
<i>GPR98</i>	c.18311-8_12delTTTT	p.?	NA	UV1
<i>GPR98</i>	c.1849G>A	p.Val617Met	NA	UV1
<i>GPR98</i>	c.18746T>G	p.Leu6249Arg	0	UV1
<i>GPR98</i>	c.18753C>T	p.Ala6251Ala	0	UV1
<i>GPR98</i>	c.18782T>C	p.Leu6261Ser	0	UV1
<i>GPR98</i>	c.18802+9G>A	p.?	NA	UV1
<i>GPR98</i>	c.18803-13A>G	p.?	NA	UV1
<i>GPR98</i>	c.2185A>G	p.Ile729Val	0	UV1
<i>GPR98</i>	c.2284C>T	p.Arg762Cys	0	UV1
<i>GPR98</i>	c.2516T>C	p.Val839Ala	0.12	UV1
<i>GPR98</i>	c.2553+11T>A	p.?	NA	UV1
<i>GPR98</i>	c.2735-10C>A	p.?	NA	UV1
<i>GPR98</i>	c.2834G>A	p.Gly945Glu	NA	UV1
<i>GPR98</i>	c.3022+8T>C	p.?	NA	UV1
<i>GPR98</i>	c.3255T>C	p.Asp1085Asp	0.12	UV1
<i>GPR98</i>	c.327C>T	p.Asp109Asp	0.24	UV1
<i>GPR98</i>	c.3289G>A*	p.Gly1097Ser	0.11	UV1
<i>GPR98</i>	c.3635-10C>A	p.?	NA	UV1
<i>GPR98</i>	c.3805T>C	p.Phe1269Leu	0	UV1
<i>GPR98</i>	c.380T>G	p.Leu127Arg	2.22% [2/90 CEPH]	UV1
<i>GPR98</i>	c.3975G>A	p.Thr1325Thr	0	UV1
<i>GPR98</i>	c.4214C>T <sup>+</sup>	p.Ser1405Ser	NA	UV1
<i>GPR98</i>	c.4260A>G	p.Glu1420Glu	0.84	Neutral
<i>GPR98</i>	c.5104C>T	p.Pro1702Ser	0	UV1
<i>GPR98</i>	c.5221T>C <sup>+</sup>	p.Leu1741Leu	0	UV1
<i>GPR98</i>	c.5525-7C>T	p.?		UV1
<i>GPR98</i>	c.5785G>T	p.Ala1929Ser	0.24	UV1
<i>GPR98</i>	c.6012G>T	p.Leu2004Phe	NA	UV1
<i>GPR98</i>	c.6289C>T	p.Arg2097Cys	1.19	Neutral
<i>GPR98</i>	c.6317C>T	p.Ala2106Val	0.69	Neutral
<i>GPR98</i>	c.6318G>A	p.Ala2106Ala	0.68	Neutral
<i>GPR98</i>	c.6608 T>C	p.Val2203Ala	0.34	Neutral
<i>GPR98</i>	c.6695A>G	p.Tyr2232Cys	NA	UV1

<i>GPR98</i>	c.6938C>T	p.Pro2313Leu	0	UV1
<i>GPR98</i>	c.6994A>T	p.Ile2332Phe	0	UV1
<i>GPR98</i>	c.7176C>T	p.Ser2392Ser	0.24	UV1
<i>GPR98</i>	c.7179C>T	p.Asp2393Asp	1.48	Neutral
<i>GPR98</i>	c.7229A>G	p.Tyr2410Cys	0.11	UV1
<i>GPR98</i>	c.7293C>T	p.Ala2431Ala	0	UV1
<i>GPR98</i>	c.7576 A>G	p.Ile2526Val	0	UV1
<i>GPR98</i>	c.7821G>A	p.Glu2607Glu	0.36	Neutral
<i>GPR98</i>	c.7874G>A	p.Arg2625His	NA	UV1
<i>GPR98</i>	c.9280G>A	p.Val3094Ile	NA	UV1
<i>GPR98</i>	c.9650C>T	p.Ala3217Val	0.8	Neutral
<i>GPR98</i>	c.9743G>A	p.Gly3248Asp	NA	UV1
<i>USH2A</i>	c.10062G>C	p.Val3354Val	0.11	UV1
<i>USH2A</i>	c.10510 C>G	p.Pro3504Ala	0.12	UV1
<i>USH2A</i>	c.11597C>T	p.Ala3866Val	0	UV1
<i>USH2A</i>	c.1174C>T	p.Pro392Ser	0	UV1
<i>USH2A</i>	c.11928G>A	p.Thr3976Thr	NA	UV1
<i>USH2A</i>	c.12505A>G	p.Thr4169Ala	0	UV1
<i>USH2A</i>	c.12823T>A	p.Ser4275Thr	0.11	UV1
<i>USH2A</i>	c.12910G>A	p.Glu4304Lys	0	UV1
<i>USH2A</i>	c.13631G>T	p.Gly4544Val	0	UV1
<i>USH2A</i>	c.1453A>G	p.Ile485Val	0	UV1
<i>USH2A</i>	c.1731C>T	p.Cys577Cys	0.24	UV1
<i>USH2A</i>	c.2844 C>G	p.Cys948Trp	0	UV1
<i>USH2A</i>	c.2844C>G	p.Cys948Trp	0	UV1
<i>USH2A</i>	c.3261 C>T	p.Ile1207Ile	0	UV1
<i>USH2A</i>	c.3945T>C	p.Asn1315Asn	1.12	Neutral
<i>USH2A</i>	c.4115C>A	p.Pro1372His	0.12	UV1
<i>USH2A</i>	c.4560C>T	p.Ile1520Ile	0.46	Neutral
<i>USH2A</i>	c.5142 T>C	p.Asn1714Asn	0	UV1
<i>USH2A</i>	c.550A>C	p.Thr184Pro	0 [0/96 CEPH]	UV1
<i>USH2A</i>	c.7130A>G	p.Asn2377Ser	0.81	Neutral
<i>USH2A</i>	c.7584C>T	p.Thr2528Thr	0.11	UV1
<i>USH2A</i>	c.785-17_- 14delAT	p.?	0	UV1
<i>USH2A</i>	c.8681+18A> G	p.?	4.35 [4/92 CEPH]	UV1
<i>USH2A</i>	c.9228C>A	p.Asp3076Glu	0	UV1
<i>USH3A</i>	c.126G>A	p.Thr42Thr	0	UV1
<i>USH3A</i>	c.6A>C	p.Pro2Pro	0.46	Neutral
<i>WHRN</i>	c.1166+18 G>A	p.?	NA	UV1
<i>WHRN</i>	c.1365T>C	p.Ser455Ser	NA	UV1

<i>WHRN</i>	c.1653 C>T <sup>†</sup>	p.Gly551Gly	NA	UV1
<i>WHRN</i>	c.2118A>G	p.Pro706Pro	NA	UV1
<i>WHRN</i>	c.2485 G>A	p.Ala829Thr	NA	UV1
<i>WHRN</i>	c.409 G>C	p.Glu137Gln	0 [0/134 ECCAC]	UV1
<i>WHRN</i>	c.755A>G	p.Gln252Arg	0 [0/98 ECCAC; 0/84 Pakistani]	UV1
<i>SLC4A7</i>	c.1740-3T>C	p.?	NA	UV1
<i>SLC4A7</i>	c.2314C>T	p.Pro772Ser	1.72	Neutral
<i>SLC4A7</i>	c.3624G>A	p.Val1208Val	0	UV1
<i>SLC4A7</i>	c.402-7C>T	p.?	NA	UV1
<i>SLC4A7</i>	c.459T>C	p.Tyr53Tyr	NA	UV1
<i>SLC4A7</i>	c.756A>T	p.Glu252Asp	0.12	UV1

### Legend to Supplementary Table 3.

\*Last nucleotide of an exon. CDH23:c.8722G>A, (p.Gly2908Arg), and GPR98:c.3289G>A (p.Gly1097Ser) had no significant effect on predicted use of splice sites.

<sup>†</sup>Novel synonymous changes in which there was no other contributing evidence to their pathogenicity ie. the patient had a single or no other pathogenic/UV3/UV4 mutations accounting for their disease in the same or another gene *and* they do not appear or have not been assessed in controls, were also analysed *in silico* (Human Splicing Finder, <http://www.umd.be/HSF/> and Splice Site Prediction by Neural Network, [www.fruitfly.org/seq\\_tools/splice.html](http://www.fruitfly.org/seq_tools/splice.html)). There was no significant effect on splicing except for the variant *WHRN*:c.1653C>T, (p.Gly551Gly); this increased splice score by >20% for creation of a novel splice site (Human Splicing Finder, <http://www.umd.be/HSF/>) - initial score of 65 for wild-type C increasing to 92 for variant T. However this variant showed no effect on splicing using Splice Site Prediction by Neural Network, [www.fruitfly.org/seq\\_tools/splice.html](http://www.fruitfly.org/seq_tools/splice.html)).

<sup>a</sup> Minimum Allele Frequency in 878 control chromosomes unless stated otherwise; NA-not assessed