Supplementary Table 1: Usher microarray nucleotide variants (page 1 of 3)

#	Exon	Nucleotide change	Amino acid change	Validated	Ref.
CDH23					
1	3	172C>T	Q58X	y	1
2	3	193delC	L65fs	y	1
3	4	336G>C	G112G/splice defect	y	2
4	4	336+1G>A	splice defect	y	1
5	5	371A>G	D124G	y	1
6	10	1087delG	V363fs	y	1
7	10	1112delT	I371fs	y	1
8	13	1355A>G	N452S	y	1
9	13	1439T>A	L480Q	n	1
10	14	1450G>C	A484P/splice defect	y	1
11	15	1745G>A	R582Q	y	1
12	20	2289+1G>A	splice defect	y	1
13	25	2968G>A	D990N	n	3
14	25	3105A>C	T1035T/splice defect	y	1
15	26	3178C>T	R1060W	y	1
16	29	3557G>A	G1186D	n	1
17	30	3617C>G	P1206R	t/n	1
18	30	3625A>G	T1209A	y	1
19	31	3841_3843del	M1281del	n	4
20	31	3842_3845dup	V1283fs	y	1
21	31	c.3880>T#	p.Q1294X#	n	3
22	31	4021G>A	D1341N	y	5
23	35	4488G>C	Q1496H/splice defect	n	4
24	36	4504C>T	R1502X	y	1
25	37	4756G>C	A1586P	n	1
26	37	4783G>A	E1595K	y	1
27	40	5237G>A	R1746Q	y	4
28	42	5536G>A	D1846N	n	1
29	42	5712G>A	T1904T/splice defect	n	2
30	45	6049+1G>A	splice defect	n	3
31	46	6050-9G>A	splice defect	y	1, 2
32	46	6133G>A	D2045N	n	3
33	46	6155delC	T2055fs	y	1
34	47	6307G>T	E2103X	y	1
35	47	6319C>T	R2107X	n	3
36	47	6442G>A	D2148N	n	1,5
37	47	6604G>A	D2202N	n	3
38	49	6933delT	T2313fs	n	2
39	49	6968delC	P2323fs	y	1
40	51	7362+5G>A	splice defect	n	4
41	52	7393C>T	R2465W	y	1
42	53	7549A>G	S2517G	y	1
43	54	7823G>A	R2608H	y	1
44	54	7872G>A	E2624E/splice defect	n	1
45	57	8230G>A	G2744S	n	1
46	58	8497C>G	R2833G	y	1
47	60	8849T>A	I2950N	n	3
48	60	8866C>T	R2956C	n	3
49	62	9175C>A	P3059T	n	3
50	66	9510+1G>A	splice defect	n	3
51	67	9524G>A	R3175H	n	1
52	67	9626_9627insC	I3210fs	y	1

#	Exon	Nucleotide change	Amino acid change	Validated	Ref.
MYO7A					
1	3	47T>A	L16X	n	6
2	3	52C>T	Q18X	y	7
3	3	73G>A	G25R	n	8
4	3	77C>A	A26E	n	9
5	3	93C>A	C31X	y	10
6	4	124-2A>G	splice defect	n	11
7	4	199G>A	V67M	n	9
8	4	223delG	D75fs	t/n	10
9	4	269G>C#	R90P#	n	9
10	5	318C>A	N106K	n	7
11	5	361delC	Q121fs	y	10
12	5	401T>A	I134N	y	9
13	5	448C>T	R150X	y	12
14	5	470+1G>A	splice defect	n	13
15	6	494C>T	T165M	y	14
16	7	613A>G	I205V	n	13
17	7	634C>T	R212C	y	10
18	7	635G>A	R212H	y	10
19	7	640G>A	G214R	y	13
20	7	652_657delGACATC	D218_I219del	n	12
21	7	700C>T	Q234X	y	12
22	7	721C>A	R241S	n	6
23	7	721C>T	R241C	n	9
24	7	722G>A	R241H	n	7
25	7	731G>C	R244P	n	11
26	8	736-3C>T	splice defect	n	7
27	8	805_807delAAG	K269del	n	9
28	9	905G>A	R302H	y	10
29	9	940G>T	E314X	y	10
30	9	999T>G	Y333X	n	10
31	10	1066_1067insATCTCTGC	A356fs	y	7
32	11	1132C>A	R378S	n	7
33	11	1157T>C	L386P	n	10
34	11	1190C>A	A397D	n	13
35	12	1258A>T	K420X	y	7
36	12	1325A>G	E442G	y	7
37	13	1348G>C	E450Q	n	10
38	13	1370C>T	A457V	n	9
39	13	1373A>T	N458I	y	15
40	13	1403_1404insGCA	R467_H468insQ	y	10
41	13	1508C>T	P503L	y	10
42	14	1555-8C>G	splice defect	n	9
43	14	1555-1G>C	splice defect	n	10
44	14	1556G>A	G519D	n	9
45	14	1563delC	D521fs	n	8,11
46	14	1595delA	H532fs	n	10
47	14	1623_1624insC	K542fs	y	9
48	15	1797G>A	M599I	n	16
49	16	1798-14_1798-11 delGTCCinsCCAG	splice defect	n	10
50	16	1798delG	G600fs	y	7
51	16	1884C>A	C628X	n	17

Supplementary Table 1: Usher microarray nucleotide variants (page 2 of 3)

#	Exon	Nucleotide change	Amino acid change	Validated	Ref.
MYO7A					
52	16	1900C>T	R634X	n	14
53	16	1935G>A	M654I/splice defect	n	16
54	16	1935+4A>G	splice defect	n	10
55	17	1945C>T	R649W	y	7
56	17	1952T>C	L651P	n	18
57	17	1969C>T	R657W	n	7
58	17	1996C>T	R666X	y	6
59	17	2005C>T	R669X	y	14
60	17	2028C>G	Y676X	n	7
61	17	2035_2036insT	E680fs	n	7
62	18	2171delC	K725fs	n	7
63	18	2187+1G>A	splice defect	n	13
64	20	2302A>T	K768X	y	7
65	20	2323C>T	Q775X	y	7
66	21	2461C>T	Q821X	y	17
67	21	2476G>A	A826T	n	13
68	22	2662_2670del	K888_K890del	n	19
69	23	2766_2679del	K923fs	y	7
70	23	2863G>A	G955S	n	8
71	23	2878G>T	E960X	n	6
72	23	2904G>T	E968D/splice defect	y	9
73	25	3109-2A>G	splice defect	n	7
74	25	3171C>G	Y1057X	n	7
75	25	3238A>T	K1080X	y	20
76	25	3260T>C	L1087P	n	14
77	25	3265delG	A1089fs	y	14
78	28	3508G>A	E1170K	y	20
79	28	3547C>A	P1183T	y	7
80	28	3596_3597insT	C1201fs	n	11
81	28	3630+2T>A	splice defect	n	8
82	29	3718C>T	R1240W	y	7
83	29	3719G>A	R1240Q	y	6
84	29	3750+2T>A	splice defect	n	7
85	30	3862G>C	A1288P	y	6
86	31	3979G>A	E1327K	n	17
87	31	4018G>A	A1340T	y	7
88	31	4029G>C	R1343S	n	6
89	31	4036_4038delTTC	F1346del	n	9
90	31	4039_4053del#	R1347_F1351del#	n	17
91	31	4073delC	E1359fs	y	7
92	34	4501_4502delGT	V1501fs	y	7
93	34	4543_4551delGAGATCATGinsGCA	E1515_M1517delinsA	n	7
94	35	4697C>T	T1566M	y	17
95	35	4770_4771insT	R1591fs	y	7
96	35	4805G>A	R1602Q	n	18
97	35	4816delA	K1606fs	n	7
98	36	4882G>T	A1628S	n	6
99	36	4918delG	G1640fs	n	7
100	37	5156A>G	Y1719C*	y	20
101	38	5215C>T	R1739X	y	7
102	38	5227C>T	R1743W	y	9
103	39	5392C>T	Q1798X	y	6
104	39	5411delT	L1804fs	n	7
105	40	5573T>C	L1858P	n	9
106	40	5581C>T	R1861X	y	13
107	40	5618G>A	R1873Q	y	7
108	41	5637-3C>G	splice defect	y	7
109	41	5637-1G>A	splice defect	n	9
110	41	5648G>A	R1883Q	y	7
111	41	5660C>T	P1887L	n	9
112	43	5886_5888del	F1963del	y	7
113	44	5945-1G>A	splice defect	n	7
114	44	5968C>T	Q1990X	n	7
115	44	6025delG	A2009fs	y	9
116	45	6193delC	Q2066fs	n	13
117	47	6410G>A	G2137E	n	7
118	48	6487G>A	G2163S	n	6
119	49	6560G>A	G2187D	n	9
PCDH15					
1	2	7C>T	R3X	n	21,22
2	5	400C>G	R134G	n	23
3	8	733C>T	R245X	y	24
4	8	785G>A	G262D	n	23
5	10	1086delT	L362fs	n	22
6	16	1927C>T	R643X	n	23
7	28	3686-2A>G	splice defect	n	21
USH1C					
1	2	91C>T	R31X	y	25
2	3	238_239insC	R80fs	y	26,27
3	5	496+1G>A	splice defect	n	25
4	6	497-2delA	splice defect	y	27
5	B	$32G>T	G431V	n	28
6	D	$293C>G	P608R	y	28
7	D	$329G>T	R620L	n	28
8	D	$376C>T	R636C	n	28
USH1G					
1	1	143T>C	L48P	n	29
2	2	186_187delCA	D62fs	n	29
3	2	395_396insG	V132fs	y	29
4	2	829_848del	S278fs	y	29
USH3A					
1	1	144T>G	N48K	y	30,31
2	1	149_152delCAGG-insTGTCCAAT	S50fs	y	30
3	1	165delC	D55fs	n	30
4	1	189C>A	Y63X	y	31
5	1	187_209del	Y63fs	n	31
6	2	359T>A	M120K	y	32
7	3	449T>C	L150P	n	30
8	3	459_461delATT	I153_L154delinsM	y	32
9	3	528T>G	Y176X	y	32

Supplementary Table 1: Usher microarray nucleotide variants (page 3 of 3)

#	Exon	Nucleotide change	Amino acid change	Validated	Ref.
USH2A					
1	2	94_130del	S33fs	y	7
2	2	100C>T	R34X	n	33
3	2	187C>T	R63X	y	33
4	2	233T>G#	F78C#	n	7
5	2	238_239insCGTA	Q81fs	y	34
6	2	244C>T	R82W	y	35
7	2	377_378delGT	S126fs	n	7
8	3	486-1G>C	splice defect	n	7
9	3	488G>A	C163Y	n	33
10	3	545_446delAA	K182fs	y	36
11	3	588_589insTC	P197fs	y	7
12	4	688G>A	V230M*	y	33
13	4	653T>A	V218E	y	37
14	4	775_776delAG	S259fs	y	36
15	4	779T>G	L260X	y	37
16	6	852_853delGA	E284fs	n	36
17	6	905G>T	C302F	n	7
18	6	921_922insCAGC	H308fs	y	33,37
19	6	949C>A	R317R/splice defect	y	35
20	6	956G>A	C319Y	y	37
21	6	1000C>T	R334W	n	33,34
22	6	1019_1023delATCCT	H340fs	n	7
23	6	1036A>C	N346H	y	33
24	6	1042A>C	N348H	n	37
25	6	1055C>T	T352I	y	7
26	6	1110_1111delTA	I371fs	y	7
27	6	1135C>T	Q379X	y	7
28	7	1144-2A>T	splice defect	y	7
29	7	1172G>T	S391I	y	7
30	7	1215_1216delTA	N405fs	n	7
31	7	1227G>A	W409X	n	37
32	7	1256G>T	C419F	y	37
33	8	1416_1441del	N472fs	y	7
34	8	1434G>C	E478D*	y	36
35	8	1550G>C	R517T/splice defect	y	36
36	9	1604G>C	R535T	n	7
37	9	1606T>C	C536R	y	33
38	10	1663C>G	L555V	n	38
39	10	1679delC	P560fs	y	37
40	10	1696C>T	Q566X	y	37
41	10	1829A>C	H610P	y	39
42	11	1841-2A>G	splice defect	n	39
43	11	1859G>T	C620F	n	7
44	11	1876C>T	R626X	y	33,37
45	11	1965delT	C655fs	n	33
46	11	1966G>A	D656N	y	7
47	12	1985G>T	C662F	n	7
48	12	2023C>T	Q675X	y	33
49	12	2052A>G	Q684Q*	y	37
50	12	2100delG	T701fs	y	36
51	12	2137G>C	G713R*	y	33
52	12	2154delA	A719fs	y	7
USH2A					
53	12	2167+5G>A	splice defect	n	7
54	13	2168-9T>G	splice defect	n	7
55	13	2242C>T	Q748X	y	35
56	13	2276G>T	C759F	y	33,40
57	13	2282C>G	P761R	n	39
58	13	2299delG	E767fs	y	41
59	13	2304C>A	C768X	y	7
60	13	2431_2432delAA	K811fs	n	7
61	13	2522C>A	S841Y*	y	35
62	13	2541C>A	C847X	n	37
63	13	2797C>T	Q933X	y	33
64	14	2878_2879delAA	N960fs	n	33
65	14	2898delG	T967fs	y	41
66	14	2920_2921delGA	D974fs	n	7
67	15	3004delT	C1002fs	y	7
68	16	3158-2A>C	splice defect	y	7
69	17	3368A>G	Y1123C	y	7
70	17	3395G>A	G1132D	y	35
71	17	3558delT	C1186fs	y	7
72	17	3589delT	S1197fs	y	7
73	17	3746C>T	P1249L	y	7
74	18	3840_3841delinsCT	M1280_R1281 delinsIX	y	37
75	18	3883C>T	R1295X	n	33,37
76	18	3896_3897delGC	S1299fs	n	7
77	19	4106C>T	S1369L	y	7
78	19	4174G>T	G1392X	y	7
79	19	4251G>T	Q1417H/splice def.	y	7
80	20	4252-1G>A	splice defect	y	7
81	20	4338_4339delCT	C1447fs	y	41
82	20	4372G>T	G1458X	y	7
83	20	4381C>T	Q1461X	y	7
84	20	4393_4394insAA-AACTTTAGCAG	A1465fs	n	7
85	21	4405C>T	Q1469X	y	35
86	21	4510_4511insA	R1504fs	n	37
87	21	4544C>T	T1515M	n	34
88	23	4810G>C	D1604H	y	42
89	44	8723_8724delTG	V2908fs	y	42
90	50	9815C>T	P3272L	y	42
91	61	11864G>A	W3955X	y	42
92	63	12343C>T	R4115C	y	42
93	63	13274C>T	T4425M	y	42
94	66	14525C>A	S4842X	y	42
VLGR1					
1	31	6901C>T	Q2301X	n	43
2	36	8290T>C	S2764P	n	16
3	37	8495C>A	S2832X	n	16
4	38	8713_8717dupAACA	I2906fs	n	43
5	39	8790delC	M2931fs	n	43
6	89	18732_18750del	Y6244X	n	43

Legend to Supplementary Table 1

Usher microarray nucleotide variants. The nucleotide and amino acid notations are according to Antonarakis and den Dunnen (http://www.hgvs.org/mutnomen/) with the exception of the cDNA ('c.') and protein ('p') annotations. n = not tested; y = validated upon testing; t/n = not validated upon testing. *Presumed polymorphic variants. #Errors in sequence or mutation annotations of the original publications. $Nucleotide numbering within the exons. These variants thus far were observed in non-syndromic deafness patients. All known mutations in the Usher genes were included, regardless of the phenotype.

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