

**Clinical Information on patients.**

Unclassified variants are denoted in brackets. EVA is enlarged vestibular aqueducts

- Indicates no information. Cauc means Caucasian.

Patient	<i>SLC26A4</i> Mutation (cDNA)	<i>SLC26A4</i> Mutation (protein)	Hearing Loss	Imaging	Thyroid function tests TFT (age at test)	Perchlorate	Family History	Goitre	Ethnicity
25215	c.1001+1G>A	-	-	-	-	-	-	-	Cauc
25278	c.1790T>C	p.Leu597Ser	Progressive	Normal	Normal (11)	-	No	No	Cauc
34515	c.716T>A	p. Val239Asp	Progressive	Bilat. EVA	Normal	-	Sibling affected. Not tested	No	Arab
28979	c.1061T>C	p.Phe354Ser	Progressive	Normal	Normal	-	Sister with unilat HL. Not tested	No	Cauc
35265	c.707T>C	p.Leu236Pro	Bilat. Hearing loss	Bilat. EVA	Normal (2)	-	Mild high freq HL in father.	No	Cauc
40257	c.1001+1G>A		Progressive	Bilat. EVA	Hypothyroid	-	No	Yes (autoimmune)	Cauc
37952	c.1151A>G	p.Glu384Gly	Bilat. Hearing loss	Bilat. EVA	-	-	1 sib hypothyroid; 1 sib hearing loss	-	Cauc

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38201	c.1790T>C	p.Leu597Ser	Bilat. Hearing loss	Unilat. EVA	Normal (5)	-	Consanguinity	No	Pakistani
42836	c.707T>C	p.Leu236Pro	Progressive	Bilat. EVA	-	-	-	-	Cauc
42564	(c.1234G>A	p.Val412Ile)	-	-	-	-	-	-	Arab
41066	c.1001+1G>A		Bilat. Hearing loss	Bilat EVA	Normal (32)	Normal	No	No	Cauc
40187	c.412G>T	p.Val138Phe	Bilat. Hearing loss	Bilat. EVA	Normal (39)	Normal	No	No	Cauc
44595	c.2127delT	p.Phe709Leufs*12	Unilat. SNHL	Unilat. EVA	-	-	-	-	Cauc
45381	c.1790T>C	p.Leu597Ser	Bilat. Hearing loss	Normal	Normal (35)	-	No	No	Turkish
13343	c.1342-2_1343dup	p.Leu450Glyfs*19	Bilat. Hearing loss	Bilat. EVA	Normal	Normal	Son affected-no mutation	Yes	Cauc
45592	c.340G>A	p.Gly114Arg	Bilat. Hearing loss	Bilat. EVA	Normal (20)	Normal	Cousin (no mutation)	No	Cauc
48799	c.1001+1G>A		Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
46182	c.707T>C	p.Leu236Pro	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
50939	c.2190G>T	p.Gln730His	asymmetrical Bilat. Hearing loss	Bilat. EVA	-	-	-	-	-
51079	c.707T>C	p.Leu236Pro	-	-	-	-	-	-	-
54165	c.2T>C	p.Met1?	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc

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59858	c.2080T>C	p.Ser694Pro	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	African
61452	c.412G>T	p.Val138Phe	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
54483	c.1790T>C	p.Leu597Ser	Unilat. Hearing loss	Unilat. EVA	Normal (43)	Normal	Daughter (no mutation)	Small autoimmune	Cauc
63420	c.1151A>G	p.Glu384Gly	Bilat. Hearing loss	-	Raised TSH	-	-	-	-
65983	c.1211C>T	p.Thr404Ile	-	-	-	-	-	-	-
50886	c.1151A>G	p.Glu384Gly	Bilat. Hearing loss	-	-	-	-	-	Cauc
66609	c.-3-2A>G	(p.Gly740Val)	Progressive	Bilat. EVA	-	-	-	-	Cauc
66643	c.[1001+1G>A(;) 2219C>T]		Bilat. Hearing loss	-	-	-	-	-	Cauc
66830	c.1790T>C	p.Leu597Ser	Unilat. Hearing loss	Unilat. EVA	Normal (1)	-	-	-	Cauc
69863	c.113T>C	p.Phe335Leu	Bilat. Hearing loss	-	-	-	-	-	Cauc
47141	c.1790T>C	p.Leu597Ser	Unilat. Hearing loss	Unilat. EVA check	-	-	No	No	Cauc
72446	c.-3-2A>G		Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
72770	c.412G>T	p.Val138Phe	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
72617	c.1151A>G	p.Glu384Gly	Unilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc

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76349	c.1790T>C	p.Leu597Ser	Bilat. Hearing loss	-	-	-	-	-	Cauc
76715	c.1826T>G	p.Val609Gly	Progressive	Bilat. EVA	Normal (4)	-	No	No	Cauc/African American
78124	c.1468A>C	p.Ile490Leu	Unilat. Hearing loss	Unilat. EVA	-	-	-	-	Cauc
78231	c.1001+1G>A		Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
23853	c.2T>C	p.Met1?	Bilat. Hearing loss	Bilat. EVA	Normal (12)	-	No	No	Cauc
71753	c.1342-2_1343dup	p.Leu450Glyfs*19	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
72950	c.1229C>T	p.Thr410Met	Bilat. Hearing loss	Bilat. EVA	-	-	Consanguinity	-	Asian
79945	(c.2219C>T)	(p.Gly740Val)	Progressive	Bilat. EVA	-	-	-	-	?Asian
80435	c.707T>C	p.Leu236Pro	Bilat. Hearing loss	-	-	-	Mother has Pendred (Bilat. Hearing loss, goitre and biallelic mutations)	-	Cauc
10576	c.[1343C>T]; [1991C>T]	p.[Ser448Leu]; [Ala664Ser]	Bilat. Hearing loss	Bilat. EVA	Normal (18)	-	No	No	Cauc
83112	c.1790T>C	p.Leu597Ser	Bilat. Hearing loss	Borderline enlarged EVA	Normal (1)	-	None	No	Cauc/Ashkenazi

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84175	c.119delT (c.918G>A)	p.Leu40Argfs X26	Bilat. Hearing loss	-	-	-	-	Yes	-
86297	c.2153G>T	p.Phe718Ser	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
86482	c.707T>C	p.Leu236Pro	Bilat. Hearing loss	-	-	-	-	-	Cauc
85020	c.1234G>T	p.Gln421Arg	Bilat. Hearing loss	-	-	-	-	-	Cauc
83883	c.1790T>C	p. Leu597Ser	Bilat. Hearing loss	Borderline enlarged EVA	Normal (16)	-	No	No	Asian
87823	c.1790T>C	p. Leu597Ser	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	-
84236	(c.73C>T)	(p.Pro25Ser)	Unilat. Hearing loss	Bilat. EVA	Normal (1)	-	No	No	Cauc
88933	c.1151A>G	p.Glu384Gly	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
89770	c.1001+1G>A		Bilat. Hearing loss	-	-	-	-	-	Cauc
90473	c.1790T>C	p.Leu597Ser	Bilat. Hearing loss	Unilat. EVA	-	-	-	-	Cauc
90511	c.1003T>C	p.Phe335Leu	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
90643	(c.970A>T)	(p.Asn324Tyr)	Bilat. Hearing loss	-	Hypothyroid	-	-	-	-

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91820	c.1790T>C	p.Leu597Ser	Bilat. Hearing loss		Hypothyroid	-	Family history of hypothyroidism	-	Cauc
40013	c.1000G>T	p.Gly334Trp	Bilat. Hearing loss	Bilat. EVA	-	-	-	-	Cauc
89620	c.[1790T>C(;) 412G>T]	p.[Val138Phe( ;) (Leu597Ser)]	Progressive	Bilat. EVA	-	-	-	-	Cauc
94065	c.1342- 2_1343dup	p.Leu450Glyfs *19	Progressive	Bilat. EVA	-	-	-	-	Cauc
89792	c.-103T>C		-	-	-	-	-	-	Cauc
94743	c.1003T>C	p.Phe335Leu	Bilat. Hearing loss	Bilat. EVA	Raised TSH	-	-	-	Cauc
96669	c.2015G>A	p.Gly672Glu	-	-	-	-	-	-	Cauc
95020	c.1363A>T	p.Ile455Phe	Unilat. Hearing loss	Unilat. EVA	Normal (16)	-	No	-	Cauc/ West Indian
99311	c.626G>T	p.Gly209Val	Progressive	Bilat. EVA	-	-	-	-	Cauc
99458	c.1334T>G	p.Leu445Trp	Bilat. Hearing loss	Bilat. EVA	Normal (6)	-	No	-	Cauc