S4 Table. Mutations identified in the *RAI1* gene in the SMS patients without any 17p11.2 deletions (literature data) and in patients with isolated HL (this study).

Nucleotide change	Mutation type	Amino acid change	(CAG)-repeats at 832 nucleotide position	Familial or *de novo*	Number (code) of patients	Common SMS clinical symptoms / Hearing status	References
N-terminal part of the RAI1 protein							
c.238C>T	nonsense	p.Arg80*	$(CAG)_{13\text{-}14}$	no data	1 (RNS78)	+ / normal hearing	[1]
c.253del19	frameshift	p.Leu85fsX60	$(CAG)_{13}/(CAG)_{13}$	*de novo*	1 (SMS153)	+ / normal hearing	[2]
c.518_519insG	frameshift	p.Gln174Profs*64	$(CAG)_{13\text{-}14}$	no data	1 (RNS103)	+ / normal hearing	[1]
c.548delT	frameshift	p.L183RfsX69	$(CAG)_{14}/(CAG)_{14}$	*de novo*	1 (M2911)	+ / HL	[3]
c.707A>T	missense	p.Y236F	$(CAG)_{14}/(CAG)_{14}$	familial	1 (M2732)	+ / normal hearing	[3]
c.725C>T and c.2907C>T [a]	missense	p.P242L and p.D969D	$(CAG)_{13}/(CAG)_{13}$	unclassified	1 (M2543)	+ / no data	[3]
c.1119delC	frameshift	p.Ser373fsX65	NA	*de novo*	1 (SMS201)	+ / normal hearing	[4]
c.1297C>T	nonsense	p.Gln433*	$(CAG)_{13\text{-}14}$	no data	1 (RNS105)	+ / normal hearing	[1]
c.1449delC (c.1308delC)	frameshift	p.E484KfsX35	$(CAG)_{14}/(CAG)_{14}$	*de novo*	1 (M2377=SMS159)	+ / HL	[3,5]
c.1500G>A [a] and c.3791A>G (rs61746214)	missense	p.P500P and p.E1264G	$(CAG)_{14}/(CAG)_{14}$	familial	1 (M2900)	+ / normal hearing	[3]
c.1973G>A	nonsense	W658X	$(CAG)_{13}/(CAG)_{13}$	*de novo*	1 (M2719)	+ / HL	[3]
c.2273G>A	nonsense	p.W758X	NA	*de novo*	1 (patient 1)	+ / no data	[6]
c.2396dupC	frameshift	p.Gly800Trpfs*36	$(CAG)_{13\text{-}14}$	no data	1 (RNS165)	+ / normal hearing	[1]
c.2643delC	frameshift	p.Glu882Serfs*68	$(CAG)_{13\text{-}14}$	no data	1 (RNS86)	+ / normal hearing	[1]
c.2763_2779dup17	frameshift	p.Leu927Glnfs*29	$(CAG)_{13\text{-}14}$	no data	1 (RNS131)	+ / normal hearing	[1]
c.2773del29	frameshift	p.Val925fsX8	$(CAG)_{13}/(CAG)_{13}$	*de novo*	1 (SMS129=BAB539)	+ / normal hearing	[5,7]
c.2836_2837delCT	frameshift	p.Leu946Valfs*7	$(CAG)_{13\text{-}14}$	no data	1 (RNS208)	+ / normal hearing	[1]
c.2869_2870insGG	frameshift	p.Asp957Glyfs*108	$(CAG)_{13\text{-}14}$	no data	1 (RNS101)	+ / normal hearing	[1]
c.2878C>T	nonsense	p.Arg960X	$(CAG)_{10}/(CAG)_{11}$	no data	1 (BAB1106)	+ / normal hearing	[7]
c.3103delC	frameshift	p.Gln1035fsX28	$(CAG)_{10}/(CAG)_{10}$ NA	*de novo* unclassified	1 (BAB2416) 1 (SMS324)	+ / normal hearing + / HL	[8] [9]
c.3103insC	frameshift	p.Q1034PfsX31 (p.Gln1035fsX30)	$(CAG)_{10}/(CAG)_{11}$ NA	*de novo* unclassified	1 (M2754=BAB1852) 1 (SMS335)	+ / HL + / HL	[3,7] [9]

S4 Table. Mutations identified in the *RAI1* gene in the SMS patients without any 17p11.2 deletions (literature data) and in patients with isolated HL (this study).

(continued)

Nucleotide change	Mutation type	Amino acid change	(CAG)-repeats at 832 nucleotide position	Familial or *de novo*	Number (code) of patients	Common SMS clinical symptoms / Hearing status	References
C-terminal part of the RAI1 protein							
c.3183G>A [a] and c.5653G>A [b]	missense	p.T1061T and p.D1885N	$(CAG)_{13}/(CAG)_{13}$	familial	1 (M2365)	+ / HL	[3]
c.3208G>A and c.4512G>T [a]	missense	p.G1070R and p.L1504L	$(CAG)_{14}/(CAG)_{14}$	familial	1 (M2826)	+ / HL	[3]
c.3386dupA	frameshift	p.Glu1130Glyfs*36	$(CAG)_{13\text{-}14}$	*de novo*	1 (RNS59)	+ / normal hearing	[1]
c.3634A>G	missense	p.Ser1212Gly	$(CAG)_{13}/(CAG)_{18}$	familial	1 (BAB2330)	+ / normal hearing	[8]
c.3650G>A	missense	R1217Q	NA	unclassified	1 (SAG4739)	+ / no data	[10]
c.3781_3783delGAG	frameshift	p.del1261E	$(CAG)_{14}/(CAG)_{14}$	familial	1 (M2867)	+ / normal hearing	[3]
c.3801delC	frameshift	p.Pro1267fsX46	$(CAG)_{10}/(CAG)_{11}$	*de novo*	1 (SMS188)	+ / normal hearing	[2]
c.4166A>G	missense	Q1389R	NA	unclassified	1 (SAG6888)	+ / no data	[10]
c.4649delC	frameshift	p.Ser1550fsX36	NA	*de novo*	1 (SMS278)	+ / normal hearing	[4]
c.4685A>G	missense	p.Gln1562Arg	$(CAG)_{9}/(CAG)_{11}$	*de novo*	1 (SMS175)	+ / normal hearing	[2]
c.4933delGCCG	frameshift	p.Ala1645fsX35	NA	*de novo*	1 (SMS300)	+ / normal hearing	[4]
c.5254G>A	missense	**p.Gly1752Arg**	$(CAG)_{13}/(CAG)_{13}$	familial	14 (10 homozygotes and 4 heterozygotes)	- / HL	**this study**
c.5265delC (c.4929delC)	frameshift	p.Pro1755fsX74	$(CAG)_{10}/(CAG)_{11}$	*de novo*	1 (SMS156=BAB526)	+ / HL	[5,7]
c.5423G>A	missense	p.Ser1808Asn	$(CAG)_{10}/(CAG)_{11}$	unclassified	1 (SMS195)	+ / normal hearing	[2]

HL – hearing loss. [a] - these silent variants are probably not related to the phenotype SMS [3]; [b] - variant in exon 4; NA – not analyzed

References for S4 Table

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