**S3 Table. The *RAI1* genotypes with allelic variations detected by Sanger sequencing in individuals homozygous and heterozygous for c.5254G>A.**

| Code | Hearing status | Genotypes | | | | | Allele |
|---|---|---|---|---|---|---|---|
| | | **rs3803763** c.269G>C p.Gly90Ala | **rs11649804** c.493C>A p.Pro165Thr | **Variations in polyQ region *** | **rs8067439** c.1992G>A p.Pro664Pro | **Mutation** c.5254G>A p.Gly1752Arg | |
| **Homozygotes for c.5254G>A** | | | | | | | |
| **38-II-5** | HL | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | A/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A |
| **40-II-1** | HL | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | A/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A |
| **42-II-1** | HL | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | A/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A |
| **43-III-1** | HL | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | A/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A |
| **Affected heterozygotes for c.5254G>A** | | | | | | | |
| **18-II-1** | HL | G/C | C/A | $Q_{13}$ [CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{14}$ [CAG CAG (CAG)$_{11}$ CAA] | G/A | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_1$: G-C-$Q_{14}$[CAG CAG (CAG)$_{11}$ CAA]-A-G |
| **37-II-1** | HL | G/C | C/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAG (CAG)$_{10}$ del(CAG) CAA] | G/A | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_2$: G-C-$Q_{13}$[CAG CAG (CAG)$_{10}$ del(CAG) CAA]-A-G |
| **38-I-1** | HL | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_3$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-G |
| **43-II-2** | HL | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_3$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-G |
| **Normal hearing heterozygotes for c.5254G>A** | | | | | | | |
| **37-I-2** | normal hearing | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_3$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-G |
| **43-II-3** | normal hearing | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_3$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-G |
| **38-II-1** | normal hearing | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_3$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-G |
| **Alt-1** | normal hearing | C/C | A/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] | G/G | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_3$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-G |
| **Alt-2** | normal hearing | C/C | C/A | $Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA] / $Q_{11}$[CAG CAG (CAG)$_8$ del(CAG)$_3$ CAA] | G/A | G/A | Allele$_{mut}$: C-A-$Q_{13}$[CAG CAA (CAG)$_{10}$ del(CAG) CAA]-G-A<br>Allele$_4$: C-C- $Q_{11}$[CAG CAG (CAG)$_8$ del(CAG)$_3$ CAA]-A-G |

HL – hearing loss, * NM_030665 ref seq.: (CAG)$_{13}$ CAA=$Q_{14}$; variations in polyQ region (starting from 832 nucleotide position) includes rs11078398: c.837G>A (caG/caA) p.Gln279Gln; codon deletions: del(CAG) and del(CAG)$_3$. The common haplotype for c.5254G>A (allele$_{mut}$) is highlighted by yellow, allele$_1$, allele$_2$, allele$_3$, and allele$_4$ - by grey, pink, blue, and green, respectively. Nucleotides embedded in common haplotype for c.5254G>A (allele$_{mut}$) are shown by red.