

Supplemental table S2. Molecular analyses FHONDA patients from literature*

Family	Descent	Mutations SLC38A8 gene
Poulter <i>et al.</i> F1:IV:1 ^{6,7,14}	<i>Pakistani</i>	Homozygous c.707T>A; p.(Val236Asp)
Poulter <i>et al.</i> F1:IV:3 ^{6,7,14}	<i>Pakistani</i>	Homozygous c.707T>A; p.(Val236Asp)
Poulter <i>et al.</i> F1:IV:4 ^{6,7,14}	<i>Pakistani</i>	Homozygous c.707T>A; p.(Val236Asp)
Poulter <i>et al.</i> F1:IV:6 ^{6,7,14}	<i>Pakistani</i>	Homozygous c.707T>A; p.(Val236Asp)
Poulter <i>et al.</i> F1:IV:7 ^{6,7,14}	<i>Pakistani</i>	Homozygous c.707T>A; p.(Val236Asp)
Poulter <i>et al.</i> F4:II:1 ⁷	<i>Pakistani</i>	Homozygous c.1029del; p.(Leu344Cysfs*7)
Poulter <i>et al.</i> F5:IV:1 ⁷	<i>Turkish</i>	Homozygous c.101T>G; p.(Met34Arg)
Poulter <i>et al.</i> F6:IV:7 ^{7,16}	<i>Indian</i>	Homozygous c.697G>A; p.(Glu233Lys)
Poulter <i>et al.</i> F6:VI:1 ^{7,16}	<i>Indian</i>	Homozygous c.697G>A; p.(Glu233Lys)
Poulter <i>et al.</i> F6:VI:2 ^{7,16}	<i>Indian</i>	Homozygous c.697G>A; p.(Glu233Lys)
Toral <i>et al.</i> II:1 ¹⁵	<i>Ashkenazi-Jewish</i>	Homozygous c.848A>C; p.(Asp283Ala)
Toral <i>et al.</i> II:2 ¹⁵	<i>Ashkenazi-Jewish</i>	Homozygous c.848A>C; p.(Asp283Ala)
Campbell <i>et al.</i> proband 2 ¹⁷	<i>British</i>	c.534C>G; p.(Ile178Met) and c.(189+1_190-1)_(690+1_691-1)del; p.(?) deletion exon 2 to 5
Weiner <i>et al.</i> F1:II:1 ¹⁸	<i>Karait Jewish</i>	Homozygous c.95T>G; p.(Ile32Ser)
Weiner <i>et al.</i> F1:II:2 ¹⁸	<i>Karait Jewish</i>	Homozygous c.95T>G; p.(Ile32Ser)
Weiner <i>et al.</i> F2:II:3 ¹⁸	<i>Karait Jewish</i>	Homozygous c.95T>G; p.(Ile32Ser)
Weiner <i>et al.</i> F3:II:1 ¹⁸	<i>Indian Jewish</i>	Homozygous c.95T>G; p.(Ile32Ser)
Weiner <i>et al.</i> F3:II:2 ¹⁸	<i>Indian Jewish</i>	Homozygous c.95T>G; p.(Ile32Ser)
Weiner <i>et al.</i> F4:II:1 ¹⁸	<i>Indian Jewish</i>	Homozygous c.95T>G; p.(Ile32Ser)
Weiner <i>et al.</i> F5:II:1 ¹⁸	<i>Indian Jewish</i>	c.95T>G; p.(Ile32Ser) and c.490-491del; p.(Leu164Valfs*41)
Lasseaux <i>et al.</i> P2 ¹³	<i>Caucasian</i>	c.527C>G; p.(Thr176Arg) and c.848A>C; p.(Asp283Ala)
Lasseaux <i>et al.</i> P4 ¹³	<i>Caucasian</i>	c.6_9del; p.(Glu2Aspfs*32) and c.923C>G; p.(Thr308Ser)
Kuht <i>et al.</i> F1:II-1 ¹⁴	<i>Korean</i>	c.692G>A; p.(Cys231Tyr) and c.964C>T; p.(Gln322*)
Kuht <i>et al.</i> F1:II-2 ¹⁴	<i>Korean</i>	c.692G>A; p.(Cys231Tyr) and c.964C>T; p.(Gln322*)

<i>Kuht et al</i> F2:II-1 ¹⁴	<i>Korean</i>	c.558C>A; p.(Tyr186*) and c.1078_1104del; p.(Ala360_Leu368del)
<i>Kuht et al</i> F3:II-1 ¹⁴	<i>Korean</i>	c.995dupG; p.(Trp333Metfs*35) and c.1214+5G>C
<i>Kuht et al</i> F4:II-1 ¹⁴	<i>Korean</i>	c.855G>C; p.(Leu285Phe) and c.995dupG; p.(Trp333Metfs*35)
<i>Kuht et al</i> F5:II-1 ¹⁴	<i>Korean</i>	c.644G>T [†] ; p.(Trp215Leu), c.682G>A [†] ; p.(Gly228Arg) and c.695A>G; p.(His232Arg)
<i>Kuht et al</i> F5:II-2 ¹⁴	<i>Korean</i>	c.644G>T [†] ; p.(Trp215Leu), c.682G>A [†] ; p.(Gly228Arg) and c.695A>G; p.(His232Arg)
<i>Kuht et al</i> F6:II-2 ¹⁴	<i>Korean</i>	c.954-1G>C and c.995dupG; p.(Trp333Metfs*35)
<i>Kuht et al</i> F7:II-3 ¹⁴	<i>Turkish</i>	Homozygous c.101T>G; p.(Met34Arg)
<i>Kuht et al</i> F8:II-1 ¹⁴	<i>Caucasian</i>	Homozygous c.632+2T>G
<i>Kuht et al</i> F9:II-1 ¹⁴	<i>Caucasian</i>	Exon 1 deletion and c.1126G>A; p.(Gly376Arg)

*FHONDA patient P1 from Lasseaux *et al* was discarded from their series.¹³ [†]These two variants were present in cis configuration¹⁴