**S1 Fig.**

*SCN9A* ex7 AATATTTTTATTACTTGGAAGGATCCAAAGATGCTCTCCTTTGTGGTTTCAGCACAGATT

||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||

NAT AATATTTTTATTACTTGGAAGGATCCAAAGATGCTCTCCTTTGTGGTTTCAGCACAGATT

*SCN9A* ex7 CAGG

||||

NAT CAGG

Nav1.7

302 YFYYLEGSKDALLCGFSTDS 321

*SCN9A* ex15 CTTAGTGTTGGCCATCATCGTCTTCATTTTTGCTGTGGTCGGCATGCAGCTCTTTGGTAA

||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||

NAT CTTAGTGTTGGCCATCATCGTCTTCATTTTTGCTGTGGTCGGCATGCAGCTCTTTGGTAA

*SCN9A* ex15 GAGCTACAAAGAATGTGTCTGCAAGATCAATGATGACTGTACGCTCCCACGGTGGCACAT

||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||

NAT GAGCTACAAAGAATGTGTCTGCAAGATCAATGATGACTGTACGCTCCCACGGTGGCACAT

*SCN9A* ex15 GAACGACTTCTTCCACTCCTTCCTGATTGTGTTCCGCGTG

||||||||||||||||||||||||||||||||||||||||

NAT GAACGACTTCTTCCACTCCTTCCTGATTGTGTTCCGCGTG

Nav1.7

860 LVLAIIVFIFAVVGMQLFGKSYKECVCKINDDCTLPRWHMNDFFHSFLIVFRV 912

A863P (erythromelalgia)[[1](#_ENREF_1)]

V872G (erythromelalgia)[[2](#_ENREF_2)]

Q875E (erythromelalgia)[[3](#_ENREF_3),[4](#_ENREF_4)]

R896Q (congenital insensitivity to pain)[[5](#_ENREF_5)]

W897X (congenital insensitivity to pain)[[6](#_ENREF_6)]

*SCN9A* ex17 AGATTAAACCGGTCAAGCTCCTCAGAGTGCAGCACAGTTGATAACCCTTTGCCTGGAGAA

||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||

NAT AGATTAAACCGGTCAAGCTCCTCAGAGTGCAGCACAGTTGATAACCCTTTGCCTGGAGAA

*SCN9A* ex17 GGAGAAGAAGCAGAGGCTGAACCTATGAATTCCGATGAGCCAGAGGC

|||||||||||||||||||||||||||||||||||||||||||||||

NAT GGAGAAGAAGCAGAGGCTGAACCTATGAATTCCGATGAGCCAGAGGC

Nav1.7

1107 RLNRSSSSECSTVDNPLPGEGEEAEAEPMNSDEPE 1141

*SCN9A* ex26 CCTGTTCCGAGTGATCCGTCTTGCCAGGATTGGCCGAATCCTACGTCTAGTCAAAGGAGC

||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||

NAT CCTGTTCCGAGTGATCCGTCTTGCCAGGATTGGCCGAATCCTACGTCTAGTCAAAGGAGC

*SCN9A* ex26 AAAGGGGATCCGCACGCTGCTCTTTGCTTTGATGATGTCCCTTCCTGCGTTGTTTAACAT

||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||

NAT AAAGGGGATCCGCACGCTGCTCTTTGCTTTGATGATGTCCCTTCCTGCGTTGTTTAACAT

*SCN9A* ex26 CGGCCTCCTGCTCTTCCTGGTCATGTTCATCTACGCCATCTTTGGAATGTCCAACTTTGC

||||||||||||||||||||||||||||||||||||||||||||||||||||||||||||

NAT CGGCCTCCTGCTCTTCCTGGTCATGTTCATCTACGCCATCTTTGGAATGTCCAACTTTGC

Nav1.7

1597 LFRVIRLARIGRILRLVKGAKGIRTLLFALMMSLPALFNIGLLLFLVMFIYAIFGMSNF 1655

R1599X (congenital insensitivity to pain)[[7](#_ENREF_7)]

L1612P (cold sensitive paroxysmal extreme pain disorder (PEPD))[[8](#_ENREF_8)]

M1627K (PEPD)[[9](#_ENREF_9)]

A1632E (overlapping phenotype of erythromelalgia and PEPD) [[10](#_ENREF_10)]

A1632T (erythromelalgia)[[11](#_ENREF_11)]

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