

Supplementary A

Table S1. Details of affected family members and sequencing method.

Family #	Age-at-onset	Sequencing Method
002	63	Perkin Elmer HiSeq
002	53	Perkin Elmer HiSeq
002	66	Nextera MiSeq
002	40 ‡	Ampliseq Ion Torrent
431	ND	Perkin Elmer HiSeq
431	62	Perkin Elmer HiSeq
431	66	Ampliseq Ion Torrent
433	60	Ampliseq Ion Torrent
433	59	Ampliseq Ion Torrent
433	81	Ampliseq Ion Torrent
447	69	Ampliseq Ion Torrent
447	52	Ampliseq Ion Torrent
447	67	Ampliseq Ion Torrent
460	70	Ampliseq Ion Torrent
460	67	Ampliseq Ion Torrent
460	52	Ampliseq Ion Torrent
484	48	Nextera MiSeq
484	87	Ampliseq Ion Torrent
484	52	Ampliseq Ion Torrent

ND: Did not disclose. ‡ Represents suggestive prodromal case.

Table S2. Primer designs.

Sanger Sequencing Verification †					
Gene	Mutation	Forward 5'-3' Primer	Reverse 5'-3' Primer	Annealing Temperature °C	Size bp
<i>KCNJ15</i>	p.R28C	TTCTCCTTTCTTGTG-TACTTCC	CAGGGTGAGTTT-GTATCTCC	57	340
<i>PASK</i>	p.P519L	ATCCTCAGCTGG-GACTGGAG	TGGTCACCCATGTCATTATC	57	233
<i>SON</i>	p.S1595P	GCTGGTACTAG-TCCTGTTGG	CTGGTGGAAATTAC-CATGTC	57	281
<i>SIPA1L1</i>	p.R236Q	GCAGAATAC-GCCAGCGAAG	TGACAGAG-TCTTCTGATCGGTTATC	60.5	465
<i>ZNF462</i>	p.I1523V	GAGGATGCAA-GACTGTCCCC	CAAAGTGCCGTGTGTG-TACG	60.5	432
<i>DUSP19</i>	p.I111R	ATGGGTTAG-TAGGTAAGGGAGG	TGCCCTCTTGA-TATGTACGAAGC	60.5	408
<i>KCTD1</i>	p.G134R	TCGAGTCGTGAT-TCCAGCC	TGATTAG-TGACATAACAA-GAAGTGG	60.5	360
<i>TAF1C</i>	p.R346Q	CACTGAGCTT-GTCCTCTAGGC	GTGCTCTTCCAGCTCTCGG	60.5	406
<i>DARS2</i>	p.S59L	ATTTCCCCTG-TAGCCCTGAC	ACGTTTTATTCTTCG-GACTGGTTG	60.5	320

<i>EXPH5</i>	p.T920S	TTGGTGTTAGCTCAGAA GCATCC	AGCAGTTTTCTCATA- CATGGCAGG	60.5	385
<i>FAM71B</i>	p.I318T	GCCATCGAGGTG- GAAGTACC	CTAG- TGCGGCTTCTCCATCC	60.5	349
<i>CCDC180</i>	p.R1684C	GAGAACTCTTGCCAG- TGCCT	TGGTAAAGCTACAG- TGCCCG	60.5	395
<i>SLC2A12</i>	p.S357L	GGGGCATGAG- TGAGCTTCTG	TGTACAAATCAC- TGGCCAACC	60.5	401
<i>DOCK3</i>	p.R392W	GCTGGG- GAACAGAGGGAAAG	CCTGACTTGGGATGTGG- GAG	60.5	532
<i>TPR</i>	p.K1038 N	TTTGTAAGGTCCAC- CAGTTCTT	AGGGAATGGA- TATGGAGTTGCA	60.5	395
<i>DNAH1</i>	p.K1792R	CCTCAAGCGA- GAAAACCCCA	CTGACCTG- GACTCCAAGGTG	60.5	390
<i>PCDHGA7</i>	p.S667G	AGAAC- GCCTGGCTGTCATAC	CAAGCCACCTTCTGAA- GCCT	60.5	398
<i>MYOT</i>	p.N30K	GGAGA- CACATCACCTTTGTCA	CCCAGCAGAT- TATTCTTACCACC	60.5	658
<i>KIF9</i>	p.R287W	GTGAATCCATGCCAC- CAGGA	GCTGACCTGAC- CACAGACTT	60.5	374
<i>DNAJC12</i>	p.T99M	ACAGGCTCTTCTGG- CACAAA	CTCCGAGAGA- GAAGTGCTCA	60.5	386
<i>SLAMF8</i>	p.V234E	TCCTGCATT- GTCTCCAACCC	GAGAGGTAGGAAGGGG- CAGA	60.5	378
<i>ZNF75A</i>	p.Q212E	GGAAACAA- GAGCTGCTCAAAC	CTCAC- CTGTGTGGGTTCTCC	60.5	392
TaqMan Genotyping					
Gene	Mutation	Forward 5'-3' Primer	Reverse 5'-3' Primer	Annealing Temperature °C	Size
<i>KCNJ15</i>	p.R28C	CCCCTGGTGAA- GCACACT	GCTGTGCCCACTCTT- GGA	62	75
		MGB-Probe(WT): ACAGACCCCGCGTCAT	MGB-Probe(MT): AACAGACCCTGCGTCAT		
High Resolution Melt Genotyping					
<i>ARL14EP</i>	p.A146V	TTGTCTGTTGTCAAGAT- TTGAGTTG	AGTTTGCCTTCTGGAT- TCG	60	105
<i>SIPA1L1</i>	p.R236Q	AAACAGGGAACATCTG- GAGAAAGC	TGCCACCACCAG- TAATGAGAAAG	60	118

† Requirements for Sanger sequencing validation was lesser in kindreds that were sequenced across multiple platforms due to likelihood of same machine-artefacts. Therefore, putative variants from families 484 and 431 did not undergo additional Sanger sequencing confirmation.

Table S3. Genotyping variants from #002 by country.

Country	Platform	Cases	Controls
Australia	TaqMan	1372	1397
Canada	MassARRAY	276	70
Germany	MassARRAY	112	60
Italy	TaqMan & MassARRAY	1243	183
Norway	MassARRAY	1379	1095

Saskatoon	MassARRAY	640	366
South Africa	MassARRAY	117	272
Sweden	MassARRAY	416	792
Taiwan	MassARRAY	637	320

Family #006

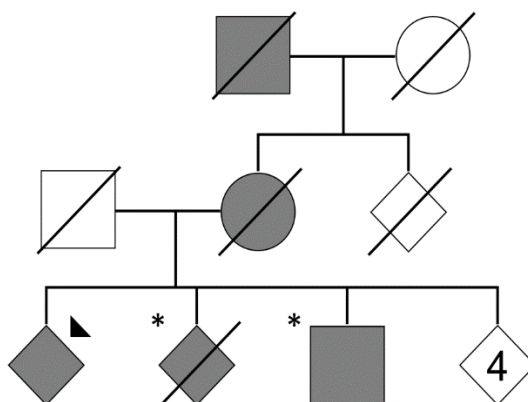


Figure S1. Queensland kindred, #006, with two affected members carrying the *ZNF75A* p.Q212E mutation, as indicated by the star. Whole exome sequencing performed on three affected siblings, finding 20 shared rare sequence variants. Full shaded shapes represent PD. Triangle suggests proband case. Gender disclosed and minimum data shown to protect the privacy of the participants.

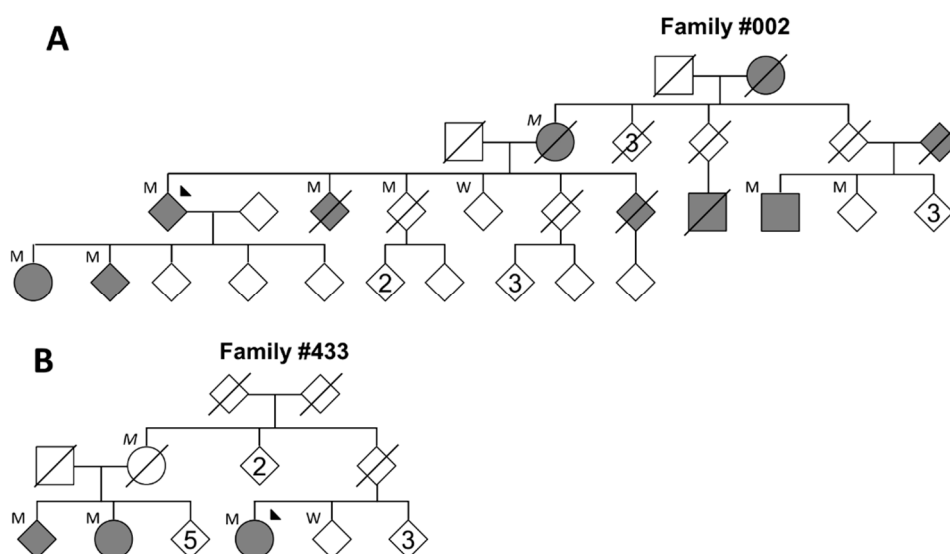


Figure S2. (A) Segregation of the *KCNJ15* p.R28C variant in family #002. **(B)** Segregation of *SIPA1L1* p.R236Q variant in family #433. Full shaded shapes represent PD. M represents heterozygous mutation carrier. Italicized M represents inferred mutation carrier >80 years. W represents confirmed homozygous reference >80 years. Triangle suggests proband case. Gender disclosed and minimum data shown to protect the privacy of the participants.