

SUPPLEMENTARY INFORMATION

Supplementary Methods: Cloning

It was necessary to express *SCN9A* with the auxiliary β_1 and β_2 subunits (encoded by *SCN1B* and *SCN2B*) as the β subunits are needed for normal function of voltage-gated sodium channels. The constructs were designed so that each gene was expressed separately (i.e. not as a fusion protein) through use of internal ribosome entry site (IRES) sequences (Fig. 4a).

The wild-type *SCN9A* → polio IRES → *DsRed2* construct (Fig. 4a) was made as follows. Human fetal brain cDNA was prepared using the Superscript First-Strand Synthesis System for RT-PCR (Invitrogen) using human fetal brain total RNA (Clontech) as template. pcDNA3 (Invitrogen) was rendered a low-copy number vector as previously described (clone name pcDNA3JC)⁶. We and previous authors observed that *SCN9A* appears to be toxic to *E. coli*, thus it was only possible to grow *E. coli* containing the *SCN9A* constructs using such an approach. Human fetal brain cDNA was used to amplify a 2333 bp product (the first third of *SCN9A*) using the forward primer (5' GGG GTA CCC CGT GAG GAG CTG AAG AGG AAT TA) and the reverse primer (5' CCG CTC GAG CGG ATT GGG TGG TGT TCC ATA GC) and Phusion High-Fidelity DNA Polymerase (New England Biolabs). This fragment was digested with *Kpn*I and *Xho*I and ligated into pcDNA3JC to generate the clone FLA. A second amplicon (3994 bp) comprising the remaining two-thirds of *SCN9A* was generated using the forward primer (5' TCA GAG GAT ATG CTG AAT GAT C) and the reverse primer (5' CCG CTC GAG CGG CAC TTT CAC AGG CTG TAA ACA) using human fetal brain cDNA as template. Following digestion with *Bst*XI and *Xho*I this fragment was ligated into clone FLA to give clone FLB, which contained the complete *SCN9A* open reading frame. To generate the polio IRES-*DsRed2* fragment which follows the *SCN9A* cDNA (Fig. 4a), a fragment from the in-house vector TRIS5EGFP

containing the polio IRES flanked by *Bgl*II and *Age*I sites was cloned into pDsRed2-N1 (Clontech) to give the clone POLRED1. The polio IRES-*DsRed2* fragment was then PCR-amplified out of POLRED1 (1487 bp) using the forward primer (5' CCG CTC GAG CGG CCGCTAGCGCTACCGGACTC) and the reverse primer (5' CTAGA GGGCCC CTA CAG GAA CAG GTG GTG GC). Following digestion with *Xho*I and *Apa*I this fragment was ligated into plasmid FLB to give the final clone FLRED (wild-type *SCN9A* → polio IRES → *DsRed2*). For these cloning steps the ligation reactions were initially transformed into XL-10 Gold Ultracompetent Cells (Stratagene) and single colonies grown up for isolation of DNA using the QIAprep Spin Miniprep Kit (Qiagen). A DNA aliquot was checked by restriction digest and then plasmid DNA was transformed into SURE2 Supercompetent Cells (Stratagene). Resultant single colonies were grown up for DNA isolation using the HiSpeed Plasmid Midi Kit (Qiagen). The final clone (FLRED) was sequenced entirely and corresponds to *SCN9A* RefSeq sequence NM_002977, the most common splice variant found in dorsal root ganglia²¹. The clone FLRED was used as template to generate the 3 nonsense mutations (Families 1, 2 and 3) using the QuikChange XL Site-Directed Mutagenesis Kit (Stratagene) according to the manufacturer's instructions.

The *SCN1B* → encephalomyocarditis virus (ECMV) IRES → *SCN2B* → polio IRES → *EGFP* construct (Fig. 4a) was made as follows. *SCN2B* was PCR-amplified from human fetal brain cDNA using the forward primer (5' CTG CAG AAC CAC AAC CAT GGA TGC ACA GAG ATG CCT GGC TAC) and the reverse primer (5' GAA GAT CTT CGT CCT AGG TCA CGG GAA GCA C) to generate a 782 bp fragment which was subsequently digested with *Bst*XI and *Bgl*III. The polio IRES-*EGFP* fragment (1488 bp) was cut out of the in-house vector TRIS5EGFP using *Bgl*III and *Not*I. The *SCN2B* and polio IRES-*EGFP* fragments were ligated into the *Bst*XI and *Not*I restriction sites of pIRES2-AcGFP1 (Clontech) to generate the clone JC3. *SCN1B* was PCR-amplified from human fetal brain cDNA using the forward primer (5' CCG CTC GAG CGG GGA CAT TCT AAC CGC CGC

CA) and the reverse primer (5' CGC GGA TCC ACC CCA GTG GCT CCG TCG GCA GG). This 906 bp fragment was digested with *Xho*I and *Bam*HI and ligated into clone JC3 to generate clone JC4. The QuikChange Site-Directed Mutagenesis Kit (Stratagene) was used to remove 4 nucleotides derived from the *Bst*XI restriction site using the forward primer (5'AAT ATG GCC ACA ACC ATG CAC AGA GAT GCC) and the reverse primer (5' GGC ATC TCT GTG CAT GGT TGT GGC CAT ATT) to generate the final clone JC5 (*SCN1B* → ECMV IRES → *SCN2B* → polio IRES → *EGFP*). This clone was sequenced entirely by standard methods.

Supplementary Figure 1

Kinetic properties of whole cell wild-type $\text{Na}_v1.7+\text{Na}_v\beta 1+\text{Na}_v\beta 2$ currents in HEK293 cells.

- a) Voltage dependence of the activation time constant τ_m and inactivation time constant τ_h , obtained by fitting current traces elicited by depolarizing pulses from -40 to +40 mV of 50 ms duration from a holding potential of -100 mV (as shown in Fig. 4) with a Hodgkin-Huxley m^3h model (n=11).
- b) Development of steady-state inactivation. Example currents elicited by a test pulse to -10 mV after a prepulse to -50 mV of increasing duration; the voltage pulse protocol is indicated below. The time course of the peak currents in response to the test pulse were fitted by a single exponential to obtain the inactivation time constant τ_{inact} at -50 mV.
- c) Voltage dependence of τ_{inact} , obtained by fitting data as shown in (b) after prepulses to different potentials (n=12-14).
- d) Recovery from inactivation. Example currents elicited by a test pulse of 20 ms duration to -20 mV after currents were initially inactivated by a similar prepulse and then allowed to recover at -100 mV for increasing time periods; the voltage pulse protocol is indicated below. The time course of the peak current in response to the test pulse was fitted with a single exponential to obtain the recovery time constant τ_{rec} at -100 mV.

e) Voltage dependence of τ_{rec} , obtained by fitting data as shown in (d) after recovery at different potentials (n=7-15).

Supplementary Figure 2

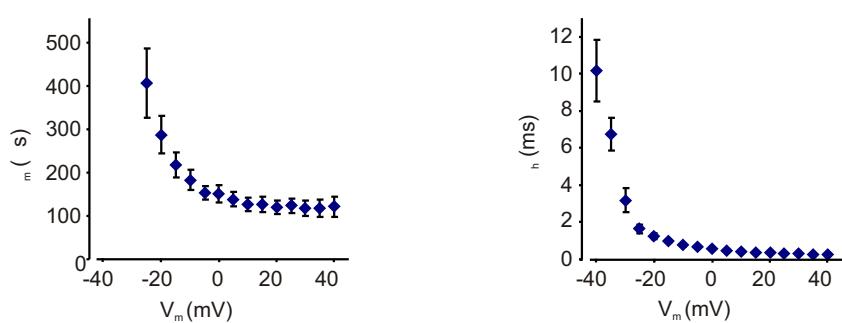
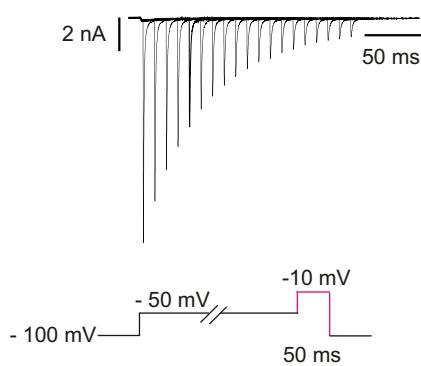
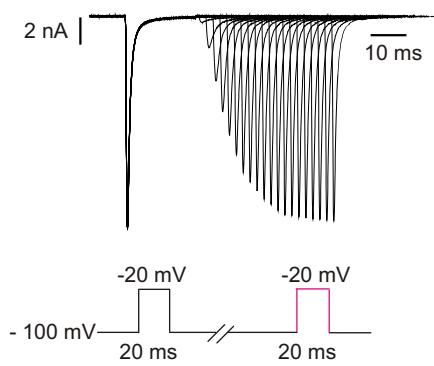
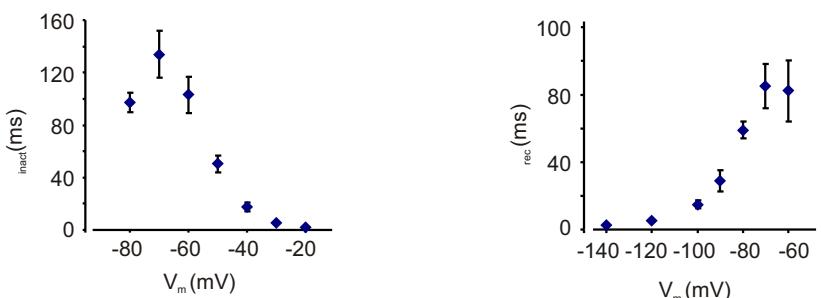
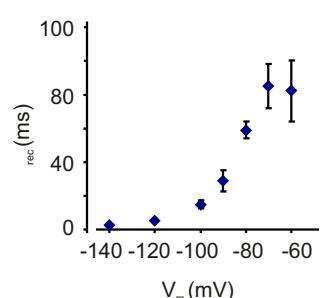
Alignment of the amino acid sequences of the corresponding common splice variants for the human (NP_002968) and mouse Na_v1.7 (XP_904764) proteins using the LALIGN program (:= identical amino acid; . = similar amino acid). The proteins are highly conserved with 92% identity and 95% similarity. Domain structures are also highly conserved between the human and mouse proteins: the putative transmembrane helices are highlighted in yellow; important residues for ion selectivity are highlighted in turquoise; the IFM motif, which is highlighted in green, is known to be important for fast-inactivation kinetics of other sodium channels; the EF-hand calcium-binding motif (InterPro 002048) and the IQ calmodulin binding motif (InterPro 000048) are highlighted in gray and red respectively; and the NEDD4L interaction site, which promotes ubiquitination and internalization of Na_v1.7, is highlighted in purple⁶.

Supplementary Table 1:

Markers used to search for common haplotype blocks between the three families. The homozygous block is bounded by a thickened line; het denotes a heterozygous microsatellite marker genotype.

Supplementary Table 2:

Primers used to amplify and sequence the coding exons and splice sites of SCN9A.

a**b****d****c****e**

Human Na_v1.7 (NP_002968)
Mouse Na_v1.7 (XP_904764)

1977 aa vs.
 1975 aa

92% identity; 95% similarity

	10	20	30	40	50	60
Human	MAMLPPPGPQSFVHFTKQLSLALIEQR	I	AERKSKEPK	EKKDDDEA	AKPSSDLEAG	KQLP
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	MAMLPPPGPQSFVHFTKQLSLALIEQR	I	RISEEKAKGH	KDEKKDDEEGP	KPSSDLEAG	KQLP
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	10	20	30	40	50	60
	70	80	90	100	110	120
Human	FIYGDIPPGMVSEPLEDLD	PYYADKKTFIVLNKG	KTIFRFNATPALYMLSPF	SPLRRISI		
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	FIYGDIPPGMVSEPLEDLD	PYYADKKTFIVLNKG	KAIFRFNATPALYMLSPF	SPLRRISI		
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	70	80	90	100	110	120
	130	140	150	160	170	180
Human	KILVHSLFSMLIMCTILTNC	I	FMTMNNPPDWTKNVEY	TFTGIYTFESLV	KILARGFCVGE	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	KILVHSLFSMLIMCTILTNC	I	FMTMSNPPDWTKNVEY	TFTGIYTFESLIK	I	LARGFCVGE
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	130	140	150	160	170	180
	190	200	210	220	230	240
Human	FTFLRDPW	NWLDFVVIVFAYLTEFVN	LGNVSALRTFRVL	RALKTISVIPGL	KTIVGALIQ	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	FTFLRDPW	NWLDFVVIVFAYLTEFVN	LGNVSALRTFRVL	RALKTISVIPGL	KTIVGALIQ	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	190	200	210	220	230	240
	250	260	270	280	290	300
Human	SVKKLSDV	MILTVFCLSVFALIGLQL	FMGNLKHKCFRNS	LENNETLESIMNT	LESEEDFR	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	SVKKLSDV	MILTVFCLSVFALIGLQL	FMGNLKHKCFRKDL	EQNETLENGHK	GQYQRKCKVK	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	250	260	270	280	290	300
	310	320	330	340	350	360
Human	K--YFYYLEGSKDALLCGF	STDGQCPEGYTCVKI	GIRNPDYGYTSFDTF	SWAFLALFR	LM	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	TMGYFYYLEGSKDALLCGF	STDGQCPEGYECVT	AGRNPDYGYTSFDT	FGWAFLALFR	LM	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	310	320	330	340	350	360
	370	380	390	400	410	420
Human	TQDYWENLYQQTLRAAGK	TYMIFFVVVIFLGSFY	LINLILA	VVAMAYEEQNQ	ANIEEAKQ	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	TQDYWENLYQQTLRAAGK	TYMIFFVVVIFLGSFY	LINLILA	VVAMAYEEQNQ	ANIEEAKQ	
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	370	380	390	400	410	420
	430	440	450	460	470	480
Human	KELEFQQMLDR	LKLKEQEEAIAAAA	EYTSIRR	SIMGLSES	SSETSKLSSK	SAKERRN
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	KELEFQQMLDR	LKLKEQEEAEVV	FIITNTKAKQVG	RIMGLSES	SSETSR	LSSKSAKERRN
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	430	440	450	460	470	480
	490	500	510	520	530	540
Human	RRKKKNQKKLSSGEEK	GDAEKLSKSE	SED	SIRRKSFHL	GVEGHRR	AHEKRLSTPNQSPLS
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
Mouse	RRKKKKQK-LSSGEEK	GDKLSKSG	SEES	SIRKSFHL	GVEGHHR	RAREKRLSTPNQSPLS
	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::	:::::::::::
	490	500	510	520	530	540

	550	560	570	580	590	600
Human	IRGSLFSARRSSRTSLFSFKGRGRDIGSETEFADDEHSIFGDNESRRGSLFVPHRPQERR :::.....:::.....:::.....:::.....:::.....:::.....:::.....:::.....:::					
Mouse	IRGSLFSARRSSRTSLFSFKGRGRDLGSETEFADDEHSIFGDNESRRGSLFVPHRPRERR 550 560 570 580 590 600					
	610	620	630	640	650	660
Human	SSNISQASRSPPMLPVNGKMHSAVDCNGVVSLVDGRSALMLPNGQLLPEGTTNQIHKKRR :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::					
Mouse	SSNISQASRSPPVLPVNGKMHSAVDCNGVVSLVDGPSALMLPNGQLLPEGTTNQMRKKRL 610 620 630 640 650 660					
	670	680	690	700	710	720
Human	CSSYLLSEDMLNDPNLRQRAMSRA SILTNTVEELEESRQKCPPWWYRFAHKFLIWCSPY :::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::					
Mouse	SSSYFLSEDMLNDPHLRQRAMSRA SILTNTVEELEESRQKCPPWWYRFAHTFLIWCSPY 670 680 690 700 710 720					
	730	740	750	760	770	780
Human	WIKFKKCIYFIVMDPFVDLAITICIVLNTLFMA MEHHPMTEEFKN VLAIGNLVFTGIFAA :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::					
Mouse	WIKFFKFIYFIVMDPFVDLAITICIVLNTLFMA MEHHPMTDEFKN VLAIGNLVFTGIFAA 730 740 750 760 770 780					
	790	800	810	820	830	840
Human	EMVLKLIAMDPYEYFQVGWNIFDSLIVTLSLVELFLADVEGL SVLRSFRLLRVFKLAKSW :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::					
Mouse	EMVLKLIAMDPYEYFQVGWNIFDSLIVTLSLVELFLADVEGL SVLRSFRLLRVFKLAKSW 790 800 810 820 830 840					
	850	860	870	880	890	900
Human	PTLNMLIKIIGNSVGNLTLVLAIIVFIFAVVGMQLFG KSYKECVCKINDDCTLPRWH :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::					
Mouse	PTLNMLIKIIGNSVGNLTLVLAIIVFIFAVVGMQLFG KSYKECVCKINENCKLPRWH 850 860 870 880 890 900					
	910	920	930	940	950	960
Human	MNDFFHSFLIVFRVLCGEWIETMWDCMEVAGQAMCLIVYMMVMVIGNLVNLFLALLLS :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::					
Mouse	MNDFFHSFLIVFRVLCGEWIETMWDCMEVAGQTMC LIVYMMVMVIGNLVNLFLALLLS 910 920 930 940 950 960					
	970	980	990	1000	1010	1020
Human	SFSSDNLTAEEDPDANNLQIAVTRIKKGINYVKQTLREFILKAFSKPKKISREIRQAED :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....::::::: :					
Mouse	SFSSDNLTAEEDTDANNLQIAVARIKRGINYVKQTLREFILKSFSKPKGSKDTKRTAD 970 980 990 1000 1010 1020					
	1030	1040	1050	1060	1070	1080
Human	LNTKKNEYISNHTLAEMSNKGHNFLKEKD KISGF GSSV DKHLMEDSDG QSF IHNPSLTVTV :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::::::.....:::					
Mouse	PNNKRENYISNRTLAEISKDHNFLKEKD KISGF GSSLDK SFMDENDYQSFIHNPSLTVTV 1030 1040 1050 1060 1070 1080					
	1090	1100	1110	1120	1130	1140
Human	PIAPGESDLENMNAEELSSDSDSEYSKVRLNRSSSECSTVDNPLPGE GE EEAEPMNSD :::::::.....:::::::.....:::::::.....:::::::.....:::::::.....::::::: :					
Mouse	PIAPGESDLENMNT EELSSDSDSDYSKERRNRSSSECSTVDNPLPGE -EEAEAE PINAD 1090 1100 1110 1120 1130 1140					

	1150	1160	1170	1180	1190	1200
Human	EPEACFTDGCVRRFSCCQVNIESGKGKIWWNIRKTCYK I VEH ^S WFESFIVL ^M ILLSSGAL ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	EPEACFTDGCVRRFPCCQVNIDSGKGKVWWTIRKTCYR I VEH ^S WFESFIVL ^M ILLSSGAL 1150 1160 1170 1180 1190 1200					
	1210	1220	1230	1240	1250	1260
Human	A FEDIYI E RKKTIK I ILEYADKIFTYIFILEMLLKWIAYGYKTYF T NAWCWLDFLIVDVS ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	A FEDIYI E KKKT I K I ILEYADKIFTYIFILEMLLKWVAYGYKTYF T NAWCWLDFLIVDVS 1210 1220 1230 1240 1250 1260					
	1270	1280	1290	1300	1310	1320
Human	L VTLVANTLGYS ^D LGPIKSLRTLRLPRLALS ^R FEGMRVVVNALIGAIPSIMNVLLVCL ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	L VTLVANTLGYS ^D LGPIKSLRTLRLPRLALS ^R FEGMRVVVNALIGAIPSIMNVLLVCL 1270 1280 1290 1300 1310 1320					
	1330	1340	1350	1360	1370	1380
Human	I FWLIFSI ^M GVNL ^F AGKFYECINTTDGSRF ^P ASQVPNRSEC ^F ALMNVSQNV ^R WKNLKVNF ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	I FWLIFSI ^M GVNL ^F AGKFYECVN ^T TDGSRF ^S V ^S QANRSEC ^F ALMNVS ^G NVR ^W KNLKVNF 1330 1340 1350 1360 1370 1380					
	1390	1400	1410	1420	1430	1440
Human	DNVGLGYL ^S LLQVATF K GW ^T IIMYAAVDSVNVDQ ^K PKY ^E YS L YMYIYFVVFIIFGSFFTL ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	DNVGLGYL ^S LLQVATF K GWMDIMYAAVDSVNNAQPIY ^E YNLYMYIYFVIFIIFGSFFTL 1390 1400 1410 1420 1430 1440					
	1450	1460	1470	1480	1490	1500
Human	N LFIGVII ^I DNFNQQKKKLGGQD I FMTEEQKKYYNAMKKLGSKKPQKPIPRPGNKIQ ^G CIF ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	N LFIGVII ^I DNFNQQKKKLGGQD I FMTEEQKKYYNAMKKLGSKKPQKPIPRPGNK ^F Q ^G CIF 1450 1460 1470 1480 1490 1500					
	1510	1520	1530	1540	1550	1560
Human	DLVTNQAFDISIMVLICLN ^M VTMMVEKEGQSQHMT E VLYWINVVFIILFTGECVLKLISL ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	DLVTNQAFDITIMVLICLN ^M VTMMVEKEGQTDYMS F VLYWINVVFIILFTGECVLKLISL 1510 1520 1530 1540 1550 1560					
	1570	1580	1590	1600	1610	1620
Human	RHYYFT V GWNIFDFVVVIISIVGMFLADLIETYFVSPTLFRVIRLARIGRILRLVKGAKG ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	RHYYFT V GWNIFDFVVVIISIVGMFLAEMIEKYFVSPTLFRVIRLARIGRILRLIKGAKG 1570 1580 1590 1600 1610 1620					
	1630	1640	1650	1660	1670	1680
Human	I RTLLFALMMSLP ^A LFNIGLLLFLVMFIYAIFGMSNFAYVK ^K EDGINDMFNFETFGNSMI ::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: ::::: :::::					
Mouse	I RTLLFALMMSLP ^A LFNIGLLLFLVMFIYAIFGMSNFAYVK ^K EA ^G INDMFNFETFGNSMI 1630 1640 1650 1660 1670 1680					

	1690	1700	1710	1720	1730	1740
Human	CLFQITTSAGWDGLLAPILNSKPPDCDPKKVHPGSSVEGDCGNP	SVGIFYFVSYIIISFL				

Mouse	CLFQITTSAGWDGLLAPILNSAPPDCDPKKVHPGSSVEGDCGNP	SVGIFYFVSYIIISFL				
	1690	1700	1710	1720	1730	1740
	1750	1760	1770	1780	1790	1800
Human	VVVNMYIAVILENSVATEESTEPLSEDDFEMFYEVWEKFDPDATQFIEFSKLSDFAAAL					

Mouse	VVVNMYIAVILENSVATEESTEPLSEDDFEMFYEVWEKFDPDATQFIEFCKLSDFAAAL					
	1750	1760	1770	1780	1790	1800
	1810	1820	1830	1840	1850	1860
Human	DPPLIAKPNKVQLIAMDLPMVSGDRIHCLDILFAFTKRVLGESGEMDSLRSQMEERFMS					

Mouse	DPPLIAKPNKVQLIAMDLPMVSGDRIHCLDILFAFTKRVLGESGEMDSLRSQMEERFMS					
	1810	1820	1830	1840	1850	1860
	1870	1880	1890	1900	1910	1920
Human	ANPSKVSYEPITTLKRKQEDVSATVIQRAYRRLRQNVKNISSIYIKDGDRDDLLNK					

Mouse	ANPSKVSYEPITTLKRKQEDVSATIIQRAYRRLRQNVKNISSIYIKDGDRDDLPNK					
	1870	1880	1890	1900	1910	1920
	1930	1940	1950	1960	1970	
Human	KDMAFDNVNENSSPEKTDATSSSTSPPSYDSVTKPDKEKYEQDRTEKEDKGKDSKESKK					

Mouse	EDIVFDNVNENSSPEKTDATASTISPSSYDSVTKPDQEKYETDKTEKEDKEKD--ESRK					
	1930	1940	1950	1960	1970	

Marker Position	Marker Name	Family 1	Family 2	Family 3	Caucasian control
159522955	D2S1353	140	156	het	
159663226	AC064843GT21	190	188	182	195/197
159880425	AC010093CA21	300	294		
160040465	D2S2370	88	88	98	91/93
160085712	D2S2190	227	227	221	221/221
160145161	AC008277AC32	329	317	333	317/317
160361795	AC009506TTA10	382	382	391	382/382
160612804	D2S156	176	188	170	190/190
160801752	AC080166GT14	172	164	164	166/172
161070953	AC092153GT17	229	235	229	227/231
161292227	AC131754TA13	250	250	244	244/266
161539521	AC096656GT26	220	200	186	215/219
161801813	AC073190AC24	224	228	218	210/224
162057568	AC009299CA22	165	167	165	165/169
162238623	AC009487GT17	192	198	196	196/204
162445601	AC062022GT23	279	279	281	279/285
162500221	AC062066GT17	258	258	258	256/258
162655195	AC096654AC20	293	297	293	295/295
162765717	AC092841TA26	170	174	168	184/186
162854397	AC008063TA16	319	317	319	309/325
162967073	AC007750TC18	232	226	232	228/228
163014807	AC007750CA14	124	126	124	124/124
163109452	AC010876CA20	265	251	239	
163279542	AC007740AAT	379	376	379	376/379
163351007	AC007740CA11	156	156	156	156/156
163397817	AC139719CTAT12	231	223	235	225/229
163536481	AC011900TG16	335	335	335	335/337
163569424	AC011900TTCC15	255	255	255	255/255
163659597	AC104822AC28	197	197	197	197/197
163745705	AC092632CA12	221	221	219	219/225
163779498	D2S2380	167	167	167	167/169
163843071	AC009439CA14	285	271	285	281/287
163882778	D2S354	178	178	180	174/178
164159039	AC079258GCA12	217	203	220	203/220
164478035	AC093727TG16	250	250	248	246/250
164593046	AC092684GT19	216	220	226	
164701834	AC016766TG21	242	256	242	229/255
164807829	AC016766AC17	167	161	167	163/163
164902460	AC016720ATCC13	289	289	293	289/293
165170192	AC110078TA12	204	202	206	202/204
165347711	AC107075AT22	219	211	209	205/209
165609555	AC019181AC22	251	253	237	237/237
165931794	D2S382	166	162	150	
166218588	AC011303AC17	252	250	246	246/246
166462626	AC019140AC22	199	211	211	209/211
166661437	D2S2330	160	156	164	164/168
166871148	AC010127TA12	306	302	288	316/316
166837116	AC107082GA17	212	214	207	
167075630	AC108146GA13	283	283	281	283/283
166,993,561	AC108146TA23	209	199	205	
167190727	AC074101AAT15	251	251	254	257/260
167277338	AC074101AT13	222	222	222	222/222
167199765	AC092538AAAT9	257	260	245	
167519277	AC104409AC20	237	249	237	249/257
167602054	AC104069GT20	151	163	153	151/161

167652758	AC104069AC13	236	238	236	238/238
167835805	D2S1379	153	153	157	153/153
168016947	AC093684TG19	203	193	199	203/203
168189596	AC073050TG23	320	300	300	300/300
168432902	AC074363TTA9	265	262	265	265/273
168684199	D2S2345	153	159	153	153/153
168911685	AC017069CA17	345	349	349	343/347
169099633	AC084371TG18	281	287	285	277/285
169372884	AC019086AC21	241	233	239	237/241
169567310	AC009475TA23	226	218	230	222/224
169609196	D2S1776	302	294	302	294/302
169734087	AC069137TG12	178	178	178	178/178
169951962	AC007556GT17	239	229	233	229/229
170219694	AC110780AC25	291	293	289	289/291
170471269	AC016772TG18	254	258	238	254/254
170712148	AC079151AC13	205	203	203	203/205
170838057	AC092641TG18	380	380	380	380/380
171191159	AC012594TG25	183	199	het	

Coding exon	Forward primer (5'-3')	Reverse primer (5'-3')	PCR product (bp)
1	CCTTCCTGGCAGGCAAAT	AAGCCAACAGAAAATGACCA	400
2	AGATCGGTTGATGACATTGG	CCAGAGTCTTCAGGTGCAA	377
3	TTCAAAGAGACAAAATAGTCTACAAGC	CTGGCAGGAAAAGGAAAGG	295
4	AAGTTATAAAGATTACATGGTGGTTG	ACCCCAGAGGTTGCTGTTA	283
5	GAAGCCCCAACGTAGAAAA	TCTTCTTCAAAAGATCAAAGTCA	444
6	AGGTTACTTAAGGTCTTGATTGA	AAAAGAGAGCAATGTTTAGCA	390
7	GGACCAGGCCTGAATTGTA	TGCAAACACTGACTGAACATTCTT	247
8	TCCCCCTATAGAAGAACCTTGA	GAGTTCCCTCATTCTCAAATAAA	353
9	CAGATTGCTCATGCCCTGTC	TCTAGCTGGAGAAGGCCAAG	386
10	TCATTGTGAAAGAAAACGATCA	AAGACATTTCTCTAGCATTCTGC	498
11	GCCAGTGGGTCAGTGGTAT	GCCATGCCTGAGCTATGTAA	488
12	TTGAACCCAGCAATCTAGGC	TGTGCCTATTAAGGTTGACCA	330
13	TTTCAATTAGAATGCCCTGACTGA	TGAAATGACAATGATGACAACAAAAA	396
14	TGATGAGCACTGACAGGACA	TGCAAAACCAAAGAAATACCC	467
15	TGCTTACCCCTTGAACAAAAA	CATCACAAATAATTCCACAGAGA	500
16	CCTGTCTCCCTATTCTACCC	TGCAATGTTAGAAGAACTTATGACAA	621
17	TTTTCATAACTTGCATGAGTCG	CGTTAAGACAACCCCCAGAA	291
18	TGAGGGAGTATCACAGAAAGCA	AGGTAAAACAACCTGCCATGA	348
19	CAGCTGGCCCATGTCAATA	TTCACAACACACAGTAAGAATAATCC	356
20	TGTTGAGTTGCTTTAGTGAGTT	TCATAATTCTACATACCCATTGTTT	300
21	TGCTGCATGGCATTTCTT	AAGAATAACCTATATCCTCGTCCAA	431
22	CAGACAAAACCTGTTATGGCTATT	TGAGTCCCAGGGCTTCAT	228
23	CCTCAACAATGCTATGCCCTC	TTGTTTCTGTGCAAAATGAAT	463
24	CTGTGTTGGAGACCCATGTT	TCCAGAAATTAAAGATGTGCATT	378
25	ATTCCTGACCAGGGTAAA	TTCAGCATATACTCCCTGAGCA	498
26A	TTGAATTATAAGAAATGAGTGACA	CAGTCGGTGGCTTACTGTT	476
26B	GACCTTGGCAACAGTATGATT	TTTGGAAAGGATTGACAGACA	577
26C	ACCGGATCCATTGTCTTGA	ACAGGCTGTAACAAATATCAAAAAA	500