The American Journal of Human Genetics, Volume *90*

Supplemental Data

De Novo Pathogenic *SCN8A* Mutation Identified by Whole-Genome Sequencing of a Family Quartet Affected by Infantile Epileptic Encephalopathy and SUDEP

Krishna R. Veeramah, Janelle E. O'Brien, Miriam H. Meisler, Xiaoyang Cheng, Sulayman D. Dib-Hajj, Stephen G. Waxman, Dinesh Talwar, Santhosh Girirajan, Evan E. Eichler, Linda L. Restifo, Robert P. Erickson, and Michael F. Hammer

Table S1. CNV Calls Found in Proband

Cytoband	Chr.	Start	Stop	Size	# Probes	Amplification	Gene Disrupting	Deletion	Gene Names	UniqueBP	SD Percent	Controls	NIMH ClinSeqCont
p22.3	1	86173346	86177248	3902	8	0.000	N	-0.917	COL24A1	3902	0.000	196	74
p21.1	1	103838243	103843177	4934	4	0.858	Y	0.000	RNPC3	4934	0.000	1	3
p13.3	1	110023792	110046030	22238	26	0.000	Y	-0.759	GSTM2 GSTM1	1	99.996	0	24
q24.2	1	167496916	167508082	11166	3	0.000	N	-1.094	NME7	11166	0.000	878	146
q31.3	1	195003634	195066067	62433	14	0.000	Y	-0.821	CFHR3 CFHR1	3099	95.036	51	16
q44	1	246686926	246715661	28735	9	0.000	Y	-0.684	OR2T3	0	100.000	0	7
q44	1	246805321	246857696	52375	4	6.661	Y	0.000	OR2T10 OR2T11	39365	24.840	253	10

q23.1	10	82202053	82206496	4443	7	0.858	N	0.000	TSPAN14	4443	0.000	0	16
q23.2	10	88688300	88695731	7431	12	0.529	Y	0.000	MMRN2	7431	0.000	0	2
q26.3	10	134882129	134897524	15395	11	0.615	Y	0.000	KNDC1 UTF1	15395	0.000	0	4
p15.4	11	5742276	5760317	18041	3	0.000	Y	-1.064	OR52N5	18041	0.000	371	39
q11	11	55124530	55167675	43145	4	1.038	Y	0.000	OR4C11 OR4P4	43145	0.000	147 9	134
q24.31	12	122317996	122321039	3043	3	0.897	N	0.000	CDK2AP1	3043	0.000	0	0
q32.3	13	99433142	99437206	4064	28	0.836	Y	0.000	ZIC2	4064	0.000	0	22
q34	13	113545189	113550676	5487	6	0.733	Y	0.000	GAS6	5487	0.000	0	7
q12	14	28305828	28308789	2961	20	0.863	Y	0.000	FOXG1	2961	0.000	0	9
q32.33	14	105280323	105584208	303885	24	0.997	N	0.000	KIAA0125 ADAM6	173090	43.041	0	0
q11.2	15	19328456	19964379	635923	83	0.648	Y	0.000	POTEB LOC727924 OR4M2 OR4N4 LOC650137	165775	73.932	12	0
q11.2	15	20908786	20936300	27514	37	0.526	N	0.000	HERC2P2	0	100.000	0	0
q13.1	15	27898898	27904843	5945	12	0.705	Y	0.000	TJP1	5945	0.000	0	20
q13.3	15	29404344	29410782	6438	12	0.623	Y	0.000	KLF13	6438	0.000	0	11
q13.3	15	29561208	29565065	3857	7	1.231	Y	0.000	OTUD7A	3857	0.000	0	22
q24.1	15	71129666	71134376	4710	7	0.614	Y	0.000	NEO1	4710	0.000	0	6
q24.1	15	72859270	72866755	7485	10	0.577	N	0.000	CSK	7485	0.000	0	0
q24.2	15	74136873	74142220	5347	8	0.688	N	0.000	C15orf27	5347	0.000	0	0
q24.3	15	74671426	74682436	11010	7	3.208	N	0.000	SCAPER	11010	0.000	39	222
q24.3	15	74678547	74680538	1991	3	5.501	N	0.000	SCAPER	1991	0.000	483	14
q24.3	15	75115608	75121278	5670	10	1.295	Y	0.000	PSTPIP1	5670	0.000	1	1
q24.3	15	75497657	75501223	3566	5	0.811	N	0.000	HMG20A	3566	0.000	0	1
q25.2	15	81664789	81669529	4740	9	0.554	Y	0.000	HDGFRP3	4740	0.000	0	20
q25.2	15	82332469	82334904	2435	5	1.029	N	0.000	ADAMTSL3	2435	0.000	3	61
q25.3	15	83090420	83095473	5053	7	0.735	N	0.000	ZNF592	5053	0.000	0	0
q26.1	15	88196414	88200643	4229	9	0.769	N	0.000	AP3S2	4229	0.000	0	0

q26.2	15	94673824	94676323	2499	5	1.791	N	0.000	NR2F2	2499	0.000	0	7
p13.3	16	937412	941075	3663	14	0.625	N	0.000	LMF1	3663	0.000	0	0
p13.3	16	2122430	2188753	66323	13	0.655	Y	0.000	PKD1 RAB26 SNORD60 TRAF7 CASKIN1	61486	7.293	3	6
p13.11	16	14956052	14992699	36647	8	0.542	Y	0.000	PDXDC1	10671	70.882	9	0
p12.1	16	21925200	21928591	3391	8	1.002	Y	0.000	C16orf52	3391	0.000	0	23
q12	17	32829927	32832842	2915	6	5.433	N	0.000	ACACA	2915	0.000	305	170
q12	17	32839956	32841975	2019	3	1.393	Y	0.000	ACACA TADA2L	2019	0.000	0	12
q12	17	32853093	32855815	2722	4	0.000	N	-0.895	TTADA2L	2722	0.000	0	68
q12	17	32923129	32925076	1947	3	0.859	N	0.000	DUSP14	1947	0.000	0	13
q21.1	17	35470593	35477090	6497	4	0.755	N	0.000	THRA THRA	6497	0.000	0	0
q21.31	17	40653853	40657215	3362	3	0.949	Y	0.000	FMNL1	3362	0.000	0	87
q21.31	17	40945119	40953419	8300	20	0.000	N	-0.848	LRRC37A4	4	99.952	22	63
q21.31	17	41521344	41665208	143864	206	0.000	Y	-0.612	KIAA1267	103942	27.750	9	7
q21.31	17	41526358	41568739	42381	59	0.000	Y	-1.083	KIAA1267	29400	30.629	0	9
q23.2	17	56832715	56842366	9651	3	0.863	Y	0.000	TBX2	9651	0.000	0	1
q24.2	17	63419284	63422115	2831	3	1.117	Y	0.000	C17orf58	0	100.000	0	0
p12	19	20399500	20493601	94101	5	0.837	N	0.000	ZNF826	92415	1.792	418	15
q13.42	19	59666684	59670297	3613	3	1.246	Y	0.000	LENG9 CDC42EP5	3613	0.000	0	1
q13.42	19	60962294	60975015	12721	4	0.723	Y	0.000	RFPL4A	1076	91.542	0	0
p24.3	2	15998302	16002326	4024	16	0.860	Y	0.000	MYCNOS MYCN	4024	0.000	0	2
q11.2	2	97507449	97522553	15104	4	0.755	Y	0.000	ANKRD36B	0	100.000	0	4
q13	2	110047302	110088127	40825	13	1.005	N	0.000	LIMS3- LOC440895	0	100.000	1	1
q13	2	110590285	110631127	40842	13	1.004	N	0.000	LIMS3- LOC440895	0	100.000	1	1

q24.2	2	162396228	162404701	8473	3	0.920	Y	0.000	SLC4A10	8473	0.000	0	1
q13.32	20	56896129	56901638	5509	15	0.501	Y	0.000	GNAS	5509	0.000	0	35
q11.21	22	17515887	17524197	8310	11	0.785	Y	0.000	GSC2	8310	0.000	0	3
q11.21	22	18127733	18135675	7942	15	0.751	Y	0.000	TBX1 TBX1 TBX1	7942	0.000	1	44
q11.21	22	18447113	18448872	1759	3	1.456	N	0.000	DGCR8	1759	0.000	0	7
q11.21	22	19121360	19124064	2704	4	0.814	Y	0.000	SCARF2	2704	0.000	0	33
q11.22	22	20717615	21293141	575526	124	0.779	Y	0.000	VPREB1 LOC96610 ZNF280B ZNF280A PRAME LOC648691	485159	15.702	16	1
q11.23	22	22677759	22725505	47746	11	0.000	Y	-0.791	LOC391322 GSTT1 GSTTP2	24593	48.492	578	80
q13.33	22	48838328	48841300	2972	6	0.872	N	0.000	MLC1 MLC1	2972	0.000	0	6
q13.33	22	49129078	49131925	2847	11	0.776	N	0.000	SAPS2	2847	0.000	33	1
q21.3	3	131271983	131287554	15571	4	0.949	Y	0.000	LOC644974	0	100.000	7	13
q23	3	144246990	144250641	3651	3	0.818	N	0.000	SR140	3651	0.000	0	1
q25.1	3	152961950	152996321	34371	3	0.734	N	0.000	LOC201651	34371	0.000	0	0
q29	3	197118167	197124124	5957	9	0.550	N	0.000	TNK2	5957	0.000	0	6
q29	3	197778183	197782079	3896	7	0.734	Y	0.000	WDR53 FBXO45	3896	0.000	0	11
q29	3	198177846	198183014	5168	7	0.628	N	0.000	PIGZ	5168	0.000	0	32
q29	3	198210680	198216872	6192	10	0.774	Y	0.000	MFI2	6192	0.000	0	1
q29	3	198728848	198730576	1728	10	0.611	N	0.000	BDH1	1728	0.000	0	7
p16.1	4	7499241	7508332	9091	8	0.614	N	0.000	SORCS2	6353	30.118	0	0
q13.2	4	69057535	69196107	138572	21	1.914	Y	0.000	UGT2B17 UGT2B15	52159	62.360	201	174
q13.2	4	69069451	69085586	16135	3	4.462	N	0.000	UGT2B17	0	100.000	0	12
q13.2	4	69113211	69117459	4248	3	0.742	Y	0.000	UGT2B17	0	100.000	0	15
p25.3	6	204118	321090	116972	19	0.000	Y	-0.779	DUSP22	116972	0.000	18	25

p25.3	6	1556514	1558506	1992	13	1.038	Y	0.000	FOXC1	1992	0.000	0	7
p21.33	6	29962649	30001190	38541	4	0.000	N	-1.008	HLA-H HCG2P7 HCG4P6	24819	35.604	463	4
q13	6	72053677	72057537	3860	5	1.057	Y	0.000	OGFRL1	3860	0.000	0	1
p21.1	7	19121452	19122466	1014	17	0.782	N	0.000	TWIST1	1014	0.000	0	0
q11.21	7	64212798	64272128	59330	11	0.000	N	-0.615	INTS4L1	0	100.000	0	5
q21.12	7	86524801	86529372	4571	7	0.808	Y	0.000	KIAA1324L	4571	0.000	0	3
q32.2	7	130067686	130070028	2342	3	1.578	Y	0.000	KLF14	2342	0.000	0	2
q34	7	142138269	142202474	64205	27	0.000	Y	-0.643	PRSS1 TRY6 PRSS2	24349	62.076	0	19
p23.1	8	7099218	7139381	40163	21	0.000	Y	-0.552	LOC349196	0	100.000	0	165
q24.11	8	119191734	119192678	944	3	1.407	Y	0.000	EXT1	944	0.000	0	17
p24.3	9	338834	346279	7445	3	0.000	N	-0.969	DOCK8	7445	0.000	0	0
q22.32	9	97306910	97319183	12273	6	0.697	Y	0.000	PTCH1	12273	0.000	0	5
p22.33	X	252640	254064	1424	5	1.183	N	0.000	PPP2R3B	1424	0.000	0	1
p11.32	Y	252840	254064	1224	4	1.409	N	0.000	PPP2R3B	1224	0.000	0	1

Table S2. Candidate Variants under a Homozygote Recessive Disease Model

Recessive Type	Chr.	Position	Ref.	Variant	KG Gene Name	KG_mRNA Transcript	KG Coding Change	KG Protein Change	Father	Mother	Sibling	Proband	dbSNP 132 rs#	Polyphen 2 Score	1000 Genome Freq	Present CGI	Autworks Xref
Simp	1	33960715	C	T	ZSCAN2	uc001bxj.3 uc009vui.2	c.C2771T c.C2768T	p.T924I p.T923I	01	01	01	11	?	0.48	?	N	
Simp	3	111901062	A	T	SLC9A10	uc010hqc.2 uc011bhu.1 uc003dyu.2	c.T2423A c.T356A c.T2567A	p.I808N p.I119N p.I856N	01	01	01	11	146697807	0.18	?	N	
Simp	3	113045485	C	A	WDR52	uc003ead.1	c.G1866T	p.W622C	01	01	01	11	?	?	?	N	
Simp	19	45206933	G	A	CEACAM16	uc010xxd.1 uc002ozq.2,	c.G352A c.G529A	p.E118K p.E177K	01	01	00	11	183860695	?	0.002	N	
Simp	1	33236824	C	T	KIAA1522	uc010ohm.1 uc001bvv.2 uc001bvu.1	c.C1900T c.C1867T c.C2044T	p.P634S p.P623S p.P682S	01	01	0N	11	?	0.50	?	N	
Comp	2	206592624	C	T	NRP2	uc002vav.2	c.C1000T	p.R334C	00	01	01	01	114144673	1.00	?	N	Ataxia, Autism, Epilepsy, Hypoxia, Stroke,
	2	206605378	C	T	NRP2	uc002vav.2	c.C1282T	p.R428W	01	00	00	01	139711818	1.00	?	N	Ataxia, Autism, Epilepsy, Hypoxia, Stroke,
Comp	15	54306012	C	G	UNC13C	uc002ack.2,	c.C912G	p.D304E	00	01	00	01	149448818	?	0.008	N	
	15	54919253	T	C	UNC13C	uc002acm.2 uc002ack.2,	c.T350C c.T6587C	p.V117A p.V2196A	01	00	00	01	146433220	?	0.004	N	