

Supplementary Appendix

This appendix has been provided by the authors to give readers additional information about their work.

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Supplementary Appendix to:

Whole-Genome Sequencing and *SH3TC2* Mutations Causing Charcot-Marie-Tooth Neuropathy

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1. Supplementary Methods

The Applied Biosystems SOLiD™ System DNA sequencing platform was employed to sequence genomic DNA, according to manufacturer's specifications. Primary sequence analysis consisting of image analysis and base-calling was performed using the standard SOLiD primary analysis pipeline for the 35bp fragment and 25x25bp mate-pair data, mapping and variant calling analysis was with the AB-provided offline analysis suite "corona_lite". For the 50 x 50 mate-pair data the Applied Biosystems Hidra pipeline was employed.

Mapping parameters for the 35 bp reads allowed for up to 2 mismatches in 25bp, and 3 mismatches in 35bp. The mapping did not allow for insertions or deletions. Variant calls were generated in a high sensitivity/low stringency mode where no coverage cut-offs were imposed for variant calls. Also, multi-allelic sites are rejected as sequencing noise, or mismapping. In making SNP calls, multiple reads with the same start point and direction are treated as a single read to avoid errors from oversampling of duplicate read alleles.

To maximize the sequence yield of the 50x50bp mate-pair data mapping was performed in 28 different iterations with reads trimmed back to progressively shorter lengths (in 5bp increments from 50bp down to 25bp) and lower stringency (from 0 to 4 mismatches)("progressive mapping"). For any given read the longest mapping with the highest stringency (longest length, fewest mismatches) was kept as the best alignment. Mapping was carried out with mapreads (the mapping tool provided by the vendor as part of their "corona_lite" analysis suite) in parallel for the different iterations and read data was merged at the end.

CNV analysis was performed with the AB single-sample CNV tool that uses read coverage corrected for mapping and amplification efficiency to identify regions likely to have increased or decreased copy number variation. Analysis of the high-density random array data used custom tools developed by Applied Biosystems and included modified bead-finding and progressive mapping using "corona_lite". Only publicly available (www.solidsoftwaretools.com) vendor software was used for the sequence data analysis presented in this study.

Supplementary Figure 1

A

G²3112 32 31302030230100112
A³112 10 313020302301001123
T² 32 313020302301001123103
T⁰22223332320200123123131002113312031222003223112 32 31302030230100112310313013002133011002202030131
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AAGAGATATCGAAGGGTCGTCGTACCCTGTATGAATGAGAAATCTACAGCTACGGAATTCGGTTTGTCTGTTACGGTAAAGTATTGTTTCTCCGGTAC
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13312031222003223112 32 313020302301^T
003223112 32 3130203023010^T

B

C²2223332320200123123131002113312031222003223112 10 3
C²020012312313100211331203222003223112 00 313020302
C⁰112312313100211331203222003223112 32 313020122301
C⁰123123131002113312031222003223112 10 3130203023010
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C¹31002113312031222003223112 10 31302030230100112310
C¹3312031222003223112 10 31
C¹3312031222003223112 10 313020302301001123103130130
C¹3312031222003223112 10 31302030212100112.....
C¹3312031222003223112 10 313020302301001123103130130
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G²3112 32 31302030230100112
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T²313020302301001123103130130021330110022020301...
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.....123123101002113310031222003223112 1^T
22223332320200123123131002123312031222003223112 32^T
2320200123123131002113312031222003223112 32 3130203^T
20200123123131002113312031222003223112 10 313020302^C
123123131002113312031222003223112 32 31302030230100^T
131002113312031222003223112 10 31302030230100112310^T
31002113312031222003223112 10 313020302301001123103^A
31002113312031222003223112 10 313020302301001123103^A
31012113312031222003223112 32 313020302301001023203^A
11002113312031222003223112 32 313020302301001123103^A
31002113312031222003223112 32 313020302301001123103^A
1002113312031222003223112 10 3130203023010011231031^C
02113312031222003223112 1^T
113312031222003223112 32 3^A
13312031222003223112 32 313020302301^T
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003223112 32 3130203023010^T
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3112 10 3130203023010011231031330300213301100220203^G

C

^A3212022120012011133311002012011100
^A202212001201113301100201
^G0221200120111330110020120111003321
^C2223100023313011112211020202113321202212001201333301100201201110033213223112211120102322221133202
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GAGATGGGGATACGGTGTGAGTGGAAGGACATAGTCCTCAGGGTCCATATAACAAAGGTCCACAAATAGTAGATGTCTGTGAACCTAGAGACATAGGA
2223100023313011112211020202113321202212001201333301100201201110033213223112211120102322221133202^A
211332120221200120133330^A
1133212022120012011133211002012011^C
1133212022100012011133011002012011^C

D

...2231000233130111122110202021133212022120012013
^C0022331301111221102020211332123221200120111330110
^T13011112211020201113311202212001101333301100....
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^T3321202212001201333301100201201110033213223112211
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^A3212022120012011133311002012011100
^C120221200120133330110020
^A202212001201113301100201
^A2022120012011133011002012011100332132231122111201
^A2022120012011133011002012011100332132231122111201
^G0221200120111330110020120111003321
^T2001201333301100201201110033213223112211120102322
^C2223100023313011112211020202113321202212001201333301100201201110033213223112211120102322221133202
CTCTACCCCTATGCCACACTCACCTTCCGTATCAGGAGTCCCAGGTAATTTGTTTCCAGGTGTTTATCATCTACAGACACTTGGATCTCTGTATCCT
GAGATGGGGATACGGTGTGAGTGGAAGGACATAGTCCTCAGGGTCCATATAACAAAGGTCCACAAATAGTAGATGTCTGTGAACCTAGAGACATAGGA
2223100023313011112211020202113321202212001201333301100201201110033213223112211120102322221133202^A
.....211332120221200120133330^A
0233110111222110202021033212022120012013333011002^G
0233130111122110202021133212022120012013333011002^G
1122110202021133212022120012011133011002012011100^A
1122110202021133212022120012013333011002012011100^A
2211020202113321202212001201333301100201201110033^A
211332120221200120133330^A
2113321202212001201113301100201201110033213223112^C
1133212022120012011133211002012011^C
1133212022100012011133011002012011^C
113321202212001201113301^C
321202212001201113301100^A
221200120111330110020120^C
212001201333301100201201^A
2120012011133011002012011100332132231122111201023^A
20012011133011002012011^A
1333301100201101110033213223112211120102322221133^A

Supplementary Figure 1. Raw read coverage at key SNP positions for initial partial-coverage analysis (7.7x) compared to final full-coverage analysis (29.6x). Both strands of the reference genome are shown in sequence space in the middle, with the color-space renderings above and below. Reference bases of interest are shown in grey. Sequence reads are shown in color-space with reads mapping in the forward direction (plus strand) above the reference, and reads mapping in the reverse direction (minus strand) shown below. Reads that agree with the reference are highlighted in green at the SNP position, and reads that show the variant allele are highlighted in red.

A. Read coverage at chr5:148,386,628 for original ~7x coverage. Initial call was reference homozygote; the single variant read was rejected as an error. B. Full read coverage at the same position for the full ~30x coverage. The final call was a heterozygote with roughly equal numbers of reference and variant alleles seen in the full-coverage read stack. C. Read coverage at chr5:148,402,474 for the original ~7x coverage. The initial call was variant homozygote; the single reference read was rejected as an error. D. Full read coverage at the same position for the full ~30x coverage. The final call was a heterozygote with roughly equal numbers of reference and variant alleles seen in the final read stack.

Supplementary Table 1. CNVs found in neuropathy subject's genome

Chr	Cytoband	Start	Stop	Size	Gain/Loss	NimbArray	Sequencing	DGV	Gene Names
chr1	p36.33	533,728	558,620	24,892	Loss	-	-	√	
chr1	p36.32	2,574,355	2,621,282	46,927	Gain	-	Yes	√	
chr1	p36.21	12,822,189	12,835,212	13,023	Gain	-	-	√	LOC649330, HNRNPCL1
chr1	p36.13	16,882,564	16,923,826	41,262	Loss	-	Yes	√	ESPFP
chr1	p36.13	17,080,364	17,152,572	72,208	Loss	-	-	√	CROCC
chr1	p36.11	25,465,575	25,531,629	66,054	Loss	Yes	Yes	√	RHD
chr1	p21.1	103,965,274	104,024,325	59,051	Loss	Yes	-	√	AMY2A, AMY1A, AMY1A, AMY1C, AMY1B
chr1	p13.1	116,970,390	116,999,657	29,267	Gain	Yes	-	√	IGSF3
chr1	p12	120,358,263	120,373,080	14,817	Gain	-	-	√	NOTCH2
chr1	p12	120,394,372	120,404,215	9,843	Gain	-	-	√	NOTCH2
chr1	p12	120,415,520	120,424,104	8,584	Gain	-	-	√	
chr1	q21.1	143,383,856	143,411,618	27,762	Gain	-	-	√	NBF20
chr1	q21.1	143,686,863	143,791,202	104,339	Gain	-	Yes	√	PDE4DIP
chr1	q21.1	143,946,161	143,997,989	51,828	Gain	-	Yes	√	NOTCH2NL
chr1	q21.1	147,308,962	147,497,322	188,360	Loss	Yes	Yes	√	
chr1	q21.3	150,824,358	150,853,324	28,966	Loss	-	Yes	√	LCE3C, LCE3B
chr1	q23.3	159,822,841	159,896,564	73,723	Loss	-	-	√	FCGR3C, HSPA7, FCGR3B
chr1	q31.1	185,985,437	185,988,338	2,901	Loss	Yes	-	√	
chr1	q31.3	194,990,451	195,089,902	99,451	Gain	-	-	√	CFHR3, CFHR1
chr1	q32.2	208,144,663	208,151,354	6,691	Loss	-	Yes	√	
chr2	p25.3	4,194,978	4,200,314	5,336	Loss	-	Yes	√	
chr2	p24.3	13,435,727	13,470,771	35,044	Gain	Yes	-	√	
chr2	p24.3	13,502,161	13,513,640	11,479	Gain	Yes	-	√	
chr2	p22.3	35,830,721	35,843,953	13,232	Loss	-	Yes	√	
chr2	p16.3	52,604,016	52,638,315	34,299	Loss	Yes	Yes	√	
chr2	p11.2	88,914,081	88,941,307	27,226	Gain	-	-	√	
chr2	p11.2	88,989,954	89,111,166	121,212	Gain	-	-	√	
chr3	p11.2	89,139,401	89,325,257	185,856	Gain	-	-	√	
chr2	p11.2	89,617,972	89,680,223	62,251	Gain	-	-	√	
chr2	p11.2	89,773,295	89,908,726	135,431	Gain	-	-	√	
chr2	q12.2	106,247,167	106,249,746	2,579	Loss	Yes	Yes	√	
chr2	q14.3	125,510,292	125,515,958	5,666	Loss	Yes	Yes	NO	
chr2	q14.3	126,160,881	126,166,108	5,227	Loss	-	-	√	
chr2	q22.3	146,579,846	146,592,834	12,988	Gain	Yes	-	√	
chr2	q24.3	167,734,684	167,782,652	47,968	Loss	Yes	Yes	NO	XIRP2
chr2	q32.3	194,397,791	194,401,129	3,338	Gain	-	-	√	
chr2	q33.3	208,059,576	208,066,304	6,728	Gain	-	-	√	
chr2	q36.3	226,874,648	226,876,563	1,915	Loss	Yes	-	√	
chr3	p26.1	6,627,047	6,628,889	1,842	Loss	Yes	-	√	
chr3	p24.1	26,409,589	26,414,428	4,839	Gain	-	-	√	
chr3	p22.2	37,955,930	37,961,169	5,239	Loss	Yes	-	√	CTDSPL
chr3	p14.1	65,165,476	65,189,354	23,878	Loss	Yes	Yes	√	
chr3	p14.1	68,824,433	68,827,577	3,144	Loss	-	-	√	
chr3	p12.3	75,840,737	75,853,111	12,374	Gain	-	Yes	√	
chr3	p12.3	75,871,755	75,883,979	12,224	Gain	-	Yes	√	ZNF717
chr3	q12.1	100,426,538	100,431,236	4,698	Loss	Yes	-	√	
chr3	q21.3	129,863,290	129,885,838	22,548	Gain	Yes	Yes	√	
chr3	q24	147,868,241	147,871,099	2,858	Gain	-	Yes	√	
chr3	q26.1	163,996,796	164,006,944	10,148	Gain	-	Yes	√	
chr3	q26.1	164,030,531	164,108,709	78,178	Gain	Yes	Yes	√	
chr3	q36.32	180,046,787	180,055,376	8,589	Gain	-	-	NO	
chr3	q29	194,358,882	194,365,707	6,825	Gain	Yes	-	√	
chr4	p16.1	9,821,327	9,842,703	21,376	Gain	Yes	Yes	√	
chr4	p16.1	10,006,532	10,009,259	2,727	Loss	-	Yes	√	
chr4	p15.2	25,901,935	25,904,465	2,530	Loss	-	Yes	√	
chr4	p15.1	34,457,648	34,502,838	45,190	Gain	-	-	√	
chr4	p13	40,922,497	40,926,391	3,894	Loss	-	-	√	
chr4	p13	42,459,138	42,462,922	3,784	Loss	Yes	-	√	
chr4	q13.1	63,817,412	63,834,857	17,445	Loss	Yes	Yes	√	

Supplementary Table 1. CNVs found in neuropathy subject's genome

Chr	Cytoband	Start	Stop	Size	Gain/Loss	NimbArray	Sequencing	DGV	Gene Names
chr4	q13.2	69,046,097	69,173,429	127,332	Gain	Yes	-	√	UGT2B17
chr4	q13.2	69,428,281	69,437,322	9,041	Gain	-	-	√	
chr4	q13.2	70,162,113	70,264,574	102,461	Gain	-	-	√	UGT2B28
chr4	q13.2	70,317,017	70,323,600	6,583	Gain	-	-	√	
chr4	q24	107,278,051	107,281,469	3,418	Gain	-	-	√	TBCKL
chr4	q25	108,731,380	108,734,061	2,681	Loss	-	-	√	
chr4	q32.3	169,047,365	169,228,589	181,224	Gain	-	-	√	
chr4	q34.1	172,613,413	172,615,132	1,719	Loss	-	Yes	√	
chr4	q34.1	173,662,973	173,666,131	3,158	Loss	-	Yes	√	GALNTL6
chr4	q35.1	185,395,604	185,404,326	8,722	Gain	-	-	√	
chr4	q35.2	187,331,981	187,333,678	1,697	Gain	-	-	√	
chr5	p15.33	796,917	819,095	22,178	Gain	-	-	√	
chr5	p15.33	848,744	877,163	28,419	Gain	-	-	√	ZDHHC11
chr5	p15.32	5,736,140	5,767,243	31,103	Gain	-	Yes	√	
chr5	p15.2	12,866,038	12,873,111	7,073	Loss	Yes	-	√	
chr5	p14.1	26,829,830	26,835,669	5,839	Loss	-	Yes	√	
chr5	p14.1	27,555,061	27,564,501	9,440	Loss	Yes	Yes	√	
chr5	q31.3	140,203,808	140,215,068	11,260	Gain	-	-	√	PCDHA1, PCDHA1, PCDHA2, PCDHA3, PCDHA4, PCDHA5, PCDHA6, PCDHA7, PCDHA8, PCDHA9, PCDHA9
chr5	q33.1	150,185,202	150,201,204	16,002	Loss	Yes	Yes	√	
chr5	q35.3	180,311,280	180,363,151	51,871	Gain	-	-	√	LOC646227, BTNL3
chr6	p25.3	202,063	239,298	37,235	Loss	Yes	-	√	DUSP22
chr6	p21.33	29,962,849	30,012,493	49,644	Gain	-	-	√	HLA-H, HCG2P7, HCG4P6
chr6	p21.33	31,344,074	31,348,964	4,890	Loss	-	-	√	HLA-C
chr6	p21.32	32,564,884	32,576,892	12,008	Loss	-	Yes	√	
chr6	p21.32	32,593,151	32,609,584	16,433	Loss	-	Yes	√	HLA-DRB5
chr6	p21.32	32,637,654	32,649,896	12,242	Gain	-	Yes	√	
chr6	p21.32	32,711,957	32,731,131	19,174	Gain	-	Yes	√	HLA-DQA1
chr6	p12.1	54,037,199	54,042,793	5,594	Gain	-	-	√	C6orf142
chr6	q12	66,208,637	66,216,847	8,210	Gain	-	-	√	EYS
chr6	q12	67,065,465	67,101,414	35,949	Loss	Yes	Yes	√	
chr6	q14.1	77,499,382	77,509,119	9,737	Gain	Yes	-	√	
chr6	q14.1	79,025,643	79,091,909	66,266	Gain	Yes	-	√	
chr6	q14.1	81,341,711	81,348,229	6,518	Loss	Yes	Yes	√	
chr6	q16.3	103,850,838	103,857,715	6,877	Loss	-	Yes	√	
chr6	q27	170,852,008	170,881,324	29,316	Loss	-	-	√	
chr7	p22.2	3,578,949	3,584,193	5,244	Gain	-	-	√	SDK1
chr7	p15.2	26,103,844	26,108,428	4,584	Gain	Yes	-	√	
chr7	p12.1	51,562,878	51,565,523	2,645	Loss	-	Yes	√	
chr7	q11.23	71,910,098	71,912,133	2,035	Loss	Yes	-	√	TYW1B
chr7	q11.23	75,984,055	75,987,589	3,534	Loss	-	Yes	√	UPK3B
chr7	q21.11	85,464,873	85,470,821	5,948	Gain	-	-	NO	
chr7	q33	133,437,464	133,447,927	10,463	Gain	Yes	Yes	√	
chr7	q34	141,700,432	141,714,227	13,795	Loss	Yes	Yes	√	
chr7	q35	143,528,075	143,692,010	163,935	Loss	-	-	√	OR2A1, OR2A42, OR2A9P, OR2A20P, OR2A7, OR2A1, ARHGEF5
chr7	q36.2	154,024,885	154,029,188	4,303	Gain	-	-	√	DPP6, DPP6, DPP6
chr7	q36.3	158,811,268	158,815,216	3,948	Gain	-	-	√	
chr8	p23.3	17,264	44,424	27,160	Loss	-	-	√	
chr8	p23.3	586,165	588,585	2,420	Gain	Yes	-	√	
chr8	p23.2	2,526,661	2,539,247	12,586	Gain	-	-	√	
chr8	p23.1	6,824,697	6,859,970	35,273	Loss	Yes	-	√	DEFA1, LOC728358, DEFA1, LOC728358
chr8	p23.1	11,250,976	11,254,937	3,961	Gain	-	-	√	TDH
chr8	p23.1	12,022,689	12,035,685	12,996	Gain	-	Yes	√	FAM66D, DUB3
chr8	p23.1	12,275,885	12,286,448	10,563	Loss	-	Yes	√	FAM66A
chr8	p12	32,800,775	32,810,135	9,360	Loss	-	Yes	NO	
chr8	q12.1	58,082,382	58,090,214	7,832	Loss	-	Yes	NO	
chr8	q12.1	58,277,102	58,285,421	8,319	Gain	-	Yes	√	
chr8	q22.1	96,946,000	96,948,094	2,094	Loss	Yes	-	√	
chr8	q23.3	115,705,054	115,711,789	6,735	Loss	Yes	-	√	
chr8	q24.3	144,771,984	144,785,852	13,868	Gain	-	-	√	

Supplementary Table 1. CNVs found in neuropathy subject's genome

Chr	Cytoband	Start	Stop	Size	Gain/Loss	NimbArray	Sequencing	DGV	Gene Names
chr9	p21.3	22,486,980	22,492,662	5,682	Gain	-	-	√	
chr9	p21.3	23,353,267	23,365,998	12,731	Loss	-	Yes	√	
chr9	p11.2	43,550,770	43,562,159	11,389	Gain	-	-	√	
chr9	q31.1	103,754,775	103,763,361	8,586	Loss	Yes	Yes	√	
chr9	q34.3	138,787,852	138,794,546	6,694	Loss	-	Yes	√	
chr10	p15.1 - p14	6,695,343	6,702,493	7,150	Gain	-	-	√	
chr10	p11.23	31,286,901	31,291,725	4,824	Gain	-	-	√	ZNF438, ZNF438, ZNF438
chr10	p11.21	37,500,439	37,508,602	8,163	Loss	Yes	-	√	ANKRD30A
chr10	p11.21	37,512,212	37,522,801	10,589	Loss	Yes	-	√	ANKRD30A
chr10	q11.22	46,321,769	46,834,292	512,523	Loss	-	Yes	√	SYT15, SYT15, GPRIN2, PPYR1, LOC728643, ANXA8, ANXA8L1, FAM25C, FAM25G, FAM25B, LOC642826, ANTXRL, ANXA8L2, FAM21B
chr10	q11.22	47,057,097	47,172,593	115,496	Loss	-	-	√	ANTXRL
chr10	q11.22	47,732,038	47,746,367	14,329	Loss	-	-	√	
chr10	q21.1	56,118,633	56,138,826	20,193	Loss	-	-	√	PCDH15, PCDH15, PCDH15
chr10	q26.13	124,335,715	124,340,556	4,841	Gain	-	-	√	DMBT1
chr11	p15.4	4,926,383	4,932,273	5,890	Loss	Yes	Yes	√	
chr11	p15.4	5,742,476	5,765,192	22,716	Gain	-	-	√	OR52N5
chr11	p15.1	18,050,268	18,058,426	8,158	Loss	Yes	Yes	NO	SAAL1
chr11	p15.1	18,906,505	18,920,218	13,713	Gain	-	-	√	MRGPRX1
chr11	p14.3	24,400,064	24,407,975	7,911	Gain	-	-	√	
chr11	p14.1	28,964,993	28,968,003	3,010	Gain	-	-	√	
chr11	p11.2	48,558,648	48,560,338	1,690	Loss	-	Yes	√	
chr11	p11.12	49,666,382	49,713,606	47,224	Gain	-	-	√	LOC440040
chr11	q11	55,124,730	55,209,599	84,869	Loss	Yes	-	√	OR4C11, OR4P4, OR4S2, OR4C6
chr11	q13.3	69,754,380	69,761,044	6,664	Loss	Yes	Yes	√	
chr11	q25	131,427,991	131,434,718	6,727	Gain	-	-	√	NTM
chr12	p13.33	2,120,303	2,125,563	5,260	Gain	-	-	√	CACNA1C
chr12	p13.31	9,528,351	9,621,934	93,583	Gain	Yes	-	√	
chr12	p13.2	11,109,511	11,147,734	38,223	Loss	-	Yes	√	PRR4, PRH1, TAS2R43
chr12	p13.2	11,398,676	11,435,698	37,022	Loss	-	-	√	PRB1, PRB2
chr12	p11.21	31,158,728	31,244,248	85,520	Gain	-	Yes	√	
chr12	p11.21	31,943,340	31,949,540	6,200	Loss	-	Yes	√	
chr12	p11.1	33,278,235	33,286,046	7,811	Gain	Yes	-	√	
chr12	q14.1	58,808,105	58,811,295	3,190	Gain	-	-	√	
chr12	q23.1	98,319,657	98,325,197	5,540	Loss	-	Yes	√	ANKS1B
chr12	q24.33	130,697,010	130,701,099	4,089	Loss	-	Yes	√	
chr13	q11	18,073,090	18,086,981	13,891	Gain	Yes	-	√	
chr13	q11	18,244,343	18,259,026	14,683	Gain	-	-	√	
chr13	q13.2	33,033,905	33,042,616	8,711	Gain	-	-	√	
chr13	q13.3	36,971,154	36,982,936	11,782	Loss	-	Yes	√	
chr13	q21.31	60,861,761	60,868,375	6,614	Gain	-	-	√	
chr13	q21.33	69,636,273	69,671,404	35,131	Gain	-	-	√	
chr13	q33.3	107,745,062	107,749,740	4,678	Loss	-	-	NO	TNFSF13B, TNFSF13B
chr14	q11.1	18,482,027	18,537,236	55,209	Loss	-	-	√	
chr14	q11.1 - q11.2	18,539,864	19,235,908	696,044	Loss	-	-	√	POTEG, P704P
chr14	q11.2	19,622,160	19,624,876	2,716	Gain	-	-	√	
chr14	q21.1	39,679,948	39,686,773	6,825	Loss	Yes	Yes	√	
chr14	q24.1	69,088,190	69,092,270	4,080	Loss	-	-	√	
chr14	q24.3	73,080,191	73,107,478	27,287	Gain	-	Yes	√	HEATR4, ACOT1, ACOT2
chr14	q32.33	105,312,580	105,393,261	80,681	Gain	-	-	√	
chr14	q32.33	105,403,001	105,428,738	25,737	Gain	-	-	√	
chr14	q32.33	105,434,073	105,599,350	165,277	Gain	Yes	-	√	KIAA0125, ADAM6
chr14	q32.33	105,602,602	105,637,941	35,339	Loss	Yes	-	√	
chr14	q32.33	105,641,753	105,894,508	252,755	Gain	-	-	√	
chr14	q32.33	105,957,708	106,002,104	44,396	Loss	Yes	-	√	
chr14	q32.33	106,219,784	106,253,703	33,919	Gain	-	-	√	
chr15	q11.2	18,464,929	20,092,742	1,627,813	Gain	Yes	Yes	√	LOC727832, GOLGA8C, LOC646214, CXADRP2, POTEB, LOC727924, OR4M2, OR4N4, LOC650137
chr15	q11.2	21,009,492	21,065,244	55,752	Gain	Yes	-	√	
chr15	q14	32,496,813	32,644,364	147,551	Gain	-	-	√	GOLGA8A, GOLGA8B, GOLGA8B
chr15	q24.3	74,678,353	74,682,295	3,942	Gain	-	-	√	SCAPER, SCAPER

Supplementary Table 1. CNVs found in neuropathy subject's genome

Chr	Cytoband	Start	Stop	Size	Gain/Loss	NimbArray	Sequencing	DGV	Gene Names
chr15	q24.3	75,117,799	75,119,772	1,973	Loss	-	Yes	√	
chr16	p12.2	21,444,541	21,498,345	53,804	Gain	-	Yes	√	
chr16	p12.1	22,530,238	22,617,276	87,038	Gain	-	Yes	√	LOC653786
chr16	p11.2	32,121,449	32,217,244	95,795	Loss	-	-	√	
chr16	p11.2	32,475,309	32,562,578	87,269	Loss	-	-	√	
chr16	p11.2	33,499,350	33,536,585	37,235	Loss	-	-	√	
chr16	p11.2 - p11.1	34,325,501	34,614,367	288,866	Loss	Yes	-	√	LOC283914, LOC283914, LOC146481
chr16	p11.1	35,102,881	35,112,822	9,941	Loss	-	-	√	
chr16	q11.2	44,943,402	44,988,282	44,880	Loss	-	-	√	
chr16	q22.1	68,707,057	68,763,721	56,664	Loss	-	-	√	PDPR
chr16	q23.1	76,929,598	76,941,510	11,912	Gain	-	-	√	WVVOX
chr16	q23.2	79,535,121	79,538,057	2,936	Loss	Yes	-	√	
chr17	q12	31,546,207	31,891,607	345,400	Loss	Yes	-	√	CCL3L1, CCL3L3, CCL4L1, CCL4L2, TBC1D3C, CCL3L1, CCL3L3, CCL4L2, CCL4L1, TBC1D3H, TBC1D3C, TBC1D3G
chr17	q12	33,333,517	33,384,910	51,393	Loss	-	-	√	TBC1D3F
chr17	q21.2	36,675,987	36,680,616	4,629	Loss	-	Yes	√	
chr17	q21.31	41,020,105	41,060,458	40,353	Gain	Yes	-	√	LOC644172, MGC57346, MGC57346
chr17	q21.31	41,568,539	41,642,565	74,026	Gain	Yes	Yes	√	KIAA1267
chr17	q21.31 - q21.32	41,670,262	42,137,274	467,012	Gain	Yes	Yes	√	LRRC37A, ARL17, LRRC37A2, ARL17P1, ARL17P1, ARL17, NSF
chr17	q24.1	60,345,240	60,357,798	12,558	Gain	Yes	-	√	LRRC37A3
chr17	q25.3	76,218,706	76,226,410	7,704	Gain	Yes	-	NO	RPTOR
chr18	p11.21	14,273,202	14,293,156	19,954	Gain	-	Yes	√	
chr18	p11.21	14,439,843	14,534,484	94,641	Gain	Yes	-	√	CXADRP3, POTEK
chr18	q12.3	40,230,827	40,235,960	5,133	Loss	Yes	Yes	√	
chr18	q21.2	49,460,662	49,464,492	3,830	Gain	-	-	√	
chr19	q13.11	38,176,810	38,188,500	11,690	Gain	Yes	-	NO	
chr19	q13.31	48,397,016	48,455,618	58,602	Gain	Yes	-	√	PSG4, PSG9
chr19	q13.41	58,212,113	58,241,832	29,719	Loss	-	-	√	
chr19	q13.42	59,418,579	59,434,216	15,637	Gain	-	-	√	LILRB3, LILRA6
chr19	q13.42	59,492,764	59,498,720	5,956	Gain	-	-	√	LILRA3
chr20	p13	1,509,459	1,541,599	32,140	Loss	-	Yes	√	SIRPB1, SIRPB1
chr21	q11.2	13,890,329	14,031,327	140,998	Gain	Yes	-	√	POTED, LOC441956
chr21	q21.2	24,756,302	25,018,441	262,139	Gain	Yes	Yes	NO	
chr21	q22.3	43,649,217	43,660,977	11,760	Loss	-	-	√	SIK1
chr22	q11.21	17,091,100	17,253,816	162,716	Gain	Yes	-	√	GGT3P
chr22	q11.21	18,705,549	18,731,613	26,064	Gain	Yes	-	√	
chr22	q11.21	18,758,765	18,854,922	96,157	Loss	-	-	√	P14KAP1, RIMBP3
chr22	q11.21	18,971,693	18,989,606	17,913	Gain	Yes	-	√	
chr22	q11.21	19,796,445	20,021,759	225,314	Gain	Yes	-	√	POM121L8P
chr22	q11.21	20,043,011	20,058,135	15,124	Loss	-	-	√	
chr22	q11.21	20,152,530	20,241,815	89,285	Loss	-	-	√	P14KAP2, RIMBP3B, RIMBP3C
chr22	q11.22	20,785,436	20,932,348	146,912	Gain	-	-	√	VPREB1
chr22	q11.22	20,999,667	21,292,917	293,250	Gain	-	-	√	VPREB1, LOC96610, ZNF280B, ZNF280A, PRAME, LOC648691
chr22	q11.22	21,327,935	21,484,188	156,253	Gain	-	-	√	POM121L1P, GGTL2
chr22	q11.22	21,485,941	21,568,919	82,978	Gain	-	-	√	
chr22	q11.23	22,606,174	22,669,363	63,189	Loss	-	Yes	√	GSTT2B, GSTT2, DDTL, DDT, DDT, GSTT2
chr22	q11.23	22,677,959	22,725,353	47,394	Gain	Yes	Yes	√	LOC391322, GSTT1, GSTTP2
chr22	q12.2	28599591	28613059	13468	Gain	Yes	-	NO	MTMR3
chr22	q13.1	37689058	37715461	26403	Loss	Yes	Yes	√	APOBEC3A, APOBEC3B
chr22	q13.33	48708883	48714608	5725	Loss	-	-	NO	
chrX	q12	67041234	67045513	4279	Loss	Yes	Yes	√	
chrX	q21.31	90377597	90398299	20702	Loss	Yes	Yes	NO	
chrX	q21.32	92683913	92687245	3332	Loss	-	Yes	√	
chrX	q26.3	134760449	134783844	23395	Loss	Yes	-	√	
chrX	q28	153075064	153177200	102136	Loss	-	-	√	OPN1LW, OPN1MW, OPN1MW2, OPN1MW, OPN1MW2, TEX28, TKTL1
chrX	q28	154447839	154449727	1888	Loss	Yes	-	√	TMLHE
chrY	p11.2	3773948	3780900	6952	Gain	-	Yes	√	
chrY	p11.2	4629543	4637623	8080	Loss	Yes	-	√	

Supplementary Table 2. nsSNPs cross-referenced to the Human Gene Mutation Database

HGMD_Disease	Type	OMIM	chr	position	strand	ref-snp	obs-snp	rs_id	HUGO_Sym	AA_change	Prot_Pos	HGMD_ID	Reference
Alcoholism, increased risk, association with ?	Beh Dis		chr4	100479812	-	T	Y	rs698	ADH1C	Ile-Val	350	CM033593	Konishi Exp Mol Pathol 74 183 2003
Mood disorders, reduced risk of childhood-onset, association with	Beh Dis		chr1	204391258	+	G	S	rs35369693	AVPR1B	Lys-Asn	65	N	Dempster Arch Gen Psychiatry 64 1189 2007
Schizophrenia, association with ?	Beh Dis		chr20	5851848	+	C	S	rs236152	CHGB	Ala-Gly	353	CM043990	Iijima Biol Psychiatry 56 10 2004
Schizophrenia, association with ?	Beh Dis		chr20	5852040	-	G	R	rs742711	CHGB	Arg-His	417	CM043991	Iijima Biol Psychiatry 56 10 2004
Schizophrenia, association with	Beh Dis		chr3	336508	+	C	T	rs2272522	CHL1	Leu-Phe	17	CM023348	Sakurai Mol Psychiatry 7 412 2002
Bipolar affective disorder, association with	Beh Dis		chr22	18331271	+	G	A	rs4680	COMT	Val-Met	158	CM960420	Lachman Am J Med Genet 67 468 1996
Schizophrenia, association with ?	Beh Dis		chr13	104917447	+	G	R	rs2391191	DAOA	Arg-Lys	30	CM045704	Wang Biochem Biophys Res Commun 319 1281 2004
Schizophrenia, risk, association with	Beh Dis		chr1	230211221	-	A	W	rs821616	DISC1	Ser-Cys	704	CM051907	Callicott Proc Natl Acad Sci U S A 102 8627 2005
Drug/alcohol abuse, association with	Beh Dis		chr1	46643348	+	C	M	rs324420	FAAH	Pro-Thr	129	CM023913	Sipe Proc Natl Acad Sci U S A 99 8394 2002
Depressive disorder, reduced risk, association with ?	Beh Dis		chr17	44942818	+	C	Y	rs2072446	NGFR	Ser-Leu	205	CM042757	Kunugi Am J Med Genet 129B 44 2004
Schizophrenia, association with	Beh Dis		chr8	20082746	+	T	K	rs2270641	SLC18A1	Thr-Pro	4	CM057213	Bly Schizophr Res 78 337 2005
Lung cancer, susceptibility to, association with	Cancer		chr17	19802050	-	C	Y	rs2108978	AKAP10	Arg-His	249	CM067336	Rudd Genome Res 16 693 2006
Colorectal cancer, increased risk, association with	Cancer		chr10	81916682	-	G	R	rs1049550	ANXA11	Arg-Cys	230	CM064971	Webb Hum Mol Genet 15 3263 2006
Cutaneous melanoma, association with ?	Cancer		chr14	19994994	+	T	G	rs1130409	APEX1	Asp-Glu	148	CM063843	Li Carcinogenesis 27 1894 2006
Lung cancer, reduced risk in Mexican Americans, association with	Cancer		chr1	155173739	+	T	Y	rs63387061	ARHGEF11	Ser-Gly	1416	CM061640	Gu Cancer 106 2707 2006
Cancer, association with	Cancer		chr13	49103026	-	T	Y	rs3803185	ARL11	Cys-Arg	148	CM062430	Frank Int J Cancer 118 2505 2006
Oesophageal cancer, association with	Cancer		chr20	54394948	+	A	W	rs2273535	AURKA	Phe-Ile	31	CM042940	Miao Cancer Res 64 2680 2004
Lung cancer, susceptibility to, association with	Cancer		chr6	31718665	-	A	G	rs60202657	BAT3	Ser-Pro	625	CM067350	Rudd Genome Res 16 693 2006
Breast cancer ?	Cancer		chr17	38497035	-	T	Y	rs28897689	BRCA1	Arg-Gly	1347	CM960181	Durocher Hum Mol Genet 5 835 1996
Breast cancer, association with	Cancer		chr13	31804729	-	A	M	rs144848	BRCA2	Asn-His	372	CM002750	Healey Nat Genet 26 362 2000
Colorectal cancer, increased risk, association with	Cancer		chr1	8939791	+	A	R	rs2274333	CA6	Ser-Gly	90	CM065045	Webb Hum Mol Genet 15 3263 2006
Lung cancer, susceptibility to, association with	Cancer		chr17	3722597	+	T	Y	rs17855558	CAMKK1	Glu-Gly	375	CM067356	Rudd Genome Res 16 693 2006
Cancer, association with	Cancer		chr6	36759949	+	C	M	rs1801270	CDKN1A	Ser-Arg	31	CM940226	Mousses Hum Mol Genet 4 1089 1995
Lung cancer, susceptibility to, association with	Cancer		chr1	145207126	+	C	S	rs17356233	CHD1L	His-Gln	350	CM067658	Rudd Genome Res 16 693 2006
Colorectal cancer, increased risk, association with	Cancer		chr1	201460809	+	C	Y	rs2297950	CNT1	Gly-Ser	102	CM065066	Webb Hum Mol Genet 15 3263 2006
Lung cancer, susceptibility to, association with	Cancer		chr6	75854022	-	C	T	rs970547	COL12A1	Gly-Ser	3058	CM067364	Rudd Genome Res 16 693 2006
Lung cancer, susceptibility to, association with	Cancer		chr1	158018690	+	G	R	rs1129923	DUSP23	Gly-Ser	131	CM067373	Rudd Genome Res 16 693 2006
Lung cancer, susceptibility to, association with	Cancer		chr17	53626093	+	G	C	rs11652709	EPX	Gln-His	122	CM067664	Rudd Genome Res 16 693 2006
Ovarian cancer susceptibility, association with	Cancer		chr2	49044545	-	C	T	rs6165	FSHR	Ala-Thr	307	CM062747	Yang Carcinogenesis 27 1502 2006
Cancer risk, association with	Cancer		chr10	106012779	+	C	M	rs4925	GSTO1	Ala-Asp	140	CM061795	Marahatta Cancer Lett 236 276 2006
Oral cancer, association with	Cancer	*134660	chr11	67109265	+	A	R	rs1695	GSTP1	Ile-Val	105	CM990713	Katoh Pharmacogenetics 9 165 1999
Lung cancer, susceptibility to, association with	Cancer		chr12	54375624	-	C	T	rs1800974	ITGA7	Arg-His	651	CM067685	Rudd Genome Res 16 693 2006
Colorectal cancer, increased risk, association with	Cancer		chr17	3579585	-	G	R	rs1716	ITGAE	Arg-Tyr	950	CM065287	Webb Hum Mol Genet 15 3263 2006
Reduced risk of multiple myeloma, association	Cancer	*601837	chr13	107661610	-	G	R	rs1805389	LIG4	Gln-Trp	3	CM023642	Roddam J Med Genet 39 900 2002
Reduced risk of multiple myeloma, association	Cancer	*601837	chr13	107661592	-	G	R	rs1805388	LIG4	Thr-Ile	9	CM023643	Roddam J Med Genet 39 900 2002
Leukaemia, risk, association with ?	Cancer		chr21	46529324	+	G	R	rs9975588	MCM3AP	Ser-Leu	102	CM065300	Rudd Blood 108 638 2006
Colorectal cancer, increased risk, association with	Cancer		chr18	28051919	-	C	Y	rs616114	MEP1B	Pro-Leu	695	CM065309	Webb Hum Mol Genet 15 3263 2006
Leukaemia, risk, association with	Cancer		chr20	44075813	+	G	C	rs2250889	MMP9	Arg-Pro	574	CM064121	Rudd Blood 108 638 2006
Lung cancer, susceptibility to, association with	Cancer		chr11	59697175	+	T	W	rs7232	MS4A6A	Asn-Ile	150	CM067701	Rudd Genome Res 16 693 2006
Lung cancer, susceptibility, association with	Cancer		chr3	9773773	+	C	S	rs1052133	OGG1	Ser-Cys	326	CM993185	Sugimura Cancer Epidemiol Biomarkers Prev 8 669 1999
Lung cancer, susceptibility to, association with	Cancer		chr17	8101874	+	C	Y	rs4791641	PFAS	Pro-Leu	367	CM067707	Rudd Genome Res 16 693 2006
Colorectal cancer, predisposition, association ?	Cancer		chr17	4667435	+	C	T	rs1052748	PLD2	Thr-Ile	577	CM030489	Yamada J Mol Med 81 126 2003
Colorectal cancer, non-polyposis	Cancer		chr7	6012153	+	C	Y	rs10254120	PMS2	Arg-Gln	20	CM941176	Nicolaides Nature 371 75 1994
Leukaemia, risk, association with ?	Cancer		chr2	170202109	+	A	G	rs8207	PP1G	Asn-Asp	699	CM063067	Rudd Blood 108 638 2006
Leukaemia, risk, association with	Cancer		chr8	53749259	+	A	R	rs17337252	RB1CC1	Met-Thr	234	CM063091	Rudd Blood 108 638 2006
Lung cancer, susceptibility to, association with	Cancer		chr13	48948621	+	A	G	rs7998427	SETDB2	Glu-Gly	117	CM067715	Rudd Genome Res 16 693 2006
Neoplastic disease, increased risk, association	Cancer		chr3	197285208	-	C	Y	rs3817672	TFR3	Gly-Ser	142	CM991171	Beckman Carcinogenesis 20 1231 1999
Colorectal cancer, increased risk, association with	Cancer		chr4	38476609	-	C	S	rs5743611	TLR1	Arg-Thr	80	CM065490	Webb Hum Mol Genet 15 3263 2006
Lung cancer, susceptibility to, association with	Cancer		chr9	116843092	-	C	Y	rs2274750	TNC	Ala-Thr	1781	CM067722	Rudd Genome Res 16 693 2006
Microangiopathy in type 2 diabetes, association	Disease		chr6	32259421	-	C	Y	rs2070600	AGER	Gly-Ser	82	CM994186	Kankova Diabetes Care 22 1745 1999
Hyperoxaluria, association with	Disease	#259900	chr2	241466189	+	A	R	rs4426527	AGXT	Ile-Met	340	CM910018	Purdue J Cell Biol 111 2341 1990
Cardiac disease, susceptibility to, association	Disease		chr17	19753133	-	T	Y	rs203462	AKAP10	Ile-Val	646	CM030773	Kammerer Proc Natl Acad Sci U S A 100 4066 2003
Arylsulphatase A pseudodeficiency, association	Disease	#250100	chr22	49411282	-	T	Y	rs2071421	ARSA	Asn-Ser	350	CM890013	Gieselmann Proc Natl Acad Sci U S A 86 9436 1989
Reduced risk of coronary artery disease, association	Disease		chr5	78457715	+	G	R	rs3733890	BHMT	Arg-Gln	239	CM031139	Weisberg Atherosclerosis 167 205 2003
Endometriosis, association with ?	Disease		chr17	38497961	-	T	Y	rs16941	BRCA1	Glu-Gly	1038	CM032861	Goumenou Fertil Steril 79 445 2003
Asthma, association with	Disease		chr17	29637007	+	G	A	rs1129844	CCL11	Ala-Thr	23	CM072923	Batra J Med Genet 44 397 2007
Multiple sclerosis, increased risk, association with	Disease	#126200	chr6	19613814	-	G	R	rs52812045	CD24	Ala-Val	57	CM035761	Zhou Proc Natl Acad Sci U S A 100 15041 2003
Alzheimer disease, association with	Disease		chr11	1739170	-	G	R	rs17571	CTSD	Ala-Val	58	CM000503	Papassotiropoulos Neurosci Lett 262 171 1999
Low renin hypertension, association with	Disease		chr8	143993541	-	T	C	rs4539	CYP11B2	Lys-Arg	173	CM962423	Fardella J Clin Endocrinol Metab 81 4347 1996
Preeclampsia, association with ?	Disease		chr1	224093029	+	A	R	rs2234922	EPHX1	His-Arg	139	CM024366	Laasanen Eur J Hum Genet 10 569 2002
Alzheimer disease, late-onset, association with	Disease		chr19	61385432	-	C	S	rs3745833	GALP	Ile-Met	72	CM071786	Grube Hum Mol Genet 16 865 2007
Coronary artery disease, association with	Disease		chr3	129687641	-	C	Y	rs2335052	GATA2	Ala-Thr	164	CM066570	Connelly PLoS Genet 2 e139 2006
Atherosclerosis, association with	Disease		chr1	35033356	+	C	Y	rs1764391	GJA4	Pro-Ser	319	CM994122	Boerma J Intern Med 246 211 1999
Asthma, increased risk, association with ?	Disease		chr17	35375519	-	G	R	rs3894194	GSDM1	Arg-Gln	18	CM073116	Moffatt Nature 448 470 2007
Diabetes, type 1, association with	Disease	610155*	chr2	162832297	+	C	T	rs1990760	IFIH1	Ala-Thr	946	CM066881	Smyth Nat Genet 38 617 2006
Sarcoidosis, increased risk, association with	Disease		chr9	21217622	-	A	C	rs9298814	IFNA17	Ile-Arg	184	CM041015	Akahoshi Hum Genet 114 503 2004
Psoriasis, increased risk, association with	Disease		chr1	67457975	+	T	C	rs7530511	IL23R	Leu-Pro	310	CM073141	Cargill Am J Hum Genet 80 273 2007
Atopy, association with	Disease		chr16	27281681	+	T	Y	rs1805015	IL4R	Ser-Pro	503	CM993667	Kruse Immunology 96 365 1999

Supplementary Table 2. nsSNPs cross-referenced to the Human Gene Mutation Database

HGMD_Disease	Type	OMIM	chr	position	strand	ref-snp	obs-snp	rs_id	HUGO_Sym	AA_change	Prot_Pos	HGMD_ID	Reference
Atopy, association with	Disease		chr16	27281901	+	A	R	rs1801275	IL4R	Gln-Arg	576	CM970783	Hershey N Engl J Med 337 1720 1997
Nephropathy, reduced risk, association with	Disease		chr3	187925712	+	T	Y	rs1656922	KNG1	Met-Thr	178	CM065290	Vionnet Diabetes 55 3166 2006
Neural tube defect, association with	Disease		chr14	63978598	-	G	R	rs2236225	MTHFD1	Arg-Gln	653	CM022820	Brody Am J Hum Genet 71 1207 2002
Cardiovascular disease and neural tube defects, association with	Disease	*607093	chr1	11778965	-	G	R	rs1801133	MTHFR	Ala-Val	222	CM950819	Frosst Nat Genet 10 111 1995
Coronary artery disease in smokers, association	Disease		chr1	235115123	+	A	G	rs1805087	MTR	Asp-Gly	919	CM983303	Wang J Cardiovasc Risk 5 289 1998
Alzheimer disease, late-onset, association with	Disease		chr17	10164439	+	T	Y	rs2074877	MTH13	Met-Val	1071	CM071869	Grube Hum Mol Genet 16 865 2007
Congenital nephrotic syndrome, Finnish type	Disease		chr19	41014441	+	T	Y	rs4806213	NPHS1	Asn-Ser	1077	CM024138	Gigante J Nephrol 15 696 2002
Minimal change nephrotic syndrome	Disease		chr19	41031849	-	G	R	.	NPHS1	Thr-Ile	294	CM044680	Lahdenkari Kidney Int 65 1856 2004
Stroke, increased risk, association with	Disease		chr1	11828655	+	A	R	rs5065	NPPA	STP-Arg	152	CM040788	Rubattu Stroke 35 814 2004
Glaucoma, open angle, association with	Disease		chr19	9829434	-	C	T	rs2303100	OLF2	Arg-Gln	106	CM066940	Funayama Invest Ophthalmol Vis Sci 47 5368 2006
Acute myocardial infarction, association with	Disease	602601	chr12	10204715	+	C	S	rs11053646	OLR1	Lys-Asn	167	CM033655	Tatsuguchi Biochem Biophys Res Commun 303 247 2003
Non-syndromic hearing impairment	Disease	*606067	chr20	16677048	+	T	Y	rs17686437	OTOR	Met-Thr	1	CM010359	Rendtorff Genomics 71 40 2001
Non-alcoholic fatty liver disease, association with	Disease		chr17	17350285	+	C	Y	rs7946	PEMT	Val-Met	175	CM054062	Song FASEB J 19 1266 2005
Asthma and atopy, high IgE levels, association with	Disease	+601690	chr6	46787262	-	A	R	rs1805018	PLA2G7	Ile-Thr	198	CM001308	Kruse Am J Hum Genet 66 1522 2000
PAF acetylhydrolase deficiency	Disease	+601690	chr6	46785057	+	C	M	rs16874954	PLA2G7	Val-Phe	279	CM961468	Stafforini J Clin Invest 97 2784 1996
Hypertension, reduced risk, association with	Disease		chr4	23424760	-	C	Y	rs8192678	PPARGC1A	Gly-Ser	482	CM013959	Ek Diabetologia 44 2220 2001
Familial haemophagocytic lymphohistiocytosis, susceptibility	Disease	*170280	chr10	72030393	+	G	R	rs35947132	PRF1	Ala-Val	91	CM022053	Busiello Int J Immunogenet 33 123 2006
Creutzfeldt-Jacob disease, Gerstmann-Straussler syndrome	Disease	*137440	chr20	4628251	+	A	R	rs1799990	PRNP	Met-Val	129	CM890104	Doh-Ura Biochem Biophys Res Commun 163 974 1989
Cleft lip / palate, association with	Disease		chr11	119015854	+	A	C	rs7940667	PVRL1	Val-Gly	361	CM066206	Avila Am J Med Genet A 140 2562 2006
Expression of allergic symptoms, association with	Disease		chr14	20430056	+	C	G	rs2073342	RNASE3	Thr-Arg	124	CM025442	Jonsson Clin Exp Allergy 32 1092 2002
Atopy, association with	Disease		chr11	65491750	-	G	S	rs660118	SART1	Gly-Ala	485	CM024558	Wheatley Hum Mol Genet 11 2143 2002
Hypertension, reduced risk, association with	Disease		chr12	6327323	-	T	C	rs2228576	SCNN1A	Thr-Ala	663	CM994637	Ambrosius Hypertension 34 631 1999
Diabetes, protection against, association with ?	Disease		chr1	167943110	-	A	R	rs1131498	SELL	Phe-Leu	193	CM004878	Kretowski Immunol Lett 74 225 2000
Higher blood pressure in males, association with	Disease		chr18	41516357	-	G	R	rs3745009	SLC14A2	Ala-Thr	880	CM013794	Ranade Hum Mol Genet 10 2157 2001
Atopy, maternally inherited, association with	Disease		chr5	147460220	+	G	R	rs2303063	SPINK5	Ser-Asn	368	CM013814	Walley Nat Genet 29 175 2001
Preeclampsia	Disease	#609404	chr10	70315382	+	A	M	rs10509305	STOX1	Glu-Asp	608	CM057372	van Dijk Nat Genet 37 514 2005
Rheumatoid arthritis, juvenile, association with	Disease		chr6	33380833	-	G	C	rs2071888	TAPBP	Thr-Arg	260	CM056992	Bukulmez Arthritis Res Ther 7 R285 2005
Cleft lip, alveolus, and palate, association	Disease		chr19	46539700	-	G	R	rs1800472	TGFB1	Thr-Ile	263	CM044946	Stoll BMC Med Genet 5 15 2004
Osteoporosis, association with	Disease		chr19	46550761	-	G	A	rs1800470	TGFB1	Pro-Leu	10	CM002115	Yamada J Bone Miner Res 13 1569 1998
Familial premature coronary artery disease, association with	Disease		chr15	37669470	+	A	R	rs2228262	THBS1	Asn-Ser	700	CM043345	Hannah J Biol Chem 279 51915 2004
Paget disease, association with	Disease		chr8	120033233	-	G	C	rs2073618	TNFRSF11B	Asn-Lys	3	CM045665	Daroszewska J Bone Miner Res 19 1506 2004
Systemic lupus erythematosus susceptibility, association with	Disease		chr19	10336652	+	C	M	rs2304256	TYK2	Val-Phe	362	CM050351	Sigurdsson Am J Hum Genet 76 528 2005
Thromboembolic disease, myocardial infarction, association with	Disease		chr8	31144196	+	T	Y	rs1346044	WRN	Cys-Arg	1367	CM971591	Ye Am J Med Genet 68 494 1997
Skin lesions in pseudoxanthoma elasticum (PXE), association with	Disease		chr17	45792455	+	C	Y	rs6504649	XYLT2	Thr-Arg	801	CM064361	Schon J Med Genet 43 745 2006
Stargardt disease	M_Disease	#248200	chr1	94341263	-	T	Y	.	ABCA4	Ile-Val	156	CM003569	Papaianou Invest Ophthalmol Vis Sci 41 16 2000
Stargardt disease	M_Disease	#248200	chr1	94316822	-	T	C	rs3112831	ABCA4	His-Arg	423	CM015072	Webster Invest Ophthalmol Vis Sci 42 1179 2001
Pseudoxanthoma elasticum, autosomal recessive	M_Disease	#177850	chr16	16159100	-	C	Y	rs2238472	ABCC6	Arg-Gln	1268	CM001044	Ringpfeil Proc Natl Acad Sci U S A 97 6001 2000
Adrenoleukodystrophy	M_Disease	#300100	chrX	152661677	.	G	R	.	ABCD1	Gly-Asp	608	CM012040	Dvorakova Hum Mutat 18 52 2001
Adenosine deaminase deficiency	M_Disease	#102700	chr20	42688634	-	T	Y	rs11555566	ADA	Lys-Arg	80	CM860001	Valerio EMBO J 5 113 1986
Metachromatic leukodystrophy	M_Disease	#250100	chr22	49410905	+	G	S	rs743616	ARSA	Thr-Ser	391	CM910052	Polten N Engl J Med 324 18 1991
Mucopolysaccharidosis VI	M_Disease	#253200	chr5	78170997	+	C	Y	rs25414	ARSB	Ser-Asn	384	CM003999	Voskoboeva Russ J Genet 36 689 2000
Progressive hearing loss, autosomal recessive	M_Disease	#610220	chr2	179033981	+	C	S	rs17304212	DFNB59	Arg-Gly	265	CM074142	Hashemzadeh Cholestori Clin Genet 72 261 2007
Mitochondrial neurogastrointestinal encephalopathy	M_Disease	#603041	chr22	49311121	-	C	Y	.	ECGF1	Ala-Thr	465	CM030064	Kocaefe Eur J Hum Genet 11 102 2003
Cockayne syndrome	M_Disease	#133540	chr10	50348375	-	T	Y	rs2228527	ERCC6	Arg-Gly	1213	CM980633	Mallery Am J Hum Genet 62 77 1998
Protoporphyrria, erythropoietic	M_Disease	#177000	chr18	53398334	+	C	M	rs3848519	FECH	Gly-Cys	55	CM910155	Lamoril Biochem Biophys Res Commun 181 594 1991
Gangliosidosis GM1	M_Disease	#230500	chr3	33113553	+	G	A	rs7637099	GLB1	Pro-Leu	10	CM055955	Gururaj J Child Neurol 20 57 2005
Spinal muscular atrophy with resp. distress 1	M_Disease	#604320	chr11	68462250	+	C	A	rs17612126	IGHMBP2	Thr-Lys	879	CM055288	Tachi Pediatr Neurol 32 288 2005
Multiple epiphyseal dysplasia (MED), modifier	M_Disease	#607078	chr2	20069022	-	C	Y	rs52826764	MATN3	Glu-Lys	252	CM040093	Jackson J Med Genet 41 52 2004
Myoclonic epilepsy of Lafora	M_Disease	#254780	chr6	18230485	+	G	A	rs10949483	NHLRC1	Pro-Leu	111	CM065338	Franceschetti Epilepsia 47 640 2006
Refsum disease	M_Disease	#266500	chr10	13380242	-	G	R	rs28938169	PHYH	Pro-Ser	29	CM001290	Jansen Hum Mol Genet 9 1195 2000
Cone-rod dystrophy	M_Disease	#608194	chr14	20859880	+	G	K	rs61722408	RPGRIP1	Ala-Ser	547	CM032029	Hameed J Med Genet 40 616 2003
Charcot-Marie-Tooth disease 4C	M_Disease	#601596	chr5	148386628	-	G	R	.	SH3TC2	Arg-STP	954	CM033084	Senderek Am J Hum Genet 73 1106 2003
Achondrogenesis 1B	M_Disease	#600972	chr5	149341414	+	A	W	rs3776070	SLC26A2	Thr-Ser	689	CM980573	Cai Am J Med Genet 78 58 1998
Peeling skin syndrome	M_Disease	#609796	chr15	41339641	.	C	M	.	TGM5	Gly-Cys	113	CM054150	Cassidy Am J Hum Genet 77 909 2005
Deafness, childhood onset	M_Disease	#601072	chr21	42682161	+	C	Y	rs45598239	TMPRSS3	Ala-Thr	90	CM054156	Hutchin Clin Genet 68 506 2005
Reduced serum noncholesterol sterols, association with	Met Trait		chr2	43952937	+	C	M	rs4148217	ABCG8	Thr-Lys	400	CM042913	Berge J Lipid Res 43 486 2002
Lower activity, association with	Met Trait		chr16	3973437	-	T	Y	rs2230739	ADCY9	Ile-Met	772	CM034003	Small Pharmacogenetics 13 535 2003
Hyperoxaluria, intermediary metabolic enzyme	Met Trait		chr2	241456987	+	C	Y	rs34116584	AGXT	Pro-Leu	11	CM910014	Purdue J Cell Biol 111 2341 1990
Elevated plasma fibrinogen levels, association with	Met Trait		chr14	24113511	+	T	Y	rs45567233	CTSG	Asn-Ser	125	CM015243	Herrmann Arterioscler Thromb Vasc Biol 21 1538 2001
Insulin resistance, association with	Met Trait		chr6	132214061	+	A	M	rs1044498	ENPP1	Lys-Gln	121	CM993455	Pizzuti Diabetes 48 1881 1999
N-acetylglucosamine binding, association with	Met Trait		chr9	136918847	+	C	Y	rs57863140	FCN2	Thr-Met	236	CM051922	Hummelshoj Hum Mol Genet 14 1651 2005
Trimethylaminuria, mild	Met Trait	#602079	chr1	169343590	+	G	R	rs2266782	FMO3	Glu-Lys	158	CM992884	Akerman Mol Genet Metab 68 24 1999
Elevated leptin levels, association with, glucose tolerance	Met Trait		chr1	65809029	+	A	R	rs1137100	LLEPR	Lys-Arg	109	CM032948	van Rossum Obes Res 11 377 2003
Mannose-binding protein deficiency, association	Met Trait		chr10	54201248	-	G	R	rs5030737	MBL2	Arg-Cys	52	CM960957	Madsen Immunogenetics 40 37 1994
Decreased enzyme activity and benzene toxicity, association with	Met Trait		chr16	68302646	-	G	R	rs1800566	NQO1	Pro-Ser	187	CM950861	Rosvold Pharmacogenetics 5 199 1995
Plasma total cholesterol, apolipoprotein B, and lipoprotein levels, assoc	Met Trait		chr1	55301775	+	G	R	rs505151	PSK9	Gly-Glu	670	CM051950	Chen J Am Coll Cardiol 45 1611 2005
Hypertriglyceridaemia, association with	Met Trait	*603937	chr8	55701948	+	A	W	rs2293869	RP1	Asn-Tyr	985	CM035763	Fujita J Hum Genet 48 305 2003
Non-inhibitory variant	Met Trait		chr18	59538313	+	T	W	rs1506419	SERPIN11	Trp-Arg	188	CM074500	Askew J Biol Chem 282 24948 2007

Supplementary Table 2. nsSNPs cross-referenced to the Human Gene Mutation Database

HGMD_Disease	Type	OMIM	chr	position	strand	ref-snp	obs-snp	rs_id	HUGO_Sym	AA_change	Prot_Pos	HGMD_ID	Reference
Non-inhibitory variant	Met Trait		chr18	59541341	+	T	Y	rs1395267	SERPINB11	Ser-Pro	303	CM074499	Askew J Biol Chem 282 24948 2007
Increased folate levels, association with	Met Trait		chr17	18172821	+	G	R	rs1979277	SHMT1	Leu-Phe	474	CM014391	Heil Mol Genet Metab 73 164 2001
Reduced function, association with	Met Trait		chr6	160463138	+	C	Y	rs12208357	SLC22A1	Arg-Cys	61	CM072040	Shu Proc Natl Acad Sci U S A 100 5902 2003
Reduced serum myo-inositol concentration, association	Met Trait		chr16	24796147	+	T	Y	rs11074656	SLC5A11	Val-Ala	182	CM044067	Groenen Mol Genet Metab 82 154 2004
Cholestasis, drug-induced, association with	Pharm Trait		chr2	169538574	-	A	G	rs2287622	ABCB11	Val-Ala	444	CM071525	Lang Pharmacogenet Genomics 17 47 2007
Gain of function, association with	Pharm Trait		chr10	115795046	+	G	C	rs1801253	ADRB1	Gly-Arg	389	CM994344	Mason J Biol Chem 274 12670 1999
ara-C resistance, association with	Pharm Trait	*123920	chr1	20788288	+	A	M	rs2072671	CDA	Lys-Gln	27	CM984152	Kirch Exp Hematol 26 421 1998
Poor paclitaxel metabolism, association with	Pharm Trait		chr10	96817020	-	C	Y	rs11572080	CYP2C8	Arg-Lys	139	CM014700	Soyama Biol Pharm Bull 24 1427 2001
Poor paclitaxel metabolism, association with	Pharm Trait		chr10	96788739	+	T	Y	rs10509681	CYP2C8	Lys-Arg	399	CM014701	Soyama Biol Pharm Bull 24 1427 2001
Poor metaboliser of sulfonylurea, warfarin sensitivity	Pharm Trait	*601130	chr10	96731043	+	A	M	rs1057910	CYP2C9	Ile-Leu	359	CM960481	Sullivan Pharmacogenetics 6 341 1996
Poor metaboliser, warfarin sensitivity	Pharm Trait	*601130	chr10	96692037	+	C	T	rs1799853	CYP2C9	Arg-Cys	144	CM994193	Aynacioglu Br J Clin Pharmacol 48 409 1999
FMO2 variant	Pharm Trait		chr1	169435169	+	T	Y	rs2307492	FMO2	Phe-Ser	182	CM033899	Furnes Drug Metab Dispos 31 187 2003
Slow acetylation	Pharm Trait	#243400	chr8	18302134	+	T	C	rs1801280	NAT2	Ile-Thr	114	CM910269	Vatsis Proc Natl Acad Sci U S A 88 6333 1991
POR deficiency	Pharm Trait		chr7	75452942	+	C	Y	rs1057868	POR	Ala-Val	500	CM057356	Miller Ann N Y Acad Sci 1061 100 2005
Thiopurine S-Methyltransferase deficiency	Pharm Trait	#610460	chr6	18251934	-	C	S	rs1800462	TPMT	Ala-Pro	80	CM951239	Krynetski Proc Natl Acad Sci U S A 92 949 1995
Decreased enzyme activity	Pharm Trait		chr2	234302542	-	T	C	rs3821242	UGT1A3	Trp-Arg	11	CM042128	Iwai J Hum Genet 49 123 2004
Reduced catalytic activity, association with	Pharm Trait		chr2	234255944	+	T	C	rs11692021	UGT1A7	Trp-Arg	208	CM004345	Guillemette Pharmacogenetics 10 629 2000
Reduced catalytic activity, association with	Pharm Trait		chr2	234255709	+	T	G	rs17868323	UGT1A7	Asn-Lys	129	CM024576	Gemignani Pharmacogenetics 12 459 2002
Blood group variation	Trait	#111200	chr19	50014584	+	A	R	rs1135062	BCAM	Thr-Ala	539	CM973377	Parsons Blood 89 4219 1997
Gov platelet antigen variation	Trait		chr6	74550153	+	A	C	rs10455097	CD109	Tyr-Ser	703	CM020385	Schuh Blood 99 1692 2002
Menstrual cycle, length, association with	Trait		chr2	49043425	-	C	T	rs6166	FSHR	Ser-Asn	680	CM052876	Greb J Clin Endocrinol Metab 90 4866 2005
Apoptosis, unable to induce, association with	Trait		chr14	24171429	-	G	S	rs11539752	GZMB	Pro-Ala	94	CM033776	Mclroy Proc Natl Acad Sci U S A 100 2562 2003
Apoptosis, unable to induce, association with	Trait		chr14	24170122	+	A	R	rs2236338	GZMB	Tyr-His	247	CM033777	Mclroy Proc Natl Acad Sci U S A 100 2562 2003
Decreased total IgE level, association with	Trait		chr16	27281373	+	A	M	rs1805011	IL4R	Glu-Ala	400	CM993665	Hackstein Hum Immunol 60 1119 1999
Increased BMI and levels of soluble receptor, association with	Trait		chr1	152693594	+	A	C	rs2228145	IL6R	Asp-Ala	358	CM034737	Wolford Mol Genet Metab 80 338 2003
Dyslexia susceptibility, association with	Trait	600202%	chr6	24696863	+	C	Y	rs4504469	KIAA0319	Ala-Thr	311	CM057333	Cope Am J Hum Genet 76 581 2005
Higher body mass index, association with	Trait		chr1	65831101	+	A	G	rs1137101	LEPR	Gln-Arg	223	CM010905	Quinton Hum Genet 108 233 2001
Exfoliation glaucoma syndrome increased risk, association with	Trait	*153456	chr15	72006599	+	G	T	rs1048661	LOXL1	Arg-Leu	141	CM074329	Thorleifsson Science 317 1397 2007
Lumbar spine bone-mineral content, association with	Trait		chr11	67957871	+	C	T	rs3736228	LRP5	Ala-Val	1330	CM067428	van Meurs J Bone Miner Res 21 141 2006
Red hair, association with	Trait		chr16	88513592	+	G	R	rs11547464	MC1R	Arg-His	142	CM994355	Schioth Biochem Biophys Res Commun 260 488 1999
Cardiac hypertrophy, protection, association	Trait		chr14	23908923	+	G	C	rs2229309	NFATC4	Gly-Ala	160	CM032294	Poirier Eur J Hum Genet 11 659 2003
Darker eye colour, association with	Trait	*611409	chr15	25903913	-	C	Y	rs1800407	OCA2	Arg-Gln	419	CM025432	Rebeck Cancer Epidemiol Biomarkers Prev 11 782 2002
Skin/hair/eye pigmentation variation, association	Trait	#227240	chr5	33999627	+	C	Y	rs26722	SLC45A2	Glu-Lys	272	CM051555	Graf Hum Mutat 25 278 2005
Mycobacterium tuberculosis, susceptibility to	Trait	#607948	chr2	230758959	-	A	G	rs3948464	SP110	Leu-Ser	425	CM066604	Tosh Proc Natl Acad Sci U S A 103 10364 2006
Phenylthiocarbamide taste sensitivity	Trait	#171200	chr7	141319814	-	C	G	rs713598	TAS2R38	Ala-Pro	49	CM031368	Kim Science 299 1221 2003
Phenylthiocarbamide taste sensitivity	Trait	#171200	chr7	141319073	+	T	C	rs10246939	TAS2R38	Ile-Val	296	CM031370	Kim Science 299 1221 2003
Altered activity, association with	Trait		chr4	187241068	-	C	Y	rs3775291	TLR3	Leu-Phe	412	CM073380	Ranjith-Kumar J Biol Chem 282 17696 2007
Legionnaires disease susceptibility, SLE resistance	Trait	#608556	chr1	221351823	-	G	R	rs5744168	TLR5	Arg-STP	392	CM034621	Hawn J Exp Med 198 1563 2003

Supplementary Table 3. Neuropathy associated genes.

Gene	Other Name	Chr	CytoBand	Start	End	All SNPs	cSNPs	
<i>NGF</i>	<i>NGFB</i>	chr1	1p13.1	115,828,537	115,880,857	59	1	
<i>YARS</i>	-	chr1	1p35.1	33,240,840	33,283,633	24	0	
<i>KIF1B</i>	-	chr1	1p36.2	10,270,764	10,441,661	17	0	
<i>MFN2</i>	-	chr1	1p36.22	12,040,238	12,073,572	52	0	
<i>PLEKHG5</i>	-	chr1	1p36.31	6,526,152	6,580,069	93	5	
<i>LMNA</i>	-	chr1	1q21.2-q21.3	156,084,461	156,109,878	6	0	
<i>NTRK1</i>	-	chr1	1q21-q22	156,785,542	156,851,642	68	1	
<i>MPZ</i>	-	chr1	1q23.3	161,274,525	161,279,762	3	0	
<i>DCTN1</i>	-	chr2	2p13	74,588,282	74,607,475	2	0	
<i>RAB7A</i>	<i>RAB7</i>	chr3	3q21.3	128,444,979	128,533,641	75	0	
<i>SH3TC2</i>	<i>KIAA1985</i>	chr5	5q32	148,361,713	148,442,737	71	3	
<i>FIG4</i>	<i>SAC3</i>	chr6	6q21	110,012,424	110,146,634	0	0	
<i>GARS</i>	-	chr7	7p15	30,634,181	30,673,649	39	3	
<i>HSPB1</i>	<i>HSP27</i>	chr7	7q11.23	75,931,875	75,933,614	8	0	
<i>NEFL</i>	<i>NFL</i>	chr8	8p21	24,808,468	24,814,131	12	0	
<i>ARHGEF10</i>	-	chr8	8p23	1,772,149	1,906,807	228	4	
<i>GDAP1</i>	-	chr8	8q21.11	75,262,618	75,279,345	30	1	
<i>NDRG1</i>	-	chr8	8q24.3	134,249,414	134,309,547	96	1	
<i>SPTLC1</i>	<i>SPT1</i>	chr9	9q22.2	94,793,427	94,877,690	10	1	
<i>IKBKAP</i>	-	chr9	9q31	111,629,800	111,696,608	60	1	
<i>SETX</i>	-	chr9	9q34.13	135,136,827	135,230,372	111	6	
<i>EGR2</i>	-	chr10	10q21.1	64,571,757	64,578,927	4	1	
<i>SBF2</i>	<i>MTMR13</i>	chr11	11p15.4	9,800,214	10,315,754	859	2	
<i>BSCL2</i>	<i>SEIPIN</i>	chr11	11q12-q13.5	62,457,747	62,477,046	20	1	
<i>IGHMBP2</i>	-	chr11	11q13.3	68,671,319	68,708,069	34	3	
<i>MTMR2</i>	-	chr11	11q22	95,566,044	95,657,371	128	2	
<i>FGD4</i>	-	chr12	12p11.21	32,655,041	32,798,984	20	1	
<i>HSN2</i>	<i>HSAN2</i>	chr12	12p13.3	976,870	978,799	2	0	
<i>HSPB8</i>	<i>HSP22</i>	chr12	12q24.23	119,616,595	119,632,551	13	0	
<i>SLC12A6</i>	-	chr15	15q13-q15	34,522,197	34,630,265	277	3	
<i>LITAF</i>	<i>SIMPLE</i>	chr16	16p13.13	11,641,578	11,681,322	75	1	
<i>GAN</i>	-	chr16	16q24.1	81,348,571	81,413,803	77	1	
<i>PMP22</i>	-	chr17	17p12-p11.2	15,133,096	15,168,644	6	0	
<i>SEPT9</i>	-	chr17	17q25	75,277,492	75,496,678	308	2	
<i>CTDP1</i>	-	chr18	18q23	77,439,801	77,514,507	143	3	
<i>DNM2</i>	-	chr19	19p13.2	10,828,755	10,942,579	82	0	
<i>PRX</i>	-	chr19	19q13.13-q13.2	40,899,671	40,919,271	19	6	
<i>SOX10</i>	-	chr22	22q13.1	38,368,319	38,380,539	5	1	
<i>GJB1</i>	<i>CX32</i>	chrX	Xq13.1	70,435,062	70,445,065	6	0	
<i>PRPS1</i>	-	chrX	Xq21.32-q24	106,871,654	106,894,256	6	0	
						Total SNPs	3,148	54
						Known SNPs	2,682 (85.19%)	45 (83.33%)
						Novel SNPs	466 (14.8%)	9 (16.66%)