Supplementary Material

Table 1. Sequences of qPCR Primers and Probes

	Position	SNP/Gene	Forward primer [5' to 3']	Reverse Primer [5' to 3']	TaqMan probe [FAM5' to 3'BHQ]
CNV 19	Left	SNP_A-1648306	GCTCATGTCTAGAGCTCCAGCAT	AACCCTGAGTTTCCCTCTGATTC	ATGACGCGCCAAGCTTCAAGACCA
	Middle	SNP_A-1683854	GCCACTGCATCAATAAAACTTCTTC	CCTACAATGGTTGCAAACAGAATAAAT	CTCACATGACCTGCATTTCACTGGTTTCC
	Right	SNP_A-1669467	CATCCTAGAAGGTGGTCCTGAAA	AGACTCAGCTCAGAGCACAGCAT	CCAAAGCACCCTGTTACCTCCTCTATGACA
CNV 57	Left	SNP_A-1652418	GGATGAAAGGGAAAGGCATTTT	TCTCAGACCAGATTGATGAAATGTG	CCCAAAGCCTTGACGCTGCCCT
	Middle	SNP_A-1644782	ACAAAGCATCTAGAGTGCCAACAC	CACCGTCCACTTACACAAATGAA	CAGCCTCACCTACATTTTATCCCTTGGCA
	Right	SNP_A-1712159	AAATCAAAGCCAGCTGCCTTAT	CTCACCATCCCAGCTCAAGAA	CCTCAGGCAGGCCACCCTTCTCAC
CNV 77	Left	SNP_A-1658092	TCTTAGCGTTCTCAGATATTTTGCA	GACCAGTCATGGGACTTCTCACA	TGAGTCCTGTCTCCAGTCCATCTCAATGTG
	Middle	SNP_A-1677992	TTCCAAAAATAGACACGGAAGGA	AAAAGCAGTTGAGGTAATGAATACTGTAGT	CCTTCCAAGCCCCAATAGTGGGCA
	Right	SNP_A-1742931	TCAGCCTCAAATTATGATACAGTACGT	TGTCCAAGTGAGAGGTCCTTCTCT	TACTACAGCAGAGCTGAACACAGCATGCC
CNV 114	Left	SNP_A-1745948	CTGGGAAACCTCTTGATAGTGGTAA	GCTCAGGCATAGGTCAATGAAAG	CCCATGCTCACCTGCTCCAATCTCCTAT
	Middle	Gene_OR4M1_exon	CCCTCATCTGACTTCTCCTATGTATTT	GGGCTGTAATGGAAGAGTACCAAA	CTGTTGGCTAATCTGGCCCTCCTTGA
	Right	SNP_A-1710646	GAGCCATGATCACATAGTTGCACTA	TCTATCTAAGGATGTCTACCTGGTCACT	AGCCTGAGTGACAGCGCAAGACCC
CNV 119	Left	SNP_A-1713638	TGTTCTCCATTCACAGAAGCATTT	GGTGATATTTTAGGAAGAGCTGTGTCA	TGTGCAGTAAACAGCAAAAGCCAACAAGC
	Middle	Gene_BC066981_exon	CAGCCACCATCTGCTTTTAACAG	AGGGTTTCCTGCTAGTTGGTGAA	AGCGAACAGATCGCGTCTACCAACCAG
	Right	SNP_A-1729906	ATGCTTTGCAGAAGTGAGCTCAT	TCTCAGGACACCATTTTTCAAGTG	TAGAAGAGGCCTGCCCACCAAGAAGTCA
CNV 138-139	Left	SNP_A-1708192	ATCAGGGTTAGCTGCAAAAGGTA	GATAAGAGGTAGGTCTCTGTCAAGTAGTTG	CTCTATGCCCAGCAAAAGATACGTGCTCA
	Middle	SNP_A-1679546	TCACAGACCTTGCTTCCTCTTTC	GCTGGACGATTTGGTTCCTTAGT	CATCCACCAGCCAGGAAGAGAGCCTT
	Right	SNP_A-1724056	AGACCCAACTCCTTCTTCAGTCA	TGGGACTCCTCATTGAAAGATGT	TGTGCTGCCCCCACAGCTAGTTTG
Others CNV 1	Left	SNP_A-1689701	CCATCAGCTAATGCTCTCATTCTG	GAGTGTGACTCTGCAGCCTTTAAAG	TTCATACCCACCCAGCTTCTCCACTGAA
	Middle	SNP_A-1666858	CCACATAGATTCAGGTGCTTTCAC	GGGCCAATTTGCAGCTATAAGTA	TTCCTCAAACTGCAGGGCCCATG
	Right	SNP_A-1650806	CCCTTGTCTTCTCGGTTCTTAGAT	AGGGTCCAAGGTTATGGCATTAA	CTTCCTCAAATCCAACCTCATTCATCCATG
Others CNV 2	Middle	SNP_A-1694209 - SNP_A-1736287	CCTTACTCACTAGCAACCCCTCTT	AGATTCCTGCTTCTTTGAGTTCAGA	CCACCCAAGCCACTCAGCTGTCC
GAPDH			GCACCGTCAAGGCTGAGAA	CAAAGCACATTTCTTCCATTCTGT	AAATCCCATCACCATCTTCCAGGAGTGA

Table 2. Unique CNVs identified in this study. Highlighted entries indicate where overlapping CNVs from different subjects were broken up into non-overlapping ones. The "Status" column indicates whether the CNV reflects copy number gain (Gain), copy number loss (Loss), or Gain in some individuals and Loss in others (Both). The "Source of concordant CNVs" column indicates the source of other CNVs that our CNV is concordant with: 1. Sebat *et al.* 2004 [5]; 2. Iafrate *et al.* 2004 [6]; 3. McCarroll *et al.* 2006 [7]; 4. Redon *et al.* 2006 [9]

Index	Cytoband	Copy Number	Num of Individuals	Individuals	CNV Size [bp]	Num of SNPs in CNV	Chrom	Start Position	End Position	Status	Source of concordant CNVs
1	1p36.21	1	1	NA17102	68	2	1	15235123	15235190	Loss	
2	1p34.2	1	1	NA17011	153652	5	1	42446117	42599768	Loss	
3	1p32.3	3	1	NA17059	112454	4	1	54898707	55011160	Gain	4
4	1p32.2	4	1	NA17221	2178	3	1	56027678	56029855	Gain	
5	1p22.1	3	1	NA17105	568	4	1	94243345	94243912	Gain	
6	1p21.1	1	1	NA17017	174982	9	1	105758069	105933050	Loss	1,4
7	1p13.3	3	1	NA17139	410558	10	1	109727774	110138331	Gain	
8	1q23.1	1	1	NA17124	656	3	1	155333810	155334465	Loss	3
9	1q25.3	3	1	NA17260	14853	3	1	181838827	181853679	Gain	
10	1q32.2	1	1	NA17011	410169	6	1	204203584	204613752	Loss	4
11	1q42.11	3	1	NA17136	3476	4	1	220208280	220211755	Gain	
12	2p24.3	3	1	NA17221	367250	6	2	13197656	13564905	Gain	4
13	2p24.3	3	1	NA17245	110245	7	2	14316043	14426287	Gain	
14	2p16.2	1	1	NA17011	20992	6	2	53763463	53784454	Loss	
15	2q12.2-2q12.3	3	1	NA17052	1102513	27	2	106658036	107760548	Gain	
16	2q13	1	1	NA17124	52770	3	2	110286548	110339317	Loss	
17	2q14.1	1	1	NA17011	53031	7	2	114922263	114975293	Loss	
18	2q14.3	1	1	NA17200	3767374	55	2	125476308	129243681	Loss	
19	2q14.3-2q21.1	1	2	NA17200, NA17017	1156082	22	2	129243681	130399762	Loss	1,4
20	2q21.1-q21.3	1	1	NA17200	5311533	75	2	130399762	135711294	Loss	4
21	2q21.3-2q22.1	1	1	NA17200	1140205	34	2	136319703	137459907	Loss	
22	2q24.2	1	1	NA17011	144299	10	2	159790667	159934965	Loss	
23	2q24.3-2q31.1	1	1	NA17196	256790	22	2	169388542	169645331	Loss	
24	2q36.3	3	1	NA17125	20507	3	2	230539530	230560036	Gain	
25	3p22.2-3p22.1	3	1	NA17148	439285	14	3	39778910	40218194	Gain	

Index	Cytoband	Copy Number	Num of Individuals	Individuals	CNV Size [bp]	Num of SNPs in CNV	Chrom	Start Position	End Position	Status	Source of concordant CNVs
26	3q13.11	1	1	NA17107	2327	4	3	105082422	105084748	Loss	
27	3q13.12	3	1	NA17196	317774	10	3	108277472	108595245	Gain	
28	3q21.3	1	1	NA17279	770	3	3	131615549	131616318	Loss	3
29	3q22.1	1	1	NA17011	78450	6	3	132997063	133075512	Loss	
30	3q25.33-3q26.1	1	1	NA17011	351600	6	3	162192183	162543782	Loss	
31	3q26.1	3	1	NA17235	49296	7	3	163373314	163422609	Gain	
32	4p15.33	1	1	NA17108	344	3	4	12051326	12051669	Loss	
33	4q13.3	1	1	NA17125	432	3	4	75448371	75448802	Loss	
34	4q24	1	3	NA17107, NA17156, NA17111	1637	5	4	104570584	104572220	Loss	
35	4q27	1	1	NA17011	86479	5	4	122370647	122457125	Loss	
36	4q32.2	1	1	NA17258	2246	4	4	162432100	162434345	Loss	
37	4q32.2	1	1	NA17147	2565	4	4	164312697	164315261	Loss	
38	4q35.2	1	1	NA17251	25694	4	4	189302971	189328664	Loss	
39	4q35.2	3	1	NA17015	546289	16	4	189670188	190216476	Gain	
40	5q12.3	1	1	NA17016	33311	3	5	63751672	63784982	Loss	
41	5q13.3	3	1	NA17204	93456	6	5	75417559	75511014	Gain	4
42	5q15	1	1	NA17013	160785	14	5	95986496	96147280	Loss	
43	5q31.1	1	1	NA17011	50726	6	5	136664008	136714733	Loss	
44	5q35.2	1	1	NA17011	3326	5	5	173907751	173911076	Loss	
45	6p24.2	1	1	NA17011	18970	6	6	10676438	10695407	Loss	
46	6p24.1-6p23	1	1	NA17013	580512	11	6	13398217	13978728	Loss	
47	6q11.1	3	1	NA17253	837721	29	6	62080207	62917927	Gain	
48	6q16.1	0	1	NA17015	80	2	6	93333402	93333481	Loss	
49	6q22.31	1	1	NA17016	673622	22	6	123934753	124608374	Loss	
50	6q23.3	1	1	NA17105	17124	4	6	136047484	136064607	Loss	
51	7p21.3	1	1	NA17235	385	3	7	8016280	8016664	Loss	
52	7p21.3	3	1	NA17013	548079	39	7	10785314	11333392	Gain	

Index	Cytoband	Copy Number	Num of Individuals	Individuals	CNV Size [bp]	Num of SNPs in CNV	Chrom	Start Position	End Position	Status	Source of concordant CNVs
53	7p21.3	3	1	NA17197	8529	13	7	12294465	12302993	Gain	
54	7p14.1	1	1	NA17011	15595	6	7	37831567	37847161	Loss	
55	7q11.21-7q11.22	3	1	NA17157	22636	4	7	66480979	66503614	Gain	
56	7q21.13	2	2	NA17115, NA17260	95391	7	7	87794639	87890029	Both	4
57	7q21.13	3	1	NA17260	1908576	75	7	87890029	89798604	Gain	
58	7q22.2	1	1	NA17054	234484	19	7	103581736	103816219	Loss	
59	7q31.33	1	1	NA17260	122487	7	7	125238607	125361093	Loss	4
60	7q36.2	3	1	NA17255	319022	5	7	152455069	152774090	Gain	4
61	8p23.2	3	1	NA17059	789246	47	8	3803529	4592774	Gain	1,4
62	8p23.2	2	2	NA17059, NA17011	8803	8	8	4592774	4601576	Both	
63	8p23.2	3	1	NA17059	1157360	66	8	4601576	5758935	Gain	1,4
64	8p23.2	2	2	NA17059, NA17011	21922	6	8	5758935	5780856	Both	
65	8p23.2	3	1	NA17059	119600	15	8	5780856	5900455	Gain	
66	8p23.2-8p23.1	1	1	NA17011	282584	10	8	5940352	6222935	Loss	4
67	8p23.1	2	2	NA17160, NA17203	793	5	8	6780732	6781524	Both	
68	8p22	1	1	NA17112	1000	4	8	16370342	16371341	Loss	
69	8p21.3	1	2	NA17013, NA17011	347090	8	8	20266438	20613527	Loss	4
70	8p21.3	3	1	NA17258	135350	5	8	20699948	20835297	Gain	
71	8p12	1	1	NA17011	10021	5	8	29138530	29148550	Loss	
72	8q12.1	1	1	NA17196	981	3	8	58654291	58655271	Loss	
73	8q21.3	3	2	NA17106, NA17148	4297	3	8	90610612	90614908	Gain	
74	8q22.1	1	1	NA17011	22915	4	8	96461491	96484405	Loss	
75	8q22.1	1	2	NA17011, NA17013	265991	14	8	96484405	96750395	Loss	

Index	Cytoband	Copy Number	Num of Individuals	Individuals	CNV Size [bp]	Num of SNPs in CNV	Chrom	Start Position	End Position	Status	Source of concordant CNVs
76	8q22.1	1	1	NA17011	46890	6	8	98888600	98935489	Loss	
77	8q24.23	1	5	NA17253, NA17251, NA17208, NA17125, NA17267	72074	12	8	137820222	137892295	Loss	
78	9p24.3	1	1	NA17112	22727	2	9	559992	582718	Loss	
79	9p24.3	1	2	NA17112, NA17101	4109	5	9	582718	586826	Loss	3
80	9p24.2	1	1	NA17011	310628	11	9	4134958	4445585	Loss	
81	9p23	1	1	NA17156	32945	8	9	9842432	9875376	Loss	
82	9p23	1	1	NA17105	61000	5	9	12194748	12255747	Loss	
83	9p22.1	3	1	NA17221	6260	4	9	19042562	19048821	Gain	
84	9p21.1	1	1	NA17124	2406	4	9	28839504	28841909	Loss	
85	9q21.13	1	1	NA17011	486655	7	9	72181725	72668379	Loss	
86	9q21.31-9q21.32	1	1	NA17017	1059268	32	9	80580925	81640192	Loss	1
87	9q31.1	3	1	NA17132	7073	6	9	103054555	103061627	Gain	
88	10q21.1	1	1	NA17115	273285	12	10	57119488	57392772	Loss	
89	10q21.1	1	1	NA17147	107053	11	10	58035421	58142473	Loss	
90	10q21.3	1	1	NA17011	85116	7	10	64222940	64308055	Loss	
91	10q26.11	3	1	NA17213	474182	17	10	119615934	120090115	Gain	
92	10q26.12	3	1	NA17109	117205	5	10	122374273	122491477	Gain	
93	10q26.13	3	1	NA17058	600782	11	10	123651653	124252434	Gain	
94	10q26.3	3	5	NA17216, NA17208, NA17134, NA17125, NA17213	64488	5	10	135164239	135228726	Gain	
95	11p15.4	1	1	NA17011	1072662	6	11	2853359	3926020	Loss	
96	11p15.4	1	1	NA17156	66807	9	11	5151240	5218046	Loss	
97	11p15.4	4	1	NA17016	20252	4	11	5860386	5880637	Gain	

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98	11q13.4-11q13.5	1	1	NA17011	661322	7	11	74393553	75054874	Loss	
99	11q14.1	3	1	NA17283	2055	3	11	81554687	81556741	Gain	
100	11q14.2	1	1	NA17132	634	3	11	87786861	87787494	Loss	
101	11q23.2-q23.3	3	1	NA17206	1589405	45	11	114047758	115637162	Gain	
102	11q23.3	2	2	NA17206, NA17011	390903	9	11	115637162	116028064	Both	2
103	11q23.3-q25	3	1	NA17206	18054780	257	11	116028064	134082843	Gain	
104	12p13.2	4	1	NA17221	886	3	12	11674778	11675663	Gain	
105	12q12	1	1	NA17011	45203	5	12	39212721	39257923	Loss	
106	12q24.21	1	1	NA17216	722	2	12	115094366	115095087	Loss	
107	13q12.11	1	1	NA17019	123988	10	13	19841224	19965211	Loss	4
108	13q13.3	1	1	NA17011	58479	9	13	36705331	36763809	Loss	
109	13q14.11	3	1	NA17275	463177	26	13	42249947	42713123	Gain	
110	13q21.31	1	1	NA17152	652	2	13	61332602	61333253	Loss	
111	13q22.3	1	1	NA17011	157787	8	13	76817590	76975376	Loss	
112	13q22.3	1	1	NA17011	70565	7	13	77045128	77115692	Loss	
113	13q33.1	1	1	NA17011	31046	6	13	101887561	101918606	Loss	
114	14q11.2	4.5	2	NA17278, NA17254	204891	4	14	19285288	19490178	Gain	1,4
115	14q21.1	1	1	NA17148	56470	12	14	37623994	37680463	Loss	
116	14q23.2	3	1	NA17247	254506	12	14	62636388	62890893	Gain	
117	14q23.3	3	1	NA17247	232391	6	14	65146283	65378673	Gain	
118	14q24.2	1	1	NA17011	200183	5	14	70281911	70482093	Loss	
119	15q11.2	3.5	2	NA17104, NA17228	780624	5	15	19208413	19989036	Gain	4
120	15q21.3	1	1	NA17019	28970	5	15	56242490	56271459	Loss	
121	15q25.2-15q25.3	3	1	NA17107	828183	12	15	82342399	83170581	Gain	4
122	16q23.1	1	1	NA17155	6188	5	16	74749038	74755225	Loss	
123	17p13.3	3	1	NA17284	110036	5	17	451209	561244	Gain	
124	17p13.3	1	1	NA17054	856105	9	17	2339117	3195221	Loss	

Index	Cytoband	Copy Number	Num of Individuals	Individuals	CNV Size [bp]	Num of SNPs in CNV	Chrom	Start Position	End Position	Status	Source of concordant CNVs
125	17q12	1	1	NA17011	397219	6	17	31064899	31462117	Loss	4
126	17q12	3	2	NA17011, NA17278	41536	4	17	31462117	31503652	Both	
127	17q12	1	1	NA17011	616434	3	17	31503652	32120085	Loss	4
128	17q21.31	5	1	NA17261	164677	5	17	41539399	41704075	Gain	1
129	17q23.3	1	1	NA17011	166049	6	17	59511039	59677087	Loss	2
130	18q12.1	1	1	NA17217	489	3	18	27636787	27637275	Loss	
131	18q22.2	1	1	NA17215	1674	4	18	65300618	65302291	Loss	
132	19q13.43	3	1	NA17052	121251	13	19	61010000	61131250	Gain	
133	20q13.2	1	1	NA17015	646	3	20	52528815	52529460	Loss	
134	21q21.1	3	1	NA17205	102411	9	21	17901508	18003918	Gain	
135	21q21.1	3	1	NA17216	31482	5	21	18979549	19011030	Gain	
136	21q22.12	3	1	NA17258	490695	13	21	35854142	36344836	Gain	
137	22q11.22	1	1	NA17160	23146	7	22	20808466	20831611	Loss	
138	22q11.22	0	3	NA17207, NA17156, NA17160	85878	3	22	21087205	21173082	Loss	
139	22q11.22	0	4	NA17207, NA17156, NA17160, NA17216	306055	5	22	21173082	21479136	Loss	1,3
140	22q12.1	1	1	NA17269	5317	6	22	24823275	24828591	Loss	
141	Xp22.31	3	1	NA17247	1323457	14	X	6437549	7761005	Gain	4
142	Xp22.2	2	1	NA17214	216154	4	X	13283378	13499531	Gain	
143	Xq27.2	0	1	NA17270	381318	11	X	140088520	140469837	Loss	4

Table 3. Significantly enriched GO categories among genes that are encompassed by CNVs. There are a total of 8775 genes that are represented on the 50K Xba SNP array of which 364 are encompassed by the CNVs identified in this study. The enrichment score indicates the ratio between the actual number of interesting genes and the expected number of interesting genes from each GO category, with the P-value calculated based on a hypergeometric distribution

Gene category	# of ref. genes in category	# of genes in CNV	Expected # of genes in CNV	Enrichment	P-value
olfactory receptor activity	173	39	7.18	5.43	4.11E-19
sensory perception of smell	179	39	7.44	5.24	1.34E-18
sensory perception of chemical stimulus	186	39	7.7	5.06	4.82E-18
rhodopsin-like receptor activity	337	47	13.98	3.36	4.02E-14
G-protein coupled receptor activity	391	48	16.2	2.96	2.70E-12
G-protein coupled receptor protein signaling pathway	486	53	20.18	2.63	1.60E-11
sensory perception	355	43	14.71	2.92	6.31E-11
transmembrane receptor activity	590	57	24.46	2.33	3.60E-10
neurophysiological process	476	49	19.75	2.48	8.66E-10
receptor activity	824	64	34.2	1.87	1.81E-07
physiological response to stimulus	781	60	32.4	1.85	6.26E-07
chemoattractant activity	3	4	0.13	30.77	8.59E-07
organismal physiological process	1113	76	46.16	1.65	1.47E-06
cell surface receptor linked signal transduction	785	59	32.57	1.81	1.71E-06
chemokine activity	31	9	1.28	7.03	2.67E-06
response to stimulus	892	64	37	1.73	2.72E-06
intrinsic to membrane	1995	116	82.77	1.4	2.90E-06
chemokine receptor binding	32	9	1.33	6.77	3.63E-06
integral to membrane	1992	115	82.64	1.39	5.03E-06
glutathione transferase activity	8	5	0.34	14.71	6.91E-06
signal transducer activity	1045	70	43.35	1.61	1.13E-05
G-protein-coupled receptor binding	37	9	1.54	5.84	1.40E-05
membrane part	2126	119	88.2	1.35	1.54E-05
membrane	2505	134	103.9	1.29	2.83E-05
chylomicron	2	3	0.09	33.33	2.99E-05
T cell receptor complex	3	3	0.13	23.08	8.58E-05
transferase activity\, transferring alkyl or aryl [other than methyl] groups	20	6	0.81	7.41	9.73E-05

Gene category	# of ref. genes in category	# of genes in CNV	Expected # of genes in CNV	Enrichment	P-value
lipoprotein metabolism	18	5	0.73	6.85	5.71E-04
immunological synapse	5	3	0.22	13.64	7.94E-04
NAD[P]+-protein-arginine ADP-ribosyltransferase activity	1	2	0.04	50	9.65E-04
glycoprotein metabolism	63	9	2.62	3.44	1.06E-03
protein amino acid glycosylation	53	8	2.19	3.65	1.33E-03
biopolymer glycosylation	53	8	2.19	3.65	1.33E-03
glycoprotein biosynthesis	55	8	2.28	3.51	1.72E-03
T cell receptor binding	2	2	0.09	22.22	1.85E-03
Golgi to plasma membrane transport	2	2	0.09	22.22	1.87E-03
sterol transport	2	2	0.09	22.22	1.87E-03
cholesterol transport	2	2	0.09	22.22	1.87E-03
lipid transport	33	6	1.38	4.35	2.10E-03
taxis	74	9	3.05	2.95	3.13E-03
chemotaxis	74	9	3.05	2.95	3.13E-03
T cell activation	26	5	1.08	4.63	3.70E-03
external side of plasma membrane	8	3	0.35	8.57	4.03E-03
locomotory behavior	77	9	3.18	2.83	4.15E-03
protein amino acid N-linked glycosylation	18	4	0.73	5.48	5.10E-03
voltage-gated sodium channel activity	9	3	0.38	7.89	5.35E-03
cell recognition	10	3	0.43	6.98	7.53E-03
lymphocyte activation	42	6	1.76	3.41	7.54E-03
protein biosynthesis	216	17	8.95	1.9	7.73E-03
transferase activity\, transferring glycosyl groups	117	11	4.87	2.26	9.11E-03