

## Appendix A

### List of Oligonucleotides

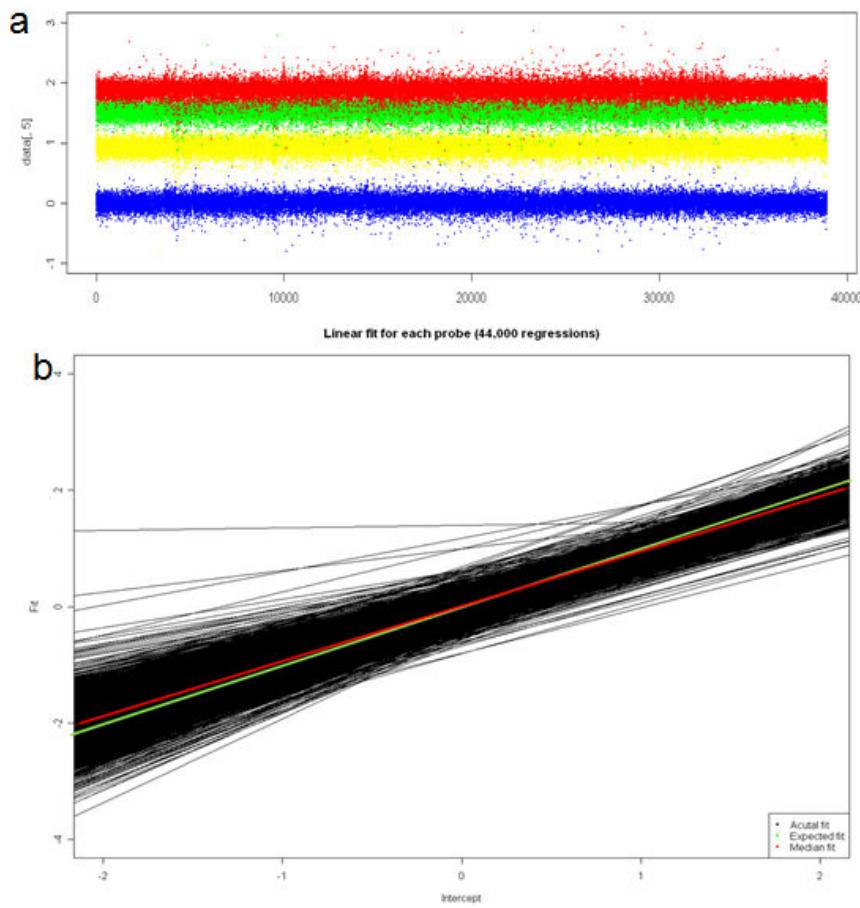
Primer/Probe Name	Sequence
<b>Long Range PCR for H6PD/SPSB1 Duplication</b>	
SPSB1-LR-f1	CTGTGTATCCAGTAGGTGCTTAATACTTT
SPSB1-LR-f2	CTTGGGAGAGATGTTTCTAGCAGTT
SPSB1-LR-f3	CTTGAGAAGCTCATTCTAACCTGTCTTAG
SPSB1-LR-f4	AGAAACTCCATCTACAGGTGCTGTC
SPSB1-LR-f5	TCTCTGTAAGTTCTGTGCCTCTCT
SPSB1-LR-f6	GTATCATGCACTAGTACTTTATGCCTTTT
SPSB1-LR-f7	AGTGTAACTCTGATGGTTTCATTAAATAG
SPSB1-LR-f8	CTTTATTAGGCAATTCTATGGTGACATC
SPSB1-LR-f9	CTATGTCCAAGAGACTGCCTTGTT
SPSB1-LR-r1	GTATGAGAGGCATCTAAAGCACCTG
SPSB1-LR-r2	CACCTGCTCACTGTAAGACTGGTACT
SPSB1-LR-r3	CCTCTGCAATGAGAACTACTGTAAGAGAAC
SPSB1-LR-r4	AGTGGGAAGAAGAGAAAAGGAAGTAAC
SPSB1-LR-r5	GTAACTGTGACGATAAAAGGGAAAGAAC
SPSB1-LR-r6	TAACATAACAACATCAGAATAACAGCAGTG
SPSB1-LR-r7	ATAACACCAGCTAACACAGCAATAAAC
SPSB1-LR-r8	GCTTAATAACAGCAATATAACACCATTGTA
<b>qPCR Validation Primers for H6PD/SPSB1 Duplication</b>	
SPSB1-I-f	GGGGGTCATCGTGAATAATG
SPSB1-I-r	CCACTTGGACCCTTCCACT
SPSB1-II-f	ATGGGTTGGTGTGGTTGT
SPSB1-II-r	GAGTGGAGTGCAGGAAGGAG
<b>qPCR Validation Primers for 10 CNV Loci</b>	
ED1176-loss-17-A-f	CGAGAGTGACTCCTGTTTCC
ED1176-loss-17-A-r	AGCCCCCTTCAAACCTAAA
ED1176-loss-17-B-f	TGGAGCCCCTCACCCTTA
ED1176-loss-17-B-r	TGCTTCTGGATTGTTGCTG
1085-loss-13-C-f	AGGCCAAATGAATCTGGAG
1085-loss-13-C-r	CTCGCTGAACCCTTGAAACA
3409-loss-23-B-f	CAGACTGCTGAAGCCATGAA
3409-loss-23-B-r	CAAACCACGAGCAAGCTGTA
4203-loss-28-B-f	TTGACCTAACCAAGGATGC
4203-loss-28-B-r	TTCTGGCTGGATGTTTC
5324-loss-21-B-f	GCATATTGCGATCCTGGAAT
5324-loss-21-B-r	CGTTGGTTGCTCCTCAAT
850-loss-22-B-f	AGCAGCCACATTGCCTTA
850-loss-22-B-r	GGCAGCCTAGTTTCATGG
3584-gain-11-A-f	AATTGGGGGTGTGATGATGT
3584-gain-11-A-r	CAAATAAAACGTCTGGTTGCAG
3584-gain-11-B-f	AAACTATGCCCTGGCCTTT
3584-gain-11-B-r	CGAAGCCACTCCTCTGACTC

Primer/Probe Name	Sequence
3766-gain-3-B-f	GCGCTGATGTTCTTCTCC
3766-gain-3-B-r	CAGCCATCAGCACTTCAGA
3789-loss-42-A-f	ACCGCGTGCTTAATTGTAT
3789-loss-42-A-r	TCTACCCCACCCCTTCTCCTT
3789-loss-42-C-f	AAAGCGATGCTCCACAAACT
3789-loss-42-C-r	CATCGAAGCACGTGAACATT
4179-gain-19-A-f	CTGAGCCCCGTGAGATCAGT
4179-gain-19-A-r	GAGAGTTCCCCAAAGCAG
<b>PCR genotyping primers for <i>CHL1</i> 5' deletion</b>	
CHL-fwd-5	TGATGATAATCCTCTTAGTTTCATT
wt-rev-2	GGGTTGGGAGTGGATTGTA
del-rev-1	AGCCACAGGTTGTTCTCCAC
<b>qPCR genotyping primers for <i>CHRFAM7A</i> deletion</b>	
CHRFAM7A-MGB-fwd1	AGTAATAGTGTAACTGTAACTTAAAATGTGTTACTTGT
CHRFAM7A-MGB-rev1	AGCCGGGATGGCTCGAT
B2M-MGB-fwd1	TGGGTTCATCCATCCGACATT
B2M-MGB-rev1	AGACAAGTCTGAATGCTCCACTTT
B2M_probe1	VIC-ATTCTTCAGTAAGTCAACTTC
CHRFAM7A_probe1	6FAM-TCCTGACTGTACACATAAAA

## **Appendix B**

### **ABCA13 Custom Designed Oligonucleotide Array Validation**

To identify copy number changes at the *ABCA13* region in families with schizophrenia, we designed a custom oligo array (Agilent Technologies) with 34,036 probes covering a 1.5-Mb interval at chromosome 7p12 spanning *ABCA13*. Validation experiments were performed to assess the probe dose response on the custom array by hybridising a series of 1x, 2x, 3x and 4x reference DNA (labelled in Cy5) against 1x reference DNA (labelled in Cy3). The expected log<sub>2</sub>ratio of each probe would be at 0, 1, 1.58 and 2 for the 4 experiments (corresponding to the 1x: 2x: 3x: 4x dosage). Analysis shows that majority of the oligo probes fall on the dosage response curves for the expected labelled DNA amount (Figure A1.1)



**Figure B-1 Dose response of the custom ABCA13 oligo array. a)** A series of experiments were performed with 1x (blue), 2x (yellow), 3x (green) and 4x (red) reference DNA hybridised against 1x reference DNA. The expected log<sub>2</sub>ratio of each probe for the 4 experiments would be 0, 1, 1.58 and 2 respectively. The dose response for the majority of probes falls on the expected values. (x-axis: log<sub>2</sub>ratio, y-axis: genomic coordinates of the probes) **b)** Best-fit linear regression lines were generated for the expected and actual dosage response. The red line represents the expected fit. The actual fit for each probe is shown in black, and the median best-fit linear regression line for all probes is shown in green. The actual median fit (green) is close to the expected fit (red).

## **Appendix C**

### **Whole Genome Tiling Path (WGTP) Array Quality Control**

#### **C1 Data Normalization**

Data normalization, which includes background subtraction and spatial normalization, were performed as previously described (Fiegler et al. 2006, Redon et al. 2006). Some array profiles in our dataset demonstrated genomic “waves” in the signal intensities, a phenomenon which appeared to be a general feature of array CGH datasets (Marioni et al. 2007). One characteristic of these “waves” were their correlation to GC content of the array probes. All array profiles were therefore GC-normalized: the log2ratio of each clone in every sample is normalized against the GC content of the BAC clone using a custom PERL script (developed by Dr Richard Redon and Armand Valsesia).

#### **C2 Clone Filtering**

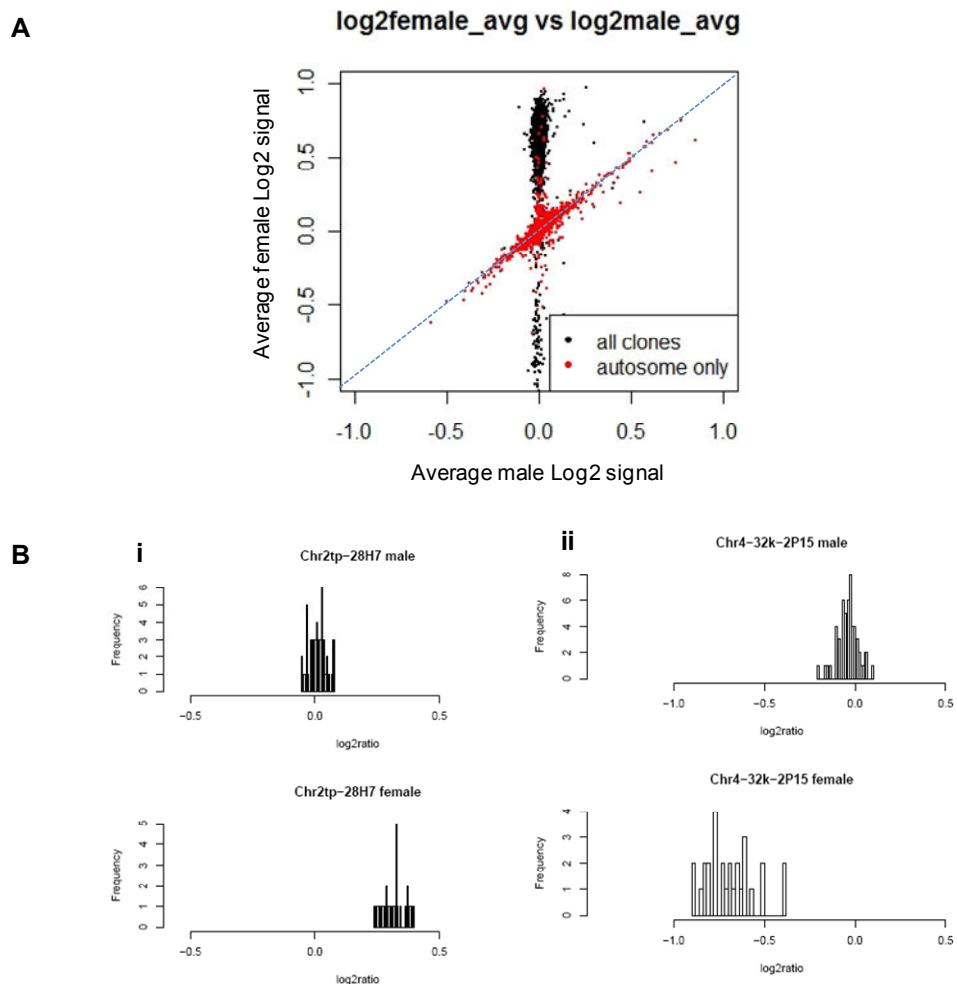
Each WGTP array constructed at the Sanger Institute is spotted with 29043 large insert clones from the “Golden Path” library (Fiegler et al., 2006). To eliminate possible artefacts due to individual clones, the following inspection and filtering steps were taken.

##### *C2.1 Inconsistent and frequently excluded clones*

Clones that showed poor morphology, spot intensities or low signal to background noise in individual experiment, or behaved inconsistently between dye swap experiments of a given sample, would be flagged as being excluded from the analysis of that DNA sample. Clones that were frequently excluded (in >75% DNA samples in both case and control cohorts) were subsequently filtered from the CNV dataset. 93 clones were filtered based on this criterion.

##### *C2.2 Autosomal clones with homology to X/Y chromosome*

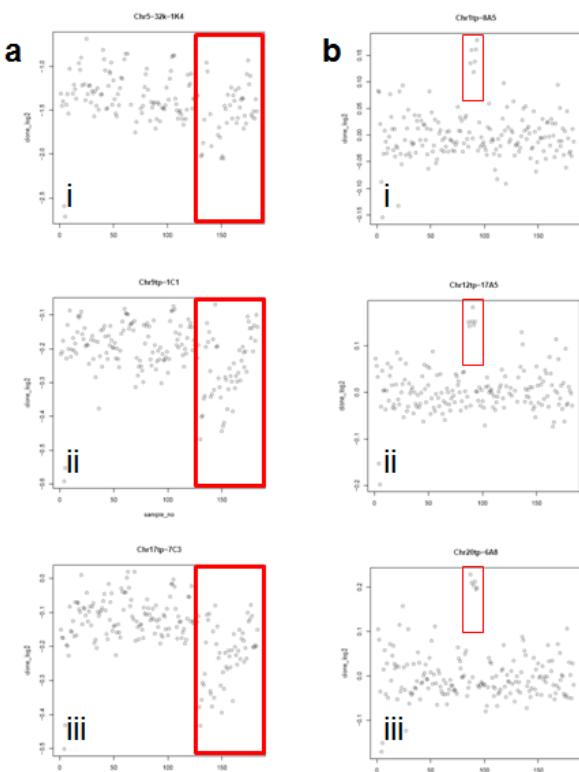
Autosomal clones that have homology to either sex chromosome would give variable log<sub>2</sub> signal ratios depending on whether hybridisations were performed with a male or female test DNA. These “pseudo-X/Y clones” were detected using a custom R script based on the difference between the average log<sub>2</sub>ratio of all female versus male test DNA hybridisations (see Fig C-1). 100 clones with a difference of >0.1 between average male and female log<sub>2</sub>ratio were excluded using this criterion. Examples of the histograms of two removed clones are shown in Fig C-1b.



**Fig C-1 Autosomal clones with homology to X/Y chromosomes.** Autosomal clones with homology to X/Y chromosomes were detected using the average log2ratio values of all female versus all male test DNA experiments. (a) A plot of the average female log2ratio against the average male log2ratio for all clones. Majority of the autosomal clones lie on the  $x=y$  line (blue line). Clones above this line are X-chromosome clones or pseudo-X autosomal clones (filtered). Clones below this line are Y-chromosome clones or pseudo-Y autosomal clones (filtered). (b) Histograms of two filtered autosomal clones. i) a pseudo-X clone Chr2tp-28H7 and ii) a pseudo-Y clone Chr4-32K-2P15 (top panel: experiments from male DNAs, bottom panel: experiments from female DNAs)

### C2.3 Artefacts due to hybridisation date

Clones were inspected with respect to the dates of hybridisation to detect artefacts due to environmental factors (e.g. variations in array batch used, ozone level and humidity level). When a set of clones with such artefact pattern was detected, the remaining 29,043 clones were screened by means of Pearson correlation coefficients to check for similar artefact patterns. A list of 110 clones showing two systematic date-specific artefact patterns were removed from further analysis (Figure C-2).



**Fig C-2 BAC clones showing systematic date-specific artefact patterns.** Two systematic date-specific artefact patterns were detected. Each plot shows clone log2ratio fluorescence signal against the DNA sample sorted by dates of hybridisation. (a) Hybridisations carried out prior to 05-Jul-2007 compared to those after 05-Jul-2007 i) Chr5-32K-1K4, ii) Chr5tp-1C1 and iii) Chr17tp-7C3. (b) Hybridisations carried out on Jun-20-2007 & June-21-2007 compared to the rest of hybridisations i) Chr1tp-8A5, ii) Chr12tp-17A5 and iii) Chr20tp-6A8).

### C2.4 Sex chromosome artefacts in male Lothian Birth Control Samples

23 samples from the Lothian Birth Control (LBC) cohort demonstrated a mosaic Y chromosome pattern, such that clones on Y chromosome exhibited lower log2ratio fluorescence signal than in a typical male DNA sample hybridisation. These chromosomal artefacts may be due to Y chromosome loss from the LBC subjects, who were at relatively advanced age (~90 years old) at the time of DNA extraction. All non-autosomal clones were removed from these 23 LBC male samples from further analysis.

## Appendix D

### WGTP Array Supplementary Tables and Figures

**Table D-1 Number and types of CNVs (Gain/ Loss) per DNA sample.**

(a) SCZ samples (n=91) and (b) LBC samples (n=92)

a)

SCZ Sample	Gain	Loss	Total				
211	29	22	51	3975	36	12	48
220	27	17	44	3976	27	13	40
241	20	13	33	4042	44	25	69
297	42	21	63	4100	40	36	76
302	56	24	80	4101	36	22	58
323	31	37	68	4111	48	31	79
358	34	17	51	4160	46	31	77
385	55	45	100	4179	27	14	41
513	52	31	83	4203	43	27	70
621	36	39	75	4355	43	30	73
665	27	14	41	4398	37	38	75
850	33	16	49	4495	37	36	73
899	36	26	62	4710	42	28	70
1085	33	19	52	4716	51	33	84
1278	54	24	78	4748	54	22	76
1295	26	19	45	4773	47	19	66
1784	28	26	54	5307	34	26	60
3071	58	41	99	5324	57	24	81
3138	41	30	71	5379	41	20	61
3141	48	44	92	5386	55	40	95
3198	33	16	49	5390	65	30	95
3199	47	24	71	5446	46	24	70
3333	36	37	73	5467	48	41	89
3341	27	18	45	5541	24	20	44
3377	34	23	57	5562	20	18	38
3409	24	20	44	5587	41	39	80
3443	44	33	77	5660	61	22	83
3503	45	26	71	5758	50	30	80
3559	53	41	94	5792	29	15	44
3572	34	23	57	6273	59	32	91
3581	51	37	88	6276	26	14	40
3584	45	19	64	6289	73	34	107
3594	28	15	43	6596	29	17	46
3652	39	33	72	6626	53	46	99
3661	33	26	59	6634	41	32	73
3703	30	13	43	6638	32	25	57
3751	54	28	82	6667	33	16	49
3766	31	19	50	6744	39	20	59
3776	24	43	67	7019	32	22	54
3789	54	35	89	7103	38	16	54
3802	41	21	62	7132	35	20	55
3812	57	23	80	7180	10	21	31
3815	47	28	75	7183	24	18	42
3857	46	38	84	7287	56	43	99
3931	54	17	71	7294	40	38	78
3945	24	25	49	<b>Grand Total</b>	<b>3650</b>	<b>2386</b>	<b>6036</b>

b)

LBC Sample	Gain	Loss	Total	D245	25	15	40
D003	51	28	79	D246	32	26	58
D007	39	29	68	D255	59	45	104
D008	61	26	87	D263	54	41	95
D011	56	41	97	D269	43	25	68
D012	34	22	56	D272	41	35	76
D015	40	12	52	D283	38	33	71
D019	46	27	73	D287	52	45	97
D020	41	25	66	D301	41	27	68
D026	42	30	72	D328	34	37	71
D027	24	27	51	D331	53	22	75
D032	29	22	51	D332	55	38	93
D033	58	34	92	D337	51	41	92
D035	46	26	72	D341	69	34	103
D045	37	36	73	D357	42	27	69
D049	31	18	49	D365	44	33	77
D052	31	23	54	D371	35	34	69
D053	33	32	65	D379	49	32	81
D055	37	16	53	D390	41	29	70
D058	58	34	92	D391	45	39	84
D059	47	59	106	D392	47	31	78
D060	47	32	79	D399	42	19	61
D062	52	33	85	D402	86	34	120
D075	45	39	84	D435	50	38	88
D078	56	34	90	D455	66	50	116
D086	37	37	74	D477	43	22	65
D088	44	20	64	D485	46	47	93
D098	44	31	75	D489	72	36	108
D107	56	30	86	D508	44	51	95
D127	52	30	82	D514	52	33	85
D158	29	17	46	D515	28	24	52
D161	26	6	32	D518	26	11	37
D162	28	16	44	D520	18	17	35
D164	37	18	55	D521	24	26	50
D168	29	18	47	D522	31	30	61
D176	31	8	39	D531	36	20	56
D209	34	25	59	D538	37	26	63
D211	35	32	67	D548	44	29	73
D220	35	39	74	LBC1	23	20	43
D223	46	38	84	LBC10	76	16	92
D224	45	38	83	LBC2	52	34	86
D226	43	34	77	LBC3	32	31	63
D234	45	28	73	LBC4	28	33	61
D235	50	41	91	LBC6	31	22	53
D237	54	31	85	LBC8	57	17	74
D242	44	32	76	LBC9	53	25	78
D244	41	15	56	<b>Grand Total</b>	<b>3973</b>	<b>2689</b>	<b>6662</b>

**Table D-2 113 Variants Specific to the SCZ Cohort**

	Chr Coordinates	Clones	Genes
1	chr1:2062380-2546655	Chr1tp-25A3, Chr1tp-25A4, Chr1tp-1A1, Chr1tp-1A2, Chr1tp-25A2, Chr1tp-25A1	PEX10, C1orf93, SKI, C1orf86, HES5, RER1, PRKCZ, TNFRSF14, MORN1, MMEL1, PLCH2, PANK4
2	chr1:2847618-3355092	Chr1tp-25B4, Chr1tp-1A8, Chr1tp-1A9, Chr1tp-25B3, Chr1tp-1A7	ACTRT2, PRDM16
3	chr1:9177728-9373336	Chr1tp-23A3, Chr1tp-25D10, Chr1tp-1C7	H6PD, SPSB1
4	chr1:68325266-68897278	Chr1tp-4A8, Chr1tp-20H2, Chr1tp-28G5, Chr1tp-23D1, Chr1tp-28G4, Chr1tp-28G3, Chr1tp-36B4	DEPDC1, RPE65, GPR177
5	chr1:71346768-71893785	Chr1tp-4C1, Chr1tp-4B12, Chr1tp-38E10, Chr1tp-18C6	NEGR1
6	chr1:105728511-105980971	Chr1tp-6E3, Chr1tp-6E4	CHIA, C1orf88, DENND2D, OVGP1,
7	chr1:111515826-111802379	Chr1tp-30E4, Chr1tp-6H4	WDR77, CHI3L2, ATP5F1, CEPT1
8	chr1:205624502-205991690	Chr1tp-21E1, Chr1tp-11G7	CR1L, CR2, CR1
9	chr1:232939949-233194586	Chr1tp-13G11, Chr1tp-13H1	
10	chr1:246013817-246295055	Chr1tp-15A5, Chr1tp-15A4	OR2AK2, OR2AJ1, OR2L13, OR2L3, OR14A16, OR2L8, OR2L2, OR2W3, TRIM58, OR2L5, OR11L1
11	chr2:731429-1026826	Chr2tp-3D6, Chr2tp-29G2, Chr2tp-31F10	SNTG2
12	chr2:37650158-37992443	Chr2tp-16B5, Chr2tp-5F10, Chr2tp-22C3	CDC42EP3, SLC30A6
13	chr2:57138035-57341900	Chr2tp-24G11, Chr2tp-1D9	
14	chr2:86130488-86470772	Chr2tp-5F6, Chr2tp-22D2	REEP1, PTCD3, POLR1A, IMMT, MRPL35
15	chr2:127333466-127814482	Chr2-32k-1B7, Chr2tp-7B12, Chr2tp-12G11, Chr2tp-7E6	ERCC3, BIN1, MAP3K2, CYP27C1
16	chr2:132668884-132859586	Chr2tp-16E12, Chr2tp-11B5	
17	chr2:188772739-189096411	Chr18tp-1A6, Chr2tp-7E12, Chr2tp-4H8	GULP1
18	chr2:213735598-214074098	Chr2-32k-4O4, Chr2tp-29F9	SPAG16
19	chr3:8686094-8880344	Chr3tp-27B5, Chr3tp-4D7	OXTR, CAV3, C3orf32
20	chr3:76248984-77652531	Chr3tp-11A8, Chr3tp-11H6, Chr3tp-4H9, Chr3-32k-6M10, Chr3tp-27A6	ROBO2
21	chr3:86998287-87362355	Chr3-32k-2L14, Chr3tp-20B2, Chr3tp-18G12	CHMP2B, VGLL3
22	chr3:139094479-139416525	Chr3tp-10F10, Chr3tp-18G6	ARMC8, DZIP1L, CLDN18, A4GNT, DBR1
23	chr3:143326112-143612526	Chr3tp-11B1, Chr3tp-1G2	XRN1, TFDP2, GK5

	<b>Chr Coordinates</b>	<b>Clones</b>	<b>Genes</b>
24	chr3:189546565-189761158	Chr3tp-9H12, Chr3tp-18F1	LPP
25	chr4:7039049-7320802	Chr4tp-21E8, Chr4tp-7G10	SORCS2, TBC1D14, GRPEL1, CCDC96
26	chr4:11668862-11990799	Chr4-32k-3N20, Chr4tp-9C5, Chr4tp-1B4	
27	chr4:28258974-28570510	Chr4tp-6E1, Chr4tp-16E1, Chr4tp-6C4	
28	chr4:115564534-115809774	Chr4tp-13A7, Chr4tp-14D9, Chr4-32k-4B10	UGT8
29	chr4:162138593-162324657	Chr4tp-17A12, Chr4tp-3E1	
30	chr4:162734833-163023593	Chr4tp-3D7, Chr4tp-25C4, Chr4tp-21C3	FSTL5
31	chr4:168885634-169288268	Chr4tp-10G12, Chr4tp-9F9	ANXA10
32	chr4:189077762-189851429	Chr4tp-17E1, Chr4tp-17D1, Chr4tp-7D1, Chr7tp-16B7, Chr4tp-5C8, Chr4tp-24A9	TRIML2, ZFP42, TRIML1
33	chr5:9850992-10044066	Chr5tp-18A3, Chr5tp-5H11	
34	chr5:25051484-25333181	Chr5tp-6H2, Chr5tp-3E11	
35	chr5:61438895-61723574	Chr5tp-19D4, Chr5tp-17G10	DIMT1L, KIF2A
36	chr5:84073606-84367471	Chr5tp-24B1, Chr5tp-6E5	
37	chr5:101134363-101393863	Chr5tp-25F12, Chr5tp-2A1	
38	chr5:104334472-104660077	Chr5tp-23B3, Chr5tp-1B3, Chr5tp-15B7, Chr5tp-23E12	
39	chr5:110212743-110577198	Chr5tp-15G1, Chr5tp-24F9, Chr5-32k-5C18	WDR36, TSLP
40	chr6:1994541-2304998	Chr6tp-4B5, Chr6tp-4D5, Chr6tp-5A4	GMDS
41	chr6:7088879-7351773	Chr6tp-1C9, Chr6tp-3B7	RREB1, CAGE1, RIOK1, SSR1
42	chr6:11754000-11930337	Chr6tp-5E7, Chr6tp-6A12	C6orf105
43	chr6:22212251-22607763	Chr6tp-3C9, Chr6tp-6A8, Chr6tp-3F3, Chr6tp-3B3	PRL
44	chr6:34358245-34603317	Chr6tp-2C4, Chr6tp-4H8	NUDT3, PACSIN1, RPS10
45	chr6:97469365-98475130	Chr6tp-18H7, Chr6tp-15B2, Chr6tp-8A7, Chr6tp-16H7, Chr6tp-17D11, Chr6tp-17A3, Chr6tp-11E8, Chr6tp-16D3, Chr6tp-13C2, Chr6tp-12G11	KLHL32, C6orf167
46	chr6:143958314-144219762	Chr6tp-11C7, Chr6tp-18B6	PHACTR2, LTV1
47	chr6:160857306-161154655	Chr6tp-14D11, Chr6tp-17C6	LPA, PLG
48	chr6:165840859-166673790	Chr6tp-8B6, Chr6tp-19H8, Chr6tp-14C5, Chr6tp-15E7, Chr6tp-8C3, Chr6tp-19A1, Chr6tp-10C12, Chr6tp-14F5	SFT2D1, T, PDE10A, PRR18
49	chr7:885103-1354746	Chr7tp-14F1, Chr7tp-4D8	CYP2W1, CENTA1, UNCX, C7orf50, GPR146, COX19, GPER, C7orf20, ZFAND2A
50	chr7:17320942-17609921	Chr7tp-4C1, Chr7tp-5D12	AHR

	<b>Chr Coordinates</b>	<b>Clones</b>	<b>Genes</b>
51	chr7:17795286-18873121	Chr7tp-23G6, Chr7tp-18H11, Chr7tp-10E12, Chr7tp-3E3, Chr7tp-22H9, Chr7tp-21E3, Chr7tp-1H3, Chr7tp-17H9, Chr7tp-5E2, Chr7-32k-2I12	SNX13, HDAC9
52	chr7:21281130-21501769	Chr7tp-21E8, Chr7tp-21A5	SP4
53	chr7:23686581-23846847	Chr7tp-14F2, Chr7tp-19E4	C7orf46, STK31
54	chr7:75289669-75520758	Chr7-32k-6E9, Chr7tp-17H6	TMEM120A, RHBDD2, POR, STYXL1, MDH2
55	chr7:88344724-88962788	Chr7tp-19G12, Chr7tp-10D4, Chr7tp-18A2, Chr7tp-18C10, Chr7tp-20H7, Chr7tp-22H12, Chr7tp-16C8	ZNF804B
56	chr7:93658283-94064936	Chr7tp-1E6, Chr7tp-17D3, Chr7tp-21F8	SGCE, CASD1, COL1A2
57	chr7:123672713-123975901	Chr7tp-3C10, Chr7tp-9B6, Chr7-32k-3I18	CENTG3
58	chr7:124338397-124505938	Chr7tp-3G9, Chr7tp-3F4	CENTG3, POT1
59	chr8:9670799-9988807	Chr8tp-20B12, Chr8tp-6F6	TNKS, MSRA
60	chr8:13581607-13896999	Chr8tp-7A2, Chr8tp-7D2	
61	chr8:47038753-47556339	Chr8tp-9B4, Chr8tp-19B9, Chr8tp-13H11, Chr8tp-19C4	
62	chr8:47728697-47982207	Chr8tp-5G11, Chr8tp-3C12	
63	chr8:48950846-49170567	Chr8tp-17F4, Chr8tp-16B12, Chr8tp-3E4	PRKDC, UBE2V2, MCM4
64	chr8:145496837-145824934	Chr8-32k-5I13, Chr8tp-16F9	GPT, CYHR1, GPR172A, PPP1R16A, KIFC2, FOXH1, HSF1, SCRT1, NFKBIL2, RECQL4, MFSD3, CPSF1, SLC39A4, DGAT1, ADCK5, LRRC14, VPS28, FBXL6, LRRC24
65	chr9:76652583-77315436	Chr9tp-4G5, Chr9tp-5C11, Chr9tp-6D10, Chr9tp-8C4, Chr9tp-5C12	C9orf41, C9orf95, TRPM6, OSTF1, C9orf40
66	chr9:137112070-137464520	Chr9tp-10A3, Chr9tp-7H9	C9orf62, OLFM1
67	chr9:137590464-137867305	Chr9tp-3C2, Chr9tp-3B10	LCN9, CAMSAP1, SOHLH1, KCNT1, PAEP, GLT6D1
68	chr10:3722438-4002432	Chr10tp-9H1, Chr18tp-10B5, Chr10tp-4H7	KLF6
69	chr10:42524238-42830197	Chr10tp-10B2, Chr10tp-3D8, Chr10tp-12D4	BMS1
70	chr10:76904943-77122607	Chr10tp-13C7, Chr10tp-6G6	C10orf11
71	chr11:31339431-31549584	Chr11tp-13E3, Chr11tp-13E8	DPH4, DCDC1, IMMP1L, ELP4
72	chr11:50013000-50737095	Chr11tp-7H3, Chr11tp-11A11, Chr11tp-11E7, Chr11tp-16H3, Chr11tp-3E2, Chr11tp-2B4, Chr11tp-5F6	OR4A5

	<b>Chr Coordinates</b>	<b>Clones</b>	<b>Genes</b>
73	chr11:58456668-58635415	Chr11tp-2G5, Chr11tp-13E10	FAM111B, GLYATL1
74	chr12:17761730-18154145	Chr12tp-12A7, Chr12tp-5D12	RERGL
75	chr12:51437221-51449660	Chr12tp-10A4, Chr12tp-11A11	KRT76
76	chr12:131115602-131701464	Chr0-32k-1C11, Chr12tp-7A9	EP400NL, GALNT9, EP400, MUC8, NOC4L, DDX51
77	chr13:18881229-19386914	Chr13tp-5D8, Chr13tp-1B2, Chr13tp-8F2, Chr13tp-10A5	MPHOSPH8, PSPC1, ZMYM5, TPTE2
78	chr13:51626371-51966133	Chr13tp-11D8, Chr13tp-11E5	THSD1, VPS36, CKAP2, NEK3
79	chr13:82035430-82280653	Chr13tp-8E10, Chr13tp-1B10	
80	chr13:86846920-87112550	Chr13tp-1F2, Chr13tp-5B8	
81	chr13:112568275-112939870	Chr13tp-1D12, Chr13tp-4E6, Chr13tp-3B4, Chr13tp-10G6	PROZ, ATP11A, MCF2L, PCID2, F10, CUL4A, F7
82	chr13:113166605-113658050	Chr13tp-11B6, Chr13tp-11A8, Chr13tp-3D4	TMCO3, TFDP1, GAS6, FAM70B, DCUN1D2, GRK1, ATP4B
83	chr14:74945402-75396561	Chr14tp-8C7, Chr14tp-10B10, Chr14tp-3B9	JDP2, C14orf1, TTL5, FLVCR2, BATF
84	chr14:103592318-103986703	Chr14-32k-3E3, Chr14-32k-2C12, Chr14tp-2C5	KIF26A
85	chr15:22894873-23111579	Chr15tp-7D1, Chr15tp-11C3	
86	chr15:25060215-25335769	Chr15tp-7C1, Chr15tp-4A1	GABRG3
87	chr15:41610450-41935933	Chr15tp-3H4, Chr15tp-3G7	STRC, CATSPER2, SERF2, CKMT1B, MAP1A, ELL3, SERINC4, HISPPD2A, MFAP1, CKMT1A, PDIA3, WDR76
88	chr15:83451942-84109983	Chr15tp-10F9, Chr15tp-10H5, Chr15tp-5E3, Chr15tp-7G3, Chr15tp-11G11	PDE8A, AKAP13, KLHL25
89	chr15:99707205-100036184	Chr15tp-1C4, Chr15tp-5B5	TM2D3, PCSK6, TARSL2
90	chr16:2870892-3098404	Chr16tp-13C6, Chr16-32k-2L14	KREMEN2, HCFC1R1, CLDN9, IL32, MMP25, CCDC64B, TNFRSF12A, FLYWCH1, ZSCAN10, PAQR4, FLYWCH2, THOC6, PKMYT1, CLDN6
91	chr16:57136506-57433538	Chr16tp-12E8, Chr16tp-3F2	GOT2, SLC38A7, CNOT1
92	chr17:61780311-61961765	Chr17tp-6A10, Chr17tp-11D10	PRKCA
93	chr17:74849624-75362772	Chr17-32k-2I12, Chr17-32k-2J21, Chr17tp-6B11	ENPP7
94	chr17:76741026-77562577	Chr17-32k-2P9, Chr17tp-3H2, Chr17-32k-2B20	C17orf55, NPB, SLC38A10, MRPL12, NOTUM, C17orf70, BAHCC1, THOC4, AATK, CCDC137, SLC25A10, HGS, FSCN2, ARL16, C17orf90, ANAPC11, TMEM105, AZI1, P4HB, C17orf56, PDE6G, PYCR1, TSPAN10, MAFG, ASPSCR1, ACTG1, ARHGDIA, PCYT2, DYSFIP1, NPLOC4, SIRT7

	<b>Chr Coordinates</b>	<b>Clones</b>	<b>Genes</b>
95	chr18:37767209-38078089	Chr18tp-5C12, Chr18-32k-1L20	PIK3C3
96	chr18:64349737-64656600	Chr18tp-1H10, Chr18tp-5H6	CCDC102B, TXNDC10
97	chr19:20265108-20637787	Chr19tp-6G2, Chr19tp-2B11, Chr19tp-6G2, Chr19tp-8A10	ZNF826
98	chr19:50553125-51160225	Chr19-32k-2E17, Chr19-32k-3I5	GPR4, DMPK, CD3EAP, IRF2BP1, VASP, QPCTL, EML2, NOVA2, NANOS2, DMWD, PPP1R13L, GIPR, RSHL1, SYMPK, FOXA3, FOSB, SIX5, ERCC1, OPA3, SNRPD2, ERCC2, RTN2
99	chr20:14561835-14936485	Chr20tp-5G6, Chr20tp-5G3, Chr20tp-6C12, Chr20tp-1H11	MACROD2
100	chr20:29267570-29480559	Chr20tp-6F10, Chr20tp-5G2	DEFB121, DEFB117, DEFB115, DEFB119, DEFB116, DEFB118
101	chr20:61940840-62284575	Chr20tp-1F12, Chr20tp-4B8, Chr20tp-2H9, Chr20tp-2H7	RGS19, MYT1, SAMD10, TCEA2, PRPF6, UCKL1, PRR17, DNAJC5, SOX18, OPRL1, ZNF512B, UCKL1OS, C20orf135, TPD52L2, NPBWR2
102	chr20:62108666-62376639	Chr20tp-2H9, Chr20tp-2H7, Chr20tp-6G1,	OPRL1, SOX18, RGS19, MYT1, TCEA2, PRPF6, ZNF512B, PRR17, NPBWR2, PCMTD2
103	chr21:34576662-34852873	Chr21tp-3F8, Chr21tp-3A5	KCNE2, RCAN1, C21orf51, KCNE1
104	chr21:44384865-44825939	Chr21-32k-2C5, Chr1-32k-7C17, Chr21-32k-2E17, Chr21-32k-2G11	TRPM2, C21orf33, ICOSLG, C21orf2, KRTAP10-2, PFKL, KRTAP10-5, KRTAP10-1, AIRE, C21orf29, DNMT3L, C21orf90, KRTAP10-3, LRRC3, KRTAP10-4
105	chr21:45508487-45829731	Chr21-32k-1N9, Chr21-32k-1B12, Chr21-32k-1L4	C21orf93, SLC19A1, C21orf123, COL18A1, POFUT2
106	chr22:39890708-40040762	Chr22tp-9A1, Chr22tp-8B6	CHADL, RANGAP1, L3MBTL2, EP300, ZC3H7B
107	chr22:46219057-46441810	Chr22tp-6F7, Chr22tp-4A6, Chr22- 32k-1H11, Chr22tp-4H5	
108	chr22:46903974-46908346	Chr22tp-7A3, Chr22tp-7A10	
109	chrX:51220257-51533184	ChrXtp-21H12, ChrXtp-20B10,	NUDT11, GSPT2
110	chrX:56037801-56325064	ChrXtp-4E12 ChrXtp-11B12, ChrXtp-6A3, ChrXtp- 21B4	KLF8
111	chrX:79042086-79447858	ChrXtp-9E7, ChrXtp-23D8, ChrXtp- 10A5, ChrXtp-4H6, ChrXtp-11A8	TBX22
112	chrX:105332666-105446815	ChrXtp-11H2, ChrXtp-7E1	MUM1L1
113	chrY:26625200-26913359	ChrYtp-1G9, ChrYtp-2D10	

**Table D-3 CNV Genotyping of WGTP Data With Bivariate Clustering**

31 regions demonstrate significant difference (p-value < 0.05) in the CNV genotype distributions between SCZ and LBC following bivariate cluster of log2ratio. For each region the p-value of chi-squared test was shown.

	clones	SCZ class	LBC class	p-val	chrom coordinates	Genes
1	Chr3-32k-5N10, <b>Chr3tp-9E3</b> ,Chr3tp-7F8	4_-25_42	1_-16_62	0.02711	chr3:46141:343409	CHL1*
2	Chr17p-8G2,Chr17p-12B1,Chr17p-2F6,Chr17p-14B12, <b>Chr17tp-14B6</b> ,Chr17p-2F9,Chr17p-1D11,Chr17p-11E1	31_-29_19_-22_26_23	0.03517	chr17:41747597-42632332	ARL17P1, LRRK37A2, AC139026.2, NS*, AC004098.1, WNT3, WNT9B, AC005670.1, GOSR2, RPRML, AC06815.2.11, LRRK37A4, CDC27	
3	Chr15-32k-11I4, <b>Chr15tp-7E12</b> ,Chr15tp-7G6,Chr15tp-9C6,Chr15tp-1G5,Chr15tp-1G2,Chr15tp-4F3,Chr15tp-12A3,Chr15tp-7H3	17_-72	7_-83	0.04511	chr15:26714864-28805754	AC055876.16, AC055876.16, AC055876.16, AC055876.1, APBA2, AC024474.8, NDNL2, AC102941.9, AC120451.9, TIP1, AC120452.2, AC111152.2, AC120451.9, AC120451.9, AC120451.9, AC120451.9, AC135731.6, AC019322.8, <b>CHRFAN7A*</b> , AC019322.8, AC019322.8, AC026150.7, AC026150.7, AC026150.7, AC026150.7, ARHGA11B, AC091057.6, AC091057.6, AC091057.6
4	<b>Chr13-32k-3D13</b> ,Chr13tp-7D5	50_-15_9	71_-7_0	0.00044	chr13:44704567-483370518	GTF2F2, AL138963.20, TPT1, AL138963.20, AL627107.32, SLC25A30, AL606514.3, COG3, AL139326.15, AL139326.15, SPERT, AL139320.18, AL157819.15, ZC3H13, CPB2, LCP1, AL137141.10, AL139801.17, AL137141.10, AL139801.17, C13orf18, AL139801.17, N/A, AL138686.14, RP11-159B4.3, N/A, LRCH1, ESD, HTR2A, AL160397.17, AL160397.17, AL139180.17, SUCL2, NUDT15, MED4, N/A, AL158196.24, ITM2B, RB1, AL1392048.9, P2RY5, AL136960.4, RCBTB2, CYSLTR2, AL137118.20, GTF2F2, AL138963.20, TPT1, AL138963.20, AL1627107.32, SLC25A50, AL606514.3, COG3, AL139326.15, AL139326.15, SPERT, AL139320.18, AL157819.15, ZC3H13, CPB2, LCP1, AL137141.10, AL139801.17, C13orf18, AL139801.17, N/A, AL139801.17, NUDT15, MED4, N/A, AL158196.24, ITM2B, RB1, AL1392048.9, RCBTB2, CYSLTR2, AL137118.20
5	<b>Chr8-32k-4F15</b> ,Chr8tp-21E9,Chr8tp-16F9,Chr8-32k-5I13	63_-16	75_-1	0.00044	chr8:144483554-145824934	TOP1MT, AC0087793.17, N/A, RHPNL, MAF4, ZC3H3, AC067930.7, GSDMDC1, N/A, C8orf73, N/APRT1, EEF1D, TIGD5, PYCRL, TSTA3, ZNF623, ZNF707, AC105219.6, MAPK15, FAM83H, N/A, AC105219.6, SCRIB, hsa-mir-937, PUF60, NRB22, PLEC1, hsa-mir-661, PARP10, GRIN1/A, SPATC1, AC105322.16, OPLAH, EXOSC4, GPAA1, CYCL1, SHARPIN, MAF1, KIAA1875, C8orf30A, AC145291.2, AC145291.2, AC145291.2, C8orf30A, BOP1, SCKA, HSFL, N/A, DGAT1, SCRT1, AF20589.5, FBX16, GPR172A, ADCR5, CPSF1, hsa-mir-939, SLC39A4, VPS28, NFKBIL2, GPT, AC084125.9, CYHL1, KIFC2, FOXKH1, PPARI6A, MFSD3, RECQL4, LRRK14, LRRK24, AC084125.9, AC084125.9

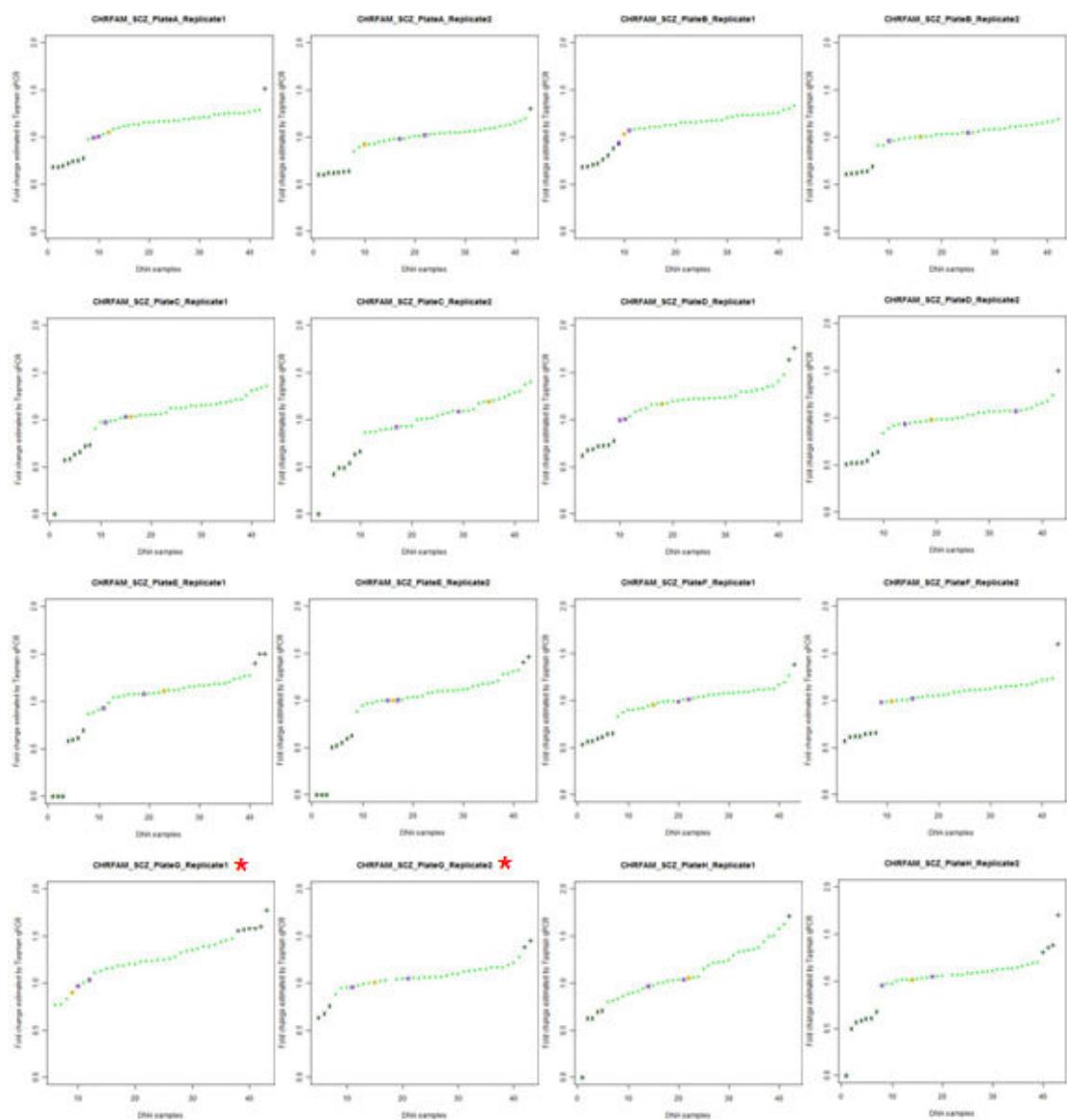
clones	SCZ class	LBC class	p-val	chrom coordinates		Genes			
				start	end				
6	Chr9tp-3C2, Chr9tp- <b>3E10</b> , Chr9tp-2k18	54_14	76_2	0.00132	chr9:137679970-138276710	LCN9, SOHH1, KCNT1, CAMSAP1, QSOX2, UBAC1, BTBD14A, N/A, LHX3, N/A			
7	<b>Chr22tp-743</b> , Chr22tp-7A10	72_9	91_0	0.00346	chr22:46903974-46908346	N/A			
8	<b>Chr4-32k-3B10</b> , Chr4-32k-4N21	0_64	13	1_84	2	0.0037	chr4:135111708-135433169	N/A	
9	<b>Chr7tp-5H9</b> , Chr7tp-11F4	35_34	0	18_50	2	0.00526	chr7:69823647-70123482	AUTS2	
10	<b>Chr19tp-6D4</b> , Chr19-32k-3I21, Chr19-32k-2N13, Chr19-32k-3O11	52_16	75_5	0.00568	chr19:1331124-2345413	NDUF57, AC005329.1, GAMT, DAZAP1, RPS15, N/A, APC2, AC027307.5, PCSK4, REEP6, ADAMTS5, AC027307.5, RKHD1, AC005943.1, MBD3, UQCR, TCF3, ONECUT3, N/A, ATP8B3, REXO1, KLF16, FAM108AA, AC012615.4, ADA13, AC012615.4, CSNK1G2, BTBD2, MKRN2, MOBK12A, C19orf36, AP3D1, DOT1L, PLEKH1, SF3A2, ANMH, JSRP1, OA21, C19orf35, N/A, LSM7, AC004410.1, TMPRSS9			
11	Chr19tp-6D4, <b>Chr19-32k-3I21</b> , Chr19-32k-2N13, Chr19-32k-3O11	48_14	79_5	0.00687	chr19:1331124-2345413	NDUF57, AC005329.1, GAMT, DAZAP1, RPS15, N/A, APC2, AC027307.5, PCSK4, REEP6, ADAMTS5, AC027307.5, RKHD1, AC005943.1, MBD3, UQCR, TCF3, ONECUT3, N/A, ATP8B3, REXO1, KLF16, FAM108AA, AC012615.4, ADA13, AC012615.4, CSNK1G2, BTBD2, MKRN2, MOBK12A, C19orf36, AP3D1, DOT1L, PLEKH1, SF3A2, ANMH, JSRP1, OA21, C19orf35, N/A, LSM7, AC004410.1, TMPRSS9			
12	Chr2tp-25B10, <b>Chr2tp-10C11</b> , Chr2tp-4E12, Chr2tp-4E12, Chr2tp-28C3, Chr2-32k-4M13	2_8	39_9	1_15	3	0.00737	chr2:87099273-87904292	AC112007, AC092651.3, AC083899.6, AC068279.6, AC133644.1, AC133644.3, N/A, PLGLB2, RGPD2	
13	<b>Chr2tp-2E3</b> , Chr2tp-19G6	1_87	0	5_78	6	0.01029	chr7:148584659-148876192	AC004941.2, AC004941.2, ZNF777, ZNF746, ZNF767	
14	<b>Chr3tp-4E6</b> , Chr3tp-10G6, Chr2tp-13tp-1D12, Chr13tp-3B4	22_49	1	10_70	0	0.01222	chr13:112568275-112939870	ATP11A, MCP2L, AL137002.19, F7, F10, PROZ, PCID2, CUL4A	
15	Chr4tp-24A9, <b>Chr14tp-17E1</b> , Chr4tp-5C8, Chr7tp-16B7	1_7	_83	0_0	_92	0.01457	chr4:189169360-189702507	TRIML2, AC138781.4, TRIML1	
16	<b>Chr5-32k-6H23</b> , Chr5tp-5F1	74_9	0	88_1	1	0.01545	chr5:33262387-3435889	N/A	
17	Chr10tp-23D1, Chr11tp-4A8, Chr11tp-28G3, Chr11tp-28G4, Chr11tp-4B12	81_6	_1	88_0	_0	0.02612	chr1:68325266-71516648	N/A, GPR177, RP65, DEPDCL4, LRRCT, AL391728.19, N/A, LRRCA0, AL353771.8, SFRS11, ANKR13C, HHLA3, CTH, AL354872.9, PTGER3, ZRANB2, hsa-mir-186	
18	<b>Chr15tp-12A11</b> , Chr15tp-4A10	85_0		84_7		0.02618	chr15:63048238-63255120	SPG21, MTIFMT, AC013552.14, RASL12, PDCD7, CLPX	
19	<b>Chr12-32k-3H21</b> , Chr12tp-18A3, Chr12tp-11G1, Chr12tp-10D9	1_5	_52	10_	2_17	_38	chr12:81598000-81881752	TMT2	
20	<b>Chr1tp-22B12</b> , Chr1tp-8V4, Chr1-32k-1C17	6_6	_47	9_	3_4	_57	1	chr1:159672939-159891049	FCGR2A, HSPA6, FCGR3A, AL451067.12, FCGR3B
21	<b>Chr4tp-14G7</b> , Chr4tp-1H7	9_45	27	8_32	46	0.02937	chr4:34313166-34632938	N/A	
22	Chr17tp-11A7p-20H2, Chr17tp-28G3, Chr17tp-28G4, Chr17tp-4B12	26_45	_12	34_31	_23	0.03077	chr17:41349822-41752876	MAPT, KIAA1267, AC005829.1, AC005829.1, AC005829.1, AR1L7P1, LRRC37A	
23	<b>Chr22tp-2A1</b> , Chr22tp-8C6	65_20		61_6		0.03138	chr22:41097245-41332085	AL022316.2, NFAM1, SERHL, CTA-126B4.5, SERHL2, Z93241.11, Z93241.11, POLDIP3	

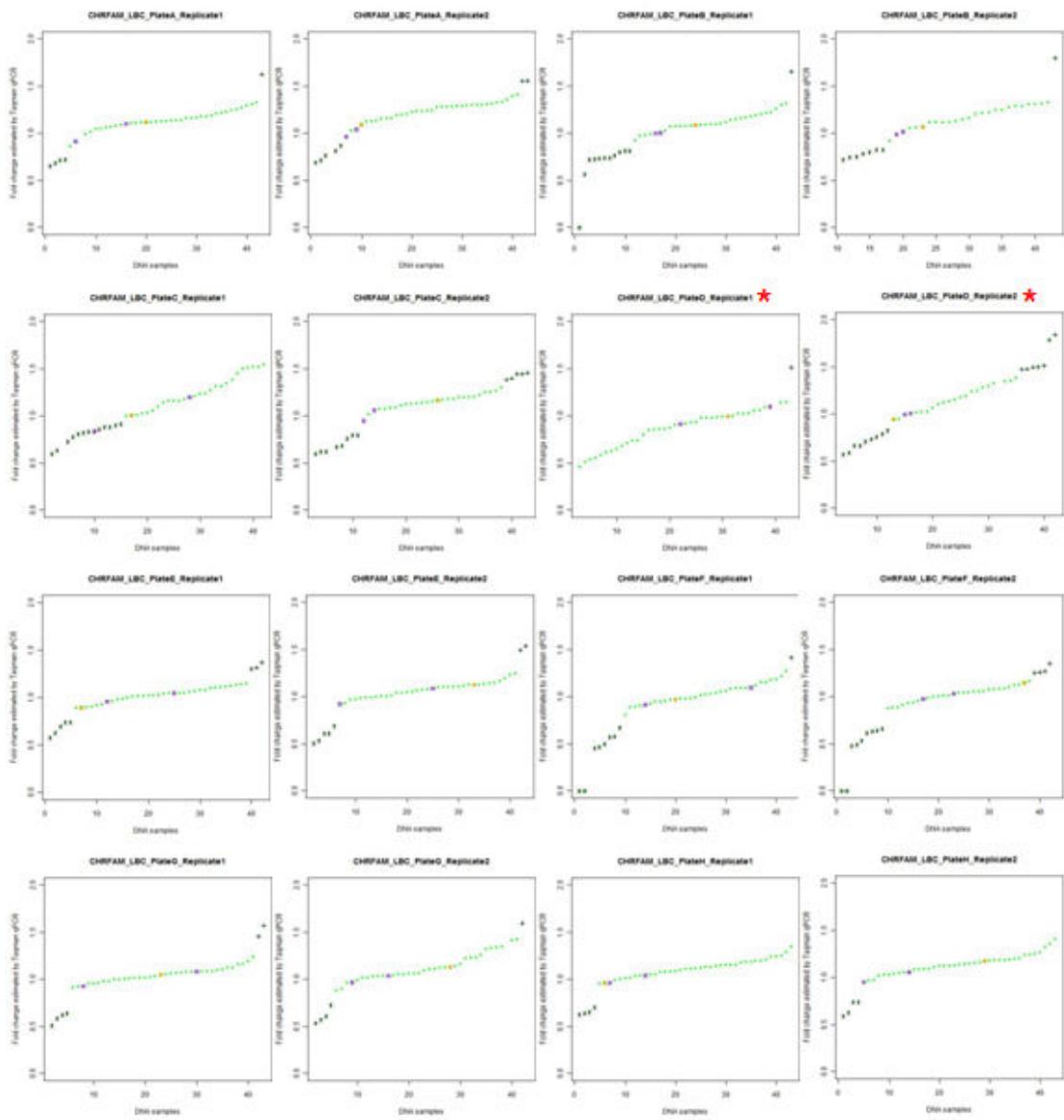
clones	SCZ class	LBC class	p-val	chrom coordinates	Genes
24	[Chr13tp-3D4, <b>Chr13tp-11B8</b> , Chr13tp-11A8	_61_9	0_77_2	0.0317	chr13:113166605-113658050
25	Chr15tp-2F10, Chr15tp-11F12, Chr15tp-1A1, <b>Chr15tp-4H6</b> , Chr15tp-12B1	4_11_4_35_19_1	1_2_10_2_23_34_6	0.03236	chr15:32330313-32775539
26	Chr12tp-2H2, <b>Chr12tp-2D2</b> , Chr12tp-6B6	51_12_0	28_18_1	0.03489	chr12:43845685-44127313
27	<b>Chr9tp-3C2</b> , Chr9tp-3E10, Chr9tp-2H8	54_16	71_7	0.03571	chr9:137679970-138276710
28	Chr20tp-6F1, Chr20tp-7A1, <b>Chr20tp-6B8</b> , Chr20tp-1E3, Chr20tp-2E8, Chr20tp-2F1	9_64_0_0	2_80_2_1	0.03897	chr20:55604184-59362789
29	Chr16tp-13A11, <b>Chr16-32k-3E20</b> , Chr16tp-11B1, Chr16tp-1C4	3_80	12_76	0.04086	chr16:28140185-28751472
30	Chr19tp-5F6, <b>Chr19-32k-1121</b> , Chr19tp-7A7, Chr19tp-2B5	2_3_61_7_1_1	0_6_68_2_2_8	0.04689	chr19:47734587-48590893
31	<b>Chr16tp-13A11</b> , Chr16-32k-3E20, Chr16tp-11B1, Chr16tp-1C4	4_84	13_78	0.04916	chr16:28140185-28751472
					DCUN1D2, A1442125.13, TMCO3, TFP1, ATP4B, GRK4, GAS6, N/A, FAM70B
					SLC12A6, AC021822.21, NOLA3, C15orf155, AGPAT7, GOLGA8A, AC025678.7, GOLGA8B, AC027139.5, AC100834.3
					PELEKA9, TMEM16F, AC063924.17
					LCN9, SOHH1, KCNT1, CAMSAP1, QSOX2, UBAC1, BYBD14A, N/A, LHC3, N/A
					ZBP1, TMEP1, AL354984.17, C20orf185, ANKR60, AL354776.15, PPP4R1L, RAB22A, VAPB, APODD1L, N/A, STX16, NEPL1, AL139349.36, hsa-mir-296, hsa-mir-298, GN/AS, GN/AS, THIL, CTSZ, TUBBL, ATPBP, C20orf145, ZNF831, EDN3, C20orf183, PHACTR3, SYCPBP2, C20orf1177, PPP1RD, CDH26, C20orf197, N/A, hsa-mir-646, AL121918.26, AL117372.35, N/A, CDH4, N/A
					SBK1, AC138904.3, EIF3CL, AC138894.2, CLN3, AC138894.2, IL27, AC130449.1, CCDC101, SULT1A2, SULT1A1, AC145285.2, AC145285.2, ATXN2L
					CEACAM8, N/A, PSG6, N/A, PSG6, N/A, PSG6, N/A, PSG6, N/A, PSG9, AC005392.2, N/A, CD177, AC005392.2, TEX101
					SBK1, AC138904.3, EIF3CL, AC138894.2, AC138894.2, CLN3, AC138894.2, IL27, AC130449.1, CCDC101, SULT1A2, SULT1A1, AC145285.2, AC145285.2, ATXN2L

**Table D-4 Results from qPCR Genotyping of a Candidate near CHRFAM7A**

Analysis of all qPCR experiments to determine *CHRFAM7A* genotypes. In each 96-well plate 40 samples (+2 controls) were genotyped. Fold change of all samples in the same experiment (sorted in ascending order) were plotted on the same graph (green dots) and CNV genotypes were estimated by the change of the slope of the graph.

(o: Homozygous deletion samples; x: heterozygous deletion samples; +: duplication samples (■: control sample NA10851; □: control sample NA12776)





## Appendix E

### Validation of SCZ Specific CNVRs

#### E1 Validation by quantitative PCR

Ten CNV regions of various sizes and types (gains/losses, single/recurrent) were selected from the 113 schizophrenia cohort-specific calls for qPCR validation. Up to three sets of primers were designed tiling across each CNV region for qPCR. SYBR quantitative PCR was performed to quantify the fold change of the patient DNA against the reference DNA NA10851. Eight regions were validated (Table E-1).

**Table E-1 10 regions selected for CNV validation by qPCR**

CNV Identifier	Chromosome Coordinates	CNV Type	Genes	Single or Recurrent	qPCR Validated
3409-loss-23	chr12:17761730-18154145	Loss	RERGL	Single	✓
850-loss-22	chr5:110212743-110577198	Loss	WDR36, TSLP	Single	✓
5324-loss-21	chr6:22212251-22607763	Loss	PRL	Single	✓
1085-loss-13	chr6:97469365-98475130	Loss	KLHL32, C6orf167	Single	✓
ED1176-loss-17	chr7:17795286-18873121	Loss	SNX13, HDAC9	Single	✓
4203-loss-28	chr7:93658283-94064936	Loss	SGCE, CASD1, COL1A2	Single	✓
3789-loss-42	chr8:9670799-9988807	Loss	TNKS, MSRA	Single	
3766-gain-3	chr1:71346768-71893785	Gain	NEGR1	Single	✓
3584-gain-11	chr3:139094479-139416525	Gain	ARMC8, DZIP1L, CLDN18, A4GNT, DBR1	Single	
323-gain-32, 4179-gain-19, 5758-gain-47	chr9:137112070-137464520	Gain	C9orf62, OLFM1	Recurrent	✓

## **E2 validation by custom oligonucleotide CGH microarrays**

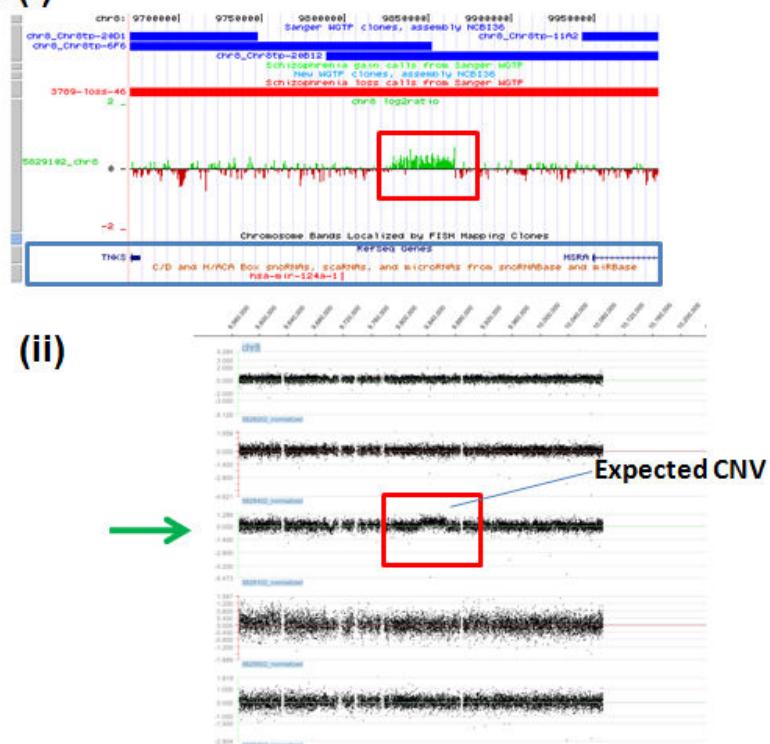
We designed a high resolution oligo array with Roche Nimblegen Inc., with a median probe spacing of 40 bp, to validate the presence and delineate the breakpoints of 7 CNV regions in the schizophrenia cohort.

Among them were two CNVRs that failed to validate by qPCR. With oligo array data we confirmed the presence of the deletion on chromosome 8 at 9 Mb, and narrowed down the CNV to ~chr8 9.83-9.87 Mb (Figure E-2a). No obvious sign of CNV was detected near the duplication call on chromosome 13 at 139Mb, suggesting a false positive call ascertained by WGTP analysis (Figure E-2b).

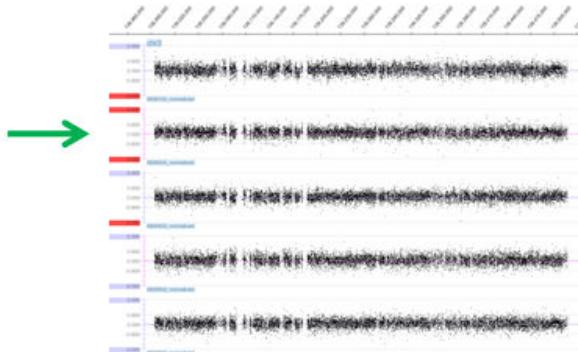
Furthermore, we confirmed the presence and resolved the boundary of another 3 CNVRs at 86-87Mb on chromosome3 (Figure E-3a), at 61 Mb on chromosome5: (Figure E-3b) and at 137 Mb on chromosome9 (Figure E-3c). WGTP array data suggested that these regions overlap the candidate genes chromatin modifying protein 2B (*CHMP2B*), kinesin heavy chain member 2 (*KIF2A*) and olfactomedin 1 (*OLFM1*), respectively. In each of these 3 cases, oligo array CGH validated the CNV regions, but restricted CNV locations to intervals either upstream or downstream of the mentioned genes.

The oligo array also indicates that two recurrent CNVRs detected on WGTP were likely false positive events. The former is a CNV in sample 5390 at chr14:103Mb overlapping the gene kinesin family member 26A (*KIF26A*), and the latter is in sample 4179 at chr20:44Mb spanning cadherin 22 precursor (*CDH22*) (data not shown). We cannot confirm whether the other affected DNA samples with CNV calls on WGTP carried any change at these loci.

### a (i) Chr8 9 Mb

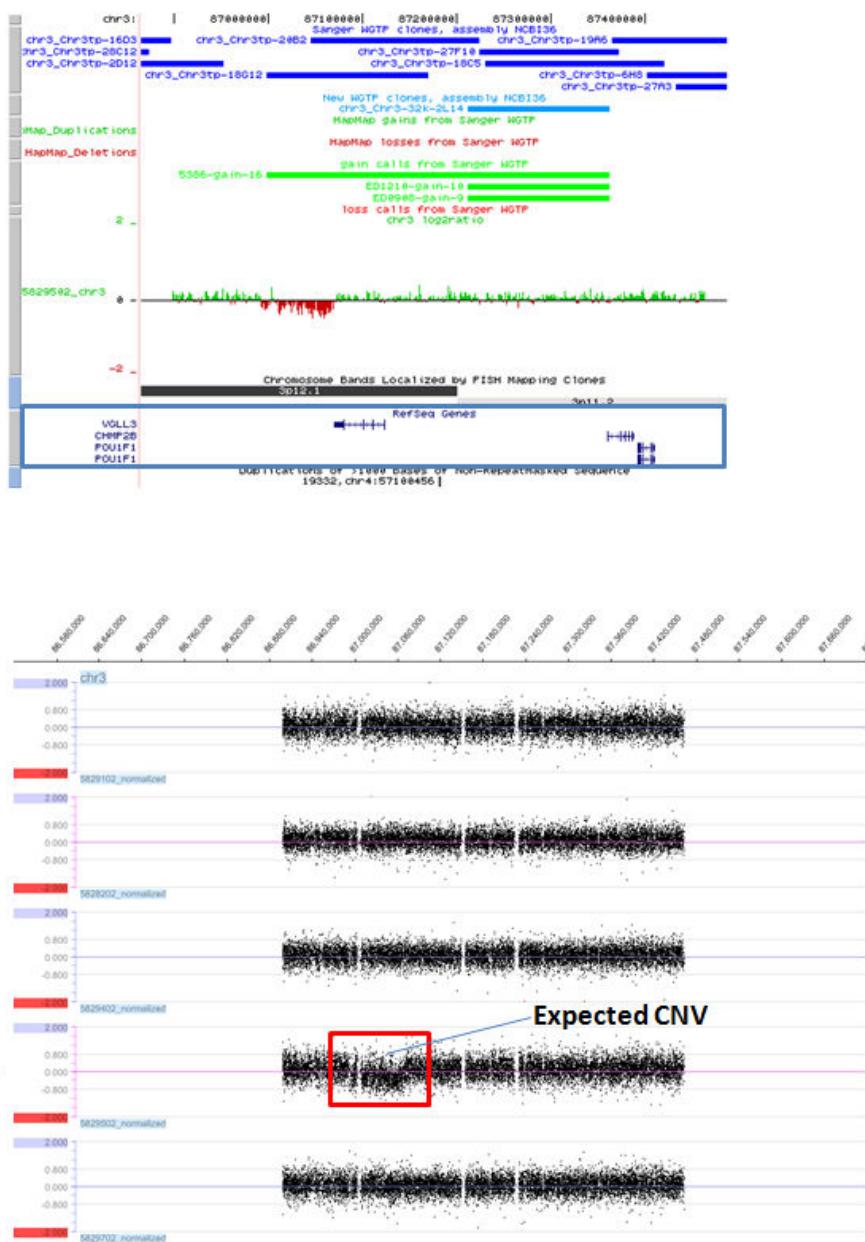


### b Chr3 139 Mb



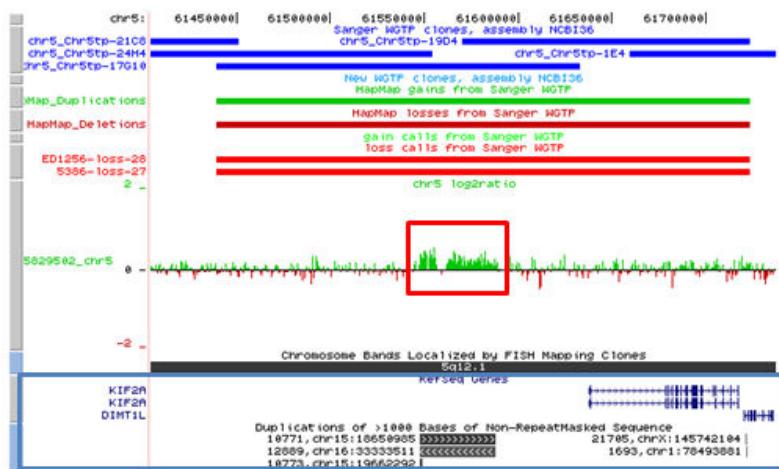
**Figure E-2 Oligonucleotide array CGH validation of 2 CNV regions that previously failed to validate with qPCR.** **a)** (i) The deletion at chr8:9Mb detected in sample 5398 was confirmed by oligo array CGH, with the CNV boundaries resolved. (ii) Results from 5 hybridization experiments on the oligo array revealed the expected deletion in sample 5398 (green arrow, hybridized as reference). **b)** The duplication call at chr3:139Mb showed no sign of CNV on the five pairs of DNA hybridized. (red filled block: CNV region as determined by WGTP array; red unfilled box: the expected CNV determined by oligo array; green arrows: pair of DNA samples we expected to see the CNV)

**a Chr3 86-87 Mb**



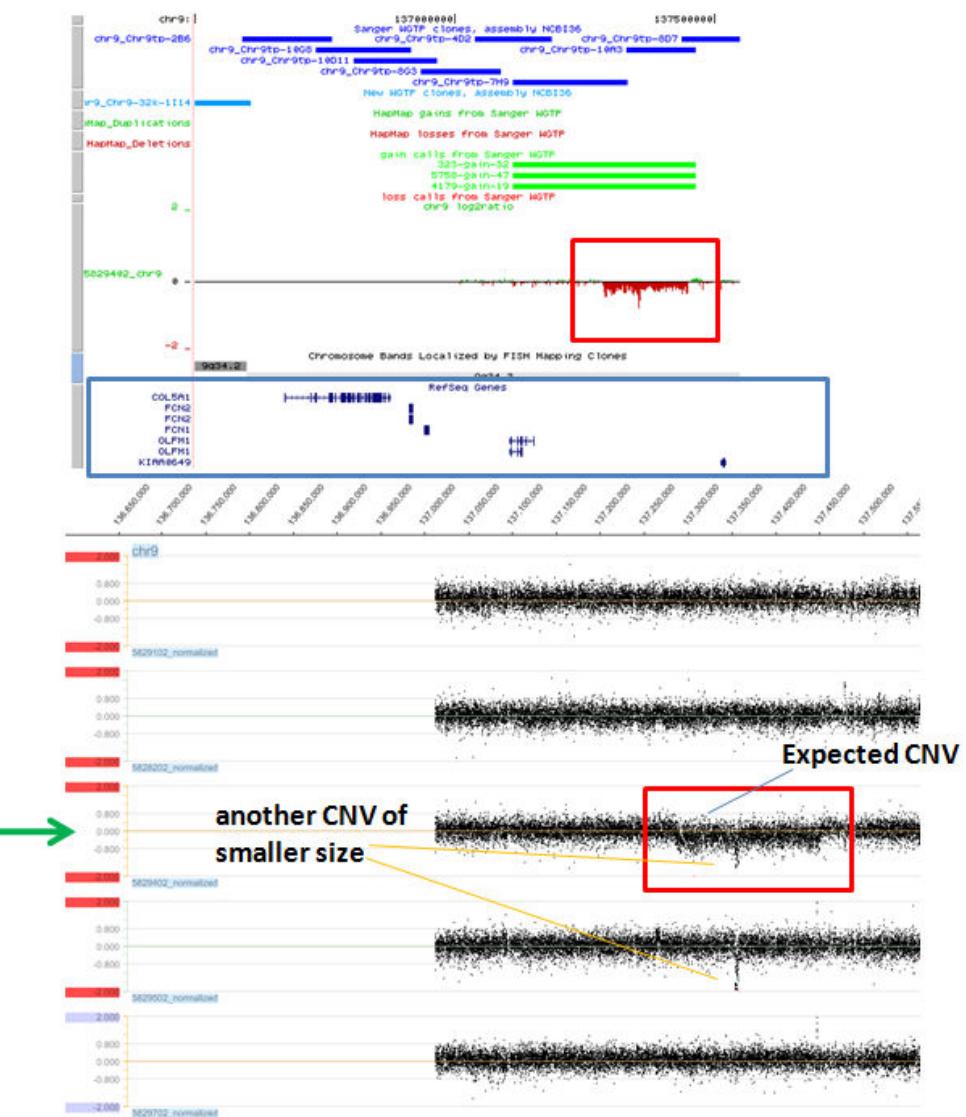
(Figure E-3 to be continued)

**b Chr5 61 Mb**



(Figure E-3 to be continued)

### c Chr9 137 Mb



**Figure E-3 Three CNVs confirmed by oligo array CGH.**

a) A duplication (sample 5386, hybridized as reference) at chr3 86-87 Mb was validated. CNV boundaries were refined to ~chr3 87.00-87.06 Mb b) A deletion (sample 5386, hybridized as reference) at chr5 61 Mb was validated. CNV boundaries were refined to ~chr5 61.55-61.58 Mb c) A duplication (sample 4179, hybridized as reference) at chr9 137 Mb was validated. CNV boundaries were refined to ~chr9 137.28-137.45 Mb  
*(red or green solid box: CNV region as determined by WGTP array; red unfilled box: expected CNV determined by oligo array; green arrows: pair of DNA samples expected to display the CNV)*

## Appendix F

### 186 Genes in the NMDA Receptor Complex (NRC/MASC)

Gene	ID	chr	start	end
GNB1	ENSG00000078369	1	1706590	1812355
CAPZB	ENSG00000077549	1	19537857	19684594
PRDX1	ENSG00000117450	1	45749295	45760196
PRKACB	ENSG00000142875	1	84316329	84476765
ATF1A1	ENSG00000163399	1	116717359	116748917
ARNT	ENSG00000143437	1	149048810	149115815
PKR	ENSG00000143627	1	153526254	153537843
PRDX6	ENSG00000117592	1	171713084	171724567
GLU1	ENSG00000135821	1	180618292	180627573
PLA2G4A	ENSG00000116711	1	185064655	185224726
ATP2B4	ENSG00000058668	1	201862312	201984201
ACTN2	ENSG00000077522	1	234916422	234993863
PRKCE	ENSG00000171132	2	45732547	46268632
CALM1	ENSG00000143933	2	47240809	47257159
SPTBN1	ENSG00000115306	2	54536958	54752086
YWHAE	ENSG00000181399	2	138762069	138762767
STK39	ENSG00000198648	2	168518777	168812365
SLC25A12	ENSG00000115840	2	172349127	172458991
MYO1B	ENSG00000128641	2	191818375	191998360
MAP2	ENSG00000078018	2	209997016	210307087
ARPC2	ENSG00000163466	2	218790119	218827324
TUBA1A	ENSG00000127824	2	219822677	219827514
IRS1	ENSG00000169047	2	227308182	227372719
ATG16L1	ENSG00000085978	2	23382958	233869059
ARPC4	none	3	9809227	9823789
RAF1	ENSG00000132155	3	12600108	12680678
CTNNB1	ENSG00000168036	3	41216000	41256938
BSN	ENSG00000164061	3	49566926	49683974
APP1	ENSG00000157500	3	57236805	577282539
SLMAP	ENSG00000163681	3	57716997	57889935
ATP6V1A	ENSG00000114573	3	114948598	115013591

Gene	ID	chr	start	end
GAP43	ENSG00000172020	3	116824861	116923024
GSK3B	ENSG00000082701	3	121028238	121295203
KALRN	ENSG00000160145	3	125296275	125922726
PIK3CA	ENSG00000121879	3	180349005	180435189
GNB4	ENSG00000114450	3	180596696	180652065
DLG1	ENSG00000075711	3	198255819	198509844
MAPK10	ENSG00000109339	4	87156655	87593320
PPP3CA	ENSG00000138814	4	102163610	102487376
SLC25A4	ENSG00000151729	4	186301392	186305418
HOMER1	ENSG00000152413	5	78704215	78845796
LMNB1	ENSG00000113368	5	126140732	126200608
PPP2CA	ENSG00000113575	5	133560047	133589849
SPINK5	ENSG00000133710	5	147423728	147497045
CAMK2A	ENSG00000070808	5	149579247	149650047
DBN1	ENSG00000113758	5	176816220	176833271
GNB2L1	ENSG00000204628	5	180596531	180603508
MOG	ENSG00000206456	6	29732788	29748128
HSPA1B	ENSG00000204388	6	31903667	31906010
EHMT2	ENSG00000204371	6	31955515	31973443
SYNGAP1	ENSG00000197283	6	33495919	33533296
VEGFA	ENSG00000112715	6	43845926	43862202
LCA5	ENSG00000135338	6	80251427	80565440
GRIK2	ENSG00000164418	6	101953385	102624651
GRM1	ENSG00000152832	6	146390611	146800427
RAC1	ENSG00000136238	7	6380651	6410120
RALA	ENSG0000006451	7	39629687	39714240
CAMK2B	ENSG00000058404	7	44223280	44331749
STX1A	ENSG00000106089	7	72751472	72771925
YWHAG	ENSG00000170027	7	75794053	75826252
ADAM22	ENSG00000008277	7	87401394	87670140
AKAP9	ENSG00000127914	7	91408128	91577925

Gene	ID	chr	start	end	Gene	ID	chr	start	end
GNB2	ENSG000000172354	7	1001109311	100114733	KLC2	ENSG000000174996	11	65781766	65791907
PRKAR2B	ENSG00000005249	7	106472375	106589491	ACTN3	ENSG000000204633	11	66087067	66087373
CAPZA2	ENSG00000198898	7	116289799	116346548	CTTN	ENSG000000085733	11	6992292	69960337
FLNC	ENSG00000128591	7	128257719	128286572	SHANK2	ENSG00000162105	11	69991609	70536020
NEFM	ENSG00000104722	8	24827188	24832508	RAB6A	ENSG00000175582	11	73064342	73149849
NEFL	ENSG00000104725	8	24864385	24870541	DLG2	ENSG00000150672	11	82843701	85015962
DPSL2	ENSG00000092964	8	26491327	26571607	GRM5	ENSG00000168959	11	87881006	88438761
PTK2B	ENSG00000120899	8	27224916	27372820	DLAT	ENSG00000150768	11	111401374	111440324
DUSP4	ENSG00000120875	8	29249539	29264104	GAPDH	none	12	6513918	6517797
RAB2A	ENSG00000104388	8	61592113	61696183	TP11	ENSG00000111669	12	6846544	6850373
RPL7	ENSG00000147604	8	74365073	74375857	PHB2	none	12	6944778	6950152
FABP5	ENSG00000164687	8	82355325	82359567	GRIN2B	ENSG00000150086	12	13605411	14024319
YWHAZ	ENSG00000164924	8	101999980	102034745	LDHB	ENSG00000111716	12	21679543	21702043
ARC	ENSG00000198576	8	143689412	143692835	FGD4	ENSG00000139132	12	32546361	32684940
GSN	ENSG00000148180	9	123009896	123134942	ARF3	ENSG00000134287	12	47616259	47637577
STXBP1	ENSG00000136854	9	129414389	129494816	MYL6	ENSG00000092841	12	54838367	54841633
DNM1	ENSG00000106976	9	130005484	1300057348	SYT1	ENSG00000067715	12	77781904	78369917
GRIN1	ENSG00000176884	9	139152663	139183028	LIN7A	ENSG00000111052	12	79715306	79855825
ATP5C1	ENSG00000165629	10	7870099	7889761	ARPC3	ENSG00000111229	12	109357090	109376185
C10orf38	ENSG00000148468	10	15293661	15453103	PPP1CC	ENSG00000186298	12	109642127	109665050
VDAC2	ENSG00000165637	10	76640569	76661208	PTPN11	ENSG00000179295	12	111340919	111432099
LGI1	ENSG00000108231	10	95507632	95547904	NOS1	ENSG00000089250	12	116120497	116283965
INA	ENSG00000148798	10	105026910	105040091	CIT	ENSG00000122966	12	118607981	118799475
ABLM1	ENSG00000099204	10	116108662	116434404	RAN	ENSG00000132341	12	129922521	129927316
HRAS	ENSG00000174775	11	522242	525591	RAP2A	ENSG00000125249	13	96884475	96918243
SLC25A22	ENSG00000177542	11	780478	786221	MYH6	ENSG00000092054	14	22946378	22974710
LDHA	ENSG00000134333	11	18372668	18386341	RTN1	ENSG00000139970	14	59132447	59407101
PTPN5	ENSG00000110786	11	18706053	18770844	AKAP5	ENSG00000179841	14	64004256	64016821
SLC1A2	ENSG00000110436	11	35229329	35398186	TJP1	ENSG00000104067	15	27778863	27901998
MAPK8IP1	ENSG00000121653	11	45863778	45884592	TP53BP1	ENSG00000067369	15	41486699	41590028
BAD	ENSG00000002330	11	63793878	63808740	MYO5A	ENSG00000197535	15	50386770	50608539

