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Supplemental Information

Refinement and Discovery of New Hotspots of Copy-Number Variation Associated with Autism Spectrum Disorder

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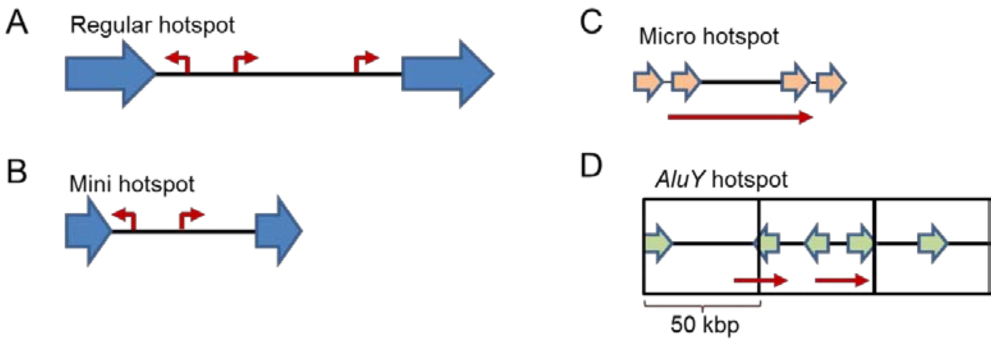


Figure S1. Definition of hotspots targeted in this study.
We identified 1,367 regions based on the repeat architecture of the human genome. These regions were classified based on the size and sequence identity of the repeats as follows: **(A)** Regular hotspots (n=120): These regions are flanked by large blocks of segmental duplications (SDs) (≥10 kbp in size) with ≥95% in sequence identity with an intervening unique segment ranging between 50 kbp – 5 Mbp. These hotspots were identified by whole-genome assembly comparison (WGAC) [1]. Based on this criteria, 143 redundant hotspots (120 non-overlapping hotspots) mapping within 115 regions were identified [2,3]. These regions were further prioritized, based on disease association, as follows: (i) out of the 115 regions, based on previous association with autism [4], 18 regions were targeted with very high density of probes; (ii) 52 sites consisted of regions where copy number variants (CNVs) are yet to be discovered; (iii) 28 hotspots were associated with canonical genomic disorders such as Williams syndrome and Smith-Magenis syndrome; and (iv) 17 hotspots are pericentromeric and potentially polymorphic (e.g., *UGT2B*, *SMN*, *PSG*, etc.). **(B)** Minihotspots (n=253): These hotspots were chosen as unique, genic regions (1–100 kbp) that are flanked by smaller (≥1 –10 kbp), directly oriented SDs with ≥90% sequence. **(C)** Microhotspots (n=410): A genome-wide scan to identify pairs of 100 bp sequences with perfect (100%) sequence identity (or k-mers) in direct orientation was performed. These 100 bp k-mers were chained, allowing two edit distances, to form sequence pairs ≥199 bp in size. Unique genic regions (1–100 kbp) flanked by such chained k-mers were defined as microhotspots. **(D)** *AluY* hotspots (n=584): A genome-wide scan was performed with a 50 kbp non-overlapping window for *AluY* and total *Alu* enrichment. (i) Top 260 *AluY*-enriched regions, i.e., with at least 15 kbp of *AluY* elements within the 50 kbp non-overlapping window and containing at least one gene, were chosen. (ii) *AluY* enriched regions (n=74) were identified on chromosome X. (iii) From a genome-wide scan of highly conserved (based on GERP scores [5]) regions, the top 250 sites were chosen that are also enriched for *AluY* repeats within a 50 kbp non-overlapping genomic window. The smaller hotspots were expanded by 5–20 kbp to accommodate at least 10 probes for improved sensitivity of CNV detection. Thus, the targeted size of *AluY* regions is 90 kbp. The block arrows indicate repeat elements (blue=SDs, orange=k-mers, green=*Alu*) and red arrows indicate genes.

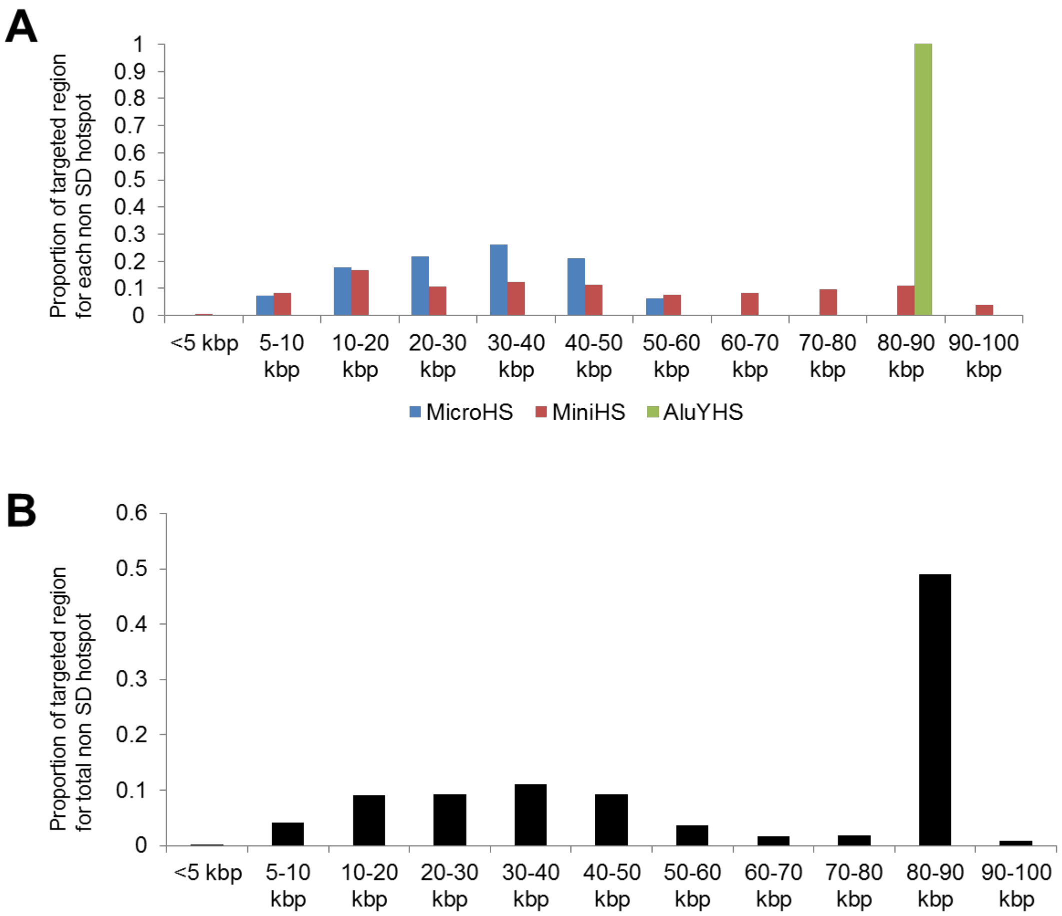


Figure S2. Size-wise distribution of targeted hotspot regions.
(A) Sizes of all smaller non-SD (n=1,247) hotspots are shown. Note that the hotspot sizes are inclusive of the extra flanks (5–20 kbp) added in the design of the array. Thus, all *AluY* hotspots are 90 kbp in size even though the intended target region is within a 50 kbp window. **(B)** Sizes of smaller non-SD hotspots as an aggregate are shown. The overall median size of these hotspots is 79 kbp (see Table 1).

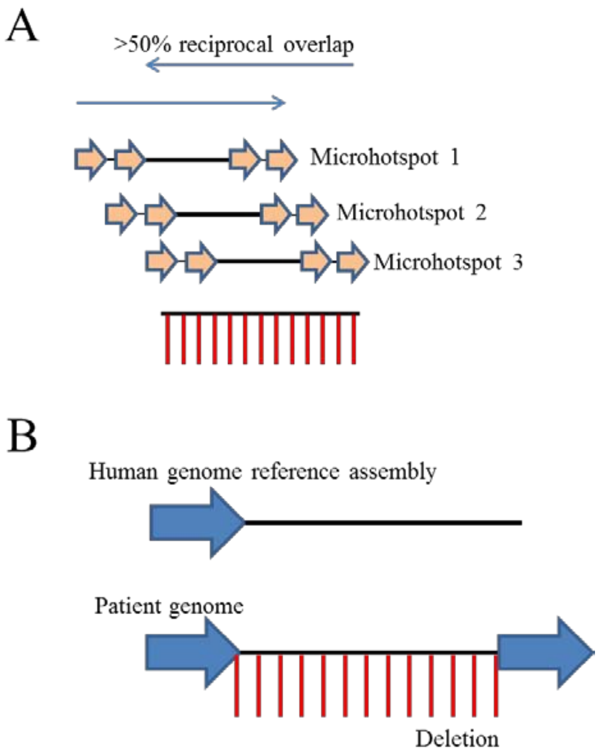


Figure S3. Identifying smaller hotspot events.
(A) Events were considered to be mediated by smaller repeats and counted only if the CNV had a reciprocal overlap of 50% or more of its length to the targeted hotspots. In this example, the deletion is mediated by one of the three hotspots, and the exact breakpoint in such cases is not determined. Note that these three hotspots will not be collapsed if they do not conform, collectively, to the definition of microhotspots (allowing for only one edit distance of sequence difference). **(B)** Since the array was designed based on the human reference genome (Build 36, hg18), we anticipated observing events where the CNV contained an unpaired repeat (SD, chained k-mer or *AluY*) element on only one side of the breakpoint. This suggests the presence of a private, predisposing genomic configuration in that individual that is not annotated in the reference genome assembly. These regions were designated as hotspot-associated events [6].

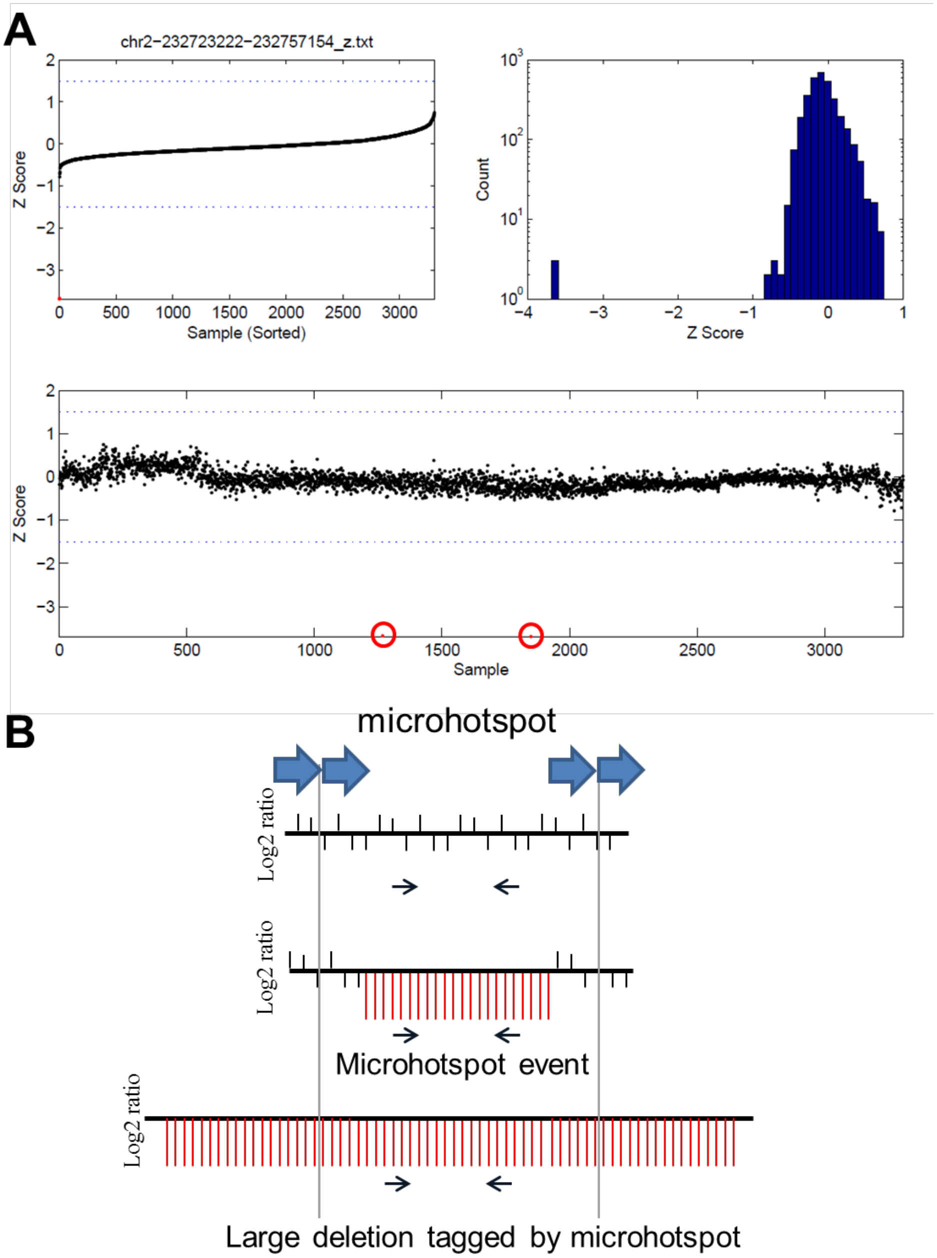


Figure S4. CNV detection within targeted regions.

(A) Transformed z-scores for one specific hotspot are shown for all the cases and controls analyzed using the targeted array. Each black dot represents a sample and the red dots represent samples with a z-score <-1.5 (circled in red) and blue dots represent samples with a z-score >1.5 (not shown). The distribution of z-scores is shown as a histogram for all the samples for that particular region. **(B)** Examples depicting variants within or traversing a microhotspot are shown. Events that are "tagged" by microhotspots are removed by manual inspection and reclassified. If no annotated repeat element is present at the flanks (as defined in this study), these "tagged" events are classified as a backbone event. Targeted genotyping of hotspots and previously implicated autism candidate genes from the SFARI gene database (sfari.org) was performed by calculating the average log intensity ratios for each targeted region. For every targeted region, the z-score was calculated using the average log intensity ratio and chromosome-specific means and standard deviations[7]. Plots were generated to manually inspect all targeted regions to select samples with an absolute z-score ≥1.5 (represented as outliers in the plot). This exercise was extended to genotype 2,090 controls but with a much more stringent filter requiring ≥10 probes to cover a region.

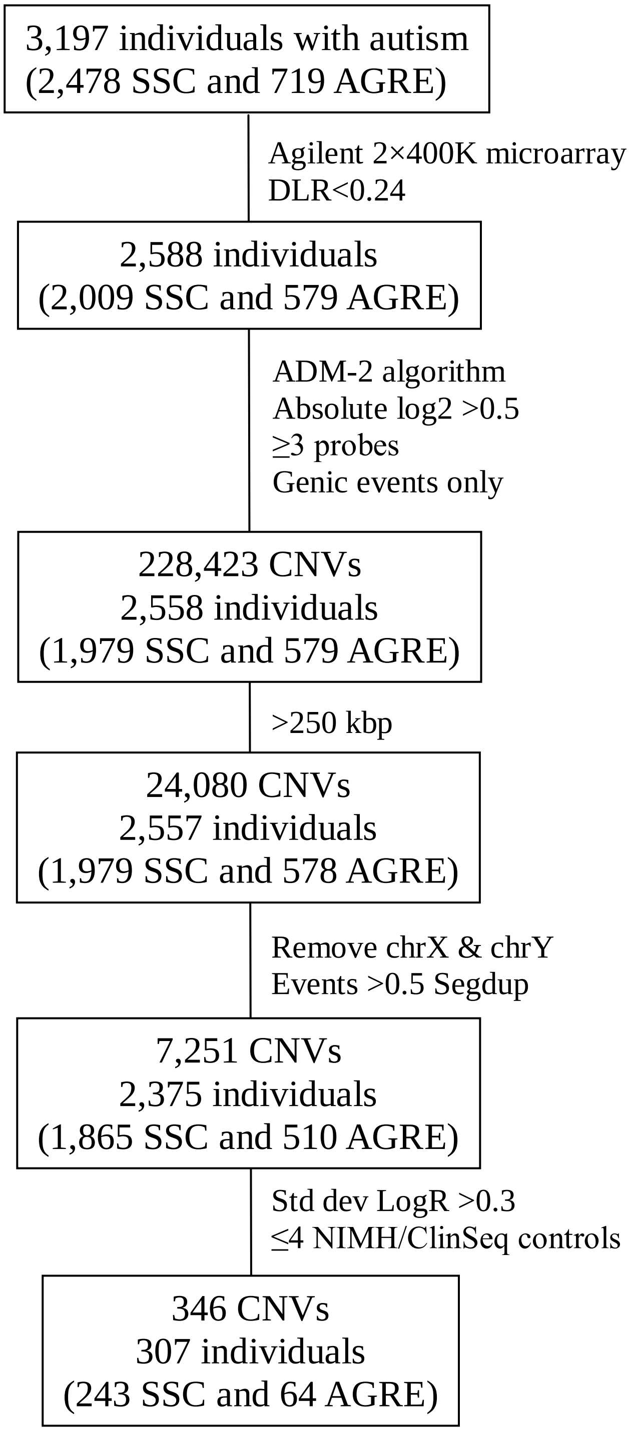
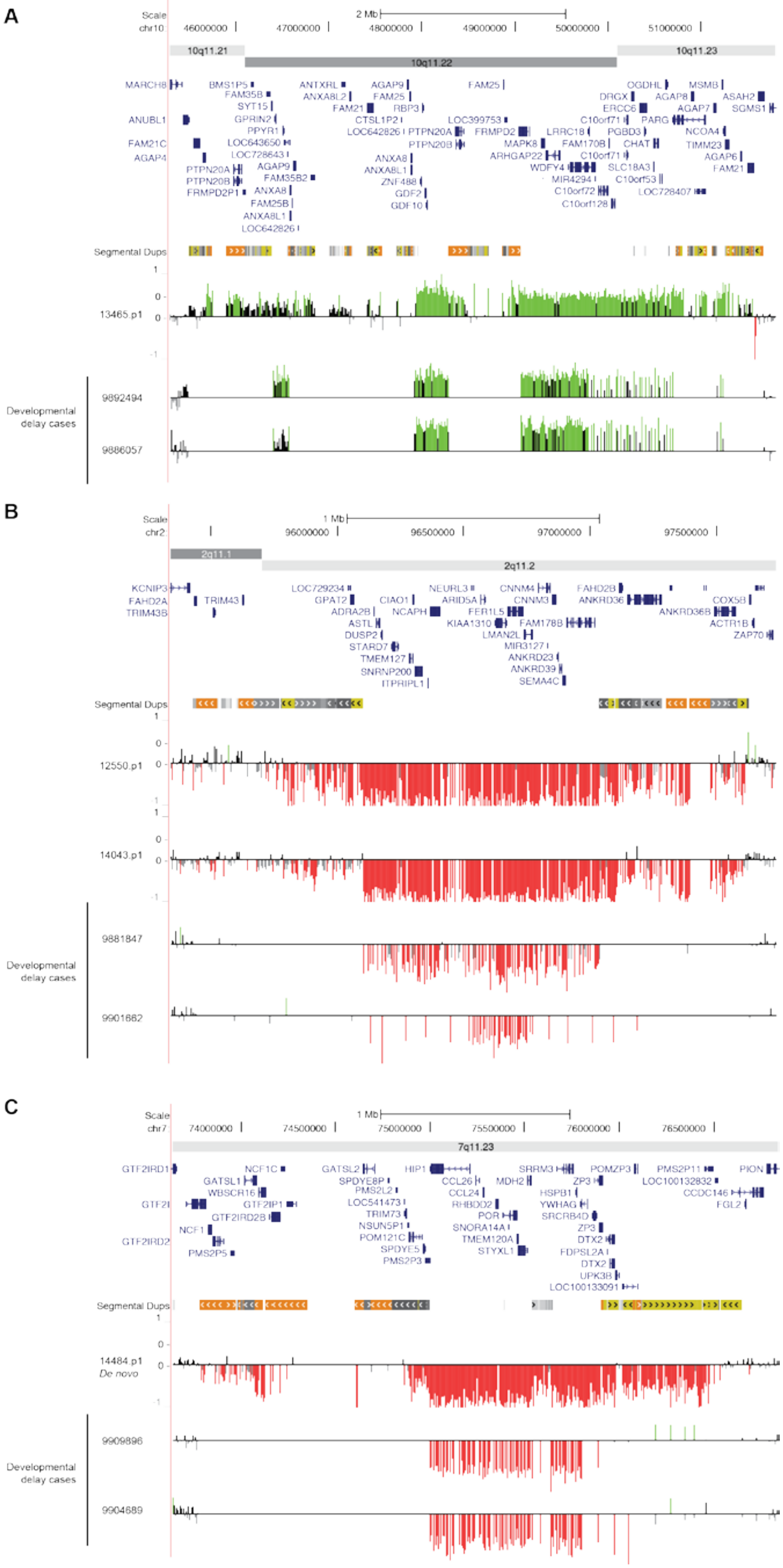


Figure S5. A work flow for CNV discovery in the genomic backbone.
Manual curation after loading the log2 signal intensities into custom UCSC genome browser was also performed to eliminate any false positive calls. Validation was performed by testing parents for inheritance status as well as by hybridization on a custom 135K NimbleGen hotspot array.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FAM35B
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PPYR1
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FAM35B2
ANXA8
FAM25B
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FAM21C
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PTPN20B
FRMPD2P1
ANTXRL
ANXA8L2
FAM21
AGAP9
FAM25
RBP3
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PTPN20B
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ANXA8L1
ZNF488
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GDF10
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AGAP7
PARG
NCOA4
TIMM23
AGAP6
FAM21
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Developmental delay cases
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9886057
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97000000
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2q11.2
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SEMA4C
CNNM4
CNNM3
FAM178B
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ANKRD36
ANKRD36B
COX5B
ACTR1B
ZAP70
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0
12550.p1
0
-1
1
0
14043.p1
0
-1
Developmental delay cases
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9901882
C
Scale
chr7
1 Mb
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74500000
75000000
75500000
76000000
76500000
7q11.23
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GTF2I
GTF2IRD2
NCF1C
GATSL1
WBSCR16
GTF2IP1
GTF2IRD2B
NCF1
PMS2P5
GATSL2
SPDYE8P
PMS2L2
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TRIM73
NSUN5P1
POM121C
SPDYE5
PMS2P3
HIP1
CCL26
CCL24
RHBDD2
POR
SNORA14A
TMEM120A
STYXL1
SRRM3
MDH2
HSPB1
YWHAG
SRCRB4D
ZP3
DTX2
FDPSL2A
DTX2
UPK3B
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ZP3
PMS2P11
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CCDC146
FGL2
PION
Segmental Dups
1
0
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De novo
0
-1
Developmental delay cases
9909896
9904689

Figure S6. Novel SD-mediated hotspots in children with autism not found in controls. We identified recurrent deletions and duplications of regions not previously reported in children with autism. Events were also found in all regions in children with developmental delay (indicated). From top to bottom, pictured are the human reference genomic coordinates (hg18), chromosome region, genes, SDs, log2 histograms depicting deletions (red) and duplications (green) for autism, and developmental delay cases. Regions depicted are recurrent **(A)** duplications of 10q11.22, **(B)** deletions 2q11.2, and **(C)** deletions of 7q11.23, distal to the Williams-Beuren syndrome locus.

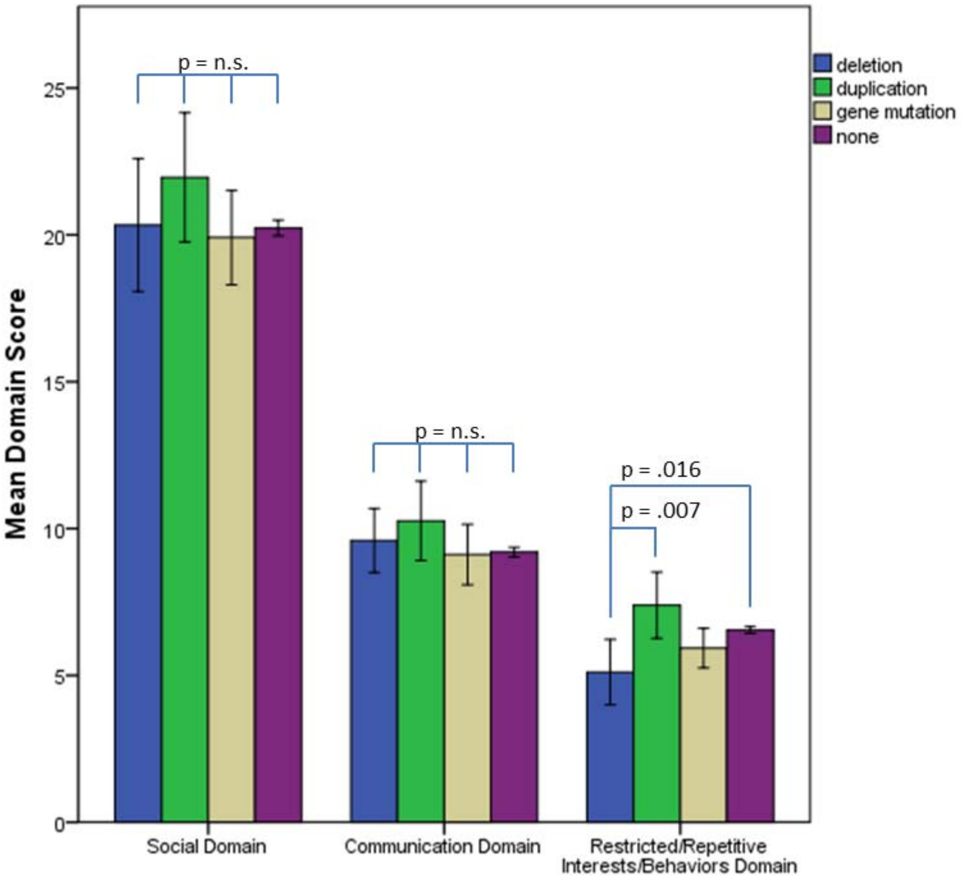


Figure S7. Autism features in individuals with deletions, duplications, *de novo* disruptive point mutations, and without any identified CNVs or mutations.
Histograms depict parent-reported autism symptoms across social, communication, and restricted/repetitive interests/behaviors domains for individuals with deletions, duplications, and *de novo* gene-disrupting mutations but without any identified CNVs or mutations.

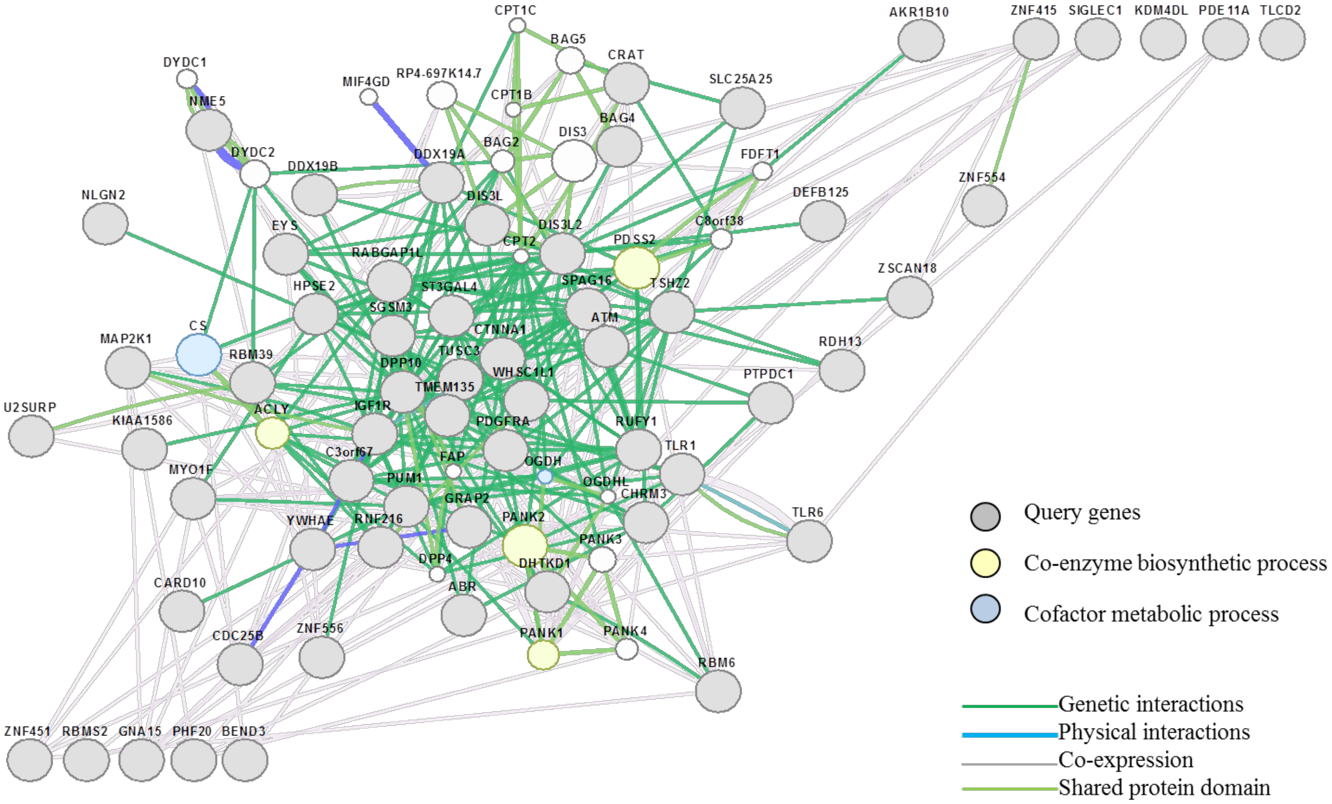
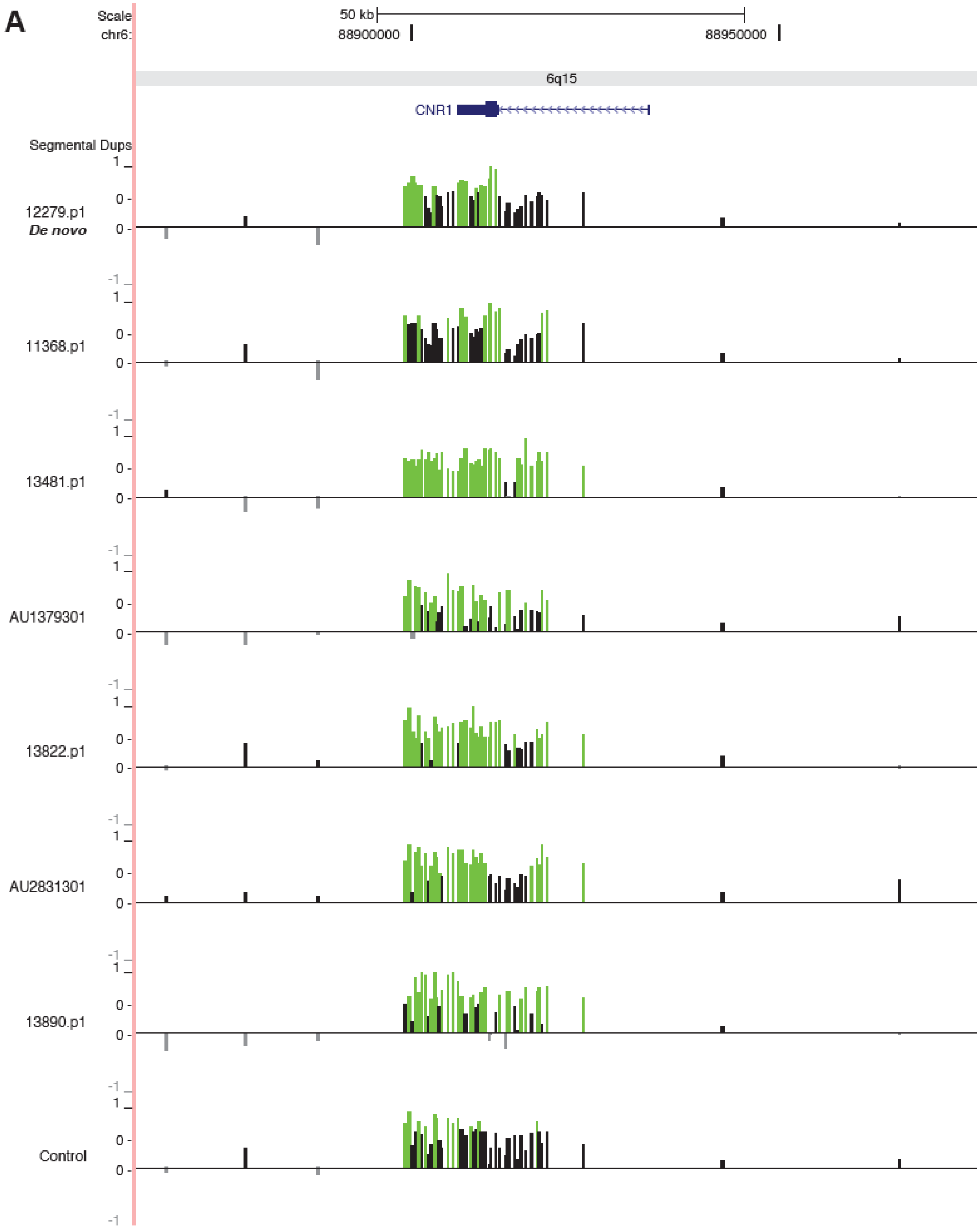


Figure S8. Pathway and gene functional analysis for the list of genes disrupted by smaller hotspot CNVs.
GeneMANIA [8] view of all the disrupted genes within smaller hotspot genes is shown.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De novo
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Control
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0
0
-1

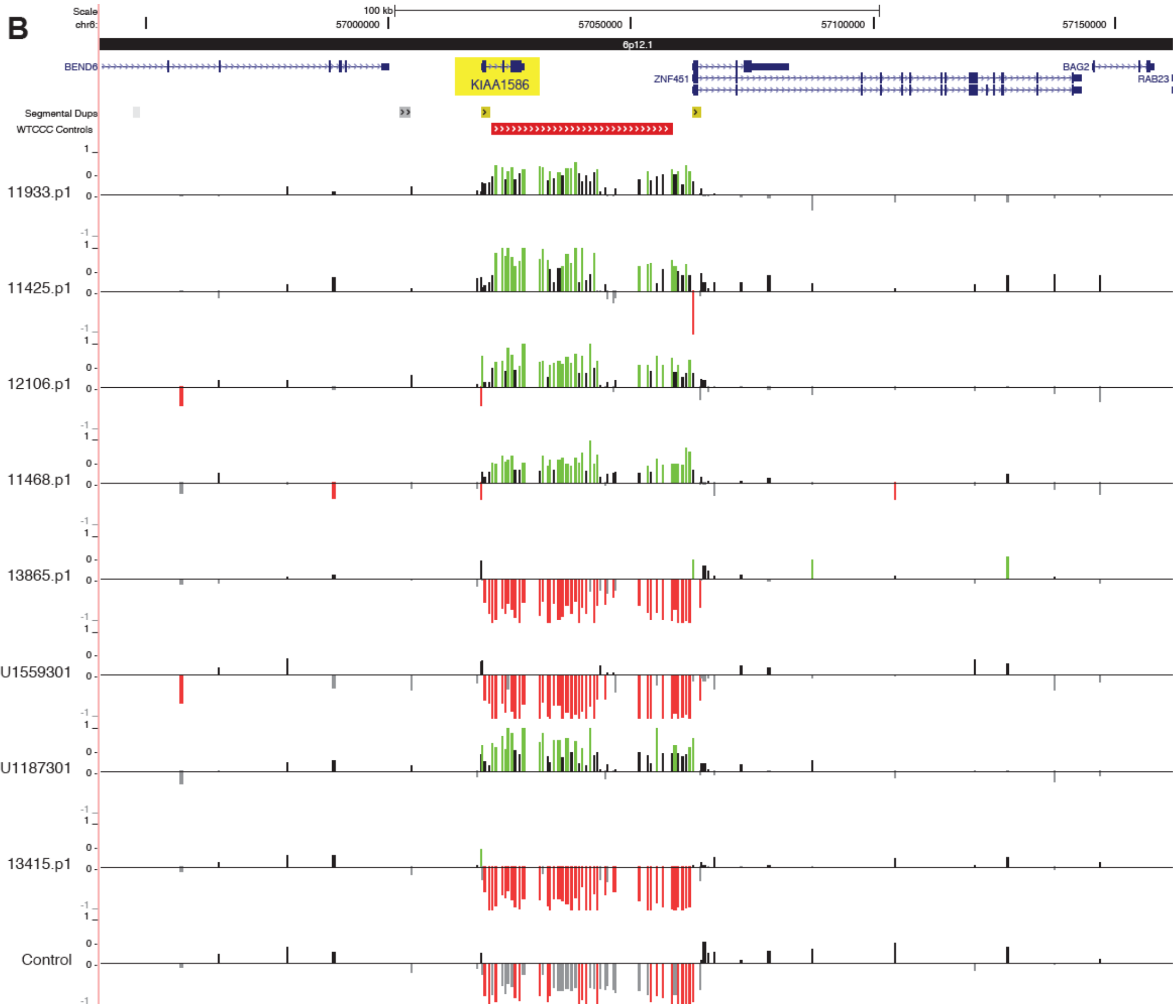

Figure S9. Autism candidate gene CNVs.
Pictured are histograms of microarray data representing log2 ratios of probes with deletions (red) and duplications (blue) depicted for candidate genes: **(A)** *CNR1* (p=0.032) and **(B)** *KIAA1586* (p=0.049).

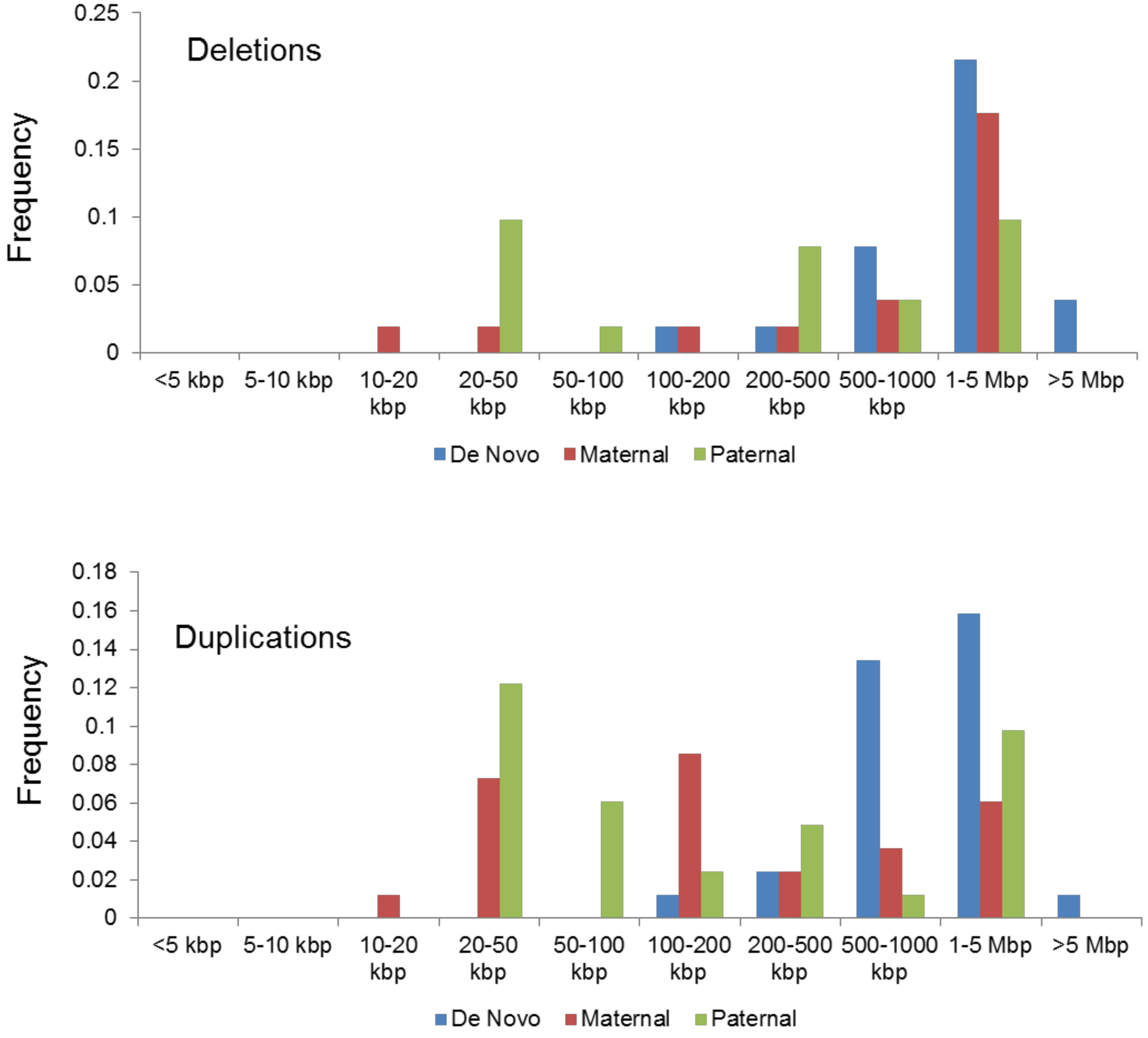

Figure S10. Inheritance status of events identified within hotspot regions.

Table S1. Genes surveyed for dosage and breakage within the 1,347 hotspots

		Breaking the gene		
	Contained within CNV	**5' end of gene**	**3' end of gene**	**Total**
Regular HS	2971	32	35	67
Mini HS	143	69	76	145
Micro HS	77	63	42	105
AluY	692	306	297	603
Total Smaller HS	912	438	415	853
HS Total	3883	470	450	920

HS=hotspot

Table S2. Cases and controls

Cohort	**Analyzed**	**Passed QC**
SSC autism	2,478	2,009
AGRE autism	719	579
NIMH controls	207	207
ClinSeq controls	373	373
WTCCC controls for targeted genotyping	2,090	

Table S3. CNVs within SD hotspots

Region	Genomic coordinates (hg18)	Samples	CNV	Inheritance	DD CNVs (n=15,767)	Total Case CNVs (n=18,355)	Control CNVs (n=8,329)	P-value	Odds ratio	Lower 95% CI	Upper 95% CI
1q21.1	chr1:144825000-146075000	12420.p1	Dup	De novo	26	33	1	4.87E-05	14.999	2.51006	608.7073
1q21.1	chr1:144825000-146075000	12907.p1	Dup	De Novo	26	33	1	4.87E-05	14.999	2.51006	608.7073
1q21.1	chr1:144825000-146075000	13975.p1	Dup	De novo	26	33	1	4.87E-05	14.999	2.51006	608.7073
1q21.1	chr1:144825000-146075000	AU3144302	Dup	De novo	26	33	1	4.87E-05	14.999	2.51006	608.7073
11q14.3	chr11:89070781-89514825	13944.p1	Dup	De novo	0	1	0	0.687865	Inf	0.011651	Inf
12p11.23	chr12:27196123-27621264	13784.p1	Dup	De novo	4	5	2	0.619403	1.134458	0.185658	11.91711
15q11.2 - q13.1	chr15:18432358-26745127	12007.p1	Dup	De novo	16	17	0	0.001724	Inf	1.873672	Inf
15q13.1	chr15:26625000-30675000	11928.p1	Dup	De novo	4	5	0	0.153973	Inf	0.415833	Inf
15q13.1	chr15:28656923-30232692	13301.p1	Del	De novo	42	44	0	6.97E-08	Inf	5.198759	Inf
15q13.1	chr15:28656923-30232692	13647.p1	Del	De novo	42	44	0	6.97E-08	Inf	5.198759	Inf
15q23-q24.1	chr15:66890869-71938416	11233.p1	Del	De novo	0	1	0	0.687865	Inf	0.011651	Inf
16p13.11_12.1	chr16:15074999-16725000	11511.p1	Dup	De novo	24	28	10	0.323242	1.270934	0.598885	2.933867
16p11.2	chr16:29425000-30275000	11090.p1	Del	De novo	64	70	3	8.48E-09	10.62435	3.487421	52.7974
16p11.2	chr16:29425000-30275000	11433.p1	Del	De novo	64	70	3	8.48E-09	10.62435	3.487421	52.7974
16p11.2	chr16:29425000-30275000	11540.p1	Del	De novo	64	70	3	8.48E-09	10.62435	3.487421	52.7974
16p11.2	chr16:29425000-30275000	12308.p1	Del	De novo	64	70	3	8.48E-09	10.62435	3.487421	52.7974
16p11.2	chr16:29425000-	12435.p1	Dup	De novo	28	37	2	7.81E-05	8.409517	2.168339	71.89444

	30275000										
16p11.2	chr16:29425000-30275000	12736.p1	Dup	De novo	28	37	2	7.81E-05	8.409517	2.168339	71.89444
16p11.2	chr16:29425000-30275000	13335.p1	Dup	De novo	28	37	2	7.81E-05	8.409517	2.168339	71.89444
16p11.2	chr16:29425000-30275000	13491.p1	Dup	De novo	28	37	2	7.81E-05	8.409517	2.168339	71.89444
16p11.2	chr16:29425000-30275000	14023.p1	Dup	De novo	28	37	2	7.81E-05	8.409517	2.168339	71.89444
16q22	chr16:68859156-72903506	12691.p1	Del	De novo	0	1	0	0.687865	Inf	0.011651	Inf
16q22	chr16:68883052-69808186	13769.p1	Dup	De novo	0	1	0	0.687865	Inf	0.011651	Inf
CMT1A	chr17:14109882-15367164	13491.p1	Dup	De novo	9	12	2	0.138459	2.723681	0.606156	25.06131
HNPP	chr17:14109882-15367164	13555.p1	Del	De novo	3	6	0	0.105903	Inf	0.534307	Inf
Potocki-Lupski syndrome	chr17:15081549-18771879	13561.p1	Dup	De novo	9	10	0	0.023697	Inf	1.017395	Inf
17q12	chr17:31355103-33660562	11353.p1	Del	De novo	14	16	2	0.048292	3.631974	0.853704	32.5675
2q12.1	chr2:103280257-108770547	12735.p1	Del	De novo	0	1	0	0.687865	Inf	0.011651	Inf
22q11.21	chr22:16925000-20075000	12239.p1	Del	De novo	96	98	0	1.10E-16	Inf	11.88983	Inf
22q11.21	chr22:16925000-20075000	13859.p1	Del	De novo	96	98	0	1.10E-16	Inf	11.88983	Inf
3q29	chr3:196789663-198943245	13064.p1	Del	De novo	6	7	0	0.07284	Inf	0.6541	Inf
Williams syndrome region	chr7:72220564-73910955	12594.p1	Dup	De novo	16	20	0	0.000561	Inf	2.241991	Inf
Williams syndrome region	chr7:72220564-73910955	AU030804	Dup	De novo	16	20	0	0.000561	Inf	2.241991	Inf
Williams syndrome region	chr7:72385594-73774582	11154.p1	Dup	De novo	16	20	0	0.000561	Inf	2.241991	Inf

distal WBS	chr7:74603980-76604725	14484.p1	Del	De novo	2	3	0	0.325453	Inf	0.187491	Inf
Williams syndrome region	chr7:74930136-75918119	14032.p1	Dup	De novo	0	1	0	0.687865	Inf	0.011651	Inf
1q21.1	chr1:144103100-144330084	14181.p1	Dup	Maternal	25	26	1	0.000541	11.81294	1.939977	483.4222
1q21.1	chr1:144825000-146075000	13507.p1	Del	Maternal	47	48	2	2.04E-06	10.91607	2.860587	92.76115
1q21.1	chr1:144825000-146075000	14473.p1	Dup	Maternal	26	33	1	4.87E-05	14.999	2.51006	608.7073
15q13.1	chr15:26996715-28157315	14074.p1	Del	Maternal	2	4	0	0.223856	Inf	0.299537	Inf
15q25.3	chr15:82625000-83675000	11348.p1	Del	Maternal	5	6	0	0.105903	Inf	0.534307	Inf
16p13.11_12.1	chr16:14301502-18700753	AU079803	Del	Maternal	6	7	3	0.616996	1.058823	0.241623	6.347529
16p13.11_12.1	chr16:15074999-16725000	14165.p1	Dup	Maternal	24	28	10	0.323242	1.270934	0.598885	2.933867
16p13.11_12.1	chr16:15074999-16725000	AU1039302	Dup	Maternal	24	28	10	0.323242	1.270934	0.598885	2.933867
16p11.2	chr16:29425000-30275000	12010.p1	Del	Maternal	64	70	3	8.48E-09	10.62435	3.487421	52.7974
16p11.2	chr16:29425000-30275000	11629.p1	Dup	Maternal	28	37	2	7.81E-05	8.409517	2.168339	71.89444
16p11.2	chr16:29425000-30275000	12417.p1	Dup	Maternal	28	37	2	7.81E-05	8.409517	2.168339	71.89444
16p11.2	chr16:29425000-30275000	13487.p1	Dup	Maternal	28	37	2	7.81E-05	8.409517	2.168339	71.89444
HNPP	chr17:14109882-15367164	14491.p1	Del	Maternal	3	6	0	0.105903	Inf	0.534307	Inf
8p22	chr8:12594361-14327206	13700.p1	Dup	Maternal	0	1	0	0.687865	Inf	0.011651	Inf
9p24.3	chr9:207823-1674250	14349.p1	Dup	Maternal	1	2	0	0.473151	Inf	0.085193	Inf
9p24.3	chr9:207823-316999	12590.p1	Del	Maternal	3	4	0	0.223856	Inf	0.299537	Inf
CMT1A	chr17:14109882-15367164	AU070705	Dup	n/a	9	12	2	0.138459	2.723681	0.606156	25.06131

HNPP	chr17:14109882-15367164	AU3006301	Del	n/a	3	6	0	0.105903	Inf	0.534307	Inf
Williams syndrome region	chr7:72220564-73910955	AU022704	Dup	n/a	16	20	0	0.000561	Inf	2.241991	Inf
1q21.1	chr1:144825000-146075000	12265.p1	Dup	Paternal	26	33	1	4.87E-05	14.999	2.51006	608.7073
1q21.1	chr1:144825000-146075000	12891.p1	Dup	Paternal	26	33	1	4.87E-05	14.999	2.51006	608.7073
10q11.23	chr10:49086581-50679930	13465.p1	Dup	Paternal	2	3	0	0.325453	Inf	0.187491	Inf
15q13.1	chr15:26996715-28160880	13585.p1	Del	Paternal	2	4	0	0.223856	Inf	0.299537	Inf
15q25.3	chr15:82625000-83675000	14252.p1	Dup	Paternal	1	2	0	0.473151	Inf	0.085193	Inf
16p13.11_12.1	chr16:15074999-16725000	11450.p1	Del	Paternal	18	21	3	0.032409	3.178647	0.948773	16.64716
16p13.11_12.1	chr16:15074999-16725000	12861.p1	Del	Paternal	18	21	3	0.032409	3.178647	0.948773	16.64716
16p13.11_12.1	chr16:15074999-16725000	14169.p1	Del	Paternal	18	21	3	0.032409	3.178647	0.948773	16.64716
16p13.11_12.1	chr16:15074999-16725000	13534.p1	Dup	Paternal	24	28	10	0.323242	1.270934	0.598885	2.933867
16p11.2	chr16:29425000-30275000	13509.p1	Dup	Paternal	28	37	2	7.81E-05	8.409517	2.168339	71.89444
16q22	chr16:68832773-70006429	AU1551302	Dup	Paternal	0	1	0	0.687865	Inf	0.011651	Inf
CMT1A	chr17:14109882-15367164	13328.p1	Dup	Paternal	9	12	2	0.138459	2.723681	0.606156	25.06131
17q12	chr17:31355103-33660562	13085.p1	Del	Paternal	14	16	2	0.048292	3.631974	0.853704	32.5675
2q11.2	chr2:96113899-97049810	12550.p1	Del	Paternal	2	4	0	0.223856	Inf	0.299537	Inf
2q11.2	chr2:96113899-97049810	14043.p1	Del	Paternal	2	4	0	0.223856	Inf	0.299537	Inf
5p15.33	chr5:369328-725118	12376.p1	Dup	Paternal	5	6	0	0.105903	Inf	0.534307	Inf
7q36.2	chr7:152199145-153299935	12185.p1	Dup	Paternal	0	1	0	0.687865	Inf	0.011651	Inf

Table S4. Comparison of head circumferences in individuals with specific CNVs

CNVs	N	Mean	Std. Deviation	Std. Error Mean	Significance	Percentile within ASD sample using normed HC z score[a]	Z score within only ASD sample
1q21.1 dup	6	1.63	0.50	0.20	t=4.176, df=5, p=.009	76.1	0.71
1q21.1 del	1	-0.62	.	.		16.3	-0.98
16p11.2 dup	8	-0.22	0.92	0.33	t=-3.565; df=11; p=.004	23.6	-0.72
16p11.2 del	5	2.30	1.66	0.74		88.9	1.22
WBS dups	3	1.58	0.84	0.48	t=1.287; df=2; p=.327	74.8	0.67
WBS del	1	0.33	.	.		39.3	-0.27

[a] From analysis of 1,962 SSC samples; Mean=0.69, Std. Deviation=1.32.

Table S5. Relationship between CNV size and proband characteristics

Region	N (with data)	Autism Severity (mean=6.95, sd=1.61)	Nonverbal IQ (mean=81.43, sd=28.15)	Size of the CNV
Duplications				
16p11.2	9	7.33	74.22	555,168
1q21.1	7	7.00	88.83	821,359
16p13.11	3	6.33	108.33	790,319
WBS	3	7.67	75.00	1,400,792
15q11.2q13	1	**10.00 (z=1.89)**	54.00	3,730,036
17p12	0	No Data	No Data	
16p12.1	0	No Data	No Data	
15q13.3	0	No Data	No Data	
Deletions				
16p11.2	6	6.67	80.00	555,168
15q13.3	6	6.50	103.50	1,344,798
16p13.11	5	6.00	72.60	790,319
17p12	0	No Data	No Data	
17p13.3	0	No Data	No Data	
17q12	3	6.67	90.67	1,415,741
1q21.1	2	8.00	78.50	821,359
2q23.1	3	7.33	79.00	570,781
3q29	2	6.00	67.50	1,616,303
DiGeorge/VCFS	5	7.00	95.60	1,273,719
Phelan-McDermid	4	6.25	**51.00 (z=-1.08)**	**8,184,170**
15q24	0	No Data	No Data	
15q25.2q25.3	1	8.00	84.00	661,819
16p12.1	0	No Data	No Data	
19p13.12	2	8.00	**110.00 (z=1.01)**	**3,617,438**
9q34	1	6.00	**34.00 (z=-1.68)**	**3,254,880**

Note: Bolded items indicate >1 sd (standard deviation) difference from mean. N is number of samples with available phenotypic data.

The table presents average severity and NVIQ data for each region identified in Figure 1. For several regions there are no participants with phenotype data. The mean severity and NVIQ for all participants in these regions are presented. Those regions with average scores greater than or less than one standard deviation from the mean are bolded.

Table S6. CNVs within microhotspots

chr	start	end	CNV counts overlapping with microhotspot
chr1	7364271	7395148	0
chr1	8976823	9005092	0
chr1	48517241	48551805	0
chr1	70123903	70146782	0
chr1	75615354	75621731	0
chr1	91610085	91630324	0
chr1	117100144	117102740	0
chr1	117100486	117103082	0
chr1	151295641	151310022	0
chr1	152231482	152274169	0
chr1	152552014	152561190	0
chr1	153186191	153189030	0
chr1	155335552	155372086	0
chr1	172828150	172865894	0
chr1	172828372	172866119	0
chr1	172829309	172867087	0
chr1	172830878	172868674	0
chr1	172848824	172866877	0
chr1	172870327	172874539	0
chr1	177431856	177437100	0
chr1	178538364	178572179	0
chr1	178538772	178572497	0
chr1	184184053	184189911	0
chr1	237853754	237868301	0
chr1	237854606	237869118	0
chr1	242722735	242753992	0
chr1	244836101	244865464	0
chr1	244837037	244866471	0
chr1	246530714	246557246	0
chr10	53463684	53469260	0
chr10	59981745	59983758	0
chr10	60617824	60631115	0
chr10	74423617	74455521	0
chr10	74510721	74512838	0
chr10	100533137	100552776	0
chr10	100738654	100765039	0
chr10	103259025	103267954	0
chr10	134985485	135031148	0
chr11	6123628	6165548	0

chr11	6123970	6165874	0
chr11	14134479	14147785	0
chr11	14827088	14861496	0
chr11	14827370	14861771	0
chr11	33681701	33686085	0
chr11	59963321	59998246	0
chr11	69864582	69869185	0
chr11	84395963	84401047	0
chr11	86631362	86659107	0
chr11	118054345	118067684	0
chr11	123391562	123399345	0
chr11	123757591	123771834	0
chr11	123757834	123772109	0
chr11	123880681	123929802	0
chr12	38471546	38516911	0
chr12	38471797	38517249	0
chr12	51150568	51170218	0
chr12	51153333	51173133	0
chr12	53075478	53077856	0
chr12	69547965	69558250	0
chr12	99400990	99433443	0
chr12	123469020	123472484	0
chr13	24299655	24340623	0
chr13	42978565	43004019	0
chr13	69259989	69285049	0
chr13	75182543	75201583	0
chr13	112491063	112492472	0
chr13	112769490	112771413	0
chr13	112769764	112771559	0
chr13	112770051	112771436	0
chr14	22929079	22959287	0
chr14	23543760	23588920	0
chr15	47435042	47477114	0
chr15	47435962	47477982	0
chr15	47436231	47478198	0
chr15	47436583	47478625	0
chr15	47437256	47479238	0
chr15	47437817	47479786	0
chr15	47439033	47481054	0
chr15	51576574	51624618	0
chr15	69175228	69214481	0

chr15	79288589	79308826	0
chr15	86882379	86890362	0
chr15	88199427	88213677	0
chr15	97735681	97776759	0
chr16	143252	154715	0
chr16	159495	164074	0
chr16	159922	164398	0
chr16	159922	164516	0
chr16	3042772	3062645	0
chr16	3043582	3063493	0
chr16	22811888	22816741	0
chr16	31200976	31228116	0
chr16	60374504	60394122	0
chr16	60374941	60394557	0
chr16	60376426	60396086	0
chr16	60376719	60397072	0
chr16	60377692	60397860	0
chr16	60378807	60398526	0
chr16	65526299	65539554	0
chr17	422781	449731	0
chr17	10346988	10374106	0
chr17	36456710	36469798	0
chr17	36469492	36475654	0
chr17	38257155	38273328	0
chr17	38261925	38280009	0
chr17	38525490	38562663	0
chr17	42830670	42853654	0
chr18	22073182	22094166	0
chr18	45827188	45865368	0
chr18	75132722	75146377	0
chr19	5782575	5795118	0
chr19	5782620	5818001	0
chr19	5794889	5817965	0
chr19	8875565	8878756	0
chr19	10135596	10138605	0
chr19	11888320	11929833	0
chr19	11901548	11941690	0
chr19	21461011	21485491	0
chr19	23335331	23336464	0
chr19	42334457	42368355	0
chr19	42334724	42368667	0

chr19	46945194	46984233	0
chr19	46945520	46984672	0
chr19	46945876	46984961	0
chr19	50366996	50368560	0
chr19	51402994	51419041	0
chr19	58291427	58321626	0
chr19	58291886	58322334	0
chr19	58570764	58597264	0
chr19	61648068	61649205	0
chr19	62409873	62453240	0
chr19	63024549	63055549	0
chr19	63655559	63671398	0
chr19	63658985	63674864	0
chr2	33253017	33278697	0
chr2	47180932	47189002	0
chr2	98965844	98989349	0
chr2	135881357	135890782	0
chr2	141508628	141518911	0
chr2	144111946	144141073	0
chr2	144112266	144141464	0
chr2	158084022	158113015	0
chr2	158084244	158113218	0
chr2	160056200	160060196	0
chr2	160564161	160613887	0
chr2	171181026	171184176	0
chr2	178423694	178431616	0
chr2	178424123	178432060	0
chr2	185138588	185181127	0
chr2	187938931	187948270	0
chr2	201867609	201877336	0
chr2	218608123	218642503	0
chr2	232953667	232982184	0
chr2	232954104	232982388	0
chr2	236959540	236983723	0
chr2	240617916	240633955	0
chr20	51523021	51554524	0
chr20	51523382	51554819	0
chr20	51524227	51555673	0
chr20	51527055	51558726	0
chr21	44818451	44845630	0
chr21	45195222	45196532	0

chr22	29217699	29251694	0
chr22	37763827	37800887	0
chr22	47355632	47366768	0
chr3	13975593	14008330	0
chr3	16972149	16982369	0
chr3	40236388	40256174	0
chr3	58807688	58815903	0
chr3	99334315	99370532	0
chr3	113078632	113096190	0
chr3	122505180	122517007	0
chr3	125568579	125569666	0
chr3	131440186	131456384	0
chr3	132562425	132582402	0
chr3	132797905	132837629	0
chr3	136204564	136230131	0
chr3	136204866	136230393	0
chr3	136206054	136231649	0
chr3	136455683	136483609	0
chr3	143574634	143580161	0
chr3	172082382	172128964	0
chr3	189984770	189985788	0
chr3	189984821	189985970	0
chr4	26216166	26261075	0
chr4	70928350	70950564	0
chr4	81078567	81108348	0
chr4	81079160	81108741	0
chr4	81079897	81109455	0
chr4	81080267	81110214	0
chr4	81081026	81110830	0
chr4	81081646	81111182	0
chr4	81082017	81111928	0
chr4	81082755	81112327	0
chr4	81083143	81112586	0
chr4	81083400	81113020	0
chr4	87280465	87312728	0
chr4	90035826	90065368	0
chr4	94733589	94753051	0
chr4	94749620	94779906	0
chr4	94750735	94780976	0
chr4	94751238	94781463	0
chr4	94751464	94781681	0

chr4	94751739	94782002	0
chr4	94753601	94783808	0
chr4	165242758	165284068	0
chr4	165244301	165285561	0
chr4	184478009	184480485	0
chr5	11724567	11729088	0
chr5	43641104	43651008	0
chr5	140188720	140202246	0
chr5	140456670	140462803	0
chr5	140517413	140561507	0
chr5	140537787	140574332	0
chr5	140538286	140574571	0
chr5	140548722	140585108	0
chr5	140553938	140561372	0
chr5	140553938	140585108	0
chr5	140561151	140585102	0
chr5	140584905	140607233	0
chr5	140705710	140735978	0
chr5	140711652	140721853	0
chr5	140747591	140758328	0
chr5	156017294	156025739	0
chr5	156017528	156026339	0
chr5	167901028	167933856	0
chr5	169574284	169600754	0
chr5	170418132	170444346	0
chr5	170419582	170445669	0
chr6	3099938	3122867	0
chr6	3101155	3124985	0
chr6	3101686	3125544	0
chr6	3122411	3170825	0
chr6	3122657	3171036	0
chr6	16722062	16723535	0
chr6	26476890	26514705	0
chr6	29650348	29664610	0
chr6	31891506	31905415	0
chr6	35212306	35213386	0
chr6	44947756	44962098	0
chr6	44962221	44977866	0
chr6	57019532	57064336	0
chr6	64848481	64860922	0
chr6	64848726	64861129	0

chr6	69459326	69464703	0
chr6	72041915	72085031	0
chr6	119459218	119499070	0
chr6	123808102	123840182	0
chr6	125651768	125684597	0
chr7	14288126	14322660	0
chr7	14288389	14322865	0
chr7	14289503	14324024	0
chr7	34805846	34823910	0
chr7	38245643	38265928	0
chr7	38246008	38266313	0
chr7	38246333	38266728	0
chr7	65737838	65775229	0
chr7	65738122	65775444	0
chr7	65738736	65776215	0
chr7	83547822	83589191	0
chr7	86489826	86530372	0
chr7	88323782	88341905	0
chr7	92792961	92814821	0
chr7	103947905	103987846	0
chr7	103948143	103988091	0
chr7	133873079	133912213	0
chr7	146238702	146261723	0
chr7	147276949	147280392	0
chr7	148885320	148926792	0
chr7	148937414	148971628	0
chr8	13195491	13198821	0
chr8	18653394	18659079	0
chr8	48486461	48492639	0
chr8	56537388	56553515	0
chr8	69493708	69539903	0
chr8	81060262	81081490	0
chr8	85495657	85540471	0
chr8	85495874	85540875	0
chr8	85496370	85541322	0
chr8	85497645	85542449	0
chr8	85643362	85691145	0
chr8	88458946	88503303	0
chr8	100133397	100165415	0
chr8	104834321	104866887	0
chr8	133161672	133167273	0

chr8	143955598	143993844	0
chr9	3462171	3466988	0
chr9	3462402	3467191	0
chr9	98603551	98627238	0
chr9	98603877	98627503	0
chr9	118315809	118338398	0
chr9	120936425	120975899	0
chr9	123592315	123598854	0
chr9	124526518	124552590	0
chr9	124668954	124669998	0
chrX	26089560	26123017	0
chrX	26090420	26123958	0
chrX	26091009	26124663	0
chrX	26091909	26125434	0
chrX	26092330	26125807	0
chrX	26093404	26126882	0
chrX	31439771	31459337	0
chrX	46781053	46817576	0
chrX	54759986	54804258	0
chrX	57160421	57176358	0
chrX	57163088	57179350	0
chrX	57163383	57180040	0
chrX	63391120	63435577	0
chrX	63391868	63436313	0
chrX	63392187	63436660	0
chrX	65274020	65318107	0
chrX	67366634	67406710	0
chrX	68920960	68960594	0
chrX	68923507	68963190	0
chrX	74647426	74674859	0
chrX	102766360	102780561	0
chrX	107452563	107454539	0
chrX	117028299	117055503	0
chrX	140795773	140811886	0
chrX	146147750	146149743	0
chrX	149713749	149733646	0
chrX	150531647	150547271	0
chrX	150574425	150586165	0
chrY	5069123	5103983	0
chrY	9967798	10009069	0
chrY	15167127	15193190	0

chrY	15167675	15193829	0
chr1	31181042	31195047	1
chr13	51122056	51164168	1
chr13	66471619	66500012	1
chr15	60083019	60098128	1
chr16	87956630	87986100	1
chr17	11690163	11706471	1
chr2	213918176	213941138	1
chr3	58776011	58807951	1
chr3	58776031	58815772	1
chr3	58779367	58805501	1
chr3	58782388	58816295	1
chr3	58782526	58808613	1
chr3	162176742	162205106	1
chr4	38475250	38506096	1
chr4	54796337	54814034	1
chr11	107736766	107753751	2
chr11	107737766	107754732	2
chr11	125749831	125771417	2
chr16	68924327	68963148	2
chr19	58600470	58647878	2
chr19	61401731	61432513	2
chr19	62572652	62598491	2
chr2	116264115	116287283	2
chr2	232722656	232752016	2
chr2	232724406	232752723	2
chr2	232725283	232753281	2
chr2	232726222	232754154	2
chr20	63848	91170	2
chr3	144248009	144287782	2
chr6	26514226	26552479	2
chr9	17248979	17277215	2
chr9	17254976	17277216	2
chr9	95891470	95916186	2
chr9	95892664	95917517	2
chr9	95895726	95920532	2
chr11	94398668	94422793	3
chr13	66213547	66248610	3
chr6	52729617	52771620	3
chr11	56075176	56113063	4
chr11	56100952	56136937	4

chr12	51130292	51152169	4
chr12	51131038	51153030	4
chr12	51131713	51153641	4
chr12	51131713	51173092	4
chr6	52758032	52797115	8
chr17	36756663	36775230	10
chr1	70146799	70164769	11
chr6	52738196	52778804	11
chr6	52739527	52780178	11
chr8	71771170	71811746	12
chr8	71772142	71812740	12
chr11	93541125	93550334	13
chr8	15459053	15479728	14
chr8	15459644	15480527	14
chr8	15460924	15481502	14
chr8	15461520	15482427	14
chr8	15462411	15483151	14
chr19	58635421	58673115	17
chr19	58637062	58674821	17
chr22	26920578	26939753	17
chr22	26922186	26940238	17
chr6	71702475	71713033	18
chr6	71703270	71713808	18
chr6	71703672	71714238	18
chr6	71704421	71715059	18
chr6	71705104	71715710	18
chr5	126254342	126268089	26
chr5	126254994	126268755	26
chr5	126255360	126269105	26
chr5	126255700	126269427	26
chr5	126257139	126271113	26
chr5	126257577	126271311	26
chr9	71282100	71313721	42
chr9	71284957	71314452	42
chr9	71285631	71314852	42
chr9	71286629	71315881	42
chr9	5292794	5329656	43
chr16	74090198	74130702	51
chr16	74090525	74131014	51
chr16	74096080	74137424	51
chr1	86178391	86186818	60

chr17	36456655	36475593	82
chr17	36464664	36475593	82

Overlap with microhotspot is based on 50% reciprocal overlap.

Table S7. List of CNVs within minihotspots

chr	start	end	No of events within minihotspot
chr1	2469724	2499885	0
chr1	2472668	2491592	0
chr1	12917368	12975585	0
chr1	12970110	13031697	0
chr1	92368743	92384138	0
chr1	143326372	143333769	0
chr1	179200022	179286898	0
chr1	179203479	179298725	0
chr1	194925159	195016547	0
chr1	204082120	204091133	0
chr1	224141032	224195718	0
chr1	244835333	244866508	0
chr10	26944305	27026530	0
chr10	26951716	27007191	0
chr10	27424752	27436385	0
chr10	96759116	96795997	0
chr10	101682117	101713759	0
chr11	297319	316827	0
chr11	1638691	1668697	0
chr11	69864258	69869377	0
chr11	70914580	70954897	0
chr11	116509420	116587255	0
chr11	118033832	118066537	0
chr11	118053959	118067914	0
chr11	125748401	125772450	0
chr12	2660503	2722138	0
chr12	2674271	2723629	0
chr12	10924360	10976655	0
chr12	39557392	39562725	0
chr12	51127117	51154381	0
chr12	69486558	69489734	0
chr12	92410418	92417415	0
chr13	18433321	18523994	0
chr15	43190117	43215922	0
chr15	73300670	73356714	0
chr15	79287785	79309283	0

chr16	142720	155198	0
chr16	2601585	2687324	0
chr16	2804128	2817953	0
chr16	3042159	3066812	0
chr16	31280833	31330347	0
chr16	65526140	65541618	0
chr16	73433985	73501309	0
chr16	73796065	73815693	0
chr17	1422235	1439016	0
chr17	2985602	3070764	0
chr17	16754112	16817813	0
chr17	20333775	20371591	0
chr17	25940403	25988412	0
chr17	36831120	36849242	0
chr17	37623684	37707008	0
chr17	38256993	38274423	0
chr17	55436696	55529713	0
chr19	5782442	5796073	0
chr19	5814345	5828859	0
chr19	15699716	15766768	0
chr19	21347870	21381429	0
chr19	40532345	40546777	0
chr19	42333940	42370269	0
chr19	49604409	49612307	0
chr19	51390726	51457278	0
chr19	51408914	51430672	0
chr19	57983031	58070109	3
chr19	58150519	58176482	0
chr19	58460834	58512214	0
chr19	58472882	58500946	0
chr19	58612185	58697001	0
chr19	58616887	58700918	0
chr19	58621084	58710505	0
chr19	59776560	59801094	0
chr19	59793199	59836638	0
chr19	60142339	60186642	0
chr19	63010931	63046050	1
chr19	63022929	63057804	1
chr2	73679654	73757822	9
chr2	73728987	73794760	0
chr2	85394272	85425835	0

chr2	178123722	178192420	0
chr2	232899346	232938956	0
chr2	232937816	233013878	0
chr2	232950578	232984396	0
chr2	234158915	234230618	1
chr2	240617598	240634311	0
chr20	5399985	5433107	0
chr20	23701289	23789231	0
chr20	23709603	23799725	0
chr21	44782773	44824908	0
chr21	44782778	44858062	0
chr21	44783579	44872395	0
chr21	44794095	44873311	0
chr21	44795241	44882308	0
chr21	44817995	44846077	0
chr21	44818057	44836784	0
chr21	44835730	44846042	0
chr21	44899080	44930665	0
chr21	44910115	44926641	0
chr22	28086303	28150882	0
chr22	34950608	34995068	0
chr22	41172142	41254394	0
chr22	43632564	43642175	1
chr3	2543406	2552203	0
chr3	9997452	10042081	0
chr3	10002598	10043470	0
chr3	10004270	10066016	0
chr3	50433413	50472473	0
chr3	127283237	127331288	0
chr3	130286773	130330598	0
chr3	140206973	140246402	0
chr3	140221400	140246599	0
chr3	181868737	181890801	0
chr3	187109666	187192347	1
chr4	54795931	54816976	0
chr4	69764187	69860146	0
chr4	73625313	73693774	0
chr4	77019036	77028180	0
chr5	68806945	68812625	1
chr5	115593843	115608240	0
chr5	139347842	139432547	0

chr5	140168129	140190184	0
chr5	140187910	140203418	0
chr5	140455788	140462884	0
chr5	140455788	140484070	0
chr5	140455788	140512338	0
chr5	140455802	140540105	0
chr5	140455807	140497509	0
chr5	140455809	140544640	0
chr5	140455812	140534913	0
chr5	140461510	140512338	0
chr5	140461630	140518007	0
chr5	140461644	140540105	0
chr5	140461662	140534913	0
chr5	140461693	140544640	0
chr5	140482877	140554618	0
chr5	140482952	140544624	0
chr5	140482953	140512322	0
chr5	140482958	140497503	0
chr5	140482958	140540121	0
chr5	140482967	140534903	0
chr5	140482992	140518007	0
chr5	140496286	140544630	0
chr5	140496389	140576191	0
chr5	140496391	140540063	0
chr5	140496421	140512328	0
chr5	140496421	140570935	0
chr5	140496424	140518007	0
chr5	140496442	140534855	0
chr5	140511210	140518007	0
chr5	140511231	140549386	0
chr5	140511267	140596617	0
chr5	140511277	140544655	0
chr5	140516957	140570905	0
chr5	140516970	140601903	0
chr5	140516973	140607572	0
chr5	140516975	140554570	0
chr5	140516990	140561773	0
chr5	140517008	140534849	0
chr5	140533845	140554627	0
chr5	140533845	140561846	0
chr5	140533845	140607606	0

chr5	140533854	140544637	0
chr5	140539013	140561837	0
chr5	140539029	140607606	0
chr5	140539057	140596613	0
chr5	140543562	140596617	0
chr5	140543569	140561846	0
chr5	140543579	140585579	0
chr5	140548290	140607603	0
chr5	140548297	140561813	0
chr5	140548297	140596617	0
chr5	140553533	140561928	0
chr5	140553533	140596613	0
chr5	140559529	140570936	0
chr5	140560706	140607628	0
chr5	140560746	140576194	0
chr5	140560750	140601979	0
chr5	140569878	140576202	0
chr5	140569882	140601964	0
chr5	140569885	140607606	0
chr5	140575106	140607636	0
chr5	140575118	140601965	0
chr5	140575124	140596617	0
chr5	140584373	140596617	0
chr5	140584485	140607603	0
chr5	140595561	140601965	0
chr5	140595561	140607606	0
chr5	140600784	140607606	0
chr5	148395217	148475903	0
chr5	150255197	150291579	2
chr5	168075710	168102572	0
chr5	177092919	177105825	0
chr6	2943089	3002381	0
chr6	2948262	2997963	0
chr6	3099557	3125799	0
chr6	27883284	27891179	0
chr6	29170622	29251227	5
chr6	29649385	29664662	5
chr6	31357342	31439187	0
chr6	31359708	31444438	0
chr6	31890981	31905640	0
chr6	32125017	32144424	0

chr6	32125018	32138191	0
chr6	32131597	32144424	0
chr6	32131599	32138191	0
chr6	32137145	32144424	0
chr6	41273124	41326004	0
chr6	57019262	57064537	8
chr6	73999317	74017295	0
chr6	150354339	150428016	0
chr6	160800000	160880088	0
chr7	23592668	23685917	0
chr7	63323409	63399303	0
chr7	65735947	65777648	2
chr7	99791060	99810042	0
chr7	148884150	148927431	0
chr7	148922048	148994099	0
chr7	149931381	150010719	0
chr8	43328990	43346982	0
chr8	82709110	82769536	0
chr8	133390938	133400130	0
chr8	143910899	143916979	0
chr8	143951581	143996301	0
chr9	21356566	21431942	0
chr9	34234189	34265006	0
chr9	93926381	94008385	0
chr9	95125471	95169379	0
chr9	135093007	135175242	0
chr9	135099431	135170543	0
chrX	15573874	15633339	0
chrX	15592857	15648330	0
chrX	46203715	46279508	0
chrX	47698097	47777330	0
chrX	49487526	49530291	0
chrX	55261848	55327510	0
chrX	57159758	57180652	0
chrX	67766128	67826319	0
chrX	70486067	70553593	0
chrX	72132628	72223522	0
chrX	99996225	100030273	0
chrX	100637454	100697586	0
chrX	101266440	101284971	0
chrX	101791364	101856584	0

chrX	101792159	101888286	0
chrX	101801146	101864174	0
chrX	101853118	101888307	0
chrX	133983100	134013835	0
chrX	134773233	134858665	0
chrX	134790484	134858665	0
chrX	148376861	148417456	0
chrX	148392252	148415841	0
chrX	148577649	148650867	0
chrX	148578927	148652952	0
chrX	151985036	152003088	0
chrX	153465653	153535994	0

Table S8. CNVs within *AluY* hotspots

Chr	Start	End	CNVs with 50% reciprocal overlap with *AluY*
chr10	12130000	12220000	4
chr10	69830000	69920000	0
chr10	70080000	70170000	0
chr10	103730000	103820000	0
chr10	103780000	103870000	0
chr10	103880000	103970000	0
chr10	126430000	126520000	0
chr11	680000	770000	0
chr11	9330000	9420000	0
chr11	9430000	9520000	0
chr11	62130000	62220000	0
chr11	62230000	62320000	0
chr11	63180000	63270000	0
chr11	65180000	65270000	0
chr11	66080000	66170000	0
chr11	66580000	66670000	0
chr12	2880000	2970000	0
chr12	7930000	8020000	155
chr12	19380000	19470000	50
chr12	31780000	31870000	0
chr12	47780000	47870000	0
chr12	52030000	52120000	0
chr12	54930000	55020000	0
chr12	55330000	55420000	0
chr12	103130000	103220000	0
chr12	109280000	109370000	0

chr12	109330000	109420000	0
chr12	109380000	109470000	0
chr12	110480000	110570000	0
chr12	111280000	111370000	0
chr12	119180000	119270000	0
chr12	119380000	119470000	0
chr12	119430000	119520000	0
chr12	121480000	121570000	0
chr12	122230000	122320000	1
chr12	122430000	122520000	0
chr14	34080000	34170000	0
chr14	34130000	34220000	0
chr14	49180000	49270000	0
chr14	72980000	73070000	0
chr14	103380000	103470000	0
chr15	20580000	20670000	0
chr15	39430000	39520000	0
chr15	39480000	39570000	0
chr15	41830000	41920000	0
chr15	62430000	62520000	0
chr15	62630000	62720000	0
chr15	64380000	64470000	1
chr15	72980000	73070000	0
chr16	80000	170000	0
chr16	130000	220000	0
chr16	180000	270000	0
chr16	380000	470000	0
chr16	2180000	2270000	0
chr16	2830000	2920000	0
chr16	4230000	4320000	0
chr16	11780000	11870000	0
chr16	29580000	29670000	0
chr16	30280000	30370000	0
chr16	57180000	57270000	0
chr16	66130000	66220000	0
chr16	66330000	66420000	0
chr16	67780000	67870000	0
chr16	68830000	68920000	0
chr16	68880000	68970000	0
chr16	70380000	70470000	0
chr16	73980000	74070000	0

chr16	87780000	87870000	0
chr16	88230000	88320000	0
chr16	88330000	88420000	0
chr16	88380000	88470000	0
chr16	88430000	88520000	0
chr17	780000	870000	0
chr17	1080000	1170000	0
chr17	1130000	1220000	0
chr17	1180000	1270000	0
chr17	1230000	1320000	0
chr17	1480000	1570000	0
chr17	1580000	1670000	0
chr17	2080000	2170000	0
chr17	2130000	2220000	0
chr17	4630000	4720000	0
chr17	7080000	7170000	0
chr17	17080000	17170000	0
chr17	17130000	17220000	0
chr17	19680000	19770000	0
chr17	24130000	24220000	0
chr17	24680000	24770000	0
chr17	27230000	27320000	0
chr17	27280000	27370000	0
chr17	34180000	34270000	0
chr17	35580000	35670000	0
chr17	38380000	38470000	0
chr17	38480000	38570000	0
chr17	38530000	38620000	0
chr17	39530000	39620000	0
chr17	40580000	40670000	0
chr17	42930000	43020000	0
chr17	44330000	44420000	0
chr17	54430000	54520000	1
chr17	70630000	70720000	0
chr17	70680000	70770000	0
chr17	70880000	70970000	0
chr17	77130000	77220000	0
chr17	77180000	77270000	0
chr17	78030000	78120000	1
chr18	17430000	17520000	0
chr19	680000	770000	0

chr19	1230000	1320000	0
chr19	1830000	1920000	0
chr19	1930000	2020000	0
chr19	2430000	2520000	0
chr19	2780000	2870000	1
chr19	2830000	2920000	0
chr19	2980000	3070000	0
chr19	4030000	4120000	0
chr19	4180000	4270000	0
chr19	4380000	4470000	0
chr19	4680000	4770000	0
chr19	4730000	4820000	0
chr19	4780000	4870000	0
chr19	5630000	5720000	0
chr19	6230000	6320000	0
chr19	6280000	6370000	0
chr19	7730000	7820000	0
chr19	8330000	8420000	0
chr19	10180000	10270000	0
chr19	10280000	10370000	0
chr19	10430000	10520000	0
chr19	10530000	10620000	0
chr19	10780000	10870000	0
chr19	10880000	10970000	0
chr19	11030000	11120000	0
chr19	11080000	11170000	0
chr19	11530000	11620000	0
chr19	11980000	12070000	0
chr19	12480000	12570000	0
chr19	12580000	12670000	0
chr19	12730000	12820000	0
chr19	12880000	12970000	0
chr19	13730000	13820000	0
chr19	41280000	41370000	0
chr19	46330000	46420000	0
chr19	50980000	51070000	0
chr19	51580000	51670000	0
chr19	52280000	52370000	0
chr19	52330000	52420000	0
chr19	54030000	54120000	0
chr19	54680000	54770000	0

chr19	54880000	54970000	0
chr19	58080000	58170000	0
chr19	60080000	60170000	0
chr19	63130000	63220000	0
chr19	63280000	63370000	1
chr2	32380000	32470000	0
chr2	61580000	61670000	0
chr2	131530000	131620000	0
chr2	202880000	202970000	0
chr20	3730000	3820000	1
chr20	29630000	29720000	0
chr20	30730000	30820000	0
chr20	32680000	32770000	0
chr20	33830000	33920000	0
chr20	47180000	47270000	0
chr20	49830000	49920000	0
chr22	16330000	16420000	0
chr22	16380000	16470000	0
chr22	16430000	16520000	0
chr22	16480000	16570000	0
chr22	18780000	18870000	0
chr22	19530000	19620000	0
chr22	20030000	20120000	0
chr22	20180000	20270000	0
chr22	20430000	20520000	0
chr22	20530000	20620000	0
chr22	23480000	23570000	0
chr22	23580000	23670000	0
chr22	36280000	36370000	1
chr22	36630000	36720000	0
chr22	38830000	38920000	0
chr22	39630000	39720000	0
chr22	39980000	40070000	0
chr22	40080000	40170000	0
chr22	40380000	40470000	0
chr22	40430000	40520000	0
chr22	49080000	49189700	0
chr22	49130000	49220000	0
chr22	49380000	49470000	0
chr3	10130000	10220000	0
chr3	10180000	10270000	0

chr3	32680000	32770000	0
chr3	47480000	47570000	0
chr3	47730000	47820000	0
chr3	47780000	47870000	0
chr3	48180000	48270000	0
chr3	48880000	48970000	0
chr3	48930000	49020000	0
chr3	49030000	49120000	0
chr3	49180000	49270000	0
chr3	49330000	49420000	0
chr3	49380000	49470000	0
chr3	49430000	49520000	0
chr3	49730000	49820000	0
chr3	49930000	50020000	1
chr3	129830000	129920000	0
chr3	130330000	130420000	0
chr4	2430000	2520000	0
chr4	39380000	39470000	0
chr4	39430000	39520000	0
chr4	39630000	39720000	0
chr4	140380000	140470000	0
chr5	43180000	43270000	0
chr5	126080000	126170000	0
chr5	132330000	132420000	0
chr5	172330000	172420000	0
chr5	176330000	176420000	0
chr5	177480000	177570000	0
chr5	178880000	178970000	1
chr5	178930000	179020000	1
chr6	17380000	17470000	0
chr6	17780000	17870000	0
chr6	33380000	33470000	0
chr6	42880000	42970000	0
chr6	42930000	43020000	0
chr6	107430000	107520000	1
chr7	780000	870000	0
chr7	2280000	2370000	0
chr7	5130000	5220000	0
chr7	5430000	5520000	0
chr7	5530000	5620000	0
chr7	5780000	5870000	5

chr7	6030000	6120000	0
chr7	6280000	6370000	0
chr7	6380000	6470000	0
chr7	23480000	23570000	0
chr7	55830000	55920000	0
chr7	65130000	65220000	0
chr7	65180000	65270000	0
chr7	65230000	65320000	0
chr7	65680000	65770000	0
chr7	72530000	72620000	0
chr7	73130000	73220000	0
chr7	73630000	73720000	0
chr7	104780000	104870000	0
chr7	104880000	104970000	0
chr7	139780000	139870000	0
chr7	148330000	148420000	0
chr7	148380000	148470000	0
chr7	148430000	148520000	0
chr8	42680000	42770000	0
chr8	95780000	95870000	0
chr9	33980000	34070000	0
chr9	34130000	34220000	0
chr9	36530000	36620000	0
chr9	130430000	130520000	0
chr9	130630000	130720000	0
chr9	130780000	130870000	0
chr9	131830000	131920000	0
chr9	135930000	136020000	0
chr9	139580000	139670000	0
chr3	52530000	52620000	0
chr17	34780000	34870000	0
chr8	67680000	67770000	0
chr15	42580000	42670000	0
chr5	138630000	138720000	0
chr7	99480000	99570000	0
chr17	57330000	57420000	0
chr17	1480000	1570000	0
chr12	52080000	52170000	0
chrX	23880000	23970000	0
chr16	66180000	66270000	0
chr22	37180000	37270000	0

chr17	23980000	24070000	0
chr17	46580000	46670000	0
chr20	33680000	33770000	0
chr16	30580000	30670000	0
chr15	41830000	41920000	0
chr20	33730000	33820000	0
chr17	34880000	34970000	0
chr12	52880000	52970000	0
chr6	43030000	43120000	0
chr12	52230000	52320000	0
chr16	70380000	70470000	0
chr11	62130000	62220000	0
chr11	62180000	62270000	0
chr17	37230000	37320000	0
chr16	30530000	30620000	0
chr5	178930000	179020000	1
chr11	47380000	47470000	0
chr2	27430000	27520000	0
chr2	47830000	47920000	0
chr7	44830000	44920000	0
chr15	62480000	62570000	0
chr16	66430000	66520000	0
chr12	52030000	52120000	0
chr12	52930000	53020000	0
chr11	66130000	66220000	0
chr12	51880000	51970000	0
chr11	118380000	118470000	0
chr12	54680000	54770000	0
chr5	137730000	137820000	0
chr20	29730000	29820000	0
chr12	54980000	55070000	0
chr12	51930000	52020000	0
chr19	47480000	47570000	0
chr9	130330000	130420000	0
chr12	54480000	54570000	0
chr7	99880000	99970000	0
chr6	149980000	150070000	0
chr17	24830000	24920000	0
chr16	30930000	31020000	0
chr2	27380000	27470000	0
chr14	20980000	21070000	0

chr14	72580000	72670000	8
chr7	100080000	100170000	0
chr20	3030000	3120000	0
chr11	46680000	46770000	0
chr14	101530000	101620000	0
chr17	27680000	27770000	0
chr9	127080000	127170000	0
chr19	10630000	10720000	0
chr11	117880000	117970000	0
chr2	203080000	203170000	0
chr11	62280000	62370000	0
chr12	54930000	55020000	0
chr6	31830000	31920000	0
chr7	100530000	100620000	0
chr5	137830000	137920000	0
chr19	8380000	8470000	0
chr22	30030000	30120000	0
chr10	74880000	74970000	0
chr17	35530000	35620000	0
chr10	75030000	75120000	0
chr17	4730000	4820000	0
chr17	35430000	35520000	0
chr17	2130000	2220000	0
chr11	9380000	9470000	0
chr12	47330000	47420000	0
chr20	45380000	45470000	0
chr12	47730000	47820000	0
chr6	37180000	37270000	0
chr11	46480000	46570000	0
chr12	6630000	6720000	0
chr16	30980000	31070000	0
chr17	37380000	37470000	0
chr16	30780000	30870000	0
chr3	49480000	49570000	0
chr12	121530000	121620000	0
chr2	61530000	61620000	0
chr16	31080000	31170000	0
chr12	54830000	54920000	0
chr10	93930000	94020000	0
chr7	101630000	101720000	0
chr11	17030000	17120000	0

chr9	130680000	130770000	0
chr14	22430000	22520000	0
chr3	187930000	188020000	0
chr12	55280000	55370000	0
chrX	70480000	70570000	0
chr14	21030000	21120000	0
chr17	40180000	40270000	0
chr2	70230000	70320000	0
chr19	43980000	44070000	0
chr2	61580000	61670000	0
chr15	39580000	39670000	0
chr19	12630000	12720000	0
chr11	57180000	57270000	0
chr10	70130000	70220000	0
chr19	50630000	50720000	0
chr19	45530000	45620000	0
chr10	103730000	103820000	0
chr22	39830000	39920000	0
chr6	30680000	30770000	0
chr18	17330000	17420000	0
chr15	62530000	62620000	0
chr17	24080000	24170000	0
chr17	33730000	33820000	0
chr3	171530000	171620000	0
chr11	47180000	47270000	0
chr11	47530000	47620000	0
chr10	69730000	69820000	0
chr10	103980000	104070000	0
chr17	7080000	7170000	0
chr6	43180000	43270000	0
chr11	122380000	122470000	0
chr19	12680000	12770000	0
chr17	31930000	32020000	0
chr10	105080000	105170000	0
chr19	39480000	39570000	0
chr20	43880000	43970000	0
chr19	51030000	51120000	0
chr15	50080000	50170000	0
chr17	38380000	38470000	0
chr17	63330000	63420000	0
chr19	13880000	13970000	0

chr10	74080000	74170000	0
chr6	36630000	36720000	0
chrX	48280000	48370000	0
chr17	40080000	40170000	0
chr12	55080000	55170000	0
chr5	134030000	134120000	0
chr12	52180000	52270000	0
chr20	47230000	47320000	0
chr10	70280000	70370000	0
chr11	64280000	64370000	0
chr19	43630000	43720000	1
chr14	30580000	30670000	0
chr12	51980000	52070000	0
chr22	39880000	39970000	0
chr16	29730000	29820000	0
chr17	44680000	44770000	0
chrX	135730000	135820000	0
chr10	69280000	69370000	0
chr5	176430000	176520000	0
chr12	110930000	111020000	0
chr17	2080000	2170000	0
chr3	49780000	49870000	0
chr11	3630000	3720000	0
chr12	119430000	119520000	0
chr15	42530000	42620000	0
chr16	70430000	70520000	0
chr11	60880000	60970000	0
chr17	38430000	38520000	0
chr9	130480000	130570000	0
chr10	104030000	104120000	0
chr16	28780000	28870000	0
chr16	29780000	29870000	0
chr2	38780000	38870000	0
chr6	107630000	107720000	0
chrX	100180000	100270000	0
chr19	54730000	54820000	0
chrX	48780000	48870000	0
chr17	38080000	38170000	0
chr12	56080000	56170000	0
chr17	57380000	57470000	0
chr16	69030000	69120000	0

chrX	70380000	70470000	0
chr7	5330000	5420000	0
chr17	55230000	55320000	0
chr19	12780000	12870000	0
chr17	4630000	4720000	0
chr17	8080000	8170000	0
chr6	42880000	42970000	0
chr12	111080000	111170000	0
chr16	11830000	11920000	0
chr22	30130000	30220000	0
chr19	19580000	19670000	0
chr17	18130000	18220000	0
chr4	57430000	57520000	0
chr19	12880000	12970000	0
chrX	122880000	122970000	0
chr12	97430000	97520000	0
chr9	130780000	130870000	0
chr11	66080000	66170000	0
chr14	20830000	20920000	0
chrX	70280000	70370000	0
chr17	54580000	54670000	0
chr22	40130000	40220000	0
chr19	14030000	14120000	0
chr17	59130000	59220000	0
chr10	94180000	94270000	0
chr12	122330000	122420000	0
chr9	127480000	127570000	0
chr16	66380000	66470000	0
chr17	24180000	24270000	0
chr16	69080000	69170000	1
chr2	172480000	172570000	0
chr14	73430000	73520000	0
chr16	11730000	11820000	0
chr11	47680000	47770000	0
chr6	90580000	90670000	0
chr12	121330000	121420000	0
chr22	37230000	37320000	0
chr19	40980000	41070000	0
chr17	24980000	25070000	0
chr7	99830000	99920000	0
chr8	38180000	38270000	1

chr20	43930000	44020000	0
chr7	99530000	99620000	0
chr3	49880000	49970000	0
chr12	6580000	6670000	0
chr11	47130000	47220000	0
chr10	104180000	104270000	0
chr20	30330000	30420000	0
chr7	101180000	101270000	0
chr16	30880000	30970000	0
chr5	159430000	159520000	0
chr17	54130000	54220000	0
chr12	109030000	109120000	0
chr12	49730000	49820000	0
chr5	134130000	134220000	0
chr14	38680000	38770000	0
chr5	138930000	139020000	0
chr9	130730000	130820000	0
chr17	1930000	2020000	0
chr17	1230000	1320000	0
chr16	67730000	67820000	0
chr19	12730000	12820000	0
chr17	63380000	63470000	0
chr16	23430000	23520000	0
chr16	4780000	4870000	0
chr7	75730000	75820000	0
chr6	43580000	43670000	0
chr17	7980000	8070000	0
chr17	1330000	1420000	0
chr17	37180000	37270000	0
chr16	66230000	66320000	0
chr12	119530000	119620000	0
chr19	50980000	51070000	0

Table S9. List of all smaller hotspot CNVs

Genomic region (hg18)	Cases n=2,588	Controls n=580	Description	P-value	Odd ratio	Lower	Upper
chr12:7930000-8020000	22	4	AluYDels	0.468897	1.234528	0.417056	4.946843
chr14:72580000-72670000	5	3	AluYDels	0.958152	0.372458	0.072246	2.405182
chr7:5738853-5871832	2	0	AluYDels	0.66731	Inf	0.042067	Inf
chr10:12130000-12220000	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr12:19380000-19470000	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr19:2769955-3461478	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr19:2777247-2838715	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr19:43630000-43720000	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr22:36277660-36330258	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr22:38773641-39148257	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr6:107430000-107518731	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr7:5120000-5170000	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr7:5790188-5870000	1	0	AluYDels	0.816919	Inf	0.005762	Inf
chr16:69080000-69170000	0	1	AluYDels	1	0	0	8.740345
chr15:64375665-64479422	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr17:78030000-78250000	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr17:902154-1567639	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr17:987984-1250631	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr19:63295548-63370953	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr20:33776065-33833342	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr20:33839309-34020687	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr20:3725594-3842583	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr3:49900360-50063973	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr5:178871628-179015299	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr7:5595608-5889320	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr8:38158036-38286608	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr9:129863151-130919094	1	0	AluYDups	0.816919	Inf	0.005762	Inf
chr7:5738853-5871832	2	0	AluYDups	0.66731	Inf	0.042067	Inf

chr10:12127247-12207883	3	0	AluYDups	0.545061	Inf	0.09253	Inf
chr12:19380000-19470000	43	6	AluYDups	0.180159	1.616148	0.68047	4.668758
chr12:7930000-8020000	106	23	AluYDups	0.498171	1.034263	0.647082	1.718025
chr12:122230000-122320000	0	1	AluYDups	1	0	0	8.740345
chr17:54430000-54520000	0	1	AluYDups	1	0	0	8.740345
chr17:77975000-78100000	0	1	AluYDups	1	0	0	8.740345
chr10:100703151-100940746	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr11:125746831-125774417	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr16:74087525-74134014	1	1	MicroHSDels	0.966529	0.223961	0.002858	17.59861
chr19:58288427-58600264	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr2:178421751-178469907	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr2:213915176-213944138	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr3:162173742-162208106	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr4:38475453-38505894	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr4:54793337-54817034	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr4:54793337-54838343	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr6:26511226-26555479	1	1	MicroHSDels	0.966529	0.223961	0.002858	17.59861
chr8:15141259-16659481	1	0	MicroHSDels	0.816919	Inf	0.005762	Inf
chr19:58597470-58650878	2	0	MicroHSDels	0.66731	Inf	0.042067	Inf
chr19:62569652-62601491	2	0	MicroHSDels	0.66731	Inf	0.042067	Inf
chr2:232719656-232757154	2	0	MicroHSDels	0.66731	Inf	0.042067	Inf
chr20:60848-94170	2	0	MicroHSDels	0.66731	Inf	0.042067	Inf
chr6:52726617-52774620	2	1	MicroHSDels	0.911807	0.447936	0.023283	26.46388
chr9:17251976-17280216	2	0	MicroHSDels	0.66731	Inf	0.042067	Inf
chr11:56097952-56139937	3	1	MicroHSDels	0.844715	0.67205	0.053856	35.33607
chr11:56072176-56116063	4	0	MicroHSDels	0.445176	Inf	0.14783	Inf
chr12:51128713-51156641	4	0	MicroHSDels	0.445176	Inf	0.14783	Inf
chr19:58634062-58677821	4	1	MicroHSDels	0.771601	0.896294	0.08846	44.21515
chr6:52755032-52800115	7	1	MicroHSDels	0.553835	1.570119	0.201096	70.87615
chr17:36753663-36778230	9	0	MicroHSDels	0.161623	Inf	0.442469	Inf
chr8:71769142-71815740	11	1	MicroHSDels	0.325397	2.470979	0.357849	106.5229

chr9:5289794-5332656	24	8	MicroHSDels	0.884644	0.669363	0.289077	1.73274
chr17:36461664-36478593	63	17	MicroHSDels	0.800902	0.826354	0.473191	1.518468
chr8:48483461-48750000	0	1	MicroHSDels	1	0	0	8.740345
chr1:172813690-172929227	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr1:31178042-31198047	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr11:125746831-125774417	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr11:86628362-87450467	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr13:51119056-51167168	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr13:66468619-66503012	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr15:97274780-97826232	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr16:68923105-68964107	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr16:68924395-68961849	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr17:11687163-11709471	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr17:36753663-36778230	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr19:58288427-58600264	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr3:58773011-58810951	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr8:88455946-88680000	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr9:93399807-95962364	1	0	MicroHSDups	0.816919	Inf	0.005762	Inf
chr11:107733766-107757732	2	0	MicroHSDups	0.66731	Inf	0.042067	Inf
chr17:36461664-36478593	2	0	MicroHSDups	0.66731	Inf	0.042067	Inf
chr19:61398731-61435513	2	0	MicroHSDups	0.66731	Inf	0.042067	Inf
chr2:116261115-116290283	2	0	MicroHSDups	0.66731	Inf	0.042067	Inf
chr3:144245009-144290782	2	0	MicroHSDups	0.66731	Inf	0.042067	Inf
chr9:95888470-95919186	2	0	MicroHSDups	0.66731	Inf	0.042067	Inf
chr11:94395668-94425793	3	0	MicroHSDups	0.545061	Inf	0.09253	Inf
chr2:116241694-116394317	3	0	MicroHSDups	0.545061	Inf	0.09253	Inf
chr19:58634062-58677821	8	4	MicroHSDups	0.948148	0.446652	0.119177	2.033917
chr9:5289794-5332656	10	1	MicroHSDups	0.374287	2.245512	0.318355	97.60173
chr16:74087525-74134014	41	8	MicroHSDups	0.445431	1.150912	0.528909	2.858146
chr15:60080019-60101128	0	1	MicroHSDups	1	0	0	8.740345
chr22:43632564-43642175	1	0	MiniHSDels	0.816919	Inf	0.005762	Inf

chr3:187109666-187192347	1	0	MiniHSDels	0.816919	Inf	0.005762	Inf
chr5:150255197-150291579	1	1	MiniHSDels	0.966529	0.223961	0.002858	17.59861
chr6:29150022-29251227	1	0	MiniHSDels	0.816919	Inf	0.005762	Inf
chr6:57016532-57067336	1	0	MiniHSDels	0.816919	Inf	0.005762	Inf
chr7:149750000-149955000	1	0	MiniHSDels	0.816919	Inf	0.005762	Inf
chr6:29170622-29251227	3	1	MiniHSDels	0.844715	0.67205	0.053856	35.33607
chr6:29649385-29664662	3	2	MiniHSDels	0.954387	0.335569	0.038372	4.025953
chr6:57019262-57064537	3	1	MiniHSDels	0.844715	0.67205	0.053856	35.33607
chr2:73679654-73757822	9	0	MiniHSDels	0.161623	Inf	0.442469	Inf
chr19:63022929-63057804	1	0	MiniHSDups	0.816919	Inf	0.005762	Inf
chr6:57019262-57064537	1	0	MiniHSDups	0.816919	Inf	0.005762	Inf
chr6:57016532-57067336	8	1	MiniHSDups	0.487144	1.80006	0.240627	80.02642
chr7:65735947-65777648	2	0	MiniHSDups	0.66731	Inf	0.042067	Inf
chr19:58021000-58078109	3	0	MiniHSDups	0.545061	Inf	0.09253	Inf
chr2:234158915-234230618	0	1	MiniHSDups	1	0	0	8.740345

Note that none of the non-SD hotspot events were significantly enriched in cases compared to controls. Burden for smaller hotspot CNVs in cases versus controls: total in cases=490/2,588 versus 94/580; Fisher’s exact test, p=0.13, OR=1.2.

Table S10. Novel gene-disrupting smaller hotspot events exclusive to autism cases

Chr	Start	End	Size	Sample	Region	Breaking genes	Event	Inheritance	Control probes	Control CNVs
chr1	31178042	31198047	20005	AU020705	1p35.2	*PUM1*	Dup	Paternal	9	0
chr1	172813690	172929227	115537	14019.p1	1q25.1	*RABGAP1L*	Dup	Maternal	35	0
chr1	237850754	237872118	21364	12162.p1	1q43	*CHRM3*	Del	Paternal	12	1
chr2	213921245	213939591	18346	12239.p1	2q34	*SPAG16*	Dup	Maternal	7	0
chr2	232719656	232757154	37498	13551.p1	2q37.1	*DIS3L2*	Del	Paternal	10	0
chr2	232719656	232757154	37498	12392.p1	2q37.1	*DIS3L2*	Del	n/a	10	0
chr3	49900360	50063973	163613	11415.p1	3p21.31	*MONA, RBM6*	Dup	Maternal	84	0
chr3	58773011	58810951	37940	14265.p1	3p14.2	*C3Orf67*	Dup	Maternal	12	0
chr3	144245009	144290782	45773	12831.p1	3q23	*U2SURP*	Dup	Paternal	11	0
chr3	144245009	144290782	45773	11948.p1	3q23	*U2SURP*	Dup	Paternal	11	0
chr4	38475453	38505894	30441	14059.p1	4p14	*TLR1,TLR6*	Del	Paternal	35	0
chr4	54793337	54838343	45006	AU055005	4q12	*PDGFRA*	Del	Maternal	10	0
chr5	178871628	179015299	143671	13341.p1	5q35.3	*RUFY1*	Dup	Maternal	62	0
chr6	64845481	64864129	18648	AU029303	6q12	*EYS*	Del	Maternal	2	1
chr6	107250000	107630000	380000	12212.p1	6q21	*PDSS2*	Del	Paternal	182	0
chr6	107250000	107630000	88731	AU0039301	6q21	*BEND3*	Del	Maternal	37	0
chr7	5595608	5889320	293712	11121.p1	7p22.1	*RNF216*	Dup	Maternal	113	0
chr7	5738853	5871832	132979	12894.p1	7p22.1	*RNF216*	Dup	Maternal	54	0
chr7	5738853	5871832	132979	13651.p1	7p22.1	*RNF216*	Dup	Paternal	54	0
chr7	5738853	5871832	132979	13906.p1	7p22.1	*RNF216*	Del	De novo	54	0
chr7	5738853	5871832	132979	13995.p1	7p22.1	*RNF216*	Del	Paternal	54	0
chr7	5790188	5870000	79812	AU2157301	7p22.1	*No gene breaks*	Del	Paternal	40	0
chr7	133870079	133915213	45134	13503.p1	7q33	*AKR1B10*	Dup	Maternal	17	0
chr7	133870079	133915213	45134	13174.p1	7q33	*AKR1B10*	Dup	Maternal	17	0
chr8	38158036	38286608	128572	AU1631301	8p12	*WHSC1L1, BAG4*	Dup	Maternal	60	0
chr9	95888470	95919186	30716	13671.p1	9q22.31	*PTPDC1*	Dup	Maternal	15	0
chr9	95888470	95919186	30716	14486.p1	9q22.32	*PTPDC1*	Dup	Paternal	15	0

chr10	12127247	12207883	80636	13990.p1	10p14	*DHTKD1*	Dup	Paternal	35	0
chr10	12127247	12207883	80636	14209.p1	10p14	*DHTKD1*	Dup	Maternal	35	0
chr10	12127247	12207883	80636	AU022704	10p14	*DHTKD1*	Dup	n/a	35	0
chr11	94395668	94425793	30125	11227.p1	11q21	*KDM4DL*	Dup	Maternal	18	0
chr11	94395668	94425793	30125	14354.p1	11q21	*KDM4DL*	Dup	Maternal	18	0
chr11	94395668	94425793	30125	13612.p1	11q21	*KDM4DL*	Dup	Paternal	18	0
chr11	107733766	107757732	23966	11044.p1	11q22.3	*ATM*	Dup	Paternal	9	0
chr11	107733766	107757732	23966	11433.p1	11q22.3	*ATM*	Dup	Paternal	9	0
chr11	107733766	107757732	23966	12211.p1	11q22.3	*ATM*	Dup	Paternal	9	0
chr11	125746831	125774417	27586	AU1686301	11q24.2	*ST3GAL4*	Dup	Paternal	11	0
chr11	125746831	125774417	27586	AU2050301	11q24.2	*ST3GAL4*	Del	Maternal	11	0
chr12	54935839	55210466	274627	13006.p1	12q13.2	*RBMS2, CS*	Dup	Paternal	153	0
chr15	64375665	64479422	103757	11734.p1	15q22.31	*DIS3L, MAP2K1*	Dup	Maternal	46	0
chr16	68923105	68964107	41002	AU1699302	*16q22.1*	*DDX19A*	Dup	Maternal	26	0
chr16	68924395	68961849	37454	AU1300302	16q22.1	*DDX19*	Dup	Paternal	22	0
chr17	902154	1567639	665485	14226.p1	17p13.3	*TLCD2, ABR*	Dup	De novo	471	0
chr17	987984	1250631	262647	AU1575302	17p13.3	*YWHAE, ABR*	Dup	Paternal	147	0
chr19	2769955	3461478	691523	14458.p1	19p13.3	*GNA15, ZNF556*	Del	n/a	401	0
chr19	2777247	2838715	61468	14491.p1	19p13.3	*ZNF554*	Del	Paternal	42	0
chr19	58288427	58600264	311837	11233.p1	*19q13.41*	*ZNF415*	Dup	Paternal	190	0
chr19	58288427	58600264	311837	AU1400301	*19q13.41*	*ZNF415*	Del	Maternal	190	0
chr19	60121654	60273674	152020	12960.p1	19q13.42	*RDH13*	Dup	De novo	124	0
chr19	61398731	61435513	36782	14046.p1	*19q13.42*	*No gene breaks*	Dup	Paternal	12	0
chr19	61398731	61435513	36782	AU1107301	*19q13.42*	*No gene breaks*	Dup	n/a	12	0
chr19	63295548	63370953	75405	12716.p1	19p13.42	*ZSCAN18*	Dup	Paternal	30	0
chr20	3725594	3842583	116989	13664.p1	20p13	*CDC25B, PANK2*	Dup	Maternal	95	0
chr20	3725594	3842583	116989	AU022704	20p13	*SIGLEC1, PANK2*	Dup	n/a	95	0
chr20	33776065	33833342	57277	14473.p1	20q11.22	*RBM39, PHF20*	Dup	n/a	17	0
chr20	33839309	34020687	181378	13782.p1	20q11.22	*PHF20*	Dup	Maternal	51	0

					q11.23					
chr20	51520021	51561726	41705	AU1512301	20q13.2	*TSHZ2*	Dup	Maternal	15	0
chr22	36230567	36730248	499681	14440.p1	22q13	*CARD10*	Dup	Paternal	290	0
chr22	36277660	36330258	52598	AU3068301	22q13	*No gene breaks*	Dup	Paternal		
chr10[a]	100703151	100940746	237595	AU1959302	10q24.2	*HPSE2*	Del	Paternal	62	0
chr11[a]	86628362	87450467	822105	13631.p1	11q14.2	*TMEM135*	Dup	De novo	321	0
chr15[a]	97274780	97826232	551452	14443.p1	*15q26.3*	*IGF1R*	Dup	De novo	327	1
chr17[a]	5962718	7286326	1323608	11532.p1	17p13.1p 13.2	*NLGN2*	Dup	De novo	869	0
chr19[a]	7970001	8780000	809999	12426.p1	19p13.2	*FBN1, MYO1F*	Del	Maternal	442	0
chr2[a]	116241694	116394317	152623	13103.p1	2q14.2	*DPP10*	Dup	Paternal	56	0
chr2[a]	116241694	116394317	152623	13418.p1	2q14.2	*DPP10*	Dup	Paternal	56	0
chr2[a]	116241694	116394317	152623	14013.p1	2q14.2	*DPP10*	Dup	Paternal	56	0
chr2[a]	116261115	116290283	29168	11324.p1	2q14.2	*DPP10*	Dup	Maternal	8	0
chr2[a]	178421751	178469907	48156	12529.p1	2q31.2	*PDE11A*	Del	Paternal	14	0
chr20[a]	60848	94170	33322	11558.p1	20p13	*DEFB125 cluster*	Del	Paternal	25	0
chr20[a]	60848	94170	33322	13014.p1	20p13	*DEFB125 cluster*	Del	Paternal	25	0
chr22[a]	38773641	39148257	374616	12224.p1	22q13	*SGSM3, GRAP2*	Del	De novo	143	0
chr5[a]	1.37E+08	1.38E+08	691037	14478.p1	5q31.2	*CTNNA1, NME5*	Dup	De novo	307	0
chr8[a]	15141259	16659481	1518222	11501.p1	8p22	*TUSC3*	Del	Maternal	941	0
chr9[a]	93399807	95962364	2562557	13727.p1	9q22.31	*PTPDC1*	Dup	De novo	1095	0
chr9[a]	129863151	130919094	1055943	12235.p1	9q34	*CRAT, SLC25A25*	Dup	De novo	660	0

[a] Indicates hotspot-associated rearrangements. None of the events were observed in controls assayed using our hotspot-targeted array (N=580). Control events, defined here as CNVs that appear to be mediated by the repeat sequences of the hotspot, were identified from population controls ran on Illumina 1M SNP array platform (N=2,090) (Cooper et al., 2011).

Table S11. Exon-disrupting CNVs for autism candidate genes

Candidate gene(s)	Genomic coordinates (hg18)	Size (bp)	Samples	Gene breaking	CNV	Inheritance[a]	WTCCC Control Probes
A2BP1	chr16:6926113-7087773	161,660	AU1393301	yes	Del	Maternal	88
A2BP1	chr16:7049351-7151263	101,912	AU015903	yes	Del	Maternal	97
A2BP1	chr16:6745494-6846684	101190	11464.p1	yes	Del	Maternal	74
A2BP1	chr16:7506551-7592706	86,155	13861.p1	yes	Del	Maternal	95
A2BP1	chr16:7044796-7066958	22,162	14073.p1	yes	Del	Maternal	12
A2BP1	chr16:6306867-6328849	21,982	13414.p1	yes	Del	Maternal	13
ANO5	chr11:21806758-26716891	4,910,133	12066.p1	no	Dup	Maternal	2036
CACNA2D3	chr3:54575478-54781732	206,254	13358.p1	yes	Del	Maternal	92
CADPS2	chr7:121509923-121938724	428,801	11026.p1	yes	Dup	Paternal	143
CNR1	chr6:88898857-88923579	24,722	12279.p1	yes	Dup	n/a	21
CNR1	chr6:88898857-88923579	24,722	AU2831301	yes	Dup	Maternal	21
CNR1	chr6:88898857-88910225	11,368	13890.p1	yes	Dup	Maternal	13
CNR1	chr6:88898857-88923579	24,722	11368.p1	yes	Dup	Maternal	21
CNR1	chr6:88898857-88923579	24,722	AU1379301	yes	Dup	Maternal	21
CNR1	chr6:88898857-88923579	24,722	13481.p1	yes	Dup	Paternal	21
CNR1	chr6:88898857-88923579	24,722	13822.p1	yes	Dup	Paternal	21
CNR1	chr6:88898857-88923579	24,722	control	yes	Dup	n/a	21
CNTNAP2	chr7:144778600-145608237	829,637	13568.p1	yes	Dup	Maternal	269
CNTNAP2	chr7:147660726-147707361	46,635	13395.p1	yes	Del	Paternal	45
CTNNA3	chr10:67981151-68126494	145,343	13001.p1	yes	Del	De novo	72
CTNNA3	chr10:67726795-67869383	142,588	12108.p1	yes	Del	Maternal	80
CTNNA3	chr10:67726795-67869383	142,588	13557.p1	yes	Del	Maternal	80
CTNNA3	chr10:67981151-68085796	104,645	13494.p1	yes	Del	Maternal	46
CTNNA3	chr10:67953343-68029641	76,298	AU1391302	yes	Del	Maternal	30
CTNNA3	chr10:68186325-68228806	42,481	11232.p1	yes	Del	Maternal	20
CTNNA3	chr10:67953343-68167013	213670	11711.p1	yes	Del	Paternal	100
CTNNA3	chr10:67992425-68186725	194,300	13619.p1	yes	Del	Paternal	95
CTNNA3	chr10:67953343-68114041	160,698	13043.p1	yes	Del	Paternal	79
CTNNA3	chr10:67726795-67869383	142,588	11463.p1	yes	Del	Paternal	80
CTNNA3	chr10:67726795-67869383	142,588	12153.p1	yes	Del	Paternal	80
CTNNA3	chr10:68029241-68126494	97,253	11156.p1	yes	Del	Paternal	51
CTNNA3	chr10:67932135-68018603	86,468	11299.p1	yes	Del	Paternal	37
CTNNA3	chr10:67992425-68064617	72,192	13005.p1	yes	Del	Paternal	33

CTNNA3	chr10:68018203-68085796	67,593	13541.p1	yes	Del	Paternal	29
CTNNA3	chr10:67525235-67568916	43,681	13526.p1	yes	Del	Paternal	17
CTNNA3	chr10:67992425-68283921	291,496	14487.p1	yes	Del	Paternal	149
CTNNA3	chr10:67881357-68018603	137,246	AU0911301	yes	Dup	n/a	58
CTNNA3	chr10:68046321-68155756	109,435	AU0946302	yes	Del	Paternal	57
CTNNA3	chr10:67843269-67932535	89,266	14260.p1	yes	Del	Paternal	34
CTNNA3	chr10:67992425-68077952	85,527	13997.p1	yes	Del	Maternal	40
CTNNA3	chr10:68029241-68064617	35,376	13658.p1	yes	Del	Paternal	17
CTNNA3	chr10:67726795-67921741	194,946	control	yes	Del	n/a	101
CTNNA3	chr10:67953343-68018603	65,260	control	yes	Del	n/a	26
CTNNA3	chr10:68029241-68064617	35,376	control	yes	Del	n/a	17
CTNNA3	chr10:68097560-68316501	218,941	control	yes	Del	n/a	117
CTNND2	chr5:11398907-11491980	93073	12289.p1	yes	Del	De novo	63
DIAPH3	chr13:59572895-59589781	16,886	12628.p1	yes	Del	Maternal	5
DISC1	chr1:229898644-229999798	101,154	11712.p1	yes	Del	Maternal	45
DISC1	chr1:228187721-230201264	2,013,543	12162.p1	yes	Del	Paternal	1066
DNAH5	chr5:13855108-14232579	377,471	11710.p1	yes	Dup	Paternal	242
DOCK1	chr10:128130613-135228892	7,098,279	14091.p1	no	Del	De novo	4385
DOCK1	chr10:128775742-128905024	129,282	14002.p1	yes	Del	Paternal	67
DPP10	chr2:115810880-116702422	891,542	11033.p1	yes	Dup	Maternal	291
DPP10[b]	chr2:116261115-116290283	29,168	11324.p1	yes	Dup	Maternal	8
DPP10[b]	chr2:116241694-116394317	152,623	13103.p1	yes	Dup	Paternal	56
DPP10[b]	chr2:116241694-116394317	152,623	13418.p1	yes	Dup	Paternal	56
DPP10[b]	chr2:116241694-116394317	152,623	14013.p1	yes	Dup	Paternal	56
DPP6	chr7:153590919-154251382	660,463	11843.p1	yes	Del	Maternal	404
DPP6	chr7:153084786-153282406	197,620	AU079104	yes	Dup	Maternal	73
DPP6	chr7:153388658-153569013	180,355	12351.p1	yes	Dup	Maternal	61
DPP6[b]	chr7:152168193-153286018	1,117,825	12185.p1	yes	Dup	Paternal	504
DPP6	chr7:153117996-153291033	173,037	AU025803	yes	Dup	Maternal	72
DPP6	chr7:153095638-153385049	289,411	control	yes	Dup	n/a	84
EML1	chr14:99464769-99483955	19,186	AU012204	yes	Dup	n/a	10
EML1	chr14:99464769-99483955	19,186	AU022203	yes	Dup	Paternal	10
EML1	chr14:99464769-99483955	19,186	AU071803	yes	Dup	Paternal	10
EML1	chr14:99464769-99483955	19,186	AU1053302	yes	Dup	n/a	10
EPHA6	chr3:96570608-100896278	4,325,670	14204.p1	no	Dup	Maternal	1384
ERBB4	chr2:212770528-213122490	351,962	11811.p1	yes	Del	Maternal	181

F13A1	chr6:5840343-6112556	272,213	14442.p1	yes	Dup	De novo	161
FHIT	chr3:60530016-60800676	270,660	12058.p1	yes	Del	De novo	186
FHIT	chr3:58463257-60148958	1,685,701	AU2554301	yes	Dup	Maternal	948
FHIT	chr3:60768687-66402436	5,633,749	13830.p1	yes	Del	De novo	2735
FOXP1	chr3:71209239-71450066	240,827	AU1718301	yes	Del	n/a	110
FOXP1, SUCLG2	chr3:67226398-72324318	5,097,920	12032.p1	no	Del	Maternal	2068
GABRB3	chr15:24521070-25133204	612,134	13355.p1	yes	Dup	De novo	308
GALNT13	chr2:154968071-154990525	22,454	14184.p1	yes	Del	Paternal	8
GIMAP8	chr7:149764636-149968584	203,948	AU016204	no	Del	Paternal	90
GPC6	chr13:93475099-93551130	76,031	AU1165302	yes	Del	n/a	24
GRID2	chr4:93501225-94634131	1,132,906	AU1286301	yes	Dup	De novo	328
GRID2	chr4:93607735-93750838	143,104	13638.p1	yes	Del	Paternal	44
GRIN2A	chr16:9999349-10427936	428,587	AU063004	yes	Dup	Maternal	217
HYDIN[b]	chr16:68859156-72903506	4,044,350	12691.p1	no	Del	De novo	1811
HYDIN[b]	chr16:68832773-70006429	1,173,656	AU1551302	no	Dup	Paternal	512
HYDIN[b]	chr16:68883052-69808186	925,134	13769.p1	yes	Dup	De novo	395
IARS	chr9:93459971-95893617	2,433,646	13727.p1	no	Dup	De novo	1008
IARS	chr9:94077839-94124216	46,377	14039.p1	yes	Dup	Paternal	21
ICA1, NXPH1	chr7:8148775-8495272	346,497	14195.p1	yes	Dup	Paternal	205
ICA1, NXPH1	chr7:8148775-8495272	346,497	AU003705	yes	Dup	Paternal	205
ICA1, NXPH1	chr7:8159219-8461473	302,254	AU1081301	yes	Dup	Paternal	183
IQGAP2	chr5:75865663-75950687	85,024	AU0980302	yes	Del	Paternal	50
KANK1[b]	chr9:193904-1655435	1,461,531	14349.p1	no	Dup	Maternal	1206
KANK1	chr9:418382-593694	175,312	13879.p1	yes	Del	Maternal	176
KANK1	chr9:425727-499569	73,842	AU1483302	yes	Del	Maternal	62
KANK1	chr9:329883-524731	194,848	11630.p1	yes	Dup	Maternal	189
KANK1	chr9:622915-1053928	431,013	12316.p1	yes	Dup	Paternal	330
KANK1	chr9:162164-546825	384,661	AU1496302	yes	Dup	Paternal	351
KANK1	chr9:524331-730114	205,783	11909.p1	yes	Dup	Paternal	240
KANK1	chr9:406092-607282	201,190	AU001505	yes	Dup	Paternal	214
KANK1	chr9:524331-803740	279,409	control	yes	Dup	n/a	293
KHDRBS2	chr6:57643044-62975372	5,332,328	13521.p1	yes	Dup	Maternal	511
KHDRBS2	chr6:57591164-62500813	4,909,649	12430.p1	yes	Dup	Maternal	400
KHDRBS2	chr6:62694594-62985285	290,691	AU062003	yes	Dup	Paternal	88
KIAA1586[b]	chr6:57019780-57064460	44,680	AU1559301	yes	Del	Maternal	16
KIAA1586[b]	chr6:57019780-57064460	44,680	11933.p1	yes	Dup	n/a	16

KIAA1586[b]	chr6:57019780-57064460	44,680	11425.p1	yes	Dup	Paternal	16
KIAA1586[b]	chr6:57019780-57064460	44,680	11468.p1	yes	Dup	Paternal	16
KIAA1586[b]	chr6:57019780-57064460	44,680	12106.p1	yes	Dup	Paternal	16
KIAA1586[b]	chr6:57019780-57064460	44,680	13415.p1	yes	Del	Paternal	16
KIAA1586[b]	chr6:57019780-57064460	44,680	AU1187301	yes	Dup	Paternal	16
KIAA1586[b]	chr6:57019780-57064460	44,680	13865.p1	yes	Del	Maternal	16
KIAA1586[b]	chr6:57019780-57064460	44,680	control	yes	Del	n/a	16
MBD5	chr2:148438643-148546784	108,141	13595.p1	yes	Del	De novo	40
MBD5	chr2:148538759-148686256	147,497	11917.p1	yes	Dup	Maternal	33
MBD5	chr2:148566978-148768357	201,379	12390.p1	yes	Del	Paternal	45
MCC	chr5:112383353-112468610	85,257	14013.p1	yes	Del	Paternal	53
MCC	chr5:112486865-112516303	29,438	13833.p1	yes	Del	Maternal	22
MCPH1	chr8:6117093-6290589	173,496	12861.p1	yes	Dup	Maternal	147
MCPH1	chr8:6254308-6267453	13,145	13930.p1	yes	Del	Maternal	13
MCPH1	chr8:6274682-6277754	3,072	11818.p1	yes	Del	Maternal	6
MCPH1	chr8:6117093-6290589	173,496	13802.p1	yes	Dup	Paternal	147
MCPH1, DLGAP2	chr8:166252-6913476	6,747,224	13700.p1	no	Del	Maternal	6259
MET	chr7:114451937-116143324	1,691,387	12053.p1	yes	Dup	Maternal	483
MKL2	chr16:13460682-14687836	1,227,154	13691.p1	no	Dup	Maternal	520
MLL3	chr7:151635262-151686760	51,498	12957.p1	yes	Del	De novo	24
MLL5	chr7:104379293-104510086	130,793	14255.p1	yes	Dup	Maternal	30
MPHOSPH8	chr13:19027522-19503622	476,100	AU1744301	no	Dup	Maternal	115
MPHOSPH8	chr13:18991146-19470342	479,196	14044.p1	no	Dup	Paternal	117
MPHOSPH8	chr13:19133515-19223738	90,223	13784.p1	yes	Dup	De novo	20
NLGN1	chr3:175418372-175993222	574,850	13697.p1	yes	Dup	Maternal	236
NRXN1	chr2:50645921-51105202	459,281	13962.p1	yes	Del	De novo	198
NRXN1	chr2:51099065-51161671	62606	13580.p1	yes	Del	De novo	16
NRXN1	chr2:50886326-51115410	229,084	AU2681301	yes	Del	Maternal	88
NRXN1	chr2:50882413-51256102	373,689	AU1495303	yes	Del	Maternal	135
NRXN1	chr2:50996153-51079874	83,721	12232.p1	yes	Del	Maternal	32
NRXN1	chr2:51001812-51105202	103,390	14443.p1	yes	Del	Paternal	42
NRXN1	chr2:51090304-51182095	91,791	13609.p1	yes	Del	Paternal	28
NRXN3	chr14:78505420-78574596	69,176	13367.p1	yes	Del	Paternal	21
NTRK3	chr15:86443548-86546448	102,900	11944.p1	yes	Del	Paternal	48
NXPH1	chr7:8751880-8791093	39,213	14240.p1	yes	Del	Paternal	40
PARK2	chr6:162641150-162832556	191,406	14091.p1	yes	Dup	De novo	100

PARK2	chr6:162488727-162771149	282,422	12996.p1	yes	Del	Maternal	159
PARK2	chr6:162641150-162832556	191,406	11577.p1	yes	Dup	Maternal	100
PARK2	chr6:162304379-162481364	176,985	12937.p1	yes	Del	Maternal	73
PARK2	chr6:162677927-162787382	109,455	AU0039301	yes	Del	Maternal	55
PARK2	chr6:162256999-162711888	454,889	12864.p1	yes	Del	Paternal	224
PARK2	chr6:162623916-162979393	355,477	AU079605	yes	Dup	Paternal	168
PARK2	chr6:162168121-162496425	328,304	13151.p1	yes	Dup	Paternal	154
PARK2	chr6:162692702-162951135	258,433	11947.p1	yes	Dup	Paternal	136
PARK2	chr6:162374443-162614988	240,545	AU1056301	yes	Del	Paternal	144
PARK2	chr6:162598188-162762550	164,362	AU1340302	yes	Del	Paternal	74
PARK2	chr6:162781786-162841793	60,007	AU0839301	yes	Del	Paternal	37
PARK2	chr6:162605130-162661068	55,938	AU011704	yes	Del	Paternal	24
PARK2	chr6:162545343-162795400	250,057	12389.p1	yes	Dup	Paternal	116
PARK2	chr6:162641150-162832556	191,406	13115.p1	yes	Dup	Paternal	100
PARK2	chr6:162641150-162832556	191,406	13874.p1	yes	Dup	Maternal	100
PARK2	chr6:162641150-162832556	191,406	14268.p1	yes	Dup	Paternal	100
PARK2	chr6:162291421-162414184	122,763	13407.p1	yes	Del	Paternal	50
PARK2	chr6:162248070-162366075	118,005	13058.p1	yes	Dup	Paternal	43
PARK2	chr6:162556092-162657434	101,342	14153.p1	yes	Del	Paternal	38
PARK2	chr6:162734379-162816234	81,855	11236.p1	yes	Dup	Maternal	51
PARK2	chr6:162978993-163060311	81,318	14056.p1	yes	Del	Maternal	14
PARK2	chr6:162568520-162614988	46,468	13559.p1	yes	Dup	Paternal	18
PARK2	chr6:162545343-162568920	23,577	14234.p1	yes	Del	Maternal	10
PARK2	chr6:162641150-162832556	191,406	control	yes	Dup	n/a	100
PARK2	chr6:162641150-162826779	185,629	control	yes	Dup	n/a	98
PARK2	chr6:162677927-162787382	109,455	control	yes	Del	n/a	55
PCDH9	chr13:66380470-66595448	214,978	13708.p1	yes	Dup	Paternal	75
PDE4A	chr19:10437100-10447425	10,325	14075.p1	yes	Dup	Maternal	3
PLCB1	chr20:8043384-8523661	480,277	13817.p1	yes	Del	Maternal	265
PLCB1	chr20:8043384-8523661	480,277	14249.p1	yes	Del	Maternal	265
PLCB1	chr20:8043384-8523661	480,277	AU2689306	yes	Dup	Maternal	265
PLCB1	chr20:7514079-8271234	757,155	11013.p1	yes	Dup	Maternal	358
PLCB1	chr20:8043384-8523661	480,277	AU1887301	yes	Dup	Paternal	265
PLCB1	chr20:7065537-8523661	1,458,124	13085.p1	yes	Dup	Paternal	696
POGZ	chr1:149599548-149673824	74,276	12691.p1	yes	Dup	Maternal	39
RGS7	chr1:239564337-239904109	339,772	14240.p1	yes	Dup	Maternal	173

ROBO1	chr3:77925115-84485593	6,560,478	13877.p1	no	Del	De novo	1736
ROBO1	chr3:78300538-78990968	690,430	13094.p1	yes	Dup	Paternal	226
RSRC1	chr3:159632251-160281110	648,859	12215.p1	yes	Dup	Paternal	249
SEMA5A	chr5:9152479-9300739	148,260	13642.p1	yes	Del	De novo	89
SLC1A1	chr9:4313244-5015973	702,730	14363.p1	yes	Dup	Maternal	504
SLC1A1	chr9:4461017-4735859	274,843	13572.p1	yes	Dup	Paternal	226
TBC1D4	chr13:74826997-74918087	91,090	AU1565302	yes	Del	Paternal	39
THSD7A	chr7:11167884-12440226	1,272,342	11398.p1	yes	Dup	Paternal	780
ZMYND11	chr10:109596-245317	135,721	12843.p1	yes	Dup	De novo	41
ZMYND11	chr10:109596-245317	135,721	12956.p1	yes	Dup	Paternal	41

[a] Inheritance information could not be assessed in controls as well a subset of autistic probands in which parental DNA samples were unavailable or the CNV call was unclear. [b] Hotspot-mediated or -associated CNV.

Table S12. List of CNVs identified in the genomic backbone (non-hotspot regions)

SSC ID	Chr	Start (kbp)	Stop (kbp)	Size (kbp)	#Probes	Score	Gene Names
11000.p1	chr1	920.5	923.5	3	49	Dup	*TGFBR3, BRDT, EPHX4, BTBD8*
11008.p1	chr18	3.6	6.5	2.9	41	Dup	*COLEC12, CETN1, CLUL1, C18orf56, TYMS*
11014.p1	chr4	570.5	573.1	2.55	21	Dup	*SRP72, ARL9, HOPX*
11026.p1	chr7	1215.1	1219.4	4.29	74	Dup	*AASS, FEZF1, CADPS2*
11033.p1	chr2	1158.1	1167	8.92	137	Dup	*DPP10*
11046.p1	chr3	41.4	48.4	7.08	75	Del	*SETMAR, SUMF1, ITPR1, EGO*
11047.p1	chr14	937.1	945.8	8.76	59	Dup	*PPP4R4, SERPINA10, SERPINA6, SERPINA1, SERPINA9, SERPINA12, SERPINA4, SERPINA5, SERPINA3, SERPINA13, GSC*
11064.p1	chr2	336	343.5	7.53	60	Dup	*RASGRP3, FAM98A, MYADML*
11141.p1	chr1	2264	2268	3.99	66	Dup	*GUK1, GJC2, C1orf69, OBSCN, TRIM11, TRIM17, HIST3H3, HIST3H2A, HIST3H2BB, RNF187*
11141.p1	chr2	2406.2	2425.7	19.52	402	Dup	*OR6B2, LOC643905, OR6B3, MYEOV2, OTOS, GPC1, PP14571, ANKMY1, DUSP28, RNPEPL1, CAPN10, GPR35, AQP12B, AQP12A, KIF1A, AGXT, C2orf54, SNED1, MTERFD2, PASK, PPP1R7, ANO7, HDLBP, SEPT2, FARP2, STK25, BOK, THAP4, ATG4B, DTYMK, ING5, D2HGDH, GAL3ST2, NEU4, PDCD1, C2orf85*
11141.p1	chr8	134	147.1	13.01	108	Dup	*DLC1, C8orf48, SGCZ*
11141.p1	chr8	433.3	476.6	43.24	98	Dup	*POTEA*
11141.p1	chr8	1438.2	1456.7	18.41	286	Dup	*LYPD2, LYNX1, LY6D, GML, CYP11B1, CYP11B2, LOC100133669, LY6E, C8orf31, LY6H, GPIHBP1, ZFP41, GLI4, ZNF696, TOP1MT, C8orf51, RHPN1, MAFA, ZC3H3, GSDMD, C8orf73, NAPRT1, EEF1D, TIGD5, PYCRL, TSTA3, ZNF623, ZNF707, BREA2, MAPK15, FAM83H, SCRIB, PUF60, NRBP2, EPPK1, PLEC1, PARP10, GRINA, SPATC1, OPLAH, EXOSC4, GPAA1, CYC1, SHARPIN, MAF1, KIAA1875, C8orf30A, HEATR7A, SCXB, SCXA, C8orf30A, BOP1, SCXB, SCXA, HSF1, DGAT1, SCRT1, C8ORFK29, FBXL6, GPR172A, ADCK5, CPSF1, SLC39A4, VPS28, NFKBIL2, CYHR1, KIFC2*
11161.p1	chr1	2454.6	2467.9	13.3	206	Del	*LOC441931, VN1R5, ZNF496, NLRP3, OR2B11, OR2W5, LOC148824, OR2C3, C1orf150, OR2G2, OR2G3, OR13G1, OR6F1, OR1C1, OR14A16, OR11L1, TRIM58, OR2W3, OR2T8, OR2L13, OR2L8, OR2AK2, OR2L1P, OR2L2, OR2L3, OR2M1P, OR2M5, OR2M2, OR2M3, OR2M4, OR2T33, OR2T12, OR2M7, OR14C36, OR2T4, OR2T6, OR2T1, OR2T2, OR2T3, OR2T5, OR2G6*
11181.p1	chr14	882.4	885.3	2.94	22	Dup	*EML5, TTC8*
11190.p1	chr4	1087.2	1090.9	3.75	29	Dup	*PAPSS1, SGMS2, CYP2U1*

11198.p1	chr8	133.9	136.5	2.61	21	Dup	*DLC1, C8orf48*
11219.p1	chr4	28.8	31.6	2.79	53	Dup	*ADD1, MFSD10, C4orf10, NOP14, GRK4, HTT*
11225.p1	chr8	39.9	43.5	3.59	175	Del	*CSMD1*
11233.p1	chr15	668.9	719.4	50.48	2,102	Del	*ANP32A, NOX5, SPESP1, TMEM84, GLCE, PAQR5KIF23, RPLP1, LOC145837, C15orf50, TLE3, UACA, LARP6, THAP10, LRRC49, LOC196993, THSD4, NR2E3, MYO9A, SENP8, GRAMD2, PKM2, PARP6, BRUNOL6, HEXA, C15orf34, TMEM202, ARIH1, GOLGA6B, HIGD2B, BBS4, ADPGK, NEO1, HCN4, C15orf60, NPTN, CD276, C15orf59*
11242.p1	chr14	211.9	214.8	2.94	29	Dup	*OR4E2*
11299.p1	chr6	1187.1	1192.3	5.16	68	Dup	*SLC35F1, C6orf204, BRD7P3, PLN*
11301.p1	chr1	2434.5	2438	3.48	27	Del	*KIF26B*
11353.p1	chr17	318.4	333.5	15.18	2,570	Del	*TBC1D3C, TBC1D3G, ZNHIT3, MYO19, PIGW, GGNBP2, DHRS11, MRM1, LHX1, AATF, ACACA, C17orf78, TADA2L, DUSP14, AP1GBP1, DDX52, HNF1B, LOC284100*
11415.p1	chr3	112.1	114.9	2.74	58	Dup	*HRH1, ATG7*
11444.p1	chr17	99.2	103.6	4.32	114	Dup	*GAS7, MYH13, MYH8, MYH4, MYH1*
11464.p1	chr3	650.9	653.7	2.76	28	Dup	*MAGI1*
11469.p1	chr1	1737	1740.3	3.28	48	Dup	*TNR*
11480.p1	chr6	260.8	263.9	3.1	25	Dup	*TRIM38, HIST1H1A, HIST1H3A, HIST1H4A, HIST1H4B, HIST1H3B, HIST1H2AB, HIST1H2BB, HIST1H3C, HIST1H1C, HFE, HIST1H4C, HIST1H1T, HIST1H2BC, HIST1H2AC, HIST1H1E, HIST1H2BD, HIST1H2BD, HIST1H2BE, HIST1H4D, HIST1H3D, HIST1H2AD, HIST1H2BF, HIST1H4E, HIST1H2BG, HIST1H2AE, HIST1H3E, HIST1H1D, HIST1H4F, HIST1H4G, HIST1H3F, HIST1H2BH, HIST1H3G, HIST1H2BI*
11483.p1	chr12	1233.4	1236.7	3.31	80	Dup	*FAM101A, NCOR2*
11483.p1	chr22	363.4	367	3.63	52	Dup	*GGA1, SH3BP1, PDXP, LGALS1, NOL12, TRIOBP, H1F0, GCAT, GALR3, ANKRD54, EIF3L, MICALL1, C22orf23, POLR2F, SOX10*
11483.p1	chr4	234.7	237.3	2.54	20	Dup	*PPARGC1A*
11483.p1	chr5	1389.9	1392.7	2.75	28	Dup	*CXXC5, PSD2, NRG2*
11496.p1	chr14	651.2	654.7	3.59	24	Dup	*FUT8*
11519.p1	chr10	1218.1	1224.8	6.72	51	Del	*PPAPDC1A*
11519.p1	chr6	1689.9	1695.4	5.45	51	Dup	*THBS2*
11532.p1	chr17	60.4	72.6	12.2	137	Dup	*AIPL1, FAM64A, PITPNM3, KIAA0753, TXNDC17, MED31, C17orf100, SLC13A5, XAF1, FBXO39, TEKT1, ALOX12P2, ALOX12, RNASEK, C17orf49,*

							BCL6B, SLC16A13, SLC16A11, CLEC10A, ASGR2, ASGR1, DLG4, ACADVL, DVL2, PHF23, GABARAP, DULLARD, C17orf81, CLDN7, SLC2A4, YBX2, EIF5A, GPS2, NEURL4, ACAP1, KCTD11, TMEM95, TNK1, PLSCR3, C17orf61, NLGN2
11542.p1	chr11	1070.5	1073.5	3.04	58	Dup	*SLN, SLC35F2, RAB39*
11544.p1	chr6	1689.9	1695.4	5.45	51	Dup	*THBS2*
11581.p1	chr16	752.1	763.2	11.07	118	Dup	*MON1B, LOC100130958, ADAMTS18, NUDT7*
11651.p1	chr21	434.7	437.5	2.75	12	Dup	*SIK1, C21orf125, C21orf84*
11651.p1	chr9	1047.5	1050.3	2.82	30	Dup	*CYLC2*
11689.p1	chr10	9	13.8	4.76	58	Dup	*LARP5, GTPBP4, IDI2, C10orf110, IDI1, WDR37, C10orf139, ADARB2*
11756.p1	chr6	577.7	621.8	44.05	62	Dup	*GUSBL2*
11756.p1	chr6	1003.9	1007.5	3.64	29	Dup	*MCHR2*
11843.p1	chr7	1535.9	1542.5	6.6	51	Del	*DPP6*
11851.p1	chr3	5.5	14.3	8.84	75	Dup	*CNTN6*
11878.p1	chr3	605.1	608	2.86	21	Del	*FHIT*
11905.p1	chr3	124.5	128.3	3.76	57	Dup	*TSEN2, MKRN2, RAF1, TMEM40, CAND2*
11944.p1	chr17	153.8	165.3	11.49	131	Dup	*FAM18B2, CDRT1, TRIM16, ZNF286A, TBC1D26, MEIS3P1, ADORA2B, ZSWIM7, TTC19, NCOR1, PIGL, CENPV, UBB, TRPV2, C17orf45, SNORD49B, SNORD49A, SNORD65, C17orf76, ZNF287, ZNF624*
11962.p1	chr10	527	544.2	17.23	252	Dup	*PRKG1, CSTF2T, DKK1, MBL2*
11986.p1	chr20	126.9	132.8	5.89	46	Del	*SPTLC3, ISM1*
11989.p1	chr5	151.6	159.2	7.63	61	Dup	*FBXL7*
11999.p1	chr3	5.5	14.3	8.84	75	Dup	*CNTN6*
12007.p1	chr15	184.3	267.5	83.13	3,479	Dup	*LOC727832, GOLGA8C, LOC646214, CXADRP2, POTEB, LOC727924, OR4M2, OR4N4, LOC650137, GOLGA8D, LOC283767, TUBGCP5, CYFIP1, NIPA2, NIPA1, WHAMML1, GOLGA9P, HERC2P2, GOLGA8E, MKRN3, MAGEL2, NDN, PWRN2, PWRN1, C15orf2, SNRPN, SNURF, SNORD107, PAR-SN, PAR5, SNORD64, SNORD108, SNORD109A, SNORD109B, SNORD116-1, SNORD116-2, SNORD116-9, SNORD116-3, SNORD116-4, SNORD116-5, SNORD116-6, SNORD116-2, SNORD116-7, SNORD116-5, SNORD116-8, SNORD116-10, SNORD116-11, SNORD116-12, SNORD116-13, SNORD116-14, SNORD116-15, SNORD116-16, SNORD116-19, SNORD116-17, SNORD116-18, SNORD116-19, SNORD116-17, SNORD116-20, SNORD116-21, SNORD116-22, SNORD116-23, SNORD116-24, SNORD116-25, SNORD116-26, SNORD116-27, SNORD116-28, SNORD116-29, IPW, PAR1, SNORD115-1, SNORD115-2, SNORD115-3,*

							SNORD115-4, SNORD115-5, SNORD115-12, SNORD115-10, SNORD115-9, SNORD115-6, SNORD115-7, SNORD115-8, SNORD115-12, SNORD115-10, SNORD115-9, SNORD115-5, SNORD115-10, SNORD115-11, SNORD115-29, SNORD115-36, SNORD115-43, SNORD115-12, SNORD115-10, SNORD115-9, SNORD115-5, SNORD115-13, SNORD115-14, SNORD115-15, SNORD115-16, SNORD115-19, SNORD115-18, SNORD115-17, SNORD115-19, SNORD115-18, SNORD115-17, SNORD115-19, SNORD115-18, SNORD115-17, SNORD115-21, SNORD115-20, SNORD115-22, PAR4, SNORD115-23, HBII-52-24, SNORD115-25, SNORD115-26, HBII-52-27, HBII-52-28, SNORD115-11, SNORD115-29, SNORD115-36, SNORD115-30, SNORD115-31, SNORD115-32, SNORD115-33, SNORD115-34, SNORD115-35, SNORD115-11, SNORD115-29, SNORD115-37, SNORD115-38, SNORD115-39, SNORD115-40, SNORD115-41, SNORD115-42, SNORD115-10, SNORD115-15, SNORD115-11, SNORD115-36, SNORD115-29, SNORD115-43, SNORD115-44, HBII-52-45, HBII-52-46, SNORD115-48, SNORD109A, SNORD109B, UBE3A, ATP10A, GABRB3, GABRA5, GABRG3, OCA2, HERC2, GOLGA8F, GOLGA8G
12032.p1	chr3	672.3	723.2	50.98	587	Del	*SUCLG2, FAM19A1, FAM19A4, C3orf64, TMF1, UBA3, ARL6IP5, LMOD3, FRMD4B, MITF, FOXP1, EIF4E3, GPR27, PROK2*
12033.p1	chr4	1659.2	1663.6	4.41	34	Dup	*TRIM61, C4orf39, TRIM60, TRIM75, TMEM192, KLHL2*
12066.p1	chr11	218.1	267.2	49.1	387	Dup	*ANO5, SLC17A6, FANCF, GAS2, SVIP, LUZP2, ANO3, MUC15, SLC5A12*
12118.p1	chr10	1334.3	1343.4	9.12	56	Dup	*PPP2R2D, BNIP3, JAKMIP3, DPYSL4, STK32C, LRRC27, PWWP2B, C10orf91, INPP5A*
12132.p1	chr14	435.7	441.3	5.57	42	Dup	*FSCB*
12144.p1	chr7	1580.9	1584.2	3.35	35	Dup	*NCAPG2, FAM62B, WDR60*
12185.p1	chr7	1521.7	1532.9	11.18	231	Dup	*ACTR3B, DPP6*
12215.p1	chr3	1596.3	1602.8	6.49	48	Dup	*RSRC1, MLF1, GFM1, LXN, RARRES1, MFSD1, IQCJ*
12224.p1	chr19	204	207.8	3.77	19	Dup	*ZNF826, ZNF737, ZNF626*
12224.p1	chr22	387.8	391	3.17	46	Del	*TNRC6B, ADSL, SGSM3*
12232.p1	chr21	450.6	455.6	5.01	114	Dup	*PTTG1IP, ITGB2, C21orf67, C21orf70, NCRNA00162, C21orf122, ADARB1, POFUT2, LOC642852*
12235.p1	chr9	1299.2	1305.2	5.97	98	Dup	*PTGES2, LOC389791, LCN2, C9orf16, CIZ1, DNM1, GOLGA2, C9orf119, TRUB2, COQ4, SLC27A4, URM1, CERCAM, ODF2, GLE1, SPTAN1, WDR34, SET, PKN3*
12239.p1	chr22	174.1	180.6	6.49	945	Del	*DGCR2, DGCR11, DGCR14, TSSK2, GSC2, SLC25A1, CLTCL1, HIRA, MRPL40, C22orf39, UFD1L, CDC45L, CLDN5, LOC150185*
12242.p1	chr17	289.9	299.9	9.99	62	Dup	*ACCN1, CCL2, CCL7, CCL11, CCL8, CCL13, CCL1, C17orf102, TMEM132E*

12279.p1	chr3	600.6	603.9	3.23	24	Del	*FHIT*
12296.p1	chr6	577.7	622.5	44.83	63	Dup	*GUSBL2*
12316.p1	chr9	6.2	10.5	4.31	88	Dup	*KANK1, DMRT1, DMRT3, DMRT2*
12323.p1	chr12	1315.6	1318.3	2.74	19	Dup	*FBRSL1, P2RX2, POLE, PXMP2, PGAM5, ANKLE2*
12334.p1	chr6	1188	1191.8	3.75	58	Dup	*C6orf204, BRD7P3, PLN*
12341.p1	chr17	587.3	596.7	9.41	95	Dup	*TANC2, CYB561, CYB561, CYB561, ACE, ACE, KCNH6, KCNH6, WDR68, CCDC44, MAP3K3, LIMD2, STRADA, CCDC47, DDX42, FTSJ3, PSMC5, SMARCD2, TCAM1, CSH2, CSH2, GH2, GH2, GH2, CSH1, CSHL1, GH1, CD79B, SCN4A, C17orf72, ICAM2, ICAM2, ERN1, SNORD104, SNORA76, TEX2*
12376.p1	chr5	3.7	10	6.32	189	Dup	*AHRR, C5orf55, EXOC3, LOC25845, SLC9A3, CEP72, TPPP, ZDHHC11, BRD9, TRIP13*
12434.p1	chr13	1137.7	1141.2	3.49	63	Dup	*RASA3, CDC16, UPF3A, ZNF828*
12465.p1	chr6	10.2	15.6	5.39	52	Dup	*LOC285768, FOXQ1, FOXF2, FOXC1*
12594.p1	chr7	722.6	738.6	15.99	511	Dup	*NCF1B, GTF2IRD2P, NSUN5, TRIM50, FKBP6, FZD9, BAZ1B, BCL7B, TBL2, MLXIPL, VPS37D, DNAJC30, WBSCR22, STX1A, WBSCR26, ABHD11, CLDN3, CLDN4, WBSCR27, WBSCR28, ELN, LIMK1, EIF4H, LAT2, RFC2, CLIP2, GTF2IRD1, GTF2I, NCF1, GTF2IRD2*
12604.p1	chr10	160.2	166.2	5.92	42	Del	*PTER, C1QL3*
12604.p1	chr7	1429.2	1434.8	5.62	77	Dup	*LOC441294, FAM115C, CTAGE6, LOC154761, FAM115A, OR2F2, OR2F1, OR6B1, OR2A5, OR2A25, OR2A12, OR2A2, OR2A14*
12689.p1	chr5	970.8	982.7	11.97	91	Dup	*RGMB, CHD1*
12691.p1	chr16	688.4	715	26.52	678	Del	*AARS, DDX19B, DDX19A, ST3GAL2, FUK, COG4, SF3B3, SNORD111B, SNORD111, IL34, MTSS1L, VAC14, HYDIN, FTSJD1, CALB2, ZNF23, ZNF19, CHST4, TAT, MARVELD3, PHLPPL, AP1G1, SNORD71, ATXN1L, ZNF821, KIAA0174, PKD1L3, DHODH, HP, HPR, TXNL4B, DHX38, PMFBP1, ZFHX3*
12729.p1	chr2	452.7	458.2	5.56	133	Dup	*SRBD1, PRKCE*
12735.p1	chr2	1032.6	1087	54.38	1,229	Del	*LOC150568, POU3F3, MRPS9, GPR45, TGFBRAP1, C2orf49, FHL2, NCK2, C2orf40, UXS1, PLGLA, RGPD3, ST6GAL2, LOC729121, RGPD4, SLC5A7, SULT1C3, SULT1C2, SULT1C4, GCC2, LIMS1*
12750.p1	chr4	1519	1522.1	3.1	51	Del	*LRBA*
12763.p1	chr12	737	743.9	6.92	54	Dup	*KCNC2, CAPS2, GLIPR1L1, GLIPR1L2, GLIPR1, KRR1*
12766.p1	chr10	61.1	65	3.95	66	Dup	*IL2RA, RBM17, PFKFB3*
12775.p1	chr8	45.9	59.2	13.36	139	Del	*CSMD1*
12831.p1	chr7	699.4	702.4	2.98	61	Dup	*WBSCR17*

12860.p1	chr6	20	22.7	2.64	19	Dup	*GMDS*
12864.p1	chr6	1622.6	1627.1	4.55	94	Del	*PARK2*
12885.p1	chr5	1132.4	1140	7.62	60	Del	*KCNN2*
12892.p1	chr11	994.2	1001.3	7.14	49	Dup	*CNTN5, FLJ32810*
12916.p1	chr13	694.8	704.9	10.16	85	Dup	*KLHL1, ATXN8OS*
12940.p1	chr3	22.8	27.9	5.07	151	Dup	*CNTN4*
12991.p1	chr5	24.3	32.2	7.92	53	Dup	*IRX2, C5orf38*
12996.p1	chr6	1624.9	1627.7	2.82	56	Del	*PARK2*
13008.p1	chr12	1.8	6.5	4.7	105	Dup	*SLC6A12, SLC6A13, KDM5A, CCDC77, B4GALNT3, NINJ2*
13028.p1	chr4	1231.7	1234.2	2.52	18	Dup	*KIAA1109*
13038.p1	chr2	1193	1197.4	4.39	34	Dup	*EN1, MARCO, C1QL2, STEAP3*
13064.p1	chr15	964.6	969.9	5.3	32	Dup	*FAM169B*
13064.p1	chr3	1971.5	1988.8	17.31	2,387	Del	*SDHALP1, TFRC, ZDHHC19, OSTalpha, PCYT1A, TCTEX1D2, TM4SF19, UBXN7, RNF168, C3orf43, WDR53, FBXO45, LRRC33, C3orf34, PIGX, PAK2, SENP5, NCBP2, LOC152217, PIGZ, MFI2, DLG1, BDH1, LOC220729*
13085.p1	chr17	314.3	336.6	22.29	3,558	Del	*CCL3, CCL4, TBC1D3B, CCL3L1, CCL3L3, CCL4L1, CCL4L2, TBC1D3C, CCL3L1, CCL3L3, CCL4L2, CCL4L1, TBC1D3H, TBC1D3C, TBC1D3G, ZNHIT3, MYO19, MYO19, PIGW, GGNBP2, DHRS11, MRM1, LHX1, AATF, ACACA, C17orf78, TADA2L, DUSP14, AP1GBP1, DDX52, HNF1B, LOC284100, TBC1D3E, TBC1D3, TBC1D3F, TBC1D3E, TBC1D3D*
13085.p1	chr20	70.7	85.2	14.58	98	Dup	*HAO1, TMX4, PLCB1*
13094.p1	chr3	783	789.9	6.9	56	Dup	*ROBO1*
13106.p1	chr17	99.3	103.6	4.22	112	Dup	*GAS7, MYH13, MYH8, MYH4, MYH1*
13119.p1	chr22	460.7	495.6	34.8	4,710	Del	*FAM19A5, FAM19A5, C22orf34, BRD1, LOC90834, ZBED4, ALG12, CRELD2, PIM3, IL17REL, TTLL8, MLC1, MOV10L1, PANX2, TRABD, SELO, TUBGCP6, HDAC10, MAPK12, MAPK11, PLXNB2, FAM116B, SAPS2, SBF1, ADM2, MIOX, LMF2, NCAPH2, NCAPH2, SCO2, TYMP, TYMP, ODF3B, KLHDC7B, C22orf41, CHKB, CPT1BCHKB, LOC100144603, MAPK8IP2, ARSA, SHANK3, ACR, RPL23AP82, RABL2B*
13134.p1	chr3	64.6	68.8	4.28	36	Dup	*GRM7*
13137.p1	chr1	336.4	341.9	5.44	38	Dup	*ZSCAN20, CSMD2, HMGB4*
13150.p1	chr5	1303.1	1307.3	4.21	29	Dup	*HINT1, LYRM7, CDC42SE2*
13151.p1	chr6	1621.7	1625	3.28	68	Dup	*PARK2*

13180.p1	chr12	330.8	336.9	6.11	73	Dup	*SYT10*
13214.p1	chr10	815.8	819.6	3.85	534	Del	*MBL1P1, SFTPD, LOC219347, C10orf57, PLAC9, ANXA11*
13287.p1	chr13	185.4	188.4	3.02	22	Del	*DKFZp686A1627, TUBA3C, LOC100101938*
13303.p1	chr20	234.5	241.6	7.1	75	Dup	*LOC164380, CST9L, CST9, CST3, CST4, CST1, CST2, CST5, GGTLC1*
13327.p1	chr1	1194.7	1199.7	4.94	34	Dup	*WARS2, HAO2, HSD3B2, HSD3B1, ZNF697*
13328.p1	chr17	140.2	153.6	13.41	188	Dup	*COX10, CDRT15, HS3ST3B1, MGC12916, PMP22, TEKT3, CDRT4, FAM18B2*
13337.p1	chr2	452.7	458.3	5.63	135	Dup	*SRBD1, PRKCE*
13338.p1	chr7	1415.8	1420.7	4.86	50	Dup	*MOXD2, TRYX3*
13338.p1	chr8	550	553.7	3.75	30	Dup	*RGS20, TCEA1, LYPLA1, MRPL15*
13346.p1	chr1	933.2	963.3	30.11	222	Del	*MTF2, TMED5, CCDC18, DR1, FNBP1L, BCAR3, DNTTIP2, GCLM, ABCA4, ARHGAP29, ABCD3, F3, SLC44A3, CNN3, ALG14, TMEM56, RWDD3*
13346.p1	chr1	990.1	1008.3	18.27	138	Del	*PAP2D, LPPR4, PALMD, FRRS1, AGL, SLC35A3, HIAT1, SASS6, CCDC76, LRRC39, DBT, RTCD1, CDC14A, GPR88*
13346.p1	chr1	1123.7	1133.2	9.48	84	Del	*CTTNBP2NL, WNT2B, ST7L, CAPZA1, MOV10, RHOC, PPM1J, FAM19A3, SLC16A1, AFARP1*
13355.p1	chr15	245.2	251.3	6.12	95	Dup	*GABRB3, GABRA5, GABRG3*
13368.p1	chr7	815.6	832.3	16.72	159	Dup	*CACNA2D1, PCLO, PCLO, SEMA3E*
13375.p1	chr16	740.9	774.2	33.27	439	Dup	*CHST5, FLJ22167, GABARAPL2, ADAT1, KARS, TERF2IP, CNTNAP4, MON1B, LOC100130958, ADAMTS18, NUDT7, VAT1L, CLEC3A, WWOX*
13437.p1	chr5	501.3	503.9	2.6	55	Dup	*PARP8*
13451.p1	chr8	852.3	931.8	79.51	796	Del	*RALYL, LRRCC1, E2F5, E2F5, C8orf59, CA13, CA1, CA3, CA2, REXO1L2P, REXO1L1, PSKH2, ATP6V0D2, SLC7A13, WWP1, FAM82B, CPNE3, CNGB3, CNBD1, WDR21C, MMP16, RIPK2, OSGIN2, NBN, DECR1, CALB1, TMEM64, NECAB1, TMEM55A, OTUD6B, LRRC69, SLC26A7, RUNX1T1*
13491.p1	chr17	140.2	154.3	14.12	199	Dup	*COX10, CDRT15, HS3ST3B1, MGC12916, PMP22, TEKT3, CDRT4, FAM18B2*
13499.p1	chr5	1791.9	1794.4	2.54	20	Dup	*SQSTM1, C5orf45, TBC1D9B, RNF130*
13503.p1	chr8	31.1	39	7.9	164	Del	*CSMD1*
13508.p1	chr18	729.4	737.7	8.38	50	Dup	*MBP, GALR1*
13518.p1	chr18	728.7	731.7	2.92	30	Dup	*MBP, GALR1*
13521.p1	chr6	576.4	626.8	50.4	129	Dup	*GUSBL2, KHDRBS2*
13521.p1	chr6	626.9	629.8	2.81	24	Dup	*KHDRBS2*
13554.p1	chr2	1871.6	1881.5	9.89	124	Dup	*ITGAV, FAM171B, ZSWIM2, CALCRL, TFPI*
13555.p1	chr14	344.3	347.5	3.17	44	Dup	*C14orf19, SRP54, FAM177A1, PPP2R3C, KIAA0391*

13555.p1	chr17	140.4	154	13.56	193	Del	*COX10, CDRT15, HS3ST3B1, MGC12916, PMP22, TEKT3, CDRT4, FAM18B2*
13568.p1	chr1	1186.2	1196.5	10.34	79	Del	*TBX15, WARS2*
13568.p1	chr7	1447.8	1456.1	8.3	94	Dup	*CNTNAP2*
13585.p1	chr15	270	281.6	11.64	1,717	Del	*APBA2, KIAA0574, NDNL2, TJP1*
13587.p1	chr1	186.9	189.9	3.04	18	Dup	*PAX7*
13590.p1	chr13	321.9	339.6	17.73	139	Dup	*PDS5B, KL, STARD13, RFC3*
13601.p1	chr2	1281.4	1290.8	9.45	64	Dup	*LIMS2, SFT2D3, WDR33, POLR2D, AMMECR1L, SAP130, UGCGL1, HS6ST1*
13603.p1	chr4	95.3	100.7	5.42	77	Dup	*SLC2A9, WDR1, ZNF518B*
13616.p1	chr11	173.7	177.4	3.72	55	Dup	*ABCC8, USH1C, MYOD1, KCNC1*
13616.p1	chr7	977.1	979.9	2.78	18	Dup	*TECPR1, BRI3, BAIAP2L1*
13624.p1	chr20	145.8	149.9	4.13	59	Del	*MACROD2*
13641.p1	chr19	620.8	623.5	2.78	26	Dup	*USP29, ZIM3*
13647.p1	chr15	286.6	302.3	15.76	2,445	Del	*FAM7A1, FAM7A2, ARHGAP11B, MTMR15, MTMR10, TRPM1, KLF13, OTUD7A, CHRNA7*
13676.p1	chr1	1737	1740.3	3.28	48	Dup	*TNR*
13688.p1	chr8	133.9	136.5	2.61	21	Dup	*DLC1, C8orf48*
13691.p1	chr16	134.6	146.9	12.27	267	Dup	*ERCC4, MKL2, PARN, BFAR, PLA2G10*
13696.p1	chr6	654.8	660	5.24	43	Dup	*EYS*
13697.p1	chr3	1754.2	1759.9	5.75	45	Dup	*NLGN1*
13700.p1	chr8	1.7	69.1	67.47	1,103	Del	*RPL23AP53, ZNF596, FBXO25, C8orf42, ERICH1, DLGAP2, CLN8, ARHGEF10, KBTBD11, MYOM2, CSMD1, MCPH1, ANGPT2, AGPAT5, XKR5, DEFB1, DEFA6, DEFA4, DEFA1, LOC728358, DEFA1, LOC728358, DEFA3, DEFA5*
13700.p1	chr8	126.1	140.7	14.59	168	Dup	*LONRF1, LOC340357, C8orf79, DLC1, C8orf48, SGCZ*
13716.p1	chr7	249.1	253.3	4.17	33	Dup	*OSBPL3, CYCS, C7orf31, NPVF*
13716.p1	chr8	3.9	7	3.15	78	Dup	*FBXO25, C8orf42, ERICH1*
13722.p1	chr3	1621.5	1626.5	5.04	85	Del	*PPM1L, B3GALNT1, NMD3, C3orf57*
13724.p1	chr9	1032.4	1056.1	23.65	213	Dup	*C9orf125, RNF20, GRIN3A, PPP3R2, CYLC2*
13726.p1	chr11	566	599.9	33.91	316	Del	*LRRC55, APLNR, TNKS1BP1, SSRP1, P2RX3, PRG3, PRG2, SLC43A3, RTN4RL2, SLC43A1, TIMM10, SMTNL1, UBE2L6, SERPING1, YPEL4, CLP1, ZDHHC5, MED19, TMX2, C11orf31, LOC643376, CTNND1, OR9Q1, OR9I1, OR9Q2, OR1S2, OR1S1, OR10Q1, OR10W1, OR5B17, OR5B3, OR5B2, OR5B12, OR5B21, LPXN, ZFP91, ZFP91-CNTF, CNTF, GLYAT, GLYATL2, GLYATL1, FAM111B,*

							FAM111A, DTX4, MPEG1, OR5AN1, OR5A2, OR5A1, OR4D6, OR4D10, OR4D11, OR4D9, OSBP, PATL1, OR10V1, STX3, MRPL16, GIF, TCN1, PLAC1L, MS4A3, MS4A2, MS4A6A, MS4A4A, MS4A6E, MS4A7, MS4A14, MS4A5, MS4A1
13739.p1	chr3	22.8	27.9	5.07	151	Dup	*CNTN4*
13757.p1	chr6	1187.5	1191.2	3.68	54	Dup	*C6orf204, BRD7P3, PLN*
13759.p1	chr17	781.8	785.2	3.45	29	Dup	*WDR45L, RAB40B, FN3KRP, FN3K, TBCD, ZNF750, B3GNTL1*
13760.p1	chr2	452.7	458.3	5.63	135	Dup	*SRBD1, PRKCE*
13783.p1	chr3	10.7	14.5	3.78	33	Dup	*CNTN6*
13786.p1	chr9	258.9	269.1	10.17	80	Dup	*C9orf82, PLAA*
13815.p1	chr16	748.1	750.7	2.59	24	Del	*CNTNAP4, CNTNAP4*
13817.p1	chr20	80.4	85.2	4.8	54	Del	*PLCB1*
13830.p1	chr3	607.7	664	56.34	461	Del	*FHIT, PTPRG, ID2B, C3orf14, FEZF2, CADPS, LOC285401, SYNPR, S100A1L, C3orf49, THOC7, ATXN7, PSMD6, PRICKLE2, ADAMTS9, MAGI1, SLC25A26*
13845.p1	chr3	18.8	25.6	6.79	153	Dup	*CNTN4*
13871.p1	chr4	565.4	569.3	3.97	33	Del	*CEP135, KIAA1211, AASDH*
13876.p1	chr8	358.8	479.8	121.07	856	Dup	*KCNU1, FKSG2, ZNF703, ERLIN2, LOC728024, PROSC, GPR124, BRF2, RAB11FIP1, GOT1L1, ADRB3, EIF4EBP1, ASH2L, ASH2L, STAR, LSM1, BAG4, DDHD2, PPAPDC1B, WHSC1L1, LETM2, FGFR1, C8orf86, RNF5P1, TACC1, PLEKHA2, HTRA4, TM2D2, ADAM9, ADAM32, ADAM5P, ADAM3A, ADAM18, ADAM2, IDO1, IDO2, C8orf4, ZMAT4, SFRP1, GOLGA7, GINS4, AGPAT6, NKX6-3, ANK1, MYST3, AP3M2, PLAT, IKBKB, POLB, DKK4, VDAC3, SLC20A2, C8orf40, CHRNB3, CHRNA6, THAP1, RNF170, HOOK3, FNTA, SGK196, HGSNAT, POTEA, BEYLA, BEYLA*
13876.p1	chr8	479.8	506	26.18	267	Dup	*KIAA0146, CEBPD, PRKDC, MCM4, UBE2V2, EFCAB1, SNAI2, C8orf22*
13877.p1	chr3	779.3	844.9	65.6	534	Del	*ROBO1, GBE1*
13887.p1	chr10	348.5	352.1	3.61	33	Dup	*PARD3*
13907.p1	chr7	466.4	474.9	8.46	182	Dup	*TNS3*
13915.p1	chr20	181.4	184.2	2.79	15	Dup	*ZNF133, C20orf12, POLR3F, RBBP9*
13926.p1	chr11	312.5	315	2.51	24	Del	*DCDC1, DNAJC24, IMMP1L, ELP4*
13935.p1	chr8	134	146.9	12.87	107	Dup	*DLC1, C8orf48, SGCZ*
13944.p1	chr3	894.6	903.9	9.27	102	Dup	*EPHA3*
13945.p1	chr12	425.3	428.9	3.58	27	Dup	*TMEM117*
13948.p1	chr11	331.4	334.5	3.07	29	Dup	*CSTF3, HIPK3*

13949.p1	chr3	20.1	23.6	3.49	60	Dup	*CNTN4*
13951.p1	chr11	42.2	44.9	2.69	31	Del	*OR52B4, TRIM21, OR52K2*
13962.p1	chr2	506.5	511.1	4.59	95	Del	*NRXN1*
13982.p1	chr5	993.3	1007.1	13.82	115	Dup	*LOC100133050, FAM174A, ST8SIA4*
13987.p1	chr13	1097.5	1100.4	2.91	20	Dup	*COL4A1, COL4A2, RAB20*
13988.p1	chr3	0.8	3.9	3.14	63	Dup	*CHL1*
13988.p1	chr6	1003.9	1007.5	3.64	29	Dup	*MCHR2*
13999.p1	chr18	22.7	27.6	4.95	47	Dup	*METTL4, NDC80, SMCHD1*
13999.p1	chr2	2388	2393.3	5.3	60	Del	*LOC151174, LOC643387, HES6, PER2, TRAF3IP1, ASB1*
14002.p1	chr20	494.7	498	3.37	15	Dup	*NFATC2, ATP9A*
14016.p1	chr6	14.4	16.9	2.54	36	Dup	*FOXC1, GMDS*
14018.p1	chr3	22.6	27.6	5.03	150	Dup	*CNTN4*
14019.p1	chr7	1576.3	1579.6	3.28	23	Dup	*PTPRN2*
14032.p1	chr7	749.9	760.8	10.91	283	Dup	*PMS2L3, HIP1, CCL26, CCL24, RHBDD2, POR, SNORA14A, TMEM120A, STYXL1, MDH2, FLJ37078, HSPB1, YWHAG, SRCRB4D, ZP3, DTX2, FDPSL2A, DTX2, UPK3B, POMZP3*
14044.p1	chr13	189.9	194.7	4.79	116	Dup	*TPTE2, MPHOSPH8, PSPC1, ZMYM5, ZMYM2*
14046.p1	chr11	1037.6	1046.1	8.51	154	Dup	*CASP12, CASP4, CASP5, CASP1, CARD16, CARD17, CARD18*
14053.p1	chr10	787.6	793.1	5.45	36	Dup	*KCNMA1, KCNMA1, DLG5*
14066.p1	chr1	1737	1740.3	3.28	48	Dup	*TNR*
14073.p1	chr12	647.9	657.5	9.65	133	Dup	*LLPH, TMBIM4, IRAK3, HELB, GRIP1*
14073.p1	chr15	970	975.4	5.48	81	Del	*IGF1R, LOC145814, SYNM, TTC23*
14074.p1	chr15	270	281.6	11.61	1,713	Del	*APBA2, KIAA0574, NDNL2, TJP1*
14084.p1	chr17	709.5	720.6	11.15	74	Del	*KIAA0195, CASKIN2, TSEN54, LLGL2, MYO15B, RECQL5, LOC643008, RECQL5, SAP30BP, ITGB4GALK1, H3F3B, UNK, UNC13D, WBP2, TRIM47, TRIM65, MRPL38, FBF1, ACOX1, LOC100134934, CDK3, EVPL, SRP68, GALR2, ZACN, EXOC7, FOXJ1, RNF157, FAM100B, QRICH2, PRPSAP1, SPHK1, UBE2O, AANAT, RHBDF2, CYGB, PRCD*
14091.p1	chr10	1281.3	1352.3	70.98	764	Del	*C10orf90, DOCK1, C10orf141, NPS, FOXI2, CLRN3, PTPRE, MKI67, MGMT, EBF3, GLRX3, TCERG1L, PPP2R2D, BNIP3, JAKMIP3, DPYSL4, STK32C, LRRC27, PWWP2B, C10orf91, INPP5A, NKX6-2, C10orf92, C10orf93, GPR123, KNDC1, UTF1, VENTX, ADAM8, TUBGCP2, ZNF511, CALY, PRAP1, C10orf125, ECHS1, PAOX, MTG1, SPRN, LOC619207, CYP2E1, SYCE1*

14091.p1	chr11	1304.1	1344.5	40.45	307	Dup	*NTM, OPCML, OPCML, SPATA19, LOC283174, IGSF9B, LOC100128239, JAM3, NCAPD3, VPS26B, THYN1, ACAD8, GLB1L3, GLB1L2, B3GAT1*
14098.p1	chr5	8.7	11.5	2.79	59	Dup	*ZDHHC11, BRD9, TRIP13, NKD2, SLC12A7*
14112.p1	chr11	853.5	862.6	9.16	84	Dup	*PICALM, EED, C11orf73, C11orf73, CCDC81, ME3, PRSS23*
14152.p1	chr8	30.2	34.8	4.66	41	Dup	*CSMD1*
14152.p1	chr8	47.6	51.1	3.47	54	Dup	*CSMD1*
14155.p1	chr1	1737	1740.3	3.28	48	Dup	*TNR*
14159.p1	chr15	190.7	261.2	70.53	2,693	Dup	*LOC646214, CXADRP2, POTEB, LOC727924, OR4M2, OR4N4, LOC650137, GOLGA8D, LOC283767, TUBGCP5, CYFIP1, NIPA2, NIPA1, WHAMML1, GOLGA9P, HERC2P2, GOLGA8E, MKRN3, MAGEL2, NDN, PWRN2, PWRN1, C15orf2, SNRPN, SNRPN, SNURF, SNORD107, PAR-SN, PAR5, SNORD64, SNORD108, SNORD109A, SNORD109B, SNORD116-1, SNORD116-2, SNORD116-9, SNORD116-3, SNORD116-4, SNORD116-7, SNORD116-5, SNORD116-6, SNORD116-2, SNORD116-7, SNORD116-5, SNORD116-8, SNORD116-9, SNORD116-3, SNORD116-10, SNORD116-11, SNORD116-12, SNORD116-13, SNORD116-14, SNORD116-15, SNORD116-16, SNORD116-19, SNORD116-17, SNORD116-18, SNORD116-19, SNORD116-17, SNORD116-20, SNORD116-21, SNORD116-22, SNORD116-23, SNORD116-24, SNORD116-25, SNORD116-26, SNORD116-27, SNORD116-28, SNORD116-29, IPW, PAR1, SNORD115-1, SNORD115-2, SNORD115-3, SNORD115-4, SNORD115-5, SNORD115-12, SNORD115-10, SNORD115-9, SNORD115-6, SNORD115-7, SNORD115-8, SNORD115-12, SNORD115-10, SNORD115-9, SNORD115-5, SNORD115-10, SNORD115-11, SNORD115-29, SNORD115-36, SNORD115-43, SNORD115-12, SNORD115-10, SNORD115-9, SNORD115-5, SNORD115-13, SNORD115-14, SNORD115-15, SNORD115-16, SNORD115-19, SNORD115-18, SNORD115-17, SNORD115-19, SNORD115-18, SNORD115-17, SNORD115-19, SNORD115-18, SNORD115-17, SNORD115-21, SNORD115-20, SNORD115-22, PAR4, SNORD115-23, HBII-52-24, SNORD115-25, SNORD115-26, HBII-52-27, HBII-52-28, SNORD115-11, SNORD115-29, SNORD115-36, SNORD115-43, SNORD115-30, SNORD115-31, SNORD115-32, SNORD115-33, SNORD115-34, SNORD115-35, SNORD115-11, SNORD115-29, SNORD115-36, SNORD115-43, SNORD115-37, SNORD115-38, SNORD115-39, SNORD115-40, SNORD115-41, SNORD115-42, SNORD115-10, SNORD115-15, SNORD115-11, SNORD115-36, SNORD115-29, SNORD115-43, SNORD115-44, HBII-52-45, HBII-52-46, SNORD115-48, SNORD109A, SNORD109B, UBE3A, UBE3A, UBE3A, ATP10A, GABRB3, GABRB3, GABRA5, GABRG3, OCA2, HERC2*
14161.p1	chr17	99.3	103.6	4.22	111	Dup	*GAS7, MYH13, MYH8, MYH4, MYH1*
14161.p1	chr7	1575.6	1580.8	5.18	46	Dup	*PTPRN2*

14166.p1	chr3	818.7	833.9	15.13	126	Del	*GBE1*
14174.p1	chr5	456.9	461.5	4.64	54	Dup	*HCN1*
14175.p1	chr1	1737	1740.3	3.28	48	Dup	*TNR*
14176.p1	chr6	1188.2	1191.4	3.13	51	Dup	*C6orf204, BRD7P3, PLN*
14195.p1	chr7	81.5	85	3.46	66	Dup	*ICA1, NXPH1*
14196.p1	chr18	129.1	131.9	2.78	14	Dup	*SEH1L, CEP192*
14204.p1	chr3	965.7	1009	43.26	372	Dup	*EPHA6, ARL6, CRYBG3, MINA, GABRR3, OR5AC2, OR5H1, OR5H14, OR5H15, OR5H6, OR5H2, OR5K4, OR5K3, OR5K1, OR5K2, CLDND1, GPR15, CPOX, ST3GAL6, DCBLD2, COL8A1*
14226.p1	chr11	749.7	755.2	5.49	41	Dup	*MAP6, MOGAT2, DGAT2, UVRAG*
14232.p1	chr22	465.7	495.7	30.03	4,084	Del	*FAM19A5, C22orf34, BRD1, LOC90834, ZBED4, ALG12, CRELD2, PIM3, IL17REL, TTLL8, MLC1, MOV10L1, PANX2, TRABD, SELO, TUBGCP6, HDAC10, MAPK12, MAPK11, PLXNB2, FAM116B, SAPS2, SBF1, ADM2, MIOX, LMF2, NCAPH2, SCO2, TYMP, ODF3B, KLHDC7B, C22orf41, CHKB, CPT1B, LOC100144603, MAPK8IP2, ARSA, SHANK3, ACR, RPL23AP82, RABL2B, RPL23AP82*
14237.p1	chr7	291.8	295.1	3.27	35	Dup	*CHN2*
14238.p1	chr18	92.8	95.4	2.61	22	Del	*TWSG1, RALBP1, PPP4R1*
14240.p1	chr1	2395.6	2399	3.4	26	Dup	*RGS7, FH, KMO, OPN3, CHML*
14249.p1	chr20	80.4	85.2	4.8	54	Del	*PLCB1*
14250.p1	chr2	1062.6	1073.8	11.25	259	Dup	*PLGLA, RGPD3, ST6GAL2*
14252.p1	chr15	829.4	835.3	5.85	897	Dup	*ZSCAN2, SCAND2, WDR73, NMB, SEC11A, ZNF592, ALPK3, SLC28A1, PDE8A*
14254.p1	chr9	710.4	759.1	48.61	481	Del	*TJP2, C9orf61, APBA1, PTAR1, C9orf135, MAMDC2, SMC5, KLF9, TRPM3, TMEM2, FAM108B1, C9orf85, HSPBL2, C9orf57, GDA, ZFAND5, TMC1, ALDH1A1, ANXA1*
14266.p1	chr13	224.3	238.6	14.27	312	Del	*SGCG, SACS, TNFRSF19, MIPEP, PCOTH, RP11-45B20.2, SPATA13, C1QTNF9*
14267.p1	chr22	213.3	219.9	6.57	153	Dup	*RTDR1, GNAZ, RAB36, BCR*
14270.p1	chr11	459.2	470.6	11.39	148	Del	*PHF21A, CREB3L1, DGKZ, MDK, CHRM4, AMBRA1, HARBI1, KIAA0652, ARHGAP1, ZNF408, F2, CKAP5, SNORD67, LRP4, C11orf49*
14274.p1	chr3	413.1	416	2.86	23	Del	*ULK4*
14286.p1	chr2	2424.1	2427	2.94	60	Dup	*NEU4, PDCD1, C2orf85, LOC728323*
14293.p1	chr18	638.5	646.8	8.36	68	Dup	*TMX3, CCDC102B, CCDC102B*
14297.p1	chr1	1829.7	1833.9	4.24	30	Dup	*EDEM3, FAM129A, RNF2, C1orf25*

14349.p1	chr9	1.9	16.6	14.62	229	Dup	*C9orf66, DOCK8, KANK1 DMRT1, DMRT3, DMRT2*
14365.p1	chr1	2041.5	2044.9	3.45	15	Del	*SLC26A9, FAM72A, AVPR1B, C1orf186, CTSE*
14376.p1	chr17	100.1	105.3	5.23	138	Dup	*GAS7, MYH13, MYH8, MYH4, MYH1, MYH2, MYH3, SCO1*
14376.p1	chr9	701.7	778	76.33	785	Del	*PGM5, C9orf71, PIP5K1B, FAM122A, PRKACG, FXN, TJP2, C9orf61, APBA1, PTAR1, C9orf135, MAMDC2, SMC5, KLF9, TRPM3, TMEM2, FAM108B1, C9orf85, HSPBL2, C9orf57, GDA, ZFAND5, TMC1, ALDH1A1, ANXA1, RORB, TRPM6, C9orf40, C9orf41, C9orf95, OSTF1, PCSK5*
14415.p1	chr16	762.5	769.7	7.16	115	Dup	*NUDT7, NUDT7, VAT1L, CLEC3A, WWOX*
14438.p1	chr2	1902.1	1907.3	5.27	41	Del	*ASNSD1, ANKAR, OSGEPL1, ORMDL1, PMS1, MSTN, C2orf88*
14438.p1	chr5	654.8	663.1	8.28	61	Dup	*SFRS12, MAST4*
14440.p1	chr22	362.5	367.1	4.66	81	Dup	*CDC42EP1, LGALS2, GGA1, SH3BP1, PDXP, LGALS1, NOL12, TRIOBP, H1F0, GCAT, GALR3, ANKRD54, EIF3L, MICALL1, C22orf23, POLR2F, SOX10*
14442.p1	chr6	58.4	61.1	2.72	18	Dup	*NRN1, F13A1*
14443.p1	chr15	973	978.1	5.15	81	Dup	*IGF1R, LOC145814, SYNM, TTC23, LRRC28*
14452.p1	chr13	197	200	3.04	67	Del	*GJB6, CRYL1*
14473.p1	chr6	1254.8	1257.5	2.69	67	Del	*TPD52L1, HDDC2*
14478.p1	chr5	1374.9	1381.6	6.69	105	Dup	*NME5, BRD8, KIF20A, CDC23, GFRA3, CDC25C, FAM53C, KDM3B, REEP2, EGR1, ETF1, HSPA9, SNORD63, CTNNA1*
14487.p1	chr10	679.9	682.8	2.91	24	Del	*CTNNA3*
14491.p1	chr17	140.2	153.8	13.62	194	Del	*COX10, CDRT15, HS3ST3B1, MGC12916, PMP22, TEKT3, CDRT4, FAM18B2*
AU1813301	chr1	59.5	64.9	5.4	36	Dup	*NPHP4, KCNAB2, CHD5, RPL22, RNF207, ICMT, HES3, GPR153, ACOT7, HES2, ESPN, TNFRSF25, TNFRSF25, PLEKHG5*
AU1813301	chr12	1233.6	1236.7	3.2	79	Dup	*FAM101A, NCOR2*
AU1813301	chr16	161.3	187	25.7	2,069	Dup	*ABCC1, ABCC6, NOMO3, LOC339047, XYLT1, LOC339047, NOMO2, ABCC6P1*
AU062003	chr6	626.9	629.9	2.91	25	Dup	*KHDRBS2*
AU083204	chr15	998.5	1001.3	2.78	52	Dup	*TM2D3, TARSL2*
AU074403	chr13	197	200	3.04	67	Del	*GJB6, CRYL1*
AU079605	chr16	807.8	812.9	5.09	34	Dup	*CDH13*
AU028905	chr17	99.3	103.6	4.22	112	Dup	*GAS7, MYH13, MYH8, MYH4, MYH1*
AU026604	chr14	804.7	870.7	65.95	518	Dup	*C14orf145, TSHR, GTF2A1, SNORA79, STON2, SEL1L, FLRT2*
AU3019302	chr19	496	503.7	7.66	136	Del	*ZNF229, ZNF180, CEACAM20, PVR, PVR, CEACAM19, CEACAM16, BCL3, CBLC, BCAM, BCAM, PVRL2, PVRL2, TOMM40, APOE, APOC1, APOC4, APOC2, CLPTM1, RELB, SFRS16, ZNF296, GEMIN7, GEMIN7, NKPD1, NKPD1,*

							TRAPPC6A
AU1172301	chr6	1445.8	1448.4	2.59	23	Dup	*UTRN*
AU0881301	chr3	1375.4	1379.3	3.85	38	Dup	*STAG1*
AU072203	chr16	344.6	349.9	5.33	58	Dup	*LOC283914, LOC146481*
AU1039302	chr4	739.7	742.8	3.1	53	Dup	*COX18, ANKRD17*
AU0915301	chr19	234.2	238.5	4.37	22	Del	*ZNF675, ZNF681, RPSAP58*
AU0979301	chr7	25.9	30.3	4.45	35	Dup	*IQCE, TTYH3, AMZ1, GNA12, CARD11*
AU1286301	chr4	935	946.3	11.33	131	Dup	*GRID2*
AU1181301	chr4	1585.4	1595.7	10.26	79	Dup	*LOC340017, C4orf18, TMEM144*
AU0981301	chr10	949.7	953.5	3.78	26	Dup	*MYOF, CEP55GPR120, RBP4*
AU1361302	chr20	293	296.3	3.34	52	Dup	*DEFB115, DEFB116, DEFB118, DEFB119, DEFB119, DEFB121, DEFB122, DEFB123, DEFB124, REM1, NCRNA00028, HM13, PSIMCT-1*
AU044004	chr7	170.3	174.1	3.8	44	Dup	*AHR*
AU044004	chr9	1398.9	1402.2	3.23	68	Dup	*CACNA1B, LOC643224*
AU029303	chr5	601.4	608.4	6.99	54	Dup	*ELOVL7, ERCC8, NDUFAF2, C5orf43, ZSWIM6*
AU008003	chr5	373.1	375.7	2.66	24	Dup	*NUP155, NUP155, WDR70*
AU1765302	chr3	10.6	23.6	12.96	127	Dup	*CNTN6, CNTN4*
AU083503	chr3	1290.2	1296.6	6.36	136	Dup	*MGLL, KLHDC6, SEC61A1, RUVBL1, EEFSEC*
AU020604	chr4	1887.1	1898.1	11.02	82	Dup	*ZFP42, TRIML2, TRIML1*
AU064104	chr2	1214	1222.8	8.76	78	Dup	*GLI2, TFCP2L1, CLASP1, RNU4ATAC, MKI67IP, TSN*
AU2050301	chr20	181.4	184.2	2.79	15	Dup	*ZNF133, C20orf12, POLR3F, RBBP9*
AU1551302	chr16	688.5	700	11.48	336	Dup	*AARS, DDX19B, DDX19A, ST3GAL2, FUK, COG4, SF3B3, SNORD111B, SNORD111, IL34, MTSS1L, VAC14, HYDIN, FTSJD1, CALB2*
AU1492301	chr5	191.6	197.8	6.17	54	Dup	*CDH18*
AU1534301	chr5	783.8	788	4.21	59	Dup	*DMGDH, BHMT2, BHMT, JMY, HOMER1*
AU1379301	chr8	1309.3	1312	2.67	52	Dup	*FAM49B, ASAP1*
AU1203303	chr5	66.3	72.2	5.88	51	Dup	*LOC255167, NSUN2, SRD5A1, POLS*
AU1482302	chr12	1311.5	1320.3	8.78	64	Del	*EP400NL, DDX51, NOC4L, GALNT9LOC100130238, FBRSL1, P2RX2, POLE, PXMP2, PGAM5, ANKLE2, GOLGA3, CHFR, ZNF605*
AU1495303	chr2	508.8	512.6	3.74	57	Del	*NRXN1*
AU1300302	chr9	97.4	102.9	5.47	108	Del	*PTPRD*

AU2557302	chr12	1006.2	1011.4	5.17	43	Dup	*CHPT1, SYCP3, GNPTAB, DRAM, CCDC53, NUP37, C12orf48, PMCH*
AU2482301	chr2	2165.6	2168.7	3.12	23	Dup	*MREG, PECR, TMEM169, XRCC5, MARCH4*
AU2908301	chr4	1231.7	1234.4	2.7	19	Dup	*KIAA1109*
AU1536304	chr8	134	146.9	12.87	107	Dup	*DLC1, C8orf48, SGCZ*
AU005303	chr3	35.7	38.6	2.88	47	Dup	*LRRN1*
AU063004	chr16	100	104.3	4.29	32	Dup	*GRIN2A*
AU2581302	chr4	482.8	487.8	4.94	64	Dup	*FRYL, OCIAD1, OCIAD2, FLJ21511*
AU020103	chr3	12.9	15.6	2.72	20	Dup	*CNTN6*
AU1631301	chr7	178.2	183.3	5.04	41	Dup	*SNX13, PRPS1L1*
AU1887301	chr20	80.4	85.2	4.8	54	Dup	*PLCB1*
AU1744301	chr13	190.3	195	4.76	117	Dup	*MPHOSPH8, PSPC1, ZMYM5, ZMYM2*
AU0884301	chr3	1239	1242.3	3.24	25	Dup	*PARP14, HSPBAP1, DIRC2, LOC100129550, SEMA5B*
AU0962303	chr4	1650.3	1656.9	6.62	104	Dup	*ANP32C*
AU1448301	chr19	496	503.7	7.66	136	Del	*ZNF229, ZNF180, CEACAM20, PVR, PVR, CEACAM19, CEACAM16, BCL3, CBLC, BCAM, PVRL2, TOMM40, APOE, APOC1, APOC4, APOC2, CLPTM1, RELB, SFRS16, ZNF296, GEMIN7, NKPD1, NKPD1, TRAPPC6A*
AU2372301	chr9	65.6	70.1	4.47	107	Dup	*GLDC, KDM4C*
AU1081301	chr12	898.3	901.9	3.59	30	Del	*C12orf12, EPYC, KERA, LUM, DCN*
AU1081301	chr7	81.6	84.6	3.02	58	Dup	*ICA1, NXPH1*
AU2182302	chr10	426.8	429.4	2.66	43	Del	*RET*
AU2554301	chr3	584.6	601.5	16.86	266	Dup	*ACOX2, FAM107A, FAM3D, C3orf67, FHIT*
AU2689306	chr20	80.4	85.2	4.8	54	Dup	*PLCB1*
AU2814301	chr5	373.3	376.2	2.93	27	Dup	*NUP155, NUP155, WDR70*
AU021005	chr10	426.3	429.4	3.1	45	Del	*BMS1, RET*
AU082505	chr3	21.9	24.5	2.6	54	Dup	*CNTN4*
AU1977303	chr18	638.5	646.8	8.36	68	Dup	*TMX3, CCDC102B*
AU1802301	chr15	287	302.3	15.31	2,334	Del	*ARHGAP11B, MTMR15, MTMR15, MTMR10, TRPM1, KLF13, OTUD7A, CHRNA7*
AU1230301	chr11	330.7	334.3	3.51	33	Dup	*CSTF3, HIPK3*
AU002804	chr14	698.9	701.9	3.04	22	Dup	*COX16, SYNJ2BP, ADAM21, ADAM20, MED6, TTC9*
13521.p1	chr6	576.4	629.8	53.32		Dup	*GUSBL2, KHDRBS2*

12430.p1	chr6	575.9	625	49.1		Dup	*PRIM2, GUSBL2, KHDRBS2*
12691.p1	chr16	688.6	729	40.44		Del	*HYDIN*
AU004104	chr22	207.2	233.3	26.13		Dup	*MANY GENES*
13727.p1	chr9	934.6	958.9	24.34		Dup	*RORS, SPTLC1, IARS, NOL8, CENPP, OGN, OMD, ASPN, ECM2, IPPK, BICD2, ZNF484, FGD3, SUSD3, C9orf89, NINJ1, WNK2, C9orf129, FAM120AOS, FAM120A, PHF2, BARX1, PTPDC1*
12162.p1	chr1	2281.9	2302	20.14		Del	*GALNT2, PGBD5, COG2, AGT, CAPN9, CAPN9, C1orf198, C1orf198, C1orf198, TTC13, ARV1, FAM89A, TRIM67, C1orf131, GNPAT, EXOC8, C1orf124, EGLN1, TSNAX, DISC1, DISC1, DISC1, DISC1, DISC2*
12053.p1	chr7	1144.5	1161.4	16.91		Dup	*TFEC, TES, CAV2, CAV1, MET*
11398.p1	chr7	111.7	124.4	12.72		Dup	*PHF14, THSD7A, TMEM106B, VWDE*
AU1551302	chr16	688.3	700.1	11.74		Dup	*HYDIN*
13804.p1	chr22	221.6	233.3	11.71		Dup	*IGLL1, C22orf43, LOC91316, RGL4, ZNF70, VPREB3, C22orf15, CHCHD10, MMP11, SMARCB1, DERL3, SLC2A11, SLC2A11, MIF, GSTT2B, GSTT2, DDTL, DDT, DDT, GSTT2, GSTTP1, LOC391322, GSTT1, GSTTP2, CABIN1, SUSD2, GGT5, POM121L9P, CYTSA, ADORA2A, UPB1, C22orf13, SNRPD3, GGT1, C22orf36, GGT1*
13769.p1	chr16	688.8	698.1	9.25		Dup	*HYDIN*
11013.p1	chr20	75.1	82.7	7.57		Dup	*HAO1, TMX4, PLCB1*
14363.p1	chr9	43.1	50.2	7.03		Dup	*SLC1A1, C9orf68, PPAPDC2, CDC37L1, AK3, RCL1, JAK2*
11551.p1	chr16	87.1	92.5	5.45		Dup	*ABAT, C16orf72, CARHSP1, PMM2, TMEM186, USP7*
14221.p1	chr7	1106	1110.3	4.3		Del	*IMMP2L*
AU1496302	chr9	1.6	5.5	3.85		Dup	*CBWD1, C9orf66, DOCK8, KANK1*
11710.p1	chr5	138.6	142.3	3.77		Dup	*DNAH5, TRIO*
AU079605	chr6	1626.2	1629.8	3.55		Dup	*PARK2*
11811.p1	chr2	2127.7	2131.2	3.52		Del	*ERBB4*
AU1453302	chr7	1107.6	1111	3.48		Del	*IMMP2L*
AU003705	chr7	81.5	85	3.46		Dup	*ICA1, NXPH1*
11479.p1	chr7	1107.9	1110.9	2.99		Del	*IMMP2L*
14326.p1	chr7	1108.4	1111.4	2.98		Del	*IMMP2L*
12265.p1	chr16	86.6	89.5	2.94		Dup	*ABAT*
13572.p1	chr9	44.6	47.4	2.75		Dup	*SLC1A1, C9orf68, PPADC2, CDC37LL1, AK3*

12058.p1	chr3	605.3	608	2.71		Del	*FHIT*
11947.p1	chr6	1626.9	1629.5	2.58		Dup	*PARK2*
12389.p1	chr6	1625.5	1628	2.5		Dup	*PARK2*

REFERENCES

1. Bailey, J.A., Yavor, A.M., Massa, H.F., Trask, B.J., and Eichler, E.E. (2001). Segmental duplications: organization and impact within the current human genome project assembly. Genome Res *11*, 1005-1017.
2. Bailey, J.A., Gu, Z., Clark, R.A., Reinert, K., Samonte, R.V., Schwartz, S., Adams, M.D., Myers, E.W., Li, P.W., and Eichler, E.E. (2002). Recent segmental duplications in the human genome. Science *297*, 1003-1007.
3. Sharp, A.J., Hansen, S., Selzer, R.R., Cheng, Z., Regan, R., Hurst, J.A., Stewart, H., Price, S.M., Blair, E., Hennekam, R.C., et al. (2006). Discovery of previously unidentified genomic disorders from the duplication architecture of the human genome. Nat Genet *38*, 1038-1042.
4. Girirajan, S., and Eichler, E.E. (2010). Phenotypic variability and genetic susceptibility to genomic disorders. Hum Mol Genet.
5. Cooper, G.M., Stone, E.A., Asimenos, G., Green, E.D., Batzoglou, S., and Sidow, A. (2005). Distribution and intensity of constraint in mammalian genomic sequence. Genome Res *15*, 901-913.
6. Itsara, A., Cooper, G.M., Baker, C., Girirajan, S., Li, J., Absher, D., Krauss, R.M., Myers, R.M., Ridker, P.M., Chasman, D.I., et al. (2009). Population analysis of large copy number variants and hotspots of human genetic disease. Am J Hum Genet *84*, 148-161.
7. Cooper, G.M., Coe, B.P., Girirajan, S., Rosenfeld, J.A., Vu, T.H., Baker, C., Williams, C., Stalker, H., Hamid, R., Hannig, V., et al. (2011). A copy number variation morbidity map of developmental delay. Nat Genet *43*, 838-846.
8. Warde-Farley, D., Donaldson, S.L., Comes, O., Zuberi, K., Badrawi, R., Chao, P., Franz, M., Grouios, C., Kazi, F., Lopes, C.T., et al. (2010). The GeneMANIA prediction server: biological network integration for gene prioritization and predicting gene function. Nucleic Acids Res *38*, W214-220.