Supplemental Data

Structural Variation of Chromosomes

in Autism Spectrum Disorder

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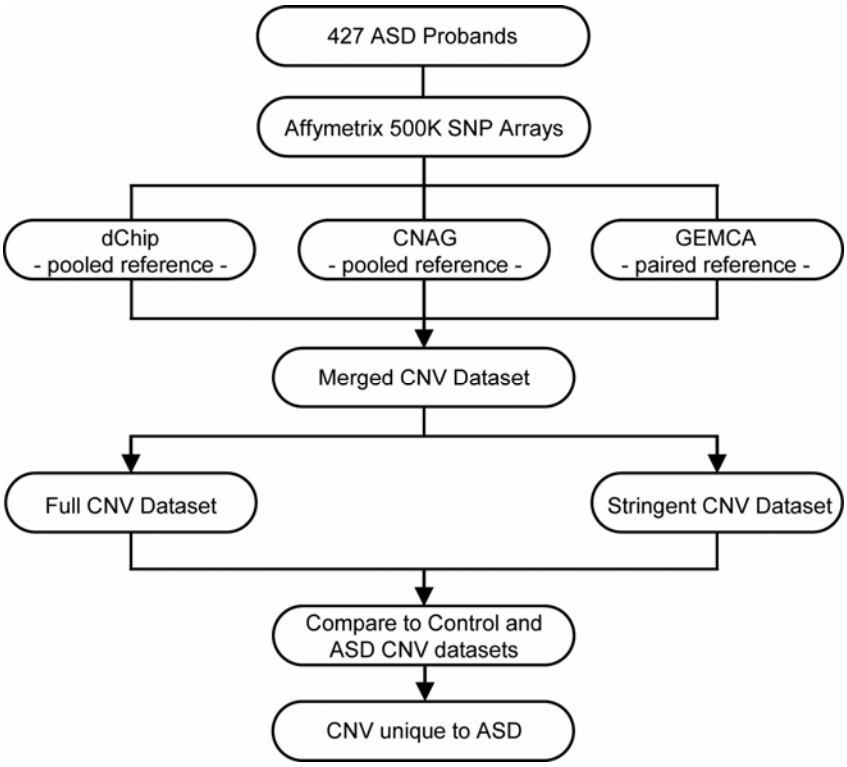

Figure S1. Flow Chart for CNV Analyses of 427 Unrelated-ASD Cases
DNA samples were genotyped using the Affymetrix 500K arrays and analyzed for CNV content using dChip, CNAG, and GEMCA (see Materials and Methods and Table 1 for details). For dChip and CNAG *NspI* and *StyI* arrays were analyzed separately, whereas for GEMCA the *NspI* and *StyI* arrays were merged before CNV analysis. We detected a merged (*NspI* and *StyI*) average of 3.0, 5.6, and 5.5 CNVs with dChip, CNAG and GEMCA, respectively. Results from the three algorithms were merged into a unique full CNV dataset (see Table 1 for statistics). A high confidence stringent dataset was defined as one in which a CNV was detected by 2 or more algorithms or on both arrays. Both the full and stringent dataset were compared to controls (see Materials and Methods) to define CNVs unique to ASD (see Table 1 and Figure 2).

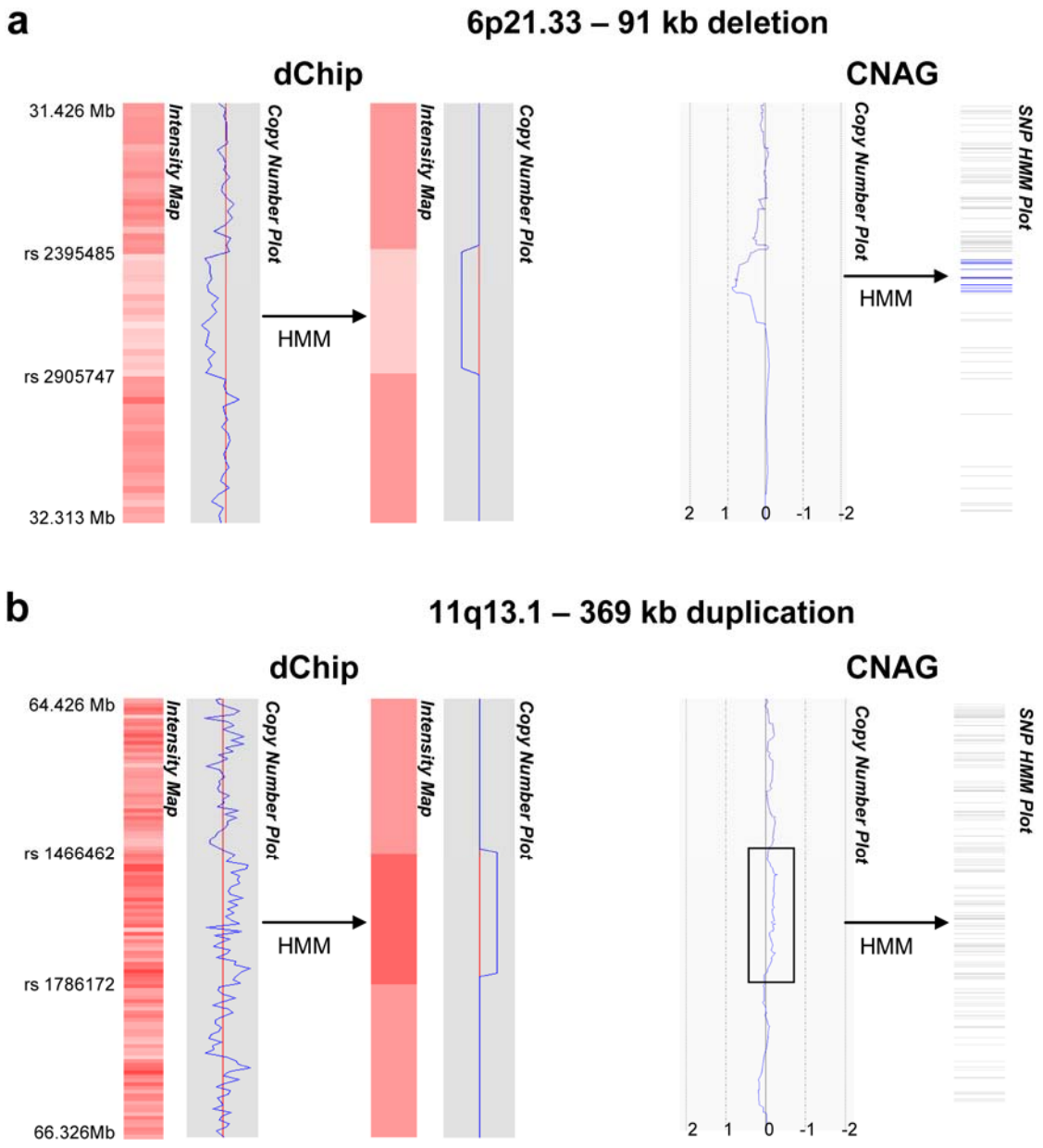

Figure S2. CNV Calling of Stringent versus Nonstringent Data
A 90 kb deletion was called by both dChip and CNAG (a) whereas a 369 kb duplication was only called by dChip (b). For dChip displays, a SNP intensity heat map (red = copy gain; white = copy loss) is accompanied by a raw copy number plot (blue). For CNAG, a $\log_2$ intensity plot of copy number is shown (blue). For both dChip and CNAG the actual copy number is inferred from the underlying raw copy number using a Hidden Markov model (HMM). The HMM calls in (a) agree with the underlying raw intensities whereas the dChip duplication call in (b) is likely a false positive resulting from noisy underlying data (box depicts the approximate location of the dChip CNV call). Genomic coordinates are from NCBI Build 35 and SNPs are depicted as 'rs' numbers from NCBI (Build 35).

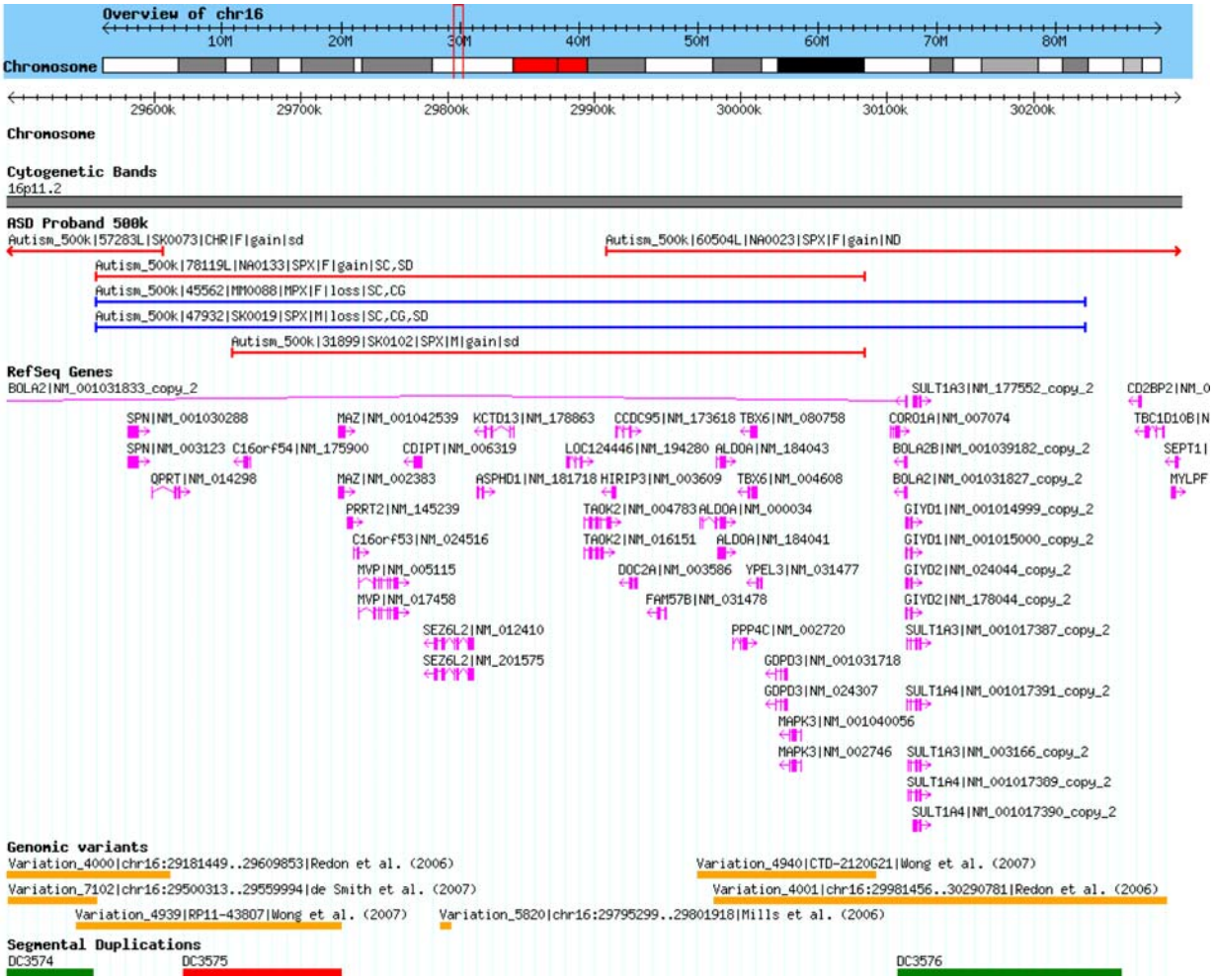

Figure S3. The Chromosome 16p11.2 Region as Depicted in the Autism Chromosome Rearrangement Database

The red box on chromosome ideogram shows approximate location of region (top) with scaled coordinates (NCBI Build 35) and cytogenetic band (below). CNV deletions (blue) and gains (red) are shown for ASD probands overlapping genes (pink). CNV found in healthy individuals (orange) is from the Database of Genomic Variants, with tags denoting study origin. Segmental duplications are depicted as being either inter-(red) or intra-(green) chromosomal.

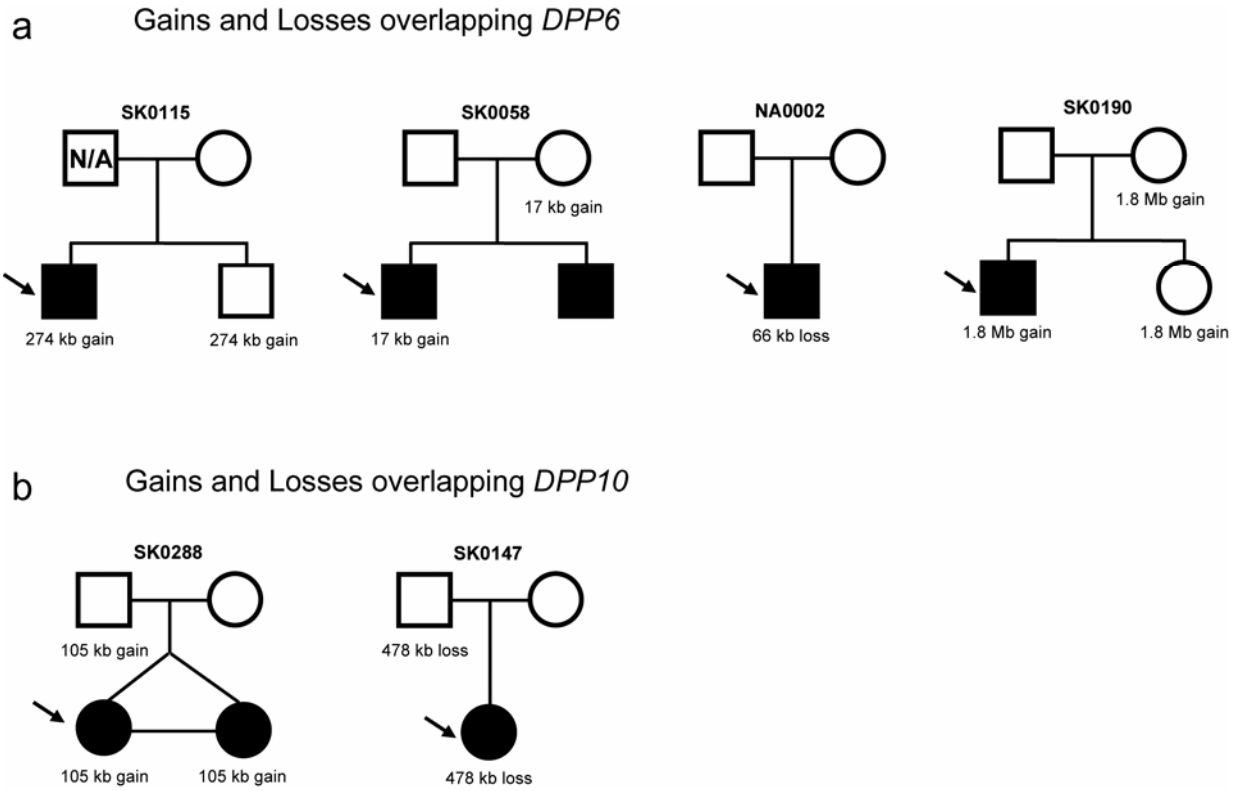

Figure S4. *DPP6* and *DPP10* CNVs
Overlapping gains and losses were found at *DPP6* (a) and *DPP10* (b), are shown. Males are denoted by squares and females by circles. The estimated size of each *de novo* or inherited event is shown below each family member. Arrows denote probands, open shapes are unaffected, filled have ASD diagnosis, and grey denotes developmental delay but not a definitive ASD diagnosis. Diamonds indicate number of older unaffected siblings of unspecified gender.

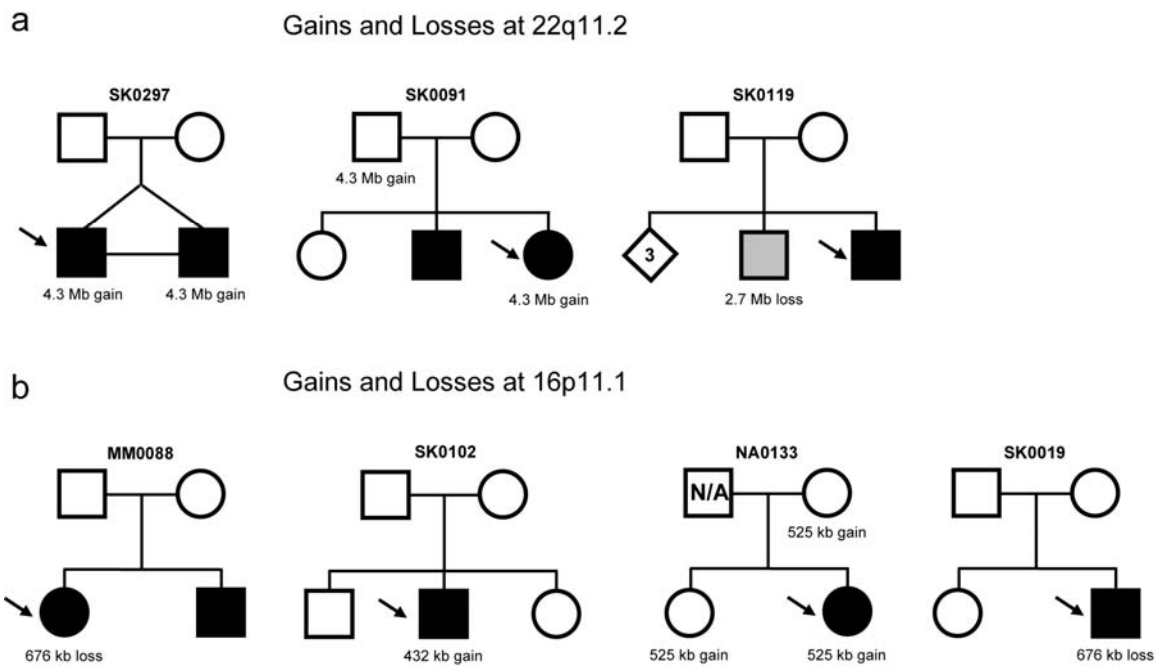

Figure S5. Chromosome 22q11.2 and 16p11.2 CNVs
Overlapping gains and losses were found at 22q11.2 (a), 16p11.2 (b), are shown. Males are denoted by squares and females by circles. The estimated size of each *de novo* or inherited event is shown below each family member. Arrows denote probands, open shapes are unaffected, filled have ASD diagnosis, and grey denotes developmental delay but not a definitive ASD diagnosis. Diamonds indicate number of older unaffected siblings of unspecified gender. Note that SK0119-003 originally entered the study with an ASD diagnosis but upon re-examination after CNV detection was assessed to be below cutoffs for ASD.

Table S1. Cohort Breakdown by Institution and Family Type

Site / Family Type		The Hospital for Sick Children	McMaster University	Memorial University	Other Sites[1]	Total[2]
CHR	SPX	9	0	4	11	24
	MPX	5	0	0	3	8
Non-CHR	SPX	132	7	75	0	214
	MPX	82	92	7	0	181
ALL		228	99	86	14	427

[1]Other sites refer to those 14 cases imported into the study because they already had known chromosome rearrangements.
[2]Analysis of population ancestry using STRUCTURE revealed ~90% (386/427) of probands were mainly of European origin, while 4.5% (19/427) were of European-mixed origin, 4.5% (19/427) were mainly of Asian origin and 0.07% (3/427) were mainly of African origin. Probands were clustered without regards to their original geographical origin using 780 unlinked SNPs, assuming three ancestral populations, and using 209 unrelated HapMap individuals (African, European and Asian) as reference in the same clustering.

Table S2. CNV in ASD Probands with Abnormal Karyotypes

	Sample ID	Phenotype/Family type	Cytogenetic Analysis			CNV Analysis						Comments
			Karyotype	Breakpoint Location	RefSeq Genes	Chr	CNV	Size (bp)	Location	AS/Str[a]	RefSeq Genes	
1	NA0008-000	Simplex family ASD, developmental dyspraxia	46, XX, t(2;6)(q32;p22) Inheritance unknown	2q33.1: 200,096,682 – 200,154,790	*SATB2*	2p11.2	Loss	917,200	89,056,400-89,973,600	No/NS	No known genes	
				6p22.3: 21,561,566 – 21,644,040	No known genes	6p21.33	Gain	54,600	30,134,300-30,188,900	Yes/NS	*ZNRD1, PPP1R11, RNF39, TRIM31*	
						11p13	Gain	54,200	35,332,700-35,386,900	No/NS	*SLC1A2*	
						13q21.33	Loss	28,200	69,642,500 - 69,670,700	No/NS	No known genes	
						14q11.2	Gain	549,300	21,490,300-22,039,600	No/NS	No known genes	
						14q32.33	Loss	64,000	106,152,000 - 106,216,000	No/NS	No known genes	
2	NA0005-000	Simplex family ASD, seizure disorder, obesity, macrocephaly	46,XX,t(4;5)(q21;q13) Inheritance unknown	4q21.3	Several	1p13.2	Gain	128,963	112,783,876-112,912,839	Yes/NS	*ST7L, CAPZA1*	
						2q37.3	Loss	602,914	242,127,468-242,730,382	No/S	10 genes	
						3q29	Loss	43,033	196,922,636-196,965,669	No/NS	*MUC20, MUC4*	
				5q14.2-q14.3: 82,802,678 – 91,285,973	Several	5q15	Loss	48,627	97,076,449-97,125,076	No/NS	No known genes	
						5q21.3	Loss	13,000	109,391,000-109,404,000	Yes/NS	No known genes	
						8p23.1	Gain	448,146	12,039,387-12,487,533	No/S	*FAM86B1, DEFB130, LOC440053*	
						14q11.2	Gain	223,579	19,272,965-19,496,544	No/S	6 *OR* genes	
						14q11.2	Gain	650,430	21,407,981-22,058,411	No/S	No known genes	
						15q11.2	Gain	1,642,961	18,446,422-20,089,383	No/NS	*LOC283755, POTE15, OR4M2, OR4N4*	
3	NA0039-000	Simplex family ASD, submucous cleft, globally developmentally delayed, large ears, short forehead, distally tapere fingers, severe pes planovalgus	46,XX,der(22)t(14;22)(q32;q13) pat Paternal inheritance	See CNV	See CNV	9q32	Gain	498,000	114,038,000-114,536,000	No/NS	7 genes	See Table 2 Unaffected sibling with ADHD has 46,XX,der(14)t(14;22)(q32;q13)
						14q32.33	Gain	1,436,000	104,920,000-106,356,000	No/NS	6 genes	
						15q13.3	Gain	502,500	29,796,300-30,298,800	No/NS	*CHRNA7*	
						22q13.31 – q31.33	Loss	3,231,700	46,277,400-49,509,100	Yes/NS	40 genes + *SHANK3*	
4	SK0283-003	Simplex family ASD	47,XX, ring chromosome 1 de novo	See CNV	See CNV	1p22.3	Gain	23,993	87,417,351-87,441,344	Yes/NS	No known genes	See Table 2
						1q21.2-q21.3	Gain	1,451,926	148,095,537-149,547,463	Yes/S	36 genes	
						3p26.1	Loss	44,458	5,365,506-5,409,964	Yes/S	No known genes	
						4p13	Gain	95,508	44,762,996-44,858,504	Yes/S	No known genes	
						4q33	Loss	82,224	171,715,627-171,797,851	Yes/NS	No known genes	
						5q31.3	Loss	355,649	140,658,658-141,014,307	Yes/NS	6 genes	
						6p12.3	Gain	13,950	46,962,122-46,976,072	No/NS	*GPR116*	
						7p14.1	Loss	102,939	38,041,635-38,144,574	No/NS	*STARD3NL, TARP*	
						7q34	Loss	169,191	141,813,948-141,983,139	No/NS	*PRSS1*	
						14q11.2	Loss	583,148	21,455,546-22,038,694	No/S	No known genes	
						15q11.2	Loss	1,632,769	18,427,103-20,059,872	No/S	*LOC283755, POTE15, OR4M2,*	

											OR4N4	
						17q21.31	Loss	140,746	41,570,665-41,711,411	No/NS	*KIAA1267*	
5	SK0044-003	Simplex family ASD	46, XY, t(1;2)(p22.1;p23)pat der(13;15)(q10;q10)mat Maternal/Paternal inheritance	1p31.1 : 72,065,578 – 72,163,007	*NEGR1*	7p14.1	Gain	85,900	39,828,000-39,913,900	No/NS	*CDC2L5*	
				2p24.3 : 12,376,807 – 12,733,637	No known genes							
				13q10 : in progress								
				15q10 : in progress								
6	SK0182-003	Simplex family ASD	46 XY, t(1;9)(q25;p13) Inherited	1q24.2: 167,452,268 - 167,522,136	No known genes	2p24.3	Gain	15,100	14,304,500-14,319,600	No/NS	No known genes	Younger sister has the same translocation and severe speech and language disorder but does not meet ASD criteria on ADOS.
				9p12: 45,695,701 – 45,737,008	No known genes	14q11.2	Gain	288,100	19,204,300-19,492,400	No/S	6 genes	
7	SK0335-003	Simplex Family ASD, mental retardation	46,XX,t(2;10)(q22;q22.3) Inheritance unknown	2q23.1: 148,938,284 – 149,125,547	*LOC401431, ATP6VOE2*	2p13.3	Gain	374,900	70,152,900-70,527,800	Yes/NS	6 genes	Non-Canadian family
				10q23.31: 91,265,490 – 91,461,660	*SLC16A12, PANK1, MPHOSPH1*	3q29	Gain	43,033	196,922,636-196,965,669	No/NS	*MUC20, MUC4*	
						5p13.1	Loss	272,618	38,534,384-38,807,002	Yes/S	*LIFR*	
						6p21.32	Gain	162,900	32,344,099-32,506,999	Yes/NS	*C6orf10, BTNL2*	
						8p23.1	Gain	21,783	12,264,620-12,286,403	No/NS	No known genes	
						9q32	Gain	22,000	114,153,000-114,175,000	No/S	*ORM1, ORM2*	
						14q11.2	Gain	331,503	21,717,112-22,048,615	No/S	No known genes	
						15q11.2	Gain	1,516,085	18,427,100-19,943,185	No/S	*LOC283755, POTE15, OR4M2, OR4N4*	
						16p11.2-11.1	Gain	266,336	34,325,041-34,591,377	No/NS	No known genes	
						17q21.31	Gain	201,731	41,518,102-41,719,833	No/S	*KIAA1267*	
						20p12.1	Loss	27,500	14,973,800 - 15,001,300	Yes/S	*C20orf133*	
8	SK0126-003	Multiplex family ASD	46,XY, t(2;11)(p11.2;q13.3) pat Paternal inheritance	2p11.2: 89,117,655 - 89,158,494	No known genes	2q34	Loss	3,000	213,013,000 - 213,016,000	Yes/NS	*ERBB4*	
				11q13.1: 64,821,333 - 64,861,285	*POLA2, CDC42EP2, DPF2*							
9	SK0152-003	Multiplex family ASD, oral motor apraxia, poor balance and coordination, mild hypotonia, walks with a wide gait, severe language delay, moderate intellectual disability, some facial features of Cri du Chat	46, XY, inv(3)(p24;q24), t(5;7)(p15p13) de novo	3p24 : not available		3p25.1-p24.3	Loss	1,409,600	15,125,800-16,535,400	Yes/S	12 genes	Previously described in a manuscript by Harvard *et al*[1]. The 3p25.1, 5p15.31-p15.2 and 18q12.2 deletions were identified using BAC CGH. The deletion size has been refined here using SNPs. Older sibling also has ASD but has a normal 46,XX karyotype Maternal aunt with schizophrenia and a maternal uncle with Down syndrome See Table 2
				3q24: not available		3p12.3	Gain	55,000	78,902,000 - 78,957,000	Yes/S	*ROBO1*	
				5p14.3 : 19,825,926 – 19,883,410	*CDH18*	5p15.31 – p15.2	Loss	3,429,389	9,275,811 – 12,705,200	Yes/S	8 genes	
				7p13: 46,618,434 - 46,733,542	No known genes	6q16.1	Loss	60,058	95,556,287-95,616,345	No/S	No known genes	
						7p14.1	Gain	35,243	38,096,725-38,131,968	No/NS	No known genes	
						10q11.22	Gain	455,130	47,030,119-47,485,249	No/S	*ANXA8*	
						12p11.21	Gain	63,728	31,904,362-31,968,090	No/S	No known genes	
						12q12	Loss	422,842	40,584,198-41,007,040	Yes/S	*YAF2, ZCRB1*	
						14q11.2	Gain	491,397	21,584,229-22,075,626	No/S	No known genes	
						14q32.33	Gain	22,269	106,223,861-106,246,130	No/NS	No known genes	
						15q11.2	Loss	1,632,718	18,446,422-20,079,140	No/S	*LOC283755, POTE15, OR4M2,*	
						16q21	Loss	91,432	63,768,909-63,860,341	Yes/NS	No known genes	

						17q21.31	Gain	219,797	41,500,036-41,719,833	No/NS	*KIAA1267*	
						18q12.2	Loss	816,914	32,174,061-32,990,975	Yes/S	*KIAA1328, C18orf10, FHOD3*	
10	SK0105-003	Multiplex family ASD, primarily non-verbal, profound developmental delay	46,XY,inv(4)(p12;p15.3)mat Maternal inheritance	4p15.3: 12,173,445 - 12,335,572	No known genes	10q11.21	Gain	1,098,400	41,956,500-43,054,900	Yes/NS	*RET, RASGEF1A, BMS1L, ZNF11B, MGC16291, GALNACT-2*	Described previously in Vincent *et al.*[2] Affected brother, apparently unaffected mother and unaffected maternal grandfather all have the same inversion. Distal 4p15.3 breakpoint maps ~12Mb to a region previously indicated to show linkage to autism.
				4p12: 44,876,353 – 46,024,486	*GABRG1* (breakpoint region is located in intron 7)	13q14.2	Gain	162,300	47,414,800 - 47,577,100	Yes/NS	*MED4, NUDT15, SUCLA2*	
						16q21	Loss	56,600	61,854,900-61,911,500	Yes/NS	No known genes	
						17q21.31	Gain	238,600	41,521,600-41,760,200	No/NS	*KIAA1267*	
11	SK0205-004	Simplex family ASD	46,XX,del(5)(p15.1) de novo	See CNV	See CNV	3q29	Gain	96,068	199,226,000-199,322,068	No/NS	*LMLN, LOC348840*	See Table 2 FISH analysis with subtelomeric probe (containing D5S2488) was consistent with a terminal deletion on 5p.
						5p15.33 – p15.2	Loss	13,800,984	81,949 – 13,882,933	Yes/S	>50 genes	
						5q15	Loss	70,891	97,054,185-97,125,076	No/NS	No known genes	
						10q11.22	Gain	1,121,866	46,363,383 - 47,485,249	No/S	*SYT15, ANXA8, ANXA8L1, PPYR1, GPRIN2*	
						10q21.3	Loss	29,732	67,747,770-67,777,502	No/NS	*CTNNA3*	
						10q26.3	Gain	244,432	135,079,000-135,323,432	No/S	*SYCE1;CYP2E1*	
						14q11.2	Gain	217,035	19,272,965-19,490,000	No/S	*OR4K1, OR4N2, OR4K5, OR4K2*	
						15q11.2	Gain	1,662,300	18,427,100-20,089,400	No/S	*LOC283755, POTE15, OR4M2, OR4N4*	
						17q21.31	Gain	65,845	41,006,823-41,072,668	No/S	No known genes	
						17q21.31	Gain	187,028	41,521,621-41,708,649	No/NS	*KIAA1267*	
						22q11.21	Gain	150,753	17,265,500-17,416,253	No/S	*DGCR6, PRODH, DGCR2*	
12	SK0061-003	Simplex family ASD, developmental delay	46, XY, t(5;7)(q15;q31.32) Inheritance unknown	7q31.31: 118,928,065 – 119,006,076	No known genes	No CNV detected						Non-Canadian Family
				5q14.3: 88,849,193 – 88,891,151	No known genes							
13	SK0195-003	Simplex family ASD	46,XY,t(5;8;17)(q31.1;q24.1;q21.3) de novo	5q31.1: 136,979,583 – 137,038,092	*KLHL3*	2p16.1	Gain	47,900	57,314,000-57,361,900	No/NS	No known genes	See Table 2
				8q24.22: 132,448,049 – 132,512,973	No known genes	10q23.1	Loss	17,500	83,772,000-83,789,500	Yes/NS	*NRG*	
				17q21.31: 41,893,216 - 42,093,636	*LRRC37A2, ARL17P1, LOC641522, NSF*	14q11.2	Gain	288,100	19,204,300-19,492,400	No/NS	*OR4K1, OR4N2, OR4M1, OR4K5, OR4Q3, OR4K2*	
						17q21.31	Gain	644,700	41,521,600-42,166,300	No/S	*KIAA1267*	
14	SK0133-003	Simplex family ASD	46,XY, t(6;7)(p11.2;q22)pat Paternal inheritance	6p12.1: 56,805,919 - 56,967,398	*DST, c6orf65*	2q37.1	Gain	314,000	232,076,000 - 232,390,000	Yes/NS	*MGC43122, NMUR1, MGC35154, NCL, B3GNT7*	CNV seen at 11q25 is in the same breakpoint region as SK0145-003
				7q22.1: 97,933,646 - 97,973,368	No known genes	5q14.3	Gain	633,400	89,492,800-90,126,200	Yes/NS	*CETN3, LOC153364, POLR3G, MASS1*	
						7q33	Loss	3,000	136,255,000-136,258,000	No/NS	No known genes	
						8q23.2	Loss	32,000	111,182,000 - 111,214,000	No/NS	No known genes	
						9p21.3	Loss	8,200	25,073,900 -	Yes/NS	No known genes	

									25,082,100			
						11q25	Gain	369,000	133,855,000 - 134,224,000	No/S	No known genes	
						12q21.33	Gain	19,700	90,807,700 - 90,827,400	Yes/NS	No known genes	
						13q21.32	Loss	2,500	65,576,300 - 65,578,800	Yes/NS	No known genes	
15	SK0043-003	Multiplex family ASD	46,XY,t(6;9)(q10;q12) Inheritance unknown	6q11.2-q12: 63,464,452 – 63,511,410	No known genes	8p23.2	Loss	35,040	3,984,190-4,019,230	No/NS	*CSMD1*	Sibling also has ASD but a normal 46,XY karyotype
				9q21.11: 68,599,032 – 68,682,365	*PIP5K1B*	15q11.2	Gain	1,713,200	18,376,200 - 20,089,400	No/S	*LOC283755, POTE15, OR4M2, OR4N4*	
16	SK0181-004	Simplex family ASD	46,XY,t(6;14)(q13;q21) de novo	6q12: 69,241,818-69,279,457	No known genes	3p14.1 – p13	Loss	5,346,900	65,286,300-70,633,200	Yes/S	13 genes	See Table 2
				14q21.1-q21.2: 40,807,716 – 44,806,460	*LRFN5, c14orf155, c14orf28, BTBD5, KIAA0423, PRPF39, FKBP3, AK093422, KIAA1596,FANCM, c14orf106*	4q28.3	Loss	254,000	135,282,000 - 135,536,000	No/NS	No known genes	
17	SK0083-003	Simplex family ASD, craniosynostosis, developmental verbal dyspraxia, motor delay	46, XY, del(7)(q31.1q31.32) de novo	7q31.1: 108,272,363 – 108,337,904	*IMMP2L, LRRN3, DOCK4, ZNF277P, IFRD1 … to … ASZ1, CFTR, CTTNBP2, LSM8, ANKRD7*	1q31.1	Loss	15,000	186,702,000 - 186,717,000	No/S	No known genes	See Table 2 Described previously in Feuk *et al.*[3]
				7q31.31: 119,007,999 – 119,335,246		2p23.3	Gain	26,300	25,138,000-25,164,300	Yes/NS	No known genes	
						4q35.2	Gain	21,314	188,232,000 - 188,253,314	Yes/S	No known genes	
						6p24.2	Gain	188,500	11,479,600-11,668,100	Yes/NS	No known genes	
						7q31.1 – q31.31	Loss	11,023,506	108,200,381-119,223,887	Yes/S	>50 genes	
						7q36.2	Loss	26,297	152,027,450-152,053,747	Yes/NS	No known genes	
						8q24.21	Gain	48,000	127,951,000 - 127,999,000	Yes/NS	No known genes	
						10p11.23	Gain	26,700	30,893,400 - 30,920,100	Yes/NS	No known genes	
						14q11.2	Loss	219,458	19,272,965-19,492,423	No/S	*OR4K1, OR4N2, OR4M1, OR4K5, OR4Q3, OR4K2*	
						17q21.31	Loss	117,521	40,897,617-41,015,138	No/NS	*PLEKHM1*	
18	SK0131-003	Simplex family Autistic features, speech-language disorder (developmental verbal dyspraxia), dysmorphic features, mild developmental delay, unable to cough/sneeze/laugh spontaneously	46, XX, del(7)(q31.2q32.2)(D7S486-, D7S522-) de novo, WBS inv-2 de novo	7q31.1: 113,181,975 – 113,518,235	*FOXP2, MDFIC, TFEC, TES, CAV2, CAV1...to...IRF5, TNPO3, TSPAN33, SMO, FAM40B, KIAA0828*	2p22.2	Gain	67,740	37,848,232-37,915,972	No/NS	No known genes	See Table 2 Described previously in Feuk *et al.*[3]
				7q32.2: 128,540,690 – 128,796,716		3p21.31	Gain	52,599	147,754,068-147,806,667	Yes/NS	*CCR5, CCRL2, CCR2*	
						4q31.21	Gain	120,171	145,146,000-145,266,171	No/S	*GYPE*	
						7p14.1	Gain	147,076	38,096,725-38,243,801	No/NS	*AMPH*	
						7q31.1-q32.2	Loss	15,486,721	113,335,000-128,821,721	Yes/S	>50 genes	
						8q13.3	Gain	261,985	72,881,221-73,143,206	Yes/NS	*MSC, TRPA1*	
						10q11.22	Gain	455,100	47,030,100-47,485,200	No/NS	ANXA8	
						10q26.2	Gain	91,077	128,501,014-128,592,091	Yes/S	*DOCK1*	
						13q21.33	Loss	44,235	69,634,065-69,678,300	No/NS	No known genes	
						14q11.2	Loss	222,786	19,272,965-19,495,751	No/NS	*OR4K1, OR4N2, OR4M1, OR4K5, OR4Q3, OR4K2*	
						14q11.2	Gain	637,249	21,462,466-22,099,715	No/S	No known genes	
						15q11.2	Gain	1,662,280	18,427,103-20,089,383	No/NS	*LOC283755, POTE15, OR4M2,*	

											OR4N4	
						17q12	Gain	29,984	31,471,515-31,501,499	No/NS	No known genes	
						22q11.22	Gain	810,876	20,772,047-21,582,923	No/NS	6 genes	
19	SK0002-003	Simplex family ASD, psychosis	46,XX,inv(7)(p15.3;q22.1) Inheritance unknown	7p21.1: 18,284,397 – 18,302,387	No known genes	4q28.3	Gain	765,000	132,195,000-132,960,000	No/S	No known genes	Non Canadian-Family
				7q22.3: 104,360,659-104,549,945	*SPRK2*	5p15.1-15.2	Gain	239,100	14,940,400-15,179,500	No/S	No known genes	
						15q11.2	Gain	1,713,200	18,376,200 – 20,089,400	Yes/S	*LOC283755, POTE15, OR4M2, OR4N4*	
20	SK0211-003	Simplex family ASD, mild elevation of lactate	46,XX,inv(7)(q22q34)mat Maternal Inheritance	7q21.3: 96,943,657 - 96,985,663	No known genes	7q22.1	Gain	379,000	100,393,000-100,772,000	No/NS	10 genes	Non Canadian Family Mother and unaffected twin sister have the same karyotype; 7q34 breakpoint overlaps with a ASD translocation patient
				7q34: 140,920,721 - 140,958,207	*TAS2R4, TAS2R5*	9p21.1	Loss	135,100	30,408,400-30,543,500	No/NS	No known genes	
21	SK0040-003	Multiplex family ASD, ADHD, severe anxiety attacks, seizures, difficulties with fine and gross motor skills	46, XY, t(7;8)(p15;q22), t (10;11)(q26;q23) Inheritance unknown	7p15.3: 21,825,126-21,869,196	No known genes	2q37.3	Loss	95,959	242,634,423-242,730,382	No/S	No known genes	Non-Canadian Family Unaffected sister with normal female karyotype, has difficulties in some muscles, difficulties with fine and gross motor skills, severe anxiety attacks, not able to relate to peers and is affected by noise
				8q22.2: 99,652,299-99,823,618	*STK3*	10q21.3	Loss	144,903	67,734,600-67,879,503	No/S	*CTNNA3*	
						11q22.3	Loss	62,995	104,729,456-104,792,451	No/NS	No known genes	
				10q26: 127,985,179 – 131,365,091	Multiple genes	14q11.2	Gain	219,458	19,272,965-19,492,423	No/NS	*OR4K2, OR4N2, OR4K1, OR4K5*	
						14q11.2	Gain	224,329	21,784,072-22,008,401	No/NS	No known genes	
				11q23: 109,979,883 – 111,597,476	Multiple genes	15q11.2	Gain	1,662,280	18,427,103-20,089,383	No/S	*LOC283755, POTE15, OR4M2, OR4N4*	
						22q11.22	Loss	515,645	21,031,117-21,546,762	No/NS	*PRAME, SUHW2, SUHW1, GGTL4*	
						22q11.23	Gain	269,129	23,975,202-24,244,331	No/S	*CTA, LRP5L*	
22	SK0145-003	Simplex family ASD	46, XX, t(7;11)(q31;q25)mat Maternal inheritance	7q31.2: 114,573,150 – 114,611,613	No known genes	1p36.11	Gain	192,600	26,231,500-26,424,100	Yes/NS	8 genes	Apparently unaffected mother has the same 7q31.2 and 11q25 breakpoints
						2p24.2	Gain	14,233	17,416,366-17,430,599	Yes/NS	No known genes	
				11q25: 133,882,647 - 134,001,155	No known genes	3p23	Gain	28,509	34,844,620-34,873,129	Yes/NS	No known genes	
						5p15.33	Gain	3,029,476	165,712-3,195,188	Yes/NS	28 genes	
						6p22.2	Gain	25,841	25,576,804-25,602,645	Yes/NS	*LRRC16*	
						7p14.1	Gain	20,412	37,494,999-37,515,411	No/NS	No known genes	
						8q13.3	Gain	28,933	72,911,162-72,940,095	Yes/NS	*MSC*	
						10p12.1	Loss	98,961	27,642,965-27,741,926	No/S	*PTCHD3*	
						12p12.3	Gain	37,831	18,855,833-18,893,664	No/NS	No known genes	
						14q11.2	Gain	464,929	21,551,291-22,016,220	No/NS	No known genes	
						15q23-24.1	Gain	435,603	70,053,228-70,488,831	Yes/NS	9 genes	
						19q13.43	Gain	308,600	63,476,500-63,785,100	Yes/NS	18 genes	
23	SK0031-003	Simplex family ASD, very little language, global developmental delays	46, XY, t(7;13)(q31.3;q21) mat Maternal inheritance	7q31.2: 116,270,156 – 116,458,896	*ST7*	5p13.2	Loss	3,000	36,495,800 – 36,498,800	Yes/NS	No known genes	Non Canadian Family
						6p22.1-21.33	Gain	79,600	29,967,200-30,046,800	No/NS	*HLA-A*	
				13q21.1: 54,559,087 –	No known genes	9p23	Loss	112,000	11,895,600	No/NS	No known genes	

									12,008,400			
				54,739,454		14q32.2	Gain	772,400	99,015,100-99,787,500	Yes/S	8 genes	
						15q11.2	Gain	1,378,000	18,711,400-20,089,400	No/S	*LOC283755, POTE15, OR4M2, OR4N4*	
						17q21.31	Gain	597,300	41,569,000-42,166,300	No/NS	6 genes	
						22q11.23	Gain	251,200	23,989,000-24,240,200	No/S	*CTA-246H3.1, LRP5L*	
24	SK0073-003	Simplex family ASD, developmental delay, delayed expressive and receptive language	47,XX,idic(15)(q13) de novo	15q13: 28,918,525 – 31,848,963	*LOC400968, LOC283755, POTE15, OR4M2, OR4N4…to…ARHGAP11A, c15orf45, GREM1, RYR3*	1q25.2	Gain	424,000	176,522,000-176,946,000	Yes/NS	6 genes	See Tables 2 and 3 Described previously in Kwasnicka-Crawford *et al.*[4]
						2p23.3	Gain	703,500	24,701,300-25,404,800	Yes/NS	7 genes	
						4p16.3	Gain	997,460	1,692,240-2,689,700	Yes/NS	12 genes	
						4q35.1	Gain	311,000	185,856,000-186,167,000	Yes/NS	*CASP3, CCDC111, MLF1IP, ACSL1*	
						5q31.1	Gain	93,000	134,426,000-134,519,000	Yes/S	No known genes	
						9p21.1	Loss	362,900	30,452,800-30,815,700	Yes/NS	No known genes	
						14q11.2	Gain	414,900	21,660,700-22,075,600	No/NS	No known genes	
						15q11.2-13.3	Gain	11,922,600	18,376,200-30,298,800	Yes/S	>50 genes	
						16p11.2	Gain	1,543,900	28,062,200-29,606,100	No/NS	>20 genes	
						16p11.2	Gain	658,600	30,589,900-31,248,500	No/NS	>20 genes	
25	SK0218-003	Multiplex family ASD, cleft palate, club feet, mild-facial hypoplasia, heart defect	46,XX,del(18)(q21) de novo	18q21.32: 55,690,398-55,884,029	See CNV	12p13.33	Loss	92,328	1,760,084-1,852,412	Yes/S	*CACNA2D4, ADIPOR2, LRTM2*	See Table 2 As noted in the Autism Chromosome Rearrangment Database there are 5 addition reported cases of abnormalities involving 18q; Sibling has a normal 46,XY karyotype also is affected with autism and has oromotor difficulties .
						15q11.2	Loss	1,613,450	18,446,422-20,059,872	No/S	*LOC283755, POTE15, OR4M2, OR4N4*	
						17q21.31	Gain	190,234	41,518,415-41,708,649	No/NS	*KIAA1267*	
						18q21.32-q23	Loss	20,358,999	55,756,601-76,115,600	Yes/S	>50 genes	
						19q13.42	Loss	68,786	59,971,717-60,040,503	No/NS	*KIR3DP1, KIR2DL1, KIR3DL1, KIR2DL4, KIR2DS4*	
						20p11.23	Gain	128,457	19,740,012-19,868,469	Yes/NS	*RIN2*	
26	SK0215-006	Simplex family ASD	46,XY,t(19;21)(p13.2;q22.12) Inherited	19p13.2: 7,804,294 – 7,896,711	*EVI5L, FLJ22184, LRRC8E, MAP2K7, SNAPC2, CTXN1*	1p21.3	Loss	1,092,500	97,271,600-98,364,100	Yes/S	*FLJ35409, DPYD*	Patient has an unaffected sister with the same karyotype
				21q22.12: 36,091,999 – 36,191,098	No known genes	17p11.1-p11.2	Gain	503,100	21,634,900-22,138,000	Yes/NS	*FAM27L*	
27	SK0136-003	Simplex family ASD	46,X,der(Y)t(Y;15)(q12;p11.2) pat Paternal inheritance	Not available		4p13	Gain	42,400	44,809,500-44,851,900	No/NS	No known genes	
						8p23.2	Gain	234,580	2,335,310 - 2,569,890	No/NS	No known genes	
						8q24.23	Loss	138,000	137,757,000-137,895,000	No/NS	No known genes	
						10p12.1	Loss	51,400	27,690,500-27,741,900	No/NS	*PTCHD3*	
						15q11.2	Loss	558,300	18,676,700-19,235,000	No/NS	*LOC283755*	
						15q26.3	Gain	388,100	99,827,900-100,216,000	No/NS	*PCSK6, TARSL2, TM2D3, OR4F6*	
28	SK0243-003	Simplex Family ASD	46,XY,del(15)(q23q24.2) de novo	See CNV	See CNV	1q21.1	Loss	333,539	145,700,996-146,034,535	No/NS	No known genes	See Table 2
						2p22.2	Gain	52,951	37,847,789 -	No/NS	No known genes	

									37,900,740			
						3q27.3	Gain	91,422	187,897,578-187,989,000	No/S	*KNG1, EIF4A2*	
						7p22.3	Gain	29,778	141,322-171,100	No/NS	No known genes	
						7p14.1	Loss	32,636	38,092,579-38,125,215	No/NS	No known genes	
						10p13	Loss	1,570	13,096,593-13,098,163	No/NS	No known genes	
						11p15.1	Gain	21,766	18,905,796-18,927,562	No/NS	*MRGPRX1*	
						15q23-q24.2	Loss	4,289,500	69,601,300-73,890,800	Yes/S	55 genes	
						17q12	Gain	38,247	31,463,252-31,501,499	No/NS	No known genes	
						17q21.31	Gain	83,359	41,636,474-41,719,833	No/NS	No known genes	
29	SK0245-005	Simplex Family ASD, epicanthal folds, drooping eyes	46,XY,trp(15)(q11.2q13) de novo	See CNV	See CNV	6q14.1	Loss	47,288	79,036,117-79,083,405	No/NS	No known genes	See Tables 2 and 3
						7p14.1	Loss	57,861	38,067,354-38,125,215	No/NS	No known genes	
						10p13	Loss	2,538	13,095,625-13,098,163	No/NS	*TARP*	
						11p15.1	Loss	12,459	18,905,796-18,918,255	No/NS	*MRGPRX1*	
						14q11.2	Loss	219,458	19,272,965-19,492,423	No/S	6 genes	
						14q32.33	Gain	27,408	106,223,861 106,251,269	No/NS	No known genes	
						15q11.2-q13.3	Gain	11,871,747	18,427,100 30,298,847	Yes/S	>50 genes	
						19p13.2	Loss	132,251	6,902,567 7,034,818	No/S	*EMR4, FLG25758, MBD3L2, ZF557*	
30	NA0097-000	Simplex Family ASD	46,XX,t(11;12)(q23.3;p13.3) Inheritance unknown	11q23: not available		2p25.3-2p15	Gain	63,451,406[b]	2,994 63,454,400	Yes/S	>50 genes	
						3p24.2	Loss	159,273	25,980,400-26,139,673	No/NS	No known genes	
						12p11.21	Gain	236,006	31,065,545-31,301,551	No/S	*DDX11, OVOS2*	
				12p13.32-p13.31: 4,341,718 - 7,918,138	Multiple genes	14q11.2	Gain	489,269	21,498,204 21,987,473	No/NS	No known genes	
						Xp22.33-Xp22.31	Loss	5,825,311	34,419-5,859,730	Yes/S	21 genes	
31	SK0300-003	Multiplex Family ASD, NF1	46,X,inv(Y)(p11.2q11.2)pat Paternal inheritance	Not available		4p16.1	Gain	35,832	7,801,488-7,837,320	Yes/NS	*SORCS2*	
						5p15.33	Gain	124,630	752,190-876,820	No/S	*ZDHHC11*	
						6p25.1	Loss	215,567	4,200,904-4,416,471	Yes/S	No known genes	
						8q24.23	Loss	198,193	137,757,137-137,955,330	No/S	No known genes	
						11p15.4	Loss	54,390	6,845,440-6,899,830	Yes/S	*OR10A2, OR10A4, OR2D2, OR2D3*	
						14q11.2	Loss	229,676	19,272,965-19,502,641	No/NS	6 genes	
						15q11.2	Loss	1,908,356	18,427,103-20,335,459	No/S	*LOC283755, POTE15, OR4M2, OR4N4*	
						15q21.2	Gain	183,903	48,583,127-48,767,030	Yes/S	*TRPM7, USP50*	
						Xp11.23	Loss	83,750	47,643,250-47,727,000	No/S	*ZNF630, SSX6*	
32	SK0094-005	Multiplex Family ASD	46,XX,ins(21;?)(p11.2;?) Inheritance unknown	Not available		7q21.2	Loss	509,800	90,919,200-91,429,000	Yes/NS	*MTERF, AKAP9, CYP51A1, LOC401387*	
						9q32	Gain	211,000	112,463,000	No/NS	*KIAA1958, C9orf80*	

									112,674,000			
						10q11.22	Gain	124,800	47,030,100-47,154,900	No/NS	No known genes	
						14q32.33	Gain	186,000	105,829,000-106,015,000	No/NS	No known genes	
						Xq23	Loss	888,000	112,325,000-113,213,000	Yes/NS	No known genes	

[a]AS/Str indicates if the CNV is Autism Specific (AS) (Yes or No) and if it belongs to the stringent (S) or non-stringent dataset (NS).
[b]63 Mb gain on chromosome 2 is suspected to be a cell line artifact.

[1]Harvard C, Malenfant P, Koochek M, et al. A variant Cri du Chat phenotype and autism spectrum disorder in a subject with de novo cryptic microdeletions involving 5p15.2 and 3p24.3-25 detected using whole genomic array CGH. Clin Genet 2005;67(4): 341-51.
[2]Vincent JB, Horike SI, Choufani S, et al. An inversion inv(4)(p12-p15.3) in autistic siblings implicates the 4p GABA receptor gene cluster. J Med Genet 2006;43(5): 429-34.
[3]Feuk L, Kalervo A, Lipsanen-Nyman M, et al. Absence of a paternally inherited FOXP2 gene in developmental verbal dyspraxia. Am J Hum Genet 2006;79(5): 965-72.
[4]Kwasnicka-Crawford DA, Roberts W, Scherer SW et al. Characterization of an autism-associated segmental maternal heterodisomy of the chromosome 15q11-13 region. J Autism Dev Disord 2007;37(4):694-702.

Table S3. Table of All Autism-Specific CNV (NCBI build 35) Displayed in Figure 1

FamID	Sex	Type	Chr	Start	Stop	Size	CNV	CNV Category
NA0074-000	M	SPX	1	41,463,611	41,924,314	460,704	gain	CNVs that are Singletons
SK0036-003	F	SPX	1	57,936,233	58,514,629	578,396	gain	CNVs that are Singletons
MM0236-004	M	MPX	1	60,369,200	61,426,300	1,057,101	gain	CNVs that are Singletons
MM0020-004	M	MPX	1	65,649,086	65,713,423	64,338	gain	CNVs that are Singletons
NA0076-000	M	SPX	1	91,930,266	92,330,344	400,078	gain	CNVs that are Singletons
SK0215-006	M	CHR	1	97,271,600	98,364,100	1,092,500	loss	CNVs confirmed *de novo*
SK0174-003	M	SPX	1	108,046,000	108,246,283	200,284	loss	CNVs that are Singletons
SK0283-003	F	CHR	1	148,095,537	149,547,463	1,451,926	gain	CNVs that are Singletons
MM0011-003	M	MPX	1	165,908,677	166,028,402	119,726	loss	CNVs that are Singletons
SK0132-003	M	MPX	1	186,673,899	186,716,570	42,672	loss	CNVs that are Singletons
SK0278-003	M	SPX	1	188,543,244	188,935,335	392,092	gain	CNVs that overlap the ACRD
MM0149-003	M	MPX	1	191,030,551	191,223,110	192,560	gain	CNVs that overlap the ACRD
NA0109-000	M	SPX	1	212,037,558	212,471,000	433,443	loss	CNVs that are Singletons
SK0183-004	M	SPX	1	238,633,145	239,606,926	973,781	loss	CNVs that are Singletons
SK0229-003	M	SPX	1	242,451,000	243,113,489	662,489	gain	CNVs that overlap the ACRD
NA0016-000	F	SPX	1	243,172,012	243,301,056	129,044	gain	CNVs that overlap the ACRD
MM0219-003	M	MPX	2	34,155,700	34,253,221	97,522	loss	CNVs that are Singletons
MM0295-003	M	MPX	2	34,662,196	34,780,515	118,320	loss	CNVs that are Singletons
NA0083-000	M	SPX	2	34,858,330	34,937,455	79,125	loss	CNVs that are Singletons
SK0270-003	M	SPX	2	39,992,374	40,053,300	60,926	loss	CNVs that are Singletons
NA0055-000	M	SPX	2	41,958,200	42,088,448	130,249	loss	CNVs that are Singletons
MM0063-003	F	MPX	2	50,780,202	50,859,200	78,999	loss	CNVs that overlap the ACRD
SK0301-003	M	MPX	2	52,856,046	52,969,575	113,530	loss	CNVs that are Singletons
SK0234-003	M	MPX	2	54,171,783	54,345,700	173,917	gain	CNVs that overlap the ACRD
SK0188-003	M	SPX	2	112,415,581	112,510,212	94,632	loss	CNVs that overlap the ACRD
SK0147-003	F	SPX	2	114,855,796	115,334,166	478,371	loss	CNVs Recurrent/Overlapping
SK0288-003	F	SPX-MZ	2	115,141,880	115,247,000	105,121	gain	CNVs Recurrent/Overlapping
NA0027-000	M	MPX	2	121,623,000	121,684,915	61,915	loss	CNVs that are Singletons
NA0057-000	M	SPX	2	125,496,832	125,890,571	393,740	loss	CNVs that are Singletons
MM0176-003	M	MPX	2	135,358,000	135,471,070	113,071	loss	CNVs that are Singletons
SK0225-003	M	SPX	2	155,849,451	155,988,560	139,109	loss	CNVs that are Singletons
SK0192-003	M	SPX	2	181,771,621	181,944,065	172,445	loss	CNVs that are Singletons
NA0030-000	M	SPX	2	186,674,000	186,786,323	112,324	loss	CNVs Recurrent/Overlapping

SK0306-004	F	SPX	2	186,674,000	186,771,130	97,131	loss	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
NA0007-000	M	SPX	2	195,170,000	195,217,247	47,248	gain	CNVs that are Singletons
MM0019-003	M	MPX	2	201,286,000	201,317,066	31,067	loss	CNVs that overlap the ACRD
MM0296-003	M	MPX	2	221,429,610	221,551,000	121,391	loss	CNVs that overlap the ACRD
NA0004-000	M	SPX	2	235,797,267	236,239,000	441,734	gain	CNVs that overlap the ACRD
MM0068-003	M	MPX	3	1,720,948	1,795,234	74,287	gain	CNVs that overlap the ACRD
SK0283-003	F	CHR	3	5,365,506	5,409,964	44,458	loss	CNVs that are Singletons
MM0210-004	M	MPX	3	7,957,390	8,250,541	293,151	gain	CNVs that are Singletons
SK0152-003	M	CHR	3	15,125,800	16,535,400	1,409,600	loss	CNVs confirmed *de novo*
NA0044-000	M	SPX	3	35,613,300	35,928,200	314,901	gain	CNVs that are Singletons
SK0021-008	M	MPX	3	36,110,965	36,215,909	104,945	loss	CNVs that are Singletons
MM0154-003	F	MPX	3	50,089,500	50,199,200	109,701	gain	CNVs that are Singletons
NA0067-000	M	SPX	3	61,075,295	61,581,100	505,806	gain	CNVs that overlap the ACRD
SK0181-003	M	CHR	3	65,286,300	70,633,200	5,346,900	loss	CNVs confirmed *de novo*
SK0152-003	M	CHR	3	78,902,000	78,957,000	55,000	gain	CNVs that are Singletons
NA0044-000	M	SPX	3	82,866,400	84,544,763	1,678,364	gain	CNVs that are Singletons
SK0023-003	M	SPX	3	99,400,957	99,484,400	83,443	gain	CNVs that are Singletons
NA0018-000	M	SPX	3	117,838,700	117,937,000	98,301	gain	CNVs that are Singletons
NA0003-000	M	SPX	3	124,386,373	124,456,000	69,628	gain	CNVs that are Singletons
NA0090-000	M	SPX	3	183,837,706	183,940,069	102,364	gain	CNVs that are Singletons
MM0296-003	M	MPX	4	328,851	542,862	214,012	gain	CNVs that overlap the ACRD
MM0228-004	M	MPX	4	11,820,924	11,983,053	162,130	loss	CNVs that overlap the ACRD
NA0129-000	M	SPX	4	38,109,899	38,349,444	239,546	gain	CNVs that overlap the ACRD
SK0283-003	F	CHR	4	44,762,996	44,858,504	95,508	gain	CNVs that overlap the ACRD
MM0010-005	M	MPX	4	44,773,367	44,846,800	73,434	gain	CNVs that overlap the ACRD
NA0093-000	M	SPX	4	44,773,367	44,846,800	73,433	gain	CNVs that overlap the ACRD
NA0044-000	M	SPX	4	55,718,164	55,811,710	93,547	loss	CNVs that are Singletons
SK0188-003	M	SPX	4	61,408,094	61,758,800	350,707	loss	CNVs that overlap the ACRD
SK0057-003	M	SPX	4	74,105,700	74,464,300	358,600	gain	CNVs that overlap the ACRD
MM0176-003	M	MPX	4	91,220,121	91,309,602	89,482	loss	CNVs that overlap the ACRD
NA0016-000	F	SPX	4	114,333,509	114,416,051	82,542	loss	CNVs that are Singletons
SK0012-003	M	SPX	4	152,993,000	153,381,007	388,008	gain	CNVs that are Singletons
SK0103-005	M	SPX	4	157,615,000	157,683,000	68,000	gain	CNVs that are Singletons
SK0012-003	M	SPX	4	162,387,402	163,362,655	975,254	gain	CNVs that overlap the ACRD
SK0012-003	M	SPX	4	173,324,616	174,954,056	1,629,441	gain	CNVs that overlap the ACRD

NA0037-000	M	SPX	4	179,692,000	179,865,679	173,680	gain	CNVs that are Singletons
MM0299-003	F	MPX	4	181,968,784	182,095,665	126,882	loss	CNVs that are Singletons
SK0266-003	M	SPX	4	183,466,000	183,517,000	51,000	loss	CNVs that are Singletons
SK0166-003	M	SPX	4	186,788,000	187,118,000	330,001	gain	CNVs that overlap the ACRD
SK0074-003	M	MPX	4	188,230,567	190,154,000	1,923,434	gain	CNVs that overlap the ACRD
SK0083-003	M	CHR	4	188,232,000	188,253,314	21,315	gain	CNVs that overlap the ACRD
MM0109-003	F	SPX	4	189,538,747	189,825,000	286,254	gain	CNVs that overlap the ACRD
SK0112-003	M	MPX	4	189,580,553	190,228,000	647,447	gain	CNVs that overlap the ACRD
MM0019-003	M	MPX	4	190,172,765	191,306,043	1,133,279	gain	CNVs that overlap the ACRD
SK0205-004	F	CHR	5	81,949	13,882,933	13,800,984	loss	CNVs confirmed *de novo*
SK0152-003	M	CHR	5	9,275,811	12,705,200	3,429,389	loss	CNVs confirmed *de novo*
SK0188-003	M	SPX	5	13,832,700	14,237,600	404,901	gain	CNVs that overlap the ACRD
SK0002-003	F	CHR	5	14,940,400	15,179,500	239,100	gain	CNVs that are Singletons
NA0078-000	M	MPX	5	25,125,371	25,450,672	325,302	gain	CNVs that are Singletons
NA0076-000	M	SPX	5	37,409,881	37,778,834	368,953	gain	CNVs that are Singletons
SK0335-003	F	CHR	5	38,534,384	38,807,002	272,619	loss	CNVs that are Singletons
NA0078-000	M	MPX	5	79,336,190	79,613,516	277,327	loss	CNVs that overlap the ACRD
NA0145-000	M	SPX	5	89,445,869	90,172,900	727,032	gain	CNVs that overlap the ACRD
MM0143-004	M	MPX	5	110,440,484	110,471,180	30,697	gain	CNVs that are Singletons
NA0023-000	F	SPX	5	113,104,916	113,178,000	73,084	loss	CNVs that are Singletons
SK0167-003	F	MPX	5	120,343,925	120,474,000	130,076	gain	CNVs that overlap the ACRD
NA0019-000	M	SPX	5	120,964,000	121,095,213	131,214	gain	CNVs that overlap the ACRD
SK0118-003	M	SPX	5	122,834,399	123,029,036	194,638	loss	CNVs that are Singletons
SK0077-003	M	SPX	5	128,968,799	129,433,000	464,201	gain	CNVs that are Singletons
MM0215-004	M	MPX	5	132,619,430	132,732,003	112,574	loss	CNVs that overlap the ACRD
SK0073-003	F	CHR	5	134,426,000	134,519,000	93,000	gain	CNVs that overlap the ACRD
SK0300-003	M	CHR	6	4,200,904	4,416,471	215,568	loss	CNVs that are Singletons
MM0212-004	F	MPX	6	17,505,095	17,703,208	198,114	gain	CNVs that are Singletons
MM0300-003	F	MPX	6	27,827,354	28,119,631	292,278	gain	CNVs that are Singletons
MM0225-004	M	MPX	6	69,929,900	70,278,043	348,144	gain	CNVs that are Singletons
SK0272-003	F	SPX	6	77,622,920	77,673,932	51,012	loss	CNVs that overlap the ACRD
MM0225-004	M	MPX	6	93,087,482	98,011,900	4,924,419	gain	CNVs that overlap the ACRD
SK0077-003	M	SPX	6	95,461,800	95,581,304	119,504	loss	CNVs that overlap the ACRD
SK0087-003	M	MPX	6	97,566,274	97,658,527	92,253	loss	CNVs that overlap the ACRD
SK0217-003	M	SPX	6	112,679,982	112,776,094	96,112	gain	CNVs that are Singletons

MM0220-003	M	MPX	6	118,799,000	119,117,000	318,001	gain	CNVs Recurrent/Overlapping
NA0025-000	M	SPX	6	118,823,011	119,117,000	293,990	gain	CNVs Recurrent/Overlapping
SK0326-003	M	SPX	6	137,930,847	138,011,644	80,798	gain	CNVs that are Singletons
SK0216-003	M	SPX	6	153,519,631	153,791,029	271,398	gain	CNVs that overlap the ACRD
MM0088-003	F	MPX	7	2,922,139	2,964,895	42,757	loss	CNVs that are Singletons
NA0147-000	M	SPX	7	3,946,854	4,002,686	55,833	loss	CNVs that are Singletons
SK0049-004	M	MPX	7	11,526,500	11,560,300	33,800	gain	CNVs that are Singletons
SK0132-003	M	MPX	7	20,242,925	20,345,800	102,876	gain	CNVs that are Singletons
NA0145-000	M	SPX	7	47,742,927	48,775,200	1,032,274	loss	CNVs that are Singletons
SK0083-003	M	CHR	7	108,200,381	119,223,887	11,023,507	loss	CNVs confirmed *de novo*
NA0061-000	M	SPX	7	108,357,049	108,597,525	240,477	loss	CNVs that overlap the ACRD
SK0131-003	F	CHR	7	113,335,000	128,821,721	15,486,722	loss	CNVs confirmed *de novo*
SK0226-005	M	SPX	7	118,462,717	118,679,189	216,473	loss	CNVs that overlap the ACRD
SK0190-003	M	SPX	7	152,698,000	154,478,000	1,780,000	gain	CNVs Recurrent/Overlapping
SK0115-003	M	SPX	7	153,098,000	153,372,000	274,001	gain	CNVs Recurrent/Overlapping
SK0058-003	M	MPX	7	153,539,745	153,556,533	16,789	gain	CNVs Recurrent/Overlapping
NA0002-000	M	SPX	7	153,585,000	153,651,462	66,463	loss	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
SK0262-003	M	SPX	8	710,491	1,501,580	791,089	gain	CNVs confirmed *de novo*
SK0119-003	M	MPX	8	17,706,313	17,738,524	32,211	loss	CNVs that are Singletons
SK0262-003	M	SPX	8	18,623,000	19,442,500	819,500	gain	CNVs that are Singletons
SK0077-003	M	SPX	8	42,971,601	43,820,300	848,699	gain	CNVs that are Singletons
SK0143-003	M	SPX	8	53,481,200	53,766,400	285,201	gain	CNVs Recurrent/Overlapping
MM0236-004	M	MPX	8	53,724,445	53,996,124	271,680	gain	CNVs Recurrent/Overlapping
SK0294-003	M	SPX	8	73,762,894	73,798,241	35,348	gain	CNVs that are Singletons
SK0076-003	F	SPX	8	83,989,256	84,141,278	152,022	gain	CNVs that are Singletons
MM0241-004	M	MPX	8	87,230,811	87,498,988	268,178	gain	CNVs that are Singletons
MM0218-004	M	MPX	8	89,598,961	89,678,800	79,840	loss	CNVs that overlap the ACRD
MM0210-004	M	MPX	8	104,166,572	104,947,190	780,618	gain	CNVs that are Singletons
SK0194-003	M	SPX	8	123,539,127	123,644,422	105,296	loss	CNVs that are Singletons
SK0292-003	F	MPX	8	130,467,000	130,529,193	62,194	loss	CNVs that are Singletons
MM0007-003	M	MPX	9	5,099,530	5,235,490	135,961	gain	CNVs that are Singletons
SK0270-003	M	SPX	9	7,725,280	7,764,180	38,900	loss	CNVs Recurrent/Overlapping
MM0103-003	M	MPX	9	7,725,283	7,760,233	34,951	loss	CNVs Recurrent/Overlapping
MM0711-003	M	MPX	9	16,092,066	16,379,100	287,035	gain	CNVs that are Singletons
SK0015-003	M	MPX	9	19,284,100	19,511,500	227,400	gain	CNVs that are Singletons

SK0015-003	M	MPX	9	19,702,200	24,674,100	4,971,900	loss	CNVs that are Singletons
SK0278-003	M	SPX	9	22,626,541	22,747,714	121,174	loss	CNVs that are Singletons
SK0148-005	F	SPX	9	24,607,036	24,682,114	75,078	loss	CNVs that are Singletons
MM0020-004	M	MPX	9	25,439,100	25,535,000	95,901	loss	CNVs that are Singletons
SK0210-004	M	MPX	9	28,577,800	29,218,800	641,000	loss	CNVs that overlap the ACRD
NA0105-000	M	SPX	9	33,054,336	33,294,800	240,465	gain	CNVs that are Singletons
SK0273-003	M	MPX	9	70,739,231	70,870,084	130,854	loss	CNVs that overlap the ACRD
NA0147-000	M	SPX	9	84,957,060	85,054,672	97,613	loss	CNVs that are Singletons
SK0045-003	M	MPX	9	109,446,000	109,837,000	391,000	gain	CNVs that are Singletons
SK0118-003	M	SPX	9	111,652,000	112,212,452	560,453	gain	CNVs that overlap the ACRD
NA0066-000	M	SPX	9	116,528,784	116,612,329	83,546	loss	CNVs that overlap the ACRD
MM0117-003	M	MPX	10	2,313,505	2,407,102	93,598	loss	CNVs that are Singletons
MM0225-004	M	MPX	10	4,976,040	5,124,511	148,472	gain	CNVs that are Singletons
MM1086-004	M	MPX	10	31,256,118	31,604,509	348,392	loss	CNVs that are Singletons
SK0102-004	M	SPX	10	42,611,900	43,266,300	654,400	gain	CNVs that overlap the ACRD
SK0102-004	M	SPX	10	44,988,900	45,468,800	479,900	gain	CNVs that overlap the ACRD
MM0068-003	M	MPX	10	68,139,200	68,246,027	106,828	loss	CNVs that are Singletons
NA0037-000	M	SPX	10	104,641,000	104,786,777	145,778	loss	CNVs that are Singletons
NA0109-000	M	SPX	10	112,267,330	112,405,408	138,079	gain	CNVs that overlap the ACRD
SK0131-003	F	CHR	10	128,501,014	128,592,091	91,078	gain	CNVs that overlap the ACRD
NA0138-000	M	SPX	10	133,285,000	133,604,999	320,000	gain	CNVs that overlap the ACRD
SK0300-003	M	CHR	11	6,845,440	6,899,830	54,391	loss	CNVs that are Singletons
NA0113-000	M	SPX	11	9,984,119	10,667,800	683,682	loss	CNVs that overlap the ACRD
SK0322-003	M	SPX	11	33,159,190	33,462,070	302,881	gain	CNVs that are Singletons
MM0272-003	M	MPX	11	40,285,800	40,548,738	262,939	loss	CNVs Recurrent/Overlapping
SK0167-003	F	MPX	11	40,417,554	40,610,400	192,847	loss	CNVs Recurrent/Overlapping
MM0305-003	M	MPX	11	68,053,777	68,204,900	151,123	gain	CNVs that are Singletons
NA0032-000	M	SPX	11	76,114,600	76,140,500	25,900	gain	CNVs that are Singletons
MM0212-004	F	MPX	11	99,148,202	99,289,243	141,042	loss	CNVs that are Singletons
SK0167-003	F	MPX	11	101,131,785	101,246,901	115,117	loss	CNVs that are Singletons
MM0112-005	M	MPX	11	116,789,980	116,855,347	65,368	gain	CNVs that are Singletons
MM0240-003	F	MPX	11	117,452,000	117,539,000	87,001	gain	CNVs that are Singletons
SK0255-003	M	SPX	11	124,303,460	124,719,976	416,517	gain	CNVs that are Singletons
NA0065-000	M	SPX	11	125,639,908	126,102,027	462,120	gain	CNVs that are Singletons
SK0218-003	F	CHR	12	1,760,084	1,852,412	92,328	loss	CNVs that overlap the ACRD

NA0172-000	M	SPX	12	3,727,911	3,879,230	151,320	loss	CNVs that are Singletons
SK0059-003	M	SPX	12	10,431,082	10,445,300	14,218	gain	CNVs that are Singletons
SK0152-003	M	CHR	12	40,584,198	41,007,040	422,842	loss	CNVs confirmed *de novo*
SK0326-003	M	SPX	12	46,170,200	46,365,774	195,575	gain	CNVs that are Singletons
SK0110-003	M	SPX	12	50,520,400	50,573,516	53,116	gain	CNVs that are Singletons
NA0071-000	F	SPX	12	57,408,270	58,532,356	1,124,087	gain	CNVs that are Singletons
SK0305-003	F	SPX	12	77,239,265	77,364,400	125,136	loss	CNVs that are Singletons
SK0301-003	M	MPX	12	83,388,935	83,428,800	39,866	gain	CNVs that are Singletons
NA0093-000	M	SPX	12	96,496,784	96,568,500	71,716	loss	CNVs that are Singletons
MM0711-003	M	MPX	12	96,576,486	96,639,686	63,201	loss	CNVs that are Singletons
SK0292-003	F	MPX	12	101,568,000	101,586,000	18,001	gain	CNVs that are Singletons
NA0109-000	M	SPX	12	110,646,607	110,800,000	153,394	gain	CNVs that are Singletons
MM0278-003	M	SPX	12	114,170,000	132,388,000	18,218,001	gain	CNVs confirmed *de novo*
MM0210-004	M	MPX	12	125,446,000	125,757,000	311,000	gain	CNVs that are Singletons
SK0079-003	M	MPX	13	17,960,300	18,492,994	532,694	gain	CNVs that are Singletons
NA0122-000	F	SPX	13	32,965,700	33,137,655	171,956	gain	CNVs that overlap the ACRD
NA0117-000	M	SPX	13	42,511,458	42,599,200	87,743	gain	CNVs that overlap the ACRD
MM0154-003	F	MPX	13	54,651,953	55,025,229	373,277	gain	CNVs that overlap the ACRD
NA0028-000	M	SPX	13	62,915,912	62,977,748	61,837	loss	CNVs that are Singletons
SK0023-003	M	SPX	13	66,470,851	66,660,289	189,438	gain	CNVs Recurrent/Overlapping
MM0299-003	F	MPX	13	66,487,899	66,660,300	172,402	gain	CNVs Recurrent/Overlapping
SK0326-003	M	SPX	13	89,726,966	90,134,219	407,254	gain	CNVs that are Singletons
NA0048-000	M	SPX	13	93,288,520	93,344,600	56,081	gain	CNVs that are Singletons
SK0326-003	M	SPX	13	93,497,400	93,732,931	235,532	gain	CNVs that are Singletons
SK0328-003	M	SPX	13	103,896,769	103,930,492	33,724	loss	CNVs that overlap the ACRD
SK0254-003	M	SPX	13	105,172,000	105,357,000	185,000	gain	CNVs that are Singletons
MM0295-003	M	MPX	13	113,361,712	113,646,000	284,289	gain	CNVs that overlap the ACRD
SK0305-004	F	SPX	14	42,022,286	42,210,026	187,741	loss	CNVs that overlap the ACRD
SK0320-003	M	MPX	14	45,537,581	45,653,418	115,838	loss	CNVs that overlap the ACRD
SK0121-003	M	SPX	14	76,007,842	76,924,400	916,558	gain	CNVs that are Singletons
MM0225-004	M	MPX	14	83,373,278	83,435,200	61,923	gain	CNVs that overlap the ACRD
SK0031-003	M	CHR	14	99,015,100	99,787,500	772,400	gain	CNVs that are Singletons
MM0154-003	F	MPX	14	106,223,861	106,356,482	132,622	gain	CNVs that overlap the ACRD
SK0073-003	F	CHR	15	18,376,200	30,298,800	11,922,600	gain	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
SK0245-005	M	CHR	15	18,427,100	30,298,847	11,871,747	gain	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*

SK0300-003	M	CHR	15	48,583,127	48,767,030	183,904	gain	CNVs that are Singletons
SK0243-003	M	CHR	15	69,601,300	73,890,800	4,289,500	loss	CNVs confirmed *de novo*
NA0064-000	M	SPX	15	82,573,421	83,631,697	1,058,276	loss	CNVs that overlap the ACRD
MM0256-004	M	MPX	15	87,922,400	87,993,909	71,510	gain	CNVs that overlap the ACRD
SK0326-003	M	SPX	15	97,406,000	97,961,522	555,523	gain	CNVs that are Singletons
SK0266-003	M	SPX	16	6,813,789	6,898,849	85,060	loss	CNVs that overlap the ACRD
MM0109-003	F	SPX	16	21,441,805	22,688,093	1,246,289	gain	CNVs Recurrent/Overlapping
MM0289-003	F	MPX	16	21,808,808	22,611,363	802,556	loss	CNVs Recurrent/Overlapping
MM0088-003	F	MPX	16	29,559,989	30,235,818	675,830	loss	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
SK0019-004	M	SPX	16	29,559,989	30,235,818	675,830	loss	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
NA0133-000	F	SPX	16	29,559,989	30,085,308	525,320	gain	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
SK0281-003	M	SPX	16	57,542,779	57,579,900	37,122	loss	CNVs that are Singletons
NA0063-000	M	SPX	16	73,397,667	73,657,067	259,400	loss	CNVs that overlap the ACRD
NA0095-000	M	SPX	16	74,576,356	74,613,000	36,645	loss	CNVs that overlap the ACRD
MM0310-005	M	MPX	16	80,972,252	80,983,135	10,884	loss	CNVs that are Singletons
SK0203-004	M	MPX	16	82,603,600	82,687,900	84,300	gain	CNVs that are Singletons
NA0067-000	M	SPX	16	87,800,593	88,066,260	265,668	loss	CNVs confirmed *de novo*
SK0085-004	M	MPX	17	3,836,592	3,998,867	162,276	gain	CNVs that are Singletons
SK0284-003	F	SPX	17	28,985,300	29,960,700	975,400	gain	CNVs that overlap the ACRD
MM0109-003	F	SPX	17	40,555,289	41,089,766	534,478	loss	CNVs that are Singletons
MM0240-003	F	MPX	17	40,555,289	41,128,323	573,035	loss	CNVs that are Singletons
SK0298-003	M	SPX	17	76,914,079	77,771,141	857,063	gain	CNVs that are Singletons
SK0328-003	M	SPX	18	13,794,043	14,743,900	949,858	gain	CNVs that are Singletons
SK0012-003	M	SPX	18	27,565,032	27,781,900	216,869	gain	CNVs that overlap the ACRD
SK0303-003	F	MPX	18	28,383,551	28,448,100	64,550	loss	CNVs that are Singletons
SK0152-003	M	CHR	18	32,174,061	32,990,975	816,914	loss	CNVs that overlap the ACRD
SK0147-003	F	SPX	18	37,509,556	37,950,450	440,895	gain	CNVs that overlap the ACRD
SK0304-003	M	SPX	18	46,101,841	46,218,000	116,160	gain	CNVs that overlap the ACRD
SK0014-003	M	SPX	18	52,531,252	53,165,421	634,169	gain	CNVs that are Singletons
SK0218-003	F	CHR	18	55,756,601	76,115,600	20,358,999	loss	CNVs confirmed *de novo*
NA0138-000	M	SPX	18	69,282,461	69,330,584	48,124	loss	CNVs that overlap the ACRD
SK0121-003	M	SPX	19	33,693,363	33,762,805	69,442	loss	CNVs that are Singletons
NA0111-000	M	SPX	19	57,836,600	58,246,200	409,601	gain	CNVs that are Singletons
NA0004-000	M	SPX	19	58,634,965	58,958,584	323,620	gain	CNVs that are Singletons
NA0070-000	F	SPX	19	60,499,398	60,742,656	243,259	loss	CNVs that are Singletons

SK0047-003	F	SPX	19	61,910,800	62,644,900	734,100	loss	CNVs that are Singletons
NA0110-000	M	SPX	19	63,050,356	63,193,800	143,445	loss	CNVs that are Singletons
SK0232-003	M	MPX	19	63,483,000	63,771,100	288,100	gain	CNVs that are Singletons
MM0018-003	M	MPX	20	11,319,093	11,424,900	105,808	loss	CNVs that are Singletons
SK0335-003	F	CHR	20	14,955,730	15,011,214	55,485	loss	CNVs that are Singletons
SK0258-004	M	SPX	20	45,468,000	45,673,300	205,300	gain	CNVs that are Singletons
MM0109-003	F	SPX	20	60,949,339	62,377,000	1,427,662	gain	CNVs confirmed *de novo*
MM0126-003	M	MPX	21	22,839,570	22,938,377	98,808	loss	CNVs that are Singletons
SK0118-003	M	SPX	21	28,060,406	28,250,400	189,995	loss	CNVs that are Singletons
SK0244-003	M	SPX	21	42,974,148	43,328,084	353,936	gain	CNVs confirmed *de novo*
SK0023-003	M	SPX	21	46,497,675	46,678,820	181,145	gain	CNVs that overlap the ACRD
SK0119-003	M	MPX	22	17,014,900	19,786,200	2,771,300	loss	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
SK0091-004	F	MPX	22	17,265,500	21,546,762	4,281,262	gain	CNVs Recurrent/Overlapping
SK0297-003	M	SPX-MZ	22	17,265,500	21,546,762	4,281,263	gain	CNVs Recurrent/Overlapping/CNVs confirmed *de novo*
SK0323-003	M	MPX	22	18,683,900	19,427,000	743,101	gain	CNVs Recurrent/Overlapping
NA0039-000	F	CHR	22	46,277,400	49,509,100	3,231,700	loss	CNVs confirmed *de novo*
SK0123-004	M	MPX	22	47,717,300	48,318,828	601,528	gain	CNVs Recurrent/Overlapping
MM0102-003	M	MPX	22	48,152,289	48,232,669	80,380	loss	CNVs Recurrent/Overlapping
MM0109-003	F	SPX	22	49,243,247	49,519,949	276,703	loss	CNVs confirmed *de novo*
NA0097-000	F	CHR	X	34,419	5,859,730	5,825,312	loss	CNVs confirmed *de novo*
SK0186-004	M	SPX	X	22,962,800	23,119,000	156,200	loss	CNVs that are Singletons
MM0087-003	M	MPX	X	25,516,263	25,620,400	104,138	loss	CNVs that are Singletons
NA0112-000	M	SPX	X	38,250,331	38,371,333	121,003	gain	CNVs that overlap the ACRD
NA0100-000	M	SPX	X	44,395,900	45,060,800	664,901	gain	CNVs that are Singletons
SK0306-004	F	SPX	X	48,073,600	52,716,966	4,643,367	gain	CNVs confirmed *de novo*
SK0087-003	F	MPX	X	83,866,300	92,175,100	8,308,800	loss	CNVs that are Singletons
MM0020-004	M	MPX	X	87,452,050	87,595,200	143,151	gain	CNVs that are Singletons
SK0228-003	M	SPX	X	104,153,000	104,638,000	485,000	gain	CNVs that are Singletons
SK0088-003	M	SPX	X	114,042,922	114,215,435	172,513	gain	CNVs that are Singletons
MM0087-003	M	MPX	X	130,406,000	130,695,499	289,500	gain	CNVs that are Singletons
NA0016-000	F	SPX	X	140,600,370	140,907,495	307,125	gain	CNVs that are Singletons
SK0234-003	M	MPX	X	142,561,000	142,682,000	121,000	loss	CNVs that are Singletons
SK0320-003	M	MPX	X	143,059,574	143,399,300	339,727	gain	CNVs that are Singletons
SK0123-004	M	MPX	X	147,974,000	148,479,449	505,449	gain	CNVs that are Singletons