Data supplement for Brownstein et al., Rate of Deleterious Copy Number Variants Similar in Early Onset Psychosis and Autism Spectrum Disorders: Implications for Clinical Practice. Am J Psychiatry (doi: 10.1176/appi.ajp.21111175)

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TABLE S1. Neurodevelopmental or neuropsychiatric-associated loci and genes investigated

Locus	CHR	START	STOP	TYPE	References for the association with NPD
1p36	1	1	2500000	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
1q21.1 TAR	1	145394955	145807817	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
1q21.1 distal+TAR	1	145394955	147394444	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
1q21.1 distal	1	146527987	147394444	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
NRXN1	2	50145643	51259674	Gene	Satterstrom et al. 2020
2q11.2	2	96742409	97677516	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
2q13	2	111394040	112012649	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
2q21.1	2	131481308	131930677	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
2q37	2	239716679	243199373	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
3q29	3	195720167	197354826	DEL	Sanders et al. 2019
				DUP	Coe et al. 2014
4p16.3	4	1552030	2091303	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
5q35	5	175720924	177052594	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
SIM1	6	100836750	100911811	Gene	Coe et al. 2014
7q11.23 WBS	7	72744915	74142892	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
7q11.23 distal	7	75138294	76064412	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
8p23.1	8	8098990	11872558	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
9q34	9	140513444	140730578	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
10q11.21q11.23	10	49390199	51058796	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
10q22q23	10	82045472	88931651	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
11p11.2	11	43940000	46020000	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
CRYL1	13	20977806	21100012	Gene	Coe et al. 2014
13q12.12	13	23555358	24884622	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
15q11.2	15	22805313	23094530	DEL	Sanders et al. 2019
15q11.2q12	15	22805313	28390339	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
15q13.1q13.2 BP3-BP4	15	29161368	30375967	DEL	Stefansson et al. 2014
				DUP	Stefansson et al. 2014
15q13.1q13.3 BP3-BP5	15	29161368	32462776	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
15q13.3 BP4-BP5	15	31080645	32462776	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
15q24	15	72900171	78151253	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
15q25.2	15	83219735	85722039	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
CREBBP	16	3775056	3930121	Gene	Satterstrom et al. 2020
16p13.11	16	15511655	16293689	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
16p12.1	16	21950135	22431889	DEL	Coe et al. 2014
				DUP	Coe et al. 2014

16p11.2 distal	16	28823196	29046783	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
16p11.2 distal+proximal	16	28823196	30200773	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
16p11.2 proximal	16	29650840	30200773	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
16p11.2p12.1	16	21596415	28347808	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
YWHAE	17	1247834	1303556	Gene	Coe et al. 2014
PAFAH1B1	17	2496923	2588909	Gene	Coe et al. 2014
17p12	17	14141387	15426961	DEL	Stefansson et al. 2014
				DUP	Stefansson et al. 2014
17p11.2	17	16812771	20211017	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
17q11.2 *NF1*	17	29107491	30265075	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
17q12	17	34815904	36217432	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
17q21.31	17	43705356	44164691	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
17q23.1q23.2	17	58302389	60289141	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
22q11.2 distal	22	21920127	23653646	DEL	Coe et al. 2014
				DUP	Coe et al. 2014
22q11.2 proximal	22	19037332	21466726	DEL	Sanders et al. 2019
				DUP	Sanders et al. 2019
SHANK3	22	51113070	51171640	Gene	Satterstrom et al. 2020

Legend: List of recurrent loci and genes previously associated with neuropsychiatric disorders.[1–4] Coordinates are presented in hg19 (Homo sapiens (human) genome assembly GRCh37) from Genome Reference Consortium; CHR: chromosome; BP: break points; WBS: William-Beuren syndrome.

TABLE S2. List of the recurrent neurodevelopmental or neuropsychiatric-associated CNVs identified in the EOP sample

Locus	Start (Hg19)	Stop (Hg19)	Type	Genes totally encompassed	Other CNV≥50Kb	Sex	Age of onset for EOP	ASD	ID
1q21.1 distal	146618800	147820000	DUP	PRKAB2, GJA5, FMO5, CHD1L, ACP6, NBPF24, BCL9, GJA8, GPR89B, AC242628.1	no	Male	13	no	no
1q21.1 distal	146618988	147825855	DUP	PRKAB2, ACP6, FMO5, BCL9, AC242628.1, CHD1L, GJA5, GPR89B, NBPF24, GJA8	no	Male	4	no	no
1q21.1 distal	146535353	147857135	DUP	GPR89B, GJA8, ACP6, AC242628.1, BCL9, PRKAB2, NBPF24, GJA5, CHD1L, FMO5	no	Male	15	yes	no
15q11.2 BP1-BP2	22762571	23080867	DEL	CYFIP1, NIPA2, TUBGCP5	no	Male	5	no	no
16p11.2 proximal	29592843	30264892	DEL	PAGR1, SEZ6L2, SLX1A, TBX6, MVP, TAOK2, BOLA2B, AC093512.2, HIRIP3, SULT1A3, YPEL3, MAZ, CDIPT, PRRT2, INO80E, C16orf54, SPN, NPIPB13, KIF22, C16orf92, CORO1A, ALDOA, TMEM219, ASPHD1, QPRT, KCTD13, ZG16, DOC2A, PPP4C, MAPK3, GDPD3, TLCD3B	no	Male	4	yes	yes
16p11.2 proximal	29652999	30357820	DUP	MVP, C16orf92, MAPK3, SLX1A, KCTD13, ZG16, C16orf54, AC093512.2, TBX6, CORO1A, PAGR1, CDIPT, PRRT2, ASPHD1, PPP4C, ALDOA, KIF22, YPEL3, SULT1A3, TAOK2, TMEM219, GDPD3, QPRT, MAZ, BOLA2B, SEZ6L2, NPIPB13, HIRIP3, SPN, TLCD3B, INO80E, DOC2A	no	Male	13	yes	no
16p13.11	14968855	16267250	DEL	MARF1, BMERB1, RRN3, ABCC1, NTAN1, CEP20, PDXDC1, MYH11, NDE1, MPV17L, NPIPA5, NPIPA1	yes (2)	Male	12	no	yes
16p13.11	14906734	16388596	DEL	RRN3, MPV17L, BMERB1, NOMO1, ABCC1, ABCC6, NPIPA5, NDE1, NTAN1, CEP20, MYH11, NPIPA1, PDXDC1, MARF1	no	Female	6	yes	--
16p13.11	14897788	16293305	DUP	MPV17L, MARF1, BMERB1, NPIPA5, CEP20, RRN3, NPIPA1, MYH11, NTAN1, NDE1, NOMO1, ABCC1, PDXDC1	no	Female	13	no	no
16p13.11	14897761	16276117	DUP	NTAN1, PDXDC1, NPIPA1, RRN3, MPV17L, MYH11, CEP20, NOMO1, ABCC1, NDE1, MARF1, BMERB1, NPIPA5	no	Female	4	yes	yes
22q11.2 proximal	18894835	20311763	DEL	ARVCF, PRODH, TANGO2, COMT, DGCR6L, DGCR8, ZDHHC8, TBX1, CLDN5, CLTCL1, SEPTIN5, MRPL40, RANBP1, HIRA, RTN4R, GSC2, UFD1, TXNRD2, DGCR2, CDC45, TRMT2A, SLC25A1, GP1BB, RTL10, ESS2, C22orf39, GNB1L, CCDC188, TSSK2	yes (1)	Male	13	no	yes

Legend: A CNV was considered as recurrent only if it overlapped at more than 40% with a loci previously associated with neuropsychiatric disorders.[1–4] Coordinates are based on Hg19 map of the genome (Homo sapiens (human) genome assembly GRCh37 from Genome Reference Consortium). CNV: Copy number variant; DEL: deletion; DUP: duplication; ASD: autism spectrum disorder; ID: intellectual disability; --: unknown.

TABLE S3. Enrichment of individual neurodevelopmental or neuropsychiatric-associated CNVs in EOP relative to ASD and controls

Cohort (Total N in cohort)	EOP (n=137)		ASD (n=5,540)		Controls (n=16,504)		EOP vs. Controls OR [95%CI] p-value		EOP vs. ASD OR [95%CI] p-value	
Loci	**DEL**	**DUP**	**DEL**	**DUP**	**DEL**	**DUP**	**DEL**	**DUP**	**DEL**	**DUP**
1q21.1	--	3	--	19	--	7	--	**52.60 [8.69-233.00] $p=6\times10^{-5}$**	--	6.50 [1.22-22.47] p=0.01
15q11.2 BP1-BP2	1	--	16	--	70	--	n.s.	--	n.s.	--
16p11.2 proximal	1	1	11	14	5	6	24.23 [0.51-218.14] p=0.05	n.s.	n.s.	n.s.
16p13.11	2	2	7	12	8	31	**30.51 [3.13-155.48] $p=3\times10^{-3}$**	7.87 [0.90-31.39] p=0.03	11.69 [1.17-62.20] p=0.02	6.82 [0.73-31.10] p=0.04
22q11.2 proximal	1	--	1	--	0	--	∞ **[3.09-inf] $p=8\times10^{-3}$**	--	40.54 [0.52-inf] p=0.05	--

Legend: Odds ratios are computed using Fisher's exact test for deletions and duplications. Significant p-values after FDR correction are in bold (≤ 0.009 when comparing EOP to controls and ≤ 0.005 when comparing EOP to ASD). EOP: early-onset psychosis; Controls: unselected population; ASD: autism spectrum disorder; DEL: deletion; DUP: duplication; OR: Odds ratio; 95% CI: 95% confidence intervals; --: not applicable; n.s.: non-significant

TABLE S4. Phenotypes of 1q21 duplication patients in comparison to Rosenfeld, et al. 2012[5]

Feature	Distal (BP3–BP4) duplications (Rosenfeld, et al.)	1440-01	1464-01	1468-01
Short stature	9.5%	-	-	-
Failure to thrive/feeding problems	20%	?	?	?
Microcephaly	8.7%	-	-	-
Macrocephaly	43.5%	-	-	-
Developmental delay/intellectual disability	77.8%	-	+	-
Hypotonia	14.8%	-	-	-
Seizures	17.9%	-	-	-
Autistic features	41.2%	-	-	+
Other behavioral problems	4%	+	+	+
Hearing loss	0%	-	-	-
Brain abnormalities	75%	-	(left temporal slowing on EEG)	-
Dysmorphic features	51.9%	-	-	-
Cataracts	0%	-	-	-
Other ophthalmologic abnormalities	14.8%	-	-	-
Craniosynostosis	0%	-	-	-
Skeletal limb abnormalities	0%	-	-	-
Other skeletal anomalies	7.4%	-	-	-
Clinodactyly	3.7%	-	-	-
Ligamentous laxity	3.7%	-	-	-
Cardiac anomalies	28.6%	-	-	-
Lung abnormalities	0%	-	-	-
Renal anomalies	3.7%	-	-	-
Genital anomalies	11.1%	-	-	-
Blood disorders	0%	-	-	-

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