Supplementary Table 1.

No.	Location (hg18)	Type of CNV	Exons / Introns	Size (Kb)	Sex	Inheritance	Phenotype	Reference
1.	Chr16:74908793-74928800	Del	I2	24	N/D	N/D	Schizophrenia	This study
2.	Chr16:74908793-74928800	Del	I2	20	N/D	N/D	Schizophrenia	This study
3.	Chr16:74919514-74924687	Del	I2	5	N/D	N/D	ADHD	This study
4.	Chr16:74913896-74924687	Del	I2	11	N/D	Maternal	ADHD	This study
5.	Chr16:74919514-74924687	Del	I2	5	N/D	N/D	ASD	This study
6.	Chr16:74913896-74924687	Del	I2	11	N/D	Inherited	ASD; Raven Non-verbal IQ of 88	This study
7. *	Chr16:74997300-75007489	Del	I3	10	Male	Maternal	ASD, above average nonverbal IQ	Pinto *et al.,* 2010
8	Chr16:74323589-75239764	Del	E1-E23	916	Male	Maternal	Autism and mild intellectual disability	This study
9	Chr16:75134230-75235873	Del	E21-E23	191	Male	Maternal	Asperger syndrome	This study
10. **	Chr16:74808505-75067064	Del	E1-E10	259	Male	*de novo*	ASD, Non-verbal IQ of 56	O'Roak *et al.*, 2012
11. ***	Chr16:74487036-75272887	Dup	E1-E23	786	Female	Unknown	Seizure disorder, mood disorder, ADHD, possibly also sleep-related breathing disorder	Mefford *et al.,* 2010
12. ****	Chr16:74558570-75020751	Dup	E1-E3	462	Male	*de novo*	ASD, mild developmental delay, MR	Hanemaaijer *et al.,* 2012

* No 7. is a male patient who harbors both a paternally inherited 78.2 kb exonic duplication on 16p13.2 affecting the ABAT gene as well as a maternally inherited 10.2 kb intronic deletion on 16q23.1 affecting solely the CNTNAP4 gene. The mother who reportedly carries the same CNTNAP4 deletion also suffers from a seizure disorder for which she was treated with valproic acid (VPA) throughout her pregnancy with patient no.7. For patient no.7, the phenotype could be partially/completely explained by the CNTNAP4 deletion, the duplication affecting ABAT, exposure to VPA during pregnancy, or any combination thereof. Further complicating the interpretation of these results is that pt no. 7 is listed as having a sibling with ASD but who does that harbor any CNVs. The patient displayed low average language, no epilepsy; mother treated with valproic acid for epilepsy during pregnancy, fetal tachycardia during labor, forceps delivery at 33 1/2 wks, poor respiratory effort, on ventilation; CT scan: mild cortical atrophy, normal EEGs; neurological exam: right hamstrings, toe walking; dysmorphic features: thin upper lip, small chin, flattened naso-labial fold, flattened occiput, small hands/fingers

** Patient no.10 harbors a *de novo* deletion that besides CNTNAP4, also affects the TERF2IP gene.

*** No. 11 is a pt with childhood-onset seizure disorder as observed clinically and verified on EEG. Seizure phenotype described as generalized tonic-clonic seizures that appear to be related to sleep. Generalized spike and wave on EEG when pt was drowsy/at sleep-wake transition. Seizures reportedly well-controlled on

carbamazepine but the patient opted for lamotrigine because of co-morbid mood disorder. There was also clinical suspicion for ADHD as well as possibly a sleep-related breathing disorder. The patient's father also reportedly suffered from a seizure disorder (grand mal) as well as her paternal great-grandfather (single grand mal seizure) but there were no samples available from either of these two family members for genetic testing (Mefford and Buono, personal communication; see also: http://ccr.coriell.org/Sections/Collections/NINDS/Epilepsy.aspx?Pgld=191&coll=ND).

**** Patient 12 is a male patient was studied by 105k Agilent array (hg18)

- a duplication 10p11.21 of 648kb (38.1-38.8Mb) – paternally inherited
- a duplication 16q23.1 of 462 kb (74.6-75.0 Mb) – *de novo*
  (with partial duplication of CNTNAP4, not other genes in the duplication)
- a deletion 20q13.33 of 483 kb (60.9-61.4 Mb) – *de novo* (including ARFGAP1)

Reason for referral for cytogenetic studies: developmental delay, Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS) versus ASD. IQ testing in Aug 2008 revealed FSIQ of 76 and SON-IQ in November 2008 was 89. The behavioral problems are the most prominent problems in this patient for which he is treated with several psychotropic medications. The patient has no obvious dysmorphic features (van Ravenswaaij-Arts, personal communication).