

Supplementary Figure 1: Manhattan plot for autism genome-wide CNV association analysis. Among 1,068,909 probes, about 105,000 probes were estimated to have CNV frequency ranging between 1% and 5%. Shown in the figure are the $-\log(p)$ for these 105,000 probes, ordered by chromosome and base pair position of the probes. The probe with the strongest p-value was on chromosome 15 but it seemed to be isolated and might be caused by data quality issue. The most promising region is located on chromosome 17. The detailed p-values in the region are in Figure 5.

Supplementary Table 1: Probe-wise type-I error rates for studies based on nuclear families, estimated based 10,000 simulations. α =0.001. Each simulation has 1000 nuclear families. Population frequency of CNV=0.025.

	Ω		Ω1		Ω2	
#sib	CN1 ^a	CN3 ^b	CN1 ^a	CN3 ^b	CN1 ^a	CN3 ^b
1	0.0015	0.0011	0.0015	0.0022	0.0010	0.0016
3	0.0014	0.0012	0.0009	0.0008	0.0014	0.0004
5	0.0010	0.0012	0.0011	0.0012	0.0010	0.0013
1	0.0017	0.0011	0.0009	0.0015	0.0009	0.0013
3	0.0012	0.0008	0.0010	0.0008	0.0008	0.0018
5	0.0015	0.0010	0.0015	0.0017	0.0010	0.0012
1	0.0011	0.0013	0.0009	0.0010	0.0012	0.0011
3	0.0010	0.0014	0.0009	0.0007	0.0013	0.0013
5	0.0011	0.0017	0.0009	0.0006	0.0010	0.0006
1	0.0004	0.0013	0.0012	0.0012	0.0013	0.0004
3	0.0008	0.0013	0.0009	0.0013	0.0010	0.0009
5	0.0012	0.0017	0.0007	0.0009	0.0011	0.0009
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^a deletion with copy number=1;

^b duplication with copy number=3