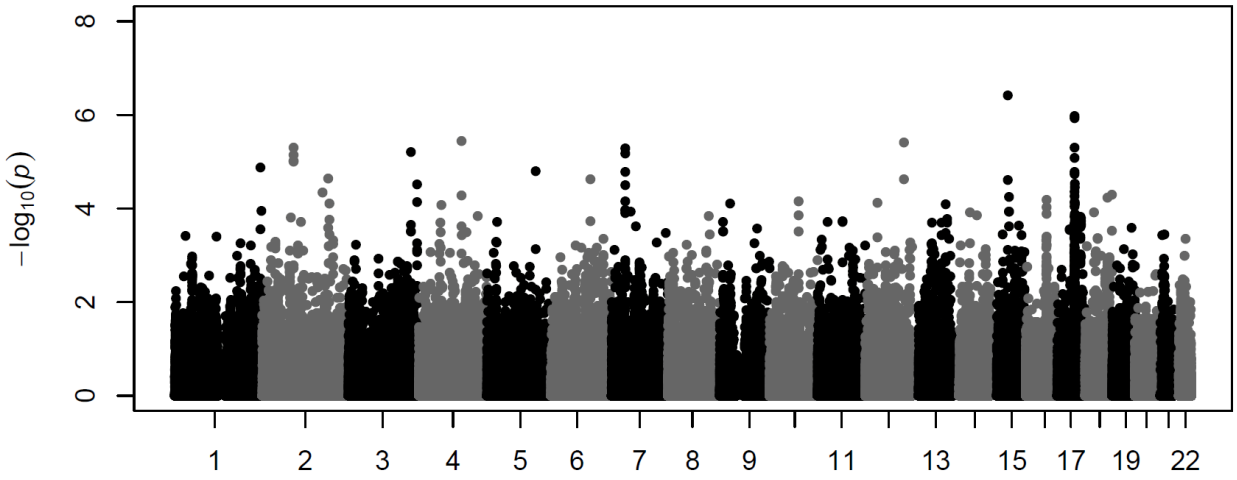

Supplementary Figure 1: Manhattan plot for autism genome-wide CNV association analysis. Among 1,068,909 probes, about 105,000 probes were estimated to have CNV frequency ranging between 1% and 5%. Shown in the figure are the –log(p) for these 105,000 probes, ordered by chromosome and base pair position 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		Ω_1		Ω_2	
#probes	#sib	CN1[a]	CN3[b]	CN1[a]	CN3[b]	CN1[a]	CN3[b]
4	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
5	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
6	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
8	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

[a] deletion with copy number=1;

[b] duplication with copy number=3