A genomewide scan for common alleles affecting risk for autism

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Supplementary Figure 1. Results from ancestry analysis using genome-wide genotype data. Presented are the first and second eigenvectors from the spectral decomposition of the genotype data. Red maps to Europeans versus non-European (blue) ancestry. Letters correspond (roughly) to Northwestern Europe (a), Southern Europe (b), other European ancestry (c), African/African-American (d), Asian (e) and Latino (f).
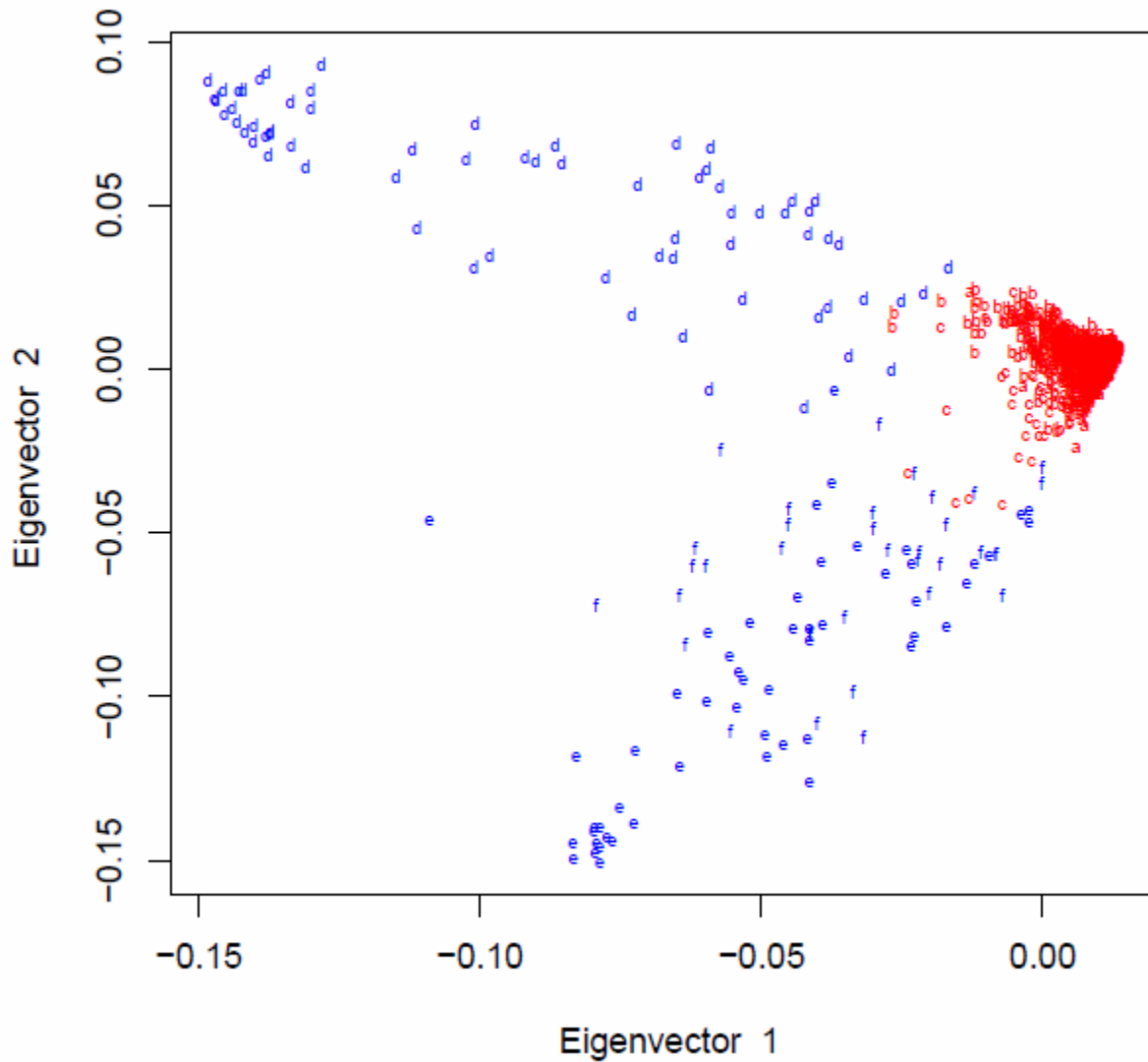
Supplementary Table 1. Quality control (QC) steps for genome-wide association.

QC evaluation	Level for QC	Number with QC failure		Number	
		Families	Individuals	Families	Individuals
Starting count	None	0	0	1558	4712
Ambiguous gender	Family	9	--	-9	-28
Pedigree error	Individual	--	1	0	-1
Proband has probably damaging CNV	Family	80	--	-80	-240
Excess Mendelian errors	Family	19	--	-19	-58
Duplicates	Family	4		-4	-17
Not genotyped	Individual		37		-37
Genotype completion rate	Individual		27	-1	-27
Families without genotyped children	Family	24	--	-24	-40
Families with genotyped child but missing parents	Family	23		-23	-45
Failed to meet diagnostic criteria	Individual	--	34	-29	-92
Final count	None	0	0	1369[1]	4127

[1] The breakdown of families was 1,369 total families; 1,353 nuclear families with one proband, 14 nuclear families with two probands, one family with cousins as probands, and one family with second cousins as probands (estimated from genetic data.)

Supplementary Figure 2. Manhattan plots for primary analyses.
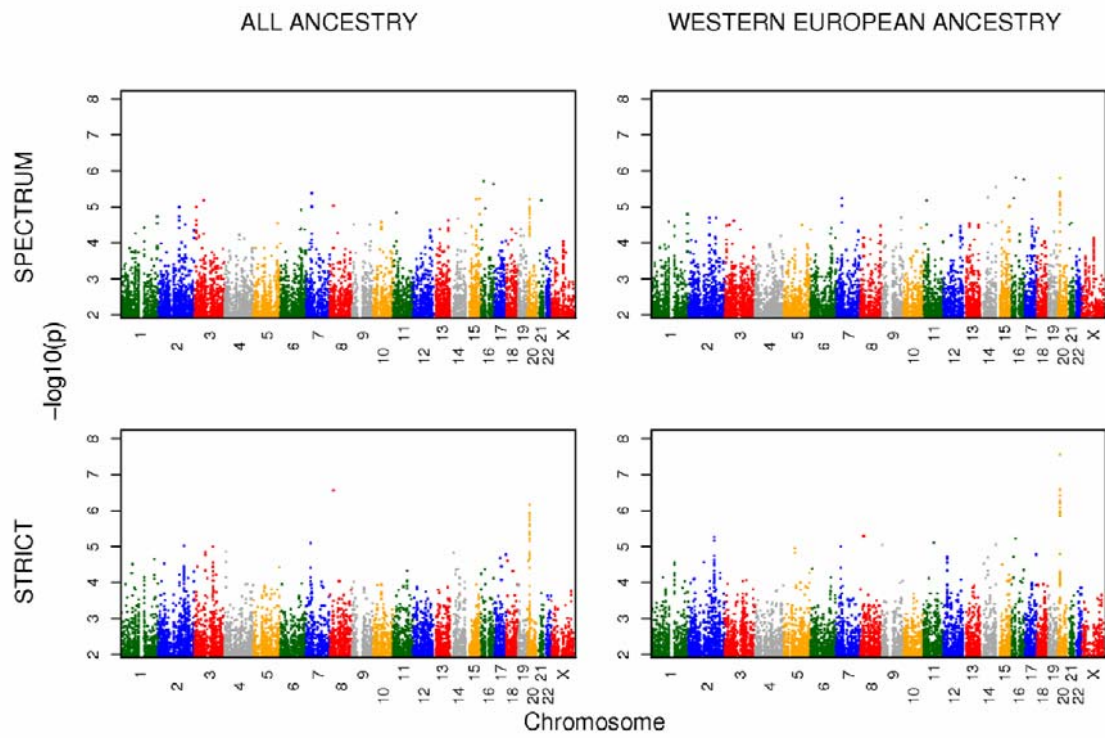
Supplementary Figure 3. QQ plots of genome-wide association data from primary analyses.
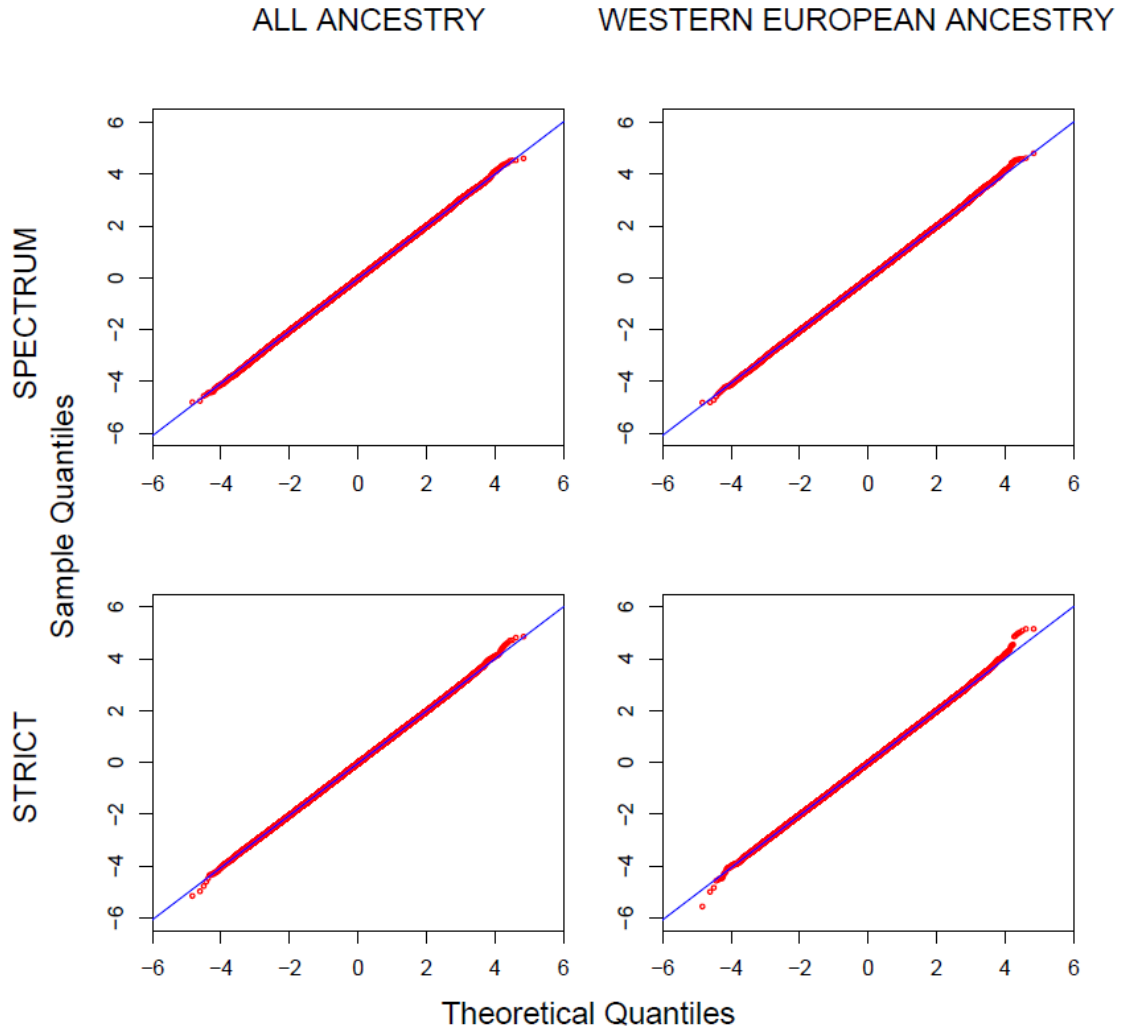
Supplementary Table 2. Summary of top association signals from primary analyses across all primary analytic subgroups. Reporting restricted to SNPs that meet or fall below the association threshold of P<5x10⁻⁶. Spc=spectrum, Str=strict, All=all ancestries, Eur=European ancestries.

SNP	Chr.	Position	Gene	MAF	Allele	Spc|All			Str|All			Spc|Eur			Str|Eur		
						OR	95%CI	P	OR	95%CI	P	OR	95%CI	P	OR	95%CI	P
rs6731562	2	172506507	*HAT1*	0.34	G	1.23	1.08-1.41	6.00E-03	1.51	1.27-1.81	3.30E-05	1.33	1.15-1.54	4.50E-04	1.64	1.35-1.99	4.70E-06
rs10258862	7	39083322	*POU6F2*	0.33	G	1.41	1.23-1.61	3.70E-06	1.43	1.21-1.71	1.80E-04	1.43	1.25-1.65	5.10E-06	1.44	1.20-1.73	3.90E-04
rs6557675	8	23396490	-	0.31	A	0.72	0.63-0.82	8.70E-06	0.61	0.51-0.72	2.20E-07	0.74	0.64-0.85	1.20E-04	0.62	0.52-0.75	4.20E-06
rs4078417	14	50826631	-	0.34	C	1.38	1.21-1.57	1.90E-05	1.50	1.26-1.79	4.10E-05	1.43	1.25-1.64	4.80E-06	1.56	1.29-1.88	1.90E-05
rs7142002	14	101430497	*PPP2R5C*	0.06	G	0.60	0.47-0.77	3.30E-05	0.55	0.39-0.76	2.00E-04	0.54	0.41-0.7 0	1.90E-06	0.45	0.31-0.65	6.30E-06
rs17284809	16	15730300	*MYH11*	0.04	A	0.52	0.39-0.69	1.70E-06	0.53	0.36-0.76	2.60E-04	0.53	0.40-0.70	4.90E-06	0.55	0.37-0.79	7.20E-04
rs205409	16	27971448	*GSG1L*	0.43	G	0.72	0.64-0.82	9.60E-06	0.69	0.58-0.81	1.10E-04	0.69	0.60-0.79	1.10E-06	0.63	0.53-0.75	4.90E-06
rs4150167	16	82771184	*TAF1C*	0.02	A	0.40	0.26-0.59	1.30E-06	0.36	0.20-0.62	4.30E-05	0.38	0.24-0.58	1.00E-06	0.33	0.18-0.59	2.50E-05
rs6079536	20	14644881	*MACROD2*	0.44	A	0.73	0.65-0.83	8.50E-06	0.64	0.54-0.75	1.60E-06	0.72	0.63-0.82	9.50E-06	0.60	0.50-0.72	5.10E-07
rs6079537	20	14645385	*MACROD2*	0.49	G	1.32	1.17-1.49	5.40E-05	1.52	1.29-1.79	6.00E-06	1.38	1.21-1.57	1.40E-05	1.64	1.38-1.96	5.10E-07
rs6079540	20	14653887	*MACROD2*	0.5	A	0.75	0.66-0.84	2.90E-05	0.65	0.55-0.77	3.70E-06	0.73	0.64-0.83	2.20E-05	0.62	0.52-0.73	1.10E-06
rs6079544	20	14662456	*MACROD2*	0.49	A	1.35	1.20-1.53	1.30E-05	1.57	1.33-1.84	1.20E-06	1.41	1.24-1.60	3.60E-06	1.67	1.40-1.99	2.10E-07
rs6074787	20	14671008	*MACROD2*	0.49	A	1.33	1.18-1.50	3.40E-05	1.53	1.30-1.80	4.10E-06	1.38	1.21-1.57	1.30E-05	1.62	1.36-1.93	8.70E-07
rs10446030	20	14675385	*MACROD2*	0.49	G	1.34	1.19-1.52	2.20E-05	1.54	1.30-1.81	3.20E-06	1.39	1.22-1.58	7.40E-06	1.63	1.37-1.95	6.80E-07
rs6079553	20	14680605	*MACROD2*	0.49	A	1.35	1.19-1.52	1.70E-05	1.55	1.31-1.82	2.10E-06	1.40	1.23-1.60	4.10E-06	1.66	1.39-1.97	3.00E-07
rs4814324	20	14690829	*MACROD2*	0.49	A	1.36	1.20-1.54	1.10E-05	1.58	1.34-1.86	9.80E-07	1.41	1.24-1.61	3.90E-06	1.67	1.40-2.00	2.00E-07
rs4141463	20	14695470	***MACROD2***	0.43	A	0.75	0.66-0.85	4.30E-05	0.62	0.52-0.73	5.50E-07	0.71	0.62-0.81	5.80E-06	0.56	0.47-0.67	**2.10E-08**
rs6074798	20	14709709	*MACROD2*	0.48	A	1.38	1.22-1.56	4.80E-06	1.56	1.32-1.84	2.10E-06	1.43	1.26-1.63	1.20E-06	1.65	1.39-1.97	4.40E-07
rs980319	20	14724939	*MACROD2*	0.48	G	1.36	1.20-1.54	1.00E-05	1.52	1.29-1.79	5.10E-06	1.41	1.24-1.61	3.00E-06	1.61	1.35-1.92	1.00E-06

Supplementary Table 3. Summary of top association signals from exploratory analyses across Discovery and Replication data. Reporting restricted to SNPs that meet primary analyses association signal at P<5x10^{-6}.

SNP	Chr	Position	Gene	MAF	Minor	Diagnosis	AGP Discovery			AGRE			Combined		
							OR	95%CI	P	OR	95%CI	P	OR	95%CI	P
rs2196826	1	240612956	**_PLD5_**	0.305	A	Spc|All|Vrb	0.58	0.33-0.69	1.1E-08	0.89	0.43-1.07	2.7E-01	0.71	0.39-0.81	1.1E-06
rs2580237	1	240619604	_PLD5_	0.48	G	Spc|All|Vrb	1.52	1.3-1.78	2.5E-06	1.00	0.83-1.19	9.2E-01	1.26	1.12-1.42	5.2E-04
rs11809951	1	240621154	_PLD5_	0.32	G	Spc|All|Vrb	0.64	0.54-0.75	8.0E-07	0.92	0.76-1.1	3.8E-01	0.75	0.66-0.85	2.6E-05
rs2654862	1	240621187	_PLD5_	0.48	G	Spc|All|Vrb	1.52	1.3-1.78	2.3E-06	1.00	0.83-1.19	9.2E-01	1.26	1.12-1.42	4.9E-04
rs2580222	1	240625569	_PLD5_	0.41	C	Spc|All|Vrb	0.65	0.56-0.76	2.0E-06	0.97	0.81-1.15	7.5E-01	0.78	0.69-0.88	2.5E-04
rs2809985	1	240629344	_PLD5_	0.39	G	Spc|All|Vrb	0.65	0.56-0.77	2.5E-06	0.97	0.82-1.16	8.0E-01	0.78	0.7-0.88	2.6E-04
rs9461347	6	27384557	_POM121L2_	0.04	A	Spc|All|Vrb	2.61	1.71-4.09	4.4E-06	1.48	0.89-2.51	1.1E-01	1.97	1.42-2.74	2.5E-05
rs11155089	6	140220670	_LOC100132735_	0.27	A	Spc|All|Vrb	0.64	0.53-0.75	2.1E-06	0.98	0.8-1.2	8.6E-01	0.77	0.67-0.87	2.0E-04
rs11624358	14	46563202	_MDGA2_	0.19	A	Spc|All|Vrb	0.63	0.53-0.76	4.5E-06	0.94	0.76-1.16	5.6E-01	0.76	0.66-0.87	1.8E-04
rs3784730	15	90788132	**_ST8SIA2_**	0.43	G	Spc|All|Vrb	1.58	1.35-1.86	4.0E-07	1.36	1.14-1.64	1.6E-03	1.47	1.31-1.66	**1.1E-08**
rs2045268	15	90790651	_ST8SIA2_	0.40	A	Spc|All|Vrb	1.56	1.32-1.83	1.5E-06	1.32	0.92-1.89	1.5E-01	1.51	1.3-1.76	1.4E-06
rs4284229	1	38622891	-	0.15	G	Spc|Al|Nvrb	2.12	1.57-2.89	1.1E-06	1.16	0.85-1.58	3.6E-01	1.60	1.29-1.99	2.6E-05
rs16848148	2	212851861	_ERBB4_	0.05	G	Spc|Al|Nvrb	3.60	2-6.87	1.3E-06	0.86	0.52-1.43	5.3E-01	1.65	1.15-2.38	3.7E-03
rs10258862	7	39083323	_POU6F2_	0.33	G	Spc|Al|Nvrb	1.95	1.54-2.49	4.4E-07	0.73	0.56-0.95	2.3E-02	1.23	1.04-1.46	3.0E-02
rs1404999	7	39087990	_POU6F2_	0.33	A	Spc|Al|Nvrb	1.90	1.49-2.42	1.5E-06	0.72	0.55-0.94	1.9E-02	1.20	1.01-1.43	5.2E-02
rs12864778	13	72858013	-	0.46	A	Spc|Al|Nvrb	0.54	0.43-0.68	2.1E-06	1.03	0.79-1.33	8.6E-01	0.72	0.61-0.86	6.9E-04

Supplementary Table 3, continued.

SNP	Chr	Position	Gene	MAF	Minor	Diagnosis	AGP + SAGE			AGP + AGRE + SAGE		
							OR	95%CI	P	OR	95%CI	P
rs2196826	1	240612956	*PLD5*	0.305	A	Spc|All|Vrb	1.37	1.20-1.57	2.5E-06	1.26	1.13-1.39	1.2E-05
rs2580237	1	240619604	*PLD5*	0.48	G	Spc|All|Vrb	1.29	1.14-1.45	3.1E-05	1.16	1.06-1.28	1.9E-03
rs11809951	1	240621154	*PLD5*	0.32	G	Spc|All|Vrb	0.77	0.68-0.88	7.2E-05	0.83	0.75-0.91	1.9E-04
rs2654862	1	240621187	*PLD5*	0.48	G	Spc|All|Vrb	1.28	1.14-1.44	3.8E-05	1.16	1.06-1.28	1.8E-03
rs2580222	1	240625569	*PLD5*	0.41	C	Spc|All|Vrb	0.79	0.70-0.90	1.7E-04	0.86	0.78-0.94	1.2E-03
rs2809985	1	240629344	*PLD5*	0.39	G	Spc|All|Vrb	0.80	0.71-0.90	2.5E-04	0.86	0.78-0.94	1.3E-03
rs9461347	6	27384557	*POM121L2*	0.04	A	Spc|All|Vrb	0.52	0.38-0.72	6.3E-05	0.58	0.44-0.76	8.2E-05
rs11155089	6	140220670	*LOC100132735*	0.27	A	Spc|All|Vrb	1.33	1.16-1.52	2.9E-05	1.21	1.09-1.35	4.5E-04
rs11624358	14	46563202	*MDGA2*	0.19	A	Spc|All|Vrb	1.37	1.17-1.60	6.4E-05	1.24	1.10-1.40	5.9E-04
rs3784730	15	90788132	*ST8SIA2*	0.43	G	Spc|All|Vrb	1.30	1.15-1.47	2.2E-05	1.26	1.14-1.38	2.0E-06
rs2045268	15	90790651	*ST8SIA2*	0.40	A	Spc|All|Vrb	0.78	0.69-0.88	6.3E-05	0.78	0.69-0.88	2.9E-05
rs4284229	1	38622891	-	0.15	G	Spc|Al|Nvrb	1.65	1.31-2.08	2.3E-05	1.51	1.26-1.80	4.8E-06
rs16848148	2	212851861	*ERBB4*	0.05	G	Spc|Al|Nvrb	2.09	1.42-3.08	2.3E-04	1.42	1.05-1.93	2.4E-02
rs10258862	7	39083323	*POU6F2*	0.33	G	Spc|Al|Nvrb	1.54	1.28-1.85	5.1E-06	1.14	0.99-1.31	7.0E-02
rs1404999	7	39087990	*POU6F2*	0.33	A	Spc|Al|Nvrb	0.66	0.55-0.80	1.1E-05	0.89	0.77-1.02	1.0E-01
rs12864778	13	72858013	-	0.46	A	Spc|Al|Nvrb	1.44	1.21-1.71	3.6E-05	1.23	1.08-1.41	2.1E-03

Supplementary Table 4. Summary of top association signals from exploratory analyses across Discovery and Replication data. Reporting restricted to SNPs that meet primary analyses association signal at P<5x10⁻⁶ for parent of origin.

							AGP Discovery			AGRE			Combined		
SNP	Chr	Position	Gene	MAF	Minor	Diagnosis	OR	95%CI	P	OR	95%CI	P	OR	95%CI	P
rs9532931	13	41298079	KIAA0564	0.08	G	Spc|Eur|Mat	0.46	0.34-0.63	1.7E-07	0.63	0.42-0.93	4.0E-03	0.52	0.41-0.67	5.2E-09
rs7329533	13	41456003		0.05	A	Spc|Eur|Mat	0.40	0.26-0.59	9.7E-07	0.88	0.55-1.4	5.1E-01	0.55	0.41-0.74	1.3E-05
rs1475531	20	14815422	MACROD2	0.36	C	Spc|Eur|Mat	1.62	1.33-1.99	7.7E-07	1.02	0.79-1.31	8.8E-01	1.36	1.17-1.6	2.8E-05
rs11155089	6	140220670	LOC100132735	0.27	A	Spc|Eur|Pat	0.62	0.51-0.76	2.3E-06	1.05	0.81-1.37	6.5E-01	0.75	0.64-0.89	2.1E-04
rs10258862	7	39083323	POU6F2	0.33	G	Spc|Eur|Pat	1.63	1.33-2.01	1.3E-06	0.79	0.61-1.02	3.0E-02	1.23	1.05-1.44	6.5E-03
rs1404999	7	39087990	POU6F2	0.33	A	Spc|Eur|Pat	1.62	1.32-2	1.8E-06	0.79	0.61-1.02	3.6E-02	1.23	1.05-1.44	6.5E-03
rs205409	16	27971449	GSG1L	0.43	G	Spc|Eur|Pat	0.64	0.53-0.77	1.6E-06	1.23	0.98-1.56	3.5E-02	0.83	0.72-0.96	7.6E-03
rs9348512	6	10564692	-	0.33	A	Str|Eur|Mat	0.55	0.42-0.71	2.6E-06	0.84	0.62-1.15	2.1E-01	0.65	0.53-0.79	4.3E-06
rs9532931	13	41298079	KIAA0564	0.08	G	Str|Eur|Mat	0.39	0.26-0.57	3.1E-07	0.57	0.35-0.93	5.1E-03	0.45	0.33-0.62	1.3E-08
rs6074798	20	14709710	MACROD2	0.48	A	Str|Eur|Mat	1.79	1.38-2.34	4.5E-06	0.99	0.73-1.33	9.3E-01	1.42	1.17-1.73	1.4E-04
rs1475531	20	14815422	MACROD2	0.36	C	Str|Eur|Mat	1.84	1.41-2.4	1.9E-06	1.18	0.85-1.63	2.4E-01	1.54	1.25-1.89	7.2E-06
rs4836231	5	122124002	-	0.36	G	Str|Eur|Pat	0.55	0.42-0.71	2.7E-06	0.86	0.63-1.17	2.7E-01	0.66	0.54-0.81	1.4E-05
rs28603112	5	122126587	-	0.36	C	Str|Eur|Pat	0.55	0.42-0.71	2.7E-06	0.86	0.63-1.17	2.7E-01	0.66	0.54-0.81	1.4E-05
rs1363614	5	122176254	SNX2	0.36	G	Str|Eur|Pat	0.55	0.42-0.72	4.5E-06	0.86	0.63-1.17	2.7E-01	0.67	0.55-0.82	2.3E-05
rs8065403	17	31487436	-	0.17	A	Str|Eur|Pat	0.50	0.36-0.68	3.6E-06	0.72	0.5-1.02	2.5E-02	0.59	0.46-0.74	1.0E-06
rs4141463	20	14695471	MACROD2	0.43	A	Str|Eur|Pat	0.54	0.42-0.7	8.6E-07	0.94	0.69-1.27	6.2E-01	0.67	0.55-0.82	1.4E-05
rs11675023	2	140382810	-	0.20	G	Spc|All|Mat	0.61	0.49-0.75	2.2E-06	1.02	0.81-1.29	8.4E-01	0.78	0.66-0.91	7.0E-04
rs11677529	2	140406237	-	0.20	A	Spc|All|Mat	0.61	0.49-0.76	3.3E-06	1.02	0.81-1.29	8.4E-01	0.78	0.67-0.92	9.1E-04
rs1475531	20	14815422	*MACROD2*	0.36	C	Spc|All|Mat	1.59	1.32-1.91	4.9E-07	0.99	0.8-1.23	9.5E-01	1.31	1.14-1.51	4.7E-05
rs3817118	4	77418235	*LOC100129583*	0.10	C	Spc|All|Pat	2.02	1.48-2.78	3.2E-06	1.03	0.77-1.37	8.0E-01	1.38	1.12-1.7	7.9E-04
rs10258862	7	39083323	*POU6F2*	0.33	G	Spc|All|Pat	1.56	1.28-1.89	3.0E-06	0.78	0.63-0.97	9.5E-03	1.15	1-1.33	4.0E-02
rs7297890	12	115694448	*TMEM118*	0.10	G	Spc|All|Pat	0.54	0.41-0.71	4.1E-06	0.87	0.65-1.16	2.5E-01	0.69	0.56-0.84	3.2E-05
rs10006242	4	176783314	-	0.40	A	Str|All|Mat	1.73	1.36-2.22	3.0E-06	1.02	0.79-1.32	8.3E-01	1.36	1.14-1.62	2.4E-04
rs9348512	6	10564692	-	0.33	A	Str|All|Mat	0.57	0.45-0.74	4.3E-06	0.87	0.67-1.14	2.4E-01	0.70	0.58-0.84	2.9E-05
rs6557675	8	23396491	-	0.31	A	Str|All|Mat	0.54	0.41-0.69	5.2E-07	0.99	0.76-1.29	9.4E-01	0.72	0.6-0.87	1.3E-04
rs10507103	12	97887773	*ANKS1B*	0.17	G	Str|All|Mat	2.02	1.47-2.8	4.7E-06	1.27	0.9-1.81	1.0E-01	1.61	1.27-2.04	1.3E-05
rs9532931	13	41298079	*KIAA0564*	0.08	G	Str|All|Mat	0.45	0.31-0.65	5.0E-06	0.58	0.38-0.87	1.2E-03	0.50	0.38-0.66	**2.6E-08**
rs4807188	19	1926217	*CSNK1G2*	0.02	A	Str|All|Mat	12.00	2.98-104.76	2.8E-06	2.33	0.53-13.98	1.5E-01	5.80	2.22-19.2	8.3E-06
rs6077999	20	10923285	-	0.30	G	Str|All|Mat	0.57	0.44-0.73	3.6E-06	0.84	0.64-1.09	1.3E-01	0.68	0.56-0.81	7.7E-06
rs1475531	20	14815422	*MACROD2*	0.36	C	Str|All|Mat	1.77	1.39-2.26	1.6E-06	1.11	0.84-1.46	4.0E-01	1.44	1.2-1.73	2.0E-05
rs10006242	4	176783314	-	0.40	A	Str|All|Mat	1.73	1.36-2.22	3.0E-06	1.02	0.79-1.32	8.3E-01	1.36	1.14-1.62	2.4E-04
rs4836231	5	122124002	-	0.36	G	Str|All|Pat	0.56	0.44-0.73	3.2E-06	0.91	0.71-1.18	4.3E-01	0.71	0.59-0.85	5.6E-05
rs28603112	5	122126587	-	0.36	C	Str|All|Pat	0.56	0.44-0.73	3.2E-06	0.91	0.71-1.18	4.3E-01	0.71	0.59-0.85	5.6E-05
rs2125903	17	45890913	*ACSF2*	0.26	A	Str|All|Pat	1.76	1.37-2.27	4.9E-06	1.23	0.95-1.6	6.1E-02	1.46	1.21-1.75	1.1E-05

Supplementary Table 5. Candidate genes identified from previous association studies. Results from candidate genes culled from the literature, selected because they show evidence for association from GWA studies or from multiple candidate gene studies [z-score (*P*-value)]. Presented is the SNP within each gene showing the strongest association (smallest p-value) for each of the four primary analyses. Negative z-score indicates the minor allele is under-transmitted.

Gene	SNP	MAF	Spc|All	Str|All	Spc|Eur	Str|Eur
SLC6A4	rs11657536	0.024	-1.69 (0.095)	-2.32 (0.020)	-1.92 (0.055)	-2.63 (0.011)
AVPR1A	rs11174810	0.027	-0.56 (0.578)	-1.53 (0.125)	-0.89 (0.376)	-0.89 (0.371)
EN2	rs1861958	0.122	0.49 (0.624)	0.86 (0.388)	0.29 (0.769)	0.40 (0.689)
RELN	rs39334	0.373	-3.897 ($9.7 \cdot 10^{-5}$)	-2.34 (0.019)	-3.61 ($3.1 \cdot 10^{-4}$)	-1.57 (0.116)
SLC25A12	rs908670	0.288	4.107 ($4.0 \cdot 10^{-5}$)	2.34 (0.019)	4.15 ($3.4 \cdot 10^{-5}$)	2.53 (0.011)
MET	rs2237712	0.047	-2.35 (0.019)	-2.55 (0.011)	-1.79 (0.074)	-2.17 (0.030)
GABRB3	rs7172653	0.45	2.37 (0.018)	1.00 (0.316)	2.48 (0.013)	0.86 (0.393)
OXTR	rs2301261	0.103	-2.42 (0.016)	-1.54 (0.124)	-2.69 (0.007)	-1.78 (0.075)
ITGB3	rs2056131	0.305	-2.07 (0.039)	-1.71 (0.087)	-2.08 (0.038)	-2.02 (0.043)
CNTNAP2	rs929353	0.321	2.31 (0.021)	2.84 (0.004)	3.09 (0.002)	4.07 ($4.6 \cdot 10^{-5}$)
GRIK2	rs17828670	0.109	-1.25 (0.211)	-2.46 (0.014)	-1.25 (0.210)	-2.54 (0.011)
CDH9	rs12652586	0.165	-1.22 (0.223)	-0.04 (0.970)	-1.46 (0.146)	-0.19 (0.848)
CDH10	rs16893429	0.03	-2.59 (0.012)	-1.53 (0.127)	-2.43 (0.015)	-0.95 (0.344)
SEMA5A	rs415024	0.285	-2.33 (0.020)	-3.04 (0.002)	-2.12 (0.034)	-3.10 (0.002)
GABRA4	rs1512130	0.358	2.33 (0.020)	2.93 (0.003)	2.19 (0.029)	2.57 (0.010)
GABRB1	rs4694846	0.423	3.21 (0.001)	1.35 (0.177)	3.35 ($8.1 \cdot 10^{-4}$)	2.00 (0.045)
ITGA4	*rs1449259*	*0.109*	*2.43 (0.015)*	*1.70 (0.090)*	*2.81 (0.005)*	2.08 (0.038)

Table 6a. Family-based association results by SNP and sites contributing samples for comparison to Wang et al. (PMID: 19404256). SNPs presented were found to be associated with risk for autism in the previous study. Cells contain z-score, *P*-value, relative allele frequency. For column 3, N is sample size in trios, P is *P*-value, Z is the z-statistics for biased transmission and f is the allele frequency for a selected allele. Fst is Wright's measure of differentiation computed across the sites.

SNP	F_{st}		ALL	AGRE[1]	IMGSAC	Miami[1]	Canagen	Paris	CPEA[1]	Ireland	Portugal
		N	1217	53	224	108	290	118	63	131	213
rs7704904	0.0019	P	0.953	0.668	0.444	0.178	0.724	0.472	0.793	0.927	0.254
		Z	0.059	-0.429	-0.765	1.347	-0.354	-0.718	0.263	0.092	1.140
		f	0.640	0.648	0.624	0.636	0.630	0.619	0.698	0.649	0.659
rs1896731	0.0051	P	0.458	0.354	0.315	0.026	0.106	0.567	0.310	0.843	0.421
		Z	0.742	0.926	1.001	-2.223	1.616	0.572	1.016	-0.198	-0.804
		f	0.637	0.630	0.677	0.603	0.637	0.673	0.611	0.660	0.587
rs10038113	0.0047	P	0.460	0.631	0.362	0.010	0.141	0.787	0.352	0.795	0.889
		Z	0.739	0.480	0.911	-2.578	1.471	0.271	0.930	0.260	-0.140
		f	0.582	0.571	0.621	0.559	0.585	0.627	0.547	0.583	0.534
rs12518194	0.0022	P	0.929	0.777	0.586	0.110	0.637	0.660	0.793	0.710	0.639
		Z	0.089	-0.283	-0.544	1.600	-0.471	-0.440	0.263	0.371	0.469
		f	0.646	0.651	0.624	0.651	0.635	0.627	0.705	0.652	0.669
rs4307059	0.0020	P	0.739	0.777	0.779	0.131	0.760	0.531	0.500	0.925	0.314
		Z	0.333	-0.283	-0.280	1.508	-0.306	-0.626	0.674	-0.094	1.006
		f	0.648	0.652	0.627	0.652	0.637	0.632	0.707	0.653	0.669
rs4327572	0.0020	P	1.000	0.668	0.494	0.113	0.603	0.660	0.896	0.859	0.440
		Z	0.000	0.429	0.684	-1.584	0.520	0.440	-0.130	-0.178	-0.772
		f	0.355	0.352	0.376	0.349	0.368	0.371	0.302	0.350	0.330

1) Families from these sites overlap somewhat with those in Wang et al. (111 trios) Note that 943 families from the AGRE repository were genotyped in Wang et al, but only a small subset of those families are contained in the data reported here. The most substantial overlap is for the Miami families: 108 families were reported on here, whereas 487 families from the Miami group were analyzed in (Wang PMID: 19404256).

Table 6b. Family-based association results by sites contributing samples for comparison to Weiss et al. (PMID: 19812673). The SNP in the table is in perfect or almost perfect linkage disequilibrium with the SNP P is *P*-value, Z is the z-statistics for biased transmission and f is the allele frequency for a selected allele. Fst is Wright's measure of differentiation computed across the sites.

SNP	F_{st}		ALL	AGRE[2]	IMGSAC	Miami	Canagen	Paris	CPEA	Ireland	Portugal
		N	1217	53	224	108	290	118	63	131	213
rs2234235	0.0014	P	0.160	0.480	0.433	0.297	0.020	0.491	0.705	0.763	1.000
		Z	1.406	0.707	0.784	-1.043	2.333	0.688	0.378	0.302	0.000
		f	0.034	0.035	0.035	0.042	0.031	0.040	0.023	0.022	0.042

2) Families from this site overlap somewhat with those in Weiss et al. (106 trios) (Weiss PMID: 19812673).

Supplementary Figure 4. Power curves by sample size (N, panels), allele frequency and allelic odds ratio (lines). Power was calculated for the binary trait (ASD, yes/no) under the assumption that the risk locus is the genotyped SNP, a log additive model, the population prevalence is 0.005, and the rejection region is $P = 5 \cdot 10^{-8}$. The colored lines map to different allelic odds ratios: black = 1.2, grey = 1.4, red = 1.6, blue = 1.8 and green = 2.0.