Data Supplement

Levinson et al., Genome-wide Association Study of Multiplex Schizophrenia Pedigrees

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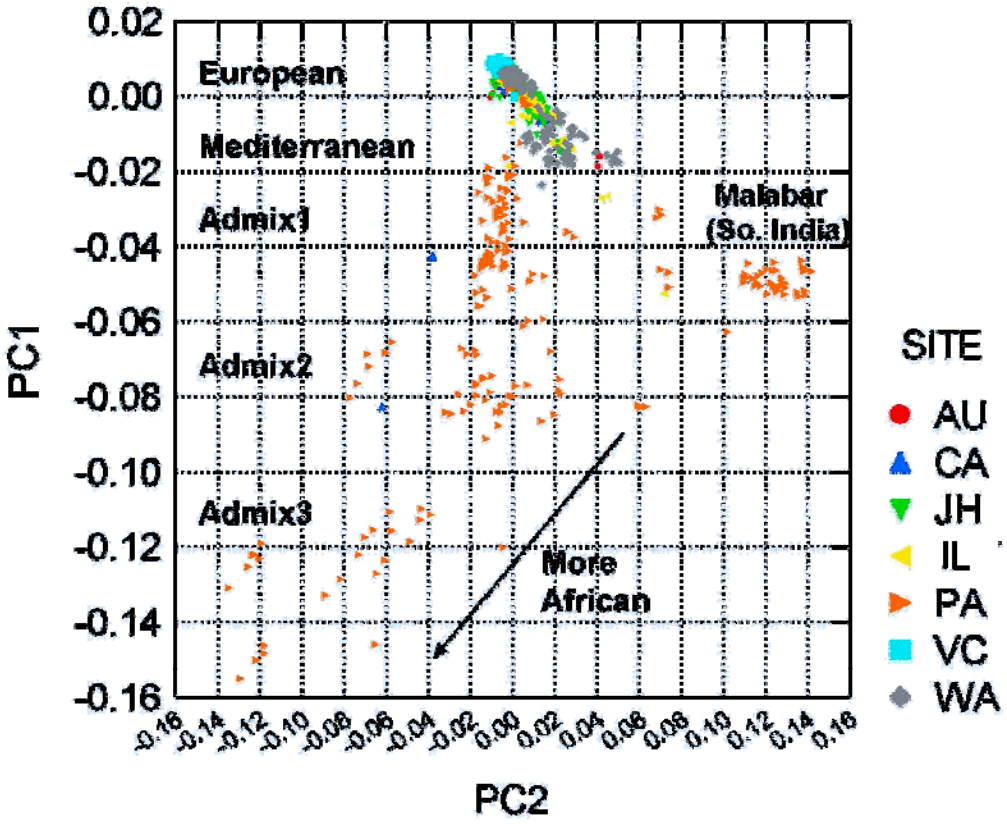
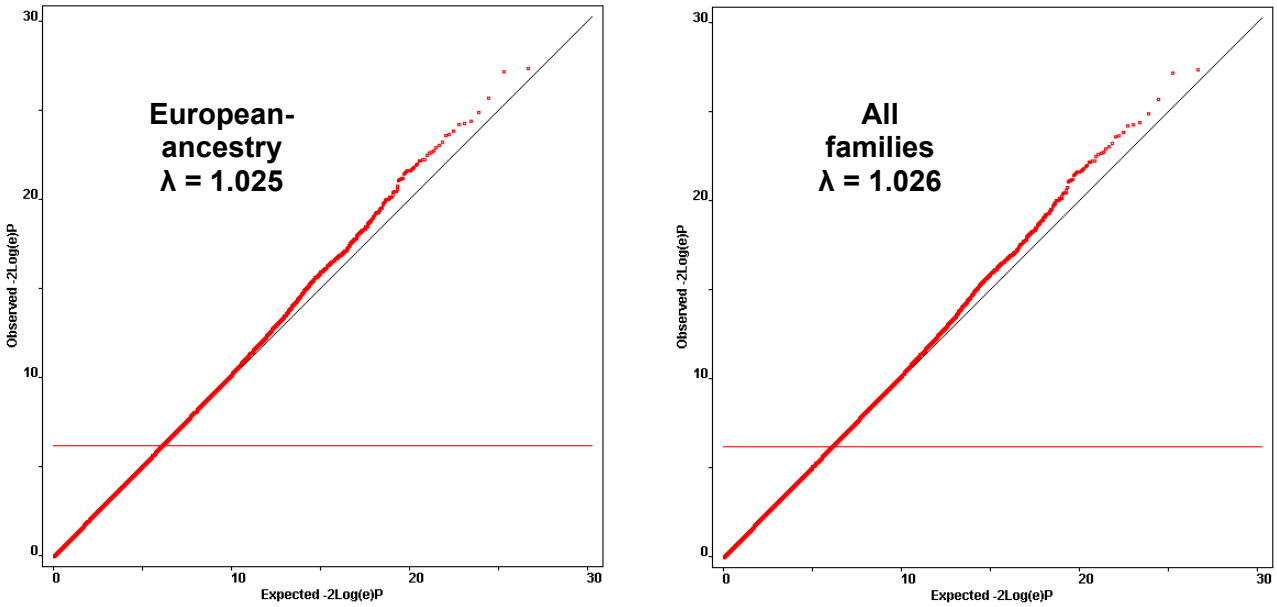
Figure S1: Ancestry groups defined by Principal Components Analysis



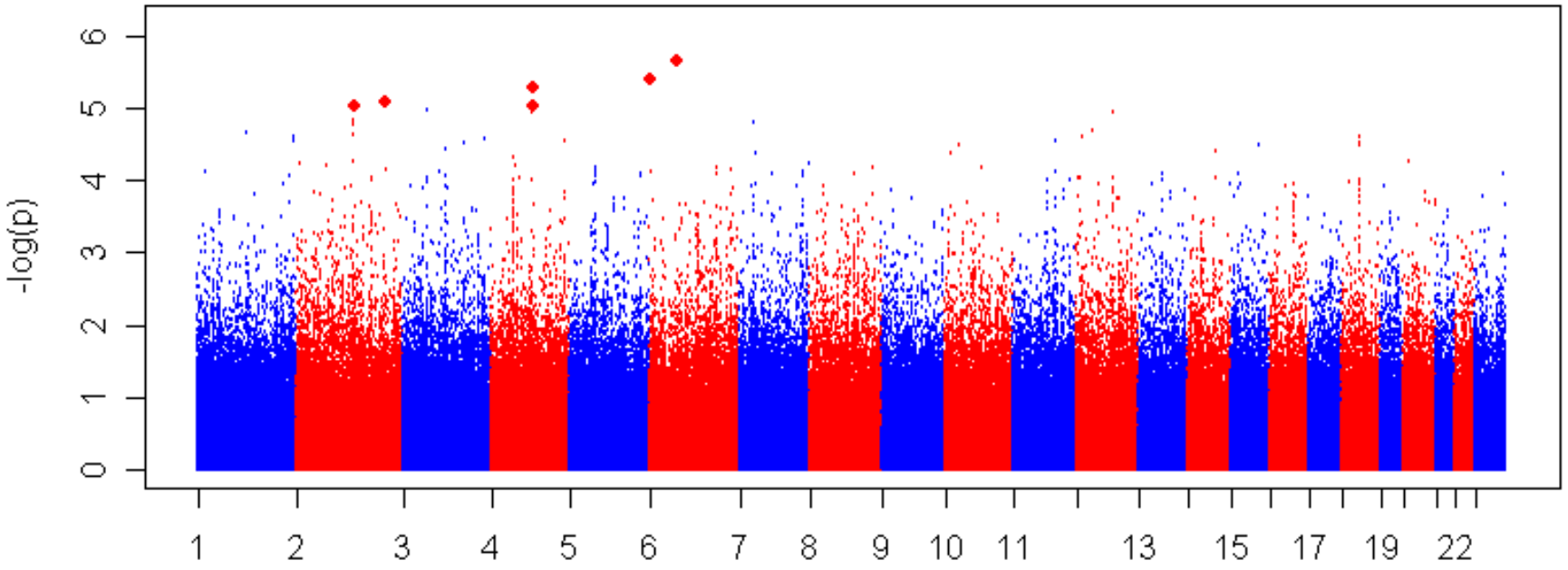
Each symbol represents one subject from the family-based GWAS (see main text, Table 1), plotted according to the first two Principal Component scores from an EIGENSTRAT analysis (1) of 55,010 autosomal SNPs with low pairwise linkage disequilibrium. Six ancestry groups were formed as shown. Each ancestry group was analyzed separately, so that the allele frequencies used by TRANSMIT or UNPHASED (in addition to the family's data) to estimate non-transmitted parental alleles would reflect the appropriate ancestral background. The primary SNP analysis included only European-ancestry families, while the all-family analysis included all six groups (with expected and observed counts of transmitted alleles combined across groups).

Figure S2: Q-Q Plots for European-ancestry (EUR) and ALL families



Shown are quantile-quantile plots for whole-genome SNP analyses of European-ancestry and of all families, generated by WGAViewer (http://people.chgv.lsrc.duke.edu/~dg48//WGAViewer/download.php) (2). Lambda values were computed by dividing the median chi-square value for all SNPs (544,131 for European-ancestry, 541,499 for all families) by the expected value of the chi-square distribution with 1 df under the null hypothesis. 95% of values lie below the red horizontal lines.

Figure S3: GWAS results for all 631 families



Each dot represents the -log(P) value of one SNP in the GWAS analysis of all ancestries. P-values < 10^{-5} are represented by large diamonds. Chromosomes are shown on the X axis.

Table S1: All SNPs with P < 0.0001 (European-ancestry families)

SNP	LOC	Allele 1 total counts				Allele 2 total counts				T from het parents		OR	P-value	Genes(+-50kb)
		A1	Frq	T	NT	A2	Frq	T	NT	A1	A2			
rs10429924	chr1:242457187	T	0.071	130	213	C	0.929	2208	2125	118.2	190.7	0.620	1.3E-06	
rs3847375	chr10:21607571	A	0.143	282	389	C	0.857	2056	1949	233.9	340.4	0.687	1.4E-06	
rs12210050	chr6:420489	T	0.227	565	459	C	0.773	1763	1869	445.1	371.7	1.197	2.9E-06	EXOC2,9648
rs12426725	chr12:80367259	A	0.152	308	411	G	0.848	2030	1927	254.1	347.9	0.731	4.2E-06	PPFIA2,within
rs16869652	chr6:33959151	A	0.088	239	163	G	0.912	2099	2175	221.1	152.6	1.448	5.4E-06	
rs1170612	chr2:124699526	T	0.225	593	481	C	0.775	1745	1857	475.1	339.3	1.400	5.9E-06	CNTNAP5,within
rs2726807	chr4:183374392	T	0.350	887	761	C	0.650	1451	1577	599.9	464.5	1.291	5.9E-06	
rs16934812	chr12:29763585	G	0.126	331	248	T	0.874	2003	2086	294.0	219.6	1.339	7.2E-06	TMTC1,within
rs1170611	chr2:124698947	G	0.227	597	486	T	0.773	1741	1852	476.6	343.8	1.386	8.0E-06	CNTNAP5,within
rs2048485	chr18:40344665	C	0.145	291	390	T	0.855	2045	1946	241.8	337.8	0.716	8.2E-06	
rs11082399	chr18:40364087	G	0.163	334	435	A	0.837	2004	1903	271.5	367.8	0.738	9.7E-06	
rs12545451	chr8:16218065	C	0.041	70	132	T	0.959	2268	2206	66.1	116.1	0.570	1.1E-05	
rs9759759	chr4:101792711	C	0.130	263	359	A	0.870	2075	1979	223.6	304.6	0.734	1.1E-05	
rs906600	chr4:101815546	A	0.129	261	356	C	0.871	2077	1982	222.1	303.0	0.733	1.3E-05	
rs17011040	chr2:124660578	T	0.222	587	479	G	0.778	1745	1853	472.2	333.0	1.418	1.3E-05	CNTNAP5,within
rs1921044	chr12:80240357	C	0.239	509	622	A	0.761	1829	1716	375.1	476.3	0.788	1.3E-05	PPFIA2,within
rs12511372	chr4:45811189	G	0.500	1229	1106	A	0.500	1107	1230	643.8	524.2	1.228	1.4E-05	GABRG1,within, and GABRA2 in cluster
rs7662743	chr4:45871585	A	0.499	1226	1103	G	0.501	1112	1235	644.5	524.5	1.229	1.6E-05	
rs3197999	chr3:49696536	T	0.298	747	628	C	0.702	1587	1706	539.6	437.1	1.235	1.6E-05	BSN,12550; APEH,598; MST1,within; RNF123,-5457; AMIGO3,33432; GMPPB,37399; IHPK1,40195
rs1529299	chr2:124658849	C	0.222	587	480	A	0.778	1751	1858	471.6	336.4	1.402	1.6E-05	CNTNAP5,within
rs13093713	chr3:140959282	G	0.334	809	701	A	0.666	1525	1633	549.2	488.6	1.124	1.6E-05	
rs10927166	chr1:242449321	G	0.146	299	402	A	0.854	2037	1934	249.4	332.0	0.751	1.8E-05	
rs2216959	chr2:204892416	G	0.186	491	389	A	0.814	1847	1949	410.5	296.0	1.387	1.9E-05	
rs954794	chr18:40335923	T	0.138	279	372	C	0.862	2059	1966	234.3	322.9	0.726	2.0E-05	
rs4716801	chr7:157381124	G	0.462	1151	1022	A	0.538	1185	1314	652.2	509.0	1.281	2.1E-05	PTPRN2,within
rs11902121	chr2:124665055	G	0.220	580	475	A	0.780	1754	1859	466.9	334.6	1.395	2.1E-05	CNTNAP5,within
rs1357262	chr12:80372931	A	0.217	458	565	G	0.783	1876	1769	347.7	446.6	0.779	2.2E-05	PPFIA2,within
rs7805806	chr7:20693853	G	0.123	334	250	A	0.877	2004	2088	298.9	203.8	1.467	2.2E-05	ABCB5,within
rs12239401	chr1:235261146	T	0.443	953	1080	C	0.557	1383	1256	495.2	657.5	0.753	2.2E-05	RYR2,-11178
rs6433323	chr2:172581306	G	0.381	963	842	A	0.619	1375	1496	623.6	479.2	1.301	2.3E-05	HAT1,24460; MAP1D,within

rs10502829	chr18:40289840	A	0.118	234	322	C	0.882	2104	2016	201.5	284.5	0.708	2.3E-05	
rs7653963	chr4:101757162	A	0.123	250	341	C	0.877	2088	1997	214.6	290.0	0.740	2.4E-05	
rs1397988	chr18:40276800	T	0.117	233	320	G	0.883	2103	2016	200.8	283.3	0.709	2.7E-05	
rs1000933	chr12:80342954	C	0.241	513	622	T	0.759	1825	1716	377.3	477.7	0.790	2.7E-05	PPFIA2,within
rs10784514	chr12:64739146	C	0.327	830	722	T	0.673	1508	1616	579.8	449.4	1.290	2.8E-05	
rs12606712	chr18:40225197	A	0.114	224	310	G	0.886	2102	2016	193.7	276.5	0.700	2.8E-05	
rs7305426	chr12:12047589	A	0.442	1088	970	C	0.558	1248	1366	632.1	520.0	1.216	2.8E-05	
rs1037231	chr3:85845797	A	0.417	913	1038	G	0.583	1421	1296	506.2	629.0	0.805	3.3E-05	CADM2,-12524
rs3892156	chr16:48877496	A	0.253	659	552	G	0.747	1669	1776	509.5	371.3	1.372	3.7E-05	ADCY7,-1827; BRD7,32945
rs2396465	chr2:228234344	G	0.098	267	198	A	0.902	2071	2140	244.6	168.1	1.456	3.8E-05	DKFZp547H025,-28212; SLC19A3,23825
rs6749984	chr2:124156863	G	0.058	99	161	A	0.942	2223	2161	91.3	160.5	0.569	3.9E-05	
rs6549051	chr3:85823124	C	0.415	911	1035	T	0.585	1427	1303	507.7	627.8	0.809	3.9E-05	CADM2,-35197
rs1881302	chr12:39267898	G	0.084	230	168	T	0.916	2108	2170	213.5	146.0	1.463	4.0E-05	
rs534459	chr4:45951562	T	0.431	1075	957	C	0.569	1261	1379	641.8	503.7	1.274	4.3E-05	GABRA2,within
rs572227	chr4:45946150	A	0.425	1059	942	G	0.575	1277	1394	637.4	504.3	1.264	4.5E-05	GABRA2,188
rs12565770	chr1:19427647	A	0.116	229	312	G	0.884	2109	2026	197.6	281.8	0.701	4.5E-05	UBR4,-18314; KIAA0090,within; MRTO4,-23014; AFAR3,37415
rs294580	chr5:162946628	T	0.331	837	719	C	0.669	1501	1619	581.0	454.2	1.279	4.5E-05	
rs488447	chr4:45887802	G	0.422	1050	936	A	0.578	1286	1400	634.9	504.4	1.259	4.6E-05	
rs11899935	chr2:124651419	T	0.223	587	485	C	0.777	1751	1853	470.8	339.1	1.389	4.8E-05	CNTNAP5,within
rs1851185	chr2:212235974	T	0.236	593	490	C	0.764	1745	1848	462.8	380.3	1.217	4.8E-05	ERBB4,within
rs522636	chr4:45922604	C	0.425	1059	943	T	0.575	1279	1395	636.9	505.7	1.259	4.8E-05	GABRA2,23734
rs1456614	chr18:40160422	T	0.144	379	298	C	0.856	1959	2040	330.8	244.1	1.355	4.8E-05	
rs12321966	chr12:8592432	T	0.094	264	196	G	0.906	2072	2140	243.5	153.6	1.585	5.1E-05	CLEC4D,26205; CLEC4E,-7607
rs1540416	chr8:16256536	G	0.052	93	156	A	0.948	2245	2182	86.7	142.5	0.609	5.3E-05	
rs4805453	chr19:34814743	C	0.421	1050	927	T	0.579	1286	1409	635.7	503.2	1.263	5.3E-05	POP4,16196; PLEKHF1,-33423
rs534787	chr4:45927453	C	0.425	1059	943	T	0.575	1279	1395	636.7	506.0	1.258	5.4E-05	GABRA2,18885
rs6901207	chr6:3798905	G	0.443	1110	995	A	0.557	1228	1343	650.3	503.8	1.291	5.6E-05	FAM50B,2355
rs6443997	chr3:186016225	A	0.058	106	156	G	0.942	2232	2182	98.0	159.2	0.616	6.1E-05	VPS8,within
rs8093196	chr18:40367268	G	0.316	678	782	T	0.684	1660	1556	444.5	566.3	0.785	6.1E-05	
rs795955	chr12:77160181	T	0.403	883	993	C	0.597	1455	1345	503.7	621.1	0.811	6.2E-05	NAV3,29260
rs10507070	chr12:94873188	A	0.172	451	359	G	0.828	1885	1977	381.6	285.1	1.338	6.3E-05	CCDC38,-12629; AMDHD1,within; HAL,18084; LTA4H,45553
rs7179849	chr15:22589304	T	0.182	383	475	C	0.818	1955	1863	305.7	389.8	0.784	6.6E-05	SNRPN,-30582
rs1432655	chr5:162961360	T	0.473	1040	1163	C	0.527	1298	1175	515.9	649.9	0.794	6.7E-05	
rs7612090	chr3:85812192	A	0.426	941	1059	G	0.574	1397	1279	517.5	625.7	0.827	6.9E-05	CADM2,-46129
rs9883807	chr3:85820336	A	0.426	941	1059	C	0.574	1397	1279	517.5	625.6	0.827	7.0E-05	CADM2,-37985
rs1391168	chr4:45809916	G	0.480	1065	1178	A	0.520	1267	1154	527.7	636.4	0.829	7.0E-05	GABRG1,within
rs502038	chr4:45975075	G	0.426	1064	950	T	0.574	1274	1388	640.3	502.9	1.273	7.0E-05	GABRA2,within
rs6927764	chr6:133518471	G	0.206	522	433	A	0.794	1816	1905	422.6	342.9	1.232	7.1E-05	

rs7180015	chr15:85305969	G	0.087	162	226	A	0.913	2176	2112	144.4	225.8	0.640	7.1E-05	AGBL1,within
rs4695140	chr4:45670100	C	0.399	1003	890	T	0.601	1335	1448	630.4	491.1	1.284	7.2E-05	
rs2249112	chr4:183378729	T	0.301	757	649	C	0.699	1579	1687	545.7	436.7	1.250	7.6E-05	
rs1782	chr6:90124434	C	0.116	311	240	T	0.884	2017	2088	279.7	197.7	1.415	7.7E-05	GABRR2,-42748; UBE2J1,-5096; RRAGD,9878
rs1598665	chr18:40183207	T	0.144	378	298	A	0.856	1960	2040	329.7	246.1	1.340	7.7E-05	
rs2362643	chr16:68503033	G	0.318	815	701	A	0.682	1523	1637	578.2	436.4	1.325	7.8E-05	WWP2,within; LOC348174,-39277
rs6718013	chr2:172577431	A	0.442	1101	984	G	0.558	1237	1354	643.4	510.1	1.261	8.2E-05	HAT1,20585; MAP1D,within
rs2211871	chr21:38744520	G	0.092	242	179	T	0.908	2094	2157	222.4	166.6	1.335	8.3E-05	ERG,within
rs7673537	chr4:45883535	T	0.509	1245	1134	C	0.491	1093	1204	639.5	529.1	1.209	8.4E-05	
rs2890851	chr15:35347654	G	0.399	986	877	A	0.601	1352	1461	614.6	506.2	1.214	8.5E-05	
rs1008927	chr21:38746043	C	0.091	238	175	A	0.909	2090	2153	218.9	164.4	1.331	8.6E-05	ERG,within
rs4925449	chr22:47486086	A	0.084	170	237	G	0.916	2166	2099	153.4	207.6	0.739	8.8E-05	FAM19A5,within
rs1591956	chr13:65207677	A	0.415	1021	908	G	0.585	1317	1430	617.9	517.5	1.194	8.8E-05	
rs175	chr7:25000316	C	0.466	1142	1031	A	0.534	1194	1305	635.4	527.1	1.206	9.2E-05	OSBPL3,-14031
rs10760120	chr9:99908721	G	0.468	1024	1143	A	0.532	1308	1189	514.3	646.8	0.795	9.3E-05	NANS,23543; TRIM14,within; CORO2A,17575
rs1433781	chr5:162966587	A	0.473	1037	1158	G	0.527	1301	1180	514.8	650.7	0.791	9.3E-05	
rs632608	chr11:81579961	C	0.379	947	828	A	0.621	1391	1510	612.0	488.0	1.254	9.4E-05	
rs10489577	chr1:231021449	C	0.036	103	61	T	0.964	2235	2277	100.0	62.3	1.604	9.5E-05	KIAA1383,8734
rs921383	chr11:77388489	A	0.470	1172	1057	G	0.530	1162	1277	656.4	506.4	1.296	9.8E-05	INTS4,-5124; KCTD14,15919

Shown are association test results for all SNPs with P<0.0001 in the EUR analysis of 583 families. A1=allele 1. Frq = allele frequency in founders. T = transmitted alleles. NT = estimated count of alleles not transmitted by parents. OR=odds ratio. Transmissions from heterozygous parents have been estimated as described in the main text.

Table S2: SNP association results in all families (in or within 50kb of genes)

SNP	LOC	Allele 1 total counts			Allele 2 total counts			T from het parents		OR	P-value	SNPs	Genew (within 50kb)
		A1	T1	NT1	A2	T1	NT2	T1	T2				
rs12210050	chr6:420489	T	582	476	C	1940	2046	459.7	390.6	1.18	4.01E-06	1	EXOC2,9648
rs1170612	chr2:124699526	T	648	533	C	1962	2077	518.7	377.3	1.37	9.15E-06	6	CNTNAP5
rs3197999	chr3:49696536	T	805	676	C	1801	1930	587.0	471.9	1.24	1.10E-05	1	BSN,12550; APEH,598; MST1; RNF123,-5457; AMIGO3,33432; GMPPB,37399; IHPK1,40195
rs12426725	chr12:80367259	A	358	461	G	2252	2149	293.2	389.7	0.75	1.13E-05	1	PPFIA2
rs175	chr7:25000316	C	1273	1144	A	1335	1464	711.9	579.6	1.23	1.58E-05	1	OSBPL3,-14031
rs16934812	chr12:29763585	G	354	271	T	2222	2305	314.9	235.6	1.34	2.01E-05	1	TMTC1
rs1574509	chr1:114432019	T	1190	1063	C	1418	1545	694.5	583.4	1.19	2.19E-05	1	SYT6,1417
rs6443997	chr3:186016225	A	135	196	G	2475	2414	120.0	194.3	0.62	2.70E-05	1	VPS8
rs2656193	chr11:98837693	A	913	1036	G	1697	1574	543.3	667.0	0.81	2.94E-05	2	CNTN5
rs7180015	chr15:85305969	G	202	274	A	2408	2336	176.0	268.9	0.65	3.33E-05	1	AGBL1
rs1037231	chr3:85845797	A	1028	1161	G	1578	1445	563.7	705.2	0.80	3.59E-05	3	CADM2,-12524
rs10225163	chr7:27892170	C	1013	1138	G	1597	1472	574.6	677.9	0.85	4.23E-05	1	JAZF1
rs502038	chr4:45975075	G	1165	1042	T	1445	1568	705.7	554.7	1.27	4.71E-05	1	GABRA2
rs1569604	chr20:8456036	T	1112	1231	C	1488	1369	570.8	711.8	0.80	5.43E-05	1	PLCB1
rs4716801	chr7:157381124	G	1281	1154	A	1327	1454	715.3	579.6	1.23	5.88E-05	1	PTPRN2
rs17086658	chr4:56864663	T	266	191	C	2312	2387	246.4	161.6	1.52	6.36E-05	1	KIAA1211; AASDH,34550
rs10999882	chr10:72996346	T	1125	1245	C	1481	1361	593.0	695.7	0.85	6.53E-05	1	CDH23
rs32181	chr5:61861390	C	940	829	T	1670	1781	645.9	517.8	1.25	6.66E-05	1	IPO11
rs2064712	chr6:161136598	T	379	473	C	2231	2137	307.1	409.0	0.75	6.93E-05	1	PLG,42261
rs1851185	chr2:212235974	T	675	566	C	1935	2044	515.3	435.1	1.18	7.21E-05	1	ERBB4
rs2620440	chr7:146629226	T	582	483	C	2028	2127	457.9	358.5	1.28	7.48E-05	3	CNTNAP2
rs318477	chr6:2832651	A	1263	1142	G	1347	1468	711.9	580.7	1.23	7.60E-05	1	SERPINB1,-45571; SERPINB9
rs12565770	chr1:19427647	A	253	337	G	2357	2273	219.4	302.9	0.72	7.78E-05	1	UBR4,-18314; KIAA0090; MRTO4,-23014; AFAR3,37415
rs247235	chr5:61775220	C	943	833	T	1663	1773	644.0	521.6	1.23	7.79E-05	3	DIMT1L,-39735; IPO11
rs1252216	chr8:85639791	G	663	560	A	1943	2046	520.2	400.3	1.30	8.23E-05	1	RALYL
rs12239401	chr1:235261146	T	1043	1167	C	1565	1441	558.5	708.6	0.79	8.60E-05	1	RYR2,-11178
rs795955	chr12:77160181	T	1002	1113	C	1606	1495	569.0	683.3	0.83	9.16E-05	1	NAV3,29260
rs11695630	chr2:172559798	C	755	873	T	1851	1733	497.2	612.9	0.81	9.19E-05	1	HAT1,2952; MAP1D,-13251
rs9855505	chr3:49767162	G	1423	1295	T	1187	1315	709.3	592.2	1.20	9.31E-05	1	RNF123,33196; AMIGO3,-35035; GMPPB,-30774; IHPK1; LOC389118,43506; C3orf54,-48528
rs939207	chr4:68850266	A	289	215	G	2319	2393	263.4	200.0	1.32	9.37E-05	1	YTHDC1,8433
rs12321966	chr12:8592432	T	271	204	G	2307	2374	250.1	163.4	1.53	9.41E-05	1	CLEC4D,26205; CLEC4E,-7607
rs11063077	chr12:4293296	G	626	730	A	1982	1878	445.4	551.4	0.81	9.43E-05	1	CCND2,8519; C12orf5,-7323
rs731716	chr11:124764510	A	1173	1285	G	1431	1319	569.2	703.9	0.81	9.69E-05	1	PKNOX2

Shown are association test results for all SNPs with P<0.0001 in the all-family analysis of 631 families. See Table S1 legend for abbreviations. SNPs with P<1.0E-05 were also observed in non-genic regions including chr6:33959151, chr4: 101792711-101815546, and chr2:204892416.

Table S3: Results of European-ancestry polygenic score analysis (family study results predicting Psychiatric GWAS Consortium case-control status)

Threshold	P-value for effect of polygenic score (accounting for covariates)	R^2 difference (R^2 for model with score+covariates, minus R^2 for model with only covariates)	N SNPs
0.0001	0.299	0.00006	35
0.001	0.058	0.00019	311
0.01	4.9E-05	0.00089	2480
0.05	1.1E-12	0.00276	10411
0.1	1.2E-14	0.00324	19152
0.2	***1.0E-17***	***0.00400***	34937
0.3	2.3E-15	0.00342	49057
0.4	5.0E-15	0.00334	61852
0.5	2.9E-14	0.00315	73530
1	2.1E-14	0.00319	112869

Shown in the table are the details of results that are illustrated in the main text in Figure 3 (see also the main text and Figure 3 legend). The "R^2 difference" is the variance in case-control status in the PGC GWAS sample attributable to polygenic scores that were computed based on family-study association results (see below), in a logistic regression model predicting PGC case-control status with polygenic scores plus covariates designating 17 PGC study samples (so that each study's controls would be contrasted with its controls, because technical factors and ancestral origins were related to study), plus principal component scores for each PGC subject reflecting ancestral origins (computed across the entire PGC sample). The Nagelkerke's R^2 for covariates-only was subtracted from the R^2 for the full model. Covariates alone explained an R^2 of ~ 0.16 (which is why correcting for them is critical in all analyses).

The analysis was repeated 10 times. The last row includes all 112,869 SNPs, selected because they were genotyped in the family sample, were genotyped or imputed (based on HapMap 3 data, with information content >0.9) in the PGC stage 1 dataset (9,394 European-ancestry cases and 12,462 controls), with minor allele frequency > 2% in both samples, and had pairwise linkage disequilibrium (in 500-SNP windows) restricted to < 0.25; and then the SNPs meeting each other p-value threshold (col 1), e.g., the row for threshold "0.2" includes SNPs whose p-values in the family study were among the 20% best observed p-values.

The maximum effect was 0.4% of variance explained, using SNPs with the best 20% of p-values in the family study.

Table S4: PGC GWAS top SNPs and consistency of direction of effect in the family study

--------Top PGC phase 1 independent SNPs--------							Best fam study proxy			PGC result for proxy				Fam study result				Same:			Corr
SNP	CHR	BP	A	FrA1	OR	P	SNP	BP	R^2	A	FrA1	OR	P	A	OR	FrA1	P	Strnd	Test	DIR	OR
rs2252865	1	8345263	TC	0.337	1.104	4.9E-06	rs10779702	8346097	1.000	AG	0.337	1.102	6.4E-06	GA	0.964	0.690	0.609	Y	N	**Y**	1.037
rs1009080	1	30204147	AG	0.707	0.904	5.8E-06								TC	1.065	0.698	0.414	N	Y	**N**	1.065
rs1625579	1	98275522	TG	0.800	1.142	5.7E-07								CA	0.887	0.187	0.149	N	N	**Y**	1.127
rs7540658	1	181010828	AC	0.698	0.898	1.5E-05								CA	1.042	0.335	0.561	Y	N	**Y**	0.960
rs3818802	1	241516504	AG	0.528	1.101	3.8E-06	rs2484639	241528990	0.762	AG	0.516	1.098	7.2E-06	GA	1.064	0.456	0.372	Y	N	**N**	0.940
rs6703335	1	241675590	AG	0.435	0.907	3.3E-06	rs3006925	241676550	0.436	TC	0.225	0.895	1.2E-05	TC	0.916	0.217	0.260	Y	Y	**Y**	0.916
rs11682175	2	57841097	TC	0.533	0.907	4.1E-06								TC	1.072	0.501	0.278	Y	Y	**N**	1.072
rs17180327	2	180724378	AG	0.653	0.896	6.8E-07								GA	1.025	0.335	0.714	Y	N	**Y**	0.976
rs17662626	2	193692866	AG	0.911	1.222	3.1E-06								GA	1.022	0.080	0.859	Y	N	**N**	0.978
rs2675968	2	233444488	TC	0.302	1.110	2.6E-06	rs709937	233452265	0.989	AG	0.304	1.104	9.7E-06	TC	0.913	0.305	0.215	N	Y	**N**	0.913
rs13025591	2	236460082	AC	0.605	0.902	1.1E-06	rs6741609	236442364	0.723	AG	0.540	0.922	7.8E-05	GA	1.001	0.489	0.992	Y	N	**Y**	0.999
rs4624519	3	36837984	TC	0.351	1.102	6.3E-06								TC	0.996	0.374	0.958	Y	Y	**N**	0.996
rs2239547	3	52830269	TC	0.724	1.117	2.3E-06	rs4687657	52827578	0.988	TG	0.274	0.897	3.7E-06	TG	0.889	0.274	0.118	Y	Y	**Y**	1.125
rs11130874	3	62039809	AG	0.792	1.145	2.1E-07	rs191558	62054056	0.966	AG	0.207	0.876	3.2E-07	TC	0.906	0.201	0.246	N	Y	**Y**	1.104
rs1879248	3	182033908	TC	0.731	1.128	1.3E-06	rs9838229	182015945	1.000	AC	0.732	1.128	1.3E-06	CA	1.037	0.243	0.642	Y	N	**N**	0.964
rs4295265	4	103054668	TC	0.339	1.103	1.1E-05	rs2850377	103131182	0.699	TC	0.424	1.094	2.4E-05	TC	0.975	0.454	0.711	Y	Y	**N**	0.975
rs7730479	5	21955293	TC	0.141	1.202	1.5E-05								TC	1.067	0.147	0.527	Y	Y	**Y**	1.067
rs1433019	5	171917518	AC	0.781	1.125	3.1E-06								CA	1.040	0.196	0.634	Y	N	**N**	0.962
rs2021722	6	30282110	TC	0.215	0.843	4.3E-11	rs2844776	30279806	1.000	TC	0.785	1.186	5.0E-11	GA	0.910	0.216	0.229	N	N	**Y**	0.910
rs9462875	6	43276095	AG	0.823	1.153	1.5E-06								GA	1.064	0.161	0.474	Y	N	**N**	0.940
rs10226475	7	2192688	AG	0.605	1.124	5.1E-08	rs12666575	1970947	0.495	TC	0.390	0.894	2.3E-07	TC	1.044	0.384	0.540	Y	Y	**N**	0.958
rs12699131	7	71389252	AG	0.472	1.098	5.2E-06	rs756912	71379733	0.990	TC	0.527	0.911	5.5E-06	TC	0.953	0.503	0.493	Y	Y	**Y**	1.049
rs4415249	7	134347420	AC	0.150	0.874	4.2E-06	rs4329203	134347280	1.000	AC	0.851	1.141	7.1E-06	CA	0.886	0.149	0.186	Y	N	**Y**	0.886
rs10503253	8	4168252	AC	0.191	1.141	3.8E-07								CA	0.976	0.782	0.771	Y	N	**Y**	1.025
rs10503256	8	4201587	AG	0.349	1.118	2.0E-07								GA	0.911	0.628	0.197	Y	N	**Y**	1.098
rs500115	8	8672925	TC	0.862	1.152	5.8E-06								GA	0.995	0.135	0.961	N	N	**Y**	1.005
rs12234997	8	9275118	AG	0.435	1.105	3.1E-06	rs12235038	9274813	0.931	TG	0.557	0.909	6.9E-06	TG	1.049	0.555	0.479	Y	Y	**N**	0.953
rs7004633	8	89829427	AG	0.819	0.858	1.5E-08								GA	0.962	0.194	0.651	Y	N	**N**	1.040
rs10098073	8	143307411	AC	0.474	1.098	5.4E-06	rs4129585	143310840	0.775	AC	0.441	1.094	1.5E-05	TG	1.047	0.443	0.501	N	Y	**Y**	1.047
rs12352353	9	4733341	AG	0.848	1.161	6.6E-07	rs396861	4733626	0.981	AG	0.846	1.160	7.5E-07	GA	1.109	0.143	0.306	Y	N	**N**	0.902
rs6602217	10	6986269	TC	0.930	0.843	1.8E-05								TC	0.963	0.925	0.746	Y	Y	**Y**	0.963
rs41441548	10	34190827	AG	0.069	1.245	2.7E-06	rs10508791	34187301	0.465	TC	0.865	0.896	2.6E-04	TC	1.010	0.853	0.910	Y	Y	**N**	0.990
rs16915157	10	62016644	TC	0.195	1.126	4.4E-06	rs2068043	61990336	0.934	AG	0.185	1.127	5.4E-06	GA	0.882	0.820	0.136	Y	N	**Y**	1.134
rs7914558	10	104765898	AG	0.414	0.896	1.6E-07	rs4532960	104657396	0.990	TC	0.415	0.898	2.4E-07	TC	0.839	0.413	0.014	Y	Y	**Y**	0.839
rs11191580	10	104896201	TC	0.909	1.227	2.2E-08	rs12413409	104709086	1.000	AG	0.091	0.828	2.1E-07	GA	1.122	0.925	0.357	Y	N	**Y**	1.122
rs11191732	10	105321751	AG	0.204	1.142	2.3E-07								GA	0.810	0.806	0.014	Y	N	**Y**	1.235
rs1025641	10	128297182	TC	0.667	1.117	8.3E-07								GA	0.935	0.319	0.349	N	N	**Y**	1.070
rs4356203	11	17116724	AG	0.591	0.905	1.9E-06	rs621246	17160341	0.859	TG	0.537	0.930	3.9E-04	TG	1.032	0.526	0.640	Y	Y	**N**	1.032
rs7938219	11	29159016	AG	0.111	1.159	5.7E-06								GA	0.946	0.896	0.589	Y	N	**Y**	1.057
rs2509843	11	97630614	AG	0.608	0.901	1.1E-06	rs2852034	97617703	0.895	AC	0.622	0.903	1.3E-06	CA	1.102	0.377	0.169	Y	N	**Y**	0.907
rs11220082	11	124829175	TC	0.528	1.112	2.6E-07	rs671789	124799824	0.480	AC	0.301	0.898	1.8E-06	TG	0.974	0.276	0.726	N	Y	**Y**	1.027
rs548181	11	124966919	AG	0.120	0.832	2.9E-08	rs540723	124994831	0.948	AG	0.118	0.841	1.5E-07	TC	0.887	0.108	0.270	N	Y	**Y**	0.887
rs10894294	11	130335958	AC	0.518	0.911	6.5E-06	rs7111478	130324838	0.960	AG	0.484	1.095	1.1E-05	GA	0.978	0.485	0.737	Y	N	**Y**	0.978

--------Top PGC phase 1 independent SNPs--------							Best fam study proxy			PGC result for proxy				Fam study result				Same:			Corr
SNP	CHR	BP	A	FrA1	OR	P	SNP	BP	R^2	A	FrA1	OR	P	A	OR	FrA1	P	Strnd	Test	DIR	OR
rs7972947	12	2040694	AC	0.220	0.865	7.8E-07								CA	0.886	0.814	0.193	Y	N	**N**	1.129
rs4765905	12	2219845	CG	0.328	1.113	8.9E-07	rs2159100	2216654	0.976	TC	0.327	1.111	1.5E-06	TC	1.040	0.346	0.579	Y	Y	**Y**	1.040
rs10135277	14	34892982	TC	0.426	0.901	5.1E-07	rs12436216	34879165	0.867	AG	0.429	0.902	8.1E-07	GA	0.946	0.595	0.417	Y	N	**N**	1.057
rs1869901	15	38382919	AG	0.608	0.898	3.5E-07								TC	0.872	0.626	0.050	N	Y	**Y**	0.872
rs4775413	15	59627395	TC	0.540	1.101	4.0E-06	rs2414718	59650425	0.691	AG	0.585	1.099	7.1E-06	GA	0.937	0.387	0.346	Y	N	**Y**	1.067
rs1078163	15	86269935	TC	0.850	0.879	5.1E-06								GA	0.869	0.178	0.112	N	N	**N**	1.151
rs16957445	18	49657366	TC	0.301	0.903	8.2E-06	rs1364467	49494095	0.347	TG	0.813	1.114	5.9E-05	CA	1.224	0.177	0.019	N	N	**N**	1.224
rs12966547	18	50903015	AG	0.417	0.902	1.0E-06	rs4309482	50901467	1.000	AG	0.583	1.108	1.1E-06	GA	0.985	0.397	0.827	Y	N	**Y**	0.985
rs17512836	18	51345959	TC	0.975	0.711	2.4E-08	rs17594721	51216890	0.768	AG	0.972	0.753	1.1E-06	GA	1.022	0.032	0.895	Y	N	**Y**	0.978
rs7248806	19	946943	TG	0.659	1.142	3.0E-05	rs2240152	943903	0.580	TC	0.533	1.082	1.7E-03	TC	1.098	0.522	0.175	Y	Y	**Y**	1.098
rs8112050	19	37323114	TC	0.704	0.906	1.8E-05								TC	0.970	0.693	0.678	Y	Y	**Y**	0.970
rs16997475	21	26430592	AG	0.063	1.209	9.6E-06	rs7283136	26365981	0.869	TC	0.933	0.841	1.6E-05	TC	0.909	0.926	0.448	Y	Y	**Y**	1.100
rs2833899	21	32855134	TC	0.821	1.134	4.1E-06	rs11702343	32859759	1.000	AC	0.179	0.883	4.9E-06	CA	0.954	0.849	0.620	Y	N	**N**	0.954
rs7289747	22	18290716	AC	0.937	0.835	9.7E-06								CA	1.025	0.063	0.843	Y	N	**Y**	0.976

For the PGC phase 1 GWAS of 17 schizophrenia datasets comprising 9,394 cases and 12,462 controls of European ancestry (3), data are shown for the 58 SNPs with the top p-values, selecting the best SNP in any region while excluding additional SNPs with pairwise LD > 0.2 (and selecting only the SNP with the best p-value in the Major Histocompatibility Complex region on chromosome 6). Shown for these 58 SNPs are:

The SNP ID, chromosome (CHR), base pair (BP) position (HG18 locations), the two SNP alleles (A), frequency of Allele 1 (FrA1), odds ratio (OR) and P-value (P) in the PGC GWAS, which was a combined analysis of genotyped and imputed data, correcting for ancestry and study.

For SNPs not genotyped in the family sample, the closest proxy (nearby SNP with $r^2 > 0.3$) genotyped in that sample was selected, and the data shown include the SNP ID, base pair position, and r^2 value between the PGC best SNP and the proxy, and then the PGC data for that SNP.

The family study data (for the PGC SNP or proxy) are then shown, followed by columns showing whether the PGC and family study analysis were for the same strand (Yes or No) and the same test allele, and whether the direction of the effect was in the same direction (both odds ratios>1 or both odds ratios<1) after correcting for strand or test allele differences.

The “Corr OR” column is the odds ratio for the family study after correcting for strand and/or test allele.

PGC and family study odds ratios were in the same direction for 37 of 58 SNPs. A one-sided binomial test yielded p=0.024 for the probability of observing 37 or more agreements. If we restricted the analysis to family study proxies with $r^2 > 0.8$, there were 29 agreements out of 45 SNPs (p=0.036).

Summary of pathway-based analyses (ALIGATOR)

The ALIGATOR software package (4) was used to evaluate whether more significant p-values were observed for SNPs in genes in genetic pathways as defined by the GO, KEGG, MGI, PANTHER, BioCarta and Reactome databases, plus two additional sets of interest; a locally-curated list of 58 genes whose products are known to interact with neurexins, because of the significant associated of schizophrenia with chromosomal deletions of exons of NRXN1 (5, 6); and a set of 386 genes (based on the Aceview database) whose products have immunoglobulin structures, which may play a role in brain development (7). Briefly, we selected a set of SNPs based on a threshold of $P < 0.00667$ (chosen because this selected 5% of genes as containing at least one such SNP). ALIGATOR then defined a list of "significant" genes as the set of genes which contain at least one such SNP, counting gene only once, regardless of how many "significant" SNPs it contains, since multiple significant SNPs may reflect linkage disequilibrium within the gene, rather than independent signals. Note that in this analysis we considered only SNPs that lay within the largest transcript of each gene, without allowing for any margin around genes. A large number of replicate gene lists of the same length as the list of significant genes from the actual data were then generated by selecting SNPs (not genes) at random. This allows for large genes containing several SNPs being more likely to contain a significant SNP by chance than small genes. The number of significant genes in a pathway in the actual data was compared to the number of genes from that pathway in the replicate gene lists, thereby giving a pathway-specific p-value for enrichment. These were corrected for multiple testing of non-independent pathways by a bootstrapping method (for more details, see (4)), which also gives a test of whether the number of pathways achieving a given level of significance is higher than expected. Importantly, genes from the same pathway lying less than 1Mb apart were counted as the same signal, to correct for the possibility that their associations were due to intergenic LD. A Simes-corrected p-value was then determined for each gene -- i.e., this p-value serves as a correction for the number of SNPs in a gene and thus gives information about the relative significance of genes, but does not correct for the number of genes tested.

Results are summarized in Tables S4 and S5 below. There were no significant results: there was no pathway with a significant p-value after permutation-based study-wide correction (i.e., across pathways), and no significant excess of the number of pathways exceeding any of three thresholds (Table S4a). However, results may be of interest for comparison with other findings

Table S5a: ALIGATOR analysis of the total number of "significant" pathways

Families	*Best pathway corrected P*	*Uncorrected threshold for declaring a pathway to contain an excess of genes with ≥ 1 SNP with P<0.00667 (i.e., in top 5% of genes)*					
		P<0.05		*P<0.01*		*P<0.001*	
		#pathways	p	#pathways	p	#pathways	p
European-ancestry	0.796	325	0.244	78	0.130	8	0.309
All families	0.662	405	0.064	93	0.058	9	0.266

In this analysis using the methods described above, 861 genes were declared "significant" because they contained at least one SNP with uncorrected P<0.00667 (a threshold which selected 5% of genes as "significant", with 4,014 SNPs meeting this criterion).

Permutation-based procedures were used to determine which pathways had a significant (uncorrected) excess of genes containing at least one "significant" SNP based on three different thresholds for declaring the excess in each pathway to be significant (0.05, 0.01 and 0.001), and to determine whether the total number of such pathways exceeded chance expectation. The corrected significance (across all pathways) for each pathway was also determined and the best value is shown for each analysis (European-ancestry and all families) -- see further details below for the primary European-ancestry analysis (all-family results available on request).

No single pathway achieved corrected significance, and the number of pathways with an excess of "significant" genes was not significant at any of the three thresholds.

Table S5b: Top pathway results
(European-ancestry families, pathways containing ≥10 genes)

PATHWAY	# GENES	# with P<0.05	EXPECTED # GENES	PATHWAY-SPECIFIC P	CORRECTED P	EXPECTED PATHWAYS PER STUDY	PATHWAY FUNCTION
GO: 6898	51	11	3.95	2E-04	0.796	2.36	receptor-mediated endocytosis
GO: 1755	18	5	0.79	4E-04	0.886	3.31	neural crest cell migration
GO: 34707	51	11	4.37	6E-04	0.936	4.27	chloride channel complex
GO: 48705	100	16	7.3	1E-03	0.979	6.19	skeletal system morphogenesis
GO: 1667	29	6	1.33	1E-03	0.979	6.19	ameboidal cell migration
MGI: 1407	26	9	4.16	1E-03	0.989	7.23	short stride length
GO: 48306	25	5	0.95	1E-03	0.992	8.21	calcium-dependent protein binding
MGI: 79	19	6	1.72	2E-03	0.995	11.22	abnormal basioccipital bone morphology
GO: 34660	227	13	5.13	2E-03	0.998	13.29	ncRNA metabolic process
GO: 6364	76	5	1.03	2E-03	0.998	13.29	rRNA processing
GO: 16072	79	5	1.06	2E-03	0.998	13.29	rRNA metabolic process
GO: 5689	18	3	0.3	2E-03	0.998	13.29	U12-type spliceosomal complex
GO: 5246	10	4	1.14	3E-03	0.999	15.41	calcium channel regulator activity

For the analysis described in the Table S4a legend, Table S4b shows results for the top pathways, excluding those containing <10 genes. See Table S5 for information on individual genes. Note that some genes were considered part of more than one of the pathways shown here (see Table S5), although this overlap is accounted for by the permutation-based procedure to determine P-values.

Table S6: Genes with SIMES-corrected P<0.05 in the 13 pathways listed in Table S5b

GENE	CHR	START (BP)	END (BP)	N(SNPS)	MOST SIG P	SIMES-CORR P	MOST SIG SNP	DESCRIPTION
GO: 6898 (receptor-mediated endocytosis)								
CAV1	7	115952075	115988466	13	4.9E-04	4.9E-03	rs3807986	caveolin 1
IGF2R	6	160310121	160447573	39	4.0E-04	1.3E-02	rs435612	insulin-like growth factor 2 receptor precursor
HTR2B	2	231681199	231698068	3	4.5E-03	1.4E-02	rs17586405	5-hydroxytryptamine (serotonin) receptor 2B
ARHGAP27	17	40827058	40858780	5	3.2E-03	1.6E-02	rs1059504	Rho GTPase activating protein 27
HIP1R	12	121885992	121913460	5	3.4E-03	1.7E-02	rs2271051	huntingtin interacting protein-1-related
CETP	16	55553263	55575257	14	1.4E-03	2.0E-02	rs5882	cholesteryl ester transfer protein, plasma
MSR1	8	16009758	16094671	17	1.4E-03	2.4E-02	rs17484273	macrophage scavenger receptor 1 isoform type 1
PPT1	1	40310969	40335555	12	2.3E-03	2.7E-02	rs7543269	palmitoyl-protein thioesterase 1
DNM1	9	130005484	130057348	13	2.9E-03	3.8E-02	rs3003569	dynamin 1 isoform 1
GO: 1755 (neural crest cell migration)								
HTR2B	2	231681199	231698068	3	4.5E-03	1.4E-02	rs17586405	5-hydroxytryptamine (serotonin) receptor 2B
KITLG	12	87410697	87498369	18	1.4E-03	2.5E-02	rs10777131	KIT ligand isoform a precursor
ACVR1	2	158301207	158403036	13	3.2E-03	4.2E-02	rs10497192	activin A type I receptor precursor
GO: 34707 (chloride channel complex)								
GABRG1	4	45732543	45820839	14	1.4E-05	2.0E-04	rs12511372	gamma-aminobutyric acid A receptor, gamma 1
GABRA2	4	45946463	46086702	10	4.3E-05	3.5E-04	rs534459	gamma-aminobutyric acid A receptor, alpha 2,gamma-aminobutyric acid A receptor, alpha 2
ANO5	11	22171298	22257975	9	4.0E-03	1.6E-02	rs7951981	anoctamin 5
GABRB1	4	46728336	47123202	71	2.6E-04	1.8E-02	rs4396968	gamma-aminobutyric acid (GABA) A receptor, beta
GABRE	23	150872252	150893807	10	2.6E-03	2.6E-02	rs2266856	gamma-aminobutyric acid (GABA) A receptor
GABRG2	5	161427295	161515106	19	1.5E-03	2.9E-02	rs211029	gamma-aminobutyric acid A receptor, gamma 2
ANO10	3	43382822	43638564	16	3.0E-03	4.9E-02	rs17075727	anoctamin 10
GO: 48705 (skeletal system morphogenesis)								
THRA	17	35472589	35503646	3	3.5E-04	1.0E-03	rs3744805	thyroid hormone receptor, alpha isoform 1
RARG	12	51890620	51912303	5	2.7E-03	8.1E-03	rs1465057	retinoic acid receptor, gamma isoform 1
COL9A1	6	70982529	71069494	41	2.8E-04	8.1E-03	rs544179	alpha 1 type IX collagen isoform 1 precursor
MDFI	6	41714231	41729959	5	2.3E-03	1.1E-02	rs4714501	MyoD family inhibitor
DSCAML1	11	116803699	117173186	150	1.8E-04	1.7E-02	rs558582	Down syndrome cell adhesion molecule like 1
BMP7	20	55178962	55274708	29	8.6E-04	2.5E-02	rs6025446	bone morphogenetic protein 7 precursor
HOXD3	2	176737051	176746072	1	2.5E-02	2.5E-02	rs2301301	homeobox D3
SIX4	14	60246009	60260545	1	2.8E-02	2.8E-02	rs17834412	sine oculis homeobox homolog 4
PEX7	6	137185416	137276752	9	3.4E-03	3.0E-02	rs3799479	peroxisomal biogenesis factor 7
BMP6	6	7672010	7826960	48	1.9E-03	3.3E-02	rs270392	bone morphogenetic protein 6 preproprotein
PRRX2	9	131467741	131524774	10	3.9E-03	3.9E-02	rs7858199	paired related homeobox 2
WNT9B	17	42283967	42309436	5	1.0E-02	4.1E-02	rs2165846	wingless-type MMTV integration site family
GO: 1667 (ameboidal cell migration)								
HTR2B	2	231681199	231698068	3	4.5E-03	1.4E-02	rs17586405	5-hydroxytryptamine (serotonin) receptor 2B
KITLG	12	87410697	87498369	18	1.4E-03	2.5E-02	rs10777131	KIT ligand isoform a precursor

ACVR1	2	158301207	158403036	13	3.2E-03	4.2E-02	rs10497192	activin A type I receptor precursor
MGI: 1407 (short stride length)								
CAV1	7	115952075	115988466	13	4.9E-04	4.9E-03	rs3807986	caveolin 1
HMBS	11	118460797	118469469	1	1.9E-02	1.9E-02	rs1784304	hydroxymethylbilane synthase isoform 1
NPAS3	14	32478200	33340702	367	1.1E-04	4.1E-02	rs12147692	neuronal PAS domain protein 3 isoform 1
GO: 48306 (calcium-dependent protein binding)								
DMBT1	10	124310171	124393242	4	5.6E-04	2.3E-03	rs8441	deleted in malignant brain tumors 1 isoform a
ANXA11	10	81904860	81955308	12	2.9E-04	3.4E-03	rs6585424	annexin A11
STX2	12	129840098	129889764	8	4.9E-03	2.5E-02	rs1554807	syntaxin 2 isoform 1
TNNT3	11	1897445	1916514	3	1.1E-02	3.2E-02	rs909116	troponin T3, skeletal, fast isoform 1
MASP1	3	188418632	188492446	35	1.1E-03	3.8E-02	rs12489890	mannan-binding lectin serine protease 1 isoform
MGI: 79 (abnormal basioccipital bone morphology)								
RARG	12	51890620	51912303	5	2.7E-03	8.1E-03	rs1465057	retinoic acid receptor, gamma isoform 1
BMP7	20	55178962	55274708	29	8.6E-04	2.5E-02	rs6025446	bone morphogenetic protein 7 precursor
GO: 34660 (ncRNA metabolic process)								
AARS	16	68843798	68880913	2	3.1E-04	6.3E-04	rs2070203	alanyl-tRNA synthetase
BOP1	8	145456864	145485928	4	5.8E-04	2.3E-03	rs11781564	block of proliferation 1,block of proliferation 1,
RARS2	6	88280820	88356440	6	5.0E-04	3.0E-03	rs7757636	arginyl-tRNA synthetase-like
DIMT1L	5	61720108	61735485	2	1.7E-03	3.4E-03	rs35015	dimethyladenosine transferase
SNRPF	12	94776840	94784369	2	2.2E-03	4.4E-03	rs3751264	small nuclear ribonucleoprotein polypeptide F
PES1	22	29302612	29317894	3	2.0E-03	6.1E-03	rs4820018	pescadillo homolog 1, containing BRCT domain
CDK5RAP1	20	31410306	31452998	5	1.5E-03	7.6E-03	rs291671	CDK5 regulatory subunit associated protein 1
TPR	1	184547407	184611080	8	3.7E-03	1.2E-02	rs3820182	nuclear pore complex-associated protein TPR
ADAT3	19	1856417	1864446	1	1.5E-02	1.5E-02	rs7260336	hypothetical protein LOC113179
C4orf23	4	8507043	8529181	3	5.8E-03	1.7E-02	rs16842315	hypothetical protein LOC152992 isoform 2
SIP1	14	38653239	38675928	2	1.9E-02	3.8E-02	rs9322993	SMN-interacting protein 1 isoform alpha
ERI1	8	8897860	8925899	6	6.5E-03	3.9E-02	rs2953807	exoribonuclease 1
UTP11L	1	38250971	38263084	1	4.2E-02	4.2E-02	rs11211383	UTP11-like, U3 small nucleolar
NSA2	5	74098859	74108490	1	4.3E-02	4.3E-02	rs1164694	NSA2 ribosome biogenesis homolog (S. cerevisiae)
SNRPG	2	70362009	70374373	1	4.5E-02	4.5E-02	rs6708754	small nuclear ribonucleoprotein polypeptide G
GO: 6364 (rRNA processing)								
BOP1	8	145456864	145485928	4	5.8E-04	2.3E-03	rs11781564	block of proliferation 1
DIMT1L	5	61720108	61735485	2	1.7E-03	3.4E-03	rs35015	dimethyladenosine transferase
PES1	22	29302612	29317894	3	2.0E-03	6.1E-03	rs4820018	pescadillo homolog 1, containing BRCT domain
ERI1	8	8897860	8925899	6	6.5E-03	3.9E-02	rs2953807	exoribonuclease 1
UTP11L	1	38250971	38263084	1	4.2E-02	4.2E-02	rs11211383	UTP11-like, U3 small nucleolar
NSA2	5	74098859	74108490	1	4.3E-02	4.3E-02	rs1164694	NSA2 ribosome biogenesis homolog (S. cerevisiae)

Shown are data for each gene with a Simes-corrected gene-wise p-value < 0.05 in the 13 pathways listed in Table S4b. There are 58 unique genes, of which 6 (shown in italics) are listed in more than one pathway.

Table S7: Functional annotation of genes with ≥ 1 SNP with P < 0.0001 (European-ancestry families)

Gene	Description	Comment (function)
EXOC2	Sec5 protein	vesicle transport
PPFIA2	PTPRF interacting protein alpha 2	axon guidance
CNTNAP5	contactin associated protein-like 5 isoform 1	neurexin family, cell adhesion, signaling
TMTC1	ARG99 protein	
GABRG1	gamma-aminobutyric acid A receptor, gamma 1	
GABRA2	gamma-aminobutyric acid A receptor, alpha 2	
BSN	bassoon protein	neuronal glutamate release
APEH	N-acylaminoacyl-peptide hydrolase	cancer; destroy oxidation-damaged proteins
MST1	macrophage stimulating 1 (hepatocyte growth	ciliary motility lung
RNF123	ring finger protein 123	ubiquitin function
AMIGO3	amphoterin-induced gene and ORF 3	cell adhesion (neuronal)
GMPPB	GDP-mannose pyrophosphorylase B isoform 1	oligosaccharide formation
IHPK1	inositol hexaphosphate kinase 1 isoform 1	
PTPRN2	protein tyrosine phosphatase, receptor type, N	cellular growth, differentiation, autoantigen (DM)
ABCB5	ATP-binding cassette, sub-family B, member 5	diverse; pigmented cells
RYR2	cardiac muscle ryanodine receptor	cardiac contraction ; also expressed in brain
HAT1	histone acetyltransferase 1 isoform a	?role in telomeric silencing
MAP1D	methionine aminopeptidase 1D	
CADM2	immunoglobulin superfamily, member 4D	cell adhesion molecule
ADCY7	adenylate cyclase 7	
BRD7	bromodomain containing 7	chromatin remodeling; p53 tumor suppression
DKFZp547H025	hypothetical protein LOC56918	folate-transporter-like protein
SLC19A3	solute carrier family 19, member 3	biotin-responsive basal ganglia disease; Wernicke's-like encephalopathy
UBR4	retinoblastoma-associated factor 600	clathrin-related scaffolding; integrin-mediated signaling
KIAA0090	hypothetical protein LOC23065	
MRTO4	ribosomal protein P0-like protein	? role in ribosome assembly
AFAR3	aflatoxin B1 aldehyde reductase 3	
ERBB4	v-erb-a erythroblastic leukemia viral oncogene	neuregulin receptor; neuronal differentiation
CLEC4D/E	C-type lectin domain family 4, member D / member E	cell adhesion; antigen uptake
POP4	POP4 (processing of precursor, S. cerevisiae)	
PLEKHF1	apoptosis-inducing protein D	induce apoptosis; weakly expressed in brain
FAM50B	family with sequence similarity 50, member B	fetal brain
VPS8	vacuolar protein sorting 8 homolog isoform b	
NAV3	neuron navigator 3	?neuronal regeneration; fetal brain
CCDC38	coiled-coil domain containing 38	
AMDHD1	amidohydrolase domain containing 1	
HAL	histidine ammonia-lyase	
LTA4H	leukotriene A4 hydrolase	
SNRPN	small nuclear ribonucleoprotein polypeptide N	pre-mRNA processing; SLE autoantigen
AGBL1	ATP/GTP binding protein-like 1	tubulin processing
GABRR2	gamma-aminobutyric acid (GABA) receptor, rho 2	brain and esp retina
UBE2J1	ubiquitin-conjugating enzyme E2, J1	attach of ubiq to proteins -- ER
RRAGD	Ras-related GTP binding D	G protein
WWP2	WW domain containing E3 ubiquitin protein ligase	NEDD4-like fam; ubiq prot ligase; inhib Tcell activ-induced cell death; atrophin-1-interacting protein
LOC348174/ CLEC18A	secretory protein LOC348174	

ERG	v-ets erythroblastosis virus E26 oncogene like	transcr regulator; chromatin modification
FAM19A5	family with sequence similarity 19	brain-spec chemokine/neurokine
OSBPL3	oxysterol-binding protein-like protein 3 isoform	lipid receptor
NANS	N-acetylneuraminic acid phosphate synthase	Produces N-acetylneuraminic acid (Neu5Ac)
TRIM14	tripartite motif protein TRIM14 isoform alpha	inhibits SPI1 (TF)
CORO2A	coronin, actin binding protein, 2A	tf in diff and activation of macrophages or B-cells
KIAA1383		
INTS4	integrator complex subunit 4	RNA splicing
KCTD14	potassium channel tetramerisation domain	

Supplementary Methods for Candidate CNV Analyses

Case-control analyses of rare CNVs (frequency < 1% in controls) were carried out to identify new candidate CNVs. These analyses produced no statistically significant results given the number of tests that were performed (as expected given the small sample size), and thus are presented as supplementary material for the consideration of investigators in the field.

Subjects. The initial analysis considered one proband from each European-ancestry family (N=585, including 581 probands from families who were eligible for SNP association analyses plus 4 married-in cases who were not part of informative sibships) vs. 2,682 unrelated European-ancestry controls recruited (with informed consent) from general pediatric practices affiliated with Children's Hospital of Philadelphia (CHOP), with no congenital anomaly or neurological diagnoses in blinded electronic medical research records. CNVs observed in 1-2 probands and 0 controls, or 3 probands and 0-1 controls, were further examined for familial segregation: for all families with at least one carrier of the CNV (for all ancestries, whether or not the proband was the carrier), the number of carriers vs. non-carriers among siblings with a Narrow diagnosis (schizophrenia or schizoaffective disorder), with a "Broad" diagnoses in the three datasets for which these diagnoses were available (other non-affective psychoses or schizotypal or paranoid personality disorders; this information was available only for the Australia/US, Paris and VCU/Ireland datasets), and with no known schizophrenia spectrum disorder. We also examined the frequency of selected CNVs in data from the Molecular Genetics of Schizophrenia (6) study (3,945 schizophrenia or schizoaffective cases, 3,611 controls) and from the International Schizophrenia Consortium (8) (3,391 schizophrenia cases, 3,181 controls) (http://pngu.mgh.harvard.edu/isc/isc-r1.cnv.bed). Results of these analyses are shown in Table S8, with deletions and duplications each grouped according to the degree of familial segregation (>75% or <75% of narrow-diagnosis siblings carrying the CNV).

Methods. QC filtering was carried out for families and CHOP controls. Subjects were excluded if they had ≥50 CNV calls or if the standard deviation of the log(R) ratio was >0.4

(increased signal variability across all probes). Based on call concordance between 27 pairs of duplicate assays, we retained CNVs spanning ≥5 probes for homozygous deletions (88% concordance); or ≥9 for heterozygous deletions (91%) or for any duplication (81%). Higher thresholds did not improve concordance. Adjacent deletions (copy numbers [CN] 0 and 1) and duplications (3 and 4) were merged. Neighboring deletions or duplications were merged if an intervening segment with copy number=2 contained <30% of probes within the merged CNV. CNVs were excluded for 50% overlap with telomeric or centromeric regions or any overlap with immunoglobulin or olfactory receptor gene clusters. We excluded CNVs detected in >50% of family or control subjects; and CNVs in regions with patterns of multiple CNVs in multiple individuals (mostly known segmental duplication regions) or in regions with common CNVs (>1% in either group) with a ten-fold or greater difference between family vs. control subjects suggesting technical artifact given that such high odds ratios are extremely unlikely for common variants. Also, we sought to minimize the possible effect of cell line artifacts. For the family sample, the source of extracted DNA (lymphoblastic cell line vs. fresh blood) was definitively known for approximately two-thirds of the specimens, of which 59% were extracted from blood and 41% from cell lines. There was no overall difference in CNV count between these two groups, but we excluded CNVs that were greater than 4 Mb long (which are usually cell line artifacts (6)), and CNVs in regions with: significantly different prevalence in specimens known to be of cell line vs. blood origin. All excluded regions are listed in **Table S10**, below.

Post-QC, family members had more total CNVs than controls (7.28±3.22 vs. 5.98±2.50, $p<10^{-15}$), primarily deletions <100,000 bp ($p<10^{-15}$; duplications, p=0.0028). However, this is consistent with the larger SD(log(R) ratio) in family members (0.156±0.034 vs. 0.138±0.029, $p<10^{-15}$), suggesting a technical difference between family and control samples which were genotyped separately. Thus this is not an appropriate dataset for testing the hypothesis of a genome-wide excess of rare CNVs in schizophrenia (6, 8, 9), including familial schizophrenia (10).

Rare CNVs selected as possible candidates (see below) were visualized in all family members by plotting log(R) ratio, B-allele frequency (BAF) and point-by-point CN estimates using a second algorithm (11). Regions were discarded if the PennCNV calls were not confirmed and/or if the data for family members without CNV calls was ambiguous, suggesting technical artifacts in the region. Because this was an exploratory analysis and no statistically significant results were observed, independent biological validation was not carried out.

Analysis of association. We identified rare (present in <1% of controls) candidate deletions and of duplications that disrupted exons of RefSeq genes (HG18). Separately for deletions and duplications, PLINK (12) counted exonic CNVs for each gene for 585 European-ancestry unrelated cases vs. 2,682 CHOP controls, and computed empirical pointwise and genome-wide P-values by permuting case-control status. Genes with exonic CNVs in less than 1% of controls were considered further. Exploratory analyses added the remaining 578 narrow-diagnosis relatives and then 152 broad cases; analyses were repeated for all ancestries (adding 47 probands, 89 other narrow cases, and 10 broad cases). For genes with pointwise $p < 0.05$ (EUR) or control counts of 0-1 and a clear excess in ALL narrow+broad cases: all CNVs in family members were plotted (visualization of log(R) ratio and B allele frequency, and analysis of point-by-point copy number estimate by a second method ; regions with multiple ambiguous calls excluded; familial segregation examined; and MGS and ISC case-control counts inspected.

Table S8: Candidate CNVs

GENE(S)	LOCATION (HG18)	Size(s)	Fams	European-ancestry case-control analysis: N with CNV Case N=585	N with CNV Cont N=2682	Empirical P Point-Wise	Empirical P Genome-Wide	All Siblings with/without CNV: Narrow +	Narrow -	Broad +	Broad -	Unaff +	Unaff -	ISC study Ca	ISC study Co	MGS Study Ca	MGS Study Co
DELETIONS observed in at least 75% of narrow-diagnosis siblings in carrier families																	
TXNIP... POLR3C†	chr1:144.11-144.48	121K, 330K,376K	2	2	0	0.031	0.974	2	1			0	1	1	1	2	0
LY75	chr2:160.42-160.46	45K	1	0	0	1.000	1.000	4	0							3	1
SGCD	chr5:155.46-155.69	232K	1	1	0	0.177	1.000	3	0					0	0	0	0
LOC729920,SOSTDC1‡	chr7:16.39-16.51	115K	1	0	0	1.000	1.000	4	0					0	0	4	2
PSD3	chr8:18.61-18.72	62K,110K	3	2	0	0.034	0.974	4	0	1	0			0	0	0	0
GOLSYN,KCNV1	chr8:110.74-112.81	2071K	1	1	0	0.185	1.000	2	0					0	0	0	1
NUDT7	chr16:76.06-76.36	299K	1	1	0	0.180	1.000	2	0					0	1	0	0
TCF4	chr18:50.98-51.04	61K	1	1	0	0.184	1.000	2	0							0	0
ZNF100,ZNF43	chr19:21.72-21.83	118K	1	1	0	0.178	1.000	2	0	1	0			0	2	0	0
ZNF600,ZNF28,ZNF468*	chr19:57.96-58.05	27K-91K	5	2	0	0.035	0.974	6	2			0	1	1	0	5	0
APOL3	chr22:34.87-34.91	37K,43K	2	2	0	0.030	0.974	3	0							0	0
DELETIONS observed in < 75% of narrow-diagnosis siblings in carrier families																	
PRSS35	chr6:84.21-84.29	75K	2	2	0	0.032	0.974	2	2	0	2					0	0
PACRG**	chr6:163-163.32	38K, 315K	2	1	0	0.182	1.000	2	3					0	1	1	1
C9orf11,MOBKL2B	chr9:27.22-27.32	100K	1	1	0	0.178	1.000	1	1	2	0	1	1	0	0	1	0
DUPLICATIONS observed in at least 75% of narrow-diagnosis siblings in carrier families																	
FBLN2	chr3:13.6-13.69	91K	1	0	0	1.000	1.000	3	0					1	0	0	1
LSM8,ANKRD7	chr7:117.17-118.95	177K	2	2	0	0.030	0.999	3	0	0	1			0	2	0	0
MSR1	chr8:15.73-16.39	650K-660K	2	1	0	0.180	1.000	3	1	2	0	1	1	0	1	0	0
WDR89,SGPP1	chr14:63.14-63.35	208K	1	1	0	0.185	1.000	3	0					0	0	1	0
ADAMTS17	chr15:98.17-98.4	229K	1	0	0	1.000	1.000	3	0					2	1	9	4
RICH2,ELAC2	chr17:12.73-12.9	148K,168K	2	2	0	0.032	0.999	3	1					1	0	1	0
MKKS,C20orf94	chr20:10.24-10.37	131K	1	1	0	0.184	1.000	3	0	1	0			0	0	0	0
CRYBB2	chr22:23.95-24.4	452K	1	1	0	0.180	1.000	2	0	1	0			0	1	1	0
DUPLICATIONS observed in < 75% of narrow-diagnosis siblings in carrier families																	
MAST2,PIK3R3	chr1:46.17-46.35	58K, 175K	2	2	0	0.034	0.999	2	3					1	2	8	1
SRBD1,PRKCE	chr2:45.26-45.83	135K,527K,563K	4	3	0	0.006	0.358	7	3			3	2	4	7	3	1
COBLL1	chr2:165.34-165.45	103K,110K	2	2	0	0.035	0.999	3	2			4	2	0	0	0	1
TRIM50,FKBP6	chr7:72.34-72.46	102K-142K	2	0	0	1.000	1.000	3	2					1	0	6	2
LILRB2***; LILRB3, LILRB5, LILRA3 ,LILRA5##	chr19:59.42-59.54	30K,110K,114K	7	3	1	0.019	0.999	6	3	1	0	1	1	5	0	6	16

See legend on the next page.

Table S9 legend. The table provides information about the CNV regions that were selected as potential candidates for association with schizophrenia, based on uncorrected p-values < 0.05 in the EUR case-control analysis, or control counts of 0-1 and a clear excess in all narrow+broad cases. For these potential candidate CNVs, we examined within-family segregation and case-control counts in ISC and MGS datasets. Note that, in order to search as systematically as possible for new "candidate" CNVs, we carrfor candidate CNV analyses was repeated using a "broad" diagnoses (other non-affective psychoses; schizotypal/paranoid personality disorders) that were available for the Australia/US, Paris and VCU/Ireland datasets.

"Gene(s)" lists the gene or genes in which exons are disrupted by the typical CNVs in the region (for those with variable length, there is some variability in which genes are disrupted).

"Size(s)" are the CNV length or lengths observed in carriers.

"Fams" is the number of families in which at least one individual carried the CNV.

For the European-ancestry case-control analysis, shown are the numbers of cases and of controls carrying the CNV, and the uncorrected ("point-wise") and corrected ("genome-wide") empirical P-values as determined by PLINK based on permutations of case-control status. Note that there are sometimes fewer CNV-carrying cases than families, because only one proband per family was included in the case-control analysis, and a different case in the family could have carried the CNV.

The next set of columns summarize within-family segregation with disease, for all ancestries. Shown are counts of siblings with and without the CNV, for those with Narrow diagnoses, Broad diagnoses (not including Narrow), and Unaffected siblings.

Case and control counts for these CNVs are shown for the ISC and MGS dataset, except that the publicly-available data for ISC includes only CNVs >100kb in length, so no ISC results are shown for shorter CNVs.

None of these CNVs produced genome-wide significant evidence for association in the family dataset. Larger datasets will be required to determine whether any are associated with schizophrenia.

† Genes include TXNIP, POLR3GL, ANKRD34A, LIX1L, RBM8A, PEX11B, ITGA10, ANKRD35, PIAS3, NUDT17, POLR3C; for ISC, counts exclude the 2 cases with long schizophrenia-associated 1q21 deletions extending to this more proximal region. ‡non-Eur.

* MGS: 4 EA, 1 AA case; lower case-control ratios distally (6:4,18:11).

** MGS: EA aff, AA unaff. #MGS 4:0 distally, 4EA, 4AA cases, 1 AA cont.

ISC 9:0, MGS 4:5 more distally.

*** Counts shown are for LILRB2.

Table S9: Functional annotation of genes in candidate CNVs

Gene	Description	Chr	Strand	Gene_start	Gene_end	Band	UniProt_Function
TXNIP	thioredoxin interacting protein	1	+	119402349	119406521	1q21.1	May act as an oxidative stress mediator by inhibiting thioredoxin activity or by limiting its bioavailability. Required for the maturation of natural killer cells
POLR3GL	polymerase (RNA) III (DNA directed) polypeptide	1	-	119420102	119434231	1q21.1	
ANKRD34A	ankyrin repeat domain 34	1	+	119434352	119439409	1q21.1	
LIX1L	Lix1 homolog (mouse) like	1	+	119440847	119462785	1q21.1	
RBM8A	RNA binding motif protein 8A	1	+	119471328	119475134	1q12	Component of a splicing-dependent multiprotein exon junction complex (EJC) deposited at splice junction on mRNAs.
PEX11B	peroxisomal biogenesis factor 11B	1	+	119479859	119487437	1q21.1	Involved in peroxisomal proliferation. May regulate peroxisomes division by recruiting the dynamin-related GTPase DNM1L to the peroxisomal membrane
ITGA10	integrin, alpha 10 precursor	1	+	119488695	119507577	1q21	Integrin alpha-10/beta-1 is a receptor for collagen
ANKRD35	ankyrin repeat domain 35	1	+	119512931	119532243	1q21.1	
PIAS3	protein inhibitor of activated STAT, 3	1	+	119539725	119550284	1q21	Functions as an E3-type small ubiquitin-like modifier (SUMO) ligase. Plays a crucial role as a transcriptional coregulation in various cellular pathways, including the STAT pathway and the steroid hormone signaling pathway.
NUDT17	nudix (nucleoside diphosphate linked moiety	1	-	119550229	119553173	1q21.1	Probably mediates the hydrolysis of some nucleoside diphosphate derivatives (By similarity)
POLR3C	polymerase (RNA) III (DNA directed) polypeptide	1	-	119556179	119574446	1q21.1	DNA-dependent RNA polymerase catalyzes the transcription of DNA into RNA. Plays a key role in sensing and limiting infection by intracellular bacteria and DNA viruses.
LY75	lymphocyte antigen 75	2	-	152542810	152644950	2q24	Acts as an endocytic receptor to direct captured antigens from the extracellular space to a specialized antigen-processing compartment (By similarity). Causes reduced proliferation of B-lymphocytes.
SGCD	delta-sarcoglycan	5	+	150847340	151288441	5q33-q34	Component of the sarcoglycan complex, a subcomplex of the dystrophin-glycoprotein complex which forms a link between the F-actin cytoskeleton and the extracellular matrix
ISPD	Isoprenoid synthase domain-containing protein	7	-	16012324	16460947	7p21.2	
SOSTDC1	sclerostin domain containing 1 precursor	7	-	16387637	16392005	7p21.1	May be involved in the onset of endometrial receptivity for implantation/sensitization for the decidual cell reaction. Enhances Wnt signaling and inhibits TGF-beta signaling (By similarity).
PSD3	ADP-ribosylation factor guanine nucleotide	8	-	16929317	17412683	8p21.3	Guanine nucleotide exchange factor for ARF6 (By similarity)
GOLSYN	hypothetical protein FLJ20366	8	-	105908387	106025974	8q23.2	Part of a kinesin motor-adapter complex that is critical for the anterograde axonal transport of active zone components and contributes to activity-dependent presynaptic assembly during neuronal development (By similarity)
KCNV1	potassium channel, subfamily V, member 1	8	-	106301223	106308930	8q23.2	Potassium channel subunit that does not form functional channels by itself. Modulates KCNB1 and KCNB2 channel activity by shifting the threshold for inactivation to more negative values and by slowing the rate of inactivation.
NUDT7	nudix (nucleoside diphosphate linked moiety X)-type motif 7	16	+	63512561	63532312	16q23.1	Coenzyme A diphosphatase which mediates the cleavage of CoA, CoA esters and oxidized CoA with similar efficiencies.
TCF4	transcription factor 4	18	-	49599225	49966085	18q21.1	Transcription factor that binds to the immunoglobulin enchancer Mu-E5/KE5-motif.

ZNF100	zinc finger protein 100	19	-	21446567	21490144	19p12	May be involved in transcriptional regulation
ZNF43	zinc finger protein 43 (HTF6)	19	-	21526978	21558296	19p13.1-p12	May be involved in transcriptional regulation
ZNF600	zinc finger protein 600	19	-	49596961	49618126	19q13.41	May be involved in transcriptional regulation
ZNF28	zinc finger protein 28 (KOX 24)	19	-	49628742	49652685	19q	May be involved in transcriptional regulation
ZNF468	zinc finger protein ZNF468	19	-	49669384	49688491	19q13.41	May be involved in transcriptional regulation
APOL3	apolipoprotein L3	22	-	19504519	19530392	22q13.1	May affect the movement of lipids in the cytoplasm or allow the binding of lipids to organelles
PRSS35	protease, serine, 35	6	+	84222194	84235423	6q14.2	
PACRG	parkin co-regulated gene protein	6	+	160600313	161189795	6q26	Suppresses cell death induced by accumulation of unfolded Pael receptor (Pael-R, a substrate of Parkin). May play an important role in the formation of Lewy bodies and protection of dopaminergic neurons against Parkinson disease.
C9orf11	Acr formation associated factor	9	-	27237155	27249644	9p21	Involved in acrosome biogenesis (By similarity)
MOBKL2B	MOB1, Mps One Binder kinase activator-like 2B	9	-	27277716	27529779	9p21.2	May regulate the activity of kinases (By similarity)
FBLN2	fibulin 2 precursor,	3	+	13524043	13679922	3p25.1	Its binding to fibronectin and some other ligands is calcium dependent
LSM8	U6 snRNA-associated Sm-like protein LSm8	7	+	112190045	112210037	7q31.1-q31.3	Binds specifically to the 3'-terminal U-tract of U6 snRNA
ANKRD7	ankyrin repeat domain 7	7	+	112230622	112248684	7q31	
MSR1	macrophage scavenger receptor 1	8	-	14509923	14594638	8p22	Membrane glycoproteins implicated in the pathologic deposition of cholesterol in arterial walls during atherogenesis. Two types of receptor subunits exist. These receptors mediate the endocytosis of a diverse group of macromolecules, including modified low density lipoproteins (LDL).
WDR89	WD repeat domain 89	14	-	44230434	44274925	14q23.2	
SGPP1	sphingosine-1-phosphatase	14	-	44317325	44361142	14q23.2	Has enzymatic activity against both sphingosine 1-phosphate (S1P) and dihydro-S1P. Regulates intracellular and extracellular S1P levels
ADAMTS17	ADAM metallopeptidase with thrombospondin type 1	15	-	76635097	77005861	15q24	
RICH2	hypothetical protein LOC9912	17	+	12588245	12894960	17p12	GTPase activator for the Rho-type GTPases by converting them to an inactive GDP-bound state.
ELAC2	elaC homolog 2	17	-	12790637	12816700	17p11.2	Zinc phosphodiesterase, which displays some tRNA 3'-processing endonuclease activity.
MKKS	McKusick-Kaufman syndrome protein	20	-	10337605	10366635	20p12	Probable molecular chaperone. Assists the folding of proteins upon ATP hydrolysis.
C20orf94	hypothetical protein LOC128710	20	+	10367720	10617477	20p12.2	
CRYBB2	crystallin, beta B2	22	+	8563281	8575505	22q11.2-q12.1|22q11.23	Crystallins are the dominant structural components of the vertebrate eye lens
MAST2	microtubule associated serine/threonine kinase	1	+	44384241	44616807	1p34.1	Appears to link the dystrophin/utrophin network with microtubule filaments via the syntrophins.
PIK3R3	phosphoinositide-3-kinase, regulatory subunit 3	1	-	44620825	44713687	1p34.1	Binds to activated (phosphorylated) protein-tyrosine kinases through its SH2 domain and regulates their kinase activity. During insulin stimulation, it also binds to IRS-1
SRBD1	S1 RNA binding domain 1	2	-	45353674	45576490	2p21	
PRKCE	protein kinase C, epsilon	2	+	45617217	46415129	2p21	PKC is activated by diacylglycerol which in turn phosphorylates a range of cellular proteins. PKC also serves as the receptor for phorbol esters, a class of tumor promoters

COBLL1	COBL-like 1	2	-	157423744	157580438	2q24.3	
TRIM50	tripartite motif protein 50A	7	-	68608277	68623822	7q11.23	E3 ubiquitin-protein ligase
FKBP6	FK506-binding protein 6	7	+	68623892	68654367	7q11.23	PPIases accelerate the folding of proteins
LILRB2	leukocyte immunoglobulin-like receptor,	19	-	51105316	51112671	19q13.4	Receptor for class I MHC antigens. Involved in the down-regulation of the immune response and the development of tolerance.
LILRB3	leukocyte immunoglobulin-like receptor	19	-	54720147	54746602	19q13.4	May act as receptor for class I MHC antigens
LILRB5	leukocyte immunoglobulin-like receptor	19	-	51081906	51088803	19q13.4	May act as receptor for class I MHC antigens
LILRA3	leukocyte immunoglobulin-like receptor	19	-	54797644	54809952	19q13.4	May act as soluble receptor for class I MHC antigens
LILRA5	leukocyte immunoglobulin-like receptor subfamily	19	-	51140227	51146290	19q13.4	May plays a role in triggering innate immune responses. Seems not play a role for any class I MHC antigens recognition

Table S10: regions excluded from CNV analyses (HG18 locations)

(A) Telomeric and centromeric regions
(CNVs with >50% overlap with these regions were excluded)

Telomeres
chr1:121100001-128000000
chr2:91000001-95700000
chr3:89400001-93200000
chr4:48700001-52400000
chr5:45800001-50500000
chr6:58400001-63400000
chr7:57400001-61100000
chr8:43200001-48100000
chr9:46700001-60300000
chr10:38800001-42100000
chr11:51400001-56400000
chr12:33200001-36500000
chr13:13500001-18400000
chr14:13600001-19100000
chr15:14100001-18400000
chr16:34400001-40700000
chr17:22100001-23200000
chr18:15400001-17300000
chr19:26700001-30200000
chr20:25700001-28400000
chr21:10000001-13200000
chr22:9600001-16300000
chr1:1-100000
chr2:1-100000
chr3:1-100000
chr4:1-100000
chr5:1-100000
chr6:1-100000
chr7:1-100000
chr8:1-100000
chr9:1-100000
chr10:1-100000
chr11:1-100000
chr12:1-100000
chr13:1-100000
chr14:1-100000
chr15:1-100000
chr16:1-100000
chr17:1-100000
chr18:1-100000
chr19:1-100000
chr20:1-100000
chr21:1-100000
chr22:1-100000
chrX:1-100000
chrY:1-100000

Centromeres
chr1:247149719-247249719
chr2:242851149-242951149
chr3:199401827-199501827
chr4:191173063-191273063
chr5:180757866-180857866
chr6:170799992-170899992
chr7:158721424-158821424
chr8:146174826-146274826
chr9:140173252-140273252
chr10:135274737-135374737
chr11:134352384-134452384
chr12:132249534-132349534
chr13:114042980-114142980
chr14:106268585-106368585
chr15:100238915-100338915
chr16:88727254-88827254
chr17:78674742-78774742
chr18:76017153-76117153
chr19:63711651-63811651
chr20:62335964-62435964
chr21:46844323-46944323
chr22:49591432-49691432

(B) Regions containing immunoglobulin or olfactory receptor gene clusters
(CNVs with any overlap with these regions were excluded)

Olfactory receptor gene clusters
chr1:58953-611897
chr1:156634935-157014049
chr1:157550083-157772421
chr1:245680953-246912228
chr3:99288706-99700165
chr5:180726893-180727832
chr6:27987002-28033939
chr6:29119968-29664724
chr7:142433408-142460501
chr7:143263258-143458071
chr7:143559936-143587654

chr7:143646150-143647083
chr9:106306364-106497564
chr9:124279057-124603173
chr11:4345156-6177992
chr11:6745813-6899801
chr11:7774096-7917643
chr11:48194937-56513894
chr11:57547928-58032154
chr11:58888507-59039906
chr11:123129497-123946155
chr12:53809819-54317884
chr14:19285426-19781765
chr14:21107773-21204078
chr15:19869939-19884787
chr15:100163445-100280785
chr17:2912712-3283885
chr17:53587513-53602939
chr19:9064920-9223739
chr19:14770985-14914260
chr19:15699833-15921768

Immunoglobulin gene clusters

chr2:88937989-89411302
chr14:21159897-22090937
chr14:105065301-106352275
chr22:20715572-21595082

(C) Additional regions excluded by QC analyses (see main text)
(CNVs in these regions were excluded)

chr1:137757137-137921433
chr1:151493576-151503071
chr4:69064675-69163188
chr4:70164518-70246877
chr4:161258794-161291569
chr4:162099909-162103486
chr5:32142841-32202977
chr5:97073409-97127572
chr5:104461415-104528155
chr6:29775000-33225000
chr6:44493419-44530667
chr6:67075448-67105019
chr6:79029649-79090197
chr7:61060840-62060344
chr7:101999689-102032517
chr7:141401664-141441259
chr8:5579321-5594564
chr8:7808665-7830417
chr8:39341981-39509376
chr8:115704806-115711712
chr9:43515795-43730292
chr10:20890630-20895015
chr10:47197304-47211888
chr11:81181640-81194909
chr12:31149819-31302088
chr13:56638784-56680301
chr15:22125445-22288804
chr15:32505886-32563312
chr16:32310544-32474045
chr16:32505196-32530051
chr16:33744011-33820307
chr16:54390068-54420550
chr19:20413668-20507068
chr19:32455280-32810457
chr19:48383158-48498835
chr20:52080333-52088118
chr21:13391465-13425083
chr22:22676385-22715105

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