

Target Genes of Autism Risk Loci in Brain Frontal Cortex

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Supplementary Material

Supplementary Table 1. Candidate ASD target genes that showed significant differential expression in microarray datasets.

ASD SNP	GWAS P-value	GWAS reference	Target gene	eQTL P-value	Microarray P-value
rs8321	8.00E-09	[1]	<i>ABCF1</i>	0.029	0.0379
rs2535629	3.00E-12	[1]	<i>ABHD14A</i>	0.015	0.0466
rs11191419	1.00E-16	[2]	<i>ACTR1A</i>	0.014	0.0249
rs8009147	2.00E-09	[2]	<i>ADSSL1</i>	0.0029	0.0315
rs1104918	6.00E-06	[1]	<i>AEN</i>	0.026	0.0176
rs6453278	5.00E-06	[3]	<i>AGGF1</i>	0.019	0.0444
rs11587682	7.00E-06	[1]	<i>APH1A</i>	0.037	0.0166
rs72687362	4.00E-09	[2]	<i>ARC</i>	0.013	0.0499
rs1080500	6.00E-09	[2]	<i>CACNA2D3</i>	0.0081	0.0334
rs4702	1.00E-10	[2]	<i>CIB1</i>	0.033	0.0388
rs115558405	2.00E-13	[2]	<i>CLIC1</i>	0.0018	0.0415
rs4773054	5.00E-08	[4]	<i>COL4A1</i>	0.0034	0.0117
rs12045323	7.00E-06	[5]	<i>DHRS3</i>	0.0092	0.0424
rs6071524	1.00E-09	[2]	<i>DHX35</i>	0.047	0.0343
rs12704290	1.00E-10	[2]	<i>DMTF1</i>	0.037	0.0443
rs11210892	4.00E-10	[2]	<i>DPH2</i>	0.0024	0.0315
rs7711337	8.00E-07	[6]	<i>GABRG2</i>	0.019	0.0315
rs171748	6.00E-09	[2]	<i>GNL3L</i>	0.048	0.0335
rs7184114	2.00E-08	[2]	<i>GPR56</i>	0.023	0.0173
rs2851447	8.00E-12	[2]	<i>HIP1R</i>	0.018	0.0315
rs75782365	8.00E-24	[2]	<i>HIST1H1C</i>	0.0015	0.0316
rs2021722	2.00E-12	[1]	<i>HLA-A</i>	0.022	0.0136
rs385492	4.00E-09	[2]	<i>HLA-F</i>	0.00097	0.002
rs4150167	3.00E-07	[6]	<i>HSDL1</i>	NaN	0.0477
rs3132581	2.00E-07	[1]	<i>HSPA1A</i>	0.0069	0.00247
rs3132581	2.00E-07	[1]	<i>HSPA1B</i>	0.0058	0.0121
rs548181	9.00E-07	[1]	<i>HYLS1</i>	0.041	0.0203
rs4905226	4.00E-06	[7]	<i>IFI27</i>	0.0011	0.0428
rs4650608	1.00E-06	[1]	<i>IFI44</i>	0.013	0.0315
rs1080500	6.00E-09	[2]	<i>IL17RB</i>	0.017	0.00776
rs17292804	6.00E-09	[2]	<i>INF2</i>	0.024	0.0401

rs2233375	4.00E-06	[8]	<i>INPP5D</i>	0.011	0.0104
rs609412	2.00E-06	[1]	<i>LARS</i>	0.015	0.0219
rs12871532	1.00E-06	[1]	<i>LIG4</i>	0.048	0.0445
rs6538761	2.00E-06	[9]	<i>LTA4H</i>	0.031	0.0314
rs11587682	7.00E-06	[1]	<i>MCL1</i>	0.011	0.0374
rs221902	4.00E-08	[2]	<i>MED6</i>	0.016	0.0303
rs73416724	3.00E-08	[2]	<i>MRPL2</i>	0.0067	0.0194
rs11587682	7.00E-06	[1]	<i>MTMR11</i>	0.026	0.0329
rs7700191	4.00E-06	[1]	<i>NFKB1</i>	0.015	0.0137
rs7254215	7.00E-06	[1]	<i>NOTCH3</i>	0.036	0.0344
rs1550976	2.00E-06	[8]	<i>NTM</i>	0.0099	0.0346
rs169738	8.00E-10	[2]	<i>NUDT3</i>	0.0036	0.0104
rs7914558	2.00E-09	[1]	<i>OBFC1</i>	0.0018	0.0172
rs760648	3.00E-08	[2]	<i>PACSIN2</i>	0.0055	0.0346
rs11735612	4.00E-06	[5]	<i>PCDH18</i>	0.005	0.0147
rs7746199	1.00E-20	[2]	<i>PGBD1</i>	0.017	0.0419
rs133047	1.00E-08	[2]	<i>PHF5A</i>	0.0082	0.0499
rs1797052	8.00E-09	[10]	<i>POLR3C</i>	0.03	0.0248
rs7700191	4.00E-06	[1]	<i>PPP3CA</i>	0.0018	0.0427
rs4702	1.00E-10	[2]	<i>PRC1</i>	0.01	0.0465
rs11587682	7.00E-06	[1]	<i>PRPF3</i>	0.017	0.0145
rs8054556	4.00E-10	[2]	<i>PRR14</i>	0.042	0.02
rs3849046	2.00E-09	[2]	<i>REEP2</i>	0.024	0.0359
rs2332700	2.00E-09	[2]	<i>RGS6</i>	0.024	0.042
rs116254153	4.00E-11	[2]	<i>RING1</i>	0.0015	0.0173
rs76994193	4.00E-08	[2]	<i>RPS6</i>	0.037	0.0349
rs1080500	6.00E-09	[2]	<i>SELK</i>	0.012	0.0285
rs4905226	4.00E-06	[7]	<i>SERPINA3</i>	0.014	0.0376
rs8054556	4.00E-10	[2]	<i>SEZ6L2</i>	0.014	0.0404
rs7914558	2.00E-09	[1]	<i>SH3PXD2A</i>	0.013	0.0277
rs12826178	4.00E-10	[2]	<i>SHMT2</i>	0.0084	0.0427
rs2898883	2.00E-08	[11]	<i>SPOP</i>	0.0081	0.0192
rs548181	9.00E-07	[1]	<i>SRPR</i>	0.035	0.0362
rs114882497	2.00E-16	[2]	<i>TAP1</i>	0.0012	0.0137
rs880446	4.00E-08	[2]	<i>TCEA2</i>	0.02	0.0319
rs2297909	2.00E-06	[1]	<i>TNNT2</i>	0.0099	0.0308
rs9607782	9.00E-09	[2]	<i>TNRC6B</i>	0.014	0.0474
rs2851447	8.00E-12	[2]	<i>VPS37B</i>	0.047	0.0499
rs72934570	1.00E-09	[2]	<i>WDR7</i>	0.047	0.0401
rs133047	1.00E-08	[2]	<i>XRCC6</i>	0.00082	0.0453
rs11210892	4.00E-10	[2]	<i>YBX1</i>	0.0013	0.0269
rs880446	4.00E-08	[2]	<i>ZBTB46</i>	0.035	0.0211
rs10255295	5.00E-06	[1]	<i>ZC3HAV1</i>	0.011	0.0315
rs12887734	2.00E-12	[2]	<i>ZFYVE21</i>	0.046	0.0226

rs56223946	3.00E-09	[2]	ZNF395	0.0097	0.0171
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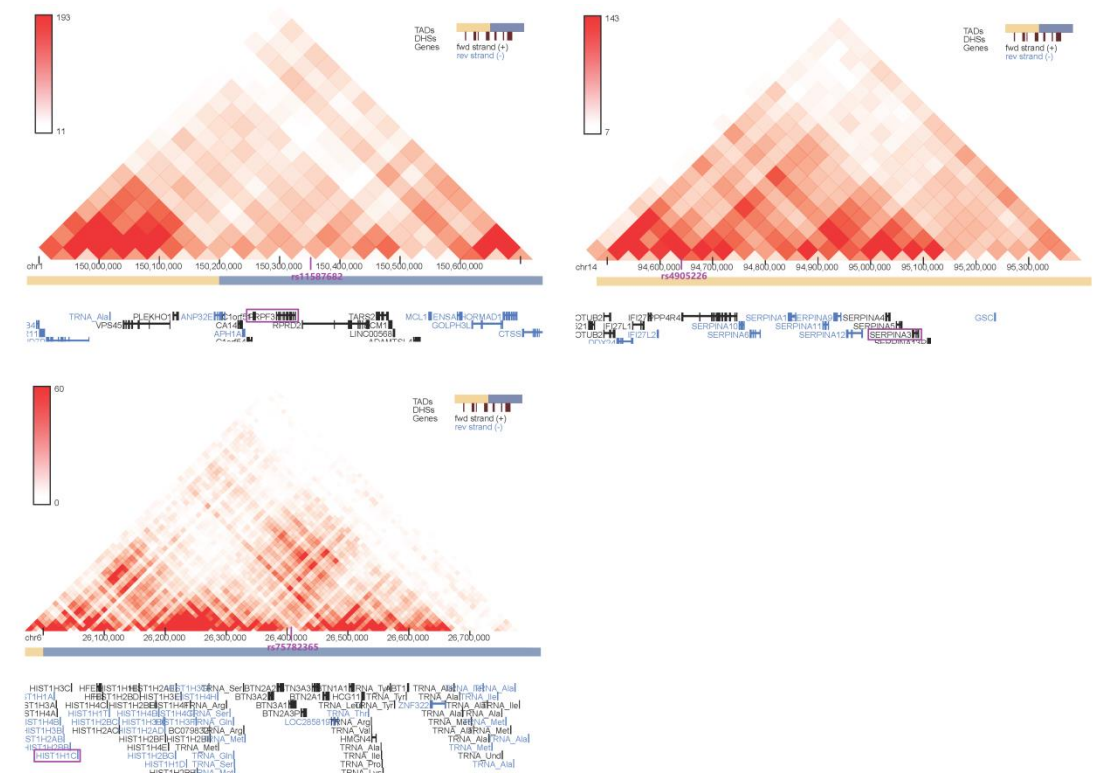
SNP = single nucleotide polymorphism; GWAS P-value = P-value of each ASD SNP in their original GWAS study; GWAS reference = reference for each ASD GWAS SNP; Target Gene = candidate target gene identified for each ASD GWAS SNP; eQTL P-value = P-value of correlation between gene expression level and GWAS SNP genotype in GTEx database or Braineac database; Microarray P-value = P-value of the differentially expressed genes in microarray meta-analysis.

Supplementary Table 2. Pathways significantly enriched among the 76 genes. The analyses were performed using web portal DAVID [12, 13] and PANTHER[14].

Web Portal	Pathway Database	Enriched Pathway	Count	Genes	P-value	Adjusted P-value
DAVID	KEGG	Antigen processing and presentation	5	HSPA1A, HSPA1B, HLA-A, HLA-F, TAP1	3.00E-04	3.60E-02
PANTHER	Reactome	Noncanonical activation of NOTCH3	3	APH1A, YBX1, NOTCH3	7.53E-06	1.65E-02

Count = The number of potential ASD target genes involved in each pathway; P-value = nominal P-value; Adjusted P-value = P-value after multiple testing adjustment.

Supplementary Figure 1. The ASD SNPs and target genes locate in same topologically associating domain (TAD) as reported in brain Hi-C data.



Reference

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