

Supplementary Data

Exome sequencing of extended families with Alzheimer's disease identifies novel genes implicated in cell immunity and neuronal function

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The authors have no conflict of interest to declare.

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Supplementary Tables

Supplementary Table 1. LOAD Families

family	diagnosed AD & MCI family members	Individuals Exome Sequenced		
		AD cases	MCI cases	unaffected relatives
191	5	2	0	0
419	6	2	0	1
609	7	2	0	1
701	9	3	0	0
713	9	6	0	1
716	6	3	0	1
757	14	10	1	1
803	6	2	1	0
911	11	7	0	1
974	11	7	2	0
1008	4	2	0	1
1086	9	4	0	3
1201	16	5	0	0
1240	6	2	0	1
1399	9	2	0	1
1743	7	2	0	1
1893	5	3	0	0
2119	6	2	0	0
2120	5	2	0	1
2349	5	2	0	1
26044	7	3	0	1
26284	6	2	0	1
33035	7	2	0	1
176		77	4	18

Supplementary Table 2. Known AD genes and loci

gene	chr	reference
<i>ABCA7</i>	19	Lambert, et al, Nat Genet, 2013
<i>AKAP9</i>	7	Logue, et al, Alzheimers Dement, 2014
<i>APOE</i>	19	Corder, et al, Science, 1993
<i>APP</i>	21	Goate, et al, Nature, 1991
<i>BIN1</i>	2	Lambert, et al, Nat Genet, 2013
<i>CASS4</i>	20	Lambert, et al, Nat Genet, 2013
<i>CD2AP</i>	6	Lambert, et al, Nat Genet, 2013
<i>CD33</i>	19	Lambert, et al, Nat Genet, 2013
<i>CELF1</i>	11	Lambert, et al, Nat Genet, 2013
<i>CLU</i>	8	Lambert, et al, Nat Genet, 2013
<i>CR1</i>	1	Brouwers, et al, Mol Psychiatry, 2012
<i>EPHA1</i>	7	Lambert, et al, Nat Genet, 2013
<i>FERMT2</i>	14	Lambert, et al, Nat Genet, 2013
<i>INPP5D</i>	2	Lambert, et al, Nat Genet, 2013
<i>MADD</i>	11	Lambert, et al, Nat Genet, 2013
<i>MAPT</i>	17	Jun, et al, Molec Psychiatry, 2015
<i>MEF2C</i>	5	Lambert, et al, Nat Genet, 2013
<i>MS4A2</i>	11	Lambert, et al, Nat Genet, 2013
<i>MS4A3</i>	11	Lambert, et al, Nat Genet, 2013
<i>MS4A4A</i>	11	Lambert, et al, Nat Genet, 2013
<i>MS4A4E</i>	11	Lambert, et al, Nat Genet, 2013
<i>MS4A6A</i>	11	Lambert, et al, Nat Genet, 2013
<i>MS4A6E</i>	11	Lambert, et al, Nat Genet, 2013
<i>NME8</i>	7	Lambert, et al, Nat Genet, 2013
<i>NYAP1</i>	7	Lambert, et al, Nat Genet, 2013
<i>PICALM</i>	11	Lambert, et al, Nat Genet, 2013
<i>PSEN1</i>	14	Sherrington, et al, Nature, 1995
<i>PSEN2</i>	1	Levy-Lahad, et al, Science, 1995
<i>PTK2B</i>	8	Lambert, et al, Nat Genet, 2013
<i>RIN3</i>	14	Lambert, et al, Nat Genet, 2013
<i>SLC24A4</i>	14	Lambert, et al, Nat Genet, 2013
<i>SORL1</i>	11	Rogaeva, et al, Nat Genet, 2007
<i>TREM2</i>	6	Lambert, et al, Nat Genet, 2013
<i>UNC5C</i>	4	Wetzel-Smith, et al, Nat Med, 2014
<i>ZCWPW1</i>	7	Lambert, et al, Nat Genet, 2013

Supplementary Table 3. SNPs tested in the ADGC

dbSNP	MAF	effect	StdErr	p-value	HetPVal
rs150893072	0.0103	0.0508	0.2508	0.8396	0.6873
rs143332484	0.007	0.0797	0.3541	0.8219	0.4185
rs17125721	0.026	-0.0224	0.1353	0.8684	0.0826
rs41274770	0.021	-0.0971	0.2098	0.6436	0.0113
rs118152239	0.0126	-0.0193	0.1568	0.9020	0.5985
rs34585936	0.0219	0.2669	0.1735	0.1240	1.0000
rs62001869	0.0129	0.1814	0.194	0.3497	0.0216
rs11768549	0.0153	0.0388	0.1884	0.8369	0.8544
rs142692233	0.0563	-0.1794	0.1169	0.1250	0.3298
rs2232088	0.0158	0.1606	0.1997	0.4213	0.0020

Supplementary Table 4. Genes tested in the ADGC

gene	chr	start	end	SNPs	p-value
<i>CD163L1</i>	12	7499281	7632493	169	0.6139
<i>CD33</i>	19	51728320	51747115	64	0.7228
<i>CLECL1</i>	12	9868456	9885895	40	0.6040
<i>CR1</i>	1	207669492	207813992	134	0.9010
<i>CTNNA1</i>	5	137946656	138270723	284	0.0990
<i>DAAM2</i>	6	39760142	39872648	193	0.1782
<i>EPHA1</i>	7	143087382	143105985	71	0.2970
<i>MIEF1</i>	22	39895437	39914137	57	0.0495
<i>MKL2</i>	16	14165178	14360630	203	0.5149
<i>NME8</i>	7	37888199	37940003	106	0.8911
<i>PLEKHG5</i>	1	6526152	6580121	95	0.1485
<i>PSEN1</i>	14	73603126	73690399	119	0.2376
<i>SORL1</i>	11	121322912	121504402	231	0.1089