

Supplementary Table 2. Enrichment of GO terms and KEGG pathways of the 58 MS-associated genes

Category	Term	Count	%	Genes	List total	Relative enrichment	Bonferroni*
GOTERM_BP_FAT	GO:0019882~antigen processing and presentation	12	21.43	3112, 285830, 3117, 3118, 6891, 3107, 3109, 3108, 3134, 3105, 3132, 3122	25	78.23	1.09×10^{-16}
GOTERM_CC_FAT	GO:0042611~MHC protein complex	11	19.64	3112, 285830, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3132, 3122	24	102.78	1.06×10^{-16}
SP_PIR_KEYWORDS	mhc ii	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3105, 3132, 3122	34	164.25	1.84×10^{-14}
UP_SEQ_FEATURE	Region of interest:Connecting peptide	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	33	137.17	3.39×10^{-13}
UP_SEQ_FEATURE	Domain:Ig-like C1-type	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	33	130.32	5.42×10^{-13}
KEGG_PATHWAY	hsa05330:Allograft rejection	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	14	90.80	7.23×10^{-14}
KEGG_PATHWAY	hsa05332:Graft-versus-host disease	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	14	83.82	1.47×10^{-13}
KEGG_PATHWAY	hsa04940:Type I diabetes mellitus	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	14	77.83	2.80×10^{-13}
KEGG_PATHWAY	hsa04612:Antigen processing and presentation	10	17.86	3112, 3117, 3118, 6891, 3107, 3109, 3108, 3134, 3105, 3122	14	43.76	7.53×10^{-13}
KEGG_PATHWAY	hsa05320:Autoimmune thyroid disease	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	14	64.10	1.50×10^{-12}
INTERPRO	IPR003597:Immunoglobulin C1-set	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	29	76.03	1.16×10^{-11}
UP_SEQ_FEATURE	Region of interest:Alpha-1	7	12.50	3117, 3118, 3107, 3108, 3134, 3105, 3122	33	202.71	1.56×10^{-10}
UP_SEQ_FEATURE	Region of interest:Alpha-2	7	12.50	3117, 3118, 3107, 3108, 3134, 3105, 3122	33	202.71	1.56×10^{-10}
INTERPRO	IPR014745:MHC class II, alpha/beta chain, N-terminal	7	12.50	3112, 3117, 3118, 3109, 3108, 3132, 3122	29	182.78	7.35×10^{-11}
INTERPRO	IPR003006:Immunoglobulin/major histocompatibility complex, conserved site	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	29	59.43	8.98×10^{-11}
KEGG_PATHWAY	hsa05416:Viral myocarditis	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	14	46.04	2.47×10^{-11}
SMART	SM00407:IGc1	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	24	50.07	7.27×10^{-11}
GOTERM_CC_FAT	GO:0042613~MHC class II protein complex	7	12.50	3112, 3117, 3118, 3109, 3108, 3132, 3122	24	128.55	5.56×10^{-10}
GOTERM_BP_FAT	GO:0002504~antigen processing and presentation of peptide or polysaccharide antigen via MHC class II	7	12.50	3112, 3117, 3118, 3109, 3108, 3132, 3122	25	114.78	3.42×10^{-9}
GOTERM_MF_FAT	GO:0032395~MHC class II receptor activity	6	10.71	3112, 3117, 3118, 3107, 3108, 3122	23	178.26	7.85×10^{-9}
SP_PIR_KEYWORDS	Immune response	10	17.86	3112, 3117, 3118, 6891, 3107, 3109, 3108, 3134, 3105, 3122	34	25.26	8.42×10^{-9}
KEGG_PATHWAY	hsa04514:Cell adhesion molecules (CAMs)	9	16.07	3112, 3117, 3118, 3107, 3109, 3108, 3134, 3105, 3122	14	24.76	4.04×10^{-9}
GOTERM_CC_FAT	GO:0042825~TAP complex	5	8.93	3112, 6891, 3109, 3108, 3122	24	380.42	2.00×10^{-8}
GOTERM_CC_FAT	GO:0042824~MHC class I peptide loading complex	5	8.93	3112, 6891, 3109, 3108, 3122	24	295.88	7.18×10^{-8}
GOTERM_BP_FAT	GO:0048002~antigen processing and presentation of peptide antigen	6	10.71	6891, 3107, 3108, 3134, 3105, 3122	25	115.95	2.17×10^{-7}
PIR_SUPERFAMILY	PIRSF001991:Class II histocompatibility antigen	6	10.71	3112, 3117, 3118, 3109, 3108, 3122	18	94.82	2.79×10^{-8}
INTERPRO	IPR007110:Immunoglobulin-like	11	19.64	3112, 3117, 3118, 4340, 3107, 3109, 3108, 3134, 56244, 3105, 3122	29	12.61	3.53×10^{-7}
KEGG_PATHWAY	hsa05310:Asthma	6	10.71	3112, 3117, 3118, 3109, 3108, 3122	14	75.15	1.10×10^{-7}
GOTERM_BP_FAT	GO:0006955~immune response	12	21.43	3112, 285830, 3117, 3118, 6891, 3107, 3109, 3108, 3134, 3105, 3132, 3122	25	9.41	1.53×10^{-6}
GOTERM_MF_FAT	GO:0042288~MHC class I protein binding	5	8.93	3112, 6891, 3109, 3108, 3122	23	176.40	8.88×10^{-7}
INTERPRO	IPR013783:Immunoglobulin-like fold	11	19.64	3112, 3117, 3118, 4340, 3107, 3109, 3108, 3134, 56244, 3105, 3122	29	11.43	9.06×10^{-7}

Supplementary Table 2. Enrichment of GO terms and KEGG pathways of the 58 MS-associated genes (continued)

Category	Term	Count	%	Genes	List total	Relative enrichment	Bonferroni*
GOTERM_MF_FAT	GO:0042287~MHC protein binding	5	8.93	3112, 6891, 3109, 3108, 3122	23	112.90	6.11×10^{-6}
KEGG_PATHWAY	hsa04672:Intestinal immune network for IgA production	6	10.71	3112, 3117, 3118, 3109, 3108, 3122	14	44.48	1.72×10^{-6}
SP_PIR_KEYWORDS	Heterodimer	6	10.71	3117, 3118, 3107, 3134, 3105, 3122	34	32.96	6.98×10^{-5}
SP_PIR_KEYWORDS	Transmembrane protein	10	17.86	3112, 3117, 3118, 4340, 6891, 3107, 3108, 3134, 3105, 3122	34	8.81	7.59×10^{-5}
INTERPRO	IPR001003:MHC class II, alpha chain, N-terminal	4	7.14	3117, 3118, 3108, 3122	29	191.48	7.32×10^{-5}
KEGG_PATHWAY	hsa05322:Systemic lupus erythematosus	6	10.71	3112, 3117, 3118, 3109, 3108, 3122	14	22.01	6.04×10^{-5}
GOTERM_BP_FAT	GO:0002474~antigen processing and presentation of peptide antigen via MHC class I	4	7.14	6891, 3107, 3134, 3105	25	127.32	6.60×10^{-4}
GOTERM_CC_FAT	GO:0044459~plasma membrane part	15	26.79	4855, 3112, 3117, 3118, 6891, 394263, 285830, 199, 3107, 3109, 3108, 3134, 3105, 3132, 3122	24	3.63	2.43×10^{-4}
INTERPRO	IPR001039:MHC class I, alpha chain, alpha1, and alpha2	4	7.14	285830, 3107, 3134, 3105	29	85.10	9.56×10^{-4}
INTERPRO	IPR011161:MHC class I-like antigen recognition	4	7.14	285830, 3107, 3134, 3105	29	82.06	1.07×10^{-3}
GOTERM_CC_FAT	GO:0042612~MHC class I protein complex	4	7.14	285830, 3107, 3134, 3105	24	76.08	1.17×10^{-3}
BIOCARTA	h_mhcPathway:Antigen processing and presentation	3	5.36	6891, 3105, 3122	3	143.70	5.67×10^{-4}
UP_SEQ_FEATURE	Sequence variant	31	55.36	170679, 3112, 4855, 11074, 54535, 6891, 56244, 7148, 10919, 6941, 199, 6046, 83463, 5460, 3107, 3109, 3108, 3134, 3105, 401250, 3117, 3118, 4340, 29113, 8449, 10665, 7916, 394263, 25833, 3122, 7917	33	1.50	1.94×10^{-2}
SP_PIR_KEYWORDS	Polymorphism	31	55.36	170679, 3112, 4855, 11074, 54535, 6891, 56244, 7148, 10919, 6941, 199, 6046, 83463, 5460, 3107, 3109, 3108, 3134, 3105, 401250, 3117, 3118, 4340, 29113, 8449, 10665, 7916, 394263, 25833, 3122, 7917	34	1.52	7.47×10^{-3}
UP_SEQ_FEATURE	Region of interest:Alpha-3	3	5.36	3107, 3134, 3105	33	193.06	2.92×10^{-2}
SP_PIR_KEYWORDS	mhc i	3	5.36	3107, 3134, 3105	34	169.72	1.05×10^{-2}
GOTERM_BP_FAT	GO:0002478~antigen processing and presentation of exogenous peptide antigen	3	5.36	6891, 3108, 3122	25	147.58	3.25×10^{-2}
PIR_SUPERFAMILY	PIRSF001990:Class I histocompatibility antigen	3	5.36	3107, 3134, 3105	18	136.96	2.30×10^{-3}
GOTERM_BP_FAT	GO:0019884~antigen processing and presentation of exogenous antigen	3	5.36	6891, 3108, 3122	25	115.95	5.30×10^{-2}
GOTERM_MF_FAT	GO:0042277~peptide binding	5	8.93	3112, 6891, 3109, 3108, 3122	23	13.90	2.69×10^{-2}
INTERPRO	IPR000353:MHC class II, beta chain, N-terminal	3	5.36	3112, 3109, 3132	29	101.37	2.84×10^{-2}
GOTERM_MF_FAT	GO:0032393~MHC class I receptor activity	3	5.36	3107, 3134, 3105	23	99.61	2.89×10^{-2}
GOTERM_CC_FAT	GO:0005886~plasma membrane	16	28.57	4855, 3112, 3117, 3118, 4340, 6891, 394263, 285830, 199, 3107, 3109, 3108, 3134, 3105, 3132, 3122	24	2.26	2.98×10^{-2}

Count, the number of genes enriched in particular GO terms or KEGG pathways; Genes, potential genes enriched in particular GO terms or KEGG pathways.

*p value with Bonferroni correction for multiple tests. Only significant results with $p < 0.05$ are listed.

GO: Gene Ontology, KEGG: Kyoto Encyclopedia of Genes and Genomes, MHC: major histocompatibility complex, MS: multiple sclerosis.