Supplemental Data

Gene Ontology Analysis of GWAS Data Sets

Provides Insights into the Biology of Bipolar Disorder

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Tables S1 and S2 are available as Excel files and may also be found online.

Table S3. **Top 30 Overrepresented GO Categories: CD Data Set Excluding MHC**

GO category	Type	total genes in category	#genes on list	expected #genes on list	p-value	expected hits/study	Function
GO06511	PROCESS	142	5	0.56	0.0002	0.5	ubiquitin-dependent protein catabolic process
GO19941	PROCESS	142	5	0.56	0.0002	0.5	modification-dependent protein catabolic process
GO43632	PROCESS	142	5	0.56	0.0002	0.5	modification-dependent macromolecule catabolic process
GO51603	PROCESS	143	5	0.56	0.0002	0.5	proteolysis involved in cellular protein catabolic process
GO44257	PROCESS	145	5	0.56	0.0002	0.5	cellular protein catabolic process
GO 51181	PROCESS	8	2	0.02	0.0002	0.5	cofactor transport
GO15226	FUNCTION	3	2	0.02	0.0002	0.5	carnitine transporter activity
GO15879	PROCESS	3	2	0.02	0.0002	0.5	carnitine transport
GO04221	FUNCTION	53	3	0.16	0.0004	0.71	ubiquitin thiolesterase activity
GO04843	FUNCTION	56	3	0.16	0.0004	0.71	ubiquitin-specific protease activity
GO19783	FUNCTION	57	3	0.16	0.0004	0.71	small conjugating protein-specific protease activity
GO51183	FUNCTION	8	2	0.02	0.0004	0.71	vitamin transporter activity
GO51184	FUNCTION	9	2	0.04	0.0006	0.9	cofactor transporter activity
GO30163	PROCESS	177	5	0.67	0.0006	0.9	protein catabolic process
GO16790	FUNCTION	66	3	0.19	0.0006	0.9	thiolester hydrolase activity
GO51180	PROCESS	11	2	0.06	0.0014	1.61	vitamin transport
GO43285	PROCESS	240	5	0.86	0.0016	1.83	biopolymer catabolic process
GO44265	PROCESS	243	5	0.86	0.0020	2.16	cellular macromolecule catabolic process
GO06952	PROCESS	318	5	1.1	0.0028	2.95	defense response
GO06512	PROCESS	383	6	1.52	0.0032	3.28	ubiquitin cycle
GO07249	PROCESS	31	2	0.09	0.0038	3.76	I-kappaB kinase/NF-kappaB cascade
GO09057	PROCESS	291	5	1.02	0.0040	3.93	macromolecule catabolic process
GO06955	PROCESS	343	5	1.16	0.0054	5.05	immune response
GO08234	FUNCTION	117	3	0.38	0.0058	5.36	cysteine-type peptidase activity
GO44248	PROCESS	439	6	1.57	0.0066	5.98	cellular catabolic process
GO31347	PROCESS	35	2	0.13	0.0068	6.15	regulation of defense response
GO05737	CELLULAR	4531	32	22.41	0.0128	10.28	cytoplasm
GO19901	FUNCTION	52	2	0.18	0.0130	10.4	protein kinase binding
GO09056	PROCESS	534	6	1.99	0.0158	12.32	catabolic process
GO65003	PROCESS	343	5	1.48	0.0168	13	macromolecular complex assembly

List of 30 most significantly over-represented GO categories for Crohn's disease, excluding the MHC region (cutoff for significant SNPs: p<1e-4). The type of category, and the expected number of categories with a category-specific over-representation p-value at least as significant as that observed in the absence of any true over-representation, are also shown.

Table S4. Number of Significantly Overrepresented GO Categories Using 20 kb Gene Window: CD Data Set

p-value criterion for SNPs	*# top SNPs*	*#genes*	*p<0.05*		*p<0.01*		*p<0.001*	
			#cat	p	#cat	p	#cat	p
0.0001	308	140	50	0.382	27	0.075	1	0.609
0.001	1226	480	99	0.280	28	0.195	5	0.159
0.005	3905	1517	145	0.258	37	0.105	11	0.025

Number of GO categories reaching various levels of significance for over-representation on the list of significant SNPs in the WTCCC CD dataset and their corresponding genes, together with p-values indicating whether this number is significantly greater than expected by chance. Only categories containing two or more significant genes are counted. SNPs assigned to genes if they lie within 20kb of that gene. Genotyped SNPs only.

Table S5. **Number of Significantly Overrepresented GO Categories: WTCCC BD Data Set**

p-value criterion for SNPs	*# top SNPs*	*#genes*	*p<0.05*		*p<0.01*		*p<0.001*	
			#cat	p	#cat	p	#cat	p
0.0001	667	80	39	0.312	15	0.157	2	0.314
0.001	3622	351	58	0.611	13	0.556	1	0.650
0.005	15569	1261	110	0.370	18	0.535	5	0.144

Number of GO categories reaching various levels of significance for over-representation on the list of significant SNPs in the WTCCC BD dataset and their corresponding genes, together with p-values indicating whether this number is significantly greater than expected by chance. Only categories containing two or more significant genes are counted. SNPs assigned to genes if they lie within that gene. Genotyped and imputed SNPs .

Table S6. **Number of Significantly Overrepresented GO Categories: BD Meta-Analysis Data Set, Genotyped SNPs Only**

p-value criterion for SNPs	*# top SNPs*	*#genes*	*p<0.05*		*p<0.01*		*p<0.001*	
			#cat	p	#cat	p	#cat	p
0.0001	103	31	14	0.512	3	0.583	0	1
0.001	687	176	43	0.437	12	0.342	0	1
0.005	2780	633	79	0.344	20	0.213	5	0.086

Number of GO categories reaching various levels of significance for over-representation on the list of significant SNPs in the BD meta-analysis dataset and their corresponding genes, together with p-values indicating whether this number is significantly greater than expected by chance. Only categories containing two or more significant genes are counted. SNPs assigned to genes if they lie within that gene. Genotyped SNPs only.

Table S7. **Number of Significantly Overrepresented GO Categories Using 20 kb Gene Window: BD Meta-Analysis Data Set**

p-value criterion for SNPs	*# top SNPs*	*#genes*	*p<0.05*		*p<0.01*		*p<0.001*	
			#cat	p	#cat	p	#cat	p
0.0001	593	73	19	0.804	8	0.527	0	1
0.001	3759	474	118	0.116	42	0.027	0	1
0.005	15979	1737	185	0.041	74	0.001	24	<0.001
0.01	29073	2866	257	0.002	72	0.001	10	0.025

Number of GO categories reaching various levels of significance for over-representation on the list of significant SNPs in the BD meta-analysis dataset and their corresponding genes, together with p-values indicating whether this number is significantly greater than expected by chance. Only categories containing two or more significant genes are counted. SNPs assigned to genes if they lie within 20kb of that gene. Genotyped and imputed SNPs.

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Table S8. Number of Significantly Overrepresented GO Categories: CD Data Set, LD-Pruned SNPs

p-value criterion for SNPs	*# significant SNPs*	*p<0.05*		*p<0.01*		*p<0.001*	
		#cat	p	#cat	p	#cat	p
0.0001	65	53	0.129	32	0.023	17	0.003
0.001	239	73	0.344	22	0.240	2	0.437
0.005	893	130	0.231	28	0.277	4	0.267

Number of GO categories reaching various levels of significance for over-representation on the LD-pruned list of significant SNPs in the WTCCC CD dataset, together with p-values indicating whether this number is significantly greater than expected by chance. SNPs assigned to genes if they lie within that gene. Genotyped SNPs only.

Table S9. Top 30 Overrepresented GO Categories: CD Data Set, LD-Pruned SNPs

GO category	*Type*	*total genes in category*	*#sig. SNPs on list*	*expected #sig SNPs on list*	*p-value*	*expected hits/study*	*Function*
GO06955	PROCESS	335	8	1.19	0.0000	0.36	immune response
GO02376	PROCESS	454	9	1.81	0.0002	0.54	immune system process
GO04221	FUNCTION	48	3	0.15	0.0002	0.54	ubiquitin thiolesterase activity
GO04843	FUNCTION	51	3	0.15	0.0002	0.54	ubiquitin-specific protease activity
GO19783	FUNCTION	52	3	0.15	0.0002	0.54	small conjugating protein-specific protease activity
GO02504	PROCESS	9	2	0.02	0.0002	0.54	antigen processing and presentation of peptide or polysaccharide antigen via MHC class II
GO32395	FUNCTION	8	2	0.02	0.0002	0.54	MHC class II receptor activity
GO42613	CELLULAR	8	2	0.02	0.0002	0.54	MHC class II protein complex
GO06878	PROCESS	8	2	0.04	0.0004	0.77	cellular copper ion homeostasis
GO55070	PROCESS	9	2	0.04	0.0004	0.77	copper ion homeostasis
GO06511	PROCESS	126	4	0.49	0.0008	1.2	ubiquitin-dependent protein catabolic process
GO19941	PROCESS	126	4	0.49	0.0008	1.2	modification-dependent protein catabolic process
GO43632	PROCESS	126	4	0.49	0.0008	1.2	modification-dependent macromolecule catabolic process
GO51603	PROCESS	126	4	0.49	0.0008	1.2	proteolysis involved in cellular protein catabolic process
GO44257	PROCESS	128	4	0.5	0.0008	1.2	cellular protein catabolic process
GO16790	FUNCTION	60	3	0.19	0.0008	1.2	thiolester hydrolase activity
GO42611	CELLULAR	17	2	0.03	0.0010	1.39	MHC protein complex
GO06952	PROCESS	297	6	1.15	0.0012	1.58	defense response
GO45087	PROCESS	71	3	0.27	0.0020	2.3	innate immune response
GO19882	PROCESS	30	2	0.06	0.0030	3.26	antigen processing and presentation
GO30163	PROCESS	152	4	0.6	0.0032	3.43	protein catabolic process
GO08234	FUNCTION	102	3	0.33	0.0038	3.98	cysteine-type peptidase activity
GO07219	PROCESS	29	3	0.29	0.0040	4.13	Notch signaling pathway
GO07249	PROCESS	29	2	0.09	0.0040	4.13	I-kappaB kinase/NF-kappaB cascade
GO06954	PROCESS	173	4	0.66	0.0048	4.87	inflammatory response
GO04857	FUNCTION	161	4	0.69	0.0050	5.02	enzyme inhibitor activity
GO44265	PROCESS	218	4	0.8	0.0056	5.53	cellular macromolecule catabolic process
GO09986	CELLULAR	107	4	0.73	0.0060	5.82	cell surface
GO31347	PROCESS	32	2	0.12	0.0072	6.8	regulation of defense response
GO43285	PROCESS	211	4	0.8	0.0076	7.12	biopolymer catabolic process

List of 30 most significantly over-represented GO categories for Crohn's disease, based on the LD-pruned SNP map. (cutoff for significant SNPs: p<1e-4). The type of category, and the expected number of categories with a category-specific over-representation p-value at least as significant as that observed in the absence of any true over-representation, are also shown.