

**S2 Table. Overrepresented functions and diseases in the gene sets suggestively associated in joint linkage and LD analyses.**

Diseases or Functions	p-value	Genes	# genes	Phenotype
Insulin-dependent diabetes mellitus	4.33E-24	<i>ABCF1, ABCG1, ABHD16A, ACTA2, AIF1, APOM, ATAT1, BAG6, C6orf15, C6orf47, C6orf48, CBS/LOC102724560, CDSN, CFB, COL27A1, DDR1, DDX39B, DHX16, DPCRI, EHMT2, FLOT1, GPANK1, GTF2H4, HCG22, HCP5, HLA-B,HLA-C, HSPA1A/HSPA1B, HSPA1L, IFIT1B, IFIT2, IFIT3, LSM2, LST1, LTA, LY6G5B, LY6G5C, LY6G6C, LY6G6D, MCCD1, MDC1, MICA, MICB, MSH5, MUC21, MZB1,NEBL, NELFE, PCDH15, POU5F1, PRRC2A, PSORS1C1, SKIV2L, SLC44A4, STK19,TCF19,TNF, TNFSF8, TUBB, UBASH3A, VARS2,VWA7, ZBTB12</i>	63	Other creat.
Systemic autoimmune syndrome	1,01E-16	<i>ABCF1,ABCG1, ABHD16A, ACTA2, AIF1, APOM, ATAT1, BAG6, BLNK, C2, C4A/C4B, C6orf15, C6orf47, C6orf48, C9orf152, CBS/LOC102724560, CCHCR1, CDSN, CFB, CLU, COL27A1, DDR1, DDX39B, DHX16,DPCRI,EGR2,EHMT2,EPHX2, FAS, FLOT1, GNL1, GPANK1, GTF2H4, HCG22, HCG27, HCP5, HLA-B, HLA-C,HSPA1A/ HSPA1B, HSPA1L,IFIT1,IFIT1B, IFIT2, IFIT3, LSM2, LST1, LTA, LTB, LY6G5B, LY6G5C,LY6G6C,LY6G6D, MBL2, MCCD1, MDC1, MICA, MICB, MSH5, MUC21,MX1,MZB1,NCR3, NEBL, NELFE, NFKBIL1, NRM, PCDH15,PLAC4, POU5F1, PRRC2A, PSORS1C1, PTK2B, SIL1, SKIV2L, SLC44A4, STK19, TCF19, TMEM173, TNF, TNFSF11, TNFSF15, TNFSF8 ,TUBB, TXN, UBASH3A, VARS, VARS2, VIM, VWA7, ZBTB12</i>	90	Other creat.
Rheumatoid arthritis	8,08E-12	<i>AIF1, APOM, ATAT1, BAG6, BLNK, C2, C4A/C4B, C6orf15, C6orf47, C9orf152, CCHCR1, CFB, CLU, DDR1, DDX39B, DHX16, DPCRI, EHMT2, EPHX2, GNL1, GPANK1, GTF2H4, HCG22, HCG27, HLA-C, HSPA1A/ HSPA1B, HSPA1L, LST1, LTA,LTB, LY6G5C, LY6G6C, LY6G6D, MBL2, MCCD1, MDC1, MICA, MSH5, NELFE, NFKBIL1, NRM, PCDH15, PLAC4,PRRC2A,PSORS1C1,PTK2B, SIL1, SKIV2L, SLC44A4, STK19,TMEM173,TNF, TNFSF11, TNFSF15, TXN ,VARS, VARS2,VIM, ZBTB12</i>	59	Other creat.
Chronic inflammatory disease	9,56E-09	<i>ACTA2, ADRA1A, AIF1, APOM, ATAT1, BAG6, BLNK, BM11, C2, C4A/ C4B, C6orf15, C6orf47, C9orf152, CCHCR1, CFB, CLU, DDR1, DDX39B, DHX16, DPCRI, DSCI, EHMT2, EPHX2, FAS, GNL1, GPANK1, GTF2H4, HCG22, HCG27, HGF, HLA-C, HSPA1A/ HSPA1B, HSPA1L, LST1,LTA, LTB,LY6G5C, LY6G6C, LY6G6D, MBL2, MCCD1, MDC1, MICA, MSH5,NELFE, NFKBIL1, NRM, PBLD, PCDH15,PLAC4, PRRC2A, PSORS1C1, PTK2B, SIL1, SKIV2L, SLC44A4, STK19, TMEM173,TNF, TNFSF11, TNFSF15, TNFSF8, TXN, VARS, VARS2, VIM, ZBTB12</i>	67	Other creat.
Multiple sclerosis	1,65E-08	<i>ADRA1A, BAG6, C4A/C4B, C6orf15, CDSN, DHX16,EGR1, EHMT2, EPST11,FAS,HCG22, HLA-E, IFIT1,MCCD1, MX1, NELFE ,POLE3, POU5F1, PRRC2A, PSORS1C1, RRM2B, TCF19, TNF</i>	23	Other creat.
Acquired immunodeficiency syndrome	7,05E-08	<i>C4A/C4B, C6orf48, CLU, FAS, HCP5, HLA-C, MICB, PSORS1C1, TNF</i>	9	Other creat.
Malignant solid tumor	1.31E-07	*	372	NCNA
	1.85E-06		423	Composing
	1.94E-05		432	Arranging
	1.54E-05		427	Other creat.
Cleavage of collagen fiber	1.02E-06	<i>MMP1, MMP13, MMP3, MMP8</i>	4	NCNA
Cancer	1.52E-06	*	436	Composing
	2.77E-06		378	NCNA
	4.81E-06		448	Arranging
	9,30E-05			Other creat.
X linked autism	2.09E-06	<i>MECP2, NLGN3, RPL10, TMLHE</i>	4	Arranging
Abdominal adenocarcinoma	3.55E-06	*	228	NCNA
X-linked hereditary disease	4.01E-06	<i>ABCD1, ADRA1A, AVPR2, BNIP3L, CLIC2, DKC1, EMD, F8, FLNA, G6PD, GDH1, GJB1, HCF1, HDAC8, IKBK, IL2RG, L1CAM, MECP2, MED12, NAA10, OPN1LW, OPN1MW, RAB39B, TAZ, TGFB1</i>	25	Arranging

Development of multicellular organism	4.19E-06	<i>DRG1, EN2, HOXA1, HOXA10, HOXA11, HOXA4, HOXA9, LIF, MYCNOS, NEUROG3, OSM, PLXNA3, RPS4X, TP53, ZMYM3</i>	15	Arranging
Cleavage of glycoprotein	4.91E-06	<i>MMPI, MMP13, MMP3, MMP8, PCSK5</i>	5	NCNA
Cell death	8.83E-06	*	157	Other creat.
	2.39E-05		159	Arranging
	6.29E-05		134	NCNA
Apoptosis	8.90E-06	*	129	Other creat.
	9.73E-05		127	Arranging
Morphogenesis of embryonic skeleton	9.95E-06	<i>HOXA2, HOXA3, HOXA4, HOXA5, HOXA6, HOXA7, MEGF8, MYCN, WNT9A</i>	9	Arranging
Cell death of male germ cells	1.40E-05	<i>BCL6, DMRTC2, ETV5, HORMAD2, HOXA11, LIMK2, MOV10L1, OSBP2, PAFAH1B3, SHBG, TASP1, TP53, YBX2</i>	13	Arranging
Compartmentalization of B lymphocytes	1.48E-05	<i>LTA, LTB, TNF</i>	3	Other creat.
Proteolysis of Gelatin	1.49E-05	<i>MMP1, MMP10, MMP12, MMP13, MMP3, MMP7</i>	6	NCNA
Cell death of germ cells	1.53E-05	<i>AXL, BCL6, DMRTC2, ETV5, HORMAD2, HOXA11, LIF, LIMK2, MOV10L1, OSBP2, PAFAH1B3, SHBG, TASP1, TP53, YBX2</i>	15	Arranging
Quantity of rib	2.07E-05	<i>BMPER, HOXA10, HOXA11, HOXA4, HOXA5, HOXA6, HOXA9, SHH, TASP1</i>	9	Arranging
Unwinding of RNA	2.76E-05	<i>DDX21, DDX50, EIF4A1, EIF4A2</i>	4	Arranging
Patterning of rostrocaudal axis	2.81E-05	<i>HOXA1, HOXA10, HOXA11, HOXA2, HOXA3, HOXA4, HOXA5, HOXA6, HOXA7, HOXA9, SHH, WNT3A</i>	12	Arranging
Morphology of vertebrae	2.84E-05	<i>BMPER, DVL2, HOXA10, HOXA11, HOXA3, HOXA4, HOXA5, HOXA6, HOXA9, SHH, TASP1, TGFB1, WNT3A, WNT9A</i>	14	Arranging
Morphology of rhombencephalon	3.15E-05	<i>AXL, CLN3, EBF2, EN2, GABRB3, HOXA1, HOXA2, KHDRBS2, LICAM, LIF, PATZ1, PLXNB2, TP53, UBE3A, WNT3A</i>	15	Arranging
Proliferation of B lymphocytes	3.65E-05	<i>ARHGEF1, AXL, BANK1, BCL6, BLNK, CD19, CD79A, GAPT, IL21R, IL27, IL2RG, IL4R, MYCN, PATZ1, PIK3AP1, PIM3, POU2F2, SHH, ST6GAL1, TGFB1, TNFSF13, TP53, WNT3A</i>	23	Arranging
Apoptosis of male germ cells	3.67E-05	<i>BCL6, DMRTC2, ETV5, HORMAD2, HOXA11, LIMK2, OSBP2, PAFAH1B3, SHBG, TASP1, TP53, YBX2</i>	12	Arranging
Brachydactyly mental retardation syndrome	3.84E-05	<i>GPC1, GPR35, HDAC4</i>	3	NCNA
Hypertrophy of connective tissue	5.11E-05	<i>HDAC4, MC4R, MMP3, NOG, STC1, WNT5A, XBP1</i>	7	NCNA
Killing of E. coli	5.22E-05	<i>DEFA1, DEFB1, DEFB103A/DEFB103B, TICAM1, WNT5A</i>	5	NCNA
Hallucinations	5.96E-05	<i>CCKAR, HTR6, NPAS3</i>	3	Composing
Transport of histamine	5.96E-05	<i>SLC22A1, SLC22A2, SLC22A3</i>	3	Composing
Demyelination of fiber tract	6.58E-05	<i>CXCR3, GAL3ST1, TGFB1</i>	3	Arranging
Cerebral malaria	6.77E-05	<i>C4A/C4B, CFB, LTA, TNF</i>	4	Other creat.
Cell death of gonadal cells	6.92E-05	<i>AXL, BCL6, DMRTC2, ETV5, HORMAD2, HOXA11, LIF, LIMK2, MOV10L1, OSBP2, PAFAH1B3, SHBG, TASP1, TGFB1, TP53, YBX2</i>	16	Arranging
Morphology of cells	7.02E-05	*	100	Arranging
Abnormal morphology of glossopharyngeal nerve	7.93E-05	<i>HOXA1, HOXA2, HOXA3, MEGF8</i>	4	Arranging
Dysmyelination	8.21E-05	<i>ABCD1, ADIPOQ, CD19, CTC1, CXCR3, EBF2, GAL3ST1, GJB1, GJC2, IKBKG, LIF, MLC1, TGFB1</i>	13	Arranging
Adenocarcinoma	8.46E-05	*	239	NCNA
Abnormal morphology of	8.76E-05	<i>AXL, CLN3, EBF2, EN2, GABRB3, HOXA1, HOXA2, KHDRBS2, LICAM, LIF, PATZ1, TP53, UBE3A, WNT3A</i>	14	Arranging

rhombencephalon				
Necrosis	9.27E-05	*	121	Other creat.
Neutralization of Influenza A virus	9.44E-05	<i>DEFA1, DEFA5, DEFA6</i>	3	NCNA
Morphology of gonadal cells	9.54E-05	<i>AXL, CTDNEP1, DMRTC2, ETV5, FETUB, HORMAD2, HOXA10, LIMK2, LIPE, MNS1, OSBP2, PAFAH1B3, PLA2G3, SH2B1, SPEM1, TASPI, TP53, YBX2</i>	18	Arranging
Morphology of reproductive system	9.55E-05	<i>AXL, BMPR1B, BRAF, CITED1, CTDNEP1, DMRTC2, EBF2, ERF, ETV5, FETUB, G6PD, GAL3ST1, HORMAD2, HOXA10, HOXA11, IL2RG, LIF, LIMK2, LIPE, MNS1, MOV10L1, NUPR1, OSBP2, PAFAH1B3, PATZ1, PLA2G3, PLK2, SH2B1, SLC34A2, SLIT2, SPEM1, TASPI, TGFB1, TP53, YBX2</i>	35	Arranging

\* List of associated genes is not shown for functions with 100 or more genes.