Geneset	stat	num.genes	pval	p.adj	gene.vals
Skin Development (GO:0043588)	0.17289050	66	1.210e-06		DSP:51 OPN3:367 ITGB4:406 MED1:466 COL3A1:815 EVPL:861
NADH Dehydrogenase Complex Assembly (GO:	0.18430286	49		1.469e-02	TMEM126B:9 NDUFA11:112 ECSIT:136 NDUFAF1:152 NDUFB8:335 NDUFB5:419
Mitochondrial Respiratory Chain Complex	0.18430286	49	8.153e-06	1.469e-02	TMEM126B:9 NDUFA11:112 ECSIT:136 NDUFAF1:152 NDUFB8:335 NDUFB5:419
Calcium Ion Transmembrane Import Into Cy	0.13956176	79	1.829e-05	2.073e-02	TRPV6:52 CACNA1I:278 CACNA1S:283 CACNA1E:310 SLC24A4:313 CACNA1G:528
Calcium Ion Transmembrane Transport (GO:	0.14010667	78	1.917e-05	2.073e-02	TRPM5:19 TRPV6:52 SLC24A3:237 CACNA1S:283 SLC24A4:313 ATP2C2:397
Keratinocyte Differentiation (GO:0030216	0.18654782	40	4.483e-05	4.040e-02	DSP:51 OPN3:367 MED1:466 EVPL:861 KRT16:1278 TGM1:1291
Natural Killer Cell Activation Involved	0.24586600	22	6.564e-05	4.436e-02	IFNA5:414 IFNA10:985 IFNA1:1066 IFNA2:1093 FCGR3A:1832 IFNA7:2164
Regulation Of Peptidyl–Serine Phosphoryl	0.27234135	18	6.338e-05	4.436e-02	IFNA5:414 IFNA10:985 IFNA1:1066 IFNA2:1093 IFNG:1395 IFNA7:2164
Positive Regulation Of Intracellular Tra	0.18140351	39	8.901e-05	5.347e-02	SORL1:6 SPAG5:369 MBD1:686 NEDD4:1044 PRR5L:1322 KIF20B:1349
Epidermis Development (GO:0008544)	0.11824992	84	1.819e-04	7.632e-02	DSP:51 HOXB13:169 FLOT2:610 EVPL:861 LCE2B:935 MANSC4:968
Intermediate Filament Organization (GO:0	0.13516074	65	1.659e-04	7.632e-02	PKP1:12 DSP:51 KRT17:196 KRT80:222 KRT20:247 DES:402
mRNA Methylation (GO:0080009)	0.27758403	15	1.976e-04	7.632e-02	TRMT61B:632 TRMT10C:646 METTL8:787 NSUN2:1916 CMTR1:1970 TRMT61A:2821
Mitochondrial Respiratory Chain Complex	0.11951587	82	1.855e-04	7.632e-02	TMEM126B:9 NDUFA11:112 ECSIT:136 NDUFAF1:152 NDUFB8:335 NDUFB5:419
Positive Regulation Of Peptidyl-Serine P	0.26217905	17	1.824e-04	7.632e-02	IFNA5:414 IFNA10:985 IFNA2:1093 IFNG:1395 IFNA7:2164 IFNA14:2417
Epidermal Cell Differentiation (GO:00099	0.14786169	52	2.273e-04	7.951e-02	DSP:51 OPN3:367 MED1:466 EVPL:861 OVOL3:1046 KRT16:1278
Homophilic Cell Adhesion Via Plasma Memb	0.13678365	60	2.500e-04	7.951e-02	SDK1:32 TRO:59 DSG2:195 ITGB1:346 NEXN:371 L1CAM:594
Positive Regulation Of Neuron Apoptotic	-0.22115776	23	2.416e-04	7.951e-02	ITGAM:427 FBXW7:508 BAX:877 TFAP2A:1104 TNF:1615 CDK5R1:2334
Regulation Of Protein Localization To Nu	-0.14268264	51	4.263e-04	1.281e-01	PARP9:167 KAT7:263 DTX3L:369 YWHAZ:437 OTUD7B:839 TYK2:887
Internal Protein Amino Acid Acetylation	-0.35641964	8	4.811e-04	1.369e-01	KAT7:263 MAPT:350 NAA10:955 KAT5:1702 KAT2A:2218 KAT2B:2985
Calcium Ion Transport (GO:0006816)	0.09393830	110	6.753e-04	1.826e-01	TRPM5:19 TRPV6:52 SLC24A3:237 CACNA1S:283 SLC24A4:313 ATP2C2:397
Actin-Myosin Filament Sliding (GO:003327	0.30312187	10	9.027e-04	2.236e-01	MYH2:16 MYL6B:461 TNNT2:704 MYH3:783 MYH4:944 MYH8:3483
Calcium Ion Import Across Plasma Membran	0.16092912	34	1.168e-03	2.236e-01	TRPV6:52 CACNA1I:278 CACNA1S:283 CACNA1E:310 SLC24A4:313 CACNA1G:528
Gene Expression (GO:0010467)	-0.05890262	266	9.807e-04	2.236e-01	RBM39:16.5 RPS4Y2:16.5 GSPT2:98 CHTOP:128 MRPS21:169 EXOSC9:198
Lymphocyte Activation Involved In Immune	0.19584182	23	1.151e-03	2.236e-01	IFNA5:414 IFNA10:985 IFNA1:1066 IFNA2:1093 IFNA7:2164 IFNA14:2417
mRNA Modification (GO:0016556)	0.18349639	27	9.681e-04	2.236e-01	TRMT61B:632 TRMT10C:646 METTL8:787 PUS7L:1000 DKC1:1209 NSUN2:1916
Natural Killer Cell Activation (GO:00301	0.14090453	44	1.227e-03	2.236e-01	IFNA5:414 IFNA10:985 IFNA1:1066 IFNA2:1093 CD2:1483 NCR3:1616
Protein Insertion Into ER Membrane (GO:0	-0.17792637	28	1.122e-03	2.236e-01	MMGT1:252 TRAM1:1127 CCDC47:1414 RAB5IF:1611 GET3:1771 EMC4:2305.5
Regulation Of Vesicle Size (GO:0097494)	-0.24928030	14	1.241e-03	2.236e-01	BLOC1S1:1154 KXD1:1893 BORCS8:2110 BORCS7:2355 RAB5A:2966 AP2M1:3456
Semaphorin-Plexin Signaling Pathway Invo	0.27245284	12	1.083e-03	2.236e-01	PLXNC1:766 PLXNA4:842 PLXNB2:1102 PLXNB3:2191 SEMA3A:2999 PLXNA1:3281
Transcription Initiation At RNA Polymera	-0.10953006	73	1.224e-03	2.236e-01	MED20:142 TAF11L3:539 GTF2H2:722 GTF2B:735 TAF11L5:752 MED30:927
Mitotic Sister Chromatid Segregation (GO	0.08903716	109	1.339e-03	2.263e-01	DLGAP5:27 KIF18A:78 CEP192:96 KIF22:192 OFD1:224 KIF2C:231
Regulation Of Neuron Migration (GO:20012	0.17516094	28	1.339e-03	2.263e-01	KIF26A:120 TNN:456 COL3A1:815 ULK4:962 IGSF10:1014 PLXNB2:1102
ERBB Signaling Pathway (GO:0038127)	0.12674094	53	1.422e-03	2.330e-01	PTK2:705 AKT1:1040 PLCG1:1068 PLCE1:1177 EGFR:1249 PIK3C2A:1328
mRNA Processing (GO:0006397)	-0.06716858	187	1.575e-03	2.505e-01	RBM22:16.5 MTREX:135 SFPQ:265 WDR83:321 CPSF7:699 SART3:847
Positive Regulation Of Calcium Ion Trans	0.14187187	41	1.677e-03	2.591e-01	CASQ1:277 GPER1:336 CAPN3:834 CEMIP:909 P2RX7:1001 PLCG1:1068
Muscle Contraction (GO:0006936)	0.09468339	91	1.817e-03	2.730e-01	MYH2:16 MYH13:89 MYOF:243 CACNA1S:283 DES:402 MYH1:438
Establishment Or Maintenance Of Apical/B	0.18325259	24	1.889e-03	2.734e-01	PATJ:228 SCRIB:258 PARD3B:633 DLG3:1710 LLGL1:1783 OOEP:1819
Negative Regulation Of Inflammatory Resp	-0.08650726	107	2.016e-03	2.734e-01	TRIM65:45 FNDC4:133 TNFAIP6:278 APOA1:286 IL22:322 PRKCD:357
Presynaptic Active Zone Organization (GO	-0.39910733	5	1.996e-03	2.734e-01	ERC2:277 ERC1:545 CAST:2550 PCLO:2855 RAB3A:3194 NA
Regulation Of Smooth Muscle Contraction	0.19941021	20	2.022e-03	2.734e-01	CTTN:279 TACR2:615 PLCE1:1177 KCNB2:1411 SETD3:1793 CHRM2:1938

## EnrichmentHsSymbolsFile2 Top pathways by non-permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
HOUNKPE_HOUSEKEEPING_GENES	-0.06632010	1063	3.793e-13	2.463e-09	ARL8B:16.5 CAB39:16.5 CUL3:16.5 TMEM203:16.5 PDCD7:54 PSMD10:76
REACTOME_METABOLISM_OF_RNA	-0.07958533	645	6.273e-12	2.037e-08	RBM39:16.5 RBM22:16.5 SNRPC:16.5 RPS4Y2:16.5 PDCD7:54 PSMD10:76
REACTOME_KERATINIZATION	0.12923156	214	7.558e-11	1.636e-07	PKP1:12 DSP:51 KRTAP6-1:55 KRTAP10-1:163 KRTAP10-3:164 DSG2:195
REACTOME_FORMATION_OF_THE_CORNIFIED_ENVE	0.15619422	128	1.065e-09	1.730e-06	PKP1:12 DSP:51 DSG2:195 KRT17:196 KRT80:222 KRT20:247
BLALOCK_ALZHEIMERS_DISEASE_DN	-0.05241809	1180	1.672e-09	2.172e-06	CAB39:16.5 STAM:16.5 NQO1:16.5 TBPL1:16.5 MEMO1:16.5 SEC61A2:16.5
REACTOME_CELLULAR_RESPONSES_TO_STIMULI	-0.06327535	749	4.334e-09	4.020e-06	CUL3:16.5 NQO1:16.5 H2AC18:16.5 H2BC3:16.5 RPS4Y2:16.5 AKT3:34.5
DIAZ_CHRONIC_MYELOGENOUS_LEUKEMIA_UP	-0.04846457	1329	3.933e-09	4.020e-06	CAB39:16.5 STAM:16.5 RBM39:16.5 TBPL1:16.5 PSMD10:76 SAV1:103
NIKOLSKY_BREAST_CANCER_7P22_AMPLICON	0.27003870	37	1.316e-08	1.003e-05	INTS1:203 GPER1:336 ZFAND2A:357 SNX8:722 MAD1L1:854 AP5Z1:942
MARTENS_TRETINOIN_RESPONSE_DN	-0.06179440	733	1.390e-08	1.003e-05	TRIM65:45 OR7G2:55 HDGF:77 IRAK3:105 ATG4D:107 CHMP2A:119
REACTOME_INFECTIOUS_DISEASE	-0.05651316	876	1.610e-08	1.045e-05	CUL3:16.5 STAM:16.5 H2AC18:16.5 H2BC3:16.5 RPS4Y2:16.5 AKT3:34.5
REACTOME_POST_TRANSLATIONAL_PROTEIN_MODI	-0.04114297	1379	3.715e-07	2.027e-04	CUL3:16.5 STAM:16.5 H2AC18:16.5 TRAPPC2:16.5 WDR48:16.5 COPZ2:16.5
REACTOME_REGULATION_OF_EXPRESSION_OF_SLI	-0.12190487	146	3.746e-07	2.027e-04	RPS4Y2:16.5 PSMD10:76 GSPT2:98 RPLP0:232 PSMB6:282 PSMA4:492
PUJANA_BRCA1_PCC_NETWORK	-0.03907607	1514	4.714e-07	2.355e-04	RAP1B:16.5 TBPL1:16.5 SNRPC:16.5 FAM120A:41.5 MMP20:48 BRAF:67
HSIAO_HOUSEKEEPING_GENES	-0.07790012	349	5.994e-07	2.780e-04	H2AC18:16.5 PSD:64 CD164:65 STOM:176 AARS1:184 RPLP0:232
SPIELMAN_LYMPHOBLAST_EUROPEAN_VS_ASIAN_D	-0.05830100	573	1.977e-06	8.560e-04	CD164:65 YME1L1:106 CNOT2:181 ARPC5L:196 CCNC:216 STK17A:218
DANG_BOUND_BY_MYC	-0.04443417	1001	2.237e-06	9.080e-04	RBM39:16.5 H2AC18:16.5 CD164:65 PSMD10:76 HMOX1:122 CNPY2:163
BENPORATH_MYC_MAX_TARGETS	-0.05146216	727	2.511e-06	9.592e-04	RBM39:16.5 H2AC18:16.5 CD164:65 PSMD10:76 SAV1:103 MED20:142
REACTOME_UB_SPECIFIC_PROCESSING_PROTEASE	-0.09202678	214	3.566e-06	1.286e-03	H2AC18:16.5 WDR48:16.5 H2BC3:16.5 PSMD10:76 USP17L24:209 H2AC12:233
REACTOME_TRANSCRIPTIONAL_REGULATION_BY_R	-0.08916174	221	5.046e-06	1.725e-03	H2AC18:16.5 H2BC3:16.5 PSMD10:76 KMT2D:172 GPAM:178 PSMB6:282
REACTOME_PROCESSING_OF_CAPPED_INTRON_CON	-0.08315536	250	6.116e-06	1.986e-03	RBM39:16.5 RBM22:16.5 SNRPC:16.5 PDCD7:54 CHTOP:128 MTREX:135
REACTOME_METABOLISM_OF_AMINO_ACIDS_AND_D	-0.07042464	347	6.804e-06	2.008e-03	NQO1:16.5 RPS4Y2:16.5 PSMD10:76 BCKDK:91 RPLP0:232 OAZ3:235
REACTOME_SARS_COV_1_INFECTION	-0.11858563	121	6.694e-06	2.008e-03	RPS4Y2:16.5 CHMP2A:119 PARP9:167 NMI:201 ST3GAL4:289 YWHAZ:437
REACTOME_RUNX1_REGULATES_TRANSCRIPTION_O	-0.12133253	114	7.708e-06	2.176e-03	H2AC18:16.5 H2BC3:16.5 PSMD10:76 PSMB6:282 H2BC10:447 PSMA4:492
REACTOME_RRNA_PROCESSING	-0.09366094	182	1.334e-05	3.609e-03	RPS4Y2:16.5 MTREX:135 EXOSC9:198 RPLP0:232 WDR3:498 UTP14A:538
FLORIO_NEOCORTEX_BASAL_RADIAL_GLIA_DN	0.09138853	186	1.749e-05	4.542e-03	MKI67:14 DLGAP5:27 PARPBP:76 KIF18A:78 SGO2:135 KIF22:192
DAZARD_RESPONSE_TO_UV_SCC_UP	-0.12310743	100	2.115e-05	5.282e-03	CCNC:216 TACSTD2:225 KYAT3:379 YWHAZ:437 PMAIP1:548 MAP2K1:558
REACTOME_CYCLIN_A_CDK2_ASSOCIATED_EVENTS	-0.13648570	80	2.452e-05	5.897e-03	AKT3:34.5 PSMD10:76 PSMB6:282 PSMA4:492 MNAT1:728 RBL2:790
BRUINS_UVC_RESPONSE_LATE	-0.03767346	1077	3.311e-05	7.194e-03	ARL8B:16.5 CAB39:16.5 MEMO1:16.5 TRAPPC2:16.5 COPZ2:16.5 TCF19:50
REACTOME_SRP_DEPENDENT_COTRANSLATIONAL_P	-0.12561462	92	3.144e-05	7.194e-03	SEC61A2:16.5 RPS4Y2:16.5 RPLP0:232 RPL36:769 SSR3:891 RPL3L:897
REACTOME_TRANSCRIPTIONAL_REGULATION_BY_R	-0.11112684	117	3.324e-05	7.194e-03	AKT3:34.5 PSMD10:76 PSMB6:282 RUNX2:429.5 PSMA4:492 FBXW7:508
REACTOME_INFLUENZA_INFECTION	-0.10309685	132	4.354e-05	9.122e-03	RPS4Y2:16.5 RPLP0:232 KPNA4:296 SLC25A6:299 NUP35:541 SEC13:623
REACTOME_TCF_DEPENDENT_SIGNALING_IN_RESP	-0.08093347	214	4.573e-05	9.279e-03	CUL3:16.5 H2AC18:16.5 H2BC3:16.5 PSMD10:76 KMT2D:172 CSNK1G2:257
REACTOME_HIV_INFECTION	-0.08053527	215	4.788e-05	9.422e-03	PSMD10:76 CHMP2A:119 TAF15:159 PSMB6:282 SLC25A6:299 LCK:360
REACTOME_HCMV_LATE_EVENTS	-0.11552157	103	5.133e-05	9.805e-03	H2AC18:16.5 H2BC3:16.5 CHMP2A:119 H2AC12:233 H2BC10:447 NUP35:541
THUM_SYSTOLIC_HEART_FAILURE_UP	-0.05969294	389	5.497e-05	1.020e-02	ARL8B:16.5 CP:102 IRF8:109 ZNF672:200 KPNA4:296 FAS:361
REACTOME_SPERM_MOTILITY_AND_TAXES	0.38418630	9	6.567e-05	1.057e-02	CATSPER2:99 CATSPERB:536 CATSPER3:587 CATSPERD:760 CATSPERG:1188 HVCN1:210
REACTOME_SIGNALING_BY_ROBO_RECEPTORS	-0.08404975	188	7.161e-05	1.057e-02	RPS4Y2:16.5 PSMD10:76 GSPT2:98 RPLP0:232 PSMB6:282 PSMA4:492
REACTOME_OTHER_SEMAPHORIN_INTERACTIONS	0.27149927	18	6.661e-05	1.057e-02	SEMA7A:97 ITGB1:346 PLXNC1:766 PLXNA4:842 PTPRC:1714 SEMA4D:2068

## DisGeNET Top pathways by non-permulation

H2AC18:16.5 H2BC3:16.5 GPAM:178 STAG2:347 H2BC10:447 SP1:593

RPS4Y2:16.5 GSPT2:98 RPLP0:232 RPL36:769 RPL3L:897 RPS3:948.5

REACTOME\_ESTROGEN\_DEPENDENT\_GENE\_EXPRESS -0.10093025 131 6.696e-05 1.057e-02

Geneset	stat	num.genes	pval	p.adj	gene.vals
Cerebral Ischemia	-0.07316557	240	9.904e-05	4.867e-01	ASIC1:16.5 KCNK3:56 REN:132 KCNJ13:171 BDNF:193 CREB1:221
Esophagitis	-0.14786041	58	9.914e-05	4.867e-01	NQO1:16.5 PDLIM3:188 PLCL1:242 YWHAZ:437 IL10:581 GPHN:900.5
Bell Palsy	0.11296138	87	2.742e-04	4.921e-01	SMCHD1:293 COL6A1:294 DES:402 DNA2:425 SLC18A3:454 MGME1:485
Erythrokeratoderma	0.16605376	41	2.353e-04	4.921e-01	DSP:51 DSG2:195 KRT17:196 DES:402 SLURP1:458 GJB4:521
Leukokeratosis	0.21915410	23	2.750e-04	4.921e-01	KRT17:196 CTTN:279 CTC1:756 DKC1:1209 KRT16:1278 ACD:1302
Thyroid Agenesis	-0.20101517	27	3.007e-04	4.921e-01	FOXE1:70 THRA:346 PRKAR1A:637 TSHB:870 LHX4:1055 DUOX2:1201
Stiff-Person Syndrome	-0.27551525	14	3.581e-04	5.023e-01	GAD1:63 GP6:514.5 GPHN:900.5 NUDT10:2115 SLC6A5:2189 HLA-DQB1:2574
Anaphylaxis (non medication)	-0.16535181	38	4.218e-04	5.176e-01	CD63:306 F9:423 IL10:581 HACD1:668 STAT6:823 TM7SF2:1153
Abnormality of the skull	0.11904777	68	6.928e-04	5.233e-01	CENPJ:337 ESCO2:567 CEP152:831 CENPE:846 IFT122:880 RBBP8:1122
Facial Paresis	0.10632062	88	5.729e-04	5.233e-01	SMCHD1:293 COL6A1:294 DES:402 DNA2:425 SLC18A3:454 MGME1:485
Hyperdiploid B Acute Lymphoblastic Leuke	-0.18773742	28	5.865e-04	5.233e-01	CEBPE:154 KRAS:970 CD19:991 ASPG:1413 PIP4K2A:1568 TNF:1615
Long Qt Syndrome 2	0.29593776	11	6.773e-04	5.233e-01	GOLGA2:250 KCNH2:663 ALG10:923 KCNQ1:2351 SCN5A:3466 KCNE1:3620
Sarcoma	-0.04123981	605	5.862e-04	5.233e-01	NQO1:16.5 BRAF:67 HDGF:77 LIN28A:78 IRF8:109 MITF:111
Facial muscle weakness of muscles innerv	0.10447054	84	9.434e-04	6.617e-01	SMCHD1:293 COL6A1:294 DES:402 DNA2:425 SLC18A3:454 MGME1:485
46, XX Testicular Disorders of Sex Devel	-0.24074597	9	1.238e-02	6.715e-01	SOX10:694 FOXL2:1561 NR5A1:2066 SOX9:3151 AR:3201 RSPO1:4096
Bulbocavernosus Reflex, Decreased	0.32153499	5	1.277e-02	6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Child Development Disorders, Specific	0.15439931	23	1.038e-02	6.715e-01	CHRNA4:732 DRD2:964 KCNT1:1030 KCNQ2:1366 SLC6A8:1549 SHANK3:1682
Conjunctivitis, recurrent	-0.11712136	39	1.140e-02	6.715e-01	BLNK:551 ERCC6:765 CD19:991 TP63:1069 XPC:1332 SAT1:1846
Creatinine measurement, serum (procedure	0.09668360	63	7.993e-03	6.715e-01	WDR72:93 ALMS1:194 PTPN12:280 CACNA1S:283 CDK12:382 SPATA5L1:520
Epilepsy, Rolandic	0.13180789	30	1.248e-02	6.715e-01	KCNQ3:240 SNIP1:1205 RBFOX3:1299 KCNQ2:1366 ADAMTS4:1402 SHANK3:168
Keratoderma, Palmoplantar, Diffuse	0.23389964	12	5.024e-03	6.715e-01	SLURP1:458 RHBDF2:1174 KRT16:1278 TSC1:2208 KRT9:3404 SNAP29:3838
Mental Retardation, Psychosocial	0.06871815	130	6.901e-03	6.715e-01	SHANK2:4 NSD1:62 CALCA:109 AP4E1:230 NDST1:365 CACNA1G:528
Neoplasm Recurrence, Local	-0.12910220	36	7.366e-03	6.715e-01	LIMD2:72 YWHAZ:437 CXCL9:1338 TPD52:1509 SOX2:1797.5 DNMT3A:2024
Nephritis, Interstitial	-0.16242781	31	1.753e-03	6.715e-01	APOA1:286 BAX:877 CCL7:988 CYP2C9:990 TM7SF2:1153 CCL1:1704
ALMOPLANTAR KERATODERMA, NONEPIDERMOLYT	0.31773886	7	3.599e-03	6.715e-01	KRT80:222 RHBDF2:1174 KRT16:1278 TSC1:2208 AQP5:4073 KRT1:5086
Periodontitis, Juvenile	0.08736900	85	5.405e-03	6.715e-01	MUC2:342 NOD2:571 CAMP:920 HTRA1:995 IL12RB2:1019 AKT1:1040
PERIODONTITIS, LOCALIZED AGGRESSIVE	0.39580980	4	6.111e-03	6.715e-01	MYOM2:1389 CLU:1989 DGKA:2155 IL1B:2255 NA NA
Psychomotor retardation, mild	0.17607902	19	7.887e-03	6.715e-01	RNF168:117 CENPJ:337 CEP152:831 CENPE:846 RBBP8:1122 ATR:1306
Reflex, Abnormal	0.32153499	5	1.277e-02	6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Acoustic, Abnormal	0.32153499	5	1.277e-02	6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Anal, Absent	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Anal, Decreased	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Ankle, Abnormal	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Ankle, Decreased	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Biceps, Abnormal	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Biceps, Absent	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Biceps, Decreased	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Corneal, Absent	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Gag, Absent	0.32153499	5		6.715e-01	PMP22:1991 CHRNB2:2399 TTPA:2852 ABAT:4487 ATXN7:5003 NA
Reflex, Gag, Decreased	0.32153499	5		6.715e-01	

## MGI\_Mammalian\_Phenotype\_Level\_4 Top pathways by non-permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
MP0000467 abnormal esophagus morphology	0.13042352	42	3.538e-03	3.439e-01	IGHMBP2:322 ITGB1:346 ITGB4:406 BRCA1:669 EGFR:1249 RIPK4:1276
MP0000955 abnormal spinal cord	-0.05740173	211	4.615e-03	3.439e-01	CP:102 LRP8:164 ABCD1:237 RIMS1:245 AFG3L2:398 GRIA1:459
MP0004811 abnormal neuron physiology	-0.05741780	284	1.095e-03	3.439e-01	AKT3:34.5 GAD1:63 BRAF:67 LRP8:164 XRCC2:186 BDNF:193
MP0001502 abnormal circadian rhythm	0.11128155	49	7.225e-03	3.439e-01	PER2:3 TNC:557 PER1:578 RASD1:616 RAI1:712 MAGEL2:807
MP0000631 abnormal neuroendocrine gland	-0.08332869	92	6.043e-03	3.439e-01	HMX3:16.5 BRAF:67 THRA:346 SCG5:381 OTP:522 B4GALT1:720
MP0004142 abnormal muscle tone	-0.11952662	43	6.844e-03	3.439e-01	MOG:575 NFATC2:576 ERCC6:765 SLC7A7:819 PEX7:1483 DST:1711
MP0000749 muscle degeneration	0.14384471	36	2.884e-03	3.439e-01	PLEC:21 MYOF:243 CACNA1S:283 IGHMBP2:322 DES:402 PFKM:723
MP0002082 postnatal lethality	-0.02879610	926	5.592e-03	3.439e-01	HMX3:16.5 IRS1:43 NDRG3:59 BRAF:67 FOXE1:70 LIN28A:78
MP0010770 preweaning lethality	-0.02879610	926	5.592e-03	3.439e-01	HMX3:16.5 IRS1:43 NDRG3:59 BRAF:67 FOXE1:70 LIN28A:78
MP0002019 abnormal tumor incidence	-0.04037287	441	4.836e-03	3.439e-01	NQO1:16.5 AKT3:34.5 BRAF:67 SAV1:103 IRF8:109 SUV39H1:227
MP0002269 muscular atrophy	0.07248473	86	2.087e-02	5.525e-01	PLEC:21 CTH:242 CASQ1:277 ADARB1:290 IGHMBP2:322 ITGB1:346
MP0008961 abnormal basal metabolism	0.15294578	18	2.483e-02	5.525e-01	PER2:3 PER1:578 FTO:1418 HTR6:1647 NCOR1:2489 PTPN1:3672
MP0004036 abnormal muscle relaxation	0.10650263	40	2.009e-02	5.525e-01	CASQ1:277 ITGB1:346 DES:402 APOE:856 CLCN1:1117 NPR2:1482
MP0005381 digestive/alimentary phenotyp	0.06288275	111	2.310e-02	5.525e-01	IGHMBP2:322 MUC2:342 HSF1:560 SH3BP2:584 SLC27A4:770 TNS3:843
MP0002169 no abnormal phenotype	-0.02097726	1179	2.679e-02	5.525e-01	HMX3:16.5 AKT3:34.5 GAD1:63 BRAF:67 XRCC1:97 SNX2:99
MP0008775 abnormal heart ventricle	0.10236976	37	3.158e-02	5.525e-01	HIF3A:375 FSTL3:380 TNNT2:704 EGFR:1249 TTN:1492 SLC2A4:1493
MP0004087 abnormal muscle fiber	0.04144344	243	2.865e-02	5.525e-01	PLEC:21 XIRP2:70 CTH:242 MYOF:243 CASQ1:277 CACNA1S:283
MP0009672 abnormal birth weight	0.07494056	62	4.210e-02	5.525e-01	XIRP2:70 KIF1A:300 DHCR7:792 PLAGL1:906 LAMA1:939 AKT1:1040
MP0001286 abnormal eye development	-0.04662254	160	4.410e-02	5.525e-01	MITF:111 IKZF2:121 ABI2:246 EXT1:368 RHO:424 ZIC5:534
MP0000462 abnormal digestive system	0.11859357	30	2.484e-02	5.525e-01	HSF1:560 SLC27A4:770 TNS3:843 CLDN15:1211 APC:1871 LAMC2:2185
MP0000681 abnormal thyroid gland	-0.09694235	44	2.652e-02	5.525e-01	FOXE1:70 THRA:346 FAS:361 RUNX2:429.5 HOXA5:625 PAX9:1124
MP0000689 abnormal spleen morphology	-0.03090261	552	1.700e-02	5.525e-01	BRAF:67 CCR2:75 CP:102 IRF8:109 MITF:111 HMOX1:122
MP0004145 abnormal muscle electrophysio	0.16906051	15	2.352e-02	5.525e-01	CASQ1:277 KCNH2:663 CLCN1:1117 MUSK:1681 ENTPD1:1802 CACNG1:22
MP0005075 abnormal melanosome morpholog	0.12918043	21	4.070e-02	5.525e-01	ADAMTS20:25 KRT17:196 GPR143:361 RXRA:2231 HPS1:2309 LYST:2541
MP0003956 abnormal body size	-0.01767078	1482	4.361e-02	5.525e-01	NQO1:16.5 RAP1B:16.5 IRS1:43 BRAF:67 CCR2:75 BCKDK:91
MP0008260 abnormal autophagy	0.14839379	16	4.007e-02	5.525e-01	NOD2:571 LRRK2:824 CLCN7:1233 ATG4B:1974 PMP22:1991 PTHLH:4138
MP0002081 perinatal lethality	-0.02195086	844	4.193e-02	5.525e-01	RAP1B:16.5 GAD1:63 NPHS2:93 HMOX1:122 TRIP11:174 XRCC2:186
MP0002083 premature death	-0.02428955	821	2.605e-02	5.525e-01	STAM:16.5 IRS1:43 GAD1:63 BRAF:67 CCR2:75 BCKDK:91
MP0003252 abnormal bile duct	0.13791757	25	1.716e-02	5.525e-01	ABCB11:726 ATXN2:1094 PCTP:1244 SCP2:2223 PKHD1:2344 CASP8:295
MP0001145 abnormal male reproductive	-0.02929892	457	3.767e-02	5.525e-01	TBPL1:16.5 HMOX1:122 DMRT1:202 PICK1:239 THRA:346 SCG5:381
MP0001873 stomach inflammation	0.13042768	21	3.879e-02	5.525e-01	SH3BP2:584 IFNG:1395 IL4:2114 CTLA4:2161 TOP3B:3142 AIRE:3431
MP0000163 abnormal cartilage morphology	-0.03795365	250	4.222e-02	5.525e-01	IRS1:43 MITF:111 TRIP11:174 CLEC2D:259 COL11A2:305 THRA:346
MP0002873 normal phenotype	-0.02082545	1184	2.767e-02	5.525e-01	HMX3:16.5 AKT3:34.5 GAD1:63 BRAF:67 XRCC1:97 SNX2:99
MP0010769 abnormal survival	-0.02180342	1016		5.525e-01	HMX3:16.5 IRS1:43 NDRG3:59 BRAF:67 FOXE1:70 LIN28A:78
MP0010768 mortality/aging	-0.02111062	1039	3.356e-02	5.525e-01	HMX3:16.5 IRS1:43 NDRG3:59 BRAF:67 FOXE1:70 LIN28A:78
MP0005375 adipose tissue phenotype	-0.07624046	66	3.296e-02		BCKDK:91 TNKS2:303 PRKCI:337 SCG5:381 BGLAP:571 PRKAR1A:637
MP0000778 abnormal nervous system	-0.06142323	121		5.525e-01	AKT3:34.5 ABI2:246 EXT1:368 GAP43:382.5 EOMES:849 PURA:908
MP0002152 abnormal brain morphology	-0.02076955	968	4.195e-02	5.525e-01	HMX3:16.5 AKT3:34.5 GAD1:63 BRAF:67 BCKDK:91 SNX2:99
MP0002210 abnormal sex determination	-0.03035145	350		6.302e-01	TBPL1:16.5 HMOX1:122 DMRT1:202 SUV39H1:227 PICK1:239 THRA:346
MP0010155 abnormal intestine physiology	-0.04798876	136	5.554e-02	6.302e-01	CCR2:75 SAV1:103 CEBPE:154 IL22:322 MAP3K7:338 THRA:346

tissue\_specific Top pathways by non-permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
leg.skin	0.06510063	288	1.709e-04	9.228e-03	PKP1:12 INAVA:34 BNC1:40 DSP:51 TRIM29:56 PSAPL1:121
cerebral.cortex	0.08534221	110	2.074e-03	3.733e-02	ADGRB1:63 NCAN:82 KIF1A:300 DPYSL4:470 POU6F2:791 HECW1:1007
EBV.lymphocyte	0.03511128	753	1.477e-03	3.733e-02	SORL1:6 MKI67:14 DLGAP5:27 ZNF267:67 ATAD2:72 PARPBP:76
suprapubic.skin	0.05164577	259	4.613e-03	6.228e-02	PKP1:12 BNC1:40 PSAPL1:121 AADACL2:171 NKPD1:186 KRT17:196
eye.development	-0.07446193	69	3.289e-02	3.552e-01	MITF:111 SP1:593 RAX:709 CHRDL1:1267 FGF1:1477 FOXC1:1481
blood	-0.02828284	347	7.416e-02	6.674e-01	LIMD2:72 CEBPE:154 SLC25A37:203 C5AR2:211 S1PR4:269 TNFAIP6:278
endocervix	0.05579029	79	8.740e-02	6.742e-01	MRGPRE:2 ANGPT4:45 ELFN1:65 COL24A1:91 KIF26A:120 ZNF516:177
cerebellar.hemisphere	-0.02024614	542	1.147e-01	7.743e-01	NDRG3:59 CDH7:85 CTXN2:155 SEZ6L:177 BDNF:193 UNCX:195
adrenal.gland	-0.03586957	116	1.840e-01	7.808e-01	KCNK3:56 FNDC4:133 TMEM52:258 C4B:409 FDXR:514.5 HTR2B:621
amygdala	-0.06403002	9	5.060e-01	7.808e-01	LHFPL3:34.5 BCAN:2016 NEUROD2:4143 SLC38A8:5756 SLC17A7:6347 NEUROD6:7690
atrium.auricle	-0.03135908	95	2.926e-01	7.808e-01	SYNPO2L:44 SBK3:348 LMOD3:353 ADPRHL1:732 RPL3L:897 KLHL31:968
bladder	-0.05801266	30	2.719e-01	7.808e-01	TMC7:862 UPK1A:1455 SIGLEC15:2287 PLA2G4A:2816 AKR1B10:2837 CYP4B1:3255
breast	0.06554570	20	3.105e-01	7.808e-01	TNN:456 ANKRD30A:629 THRSP:1155 ABCB5:2321 KRT5:3324 NPY2R:3763
Brodmann.area.24	-0.06988017	33	1.652e-01	7.808e-01	GAD1:63 HRK:272 CRH:1565 NCS1:2762.5 NPTXR:2932 FAM107A:3028
C1.spinal.cord	-0.02138066	194	3.081e-01	7.808e-01	PIEZO2:73 ANKS1B:95 MDGA2:230 PLCL1:242 MOG:575 HSPA2:595
caudate.nucleus	-0.07159151	28	1.902e-01	7.808e-01	ACBD7:366 SLC1A2:982 RGS14:2033 PPP1R1B:2459 SYNDIG1L:2742 SLC35D3:3046
cerebellum	-0.01138126	409	4.372e-01	7.808e-01	ASIC1:16.5 RGS11:108 BRSK2:115 PRAG1:165 UNCX:195 SEMA6C:240
ectocervix	-0.03986128	27	4.738e-01	7.808e-01	ADAMTSL5:652 PRLR:1772 ADRA2A:1956 FGF10:3141 CPXM1:5277 HOXD13:5626
esophagogastric.junction	-0.07928404	9	4.103e-01	7.808e-01	HOXA4:1306 COL4A5:3299 ADCY5:3789 GADL1:6176 CHRM3:6994 NKX6-1:9167
esophagus.mucosa	0.01287609	263	4.768e-01	7.808e-01	PKP1:12 BNC1:40 ADH7:60 LAD1:66 FAM83H:234 RNF222:345
fallopian.tube	-0.02758934	60	4.608e-01	7.808e-01	TCF23:553 HOXA5:625 RARRES1:757 CCDC71L:810 CFAP73:1257 PAGE4:1938
hippocampus.proper	-0.07244970	13	3.659e-01	7.808e-01	HRK:272 NEUROG3:4029 NEUROD2:4143 NTS:4886 FEZF2:5608 SLC17A7:6347
hypothalamus	-0.02203188	78	5.024e-01	7.808e-01	CTXN2:155 CALCR:260 OTP:522 ADCYAP1:697 CENPVL1:967 CALY:1165
liver	-0.01390833	399	3.480e-01	7.808e-01	UGT2B7:82 CP:102 FGB:285 APOA1:286 ALDH8A1:288 INSIG1:295
lung	0.02591768	161	2.594e-01	7.808e-01	SFTPB:423 SCGB3A1:618 FCN3:641 TFEC:706 BPIFA1:784 HGF:817
minor.salivary.gland	-0.03027475	91	3.198e-01	7.808e-01	AMTN:46 TMPRSS11E:168 BPIFB2:569 ENTPD3:589 OPRPN:635 SLC6A14:691
pancreas	-0.01976626	118	4.603e-01	7.808e-01	TMEM52:258 SHISAL2B:279 SLC4A4:613 SCTR:687 PDIA2:714 GPHA2:1131
sigmoid.colon	0.06062623	51	1.349e-01	7.808e-01	DES:402 HDAC4:537 TACR2:615 MRGPRD:666 NTSR1:958 TES:1317
skeletal.muscle	0.02416037	258	1.859e-01	7.808e-01	MYH2:16 PPP1R3A:46 XIRP2:70 MYBPC2:268 CASQ1:277 CACNA1S:283
spleen	-0.01127787	367	4.646e-01	7.808e-01	VPREB3:53 LIMD2:72 CCR2:75 IRAK3:105 HMOX1:122 CD180:329
stomach	0.03256159	99	2.647e-01	7.808e-01	MYH13:89 PIK3C2G:107 PSAPL1:121 EPS8L3:251 CAPN8:316 TMED6:416
thyroid	0.01498167	176	4.959e-01	7.808e-01	WDR72:93 PLA2R1:227 GOLGA8N:291 ZNF486:341 TLE6:344 HIRA:435
ansformed.skin.fibroblast	-0.01541238	276	3.833e-01	7.808e-01	NQO1:16.5 IRS1:43 PCDHGA10:79 RCN3:100 RUNX2:429.5 AJUBA:444
uterus	-0.04110669	63	2.603e-01	7.808e-01	LINGO2:302 LIPI:385 WT1:433 TCF23:553 GATA2:1084 SHISAL1:1820

PKP1:12 BNC1:40 RNF222:345 SLURP1:458 LCE3D:558 NOD2:571

CCDC190:104 TIMP1:3495.5 PCOLCE2:4552 SUSD5:5134 CCN3:5331 SOST:5420

CUBN:28 CALCA:109 RNF186:260 CLRN3:648 KLHDC7A:658 TM4SF5:702

RGS2:646.5 SMTN:2093 TPM1:2227 PGM5:2996 NTN1:4471 KCNMB1:5729

SYNPO2L:44 LMOD3:353 HSPB7:693 ADPRHL1:732 RPL3L:897 HJV:1041

38 5.304e-01 7.957e-01 BNC1:40 AADACL4:713 ITLN1:785 THRSP:1155 ADAMTS4:1402 FAM89A:1680

0.03231663

greater.omentum

esophagus.muscularis.mucosa -0.02935970

coronary.artery cortex.kidney

left.ventricle

0.02944769

-0.04211088

0.01389819

-0.01802245

119 2.254e-01 7.808e-01

28

5.725e-01 7.984e-01

5.833e-01 7.984e-01

5.911e-01 7.984e-01 6.082e-01 7.984e-01