





COPZ2 0.2881240 57 8.849e-03 9.398e-02 2.887e-03 9.94	4.918e-01
DZIP1 0.3280493 110 2.970e-05 2.450e-03 3.186e-03 2.00	07e-01 4.918e-01
FXYD6 0.2185245 68 2.965e-02 1.860e-01 3.372e-03 1.00	00e+00 4.918e-01
FNDC11 0.3917569 98 2.582e-06 5.228e-04 3.602e-03 1.05	55e-01 4.918e-01
RAB3B 0.3379367 42 8.814e-03 9.382e-02 3.634e-03 9.94	4.918e-01
RASL11B 0.2680241 54 1.789e-02 1.400e-01 3.851e-03 1.00	00e+00 4.918e-01

p.adj

1.999e-02 1.496e-01

1.725e-02 1.375e-01

AC010255

CEP19

0.2862415

0.2620941

57

MARCHF4 0.2118844 91 1.435e-02 1.238e-01

permPValue

2.233e-03

2.257e-03

2.456e-03

2.460e-03

qValueNoperm

1.000e+00

1.000e+00

9.946e-01

1.000e+00

qValuePerm

4.918e-01

4.918e-01

4.918e-01

4.918e-01

ADARB2	-0.2328861	94	6.217e-03	7.642e-02	4.040e-03	9.385e-01	4.918e-01
CMTR1	-0.2243401	65	2.916e-02	1.838e-01	4.175e-03	1.000e+00	4.918e-01
APCDD1	-0.2857765	91	9.589e-04	2.473e-02	4.234e-03	6.377e-01	4.918e-01
SPIRE1	-0.1793261	101	2.885e-02	1.828e-01	4.335e-03	1.000e+00	4.918e-01
IQGAP2	-0.1877200	123	1.143e-02	1.089e-01	4.458e-03	1.000e+00	4.918e-01
PRODH	-0.2070884	110	8.388e-03	9.150e-02	4.640e-03	9.946e-01	4.918e-01
SLC26A3	-0.2552085	107	1.359e-03	3.050e-02	4.656e-03	6.825e-01	4.918e-01
COMT	-0.2792737	95	9.707e-04	2.495e-02	4.764e-03	6.397e-01	4.918e-01

SLC14A2 -0.3020572 118 6.757e-05 4.309e-03

SAV1 -0.4623103 42 3.395e-04 1.278e-02 3.692e-03

permPValue qValueNoperm

2.748e-01

4.731e-01

4.918e-01

4.918e-01

3.069e-03

Geneset	stat	num.genes	pval	p.adj	gene.vals
Negative Regulation Of Leukocyte Prolife	-0.4471042	4	1.955e-03	2.522e-01	TNFAIP3:492 IL33:554 ENPP3:986 LYN:1062 NA NA
Nephron Tubule Development (GO:0072080)	0.4332989	3	9.340e-03	3.466e-01	HNF1B:415 MTSS1:1287 OXSR1:1435 NA NA NA
Cellular Response To Nitrogen Levels (GO	-0.4312031	4	2.819e-03	2.984e-01	GABARAPL1:550 BECN2:840 ATG7:952 BECN1:1697 NA NA
Cellular Response To Nitrogen Starvation	-0.4312031	4	2.819e-03	2.984e-01	GABARAPL1:550 BECN2:840 ATG7:952 BECN1:1697 NA NA
Positive Regulation Of Intrinsic Apoptot	-0.4135717	2	4.278e-02	4.893e-01	MYC:858 MSX1:1679 NA NA NA NA
Positive Regulation Of Osteoblast Prolif	-0.4095427	3	1.402e-02	3.789e-01	SOX8:277 CCNA2:1839 HPSE:1896 NA NA NA
Response To Peptidoglycan (GO:0032494)	-0.4047233	5	1.723e-03	2.506e-01	INAVA:402 IL6:430 IRAK3:721 IRF5:2460 RELA:2974 NA
Positive Regulation Of Toll-Like Recepto	0.4033954	3	1.552e-02	3.834e-01	SLC15A4:1238 PTPN22:1240 RSAD2:2062 NA NA NA
Regulation Of Toll-Like Receptor 7 Signa	0.4033954	3	1.552e-02	3.834e-01	SLC15A4:1238 PTPN22:1240 RSAD2:2062 NA NA NA
Cilium Movement Involved In Cell Motilit	0.3980911	7	2.650e-04	1.002e-01	TEKT2:365 TEKT5:791 GAS8:1163 TEKT3:1764 TEKT1:2092 TEKT4:2462
Axonemal Central Apparatus Assembly (GO:	0.3936735	5	2.299e-03	2.689e-01	SPAG17:247 SPEF1:1441 DNAJB13:1934 HYDIN:2157 RSPH9:2559 NA
Valine Metabolic Process (GO:0006573)	-0.3917083	5	2.418e-03	2.711e-01	BCAT2:101 ILVBL:561 HIBCH:2203 HIBADH:2419 BCAT1:2663 NA
Cell Junction Disassembly (GO:0150146)	-0.3865811	4	7.412e-03	3.416e-01	C1QB:98 CX3CR1:274 C1QC:489 DKK1:5797 NA NA
Cellular Response To Potassium Ion (GO:0	0.3816254	4	8.208e-03	3.416e-01	DLG4:135 SLC12A2:756 DLG2:890 CRHBP:5565 NA NA
Response To Potassium Ion (GO:0035864)	0.3816254	4	8.208e-03	3.416e-01	DLG4:135 SLC12A2:756 DLG2:890 CRHBP:5565 NA NA
Protein Retention In ER Lumen (GO:000662	0.3732334	3	2.515e-02	4.371e-01	GPAA1:64 KDELR3:927 OS9:4892 NA NA NA
Toll-Like Receptor 9 Signaling Pathway (0.3713153	5	4.034e-03	3.191e-01	IRAK4:591 PIK3AP1:749 IRAK1:2048 UNC93B1:2736 TNIP2:3935 NA
Gastrulation With Mouth Forming Second (-0.3695565	3	2.663e-02	4.408e-01	LRP5:674 UGDH:2075 MEGF8:3006 NA NA NA
Positive Regulation Of Gastrulation (GO:	0.3684619	2	7.110e-02	5.639e-01	OXSR1:1435 SCX:2683 NA NA NA NA
TRAIL-activated Apoptotic Signaling Path	-0.3657934	3	2.821e-02	4.503e-01	FADD:1220 SPI1:1479 ZDHHC3:3229 NA NA NA
Protein Modification By Small Protein Co	-0.3594737	3	3.105e-02	4.695e-01	ATG7:952 UBA6:1503 SENP6:3735 NA NA NA
Regulation Of Translation In Response To	-0.3590882	4	1.287e-02	3.681e-01	EIF4G1:563 SESN2:1036 NCK1:2637 NCK2:4026 NA NA
Regulation Of Cilium Beat Frequency (GO:	0.3519910	5	6.414e-03	3.320e-01	DNAH11:19 CFAP206:481 CFAP43:1915 CYB5D1:2519 CCDC40:6549 NA
snRNA Modification (GO:0040031)	-0.3516117	4	1.487e-02	3.834e-01	MEPCE:290 METTL16:2367 METTL4:2585 NHP2:3460 NA NA
L-phenylalanine Catabolic Process (GO:00	-0.3507499	5	6.604e-03	3.320e-01	TAT:33 HGD:198 QDPR:640 GSTZ1:2049 IL4I1:8036 NA
L-phenylalanine Metabolic Process (GO:00	-0.3507499	5	6.604e-03	3.320e-01	TAT:33 HGD:198 QDPR:640 GSTZ1:2049 IL4I1:8036 NA
Erythrose 4-Phosphate/Phosphoenolpyruvat	-0.3507499	5	6.604e-03	3.320e-01	TAT:33 HGD:198 QDPR:640 GSTZ1:2049 IL4I1:8036 NA
Negative Regulation Of Actin Filament De	0.3475394	4	1.607e-02	3.834e-01	SCIN:355 PLEKHH2:378 VIL1:3087 LIMA1:5643 NA NA
Negative Regulation Of CD8-positive, Alp	0.3473551	6	3.214e-03	3.116e-01	SOCS1:275 DAPL1:351 VSIR:1791 HFE:2339 ZBTB7B:3804 SLC4A2:5687
Regulation Of Monoatomic Anion Transport	-0.3471947	4	1.618e-02	3.834e-01	CA2:263 ATP8B1:1350 STC1:2071 PDZK1:5295 NA NA
ATP Synthesis Coupled Electron Transport	0.3460955	5	7.359e-03	3.416e-01	NDUFV1:76 NDUFB6:2653 NDUFS2:2764 NDUFA12:3041 NDUFV3:3480 N
Positive Regulation Of CD4–positive, CD2	-0.3454843	3	3.821e-02	4.809e-01	KLHL25:255 HLA-DRA:1410 IFNG:5140 NA NA NA
Negative Regulation Of Mitochondrial Fus	-0.3408040	6	3.841e-03	3.191e-01	OMA1:121 SLC18A1:272 HUWE1:288 PRKN:3226 TFRC:3836 MUL1:6284
Signal Complex Assembly (GO:0007172)	-0.3373901	5	8.982e-03	3.466e-01	PXN:835 MAPK8IP2:1323 NCK1:2637 SRC:3111 NCK2:4026 NA
Cellular Response To Histamine (GO:00714	-0.3369591	5	9.070e-03	3.466e-01	GABRB3:1344 DIAPH1:1407 GABRG2:1470 DHX8:3701 GABRB1:4049 NA
Positive Regulation Of Epithelial Cell D	-0.3366368	3	4.344e-02	4.899e-01	PROM1:269 LIF:566 PAX8:6354 NA NA NA
Positive Regulation Of Translation In Re	-0.3326295	4		4.167e-01	EIF4G1:563 IMPACT:2587 NCK1:2637 NCK2:4026 NA NA
Regulation Of Lysosomal Protein Cataboli	-0.3311640	6	4.967e-03	3.191e-01	LRP1:91 LAPTM4B:236 USP8:832 MARCHF2:2253 MGAT3:2960 LDLR:853
Mesodermal Cell Differentiation (GO:0048	0.3306751	8	1.201e-03		ITGA3:358 ITGB4:951 ITGB1:1752 HMGA2:2252 ITGB3:2909 KDM6B:3230
Alpha-Amino Acid Biosynthetic Process (G	-0.3304389	10		1.002e-01	BCAT2:101 SDS:167 SDSL:384 ILVBL:561 CPS1:909 MTHFD1:2006

EnrichmentHsSymbolsFile2 Top pathways by permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
REACTOME_ERYTHROCYTES_TAKE_UP_OXYGEN_AND	-0.4606602	6	9.310e-05	1.232e-02	SLC4A1:27 RHAG:75 CA4:132 CA2:263 CA1:1219 AQP1:1829
WP_CELLULAR_PROTEOSTASIS	0.4482417	2	2.811e-02	3.183e-01	VBP1:530 PFDN2:1028 NA NA NA NA
WP_ARACHIDONATE_EPOXYGENASE_EPOXIDE_HYDR	-0.4090380	2	4.511e-02	3.770e-01	EPHX2:1139 COX5A:1574 NA NA NA NA
BIOCARTA_TERT_PATHWAY	-0.4064353	6	5.650e-04	4.071e-02	SP1:558 MZF1:811 MYC:858 SP3:1257 WT1:1910 TP53:2991
NIKOLSKY_BREAST_CANCER_12Q24_AMPLICON	0.4059417	15	5.230e-08	2.609e-05	NOC4L:116 GALNT9:313 POLE:580 SFSWAP:595 ULK1:660 CHFR:712
REACTOME_LOSS_OF_FUNCTION_OF_SMAD2_3_IN_	-0.4020973	4	5.347e-03	1.425e-01	ZFYVE9:588 TGFB1:994 TGFBR1:1921 TGFBR2:2348 NA NA
REACTOME_SIGNALING_BY_TGF_BETA_RECEPTOR_	-0.4020973	4	5.347e-03	1.425e-01	ZFYVE9:588 TGFB1:994 TGFBR1:1921 TGFBR2:2348 NA NA
STARK_HYPPOCAMPUS_22Q11_DELETION_DN	-0.3836416	20	2.866e-09	3.098e-06	PRODH:8 COMT:10 ARVCF:32 TRMT2A:62 RTN4R:102 TANGO2:251
WP_METHIONINE_METABOLISM_LEADING_TO_SULF	-0.3668945	11	2.514e-05	4.406e-03	BHMT:153 AHCY:195 CSAD:494 CBS:621 MAT1A:666 GNMT:868
TESAR_ALK_TARGETS_EPISC_4D_UP	-0.3607690	2	7.719e-02	4.657e-01	MSX1:1679 PAX6:2478 NA NA NA NA
TESAR_ALK_AND_JAK_TARGETS_MOUSE_ES_D4_UP	-0.3607690	2	7.719e-02	4.657e-01	MSX1:1679 PAX6:2478 NA NA NA NA
MIKHAYLOVA_OXIDATIVE_STRESS_RESPONSE_VIA	-0.3600579	6	2.255e-03	8.970e-02	HSPB1:301 AKR1B1:559 PGAM1:889 CTSD:2433 CALU:3489 OAT:4869
REACTOME_ABACAVIR_TRANSMEMBRANE_TRANSPOR	-0.3582866	4	1.307e-02	2.202e-01	ABCB1:209 SLC22A1:1671 SLC22A3:1984 SLC22A2:4595 NA NA
BIOCARTA_TERC_PATHWAY	-0.3541296	4	1.416e-02	2.294e-01	SP1:558 SP3:1257 NFYC:2399 RB1:4502 NA NA
RANKIN_ANGIOGENIC_TARGETS_OF_VHL_HIF2A_D	-0.3413519	6	3.782e-03	1.179e-01	VEGFA:446 ANGPTL3:882 PLXND1:1391 CDH5:2731 EGFL7:3442 ITGAV:5318
WP_EICOSANOID_METABOLISM_VIA_CYTOCHROME_	-0.3398905	3	4.144e-02	3.647e-01	PPARA:715 EPHX2:1139 PPARG:5310 NA NA NA
MANN_RESPONSE_TO_AMIFOSTINE_DN	-0.3396223	5	8.534e-03	1.802e-01	TWF1:908 BLMH:1352 PRKACB:1473 RRM2:2752 CCNB1:5482 NA
CASTELLANO_HRAS_TARGETS_UP	-0.3379291	3	4.263e-02	3.682e-01	NNAT:1319 PRDX2:1424 GFER:4513 NA NA NA
REACTOME_SARS_COV_2_MODULATES_AUTOPHAGY	0.3370961	10	2.231e-04	2.372e-02	VPS11:237 VPS16:578 VPS33B:1304 VPS45:1538 TUFM:2250 VPS41:2824
WUNDER_INFLAMMATORY_RESPONSE_AND_CHOLEST	-0.3309688	7	2.425e-03	9.361e-02	ALB:80 ADIPOQ:770 CDKN1C:1457 CXCL9:1604 JCHAIN:2639 MPEG1:5451
REACTOME_HIGHLY_SODIUM_PERMEABLE_POSTSYN	0.3306169	7	2.451e-03	9.405e-02	CHRNE:42 CHRNA4:143 CHRND:514 CHRNB4:3111 CHRNA3:4299 CHRNG:4724
REACTOME_DOPAMINE_CLEARANCE_FROM_THE_SYN	-0.3299747	4	2.227e-02	2.877e-01	COMT:10 SLC6A3:680 LRTOMT:2240 MAOA:7220 NA NA
CHEN_HOXA5_TARGETS_6HR_DN	-0.3289429	3	4.845e-02	3.841e-01	ZBED8:1791 NPTXR:2422 FJX1:3450 NA NA NA
SASAI_TARGETS_OF_CXCR6_AND_PTCH1_DN	-0.3252589	7	2.880e-03	1.032e-01	TFPI:1300 MDK:1326 PIP5K1A:1926 OTX1:2352 APOD:2436 ADAM28:3069
WU_HBX_TARGETS_3_DN	-0.3199754	10	4.586e-04	3.765e-02	IL6:430 TGFB1:994 GLG1:1289 GSTA4:1376 GAS6:1596 MAP2K2:1606
BIOCARTA_VOBESITY_PATHWAY	-0.3127738	8	2.187e-03	8.809e-02	LPL:67 RXRA:229 ADIPOQ:770 TNF:1662 RETN:1845 HSD11B1:3473
REACTOME_CIPROFLOXACIN_ADME	-0.3123888	4	3.047e-02	3.219e-01	ALB:80 SLCO1A2:681 SLC22A1:1671 SLC22A8:8754 NA NA
REACTOME_RUNX3_REGULATES_BCL2L11_BIM_TRA	-0.3116319	3	6.155e-02	4.237e-01	BCL2L11:585 FOXO3:3625 RUNX3:4215 NA NA NA
KUROKAWA_LIVER_CANCER_EARLY_RECURRENCE_D	-0.3027063	4	3.601e-02	3.437e-01	VPS35:1269 IGF2R:2739 LCLAT1:3269 RB1:4502 NA NA
REACTOME_LEUKOTRIENE_RECEPTORS	0.3023814	5	1.919e-02	2.688e-01	LTB4R:329 CYSLTR2:855 LTB4R2:1490 CYSLTR1:2852 GPR17:9372 NA
MIKKELSEN_PLURIPOTENT_STATE_DN	-0.3023770	6	1.031e-02	1.967e-01	CASP8:191 CCND2:1127 TGFBR2:2348 CDK6:2438 JAK2:3096 CAMK2D:8492
REACTOME_ABACAVIR_ADME	-0.3009328	7	5.827e-03	1.482e-01	ABCB1:209 ADAL:853 NT5C2:1106 SLC22A1:1671 SLC22A3:1984 SLC22A2:4595
CHOI_ATL_ACUTE_STAGE	-0.3006842	3	7.125e-02	4.532e-01	MET:668 ETV1:3177 MBD1:5074 NA NA NA
REACTOME_ACTIVATION_OF_THE_PHOTOTRANSDUC	0.2979060	5	2.105e-02	2.840e-01	PDE6A:289 CNGA1:443 SAG:1604 RHO:4270 PDE6B:8648 NA
BRUNEAU_HEART_GREAT_VESSELS_AND_VALVULOG	-0.2974618	7	6.420e-03	1.549e-01	JAG1:993 MED13L:1282 TBX1:1600 NOTCH1:1701 PTPN11:3352 NOTCH2:3556
REACTOME_RUNX1_REGULATES_ESTROGEN_RECEPT	-0.2941700	4	4.158e-02	3.647e-01	AXIN1:360 GPAM:1197 RUNX1:3974 ESR1:6753 NA NA
REACTOME_PP2A_MEDIATED_DEPHOSPHORYLATION	0.2941187	4	4.161e-02	3.647e-01	PPP2R1B:1285 PPP2R5D:2209 MLXIPL:3436 PFKFB1:5540 NA NA
WP_COVID19_THROMBOSIS_AND_ANTICOAGULATIO	-0.2939023	6	1.266e-02	2.193e-01	FGG:638 F13B:706 PLG:829 FGB:4689 F13A1:4776 F2:6811
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DisGeNET Top pathways by permulation

4.382e-02 3.724e-01

8.998e-04 5.453e-02

LSM10:1761 LSM11:3384 NCBP1:3576 SLBP:3750 NA NA

MDM2:345 LIF:566 BTG2:1699 DDB2:1792 CTSD:2433 XPC:2925

REACTOME_SLBP_DEPENDENT_PROCESSING_OF_RE -0.2910052

AMUNDSON_DNA_DAMAGE_RESPONSE_TP53

Geneset
HP_SPHEROCYTOSIS

GOCC_9PLUS0_MOTILE_CILIUM

MP0001661 extended life span

MP0005171 absent coat pigmentation

MP0002090 abnormal vision

MP0002877 abnormal melanocyte morpholog -0.1031842

0.1034317

0.1033343

-0.1030260

17

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Ochicaet .	Jiai	mann.genes	Pvai	p.aaj	gene. vais
Anemia, hereditary spherocytic hemolytic	-0.4941793	5	1.297e-04	4.956e-02	SLC4A1:27 EPB42:42 SPTB:85 ANK1:119 SPTA1:135 NA
Spherocytosis	-0.4941793	5	1.297e-04	4.956e-02	SLC4A1:27 EPB42:42 SPTB:85 ANK1:119 SPTA1:135 NA
Angiokeratoma	-0.4740601	3	4.457e-03	2.579e-01	MANBA:142 VEGFA:446 GLA:507 NA NA NA
Abnormality of cranial vault shape	-0.4169462	2	4.112e-02	4.572e-01	SPECC1L:151 PEX19:2323 NA NA NA NA
Abnormality of cranium shape	-0.4169462	2	4.112e-02	4.572e-01	SPECC1L:151 PEX19:2323 NA NA NA NA
Abnormality of head shape	-0.4169462	2	4.112e-02	4.572e-01	SPECC1L:151 PEX19:2323 NA NA NA NA
Asymmetry of head	-0.4169462	2	4.112e-02	4.572e-01	SPECC1L:151 PEX19:2323 NA NA NA NA
Malformation of cranial vault shape	-0.4169462	2	4.112e-02	4.572e-01	SPECC1L:151 PEX19:2323 NA NA NA NA
Malformation of cranium shape	-0.4169462	2	4.112e-02	4.572e-01	SPECC1L:151 PEX19:2323 NA NA NA NA
Malformation of head shape	-0.4169462	2	4.112e-02	4.572e-01	SPECC1L:151 PEX19:2323 NA NA NA NA
Aase Smith syndrome 2	-0.4099371	3	1.392e-02	3.404e-01	TSR2:1233 RPS10:1273 GATA1:1475 NA NA NA
Monilethrix	0.4092319	2	4.501e-02	4.695e-01	KRT80:954 DSG4:1826 NA NA NA NA
Maple Syrup Urine Disease, Thiamine Resp	-0.4079594	5	1.582e-03	1.782e-01	BCAT2:101 PPM1K:861 BCKDHA:1038 DBT:1399 BCKDHB:3382 NA
Classic Maple Syrup Urine Disease	-0.4079594	5	1.582e-03	1.782e-01	BCAT2:101 PPM1K:861 BCKDHA:1038 DBT:1399 BCKDHB:3382 NA
Intermediate Maple Syrup Urine Disease	-0.4079594	5	1.582e-03	1.782e-01	BCAT2:101 PPM1K:861 BCKDHA:1038 DBT:1399 BCKDHB:3382 NA
Intermittent Maple Syrup Urine Disease	-0.4079594	5	1.582e-03	1.782e-01	BCAT2:101 PPM1K:861 BCKDHA:1038 DBT:1399 BCKDHB:3382 NA
Herlitz Disease	0.4026420	5	1.820e-03	1.959e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 ITGA6:2330 NA
delta beta^0^ Thalassemia	0.3967382	2	5.197e-02	4.927e-01	PSMB6:543 DLL1:2626 NA NA NA NA
Miller Fisher Syndrome	0.3954018	3	1.769e-02	3.684e-01	PSMB6:543 SMUG1:1645 DLL1:2626 NA NA NA
Junctional split	0.3951658	6	8.020e-04	1.289e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 PLEC:2200 ITGA6:2330
Desmoplastic infantile astrocytoma	-0.3921741	6	8.787e-04	1.367e-01	HDAC6:158 DIAPH2:411 LIF:566 BRAF:2130 TP53:2991 PTPN11:3352
PAROXYSMAL EXTREME PAIN DISORDER	-0.3909838	5	2.464e-03	2.147e-01	SCN9A:735 SCN10A:766 SCN11A:1111 IDS:1200 MCF2L2:4195 NA
Progression of non-small cell lung cance	0.3900721	2	5.604e-02	4.970e-01	CCR7:1133 CD274:2251 NA NA NA NA
Sepsis of the newborn	-0.3898952	3	1.934e-02	3.809e-01	IL6:430 SERAC1:1490 ST14:2980 NA NA NA
Occipital myelomeningocele	-0.3893550	9	5.241e-05	3.022e-02	COMT:10 ARVCF:32 UFD1:383 HIRA:1025 TBX1:1600 GP1BB:2218
22q11 partial monosomy syndrome	-0.3889745	11	7.942e-06	7.786e-03	COMT:10 ARVCF:32 UFD1:383 PI4KA:789 HIRA:1025 TBX1:1600
Bacterial sepsis of newborn	-0.3852926	2	5.912e-02	5.005e-01	IL6:430 ST14:2980 NA NA NA NA
Visually threatening diabetic retinopath	-0.3811640	4	8.285e-03	3.088e-01	VEGFA:446 TNF:1662 CXCL12:2465 LTA:2524 NA NA
EPIDERMOLYSIS BULLOSA, JUNCTIONAL, LOCAL	0.3810702	5	3.167e-03	2.197e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 COL17A1:3959 NA
Adult junctional epidermolysis bullosa (0.3810702	5	3.167e-03	2.197e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 COL17A1:3959 NA
Epidermolysis Bullosa Progressiva	0.3810702	5	3.167e-03	2.197e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 COL17A1:3959 NA
JEB–I	0.3810702	5	3.167e-03	2.197e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 COL17A1:3959 NA
Palmar hyperhidrosis	0.3810702	5	3.167e-03	2.197e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 COL17A1:3959 NA
Selenium measurement	-0.3777256	4	8.886e-03	3.174e-01	BHMT:153 CBS:621 DMGDH:2503 ARSB:3977 NA NA
Plantar hyperkeratosis	0.3771878	6	1.376e-03	1.752e-01	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 PLEC:2200 COL17A1:3959
Vulvar Neoplasms	-0.3770650	3	2.370e-02	4.097e-01	CA9:1415 CASP3:1995 PTGS2:2086 NA NA NA
Exudative vitreoretinopathy	-0.3767089	5	3.531e-03	2.355e-01	ZNF408:671 LRP5:674 NDP:2526 FZD4:2574 TSPAN12:2774 NA
Chronic iridocyclitis	-0.3764464	2	6.519e-02	5.133e-01	IL6:430 RBM45:3228 NA NA NA NA
prenatal alcohol exposure	-0.3759644	4	9.209e-03	3.174e-01	VEGFA:446 IGF2:1206 HAND1:2027 NTRK2:3675 NA NA
Myelitis	0.3742033	4	9.542e-03	3.174e-01	LAMC2:427 CSF2:1157 POU2F3:1450 AQP4:4657 NA NA

1.522e-06 7.105e-04 SLC4A1:27 EPB42:42 RHAG:75 SPTB:85 ANK1:119 SPTA1:135

0.4706930 4 1.112e-03 7.732e-02 DNAH11:19 CFAP45:132 DNAH5:728 ENKUR:916 NA NA

GSEA-c5-HsSymbols Top pathways by permulation

GOMF_OXIDOREDUCTASE_ACTIVITY_ACTING_ON_P		•		1.1326-02	DNAH11.19 CFAF45.132 DNAH5.726 ENKOK.916 NA NA
	-0.4502446	2	2.742e-02	3.489e-01	CYP11A1:14 CYP2U1:1454 NA NA NA NA
GOBP_BRANCHED_CHAIN_AMINO_ACID_BIOSYNTHE	-0.4478230	5	5.242e-04	5.041e-02	BCAT2:101 SDS:167 SDSL:384 ILVBL:561 BCAT1:2663 NA
GOBP_REGULATION_OF_PROTEIN_TYROSINE_PHOS	-0.4360294	3	8.902e-03	2.205e-01	SLC39A10:471 PTPRC:946 MGAT5:1418 NA NA NA
GOBP_ESTABLISHMENT_OF_LEFT_RIGHT_ASYMMET	0.4315091	4	2.799e-03	1.237e-01	CFAP45:132 CFAP52:536 ENKUR:916 CCDC39:2623 NA NA
GOBP_L_CYSTEINE_METABOLIC_PROCESS	-0.4274072	4	3.070e-03	1.313e-01	CSAD:494 CBS:621 AGXT:1531 CDO1:1648 NA NA
GOBP_POSITIVE_REGULATION_OF_CONNECTIVE_T	0.4179378	2	4.063e-02	4.023e-01	ROCK2:1193 ROCK1:1322 NA NA NA NA
GOBP_NEURON_NEURON_SYNAPTIC_TRANSMISSION	0.4071315	4	4.800e-03	1.643e-01	DRD2:170 KIF1B:1181 TMOD2:1280 DLGAP2:3057 NA NA
GOBP_REGULATION_OF_TOLL_LIKE_RECEPTOR_7_	0.4013936	3	1.604e-02	2.782e-01	SLC15A4:1238 PTPN22:1240 RSAD2:2062 NA NA NA
GOBP_LEUKOTRIENE_CATABOLIC_PROCESS	0.3946611	2	5.321e-02	4.493e-01	DPEP2:1230 DPEP1:2003 NA NA NA NA
GOMF_C3HC4_TYPE_RING_FINGER_DOMAIN_BINDI	0.3941574	2	5.351e-02	4.500e-01	KCNH2:878 PINK1:2374 NA NA NA NA
HP_ABNORMAL_ISOHEMAGGLUTININ_LEVEL	-0.3926089	5	2.362e-03	1.135e-01	SLC35C1:350 CD19:565 ARHGEF1:857 CR2:2474 PRKCD:3712 NA
HP_MULTIPLE_GLOMERULAR_CYSTS	0.3923749	3	1.858e-02	2.981e-01	HNF1B:415 NPHP3:1504 MYOCD:3029 NA NA NA
HP_DECREASED_SERUM_COMPLEMENT_FACTOR_I	-0.3917854	4	6.650e-03	1.925e-01	C1QB:98 C1QA:335 C1QC:489 CFI:5494 NA NA
HP_ABNORMAL_PERIFOLLICULAR_MORPHOLOGY	-0.3876736	3	2.003e-02	3.045e-01	LRP1:91 HLA-DRA:1410 MBTPS2:3498 NA NA NA
GOCC_INNER_DYNEIN_ARM	0.3874496	4	7.278e-03	2.012e-01	DNAH1:1176 DNHD1:1468 DNAH2:1926 DNAH7:2355 NA NA
GOCC_PREFOLDIN_COMPLEX	0.3844835	3	2.108e-02	3.091e-01	VBP1:530 PFDN2:1028 PDRG1:3752 NA NA NA
GOCC_GPI_ANCHOR_TRANSAMIDASE_COMPLEX	0.3817825	4	8.179e-03	2.113e-01	GPAA1:64 PIGK:455 PIGT:3026 PIGS:3696 NA NA
GOBP_SRP_DEPENDENT_COTRANSLATIONAL_PROTE	0.3808102	2	6.213e-02		SRPRB:1701 SRPRA:1964 NA NA NA NA
GOBP ATTACHMENT OF GPI ANCHOR TO PROTEIN	0.3807917	5	3.188e-03	1.331e-01	GPAA1:64 PIGK:455 PGAP1:1898 PIGT:3026 PIGS:3696 NA
GOBP_LEUKOTRIENE_SIGNALING_PATHWAY	0.3769341	5	3.511e-03		LTB4R:329 CYSLTR2:855 LTB4R2:1490 CYSLTR1:2852 RGS1:3903 NA
HP_FALCIFORM_RETINAL_FOLD	-0.3758978	5	3.602e-03	1.422e-01	ZNF408:671 LRP5:674 NDP:2526 FZD4:2574 TSPAN12:2774 NA
HP_HYPOPLASTIC_DERMOEPIDERMAL_HEMIDESMOS	0.3732250	7		5.456e-02	LAMC2:427 ITGB4:951 LAMA3:1863 LAMB3:1910 PLEC:2200 ITGA6:2330
GOMF_RIBOSOMAL_PROTEIN_S6_KINASE_ACTIVIT	0.3723733	4	9.897e-03		RPS6KA1:154 RPS6KA6:157 RPS6KA2:2241 RPS6KA4:5263 NA NA
HP_PERIPHERAL_RETINAL_AVASCULARIZATION	-0.3711074	6	1.643e-03	9.585e-02	ZNF408:671 LRP5:674 DLK1:2272 NDP:2526 FZD4:2574 TSPAN12:2774
GOBP_POSITIVE_REGULATION_OF_GLYCOGEN_STA	-0.3678086	5	4.394e-03		ADIPOQ:770 EPM2AIP1:1155 IGF2:1206 GSK3A:2713 PPP1R3G:3948 NA
GOBP_NEGATIVE_REGULATION_OF_PROTEIN_CATA	-0.3669162	5	4.490e-03		LAPTM4B:236 USP8:832 VPS35:1269 MGAT3:2960 ATP13A2:4568 NA
HP_FAILURE_TO_THRIVE_SECONDARY_TO_RECURR	0.3667770	6	1.862e-03	1.012e-01	CD3E:721 RAG1:781 CD3D:1763 IL7R:2213 CD247:3108 RAG2:3676
GOMF_PHOSPHATIDYLSERINE_FLIPPASE_ACTIVIT	-0.3654336	5	4.654e-03	1.636e-01	ATP11A:138 ATP11C:1005 ATP8B1:1350 ATP8A1:1558 ATP8A2:5920 NA
GOMF_MHC_CLASS_II_RECEPTOR_ACTIVITY	-0.3653929	4	1.137e-02	2.485e-01	HLA-DOB:673 HLA-DRA:1410 HLA-DOA:2845 HLA-DPA1:3070 NA NA
HP RECURRENT	-0.3641814	5	4.797e-03		ADORA2A:176 BCKDHA:1038 DBT:1399 BCKDHB:3382 PTH:4065 NA
HP_PARTIAL_ABSENCE_OF_SPECIFIC_ANTIBODY_	-0.3622382	5		1.679e-01	TNFRSF13B:258 CD19:565 CR2:2474 ICOS:2676 SASH3:4241 NA
GOMF MINUS END DIRECTED MICROTUBULE MOTO	0.3617653	16	5.451e-07	3.521e-04	DNAH11:19 DNAH3:232 DNAH10:368 DNAH9:435 DYNC2H1:487 DNAH5:728
GOCC_AXONEMAL_DYNEIN_COMPLEX	0.3615223	16			DNAI2:36 DNAH3:232 CCDC65:326 DNAH9:435 DNAH5:728 DNAH17:1027
GOBP_RNA_MEDIATED_HETEROCHROMATIN_FORMAT	-0.3613890	5		1.692e-01	SIRT6:866 FAM172A:1620 HELLS:1683 ZNFX1:2720 CENPV:3392 NA
GOBP_PARATHYROID_GLAND_DEVELOPMENT	-0.3581282	6		1.135e-01	GATA3:1405 TBX1:1600 GCM2:1880 TGFBR1:1921 HOXA3:2606 CRKL:3210
	-0.3522943	3	3.456e-02		FMR1:1021 BRAF:2130 SYT4:3421 NA NA NA
GOBP NEGATIVE REGULATION OF SYNAPTIC VES	0.0022010			5.181e-01	DHX9:2018 NRDE2:2382 NA NA NA NA
GOBP_NEGATIVE_REGULATION_OF_SYNAPTIC_VES GOBP_POSITIVE_REGULATION_OF_RNA_EXPORT_F	-0.3517053	2			

MGI_Mammalian_Phenotype_Level_4 Top pathways by permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
MP0006054 spinal hemorrhage	-0.2188257	8	3.219e-02	3.058e-01	SPHK1:725 TFPI:1300 C1GALT1:1852 FLI1:2179 PSEN1:3166 FBLN1:4638
MP0003121 genomic imprinting	-0.2068265	20	1.387e-03	6.818e-02	AXIN1:360 SGCE:1001 ARID4A:1205 CDKN1C:1457 HELLS:1683 NDN:1893
MP0003122 maternal imprinting	-0.1997596	11	2.189e-02	2.692e-01	AXIN1:360 SGCE:1001 ARID4A:1205 IGF2:1206 NDN:1893 DLK1:2272
MP0003724 increased susceptibility to	-0.1667610	15	2.550e-02	2.692e-01	PECAM1:312 IFNGR1:341 FPR2:610 SOD3:2172 SOCS3:2847 TNFSF10:3272
MP0005646 abnormal pituitary gland	0.1657007	18	1.507e-02	2.170e-01	GNRH1:106 DRD2:170 RASGRF1:370 ARNT2:413 NF1:1248 PROP1:1599
MP0003303 peritoneal inflammation	-0.1607388	10	7.860e-02	4.560e-01	PECAM1:312 LYN:1062 PTGS2:2086 SOCS3:2847 ST6GAL1:3151 IL10:4251
MP0010386 abnormal urinary bladder	0.1592785	14	3.927e-02	3.406e-01	CHRM3:1023 FBN1:2053 PSAP:2058 CHRNB4:3111 UPK2:3431 KCNMA1:4062
MP0002254 reproductive system inflammat	-0.1580762	5	2.211e-01	7.048e-01	MFGE8:1508 RELB:2427 EPHA1:4254 PGR:4342 AIRE:12758 NA
MP0005275 abnormal skin tensile	0.1559055	17	2.623e-02	2.692e-01	OGN:224 LAMC2:427 COL5A2:1543 DSE:1639 DSG4:1826 FBN1:2053
MP0004043 abnormal pH regulation	-0.1486003	14	5.446e-02	4.116e-01	RHCG:202 SLC12A1:221 SLC26A7:1687 WNK4:1842 SLC4A10:2102 SLC4A7:2631
MP0005174 abnormal tail pigmentation	0.1482395	14	5.505e-02	4.116e-01	MYO5A:725 CBL:999 MC1R:2073 DOCK7:3084 BLOC1S3:3615 KIT:4099
MP0008438 abnormal cutaneous collagen	0.1429851	15	5.546e-02	4.116e-01	OGN:224 PLOD1:610 COL5A2:1543 DSE:1639 FBN1:2053 TNXB:2066
MP0003172 abnormal lysosome physiology	-0.1429487	21	2.356e-02	2.692e-01	NAGPA:12 MANBA:142 AP3B1:962 IDS:1200 HPS4:1403 CTSD:2433
MP0000751 myopathy	0.1425507	18	3.652e-02	3.336e-01	TCAP:145 DMD:228 ITGA7:555 PLEC:2200 COL6A1:2205 LDB3:3414
MP0006292 abnormal olfactory placode	-0.1328421	10	1.461e-01	5.640e-01	HHEX:599 HESX1:2469 PAX6:2478 POU2F1:3963 DKK1:5797 GBX2:6535
MP0008260 abnormal autophagy	-0.1311174	15	7.904e-02	4.560e-01	ATG4C:874 ATG7:952 PMP22:1059 BNIP3L:1187 BECN1:1697 ATG9A:3124
MP0005645 abnormal hypothalamus physiol	0.1309957	14	9.001e-02	4.706e-01	NOS1:1025 NF1:1248 UCP2:1627 AVP:1941 PROKR2:2996 FGF21:4787
MP0003123 paternal imprinting	-0.1309116	7	2.306e-01	7.087e-01	AXIN1:360 CDKN1C:1457 HELLS:1683 GRB10:2987 UBE3A:3320 GPC3:13904
MP0003300 gastrointestinal ulcer	-0.1295960	14	9.350e-02	4.706e-01	TGFB1:994 VDR:1467 PKHD1:2431 CYLD:3313 IL2:3749 IL10:4251
MP0010234 abnormal vibrissa follicle	0.1268649	4	3.797e-01	8.016e-01	PKP3:866 CHUK:3642 SPINK5:6419 ST14:12007 NA NA
MP0004134 abnormal chest morphology	-0.1263750	22	4.050e-02	3.406e-01	PTCH1:718 HOXB4:973 NKX3-2:1164 PDGFA:1165 SLC35D1:1358 UNCX:1439
MP0003646 muscle fatigue	0.1263482	9	1.896e-01	6.433e-01	NOS1:1025 MB:2632 SLC2A4:2829 MTOR:3196 SOD2:3555 PPARGC1A:5051
MP0001529 abnormal vocalization	0.1248371	18	6.707e-02	4.301e-01	E2F4:92 DRD2:170 FOXP2:743 CHRM2:1927 TOR1A:2129 NDUFS4:2154
MP0001188 hyperpigmentation	0.1215453	7	2.657e-01	7.294e-01	DRD2:170 TERF1:1095 MC1R:2073 KIT:4099 CDKN2A:9791 TERF2IP:10244
MP0003787 abnormal imprinting	-0.1205357	8	2.380e-01	7.087e-01	ARID4A:1205 CDKN1C:1457 ATRX:2634 ARID4B:4400 RB1:4502 SMCHD1:5014
MP0003890 abnormal embryonic-extraembry	-0.1170296	16	1.055e-01	4.891e-01	GCLC:215 HSD17B12:487 HIRA:1025 PALB2:1287 FOXH1:1619 FOXA2:2417
MP0003656 abnormal erythrocyte physiolo	-0.1168030	21	6.430e-02	4.301e-01	SLC4A1:27 ANK1:119 PFKM:194 MAN2A1:784 FECH:934 BNIP3L:1187
MP0000750 abnormal muscle regeneration	0.1159244	31	2.584e-02	2.692e-01	MYF5:82 DMD:228 CAV2:251 MYOZ1:1279 RNF5:2232 IFRD1:3245
MP0000015 abnormal ear pigmentation	0.1150872	15	1.232e-01	5.212e-01	MYO5A:725 CBL:999 SLC24A5:1481 MC1R:2073 POLH:3201 BLOC1S3:3615
MP0001835 abnormal antigen presentation	0.1149986	48	6.027e-03	1.301e-01	TNFRSF11B:18 PSMB8:401 PDCD1LG2:758 RAG1:781 CSF2:1157 MAP3K14:1677
MP0002396 abnormal hematopoietic system	-0.1107924	19	9.501e-02	4.706e-01	CASP8:191 MAN2A1:784 MYC:858 ARHGAP1:1413 CDK6:2438 SNAI2:2571
MP0005084 abnormal gallbladder morpholo	0.1102309	14	1.537e-01	5.748e-01	VTI1B:196 HNF1B:415 HLX:513 SOX17:1299 KLB:1515 NPC1L1:2855
MP0004270 analgesia	0.1087706	17	1.210e-01	5.192e-01	MUSK:810 DLG2:890 KIF1B:1181 PLCB3:1902 NRG1:2188 RYR1:2891
MP0003806 abnormal nucleotide metabolis	0.1080727	9	2.619e-01	7.289e-01	TPMT:2493 TALDO1:2842 VPS54:2843 UPP1:3651 NQO1:6671 OTC:7365
MP0000383 abnormal hair follicle	0.1053727	21	9.511e-02	4.706e-01	FGFR2:39 PKP3:866 TRPV3:1211 KSR1:1234 KRT14:1318 DICER1:1759
MP0001879 abnormal lymphatic vessel	-0.1038005	27	6.244e-02	4.301e-01	LYVE1:180 HEG1:261 VEGFA:446 PLCG2:1090 PDPN:1217 SOX18:1227

1.403e-01 5.508e-01

3.118e-01 7.484e-01

2.841e-01 7.312e-01 2.459e-02 2.692e-01 DMD:228 INHA:295 GHR:526 DSG4:1826 MTOR:3196 SHC1:3560

FBN1:2053 ATP7A:3422 EDNRB:4377 PAX3:4394 MITF:5178 RECQL4:6611 MREG:1355 RAB27A:1786 PTPN11:3352 ECE1:4428 RB1:4502 CDKN2A:5196

SLC6A6:53 ADORA2A:176 LRAT:524 TYR:626 POU4F2:843 AP3B1:962