

EnrichmentHsSymbolsFile2 Top pathways by non-permutation

| Geneset                                  | stat        | num.genes | pval      | p.adj     | gene.vals                                                      |
|------------------------------------------|-------------|-----------|-----------|-----------|----------------------------------------------------------------|
| REACTOME_KERATINIZATION                  | -0.20873189 | 202       | 1.659e-24 | 1.077e-20 | EVPL5: KRT20:47 DSP:49 KRT80:78 KRT17:80 KRT13:86              |
| REACTOME_FORMATION_OF_THE_CORNIFIED_ENVE | -0.25072635 | 126       | 2.625e-22 | 8.523e-19 | EVPL5: KRT20:47 DSP:49 KRT80:78 KRT17:80 KRT13:86              |
| DODD_NASOPHARYNGEAL_CARINOMA_DN          | -0.22031450 | 1273      | 4.423e-18 | 9.574e-15 | KIF18A:2 FGD6:14 TFAM:29 LRPPRC:41 ZNF557:45 TPX2:52           |
| FISCHER_DREAM_TARGETS                    | 0.08327049  | 920       | 1.632e-17 | 2.649e-14 | KIF18A:2 OBH1:11 TRIM59:33 CENPC:51 WDR76:54                   |
| RODRIGUES_THYROID_CARINOMA_POORLY_DIFFE  | 0.10155905  | 602       | 2.198e-17 | 2.855e-14 | KIF18A:2 ZNF644:25 TFAM:29 LRPPRC:41 WDR76:54 NIFK:58          |
| JOHNSTONE_PARVB_TARGETS_3_DN             | 0.08421993  | 793       | 9.888e-16 | 1.070e-12 | KIF18A:2 SENP7:7 OBH1:11 CEP350:20 TFAM:29 UBAP2:47            |
| REACTOME_CELL_CYCLE                      | 0.08768495  | 647       | 3.384e-14 | 3.140e-11 | DAXX:1 KIF18A:2 CENP7:8 UIMC1:10 ESCO1:32 CENPC:51             |
| ZHANG_BREAST_CANCER_PROGENITORS_UP       | 0.09460831  | 414       | 4.419e-11 | 1.888e-08 | SYDE2:3 TRIM59:33 SLF1:60 CTH:62 KIF2C:64 SMCHD1:74            |
| REACTOME_CELL_CYCLE_CHECKPOINTS          | 0.11733024  | 268       | 4.036e-11 | 3.188e-08 | KIF18A:2 CENP7:8 UIMC1:10 CENPC:51 KIF2C:64 PHF20:65           |
| WP_GPCRS_CLASS_A_RHOODPSINLIKE           | -0.11865440 | 249       | 1.197e-10 | 6.793e-08 | P2RY4:11 SSTR4:67 PTAFR:70 BDKR1:238 SSTR3:294 ACKR3:299       |
| HAMAI_APOPTOSIS_VIA_TRAIL_UP             | 0.07569126  | 624       | 1.255e-10 | 6.793e-08 | KIF18A:2 OBH1:11 CEP350:20 TASOR:23 ZNF644:25 CENPC:51         |
| SHEDDEN_LUNG_CANCER_POOR_SURVIVAL_A6     | 0.08967520  | 440       | 1.243e-10 | 6.793e-08 | KIF18A:2 LRPPRC:41 GLMN:48 TPX2:52 WDR76:54 KIF2C:64           |
| KINSEY_TARGETS_OF_EWSR1_FLII_FUSION_UP   | 0.05499208  | 1210      | 1.611e-10 | 8.046e-08 | KIF18A:2 OBH1:11 TPX2:52 WDR76:54 NIFK:58 SLF1:60              |
| MARTENS_TRETINOLIN_RESPONSE_UP           | -0.06711415 | 782       | 2.082e-10 | 9.656e-08 | PIK3R5:23 CEP170B:26 MUC2:32 RNF186:36 CARD14:38 TRIM29:53     |
| RODRIGUES_THYROID_CARINOMA_ANAPLASTIC_U  | 0.07194496  | 673       | 2.291e-10 | 9.917e-08 | KIF18A:2 ZNF644:25 ATP13A3:35 TPX2:52 NIFK:58 RESF1:76         |
| MARSON_BOUND_BY_E2F4_UNSTIMULATED        | 0.07182736  | 664       | 3.189e-10 | 1.294e-07 | UBAP2:47 TPX2:52 WDR76:54 KIF2C:64 TTK:84 KIF2P3:85            |
| REACTOME_CELL_CYCLE_MITOTIC              | 0.08034050  | 520       | 4.058e-10 | 1.550e-07 | KIF18A:2 CENP7:8 ESCO1:32 CENPC:51 TPX2:52 KIF2C:64            |
| WP_MITOCHONDRIAL_COMPLEX_I_ASSEMBLY_MODE | 0.25677589  | 47        | 1.131e-09 | 4.079e-07 | NDUFB5:6 NDUFB10:12 NDUFB8:31 TMEM126B:43 NDUF5:59 NDUFM70:101 |
| REACTOME_RESPIRATORY_ELECTRON_TRANSPORT  | 0.16613187  | 112       | 1.272e-09 | 4.284e-07 | NDUFB5:6 NDUFB10:12 NDUFB8:31 LRPPRC:41 TMEM126B:43 NDUF5:59   |
| REACTOME_RESPIRATORY_ELECTRON_TRANSPORT  | 0.18503144  | 90        | 1.319e-09 | 4.284e-07 | NDUFB5:6 NDUFB10:12 NDUFB8:31 LRPPRC:41 TMEM126B:43 NDUF5:59   |
| PUJANA_BRCA1_PCC_NETWORK                 | 0.04611602  | 1515      | 2.778e-09 | 8.590e-07 | DAXX:1 TFAM:29 ATP13A3:35 GLMN:48 CENPC:51 TPX2:52             |
| WONG_MITOCHONDRIA_GENE_MODULE            | 0.11585824  | 214       | 6.580e-09 | 1.942e-06 | NDUFB5:6 NDUFB10:12 ZNF644:25 NDUFB8:31 TPX2:52                |
| GRAESSMANN_APOPTOSIS_BY_DOXORUBICIN_DN   | 0.04251398  | 1698      | 8.005e-09 | 2.260e-06 | MGA1:7 CRLF2:11 ZNF644:25 PLAZ2:144 PHF20:65 PHF3:72           |
| KEGG_OXIDATIVE_PHOSPHORYLATION           | 0.15484821  | 114       | 1.139e-08 | 3.083e-06 | NDUFB5:6 NDUFB10:12 NDUFB8:31 NDUF5:59 NDUFM11:108 ATP5PB:155  |
| REACTOME_MITOTIC_PROMETAPHASE            | 0.11708675  | 195       | 1.778e-08 | 4.620e-06 | KIF18A:2 CENP7:8 CENPC:51 KIF2C:64 NDC80:142 AKAP9:149         |
| WP_ELECTRON_TRANSPORT_CHAIN_OXPHOS_SYSTE | 0.17225577  | 89        | 1.966e-08 | 4.911e-06 | NDUFB5:6 NDUFB10:12 NDUFB8:31 NDUF5:59 ATP5PB:155 NDUFM70:101  |
| KIM_WT1_TARGETS_DN                       | 0.07765376  | 445       | 2.116e-08 | 4.989e-06 | KIF18A:2 FGD6:14 CEP350:20 CENPC:51 KIF2C:64 PHF3:72           |
| MOOTHA_VOXPHOS                           | 0.17473315  | 86        | 2.151e-08 | 4.989e-06 | NDUFB5:6 NDUFB10:12 NDUF5:59 ATP5PB:155 NDUFM70:101            |
| REACTOME_CELL_CYCLE_LBIogenesis          | 0.23307878  | 48        | 2.322e-08 | 5.201e-06 | NDUFB5:6 NDUFB10:12 NDUFB8:31 TMEM126B:43 NDUF5:59 NDUFM11:108 |
| REACTOME_CLASS_A_1_RHOODPSIN_LIKE_RECEPT | -0.08959244 | 324       | 3.310e-08 | 6.775e-06 | RXFPA:3 P2RY4:11 SSTR4:67 PTAFR:70 TACR2:148 GPR132:188        |
| REACTOME_SENSORY_PERCEPTION              | -0.06642596 | 596       | 3.361e-08 | 7.040e-06 | CALHM1:46 PCDH15:57 OTOF:159 OR10S1:127 OR10R2:225 OR5M10:260  |
| REACTOME_M_PHASE                         | 0.08024169  | 384       | 3.631e-08 | 7.688e-06 | KIF18A:2 CENP7:8 CENPC:51 KIF2C:64 UMP205:87 TPR:97            |
| SHEN_SMARCA2_TARGETS_UP                  | 0.07965747  | 409       | 4.391e-08 | 8.641e-06 | CEP350:20 TMEM126B:43 CENPC:51 PHF3:72 GPR132:188              |
| REACTOME_SIGNALING_BY_GPCR               | -0.06072576 | 682       | 7.135e-08 | 1.363e-05 | RXFPA:3 P2RY4:11 ARHGEF11:21 KIF18A:2 SSTR4:67 PTAFR:70        |
| WONG_EMBRYONIC_STEM_CELL_CORE            | 0.08774903  | 318       | 7.872e-08 | 1.461e-05 | NDUFB10:12 NDUFB8:31 NIFK:58 TTK:84 NDUFM11:108 NDC80:142      |
| WP_OXIDATIVE_PHOSPHORYLATION             | 0.21587730  | 51        | 9.681e-08 | 1.746e-05 | NDUFB5:6 NDUFB10:12 NDUFB8:31 NDUF5:59 NDUFM11:108 ATP5PB:155  |
| ROSTY_CERVICAL_CANCER_PROLIFERATION_CLUS | 0.13120776  | 138       | 1.048e-07 | 1.839e-05 | TPX2:52 KIF2C:64 TTK:84 NDC80:142 KIF4A:220 DLGAP5:274         |
| NYUTTEN_EZH2_TARGETS_DN                  | 0.05082026  | 959       | 1.152e-07 | 1.917e-05 | NDUFB10:12 ZNF644:25 TFAM:29 TPX2:52 WDR76:54 SLF1:60          |
| FEVR_CTNNB1_TARGETS_DN                   | 0.06758516  | 529       | 1.138e-07 | 1.917e-05 | KIF18A:2 TFAM:29 TTK:84 LRPA1:118 CD2AP:119 GRSF1:123          |
| REACTOME_THE_CITRIC_ACID_TCA_CYCLE_AND_R | 0.12005591  | 162       | 1.373e-07 | 2.229e-05 | NDUFB5:6 NDUFB10:12 NDUFB8:31 LRPPRC:41 TMEM126B:43 NDUF5:59   |

DisGeNET Top pathways by non-permutation

| Geneset                                   | stat         | num.genes | pval      | p.adj     | gene.vals                                                           |
|-------------------------------------------|--------------|-----------|-----------|-----------|---------------------------------------------------------------------|
| Mitochondrial Diseases                    | 0.07876704   | 363       | 2.883e-07 | 2.831e-03 | NDUFB5:6 NDUFB10:12 NDUFB8:31 LRPPRC:41 TMEM126B:43 NDUF5:59        |
| Increased CSF lactate                     | 0.16384078   | 56        | 2.249e-05 | 4.417e-02 | LRPPRC:41 TMEM126B:43 NDUFM11:108 FASTKD2:143 TIMMD01:211 NDUF5:59  |
| Left ventricular non-compaction cardiomyo | -0.22031450  | 31        | 2.191e-05 | 4.417e-02 | PLEC:9 DSP:49 MUL1:331 TTN:480 PKRIN:605 MYH7B:1066                 |
| Palmoaplanar Keratosis                    | -0.12860891  | 95        | 1.508e-05 | 4.417e-02 | PLEC:9 CARD14:38 DSP:49 KRT17:80 PKP1:120 ALOX12B:122               |
| Primary myocardiophaly                    | 0.12050223   | 110       | 1.296e-05 | 4.417e-02 | NIPBL:124 TRMT10A:194 THADA:297 MTOT1:394 TRMT10B:494               |
| Erythrokeratoderma                        | -0.18748458  | 41        | 3.290e-05 | 5.385e-02 | CARD14:38 DSP:49 KRT17:80 ALOX12B:122 FAM83G:285 RHBDP2:302         |
| Increased serum lactate                   | 0.11595816   | 99        | 6.818e-05 | 8.369e-02 | LRPPRC:41 TMEM70:101 FASTKD2:143 TWNK:178 NDUF5:59 TMEM70:101       |
| Parakeratosis                             | -0.28835621  | 16        | 6.520e-05 | 8.369e-02 | CARD14:38 DSP:49 TNC:60 NLRP1:300 RHBDP2:302 TRPV3:1493             |
| Epidermolytic Bullosa Simplex             | -0.23341254  | 23        | 1.069e-04 | 1.166e-01 | PLEC:9 ITGB4:13 KRT80:78 KRT17:80 TGM5:249 KRT9:43                  |
| Hypotrichosis                             | -0.14484698  | 57        | 1.566e-04 | 1.398e-01 | PKP1:120 ALOX12B:122 TGM1:190 ABCA12:368 KRT17:502 DSG4:671         |
| Seckel syndrome                           | 0.18787626   | 34        | 1.505e-04 | 1.398e-01 | CEP152:206 DNA2:473 CENPJ:475 DNMT1:654 CENPE:781 ATR:783           |
| Pachonychia Congenita                     | -0.30902012  | 12        | 2.101e-04 | 1.719e-01 | KRT80:78 KRT17:80 KRT6B:372 KRT9:413 SLURP1:1017 KRT6A:1244         |
| Anemia, Macrocytic                        | 0.18542640   | 33        | 2.283e-04 | 1.724e-01 | SFXN4:263 TET2:348 SF3B1:702 ADH1B:776 RPL26:920 RPS26:1416         |
| Psychomotor retardation, mild             | 0.23860386   | 19        | 3.179e-04 | 1.856e-01 | RNF168:66 CEP152:206 CENPJ:475 CDC6:833 CENPE:781 ATR:783           |
| Acanthosis                                | -0.18134126  | 33        | 3.130e-04 | 1.856e-01 | CARD14:38 KRT13:86 ALOX12B:122 TGM1:190 NLRP1:300 GJB4:433          |
| Amphetamine-Related Disorders             | -0.12343500  | 70        | 3.591e-04 | 1.856e-01 | MYO5B:353 ZNF423:402 HTR6:440 SLCO6A:558 PRDM2:610 HTR1B:806        |
| Amphetamine Abuse                         | -0.12343500  | 70        | 3.591e-04 | 1.856e-01 | MYO5B:353 ZNF423:402 HTR6:440 SLCO6A:558 PRDM2:610 HTR1B:806        |
| Amphetamine Addiction                     | -0.12343500  | 70        | 3.591e-04 | 1.856e-01 | MYO5B:353 ZNF423:402 HTR6:440 SLCO6A:558 PRDM2:610 HTR1B:806        |
| Mild global developmental delay           | 0.23860386   | 19        | 3.179e-04 | 1.856e-01 | RNF168:66 CEP152:206 CENPJ:475 CDC6:833 CENPE:781 ATR:783           |
| Glycogen Storage Disease Type V           | -0.227163278 | 14        | 4.334e-04 | 2.128e-01 | SLC2A4:178 PYGM:309 ATP2A1:774 MADD:867 AMPD1:878 PYGL:944          |
| Absent earlobe                            | -0.08882769  | 12        | 5.305e-04 | 2.429e-01 | CEP152:206 CENPJ:475 CENPE:781 ATR:783 RBP8:899 PLK4:1051           |
| Cornelia De Lange Syndrome                | 0.18884307   | 28        | 5.442e-04 | 2.429e-01 | NIPBL:124 AFFA:360 CENPJ:475 ATR:783 ESCO2:1248 NAALADL2:1294       |
| Arthrogryposis                            | -0.05783313  | 297       | 6.358e-04 | 2.715e-01 | PLEC:9 ITGB4:13 OPD1:59 TSC1:79 CHRNA4:138 COL6A1:192               |
| NADH:(1) Oxidoreductase deficiency        | 0.19555160   | 25        | 7.146e-04 | 2.825e-01 | NDUFB10:12 TMEM126B:43 NDUFM11:108 TIMMD01:211 NDUF5:59 NDUFM70:101 |
| Poor school performance                   | 0.03266879   | 946       | 7.191e-04 | 2.825e-01 | NSD1:5 MAGEL2:36 CLCNKA:75 CLCNKB:90 SRCAP:93 TPR:97                |
| Coughing                                  | -0.10457802  | 86        | 8.106e-04 | 3.062e-01 | PER2:4 DSP:49 TSC1:79 RTEL1:170 TACR2:184 PRNT3:229                 |
| Hyperkeratosis, Epidermolytic             | -0.24087328  | 16        | 8.511e-04 | 3.095e-01 | DSP:49 KRT80:78 ALOX12B:122 TGM1:190 KRT9:413 GJB4:433              |
| Monilethrix                               | -0.42754797  | 5         | 9.294e-04 | 3.260e-01 | KRT80:78 DSG4:671 KRT81:921 KRT86:2405 KRT83:2689 NA                |
| Periodic syndrome                         | 0.14850474   | 5         | 1.191e-03 | 4.033e-01 | TNF:515 XBP1:1018 NLRP3:1456 MEV1:900 TNFRSF1A:2981 NA              |
| Hypertrichosis                            | 0.13335447   | 49        | 1.249e-03 | 4.088e-01 | NDUF5:59 NDUFB10:12 NDUFB8:31 NDUF5:59 NDUFM11:108                  |
| Junctional split                          | -0.37872272  | 6         | 1.315e-03 | 4.164e-01 | PLEC:9 ITGB4:13 LAMC2:745 ITGA6:1729 LAMB3:2165 LAMA3:8811.5        |
| Bulbous nasal tip                         | 0.10253796   | 80        | 1.536e-03 | 4.308e-01 | AP4E1:15 SRCAP:93 JMD1C:126 SEC24C:413 DOCK6:448 DYRK1A:565         |
| Bulbous nose                              | 0.10253796   | 80        | 1.536e-03 | 4.308e-01 | AP4E1:15 SRCAP:93 JMD1C:126 SEC24C:413 DOCK6:448 DYRK1A:565         |
| Potato nose                               | 0.10253796   | 80        | 1.536e-03 | 4.308e-01 | AP4E1:15 SRCAP:93 JMD1C:126 SEC24C:413 DOCK6:448 DYRK1A:565         |
| T-lymphocyte immunodeficiency             | -0.37444618  | 6         | 1.491e-03 | 4.308e-01 | CD3E:214 SIPA1:545 LBR:911 RAG1:1117 SFTPA1:2368 SMARCA1:8811.5     |
| Dull intelligence                         | 0.03111849   | 911       | 1.653e-03 | 4.386e-01 | NSD1:5 MAGEL2:36 CLCNKA:75 CLCNKB:90 SRCAP:93 NIPBL:124             |
| Low intelligence                          | 0.03111849   | 911       | 1.653e-03 | 4.386e-01 | NSD1:5 MAGEL2:36 CLCNKA:75 CLCNKB:90 SRCAP:93 NIPBL:124             |
| Acidosis, Lactic                          | 0.07253296   | 153       | 1.996e-03 | 5.100e-01 | TFAM:29 LRPPRC:41 TMEM126B:43 TMEM70:101 NDUFM11:108 FASTKD2:143    |
| Conduction disorder of the heart          | -0.10081556  | 78        | 2.100e-03 | 5.100e-01 | DSP:49 DSG2:364 MPO:431 FLNC:441 DES:451 TTN:480                    |
| Granulomatosis                            | -0.29561828  | 9         | 2.133e-03 | 5.100e-01 | PRNT3:229 NOD2:289 CYBB:811 NCF2:945 NCF1:1055 PTPN22:1299          |

customGeneSet Top pathways by non-permutation

| Geneset                        | stat        | num.genes | pval      | p.adj     | gene.vals                                                                     |
|--------------------------------|-------------|-----------|-----------|-----------|-------------------------------------------------------------------------------|
| HumanLocalAdaptionDietAll      | -0.06950067 | 13        | 4.367e-01 | 8.580e-01 | SLC22A5:2454 AS3MT:8811.5 GPX1:8811.5 GPX3:8811.5 CELF1:8811.5 SEPSECS:8811.5 |
| NAFLDQWAS                      | 0.01515152  | 15        | 8.580e-01 | 8.580e-01 | ADH1B:776 MTARC1:1149 INSR:1200 TOR1B:2862 TM6SF2:9694.5 APOE:9694.5          |
| expressionDirectionalSelection | 0.03316327  | 42        | 6.401e-01 | 8.580e-01 | HLA-DOA1:379 TLR10:521 SCAPER:605 UBE2U:1672 HLA-DQA2:2216 ZBTB12:2383        |
| NA                             | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.1                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.2                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.3                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.4                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.5                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.6                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.7                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.8                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.9                           | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.10                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.11                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.12                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.13                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.14                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.15                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.16                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.17                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.18                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.19                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.20                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.21                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.22                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.23                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.24                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.25                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.26                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.27                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.28                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.29                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.30                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.31                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.32                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.33                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.34                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.35                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |
| NA.36                          | NA          | NA        | NA        | NA        | NA NA NA NA NA NA                                                             |

GO\_Biological\_Process\_2023 Top pathways by non-permutation

|                                                    |             |      |           |           |                                                                      |
|----------------------------------------------------|-------------|------|-----------|-----------|----------------------------------------------------------------------|
| RNA Cation Metylation (GO:0030488)                 | 0.18435309  | 36   | 1.299e-04 | 3.697e-02 | TRMT1L:106 TRMT61B:111 TRMT10A:194 THADA:297 MTOT1:394 TRMT10B:494   |
| Monocytic Catalon Transmembrane Transport          | -0.06661224 | 277  | 1.436e-04 | 3.698e-02 | KCNK3:45 CACNA1G:96 P2RX1:734 SLC6A18:175 KCN3:181 SLC24A4:269       |
| Peptide Cross-Linking (GO:0018149)                 | -0.22441734 | 24   | 1.418e-04 | 3.698e-02 | EBV1L5 DSP:49 TGM1:190 TGM5:249 TGM2:254 COL3A1:649                  |
| Regulation of DNA Replication (GO:000627)          | 0.12776350  | 73   | 1.624e-04 | 3.817e-02 | OPL1:5 ESOC1:32 BRP3:163 STON1:452 DNA2:473 USP37:501                |
| Regulation of Heart Rate by Cardiac Cond           | -0.17263087 | 40   | 1.590e-04 | 3.817e-02 | DSP:49 CACNA1G:96 DSG2:364 KCNH2:685 CACNB2:797 KCNE5:1294           |
| Cytoplasmic Translation (GO:0002181)               | 0.12602212  | 74   | 1.802e-04 | 4.061e-02 | RPL30:617 RPL23A:722 FTSJ1:764 RPL26L1:890 RPL26:920 RPS9:1135       |
| Regulation of Transcription by RNA Polym           | 0.02635899  | 1924 | 2.013e-04 | 4.356e-02 | NSD1:5 ZNF644:25 MAGEL2:364 ZSCAN2:492 ZNF557:45 HCF2C:61            |
| Peptide Biosynthetic Process (GO:0043043)          | 0.09097471  | 137  | 2.423e-04 | 5.038e-02 | SRBD1:29 PML3:243 ZAR1:282 ZAR1L:350 RPL2M:582 SPS1:30617            |
| Chemical Synaptic Transmission (GO:00072)          | -0.06486298 | 263  | 3.073e-04 | 5.684e-02 | CHRN2B:39 GABRR3:161 RIMBP2:171 CHRNA4:183 GRIN3B:208 CORT:240       |
| Long-Chain Fatty Acid Biosynthetic Proce           | -0.216098   | 24   | 3.123e-04 | 5.684e-02 | ALOX12B:122 ACSBG1:165 GSTM4:544 ALOX5:380 GSTM2:1329 ALOX5:1436     |
| Mitochondrial RNA Metabolic Process (GO:0000000)   | 0.23270540  | 20   | 3.154e-04 | 5.684e-02 | TFAM:29 TEFM:129 FASTKD3:143 TVNKL:178 TFB2M:231 POLRM7:797          |
| Potassium Ion Transmembrane Transport (G           | -0.09051281 | 135  | 2.887e-04 | 5.684e-02 | KCNK3:45 KCN3:181 SLC24A4:269 KNG2:471 KCN2V:552 KCN82:580           |
| Adenylyte Cyclase-Modulating G Protein-C           | -0.08626808 | 158  | 3.473e-04 | 6.057e-02 | PSAPL1:2 CALCA:84 ADGRG4:141 GPR78:200 CORT:240 GRM6:263             |
| Positive Regulation of Cell Cycle Process          | 0.09645366  | 114  | 3.813e-04 | 6.443e-02 | TPR:97 NCAPD3:128 NCDC80:142 ZNF16:186 USP19:284 SPAS5:351           |
| Gene Expression (GO:0010467)                       | 0.06299172  | 266  | 4.242e-04 | 6.915e-02 | NOL9:83 TPR:97 FASTKD3:143 CASC3:180 RBM6:187 SRBD1:239              |
| Mitochondrial Translation (GO:0032543)             | 0.10323442  | 97   | 4.487e-04 | 7.136e-02 | FASTKD3:143 MRLP3:243 MRL1:370 MRLP4:582 MRP528:60 MTG1:731          |
| Positive Regulation of DNA Metabolic Pro           | 0.09934479  | 104  | 4.718e-04 | 7.289e-02 | UIMC1:10 RNF168:66 SMCHD1:474 BRP3:163 RNF8:374 STON1:452            |
| Long-Chain Fatty Acid Metabolic Process            | -0.11637930 | 75   | 4.980e-04 | 7.480e-02 | ALOX12B:122 ACSBG1:165 SLC27A2:472 BRP3:164 ALOX5:380 ELOVL2:1077    |
| tRNA Modification (GO:0006040)                     | 0.12277072  | 67   | 5.154e-04 | 7.532e-02 | TRMT10A:194 THADA:297 MTOT1:394 TYW5:593 TWD1D:1743 TSJ1:654         |
| Hexoplin Biosynthetic Process (GO:00511)           | -0.37628755 | 7    | 5.653e-04 | 7.837e-02 | ALOX12B:122 ALOX5:380 GSTM2:1329 ALOX15:1514 GSTM1:1776 ALOX15B:1836 |
| Hexoplin Metabolic Process (GO:0051121)            | -0.37628755 | 7    | 5.653e-04 | 7.837e-02 | ALOX12B:122 ALOX5:380 GSTM2:1329 ALOX15:1514 GSTM1:1776 ALOX15B:1836 |
| Mitotic Sister Chromatid Segregation (GO:009424676 | 0.09424676  | 116  | 6.130e-04 | 8.286e-02 | KIF18A:2 TPX2:52 KIF2C:64 NCAPD3:128 NCDC80:142 KIF22:1628           |

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

| Geneset                                | stat        | num.genes | pval      | p.adj     | gene.vals                                                  |
|----------------------------------------|-------------|-----------|-----------|-----------|------------------------------------------------------------|
| MP0002080 prenatal lethality           | 0.04485643  | 1335      | 7.516e-07 | 3.578e-04 | DAXX:1 NSD1:5 P2RX1:29 LTN1:50 PTPR8:56 SMCHD1:74          |
| MP0001970 abnormal pain threshold      | -0.08582857 | 152       | 3.007e-04 | 3.578e-02 | CHRN2B:39 SSTR4:67 MGRPRD:83 CALCA:84 PTPR2:110 P2RX1:734  |
| MP0002085 abnormal embryonic tissue    | 0.04545307  | 654       | 1.558e-04 | 3.578e-02 | DAXX:1 NSD1:5 TFAM:29 CHD8:81 ADAMTS:52113 ARL13B:139      |
| MP0002086 abnormal extraembryonic tiss | 0.04970310  | 486       | 2.885e-04 | 3.578e-02 | DAXX:1 NSD1:5 PTPR8:56 SMCHD1:74 NFATC3:125 SETH2:261      |
| MP0002088 abnormal embryonic growth/we | 0.05716094  | 343       | 3.826e-04 | 3.643e-02 | DAXX:1 PTPR8:56 NFATC3:125 SETH2:261 CDC25A:286 AFF4:380   |
| MP0003121 genomic imprinting           | 2.18590573  | 28        | 6.768e-04 | 6.371e-02 | MAGEL2:364 SMCHD1:74 ATRX:620 DNMT1:654 C12orf171 PEG3:856 |
| MP0003984 embryonic growth retardation | 0.05325063  | 336       | 1.049e-03 | 5.240e-02 | DAXX:1 PTPR8:56 NFATC3:125 SETH2:261 CDC25A:286 AFF4:380   |