

DisGeNET Top pathways by non-permutation

| Geneset | stat | num.genes | pval | p.adj | gene.vals |
|--|--------------|-----------|-----------|-----------|--|
| Leukopenia | 0.095622174 | 200 | 3.369e-06 | 3.305e-02 | BRCA2:10 MCPH1:16 USB1:64 TG:68 ERCC6L2:102 LYST:233 |
| hypopigmented skin patch | 0.149858976 | 64 | 3.439e-05 | 1.124e-01 | BRCA2:10 USB1:64 FAS:113 SLX4:285 BRIP1:308 TSC2:403 |
| Thrombocytopenia | 0.072465305 | 286 | 2.710e-05 | 1.124e-01 | ITGA2B:8 BRCA2:10 USB1:64 ERCC6L2:102 NAAA:103 FAS:113 |
| Pyridoxine-responsive sideroblastic anem | 0.250798831 | 21 | 6.955e-05 | 1.706e-01 | BRCA2:10 SLX4:285 BRIP1:308 FANCM:491 FANCA:568 FANCL:738 |
| Pre-Eclampsia | 0.098433430 | 129 | 1.168e-04 | 2.291e-01 | ADAMTS13:174 ENG:183 TNFRSF8:194 NOD2:241 LBR:275 CPB2:282 |
| Anemia | 0.055702188 | 397 | 1.547e-04 | 2.421e-01 | ITGA2B:8 F11:24 NPHP4:36 USB1:64 TP53BP1:67 LIPA:70 |
| Tracheoesophageal Fistula | 0.167585484 | 42 | 1.728e-04 | 2.421e-01 | BRCA2:10 CYBA:37 USB1:64 SLX4:285 BRIP1:308 CTC1:479 |
| Chromosome Breakage | 0.167629355 | 39 | 2.939e-04 | 3.129e-01 | BRCA2:10 SLX4:285 BRIP1:308 ATM:475 FANCM:491 FANCA:568 |
| Ciliopathies | 0.080473134 | 169 | 3.189e-04 | 3.129e-01 | EVC2:11 PKHD1:27 NPHP4:36 NME8:38 CCDC151:42 C2CD3:78 |
| Decreased platelet count | 0.089447282 | 138 | 2.948e-04 | 3.129e-01 | BRCA2:10 USB1:64 ERCC6L2:102 ADAMTS13:174 ABL1:189 LYST:233 |
| Pancytopenia | 0.101964114 | 100 | 4.349e-04 | 3.879e-01 | CD48:53 GLB1:79 ERCC6L2:102 WRN:120 TNFRSF8:194 SLX4:285 |
| Almond-shaped palpebral fissure | 0.203000661 | 23 | 7.536e-04 | 6.161e-01 | BRCA2:10 SLX4:285 BRIP1:308 FANCM:491 FANCA:568 FANCL:738 |
| Longevity | 0.200844228 | 23 | 8.576e-04 | 6.472e-01 | SUSD3:80 WRN:120 TCF19:138 TTC6:235 MFRP:451 ECHS1:558 |
| Autoimmune Primary Adrenal Insufficiency | 0.165917156 | 31 | 1.393e-03 | 9.047e-01 | DDC:198 CXCL10:628 CIITA:761 TPO:771 CD274:907 HLA-DOA:984 |
| Cherry red spot of the macula | 0.327180741 | 8 | 1.353e-03 | 9.047e-01 | GLB1:79 HEXB:433 ASAH1:498 GM2A:626 HEXA:1095 NEU1:1376 |
| Renal Insufficiency | 0.061082839 | 230 | 1.476e-03 | 9.047e-01 | PKHD1:27 CYBA:37 CD5L:101 RASAL1:140 ROS1:144 BBS10:168 |
| Carotid Artery Plaque | 0.190155013 | 22 | 2.023e-03 | 9.450e-01 | CX3CR1:311 TLR4:382 CCR2:520 ADIPOQ:596 F3:1353 STAT1:1696 |
| Common atrium | 0.365002385 | 6 | 1.960e-03 | 9.450e-01 | EVC2:11 MMP21:410 GDF1:460 EVC:492 ZIC3:2570 PITX2:8960.5 |
| Confusion | 0.144633167 | 39 | 1.785e-03 | 9.450e-01 | BRCA2:10 FAS:113 ADAMTS13:174 TLR4:382 IL10:553 CUBN:841 |
| Fever | 0.073241049 | 151 | 1.943e-03 | 9.450e-01 | CYBA:37 FAS:113 ADAMTS13:174 ABL1:189 LYST:233 NOD2:241 |
| Irregular hyperpigmentation | 0.150738940 | 36 | 1.757e-03 | 9.450e-01 | BRCA2:10 LBR:275 SLX4:285 BRIP1:308 TNFRSF1B:442 FANCM:491 |
| Chronic stable plaque psoriasis | 0.441096979 | 4 | 2.248e-03 | 9.586e-01 | CYP1A1:344 UGT1A1:795 EPHX1:885 CYP1B1:1799 NA NA |
| Familial Thrombotic Thrombocytopenic Pur | 0.441693164 | 4 | 2.217e-03 | 9.586e-01 | ADAMTS13:174 TFPI:760 F3:1353 THBD:1521 NA NA |
| Cystic Kidney Diseases | 0.106515425 | 68 | 2.411e-03 | 9.854e-01 | PKHD1:27 NPHP4:36 BBS10:168 TMEM231:217 PKD1:227 CEP41:325 |
| Gastrointestinal pain | 0.084210426 | 105 | 2.915e-03 | 9.875e-01 | BRCA2:10 ECE1:75 FAS:113 PMS2:350 TLR4:382 TSC2:403 |
| Hemoglobin low | 0.070789158 | 147 | 3.121e-03 | 9.875e-01 | ITGA2B:8 NPHP4:36 USB1:64 LIPA:70 ERCC6L2:102 FASTKD2:158 |
| Infection | 0.044336420 | 387 | 2.935e-03 | 9.875e-01 | IL10RA:51 TG:68 FAS:113 LAMA5:176 ENG:183 ABL1:189 |
| Molar tooth sign on MRI | 0.172722351 | 25 | 2.803e-03 | 9.875e-01 | C2CD3:78 CPLANE1:216 TMEM231:217 CEP41:325 OFD1:427 KIF7:707 |
| Renal failure in adulthood | 0.090973445 | 90 | 2.893e-03 | 9.875e-01 | PKHD1:27 BBS10:168 PKD1:227 WDR73:320 SMARCAL1:343 ABCC6:507 |
| Sexually Transmitted Diseases | 0.220794547 | 15 | 3.073e-03 | 9.875e-01 | APRT:513 CXCL10:628 IFNE:767 CD38:966 IL1B:1532 EMB:1747 |
| Stricture of artery | 0.304270038 | 8 | 2.881e-03 | 9.875e-01 | ABCC6:507 SLC2A10:622 YY1AP1:1070 ENPP1:1081 ADA2:1823 ATP7A:1959 |
| 46, XX Testicular Disorders of Sex Devel | -0.127880826 | 7 | 2.413e-01 | 9.993e-01 | NR5A1:47 SOX3:6810.5 RSPO1:6810.5 SOX9:6810.5 FOXL2:6810.5 NR0B1:6810.5 |
| 46, XY Disorders of Sex Development | 0.064807595 | 15 | 3.849e-01 | 9.993e-01 | CYP11A1:476 SRD5A2:601 MAMLD1:1763 HSD17B13:1909 HSD17B7:2257 SOX9:8960.5 |
| 46, XY female | -0.033224260 | 16 | 6.455e-01 | 9.993e-01 | SOX9:6810.5 NR0B1:6810.5 HSD17B3:6810.5 DMRT2:6810.5 DMRT1:6810.5 UBL4A:6810.5 |
| 46,XY partial gonadal dysgenesis | -0.109424209 | 10 | 2.309e-01 | 9.993e-01 | NR5A1:47 SOX9:6810.5 NR0B1:6810.5 MAP3K1:6810.5 WT1:6810.5 ZFPM2:6810.5 |
| 5,10-Methylenetetrahydrofolate reductase | -0.009222417 | 18 | 8.923e-01 | 9.993e-01 | SERPINE1:396 GPX3:6810.5 ABCB1:6810.5 XRCC3:6810.5 XRCC4:6810.5 MTHFR:6810.5 |
| Abnormalities, Drug-Induced | 0.061804452 | 4 | 6.686e-01 | 9.993e-01 | EPHX1:885 WNT11:8960.5 CAT:8960.5 CRBN:8960.5 NA NA |
| Abortion, Habitual | -0.066255962 | 4 | 6.463e-01 | 9.993e-01 | MTHFR:6810.5 F2:6810.5 F5:6810.5 VEGFA:6810.5 NA NA |
| Abortion, Tubal | 0.026780272 | 91 | 3.779e-01 | 9.993e-01 | CD69:223 HSD17B1:330 CYP1A1:344 ECM2:366 CYP24A1:580 CXCL10:628 |
| Achondrogenesis, type IB (disorder) | -0.066261229 | 5 | 6.079e-01 | 9.993e-01 | SLC26A3:6810.5 SLC26A2:6810.5 SLC26A5:6810.5 SLC26A4:6810.5 DCN:6810.5 NA |