Geneset	stat	num.genes	pval	p.adj	gene.vals
Ciliary Motility Disorders	0.20709555	55	1.109e-07	1.023e-03	DNAH11:19 DNAI2:36 CCDC65:326 DNAH9:435 DNAH5:728 SPEF2:820
DiGeorge Syndrome	-0.19898041	57	2.088e-07	1.023e-03	PRODH:8 COMT:10 ARVCF:32 ALB:80 SPECC1L:151 NDST1:327
Impaired T cell function	-0.35315601	17	4.649e-07	1.519e-03	COMT:10 ARVCF:32 TNFRSF13B:258 UFD1:383 UMPS:428 CD19:565
Posterior embryotoxon	-0.25495313	31	9.072e-07	2.223e-03	COMT:10 ARVCF:32 UFD1:383 PEX2:407 JAG1:993 HIRA:1025
Seborrheic dermatitis	-0.31300806	20	1.267e-06	2.484e-03	COMT:10 ARVCF:32 SLCO2A1:46 UFD1:383 HIRA:1025 TBX1:1600
Embryotoxon	-0.25222446	30	1.758e-06	2.873e-03	COMT:10 ARVCF:32 UFD1:383 PEX2:407 JAG1:993 HIRA:1025
Polynesian Bronchiectasis	0.23541034	34	2.054e-06	2.876e-03	DNAH11:19 DNAI2:36 CCDC65:326 DNAH5:728 STK36:928 RSPH3:1118
Tetany	-0.30203645	20	2.939e-06	3.602e-03	COMT:10 ARVCF:32 SLC12A1:221 CASR:265 UFD1:383 TRPM6:904
22q11 Deletion Syndrome	-0.31090120	18	4.979e-06	5.424e-03	PRODH:8 COMT:10 ARVCF:32 UFD1:383 CXCR4:425 ZDHHC8:434
22q11 partial monosomy syndrome	-0.38897450	11	7.942e-06	7.786e-03	COMT:10 ARVCF:32 UFD1:383 PI4KA:789 HIRA:1025 TBX1:1600
Trisomy	-0.10341521	156	8.794e-06	7.837e-03	LPL:67 ALB:80 PAX5:231 MDM2:345 IL6:430 LTBR:454
Small earlobe	-0.33504955	13	2.886e-05	2.357e-02	COMT:10 ARVCF:32 UFD1:383 KCTD1:694 HIRA:1025 TBX1:1600
Dysseborrheic dermatitis	-0.30832704	15	3.567e-05	2.498e-02	COMT:10 ARVCF:32 SLCO2A1:46 UFD1:383 HIRA:1025 TBX1:1600
Shprintzen syndrome	-0.21873424	30	3.401e-05	2.498e-02	PRODH:8 COMT:10 ARVCF:32 UFD1:383 HIRA:1025 DGCR6L:1305
Hereditary spherocytosis	-0.24775670	23	3.924e-05	2.565e-02	SLC4A1:27 EPB42:42 SPTB:85 ANK1:119 SPTA1:135 SDS:167
Occipital myelomeningocele	-0.38935500	9	5.241e-05	3.022e-02	COMT:10 ARVCF:32 UFD1:383 HIRA:1025 TBX1:1600 GP1BB:2218
Reticulocyte count (procedure)	-0.09306753	160	5.113e-05	3.022e-02	IQGAP2:7 SLC4A1:27 CD2AP:69 SPTB:85 ANK1:119 SPTA1:135
Abnormality of the tonsils	-0.34467567	11	7.557e-05	4.116e-02	COMT:10 ARVCF:32 UFD1:383 HIRA:1025 TBX1:1600 GP1BB:2218
Anemia, Sickle Cell	-0.08760053	169	8.976e-05	4.543e-02	COMT:10 ALB:80 HBE1:124 SPTA1:135 KEL:275 RHD:296
Ewings sarcoma	-0.06934227	271	9.268e-05	4.543e-02	PAK3:39 LRP1:91 CASP8:191 ABCB1:209 LOX:227 FCGRT:291
Anemia, hereditary spherocytic hemolytic	-0.49417932	5	1.297e-04	4.956e-02	SLC4A1:27 EPB42:42 SPTB:85 ANK1:119 SPTA1:135 NA
Congenital atresia of nasopharynx	0.34929518	10	1.310e-04	4.956e-02	DNAH11:19 DNAH5:728 GAS8:1163 DRC1:1213 ARMC4:1815 DNAI1:2131
Kartagener Syndrome	0.11965410	88	1.072e-04	4.956e-02	DNAH11:19 DNAI2:36 CFTR:81 DNAH3:232 CCDC65:326 DNAH9:435
Mental deficiency	-0.03846442	890	1.314e-04	4.956e-02	PRODH:8 TAT:33 PAK3:39 CLCNKB:50 CLCNKA:51 GTF2IRD1:54
Other specified congenital malformations	0.34929518	10	1.310e-04	4.956e-02	DNAH11:19 DNAH5:728 GAS8:1163 DRC1:1213 ARMC4:1815 DNAI1:2131
Spherocytosis	-0.49417932	5	1.297e-04	4.956e-02	SLC4A1:27 EPB42:42 SPTB:85 ANK1:119 SPTA1:135 NA
Poor school performance	-0.03912154	849	1.415e-04	5.126e-02	PRODH:8 TAT:33 PAK3:39 CLCNKB:50 CLCNKA:51 GTF2IRD1:54
Upward slant of palpebral fissure	-0.10595977	108	1.464e-04	5.126e-02	COMT:10 ARVCF:32 EHMT1:273 UFD1:383 PEX2:407 CDK13:417
Solid Neoplasm	-0.04382415	658	1.536e-04	5.194e-02	TAT:33 LPL:67 BUB1:76 ALB:80 PGLYRP1:83 ANK1:119
Liver neoplasms	-0.03547721	1006	1.916e-04	6.261e-02	IQGAP2:7 COMT:10 KDM4B:21 TAT:33 LPL:67 ALB:80
Hepatocarcinogenesis	-0.05426388	395	2.393e-04	7.358e-02	KDM4B:21 NUP62:63 ALB:80 CLU:114 PTGES:117 ARG1:125
Increased variability in muscle fiber di	0.21663236	24	2.402e-04	7.358e-02	TCAP:145 ITGA7:555 SELENON:829 TWNK:1469 SQSTM1:2146 COL6A1:2205
Leukemia, Myelocytic, Acute	-0.03105901	1265	2.970e-04	7.770e-02	TAT:33 PAK3:39 RHEBL1:60 BUB1:76 WDHD1:88 LRP1:91
Pain, Postoperative	-0.22973881	21	2.688e-04	7.770e-02	COMT:10 TRPV1:133 ABCB1:209 LIF:566 SCN9A:735 AP3B1:962
Abnormality of aortic arch	-0.31474135	11	3.012e-04	7.770e-02	COMT:10 ARVCF:32 UFD1:383 HIRA:1025 TBX1:1600 GP1BB:2218
Bilateral cataracts (disorder)	-0.10549628	100	2.737e-04	7.770e-02	ADAM9:47 PAX5:231 CASR:265 MGST1:333 IFNGR1:341 GSTO1:389
Breast adenocarcinoma	-0.08948959	139		7.770e-02	CASP8:191 ABCB1:209 GCLC:215 HSPB1:301 MDM2:345 VEGFA:446
Fabry Disease	-0.16580585	40	2.874e-04	7.770e-02	MANBA:142 ABCB1:209 NAGA:287 VCAM1:305 IL6:430 VEGFA:446
Leprosy, Paucibacillary	-0.29684877	12		8.859e-02	LRP1:91 SCAF1:104 CHERP:798 SLC11A1:808 C1D:1441 CYFIP1:1487

GSEA-c5-HsSymbols Top pathways by permulation

Geneset	stat	num.genes	pval	p.adj	gene.vals
GOCC_CILIUM	0.09555095	623	6.795e-16	1.078e-11	CEP19:2 TMEM216:3 DZIP1:6 DNAH11:19 TPPP2:33 DNAI2:36
GOBP_CILIUM_MOVEMENT	0.18074911	157	6.156e-15	4.884e-11	DZIP1:6 DNAH11:19 TPPP2:33 DNAI2:36 TMF1:95 ATP1A4:123
GOCC_MOTILE_CILIUM	0.15261536	209	3.324e-14	1.758e-10	DNAH11:19 TPPP2:33 DNAI2:36 SAXO1:99 ACTL7A:101 ATP1A4:123
GOCC_9PLUS2_MOTILE_CILIUM	0.18178260	140	1.235e-13	4.899e-10	DNAH11:19 TPPP2:33 DNAI2:36 SAXO1:99 ATP1A4:123 IFT172:131
GOBP_CILIUM_OR_FLAGELLUM_DEPENDENT_CELL_	0.17880709	128	3.055e-12	9.694e-09	DZIP1:6 DNAH11:19 TPPP2:33 TMF1:95 ATP1A4:123 CFAP45:132
GOBP_AXONEME_ASSEMBLY	0.22834182	74	1.152e-11	3.048e-08	DNAI2:36 LRGUK:141 SPAG17:247 CCDC65:326 TEKT2:365 CFAP206:481
GOBP_SPERM_MOTILITY	0.17530259	107	3.921e-10	8.887e-07	DZIP1:6 DNAH11:19 TPPP2:33 TMF1:95 ATP1A4:123 CFAP45:132
GOBP_MICROTUBULE_BUNDLE_FORMATION	0.17154411	102	2.241e-09	3.951e-06	TPPP2:33 DNAI2:36 AAAS:63 TRIM46:71 LRGUK:141 FES:198
GOBP_ORGANONITROGEN_COMPOUND_CATABOLIC_P	-0.05391880	1117	2.148e-09	3.951e-06	PRODH:8 COMT:10 TAT:33 GBA2:45 ADAM9:47 USP30:48
GOCC_AXONEME	0.15554971	122	3.117e-09	4.947e-06	DNAH11:19 DNAI2:36 SAXO1:99 IFT172:131 CFAP45:132 AK8:168
GOBP_CILIUM_ORGANIZATION	0.08889188	375	4.057e-09	5.852e-06	CEP19:2 TMEM216:3 DZIP1:6 DNAI2:36 E2F4:92 SAXO1:99
GOBP_MICROTUBULE_BASED_MOVEMENT	0.09229572	338	6.402e-09	8.466e-06	DZIP1:6 DNAH11:19 TPPP2:33 DNAI2:36 TRIM46:71 TMF1:95
HP_ABNORMAL_SPERM_MOTILITY	0.17828241	87	9.344e-09	1.141e-05	DZIP1:6 DNAH11:19 DNAI2:36 CFAP45:132 TTC21A:240 SPAG17:247
GOBP_LIPID_METABOLIC_PROCESS	-0.04778256	1194	4.547e-08	5.153e-05	COMT:10 CYP11A1:14 ANGPTL8:22 HADHB:24 BPNT1:26 AVPR1A:28
GOCC_DYNEIN_COMPLEX	0.25171237	38	7.981e-08	8.443e-05	DNAH11:19 DNAI2:36 DYNC2LI1:190 DNAH3:232 CCDC65:326 DNAH10:368
GOBP_MOTILE_CILIUM_ASSEMBLY	0.19877875	59	1.308e-07	1.297e-04	DZIP1:6 E2F4:92 SPAG17:247 CFAP206:481 CFAP69:669 SPEF2:820
GOBP_ALPHA_AMINO_ACID_METABOLIC_PROCESS	-0.11007972	193	1.437e-07	1.341e-04	PRODH:8 TAT:33 RIDA:65 NAGS:77 BCAT2:101 ARG1:125
GOBP_ORGANIC_ACID_CATABOLIC_PROCESS	-0.09928539	236	1.620e-07	1.428e-04	PRODH:8 HADHB:24 TAT:33 RIDA:65 NUDT7:86 BCAT2:101
GOBP_MALE_GAMETE_GENERATION	0.07071490	470	1.790e-07	1.495e-04	DZIP1:6 TPPP2:33 ZBTB16:47 RAI14:62 CFTR:81 SPPL2C:90
GOCC_SUPRAMOLECULAR_POLYMER	0.05504118	769	2.764e-07	2.193e-04	DNAH11:19 TPPP2:33 DNAI2:36 NEXN:45 SCN5A:49 TOGARAM2:73
HP_INFERTILITY	0.10683192	189	4.353e-07	3.289e-04	CEP19:2 DZIP1:6 DNAH11:19 DNAI2:36 CFTR:81 GNRH1:106
GOBP_ALPHA_AMINO_ACID_CATABOLIC_PROCESS	-0.15393324	89	5.324e-07	3.521e-04	PRODH:8 TAT:33 RIDA:65 BCAT2:101 ARG1:125 IDO2:165
GOCC_AXONEMAL_DYNEIN_COMPLEX	0.36152226	16	5.548e-07	3.521e-04	DNAI2:36 DNAH3:232 CCDC65:326 DNAH9:435 DNAH5:728 DNAH17:1027
GOCC_TRANSFERASE_COMPLEX	-0.05918915	629	5.049e-07	3.521e-04	ELOVL6:55 ATG3:59 CUL4B:96 KLHL2:97 PRKAG1:107 CDK7:118
GOMF_MINUS_END_DIRECTED_MICROTUBULE_MOTO	0.36176534	16	5.451e-07	3.521e-04	DNAH11:19 DNAH3:232 DNAH10:368 DNAH9:435 DYNC2H1:487 DNAH5:728
GOBP_NEUROPEPTIDE_SIGNALING_PATHWAY	0.14430626	99	7.198e-07	4.393e-04	SORCS1:16 PROKR1:17 RXFP3:35 GPR37:226 LTB4R:329 OPRK1:635
HP_CHRONIC_SINUSITIS	0.19820968	52	7.739e-07	4.548e-04	DNAH11:19 DNAI2:36 CFTR:81 CFAP45:132 CCDC65:326 DNAH9:435
HP_ABNORMAL_ATRIAL_ARRANGEMENT	0.22010494	42	8.069e-07	4.573e-04	DNAH11:19 DNAI2:36 CCDC65:326 DNAH9:435 DNAH5:728 SPEF2:820
GOBP_SMALL_MOLECULE_CATABOLIC_PROCESS	-0.07539102	362	9.408e-07	5.148e-04	PRODH:8 HADHB:24 BPNT1:26 TAT:33 RIDA:65 NUDT7:86
GOBP_SPERMATID_DIFFERENTIATION	0.11259537	159	1.009e-06	5.338e-04	DZIP1:6 CFTR:81 SPPL2C:90 TMF1:95 ACTL7A:101 ADAD1:105
HP_NASAL_CONGESTION	0.20190902	48	1.316e-06	6.738e-04	DNAH11:19 DNAI2:36 CCDC65:326 DNAH9:435 DNAH5:728 SPEF2:820
GOBP_AXONEMAL_DYNEIN_COMPLEX_ASSEMBLY	0.26814276	27	1.423e-06	7.058e-04	DNAI2:36 CCDC65:326 TEKT2:365 DAW1:708 DNAH5:728 DNAH17:1027
GOCC_CHROMATIN	-0.04525291	1015	1.522e-06	7.105e-04	TCF15:19 NSD1:106 WAPL:123 CUX2:150 DDX21:155 KMT5C:162
HP_SPHEROCYTOSIS	-0.49091204	8	1.522e-06	7.105e-04	SLC4A1:27 EPB42:42 RHAG:75 SPTB:85 ANK1:119 SPTA1:135
GOBP_POSITIVE_REGULATION_OF_CELL_CYCLE_P	-0.09534216	214	1.635e-06	7.412e-04	NUP62:63 CUL4B:96 BRD4:181 WIZ:282 MEPCE:290 MDM2:345
HP_ABSENT_SPERM_FLAGELLA	0.30918588	20	1.699e-06	7.489e-04	DZIP1:6 TTC21A:240 DNAH10:368 SPEF2:820 TTC29:983 DNAH17:1027
GOBP_ORGANIC_ACID_METABOLIC_PROCESS	-0.04900783	840	1.850e-06	7.730e-04	PRODH:8 HADHB:24 BPNT1:26 SLC4A1:27 AVPR1A:28 TAT:33
GOBP_MICROTUBULE_BASED_PROCESS	0.05149941	754	1.900e-06	7.730e-04	CEP19:2 DZIP1:6 PHLDB1:12 DNAH11:19 TPPP2:33 DNAI2:36
HP_DECREASED_FERTILITY_IN_MALES	0.11260181	151	1.871e-06	7.730e-04	DZIP1:6 DNAH11:19 DNAI2:36 CFTR:81 TTC21A:240 SPAG17:247
CORD EVERACELLILAR TRANSPORT	0.00044040	0.0	0.470 00	0.400 0.4	DNALIA AO OFADAS AOO ODAOAT DATA DNALIO AOS DANAS TOO DATA SE

tissue\_specific Top pathways by permulation

0.22211043

num.genes pval p.adj

2.050e-18 1.107e-16

1352

GOBP\_EXTRACELLULAR\_TRANSPORT

Geneset

testis

0.07674690

2.176e-06 8.423e-04 DNAH11:19 CFAP45:132 SPAG17:247 DNAH9:435 DAW1:708 DNAH5:728

CEP19:2 FNDC11:8 FBXO24:23 OPLAH:32 TPPP2:33 DNAI2:36

, , ,	-0.05642624	646	2.278e-06	6.151e-05	MIS18BP1:29 RHEBL1:60 CENPE:74 BUB1:76 HAUS8:81 WDHD1:88
liver –	0.00440040				
	-0.06448218	324	8.711e-05	1.279e-03	TM4SF4:16 ANGPTL8:22 TAT:33 SLC38A3:38 ITIH2:49 RIDA:65
thyroid –	-0.09365461	148	9.478e-05	1.279e-03	CLCNKA:51 GPAT3:143 IDO2:165 GGT2:248 VEGFA:446 INPP5J:501
cerebellar.hemisphere –	-0.05186306	451	2.269e-04	2.450e-03	SPRN:13 ARVCF:32 ANK1:119 CA4:132 PISD:157 PCDHGC4:160
skeletal.muscle (	0.06522793	230	7.561e-04	6.805e-03	SLC8A3:13 NEXN:45 ASB15:53 TCAP:145 LMOD3:204 NRAP:231
left.ventricle (	0.10636034	64	3.352e-03	2.586e-02	SCN5A:49 ASB15:53 TCAP:145 LMOD3:204 NRAP:231 MYLK3:325
esophagus.mucosa (	0.04663158	218	1.895e-02	1.279e-01	PTK6:122 RHOD:187 SCEL:211 EVPLL:221 THSD4:255 RNF222:344
atrium.auricle (	0.06922450	87	2.627e-02	1.418e-01	CHRNE:42 SCN5A:49 ASB15:53 TCAP:145 LMOD3:204 NRAP:231
bladder -	-0.13534444	23	2.480e-02	1.418e-01	UGT1A6:625 SLC14A1:796 UPK1A:1996 ACER2:2283 ID1:3095 DHRS2:3160
ectocervix -	-0.12891639	24	2.898e-02	1.423e-01	LRFN5:814 RIPOR3:862 MMRN1:1023 KRT34:1216 TRHDE:1226 PAPPA2:1583
greater.omentum -	-0.10408703	31	4.519e-02	1.958e-01	TCF15:19 BTNL9:36 LPL:67 IL6:430 ADIPOQ:770 GFPT2:813
ovary (	0.05890821	96	4.713e-02	1.958e-01	RASL11B:10 SGCZ:84 C21orf62:171 SUSD4:172 CCDC170:173 ADRB3:200
Brodmann.area.9 –	-0.04338454	158	6.190e-02	2.089e-01	SERPINI1:468 INSM2:486 RXFP1:710 SYT16:717 FREM3:856 LRRTM4:876
vagina (	0.05828431	90	5.709e-02	2.089e-01	SCEL:211 RNF222:344 KRT13:524 TMPRSS11A:621 MAB21L4:685 LEXM:750
eye.development –	-0.07339557	56	5.813e-02	2.089e-01	SOX8:277 SP1:558 TYR:626 WNT5A:689 MSX1:1679 NOTCH1:1701
	-0.03322162	247	7.581e-02	2.408e-01	ADAM9:47 DNAJB4:52 LRP1:91 PTGER2:137 NRP1:173 LOX:227
leg.skin (	0.03294470	213	1.012e-01	3.035e-01	ANKRD35:26 PSORS1C2:44 POF1B:140 SCEL:211 EPHB3:261 RNF222:344
	-0.03378932	170	1.318e-01	3.747e-01	ADARB2:3 PAQR4:128 SEC14L5:278 ZNF488:511 LRAT:524 SCD:557
cortex.kidney –	-0.04033182	108	1.498e-01	3.916e-01	CLCNKB:50 BHMT:153 SLC12A1:221 CASR:265 SLC22A6:660 SLC5A10:822
prostate (	0.06812433	37	1.523e-01	3.916e-01	STEAP2:54 ACP3:55 NTF4:159 CHRNA2:167 TULP1:270 LMAN1L:359
esophagus.muscularis.mucosa (	0.07127844	26	2.089e-01	4.704e-01	KCNMB1:703 NTN1:952 CHRM3:1023 PRUNE2:1053 SYNM:1155 FAM83D:1376
minor.salivary.gland -	-0.04224839	69	2.265e-01	4.704e-01	BPIFB1:31 PROM1:269 ENPP3:986 TRPV4:1271 OVOL2:1484 GABRP:1666
sigmoid.colon (	0.05158618	47	2.222e-01	4.704e-01	PLA2G2C:436 HLX:513 FOXP2:743 BMP3:962 PDE9A:1066 PLEKHO1:1283
substantia.nigra –	-0.10772634	11	2.162e-01	4.704e-01	SLC6A3:680 DBX2:3240 TH:3510 C10orf105:4764 SLC6A11:5281 KCNE5:6143
suprapubic.skin (	0.02600009	186	2.257e-01	4.704e-01	PSORS1C2:44 SCEL:211 LTB4R:329 RNF222:344 RNF39:396 ANXA8L1:529
blood –	-0.02034884	267	2.589e-01	4.821e-01	SLC4A1:27 SLA:41 EPB42:42 SLC6A6:53 RHAG:75 PGLYRP1:83
caudate.nucleus -	-0.06417602	26	2.579e-01	4.821e-01	PSD2:35 MLC1:299 SYNDIG1L:915 ANO3:1003 RGS14:2064 SLCO1C1:2151
putamen –	-0.08925691	14	2.478e-01	4.821e-01	SYNDIG1L:915 ANO3:1003 KCNH4:3261 FAM237A:4721 SLC18A3:4795 GPR88:5681
peyers.patch (	0.02381057	164	2.967e-01	5.341e-01	TRPM5:342 HTR4:528 NLRP6:568 BCO1:713 KRT20:755 MUSK:810
aorta	0.02146840	150	3.677e-01	6.018e-01	TNFRSF11B:18 PDGFD:25 PCDH11X:38 PPP1R14A:83 S100A4:113 NOX4:156
nucleus.accumbens (	0.02859555	91	3.480e-01	6.018e-01	PCDH11X:38 CACNG4:85 KIRREL3:209 DACH2:383 POU3F4:459 OPRK1:635
pancreas -	-0.02740500	93	3.633e-01	6.018e-01	PNLIPRP1:120 CASR:265 CPA2:300 TMED6:497 GNMT:868 MATN4:1028
subcutaneous.adipose -	-0.02759553	70	4.262e-01	6.770e-01	ADGRL4:462 ACACB:611 ADIPOQ:770 GPAM:1197 AGTR1:1225 PRKAR2B:1375
cerebellum -	-0.01198493	356	4.457e-01	6.876e-01	SPRN:13 RTN4R:102 MTCL1:190 SLC1A6:226 ATP1A3:321 CBLN3:396
stomach -	-0.02194986	85	4.861e-01	7.291e-01	EPN3:82 MFSD4A:380 TMED6:497 PIK3C2G:1294 ATP4B:1371 GPRC5C:1372
breast	0.05113863	14	5.079e-01	7.413e-01	THRSP:386 ABCB5:1190 ALX4:1388 TNN:1402 HCAR1:2065 KRT5:3827
transverse.colon (	0.01537597	145	5.258e-01	7.471e-01	TRIM15:400 KRT20:755 SMPDL3B:766 LLGL2:858 ARHGEF16:929 LRRC31:1074
lung –	-0.01521252	138	5.401e-01	7.478e-01	PTPRB:34 SLCO2A1:46 GP9:242 PECAM1:312 IL18R1:475 GGTLC1:545
adrenal.gland -	-0.01358864	107	6.291e-01	7.710e-01	SLC14A2:1 CYP11A1:14 MCOLN3:23 ABCB1:209 MGST1:333 AKR1B1:559