|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Supplementary Table 8. Gene set enrichment analysis (MAGENTA) for African Americans of biological pathways with at least one gene from the confirmed loci1.** | | | | | | | | |
| Database | Gene set | Original No. of genes in gene set | No. of genes in gene set analyzed by MAGENTA | Nominal GSEA P value | FDR q-value2 | No. of genes expected above 75th percentile cutoff | No. of genes observed above 75th percentile cutoff | Genes within 300 kb from the confirmed SNPs |
| BIOCARTA |  |  |  |  |  |  |  |  |
|  | NKT\_PATHWAY | 28 | 25 | 0.002 | 0.03 | 6 | 13 | CSF2, IL5 |
|  | ERYTH\_PATHWAY | 15 | 14 | 0.009 | 0.12 | 4 | 8 | CSF2, IL3 |
| GOTERM |  |  |  |  |  |  |  |  |
|  | positive regulation of tyrosine phosphorylation of Stat5 protein | 10 | 9 | 0.009 | 0.15 | 2 | 6 | CSF2, IL3 |
|  | positive regulation of gene-specific transcription | 36 | 34 | 0.01 | 0.15 | 9 | 15 | IL5 |
|  | glucose transport | 23 | 23 | 0.01 | 0.15 | 6 | 11 | SLC2A5 |
|  | one-carbon metabolic process | 30 | 26 | 0.02 | 0.15 | 7 | 12 | CA6 |
|  | oxidoreductase activity, acting on paired donors, with incorporation or reduction of molecular oxygen | 16 | 16 | 0.03 | 0.15 | 4 | 8 | P4HA2 |
|  | magnesium ion binding | 157 | 147 | 0.03 | 0.15 | 37 | 47 | ENO1 |
|  | transcription repressor activity | 162 | 154 | 0.03 | 0.15 | 39 | 49 | ENO1 |
|  | glucose transmembrane transporter activity | 11 | 11 | 0.03 | 0.15 | 3 | 6 | SLC2A5 |
|  | glycolysis | 42 | 35 | 0.04 | 0.15 | 9 | 14 | ENO1 |
|  | positive regulation of transcription factor activity | 24 | 23 | 0.04 | 0.15 | 6 | 10 | IL5 |
|  | symporter activity | 113 | 103 | 0.04 | 0.15 | 26 | 34 | SLC22A4, SLC22A5 |
|  | negative regulation of cell growth | 88 | 80 | 0.04 | 0.15 | 20 | 27 | ENO1 |
|  | oxidoreductase activity, acting on single donors with incorporation of molecular oxygen, incorporation of two atoms of oxygen | 53 | 46 | 0.05 | 0.16 | 12 | 17 | P4HA2 |
| Ingenuity |  |  |  |  |  |  |  |  |
|  | GM-CSF.Signaling | 23 | 20 | 0.04 | 0.30 | 5 | 9 | CSF2 |
| KEGG |  |  |  |  |  |  |  |  |
|  | KEGG\_APOPTOSIS | 88 | 77 | 0.01 | 0.11 | 19 | 29 | IL3 |
|  | KEGG\_JAK\_STAT\_SIGNALING\_PATHWAY | 155 | 120 | 0.04 | 0.13 | 30 | 39 | CSF2, IL3, IL5 |
|  | KEGG\_ARGININE\_AND\_PROLINE\_METABOLISM | 54 | 48 | 0.04 | 0.13 | 12 | 18 | P4HA2 |
| PANTHER, BP |  |  |  |  |  |  |  |  |
|  | Carbohydrate\_transport | 50 | 49 | 0.02 | 0.36 | 12 | 19 | SLC2A5, SLC2A7 |
| PANTHER, MF |  |  |  |  |  |  |  |  |
|  | Carbohydrate\_transporter | 34 | 31 | 0.01 | 0.15 | 8 | 14 | SLC2A5, SLC2A7 |
|  | Other\_ligase | 146 | 121 | 0.04 | 0.26 | 30 | 39 | ACSL6 |
| 1. Pathways with a nominal P value < 0.05 are summarized. | | | | | | | | |
| 2. Multiple testing correction was conducted separately for each database due to considerable overlap of gene sets between databases. | | | | | | | | |