

# #606176 DIABETES MELLITUS, PERMANENT NEONATAL; PNDM

| OMIM Gene ID | HGNC   | UniProtAC |
|--------------|--------|-----------|
| 138079       | GCK    | P35557    |
| 176730       | INS    | P01308    |
| 600509       | ABCC8  | Q09428    |
| 600937       | KCNJ11 | Q14654    |

Table 1: OMIM - UniProtAC mapping

## Legend

- N1: #input proteins associated to the significant GO term
- N2: #proteins associated to the significant GO term
- P-value: Bonferroni-corrected p-value of Fisher's exact test
- *red*: go terms not related to the input proteins
- *blue*: go terms related to the input proteins (enriched uniquely by network-based method)
- *green*: go terms ancestors of terms enriched with the standard method (enriched uniquely by network-based method)

# 1 Standard enrichment

| GO Term    | N1 | N2   | P-value     | Description                                                    |
|------------|----|------|-------------|----------------------------------------------------------------|
| GO:0006112 | 4  | 185  | 3.28559e-07 | energy reserve metabolic process                               |
| GO:0015980 | 4  | 231  | 8.03914e-07 | energy derivation by oxidation of organic compounds            |
| GO:0050796 | 4  | 258  | 1.25439e-06 | regulation of insulin secretion                                |
| GO:0090276 | 4  | 279  | 1.71846e-06 | regulation of peptide hormone secretion                        |
| GO:0002791 | 4  | 284  | 1.8457e-06  | regulation of peptide secretion                                |
| GO:0090087 | 4  | 286  | 1.89852e-06 | regulation of peptide transport                                |
| GO:0046883 | 4  | 336  | 3.62807e-06 | regulation of hormone secretion                                |
| GO:0006091 | 4  | 531  | 2.27804e-05 | generation of precursor metabolites and energy                 |
| GO:0051046 | 4  | 829  | 0.000135885 | regulation of secretion                                        |
| GO:0007154 | 4  | 1103 | 0.000426617 | cell communication                                             |
| GO:0045721 | 2  | 14   | 0.000450562 | negative regulation of gluconeogenesis                         |
| GO:0045821 | 2  | 14   | 0.000450562 | positive regulation of glycolytic process                      |
| GO:0006006 | 3  | 225  | 0.000489528 | glucose metabolic process                                      |
| GO:0051048 | 3  | 225  | 0.000489528 | negative regulation of secretion                               |
| GO:0045725 | 2  | 15   | 0.000519861 | positive regulation of glycogen biosynthetic process           |
| GO:0070875 | 2  | 17   | 0.000673295 | positive regulation of glycogen metabolic process              |
| GO:0033500 | 3  | 264  | 0.000791707 | carbohydrate homeostasis                                       |
| GO:0042593 | 3  | 264  | 0.000791707 | glucose homeostasis                                            |
| GO:0060341 | 4  | 1363 | 0.000995796 | regulation of cellular localization                            |
| GO:0019318 | 3  | 321  | 0.00142448  | hexose metabolic process                                       |
| GO:0005975 | 4  | 1491 | 0.00142647  | carbohydrate metabolic process                                 |
| GO:0005996 | 3  | 362  | 0.00204348  | monosaccharide metabolic process                               |
| GO:0006110 | 2  | 32   | 0.00245425  | regulation of glycolytic process                               |
| GO:0005979 | 2  | 39   | 0.00366562  | regulation of glycogen biosynthetic process                    |
| GO:0010962 | 2  | 39   | 0.00366562  | regulation of glucan biosynthetic process                      |
| GO:0010907 | 2  | 40   | 0.00385841  | positive regulation of glucose metabolic process               |
| GO:0031018 | 2  | 42   | 0.00425879  | endocrine pancreas development                                 |
| GO:0043470 | 2  | 42   | 0.00425879  | regulation of carbohydrate catabolic process                   |
| GO:0043471 | 2  | 42   | 0.00425879  | regulation of cellular carbohydrate catabolic process          |
| GO:0010677 | 2  | 44   | 0.0046789   | negative regulation of cellular carbohydrate metabolic process |
| GO:0032885 | 2  | 45   | 0.00489635  | regulation of polysaccharide biosynthetic process              |
| GO:0051051 | 3  | 493  | 0.00515948  | negative regulation of transport                               |
| GO:0070873 | 2  | 47   | 0.00534604  | regulation of glycogen metabolic process                       |
| GO:0051049 | 4  | 2081 | 0.00541921  | regulation of transport                                        |
| GO:0055114 | 4  | 2084 | 0.00545055  | oxidation-reduction process                                    |
| GO:0045912 | 2  | 51   | 0.00630459  | negative regulation of carbohydrate metabolic process          |
| GO:0032881 | 2  | 53   | 0.00681339  | regulation of polysaccharide metabolic process                 |
| GO:0006111 | 2  | 57   | 0.00789014  | regulation of gluconeogenesis                                  |
| GO:0071417 | 3  | 588  | 0.00874574  | cellular response to organonitrogen compound                   |
| GO:0010676 | 2  | 61   | 0.00904573  | positive regulation of cellular carbohydrate metabolic process |
| GO:0045913 | 2  | 68   | 0.0112574   | positive regulation of carbohydrate metabolic process          |
| GO:1901699 | 3  | 645  | 0.0115357   | cellular response to nitrogen compound                         |
| GO:0046888 | 2  | 79   | 0.0152197   | negative regulation of hormone secretion                       |
| GO:0001678 | 2  | 86   | 0.0180507   | cellular glucose homeostasis                                   |
| GO:0032879 | 4  | 2827 | 0.0184706   | regulation of localization                                     |
| GO:0015758 | 2  | 88   | 0.0189038   | glucose transport                                              |
| GO:0043467 | 2  | 93   | 0.0211223   | regulation of generation of precursor metabolites and energy   |
| GO:0008645 | 2  | 95   | 0.022044    | hexose transport                                               |
| GO:0015749 | 2  | 97   | 0.0229854   | monosaccharide transport                                       |
| GO:0090277 | 2  | 101  | 0.0249269   | positive regulation of peptide hormone secretion               |
| GO:0002793 | 2  | 104  | 0.0264345   | positive regulation of peptide secretion                       |
| GO:0007267 | 3  | 859  | 0.0271627   | cell-cell signaling                                            |
| GO:0043255 | 2  | 111  | 0.0301238   | regulation of carbohydrate biosynthetic process                |
| GO:0023052 | 3  | 913  | 0.0325853   | signaling                                                      |
| GO:0044700 | 3  | 913  | 0.0325853   | single organism signaling                                      |
| GO:0051050 | 3  | 919  | 0.0332286   | positive regulation of transport                               |
| GO:0010827 | 2  | 128  | 0.0400817   | regulation of glucose transport                                |
| GO:0010906 | 2  | 133  | 0.0432794   | regulation of glucose metabolic process                        |
| GO:0046887 | 2  | 133  | 0.0432794   | positive regulation of hormone secretion                       |
| GO:0030001 | 3  | 1036 | 0.0475089   | metal ion transport                                            |

Table 2: Overrepresented GO terms with the standard enrichment

| GO Term    | N1 | N2   | P-value   | Description                                    |
|------------|----|------|-----------|------------------------------------------------|
| GO:0008643 | 2  | 140  | 0.0479612 | carbohydrate transport                         |
| GO:0044723 | 3  | 1049 | 0.0493087 | single-organism carbohydrate metabolic process |

Table 3: Overrepresented GO terms with the standard enrichment

## 2 Network-based enrichment

| GO Term    | N1 | N2   | P-value     | Description                                    |
|------------|----|------|-------------|------------------------------------------------|
| GO:0009743 | 4  | 561  | 0.000146    | response to carbohydrate                       |
| GO:0009730 | 2  | 5    | 0.00019838  | detection of carbohydrate stimulus             |
| GO:0009732 | 2  | 5    | 0.00019838  | detection of hexose stimulus                   |
| GO:0034287 | 2  | 5    | 0.00019838  | detection of monosaccharide stimulus           |
| GO:0051594 | 2  | 5    | 0.00019838  | detection of glucose                           |
| GO:0046676 | 3  | 105  | 0.00022278  | negative regulation of insulin secretion       |
| GO:0032811 | 2  | 9    | 0.000714055 | negative regulation of epinephrine secretion   |
| GO:0014060 | 2  | 11   | 0.00109083  | regulation of epinephrine secretion            |
| GO:0051156 | 2  | 20   | 0.00376696  | glucose 6-phosphate metabolic process          |
| GO:0033604 | 2  | 21   | 0.00416332  | negative regulation of catecholamine secretion |
| GO:0055082 | 4  | 1433 | 0.00625634  | cellular chemical homeostasis                  |
| GO:0071804 | 3  | 351  | 0.00844502  | cellular potassium ion transport               |
| GO:0071805 | 3  | 351  | 0.00844502  | potassium ion transmembrane transport          |
| GO:0043269 | 4  | 1590 | 0.00948644  | regulation of ion transport                    |
| GO:0044320 | 2  | 32   | 0.00982903  | cellular response to leptin stimulus           |
| GO:0032355 | 3  | 374  | 0.0102164   | response to estradiol                          |
| GO:0044321 | 2  | 37   | 0.0131952   | response to leptin                             |
| GO:0009749 | 3  | 416  | 0.0140573   | response to glucose                            |
| GO:0006813 | 3  | 419  | 0.0143633   | potassium ion transport                        |
| GO:0043279 | 3  | 424  | 0.0148833   | response to alkaloid                           |
| GO:0009746 | 3  | 439  | 0.0165178   | response to hexose                             |
| GO:0051953 | 2  | 42   | 0.0170553   | negative regulation of amine transport         |
| GO:0003013 | 3  | 444  | 0.017088    | circulatory system process                     |
| GO:0019725 | 4  | 1855 | 0.0175843   | cellular homeostasis                           |
| GO:0019932 | 3  | 457  | 0.0186315   | second-messenger-mediated signaling            |
| GO:0034284 | 3  | 458  | 0.0187539   | response to monosaccharide                     |
| GO:0046683 | 3  | 488  | 0.0226795   | response to organophosphorus                   |
| GO:0070509 | 2  | 49   | 0.0232885   | calcium ion import                             |
| GO:0005978 | 2  | 56   | 0.0304882   | glycogen biosynthetic process                  |
| GO:0009250 | 2  | 56   | 0.0304882   | glucan biosynthetic process                    |
| GO:0014074 | 3  | 555  | 0.0333359   | response to purine-containing compound         |
| GO:0009607 | 4  | 2197 | 0.0346169   | response to biotic stimulus                    |

Table 4: Overrepresented terms with the network-based enrichment. Only terms not detected with the standard method.