

# #210250 SITOSTEROLEMIA

| OMIM Gene ID | HGNC  | UniProtAC |
|--------------|-------|-----------|
| 605459       | ABCG5 | Q9H222    |
| 605460       | ABCG8 | Q9H221    |

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:0010949 | 2  | 2   | 1.88137e-07 | negative regulation of intestinal phytosterol absorption |
| GO:0045796 | 2  | 2   | 1.88137e-07 | negative regulation of intestinal cholesterol absorption |
| GO:0030299 | 2  | 8   | 5.26782e-06 | intestinal cholesterol absorption                        |
| GO:0030300 | 2  | 9   | 6.77292e-06 | regulation of intestinal cholesterol absorption          |
| GO:0032372 | 2  | 12  | 1.2417e-05  | negative regulation of sterol transport                  |
| GO:0032375 | 2  | 12  | 1.2417e-05  | negative regulation of cholesterol transport             |
| GO:0060457 | 2  | 18  | 2.87849e-05 | negative regulation of digestive system process          |
| GO:0050892 | 2  | 27  | 6.6036e-05  | intestinal absorption                                    |
| GO:0032369 | 2  | 28  | 7.11157e-05 | negative regulation of lipid transport                   |
| GO:0033344 | 2  | 31  | 8.74835e-05 | cholesterol efflux                                       |
| GO:0044058 | 2  | 38  | 0.00013226  | regulation of digestive system process                   |
| GO:0032371 | 2  | 48  | 0.000212218 | regulation of sterol transport                           |
| GO:0032374 | 2  | 48  | 0.000212218 | regulation of cholesterol transport                      |
| GO:0007588 | 2  | 51  | 0.000239875 | excretion                                                |
| GO:0022600 | 2  | 65  | 0.000391324 | digestive system process                                 |
| GO:0015918 | 2  | 66  | 0.000403553 | sterol transport                                         |
| GO:0030301 | 2  | 66  | 0.000403553 | cholesterol transport                                    |
| GO:0042632 | 2  | 95  | 0.00084003  | cholesterol homeostasis                                  |
| GO:0055092 | 2  | 96  | 0.000857903 | sterol homeostasis                                       |
| GO:0032368 | 2  | 112 | 0.00116946  | regulation of lipid transport                            |
| GO:0015850 | 2  | 139 | 0.00180442  | organic hydroxy compound transport                       |
| GO:0055088 | 2  | 140 | 0.00183057  | lipid homeostasis                                        |
| GO:0007584 | 2  | 307 | 0.00883697  | response to nutrient                                     |
| GO:0006869 | 2  | 348 | 0.0113593   | lipid transport                                          |
| GO:0031667 | 2  | 493 | 0.0228168   | response to nutrient levels                              |
| GO:0051051 | 2  | 493 | 0.0228168   | negative regulation of transport                         |
| GO:0009991 | 2  | 532 | 0.0265735   | response to extracellular stimulus                       |
| GO:0051241 | 2  | 547 | 0.0280946   | negative regulation of multicellular organismal process  |
| GO:0044057 | 2  | 554 | 0.028819    | regulation of system process                             |
| GO:0006200 | 2  | 569 | 0.0304022   | ATP catabolic process                                    |
| GO:0009158 | 2  | 574 | 0.0309393   | ribonucleoside monophosphate catabolic process           |
| GO:0009169 | 2  | 574 | 0.0309393   | purine ribonucleoside monophosphate catabolic process    |
| GO:0009128 | 2  | 575 | 0.0310473   | purine nucleoside monophosphate catabolic process        |
| GO:0009125 | 2  | 578 | 0.0313723   | nucleoside monophosphate catabolic process               |
| GO:0042493 | 2  | 633 | 0.0376326   | response to drug                                         |
| GO:0046034 | 2  | 656 | 0.0404192   | ATP metabolic process                                    |
| GO:0009167 | 2  | 698 | 0.0457648   | purine ribonucleoside monophosphate metabolic process    |
| GO:0009126 | 2  | 699 | 0.0458961   | purine nucleoside monophosphate metabolic process        |
| GO:0009161 | 2  | 727 | 0.0496494   | ribonucleoside monophosphate metabolic process           |

Table 2: Overrepresented GO terms with the standard enrichment

# 2 Network-based enrichment

*No novel enriched terms*