

## #202400 AFIBRINOGENEMIA, CONGENITAL

| OMIM Gene ID | HGNC | UniProtAC |
|--------------|------|-----------|
| 134820       | FGA  | P02671    |
| 134830       | FGB  | P02675    |

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:0034116 | 2  | 9   | 9.40122e-06 | positive regulation of heterotypic cell-cell adhesion                                   |
| GO:0034114 | 2  | 17  | 3.55158e-05 | regulation of heterotypic cell-cell adhesion                                            |
| GO:2000352 | 2  | 23  | 6.60696e-05 | negative regulation of endothelial cell apoptotic process                               |
| GO:1900026 | 2  | 31  | 0.000121432 | positive regulation of substrate adhesion-dependent cell spreading                      |
| GO:1902042 | 2  | 34  | 0.000146502 | negative regulation of extrinsic apoptotic signaling pathway via death domain receptors |
| GO:2000351 | 2  | 34  | 0.000146502 | regulation of endothelial cell apoptotic process                                        |
| GO:0045907 | 2  | 37  | 0.000173922 | positive regulation of vasoconstriction                                                 |
| GO:1900024 | 2  | 37  | 0.000173922 | regulation of substrate adhesion-dependent cell spreading                               |
| GO:0070527 | 2  | 48  | 0.000294572 | platelet aggregation                                                                    |
| GO:0022409 | 2  | 54  | 0.000373698 | positive regulation of cell-cell adhesion                                               |
| GO:0045921 | 2  | 55  | 0.000387801 | positive regulation of exocytosis                                                       |
| GO:1902041 | 2  | 59  | 0.000446819 | regulation of extrinsic apoptotic signaling pathway via death domain receptors          |
| GO:0034109 | 2  | 67  | 0.000577392 | homotypic cell-cell adhesion                                                            |
| GO:0019229 | 2  | 68  | 0.000594888 | regulation of vasoconstriction                                                          |
| GO:0010770 | 2  | 73  | 0.00068629  | positive regulation of cell morphogenesis involved in differentiation                   |
| GO:0002576 | 2  | 82  | 0.000867262 | platelet degranulation                                                                  |
| GO:0090277 | 2  | 101 | 0.00131878  | positive regulation of peptide hormone secretion                                        |
| GO:0002793 | 2  | 104 | 0.00139869  | positive regulation of peptide secretion                                                |
| GO:0051258 | 2  | 107 | 0.00148095  | protein polymerization                                                                  |
| GO:0022407 | 2  | 120 | 0.00186458  | regulation of cell-cell adhesion                                                        |
| GO:0046887 | 2  | 133 | 0.00229234  | positive regulation of hormone secretion                                                |
| GO:0051592 | 2  | 145 | 0.00272635  | response to calcium ion                                                                 |
| GO:0010811 | 2  | 150 | 0.0029183   | positive regulation of cell-substrate adhesion                                          |
| GO:0070374 | 2  | 150 | 0.0029183   | positive regulation of ERK1 and ERK2 cascade                                            |
| GO:0050714 | 2  | 162 | 0.00340559  | positive regulation of protein secretion                                                |
| GO:2001237 | 2  | 165 | 0.00353329  | negative regulation of extrinsic apoptotic signaling pathway                            |
| GO:0017157 | 2  | 167 | 0.00361973  | regulation of exocytosis                                                                |
| GO:0030168 | 2  | 216 | 0.00606379  | platelet activation                                                                     |
| GO:0007160 | 2  | 228 | 0.0067579   | cell-matrix adhesion                                                                    |
| GO:0070372 | 2  | 231 | 0.00693732  | regulation of ERK1 and ERK2 cascade                                                     |
| GO:0010810 | 2  | 243 | 0.00767845  | regulation of cell-substrate adhesion                                                   |
| GO:0045785 | 2  | 253 | 0.00832478  | positive regulation of cell adhesion                                                    |
| GO:2001236 | 2  | 253 | 0.00832478  | regulation of extrinsic apoptotic signaling pathway                                     |
| GO:0050708 | 2  | 273 | 0.00969579  | regulation of protein secretion                                                         |
| GO:0072377 | 1  | 1   | 0.00985614  | blood coagulation, common pathway                                                       |
| GO:2000260 | 1  | 1   | 0.00985614  | regulation of blood coagulation, common pathway                                         |
| GO:2000261 | 1  | 1   | 0.00985614  | negative regulation of blood coagulation, common pathway                                |
| GO:0090276 | 2  | 279 | 0.0101275   | regulation of peptide hormone secretion                                                 |
| GO:0002791 | 2  | 284 | 0.0104944   | regulation of peptide secretion                                                         |
| GO:0090087 | 2  | 286 | 0.010643    | regulation of peptide transport                                                         |
| GO:2001234 | 2  | 286 | 0.010643    | negative regulation of apoptotic signaling pathway                                      |
| GO:0031589 | 2  | 292 | 0.011095    | cell-substrate adhesion                                                                 |
| GO:0046883 | 2  | 336 | 0.0146972   | regulation of hormone secretion                                                         |
| GO:0006887 | 2  | 346 | 0.0155864   | exocytosis                                                                              |
| GO:0010720 | 2  | 348 | 0.0157674   | positive regulation of cell development                                                 |
| GO:0051222 | 2  | 349 | 0.0158583   | positive regulation of protein transport                                                |
| GO:0010769 | 2  | 381 | 0.0189043   | regulation of cell morphogenesis involved in differentiation                            |
| GO:0016337 | 2  | 388 | 0.0196063   | single organismal cell-cell adhesion                                                    |
| GO:0051047 | 2  | 392 | 0.020013    | positive regulation of secretion                                                        |
| GO:0043623 | 2  | 393 | 0.0201155   | cellular protein complex assembly                                                       |
| GO:0010038 | 2  | 402 | 0.0210485   | response to metal ion                                                                   |
| GO:0098602 | 2  | 425 | 0.0235292   | single organism cell adhesion                                                           |
| GO:0060627 | 2  | 451 | 0.0264996   | regulation of vesicle-mediated transport                                                |
| GO:0030155 | 2  | 485 | 0.0306506   | regulation of cell adhesion                                                             |
| GO:0030198 | 2  | 486 | 0.0307772   | extracellular matrix organization                                                       |
| GO:0043062 | 2  | 487 | 0.0309041   | extracellular structure organization                                                    |
| GO:0007596 | 2  | 501 | 0.0327085   | blood coagulation                                                                       |
| GO:0050817 | 2  | 501 | 0.0327085   | coagulation                                                                             |
| GO:0007599 | 2  | 510 | 0.0338953   | hemostasis                                                                              |
| GO:0043410 | 2  | 539 | 0.0378636   | positive regulation of MAPK cascade                                                     |

Table 2: Overrepresented GO terms with the standard enrichment

| GO Term    | N1 | N2  | P-value   | Description                               |
|------------|----|-----|-----------|-------------------------------------------|
| GO:2001233 | 2  | 541 | 0.0381454 | regulation of apoptotic signaling pathway |
| GO:0044057 | 2  | 554 | 0.0400025 | regulation of system process              |
| GO:0032940 | 2  | 592 | 0.0456836 | secretion by cell                         |
| GO:0022604 | 2  | 593 | 0.0458382 | regulation of cell morphogenesis          |
| GO:0051223 | 2  | 595 | 0.0461483 | regulation of protein transport           |
| GO:0010035 | 2  | 612 | 0.0488252 | response to inorganic substance           |

Table 3: Overrepresented GO terms with the standard enrichment

## 2 Network-based enrichment

| GO Term                    | N1 | N2  | P-value     | Description                          |
|----------------------------|----|-----|-------------|--------------------------------------|
| <a href="#">GO:0044320</a> | 2  | 32  | 0.000331604 | cellular response to leptin stimulus |
| <a href="#">GO:0044321</a> | 2  | 37  | 0.000445259 | response to leptin                   |
| <a href="#">GO:0070555</a> | 2  | 246 | 0.0201469   | response to interleukin-1            |

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