

# #163200 SCHIMMELPENNING-FEUERSTEIN-MIMS SYNDROME; SFM

| OMIM Gene ID | HGNC | UniProtAC |
|--------------|------|-----------|
| 190020       | HRAS | P01112    |
| 190070       | KRAS | P01116    |

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:0035022 | 2  | 4   | 3.46228e-06 | positive regulation of Rac protein signal transduction           |
| GO:0035020 | 2  | 18  | 8.82881e-05 | regulation of Rac protein signal transduction                    |
| GO:0048169 | 2  | 37  | 0.000384313 | regulation of long-term neuronal synaptic plasticity             |
| GO:0060441 | 2  | 39  | 0.000427592 | epithelial tube branching involved in lung morphogenesis         |
| GO:0032228 | 2  | 48  | 0.000650909 | regulation of synaptic transmission, GABAergic                   |
| GO:0035176 | 2  | 50  | 0.000706883 | social behavior                                                  |
| GO:0051703 | 2  | 50  | 0.000706883 | intraspecies interaction between organisms                       |
| GO:0008542 | 2  | 58  | 0.000953857 | visual learning                                                  |
| GO:0007632 | 2  | 63  | 0.00112697  | visual behavior                                                  |
| GO:0048168 | 2  | 66  | 0.00123776  | regulation of neuronal synaptic plasticity                       |
| GO:0046579 | 2  | 67  | 0.00127585  | positive regulation of Ras protein signal transduction           |
| GO:0051057 | 2  | 76  | 0.00164458  | positive regulation of small GTPase mediated signal transduction |
| GO:0000186 | 2  | 83  | 0.00196369  | activation of MAPKK activity                                     |
| GO:0008306 | 2  | 89  | 0.00225971  | associative learning                                             |
| GO:0007146 | 2  | 93  | 0.0024686   | striated muscle cell differentiation                             |
| GO:0051705 | 2  | 116 | 0.0038489   | multi-organism behavior                                          |
| GO:0007612 | 2  | 163 | 0.00761875  | learning                                                         |
| GO:0008286 | 2  | 195 | 0.0109148   | insulin receptor signaling pathway                               |
| GO:0042692 | 2  | 200 | 0.0114832   | muscle cell differentiation                                      |
| GO:0007265 | 2  | 201 | 0.0115986   | Ras protein signal transduction                                  |
| GO:0007173 | 2  | 202 | 0.0117146   | epidermal growth factor receptor signaling pathway               |
| GO:0046578 | 2  | 203 | 0.0118312   | regulation of Ras protein signal transduction                    |
| GO:0038127 | 2  | 205 | 0.012066    | ERBB signaling pathway                                           |
| GO:0048167 | 2  | 210 | 0.0126633   | regulation of synaptic plasticity                                |
| GO:0008543 | 2  | 211 | 0.0127844   | fibroblast growth factor receptor signaling pathway              |
| GO:0043524 | 2  | 216 | 0.013399    | negative regulation of neuron apoptotic process                  |
| GO:0044344 | 2  | 237 | 0.0161377   | cellular response to fibroblast growth factor stimulus           |
| GO:0071774 | 2  | 243 | 0.0169669   | response to fibroblast growth factor                             |
| GO:0048754 | 2  | 244 | 0.0171071   | branching morphogenesis of an epithelial tube                    |
| GO:1901215 | 2  | 244 | 0.0171071   | negative regulation of neuron death                              |
| GO:0000165 | 2  | 249 | 0.0178169   | MAPK cascade                                                     |
| GO:0050900 | 2  | 265 | 0.0201851   | leukocyte migration                                              |
| GO:0032869 | 2  | 270 | 0.0209554   | cellular response to insulin stimulus                            |
| GO:0043406 | 2  | 275 | 0.0217402   | positive regulation of MAP kinase activity                       |
| GO:0048011 | 2  | 276 | 0.0218989   | neurotrophin TRK receptor signaling pathway                      |
| GO:0038179 | 2  | 285 | 0.0233531   | neurotrophin signaling pathway                                   |
| GO:0061138 | 2  | 286 | 0.0235175   | morphogenesis of a branching epithelium                          |
| GO:0038095 | 2  | 294 | 0.024854    | Fc-epsilon receptor signaling pathway                            |
| GO:0043523 | 2  | 300 | 0.0258805   | regulation of neuron apoptotic process                           |
| GO:0001763 | 2  | 303 | 0.0264016   | morphogenesis of a branching structure                           |
| GO:0023014 | 2  | 309 | 0.0274593   | signal transduction by phosphorylation                           |
| GO:0007611 | 2  | 329 | 0.0311351   | learning or memory                                               |
| GO:1901214 | 2  | 348 | 0.0348409   | regulation of neuron death                                       |
| GO:0038093 | 2  | 350 | 0.0352431   | Fc receptor signaling pathway                                    |
| GO:0032147 | 2  | 357 | 0.036669    | activation of protein kinase activity                            |
| GO:0071902 | 2  | 357 | 0.036669    | positive regulation of protein serine/threonine kinase activity  |
| GO:0050890 | 2  | 365 | 0.0383332   | cognition                                                        |
| GO:0071375 | 2  | 372 | 0.0398197   | cellular response to peptide hormone stimulus                    |
| GO:0032868 | 2  | 376 | 0.0406818   | response to insulin                                              |
| GO:0043405 | 2  | 381 | 0.0417724   | regulation of MAP kinase activity                                |
| GO:0050804 | 2  | 390 | 0.0437719   | regulation of synaptic transmission                              |
| GO:1901653 | 2  | 391 | 0.0439967   | cellular response to peptide                                     |
| GO:0051056 | 2  | 404 | 0.0469748   | regulation of small GTPase mediated signal transduction          |

Table 2: Overrepresented GO terms with the standard enrichment

## 2 Network-based enrichment

| GO Term    | N1 | N2  | P-value    | Description                                    |
|------------|----|-----|------------|------------------------------------------------|
| GO:0002548 | 2  | 44  | 0.00220933 | monocyte chemotaxis                            |
| GO:0019320 | 2  | 84  | 0.00814134 | hexose catabolic process                       |
| GO:0045055 | 2  | 85  | 0.00833752 | regulated secretory pathway                    |
| GO:0046365 | 2  | 93  | 0.00999101 | monosaccharide catabolic process               |
| GO:0008361 | 2  | 104 | 0.0125086  | regulation of cell size                        |
| GO:0051353 | 2  | 104 | 0.0125086  | positive regulation of oxidoreductase activity |
| GO:0030593 | 2  | 113 | 0.0147787  | neutrophil chemotaxis                          |
| GO:0048640 | 2  | 122 | 0.0172378  | negative regulation of developmental growth    |
| GO:1990266 | 2  | 123 | 0.0175228  | neutrophil migration                           |
| GO:0042446 | 2  | 141 | 0.0230508  | hormone biosynthetic process                   |
| GO:0071621 | 2  | 142 | 0.0233801  | granulocyte chemotaxis                         |
| GO:0018279 | 2  | 145 | 0.024382   | protein N-linked glycosylation via asparagine  |
| GO:0018196 | 2  | 146 | 0.0247206  | peptidyl-asparagine modification               |
| GO:0097530 | 2  | 154 | 0.0275138  | granulocyte migration                          |
| GO:0032330 | 2  | 157 | 0.0285997  | regulation of chondrocyte differentiation      |
| GO:0032411 | 2  | 158 | 0.0289664  | positive regulation of transporter activity    |
| GO:0002028 | 2  | 181 | 0.0380443  | regulation of sodium ion transport             |
| GO:0030072 | 2  | 189 | 0.0414914  | peptide hormone secretion                      |
| GO:0002790 | 2  | 199 | 0.0460105  | peptide secretion                              |
| GO:0061035 | 2  | 207 | 0.0497939  | regulation of cartilage development            |

Table 3: Overrepresented terms with the network-based enrichment. Only terms not detected with the standard method.