

# #187500 TETRALOGY OF FALLOT; TOF

| OMIM Gene ID | HGNC   | UniProtAC |
|--------------|--------|-----------|
| 600584       | NKX2-5 | P52952    |
| 601656       | GATA6  | Q92908    |
| 601920       | JAG1   | P78504    |
| 602054       | TBX1   | O43435    |
| 602880       | GDF1   | P27539    |
| 603693       | ZFPM2  | Q8WW38    |

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:0060411 | 5  | 61   | 3.35223e-11 | cardiac septum morphogenesis                                            |
| GO:0003148 | 4  | 18   | 3.25567e-10 | outflow tract septum morphogenesis                                      |
| GO:0003221 | 2  | 2    | 1.26361e-05 | right ventricular cardiac muscle tissue morphogenesis                   |
| GO:0060415 | 3  | 71   | 7.62342e-05 | muscle tissue morphogenesis                                             |
| GO:0051891 | 2  | 5    | 0.000126334 | positive regulation of cardioblast differentiation                      |
| GO:0045944 | 5  | 1312 | 0.000176146 | positive regulation of transcription from RNA polymerase II promoter    |
| GO:0051094 | 5  | 1326 | 0.000185702 | positive regulation of developmental process                            |
| GO:2000736 | 3  | 117  | 0.000346014 | regulation of stem cell differentiation                                 |
| GO:0051890 | 2  | 9    | 0.000454673 | regulation of cardioblast differentiation                               |
| GO:0048729 | 4  | 578  | 0.000478111 | tissue morphogenesis                                                    |
| GO:0035239 | 3  | 141  | 0.000607422 | tube morphogenesis                                                      |
| GO:0045893 | 5  | 1762 | 0.000763152 | positive regulation of transcription, DNA-templated                     |
| GO:0042693 | 2  | 12   | 0.000833388 | muscle cell fate commitment                                             |
| GO:1902680 | 5  | 1811 | 0.000874488 | positive regulation of RNA biosynthetic process                         |
| GO:0051254 | 5  | 1838 | 0.000941142 | positive regulation of RNA metabolic process                            |
| GO:0048513 | 5  | 1910 | 0.00113884  | organ development                                                       |
| GO:2000738 | 2  | 14   | 0.0011489   | positive regulation of stem cell differentiation                        |
| GO:0010628 | 5  | 1919 | 0.00116571  | positive regulation of gene expression                                  |
| GO:0010557 | 5  | 2042 | 0.00158635  | positive regulation of macromolecule biosynthetic process               |
| GO:0045935 | 5  | 2060 | 0.00165689  | positive regulation of nucleobase-containing compound metabolic process |
| GO:0042692 | 3  | 200  | 0.00173838  | muscle cell differentiation                                             |
| GO:0016202 | 3  | 203  | 0.00181787  | regulation of striated muscle tissue development                        |
| GO:1901861 | 3  | 204  | 0.00184489  | regulation of muscle tissue development                                 |
| GO:0051173 | 5  | 2108 | 0.00185727  | positive regulation of nitrogen compound metabolic process              |
| GO:0045595 | 5  | 2111 | 0.00187041  | regulation of cell differentiation                                      |
| GO:0048634 | 3  | 207  | 0.00192755  | regulation of muscle organ development                                  |
| GO:0009653 | 5  | 2131 | 0.00195989  | anatomical structure morphogenesis                                      |
| GO:0045596 | 4  | 834  | 0.00205631  | negative regulation of cell differentiation                             |
| GO:0031328 | 5  | 2177 | 0.00217864  | positive regulation of cellular biosynthetic process                    |
| GO:0009891 | 5  | 2208 | 0.00233671  | positive regulation of biosynthetic process                             |
| GO:2000026 | 5  | 2229 | 0.00244887  | regulation of multicellular organismal development                      |
| GO:0006357 | 5  | 2379 | 0.00338062  | regulation of transcription from RNA polymerase II promoter             |
| GO:0035909 | 2  | 25   | 0.00378466  | aorta morphogenesis                                                     |
| GO:0060037 | 2  | 25   | 0.00378466  | pharyngeal system development                                           |
| GO:0030154 | 5  | 2446 | 0.00387862  | cell differentiation                                                    |
| GO:0009888 | 4  | 984  | 0.00396341  | tissue development                                                      |
| GO:0045597 | 4  | 985  | 0.00397939  | positive regulation of cell differentiation                             |
| GO:0060412 | 2  | 26   | 0.00409976  | ventricular septum morphogenesis                                        |
| GO:0048534 | 3  | 272  | 0.00437147  | hematopoietic or lymphoid organ development                             |
| GO:0051093 | 4  | 1051 | 0.00514528  | negative regulation of developmental process                            |
| GO:0030878 | 2  | 34   | 0.0070728   | thyroid gland development                                               |
| GO:0060548 | 4  | 1147 | 0.00727194  | negative regulation of cell death                                       |
| GO:0048646 | 4  | 1201 | 0.00872274  | anatomical structure formation involved in morphogenesis                |
| GO:0050793 | 5  | 2884 | 0.00875346  | regulation of developmental process                                     |
| GO:0048732 | 3  | 391  | 0.0129365   | gland development                                                       |
| GO:0003007 | 2  | 50   | 0.0154267   | heart morphogenesis                                                     |
| GO:0010604 | 5  | 3285 | 0.0166317   | positive regulation of macromolecule metabolic process                  |
| GO:0055010 | 2  | 53   | 0.0173498   | ventricular cardiac muscle tissue morphogenesis                         |
| GO:0001974 | 2  | 55   | 0.0186944   | blood vessel remodeling                                                 |
| GO:0060043 | 2  | 56   | 0.0193854   | regulation of cardiac muscle cell proliferation                         |
| GO:0030855 | 3  | 451  | 0.0198014   | epithelial cell differentiation                                         |
| GO:0031325 | 5  | 3418 | 0.0202206   | positive regulation of cellular metabolic process                       |
| GO:0051239 | 5  | 3432 | 0.0206315   | regulation of multicellular organismal process                          |
| GO:0055006 | 2  | 58   | 0.0208049   | cardiac cell development                                                |
| GO:0055007 | 2  | 59   | 0.0215333   | cardiac muscle cell differentiation                                     |
| GO:0055021 | 2  | 62   | 0.0237937   | regulation of cardiac muscle tissue growth                              |
| GO:0001709 | 2  | 64   | 0.0253629   | cell fate determination                                                 |
| GO:0048844 | 2  | 65   | 0.0261662   | artery morphogenesis                                                    |
| GO:0055008 | 2  | 65   | 0.0261662   | cardiac muscle tissue morphogenesis                                     |
| GO:0009893 | 5  | 3630 | 0.0271855   | positive regulation of metabolic process                                |

Table 2: Overrepresented GO terms with the standard enrichment

| GO Term    | N1 | N2   | P-value   | Description                                     |
|------------|----|------|-----------|-------------------------------------------------|
| GO:0007498 | 2  | 67   | 0.0278102 | mesoderm development                            |
| GO:0048869 | 5  | 3694 | 0.0296239 | cellular developmental process                  |
| GO:0060420 | 2  | 70   | 0.0303697 | regulation of heart growth                      |
| GO:0045599 | 2  | 72   | 0.0321383 | negative regulation of fat cell differentiation |
| GO:0035051 | 2  | 76   | 0.0358249 | cardiocyte differentiation                      |
| GO:0055024 | 2  | 77   | 0.0367776 | regulation of cardiac muscle tissue development |
| GO:0060485 | 2  | 77   | 0.0367776 | mesenchyme development                          |

Table 3: Overrepresented GO terms with the standard enrichment

## 2 Network-based enrichment

| GO Term    | N1 | N2   | P-value     | Description                                                               |
|------------|----|------|-------------|---------------------------------------------------------------------------|
| GO:0001701 | 5  | 893  | 6.36773e-05 | in utero embryonic development                                            |
| GO:0043009 | 5  | 909  | 6.95753e-05 | chordate embryonic development                                            |
| GO:0009792 | 5  | 937  | 8.09407e-05 | embryo development ending in birth or egg hatching                        |
| GO:0009790 | 5  | 1155 | 0.000229521 | embryo development                                                        |
| GO:0010621 | 2  | 6    | 0.000321425 | negative regulation of transcription by transcription factor localization |
| GO:0003151 | 3  | 129  | 0.000891919 | outflow tract morphogenesis                                               |
| GO:0030182 | 4  | 911  | 0.00632372  | neuron differentiation                                                    |
| GO:0001570 | 3  | 258  | 0.00715676  | vasculogenesis                                                            |
| GO:0003215 | 2  | 28   | 0.00808567  | cardiac right ventricle morphogenesis                                     |
| GO:1902533 | 5  | 2418 | 0.0089701   | positive regulation of intracellular signal transduction                  |
| GO:2000018 | 2  | 30   | 0.00930348  | regulation of male gonad development                                      |
| GO:0048514 | 3  | 294  | 0.0105794   | blood vessel morphogenesis                                                |
| GO:0090288 | 3  | 301  | 0.0113505   | negative regulation of cellular response to growth factor stimulus        |
| GO:0048731 | 5  | 2612 | 0.0131301   | system development                                                        |
| GO:0014706 | 3  | 333  | 0.0153505   | striated muscle tissue development                                        |
| GO:0002009 | 4  | 1159 | 0.0163888   | morphogenesis of an epithelium                                            |
| GO:0001101 | 4  | 1170 | 0.0170115   | response to acid chemical                                                 |
| GO:0060537 | 3  | 387  | 0.0240367   | muscle tissue development                                                 |
| GO:0007389 | 4  | 1481 | 0.0430524   | pattern specification process                                             |
| GO:0001932 | 5  | 3361 | 0.0454277   | regulation of protein phosphorylation                                     |
| GO:0003208 | 2  | 68   | 0.048572    | cardiac ventricle morphogenesis                                           |

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