

## GeDiPNet Cluster 117 -- Top Shared Genes

Curated genes (disease\_gdp) linked to 2 or more of this cluster's member diseases, most widely shared first. Data as of: 2026-10-08 03:15:01. · Rows: 77 · Generated: 2026-10-08 15:00:00

| Gene     | Member Diseases Linked | Cluster Size | Linked Diseases                                                                                                                                                                                |
|----------|------------------------|--------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| P4HB     | 6                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Cole-carpenter syndrome; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome      |
| B4GALT7  | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; ehlers-danlos syndrome, spondylodysplastic type, 1; Larsen syndrome; Osteoporosis-pseudoglioma syndrome |
| COL1A1   | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| COL1A2   | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| CRTAP    | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Cole-carpenter syndrome; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                    |
| GORAB    | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Larsen syndrome; Osteoporosis; Osteoporosis-pseudoglioma syndrome                                       |
| IFITM5   | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| LRP5     | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| PLS3     | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| SEC24D   | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Cole-carpenter syndrome; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                    |
| SERPINF1 | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| SP7      | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| WNT1     | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis; Osteoporosis-pseudoglioma syndrome                               |
| XYLT2    | 5                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Larsen syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                            |
| ALPL     | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| B3GALT6  | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Larsen syndrome; Osteoporosis-pseudoglioma syndrome                                                     |
| B3GAT3   | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Larsen syndrome; Osteoporosis-pseudoglioma syndrome                                                     |
| BMP1     | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| CCDC134  | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| CHST3    | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Larsen syndrome; Osteoporosis-pseudoglioma syndrome                                                     |
| CREB3L1  | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| FKBP10   | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| KDELR2   | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| MBTPS2   | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| MESD     | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| P3H1     | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| PLOD2    | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| PPIB     | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |
| SERPINH1 | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                                             |

| Gene       | Member Diseases Linked | Cluster Size | Linked Diseases                                                                                                                                    |
|------------|------------------------|--------------|----------------------------------------------------------------------------------------------------------------------------------------------------|
| SPARC      | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome |
| TENT5A     | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome |
| TMEM38B    | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome |
| XYLT1      | 4                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Larsen syndrome; Osteoporosis-pseudoglioma syndrome         |
| COPB2      | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis; Osteoporosis-pseudoglioma syndrome                                 |
| CSGALNACT1 | 3                      | 10           | Desbuquois syndrome; Larsen syndrome; skeletal dysplasia, mild, with joint laxity and advanced bone age                                            |
| LIFR       | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteoporosis-pseudoglioma syndrome                          |
| NBAS       | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteoporosis-pseudoglioma syndrome                          |
| P4HA1      | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Larsen syndrome; Osteoporosis-pseudoglioma syndrome                              |
| PHLDB1     | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteogenesis imperfecta; Osteoporosis-pseudoglioma syndrome                      |
| PLOD3      | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Larsen syndrome; Osteoporosis-pseudoglioma syndrome                              |
| SGMS2      | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteoporosis-pseudoglioma syndrome                          |
| SLC10A7    | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteoporosis-pseudoglioma syndrome                          |
| TAPT1      | 3                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Desbuquois syndrome; Osteoporosis-pseudoglioma syndrome                          |
| AMBN       | 2                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis-pseudoglioma syndrome                                               |
| ANO5       | 2                      | 10           | Desbuquois syndrome; Osteogenesis imperfecta                                                                                                       |
| ANTXR2     | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| BMP2       | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| CA2        | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| CANT1      | 2                      | 10           | Desbuquois syndrome; Larsen syndrome                                                                                                               |
| COL11A1    | 2                      | 10           | Desbuquois syndrome; Osteogenesis imperfecta                                                                                                       |
| DSPP       | 2                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis-pseudoglioma syndrome                                               |
| EMILIN1    | 2                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis-pseudoglioma syndrome                                               |
| FGFR3      | 2                      | 10           | Desbuquois syndrome; Larsen syndrome                                                                                                               |
| FLNB       | 2                      | 10           | Desbuquois syndrome; Larsen syndrome                                                                                                               |
| GALNT3     | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| GPC6       | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| GZF1       | 2                      | 10           | Desbuquois syndrome; Larsen syndrome                                                                                                               |
| IDH2       | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| IGF1R      | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| IL1RN      | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                                                                  |
| KIF22      | 2                      | 10           | Desbuquois syndrome; Larsen syndrome                                                                                                               |
| KIF5B      | 2                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis-pseudoglioma syndrome                                               |

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|-----------|------------------------|--------------|--------------------------------------------------------------------------------------------------------------|
| KL        | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| MECOM     | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| MIA3      | 2                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis-pseudoglioma syndrome         |
| OFD1      | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| PGGHG     | 2                      | 10           | Osteogenesis imperfecta; Osteoporosis                                                                        |
| PIEZO2    | 2                      | 10           | Larsen syndrome; Osteoporosis                                                                                |
| PPP1R21   | 2                      | 10           | Desbuquois syndrome; neurodevelopmental disorder with hypotonia, facial dysmorphism, and brain abnormalities |
| SEC16B    | 2                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis-pseudoglioma syndrome         |
| SFRP4     | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| SKI       | 2                      | 10           | Desbuquois syndrome; Larsen syndrome                                                                         |
| TGFB1     | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| TNFRSF11A | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| TNFRSF11B | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| TNFSF11   | 2                      | 10           | Desbuquois syndrome; Osteoporosis                                                                            |
| WNT11     | 2                      | 10           | Bone fragility with contractures, arterial rupture, and deafness; Osteoporosis-pseudoglioma syndrome         |