

## GeDiPNet Cluster 12 -- 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: 2026-10-10 03:15:01. · Rows: 116 · Generated: 2026-10-10 06:05:12

| Gene    | Member Diseases Linked | Cluster Size | Linked Diseases                                                                                                                                                                                                                                                                                                                                                              |
|---------|------------------------|--------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| FBN1    | 13                     | 24           | Aneurysm; Aortic aneurysm; Congenital aneurysm of ascending aorta; Congenital contractural arachnodactyly; Connective tissue disease; Ehlers-danlos syndrome; familial thoracic aortic aneurysm and aortic dissection; Loeys-dietz syndrome; Marfan syndrome; Mitral valve prolapse; Mixed connective tissue disease; Myopia; Thoracic aortic aneurysm and aortic dissection |
| SMAD3   | 10                     | 24           | Aneurysm; Aortic aneurysm; Congenital aneurysm of ascending aorta; Congenital contractural arachnodactyly; Connective tissue disease; Ehlers-danlos syndrome; Keratoconus; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                             |
| LOX     | 9                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; Congenital contractural arachnodactyly; Connective tissue disease; Ehlers-danlos syndrome; familial thoracic aortic aneurysm and aortic dissection; Keratoconus; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                    |
| MYH11   | 9                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; Connective tissue disease; Ehlers-danlos syndrome; familial thoracic aortic aneurysm and aortic dissection; Loeys-dietz syndrome; Marfan syndrome; Myopia; Thoracic aortic aneurysm and aortic dissection                                                                                                           |
| COL3A1  | 8                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; Congenital contractural arachnodactyly; Connective tissue disease; Ehlers-danlos syndrome; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                    |
| FBN2    | 8                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Connective tissue disease; Ehlers-danlos syndrome; Loeys-dietz syndrome; Marfan syndrome; Myopia; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                    |
| FLNA    | 8                      | 24           | Aortic aneurysm; Connective tissue disease; Ehlers-danlos syndrome; familial thoracic aortic aneurysm and aortic dissection; Marfan syndrome; Mitral valve prolapse; Myopia; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                  |
| TGFB R2 | 8                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; Congenital contractural arachnodactyly; Connective tissue disease; Ehlers-danlos syndrome; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                    |
| COL5A1  | 7                      | 24           | Connective tissue disease; Ehlers-danlos syndrome; Keratoconus; Loeys-dietz syndrome; Marfan syndrome; Mitral valve prolapse; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                 |
| MYLK    | 7                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; Connective tissue disease; familial thoracic aortic aneurysm and aortic dissection; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                           |
| TGFB 2  | 7                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Ehlers-danlos syndrome; familial thoracic aortic aneurysm and aortic dissection; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                              |
| COL5A2  | 6                      | 24           | Connective tissue disease; Ehlers-danlos syndrome; Keratoconus; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                                        |
| MFAP5   | 6                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Connective tissue disease; familial thoracic aortic aneurysm and aortic dissection; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                 |
| TGFB R1 | 6                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Ehlers-danlos syndrome; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                       |
| ACTA2   | 5                      | 24           | Aneurysm; Aortic aneurysm; Connective tissue disease; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                                                                        |
| COL1A1  | 5                      | 24           | Connective tissue disease; Ehlers-danlos syndrome; Keratoconus; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                                                              |
| NDE1    | 5                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; Connective tissue disease; Myopia; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                                                   |
| PLOD1   | 5                      | 24           | Congenital contractural arachnodactyly; Ehlers-danlos syndrome; Keratoconus; Mitral valve prolapse; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                                           |
| PRKG1   | 5                      | 24           | Aortic aneurysm; Connective tissue disease; familial thoracic aortic aneurysm and aortic dissection; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                         |
| SLC2A10 | 5                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Ehlers-danlos syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                                                                                                                                                                             |

| Gene     | Member Diseases Linked | Cluster Size | Linked Diseases                                                                                                                                                                        |
|----------|------------------------|--------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| SMAD2    | 5                      | 24           | Congenital contractural arachnodactyly; Loeys-dietz syndrome; loeys-dietz syndrome 6; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                  |
| TGFB3    | 5                      | 24           | Congenital contractural arachnodactyly; familial thoracic aortic aneurysm and aortic dissection; Loeys-dietz syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection |
| BGN      | 4                      | 24           | Congenital contractural arachnodactyly; familial thoracic aortic aneurysm and aortic dissection; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                       |
| COL2A1   | 4                      | 24           | Congenital aneurysm of ascending aorta; Connective tissue disease; Marfan syndrome; Myopia                                                                                             |
| FOXE3    | 4                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; familial thoracic aortic aneurysm and aortic dissection; Thoracic aortic aneurysm and aortic dissection                       |
| NOTCH1   | 4                      | 24           | Connective tissue disease; Ehlers-danlos syndrome; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                                     |
| SLC39A13 | 4                      | 24           | Connective tissue disease; Ehlers-danlos syndrome; Mitral valve prolapse; Myopia                                                                                                       |
| SRFBP1   | 4                      | 24           | Aortic aneurysm; Congenital aneurysm of ascending aorta; Connective tissue disease; Thoracic aortic aneurysm and aortic dissection                                                     |
| THSD4    | 4                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                               |
| ZNF469   | 4                      | 24           | Connective tissue disease; Ehlers-danlos syndrome; Keratoconus; Myopia                                                                                                                 |
| AEBP1    | 3                      | 24           | Congenital contractural arachnodactyly; Ehlers-danlos syndrome; Myopia                                                                                                                 |
| CBS      | 3                      | 24           | Congenital contractural arachnodactyly; Connective tissue disease; Thoracic aortic aneurysm and aortic dissection                                                                      |
| DAB2IP   | 3                      | 24           | Aneurysm; Aortic aneurysm; Keratoconus                                                                                                                                                 |
| EFEMP1   | 3                      | 24           | Congenital contractural arachnodactyly; Connective tissue disease; Myopia                                                                                                              |
| EFEMP2   | 3                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Thoracic aortic aneurysm and aortic dissection                                                                                |
| IPO8     | 3                      | 24           | Congenital contractural arachnodactyly; Loeys-dietz syndrome; Marfan syndrome                                                                                                          |
| KCNH5    | 3                      | 24           | Aneurysm; Aortic aneurysm; neonatal/infantile epilepsy syndrome                                                                                                                        |
| LTBP3    | 3                      | 24           | Congenital contractural arachnodactyly; Marfan syndrome; Mitral valve prolapse                                                                                                         |
| MAT2A    | 3                      | 24           | familial thoracic aortic aneurysm and aortic dissection; Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                                                               |
| MMP2     | 3                      | 24           | Aortic aneurysm; Marfan syndrome; Myopia                                                                                                                                               |
| NCKAIP5  | 3                      | 24           | Aneurysm; Aortic aneurysm; Vitreous body disease                                                                                                                                       |
| NPR3     | 3                      | 24           | Boudin-mortier syndrome; Congenital contractural arachnodactyly; Marfan syndrome                                                                                                       |
| SKI      | 3                      | 24           | Aortic aneurysm; Congenital contractural arachnodactyly; Thoracic aortic aneurysm and aortic dissection                                                                                |
| ABL1     | 2                      | 24           | Congenital contractural arachnodactyly; Connective tissue disease                                                                                                                      |

| Gene    | Member Diseases Linked | Cluster Size | Linked Diseases                                                                     |
|---------|------------------------|--------------|-------------------------------------------------------------------------------------|
| ADAMTS8 | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| AGT     | 2                      | 24           | Aortic aneurysm; Mitral valve prolapse                                              |
| APOE    | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| ARIH1   | 2                      | 24           | Aortic aneurysm; Marfan syndrome                                                    |
| BASP1   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                          |
| CAST    | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| CDH4    | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                          |
| CDKN1A  | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| CDYL    | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                          |
| CELSR2  | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| COL11A1 | 2                      | 24           | Connective tissue disease; Myopia                                                   |
| COL12A1 | 2                      | 24           | Ehlers-danlos syndrome; Keratoconus                                                 |
| COL1A2  | 2                      | 24           | Connective tissue disease; Ehlers-danlos syndrome                                   |
| COL9A1  | 2                      | 24           | Connective tissue disease; Myopia                                                   |
| CSMD1   | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| DOT1L   | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| DSE     | 2                      | 24           | Ehlers-danlos syndrome; ehlers-danlos syndrome, musculocontractural type 2          |
| DUT     | 2                      | 24           | Bone marrow failure and diabetes mellitus syndrome; Myopia                          |
| ELN     | 2                      | 24           | Aortic aneurysm; Thoracic aortic aneurysm and aortic dissection                     |
| EML6    | 2                      | 24           | Keratoconus; Myopia                                                                 |
| FKBP14  | 2                      | 24           | Congenital contractural arachnodactyly; Ehlers-danlos syndrome                      |
| FLII    | 2                      | 24           | cardiomyopathy, dilated, 2j; Myopia                                                 |
| GDF7    | 2                      | 24           | Aneurysm; Aortic aneurysm                                                           |
| GRIA4   | 2                      | 24           | Myopia; neurodevelopmental disorder with or without seizures and gait abnormalities |
| HCN4    | 2                      | 24           | Marfan syndrome; Thoracic aortic aneurysm and aortic dissection                     |
| HDAC4   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                          |

| Gene    | Member Diseases Linked | Cluster Size | Linked Diseases                                                        |
|---------|------------------------|--------------|------------------------------------------------------------------------|
| HEY2    | 2                      | 24           | Marfan syndrome; Thoracic aortic aneurysm and aortic dissection        |
| HGF     | 2                      | 24           | Keratoconus; Myopia                                                    |
| HHEX    | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| HLA-B   | 2                      | 24           | Connective tissue disease; Mitral valve prolapse                       |
| JAG1    | 2                      | 24           | Marfan syndrome; Thoracic aortic aneurysm and aortic dissection        |
| KCNMB2  | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| LPA     | 2                      | 24           | Aneurysm; Aortic aneurysm                                              |
| LPAR3   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| LRP1    | 2                      | 24           | Aneurysm; Aortic aneurysm                                              |
| LRP11   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| LRP1B   | 2                      | 24           | Keratoconus; Myopia                                                    |
| LRP6    | 2                      | 24           | Keratoconus; Myopia                                                    |
| LTBP2   | 2                      | 24           | Marfan syndrome; Mitral valve prolapse                                 |
| MED12   | 2                      | 24           | Ehlers-danlos syndrome; Thoracic aortic aneurysm and aortic dissection |
| MMP13   | 2                      | 24           | Aneurysm; Aortic aneurysm                                              |
| MMP9    | 2                      | 24           | Aortic aneurysm; Marfan syndrome                                       |
| MRC2    | 2                      | 24           | Aneurysm; Aortic aneurysm                                              |
| MYRIP   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| NSDHL   | 2                      | 24           | Connective tissue disease; Mitral valve prolapse                       |
| PBLD    | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| PCLO    | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| PFKP    | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| PLCE1   | 2                      | 24           | Aneurysm; Aortic aneurysm                                              |
| PMEP A1 | 2                      | 24           | Congenital contractural arachnodactyly; Marfan syndrome                |
| PRDM5   | 2                      | 24           | Connective tissue disease; Ehlers-danlos syndrome                      |
| PSRC1   | 2                      | 24           | Aneurysm; Aortic aneurysm                                              |
| PTGIS   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |
| PTPRN2  | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease             |

| Gene    | Member Diseases Linked | Cluster Size | Linked Diseases                                                           |
|---------|------------------------|--------------|---------------------------------------------------------------------------|
| ROBO4   | 2                      | 24           | Aortic aneurysm; Thoracic aortic aneurysm and aortic dissection           |
| SLC35D1 | 2                      | 24           | Connective tissue disease; schneckenbecken dysplasia                      |
| SLC4A10 | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                |
| SMAD4   | 2                      | 24           | Marfan syndrome; Thoracic aortic aneurysm and aortic dissection           |
| SNRPB   | 2                      | 24           | Cerebrocostomandibular syndrome; Mixed connective tissue disease          |
| SNRPC   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                |
| SOD2    | 2                      | 24           | Aortic aneurysm; Marfan syndrome                                          |
| SOX14   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                |
| SPOP    | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                |
| SPSB1   | 2                      | 24           | Aneurysm; Aortic aneurysm                                                 |
| SSX2IP  | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                |
| TGFB R3 | 2                      | 24           | Connective tissue disease; Thoracic aortic aneurysm and aortic dissection |
| THSD1   | 2                      | 24           | Aneurysm; Aortic aneurysm                                                 |
| TRIM55  | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                |
| UIMC1   | 2                      | 24           | Connective tissue disease; Mixed connective tissue disease                |
| WNT1    | 2                      | 24           | Connective tissue disease; Keratoconus                                    |
| ZNF335  | 2                      | 24           | Aneurysm; Aortic aneurysm                                                 |
| ZPR1    | 2                      | 24           | Aneurysm; Aortic aneurysm                                                 |