

GeDiPNet Shared-Gene Disease Pairs

Similarity: Jaccard index on shared curated genes (disease_gdp). Significance: upper-tail hypergeometric test ($P(X \geq k)$), Benjamini-Hochberg FDR-corrected across all tested pairs. Corroborated Shared Genes: count of shared genes backed by ≥ 2 distinct disease_gdp.main_source values combined across both diseases (e.g. GWAS-backed for one, ClinVar-backed for the other still counts); Shared Gene Names lists each gene's own source count in parentheses. Overlap Coefficient: shared genes / MIN(each disease's own total gene count) -- complements the Jaccard-based Similarity Score by showing how much of the SMALLER disease's entire known gene-set is covered by this pair, which Jaccard (dividing by the union) can understate. Shared Cluster: cluster id if both diseases fall in the same label-propagation cluster (scripts/build_disease_clusters.php), blank otherwise. See scripts/build_related.php on gedipnet.bicnirrh.res.in. Data as of: 2026-10-20 03:00:20. Filters applied: disease name contains "Bone fragility" (41 matching pairs). · Rows: 41 · Generated: 2026-10-20 04:43:52

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficie nt	p- value	FDR Q- value	Shared Cluster
Desbuquois syndrome	Doughnut lesion of calvaria and bone fragility syndrome	1	1	SGMS2 (2)	0.00177	1.00000	3.648 e-2	3.803 e-2	
Calvarial doughnut lesions with bone fragility	Desbuquois syndrome	1	1	SGMS2 (5)	0.00177	1.00000	3.648 e-2	3.803 e-2	
Bone fragility with contractures, arterial rupture, and deafness	Larsen-like syndrome, b3gat3 type	1	1	B3GAT3 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Deafness, autosomal dominant, 39, with dentinogenesis imperfecta 1	1	1	DSPP (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Doughnut lesion of calvaria and bone fragility syndrome	Osteoporosis-pseudoglioma syndrome	1	1	SGMS2 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Ehlers-danlos syndrome with short stature and limb anomalies	1	1	B4GALT7 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Ehlers-danlos syndrome, spondylodysplastic type, 1	1	1	B4GALT7 (3)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Increased susceptibility to fractures	1	1	KDEL2 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Joint hypermobility	1	1	KDEL2 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Doughnut lesion of calvaria and bone fragility syndrome	1	1	SGMS2 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Multiple joint dislocations, short stature, and craniofacial dysmorphism with or without congenital heart defects	1	1	B3GAT3 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Osteochondrodysplasia, complex lethal, symoens-barnes-gistelinck type	1	1	TAPT1 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Osteogenesis imperfecta type 3	1	1	SERPINH1 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Preterm premature rupture of the membranes, susceptibility to	1	1	SERPINH1 (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Short stature-optic atrophy-pelger-huc+t anomaly syndrome	1	1	NBAS (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis	1	1	SLC10A7 (3)	0.02000	1.00000	3.164 e-3	4.003 e-3	
Bone fragility with contractures, arterial rupture, and deafness	Short stature, optic nerve atrophy, pelger-huet anomaly and/or infantile liver failure	1	1	NBAS (2)	0.02000	1.00000	3.164 e-3	4.003 e-3	

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Bone fragility with contractures, arterial rupture, and deafness	Spondyloocular syndrome	1	1	XYLT2 (2)	0.02000	1.00000	3.164e-3	4.003e-3	
Bone fragility with contractures, arterial rupture, and deafness	Complex lethal osteochondrodysplasia	1	1	TAPT1 (2)	0.02000	1.00000	3.164e-3	4.003e-3	
Bone fragility with contractures, arterial rupture, and deafness	Chondrodysplasia with multiple dislocations and amelogenesis imperfecta	1	1	SLC10A7 (2)	0.02000	1.00000	3.164e-3	4.003e-3	
Calvarial doughnut lesions with bone fragility	Osteoporosis-pseudoglioma syndrome	1	1	SGMS2 (5)	0.02000	1.00000	3.164e-3	4.003e-3	
Bone fragility with contractures, arterial rupture, and deafness	Calvarial doughnut lesions with bone fragility	1	1	SGMS2 (5)	0.02000	1.00000	3.164e-3	4.003e-3	
Bone fragility	Bone fragility with contractures, arterial rupture, and deafness	1	1	NBAS (2)	0.02000	1.00000	3.164e-3	4.003e-3	
Bone fragility	Pituitary stalk interruption syndrome	1	1	NBAS (2)	0.03030	1.00000	2.066e-3	2.785e-3	
Bone fragility	Male reproductive organ cancer	1	1	NBAS (2)	0.05556	1.00000	1.098e-3	1.651e-3	
Frontonasal dysplasia	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	1	IRX5 (3)	0.12500	1.00000	4.520e-4	8.260e-4	345
Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	Hypochromic microcytic anemia	1	1	IRX5 (3)	0.16667	1.00000	3.228e-4	6.412e-4	
Anodontia	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	1	IRX5 (2)	0.20000	1.00000	2.583e-4	5.384e-4	345
Bone fragility	Short stature, optic nerve atrophy, pelger-huet anomaly and/or infantile liver failure	1	0	NBAS (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Craniofacial dysplasia - osteopenia syndrome	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	1	IRX5 (4)	0.50000	1.00000	6.457e-5	1.985e-4	345
Bone fragility	Short stature-optic atrophy-pelger-huc+t anomaly syndrome	1	0	NBAS (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Calvarial doughnut lesions with bone fragility	Doughnut lesion of calvaria and bone fragility syndrome	1	1	SGMS2 (5)	0.50000	1.00000	6.457e-5	1.985e-4	
Hamamy syndrome	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	0	IRX5 (1)	0.50000	1.00000	6.457e-5	1.985e-4	345
Bone fragility with contractures, arterial rupture, and deafness	Recurrent fractures	2	2	COPB2 (2), WNT11 (2)	0.04000	1.00000	9.806e-6	5.309e-5	
Bone fragility with contractures, arterial rupture, and deafness	Cole-carpenter syndrome	3	3	CRTAP (3), P4HB (7), SEC24D (7)	0.06000	1.00000	2.976e-8	2.220e-7	115
Bone fragility with contractures, arterial rupture, and deafness	Skeletal dysplasia	8	8	GORAB (2), COL1A1 (2), SLC10A7 (2), PLOD2 (2), COL1A2 (2), CHST3 (3), B4GALT7 (2), SERPINH1 (2)	0.09524	0.19048	5.993e-13	6.523e-12	115

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Bone fragility with contractures, arterial rupture, and deafness	Osteoporosis	12	12	GORAB (2), SERPINF1 (2), COL1A1 (4), COL1A2 (3), WNT1 (6), LRP5 (8), COPB2 (6), WNT11 (2), IFITM5 (2), P4HB (2), PLS3 (3), SP7 (2)	0.06030	0.24490	6.966 e-14	7.947 e-13	
Bone fragility with contractures, arterial rupture, and deafness	Larsen syndrome	9	3	GORAB (1), XYLT1 (1), P4HA1 (1), CHST3 (3), XYLT2 (1), B3GALT6 (1), B3GAT3 (5), B4GALT7 (1), PLOD3 (3)	0.15517	0.52941	3.477 e-19	4.942 e-18	115
Bone fragility with contractures, arterial rupture, and deafness	Desbuquois syndrome	38	1	ALPL (1), BMP1 (1), CCDC134 (1), GORAB (1), NBAS (1), SERPINF1 (1), TENT5A (1), XYLT1 (3), COL1A1 (1), SLC10A7 (1), PLOD2 (1), FKBP10 (1)	0.06586	0.77551	1.343 e-45	3.402 e-44	
Bone fragility with contractures, arterial rupture, and deafness	Osteogenesis imperfecta	30	30	ALPL (2), BMP1 (7), CCDC134 (6), SERPINF1 (7), TENT5A (6), COL1A1 (8), PLOD2 (3), FKBP10 (6), COL1A2 (8), CRTAP (7), KDELR2 (6), KIF5B (2)	0.49180	0.73171	3.201 e-71	1.059 e-69	115
Bone fragility with contractures, arterial rupture, and deafness	Osteoporosis-pseudoglioma syndrome	49	2	ALPL (1), BMP1 (1), CCDC134 (1), GORAB (1), NBAS (1), SERPINF1 (1), TENT5A (1), XYLT1 (1), AMBN (1), SEC16B (1), COL1A1 (1), SLC10A7 (1)	0.98000	1.00000	3.216 e-143	1.756 e-141	115