

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  $\geq 2$  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: 2026-10-10 03:00:13. Filters applied: disease name contains "Calvarial doughnut lesions with bone fragility" (4 matching pairs). · Rows: 4 · Generated: 2026-10-10 11:43:56

| Disease A                                                        | Disease B                                               | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|------------------------------------------------------------------|---------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Calvarial doughnut lesions with bone fragility                   | Doughnut lesion of calvaria and bone fragility syndrome | 1            | 1                         | SGMS2 (4)         | 0.50000          | 1.00000             | 6.494e-5 | 2.331e-4    |                |
| Bone fragility with contractures, arterial rupture, and deafness | Calvarial doughnut lesions with bone fragility          | 1            | 1                         | SGMS2 (4)         | 0.02000          | 1.00000             | 3.182e-3 | 4.168e-3    |                |
| Calvarial doughnut lesions with bone fragility                   | Osteoporosis-pseudoglioma syndrome                      | 1            | 1                         | SGMS2 (4)         | 0.02000          | 1.00000             | 3.182e-3 | 4.168e-3    |                |
| Calvarial doughnut lesions with bone fragility                   | Desbuquois syndrome                                     | 1            | 1                         | SGMS2 (4)         | 0.00176          | 1.00000             | 3.682e-2 | 3.867e-2    |                |