

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: 2026-10-11 03:00:14. Filters applied: disease name contains "Anauxetic dysplasia" (5 matching pairs). · Rows: 5 · Generated: 2026-10-11 19:22:00

| Disease A           | Disease B                 | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|---------------------|---------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Anauxetic dysplasia | Cartilage-hair hypoplasia | 1            | 0                         | CCDC107 (1)       | 0.16667          | 0.50000             | 5.195e-4 | 1.061e-3    |                |
| Anauxetic dysplasia | Metaphyseal dysplasia     | 1            | 0                         | CCDC107 (1)       | 0.14286          | 0.33333             | 7.792e-4 | 1.405e-3    |                |
| Anauxetic dysplasia | Prion disease             | 1            | 1                         | POP1 (6)          | 0.04762          | 0.25000             | 4.409e-3 | 5.528e-3    |                |
| Anauxetic dysplasia | Desbuquois syndrome       | 1            | 1                         | POP1 (5)          | 0.00175          | 0.25000             | 1.394e-1 | 1.406e-1    |                |
| Anauxetic dysplasia | Diabetes mellitus type 2  | 1            | 1                         | CCDC107 (2)       | 0.00032          | 0.25000             | 5.905e-1 | 5.906e-1    |                |