

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 "Timothy syndrome" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 22:34:20

| Disease A                                             | Disease B        | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|-------------------------------------------------------|------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Concentric hypertrophic cardiomyopathy                | Timothy syndrome | 1            | 1                         | CACNA1C (6)       | 0.25000          | 1.00000             | 1.948e-4 | 5.354e-4    |                |
| Short qt syndrome                                     | Timothy syndrome | 1            | 1                         | CACNA1C (7)       | 0.08333          | 1.00000             | 7.144e-4 | 1.335e-3    |                |
| Polymorphic catecholaminergic ventricular tachycardia | Timothy syndrome | 1            | 1                         | CACNA1C (6)       | 0.06250          | 1.00000             | 9.742e-4 | 1.665e-3    |                |
| Congenital anomaly of limb                            | Timothy syndrome | 1            | 1                         | CACNA1C (6)       | 0.05882          | 1.00000             | 1.039e-3 | 1.739e-3    |                |
| Restrictive cardiomyopathy                            | Timothy syndrome | 1            | 1                         | CACNA1C (6)       | 0.04167          | 1.00000             | 1.494e-3 | 2.313e-3    |                |