

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: 2026-10-12 03:00:13. Filters applied: disease name contains "Hypotonia-cystinuria syndrome" (5 matching pairs). · Rows: 5 · Generated: 2026-10-12 00:39:12

| Disease A                           | Disease B                     | Shared Genes | Corroborated Shared Genes | Shared Gene Names     | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|-------------------------------------|-------------------------------|--------------|---------------------------|-----------------------|------------------|---------------------|----------|-------------|----------------|
| Hypotonia-cystinuria syndrome       | Mitochondrial myopathy        | 1            | 1                         | CAMKMT (3)            | 0.05556          | 0.33333             | 2.920e-3 | 3.876e-3    |                |
| Hypotonia-cystinuria syndrome       | Motor skills disorder         | 1            | 1                         | CAMKMT (3)            | 0.06250          | 0.33333             | 2.531e-3 | 3.446e-3    |                |
| Developmental coordination disorder | Hypotonia-cystinuria syndrome | 1            | 1                         | CAMKMT (2)            | 0.06667          | 0.33333             | 2.336e-3 | 3.243e-3    |                |
| Cystinuria                          | Hypotonia-cystinuria syndrome | 2            | 2                         | PREPL (2), SLC3A1 (7) | 0.33333          | 0.66667             | 1.518e-7 | 1.030e-6    |                |
| Cystine urolithiasis                | Hypotonia-cystinuria syndrome | 2            | 2                         | PREPL (2), SLC3A1 (2) | 0.40000          | 0.66667             | 7.592e-8 | 5.413e-7    |                |