

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 "Smith-lemli-opitz syndrome" (7 matching pairs). · Rows: 7 · Generated: 2026-10-11 10:42:03

| Disease A                                             | Disease B                                            | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|-------------------------------------------------------|------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Congenital myotonia                                   | Smith-lemli-opitz syndrome                           | 1            | 1                         | CLCN1 (2)         | 0.20000          | 1.00000             | 2.598e-4 | 6.509e-4    |                |
| Myotonia congenita                                    | Smith-lemli-opitz syndrome                           | 1            | 1                         | CLCN1 (3)         | 0.20000          | 1.00000             | 2.598e-4 | 6.509e-4    |                |
| Becker generalized myotonia                           | Smith-lemli-opitz syndrome                           | 1            | 0                         | CLCN1 (1)         | 0.16667          | 0.50000             | 5.195e-4 | 1.061e-3    |                |
| Smith-lemli-opitz syndrome                            | Syndactyly of the toes                               | 1            | 1                         | DHCR7 (7)         | 0.16667          | 0.50000             | 5.195e-4 | 1.061e-3    |                |
| Congenital vertebral-cardiac-renal anomalies syndrome | Smith-lemli-opitz syndrome                           | 1            | 1                         | NADSYN1 (2)       | 0.14286          | 0.33333             | 7.792e-4 | 1.405e-3    |                |
| Hyperkalemic periodic paralysis                       | Smith-lemli-opitz syndrome                           | 1            | 0                         | CLCN1 (1)         | 0.14286          | 0.33333             | 7.792e-4 | 1.405e-3    |                |
| Smith-lemli-opitz syndrome                            | Vertebral, cardiac, renal, and limb defects syndrome | 1            | 1                         | NADSYN1 (4)       | 0.12500          | 0.25000             | 1.039e-3 | 1.739e-3    |                |