

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 "multiple congenital anomalies-hypotonia-seizures syndrome 3" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 15:25:03

| Disease A                                                        | Disease B                                                   | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|------------------------------------------------------------------|-------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Cold paroxysmal hemoglobinuria                                   | multiple congenital anomalies-hypotonia-seizures syndrome 3 | 1            | 1                         | PIGT (2)          | 0.14286          | 1.00000             | 3.897e-4 | 8.665e-4    |                |
| Hemoglobinuria paroxysmal                                        | multiple congenital anomalies-hypotonia-seizures syndrome 3 | 1            | 1                         | PIGT (3)          | 0.14286          | 1.00000             | 3.897e-4 | 8.665e-4    |                |
| multiple congenital anomalies-hypotonia-seizures syndrome 3      | Paroxysmal nocturnal hemoglobinuria                         | 1            | 1                         | PIGT (4)          | 0.14286          | 1.00000             | 3.897e-4 | 8.665e-4    |                |
| Intellectual developmental disorder seizures hypotonia skeletal  | multiple congenital anomalies-hypotonia-seizures syndrome 3 | 1            | 1                         | PIGT (2)          | 0.33333          | 1.00000             | 1.299e-4 | 3.931e-4    |                |
| Intellectual developmental disorder seizures dysmorphic skeletal | multiple congenital anomalies-hypotonia-seizures syndrome 3 | 1            | 1                         | PIGT (2)          | 0.50000          | 1.00000             | 6.494e-5 | 2.331e-4    |                |