

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 "progressive microcephaly-seizures-cortical blindness-developmental delay syndrome" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 21:35:35

| Disease A                                                                         | Disease B                                                                         | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|-----------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| progressive microcephaly-seizures-cortical blindness-developmental delay syndrome | Sensorineural hearing loss thrombocytopenia syndrome                              | 1            | 1                         | DIAPH1 (3)        | 0.50000          | 1.00000             | 6.494e-5 | 2.331e-4    |                |
| Neonatal convulsions                                                              | progressive microcephaly-seizures-cortical blindness-developmental delay syndrome | 1            | 1                         | DIAPH1 (2)        | 0.25000          | 1.00000             | 1.948e-4 | 5.354e-4    |                |
| Pendred syndrome                                                                  | progressive microcephaly-seizures-cortical blindness-developmental delay syndrome | 1            | 1                         | DIAPH1 (2)        | 0.12500          | 1.00000             | 4.546e-4 | 9.726e-4    |                |
| Auditory neuropathy                                                               | progressive microcephaly-seizures-cortical blindness-developmental delay syndrome | 1            | 1                         | DIAPH1 (2)        | 0.03125          | 1.00000             | 2.013e-3 | 2.901e-3    |                |
| Hereditary hearing loss                                                           | progressive microcephaly-seizures-cortical blindness-developmental delay syndrome | 1            | 1                         | DIAPH1 (2)        | 0.01961          | 1.00000             | 3.247e-3 | 4.245e-3    |                |