

## 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: 2026-10-20 03:00:20. Filters applied: disease name contains "Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset" (14 matching pairs). · Rows: 14 · Generated: 2026-10-20 10:51:22

| Disease A                                                                   | Disease B                                                                     | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|-----------------------------------------------------------------------------|-------------------------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Global developmental delay                                                  | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset   | 1            | 1                         | PTRH2 (2)         | 0.00229          | 1.00000             | 2.815e-2 | 2.962e-2    |                |
| Global developmental delay                                                  | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 1 | 1            | 1                         | PTRH2 (2)         | 0.00229          | 1.00000             | 2.815e-2 | 2.962e-2    |                |
| Hearing loss                                                                | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset   | 1            | 1                         | PTRH2 (2)         | 0.00251          | 1.00000             | 2.563e-2 | 2.720e-2    |                |
| Hearing loss                                                                | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 1 | 1            | 1                         | PTRH2 (2)         | 0.00251          | 1.00000             | 2.563e-2 | 2.720e-2    |                |
| Charcot-marie-tooth disease                                                 | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset   | 1            | 1                         | PTRH2 (2)         | 0.00741          | 1.00000             | 8.652e-3 | 9.793e-3    |                |
| Charcot-marie-tooth disease                                                 | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 1 | 1            | 1                         | PTRH2 (2)         | 0.00741          | 1.00000             | 8.652e-3 | 9.793e-3    |                |
| Cerebellar ataxia                                                           | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset   | 1            | 1                         | PTRH2 (2)         | 0.00820          | 1.00000             | 7.813e-3 | 8.931e-3    |                |
| Cerebellar ataxia                                                           | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 1 | 1            | 1                         | PTRH2 (2)         | 0.00820          | 1.00000             | 7.813e-3 | 8.931e-3    |                |
| Anemia                                                                      | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 2 | 1            | 1                         | YARS1 (2)         | 0.01163          | 1.00000             | 5.488e-3 | 6.515e-3    |                |
| Liver failure                                                               | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 2 | 1            | 1                         | YARS1 (2)         | 0.01818          | 1.00000             | 3.487e-3 | 4.343e-3    |                |
| Cholestasis                                                                 | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 2 | 1            | 1                         | YARS1 (2)         | 0.05556          | 1.00000             | 1.098e-3 | 1.651e-3    |                |
| Hyperinsulinemic hypoglycemia                                               | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 2 | 1            | 1                         | YARS1 (2)         | 0.08333          | 1.00000             | 7.102e-4 | 1.174e-3    |                |
| Metachromatic leukodystrophy                                                | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 2 | 1            | 1                         | YARS1 (2)         | 0.11111          | 1.00000             | 5.165e-4 | 9.125e-4    | 214            |
| Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset | Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset 1 | 1            | 0                         | PTRH2 (1)         | 0.50000          | 1.00000             | 6.457e-5 | 1.985e-4    |                |