

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 "microcephalic primordial dwarfism due to RTTN deficiency" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 08:32:06

| Disease A                                                | Disease B                                                | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|----------------------------------------------------------|----------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| microcephalic primordial dwarfism due to RTTN deficiency | Neurodevelopmental disorder                              | 1            | 1                         | RTTN (2)          | 0.00106          | 1.00000             | 6.092e-2 | 6.246e-2    |                |
| microcephalic primordial dwarfism due to RTTN deficiency | Microcephaly                                             | 1            | 1                         | RTTN (4)          | 0.00935          | 1.00000             | 6.884e-3 | 8.163e-3    |                |
| Congenital microcephaly                                  | microcephalic primordial dwarfism due to RTTN deficiency | 1            | 1                         | RTTN (2)          | 0.03125          | 1.00000             | 2.013e-3 | 2.901e-3    |                |
| Laryngeal carcinoma                                      | microcephalic primordial dwarfism due to RTTN deficiency | 1            | 1                         | RTTN (2)          | 0.09091          | 1.00000             | 6.494e-4 | 1.241e-3    |                |
| Bilateral generalized polymicrogyria                     | microcephalic primordial dwarfism due to RTTN deficiency | 1            | 1                         | RTTN (3)          | 0.33333          | 1.00000             | 1.299e-4 | 3.931e-4    |                |