

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 "Developmental delay with impaired growth and dysmorphic facies" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 21:35:34

| Disease A                                                      | Disease B                                                      | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|----------------------------------------------------------------|----------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Charcot-Marie-Tooth disease axonal type 2Z                     | Developmental delay with impaired growth and dysmorphic facies | 1            | 1                         | MORC2 (5)         | 0.20000          | 1.00000             | 2.598e-4 | 6.509e-4    |                |
| Central serous retinopathy                                     | Developmental delay with impaired growth and dysmorphic facies | 1            | 1                         | ARFGEF1 (5)       | 0.06667          | 0.25000             | 2.855e-3 | 3.806e-3    |                |
| Developmental delay with impaired growth and dysmorphic facies | Distal spinal muscular atrophy                                 | 1            | 1                         | MORC2 (4)         | 0.02857          | 0.25000             | 8.029e-3 | 9.332e-3    |                |
| Developmental delay with impaired growth and dysmorphic facies | Distal hereditary motor neuropathy                             | 1            | 1                         | MORC2 (4)         | 0.02500          | 0.25000             | 9.320e-3 | 1.062e-2    |                |
| Developmental delay with impaired growth and dysmorphic facies | Mitral valve prolapse                                          | 1            | 1                         | SPTBN1 (5)        | 0.01961          | 0.25000             | 1.215e-2 | 1.361e-2    |                |