

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 "Intellectual disability with craniofacial dysmorphism and macrocephaly" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 20:16:40

| Disease A                                                               | Disease B                                                              | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|-------------------------------------------------------------------------|------------------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Intellectual disability with craniofacial anomalies and cardiac defects | Intellectual disability with craniofacial dysmorphism and macrocephaly | 1            | 1                         | H1-4 (2)          | 0.33333          | 1.00000             | 1.299e-4 | 3.931e-4    |                |
| Auditory neuropathy                                                     | Intellectual disability with craniofacial dysmorphism and macrocephaly | 1            | 1                         | H1-4 (2)          | 0.03125          | 1.00000             | 2.013e-3 | 2.901e-3    |                |
| Intellectual disability with craniofacial dysmorphism and macrocephaly  | syndromic intellectual disability                                      | 1            | 1                         | H1-4 (2)          | 0.02703          | 1.00000             | 2.338e-3 | 3.243e-3    |                |
| Adenoid cystic carcinoma                                                | Intellectual disability with craniofacial dysmorphism and macrocephaly | 1            | 1                         | H1-4 (2)          | 0.00990          | 1.00000             | 6.494e-3 | 7.751e-3    |                |
| Intellectual disability with craniofacial dysmorphism and macrocephaly  | Neurodevelopmental disorder                                            | 1            | 1                         | H1-4 (2)          | 0.00106          | 1.00000             | 6.092e-2 | 6.246e-2    |                |