

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 "microcephaly 2, primary, autosomal recessive, with or without cortical malformations" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 08:32:25

| Disease A                                                                            | Disease B                                                                            | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|--------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| microcephaly 2, primary, autosomal recessive, with or without cortical malformations | Primary microcephaly                                                                 | 1            | 1                         | WDR62 (4)         | 0.02778          | 1.00000             | 2.273e-3 | 3.191e-3    |                |
| Congenital microcephaly                                                              | microcephaly 2, primary, autosomal recessive, with or without cortical malformations | 1            | 1                         | WDR62 (2)         | 0.03125          | 1.00000             | 2.013e-3 | 2.901e-3    |                |
| Cortical development malformation                                                    | microcephaly 2, primary, autosomal recessive, with or without cortical malformations | 1            | 1                         | WDR62 (3)         | 0.03571          | 1.00000             | 1.753e-3 | 2.609e-3    |                |
| microcephaly 2, primary, autosomal recessive, with or without cortical malformations | Polymicrogyria                                                                       | 1            | 1                         | WDR62 (2)         | 0.03846          | 1.00000             | 1.624e-3 | 2.461e-3    |                |
| microcephaly 2, primary, autosomal recessive, with or without cortical malformations | Skraban-deardorff syndrome                                                           | 1            | 1                         | WDR62 (2)         | 0.33333          | 1.00000             | 1.299e-4 | 3.931e-4    |                |