

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 "Digeorge syndrome" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 15:07:35

| Disease A                                    | Disease B                       | Shared Genes | Corroborated Shared Genes | Shared Gene Names                                                                               | Similarity Score | Overlap Coefficient | P-value   | FDR Q-value | Shared Cluster |
|----------------------------------------------|---------------------------------|--------------|---------------------------|-------------------------------------------------------------------------------------------------|------------------|---------------------|-----------|-------------|----------------|
| 22q11.2 deletion syndrome                    | Digeorge syndrome               | 9            | 9                         | ARVCF (2), COMT (3), GP1BB (2), HIRA (2), JMJD1C (3), RREB1 (3), SEC24C (3), TBX1 (6), UFD1 (2) | 0.45000          | 1.00000             | 6.905e-28 | 1.945e-26   |                |
| Digeorge syndrome                            | Velocardiofacial syndrome       | 5            | 5                         | TBX1 (6), DGCR8 (4), DGCR2 (4), DGCR6 (4), ESS2 (4)                                             | 0.25000          | 1.00000             | 1.613e-15 | 2.379e-14   |                |
| 22q11 deletion syndrome                      | Digeorge syndrome               | 1            | 1                         | FGF8 (2)                                                                                        | 0.05000          | 1.00000             | 1.234e-3  | 1.986e-3    |                |
| Digeorge syndrome                            | Intermittent explosive disorder | 1            | 1                         | COMT (2)                                                                                        | 0.05000          | 1.00000             | 1.234e-3  | 1.986e-3    |                |
| Chromosome 22q11.2 microduplication syndrome | Digeorge syndrome               | 1            | 1                         | TBX1 (5)                                                                                        | 0.05000          | 1.00000             | 1.234e-3  | 1.986e-3    |                |