

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 "SETD2-related neurodevelopmental disorder without or with macrocephaly/overgrowth" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 02:33:40

| Disease A                                                                                        | Disease B                                                                         | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|--------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| SETD2-related microcephaly-severe intellectual disability-multiple congenital anomalies syndrome | SETD2-related neurodevelopmental disorder without or with macrocephaly/overgrowth | 1            | 0                         | SETD2 (1)         | 0.50000          | 1.00000             | 6.494e-5 | 2.331e-4    |                |
| SETD2-related neurodevelopmental disorder without or with macrocephaly/overgrowth                | Sotos syndrome                                                                    | 1            | 1                         | SETD2 (3)         | 0.16667          | 1.00000             | 3.247e-4 | 7.709e-4    |                |
| Dandy-walker syndrome                                                                            | SETD2-related neurodevelopmental disorder without or with macrocephaly/overgrowth | 1            | 1                         | SETD2 (2)         | 0.04762          | 1.00000             | 1.299e-3 | 2.063e-3    |                |
| Rhabdomyosarcoma                                                                                 | SETD2-related neurodevelopmental disorder without or with macrocephaly/overgrowth | 1            | 1                         | SETD2 (2)         | 0.03571          | 1.00000             | 1.753e-3 | 2.609e-3    |                |
| Cerebellar hypoplasia/atrophy, epilepsy, and global developmental delay                          | SETD2-related neurodevelopmental disorder without or with macrocephaly/overgrowth | 1            | 1                         | SETD2 (2)         | 0.02703          | 1.00000             | 2.338e-3 | 3.243e-3    |                |