

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 "mosaic variegated aneuploidy syndrome 1" (5 matching pairs). · Rows: 5 · Generated: 2026-10-10 15:25:03

| Disease A                               | Disease B                               | Shared Genes | Corroborated Shared Genes | Shared Gene Names | Similarity Score | Overlap Coefficient | P-value  | FDR Q-value | Shared Cluster |
|-----------------------------------------|-----------------------------------------|--------------|---------------------------|-------------------|------------------|---------------------|----------|-------------|----------------|
| Microcephaly                            | mosaic variegated aneuploidy syndrome 1 | 1            | 1                         | BUB1B (3)         | 0.00935          | 1.00000             | 6.884e-3 | 8.163e-3    |                |
| Cerebral palsy                          | mosaic variegated aneuploidy syndrome 1 | 1            | 1                         | BUB1B (2)         | 0.02128          | 1.00000             | 2.987e-3 | 3.949e-3    |                |
| Growth disorder                         | mosaic variegated aneuploidy syndrome 1 | 1            | 1                         | BUB1B (2)         | 0.02778          | 1.00000             | 2.273e-3 | 3.191e-3    |                |
| mosaic variegated aneuploidy syndrome 1 | Rhabdomyosarcoma                        | 1            | 1                         | BUB1B (3)         | 0.03571          | 1.00000             | 1.753e-3 | 2.609e-3    |                |
| Mosaic variegated aneuploidy            | mosaic variegated aneuploidy syndrome 1 | 1            | 1                         | BUB1B (7)         | 0.08333          | 1.00000             | 7.144e-4 | 1.335e-3    |                |