

## 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: 2026-10-12 03:00:13. Filters applied: disease name contains "Periventricular heterotopia, x-linked" (7 matching pairs). · Rows: 7 · Generated: 2026-10-11 23:40:42

| Disease A                                                          | Disease B                             | Shared Genes | Corroborated Shared Genes | Shared Gene Names                                                  | Similarity Score | Overlap Coefficient | P-value   | FDR Q-value | Shared Cluster |
|--------------------------------------------------------------------|---------------------------------------|--------------|---------------------------|--------------------------------------------------------------------|------------------|---------------------|-----------|-------------|----------------|
| Periventricular heterotopia, x-linked                              | Periventricular nodular heterotopia   | 6            | 6                         | ARFGEF2 (3), FLNA (7), MAP1B (6), NEDD4L (6), ARF1 (6), ERMARD (8) | 0.42857          | 0.85714             | 3.496e-19 | 6.366e-18   |                |
| hereditary von Willebrand disease                                  | Periventricular heterotopia, x-linked | 1            | 1                         | VWF (2)                                                            | 0.12500          | 1.00000             | 4.546e-4  | 9.726e-4    |                |
| Periventricular heterotopia with microcephaly, autosomal recessive | Periventricular heterotopia, x-linked | 1            | 1                         | ARFGEF2 (4)                                                        | 0.12500          | 1.00000             | 4.546e-4  | 9.726e-4    |                |
| Periventricular heterotopia, x-linked                              | Periventricular laminar heterotopia   | 1            | 0                         | ARFGEF2 (1)                                                        | 0.12500          | 1.00000             | 4.546e-4  | 9.726e-4    |                |
| Congenital idiopathic intestinal pseudoobstruction                 | Periventricular heterotopia, x-linked | 1            | 1                         | FLNA (2)                                                           | 0.12500          | 1.00000             | 4.546e-4  | 9.726e-4    |                |
| Otopalatodigital spectrum disorder                                 | Periventricular heterotopia, x-linked | 1            | 1                         | FLNA (2)                                                           | 0.12500          | 1.00000             | 4.546e-4  | 9.726e-4    |                |
| Periventricular heterotopia, x-linked                              | periventricular nodular heterotopia 7 | 1            | 1                         | NEDD4L (2)                                                         | 0.12500          | 1.00000             | 4.546e-4  | 9.726e-4    |                |