

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 ≥ 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 "Short syndrome" (9 matching pairs). · Rows: 9 · Generated: 2026-10-12 04:46:40

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
PIK3R1-related immunodeficiency and SHORT syndrome	Short syndrome	1	1	PIK3R1 (6)	0.50000	1.00000	6.494e-5	2.331e-4	
Activated pi3k-delta syndrome	Short syndrome	1	1	PIK3R1 (5)	0.25000	1.00000	1.948e-4	5.354e-4	
Activated pi3k-delta syndrome	PIK3R1-related immunodeficiency and SHORT syndrome	1	1	PIK3R1 (2)	0.25000	1.00000	1.948e-4	5.354e-4	
Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi	Short syndrome	1	1	PIK3R1 (5)	0.25000	1.00000	1.948e-4	5.354e-4	
Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi	PIK3R1-related immunodeficiency and SHORT syndrome	1	1	PIK3R1 (2)	0.25000	1.00000	1.948e-4	5.354e-4	
Diffuse idiopathic skeletal hyperostosis	Short syndrome	1	1	PIK3R1 (6)	0.08333	1.00000	7.144e-4	1.335e-3	
Diffuse idiopathic skeletal hyperostosis	PIK3R1-related immunodeficiency and SHORT syndrome	1	1	PIK3R1 (2)	0.08333	1.00000	7.144e-4	1.335e-3	
Agammaglobulinemia	Short syndrome	1	1	PIK3R1 (7)	0.07143	1.00000	8.443e-4	1.499e-3	
Agammaglobulinemia	PIK3R1-related immunodeficiency and SHORT syndrome	1	1	PIK3R1 (4)	0.07143	1.00000	8.443e-4	1.499e-3	