

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: 2026-10-20 03:00:20. Filters applied: disease name contains "Hypotonia, infantile, with psychomotor retardation and characteristic facies" (17 matching pairs). · Rows: 17 · Generated: 2026-10-20 06:17:49

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Autism	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	1	1	UNC80 (4)	0.00068	1.00000	9.504e-2	9.587e-2	
Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	Intellectual developmental disorder	1	1	UNC80 (3)	0.00123	1.00000	5.262e-2	5.418e-2	3
Global developmental delay	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2	1	1	UNC80 (3)	0.00229	1.00000	2.815e-2	2.962e-2	3
Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	Seizures	1	0	TBCK (1)	0.00433	1.00000	1.485e-2	1.628e-2	
Congenital neurologic anomalies	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	1	1	TBCK (2)	0.00862	1.00000	7.425e-3	8.572e-3	
Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	Ventricular septal defect	1	1	TBCK (2)	0.02381	1.00000	2.647e-3	3.413e-3	
Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	Syndromic complex neurodevelopmental disorder	1	1	TBCK (2)	0.05000	1.00000	1.227e-3	1.808e-3	
Distal arthrogryposis	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	1	1	NALCN (3)	0.05556	1.00000	1.098e-3	1.651e-3	
Distal arthrogryposis	Hypotonia, infantile, with psychomotor retardation and characteristic facies	1	1	NALCN (3)	0.05556	1.00000	1.098e-3	1.651e-3	
Digitotalar dysmorphism	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	1	1	NALCN (3)	0.14286	1.00000	3.874e-4	7.329e-4	
Digitotalar dysmorphism	Hypotonia, infantile, with psychomotor retardation and characteristic facies	1	1	NALCN (3)	0.14286	1.00000	3.874e-4	7.329e-4	
Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	Sheldon-hall syndrome	1	1	NALCN (3)	0.16667	1.00000	3.228e-4	6.412e-4	
Hypotonia, infantile, with psychomotor retardation and characteristic facies	Sheldon-hall syndrome	1	1	NALCN (3)	0.16667	1.00000	3.228e-4	6.412e-4	
Congenital contracture of limbs and face, hypotonia, developmental delay syndrome	Hypotonia, infantile, with psychomotor retardation and characteristic facies	1	1	NALCN (6)	0.50000	1.00000	6.457e-5	1.985e-4	
Hypotonia, infantile, with psychomotor retardation and characteristic facies 3	Severe infantile syndromic encephalopathy	1	0	TBCK (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Hypotonia, infantile, with psychomotor retardation and characteristic facies	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	1	0	NALCN (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Congenital contracture of limbs and face, hypotonia, developmental delay syndrome	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1	1	1	NALCN (6)	0.50000	1.00000	6.457e-5	1.985e-4	