

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: 2026-10-07 17:17:47. Filters applied: disease name contains "Brain atrophy" (19 matching pairs). · Rows: 19 · Generated: 2026-10-07 15:32:26

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	Seborrhea-like dermatitis with psoriasiform elements	2	2	TBCD (5), ZNF750 (5)	0.50000	1.00000	2.531e-8	1.932e-7	222
Brain atrophy	Niemann-pick disease	2	1	ACYP1 (1), NPC2 (8)	0.11765	0.25000	1.061e-5	5.252e-5	
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	encephalopathy, progressive, with amyotrophy and optic atrophy	1	1	TBCE (3)	0.25000	1.00000	1.948e-4	5.325e-4	222
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	Sanjad-sakati syndrome	1	1	TBCE (3)	0.25000	1.00000	1.948e-4	5.325e-4	222
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	Kenny caffey syndrome	1	1	TBCE (5)	0.25000	1.00000	1.948e-4	5.325e-4	222
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	Early-onset progressive encephalopathy-spastic ataxia-distal spinal muscular atrophy syndrome	1	1	TBCE (3)	0.25000	1.00000	1.948e-4	5.325e-4	222
Early-onset progressive diffuse brain atrophy-microcephaly-muscle weakness-optic atrophy syndrome	Thiel-behnke corneal dystrophy	1	1	TBCD (5)	0.20000	0.50000	3.896e-4	8.637e-4	
Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	Lipoyltransferase deficiency	1	1	LIPT2 (2)	0.20000	0.50000	3.896e-4	8.637e-4	
Brain atrophy	optic atrophy 9	1	1	ACO2 (2)	0.09091	1.00000	6.494e-4	1.239e-3	
Brain atrophy	Early onset alzheimers disease with behavioral disturbance	1	0	LRRK2 (1)	0.08333	0.50000	1.298e-3	2.060e-3	
Ap4-related intellectual disability and spastic paraplegia	Brain atrophy	1	1	AP4M1 (2)	0.07143	0.25000	2.595e-3	3.512e-3	
Ap-4 deficiency syndrome	Brain atrophy	1	1	AP4M1 (3)	0.07143	0.25000	2.595e-3	3.512e-3	
Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	Ventricular fibrillation	1	0	LIPT2 (1)	0.02632	0.50000	4.671e-3	5.786e-3	
Brugada syndrome	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	1	0	LIPT2 (1)	0.01538	0.50000	8.166e-3	9.478e-3	
Cerebral amyloid angiopathy	Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	1	1	TRAPPC12 (4)	0.01370	0.50000	9.201e-3	1.052e-2	
neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	Non-specific syndromic intellectual disability	1	1	TRAPPC4 (3)	0.00271	1.00000	2.390e-2	2.570e-2	6
Early-onset progressive encephalopathy-hearing loss-pons hypoplasia-brain atrophy syndrome	Mitochondrial disease	1	1	LIPT2 (3)	0.00410	0.50000	3.119e-2	3.309e-2	
Neurodevelopmental disorder	neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	1	1	TRAPPC4 (5)	0.00106	1.00000	6.092e-2	6.245e-2	6

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Neurodevelopmental disorder	neurodevelopmental disorder with microcephaly, epilepsy, and brain atrophy	1	1	TRAPPC6B (4)	0.00106	1.00000	6.092e-2	6.245e-2	6