

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: 2026-10-11 03:00:14. Filters applied: disease name contains "Neural tube defect" (40 matching pairs). · Rows: 40 · Generated: 2026-10-11 01:42:20

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	p-value	FDR Q-value	Shared Cluster
Bifid uvula	Neural tube defect	1	1	GRHL3 (2)	0.02000	0.50000	6.225e-3	7.479e-3	
Bifid uvula	Neural tube defects, x-linked	1	0	GRHL3 (1)	0.03030	0.50000	4.023e-3	5.086e-3	
Cerebral folate deficiency	Neural tube defects, x-linked	1	1	FOLR1 (2)	0.03030	0.50000	4.023e-3	5.086e-3	
Neural tube defect	neurodegenerative syndrome due to cerebral folate transport deficiency	1	1	FOLR1 (3)	0.02041	1.00000	3.117e-3	4.096e-3	
Intellectual developmental disorder dysmorphic hypotonia	Neural tube defect	1	1	KMT5B (2)	0.02041	1.00000	3.117e-3	4.096e-3	
GRHL3-related orofacial clefting	Neural tube defect	1	1	GRHL3 (3)	0.02041	1.00000	3.117e-3	4.096e-3	
frontorhiny	Neural tube defect	1	1	ALX3 (3)	0.02041	1.00000	3.117e-3	4.096e-3	
Neural tube defects, x-linked	neurodegenerative syndrome due to cerebral folate transport deficiency	1	1	FOLR1 (2)	0.03125	1.00000	2.013e-3	2.901e-3	
Homocystinuria	Neural tube defects, x-linked	1	1	CBS (4)	0.03125	1.00000	2.013e-3	2.901e-3	
Combined immunodeficiency-megaloblastic anemia due to methylenetetrahydrofolate dehydrogenase 1 deficiency	Neural tube defects, x-linked	1	1	MTHFD1 (2)	0.03125	1.00000	2.013e-3	2.901e-3	
frontorhiny	Neural tube defects, x-linked	1	1	ALX3 (2)	0.03125	1.00000	2.013e-3	2.901e-3	
GRHL3-related orofacial clefting	Neural tube defects, x-linked	1	1	GRHL3 (2)	0.03125	1.00000	2.013e-3	2.901e-3	
Neural tube defects, susceptibility to	Sacral agenesis	1	1	TBXT (5)	0.11111	0.25000	1.298e-3	2.063e-3	
Aprosencephaly	Neural tube defects, folate-sensitive	1	1	MTRR (2)	0.12500	0.25000	1.039e-3	1.739e-3	121
Neural tube defects, susceptibility to	Sacral defect	1	1	VANGL1 (5)	0.14286	0.50000	6.494e-4	1.241e-3	94
Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	Neural tube defects, folate-sensitive	1	1	MTHFD1 (5)	0.16667	0.50000	5.195e-4	1.061e-3	
Neural tube defects, susceptibility to	Yellow nail syndrome	1	1	CELSR1 (2)	0.16667	1.00000	3.247e-4	7.709e-4	94

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Combined immunodeficiency-megaloblastic anemia due to methylenetetrahydrofolate dehydrogenase 1 deficiency	Neural tube defects, folate-sensitive	1	1	MTHFD1 (2)	0.20000	1.00000	2.598e-4	6.509e-4	
methylcobalamin deficiency type cblE	Neural tube defects, folate-sensitive	1	1	MTRR (2)	0.20000	1.00000	2.598e-4	6.509e-4	121
methylcobalamin deficiency type cblG	Neural tube defects, folate-sensitive	1	1	MTR (2)	0.20000	1.00000	2.598e-4	6.509e-4	121
homocystinuria due to methylene tetrahydrofolate reductase deficiency	Neural tube defects, folate-sensitive	1	1	MTHFR (2)	0.20000	1.00000	2.598e-4	6.509e-4	
Microvascular angina	Neural tube defects, folate-sensitive	1	1	MTHFR (3)	0.20000	1.00000	2.598e-4	6.509e-4	
Neural tube defects, folate-sensitive	Neural tube defects, x-linked	2	2	MTHFR (2), MTHFD1 (2)	0.05882	0.50000	2.348e-5	1.105e-4	
Caudal regression syndrome	Neural tube defects, x-linked	2	2	FUZ (2), VANGL1 (2)	0.06061	0.66667	1.175e-5	5.746e-5	
Malnutrition	Neural tube defects, x-linked	2	2	MTHFR (2), CBS (2)	0.06061	0.66667	1.175e-5	5.746e-5	
Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	Neural tube defect	2	1	ZBTB25 (1), MTHFD1 (5)	0.04082	1.00000	9.516e-6	4.722e-5	
Down syndrome	Neural tube defects, folate-sensitive	2	2	MTHFR (3), MTHFD1 (2)	0.09091	0.50000	8.642e-6	4.300e-5	
Hematologic disease	Neural tube defects, folate-sensitive	2	2	MTHFR (3), MTR (3)	0.09524	0.50000	7.733e-6	3.896e-5	121
Anencephaly	Neural tube defects, folate-sensitive	2	2	MTHFR (2), MTRR (2)	0.16667	0.50000	1.821e-6	1.021e-5	121
Intracellular cobalamin metabolism disorder	Neural tube defects, folate-sensitive	2	0	MTRR (1), MTR (1)	0.16667	0.50000	1.821e-6	1.021e-5	
Homocystinuria with megaloblastic anemia	Neural tube defects, folate-sensitive	2	2	MTRR (4), MTR (2)	0.28571	0.50000	3.036e-7	1.951e-6	121
Caudal regression syndrome	Neural tube defects, susceptibility to	2	2	FUZ (4), VANGL1 (4)	0.28571	0.66667	2.530e-7	1.653e-6	94
Malnutrition	Neural tube defects, folate-sensitive	2	2	MTHFR (3), MTR (3)	0.33333	0.66667	1.518e-7	1.030e-6	121
Hyperhomocysteinemia	Neural tube defect	4	4	MTHFR (2), MTRR (2), CBS (2), MTR (2)	0.06897	0.30769	5.821e-8	4.240e-7	
Malnutrition	Neural tube defect	3	3	MTHFR (2), CBS (2), MTR (2)	0.06122	1.00000	2.843e-8	2.158e-7	
Hyperhomocysteinemia	Neural tube defects, folate-sensitive	3	3	MTHFR (3), MTRR (3), MTR (2)	0.20000	0.75000	1.880e-9	1.650e-8	121
Neural tube defect	Neural tube defects, susceptibility to	4	4	VANGL2 (5), TBXT (3), FUZ (4), VANGL1 (4)	0.08000	0.80000	4.146e-10	3.897e-9	
Neural tube defect	Neural tube defects, folate-sensitive	4	4	MTHFR (3), MTRR (3), MTR (3), MTHFD1 (3)	0.08163	1.00000	8.310e-11	8.352e-10	

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Neural tube defects, susceptibility to	Neural tube defects, x-linked	4	4	VANGL2 (4), TBXT (2), FUZ (3), VANGL1 (3)	0.12121	0.80000	6.710e-11	6.792e-10	
Neural tube defect	Neural tube defects, x-linked	31	29	SKI (2), INS (2), GLI3 (2), CYP1A2 (2), MTHFD1L (1), PAX3 (2), ZIC5 (2), MTHFR (2), PYY (2), CSF2 (2), NPY1R (2), IFNG (2)	0.63265	1.00000	5.554e-84	4.623e-82	