

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 "Multiple malformations" (21 matching pairs). · Rows: 21 · Generated: 2026-10-20 04:56:11

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Multiple malformations of neural tube, ear, genitourinary and gastrointestinal systems	Scoliosis	1	1	DACT1 (2)	0.00096	1.00000	6.708e-2	6.836e-2	
Colorectal neoplasms	Multiple malformations of neural tube, ear, genitourinary and gastrointestinal systems	1	1	DACT1 (2)	0.00347	1.00000	1.853e-2	1.996e-2	
Craniosynostosis	Multiple malformations of neural tube, ear, genitourinary and gastrointestinal systems	1	1	DACT1 (2)	0.01408	1.00000	4.520e-3	5.478e-3	
Multiple malformations of neural tube, ear, genitourinary and gastrointestinal systems	Neural tube defect	1	0	DACT1 (1)	0.01887	1.00000	3.357e-3	4.219e-3	
Hydatidiform mole	Multiple malformations syndrome (bcahh)	1	0	KMT2D (1)	0.08333	1.00000	7.102e-4	1.174e-3	36
Multiple malformations syndrome (bcahh)	Spinal cord ependymoma	1	0	KMT2D (1)	0.12500	1.00000	4.520e-4	8.260e-4	36
Multiple malformations syndrome (bcahh)	Vein of galen aneurysm	1	1	KMT2D (2)	0.14286	1.00000	3.874e-4	7.329e-4	36
Congenital cystic eyeball	Multiple malformations	1	1	GDF3 (2)	0.14286	1.00000	3.874e-4	7.329e-4	
Congenital small ears	Multiple malformations syndrome (bcahh)	1	1	KMT2D (2)	0.16667	1.00000	3.228e-4	6.412e-4	36
Minor salivary gland adenocarcinoma	Multiple malformations syndrome (bcahh)	1	0	KMT2D (1)	0.20000	1.00000	2.583e-4	5.384e-4	36
Camptocormia	Multiple malformations	1	1	GDF3 (3)	0.20000	1.00000	2.583e-4	5.384e-4	
Intraocular medulloepithelioma	Multiple malformations syndrome (bcahh)	1	0	KMT2D (1)	0.20000	1.00000	2.583e-4	5.384e-4	36
Liver cavernous hemangioma	Multiple malformations syndrome (bcahh)	1	0	KMT2D (1)	0.25000	1.00000	1.937e-4	4.414e-4	36
Multiple malformations syndrome (bcahh)	Vissers-bodmer syndrome	1	1	KMT2D (2)	0.25000	1.00000	1.937e-4	4.414e-4	36
Microphthalmia, isolated, with coloboma 7	Multiple malformations	1	0	GDF3 (1)	0.33333	1.00000	1.291e-4	3.269e-4	
Multiple malformations of neural tube, ear, genitourinary and gastrointestinal systems	Townes-brocks syndrome	1	1	DACT1 (7)	0.33333	1.00000	1.291e-4	3.269e-4	
Microphthalmia with coloboma 6	Multiple malformations	1	0	GDF3 (1)	0.33333	1.00000	1.291e-4	3.269e-4	
Branchial arch abnormalities syndrome	Multiple malformations syndrome (bcahh)	1	1	KMT2D (4)	0.50000	1.00000	6.457e-5	1.985e-4	36

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Microphthalmia, isolated, with coloboma 6	Multiple malformations	1	0	GDF3 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Branchial cleft anomalies	Multiple malformations syndrome (bcahh)	1	1	KMT2D (2)	0.50000	1.00000	6.457e-5	1.985e-4	36
Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome	Multiple malformations syndrome (bcahh)	1	0	KMT2D (1)	0.50000	1.00000	6.457e-5	1.985e-4	36