

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 "Hypertelorism" (12 matching pairs). · Rows: 12 · Generated: 2026-10-20 04:10:34

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Craniofacial dysplasia - osteopenia syndrome	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	1	IRX5 (4)	0.50000	1.00000	6.457e-5	1.985e-4	345
Disproportionate tall stature	Hypertelorism	1	0	PDZD8 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Hamamy syndrome	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	0	IRX5 (1)	0.50000	1.00000	6.457e-5	1.985e-4	345
Intellectual developmental disorder with hypertelorism and distinctive facies	Syndromic neurodevelopmental disorder with distinctive facial dysmorphism	1	0	CCNK (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Hypertelorism	Intellectual developmental disorder autism dysmorphic	1	1	PDZD8 (5)	0.33333	1.00000	1.291e-4	3.269e-4	
Anodontia	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	1	IRX5 (2)	0.20000	1.00000	2.583e-4	5.384e-4	345
Intellectual developmental disorder dysmorphic facial	Intellectual developmental disorder with hypertelorism and distinctive facies	1	1	CCNK (5)	0.20000	1.00000	2.583e-4	5.384e-4	
Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	Hypochromic microcytic anemia	1	1	IRX5 (3)	0.16667	1.00000	3.228e-4	6.412e-4	
Abnormality of the face	Hypertelorism	1	0	PDZD8 (1)	0.12500	1.00000	4.520e-4	8.260e-4	
Frontonasal dysplasia	Hypertelorism, severe, with midface prominence, myopia, intellectual developmental disorder, and bone fragility	1	1	IRX5 (3)	0.12500	1.00000	4.520e-4	8.260e-4	345
Autistic behavior	Hypertelorism	1	0	PDZD8 (1)	0.10000	1.00000	5.811e-4	1.000e-3	
Behavioral abnormality	Hypertelorism	1	0	PDZD8 (1)	0.06667	1.00000	9.039e-4	1.411e-3	