

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 "Pena-shokeir syndrome" (11 matching pairs). · Rows: 11 · Generated: 2026-10-11 02:17:23

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Alpha-n-acetylgalactosaminidase deficiency	Pena-shokeir syndrome	1	1	NAGA (4)	0.01887	1.00000	3.377e-3	4.374e-3	
Hypo-akinesia disorder of prenatal onset	Pena-shokeir syndrome	1	1	CHRNA3 (3)	0.01887	1.00000	3.377e-3	4.374e-3	
Mowat-wilson syndrome	Pena-shokeir syndrome	1	1	ZEB2 (7)	0.01887	1.00000	3.377e-3	4.374e-3	
Pena-shokeir syndrome	Schaaf-yang syndrome	1	1	MAGEL2 (5)	0.01887	1.00000	3.377e-3	4.374e-3	
Pena-shokeir syndrome	Schindler disease	1	1	NAGA (3)	0.01887	1.00000	3.377e-3	4.374e-3	
Pena-shokeir syndrome	succinic semialdehyde dehydrogenase deficiency	1	1	ALDH5A1 (2)	0.01887	1.00000	3.377e-3	4.374e-3	
Congenital myasthenic syndrome	Pena-shokeir syndrome	6	6	SCN4A (3), RAPSN (3), CHRND (3), MUSK (4), SLC18A3 (3), DOK7 (5)	0.07895	0.20690	4.930e-10	4.595e-9	
Pena-shokeir syndrome	Postsynaptic congenital myasthenic syndrome	5	5	SCN4A (3), RAPSN (3), CHRND (3), MUSK (4), DOK7 (2)	0.08333	0.41667	2.805e-10	2.672e-9	
Myasthenic syndrome	Pena-shokeir syndrome	8	7	SCN4A (3), RAPSN (3), RYR1 (1), CHRNA3 (2), CHRND (3), MUSK (3), SLC18A3 (3), DOK7 (2)	0.09412	0.20000	6.819e-13	8.338e-12	
Fetal akinesia deformation sequence	Pena-shokeir syndrome	10	10	RAPSN (5), MAGEL2 (2), GLDN (2), KIF21A (2), MYOD1 (3), MUSK (6), TUBA1A (2), SLC18A3 (3), DOK7 (5), NUP88 (5)	0.18182	0.83333	5.046e-24	1.167e-22	
Arthrogryposis multiplex congenita	Pena-shokeir syndrome	44	2	SCN4A (1), GBE1 (1), BLTP1 (1), ACTA1 (1), NAGA (1), ASCC1 (1), PRICKLE1 (1), RAPSN (1), ROR2 (1), SCN8A (1), SPAG16 (1), DYNC1H1 (1)	0.54321	0.84615	8.942e-102	1.046e-99	