

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 "H syndrome" (202 matching pairs). · Rows: 202 · Generated: 2026-10-10 23:44:40

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficie nt	p- value	FDR Q- value	Shared Cluster
Pash syndrome	Schizophrenia	1	1	NCSTN (2)	0.00039	1.00000	1.650 e-1	1.657 e-1	
Alzheimer disease	Pash syndrome	1	1	NCSTN (2)	0.00045	1.00000	1.440 e-1	1.450 e-1	
Barth syndrome	Coronary artery disease	1	1	TAFAZZIN (6)	0.00087	1.00000	7.423 e-2	7.553 e-2	
Deeah syndrome	Kidney disease	1	1	MADD (4)	0.00235	1.00000	2.760 e-2	2.941 e-2	
Deeah syndrome	Migraine	1	1	MADD (4)	0.00247	1.00000	2.624 e-2	2.802 e-2	
Deeah syndrome	Open angle glaucoma	1	1	MADD (4)	0.00270	1.00000	2.396 e-2	2.575 e-2	240
Global developmental delay	marshall-smith syndrome	1	1	NFIX (2)	0.00270	1.00000	2.403 e-2	2.575 e-2	
Deeah syndrome	Non-specific syndromic intellectual disability	1	1	MADD (4)	0.00271	1.00000	2.390 e-2	2.570 e-2	
Leigh syndrome	Lipoyltransferase deficiency	1	1	LIPT1 (7)	0.00909	0.33333	2.070 e-2	2.250 e-2	
Barth syndrome	Dilated cardiomyopathy	1	1	TAFAZZIN (6)	0.00365	1.00000	1.773 e-2	1.940 e-2	
Barth syndrome	Cardiomyopathy	1	1	TAFAZZIN (5)	0.00408	1.00000	1.585 e-2	1.746 e-2	
Leigh syndrome	Sandhoff disease	1	1	GFM2 (2)	0.00917	0.50000	1.385 e-2	1.542 e-2	50
Cardioembolic stroke	Wiskott-aldrich syndrome	1	1	WIPF1 (6)	0.00971	0.50000	1.308 e-2	1.460 e-2	
Atrial flutter	Wiskott-aldrich syndrome	1	1	WIPF1 (6)	0.01205	0.50000	1.049 e-2	1.186 e-2	
Hodgkin lymphoma	marshall-smith syndrome	1	1	NFIX (2)	0.00893	1.00000	7.209 e-3	8.515 e-3	
Proliferative diabetic retinopathy	Pseudo-torch syndrome	1	1	STAT2 (5)	0.02564	0.33333	6.998 e-3	8.284 e-3	
3-hydroxyisobutyryl-coa hydrolase deficiency	Leigh syndrome	1	1	HIBCH (3)	0.00926	1.00000	6.949 e-3	8.229 e-3	

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficient	p- value	FDR Q- value	Shared Cluster
Aminoglycoside-induced deafness	Leigh syndrome	1	1	TRMU (3)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Amish lethal microcephaly	Leigh syndrome	1	1	SLC25A19 (6)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Biotin-thiamine-responsive basal ganglia disease	Leigh syndrome	1	1	SLC19A3 (5)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Biotinidase deficiency	Leigh syndrome	1	1	BTD (8)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Bjornstad syndrome	Leigh syndrome	1	1	BCS1L (6)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Cataract-growth hormone deficiency-skeletal dysplasia syndrome	Leigh syndrome	1	1	IARS2 (6)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Charcot-Marie-Tooth disease axonal type 2Z	Leigh syndrome	1	1	MORC2 (2)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Childhood-onset dystonia	Leigh syndrome	1	1	MECR (2)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Childhood-onset dystonia with optic atrophy and basal ganglia abnormalities	Leigh syndrome	1	1	MECR (5)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Codas syndrome	Leigh syndrome	1	1	LONP1 (7)	0.00926	1.00000	6.949 e-3	8.229 e-3	50
Hepatoencephalopathy due to combined oxidative phosphorylation defect	Leigh syndrome	1	1	GFM1 (5)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Hurthle cell thyroid cancer	Leigh syndrome	1	1	NDUFA13 (4)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Leigh syndrome	mitochondrial short-chain enoyl-coa hydratase 1 deficiency	1	1	ECHS1 (4)	0.00926	1.00000	6.949 e-3	8.229 e-3	50
Leigh syndrome	optic atrophy 13 with retinal and foveal abnormalities	1	1	SSBP1 (2)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Leigh syndrome	pyruvate dehydrogenase E3 deficiency	1	1	DLD (2)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Leigh syndrome	Thiamine-responsive encephalopathy	1	1	SLC19A3 (4)	0.00926	1.00000	6.949 e-3	8.229 e-3	
Congenital epithelial dysplasia of intestine	Lynch syndrome	1	1	EPCAM (8)	0.01961	0.50000	6.355 e-3	7.624 e-3	
Cystic leukoencephalopathy	Lynch syndrome	1	1	RNASET2 (4)	0.01961	0.50000	6.355 e-3	7.624 e-3	166
Interstitial nephritis	Lynch syndrome	1	1	FAN1 (5)	0.01961	0.50000	6.355 e-3	7.624 e-3	
Lynch syndrome	Sveinsson chorioretinal atrophy	1	1	SEMA4A (2)	0.01961	0.50000	6.355 e-3	7.624 e-3	
Congenital microcephaly	Pseudo-torch syndrome	1	1	OCLN (6)	0.02941	0.33333	6.028 e-3	7.260 e-3	

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficie nt	p- value	FDR Q- value	Shared Cluster
Chagas cardiomyopathy	Chudley-mccullough syndrome	1	1	SPTB (2)	0.03571	0.33333	4.863 e-3	5.987 e-3	400
Corneal astigmatism	Deeah syndrome	1	1	MADD (4)	0.01389	1.00000	4.611 e-3	5.734 e-3	240
Congenital heart defects	H syndrome	1	1	SLC29A3 (2)	0.01408	1.00000	4.546 e-3	5.668 e-3	
Barth syndrome	Left ventricular disease	1	1	TAFAZZIN (5)	0.01493	1.00000	4.286 e-3	5.383 e-3	4
Pseudo-torch syndrome	Splenomegaly	1	1	OCLN (6)	0.04348	0.33333	3.892 e-3	4.941 e-3	
H syndrome	Hepatomegaly	1	1	SLC29A3 (2)	0.01695	1.00000	3.767 e-3	4.809 e-3	
Movement disorder	Wiedemann-rautenstrauch syndrome	1	1	POLR3A (3)	0.01724	1.00000	3.702 e-3	4.735 e-3	
Colorectal adenomatous polyposis	Lynch syndrome	1	1	MUTYH (2)	0.02000	1.00000	3.182 e-3	4.168 e-3	
colorectal cancer, hereditary nonpolyposis, type 7	Lynch syndrome	1	1	MLH3 (6)	0.02000	1.00000	3.182 e-3	4.168 e-3	
Congenital tufting enteropathy	Lynch syndrome	1	1	EPCAM (8)	0.02000	1.00000	3.182 e-3	4.168 e-3	
karyomegalic interstitial nephritis	Lynch syndrome	1	1	FAN1 (3)	0.02000	1.00000	3.182 e-3	4.168 e-3	
Lynch syndrome	RAD51D-related cancer predisposition	1	1	RAD51D (2)	0.02000	1.00000	3.182 e-3	4.168 e-3	
Lynch syndrome	thrombocytopenia 10	1	1	PTPRJ (2)	0.02000	1.00000	3.182 e-3	4.168 e-3	
Knobloch syndrome	Megaloblastic anemia	1	1	SLC19A1 (4)	0.06250	0.25000	3.114 e-3	4.096 e-3	
Knobloch syndrome	Transposition of the great arteries	1	0	SLC19A1 (1)	0.06667	0.25000	2.855 e-3	3.806 e-3	113
Severe congenital neutropenia	Wiskott-aldrich syndrome	1	1	WAS (7)	0.04167	0.50000	2.856 e-3	3.806 e-3	
Barth syndrome	Left ventricular noncompaction cardiomyopathy	1	1	TAFAZZIN (5)	0.02564	1.00000	2.468 e-3	3.371 e-3	4
marshall-smith syndrome	Strabismus	1	1	NFIX (2)	0.02857	1.00000	2.208 e-3	3.113 e-3	
Chromodomain helicase dna binding protein 8 overgrowth syndrome	Gastrointestinal disease	1	1	CHD8 (2)	0.03030	1.00000	2.078 e-3	2.969 e-3	
Chudley-mccullough syndrome	Spherocytosis	1	1	SPTB (3)	0.07692	0.33333	1.947 e-3	2.828 e-3	
Hypomyelinating leukodystrophy	Wiedemann-rautenstrauch syndrome	1	1	POLR3A (4)	0.03226	1.00000	1.948 e-3	2.828 e-3	

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	p- value	FDR Q- value	Shared Cluster
Chromodomain helicase dna binding protein 8 overgrowth syndrome	Congenital myasthenic syndrome	1	1	CHD8 (2)	0.03333	1.00000	1.883 e-3	2.762 e-3	
overgrowth syndrome and/or cerebral malformations due to abnormalities in MTOR pathway genes	Small cell carcinoma	1	1	MTOR (2)	0.08333	0.33333	1.753 e-3	2.609 e-3	
Acro-dermo-ungual-lacrima-tooth syndrome	Bladder exstrophy and epispadias complex	1	1	TP63 (2)	0.03571	1.00000	1.753 e-3	2.609 e-3	27
Leukodystrophy	Wiedemann-rautenstrauch syndrome	1	1	POLR3A (3)	0.03704	1.00000	1.689 e-3	2.531 e-3	
Brown tendon sheath syndrome	Torsion dystonia	1	1	TUBB4A (3)	0.10000	0.33333	1.363 e-3	2.148 e-3	
Chudley-mccullough syndrome	Hereditary elliptocytosis	1	1	SPTB (4)	0.10000	0.33333	1.363 e-3	2.148 e-3	400
Focal cortical dysplasia	overgrowth syndrome and/or cerebral malformations due to abnormalities in MTOR pathway genes	1	1	MTOR (4)	0.10000	0.33333	1.363 e-3	2.148 e-3	
Hepatic veno occlusive disease	Pseudo-torch syndrome	1	1	OCLN (6)	0.10000	0.33333	1.363 e-3	2.148 e-3	
Leiomyosarcoma	Pseudo-torch syndrome	1	1	USP18 (6)	0.10000	0.33333	1.363 e-3	2.148 e-3	213
Acro-dermo-ungual-lacrima-tooth syndrome	Bladder exstrophy	1	1	TP63 (2)	0.04545	1.00000	1.364 e-3	2.148 e-3	27
Aprosencephaly	Coach syndrome	1	1	RPGRIP1L (3)	0.11111	0.25000	1.298 e-3	2.063 e-3	
Coach syndrome	Encephalocele	1	1	CC2D2A (3)	0.11111	0.25000	1.298 e-3	2.063 e-3	
Dyslipidemias	H syndrome	1	1	SLC29A3 (4)	0.05263	1.00000	1.169 e-3	1.895 e-3	
Clear cell papillary renal cell carcinoma	Waardenburg-shah syndrome	1	1	MITF (2)	0.12500	0.25000	1.039 e-3	1.739 e-3	
Discordant ventriculoarterial connection	Knobloch syndrome	1	0	SLC19A1 (1)	0.12500	0.25000	1.039 e-3	1.739 e-3	113
Bonnevie-ullrich syndrome	Congenital isolated growth hormone deficiency	1	0	GH1 (1)	0.11111	0.50000	9.090 e-4	1.576 e-3	
Acro-dermo-ungual-lacrima-tooth syndrome	Omphalocele exstrophy imperforate anus	1	1	TP63 (2)	0.07143	1.00000	8.443 e-4	1.499 e-3	27
Brown tendon sheath syndrome	Parathyroid disease	1	1	MAFB (2)	0.14286	0.33333	7.792 e-4	1.405 e-3	
Aortic arch syndrome	Parapsoriasis	1	1	HLA-A (2)	0.12500	0.50000	7.792 e-4	1.405 e-3	1
Aortic arch syndrome	Vaginal neoplasms	1	1	IL2 (2)	0.12500	0.50000	7.792 e-4	1.405 e-3	

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficient	p- value	FDR Q- value	Shared Cluster
Specific learning disability	Tatton-Brown-Rahman overgrowth syndrome	1	1	DNMT3A (2)	0.07692	1.00000	7.793 e-4	1.405 e-3	268
H syndrome	Hypertrichosis	1	1	SLC29A3 (3)	0.08333	1.00000	7.144 e-4	1.335 e-3	
Boichis syndrome	Coach syndrome	1	1	TMEM67 (5)	0.14286	0.50000	6.494 e-4	1.241 e-3	306
Cerebellar malformation	Coach syndrome	1	1	TMEM67 (5)	0.14286	0.50000	6.494 e-4	1.241 e-3	306
Coach syndrome	Senior-boichis syndrome	1	1	TMEM67 (6)	0.14286	0.50000	6.494 e-4	1.241 e-3	306
Acro-dermo-ungual-lacrima-tooth syndrome	Congenital skin anomaly	1	1	TP63 (2)	0.09091	1.00000	6.494 e-4	1.241 e-3	
Denys drash syndrome	Peritoneal neoplasms	1	1	WT1 (6)	0.09091	1.00000	6.494 e-4	1.241 e-3	35
Drash syndrome	Peritoneal neoplasms	1	1	WT1 (2)	0.09091	1.00000	6.494 e-4	1.241 e-3	35
Acro-dermo-ungual-lacrima-tooth syndrome	Skin abnormalities	1	1	TP63 (2)	0.09091	1.00000	6.494 e-4	1.241 e-3	
Dwarfism	Tatton-Brown-Rahman overgrowth syndrome	1	1	DNMT3A (3)	0.09091	1.00000	6.494 e-4	1.241 e-3	
Polycythemia vera	Tatton-Brown-Rahman overgrowth syndrome	1	1	DNMT3A (2)	0.10000	1.00000	5.845 e-4	1.156 e-3	
Digestive system neoplasms	H syndrome	1	1	SLC29A3 (2)	0.11111	1.00000	5.195 e-4	1.061 e-3	
Acro-dermo-ungual-lacrima-tooth syndrome	Split hand-foot malformation	1	1	TP63 (5)	0.11111	1.00000	5.195 e-4	1.061 e-3	
Acatalasia	Bonnevie-ullrich syndrome	1	1	CAT (3)	0.12500	1.00000	4.546 e-4	9.726 e-4	285
amyotrophic lateral sclerosis type 1	Bonnevie-ullrich syndrome	1	1	SOD1 (2)	0.12500	1.00000	4.546 e-4	9.726 e-4	285
Ateleiotic dwarfism	Bonnevie-ullrich syndrome	1	1	GH1 (2)	0.12500	1.00000	4.546 e-4	9.726 e-4	
Bonnevie-ullrich syndrome	Esophageal stenosis	1	1	SOD2 (2)	0.12500	1.00000	4.546 e-4	9.726 e-4	
Acro-dermo-ungual-lacrima-tooth syndrome	Breast disease	1	1	TP63 (3)	0.12500	1.00000	4.546 e-4	9.726 e-4	27
Bonnevie-ullrich syndrome	Isolated somatotropin deficiency	1	1	GH1 (2)	0.12500	1.00000	4.546 e-4	9.726 e-4	
Bonnevie-ullrich syndrome	Peptic esophagitis	1	0	VDR (1)	0.12500	1.00000	4.546 e-4	9.726 e-4	

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Cebalid syndrome	overgrowth syndrome and/or cerebral malformations due to abnormalities in MTOR pathway genes	1	1	MTOR (2)	0.20000	0.50000	3.896 e-4	8.665 e-4	
Aortic arch syndrome	Autoimmune pulmonary alveolar proteinosis	1	1	HLA-DRB1 (2)	0.14286	1.00000	3.897 e-4	8.665 e-4	1
Aortic arch syndrome	Heerfordt syndrome	1	0	HLA-DRB1 (1)	0.14286	1.00000	3.897 e-4	8.665 e-4	1
Aortic arch syndrome	Carbamazepine hypersensitivity	1	1	HLA-B (2)	0.14286	1.00000	3.897 e-4	8.665 e-4	
Aortic arch syndrome	Middle ear cholesteatoma	1	0	IL2 (1)	0.14286	1.00000	3.897 e-4	8.665 e-4	
Aortic arch syndrome	Thrombophlebitis	1	0	HLA-B (1)	0.14286	1.00000	3.897 e-4	8.665 e-4	
Ambiguous genitalia	Denys drash syndrome	1	1	WT1 (6)	0.16667	1.00000	3.247 e-4	7.709 e-4	35
Ambiguous genitalia	Drash syndrome	1	1	WT1 (2)	0.16667	1.00000	3.247 e-4	7.709 e-4	35
Bifid nail	Coach syndrome	1	0	OFD1 (1)	0.16667	1.00000	3.247 e-4	7.709 e-4	
Chromodomain helicase dna binding protein 8 overgrowth syndrome	Congenital ptosis	1	0	CHD8 (1)	0.16667	1.00000	3.247 e-4	7.709 e-4	279
Coach syndrome	Rhyns syndrome	1	1	TMEM67 (6)	0.16667	1.00000	3.247 e-4	7.709 e-4	306
Coach syndrome	joubert syndrome 1	1	1	INPP5E (2)	0.16667	1.00000	3.247 e-4	7.709 e-4	
Coach syndrome	MORM syndrome	1	1	INPP5E (2)	0.16667	1.00000	3.247 e-4	7.709 e-4	
Coach syndrome	OFD1-related ciliopathy	1	1	OFD1 (2)	0.16667	1.00000	3.247 e-4	7.709 e-4	
Congenital or early infantile cach syndrome	leukoencephalopathy with vanishing white matter 3	1	1	EIF2B3 (3)	0.16667	1.00000	3.247 e-4	7.709 e-4	170
Congenital or early infantile cach syndrome	leukoencephalopathy with vanishing white matter 1	1	1	EIF2B1 (3)	0.16667	1.00000	3.247 e-4	7.709 e-4	170
Congenital or early infantile cach syndrome	leukoencephalopathy with vanishing white matter 2	1	1	EIF2B2 (3)	0.16667	1.00000	3.247 e-4	7.709 e-4	170
Congenital or early infantile cach syndrome	leukoencephalopathy with vanishing white matter 4	1	1	EIF2B4 (3)	0.16667	1.00000	3.247 e-4	7.709 e-4	170
Congenital or early infantile cach syndrome	leukoencephalopathy with vanishing white matter 5	1	1	EIF2B5 (3)	0.16667	1.00000	3.247 e-4	7.709 e-4	170
Abcd syndrome	Waardenburg-shah syndrome	1	1	EDNRB (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficient	p- value	FDR Q- value	Shared Cluster
Acro-dermo-ungual-lacrimal-tooth syndrome	Cleft lip and cleft of alveolar process of maxilla	1	1	TP63 (2)	0.20000	1.00000	2.598 e-4	6.509 e-4	27
Bone mineral density quantitative trait locus	Worth syndrome	1	1	LRP5 (4)	0.20000	1.00000	2.598 e-4	6.509 e-4	98
Acro-dermo-ungual-lacrimal-tooth syndrome	Congenital foot deformity	1	1	TP63 (2)	0.20000	1.00000	2.598 e-4	6.509 e-4	27
Deaf blind hypopigmentation syndrome	Waardenburg-shah syndrome	1	1	SOX10 (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	
immunodeficiency 114, folate-responsive	Knobloch syndrome	1	1	SLC19A1 (2)	0.20000	1.00000	2.598 e-4	6.509 e-4	113
Intestinal aganglionosis	Waardenburg-shah syndrome	1	1	EDNRB (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	
Knobloch syndrome	microcornea-myopic chorioretinal atrophy	1	1	ADAMTS18 (2)	0.20000	1.00000	2.598 e-4	6.509 e-4	113
Acro-dermo-ungual-lacrimal-tooth syndrome	Post-operative stroke	1	1	TP63 (2)	0.20000	1.00000	2.598 e-4	6.509 e-4	27
Waardenburg syndrome type 2	Waardenburg-shah syndrome	1	1	MITF (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	229
Waardenburg syndrome type 4A	Waardenburg-shah syndrome	1	1	EDNRB (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	
Waardenburg syndrome type 4B	Waardenburg-shah syndrome	1	1	EDN3 (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	229
Waardenburg syndrome type 4C	Waardenburg-shah syndrome	1	1	SOX10 (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	
Denys drash syndrome	Wagr syndrome	1	1	WT1 (6)	0.20000	1.00000	2.598 e-4	6.509 e-4	35
Drash syndrome	Wagr syndrome	1	1	WT1 (4)	0.20000	1.00000	2.598 e-4	6.509 e-4	35
Waardenburg-shah syndrome	Yemenite deaf-blind hypopigmentation syndrome	1	1	SOX10 (3)	0.20000	1.00000	2.598 e-4	6.509 e-4	
14q11.2 microduplication syndrome	Chromodomain helicase dna binding protein 8 overgrowth syndrome	1	1	CHD8 (2)	0.25000	1.00000	1.948 e-4	5.354 e-4	279
Acne inversa	Pash syndrome	1	1	NCSTN (2)	0.25000	1.00000	1.948 e-4	5.354 e-4	116
Brown tendon sheath syndrome	Whispering dysphonia	1	1	TUBB4A (2)	0.25000	1.00000	1.948 e-4	5.354 e-4	
Brown tendon sheath syndrome	TUBB4A-related neurologic disorder	1	1	TUBB4A (2)	0.25000	1.00000	1.948 e-4	5.354 e-4	
Brown tendon sheath syndrome	Osteolysis, hereditary, of carpal bones with or without nephropathy	1	0	MAFB (1)	0.25000	1.00000	1.948 e-4	5.354 e-4	
Chudley-mccullough syndrome	Neonatal anemia	1	1	SPTB (2)	0.25000	1.00000	1.948 e-4	5.354 e-4	400

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Chudley-mccullough syndrome	Perinatal hemolytic anemia	1	0	SPTB (1)	0.25000	1.00000	1.948 e-4	5.354 e-4	400
Clonal cytopenia of undetermined significance	Tatton-Brown-Rahman overgrowth syndrome	1	1	DNMT3A (2)	0.25000	1.00000	1.948 e-4	5.354 e-4	268
Vitamin d deficiency	Worth syndrome	1	1	LRP5 (3)	0.25000	1.00000	1.948 e-4	5.354 e-4	98
Camurati-engelmann syndrome	Worth syndrome	1	1	LRP5 (3)	0.33333	1.00000	1.299 e-4	3.931 e-4	98
Chromodomain helicase dna binding protein 8 overgrowth syndrome	Intellectual developmental disorder autism dysmorphic	1	1	CHD8 (4)	0.33333	1.00000	1.299 e-4	3.931 e-4	279
Congenital aniridia	Denys drash syndrome	1	1	WT1 (7)	0.33333	1.00000	1.299 e-4	3.931 e-4	35
Congenital aniridia	Drash syndrome	1	0	WT1 (1)	0.33333	1.00000	1.299 e-4	3.931 e-4	35
Denys drash syndrome	Desmoplastic small round cell tumor	1	1	WT1 (6)	0.33333	1.00000	1.299 e-4	3.931 e-4	35
Desmoplastic small round cell tumor	Drash syndrome	1	1	WT1 (2)	0.33333	1.00000	1.299 e-4	3.931 e-4	35
Osteopetrosis and infantile neuroaxonal dystrophy	Worth syndrome	1	1	LRP5 (3)	0.33333	1.00000	1.299 e-4	3.931 e-4	98
Retinopathy background	Worth syndrome	1	1	LRP5 (3)	0.33333	1.00000	1.299 e-4	3.931 e-4	98
Tremor-ataxia-central hypomyelination syndrome	Wiedemann-rautenstrauch syndrome	1	1	POLR3A (3)	0.33333	1.00000	1.299 e-4	3.931 e-4	
Van buchem disease	Worth syndrome	1	1	LRP5 (3)	0.33333	1.00000	1.299 e-4	3.931 e-4	98
Wiskott-aldrich syndrome	X-linked severe congenital neutropenia	1	1	WAS (7)	0.33333	1.00000	1.299 e-4	3.931 e-4	
Wiskott-aldrich syndrome	X-linked thrombocytopenia	1	1	WAS (7)	0.33333	1.00000	1.299 e-4	3.931 e-4	
11p partial monosomy syndrome	Denys drash syndrome	1	1	WT1 (7)	0.50000	1.00000	6.494 e-5	2.331 e-4	35
11p partial monosomy syndrome	Drash syndrome	1	0	WT1 (1)	0.50000	1.00000	6.494 e-5	2.331 e-4	35
19p13.3 microduplication syndrome	marshall-smith syndrome	1	1	NFIX (3)	0.50000	1.00000	6.494 e-5	2.331 e-4	
Acro-dermo-ungual-lacrima-tooth syndrome	Ankyloblepharon-ectodermal defects-cleft lip/palate	1	1	TP63 (5)	0.50000	1.00000	6.494 e-5	2.331 e-4	27
Acro-dermo-ungual-lacrima-tooth syndrome	Limb-mammary syndrome	1	1	TP63 (5)	0.50000	1.00000	6.494 e-5	2.331 e-4	27
Acro-dermo-ungual-lacrima-tooth syndrome	Rudiger syndrome	1	1	TP63 (2)	0.50000	1.00000	6.494 e-5	2.331 e-4	27

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficient	p- value	FDR Q- value	Shared Cluster
Acro-dermo-ungual-lacrima-tooth syndrome	Sweat gland neoplasm	1	1	TP63 (2)	0.50000	1.00000	6.494 e-5	2.331 e-4	27
Denys drash syndrome	Drash syndrome	1	1	WT1 (7)	0.50000	1.00000	6.494 e-5	2.331 e-4	35
Denys drash syndrome	wilms tumor 1	1	1	WT1 (6)	0.50000	1.00000	6.494 e-5	2.331 e-4	35
Drash syndrome	wilms tumor 1	1	1	WT1 (2)	0.50000	1.00000	6.494 e-5	2.331 e-4	35
heyn-sproul-jackson syndrome	Tatton-Brown-Rahman overgrowth syndrome	1	0	DNMT3A (1)	0.50000	1.00000	6.494 e-5	2.331 e-4	268
LRP5-related exudative vitreoretinopathy	Worth syndrome	1	1	LRP5 (4)	0.50000	1.00000	6.494 e-5	2.331 e-4	98
Malan syndrome	marshall-smith syndrome	1	1	NFIX (6)	0.50000	1.00000	6.494 e-5	2.331 e-4	
Microcephalic dwarfism	Tatton-Brown-Rahman overgrowth syndrome	1	1	DNMT3A (2)	0.50000	1.00000	6.494 e-5	2.331 e-4	268
POLR3A-related disorder	Wiedemann-rautenstrauch syndrome	1	1	POLR3A (4)	0.50000	1.00000	6.494 e-5	2.331 e-4	
polycystic liver disease 4 with or without kidney cysts	Worth syndrome	1	1	LRP5 (4)	0.50000	1.00000	6.494 e-5	2.331 e-4	98
Encephalopathy due to mitochondrial and peroxisomal fission defect	Leigh syndrome	2	2	DNM1L (3), MFF (2)	0.01852	1.00000	4.784 e-5	2.166 e-4	
Leigh syndrome	Thiamine metabolism dysfunction syndrome	2	2	SLC25A19 (4), TPK1 (4)	0.01852	1.00000	4.784 e-5	2.166 e-4	
Aortic arch syndrome	Retinopathy of prematurity	2	0	HLA-DRB1 (1), HLA-B (1)	0.08000	0.33333	2.397 e-5	1.127 e-4	
Bonnevie-ullrich syndrome	Hyperemia	2	2	CAT (2), NOS2 (2)	0.10000	0.28571	1.608 e-5	7.714 e-5	
Cortical development malformation	overgrowth syndrome and/or cerebral malformations due to abnormalities in MTOR pathway genes	2	2	AKT3 (2), MTOR (2)	0.06897	0.66667	8.873 e-6	4.408 e-5	
Aortic arch syndrome	Pemphigus	2	1	HLA-DRB1 (2), IL2 (1)	0.15385	0.33333	3.539 e-6	1.886 e-5	1
Anencephaly	Coach syndrome	2	2	CC2D2A (3), RPEGIP1L (3)	0.15385	0.40000	3.034 e-6	1.631 e-5	
Aortic arch syndrome	Birdshot chorioretinopathy	2	1	HLA-B (1), HLA-A (2)	0.20000	0.40000	1.265 e-6	7.290 e-6	
Bonnevie-ullrich syndrome	Ureteral calculi	2	2	CAT (2), SOD1 (2)	0.20000	0.50000	1.062 e-6	6.210 e-6	285
Hemimegalencephaly	overgrowth syndrome and/or cerebral malformations due to abnormalities in MTOR pathway genes	2	2	AKT3 (3), MTOR (3)	0.22222	0.66667	5.313 e-7	3.272 e-6	

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similari ty Score	Overlap Coefficie nt	p- value	FDR Q- value	Shared Cluster
Hirschsprung disease	Waardenburg-shah syndrome	3	3	EDNRB (6), EDN3 (6), SOX10 (3)	0.04348	0.75000	3.140 e-7	2.015 e-6	
Aortic arch syndrome	Dermatitis	3	3	HLA-DRB1 (2), HLA-B (2), HLA-A (2)	0.07143	0.50000	2.759 e-7	1.795 e-6	
Aortic arch syndrome	Thromboangiitis obliterans	2	0	HLA-DRB1 (1), HLA-A (1)	0.28571	1.00000	1.265 e-7	8.712 e-7	1
Aortic disease	Bonnevie-ullrich syndrome	3	3	CAT (2), SOD2 (2), SOD1 (2)	0.12500	0.42857	5.558 e-8	4.053 e-7	285
Coach syndrome	Cystic kidney disease	3	3	CC2D2A (3), RPGRIP1L (3), TMEM67 (5)	0.10345	0.60000	4.264 e-8	3.186 e-7	
Hereditary breast and ovarian cancer syndrome	Lynch syndrome	5	5	CHEK2 (4), ATM (3), MRE11 (2), PALB2 (2), RAD51D (3)	0.08333	0.33333	7.756 e-10	7.090 e-9	
Lynch syndrome	Muir-torre syndrome	4	4	MSH2 (8), MSH6 (8), PMS2 (7), MLH1 (8)	0.08000	1.00000	9.049 e-11	9.068 e-10	166
Kleins syndrome	Waardenburg-shah syndrome	3	3	EDNRB (3), MITF (3), EDN3 (3)	0.42857	0.75000	6.574 e-11	6.659 e-10	229
Waardenburg syndrome	Waardenburg-shah syndrome	4	4	EDNRB (8), MITF (7), EDN3 (8), SOX10 (8)	0.28571	1.00000	3.054 e-13	3.864 e-12	229
Combined oxidative phosphorylation deficiency	Leigh syndrome	14	14	MTRFR (7), NARS2 (6), PNPT1 (7), EARS2 (4), FARS2 (7), AIFM1 (5), TSFM (4), MTFMT (6), TARS2 (6), GTPBP3 (7), GFM2 (6), GFM1 (5)	0.08642	0.20588	2.298 e-17	3.830 e-16	50
Childhood ataxia with cns hypomyelination	Congenital or early infantile cach syndrome	5	5	EIF2B3 (2), EIF2B1 (2), EIF2B2 (2), EIF2B4 (2), EIF2B5 (2)	0.62500	1.00000	2.913 e-18	5.082 e-17	170
Cytochrome c oxidase deficiency	Leigh syndrome	11	11	LRPPRC (5), SCO2 (5), SCO1 (4), COX15 (5), SURF1 (6), PET100 (2), COX10 (5), TACO1 (4), COX411 (2), COX8A (2), PET117 (2)	0.09402	0.55000	1.716 e-19	3.169 e-18	50
Congenital or early infantile cach syndrome	Cree leukoencephalopathy	5	5	EIF2B3 (2), EIF2B1 (2), EIF2B2 (2), EIF2B4 (2), EIF2B5 (2)	0.83333	1.00000	1.387 e-19	2.573 e-18	170
Congenital or early infantile cach syndrome	Vanishing white matter disease	5	5	EIF2B3 (3), EIF2B1 (4), EIF2B2 (4), EIF2B4 (4), EIF2B5 (4)	0.83333	1.00000	1.387 e-19	2.573 e-18	170
Hereditary breast cancer	Lynch syndrome	11	11	CHEK2 (4), EPCAM (8), MSH2 (7), MSH6 (8), ATM (3), MRE11 (2), PALB2 (3), PIK3CA (3), PMS2 (6), MLH1 (8), MUTYH (2)	0.15493	0.34375	1.245 e-20	2.444 e-19	
hereditary nonpolyposis colon cancer	Lynch syndrome	8	8	CHEK2 (3), SMARCA4 (2), EPHX1 (2), PTPRJ (2), CDKN1B (2), NFKBIZ (2), XRCC4 (2), FAN1 (3)	0.16000	1.00000	5.764 e-21	1.146 e-19	
Leigh syndrome	Mitochondrial encephalomyopathy	11	11	MTRFR (3), FARS2 (3), NDUFS2 (5), POLG (2), DNMT1L (4), SCO2 (3), NDUFV2 (4), LONP1 (4), FBXL4 (3), FOXRED1 (5), MFF (2)	0.09735	0.68750	4.566 e-21	9.112 e-20	50
Constitutional mismatch repair deficiency	Lynch syndrome	10	5	MSH2 (8), MSH6 (8), APC (1), TGFB2 (5), RNASET2 (1), PMS2 (7), TAF1B (1), MLH1 (8), ASTE1 (1), SLC22A9 (1)	0.18868	0.76923	1.134 e-23	2.570 e-22	166
Bonnevie-ullrich syndrome	Turner syndrome	7	5	CAT (2), IGFBP3 (1), SOD2 (2), SOD1 (2), VDR (1), GH1 (2), NOS2 (2)	0.77778	1.00000	1.967 e-25	4.898 e-24	285
Leigh syndrome	Mitochondrial complex deficiency	48	45	NDUFA10 (6), NDUFA12 (5), NDUFA9 (5), NDUFAF2 (5), NDUFAF6 (6), ND2 (1), NDUFS3 (6), NDUFS2 (6), TTC19 (7), LRPPRC (5), NDUFA2 (6), TIMMDC1 (5)	0.26816	0.44860	3.857 e-76	2.952 e-74	50

Disease A	Disease B	Share d Genes	Corroborate d Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	p- value	FDR Q- value	Shared Cluster
Leigh syndrome	Mitochondrial disease	72	67	HIBCH (3), TRMU (3), IARS2 (4), MTRFR (3), NARS2 (3), NDUFA10 (5), NDUFA12 (2), NDUFA9 (3), NDUFAF2 (4), NDUFAF6 (4), ND2 (1), NDUFS3 (4)	0.25899	0.67290	1.619 e-107	2.080 e-105	50