

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 "Spasticity" (166 matching pairs). · Rows: 166 · Generated: 2026-10-20 04:42:58

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Anemia, sideroblastic, 3, pyridoxine-refractory	Spasticity with hyperglycinemia	1	1	GLRX5 (3)	0.50000	1.00000	6.457e-5	1.985e-4	432
Arts syndrome	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	1	1	PRPS1 (6)	0.50000	1.00000	6.457e-5	1.985e-4	347
ATP6AP2-related disorder	Parkinsonism with spasticity, x-linked	1	1	ATP6AP2 (5)	0.50000	1.00000	6.457e-5	1.985e-4	
ATP6AP2-related disorder	X-linked parkinsonism-spasticity syndrome	1	1	ATP6AP2 (4)	0.50000	1.00000	6.457e-5	1.985e-4	
Corpus callosum agenesis with abnormal genitalia	X-linked spasticity-intellectual disability-epilepsy syndrome	1	1	ARX (7)	0.50000	1.00000	6.457e-5	1.985e-4	223
Developmental delay, epilepsy, and microcephaly	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities	1	1	NSRP1 (2)	0.50000	1.00000	6.457e-5	1.985e-4	
Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	Spastic paraplegia 79b, autosomal recessive	1	1	UCHL1 (4)	0.50000	1.00000	6.457e-5	1.985e-4	
Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	Parkinson disease 5, autosomal dominant, susceptibility to	1	1	UCHL1 (4)	0.50000	1.00000	6.457e-5	1.985e-4	
Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	Spastic paraplegia 79a, autosomal dominant	1	1	UCHL1 (4)	0.50000	1.00000	6.457e-5	1.985e-4	
Epileptic encephalopathy early infantile type 1	X-linked spasticity-intellectual disability-epilepsy syndrome	1	1	ARX (4)	0.50000	1.00000	6.457e-5	1.985e-4	223
Hydranencephaly with abnormal genitalia	X-linked spasticity-intellectual disability-epilepsy syndrome	1	1	ARX (4)	0.50000	1.00000	6.457e-5	1.985e-4	223
Intellectual developmental disorder hypotonia spastic sleep	Intellectual disability-hypotonia-spasticity-sleep disorder syndrome	1	1	ANK3 (3)	0.50000	1.00000	6.457e-5	1.985e-4	
Intellectual developmental disorder with epilepsy	Parkinsonism with spasticity, x-linked	1	1	ATP6AP2 (4)	0.50000	1.00000	6.457e-5	1.985e-4	
Intellectual developmental disorder with epilepsy	X-linked parkinsonism-spasticity syndrome	1	1	ATP6AP2 (4)	0.50000	1.00000	6.457e-5	1.985e-4	
Lethal infantile epileptic encephalopathy	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	1	0	PLAA (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Leukodystrophy, hypomyelinating with or without oligodontia and/or hypogonadotropic hypogonadism	Neurodevelopmental disorder with or without seizures, ataxia, spasticity, and demyelinating neuropathy	1	0	POLR3B (1)	0.50000	1.00000	6.457e-5	1.985e-4	636
Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	Neurodevelopmental disorder with or without seizures, ataxia, spasticity, and demyelinating neuropathy	1	0	POLR3B (1)	0.50000	1.00000	6.457e-5	1.985e-4	636

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Lower limb spasticity	Spastic paraplegia 12, autosomal dominant	1	0	RTN2 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Lower limb spasticity	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity	1	0	RTN2 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	Neurodevelopmental disorder with or without structural brain anomalies, optic atrophy, seizures and spasticity	1	0	PTPN23 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with characteristic facial and ectodermal features and tetraparesis 1	Neurodevelopmental disorder with severe-profound intellectual disability, spasticity and ectodermal features	1	0	GTPBP1 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with dysmorphic features, hypotonia, and spasticity	Neurodevelopmental disorder with neuromuscular and skeletal abnormalities	1	0	NRCAM (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity	Primary microcephaly, cortical malformation, and epileptic encephalopathy	1	0	TMX2 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with or without seizures, ataxia, spasticity, and demyelinating neuropathy	POLR3B-related disorder	1	1	POLR3B (2)	0.50000	1.00000	6.457e-5	1.985e-4	636
Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities	Spastic paraplegia 83, autosomal recessive	1	0	HPDL (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities spastic paraplegia 83, autosomal recessive	1	0	HPDL (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities spastic paraplegia 83, autosomal recessive	Spastic paraplegia 83, autosomal recessive	1	0	HPDL (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with seizures, hypotonia, spasticity, and agenesis of the corpus callosum	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum	1	0	HECTD4 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental disorder with spasticity and poor growth	Severe early-onset encephalopathy with progressive microcephaly	1	0	UFC1 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Neurodevelopmental spectrum including lissencephaly with genital anomalies and epilepsy to non-syndromic intellectual disability	X-linked spasticity-intellectual disability-epilepsy syndrome	1	1	ARX (4)	0.50000	1.00000	6.457e-5	1.985e-4	223
Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity	Spastic paraplegia 12, autosomal dominant	1	0	RTN2 (1)	0.50000	1.00000	6.457e-5	1.985e-4	
Parkinsonism with spasticity, x-linked	X-linked parkinsonism-spasticity syndrome	1	1	ATP6AP2 (7)	0.50000	1.00000	6.457e-5	1.985e-4	
Phosphoribosylpyrophosphate synthetase superactivity	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	1	1	PRPS1 (5)	0.50000	1.00000	6.457e-5	1.985e-4	347
Proud syndrome	X-linked spasticity-intellectual disability-epilepsy syndrome	1	1	ARX (4)	0.50000	1.00000	6.457e-5	1.985e-4	223
PRPS1 deficiency disorder	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	1	1	PRPS1 (2)	0.50000	1.00000	6.457e-5	1.985e-4	347
Pyridoxine refractory sideroblastic anemia 3	Spasticity with hyperglycinemia	1	1	GLRX5 (3)	0.50000	1.00000	6.457e-5	1.985e-4	432
Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive	Spasticity with hyperglycinemia	1	1	GLRX5 (3)	0.33333	1.00000	1.291e-4	3.269e-4	432
Ataxia with deafness and vision loss	X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	1	0	PRPS1 (1)	0.33333	1.00000	1.291e-4	3.269e-4	347

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Axial hypotonia	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia	1	1	MED27 (2)	0.33333	1.00000	1.291e-4	3.269e-4	316
Complex neurodevelopmental disorder with spasticity and hypoplasia of corpus callosum	Periventricular leukomalacia	1	1	RPS6KC1 (2)	0.33333	1.00000	1.291e-4	3.269e-4	
Lissencephaly, x-linked	X-linked spasticity-intellectual disability-epilepsy syndrome	1	1	ARX (8)	0.33333	1.00000	1.291e-4	3.269e-4	223
Neurodevelopmental disorder with or without seizures, ataxia, spasticity, and demyelinating neuropathy	Tremor-ataxia-central hypomyelination syndrome	1	1	POLR3B (2)	0.33333	1.00000	1.291e-4	3.269e-4	636
Diencephalic mesencephalic junction dysplasia	Microcephaly, seizures, spasticity, and brain calcification	1	1	PCDH12 (6)	0.25000	1.00000	1.937e-4	4.414e-4	
Arachnoid cysts	X-linked spasticity-intellectual disability-epilepsy syndrome	1	1	ARX (4)	0.20000	1.00000	2.583e-4	5.384e-4	223
Ectopic thyroid tissue	Spasticity	1	1	FBXO31 (2)	0.20000	1.00000	2.583e-4	5.384e-4	
Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia	Spasticity	1	1	MED27 (2)	0.20000	1.00000	2.583e-4	5.384e-4	
Repetitive behaviours	Spasticity	1	0	LIG3 (1)	0.20000	1.00000	2.583e-4	5.384e-4	
X-linked intellectual disability-limb spasticity-retinal dystrophy-diabetes insipidus syndrome	X-linked nonsyndromic hearing loss	1	1	PRPS1 (3)	0.20000	1.00000	2.583e-4	5.384e-4	347
Cerebellar hypoplasia	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia	1	1	MED27 (2)	0.14286	1.00000	3.874e-4	7.329e-4	
Cerebellar hypoplasia	Neurodevelopmental disorder with spasticity, epilepsy and cerebellar hypoplasia	1	0	INPP4A (1)	0.14286	1.00000	3.874e-4	7.329e-4	
Exudative retinopathy	Microcephaly, seizures, spasticity, and brain calcification	1	1	PCDH12 (2)	0.12500	1.00000	4.520e-4	8.260e-4	
Axial hypotonia	Spasticity	1	0	MED27 (1)	0.16667	0.50000	5.165e-4	9.125e-4	
Neurogenic bladder	Spasticity	1	0	LIG3 (1)	0.16667	0.50000	5.165e-4	9.125e-4	
Sideroblastic anemia	Spasticity with hyperglycinemia	1	1	GLRX5 (7)	0.07692	1.00000	7.748e-4	1.247e-3	432
Brain atrophy	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	1	1	PTPN23 (2)	0.07143	1.00000	8.394e-4	1.336e-3	
Brain atrophy	Neurodevelopmental disorder with or without structural brain anomalies, optic atrophy, seizures and spasticity	1	1	PTPN23 (2)	0.07143	1.00000	8.394e-4	1.336e-3	
Intellectual disability-hypotonia-spasticity-sleep disorder syndrome	Prion disease	1	1	ANK3 (2)	0.05556	1.00000	1.098e-3	1.651e-3	
Gross motor development delay	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities	1	1	MFSD2A (2)	0.05263	1.00000	1.162e-3	1.729e-3	
Marshall syndrome	Microcephaly, seizures, spasticity, and brain calcification	1	1	PCDH12 (2)	0.05000	1.00000	1.227e-3	1.808e-3	

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome	Splenomegaly	1	1	EIF2AK1 (2)	0.04762	1.00000	1.291e-3	1.888e-3	90
Autosomal recessive mental retardation	Neurodevelopmental disorder with or without seizures, ataxia, spasticity, and demyelinating neuropathy	1	0	POLR3B (1)	0.04762	1.00000	1.291e-3	1.888e-3	
Cryptorchidism	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities	1	1	MFSD2A (2)	0.04762	1.00000	1.291e-3	1.888e-3	
Autosomal recessive mental retardation	Neurodevelopmental disorder with spasticity, epilepsy and cerebellar hypoplasia	1	0	INPP4A (1)	0.04762	1.00000	1.291e-3	1.888e-3	
Febrile convulsion	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities	1	1	MFSD2A (2)	0.04348	1.00000	1.420e-3	2.040e-3	
Autosomal recessive primary microcephaly	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities	1	0	MFSD2A (1)	0.04000	1.00000	1.550e-3	2.192e-3	54
Neurodevelopmental disorder with progressive spasticity and brain abnormalities	Uterine polyp	1	1	EEFSEC (2)	0.04000	1.00000	1.550e-3	2.192e-3	
Anhedonia	Intellectual disability-hypotonia-spasticity-sleep disorder syndrome	1	1	ANK3 (2)	0.03704	1.00000	1.679e-3	2.343e-3	
Dyskeratosis congenita	Neurodevelopmental disorder with spasticity, epilepsy and cerebellar hypoplasia	1	1	INPP4A (2)	0.03448	1.00000	1.808e-3	2.494e-3	
Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities	Polymicrogyria	1	1	SHMT2 (2)	0.03333	1.00000	1.872e-3	2.572e-3	230
Hypomyelinating leukodystrophy	Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome	1	1	EIF2AK1 (2)	0.03125	1.00000	2.002e-3	2.719e-3	
Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain imaging abnormalities	Primary microcephaly	1	1	MFSD2A (4)	0.02703	1.00000	2.324e-3	3.054e-3	54
Distal hereditary motor neuropathy	Lower limb spasticity	1	1	RTN2 (5)	0.02632	1.00000	2.389e-3	3.123e-3	
Distal hereditary motor neuropathy	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity	1	1	RTN2 (5)	0.02632	1.00000	2.389e-3	3.123e-3	
Chronic obstructive pulmonary disease	Neurodevelopmental disorder with progressive spasticity and brain abnormalities	1	1	EEFSEC (2)	0.02381	1.00000	2.647e-3	3.413e-3	
Hypospadias	Neurodevelopmental disorder with progressive spasticity and brain abnormalities	1	1	EEFSEC (2)	0.02326	1.00000	2.712e-3	3.483e-3	
Neurodegenerative disease	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities	1	0	HPDL (1)	0.02273	1.00000	2.776e-3	3.558e-3	
Neurodegenerative disease	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities spastic paraplegia 83, autosomal recessive	1	0	HPDL (1)	0.02273	1.00000	2.776e-3	3.558e-3	
Neurodevelopmental disorder with spasticity, epilepsy and cerebellar hypoplasia	Nystagmus	1	1	INPP4A (2)	0.02128	1.00000	2.970e-3	3.788e-3	
Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	Peripheral nervous system disease	1	1	UCHL1 (5)	0.01852	1.00000	3.422e-3	4.286e-3	
Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome	Non-neoplastic peripheral nervous system disease	1	1	UCHL1 (4)	0.01818	1.00000	3.487e-3	4.343e-3	

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Hemolytic anemia	Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome	1	1	EIF2AK1 (3)	0.01786	1.00000	3.551e-3	4.417e-3	
Neurodevelopmental disorder with seizures, hypotonia, spasticity, and agenesis of the corpus callosum	Pancreatitis	1	1	HECTD4 (2)	0.01724	1.00000	3.680e-3	4.562e-3	
Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum	Pancreatitis	1	1	HECTD4 (2)	0.01724	1.00000	3.680e-3	4.562e-3	
Combined oxidative phosphorylation deficiency	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay	1	1	TRMT5 (7)	0.01449	1.00000	4.390e-3	5.339e-3	16
Intellectual disability-hypotonia-spasticity-sleep disorder syndrome	Non-organic psychosis	1	1	ANK3 (2)	0.01429	1.00000	4.455e-3	5.410e-3	
Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities	Spastic ataxia	1	1	HPDL (2)	0.01408	1.00000	4.520e-3	5.478e-3	
Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities spastic paraplegia 83, autosomal recessive	Spastic ataxia	1	1	HPDL (2)	0.01408	1.00000	4.520e-3	5.478e-3	
Intellectual disability-hypotonia-spasticity-sleep disorder syndrome	Status epilepticus	1	1	ANK3 (3)	0.01389	1.00000	4.584e-3	5.544e-3	
Corneal astigmatism	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus	1	1	TNR (3)	0.01389	1.00000	4.584e-3	5.544e-3	430
Parkinsonism with spasticity, x-linked	X-linked intellectual disability	1	1	ATP6AP2 (5)	0.01333	1.00000	4.778e-3	5.751e-3	
X-linked intellectual disability	X-linked parkinsonism-spasticity syndrome	1	1	ATP6AP2 (4)	0.01333	1.00000	4.778e-3	5.751e-3	
Dystonia	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia	1	1	MED27 (2)	0.01235	1.00000	5.165e-3	6.162e-3	
Age-related macular degeneration	Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus	1	1	TNR (3)	0.01176	1.00000	5.424e-3	6.440e-3	
Intellectual developmental disorder, x-linked	Parkinsonism with spasticity, x-linked	1	1	ATP6AP2 (4)	0.01099	1.00000	5.811e-3	6.843e-3	
Intellectual developmental disorder, x-linked	X-linked parkinsonism-spasticity syndrome	1	1	ATP6AP2 (6)	0.01099	1.00000	5.811e-3	6.843e-3	
Neurodevelopmental disorder with progressive spasticity and brain abnormalities	Obstructive airway disease	1	1	EEFSEC (2)	0.01000	1.00000	6.392e-3	7.497e-3	
Hereditary spastic paraplegia	Lower limb spasticity	1	1	RTN2 (2)	0.00943	1.00000	6.779e-3	7.882e-3	
Hereditary spastic paraplegia	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	1	1	PTPN23 (2)	0.00943	1.00000	6.779e-3	7.882e-3	
Hereditary spastic paraplegia	Neurodevelopmental disorder with or without structural brain anomalies, optic atrophy, seizures and spasticity	1	1	PTPN23 (2)	0.00943	1.00000	6.779e-3	7.882e-3	
Hereditary spastic paraplegia	Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity	1	1	RTN2 (2)	0.00943	1.00000	6.779e-3	7.882e-3	
Leigh syndrome	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities	1	1	HPDL (3)	0.00909	1.00000	7.038e-3	8.163e-3	

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Leigh syndrome	Neurodevelopmental disorder with progressive spasticity and brain white matter abnormalities spastic paraplegia 83, autosomal recessive	1	1	HPDL (3)	0.00909	1.00000	7.038e-3	8.163e-3	
Congenital neurologic anomalies	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity	1	1	TMX2 (2)	0.00862	1.00000	7.425e-3	8.572e-3	
Cerebellar ataxia	Microcephaly, seizures, spasticity, and brain calcification	1	1	PCDH12 (2)	0.00820	1.00000	7.813e-3	8.931e-3	
Lower limb spasticity	Spastic paraplegia	1	1	RTN2 (5)	0.00800	1.00000	8.006e-3	9.125e-3	
Neuronopathy, distal hereditary motor, autosomal recessive 11, with spasticity	Spastic paraplegia	1	1	RTN2 (5)	0.00800	1.00000	8.006e-3	9.125e-3	
Endometrial neoplasms	Neurodevelopmental disorder with progressive spasticity and brain abnormalities	1	1	EEFSEC (2)	0.00752	1.00000	8.523e-3	9.682e-3	
Complex neurodevelopmental disorder	Complex neurodevelopmental disorder with spasticity and hypoplasia of corpus callosum	1	0	RPS6KC1 (1)	0.00741	1.00000	8.652e-3	9.793e-3	
Myopia	Neurodevelopmental disorder with dysmorphic features, hypotonia, and spasticity	1	1	NRCAM (2)	0.00735	1.00000	8.716e-3	9.858e-3	
Anxiety disorder	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity	1	1	TMX2 (2)	0.00680	1.00000	9.427e-3	1.059e-2	
Neurodevelopmental disorder with dysmorphic features, hypotonia, and spasticity	Orofacial cleft	1	1	NRCAM (2)	0.00633	1.00000	1.014e-2	1.134e-2	
Microcephaly	Microcephaly, seizures, spasticity, and brain calcification	1	0	PCDH12 (1)	0.00625	1.00000	1.027e-2	1.144e-2	
Microcephaly	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities	1	1	SHMT2 (2)	0.00625	1.00000	1.027e-2	1.144e-2	
Microcephaly	Neurodevelopmental disorder with spasticity and poor growth	1	0	UFC1 (1)	0.00625	1.00000	1.027e-2	1.144e-2	
Microcephaly	Neurodevelopmental disorder with spasticity, epilepsy and cerebellar hypoplasia	1	0	INPP4A (1)	0.00625	1.00000	1.027e-2	1.144e-2	
Esophageal cancer	Neurodevelopmental disorder with seizures, hypotonia, spasticity, and agenesis of the corpus callosum	1	1	HECTD4 (2)	0.00621	1.00000	1.033e-2	1.151e-2	
Esophageal cancer	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum	1	1	HECTD4 (2)	0.00621	1.00000	1.033e-2	1.151e-2	
Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus	Sarcoidosis	1	1	TNR (3)	0.00610	1.00000	1.052e-2	1.171e-2	
Ischemic heart disease	Neurodevelopmental disorder with seizures, hypotonia, spasticity, and agenesis of the corpus callosum	1	1	HECTD4 (2)	0.00588	1.00000	1.091e-2	1.212e-2	
Ischemic heart disease	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum	1	1	HECTD4 (2)	0.00588	1.00000	1.091e-2	1.212e-2	
Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies	Optic atrophy	1	1	ELOVL1 (2)	0.00526	1.00000	1.220e-2	1.348e-2	10
Celiac disease	Neurodevelopmental disorder with seizures, hypotonia, spasticity, and agenesis of the corpus callosum	1	1	HECTD4 (2)	0.00515	1.00000	1.246e-2	1.376e-2	

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Celiac disease	Neurodevelopmental disorder with seizures, spasticity, and complete or partial agenesis of the corpus callosum	1	1	HECTD4 (2)	0.00515	1.00000	1.246e-2	1.376e-2	
Cataract	Neurodevelopmental disorder with spasticity, cataracts, and cerebellar hypoplasia	1	1	MED27 (2)	0.00472	1.00000	1.362e-2	1.498e-2	
Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities	Obsessive-compulsive disorder	1	1	SHMT2 (3)	0.00427	1.00000	1.504e-2	1.643e-2	
Diverticular disease	Neurodevelopmental disorder with severe-profound intellectual disability, spasticity and ectodermal features	1	1	GTPBP1 (3)	0.00402	1.00000	1.601e-2	1.741e-2	
Mitochondrial disease	Peripheral neuropathy with variable spasticity, exercise intolerance, and developmental delay	1	1	TRMT5 (6)	0.00402	1.00000	1.601e-2	1.741e-2	16
Anorexia nervosa	Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities	1	1	SHMT2 (3)	0.00398	1.00000	1.614e-2	1.753e-2	
Neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities	Tourette syndrome	1	1	SHMT2 (3)	0.00388	1.00000	1.659e-2	1.798e-2	
Juvenile idiopathic arthritis	Neurodevelopmental disorder with dysmorphic features, hypotonia, and spasticity	1	1	NRCAM (2)	0.00376	1.00000	1.711e-2	1.851e-2	
Colorectal neoplasms	Neurodevelopmental disorder with dysmorphic features, hypotonia, and spasticity	1	1	NRCAM (2)	0.00347	1.00000	1.853e-2	1.996e-2	
Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome	Nonalcoholic fatty liver disease	1	1	EIF2AK1 (3)	0.00315	1.00000	2.040e-2	2.189e-2	
Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies	Retinitis pigmentosa	1	1	ELOVL1 (2)	0.00273	1.00000	2.357e-2	2.517e-2	10
Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	Non-specific syndromic intellectual disability	1	1	PTPN23 (3)	0.00271	1.00000	2.376e-2	2.528e-2	
Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	Non-specific syndromic intellectual disability	1	1	TRAPPC4 (4)	0.00271	1.00000	2.376e-2	2.528e-2	3
Neurodevelopmental disorder with or without structural brain anomalies, optic atrophy, seizures and spasticity	Non-specific syndromic intellectual disability	1	1	PTPN23 (3)	0.00271	1.00000	2.376e-2	2.528e-2	
Neurodevelopmental disorder with speech and motor delay and spasticity	Non-specific syndromic intellectual disability	1	1	RAB1A (2)	0.00271	1.00000	2.376e-2	2.528e-2	3
Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus	Open angle glaucoma	1	1	TNR (3)	0.00270	1.00000	2.389e-2	2.541e-2	430
Intellectual disability	Neurodevelopmental disorder and structural brain anomalies with or without seizures and spasticity	1	0	PTPN23 (1)	0.00242	1.00000	2.667e-2	2.816e-2	
Intellectual disability	Neurodevelopmental disorder with or without structural brain anomalies, optic atrophy, seizures and spasticity	1	0	PTPN23 (1)	0.00242	1.00000	2.667e-2	2.816e-2	
Complex neurodevelopmental disorder with spasticity and hypoplasia of corpus callosum	Global developmental delay	1	1	RPS6KC1 (2)	0.00229	1.00000	2.815e-2	2.962e-2	
Neurodevelopmental disorder, nonprogressive, with spasticity and transient opisthotonus	Parkinson disease	1	1	TNR (3)	0.00189	1.00000	3.409e-2	3.568e-2	
Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities	Obstructive pulmonary disease	1	1	NSRP1 (3)	0.00179	1.00000	3.609e-2	3.775e-2	

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Myocardial infarction	Neurodevelopmental disorder with cerebellar hypoplasia and spasticity	1	1	INTS8 (2)	0.00172	1.00000	3.738e-2	3.894e-2	8
Leukoencephalopathy, motor delay, spasticity, and dysarthria syndrome	Oligodendroglioma	1	1	EIF2AK1 (2)	0.00157	1.00000	4.093e-2	4.253e-2	
Hypomyelination with brain stem and spinal cord involvement and leg spasticity	Systemic lupus erythematosus	1	1	DARS1 (2)	0.00152	1.00000	4.236e-2	4.396e-2	101
Atrial fibrillation	Neurodevelopmental disorder with speech and motor delay and spasticity	1	1	RAB1A (2)	0.00116	1.00000	5.572e-2	5.722e-2	
Complex neurodevelopmental disorder with spasticity and hypoplasia of corpus callosum	Neurodevelopmental disorder	1	0	RPS6KC1 (1)	0.00099	1.00000	6.495e-2	6.624e-2	
Neurodevelopmental disorder	Neurodevelopmental disorder with cerebellar hypoplasia and spasticity	1	1	INTS8 (5)	0.00099	1.00000	6.495e-2	6.624e-2	
Neurodevelopmental disorder	Neurodevelopmental disorder with epilepsy, spasticity, and brain atrophy	1	1	TRAPPC4 (6)	0.00099	1.00000	6.495e-2	6.624e-2	3
Neurodevelopmental disorder	Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity	1	1	ESAM (5)	0.00099	1.00000	6.495e-2	6.624e-2	3
Neurodevelopmental disorder	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity	1	1	TMX2 (5)	0.00099	1.00000	6.495e-2	6.624e-2	
Neurodevelopmental disorder	Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	1	1	PLAA (4)	0.00099	1.00000	6.495e-2	6.624e-2	
Neurodevelopmental disorder	Neurodevelopmental disorder with severe-profound intellectual disability, spasticity and ectodermal features	1	1	GTPBP1 (4)	0.00099	1.00000	6.495e-2	6.624e-2	
Neurodevelopmental disorder	Neurodevelopmental disorder with spasticity and poor growth	1	1	UFC1 (5)	0.00099	1.00000	6.495e-2	6.624e-2	
Neurodevelopmental disorder	Neurodevelopmental disorder with spasticity, seizures, and brain abnormalities	1	1	NSRP1 (6)	0.00099	1.00000	6.495e-2	6.624e-2	
Neurodevelopmental disorder	Neurodevelopmental disorder with speech and motor delay and spasticity	1	1	RAB1A (2)	0.00099	1.00000	6.495e-2	6.624e-2	3
Obesity	Spasticity with hyperglycinemia	1	1	GLRX5 (4)	0.00083	1.00000	7.754e-2	7.857e-2	
Breast cancer	Complex neurodevelopmental disorder with spasticity and hypoplasia of corpus callosum	1	1	RPS6KC1 (2)	0.00080	1.00000	8.110e-2	8.205e-2	
Autism	Neurodevelopmental disorder with microcephaly, cortical malformations, and spasticity	1	1	TMX2 (2)	0.00068	1.00000	9.504e-2	9.587e-2	
Alzheimer disease	Neurodevelopmental disorder with spasticity and poor growth	1	1	UFC1 (2)	0.00045	1.00000	1.431e-1	1.438e-1	
Alzheimer disease	Neurodevelopmental disorder with speech and motor delay and spasticity	1	1	RAB1A (2)	0.00045	1.00000	1.431e-1	1.438e-1	
Neurodevelopmental disorder with intracranial hemorrhage, seizures, and spasticity	Schizophrenia	1	1	ESAM (4)	0.00039	1.00000	1.639e-1	1.643e-1	
Neurodevelopmental disorder with progressive microcephaly, spasticity, and brain anomalies	Schizophrenia	1	1	PLAA (3)	0.00039	1.00000	1.639e-1	1.643e-1	

Disease A	Disease B	Shared Genes	Corroborated Shared Genes	Shared Gene Names	Similarity Score	Overlap Coefficient	P-value	FDR Q-value	Shared Cluster
Diabetes mellitus type 2	Neurodevelopmental disorder with cerebellar hypoplasia and spasticity	1	1	INTS8 (2)	0.00032	1.00000	1.989e-1	1.990e-1	
Diabetes mellitus type 2	Neurodevelopmental disorder with speech and motor delay and spasticity	1	1	RAB1A (2)	0.00032	1.00000	1.989e-1	1.990e-1	