

Solving patients with rare diseases through programmatic reanalysis of genome-phenome data

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gene	genotype	transcript	HGVS:cDNA	HGVS:protein	ERN	patient_hpos	omim_inheritance	Family_inheritance	reported consanguinity	experimental consanguinity	in_ROH_1Mb
ANO10	1/1	NM_001346464.1	c.132dupA	p.Asp45fs	NMD	Ataxia	Spinocerebellar ataxia, autosomal recessive 10, 613728 (3), Autosomal recessive	recessive	Not available	likely-consanguineous	yes
ATM	1/1	NM_000051.3	c.3894dupT	p.Ala1299fs	ITHACA	Cryptorchidism//Triangular face//Telangiectasia//Intellectual disability//Ataxia//Global developmental delay//Abnormality of the tricuspid valve//Asthma//Abnormality of the vertebrae//Oral-pharyngeal dysphagia	Ataxia-telangiectasia, 208900 (3), Autosomal recessive; (Breast cancer, susceptibility to), 114480 (3), Autosomal dominant; Lymphoma, B-cell non-Hodgkin, somatic (3); Lymphoma, mantle cell, somatic (3); T-cell prolymphocytic leukemia, somatic (3)	recessive	TRUE	likely-consanguineous	yes
ATP13A2	1/1	NM_022089.4	c.364C>T	p.Gln122*	RND	Cerebellar atrophy//Progressive cerebellar ataxia//Cerebral cortical atrophy//Progressive spastic paraparesis//Peripheral neuropathy	Kufor-Rakeb syndrome, 606693 (3), Autosomal recessive; Spastic paraplegia 78, autosomal recessive, 617225 (3), Autosomal recessive	recessive	FALSE	uncertain	no
B4GALNT1	1/1	NM_001478.5	c.1298A>C	p.Asp433Ala	RND	Psychosis//Intellectual disability//Spasticity//Dysarthria//Lower limb spasticity//Spastic gait//Babinski sign//Progressive spastic paraparesis	Spastic paraplegia 26, autosomal recessive, 609195 (3), Autosomal recessive	recessive	FALSE	uncertain	no
CLN5	1/1	NM_006493.4	c.625T>G	p.Tyr209Asp	RND	Blindness//Spasticity//Lower limb spasticity//Spastic gait//Babinski sign//Progressive extrapyramidal movement disorder//Progressive spastic paraparesis	Ceroid lipofuscinosis, neuronal, 5, 256731 (3), Autosomal recessive	recessive	FALSE	uncertain	no
CSF1R	1/1	NM_001288705.2	c.1969+115_1969+116delAG		RND	Open mouth//High palate//Long face//Low-set ears//Spasticity//Spastic gait//Lower limb hyperreflexia//Spastic tetraplegia//Babinski sign//Hemiplegia/hemiparesis//Biconcave vertebral bodies//CNS demyelination//Periventricular white matter hyperdensities//Arachnoid cyst	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820 (3), Autosomal dominant	recessive	TRUE	consanguineous	yes
DDHD2	1/1	NM_001164232.1	c.859C>T	p.Arg287*	RND	Intellectual disability//Spasticity//Spastic tetraparesis//Lower limb spasticity//Spastic gait//Hypoplasia of the corpus callosum//Babinski sign//Progressive spastic paraparesis	Spastic paraplegia 54, autosomal recessive, 615033 (3), Autosomal recessive	recessive	TRUE	consanguineous	yes
DYSF	1/1	NM_001130987.2	c.3167G>A	p.Arg1056Gln	NMD	Not available	Miyoshi muscular dystrophy 1, 254130 (3), Autosomal recessive; Muscular dystrophy, limb-girdle, type 2B, 253601 (3), Autosomal recessive; Myopathy, distal, with anterior tibial onset, 606768 (3), Autosomal recessive	recessive	Not available	likely-consanguineous	yes
EXOSC3	1/1	NM_016042.4	c.395A>C	p.Asp132Ala	NMD	Pontocerebellar atrophy	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive	recessive	Not available	non-consanguineous	no
EXOSC3	1/1	NM_016042.4	c.395A>C	p.Asp132Ala	RND	Hypertelorism//Ophthalmoparesis//Nystagmus//Eczema//Muscular hypotonia//Dysarthria//Cerebellar atrophy//Lower limb spasticity//Limb ataxia//Progressive cerebellar ataxia//Truncal ataxia//Recurrent respiratory infections//Tetraparesis//Drooling//Psychomotor deterioration//Lower limb hyperreflexia//Thoracic scoliosis//Babinski sign//Progressive pulmonary function impairment//Cerebellar vermis atrophy//Lower limb hypertonia//Postnatal growth retardation//Seasonal allergy//Psychomotor retardation	Pontocerebellar hypoplasia, type 1B, 614678 (3), Autosomal recessive	recessive	Not available	non-consanguineous	no
FBXL4	1/1	NM_001278716.2	c.1648_1649delGA	p.Asp550fs	NMD	Not available	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive	recessive	Not available	consanguineous	no
FBXL4	1/1	NM_001278716.2	c.1648_1649delGA	p.Asp550fs	NMD	Not available	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471 (3), Autosomal recessive	recessive	Not available	consanguineous	yes
FKBP14	1/1	NM_017946.4	c.362dupC	p.Glu122fs	ITHACA	Retrognathia//Arachnodactyly//Delayed gross motor development//Scoliosis//High, narrow palate//Abnormality of the fibula//Short stature//Pes valgus//Microtia//Infantile axial hypotonia//Short metatarsal//Unilateral strabismus//Severe Myopia	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 (3), Autosomal recessive	recessive	Not available	uncertain	no
GBA2	1/1	NM_020944.3	c.700C>T	p.Arg234*	RND	Abnormality of eye movement//Intellectual disability//Dysphagia//Gait ataxia//Limb ataxia//Hypokinesia//Spastic dysarthria//Impaired vibratory sensation//Progressive spastic paraparesis//Cognitive impairment	Spastic paraplegia 46, autosomal recessive, 614409 (3), Autosomal recessive	recessive	TRUE	consanguineous	yes

GJB2	1/1	NM_004004.6	c.34delG	p.Gly12fs	ITHACA	Not available	Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Autosomal recessive; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant	recessive	Not available	NA	no
KLHL7	1/1	NM_001031710.3	c.618+1G>A		ITHACA	Pierre-Robin sequence//Abnormal facial shape//Limb hypertonial//Distal arthrogyposis//Infantile axial hypotonia	Cold-induced sweating syndrome 3, 617055 (3), Autosomal recessive; Retinitis pigmentosa 42, 612943 (3), Autosomal dominant	recessive	TRUE	likely-consanguineous	yes
MAP3K20	1/1	NM_016653.3	c.282dupT	p.Asn95fs	NMD	Scoliosis//Progressive muscle weakness//Myalgia//EMG: myopathic abnormalities//Scapular winging//Proximal muscle weakness in lower limbs//Proximal muscle weakness in upper limbs//Hypertrophy of the lower limb//Ring fibers	Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive	recessive	FALSE	consanguineous	yes
MAP3K20	1/1	NM_016653.3	c.282dupT	p.Asn95fs	NMD	Motor delay//Hyperlordosis//Progressive muscle weakness//Scapular winging//Proximal muscle weakness in lower limbs//Proximal muscle weakness in upper limbs	Centronuclear myopathy 6 with fiber-type disproportion, 617760 (3), Autosomal recessive; Split-foot malformation with mesoaxial polydactyly, 616890 (3), Autosomal recessive	recessive	FALSE	consanguineous	yes
MUTYH	1/1	NM_001128425.1	c.1147delC	p.Ala385fs	GENTURIS	Adenomatous colonic polyposis//Large intestinal polyposis//Intestinal polyposis//Colorectal polyposis	Adenomas, multiple colorectal, 608456 (3), Autosomal recessive; Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 (3), Somatic mutation; Gastric cancer, somatic, 613659 (3)	recessive	Not available	non-consanguineous	no
NPHS1	1/1	NM_004646.3	c.2417C>A	p.Ala806Asp	ITHACA	Gingival overgrowth//Dolichocephaly//Delayed speech and language development//Global developmental delay//Overlapping toe//Delayed gross motor development//Intellectual disability, moderate//Short stature//Decreased body weight//Delayed fine motor development//Torsion of the penis	Nephrotic syndrome, type 1, 256300 (3), Autosomal recessive	recessive	TRUE	consanguineous	yes
OSGEP	1/1	NM_017807.4	c.332T>C	p.Ile111Thr	ITHACA	Nephrotic syndrome//Microcephaly//Delayed speech and language development//Muscular hypotonia//Intellectual disability, mild//Abnormal facial shape//Short stature	Galloway-Mowat syndrome 3, 617729 (3), Autosomal recessive	recessive	Not available	uncertain	no
PTRH2	1/1	NM_001015509.2	c.272_273delCT	p.Ala91fs	RND	Hearing impairment//Intellectual disability//Peripheral neuropathy	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263 (3), Autosomal recessive	recessive	TRUE	consanguineous	yes
SELENON	1/1	NM_020451.3	c.1090C>T	p.Gln364Ter	NMD	High palate//Facial asymmetry//Myopia//Growth hormone deficiency//Muscular hypotonia//Hyporeflexia//Muscle weakness//Joint hypermobility//Weak cry//Pes planus//Respiratory insufficiency//Tetraparesis//Frequent falls//Hyporeflexia of lower limbs//Scoliosis//Respiratory insufficiency due to muscle weakness//Myopathy//Skeletal muscle atrophy//Generalized muscle weakness//Generalized amyotrophy//Proximal muscle weakness//Short stature//Craniofacial asymmetry//Arnold-Chiari type 1 malformation//Congenital facial diplegia//Infantile muscular hypotonia//Proximal muscle weakness in lower limbs//Decreased Achilles reflex//Abnormal elasticity of skin//Complete breech presentation//Hyporeflexia of upper limbs//Fatty replacement of skeletal muscle//Abnormality of the brachial nerve plexus	Muscular dystrophy, rigid spine, 1, 602771 (3), Autosomal recessive; Myopathy, congenital, with fiber-type disproportion, 255310 (3), Autosomal recessive, Autosomal dominant	recessive	FALSE	uncertain	no
TAF2	1/1	NM_003184.4	c.2531C>T	p.Pro844Leu	ITHACA	Wide mouth//Progressive microcephaly//Synophrys//Seizures//Profound global developmental delay	Mental retardation, autosomal recessive 40, 615599 (3), Autosomal recessive	recessive	TRUE	consanguineous	no
TMEM107	1/1	NM_183065.4	c.134A>G	p.Glu45Gly	ITHACA	Macrocephaly//Cerebellar hypoplasia//Absent speech//Ventricular septal defect//Abnormal facial shape//Delayed gross motor development//Severe global developmental delay//Hamartoma of tongue//Delayed ability to sit//Postaxial polydactyly	Leukoencephalopathy, brain calcifications, and cysts, 614561 (3), Autosomal recessive	recessive	TRUE	consanguineous	yes
TRIP4	1/1	NM_016213.5	c.760C>T	p.Arg254*	NMD	Pontocerebellar atrophy	?Muscular dystrophy, congenital, Davignon-Chauveau type, 617066 (3), Autosomal recessive; Spinal muscular atrophy with congenital bone fractures 1, 616866 (3), Autosomal recessive	recessive	Not available	uncertain	no