# 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 |
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| 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//Pschomotor 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 |

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| 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 hypertonia//Distal arthrogryposis//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 I 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 |