Orpha Code	Preferred term (English term)	Synonyms EN
Diseases		
15	Achondroplasia	Achondroplasia
17	Fatal infantile lactic acidosis with methylmalonic aciduria	
20	3-hydroxy-3-methylglutaric aciduria	3-hydroxy-3-methylglutaryl-CoA lyase deficiency|HMG-CoA lyase deficiency|Hydroxymethylglutaric aciduria
25	Glutaryl-CoA dehydrogenase deficiency	GA1|GCDHD|Glutaric acidemia type 1|Glutaric aciduria type 1|Glutaryl-coenzyme A dehydrogenase deficiency
27	Vitamin B12-unresponsive methylmalonic acidemia	Methylmalonyl-CoA mutase deficiency|Methylmalonyl-Coenzyme A mutase deficiency|Vitamin B12-unresponsive methylmalonic aciduria
33	Isovaleric acidemia	Isovaleric acid CoA dehydrogenase deficiency
37	Acrodermatitis enteropathica	AEZ|Acrodermatitis enteropathica, zinc deficiency type|Inherited zinc deficiency
42	Medium chain acyl-CoA dehydrogenase deficiency	ACADM deficiency|Carnitine deficiency secondary to medium-chain acyl-CoA dehydrogenase deficiency|MCAD deficiency|MCADD|Medium chain acyl-coenzyme A dehydrogenase deficiency|
43	X-linked adrenoleukodystrophy	ALD|X-ALD|X-linked ALD
51	Aicardi-Goutières syndrome	Encephalopathy with basal ganglia calcification|Encephalopathy with intracranial calcification and chronic lymphocytosis of cerebrospinal fluid
60	Alpha-1-antitrypsin deficiency	
63	Alport syndrome	Alport deafness-nephropathy
70	Proximal spinal muscular atrophy	SMA
95	Friedreich ataxia	FA|FRDA
97	Familial paroxysmal ataxia	Episodic ataxia type 2
100	Ataxia-telangiectasia	Louis-Bar syndrome
104	Leber hereditary optic neuropathy	LHON|Leber optic atrophy
110	Bardet-Biedl syndrome	BBS
117	Behçet disease	
124	Blackfan-Diamond anemia	Aase syndrome|Aase-Smith II syndrome|Congenital PRCA|Congenital hypoplastic anemia, Blackfan-Diamond type|Congenital pure red cell aplasia|Diamond-Blackfan anemia
125	Bloom syndrome	BSyn
130	Brugada syndrome	Bangungut|Dream disease|Idiopathic ventricular fibrillation, Brugada type|Pokkuri death syndrome|SUNDS|Sudden unexplained nocturnal death syndrome
136	CADASIL	Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy|Hereditary multi-infarct dementia
144	Lynch syndrome	
145	Hereditary breast and ovarian cancer syndrome	
154	Familial isolated dilated cardiomyopathy	Familial or idiopathic dilated cardiomyopathy

157	Carnitine palmitoyltransferase II deficiency	CPT2|CPTII|Carnitine palmitoyltransferase deficiency type 2
158	Systemic primary carnitine deficiency	CDSP|CUD|Carnitine transporter defect|Carnitine uptake deficiency|Deficiency of plasma-membrane carnitine transporter|SPCD|CACT deficiency
167	Chédiak-Higashi syndrome	Chédiak-Higashi disease|Chédiak-Higashi-Steinbrink syndrome
175	Cartilage-hair hypoplasia	Autosomal recessive metaphyseal chondrodysplasia|Metaphyseal chondrodysplasia, McKusick type
180	Choroideremia	CHM|Tapetochoroidal dystrophy
198	Occipital horn syndrome	EDS IX|Ehlers-Danlos syndrome type 9|Ehlers-Danlos syndrome type IX|X-linked cutis laxa||Brachmann-de Lange syndrome
201	Cowden syndrome	Cowden disease|Multiple hamartoma syndrome
214	Cystinuria	Cystinuria-lysinuria syndrome
215	Congenital stationary night blindness	Congenital essential nyctalopia
223	Nephrogenic diabetes insipidus	
225	Maternally-inherited diabetes and deafness	MIDD|Mitochondrial diabetes
229	Familial aortic dissection	Annuloaortic ectasia|Cystic medial necrosis of aorta
232	Sickle cell anemia	Sickle cell disease
244	Primary ciliary dyskinesia	PCD
269	Facioscapulohumeral dystrophy	FSH dystrophy|FSHD|Facioscapulohumeral muscular dystrophy|Facioscapulohumeral myopathy|Landouzy-Dejerine myopathy
270	Oculopharyngeal muscular dystrophy	OPMD
273	Steinert myotonic dystrophy	DM1|MD1|Myotonic dystrophy type 1|Steinert disease
275	Severe combined immunodeficiency due to DCLRE1C deficiency	SCID due to ARTEMIS deficiency|SCID due to DCLRE1C deficiency|SCID, Athabascan type|SCID, Athabaskan type|Severe combined immunodeficiency due to ARTEMIS deficiency|Severe combined immunodeficiency, Athabascan type|Severe combined immunodeficiency, Athabaskan type
285	Hypermobile Ehlers-Danlos syndrome	EDS III|EDS-HT|Ehlers-Danlos syndrome hypermobility type|Ehlers-Danlos syndrome type 3|hEDS
286	Vascular Ehlers-Danlos syndrome	Arterial-ecchymotic EDS|EDS IV|Ehlers-Danlos syndrome type 4|Sack-Barabas syndrome|vEDS
287	Classical Ehlers-Danlos syndrome	Classical EDS|cEDS
296	Ollier disease	Dyschondroplasia
306	Benign familial infantile epilepsy	BFIE|BFIS|Benign familial infantile convulsions|Benign familial infantile seizures

312	Autosomal dominant epidermolytic ichthyosis	BCIE|Bullous congenital ichthyosiform erythroderma|Bullous congenital ichthyosiform erythroderma of Brock|Bullous ichthyosis|EHK|EI|Epidermolytic hyperkeratosis|Ichthyosis hystrix Brocq type
313	Lamellar ichthyosis	Classic lamellar ichthyosis|Congenital lamellar ichthyosis|LI
321	Multiple osteochondromas	Bessel-Hagen disease|Multiple cartilaginous exostoses
324	Fabry disease	Alpha-galactosidase A deficiency|Anderson-Fabry disease|Angiokeratoma corporis diffusum|Diffuse angiokeratoma|FD
327	Congenital factor VII deficiency	Congenital proconvertin deficiency|Hypoproconvertinemia
329	Congenital factor XI deficiency	Hemophilia C|PTA deficiency|Plasma thromboplastin antecedent deficiency|Rosenthal factor deficiency|Rosenthal syndrome
337	Fibrodysplasia ossificans progressiva	FOP|Myositis ossificans progressiva|Stone man syndrome
342	Familial Mediterranean fever	Benign paroxysmal peritonitis|Benign recurrent polyserositis|FMF|Familial paroxysmal polyserositis|Periodic disease
358	Gitelman syndrome	Primary renal tubular hypokalemic hypomagnesemia with hypocalciuria
366	Glycogen storage disease due to glycogen debranching enzyme deficiency	Amylo-1,6-glucosidase deficiency|Cori disease|Cori-Forbes disease|Forbes disease|GDE deficiency|GSD due to glycogen debranching enzyme deficiency|GSD type 3|GSDIII|Glycogen storage disease type 3|Glycogen storage disease type III|Glycogenosis due to glycogen debranching enzyme deficiency|Glycogenosis type 3|Glycogenosis type III|Limit dextrinosis
368	Glycogen storage disease due to muscle glycogen phosphorylase deficiency	GSD due to muscle glycogen phosphorylase deficiency|GSD type 5|GSD type V|Glycogen storage disease type 5|Glycogen storage disease type V|Glycogenosis due to muscle glycogen phosphorylase deficiency|Glycogenosis type 5|Glycogenosis type V|McArdle disease|Myophosphorylase deficiency
388	Hirschsprung disease	Aganglionic megacolon|Congenital intestinal aganglionosis|HSCR
394	Classic homocystinuria	Cystathionine beta-synthase deficiency|Homocystinuria due to cystathionine beta-synthase deficiency
395	Homocystinuria due to methylene tetrahydrofolate reductase deficiency	MTHFR deficiency|Methylene tetrahydrofolate reductase deficiency
399	Huntington disease	Huntington chorea
405	Familial hypocalciuric hypercalcemia	FBH|FBHH|FHH|Familial benign hypercalcemia|Familial benign hypocalciuric hypercalcemia

414	Gyrate atrophy of choroid and retina	HOGA|Hyperornithinemia|Hyperornithinemia-gyrate atrophy of choroid and retina syndrome|Ornithine aminotransferase deficiency
423	Malignant hyperthermia of anesthesia	Hyperthermia of anesthesia
429	Hypochondroplasia	
432	Normosmic congenital hypogonadotropic hypogonadism	Gonadotropic deficiency|Isolated congenital gonadotropin deficiency|Normosmic idiopathic hypogonadotropic hypogonadism|nIHH
447	Paroxysmal nocturnal hemoglobinuria	Marchiafava-Micheli disease|PNH
461	Recessive X-linked ichthyosis	RXLI|Steroid sulfatase deficiency|X-linked ichthyosis|XLI
478	Kallmann syndrome	Congenital hypogonadotropic hypogonadism with anosmia|Olfacto-genital pathological sequence||
480	Kearns-Sayre syndrome	
512	Metachromatic leukodystrophy	Arylsulfatase A deficiency|MLD
523	Hereditary leiomyomatosis and renal cell cancer	Familial leiomyomatosis and renal cell cancer|Familial leiomyomatosis cutis et uteri|Familial leiomyomatosis with renal carcinoma|Familial multiple cutaneous leiomyomas|HLRCC|Hereditary leiomyomatosis|Hereditary leiomyomatosis with renal carcinoma|Hereditary multiple cutaneous leiomyomas|MCUL|Multiple cutaneous and uterine leiomyomas|Reed syndrome
524	Li-Fraumeni syndrome	
536	Systemic lupus erythematosus	Disseminated lupus erythematosus|SLE
550	MELAS	Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes|Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes
551	MERRF	Fukuhara syndrome|Myoclonus epilepsy associated with ragged-red fibres
552	MODY	Maturity-onset diabetes of the young
558	Marfan syndrome	MFS
562	McCune-Albright syndrome	Gonadotropin-independent female-limited sexual precocity
569	Familial or sporadic hemiplegic migraine	
570	Moebius syndrome	Congenital facial diplegia|Möbius syndrome
580	Mucopolysaccharidosis type 2	Hunter syndrome|Iduronate 2-sulfatase deficiency|MPS2|MPSII|Mucopolysaccharidosis type II
581	Mucopolysaccharidosis type 3	MPS3|MPSIII|Mucopolysaccharidosis type III|Sanfilippo disease
586	Cystic fibrosis	CF|Mucoviscidosis
587	Muir-Torre syndrome	Multiple keratoacanthoma, Muir-Torre type
590	Congenital myasthenic syndrome	CMS
597	Central core disease	
606	Proximal myotonic myopathy	Myotonic dystrophy type 2|Proximal myotonic dystrophy|Ricker disease|Ricker syndrome
610	Bethlem myopathy	Benign autosomal dominant myopathy

611	Inclusion body myositis	IBM|Sporadic inclusion body myositis|sIBM
614	Thomsen and Becker disease	Myotonia congenita
631	Non-acquired isolated growth hormone deficiency	Congenital IGHD|Congenital isolated GH deficiency|Congenital isolated growth hormone deficiency
635	Neuroblastoma	
636	Neurofibromatosis type 1	NF1|Von Recklinghausen disease
637	Neurofibromatosis type 2	Bilateral acoustic neurofibromatosis|Central neurofibromatosis|NF2
641	Multifocal motor neuropathy	MMN|MMNCB|Multifocal motor neuropathy with conduction block
644	NARP syndrome	Neurogenic muscle weakness-ataxia-retinitis pigmentosa syndrome|Neuropathy-ataxia-retinitis pigmentosa syndrome
652	Multiple endocrine neoplasia type 1	MEN1|Wermer syndrome
653	Multiple endocrine neoplasia type 2	MEN2
656	Familial idiopathic steroid-resistant nephrotic syndrome	Familial idiopathic nephrotic syndrome
661	Ondine syndrome	CCHS|Central congenital hypoventilation syndrome|Congenital central alveolar hypoventilation syndrome|Ondine curse
663	Mitochondrial DNA-related progressive external ophthalmoplegia	Maternally-inherited CPEO|Maternally-inherited chronic progressive external ophthalmoplegia|mtDNA-related progressive external ophthalmoplegia
666	Osteogenesis imperfecta	Brittle bone disease|Glass bone disease|Lobstein disease|OI|Osteopsathyrosis|Porak and Durante disease
676	Hereditary chronic pancreatitis	
684	Paramyotonia congenita of Von Eulenburg	Paramyotonia congenita
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	GSD due to phosphoglycerate kinase 1 deficiency|Glycogenosis due to phosphoglycerate kinase 1 deficiency
726	Alpers-Huttenlocher syndrome	Alpers progressive sclerosing poliodystrophy|Alpers syndrome|Progressive neuronal degeneration of childhood with liver disease
730	Autosomal dominant polycystic kidney disease	ADPKD
731	Autosomal recessive polycystic kidney disease	AR-PKD
733	Familial adenomatous polyposis	Colorectal adenomatous polyposis|FAP|Familial polyposis coli
739	Prader-Willi syndrome	Prader-Labhart-Willi syndrome|Willi-Prader syndrome
743	Severe hereditary thrombophilia due to congenital protein S deficiency	Autosomal recessive thrombophilia due to congenital protein S deficiency
750	Pseudoachondroplasia	Pseudoachondroplastic dysplasia|Pseudoachondroplastic spondyloepiphyseal dysplasia
758	Pseudoxanthoma elasticum	Gronblad-Strandberg-Touraine syndrome|PXE
760	Purine nucleoside phosphorylase deficiency	PNP deficiency|PNPase deficiency

766	Hemolytic anemia due to red cell pyruvate kinase deficiency	Pyruvate kinase deficiency of erythrocytes
767	Polyarteritis nodosa	Küssmaul-Maier disease|PAN|Periarteritis nodosa
774	Hereditary hemorrhagic telangiectasia	HHT|Rendu-Osler disease|Rendu-Osler-Weber disease
778	Rett syndrome	
790	Retinoblastoma	
791	Retinitis pigmentosa	
803	Amyotrophic lateral sclerosis	ALS|Charcot disease|Lou Gehrig disease
805	Tuberous sclerosis complex	Bourneville syndrome|Tuberous sclerosis
813	Silver-Russell syndrome	Silver-Russell dwarfism
821	Sotos syndrome	Cerebral gigantism
822	Hereditary spherocytosis	Minkowski-Chauffard disease
827	Stargardt disease	Fundus flavimaculatus|Stargardt 1
828	Stickler syndrome	Hereditary progressive arthroophthalmopathy
839	Congenital nephrotic syndrome, Finnish type	Finnish congenital nephrosis
846	Alpha-thalassemia	
847	Alpha-thalassemia-X-linked intellectual disability syndrome	ATR-X syndrome
848	Beta-thalassemia	
892	Von Hippel-Lindau disease	Familial cerebelloretinal angiomatosis|Lindau disease|VHL|Von Hippel-Lindau syndrome
897	Waardenburg-Shah syndrome	Shah-Waardenburg syndrome|WS4|Waardenburg syndrome type 4|Waardenburg-Hirschsprung syndrome
905	Wilson disease	Hepatolenticular degeneration
909	Cerebrotendinous xanthomatosis	CTX|Sterol 27-hydroxylase deficiency
912	Zellweger syndrome	Cerebrohepatorenal syndrome|ZS
1062	Hereditary neurocutaneous malformation	
1159	Progressive pseudorheumatoid arthropathy of childhood	Spondyloepiphyseal dysplasia tarda-progressive arthropathy syndrome
1180	Ataxia-hypogonadism-choroidal dystrophy syndrome	Boucher-Neuhäuser syndrome
1214	Progressive hemifacial atrophy	Hemifacial atrophy|PHA|Parry-Romberg syndrome|Progressive facial hemiatrophy|Romberg syndrome
1243	Best vitelliform macular dystrophy	BMD|BVMD|Best disease|Best macular dystrophy|Early-onset vitelliform macular dystrophy|Juvenile-onset vitelliform macular dystrophy|Polymorphic vitelline macular degeneration|Vitelliform macular dystrophy type 2
1331	Familial prostate cancer	
1359	Carney complex	Carney syndrome|Myxoma-spotty pigmentation-endocrine overactivity syndrome
1428	Familial chondromalacia patellae	
1496	Corpus callosum agenesis-neuronopathy syndrome	Andermann syndrome|Charlevoix disease
1572	Common variable immunodeficiency	CVID|Idiopathic immunoglobulin deficiency|Primary antibody deficiency|Primary hypogammaglobulinemia
1598	Monosomy 18p	18p- syndrome|De Grouchy syndrome

1652	Dent disease	Dent syndrome|Low-molecular-weight proteinuria with hypercalciuria and nephrocalcinosis|Renal Fanconi syndrome with nephrocalcinosis and renal stones|X-linked recessive hypercalciuric hypophosphatemic rickets|X-linked recessive nephrolithiasis
1775	Dyskeratosis congenita	DC|DKC|Zinsser-Engman-Cole syndrome
1826	Frontometaphyseal dysplasia	
1872	Cone rod dystrophy	
1942	Myoclonic-astastic epilepsy	Doose syndrome|EMAS|Epilepsy with myoclonic-astatic seizures|Epilepsy with myoclonic-atonic seizures|MAE|Myoclonic atonic epilepsy|Myoclonic-astatic epilepsy in early childhood
1945	Rolandic epilepsy	BECRS|BECTS|BRE|Benign epilepsy of childhood with centrotemporal spikes|Benign familial epilepsy of childhood with rolandic spikes|Benign rolandic epilepsy|Centrotemporal epilepsy
1949	Benign familial neonatal epilepsy	BFNS|Benign familial neonatal convulsions|Benign familial neonatal seizures
1980	Bilateral striopallidodentate calcinosis	BSPDC|Cerebrovascular ferrocalcinosis|Idiopathic basal ganglia calcification|PFBC|Primary familial brain calcification
2073	Narcolepsy type 1	Gélineau disease|Narcolepsy-cataplexy
2111	Cystic hamartoma of lung and kidney	Graham-Boyle-Troxell syndrome
2134	Atypical hemolytic-uremic syndrome	Atypical HUS|aHUS
2148	Lissencephaly type 1 due to doublecortin gene mutation	X-linked lissencephaly type 1
2199	Epidermolytic palmoplantar keratoderma	Diffuse erythrodermic palmoplantar keratoderma, Voerner type|Diffuse erythrodermic palmoplantar keratoderma, Vörner type|EPPK|Epidermolytic palmoplantar keratoderma of Voerner|Epidermolytic palmoplantar keratoderma of Vörner
2295	Familial articular hypermobility syndrome	Familial joint instability syndrome|Familial joint laxity|Joint instability syndrome
2309	Pachyonychia congenita	PC
2346	Angioosteohypertrophic syndrome	Klippel-Trénaunay-Weber syndrome
2612	Linear nevus sebaceus syndrome	Nevus sebaceus of Jadassohn|Nevus sebaceus syndrome|Organoid nevus syndrome|Schimmelpenning syndrome|Solomon syndrome
2721	Odonto-onycho-dermal dysplasia	OODD
2836	PEHO syndrome	Progressive encephalopathy with edema, hypsarrhythmia and optic atrophy|Progressive encephalopathy-optic atrophy syndrome
2841	Familial benign chronic pemphigus	Benign chronic familial pemphigus of Hailey-Hailey|Hailey-Hailey disease
2869	Peutz-Jeghers syndrome	Hamartomatous intestinal polyposis|PJS|Polyps and spots syndrome
2875	Phakomatosis pigmentovascularis	

2884	Piebaldism	
2897	Pityriasis rubra pilaris	
2909	Rothmund-Thomson syndrome	Poikiloderma of Rothmund-Thomson|RTS
2940	Porencephaly	
2953	Musculocontractural Ehlers-Danlos syndrome	Adducted thumb-clubfoot syndrome|Distal arthrogryposis with peculiar facies and hydronephrosis|Dündar syndrome|Ehlers-Danlos syndrome, Kosho type|mcEDS
3095	Atypical Rett syndrome	Atypical RTT|Rett syndrome variant
3143	Autoimmune polyendocrinopathy type 2	APS type 2|APS2|Autoimmune polyendocrine syndrome type 2|Autoimmune polyglandular syndrome type 2|Autoimmune thyroid disease and/or type 1 diabetes-Addison disease syndrome|Schmidt syndrome
3156	Senior-Loken syndrome	Nephronophthisis with retinal dystrophy|Renal dysplasia-retinal aplasia syndrome|SLSN
3197	Hereditary hyperekplexia	Congenital stiff man syndrome|Familial startle disease|Hereditary hyperexplexia|Hyperekplexia|Kok disease|Stiff baby syndrome
3221	Generalized resistance to thyroid hormone	Deafness-thyroid hormone resistance syndrome|Refetoff syndrome
3261	Autoimmune lymphoproliferative syndrome	ALPS|Canale-Smith syndrome
3286	Catecholaminergic polymorphic ventricular tachycardia	Bidirectional tachycardia induced by catecholamine|CPVT|Double tachycardia induced by catecholamines|Malignant paroxysmal ventricular tachycardia|Multifocal ventricular premature beats
3337	Primary Fanconi syndrome	Primary Fanconi renotubular syndrome
3440	Waardenburg syndrome	
3463	Wolfram syndrome	DIDMOAD syndrome|Diabetes insipidus-diabetes mellitus-optic atrophy-deafness syndrome
26106	Hereditary diffuse gastric cancer	FDGC|Familial diffuse cancer of stomach|Familial diffuse gastric cancer|HDGC|Hereditary diffuse cancer of stomach|Hereditary diffuse gastric adenocarcinoma
28378	Tyrosinemia type 2	Keratosis palmoplantaris-corneal dystrophy syndrome|Oculocutaneous tyrosinemia|Richner-Hanhart syndrome|Tyrosinemia due to TAT deficiency|Tyrosinemia due to tyrosine aminotransferase deficiency|Tyrosinemia type II
29072	Hereditary pheochromocytoma-paraganglioma	Familial pheochromocytoma-paraganglioma
30924	Primary hypomagnesemia with secondary hypocalcemia	HOMG1|HSH|Hypomagnesemia caused by selective magnesium malabsorption|Hypomagnesemia intestinal type 1|Intestinal hypomagnesemia with secondary hypocalcemia|PHSH

32960	Tumor necrosis factor receptor 1 associated periodic syndrome	Familial Hibernian fever|TNF receptor 1-associated periodic syndrome|TRAPS syndrome
33069	Dravet syndrome	DS|SMEI|Severe myoclonic epilepsy of infancy|Severe myoclonus epilepsy of infancy
33574	Glutamate-cysteine ligase deficiency	Gamma-glutamylcysteine synthetase deficiency
34149	Autosomal dominant tubulointerstitial kidney disease	ADTKD|Autosomal dominant medullary cystic kidney disease|MCKD
34515	Autosomal recessive limb-girdle muscular dystrophy type 2I	LGMD2I|Limb-girdle muscular dystrophy due to FKRP deficiency
35122	Congenital sucrase-isomaltase deficiency	CSID|Congenital sucrase-isomaltose malabsorption|Congenital sucrose intolerance|Disaccharide intolerance
35125	Epidermal nevus syndrome	Epidermal hamartoma syndrome
35173	X-linked dominant chondrodysplasia punctata	CDPX2|CDPXD|CPXD|Chondrodystrophia calcificans congenita|Conradi-Hünermann-Happle syndrome|X-linked chondrodysplasia punctata type 2
35708	Aromatic L-amino acid decarboxylase deficiency	AADC deficiency
36383	COL4A1-related familial vascular leukoencephalopathy	COL4A1-related brain small vessel disease with hemorrhage|COL4A1-related retinal arteriolar tortuosity-infantile hemiparesis-autosomal dominant leukoencephalopathy syndrome
36387	Generalized epilepsy with febrile seizures-plus	GEFS+
37553	Andersen-Tawil syndrome	Andersen syndrome|LQT7|Long QT syndrome type 7
39041	Omenn syndrome	Combined immunodeficiency with hypereosinophilia
45358	Congenital fibrosis of extraocular muscles	FEOM
47044	Hereditary papillary renal cell carcinoma	HPRCC
49382	Achromatopsia	ACHM|Complete or incomplete color blindness|Pingelapese blindness|Rod monochromacy|Rod monochromatism|Total color blindness
50942	Striate palmoplantar keratoderma	Keratosis palmoplantaris striata|Keratosis palmoplantaris striata et areata|Keratosis palmoplantaris varians of Wachters
51083	Familial short QT syndrome	SQTS
52430	Inclusion body myopathy with Paget disease of bone and frontotemporal dementia	IBMPFD|Limb-girdle muscular dystrophy with Paget disease of bone|Pagetoid amyotrophic lateral sclerosis|Pagetoid neuroskeletal syndrome
52503	X-linked creatine transporter deficiency	Creatine transporter deficiency|SLC6A8 deficiency
54260	Left ventricular noncompaction	LVNC|Left ventricular hypertrabeculation|Spongy myocardium
64545	Benign idiopathic neonatal seizures	BINS|Benign nonfamilial neonatal seizures
64745	Pruritic urticarial papules and plaques of pregnancy	PUPPP|Polymorphic eruption of pregnancy
64753	Spinocerebellar ataxia with axonal neuropathy type 2	AOA2|Ataxia-oculomotor apraxia type 2|SCAN 2|SCAR1
64755	Becker nevus syndrome	Pigmentary hairy epidermal nevus
69076	Familial renal glucosuria	SGLT2 deficiency

69663	Low phospholipid-associated cholelithiasis	ABCB4-related cholelithiasis|LPAC
69665	Intrahepatic cholestasis of pregnancy	Gravidic intrahepatic cholestasis|Pregnancy-related cholestasis|Recurrent intrahepatic cholestasis of pregnancy
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	IRAK4 deficiency
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency	Autosomal recessive sepiapterin reductase-deficient DRD|DRD due to SRD|SPR deficiency|Sepiapterin reductase deficiency
70595	Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	SANDO
73229	HANAC syndrome	Autosomal dominant familial hematuria-retinal arteriolar tortuosity-contractures syndrome|Hereditary angiopathy-nephropathy-aneurysms-muscle cramps syndrome
73273	Growth delay due to insulin-like growth factor I resistance	Resistance to IGF-1
75249	Familial isolated restrictive cardiomyopathy	Familial or idiopathic restrictive cardiomyopathy
77293	Niemann-Pick disease type B	
79146	Familial progressive hyperpigmentation	Melanosis diffusa congenita|Melanosis universalis hereditaria|Universal melanosis
79230	Hemochromatosis type 2	Juvenile hemochromatosis
79239	Classic galactosemia	GALT deficiency|Galactose-1-phosphate uridyltransferase deficiency|Galactosemia type 1
79276	Acute intermittent porphyria	
79278	Autosomal erythropoietic protoporphyria	EPP
79314	L-2-hydroxyglutaric aciduria	L-2-HGA|L-2-hydroxyglutaric acidemia
79323	MPDU1-CDG	CDG syndrome type If|CDG-If|CDG1F|Carbohydrate deficient glycoprotein syndrome type If|Congenital disorder of glycosylation type 1f|Congenital disorder of glycosylation type If
79397	Epidermolysis bullosa simplex with mottled pigmentation	EBS-MP
79404	Junctional epidermolysis bullosa, generalized severe	Epidermolysis bullosa letalis|JEB, generalized severe|JEB-H|Junctional epidermolysis bullosa generalisata gravis|Junctional epidermolysis bullosa, Herlitz type|Junctional epidermolysis bullosa, Herlitz-Pearson type
79430	Hermansky-Pudlak syndrome	HPS
79432	Oculocutaneous albinism type 2	OCA2
79443	Pseudohypoparathyroidism type 1A	AHO-PHP syndrome Ia|Albright hereditary osteodystrophy-PHP syndrome Ia
79445	Pseudopseudohypoparathyroidism	AHO-PPHP syndrome|Albright hereditary osteodystrophy-PPHP syndrome
79457	Maculopapular cutaneous mastocytosis	Urticaria pigmentosa
84064	Syndromic diarrhea	Phenotypic diarrhea|SD/THE|Syndromic diarrhea/Tricho-hepato-enteric syndrome|Tricho-hepato-enteric syndrome|Trichohepatoenteric syndrome

84081	Senior-Boichis syndrome	Boichis disease|Nephronophthisis-hepatic fibrosis syndrome
85128	Bothnia retinal dystrophy	Västerbotten dystrophy
85167	Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	SMD-CRD
85451	ATTRV122I amyloidosis	ATTR cardiomyopathy|ATTRV122I-related amyloidosis|TTR-related amyloid cardiomyopathy|TTR-related cardiac amyloidosis|Transthyretin amyloid cardiopathy|Transthyretin-related familial amyloid cardiomyopathy
86923	Hereditary palmoplantar keratoderma, Gamborg-Nielsen type	Hereditary palmoplantar hyperkeratosis, Gamborg-Nielsen type|PPK, Gamborg-Nielsen type
87884	Non-syndromic genetic deafness	Isolated genetic deafness
88642	Channelopathy-associated congenital insensitivity to pain	Channelopathy-associated CIP
88661	Amelogenesis imperfecta	
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	Tuberous sclerosis/polycystic kidney disease contiguous gene syndrome
89838	Epidermolysis bullosa simplex, autosomal recessive K14	EBS, autosomal recessive K14|EBS-AR|KRT14|KRT14-related autosomal recessive EBS|KRT14-related autosomal recessive epidermolysis bullosa simplex
89936	X-linked hypophosphatemia	X-linked hypophosphatemic rickets|XLH
90342	Xeroderma pigmentosum variant	XPV
90348	Autosomal dominant cutis laxa	ADCL
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Classic 21-OHD CAH
91387	Familial thoracic aortic aneurysm and aortic dissection	Familial TAAD
91492	Early-onset non-syndromic cataract	
93111	Renal cysts and diabetes syndrome	HNF1B-MODY|HNF1B-related renal cysts and diabetes syndrome|MODY5|RCAD syndrome|Renal cysts-maturity-onset diabetes of the young syndrome|Renal dysfunction-early-onset diabetes syndrome
93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	CMTDIE|Charcot-Marie-Tooth disease-nephropathy syndrome
93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis	
93284	Spondyloepiphyseal dysplasia tarda	
93311	Multiple epiphyseal dysplasia type 5	BHMED|Bilateral hereditary micro-epiphyseal dysplasia|EDM5|MED5|Polyepiphyseal dysplasia type 5
93314	Spondylometaphyseal dysplasia, Kozlowski type	
93921	Schwannomatosis	NF3|Neurilemmomatosis|Neurofibromatosis type 3
94068	Spondyloepiphyseal dysplasia congenita	Congenital spondyloepiphyseal dysplasia|SEDC|Spranger-Wiedemann disease
94089	Pseudohypoparathyroidism type 1B	
95494	Combined pituitary hormone deficiencies, genetic forms	Familial congenital hypopituitarism|Multiple pituitary hormone deficiencies, genetic forms

95716	Familial thyroid dyshormonogenesis	Thyroid dyshormonogenesis
98267	Genetic non-syndromic obesity	Monogenic obesity due to a leptin-melanocortin pathway anomaly
98673	Autosomal dominant optic atrophy, classic form	Autosomal dominant optic atrophy, Kjer type|Kjer optic atrophy|Optic atrophy type 1
98756	Spinocerebellar ataxia type 2	SCA2
98758	Spinocerebellar ataxia type 6	SCA6
98760	Spinocerebellar ataxia type 8	SCA8
98764	Spinocerebellar ataxia type 27	SCA27
98766	Spinocerebellar ataxia type 5	SCA5
98769	Spinocerebellar ataxia type 15/16	SCA15/16
98784	Autosomal dominant sleep-related hypermotor epilepsy	ADNFLE|Autosomal dominant nocturnal frontal lobe epilepsy
98808	Autosomal dominant dopa-responsive dystonia	Autosomal dominant Segawa syndrome|DYT5a|GTPCH1-deficient DRD|GTPCH1-deficient dopa-responsive dystonia|HPD with marked diurnal fluctuation|Hereditary progressive dystonia with marked diurnal fluctuation
98809	Paroxysmal kinesigenic dyskinesia	Familial PKD|Familial paroxysmal kinesigenic dyskinesia|Paroxysmal kinesigenic choreathetosis
98820	Familial focal epilepsy with variable foci	FFEVF|Familial partial epilepsy with variable foci
98878	Hemophilia A	Factor VIII deficiency
98895	Becker muscular dystrophy	BMD|Becker dystrophinopathy
98896	Duchenne muscular dystrophy	DMD|Severe dystrophinopathy, Duchenne type
98960	Thiel-Behnke corneal dystrophy	Anterior limiting membrane dystrophy type 2|Anterior limiting membrane dystrophy type II|Corneal dystrophy of Bowman layer type 2|Corneal dystrophy of Bowman layer type II|Curly fiber corneal dystrophy|Honeycomb corneal dystrophy|TBCD|Waardenburg-Jonker corneal dystrophy
99013	Spastic paraplegia type 7	SPG7
99015	Spastic paraplegia type 2	SPG2|Spastic gait type 2|Spastic paraparesis type 2|X-linked spastic paraplegia type 2
99718	Leber plus disease	LHON plus disease
99749	Kostmann syndrome	Infantile agranulocytosis|Severe congenital neutropenia type 3
99943	Autosomal dominant Charcot-Marie-Tooth disease type 2J	CMT2J
100973	FRAXE intellectual disability	Intellectual disability associated with fragile site FRAXE
100976	Bathing suit ichthyosis	BSI
100985	Autosomal dominant spastic paraplegia type 4	SPG4
101001	Autosomal recessive spastic paraplegia type 21	Mast syndrome|SPG21
101039	Female restricted epilepsy with intellectual disability	EFMR|Juberg-Hellman syndrome
101075	X-linked Charcot-Marie-Tooth disease type 1	CMT1X|CMTX1
101076	X-linked Charcot-Marie-Tooth disease type 2	CMTX2

101081	Charcot-Marie-Tooth disease type 1A	CMT1A|Microduplication 17p12
101685	Rare non-syndromic intellectual disability	Rare NSID
137678	Czech dysplasia, metatarsal type	
139455	Autosomal recessive bestrophinopathy	Retinopathy, Burgess-Black type
139536	Distal hereditary motor neuropathy type 5	Distal HMN V|Distal hereditary motor neuropathy type V|Distal spinal muscular atrophy type 5|dHMN5
140957	Autosomal dominant macrothrombocytopenia	
163634	Maffucci syndrome	
163690	Hypotonia-cystinuria syndrome	HCS
163937	X-linked intellectual disability, Najm type	MICPCH|X-linked intellectual disability-microcephaly-pontocerebellar hypoplasia syndrome
163956	X-linked intellectual disability, Nascimento type	X-linked intellectual disability-nail dystrophy-seizures syndrome
166002	Multiple epiphyseal dysplasia due to collagen 9 anomaly	
166119	Isolated osteopoikilosis	
169186	Autosomal recessive centronuclear myopathy	AR-CNM
169189	Autosomal dominant centronuclear myopathy	AD-CNM
199267	Infantile digital fibromatosis	Inclusion body fibromatosis|Recurring digital fibrous tumor of childhood|Reye tumor
199627	Atypical autism	
200418	Immunodeficiency with factor I anomaly	Complete factor I deficiency
206549	Autosomal recessive limb-girdle muscular dystrophy type 2L	LGMD2L
208513	Spinocerebellar ataxia type 29	Congenital nonprogressive spinocerebellar ataxia|SCA29
209886	Familial juvenile hyperuricemic nephropathy type 1	FJHN type 1|Familial juvenile gouty nephropathy|Familial nephropathy with gout|UMOD-associated FJHN|UMOD-associated familial juvenile hyperuricemic nephropathy
209905	Brain-lung-thyroid syndrome	Choreoathetosis-hypothyroidism-neonatal respiratory distress syndrome
220460	Attenuated familial adenomatous polyposis	AFAP|Attenuated FAP|Attenuated familial polyposis coli
221046	Poikiloderma with neutropenia	Poikiloderma with neutropenia, Clericuzio type
221150	Pitt-Hopkins-like syndrome	
227535	Hereditary breast cancer	Familial breast cancer|Familial breast carcinoma|Hereditary breast carcinoma
228285	Acquired cutis laxa	Cutis laxa acquisita
230839	Classical-like Ehlers-Danlos syndrome type 1	Classical-like EDS type 1|Ehlers-Danlos syndrome due to tenascin-X deficiency|clEDS type 1
238468	Hypohidrotic ectodermal dysplasia	Anhidrotic ectodermal dysplasia|HED
247525	Citrullinemia type I	ASS deficiency|Argininosuccinate synthase deficiency|Argininosuccinate synthetase deficiency|Argininosuccinic acid synthase deficiency|Argininosuccinic acid synthetase deficiency|CTLN1|Citrullinemia type 1|Classic citrullinemia
247868	NLRP12-associated hereditary periodic fever syndrome	FCAS2|Familial cold autoinflammatory syndrome type 2|NAPS12

252183	Neurofibroma	
254892	Autosomal dominant progressive external ophthalmoplegia	adPEO
263463	CHST3-related skeletal dysplasia	Chondrodysplasia with congenital joint dislocations, CHST3 type|SDCD, CHST3 type|Spondyloepiphyseal dysplasia with congenital joint dyslocations, CHST3 type
263534	Acral peeling skin syndrome	Acral PSS|Acral deciduous skin|Localized PSS|Localized deciduous skin
264580	Glycogen storage disease due to liver phosphorylase kinase deficiency	GSD due to liver phosphorylase kinase deficiency|GSD type 9A|GSD type 9C|GSD type IXa|GSD type IXc|Glycogen storage disease type 9A|Glycogen storage disease type 9C|Glycogen storage disease type IXa|Glycogen storage disease type IXc|Glycogenosis due to liver phosphorylase kinase deficiency|Glycogenosis type 9A|Glycogenosis type 9C|Glycogenosis type IXa|Glycogenosis type IXc|XLG
275761	Lysosomal acid lipase deficiency	LAL deficiency
275864	Behavioral variant of frontotemporal dementia	bv-FTD
275872	Frontotemporal dementia with motor neuron disease	FTD-ALS|FTD-MND|Frontotemporal dementia with amyotrophic lateral sclerosis
276580	Autosomal dominant hyperinsulinism due to Kir6.2 deficiency	Autosomal dominant hyperinsulinemic hypoglycemia due to Kir6.2 deficiency|Dominant KATP hyperinsulinism due to Kir6.2 deficiency
276598	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	Hyperinsulinemic hypoglycemia due to SUR1 deficiency, diazoxide-resistant focal form
282166	Inherited Creutzfeldt-Jakob disease	Inherited CJD
284289	Adult-onset autosomal recessive cerebellar ataxia	Autosomal recessive spinocerebellar ataxia type 10|SCAR10
284984	Aneurysm-osteoarthritis syndrome	
289266	Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation	
289548	Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency	
289601	Hereditary arterial and articular multiple calcification syndrome	CALJA|Calcification of joints and arteries
293987	Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation syndrome	ROHHAD|ROHHADNET|Rapid-onset childhood obesity-hypothalamic dysfunction-hypoventilation-autonomic dysregulation-neural tumors syndrome
300547	Autosomal recessive infantile hypercalcemia	Familial infantile hypercalcemia with suppressed intact parathyroid hormone
300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation	

313808	Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia	ALSP|Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia|Autosomal dominant leukoencephalopathy with neuroaxonal spheroids|FPSG|Familial dementia, Neumann type|Familial progressive subcortical gliosis|GPSC|HDLS|Hereditary diffuse leukoencephalopathy with spheroids|POLD|Pigmentary orthochromatic leukodystrophy|Subcortical gliosis of Neumann
313936	PENS syndrome	
314647	Non-progressive cerebellar ataxia with intellectual disability	
314795	SHOX-related short stature	
314811	Short stature due to GHSR deficiency	Ghrelin receptor deficiency|Short stature due to growth hormone secretagogue receptor deficiency
319462	Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations	
324611	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to KIF5A mutation	CMT2 due to KIF5A mutation
329235	X-linked central congenital hypothyroidism with late-onset testicular enlargement	IGSF1 deficiency syndrome|X-linked central congenital hypothyroidism with late-onset macroorchidism
329336	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy	Adult-onset CPEO with mitochondrial myopathy
329457	Distal arthrogryposis type 5D	DA5D|Distal arthrogryposis type 5 without ophthalmoparesis|Distal arthrogryposis type 5 without ophthalmoplegia
352490	Autism spectrum disorder due to AUTS2 deficiency	ASD due to AUTS2 deficiency|AUTS2 syndrome
352577	Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome	Bainbridge-Roppers syndrome
352582	Familial infantile myoclonic epilepsy	FIME|Familial infantile myoclonus epilepsy
352731	Oculocutaneous albinism type 1	OCA1
363611	Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome	
363649	Mandibular hypoplasia-deafness-progeroid features-lipodystrophy syndrome	MDP syndrome|MDPL syndrome|Mandibular hypoplasia-hearing loss-progeroid syndrome
363686	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome	
370022	Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome	Poretti-Boltshauser syndrome
370109	Ataxia-telangiectasia variant	v-AT
370334	Extraskeletal Ewing sarcoma	EOE|Extraosseous Ewing sarcoma|Extraosseous Ewing tumor|Extraskeletal Ewing tumor
391330	X-linked osteoporosis with fractures	
391651	Glomus tumor	
391665	Homozygous familial hypercholesterolemia	HoFH

401996	Karyomegalic interstitial nephritis	KIN|Systemic karyomegaly
404560	Familial atypical multiple mole melanoma syndrome	B-K mole syndrome|FAMM-PC syndrome|FAMMM syndrome|Familial Clark nevus syndrome|Familial atypical mole syndrome|Familial atypical multiple mole melanoma-pancreatic carcinoma syndrome|Familial dysplastic nevus syndrome|Melanoma-pancreatic cancer syndrome
411590	Wolfram-like syndrome	
420611	Transient myeloproliferative syndrome	TMD|Transient abnormal myelopoiesis|Transient myeloproliferative disease
435804	Short stature-advanced bone age-early-onset osteoarthritis syndrome	
439854	Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease	Fatal congenital hypertrophic cardiomyopathy due to GSD|Fatal congenital hypertrophic cardiomyopathy due to glycogenosis
440392	Interstitial lung disease due to SP-C deficiency	Interstitial lung disease due to surfactant protein C deficiency
442835	Undetermined early-onset epileptic encephalopathy	Undetermined EOEE
443197	X-linked erythropoietic protoporphyria	X-linked dominant erythropoietic protoporphyria|X-linked dominant protoporphyria|XLDPP|XLP|XLPP
444463	Autoimmune hemolytic anemia-autoimmune thrombocytopenia-primary immunodeficiency syndrome	Evans syndrome associated with primary immunodeficiency|TPPII deficiency|TPPII-related immunodeficiency, autoimmunity, and neurodevelopmental delay with impaired glycolysis and lysosomal expansion disease|TRIANGLE disease|Tripeptidyl-peptidase II deficiency
449291	Symptomatic form of fragile X syndrome in female carrier	
457260	X-linked intellectual disability-hypotonia-movement disorder syndrome	
458718	Idiopathic spontaneous coronary artery dissection	Idiopathic SCAD
464321	Multifocal lymphangioendotheliomatosis-thrombocytopenia syndrome	Cutaneovisceral angiomatosis-thrombocytopenia syndrome|MLT|Multifocal lymphangioendotheliomatosis with thrombocytopenia
465508	Symptomatic form of hemochromatosis type 1	Symptomatic form of HFE-related hereditary hemochromatosis|Symptomatic form of classic hemochromatosis
466026	Class I glucose-6-phosphate dehydrogenase deficiency	Class I G6PD deficiency|Severe hemolytic anemia due to G6PD deficiency
468678	Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome	White-Sutton syndrome
486815	Congenital muscular dystrophy-respiratory failure-skin abnormalities-joint hyperlaxity syndrome	Congenital muscular dystrophy, Davignon-Chauveau type
488333	Autosomal dominant Charcot-Marie-Tooth disease type 2W	Autosomal dominant Charcot-Marie-Tooth disease type 2 due to HARS mutation|CMT2W

494444	DIAPH1-related sensorineural hearing loss-thrombocytopenia syndrome	DIAPH1-related sensorineural deafness-thrombocytopenia syndrome
496790	Optic atrophy-peripheral neuropathy-developmental delay syndrome	Harel-Yoon syndrome
497737	Epidermolytic nevus	Epidermal nevus with epidermolytic hyperkeratosis|Epidermolytic epidermal nevus|Epidermolytic verrucous epidermal nevus
497757	MME-related autosomal dominant Charcot Marie Tooth disease type 2	MME-related autosomal dominant CMT2|MME-related autosomal dominant hereditary motor and sensory neuropathy type 2
528084	Non-specific syndromic intellectual disability	Complex neurodevelopmental disorder
530983	Lamb-Shaffer syndrome	SOX5 haploinsufficiency syndrome
536516	Myopathic Ehlers-Danlos syndrome	EDS/myopathy overlap syndrome|Myopathic EDS
544254	SYNGAP1-related developmental and epileptic encephalopathy	SYNGAP1-related DEE
289877	Transient hyperammonemia of the newborn	
576349	NLRC4-related familial cold autoinflammatory syndrome	FCAS4 ; Familial cold autoinflammatory syndrome 4 ; NLRC4-related familial cold urticaria

Category

99	Autosomal dominant cerebellar ataxia	ADCA|Autosomal dominant spinocerebellar ataxia
166	Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy	CMT/HMSN|Charcot-Marie-Tooth hereditary neuropathy
352	Galactosemia	
442	Congenital hypothyroidism	
593	Myofibrillar myopathy	
599	Distal myopathy	Distal muscular dystrophy
1531	Craniosynostosis	
34526	Familial primary hypomagnesemia	
35696	Mitochondrial disorder due to a defect in mitochondrial protein synthesis	COXPD|Combined OXPHOS defect|Combined OXPHOS deficiency|Combined oxidative phosphorylation defect
35705	Neurometabolic disorder due to serine deficiency	Serine deficiency
48471	Lissencephaly	
68335	Rare chromosomal anomaly	
68367	Rare inborn errors of metabolism	Rare metabolic disease
68373	Peroxisomal disease	
68402	Rare parkinsonian disorder	Rare hypokinetic movement disorder
79163	Classic organic aciduria	
79167	Disorder of urea cycle metabolism and ammonia detoxification	
79373	Ectodermal dysplasia syndrome	Ectodermal dysplasia
79386	Rare skin tumor or hamartoma	
87277	Rare intellectual disability	
90692	Rare endocrine growth disease	
93419	Rare bone disease	
93457	Non-syndromic limb reduction defect	Non-syndromic limb hypoplasia
93460	Overgrowth syndrome	
93547	Syndromic renal or urinary tract malformation	

93626	Rare renal disease	
93890	Rare developmental defect during embryogenesis	Malformation syndrome
96346	Anorectal malformation	
97245	Congenital myopathy	
97593	Pseudohypoparathyroidism	
97929	Rare cardiac disease	
97935	Rare gastroenterologic disease	
97966	Rare ophthalmic disorder	
97978	Rare endocrine disease	
98006	Rare neurologic disease	Rare nervous system disease
98033	Rare neurologic disease with psychiatric involvement	
98053	Rare genetic disease	
98127	Autosomal anomaly	
98158	Chromosome Y structural anomaly	
98258	Infantile epilepsy syndrome	
98473	Muscular dystrophy	
99739	Rare familial disorder with hypertrophic cardiomyopathy	Rare familial disorder with hypertrophic obstructive cardiomyopathy|Rare familial disorder with hypertrophic subaortic stenosis
100091	Adrenal/paraganglial tumor	
101998	Rare epilepsy	
102002	Rare ataxia	
102015	Autosomal recessive limb-girdle muscular dystrophy	Autosomal recessive limb-girdle muscular dystrophy
102369	Rare syndromic intellectual disability	
109007	Arthrogryposis syndrome	
140162	Inherited cancer-predisposing syndrome	
156601	Rare genetic hepatic disease	
165704	Non-syndromic urogenital tract malformation	
166472	Monogenic disease with epilepsy	
169355	Immunodeficiency syndrome with autoimmunity	
178025	Non-acquired combined pituitary hormone deficiencies without extra-pituitary malformations	
180772	Rare disease with autism	
181393	Growth hormone insensitivity syndrome	GHIS|Short stature due to a defect in growth hormone receptor or post-receptor pathway
181399	Rare hyperthyroidism	
181412	Adrenogenital syndrome	
182070	Rare neurodegenerative disease	
183518	Rare hereditary ataxia	
183757	Rare genetic intellectual disability	
183763	Rare genetic syndromic intellectual disability	
206656	Non-dystrophic myopathy	
207085	Qualitative or quantitative defects of dystrophin	Dystrophinopathy
217569	Hypertrophic cardiomyopathy	
217595	Syndrome associated with hypertrophic cardiomyopathy	
217598	Non-familial hypertrophic cardiomyopathy	
217604	Dilated cardiomyopathy	
217607	Familial dilated cardiomyopathy	

223713	Mitochondrial oxidative phosphorylation disorder	OXPHOS disease
261721	Anomaly of chromosome 9	
261748	Anomaly of chromosome 17	
261857	Partial deletion of the short arm of chromosome 1	Partial deletion of chromosome 1p|Partial monosomy of chromosome 1p|Partial monosomy of the short arm of chromosome 1
261866	Partial deletion of the short arm of chromosome 2	Partial deletion of chromosome 2p|Partial monosomy of chromosome 2p|Partial monosomy of the short arm of chromosome 2
261875	Partial deletion of the short arm of chromosome 3	Partial deletion of chromosome 3p|Partial monosomy of chromosome 3p|Partial monosomy of the short arm of chromosome 3
261902	Partial deletion of the short arm of chromosome 6	Partial deletion of chromosome 6p|Partial monosomy of chromosome 6p|Partial monosomy of the short arm of chromosome 6
261956	Partial deletion of the short arm of chromosome 16	Partial deletion of chromosome 16p|Partial monosomy of chromosome 16p|Partial monosomy of the short arm of chromosome 16
261974	Partial deletion of the short arm of chromosome 18	Partial deletion of chromosome 18p|Partial monosomy of chromosome 18p|Partial monosomy of the short arm of chromosome 18
261983	Partial deletion of the short arm of chromosome 19	Partial deletion of chromosome 19p|Partial monosomy of chromosome 19p|Partial monosomy of the short arm of chromosome 19
262010	Partial deletion of the long arm of chromosome 2	Partial deletion of chromosome 2q|Partial monosomy of chromosome 2q|Partial monosomy of the long arm of chromosome 2
262029	Partial deletion of the long arm of chromosome 4	Partial deletion of chromosome 4q|Partial monosomy of chromosome 4q|Partial monosomy of the long arm of chromosome 4
262056	Partial deletion of the long arm of chromosome 7	Partial deletion of chromosome 7q|Partial monosomy of chromosome 7q|Partial monosomy of the long arm of chromosome 7
262083	Partial monosomy of the long arm of chromosome 10	Partial deletion of chromosome 10q|Partial deletion of the long arm of chromosome 10|Partial monosomy of chromosome 10q
262092	Partial deletion of the long arm of chromosome 11	Partial deletion of chromosome 11q|Partial monosomy of chromosome 11q|Partial monosomy of the long arm of chromosome 11
262101	Partial deletion of the long arm of chromosome 13	Partial deletion of chromosome 13q|Partial monosomy of chromosome 13q|Partial monosomy of the long arm of chromosome 13

262110	Partial deletion of the long arm of chromosome 14	Partial deletion of chromosome 14q|Partial monosomy of chromosome 14q|Partial monosomy of the long arm of chromosome 14
262128	Partial deletion of the long arm of chromosome 16	Partial deletion of chromosome 16q|Partial monosomy of chromosome 16q|Partial monosomy of the long arm of chromosome 16
262137	Partial deletion of the long arm of chromosome 17	Partial deletion of chromosome 17q|Partial monosomy of chromosome 17q|Partial monosomy of the long arm of chromosome 17
262146	Partial deletion of the long arm of chromosome 18	Partial deletion of chromosome 18q|Partial monosomy of chromosome 18q|Partial monosomy of the long arm of chromosome 18
262173	Partial deletion of the long arm of chromosome 21	Partial deletion of chromosome 21q|Partial monosomy of chromosome 21q|Partial monosomy of the long arm of chromosome 21
262182	Partial deletion of the long arm of chromosome 22	Partial deletion of chromosome 22q|Partial monosomy of chromosome 22q|Partial monosomy of the long arm of chromosome 22
262643	Partial trisomy/tetrasomy of chromosome 9	Partial duplication/triplication of chromosome 9
262658	Partial trisomy/tetrasomy of the short arm of chromosome 12	Partial duplication/triplication of chromosome 12p|Partial duplication/triplication of the short arm of chromosome 12|Partial trisomy/tetrasomy of chromosome 12p
262698	Partial duplication of the short arm of chromosome 2	Partial duplication of chromosome 2p|Partial trisomy of chromosome 2p
262725	Partial trisomy/tetrasomy of the short arm of chromosome 5	Partial duplication/triplication of chromosome 5p|Partial duplication/triplication of the short arm of chromosome 5|Partial trisomy/tetrasomy of chromosome 5p
262794	Partial duplication of the short arm of chromosome 16	Partial duplication of chromosome 16p|Partial trisomy of chromosome 16p|Partial trisomy of the short arm of chromosome 16
262812	Partial trisomy/tetrasomy of the short arm of chromosome 18	Partial duplication/triplication of chromosome 18p|Partial duplication/triplication of the short arm of chromosome 18|Partial trisomy/tetrasomy of chromosome 18p
262833	Partial duplication of the long arm of chromosome 1	Partial duplication of chromosome 1q|Partial trisomy of chromosome 1q|Partial trisomy of the long arm of chromosome 1
262860	Partial duplication of the long arm of chromosome 4	Partial duplication of chromosome 4q|Partial trisomy of chromosome 4q|Partial trisomy of the long arm of chromosome 4
262869	Partial trisomy of the long arm of chromosome 5	Partial duplication of chromosome 5q|Partial duplication of the long arm of chromosome 5|Partial trisomy of chromosome 5q

262878	Partial duplication of the long arm of chromosome 6	Partial duplication of chromosome 6q|Partial trisomy of chromosome 6q|Partial trisomy of the long arm of chromosome 6
262887	Partial duplication of the long arm of chromosome 7	Partial duplication of chromosome 7q|Partial trisomy of chromosome 7q|Partial trisomy of the long arm of chromosome 7
262905	Partial trisomy of the long arm of chromosome 9	Partial duplication of chromosome 9q|Partial duplication of the long arm of chromosome 9|Partial trisomy of chromosome 9q
262932	Partial duplication of the long arm of chromosome 13	Partial duplication of chromosome 13q|Partial trisomy of chromosome 13q|Partial trisomy of the long arm of chromosome 13
262950	Partial duplication of the long arm of chromosome 15	Partial duplication of chromosome 15q|Partial trisomy of chromosome 15q|Partial trisomy of the long arm of chromosome 15
262959	Partial trisomy of the long arm of chromosome 16	Partial duplication of chromosome 16q|Partial duplication of the long arm of chromosome 16|Partial trisomy of chromosome 16q
262968	Partial duplication of the long arm of chromosome 17	Partial duplication of chromosome 17q|Partial trisomy of chromosome 17q|Partial trisomy of the long arm of chromosome 17
262977	Partial trisomy of the long arm of chromosome 18	Partial duplication of chromosome 18q|Partial duplication of the long arm of chromosome 18|Partial trisomy of chromosome 18q
263708	Complex chromosomal rearrangement	
263711	X chromosome anomaly	
263731	Partial monosomy of the short arm of chromosome X	Partial deletion of chromosome Xp|Partial deletion of the short arm of chromosome X|Partial monosomy of chromosome Xp
263756	Partial deletion of the long arm of chromosome X	Partial deletion of chromosome Xq|Partial monosomy of chromosome Xq|Partial monosomy of the long arm of chromosome X
263775	Partial duplication of the short arm of chromosome X	Partial duplication of chromosome Xp|Partial trisomy of chromosome Xp|Partial trisomy of the short arm of chromosome X
263783	Partial duplication of the long arm of chromosome X	Partial duplication of chromosome Xq|Partial trisomy of chromosome Xq|Partial trisomy of the long arm of chromosome X
281217	Autosomal ichthyosis syndrome	
284993	Marfan and Marfan-related disorders	
289899	Organic aciduria	
290842	Autoinflammatory syndrome with skin involvement	
294057	Rare nevus	
294953	Non syndromic limb overgrowth	
307061	Rare genetic tremor disorder	
363294	Genetic syndromic Pierre Robin syndrome	

370106	Rare disorder with dystonia and other neurologic or systemic manifestation	
399846	Rare disorder with female infertility due to a congenital hypogonadotropic hypogonadism	
447985	Partial duplication of the short arm of chromosome 19	Partial duplication of chromosome 19p|Partial trisomy of chromosome 19p|Partial trisomy of the short arm of chromosome 19
466658	Rare disease with malignant hyperthermia	
477759	COL4A1 or COL4A2-related cerebral small vessel disease	COL4A1 or COL4A2-related cerebral angiopathy
477811	Rare hypercholesterolemia	
485382	Genetic non-acquired premature ovarian failure	
498477	Ectrodactyly with and without other manifestations	

Clinical syndrome

1799	Familial developmental dysphasia	Billard-Toutain-Maheut syndrome|FOXP2-associated dysphasia
1934	Early infantile epileptic encephalopathy	EIEE|Early infantile epileptic encephalopathy with suppression-bursts|Ohtahara syndrome

Clinical group

19	2-hydroxyglutaric aciduria	2-hydroxyglutaric acidemia
55	Oculocutaneous albinism	OCA
247	Arrhythmogenic right ventricular cardiomyopathy	ARVC|ARVD|Arrhythmogenic right ventricular dysplasia
251	Multiple epiphyseal dysplasia	EDM|MED|Polyepiphyseal dysplasia
254	Spondylometaphyseal dysplasia	
262	Duchenne and Becker muscular dystrophy	Severe dystrophinopathy, Duchenne and Becker type
282	Frontotemporal dementia	FTD
303	Dystrophic epidermolysis bullosa	DEB|Dermolytic epidermolysis bullosa|Epidermolysis bullosa dystrophica
304	Epidermolysis bullosa simplex	EBS|EEB
418	Congenital adrenal hyperplasia	CAH
607	Nemaline myopathy	NEM|NM|Nemaline rod myopathy
685	Hereditary spastic paraplegia	Familial spastic paraplegia|HSP|Hereditary spastic paraparesis|SPG|Strümpell-Lorrain disease
768	Familial long QT syndrome	Congenital long QT syndrome
823	Isolated spina bifida	
1505	Short rib-polydactyly syndrome	
2014	Cleft palate	
3280	Syringomyelia	Hydromyelia
64747	X-linked Charcot-Marie-Tooth disease	CMTX|X-linked hereditary motor and sensory neuropathy
65753	Charcot-Marie-Tooth disease type 1	Autosomal dominant demyelinating Charcot-Marie-Tooth disease|CMT1|Charcot-Marie-Tooth neuropathy type 1|Hereditary motor and sensory neuropathy type 1

91024	Autosomal recessive axonal hereditary motor and sensory neuropathy	AR-CMT2|Autosomal recessive axonal Charcot-Marie-Tooth disease type 2
93442	Chondrodysplasia punctata	CDP
93573	Thrombotic microangiopathy	
97556	Congenital and infantile nephrotic syndrome	
98249	Ehlers-Danlos syndrome	
98306	Familial partial lipodystrophy	FPLD
98672	Autosomal dominant optic atrophy	ADOA|DOA
102009	Classic lissencephaly	Lissencephaly type 1
155867	Paramedian facial cleft	Tessier number 1-1 and 2-12 facial cleft
165985	Diazoxide-sensitive diffuse hyperinsulinism	Hyperinsulinemic hypoglycemia, diazoxide-sensitive diffuse form
166311	Benign partial infantile seizures	
206647	Myotonic dystrophy	
206973	Congenital myotonia	
238666	Isolated congenital hypogonadotropic hypogonadism	
248095	Primary hypertrophic osteoarthropathy	Idiopathic hypertrophic osteoarthropathy|PHO
254837	Unspecified mitochondrial disorder	
271861	Hereditary ATTR amyloidosis	Familial TTR-related amyloidosis|Familial transthyretin-related amyloidosis
281097	Autosomal recessive congenital ichthyosis	ARCI
281210	X-linked ichthyosis syndrome	
306498	PTEN hamartoma tumor syndrome	PHTS
320335	Pure or complex hereditary spastic paraplegia	Pure or complex familial spastic paraplegia|Pure or complicated familial spastic paraplegia|Pure or complicated hereditary spastic paraplegia
435365	Fetal lower urinary tract obstruction	LUTO
443909	Hereditary nonpolyposis colon cancer	Familial nonpolyposis colon cancer|Familial nonpolyposis colorectal cancer|HNPCC|Hereditary nonpolyposis colorectal cancer
530313	PIK3CA-related overgrowth syndrome	PROS
544458	Hemolytic uremic syndrome	HUS

Clinical subtype

47	Isolated agammaglobulinemia	BTK-deficiency|Bruton type agammaglobulinemia
383	Non-syndromic genetic deafness	Conductive deafness with stapes fixation|DFNX2|Nance deafness|X-linked deafness type 2|X-linked mixed conductive and neurosensory deafness|X-linked mixed conductive and neurosensory hearing loss|X-linked mixed conductive and sensorineural deafness|X-linked mixed conductive and sensorineural hearing loss|X-linked stapes gusher syndrome
1018	X-linked Alport syndrome-diffuse leiomyomatosis	Xq22.3 microdeletion syndrome
1860	Thanatophoric dysplasia	TD1|Thanatophoric dwarfism type 1
2028	Hyaline fibromatosis syndrome	Murray-Puretic-Drescher syndrome|Puretic syndrome

2578	Mayer-Rokitansky-Küster-Hauser syndrome	Atypical MRKH syndrome|MRKH syndrome type 2|MURCS association|Müllerian duct aplasia-renal dysplasia-cervical somite anomalies syndrome
30925	Central diabetes insipidus	Hereditary CDI|Hereditary neurogenic diabetes insipidus
77259	Gaucher disease	Non-cerebral juvenile Gaucher disease
79253	Phenylketonuria	Mild PKU|Variant PKU|Variant phenylketonuria|mPKU
79254	Phenylketonuria	Classic PKU
79255	GM1 gangliosidosis	Infantile GM1 gangliosidosis|Norman-Landing disease
79282	Methylmalonic acidemia with homocystinuria	CblC defect|Cobalamin C defect|Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cblC|Methylmalonic aciduria with homocystinuria, type cblC
79305	Progressive familial intrahepatic cholestasis	PFIC3
79651	Phenylketonuria	Mild HPA|Non-PKU HPA|mHPA
83330	Proximal spinal muscular atrophy	Infantile spinal muscular atrophy|SMA type 1|SMA type I|SMA-I|SMA1|Werdnig-Hoffmann disease
88917	Alport syndrome	
88918	Alport syndrome	
88919	Alport syndrome	
88949	Autosomal dominant tubulointerstitial kidney disease	ADTKD-MUC1|MCKD1|MUC1-related autosomal dominant medullary cystic kidney disease|MUCI-related ADTKD|Medullary cystic kidney disease type 1
90308	Angioosteohypertrophic syndrome	
90653	Stickler syndrome	
90654	Stickler syndrome	
93100	Renal agenesis	
93328	Omodysplasia	
93473	Mucopolysaccharidosis type 1	Hurler disease|MPS1H|MPSIH|Mucopolysaccharidosis type 1H|Mucopolysaccharidosis type IH
93598	Primary hyperoxaluria	Glycolic aciduria|Peroxisomal alanine-glyoxylate aminotransferase deficiency
93600	Primary hyperoxaluria	
93616	Alpha-thalassemia	Alpha-thalassemia intermedia|HbH disease
93622	Dent disease	Nephrolithiasis type 1
93623	Dent disease	Nephrolithiasis type 2
93929	Exstrophy-epispadias complex	OEIS complex|Omphalocele-cloacal exstrophy-imperforate anus-spinal defect syndrome
98892	Nodular neuronal heterotopia	PVNH
98991	Early-onset non-syndromic cataract	
99361	Multiple endocrine neoplasia type 2	Familial MTC
99858	Primary syringomyelia	
101070	Bilateral polymicrogyria	
101088	Hyper-IgM syndrome with susceptibility to opportunistic infections	HIGM1|Hyper-IgM syndrome due to CD40 ligand deficiency|Hyper-IgM syndrome due to CD40L deficiency|Hyper-IgM syndrome type 1|XHIGM

139396	X-linked adrenoleukodystrophy	X-CALD
139399	X-linked adrenoleukodystrophy	
166260	Dentinogenesis imperfecta	Capdepont teeth|DGI-2|DI-2|Dentinogenesis imperfecta, Shields type 2
171871	Pseudohypoaldosteronism type 1	Autosomal dominant pseudohypoaldosteronism type 1
171876	Pseudohypoaldosteronism type 1	Autosomal recessive pseudohypoaldosteronism type 1
216445	Non-syndromic genetic deafness	Isolated prelingual genetic deafness
216452	Non-syndromic genetic deafness	Isolated postlingual genetic deafness
216796	Osteogenesis imperfecta	Adair-Dighton syndrome|Mild osteogenesis imperfecta|Non-deforming osteogenesis imperfecta|OI type 1|Van der Hoeve syndrome
216812	Osteogenesis imperfecta	OI type 3|Progressive deforming osteogenesis imperfecta|Severe osteogenesis imperfecta
216820	Osteogenesis imperfecta	OI type 4
216828	Osteogenesis imperfecta	OI type 5
216866	Pantothenate kinase-associated neurodegeneration	NBIA1, classic form|Neurodegeneration with brain iron accumulation type 1, classic form|PKAN, classic form
216981	Niemann-Pick disease type C	Niemann-Pick disease type C, classic form
217085	Mucopolysaccharidosis type 2	Hunter syndrome type A|Iduronate 2-sulfatase deficiency type A|MPS2A|MPSIIA|Mucopolysaccharidosis type 2A|Mucopolysaccharidosis type II, severe form|Mucopolysaccharidosis type IIA
228302	Carnitine palmitoyltransferase II deficiency	CPT2, adult-onset form|CPT2, myopathic form|CPTII, adult-onset form|CPTII, myopathic form|Carnitine palmitoyl transferase II deficiency, adult-onset form|Carnitine palmitoyl transferase deficiency type 2, adult-onset form|Carnitine palmitoyl transferase deficiency type 2, myopathic form
231169	Usher syndrome	USH1
231178	Usher syndrome	USH2
231512	Hermansky-Pudlak syndrome	HPS without pulmonary fibrosis
231679	Non-acquired isolated growth hormone deficiency	Congenital IGHD type II|Congenital isolated GH deficiency type II|Congenital isolated growth hormone deficiency type II
247667	Hypophosphatasia	Childhood-onset Rathburn disease|Childhood-onset phosphoethanolaminuria
247698	Multiple endocrine neoplasia type 2	MEN2A|PTC syndrome|Sipple syndrome
247798	Attenuated familial adenomatous polyposis	MUTYH-related AFAP|MUTYH-related attenuated FAP|MUTYH-related attenuated familial polyposis coli
247806	Attenuated familial adenomatous polyposis	APC-related AFAP|APC-related attenuated FAP|APC-related attenuated familial polyposis coli
250984	Stickler syndrome	
252018	Extragonadal teratoma	

255182	Pyruvate dehydrogenase deficiency	2-oxoglutarate complex deficiency|Branched chain alpha-ketoacid dehydrogenase complex deficiency|Diaphorase deficiency|Dihydrolipoyl dehydrogenase deficiency|Glycine cleavage system L protein deficiency|Lipoamide dehydrogenase deficiency|Pyruvate dehydrogenase complex component E3 deficiency|Pyruvate dehydrogenase protein X component deficiency
268261	DYRK1A-related intellectual disability syndrome	21q22.13q22.2 microdeletion syndrome|Del(21)(q22.13q22.2)|Monosomy 21q22.13q22.2
280229	Pelizaeus-Merzbacher disease	
284343	Pleuropulmonary blastoma	DICER1 syndrome|PPB familial tumor susceptibility syndrome|PPBFTDS|Pleuro-pulmonary blastoma familial tumor susceptibility syndrome
284408	Isolated glycerol kinase deficiency	
284963	Marfan syndrome	MFS1
293910	Familial isolated arrhythmogenic right ventricular dysplasia	Familial isolated arrhythmogenic ventricular cardiomyopathy, classic form|Familial isolated arrhythmogenic ventricular cardiomyopathy, right dominant form|Familial isolated arrhythmogenic ventricular dysplasia, classic form
309185	Tay-Sachs disease	GM2 gangliosidosis, B variant, juvenile form|Hexosaminidase A deficiency, juvenile form
309282	Alpha-mannosidosis	Lysosomal alpha-D-mannosidase deficiency, infantile form
309297	Mucopolysaccharidosis type 4	GALNS deficiency|Galactosamine-6-sulfatase deficiency|MPS4A|MPSIVA|Morquio disease type A|Mucopolysaccharidosis type IVA|N-acetylgalactosamine-6-sulfate sulfatase deficiency
315306	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Classic 21-OHD CAH, salt wasting form
315311	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Classic 21-OHD CAH, simple virilizing form
329918	Primary membranoproliferative glomerulonephritis	Non-Ig-mediated MPGN|Non-Ig-mediated membranoproliferative glomerulonephritis|Non-immunoglobulin-mediated MPGN|Non-immunoglobulin-mediated membranoproliferative glomerulonephritis
353284	Rubinstein-Taybi syndrome	
391646	Feingold syndrome	Brachydactyly-short stature-microcephaly syndrome|Brunner-Winter syndrome type 2|FGLDS2|FS2|MMT type 2|Microcephaly-digital anomalies-normal intelligence syndrome type 2|Microcephaly-intellectual disability-tracheoesophageal fistula syndrome type 2
402041	Distal renal tubular acidosis	AR dRTA|Autosomal recessive distal RTA

454840	Attenuated familial adenomatous polyposis	NTHL1-related AFAP|NTHL1-related attenuated FAP
464311	DYRK1A-related intellectual disability syndrome	DYRK1A-related intellectual disability syndrome due to a point mutation
466950	WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	
556030	Familial hypoaldosteronism	Early-onset familial hyperreninemic hypoaldosteronism|Severe aldosterone synthase deficiency

Malformation syndrome

8	47,XYY syndrome	Double Y syndrome|XYY syndrome|Y disomy
40	Acromesomelic dysplasia, Maroteaux type	
52	Alagille syndrome	Alagille-Watson syndrome|Arteriohepatic dysplasia|Syndromic bile duct paucity
53	Albers-Schönberg osteopetrosis	Osteopetrosis autosomal dominant type 2
72	Angelman syndrome	
84	Fanconi anemia	Fanconi pancytopenia
87	Apert syndrome	ACS1|Acrocephalosyndactyly type 1
107	BOR syndrome	Branchiootorenal syndrome
109	Bannayan-Riley-Ruvalcaba syndrome	BRRS|Myhre-Riley-Smith syndrome
115	Congenital contractural arachnodactyly	Beals syndrome|Beals-Hecht syndrome|CCA syndrome|Distal arthrogryposis type 9
116	Beckwith-Wiedemann syndrome	BWS|Exomphalos-macroglossia-gigantism syndrome|Wiedemann-Beckwith syndrome
122	Birt-Hogg-Dubé syndrome	Fibrofolliculomas with trichodiscomas and acrochordons
126	Blepharophimosis-epicanthus inversus-ptosis syndrome	Blepharophimosis types 1 and 2
127	Borjeson-Forssman-Lehmann syndrome	BFLS|Intellectual disability-epilepsy-endocrine disorders syndrome
138	CHARGE syndrome	CHARGE association|Coloboma-heart defects-atresia choanae-retardation of growth and development-genitourinary problems-ear abnormalities syndrome|Hall-Hittner syndrome
140	Campomelic dysplasia	Campomelic dwarfism
162	Cataract-glaucoma syndrome	
192	Coffin-Lowry syndrome	CLS
193	Cohen syndrome	
195	Cat-eye syndrome	CES
199	Cornelia de Lange syndrome	Brachmann-de Lange syndrome
207	Crouzon disease	Crouzon craniofacial dysostosis
233	Duane retraction syndrome	DRS|DURS|Duane syndrome|Stilling-Turk-Duane syndrome
236	Trisomy 9p	Duplication 9p|Duplication of the short arm of chromosome 9|Trisomy of the short arm of chromosome 9
240	Léri-Weill dyschondrosteosis	Léri-Weill syndrome
249	Fibrous dysplasia of bone	
280	Wolf-Hirschhorn syndrome	4p- syndrome|Distal deletion 4p|Distal monosomy 4p|Telomeric deletion 4p
281	Monosomy 5p	Cri du chat syndrome|Deletion 5p
289	Ellis Van Creveld syndrome	Chondroectodermal dysplasia|Mesodermic dysplasia

308	Unverricht-Lundborg disease	PME type 1|Progressive myoclonic epilepsy type 1|Progressive myoclonus epilepsy type 1|ULD|||
373	Simpson-Golabi-Behmel syndrome	DGSX|Golabi-Rosen syndrome|SDYS|SGBS|SGBS1|Simpson dysmorphia syndrome|Simpson-Golabi-Behmel syndrome type 1|X-linked dysplasia gigantism syndrome
374	Goldenhar syndrome	Expanded spectrum hemifacial microsomia|Facioauriculovertebral dysplasia|OAV dysplasia|OAVS|Oculoauriculovertebral dysplasia|Oculoauriculovertebral syndrome||Anti-GBM syndrome|Goodpasture syndrome
377	Gorlin syndrome	Basal cell nevus syndrome|Gorlin-Goltz syndrome|NBCCS|Nevoid basal cell carcinoma syndrome
380	Greig cephalopolysyndactyly syndrome	GCPS
392	Holt-Oram syndrome	Atriodigital dysplasia type 1|HOS|Heart-hand syndrome type 1
464	Incontinentia pigmenti	Bloch-Siemens syndrome|Bloch-Sulzberger syndrome||Congenital PAI-1 deficiency
474	Jeune syndrome	Asphyxiating thoracic dystrophy of the newborn|JATD|Jeune asphyxiating thoracic dystrophy
475	Joubert syndrome	CPD IV|Cerebelloparenchymal disorder IV|Classic Joubert syndrome|Joubert syndrome type A|Joubert-Boltshauser syndrome|Pure Joubert syndrome
500	Noonan syndrome with multiple lentigines	Cardiomyopathic lentiginosis|Familial multiple lentigines syndrome|LEOPARD syndrome
502	Trichorhinophalangeal syndrome type 2	Deletion 8q24.1|Langer-Giedion syndrome|Monosomy 8q24.1
534	Oculocerebrorenal syndrome of Lowe	Lowe disease|Lowe oculo-cerebro-renal syndrome|Lowe syndrome|OCR|OCRL|Oculo-cerebro-renal dystrophy|Oculo-cerebro-renal syndrome|Oculocerebrorenal dystrophy|Phosphatidylinositol 4,5-biphosphate 5-phosphatase deficiency
567	22q11.2 deletion syndrome	22q11DS|CATCH 22|Cayler cardiofacial syndrome|Conotruncal anomaly face syndrome|DiGeorge sequence|DiGeorge syndrome|Microdeletion 22q11.2|Monosomy 22q11|Sedlackova syndrome|Shprintzen syndrome|Takao syndrome|Velocardiofacial syndrome
627	Nance-Horan syndrome	
640	Hereditary neuropathy with liability to pressure palsies	Current pressure-sensitive neuropathy|HNPP|Heterozygous microdeletion 17p11.2p12|Potato-grubbing palsy|Tomaculous neuropathy|Tulip-bulb digger's palsy
648	Noonan syndrome	
705	Pendred syndrome	Goiter-deafness syndrome

709	Peters plus syndrome	Krause-Kivlin syndrome|Krause-van Schooneveld-Kivlin syndrome|Peters anomaly with short limb dwarfism
710	Pfeiffer syndrome	ACS5|Acrocephalosyndactyly type 5
718	Isolated Pierre Robin syndrome	Isolated Pierre Robin sequence
744	Proteus syndrome	Partial gigantism-nevi-hemihypertrophy-macrocephaly syndrome
782	Axenfeld-Rieger syndrome	Axenfeld syndrome|Rieger syndrome
783	Rubinstein-Taybi syndrome	Broad thumb-hallux syndrome|Broad thumbs-halluces syndrome
792	X-linked retinoschisis	X-linked juvenile retinoschisis|XLRS
794	Saethre-Chotzen syndrome	ACS3|Acrocephalosyndactyly type 3|SCS
818	Smith-Lemli-Opitz syndrome	7-dehydrocholesterol reductase deficiency|RSH syndrome|SLOS
819	Smith-Magenis syndrome	17p11.2 microdeletion syndrome
857	Townes-Brocks syndrome	Imperforate anus-hand, foot and ear anomalies syndrome|REAR syndrome|Renal-ear-anal-radial syndrome|Sensorineural deafness with imperforate anus and hypoplastic thumbs|TBS|Townes syndrome
861	Treacher-Collins syndrome	Franceschetti-Klein syndrome|Mandibulofacial dysostosis without limb anomalies
870	Down syndrome	Trisomy 21
881	Turner syndrome	45,X syndrome|45,X/46,XX syndrome
884	Tetrasomy 12p	Isochromosome 12p mosaicism|Isochromosome 12p syndrome|Pallister-Killian syndrome
887	VACTERL/VATER association	VACTERL association|VATER association
888	Van der Woude syndrome	Cleft lip/palate with mucous cysts of lower lip|Lip-pit syndrome|VWS
893	WAGR syndrome	Del(11)(p13)|Deletion 11p13|Monosomy 11p13|Wilms tumor-aniridia-genitourinary anomalies-intellectual disability syndrome
904	Williams syndrome	Deletion 7q11.23|Monosomy 7q11.23|Williams-Beuren syndrome
908	Fragile X syndrome	FRAXA syndrome|FXS|FraX syndrome|Martin-Bell syndrome
915	Aarskog-Scott syndrome	Aarskog syndrome|Faciodigitogenital syndrome|Faciogenital dysplasia
955	Acroosteolysis dominant type	Acrodentoosteodysplasia|Acroosteolysis with osteoporosis and changes in skull and mandible|Arthrodentoosteodysplasia|Cheney syndrome|Hajdu-Cheney syndrome
959	Acro-renal-ocular syndrome	
974	Adams-Oliver syndrome	AOS|Congenital scalp defects with distal limb anomalies|Congenital scalp defects with distal limb reduction anomalies|Limb, scalp and skull defects
1001	2q37 microdeletion syndrome	Albright hereditary osteodystrophy type 3|Albright hereditary osteodystrophy-like syndrome|Brachydactyly-intellectual disability syndrome|Del(2)(q37)|Deletion 2q37|Monosomy 2q37qter
1031	Enamel-renal syndrome	Amelogenesis imperfecta-nephrocalcinosis syndrome

1114	Aplasia cutis congenita	
1190	Atelosteogenesis type I	AO1|AOI|Atelosteogenesis type 1|Giant cell chondrodysplasia|Spondylo-humero-femoral dysplasia
1276	Brachydactyly-arterial hypertension syndrome	Bilginturan brachydactyly|Bilginturan syndrome|Brachydactyly type E, with short stature and hypertension
1305	Feingold syndrome	Brunner-Winter syndrome|Digital anomalies with short palpebral fissures and atresia of esophagus or duodenum|FGLDS|FS|MMT|MODED syndrome|Microcephaly-digital anomalies-normal intelligence syndrome|Microcephaly-intellectual disability-tracheoesophageal fistula syndrome|Microcephaly-intellectual disability-tracheoesophageal fistula syndrome|ODED syndrome|Oculo-digito-esophageal-duodenal syndrome
1306	Buschke-Ollendorff syndrome	Disseminated dermatofibrosis with osteopoikilosis
1328	Camurati-Engelmann disease	Progressive diaphyseal dysplasia
1340	Cardiofaciocutaneous syndrome	CFC syndrome
1425	Desbuquois syndrome	DBQD|Desbuquois dysplasia
1442	Ring chromosome 18 syndrome	Ring 18|Ring chromosome 18
1444	Ring chromosome 20 syndrome	Ring 20|Ring chromosome 20
1446	Ring chromosome 22 syndrome	Ring 22|Ring chromosome 22|r(22) syndrome
1452	Cleidocranial dysplasia	Cleidocranial dysostosis
1465	Coffin-Siris syndrome	CSS
1520	Craniofrontonasal dysplasia	CFND|CFNS|Craniofrontonasal syndrome
1522	Craniometaphyseal dysplasia	
1552	Currarino syndrome	Currarino triad
1570	Symbrachydactyly of hands and feet	De Smet-Fabry-Fryns syndrome
1571	Knobloch syndrome	Knobloch-Layer syndrome|Retinal detachment-occipital encephalocele syndrome
1590	Distal monosomy 13q	13q32 deletion|Deletion 13q32|Distal 13q deletion|Monosomy 13q32|Telomeric deletion13q
1600	Monosomy 18q	18q deletion syndrome|18q-syndrome|Deletion 18q
1606	1p36 deletion syndrome	Del(1)(p36)|Deletion 1p36|Deletion 1pter|Monosomy 1p36|Monosomy 1pter|Subtelomeric 1p36 deletion
1636	Distal monosomy 7q36	Distal deletion 7q36|Monosomy 7qter|Telomeric deletion 7q36
1642	Distal monosomy 9p	Distal deletion 9p|Monosomy 9pter|Telomeric deletion 9p
1708	Mosaic trisomy 16	Mosaic trisomy chromosome 16|Trisomy 16 mosaicism
1713	17p11.2 microduplication syndrome	Potocki-Lupski syndrome|Trisomy 17p11.2
1717	Distal trisomy 19q	Distal duplication 19q|Telomeric duplication 19q|Trisomy 19qter
1727	22q11.2 microduplication syndrome	Dup(22)(q11)|Duplication 22q11.2|Trisomy 22q11.2
1756	Caudal duplication	Dipygus|Split notochord syndrome
1762	Trisomy Xq28	Distal duplication Xq|Telomeric duplication Xq

1797	Autosomal dominant spondylocostal dysostosis	Autosomal dominant spondylocostal dysplasia
1885	Isolated ectopia lentis	Ectopia lentis syndrome|Familial ectopia lentis
1892	Ectrodactyly-polydactyly syndrome	
1906	Fetal valproate syndrome	Fetal valproic acid syndrome
1915	Fetal alcohol syndrome	ARBD|ARND|Alcohol-related birth defects|Alcohol-related neurodevelopmental disorder|FAS|FASD|Fetal alcohol spectrum disorders
2019	Femur-fibula-ulna complex	FFU complex|Femur-fibula-ulna dysostosis|Femur-fibula-ulna syndrome|PFFD
2044	Floating-Harbor syndrome	
2065	Galloway-Mowat syndrome	Galloway syndrome|Microcephaly-hiatus hernia-nephrotic syndrome|Nephrosis-neuronal dysmigration syndrome
2108	Hallermann-Streiff syndrome	François dyscephalic syndrome|Oculomandibulofacial syndrome
2152	Mowat-Wilson syndrome	Hirschsprung disease-intellectual disability syndrome
2237	Hypoparathyroidism-sensorineural deafness-renal disease syndrome	Barakat syndrome|HDR syndrome
2254	Pontocerebellar hypoplasia type 1	Norman disease|PCH1
2322	Kabuki syndrome	Kabuki make-up syndrome|Niikawa-Kuroki syndrome
2332	KBG syndrome	Short stature-facial and skeletal anomalies-intellectual disability-macrodontia syndrome
2345	Isolated Klippel-Feil syndrome	Congenital cervical vertebral fusion|Congenital fused cervical segments|Klippel-Feil malformation|Klippel-Feil sequence
2412	Dislocation of the hip-dysmorphism syndrome	Collins-Pope syndrome
2440	Isolated split hand-split foot malformation	Ectrodactyly|SHFM|Split hand foot malformation
2499	Metachondromatosis	
2526	Microcephaly-lymphedema-chorioretinopathy syndrome	MLCRD
2588	Myhre syndrome	Facial dysmorphism-intellectual disability-short stature-hearing loss syndrome
2614	Nail-patella syndrome	Onychoosteodysplasia|Turner-Kieser syndrome
2616	3M syndrome	3-M syndrome|Dolichospondylic dysplasia|Gloomy face syndrome|Le Merrer syndrome|Yakut short stature syndrome
2637	Microcephalic osteodysplastic primordial dwarfism type II	MOPD type II|Majewski osteodysplastic primordial dwarfism type II
2662	Keipert syndrome	Nasodigitoacoustic syndrome
2701	Noonan syndrome-like disorder with loose anagen hair	NS/LAH|Tosti syndrome
2707	Oculocerebrofacial syndrome, Kaufman type	
2710	Oculodentodigital dysplasia	Meyer-Schwickerath syndrome|ODDD syndrome|Oculodentoosseous dysplasia

2712	Oculofaciocardiodental syndrome	Cataract-microphthalmia-radiculomegaly-cardiac septal defect syndrome|OFCD syndrome
2750	Orofaciodigital syndrome type 1	OFD1|OFDI|OFDSI|Oral-facial-digital syndrome type 1|Papillon-Léage-Psaume syndrome
2771	Bruck syndrome	Osteogenesis imperfecta-congenital joint contractures syndrome
2856	Persistent Müllerian duct syndrome	PMDS|Persistent Müllerian derivatives
2886	TARP syndrome	Pierre Robin sequence-congenital heart defect-talipes syndrome|Pierre Robin syndrome-congenital heart defect-talipes syndrome|Talipes equinovarus-atrial septal defect-Robin sequence-persistence of the left superior vena cava syndrome
2896	Pitt-Hopkins syndrome	
2911	Poland syndrome	Poland anomaly|Poland sequence
2924	Isolated polycystic liver disease	ADPCLD|Autosomal dominant polycystic liver disease|PCLD
2990	Autosomal recessive multiple pterygium syndrome	Autosomal recessive non-lethal multiple pterygium syndrome|EVMPS|Escobar syndrome|Escobar variant multiple pterygium syndrome
2995	Baraitser-Winter cerebrofrontofacial syndrome	
3027	Caudal regression sequence	Caudal dysplasia|Sacral agenesis syndrome|Sacral regression syndrome
3047	Blepharophimosis-intellectual disability syndrome, SBBYS type	Hypothyroidism-dysmorphism-postaxial polydactyly-intellectual disability syndrome|SBBYSS|Say-Barber-Biesecker-Young-Simpson syndrome
3051	Intellectual disability-sparse hair-brachydactyly syndrome	Nicolaides-Baraitser syndrome
3071	Costello syndrome	FCS syndrome|Faciocutaneoskeletal syndrome
3103	Roberts syndrome	Pseudothalidomide syndrome|Roberts-SC phocomelia syndrome|SC phocomelia|SC pseudothalidomide syndrome
3138	Ulnar-mammary syndrome	Pallister ulnar-mammary syndrome|Schinzel syndrome|UMS
3157	Septo-optic dysplasia spectrum	De Morsier syndrome|SOD|Septo-optic dysplasia
3205	Sturge-Weber syndrome	Encephalofacial angiomatosis|Encephalotrigeminal angiomatosis|SWS|Sturge-Weber-Dimitri syndrome|Sturge-Weber-Krabbe angiomatosis|Sturge-Weber-Krabbe syndrome
3206	Stüve-Wiedemann syndrome	Neonatal Schwartz-Jampel syndrome|SJS2|Schwartz-Jampel syndrome type 2|Stüve-Wiedemann dysplasia
3238	Cardiospondylocarpofacial syndrome	Forney syndrome|Forney-Robinson-Pascoe syndrome|Mitral regurgitation-deafness-skeletal anomalies syndrome
3275	Spondylocarpotarsal synostosis	Synspondylism
3306	Duplication/inversion 15q11	Invdup(15)|Isodicentric 15 chromosome|Non-distal tetrasomy 15q|Non-telomeric tetrasomy 15q|idic(15)
3307	Tetrasomy 18p	Isochromosome 18p

3309	Tetrasomy 5p	Isochromosome 5p
3310	Tetrasomy 9p	Isochromosome 9p
3329	Tibial aplasia-ectrodactyly syndrome	Aplasia of tibia with split-hand/split-foot deformity|SHFLD syndrome|SHFM associated with aplasia of long bones|Split hand/foot malformation with long bone deficiency|Split-hand/foot malformation associated with aplasia of long bones|TH-SHFM|Tibial hemimelia with split hand/foot malformation|Tibial hemimelia-ectrodactyly syndrome
3339	Toriello-Lacassie-Droste syndrome	Aplasia cutis congenita-epibulbar dermoids syndrome|Oculoectodermal syndrome
3342	Arterial tortuosity syndrome	ATS
3375	Trisomy X	47,XXX syndrome|Triple X syndrome|Triplo-X syndrome|XXX syndrome
3377	Trismus-pseudocamptodactyly syndrome	Distal arthrogryposis type 7|Dutch-Kentucky syndrome|Hecht syndrome|Hecht-Beals syndrome
3378	Trisomy 13	Patau syndrome
3454	Intellectual disability-developmental delay-contractures syndrome	Foot contractures-muscle atrophy-oculomotor apraxia syndrome|Wieacker-Wolff syndrome
36367	Distal monosomy 1q	Distal deletion 1q|Monosomy 1qter|Telomeric deletion 1q
42775	PHACE syndrome	Pascual-Castroviejo syndrome type 2
48652	Monosomy 22q13	22q13 deletion|Phelan-McDermid syndrome
60030	Loeys-Dietz syndrome	Aortic aneurysm syndrome due to TGF-beta receptors anomalies
60040	Megalencephaly-capillary malformation-polymicrogyria syndrome	MCAP|MCM|MCMTC|Macrocephaly-capillary malformation syndrome|Macrocephaly-cutis marmorata telangiectatica congenita syndrome|Megalencephaly-capillary malformation syndrome|Megalencephaly-cutis marmorata telangiectatica congenita syndrome
65286	3q29 microdeletion syndrome	3q subtelomere deletion syndrome|3qter deletion|Del(3)(q29)|Monosomy 3q29|Monosomy 3qter
75857	6q terminal deletion syndrome	
77258	Trichorhinophalangeal syndrome type 1 and 3	
79113	Mandibulofacial dysostosis-microcephaly syndrome	MFDM syndrome|Mandibulofacial dysostosis, Guion-Almeida type
79490	Microcystic lymphatic malformation	Capillary lymphangioma|Capillary lymphatic malformation|Cutaneous lymphangioma circumscriptum|Microcystic infiltrating lymphatic malformation|Microcystic lymphangioma|Superficial lymphangioma|Superficial lymphatic malformation

83454	Glomuvenous malformation	Glomangiomatosis|Hereditary multiple glomangiomas|Multiple glomus tumors|VMGLOM|Venous malformations with glomus cells
85193	Idiopathic juvenile osteoporosis	IJO|Juvenile osteoporosis
85200	Ischiovertebral syndrome	Ischiospinal dysostosis|Ischiovertebral dysplasia
85279	Syndromic X-linked intellectual disability due to JARID1C mutation	
85287	X-linked intellectual disability, Siderius type	
85328	X-linked intellectual disability, Turner type	
85329	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome	
93256	Fragile X-associated tremor/ataxia syndrome	FXTAS syndrome
93304	Autosomal dominant brachyolmia	Brachyolmia type 3
93383	Brachydactyly type B	
93388	Brachydactyly type A1	Brachydactyly, Farabee type
96061	Mosaic trisomy 8	Mosaic trisomy chromosome 8|Trisomy 8 mosaicism
96072	4p16.3 microduplication syndrome	Distal duplication 4p|Distal trisomy 4p|Telomeric duplication 4p|Trisomy 4pter
96096	Distal trisomy 4q	Distal duplication 4q|Telomeric duplication 4q|Trisomy 4qter
96102	Distal trisomy 10q	Distal duplication 10q|Telomeric duplication 10q|Trisomy 10qter
96121	7q11.23 microduplication syndrome	Dup(7)(q11.23)|Trisomy 7q11.23
96125	Distal monosomy 6p	6p subtelomeric deletion syndrome|6p25 microdeletion syndrome|Distal deletion 6p|Monosomy 6p25
96129	Distal monosomy 19p13.3	Distal deletion 19p|Telomeric deletion 19p
96145	Distal monosomy 4q	Distal deletion 4q|Monosomy 4qter|Telomeric deletion 4q
96148	Distal monosomy 10q	Distal deletion 10q|Monosomy 10qter|Telomeric deletion 10q
96168	Monosomy 13q34	Del(13)(q34)|Distal deletion 13q34|Subtelomeric deletion 13q34
96169	Koolen-De Vries syndrome	KdVS
96170	Emanuel syndrome	Der(22)t(11;22) syndrome|Supernumerary der(22) syndrome
96181	Maternal uniparental disomy of chromosome 6	UPD(6)mat
97297	Bohring-Opitz syndrome	BOS syndrome|Bohring syndrome|C-like syndrome|Oberklaid-Danks syndrome|Opitz trigonocephaly-like syndrome
98797	Isochromosomy Yp	
98938	Colobomatous microphthalmia	MAC|Microphthalmia with colobomatous cyst|Microphthalmia-anophthalmia-coloboma syndrome
99741	King-Denborough syndrome	Koussef-Nichols syndrome
99776	Mosaic trisomy 9	Mosaic trisomy chromosome 9|Trisomy 9 mosaicism
137605	Legius syndrome	NF1-like syndrome|Neurofibromatosis 1-like syndrome

137667	Capillary malformation-arteriovenous malformation	CM-AVM
137888	Auriculocondylar syndrome	Question mark ear syndrome
139474	17q11.2 microduplication syndrome	Dup(17)(q11.2)|Grisart-Destrée syndrome|Trisomy 17q11.2
140944	CLOVES syndrome	Congenital lipomatous overgrowth-vascular malformation-epidermal nevi-skeletal anomaly syndrome|Congenital lipomatous overgrowth-vascular malformation-epidermal nevi-spinal anomaly syndrome
141136	Otomandibular syndrome	First branchial arch syndrome|Hemifacial microsomia|Laterofacial microsomia|Otomandibular dysostosis
163976	X-linked intellectual disability, Van Esch type	
171680	Lissencephaly due to TUBA1A mutation	
199318	15q13.3 microdeletion syndrome	Del(15)(q13.3)|Monosomy 15q13.3
217340	17q21.31 microduplication syndrome	Dup(17)(q21.31)|Trisomy 17q21.31
217377	Microduplication Xp11.22p11.23 syndrome	Dup(X)(p11.22p11.23)|Trisomy Xp11.22p11.23
221061	Familial cerebral cavernous malformation	Familial brain cavernous angioma|Familial cerebral cavernoma|Hereditary brain cavernous angioma|Hereditary cerebral cavernoma|Hereditary cerebral cavernous malformation
228384	5q14.3 microdeletion syndrome	Del(5)(q14.3)|Monosomy 5q14.3
228402	2q23.1 microdeletion syndrome	Del(2)(q23.1)|Monosomy 2q23.1|Pseudo-Angelman syndrome
228410	Polyvalvular heart disease syndrome	PHD syndrome
238446	15q11q13 microduplication syndrome	15q11q13 duplication syndrome|Dup(15)(q11q13)|Trisomy 15q11q13
238769	1q44 microdeletion syndrome	Del(1)(q44)|Monosomy 1q44
250989	1q21.1 microdeletion syndrome	Del(1)(q21)|Monosomy 1q21.1
250994	1q21.1 microduplication syndrome	Dup(1)(q21.1)|Trisomy 1q21.1
251014	2q31.1 microdeletion syndrome	Del(2)(q31.1)|Monosomy 2q31.1
251038	3q29 microduplication syndrome	Trisomy 3q29
251071	8p23.1 microdeletion syndrome	Del(8)(p23.1)|Monosomy 8p23.1
251076	8p23.1 duplication syndrome	Dup(8)(p23.1p23.1)|Trisomy 8p23.1
254516	Temple syndrome	
261183	15q11.2 microdeletion syndrome	15q11.2 BP1-BP2 microdeletion syndrome|Del(15)(q11.2)|Monosomy 15q11.2
261190	15q14 microdeletion syndrome	Del(15)(q14)|Monosomy 15q14
261197	Proximal 16p11.2 microdeletion syndrome	Proximal del(16)(p11.2)|Proximal monosomy 16p11.2
261204	16p11.2p12.2 microduplication syndrome	Dup(16)(p11.2p12.2)|Trisomy 16p11.2p12.2
261211	16p11.2p12.2 microdeletion syndrome	Del(16)(p11.2p12.2)|Monosomy 16p11.2p12.2
261222	Distal 16p11.2 microdeletion syndrome	Distal del(16)(p11.2)|Distal monosomy 16p11.2
261236	16p13.11 microdeletion syndrome	Del(16)(p13.11)|Monosomy 16p13.11
261243	16p13.11 microduplication syndrome	Dup(16)(p13.11)|Trisomy 16p13.11
261257	Distal 17p13.3 microdeletion syndrome	Distal del(17)(p13.3)|Distal monosomy 17p13.3
261265	17q12 microdeletion syndrome	Del(17)(q12)|Monosomy 17q12

261272	17q12 microduplication syndrome	Dup(17)(q12)|Trisomy 17q12
261295	20p12.3 microdeletion syndrome	Del(20)(p12.3)|Monosomy 20p12.3
261330	Distal 22q11.2 microdeletion syndrome	Distal del(22)(q11.2)|Distal monosomy 22q11.2
261337	Distal 22q11.2 microduplication syndrome	Distal dup(22)(q11.2)|Distal trisomy 22q11.2
261494	Kleefstra syndrome	
276413	10q22.3q23.3 microdeletion syndrome	Del(10)(q22.3q23.3)|Deletion 10q22.3q23.3|Monosomy 10q22.3q23.3
280640	Occipital pachygyria and polymicrogyria	Occipital MCD|Occipital malformations of cortical development
293939	Distal Xq28 microduplication syndrome	Distal dup(X)q(28)|Distal trisomy Xq28
293948	1p21.3 microdeletion syndrome	Del(1)(p21.3)|Monosomy 1p21.3
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	MCAHS type 2
314034	7p22.1 microduplication syndrome	Dup(7)(p22.1)|Trisomy 7p22.1
319182	Wiedemann-Steiner syndrome	Hypertrichosis-short stature-facial dysmorphism-developmental delay syndrome
329224	Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome	
363972	Noonan syndrome-like disorder with juvenile myelomonocytic leukemia	CBL syndrome|Noonan syndrome-like disorder with JMML
369891	Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	
370079	Proximal 16p11.2 microduplication syndrome	Proximal dup(16)(p11.2)|Proximal trisomy 16p11.2
391372	Intellectual disability-severe speech delay-mild dysmorphism syndrome	FOXP1 syndrome
391677	Short stature-optic atrophy-Pelger-Huët anomaly syndrome	SOPH syndrome
401973	MEND syndrome	Male EBP disorder with neurological defects
401986	1p31p32 microdeletion syndrome	Del(1)(p31p32)|Monosomy 1p31p32
404440	Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency	
404443	Tall stature-intellectual disability-facial dysmorphism syndrome	DNMT3A-related overgrowth syndrome|Tatton-Brown-Rahman overgrowth syndrome
404448	ADNP syndrome	ADNP-related syndromic intellectual disability-autism spectrum disorder|HVDAS|Helsmoortel-Van Der Aa Syndrome
404473	Severe intellectual disability-progressive spastic diplegia syndrome	
412069	AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome	Xia-Gibbs syndrome
420179	Malan overgrowth syndrome	Sotos syndrome 2
435638	3p25.3 microdeletion syndrome	Del(3)p(25.3)|Intellectual disability-epilepsy-stereotypic hand movement syndrome|Monosomy 3p25.3
447980	19p13.3 microduplication syndrome	Dup(19)(p13.13)
453499	Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-skeletal anomalies syndrome	Au-Kline syndrome
457193	Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome	

457485	Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome	MINDS syndrome|Smith-Kingsmore syndrome
466943	WAC-related facial dysmorphism-developmental delay-behavioral abnormalities syndrome	
476126	Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome	
480520	Caroli syndrome	
481152	PYCR2-related microcephaly-progressive leukoencephalopathy	
485350	CLCN4-related X-linked intellectual disability syndrome	
488586	Congenital amyoplasia	Amyoplasia congenita
488632	TBCK-related intellectual disability syndrome	
508498	Intellectual disability-cardiac anomalies-short stature-joint laxity syndrome	
521258	Xq25 microduplication syndrome	Dup(X)(q25)|Xq25 microtriplication

Morphological anomaly

619	Common mesentery	Universal mesentery
741	Familial mitral valve prolapse	
1478	Interatrial communication	ASD|Atrial septal defect|Interauricular communication
1880	Ebstein malformation	Ebstein anomaly of the tricuspid valve
2128	Isolated hemihyperplasia	Hemi 3 syndrome|Hemicorporal hypertrophy|Isolated hemihypertrophy
2149	Nodular neuronal heterotopia	
3181	Sprengel deformity	High scapula
3193	Supravalvular aortic stenosis	SVAS
3366	Isolated trigonocephaly	Non-syndromic metopic craniosynostosis
30391	Isolated biliary atresia	Isolated atresia of bile ducts|Non-syndromic biliary atresia
35098	Isolated plagiocephaly	Non-syndromic unicoronal synostosis|Synostotic plagiocephaly
35099	Isolated brachycephaly	Non-syndromic bicoronal synostosis
83463	Microtia	
93108	Renal dysplasia	
93338	Polysyndactyly	PPD4|Preaxial polydactyly type 4
95496	Pituitary stalk interruption syndrome	Ectopic neurohypophysis|PSIS
95706	Posterior hypospadias	Perineal, scrotal or penoscrotal hypospadias
99796	Subcortical band heterotopia	Subcortical laminar heterotopia
99798	Oligodontia	Selective tooth agenesis
101023	Cleft hard palate	
137902	Isolated optic nerve hypoplasia/aplasia	
199306	Cleft lip/palate	Alveolar cleft lip and palate|Cleft lip and palate|Cleft lip-alveolus-palate syndrome|FLP
250923	Isolated aniridia	
268820	Cranial meningocele	
268835	Lipomyelomeningocele	
268861	Primary tethered cord syndrome	Primary tethered spinal cord syndrome
294983	Acheiria	Congenital absence of hand
294988	Congenital absence/hypoplasia of thumb	Thumb hypodactyly|Thumb oligodactyly
295047	Macrodactyly of toes	Macrodactyly of foot
295051	Lower limb hypertrophy	

Histopathological subtype

93213	Familial idiopathic steroid-resistant nephrotic syndrome	Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis
93218	Sporadic idiopathic steroid-resistant nephrotic syndrome	Sporadic idiopathic steroid-resistant nephrotic syndrome with focal segmental glomerulosclerosis
329931	Primary membranoproliferative glomerulonephritis	

Etiological subtype

181	Hypohidrotic ectodermal dysplasia	Christ-Siemens-Touraine syndrome|X-linked anhidrotic ectodermal dysplasia|XHED
248	Hypohidrotic ectodermal dysplasia	AR-HED|Autosomal recessive anhidrotic ectodermal dysplasia
777	Rare non-syndromic intellectual disability	
2512	Isolated congenital microcephaly	MCPH|Microcephalia vera|Microcephaly vera|True microcephaly
71529	Genetic non-syndromic obesity	MC4R deficiency
79269	Mucopolysaccharidosis type 3	Heparan sulfamidase deficiency|MPS3A|MPSIIIA|Mucopolysaccharidosis type 3A|Mucopolysaccharidosis type IIIA
79270	Mucopolysaccharidosis type 3	MPS3B|MPSIIIB|Mucopolysaccharidosis type 3B|Mucopolysaccharidosis type IIIB|N-acetyl-alpha-glucosaminidase deficiency
88616	Rare non-syndromic intellectual disability	AR-NSID|NS-ARID
90635	Non-syndromic genetic deafness	Autosomal dominant isolated neurosensory deafness type DFNA|Autosomal dominant isolated neurosensory hearing loss type DFNA|Autosomal dominant isolated sensorineural deafness type DFNA|Autosomal dominant isolated sensorineural hearing loss type DFNA|Autosomal dominant non-syndromic neurosensory deafness type DFNA|Autosomal dominant non-syndromic neurosensory hearing loss type DFNA|Autosomal dominant non-syndromic sensorineural hearing loss type DFNA
90636	Non-syndromic genetic deafness	Autosomal recessive isolated neurosensory deafness type DFNB|Autosomal recessive isolated sensorineural deafness type DFNB|Autosomal recessive non-syndromic neurosensory deafness type DFNB
90641	Non-syndromic genetic deafness	Isolated mitochondrial neurosensory deafness|Isolated mitochondrial sensorineural deafness|Mitochondrial non-syndromic neurosensory deafness

93579	Atypical hemolytic-uremic syndrome	Atypical HUS with H factor anomaly|D- HUS with H factor anomaly|Hemolytic uremic syndrome without diarrhea with H factor anomaly|aHUS with H factor anomaly
93613	Cystinuria	
94065	SIN3A-related intellectual disability syndrome	Del(15)(q24)|Monosomy 15q24
96147	Kleefstra syndrome	9q subtelomeric deletion syndrome|9qSTDS|Kleefstra syndrome due to 9q subtelomeric deletion|Kleefstra syndrome due to del(9)(q34)|Kleefstra syndrome due to monosomy 9q34
96182	Silver-Russell syndrome	UPD(7)mat
96184	Temple syndrome	UPD(14)mat
96193	Beckwith-Wiedemann syndrome	Mosaic paternal uniparental disomy of chromosome 11|UPD(11)pat
96334	Kagami-Ogata syndrome	UPD(14)pat
98754	Prader-Willi syndrome	UPD(15)mat
98793	Prader-Willi syndrome	
98794	Angelman syndrome	Angelman syndrome due to maternal monosomy 15q11q13
98913	Congenital myasthenic syndrome	
99413	Turner syndrome	
99810	Porencephaly	
100050	Hereditary angioedema with C1Inh deficiency	HAE 1|HAE-I|Hereditary angioneurotic edema type 1
177901	Prader-Willi syndrome	
177910	Prader-Willi syndrome	
178469	Rare non-syndromic intellectual disability	
228363	Late infantile neuronal ceroid lipofuscinosis	
231117	Beckwith-Wiedemann syndrome	
231140	Silver-Russell syndrome	
231144	Silver-Russell syndrome	
261537	Mowat-Wilson syndrome	Hirschsprung disease and intellectual disability due to 2q22 microdeletion|Hirschsprung disease and intellectual disability due to del(2)(q22)|Hirschsprung disease and intellectual disability due to monosomy 2q22|Mowat-Wilson syndrome due to 2q22 microdeletion|Mowat-Wilson syndrome due to del(2)q(22)
261552	Mowat-Wilson syndrome	Hirschsprung disease and intellectual disability due to a ZEB2 point mutation
261572	Blepharophimosis-epicanthus inversus-ptosis syndrome	Blepharophimosis types 1 and 2 due to a point mutation
261600	Alagille syndrome	Alagille syndrome due to del(20)(p12)|Alagille syndrome due to monosomy 20p12|Alagille-Watson syndrome due to monosomy 20p12|Arteriohepatic dysplasia due to monosomy 20p12|Syndromic bile duct paucity due to monosomy 20p12

261619	Alagille syndrome	Alagille-Watson syndrome due to a JAG1 point mutation|Arteriohepatic dysplasia due to a JAG1 point mutation|Syndromic bile duct paucity due to a JAG1 point mutation
261652	Kleefstra syndrome	
293633	De Barsy syndrome	PYCR1 deficiency|Pyrroline-5-carboxylate reductase 1 deficiency
309789	Rhizomelic chondrodysplasia punctata	
330050	Encephalopathy due to mitochondrial and peroxisomal fission defect	
363700	Neurofibromatosis type 1	Von Recklinghausen disease due to NF1 mutation or intragenic deletion
363958	Koolen-De Vries syndrome	Del(17)(q21.31)|Monosomy 17q21.31
398069	Prader-Willi syndrome	PWS due to a point mutation|Schaaf-Yang syndrome
438216	PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome	

Biological anomaly

206599	Isolated asymptomatic elevation of creatine phosphokinase	Idiopathic asymptomatic hyperCKemia|Isolated asymptomatic hyperCKemia

Non rare in Europe

106	NON RARE IN EUROPE: Autism	
155	NON RARE IN EUROPE: Familial isolated hypertrophic cardiomyopathy	NON RARE IN EUROPE: Familial isolated hypertrophic obstructive cardiomyopathy|Familial isolated hypertrophic subaortic stenosis|Familial or idiopathic hypertrophic subaortic stenosis|Familila or idiopathic hypertrophic obstructive cardiomyopathy|Hypertrophic obstructive cardiomyopathy|Primitive hypertrophic obstructive cardiomyopathy|Primitive hypertrophic subaortic stenosis
164	NON RARE IN EUROPE: Cerebral cavernous malformations	NON RARE IN EUROPE: Brain cavernous angioma|Brain cavernous hemangioma|Cerebral cavernoma
336	NON RARE IN EUROPE: Fibromuscular dysplasia of arteries	
357	NON RARE IN EUROPE: Gilbert syndrome	NON RARE IN EUROPE: Familial cholemia|Hyperbilirubinemia type 1||Primary renal tubular hypokalemic hypomagnesemia with hypocalciuria
362	NON RARE IN EUROPE: Glucose-6-phosphate-dehydrogenase deficiency	NON RARE IN EUROPE: Favism|G6PD deficiency
406	NON RARE IN EUROPE: Heterozygous familial hypercholesterolemia	HeFH
462	NON RARE IN EUROPE: Autosomal dominant ichthyosis vulgaris	
484	NON RARE IN EUROPE: Klinefelter syndrome	NON RARE IN EUROPE: 47,XXY syndrome

555	NON RARE IN EUROPE: Celiac disease	NON RARE IN EUROPE: CD|Celiac sprue|Coeliac disease|Coeliac sprue|Gluten intolerance|Gluten-induced enteropathy|Gluten-sensitive enteropathy|Idiopathic steatorrhea|Nontropical sprue
618	NON RARE IN EUROPE: Primary ovarian failure	NON RARE IN EUROPE: Hypergonadotropic ovarian failure|POF|Premature menopause|Premature ovarian failure|Premature ovarian insufficiency|Primary ovarian insufficiency
64738	NON RARE IN EUROPE: Non rare thrombophilia	
95698	NON RARE IN EUROPE: Non-classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	NON RARE IN EUROPE: NCAH
97562	NON RARE IN EUROPE: Benign familial hematuria	
319658	NON RARE IN EUROPE: Unexplained intellectual disability	

Obsolete

77	OBSOLETE: Aniridia	
1034	OBSOLETE: Amniotic bands	OBSOLETE: ADAM syndrome|Amniotic deformity-adhesion-mutilation syndrome
98335	OBSOLETE: Male infertility with normal virilization due to an acquired testicular defect	
98670	OBSOLETE: Vitreoretinal degeneration	
99876	OBSOLETE: Arthrochalasia Ehlers-Danlos syndrome	
247871	OBSOLETE: Vitiligo-associated autoimmune disease	
294937	OBSOLETE: Brachydactyly	
294992	OBSOLETE: Isolated split hand-split foot malformation	OBSOLETE: Ectrodactyly of hand
308604	OBSOLETE : Glycogen storage disease due to acid maltase deficiency	OBSOLETE : Alpha-1,4-glucosidase acid deficiency, adult onset|GSD due to acid maltase deficiency, adult onset|GSD type 2, adulte onset|Glycogen storage disease type 2, adult onset|Glycogenosis due to acid maltase deficiency, adult onset|Glycogenosis type 2, adult onset|Pompe disease, adult onset
480773	OBSOLETE: FATCO syndrome	