ORPHAcode	Preferred term (english label)	Synonyms EN	TypTag	Preferential parent
636	Neurofibromatosis type 1	NF1|Von Recklinghausen disease	Disease	Rare developmental defect during embryogenesis
145	Hereditary breast and ovarian cancer syndrome		Disease	Rare neoplastic disease
285	Hypermobile Ehlers-Danlos syndrome	EDS III|EDS-HT|Ehlers-Danlos syndrome hypermobility type|Ehlers-Danlos syndrome type 3|Hypermobile EDS|hEDS	Disease	Rare systemic or rheumatologic disease
870	Down syndrome	Trisomy 21	Malformation syndrome	Rare developmental defect during embryogenesis
648	Noonan syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
730	Autosomal dominant polycystic kidney disease	ADPKD	Disease	Rare renal disease
217569	Hypertrophic cardiomyopathy		Category	0
68335	Rare chromosomal anomaly		Category	0
144	Lynch syndrome		Disease	Rare neoplastic disease
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	Malformation syndrome	Rare developmental defect during embryogenesis

87277	Rare intellectual disability		Category	0
558	Marfan syndrome	MFS	Disease	Rare systemic or rheumatologic disease
217604	Dilated cardiomyopathy		Category	0
399	Huntington disease	Huntington chorea	Disease	Rare neurologic disease
102369	Rare syndromic intellectual disability		Category	0
758	Pseudoxanthoma elasticum	Gronblad-Strandberg-Touraine syndrome|PXE	Disease	Rare systemic or rheumatologic disease
908	Fragile X syndrome	FRAXA syndrome|FXS|FraX syndrome|Martin-Bell syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
739	Prader-Willi syndrome	Prader-Labhart-Willi syndrome	Disease	Rare developmental defect during embryogenesis
2322	Kabuki syndrome	Kabuki make-up syndrome|Niikawa-Kuroki syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
324	Fabry disease	Alpha-galactosidase A deficiency|Anderson-Fabry disease|Angiokeratoma corporis diffusum|Diffuse angiokeratoma|FD	Disease	Rare inborn errors of metabolism
15	Achondroplasia		Disease	Rare bone disease
88918	Autosomal dominant Alport syndrome		Clinical subtype	Rare renal disease
99999	Rare disease: no mapping possible		Category	Category
774	Hereditary hemorrhagic telangiectasia	HHT|Rendu-Osler disease|Rendu-Osler-Weber disease	Disease	Rare developmental defect during embryogenesis
122	Birt-Hogg-Dubé syndrome	Fibrofolliculomas with trichodiscomas and acrochordons|Hornstein-Knickenberg syndrome	Malformation syndrome	Rare neoplastic disease
130	Brugada syndrome	Idiopathic ventricular fibrillation, Brugada type	Disease	Rare cardiac disease

273	Steinert myotonic dystrophy	Myotonic dystrophy type 1|Steinert disease	Disease	Rare neurologic disease
178469	Autosomal dominant non-syndromic intellectual disability		Etiological subtype	Rare neurologic disease
321	Multiple osteochondromas	Bessel-Hagen disease|Multiple cartilaginous exostoses	Disease	Rare bone disease
768	Familial long QT syndrome	Congenital long QT syndrome|LQTS	Clinical group	Rare cardiac disease
904	Williams syndrome	Deletion 7q11.23|Monosomy 7q11.23|Williams-Beuren syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
778	Rett syndrome		Disease	Rare neurologic disease
88917	X-linked Alport syndrome		Clinical subtype	Rare renal disease
90636	Autosomal recessive non-syndromic sensorineural deafness type DFNB	Autosomal recessive isolated neurosensory deafness type DFNB|Autosomal recessive isolated neurosensory hearing loss type DFNB|Autosomal recessive isolated sensorineural deafness type DFNB|Autosomal recessive isolated sensorineural hearing loss type DFNB|A	Etiological subtype	Rare otorhinolaryngologic disease
821	Sotos syndrome	Cerebral gigantism	Disease	Rare developmental defect during embryogenesis
1465	Coffin-Siris syndrome	CSS	Malformation syndrome	Rare developmental defect during embryogenesis
881	Turner syndrome	45,X syndrome|45,X/46,XX syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
227535	Hereditary breast cancer	Familial breast cancer|Familial breast carcinoma|Hereditary breast carcinoma	Disease	Rare neoplastic disease

201	Cowden syndrome	Cowden disease|Multiple hamartoma syndrome	Disease	Rare developmental defect during embryogenesis
287	Classical Ehlers-Danlos syndrome	Classical EDS|cEDS|	Disease	Rare systemic or rheumatologic disease
101081	Charcot-Marie-Tooth disease type 1A	CMT1A|Microduplication 17p12	Disease	Rare neurologic disease
63	Alport syndrome	Alport deafness-nephropathy|Alport hearing loss-nephropathy	Disease	Rare renal disease
550	MELAS	Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes|Mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes|Mitochondrial myopathy, encephalopathy, lactic acidosis and stroke-like episodes	Disease	Rare neurologic disease
805	Tuberous sclerosis complex	Bourneville syndrome|Tuberous sclerosis	Disease	Rare developmental defect during embryogenesis
1727	22q11.2 duplication syndrome	22q11.2 microduplication syndrome|Dup(22)(q11)|Duplication 22q11.2|Trisomy 22q11.2	Malformation syndrome	Rare developmental defect during embryogenesis
90635	Autosomal dominant non-syndromic sensorineural deafness type DFNA	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|Autos	Etiological subtype	Rare otorhinolaryngologic disease

240	Léri-Weill dyschondrosteosis	Léri-Weill syndrome	Malformation syndrome	Rare bone disease
261183	15q11.2 microdeletion syndrome	15q11.2 BP1-BP2 microdeletion syndrome|Del(15)(q11.2)|Monosomy 15q11.2	Malformation syndrome	Rare developmental defect during embryogenesis
154	Familial isolated dilated cardiomyopathy	Familial or idiopathic dilated cardiomyopathy	Disease	Rare cardiac disease
199	Cornelia de Lange syndrome	Brachmann-de Lange syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
247	Arrhythmogenic right ventricular cardiomyopathy	ARVC|ARVD|Arrhythmogenic right ventricular dysplasia	Clinical group	0
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	Malformation syndrome	Rare neurologic disease
733	Familial adenomatous polyposis	Colorectal adenomatous polyposis|FAP|Familial polyposis coli	Disease	Rare gastroenterologic disease
101685	Rare non-syndromic intellectual disability	Rare NSID	Disease	Rare neurologic disease
244	Primary ciliary dyskinesia	PCD	Disease	Rare respiratory disease
269	Facioscapulohumeral dystrophy	FSH dystrophy|FSHD|Facioscapulohumeral muscular dystrophy|Facioscapulohumeral myopathy|Landouzy-Dejerine dystrophy|Landouzy-Dejerine myopathy	Disease	Rare neurologic disease

887	VACTERL/VATER association	VACTERL association|VATER association	Malformation syndrome	Rare developmental defect during embryogenesis
98053	Rare genetic disease		Category	0
221061	Familial cerebral cavernous malformation	Familial brain cavernous angioma|Familial cerebral cavernoma|Hereditary brain cavernous angioma|Hereditary cerebral cavernoma|Hereditary cerebral cavernous malformation	Malformation syndrome	Rare developmental defect during embryogenesis
3286	Catecholaminergic polymorphic ventricular tachycardia	Bidirectional ventricular tachycardia induced by catecholamine|CPVT|Malignant paroxysmal ventricular tachycardia|Polymorphic ventricular tachycardia induced by catecholamines	Disease	Rare cardiac disease
87884	Non-syndromic genetic deafness	Isolated genetic deafness|Isolated genetic hearing loss|Non-syndromic genetic hearing loss	Disease	Rare otorhinolaryngologic disease
91387	Familial thoracic aortic aneurysm and aortic dissection	Familial TAAD	Disease	Rare developmental defect during embryogenesis
199318	15q13.3 microdeletion syndrome	Del(15)(q13.3)|Monosomy 15q13.3	Malformation syndrome	Rare developmental defect during embryogenesis
250989	1q21.1 microdeletion syndrome	Del(1)(q21)|Monosomy 1q21.1	Malformation syndrome	Rare developmental defect during embryogenesis
116	Beckwith-Wiedemann syndrome	BWS|Exomphalos-macroglossia-gigantism syndrome|Wiedemann-Beckwith syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
652	Multiple endocrine neoplasia type 1	MEN1|Wermer syndrome	Disease	Rare neoplastic disease

29072	Hereditary pheochromocytoma-paraganglioma	Familial pheochromocytoma-paraganglioma	Disease	Rare neoplastic disease
60030	Loeys-Dietz syndrome	Aortic aneurysm syndrome due to TGF-beta receptors anomalies	Malformation syndrome	Rare systemic or rheumatologic disease
141132	Oculo-auriculo-vertebral spectrum	OAV spectrum|Oculoauriculovertebral spectrum	Malformation syndrome	0
48652	Monosomy 22q13.3	22q13.3 deletion|Phelan-McDermid syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
94068	Spondyloepiphyseal dysplasia congenita	Congenital spondyloepiphyseal dysplasia|SEDC|Spranger-Wiedemann disease	Disease	Rare bone disease
216796	Osteogenesis imperfecta type 1	Adair-Dighton syndrome|Mild osteogenesis imperfecta|Non-deforming osteogenesis imperfecta|OI type 1|Van der Hoeve syndrome	Clinical subtype	Rare bone disease
250994	1q21.1 microduplication syndrome	Dup(1)(q21.1)|Trisomy 1q21.1	Malformation syndrome	Rare developmental defect during embryogenesis
110	Bardet-Biedl syndrome	BBS	Disease	Rare developmental defect during embryogenesis
464	Incontinentia pigmenti	Bloch-Siemens syndrome|Bloch-Sulzberger syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
783	Rubinstein-Taybi syndrome	Broad thumb-hallux syndrome|Broad thumbs-halluces syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
791	Retinitis pigmentosa		Disease	Rare ophthalmic disorder
803	Amyotrophic lateral sclerosis	ALS|Charcot disease|Lou Gehrig disease	Disease	Rare neurologic disease
828	Stickler syndrome	Hereditary progressive arthroophthalmopathy	Disease	Rare bone disease

79254	Classic phenylketonuria	Classic PKU	Clinical subtype	Rare inborn errors of metabolism
98006	Rare neurologic disease	Rare nervous system disease	Category	0
137605	Legius syndrome	NF1-like syndrome|Neurofibromatosis 1-like syndrome	Malformation syndrome	Rare skin disease
247698	Multiple endocrine neoplasia type 2A	MEN2A|PTC syndrome|Sipple syndrome	Clinical subtype	Rare neoplastic disease
261197	Proximal 16p11.2 microdeletion syndrome	Proximal del(16)(p11.2)|Proximal monosomy 16p11.2	Malformation syndrome	Rare developmental defect during embryogenesis
306498	PTEN hamartoma tumor syndrome	PHTS	Clinical group	0
60	Alpha-1-antitrypsin deficiency	Alpha-1-proteinase inhibitor deficiency|Alpha1-antitrypsin deficiency	Disease	Rare respiratory disease
138	CHARGE syndrome	CHARGE association|Coloboma-heart defects-atresia choanae-retardation of growth and development-genitourinary problems-ear abnormalities syndrome|Hall-Hittner syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
478	Kallmann syndrome	Congenital hypogonadotropic hypogonadism with anosmia|Olfacto-genital pathological sequence	Clinical subtype	Rare endocrine disease
819	Smith-Magenis syndrome	17p11.2 microdeletion syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2614	Nail-patella syndrome	Onychoosteodysplasia|Turner-Kieser syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3095	Atypical Rett syndrome	Atypical RTT|Rett syndrome variant	Disease	Rare developmental defect during embryogenesis

72	Angelman syndrome		Malformation syndrome	Rare neurologic disease
429	Hypochondroplasia		Disease	Rare bone disease
552	MODY	Maturity-onset diabetes of the young	Disease	Rare endocrine disease
637	Neurofibromatosis type 2	NF2	Disease	Rare otorhinolaryngologic disease
666	Osteogenesis imperfecta	Brittle bone disease|Glass bone disease|Lobstein disease|OI	Disease	Rare bone disease
777	X-linked non-syndromic intellectual disability		Etiological subtype	Rare neurologic disease
93921	Schwannomatosis	NF3|Neurilemmomatosis|Neurofibromatosis type 3	Disease	Rare developmental defect during embryogenesis
97929	Rare cardiac disease		Category	0
52	Alagille syndrome	Alagille-Watson syndrome|Arteriohepatic dysplasia|Syndromic bile duct paucity	Malformation syndrome	Rare developmental defect during embryogenesis
104	Leber hereditary optic neuropathy	LHON|Leber optic atrophy	Disease	Rare ophthalmic disorder
136	Cerebral autosomal dominant arteriopathy-subcortical infarcts-leukoencephalopathy	CADASIL|Hereditary multi-infarct dementia	Disease	Rare neurologic disease
286	Vascular Ehlers-Danlos syndrome	Arterial-ecchymotic EDS|EDS IV|Ehlers-Danlos syndrome type 4|Sack-Barabas syndrome|Vascular EDS|vEDS	Disease	Rare systemic or rheumatologic disease
380	Greig cephalopolysyndactyly syndrome	GCPS	Malformation syndrome	Rare developmental defect during embryogenesis
423	Malignant hyperthermia of anesthesia	Hyperthermia of anesthesia	Disease	Rare neurologic disease
586	Cystic fibrosis	CF|Mucoviscidosis	Disease	Rare respiratory disease

610	Bethlem myopathy	Benign autosomal dominant myopathy	Disease	Rare neurologic disease
827	Stargardt disease	Fundus flavimaculatus|Stargardt 1	Disease	Rare ophthalmic disorder
892	Von Hippel-Lindau disease	Familial cerebelloretinal angiomatosis|Lindau disease|VHL|Von Hippel-Lindau syndrome	Disease	Rare neoplastic disease
1906	Fetal valproate spectrum disorder	Fetal valproate syndrome|Fetal valproic acid syndrome|Valproic acid embryopathy	Malformation syndrome	Rare developmental defect during embryogenesis
137667	Capillary malformation-arteriovenous malformation	CM-AVM	Malformation syndrome	Rare developmental defect during embryogenesis
261265	17q12 microdeletion syndrome	Del(17)(q12)|Monosomy 17q12	Malformation syndrome	Rare developmental defect during embryogenesis
193	Cohen syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
731	Autosomal recessive polycystic kidney disease	AR-PKD	Disease	Rare renal disease
813	Silver-Russell syndrome	Silver-Russell dwarfism	Disease	Rare developmental defect during embryogenesis
1340	Cardiofaciocutaneous syndrome	CFC syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2512	Autosomal recessive primary microcephaly	MCPH|Microcephalia vera|Microcephaly vera|True microcephaly	Etiological subtype	Rare developmental defect during embryogenesis
2911	Poland syndrome	Poland anomaly|Poland sequence	Malformation syndrome	Rare developmental defect during embryogenesis
3342	Arterial tortuosity syndrome	ATS	Malformation syndrome	Rare circulatory system disease
183757	Rare genetic intellectual disability		Category	0

238446	15q11q13 microduplication syndrome	15q11q13 duplication syndrome|Dup(15)(q11q13)|Trisomy 15q11q13	Malformation syndrome	Rare developmental defect during embryogenesis
238468	Hypohidrotic ectodermal dysplasia	Anhidrotic ectodermal dysplasia|HED	Disease	Rare developmental defect during embryogenesis
261211	16p11.2p12.2 microdeletion syndrome	Del(16)(p11.2p12.2)|Monosomy 16p11.2p12.2	Malformation syndrome	Rare developmental defect during embryogenesis
261222	Distal 16p11.2 microdeletion syndrome	Distal del(16)(p11.2)|Distal monosomy 16p11.2	Malformation syndrome	Rare developmental defect during embryogenesis
284963	Marfan syndrome type 1	MFS1	Clinical subtype	Rare systemic or rheumatologic disease
280	Wolf-Hirschhorn syndrome	4p- syndrome|Distal deletion 4p|Distal monosomy 4p|Telomeric deletion 4p	Malformation syndrome	Rare developmental defect during embryogenesis
281	Monosomy 5p	Cri du chat syndrome|Deletion 5p	Malformation syndrome	Rare developmental defect during embryogenesis
306	Benign familial infantile epilepsy	BFIE|BFIS|Benign familial infantile convulsions|Benign familial infantile seizures	Disease	Rare neurologic disease
342	Familial Mediterranean fever	Benign paroxysmal peritonitis|Benign recurrent polyserositis|FMF|Familial paroxysmal polyserositis|Periodic disease	Disease	Rare systemic or rheumatologic disease
377	Gorlin syndrome	Basal cell nevus syndrome|Gorlin-Goltz syndrome|NBCCS|Nevoid basal cell carcinoma syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
1606	1p36 deletion syndrome	Del(1)(p36)|Deletion 1p36|Deletion 1pter|Monosomy 1p36|Monosomy 1pter|Subtelomeric 1p36 deletion	Malformation syndrome	Rare developmental defect during embryogenesis

1713	17p11.2 microduplication syndrome	Potocki-Lupski syndrome|Trisomy 17p11.2	Malformation syndrome	Rare developmental defect during embryogenesis
2295	Familial articular hypermobility syndrome	Familial joint instability syndrome|Familial joint laxity|Joint instability syndrome	Disease	Rare systemic or rheumatologic disease
2345	Isolated Klippel-Feil syndrome	Congenital cervical vertebral fusion|Congenital fused cervical segments|Klippel-Feil malformation|Klippel-Feil sequence	Malformation syndrome	Rare developmental defect during embryogenesis
79373	Ectodermal dysplasia syndrome	Ectodermal dysplasia	Category	0
93622	Dent disease type 1	Nephrolithiasis type 1	Clinical subtype	Rare renal disease
98673	Autosomal dominant optic atrophy, classic form	Autosomal dominant optic atrophy, Kjer type|Kjer optic atrophy|Optic atrophy type 1	Disease	Rare ophthalmic disorder
261494	Kleefstra syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
8	47,XYY syndrome	Double Y syndrome|XYY syndrome|Y disomy	Malformation syndrome	Rare developmental defect during embryogenesis
248	Autosomal recessive hypohidrotic ectodermal dysplasia	AR-HED|Autosomal recessive anhidrotic ectodermal dysplasia	Etiological subtype	Rare developmental defect during embryogenesis
313	Lamellar ichthyosis	Classic lamellar ichthyosis|Congenital lamellar ichthyosis|LI	Disease	Rare skin disease
432	Normosmic congenital hypogonadotropic hypogonadism	Normosmic idiopathic hypogonadotropic hypogonadism|nIHH	Clinical subtype	Rare endocrine disease
461	Recessive X-linked ichthyosis	RXLI|Steroid sulfatase deficiency|X-linked ichthyosis|XLI	Disease	Rare skin disease
524	Li-Fraumeni syndrome		Disease	Rare neoplastic disease

656	Genetic steroid-resistant nephrotic syndrome	Familial idiopathic steroid-resistant nephrotic syndrome|Genetic SRNS|Hereditary steroid-resistant nephrotic syndrome	Disease	Rare renal disease
685	Hereditary spastic paraplegia	Familial spastic paraplegia|HSP|Hereditary spastic paraparesis|SPG|Strümpell-Lorrain disease	Clinical group	0
782	Axenfeld-Rieger syndrome	Axenfeld syndrome|Rieger syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
1762	Proximal Xq28 duplication syndrome	MECP2 duplication syndrome|X-linked intellectual disability syndrome, Lubs type	Malformation syndrome	Rare developmental defect during embryogenesis
1915	Fetal alcohol syndrome	ARBD|ARND|Alcohol-related birth defects|Alcohol-related neurodevelopmental disorder|FAS|FASD|Fetal alcohol spectrum disorders	Malformation syndrome	Rare developmental defect during embryogenesis
77258	Trichorhinophalangeal syndrome type 1 and 3		Malformation syndrome	Rare developmental defect during embryogenesis
79113	Mandibulofacial dysostosis-microcephaly syndrome	MFDM syndrome|Mandibulofacial dysostosis, Guion-Almeida type	Malformation syndrome	Rare developmental defect during embryogenesis
88888	Carrier of disease		Disease	Disease
88919	Autosomal recessive Alport syndrome		Clinical subtype	Rare renal disease
93111	HNF1B-related autosomal dominant tubulointerstitial kidney disease	ADTKD-HNF1B|HNF1B-MODY|MODY5|Maturity-onset diabetes of the young type 5|RCAD syndrome|Renal cysts and diabetes syndrome|Renal dysfunction-early-onset diabetes syndrome	Clinical subtype	Rare endocrine disease

96121	7q11.23 microduplication syndrome	Dup(7)(q11.23)|Trisomy 7q11.23	Malformation syndrome	Rare developmental defect during embryogenesis
101075	X-linked Charcot-Marie-Tooth disease type 1	CMT1X|CMTX1	Disease	Rare neurologic disease
231117	Beckwith-Wiedemann syndrome due to imprinting defect of 11p15		Etiological subtype	Rare developmental defect during embryogenesis
261330	Distal 22q11.2 microdeletion syndrome	Distal del(22)(q11.2)|Distal monosomy 22q11.2	Malformation syndrome	Rare developmental defect during embryogenesis
300751	Familial dilated cardiomyopathy with conduction defect due to LMNA mutation		Disease	Rare cardiac disease
84	Fanconi anemia	Fanconi pancytopenia	Malformation syndrome	Rare developmental defect during embryogenesis
87	Apert syndrome	ACS1|Acrocephalosyndactyly type 1	Malformation syndrome	Rare developmental defect during embryogenesis
100	Ataxia-telangiectasia	Louis-Bar syndrome	Disease	Rare neurologic disease
126	Blepharophimosis-ptosis-epicanthus inversus syndrome	BPES	Malformation syndrome	Rare developmental defect during embryogenesis
166	Charcot-Marie-Tooth disease/Hereditary motor and sensory neuropathy	CMT/HMSN|Charcot-Marie-Tooth hereditary neuropathy	Category	0
223	Nephrogenic diabetes insipidus		Disease	Rare renal disease
718	Isolated Pierre Robin syndrome	Isolated Pierre Robin sequence	Malformation syndrome	Rare developmental defect during embryogenesis
822	Hereditary spherocytosis	Minkowski-Chauffard disease	Disease	Rare hematologic disease
847	Alpha-thalassemia-X-linked intellectual disability syndrome	ATR-X syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
848	Beta-thalassemia		Disease	Rare hematologic disease

915	Aarskog-Scott syndrome	Aarskog syndrome|Faciodigitogenital syndrome|Faciogenital dysplasia	Malformation syndrome	Rare developmental defect during embryogenesis
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 fis	Malformation syndrome	Rare developmental defect during embryogenesis
2152	Mowat-Wilson syndrome	Hirschsprung disease-intellectual disability syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2896	Pitt-Hopkins syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
3193	Supravalvular aortic stenosis	SVAS	Morphological anomaly	Rare developmental defect during embryogenesis
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	Disease	Rare neurologic disease
36387	Generalized epilepsy with febrile seizures-plus	GEFS+|Genetic epilepsy with febrile seizures-plus	Disease	Rare neurologic disease

49382	Achromatopsia	ACHM|Complete or incomplete color blindness|Pingelapese blindness|Rod monochromacy|Rod monochromatism|Total color blindness	Disease	Rare ophthalmic disorder
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	Malformation syndrome	Rare developmental defect during embryogenesis
83454	Glomuvenous malformation	Glomangiomatosis|Hereditary multiple glomangiomas|Multiple glomus tumors|VMGLOM|Venous malformations with glomus cells	Malformation syndrome	Rare developmental defect during embryogenesis
93890	Rare developmental defect during embryogenesis	Malformation syndrome	Category	0
94089	Pseudohypoparathyroidism type 1B		Disease	Rare endocrine disease
97935	Rare gastroenterologic disease		Category	0
98758	Spinocerebellar ataxia type 6	SCA6	Disease	Rare neurologic disease
98892	Periventricular nodular heterotopia	PVNH	Clinical subtype	Rare developmental defect during embryogenesis
98895	Becker muscular dystrophy	BMD|Becker dystrophinopathy	Disease	Rare neurologic disease

99739	Rare familial disorder with hypertrophic cardiomyopathy	Rare familial disorder with hypertrophic obstructive cardiomyopathy|Rare familial disorder with hypertrophic subaortic stenosis	Category	0
99798	Oligodontia	Selective tooth agenesis	Morphological anomaly	Rare odontologic disease
183518	Rare hereditary ataxia		Category	0
261272	17q12 microduplication syndrome	Dup(17)(q12)|Trisomy 17q12	Malformation syndrome	Rare developmental defect during embryogenesis
261619	Alagille syndrome due to a JAG1 point mutation	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	Etiological subtype	Rare developmental defect during embryogenesis
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	Category	0
284984	Aneurysm-osteoarthritis syndrome		Disease	Rare systemic or rheumatologic disease
289539	BAP1-related tumor predisposition syndrome	Tumor susceptibility linked to germline BAP1 mutations	Disease	Rare neoplastic disease
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	Disease	Rare inborn errors of metabolism

55	Oculocutaneous albinism	OCA	Clinical group	Rare skin disease
97	Familial paroxysmal ataxia	Episodic ataxia type 2	Disease	Rare neurologic disease
115	Congenital contractural arachnodactyly	Beals syndrome|Beals-Hecht syndrome|CCA syndrome|Distal arthrogryposis type 9	Malformation syndrome	Rare developmental defect during embryogenesis
174	Metaphyseal chondrodysplasia, Schmid type		Disease	Rare bone disease
175	Cartilage-hair hypoplasia	Autosomal recessive metaphyseal chondrodysplasia|Metaphyseal chondrodysplasia, McKusick type|Autosomal recessive metaphyseal chondrodysplasia|Metaphyseal chondrodysplasia, McKusick type	Disease	Rare bone disease
215	Congenital stationary night blindness	Congenital essential nyctalopia	Disease	Rare ophthalmic disorder
388	Hirschsprung disease	Aganglionic megacolon|Congenital intestinal aganglionosis|HSCR	Disease	Rare gastroenterologic disease
403	Familial hyperaldosteronism type I	Dexamethasone-sensitive hypertension|FH-I|FH1|Familial hyperaldosteronism type 1|GRA|Glucocorticoid-remediable aldosteronism|Glucocorticoid-sensitive hypertension	Disease	Rare endocrine disease
500	Noonan syndrome with multiple lentigines	Cardiomyopathic lentiginosis|Familial multiple lentigines syndrome|LEOPARD syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
590	Congenital myasthenic syndrome	CMS	Disease	Rare neurologic disease
684	Paramyotonia congenita of Von Eulenburg	Paramyotonia congenita	Disease	Rare neurologic disease

709	Peters plus syndrome	Krause-Kivlin syndrome|Krause-van Schooneveld-Kivlin syndrome|Peters anomaly with short limb dwarfism	Malformation syndrome	Rare developmental defect during embryogenesis
750	Pseudoachondroplasia	Pseudoachondroplastic dysplasia|Pseudoachondroplastic spondyloepiphyseal dysplasia	Disease	Rare bone disease
794	Saethre-Chotzen syndrome	ACS3|Acrocephalosyndactyly type 3|SCS	Malformation syndrome	Rare developmental defect during embryogenesis
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	Malformation syndrome	Rare developmental defect during embryogenesis
1452	Cleidocranial dysplasia	Cleidocranial dysostosis	Malformation syndrome	Rare bone disease
1531	Craniosynostosis		Category	0
1934	Early infantile epileptic encephalopathy	EIEE|Early infantile epileptic encephalopathy with suppression-bursts|Ohtahara syndrome	Clinical syndrome	Rare neurologic disease
2134	Atypical hemolytic uremic syndrome	Atypical HUS|aHUS	Disease	Rare renal disease
2309	Pachyonychia congenita	PC	Disease	Rare skin disease
2588	Myhre syndrome	Facial dysmorphism-intellectual disability-short stature-deafness syndrome|Facial dysmorphism-intellectual disability-short stature-hearing loss syndrome	Malformation syndrome	Rare developmental defect during embryogenesis

2612	Linear nevus sebaceus syndrome	Nevus sebaceus of Jadassohn|Nevus sebaceus syndrome|Organoid nevus syndrome|Schimmelpenning syndrome|Solomon syndrome	Disease	Rare developmental defect during embryogenesis
2924	Isolated polycystic liver disease	ADPCLD|Autosomal dominant polycystic liver disease|PCLD	Malformation syndrome	Rare hepatic disease
3071	Costello syndrome	FCS syndrome|Faciocutaneoskeletal syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3375	Trisomy X	47,XXX syndrome|Triple X syndrome|Triplo-X syndrome|XXX syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
34149	Autosomal dominant tubulointerstitial kidney disease	ADTKD|Autosomal dominant medullary cystic kidney disease|MCKD	Disease	Rare renal disease
65286	3q29 microdeletion syndrome	3q subtelomere deletion syndrome|3qter deletion|Del(3)(q29)|Monosomy 3q29|Monosomy 3qter	Malformation syndrome	Rare developmental defect during embryogenesis
73229	HANAC syndrome	Autosomal dominant familial hematuria-retinal arteriolar tortuosity-contractures syndrome|Hereditary angiopathy-nephropathy-aneurysms-muscle cramps syndrome	Disease	Rare neurologic disease
75857	6q terminal deletion syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
79432	Oculocutaneous albinism type 2	OCA2	Disease	Rare skin disease

79443	Pseudohypoparathyroidism type 1A	AHO-PHP syndrome Ia|Albright hereditary osteodystrophy-PHP syndrome Ia	Disease	Rare endocrine disease
85287	X-linked intellectual disability, Siderius type		Malformation syndrome	Rare developmental defect during embryogenesis
90692	Rare endocrine growth disease		Category	0
91492	Early-onset non-syndromic cataract		Disease	Rare ophthalmic disorder
93108	Renal dysplasia	Kidney dysplasia	Morphological anomaly	Rare developmental defect during embryogenesis
98127	Autosomal anomaly		Category	0
98196	Malformation syndrome with hamartosis	Dysmorphologic diseases with phakomatosis	Category	0
98672	Autosomal dominant optic atrophy	ADOA|DOA	Clinical group	0
98991	Early-onset nuclear cataract		Clinical subtype	Rare ophthalmic disorder
99013	Spastic paraplegia type 7	SPG7	Disease	Rare neurologic disease
100985	Autosomal dominant spastic paraplegia type 4	SPG4	Disease	Rare neurologic disease
139474	17q11.2 microduplication syndrome	Dup(17)(q11.2)|Grisart-Destrée syndrome|Trisomy 17q11.2	Malformation syndrome	Rare developmental defect during embryogenesis
140944	CLOVES syndrome	Congenital lipomatous overgrowth-vascular malformation-epidermal nevi-skeletal anomaly syndrome|Congenital lipomatous overgrowth-vascular malformation-epidermal nevi-spinal anomaly syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
171680	Lissencephaly due to TUBA1A mutation		Malformation syndrome	Rare developmental defect during embryogenesis
180772	Rare disease with autism		Category	0

209905	Brain-lung-thyroid syndrome	Choreoathetosis-hypothyroidism-neonatal respiratory distress syndrome	Disease	Rare developmental defect during embryogenesis
231169	Usher syndrome type 1	USH1	Clinical subtype	Rare developmental defect during embryogenesis
247667	Childhood-onset hypophosphatasia	Childhood-onset Rathbun disease|Childhood-onset phosphoethanolaminuria	Clinical subtype	Rare bone disease
250923	Isolated aniridia		Morphological anomaly	Rare developmental defect during embryogenesis
251071	8p23.1 microdeletion syndrome	Del(8)(p23.1)|Monosomy 8p23.1	Malformation syndrome	Rare developmental defect during embryogenesis
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	Category	0
262698	Partial duplication of the short arm of chromosome 2	Partial duplication of chromosome 2p|Partial trisomy of chromosome 2p|Saal-Greenstein syndrome	Category	0
263708	Complex chromosomal rearrangement		Category	0
51	Aicardi-Goutières syndrome	Encephalopathy with basal ganglia calcification|Encephalopathy with intracranial calcification and chronic lymphocytosis of cerebrospinal fluid	Disease	Rare neurologic disease
95	Friedreich ataxia	FA|FRDA	Disease	Rare neurologic disease
181	X-linked hypohidrotic ectodermal dysplasia	Christ-Siemens-Touraine syndrome|X-linked anhidrotic ectodermal dysplasia|XHED	Etiological subtype	Rare developmental defect during embryogenesis

214	Cystinuria	Cystinuria-lysinuria syndrome	Disease	Rare renal disease
232	Sickle cell anemia		Disease	Rare hematologic disease
251	Multiple epiphyseal dysplasia	EDM|MED|Polyepiphyseal dysplasia	Clinical group	0
289	Ellis Van Creveld syndrome	Chondroectodermal dysplasia|Mesodermic dysplasia	Malformation syndrome	Rare bone disease
312	Autosomal dominant epidermolytic ichthyosis	BCIE|Bullous congenital ichthyosiform erythroderma|Bullous congenital ichthyosiform erythroderma of Brock|Bullous ichthyosis|EHK||Epidermolytic hyperkeratosis|Ichthyosis hystrix Brocq type	Disease	Rare skin disease
352	Galactosemia		Category	Rare inborn errors of metabolism
358	Gitelman syndrome	Primary renal tubular hypokalemic hypomagnesemia with hypocalciuria	Disease	Rare renal disease
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|	Disease	Rare inborn errors of metabolism
392	Holt-Oram syndrome	Atriodigital dysplasia type 1|HOS|Heart-hand syndrome type 1	Malformation syndrome	Rare developmental defect during embryogenesis

394	Classic homocystinuria	Cystathionine beta-synthase deficiency|Homocystinuria due to cystathionine beta-synthase deficiency	Disease	Rare inborn errors of metabolism
474	Jeune syndrome	Asphyxiating thoracic dystrophy of the newborn|JATD|Jeune asphyxiating thoracic dystrophy	Malformation syndrome	Rare bone disease
534	Oculocerebrorenal syndrome of Lowe	Lowe disease|Lowe oculo-cerebro-renal dystrophy|Lowe oculo-cerebro-renal syndrome|Lowe oculocerebrorenal dystrophy|Lowe syndrome|OCRL|Phosphatidylinositol 4,5-biphosphate 5-phosphatase deficiency	Malformation syndrome	Rare developmental defect during embryogenesis
562	McCune-Albright syndrome	Gonadotropin-independent female-limited sexual precocity	Disease	Rare bone disease
618	Familial melanoma		Disease	Rare neoplastic disease
726	Alpers-Huttenlocher syndrome	Alpers progressive sclerosing poliodystrophy|Alpers syndrome|Progressive neuronal degeneration of childhood with liver disease	Disease	Rare inborn errors of metabolism
790	Retinoblastoma		Disease	Rare neoplastic disease
818	Smith-Lemli-Opitz syndrome	7-dehydrocholesterol reductase deficiency|RSH syndrome|SLOS	Malformation syndrome	Rare developmental defect during embryogenesis
839	Congenital nephrotic syndrome, Finnish type	Finnish congenital nephrosis	Disease	Rare renal disease
846	Alpha-thalassemia		Disease	Rare hematologic disease

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|Sensorineural hearing loss with imperforate anus and hypoplastic thumbs|TBS|Townes synd	Malformation syndrome	Rare developmental defect during embryogenesis
861	Treacher-Collins syndrome	Franceschetti-Klein syndrome|Mandibulofacial dysostosis without limb anomalies	Malformation syndrome	Rare developmental defect during embryogenesis
888	Van der Woude syndrome	Cleft lip/palate with mucous cysts of lower lip|Lip-pit syndrome|VWS	Malformation syndrome	Rare developmental defect during embryogenesis
1020	Early-onset autosomal dominant Alzheimer disease	EOFAD|Early-onset familial autosomal dominant Alzheimer disease|Familial Alzheimer disease	Disease	Rare neurologic disease
1306	Buschke-Ollendorff syndrome	Disseminated dermatofibrosis with osteopoikilosis	Malformation syndrome	Rare bone disease
1328	Camurati-Engelmann disease	Progressive diaphyseal dysplasia	Malformation syndrome	Rare bone disease
1598	Monosomy 18p	18p- syndrome|De Grouchy syndrome	Disease	Rare developmental defect during embryogenesis
1600	Monosomy 18q	18q deletion syndrome|18q- syndrome|Deletion 18q	Malformation syndrome	Rare developmental defect during embryogenesis
1799	Familial developmental dysphasia	Billard-Toutain-Maheut syndrome|FOXP2-associated dysphasia	Clinical syndrome	Rare neurologic disease
1885	Isolated ectopia lentis	Ectopia lentis syndrome|Familial ectopia lentis	Malformation syndrome	Rare ophthalmic disorder
2032	Idiopathic pulmonary fibrosis	IPF	Disease	Rare respiratory disease

2440	Isolated split hand-split foot malformation	Ectrodactyly|SHFM|Split hand foot malformation	Malformation syndrome	Rare developmental defect during embryogenesis
2526	Microcephaly-lymphedema-chorioretinopathy syndrome	MLCRD	Malformation syndrome	Rare developmental defect during embryogenesis
2578	Mayer-Rokitansky-Küster-Hauser syndrome type 2	Atypical MRKH syndrome|MRKH syndrome type 2|MURCS association|Müllerian duct aplasia-renal dysplasia-cervical somite anomalies syndrome	Clinical subtype	Rare developmental defect during embryogenesis
2710	Oculodentodigital dysplasia	Meyer-Schwickerath syndrome|ODDD syndrome|Oculodentoosseous dysplasia	Malformation syndrome	Rare developmental defect during embryogenesis
2721	Odonto-onycho-dermal dysplasia	OODD	Disease	Rare developmental defect during embryogenesis
2869	Peutz-Jeghers syndrome	Hamartomatous intestinal polyposis|PJS|Polyps and spots syndrome	Disease	Rare gastroenterologic disease
3138	Ulnar-mammary syndrome	Pallister ulnar-mammary syndrome|Schinzel syndrome|UMS	Malformation syndrome	Rare developmental defect during embryogenesis
3157	Septo-optic dysplasia spectrum	De Morsier syndrome|SOD|Septo-optic dysplasia	Malformation syndrome	Rare developmental defect during embryogenesis
3306	Inverted duplicated chromosome 15 syndrome	Duplication/inversion 15q11|Inv dup (15) syndrome|Isodicentric chromosome 15 syndrome|Non-distal tetrasomy 15q|Non-telomeric tetrasomy 15q|idic (15) syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3307	Tetrasomy 18p	Isochromosome 18p	Malformation syndrome	Rare developmental defect during embryogenesis

3440	Waardenburg syndrome		Disease	Rare developmental defect during embryogenesis
3463	Wolfram syndrome	DIDMOAD syndrome|Diabetes insipidus-diabetes mellitus-optic atrophy-deafness syndrome|Diabetes insipidus-diabetes mellitus-optic atrophy-hearing loss syndrome	Disease	Rare endocrine disease
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	Disease	Rare neoplastic disease
33069	Dravet syndrome	SMEI|Severe myoclonic epilepsy of infancy|Severe myoclonus epilepsy of infancy	Disease	Rare neurologic disease
36367	Distal monosomy 1q	Distal deletion 1q|Monosomy 1qter|Telomeric deletion 1q	Malformation syndrome	Rare developmental defect during embryogenesis
69663	Low phospholipid-associated cholelithiasis	ABCB4-related cholelithiasis|LPAC	Disease	Rare hepatic disease
77293	Niemann-Pick disease type B		Disease	Rare inborn errors of metabolism
79253	Mild phenylketonuria	Mild PKU|Variant PKU|Variant phenylketonuria|mPKU	Clinical subtype	Rare inborn errors of metabolism
79278	Autosomal erythropoietic protoporphyria	EPP	Disease	Rare inborn errors of metabolism
79282	Methylmalonic acidemia with homocystinuria, type cblC	CblC defect|Cobalamin C defect|Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cblC|Methylmalonic aciduria with homocystinuria, type cblC	Clinical subtype	Rare inborn errors of metabolism

79305	Progressive familial intrahepatic cholestasis type 3	PFIC3	Clinical subtype	Rare hepatic disease
79394	Congenital non-bullous ichthyosiform erythroderma	CIE|Erythrodermic ichthyosis|Non-bullous congenital ichthyosiform erythroderma	Disease	Rare skin disease
88616	Autosomal recessive non-syndromic intellectual disability	AR-NSID|NS-ARID	Etiological subtype	Rare neurologic disease
90653	Stickler syndrome type 1		Clinical subtype	Rare bone disease
91024	Autosomal recessive axonal hereditary motor and sensory neuropathy	AR-CMT2|Autosomal recessive axonal Charcot-Marie-Tooth disease type 2	Clinical group	0
93100	Renal agenesis, unilateral		Clinical subtype	Rare developmental defect during embryogenesis
93314	Spondylometaphyseal dysplasia, Kozlowski type		Disease	Rare bone disease
93545	Renal or urinary tract malformation	CAKUT|Congenital anomalies of kidney and urinary tract	Category	0
96061	Mosaic trisomy 8	Mosaic trisomy chromosome 8|Trisomy 8 mosaicism|Warkany syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
98306	Familial partial lipodystrophy	FPLD	Clinical group	0
98754	Prader-Willi syndrome due to maternal uniparental disomy of chromosome 15	UPD(15)mat	Etiological subtype	Rare developmental defect during embryogenesis
98896	Duchenne muscular dystrophy	DMD|Severe dystrophinopathy, Duchenne type	Disease	Rare neurologic disease
98913	Postsynaptic congenital myasthenic syndromes		Etiological subtype	Rare neurologic disease
99413	Turner syndrome due to structural X chromosome anomalies		Etiological subtype	Rare developmental defect during embryogenesis

99810	Familial porencephaly		Etiological subtype	Rare developmental defect during embryogenesis
100050	Hereditary angioedema type 1	HAE 1|HAE-I|Hereditary angioneurotic edema type 1	Etiological subtype	Rare systemic or rheumatologic disease
163956	X-linked intellectual disability, Nascimento type	X-linked intellectual disability-nail dystrophy-seizures syndrome	Disease	Rare developmental defect during embryogenesis
169189	Autosomal dominant centronuclear myopathy	AD-CNM	Disease	Rare neurologic disease
217340	17q21.31 microduplication syndrome	Dup(17)(q21.31)|Trisomy 17q21.31	Malformation syndrome	Rare developmental defect during embryogenesis
217607	Familial dilated cardiomyopathy		Category	0
247806	APC-related attenuated familial adenomatous polyposis	APC-related AFAP|APC-related attenuated FAP|APC-related attenuated familial polyposis coli	Clinical subtype	Rare gastroenterologic disease
250908	Rare neoplastic disease	Rare tumoral disease	Category	0
261204	16p11.2p12.2 microduplication syndrome	Dup(16)(p11.2p12.2)|Trisomy 16p11.2p12.2	Malformation syndrome	Rare developmental defect during embryogenesis
261236	16p13.11 microdeletion syndrome	Del(16)(p13.11)|Monosomy 16p13.11	Malformation syndrome	Rare developmental defect during embryogenesis
261243	16p13.11 microduplication syndrome	Dup(16)(p13.11)|Trisomy 16p13.11	Malformation syndrome	Rare developmental defect during embryogenesis
261337	Distal 22q11.2 microduplication syndrome	Distal dup(22)(q11.2)|Distal trisomy 22q11.2	Malformation syndrome	Rare developmental defect during embryogenesis
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	Category	0

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	Category	0
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	Category	0
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	Category	0
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	Category	0
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	Category	0
281097	Autosomal recessive congenital ichthyosis	ARCI	Clinical group	0

293910	Familial isolated arrhythmogenic ventricular dysplasia, right dominant form	Familial isolated arrhythmogenic ventricular cardiomyopathy, classic form|Familial isolated arrhythmogenic ventricular cardiomyopathy, right dominant form|Familial isolated arrhythmogenic ventricular dysplasia, classic form	Clinical subtype	Rare cardiac disease
293939	Distal Xq28 microduplication syndrome	Distal dup(X)q(28)|Distal trisomy Xq28|Int22h1/Int22h2 mediated-Xq28 microduplication syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
19	2-hydroxyglutaric aciduria	2-hydroxyglutaric acidemia	Clinical group	0
25	Glutaryl-CoA dehydrogenase deficiency	GA1|GCDHD|Glutaric acidemia type 1|Glutaric aciduria type 1|Glutaryl-coenzyme A dehydrogenase deficiency	Disease	Rare inborn errors of metabolism
33	Isovaleric acidemia	Isovaleric acid CoA dehydrogenase deficiency	Disease	Rare inborn errors of metabolism
99	Autosomal dominant cerebellar ataxia	ADCA|Autosomal dominant spinocerebellar ataxia	Category	0
107	BOR syndrome	Branchiootorenal syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
109	Bannayan-Riley-Ruvalcaba syndrome	BRRS|Myhre-Riley-Smith syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
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	Disease	Rare hematologic disease

125	Bloom syndrome	BSyn	Disease	Rare developmental defect during embryogenesis
157	Carnitine palmitoyltransferase II deficiency	CPT2|CPTII|Carnitine palmitoyltransferase deficiency type 2	Disease	Rare inborn errors of metabolism
158	Systemic primary carnitine deficiency	CDSP|CUD|Carnitine transporter defect|Carnitine uptake deficiency|Deficiency of plasma-membrane carnitine transporter|SPCD	Disease	Rare inborn errors of metabolism
192	Coffin-Lowry syndrome	CLS	Malformation syndrome	Rare developmental defect during embryogenesis
207	Crouzon disease	Crouzon craniofacial dysostosis	Malformation syndrome	Rare developmental defect during embryogenesis
225	Maternally-inherited diabetes and deafness	MIDD|Maternally-inherited diabetes and hearing loss|Mitochondrial diabetes	Disease	Rare inborn errors of metabolism
296	Ollier disease	Dyschondroplasia	Disease	Rare bone disease
327	Congenital factor VII deficiency	Congenital proconvertin deficiency|Hypoproconvertinemia	Disease	Rare hematologic disease
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 glyco	Disease	Rare inborn errors of metabolism
393	46,XX testicular disorder of sex development	46,XX testicular DSD|De la Chapelle syndrome|XX, male syndrome	Malformation syndrome	Rare developmental defect during embryogenesis

405	Familial hypocalciuric hypercalcemia	FBH|FBHH|FHH|Familial benign hypercalcemia|Familial benign hypocalciuric hypercalcemia	Disease	Rare endocrine disease
427	Familial hypoaldosteronism		Disease	Rare endocrine disease
475	Joubert syndrome	CPD IV|Cerebelloparenchymal disorder IV|Classic Joubert syndrome|Joubert syndrome type A|Joubert-Boltshauser syndrome|Pure Joubert syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
502	Trichorhinophalangeal syndrome type 2	Deletion 8q24.1|Langer-Giedion syndrome|Monosomy 8q24.1	Malformation syndrome	Rare developmental defect during embryogenesis
512	Metachromatic leukodystrophy	Arylsulfatase A deficiency|MLD	Disease	Rare neurologic disease
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|Heredit	Disease	Rare neoplastic disease
536	Systemic lupus erythematosus	Disseminated lupus erythematosus|SLE	Disease	
569	Familial or sporadic hemiplegic migraine		Disease	Rare neurologic disease
593	Myofibrillar myopathy		Category	0
606	Proximal myotonic myopathy	Myotonic dystrophy type 2|Proximal myotonic dystrophy|Ricker disease|Ricker syndrome	Disease	Rare neurologic disease

614	Thomsen and Becker disease	Myotonia congenita	Disease	Rare neurologic disease
627	Nance-Horan syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
635	Neuroblastoma		Disease	Rare neoplastic disease
644	NARP syndrome	Neurogenic muscle weakness-ataxia-retinitis pigmentosa syndrome|Neuropathy-ataxia-retinitis pigmentosa syndrome	Disease	Rare neurologic disease
661	Ondine syndrome	CCHS|Central congenital hypoventilation syndrome|Congenital central alveolar hypoventilation syndrome|Ondine curse	Disease	Rare neurologic disease
676	Hereditary chronic pancreatitis		Disease	Rare gastroenterologic disease
705	Pendred syndrome	Goiter-deafness syndrome|Goiter-hearing loss syndrome	Malformation syndrome	Rare otorhinolaryngologic disease
741	Familial mitral valve prolapse		Morphological anomaly	Rare developmental defect during embryogenesis
743	Severe hereditary thrombophilia due to congenital protein S deficiency	Autosomal recessive thrombophilia due to congenital protein S deficiency	Disease	Rare hematologic disease
766	Hemolytic anemia due to red cell pyruvate kinase deficiency	Pyruvate kinase deficiency of erythrocytes	Disease	Rare hematologic disease
884	Tetrasomy 12p	Isochromosome 12p mosaicism|Isochromosome 12p syndrome|Pallister-Killian syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
897	Waardenburg-Shah syndrome	Shah-Waardenburg syndrome|WS4|Waardenburg syndrome type 4|Waardenburg-Hirschsprung syndrome	Disease	Rare developmental defect during embryogenesis

905	Wilson disease	Hepatolenticular degeneration	Disease	Rare hepatic disease
955	Hajdu-Cheney syndrome	Acroosteolysis dominant type|Acroosteolysis with osteoporosis and changes in skull and mandible|Arthrodentoosteodysplasia|Cheney syndrome	Malformation syndrome	Rare bone disease
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	Malformation syndrome	Rare developmental defect during embryogenesis
1031	Enamel-renal syndrome	Amelogenesis imperfecta-nephrocalcinosis syndrome	Malformation syndrome	Rare renal disease
1114	Aplasia cutis congenita		Malformation syndrome	Rare skin disease
1359	Carney complex	Carney syndrome|Myxoma-spotty pigmentation-endocrine overactivity syndrome	Disease	Rare endocrine disease
1519	SPECC1L-related hypertelorism syndrome	Brachycephalofrontonasal dysplasia|Teebi hypertelorism syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
1552	Currarino syndrome	Currarino triad	Malformation syndrome	Rare developmental defect during embryogenesis
1568	X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
1570	Symbrachydactyly of hands and feet	De Smet-Fabry-Fryns syndrome	Malformation syndrome	Rare developmental defect during embryogenesis

1636	Distal monosomy 7q36	Distal deletion 7q36|Monosomy 7qter|Telomeric deletion 7q36	Malformation syndrome	Rare developmental defect during embryogenesis
1797	Autosomal dominant spondylocostal dysostosis	Autosomal dominant spondylocostal dysplasia	Malformation syndrome	Rare developmental defect during embryogenesis
1872	Cone rod dystrophy		Disease	Rare ophthalmic disorder
1942	Myoclonic-astatic epilepsy	Doose syndrome|EMAS|Epilepsy with myoclonic-astatic seizures|Epilepsy with myoclonic-atonic seizures|MAE|Myoclonic atonic epilepsy|Myoclonic-astatic epilepsy in early childhood	Disease	Rare neurologic disease
1949	Benign familial neonatal epilepsy	BFNS|Benign familial neonatal convulsions|Benign familial neonatal seizures	Disease	Rare neurologic disease
2014	Cleft palate		Clinical group	0
2028	Juvenile hyaline fibromatosis	Murray-Puretic-Drescher syndrome|Puretic syndrome	Clinical subtype	Rare bone disease
2044	Floating-Harbor syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
2065	Galloway-Mowat syndrome	Galloway syndrome|Microcephaly-hiatus hernia-nephrotic syndrome|Nephrosis-neuronal dysmigration syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2073	Narcolepsy type 1	Gélineau disease|Narcolepsy-cataplexy	Disease	Rare neurologic disease
2108	Hallermann-Streiff syndrome	François dyscephalic syndrome|Oculomandibulofacial syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2128	Isolated hemihyperplasia	Hemi 3 syndrome|Hemicorporal hypertrophy|Isolated hemihypertrophy	Morphological anomaly	Rare developmental defect during embryogenesis

2228	Hypodontia-dysplasia of nails syndrome	Hypodontia-nail dysgenesis syndrome|Tooth and nail syndrome|Witkop syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2308	Jacobsen syndrome	Del(11)(q23.3)|Del(11)(qter)|Distal deletion 11q|Distal monosomy 11q|Monosomy 11qter|Telomeric deletion 11q	Malformation syndrome	Rare developmental defect during embryogenesis
2346	Angioosteohypertrophic syndrome	Klippel-Trénaunay-Weber syndrome	Disease	Rare developmental defect during embryogenesis
2492	FATCO syndrome	Fibular aplasia-tibial campomelia-oligosyndactyly syndrome|Hecht-Scott syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2616	3M syndrome	3-M syndrome|Yakut short stature syndrome	Malformation syndrome	Rare bone disease
2701	Noonan syndrome-like disorder with loose anagen hair	Mazzanti syndrome|NS/LAH	Malformation syndrome	Rare developmental defect during embryogenesis
2841	Familial benign chronic pemphigus	Benign chronic familial pemphigus of Hailey-Hailey|Hailey-Hailey disease	Disease	Rare skin disease
2856	Persistent Müllerian duct syndrome	PMDS|Persistent Müllerian derivatives	Malformation syndrome	Rare developmental defect during embryogenesis
2884	Piebaldism		Disease	Rare skin disease
2953	Musculocontractural Ehlers-Danlos syndrome	Adducted thumb-clubfoot syndrome|Distal arthrogryposis with peculiar facies and hydronephrosis|Dündar syndrome|Ehlers-Danlos syndrome, Kosho type|Musculocontractural EDS|mcEDS	Disease	Rare systemic or rheumatologic disease

3047	Blepharophimosis-intellectual disability syndrome, SBBYS type	Hypothyroidism-dysmorphism-postaxial polydactyly-intellectual disability syndrome|SBBYS variant of Ohdo syndrome|SBBYSS|Say-Barber-Biesecker-Young-Simpson syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3051	Nicolaides-Baraitser syndrome	Intellectual disability-sparse hair-brachydactyly syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3077	X-linked intellectual disability-psychosis-macroorchidism syndrome	Lindsay-Burn syndrome|PPM-X	Malformation syndrome	Rare developmental defect during embryogenesis
3156	Senior-Loken syndrome	Nephronophthisis with retinal dystrophy|Renal dysplasia-retinal aplasia syndrome|SLSN	Disease	Rare renal disease
3206	Stüve-Wiedemann syndrome	Neonatal Schwartz-Jampel syndrome|SJS2|Schwartz-Jampel syndrome type 2|Stüve-Wiedemann dysplasia	Malformation syndrome	Rare bone disease
3220	Deafness-enamel hypoplasia-nail defects syndrome	Hearing loss-enamel hypoplasia-nail defects syndrome|Heimler syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3242	Renpenning syndrome	X-linked intellectual disability due to PQBP1 mutations|X-linked intellectual disability, Renpenning type	Malformation syndrome	Rare developmental defect during embryogenesis
3261	Autoimmune lymphoproliferative syndrome	ALPS|Canale-Smith syndrome	Disease	Rare immune disease
3337	Primary Fanconi renotubular syndrome	DeToni-Debré-Fanconi syndrome|Primary Fanconi renal syndrome	Disease	Rare renal disease

3366	Isolated trigonocephaly	Non-syndromic metopic craniosynostosis	Morphological anomaly	Rare developmental defect during embryogenesis
3378	Trisomy 13	Patau syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
32960	Tumor necrosis factor receptor 1 associated periodic syndrome	Familial Hibernian fever|TNF receptor 1-associated periodic syndrome|TRAPS syndrome	Disease	Rare systemic or rheumatologic disease
34515	FKRP-related limb-girdle muscular dystrophy R9	Autosomal recessive limb-girdle muscular dystrophy type 2I|FKRP-related LGMD R9|LGMD due to FKRP deficiency|LGMD type 2I|LGMD2I|Limb-girdle muscular dystrophy due to FKRP deficiency|Limb-girdle muscular dystrophy type 2I	Disease	Rare neurologic disease
35099	Isolated brachycephaly	Non-syndromic bicoronal synostosis	Morphological anomaly	Rare developmental defect during embryogenesis
37553	Andersen-Tawil syndrome	Andersen syndrome|LQT7|Long QT syndrome type 7	Disease	Rare neurologic disease
42775	PHACE syndrome	PHACES syndrome|Pascual-Castroviejo syndrome type 2	Malformation syndrome	Rare developmental defect during embryogenesis
48471	Lissencephaly		Category	0
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	Disease	Rare neurologic disease
53271	Muenke syndrome		Malformation syndrome	Rare developmental defect during embryogenesis

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	Clinical group	0
68363	Rare dystonia	Rare dystonic disorder	Category	0
68402	Rare parkinsonian disorder	Rare hypokinetic movement disorder	Category	0
69076	Familial renal glucosuria	Familial renal glycosuria|SGLT2 deficiency	Disease	Rare renal disease
71529	Obesity due to melanocortin 4 receptor deficiency	MC4R deficiency	Etiological subtype	Rare endocrine disease
77828	Genetic obesity		Category	0
79239	Classic galactosemia	GALT deficiency|Galactose-1-phosphate uridyltransferase deficiency|Galactosemia type 1|	Disease	Rare inborn errors of metabolism
79255	GM1 gangliosidosis type 1	Infantile GM1 gangliosidosis|Norman-Landing disease	Clinical subtype	Rare inborn errors of metabolism
79314	L-2-hydroxyglutaric aciduria	L-2-HGA|L-2-hydroxyglutaric acidemia	Disease	Rare inborn errors of metabolism
79404	Severe generalized junctional epidermolysis bullosa	Epidermolysis bullosa letalis|JEB-H|Junctional epidermolysis bullosa generalisata gravis|Junctional epidermolysis bullosa, Herlitz type|Junctional epidermolysis bullosa, Herlitz-Pearson type|Severe generalized JEB	Disease	Rare skin disease
79435	Oculocutaneous albinism type 4	OCA4	Disease	Rare skin disease

79501	Punctate palmoplantar keratoderma type 1	Buschke-Fischer-Brauer syndrome|Keratodermia palmoplantaris papulosa, Buschke-Fischer-Brauer type|PPKP1	Disease	Rare skin disease
84081	Senior-Boichis syndrome	Boichis disease|Nephronophthisis-hepatic fibrosis syndrome	Disease	Rare renal disease
85193	Idiopathic juvenile osteoporosis	IJO|Juvenile osteoporosis	Malformation syndrome	Rare bone disease
85277	X-linked intellectual disability, Cantagrel type		Malformation syndrome	Rare developmental defect during embryogenesis
85279	Syndromic X-linked intellectual disability due to JARID1C mutation		Malformation syndrome	Rare developmental defect during embryogenesis
88661	Amelogenesis imperfecta		Disease	Rare odontologic disease
88924	Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis	PKDTS|TSC2/PKD1 contiguous gene syndrome|Tuberous sclerosis/polycystic kidney disease contiguous gene syndrome	Disease	Rare developmental defect during embryogenesis
89838	Autosomal recessive generalized epidermolysis bullosa simplex	Autosomal recessive generalized EBS	Disease	Rare skin disease
89936	X-linked hypophosphatemia	X-linked hypophosphatemic rickets|XLH	Disease	Rare endocrine disease
90348	Autosomal dominant cutis laxa	ADCL	Disease	Rare skin disease
93101	Renal hypoplasia		Morphological anomaly	Rare developmental defect during embryogenesis
93256	Fragile X-associated tremor/ataxia syndrome	FXTAS syndrome	Malformation syndrome	Rare neurologic disease
93276	Polyostotic fibrous dysplasia		Clinical subtype	Rare bone disease
93315	Spondylometaphyseal dysplasia, 'corner fracture' type	Spondylometaphyseal dysplasia, Sutcliffe type	Disease	Rare bone disease

93328	Autosomal dominant omodysplasia		Clinical subtype	Rare bone disease
93419	Rare bone disease		Category	0
93460	Overgrowth syndrome		Category	0
93473	Hurler syndrome	Hurler disease|MPS1H|MPSIH|Mucopolysaccharidosis type 1H|Mucopolysaccharidosis type IH	Clinical subtype	Rare inborn errors of metabolism
93547	Syndromic renal or urinary tract malformation		Category	0
93573	Thrombotic microangiopathy	TMA	Clinical group	0
93603	Rare renal tubular disease		Category	0
94064	Deafness-infertility syndrome	DIS|Hearing loss-infertility syndrome	Malformation syndrome	Rare otorhinolaryngologic disease
94083	Partington syndrome	Partington-Mulley syndrome|X-linked intellectual disability-dystonia-dysarthria syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
96102	Distal trisomy 10q	Distal duplication 10q|Telomeric duplication 10q|Trisomy 10qter	Malformation syndrome	Rare developmental defect during embryogenesis
96125	Distal monosomy 6p	6p subtelomeric deletion syndrome|6p25 microdeletion syndrome|Distal deletion 6p|Monosomy 6p25	Malformation syndrome	Rare developmental defect during embryogenesis
96129	Distal monosomy 19p13.3	Distal deletion 19p|Telomeric deletion 19p	Malformation syndrome	Rare developmental defect during embryogenesis
96148	Distal monosomy 10q	Distal deletion 10q|Monosomy 10qter|Telomeric deletion 10q	Malformation syndrome	Rare developmental defect during embryogenesis
96169	Koolen-De Vries syndrome	KdVS	Malformation syndrome	Rare developmental defect during embryogenesis

96170	Emanuel syndrome	Der(22)t(11;22) syndrome|Supernumerary der(22) syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
96334	Kagami-Ogata syndrome due to paternal uniparental disomy of chromosome 14	UPD(14)pat	Etiological subtype	Rare developmental defect during embryogenesis
97978	Rare endocrine disease		Category	0
98033	Rare neurologic disease with psychiatric involvement		Category	0
98760	Spinocerebellar ataxia type 8	SCA8	Disease	Rare neurologic disease
98764	Spinocerebellar ataxia type 27	SCA27	Disease	Rare neurologic disease
98784	Autosomal dominant nocturnal frontal lobe epilepsy	ADNFLE|Autosomal dominant sleep-related hypermotor epilepsy	Disease	Rare neurologic disease
98793	Prader-Willi syndrome due to paternal 15q11q13 deletion		Etiological subtype	Rare developmental defect during embryogenesis
98794	Angelman syndrome due to maternal 15q11q13 deletion	Angelman syndrome due to maternal monosomy 15q11q13	Etiological subtype	Rare developmental defect during embryogenesis
98809	Paroxysmal kinesigenic dyskinesia	Familial PKD|Familial paroxysmal kinesigenic dyskinesia|Paroxysmal kinesigenic choreathetosis	Disease	Rare neurologic disease
98878	Hemophilia A	FVIII deficiency|Factor VIII deficiency	Disease	Rare hematologic disease
99741	King-Denborough syndrome	Koussef-Nichols syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
99796	Subcortical band heterotopia	Subcortical laminar heterotopia	Morphological anomaly	Rare developmental defect during embryogenesis
99943	Autosomal dominant Charcot-Marie-Tooth disease type 2J	CMT2J	Disease	Rare neurologic disease

101088	X-linked hyper-IgM syndrome	HIGM1|Hyper-IgM syndrome due to CD40 ligand deficiency|Hyper-IgM syndrome due to CD40L deficiency|Hyper-IgM syndrome type 1|XHIGM	Clinical subtype	Rare immune disease
101351	Familial isolated congenital asplenia		Morphological anomaly	Rare developmental defect during embryogenesis
101998	Rare epilepsy		Category	0
156643	Genetic endocrine growth disease		Category	0
169186	Autosomal recessive centronuclear myopathy	AR-CNM	Disease	Rare neurologic disease
169793	Severe hemophilia B	Severe factor IX deficiency	Clinical subtype	Rare hematologic disease
171848	Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome	PHARC syndrome|Peripheral neuropathy, Fiskerstrand type|Polyneuropathy-deafness-ataxia-retinitis pigmentosa-cataract syndrome	Disease	Rare neurologic disease
177910	Prader-Willi syndrome due to imprinting mutation		Etiological subtype	Rare developmental defect during embryogenesis
183763	Rare genetic syndromic intellectual disability		Category	0
199306	Cleft lip/palate	Alveolar cleft lip and palate|Cleft lip and palate|Cleft lip-alveolus-palate syndrome|FLP	Morphological anomaly	Rare developmental defect during embryogenesis
206656	Non-dystrophic myopathy		Category	0
216820	Osteogenesis imperfecta type 4	OI type 4	Clinical subtype	Rare bone disease
216828	Osteogenesis imperfecta type 5	OI type 5	Clinical subtype	Rare bone disease
217595	Syndrome associated with hypertrophic cardiomyopathy		Category	0

217656	Familial isolated arrhythmogenic right ventricular dysplasia	Familial isolated ARVC|Familial isolated ARVD|Familial isolated arrhythmogenic right ventricular cardiomyopathy|Familial isolated arrhythmogenic ventricular cardiomyopathy|Familial isolated arrhythmogenic ventricular dysplasia	Disease	Rare cardiac disease
220460	Attenuated familial adenomatous polyposis	AFAP|Attenuated FAP|Attenuated familial polyposis coli	Disease	Rare gastroenterologic disease
228302	Carnitine palmitoyl transferase II deficiency, myopathic form	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 d	Clinical subtype	Rare inborn errors of metabolism
228402	2q23.1 microdeletion syndrome	Del(2)(q23.1)|Monosomy 2q23.1|	Malformation syndrome	Rare developmental defect during embryogenesis
230839	Classical-like Ehlers-Danlos syndrome type 1	Classical-like EDS type 1|Ehlers-Danlos syndrome due to tenascin-X deficiency|clEDS type 1	Disease	Rare systemic or rheumatologic disease
231178	Usher syndrome type 2	USH2	Clinical subtype	Rare developmental defect during embryogenesis
238666	Isolated congenital hypogonadotropic hypogonadism	Gonadotropic deficiency|Isolated congenital gonadotropin deficiency|Isolated gonadotropin-releasing hormone deficiency|	Disease	0

238769	1q44 microdeletion syndrome	Del(1)(q44)|Monosomy 1q44	Malformation syndrome	Rare developmental defect during embryogenesis
251038	3q29 microduplication syndrome	Trisomy 3q29	Malformation syndrome	Rare developmental defect during embryogenesis
251076	8p23.1 duplication syndrome	Dup(8)(p23.1p23.1)|Trisomy 8p23.1	Malformation syndrome	Rare developmental defect during embryogenesis
252183	Neurofibroma		Disease	Rare neoplastic disease
254892	Autosomal dominant progressive external ophthalmoplegia	adPEO	Disease	Rare inborn errors of metabolism
261476	Xp21 deletion syndrome	Complex GKD|Complex glycerol kinase deficiency|Del(X)(p21)|Xp21 contiguous gene deletion syndrome|Xp21 microdeletion syndrome	Disease	Rare developmental defect during embryogenesis
261552	Mowat-Wilson syndrome due to a ZEB2 point mutation	Hirschsprung disease and intellectual disability due to a ZEB2 point mutation	Etiological subtype	Rare developmental defect during embryogenesis
261652	Kleefstra syndrome due to a point mutation		Etiological subtype	Rare developmental defect during embryogenesis
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	Category	0
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	Category	0

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	Category	0
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	Category	0
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	Category	0
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	Category	0
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	Category	0
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	Category	0

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	Category	0
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	Category	0
275864	Behavioral variant of frontotemporal dementia	bv-FTD	Disease	Rare neurologic disease
276413	10q22.3q23.3 microdeletion syndrome	Del(10)(q22.3q23.3)|Deletion 10q22.3q23.3|Monosomy 10q22.3q23.3	Malformation syndrome	Rare developmental defect during embryogenesis
281210	X-linked ichthyosis syndrome		Clinical group	0
289266	Early-onset epileptic encephalopathy and intellectual disability due to GRIN2A mutation		Disease	Rare developmental defect during embryogenesis
306661	Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome	Hypercalcemic tumoral calcinosis	Clinical subtype	Rare skin disease
9	Tetrasomy X	48,XXXX syndrome|Quadruple X|Tetra X	Malformation syndrome	Rare developmental defect during embryogenesis
10	48,XXYY syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
17	Fatal infantile lactic acidosis with methylmalonic aciduria		Disease	Rare inborn errors of metabolism
20	3-hydroxy-3-methylglutaric aciduria	3-hydroxy-3-methylglutaryl-CoA lyase deficiency|HMG-CoA lyase deficiency|Hydroxymethylglutaric aciduria	Disease	Rare inborn errors of metabolism

27	Vitamin B12-unresponsive methylmalonic acidemia	Methylmalonyl-CoA mutase deficiency|Methylmalonyl-Coenzyme A mutase deficiency|Vitamin B12-unresponsive methylmalonic aciduria	Disease	Rare inborn errors of metabolism
37	Acrodermatitis enteropathica	AEZ|Acrodermatitis enteropathica, zinc deficiency type|Inherited zinc deficiency	Disease	Rare skin disease
40	Acromesomelic dysplasia, Maroteaux type		Malformation syndrome	Rare bone disease
43	X-linked adrenoleukodystrophy	ALD|X-ALD|X-linked ALD	Disease	Rare inborn errors of metabolism
47	X-linked agammaglobulinemia	BTK-deficiency|Bruton type agammaglobulinemia	Clinical subtype	Rare immune disease
48	Congenital bilateral absence of vas deferens	Congenital bilateral agenesis of vas deferens|Congenital bilateral aplasia of vas deferens	Morphological anomaly	Rare developmental defect during embryogenesis
59	Allan-Herndon-Dudley syndrome		Disease	Rare neurologic disease
70	Proximal spinal muscular atrophy	SMA	Disease	Rare neurologic disease
82	Hereditary thrombophilia due to congenital antithrombin deficiency	Hereditary thrombophilia due to congenital antithrombin 3 deficiency	Disease	Rare hematologic disease
114	Auriculoosteodysplasia		Malformation syndrome	Rare bone disease
117	Behçet disease		Disease	Rare systemic or rheumatologic disease
127	Borjeson-Forssman-Lehmann syndrome	BFLS|Intellectual disability-epilepsy-endocrine disorders syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
140	Campomelic dysplasia	Campomelic dwarfism	Malformation syndrome	Rare bone disease
162	Cataract-glaucoma syndrome		Malformation syndrome	Rare ophthalmic disorder

167	Chédiak-Higashi syndrome	Chédiak-Higashi disease|Chédiak-Higashi-Steinbrink syndrome	Disease	Rare immune disease
180	Choroideremia	CHM|Tapetochoroidal dystrophy	Disease	Rare ophthalmic disorder
195	Cat-eye syndrome	CES	Malformation syndrome	Rare developmental defect during embryogenesis
198	Occipital horn syndrome		Disease	Rare inborn errors of metabolism
211	Familial cylindromatosis	Turban tumor syndrome	Clinical subtype	Rare skin disease
218	Darier disease	Darier-White disease|Keratosis follicularis	Disease	Rare skin disease
220	Denys-Drash syndrome	Drash syndrome|Wilms tumor-DSD syndrome|Wilms tumor-disorder of sex development syndrome	Disease	Rare renal disease
229	Familial aortic dissection	Annuloaortic ectasia|Cystic medial necrosis of aorta	Disease	Rare circulatory system disease
233	Duane retraction syndrome	DRS|DURS|Duane syndrome|Stilling-Turk-Duane syndrome	Malformation syndrome	Rare ophthalmic disorder
236	Trisomy 9p	Duplication 9p|Duplication of the short arm of chromosome 9|Trisomy of the short arm of chromosome 9	Malformation syndrome	Rare developmental defect during embryogenesis
249	Fibrous dysplasia of bone		Malformation syndrome	Rare bone disease
254	Spondylometaphyseal dysplasia		Clinical group	0
262	Duchenne and Becker muscular dystrophy	Severe dystrophinopathy, Duchenne and Becker type	Clinical group	0
270	Oculopharyngeal muscular dystrophy	OPMD	Disease	Rare neurologic 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, Athaba	Disease	Rare immune disease
282	Frontotemporal dementia	FTD	Clinical group	0
300	Bifunctional enzyme deficiency		Disease	Rare inborn errors of metabolism
303	Dystrophic epidermolysis bullosa	DEB|Dermolytic epidermolysis bullosa|Epidermolysis bullosa dystrophica||Epidermolysis bullosa atrophicans|JEB	Clinical group	0
308	Unverricht-Lundborg disease	PME type 1|Progressive myoclonic epilepsy type 1|Progressive myoclonus epilepsy type 1|ULD	Malformation syndrome	Rare neurologic disease
329	Congenital factor XI deficiency	Hemophilia C|PTA deficiency|Plasma thromboplastin antecedent deficiency|Rosenthal factor deficiency|Rosenthal syndrome	Disease	Rare hematologic disease
337	Fibrodysplasia ossificans progressiva	FOP|Myositis ossificans progressiva|Stone man syndrome	Disease	Rare bone disease

365	Glycogen storage disease due to acid maltase deficiency	Alpha-1,4-glucosidase acid deficiency|GSD due to acid maltase deficiency|GSD type 2|GSD type II|Glycogen storage disease type 2|Glycogen storage disease type II|Glycogenosis due to acid maltase deficiency|Glycogenosis type 2|Glycogenosis type II|Pompe dis	Disease	Rare inborn errors of metabolism
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	Malformation syndrome	Rare developmental defect during embryogenesis
389	Langerhans cell histiocytosis	Histiocytosis X|Langerhans cell granulomatosis	Disease	0
395	Homocystinuria due to methylene tetrahydrofolate reductase deficiency	MTHFR deficiency|Methylene tetrahydrofolate reductase deficiency	Disease	Rare inborn errors of metabolism
414	Gyrate atrophy of choroid and retina	HOGA|Hyperornithinemia|Hyperornithinemia-gyrate atrophy of choroid and retina syndrome|Ornithine aminotransferase deficiency	Disease	Rare ophthalmic disorder
442	Congenital hypothyroidism		Category	0
447	Paroxysmal nocturnal hemoglobinuria	Marchiafava-Micheli disease|PNH	Disease	Rare hematologic disease
467	Non-acquired combined pituitary hormone deficiency	Congenital combined pituitary hormone deficiency|Congenital hypopituitarism	Category	0

477	KID syndrome	Ichthyosis hystrix Rheydt type|KID/HID syndrome|Keratitis-ichthyosis-deafness/Hystrix-like ichthyosis-deafness syndrome|Keratitis-ichthyosis-hearing loss/Hystrix-like ichthyosis-hearing loss syndrome|Senter syndrome	Disease	Rare developmental defect during embryogenesis
480	Kearns-Sayre syndrome		Disease	Rare inborn errors of metabolism
486	Autosomal dominant severe congenital neutropenia		Disease	Rare immune disease
551	MERRF	Fukuhara syndrome|Myoclonus epilepsy associated with ragged-red fibres	Disease	Rare neurologic disease
570	Moebius syndrome	Möbius syndrome	Disease	Rare developmental defect during embryogenesis
580	Mucopolysaccharidosis type 2	Hunter syndrome|Iduronate 2-sulfatase deficiency|MPS2|MPSII|Mucopolysaccharidosis type II	Disease	Rare inborn errors of metabolism
581	Mucopolysaccharidosis type 3	MPS3|MPSIII|Mucopolysaccharidosis type III|Sanfilippo disease	Disease	Rare inborn errors of metabolism
584	Mucopolysaccharidosis type 7	Beta-glucuronidase deficiency|MPS7|MPSVII|Mucopolysaccharidosis type VII|Sly disease	Disease	Rare inborn errors of metabolism
587	Muir-Torre syndrome	Multiple keratoacanthoma, Muir-Torre type	Disease	Rare neoplastic disease
597	Central core disease		Disease	Rare neurologic disease
599	Distal myopathy	Distal muscular dystrophy	Category	0
611	Inclusion body myositis	IBM|Sporadic inclusion body myositis|sIBM	Disease	Rare neurologic disease

621	Hereditary methemoglobinemia	Autosomal recessive methemoglobinemia|Congenital methemoglobinemia	Disease	Rare hematologic disease
631	Non-acquired isolated growth hormone deficiency	Congenital IGHD|Congenital isolated GH deficiency|Congenital isolated growth hormone deficiency	Disease	Rare endocrine disease
646	Niemann-Pick disease type C		Disease	Rare inborn errors of metabolism
653	Multiple endocrine neoplasia type 2	MEN2	Disease	Rare neoplastic disease
663	Mitochondrial DNA-related progressive external ophthalmoplegia	Maternally-inherited CPEO|Maternally-inherited chronic progressive external ophthalmoplegia|mtDNA-related progressive external ophthalmoplegia	Disease	Rare inborn errors of metabolism
681	Hypokalemic periodic paralysis	Westphall disease	Disease	Rare neurologic disease
702	Pelizaeus-Merzbacher disease	Diffuse familial brain sclerosis|PMD|Pelizaeus-Merzbacher brain sclerosis|Sudanophilic leukodystrophy, Paelizeus-Merzbacher type	Disease	Rare neurologic disease
708	Peters anomaly	Peters congenital glaucoma	Morphological anomaly	Rare developmental defect during embryogenesis
710	Pfeiffer syndrome	ACS5|Acrocephalosyndactyly type 5	Malformation syndrome	Rare developmental defect during embryogenesis
713	Glycogen storage disease due to phosphoglycerate kinase 1 deficiency	GSD due to phosphoglycerate kinase 1 deficiency|Glycogenosis due to phosphoglycerate kinase 1 deficiency	Disease	Rare inborn errors of metabolism

744	Proteus syndrome	Partial gigantism-nevi-hemihypertrophy-macrocephaly syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
753	46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency	46,XY DSD due to 5-alpha-reductase 2 deficiency|Pseudovaginal perineoscrotal hypospadias|Steroid 5-alpha-reductase deficiency	Disease	Rare developmental defect during embryogenesis
760	Purine nucleoside phosphorylase deficiency	PNP deficiency|PNPase deficiency	Disease	Rare immune disease
761	Immunoglobulin A vasculitis	Anaphylactoid purpura|Henoch-Schönlein purpura|IgA vasculitis|Purpura rheumatica|Rheumatoid purpura	Disease	Rare systemic or rheumatologic disease
767	Polyarteritis nodosa	Küssmaul-Maier disease|PAN|Periarteritis nodosa	Disease	Rare systemic or rheumatologic disease
776	Lujan-Fryns syndrome	X-linked intellectual disability with marfanoid habitus	Malformation syndrome	Rare developmental defect during embryogenesis
792	X-linked retinoschisis	X-linked juvenile retinoschisis|XLRS	Malformation syndrome	Rare ophthalmic disorder
823	Isolated spina bifida		Clinical group	0
893	WAGR syndrome	Del(11)(p13)|Deletion 11p13|Monosomy 11p13|Wilms tumor-aniridia-genitourinary anomalies-intellectual disability syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
894	Waardenburg syndrome type 1	WS1|Waardenburg syndrome type I	Clinical subtype	Rare developmental defect during embryogenesis
895	Waardenburg syndrome type 2	WS2|Waardenburg syndrome type II	Clinical subtype	Rare developmental defect during embryogenesis

909	Cerebrotendinous xanthomatosis	CTX|Sterol 27-hydroxylase deficiency	Disease	Rare inborn errors of metabolism
910	Xeroderma pigmentosum		Disease	Rare skin disease
912	Zellweger syndrome	Cerebrohepatorenal syndrome|ZS	Disease	Rare inborn errors of metabolism
959	Acro-renal-ocular syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
1018	X-linked Alport syndrome-diffuse leiomyomatosis	Xq22.3 microdeletion syndrome	Clinical subtype	Rare renal disease
1143	Neurogenic arthrogryposis multiplex congenita		Disease	Rare developmental defect during embryogenesis
1147	Sheldon-Hall syndrome	Distal arthrogryposis type 2B|Freeman-Sheldon syndrome variant	Malformation syndrome	Rare developmental defect during embryogenesis
1154	Arthrogryposis-oculomotor limitation-electroretinal anomalies syndrome	Distal arthrogryposis type 5|Distal arthrogryposis type IIB|Distal arthrogryposis with ophthalmoplegia|Oculomelic amyoplasia	Malformation syndrome	Rare developmental defect during embryogenesis
1159	Progressive pseudorheumatoid arthropathy of childhood	Spondyloepiphyseal dysplasia tarda-progressive arthropathy syndrome	Disease	Rare bone disease
1166	Congenital unilateral hypoplasia of depressor anguli oris	Isolated asymmetric crying facies	Morphological anomaly	Rare developmental defect during embryogenesis
1180	Ataxia-hypogonadism-choroidal dystrophy syndrome	Boucher-Neuhäuser syndrome	Disease	Rare developmental defect during embryogenesis
1190	Atelosteogenesis type I	AO1|AOI|Atelosteogenesis type 1|Giant cell chondrodysplasia|Spondylo-humero-femoral dysplasia	Malformation syndrome	Rare bone disease
1199	Esophageal atresia		Morphological anomaly	Rare developmental defect during embryogenesis

1214	Progressive hemifacial atrophy	Hemifacial atrophy|PHA|Parry-Romberg syndrome|Progressive facial hemiatrophy|Romberg syndrome	Disease	Rare neurologic disease
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	Disease	Rare ophthalmic disorder
1276	Brachydactyly-arterial hypertension syndrome	Bilginturan brachydactyly|Bilginturan syndrome|Brachydactyly type E, with short stature and hypertension	Malformation syndrome	Rare developmental defect during embryogenesis
1310	Caffey disease	Infantile cortical hyperostosis	Malformation syndrome	Rare bone disease
1331	Familial prostate cancer		Disease	Rare neoplastic disease
1425	Desbuquois syndrome	DBQD|Desbuquois dysplasia	Malformation syndrome	Rare bone disease
1428	Familial chondromalacia patellae		Disease	Rare developmental defect during embryogenesis
1442	Ring chromosome 18 syndrome	Ring 18|Ring chromosome 18	Malformation syndrome	Rare developmental defect during embryogenesis
1444	Ring chromosome 20 syndrome	Ring 20|Ring chromosome 20	Malformation syndrome	Rare developmental defect during embryogenesis
1446	Ring chromosome 22 syndrome	Ring 22|Ring chromosome 22|r(22) syndrome	Malformation syndrome	Rare developmental defect during embryogenesis

1457	Aorta coarctation		Morphological anomaly	Rare developmental defect during embryogenesis
1475	Renal coloboma syndrome	Coloboma of optic nerve with renal disease|Papillo-renal syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
1478	Interatrial communication	ASD|Atrial septal defect|Interauricular communication	Morphological anomaly	Rare developmental defect during embryogenesis
1496	Corpus callosum agenesis-neuronopathy syndrome	Andermann syndrome|Charlevoix disease	Disease	Rare neurologic disease
1505	Short rib-polydactyly syndrome		Clinical group	0
1520	Craniofrontonasal dysplasia	CFND|CFNS|Craniofrontonasal syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
1522	Craniometaphyseal dysplasia		Malformation syndrome	Rare bone disease
1571	Knobloch syndrome	Knobloch-Layer syndrome|Retinal detachment-occipital encephalocele syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
1572	Common variable immunodeficiency	CVID|Idiopathic immunoglobulin deficiency|Primary antibody deficiency|Primary hypogammaglobulinemia	Disease	Rare immune disease
1573	Hypotrichosis with juvenile macular degeneration	HJMD|Hypotrichosis with juvenile macular dystrophy	Malformation syndrome	Rare ophthalmic disorder
1590	Distal monosomy 13q	13q32 deletion|Deletion 13q32|Distal 13q deletion|Monosomy 13q32|Telomeric deletion13q	Malformation syndrome	Rare developmental defect during embryogenesis
1642	Distal monosomy 9p	Distal deletion 9p|Monosomy 9pter|Telomeric deletion 9p	Malformation syndrome	Rare developmental defect during embryogenesis
1646	Partial chromosome Y deletion	Male sterility due to chromosome Y deletion	Malformation syndrome	Rare infertility

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	Disease	Rare renal disease
1708	Mosaic trisomy 16	Mosaic trisomy chromosome 16|Trisomy 16 mosaicism	Malformation syndrome	Rare developmental defect during embryogenesis
1717	Distal trisomy 19q	Distal duplication 19q|Telomeric duplication 19q|Trisomy 19qter	Malformation syndrome	Rare developmental defect during embryogenesis
1756	Caudal duplication		Malformation syndrome	Rare developmental defect during embryogenesis
1775	Dyskeratosis congenita	DC|DKC|Zinsser-Engman-Cole syndrome	Disease	Rare immune disease
1826	Frontometaphyseal dysplasia		Disease	Rare bone disease
1851	Multicystic dysplastic kidney	MCDK|Multicystic renal dysplasia	Morphological anomaly	Rare renal disease
1860	Thanatophoric dysplasia type 1	TD1|Thanatophoric dwarfism type 1	Clinical subtype	Rare bone disease
1880	Ebstein malformation of the tricuspid valve	Ebstein anomaly of the tricuspid valve	Morphological anomaly	Rare developmental defect during embryogenesis
1896	EEC syndrome	Ectrodactyly-ectodermal dysplasia-cleft lip/palate syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
1899	Arthrochalasia Ehlers-Danlos syndrome	Arthrochalasia EDS|Arthrochalasis multiplex congenita|EDS VII|Ehlers-Danlos syndrome type 7|Ehlers-Danlos syndrome, arthrochalasia type|aEDS	Disease	Rare systemic or rheumatologic disease

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	Disease	Rare neurologic disease
1980	Bilateral striopallidodentate calcinosis	BSPDC|Cerebrovascular ferrocalcinosis|Idiopathic basal ganglia calcification|PFBC|Primary familial brain calcification	Disease	Rare neurologic disease
1997	Blepharo-cheilo-odontic syndrome	BCD syndrome|Blepharocheilodontic syndrome|Clefting-ectropion-conical teeth syndrome|Ectropion inferior-cleft lip and/or palate syndrome|Elschnig syndrome|Lagophthalmia-cleft lip and palate syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2019	Femur-fibula-ulna complex	FFU complex|Femur-fibula-ulna dysostosis|Femur-fibula-ulna syndrome|PFFD	Malformation syndrome	Rare developmental defect during embryogenesis
2052	Fraser syndrome	Cryptophthalmos-syndactyly syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2148	Lissencephaly type 1 due to doublecortin gene mutation	X-linked lissencephaly type 1	Disease	Rare developmental defect during embryogenesis
2149	Nodular neuronal heterotopia		Morphological anomaly	Rare developmental defect during embryogenesis

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	Disease	Rare skin disease
2237	Hypoparathyroidism-sensorineural deafness-renal disease syndrome	Barakat syndrome|HDR syndrome|Hypoparathyroidism-sensorineural hearing loss-renal disease syndrome	Malformation syndrome	Rare renal disease
2254	Pontocerebellar hypoplasia type 1	Norman disease|PCH1	Malformation syndrome	Rare developmental defect during embryogenesis
2315	Johanson-Blizzard syndrome	JBS	Malformation syndrome	Rare developmental defect during embryogenesis
2379	Early-onset parkinsonism-intellectual disability syndrome	Laxova-Opitz syndrome|Waisman syndrome	Disease	Rare developmental defect during embryogenesis
2412	Dislocation of the hip-dysmorphism syndrome	Collins-Pope syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2477	Megalencephaly	Macroencephaly	Malformation syndrome	Rare developmental defect during embryogenesis
2499	Metachondromatosis		Malformation syndrome	Rare bone disease
2542	Isolated microphthalmia-anophthalmia-coloboma	Isolated anophthalmia-microphthalmia syndrome	Clinical group	0
2637	Microcephalic osteodysplastic primordial dwarfism type II	MOPD type II|Majewski osteodysplastic primordial dwarfism type II	Malformation syndrome	Rare bone disease

2662	Keipert syndrome	Nasodigitoacoustic syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2671	Neu-Laxova syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
2707	Oculocerebrofacial syndrome, Kaufman type		Malformation syndrome	Rare developmental defect during embryogenesis
2712	Oculofaciocardiodental syndrome	Cataract-microphthalmia-radiculomegaly-cardiac septal defect syndrome|OFCD syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2750	Orofaciodigital syndrome type 1	OFD1|OFDI|OFDSI|Oral-facial-digital syndrome type 1|Papillon-Léage-Psaume syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2771	Bruck syndrome	Osteogenesis imperfecta-congenital joint contractures syndrome	Malformation syndrome	Rare bone disease
2780	Osteopathia striata-cranial sclerosis syndrome	Hyperostosis generalisata with striations|Robinow-Unger syndrome	Malformation syndrome	Rare bone disease
2781	Osteopetrosis and related disorders		Clinical group	0
2788	Osteoporosis-pseudoglioma syndrome	OPPG|Ocular form of osteogenesis imperfecta	Disease	Rare bone disease
2822	Autosomal recessive spastic paraplegia type 11	Nakamura-Osame syndrome|SPG11|Spastic paraplegia-intellectual disability-thin corpus callosum syndrome	Disease	Rare neurologic disease
2836	PEHO syndrome	Progressive encephalopathy with edema, hypsarrhythmia and optic atrophy|Progressive encephalopathy-optic atrophy syndrome	Disease	Rare neurologic disease

2875	Phakomatosis pigmentovascularis		Disease	Rare developmental defect during embryogenesis
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	Malformation syndrome	Rare developmental defect during embryogenesis
2897	Pityriasis rubra pilaris		Disease	Rare skin disease
2909	Rothmund-Thomson syndrome	Poikiloderma of Rothmund-Thomson|RTS	Disease	Rare developmental defect during embryogenesis
2940	Porencephaly		Disease	Rare developmental defect during embryogenesis
2990	Autosomal recessive multiple pterygium syndrome	Autosomal recessive non-lethal multiple pterygium syndrome|EVMPS|Escobar syndrome|Escobar variant multiple pterygium syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
2995	Baraitser-Winter cerebrofrontofacial syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
3027	Caudal regression sequence	Caudal dysplasia|Sacral agenesis syndrome|Sacral regression syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3042	Intellectual disability-cataracts-calcified pinnae-myopathy syndrome	Primrose syndrome	Malformation syndrome	Rare neurologic disease
3057	Monoamine oxidase A deficiency	Brunner syndrome	Disease	Rare inborn errors of metabolism

3103	Roberts syndrome	Pseudothalidomide syndrome|Roberts-SC phocomelia syndrome|SC phocomelia|SC pseudothalidomide syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3109	Mayer-Rokitansky-Küster-Hauser syndrome	MRKH syndrome|Rokitansky syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
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	Disease	Rare endocrine disease
3181	Sprengel deformity	High scapula	Morphological anomaly	Rare developmental defect during embryogenesis
3197	Hereditary hyperekplexia	Congenital stiff man syndrome|Familial startle disease|Hereditary hyperexplexia|Kok disease|Stiff baby syndrome	Disease	Rare neurologic disease
3205	Sturge-Weber syndrome	Encephalofacial angiomatosis|Encephalotrigeminal angiomatosis|SWS|Sturge-Weber-Dimitri syndrome|Sturge-Weber-Krabbe angiomatosis|Sturge-Weber-Krabbe syndrome	Malformation syndrome	Rare developmental defect during embryogenesis

3238	Cardiospondylocarpofacial syndrome	Forney syndrome|Forney-Robinson-Pascoe syndrome|Mitral regurgitation-deafness-skeletal anomalies syndrome|Mitral regurgitation-hearing loss-skeletal anomalies syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3275	Spondylocarpotarsal synostosis	Synspondylism	Malformation syndrome	Rare bone disease
3280	Syringomyelia	Hydromyelia	Clinical group	Rare neurologic disease
3309	Tetrasomy 5p	Isochromosome 5p	Malformation syndrome	Rare developmental defect during embryogenesis
3310	Tetrasomy 9p	Isochromosome 9p	Malformation syndrome	Rare developmental defect during embryogenesis
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 hemime	Malformation syndrome	Rare developmental defect during embryogenesis
3339	Toriello-Lacassie-Droste syndrome	Aplasia cutis congenita-epibulbar dermoids syndrome|Oculoectodermal syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
3377	Trismus-pseudocamptodactyly syndrome	Distal arthrogryposis type 7|Dutch-Kentucky syndrome|Hecht syndrome|Hecht-Beals syndrome	Malformation syndrome	Rare developmental defect during embryogenesis

3454	Intellectual disability-developmental delay-contractures syndrome	Foot contractures-muscle atrophy-oculomotor apraxia syndrome|Wieacker-Wolff syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
26791	Multiple acyl-CoA dehydrogenase deficiency	Glutaric acidemia type 2|Glutaric aciduria type 2|MAD deficiency|MADD	Disease	Rare inborn errors of metabolism
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	Disease	Rare inborn errors of metabolism
30391	Isolated biliary atresia	Isolated atresia of bile ducts|Non-syndromic biliary atresia	Morphological anomaly	Rare developmental defect during embryogenesis
30924	Primary hypomagnesemia with secondary hypocalcemia	HOMG1|HSH|Hypomagnesemia caused by selective magnesium malabsorption|Hypomagnesemia intestinal type 1|Intestinal hypomagnesemia with secondary hypocalcemia|PHSH	Disease	Rare renal disease
30925	Hereditary central diabetes insipidus	Hereditary CDI|Hereditary neurogenic diabetes insipidus	Clinical subtype	Rare endocrine disease
33574	Glutamate-cysteine ligase deficiency	Gamma-glutamylcysteine synthetase deficiency	Disease	Rare hematologic disease
34526	Genetic primary hypomagnesemia		Category	0
35098	Isolated plagiocephaly	Non-syndromic unicoronal synostosis|Synostotic plagiocephaly	Morphological anomaly	Rare developmental defect during embryogenesis

35122	Congenital sucrase-isomaltase deficiency	CSID|Congenital sucrose intolerance|Disaccharide intolerance	Disease	Rare gastroenterologic disease
35125	Epidermal nevus syndrome	Epidermal hamartoma syndrome	Disease	Rare developmental defect during embryogenesis
35173	X-linked dominant chondrodysplasia punctata	CDPX2|CDPXD|CPXD|Chondrodystrophia calcificans congenita|Conradi-Hünermann-Happle syndrome|X-linked chondrodysplasia punctata type 2	Disease	Rare bone disease
35656	Coenzyme Q10 deficiency	CoQ10 deficiency	Clinical group	0
35696	Mitochondrial disorder due to a defect in mitochondrial protein synthesis	COXPD|Combined OXPHOS defect|Combined OXPHOS deficiency|Combined oxidative phosphorylation defect	Category	0
35705	Neurometabolic disorder due to serine deficiency	Serine deficiency	Category	0
35708	Aromatic L-amino acid decarboxylase deficiency	AADC deficiency	Disease	Rare inborn errors of metabolism
35858	Imerslund-Gräsbeck syndrome	Familial megaloblastic anemia|Selective cobalamin malabsorption with proteinuria	Disease	Rare hematologic disease
36899	Myoclonus-dystonia syndrome	Alcohol-responsive dystonia|Hereditary essential myoclonus|Myoclonic dystonia	Disease	Rare neurologic disease
39041	Omenn syndrome	Combined immunodeficiency with hypereosinophilia	Disease	Rare immune disease
45358	Congenital fibrosis of extraocular muscles	FEOM	Disease	Rare ophthalmic disorder
45448	Miyoshi myopathy		Disease	Rare neurologic disease

47044	Hereditary papillary renal cell carcinoma	HPRCC	Disease	Rare neoplastic disease
48431	Congenital cataracts-facial dysmorphism-neuropathy syndrome	CCFDN	Malformation syndrome	Rare developmental defect during embryogenesis
50942	Striate palmoplantar keratoderma	Keratosis palmoplantaris striata|Keratosis palmoplantaris striata et areata|Keratosis palmoplantaris varians of Wachters	Disease	Rare skin disease
51083	Familial short QT syndrome	SQTS	Disease	Rare cardiac disease
52503	X-linked creatine transporter deficiency	Creatine transporter deficiency|SLC6A8 deficiency	Disease	Rare inborn errors of metabolism
54260	Left ventricular noncompaction	LVNC|Left ventricular hypertrabeculation|Spongy myocardium	Disease	Rare cardiac disease
57146	Rare hepatic disease		Category	0
64545	Benign idiopathic neonatal seizures	BINS|Benign nonfamilial neonatal seizures	Disease	Rare neurologic disease
64745	Pruritic urticarial papules and plaques of pregnancy	PUPPP|Polymorphic eruption of pregnancy	Disease	Rare skin disease
64747	X-linked Charcot-Marie-Tooth disease	CMTX|X-linked hereditary motor and sensory neuropathy	Clinical group	0
64749	Charcot-Marie-Tooth disease type 4	AR-CMT1|Autosomal recessive demyelinating Charcot-Marie-Tooth|CMT4	Clinical group	0
64753	Spinocerebellar ataxia with axonal neuropathy type 2	AOA2|Ataxia-oculomotor apraxia type 2|SCAN 2|SCAR1	Disease	Rare neurologic disease
64755	Becker nevus syndrome	Pigmentary hairy epidermal nevus	Disease	Rare skin disease
66628	Obesity due to congenital leptin deficiency		Etiological subtype	Rare endocrine disease
68361	Rare deafness	Rare hearing loss	Category	0

68367	Rare inborn errors of metabolism	Rare metabolic disease	Category	0
68380	Mitochondrial disease		Category	0
68381	Neuromuscular disease		Category	0
68416	Rare infectious disease		Category	0
69061	Idiopathic steroid-sensitive nephrotic syndrome		Clinical syndrome	Rare renal disease
69665	Intrahepatic cholestasis of pregnancy	Gravidic intrahepatic cholestasis|Pregnancy-related cholestasis|Recurrent intrahepatic cholestasis of pregnancy	Disease	Rare hepatic disease
70592	Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency	IRAK4 deficiency	Disease	Rare immune disease
70594	Dopa-responsive dystonia due to sepiapterin reductase deficiency	Autosomal recessive sepiapterin reductase-deficient DRD|DRD due to SRD|SPR deficiency|Sepiapterin reductase deficiency	Disease	Rare neurologic disease
70595	Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome	SANDO	Disease	Rare neurologic disease
71274	Disseminated peritoneal leiomyomatosis	DPL|Diffuse peritoneal leiomyomatosis|LPD|Leiomyomatosis peritonealis disseminate	Disease	Rare neoplastic disease
71277	Classic glucose transporter type 1 deficiency syndrome	Classic GLUT1 deficiency syndrome|Classic GLUT1-DS|De Vivo disease|Encephalopathy due to GLUT1 deficiency	Disease	Rare neurologic disease
71290	Familial platelet disorder with associated myeloid malignancy		Disease	Rare hematologic disease
71859	Rare genetic neurological disorder		Category	0
71862	Inherited retinal disorder	Retinal dystrophy	Category	0

73273	Growth delay due to insulin-like growth factor I resistance	Resistance to IGF-1	Disease	Rare endocrine disease
75249	Familial isolated restrictive cardiomyopathy	Familial or idiopathic restrictive cardiomyopathy	Disease	Rare cardiac disease
77259	Gaucher disease type 1	Non-cerebral juvenile Gaucher disease	Clinical subtype	Rare inborn errors of metabolism
79083	PPARG-related familial partial lipodystrophy	FPLD3|Familial partial lipodystrophy type 3|PPARG-related FPLD	Disease	Rare endocrine disease
79146	Familial progressive hyperpigmentation	Melanosis diffusa congenita|Melanosis universalis hereditaria|Universal melanosis	Disease	Rare skin disease
79163	Classic organic aciduria		Category	0
79167	Disorder of urea cycle metabolism and ammonia detoxification		Category	0
79230	Hemochromatosis type 2	Juvenile hemochromatosis	Disease	Rare hepatic disease
79269	Sanfilippo syndrome type A	Heparan sulfamidase deficiency|MPS3A|MPSIIIA|Mucopolysaccharidosis type 3A|Mucopolysaccharidosis type IIIA	Etiological subtype	Rare inborn errors of metabolism
79270	Sanfilippo syndrome type B	MPS3B|MPSIIIB|Mucopolysaccharidosis type 3B|Mucopolysaccharidosis type IIIB|N-acetyl-alpha-glucosaminidase deficiency	Etiological subtype	Rare inborn errors of metabolism
79276	Acute intermittent porphyria		Disease	Rare inborn errors of metabolism

79318	PMM2-CDG	CDG syndrome type Ia|CDG-Ia|CDG1A|Carbohydrate deficient glycoprotein syndrome type Ia|Congenital disorder of glycosylation type 1a|Congenital disorder of glycosylation type Ia|Phosphomannomutase 2 deficiency	Disease	Rare inborn errors of metabolism
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	Disease	Rare inborn errors of metabolism
79361	Inherited epidermolysis bullosa	Epidermolysis bullosa hereditaria|Hereditary epidermolysis bullosa	Category	0
79397	Epidermolysis bullosa simplex with mottled pigmentation	EBS with mottled pigmentation|EBS-MP	Disease	Rare skin disease
79430	Hermansky-Pudlak syndrome	HPS	Disease	Rare immune disease
79431	Oculocutaneous albinism type 1A	OCA1A|Tyrosinase-negative oculocutaneous albinism	Clinical subtype	Rare skin disease
79434	Oculocutaneous albinism type 1B	OCA1B|Oculocutaneous albinism, Amish type|Platinum oculocutaneous albinism|Yellow oculocutaneous albinism	Clinical subtype	Rare skin disease
79445	Pseudopseudohypoparathyroidism	AHO-PPHP syndrome|Albright hereditary osteodystrophy-PPHP syndrome	Disease	Rare endocrine disease
79452	Milroy disease	Hereditary lymphedema type I|Nonne-Milroy lymphedema	Disease	Rare circulatory system disease

79457	Maculopapular cutaneous mastocytosis	Urticaria pigmentosa	Disease	Rare skin disease
79490	Microcystic lymphatic malformation	Capillary lymphangioma|Capillary lymphatic malformation|Cutaneous lymphangioma circumscriptum|Microcystic infiltrating lymphatic malformation|Microcystic lymphangioma|Superficial lymphangioma|Superficial lymphatic malformation	Malformation syndrome	Rare developmental defect during embryogenesis
79493	Brooke-Spiegler syndrome	CYLD cutaneous syndrome	Disease	Rare skin disease
79651	Mild hyperphenylalaninemia	Mild HPA|Non-PKU HPA|mHPA	Clinical subtype	Rare inborn errors of metabolism
83330	Proximal spinal muscular atrophy type 1	Infantile spinal muscular atrophy|Infantile-onset spinal muscular atrophy|SMA type 1|SMA type I|SMA-I|SMA1|Werdnig-Hoffmann disease	Clinical subtype	Rare neurologic disease
83419	Proximal spinal muscular atrophy type 3	Juvenile spinal muscular atrophy|Kugelberg-Welander disease|SMA type 3|SMA type III|SMA-III|SMA3	Clinical subtype	Rare neurologic disease
83463	Microtia		Morphological anomaly	Rare developmental defect during embryogenesis

84064	Syndromic diarrhea	Phenotypic diarrhea|SD/THE|Syndromic diarrhea/Tricho-hepato-enteric syndrome|Tricho-hepato-enteric syndrome|Trichohepatoenteric syndrome	Disease	Rare gastroenterologic disease
85128	Bothnia retinal dystrophy	Västerbotten dystrophy	Disease	Rare ophthalmic disorder
85167	Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome	SMD-CRD	Disease	Rare bone disease
85200	Ischiovertebral syndrome	Ischiospinal dysostosis|Ischiovertebral dysplasia	Malformation syndrome	Rare developmental defect during embryogenesis
85329	X-linked intellectual disability-hypotonia-facial dysmorphism-aggressive behavior syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
85451	ATTRV122I amyloidosis	ATTR cardiomyopathy|ATTRV122I-related amyloidosis|TTR-related amyloid cardiomyopathy|TTR-related cardiac amyloidosis|Transthyretin amyloid cardiopathy|Transthyretin-related familial amyloid cardiomyopathy	Disease	Rare cardiac disease
86923	Hereditary palmoplantar keratoderma, Gamborg-Nielsen type	Hereditary palmoplantar hyperkeratosis, Gamborg-Nielsen type|PPK, Gamborg-Nielsen type	Disease	Rare skin disease
88632	Anterior segment developmental anomaly	Anterior segment dysgenesis	Category	Rare developmental defect during embryogenesis
88637	Hypomyelination-hypogonadotropic hypogonadism-hypodontia syndrome	4H syndrome	Clinical subtype	Rare neurologic disease

88642	Channelopathy-associated congenital insensitivity to pain	Channelopathy-associated CIP	Disease	Rare neurologic disease
88949	MUC1-related 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	Clinical subtype	Rare renal disease
88950	UMOD-related autosomal dominant tubulointerstitial kidney disease	ADTKD-UMOD|Autosomal dominant medullary cystic kidney disease type 2|Familial juvenile hyperuricemic nephropathy type 1|MCKD2|UMOD-related ADTKD|Uromodulin kidney disease|Uromodulin-associated kidney disease	Clinical subtype	Rare renal disease
90117	Hereditary motor and sensory neuropathy, Okinawa type	HMSNP|Hereditary motor and sensory neuropathy, proximal type	Disease	Rare neurologic disease
90308	Klippel-Trénaunay syndrome		Clinical subtype	Rare developmental defect during embryogenesis
90342	Xeroderma pigmentosum variant	XPV	Disease	Rare skin disease
90625	X-linked non-syndromic sensorineural deafness type DFN	X-linked isolated neurosensory deafness type DFN|X-linked isolated neurosensory hearing loss type DFN|X-linked isolated sensorineural deafness type DFN|X-linked isolated sensorineural hearing loss type DFN|X-linked non-syndromic neurosensory deafness type	Etiological subtype	Rare otorhinolaryngologic disease

90641	Mitochondrial non-syndromic sensorineural deafness	Isolated mitochondrial neurosensory deafness|Isolated mitochondrial neurosensory hearing loss|Isolated mitochondrial sensorineural deafness|Isolated mitochondrial sensorineural hearing loss|Mitochondrial non-syndromic neurosensory deafness|Mitochondrial n	Etiological subtype	Rare otorhinolaryngologic disease
90654	Stickler syndrome type 2		Clinical subtype	Rare bone disease
90771	Disorder of sex development	DSD	Category	0
90794	Classic congenital adrenal hyperplasia due to 21-hydroxylase deficiency	Classic 21-OHD CAH	Disease	Rare developmental defect during embryogenesis
90795	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	CAH due to 11-beta-hydroxylase deficiency|CYP11B1 deficiency	Disease	Rare developmental defect during embryogenesis
91359	Chronic pneumonitis of infancy	CPI	Disease	Rare respiratory disease
93114	Autosomal dominant intermediate Charcot-Marie-Tooth disease type E	CMTDIE|Charcot-Marie-Tooth disease-nephropathy syndrome	Disease	Rare neurologic disease
93271	Short rib-polydactyly syndrome, Verma-Naumoff type	Short rib-polydactyly syndrome type 3	Malformation syndrome	Rare bone disease
93279	Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis		Disease	Rare bone disease
93283	Spondyloepiphyseal dysplasia, Kimberley type		Disease	Rare bone disease
93284	Spondyloepiphyseal dysplasia tarda		Disease	Rare bone disease
93293	Okihiro syndrome	Duane-radial ray syndrome	Malformation syndrome	Rare developmental defect during embryogenesis

93304	Autosomal dominant brachyolmia	Brachyolmia type 3	Malformation syndrome	Rare bone disease
93311	Multiple epiphyseal dysplasia type 5	BHMED|Bilateral hereditary micro-epiphyseal dysplasia|EDM5|MED5|Polyepiphyseal dysplasia type 5	Disease	Rare bone disease
93338	Polysyndactyly	PPD4|Preaxial polydactyly type 4	Morphological anomaly	Rare developmental defect during embryogenesis
93360	Spondyloepimetaphyseal dysplasia with multiple dislocations	SEMD-MD|SEMDJL2|Spondyloepimetaphyseal dysplasia with joint laxicity, Hall type|Spondyloepimetaphyseal dysplasia with joint laxity type 2|Spondyloepimetaphyseal dysplasia with joint laxity, leptodactylic type|Spondyloepimetaphyseal dysplasia with multiple	Disease	Rare bone disease
93372	Familial hypocalciuric hypercalcemia type 1	FHH type 1	Etiological subtype	Rare endocrine disease
93383	Brachydactyly type B		Malformation syndrome	Rare developmental defect during embryogenesis
93388	Brachydactyly type A1	Brachydactyly, Farabee type	Malformation syndrome	Rare developmental defect during embryogenesis
93437	Acromesomelic dysplasia		Clinical group	0
93442	Chondrodysplasia punctata	CDP	Clinical group	0
93454	Dysostosis with predominant vertebral and costal involvement		Category	0
93457	Non-syndromic limb reduction defect	Non-syndromic limb hypoplasia	Category	0
93548	Glomerular disease		Category	0

93591	Infantile nephronophthisis	Autosomal recessive infantile NPHP|Autosomal recessive infantile nephronophthisis	Clinical subtype	Rare renal disease
93593	Nephropathy secondary to a storage or other metabolic disease		Category	0
93598	Primary hyperoxaluria type 1	Glycolic aciduria|Peroxisomal alanine-glyoxylate aminotransferase deficiency	Clinical subtype	Rare renal disease
93600	Primary hyperoxaluria type 3		Clinical subtype	Rare renal disease
93613	Cystinuria type B		Etiological subtype	Rare renal disease
93616	Hemoglobin H disease	Alpha-thalassemia intermedia|HbH disease	Clinical subtype	Rare hematologic disease
93623	Dent disease type 2	Nephrolithiasis type 2	Clinical subtype	Rare renal disease
93626	Rare renal disease		Category	0
93924	Lobar holoprosencephaly		Clinical subtype	Rare developmental defect during embryogenesis
93929	Cloacal exstrophy	OEIS complex|Omphalocele-cloacal exstrophy-imperforate anus-spinal defect syndrome	Clinical subtype	Rare developmental defect during embryogenesis
93964	Blepharospasm-oromandibular dystonia syndrome	Meige dystonia|Meige syndrome	Disease	Rare neurologic disease
94065	15q24 microdeletion syndrome	Del(15)(q24)|Monosomy 15q24	Etiological subtype	Rare developmental defect during embryogenesis
94147	Spinocerebellar ataxia type 7	Ataxia with pigmentary retinopathy|Cerebellar syndrome-pigmentary maculopathy syndrome|SCA7	Disease	Rare neurologic disease

95494	Combined pituitary hormone deficiencies, genetic forms	Familial congenital hypopituitarism|Multiple pituitary hormone deficiencies, genetic forms	Disease	Rare endocrine disease
95496	Pituitary stalk interruption syndrome	Ectopic neurohypophysis|PSIS	Morphological anomaly	Rare endocrine disease
95702	X-linked adrenal hypoplasia congenita	X-linked AHC|X-linked congenital adrenal hypoplasia	Disease	Rare endocrine disease
95706	Posterior hypospadias	Hypospadias, severe form|Perineal, scrotal or penoscrotal hypospadias	Morphological anomaly	Rare developmental defect during embryogenesis
95716	Familial thyroid dyshormonogenesis	Thyroid dyshormonogenesis	Disease	Rare endocrine disease
96072	4p16.3 microduplication syndrome	Distal duplication 4p|Distal trisomy 4p|Telomeric duplication 4p|Trisomy 4pter	Malformation syndrome	Rare developmental defect during embryogenesis
96092	8p inverted duplication/deletion syndrome	Invdupdel(8p)|Inverted 8p duplication/deletion syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
96096	Distal trisomy 4q	Distal duplication 4q|Telomeric duplication 4q|Trisomy 4qter	Malformation syndrome	Rare developmental defect during embryogenesis
96145	Distal monosomy 4q	Distal deletion 4q|Monosomy 4qter|Telomeric deletion 4q	Malformation syndrome	Rare developmental defect during embryogenesis
96147	Kleefstra syndrome due to 9q34 microdeletion	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	Etiological subtype	Rare developmental defect during embryogenesis
96168	Monosomy 13q34	Del(13)(q34)|Distal deletion 13q34|Subtelomeric deletion 13q34	Malformation syndrome	Rare developmental defect during embryogenesis

96176	Ring chromosome 13 syndrome	Ring 13|Ring chromosome 13	Malformation syndrome	Rare developmental defect during embryogenesis
96181	Maternal uniparental disomy of chromosome 6	UPD(6)mat	Malformation syndrome	Rare developmental defect during embryogenesis
96182	Silver-Russell syndrome due to maternal uniparental disomy of chromosome 7	UPD(7)mat	Etiological subtype	Rare developmental defect during embryogenesis
96184	Temple syndrome due to maternal uniparental disomy of chromosome 14	UPD(14)mat	Etiological subtype	Rare developmental defect during embryogenesis
96193	Beckwith-Wiedemann syndrome due to paternal uniparental disomy of chromosome 11	Mosaic paternal uniparental disomy of chromosome 11|UPD(11)pat	Etiological subtype	Rare developmental defect during embryogenesis
96263	48,XXXY syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
96264	49,XXXXY syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
96346	Anorectal malformation	ARM	Category	0
97245	Congenital myopathy		Category	0
97297	Bohring-Opitz syndrome	BOS syndrome|Bohring syndrome|C-like syndrome|Oberklaid-Danks syndrome|Opitz trigonocephaly-like syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
97361	Renal hypoplasia, unilateral		Clinical subtype	Rare developmental defect during embryogenesis
97363	Unilateral multicystic dysplastic kidney	Unilateral MCDK|Unilateral multicystic renal dysplasia	Clinical subtype	Rare developmental defect during embryogenesis
97548	Right sided atrial isomerism	Isomerism of right atrial appendage|Ivemark syndrome|RAI	Malformation syndrome	Rare developmental defect during embryogenesis
97593	Pseudohypoparathyroidism		Category	0
97966	Rare ophthalmic disorder		Category	0

98048	Rare male infertility		Category	0
98057	Rare tumor	Rare neoplasm	Category	0
98132	Partial autosomal trisomy/tetrasomy		Category	0
98158	Chromosome Y structural anomaly		Category	0
98159	Chromosome X structural anomaly		Category	0
98249	Ehlers-Danlos syndrome	EDS	Clinical group	0
98258	Infantile epilepsy syndrome		Category	0
98267	Genetic non-syndromic obesity	Monogenic obesity due to a leptin-melanocortin pathway anomaly	Disease	0
98345	Rare idiopathic male infertility		Disease	Rare infertility
98473	Muscular dystrophy		Category	0
98668	Vitreoretinopathy		Category	0
98688	Oculomotor apraxia		Category	0
98756	Spinocerebellar ataxia type 2	SCA2	Disease	Rare neurologic disease
98766	Spinocerebellar ataxia type 5	SCA5	Disease	Rare neurologic disease
98769	Spinocerebellar ataxia type 15/16	SCA15/16	Disease	Rare neurologic disease
98791	Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16	ATR syndrome linked to chromosome 16|ATR syndrome, deletion type|ATR-16 syndrome|Alpha thalassemia-intellectual disability syndrome, deletion type	Malformation syndrome	Rare developmental defect during embryogenesis
98797	Isochromosomy Yp		Malformation syndrome	Rare developmental defect during embryogenesis

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	Disease	Rare neurologic disease
98820	Familial focal epilepsy with variable foci	FFEVF|Familial partial epilepsy with variable foci	Disease	Rare neurologic disease
98938	Colobomatous microphthalmia	MAC|Microphthalmia with colobomatous cyst|Microphthalmia-anophthalmia-coloboma syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
98942	Coloboma of choroid and retina		Morphological anomaly	Rare developmental defect during embryogenesis
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 c	Disease	Rare ophthalmic disorder
98969	Macular corneal dystrophy	Corneal dystrophy Groenouw type II|Fehr corneal dystrophy|MCD	Disease	Rare ophthalmic disorder
98976	Congenital glaucoma	Buphthalmia|Buphthalmos|Buphthalmus|Primary congenital glaucoma	Disease	Rare ophthalmic disorder

98994	Total early-onset cataract		Clinical subtype	Rare ophthalmic disorder
99015	Spastic paraplegia type 2	SPG2|Spastic gait type 2|Spastic paraparesis type 2|X-linked spastic paraplegia type 2	Disease	Rare neurologic disease
99361	Familial medullary thyroid carcinoma	Familial MTC	Disease	Rare neoplastic disease
99718	Leber plus disease	LHON plus disease	Disease	Rare ophthalmic disorder
99749	Kostmann syndrome	Infantile agranulocytosis|Severe congenital neutropenia type 3	Disease	Rare immune disease
99776	Mosaic trisomy 9	Mosaic trisomy chromosome 9|Trisomy 9 mosaicism	Malformation syndrome	Rare developmental defect during embryogenesis
99858	Idiopathic syringomyelia		Clinical subtype	Rare neurologic disease
100091	Adrenal/paraganglial tumor		Category	0
100973	FRAXE intellectual disability	Intellectual disability associated with fragile site FRAXE	Disease	Rare developmental defect during embryogenesis
100976	Bathing suit ichthyosis	BSI	Disease	Rare skin disease
100980	Autosomal dominant pure spastic paraplegia	Autosomal dominant pure HSP|Autosomal dominant pure SPG|Autosomal dominant uncomplicated HSP|Autosomal dominant uncomplicated SPG|Autosomal dominant uncomplicated spastic paraplegia	Clinical group	0
100984	Autosomal dominant spastic paraplegia type 3	Strümpell disease	Disease	Rare neurologic disease
100991	Autosomal dominant spastic paraplegia type 10	SPG10	Disease	Rare neurologic disease
101001	Autosomal recessive spastic paraplegia type 21	Mast syndrome|SPG21	Disease	Rare neurologic disease

101011	Autosomal dominant spastic paraplegia type 31	SPG31	Disease	Rare neurologic disease
101023	Cleft hard palate		Morphological anomaly	Rare developmental defect during embryogenesis
101039	Female restricted epilepsy with intellectual disability	EFMR|Juberg-Hellman syndrome	Disease	Rare developmental defect during embryogenesis
101063	Situs inversus totalis	Complete situs inversus|Complete situs inversus viscerum|Situs inversus	Morphological anomaly	Rare developmental defect during embryogenesis
101070	Bilateral frontoparietal polymicrogyria		Clinical subtype	Rare developmental defect during embryogenesis
101076	X-linked Charcot-Marie-Tooth disease type 2	CMTX2	Disease	Rare neurologic disease
101433	Rare urogenital disease		Category	0
102002	Rare ataxia		Category	0
102009	Classic lissencephaly	Lissencephaly type 1	Clinical group	0
102015	Autosomal recessive limb-girdle muscular dystrophy		Category	0
109007	Arthrogryposis syndrome		Category	0
137678	Spondyloepiphyseal dysplasia with metatarsal shortening	Czech dysplasia, metatarsal type|SED with metatarsal shortening	Disease	Rare bone disease
137888	Auriculocondylar syndrome	Question mark ear syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
137902	Isolated optic nerve hypoplasia/aplasia		Morphological anomaly	Rare ophthalmic disorder
139396	X-linked cerebral adrenoleukodystrophy	X-CALD	Clinical subtype	Rare inborn errors of metabolism
139399	Adrenomyeloneuropathy		Clinical subtype	Rare inborn errors of metabolism
139455	Autosomal recessive bestrophinopathy	Retinopathy, Burgess-Black type	Disease	Rare ophthalmic disorder

139536	Distal hereditary motor neuropathy type 5	Distal HMN V|Distal hereditary motor neuropathy type V|Distal spinal muscular atrophy type 5|dHMN5	Disease	Rare neurologic disease
140957	Autosomal dominant macrothrombocytopenia		Disease	Rare hematologic disease
155867	Paramedian facial cleft	Tessier number 1-1 and 2-12 facial cleft	Clinical group	0
156601	Rare genetic hepatic disease		Category	0
156728	Spondyloepimetaphyseal dysplasia, matrilin-3 type	SEMD, MATN3-related|SEMD, matrilin-3 type	Disease	Rare bone disease
157215	Hereditary hypophosphatemic rickets with hypercalciuria	HHRH	Disease	Rare endocrine disease
157794	Hereditary mixed polyposis syndrome	HMPS	Disease	Rare gastroenterologic disease
157798	Serrated polyposis syndrome	Hyperplastic polyposis syndrome	Disease	Rare gastroenterologic disease
157973	Congenital muscular dystrophy due to LMNA mutation	L-CMD|LMNA-related congenital muscular dystrophy	Disease	Rare neurologic disease
163634	Maffucci syndrome		Disease	Rare bone disease
163690	Hypotonia-cystinuria syndrome	HCS	Disease	Rare inborn errors of metabolism
163937	X-linked intellectual disability, Najm type	MICPCH|X-linked intellectual disability-microcephaly-pontocerebellar hypoplasia syndrome	Disease	Rare developmental defect during embryogenesis
163976	X-linked intellectual disability, Van Esch type		Malformation syndrome	Rare developmental defect during embryogenesis
163985	Hyperekplexia-epilepsy syndrome		Disease	Rare developmental defect during embryogenesis
165704	Non-syndromic urogenital tract malformation		Category	0

166002	Multiple epiphyseal dysplasia due to collagen 9 anomaly		Disease	Rare bone disease
166100	Autosomal dominant otospondylomegaepiphyseal dysplasia	AD OSMED|Stickler syndrome type 3|Stickler syndrome, non-ocular type	Malformation syndrome	Rare bone disease
166108	Intellectual disability, Birk-Barel type	Intellectual disability-hypotonia-facial dysmorphism syndrome	Disease	Rare developmental defect during embryogenesis
166119	Isolated osteopoikilosis		Disease	Rare bone disease
166260	Dentinogenesis imperfecta type 2	Capdepont teeth|DGI-2|DI-2|Dentinogenesis imperfecta, Shields type 2	Clinical subtype	Rare odontologic disease
166472	Monogenic disease with epilepsy		Category	0
168569	H syndrome		Malformation syndrome	Rare skin disease
169147	Immunodeficiency due to a classical component pathway complement deficiency	Immunodeficiency due to C1, C4, or C2 component complement deficiency|Immunodeficiency due to an early component of complement deficiency	Disease	Rare immune disease
169355	Immunodeficiency syndrome with autoimmunity		Category	0
169802	Severe hemophilia A	Severe factor VIII deficiency	Clinical subtype	Rare hematologic disease
171871	Renal pseudohypoaldosteronism type 1	Autosomal dominant pseudohypoaldosteronism type 1	Clinical subtype	Rare renal disease
171876	Generalized pseudohypoaldosteronism type 1	Autosomal recessive pseudohypoaldosteronism type 1	Clinical subtype	Rare renal disease
174590	Congenital hypogonadotropic hypogonadism		Category	0
177901	Prader-Willi syndrome due to paternal deletion of 15q11q13 type 1		Etiological subtype	Rare developmental defect during embryogenesis

178025	Non-acquired combined pituitary hormone deficiencies without extrapituitary malformations		Category	0
178303	8q22.1 microdeletion syndrome	Monosomy 8q22.1|Nablus mask-like facial syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
181393	Growth hormone insensitivity syndrome	GHIS|Short stature due to a defect in growth hormone receptor or post-receptor pathway	Category	0
181396	Rare hypothyroidism		Category	0
181399	Rare hyperthyroidism		Category	0
181412	Adrenogenital syndrome		Category	0
183484	Genetic subcutaneous tissue disorder		Category	0
183539	Genetic renal or urinary tract malformation		Category	0
189439	Primary pigmented nodular adrenocortical disease	PPNAD|Primary pigmented nodular adrenal dysplasia	Disease	Rare endocrine disease
199267	Infantile digital fibromatosis	Inclusion body fibromatosis|Recurring digital fibrous tumor of childhood|Reye tumor	Disease	Rare neoplastic disease
199627	Atypical autism		Disease	Rare neurologic disease
200418	Immunodeficiency with factor I anomaly	Complete factor I deficiency	Disease	Rare immune disease
206549	Anoctamin-5-related limb-girdle muscular dystrophy R12	Anoctamin-5-related LGMD R12|Autosomal recessive limb-girdle muscular dystrophy type 2L|LGMD type 2L|LGMD2L|Limb-girdle muscular dystrophy type 2L	Disease	Rare neurologic disease

206599	Isolated asymptomatic elevation of creatine phosphokinase	Idiopathic asymptomatic hyperCKemia|Isolated asymptomatic hyperCKemia	Biological anomaly	Rare neurologic disease
206647	Myotonic dystrophy		Clinical group	0
206973	Congenital myotonia		Clinical group	0
208513	Spinocerebellar ataxia type 29	Congenital nonprogressive spinocerebellar ataxia|SCA29	Disease	Rare neurologic disease
210548	Macrocephaly-intellectual disability-autism syndrome		Disease	Rare developmental defect during embryogenesis
216812	Osteogenesis imperfecta type 3	OI type 3|Progressive deforming osteogenesis imperfecta|Severe osteogenesis imperfecta	Clinical subtype	Rare bone disease
216866	Classic pantothenate kinase-associated neurodegeneration	NBIA1, classic form|Neurodegeneration with brain iron accumulation type 1, classic form|PKAN, classic form	Clinical subtype	Rare neurologic disease
216981	Niemann-Pick disease type C, juvenile neurologic onset	Niemann-Pick disease type C, classic form	Clinical subtype	Rare inborn errors of metabolism
217085	Mucopolysaccharidosis type 2, severe form	Hunter syndrome type A|Iduronate 2-sulfatase deficiency type A|MPS2A|MPSIIA|Mucopolysaccharidosis type 2A|Mucopolysaccharidosis type II, severe form|Mucopolysaccharidosis type IIA	Clinical subtype	Rare inborn errors of metabolism
217377	Microduplication Xp11.22p11.23 syndrome	Dup(X)(p11.22p11.23)|Trisomy Xp11.22p11.23	Malformation syndrome	Rare developmental defect during embryogenesis
217557	Pulmonary interstitial glycogenosis	Infantile cellular interstitial pneumonitis|PIG	Disease	Rare respiratory disease

217572	Glycogen storage disease with hypertrophic cardiomyopathy	GSD with hypertrophic cardiomyopathy|Glycogenosis with hypertrophic cardiomyopathy	Category	0
217598	Non-familial hypertrophic cardiomyopathy		Category	0
220489	Rare hereditary hemochromatosis	Iron overload disease	Category	0
221046	Poikiloderma with neutropenia	Poikiloderma with neutropenia, Clericuzio type	Disease	Rare skin disease
223713	Mitochondrial oxidative phosphorylation disorder	OXPHOS disease	Category	0
228285	Acquired cutis laxa	Cutis laxa acquisita	Disease	Rare skin disease
228363	CLN6 disease		Etiological subtype	Rare neurologic disease
228384	5q14.3 microdeletion syndrome	Del(5)(q14.3)|Monosomy 5q14.3	Malformation syndrome	Rare developmental defect during embryogenesis
228410	Polyvalvular heart disease syndrome	PHD syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
231140	Silver-Russell syndrome due to an imprinting defect of 11p15		Etiological subtype	Rare developmental defect during embryogenesis
231144	Silver-Russell syndrome due to 11p15 microduplication		Etiological subtype	Rare developmental defect during embryogenesis
231512	Hermansky-Pudlak syndrome without pulmonary fibrosis	HPS without pulmonary fibrosis	Clinical subtype	Rare immune disease
231679	Isolated growth hormone deficiency type II	Congenital IGHD type II|Congenital isolated GH deficiency type II|Congenital isolated growth hormone deficiency type II	Clinical subtype	Rare endocrine disease
238750	4q21 microdeletion syndrome	Del(4)(q21)|Monosomy 4q21	Malformation syndrome	Rare developmental defect during embryogenesis
240371	Syndromic obesity		Category	0

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	Disease	Rare inborn errors of metabolism
247868	NLRP12-associated hereditary periodic fever syndrome	FCAS2|Familial cold autoinflammatory syndrome type 2|NAPS12	Disease	Rare systemic or rheumatologic disease
248095	Primary hypertrophic osteoarthropathy	Idiopathic hypertrophic osteoarthropathy|PHO	Clinical group	0
250984	Autosomal recessive Stickler syndrome		Clinical subtype	Rare bone disease
251014	2q31.1 microdeletion syndrome	Del(2)(q31.1)|Monosomy 2q31.1	Malformation syndrome	Rare developmental defect during embryogenesis
251028	SATB2-associated syndrome due to a chromosomal rearrangement	2q33.1 microdeletion syndrome|Del(2)(q33.1)|Monosomy 2q33.1	Etiological subtype	Rare developmental defect during embryogenesis
252018	Teratoma of the central nervous system		Clinical subtype	Rare neoplastic disease
254351	Distal 7q11.23 microdeletion syndrome	Distal del(7)(q11.23)|Distal monosomy 7q11.23	Malformation syndrome	Rare developmental defect during embryogenesis
254516	Temple syndrome		Malformation syndrome	Rare developmental defect during embryogenesis
254837	Unspecified mitochondrial disorder		Clinical group	0

255182	Pyruvate dehydrogenase E3-binding protein 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 dehydrog	Clinical subtype	Rare inborn errors of metabolism
261190	15q14 microdeletion syndrome	Del(15)(q14)|Monosomy 15q14	Malformation syndrome	Rare developmental defect during embryogenesis
261257	Distal 17p13.3 microdeletion syndrome	Distal del(17)(p13.3)|Distal monosomy 17p13.3	Malformation syndrome	Rare developmental defect during embryogenesis
261279	17q23.1q23.2 microdeletion syndrome	Del(17)(q23.1q23.2)|Monosomy 17q23.1q23.2	Malformation syndrome	Rare developmental defect during embryogenesis
261295	20p12.3 microdeletion syndrome	Del(20)(p12.3)|Monosomy 20p12.3	Malformation syndrome	Rare developmental defect during embryogenesis
261311	20q13.33 microdeletion syndrome	Del(20)(q13.33)|Monosomy 20q13.33	Malformation syndrome	Rare developmental defect during embryogenesis
261537	Mowat-Wilson syndrome due to monosomy 2q22	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 microdel	Etiological subtype	Rare developmental defect during embryogenesis

261600	Alagille syndrome due to 20p12 microdeletion	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	Etiological subtype	Rare developmental defect during embryogenesis
261821	Partial deletion of the long arm of chromosome 12	Partial deletion of chromosome 12q|Partial monosomy of chromosome 12q|Partial monosomy of the long arm of chromosome 12	Category	0
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	Category	0
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	Category	0
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	Category	0

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	Category	0
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	Category	0
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	Category	0
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	Category	0
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	Category	0
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	Category	0

262201	Partial duplication of chromosome 3	Partial trisomy of chromosome 3	Category	0
262643	Partial trisomy/tetrasomy of chromosome 9	Partial duplication/triplication of chromosome 9	Category	0
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	Category	0
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	Category	0
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	Category	0
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	Category	0
262923	Partial duplication of the long arm of chromosome 11	Partial duplication of chromosome 11q|Partial trisomy of chromosome 11q|Partial trisomy of the long arm of chromosome 11	Category	0

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	Category	0
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	Category	0
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	Category	0
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	Category	0
263463	CHST3-related skeletal dysplasia	Chondrodysplasia with congenital joint dislocations, CHST3 type|SDCD, CHST3 type|Spondyloepiphyseal dysplasia with congenital joint dyslocations, CHST3 type	Disease	Rare bone disease
263516	Progressive myoclonic epilepsy type 3	CLN14 disease|EPM3|PME type 3|Progressive myoclonic epilepsy due to KCTD7 deficiency|Progressive myoclonus epilepsy type 3	Clinical subtype	Rare neurologic disease
263534	Acral peeling skin syndrome	Acral PSS|Acral deciduous skin|Localized PSS|Localized deciduous skin	Disease	Rare skin disease

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	Category	0
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	Category	0
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	Category	0
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 li	Disease	Rare inborn errors of metabolism
268261	DYRK1A-related intellectual disability syndrome due to 21q22.13q22.2 microdeletion	21q22.13q22.2 microdeletion syndrome|Del(21)(q22.13q22.2)|Monosomy 21q22.13q22.2	Etiological subtype	Rare developmental defect during embryogenesis
268820	Cranial meningocele		Morphological anomaly	Rare developmental defect during embryogenesis
268823	Occipital encephalocele		Clinical subtype	Rare developmental defect during embryogenesis
268835	Lipomyelomeningocele		Morphological anomaly	Rare developmental defect during embryogenesis

268861	Primary tethered cord syndrome	Primary tethered spinal cord syndrome	Morphological anomaly	Rare developmental defect during embryogenesis
271861	Hereditary ATTR amyloidosis	Familial TTR-related amyloidosis|Familial transthyretin-related amyloidosis	Clinical group	0
275761	Lysosomal acid lipase deficiency	LAL deficiency	Disease	Rare inborn errors of metabolism
275777	Heritable pulmonary arterial hypertension	FPAH|Familial pulmonary arterial hypertension|HPAH|Hereditary pulmonary arterial hypertension	Etiological subtype	Rare respiratory disease
275872	Frontotemporal dementia with motor neuron disease	FTD-ALS|FTD-MND|Frontotemporal dementia with amyotrophic lateral sclerosis	Disease	Rare neurologic disease
276432	Ogden syndrome	Premature aging appearance-developmental delay-cardiac arrhythmia syndrome	Malformation syndrome	Rare developmental defect during embryogenesis
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	Disease	Rare inborn errors of metabolism
276598	Diazoxide-resistant focal hyperinsulinism due to SUR1 deficiency	Hyperinsulinemic hypoglycemia due to SUR1 deficiency, diazoxide-resistant focal form	Disease	Rare endocrine disease
280229	Pelizaeus-Merzbacher disease in female carriers		Clinical subtype	Rare neurologic disease
280640	Occipital pachygyria and polymicrogyria	Occipital MCD|Occipital malformations of cortical development	Malformation syndrome	Rare developmental defect during embryogenesis
280763	Severe intellectual disability and progressive spastic paraplegia	AP4 deficiency syndrome	Disease	Rare developmental defect during embryogenesis

282166	Inherited Creutzfeldt-Jakob disease	Inherited CJD	Disease	Rare neurologic disease
284289	Adult-onset autosomal recessive cerebellar ataxia	Autosomal recessive spinocerebellar ataxia type 10|SCAR10	Disease	Rare neurologic disease
284343	Pleuropulmonary blastoma familial tumor susceptibility syndrome	DICER1 syndrome|PPB familial tumor susceptibility syndrome|PPBFTDS|Pleuro-pulmonary blastoma familial tumor susceptibility syndrome	Clinical subtype	Rare neoplastic disease
284993	Marfan syndrome and Marfan-related disorders		Category	0
289157	Hypocalcemic vitamin D-dependent rickets	1-alpha-hydroxylase deficiency|PDDRI|Pseudovitamin D-deficient rickets|VDDI|VDDR-I|Vitamin D dependent rickets type I|Vitamin D-dependency type I	Disease	Rare endocrine disease
289548	Inherited isolated adrenal insufficiency due to partial CYP11A1 deficiency		Disease	Rare endocrine disease
289601	Hereditary arterial and articular multiple calcification syndrome	CALJA|Calcification of joints and arteries	Disease	Rare circulatory system disease
289877	Transient hyperammonemia of the newborn		Particular clinical situation in a disease or syndrome	Rare inborn errors of metabolism
289899	Organic aciduria		Category	0
293633	PYCR1-related De Barsy syndrome	PYCR1 deficiency|Pyrroline-5-carboxylate reductase 1 deficiency	Etiological subtype	Rare inborn errors of metabolism
293948	1p21.3 microdeletion syndrome	Del(1)(p21.3)|Monosomy 1p21.3	Malformation syndrome	Rare developmental defect during embryogenesis

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	Disease	Rare endocrine disease
294057	Rare nevus		Category	0
294953	Non syndromic limb overgrowth		Category	0
294983	Acheiria	Congenital absence of hand	Morphological anomaly	Rare developmental defect during embryogenesis
294988	Congenital hypoplasia of thumb	Congenital absence/hypoplasia of thumb|Thumb hypodactyly|Thumb oligodactyly	Morphological anomaly	Rare developmental defect during embryogenesis
295047	Macrodactyly of toes	Macrodactyly of foot	Morphological anomaly	Rare developmental defect during embryogenesis
295051	Lower limb hypertrophy		Morphological anomaly	Rare developmental defect during embryogenesis
300496	Multiple congenital anomalies-hypotonia-seizures syndrome type 2	MCAHS type 2	Malformation syndrome	Rare developmental defect during embryogenesis
300547	Autosomal recessive infantile hypercalcemia	Familial infantile hypercalcemia with suppressed intact parathyroid hormone	Disease	Rare endocrine disease
528084	Non-specific syndromic intellectual disability	Complex neurodevelopmental disorder	Category	0
418	Congenital adrenal hyperplasia	CAH	Clinical group	0