21/05/2025

רשימת מחלות נדירות

לפי תקנות הביטוח הלאומי (ילד נכה), התש"ע-2010, סעיף 13 לתוספת הראשונה, בסמכות מנהל השירותים הרפואיים במוסד, או רופא שהוא הסמיכו לכך, לקבוע מהי מחלה או תסמונת נדירה.
מחלה או תסמונת נדירה, לפי תקנות אלה, היא כזו הקיימת ביחס של אחד, לכל היותר, בכל מאה אלף לידות חי.
בהתאם לכך, להלן רשימת מחלות ששכיחותן ביילודים עונה להגדרת מחלה נדירה. יש לזכור כי הגדרה זו לבדה, אין בה די כדי לזכות בגמלה. על רופא מוסמך לקבוע האם הילד הנדון זקוק לטיפול מיוחד, במידה המטילה עומס כבד ביותר על המשפחה.
רשימת המחלות המוגדרות כמחלות נדירות אינה קבועה, והיא מתעדכנת בלשכה הרפואית באופן שוטף. הרשימה המצורפת כאן נכונה למועד פרסומה. ייתכן שישנן מחלות נוספות שאינן מופיעות ברשימה, וכן מחלות שמופיעות ברשימה אך אינן מזכות בקצבה, כיוון שהתגלה כי שכיחותן גבוהה מהסף שהוגדר.
הרשימה המהווה בסיס לקבלת החלטה בדבר קיומה של נדירות המחלה הינה הרשימה של הלשכה הרפואית ביום עריכת האבחון הרפואי או ועדת הערר.

#	מחלה	שמות נרדפים
1	16p11.2p12.2 microdeletion syndrome	Monosomy 16p11.2p12.2
2	16p13.11 microdeletion syndrome	Monosomy 16p13.11
3	3p25.3 deletion syndrome	Del(3)p(25.3) 3p25.3 microdeletion syndrome Monosomy 3p25.3 Mental retardation-epilepsy-stereotypic hand movement syndrome
4	3q26 microduplication syndrome	Dup(3q) syndrome Trisomy 3q26 Dup(3)(q26)
5	5q14.3 microdeletion syndrome	MEF2C haploinsufficiency syndrome
6	8p inverted duplication deletion syndrome	
7	9q22.3 deletion syndrome	Monosomy 9q22.3 syndrome Microdeletion 9q22.3
8	Abetalipoproteinemia	
9	Acquired epileptic aphasia	Landau-Kleffner syndrome
10	Acrocephalosyndactyly	Apert syndrome
11	Acrocephalosyndactyly type 5	Pfeiffer syndrome
12	Acrodysostosis	
13	Adams-Oliver syndrome	
14	Adenosine deaminase 2 (ADA2) gene mutation	Deficiency of adenosine deaminase 2 (DADA2)
15	Aggressive fibromatosis	Desmoid fibromatosis CTNNB1 gene mutation
16	Aicardi Goutieres syndrome	
17	Aicardi syndrome (AIC)	
18	Al Kaissi syndrome (ALKAS)	Cyclin-dependent kinase 10 (CDK10) gene mutation
19	Alexander's disease	Alexander syndrome
20	Allan-Herndon-Dudley syndrome	Monocarboxylate transporter 8 deficiency
21	Alstrom syndrome	

22	Alternating hemiplegia of childhood	
23	Ameloblastome	
24	Arboleda Tham syndrome (ARTHS)	KAT6A gene mutation
25	Aromatic amino acid decarboxylase deficiency (AADC)	
26	ATR-X syndrome	Alpha-thalassemia/impaired intellectual development syndrome, X-linked Alpha-thalassemia/mental retardation syndrome, X-linked Alpha-thalassemia/mental retardation syndrome, nondeletion type
27	Au Kline syndrome	Okamoto syndrome
28	Autosomal dominant central core disease	Ryanodine receptor 1 (RYR1) gene receptor Periodic paralysis
29	Autosomal dominant Charcot-Marie-Tooth disease type 2U	Methionyl-tRNA synthetase (MARS) gene mutation
30	Autosomal dominant HYPER-IgE syndrome due to STAT3 deficiency (HIES)	HYPER-IgE syndrome 1, AD Job syndrome
31	Autosomal recessive cerebellar ataxia with oculomotor apraxia type 1 (AOA1)	
32	Autosomal recessive cerebellar ataxia, epilepsy, intellectual disability syndrome - WWOX deficiency	Autosomal recessive spinocerebellar ataxia type 12 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12; SCAR12
33	Autosomal recessive spastic paraplegia type 15	Hereditary spastic paraparesis Kjellin syndrome
34	Axenfeld-Rieger syndrome	
35	Aymé-Gripp syndrome (AYGRP)	
36	Bainbridge Ropers syndrome	ASXL transcriptional regulator 3 deficiency syndrome
37	Baker Gordon syndrome	Synaptotagmin (SYT) 1-related neurodevelopmental disorder
38	Baraitser Winter cerebrofrontofacial syndrome	
39	Barakat syndrome	Hypoparathyroidism, sensorineural hearing loss, renal disease syndrome Hypoparathyroidism, deafness, renal disease (HDR) syndrome
40	Bardet-Biedl syndrome (BBS)	Laurence-Moon-Biedl syndrome Laurence-Moon-Bardet-Biedl syndrome (LMBB)
41	Bartter syndrome type 2	Hyperprostaglandin E syndrome type 2 Hypokalemic alkalosis with hypercalciuria antenatal type 2
42	Bartter syndrome type 3	Bartter syndrome CLCNKB related disorder
43	BCL2L11-related disorder	2q13 deleteion syndrome
44	Beals syndrome	Congenital contractural arachnodactyly (CCA) Beals Hecht syndrome
45	Bernard Soulier syndrome	Hemorrhagiparous thrombocytic dystrophy Giant platelet syndrome
46	Blackfan-Diamond anemia	Aase syndrome Congenital pure red cell aplasia

47	Bohring-Opitz syndrome (BOS)	C-like syndrome
48	Borjeson-Forssman-Lehmann syndrome	
49	Brain lung thyroid syndrome	Choreoathetosis with congenital hypothyroidism and neonatal respiratoy distress syndrome
50	Brain malformation with or without urinary tract defects	Nuclear factor I/A (NFIA) gene mutation
51	BRD4 related syndrome	Cornelia de Lange syndrome 6 (CDLS6)
52	Bromodomain and WD repeat-containing protein 3 (BRWD3)	
53	Bruton's agammaglobulinemia	X-linked agammaglobulinemia (XLA)
54	CACNA1A -related epilepsy	Developmental and epileptic encephalopathy 42 (DEE42) Early infantile epileptic encephalopathy 42 (EIEE42)
55	Camptodactyly, arthropathy, coxa-vara, pericarditis syndrome (CACP)	Jacobs syndrome
56	Capillary malformation	Congenital anomaly of capillary
57	CAPOS syndrome	Cerebellar ataxia, areflexia, pes cavus, optic atrophy, sensorineural hearing loss syndrome
58	Cardiofaciocutaneous syndrome 1 (CFC1)	Noonan syndrome 7
59	Caudal duplication syndrome	Caudal duplication anomaly
60	CCR4-not transcription complex, subunit 3 (CNOT3) mutation	
61	Central core disease	Central core myopathy CONGENITAL MYOPATHY 1A, AUTOSOMAL DOMINANT, WITH SUSCEPTIBILITY TO MALIGNANT HYPERTHERMIA; CMYP1A
62	Cerebellar atrophy, developmental delay, and seizures (CADEDS)	
63	Cerebellar dysfunction with variable cognitive and behavioral abnormalities (CECBA)	CAMTA1 ASSOCIATED DISORDER Non-progressive cerebellar ataxia with intellectual disability
64	Cerebral folate transport deficiency	Cerebral folate deficiency
65	Cerebro-oculo-dento-auriculo-skeletal (CODAS) syndrome	
66	Cerebrotendinous xanthomatosis (CTX)	Cholestanol storage disease Van Bogaert-Scherer-Epstein syndrome Cerebral cholesterinosis Cerebrotendinous cholesterinosis
67	CHAMP1 related disease	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, IMPAIRED LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILD
68	Charcot-Marie-Tooth disease type 2N	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL DOMINANT, TYPE 2A1; CMT2A1 HEREDITARY MOTOR AND SENSORY NEUROPATHY IIA1; HMSN IIA1
69	Charcot-Marie-Tooth disease, axonal, type 2O	DYNC1H1-related Charcot-Marie-Tooth

70	CHD8 mutation related disorder	Intellectual developmental disorder with autism and macrocephaly (IDDAM) Chromodomain helicase DNA binding protein 8 overgrowth syndrome CHD8-related intellectual disability-autism-macrocephaly-tall stature syndrome
71	CHOPS syndrome	
72	Chronic bullous dermatosis of childhood	Linear IgA bullous disease in children Childhood linear IgA disease
73	Chronic granulomatous disease type II (CGD2)	
74	Chronic recurrent multifocal osteomyelitis (CRMO)	Majeed syndrome Synovitis acne pustulosis hyperostosis osteitis syndrome (SAPHO)
75	Ciliary dyskinesia, primary, 3 (CILD3)	DNAH5-related disorder
76	Citrullinemia type 2	Adult onset citrin deficiency
77	Clark Baraitser syndrome	
78	Clathrin (CLTC) gene mutation	Intelectual developmental disorder, autosomal dominant 56
79	Cleidocranial dysplasia (CCD)	Cleidocranial dysostosis (CLCD)
80	CLOVES syndrome	Congenital lipomatous overgrowth, vascular malformation, epidermal nevi, skeletal anomaly syndrome
81	CNOT3 related disorder	
82	Coats' disease	Leber's miliary aneurysms Miliary aneurysms of retina
83	Cobalamin C disease	Cobalamin locus C variant CblC methylmalonic acidemia and homocystinuria Methylmalonic acidemia with homocystinuria, type CBLC
84	Coffin-Lowry syndrome (CLS)	
85	Coffin-Siris syndrome	Fifth digit syndrome
86	Collagen type IV alpha 1 chain related familial vascular leukoencephalopathy	COL4A1-related brain small vessel disease with hemmorrhage COL4A1-related retinal arteriolar tortuosity, infantile hemiparesis, autosomal dominant leukoencephalopathy syndrome
87	Combined immunodeficiency due to DOCK8 deficiency	
88	Combined oxidative phosphorilation deficiency 37 (COXPD37)	MICOS13-related disorder
89	Congenital central hypoventilation syndrome (CCHS)	Ondine curse Congenital pulmonary hypoventilation
90	Congenital chronic diarrhea with protein-losing enteropathy	Congenital chronic diarrhea with exudative enteropathy
91	Congenital ichthyosiform erythroderma (CIE)	Congenital recessive ichthyosis
92	Congenital ichtyosis with hypotrichosis syndrome	Autosomal recessive congenital ichthyosis
93	Congenital insensitivity to pain with anhidrosis (CIPA)	Hereditary sensory and autonomic neuropathy (HSAN) type 4 Swanson-Buchanan-Alvord neuropathy syndrome

94	Congenital malformation of dural sinus	
95	Congenital myasthenic syndrome	Congenital myasthenia syndrome
96	Congenital myasthenic syndrome type 9 (CMS9)	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency MUSK-related disorder
97	Congenital myopathy 13 (CMYO13)	STAC3-related congenital myopathy Native American myopathy Myopathy, congenital, Bailey-Bloch (MYPBB) Myopathy, congenital, with myopathic facies, scoliosis and malignant hyperthermia
98	Congenital pseudoarthrosis of the tibia	
99	Congenital secretory diarrhea, chloride type	Congenital chloride diarrhea
100	Congenital stationary night blindness (CSNB)	Oguchi's disease
101	Convulsions, familial infantile, with paroxysmal choreoathetosis	PRRT2-related familial convulsions
102	COPB2 gene mutation-related disorder	Autosomal recessive primary microcephaly-19 (MCPH19)
103	Cortical dysplasia, complex, with other brain malformation 7 (CDCBM7)	TUBB2B-related disorder
104	Cortical dysplasia, complex, with other brain malformations 13	DYNC1H1-related cortical dysplasia
105	Craniofrontonasal dysplasia	Craniofrontonasal syndrome Craniofrontonasal dysostosis
106	Craniometaphyseal dysplasia	
107	Currarino triad	Currarino syndrome
108	Cutis marmorata telangiectatica congenita (CMTC)	Congenital livedo reticularis Van Lohuizen's syndrome
109	Cystathionine beta-synthase (CBS) deficiency	Deficiency of beta-thionase Deficiency of methylcysteine synthase Deficiency of serine sulphydrase
110	Cystic lymphangioma	Cystic hygroma
111	Deficiency of dihydrofolate reductase	Dihydrofolate reductase deficiency
112	Deficiency of isomaltase	
113	Deficiency of prolidase	
114	Dehydrodolichyl diphosphate sythase (DHDDS) gene mutation	
115	Deletion of part of autosome	
116	Dent disease type 1	Nephrolithiasis, hypercalciuric, X-linked (NPHL2)
117	Desmin myopathy	Desmin-related myofibrillar myopathy Desminopathy
118	Developmental and epileptic encephalopathy 64 (DEE64)	Epileptic encephalopathy, early infantile, 64 (EIEE64) RHOBTB2-related disorder
119	Developmental delay with or without dysmorphic facies and autism (DEDDFA)	Transformation/Transcription Domain-associated protein (TRRAP) associated disorder

120	Developmental delay-facial dysmorphism syndrome due to MED13L deficiency	Developmental delay, facial dysmorphism syndrome due to mediator complex subunit 13 like deficiency MED13L-related intellectual disability syndrome
121	Diaphyseal dysplasia	Camurati Engelmann syndrome
122	Diets Jongmans syndrome	KDM3B-related intellectual disability, facial dysmorphism, short stature syndrome
123	Diffuse intrinsic pontine glioma (DIPG)	
124	Distal anoctaminopathy	Miyoshi muscular dystrophy type 3 (MMD3) ANO5-related disorder
125	Distal trisomy 10q	Distal duplication 10q 10q trisomy
126	Distal Xq28 microduplication syndrome	Distal trisomy Xq28
127	DYRK1A-related intellectual disability syndrome	DYRK1A (dual specificity tyrosine phosphorylation regulated kinase 1A)
128	Dyskeratosis congenita (DKC)	Cole-Engmann-Zinsser syndrome
129	Dystonia 11, myoclonic (DYT11)	
130	Early infantile epileptic encephalopathy with suppression bursts	Ohtahara syndrome
131	Ectopia lentis syndrome	Familial ectopia lentis, Isolated ectopia lentis (IEL) ADAMTSL4 related lens ectopia
132	Emanuel syndrome	Supernumerary der(22)t(11;22) syndrome Der(22) syndrome due to 3:1 meiotic disjunction events Supernumerary derivative 22 chromosome syndrome
133	Encephalopathy due to defective mitochondrial and peroxisomal fission 2 (EMPF2)	DNM1L gene mutation 2
134	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1 (EMPF1)	DNM1L gene mutation 1
135	Enchondromatosis	Ollier disease
136	Epidermolysis bullosa	Acantholysis bullosa Fox disease
137	Epidermolysis bullosa acquisita	Acquired epidermolysis bullosa
138	Epidermolysis bullosa simplex 2A, generalized severe	Epidermolysis bullosa simplex 2A, Dowling-Meara type KRT5 related epidermolysis bullosa simplex, severe
139	Epilepsy, idiopathic generalized (EIG16)	
140	Epileptic encephalopathy due to KCNT1	Epilepsy of infancy with migrating focal seizures (EIMFS) Malignant migrating partial epilepsy of infancy (MMPEI) Malignant migrating partial seizures of infancy (MMPSI) Developmental and epileptic encephalopathy 14 (DEE14)

141	Episodic kinesigenic dyskinesia 1	PRRT2-related kinesigenic dyskinesia
142	Erythema annulare	Figurate erythema
143	Erythropoietic protoporphyria (EPP)	Magnus syndrome Heme synthase deficiency
144	Facioscapulohumeral muscular dystrophy (FMD, FSHD)	Landouzy-Dejerine muscular dystrophy
145	Factor X deficiency	Factor 10 deficiency Stuart-Prower factor deficiency
146	Familial amyloid nephropathy with urticaria and deafness	Muckle-Wells syndrome
147	Familial hemiplagic migraine type 1	Migraine type caused by mutations on the CACNA1A gene
148	Familial Hemophagocytic Lymphohistiocytosis (FHL)	Familial haemophagocytic reticulosis Familial HLH
149	Familial hypercholanemia	
150	Familial infantile bilateral striatal necrosis (IBSN)	
151	Familial lipoprotein lipase deficiency	Fredrickson type 1 hyperlipoproteinemia
152	Familial restrictive cardiomyopathy	
153	Familial Short QT syndrome	
154	FBXO11 related disorder	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (IDDFBA)
155	Febrile infection related epilepsy syndrome (FIRES)	Fever-induced refractory epileptic encephalopathy in school-aged children (FIRES)
156	Feingold syndrome	Microcephaly-oculo-digito-esophageal-duodenal syndrome MYCN-related disorder
157	Fibrodysplasia Ossificans Progressiva (FOP)	Progressive myositis ossificans Munchmeyer disease
158	Focal dermal hypoplasia (FDH)	Goltz-Gorlin syndrome
159	Forkhead box P1 (FOXP1) gene mutation	Intellectual disability, severe speech delay, mild dysmorphism syndrome
160	Forsius-Eriksson syndrome	Aland Island Eye Disease (AIED)
161	Fructose-1,6-bisphosphate aldolase B (ALDOB) deficiency	Hereditary fructosuria Aldolase B (ALDB) deficiency
162	GABA-transaminase deficiency	Gamma-aminobutiric acid transaminase deficiency
163	Galloway-Mowat syndrome	Nephrosis, neuronal dysmigration syndrome Galloway syndrome Microcephaly, hiatus hernia, nephrotic syndrome
164	Gastroparesis	Gastric atony Gastric stasis
165	GATAD2B-associated neurodevelopmental disorder (GAND)	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome GAND syndrome Mental retardation, autosomal dominant 18 (MRD18)
166	Genitopatellar syndrome (GTPTS)	Absent patellae, scrotal hypoplasia, renal anomalies, facial dysmorphism, and mental retardation KAT6B-related disorder

167	Ghosal hematodiaphyseal dysplasia	Ghosal syndrome TBXAS1-related disorder
168	Giant congenital melanocytic nevus (GCMN)	שומה מלנוציטית מולדת ענקית
169	Gillespie syndrome	Aniridia, cerebellar ataxia, intellectual disability syndrome
170	Glanzmann's thrombasthenia	Glanzmann's disease Hereditary hemorrhagic thromboasthenia
171	Glucocorticoid deficiency with achalasia	Allgrove syndrome Triple A syndrome
172	Glutamate receptor, ionotropuc, N-methyl-D-aspartate, subunit 2B (GRIN2B) gene mutation	Intellectual developmental disorder, autosomal dominant 6
173	Glutaric aciduria, type 1 (GA1)	Glutaric acidemia type 1 (GA1) Glutaryl-coenzyme A dehydrogenase deficiency (GCDHD)
174	Glycogen storage disease III (GSD3)	Glycogen storage disease 3 Amylo-1,6-glucosidase deficiency Cori disease Debrancher deficiency
175	Glycogen storage disease IXa (GSD9A)	
176	Gordon hyperkalemia-hypertension syndrome	Pseudohypoaldosteronism type 2
177	Gorham syndrome	Gorham's disease Hemangiomata with osteolysis Phantom bone disease
178	Greig cephalopolysyndactyly syndrome	
179	Growth factor, ERV1-like (GFER) gene mutation	Congenital cataract, progressive muscular hypotonia, hearing loss, developmental delay syndrome
180	Hamartoma of hypothalamus	Tuber cinereum hamartoma
181	Hao-Fountain syndrome	Ubiquitin-specific protease 7 (USP7) gene mutation Chromosome 16p13.2 deletion syndrome Intellectual developmental disorder with impaired speech, behavioral abnormalities and dysmorphic facies
182	Helsmoortel-van der Aa syndrome	Activity dependent neuroprotector homeobox related multiple congenital anomalies, intellectual disability, autism spectrum disorder ADNP-related multiple congenital anomalies, intellectual disability, autism spectrum disorder
183	Hemihyperplasia with multiple lipomatosis (HHML) syndrome	
184	Hereditary hyperekplexia	Congenital stiff man syndrome Familial startle disease Kok disease
185	Hereditary Hypophosphatemic Rickets with Hypercalciuria (HHRH)	
186	Hereditary limphedema type 1	Milroy lymphedema Nonne-Milroy lymphedema

187	Hereditary pancreatitis	Protease, serine 1 (PRSS1) gene mutation
188	Hereditary pheochromocytoma and paraganglioma	
189	Hereditary sensory and autonomic neuropathy (HSAN)	
190	Hereditary sensory and autonomic neuropathy (HSAN) type 9	Tectonin beta-propeller repeat-containing protein 2 (TECPR2) gene mutation Autosomal recessive spastic paraplegia type 49
191	Hereditary spastic paraplegia (HSP)	Strumpell-Lorrain disease
192	Hermansky-Pudlak syndrome (HPS)	Albinism with hemorrhagic diathesis Alpha storage pool disease
193	Heterogeneous nuclear ribonucleoprotein D (HNRNPD) gene mutation	
194	HHH syndrome (Hyperornithinemia, Hyperammonemia, Homocitrullinuria)	Triple H syndrome Ornithine translocase ORNT1 deficiency Ornithine carrier deficiency
195	Histiocytosis-lymphadenopathy plus syndrome	H syndrome SLC29A3 spectrum disorder
196	Houge-Janssens syndrome 1 (HJS1)	
197	Houge-Janssens syndrome 2 (HJS2)	
198	Houge-Janssens syndrome 3 (HJS3)	
199	Human immunodeficiency virus (HIV) disease	איידס
200	Hyperekplexia epilepsy syndrome	ARHGEF9 RELATED DISORDER
201	Hyperinsulinemic hypoglycemia, familial, 1 (HHF1)	ABCC8-related disorder
202	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency (SAHHD)	
203	Hyperphosphatemic familial tumoral calcinosis	
204	Hypertrophy (benign) of prostate with urinary obstruction and other lower urinary tract symptoms (LUTS)	
205	Hyperuricemia, pulmonary hypertension, renal failure, alkalosis syndrome	HUPRA syndrome
206	Hypotonia Hypoventilation Impaired Intellectual Development Dysautonomia Epilepsy and Eye	HIDEA syndrome
207	Hypotonia, Ataxia and Delayed Development syndrome (HADDS)	
208	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome (HADDTS)	CTBP1-related disorder
209	I-cell disease	Mucolipidosis II GNPTAB-related disorder
210	Ichthyosis, Congenital; Autosomal Recessive 1 (ARCI1)	COLLODION BABY, SELF-HEALING; SHCB ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 1, WITH BATHING SUIT DISTRIBUTION

211	IDDHBA syndrome	Intellectual developmental disorder with hypotonia and behavioral abnormalities
212	Idiopathic non-lupus full-house nephropathy (FHN)	
213	Immunodeficiency 14	Activated PI3K-delta syndrome
214	Immunodysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome	Autoimmune enteropathy type 1
215	Impaired intellectual development and distinctive facial features with or without cardiac defects (MRFACD)	Asadollahi-Rauch syndrome
216	Infantile Neuroaxonal Dystrophy (INAD1)	Seitelberger's disease
217	Intellectual developmenta disorder with severe speech and ambulation defects (IDDSSAD)	ACTL6B gene mutation
218	Intellectual developmental disorder, AR 13	TRAPPC9-related disorder
219	Intellectual developmental disorder, autosomal dominant 29 (MRD29)	SETBP1 haploinsufficiency disorder (SETBP1-HD)
220	Intellectual developmental disorder, autosomal dominant 45 (MRD45)	CIC gene mutation-related disorder
221	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type	KDM5C related syndromic X-linked intellectual disability Claes Jensen X-linked De Novo syndrome Mental Retardation, X-Linked, syndromic, Claes-Jensen type (MRXSCJ)
222	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type	DDX3X related disorder
223	Intellectual disability Birk-Barel type	Intellectual disability, hypotonia, facial dysmorphism syndrome Birk Barel mental retardation dysmorphism syndrome KCNK9 (potassium two pore domain channel subfamily K member 9) imprinting syndrome
224	Intellectual disability, expressive aphasia, facial dysmorphism syndrome	
225	Intellectual disability, facial dysmorphism syndrome due to SETD5 haploinsufficiency	
226	Intellectual disability-hypotonic facies syndrome, X-linked (MRXHF1)	Smith-Fineman-Myers syndrome 1 (SFMS) XLMR-hypotonic facies syndrome Carpenter-Waziri syndrome Holmes-Gang syndrome Chudley-Lowry syndrome
227	Intractable diarrhea of infancy syndrome (IDIS)	Diarrhea 11, malabsorptive, congenital (DIAR11) Proline- and glutamate-rich protein with coiled-coil domain 1 (PERCC1) gene mutation

228	Intrauterine restriction, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomaly (IMAGe) syndrome	
229	Isovaleric acidemia	Isovaleryl-coenzyme A dehydrogenase deficiency Isovaleric aciduria
230	ITPR1 mutation related spinocerebellar ataxia 29	Spinocerebellar ataxia type 29 (SCA29) Congenital nonprogressive spinocerebellar ataxia
231	Jansen-de Vries syndrome (JDVS)	
232	Joubert syndrome	
233	Joubert syndrome with renal defect	NPHP1-related nephronophthisis
234	Juvenile Hypophosphatasia	Hypophosphatasia, childhood type
235	KBG syndrome	Short stature, facial and skeletal anomalies, intellectual disability, macrodontia syndrome
236	KIF1A associated neurological disorder (KAND)	
237	Kindler's syndrome	
238	KINSSHIP syndrome	
239	Kleefstra syndrome	due to del(9)(q34) due to monosomy 9q34
240	Kleefstra syndrome 2	
241	Klinefelter syndrome variant	49 XXXXY
242	Klippel Trenaunay syndrome	Klippel Trenaunay Weber syndrome
243	Kohlschutter syndrome	Amelocerebrohypohidrotic syndrome Epilepsy, mental deterioration and yellow teeth Epilepsy, dementia, amelogenesis, imperfecta syndrome Kohlschutter-Tonz syndrome
244	Kostmann syndrome	Infantile agranulocytosis
245	Kozlowski spondylometaphyseal dysplasia	TRPV4 related skeletal dysplasia
246	KREMEN1 related ectodermal dysplasia 13	Ectodermal dysplasia 13, hair/tooth type (ECTD13)
247	Lamb Shaffer syndrome	SOX5 haploinsufficiency syndrome
248	Lamellar Ichthyosis	Collodion baby
249	Lateral meningocele syndrome (LMNS)	NOTCH3-related lateral meningocele syndrome Lehman syndrome
250	Legius syndrome	Neurofibromatosis type 1-like syndrome (NFLS)
251	Leukocyte adhesion deficiency (LAD) type 2	
252	Liang-Wang syndrome (LIWAS)	
253	Limb-girdle Muscular Dystrophy, Type 28, Autosomal Recessive	LGMDR28
254	Lipid transport defect of intestine	Chylomicron retention disease Anderson syndrome
255	Lissencephaly, X-linked, 1 (LISX1)	Lissencephaly and agenesis of corpus callosum (XLIS) Subcortical laminar heterotopia, X-linked (SCLH) Subcortical band heterotopia, X-linked (SBH) Double cortex (DC) syndrome DCX-related disorder
256	Loeys-Dietz syndrome	

257	Long chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency (LCHADD)	
258	Lowe syndrome	Cerebro-oculorenal dystrophy Oculocerebrorenal syndrome Lowe-Bickel syndrome Lowe-Terrey-MacLachlan syndrome Phosphatidylinositol-4,5-bisphosphate-5-phosphatase deficiency
259	Lysine demethylase 5c (KDM5C) gene mutation	
260	Lysinuric protein intolerance (LPI)	Congenital lysinuria
261	Malan overgrowth syndrome	Sotos syndrome 2
262	Mandibulofacial dysostosis with microcephaly	Mandibulofacial dysostosis, Guion-Almeida type Elongation factor Tu GTP-binding comain-containing 2 (EFTUD2) gene mutation
263	Maple syrup urine disease (MSUD)	Branched chain 2-ketoacid dehydrogenase deficiency (BCKD) Ketoacidemia
264	Mast cell activation syndrome (MCAS)	Mast cell activation disorder (MCAD)
265	Maternally inherited Leigh syndrome (MILS)	Maternally inherited infantile subacute necrotizing encephalopathy
266	MBD5-associated neurodevelopmental disorder (MAND)	Intellectual developmental disorder, autosomal dominant 1
267	McCune Albright syndrome	
268	Megalencephaly capillary malformation (MCAP)	Macrocephaly-capillary malformation (M-CM, MCM) Megalencephaly-cutis marmorata telangiectatica congenita syndrome (MCMTC)
269	Meier-Gorlin syndrome	Ear, patella, short stature syndrome Microtia, absent patellae, micrognathia syndrome
270	MEIS2 syndrome	Cleft palate, cardiac defects, and impaired intellectual development Cardiac malformation, cleft lip/palate, microcephaly and digital anomalies
271	Metaphyseal anadysplasia type 2	Matrix metalloproteinase 9 (MMP9) gene mutation
272	Methyl-CpG-binding protein 2 (MECP2) gene mutation	
273	Methylmalonic acidemia	Methylmalonic aciduria (MMA)
274	Microcephalic primordial dwarfism Alazami type	Alazmai syndrome
275	Microcephaly, corpus callosum hypoplasia, intellectual disability, facial dysmorphism syndrome	
276	Minor partial trisomy	Minor partial chromosome duplication
277	Mitochondrial enoyl coA reductase protein-associated neurodegeneration (MEPAN) syndrome	Mitochondrial enoyl CoA hydratase 1 deficiency (ECHS1D) Mitochondrial enoyl coA reductase (MECP) gene mutation
278	Mixed gonadal dysgenesis	Mosaicism 45, X; 46, XY
279	Moebius syndrome	Congenital facial diplegia

280	Molybdenum cofactor deficiency (MoCD)	Combined molybdenoflavoprotein enzyme deficiency Combined xanthine oxidase and aldehyde oxidase deficiency Hereditary xanthinuria type 2
281	Mosaic trisomy 16 syndrome	
282	Moyamoya disease	
283	MPPH syndrome	Megalencephaly, polymicrogyria, postaxial polydactyly, hydrocephalus syndrome
284	MULLEGAMA-KLEIN-MARTINEZ SYNDROME STAG2 RELATED DISORDER (MKMS)	
285	Multicentric carpotarsal osteolysis syndrome	
286	Multiple congenital exostosis	Multiple osteochondromatosis syndrome
287	Multiple endocrine neoplasia, type 3 (MEN3)	Multiple endocrine neoplasia, type 2B (MEN2B)
288	Multiple sulfatase deficiency (MSD)	Juvenile sulfatidosis, Austin type Mucosulphatidosis
289	Multisystemic smooth muscle dysfunction syndrome	Actin, alpha-2, smooth muscle, aorta (ACTA2) gene mutation
290	Muscle eye brain disease	
291	Muscle phosphoglycerate mutase deficiency	Phosphoglucomutase deficiency
292	Myhre syndrome	
293	Myoclonus Epilepsy and Ataxia due to potassium channel mutation (MEAK)	Progressive myoclonic epilepsy type 7 Progressive myoclonic epilepsy due to KV3.1 deficiency Epilepsy progressive myoclonic 7 (EPM7)
294	Myofibrillar myopathy	
295	Myosin, cardiac, heavy chain, beta (MYHCB, MYH7) gene mutation	
296	Nabais Sa-de Vries syndrome, type 2 (NSDVS2)	NEURODEVELOPMENTAL DISORDER WITH RELATIVE MACROCEPHALY AND WITH OR WITHOUT CARDIAC OR ENDOCRINE ANOMALIES, NEDMACE
297	NARS2 related disorder	Deafness. Autosomal recessive 94 (DFNB94)
298	Nemo syndrome	X-linked MSMD due to IKBKG deficiency X-linked MSMD due to NEMO deficiency X-linked mendelian susceptibility to mycobacterial diseases due to NEMO deficiency
299	Neurodegeneration due to 3-hydroxyisobutyryl coenzyme A hydroxylase (HIBCH) deficiency	Methacrylic aciduria Valine metabolic defect
300	Neurodegeneration, childhood-onset, with brain atrophy (CONDBA)	UBTF related motor and cognitive regression syndrome
301	Neurodevelopmental disorder w/ hypotonia, variable intellectual + behavioral abnormalities (NEDHIB)	POLR2A-related disorder
302	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies (NEDDFL)	BPTF-related disorder

303	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies (NEDDFSA)	Zinc finger miz-domain containig 1(ZMIZ1) gene mutation
304	Neurodevelopmental disorder with eye movement abnormalities and ataxia (NEDEMA)	FRMD5-related disorder
305	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures (NEDFSS)	TRPM3-related disorder
306	Neuroendocrine cell hyperplasia of infancy	NEHI syndrome
307	Nicolaides-Baraitser syndrome	SWI/SNF-related, matrix-associated, actin-dependant regulator of chromatin, subfamily A, member 2 (SMARCA2) gene mutation
308	Niemann-Pick disease, type C	Supraoptic vertical ophthalmoplegia
309	NOG related disorder	Teunissen-Cremers syndrome Stapes ankylosis with broad thumbs and toes Ankylosis of stapes, hyperopia, broad thumbs, broad first toes and syndactyly
310	NPHS2 related nephrotic syndrome	Familial idiopathic steroid-resistant nephrotic syndrome Genetic SRNS Hereditary steroid-resistant nephrotic syndrome
311	Occipital pachygyria and polymicrogyria	LAMB1-related disorder
312	Oculo-facio-cardio-dental syndrome	Oculofaciocardiodental syndrome
313	O'Donnell-Luria-Rodan syndrome (ODLURO)	Lysine-specific methyltransferase 2E (KMT2E) gene mutation
314	Odonto-onycho-dermal dysplasia	Ectodermal dysplasia 16, hair/tooth/nail type (ECTD16) WNT10A-related disorder
315	Ogden syndrome	
316	Ohdo sydrome, Say-Barber-Biesecker-Young-Simpson (SBBYS) variant	Blepharophimosis, intellectual disability syndrome Say-Barber-Biesecker-Young-Simpson (SBBYS) variant KAT6B-related disorder
317	Okur-Chung neurodevelopmental syndrome (OCNDS)	OKUR CHUNG SYNDROME -CSNK2A1 MUTATION
318	Pallister Killian syndrome	Tetrasomy 12p syndrome Killian-Teschler-Nicola syndrome Pallister mosaic syndrome
319	Paraneoplastic opsoclonus-myoclonus-ataxia syndrome; POMA	Dancing eye syndrome Dancing eye-dancing feet syndrome Opsoclonus-myoclonus-ataxia syndrome; OMA Kinsbourne syndrome
320	Parkes Weber syndrome	
321	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy (PNKD3)	Generalized epilepsy and paroxysmal dyskinesia (GEPD)
322	Pearson's syndrome	
323	Pediatric multiple sclerosis	

324	Pelizaeus-Merzbacher disease (PMD)	Sudanophilic leukodystrophy
325	Penta X syndrome	XXXXX syndrome
326	Perching syndrome	Kelch like family member 7-related Bohring-Opitz-like and Crisponi/cold-induced sweating-like overlap syndrome
327	Perlman syndrome	
328	Permanent neonatal diabetes mellitus	Developmental delay, epilepsy and neonatal diabetes 1 (DEND)
329	Peutz Jeghers syndrome PJS	Peutz-Jeghers polyposis Peutz-Jeghers lentiginosis syndrome
330	PHACE syndrome	Phace syndrome and Moya Moya Disease Pascual-Castroviejo syndrome type 2
331	PHACES syndrome	
332	PHD finger protein 21A (PHF21A) gene mutation	
333	Phelan-McDermid syndrome	22q13.3 deletion syndrome SH3 and multiple ankyrin repeat domains 3 (SHANK3) gene mutation
334	PHIP-related syndrome	Pleckstrin homology domain interacting protein-related behavioral problems, intellectual disability, obesity, dysmorphic features syndrome PHIP-related behavioral problems, intellectual disability, obesity, dysmorphic features syndrome Chung Jansen syndrome; CHUJANs DEVELOPMENTAL DELAY, INTELLECTUAL DISABILITY, OBESITY, AND DYSMORPHISM; DIDOD
335	Phocomelia - Partial congenital absence of limb	Femorotibiofibular intercalary transverse meromelia Humero-radio-ulnar intercalary transverse meromelia Congenital absence of thigh and lower leg with foot present Congenital absence of upper arm and forearm with hand present
336	Phosphatase and tensin homolog (PTEN) gene mutation	
337	Phosphoenolpyruvate carboxykinase (PEPCK-C) deficiency	
338	Pitt-Hopkins syndrome (PTHS)	
339	Pitt-Hopkins-like syndrome 1 (PTHSL1)	
340	Pituitary stalk interruption syndrome	Ectopis neurohypophysis
341	Pleuropulmonary blastoma (PPB)	
342	Pleuropulmonary blastoma familial tumor and dysplasia syndrome (PPBFTDS)	DICER1 tumor-predisposition syndrome PPB familial tumor and dysplasia syndrome
343	Poirier-Bienvenu neurodevelopmental syndrome (POBINDS)	CSNK2B-related disorder

344	POMT1-related limb-girdle muscular dystrophy R11	Limb-girdle muscular dystrophy type 2K; LGMD type 2K POMT1-related LGMD R11
345	Pontocerebellar hypoplasia type 9 (PCH9)	
346	Popliteal pterygium syndrome	
347	Port-wine stain with oculocutaneous melanosis	Phakomatosis pigmentovascularis
348	Potocki Lupski syndrome	17p11.2 microduplication
349	Potocki-Shaffer syndrome	Chromosome 11p11.2 deletion syndrome Proximal 11p deletion syndrome (P11pDS)
350	Primary hyperoxaluria type 1	Glycolic aciduria
351	Primary microcephaly, mild intellectual disability, young-onset diabetes syndrome	
352	Primrose syndrome	Intellectual disability, cataract, calcified pinna, myopathy syndrome ZBTB20-related syndrome
353	PRKAG2-related hypertrophic cardiomyopathy	Familial hypertrophic cardiomyopathy 6
354	Progeria syndrome	Hutchinson-Gilford syndrome
355	Propionic Acidemia	Ketotic hyperglycinaemia Hyperglycinaemia with ketosis and leucopenia Propionyl-CoA carboxylase (PCC) deficiency
356	Proteolipid protein 1(PLP1) gene mutation	
357	Pseudohypoaldosteronism type 1, recessive form	Pseudohypoaldosteronism type 1B Pseudohypoaldosteronism, Persian-Jewish type
358	Pseudohypoparathyroidism	Guanine nucleotide-binding protein, alpha-stimulating activity polypeptide 1 (GNAS) gene mutation Parathyroid hormone resistant hypoparathyroidism
359	Pseudohypoparathyroidism type 1A	Albright hereditary osteodystrophy (AHO)
360	Pseudohypoparathyroidism type 2	
361	Pseudotumor cerebri	Benign intracranial hypertension; BIH Nonne's syndrome Noninfective serous meningitis
362	PTEN hamartoma tumor syndrome (PHTS)	Bannayan syndrome Bannayan-Riley-Ruvalcaba syndrome Macrocephaly with multiple lipomas and hemangiomas Cowden syndrome Proteus like syndrome Cohen-Hayden syndrome Segmental outgrowth, lipomatosis, arteriovenous malformation, epidermal nevus syndrome (SOLAMEN)
363	Pulmonary alveolar proteinosis (PAP)	
364	Purine rich element binding protein A (PURA) syndrome	
365	Pyknodysostosis	Maroteaux-Lamy syndrome type II Cathepsin k (CTSK) gene mutation

366	Radial aplasia-trhombocytopenia (TAR) syndrome	
367	Radioulnar synostosis with amegakayocytic thrombocytopenia (RUSAT) 2	MDS1 and EVI1 complex (MECOM) gene mutation
368	Rahman syndrome	H1-4-related neurodevelopmental disorder HIST1H1E-related disorder
369	RAS-associated autoimmune leukoproliferative disease (RALD)	Autoimmune leukoproliferative syndrome type 4
370	Rasmussen subacute encephalitis	Rasmussen syndrome Rasmussen encephalitis
371	Rauch-Steindl syndrome	
372	Raynaud-Claes syndrome	Chloride voltage-gated channel 4-related X-linked intellectual disability syndrome CLCN4-related X-linked intellectual disability syndrome Raynaud-Claes syndrome related to CLCN4 gene mutation
373	Renal nutcracker syndrome	Left renal vein entrapment syndrome Mesoaortic compression of left renal vein
374	Renpenning syndrome	Golabi-Ito-Hall syndrome Hamel cerebropalatocardiac syndrome Polyglutamine-binding protein 1 (PQBP1) gene mutation
375	Resistance to insulin-like growth factor 1 (IGF1)	Insulin-like growth factor 1 receptor (IGF1R) gene mutation
376	Rett syndrome; RTS	Rett's disorder Cerebroatrophic hyperammonemia
377	ROHHAD syndrome	Rapid-onset childhood obesity, hypothalamic dysfunction, hypoventilation, autonomic dysregulation syndrome
378	Rubinstein-Taybi syndrome	
379	Russell-Silver syndrome	Silver syndrome Silver-Russell dwarfism
380	Schaaf Yang syndrome	MAGE family member L2-related Prader-Willi-like syndrome MAGEL2 related Prader Willi-like syndrome
381	Schinzel-Giedion syndrome	
382	Schuurs-Hoeijmakers syndrome	
383	Scimitar syndrome	Pulmonary venolobar syndrome
384	Seizures, benign familial infantile, 2	PRRT2-related benign familial seizures
385	Selective malabsorption of cyanocobalamin (B12)	Imerslund-Grasbeck sydrome (IGS)
386	Sensenbrenner–Dorst–Owens syndrome	Sensenbrenner's syndrome Levin syndrome Cranioectodermal dysplasia (WDR35 mutation)
387	Sepiapterin reductase deficiency	
388	Set domain-containing protein 5 (SETD5) gene mutation	
389	Severe congenital nemaline myopathy (TNNT1 mutation)	Nemaline myopathy, Amish type (ANM) Nemaline myopathy 5A, autosomal recessive, severe infantile (NEM5A)

390	Severe infantile form of carnitine palmitoyltransferase II deficiency	
391	SH3 and multiple ankyrin repeat domains 2 (SHANK2) gene mutation	
392	Sharp's syndrome	Mixed collagen vascular disease Mixed connective tissue disease (MCTD)
393	Short QT syndrome 2	KCNQ1-related Short QT syndrome
394	SHORT syndrome	
395	Shprintzen-Goldberg syndrome (SGS)	Marfanoid craniosynostosis syndrome Shprintzen Goldberg craniosynostosis syndrome
396	Shukla-Vernon syndrome (SHUVER)	
397	Sialuria	
398	Sifrim-Hitz-Weiss syndrome	CHD4-related neurodevelopmental disorder
399	Signal transducer and activator of trascription 3 (STAT3) gene mutation	
400	Sjogren-Larsson syndrome	Fatty alcohol-nicotinamide adenine dinucleotide oxidoreductase deficiency
401	Skraban Deardorff syndrome	Intellectual disability, seizures, abnormal gait, facial dysmorphism syndrome
402	Small G protein signaling modulator 3 (SGSM3) mutation	
403	SMC3-related Cornelia de Lange syndrome (CdLS)	Cornelia de Lange syndrome 3
404	Smith Kingsmore syndrome	Macrocephaly, intellectual disability, neurodevelopmental disorder, small thorax (MINDS) syndrome
405	Smith-Lemli-Opitz syndrome	7-dehydrocholesterol reductase deficiency
406	Snijders Blok-Campeau syndrome	CHD3-related developmental delay, speech delay, intellectual disability, abnormalities of vision, facial dysmorphism syndrome
407	Snyder-Robinson syndrome	X-linked intellectual disability Snyder type
408	Sodium voltage-gated channel, alpha subunit 4 (SCN4A) gene mutation	
409	Solute carrier family 9, member 7 (SLC9A7) gene mutation	
410	Sotos' syndrome	Cerebral gigantism Nuclear receptor-binding set domain protein 1 (NSD1) gene mutation
411	SOX2 anophthalmia syndrome	Syndromic microphthalmia 3 Anophthalmia-esophageal-genital (AEG) syndrome
412	Special AT-rich sequence-bindind protein 2 (SATB2) associated syndrome	Glass syndrome Chromosome 2q32-q33 deletion syndrome Del(2)(q32q33) Monosomy 2q32q33
413	Spinal muscular atrophy, lower extremity-predominant 1, AD	DYNC1H1-related spinal muscular atrophy
414	Spinocerebellar ataxia type 21	SCY1-lime (SCYL1) gene mutation
415	Spinocerebellar ataxia type 26 (SCA26)	EEF2 related neurodevelopmental disorder
416	Spondilometaphyseal dysplasia	
417	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type (SEMDIST)	Spondyloepimetaphyseal dysplasia with severe short stature
418	Spondyloepiphyseal dysplasia (SED)	Spondyloepiphyseal dysplasia congenita (SEDC)

419	Spondylometaphyseal dysplasia with combined immunodeficiency; SPENCDI	Roifman-Melamed syndrome Spondyloenchondrodysplasia with immune dysregulation
420	SPTBN1 related developmental delay, impaired speech and behavioral abnormalities (DDISBA)	
421	Stankiewicz-Isidor syndrome (STISS)	PSMD12-related disorder
422	Streptococcus infection in conditions classified elsewhere and of unspecified site, Streptococcus, unspecified	
423	Synaptic RAS-GTPase-activating protein 1 (SYNGAP1) gene mutation	
424	Systematized epidermal nevus	Nevus unius lateris
425	Systemic mast cell disease (SMCD)	Systemic mastocytosis
426	SZT2 related developmental and epileptic encephalopathy 18	DEE18
427	SZT2 subunit of KICSTOR complex gene mutation	
428	Tatton Brown Rahman syndrome	DNA methyltranspherase 3A (DNMT3A) gene mutation Tall stature, intellectual disability, facial dysmorphism syndrome
429	Temple syndrome	
430	TET3-related Beck-Fahrner syndrome; TET3-BEFAHRS	
431	THUMPD1 related disorder	Neurodevelopmental disorder with speech delay and variable ocular anomalies (NEDSOA)
432	Timothy syndrome	Long QT syndrome type 8; LQT8 Long QT syndrome-syndactyly syndrome
433	Tooth agenesis-colorectal cancer syndrome (ODCRCS)	AXIN2-related disorder
434	TRAF7-associated heart defect, digital anomalies, facial dysmorphism, motor/speech delay	Tumor necrosis factor receptor associated factor 7-associated heart defect, digital anomalies, facial dysmorphism, motor and speech delay syndrome Cardiac, facial, and digital anomalies with developmental delay (CAFDADD)
435	Transcobalamin 2 (TCN2) deficiency	
436	Transketolase (TKT) deficiency	Short stature-developmental delay-congenital heart defect syndrome
437	Trichohepatoenteric syndrome	
438	Trichorhinophalangeal syndrome	
439	tRNA methyltransferase 10A (TRMT10A) gene mutation	
440	Turner's phenotype - ring chromosome karyotype	
441	Tyrosinemia type 1	FAH-related tyrosinemia Hepatorenal tyrosinemia
442	Tyrosinemia type 2	Hypertyrosinemia, Richner-Hanhart type Hypertyrosinemia, Oregon type
443	Tyrosinemia type 3	
444	Unverricht-Lundborg syndrome	Progressive myoclonic epilepsy type 1 Baltic myoclonus epilepsy

445	Usher syndrome type 3; USH3	Retinitis pigmentosa-deafness syndrome type 3
446	Ventriculomegaly with cystic kidney disease (VMCKD)	Heterozygous CRB2-related disorder
447	Verheij syndrome	8q24.3 microdeletion syndrome Del(8)(q24.3) Monosomy 8q24.3
448	Verloes Bourguignon syndrome	Autosomal recessive brachyolmia and amelogenesis imperfecta syndrome Dental anomalies and short stature (DAAS)
449	Ververi Brady syndrome	Glutamine rich 1-related intellectual disability, chondrodysplasia syndrome
450	Vici syndrome	Dionisi Vici Sabetta Gambarara syndrome
451	VPS11-related autosomal recessive hypomyelinating leukoencephalopathy	Leukodystrophy, hypomyelinating, 12 (HLD12) VPS11 core subunit of CORVET and HOPS complexes-related autosomal recessive hypomyelinating leukodystrophy
452	Waardenburg Shah syndrome	Waardenburg Hirschsprung syndrome
453	Weaver syndrome (WVS)	
454	Weill-Marchesani syndrome	Brachydactyly-spherophakia syndrome
455	Weiss-Kruszka syndrome (WSKA)	Zinc finger protein 462 (ZNF462) gene mutation
456	Wieacker Wolff syndrome	Intellectual disability, developmental delay, contracture syndrome Foot contracture, muscle atrophy, oculomotor apraxia syndrome
457	Wiedemann Steiner syndrome	Hypertrichosis, short stature, facial dysmorphism, developmental delay syndrome
458	Wilms tumor, aniridia, genitourinary anomalies and mental retardation (WAGR) syndrome	Chromosome 11p13 deletion syndrome
459	Wiscott-Aldrich syndrome	Aldrich syndrome
460	Witteveen Kolk syndrome (WITKOS)	SIN3A-related intellectual disability syndrome
461	Wolfram syndrome	Marquardt-Loriaux syndrome DIDMOAD (diabetes insipidus, diabetes mellitus, optic atrophy, deafness) syndrome
462	Wolman disease	Primary familial xanthomatosis with adrenal calcification Familial visceral xanthomatosis Deficiency of cholesterol esterase AND triacylglycerol lipase Wolman xanthomatosis Acid esterase deficiency
463	Xia-Gibbs syndrome	
464	X-linked intellectual disability Cabezas type	Cullin 4b (CUL4B) gene mutation
465	X-linked intellectual disability due to glutamate ionotropic receptor AMPA type subunit 3 mutations	GRIA3 related ASD Intellectual developmental disorder, X-linked syndromic, Wu type
466	X-linked intellectual disability Siderius type	
467	X-linked intellectual disability with cerebellar hypoplasia syndrome	Oligophrenin-1 syndrome (OPHN1)

468	X-linked intellectual disability, hypotonia, movement disorder syndrome	
469	X-linked non progressive cerebellar ataxia	
470	X-linked reticulate pigmentary disorder with systemic manifestation syndrome	Partington disease
471	Zhu Tokita Takenouchi Kim (ZTTK) syndrome	
472	Zinc Finger Homeobox 3 (ZFHX3)	
473	Zinc finger protein 292 (ZNF292) gene mutation	Intelectual developmental disorder, autosomal dominant 64
474	Zinc finger protein 335 (ZNF335) gene mutation	Microcephaly 10, primary, autosomal recessive