11/09/2025

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	14q22q23 microdeletion syndrome	Monosomy 14q22-q23 Frias Syndrome
2	16p11.2p12.2 microdeletion syndrome	Monosomy 16p11.2p12.2
3	16p13.11 microdeletion syndrome	Monosomy 16p13.11
4	3p25.3 deletion syndrome	Del(3)p(25.3) 3p25.3 microdeletion syndrome Monosomy 3p25.3 Mental retardation-epilepsy-stereotypic hand movement syndrome
5	3q26 microduplication syndrome	Dup(3q) syndrome Trisomy 3q26 Dup(3)(q26)
6	5q14.3 microdeletion syndrome	MEF2C haploinsufficiency syndrome
7	8p inverted duplication deletion syndrome	
8	9q22.3 deletion syndrome	Monosomy 9q22.3 syndrome Microdeletion 9q22.3
9	Abetalipoproteinemia	
10	Acetyl-coenzyme A acyltransferase	Acetyl-CoA acyltransferase 3-ketoacyl-CoA thiolase Acetyl-coenzyme A acyltransferase Beta-ketothiolase
11	Acquired epileptic aphasia	Landau-Kleffner syndrome
12	Acrocephalosyndactyly	Apert syndrome
13	Acrocephalosyndactyly type 5	Pfeiffer syndrome
14	Acrodysostosis	
15	Acute myeloid leukemia with RUNX1::RUNX1T1 fusion (morphologic abnormality)	Acute myeloid leukemia, t(8;21) (q22;q22.1)
16	Adams-Oliver syndrome	

17	Adenosine deaminase 2 (ADA2) gene mutation	Deficiency of adenosine deaminase 2 (DADA2)
18	Aggressive fibromatosis	Desmoid fibromatosis CTNNB1 gene mutation
19	Aicardi Goutieres syndrome	
20	Aicardi syndrome (AIC)	
21	Aicardi-Goutieres AGS syndrome 1, dominant and recessive (TREX1)	Chilblain lupus Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestation
22	Al Kaissi syndrome (ALKAS)	Cyclin-dependent kinase 10 (CDK10) gene mutation
23	Alacrima, achalasia, and impaired intellectual development syndrome (GMPPA)	GDP-MANNOSE PYROPHOSPHORYLASE A
24	Alexander's disease	Alexander syndrome
25	Allan-Herndon-Dudley syndrome	Monocarboxylate transporter 8 deficiency
26	Alstrom syndrome	
27	Alternating hemiplegia of childhood	
28	Ameloblastome	
29	Ankyrin 3 related intellectual disability, sleep disturbance syndrome (ANK-3)	
30	Arboleda Tham syndrome (ARTHS)	KAT6A gene mutation
31	Aromatic amino acid decarboxylase deficiency (AADC)	
32	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
33	Au Kline syndrome	Okamoto syndrome
34	Autosomal dominant central core disease	Ryanodine receptor 1 (RYR1) gene receptor Periodic paralysis
35	Autosomal dominant Charcot-Marie-Tooth disease type 2U	Methionyl-tRNA synthetase (MARS) gene mutation
36	Autosomal dominant HYPER-IgE syndrome due to STAT3 deficiency (HIES)	HYPER-IgE syndrome 1, AD Job syndrome
37	Autosomal recessive cerebellar ataxia with oculomotor apraxia type 1 (AOA1)	
38	Autosomal recessive cerebellar ataxia, epilepsy, intellectual disability syndrome - WWOX deficiency	Autosomal recessive spinocerebellar ataxia type 12 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12; SCAR12
39	Autosomal recessive spastic paraplegia type 15	Hereditary spastic paraparesis Kjellin syndrome
40	Axenfeld-Rieger syndrome	
41	Aymé-Gripp syndrome (AYGRP)	
42	Bainbridge Ropers syndrome	ASXL transcriptional regulator 3 deficiency syndrome

43	Baker Gordon syndrome	Synaptotagmin (SYT) 1-related neurodevelopmental disorder
44	Baraitser Winter cerebrofrontofacial syndrome	
45	Barakat syndrome	Hypoparathyroidism, sensorineural hearing loss, renal disease syndrome Hypoparathyroidism, deafness, renal disease (HDR) syndrome
46	Bardet-Biedl syndrome (BBS)	Laurence-Moon-Biedl syndrome Laurence-Moon-Bardet-Biedl syndrome (LMBB)
47	Bartter syndrome type 1	Hyperprostaglandin E syndrome type 1
48	Bartter syndrome type 2	Hyperprostaglandin E syndrome type 2 Hypokalemic alkalosis with hypercalciuria antenatal type 2
49	Bartter syndrome type 3	Bartter syndrome CLCNKB related disorder
50	BCL2L11-related disorder	2q13 deleteion syndrome
51	Beals syndrome	Congenital contractural arachnodactyly (CCA) Beals Hecht syndrome
52	Bernard Soulier syndrome	Hemorrhagiparous thrombocytic dystrophy Giant platelet syndrome
53	Blackfan-Diamond anemia	Aase syndrome Congenital pure red cell aplasia
54	Bohring-Opitz syndrome (BOS)	C-like syndrome
55	Borjeson-Forssman-Lehmann syndrome	
56	Brachydactyly mental retardation syndrome	Chromosome 2q37 deletion syndrome Albright hereditary osteodystrophy-like syndrome
57	Brain lung thyroid syndrome	Choreoathetosis with congenital hypothyroidism and neonatal respiratoy distress syndrome
58	Brain malformation with or without urinary tract defects	Nuclear factor I/A (NFIA) gene mutation
59	BRD4 related syndrome	Cornelia de Lange syndrome 6 (CDLS6)
60	Bromodomain and WD repeat-containing protein 3 (BRWD3)	
61	Bruton's agammaglobulinemia	X-linked agammaglobulinemia (XLA)
62	CACNA1A -related epilepsy	Developmental and epileptic encephalopathy 42 (DEE42) Early infantile epileptic encephalopathy 42 (EIEE42)
63	Camptodactyly, arthropathy, coxa-vara, pericarditis syndrome (CACP)	Jacobs syndrome
64	Capillary malformation	Congenital anomaly of capillary
65	CAPOS syndrome	Cerebellar ataxia, areflexia, pes cavus, optic atrophy, sensorineural hearing loss syndrome
66	Cardiofaciocutaneous syndrome 1 (CFC1)	Noonan syndrome 7
67	Cardiomyopathy, dilated, 1GG (SDHA)	Mitochondrial complex II deficiency, nuclear type 1 (SDH1) Neurodegeneration with ataxia and late-onset optic atrophy Pheochromocytoma/paraganglioma syndrome 5
68	Cat eye syndrome (CES)	Schachenmann's syndrome
69	Caudal duplication syndrome	Caudal duplication anomaly

70	CCR4-not transcription complex, subunit 3 (CNOT3) mutation	
71	Central core disease	Central core myopathy CONGENITAL MYOPATHY 1A, AUTOSOMAL DOMINANT, WITH SUSCEPTIBILITY TO MALIGNANT HYPERTHERMIA; CMYP1A
72	Cerebellar atrophy, developmental delay, and seizures (CADEDS)	
73	Cerebellar dysfunction with variable cognitive and behavioral abnormalities (CECBA)	CAMTA1 ASSOCIATED DISORDER Non-progressive cerebellar ataxia with intellectual disability
74	Cerebral folate transport deficiency	Cerebral folate deficiency
75	Cerebro-oculo-dento-auriculo-skeletal (CODAS) syndrome	
76	Cerebrotendinous xanthomatosis (CTX)	Cholestanol storage disease Van Bogaert-Scherer-Epstein syndrome Cerebral cholesterinosis Cerebrotendinous cholesterinosis
77	CHAMP1 related disease	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, IMPAIRED LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILD
78	Charcot-Marie-Tooth disease type 2N	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL DOMINANT, TYPE 2A1; CMT2A1 HEREDITARY MOTOR AND SENSORY NEUROPATHY IIA1; HMSN IIA1
79	Charcot-Marie-Tooth disease, axonal, type 2O	DYNC1H1-related Charcot-Marie-Tooth
80	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
81	Cholestasis, progressive familial intrahepatic 3 (PFIC3)	MDR3 DEFICIENCY ABCB4-related disorder
82	CHOPS syndrome	
83	Chromosomal imbalance syndrome	
84	Chronic bullous dermatosis of childhood	Linear IgA bullous disease in children Childhood linear IgA disease
85	Chronic granulomatous disease type II (CGD2)	
86	Chronic recurrent multifocal osteomyelitis (CRMO)	Majeed syndrome Synovitis acne pustulosis hyperostosis osteitis syndrome (SAPHO)
87	Ciliary dyskinesia, primary, 3 (CILD3)	DNAH5-related disorder
88	Citrullinemia type 2	Adult onset citrin deficiency
89	Clark Baraitser syndrome	
90	Clathrin (CLTC) gene mutation	Intelectual developmental disorder, autosomal dominant 56
91	Cleidocranial dysplasia (CCD)	Cleidocranial dysostosis (CLCD)
92	CLOVES syndrome	Congenital lipomatous overgrowth, vascular malformation, epidermal nevi, skeletal anomaly syndrome

93	CNOT 1 MUTATION RELATED VISSERS BODMER SYNDROME (VIBOS)	
94	CNOT3 related disorder	
95	Coats' disease	Leber's miliary aneurysms Miliary aneurysms of retina
96	Cobalamin C disease	Cobalamin locus C variant CblC methylmalonic acidemia and homocystinuria Methylmalonic acidemia with homocystinuria, type CBLC
97	Cockayne syndrome	
98	Coffin-Lowry syndrome (CLS)	
99	Coffin-Siris syndrome	Fifth digit syndrome
100	COL6A2 Betlehem Myopathy	Intermediate collagen VI-related muscular dystrophy
101	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
102	Combined immunodeficiency due to DOCK8 deficiency	
103	Combined oxidative phosphorilation deficiency 37 (COXPD37)	MICOS13-related disorder
104	Congenital central hypoventilation syndrome (CCHS)	Ondine curse Congenital pulmonary hypoventilation
105	Congenital chronic diarrhea with protein-losing enteropathy	Congenital chronic diarrhea with exudative enteropathy
106	Congenital dyserythropoietic anemia (CDA) type I	
107	Congenital ichthyosiform erythroderma (CIE)	Congenital recessive ichthyosis
108	Congenital ichtyosis with hypotrichosis syndrome	Autosomal recessive congenital ichthyosis
109	Congenital insensitivity to pain with anhidrosis (CIPA)	Hereditary sensory and autonomic neuropathy (HSAN) type 4 Swanson-Buchanan-Alvord neuropathy syndrome
110	Congenital malformation of dural sinus	
111	Congenital myasthenic syndrome	Congenital myasthenia syndrome
112	Congenital myasthenic syndrome type 9 (CMS9)	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency MUSK-related disorder
113	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
114	Congenital myopathy with myasthenic-like onset (RYR1)	
115	Congenital pseudoarthrosis of the tibia	

116	Congenital secretory diarrhea, chloride type	Congenital chloride diarrhea
117	Congenital stationary night blindness (CSNB)	Oguchi's disease
118	Convulsions, familial infantile, with paroxysmal choreoathetosis	PRRT2-related familial convulsions
119	COPB2 gene mutation-related disorder	Autosomal recessive primary microcephaly-19 (MCPH19)
120	Cortical dysplasia, complex, with other brain malformation 7 (CDCBM7)	TUBB2B-related disorder
121	Cortical dysplasia, complex, with other brain malformations 13	DYNC1H1-related cortical dysplasia
122	Craniofrontonasal dysplasia	Craniofrontonasal syndrome Craniofrontonasal dysostosis
123	Craniometaphyseal dysplasia	
124	Currarino triad	Currarino syndrome
125	Cutis marmorata telangiectatica congenita (CMTC)	Congenital livedo reticularis Van Lohuizen's syndrome
126	Cystathionine beta-synthase (CBS) deficiency	Deficiency of beta-thionase Deficiency of methylcysteine synthase Deficiency of serine sulphydrase
127	Cystic lymphangioma	Cystic hygroma
128	De Barsey syndrome (DBS)	Autosomal recessive cutis laxa type III Cutis laxa-corneal clouding-oligophrenia syndrome Progeroid syndrome of de Barsey de Barsey-Moens-Dierckx syndrome
129	Deficiency of dihydrofolate reductase	Dihydrofolate reductase deficiency
130	Deficiency of isomaltase	
131	Deficiency of prolidase	
132	Dehydrodolichyl diphosphate sythase (DHDDS) gene mutation	
133	Deletion of part of autosome	
134	Dent disease type 1	Nephrolithiasis, hypercalciuric, X-linked (NPHL2)
135	Dent's disease (DENT)	
136	Desmin myopathy	Desmin-related myofibrillar myopathy Desminopathy
137	Developmental and epileptic encephalopathy (GABRB3)	Epilepsy, childhood absence, susceptibility to, 5
138	Developmental and epileptic encephalopathy 64 (DEE64)	Epileptic encephalopathy, early infantile, 64 (EIEE64) RHOBTB2-related disorder
139	Developmental delay with or without dysmorphic facies and autism (DEDDFA)	Transformation/Transcription Domain-associated protein (TRRAP) associated disorder
140	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
141	Diaphyseal dysplasia	Camurati Engelmann syndrome
142	Diets Jongmans syndrome	KDM3B-related intellectual disability, facial dysmorphism, short stature syndrome

143	Diffuse intrinsic pontine glioma (DIPG)	
144	Distal anoctaminopathy	Miyoshi muscular dystrophy type 3 (MMD3) ANO5-related disorder
145	Distal trisomy 10q	Distal duplication 10q 10q trisomy
146	Distal Xq28 microduplication syndrome	Distal trisomy Xq28
147	DYRK1A-related intellectual disability syndrome	DYRK1A (dual specificity tyrosine phosphorylation regulated kinase 1A)
148	Dyskeratosis congenita (DKC)	Cole-Engmann-Zinsser syndrome
149	Dystonia 11, myoclonic (DYT11)	
150	Early infantile epileptic encephalopathy with suppression bursts	Ohtahara syndrome
151	Ectopia lentis syndrome	Familial ectopia lentis, Isolated ectopia lentis (IEL) ADAMTSL4 related lens ectopia
152	Emanuel syndrome	Supernumerary der(22)t(11;22) syndrome Der(22) syndrome due to 3:1 meiotic disjunction events Supernumerary derivative 22 chromosome syndrome
153	Encephalopathy due to defective mitochondrial and peroxisomal fission 2 (EMPF2)	DNM1L gene mutation 2
154	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1 (EMPF1)	DNM1L gene mutation 1
155	Enchondromatosis	Ollier disease
156	Epidermolysis bullosa	Acantholysis bullosa Fox disease
157	Epidermolysis bullosa acquisita	Acquired epidermolysis bullosa
158	Epidermolysis bullosa simplex 2A, generalized severe	Epidermolysis bullosa simplex 2A, Dowling-Meara type KRT5 related epidermolysis bullosa simplex, severe
159	Epilepsy, idiopathic generalized (EIG16)	
160	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)
161	Episodic kinesigenic dyskinesia 1	PRRT2-related kinesigenic dyskinesia
162	Erythema annulare	Figurate erythema
163	Erythropoietic protoporphyria (EPP)	Magnus syndrome Heme synthase deficiency
164	Factor X deficiency	Factor 10 deficiency Stuart-Prower factor deficiency

165	Familial amyloid nephropathy with urticaria and deafness	Muckle-Wells syndrome
166	Familial hemiplagic migraine type 1	Migraine type caused by mutations on the CACNA1A gene
167	Familial Hemophagocytic Lymphohistiocytosis (FHL)	Familial haemophagocytic reticulosis Familial HLH
168	Familial hypercholanemia	
169	Familial infantile bilateral striatal	
170	Familial lipoprotein lipase deficiency	Fredrickson type 1 hyperlipoproteinemia
171	Familial restrictive cardiomyopathy	
172	Familial Short QT syndrome	
173	FASTKD2 MUTATION RELATED	FAST kinase domains 2-related infantile
174	FBXO11 related disorder	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (IDDFBA)
175	Febrile infection related epilepsy syndrome (FIRES)	Fever-induced refractory epileptic encephalopathy in school-aged children (FIRES)
176	Feingold syndrome	Microcephaly-oculo-digito-esophageal-duodenal syndrome
177	Fibrodysplasia Ossificans Progressiva (FOP)	Progressive myositis ossificans Munchmeyer disease
178	Focal dermal hypoplasia (FDH)	Goltz-Gorlin syndrome
179	Forkhead box P1 (FOXP1) gene	Intellectual disability, severe speech delay, mild
180	Forsius-Eriksson syndrome	Aland Island Eye Disease (AIED)
181	Fructose-1,6-bisphosphate aldolase B	Hereditary fructosuria
182	GABA-transaminase deficiency	Gamma-aminobutiric acid transaminase deficiency
183	Galloway-Mowat syndrome	Nephrosis, neuronal dysmigration syndrome Galloway syndrome
184	Gastroparesis	Gastric atony Gastric stasis
185	GATAD2B-associated neurodevelopmental disorder (GAND)	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome
186	GBE1 GSD4	
187	Genitopatellar syndrome (GTPTS)	Absent patellae, scrotal hypoplasia, renal
188	Ghosal hematodiaphyseal dysplasia	Ghosal syndrome TBXAS1-related disorder
189	Giant congenital melanocytic nevus (GCMN)	שומה מלנוציטית מולדת ענקית

190	Gillespie syndrome	Aniridia, cerebellar ataxia, intellectual disability syndrome
191	Glucocorticoid deficiency with achalasia	Allgrove syndrome Triple A syndrome
192	Glutamate receptor, ionotropuc, N-methyl-D-aspartate, subunit 2B	Intellectual developmental disorder, autosomal dominant 6
193	Glutaric aciduria, type 1 (GA1)	Glutaric acidemia type 1 (GA1) Glutaryl-coenzyme A dehydrogenase deficiency
194	Glycogen storage disease III (GSD3)	Glycogen storage disease 3 Amylo-1,6-glucosidase deficiency Cori disease
195	Glycogen storage disease IXa (GSD9A)	
196	GMPPB HOMOZYGOUS VARIANT RELATED LGMB	Muscular dystrophy-dystroglycanopathy
197	Gordon hyperkalemia-hypertension syndrome	Pseudohypoaldosteronism type 2
198	Gorham syndrome	Gorham's disease Hemangiomata with osteolysis
199	GPATCH11RETINITIS PIGMENTOSA	
200	Greig cephalopolysyndactyly syndrome	
201	Growth factor, ERV1-like (GFER)	Congenital cataract, progressive muscular
202	Hamartoma of hypothalamus	Tuber cinereum hamartoma
203	Hao-Fountain syndrome	Ubiquitin-specific protease 7 (USP7) gene mutation Chromosome 16p13.2 deletion syndrome Intellectual developmental disorder with impaired
204	Harlequin ichthyosis	
205	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
206	Hemihyperplasia with multiple	
207	Hereditary hyperekplexia	Congenital stiff man syndrome Familial startle disease Kok disease
208	Hereditary Hypophosphatemic Rickets with Hypercalciuria (HHRH)	
209	Hereditary limphedema type 1	Milroy lymphedema Nonne-Milroy lymphedema
210	Hereditary pancreatitis	Protease, serine 1 (PRSS1) gene mutation

211	Hereditary pheochromocytoma and paraganglioma	
212	Hereditary sensory and autonomic	
213	Hereditary sensory and autonomic neuropathy (HSAN) type 9	Tectonin beta-propeller repeat-containing protein 2 (TECPR2) gene mutation
214	Hereditary spastic paraplegia (HSP)	Strumpell-Lorrain disease
215	Hermansky-Pudlak syndrome (HPS)	Albinism with hemorrhagic diathesis Alpha storage pool disease
216	Heterogeneous nuclear	
217	HHH syndrome (Hyperornithinemia, Hyperammonemia, Homocitrullinuria)	Triple H syndrome Ornithine translocase ORNT1 deficiency
218	Histiocytosis-lymphadenopathy plus syndrome	H syndrome SLC29A3 spectrum disorder
219	Homocystinuria	
220	Houge-Janssens syndrome 1 (HJS1)	
221	Houge-Janssens syndrome 2 (HJS2)	
222	Houge-Janssens syndrome 3 (HJS3)	
223	Human immunodeficiency virus (HIV)	אידס
224	Hyperekplexia epilepsy syndrome	ARHGEF9 RELATED DISORDER
225	Hyperinsulinemic hypoglycemia, familial, 1 (HHF1)	ABCC8-related disorder
226	Hypermethioninemia with S-	
227	Hyperphosphatemic familial tumoral calcinosis	
228	Hypertrophy (benign) of prostate with urinary obstruction and other lower urinary tract symptoms (LUTS)	
229	Hyperuricemia, pulmonary hypertension, renal failure, alkalosis	HUPRA syndrome
230	Hypotonia Hypoventilation Impaired Intellectual Development Dysautonomia Epilepsy and Eye	HIDEA syndrome
231	Hypotonia, Ataxia and Delayed Development syndrome (HADDS)	
232	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome (HADDTS)	CTBP1-related disorder
233	I-cell disease	Mucolipidosis II GNPTAB-related disorder
234	Ichthyosis, Congenital; Autosomal Recessive 1 (ARCI1)	COLLODION BABY, SELF-HEALING; SHCB ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 1, WITH BATHING SUIT
235	IDDHBA syndrome	Intellectual developmental disorder with hypotonia and behavioral abnormalities

236	Idiopathic non-lupus full-house nephropathy (FHN)	
237	Immunodeficiency 14	Activated PI3K-delta syndrome
238	Immunodysregulation, polyendocrinopathy, enteropathy, X-	Autoimmune enteropathy type 1
239	Impaired intellectual development and	Asadollahi-Rauch syndrome
240	Infantile Neuroaxonal Dystrophy (INAD1)	Seitelberger's disease
241	INOSITOL POLYPHOSPHATE-5-PHOSPHATASE K (INPP5K)	SKELETAL MUSCLE- AND KIDNEY-ENRICHED INOSITOL PHOSPHATASE (SKIP)
242	Intellectual developmenta disorder with severe speech and ambulation	ACTL6B gene mutation
243	INTELLECTUAL DEVELOPMENTAL DISORDER WITH IMPAIRED	
244	Intellectual developmental disorder, AR 13	TRAPPC9-related disorder
245	Intellectual developmental disorder, autosomal dominant 29 (MRD29)	SETBP1 haploinsufficiency disorder (SETBP1-HD)
246	Intellectual developmental disorder, autosomal dominant 45 (MRD45)	CIC gene mutation-related disorder
247	Intellectual developmental disorder, X-linked 96 (SYP)	SYNAPTOPHYSIN SYP gene mutation
248	Intellectual developmental disorder, X-linked 99 (USP9X)	UBIQUITIN-SPECIFIC PROTEASE 9, X-LINKED DROSOPHILA FAT FACETS-RELATED, X-LINKED
249	Intellectual developmental disorder, X-linked syndromic, Claes-Jensen type	KDM5C related syndromic X-linked intellectual disability Claes Jensen X-linked De Novo syndrome
250	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type	DDX3X related disorder
251	Intellectual disability Birk-Barel type	Intellectual disability, hypotonia, facial dysmorphism syndrome Birk Barel mental retardation dysmorphism syndrome KCNK9 (potassium two pore domain channel
252	Intellectual disability, expressive aphasia, facial dysmorphism syndrome	
253	Intellectual disability, facial dysmorphism syndrome due to SETD5 haploinsufficiency	

254	Intellectual disability-hypotonic facies syndrome, X-linked (MRXHF1)	Smith-Fineman-Myers syndrome 1 (SFMS) XLMR-hypotonic facies syndrome Carpenter-Waziri syndrome
255	Intractable diarrhea of infancy syndrome (IDIS)	Diarrhea 11, malabsorptive, congenital (DIAR11) Proline- and glutamate-rich protein with coiled-coil domain 1 (PERCC1) gene mutation
256	Intrauterine restriction, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomaly (IMAGe) syndrome	
257	Isovaleric acidemia	Isovaleryl-coenzyme A dehydrogenase deficiency Isovaleric aciduria
258	ITPR1 mutation related spinocerebellar ataxia 29	Spinocerebellar ataxia type 29 (SCA29) Congenital nonprogressive spinocerebellar ataxia
259	Jacobsen Syndrome	Distal partial deletion of long arm of chromosome 11
260	Jansen-de Vries syndrome (JDVS)	
261	Joubert syndrome	
262	Joubert syndrome with renal defect	NPHP1-related nephronophthisis
263	Juvenile Hypophosphatasia	Hypophosphatasia, childhood type
264	KBG syndrome	Short stature, facial and skeletal anomalies,
265	KIF1A associated neurological	
266	Kindler's syndrome	
267	KINSSHIP syndrome	
268	Kleefstra syndrome	due to del(9)(q34) due to monosomy 9q34
269	Kleefstra syndrome 2	
270	Klinefelter syndrome variant	49 XXXXY
271	Klippel Trenaunay syndrome	Klippel Trenaunay Weber syndrome
272	Kohlschutter syndrome	Amelocerebrohypohidrotic syndrome
273	Kostmann syndrome	Infantile agranulocytosis
274	Kozlowski spondylometaphyseal dysplasia	TRPV4 related skeletal dysplasia
275	KREMEN1 related ectodermal	Ectodermal dysplasia 13, hair/tooth type (ECTD13)
276	Lamb Shaffer syndrome	SOX5 haploinsufficiency syndrome
277	Lamellar Ichthyosis	Collodion baby
278	Lateral meningocele syndrome (LMNS)	NOTCH3-related lateral meningocele syndrome
279	Legius syndrome	Neurofibromatosis type 1-like syndrome (NFLS)
280	Leukocyte adhesion deficiency (LAD) type 2	
281	Liang-Wang syndrome (LIWAS)	

282	Limb-girdle Muscular Dystrophy, Type 28, Autosomal Recessive	LGMDR28
283	Lipid transport defect of intestine	Chylomicron retention disease
284	Lissencephaly, X-linked, 1 (LISX1)	Lissencephaly and agenesis of corpus callosum (XLIS)
285	Loeys-Dietz syndrome	
286	Long chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency (LCHADD)	
287	Low density lipoprotein receptor-	LRP5-related primary osteoporosis
288	Lowe syndrome	Cerebro-oculorenal dystrophy Oculocerebrorenal syndrome Lowe-Bickel syndrome
289	LSM1 GENE MUTATION RELATED DISORDER	LSM1 PROTEIN CANCER-ASSOCIATED SM-LIKE PROTEIN (CASM)
290	Lysine demethylase 5c (KDM5C) gene mutation	
291	LYSINE METHYLTRANSFERASE 5B (KMT5B)	Intellectual developmental disorder, autosomal dominant 51 SUPPRESSOR OF VARIEGATION 4-20, DROSOPHILA, HOMOLOG OF, 1 (SUV420H1) LYSINE-SPECIFIC METHYLTRANSFERASE 5B
292	Lysinuric protein intolerance (LPI)	Congenital lysinuria
293	MACROPHAGE EXPRESSED GENE 1 (MPEG1)	PERFORIN 2 (PRF2) MPG1 Immunodeficiency 77
294	Malan overgrowth syndrome	Sotos syndrome 2
295	Mandibulofacial dysostosis with microcephaly	Mandibulofacial dysostosis, Guion-Almeida type Elongation factor Tu GTP-binding comain-containing 2 (EFTUD2) gene mutation
296	MAP/MICROTUBULE AFFINITY-	ELKL MOTIF KINASE (EMK1)
297	Maple syrup urine disease (MSUD)	Branched chain 2-ketoacid dehydrogenase deficiency (BCKD) Ketoacidemia
298	Mast cell activation syndrome (MCAS)	Mast cell activation disorder (MCAD)

299	Maternally inherited Leigh syndrome (MILS)	Maternally inherited infantile subacute necrotizing encephalopathy
300	MBD5-associated neurodevelopmental disorder (MAND)	Intellectual developmental disorder, autosomal dominant 1
301	McCune Albright syndrome	
302	Megalencephaly capillary malformation (MCAP)	Macrocephaly-capillary malformation (M-CM, MCM) Megalencephaly-cutis marmorata telangiectatica
303	Meier-Gorlin syndrome	Ear, patella, short stature syndrome
304	MEIS2 syndrome	Cleft palate, cardiac defects, and impaired intellectual development Cardiac malformation, cleft lip/palate, microcephaly and digital anomalies
305	Metaphyseal anadysplasia type 2	Matrix metalloproteinase 9 (MMP9) gene mutation
306	Methyl-CpG-binding protein 2 (MECP2) gene mutation	
307	Methylmalonic acidemia	Methylmalonic aciduria (MMA)
308	Microcephalic osteodysplastic primordial dwarfism type II (MOPDII)	Majewski osteodysplastic primordial dwarfism type II
309	Microcephalic primordial dwarfism	Alazmai syndrome
310	Microcephaly, corpus callosum hypoplasia, intellectual disability, facial	
311	Minor partial trisomy	Minor partial chromosome duplication
312	Mitochondrial complex I deficiency, nuclear type 33 (NDUFA6)	
313	Mitochondrial	
314	MITOCHONDRIAL ENCEPHALOPATHY LACTIC	Juvenile myopathy, encephalopathy, lactic acidosis, stroke
315	Mitochondrial enoyl coA reductase protein-associated neurodegeneration (MEPAN) syndrome	Mitochondrial enoyl CoA hydratase 1 deficiency (ECHS1D) Mitochondrial enoyl coA reductase (MECP) gene mutation
316	Mixed gonadal dysgenesis	Mosaicism 45, X; 46, XY
317	Moebius syndrome	Congenital facial diplegia
318	Molybdenum cofactor deficiency	Combined molybdenoflavoprotein enzyme deficiency
319	Mosaic trisomy 16 syndrome	
320	Moyamoya disease	

321	MPPH syndrome	Megalencephaly, polymicrogyria, postaxial
322	Mulchandani-Bhoj-Conlin syndrome	maternal uniparental disomy of chromosome 20
323	MULLEGAMA-KLEIN-MARTINEZ SYNDROME STAG2 RELATED	
324	Multicentric carpotarsal osteolysis syndrome	
325	Multiple congenital exostosis	Multiple osteochondromatosis syndrome
326	Multiple endocrine neoplasia, type 3 (MEN3)	Multiple endocrine neoplasia, type 2B (MEN2B)
327	Multiple sulfatase deficiency (MSD)	Juvenile sulfatidosis, Austin type
328	Multisystemic smooth muscle dysfunction syndrome	Actin, alpha-2, smooth muscle, aorta (ACTA2) gene mutation
329	Muscle eye brain disease	
330	Muscle phosphoglycerate mutase deficiency	Phosphoglucomutase deficiency
331	Myhre syndrome	
332	Myoclonus Epilepsy and Ataxia due to potassium channel mutation (MEAK)	Progressive myoclonic epilepsy type 7 Progressive myoclonic epilepsy due to KV3.1
333	Myofibrillar myopathy	
334	Myosin, cardiac, heavy chain, beta (MYHCB, MYH7) gene mutation	
335	Nabais Sa-de Vries syndrome (SPOP)	SPECKLE-TYPE BTB/POZ PROTEIN (SPOP)
336	Nabais Sa-de Vries syndrome, type 2 (NSDVS2)	NEURODEVELOPMENTAL DISORDER WITH RELATIVE MACROCEPHALY AND WITH OR
337	NARS2 related disorder	Deafness. Autosomal recessive 94 (DFNB94)
338	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
339	Neurodegeneration due to 3-	Methacrylic aciduria
340	Neurodegeneration, childhood-onset, with brain atrophy (CONDBA)	UBTF related motor and cognitive regression syndrome
341	Neurodevelopmental disorder w/ hypotonia, variable intellectual + behavioral abnormalities (NEDHIB)	POLR2A-related disorder
342	Neurodevelopmental disorder with dysmorphic facies and distal limb	BPTF-related disorder
343	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies (NEDDFSA)	Zinc finger miz-domain containig 1(ZMIZ1) gene mutation
344	Neurodevelopmental disorder with eye movement abnormalities and ataxia (NEDEMA)	FRMD5-related disorder

345	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without	TRPM3-related disorder
346	Neuroendocrine cell hyperplasia of infancy	NEHI syndrome
347	N-glycanase 1 congenital disorder of deglycosylation (NGLY1)	Alacrimia, choreoathetosis, liver dysfunction syndrom Deficiency of N-glycanase 1
348	Nicolaides-Baraitser syndrome	SWI/SNF-related, matrix-associated, actin-dependant regulator of chromatin, subfamily A,
349	Niemann-Pick disease, type A	
350	Niemann-Pick disease, type C	Supraoptic vertical ophthalmoplegia
351	NOG related disorder	Teunissen-Cremers syndrome
352	NPHS2 related nephrotic syndrome	Familial idiopathic steroid-resistant nephrotic
353	Obesity, morbid, due to leptin receptor deficiency (LEPR)	OBR
354	Occipital pachygyria and polymicrogyria	LAMB1-related disorder
355	Oculo-facio-cardio-dental syndrome	Oculofaciocardiodental syndrome
356	O'Donnell-Luria-Rodan syndrome (ODLURO)	Lysine-specific methyltransferase 2E (KMT2E) gene mutation
357	Odonto-onycho-dermal dysplasia	Ectodermal dysplasia 16, hair/tooth/nail type
358	Ogden syndrome	
359	Ohdo sydrome, Say-Barber-Biesecker-Young-Simpson (SBBYS) variant	Blepharophimosis, intellectual disability syndrome Say-Barber-Biesecker-Young-Simpson (SBBYS) variant
360	Okur-Chung neurodevelopmental	OKUR CHUNG SYNDROME -CSNK2A1
361	PACS1 NEURODEVELOPMENTAL DISORDER	PHOSPHOFURIN ACIDIC CLUSTER SORTING PROTEIN Schuurs-Hoeijmakers syndrome PACS RELATED EPILEPTIC ENCEPHALOPATHY
362	Pallister Killian syndrome	Tetrasomy 12p syndrome Killian-Teschler-Nicola syndrome
363	Paraneoplastic opsoclonus-myoclonus-ataxia syndrome; POMA	Dancing eye syndrome Dancing eye-dancing feet syndrome Opsoclonus-myoclonus-ataxia syndrome; OMA Kinsbourne syndrome
364	Parkes Weber syndrome	

365	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy (PNKD3)	Generalized epilepsy and paroxysmal dyskinesia (GEPD)
366	Pearson's syndrome	
367	Pediatric multiple sclerosis	
368	Pelizaeus-Merzbacher disease (PMD)	Sudanophilic leukodystrophy
369	Penta X syndrome	XXXXX syndrome
370	Perching syndrome	Kelch like family member 7-related Bohring-Opitz-like and Crisponi/cold-induced sweating-like overlap
371	Perlman syndrome	
372	Permanent neonatal diabetes mellitus	Developmental delay, epilepsy and neonatal diabetes 1 (DEND)
373	Peutz Jeghers syndrome PJS	Peutz-Jeghers polyposis
374	PHACE syndrome	Phace syndrome and Moya Moya Disease Pascual-Castroviejo syndrome type 2
375	PHACES syndrome	
376	PHD finger protein 21A (PHF21A) gene mutation	
377	Phelan-McDermid syndrome	22q13.3 deletion syndrome
378	PHIP-related syndrome	Pleckstrin homology domain interacting protein-related behavioral problems, intellectual disability,
379	Phocomelia - Partial congenital absence of limb	Femorotibiofibular intercalary transverse meromelia Humero-radio-ulnar intercalary transverse meromelia
380	Phosphatase and tensin homolog (PTEN) gene mutation	
381	Phosphoenolpyruvate carboxykinase (PEPCK-C) deficiency	
382	Pitt-Hopkins syndrome (PTHS)	
383	Pitt-Hopkins-like syndrome 1 (PTHSL1)	
384	Pituitary stalk interruption syndrome	Ectopis neurohypophysis
385	Pleuropulmonary blastoma (PPB)	

386	Pleuropulmonary blastoma familial	DICER1 tumor-predisposition syndrome
387	Poirier-Bienvenu neurodevelopmental	CSNK2B-related disorder
388	POMT1-related limb-girdle muscular dystrophy R11	Limb-girdle muscular dystrophy type 2K; LGMD type 2K POMT1-related LGMD R11
389	Pontocerebellar hypoplasia type 9 (PCH9)	
390	Popliteal pterygium syndrome	
391	Port-wine stain with oculocutaneous melanosis	Phakomatosis pigmentovascularis
392	Potocki Lupski syndrome	17p11.2 microduplication
393	Potocki-Shaffer syndrome	Chromosome 11p11.2 deletion syndrome Proximal 11p deletion syndrome (P11pDS)
394	PPP2R1A	Houge-Janssens syndrome 2
395	PRICKLE1 HOMOZYGOUS MUTATION RELATED DISORDER	Progressive myoclonus epilepsy with ataxia
396	Primary hyperoxaluria type 1	Glycolic aciduria
397	Primary microcephaly, mild intellectual disability, young-onset diabetes	
398	Primrose syndrome	Intellectual disability, cataract, calcified pinna,
399	PRKAG2-related hypertrophic cardiomyopathy	Familial hypertrophic cardiomyopathy 6
400	Progeria syndrome	Hutchinson-Gilford syndrome
401	Progressive myoclonic epilepsy (PME) type 8 (CERS1)	
402	Propionic Acidemia	Ketotic hyperglycinaemia
403	Proteolipid protein 1(PLP1) gene mutation	
404	Pseudohypoaldosteronism type 1, recessive form	Pseudohypoaldosteronism type 1B Pseudohypoaldosteronism, Persian-Jewish type
405	Pseudohypoparathyroidism	Guanine nucleotide-binding protein, alpha-stimulating activity polypeptide 1 (GNAS) gene
406	Pseudohypoparathyroidism type 1A (PHP1A)	Albright hereditary osteodystrophy (AHO)
407	Pseudohypoparathyroidism type 2	
408	Pseudotumor cerebri	Benign intracranial hypertension; BIH Nonne's syndrome
409	PTEN hamartoma tumor syndrome	Bannayan syndrome
410	Pulmonary alveolar proteinosis (PAP)	

411	Purine rich element binding protein A (PURA) syndrome	
412	Pyknodysostosis	Maroteaux-Lamy syndrome type II
413	PYRUVATE DEHYDROGENASE COMPLEX, COMPONENT X (PDHX)	PDX1 PYRUVATE DEHYDROGENASE COMPLEX, E3-
414	Radial aplasia-trhombocytopenia (TAR) syndrome	
415	Radioulnar synostosis with amegakayocytic thrombocytopenia (RUSAT) 2	MDS1 and EVI1 complex (MECOM) gene mutation
416	Rahman syndrome	H1-4-related neurodevelopmental disorder HIST1H1E-related disorder
417	RAS-associated autoimmune leukoproliferative disease (RALD)	Autoimmune leukoproliferative syndrome type 4
418	Rasmussen subacute encephalitis	Rasmussen syndrome Rasmussen encephalitis
419	Rauch-Steindl syndrome	
420	Raynaud-Claes syndrome	Chloride voltage-gated channel 4-related X-linked intellectual disability syndrome
421	Renal nutcracker syndrome	Left renal vein entrapment syndrome
422	Renpenning syndrome	Golabi-Ito-Hall syndrome Hamel cerebropalatocardiac syndrome Polyglutamine-binding protein 1 (PQBP1) gene mutation
423	Resistance to insulin-like growth factor 1 (IGF1)	Insulin-like growth factor 1 receptor (IGF1R) gene mutation
424	Rett syndrome; RTS	Rett's disorder Cerebroatrophic hyperammonemia
425	ROHHAD syndrome	Rapid-onset childhood obesity, hypothalamic dysfunction, hypoventilation, autonomic
426	Rubinstein-Taybi syndrome	
427	Russell-Silver syndrome	Silver syndrome Silver-Russell dwarfism
428	SARCOGLYCAN, ALPHA (SGCA)	ADHALIN (ADL)

429	Schaaf Yang syndrome	MAGE family member L2-related Prader-Willi-like syndrome
430	Schinzel-Giedion syndrome	
431	Schuurs-Hoeijmakers syndrome	
432	Scimitar syndrome	Pulmonary venolobar syndrome
433	Seizures, benign familial infantile, 2	PRRT2-related benign familial seizures
434	Selective malabsorption of	Imerslund-Grasbeck sydrome (IGS)
435	Sensenbrenner–Dorst–Owens	Sensenbrenner's syndrome
436	Sepiapterin reductase deficiency	
437	Set domain-containing protein 5 (SETD5) gene mutation	
438	Severe congenital nemaline myopathy	Nemaline myopathy, Amish type (ANM)
439	Severe infantile form of carnitine palmitoyltransferase II deficiency	
440	SH3 and multiple ankyrin repeat domains 2 (SHANK2) gene mutation	
441	Sharp's syndrome	Mixed collagen vascular disease Mixed connective tissue disease (MCTD)
442	Short QT syndrome 2	KCNQ1-related Short QT syndrome
443	SHORT syndrome	
444	Shprintzen-Goldberg syndrome (SGS)	Marfanoid craniosynostosis syndrome
445	Shukla-Vernon syndrome (SHUVER)	
446	Sialuria	
447	Sifrim-Hitz-Weiss syndrome	CHD4-related neurodevelopmental disorder
448	Signal transducer and activator of	
449	Sjogren-Larsson syndrome	Fatty alcohol-nicotinamide adenine dinucleotide
450	Skraban Deardorff syndrome	Intellectual disability, seizures, abnormal gait, facial dysmorphism syndrome
451	Small G protein signaling modulator 3 (SGSM3) mutation	
452	SMC3-related Cornelia de Lange syndrome (CdLS)	Cornelia de Lange syndrome 3
453	Smith Kingsmore syndrome	Macrocephaly, intellectual disability, neurodevelopmental disorder, small thorax (MINDS)
454	Snijders Blok-Campeau syndrome	CHD3-related developmental delay, speech delay, intellectual disability, abnormalities of vision, facial
455	Snyder-Robinson syndrome	X-linked intellectual disability Snyder type

456	Sodium voltage-gated channel, alpha	
457	Solute carrier family 9, member 7 (SLC9A7) gene mutation	
458	Sotos' syndrome	Cerebral gigantism
459	SOX2 anophthalmia syndrome	Syndromic microphthalmia 3 Anophthalmia-esophageal-genital (AEG) syndrome
460	Special AT-rich sequence-bindind protein 2 (SATB2) associated	Glass syndrome Chromosome 2q32-q33 deletion syndrome
461	Spinal muscular atrophy, lower extremity-predominant 1, AD	DYNC1H1-related spinal muscular atrophy
462	Spinocerebellar ataxia type 21	SCY1-lime (SCYL1) gene mutation
463	Spinocerebellar ataxia type 26 (SCA26)	EEF2 related neurodevelopmental disorder
464	Spondilometaphyseal dysplasia	
465	Spondyloepimetaphyseal dysplasia,	Spondyloepimetaphyseal dysplasia with severe
466	Spondyloepiphyseal dysplasia (SED)	Spondyloepiphyseal dysplasia congenita (SEDC)
467	Spondylometaphyseal dysplasia	Spondylometaphyseal dysplasia Algerian type
468	Spondylometaphyseal dysplasia with combined immunodeficiency;	Roifman-Melamed syndrome Spondyloenchondrodysplasia with immune
469	SPTBN1 related developmental delay,	
470	Stankiewicz-Isidor syndrome (STISS)	PSMD12-related disorder
471	Streptococcus infection in conditions classified elsewhere and of unspecified site, Streptococcus,	
472	Synaptic RAS-GTPase-activating protein 1 (SYNGAP1) gene mutation	
473	Systematized epidermal nevus	Nevus unius lateris
474	Systemic mast cell disease (SMCD)	Systemic mastocytosis
475	SZT2 related developmental and epileptic encephalopathy 18	DEE18
476	SZT2 subunit of KICSTOR complex	
477	Tatton Brown Rahman syndrome	DNA methyltranspherase 3A (DNMT3A) gene
478	Temple syndrome	
479	TET3-related Beck-Fahrner syndrome; TET3-BEFAHRS	
480	TETRATRICOPEPTIDE REPEAT DOMAIN-CONTAINING PROTEIN 2 (TTC26)	INTRAFLAGELLAR TRANSPORT 56, CHLAMYDOMONAS, HOMOLOG OF (IFT56) Biliary, renal, neurologic, and skeletal syndrome (BRENS)

481	THUMPD1 related disorder	Neurodevelopmental disorder with speech delay
482	THYROID HORMONE RECEPTOR, ALPHA-1 (THRA)	ERBA-ALPHA
483	Timothy syndrome	Long QT syndrome type 8; LQT8 Long QT syndrome-syndactyly syndrome
484	Tooth agenesis-colorectal cancer syndrome (ODCRCS)	AXIN2-related disorder
485	TRAF7-associated heart defect, digital anomalies, facial dysmorphism,	Tumor necrosis factor receptor associated factor 7-associated heart defect, digital anomalies, facial
486	Transcobalamin 2 (TCN2) deficiency	
487	Transketolase (TKT) deficiency	Short stature-developmental delay-congenital heart defect syndrome
488	Trichohepatoenteric syndrome	
489	Trichorhinophalangeal syndrome	
490	tRNA methyltransferase 10A (TRMT10A) gene mutation	
491	tRNA-VAL, MITOCHONDRIAL(MT-	
492	Turner's phenotype - ring	
493	Tyrosinemia type 1	FAH-related tyrosinemia Hepatorenal tyrosinemia
494	Tyrosinemia type 2	Hypertyrosinemia, Richner-Hanhart type Hypertyrosinemia, Oregon type
495	Tyrosinemia type 3	
496	Unverricht-Lundborg syndrome	Progressive myoclonic epilepsy type 1 Baltic myoclonus epilepsy
497	Usher syndrome type 3; USH3	Retinitis pigmentosa-deafness syndrome type 3
498	Ventriculomegaly with cystic kidney	Heterozygous CRB2-related disorder
499	Verheij syndrome	8q24.3 microdeletion syndrome Del(8)(q24.3)
500	Verloes Bourguignon syndrome	Autosomal recessive brachyolmia and
501	Ververi Brady syndrome	Glutamine rich 1-related intellectual disability, chondrodysplasia syndrome
502	Vici syndrome	Dionisi Vici Sabetta Gambarara syndrome
503	VPS11-related autosomal recessive hypomyelinating leukoencephalopathy	Leukodystrophy, hypomyelinating, 12 (HLD12) VPS11 core subunit of CORVET and HOPS complexes-related autosomal recessive
504	Waardenburg Shah syndrome	Waardenburg Hirschsprung syndrome
505	Weaver syndrome (WVS)	

506	Weill-Marchesani syndrome	Brachydactyly-spherophakia syndrome
507	Weiss-Kruszka syndrome (WSKA)	Zinc finger protein 462 (ZNF462) gene mutation
508	Wieacker Wolff syndrome	Intellectual disability, developmental delay,
509	Wiedemann Steiner syndrome	Hypertrichosis, short stature, facial dysmorphism,
510	Wilms tumor, aniridia, genitourinary	Chromosome 11p13 deletion syndrome
511	Wiscott-Aldrich syndrome	Aldrich syndrome
512	Wiskott-Aldrich syndrome (WAS)	Eczema, thrombocytopenia, immunodeficiency syndrome
513	Witteveen Kolk syndrome (WITKOS)	SIN3A-related intellectual disability syndrome
514	Wolfram syndrome	Marquardt-Loriaux syndrome
515	Wolman disease	Primary familial xanthomatosis with adrenal calcification
516	X LINKED CLCN5 RELATED DENT 1	CHLORIDE CHANNEL, VOLTAGE-GATED, K2
517	X LINKED IQSEC2	IQSEC2-related syndromic intellectual disability IQSEC2 (IQ motif and Sec7 domain 2) related syndromic intellectual disability
518	Xeroderma Pigmentosum (XP)	Angioma pigmentosum atrophicum Atrophoderma pigmentosum Kaposi dermatosis Melanosis lenticularis progressiva Pigmented epitheliomatosis Xeroderma of Kaposi
519	Xia-Gibbs syndrome	
520	X-linked intellectual disability Cabezas type	Cullin 4b (CUL4B) gene mutation
521	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
522	X-linked intellectual disability Siderius	
523	X-linked intellectual disability with cerebellar hypoplasia syndrome	Oligophrenin-1 syndrome (OPHN1)

524	X-linked intellectual disability, hypotonia, movement disorder syndrome	
525	X-linked non progressive cerebellar ataxia	
526	X-linked reticulate pigmentary disorder with systemic manifestation syndrome	Partington disease
527	Zhu Tokita Takenouchi Kim (ZTTK) syndrome	
528	ZINC FINGER- AND BTB DOMAIN-CONTAINING PROTEIN 18 (ZBTB18)	Intellectual developmental disorder, autosomal dominant 22
529	Zinc Finger Homeobox 3 (ZFHX3)	
530	ZINC FINGER MYND DOMAIN-CONTAINING PROTEIN 11	BS69
531	Zinc finger protein 292 (ZNF292) gene mutation	Intellectual developmental disorder, autosomal dominant 64
532	Zinc finger protein 335 (ZNF335) gene	Microcephaly 10, primary, autosomal recessive
533	#REF!	#REF!
534	#REF!	#REF!