18/02/2025

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, בהתאם לשאלון עומס טיפולי שפורסם ע"י הלשכה הרפואית.

#	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	Alagille syndrome	Arteriohepatic dysplasia Watson-Alagille syndrome
20	Alexander's disease	Alexander syndrome
21	Allan-Herndon-Dudley syndrome	Monocarboxylate transporter 8 deficiency
22	Alstrom syndrome	
23	Alternating hemiplegia of childhood	
24	Ameloblastome	

25	Arboleda Tham syndrome (ARTHS)	KAT6A gene mutation
26	Aromatic amino acid decarboxylase deficiency (AADC)	
27	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
28	Au Kline syndrome	Okamoto syndrome
29	Autosomal dominant central core disease	Ryanodine receptor 1 (RYR1) gene receptor Periodic paralysis
30	Autosomal dominant Charcot-Marie-Tooth disease type 2U	Methionyl-tRNA synthetase (MARS) gene mutation
31	Autosomal dominant HYPER-IgE syndrome due to STAT3 deficiency (HIES)	HYPER-IgE syndrome 1, AD Job syndrome
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 malformation with or without urinary tract defects	Nuclear factor I/A (NFIA) gene mutation

50	BRD4 related syndrome	Cornelia de Lange syndrome 6 (CDLS6)
51	Bromodomain and WD repeat-containing protein 3 (BRWD3)	
52	Bruton's agammaglobulinemia	X-linked agammaglobulinemia (XLA)
53	CACNA1A -related epilepsy	Developmental and epileptic encephalopathy 42 (DEE42) Early infantile epileptic encephalopathy 42 (EIEE42)
54	Capillary malformation	Congenital anomaly of capillary
55	CAPOS syndrome	Cerebellar ataxia, areflexia, pes cavus, optic atrophy, sensorineural hearing loss syndrome
56	Cardiofaciocutaneous syndrome 1 (CFC1)	Noonan syndrome 7
57	Caudal duplication syndrome	Caudal duplication anomaly
58	CCR4-not transcription complex, subunit 3 (CNOT3) mutation	
59	Central core disease	Central core myopathy CONGENITAL MYOPATHY 1A, AUTOSOMAL DOMINANT, WITH SUSCEPTIBILITY TO MALIGNANT HYPERTHERMIA; CMYP1A
60	Cerebellar atrophy, developmental delay, and seizures (CADEDS)	
61	Cerebellar dysfunction with variable cognitive and behavioral abnormalities (CECBA)	CAMTA1 ASSOCIATED DISORDER Non-progressive cerebellar ataxia with intellectual disability
62	Cerebral folate transport deficiency	Cerebral folate deficiency
63	Cerebro-oculo-dento-auriculo-skeletal (CODAS) syndrome	
64	Cerebrotendinous xanthomatosis (CTX)	Cholestanol storage disease Van Bogaert-Scherer-Epstein syndrome Cerebral cholesterinosis Cerebrotendinous cholesterinosis
65	CHAMP1 related disease	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, IMPAIRED LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILD
66	Charcot-Marie-Tooth disease type 2N	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL DOMINANT, TYPE 2A1; CMT2A1 HEREDITARY MOTOR AND SENSORY NEUROPATHY IIA1; HMSN IIA1
67	Charcot-Marie-Tooth disease, axonal, type 2O	DYNC1H1-related Charcot-Marie-Tooth
68	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
69	CHOPS syndrome	
70	Chronic recurrent multifocal osteomyelitis (CRMO)	Majeed syndrome Synovitis acne pustulosis hyperostosis osteitis syndrome (SAPHO)
71	Ciliary dyskinesia, primary, 3 (CILD3)	DNAH5-related disorder
72	Citrullinemia type 2	Adult onset citrin deficiency

73	Clark Baraitser syndrome	
74	Clathrin (CLTC) gene mutation	Intelectual developmental disorder, autosomal dominant 56
75	CLOVES syndrome	Congenital lipomatous overgrowth, vascular malformation, epidermal nevi, skeletal anomaly syndrome
76	CNOT3 related disorder	
77	Coats' disease	Leber's miliary aneurysms Miliary aneurysms of retina
78	Cobalamin C disease	Cobalamin locus C variant CblC methylmalonic acidemia and homocystinuria Methylmalonic acidemia with homocystinuria, type CBLC
79	Coffin-Lowry syndrome (CLS)	
80	Coffin-Siris syndrome	Fifth digit syndrome
81	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
82	Congenital central hypoventilation syndrome (CCHS)	Ondine curse Congenital pulmonary hypoventilation
83	Congenital chronic diarrhea with protein-losing enteropathy	Congenital chronic diarrhea with exudative enteropathy
84	Congenital ichthyosiform erythroderma (CIE)	Congenital recessive ichthyosis
85	Congenital ichtyosis with hypotrichosis syndrome	Autosomal recessive congenital ichthyosis
86	Congenital insensitivity to pain with anhidrosis (CIPA)	Hereditary sensory and autonomic neuropathy (HSAN) type 4 Swanson-Buchanan-Alvord neuropathy syndrome
87	Congenital malformation of dural sinus	
88	Congenital myasthenic syndrome	Congenital myasthenia syndrome
89	Congenital myasthenic syndrome type 9 (CMS9)	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency MUSK-related disorder
90	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
91	Congenital pseudoarthrosis of the tibia	
92	Congenital secretory diarrhea, chloride type	Congenital chloride diarrhea
93	Congenital stationary night blindness (CSNB)	Oguchi's disease
94	Convulsions, familial infantile, with paroxysmal choreoathetosis	PRRT2-related familial convulsions
95	COPB2 gene mutation-related disorder	Autosomal recessive primary microcephaly-19 (MCPH19)

96	Cortical dysplasia, complex, with other brain malformation 7 (CDCBM7)	TUBB2B-related disorder
97	Cortical dysplasia, complex, with other brain malformations 13	DYNC1H1-related cortical dysplasia
98	Craniofrontonasal dysplasia	Craniofrontonasal syndrome Craniofrontonasal dysostosis
99	Craniometaphyseal dysplasia	
100	Currarino triad	Currarino syndrome
101	Cutis marmorata telangiectatica congenita (CMTC)	Congenital livedo reticularis Van Lohuizen's syndrome
102	Cystathionine beta-synthase (CBS) deficiency	Deficiency of beta-thionase Deficiency of methylcysteine synthase Deficiency of serine sulphydrase
103	Cystic lymphangioma	Cystic hygroma
104	Deficiency of dihydrofolate reductase	Dihydrofolate reductase deficiency
105	Deficiency of isomaltase	
106	Deficiency of prolidase	
107	Dehydrodolichyl diphosphate sythase (DHDDS) gene mutation	
108	Deletion of part of autosome	
109	Dent disease type 1	Nephrolithiasis, hypercalciuric, X-linked (NPHL2)
110	Desmin myopathy	Desmin-related myofibrillar myopathy Desminopathy
111	Developmental delay with or without dysmorphic facies and autism (DEDDFA)	Transformation/Transcription Domain-associated protein (TRRAP) associated disorder
112	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
113	Diaphyseal dysplasia	Camurati Engelmann syndrome
114	Diffuse intrinsic pontine glioma (DIPG)	
115	Distal trisomy 10q	Distal duplication 10q 10q trisomy
116	Distal Xq28 microduplication syndrome	Distal trisomy Xq28
117	DYRK1A-related intellectual disability syndrome	DYRK1A (dual specificity tyrosine phosphorylation regulated kinase 1A)
118	Dyskeratosis congenita (DKC)	Cole-Engmann-Zinsser syndrome
119	Dystonia 11, myoclonic (DYT11)	
120	Early infantile epileptic encephalopathy with suppression bursts	Ohtahara syndrome
121	Ectopia lentis syndrome	Familial ectopia lentis, Isolated ectopia lentis (IEL) ADAMTSL4 related lens ectopia
122	Emanuel syndrome	Supernumerary der(22)t(11;22) syndrome Der(22) syndrome due to 3:1 meiotic disjunction events Supernumerary derivative 22 chromosome syndrome

123	Encephalopathy due to defective mitochondrial and peroxisomal fission 2 (EMPF2)	DNM1L gene mutation 2
124	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1 (EMPF1)	DNM1L gene mutation 1
125	Enchondromatosis	Ollier disease
126	Epidermolysis bullosa	Acantholysis bullosa Fox disease
127	Epidermolysis bullosa acquisita	Acquired epidermolysis bullosa
128	Epidermolysis bullosa simplex 2A, generalized severe	Epidermolysis bullosa simplex 2A, Dowling-Meara type KRT5 related epidermolysis bullosa simplex, severe
129	Epilepsy, idiopathic generalized (EIG16)	
130	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)
131	Episodic kinesigenic dyskinesia 1	PRRT2-related kinesigenic dyskinesia
132	Erythema annulare	Figurate erythema
133	Erythropoietic protoporphyria (EPP)	Magnus syndrome Heme synthase deficiency
134	Facioscapulohumeral muscular dystrophy (FMD, FSHD)	Landouzy-Dejerine muscular dystrophy
135	Factor X deficiency	Factor 10 deficiency Stuart-Prower factor deficiency
136	Familial amyloid nephropathy with urticaria and deafness	Muckle-Wells syndrome
137	Familial hemiplagic migraine type 1	Migraine type caused by mutations on the CACNA1A gene
138	Familial Hemophagocytic Lymphohistiocytosis (FHL)	Familial haemophagocytic reticulosis Familial HLH
139	Familial hypercholanemia	
140	Familial infantile bilateral striatal necrosis (IBSN)	
141	Familial lipoprotein lipase deficiency	Fredrickson type 1 hyperlipoproteinemia
142	Familial restrictive cardiomyopathy	
143	Familial Short QT syndrome	
144	FBXO11 related disorder	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (IDDFBA)
145	Febrile infection related epilepsy syndrome (FIRES)	Fever-induced refractory epileptic encephalopathy in school-aged children (FIRES)
146	Fibrodysplasia Ossificans Progressiva (FOP)	Progressive myositis ossificans Munchmeyer disease
147	Focal dermal hypoplasia (FDH)	Goltz-Gorlin syndrome

148	Forkhead box P1 (FOXP1) gene mutation	Intellectual disability, severe speech delay, mild dysmorphism syndrome
149	Forsius-Eriksson syndrome	Aland Island Eye Disease (AIED)
150	Fructose-1,6-bisphosphate aldolase B (ALDOB) deficiency	Hereditary fructosuria Aldolase B (ALDB) deficiency
151	GABA-transaminase deficiency	Gamma-aminobutiric acid transaminase deficiency
152	Galloway-Mowat syndrome	Nephrosis, neuronal dysmigration syndrome Galloway syndrome Microcephaly, hiatus hernia, nephrotic syndrome
153	Gastroparesis	Gastric atony Gastric stasis
154	GATAD2B-associated neurodevelopmental disorder (GAND)	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome GAND syndrome Mental retardation, autosomal dominant 18 (MRD18)
155	Genitopatellar syndrome (GTPTS)	Absent patellae, scrotal hypoplasia, renal anomalies, facial dysmorphism, and mental retardation KAT6B-related disorder
156	Ghosal hematodiaphyseal dysplasia	Ghosal syndrome TBXAS1-related disorder
157	Giant congenital melanocytic nevus (GCMN)	שומה מלנוציטית מולדת ענקית
158	Gillespie syndrome	Aniridia, cerebellar ataxia, intellectual disability syndrome
159	Glanzmann's thrombasthenia	Glanzmann's disease Hereditary hemorrhagic thromboasthenia
160	Glucocorticoid deficiency with achalasia	Allgrove syndrome Triple A syndrome
161	Glutamate receptor, ionotropuc, N-methyl-D-aspartate, subunit 2B (GRIN2B) gene mutation	Intellectual developmental disorder, autosomal dominant 6
162	Glutaric aciduria, type 1 (GA1)	Glutaric acidemia type 1 (GA1) Glutaryl-coenzyme A dehydrogenase deficiency (GCDHD)
163	Glycogen storage disease III (GSD3)	Glycogen storage disease 3 Amylo-1,6-glucosidase deficiency Cori disease Debrancher deficiency
164	Glycogen storage disease IXa (GSD9A)	
165	Gordon hyperkalemia-hypertension syndrome	Pseudohypoaldosteronism type 2
166	Gorham syndrome	Gorham's disease Hemangiomata with osteolysis Phantom bone disease
167	Greig cephalopolysyndactyly syndrome	
168	Growth factor, ERV1-like (GFER) gene mutation	Congenital cataract, progressive muscular hypotonia, hearing loss, developmental delay syndrome

169	Hamartoma of hypothalamus	Tuber cinereum hamartoma
170	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
171	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
172	Hemihyperplasia with multiple lipomatosis (HHML) syndrome	
173	Hereditary hyperekplexia	Congenital stiff man syndrome Familial startle disease Kok disease
174	Hereditary Hypophosphatemic Rickets with Hypercalciuria (HHRH)	
175	Hereditary limphedema type 1	Milroy lymphedema Nonne-Milroy lymphedema
176	Hereditary pancreatitis	Protease, serine 1 (PRSS1) gene mutation
177	Hereditary pheochromocytoma and paraganglioma	
178	Hereditary sensory and autonomic neuropathy (HSAN)	
179	Hereditary sensory and autonomic neuropathy (HSAN) type 9	Tectonin beta-propeller repeat-containing protein 2 (TECPR2) gene mutation Autosomal recessive spastic paraplegia type 49
180	Hereditary spastic paraplegia (HSP)	Strumpell-Lorrain disease
181	Hermansky-Pudlak syndrome (HPS)	Albinism with hemorrhagic diathesis Alpha storage pool disease
182	Heterogeneous nuclear ribonucleoprotein D (HNRNPD) gene mutation	
183	HHH syndrome (Hyperornithinemia, Hyperammonemia, Homocitrullinuria)	Triple H syndrome Ornithine translocase ORNT1 deficiency Ornithine carrier deficiency
184	Histiocytosis-lymphadenopathy plus syndrome	H syndrome SLC29A3 spectrum disorder
185	Houge-Janssens syndrome 1 (HJS1)	
186	Houge-Janssens syndrome 2 (HJS2)	
187	Houge-Janssens syndrome 3 (HJS3)	
188	Human immunodeficiency virus (HIV) disease	איידס
189	Hyperekplexia epilepsy syndrome	ARHGEF9 RELATED DISORDER
190	Hyperphosphatemic familial tumoral calcinosis	
191	Hypertrophy (benign) of prostate with urinary obstruction and other lower urinary tract symptoms (LUTS)	

192	Hyperuricemia, pulmonary hypertension, renal failure, alkalosis syndrome	HUPRA syndrome
193	Hypotonia Hypoventilation Impaired Intellectual Development Dysautonomia Epilepsy and Eye	HIDEA syndrome
194	Hypotonia, Ataxia and Delayed Development syndrome (HADDS)	
195	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome (HADDTS)	CTBP1-related disorder
196	Ichthyosis, Congenital; Autosomal Recessive 1 (ARCI1)	COLLODION BABY, SELF-HEALING; SHCB ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 1, WITH BATHING SUIT DISTRIBUTION
197	IDDHBA syndrome	Intellectual developmental disorder with hypotonia and behavioral abnormalities
198	Idiopathic non-lupus full-house nephropathy (FHN)	
199	Immunodeficiency 14	Activated PI3K-delta syndrome
200	Immunodysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome	Autoimmune enteropathy type 1
201	Impaired intellectual development and distinctive facial features with or without cardiac defects (MRFACD)	Asadollahi-Rauch syndrome
202	Infantile Neuroaxonal Dystrophy (INAD1)	Seitelberger's disease
203	Intellectual developmenta disorder with severe speech and ambulation defects (IDDSSAD)	ACTL6B gene mutation
204	Intellectual developmental disorder, autosomal dominant 29 (MRD29)	SETBP1 haploinsufficiency disorder (SETBP1-HD)
205	Intellectual developmental disorder, autosomal dominant 45 (MRD45)	CIC gene mutation-related disorder
206	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)
207	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type	DDX3X related disorder
208	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
209	Intellectual disability, expressive aphasia, facial dysmorphism syndrome	

210	Intellectual disability, facial dysmorphism syndrome due to SETD5 haploinsufficiency	
211	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
212	Intractable diarrhea of infancy syndrome (IDIS)	Diarrhea 11, malabsorptive, congenital (DIAR11) Proline- and glutamate-rich protein with coiled-coil domain 1 (PERCC1) gene mutation
213	Intrauterine restriction, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomaly (IMAGe) syndrome	
214	Isovaleric acidemia	Isovaleryl-coenzyme A dehydrogenase deficiency Isovaleric aciduria
215	ITPR1 mutation related spinocerebellar ataxia 29	Spinocerebellar ataxia type 29 (SCA29) Congenital nonprogressive spinocerebellar ataxia
216	Jansen-de Vries syndrome (JDVS)	
217	Joubert syndrome	
218	Joubert syndrome with renal defect	NPHP1-related nephronophthisis
219	Juvenile Hypophosphatasia	Hypophosphatasia, childhood type
220	KBG syndrome	Short stature, facial and skeletal anomalies, intellectual disability, macrodontia syndrome
221	KIF1A associated neurological disorder (KAND)	
222	Kindler's syndrome	
223	KINSSHIP syndrome	
224	Kleefstra syndrome	due to del(9)(q34) due to monosomy 9q34
225	Kleefstra syndrome 2	
226	Klinefelter syndrome variant	49 XXXXY
227	Klippel Trenaunay syndrome	Klippel Trenaunay Weber syndrome
228	Kohlschutter syndrome	Amelocerebrohypohidrotic syndrome Epilepsy, mental deterioration and yellow teeth Epilepsy, dementia, amelogenesis, imperfecta syndrome Kohlschutter-Tonz syndrome
229	Kostmann syndrome	Infantile agranulocytosis
230	Kozlowski spondylometaphyseal dysplasia	TRPV4 related skeletal dysplasia
231	KREMEN1 related ectodermal dysplasia 13	Ectodermal dysplasia 13, hair/tooth type (ECTD13)
232	Lamb Shaffer syndrome	SOX5 haploinsufficiency syndrome
233	Lamellar Ichthyosis	Collodion baby
234	Lateral meningocele syndrome (LMNS)	NOTCH3-related lateral meningocele syndrome Lehman syndrome
235	Legius syndrome	Neurofibromatosis type 1-like syndrome (NFLS)
236	Leukocyte adhesion deficiency (LAD) type 2	
237	Liang-Wang syndrome (LIWAS)	
238	Limb-girdle Muscular Dystrophy, Type 28, Autosomal Recessive	LGMDR28

239	Lipid transport defect of intestine	Chylomicron retention disease Anderson syndrome
240	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
241	Loeys-Dietz syndrome	
242	Long chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency (LCHADD)	
243	Lowe syndrome	Cerebro-oculorenal dystrophy Oculocerebrorenal syndrome Lowe-Bickel syndrome Lowe-Terrey-MacLachlan syndrome Phosphatidylinositol-4,5-bisphosphate-5-phosphatase deficiency
244	Lysine demethylase 5c (KDM5C) gene mutation	
245	Lysinuric protein intolerance (LPI)	Congenital lysinuria
246	Malan overgrowth syndrome	Sotos syndrome 2
247	Mandibulofacial dysostosis with microcephaly	Mandibulofacial dysostosis, Guion-Almeida type Elongation factor Tu GTP-binding comain-containing 2 (EFTUD2) gene mutation
248	Maple syrup urine disease (MSUD)	Branched chain 2-ketoacid dehydrogenase deficiency (BCKD) Ketoacidemia
249	Maternally inherited Leigh syndrome (MILS)	Maternally inherited infantile subacute necrotizing encephalopathy
250	MBD5-associated neurodevelopmental disorder (MAND)	Intellectual developmental disorder, autosomal dominant 1
251	McCune Albright syndrome	
252	Megalencephaly capillary malformation (MCAP)	Macrocephaly-capillary malformation (M-CM, MCM) Megalencephaly-cutis marmorata telangiectatica congenita syndrome (MCMTC)
253	Meier-Gorlin syndrome	Ear, patella, short stature syndrome Microtia, absent patellae, micrognathia syndrome
254	MEIS2 syndrome	Cleft palate, cardiac defects, and impaired intellectual development Cardiac malformation, cleft lip/palate, microcephaly and digital anomalies
255	Metaphyseal anadysplasia type 2	Matrix metalloproteinase 9 (MMP9) gene mutation
256	Methyl-CpG-binding protein 2 (MECP2) gene mutation	
257	Methylmalonic acidemia	Methylmalonic aciduria (MMA)
258	Microcephalic primordial dwarfism Alazami type	Alazmai syndrome
259	Microcephaly, corpus callosum hypoplasia, intellectual disability, facial dysmorphism syndrome	
260	Minor partial trisomy	Minor partial chromosome duplication

261	Mitochondrial enoyl coA reductase protein-associated neurodegeneration (MEPAN) syndrome	Mitochondrial enoyl CoA hydratase 1 deficiency (ECHS1D) Mitochondrial enoyl coA reductase (MECP) gene mutation
262	Mixed gonadal dysgenesis	Mosaicism 45, X; 46, XY
263	Moebius syndrome	Congenital facial diplegia
264	Mosaic trisomy 16 syndrome	
265	Moyamoya disease	
266	MPPH syndrome	Megalencephaly, polymicrogyria, postaxial polydactyly, hydrocephalus syndrome
267	MULLEGAMA-KLEIN-MARTINEZ SYNDROME STAG2 RELATED DISORDER (MKMS)	
268	Multicentric carpotarsal osteolysis syndrome	
269	Multiple congenital exostosis	Multiple osteochondromatosis syndrome
270	Multiple endocrine neoplasia, type 3 (MEN3)	Multiple endocrine neoplasia, type 2B (MEN2B)
271	Multiple sulfatase deficiency (MSD)	Juvenile sulfatidosis, Austin type Mucosulphatidosis
272	Multisystemic smooth muscle dysfunction syndrome	Actin, alpha-2, smooth muscle, aorta (ACTA2) gene mutation
273	Muscle eye brain disease	
274	Muscle phosphoglycerate mutase deficiency	Phosphoglucomutase deficiency
275	Myhre syndrome	
276	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)
277	Myofibrillar myopathy	
278	Myosin, cardiac, heavy chain, beta (MYHCB, MYH7) gene mutation	
279	Nabais Sa-de Vries syndrome, type 2 (NSDVS2)	NEURODEVELOPMENTAL DISORDER WITH RELATIVE MACROCEPHALY AND WITH OR WITHOUT CARDIAC OR ENDOCRINE ANOMALIES, NEDMACE
280	NARS2 related disorder	Deafness. Autosomal recessive 94 (DFNB94)
281	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
282	Neurodegeneration due to 3-hydroxyisobutyryl coenzyme A hydroxylase (HIBCH) deficiency	Methacrylic aciduria Valine metabolic defect
283	Neurodegeneration, childhood-onset, with brain atrophy (CONDBA)	UBTF related motor and cognitive regression syndrome
284	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies (NEDDFSA)	Zinc finger miz-domain containig 1(ZMIZ1) gene mutation
285	Neurodevelopmental disorder with eye movement abnormalities and ataxia (NEDEMA)	FRMD5-related disorder

286	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures (NEDFSS)	TRPM3-related disorder
287	Neuroendocrine cell hyperplasia of infancy	NEHI syndrome
288	Nicolaides-Baraitser syndrome	SWI/SNF-related, matrix-associated, actin-dependant regulator of chromatin, subfamily A, member 2 (SMARCA2) gene mutation
289	Niemann-Pick disease, type C	Supraoptic vertical ophthalmoplegia
290	NOG related disorder	Teunissen-Cremers syndrome Stapes ankylosis with broad thumbs and toes Ankylosis of stapes, hyperopia, broad thumbs, broad first toes and syndactyly
291	NPHS2 related nephrotic syndrome	Familial idiopathic steroid-resistant nephrotic syndrome Genetic SRNS Hereditary steroid-resistant nephrotic syndrome
292	Oculo-facio-cardio-dental syndrome	Oculofaciocardiodental syndrome
293	O'Donnell-Luria-Rodan syndrome (ODLURO)	Lysine-specific methyltransferase 2E (KMT2E) gene mutation
294	Ogden syndrome	
295	Ohdo sydrome, Say-Barber-Biesecker-Young-Simpson (SBBYS) variant	Blepharophimosis, intellectual disability syndrome Say-Barber-Biesecker-Young-Simpson (SBBYS) variant KAT6B-related disorder
296	Okur-Chung neurodevelopmental syndrome (OCNDS)	OKUR CHUNG SYNDROME -CSNK2A1 MUTATION
297	Pallister Killian syndrome	Tetrasomy 12p syndrome Killian-Teschler-Nicola syndrome Pallister mosaic syndrome
298	Paraneoplastic opsoclonus-myoclonus-ataxia syndrome; POMA	Dancing eye syndrome Dancing eye-dancing feet syndrome Opsoclonus-myoclonus-ataxia syndrome; OMA Kinsbourne syndrome
299	Parkes Weber syndrome	
300	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy (PNKD3)	Generalized epilepsy and paroxysmal dyskinesia (GEPD)
301	Pearson's syndrome	
302	Pediatric multiple sclerosis	
303	Pelizaeus-Merzbacher disease (PMD)	Sudanophilic leukodystrophy
304	Penta X syndrome	XXXXX syndrome
305	Perching syndrome	Kelch like family member 7-related Bohring-Opitz-like and Crisponi/cold-induced sweating-like overlap syndrome
306	Permanent neonatal diabetes mellitus	Developmental delay, epilepsy and neonatal diabetes 1 (DEND)
307	Peutz Jeghers syndrome PJS	Peutz-Jeghers polyposis Peutz-Jeghers lentiginosis syndrome
308	PHACE syndrome	Phace syndrome and Moya Moya Disease Pascual-Castroviejo syndrome type 2

309	PHACES syndrome	
310	PHD finger protein 21A (PHF21A) gene mutation	
311	Phelan-McDermid syndrome	22q13.3 deletion syndrome SH3 and multiple ankyrin repeat domains 3 (SHANK3) gene mutation
312	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
313	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
314	Phosphatase and tensin homolog (PTEN) gene mutation	
315	Phosphoenolpyruvate carboxykinase (PEPCK-C) deficiency	
316	Pitt-Hopkins-like syndrome 1 (PTHSL1)	
317	Pituitary stalk interruption syndrome	Ectopis neurohypophysis
318	Pleuropulmonary blastoma (PPB)	
319	Pleuropulmonary blastoma familial tumor and dysplasia syndrome (PPBFTDS)	DICER1 tumor-predisposition syndrome PPB familial tumor and dysplasia syndrome
320	Poland anomaly	Poland syndrome Unilateral defect of pectoralis muscle and syndactyly of hand
321	POMT1-related limb-girdle muscular dystrophy R11	Limb-girdle muscular dystrophy type 2K; LGMD type 2K POMT1-related LGMD R11
322	Pontocerebellar hypoplasia type 9 (PCH9)	
323	Popliteal pterygium syndrome	
324	Potocki Lupski syndrome	17p11.2 microduplication
325	Potocki-Shaffer syndrome	Chromosome 11p11.2 deletion syndrome Proximal 11p deletion syndrome (P11pDS)
326	Primary hyperoxaluria type 1	Glycolic aciduria
327	Primary microcephaly, mild intellectual disability, young-onset diabetes syndrome	
328	Primrose syndrome	Intellectual disability, cataract, calcified pinna, myopathy syndrome ZBTB20-related syndrome

329	PRKAG2-related hypertrophic cardiomyopathy	Familial hypertrophic cardiomyopathy 6
330	Progeria syndrome	Hutchinson-Gilford syndrome
331	Propionic Acidemia	Ketotic hyperglycinaemia Hyperglycinaemia with ketosis and leucopenia Propionyl-CoA carboxylase (PCC) deficiency
332	Proteolipid protein 1(PLP1) gene mutation	
333	Pseudohypoaldosteronism type 1, recessive form	Pseudohypoaldosteronism type 1B Pseudohypoaldosteronism, Persian-Jewish type
334	Pseudohypoparathyroidism	Guanine nucleotide-binding protein, alpha-stimulating activity polypeptide 1 (GNAS) gene mutation Parathyroid hormone resistant hypoparathyroidism
335	Pseudohypoparathyroidism type 1A	Albright hereditary osteodystrophy (AHO)
336	Pseudohypoparathyroidism type 2	
337	Pseudotumor cerebri	Benign intracranial hypertension; BIH Nonne's syndrome Noninfective serous meningitis
338	Pulmonary alveolar proteinosis (PAP)	
339	Purine rich element binding protein A (PURA) syndrome	
340	Pyknodysostosis	Maroteaux-Lamy syndrome type II Cathepsin k (CTSK) gene mutation
341	Radial aplasia-trhombocytopenia (TAR) syndrome	
342	Radioulnar synostosis with amegakayocytic thrombocytopenia (RUSAT) 2	MDS1 and EVI1 complex (MECOM) gene mutation
343	Rahman syndrome	H1-4-related neurodevelopmental disorder HIST1H1E-related disorder
344	RAS-associated autoimmune leukoproliferative disease (RALD)	Autoimmune leukoproliferative syndrome type 4
345	Rasmussen subacute encephalitis	Rasmussen syndrome Rasmussen encephalitis
346	Rauch-Steindl syndrome	
347	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
348	Renal nutcracker syndrome	Left renal vein entrapment syndrome Mesoaortic compression of left renal vein
349	Renpenning syndrome	Golabi-Ito-Hall syndrome Hamel cerebropalatocardiac syndrome Polyglutamine-binding protein 1 (PQBP1) gene mutation
350	Resistance to insulin-like growth factor 1 (IGF1)	Insulin-like growth factor 1 receptor (IGF1R) gene mutation
351	Rett syndrome; RTS	Rett's disorder Cerebroatrophic hyperammonemia

352	ROHHAD syndrome	Rapid-onset childhood obesity, hypothalamic dysfunction, hypoventilation, autonomic dysregulation syndrome
353	Rubinstein-Taybi syndrome	
354	Russell-Silver syndrome	Silver syndrome Silver-Russell dwarfism
355	Schaaf Yang syndrome	MAGE family member L2-related Prader-Willi-like syndrome MAGEL2 related Prader Willi-like syndrome
356	Schinzel-Giedion syndrome	
357	Schuurs-Hoeijmakers syndrome	
358	Scimitar syndrome	Pulmonary venolobar syndrome
359	Seizures, benign familial infantile, 2	PRRT2-related benign familial seizures
360	Sensenbrenner–Dorst–Owens syndrome	Sensenbrenner's syndrome Levin syndrome Cranioectodermal dysplasia (WDR35 mutation)
361	Sepiapterin reductase deficiency	
362	Set domain-containing protein 5 (SETD5) gene mutation	
363	Severe congenital nemaline myopathy (TNNT1 mutation)	Nemaline myopathy, Amish type (ANM) Nemaline myopathy 5A, autosomal recessive, severe infantile (NEM5A)
364	Severe infantile form of carnitine palmitoyltransferase II deficiency	
365	SH3 and multiple ankyrin repeat domains 2 (SHANK2) gene mutation	
366	Sharp's syndrome	Mixed collagen vascular disease Mixed connective tissue disease (MCTD)
367	Short QT syndrome 2	KCNQ1-related Short QT syndrome
368	SHORT syndrome	
369	Shprintzen-Goldberg syndrome (SGS)	Marfanoid craniosynostosis syndrome Shprintzen Goldberg craniosynostosis syndrome
370	Shukla-Vernon syndrome (SHUVER)	
371	Sialuria	
372	Sifrim-Hitz-Weiss syndrome	CHD4-related neurodevelopmental disorder
373	Signal transducer and activator of trascription 3 (STAT3) gene mutation	
374	Sjogren-Larsson syndrome	Fatty alcohol-nicotinamide adenine dinucleotide oxidoreductase deficiency
375	Small G protein signaling modulator 3 (SGSM3) mutation	
376	SMC3-related Cornelia de Lange syndrome (CdLS)	Cornelia de Lange syndrome 3
377	Smith Kingsmore syndrome	Macrocephaly, intellectual disability, neurodevelopmental disorder, small thorax (MINDS) syndrome
378	Smith-Lemli-Opitz syndrome	7-dehydrocholesterol reductase deficiency
379	Snijders Blok-Campeau syndrome	CHD3-related developmental delay, speech delay, intellectual disability, abnormalities of vision, facial dysmorphism syndrome
380	Sodium voltage-gated channel, alpha subunit 4 (SCN4A) gene mutation	

381	Solute carrier family 9, member 7 (SLC9A7) gene mutation	
382	Sotos' syndrome	Cerebral gigantism Nuclear receptor-binding set domain protein 1 (NSD1) gene mutation
383	SOX2 anophthalmia syndrome	Syndromic microphthalmia 3 Anophthalmia-esophageal-genital (AEG) syndrome
384	Special AT-rich sequence-bindind protein 2 (SATB2) associated syndrome	Glass syndrome Chromosome 2q32-q33 deletion syndrome Del(2)(q32q33) Monosomy 2q32q33
385	Spinal muscular atrophy, lower extremity-predominant 1, AD	DYNC1H1-related spinal muscular atrophy
386	Spinocerebellar ataxia type 21	SCY1-lime (SCYL1) gene mutation
387	Spinocerebellar ataxia type 26 (SCA26)	EEF2 related neurodevelopmental disorder
388	Spondilometaphyseal dysplasia	
389	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type (SEMDIST)	Spondyloepimetaphyseal dysplasia with severe short stature
390	Spondyloepiphyseal dysplasia (SED)	Spondyloepiphyseal dysplasia congenita (SEDC)
391	Spondylometaphyseal dysplasia with combined immunodeficiency; SPENCDI	Roifman-Melamed syndrome Spondyloenchondrodysplasia with immune dysregulation
392	SPTBN1 related developmental delay, impaired speech and behavioral abnormalities (DDISBA)	
393	Stankiewicz-Isidor syndrome (STISS)	PSMD12-related disorder
394	Streptococcus infection in conditions classified elsewhere and of unspecified site, Streptococcus, unspecified	
395	Synaptic RAS-GTPase-activating protein 1 (SYNGAP1) gene mutation	
396	Systematized epidermal nevus	Nevus unius lateris
397	Systemic mast cell disease (SMCD)	Systemic mastocytosis
398	SZT2 related developmental and epileptic encephalopathy 18	DEE18
399	SZT2 subunit of KICSTOR complex gene mutation	
400	Tatton Brown Rahman syndrome	DNA methyltranspherase 3A (DNMT3A) gene mutation Tall stature, intellectual disability, facial dysmorphism syndrome
401	Temple syndrome	
402	TET3-related Beck-Fahrner syndrome; TET3-BEFAHRS	
403	THUMPD1 related disorder	Neurodevelopmental disorder with speech delay and variable ocular anomalies (NEDSOA)
404	Timothy syndrome	Long QT syndrome type 8; LQT8 Long QT syndrome-syndactyly syndrome
405	Tooth agenesis-colorectal cancer syndrome (ODCRCS)	AXIN2-related disorder

406	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)
407	Transketolase (TKT) deficiency	Short stature-developmental delay-congenital heart defect syndrome
408	Trichohepatoenteric syndrome	
409	Trichorhinophalangeal syndrome	
410	tRNA methyltransferase 10A (TRMT10A) gene mutation	
411	Turner's phenotype - ring chromosome karyotype	
412	Tyrosinemia type 2	Hypertyrosinemia, Richner-Hanhart type Hypertyrosinemia, Oregon type
413	Tyrosinemia type 3	
414	Unverricht-Lundborg syndrome	Progressive myoclonic epilepsy type 1 Baltic myoclonus epilepsy
415	Usher syndrome type 3; USH3	Retinitis pigmentosa-deafness syndrome type 3
416	Ventriculomegaly with cystic kidney disease (VMCKD)	Heterozygous CRB2-related disorder
417	Verheij syndrome	8q24.3 microdeletion syndrome Del(8)(q24.3) Monosomy 8q24.3
418	Verloes Bourguignon syndrome	Autosomal recessive brachyolmia and amelogenesis imperfecta syndrome Dental anomalies and short stature (DAAS)
419	Ververi Brady syndrome	Glutamine rich 1-related intellectual disability, chondrodysplasia syndrome
420	Vici syndrome	Dionisi Vici Sabetta Gambarara syndrome
421	VPS11-related autosomal recessive hypomyelinating leukoencephalopathy	Leukodystrophy, hypomyelinating, 12 (HLD12) VPS11 core subunit of CORVET and HOPS complexes-related autosomal recessive hypomyelinating leukodystrophy
422	Waardenburg Shah syndrome	Waardenburg Hirschsprung syndrome
423	Weaver syndrome (WVS)	
424	Weill-Marchesani syndrome	Brachydactyly-spherophakia syndrome
425	Weiss-Kruszka syndrome (WSKA)	Zinc finger protein 462 (ZNF462) gene mutation
426	Wieacker Wolff syndrome	Intellectual disability, developmental delay,
427	Wiedemann Steiner syndrome	Hypertrichosis, short stature, facial dysmorphism, developmental delay syndrome
428	Wilms tumor, aniridia, genitourinary anomalies and mental retardation	Chromosome 11p13 deletion syndrome
429	Wiscott-Aldrich syndrome	Aldrich syndrome
430	Witteveen Kolk syndrome (WITKOS)	SIN3A-related intellectual disability syndrome
431	Wolfram syndrome	Marquardt-Loriaux syndrome DIDMOAD (diabetes insipidus, diabetes mellitus, optic atrophy, deafness) syndrome

432	Wolman disease	Primary familial xanthomatosis with adrenal calcification Familial visceral xanthomatosis Deficiency of cholesterol esterase AND triacylglycerol lipase Wolman xanthomatosis Acid esterase deficiency
433	Xia-Gibbs syndrome	
434	X-linked intellectual disability Cabezas type	Cullin 4b (CUL4B) gene mutation
435	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
436	X-linked intellectual disability Siderius type	
437	X-linked intellectual disability with cerebellar hypoplasia syndrome	Oligophrenin-1 syndrome (OPHN1)
438	X-linked intellectual disability, hypotonia, movement disorder syndrome	
439	X-linked non progressive cerebellar ataxia	
440	X-linked reticulate pigmentary disorder with systemic manifestation syndrome	Partington disease
441	Zhu Tokita Takenouchi Kim (ZTTK) syndrome	
442	Zinc Finger Homeobox 3 (ZFHX3)	
443	Zinc finger protein 292 (ZNF292) gene mutation	Intelectual developmental disorder, autosomal dominant 64
444	Zinc finger protein 335 (ZNF335) gene mutation	Microcephaly 10, primary, autosomal recessive