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 שהוכרו כנדירות ואינן מופיעות ברשימה, וכן מחלות שמופיעות ברשימה אך אינן מזכות בקצבה, כיוון שהתגלה כי שכיחותן גבוהה מהסף שהוגדר. בנספח למסמך זה מופיעה רשימה של אבחנות אשר הוסרו מן הרשימה, כיוון שהתגלה כי שכיחותן עלתה.
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	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	9q22.3 deletion syndrome	Monosomy 9q22.3 syndrome Microdeletion 9q22.3
7	Abetalipoproteinemia	
8	Acetyl-coenzyme A acyltransferase	Acetyl-CoA acyltransferase 3-ketoacyl-CoA thiolase Acetyl-coenzyme A acyltransferase Beta-ketothiolase
9	Acquired epileptic aphasia	Landau-Kleffner syndrome
10	Acrocephalosyndactyly type 5	Pfeiffer syndrome
11	Acrodysostosis	
12	ACTB Dystonia-deafness syndrome 1	Baraitser-Winter syndrome 1 Becker nevus, syndromic or isolated, somatic mosaic Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic Thrombocytopenia 8, with dysmorphic features and developmental delay
13	Acute myeloid leukemia with RUNX1::RUNX1T1 fusion (morphologic abnormality)	Acute myeloid leukemia, t(8;21) (q22;q22.1)
14	Adams-Oliver syndrome	

15	Adenosine deaminase 2 (ADA2) gene mutation	Deficiency of adenosine deaminase 2 (DADA2)
16	Adenylosuccinase deficiency ADSL	
17	AFF2	
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	AKT2 serine threonine kinase 2 related familial partial lipodystrophy	
23	Al Kaissi syndrome (ALKAS)	Cyclin-dependent kinase 10 (CDK10) gene mutation
24	Alacrima, achalasia, and impaired intellectual development syndrome (GMPPA)	GDP-MANNOSE PYROPHOSPHORYLASE A
25	Alexander's disease	Alexander syndrome
26	Allan-Herndon-Dudley syndrome	Monocarboxylate transporter 8 deficiency
27	ALMS1	Alstrom syndrome
28	Alternating hemiplegia of childhood	
29	Ameloblastome	
30	Ankyrin 3 related intellectual disability, sleep disturbance syndrome (ANK-3)	
31	Arboleda Tham syndrome (ARTHS)	KAT6A gene mutation
32	ARI1 Smith-Magenis syndrome	
33	Aromatic amino acid decarboxylase deficiency (AADC)	
34	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
35	Au Kline syndrome	Okamoto syndrome
36	Autoimmune neutropenia	
37	Autosomal dominant central core disease	Ryanodine receptor 1 (RYR1) gene receptor Periodic paralysis
38	Autosomal dominant HYPER-IgE syndrome due to STAT3 deficiency (HIES)	HYPER-IgE syndrome 1, AD Job syndrome
39	Autosomal recessive agammaglobulinemia PIK3R1 p85 deficiency	
40	Autosomal recessive cerebellar ataxia with oculomotor apraxia type 1 (AOA1)	
41	Autosomal recessive cerebellar ataxia, epilepsy, intellectual disability syndrome - WWOX deficiency	Autosomal recessive spinocerebellar ataxia type 12 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12; SCAR12

42	Autosomal recessive spastic paraplegia type 15	Hereditary spastic paraparesis Kjellin syndrome
43	Axenfeld-Rieger syndrome	
44	Aymé-Gripp syndrome (AYGRP)	
45	Bainbridge Ropers syndrome	ASXL transcriptional regulator 3 deficiency syndrome
46	Baker Gordon syndrome	Synaptotagmin (SYT) 1-related neurodevelopmental disorder
47	Baraitser Winter cerebrofrontofacial syndrome	
48	Barakat syndrome	Hypoparathyroidism, sensorineural hearing loss, renal disease syndrome Hypoparathyroidism, deafness, renal disease (HDR) syndrome
49	Bardet-Biedl syndrome (BBS)	Laurence-Moon-Biedl syndrome Laurence-Moon-Bardet-Biedl syndrome (LMBB)
50	Barth syndrome	3-Methylglutaconic aciduria type 2
51	Bartter syndrome type 1	Hyperprostaglandin E syndrome type 1
52	Bartter syndrome type 2	Hyperprostaglandin E syndrome type 2 Hypokalemic alkalosis with hypercalciuria antenatal type 2
53	Bartter syndrome type 3	Bartter syndrome CLCNKB related disorder
54	BCL11A Dias Logan syndrome	
55	BCL2L11-related disorder	2q13 deleteion syndrome
56	Beals syndrome	Congenital contractural arachnodactyly (CCA) Beals Hecht syndrome
57	Bernard Soulier syndrome	Hemorrhagiparous thrombocytic dystrophy Giant platelet syndrome
58	Blackfan-Diamond anemia	Aase syndrome Congenital pure red cell aplasia
59	BLOOM syndrome	
60	Bohring-Opitz syndrome (BOS)	C-like syndrome
61	Borjeson-Forssman-Lehmann syndrome	
62	Brachydactyly mental retardation syndrome	Chromosome 2q37 deletion syndrome Albright hereditary osteodystrophy-like syndrome
63	BRAF craniofacial syndrome	Cardiofaciocutaneous syndrome
64	Brain lung thyroid syndrome	Choreoathetosis with congenital hypothyroidism and neonatal respiratoy distress syndrome
65	Brain malformation with or without urinary tract defects	Nuclear factor I/A (NFIA) gene mutation
66	BRD4 related syndrome	Cornelia de Lange syndrome 6 (CDLS6)
67	Bromodomain and WD repeat-containing protein 3 (BRWD3)	
68	Bruton's agammaglobulinemia	X-linked agammaglobulinemia (XLA)
69	Camptodactyly, arthropathy, coxa-vara, pericarditis syndrome (CACP)	Jacobs syndrome
70	CAPOS syndrome	Cerebellar ataxia, areflexia, pes cavus, optic atrophy, sensorineural hearing loss syndrome ATP1A3
71	Cardiofaciocutaneous syndrome 1 (CFC1)	Noonan syndrome 7

72	Cardiomyopathy, dilated, 1GG (SDHA)	Mitochondrial complex II deficiency, nuclear type 1 (SDH1) Neurodegeneration with ataxia and late-onset optic atrophy Pheochromocytoma/paraganglioma syndrome 5
73	Caudal duplication syndrome	Caudal duplication anomaly
74	CCR4-not transcription complex, subunit 3 (CNOT3) mutation	
75	Central core disease	Central core myopathy CONGENITAL MYOPATHY 1A, AUTOSOMAL DOMINANT, WITH SUSCEPTIBILITY TO MALIGNANT HYPERTHERMIA; CMYP1A
76	Cerebellar atrophy, developmental delay, and seizures (CADEDS)	
77	Cerebellar dysfunction with variable cognitive and behavioral abnormalities (CECBA)	CAMTA1 ASSOCIATED DISORDER Non-progressive cerebellar ataxia with intellectual disability
78	Cerebral folate transport deficiency	Cerebral folate deficiency
79	Cerebro-oculo-dento-auriculo-skeletal (CODAS) syndrome	
80	CHAMP1 related disease	NEURODEVELOPMENTAL DISORDER WITH HYPOTONIA, IMPAIRED LANGUAGE, AND DYSMORPHIC FEATURES; NEDHILD
81	Chanarin-Dorfman disease	Triglyceride storage disease with ichthyosis Chanarin-Miranda syndrome
82	Charcot-Marie-Tooth disease type 2N	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, AUTOSOMAL DOMINANT, TYPE 2A1; CMT2A1 HEREDITARY MOTOR AND SENSORY NEUROPATHY IIA1; HMSN IIA1
83	Charcot-Marie-Tooth disease, axonal, type 2O	DYNC1H1-related Charcot-Marie-Tooth
84	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
85	Cholestasis, progressive familial intrahepatic 3 (PFIC3)	MDR3 DEFICIENCY ABCB4-related disorder
86	CHOPS syndrome	
87	Chromosome 6q11-q14 deletion syndrome	include 6q13-14
88	Chronic bullous dermatosis of childhood	Linear IgA bullous disease in children Childhood linear IgA disease
89	Chronic granulomatous disease type II (CGD2)	
90	Chronic recurrent multifocal osteomyelitis (CRMO)	Majeed syndrome Synovitis acne pustulosis hyperostosis osteitis syndrome (SAPHO)
91	Ciliary dyskinesia, primary, 3 (CILD3)	DNAH5-related disorder
92	Citrullinemia type 2	Adult onset citrin deficiency
93	Clark Baraitser syndrome	

94	Clathrin (CLTC) gene mutation	Intelectual developmental disorder, autosomal dominant 56
95	Cleidocranial dysplasia (CCD)	Cleidocranial dysostosis (CLCD)
96	CLOVES syndrome	Congenital lipomatous overgrowth, vascular malformation, epidermal nevi, skeletal anomaly syndrome
97	CNOT 1 MUTATION RELATED VISSERS BODMER SYNDROME (VIBOS)	
98	CNOT3 related disorder	
99	Cockayne syndrome	
100	Coffin-Siris syndrome	Fifth digit syndrome
101	COL6A2 Betlehem Myopathy	Intermediate collagen VI-related muscular dystrophy
102	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
103	Combined immunodeficiency CARMIL2	
104	Combined immunodeficiency due to DOCK8 deficiency	
105	Combined oxidative phosphorilation deficiency 37 (COXPD37)	MICOS13-related disorder
106	Congenital central hypoventilation syndrome (CCHS)	Ondine curse Congenital pulmonary hypoventilation
107	Congenital chronic diarrhea with protein-losing enteropathy	Congenital chronic diarrhea with exudative enteropathy
108	Congenital dyserythropoietic anemia (CDA) type I	
109	Congenital ichthyosiform erythroderma (CIE)	Congenital recessive ichthyosis
110	Congenital ichthyosis of skin	Ichthyosis congenita Fish scale disease Fish skin
111	Congenital ichtyosis with hypotrichosis syndrome	Autosomal recessive congenital ichthyosis
112	Congenital insensitivity to pain with anhidrosis (CIPA)	Hereditary sensory and autonomic neuropathy (HSAN) type 4 Swanson-Buchanan-Alvord neuropathy syndrome
113	Congenital malformation of dural sinus	
114	Congenital muscular dystrophy due to lamin A/C mutation LMNA	
115	Congenital Muscular Dystrophy Merosin-deficient (LAMA2 mutation)	
116	Congenital myasthenic syndrome	Congenital myasthenia syndrome
117	Congenital myasthenic syndrome type 9 (CMS9)	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency MUSK-related disorder

118	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
119	Congenital myopathy with myasthenic-like onset (RYR1)	
120	Congenital pseudoarthrosis of the tibia	
121	Congenital secretory diarrhea, chloride type	Congenital chloride diarrhea
122	Convulsions, familial infantile, with paroxysmal choreoathetosis	PRRT2-related familial convulsions
123	COPB2 gene mutation-related disorder	Autosomal recessive primary microcephaly-19 (MCPH19)
124	Cortical dysplasia, complex, with other brain malformation 7 (CDCBM7)	TUBB2B-related disorder
125	Cortical dysplasia, complex, with other brain malformations 13	DYNC1H1-related cortical dysplasia
126	Craniofrontonasal dysplasia	Craniofrontonasal syndrome Craniofrontonasal dysostosis
127	Craniometaphyseal dysplasia	
128	Currarino triad	Currarino syndrome
129	Cutis laxa type 1	Urban Rifkin Davis syndrome ARCL1C
130	Cutis marmorata telangiectatica congenita (CMTC)	Congenital livedo reticularis Van Lohuizen's syndrome
131	DASS Dental anomalies and short stature LTBP3	
132	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
133	Deficiency of dihydrofolate reductase	Dihydrofolate reductase deficiency
134	Deficiency of prolidase	
135	Deletion of part of autosome	
136	Dent disease type 1	Nephrolithiasis, hypercalciuric, X-linked (NPHL2)
137	Dent's disease (DENT)	
138	Desmin myopathy	Desmin-related myofibrillar myopathy Desminopathy
139	Developmental and epileptic encephalopathy (GABRB3)	Epilepsy, childhood absence, susceptibility to, 5
140	Developmental and epileptic encephalopathy 64 (DEE64)	Epileptic encephalopathy, early infantile, 64 (EIEE64) RHOBTB2-related disorder
141	Developmental and epileptic encephalopathy ATP6V0A1	
142	Developmental delay with or without dysmorphic facies and autism (DEDDFA)	Transformation/Transcription Domain-associated protein (TRRAP) associated disorder
143	Developmental delay, dysmorphic facies, and brain anomalies (U2AF2)	

144	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
145	Diaphyseal dysplasia	Camurati Engelmann syndrome
146	Diarrhea 7, protein-losing enteropathy type DGAT1	
147	Diets Jongmans syndrome	KDM3B-related intellectual disability, facial dysmorphism, short stature syndrome
148	Diffuse intrinsic pontine glioma (DIPG)	
149	DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY (DLDD)	E3 DEFICIENCY LIPOAMIDE DEHYDROGENASE DEFICIENCY, LACTIC ACIDOSIS DUE TO MAPLE SYRUP URINE DISEASE, TYPE III (MSUD3)
150	Distal anoctaminopathy	Miyoshi muscular dystrophy type 3 (MMD3) ANO5-related disorder
151	Distal trisomy 10q	Distal duplication 10q 10q trisomy
152	Distal Xq28 microduplication syndrome	Distal trisomy Xq28
153	DMD - Duchenne muscular dystrophy	
154	DYRK1A-related intellectual disability syndrome	DYRK1A (dual specificity tyrosine phosphorylation regulated kinase 1A)
155	Dyskeratosis congenita (DKC)	Cole-Engmann-Zinsser syndrome
156	Dystonia 11, myoclonic (DYT11)	
157	Early infantile epileptic encephalopathy with suppression bursts	Ohtahara syndrome
158	Ectopia lentis syndrome	Familial ectopia lentis, Isolated ectopia lentis (IEL) ADAMTSL4 related lens ectopia
159	Elane Neutropenia cyclic	
160	Emanuel syndrome	Supernumerary der(22)t(11;22) syndrome Der(22) syndrome due to 3:1 meiotic disjunction events Supernumerary derivative 22 chromosome syndrome
161	Encephalopathy due to defective mitochondrial and peroxisomal fission 2 (EMPF2)	DNM1L gene mutation 2
162	Encephalopathy, lethal, due to defective mitochondrial peroxisomal fission 1 (EMPF1)	DNM1L gene mutation 1
163	Enchondromatosis	Ollier disease
164	Epidermolysis bullosa acquisita	Acquired epidermolysis bullosa
165	Epidermolysis bullosa simplex 2A, generalized severe	Epidermolysis bullosa simplex 2A, Dowling-Meara type KRT5 related epidermolysis bullosa simplex, severe
166	Epilepsy, idiopathic generalized (EIG16)	

167	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)
168	Episodic kinesigenic dyskinesia 1	PRRT2-related kinesigenic dyskinesia
169	Erythema annulare	Figurate erythema
170	Erythropoietic protoporphyria (EPP)	Magnus syndrome Heme synthase deficiency
171	Evans syndrome	
172	Factor X deficiency	Factor 10 deficiency Stuart-Prower factor deficiency
173	Familial amyloid nephropathy with urticaria and deafness	Muckle-Wells syndrome
174	Familial Hemophagocytic Lymphohistiocytosis (FHL)	Familial haemophagocytic reticulosis Familial HLH
175	Familial hypercholanemia	
176	Familial infantile bilateral striatal necrosis (IBSN)	
177	Familial lipoprotein lipase deficiency	Fredrickson type 1 hyperlipoproteinemia
178	Familial restrictive cardiomyopathy	
179	Familial Short QT syndrome	
180	Fanconi-Bickel syndrome	Glycogenosis with glucoaminophosphaturia Hepatic glycogenosis with de Toni-Debré-Fanconi syndrome Pseudo-phlorizin diabetes Renal glucose-losing syndrome
181	FASTKD2 MUTATION RELATED CARDIOMYOPATHY AND MITOCHONDRIAL DISORDER	FAST kinase domains 2-related infantile mitochondrial encephalomyopathy
182	FBN1 Weill-Marchesani syndrome 2	
183	FBP1 Fructose-1,6-bisphosphatase deficiency	
184	FBXO11 related disorder	Intellectual developmental disorder with dysmorphic facies and behavioral abnormalities (IDDFBA)
185	Febrile infection related epilepsy syndrome (FIRES)	Fever-induced refractory epileptic encephalopathy in school-aged children (FIRES)
186	Feingold syndrome	Microcephaly-oculo-digito-esophageal-duodenal syndrome MYCN-related disorder
187	Fibrodysplasia Ossificans Progressiva (FOP)	Progressive myositis ossificans Munchmeyer disease
188	FKBP14 Ehlers Danlos syndrome kyphoscoliotic type 2	
189	Floating-Harbor syndrome (FLHS)	SCRAP mutation-related disorder
190	Focal dermal hypoplasia (FDH)	Goltz-Gorlin syndrome
191	Forkhead box P1 (FOXP1) gene mutation	Intellectual disability, severe speech delay, mild dysmorphism syndrome
192	Forsius-Eriksson syndrome	Aland Island Eye Disease (AIED)

193	Fructose-1,6-bisphosphate aldolase B (ALDOB) deficiency	Hereditary fructosuria Aldolase B (ALDB) deficiency
194	GABA-transaminase deficiency	Gamma-aminobutiric acid transaminase deficiency
195	Galloway-Mowat syndrome	Nephrosis, neuronal dysmigration syndrome Galloway syndrome Microcephaly, hiatus hernia, nephrotic syndrome
196	GATAD2B-associated neurodevelopmental disorder (GAND)	Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome GAND syndrome Mental retardation, autosomal dominant 18 (MRD18)
197	GBE1 GSD4	
198	Genitopatellar syndrome (GTPTS)	Absent patellae, scrotal hypoplasia, renal anomalies, facial dysmorphism, and mental retardation KAT6B-related disorder
199	Ghosal hematodiaphyseal dysplasia	Ghosal syndrome TBXAS1-related disorder
200	Gillespie syndrome	Aniridia, cerebellar ataxia, intellectual disability syndrome
201	Glucocorticoid deficiency with achalasia	Allgrove syndrome Triple A syndrome
202	Glucose/galactose malabsorption SLC5A1	
203	Glutamate receptor, ionotropuc, N-methyl-D-aspartate, subunit 2B (GRIN2B) gene mutation	Intellectual developmental disorder, autosomal dominant 6
204	Glutaric aciduria, type 1 (GA1)	Glutaric acidemia type 1 (GA1) Glutaryl-coenzyme A dehydrogenase deficiency (GCDHD)
205	Glycogen storage disease IXa (GSD9A)	
206	Glycogen storage disease type 1a (GSD1a)	Glycogen storage disease, type I (GSD I) von Gierke disease Hepatorenal glycogen storage disease
207	GMPPB HOMOZYGOUS VARIANT RELATED LGMB	Muscular dystrophy-dystroglycanopathy
208	Gordon hyperkalemia-hypertension syndrome	Pseudohypoaldosteronism type 2
209	Gorham syndrome	Gorham's disease Hemangiomata with osteolysis Phantom bone disease
210	GPATCH11RETINITIS PIGMENTOSA	
211	Greig cephalopolysyndactyly syndrome	
212	Growth factor, ERV1-like (GFER) gene mutation	Congenital cataract, progressive muscular hypotonia, hearing loss, developmental delay syndrome
213	GYS2	
214	Hamartoma of hypothalamus	Tuber cinereum hamartoma

215	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
216	Harlequin ichthyosis	
217	Hemihyperplasia with multiple lipomatosis (HHML) syndrome	
218	Hemolytic uremic CFH	
219	Hereditary hyperekplexia	Congenital stiff man syndrome Familial startle disease Kok disease
220	Hereditary Hypophosphatemic Rickets with Hypercalciuria (HHRH)	
221	Hereditary limphedema type 1	Milroy lymphedema Nonne-Milroy lymphedema
222	Hereditary pancreatitis	Protease, serine 1 (PRSS1) gene mutation
223	Hereditary pheochromocytoma and paraganglioma	
224	Hereditary sensory and autonomic neuropathy (HSAN) type 9	Tectonin beta-propeller repeat-containing protein 2 (TECPR2) gene mutation Autosomal recessive spastic paraplegia type 49
225	Hermansky-Pudlak syndrome (HPS)	Albinism with hemorrhagic diathesis Alpha storage pool disease
226	Heterogeneous nuclear ribonucleoprotein D (HNRNPD) gene mutation	
227	HHH syndrome (Hyperornithinemia, Hyperammonemia, Homocitrullinuria)	Triple H syndrome Ornithine translocase ORNT1 deficiency Ornithine carrier deficiency
228	Histiocytosis-lymphadenopathy plus syndrome	H syndrome SLC29A3 spectrum disorder
229	HIVEP2 intellectual disability	
230	Houge-Janssens syndrome 1 (HJS1)	
231	Houge-Janssens syndrome 2 (HJS2)	
232	Houge-Janssens syndrome 3 (HJS3)	
233	HPDL HOMOZYGOUS MUTATION RELATED SPASTIC PARAPLEGIA 83	HPDL-related Leigh-like encephalopathy
234	Human immunodeficiency virus (HIV) disease	איידס
235	Hunter disease	Mucopolysaccharidosis, MPS-II MPS2 Iduronate 2-sulfatase deficiency
236	Hyperekplexia epilepsy syndrome	ARHGEF9 RELATED DISORDER
237	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency (SAHHD)	
238	Hyperphosphatemic familial tumoral calcinosis	
239	Hyperuricemia, pulmonary hypertension, renal failure, alkalosis syndrome	HUPRA syndrome

240	Hypotonia Hypoventilation Impaired Intellectual Development Dysautonomia Epilepsy and Eye	HIDEA syndrome
241	Hypotonia, Ataxia and Delayed Development syndrome (HADDS)	
242	Hypotonia, ataxia, developmental delay, and tooth enamel defect syndrome (HADDTS)	CTBP1-related disorder
243	I-cell disease	Mucolipidosis II GNPTAB-related disorder
244	Ichthyosis, Congenital; Autosomal Recessive 1 (ARCI1)	COLLODION BABY, SELF-HEALING; SHCB ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 1, WITH BATHING SUIT DISTRIBUTION
245	IDDHBA syndrome	Intellectual developmental disorder with hypotonia and behavioral abnormalities
246	Idiopathic non-lupus full-house nephropathy (FHN)	
247	ILR6 deficiency Hyper-IgE syndrome 5	
248	Immunodeficiency 14	Activated PI3K-delta syndrome
249	Immunodysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome	Autoimmune enteropathy type 1
250	Impaired intellectual development and distinctive facial features with or without cardiac defects (MRFACD)	Asadollahi-Rauch syndrome
251	Infantile Neuroaxonal Dystrophy (INAD1)	Seitelberger's disease
252	INFLAMMATORY BOWEL DISEASE 28, AUTOSOMAL RECESSIVE; IBD28 (IL10RA)	
253	INOSITOL POLYPHOSPHATE-5-PHOSPHATASE K (INPP5K)	SKELETAL MUSCLE- AND KIDNEY-ENRICHED INOSITOL PHOSPHATASE (SKIP)
254	INPPL1 Opsismodysplasia	
255	Intellectual developmenta disorder with severe speech and ambulation defects (IDDSSAD)	ACTL6B gene mutation
256	Intellectual developmental disorder with dysmorphic facies and ptosis (BRPF1)	
257	INTELLECTUAL DEVELOPMENTAL DISORDER WITH IMPAIRED LANGUAGE AND DYSMORPHIC FACIES (IDDILF) (DDX6)	
258	Intellectual developmental disorder, AR 13	TRAPPC9-related disorder
259	Intellectual developmental disorder, autosomal dominant 29 (MRD29)	SETBP1 haploinsufficiency disorder (SETBP1-HD)
260	Intellectual developmental disorder, autosomal dominant 45 (MRD45)	CIC gene mutation-related disorder
261	Intellectual developmental disorder, autosomal dominant 53 (CAMK2A)	

262	Intellectual developmental disorder, X-linked 21(IL1RAPL1)	
263	Intellectual developmental disorder, X-linked 96 (SYP)	SYNAPTOPHYSIN SYP gene mutation XLID 96
264	Intellectual developmental disorder, X-linked 99 (USP9X)	UBIQUITIN-SPECIFIC PROTEASE 9, X-LINKED DROSOPHILA FAT FACETS-RELATED, X-LINKED (DFFRX FAM)
265	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)
266	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type	DDX3X related disorder
267	Intellectual developmental disorder, X-linked syndromic, Turner type HUWE1	
268	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
269	Intellectual disability, expressive aphasia, facial dysmorphism syndrome	
270	Intellectual disability, facial dysmorphism syndrome due to SETD5 haploinsufficiency	
271	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
272	Intractable diarrhea of infancy syndrome (IDIS)	Diarrhea 11, malabsorptive, congenital (DIAR11) Proline- and glutamate-rich protein with coiled-coil domain 1 (PERCC1) gene mutation
273	Intrauterine restriction, metaphyseal dysplasia, adrenal hypoplasia congenita, and genital anomaly (IMAGe) syndrome	
274	IRAK4 Immunodeficiency	
275	ITPR1 mutation related spinocerebellar ataxia 29	Spinocerebellar ataxia type 29 (SCA29) Congenital nonprogressive spinocerebellar ataxia
276	ITPR3 IMMUNODEFICIENCY 133 WITH ECTODERMAL DYSPLASIA WITH OR WITHOUT PERIPHERAL NEUROPATHY	
277	Jacobsen Syndrome	Distal partial deletion of long arm of chromosome 11
278	Jansen-de Vries syndrome (JDVS)	
279	Joubert syndrome with renal defect	NPHP1-related nephronophthisis

280	KBG syndrome	Short stature, facial and skeletal anomalies, intellectual disability, macrodontia syndrome
281	KCNH5 Developmental and epileptic encephalopathy 112	
282	KCNJ hyperinsulinism	
283	KCNQ2	
284	KDM2B Neurodevelopmental disorder with congenital cardiac defects and variable renal and ocular abnormalities	
285	KIF1A associated neurological disorder (KAND)	
286	Kindler's syndrome	
287	KINSSHIP syndrome	
288	Kleefstra syndrome	due to del(9)(q34) due to monosomy 9q34
289	Kleefstra syndrome 2	
290	Klinefelter syndrome variant	49 XXXXY
291	Klippel Trenaunay syndrome	Klippel Trenaunay Weber syndrome
292	KMT2B Dystonia Intellectual developmental disorder	
293	Kohlschutter syndrome	Amelocerebrohypohidrotic syndrome Epilepsy, mental deterioration and yellow teeth Epilepsy, dementia, amelogenesis, imperfecta syndrome Kohlschutter-Tonz syndrome
294	Kostmann syndrome	Infantile agranulocytosis
295	Kozlowski spondylometaphyseal dysplasia	TRPV4 related skeletal dysplasia
296	KREMEN1 related ectodermal dysplasia 13	Ectodermal dysplasia 13, hair/tooth type (ECTD13)
297	Lamb Shaffer syndrome	SOX5 haploinsufficiency syndrome
298	Lamellar Ichthyosis	Collodion baby
299	Lateral meningocele syndrome (LMNS)	NOTCH3-related lateral meningocele syndrome Lehman syndrome
300	Leukocyte adhesion deficiency (LAD) type 2	
301	Liang-Wang syndrome (LIWAS)	
302	Limb-girdle Muscular Dystrophy, Type 28, Autosomal Recessive	LGMDR28
303	Lipid transport defect of intestine	Chylomicron retention disease Anderson syndrome
304	Lissencephaly TUBA1A	
305	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
306	Loeys-Dietz syndrome	
307	Long chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency (LCHADD)	

308	Low density lipoprotein receptor-related protein 5 related primary osteoporosis (LRP5)	LRP5-related primary osteoporosis
309	Lowe syndrome	Cerebro-oculorenal dystrophy Oculocerebrorenal syndrome Lowe-Bickel syndrome Lowe-Terrey-MacLachlan syndrome Phosphatidylinositol-4,5-bisphosphate-5-phosphatase deficiency
310	LSM1 GENE MUTATION RELATED DISORDER	LSM1 PROTEIN CANCER-ASSOCIATED SM-LIKE PROTEIN (CASM) LSM1 HOMOLOG, mRNA DEGRADATION-ASSOCIATED
311	Lymphangiomatosis	Generalized Lymphatic Anomaly GLA
312	Lysine demethylase 5c (KDM5C) gene mutation	
313	Lysinuric protein intolerance (LPI)	Congenital lysinuria
314	MACROPHAGE EXPRESSED GENE 1 (MPEG1)	PERFORIN 2 (PRF2) MPG1 Immunodeficiency 77
315	MAJEED SYNDROME (LPIN2)	
316	Malan overgrowth syndrome	Sotos syndrome 2
317	Mandibulofacial dysostosis with microcephaly	Mandibulofacial dysostosis, Guion-Almeida type Elongation factor Tu GTP-binding comain-containing 2 (EFTUD2) gene mutation
318	MAP/MICROTUBULE AFFINITY-REGULATING KINASE 2 (MARK2)	ELKL MOTIF KINASE (EMK1) PAR1, C. ELEGANS, HOMOLOG OF, B (PAR1B)
319	Maroteaux-Lamy syndrome (ARSB)	MPS VI - Mucopolysaccharidosis VI
320	Maternally inherited Leigh syndrome (MILS)	Maternally inherited infantile subacute necrotizing encephalopathy
321	MBD5-associated neurodevelopmental disorder (MAND)	Intellectual developmental disorder, autosomal dominant 1
322	McCune Albright syndrome	
323	MED12 FGS1	Opitz-Kaveggia syndrome
324	Megalencephaly capillary malformation (MCAP)	Macrocephaly-capillary malformation (M-CM, MCM) Megalencephaly-cutis marmorata telangiectatica congenita syndrome (MCMTC)
325	Meier-Gorlin syndrome	Ear, patella, short stature syndrome Microtia, absent patellae, micrognathia syndrome
326	MEIS2 syndrome	Cleft palate, cardiac defects, and impaired intellectual development Cardiac malformation, cleft lip/palate, microcephaly and digital anomalies
327	Menke Hennekam syndrome	
328	Metaphyseal anadysplasia type 2	Matrix metalloproteinase 9 (MMP9) gene mutation
329	Microcephalic osteodysplastic primordial dwarfism type II (MOPDII)	Majewski osteodysplastic primordial dwarfism type II
330	Microcephalic primordial dwarfism Alazami type	Alazmai syndrome

331	Microcephaly, corpus callosum hypoplasia, intellectual disability, facial dysmorphism syndrome	
332	Miller-Dieker lissencephaly syndrome	
333	Mitochondrial encephalocardiomyopathy due to transmembrane protein 70 mutation (TMEM70)	
334	MITOCHONDRIAL ENCEPHALOPATHY LACTIC ACIDOSIS AND STROKE LIKE EPISODES (MELAS)	Juvenile myopathy, encephalopathy, lactic acidosis, stroke
335	Mitochondrial enoyl coA reductase protein-associated neurodegeneration (MEPAN) syndrome	Mitochondrial enoyl CoA hydratase 1 deficiency (ECHS1D) Mitochondrial enoyl coA reductase (MECP) gene mutation
336	Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to mitochondrial transfer ribonucleic acid translation optimization 1 deficiency MTO1	COXPD10
337	Molybdenum cofactor deficiency (MoCD)	Combined molybdenoflavoprotein enzyme deficiency Combined xanthine oxidase and aldehyde oxidase deficiency Hereditary xanthinuria type 2
338	Mosaic trisomy 16 syndrome	
339	Moyamoya disease	
340	MPPH syndrome	Megalencephaly, polymicrogyria, postaxial polydactyly, hydrocephalus syndrome
341	MSN IMD50	
342	MT-ATP6 mitochondrial disease	
343	Mulchandani-Bhoj-Conlin syndrome	maternal uniparental disomy of chromosome 20 (Upd(20)mat)
344	MULLEGAMA-KLEIN-MARTINEZ SYNDROME STAG2 RELATED DISORDER (MKMS)	
345	Multicentric carpotarsal osteolysis syndrome	
346	Multiple endocrine neoplasia, type 3 (MEN3)	Multiple endocrine neoplasia, type 2B (MEN2B)
347	Multiple sulfatase deficiency (MSD)	Juvenile sulfatidosis, Austin type Mucosulphatidosis
348	Multisystemic smooth muscle dysfunction syndrome	Actin, alpha-2, smooth muscle, aorta (ACTA2) gene mutation
349	Muscle eye brain disease	
350	Muscle phosphoglycerate mutase deficiency	Phosphoglucomutase deficiency
351	Myhre syndrome	
352	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)

353	Myofibrillar myopathy	
354	Myopathy X linked with excessive autophagy VMA21	
355	Myosin, cardiac, heavy chain, beta (MYHCB, MYH7) gene mutation	
356	Nabais Sa-de Vries syndrome (SPOP)	SPECKLE-TYPE BTB/POZ PROTEIN (SPOP)
357	Nabais Sa-de Vries syndrome, type 2 (NSDVS2)	NEURODEVELOPMENTAL DISORDER WITH RELATIVE MACROCEPHALY AND WITH OR WITHOUT CARDIAC OR ENDOCRINE ANOMALIES, NEDMACE
358	NALCN Syndrome	Congenital contractures of the limbs and face, hypotonia, and developmental delay Hypotonia, infantile, with psychomotor retardation and characteristic facies 1
359	NARS2 related disorder	Deafness. Autosomal recessive 94 (DFNB94)
360	NEDAMSS	
361	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
362	Neurodegeneration due to 3-hydroxyisobutyryl coenzyme A hydroxylase (HIBCH) deficiency	Methacrylic aciduria Valine metabolic defect
363	Neurodegeneration, childhood-onset, with brain atrophy (CONDBA)	UBTF related motor and cognitive regression syndrome
364	Neurodevelopmental disorder w/ hypotonia, variable intellectual + behavioral abnormalities (NEDHIB)	POLR2A-related disorder
365	Neurodevelopmental disorder with dysmorphic facies and distal limb anomalies (NEDDFL)	BPTF-related disorder
366	Neurodevelopmental disorder with dysmorphic facies and distal skeletal anomalies (NEDDFSA)	Zinc finger miz-domain containig 1(ZMIZ1) gene mutation
367	Neurodevelopmental disorder with eye movement abnormalities and ataxia (NEDEMA)	FRMD5-related disorder
368	Neurodevelopmental disorder with hypotonia, dysmorphic facies, and skeletal anomalies, with or without seizures (NEDFSS)	TRPM3-related disorder
369	Neurodevelopmental disorder with microcephaly, short stature, and speech delay TRAPPC10	
370	Neuroendocrine cell hyperplasia of infancy	NEHI syndrome
371	N-glycanase 1 congenital disorder of deglycosylation (NGLY1)	Alacrimia, choreoathetosis, liver dysfunction syndrom Deficiency of N-glycanase 1
372	Nicolaides-Baraitser syndrome	SWI/SNF-related, matrix-associated, actin-dependant regulator of chromatin, subfamily A, member 2 (SMARCA2) gene mutation

373	Niemann-Pick disease, type A	
374	Niemann-Pick disease, type C	Supraoptic vertical ophthalmoplegia
375	NOG related disorder	Teunissen-Cremers syndrome Stapes ankylosis with broad thumbs and toes Ankylosis of stapes, hyperopia, broad thumbs, broad first toes and syndactyly
376	NPHS2 related nephrotic syndrome	Familial idiopathic steroid-resistant nephrotic syndrome Genetic SRNS Hereditary steroid-resistant nephrotic syndrome
377	NPR2 HOMOZYGOUS MUTATION RELATED ACROMESOMELIC DYSPLASIA TYPE 1 Maroteaux type	Epiphyseal chondrodysplasia, Miura type Short stature with nonspecific skeletal abnormalities
378	Obesity, morbid, due to leptin receptor deficiency (LEPR)	OBR
379	Occipital pachygyria and polymicrogyria	LAMB1-related disorder
380	Oculocerebrofacial syndrome Kaufman type	
381	Oculo-facio-cardio-dental syndrome	Oculofaciocardiodental syndrome
382	O'Donnell-Luria-Rodan syndrome (ODLURO)	Lysine-specific methyltransferase 2E (KMT2E) gene mutation
383	Odonto-onycho-dermal dysplasia	Ectodermal dysplasia 16, hair/tooth/nail type (ECTD16) WNT10A-related disorder
384	Ogden syndrome	
385	Ohdo sydrome, Say-Barber-Biesecker-Young-Simpson (SBBYS) variant	Blepharophimosis, intellectual disability syndrome Say-Barber-Biesecker-Young-Simpson (SBBYS) variant KAT6B-related disorder
386	Okur-Chung neurodevelopmental syndrome (OCNDS)	OKUR CHUNG SYNDROME -CSNK2A1 MUTATION
387	PACS1 NEURODEVELOPMENTAL DISORDER	PHOSPHOFURIN ACIDIC CLUSTER SORTING PROTEIN Schuurs-Hoeijmakers syndrome PACS RELATED EPILEPTIC ENCEPHALOPATHY
388	PACS2 Developmental and epileptic encephalopathy 66	
389	Pallister Killian syndrome	Tetrasomy 12p syndrome Killian-Teschler-Nicola syndrome Pallister mosaic syndrome
390	PAN Polyarteritis nodosa	Periarteritis nodosa
391	Paraneoplastic opsoclonus-myoclonus-ataxia syndrome; POMA	Dancing eye syndrome Dancing eye-dancing feet syndrome Opsoclonus-myoclonus-ataxia syndrome; OMA Kinsbourne syndrome
392	Parkes Weber syndrome	
393	Paroxysmal nonkinesigenic dyskinesia, 3, with or without generalized epilepsy (PNKD3)	Generalized epilepsy and paroxysmal dyskinesia (GEPD)

394	PCDHGC4 Neurodevelopmental disorder with poor growth and skeletal anomalies	
395	PDCD10 Cerebral cavernous malformations-3 (CCM3)	
396	Pearson's syndrome	
397	Pelizaeus-Merzbacher disease (PMD)	Sudanophilic leukodystrophy
398	Penta X syndrome	XXXXX syndrome
399	Perching syndrome	Kelch like family member 7-related Bohring-Opitz-like and Crisponi/cold-induced sweating-like overlap syndrome
400	Perlman syndrome	
401	Permanent neonatal diabetes mellitus	Developmental delay, epilepsy and neonatal diabetes 1 (DEND)
402	Peutz Jeghers syndrome PJS	Peutz-Jeghers polyposis Peutz-Jeghers lentiginosis syndrome
403	PFIC4 Cholestasis progressive familial intrahepatic 4	
404	PHACE syndrome	Phace syndrome and Moya Moya Disease Pascual-Castroviejo syndrome type 2
405	PHACES syndrome	
406	PHD finger protein 21A (PHF21A) gene mutation	
407	Phelan-McDermid syndrome	22q13.3 deletion syndrome SH3 and multiple ankyrin repeat domains 3 (SHANK3) gene mutation
408	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
409	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
410	Phosphoenolpyruvate carboxykinase (PEPCK-C) deficiency	
411	Pierpont syndrome	Plantar lipomatosis, facial dysmorphism, developmental delay syndrome
412	Pitt-Hopkins syndrome (PTHS)	
413	Pitt-Hopkins-like syndrome 1 (PTHSL1)	
414	Pituitary stalk interruption syndrome	Ectopis neurohypophysis
415	Pleuropulmonary blastoma (PPB)	

416	Pleuropulmonary blastoma familial tumor and dysplasia syndrome (PPBFTDS)	DICER1 tumor-predisposition syndrome PPB familial tumor and dysplasia syndrome
417	POGZ White-Sutton syndrome	
418	Poirier-Bienvenu neurodevelopmental syndrome (POBINDS)	CSNK2B-related disorder
419	POMT1-related limb-girdle muscular dystrophy R11	Limb-girdle muscular dystrophy type 2K; LGMD type 2K POMT1-related LGMD R11
420	Pontocerebellar hypoplasia type 9 (PCH9)	
421	Popliteal pterygium syndrome	
422	Port-wine stain with oculocutaneous melanosis	Phakomatosis pigmentovascularis
423	Potocki-Shaffer syndrome	Chromosome 11p11.2 deletion syndrome Proximal 11p deletion syndrome (P11pDS)
424	PPP2R1A	Houge-Janssens syndrome 2
425	PRICKLE1 HOMOZYGOUS MUTATION RELATED DISORDER	Progressive myoclonus epilepsy with ataxia
426	Primary hyperoxaluria type 1	Glycolic aciduria
427	Primary microcephaly, mild intellectual disability, young-onset diabetes syndrome	
428	Primrose syndrome	Intellectual disability, cataract, calcified pinna, myopathy syndrome ZBTB20-related syndrome
429	PRKAG2-related hypertrophic cardiomyopathy	Familial hypertrophic cardiomyopathy 6
430	Progeria syndrome	Hutchinson-Gilford syndrome
431	Progressive myoclonic epilepsy (PME) type 8 (CERS1)	
432	Propionic Acidemia	Ketotic hyperglycinaemia Hyperglycinaemia with ketosis and leucopenia Propionyl-CoA carboxylase (PCC) deficiency
433	Proteolipid protein 1(PLP1) gene mutation	
434	Pseudohypoaldosteronism type 1, recessive form	Pseudohypoaldosteronism type 1B Pseudohypoaldosteronism, Persian-Jewish type
435	Pseudohypoparathyroidism	Guanine nucleotide-binding protein, alpha-stimulating activity polypeptide 1 (GNAS) gene mutation Parathyroid hormone resistant hypoparathyroidism
436	Pseudohypoparathyroidism type 1A (PHP1A)	Albright hereditary osteodystrophy (AHO)
437	Pseudohypoparathyroidism type 2	

438	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)
439	Pulmonary alveolar proteinosis (PAP)	
440	Purine rich element binding protein A (PURA) syndrome	
441	Pyknodysostosis	Maroteaux-Lamy syndrome type II Cathepsin k (CTSK) gene mutation
442	PYRUVATE DEHYDROGENASE COMPLEX, COMPONENT X (PDHX)	PDX1 PYRUVATE DEHYDROGENASE COMPLEX, E3-BINDING PROTEIN SUBUNIT (E3BP) PYRUVATE DEHYDROGENASE COMPLEX, LIPOYL-CONTAINING COMPONENT X Lacticacidemia due to PDX1 deficiency
443	Radial aplasia-trhombocytopenia (TAR) syndrome	
444	Radioulnar synostosis with amegakayocytic thrombocytopenia (RUSAT) 2	MDS1 and EVI1 complex (MECOM) gene mutation
445	Rahman syndrome	H1-4-related neurodevelopmental disorder HIST1H1E-related disorder
446	RASA1 Parkes Weber syndrome	
447	RAS-associated autoimmune leukoproliferative disease (RALD)	Autoimmune leukoproliferative syndrome type 4
448	Rasmussen subacute encephalitis	Rasmussen syndrome Rasmussen encephalitis
449	Rauch-Steindl syndrome (RAUST) NSD2	
450	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
451	Renal coloboma Papillorenal	
452	Renpenning syndrome	Golabi-Ito-Hall syndrome Hamel cerebropalatocardiac syndrome Polyglutamine-binding protein 1 (PQBP1) gene mutation
453	Resistance to insulin-like growth factor 1 (IGF1)	Insulin-like growth factor 1 receptor (IGF1R) gene mutation
454	RFX3 Autism	
455	ROHHAD syndrome	Rapid-onset childhood obesity, hypothalamic dysfunction, hypoventilation, autonomic dysregulation syndrome
456	ROR2 skeletal dysplasia	Robinow syndrome

457	Rubinstein-Taybi syndrome	
458	SARCOGLYCAN, ALPHA (SGCA)	ADHALIN (ADL) DYSTROGLYCAN 2 (DAG2) DYSTROPHIN-ASSOCIATED GLYCOPROTEIN, 50-KD 50-DAG Muscular dystrophy, limb-girdle, autosomal recessive 3
459	Schaaf Yang syndrome	MAGE family member L2-related Prader-Willi-like syndrome MAGEL2 related Prader Willi-like syndrome
460	Schinzel-Giedion syndrome	
461	Schuurs-Hoeijmakers syndrome	
462	SCN5A Long QT syndrome 3	Brugada syndrome
463	Seizures, benign familial infantile, 2	PRRT2-related benign familial seizures
464	Selective malabsorption of cyanocobalamin (B12)	Imerslund-Grasbeck sydrome (IGS)
465	Sensenbrenner–Dorst–Owens syndrome	Sensenbrenner's syndrome Levin syndrome Cranioectodermal dysplasia (WDR35 mutation)
466	Sepiapterin reductase deficiency	
467	Severe congenital nemaline myopathy (TNNT1 mutation)	Nemaline myopathy, Amish type (ANM) Nemaline myopathy 5A, autosomal recessive, severe infantile (NEM5A)
468	Severe infantile form of carnitine palmitoyltransferase II deficiency	
469	SH3 and multiple ankyrin repeat domains 2 (SHANK2) gene mutation	
470	Short QT syndrome 2	KCNQ1-related Short QT syndrome
471	SHORT syndrome	
472	Shprintzen-Goldberg syndrome (SGS)	Marfanoid craniosynostosis syndrome Shprintzen Goldberg craniosynostosis syndrome
473	Shukla-Vernon syndrome (SHUVER)	
474	Sialuria	
475	Sifrim-Hitz-Weiss syndrome	CHD4-related neurodevelopmental disorder
476	Signal transducer and activator of trascription 3 (STAT3) gene mutation	
477	Sjogren-Larsson syndrome	Fatty alcohol-nicotinamide adenine dinucleotide oxidoreductase deficiency
478	Skraban Deardorff syndrome	Intellectual disability, seizures, abnormal gait, facial dysmorphism syndrome
479	Skraban-Deardorff syndrome WDR26	
480	SMARCAL1 Schimke immunoosseous dysplasia	
481	SMC3-related Cornelia de Lange syndrome (CdLS)	Cornelia de Lange syndrome 3
482	Smith Kingsmore syndrome	Macrocephaly, intellectual disability, neurodevelopmental disorder, small thorax (MINDS) syndrome
483	Snijders Blok-Campeau syndrome	CHD3-related developmental delay, speech delay, intellectual disability, abnormalities of vision, facial dysmorphism syndrome
484	Snyder-Robinson syndrome	X-linked intellectual disability Snyder type

485	Sodium voltage-gated channel, alpha subunit 4 (SCN4A) gene mutation	
486	Solute carrier family 9, member 7 (SLC9A7) gene mutation	
487	SOX2 anophthalmia syndrome	Syndromic microphthalmia 3 Anophthalmia-esophageal-genital (AEG) syndrome
488	SPAST VARIANT Spastic paraplegia 4	
489	Spinal muscular atrophy, lower extremity-predominant 1, AD	DYNC1H1-related spinal muscular atrophy
490	Spinocerebellar ataxia type 21	SCY1-lime (SCYL1) gene mutation
491	Spinocerebellar ataxia type 26 (SCA26)	EEF2 related neurodevelopmental disorder
492	Spondilometaphyseal dysplasia	
493	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type (SEMDIST)	Spondyloepimetaphyseal dysplasia with severe short stature
494	Spondyloepiphyseal dysplasia (SED)	Spondyloepiphyseal dysplasia congenita (SEDC)
495	Spondylometaphyseal dysplasia Schmidt type	Spondylometaphyseal dysplasia Algerian type Spondylometaphyseal dysplasia with severe genu valgum
496	Spondylometaphyseal dysplasia with combined immunodeficiency; SPENCDI	Roifman-Melamed syndrome Spondyloenchondrodysplasia with immune dysregulation
497	SPTBN1 related developmental delay, impaired speech and behavioral abnormalities (DDISBA)	
498	Stankiewicz-Isidor syndrome (STISS)	PSMD12-related disorder
499	Streptococcus infection in conditions classified elsewhere and of unspecified site, Streptococcus, unspecified	
500	Synaptic RAS-GTPase-activating protein 1 (SYNGAP1) gene mutation	
501	Systematized epidermal nevus	Nevus unius lateris
502	SZT2 related developmental and epileptic encephalopathy 18	DEE18
503	SZT2 subunit of KICSTOR complex gene mutation	
504	Tatton Brown Rahman syndrome	DNA methyltranspherase 3A (DNMT3A) gene mutation Tall stature, intellectual disability, facial dysmorphism syndrome
505	TCF20	
506	Temple syndrome	
507	TET3-related Beck-Fahrner syndrome; TET3-BEFAHRS	
508	TETRATRICOPEPTIDE REPEAT DOMAIN-CONTAINING PROTEIN 2 (TTC26)	INTRAFLAGELLAR TRANSPORT 56, CHLAMYDOMONAS, HOMOLOG OF (IFT56) Biliary, renal, neurologic, and skeletal syndrome (BRENS)
509	THUMPD1 related disorder	Neurodevelopmental disorder with speech delay and variable ocular anomalies (NEDSOA)
510	Tietze's disease	

511	Timothy syndrome	Long QT syndrome type 8; LQT8 Long QT syndrome-syndactyly syndrome
512	Tooth agenesis-colorectal cancer syndrome (ODCRCS)	AXIN2-related disorder
513	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)
514	Transaldolase deficiency (TALDO1)	
515	Transcobalamin 2 (TCN2) deficiency	
516	Transketolase (TKT) deficiency	Short stature-developmental delay-congenital heart defect syndrome
517	Trichohepatoenteric syndrome	
518	Trichorhinophalangeal syndrome	
519	Trio Gene Intellectual developmental disorder autosomal dominant 44 with microcephaly	TRIPLE FUNCTIONAL DOMAIN
520	tRNA methyltransferase 10A (TRMT10A) gene mutation	
521	tRNA-VAL, MITOCHONDRIAL(MT-TV) (MTTV)	
522	TUBB3	Cortical dysplasia, complex, with other brain malformations 1 Fibrosis of extraocular muscles, congenital, 3A
523	Turner's phenotype - ring chromosome karyotype	
524	Twin reversal arterial perfusion syndrome TRAP	
525	Tyrosinemia type 1	FAH-related tyrosinemia Hepatorenal tyrosinemia
526	Tyrosinemia type 2	Hypertyrosinemia, Richner-Hanhart type Hypertyrosinemia, Oregon type
527	Tyrosinemia type 3	
528	UBE2A Intellectual developmental disorder Nascimento type	
529	Usher syndrome type 3; USH3	Retinitis pigmentosa-deafness syndrome type 3
530	Ventriculomegaly with cystic kidney disease (VMCKD)	Heterozygous CRB2-related disorder
531	Verheij syndrome	8q24.3 microdeletion syndrome Del(8)(q24.3) Monosomy 8q24.3
532	Verloes Bourguignon syndrome	Autosomal recessive brachyolmia and amelogenesis imperfecta syndrome Dental anomalies and short stature (DAAS)
533	Ververi Brady syndrome	Glutamine rich 1-related intellectual disability, chondrodysplasia syndrome
534	Vici syndrome	Dionisi Vici Sabetta Gambarara syndrome
535	VPS11-related autosomal recessive hypomyelinating leukoencephalopathy	Leukodystrophy, hypomyelinating, 12 (HLD12) VPS11 core subunit of CORVET and HOPS complexes-related autosomal recessive hypomyelinating leukodystrophy
536	Waardenburg Shah syndrome	Waardenburg Hirschsprung syndrome

537	WDFY3	
538	Weaver syndrome (WVS)	
539	Weill-Marchesani syndrome	Brachydactyly-spherophakia syndrome
540	Weiss-Kruszka syndrome (WSKA)	Zinc finger protein 462 (ZNF462) gene mutation
541	Wieacker Wolff syndrome	Intellectual disability, developmental delay, contracture syndrome Foot contracture, muscle atrophy, oculomotor apraxia syndrome
542	Wiedemann Steiner syndrome	Hypertrichosis, short stature, facial dysmorphism, developmental delay syndrome
543	Wilms tumor, aniridia, genitourinary anomalies and mental retardation (WAGR) syndrome	Chromosome 11p13 deletion syndrome
544	Wiscott-Aldrich syndrome	Aldrich syndrome
545	Wiskott-Aldrich syndrome (WAS)	Eczema, thrombocytopenia, immunodeficiency syndrome
546	Witteveen Kolk syndrome (WITKOS)	SIN3A-related intellectual disability syndrome
547	Wolfram syndrome	Marquardt-Loriaux syndrome DIDMOAD (diabetes insipidus, diabetes mellitus, optic atrophy, deafness) syndrome
548	Wolman disease	Primary familial xanthomatosis with adrenal calcification Familial visceral xanthomatosis Deficiency of cholesterol esterase AND triacylglycerol lipase Wolman xanthomatosis Acid esterase deficiency
549	X LINKED CLCN5 RELATED DENT 1 DISEASE	CHLORIDE CHANNEL, VOLTAGE-GATED, K2 (CLCK2) Hypophosphatemic rickets Nephrolithiasis, type I Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis
550	X LINKED IQSEC2	IQSEC2-related syndromic intellectual disability IQSEC2 (IQ motif and Sec7 domain 2) related syndromic intellectual disability Severe intellectual disability, progressive postnatal microcephaly, midline stereotypic hand movements syndrome
551	Xia-Gibbs syndrome	
552	X-linked intellectual disability Cabezas type	Cullin 4b (CUL4B) gene mutation
553	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
554	X-linked intellectual disability Siderius type	
555	X-linked intellectual disability with cerebellar hypoplasia syndrome	Oligophrenin-1 syndrome (OPHN1)
556	X-linked intellectual disability, hypotonia, movement disorder syndrome	

557	X-linked non progressive cerebellar ataxia	
558	X-linked reticulate pigmentary disorder with systemic manifestation syndrome	Partington disease
559	Xq25 microduplication	
560	Zhu Tokita Takenouchi Kim (ZTTK) syndrome	
561	ZINC FINGER- AND BTB DOMAIN-CONTAINING PROTEIN 18 (ZBTB18)	Intellectual developmental disorder, autosomal dominant 22
562	Zinc Finger Homeobox 3 (ZFHX3)	
563	ZINC FINGER MYND DOMAIN-CONTAINING PROTEIN 11 (ZMYND11)	BS69
564	Zinc finger protein 292 (ZNF292) gene mutation	Intellectual developmental disorder, autosomal dominant 64
565	Zinc finger protein 335 (ZNF335) gene mutation	Microcephaly 10, primary, autosomal recessive
566	ZMYM2	

נספח - רשימת מחלות שכבר אינן מוכרות כנדירות

#	מחלה	תאריך הסרה מהרשימה
1	Pediatric multiple sclerosis Monosomy 14q22-q23 Frias Syndrome	14/04/2026
2	Phosphatase and tensin homolog (PTEN) gene mutation Monosomy 16p11.2p12.2	14/04/2026
3	Potocki Lupski syndrome Del(3)p(25.3) 3p25.3 microdeletion syndrome Monosomy 3p25.3 Mental retardation-epilepsy-stereotypic hand movement syndrome	14/04/2026
4	Pseudotumor cerebri Dup(3q) syndrome Trisomy 3q26 Dup(3)(q26)	14/04/2026
5	Renal nutcracker syndrome MEF2C haploinsufficiency syndrome	14/04/2026
6	Rett syndrome; RTS Monosomy 9q22.3 syndrome Microdeletion 9q22.3	14/04/2026
7	Scimitar syndrome	14/04/2026
8	Set domain-containing protein 5 (SETD5) gene mutation Acetyl-CoA acyltransferase 3-ketoacyl-CoA thiolase Acetyl-coenzyme A acyltransferase Beta-ketothiolase	14/04/2026
9	Sharp's syndrome Landau-Kleffner syndrome	14/04/2026
10	Small G protein signaling modulator 3 (SGSM3) mutation Pfeiffer syndrome	14/04/2026
11	Sotos' syndrome	14/04/2026
12	Special AT-rich sequence-bindind protein 2 (SATB2) associated syndrome Baraitser-Winter syndrome 1 Becker nevus, syndromic or isolated, somatic mosaic Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic Thrombocytopenia 8, with dysmorphic features and developmental delay	14/04/2026
13	Systemic mast cell disease (SMCD) Acute myeloid leukemia, t(8;21) (q22;q22.1)	14/04/2026
14	THYROID HORMONE RECEPTOR, ALPHA-1 (THRA)	14/04/2026
15	Unverricht-Lundborg syndrome Deficiency of adenosine deaminase 2 (DADA2)	14/04/2026
16	Xeroderma Pigmentosum (XP)	14/04/2026
17	16p13.11 microdeletion syndrome	14/04/2026
18	8p inverted duplication deletion syndrome Desmoid fibromatosis CTNNB1 gene mutation	14/04/2026
19	Acrocephalosyndactyly	14/04/2026

20	Autosomal dominant Charcot-Marie-Tooth disease type 2U	14/04/2026
21	CACNA1A -related epilepsy Chilblain lupus Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestation	14/04/2026
22	Capillary malformation	14/04/2026
23	Cat eye syndrome (CES) Cyclin-dependent kinase 10 (CDK10) gene mutation	14/04/2026
24	Cerebrotendinous xanthomatosis (CTX) GDP-MANNOSE PYROPHOSPHORYLASE A	14/04/2026
25	Chromosomal imbalance syndrome Alexander syndrome	14/04/2026
26	Coats' disease Monocarboxylate transporter 8 deficiency	14/04/2026
27	Cobalamin C disease Alstrom syndrome	14/04/2026
28	Coffin-Lowry syndrome (CLS)	14/04/2026
29	Congenital stationary night blindness (CSNB)	14/04/2026
30	Cystathionine beta-synthase (CBS) deficiency	14/04/2026
31	Cystic lymphangioma KAT6A gene mutation	14/04/2026
32	Deficiency of isomaltase	14/04/2026
33	Dehydrodolichyl diphosphate sythase (DHDDS) gene mutation	14/04/2026
34	Epidermolysis bullosa Alpha-thalassemia/impaired intellectual development syndrome, X-linked Alpha-thalassemia/mental retardation syndrome, X-linked Alpha-thalassemia/mental retardation syndrome, nondeletion type	14/04/2026
35	Familial hemiplagic migraine type 1 Okamoto syndrome	14/04/2026
36	Gastroparesis	14/04/2026
37	Giant congenital melanocytic nevus (GCMN) Ryanodine receptor 1 (RYR1) gene receptor Periodic paralysis	14/04/2026
38	Glycogen storage disease III (GSD3) HYPER-IgE syndrome 1, AD Job syndrome	14/04/2026
39	Helsmoortel-van der Aa syndrome	14/04/2026
40	Hereditary sensory and autonomic neuropathy (HSAN)	14/04/2026
41	Hereditary spastic paraplegia (HSP) Autosomal recessive spinocerebellar ataxia type 12 SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 12; SCAR12	14/04/2026
42	Homocystinuria Hereditary spastic paraparesis Kjellin syndrome	14/04/2026
43	Hyperinsulinemic hypoglycemia, familial, 1 (HHF1)	14/04/2026
44	Hypertrophy (benign) of prostate with urinary obstruction and other lower urinary tract symptoms (LUTS)	14/04/2026
45	Isovaleric acidemia ASXL transcriptional regulator 3 deficiency syndrome	14/04/2026
46	Joubert syndrome Synaptotagmin (SYT) 1-related neurodevelopmental disorder	14/04/2026
47	Juvenile Hypophosphatasia	14/04/2026

48	Legius syndrome Hypoparathyroidism, sensorineural hearing loss, renal disease syndrome Hypoparathyroidism, deafness, renal disease (HDR) syndrome	14/04/2026
49	LYSINE METHYLTRANSFERASE 5B (KMT5B) Laurence-Moon-Biedl syndrome Laurence-Moon-Bardet-Biedl syndrome (LMBB)	14/04/2026
50	Maple syrup urine disease (MSUD) 3-Methylglutaconic aciduria type 2	14/04/2026
51	Mast cell activation syndrome (MCAS) Hyperprostaglandin E syndrome type 1	14/04/2026
52	Methyl-CpG-binding protein 2 (MECP2) gene mutation Hyperprostaglandin E syndrome type 2 Hypokalemic alkalosis with hypercalciuria antenatal type 2	14/04/2026
53	Methylmalonic acidemia Bartter syndrome CLCNKB related disorder	14/04/2026
54	Minor partial trisomy	14/04/2026
55	Mitochondrial complex I deficiency, nuclear type 33 (NDUFA6) 2q13 deleteion syndrome	14/04/2026
56	Mixed gonadal dysgenesis Congenital contractural arachnodactyly (CCA) Beals Hecht syndrome	14/04/2026
57	Moebius syndrome Hemorrhagiparous thrombocytic dystrophy Giant platelet syndrome	14/04/2026
58	Multiple congenital exostosis Aase syndrome Congenital pure red cell aplasia	14/04/2026