

MDA's Portfolio of Diseases 2024

To learn more, see <https://www.mda.org/disease/list>

Amyotrophic Lateral Sclerosis (ALS): (ALS, subtype not named)
Amyotrophic Lateral Sclerosis (ALS): ALS (subtype unknown)
Amyotrophic Lateral Sclerosis (ALS): ALS1
Amyotrophic Lateral Sclerosis (ALS): ALS2, Juvenile ALS
Amyotrophic Lateral Sclerosis (ALS): ALS4
Amyotrophic Lateral Sclerosis (ALS): ALS5
Amyotrophic Lateral Sclerosis (ALS): ALS6
Amyotrophic Lateral Sclerosis (ALS): ALS8
Amyotrophic Lateral Sclerosis (ALS): ALS9
Amyotrophic Lateral Sclerosis (ALS): ALS10
Amyotrophic Lateral Sclerosis (ALS): ALS11
Amyotrophic Lateral Sclerosis (ALS): ALS12
Amyotrophic Lateral Sclerosis (ALS): ALS13
Amyotrophic Lateral Sclerosis (ALS): ALS14
Amyotrophic Lateral Sclerosis (ALS): ALS15
Amyotrophic Lateral Sclerosis (ALS): ALS16
Amyotrophic Lateral Sclerosis (ALS): ALS17
Amyotrophic Lateral Sclerosis (ALS): ALS18
Amyotrophic Lateral Sclerosis (ALS): ALS20
Amyotrophic Lateral Sclerosis (ALS): ALS21
Amyotrophic Lateral Sclerosis (ALS): ALS22
Amyotrophic Lateral Sclerosis (ALS): ALS23
Amyotrophic Lateral Sclerosis (ALS): ALS24
Amyotrophic Lateral Sclerosis (ALS): ALSDC
Andersen-Tawill Syndrome: Andersen-Tawill Syndrome/Periodic paralysis, potassium sensitive, cardiodysrhythmic
Centronuclear Myopathy (CNM): Centronuclear Myopathy (subtype unknown)
Centronuclear Myopathy (CNM): Centronuclear myopathy, BIN 1 related/CNM2
Centronuclear Myopathy (CNM): Centronuclear myopathy, RYR1 related
Centronuclear Myopathy (CNM): Centronuclear myopathy, TTN related
Centronuclear Myopathy (CNM): Centronuclear myopathy/CNM1
Centronuclear Myopathy (CNM): Myotubular myopathy/MTM1/ CNMX/ X-linked myotubular myopathy
Charcot-Marie-Tooth Disease (CMT): Charcot-Marie-Tooth Disease (subtype unknown)
Charcot-Marie-Tooth Disease (CMT): CMT-1A
Charcot-Marie-Tooth Disease (CMT): CMT-1B
Charcot-Marie-Tooth Disease (CMT): CMT-1C
Charcot-Marie-Tooth Disease (CMT): CMT1D
Charcot-Marie-Tooth Disease (CMT): CMT1E
Charcot-Marie-Tooth Disease (CMT): CMT1F
Charcot-Marie-Tooth Disease (CMT): CMT2A1
Charcot-Marie-Tooth Disease (CMT): CMT2A2
Charcot-Marie-Tooth Disease (CMT): CMT2B
Charcot-Marie-Tooth Disease (CMT): CMT2C
Charcot-Marie-Tooth Disease (CMT): CMT2D
Charcot-Marie-Tooth Disease (CMT): CMT2DD
Charcot-Marie-Tooth Disease (CMT): CMT2E
Charcot-Marie-Tooth Disease (CMT): CMT2F
Charcot-Marie-Tooth Disease (CMT): CMT2I
Charcot-Marie-Tooth Disease (CMT): CMT2J
Charcot-Marie-Tooth Disease (CMT): CMT2K

Charcot-Marie-Tooth Disease (CMT): CMT2L
Charcot-Marie-Tooth Disease (CMT): CMT2N
Charcot-Marie-Tooth Disease (CMT): CMT2O
Charcot-Marie-Tooth Disease (CMT): CMT2P
Charcot-Marie-Tooth Disease (CMT): CMT2Q
Charcot-Marie-Tooth Disease (CMT): CMT2R
Charcot-Marie-Tooth Disease (CMT): CMT2T
Charcot-Marie-Tooth Disease (CMT): CMT2V
Charcot-Marie-Tooth Disease (CMT): CMT2W
Charcot-Marie-Tooth Disease (CMT): CMT2X
Charcot-Marie-Tooth Disease (CMT): CMT2Z
Charcot-Marie-Tooth Disease (CMT): CMT3/Dejerine-Sottas syndrome (subtype unknown)
Charcot-Marie-Tooth Disease (CMT): CMT3/Dejerine-Sottas syndrome A (DSSA)
Charcot-Marie-Tooth Disease (CMT): CMT3/Dejerine-Sottas syndrome B (DSSB)
Charcot-Marie-Tooth Disease (CMT): CMT3/Dejerine-Sottas syndrome C (DSSC)
Charcot-Marie-Tooth Disease (CMT): CMT3/Dejerine-Sottas syndrome E (DSSE)
Charcot-Marie-Tooth Disease (CMT): CMT4A
Charcot-Marie-Tooth Disease (CMT): CMT4B1
Charcot-Marie-Tooth Disease (CMT): CMT4B2
Charcot-Marie-Tooth Disease (CMT): CMT4B3
Charcot-Marie-Tooth Disease (CMT): CMT4C
Charcot-Marie-Tooth Disease (CMT): CMT4D
Charcot-Marie-Tooth Disease (CMT): CMT4E/congenital hypomyelinating myopathy
Charcot-Marie-Tooth Disease (CMT): CMT4F
Charcot-Marie-Tooth Disease (CMT): CMT4G
Charcot-Marie-Tooth Disease (CMT): CMT4H
Charcot-Marie-Tooth Disease (CMT): CMT4J
Charcot-Marie-Tooth Disease (CMT): CMT4K
Charcot-Marie-Tooth Disease (CMT): CMTX1
Charcot-Marie-Tooth Disease (CMT): CMTX4
Charcot-Marie-Tooth Disease (CMT): CMTX5
Charcot-Marie-Tooth Disease (CMT): CMTX6
Charcot-Marie-Tooth Disease (CMT): Dominant intermediate CMT
Charcot-Marie-Tooth Disease (CMT): Hereditary neuropathy with liability to pressure palsies (HNPP)
Congenital Muscular Dystrophy (CMD): Bethlem myopathy
Congenital Muscular Dystrophy (CMD): Bethlem myopathy 2
Congenital Muscular Dystrophy (CMD): CMD (subtype unknown)
Congenital Muscular Dystrophy (CMD): CMD, dynamin2 related
Congenital Muscular Dystrophy (CMD): CMD, integrin related
Congenital Muscular Dystrophy (CMD): CMD, LMNA related
Congenital Muscular Dystrophy (CMD): CMD, telethonin related
Congenital Muscular Dystrophy (CMD): CMD with hypoglycosylation of dystroglycan
Congenital Muscular Dystrophy (CMD): CMD with hypoglycosylation of dystroglycan, type A9
Congenital Muscular Dystrophy (CMD): Fukuyama CMD
Congenital Muscular Dystrophy (CMD): Merosin-Deficient CMD type 1A (MDC1A)/CMD with merosin deficiency
Congenital Muscular Dystrophy (CMD): Muscle-Eye-Brain disease (MEB)
Congenital Muscular Dystrophy (CMD): Rigid Spine Syndrome, FHL1 related
Congenital Muscular Dystrophy (CMD): Rigid Spine Syndrome, SEPN1 related
Congenital Muscular Dystrophy (CMD): Ullrich CMD 2
Congenital Muscular Dystrophy (CMD): Ullrich syndrome/UCMD
Congenital Muscular Dystrophy (CMD): Walker-Warburg Syndromes (WWS)
Congenital Myasthenic Syndrome (CMS): Acetylcholine receptor deficiency
Congenital Myasthenic Syndrome (CMS): ChAT CMS

Congenital Myasthenic Syndrome (CMS): CMS with Acetylcholine receptor deficiency
Congenital Myasthenic Syndrome (CMS): COLQ CMS
Congenital Myasthenic Syndrome (CMS): Congenital Myasthenic Syndrome (subtype unknown)
Congenital Myasthenic Syndrome (CMS): Congenital myasthenic syndrome related to GMPPB
Congenital Myasthenic Syndrome (CMS): Congenital myasthenic syndrome related to RPH3A
Congenital Myasthenic Syndrome (CMS): Congenital myasthenic syndrome related to SLC24A1
Congenital Myasthenic Syndrome (CMS): Congenital myasthenic syndrome with nephrotic syndrome
Congenital Myasthenic Syndrome (CMS): Congenital myasthenic syndrome (subtype unknown)
Congenital Myasthenic Syndrome (CMS): Escobar syndrome (multiple pterygium syndrome)
Congenital Myasthenic Syndrome (CMS): Familial limb girdle myasthenia with tubular aggregates, Familial limb girdle myasthenic syndrome with tubular aggregates
Congenital Myasthenic Syndrome (CMS): Fast channel CMS
Congenital Myasthenic Syndrome (CMS): Fast channel syndromes
Congenital Myasthenic Syndrome (CMS): MuSK CMS
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 5
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 6, presynaptic
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 7, presynaptic
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 8, with pre and postsynaptic deficits
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 9
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 10
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 12, with tubular aggregates
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 13, with tubular aggregates
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 14, with tubular aggregates
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 15, without tubular aggregates
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 16
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 17
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 18
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 19
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 20
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 21, presynaptic
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, congenital, 22
Congenital Myasthenic Syndrome (CMS): Myasthenic syndrome, with plectin defect
Congenital Myasthenic Syndrome (CMS): Presynaptic congenital myasthenic syndrome
Congenital Myasthenic Syndrome (CMS): Presynaptic congenital myasthenic syndrome related to MUNC13-1
Congenital Myasthenic Syndrome (CMS): RAPSN CMS
Congenital Myasthenic Syndrome (CMS): Slow channel CMS
Congenital Myasthenic Syndrome (CMS): Slow channel syndromes
Core Myopathy: Central Core Disease (CCD)
Core Myopathy: Core Disease (subtype unknown)
Core Myopathy: Multiminicore disease (MmD), classic
Core Myopathy: Multiminicore disease (MmD) w/external opthalmoplegia
Core Myopathy: Transient Multiminicore myopathy/recessive CCD
Disorders of lipid metabolism: Carnitine Deficiency, primary systemic/CDSP
Disorders of lipid metabolism: Carnitine Palmitoyltransferase Deficiency
Distal Myopathy: Desmin associated distal myopathy
Distal Myopathy: Distal Anoctaminopathy/Early Onset Calf Distal Myopathy
Distal Myopathy: Distal Myopathy (subtype unknown)
Distal Myopathy: Distal myopathy, Alpha-B crystallin related
Distal Myopathy: Distal myopathy, Dynamin 2 related/Distal ABC-filaminopathy
Distal Myopathy: Distal myopathy, Filamin C related/Distal ABC-filaminopathy
Distal Myopathy: Distal myopathy with caveolin defect

Distal Myopathy: Distal myopathy with myotilin defect
Distal Myopathy: Distal myopathy with nebulin defect
Distal Myopathy: Distal Myopathy with VCP defect
Distal Myopathy: Early onset distal myopathy w/KLHL9 defect
Distal Myopathy: GNE Myopathy/Hereditary Inclusion Body Myositis
Distal Myopathy: Laing distal myopathy/MPD1
Distal Myopathy: Late-onset distal myopathy/Markesbery-Griggs/ZASPopathy
Distal Myopathy: Miyoshi Myopathy/Distal Recessive Myopathy
Distal Myopathy: Myopathy, distal, with rimmed vacuoles
Distal Myopathy: Nonaka Myopathy/Hereditary Inclusion Body Myopathy (HIBM)/GNE Myopathy/Distal Mopathy with Rimmed Vacuoles (DMRV)/IBM2/ hIBM2
Distal Myopathy: Udd myopathy/Tibial muscular dystrophy (TMD)
Distal Myopathy: Vocal cord and pharyngeal distal myopathy (VCPDM)/MPD2
Distal Myopathy: Welander distal myopathy (WDM)
Duchenne/Becker Muscular Dystrophy (BMD): Becker muscular dystrophy
Duchenne/Becker Muscular Dystrophy (DBMD): Becker muscular dystrophy (BMD)
Duchenne/Becker Muscular Dystrophy (DBMD): Duchenne muscular dystrophy (DMD)
Duchenne/Becker Muscular Dystrophy (DBMD): Manifesting Carrier
Duchenne/Becker Muscular Dystrophy (DMD): Duchenne muscular dystrophy
Emery-Dreifuss Muscular Dystrophy (EDMD): EDMD (subtype unknown)
Emery-Dreifuss Muscular Dystrophy : EDMD (subtype unknown)
Emery-Dreifuss Muscular Dystrophy : EDMD1, X-linked, type 1
Emery-Dreifuss Muscular Dystrophy : EDMD2, autosomal dominant
Emery-Dreifuss Muscular Dystrophy : EDMD3, autosomal recessive
Emery-Dreifuss Muscular Dystrophy : EDMD4/Nesprin-1 related muscular dystrophy
Emery-Dreifuss Muscular Dystrophy : EDMD5/Nesprin-2 related muscular dystrophy
Emery-Dreifuss Muscular Dystrophy : EDMD6, X-linked, type 2
Emery-Dreifuss Muscular Dystrophy : EDMD7/LUMA related muscular dystrophy
Endocrine myopathy: Hyperthyroid Myopathy
Endocrine myopathy: Hypothyroid Myopathy
Facioscapulohumeral Muscular Dystrophy (FSHD): FSHD (subtype unknown)
Facioscapulohumeral Muscular Dystrophy (FSHD): FSHD, type 1
Facioscapulohumeral Muscular Dystrophy (FSHD): FSHD, type 2
Friedreich's Ataxia (FA): Friedreich's Ataxia
Giant Axonal Neuropathy (GAN): Giant Axonal Neuropathy
Glycogen Storage Diseases (GSD): Debrancher Enzyme Deficiency/GSD IIIa, Glycogenosis type III
Glycogen Storage Diseases (GSD): Enolase deficiency/GSD XIII
Glycogen Storage Diseases (GSD): Glycogen Branching Enzyme/GSD IV/adult polyglucosa body disease
Glycogen Storage Diseases (GSD): Glycogenosis type XIV/GSD XIV/Glycogenosis type XIV
Glycogen Storage Diseases (GSD): Glycogen storage disease of heart, lethal congenital
Glycogen Storage Diseases (GSD): Glycogen synthase 1 deficiency/GSD O
Glycogen Storage Diseases (GSD): GSD (subtype unknown)
Glycogen Storage Diseases (GSD): Gycogenin 1 deficiency/GSD XV/ Glycogenosis type XV
Glycogen Storage Diseases (GSD): Lactate Dehydrogenase Deficiency/GSD XI
Glycogen Storage Diseases (GSD): Lysosome-associated membrane protein 2 deficiency/Danon Disease/LAMP2 deficiency/GSD IIB/Vacuolar X-linked Cardiomyopathy
Glycogen Storage Diseases (GSD): McArdle's/Phosphorylase deficiency/myophosphorylase deficiency/GSD V/Glycogenosis type V
Glycogen Storage Diseases (GSD): Muscle phosphorylase kinase deficiency/GSD IXd
Glycogen Storage Diseases (GSD): Phosphofructokinase Deficiency/Tauri's/GSD VII, Glycogenosis type VII
Glycogen Storage Diseases (GSD): Phosphoglycerate Kinase Deficiency/Glycogenosis type IX
Glycogen Storage Diseases (GSD): Phosphoglycerate Mutase Deficiency/GSD X, Glycogenosis type X
Glycogen Storage Diseases (GSD): Polyglucosan storage myopathy

Glycogen Storage Diseases (GSD): Pompe/Acid Maltase Deficiency (AMD)/GSD II/ Glycogenosis type II

Hyperkalemic Periodic Paralysis : Hyperkalemic Periodic Paralysis

Hypokalemic Periodic Paralysis (HypoKPP): Hypokalemic Periodic Paralysis (subtype unknown)

Hypokalemic Periodic Paralysis (HypoKPP): HypoKPP Type 1

Hypokalemic Periodic Paralysis (HypoKPP): HypoKPP Type 2

Hypokalemic Periodic Paralysis (HypoKPP): HypoKPP Type 3

Hypokalemic Periodic Paralysis (HypoKPP): Thyrotoxic HypoKPP

Inflammatory myopathy: Dermatomyositis

Inflammatory myopathy: Inclusion Body Myositis

Inflammatory myopathy: Polymyositis

Inflammatory myopathy: VCP/Myopathy/IBMPFD

Lambert-Eaton Syndrome (LEMS): Lambert-Eaton Syndrome/Lambert-Eaton Myasthenic Syndrome

Limb-Girdle Muscular Dystrophy (LGMD): LGMD (subtype unknown)

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1A

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1B

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1C

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1D

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1E

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1F

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1G

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-1H

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2A

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2B

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2C

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2D

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2E

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2F

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2G

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2H

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2I

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2J

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2K

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2L

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2M

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2N

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2O

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2P

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2Q

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2R

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2S

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2T

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2U

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2V

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2W

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2X

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2Y

Limb-Girdle Muscular Dystrophy (LGMD): LGMD-2Z

Miscellaneous Congenital Myopathies: Cap myopathy

Miscellaneous Congenital Myopathies: Compton-North congenital myopathy

Miscellaneous Congenital Myopathies: Congenital Myopathy (subtype unknown)

Miscellaneous Congenital Myopathies: Congenital myopathy related to PTPLA

Miscellaneous Congenital Myopathies: Congenital myopathy with fatal cardiomyopathy

Miscellaneous Congenital Myopathies: Congenital myopathy with Fiber type disproportion

Miscellaneous Congenital Myopathies: Congenital skeletal myopathy and fatal cardiomyopathy
Miscellaneous Congenital Myopathies: Myosin IIA myopathy
Miscellaneous Congenital Myopathies: Myosin storage myopathy and cardiomyopathy
Miscellaneous Congenital Myopathies: Myosin storage myopathy/Hyaline body myopathy
Miscellaneous metabolic disorders: Myoadenylate Deaminase Deficiency/Myopathy due to AMPD1 Deficiency/
Adenosine Monophosphate Deaminase 1 Deficiency
Miscellaneous Myopathy: Edstrom myopathy/Hereditary myopathy with early respiratory failure
Miscellaneous Myopathy: HyperCKemia
Miscellaneous Myopathy: Inclusion body myopathy associated with Paget disease of bone and FTD (IBMPFD)
Miscellaneous Myopathy: Inclusion body myopathy with early-onset Paget disease w/o w/o FTD
Miscellaneous Myopathy: Myopathy with excessive autophagia
Miscellaneous Myopathy: Myopathy with exercise intolerance, Swedish
Miscellaneous Myopathy: Reducing body myopathy
Miscellaneous Myopathy: Scapuloperoneal Myopathy/XPMD
Miscellaneous Myopathy: Tubular aggregate myopathy
Mitochondrial Myopathy: Kearns-Sayre syndrome (KSS)
Mitochondrial Myopathy: Leigh's / Leigh Disease/ Leigh syndrome/Subacute necrotizing encephalomyopathy
Mitochondrial Myopathy: Mitochondrial DNA depletion syndrome
Mitochondrial Myopathy: Mitochondrial encephalomyopathy lactic acidosis and stroke-like episodes (MELAS)
Mitochondrial Myopathy: Mitochondrial Myopathy
Mitochondrial Myopathy: Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)/myoneurogastrointestinal
encephalopathy syndrome
Mitochondrial Myopathy: Myoclonus epilepsy with ragged red fibers (MERRF)
Mitochondrial Myopathy: Neuropathy, ataxia and retinitis pigmentosa (NARP)
Mitochondrial Myopathy: Pearson syndrome
Mitochondrial Myopathy: PEOA1
Mitochondrial Myopathy: PEOA2
Mitochondrial Myopathy: PEOA3
Mitochondrial Myopathy: PEOA4
Mitochondrial Myopathy: PEOA5
Mitochondrial Myopathy: PEOB1
Mitochondrial Myopathy: PEOB2
Mitochondrial Myopathy: PEOB3
Mitochondrial Myopathy: PEOB4
Mitochondrial Myopathy: Progressive external ophthalmoplegia (PEO)/Chronic Progressive external ophthalmoplegia
(CPEO)
Myasthenia Gravis (MG): Acetylcholine receptor/AChR antibody-mediated MG
Myasthenia Gravis (MG): Infantile MG
Myasthenia Gravis (MG): Inherited - familial MG
Myasthenia Gravis (MG): LRP4 antibody-mediated MG
Myasthenia Gravis (MG): MuSk antibody-mediated MG
Myasthenia Gravis (MG): Myasthenia Gravis (subtype unknown)
Myasthenia Gravis (MG): Ocular MG
Myasthenia Gravis (MG): Seronegative MG
Myofibrillar Myopathy (MFM): MFM (subtype unknown)
Myofibrillar Myopathy (MFM): MFM, alpha-B crystallin related/MFM2
Myofibrillar Myopathy (MFM): MFM, BAG3 related
Myofibrillar Myopathy (MFM): MFM, desmin related/Desmin-related myopathy/ Desminopathy
Myofibrillar Myopathy (MFM): MFM, filamin-C related/Filaminopathy/MFM5
Myofibrillar Myopathy (MFM): MFM, myotillin related/Myotilinopathy/MFM3
Myofibrillar Myopathy (MFM): MFM, SEPN related/Desmin-related myopathy with Mallory bodies
Myofibrillar Myopathy (MFM): MFM LDB3, ZASP related/Zaspopathy/MFM4
Myofibrillar Myopathy (MFM): Myopathy microfibrillar type 7/MFM7

Myofibrillar Myopathy (MFM): Spheroid body myopathy
Myotonia Congenita (MC): MC, dominant (Thomsen myotonia)
Myotonia Congenita (MC): MC, recessive (Becker myotonia)
Myotonia Congenita (MC): Myotonia Congenita (subtype unknown)
Myotonic Dystrophy (DM): DM1
Myotonic Dystrophy (DM): DM2
Myotonic Dystrophy (DM): Myotonic Dystrophy (subtype unknown)
Nemaline Myopathy (NEM): NEM1
Nemaline Myopathy (NEM): NEM2
Nemaline Myopathy (NEM): NEM3
Nemaline Myopathy (NEM): NEM4
Nemaline Myopathy (NEM): NEM5
Nemaline Myopathy (NEM): NEM6
Nemaline Myopathy (NEM): NEM7
Nemaline Myopathy (NEM): NEM8
Nemaline Myopathy (NEM): NEM9
Nemaline Myopathy (NEM): NEM10
Nemaline Myopathy (NEM): Nemaline Myopathy (subtype unknown)
Oculopharyngeal Muscular Dystrophy (OPMD): OPMD
Other: Covered diagnosis not listed
Other: Diagnosis not listed / Unknown
Other: Diagnosis unknown
Other: Non-covered diagnosis
Paramyotonia Congenita (PMC): Paramyotonia Congenita
Potassium-Aggravated Myotonia: Potassium-Aggravated Myotonia/myotonia fluctuans, myotonia congenita acetazolamide-responsive
Spinal Bulbar Muscular Atrophy (SBMA): Spinal Bulbar Muscular Atrophy/Kennedy Disease
Spinal Muscular Atrophy (SMA): Distal SMA
Spinal Muscular Atrophy (SMA): Distal SMA/dHMN (distal hereditary motor neuropathy)
Spinal Muscular Atrophy (SMA): Distal SMA/HMN2A (hereditary motor neuropathy 2a)
Spinal Muscular Atrophy (SMA): Distal SMA/HMN2B (hereditary motor neuropathy 2b)
Spinal Muscular Atrophy (SMA): Distal SMA/HMN2C (hereditary motor neuropathy 2c)
Spinal Muscular Atrophy (SMA): Distal SMA/HMN5 (hereditary motor neuropathy 5)
Spinal Muscular Atrophy (SMA): Distal SMA/HMN7B (hereditary motor neuropathy 7b)
Spinal Muscular Atrophy (SMA): Distal SMA/X-linked
Spinal Muscular Atrophy (SMA): Distal X-linked infantile SMA/SMAX2
Spinal Muscular Atrophy (SMA): SMA (subtype unknown)
Spinal Muscular Atrophy (SMA): SMA1/Werdnig Hoffman Disease/Severe SMA
Spinal Muscular Atrophy (SMA): SMA2/Intermediate SMA
Spinal Muscular Atrophy (SMA): SMA3/Kugelberg Welander Disease/Mild SMA
Spinal Muscular Atrophy (SMA): SMA4/Adult SMA
Spinal Muscular Atrophy (SMA): SMA with respiratory distress (SMARD1)