

Neuromuscular Diseases Included in MDA's Programs

Group/Type	Usual Age of Onset	Disease Characteristics
Muscular Dystrophies		
Myotonic	Birth to adulthood	Weakness of all muscle groups accompanied by delayed relaxation of muscles after contraction. Affects face, feet, hands and neck first. Progression is slow, sometimes spanning 50 to 60 years.
Duchenne	2 to 6 years	General muscle weakness and wasting, affecting pelvis, upper arms and upper legs first. Duchenne progresses slowly, yet eventually involves all voluntary muscles. Survival is rare beyond the late 20s.
Becker	2 to 16 years	Symptoms almost identical to Duchenne yet less severe. Affects pelvis, upper arms and upper legs first. Becker progresses more slowly than Duchenne and survival runs well into middle age.
Limb-Girdle	Late childhood to middle age	Weakness and wasting, affecting shoulder girdle and pelvic girdle first. Disease usually progresses slowly. Variable cardiopulmonary complications may occur in later stages.
Facioscapulohumeral	Childhood to early adulthood	Facial muscle weakness, with weakness and wasting of the shoulders and upper arms. Progressing slowly with some periods of rapid deterioration, disease may span many decades.
Congenital	At birth or infancy	Generalized muscle weakness, with possible joint contractures resulting from shortening of muscles. Disease progresses very slowly. Weakness is variable.
Oculopharyngeal	Early adulthood to middle age	First affects muscles of eyelids and throat. While progression is slow, weakening of throat muscles in time causes swallowing difficulties.
Distal	Early adulthood to middle age	Weakness and wasting of muscles of the hands, forearms and lower legs. Progresses slowly and is rarely life-threatening.
Emery-Dreifuss	Childhood to early teens	Weakness and wasting of shoulder, upper arm and shin muscles. Joint contractures are common. Disease progresses slowly, with cardiac complications common.
Motor Neuron Diseases		
Amyotrophic Lateral Sclerosis (ALS)	Adulthood	Progressive wasting and weakness of all voluntary muscles, with cramps and muscle twitches common. ALS first affects legs, arms and/or throat muscles. Survival rarely exceeds five years after onset, without respiratory intervention.
Infantile Progressive Spinal Muscular Atrophy	Birth to 3 months	Generalized muscle weakness, weak cry, trouble swallowing and sucking, and breathing distress. Life span rarely exceeds age 2.

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Intermediate Spinal Muscular Atrophy	6 months to 3 years	Weakness in arms, legs, upper and lower torso, often with skeletal deformities. Disease progresses rapidly. Respiratory problems can shorten life.
Juvenile Spinal Muscular Atrophy	1 to 15 years	Weakness in leg, hip, shoulder, arm and respiratory muscles. Disease progresses slowly. Life span is unaffected.
Adult Spinal Muscular Atrophy	18 to 50 years	Generalized muscle weakness with muscle twitches common. Disease progression varies.
Spinal-Bulbar Muscular Atrophy	15 to 60 years	Weakness of limb muscles, especially legs, and of muscles involved in talking, chewing and swallowing. Occurs in men. Slowly progressive over decades.

Inflammatory Myopathies

Polymyositis	Childhood to late adulthood	Weakness of neck and limb muscles, sometimes with pain. Disease severity and progression vary by individual. Sometimes associated with malignancy. Often responds to drug therapy.
Dermatomyositis	Childhood to late adulthood	Weakness of neck and limb muscles, often with pain. Skin rash typically affects cheeks, eyelids, neck, chest and limbs. Disease severity and progression vary. Sometimes associated with malignancy. Often responds to drug therapy.
Inclusion-Body Myositis	After age 50	Weakness of arms, legs and hands, especially thighs, wrists and fingers. Sometimes involves swallowing muscles. Slowly progressive. More common in men than women.

Diseases of the Neuromuscular Junction

Myasthenia Gravis	Childhood to adulthood	Weakness and fatigability of muscles of the eyes, face, neck, throat, limbs and/or trunk. Weakness may fluctuate. Disease progression varies. Drug therapy and/or removal of thymus gland often effective.
Lambert-Eaton Myasthenic Syndrome	Adulthood	Weakness and fatigue of hip and leg muscles with aching back and thigh muscles common. Lung tumor is often present. Progression varies with success of drug therapy and treatment of any malignancy.
Congenital Myasthenic Syndromes	Infancy or childhood, can be later	Generalized weakness and fatigability of voluntary muscles, including those controlling eye movement, swallowing and breathing. Varies in severity, and weakness can fluctuate.

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<i>Diseases of the Peripheral Nerves</i>		
Charcot-Marie-Tooth Disease	Childhood to young adulthood	Weakness and atrophy of muscles of hands and lower legs, with foot deformities and some loss of sensation in feet. Disease progression usually slow.
Friedreich's Ataxia	Childhood to adolescence	Impairment of balance and limb coordination, muscle weakness and loss of sensation. Severity and progression of disorder vary. Often associated with diabetes and heart disease.
Dejerine-Sottas Disease	Infancy to early childhood	Delayed development of motor skills. Muscle weakness affects hands and legs and may involve impairment of sensation. Severity and progression of disease vary.
<i>Metabolic Diseases of Muscle</i>		
Phosphorylase Deficiency	Childhood to adulthood	Muscle cramps often occur after exercise. Intense exercise can cause muscle destruction and possible damage to kidneys. Reducing strenuous exercise can lessen severity.
Acid Maltase Deficiency	Infancy to adulthood	For infants, disease is generalized and severe with heart, liver and tongue enlargement common. Adult form involves weakness of upper arms, legs, trunk and respiratory muscles. Progression varies.
Phosphofructokinase Deficiency	Childhood to adulthood	Muscle fatigue which upon exercise can lead to severe cramps, nausea, vomiting, muscle damage and discoloration of urine. Progression varies.
Debrancher Enzyme Deficiency	Early childhood to adulthood	Generalized weakness and muscle wasting. Heart involvement and enlarged liver may occur with infantile form. Episodes of low blood sugar are common. Variable progression. Muscle symptoms may be delayed until teens or adulthood.
Mitochondrial Myopathy	Early childhood to adulthood	Generalized muscle weakness with droopy eyelids and inability to walk. Brain is often involved, with seizures, deafness, loss of balance and vision, and retardation common. Progression and severity vary widely.
Carnitine Deficiency	Early childhood	Varied weakness of shoulder, hip, face and neck muscles. Progression varies and carnitine supplementation can be effective.
Carnitine Palmityl Transferase Deficiency	Young adulthood	Inability to sustain moderate prolonged exercise. Prolonged exercise and/or fasting can cause severe muscle destruction with urine discoloration and kidney damage. Severity varies.
Phosphoglycerate Kinase Deficiency	Childhood to adolescence	Muscle pain and weakness, with muscle damage and urine discoloration possible after vigorous exercise. Severity varies.

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Phosphoglycerate Mutase Deficiency	Childhood to adulthood	Muscular pain, cramps, muscle damage and urine discoloration possible following intense exercise of brief duration. Severity varies.
Lactate Dehydrogenase Deficiency	Childhood to adolescence	Intolerance of intense exercise with muscle damage and urine discoloration possible following strenuous physical activity. Severity of disorder varies.
Myoadenylate Deaminase Deficiency	Early adulthood to middle age	Muscle fatigue and weakness during and after exertion, with muscle soreness or cramping. Condition is nonprogressive and severity varies.

Myopathies Due to Endocrine Abnormalities

Hyperthyroid Myopathy	Childhood to adulthood	Weakness in upper arm and upper leg muscles with some evidence of wasting. Usually improves with treatment of underlying thyroid condition.
Hypothyroid Myopathy	Childhood to adulthood	Weakness of arm and leg muscles. Stiffness, muscle pain and cramps common. Usually improves with treatment of underlying thyroid condition.

Other Myopathies

Myotonia Congenita	Early childhood	Muscle stiffness and cramps after periods of rest. Condition causes discomfort but is not life-threatening.
Paramyotonia Congenita	Childhood to early adulthood	Poor or difficult relaxation of muscles, which may worsen after exposure to cold or exercise. Often associated with hyperkalemic periodic paralysis. Condition causes discomfort but isn't life-threatening.
Central Core Disease	Infancy to childhood	Delayed motor development. Hip displacement is not uncommon. Condition can be stable to slowly progressive.
Nemaline Myopathy	Infancy to childhood	Delayed motor development. Weakness of arm, leg, trunk, face and throat muscles. Respiratory involvement common. Severity and progression vary.
Myotubular Myopathy	Infancy	Drooping of upper eyelids, facial weakness. Weakness of the limbs and trunk muscles. Patients almost always have no reflexes. Respiratory involvement is possible. Disease progresses slowly.
Periodic Paralysis – Hypokalemic – Hyperkalemic	Childhood to adulthood	Episodes of generalized muscle weakness. Hyperkalemic type may be associated with paramyotonia congenita. Frequency of attacks and severity vary. May respond to drug therapy.