Muscular Dystrophy Australia supports people with any one of more than 60 different neuromuscular conditions, about half of which are classified as muscular dystrophies. These conditions affect approximately 1 in every 1000 people.

The muscle conditions can be divided into nine broad categories: their main features are described in the table below. All of the conditions have in common that they cause muscle weakness which has a profound effect on people’s lives. However, this is where the similarity ends. Some neuromuscular conditions are so severe that babies die within the first few years of life, whereas others only affect people in old age and don’t cause severe disability. Most of the conditions have a genetic cause and are passed down through families (inherited) but sometimes the genetic change occurs spontaneously in an individual without a family history of muscle disease. Other conditions are autoimmune or the cause is unknown or poorly understood.

The source of the muscle weakness within the body differs too. For example, muscular dystrophy is primarily a disease of muscle, often caused by the lack of an important structural protein and as a result, the muscles are fragile and easily damaged. In other types of muscular dystrophy such as facioscapulohumeral muscular dystrophy (FSHD) the genetic change results in the production of a substance that is toxic to muscle.

For some other neuromuscular conditions the muscle weakness originates in the nerves that control the muscles. For example, amyotrophic lateral sclerosis (ALS) and spinal muscular atrophy (SMA) affect the motor neurons in the spinal cord. Others, such as Charcot-Marie-Tooth disease affect the peripheral nerves which carry signals from the spinal cord to the limbs. A structure called the ‘neuromuscular junction’ that connects the nerves to the muscles is affected in some conditions, for example myasthenia gravis.

Each neuromuscular condition causes a characteristic pattern of muscle weakness: it could be just the legs or the muscles around the eyes and throat. The most severe conditions affect all the muscles of the body, including the heart and the muscles used for breathing. In most cases intelligence is not affected. Some conditions such as myotonic dystrophy affect many parts of the body as well as the muscles. This results in very diverse care needs depending on the condition and the stage of progression. Often the management of symptoms is complex requiring input from a wide variety of health care professionals.

There are no specific treatments for most neuromuscular conditions but considerable research efforts are working towards developing them and encouragingly, many clinical trials are now starting. Treatments, such as gene therapy to specifically target the underlying cause are being developed for some conditions, as well as more generalised treatments which aim to reduce the severity of symptoms by, for example, boosting muscle regeneration. Read about the research MDA funds.
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A brief description of some muscular dystrophies and related neuromuscular conditions

Muscular Dystrophies:

**BECKER MUSCULAR DYSTROPHY**

Age of onset: 2 to 16 years. Symptoms are almost identical to Duchenne yet less severe. Affects pelvis, upper arms and upper legs. Becker progresses more slowly than Duchenne and survival runs well into middle age.

**CONGENITAL MUSCULAR DYSTROPHY**

Age of onset: at birth. Generalised muscle weakness, with possible joint deformities from shortening of muscles. There are at least 30 different types - many types progress very slowly; some shorten life span.

**DUCHENNE MUSCULAR DYSTROPHY**

Age of onset: 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. A wheelchair is required by about age 8 to 11 years and the condition is severe enough to shorten life expectancy. With high standards of medical care survival is often into the 30s.

**DISTAL MUSCULAR DYSTROPHY**

Age of onset: 40 to 60 years. A group of conditions that cause weakness and wasting of muscles of the hands, forearms and lower legs. Progresses slowly and rarely leads to total incapacity.

**EMERY-DREIFUSS MUSCULAR DYSTROPHY**

Age of onset: childhood to early teens. Weakness and wasting of shoulder, upper arm and shin muscles. Joint deformities are common. Disease progresses slowly, yet sudden death can result from cardiac complications.

**FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY**

Age of onset: teens to early adulthood. There is also an infantile-onset form. Muscles of the face, shoulder blades and upper arms are among the most affected but other muscles are usually affected. Progresses slowly with some periods of rapid deterioration, disease may span many decades. Most people with the disease have a normal life span.

**LIMB-GIRDLE MUSCULAR DYSTROPHY**

Age of onset: late childhood to middle age. Weakness and wasting, affecting muscles around the shoulders and hips first. There are more than 20 different subtypes - some progress to loss
of walking ability within a few years and cause serious disability, while others progress very slowly over many years and cause minimal disability.

**OCULOPHARYNGEAL MUSCULAR DYSTROPHY**

Age of onset: 40 to 70 years. First affects muscles of the eyelids and throat. Progression is slow, but weakening of throat muscles causes inability to swallow and mobility can be affected later on.

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**Myotonic disorders**

**MYOTONIC DYSTROPHY**

Age of onset: 20 to 40 years. Muscle weakness is accompanied by myotonia (delayed relaxation of muscles after contraction) and by a variety of symptoms that affect other parts of the body including the heart, eyes and brain. Muscle weakness affects face, feet, hands and neck first. Progression is slow, sometimes spanning 50 to 60 years. More severe infantile and childhood forms also exist.

**MYOTONIA CONGENITA**

Age of onset: infancy to childhood. Muscle stiffness and difficulty in moving after periods of rest. With exercise, muscle strength and movement return to normal. Condition causes discomfort throughout life but is not life-threatening.

**PARAMYOTONIA CONGENITA**

Age of onset: adulthood. Poor or difficult relaxation of muscle which usually worsens after repeated use or exercise. Condition causes discomfort throughout life but is not life-threatening.

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**Spinal Muscular Atrophies (SMA):**

**SMA TYPE 1 (INFANTILE PROGRESSIVE)**

Age of onset: birth to 6 months. Generalised muscle weakness, weak cry, trouble breathing, swallowing and sucking. Do not reach the developmental milestone of being able to sit up unassisted. Life span rarely exceeds age of two.

**SMA TYPE 2 (INTERMEDIATE)**

Age of onset: 7 to 18 months. Weakness in arms, legs and lower torso, often with skeletal deformities. Children learn to sit unassisted but do not stand or walk independently. Although respiratory complications are a constant threat, children with type 2 SMA usually live to young adulthood and many live longer.

**SMA TYPE 3 (JUVENILE)**
Age of onset: 1 to 15 years. Weakness in leg, hip, shoulder, arm and respiratory muscles. Children learn to stand and walk but some lose the ability to walk in adolescence, others walk well into their adult years. Life span is unaffected.

**SMA ADULT SPINAL MUSCULAR ATROPHY**

Age of onset: 18 to 50 years. Weakness in the tongue, hands or feet which slowly spreads to other parts of the body. A relatively mild form of spinal muscular atrophy, it has little impact on life expectancy.

**Inflammatory Myopathies:**

**DERMATOMYOTISIS**

Age of onset: childhood to 60 years. Inflammatory condition of the muscles that mostly affects the shoulders, upper arms, pelvis and thighs. Resulting disability is very variable. Other symptoms include a muscle pain, rash, fever, malaise and weight loss. Usually responds to treatment with corticosteroid and other immune suppressing drugs.

**POLYMYOSITIS**

Age of onset: mostly 30 to 50. Symptoms are similar to dermatomyositis except usually there is no rash or muscle pain. Disease severity and progression is very variable. More women than men are affected.

**INCLUSION BODY MYOSITIS**

Age of onset: over 50. A slowly progressive condition causing weakness primarily in the quadriceps and forearm muscles. Difficulty with stairs, getting out of a chair and a poor grip are common problems. Swallowing muscles are sometimes affected. In general patients do not die of the disease, but most meet with some degree of disability as the disease progresses.

**Neuropathies (Diseases of Peripheral Nerve):**

**CHARCOT-MARIE-TOOTH DISEASE**

Age of onset: teens to 20 years (occasionally in childhood or infancy). Damage to the peripheral nerves causes muscle weakness and wasting, and some loss of sensation, in the extremities of the body: the feet, the lower legs, the hands and the forearms. There are several different types of CMT and disease progression varies. Dejerine-Sottas Disease is a severe form.

**HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSYSES (HNPP)**

Age of onset: 20 to 40. Recurrent episodes of numbness, tingling, and/or loss of muscle function (palsy). An episode can last from several minutes to several months, but recovery is
usually complete. Repeated incidents, however, can cause permanent muscle weakness or loss of sensation.

**GUILLAIN-BARRE SYNDROME**

Age of onset: all ages. An autoimmune condition in which the nerves are attacked by the body's own immune system causing paralysis, muscular weakness and tingling sensations. The disorder can be mild, moderate or severe, with life support needed in the worst cases. Most people spontaneously recover, though some will be left with permanent disabilities.

**CHRONIC INFLAMMATORY DEMYELINATING POLYNEUROPATHY (CIPD)**

Age of onset: any age but more common in the 5th and 6th decades. An autoimmune condition which causes slowly progressing weakness and loss of feeling in the legs and arms. Numbness and tingling usually starts in the feet. Balance may also be affected. Severity varies widely among individuals. Some may have a bout of CIDP followed by spontaneous recovery, while others may have many bouts with partial recovery in between relapses. CIPD is treatable by suppressing the immune system although some individuals are left with some residual numbness or weakness.

**Disorders of the neuromuscular junction:**

**MYASTHENIA GRAVIS**

Age of onset: 30 to 50 years. An autoimmune condition where the junction between the nerve and muscle is damaged resulting in weakness of muscles of the eyes, face, neck, throat, limbs and/or trunk. Disease progression varies. Drug therapy and/or removal of thymus gland is often effective. There is a juvenile onset form of the condition.

**CONGENITAL MYASTHENIC SYNDROMES**

Age of onset: birth or early childhood. Genetic condition causing problems with the way messages are transmitted from the nerves to the muscles, causing weakness (myasthenia) and the muscles tire easily (fatigue). Muscle weakness varies depending on the type of genetic defect, so impact on mobility ranges from mild to severe.

**LAMBERT-EATON SYNDROME**

Age of onset: over 40 years. Weakness and fatigue of hip and thigh muscles is common. Lung tumour is often present. Progression varies with success of drug therapy and treatment of any malignancy.

**Other conditions affecting the nervous system**

**AMYOTROPIC LATERAL SCLEROSIS (ALS/MOTOR NEURONE DISEASE/LOU GEHRIG’S DISEASE)**
Age of onset: 35 to 65 years. Motor neurons (nerve cells that control muscle cells) are gradually lost. Wasting and weakness of all body muscles, with cramps and muscle twitches common. Progressive, ALS first affects legs, arms and/or throat muscles. Survival rarely exceeds five years after onset.

**FRIEDREICH'S ATAXIA**

Age of onset: 4 to 16 years. Inherited disease of the nervous system resulting in impairment of limb coordination, muscle weakness and loss of sensation. Severity and progression of disorder vary. Often associated with diabetes and heart disease.

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**Metabolic Diseases of the Muscle:**

**ACID MALTASE DEFICIENCY (Type II Glycogen storage disease, Pompe’s disease)**

Age of onset: Infancy to adulthood. For infants, disease is generalised and severe with heart, liver and tongue enlargement common. Adult form involves weakness of mid-body and respiratory muscles. Progression varies. Enzyme replacement therapy is available.

**CARNITINE DEFICIENCY**

Age of onset: Early childhood. Varied weakness of shoulder, hip, face and neck muscles. Often a secondary metabolic condition, progression varies and carnitine supplementation can be effective.

**CARNITINE PALMITYL TRANSFERASE DEFICIENCY**

Age of onset: young adulthood. Inability to sustain moderate prolonged exercise. Prolonged exercise and/or fasting can cause severe muscle damage with urine discoloration and kidney damage.

**DEBRANCHER ENZYME DEFICIENCY (Type III Glycogen storage disease, Cori’s disease)**

Age of onset: 1 year. General muscle weakness, poor muscle control and an enlarged liver with low blood sugar. Slow progression. Some patients do not experience muscular weakness until late teens or early adulthood.

**LACTATE DEHYDROGENASE DEFICIENCY**

Age of onset: childhood to adolescence. Intolerance of intense exercise with muscle damage and urine discoloration possible following strenuous physical activity. Severity of disorder varies and intense exercise should be avoided.

**MITOCHONDRIAL MYOPATHY**
Age of onset: birth to adulthood. Severe muscle weakness, flaccid neck muscles and inability to walk. Brain is often involved, with seizures, deafness, loss of balance and vision, and retardation common. Progression and severity vary.

**MYOADENYLATE DEAMINASE DEFICIENCY**

Age of onset: early adulthood to middle age. Muscle fatigue and weakness during and after exertion, with muscle soreness or cramping. Patients are often unable to attain previous performance levels yet condition is non-debilitating and non-progressive.

**PHOSPHORYLASE DEFICIENCY (Type V glycogen storage disease, McArdle disease)**

Age of onset: Adolescence. Low tolerance for exercise, with cramps often occurring after exercise. Intense exercise can cause muscle damage and possible damage to kidneys. Reducing strenuous exercise can lessen severity.

**PHOSPHOFRACTOKINASE DEFICIENCY (Type VII glycogen storage disease)**

Age of onset: Childhood. Muscle fatigue which upon exercise can lead to severe cramps, nausea, vomiting, muscle damage and discoloration of urine. Disease varies widely in severity and progression.

**PHOSPHOGLYCERATE KINASE DEFICIENCY**

Age of onset: Childhood to adulthood. Muscular pain, cramps, muscle damage and urine discoloration possible following intense exercise of brief duration. Severity varies and intense exercise should be avoided.

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**Other Myopathies:**

**BETHLEM MYOPATHY**

Age of onset: birth through to adulthood. Considered a type of congenital muscular dystrophy. In childhood the symptoms can be hypotonia (floppiness), muscle weakness, delayed motor milestones, talipes (clubfoot), torticollis (stiff neck) and contractures (tightness) in the ankles, hips, knees and elbows. The main symptoms in adults include tight tendons at the back of their ankles, as well as tightness of various other joints especially in the hands and mild muscle weakness.

**CENTRAL CORE DISEASE**

Age of onset: at birth or early infancy. Motor skill milestones are reached very slowly and hip displacement is not uncommon. Condition is disabling but not life-threatening.

**CONGENITAL FIBRE TYPE DISPROPORTION**
Age of onset: at birth or before age of 1. A rare type of myopathy characterized by hypotonia and mild to severe generalized muscle weakness.

**FIBRODYSPLASIA OSSIFICANS PROGRESSIVA (FOP)**

Age of onset: early childhood. An extremely rare disorder where a person's muscle and connective tissues, such as ligaments and tendons, are slowly replaced by bone. Starts in the person's shoulders and neck, progressing along their back, trunk, and limbs.

**HYPER/HYPO THYROID MYOPATHY -**

Age of onset: childhood to adulthood. Muscle disease caused by under or overproduction of thyroid hormones from the thyroid gland. Weakness in upper arm and upper leg muscles with some evidence of wasting. Severity depends on success in treating underlying thyroid condition.

**MINICORE (MULTICORE) MYOPATHY (multi-minicore disease)**

Age of onset: infancy or childhood. Rare myopathy with variable degrees of muscle weakness and wasting. There are different subtypes each with varying symptoms and severity. Most common type is characterized by spinal rigidity, early scoliosis and respiratory impairment. Other types may involve the muscle around the eyes, distal weakness and wasting or weakness around the hips. Weakness is static or only slowly progressive.

**MYOTUBULAR (CENTRONUCLEAR) MYOPATHY**

Age of onset: birth to early adulthood. There are three different types. In the most severe form babies are born floppy with breathing difficulties and the bones of their head are malformed. The most common type only affects males. More mildly affected people are often able to walk well into adulthood but do find themselves in a wheelchair in later life.

**NEMALINE MYOPATHY -**

Age of onset: at birth or early infancy. Hypotonia (poor muscle tone or floppiness) and weakness of arm, leg, trunk, face and throat muscles. In severe cases, children have marked respiratory weakness. Children rarely survive more than a few years, yet some live into teens.

**PERIODIC PARALYSIS - HYPOKALEMIC - HYPERKALEMIC -**

Age of onset: infancy to 30 years. Severe generalised weakness of legs and other muscle groups with periods of paralysis affecting arms, legs and neck. Severity varies by age of onset and success of drug therapy.

**If you have any questions, please contact us:**

**Email:** info@mda.org.au  
**Phone:** +61 3 9320 9555