LIST OF NEUROMUSCULAR CONDITIONS

Muscular Dystrophy South Australia provides a range of services to people living with a variety of neuromuscular conditions.
This is a list of some of the neuromuscular conditions for which Muscular Dystrophy SA provides support, but is not a complete list.

These neuromuscular conditions have certain features in common:

- Most are hereditary
- Most are progressive
- Each causes a characteristic selective pattern of muscle weakness
- There are currently no cures
- Each condition varies in severity but can result in significant physical disability and may reduce lifespan
Muscular Dystrophy

1. Adrenoleukodystrophy
   - Changes in muscle tone, especially muscle spasms and spasticity
   - Crossed eyes (strabismus)
   - Hearing loss
   - Hyperactivity
   - Worsening nervous system deterioration
   - Coma
   - Decreased fine motor control
   - Paralysis
   - Seizures
   - Swallowing difficulties
   - Visual impairment or blindness

2. Bethlem Myopathy (BM)
   - Low muscle tone and a stiff neck that causes the head to lean to one side (torticollis)
   - Delayed developmental milestones, such as sitting or walking.
   - Skin abnormalities such as small bumps called follicular hyperkeratosis that develop around the elbows and knees

3. Becker Muscular Dystrophy (BMD)
   - Muscle loss in begins with the hips and pelvic area, and the thighs and the shoulders
   - To compensate for weakening muscles, the person may walk with a waddling gait, walk on his toes or stick out the abdomen
4. **Congenital Muscular Dystrophy (CMD)**

- Begins in infancy or very early childhood (typically before age 20)
- Identified as hypotonia, or lack of muscle tone, can make an infant seem “floppy.”
- Slow to meet motor milestones
- Significant learning disabilities, or mental retardation

5. **Distal Myopathy (DM)**

- Begins in adulthood
- Muscle weakness in the throat, lower legs, and forearms
- Muscle weakness in the ankles
- Muscles weakness in the hands, wrists, and shoulders
- Weakness of the vocal cords and throat
- Difficulty swallowing (dysphagia)

6. **Distal Muscular Dystrophy (DD)**

- Late walkers
- Enlarged calf muscles (known as pseudohypertrophy, or “false enlargement”)
- Clumsy and fall often
- Trouble climbing stairs, getting up from the floor or running
- Difficulty raising their arms

7. **Duchenne Muscular Dystrophy (DMD)**

- Frequent falls
- Difficulty getting up from a lying or sitting position
- Trouble running and jumping
- Waddling gait
- Large calf muscles
- Learning disabilities
- Generally lose the ability to walk by 12 years of age, after which they need to use a wheelchair
8. **Emery-Dreifuss muscular dystrophy**

- Contractures become noticeable in early childhood
- Muscles weakness in the upper arms and lower legs and progressing to muscles in the shoulders and hips.
- Heart problems by adulthood

9. **Facioscapulohumeral muscular dystrophy (also known as Landouzy-Dejerine Muscular Dystrophy)**

- Affected most often: muscles in the face (facio-)
- Muscle weakness around the shoulder blades (scapulo-), and
- Muscle weakness in the upper arms (humeral)

10. **Limb-Girdle Muscular Dystrophies**

- Difficulty standing from a sitting position without using the arms,
- Difficulty climbing stairs
- Abnormal, sometimes waddling, walk
- Large and muscular-looking calves (pseudohypertrophy), which are not actually strong
- Loss of muscle mass, thinning of certain body parts
- Palpitations or passing-out spells
- Shoulder weakness

11. **Myotonic Muscular Dystrophy Type I (also called Steinert’s Disease)**

- Prolonged muscle contractions (myotonia) and are not able to relax certain muscles after use
- Clouding of the lens of the eye (cataracts)
- Abnormalities of the electrical signals that control the heartbeat (cardiac conduction defects)
- Muscle weakness in the lower legs, hands, neck, and face
12. Myotonic Muscular Dystrophy Type II (also known as Proximal Myotonic Myopathy)

- Begins in a person’s twenties
- Prolonged muscle contractions (myotonia) and are not able to relax certain muscles after use
- Difficulty releasing their grip on a doorknob or handle
- Slurred speech or temporary locking of their jaw
- Muscle pain and weakness that mainly affects the neck, shoulders, elbows, and hips
- Cardiac (heart) problems
- Clouding of the lens in the eyes (cataracts)
- Diabetes
- Males may experience balding and infertility

13. Myotonic Muscular Dystrophy

- Delayed muscle relaxation after contraction
- Impaired nourishment of nonmuscular tissue
- Weaknesses in the facial muscles, arms and legs, and muscles affecting speech and swallowing
- Baldness in men and women
- Intellectual impairment
- Respiratory problems
- Heart abnormalities in early adulthood

14. Manifesting carrier of Muscular Dystrophy

- Mild degree of muscle weakness
- Inability to walk, differing from individual to individual
- May have problems such as:
  - Drooping eyelids (a condition known as ptosis)
  - Choke frequently
  - Difficulty swallowing (called dysphagia)
  - Eventual weakness of the muscles in the face and limbs
  - Problems with kneeling, bending, squatting, walking and climbing stairs.
  - Double vision and a "breathy" quality of the voice
15. Miyoshi Myopathy

- Appears in mid to late childhood or early-adulthood,
- Most marked in the distal parts of the legs,
- Not able to stand on tiptoe
- Weakness and atrophy in the thighs and gluteal muscles
- Difficult to climbing stairs, standing, and walking
- Weakness in shoulder girdle muscles

16. Ophthalmoplegic Muscular Dystrophy

- Droopy eyelids
- Difficulty moving eyes
- Difficulty swallowing

17. Tibial Muscular Dystrophy (also known as Udd Distal Myopathy)

- Appears after age 35
- Affects the muscles at the front of the lower leg
- Difficult to lift the toes while walking
- Affecting muscles in the arms
List of Neuromuscular Conditions

Leukodystrophies

18. Metachromatic leukodystrophy

- Loss of sensation in the extremities (peripheral neuropathy)
- Incontinence
- Seizures
- Paralysis
- Inability to speak
- Blindness
- Hearing loss

19. Adrenoleukodystrophy (also known as X-linked adrenoleukodystrophy)

There are three distinct types of X-linked adrenoleukodystrophy

**First type is a childhood cerebral form**
- Appear by the age of 10
- Learning and behavioral problems
- Difficulty reading, writing, understanding speech, and comprehending written material
- Aggressive behavior
- Vision problems
- Impaired adrenal gland function

**Second type is Adrenomyeloneuropathy**
- Appear between early adulthood and middle age
- Progressive stiffness and weakness in their legs (paraparesis)
- Urinary and genital tract disorders
- Some degree of brain dysfunction
- Adrenocortical insufficiency

**Third type is Addison disease**
- Weakness
- Weight loss
- Skin changes
- Vomiting
- Coma
20. **Amyotrophic Lateral Sclerosis (ALS)** (also known as Lou Gehrig’s Disease or Motor Neurone Disease)

- Appears in one's late forties or early fifties
- Muscle twitching, cramping, stiffness, or weakness
- May develop slurred speech
- Difficulty chewing or swallowing (dysphagia)
- Arms and legs begin to look thinner as muscle tissue wastes away (atrophies)
- Lose their strength and the ability to walk
- Breathing difficulty

21. **Distal Spinal Muscular Atrophy** (also known as Spinal Muscular Atrophy and Hereditary motor neuropathy type)

- Difficult and noisy breathing
- Weak cry
- Problems feeding
- Recurrent episodes of pneumonia
- Weakness spreads to all muscles and lose all ability to move muscles
- Weakness severely impairs motor development, such as sitting, standing, and walking

22. **Spinal Bulbar (Muscular) Atrophy** (also known as Kennedy’s Disease and X-Linked Myotubular myopathy)

- Begins in adulthood and worsens slowly over time
- Muscle weakness and wasting (atrophy)
- Muscle wasting in the arms and legs results in cramping; leg muscle weakness
- Difficulty walking and a tendency to fall
- Certain muscles in the face and throat (bulbar muscles)
- Problems with swallowing and speech
- Muscle twitches
- Unusual breast development
23. Spinal Muscular Atrophy Type I (also known as Werdnig-Hoffman disease, Acute Spinal Muscular Atrophy or Scapuloperoneal Muscular Atrophy)

- Appears after birth
- Difficulty breathing,
- Sucking and swallowing difficulty
- Unable to support their head or sit unassisted
- Weak muscle of thighs

24. Spinal Muscular Atrophy Type II (also known as Intermediate Spinal Muscular Atrophy)

- Develops in children between ages 6 and 12 months
- Can sit without support, although they may need help getting to a seated position
- Unable to stand or walk without support

25. Spinal Muscular Atrophy Type III (also known as Kugelberg-Welander disease)

- Develop between early childhood and adolescence
- Difficulty to climbing stairs

26. Spinal Muscular Atrophy Type IV

- Appears after age 30
- Mild to moderate muscle weakness
- Tremor, twitching, or mild breathing problems
- Muscle weakness in upper arms and legs muscles
Condition of the Peripheral Nerve

27. **Andermann Syndrome** Peripheral neuropathy and agenesis of the corpus callosum (also known as Charlevoix - Saguenay Syndrome/Disease)

- Abnormal or absent reflexes (areflexia) and weak muscle tone (hypotonia)
- Severe progressive weakness and loss of sensation in the limbs
- Rhythmic shaking (tremors)
- Lose this ability of walking by their teenage years
- Abnormal curvature of the spine (scoliosis)
- Abnormal function of certain cranial nerves
- Facial muscle weakness,
- Drooping eyelids (ptosis),
- Difficulty following movements with the eyes (gaze palsy)

28. **Charcot-Marie-Tooth Disease** (also known as Hereditary Motor and Sensory Neuropathy, or Peroneal Muscular Dystrophy)

- Appears in adolescence or early adulthood
- Balance difficulties,
- Clumsiness,
- Muscle weakness in the feet
- Muscles weakness in the lower legs
- Foot abnormalities such as high arches (pes cavus), flat feet (pes planus), or curled toes (hammer toes)
- Difficulty flexing the foot or walking on the heel of the foot
- Gradual hearing loss, deafness, or loss of vision

29. **Critical illness polyneuropathy and/or myopathy**

- Flaccid, predominantly distal tetra paresis or tetraplegia: Lower limbs more affected than upper limbs
- Weakness of the respiratory muscles
- Deep tendon reflexes reduced
- Sensory loss may be present
- Loss of pain, temperature, and vibrations sense in the distal limbs
30. Chronic Inflammatory Demyelinating Polyneuropathies

- Muscle weakness in the ankles
- Numbness and tingling in the feet and toes
- Abdominal fullness or bloating, diarrhea, or constipation
- Difficulty with walking
- Low blood with dizziness, or trouble maintaining an erection

31. Congenital insensitivity to pain and anhidrosis (also called Hereditary sensory and autonomic neuropathy type IV)

- Analgesia (inability to feel pain)
- Unintentional self-injury
- Biting the tongue, lips, or fingers
- Seizures
- Reduced reflexes
- Pupillary abnormalities
- Vasomotor instability
- Lack of sweating
- Aplasia of dental enamel

32. Dejerine-Sottas Disease

- Severe neuropathy (disease or abnormality of the nerves)
- Muscle weakness
- Loss of or changes in sensation,
- Curvature of the spine
- Mild hearing loss

33. Familial amyloid neuropathy

- Polyneuropathy
- Impaired pain and temperature sensation
- High blood pre-albumin levels
- Loss of sensation in extremities
- Foot ulcers
- Diarrhea
- Orthostatic hypotension
34. Familial Dysautonomia (also known as Hereditary Sensory and Autonomic Neuropathy and also as Riley Day syndrome)

- Motional stress or infection
- Poor muscle tone
- Impaired temperature regulation
- Feeding difficulties
- Poor motor coordination
- Inadequate eye moisture
- Anxiety and learning difficulties
- Abnormal spinal curvature
- Problems with bone health and physical coordination

35. Giant Axonal Neuropathy

- Appears in infancy or early childhood
- Loose sensation in the arms, legs, and other parts of the body.
- Problems with walking
- Lose sensation, coordination, strength, and reflexes in their limbs
- Hearing and visual problem

36. Guillain-Barré Syndrome (also known as Acute Inflammatory Demyelinating Polyradiculoneuropathy)

- Muscle weakness or paralysis
- Muscle weakness in the legs and spreads to the arms, torso, and face
- Numbness, tingling, or pain
- Swallowing difficulty
- Difficulty breathing
- Blood pressure or an abnormal heartbeat (cardiac arrhythmia)
37. Hereditary Neuropathy with liability to pressure palsy

- Episodes of numbness, tingling, and/or loss of muscle function (palsy) (episode can last from several minutes to several months)
- Permanent muscle weakness or loss of sensation
- Pain in the limbs, especially the hands
- Problem sites involve nerves in wrists, elbows, knees and fingers, shoulders, hands, feet, and the scalp

38. Hereditary Sensory and Autonomic Neuropathy Type II

- Acroosteolysis
- Loss of sense of touch
- Loss of sense of pain
- Loss of sense of temperature
- Amyotrophy
- Abnormal muscle development
Inflammatory Myopathies

39. Dermatomyositis

- Skin changes
- Muscle weakness in hips, thighs, shoulders, upper arms and neck
- Difficulty swallowing (dysphagia)
- Muscle pain or tenderness
- Fatigue, fever and weight loss
- Hardened deposits of calcium under the skin (calcinosis), especially in children
- Gastrointestinal ulcers and intestinal perforations
- Lung problems

40. Inclusion Body Myositis

- Lowly progressive weakness in the muscles of the wrists and fingers
- Muscle weakness in the front of the thigh (quadriceps)
- Trouble with gripping, such as a shopping bag or briefcase
- Frequent stumble
- Weakness of the swallowing muscles

41. Myositis Ossificans Polymyositis

- Muscle weakness
- Rigid muscles
- Tendon weakness
- Rigid tendons
- Calcium deposits in muscles
- Movement pain
- Tenderness
- Skin swelling over calcified site
- Shortened digits
- Skeletal malformations
- Malformed fingers
- Malformed toes
- Limited joint movement
- Skin swelling
42. Lyme Neuropathy
   - Fatigue
   - Fever and chills
   - Muscle and joint pain
   - Red circular rash
   - Stiff neck
   - Swollen lymph nodes
   - Numbness and tingling
   - Peripheral neuropathy
   - Pain, numbness and tingling in limbs
   - Paralysis of facial muscles (Bell's palsy)
43. Congenital Myasthenic Syndrome (also known as Congenital Myasthenia)

- Appears in adolescence or adulthood
- Weak facial muscles, including muscles that control the eyelids, muscles that move the eyes
- Chewing and swallowing difficulty
- Feeding difficulties
- Crawling or walking may be delayed
- Unable to walk in severe weakness
- Episodes of breathing problems

44. Lambert-Eaton Myasthenic Syndrome

- Weakness or loss of movement that can be more or less severe, including:
  - Difficulty chewing
  - Difficulty climbing stairs
  - Difficulty lifting objects
  - Difficulty talking
  - Drooping head
  - Need to use hands to get up from sitting or lying positions
- Swallowing difficulty, gagging, or choking
- Vision changes such as:
  - Blurry vision
  - Double vision
  - Problems keeping a steady gaze

45. Myasthenia Gravis

- Muscles weakness
- Muscles weakness in the Eye
  - Drooping of one or both eyelid
  - Double vision (diplopia)
List of Neuromuscular Conditions

- Muscles weakness in the throat
- Altered speaking
- Difficulty swallowing
- Problems chewing
- Muscles weakness in the Neck and limb
- Breathing difficulty
- Difficulty using arms or hands
- Difficulty with head control
46. Central core disease

- Mild muscle weakness that does not worsen with time
- Muscles in the upper legs and hips
- Delay of motor development
- Weak muscle tone (hypotonia)
- Breathing problems
- Abnormal curvature of the spine (scoliosis)
- Hip dislocation, and joint deformities

47. Centronuclear myopathy (also known as Myotubular Myopathy)

There are two forms, which are differentiated by their pattern of inheritance:

**First form: Autosomal dominant**
- Appears at adolescence or early adulthood
- Muscle pain during exercise
- Difficulty walking
- Weakness in the muscles that control eye movement (ophthalmoplegia)
- Droopy eyelids (ptosis)
- Disturbances in nerve function (neuropathy) or intellectual disability

**Second form: Autosomal recessive**
- Apparent at birth or begins in childhood
- Foot abnormalities
- High arch in the roof of the mouth (high-arched palate)
- Abnormal side-to-side curvature of the spine (scoliosis)
- Mild to severe breathing problems
- The heart muscle is weakened (cardiomyopathy)

48. Hyperthyroid Myopathy

- Muscles weakness around the shoulders and sometimes the hips
- Weakness in muscles of the face, throat, and the respiratory muscles
- Unable to control movement of the eye and eyelids, which can lead to vision loss
49. Hypothyroid Myopathy

- Muscle enlargement along with muscle weakness
- Muscle weakness around the hips and sometimes the shoulders
- Slowing of reflexes
- Muscle stiffness and painful muscle cramps

50. Inclusion Body Myopathy 2

- Appears in late adolescence or early adulthood
- Weakness of a muscle in the lower leg
- Weakness in the tibialis anterior muscles
- Difficulty in walking, running and climbing stairs
- Muscles weakness in the upper legs, hips, shoulders, and hands

51. Inclusion Body Myopathy with early-onset Paget disease and Frontotemporal Dementia (IBMPFD)

- Appears in mild-adulthood
- Muscle weakness
- Muscle weakness in the hips and shoulders
- Muscle weakness in the arms and legs
- Respiratory and heart muscle weakness (cardiomyopathy)
- Breathing difficulties
- None pain particularly in the hips and spine
- Trouble speaking, remembering words and names (dysnomia)
- Loss of Judgment and inappropriate social behavior

52. Isaac’s Syndrome (also known as acquired neuromytonia)

- Persistent myokymia
- Lower limb contractures
- Increased muscle tone
- Cyanotic episodes
- Transient stiffness
- Reduced motor activity with flexion
- Muscle cramps
- Difficulty relaxing muscles
53. Periodic Paralysis (also known as Hyperkalemic Periodic Paralysis or Gamstorp Disease)

- Begins in childhood or adolescence
- Episodes involve a temporary inability to move muscles in the arms and legs
- Reduced levels of potassium in their blood (hypokalemia) during episodes of muscle weakness

54. Polymyositis

- Progressive muscle weakness
- Difficulty swallowing (dysphagia)
- Difficulty speaking
- Mild joint or muscle tenderness
- Fatigue
- Shortness of breath
Metabolic Muscle Conditions

55. Acid Maltase Deficiency (also known as Pompe’s Disease or Glycogenosis Type II)

- Muscle weakness (myopathy)
- Poor muscle tone (hypotonia)
- Heart defects (mild to severe)
- Breathing problems
- Delayed motor skills
- Muscle weakness in the legs and the trunk

56. Andersen Disease/Syndrome (also known as Glycogen Storage Disease Type IV or Branching Enzyme Deficiency)

- Irregular heartbeat
- Discomfort
- Fainting caused by irregular heartbeat
- Small lower jaw
- Dental abnormalities
- Low-set ears
- Widely spaced eyes
- Abnormal curving of fingers
- Abnormal curving of toes
- Short stature
- Abnormal curvature of the spine

57. Adenylate Deaminase Deficiency Myodenylate Deaminase Deficiency

- Intolerance, cramps and muscle pain
- People with deficiencies in this enzyme may experience no symptoms
58. Barth Syndrome

- Heart muscle weakness (cardiomyopathy)
- Skeletal muscle abnormalities
- Low levels of white blood cells
- Slow development or weak muscle tone
- Increased levels of organic acids in the urine and blood
- Frequent bacterial infections, such as pneumonia

59. Carnitine Palmityl Transferase I Deficiency (CPT I Deficiency)

- Appears during early childhood
- Low blood sugar (hypoglycemia)
- Low level of ketones
- Enlarged liver (hepatomegaly),
- Liver malfunction
- Elevated levels of carnitine in the blood

60. Danon Disease or (also known as Glycogen Storage Type IIB)

- Begins in childhood or adolescence in most affected male
- Begin in early adulthood in most affected females
- Proximal muscle weakness
- Thickening of heart muscle
- Intermittent hepatomegaly
- Glycogen deposits in the heart

61. Debranching Enzyme Deficiency (also known as Forbes Disease)

- Liver malfunction
- Slowing of growth, low blood sugar levels
- Seizures.
- Muscle weakness of forearms, hands, lower legs and feet
- Heart defects
62. Glycogen Storage Disease Type III (also known as Cori Disease)

- Begins in infancy
- Low blood sugar (hypoglycemia)
- Elevated blood levels of liver enzymes
- Enlarged liver (hepatomegaly)
- Liver failure later in life
- Slow growth

63. Lactate Dehydrogenase A Deficiency (also known as glycogen storage disease XI)

- Fatigue, muscle pain, and cramps during exercise (exercise intolerance)
- Breakdown of muscle tissue (rhabdomyolysis)
- Kidney failure
- Skin rashes

64. Myoadenylate Deaminase Deficiency

- Myalgia
- Exercise-induced myalgia
- Weakness
- Muscle cramps
- Muscle pain

65. Myochondrial Myopathies

- Muscle weakness
- Exercise intolerance
- Loss of hearing
- Seizure Disorder
- Lack of balance or coordination
- Progressive weakness
- Inability to move eyes
- Heart Failure
- Learning deficits
- Blindness
List of Neuromuscular Conditions

- Stroke-like episodes
- Droopy eyelids
- Breathlessness
- Dementia
- Diabetes
- Muscle wasting

66. Primary Carnitine Deficiency

- Appears during infancy or early childhood
- Severe brain dysfunction (encephalopathy)
- Weakened and enlarged heart (cardiomyopathy)
- Confusion, vomiting, muscle weakness
- Low blood sugar (hypoglycemia)

67. Phosphofructokinase Deficiency (also known as Tauri’s disease or Glycogenosis Type VII)

- Exercise intolerance, with pain, cramps
- Muscle breakdown
- Lowering blood levels of fats
- Kidney failure

68. Phosphoglycerate Mutase Deficiency

- Begin in childhood or adolescence
- Experience muscle aches or cramping
- Kidney failure
- Permanent weakness

69. Phosphorylase Deficiency (also known as Myophosphorylase Deficiency or McArdle’s disease)

- Developmental delay
- Exercise intolerance, such as cramps, muscle pain and weakness
List of Neuromuscular Conditions

Congenital Myopathies

70. Arthrogryposis Multiplex Congenita
- The condition is always present at birth (congenital) but does not get worse over time (it is not progressive)
- Hands, wrists, elbows, shoulders, hips, feet, and knees are affected.
- Limit movement

71. Congenital fibre type disproportion
- Muscle weakness (myopathy) throughout the body
- Muscles weakness of the shoulders, upper arms, hips, and thighs
- Muscles weakness in the face and muscles that control eye movement (ophthalmoplegia), sometimes causing droopy eyelids
- Joint deformities (contractures)
- Abnormally curved lower back (lordosis) or spine that curves to the side (scoliosis)
- Mild to severe breathing problems
- Difficulty swallowing
- Weakened and enlarged heart muscle

72. Nemaline Myopathy (also known as rod myopathy or Nemaline Rod Myopathy)
- Muscles weakness of the face, neck, and limbs
- Feeding and swallowing difficulties
- Foot deformities
- Abnormal curvature of the spine (scoliosis)
- Joint deformities (contractures)
- Breathing difficulty

73. Myosin storage myopathy
- Childhood onset
- Waddling gait and difficulty climbing stairs,
- Difficulty lifting the arms above shoulder level
- Trouble breathing
74. Multiminicore disease

- Begins in infancy or early childhood
- Muscle weakness in the trunk and neck (axial muscles)
- Less severe in the arm and leg muscles
- Delay the development of motor skills
- Breathing difficulties
- Abnormal curvature of the spine (scoliosis)
- Muscle weakness and looseness of the joints, particularly in the arms and hands

75. Minicore Myopathy

- Begins in infancy or early childhood
- Muscle weakness in the trunk and neck (axial muscles)
- Muscle wasting
- Scoliosis
- Impaired breathing function
- Reduced infant muscle tone
- Delayed motor development
- Infant feeding problems
- Double-jointed
- Weak eye muscles

76. Reducing body myopathy

- Rapid progression and fatal outcome
- Delayed developmental milestone
- Limb weakness and wasting at onset
- Severe generalized muscle wasting and weakness,
- Respiratory failure
77. Walker-Warburg Syndrome (also known as Arburg syndrome, Chemke syndrome, HARD syndrome Hydrocephalus, Agyria and Retinal Dysplasia, Pagon syndrome, cerebro-ocular dysgenesis or cerebro-ocular dysplasia-muscular dystrophy syndrome)

- Muscle weakness
- Mental retardation
- Seizures
- Cataracts
- Anterior chamber malformation
- Detachment of the retina
- Glaucoma
- Abnormalities of the male genitalia
Other Myopathies

78. Andersen-Tawil syndrome

- Physical abnormalities of the head, face, and limbs
- Small lower jaw
- Dental abnormalities
- Low-set ears
- Widely spaced eyes
- Unusual curving of the fingers or toes (clinodactyly)
- Abnormal curvature of the spine (scoliosis)

79. Laing Distal Myopathy

-Appears in childhood
- Muscle weakness in the feet and ankles
- Muscle weakness in the hands and wrists
- Inability to lift the big toe, and a high-stepping walk
- Difficult to lift the fingers, especially the third and fourth fingers
- Weakness in several muscles of the neck and face.
- Weakness in the legs, hips, and shoulders

80. Myofibrillar Myopathy

- Appears anytime between infancy and late adulthood.
- Muscle weakness in the hands and feet
- Facial muscle weakness
- Difficulty to swallowing
- Difficulty to speech
- Loss of sensation
- Weakened heart muscle
- Muscle weakness in the limbs
- Respiratory failure
- Abnormal side-to-side curvature of the spine
- Clouding of the lens of the eyes
81. Mitochondrial myopathy

- Exercise intolerance
- Hearing loss
- Trouble with balance and coordination
- Seizures
- Learning deficits
- Impaired vision,
- Heart defects,
- Diabetes and stunted growth

82. Nonaka Myopathy

- Difficulty running
- Tendency to fall
- Leg muscle weakness
- Leg muscle wasting
- Difficulty walking on toes

83. Tubular Aggregate Myopathy

- Begins in childhood or adult
- Muscle weakness
- Muscle cramps
- Limb weakness
Genetically determined Ataxias

84. Abetalipoproteinemia (also known as Bassen-Kornzweig)

- Abnormal growth patterns in infants – developmental delays or “failure to thrive”
- Curving of the spine
- Problems with balance and dexterity
- Problems with coordination
- Muscle weakness
- Protruding abdomen
- Problems with vision
- Speech disorders, slurring of speech
- Fatty, frothy, foul-smelling, or otherwise irregular stools

85. Ataxia Talangiectasia

- Begins in early childhood
- Walking difficulty
- Problems with balance and hand coordination,
- Involuntary jerking movements (chorea)
- Muscle twitches (myoclonus)
- Disturbances in nerve function (neuropathy)
- Trouble moving their eyes to look side-to-side (oculomotor apraxia)
- Chronic lung infections
- Sensitive to the effects of radiation exposure, including medical x-rays

86. Ataxia with congenital glaucoma

- Glaucoma
- Generalized loss of reflexes
- High foot arch
- Muscle weakness
- Abnormal walk
- Speech problems
- Clumsy movements
- Unsteadiness
List of Neuromuscular Conditions

- Muscle wasting
- Loss of tendon reflexes
- Easily fatigued
- Rapid involuntary eye movements
- Loss of sensation

87. Ataxia with vitamin E deficiency

- Difficulty coordinating movements (ataxia)
- Speech (dysarthria)
- Loss of reflexes in the legs (lower limb areflexia)
- Loss of sensation in the extremities (peripheral neuropathy)
- Vision loss

88. Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)

- Abnormal tensing of the muscles (spasticity)
- Difficulty coordinating movements (ataxia)
- Muscle wasting (amyotrophy)
- Involuntary eye movements (nystagmus)
- Speech difficulties (dysarthria)
- Deformities of the fingers and feet
- Reduced sensation
- Muscle Weakness in the arms and legs (peripheral neuropathy)

89. Friedrech’s Ataxia

- Loss of strength and sensation in the arms and legs
- Muscle stiffness (spasticity)
- Impaired speech
- Have a form of heart disease called hypertrophic cardiomyopathy
- Diabetes,
- Impaired vision,
- Hearing loss, or an abnormal curvature of the spine (scoliosis)
- Poor balance when walking
90. Spinocerebellar Ataxia type 1

- Begins in early adulthood
- Problems with coordination and balance
- Speech and swallowing difficulties
- Muscle stiffness (spasticity)
- Muscle weakness in the muscles that control eye movement (ophthalmoplegia)
- Difficulty processing, learning, and remembering information (cognitive impairment)
- Numbness, tingling, or pain in the arms and legs (sensory neuropathy)
- Uncontrolled muscle tensing (dystonia)
- Muscle wasting (atrophy)
- Muscle twitches (fasciculations)
Myotonic disorders (distinct from channelopathies or dystrophies)

91. Brody Myopathy

- Begins in childhood
- Muscle cramping and stiffening after exercise
- Muscles weakness of the arms, legs, and face (particularly the eyelids)

92. Chondrodystrophic Myotonia (also known as Schwartz-Jampel Syndrome)

- Muscle weakness and stiffness (myotonic myopathy)
- Abnormal bone development (bone dysplasia)
- Permanent bending or extension of certain joints in a fixed position (joint contractures)
- Small, fixed facial features and various abnormalities of the eyes
- Impaired vision

93. Rippling Muscle Disease

- Rippling muscles, with rolling muscle contractions
- Muscle cramps
- Muscle pain
- Muscle stiffness
- Myotonia
Phakomatoses

94. Neurofibromatosis

- Begins in early childhood
- Multiple café-au-lait spots, which are flat patches on the skin that are darker than the surrounding area
- Freckles in the underarms
- Neurofibromas, which are noncancerous (benign) tumors that are usually located on or just under the skin
- Cancerous tumors that grow along nerves
- High blood pressure (hypertension)
- Short stature,
- Unusually large head (macrocephaly)
- Skeletal abnormalities such as an abnormal curvature of the spine (scoliosis)

95. Neurofibromatosis Type 2 (also known as vestibular schwannomas or acoustic neuromas)

- Appears during adolescence or in a person’s early twenties
- Signs and symptoms vary according to tumors location
- Hearing loss
- Ringing in the ears (tinnitus)
- Problems with balance
- Changes in vision or sensation
- Numbness or weakness in the arms or legs
- Fluid buildup in the brain

96. Schwannomatosis

- Chronic pain
- Numbness
- Tingling
- Weakness
97. Tuberous Sclerosis

- Skin abnormalities
  - Patches of light-colored skin
  - Facial lesions
- Neurological symptoms
  - Seizures,
  - Intellectual disability,
  - Learning disabilities or developmental delays
  - Trouble with communication and social interaction
- Kidney problems
- Lung problems

98. Von Hippel Lindau syndrome

- Appears during young adulthood
- Hemangioblastomas (Tumors) that develop in the brain and spinal cord can cause headaches, vomiting, weakness, and a loss of muscle coordination (ataxia)
- Cysts in the kidneys
Channelopathies

99. Myotonia Congenita (also known as Thomsen’s disease)

- Begins in childhood
- Muscles weakness in the face and tongue
- Muscle weakness in the legs
- Muscle stiffness that can interfere with movement

100. Paramyotonia Congenita

- Begins in infancy or early childhood,
- Experience bouts of sustained muscle tensing (myotonia) that prevent muscles from relaxing normally
- Stiffness chiefly affects muscles in the face, neck, arms, and hands
Other disorders

101. Congenital Fibrosis of the Extraocular Muscles

- Unable to move their eyes normally
- Difficulty looking upward
- Their side-to-side eye movement also be limited
- Droopy eyelids (ptosis)
- Limits their vision

102. Hereditary Spastic Paraplegias (HSP) (also known as Familial Spastic Paraparesis)

- Abnormal gait
- Delayed walking
- Repeated tripping or falling
- Weakness of the leg muscles
- Rigidity and increased tone of the person’s leg muscles
- Leg cramps
- Muscle spasms
- Highly arched feet
- Bladder control problems
- Relatively mild muscle wasting
- Diminished vibration sense in the feet
- Ankle clonus or abnormal reflex movements of the foot

103. Hyperkalemic periodic paralysis (also known as Impressive Syndrome)

- Begins in adolescence
- Occasional episodes of muscle weakness.
- Loss of muscle movement (paralysis) that come and go
- Most commonly occurs at the shoulders and hip
- May also involve the arms and legs but does not affect muscles of the eyes and those that help you breathe and swallow
- Most commonly occurs while resting after activity
- May occur on awakening
- Intermittent, usually lasting 1-2 hours
104. Kearns-Sayre Syndrome (also known as oculocraniosomatic disease or Oculocraniosomatic neuromuscular disease)

- Appears before age 20
- Paralysis of eye muscles
- Diabetes
- Deafness
- Myopathy
- Heart block
- Hearing loss
- Short stature
- Heart disease
- Muscle weakness
- Endocrine disorders
- Retinal pigmentation
- Difficulty walking or moving
- Cardiac conduction defects
- Progressive external ophthalmoplegia
For further information or support please do not hesitate to contact the Association.

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