MUSCULAR DYSTROPHY CANADA
DYSTROPHIE MUSCULAIRE CANADA
MUSCLE.CA

NEUROMUSCULAR DISORDER UMBRELLA

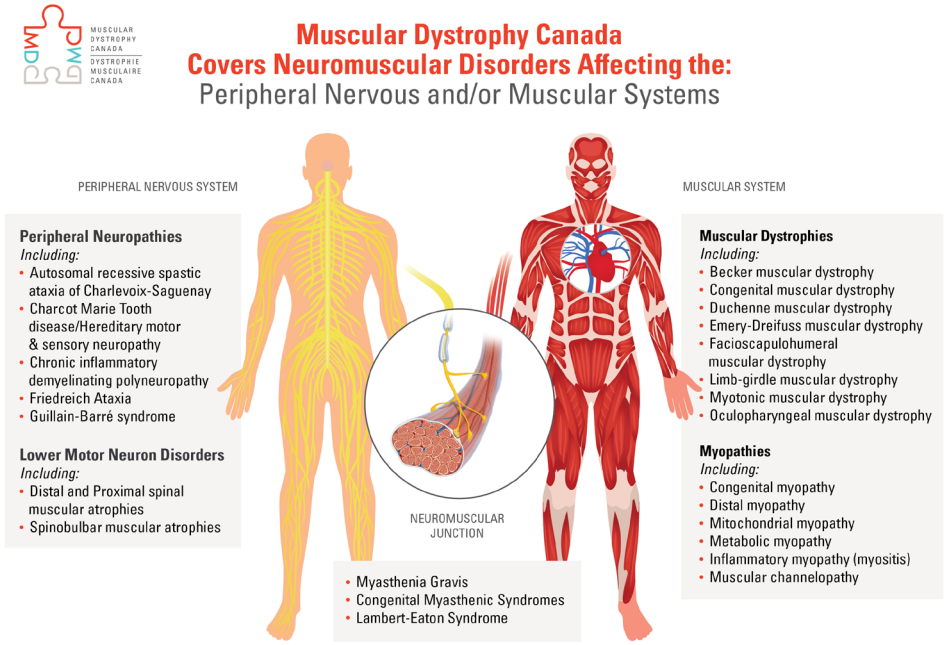
at Muscular Dystrophy Canada

Muscular Dystrophy Canada's (MDC) mission focuses on supporting Canadians affected by neuromuscular disorders. MDC defines neuromuscular disorders (NMDs) as health conditions in which the primary area impacted is the muscles and/ or the peripheral nervous system.

The list of NMDs in this document are some of the NMDs we cover, but we are always monitoring and learning about other disorders that may fall under our umbrella. If you don't see a disorder list that you feel falls under this definition, please contact research@muscle.ca.

MDC supports individuals affected by NMDs in the following groups:

- **Primary disorders of the muscle** (myopathies, muscular dystrophies);
- **Primary disorders of the lower motor neuron and peripheral nerves** (genetic or immune-mediated peripheral neuropathies; lower motor neuron disorders);
- **Neuromuscular junctions disorders** (genetic or immune-mediated myasthenic conditions)



Please contact research@muscle.ca with any concerns or questions about the disorders listed or other related disorders.

PRIMARY DISORDERS OF THE MUSCLES

Muscular Dystrophies

Congenital muscular dystrophies (CMD)
All types including:

- Congenital muscular dystrophy, collagenopathies including Collagen VI and XII related dystrophies, Bethlem, Ullrich CMD
- Congenital muscular dystrophy, dystroglycanopathies including all subtypes of Fukuyama, Muscle-eye-brain disease, Walker-Warburg syndromes, Congenital muscular dystrophy with hypoglycosylation of dystroglycan, incl related to *B4GAT1, CRPPA, DPM2, DAG1, FKTN, FKRP, GMPPB, LARGE1, POMT1, POMT2, POMGNT1, POMGNT2*
- Congenital muscular dystrophy, laminopathies CMD related to *LMNA*
- Congenital muscular dystrophy, merosinopathies CMD related to *LAMA2*
- SELENON/SEPN1 related CMD Rigid spine syndrome
- Other CMD including but not limited to *ACTA1, CHKB, DNM2, ITGA7, RYR1, SYNE1, TCAP* related CMD and CMD without genetic diagnosis

Progressive muscular dystrophies

- Becker muscular dystrophy BMD
- Duchenne muscular dystrophy DMD
- Dystrophinopathies, other including X-linked Cardiomyopathy, Dilated cardiomyopathy 3B (Manifesting Female)
- Emery Dreifuss muscular dystrophy All subtypes of EDMD
- Facioscapulohumeral muscular dystrophy FSHD1 related to D4Z4, FSHD2 related to *SMCHD1*
- Limb girdle muscular dystrophy All subtypes including but not limited to *ANO5, CAPN3, DNAJB6, DYS* (Miyoshi myopathy), *FKR, GMPPB, SGCA/D/G, TTN* –related LGMD
- Myotonic dystrophy All subtypes including DM1 Steinert disease (congenital, childhood, juvenile and adult onset DM1), DM2 proximal myotonic myopathy, Ricker syndrome
- Oculopharyngeal muscular dystrophy OPMD

Myopathies

Congenital myopathies
All types including:

- Cap myopathies
- Central core disease
- Centronuclear myopathies including recessive, dominant, and X-linked (Myotubular Myopathy) subtypes
- Congenital fiber type disproportion myopathy
- Minicore myopathy or Multiminicore myopathy *RYR1*
- Nemaline myopathy
- Tubular aggregate myopathies

Distal myopathies
All types including:

- Udd myopathy Tibial myopathy
- Nonaka myopathy Inclusion body myopathy (inherited), GNE myopathy
- Laing distal myopathy Distal myopathy type 1
- Welander myopathy

Immune-mediated myopathies/Myositis

- Dermatomyositis
- Immune-mediated necrotizing myositis
- Sporadic inclusion body myositis sIBM
- Polymyositis Note historical diagnostic term.
- Vasculitis related myopathies

Metabolic myopathies

- Carnitine palmitoyl transferase II deficiency *CP2* related
- Glycogen storage disease type II Pompe disease or acid maltase deficiency
- Glycogen storage disease III Forbe disease or Glycogen Debrancher Deficiency
- Glycogen storage disease IV Andersen disease or Glycogen branching enzyme deficiency
- Glycogen storage disease V McArdle disease or myophosphorylase deficiency
- Glycogen storage disease VII Phosphofructokinase deficiency or Tarui disease
- Glycogen storage disease 1Xd Glycogen storage disease 1Xd Glycogen storage disease due to muscle phosphorylase kinase deficiency
- Glycogen storage disease XII Aldolase deficiency
- Glycogen storage disease XIV Congenital disorder of glycosylation type 1T; or Phosphoglucomutase 1 deficiency
- Glycogen storage disease of heart, lethal congenital
- Myoadenylate deaminase deficiency

Mitochondrial disorders with primary myopathy

Muscular ion channel disorders

- Brody myopathy
- Genetic periodic paralysis Andersen-Tawil syndrome, Hyperkalemic and Hypokalemic (Gamstorp disease) periodic paralysis (Sodium, potassium, calcium channels related)
- Myotonia congenita Thompsen and Becker disease
- Neuromyotonia Isaac syndrome
- Schwartz-Jampel Syndrome Chondrodystrophic Myotonia
- Paramyotonia congenita
- Potassium aggravated myotonia

Other myopathies

- Myofibrillar myopathies including desminopathy
- Reducing body myopathy
- Hereditary distal arthrogryposis multiplex congenita
- Rippling muscle disease
- Vacuolar aggregate myopathy

NEUROMUSCULAR JUNCTION DISORDERS

Myasthenic Conditions

Genetic disorders of the neuromuscular junction

- Congenital myasthenic syndromes All subtypes including, Slow and Fast channel syndromes, Acetylcholine receptor deficiency

Immune-mediated disorders of the neuromuscular junction

- Myasthenia gravis
- Lambert-Eaton myasthenic syndromes

PRIMARY DISORDERS OF THE LOWER MOTOR NEURON & PERIPHERAL NERVE

Motor and/or Sensory Nerve Disorders

Genetic peripheral neuropathies

- Amyloid peripheral neuropathy AL and AA amyloidosis
- Andermann syndrome/Agenesis of the corpus callosum with peripheral neuropathy (SLC12A6 related), Charlevoix disease
- Ataxia with vitamin E deficiency Friedreich-like ataxia
- Autosomal recessive spastic ataxia of Charlevoix-Saguenay ARSACS
- Charcot-Marie-Tooth neuropathy/ Hereditary Motor and sensory Neuropathy All subtypes of CMT including but not limited to Hereditary neuropathy with liability to pressure palsies (HNPP), Congenital hypomyelinating neuropathy (HMSN), axonal & demyelinating HMSN and disorders historical known as peroneal neuropathy, Dejerine-Sottas disease
- Friedreich ataxia
- Giant axonal neuropathy GAN
- Hereditary neuralgic amyotrophy (HNA) *SEPT9* related
- Hereditary sensory and autonomic neuropathy (HSAN) all subtypes HSAN I-VIII including congenital insensitivity to pain (CIP), Familial dysautonomia (Riley-Day syndrome)
- Tangier disease Alphalipoproteinem

Immune-mediated peripheral neuropathies

- Antibody mediated paraneoplastic neuropathy
- Critical illness polyneuropathy and/or myopathy
- Chronic inflammatory demyelinating polyradiculoneuropathy CIDP

- Guillain-Barré syndrome including acute inflammatory demyelinating polyradiculoneuropathy (AIDP), Multifocal motor neuropathy MMN with conduction block

- Neuropathy due to monoclonal gammopathy Axonal polyneuropathy and polyradiculonearopathy polyradiculoneuropathy associated with IgG/IgM/IgA monoclonal gammopathy

- Parsonage-Turner syndrome Brachial neuritis, Neuralgic amyotrophy

Mitochondrial disorders with primary peripheral neuropathy

Lower Motor Neuron Disorders

Genetic lower motor neuron disorders:

- Distal hereditary motor neuropathy/ spinal muscular atrophy, dominant (dHMN/dSMA) including dHMN1,2,5,7

- Distal hereditary motor neuropathy/ spinal muscular atrophy, recessive (dHMN/dSMA) including dSMA1 (SMARD1/ dHMN6,) dSMA2-5

- Distal hereditary motor neuropathy/ spinal muscular atrophy, X-linked including SMARD2, SMAX2 (X-linked distal arthrogryposis multiplex congenita), SMAX3

- Proximal spinal muscular atrophy, dominant including SMALED *(BICD2, DYNC1H1)*, SMAFK

- Proximal spinal muscular atrophy, recessive all subtypes related to *SMN1*, SMA5q

- Spinobulbar muscular atrophy, X-linked SBMA, Kennedy disease, SMAX1

If you have questions about any of the disorders listed, or don't see a disorder that you feel should be included, please contact the Research Hotline at 1-800-567-2873 ext 1114 or research@muscle.ca.

The Research Hotline is a direct connection to MDC's research team who can:

- **Explain complex disorder information or recent studies in plain language**

- **Talk to you about new treatments and let you know which clinical trials are available**

- **Answer disorder specific questions**

- **Work with a panel of medical and scientific experts to gather helpful information**

For more information, visit muscle.ca/research-hotline

NOTE: Health conditions that do not fall under the umbrella include acquired traumatic conditions due to birth related injury and/or motor vehicle or other accidents (i.e. brachial plexopathy, spinal cord injuries); transient, toxic diseases (i.e., Lyme disease, Rhabdomyolysis, steroid induced conditions); acquired conditions secondary to non-neuromuscular disorder or its treatment (i.e. diabetic polyneuropathy, peripheral due to chemotherapy/cancer, sarcopenia); brain/ spine ("central nervous system") predominant diseases (i.e., Multiple sclerosis, Huntington, Leukodystrophy, Ataxias with primary CNS involvement; and, upper motor neuron disorders (ie Amyotrophic/Primary lateral sclerosis).