

## Comprehensive List of Neuromuscular Disorders Covered by Muscular Dystrophy Canada

---

Neuromuscular disorders is a general term that refers to diseases that affect any part of the nerve and muscle. These disorders vary according to characteristics such as pattern of inheritance, origin of the genetic mutation, incidence, symptoms, age of onset, rate of progression, and prognosis. Several examples included in this wide range of disorders are:

- Skeletal muscle disorders, such as muscular dystrophies and inflammatory myopathies
- Neuromuscular junction disorders, such as myasthenia gravis
- Peripheral nerve disorders, such as Charcot-Marie-Tooth disease
- Motor neuron disorders, such as Spinal Muscular Atrophy
- Genetically determined ataxias, such as Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay and Friedreich's Ataxia

The following is a comprehensive list of neuromuscular disorders that fall under the umbrella of Muscular Dystrophy Canada. The purpose of this list is to identify those disorders that are served by Muscular Dystrophy Canada and may be eligible for funding. Individuals with these disorders may register with Muscular Dystrophy Canada (free of charge) and be eligible to receive services.

The list is reviewed and updated annually by Muscular Dystrophy Canada's Medical and Scientific Advisory Committee. Disorders not currently listed may be brought forward to the Committee for consideration throughout the year by request.

Muscular Dystrophy Canada funds services to support Canadians with neuromuscular disorders. Not all disorders listed will be the focus of specific research initiatives or direct services from Muscular Dystrophy Canada. Eligibility for direct funding will be determined based on criteria of the specific program, for example, Muscular Dystrophy Canada's Equipment Program or the Neuromuscular Research Partnership. For more information about our research programs and services, please contact the Regional Office in your area.

**a**

- |   |   |
|---|---|
| 1. Abetalipoproteinemia<br>Bassen Kornzweig   | 9. Andersen Disease/Syndrome<br>Glycogen Storage Disease Type IV<br>Branching Enzyme Deficiency |
| 2. Acetylcholine Receptor Deficiency<br>Congenital Myasthenic Syndrome  | 10. Arthrogryposis Multiplex Congenita  |
| 3. Acid Maltase Deficiency<br>Pompe's Disease<br>Glycogenosis Type II   | 11. Ataxia Talangiectasia   |
| 4. Adenylate Deaminase Deficiency<br>Myoadenylate Deaminase Deficiency  | 12. Ataxia with congenital glaucoma   |
| 5. Adrenoleukodystrophy   | 13. Ataxia with optic atrophy and<br>sensorineural deafness                                     |
| 6. Alphaipoproteinemia<br>Tangier's Disease   | 14. Ataxia with vitamin E deficiency  |
| 7. Amyloidosis  | 15. Autosomal Recessive Spastic Ataxia of Charlevoix-<br>Saguenay                               |
| 8. Andermann Syndrome<br>Peripheral neuropathy and agenesis<br>of the corpus callosum<br>Charlevoix - Saguenay Syndrome/Disease | 16. Axonal Motor-Sensory Neuropathy with Deafness<br>and Mental Retardation                     |

---

**b**

- |                                |  |
|--------------------------------|--|
| 17. Barth Syndrome             | 22. Brachial Plexopathy  |
| 18. Becker Muscular Dystrophy  | 23. Branching Enzyme Deficiency<br>Glycogen Storage Disease Type IV<br>Andersen Disease/Syndrome |
| 19. Benign Congenital Myopathy | 24. Brody Disease  |
| 20. Bethlem Myopathy           |  |
| 21. Botulism                   |  |

\* Alternate name for disorder listed below in gray.



## C

- |   |   |
|---|---|
| 25. Carnitine Palmitoyl Transferase Deficiency  | 33. Chudley Syndrome  |
| 26. Carnitine Deficiency  | 34. Congenital Fibre Type Disproportion Myopathy  |
| 27. Central Core Disease  | 35. Congenital Fibrosis of the Extraocular Muscles  |
| 28. Centronuclear Myopathy<br>Myotubular Myopathy   | 36. Congenital Hypomyelinating neuropathy<br>Hereditary motor and sensory neuropathy<br>Charcot-Marie-Tooth Disease |
| 29. Charcot-Marie-Tooth Disease<br>Hereditary Motor and Sensory Neuropathy<br>Congenital insensitivity to pain and anhidrosis | 37. Congenital insensitivity to pain and anhidrosis<br>Hereditary sensory and autonomic neuropathy                  |
| 30. Charlevoix-Saguenay Syndrome<br>Peripheral neuropathy<br>and agenesis of the corpus callosum<br>Andermann syndrome        | 38. Congenital Muscular Dystrophy   |
| 31. Chondrodystrophic Myotonia<br>Schwartz-Jampel Syndrome  | 39. Congenital Myasthenic Syndrome<br>Congenital Myasthenia   |
| 32. Chronic Inflammatory Demyelinating<br>Polyneuropathies  | 40. Congenital Myopathy   |
|   | 41. Critical illness polyneuropathy and/or myopathy   |
- 

## d

- |   |   |
|---|---|
| 42. Danon Disease<br>Type IIB Glycogenosis  | 46. Desmin Storage Myopathy<br>Myofibrillar Myopathy                  |
| 43. Debranching Enzyme Deficiency<br>Type III Glycogenosis  | 47. Distal Myopathy   |
| 44. Dejerine-Sottas Disease<br>Hereditary Motor and Sensory Neuropathy<br>Charcot-Marie-Tooth Disease | 48. Distal Myopathy, other  |
| 45. Dermatomyositis   | 49. Distal Spinal Muscular Atrophy<br>Spinal Muscular Atrophy, Distal |
|   | 50. Duchenne Muscular Dystrophy                                       |

\* Alternate name for disorder listed below in gray.



e

51. Emery-Dreifuss Muscular Dystrophy
52. Exertional Myoglobinuria
- 

f

53. Facioscapulohumeral Muscular Dystrophy
54. Familial amyloid neuropathy
55. Familial dilated cardiomyopathy with muscular dystrophy
56. Familial Dysautonomia  
Hereditary Sensory and Autonomic Neuropathy  
Riley-Day syndrome
57. Fingerprint Myopathy
58. Forbe's Disease  
Glycogen Storage Disease Type III
59. Friedreich's Ataxia
- 

g

60. Gamstorp Disease  
Hyperkalemic periodic paralysis
61. Giant Axonal Neuropathy
62. Glycogen Storage Disease  
(with neuromuscular involvement)
63. Glycogenosis (with neuromuscular involvement)  
Pompe's disease  
Acid Maltase Deficiency
64. Guillain-Barré Syndrome  
Acute Inflammatory Demyelinating  
Polyradiculoneuropathy
- 

h

65. Hereditary Motor and Sensory Neuropathies Type I  
Peroneal Muscular Atrophy  
Charcot-Marie-Tooth Type I and subtypes
66. Hereditary Motor and Sensory Neuropathies Type II  
Neuronal Type of Peroneal Muscular Atrophy  
Charcot-Marie-Tooth Type II and subtypes

\* Alternate name for disorder listed below in gray.



- |   |  |
|---|--|
| 67. Hereditary Motor and Sensory Neuropathies Type III<br>Dejerine-Sottas disease or Hypertrophic neuropathy of infancy<br>Charcot-Marie-Tooth Type III       | 73. Hereditary Sensory and Autonomic Neuropathy Type II<br>Acroosteolysis                                  |
| 68. Hereditary Motor and Sensory Neuropathies Type IV<br>Refsum's Disease<br>Hypertrophic neuropathy with excess phytanic acid<br>Charcot-Marie-Tooth Type IV | 74. Hereditary Sensory and Autonomic Neuropathy Type III<br>Familial dysautonomia<br>Riley-Day syndrome    |
| 69. Hereditary Motor and Sensory Neuropathy with spastic paraplegia Type V<br>Charcot-Marie-Tooth Disease Type V  | 75. Hereditary Sensory and Autonomic Neuropathy Type IV<br>Congenital insensitivity to pain and anhidrosis |
| 70. Hereditary neuralgic amyotrophy<br>Parsonage-Turner syndrome  | 76. Hyperkalemic Periodic Paralysis<br>Gamstorp Disease  |
| 71. Hereditary Neuropathy with liability to pressure palsy  | 77. Hyperthermia<br>Malignant Hyperthermia   |
| 72. Hereditary Sensory and Autonomic Neuropathy Type I  | 78. Hypokalemic Periodic Paralysis   |
- 

**i**

- |                             |                                       |
|-----------------------------|---------------------------------------|
| 79. Iatrogenic Myopathy     | 81. Inclusion Body Myositis           |
| 80. Inclusion Body Myopathy | 82. Isaac's Syndrome<br>Neuromyotonia |
- 

**k**

- |   |  |
|---|--|
| 83. Kearns-Sayre Syndrome                                 | 85. Kugelberg Welander Disease<br>Spinal Muscular Atrophy Type III |
| 84. Kennedy's Disease<br>Spinal Bulbar (Muscular) Atrophy |  |

\* Alternate name for disorder listed below in gray.



l

- 86. Lac St-Jean Syndrome  
Progressive Polyneuropathy
  - 87. Laing Distal Myopathy
  - 88. Lambert-Eaton (myasthenic) syndrome
  - 89. Landouzy-Dejerine Muscular Dystrophy  
Facioscapulohumeral Muscular Dystrophy Type IA
  - 90. Leigh's syndrome
  - 91. Limb Girdle Muscular Dystrophy
  - 92. Lom hereditary motor and sensory neuropathy  
Charcot-Marie-Tooth Disease
  - 93. Lyme Neuropathy
- 

m

- 94. Macrophagic Myofascitis
- 95. Malignant Hyperthermia
- 96. Manifesting for Duchenne Muscular Dystrophy/  
Becker Muscular Dystrophy  
Manifesting Carrier
- 97. McArdle's Disease  
Myophosphorylase Deficiency  
Glycogen Storage Disease Type V
- 98. Minicore Myopathy
- 99. Mitochondrial myopathy and/or neuropathy  
Mitochondrial neuropathy  
Mitochondrial DNA breakage syndrome secondary  
to nuclear mutation
- 100. Mixed Connective Tissue Overlap Disease
- 101. Miyoshi Myopathy
- 102. Monoclonal gammopathy with neuropathy
- 103. Mononeuritis multiplex
- 104. Multicore Myopathy
- 105. Multifocal motor neuropathy with conduction block
- 106. Muscle-Eye-Brain Disease
- 107. Muscular Dystrophy
- 108. Myasthenia Gravis
- 109. Myoadenylate Deaminase Deficiency
- 110. Myofibrillar Myopathy  
Desmin Storage Myopathy
- 111. Myoglobinuria
- 112. Myophosphorylase Deficiency  
McArdle's disease
- 113. Myositis  
Local Nodular Myositis  
Focal Myositis
- 114. Myotonia Congenita  
Thomsen's disease

\* Alternate name for disorder listed below in gray.



115. Myotonic Muscular Dystrophy

116. Myotonic Muscular Dystrophy Type I  
Steinert's Disease

117. Myotonic Muscular Dystrophy Type II  
Proximal Myotonic Myopathy

118. Myotubular Myopathy  
Centronuclear Myopathy

---

## n

119. Neuromyotonia  
Isaac's Syndrome

120. Nemaline Myopathy

121. Nonaka Myopathy

122. Normokaliemic Periodic Paralysis

---

## o

123. Oculopharyngeal Muscular Dystrophy

124. Olivopontocerebellar Atrophy

125. Ophthalmoplegic Muscular Dystrophy

---

## p

126. Paramyotonia Congenita

127. Paraneoplastic neuropathy

128. Parsonage-Turner syndrome  
Hereditary neuralgic amyotrophy

129. Periodic Paralysis

130. Peripheral Neuropathy and agenesis of  
the corpus callosum  
Charlevoix-Saguenay Disease  
Andermann Syndrome

131. Peroneal Muscular Dystrophy  
Charcot-Marie-Tooth Disease Type I  
Hereditary Motor and Sensory Neuropathies Type I

\* Alternate name for disorder listed below in gray.



132. Phosphofructokinase Deficiency  
Tauri's disease  
Glycogenosis Type VII

133. Polymyositis

134. Pompe's Disease  
Acid Maltase Deficiency  
Glycogenosis Type II

135. Proximal Myotonic Myopathy  
Myotonic Muscular Dystrophy Type II

---

## r

136. Reducing Body Myopathy

137. Refsum's Disease  
Charot-Marie-Tooth Disease  
Hereditary Motor and Sensory Neuropathy

138. Riley-Day Syndrome  
Hereditary Sensory and Autonomic Neuropathy Type III

139. Rippling Muscle Disease

---

## S

140. Sarcotubular Myopathy

141. Scapulooperoneal Muscular Atrophy

142. Schwartz-Jampel syndrome  
Chondrodystrophic Myotonia

143. Spinal Bulbar (Muscular) Atrophy  
Kennedy's Disease

144. Spinal Muscular Atrophy Type I  
Werdnig-Hoffman disease  
Acute Spinal Muscular Atrophy

145. Spinal Muscular Atrophy Type II  
Intermediate Spinal Muscular Atrophy

146. Spinal Muscular Atrophy Type III  
Kugelberg-Welander disease  
Intermediate Spinal Muscular Atrophy

147. Spinal Muscular Atrophy Type IV

148. Spinal Muscular Atrophy, Distal

149. Spinal Muscular Atrophy, other

150. Spinocerebellar Ataxia of Charlevoix-Saguenay  
Autosomal Recessive Spastic Ataxia of  
Charlevoix-Saguenay

151. Steinert's Disease  
Myotonic Muscular Dystrophy Type I

\* Alternate name for disorder listed below in gray.



## t

- |   |   |
|---|---|
| 152. Tangier's Disease<br>Alphalipoproteinemia                                  | 155. Tibial Muscular Dystrophy<br>Udd Distal Myopathy |
| 153. Tauri's Disease<br>Glycogenosis Type VII<br>Phosphofructokinase Deficiency | 156. Tubular Aggregate Myopathy                       |
| 154. Thompsen's Disease<br>Myotonia Congenita                                   |   |
- 

## v

- |                          |                                    |
|--------------------------|------------------------------------|
| 157. Vacuolar Myopathies | 158. Vasculitis-related Myopathies |
|--------------------------|------------------------------------|
- 

## w

- |                                 |  |
|---------------------------------|--|
| 159. Walker-Warburg Syndrome    | 161. Werdnig-Hoffman Disease<br>Spinal Muscular Atrophy Type I |
| 160. Welander's Distal Myopathy |  |
- 

## z

- |                          |  |
|--------------------------|--|
| 162. Zebra Body Myopathy |  |
|--------------------------|--|