

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.

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

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

* Alternate name for disorder listed below in gray.



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| 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
Myotubular Myopathy | 36. Congenital Hypomyelinating neuropathy
Hereditary motor and sensory neuropathy
Charcot-Marie-Tooth Disease |
| 29. Charcot-Marie-Tooth Disease
Hereditary Motor and Sensory Neuropathy
Congenital insensitivity to pain and anhidrosis | 37. Congenital insensitivity to pain and anhidrosis
Hereditary sensory and autonomic neuropathy |
| 30. Charlevoix-Saguenay Syndrome
Peripheral neuropathy
and agenesis of the corpus callosum
Andermann syndrome | 38. Congenital Muscular Dystrophy |
| 31. Chondrodystrophic Myotonia
Schwartz-Jampel Syndrome | 39. Congenital Myasthenic Syndrome
Congenital Myasthenia |
| 32. Chronic Inflammatory Demyelinating
Polyneuropathies | 40. Congenital Myopathy |
| | 41. Critical illness polyneuropathy and/or myopathy |
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| 42. Danon Disease
Type IIB Glycogenosis | 46. Desmin Storage Myopathy
Myofibrillar Myopathy |
| 43. Debranching Enzyme Deficiency
Type III Glycogenosis | 47. Distal Myopathy |
| 44. Dejerine-Sottas Disease
Hereditary Motor and Sensory Neuropathy
Charcot-Marie-Tooth Disease | 48. Distal Myopathy, other |
| 45. Dermatomyositis | 49. Distal Spinal Muscular Atrophy
Spinal Muscular Atrophy, Distal |
| | 50. Duchenne Muscular Dystrophy |

* Alternate name for disorder listed below in gray.



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



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| 67. Hereditary Motor and Sensory Neuropathies Type III
Dejerine-Sottas disease or Hypertrophic neuropathy of infancy
Charcot-Marie-Tooth Type III | 73. Hereditary Sensory and Autonomic Neuropathy Type II
Acroosteolysis |
| 68. Hereditary Motor and Sensory Neuropathies Type IV
Refsum's Disease
Hypertrophic neuropathy with excess phytanic acid
Charcot-Marie-Tooth Type IV | 74. Hereditary Sensory and Autonomic Neuropathy Type III
Familial dysautonomia
Riley-Day syndrome |
| 69. Hereditary Motor and Sensory Neuropathy with spastic paraplegia Type V
Charcot-Marie-Tooth Disease Type V | 75. Hereditary Sensory and Autonomic Neuropathy Type IV
Congenital insensitivity to pain and anhidrosis |
| 70. Hereditary neuralgic amyotrophy
Parsonage-Turner syndrome | 76. Hyperkalemic Periodic Paralysis
Gamstorp Disease |
| 71. Hereditary Neuropathy with liability to pressure palsy | 77. Hyperthermia
Malignant Hyperthermia |
| 72. Hereditary Sensory and Autonomic Neuropathy Type I | 78. Hypokalemic Periodic Paralysis |
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| 79. Iatrogenic Myopathy | 81. Inclusion Body Myositis |
| 80. Inclusion Body Myopathy | 82. Isaac's Syndrome
Neuromyotonia |
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| 83. Kearns-Sayre Syndrome | 85. Kugelberg Welander Disease
Spinal Muscular Atrophy Type III |
| 84. Kennedy's Disease
Spinal Bulbar (Muscular) Atrophy | |

* Alternate name for disorder listed below in gray.



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- 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
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- 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

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119. Neuromyotonia
Isaac's Syndrome

120. Nemaline Myopathy

121. Nonaka Myopathy

122. Normokaliemic Periodic Paralysis

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123. Oculopharyngeal Muscular Dystrophy

124. Olivopontocerebellar Atrophy

125. Ophthalmoplegic Muscular Dystrophy

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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

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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

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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.



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| 152. Tangier's Disease
Alphalipoproteinemia | 155. Tibial Muscular Dystrophy
Udd Distal Myopathy |
| 153. Tauri's Disease
Glycogenosis Type VII
Phosphofructokinase Deficiency | 156. Tubular Aggregate Myopathy |
| 154. Thompsen's Disease
Myotonia Congenita | |
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| 157. Vacuolar Myopathies | 158. Vasculitis-related Myopathies |
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| 159. Walker-Warburg Syndrome | 161. Werdnig-Hoffman Disease
Spinal Muscular Atrophy Type I |
| 160. Welander's Distal Myopathy | |
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| 162. Zebra Body Myopathy |
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