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.
### List of Disorders Covered by Muscular Dystrophy Canada / page 2

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

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<tr>
<td>17.</td>
<td>Barth Syndrome</td>
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<td>Becker Muscular Dystrophy</td>
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<td>Bethlem Myopathy</td>
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<td>21.</td>
<td>Botulism</td>
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<td>22.</td>
<td>Brachial Plexopathy</td>
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</table>
| 23. | Branching Enzyme Deficiency  
   | Glycogen Storage Disease Type IV  
   | Andersen Disease/Syndrome |
| 24. | Brody Disease |

* Alternate name for disorder listed below in gray.
List of Disorders Covered by Muscular Dystrophy Canada / page 3

C

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

D

42. Danon Disease
   Type IIB Glycogenosis
43. Debranching Enzyme Deficiency
   Type III Glycogenosis
44. Dejerine-Sottas Disease
   Hereditary Motor and Sensory Neuropathy
   Charcot-Marie-Tooth Disease
45. Dermatomyositis
46. Desmin Storage Myopathy
   Myofibrillar Myopathy
47. Distal Myopathy
48. Distal Myopathy, other
49. Distal Spinal Muscular Atrophy
   Spinal Muscular Atrophy, Distal
50. Duchenne Muscular Dystrophy
List of Disorders Covered by Muscular Dystrophy Canada / page 4

51. Emery-Dreifuss Muscular Dystrophy
52. Exertional Myoglobinuria

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. Forbes Disease
   Glycogen Storage Disease Type III
59. Friedreich’s Ataxia

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

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
   Dejerine-Sottas disease or Hypertrophic neuropathy of infancy
   Charcot-Marie-Tooth Type III

68. Hereditary Motor and Sensory Neuropathies Type IV
   Refsum’s Disease
   Hypertrophic neuropathy with excess phytanic acid
   Charcot-Marie-Tooth Type IV

69. Hereditary Motor and Sensory Neuropathy with spastic paraplegia Type V
   Charcot-Marie-Tooth Disease Type V

70. Hereditary neuralgic amyotrophy
    Parsonage-Turner syndrome

71. Hereditary Neuropathy with liability to pressure palsy

72. Hereditary Sensory and Autonomic Neuropathy Type I

73. Hereditary Sensory and Autonomic Neuropathy Type II
    Acroosteolysis

74. Hereditary Sensory and Autonomic Neuropathy Type III
    Familial dysautonomia
    Riley-Day syndrome

75. Hereditary Sensory and Autonomic Neuropathy Type IV
    Congenital insensitivity to pain and anhidrosis

76. Hyperkalemic Periodic Paralysis
    Gamstorp Disease

77. Hyperthermia
    Malignant Hyperthermia

78. Hypokalemic Periodic Paralysis

79. Iatrogenic Myopathy

80. Inclusion Body Myopathy

81. Inclusion Body Myositis

82. Isaac’s Syndrome
    Neuromyotonia

83. Kearns-Sayre Syndrome

84. Kennedy’s Disease
    Spinal Bulbar (Muscular) Atrophy

85. Kugelberg Welander Disease
    Spinal Muscular Atrophy Type III

* Alternate name for disorder listed below in gray.
List of Disorders Covered by Muscular Dystrophy Canada / page 6

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

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.
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<td>Peripheral Neuropathy and agenesis of the corpus callosum</td>
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<td>Charlevoix-Saguenay Disease</td>
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<td>Andermann Syndrome</td>
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<td>131</td>
<td>Peroneal Muscular Dystrophy</td>
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<td>Charcot-Marie-Tooth Disease Type I</td>
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<td>Hereditary Motor and Sensory Neuropathies Type I</td>
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* Alternate name for disorder listed below in gray.
132. Phosphofructokinase Deficiency
   Tauri's disease
   Glycogenosis Type VII

133. Polymyositis

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

135. Proximal Myotonic Myopathy
   Myotonic Muscular Dystrophy Type II

136. Reducing Body Myopathy

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

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

139. Rippling Muscle Disease

140. Sarcomubular Myopathy

141. Scapuloperoaneal 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.
List of Disorders Covered by Muscular Dystrophy Canada / page 9

T
152. Tangier's Disease Alphalipoproteinemia
153. Tauri's Disease Glycogenosis Type VII Phosphofructokinase Deficiency
154. Thompsen's Disease Myotonia Congenita

V
157. Vacuolar Myopathies
158. Vasculitis-related Myopathies

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

Z
162. Zebra Body Myopathy

* Alternate name for disorder listed below in gray.
Revised: March 2009