Muscular Dystrophy Canada

1-866-687-2538
www.muscle.ca

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.

National Office:
2345 Yonge Street, Suite 900, Toronto, Ontario M4P 2E5
T 416.488.0030 1.866.MUSCLE.8 F 416.488.7523 W muscle.ca

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1. Abetalipoproteinemia
   Bassen Kornzwieg
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 Talengiectasia
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

b

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

* Alternate name for disorder listed below in gray.

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25. Carnitine Palmityl 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
    Congenital Myasthenia
40. Congenital Myopathy
41. Critical illness polyneuropathy and/or myopathy

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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

* 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.

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

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79. Iatrogenic Myopathy

80. Inclusion Body Myopathy

81. Inclusion Body Myositis

82. Isaac's Syndrome
    Neuromyotonia

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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.

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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
Charcot-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. Scapuloperoneal 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

153. Tauri's Disease
Glycogenosis Type VII
Phosphofructokinase Deficiency

154. Thompsen's Disease
Myotonia Congenita

155. Tibial Muscular Dystrophy
Udd Distal Myopathy

156. Tubular Aggregate Myopathy

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157. Vacuolar Myopathies

158. Vasculitis-related Myopathies

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159. Walker-Warburg Syndrome

160. Welander's Distal Myopathy

161. Werdnig-Hoffman Disease
Spinal Muscular Atrophy Type I

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162. Zebra Body Myopathy

* Alternate name for disorder listed below in gray.

Revised: May 2017