Opis choroby *

Definicja

A rare genetic multi-system disorder characterized by a wide range of muscle-related manifestations (muscle weakness, myotonia, early onset cataracts (before age 50) and systemic manifestations (cerebral, endocrine, cardiac, gastrointestinal tract, uterus, skin and immunologic involvement) that vary depending on the age of onset. The very wide clinical spectrum ranges from lethal presentations in infancy to mild, late-onset disease.

Dane

Klasyfikacja
Choroba

Synonimy
Myotonic dystrophy type 1
Choroba Steinerta
DM1
Dystrofia miotoniczna typu 1
MD1
Steinert disease

Kod ORPHA
273

Kod OMIM
160900

Kod ICD10
G71.1

Kod ICD11
8C71.0

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*Źródło

orphanet