

## 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