Opis choroby *

Definicja

A rare congenital myopathy characterized by early onset of severe muscular weakness, respiratory distress due to diaphragmatic paralysis, dysphagia and areflexia, joint contractures, and scoliosis. Decreased fetal movements are seen in some individuals. Muscle biopsy may show a combination of dystrophic and myopathic features. The clinical course is variable, with some patients becoming ventilator-dependent and never achieving ambulation.

Dane

Klasyfikacja
Choroba

Synonimy
EMARDD
EMARDD

Kod ORPHA
439212

Kod OMIM
614399

Kod ICD10
G71.2

Kod ICD11
-

---

*Źródło

orphanet