

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

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*Źródło

orphanet