

Opis choroby *

Definicja

A rare genetic motor neuron disease characterized by decreased or absent fetal movements, congenital proximal and distal joint contractures (consistent with arthrogryposis multiplex congenita), and multiple congenital fractures of the long bones. Further manifestations are neonatal respiratory distress, severe muscular hypotonia, areflexia, dysphagia, congenital heart defects, and dysmorphic facial features. Muscle biopsy shows increased fiber-size variation and grouping of larger type I fibers. The disease is usually fatal in infancy due to respiratory failure.

Dane

Klasyfikacja

Choroba

Synonimy

SMABF

SMABF

Kod ORPHA

486811

Kod OMIM

616867

Kod ICD10

G12.8

Kod ICD11

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*Źródło

orphanet