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