Opis choroby *

Definicja

A rare defect of tropomyosin characterized by decreased fetal movements and generalized muscle stiffness at birth. Additional features include joint contractures, short stature, kyphosis, dysmorphic features, temperature dysregulation, and variably severe respiratory involvement with hypoxemia. Muscle biopsy shows mild myopathic features.

Dane

Klasyfikacja
Choroba

Kod ORPHA
476406

Kod OMIM
-

Kod ICD10
G71.2

Kod ICD11
-

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*Źródło

orphanet