

## 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

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#### Kod ICD10

G71.2

#### Kod ICD11

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### \*Źródło

orphanet