

## Opis choroby \*

### Definicja

A rare genetic neuromuscular disease characterized by neonatal or infancy onset of delayed motor development, generalized muscle weakness involving also the facial muscles, pseudohypertrophy of lower limb muscles, and joint contractures, associated with childhood onset of rapidly progressive dilated cardiomyopathy with arrhythmias leading to sudden cardiac death. Muscle biopsy in early childhood shows minicore-like lesions and centralized nuclei, with dystrophic features being more conspicuous in the second decade of life.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

EOMFC

Salih myopathy

EOMFC

Salih myopathy

#### Kod ORPHA

289377

#### Kod OMIM

611705

#### Kod ICD10

G71.8

#### Kod ICD11

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#### \*Źródło

orphanet