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