Opis choroby *

Definicja

A rare congenital muscular dystrophy characterized by early onset of hypotonia, delayed motor development, and variably progressive generalized muscle weakness. Predominant involvement of pelvic and neck flexor muscles has been reported, as well as early involvement of hamstrings and medial gastrocnemius visible on muscle MRI. Serum creatine kinase levels are markedly elevated (in some cases already from early childhood). Muscle biopsy shows absence of dysferlin.

Dane

Klasyfikacja
Choroba

Kod ORPHA
199329

Kod OMIM
-

Kod ICD10
G71.2

Kod ICD11
-

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*Źródło

orphanet