Opis choroby *

Definicja

A rare, genetic, skeletal muscle disease characterized by an early-onset hypotonia, muscle weakness, global developmental delay with intellectual disability, and cardiomyopathy. Congenital structural heart defects and ichthyosiform cutaneous lesions have also been associated. Muscle biopsy shows characteristic enlarged mitochondria located at the periphery of muscle fibers.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital megaconial myopathy
Wrodzona dystrofia mięśni typu megaconial
Wrodzona dystrofia mięśniowa z zaburzeniami struktury mitochondriów
Wrodzona miopatia typu megaconial
Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect
Congenital muscular dystrophy with mitochondrial structural abnormalities

Kod ORPHA
280671

Kod OMIM
602541

Kod ICD10
G71.2

Kod ICD11
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*Źródło

orphanet