Opis choroby *

Definicja

Congenital myopathy with myasthenic-like onset is a rare, genetic, non-dystrophic myopathy characterized by fatigable muscle weakness associated with congenital myopathy. Patients present with axial hypotonia, myopathic facies with fatigable ptosis, feeding difficulties, delayed gross motor development and proximal limb weakness with a RYR1-related typical pattern of muscle involvement (i.e. severe involvement of the soleus muscle and sparring of the rectus femoris, sartorius, gracilis and semitendinous muscles). Scoliosis and frequent respiratory tract infections are additional observed features.

Dane

Klasyfikacja
Choroba

Kod ORPHA
424107

Kod OMIM
-

Kod ICD10
G71.2

Kod ICD11
-

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*Źródło

orphanet