

# Miopatia wrodzona o początku podobnym do miastenii

**Kod Orpha: 424107 Kod OMIM:**

## 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 sparing 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**

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**Kod ICD10**

G71.2

**Kod ICD11**

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[\\*Źródło](#)

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## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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