Opis choroby *

Definicja

Oculogastrointestinal muscular dystrophy is an extremely rare autosomal recessively inherited neuromuscular disease characterized by ocular manifestations such as ptosis and diplopia followed by chronic diarrhea, malnutrion and intestinal peudo-obstruction.

Dane

Klasyfikacja
Choroba

Synonimy
Visceral myopathy-familial external ophthalmoplegia syndrome
Miopatia trzewna - rodzinna zewnętrzna oftalmoplegia

Kod ORPHA
1876

Kod OMIM
277320

Kod ICD10
G71.0

Kod ICD11
DA90.2

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*Źródło

orphanet