

Opis choroby *

Definicja

A rare, genetic, mitochondrial DNA-related mitochondrial myopathy disorder characterized by slowly progressive muscular weakness (proximal greater than distal), predominantly involving the facial muscles and scapular girdle, associated with insulin-dependent diabetes mellitus. Neurological involvement and congenital myopathy may be variably observed.

Dane

Klasyfikacja

Choroba

Kod ORPHA

2596

Kod OMIM

500002

Kod ICD10

G71.3

Kod ICD11

8C73.Y

*Źródło

orphanet