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

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*Źródło

orphanet