

Opis choroby *

Definicja

A rare, genetic, neuromuscular disease characterized by adult-onset muscle weakness and atrophy in a scapulo-peroneal distribution, mild involvement of the facial muscles, dysphagia, and gynecomastia. Elevated serum CK levels and mixed myopathic and neurogenic abnormalities are associated clinical findings.

Dane

Klasyfikacja

Choroba

Synonimy

Kaeser syndrome

Zespół Kaesera

Zespół Starka i Kaesera

Stark-Kaeser syndrome

Kod ORPHA

85146

Kod OMIM

181400

Kod ICD10

G12.1

Kod ICD11

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*Źródło

orphanet