Opis choroby *

Definicja

A rare genetic disease characterized by the association of Klippel-Feil anomaly (fusion of the cervical spine), myopathy, hypotonia, short stature, microcephaly, and facial dysmorphism (including low-set ears, bulbous nose, long philtrum, high-arched palate, and low posterior hairline, among others). Cardiac abnormalities and various skeletal anomalies (such as pectus excavatum or clinodactyly) have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
447974

Kod OMIM
616549

Kod ICD10
Q76.1

Kod ICD11
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*Źródło

orphanet