## 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