## Opis choroby *

Definicja

A rare disorder characterized by pterygium colli, digital anomalies (abnormal small thumbs, widened interphalangeal joints, and broad terminal phalanges), and craniofacial abnormalities (brachycephaly, epicanthic folds, angulated eyebrows, upward slanting of the palpebral fissures, ptosis, hypertelorism, and prominent low-set, posteriorly rotated ears). It has been described in a woman and her son, but the manifestations were much less severe in the mother. The son also had intellectual deficit. The inheritance is either X-linked dominant or autosomal dominant.

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Khalifa-Graham syndrome
Zespół Khalifa i Grahama

**Kod ORPHA**
2988

**Kod OMIM**
600159

**Kod ICD10**
Q87.0

**Kod ICD11**
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*Źródło

orphanet