

Zespół Kuskokwim

Kod Orpha: 1149 Kod OMIM: 259450

Opis choroby *

Definicja

A very rare congenital contracture disorder, reported exclusively in Yup'ik Eskimos of the Kuskokwim River delta region of Alaska, characterized by multiple contractures of large joints (predominantly the knees and ankles) that present at birth or during childhood but are lifelong; deformities of the spine, pelvis and feet; and sometimes proximally or distally displaced patellae and muscle atrophy in the limbs with contractures. Additional radiological features include mild vertebral wedging, elongation of the vertebral pedicle, and clubbing of the distal clavicle. An autosomal recessive pattern of inheritance has been suggested.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Arthrogryposis-like syndrome
Choroba Kuskokwim
Kuskokwim disease

Kod ORPHA

1149

Kod OMIM

259450

Kod ICD10

Q87.2

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl