

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	Synonimy
Zespół wad wrodzonych	Arthrogryposis-like syndrome
	Choroba Kuskokwim
	Kuskokwim disease

Kod ORPHA	Kod OMIM	Kod ICD10
1149	259450	Q87.2

Kod ICD11

-

*Źródło

orphanet