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Kuskokwim syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Kuskokwim syndrome. ORPHA:1149*

Arthrogryposis-like syndrome, also known as Kuskokwim disease, is 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.