Opis choroby *

Definicja

A rare arthrogryposis syndrome characterized by the association of multiple congenital joint contractures (of the large joints, fingers and toes) and hyperkeratosis (i.e. thick, scaling and fissured skin), with death occurring in early infancy. There have been no further reports in the literature since 1993.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Johnston-Aarons-Schelley syndrome
Zespół Johnstona, Aaronsa i Schelley

Kod ORPHA
1485

Kod OMIM
208158

Kod ICD10
Q68.8

Kod ICD11
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*Źródło

orphanet