Artrogrypoza - hiperkeratoza forma śmiertelna
Kod Orpha: 1485 Kod OMIM: 208158

Opis choroby *

Definition
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

Classification
Zespół wad wrodzonych

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

ORPHA code
1485

OMIM code
208158

ICD10 code
Q68.8

ICD11 code
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.