Opis choroby *

Definicja

A rare, genetic developmental defect during embryogenesis syndrome characterized by camptodactyly, joint contractures with amyotrophy, and ectodermal anomalies (oligodontia, enamel abnormalities, longitudinally broken nails, hypohidrotic skin with tendency to excessive bruising and scarring after injuries and scratching), as well as growth retardation, kyphoscoliosis, mild facial dysmorphism, and microcephaly. There have been no further descriptions in the literature since 1992.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Stoll-Alembik-Finck syndrome
Artrogrypoza - dysplazja ektodermalna - inne wady

Kod ORPHA
3200

Kod OMIM
601701

Kod ICD10
Q68.8

Kod ICD11
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*Źródło

orphanet