

Zespół Stolla, Alembika i Fincka

Kod Orpha: 3200 Kod OMIM: 601701

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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.