

## 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 Stoll-Alembik-Finck syndrome

#### Synonimy

Artrogrypoza - dysplazja ektodermalna - inne wady

#### Kod ORPHA

3200

#### Kod OMIM

601701

#### Kod ICD10

Q68.8

#### Kod ICD11

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#### \*Źródło

orphanet