

# Kamptodaktylia - taurynuria

**Kod Orpha: 1325 Kod OMIM:**

## Opis choroby \*

### Definicja

Camptodactyly-aurinuria syndrome is a congenital malformation syndrome characterized by the association of a permanent camptodactyly of the fingers (see this term) with the over excretion of taurine in the urine. Camptodactyly mainly affects the little finger, although any finger may be involved. The disease has been described in 17 affected patients from 4 unrelated families. An autosomal dominant inheritance has been suggested. There have been no further descriptions in the literature since 1966.

### Dane

#### Klasyfikacja

Zespół wad wrodzonych

#### Synonimy

Familial streblodactyly with amino-aciduria  
Rodzinna streblodaktylia z aminoacydurią

#### Kod ORPHA

1325

#### Kod OMIM

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#### Kod ICD10

Q68.1

#### Kod ICD11

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[\\*Źródło](#)

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.