Kamptodaktylia - taurynuria
Kod Orpha: 1325 Kod OMIM:

Opis choroby *

Definicja
Camptodactyly-taurinuria 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
-

Kod ICD10
Q68.1

Kod ICD11
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.