Zespół Mononena, Karnesa i Senac
Kod Orpha: 2565 Kod OMIM: 301940

Opis choroby *

Definicja
Mononen-Karnes-Senac syndrome is characterized by skeletal dysplasia associated with finger malformations (brachydactyly with short and abducted thumbs, short index fingers, and markedly short and abducted great toes), variable mild short stature, and mild bowleg with overgrowth of the fibula. It has been described in two males, their mothers, and a maternal aunt. Females are less severely affected than males. X-linked dominant inheritance is suggested.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Skeletal dysplasia-brachydactyly syndrome
Dysplazja szkieletowa - brachydaktylia

Kod ORPHA
2565

Kod OMIM
301940

Kod ICD10
Q87.5

Kod ICD11
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.