

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
Skeletal dysplasia-brachydactyly syndrome
Dysplazja szkieletowa - brachydaktylia

Synonimy

Kod ORPHA

2565

Kod OMIM

301940

Kod ICD10

Q87.5

Kod ICD11

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*Źródło

orphanet