

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	Synonimy
Zespół wad wrodzonych	Skeletal dysplasia-brachydactyly syndrome Dysplazja szkieletowa - brachydaktylia

Kod ORPHA	Kod OMIM	Kod ICD10
2565	301940	Q87.5

Kod ICD11
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*Źródło

orphanet