

Opis choroby *

Definicja

A rare autosomal dominant heart-hand syndrome that is characterized by bisymmetric brachydactyly accompanied by long thumbs, joint anomalies (restriction of motion at the shoulder and metacarpophalangeal joints) and cardiac conduction defects. Additional features include small hands and feet, clinodactyly, narrow shoulders with short clavicles, pectus excavatum and mild shortness of the limbs, cardiomegaly and murmur of pulmonic stenosis. There have been no new reports since 1981.

Dane

Klasyfikacja

Synonimy

Zespół wad wrodzonych Brachydactyly, long thumb type

Brachydaktylia - długi kciuk, typu długiego kciuka

Kod ORPHA

2946

Kod OMIM

112430

Kod ICD10

Q87.2

Kod ICD11

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*Źródło

orphanet