

Opis choroby *

Definicja

Heart-hand syndrome type 2 is an extremely rare heart-hand syndrome (see this term) described in two families to date, that is characterized by upper limb malformations (brachytelephalangy type D, hypoplastic deltoids, mild shortening of the fourth and fifth metacarpals in some individuals, skeletal anomalies in the humerus, radius, ulnae, and thenar bones) and cardiac arrhythmias (junctional rhythms and atrial fibrillation).

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Atriodigital dysplasia type 2

Dysplazja przedsionkowo-palcowa typu 2

Zespół Tabatznika

Tabatznik syndrome

Kod ORPHA

1350

Kod OMIM

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Kod ICD10

Q87.2

Kod ICD11

LD2F.1Y

*Źródło

orphanet