

Opis choroby *

Definicja

Carney complex-trismus-pseudocamptodactyly syndrome is a rare genetic heart-hand syndrome characterized by typical manifestations of the Carney complex (spotty pigmentation of the skin, familial cardiac and cutaneous myxomas and endocrinopathy) associated with trismus and distal arthrogryposis (presenting as involuntary contraction of distal and proximal interphalangeal joints of hands evident only on dorsiflexion of wrist and similar lower-limb contractures producing foot deformities).

Dane

Klasyfikacja

Choroba

Synonimy

Carney complex variant

Wariant kompleksu Carney'a

Kod ORPHA

319340

Kod OMIM

608837

Kod ICD10

Q68.8

Kod ICD11

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*Źródło

orphanet