

Opis choroby *

Definicja

A rare autosomal dominant form of heart-hand syndrome that is characterized by adult onset, progressive cardiac conduction disease, tachyarrhythmias that can lead to sudden death, dilated cardiomyopathy and brachydactyly, with the hands less severely affected than the feet. Muscle weakness and/or myopathic electromyographic findings have been observed in some cases.

Dane

Klasyfikacja

Zespół wad wrodzonych Atriodigital dysplasia, Slovenian type

Synonimy

Choroba przewodzenia w sercu - kardiomiopatia rozstrzeniowa - brachydaktylia
Dysplazja przedsionkowo-palcowa, typ słoweński
Cardiac conduction disease-dilated cardiomyopathy-brachydactyly syndrome

Kod ORPHA

168796

Kod OMIM

610140

Kod ICD10

Q87.2

Kod ICD11

LD2F.1Y

*Źródło

orphanet