Opis choroby *

Definicja

A rare, multiple congenital anomalies syndrome with cardiac involvement as a major feature characterized by QT prolongation, congenital heart defects, syndactyly, facial dysmorphism and neurodevelopmental features. There are three clinical phenotypes recognized, the classical types that present with a prolonged QT interval and either with (TS1) or without (TS2) cutaneous syndactyly of fingers and toes. The atypical form (ATS) causes multi-system health concerns but not necessarily with prolonged QT.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
LQT8
LQT8
Zespół wydłużonego QT - syndaktylia
Zespół wydłużonego QT typu 8
Long QT syndrome type 8
Long QT syndrome-syndactyly syndrome

Kod ORPHA
65283

Kod OMIM
601005

Kod ICD10
I49.8

Kod ICD11
BC65.0

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*Źródło

orphanet