DISEASE:
Timothy syndrome

NAME:	Timothy syndrome
DESCRIPTION:	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.
ORPHACODE:	65283
SYNONYMS:	LQT8 Long QT syndrome type 8 Long QT syndrome-syndactyly syndrome
XREF(S):	Orphanet OMIM ICD-10 MeSH OMIM
ANALYTE(S):	CACNA1C
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