DISEASE:
Sinoatrial node dysfunction and deafness

NAME:	Sinoatrial node dysfunction and deafness
DESCRIPTION:	Sinoatrial node dysfunction and deafness is a rare genetic disease characterized by congenital severe to profound deafness with no evidence of vestibular dysfunction, associated with sinoatrial node dysfunction with pronounced bradycardia and increased variability of heart rate at rest and episodic syncopes that may be triggered by enhanced physical activity and stress.
ORPHACODE:	324321
SYNONYMS:	Sinoatrial node dysfunction and hearing loss
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CACNA1D
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Cardiopathies, hereditary (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- calcium voltage-gated channel subunit alpha1 D

Related Gene Panels

- Cardiopathies, hereditary (102 genes) - KUL