Opis choroby *

Definicja

A rare genetic disease characterized by intellectual disability, developmental delay, language deficits, and cardiac arrhythmia (most commonly sick sinus syndrome). Additional reported features include epilepsy, hypotonia, retinal abnormalities, nystagmus, attention deficit hyperactivity disorder, autism, and gastroesophageal reflux. The severity of the phenotype is highly variable.

Dane

Klasyfikacja
Choroba

Kod ORPHA
542306

Kod OMIM
617173

Kod ICD10
I49.8

Kod ICD11
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*Źródło

orphanet