Opis choroby *

Definicja

A rare, genetic developmental defect during embryogenesis disorder characterized by sensorineural hearing impairment, childhood-onset cataract, underdeveloped secondary sexual characteristics, spinal muscular atrophy, growth retardation, and cardiac and skeletal anomalies. Sudden death, as well as fatal cardiomyopathy and heart failure, have been described in some cases.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Deafness-cataract-skeletal anomalies syndrome
Głuchota - zaćma - wady szkieletu
Sensorineural hearing loss-cataract-skeletal anomalies-cardiomyopathy syndrome

Kod ORPHA
2663

Kod OMIM
255990

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet