

Opis choroby *

Definicja

Cataract-congenital heart disease-neural tube defect syndrome is a multiple congenital anomaly syndrome characterized by sacral neural tube defects resulting in tethered cord, atrial and/or ventricular septal heart defects (that are detected in infancy), bilateral, symmetrical hyperopia, rapidly progressive early childhood cataracts, bilateral aphakic glaucoma, and abnormal facial features (low frontal hairline, small ears, short philtrum, prominent, widely spaced central incisors, and micrognathia). Hypotonia, growth and developmental delay, seizures, and joint limitation are also reported.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA	Kod OMIM	Kod ICD10
314993	608227	Q87.8
Kod ICD11		
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*Źródło

orphanet