Opis choroby *

Definicja

A rare syndromic cardiac disease characterized by communicating hydrocephalus, endocardial fibroelastosis, and congenital cataracts. A history of upper respiratory infection in the mother during the first trimester of pregnancy and polyhydramnios in the third trimester has been associated. No evience of toxoplasmosis, rubella, cytomegalovirus, herpes simplex virus, syphilis, and galactosemia is reported. There have been no further descriptions in the literature since 1995.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hydrocephalus-endocardial fibroelastosis-cataract syndrome
Wodogłowie - fibroelastoza endokardialna - zaćma

Kod ORPHA
2119

Kod OMIM
600559

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet