

## 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 evidence 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 Hydrocephalus-endocardial fibroelastosis-cataract syndrome  
Wodogłowie - fibroelastoza endokardialna -  
zaćma

#### Synonimy

#### Kod ORPHA

2119

#### Kod OMIM

600559

#### Kod ICD10

Q87.8

#### Kod ICD11

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#### \*Źródło

orphanet