

## Opis choroby \*

### Definicja

A rare genetic syndrome with a central nervous system malformation as a major feature, characterized by a triad of high alpha-fetoprotein levels in both maternal serum and amniotic fluid, cerebral ventriculomegaly, and renal macro- and microcysts. Variable findings include congenital nephrotic syndrome, aqueductal stenosis, gray matter heterotopias, and cardiac malformations, among others.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Congenital nephrosis-cerebral ventriculomegaly syndrome  
VMCKD  
Wrodzona nefroza z wentrikulomegalią mózgową  
VMCKD

#### Kod ORPHA

443988

#### Kod OMIM

219730

#### Kod ICD10

Q04.8

#### Kod ICD11

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#### \*Źródło

orphanet