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