DISEASE:
Ventriculomegaly-cystic kidney disease

NAME:	Ventriculomegaly-cystic kidney disease
DESCRIPTION:	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.
ORPHACODE:	443988
SYNONYMS:	Congenital nephrosis-cerebral ventriculomegaly syndrome VMCKD
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	CRB2
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- crumbs cell polarity complex component 2

Related Gene Panels

- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG