Fowler syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Fowler syndrome. ORPHA:221126

A rare, genetic neurological disorder characterized by hydranencephaly, distinctive glomeruloid vasculopathy in the central nervous system and retina, polyhydramnios and fetal akinesia with arthrogryposis. The disorder is usually prenatally lethal. In rare reported cases that survived beyond infancy, severe intellectual and neurologic disability with seizures, microcephaly and absence of functional movements were reported.