Chudley-McCullough syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Chudley-McCullough syndrome. ORPHA:314597

Chudley-McCullough syndrome is a rare, genetic, syndromic deafness characterized by severe to profound, bilateral, sensorineural hearing loss (congenital or rapidly progressive in infancy) associated with a complex brain malformation including hydrocephalus, varying degrees of partial corpus callosum agenesis, colpocephaly, cerebral and cerebellar cortical dysplasia (bilateral medial frontal polymicrogyria, bilateral frontal subcortical heteropia) and, in some, arachnoid cysts. Major physical abnormalities or psychomotor delay are usually not associated.