Hydrocephaly-cerebellar agenesis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hydrocephaly-cerebellar agenesis syndrome. ORPHA:1397

This syndrome is characterised by infantile hypotonia followed by onset of ataxia, cataract and intellectual deficit by preschool age. Cerebral atrophy was also reported.