Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Craniosynostosis-hydrocephalus-Arnold-Chiari malformation type I-radioulnar synostosis syndrome. ORPHA:171839

Capra-DeMarco syndrome is characterized by sagittal craniosynostosis, hydrocephalus, Chiari I malformation and radioulnar synostosis. Other clinical findings include blepharophimosis, small low-set ears, hypoplastic philtrum, kidney malformation, and hypogenitalism.