Hunter-McAlpine craniosynostosis

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hunter-McAlpine craniosynostosis. ORPHA:97340

Hunter-McAlpine craniosynostosis is characterised by craniosynostosis, intellectual deficit, short stature, facial dysmorphism (oval face with almond-shaped palpebral fissures, droopy eyelids and a small nose) and minor distal anomalies. It has been described in 10 patients. Transmission is autosomal dominant and the syndrome is associated with partial duplication of the long arm of chromosome 5 (5q35-5qter).