Open Peer Review on Qeios

Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hydrocephalus-costovertebral dysplasia-Sprengel anomaly syndrome. ORPHA:2180

This syndrome is characterised principally by Sprengel anomaly (upward displacement of the scapula) and hydrocephaly. Other anomalies such as psychomotor retardation, psychosis, brachydactyly, and costovertebral dysplasia may also be present.