Microcephaly-cervical spine fusion anomalies syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microcephaly-cervical spine fusion anomalies syndrome. ORPHA:2522

Microcephaly-cervical spine fusion anomalies syndrome is characterized by microcephaly, facial dysmorphism (beaked nose, low-set ears, downslanting palpebral fissures, micrognathia), mild intellectual deficit, short stature, and cervical spine fusion anomalies producing spinal cord compression. It has been described in two brothers born to consanguineous parents. Transmission is likely to be autosomal recessive.