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Macrocephaly-short stature-paraplegia syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Macrocephaly-short stature-paraplegia syndrome. ORPHA:2427

Macrocephaly-short stature-paraplegia syndrome is characterized by macrocephaly and midface hypoplasia, intellectual deficit, short stature, spastic paraplegia and severe central nervous system anomalies (hydrocephalus and Dandy-Walker malformation). It has been described in two unrelated adults.