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Short stature-pituitary and cerebellar defects-small sella turcica syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Short stature-pituitary and cerebellar defects-small sella turcica syndrome. ORPHA:85442*

Short stature-pituitary and cerebellar defects-small sella turcica syndrome is characterised by short stature, anterior pituitary hormone deficiency, small sella turcica, and a hypoplastic anterior hypophysis associated with pointed cerebellar tonsils. It has been described in three generations of a large French kindred. Ectopia of the posterior hypophysis was observed in some patients. The syndrome is transmitted as a dominantly inherited trait and is caused by a germline mutation within the LIM-homeobox transcription factor LHX4 gene (1q25).