

Opis choroby *

Definicja

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).

Dane

Klasyfikacja

Choroba

Kod ORPHA

85442

Kod OMIM

262700

Kod ICD10

E23.0

Kod ICD11

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*Źródło

orphanet