

Niski wzrost - wady mózdzku i przysadki - małe siodło tureckie

Kod Orpha: 85442 Kod OMIM: 262700

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 <i>LHX4</i> gene (1q25).

Dane

Klasyfikacja

Choroba

Kod ORPHA

85442

Kod OMIM

262700

Kod ICD10

E23.0

Kod ICD11

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[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie www.orphanet.pl