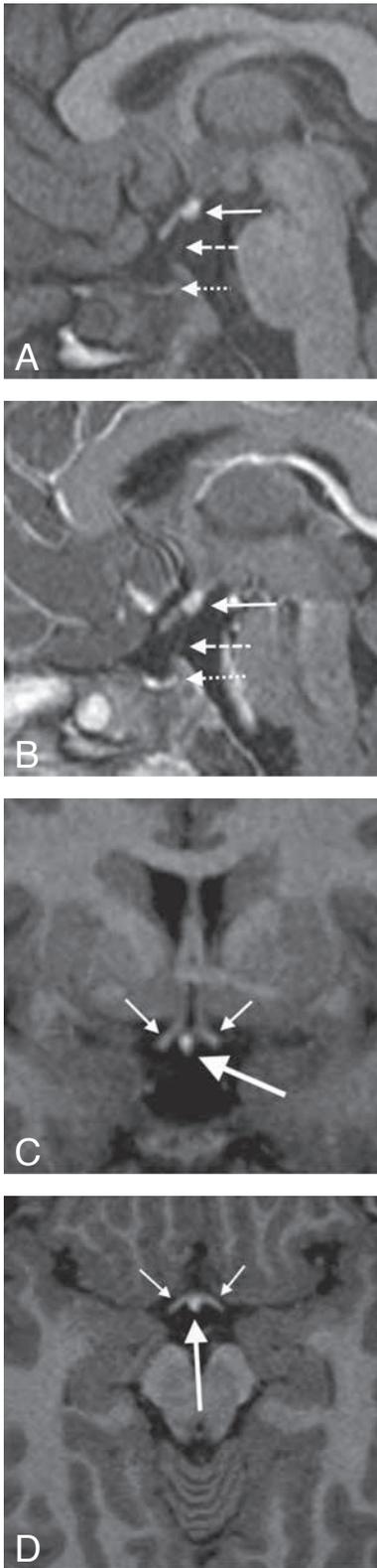


IMAGES IN CLINICAL RADIOLOGY



Pituitary stalk interruption syndrome

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A 5-year-old boy presented at the department of Pediatrics because of parental concerns about his delayed growth which had been slowing since the age of 2.5 years. The patient had no significant familial, perinatal or past medical history. At clinical examination, no abnormalities were present. His stature was below the 5th percentile. Laboratory investigations showed normal serum values of TSH, FT4 and prolactin, but an abnormally low IGF-1 concentration. Bone age (Grulich and Pyle) was 3 years and 6 months, more than one year below his chronological age. MRI of the brain was performed to rule out a hypothalamic-hypophyseal lesion (Fig. A-D). Midsagittal T1-weighted MR imaging revealed a small sella with a hypoplastic anterior pituitary gland (dotted arrow). The pituitary stalk was extremely hypoplastic and barely perceptible (dashed arrow). An ectopic posterior pituitary gland (solid arrow) was observed as an area of T1 high signal intensity at the median eminence in the floor of the third ventricle between the right and left optic tract (small arrows).

Comment

Pituitary stalk interruption syndrome (PSIS) is a congenital midline defect characterized by the triad of a small or absent anterior pituitary gland, a small or absent pituitary stalk and an ectopic posterior pituitary (EPP) lobe located at the median eminence in the floor of the third ventricle. In some patients, the abnormalities may be limited to EPP or to an interrupted pituitary stalk.

PSIS is associated with several midline and/or cerebral malformations and with pituitary endocrinopathy. Contrary to what one might expect, the most frequent presentation of the latter is not diabetes insipidus, but either isolated growth hormone deficiency (IGHD) or multiple anterior pituitary hormone deficiency (MPHD). The endocrine outcome seems to be a progressive onset of pituitary hormone deficiencies leading to panhypopituitarism, usually with maintenance of the posterior pituitary function. In general, MPDH is more frequent in cases of EPP with complete absence of the pituitary stalk, whereas IGHD is more common in cases of EPP with a visible but hypoplastic pituitary stalk.

Cerebral malformations associated with PSIS include periventricular nodular heterotopia, bilateral perisylvian syndrome (polymicrogyria of both lips of the Sylvian fissure), cerebellar dysgenesis and septo-optic-pituitary dyplasia (bilateral optic nerve hypoplasia and agenesis of the septum pellucidum). Extrapituitary abnormalities include renal anomalies, such as unilateral renal agenesis or hypoplasia, and dental anomalies, such as single central incisors.

The causes of PSIS are still unknown. Mechanical pituitary stalk rupture or pituitary stalk ischemia during breech delivery have long been implicated as major causes of PSIS. More recent studies favor defects during early embryogenesis. Those defects may be genetic or the result of exposure to environmental factors during pregnancy. This theory of abnormal organogenesis is supported by the presence of other structural brain and/or body malformations in some patients and by the existence of rare familial forms of PSIS.

MR imaging is the technique of choice in the diagnosis of children with IGHD or MGHD. If PSIS is diagnosed from MRI, there is no other possible diagnosis. The treatment is based on the replacement of the deficient hormones. In case of a partial or complete pituitary stalk agenesis, a close follow-up for evolving pituitary hormone deficiencies is needed. As patients progress from IGHD to MPHD, the stalk and adenohypophysis tend to become smaller.

Reference

1. Kulkarni C., Moorthy S., Pullara S.K., Rajeshkannan R., Unnikrishnan A.G.: Pituitary stalk transection syndrome: Comparison of clinico-radiological features in adults and children with review of literature. *Indian J Radiol Imaging*, 2012, 22: 182-185.