Photo Clinic

Missing Pituitary Stalk: A Key to the Diagnosis

A 17-year-old female patient referred to the outpatient department for an evaluation of her short stature. In her physical examination, weight and height were less than 3 percentile for age. Delayed puberty was diagnosed as Tanner breast and pubic hair stage II. Lab data showed normal results for complete blood count, thyroid function test, fasting blood sugar, and serum electrolytes. The hormonal assay revealed low levels of basal luteinizing and follicle-stimulating hormones, as well as abnormal insulin-like growth factor 1 (IGF-1), and growth hormone stimulation tests. Wrist X-ray was performed, that confirmed significantly delayed bone age.

As part of the investigation, hypothalamic-pituitary axis magnetic resonance imaging (MRI) without and with contrast injection were performed. The sagittal T1-weighted image before and after the contrast injection showed a relatively small hypophysis and an absent pituitary stalk (figure 1). The posterior coronal section through the sella demonstrated a small round hyperintense focus in suprasellar location, which together with the absence of normally located bright signals of the neurohypophysis and the pituitary stalk on the sagittal images, was indicative of an ectopic neurohypophysis. The MRI findings were in keeping with the diagnosis of pituitary stalk interruption syndrome (PSIS). Based on the clinical and paraclinical findings, the diagnosis was made and hormone replacement therapy was started with a significant growth spurt at a 6-month follow-up.

PSIS is a rare congenital disorder characterized by the absence or hypoplasia of the pituitary stalk, the hypoplasia or aplasia of the anterior pituitary, and an ectopic posterior pituitary lobe. The definite pathogenesis of PSIS is still unclear; however, several mechanisms including gene mutations resulting in abnormal embryonic development and environmental factors such as perinatal injury have been postulated as possible etiologies.[1]

The clinical presentation depends on age; nonetheless, many patients present with short stature and anterior pituitary hormone deficiencies. The most common hormonal abnormality is the deficiency of growth hormone, followed by the deficiency of gonadotropin, corticotrophin, and thyrotropin hormones.[2]

Growth hormone therapy is the standard necessary and effective treatment, even in those with delayed diagnosis. Moreover, hormonal evaluations should be performed regularly as hormonal deficiencies are worsened in patients with PSIS.[3]

A written informed consent was given by the patient's father.

Conflict of Interest: None declared.

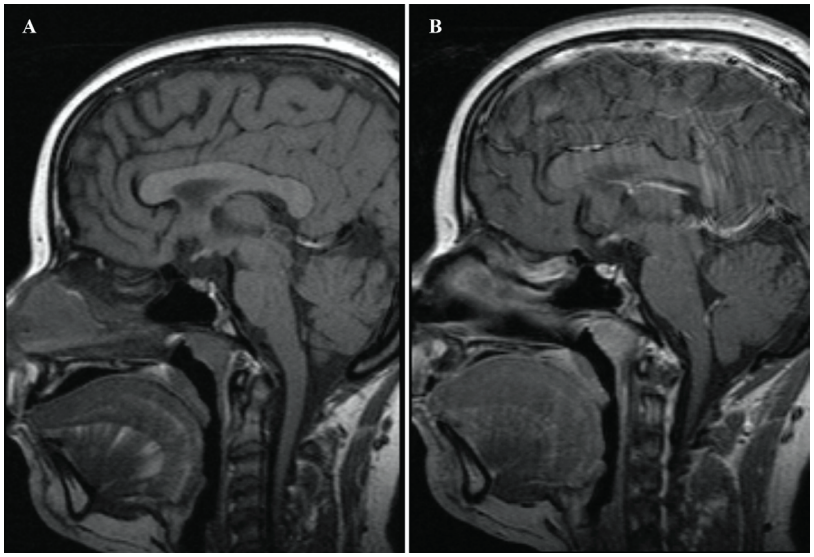

Figure 1: A & B) Sagittal T1-weighted images with and without contrast injection through the sella demonstrate loss of the normal bright spot of the neurohypophysis and an absent pituitary stalk.

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