

Pituitary Stalk Interruption Syndrome: A Case Report Highlighting Key Imaging Features

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Abstract

Case Report

Background: Pituitary stalk interruption syndrome (PSIS) is a rare congenital abnormality of pituitary development, typically diagnosed on magnetic resonance imaging (MRI). It is defined by a thin, discontinuous, or absent pituitary stalk, hypoplasia or aplasia of the anterior pituitary, and an ectopic posterior pituitary gland. This case involves a 38-year-old woman with autoimmune polyendocrinopathy who underwent pituitary MRI for routine assessment of her condition, leading to the incidental discovery of pituitary stalk interruption syndrome.

Keywords: Pituitary Stalk Interruption Syndrome, Autoimmune Polyendocrinopathy, Posterior Pituitary Ectopia, MRI, Hypogonadotropic Hypogonadism.

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INTRODUCTION

Pituitary stalk interruption syndrome (PSIS) represents an uncommon congenital malformation, presenting with variable degrees of anterior pituitary hormone deficiency [1, 2]. The reported incidence is about 0.5 cases per 100,000 live births [3, 4].

The precise etiology of PSIS is still undetermined. Proposed mechanisms include perinatal insults and disrupted organogenesis secondary to genetic mutations or environmental influences during gestation [5].

CASE PRESENTATION

We report the case of a 38-year-old married female with a history of autoimmune polyendocrinopathy, including Addison's disease and thyroiditis. The patient presented with growth retardation, amenorrhea, infertility, and recurrent episodes of hypoglycemia.

Laboratory evaluation revealed the following: low luteinizing hormone (LH, 0.12 IU/L), low follicle-stimulating hormone (FSH, 0.1 IU/L), hypocortisolism (1 µg/dL), hypoglycemia (0.73 g/L), hyperprolactinemia (44.73 ng/mL), and low thyroxine (T4, 5.2 µmol/L).

An ultrasound examination was performed, demonstrating thyroid hypotrophy with features consistent with thyroiditis.

The patient was referred by her endocrinologist for a pituitary MRI as part of the evaluation for hypogonadotropic hypogonadism.

Imaging revealed an interrupted pituitary stalk (**figure 1**), ectopic posterior pituitary, and a slightly hypoplastic anterior pituitary (**figure 2**).

The diagnosis of pituitary stalk interruption syndrome was subsequently confirmed.

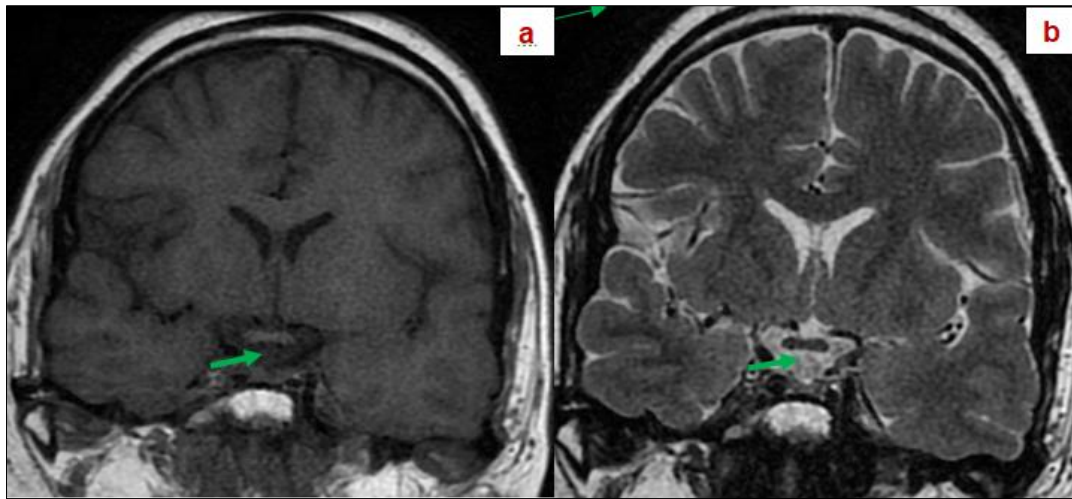


Figure 1: Coronal T1 (a) and T2 (b) weighted MRI sequences demonstrating an interrupted pituitary stalk (arrow).

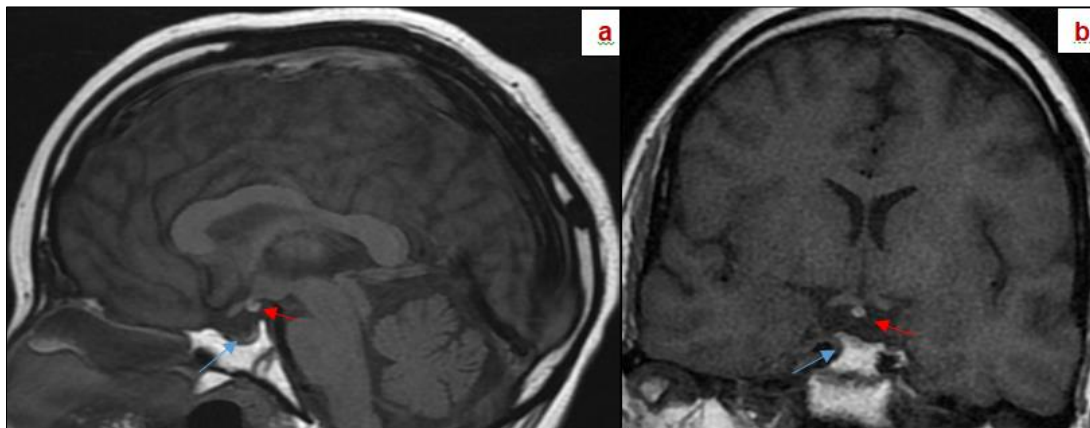


Figure 2: Sagittal (a) and coronal (b) T1-weighted MRI sequences showing a hypoplastic anterior pituitary (blue arrow) and an ectopic posterior pituitary (red arrow).

DISCUSSION

❖ *Epidemiology and Clinical Manifestations:*

PSIS is a rare congenital anomaly that may present with diverse clinical symptoms due to reduced secretion of anterior pituitary hormones.

A male predominance has been observed in PSIS, with a male-to-female ratio reported between 2.3 and 6.9:1, consistent with a potential X-linked inheritance pattern. The mean age at diagnosis is approximately 9.4 ± 11.6 years [6].

Approximately 20–50% of PSIS cases are associated with additional congenital anomalies, predominantly midline defects, including cleft lip, diaphragmatic agenesis, optic nerve hypoplasia, encephalocele, or harelip [5].

This syndrome is most frequently characterized by anterior pituitary hormone deficiencies, typically occurring in the following order of prevalence: growth hormone (100%), gonadotropins (97.2%), corticotropin (88.2%), and thyrotropin (70.3%). Mild

hyperprolactinemia (6.9%) may occasionally be observed, secondary to impaired dopaminergic inhibition. In contrast, posterior pituitary function usually remains intact [7].

❖ *MRI Features:*

During the first two years of life, the pituitary gland undergoes significant morphological changes. At birth, both the anterior and posterior lobes display high signal intensity on T1-weighted MRI images [8, 9]. By approximately six months of age, the anterior lobe loses this hyperintensity and becomes isointense, whereas the posterior lobe maintains a persistent high signal throughout life, reflecting the presence of neurosecretory granules [10, 11].

The MRI appearance of PSIS is variable. The most characteristic findings include a hypoplastic anterior pituitary, ectopic posterior pituitary, and an absent or interrupted pituitary stalk [12]. In certain patients, the anomaly may be limited to either an ectopic posterior pituitary or an isolated stalk interruption [13–15].

The location of the ectopic posterior pituitary has functional prognostic significance, as a higher number of anterior pituitary hormone deficiencies are observed when the posterior lobe is located at the median eminence or within the hypothalamic region [16].

❖ Treatment:

Prompt and appropriate hormone replacement therapy constitutes the cornerstone of management for all patients with PSIS. Depending on the specific hormonal deficiencies identified and the patient's age, treatment may include a combination of growth hormone, levothyroxine, cortisol, testosterone or estrogen, and desmopressin [17].

Hormone replacement therapy should be tailored to the specific hormonal deficiencies present, including cortisol, thyroxine, growth hormone, and, in cases of osteoporosis, calcium supplementation [18].

Early diagnosis and treatment are crucial because, first, untreated PSIS is associated with significant morbidity and mortality, and second, inadequate growth before the onset of puberty may result in reduced final adult height.

CONCLUSION

This case emphasizes the importance of performing pituitary imaging in patients with complex endocrine disorders such as autoimmune polyendocrinopathy. Identifying pituitary stalk interruption syndrome allows timely initiation of appropriate hormonal therapy and close follow-up.

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