Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Pituitary Stalk Interruption Syndrome in a Young Male with Congenital Hypopituitarism: MRI Findings and Clinical Correlation

M. Mekouar[1*], O. Fahir[1], Y. Bouktib[1], A. Elhajjami B. Boutakiout[1], M. Ouali Idrissi[1], N. Cherif Idrissi[1]

[1]Department of Radiology, Arrazi Hospital, Mohammed VI University Hospital Center, Marrakech, Morocco

DOI: https://doi.org/10.36347/sjmcr.2025.v13i05.121 | **Received:** 14.04.2025 | **Accepted:** 26.05.2025 | **Published:** 29.05.2025

***Corresponding author:** M. Mekouar
Department of Radiology, Arrazi Hospital, Mohammed VI University Hospital Center, Marrakech, Morocco

Abstract | **Case Report**

Pituitary Stalk Interruption Syndrome (PSIS) is a rare congenital disorder characterized by a triad of anatomical anomalies affecting the pituitary gland. We present the case of a 21-year-old male with congenital hypopituitarism. MRI findings were critical in confirming PSIS and guiding further management. This case highlights the essential role of imaging in evaluating patients with endocrine dysfunction and growth failure.

Keywords: Pituitary Stalk Interruption Syndrome (PSIS), Congenital Hypopituitarism, Magnetic Resonance Imaging (MRI), Ectopic Posterior Pituitary, Hypoplastic Anterior Pituitary.

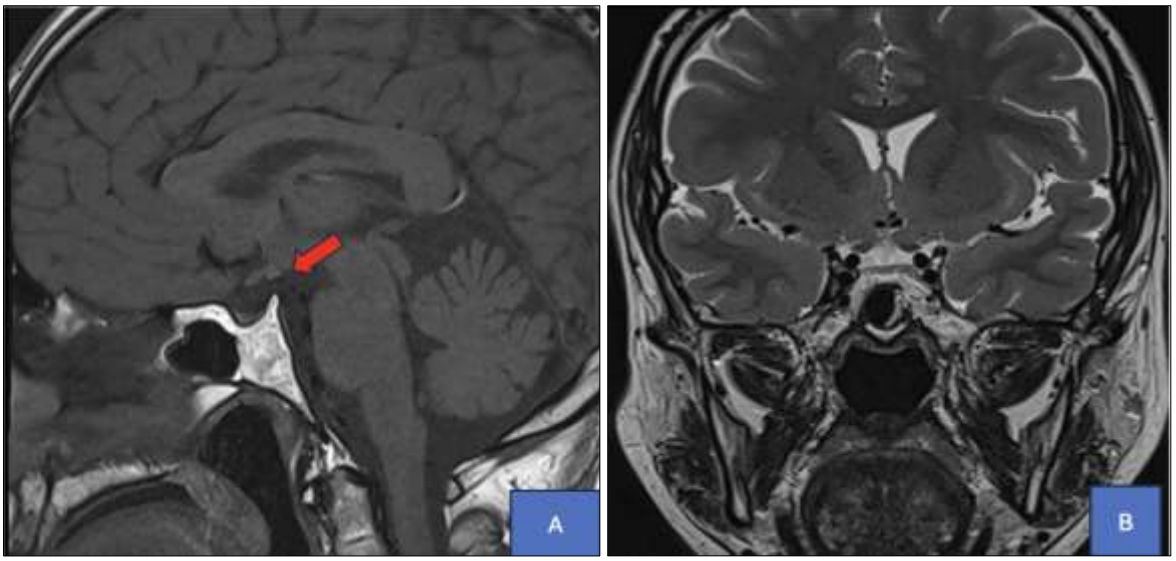
INTRODUCTION

Pituitary Stalk Interruption Syndrome (PSIS) is a rare congenital condition that presents with growth hormone deficiency and other anterior pituitary hormone deficits. First described in the late 20th century, the syndrome is defined by a triad on magnetic resonance imaging (MRI): absent or thin pituitary stalk, ectopic or absent posterior pituitary, and hypoplastic or aplastic anterior pituitary gland [1, 2]. MRI is the gold standard imaging modality due to its excellent resolution of soft tissue structures and ability to detect these hallmark abnormalities [3]. Early diagnosis enables timely hormonal replacement therapy and prevents complications related to delayed treatment.

CASE PRESENTATION

A 21-year-old male presented to the endocrinology clinic with a history of congenital hypopituitarism. He had been diagnosed at the age of four following an evaluation for growth retardation and failure to thrive. There was no significant personal or family history of endocrine or genetic disorders. Initial laboratory investigations showed hypoglycemia, low serum cortisol and thyroxine, and reduced ACTH levels, consistent with adrenal insufficiency. He was also diagnosed with central hypothyroidism and hypogonadotropic hypogonadism.

Magnetic resonance imaging of the brain and pituitary was performed using a dedicated pituitary protocol. The imaging revealed an ectopic posterior pituitary, identified as a T1-weighted hyperintense bright spot located at the median eminence. The pituitary stalk was not visualized, indicating complete absence. The anterior pituitary was hypoplastic, appearing significantly reduced in volume but still located within the sella turcica. These findings were consistent with a diagnosis of Pituitary Stalk Interruption Syndrome [4, 5].



(A) T1 sagittal image showing an hypoplastic anterior pituitary gland and a 5 mm hyperintense focus in the retrochiasmatic region, which represents the ectopic posterior pituitary gland (red arrow). (B) T1 coronal image showing non-visualisation of the pituitary stalk

DISCUSSION

PSIS is typically diagnosed during childhood or adolescence due to symptoms such as growth retardation and delayed puberty. It has a strong male predominance and is often diagnosed in patients presenting with multiple anterior pituitary hormone deficiencies. While the majority of cases are sporadic, genetic mutations involving transcription factors critical to pituitary development have been implicated in some familial and syndromic cases [1-6]. Etiological theories include both genetic mutations such as HESX1, LHX4, and PROP1, and perinatal insults such as birth trauma, breech delivery, or neonatal hypoxia [6-8].

MRI plays a central role in confirming the diagnosis of PSIS. It allows accurate characterization of the hypothalamic-pituitary axis. In typical presentations, the anterior pituitary is hypoplastic or aplastic, the pituitary stalk is absent or thin, and the posterior pituitary is ectopically located, most often along the pituitary stalk or at the median eminence [3, 4]. In addition to the classic triad, associated midline cerebral abnormalities such as septo-optic dysplasia, agenesis of the corpus callosum, hydrocephalus, and Chiari malformations may be observed [7]. In our case, none of these associated anomalies were present.

Clinically, growth hormone deficiency is the most consistent finding and may be accompanied by deficiencies in ACTH, TSH, LH, and FSH. Prolactin levels may be low or elevated depending on the degree of dopaminergic disconnection [5]. Early diagnosis and hormone replacement therapy are critical in avoiding metabolic crises and optimizing growth and development.

CONCLUSION

This case illustrates the characteristic MRI features of Pituitary Stalk Interruption Syndrome in a patient with long-standing hypopituitarism. MRI is essential for diagnosing PSIS and for guiding the appropriate endocrine management. Awareness of the typical imaging features and clinical context is vital for radiologists and endocrinologists alike, especially when evaluating patients with unexplained short stature and multiple hormonal deficiencies.

REFERENCES

1. Wang CZ, Guo LL, Han BY, et al. Pituitary stalk interruption syndrome in 55 children: clinical, hormonal and magnetic resonance imaging characterization. *Pediatr Radiol.* 2014;44(9):1133-1140. doi:10.1007/s00247-014-2977-2
2. Sani I, Kreitschmann-Andermahr I, Hoff C, et al. Pituitary stalk interruption syndrome: etiology and clinical findings. *J Pediatr Endocrinol Metab.* 2007;20(3):335-344.
3. Gupta DK, Bhatia V, Sarma D, et al. Pituitary stalk interruption syndrome: an MRI-based study in children. *Neuroradiology.* 2012;54(4):321-328.
4. Maghnie M, Ghirardello S, Genovese E, et al. Hypothalamic-pituitary MRI in children with growth hormone deficiency: correlation with clinical and laboratory findings. *J Clin Endocrinol Metab.* 2000;85(3):1384-1390.
5. Arrigo T, Wasniewska M, De Luca F. Pituitary stalk interruption syndrome: a congenital anomaly leading to hypopituitarism. *Horm Res Paediatr.* 2011;75(3):141–147.
6. Reynaud R, Gueydan M, Saveanu A, et al. Genetic analysis of hypopituitarism using next-generation sequencing: prevalence of mutations and polymorphisms in 122 patients with

hypopituitarism. *Clin Endocrinol (Oxf).* 2016;84(3):365–371.
7. Kelberman D, Rizzoti K, Avilion A, et al. Genetic regulation of pituitary gland development in human and mouse. *Endocr Rev.* 2009;30(7):790–829.
8. Liu S, Tang L, Meng F, et al. Clinical and genetic features of pituitary stalk interruption syndrome in 90 Chinese patients. *Clin Endocrinol (Oxf).* 2020;93(2):121–128.