MRI findings in a pituitary stalk interruption syndrome

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Abstract: **Pituitary Stalk Interruption Syndrome (PSIS) is a rare congenital disorder characterized by an absent or hypoplastic pituitary stalk, often leading to multiple endocrinological abnormalities. It typically presents in early childhood with growth hormone deficiency, hypothyroidism, hyperprolactinemia, and adrenal insufficiency, resulting in short stature, delayed puberty, hypoglycemia, and developmental delays. Birth complications such as breech presentation and neonatal jaundice are common. MRI plays a crucial role in diagnosis, revealing structural abnormalities, including an absent or ectopic pituitary gland, hypothalamic displacement, and third ventricle enlargement. Early diagnosis and hormonal therapy are essential for managing symptoms and preventing long-term complications.**

Key Words: **Pituitary stalk interruption syndrome (PSIS), panhypopituitarism, Ectopic Posterior Pituitary, hypoplastic pituitary stalk**

INTRODUCTION

Pituitary Stalk Interruption Syndrome (PSIS) is a rare congenital disorder characterized by anomalies in the pituitary stalk and often associated with a variety of endocrinological abnormalities. This syndrome is typically diagnosed through imaging, particularly MRI, which allows for detailed visualization of the pituitary and hypothalamic structures.

In cases of pituitary stalk interruption syndrome (PSIS), MRI typically reveals an absent or hypoplastic pituitary stalk, appearing as a thin, interrupted, or completely missing connection between the hypothalamus and pituitary gland. The pituitary gland itself may be underdeveloped, flattened, or ectopically positioned, with a small or absent anterior lobe and a posterior lobe displaced caudally. The hypothalamus may be abnormally positioned, disrupting its normal relationship with the pituitary gland. Third ventricle abnormalities, such as enlargement, may also be present. In severe cases, the anterior pituitary may be completely absent, while the posterior pituitary may appear ectopic, often found in the infundibulum or near the third ventricle. Additional findings may include septo-optic dysplasia, hydrocephalus, ventriculomegaly, or, rarely, arachnoid cysts near the pituitary region.

LEARNING OBJECTIVES

- Understand Pituitary Stalk Interruption Syndrome (PSIS) as a rare congenital disorder affecting the pituitary gland and stalk.
- Recognize key clinical features, including endocrine deficiencies (growth hormone, thyroid, adrenal insufficiency), developmental delays, and neonatal complications.
- Identify typical MRI findings, such as an absent or hypoplastic pituitary stalk, ectopic or underdeveloped pituitary gland, and associated brain abnormalities.
- Highlight the importance of early diagnosis and hormonal therapy to manage symptoms and prevent complications.

CASE REPORT

A 17-day old male neonate presented with persistent hypoglycernia and neonatal jaundice.
The neonate was a full term with breech presentation and LSCS delivery, he cried after 1 cycle of bag and mask ventilation.
Initial hormonal panel was performed
He underwent MRI Brain Plain for further evaluation.

Imaging and laboratory findings:

- MRI of the brain showed an absent anterior pituitary gland and absent pituitary stalk with an ectopic posterior pituitary gland, consistent with Pituitary Stalk Interruption Syndrome (PSIS).
- Initial hormonal panel showed low levels of serum cortisol (0.82), TSH (0.51) and free T4 (0.7).

- After the complete work up the patient was started on hydrocortisone and resulted in reversal of the hypoglycemia and neonatal jaundice.

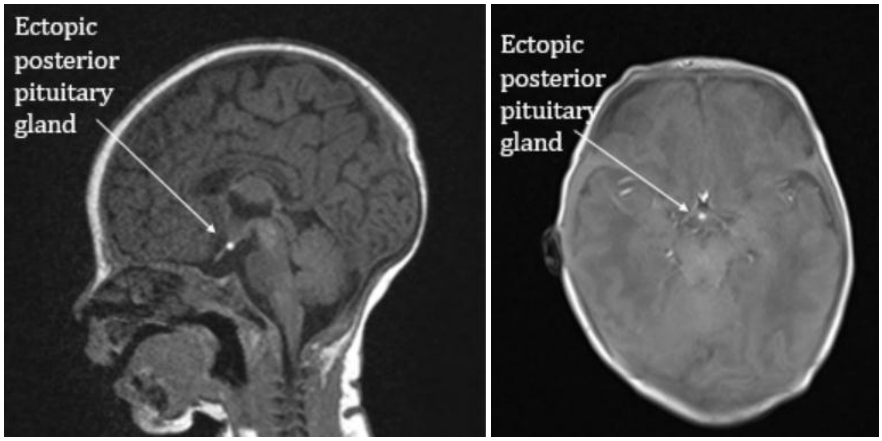

Fig: Sagittal T1 weighted image showing absent anterior pituitary gland and absent pituitary stalk with an ectopic posterior pituitary gland

DISCUSSION

PSIS typically manifests in early childhood with endocrine, developmental, and neurological symptoms. Endocrine issues include growth hormone deficiency causing short stature, multiple pituitary hormone deficiencies that may lead to panhypopituitarism, hypothyroidism, hyperprolactinemia, and adrenal insufficiency. Developmental challenges include delayed puberty, learning difficulties, seizures, and hypoglycemia. Birth complications such as breech presentation, neonatal hypoglycemia, and jaundice are common. MRI often reveals structural abnormalities like an absent or hypoplastic pituitary stalk, ectopic or underdeveloped pituitary gland, hypothalamic displacement, and third ventricle enlargement. Symptom severity varies based on hormonal and structural abnormalities.

MRI Findings:

In a case report of PSIS, the following MRI findings would commonly be observed:

1. Absence or Hypoplasia of the Pituitary Stalk:

The key MRI feature of PSIS is the absence or severe hypoplasia of the pituitary stalk. On imaging, it is seen as a thin, interrupted, or completely absent stalk, with no visible connection between the hypothalamus and the pituitary gland. In some cases, the stalk may appear discontinuous, with a gap or interruption in its continuity.

2. Abnormal Pituitary Gland:

The pituitary gland itself may be hypoplastic, meaning it is small or underdeveloped. In some cases, the gland may appear as a flattened structure or may be displaced. This could also be noted as a small anterior pituitary lobe and, in severe cases, the pituitary gland may be absent or displaced into an ectopic location. The posterior pituitary lobe might be located more caudally than usual, with an abnormal configuration.

3. Displacement of the Hypothalamus:

There may be upward displacement or abnormal positioning of the hypothalamus. The normal relationship between the hypothalamus and the pituitary gland is disturbed due to the absence of the pituitary stalk. This displacement can be identified through MRI slices showing the proximity and relationship between these structures.

4. Third Ventricle Abnormalities:

There could be associated changes in the third ventricle, such as dilatation or an enlarged third ventricle, which can be attributed to the lack of normal pituitary function and the displacement of brain structures.

5. Agenesis of the Anterior Pituitary:

In some cases, there may be complete agenesis of the anterior pituitary, which can be evident as an absence of the anterior pituitary lobe on sagittal MRI images. The posterior pituitary might still be visible, and its position may be altered or irregular.

6. Ectopic Posterior Pituitary:

The posterior pituitary gland, typically located at the base of the pituitary stalk, may be displaced, sometimes found in the infundibulum or another ectopic position within the brain. On MRI, the posterior pituitary may be visible but located in an unusual position, such as in the floor of the third ventricle.

7. Associated Abnormalities:

In some cases, other structural abnormalities may be observed, such as septo-optic dysplasia (SOD), where the optic nerves and the septum pellucidum may be underdeveloped. Hydrocephalus and ventriculomegaly may also be present, though these are less common.

8. Arachnoid Cyst (Rare):

Arachnoid cysts can occasionally be seen near the pituitary stalk area or within the suprasellar cisterns. This is a rare finding but may be noted in some patients with PSIS.

CONCLUSION

MRI findings play a crucial role in diagnosing Pituitary Stalk Interruption Syndrome (PSIS), revealing an absent or hypoplastic pituitary stalk, a small or displaced pituitary gland, an enlarged third ventricle, and possible ectopic positioning of the posterior pituitary. Structural abnormalities such as septo-optic dysplasia or ventriculomegaly may also be present. Clinically, PSIS manifests with endocrine dysfunction, including growth hormone deficiency, hypothyroidism, hyperprolactinemia, and adrenal insufficiency, leading to short stature, delayed puberty, fatigue, and hypoglycemia. Developmental and neurological symptoms such as learning difficulties, seizures, and neonatal complications like breech presentation and jaundice further contribute to the condition's complexity.

These findings are critical for guiding treatment and management, as the condition typically results in hormonal deficiencies and necessitates close endocrinological monitoring.

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REFERENCES

[1] Gutch, Manish; et al. Pituitary stalk interruption syndrome Case report of three cases with review of literature. Journal of Pediatric Neurosciences 9(23p 188-191, May-Aug 2014
[2] Wang CZ, Guo LL, Han BY, et al. Pituitary Stalk Interruption Syndrome: From Clinical Findings to Pathogenesis. J Neuroendocrinol 2017:29. doi: 10.1111/jne 12451