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Case Report

Pituitary Stalk Interruption Syndrome in a Child: A Rare Case Report with Literature Review

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Abstract

Introduction

Pituitary stalk interruption syndrome is an exceedingly rare congenital abnormality affecting the pituitary gland that is still not fully understood. This study presents a 7-year-old child with the disease.

Case presentation

A 7-year-old male child was presented with short stature, school performance postponement, and an intellectual disability at a mild level. At 6.5 years of age, he had no facial features, was wearing eyeglasses, and had a weight and height of 20 kg (25th percentile) and 101 cm (3rd percentile), respectively. At the age of 7 years, his height was 117 cm (10th percentile) and his weight was 25 kg (50th percentile). Most laboratory tests were normal. However, insulin-like growth factor-1 and glucagon levels were low. A dynamic magnetic resonance imaging of the pituitary gland revealed an ectopic posterior pituitary lobe and the absence of a pituitary stalk. The patient was put on hormone replacement therapy (Norditropin pen) to control the growth hormone deficiency, and he was put under a close follow-up to monitor growth and panhypopituitarism.

Conclusion

Although hormone replacement therapy is associated with satisfactory outcomes in treating the syndrome, lifelong follow-up is indicated as new hormone deficiencies may arise later in life.

1. Introduction

Pituitary stalk interruption syndrome (PSIS) or pituitary stalk transection syndrome is an anomalous congenital defect in the pituitary gland. The syndrome is associated with hormone deficiencies related to the pituitary gland [1,2]. It can be described by a thin or interrupted pituitary stalk, a hypoplastic or absent anterior pituitary, and an ectopic or absent posterior pituitary gland [3]. As a result, individuals with PSIS often experience a permanent growth hormone deficit, leading to a variety of symptoms and complications, with the main symptoms appearing in the first decade of life. The clinical findings can vary depending on the extent of hormonal deficiencies [2]. The estimated incidence of PSIS is

approximately 0.5 per 1,000,000 births, with males being more frequently affected than females at a ratio of 1.7-2.3:1 [1,3,4]. PSIS has a significant impact on patient health and quality of life. The underlying causes and mechanisms of this condition remain poorly understood, yet, it is thought to be a result of a complex interplay of genetic mutations and environmental factors [5,6].

This case report aims to present the clinical presentation, diagnosis, and management of a 7-year-old patient with PSIS.

2. Case Presentation

2.1. Patient information

A 7-year-old male child presented to the pediatric clinic with short stature, school performance postponement, two attacks of seizure, and an intellectual disability at a mild level. He was born at full term, without complications, to non-consanguineous parents who were healthy and lacked any medical history. His family history was unremarkable.

2.2. Clinical findings

On initial examination, at 6.5 years of age, he had no facial features, was wearing eyeglasses, and had a weight and height of 20 kg (25th percentile) and 101 cm (3rd percentile), respectively (BMI = 19.6 kg/m2). At the age of 7 years, his height was 117 cm (10th percentile) and his weight was 25 kg (50th percentile) (18.2 kg/m2).

2.3. Diagnostic assessment

Different related blood tests were done for the patient; complete blood count (CBC) and thyroid-stimulating hormone (TSH) were normal, however, insulin-like growth factor-1 (IGF-1) and glucagon levels were low. A dynamic magnetic resonance imaging (MRI) of the pituitary gland was conducted to examine the presence of any structural abnormalities, which showed that the posterior pituitary lobe was ectopic and that the pituitary stalk was absent, suggesting PSIS.

2.4. Therapeutic intervention

The patient was put on hormone replacement therapy (Norditropin pen) to control the growth hormone deficiency.

2.5. Follow-up

he was put under a close follow-up to monitor growth and panhypopituitarism.

4. Discussion

Pituitary stalk interruption syndrome is a congenital defect that was identified and described by Fujisawa et al. [1,3]. The etiology of the syndrome is still unclear, however, it has been considered that environmental and genetic factors are behind it. The suggested mechanism for pituitary stalk interruption syndrome can involve genetic mutations in various genes, such as LHX3/LHX4, PIT1, PROP1, TGIF, PROKR2, OTX2, GPR161, HESX1, and ROBO1, which are linked to the development of the anterior pituitary gland [3,6]. Additionally, some studies have suggested that Breech delivery, cesarean section, and neonatal hypoxemia may result in damage to the pituitary gland [5,7]. The parents of the existing case reported no complications during breech delivery and neonatal stage, and unfortunately, no genetic testing was performed for the case.

Individuals with PSIS may have additional congenital malformations, primarily affecting structures or organs located along the midline of the body. These may include cleft lip, absence of the diaphragm, hypoplasia of the optic nerve, bulging brain, and harelip. This suggests that the genetic defect causing PSIS may be associated with the genes involved in the embryonic development of the hypothalamus-pituitary area [1,2]. These features were absent in our case as he was brought to the pediatric clinic with only delayed performance and intellectual disability.

The primary symptoms of PSIS typically manifest during the first ten years of a person's life. The most common clinical examination findings in patients with PSIS can vary depending on the extent of hormonal deficiencies. Common manifestations may include prolonged neonatal jaundice, growth failure, delayed puberty, and features of hypogonadism such as the absence of secondary sexual characteristics, micropenis or small testes in males, and lack of breast development in females [2,7]. Hypothyroidism may present with fatigue, cold intolerance, and other signs of thyroid hormone deficiency. In addition, patients may show signs of other hormonal deficiencies such as adrenal insufficiency, growth hormone deficiency, and diabetes insipidus, depending on the involvement of other pituitary hormones [4].

The diagnosis of PSIS is typically confirmed through a combination of clinical, radiological, and endocrine evaluations. Brain MRI is the imaging modality of choice for evaluating the pituitary gland and stalk as it can reveal the absence or thinning of the pituitary stalk, absent or hypoplastic anterior pituitary, and ectopic posterior pituitary [8]. Endocrine evaluation should include measurement of basal hormone levels, dynamic testing, and stimulation tests to assess the functional status of various hormonal axes. Hormonal tests may check for deficiencies in adrenocorticotropic hormone, growth hormone, thyroid-stimulating hormone, antidiuretic hormone, luteinizing hormone, and follicle-stimulating hormone [9]. In the current case, all pituitary hormone panels were in the normal range except IGF1 which was in the lower range.

The management of PSIS involves replacement therapy for the identified hormonal deficiencies. It is typically tailored to the specific hormonal deficiencies and may include levothyroxine for hypothyroidism, glucocorticoids for adrenal insufficiency, sex hormone replacement for hypogonadism, growth hormone replacement for growth hormone deficiency, and desmopressin for diabetes insipidus. Regular monitoring of hormonal levels and clinical response is important to optimize the management and adjust hormone replacement therapy accordingly [4]. In this case, depending on his deficiency of IGF1 the patient was put on a Norditropin pen to control growth hormone levels.

The long-term prognosis of PSIS depends on the severity of hormonal deficiencies, the timeliness of diagnosis, and the initiation of appropriate hormone replacement therapy. With early diagnosis and proper management, most patients with PSIS can achieve adequate hormonal replacement, achieve normal growth, and a good quality of life. However, lifelong hormone replacement therapy may be required, and close monitoring is necessary to adjust the hormone replacement regimen as needed [10].

5. Conclusion

PSIS is a rare condition characterized by developmental abnormalities of the pituitary gland and stalk, resulting in various hormonal deficiencies. Clinical examination, radiological imaging, and endocrine evaluation are the keys to diagnosing the condition. Hormone replacement therapy is the mainstay of management and regular monitoring is essential for optimizing outcomes.

Declarations

Conflicts of interest: The author(s) have no conflicts of interest to disclose.

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