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A Case study on Pituitary Hypoplasia in a 5 Year Female Child

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Abstract: ***Background: Pituitary hypoplasia is a rare congenital anomaly characterised by underdevelopment of the pituitary gland, resulting in partial or complete hormone deficiencies. It may present as isolated growth hormone deficiency or as multiple pituitary hormone deficiencies. Early diagnosis through careful clinical evaluation, hormonal assessment, and magnetic resonance imaging of the hypothalamic–pituitary axis is essential for timely management. Hormone replacement therapy and multidisciplinary care significantly improve growth, metabolic function, and developmental outcomes. Objective: This review summarises the clinical profile, diagnostic modalities and treatment outcomes of pituitary hypoplasia in paediatric population. Methods: Clinical evaluation, hormonal investigations, bone age assessment, and magnetic resonance imaging were performed to establish the diagnosis. We report a case of isolated growth hormone deficiency secondary to pituitary hypoplasia in a five-year-old female presenting with short stature. Results: Isolated growth hormone deficiency. Bone age was delayed compared to chronological age. Magnetic resonance imaging of the brain demonstrated a hypo plastic anterior pituitary gland, confirming the structural basis of the deficiency. Child was initiated on appropriate hormone replacement therapy with planned regular endocrine follow-up. Conclusion: Early recognition of pituitary hypoplasia in children with short stature is essential. Timely diagnosis and hormone therapy can significantly improve growth outcomes.***

Keywords: Pituitary hypoplasia, Hypopituitarism, Growth hormone deficiency, Paediatric endocrinology

1. Introduction

The pituitary gland is the central regulator of the endocrine system and plays a pivotal role in growth, metabolism, stress response, and reproductive function. Hormones secreted by the anterior and posterior lobes influence peripheral endocrine organs and physiological homeostasis. Structural abnormalities of the pituitary gland can therefore lead to significant systemic dysfunction.

Pituitary hypoplasia is a rare congenital disorder characterized by underdevelopment of the anterior pituitary gland, often leading to varying degrees of hormone deficiency. This condition may present as isolated growth hormone deficiency or as part of a broader spectrum of pituitary hormone deficiencies, depending on the extent of tissue involvement. The estimated incidence of congenital pituitary hypoplasia is approximately 1 in 3,000 to 1 in 10,000 live births, with improved magnetic resonance imaging playing a key role in increased detection of structural anomalies associated with growth hormone deficiency.

Several case reports and observational studies have documented pituitary hypoplasia associated with isolated growth hormone deficiency or combined pituitary hormone deficiencies. For example, a case report described an infant with isolated growth hormone deficiency due to a novel mutation in the growth hormone-1 gene, with magnetic resonance imaging revealing anterior pituitary hypoplasia. Another series of children with growth hormone deficiency showed that a subset had pituitary hypoplasia on imaging, illustrating the structural basis for endocrine dysfunction.

The exact pathogenesis of pituitary hypoplasia is heterogeneous and may involve genetic mutations affecting pituitary development, disruptions in early embryogenesis, or perinatal insults. Abnormal development of Rathke's pouch, genetic defects in transcription factors, or pituitary stalk interruption may result in deficient hormone production. Structural imaging typically demonstrates a small anterior pituitary, thin or absent pituitary stalk, and sometimes an ectopic posterior bright spot on magnetic resonance imaging.

Clinically, affected children frequently present with short stature, reduced growth velocity, and delayed bone maturation, as in the present case. Early diagnosis is essential because untreated hormone deficiencies can lead to severe growth failure, metabolic imbalance, and life-threatening conditions such as adrenal insufficiency. This article presents a case of isolated growth hormone deficiency secondary to pituitary hypoplasia in a five-year-old female, highlighting the importance of early recognition and comprehensive management.

2. Case Presentation

A five-year-old female child presented with fever, cough, and cold for one week. The respiratory symptoms were acute in onset and associated with decreased appetite. There was no history of headache, vomiting, seizures, visual disturbances,

polyuria, polydipsia, or altered sensorium. On further evaluation, parents reported poor height gain since early childhood. She was born at term by normal vaginal delivery to non-consanguineous parents with a birth weight of 2.5 kilograms and cried immediately after birth. There was no history of neonatal hypoglycaemia, prolonged jaundice, central nervous system infection, head trauma, radiation exposure, or prolonged steroid use. Developmental milestones were appropriate for age, and immunisation was complete as per the national immunisation schedule. Family history revealed short stature in the eldest sibling, while the other sibling had normal height.

On examination, the child was alert and active with tachypnea. Respiratory system examination revealed bilateral crepitations, while cardiovascular, neurological, and abdominal examinations were normal. Anthropometric assessment showed height and weight below the third percentile for age with proportionate short stature and growth velocity less than four centimetres per year. There were no dysmorphic features or midline defects. Bone age assessment revealed significant delay compared to chronological age. Laboratory evaluation demonstrated mild microcytic hypo chromic anaemia and vitamin D deficiency. Hormonal analysis showed severely reduced growth hormone levels with markedly decreased insulin-like growth factor one levels, while thyroid function was within normal limits. Chest radiograph revealed bilateral bronchopneumonia. Magnetic resonance imaging of the brain demonstrated a small anterior pituitary gland with a thin stalk and ectopic posterior bright spot, consistent with pituitary hypoplasia. Based on clinical, biochemical, and radiological findings, a diagnosis of isolated growth hormone deficiency secondary to pituitary hypoplasia was established.

Differential diagnoses

These included constitutional growth delay, familial short stature, chronic systemic illness, malnutrition, hypothyroidism, celiac disease, chronic pulmonary disease, multiple pituitary hormone deficiencies, and structural abnormalities such as pituitary stalk interruption syndrome. These were systematically evaluated and excluded based on clinical findings, laboratory investigations, and imaging studies.

Based on clinical, biochemical and radiological findings, a diagnosis of isolated growth hormone deficiency secondary to pituitary hypoplasia was established.

3. Discussion

Pituitary hypoplasia is a rare congenital structural abnormality of the pituitary gland that may result in isolated growth hormone deficiency or multiple hormone deficiencies. Advances in magnetic resonance imaging have improved early detection of structural abnormalities in children presenting with growth failure.

In the present case, proportionate short stature, reduced growth velocity, delayed bone age, and markedly low growth hormone and insulin-like growth factor one levels established the diagnosis of isolated growth hormone deficiency. Normal thyroid function suggested preserved other hormonal axes. Magnetic resonance imaging findings of a small anterior pituitary gland with a thin stalk and ectopic posterior bright spot confirmed pituitary hypoplasia as the structural cause.

Differential diagnoses such as hypothyroidism, celiac disease, chronic systemic illness, malnutrition, and familial short stature were excluded based on clinical evaluation and laboratory findings. The episode of bronchopneumonia was likely incidental but facilitated identification of the underlying endocrine disorder.

Management of pituitary hypoplasia depends on the specific hormone deficiency. Recombinant growth hormone therapy is the mainstay of treatment in isolated growth hormone deficiency and significantly improves growth outcomes when initiated early. Regular follow-up is essential to monitor growth response and detect evolving hormone deficiencies. In this case, acute bronchopneumonia was treated with intravenous antibiotics and supportive care, and long-term endocrine follow-up with planned growth hormone therapy was advised. Early diagnosis and appropriate management are crucial to ensure optimal growth and prevent long-term complications.

4. Conclusion

Pituitary hypoplasia is an important but uncommon cause of childhood growth failure. Careful growth monitoring, timely hormonal evaluation, and magnetic resonance imaging are essential for early diagnosis. Isolated growth hormone deficiency may present without other endocrine abnormalities, emphasising the need for high clinical suspicion in children with proportionate short stature and delayed bone age. Early initiation of appropriate hormone therapy significantly improves growth outcomes and long-term prognosis.

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