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PITUITARY STALK INTERRUPTION SYNDROME: A RARE CAUSE OF SHORT STATURE

Radiology

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ABSTRACT

Pituitary stalk interruption syndrome is a rare congenital disorder and an uncommon but treatable cause of short stature. It has variable clinical, biochemical and radiological presentations. Diagnosis is usually established on magnetic resonance imaging (MRI) after clinical suspicion and biochemical evidence of hypopituitarism. Growth hormone deficiency is seen in almost all the cases with a mixture of other anterior pituitary hormones deficiency. The prognosis is favourable when diagnosed early and treated appropriately. In our case, a 5.5-year-old child was brought to the paediatrics department with complains of failure to thrive and recurrent seizures. Clinical and biochemical examination revealed stunted growth with hypopituitarism and hypoglycemia. MRI showed PSIS. The child was started on hormonal therapy and is maintaining well.

KEYWORDS

Psis, Pituitary Stalk Interruption Syndrome, Hypopituitarism, Ectopic Posterior Pituitary

INTRODUCTION

Pituitary stalk interruption syndrome (PSIS) is a rare congenital disorder with the reported incidence of 0.5/1,00,000 live births [1]. This condition was first reported in 1987 following the widespread use of MRI imaging [2]. It is characterised by the triad of thin or absent pituitary infundibulum, aplastic or hypoplastic anterior pituitary gland, and ectopic posterior pituitary gland. The patient usually presents with variable symptoms mainly secondary to anterior pituitary hormonal deficiency. The diagnosis is confirmed by magnetic resonance imaging (MRI) findings. Apart from a spectrum of presentation, the age of onset of this disease is also highly variable ranging from birth to paediatric age. High degree of suspicion is needed for early diagnosis to prevent morbidity. We present a case of 5-year-old male child who got diagnosed with PSIS with magnetic resonance imaging and was started on hormonal therapy for management.

Case Report

A 5.5-year-old male of Indian origin visited paediatric OPD of our institute with complains of recurrent seizures and failure to thrive. The first symptoms appeared at 3 years of age for which treatment was sought at outside clinic with failure to follow up.

The child was born out of non-consanguineous marriage with normal vaginal delivery at full term without any complications. There was no history of delayed cry. The child has 1 elder sibling (2 years elder) who is healthy. Parents are of normal heights. There is no significant family history.

At 3 years of age, the height, weight, body mass index (BMI) and head circumference of the child were 82.7 cm (<3rd centile), 9.3 kg (<3rd centile), 13.6 (<3rd centile) and 45 cm (<3rd centile) respectively. On recent examination, developmental milestones (motor) are slightly delayed for age The patient has normal intelligence for age and performs average in school. The current height, weight, body mass index (BMI) and head circumference are 95.2 cm (<3rd centile), 11.8 kg (<3rd centile), 13.0 (<3rd centile) and 45.5 cm respectively. These parameters show poor physical growth.

Routine blood investigations were within normal range. Random blood sugar was 47gm/dL. Growth hormone stimulation test was done which showed growth hormone deficiency (<0.05 ng/mL). Investigation for other pituitary hormones was done which showed thyroid stimulating hormone (TSH): 0.32 mIU (0.45-4.5 mIU), fasting morning cortisol: 4.8 µg/dL (range 5-23 µg/dL), prolactin: 1.18 ng/mL (<20 ng/mL), testosterone: 7.4 ng/mL (range 7-20 ng/mL), adrenocorticotropic hormone (ACTH): 6.8 pg/mL (range 11-82 pg/mL), These biochemical tests pointed towards anterior pituitary hormone deficiency. There was no symptom of diabetes insipidus like polyuria, polydipsia or nocturia. An electroencephalogram (EEG) was performed which was normal.

Radiological investigations included X-RAY of the non-dominant left hand which showed bone age of 16 months at chronological age of 3 years (determined using Tanner-Whitehouse 2 method), which signifies significant disease [Figure 1]. Contrast enhanced magnetic resonance imaging (MRI) brain was advised further which showed hypoplastic anterior pituitary with height of 3.1 mm (normal height is 5.4 ± 1.5 mm)[3] with absent pituitary stalk and T1 hyperintense focus at floor of third ventricle near optic chiasm representing the ectopic posterior pituitary [Figures 2a, 2b].

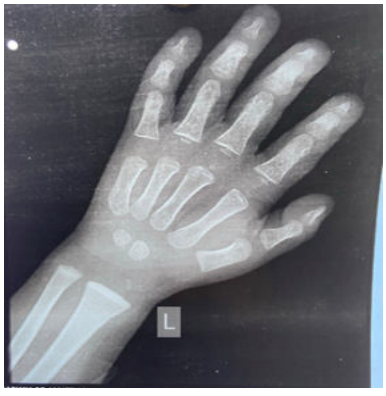

Figure 1: Radiograph of the left hand with wrist joint shows bone age of approximately 16 months (Tanner-Whitehouse 2 method) in a male child of 3 years

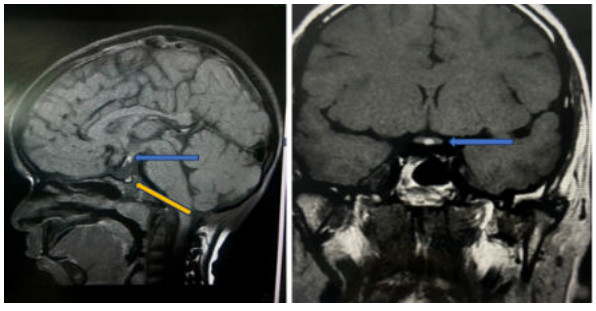

Figure 2a **Figure 2b**

Figures 2a and 2b: Mid-sagittal and coronal T1-weighted sections of the brain of the patient shows hypoplastic anterior pituitary (3.1mm) [yellow arrow], absent pituitary stalk and T1 hyperintense ectopic posterior pituitary at the floor of the 3rd ventricle near optic chiasm [blue arrows]

The patient was started on steroids, thyroid and growth hormone replacement therapy and showed increment in height of 12cm over 2 years' time with a growth velocity of 6cm/year. The child is kept under routine follow-up.

DISCUSSION

PSIS is a rare congenital cause of short stature and has variable presentation depending on age of presentation and hormone severity. It was first reported by Fujisawa et al.[2] in 1987. Less than 1000 cases were reported in the literature until 2010 [4]. There is slight male predominance as per literature. The patient in our case was also a male child.

The etiology of PSIS is not well known, however PSIS is believed to be either due to mutations in the genes involved in pituitary embryogenesis (PROP1, LHX3, HEXSX1, PROKR2 and GPR161) or perinatal asphyxia [5]. PSIS can present with anterior pituitary hormone deficiencies (growth hormone, 100%; gonadotropin, 97.2%; corticotropin, 88.2%; and thyrotropin, 70.3%); however, hyperprolactinaemia (6.9%) due to lack of dopaminergic inhibition can be seen. Posterior pituitary function is intact.

As the reported number of PSIS is limited, the clinical manifestations of this disease are diverse. The age of presentation also depends on the severity of the hormone deficiency. When PSIS occurs at birth, failure to thrive, and hypoglycemia are the most common symptoms. In childhood, stunted growth is typically the main complaint, while delayed puberty is usually the main concern when it manifests in adolescence and early adulthood [6]. Most patient lack sexual development. Our patient presented with hypoglycemic seizure, failure to thrive and mild developmental delay.

The age of presentation in our patient was 3 years, which is similar to mean age of 3.6 years reported by Gascoin-Lachambre et al.[7] and 4.0 years by Pinto et al.[8]. Others like Reynaud at al. (9.6 years) and Guo et al. (19.6 years) found different mean age groups of presentation.

Brain MRI shows a range of features that includes small sella turcica, anterior pituitary hypoplasia and absence of the pituitary stalk and the presence of high signal posterior pituitary at the ectopic location, mostly at the infundibulum or at the median eminence of the hypothalamus [9,10]. Contrast-enhanced MRI is more effective than non-contrast MRI to look for the enhancement of the pituitary stalk and to look for the integrity of the hypothalamohypophyseal pituitary vessels[11].

CONCLUSION

PSIS is a rare congenital abnormality and high degree of suspicion is required for the diagnosis. Early identification, strict growth monitoring and hormonal therapy at the soonest is critical in such conditions. Early diagnosis is of paramount importance for preventing adverse effects on long-term growth and development. These patients can reach their normal height if they present before epiphyseal closure. Close follow-up during pubertal period is necessary. MRI is confirmatory for the diagnosis of PSIS.

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