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Case Report

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PITUITARY STALK INTERRUPTION SYNDROME: CASE REPORT WITH ISOLATED GROWTH HORMONE DEFICIENCY

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ABSTRACT

Pituitary stalk interruption syndrome is characterized by a triad of thin or interrupted pituitary stalk, aplasia or hypoplasia of the anterior pituitary and absent or ectopic posterior pituitary seen on magnetic resonance imaging (MRI). We are presenting the case history and examination with MRI finding of a child who presented with sever short stature and diagnosed as isolated growth hormone deficiency and treated with growth hormone therapy. Diagnosing such a case may seem challenging however, Pituitary anomalies should be considered in the differential diagnosis of sever short stature in children.

KEYWORDS: Growth hormone treatment, pituitary stalk interruption syndrome.

INTRODUCTION

Pituitary stalk interruption syndrome (PSIS) is characterized by a triad of thin or interrupted pituitary stalk, aplasia or hypoplasia of the anterior pituitary and absent or ectopic posterior pituitary. it is a rare congenital pituitary defect manifesting with variable spectrum of pituitary hormone deficiency.[1-3] The estimated incidence of PSIS around 0.5 in 100 000 births.[4] The diagnosing such a case may seem challenging however.

Pituitary anomalies should be considered in the differential diagnosis of sever short stature in children.

CASE REPORT

A 4 years old male was presented to pediatric endocrine outpatient clinic with history of short stature and small penis. He was a product of 34 weeks of pregnancy and delivered by normal spontaneous vaginal delivery birth weight 1.67 kg (10^{th} centile), birth length 41 cm (10^{th} centile) and head circumference 31cm (50^{th} centile). He was admitted to neonatal intensive unit for one month due to prematurity .there was no history of neonatal hypoglycemia or convulsion. No pervious admission or surgeries. No pulmonary, gastroentesinal, hepatic, cardiac or renal diseases. No history of malabsorption or malnutrition symptoms. No history of head trauma or meningitis. The patient was not on any medication. There was no consanguinity between parents. he has 3 healthy sibling 2 brothers and one sister. Psychosocial history was unremarkable. no history of similar condition in the family.

Vaccination was up to date. Developmental parameters were appropriate for age. He was on family diet with average appetite.

On examination he was not dysmorphic, weight, height, and head circumference were at 3^{rd}, 3^{rd} and 25^{th} percentile respectively. Midparental height 161 cm. upper/lower segment ratio was 1.3 (normal). There was frontal bossing. Examination of chest showed equal air entry with no added sound .cardiac examination was unremarkable. There was no hepatosplenomegaly, no skin changes. Child had normal muscle tone and power. Child can walk normally. Musculoskeletal examination was unremarkable. Tanner stage examination revealed that testis were prepubertal in size, stretch penile length: 2.3 cm (below -2SD) for age. Fundus examination within normal limit. No gluteal wasting. Growth velocity average was 1cm in 6 month follow-up Investigations showed Hemoglobin 12.3 g dl, white blood cell (WBC) 10.9 $x10^3/\mu L$, neutrophil 21 % and lymphocyte 57 %. Serum urea and electrolytes were normal. Liver function

tests were normal, renal profile and electrolytes were normal .urine and stool analysis were normal .Metabolic screen was unremarkable, blood gas was normal. Bone profile was normal Arterial blood gas was normal. Celiac disease profile was negative.

Hormonal analysis revealed IGF-1 was 36 (normal 25-157 ng/ml), IGHBP3 was 0.6 (normal 0.8-3 mg/L). Stimulated growth hormone level by (clonidine and glucagone) showed peak GH level was 1.8 ng/mL (normal >10 ng/ml. Thyroid function test within normal limits. Basal FSH, LH and Free Testosterone were normal prepubertal level. serum 25 OH vitamin D was 214 nmol/L (normal 50 - 250) Fasting morning cortisol 8 mic /dl (normal 3-21 mic /dl).

Bone age was equal to 2 years according to atlas of William walter Greulich. MRI pituitary showed ectopic posterior pituitary, pituitary stalk was not visualized in expected site and small anterior pituitary size which is consistence with diagnosis of Pituitary stalk interruption syndrome (Figure 1,2 and 3).

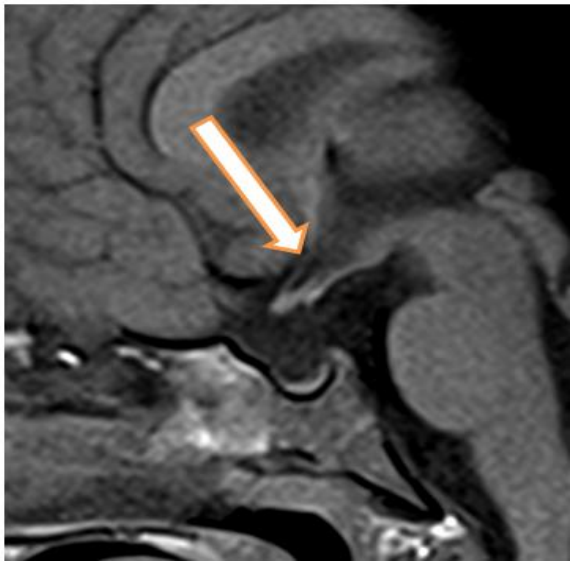

Figure 1: Sagittal pre-contrast T1-weighted image shows ectopic posterior pituitary (arrow).

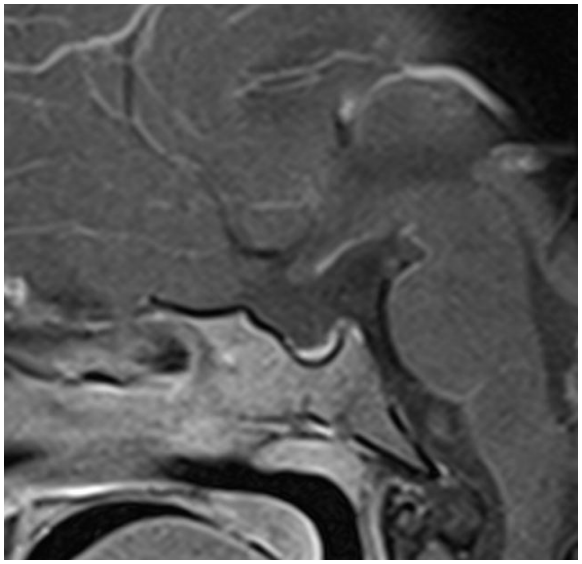

Figure 2 –a

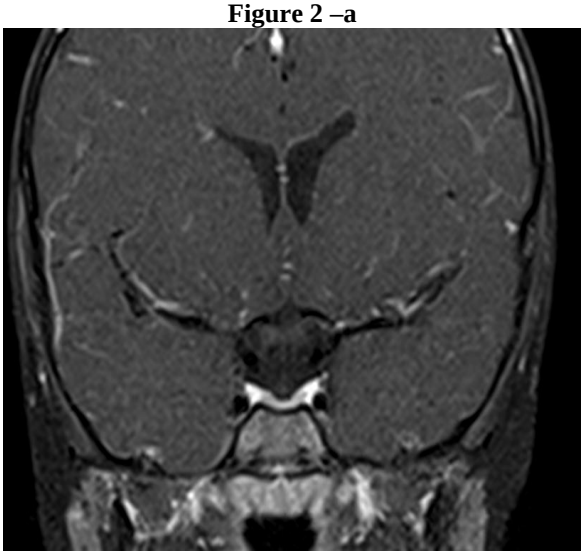

Figure 2-b

Figure 2-a and 2-b showed Postcontrast coronal and sagittal T1-weighted image – pituitary stalk not visualized in expected site.

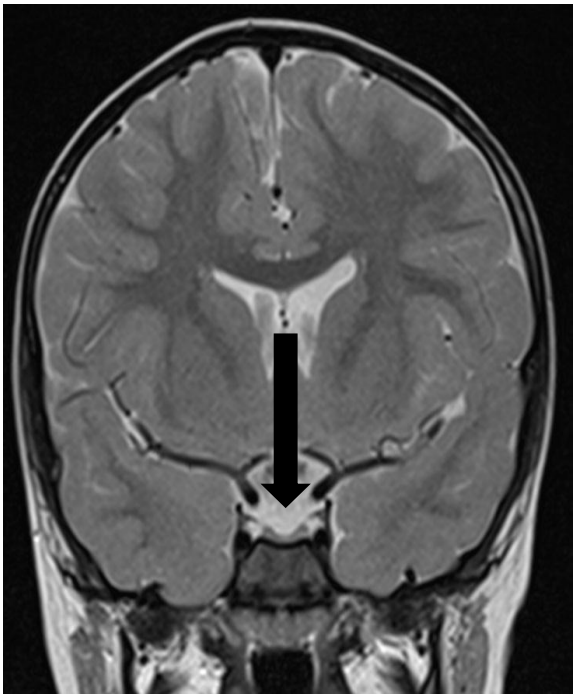

Figure 3: Coronal T2-weighted image showing small anterior pituitary measuring 1 mm in height (black arrow)

DISCUSSION

The first reported case of pituitary stalk interruption syndrome was in 1987 by Fujisawa *et al*. with the typical features: very thin or absent pituitary stalk, an ectopic posterior pituitary and hypoplasia or aplasia of the anterior pituitary gland.[5] The mean age of diagnosis of PSIS is 9.4 ± 11.6 years.[6] Most PSIS cases are sporadic however 5% are familial inherited condition.[7] The etiology of PSIS is still unknown.[8]

Many gentic mutation were associated with familial PSIS like HESX1, LH4, OTX3 and SOX3 can be the cause of PSIS.[6]

The most common manifestation of PSIS in children is growth retardation. in more than half of the patients, height and weight are below the third percentile with delayed bone age.[9-10] Most of reported PSIS cases presented with multiple anterior pituitary deficiencies.[11] Growth hormone deficiency is most frequently present most of reported cases.[12]

Anterior pituitary hypoplasia and absence of pituitary stalk are found in most of the cases (98.3%) and ectopic neurohypophysis in (91.4%) Of patients. In patient with ectopic neurohypophysis, 60.4% were located at the infundibular recess and 18.9% at the hypothalamus.[13]

In our patient who is a male male child present to endocrine OPD with complaints of short stature and micropenis. There is history of preterm vaginal delivery at hospital. There is no significant past and family history. On examination height and weight on the 3rd percentile of age. patient had micropenis. investigation showed that patient had growth hormone deficiency with delayed bone age. MRI brain suggestive of Pituitary stalk interruption syndrome On the basis of these findings, he was diagnosed as PSIS and patients were started on growth hormone treatment with no reported side effects.

Diagnosing such a case may seem challenging however, Pituitary anomalies should be considered in the differential diagnosis of sever short stature in children.

Conflict of interest: none.

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