Case Report

A rare case of short stature – pituitary stalk transection syndrome

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Abstract

Short stature due to Growth hormone deficiency could be due to multiple etiologies. One such rare cause is the Pituitary stalk transection syndrome which is characterized by a triad of thin or absent pituitary stalk, aplasia or hypoplasia of the anterior pituitary and absent or ectopic posterior pituitary seen on magnetic resonance imaging (MRI). The early identification of growth hormone deficiency through growth hormone stimulation tests, evaluation of the hypothalamic-pituitary anatomy by performing MRI brain and the early initiation of growth hormone replacement therapy may salvage the child from pathological short stature.

Keyword: Pituitary stalk, Hypoplasia or aplasia, Ectopic posterior pituitary

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Introduction

Short stature though not a life-threatening condition has significant associated comorbidities and psychosocial impact. Though there are multiple etiologies to this presentation few causes if detected early can be treated and hence totally prevent or partially correct the condition with growth [1]. Identifying these conditions assumes significance as, if left untreated these will result in a permanent short stature which can never be corrected once the child is past his growth spurt [2]. Thus, timely and correct identification of etiology is of prime importance in this condition.

One of the causes of short stature which if identified early can avoid short stature with appropriate treatment is growth hormone deficiency due to pituitary stalk transection. This condition was first described in 1987 after the widespread use of MRI imaging [3]. European data suggests that the incidence of pituitary stalk transection is nearly 0.5/10,00,000 live births [4]. With nearly 50,000 children born in India every day the disease burden is supposed to be high [5], however there are very few reported cases [6,7].

Manuscript received: 2nd February 2019
Reviewed: 11th February 2019
Author Corrected: 16th February 2019
Accepted for Publication: 21st February 2019

The possible explanation being low level of awareness of the condition that it is untreatable leading to delay in seeking medical attention. Even in those children presenting for evaluation, under-evaluation leads to missing this condition.

Pituitary stalk transection can be broadly divided into absence of pituitary stalk, hypoplasia or aplasia of the anterior pituitary and ectopic posterior pituitary gland [3]. Various hypothesis was put forward to explain the findings of absent or hypoplastic pituitary stalk.

One being a traumatic transection during delivery with attempted reformation of the stalk in later years and other being defective development of both adeno- and neurohypophysis with failure of fusion of the pituitary lobes and incomplete pituitary-hypothalamic axis [8,9].

The risk factors of developing this condition is associated with maternal factors like breech presentation, complicated labor and perinatal factors like trauma during birth, prolonged labor or forceps delivery [10]. These children present with short stature, decreased growth rate, seizures, hypotension, intellectual delay and delayed puberty [11].

MRI features of pituitary stalk transection with or without ectopic posterior pituitary was reported by many authors [12,13]. The principal treatment for this condition is early identification with imaging and growth hormone therapy [14,15]. We report a case of an Indian child with short stature who presented to us in early stage of his growth spurt, we discuss the diagnosis of his condition, treatment with growth hormone supplements and follow up along with review of literature.

Case Report

Clinical Data: A 5 year 6-month-old male child born out of non-consanguineous marriage, first in birth order brought by mother with chief complaint of not gaining weight and height since 1 year of age and complaint of short stature when compared to peers. No h/o malnutrition or any chronic illness. No significant treatment history. Child is term born, cephalic presentation through caesarian section with a birth weight of 2.75 kg and had uneventful perinatal history and normal developmental history. Child has a younger brother with normal growth and development. No history of short stature in first/second degree relatives.

Anthropometry

- Weight – 10 kg ; SD: -4.5
- Height – 110 cm ; SD: -5.9
- BMI – 14.52 (25 – 50th centile)
- US : LS – 1.18 (Proportionate)
- HC – 48 cm (normal)
- Stretched penile length – 3 cm (expected – 4 cm)
- Mid-parental height – 165cm (25-50th centile)
- Chronological age - 5 year 6 months
- Height age – 1year 6months
- Bone age -2years
- Growth velocity-no previous height records.

On general physical examination child had frontal bossing and micropenis. His systemic examination was normal. Child was diagnosed to have Pathological short stature due to endocrine cause probably Growth hormone deficiency for which he was investigated. Routine haematological workup came out to be normal.

Investigations

- CBP, CUE, Stool exam, LFT, RFT with serum electrolytes - normal
- Thyroid Profile - Normal [FT4 - 1.29 ng/dl ,TSH - 3.38 uiu/ml]
- Serum Cortisol - Normal [15.46 ug/dl]
- USG Abdomen - Normal

Growth hormone test: Clonidine stimulation test was done which showed Complete growth hormone deficiency. First sample for GH was taken at 0 minute and then 100 microgram Clonidine tablets was given orally. Second and third GH samples were taken after 1 hour and 2 hours respectively. Child was monitored for hypotension. Results were 0.178, 0.269, 0.175 ng/ml at 0-minute, one hour and 2 hours respectively which implied Complete growth hormone deficiency. (Normal > 10 ng/ml)

MRI Brain: s/o Pituitary Stalk Transection

- Hypoplastic anterior pituitary gland
- Ectopic posterior pituitary at median eminence
- Thin infundibulum

Management: The child was started on Human recombinant growth hormone @ 0.1 IU/kg/day which showed increment in height of 2.8cms over 3 months with a growth velocity of 10 cms/year. We planned to continue growth hormone replacement therapy for a duration of 2 years with close follow-up of growth and any progression of multiple pituitary hormones deficiency.

Discussion

Growth hormone deficiency is a common endocrinologic cause of shortstature. This hormone deficiency maybe idiopathic or associated with organic causes, such as tumors or surgery. Idiopathic growth hormonedeficiency occurs sporadically and maybe isolated or associated with multiplepituitary hormone deficiencies. Clinical isolated growth hormone-deficiency may progress to multiple pituitary hormone deficiency. Isolated growth hormone deficiencyand multiple pituitary hormone deficiency can be due to the pituitary stalktransection syndrome.

In a study by Van der Linden et al. among 21 patients with isolatedgrowth hormone deficiency evaluatedwith MR imaging, 19 had a thin or truncated pituitary stalk [16].

In another study by Triulzi et al., among 101 patients with congenital idiopathic growth hormone deficiency, 59 had ectopic posterior pituitary of which 30 patients had isolated growth hormone deficiency and the rest had multiple pituityary hormone deficiency [17].

The above two studies were compiled cross-sectional studies with no further followup whereas we did a prospective case study with the child started on therapy and regular followup.

In our case, the male child presented with complaint of short stature. There is no history of perinatal insult or breech presentation. There is no significant past and family history. The child had a normal development in all domains.

On examination the child had height and weight < 3 percentile for his age and had frontal bossing and micropenis. On investigation, the child had complete growth hormone deficiency along with delayed bone age. MRI brain is suggestive of hypoplastic anterior pituitary gland, ectopic posterior pituitary at median eminence and a thin pituitary stalk.

On the basis of these findings, the diagnosis of Pituitary stalk transection syndrome was made, and patients were started on hormonal replacement therapy. Growth hormone (GH) replacement therapy forms the cornerstone of management of children with growth hormone deficiency (GHD).

It is a liquid preparation given through Automated pen device at a dose of 0.33 mg/kg/week or 0.14 IU/kg/day subcutaneously, once daily administration at bedtime.

Conclusion

A high degree of suspicion is required for preventable causes of growth failure like growth hormone deficiency due to pituitary stalk transection syndrome. Early identification, strict growth monitoring and therapy at the earliest is of paramount importance in such conditions.

They have an excellent opportunity to reach their normal height if they present before epiphyseal closure. Multiple pituitary hormone deficiency should be looked out for, in all these cases. Close follow-up during pubertal period is necessary.

Funding: Nil, **Conflict of interest:** None initiated,
Perission from IRB: Yes

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How to cite this article?

Chadalawada L., Nimmagadda R., Vupputuri H., Bhimireddy V. A rare case of short stature – pituitary stalk transection syndrome. Int J Pediatr Res. 2019;6(02):87-90. doi:10.17511/ijpr.2019.i02.08

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