Case Report

Unravelling the cause of short stature by shades of grey

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Abstract

Background: Pituitary stalk transection syndrome (PSTS), also called pituitary stalk interruption syndrome (PSIS), is characterized by a triad of absent or hypoplastic anterior pituitary, hypoplastic, or thin infundibulum and ectopic posterior pituitary. There are various causes for short stature due to growth hormone deficiency and PSTS is one such rare cause. Patients with PSTS may present with additional major symptoms like neonatal hypoglycemia /prolonged physiological jaundice, cryptorchidism, micropenis, and in older children /in adults can manifest as short stature. We herein report a rare presentation of PSTS in a 6-year-old male child who presented with complaints of short stature and no weight gain. Physical examination revealed frontal bossing and micropenis. Lab investigations revealed normal thyroid profile and cortisol levels. However, clonidine stimulation test revealed GH deficiency. The patient was started on Human recombinant growth hormone which showed an increase in height on follow-up at 3 months and therapy was suggested to be continued for 2 years. Early detection and treatment are crucial for preventing permanent short stature and Magnetic resonance imaging (MRI) plays a vital part in its diagnosis.

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INTRODUCTION

PSTS was first reported by Fujisawa *et al.*[1] in 1987. It is a rare congenital abnormality with an estimated incidence of 0.5/1,000,000 births.[2] causing anterior pituitary deficiency. Hormone deficiencies can either be isolated (GH deficiency) or involving multiple anterior pituitary hormones. Posterior pituitary function is normal. Rare genetic mutations may be a cause and account for less than 5 % of cases. Depending on the age at presentation it can present as neonatal hypoglycemia/prolonged physiological jaundice, cryptorchidism, micropenis and in older children / in adults can manifest as short stature.[3,4] Early diagnosis is important as it can lead to permanent short stature and is associated with comorbidities.[5] We report a case of a child who presented to the pediatric department with short stature due to a rare cause and the role of imaging in arriving at a diagnosis.

CASE REPORT

A 6-year-old male child born out of non-consanguineous marriage presented to the pediatric department with complaints of short stature since 1 year of age and not gaining weight. Uneventful antenatal, natal and postnatal history noted. No short stature recorded in his siblings. No other significant similar history noted in his near relatives. On examination weight and height was less than 3 standard deviations below the mean for sex and age. Physical examination revealed frontal bossing and micropenis. Systemic examination was unremarkable. Laboratory investigations revealed a normal thyroid profile (FT4-1.29

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ng/dl, TSH -3.38 mIU/ nl) and cortisol levels (15.46 ug/dl). Serum creatinine -0.3 mg/dl, Serum ALT-23 U/L. USG abdomen –no significant abnormality detected. CBP, urine analysis was normal. Clonidine stimulation test for growth hormone at 0,1, and 2 hours was 0.178 ng /ml, 0.269 ng/ml (10.8 ng/ml being normal lower limit for age) revealed GH deficiency. MRI Brain revealed (shown in Fig. 1 (a-d)) shows hypoplastic anterior pituitary gland, hypoplastic infundibulum and ectopic position of the posterior pituitary bright spot at the median eminence.No corpus callosal agenesis / Arnold Chiari I malformation seen. There is no associated malformations like septo optic dysplasia (optic nerve hypoplasia, absent septum pellucidum) (Shown in Fig.2 (a,b)). Imaging findings were characteristic of pituitary stalk transection syndrome. Based on the clinical examination, laboratory results, and imaging findings, the patient was started on Human recombinant growth hormone which showed increase in height on follow up at 3 months, and therapy was suggested to be continued for 2 years.

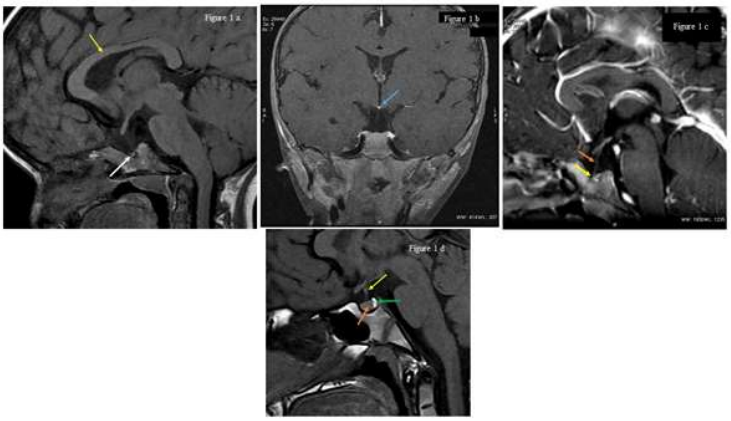

Figure 1: a-Sagittal T1W MRI reveals hypoplastic anterior pituitary gland (white arrow). Normal corpus callosum noted (yellow arrow). Fig 1. b-T1W coronal MRI shows ectopic location of posterior pituitary bright spot at the median eminence (blue arrow). Fig 1. c –post contrast T1W sagittal zoomed images- shows hypoplastic anterior pituitary gland (yellow arrow) and hypoplastic infundibulum (orange arrow). Fig 1. d- T1W sagittal images shows normal pituitary gland for comparison (orange arrow-anterior pituitary gland, green arrow-posterior pituitary bright spot at normal location and yellow arrow- pituitary stalk).

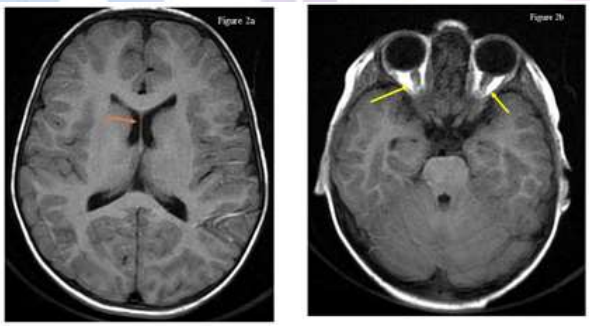

Figure 2: A and b-T1 W axial images of the same patient shows normal septum pellucidum (orange arrow) and normal appearance of optic nerves (yellow arrows).

DISCUSSION/CONCLUSION

PSTS patients manifest with symptoms in the first decade of life with male predominance of 2.3 and 6.9: 1.[6,7] Exact etiology is not known. It could be due to defective migration during intrauterine growth or could be secondary to ischemia with resultant reorganization of the infundibular axons with development of posterior pituitary in ectopic location [8]. Breech delivery may also lead to pituitary injury. Pickardt syndrome is hyperprolactinemia and hypothyroidism in combination [9]. PSTS can be associated with other malformations like (i) septo optic dysplasia comprising of optic nerve hypoplasia, absent septum pellucidum, hypothalamic pituitary dysfunction (ii) Agenesis of corpus callosum (iii) Chiari I malformation. Higher risk of congenital malformations is seen in association with isolated growth hormone deficiency.[7]

Clinical manifestations vary from hypoglycemia, prolonged physiological jaundice, cryptorchidism, micropenis in neonates to short stature and other signs of anterior pituitary hormone deficiency in older children and adults.[3,4] MRI features include absent or hypoplastic

anterior pituitary, hypoplastic or thin infundibulum and absent posterior pituitary in its normal location and situated at the median eminence or at the pituitary stalk. Anterior pituitary hormone deficiencies are related to the position of the ectopic posterior pituitary gland. Hormonal deficiency is more common when the posterior pituitary is situated at the median eminence or at the hypothalamic region.[6] Treatment consists of growth hormone replacement[10] which is the mainstay in the management of PSTS. PSTS should be considered as one of the causes for short stature. Clinical features, laboratory investigations, imaging features help to arrive at an early diagnosis of PSTS and initiation of appropriate hormone replacement (good response if treatment started before epiphyseal fusion), regular monitoring will thereby reduce morbidity and mortality.

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