Pituitary stalk transection syndrome in a 16 years old girl presenting with short stature and delayed puberty – A case report
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Section: Neuroradiology
Area of Interest: Neuroradiology brain
Procedure: Screening
Imaging Technique: MR
Special Focus: Endocrine disorders Case Type: Anatomy and Functional Imaging
Authors: Samar Hamid1, Shumaila Arooj2
Patient: 16 years, female

Clinical History:
A 16 year-old girl came to us for the evaluation of her short stature and delayed puberty. On examination she had underdeveloped secondary sexual characteristics. Her growth hormone, estradiol and vitamin D levels were subnormal.

Imaging Findings:
Contrast enhanced MRI of pituitary gland was performed which revealed an ectopic posterior pituitary gland appearing hyperintense on T1W1 MR imaging in the midline at the median eminence (Fig: 1). Additionally, the anterior pituitary appeared significantly smaller in size with a barely perceptible midline pituitary stalk which could hardly be seen on both pre and post contrast T1W1 MRI (Fig: 2, 3). Sella appeared smaller in size.

Discussion:
Background:
Pituitary stalk transection syndrome, also known as pituitary stalk interruption syndrome, is a rare entity with an estimated incidence rate of 5/1,000,000 births [1]. It is characterized by the presence of the classical triad of a thin or absent pituitary stalk, hypoplastic or aplastic anterior pituitary and ectopic posterior pituitary on MRI. This can be associated with midline defects and various pituitary endocrine deficiencies, ranging from isolated growth hormone deficiency to combined pituitary hormone deficiencies [2]. It may be congenital in origin or a sequelae of ischemic insult or head trauma. However, the exact mechanism is unknown. There is a male predominance with a male to female ratio of 6.9:1 and a mean age at diagnosis of around 9.4 ± 11.6 years. Usual presenting symptoms are short stature, delayed growth, seizures, hypotension, intellectual delay and delayed puberty [3].

Clinical Perspective:
On examination our patient had short stature, delayed puberty, no previous history of chronic medical or surgery issue or trauma. Her secondary sexual characters were underdeveloped. Her height was 138 cms and weight was 30 kgs. She was a student of class IX but was no taller in height than a student of class IV. Her hormone analysis
revealed decreased levels of growth hormone, estradiol and vitamin-D. This picture was suggestive of anterior pituitary dysfunction.

Imaging perspective:

Pituitary stalk transection syndrome should be suspected when there is lack of pituitary stalk visibility, hypoplasia of the anterior pituitary and ectopic posterior pituitary on Contrast enhanced MRI in appropriate clinical setting [4, 5].

Outcome:

Our patient was started on hormonal replacement therapy. It is important to closely follow up these patients in the long?term so that their natural history of progressive radiological and hormonal deterioration can be ascertained and managed properly.

Take home message:

Our case report highlights a rare, yet important cause of delayed puberty and short stature in children and young adults. MRI plays a key role in the diagnosis and management of this rare clinical entity. Early diagnosis and treatment of this rare pathological condition is twofold; prevention of permanent short stature and decreased morbidity and mortality. As complete anterior pituitary hormone deficiency may occur anytime during the 2nd or 3rd decade of life even after initiation of hormonal therapy, the role of follow up MRI should never be underestimated in such patients.

Differential Diagnosis List: Pituitary stalk transection syndrome, Posterior pituitary ectopia, Empty sella

Final Diagnosis: Pituitary stalk transection syndrome

References:


Miyamoto J et al (2001) Development of growth hormone and adrenocorticotropic hormone deficiencies in patients with prenatal or perinatal-onset hypothalamic hypopituitarism having invisible or thin pituitary stalk on magnetic resonance imaging. Endocr J 48: 355-362
Description: Barely perceptible midline pituitary stalk which could hardly be figured out on Coronal MR image
Origin: Department of Radiology, Jinnah Postgraduate Medical Center, Karachi, Pakistan
Figure 2

**Description:** Non enhancing imperceptible pituitary stalk with ectopic posterior pituitary

**Origin:** Department of Radiology, Jinnah Postgraduate Medical Center, Karachi, Pakistan.
**Figure 3**

**Description:** Small sized pituitary gland with absent posterior pituitary bright spot and ectopic posterior pituitary bright spot (arrow) **Origin:** Department of Radiology, Jinnah Postgraduate Medical center Karachi, Pakistan.