Case 15437

Eurorad

Atypical cause of short stature with growth hormone deficiency: Pituitary stalk interruption syndrome

Published on 25.02.2018

DOI: 10.1594/EURORAD/CASE.15437
ISSN: 1563-4086
Section: Neuroradiology
Area of Interest: Musculoskeletal bone Neuroradiology brain
Procedure: Diagnostic procedure
Imaging Technique: MR
Special Focus: Endocrine disorders Case Type: Clinical Cases
Authors: Naresh Bansal, Palak Patel, Pratik Panasara, Shikha Khandelwal, Dhara Patel, Mrugesh Doctor
Patient: 12 years, male

Clinical History:

Short stature, generalised weakness and easy fatigability.
Development milestones: normal.
X-ray of both hands with wrist for bone age: Bone age is less than chronological age of patient.
Hormonal values:
Growth Hormone: 0.127 ng/ml.
Free T4: 0.51 ng/dl.
S. TSH: 4.292 microIU/ml.
Insulin-like growth factor: <25 ng/ml.

Imaging Findings:

On X-ray of both hands ossification of four carpal bones is seen. Lower end of ulna is not ossified, suggesting bone age of less than 8 years. (Fig. 1)

Ectopic posterior pituitary (as evidenced by hyperintense focus on T1W image) located at median eminence on mid-sagittal and coronal T1WI respectively (Fig. 2 and 3).

Hypoplastic anterior pituitary gland in pituitary fossa and non-visualisation of pituitary stalk on mid-sagittal and coronal T2WI respectively (Fig. 4 and 5).

Visualised brain was normal.

Discussion:

There are many causes of short stature in childhood. Pituitary stalk interruption syndrome (PSIS) is also a cause for growth retardation with growth hormone or multiple anterior pituitary hormone deficiency.
It is a rare congenital anomaly of the pituitary gland which causes deficiency of anterior pituitary hormones. PSIS has a classical triad which consists of absent or ectopic posterior pituitary gland, interrupted pituitary stalk and anterior pituitary hypoplasia or aplasia. [1]

Pituitary stalk interruption syndrome was firstly reported by Fujisawa et al. [2]
Estimated incidence rate of PSIS is 0.5/1, 000, 000 births. [3]

The pathogenesis of PSIS is unclear. It could be acquired due to injury to this region during birth or mutation during the embryonic period. Few mutations in the genes are found to be associated in patients with congenital hypo-pituitarism and PSIS. e.g. HESX1, SOX3, LHX4, OTX2, PIT1, PROP1, PROKR2 and TGIF genes. [3, 4, 5, 6] In our patient birth history was normal.

The clinical features of PSIS in the early neonatal period include cryptorchidism, micropenis, hypoglycaemia and jaundice. The most common presenting clinical feature of PSIS in childhood is short stature. In most patients, usually weight and height are less than the third percentile as compared to normal children. Delayed bone age is also seen in patients with PSIS. [3, 6] In our patient the presentation was short stature.

X-ray of the hand can be used for the bone age estimation in childhood. In our patient, bone age was between 6-8 years, which is significantly lower than the chronological age.

On MRI imaging, T1-weighted, T2-weighted and gadolinium-enhanced MRI brain images in sagittal and coronal plane show hypoplastic adenohypophysis with ectopic posterior pituitary and absent stalk. [1, 3, 7, 8] In our patient all of the three findings were present.

It is important that the patients receive long-term follow-up for the earliest diagnosis and management of hormone deficiency as soon as symptoms manifest. [8]

Although this is a rare disorder, a clinician and radiologist should always think of pituitary stalk interruptionsyndrome in case of short stature with growth hormone deficiency alone or multiple hormone deficiencies, because PSIS patients have an excellent prognosis to achieve normal height according to their age if they are diagnosed before the fusion of epiphyses. [1, 9]

Differential Diagnosis List: Pituitary stalk interruption syndrome., Ectopic posterior pituitary gland, Empty sella

Final Diagnosis: Pituitary stalk interruption syndrome.

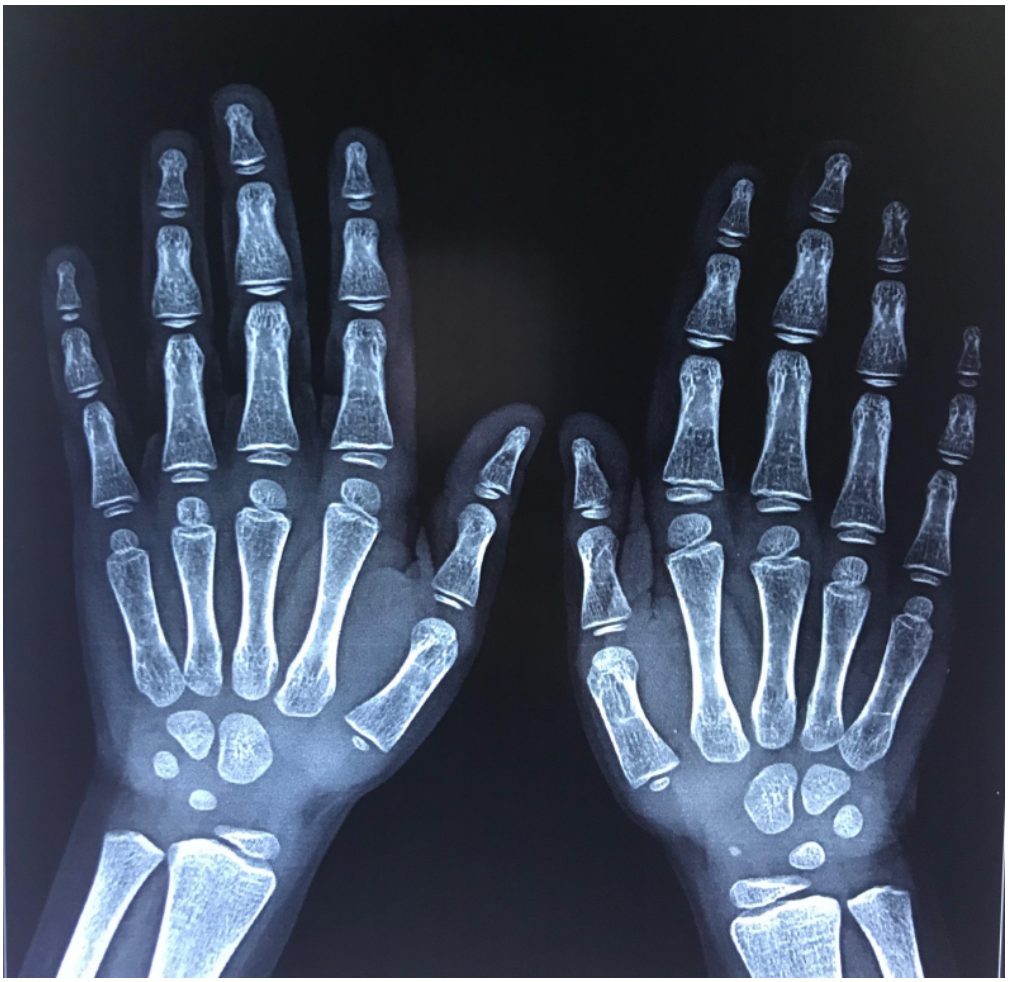
References:

Nanik Ram, Syed Ahsan Ali and Syed Zubair Hussain (2014) Pituitary stalk interruption syndrome presenting as short stature: a case report. Journal of Medical Case Reports 8:445. (PMID: 25524465)
Fujisawa I, Kikuchi K, Nishimura K, Togashi K, Itoh K, Noma S (1987) Transection of the pituitary stalk: Development of an ectopic posterior lobe assessed with MR imaging. Radiology 165:487–9 (PMID: 3659371)
Manish Gutch, Sukriti Kumar (2014) Pituitary stalk interruption syndrome: Case report of three cases with review of literature. Journal of Pediatric Neurosciences 9(2): 188–191 (PMID: 25250085)
Fabio Triulzi, Giuseppe Scotti, Berardo di Natale, Cristina Pellini, Monica Lukezic, Marialuisa Scognamiglio,

Giuseppe Chiumello (1994) Evidence of a Congenital Midline Brain Anomaly in Pituitary Dwarfs: A Magnetic Resonance Imaging Study in 101 Patients. Pediatrics 93:409–416 (PMID: 8115199)
Céline Bar, Charline Zadro (2015) Pituitary Stalk Interruption Syndrome from Infancy to Adulthood: Clinical, Hormonal, and Radiological Assessment According to the Initial Presentation. PLoS One 10(11) (PMID: 26562670)
Wang, C.-Z., Guo, L.-L., Han, B.-Y., Su, X., Guo, Q.-H. and Mu, Y.-M. (2017) Pituitary Stalk Interruption Syndrome: From Clinical Findings to Pathogenesis. Annals of Pediatric Endocrinology & Metabolism 22, 3, 208 (PMID: 27917547)
van der Linden AS, van Es HW (2017) pituitary stalk transection syndrome with ectopic posterior pituitary gland. Radiology 243(2):594-7 (PMID: 17456881)
Chinmay Kulkarni, Srikanth Moorthy, Sreekumar K Pullara, R Rajeshkannan, Ambika G Unnikrishnan (2012) Pituitary stalk transection syndrome: Comparison of clinico-radiological features in adults and children with review of literature. Indian J Radiol Imaging 22(3): 182–185 (PMID: 23599565)
Ioachimescu AG1, Hamrahian AH, Stevens M, Zimmerman RS (2012) Pituitary stalk interruption syndrome: Case report of three cases with review of literature. Pituitary 15(3):405 (PMID: 21826468)

Figure 1

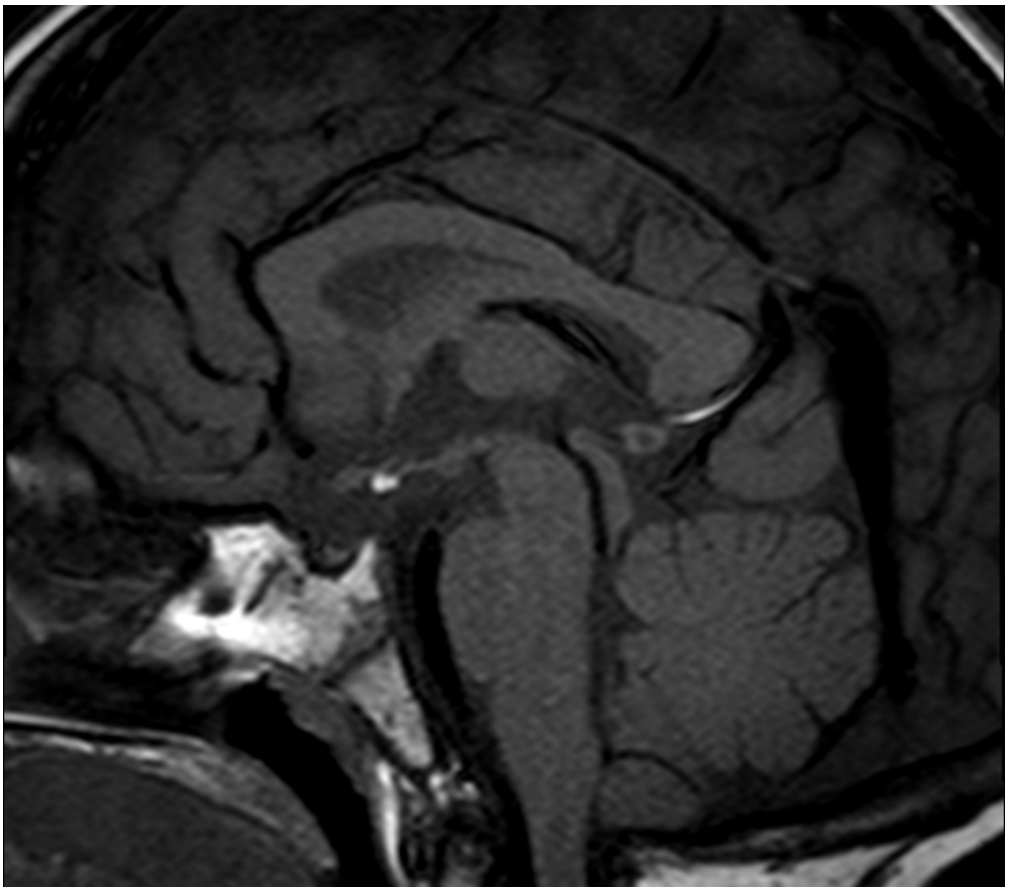
a



Description: Ossification of four carpal bones is seen. Lower end of ulna is not ossified, suggesting bone age of less than 8 years. **Origin:** GUJARAT IMAGING CENTRE, SAMVED HOSPITAL, AHMEDABAD, GUJARAT, INDIA

Figure 2

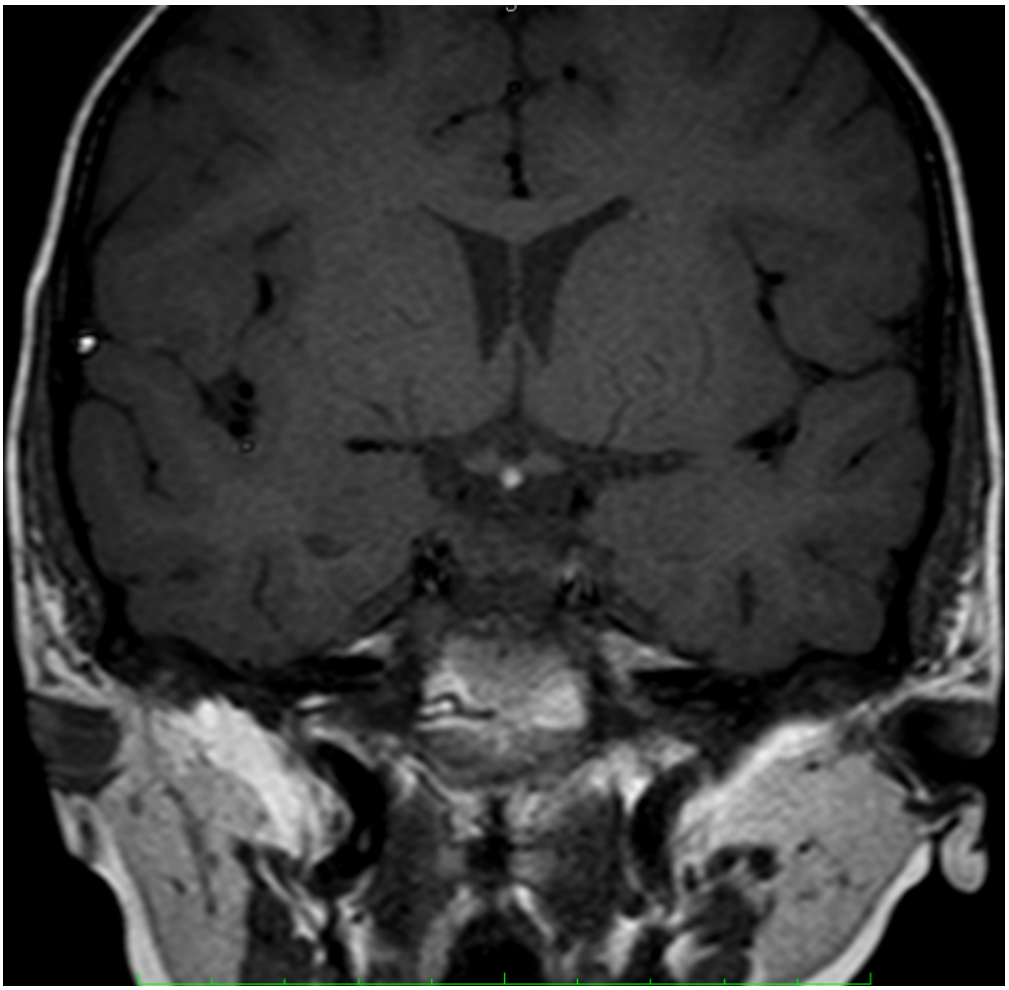
a



Description: T1W mid-sagittal image shows ectopic posterior pituitary (as evidenced by hyperintense focus) located at median eminence. **Origin:** GUJARAT IMAGING CENTRE, SAMVED HOSPITAL, AHMEDABAD, GUJARAT, INDIA

Figure 3

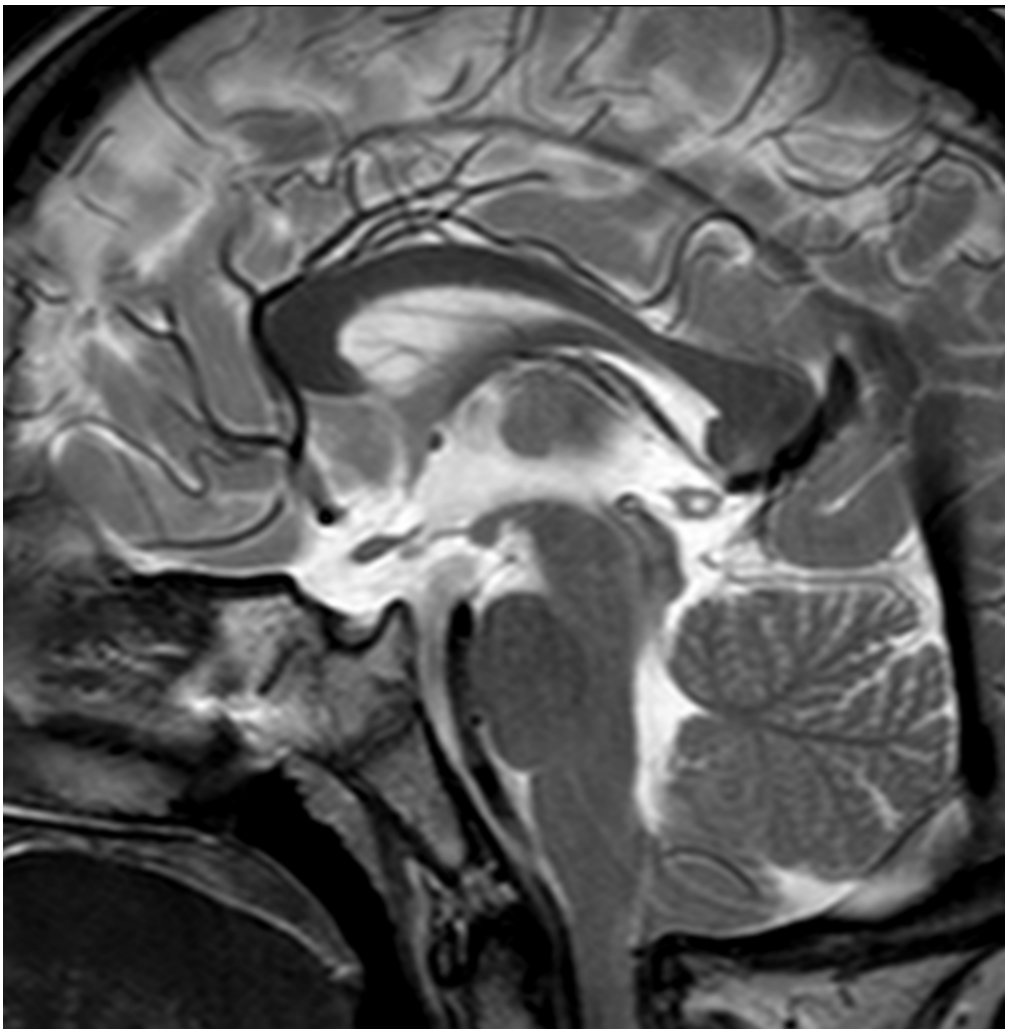
a



Description: T1W coronal image shows ectopic posterior pituitary (as evidenced by hyperintense focus) located at median eminence. **Origin:** GUJARAT IMAGING CENTRE, SAMVED HOSPITAL, AHMEDABAD, GUJARAT, INDIA

Figure 4

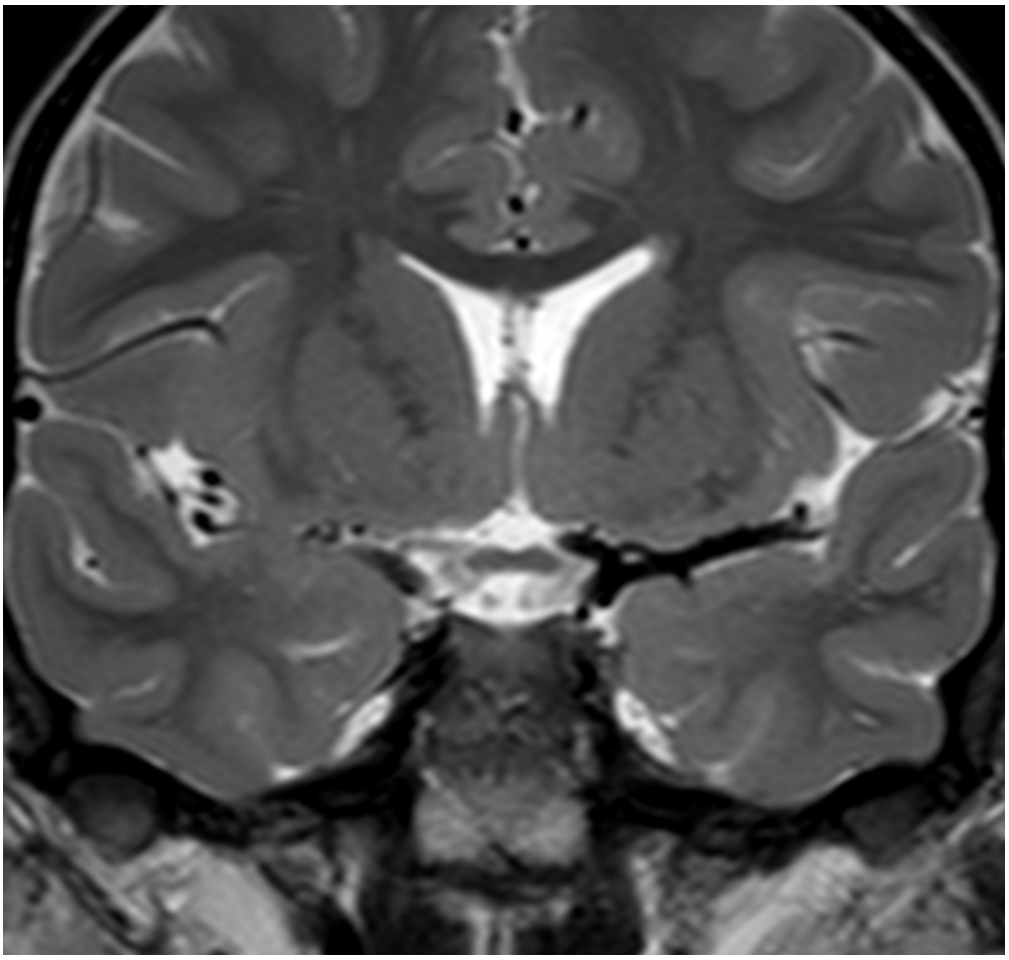
a



Description: T2W mid-sagittal image reveals hypoplastic anterior pituitary gland in pituitary fossa and non-visualisation of pituitary stalk. **Origin:** GUJARAT IMAGING CENTRE, SAMVED HOSPITAL, AHMEDABAD, GUJARAT, INDIA

Figure 5

a



Description: T2W coronal image reveals non-visualisation of pituitary stalk. **Origin:** GUJARAT IMAGING CENTRE, SAMVED HOSPITAL, AHMEDABAD, GUJARAT, INDIA