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# Imaging Findings of Pituitary Stalk Interruption Syndrome

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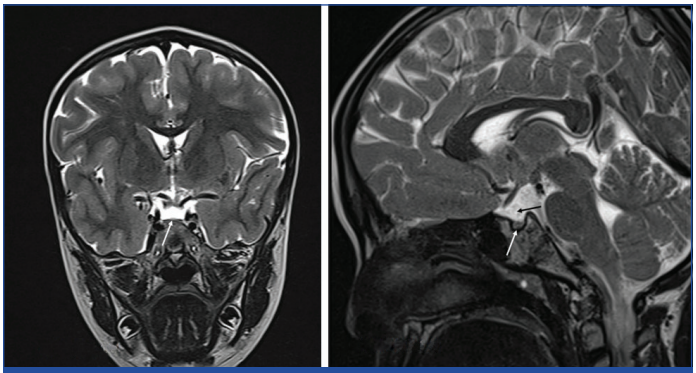
An 11-year-old male child presented with inadequate height and weight gain compared to peers. His social, familial, and peer interactions were normal, with no behavioural concerns. He was born at term via caesarean section (birth weight: 2.7 kg) to non consanguineous parents. His medical history included poor academic performance-resulting in a one-year school interruption-and developmental delay, including delayed tooth eruption. There was no history of chronic diarrhoea, vomiting, respiratory distress, recurrent hospitalisation, seizures, or bowel/bladder incontinence.

On examination, the child's height was 114 cm and weight 18 kg (both below the 3rd centile). He exhibited prepubertal genitalia, normal tone and power, and no focal neurological deficits. He was alert, active, and afebrile. Laboratory investigations showed normal peripheral smear, renal and liver function tests, serum electrolytes, and bone markers. However, reduced levels of insulin-like growth factor-1 (56 ng/mL; normal: 127-903 ng/mL), cortisol (2.60 µg/dL; normal: 5-25 µg/dL), and Thyroxine (T4) (3.48 µg/dL; normal: 5-12 µg/dL) were noted. Other anterior pituitary hormones were within normal limits. Random blood sugar and input/output charting were normal, ruling out polyuria.

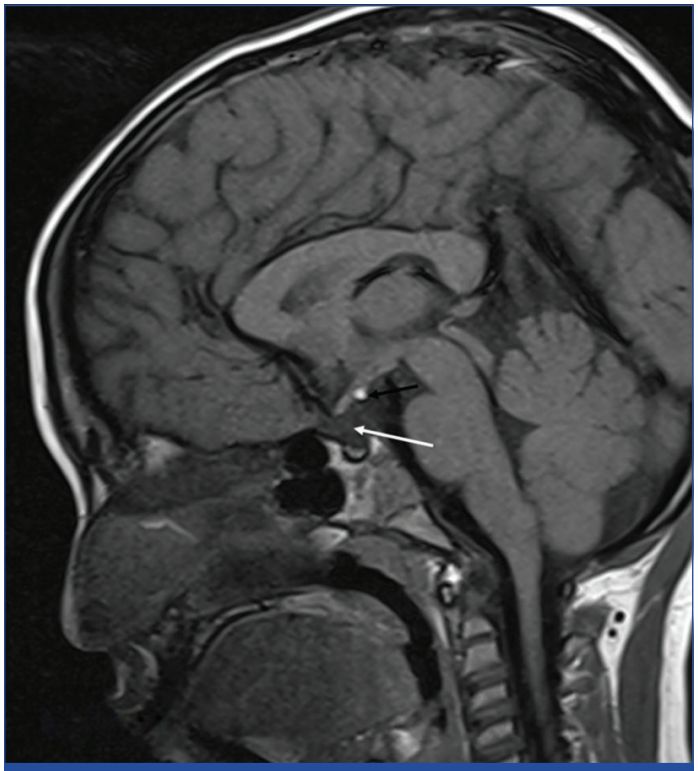
Magnetic Resonance Imaging (MRI) of brain revealed a hypoplastic anterior pituitary, absent pituitary stalk, and ectopic posterior pituitary, located near the hypothalamus, suggestive of Pituitary Stalk Interruption Syndrome (PSIS) [Table/Fig-1,2]. A hand X-ray showed four carpal bones indicating a bone age of approximately 4-6 years [Table/Fig-3].



**[Table/Fig-1]:** a) Coronal; and b) Sagittal T2 weighted MRI image showing hypoplastic anterior pituitary (white arrow) and absent pituitary stalk (black arrow). (Images from left to right)

Treatment was initiated with hydrocortisone (5 mg daily), levothyroxine (40 µg daily), and recombinant growth hormone. Regular follow-up with paediatric endocrinology was advised. PSIS is a rare congenital disorder with an incidence of 1 in 2,000,000 live births [1]. The typical MRI triad includes a hypoplastic anterior pituitary, an ectopic posterior pituitary, and an absent pituitary stalk, leading to anterior pituitary hormone deficiencies [2-4].

Though an increased incidence is associated with breech presentation, this case involved caesarean delivery [3]. The main pathology involves a hypoplastic or aplastic anterior pituitary and ectopic posterior



**[Table/Fig-2]:** Sagittal T1 weighted MRI image showing absent pituitary stalk (white arrow) and ectopic posterior pituitary bright spot appearing hyperintense on T1 weighted image (black arrow).


**[Table/Fig-3]:** Anteroposterior view of left hand showing four carpal bones suggestive of bone age of 4 to 6 years which is less than the chronological age of the patient.

pituitary with an absent pituitary stalk, impairing hypothalamic hormone transmission to the anterior pituitary, resulting in anterior pituitary hormone deficiencies [3]. Clinical diagnosis relies on isolated or combined anterior pituitary deficiencies, such as short stature, intellectual disability, and inadequate weight gain, as seen in this case. Most present in the first decade with delayed growth, potentially developing other anterior pituitary hormone deficiencies, seizures, and learning difficulties later in life [3,4]. Pickardt syndrome (hyperprolactinaemia and hypothyroidism) may also occur [4]. While clinical presentations vary, GH deficiency is almost universal [1,5]. Other deficiencies include ACTH, LH, FSH, and TSH, with reported frequencies of 95.8% (gonadotropin), 81.8% (corticotrophin), and 76.3% (thyrotropin) [6]. Hyperprolactinaemia was present in 36.4%, and 92.7% had two or more anterior pituitary hormone deficiencies [6].

Pituitary-related conditions may also involve extra-pituitary malformations, including midline central nervous system defects [7,8], such as septal agenesis, microcephaly, hydrocephalus, partial corpus callosum agenesis, Arnold-Chiari malformations, and optic nerve hypoplasia [7,8]. These abnormalities suggest underlying developmental or congenital issues affecting both the brain and pituitary gland, leading to neurological and hormonal disturbances [7,8]. The ectopic posterior pituitary location has prognostic significance, with higher anterior pituitary hormone deficiency prevalence when located at the median eminence or within the hypothalamus [7,9].

Early diagnosis and hormone replacement therapy are essential for optimising growth and developmental outcomes.

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