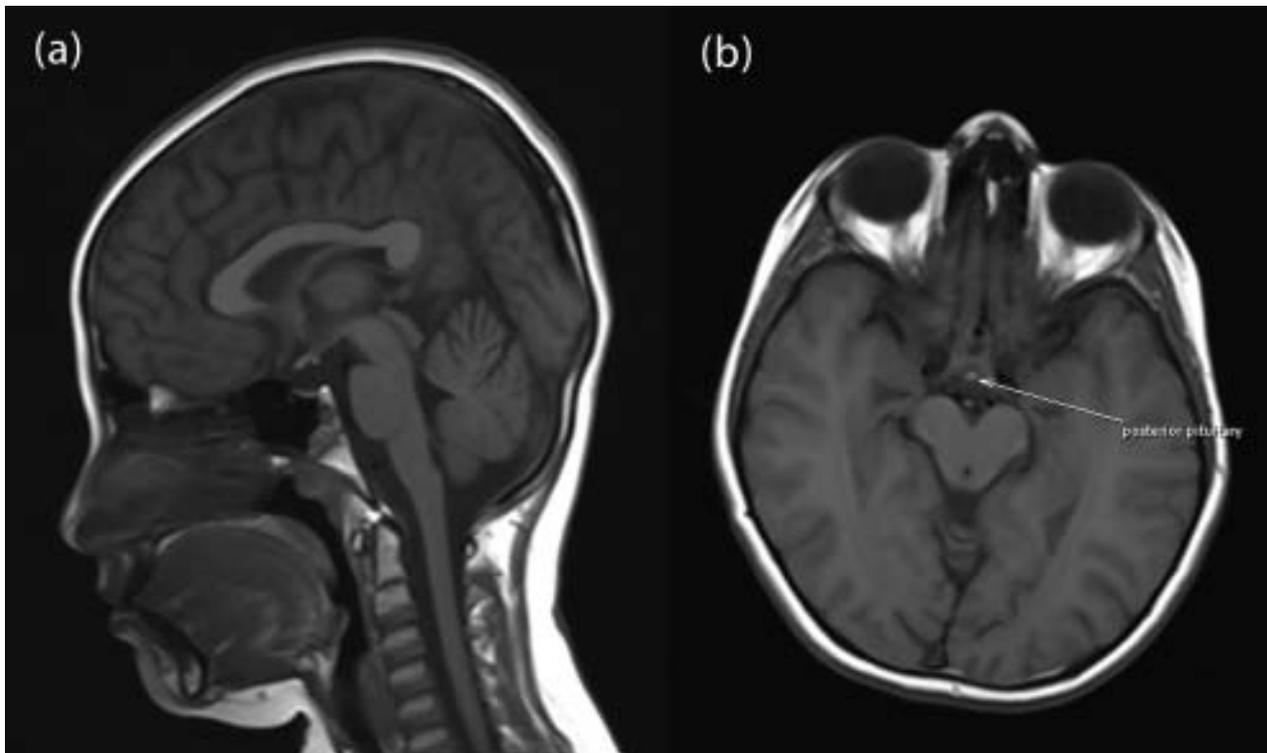


## Pituitary stalk interruption syndrome (PSIS) – a rare cause of short stature

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**Figure 1. MRI brain (a) sagittal and (b) axial images showing an absent anterior pituitary and ectopic posterior pituitary located in the region of the hypothalamus with absent pituitary stalk.**

A 16 year old girl was evaluated for short stature and delayed puberty. She was noted to be shorter than her twin sister since infancy. Her parents, twin and two other siblings had normal growth and development. Although her twin went through puberty at 12 years of age, she failed to develop any sign of puberty. There were no symptoms of hypothyroidism, adrenal insufficiency, polydipsia or polyuria.

The patient was a product of non-consanguineous marriage and the first born of twins, delivered with breech presentation. Although there was a history of early neonatal seizures with prolonged ICU stay and delayed developmental milestones compared to her twin, there had been no chronic medical problems later on and she

performed well at school. The social, family and peer interactions were normal with no behavioural problems. There was no history of short stature or primary amenorrhoea in the family.

On examination her height and weight were less than the 3rd percentile and she was pre pubertal (Tanner stage 1). White blood count (WBC), erythrocyte sedimentation rate (ESR), urinalysis (UA), and serum electrolytes were normal. Her hormone analysis is shown in table 1.

The X ray hand showed the bone age to be 6 years (Pyle and Greulich).

MRI brain revealed an absent anterior pituitary,

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**Table 1**

Thyroid stimulating hormone	1.56 $\mu$ U/ml (0.4-4.5)
FreeT4	0.5 ng/ dl (0.8-2.0)
Follicle-stimulating hormone	<0.1 U/l (1-10)
Luteinizing hormone	<0.1 U/l (2-8)
Prolactin	227 mu/l (0-460)
Peak growth hormone (GH) response (glucagon stimulation test )	<0.1 u/l

ectopic posterior pituitary located in the region of the hypothalamus with absent pituitary stalk (Figure 1).

A diagnosis of panhypopituitarism with intact posterior pituitary function was made and she was treated with thyroxine, hydrocortisone and growth hormone replacement therapy as well as low dose oestrogen therapy to induce puberty.

## Discussion

PSIS is a rare disorder of which the exact aetiology is uncertain. It is characterised by the absence of the pituitary stalk, anterior pituitary hypoplasia, and an ectopically located posterior pituitary. The clinical syndrome may be one of isolated GH deficiency or multiple anterior pituitary hormone deficiencies. The posterior pituitary function has been shown to be normal in these patients (1). A study of 55 patients with PSIS revealed documented breech delivery in 88.9% of the patients and the prevalence of deficiencies in growth hormone, gonadotropins, corticotropin, and thyrotropin were 100%, 95.8%, 81.8% and 76.3%, respectively (2). Timely diagnosis and replacement of the deficient hormones is the key to the management of these patients.

## References

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