Spot diagnosis case 3

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Quiz

A 32-year-old man
presented with short penile length

Present illness

Physical examination

Investigation

Diagnosis

Review

อายุ 13 ปี เรียนชั้น ม.1 ความสูง 119 ซม. เตี้ยที่สุดในห้อง

อายุ 15 ปี ยังเตี้ยกว่าเพื่อนในห้อง น้ำหนักปกติตามวัย สังเกตว่าอวัยวะเพศเล็ก ไม่มีขนขึ้นที่ใบหน้า รักแร้และหัวหน่าว เสียงแหลมเล็ก เคยไปปรึกษาแพทย์โรงพยาบาลประจำจังหวัดแจ้งว่าปกติ จึงไม่ได้ตรวจรักษาต่อ

อายุ 18 ปี เรียนชั้น ปี 1 ไม่ได้เตี้ยเท่าเดิม เริ่มสูงเร็วจนอายุ 20 ปี มีความสูงถึง 165 ซม. แต่ยังคงมีอวัยวะเพศและอัณฑะเล็ก ไม่มีขนที่ใบหน้า รักแร้ และหัวหน่าว เสียงยังแหลมเล็ก ไม่มีความต้องการทางเพศ ไม่เคยมีเพศสัมพันธ์ แต่มีอวัยวะเพศแข็งตัวตอนเช้าได้ 1-2 ครั้ง ต่อสัปดาห์

อายุ 32 ปี (3 เดือน ก่อนมา รพ.) ได้เข้ามาทำงานบรรจุเป็นข้าราชการครู ในกรุงเทพมหานคร จึงตัดสินใจมาปรึกษา

Additional history

Physical examination

Investigation

Diagnosis

Review

- ตั้งแต่เด็กผู้ป่วยรู้สึกว่า ปัสสาวะเยอะ มี nocturia 3-4 ครั้งต่อคืน มีความรู้สึกหิวน้ำบ่อย มีตื่นมาดื่มน้ำตอนกลางคืนเวลาตื่นมาปัสสาวะ ดื่มน้ำประมาณ 8 ลิตร/วัน
- บิดาสูง 170 ซม. มารดาสูง 150 ซม .
  Mid parental height = 166.5 cm ± 8 cm

History

Physical examination

Investigation

Diagnosis

Review

Vital signs	BT 36.6 °C, HR 98/min, BP 139/89mmHg, RR 18/min, SpO2 98% room air
General appearance	alert, well co-operative, no pallor, no jaundice,no cushingoid appearance no syndromic facies
HEENT	no puffy eyelid, no thyroid gland enlargement, no thyroid nodule no prominent Adam's apple no deepening of voice , no temporal balding.
CVS	PMI at 5th ICS MCL, no heaving, no thrill, normal S1S2, no murmur, regular 2+
RS	normal breath sounds, no adventitious sounds
Abdomen	soft, not tender, liver and spleen are not palpable, no increased splenic dullness,
Skin	no rash, no petechiae, no ecchymosis, no dry and coarse skin
Musculoskeletal	no arthritis, no deformity
Nervous system	Cranial nerves : normal Motor : motor power gr. V/V all extremities Sensory : intact all DTR : 2+ all

History

Physical examination

Investigation

Diagnosis

Review

Arthropometric measurement

Body weight 74.1 kg**, Height** 166 cm (BMI 26 kg/m2)

Mid parental height 166.5 ± 8 cm

Arm span 177 cm**, Lower body length** 89.5 cm

Arm span - height : 11 cm

Upper:Lower ratio : 0.855

Breast : Gynecomastia 3 cm in size bilaterally

Hair : No axillary hair, pubic hair tanner stage I

Genitalia

- Flaccid penile length 4 cm
- High scrotal position both sides, testicular volume 1 ml both sides by Prader orchidometer, soft consistency

History

Physical examination

Investigation

Diagnosis

Review

- Testosterone 0.171 ng/ml (2.49-8.36)
- FSH 4.05 mIU/ml (1.5-12.4)
- LH 2.08 mIU/ml (1.7-8.6)
- IGF-1 < 15 ng/ml (69.2-243)
- Prolactin 16.1 ng/ml (4.04-15.2)
- FT4 0.92 ng/dl (0.92-1.68)
- TSH 7.02 mIU/ml (0.27-4.1)
- 8 A.M. cortisol 9.7 mcg/dl (morning 6-10 A.M 6.02-18.4)
- Na 144 mmol/l
- Urine osmole 121 mOsm/kg H2O (50-1200)
- 250 mcg ACTH stimulation test : Cortisol at 0, 30, 60 min = 10.4, 21, 25.1 mcg/dl

MRI

Pituitary gland protocol

MRI pituitary gland protocol

- Relative thinning of left lobe of pituitary gland
- No demonstrate normal pituitary stalk is noted
- Thin linear enhancement above superior pituitary gland, 0.2 cm in length, (questionable hypoplastic of pituitary stalk)
- No demonstrate posterior pituitary bright spot is noted
- No definite pituitary mass is observed
- Visualized both optic nerves, optic chiasm and optic tracts appear unremarkable
- Both cavernous sinuses are unremarkable
- The screening brain parenchyma shows no space taking lesion
- Both olfactory nerves appear unremarkable

MRI pituitary gland protocol

Thining of left lobe of pituitary gland

Absent or hypoplastic pituitary stalk

No demonstrate posterior bright spot

possible

‘Pituitary stalk interruption syndrome’

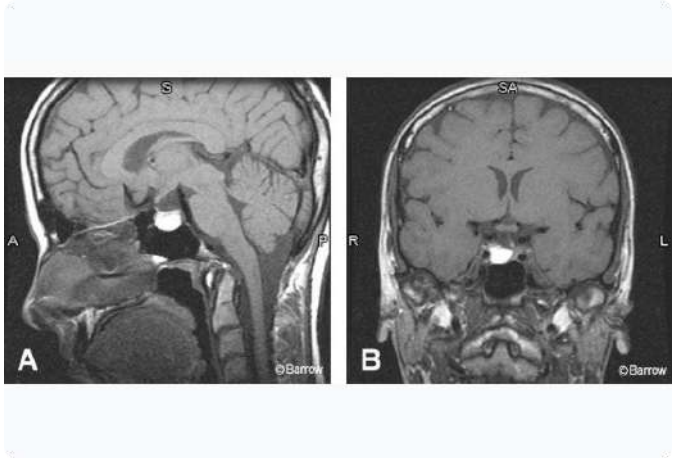
Pituitary Stalk Interruption Syndrome

Pituitary Stalk Interruption Syndrome

Rare congenital disorder of pituitary gland characterized by a specific radiologic triad

- **Interrupted Stalk**: Thin or absent pituitary stalk
- **Ectopic Posterior Lobe:** Posterior pituitary not in the sella
- **Anterior Hypoplasia:** Agenesis or dysgenesis of the anterior lobe

Typically diagnosed in childhood due to growth failure



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Pituitary Stalk Interruption Syndrome: A Case Series.Cureus 17.5 (2025)

Pathogenesis and mechanism

Genetic Factors

Mutations in the genes associated with hypothalamic-pituitary development signaling pathways (Wnt, Notch, Shh)

PIT1, PROP1, **HESX1**, LHX3/LHX4, PROKR2, OTX2, TGIF, ROBO1, and GPR161

** Rarely familial, Male predominance

Environmental Factors

Breech Delivery
Occurred in 88.9% of PSIS patients in some studies

Perinatal Asphyxia
Potential traumatic injury to the fragile pituitary stalk

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Clinical spectrum

Hypoglycemia
Jaundice
Micropenis
(MPHD : Variable presentation)

Short stature (70%) due toGHD

↓

100% prevalence followed by
GnRH deficiency (86%)
Corticotropin deficiency (75%)
Tryrotropin deficiency (79%)
Hyperprolactinemia (7%)

Absent puberty
Panhypopituitarism

Neonatal

Childhood

Adult

** Sometimes present with additional
extra-pituitary symtoms : midline abnormalities; CVS, Eye structure
extracerebral abnormalities : skin, limb, facial hypoplasia

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Diagnosis of PSIS

- Based on **radiological findings** and supported by **clinical and hormonal evaluation**

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Imaging of PSIS

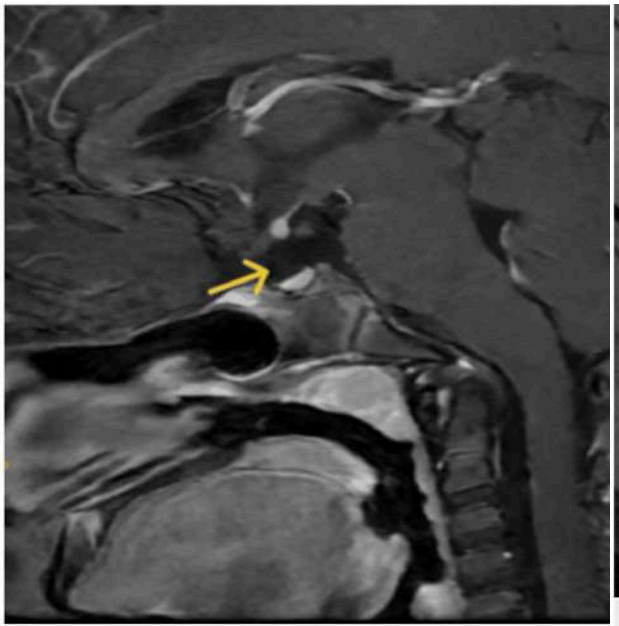

FIGURE 1: Sagittal T1-weighted contrast-enhanced magnetic resonance imaging of the sella turcica shows absent pituitary infundibulum (yellow arrow) in Case 1

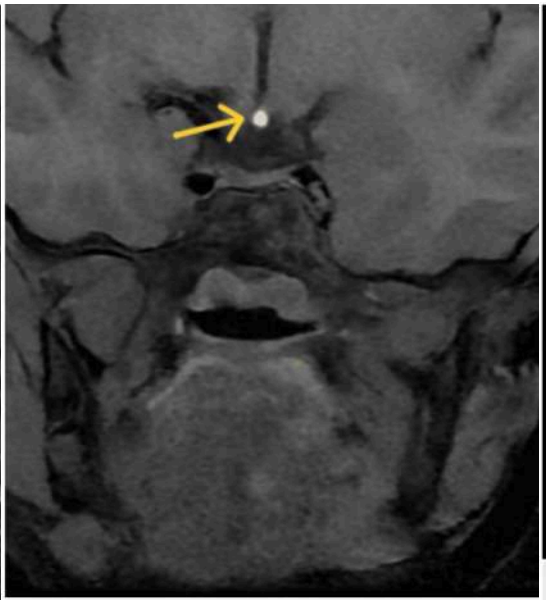

FIGURE 2: Contrast-enhanced magnetic resonance imaging of the sella turcica of Case 1

Contrast-enhanced magnetic resonance imaging of the sella turcica showing an ectopic posterior pituitary (yellow arrow) with a hypoplastic anterior pituitary measuring 2.3 mm in height, 5.4 mm in the anteroposterior dimension, and 8.1 mm in coronal diameter, and absent/thin pituitary infundibulum.

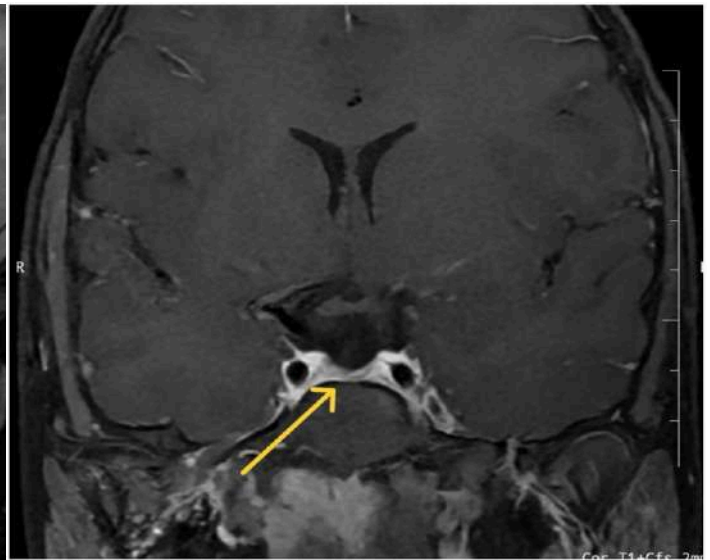

FIGURE 3: Contrast-enhanced magnetic resonance imaging of the sella turcica of Case 2

Contrast-enhanced magnetic resonance imaging of the sella turcica showing an ectopic posterior pituitary with a hypoplastic anterior pituitary (yellow arrow) measuring 1.9 mm in height, 4.6 mm in the anteroposterior dimension, and 6.8 mm in coronal diameter, and absent pituitary stalk.

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Imaging of PSIS

FIGURE 4: Sagittal T2-weighted MRI of sella turcica showing absent pituitary stalk (yellow arrow) of Case 2

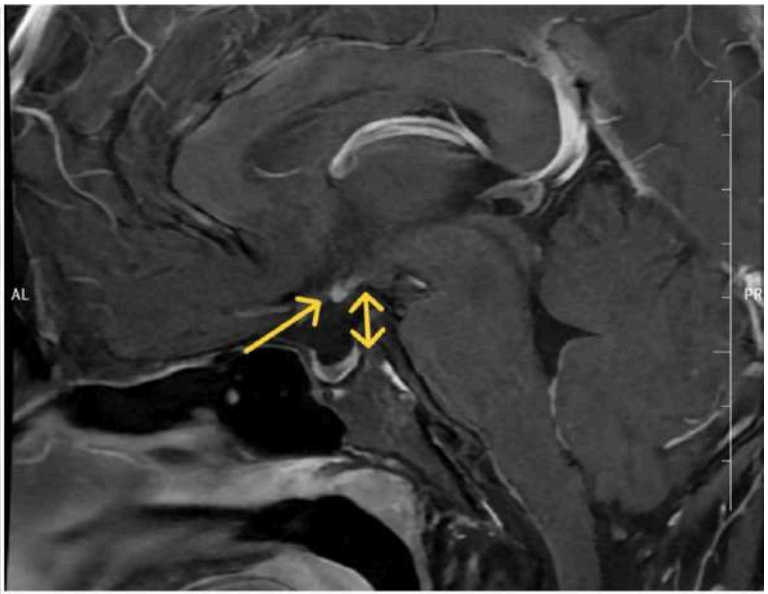

FIGURE 5: T1-weighted contrast enhanced MRI of sella turcica showing an ectopic posterior pituitary (yellow arrow) and absent pituitary infundibulum in Case 3

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Treatment of PSIS

- Early hormone replacement
  (promoted the development of secondary sexual traits and adult height at nearly normal rate)

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