Stump the Professors

Professor Panelists

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Disclosures

- No conflicts of interest or disclosures

Case: Delayed Puberty

- Chief Complaint: Delayed Puberty
- Patient presented for evaluation at age 14 with no genital hair and no breast development. She had not had any vaginal bleeding consistent with a period and no pelvic pain or cramping.
Case: Delayed Puberty

Past Medical History:
- Renal failure diagnosed at age 12, receiving home peritoneal dialysis and on the renal transplant list
- History of prior bilateral inguinal hernia repair

Family History:
- 10 year old sister has already achieved menarche
- Mother—hypothyroidism and hypercholesterolemia
- Paternal aunt—lupus

Physical Exam
- T: 37.2 HR 88 BP 129/65 RR 18 O2 sat 98%
- Ht 162cm Wt 53.5kg BMI 20
- Tanner Stage I-II breast and genital development
- Normal external female genitalia, hymen was patent and intact
- No evidence of hirsutism, acne, clitoromegaly, or thyromegaly

Social History
- Patient is in 8th grade where she earns A’s and B’s and enjoys painting and music
- Lives with her mom, dad, and siblings
- Patient is not yet sexually active

Stump the professors Case
Questions or comments from the panelists
Stump the professors Case

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Case: Delayed Puberty

Laboratory Data
- TSH 1.8 mIU/mL
- LH 6.4 mIU/mL
- FSH 161.5 mIU/mL
- Prolactin 150.7 ng/mL
- Estrogen 7.5 pg/mL

MRI Pelvis: Uterus present within the pelvis, unremarkable in appearance. Possible, though not definitive, ovarian tissue present bilaterally. Moderate to large volume of fluid in the pelvis secondary to peritoneal dialysis.

MRI Head: Normal non-contrast MRI of the head. The pituitary gland is not enlarged.

Case: Delayed Puberty
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- Additional Studies:
  - Karyotype 46XY
  - FISH SRY +

Stump the professors Case

Questions or comments from the panelists

Case: Delayed Puberty

- Differential Diagnosis for Hypergonadotropic Hypogonadism:
  - XY Pure Gonadal dysgenesis
  - Premature ovarian insufficiency
  - Turner Syndrome
  - Radiation/Chemotherapy
  - 17-alpha hydroxylase deficiency
  - Autoimmune oophoritis
  - LH and FSH receptor defects
Case: Delayed Puberty

Diagnosis
- Frasier Syndrome
  - Rare genetic syndrome characterized by
    - Steroid-resistant nephrotic syndrome
    - 46XY gonadal dysgenesis
    - WT1 mutation
    - High risk of gonadoblastoma
  - Reported risk of gonadal tumor ranges from 48-67%
- Hyperprolactinemia
  - Prolactin is renally eliminated in the renal tubules

Case: Delayed Puberty

Additional Procedures:
- Renal transplant
- Bilateral gonadectomy
  - Pathology revealed bilateral gonadoblastoma
  - Tumor confined to the gonads

Additional Studies:
- WT1 mutation: c.1432+5G>A

Case: Delayed Puberty

Treatment:
- Gynecologic
  - Started on hormone replacement therapy, now with cycling progesterone
  - Tanner stage IV breast development
- Renal
  - Creatinine and BP stable
  - No evidence of kidney transplant rejection
Case: Delayed Puberty

- **Treatment**
  - Oncology
    - Negative CT and tumor markers
  - Endocrine
    - Normalized prolactin

- **Outcome**
  - Patient is doing well, continuing in school
  - She has started modeling for local businesses around Albuquerque
  - Recently started dating her first boyfriend

Uterus measures 6.1 x 3.6 x 2.4 cm
Comments from Panelists

Case: Delayed Puberty

Take home points: Frasier Syndrome

- Characterized by renal disease and XY gonadal dysgenesis
- Majority of patients present with renal disease but diagnosis is not made until delayed puberty
- 67% of patients had gonadoblastoma at time of gonadectomy

References

NASPAG would like to thank the Expert Panel and the Resident/Fellow Challengers.