Pseudohermaphroditism due to Congenital Adrenal Hyperplasia

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Agenda

1. Case Presentation

2. Image Findings

3. Discussion
   I. Definition/Incidence
   II. Pathogenesis of Disorder of Sexual Differentiation associated to CAH
   III. Diagnosis/Radiographic Modalities
   IV. Treatment

4. Take Home Points

5. Acknowledgements

6. References
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Our Patient: Case presentation

13-month old female, who presented with:

• Clitorormegaly

• Partial fusion of the posterior labioscrotal folds

• Single aperture in urogenital area

Courtesy of Dr. David Diamond
Our Patient: History

Patient was referred with following information:

• Diagnosed:
  – *Congenital Adrenal Hyperplasia*
  • Due to:
    – High level of 17-hydroxyprogesterone
    – *Genital Ambiguity*
    – *US abdomen + Pelvis: normal*

...For evaluation and surgical management.
Our Patient: Evaluation

First step of evaluation:

- VCUG

Voiding – CystoUrethroGram
But... What is VCUG?
VCUG: General Information

• **Gold standard** method of detecting Vesicoureteric reflux.

• **Continuous radiographic test with contrast to assess and evaluate:**
  – Kidney
  – Ureter
  – Bladder
  – Urethra
VCUG: General Information

• Surgical work-up to display urogenital anatomy:
  – type of urethra
  – presence of vagina / cervix / urogenital sinus
  – fistulous communications

• Most of the time it is *all that is necessary* for anatomic evaluation of the urinary tract.
  – eliminates the need for endoscopy, CT and MRI
  • reserved for inconclusive findings

• Vallerie A, Breech L. Update in Mullerian anomalies: diagnosis, management, and outcomes. Current Opinion in Obstetrics & Gynecology. Oct 2010; 22(5); 381-387
VCUG: General Information

Complications
• Pain: Insertion of catheter / Post-Void
• Urinary tract infections
• Reaction to Iodine-based dye

Radiation? = the amount of background radiation a person gets in 9 days

Use of Midazolam
– is effective with no apparent effect on voiding dynamics.
– causes amnesia, reduces distress

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Image: Our Patient: VCUG with full Bladder

VCUG, Lateral view; with perineal opening marker. Filled Bladder. Courtesy of Dr. Mei Mei Chow.
Image: Our Patient: VCUG; Voiding

VCUG, Lateral view; with perineal opening marker. During Void. Courtesy of Dr. Mei Mei Chow

Bladder, filled with contrast

Urethra

Arrow-Shaped Metallic Marker placed on perineal opening

Vagina Vault

Urogenital Sinus
VCUG, Lateral view; with perineal opening marker.
During Void.
Courtesy of Dr. Mei Mei Chow

Distance: 1.1cm

Urogenital Sinus

Arrow-Shaped Metallic Marker placed on perineal opening
• So the finding represents the presence of Urogenital Sinus.

• But... What is *Urogenital Sinus*?
Urogenital sinus Abnormality

• Defect present at birth, vagina and urethra open into a common channel.

• This structure may persist in patients with Congenital Adrenal Hyperplasia, due to excessive androgens exposure.

• 2 types: Low vs. High
Diagnosis:

• Congenital Adrenal Hyperplasia
  – Disorder of Sexual Differentiation
    • Urogenital sinus
    • Clitomegaly
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What are some of possible causes of *Disorder of Sexual Differentiation*?
Disorders of Sexual Differentiation

- Sex chromosome
  - X0
  - XXY
  - Maternal Androgens
  - Androgen Excess
  - D. Ovarian Develop,
  - D. Testicular Develop,
  - D. Androgen Synthesis
  - D. androgen-dependent target tissue

D. = Disorder of; Develop. = Development

CAH and DSD

• Congenital Adrenal Hyperplasia (CAH) in Female
  – Most common cause disorder of sexual differentiation (DSD)
  • represents ~60-70% of ambiguous genitalia in newborn

• Incidence: 1:14500

CAH and DSD

• Most cases are due to autosomal recessive deficiency of:
  
  – 21-hydroxylase

  – Which causes steroid hormone synthesis to be diverted towards the androgen pathway

• Females: High levels of androgens causes virilization of the external genitalia.
CAH and DSD

Female:

- Around 12 weeks of gestation, there is separation of Vagina and Urethra
- This is a Non-hormone Dependent Process

- But if there is exposure to androgens, female fetus can have:
  - Labial fusion / Phallic urethra / Urogenital sinus
  - Clitoral enlargement / Scrotalisation of the labial folds.

Urogenital Sinus: Diagnosis/Radiographic Modalities

• Imaging plays an important role in depicting the internal organs and urogenital anatomy.
  – Cystourethrogram / Genitogram (lateral view)
  – Endoscopy / Cystoscopy
  – US
  – CT / MRI

Image: VCUG: Companion Patients 2, Urogenital Sinus

Urogenital Sinus: Role of Radiologist

- Provide correct/full name diagnosis
- Provide information to guide surgical approach.

Description of Genital ambiguity and urogenital sinus should include:

- Degree of masculinzation
- Phallic Size
- External genital appearance
- Location of Vaginal Confluence

R. Rink, M. Adam, R. Misseri. A new classification for genital ambiguity and urogenital sinus anomalies. BJUinternational 2005; 95, 638-642
Urogenital Sinus: Role of Radiologist

**PVE classification:**


**P**: Phallic length and width.
  - measured in cm in flaccid state

**V**: Vaginal Location
  - Distance from the bladder neck to the confluence and length of the common urogenital sinus.

**E**: External genital appearance.
  - Prader number

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Urogenital Sinus: Treatment

• Feminizing surgery (within 18 months of life)
  – genitoplasty (Clitoral reduction + vaginoplasty)

• Cosmetic results -> Good

References:
Our Patient: Plan

• With the information obtained from the VCUG, patient was planned to undergo surgery in 2 weeks.

• Due to the short length of the urogenital sinus (1.1cm), she was expected to have good outcome with low risk of complications.
Take Home Points

• We discussed case presentation of female patient with Congenital Adrenal Hyperplasia and Disorder of Sexual Differentiation, Urogenital Sinus.

• You saw images of VCUG, showing the presence of Urogenital Sinus.

• We discussed about Congenital Adrenal Hyperplasia and how it causes Disorder of Sexual differentiation.

• We discussed about different radiographic studies that can be used to assess urogenital sinus; and the important role of radiologist.

• You saw images of physical examination, VCUG and cystoscopy of other companion patients with Urogenital Sinus.
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References (1/2)

References (2/2)

• R. Rink, M. Adam, R. Misseri. A new classification for genital ambiguity and urogenital sinus anomalies. BJUinternational 2005; 95, 638-642