Williams OB Chapter 3: Congenital Genitourinary Abnormalities

**CREOG Learning Objectives:**

1) Describe the embryology of the pelvis and pelvic organs:

a. Describe normal development

b. Describe abnormal development, including ambiguous genitalia, Mullerian agenesis, and vaginal/uterine septum

2) For the Mullerian agenesis, describe the appropriate screening, diagnosis, pertinent history, focused physical examination, diagnostic testing and treatment, and indications for referral

3) Understand and be able to perform hysteroscopic resection of a uterine septum

**Practice Questions:**

1) A 14-year old patient presents with severe menstrual cramping, predominantly on the right side, that persists for the duration of menstrual flow. The pain is improved, but not relieved, with the use of NSAIDs. She had menarche 6 months before presentation and her menses occur every 5-6 weeks, lasting 5 days. She is healthy but was noted to have right renal agenesis on prenatal ultrasonography when she was a fetus. On abdominal examination, she has no masses or tenderness in the abdomen and external genitalia are normal. The best next step in her management is

A. combined oral contraceptive pills (OCPs)

B. depot medroxyprogesterone acetate (DMPA)

C. diagnostic laparoscopy

D. menstrual calendar

E. transabdominal ultrasonography

Source: PROLOG REI 8th edition #117

2) A 15-year old adolescent patient is referred to you by her pediatrician because of amenorrhea and absent breast development. She is generally healthy otherwise. She has a body mass index (BMI; 30). Her breasts are Tanner stage 1. She has a normal vaginal length on examination and Tanner stage IV pubic hair. Her FSH is 73 mIU/mL, and her estradiol concentration is less than 20 pg/mL. Her fragile X premutation screening is negative, and her karyotype is 45,X/46,XY. Her transabdominal ultrasonography shows a prepubescent uterus and ovaries are not seen. The next step in her management is to

A. initiate low-dose oral contraceptives (OCs)

B. initiate low-dose transdermal estradiol

C. measure her testosterone concentration

D. perform laparoscopic gonadectomy

Source: PROLOG REI 8th edition #78

3) You have been called to evaluate a neonate after delivery. On physical examination, the genitalia are ambiguous. You suspect congenital adrenal hyperplasia (CAH). The neonate’s electrolytes are normal and you await results of a karyotype, pelvic ultrasonography, and a steroid hormone profile. The most appropriate next step in the management of this neonate is to

A. perform a cosyntropin stimulation test

B. assign preliminary gender and meet with the family

C. consult with a pediatric surgeon

D. use a multidisciplinary team to assist in gender reassignment

E. start adrenal steroid replacement

Source: REI Prolog 7th edition #115

4) For each diagnosis, choose the appropriate karyotype and testosterone level:

1) Complete androgen insensitivity syndrome

2) Pure gonadal dysgenesis

3) Trisomy X syndrome

A. 46, XX karyotype, serum T in female range

B. 46, XX karyotype, serum T in male range

C. 46, XY karyotype, serum T in female range

D. 46, XY karyotype, serum T in male range

E. 47, XXX karyotype, serum T in female range

F. 47, XXY karyotype, serum T in male range

**High-Yield Resources:**

1) Committee Opinion #728: Mullerian agenesis: Diagnosis, Management, and Treatment

2) Committee Opinion # 729: Management of Acute Obstructive Uterovaginal anomalies

3) Committee Opinion # 780: Diagnosis and Management of Hymenal Variants

4) CREOGS over coffee Episode 80: Mullerian Anomalies and Variants <https://creogsovercoffee.com/notes/2020/3/29/mullerian-anomalies>

Answers

1. e 2. d 3. d 4.1 d 4.2 c 4.3 e