Educational Topic 42: Puberty

Rationale: Puberty consists of physical and emotional changes associated with the maturation of the reproductive system. In order to provide appropriate care and counseling, the physician must have an understanding of normal puberty, and recognize deviation from normal.

Intended Learning Outcomes:

A student should be able to:

- Describe the changes in the hypothalamic-pituitary-ovarian axis and target organs during normal puberty
- Explain the normal sequence of pubertal events and ages at which these changes occur
- Discuss the psychological issues associated with puberty
- Define precocious and delayed puberty and describe the steps in the initial evaluation of these conditions

TEACHING CASE

CASE: A 15 year-old female comes in for exam because she has not had her period. She seemed to be developing normally and had normal breast development that started about 3 years ago and she has pubic hair. She met her developmental milestones in childhood and is of normal height and weight. She has not had any significant medical illnesses. Her ROS is negative and her family history is negative.

She is active in school and is a cheerleader. She works out with the team and runs. She does well in school. She lives at home with her mom, dad and sister. She reports she has a boyfriend but has not been sexually active.

On physical exam, she is well appearing, BP is 100/60, weight 130 pounds, height 5 feet 7 inches. Breast exam: appear symmetric, areola are darkened bilaterally with nipple continuous with the areola, abdomen: soft, non-tender, no masses and external genitalia: soft straight hair covering the mons but not extending to the thighs.

COMPETENCY-BASED DISCUSSION & KEY TEACHING POINTS:

Competencies addressed:
- Patient Care
- Medical Knowledge
- Systems-based Practice
1. What Tanner stage is this patient?
   - Tanner 3
     - Breast:
       - Tanner stage 1-pre-pubertal breast
       - Tanner stage 2-breast bud
       - Tanner stage 3-further breast and areolar enlargement
       - Tanner stage 4-areola and papilla form secondary mound
       - Tanner stage 5-areola returns to level of the breast
     - Pubic Hair:
       - Tanner stage 1-no pubic hair
       - Tanner stage 2-sparse hair on labia majora
       - Tanner stage 3-pubic hair to mons pubis
       - Tanner stage 4-adult hair with no spread to thighs
       - Tanner stage 5-spread to thighs

   **KEY LEARNING POINT:** Students should be able to describe clinical findings technically and understand them when presented technically. Communication among various healthcare providers requires that we all speak the same “language.”

2. What is the normal process of puberty in girls?
   - Proceeds in sequence: thelarche, adrenarche, pubarche, growth spurt, menarche (20% of the time adrenarche precedes thelarche, most commonly in African American girls)

3. What is the differential diagnosis for this patient’s presentation?
   - Pregnancy
   - Delayed puberty defined as:
     - No secondary sexual characteristics by age 13
     - No menarche by 15-16 or 5 years after thelarche
   - Common causes in order of frequency:
     - Hypergonadotrophic hypogonadism
       - Inadequate estrogen secretion
       - Gonadal dysgenesis
       - Turner’s syndrome
       - Premature ovarian failure (younger than 40)
     - Hypogonadotropic hypogonadism
       - Inadequate GnRH (hypothalamic dysfunction), Kallman’s syndrome
       - Weight loss, excessive exercise, psychological stress, chronic illness related stress, brain or head injury, brain tumor
       - Inadequate gonadotropin secretion (pituitary dysfunction)
       - Pituitary adenoma
       - Gonadotropin deficiency
       - Inadequate body fat
       - Anorexia/bulimia
       - Excess exercise
       - Thyroid dysfunction
       - Eugonadism

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• Androgen insensitivity syndrome (Testicular feminization 46XY)
• Müllerian agenesis (Mayer-Rokitansky-Kuster-Hauser)
• Outflow tract obstruction
  • Imperforate hymen
  • Vaginal septum

  • Medications
    • Phenothiazines, antidepressants, hypertension meds, metoclopramide, all can cause elevated prolactin
    • Narcotics, estrogens can result in anovulation

4. What key test must be performed first?
   • Pregnancy test

   **KEY LEARNING POINT:** All patients are pregnant until proven otherwise.

5. What further history must be elicited?
   • Dietary and exercise habits (anorexia, bulimia)
   • Sexual history in a confidential setting

   **KEY LEARNING POINT:** Physical, psychological and cognitive development are uneven in adolescents which may increase the risk of adverse outcomes. Confidentiality can be an area of concern in approaching adolescents. Laws vary by state; however, every state allows minors to make independent decisions about treatment for reproductive healthcare (other than abortion). Both assuring an adolescent that her confidentiality will be maintained and knowing the state laws are important components of the doctor-patient relationship.

6. What further studies will help refine the diagnosis?
   • Physical exam
     • Pelvic exam: Only do the minimum amount of pelvic exam needed. If you can obtain the information with imaging, do so in this young patient.
   • Prolactin 11 ng/ml (normal range <22)
   • TSH 2.11 uIU/ml (normal range 0.5-4.0)
   • FSH 6.8 mIU/ml
   • LH 8.3 mIU/ml
   • Ultrasound shows no uterus present; bilateral ovaries 3 x 2 cm with multiple small antral follicles

7. If the patient has eugonadotropic eugonadism, what further testing should be performed?
   • Abdominal pelvic ultrasound to determine the presence/absence of a uterus and gonads
   • If uterus absent, check testosterone level, karyotype to distinguish between mullerian agenesis and androgen insensitivity (AIS)
     • If mullerian agenesis: needs renal evaluation
     • If AIS, needs removal of gonads

8. How would you define precocious puberty, what is the most common cause and what psychosocial issues does this raise?
• Precocious puberty refers to the development of any sign of secondary sexual maturation at an age earlier than 2.5 standard deviations less than the expected age of pubertal onset
• In North America, these ages are 8 years for girls.
• About 75% of cases of precocious puberty in girls prove to have a constitutional or idiopathic cause, and these patients are candidates for GnRH agonist (e.g., leuprolide acetate) therapy.
• Most children with sexual precocity have few significant behavioral problems, but emotional support is important in these children. Behavioral expectations by family members and teachers should be based on the child's chronologic age, which determines psychosocial development, and not on the presence of secondary sexual characteristics.

REFERENCES
