

Disorders of Growth and Development: Review Questions

Frank J. Genuardi, MD, MPH

QUESTIONS

Choose the single best answer for each question.

Questions 1 and 2 refer to the following case.

A 9-year-old boy presents to his primary care physician because his parents are concerned about his height. He is the shortest boy in his 4th grade class. He was born at term without complications and has never been hospitalized or had any surgeries. He does well in school and is active in sports. His father is 5 ft 8 in, and his mother is 5 ft 3 in. On physical examination, his height is 48.5 in (5th percentile for age), he weighs 52 lb (10th percentile for age), and his body mass index (BMI) is 15.5 kg/m² (35th percentile for age). He has no pubic, axillary, or facial hair. He has normal male external genitalia, and testes are descended bilaterally with a testicular volume of 2 cm³ (prepubertal). The remainder of his physical examination is normal.

- Which of the following is the most appropriate next step in the management of this patient?
 - Bone age determination
 - Computed tomography scan of the head
 - Karyotype determination
 - Testing of the serum testosterone level
 - Thyroid function testing
- Which of the following is this patient's most likely diagnosis if testing shows that bone age is consistent with chronologic age?
 - Growth hormone deficiency
 - Hypothyroidism
 - Klinefelter's syndrome
 - Malnutrition
 - Normal growth
- A 13-year-old girl presents to her primary care physician because her mother is concerned that she has not yet begun menstruating. Maternal menarche was at age 11 years. The patient is on no medications and has never been hospitalized or had any surgeries. She is in the 7th grade, does well in school, and is active in sports. She denies sexual activity. On physical examination, her height is 60 in (25th percentile for age), she weighs 94 lb (35th percentile for age), and her BMI is 18.4 kg/m² (45th percentile for age). She has Tanner stage 3 breasts and pubic hair. Which of the following is the most appropriate next step in this patient's management?
 - Bone age determination
 - Estrogen therapy
 - Follicle-stimulating hormone and luteinizing hormone determination
 - Reassurance
 - Thyroid function testing
- A 15-year-old girl presents to her primary care physician because her mother is concerned that she has not yet begun menstruating. Maternal menarche was at age 13 years. The patient is on no medications and has never been hospitalized or had any surgeries. She is in the 9th grade and does well in school. She does not participate in sports but does play in the school band. She denies sexual activity. On physical examination, her height is 54 in (< 5th percentile for age), she weighs 102 lb (25th percentile for age), and her BMI is 24.6 kg/m² (85th percentile for age). She has Tanner stage 1 breasts with widely spaced nipples and Tanner 3 stage pubic
 - Bone age determination
 - Estrogen therapy
 - Follicle-stimulating hormone and luteinizing hormone determination
 - Reassurance
 - Thyroid function testing

Dr. Genuardi is an associate professor, Department of Pediatrics, and assistant dean for educational affairs, University of Florida Health Science Center, Jacksonville, FL.

hair. Which of the following is this patient's most likely diagnosis?

- (A) Craniopharyngioma
- (B) Hypothyroidism
- (C) Normal development
- (D) Polycystic ovarian disease
- (E) Turner's syndrome

ANSWERS AND EXPLANATIONS

1. **(A) Bone age determination.** This patient should undergo bone age determination, which would help to exclude growth hormone deficiency and confirm that skeletal maturation is normal. The patient has no symptoms or physical findings to warrant obtaining a computed tomography scan of the head. Growth parameters are consistent with adequate nutrition, and hypothyroidism is unlikely given normal mental functioning and physical activity. No physical findings consistent with precocious puberty are present to prompt testing of serum testosterone levels. If Klinefelter's syndrome were suspected, karyotype testing would be indicated. Although patients with Klinefelter's syndrome have small testes, patients are tall rather than short for their age.
2. **(E) Normal growth.** If bone age determination is consistent with chronologic age, this patient is growing normally and most likely has familial short stature.

The most appropriate management at this point would be to reassure the patient and his family and to continue to follow his growth at regular intervals.

3. **(D) Reassurance.** This patient is growing and developing normally. In the presence of normal development of secondary sexual characteristic as seen in this patient, menarche is expected by age 16 years. Patients without development of secondary sexual characteristics should be evaluated earlier, generally by age 14 years. No laboratory evaluation is indicated at this time nor is any therapeutic intervention.
4. **(E) Turner's syndrome.** Amenorrhea in a patient with short stature and the presence of pubic hair but not breast development should raise suspicion of Turner's syndrome, a genetic abnormality of females in which all or part of one X chromosome is missing. Affected patients have growth problems as well as delayed and incomplete sexual development, and are generally infertile. Characteristic physical findings of Turner's syndrome, such as widely spaced nipples, makes evaluation with a karyotype determination imperative. Patients with polycystic ovarian disease may have amenorrhea but generally also have hirsutism and acne. Hypothyroidism is less likely given normal mental functioning, and the patient has no symptoms, such as headache, that would suggest a central nervous system tumor.

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PEDIATRIC ENDOCRINOLOGY

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