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**QUESTION 1**

A 10-year-old girl is brought to the office by her mother because her school nurse thinks that she may have Marfan syndrome. She is at the 95th percentile for height and 25th percentile for weight. Physical examination shows a narrow palate with dental crowding, long slender extremities, and joint laxity. Molecular testing for FBN1 shows a single nucleotide difference that does not change the amino acid at that locus. Her mother is 163 cm (5 ft 4 in) tall. There is no clinical evidence of Marfan syndrome in the mother, but she has the same single nucleotide change as the patient. The same nucleotide change is found in 15 of 200 individuals without Marfan syndrome. Which of the following best describes the single nucleotide change in the patient and her mother?

- A. It is a disease-causing mutation in the patient and her mother
- B. It is a polymorphism
- C. It is a sequence variant of unknown significance
- D. It will change the folding of the protein
- E. It will result in a truncated protein

Correct Answer: B

QUESTION 2

A couple presents to a clinic for workup of infertility after 5 years of unprotected intercourse. The wife denies any medical problems and notes regular menstrual cycles. The husband states that he has had chronic sinusitis and lower respiratory tract infections. Physical examination of the woman is unremarkable. Examination of the man is remarkable for dextrocardia. Further workup of the husband will most likely reveal

- A. azoospermia
- B. germinal cell aplasia
- C. immotile sperm
- D. isolated gonadotropin deficiency
- E. varicocele

Correct Answer: C

Explanation:

The husband is suffering from Kartagener syndrome, an autosomal recessive disorder characterized by infertility, situs inversus, chronic sinusitis, and bronchiectasis. The underlying cause of these varied manifestations is a defect in the dynein arms, which are spokes of microtubule doublets of cilia in the airways and the reproductive tract. Since sperm motility is dependent on the functioning of cilia, infertility frequently accompanies this disorder. Situs inversus occurs because ciliary function is necessary for cell migration during embryonic development. Azoospermia is not a feature of Kartagener syndrome, as sperm production or survival is not affected in this disorder. Germinal cell aplasia, also known as Sertoli-only syndrome, is characterized by oligospermia or azoospermia. Isolated gonadotropin deficiency is characterized by delayed or incomplete pubertal maturation. Varicocele results in an increased testicular temperature, decreasing the count of normal, viable sperm.

**QUESTION 3**

A 33-year-old woman comes to the physician because of a 2-day history of mild nausea, increased urinary urgency and frequency, and constipation. She also has had a 4.5-kg (10-lb) weight loss during the past 2 weeks and a 3-week history of vaginal bleeding. Pelvic examination shows a nodular cervix with an irregular, friable posterior lip, and a rock-hard, irregular, immobile pelvic mass that extends across the pelvis. Examination of biopsy specimens from the cervix and anterior wall of the vagina show well-differentiated keratinizing squamous cell carcinoma. Which of the following best describes the pathogenesis of this patient's disease?

- A. Inactivation of cellular p53
- B. Insertion of viral promoters adjacent to cellular growth factor genes
- C. Specialized transduction
- D. Transactivation of cellular growth factor genes by TAX
- E. Translocation of CMYC to an Ig gene promoter

Correct Answer: A

QUESTION 4

Which of the following is caused by a Vitamin D deficiency?

- A. Edema
- B. Anemia
- C. Lupus
- D. Rickets

Correct Answer: D

QUESTION 5

Object permanence for toddlers develops in this age range?

- A. 5–10 months
- B. 10–14 months
- C. 12–24 months
- D. 15–24 months



Correct Answer: C

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