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QUESTION 1

Which of the following syndromes corresponds to: can be caused by high doses of Tetracyclines?

- A. Dubin-Johnson syndrome
- B. Fanconi's syndrome
- C. Edward's syndrome
- D. Cri-du-chat syndrome

Correct Answer: B

QUESTION 2

Which of the following is not directly related with Goodpasture syndrome?

- A. Hematuria
- B. Anemia
- C. Adrenal atrophy
- D. Hemoptysis

Correct Answer: C

QUESTION 3

A 25-day-old male is brought to the physician due to seizures, rigidity and frequent viral infections. Physical examination reveals a neonate with cyanosis in the lower extremities. The most likely cause of these findings is a

- A. defect in the IL-2R gamma chain
- B. defect in the lysosomal trafficking regulation gene
- C. defect in the tyrosinase kinase gene
- D. deletion in chromosome 22q11.2
- E. mutation in the STAT3 gene

Correct Answer: D

Explanation: The patient most likely has thymic aplasia which is also known as DiGeorge syndrome. It is caused by a deletion in chromosome 22q11.2 which leads to failure of the third and fourth pharyngeal pouches to develop. The



features of DiGeorge syndrome include conotruncal cardiac anomalies such as tetralogy of Fallot and truncus arteriosus, an absent thymus gland which leads to T-cell deficiency and frequent viral infections, and hypocalcemia due to a hypoplastic parathyroid. The congenital heart defect may lead to cyanosis and the hypocalcemia can present with seizures and tetany. Laboratory findings include decreased T cell levels, decreased parathyroid hormone, decreased calcium levels and a thymic shadow on a chest x-ray.

QUESTION 4

A 2-year-old boy is brought to the physician for a well-child examination. He was delivered at term after an uncomplicated pregnancy. His birth weight was 3500 g (7lb11oz), and Apgar scores were 8 and 10 at 1 and 5 minutes, respectively. At the age of 15 months, physical examination showed no abnormalities, but he was not yet talking. Both of his parents had learning difficulties in school, and his mother stopped attending after the 10th grade. He has a maternal uncle with cognitive disabilities. He is at the 25th percentile for height, 15th percentile for weight, and 90th percentile for head circumference. He appears irritable, he resists making eye contact, and he is flapping his hands. Which of the following is the most likely cause of this patient's condition?

- A. Creation of an alternative splice site
- B. Frameshift mutation
- C. Missense mutation
- D. Nonsense mutation
- E. Trinucleotide repeat expansion

Correct Answer: E

QUESTION 5

Which of the following is not a risk factor for hypertension?

- A. Genetics
- B. Obesity
- C. Youth
- D. Smoking

Correct Answer: C