

Original question:

A 4-week-old boy is brought to the pediatrician by his parents for an initial evaluation. His parents are concerned that he is not feeding well and has lost weight over the last 2 weeks. {...} He does not appear to respond to visual stimuli, and further examination reveals bilateral clouding of the lens. Which of the following interventions could have avoided this patient's symptoms?

{ 'A': 'Avoiding fruit juice and sweetened foods', 'B': 'Changing to a soy based formula', 'C': 'Providing imiglucerase enzyme replacement', 'D': 'Removing phenylalanine from maternal diet during pregnancy', 'E': 'Vitamin B6 supplementation' }

Original explanation:

This patient who presents with failure to thrive, hepatosplenomegaly, and bilateral cataracts most likely has classic galactosemia. Patients with this disorder should avoid lactose-containing products by changing to a soy-based formula. Classic galactosemia is an autosomal recessive defect in galactose-1-phosphate uridylyltransferase. This enzyme is involved in the conversion of galactose to glucose, and a deficiency of this enzyme results in the accumulation of galactose 1-phosphate in the liver, kidney, and brain. This metabolite acts as a phosphate sink, meaning that it traps all free phosphate in the cytosol and inhibits the formation of other phosphate-dependent metabolites such as adenosine triphosphate. {...}

Step-by-step explanation:

Step 1: Analyze the patient's symptoms and clinical presentation. The 4-week-old boy presents with failure to thrive, hepatosplenomegaly, jaundice, and bilateral cataracts.

Step 2: Recognize the pattern of symptoms. The combination of these symptoms suggests a metabolic disorder, specifically classic galactosemia, which is characterized by the inability to metabolize galactose properly due to an enzyme deficiency.

{...}

Step 5: Select the correct answer based on the reasoning. The intervention that could have avoided the patient's symptoms is 'B': Changing to a soy-based formula.

Conclusion: The correct intervention to avoid symptoms of classic galactosemia in this patient is to switch to a soy-based formula, as this eliminates dietary galactose.

Key points:

["failure to thrive", "hepatosplenomegaly", "bilateral cataracts", "classic galactosemia", "avoid lactose-containing products", {...} "eczema", "homocystinuria", "marfanoid appearance", "arachnodactyly", "galactose-free", "lactose-free"]