A 3-day-old male neonate presents with poor feeding, vomiting, lethargy, and seizures. Laboratory investigations reveal markedly elevated plasma ammonia levels, very low blood urea nitrogen, and respiratory alkalosis. There is no evidence of hypoglycemia or metabolic acidosis.

Which of the following enzyme deficiencies is the most likely cause?

A. Arginase
B. Argininosuccinate lyase
C. Ornithine transcarbamylase
D. Argininosuccinate synthetase
E. Carbamoyl phosphate synthetase I

A 2-week-old infant presents with failure to thrive and recurrent vomiting. Laboratory tests reveal hyperammonemia and elevated urinary orotic acid levels.

Which step of the urea cycle is most likely defective?

A. Formation of carbamoyl phosphate
B. Conversion of ornithine to citrulline
C. Conversion of citrulline to argininosuccinate
D. Conversion of argininosuccinate to arginine
E. Conversion of arginine to urea

A neonate presents with severe hyperammonemia. Urinary orotic acid levels are found to be within the normal range.

Which of the following enzyme deficiencies best explains these findings?

A. Ornithine transcarbamylase
B. Carbamoyl phosphate synthetase I
C. Argininosuccinate synthetase
D. Argininosuccinate lyase
E. Arginase

A child presents with developmental delay, episodic vomiting, and brittle hair. Laboratory analysis shows elevated plasma ammonia and increased levels of argininosuccinic acid in blood and urine.

Which enzyme is deficient in this disorder?

A. Arginase
B. Carbamoyl phosphate synthetase I
C. Ornithine transcarbamylase
D. Argininosuccinate synthetase
E. Argininosuccinate lyase

A neonate presents with lethargy and seizures. Plasma amino acid analysis reveals markedly elevated citrulline levels along with hyperammonemia.

Which enzyme deficiency is the most likely diagnosis?

A. Carbamoyl phosphate synthetase I
B. Ornithine transcarbamylase
C. Argininosuccinate synthetase
D. Argininosuccinate lyase
E. Arginase

A 7-year-old child presents with progressive spasticity and mild hyperammonemia. Plasma arginine levels are markedly elevated.

Which of the following enzymes is deficient?

A. Ornithine transcarbamylase
B. Carbamoyl phosphate synthetase I
C. Argininosuccinate synthetase
D. Argininosuccinate lyase
E. Arginase

A patient with a urea cycle disorder develops cerebral edema. Which biochemical mechanism best explains the neurotoxicity caused by elevated ammonia levels?

A. Increased ATP generation
B. Increased acetylcholine synthesis
C. Depletion of α-ketoglutarate
D. Inhibition of glycolysis
E. Increased oxidative phosphorylation

A child diagnosed with ornithine transcarbamylase deficiency is treated with sodium benzoate.

What is the primary mechanism by which this therapy reduces plasma ammonia levels?

A. Activation of carbamoyl phosphate synthetase I
B. Direct conversion of ammonia to urea
C. Increased renal excretion of free ammonia
D. Formation of nitrogen-containing conjugates
E. Inhibition of intestinal ammonia production

A neonate presents with hyperammonemia and normal urinary orotic acid levels. Enzyme assays show normal carbamoyl phosphate synthetase I activity.

Deficiency of which compound is most likely responsible for this condition?

A. Arginine
B. Citrulline
C. N-acetylglutamate
D. Ornithine
E. Carbamoyl phosphate

Patients with acute hyperammonemia commonly present with respiratory alkalosis.

What is the most likely explanation for this finding?

A. Renal loss of hydrogen ions
B. Metabolic acidosis compensation
C. Direct stimulation of the respiratory center
D. Increased bicarbonate production
E. Increased lactate accumulation