PATIENT SCENARIO AND QUESTIONS

PATIENT SCENARIO:

- A male infant, initially asymptomatic after birth, deteriorated after starting feeding. He was brought to the emergency department at 3 days of age due to vomiting, failure to thrive, and altered consciousness.
- There is a history of consanguinity between the parents. The infant was born at term, weighing 3,300 grams.
- The family's first child died at the age of 14 days in another hospital, where he was investigated and treated for vomiting, weakness and seizures.
- The patient appears weak and prone to sleep during examination. No other pathological findings are noted.

QUESTION 1. Which investigations should be performed initially for the patient?

I. Complete blood count II. Biochemistry III. Acute phase reactants IV. Blood gas V. Blood sugar VI. Blood ketones VII. Ferritin VIII. Transglutaminase Ig A IX. Ammonia X. Viral hepatitis serology panel

A) I-II-III-IV-V-VI-IX
B) I-II-III-IV-V-VI-X
C) I-II-III-IV-V-VIII-X
D) I-II-III-IV-V-VII-VIII
E) II-III-IV-V-VII-VIII

QUESTION 2. The patient has respiratory alkalosis, hyperammonaemia (ammonia: 392 μmol/L), and hypoglycaemia. Acute phase reactants are negative. What should be the initial treatment approach in the emergency department?

A) The baby should be encouraged to breastfeed rapidly if hypoglycaemic, or if unable to feed, formula should be given.
B) Start the patient on 1/2 maintenance fluid (half 5% dextrose, half normal saline).
C) Start urgent total parenteral nutrition containing protein and carbohydrates by inserting an emergency umbilical catheter.
D) Rapidly cease the patient's protein intake, such as breast milk or formula, and initiate intravenous fluids with high glucose content (glucose infusion rate: 8-10 mg/kg/min).
E) Provide rapid lipid infusion to meet the patient's calorie requirement.

QUESTION 3. During follow-up, the patient's blood sugar is measured at 340 mg/dl. What would you do?

A) Immediately discontinue fluids and start normal saline.
B) Do not change the fluid, start insulin.
C) Make no changes in treatment.
D) Reduce the glucose infusion rate by half.
E) Double the volume of the fluid.

QUESTION 4. What is the possible preliminary diagnosis for the patient, and what should be the second-line treatments?

A) Organic acidaemia - Hydroxocobalamin and biotin
B) Fatty acid oxidation defects - Dietary therapy
C) Urea cycle disorder - Sodium benzoate, Sodium phenylacetate
D) Mitochondrial disease - Coenzyme Q, Riboflavin, Carnitine
E) Lysosomal disease - Substrate reduction therapy

QUESTION 5. Please provide medication orders for the patient (mark only the indicated medications and place orders)

□ Na benzoate: ..... mg/kg/dose bolus loading, then ..... mg/kg/day maintenance
□ Sodium phenylbutyrate/Na phenylacetate: ..... mg/kg/dose bolus loading, then ..... mg/kg/day maintenance
□ L-arginine: ..... mg/kg/dose bolus loading, then ..... mg/kg/day maintenance
□ Hydroxocobalamin: ..... mcg/dose
□ Biotin: ..... mg/day
□ Carglumic acid: ..... mg/kg/dose bolus loading, then ..... mg/kg/day maintenance
□ Thiamine: ..... mg/day

QUESTION 6. Despite initiating the ammonia-lowering therapies, the ammonia level continues to rise in the patient (ammonia: 680 μmol/L). What treatment is necessary at this stage?

A) Increase the dose of ammonia-lowering therapies.
B) Increase the glucose infusion rate.
C) Start the patient on a high-glucose-content oral diet.
D) Transition to extracorporeal detoxification methods (peritoneal dialysis, haemodialysis).
E) Initiate formula feeding specific to the diagnosis.

QUESTION 7. What are the priority metabolic tests to be requested for diagnosis?

A) Blood amino acids, urine organic acids, carnitine-acetylcarnitine analysis
B) Carnitine-acetylcarnitine analysis, homocysteine, vitamin B12
C) Very long-chain fatty acids, blood amino acids, urine organic acids
D) Reducing substances in urine, urine sugar chromatography, and urine organic acids
E) Urine organic acids, reducing substances in urine, homocysteine

QUESTION 8. What are your recommendations for treatment follow-up after the patient's general condition has improved and the diagnosis has been confirmed?

A) Initiate breastfeeding.
B) Start only ammonia-lowering medications.
C) Initiate only a protein-restricted diet.
D) Plan treatment only for attacks.
E) Plan ammonia-lowering treatment and a protein-restricted diet for the patient.

QUESTION 9. Which tests should be planned when patients come for follow-up?

I. Complete blood count II. Biochemistry III. Ammonia IV. Blood amino acids V. Urine organic acids VI. Nutritional parameters VII. Immunoglobulins VIII. Protein C and S IX. Homocysteine

A) II-III-IV-VII-IX
B) I-II-III-IV-IX
C) I-II-III-IV-VII
D) I-II-III-VII-IX
E) I-II-III-IV-VI

QUESTION 10. The patient's mother mentions a desire to become pregnant again in the future. How would you provide information to the family? (Write)

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