

Pearson syndrome associated with hemophagocytic syndrome in a child

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A seven-month-old boy was admitted to a local health center with a two-week history of fever, cough, vomiting, and diarrhea. The patient was the first child of healthy, unrelated parents. Pancytopenia, increased activated partial thromboplastin time (aPTT) and international normalized ratio (INR), and acidosis were detected during local health care evaluation, and the patient was referred to our hospital with the diagnosis of metabolic disorder. Physical examination revealed toxic appearance, lethargy, paleness, and hepatomegaly palpable 4 cm below the right costal margin. The results of the laboratory examinations were as follows: Hematological studies showed hemoglobin 7.4 g/dl, platelet 93x10⁹/L, white blood cells 7.4x10⁹/L with a differential count of 21% neutrophils, 70% lymphocytes, 9% monocytes, mean corpuscular volume

(MCV) 91.3 fl, and red cell distribution width (RDW) 14.7. Peripheral blood smears showed burr cell, schistocyte, and acanthocyte. Serum alanine aminotransferase was 237 U/L (5-40 U/L), aspartate aminotransferase 506 U/L (8-33 U/L), total bilirubin 1.46 mg/dl (0.10-1.20 mg/dl), conjugated bilirubin 1.04 mg/dl (0-0.30 mg/dl), blood urea nitrogen 2.0 mg/dl (5-18 mg/dl), creatinine 0.28 mg/dl (0.6-1.2 mg/dl), sodium 127 mEq/L (138-145 mEq/L), potassium 3.21 mEq/L (3.4-4.7 mEq/L), chloride 95 mEq/L (95-110 mEq/L), calcium 13.2 mg/dl (8.6-10.2 mg/dl), phosphorus 2.0 mg/dl (2.3-4.7 mg/dl), blood pH 7.282 (7.35-7.45), bicarbonate 7 mmol/L (21-28 mmol/L), lactic acid 69.4 mg/dl (10-14 mg/dl), and pyruvic acid 2.18 mg/dl (0.5-1.0 mg/dl). Bone marrow aspiration smear showed vacuolization of hematopoietic pre-

cursor (especially erythroblasts) and hemophagocytosis. Urine organic acid test determined findings consistent with Pearson syndrome. To confirm the diagnosis, mitochondrial DNA (mtDNA) from peripheral blood leukocytes of the patient was investigated. The patient showed a specific deletion in a population of mtDNA. After initial evaluation, intravenous (i.v.) fluid, fresh frozen plasma, mitochondrial cocktail, and antibacterial treatment including meropenem, amikacin, and teicoplanin were started, and peritoneal dialysis was performed for resistant acidosis. During the follow-up, mechanical ventilation was instituted due to respiratory depression. The patient died two days after admission to our hospital.

Pearson syndrome, first described in 1979 [1], is a pro-gressive multiorgan disorder that involves the hematopoietic system, exocrine pancreas, liver, and kidneys, and often presents clinically with failure to thrive caused by deletions in mtDNA [2,3]. Impairment of the mitochondrial respiratory chain may lead to lactic acidemia, high lactate/pyruvate molar ratios in plasma, and even fatal metabolic acidosis [2,3]. The disease starts during infancy and most children diagnosed with Pearson syndrome die before the age of three years [2,3].

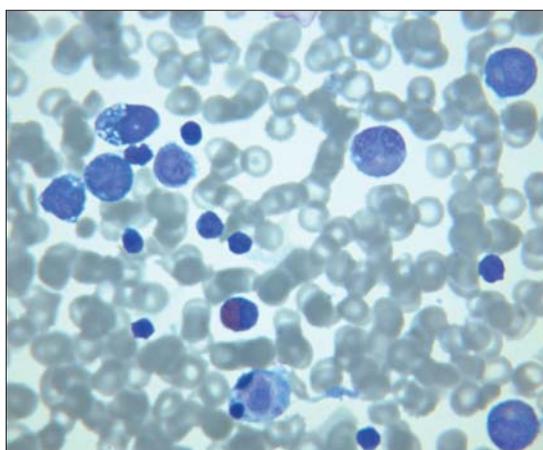


Figure 1. Bone marrow aspiration smear shows vacuolization of hematopoietic precursors and hemophagocytic histiocytes

Hemophagocytic lymphohistiocytosis (HLH) is a life-threatening condition of severe hyperinflammation caused by the uncontrolled proliferation of activated lymphocytes and histiocytes secreting high amounts of inflammatory cytokines [4]. Cardinal signs and symptoms are prolonged fever, hepatosplenomegaly and pancytopenia [4]. Characteristic biochemical markers include elevated triglycerides, ferritin and low fibrinogen [4]. Two forms of hemophagocytic syndrome (HPS), primary and secondary, have been reported [4,5]. Secondary HPS has been reported in association with many different conditions [4,5,6].

Pearson syndrome and HPS are multisystemic disorders and cytopenias are a component of both diseases.

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