Case Report

The Essential History of a Patient with Pearson Marrow, a Case Report

Fatemeh Malek1*, Parastoo Tavana1, Masoumeh Mohkam2

1Department of Pediatrics, Division of Pediatric Oncology, Shahid Beheshti University of Medical Sciences, Tehran, Iran.
2Pediatric Nephrology Research Center, Research Institute for Children's Health, Shahid Beheshti University of Medical Sciences, Tehran, Iran.

*Corresponding Author
Dr. Fatemeh Malek,
Email: fmalek@sbmu.ac.ir

Abstract
Pearson syndrome is a rare mitochondrial disorder confirmed by mt-DNA deletion which typically occurs in the first two years of life. That is to say children are at high fatal risk, most infants are marked with some common features especially anemia and pancreatitis, which results in death in early childhood. A 6-month-old Iranian female infant was presented with macrocytic anemia, required packed red blood cell transfusions. She also was affected by exocrine pancreatic dysfunction, in which she underwent Creon treatment. By first year of age she had experienced some severe metabolic crises intermittently. After hospitalized for some months she was expired unfortunately. In conclusion, Pearson syndrome, as a rare disease affects many organs, such as liver, kidney, pancreas, bone marrow, which led to anemia, failure to thrive, and multi organ failure. In such cases, a physician must consider and evaluate all possible damages, especially anemia and pancreatitis. We present a case of Pearson syndrome with anemia

Keywords: Mitochondrial disorders; Pearson syndrome; Pancreatitis; Acidosis; Infants.

Introduction
Pearson syndrome (PS) is a mitochondrial cytopathic condition, foremost defined by Dr. Pearson and his colleagues in 1979. Pearson's disease is a syndrome of refractory sideroblastic anemia in childhood with vacuolization of bone marrow precursors and exocrine pancreatic dysfunction (1-3). The frequency of PS is indefinite, with only about 100 patients termed in the literature since reported by Pearson. Severe transfusion-dependent, macrocytic anemia begins in early infancy and dissociated with some variable degrees (4-5). PS affects marrow and pancreas, by deletions in mitochondrial DNA (mtDNA), which results in metabolic acidosis and adjustable tissue dysfunction in patients. Deleted mtDNA in cells occurs in variable proportions comparative to normal mtDNA, and a mixture labeled heteroplasmy (6-8). Variations in heteroplasmy are assumed to cause alterations in disease demonstrations and progression in patients.

Case report
A six-month female infant of non-cousin parents, was referred to the department of oncology, Mofid children's hospital. Severe pallor, without organomegaly was found in physical exam at the time of presentation. Initial workups revealed macrocytic anemia and mild Hyperlactatemia. Physical and psychomotor growth were in the normal range and were appropriate for her age. The result of her laboratory tests are as follow.


HB electrophoresis showed 2% HB A2. She underwent a Bone marrow aspiration, which revealed the vacuolization of erythroid and myeloid precursors (figure 1). Moreover, no clinical signs of neuromuscular dysfunction were found.
No viral pathogens or infections, such as parvovirus B19, Epstein-Barr virus, or CMV were detected. The bone marrow sample was stained with Prussian blue technique was also in favor of ring sideroblasts, which rechecked for confirmation. The molecular analysis which was performed to confirm the diagnosis of PS, detected mitochondrial deletions.

**Discussion**

Pearson syndrome (PS) is a mitochondrial cytopathic disorder, which was defined by Dr. Pearson and his colleagues in 1979 (1-3). PS as an inexplicable syndrome presents with sideroblastic anemia, neutropenia, thrombocytopenia, and vacuolization of marrow precursors. Since it is a multisystem disease, pancreatic insufficiency, proximal renal tubular acidosis, and metabolic acidosis are common findings. As mentioned, single mitochondrial DNA deletions occur in different cells in a wide spectrum which result in inconsistency in clinical phenotype, called heteroplasmy (6-7). No specific treatment is available for patients with PS, so alertness of possible complications and early interventions may minimize PS-associated morbidity and mortality. Red blood cell transfusions are frequently needed to manage macrocytic anemia (4-8). Pancreatic enzyme replacement necessitates for patients with malabsorption because of their exocrine pancreatic dysfunction. Metabolic crises can be managed by hydration, correction of electrolyte abnormalities, and acidosis (6-8). In the differential diagnosis of congenital anemia PS syndrome has been missed for a long time (8). Hence, one must be cautious about the hallmarks of the syndrome.

**Conclusion**

No specific treatment is currently available for patients with PS. Because of clinical difficulty in diagnosis of PS, to consider and to expect the disease in advance is of excessive clinical importance (8). So, for patients with exocrine pancreatic dysfunction and mal absorption the following medicines are recommended; Pancreatic enzyme replacement such as Creon and fat-soluble vitamins.

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**Conflict of Interest**

The authors declare no conflicts of interest.

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