

Sea Blue Histiocytosis Secondary to Niemann Pick Disease

Mobina Ahsan Dodhy, Wardah Aslam, Amna Nazir, Masooma Shaheen,
Maryam Habib, Humaira Zafar

Department of Pathology, Benazir Bhutto Hospital, Rawalpindi

Abstract: Sea blue histiocytosis is a rare disorder, characterized by the presence of atypical macrophages (sea blue histiocytes) in the bonemarrow detectable by May-Giemsa staining. We report a case of a 3 years old female ,whose bone marrow examination revealed numerous sea blue histiocytes and Niemann-Pick cells (storage cells) .Niemann-Pick disease is the most common cause of secondary sea-blue histiocytosis.

Keywords: Sea-blue-histiocytosis, Niemann Pick disease

Introduction

Sea blue histiocytosis is a rare disorder characterized by the presence of atypical macrophages in the bone marrow, liver or spleen. It is categorized as primary and secondary.¹ Primary sea blue histiocytosis syndrome has no known etiology and characterized by a triad of splenomegaly, thrombocytopenia and accumulation of sea blue histiocytes in bone marrow, liver and spleen.² Secondary causes include Chronic myeloid leukemia (CML), myeloproliferative disorder, myelodysplastic syndrome, Idiopathic thrombocytopenic purpura (ITP), long term parenteral nutrition with fat emulsion and storage disorder like Gauchers and Niemann Pick disease.³ We report a case of sea blue histiocytosis secondary to Niemann Pick disease diagnosed on bone marrow aspiration in a 3 year old girl.

Case Report

A 3 year old female presented to pediatrics department on 18 January 2011 with progressive pallor for 1 month and cough for last 2 days before admission. There was no history of fever, bleeding or CNS symptoms. Her parents had nonconsanguous marriage. She was developmentally normal. Rest of the siblings were alive and healthy. She was transfused 3 pints of blood after admission.

On examination she was sick looking pale child with petechiae on abdomen and back. Laboratory investigation revealed pancytopenia with Hb 4.5g/dl, WBC

$3.6 \times 10^3/\mu\text{l}$.and platelet count $10 \times 10^3/\mu\text{l}$. Peripheral blood film revealed dimorphic blood picture and reticulocyte count 0.1%. The differential diagnosis included nutritional anaemia, leukemia, storage disorder. WBC Haemoglobin electrophoresis showed normal study. Ultrasound abdomen was normal. Chest x ray did not reveal any abnormal finding.

The bone marrow aspiration was done from right posterior superior iliac spine (PSIS). Smears were made and stained with giemsa, pearl stain and PAS stain. Bone marrow aspirate was hypercellular. Histiocytes were markedly increased showing sea blue colored cytoplasm with multiple tiny vacuoles. Niemann Pick cells were present. Iron was increased. The Histiocytes were positive for pearl stain. The diagnosis of sea blue histiocytosis secondary to Niemann Pick disease was made. Sphingomyelinase levels were suggested. She was given supported treatment but unfortunately after 2 days of diagnosis she succumbed to her disease.

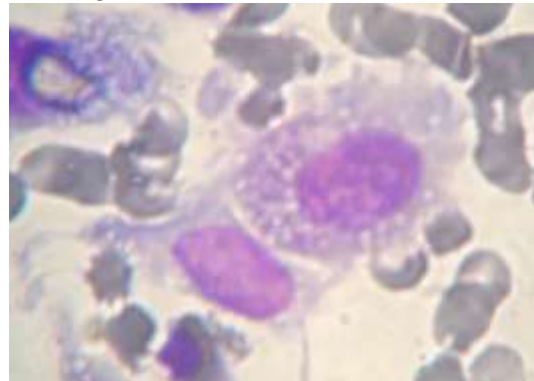


Fig 1: Bone marrow aspirate: Sea Blue Histiocyte (arrow) (May-Grnwald-Giemsa x 1000)

Correspondence: Mobina Ahsan Dodhy

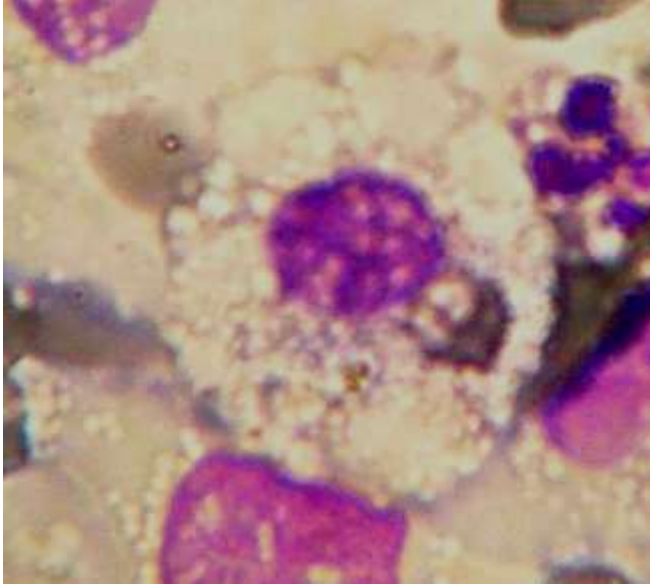


Fig 2: Bone marrow aspirate: Niemann-Pick cell (May-Grunwald-Giemsa x 1000)

Discussion

Sea blue histiocytosis was first described by Moschlin in 1947, characterized by the presence of atypical macrophages in bone marrow, liver or spleen. It is classified into primary and secondary.¹ Primary sea blue histiocyte syndrome is a rare disorder characterized by splenomegaly, thrombocytopenia and accumulation of sea blue histiocyte in multiple organs with no known etiology.² Secondary causes of sea blue histiocytosis include diseases in which there is increased haemopoietic cellular turnover like CML, myeloproliferative disorder, myelodysplastic syndrome, severe

autoimmune neutropenia, ITP other causes include long term parenteral nutrition including fat emulsion sources, storage disease and other lysosomal disorders like Niemann Pick, Gaucher disease.^{3,4,5,9}

The sea blue histiocytes are atypical macrophages having large cytoplasm, empty vacuoles and sea blue granules containing lipofusion or ceroid. The nuclei are centrally located or eccentric. They are seen in bone marrow aspirate stained with May Giemsa stain.^{3,10}

Niemann pick is a rare inherited autosomal recessive disorder. There is deficiency of sphingomyelinase resulting in accumulation of sphingomyelin in the liver, spleen, lungs, bone marrow or brain. Prognosis of the disease depends upon the organ involved. According to Crocker classification five clinical forms are de-

finied.⁶

The subtype A is a severe neurodegenerative disorder resulting from sphingomyelinase deficiency characterized by progressive psychomotor retardation, hepatosplenomegaly and early death.⁷ In type C disease there is defect in cholesterol esterification.⁸

The subtype B is characterized by splenomegaly, thrombocytopenia and sea blue histiocytes in the bone marrow. The nervous system is not involved. Rarely liver and lungs are involved. The diagnosis of Niemann pick disease should be confirmed by liver or bone marrow biopsy or sphingomyelinase activity. Prognosis of type B is variable, some patients may survive to sixth decade of life.^{6,11}

The case reported here corresponds to Niemann pick disease with secondary sea blue histiocytosis. Niemann pick disease is a cause of sea blue histiocytosis and should be considered in patients with splenomegaly and thrombocytopenia¹².

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