A rare case of sea-blue histiocytosis associated with Niemann-Pick disease type B in a 8-year and 9-month old boy with hypertension

Authors *MA Hua-mei, SU Zhe, LI Yan-hong*

Hospital The First Affiliated Hospital of Sun Yat-sen University, Guangzhou 510080, China.

OBJECTIVES

Background—Sea-blue histiocytosis is a morphological finding that can be associated both with acquired conditions of increased cellular turnover and inborn errors of lipid metabolism.

Objective—To present a Chinese boy of hypertension and sea-blue histiocytosis secondary to Niemann-Pick disease type B.

METHODS

Diagnosis was confirmed by the bone marrow aspiration and the specific enzyme assay of leukocytes (deficiency in sphingomyelinase activity).

RESULTS

The 8-year and 9-month old boy was the first child of non-consanguineous parents of Chinese Han ethnicity, who presented with hepatosplenomegaly for 4 years. General physical examination showed short stature with Ht 106 cm(-5SD) and abdominal distention. On systemic examination, hypertension with BP 158/119 mmHg was noted, and hepatosplenomegaly measuring 10cm below costal margins, respectively was present. Neurological examination and respiratory function tests were normal.Chest X-rays revealed diffuse reticular pattern. The echocardiography showed a thickening(8mm) of the interventricular septum. Ultrasonography showed a bright liver, which is usually fatty tissue. Coagulation function tests showed slightly prolonged activated partial thromboplastin time (APTT), lipid profile showed elevated LDL(3.57mmol/L), elevated TG(2.37mmol/L) and decreased HDL(0.54mmol/L). Other admission laboratory data include following: white blood cell count, hemoglolin, platelet count, serum total bilirubin, aspartate aminotransferase, alanine aminotransferase, serum albumin, were all within normal ranges. Hematoxylin-eosin staining of bone marrow showed scattered foci of foamy histiocytes. May-Giemsa staining of the bone marrow smear showed multiple blue-colored granules were found in the cytoplasm of histiocytes. Histiocytes were stained blue by the Schmorl reaction. The acidic sphingomyelinase activity seen in peripheral blood leukocytes was lower (4.75 nmol/mg/h) than normal (40.29 nmol/mg/h), confirming a diagnosis of seablue histiocytosis secondary to Niemann-Pick disease.

CONCLUSIONS

We report a rare case of sea-blue histiocytosis associated with Niemann-Pick disease type B in a 8-year and 9-month old boy with hypertension.

REFERENCES:

1.Suzuki O, Abe M. Secondary Sea-blue Histiocytosis Derived from Niemann-Pick Disease. J Clin Exp Hematop. 2007;47(1):19-21.):19-21.

2.Castañón Martínez R1, Fernández-Velilla Peña M, González Montaño MV, et al. Lung affectation in an adult patient with Niemann-Pick disease, type B. Arch Bronconeumol. 2012 ;48(6):213-215.

3.Candoni A1, Grimaz S, Doretto P, et al. Sea-blue histiocytosis secondary to Niemann-Pick disease type B: a case report. Ann Hematol. 2001;80(10):620-622.