

Successful Pregnancy in a Patient with Familial Sea-Blue Histiocyte Syndrome

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Summary

A 27-year-old Indian woman at 23 weeks' gestation presented with decompensated liver cirrhosis, coagulopathy, restrictive lung disease with cor pulmonale and preeclampsia. She was diagnosed to have sea-blue histiocyte syndrome (SBHS) at the age of 13 years and was treated conservatively. There was worsening liver, respiratory and bone marrow function as the pregnancy progressed. She underwent a successful pregnancy despite her poor medical condition and advanced disease state. We described the first case of familial SBHS in a pregnant patient from Asia.

Key Words: Sea-blue histiocyte, Cirrhosis, Hypersplenism, Restrictive lung disease, Lipidosis

Introduction

The syndrome of sea-blue histiocyte is a rare disorder first described in 1970¹ consisting of splenomegaly in association with numerous histiocytes which stained a sea-blue color with Romanowsky's stain in the bone marrow. Since then 68 cases have been reported in the literature and all patients are from the Western region except for 2 cases from Japan². The occurrence of the syndrome among family members is uncommon with less than 10 families being described³. The following describes the first case of sea-blue histiocyte syndrome (SBHS) in two Indian siblings. One of them underwent a successful pregnancy. Histiocytic involvement of the lungs, spleen, liver and bone marrow was evident in both patients.

Case report

A 27-year-old woman presented with worsening of effort intolerance and gum bleeding during the 23rd

week of her second pregnancy. She was diagnosed to have SBHS from the bone marrow aspirate at the age of 13 when she presented with empyema of the right lung that required an open drainage. She had repeated hospitalization for septicaemia. SBHS was diagnosed in the younger brother at the age of 11 when a thoracotomy was performed for non-resolving empyema. He was lost to follow-up and presented in November 1999 with decompensated liver and lung disease. The two siblings were born in Malaysia. The parents are first cousins and they are immigrants from the northern region of India. The father died of cancer of the spine and the mother who is in her fifties is well and alive. The other three siblings aged between 20 and 25 years are well and one of them has 2 healthy children. The patient is not related to her husband. Abdominal examination revealed a gravid uterus, gross splenomegaly and ascites. The fetal heart was audible. The haemoglobin was 10.3 g/dl; leucocyte count $3.2 \times 10^9/l$ and platelet count $58 \times 10^9/l$. The coagulation and liver profiles were impaired. A cirrhotic liver with portal hypertension and patent

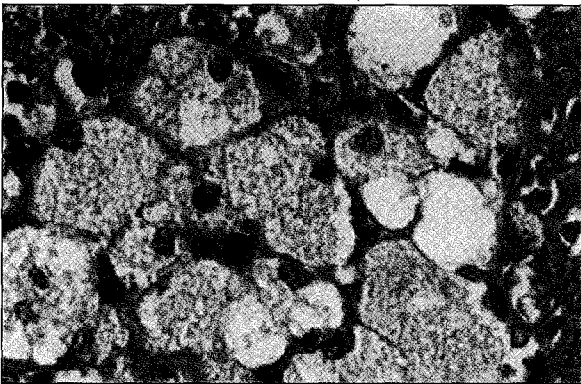


Fig. 1: Section of the spleen showing numerous large, foamy histiocytes with vacuolated and granular cytoplasm. The cytoplasm stains pink with haematoxylin and eosin.

portal, hepatic and splenic veins were evident on ultrasound. Advanced infiltrative and restrictive lung disease was confirmed by radiological and functional lung studies. Echocardiogram showed severe pulmonary hypertension and dilated right-sided chambers. Symptomatic treatment was instituted and the patient was planned for early delivery. Caesarian section was carried out at 30 weeks gestation because she developed worsening of liver and pulmonary function with superimposed pre eclampsia. A healthy baby boy was delivered and the patient was discharged well. Six months later, splenectomy was performed for painful and increasing splenomegaly. The spleen weighed 1432 g. The normal splenic architecture was lost and both the red and white pulps are diffusely infiltrated by large, foamy histiocytes (Fig. 1). Positive staining with oil red O confirmed the lipid nature of the granules. The histiocytes have not demonstrated the uniformly vacuolated appearance of Niemann-Pick cells, or the filamentous structure of Gaucher's cells. Extramedullary haemopoiesis was also evident. During the last follow-up review 15 years after diagnosis, there was no progression of the disease and her 2-year-old son was well with no evidence of similar disease.

Discussion

SBHS is a congenital disorder, which is thought to be autosomal recessive. The mechanism of blue granule storage is unknown. The storage of specific phosphosphingolipids and glycosphingolipids appeared to be the major element of this disorder. Bone marrow infiltration by SBH is the basis for the diagnosis. Other conditions that show sea-blue histiocytosis such as adult lipidosis, chronic granulocytic leukaemia were excluded in the present case. Clinical events appear to be predictable based on the degree of histiocytic involvement of the organs. The clinical course is usually benign but 15% of cases develop fatal liver failure¹. In general, the younger the patient at time of diagnosis, the poorer the prognosis. Despite the extensive organ infiltration with histiocytes, our patient successfully conceived and gave birth to a healthy baby boy.

The genetic ramifications of the primary syndrome are yet to be determined. Jones et al¹ described the syndrome in 2 siblings. Zlotnick and Fried³ have reported a father and mother, who were first cousins and their 2 children who had the syndrome. In view of the hereditary basis for this disease along with the potentially fatal complications, genetic counseling and close monitoring of the patient's offspring is essential. However, the syndrome itself appears not an absolute contraindication for pregnancy as observed in our case. The high prevalence of the syndrome in the Caribbean region is interesting and unexplained. The reason for the low number of reports among Asian patients is thought to be either a low frequency of this gene abnormality or it has been overlooked, as this disease is not generally known in this part of the world. As such, the syndrome may be more common than is currently recognised and should be considered in the differential diagnosis of young patients presenting with pulmonary infiltrates, liver cirrhosis and hypersplenism and a positive family history. To the best of our knowledge, this case represents the first description of familial SBHS in a pregnant patient from Asia.

CASE REPORT

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