



Waldmann's Disease: A Rare Form of Protein Losing Enteropathy

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ABSTRACT

Waldmann's disease or primary intestinal lymphangiectasia is a rare disorder characterised by loss of protein from gastrointestinal tract. We present a patient with left upper limb hemi hypertrophy and chylous ascites associated with lymphopenia and hypoalbuminemia. Diagnosis was made by upper gastrointestinal endoscopy and biopsy. Patient responded to nutritional therapy very well. Primary intestinal lymphangiectasia should be considered in patients with chylous ascites, hypoalbuminemia and lymphopenia.

Keywords: *Waldmann's disease, primary intestinal lymphangiectasia, protein losing enteropathy.*

INTRODUCTION

Waldmann's disease or primary intestinal lymphangiectasia is a rare form of protein losing enteropathy characterised by dilatation of lymphatics of mucosa, submucosa, serosa and mesentery of bowel resulting in secondary lymphatic leakage. The condition can present through a broad spectrum of clinical manifestations depending on the anatomical location and extent of the lymphatic anomaly.^[1] Here we report the case of a child with intestinal lymphangiectasia presenting with chylous ascites and hemi hypertrophy of left upper limb.

CASE REPORT

A 2-year-old girl was admitted with generalised weakness for last 3 months and huge ascites for last 2 weeks. The child had no history of fever or any other extra-gastrointestinal symptoms. There was no significant past or family history.

On admission the patient was found to be orthopneic due to huge abdominal distension. There was no jaundice or any other sign of hepatic failure. There was no hepato-splenomegaly but tense ascites was present. Left upper limb hemi hypertrophy was noted which involved forearm and hand. Anthropometry and nutritional assessment showed height 0.80 m (below 10th percentile) and weight 10 kg (below 5th percentile).

Investigations revealed Hb 9.7 gm% with normocytic normochromic anemia in peripheral blood smear. Total WBC count was: $6 \times 10^9/L$ with 13% lymphocytes. Absolute lymphocyte count was $0.78 \times 10^9/L$. Serum total protein was 3.1 g/dL with albumin 1.8 g/dL and globulin 1.3 g/dL. Ultrasonogram of whole abdomen showed huge ascites with a few mesenteric lymph nodes, maximum size being 4mm. Routine urine examination showed 0-2 pus cells/HPF and trace levels of albumin. Urine protein: creatinine ratio was 0.2. Kidney and

liver function tests , serum electrolytes , ultrasonogram with Doppler study of portal venous system and echocardiography were normal . Markers for sepsis were negative. Abdominal paracentesis revealed chylous fluid with cholesterol 12.2 mg/dL, triglyceride 246.9 mg/dL, protein 4 g/dL and albumin 1 g/dL. We suspected some form of protein losing enteropathy and upper gastrointestinal endoscopy revealed diffuse white specs suggestive of intestinal lymphangiectasia over the 2nd part of duodenal mucosa. Histopathological examination of the duodenal biopsy specimen showed mucosa with focal blunting and broadening of villi and many dilated lymphatics in the lamina propria with mild lymphoplasmacytic infiltrates [See Figure 1]. There was no evidence of malignant cells or any parasitic infestation .

The diagnosis of primary intestinal lymphangiectasia was clinched. Intravenous human albumin was infused to correct hypoalbuminemia and child was put on medium chain triglyceride (MCT) based diet . Patient responded to therapy very well and ascites decreased after 2 weeks of therapy . Serum albumin level was found to be 2.6 g/dL at 2 weeks follow up . Child is currently doing well and is on regular follow up.

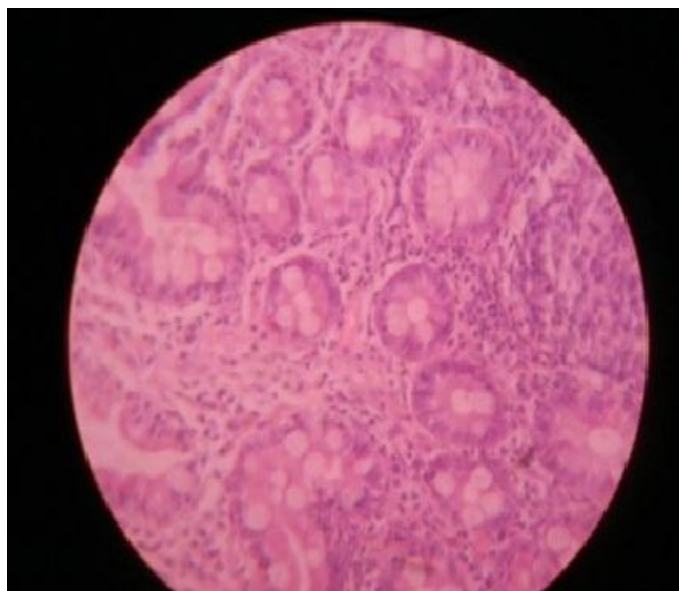


Figure 1: Histopathological picture of upper gastrointestinal biopsy specimen showing marked dilatation of numerous lymphatic channels indicative of primary intestinal lymphangiectasia.

DISCUSSION

In 1961, Waldmann et al. Described first 18 cases of idiopathic hypercatabolic hypoproteinemia.^[2] These patients had hypoproteinemia and edema with low serum albumin and immunoglobulin levels. These authors proposed the term intestinal lymphangiectasia.^[3]

Children with primary intestinal lymphangiectasia are usually diagnosed before the age of 3 years.^[1] In 95% of cases, the principal clinical feature is edema. Patients may exhibit fatigue, abdominal pain, nausea, vomiting and failure to thrive. The main digestive symptom is moderate and intermittent diarrhea.^[1]

Several syndromes including Yellow Nail syndrome, Von Recklinghausen Disease, Turner syndrome, Noonan syndrome, Klippel-Trenaunay Syndrome and Henekam syndrome have been described.^[1]

Intestinal lymphangiectasia is responsible for lymphatic leakage into the intestinal lumen leading to hypoalbuminemia, edema and lymphopenia. Also there may be hypogammaglobulinemia with low levels of IgG, IgM and IgA, low counts of CD4 and T cells.^[6] However, the diagnosis is confirmed when endoscopic findings with histologic confirmation of an intestinal biopsy show presence of intestinal lymphangiectasia.^[1] Macroscopic characteristics observed in upper digestive endoscopy consist of prominent folds (Kerckring folds) with whitish appearing villi, small white dots that are not eliminated by endoscopic wash and “cotton ball” imaging which is sometimes similar to those produced by grease build up.^[4]

The cornerstone of treatment is a reduction of long chain fatty acids in the diet combined with the addition of medium chain triglycerides. This prevents the swelling and rupture of malformed lymphatics.

For nonresponsive cases other treatment options are total parenteral nutrition, corticosteroids, octreotide, tranexamic acid etc. Supportive therapy may require infusion of albumin, diuretics, thoracentesis or paracentesis for relief of symptoms.

The long-term prognoses of patients with primary intestinal lymphangiectasia vary, but this is usually a chronic disease which slowly progresses albeit with intermittent clinical remission. It requires a continuous restrictive

diet.^[5] Poor quality of life may be due to infections or lymphedema.

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