

A Case of Primary Intestinal Lymphangiectasia

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ABSTRACT

Primary intestinal lymphangiectasia (PIL) is an uncommon disorder and important cause of protein losing enteropathy due to congenital malformation or obstruction of intestinal lymphatic drainage. No accurate serological or radiological tests available for the diagnosis of intestinal lymphangiectasia. Endoscopy and biopsy are currently the only diagnostic modality with certainty. Hence histopathological examination is the gold standard for the diagnosis of intestinal lymphangiectasia which shows dilated intestinal lymphatics with broadened villi of the small bowel. Herein we report a case of intestinal lymphangiectasia in a 4-year-male child presented with chronic diarrhea and edema. Patient was treated by resection of small intestine and postoperatively patient showed improvement with small bowel movements and disappearance of edema.

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Introduction

Primary intestinal lymphangiectasia (PIL) is a relatively rare disorder and main cause of protein losing enteropathy. It is caused by congenital malformation or secondary obstruction of intestinal lymphatic drainage and characterized by diffuse or marked dilation of the enteric lymphatics.^[1] Intestinal lymphangiectasia is manifested by an extreme loss of proteins into gastrointestinal tract (GIT) resulting in diarrhea, edema, hypoalbuminemia, hypogammaglobulinemia, sometimes pleural effusion and ascites. In 1961, Waldmann and Schwabb reported this rare entity of intestinal lymphangiectasia.^[1]

It can be primary or secondary type. Primary intestinal lymphangiectasia (PIL) usually occurs in children and adolescents resulting from a congenital deformity of the small bowel lymphatic system. Secondary intestinal lymphangiectasia is more often seen in adults and occurs due to an elevated lymphatic pressure as in lymphoma, systemic lupus erythematosus, sarcoidosis, scleroderma, inflammatory bowel disease, malignancies, constrictive pericarditis and cardiac surgery. ^[2,3]The diagnosis is based on the upper gastrointestinal endoscopy and histopathological examination which are showing diffuse scattered mucosal white lesions with typical histological findings of abnormal lymphatic dilation.

Herein we report a case of primary intestinal lymphangiectasia (PIL) in a 4-year-male child who presented with chronic diarrhea and edema.

Case History

A 4-year-male child presented with history of diarrhea with upper and lower limb edema since 5-6 months. He also complained of abdominal distension and weight loss since 2-3 months. He had 4-5 watery stools per day and mixed with mucus. There was no history of fever, vomiting, abdominal pain, skin rash or urinary symptoms. On examination he had generalized anasarca and abdominal distension. General physical examination and systemic examination were normal. His routine hematological and biochemical investigation were normal except serum protein. Total serum protein -3.7 gm/dl, albumin- 1.37gm/dl and globuline-2.4 gm/dl. Routine microscopic stool examination for common parasites was negative. A probable clinical diagnosis of malabsorption syndrome due to protein losing enteropathy was made. Patient underwent gastrointestinal endoscopy and revealed numerous whitish spots over the mucosa of ileum and biopsy was taken. Histopathological examination of ileum biopsy showed dilated lymphatics. Then affected part of ileum was resected and sent for histopathological examination. On gross examination resected loop of small intestine measured 10 cm x 3 cm. External surface showed whitish

appearance and cut surface showed multiloculated cystic areas measured 0.5 cm to 3 cm in diameter with thinning of intestinal wall. [figure 1]

Microscopically multiple serial sections showed broadened villi with dilated lymphatic channels within mucosa and submucosa. Lamina propria showed few inflammatory cells infiltrate. [Figure: 2 and 3]

Based on clinical and histopathological findings confirmed the diagnosis of lymphangiectasia. Patient was started on medium chain triglyceride diet along with high protein diet.



Fig. 1: Cut surface of piece of small bowel showed multiloculated cyst like areas with thinning of intestinal and whitish in appearance.

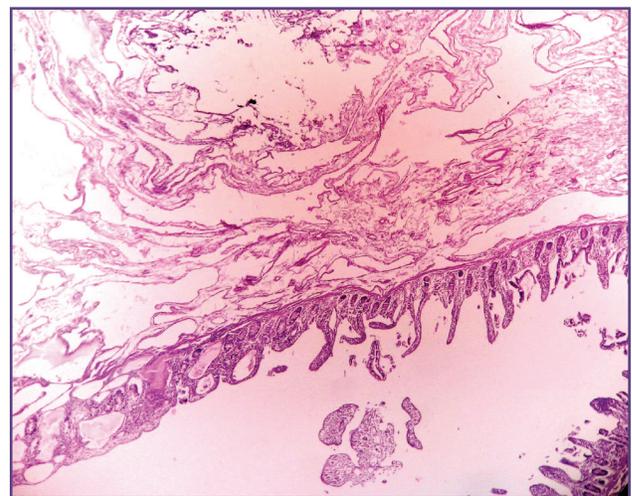


Fig. 2: Microscopy showed broadened villi with dilated lymphatic channels in mucosa and submucosa. [H & E, x 100]

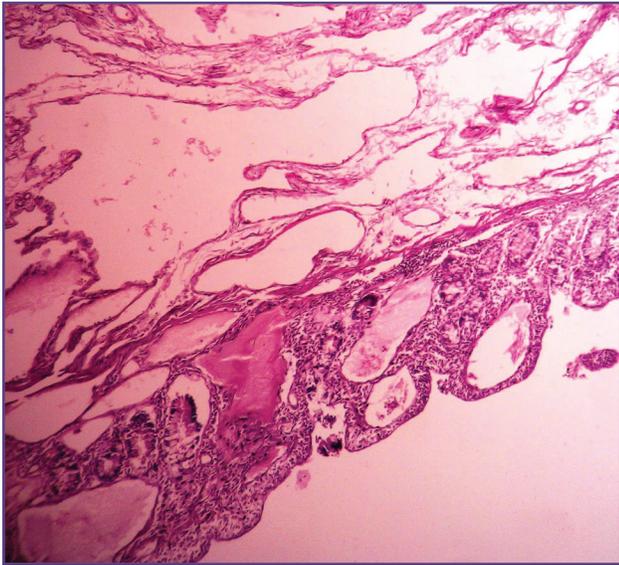


Fig. 3: Microscopy showed broadened villi with dilated lymphatic channels in mucosa and submucosa. Lamina propria showed few inflammatory cells infiltrate. [H&E, x400]

Discussion

Primary intestinal lymphangiectasia (PIL) or Waldmann's disease is characterized by markedly dilation of the mucosal, submucosal and subserosal intestinal lymphatics resulting in leakage of lymph into the lumen of small intestine that may be associated with protein losing enteropathy resulting in hypoalbuminemia, hypogammaglobulinemia, diarrhea, edema and sometimes effusions.^[4] It is diagnosed before 3 years of age but occasionally may be diagnosed in late childhood. Both sexes are equally affected. It may appear as a primary form caused by congenital malformation of lymphatic system. Primary intestinal lymphangiectasia is primarily affecting children and young adults which can occur as an isolated disorder (from birth) or as a part of syndrome such as Noonan, Klippel-Trennay-Weber, Von Recklinghausen, Hennekan and Yellow nail syndrome.^[4] Secondary form occurs due to various condition including inflammatory bowel disease, lymphoma, SLE, sarcoidosis, scleroderma, malignancies, constrictive pericarditis and cardiac surgery.^[2,3]

In the small intestine delicate thin-walled lymph vessels, known as lacteals which absorb nutritional fat and proteins. But in lymphangiectasia the mechanism of protein loss is not well understood. In the literature many theories regarding the mechanism for the increased loss of protein into the bowel in intestinal lymphangiectasia have been proposed. The most accepted theory is an increase in the pressure of the lymph channels which causes protein loss.

^[5] The lymphatic hypoplasia results in an obstruction in lymph flow, which leads to increased pressure within the lymphatics. This, in turn, causes dilation of the lymphatic channels in the intestine and finally leads to the rupture of the channels with discharge of the lymph into the lumen of the bowel. As lymphatic fluid contains a lot of protein, fat and lymphocytes and leakage of lymph causes hypoproteinemia, lymphocytopenia and decreased serum levels of immunoglobulin.^[6]

Clinically patients present as fatigue, abdominal pain, nausea, vomiting, edema, weight loss and diarrhea.^[5] Few patients can also develop hypocalcemia secondary to failure to absorb fat and fat-soluble vitamins. Laboratory investigation show lymphopenia, hypoalbuminemia and low immunoglobulin levels due to lymph leakage from the ruptured lymph vessels.^[7] Our patient presented as diarrhea with pain in abdomen, edema and hypoalbuminemia. Edema may be due to lymphatic abnormality of the affected extremity. The whole lymphatics system below the diaphragm may be abnormal and outflow towards the thoracic duct may be obstructed.^[8]

In our case based on clinical features and laboratory investigation clinical diagnosis of malabsorption syndrome due to protein losing enteropathy was considered. Protein losing enteropathies include various conditions such as Whipple's disease, Crohn's disease, Ulcerative colitis, sprue and intestinal lymphangiectasia. Gastrointestinal endoscopic findings and intestinal mucosal biopsy are essential to differentiate the various causes of protein losing enteropathy.

A diagnosis of Primary intestinal lymphangiectasia (PIL) is based on endoscopic findings and histology in intestinal biopsy specimen.^[3] Endoscopic abnormalities such as scattered white plaques covering the mucosa have been observed but it may be negative when intestinal lesions are patchy in such cases video capsule endoscopy and double balloon enteroscopy assisted biopsy helps to make the histological diagnosis. Other tests proposed to diagnose intestinal lymphangiectasia (IL) are albumin scintigraphy, which shows the albumin leakage into the bowel, CT scan showing diffuse, nodular, small bowel wall-thickening and edema.^[4] In our patient upper gastrointestinal endoscopy was done and showed numerous whitish spots over the mucosa of ileum. We did small bowel resection and marked improvement was observed.

Conclusion

PIL is a rare condition that presents in early childhood with diarrhea. It should be consider as a differential diagnosis of

diarrhea and edema in a childhood age group. Therefore, detailed clinical examination, thorough investigation and follow-up are mandatory.

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