CASE REPORT

26. Primary Intestinal Lymphangiectasia Presenting as Chylous Ascites in a Young Female: A Rare Case

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https://www.youtube.com/watch?v=hJIcIJ1w8oM&list=PLhqNq3xJClbafO0Y5bvBcgMmXpgzJxd44&index=5&t=9821s

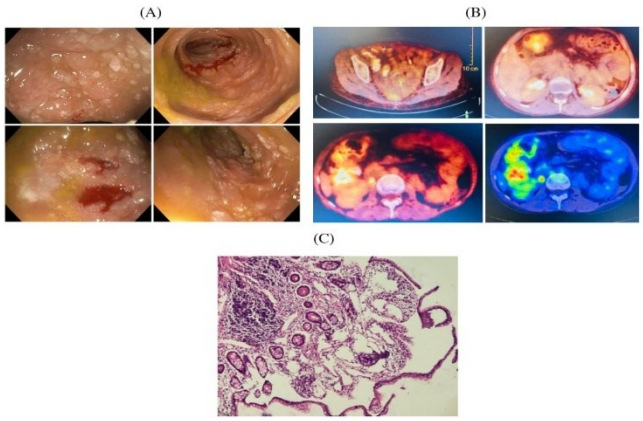
Background: Primary intestinal lymphangiectasia (PIL) is a rare condition that occurs due to the pathological dilation of intestinal lymphatics, characterized by protein losing enteropathy, and occurs more commonly in children. Adults' occurrence is less common, and misdiagnoses with other gastrointestinal disease is prevalent because of the overlapping and nonspecific features.

The Case: A 28-year-old female presented with recurrent abdominal distension and chylous ascites requiring therapeutic paracentesis over the past three years. Medical history was notable for 7 years of diffuse recurrent intermittent crampy abdominal pain more prominent periumbilical and at the right lower quadrant, bloating, loose stools, and weight loss, raising suspicion for Crohn's disease, however, no supportive findings were noted on colonoscopy or histopathology, despite this, she was treated as a case of Crohn's for a short period, but the absence of progression and the new-onset chylous ascites prompted reconsideration of the diagnosis and further investigation into alternative causes of protein-losing enteropathy and ascites.

At presentation, the patient denied fever, night sweats, diarrhea, or overt gastrointestinal bleeding. An abdominal examination showed distention with shifting dullness, consistent with ascites. Without peripheral edema, lymphadenopathy, or hepatosplenomegaly. Laboratory investigations were significant for low serum albumin level of 2.3 g/dL. Ascitic fluid was sterile, and negative for malignant cells. A PET-CT scan was ordered to exclude occult malignancy, autoimmune, inflammatory, or metabolically active conditions. Abdominal CT imaging revealed diffuse thickening of the hepatic flexure of the colon, associated mesenteric fat stranding, and lymphadenopathy (largest node 1.5 cm), in addition to moderate-volume ascites, these findings alongside the clinical picture have raised suspicion for a lymphatic telangiectasia; the patient was started on budesonide and long-acting octreotide (Sandostatin LAR 20 mg intramuscularly every 28 days). She showed clinical improvement following therapy, with reduction in abdominal distension and decreased need for paracentesis. Endoscopic evaluation (Figure 1) demonstrated multiple white plaques in the duodenum on upper endoscopy. Histopathologic examination of the duodenum and ileum was most consistent with primary intestinal lymphangiectasia and revealed lymphatic dilation with preserved villous architecture. No parasites, granulomas, dysplasia, or malignancy were noted.

Conclusion: PIL is rare in adults and can mimic other gastrointestinal disorders, leading to delayed diagnosis. Diagnosis relies on histopathologic confirmation and exclusion of secondary causes. Dietary management is the cornerstone of treatment, with adjunctive therapies reserved for refractory cases. Our patient's diagnostic journey underscored several critical knowledge gaps that continue to challenge clinicians managing intestinal lymphangiectasia. Despite presenting clinical and biochemical features suggestive of the disease, the absence of standardized approaches to quantify lymphatic dysfunction made it difficult to objectively assess disease burden or monitor response to therapy. This case reflects broader gaps in our understanding of the disease's natural history, particularly in adult-onset presentations, which remain poorly characterized. The clinical heterogeneity observed across patients suggests that individualized diagnostic and therapeutic strategies are urgently needed. Continued research is essential to uncover the underlying mechanisms driving variability in presentation and treatment response and ultimately guide the development of more targeted, personalized care.

Figure 1: Medical Image Panel: Endoscopy, PET/CT, and Histopathology Findings.



Legend: (A) Upper gastrointestinal endoscopy showing multiple white plaques in the duodenum, suggestive of lymphatic dilation. These mucosal changes were most prominent in the second part of the duodenum and raised initial concern for infiltrative or lymphatic pathology. (B) Whole-body PET/CT scan demonstrating diffuse FDG uptake in the jejunum, ileum, ascending colon, and proximal transverse colon, consistent with hypermetabolic bowel wall thickening. No discrete focal lesion or metabolically active lymphadenopathy was identified. (C) Terminal ileum biopsy shows dilated lymphatic channels (H&E, 20x)

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