Primary Intestinal Lymphangiectasia Diagnosed by Single-Balloon Enteroscopy

Abstract
Primary intestinal lymphangiectasia (PIL) is a rare protein losing gastroenteropathy that usually affects children and teenagers. There are only a few cases described in the literature. The diagnosis is confirmed by the presence of intestinal lymphangiectasia based on endoscopic findings and histology. We present a case of PIL diagnosed in a teenager patient, by small bowel capsule endoscopy (SBCE) and single balloon enteroscopy (SBE).

Case Report
A 19-year-old male was admitted at a secondary care hospital for clinical signs of protein loosing enteropathy, with diarrhea, hypoproteinemia, peripheral edema and low weight. Laboratory diagnoses showed decreased total proteins 4g/dL, albumins 2.5g/dL, zinc 55µg/dL. No previous family history of lymphangiectasia. Diffuse wall thickening of jejunal and ileal loops was described in the abdominal CT. No lesions were identified in an upper endoscopy. SBCE showed diffusely swollen mucosae, aphthae, erosions and dilatation of lymphatic vessels affecting the entire small bowel mucosa. In distal jejunum there was a polyposis-like pattern.

The patient was sent to a tertiary care hospital for a SBE. The mucosa of the small bowel had atrophy, edematous folds, loss of villi and dilated lymphatic vessels. In the jejunum there were multiple lesions suggestive of pseudopolyps occupying the entire circumference. Biopsies were taken and the microbiological study ruled out *Tropheryma whipplei* infection (PCR). The histological analysis revealed villous widening, vascular congestion and lymphangiectasia without intraepithelial lymphocytosis. These signs were suggestive of PIL.

Nutritional treatment was started with low long chain triglycerides and high protein diet supplements with medium chain triglyceride. Diarrhea and edemas improved and laboratory parameters normalized.

Discussion
PIL is a rare protein losing gastroenteropathy that usually affects children and teenagers. There are only a few cases described in the literature. It is caused by a congenital malformation or obstruction of intestinal lymphatic drainage, characterized by dilated lymphatic vessels of the gastrointestinal tract. When the pressure of lymphatic vessels increases, those dilate and rupture, resulting in leakage of lymph fluid that contains a great amount of protein, fat and lymphocytes. As a consequence, this causes hypoproteinemia, lymphocytopenia and decreased serum levels of immunoglobulin. Edema, diarrhea and ascites are the most common signs, but patients may also present chylothorax, pericarditis, lymphedema, fatigue, diarrhea, growth retardation and smaller physical constitution.

Laboratory findings include lymphopenia and decreased serum total protein and albumin along with decreased immunoglobulin levels including IgA, IgM, and especially IgG [1-3]. The diagnosis is confirmed by the presence of intestinal lymphangiectasia based on endoscopic findings and histology. Upper GI tract endoscopy and colonoscopy could be used to explore the duodenum or terminal ileum, but since intestinal lymphangiectasia often occurs at jejenum-ileum, the biopsies of duodenum or terminal ileum may be negative [1-3]. As a non-invasive procedure SBCE could be more useful to explore the entire small bowel mucosa. The typical endoscopic image is diffusely elongated, circumferential and polypoid mucosa covered with whitish enlarged villi. SBCE is very helpful, but the definite diagnosis is made by the histological examination obtained by BE, with the presence of dilated mucosal and submucosal lymphatic vessels.

Treatment of PIL is not well defined. A low-fat diet with medium-chain triglyceride is the cornerstone of PIL treatment. Medium-chain triglycerides, unlike the long chain triglycerides, are absorbed into the portal circulation and can bypass the lymphatic system, avoiding the increase of lymphatic vessel pressure and the loss of protein. In severe cases, parenteral nutrition could be a complementary treatment. In most patients dietary treatment is permanently needed, because clinical and biochemical findings recur after treatment withdrawal [4-11] (Figure 1 & 2).
Since the first description of the disease, a few cases of lymphoma have been reported suggesting its association. Patients developing lymphoma, do so after decades after the diagnosis. Treatment is surgery and postoperative chemotherapy or chemotherapy alone [12-15].

**Conclusion**

In conclusion, PIL is a rare disease where the clinical suspicion and an endoscopic approach are crucial for the diagnosis.

**Conflict of Interest**

Authors declare no conflict of interest.

**References**


**Figure 1:** Inflamed and cobblestone jejuna mucosa seen by CE.

**Figure 2:** Cobblestone patter seen by SBE.