Case Report

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).

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 after three months of treatment. Nowadays our patient is asymptomatic, under the same treatment and medical control every 6 months.

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 into the bowel lumen 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 jejunum–ileum, the biopsies of duodenum or terminal ileum may be negative [4–9]. 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 polyloid 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.

In our case we were able to visualize the lesions in the entire small bowel by the SBCE. These findings guided us to perform a SBE and mucosal biopsies were taken to achieve the final diagnosis.

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 [3,10,11].

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].

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

References

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

Figure 2: Single Balloon Enteroscopy image of the same lesions.