

CASE REPORT

Degenerated Waldmann's Small Bowel Disease: An Exceptional Entity

A Case Report

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KEYWORDS

Intestinal lymphangiectasia

Waldmann's disease

Exudative enteropathy

Macrophage activation syndrome

Abstract:

Primary intestinal lymphangiectasia, also known as Waldmann's disease, is a rare pathology first described in 1961, typically presenting in childhood or adolescence. It is characterized by exudative enteropathy of lymphatic origin. Diagnosis is based on clinical, biological, and endoscopic criteria after excluding secondary forms. Complications can arise, the most severe being malignant non-Hodgkin's lymphoma. We report the case of a 39-year-old man with a history of hypothyroidism, managed for exudative enteropathy linked to Waldmann's disease with a low-fat diet and vitamin supplementation. He was admitted with acute intestinal obstruction. An abdominal CT scan confirmed acute mechanical small bowel obstruction. Emergency surgery revealed staged small bowel stenoses and a 3 cm tumoral lesion. A carcinological bowel resection was performed, with delayed restoration of digestive continuity due to the patient's critical condition. Pathology identified mucinous adenocarcinoma associated with intestinal lymphangiectasia. Postoperative complications included a digestive fistula, fever, pancytopenia, and neurological symptoms such as seizures. A myelogram revealed hemophagocytosis, and macrophage activation syndrome was suspected. Despite treatment, the patient developed status epilepticus and died three months later.

Introduction:

Primary intestinal lymphangiectasia or Waldmann's disease is a rare pathology first described in 1961. It usually manifests during childhood or adolescence. It is characterized by exudative enteropathy of lymphatic origin [1].

Diagnosis is based on clinical-biological and endoscopic criteria, after ruling out secondary intestinal lymphangiectasia. Several complications may occur during the progress of the disease, the most dreaded of which is malignant non-Hodgkin's lymphoma.

We report a case of adenocarcinoma on exudative enteropathy revealed by acute intestinal obstruction.

Observation:

This is a 39-year-old patient with a history of hypothyroidism treated with levothyroxine, followed in the internal medicine department for exudative enteropathy associated with Waldman's disease, managed with a low-lipid diet and vitamin supplementation. He was admitted to our surgical department with an occlusive syndrome consisting of abdominal distension and pain, associated with vomiting without cessation of bowel movements and gas.

Clinical examination revealed an altered general condition with cachexia and conjunctival pallor without fever. Abdominal examination revealed a distended tympanic abdomen with diffuse abdominal tenderness and no palpable abdominal mass. There is no hernia revealed upon examination of the hernial orifices. Rectal examination revealed normo-coloured stools with no palpable mass. The remaining examination was otherwise normal.

The unprepared abdomen showed small bowel-type air-fluid levels without pneumoperitoneum.

Biological tests showed hypoalbuminemia, anemia, acute renal failure and ionic disorders without biological inflammatory syndrome.

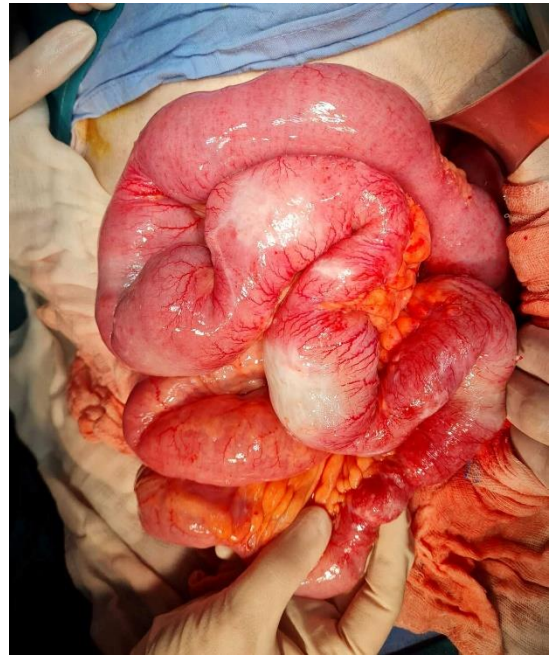
An urgent abdominal CT scan was done, showing the presence of an acute mechanical small bowel obstruction with no signs of digestive distress. The patient underwent urgent surgery. Intraoperative exploration showed the presence of staged stenoses of the small bowel, situated 120 cm from the duodeno-jejunal angle extending over 80 cm, associated with a 3 cm tumoral lesion of the small bowel with multiple adhesions to the sigmoid.

(figure1)

The patient underwent a carcinological small bowel resection, removing the pathological small bowel, and the restoration of digestive continuity was postponed because of the patient's biological disorders and cachectic state.

Pathological examination showed a mucinous adenocarcinoma of the small intestine with intestinal lymphangiectasia.

One month later, the patient underwent elective continuity restoration.



1: Intraoperative exploration showing staged small bowel stenoses

The postoperative course was marked by the appearance of a well-directed digestive fistula, for which he was taken on medical treatment. This was followed by prolonged fever, associated with deterioration in general condition and neurological signs such as irritation and convulsive seizures, the etiological investigation of which was negative. Biologically, the patient developed pancytopenia. A myelogram showed images of hemophagocytosis. Macrophagic activation syndrome was suspected and the diagnostic investigation was completed, showing hyperferritinemia, hypofibrinemia and hypertriglyceridemia. The course was marked by the onset of status epilepticus and the patient's death after 3 months of treatment.

Discussion:

Primary intestinal lymphangiectasia (PIL), or Waldmann's disease, is a sporadic disorder of unknown prevalence, first described in 1961 by Waldmann in 18 patients [2]. It typically begins in childhood before the age of 3, but it can be diagnosed later in adulthood [1]. It is characterized by exudative enteropathy of lymphatic origin, which is a clinico-biological syndrome secondary to lymphatic engorgement with exudation of chyle rich in proteins, lipids and lymphocytes exceeding the liver's capacity for synthesis [3].

Diagnosis is established based on a combination of clinical, biological and endoscopic arguments, after ruling out secondary intestinal lymphangiectasia caused by obstruction of lymphatic flow due to

neoplasia (lymphomas), infection (Whipple's disease, parasitic infection, intestinal tuberculosis), inflammation (Crohn's disease, systemic lupus erythematosus), drainage pathway fibrosis (mesenteric sclerosis, retroperitoneal fibrosis, post-radiation fibrosis), liver cirrhosis, constrictive pericarditis, abdominal surgery, among others.

Clinically, LIP is manifested by the presence of peripheral edema, of variable intensity, associated with serous effusions (pleural, pericardial, intraperitoneal) which, in major forms, can be life-threatening [1]. Other signs may include anemia, asthenia, lymphedema of the limbs, and moderate, intermittent diarrhea [1], [4].

Waldmann's disease is characterized biologically by hypo-proteinemia due to digestive loss, associated with a deficiency of calcium, iron, copper, lipoproteins and circulating lymphocytes [3], [5].

The cellular immune deficiency is linked to lymphocyte depletion, mainly affecting TCD4+ lymphocytes, but also TCD8+ lymphocytes and B lymphocytes, making the patient more susceptible to chronic viral infections and neoplasia such as lymphomas of the gastrointestinal tract. Hypogammaglobulinemia adds to this lymphopenia and predisposes to infections [3], [6].

Measuring fecal alpha1-antitrypsin clearance in 24-hour stool collections is the gold standard for the biological diagnosis of Waldmann disease, reflecting gastrointestinal protein exudation [3].

Oesogastroduodenalfibroscopy combined with staged biopsies confirms the diagnosis, showing yellow/brown lymphatic dilatations of variable density and size [1].

Diet is the main therapeutic weapon in Waldmann disease, based on a hypolipid and hyperprotein diet enriched with medium-chain triglycerides and supplemented with fat-soluble vitamins. Reducing the lipid content of the diet helps to reduce chylomicron formation, thereby reducing pressure within the splanchnic lymphatic circulation and, consequently, lymphatic leakage. However, medium-chain triglycerides are not absorbed via chylomicrons and ensure the body's necessary lipid intake without clogging the splanchnic lymphatic circulation [7], [8].

Other treatments, such as octreotide, sirolimus and everolimus, have been proposed in the literature, although there are no randomized prospective studies on their efficiency [1]. Surgical treatment plays a minor role. It is proposed for segmental and localized lymphangiectasias or the treatment of complications [1]. The occurrence of malignant non-Hodgkin's lymphoma, whether digestive or extra-digestive, although rare, represents a serious complication of PIL, the mechanisms of which are still poorly understood [1]. Other complications have been described in the literature, such as recurrent infections, the gelatinous transformation of the marrow, hemolytic uremic syndrome and malabsorption syndromes [1].

Macrophage Activation Syndrome (MAS) is a rare, severe, and potentially fatal systemic disease. It is characterized by intense and inappropriate activation of the immune system, leading to the infiltration of tissues by activated macrophages, resulting in a condition known as "Cytokine Storm" [9]. There are primarily two forms of this syndrome: primary MAS, which includes hereditary diseases of the immune system with T-lymphocyte and macrophage activation, and secondary MAS, which occurs in the course of neoplastic, autoimmune or infectious diseases [6], [9], [10]. Clinically, the manifestations are non-specific, and it is their association that should prompt a diagnosis. Currently, the Henter criteria are accepted as the diagnostic criteria for MAS, and the diagnosis of MAS is based on the presence of five of eight criteria: fever, splenomegaly, cytopenia (hemoglobin (Hb) < 9 g/dl, platelets < 100,000/mm³, neutrophils < 1,000/mm³), hypertriglyceridemia (> 3 mmol/l) and/or hypofibrinemia (< 1.5 G/l), hemophagocytosis in the marrow (or other tissues: lymph node, spleen, etc.), ferritin > 500 mg/mm³, soluble CD25 > 2,400 U/ml and absent or reduced natural killer (NK) activity [9]. The treatment of MAS is still poorly codified. It combines both symptomatic and specific treatment, notably corticosteroid therapy (dexamethasone) and immunoglobulins (of mediocre efficacy in primary forms) [6], [9], [10].

Conclusion:

Primary intestinal lymphangiectasia is a chronic and rare disease, often challenging to diagnose, relying on a combination of clinical, biological, and endoscopic evidence. Its treatment primarily revolves around dietary measures. Multiple complications can arise during the disease, some of which are fatal.

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Consent

Written informed consent was obtained from the patient for the publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Conflict of interest

No conflict of interest to disclose.

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