Primary Systemic Amyloidosis Presenting as a Colonic Stricture: Successful Treatment with Left Hemicolectomy Followed by Autologous Hematopoietic Stem-Cell Transplantation

Report of a Case

S. Rives, M.D.,* M. Pera, M.D.,† L. Rosinol, M.D.,” O. Vidal, M.D.,† R. Miquel, M.D.,‡ M. Solé, M.D.,‡ J. García-Valdecasas, M.D.,† J. Blade, M.D.*

Intestinal tract involvement by primary systemic amyloidosis is frequent but usually asymptomatic. Ischemic colitis caused by amyloid infiltration of wall blood vessels can occasionally be observed. We report a 62-year-old female with primary systemic amyloidosis who presented with intestinal obstruction caused by ischemic stricture of the sigmoid colon, secondary to submucosal amyloid deposition. The patient was successfully treated with surgical resection followed by high-dose chemotherapy and hematopoietic stem-cell transplantation. The clinical manifestations and differential diagnosis of gastrointestinal involvement of primary systemic amyloidosis, as well as its current treatment, are discussed. [Key words: Primary systemic amyloidosis; Hematopoietic stem-cell transplant; Intestinal; Colon; Surgery]

Amyloidosis are a group of disorders characterized by the extracellular deposition of an amorphous substance that stains with Congo red, showing an apple-green birefringency under polarized light microscopy. The composition of the amyloid fibrils depends on the different types of amyloidosis. In primary amyloidosis (AL) the amyloid is composed of the variable portion of the immunoglobulin light chains. These light chains are synthesized by a monoclonal population of plasma cells. The clinical symptoms are caused by the deposition of amyloid in different organs and tissues, impairing their normal function. The main clinical features associated with AL are nephrotic syndrome, cardiac failure, hepatomegaly, and sensorimotor neuropathy. Histologic involvement of the intestinal tract is common but usually asymptomatic. When gastrointestinal (GI) symptoms occur, they are more often related to automatic dysfunction than to amyloid infiltration of the GI tract. Ischemic colitis caused by amyloid infiltration of wall blood vessels presenting as abdominal pain, rectal bleeding, and intestinal ulcers can occasionally be observed. Cases of true mechanical intestinal obstruction (amyloidoma, megacolon, and volvulus) are exceedingly rare.2,4

We present a case of intestinal obstruction caused by an ischemic stricture in sigmoid colon, successfully treated with surgical resection followed by high-dose therapy and hematopoietic stem-cell transplantation as the primary treatment of the underlying disease. The clinical manifestations and differential diagnosis of GI involvement of AL as well as current treatment approaches are discussed.
REPORT OF A CASE

A 62-year-old female presented with a three-month history of dysphagia, colic abdominal pain, diarrhea, and intermittent rectal bleeding. She had lost 6 kg of weight. Physical examination showed bilateral conjunctive infiltration and marked enlargement of sub-mandibular structures. Her abdomen was distended but not tender, and peristalsis was increased. Rectal examination was normal. Laboratory investigation revealed the presence of a serum M-spike of 6 g/l (IgG λ type) and a 24-hour λ type chain urine protein excretion of 1 g. Hepatic and renal functions were normal. Factor X was normal. A bone marrow aspirate showed 57 percent of plasma cells. The skeletal survey was negative for bone lytic lesions. A radiopaque contrast enema revealed a 15-cm-long segmental sigmoid colon stricture (Fig. 1). Colonoscopy showed a severe stricture in sigma, which prevented the endoscope progression. The mucosa in the stenotic area was friable, with multiple ulcerations, the endoscopic pattern mimicking Crohn's disease. Biopsies showed submucosal deposition of amyloid. The patient was diagnosed as having primary systemic amyloidosis with extensive colonic involvement. Because of impending obstruction, surgical resection of the left colon was indicated. Surgery was started by laparoscopic approach, conversion to open surgery was decided because of submucosal hemorrhage and bleeding of the peritoneal surface, and conventional left hemicolectomy was completed. Histopathologic examination showed full-thickness infiltration of the colonic wall with AL type amyloid substance (λ light chain). In addition, the deposits occluded the blood vessel lumens (Figs. 2 and 3). After the postoperative period the patient received high-dose therapy with melphalan (140 mg/m²) followed by peripheral stem-cell rescue (HDT/SCT) as therapy for her primary...