Challenges in the diagnosis of enteropathy-associated T cell lymphoma

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CASE PRESENTATION

A 39-year-old man of Indian descent presented with a one-month history of epigastric pain, fatigue, decreased appetite and weight loss of 6 kg. Two months earlier, he was diagnosed with celiac disease by his primary care physician based on serology (antitissue transglutaminase level 90 U/L) and had since been on a gluten-free diet. Relevant history was remote exposure to tuberculosis in India with Mantoux test conversion, for which he received nine months of isoniazid. On admission to hospital, computed tomography revealed circumferential thickening of a 6 cm segment of small bowel, with significant intra-abdominal lymphadenopathy and multiple subcentimetre pulmonary nodules suggestive of previous granulomatous disease. To evaluate the small bowel abnormality, proximal double-balloon enteroscopy (DBE) (Fujinon Inc, Japan) was performed and demonstrated widespread features of celiac disease, along with multiple areas of stenoses and ulceration in the mid-jejunum (Figure 1). Further advancement of the endoscope was not possible due to significant stenosis from a circumferential mass lesion. Extensive biopsies showed ulcerative jejunitis but no evidence of malignancy. Because a malignant diagnosis was suspected, a distal DBE was performed to sample tissue from an alternative area of small bowel; however, the area of interest was unreachable from the retrograde approach. A subsequent positron emission tomography scan showed disease progression in the chest and intense uptake in the left adrenal gland. A bronchoscopy performed to rule out active tuberculosis with transbronchial biopsy was normal. Endoscopic ultrasound (EUS) using a linear echoendoscope (GF-UCT140-AL5, Olympus Inc, Japan) demonstrated a hypoechoic adrenal nodule 24 mm × 13 mm in size. EUS-guided biopsy using a 22-gauge EchoTip ProCore needle (Cook Medical, USA) revealed a mixed composition of large, atypical and small mature lymphocytes, and an intense plasmacytic infiltrate on histology. Stains for acid-fast bacilli and fungal elements were negative. Immunophenotypic analyses from flow cytometry confirmed mixed inflammation with relative predominance of a T cell population and a high mitotic index, which were highly suspicious, yet unable to confirm malignancy. Because minimally invasive modalities had failed to obtain a definitive diagnosis, the patient underwent surgical lymph node sampling using laparoscopy that was converted to a mini laparotomy because of the deep location of the lymph nodes. Intraoperatively, the jejunum appeared abnormal, with multiple transmural skip lesions. Frozen sections of the sampled lymph nodes were deemed either bland or reactive. Finally, segmental jejunal resection was performed on an area 110 cm distal to the ligament of Treitz with end-to-end anastomosis, and the diagnosis of enteropathy-associated T cell lymphoma (EATL) was ultimately confirmed (Figures 2 and 3). The patient progressed to receive two cycles of cyclophosphamide, etoposide, vincristine and prednisone. He experienced complications of a wound infection and a superficial intra-abdominal infection requiring percutaneous drainage. Because he exhibited a poor initial response, a more aggressive regimen was initiated with higher doses of chemotherapy, followed by an autologous stem cell transplant. Due to complications of small bowel obstruction and febrile neutropenia, he passed away 11 months after the diagnosis of EATL.

DISCUSSION

EATL is a rare and fatal complication of celiac disease. It often presents with multifocal involvement of the mid-small bowel, making minimally invasive attempts at diagnosis challenging (1). The aggressive nature of this disease dictates the urgency in making the diagnosis. In the past, surgery was often required for disease complications, such as bowel obstruction or perforation, but also as the primary diagnostic modality (2). Recently, attempts have been made to avoid surgery before aggressive chemotherapy to preserve patient functional status and avoid problems with wound healing. Even laparoscopic surgery...
requires prolonged recovery times due to disease-related malabsorption and ensuing chemotherapy (1,3). In 2007, Hadithi et al (4) reported five cases of EATL diagnosed by biopsies taken via DBE, while other reports have shown benefit with DBE in evaluating refractory celiac disease (5). However, as our case highlights, despite advances in small bowel endoscopy and other minimally invasive techniques, invasive surgery may still be required to confirm the diagnosis of EATL. There are, however, important rationales for avoiding diagnostic surgery in these patients, and we continue to believe that a minimally invasive approach should be pursued when possible.

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REFERENCES