A condition of unknown significance: esophageal melanocytosis in a patient with celiac disease

A 74-year-old woman was admitted to our unit with complaints of abdominal discomfort and heartburn over 2 months. She had a history of hypertension and celiac disease, but admitted that she did not comply with a gluten-free diet. She had no history of any medication, smoking, or drinking alcohol, and her family history was unremarkable.

Physical examination findings were normal. Serological studies revealed positivity for anti-tissue transglutaminase, anti-gliadin, and anti-endomysial antibodies. An upper gastrointestinal endoscopy showed multiple, focal, flat and black pigmented areas in the middle and distal esophagus (Fig. 1a). Multiple biopsies were obtained from these areas. In addition, atrophic folds and scalloping were observed in the second part of the duodenum during the endoscopic examination (Fig. 1b). Histological examination of esophageal biopsies showed melanin deposits in the basal layer and lamina propria of the squamous epithelium (Fig. 2a). Duodenal biopsy specimens revealed lymphocyte infiltration, crypt hyperplasia, and villous atrophy, compatible with celiac disease grade IIIb according to the Marsh classification (Fig. 2b) [1, 2].

Esophageal melanocytosis is a benign and rare condition characterized by melanocytic proliferation in the basal layer of esophageal squamous epithelium, with an increased aggregation of melanin pigments. The etiology and pathogenesis of esophageal melanocytosis remain uncertain; however, it may be caused by gastrointestinal reflux or other conditions that lead to continued and chronic stimuli at the esophageal mucosa. It has also been reported to be associated with Addison’s disease, Laugier–Hunziger syndrome, oral melanoma, esophageal squamous cell carcinoma, and celiac disease [3]. There are insufficient data to establish a guideline regarding treatment and surveillance of esophageal melanocytosis. However, the most important point is that esophageal melanocytosis must be differentiated from malignant melanoma [4].

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Fig. 1 Endoscopy images. a Multiple, focal, flat and black pigmented areas in the middle and distal esophagus. b Atrophic folds and scalloping in the second part of the duodenum.

Fig. 2 Histological appearance of biopsies. a Esophagus – melanin deposits in the basal layer and lamina propria of the squamous epithelium. b Duodenum – lymphocyte infiltration, crypt hyperplasia, and villous atrophy in the mucosa, compatible with celiac disease.