

Endoscopic images in Fabry disease

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Fabry disease, also called Anderson-Fabry disease, is the second most prevalent lysosomal storage disorder. It is an X-linked inborn error of the glycosphingolipid metabolic pathway [1]. The accumulation of a metabolic product, called globotriaosylceramide, within lysosomes in a wide variety of cells [2], produces the many manifestations of the disease, which include: severe neuropathic or limb pain, renal disease, cardiac and cerebral involvement [3].

Dermatological manifestations affect more than 70% of patients and include: telangiectasias and vascular lesions called angiokeratomas. The latter are characteristic of the disease and affect the groin, hip and periumbilical areas.

We report the case of a 42-year-old male, with Fabry disease, diagnosed in his twenties, according to kidney histology and the typical dermatological manifestations. He developed end-stage renal disease at the age of 38. He presented to our unit for upper and lower endoscopy for pretransplantation control. He occasionally complained of abdominal pain. At physical examination there were angiokeratomas at the periumbilical area and the groin.

We identified a few angiokeratomas at the mucosa of the esophagus (Fig. 1). The endoscopic appearance of the mucosa of the stomach was consistent with chronic gastritis with no evidence of angiokeratomas. The mucosa of the colon was otherwise normal, with a few angiokeratomas at the sigmoid and descending colon (Fig. 2). The endoscopic appearance of angiokeratomas was similar to those observed on the skin.

We conclude that angiokeratomas are not only dermatological manifestations in Fabry disease, but they are also detected at the mucosa of the gastrointestinal tract.

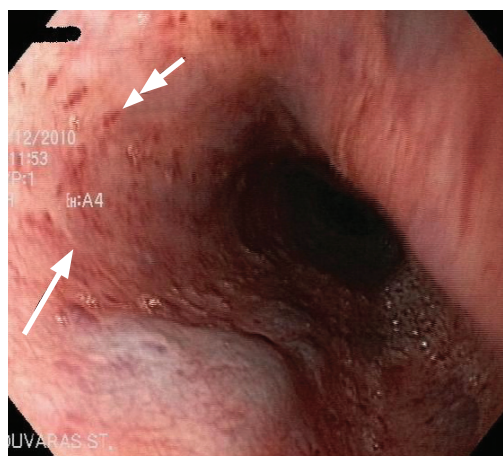


Figure 1 Angiokeratomas of esophagus



Figure 2 Angiokeratomas of colon

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