PICTURES IN DIGESTIVE PATHOLOGY

Gastric mucosa appearance in a patient with Tangier disease

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A 24-year-old male was diagnosed with Tangier disease. He had no family history, exhibited lipid changes, and was free of coronary disease. He was referred to our department for an increased number of stools (2-4/day) at daytime, mostly liquid, maybe related with the ingestion of fatty foods (hamburgers). No weight loss was present. The physical exploration was normal. Laboratory tests showed: cholesterol 33 mg/dl (normal: 220-250); triglycerides 100 mg/dl (normal: 170-350); HDL 1 mg/dL (normal: 35-55), LDL 10 mg/dL (normal: 130-159); apoprotein A < 5 mg/dl (normal: 115-190); transaminases, coagulation, orosomucoid, CRP, antigliadin and antitransglutaminase antibodies, fat-soluble vitamins, and fecal chemotrypsin were normal. A gastric endoscopy showed numerous tiny white-yellowish injuries at the antral mucosa (Fig. 1). The duodenum showed no alterations. A gastric biopsy revealed extended xanthomatosis (Tangier disease) (Fig. 2) with no injuries in the duodenal mucosa. The evolution was satisfactory, and the bowel habit regressed to normal with no need for medication.

Fig. 1. Gastric mucosa with white-yellowish fatty deposits.
Mucosa gástrica con depósitos grasos amarillo-blanquecinos.

Fig. 2. Numerous histiocytes with a foamy cytoplasm fill the lamina propria and push glandular structures wider apart (HE. x 400).
Numerosas células histiocitarias con citoplasma espumoso que ocupan la lámina propia y separan las estructuras glandulares (HE. x 400).

DISCUSSION

Tangier disease is a disorder of lipid metabolism that was first described in 1961. It is transmitted in a recessive manner. These subjects have very low plasma cholesterol levels and their HDL is frequently undetectable (1). Triglycerides are usually normal or mildly elevated. This is an unknown, rare disease with approximately one hundred cases reported in our country (2,3). The underlying molecular defect is poorly understood, although apolipoprotein A-I catabolism is increased.

Severe HDL deficiency in the blood originates cholesterol deposition in both reticuloendothelial and Schwann cells (4). Orange tonsils, hepatosplenomegaly, and peripheral neuropathy are usual manifestations (3). Our patient, who was diag-

nosed with Tangier disease based on lipid metabolism testing, had no orange tonsils. Such an absence, while unusual, has been previously described (5). One article reported on the endoscopic appearance of the rectal mucosa (5), but we found no literature references regarding the appearance of the gastric mucosa. Endoscopic findings as seen in our patient reflect a likely relationship to Tangier disease.

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