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IMAGES IN PAEDIATRICS

Gastrointestinal inflammation in glycosylation defect Ib☆

Inflamación gastrointestinal en defecto de la glicosilación Ib

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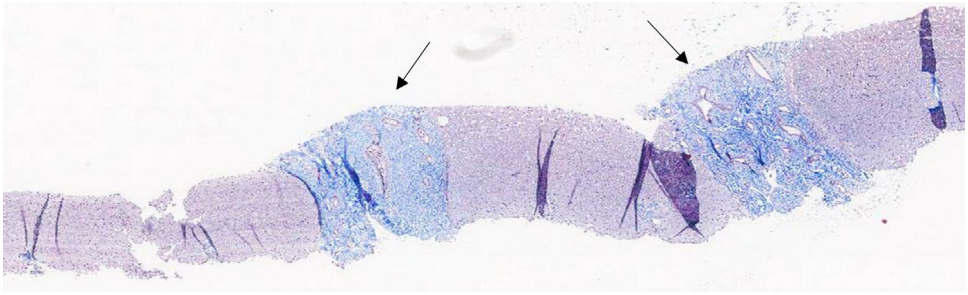

Figure 1 Masson trichrome stain. This techniques stains collagen fibers blue, allowing their visualization. Fibrosis of portal areas of the liver without formation of porto-portal fibrous bridges (arrows). Metavir fibrosis score: F1.

A boy aged 9 years presented with chronic diarrhea, failure to thrive and hepatomegaly, a calprotectin level greater than 500 mg/kg, slightly abnormal inflammatory markers and normal liver function. The ultrasound scan showed signs of chronic liver disease and the findings of the biopsy were consistent with congenital hepatic fibrosis (Fig. 1). The endoscopy revealed a polypoid lesion in the lesser curvature of the stomach (Fig. 2) 20–30 cm from the anal margin with isolated ulcers in the rest of the colon (Fig. 3), compatible with gastric polypoid foveolar hyperplasia and

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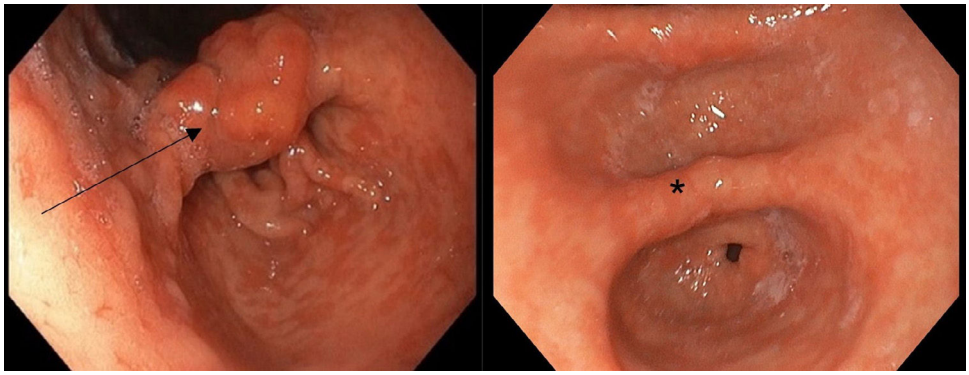

Figure 2 Stomach. Loss of vascular and mucosal pattern with erythematous areas, mucosal cobblestone appearance at the level of the lesser curvature and antrum. Edematous and congestive lesion measuring 1–2 cm compatible with multilobulated sessile polyp (arrow). Minimal residual lesion after oral mannose therapy (*).

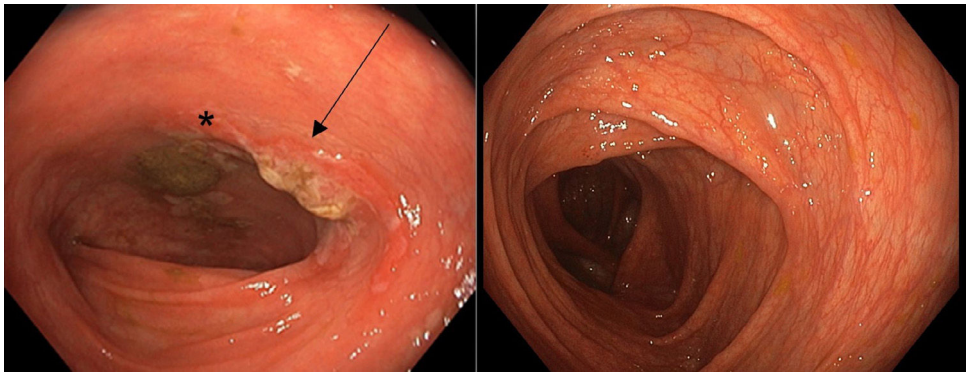

Figure 3 Colon. Mucosa with edematous appearance and associated fibrin, suggestive of sessile polyp (arrow) with isolated aphthous lesions alternating with normal-appearing mucosa (*). Normalization of endoscopic lesions after initiation of oral treatment with mannose.

nonspecific colitis. Exome sequencing and analysis identified two variants in the mannose phosphate isomerase (*MPI*) gene, associated with congenital disorder of glycosylation (CDG) type Ib, prompting initiation of oral mannose therapy. Six months later, bowel movements and calprotectin levels had normalized and the endoscopic lesions had resolved.

The manifestations of CDG Ib are mainly hepatic and gastrointestinal, including vomiting, protein-losing enteropathy and failure to thrive, in absence of neurologic manifestations.[1–3] In some cases, congenital hepatic fibrosis is the sole presenting symptom.[1] In the scant literature on the subject, the most commonly reported endoscopic findings are villous atrophy and lymphoplasmacytic inflammation.[3] We present this case due to the singularity of the endoscopic lesions, the similarities with inflammatory bowel disease and the improvement achieved with mannose therapy.

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