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IMAGES IN PAEDIATRICS

Chylomicron retention disease: A condition to keep in mind

Enfermedad por retención de quilomicrones: entidad a considerar

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A male infant aged 10 months with a history of chronic diarrhoea, vomiting and abdominal distension refractory to free amino acid formula. The physical examination revealed abdominal distension and small amount of subcutaneous fat; with a weight of 7.75 kg (z score, −1.48) and a height of 70 cm (z score, −1.32).

The results of the complete blood count, thyroid, hepatic and renal function tests and immunoglobulin tests were normal, with a negative result for tissue transglutaminase IgA. The stool elastase and microbiology tests were negative. The gastrointestinal endoscopy with biopsy did not lead to detection of macroscopic or histological lesions in the colon. The duodenal mucosa had a whitish appearance with no changes in the villi (Fig. 1) and the histological examination revealed enterocyte vacuolization (Fig. 2).

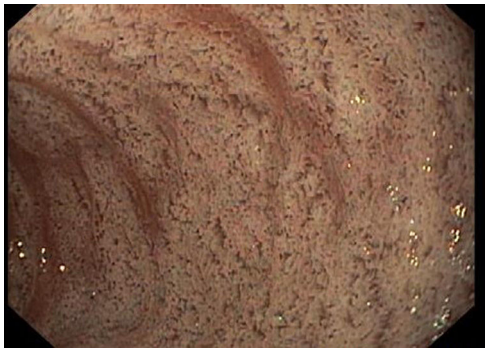

Figure 1 Endoscopic image of the descending part of the duodenum: whitish appearance of mucosa.

The salient findings of the lipid panel were: hypocholesterolaemia (total cholesterol, 81 mg/dL; LDL cholesterol, 25 mg/dL; HDL cholesterol, 23 mg/dL), a triglyceride level of 164 mg/dL, hypovitaminosis E (2 mg/L), hypovitaminosis D (18 ng/mL), decreased apolipoproteins AI and B100. The analysis of the *SAR1B* gene confirmed the diagnosis of chy-

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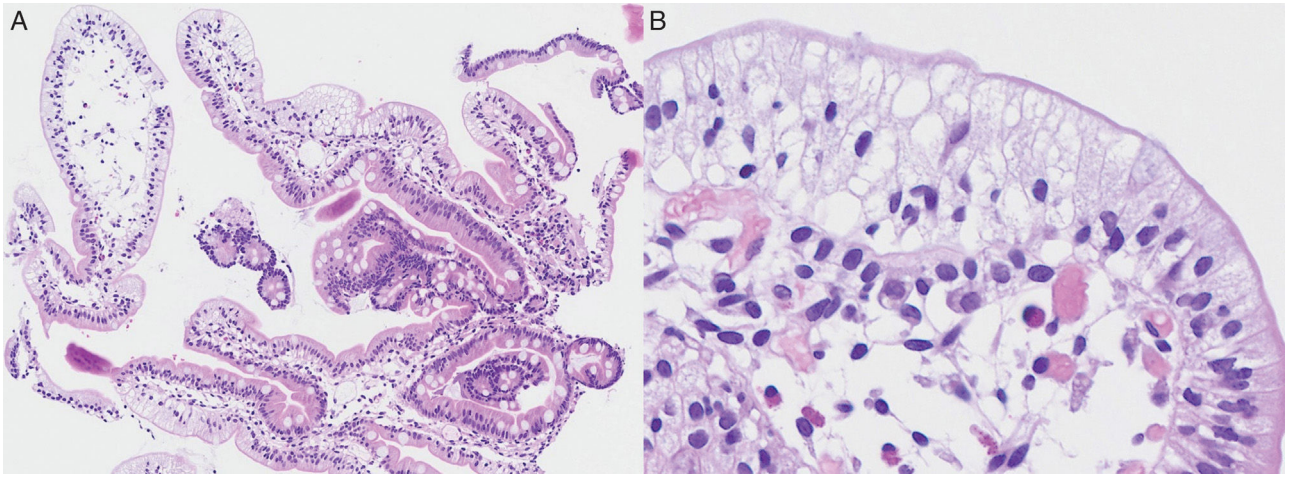

Figure 2 Duodenal histology. (A) Magnification ×10. Duodenal mucosa with preserved villous architecture and vacuolization of clusters of enterocytes in the apical portion of the villi (haematoxylin/eosin). (B) Magnification ×60. Vacuolization of enterocytes (haematoxylin/eosin).

lomicron retention disease (CRD) through the detection of the homozygous variant c.389 T > G (p.L130R) in exon 6. The patient was managed with administration of liposoluble vitamins, a low-fat diet supplemented with medium-chain triglycerides and essential fatty acids,[1] which achieved improvement of diarrhoea and anthropometric measurements: 12.8 kg (z score, +0.04) and 88 cm (z score, −0.49) at age 29 months.

Chylomicron retention disease is an autosomal recessive lipid malabsorption disorder characterised by changes in chylomicron formation due to defects in transport between the endoplasmic reticulum and the Golgi apparatus mediated by SAR1B-GTPase.[2] Patients tend to present with diarrhoea, failure to thrive, abdominal distention, vomiting, hypocholesterolaemia and normal triglyceride levels.[3]

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Conflicts of interest

The authors have no conflicts of interest to declare.

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