Fat accumulation in enterocytes: a key to the diagnosis of abetalipoproteinemia or homozygous hypobetalipoproteinemia

A 20-year-old woman was referred by her ophthalmologist to investigate the reason for her hypovitaminosis A and secondary night blindness. She had no other symptoms of deficiencies in fat-soluble vitamins, no abdominal complaints, and no weight loss.

Laboratory examination revealed a deficiency in vitamin A (< 12µg/dL, normal range 30 – 80) and vitamin E (<0.30 mg/dL, normal range 0.5 – 1.8), a very low prothrombin time (30 %), and very low levels of cholesterol (30 mg/dL), triglycerides (4 mg/dL), and LDL-cholesterol (below the level of detection). Her level of 25-hydroxy vitamin D appeared to be normal, but at the time of her first admission, vitamin D substitution had already been started. A slightly raised alanine aminotransferase was also detected (33 U/L).

Further work-up excluded cystic fibrosis, exocrine pancreas insufficiency, and celiac disease. Gastroscopy revealed a very pale duodenal mucosa; the villi, however, could be easily recognized (Fig. 1). Videocapsule endoscopy revealed a pale small bowel mucosa with extremely pronounced villi (Fig. 2, Video 1). Biopsies of the duodenal mucosa revealed areas of extended supranuclear vacuolization of the cytoplasm in the villi. These areas were interspersed with normal areas (Fig. 3 and Fig. 4).

These findings suggested a diagnosis of either abetalipoproteinemia or homozygous hypolipoproteinemia, disorders that are caused by mutations in both alleles of the microsomal triglycerides transfer protein (MTP) or in the APO-B gene, respectively [1–2] This results in the failure of APO B-100 synthesis in the liver and APO B-48 synthesis in enterocytes, leading to fat accumulation in the small intestine. This diagnosis can be con-
firmed by sequencing the MTP and APO-B genes [1].
This disorder can be treated by a low-fat diet, supplementation of essential fatty acids, and high oral doses of fat-soluble vitamins [1]. Follow-up is necessary to monitor potential ophthalmologic, neurologic, hematologic, and hepatologic complications [1–2].

This patient illustrates that the disorder is sometimes diagnosed in adulthood when the phenotype is mild [1–3]. The prognosis is variable but adherence to the treatment regime can restore neurologic function and prevent subsequent disease progression [1–2].

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