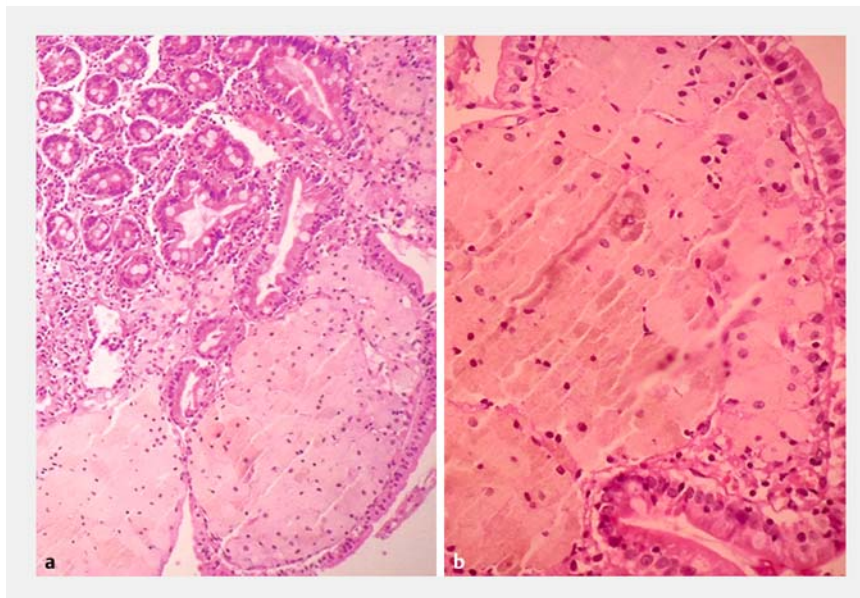


Cholesteryl ester storage disease: endoscopic findings of an orphan disease

A 23-year-old man with normal antenatal history was admitted to hospital because of right hypochondrial pain, periodic nausea, and fatigue. The patient reported an episode of idiopathic jaundice with moderate fever 5 years previously. He was a non-smoker who did not drink alcohol nor take oral or intravenous drugs. Evaluation for viral hepatitis was negative. His physical examination revealed a high body mass index (BMI) of 32.2 kg/m², his liver was palpable 2 cm below the costal margin, but his spleen was non-palpable. Laboratory test results showed a leukocytosis, as well as high levels of total bilirubin (29.2 μmol/L), conjugated bilirubin (7.7 μmol/L), AST (48 U/L), total cholesterol (7.58 mmol/L), triglycerides (1.82 mmol/L), LDL-C (5.29 mmol/L), and VLDL-C (0.83 mmol/L), but his HDL-C level was normal (0.67 mmol/L).

Abdominal ultrasonography showed hepatosplenomegaly with moderate diffuse parenchymal changes in the liver (liver steatosis) and pancreas. Arteriosclerotic vascular disease with hemodynamically relevant stenotic brachiocephalic vessels (20%–35%) was found on brachiocephalic vessel ultrasonography. Upper gastrointestinal endoscopy revealed regions of flat yellow- and brown-speckled pigmented mucosa from the descending part of the duodenum to the fourth part (▶ **Video 1**). Histopathological examination of a biopsy taken from the duodenal mucosa showed numerous macrophages containing brownish granules in their cytoplasm (▶ **Fig. 1**). Staining of the specimen with Perls' stain for iron-containing deposits was completely negative (▶ **Fig. 2**). An ultrasonography-guided liver biopsy was performed, the results of which revealed moderate histologic hepatic activity, fibrous degeneration (grade 3), and severe steatosis with an accumulation of foam cells. Homozygosity for pSer103Arg+0 IVS81G>A was found on genetic sequence analysis of the *LIPA* gene. Lysosomal acid lipase (LAL) deficiency cholesteryl ester storage disease (CESD) was confirmed.



▶ **Fig. 1** Histopathological images from a biopsy of the duodenal mucosa stained by hematoxylin and eosin (H&E) showing numerous macrophages containing brownish granules in their cytoplasm, magnification: **a** ×100; **b** ×400.



▶ **Video 1** Upper gastrointestinal endoscopy showing regions of flat yellow- and brownish-speckled pigmented mucosa in the duodenum. The biopsy showed duodenal pseudo-melanosis.

ciency cholesteryl ester storage disease (CESD) was confirmed. LAL deficiency is a rare (orphan) autosomal recessive lysosomal lipid storage disorder caused by mutations in the LAL

gene (*LIPA*), which is characterized by the accumulation of cholesteryl esters and triglycerides [1,2]. Depending on the residual enzyme activity, two different presentations may be seen: an early-



► **Fig. 2** Histopathological image of the biopsy with Perls' staining showing that the pigment was negative for iron-containing deposits.

onset severe and lethal phenotype known as Wolman disease – absent or <1% of normal LAL activity – or a late-onset attenuated phenotype known as CESD – 1%–12% of normal LAL activity [1,2]. Over 40 *LIPA* mutations on chromosome 10q23.2-23.3 that cause CESD and Wolman disease have been identified [2,3]. There have been 135 CESD patients described in the literature [4].

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Competing interests

None

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