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 tissue activity, fibrous degeneration (grade 3), and severe steatosis with an accumulation of foam cells. Homozygosity for pSer103Arg+/0 IVS8 1G > A was found on genetic sequence analysis of the LIPA gene. Lysosomal acid lipase (LAL) deficiency cholesteryl ester storage disease (CESD) was confirmed. LAL deficiency is a rare (orphan) autosomal recessive lysosomal lipid storage disorder caused by mutations in the LIPA 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-

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

**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.
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

The authors

Pavel V. Pavlov1, Andrey P. Kiryukhin1, Alexander S. Tertychnyi2

1 Endoscopy Unit, The Second University Clinic, I.M. Sechenov First Moscow State Medical University, Sechenov University, Moscow, Russia
2 Department of Pathology, I.M. Sechenov First Moscow State Medical University, Sechenov University, Moscow, Russia

Corresponding author

Andrey P. Kiryukhin, MD, PhD
Endoscopy Unit, The Second University Clinic, I.M. Sechenov First Moscow State Medical University (Sechenov University), 1 build. 1, Pogodinskaya St., Moscow, 119435, Russia
a.p.kiryukhin@gmail.com

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