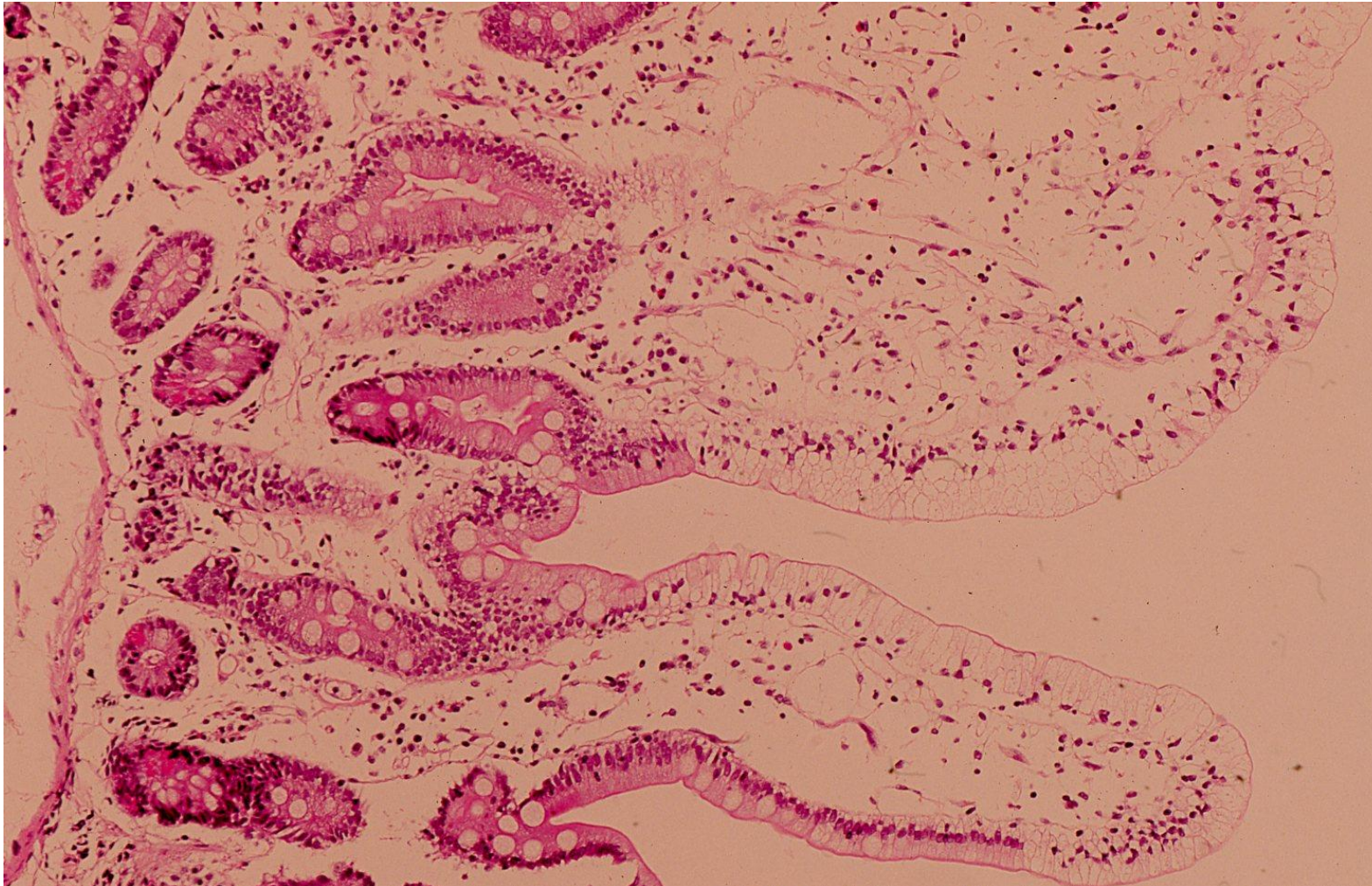


Abetalipoproteinemia

Abetalipoproteinemia is a rare autosomal recessive disorder with mutations in the MTP gene encoding a microsomal triglyceride transfer protein. The disease is marked by very low levels of plasma cholesterol, low-density lipoproteins, and very low-density lipoproteins. Symptoms include fat malabsorption (steatorrhea, vomiting, and failure to thrive in infancy), spinocerebellar degeneration, acanthocytosis in the peripheral blood and retinitis pigmentosa. The deficiency of fat-soluble vitamins progressively develops later in life. Duodenal biopsy reveals lipid accumulation in the cytoplasm of the duodenal absorptive epithelial cells. Clear cytoplasm of the villous enterocytes is quite characteristic. Duodenal biopsy from a 32-year-old female patient is displayed.

Ref.: Takahashi M, et al. Current diagnosis and management of abetalipoproteinemia. J Atheroscler Thromb 2021; 28(10): 1009-1019. doi: 10.5551/jat.RV17056



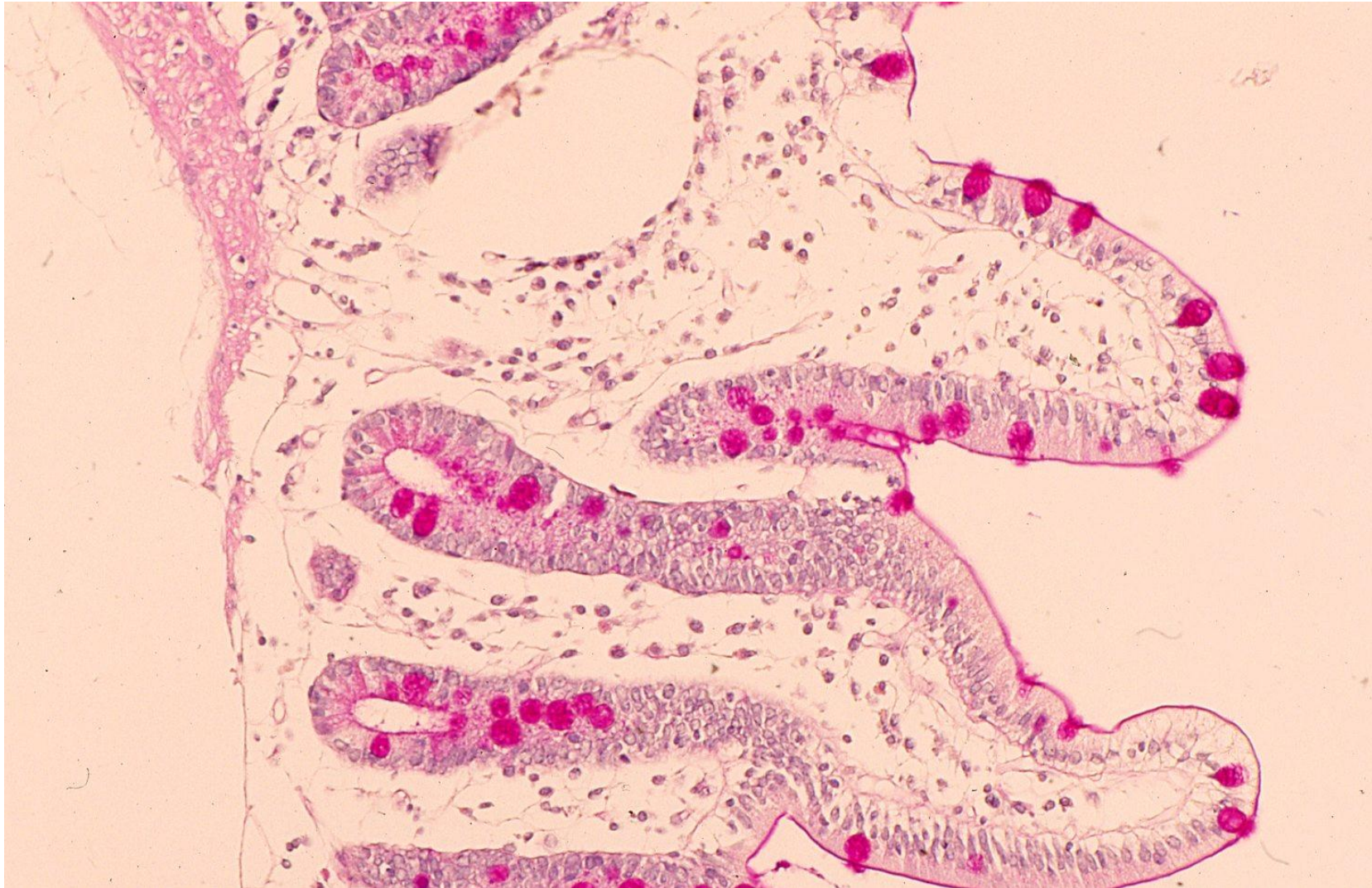
Duodenal biopsy from a 32-year-old female patient with abetalipoproteinemia, complaining of intractable steatorrheic diarrhea. Vacuolated clear cytoplasm of the villous enterocytes is evident. H&E-1



Duodenal biopsy from a 32-year-old female patient with abetalipoproteinemia, complaining of intractable steatorrheic diarrhea. Vacuolated clear cytoplasm of the villous enterocytes is evident. H&E-2



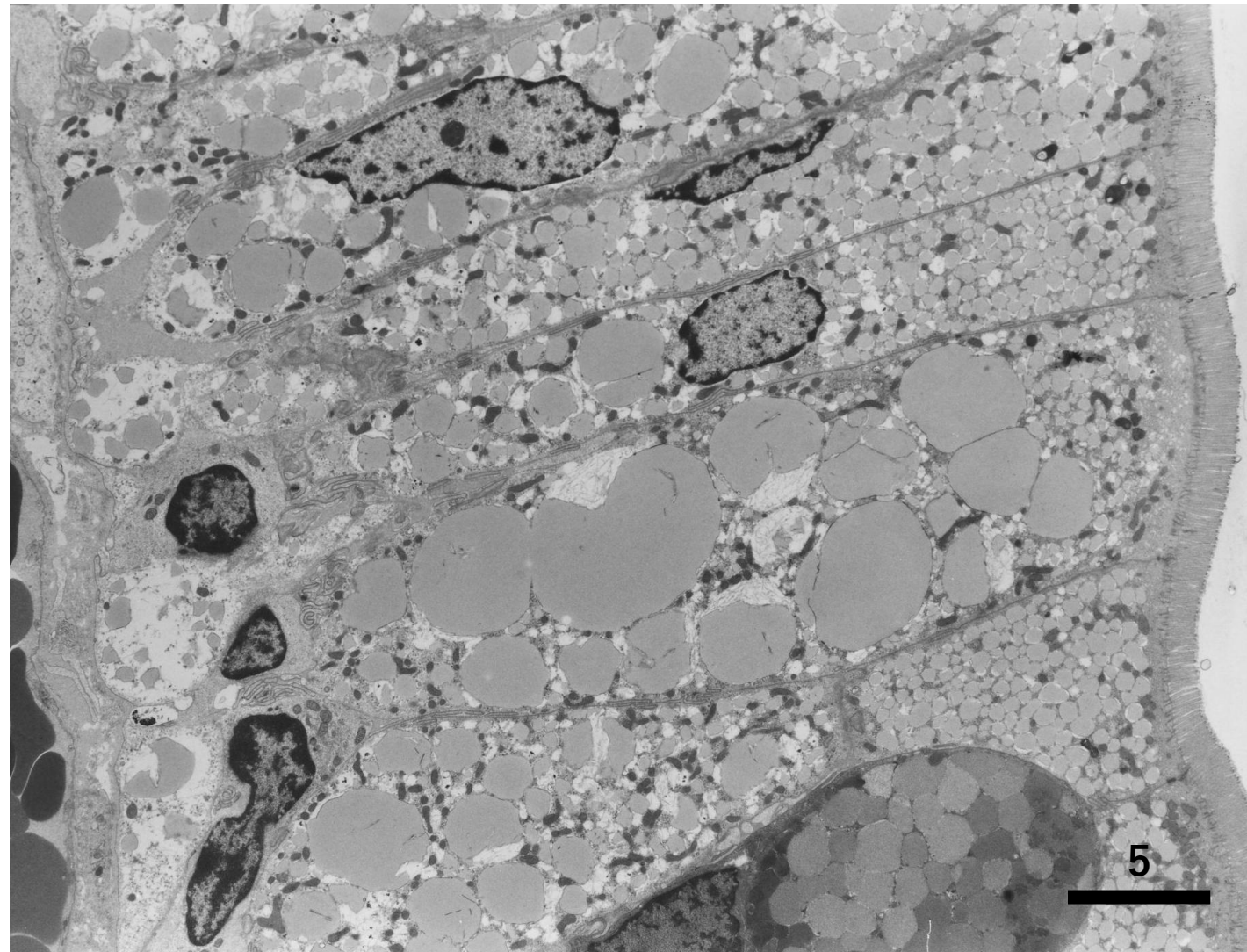
Duodenal biopsy from a 32-year-old female patient with abetalipoproteinemia, complaining of intractable steatorrheic diarrhea. Vacuolated clear cytoplasm of the villous enterocytes is evident. H&E-3



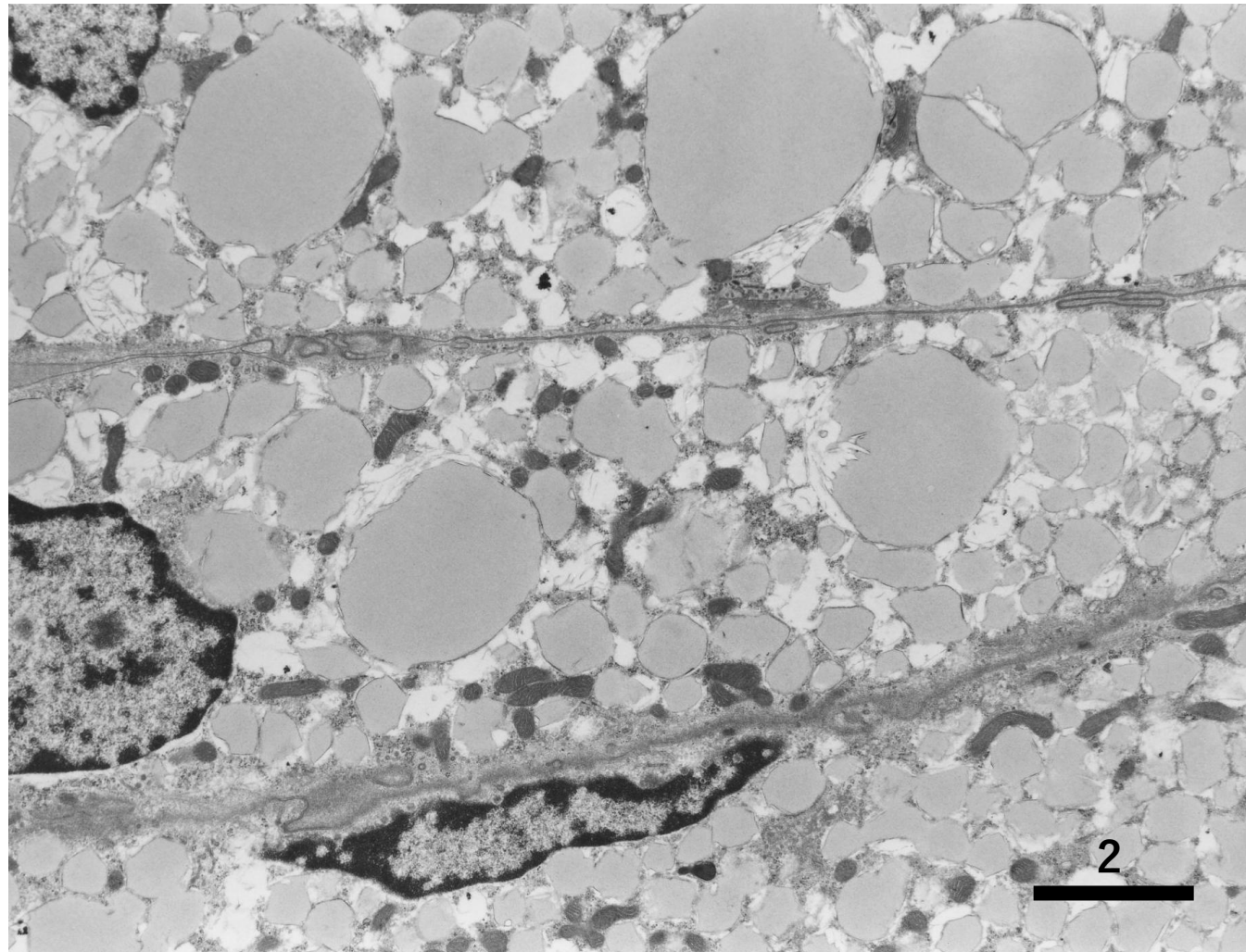
Duodenal biopsy from a 32-year-old female patient with abetalipoproteinemia, complaining of intractable steatorrheic diarrhea. Vacuolated clear cytoplasm of the villous enterocytes is negative with PAS reaction. PAS-1



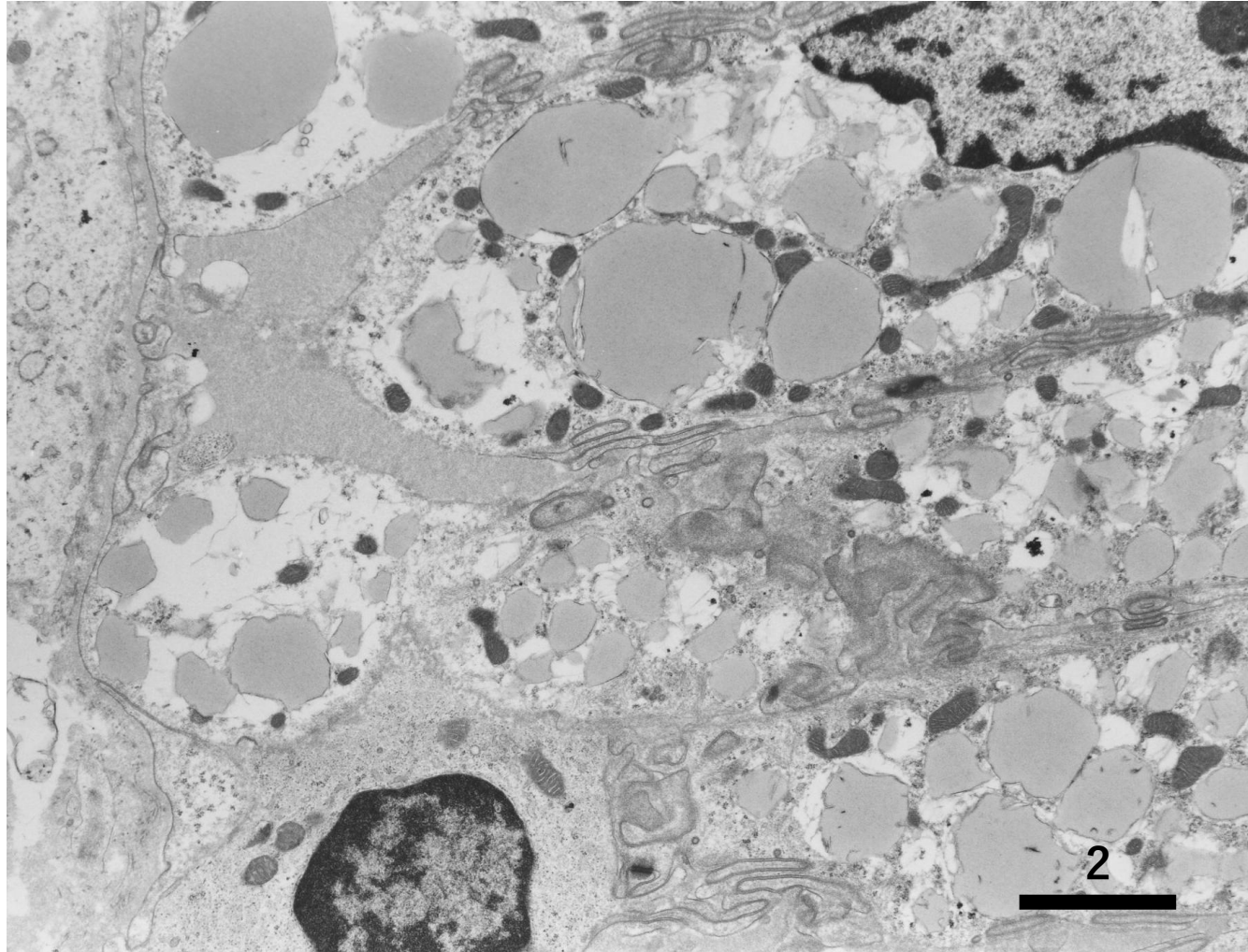
Duodenal biopsy from a 32-year-old female patient with abetalipoproteinemia, complaining of intractable steatorrheic diarrhea. Vacuolated clear cytoplasm of the villous enterocytes is negative with PAS reaction. PAS-2



EM features of the duodenal villous epithelial cells sampled from a 32-year-old female patient with abetalipoproteinemia. Lipid vacuoles are retained in the cytoplasm. EM-1



EM features of the duodenal villous epithelial cells sampled from a 32-year-old female patient with abetalipoproteinemia. Lipid vacuoles are retained in the cytoplasm. EM-2



EM features of the duodenal villous epithelial cells sampled from a 32-year-old female patient with abetalipoproteinemia. Lipid vacuoles are retained in the cytoplasm. EM-3