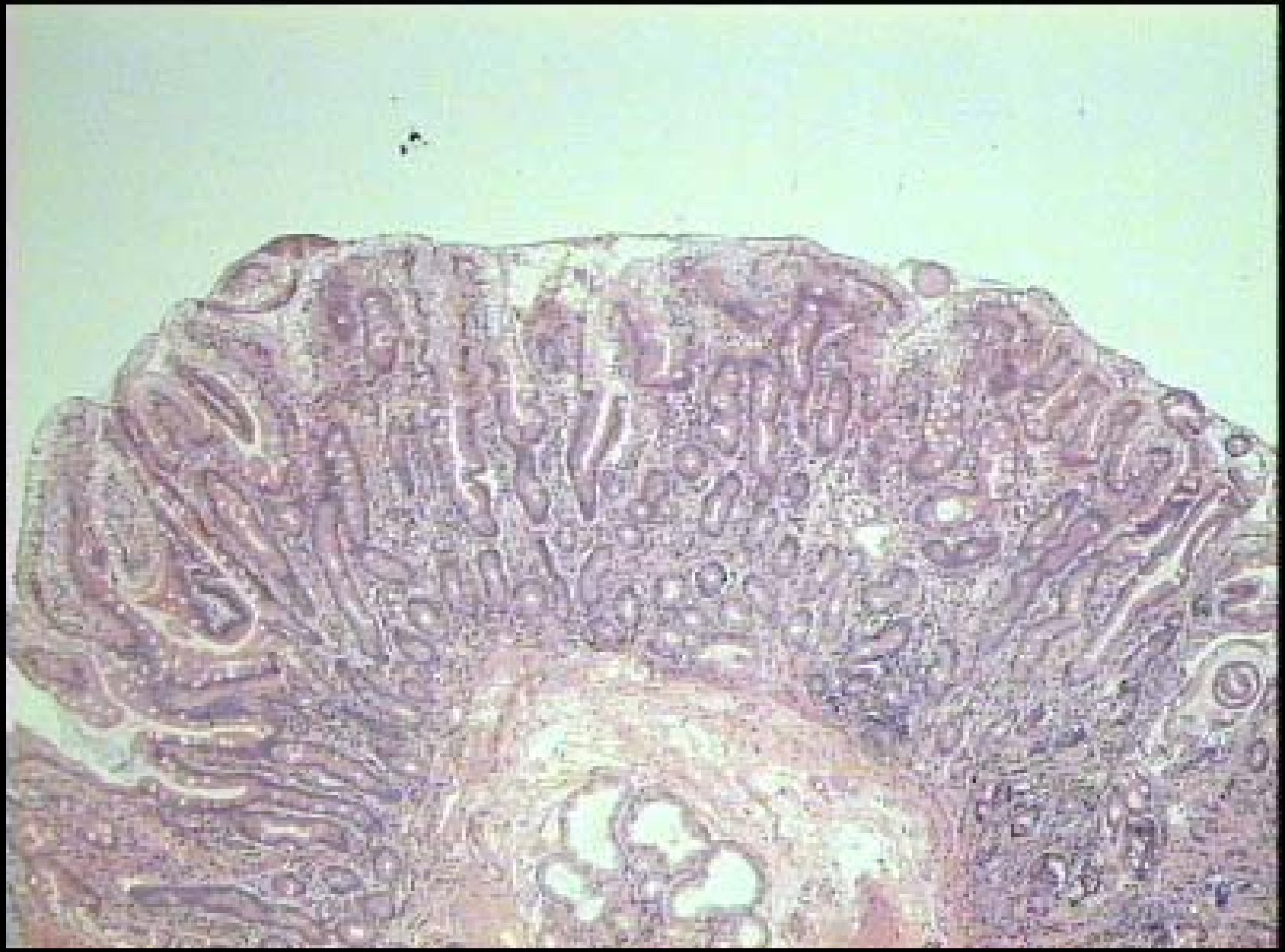
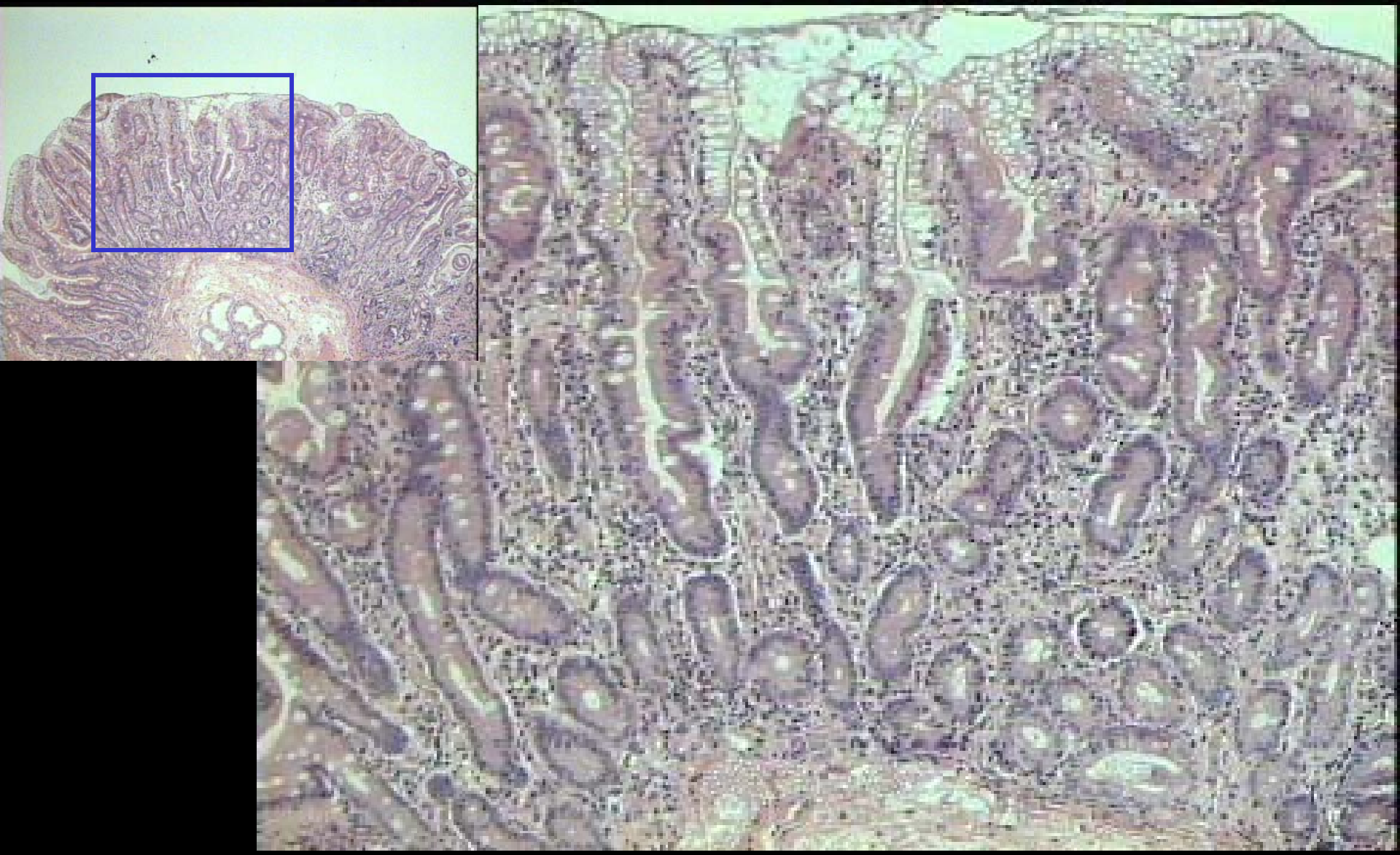
HDF Case 996741

- 31 y.o. female with a malabsorption syndrome. Duodenal mucosa is pale looking in endoscopy, with a mosaic pattern.
- Endocopic biopsy.



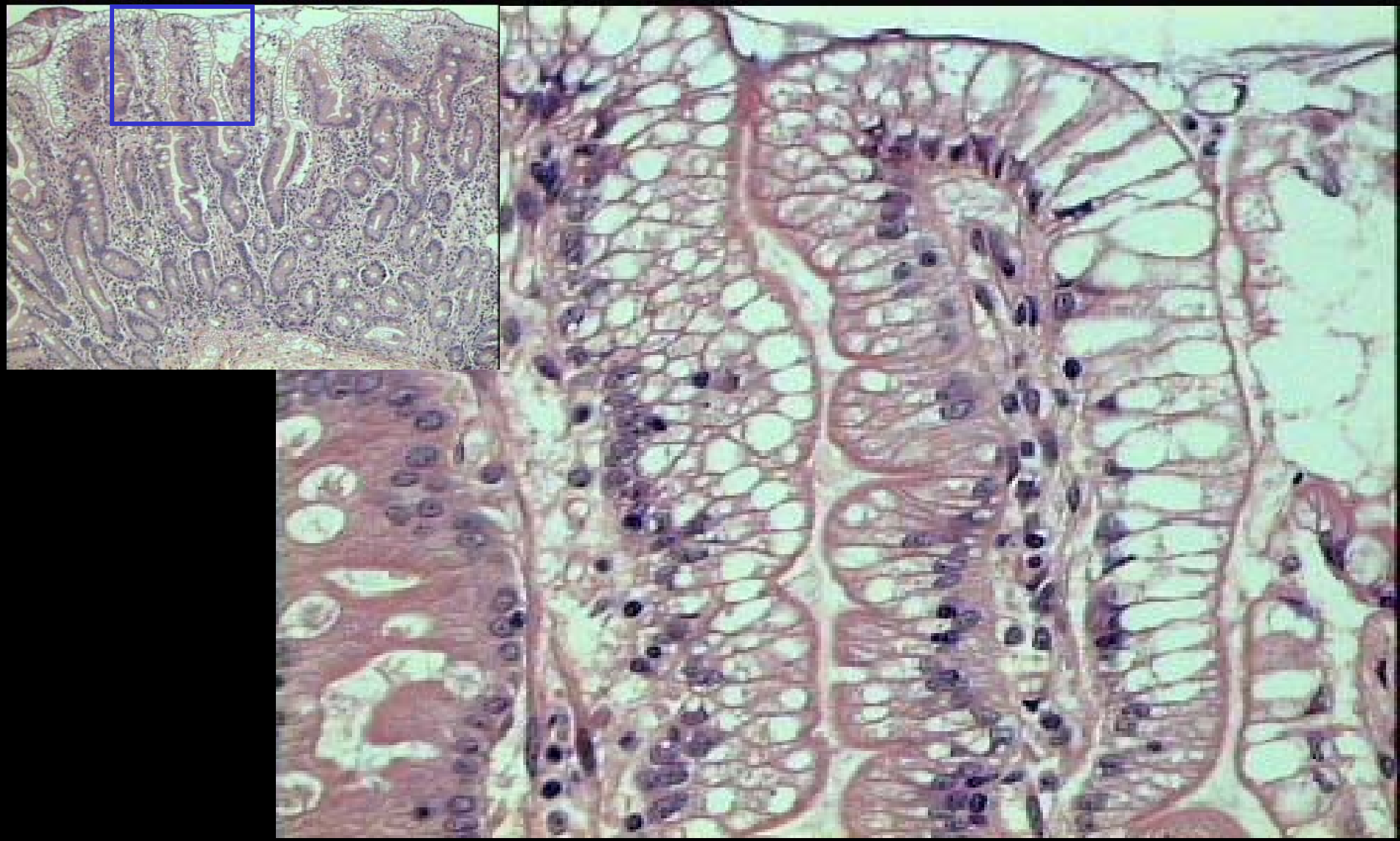
- Scanning view: Duodenal villous pattern preserved.



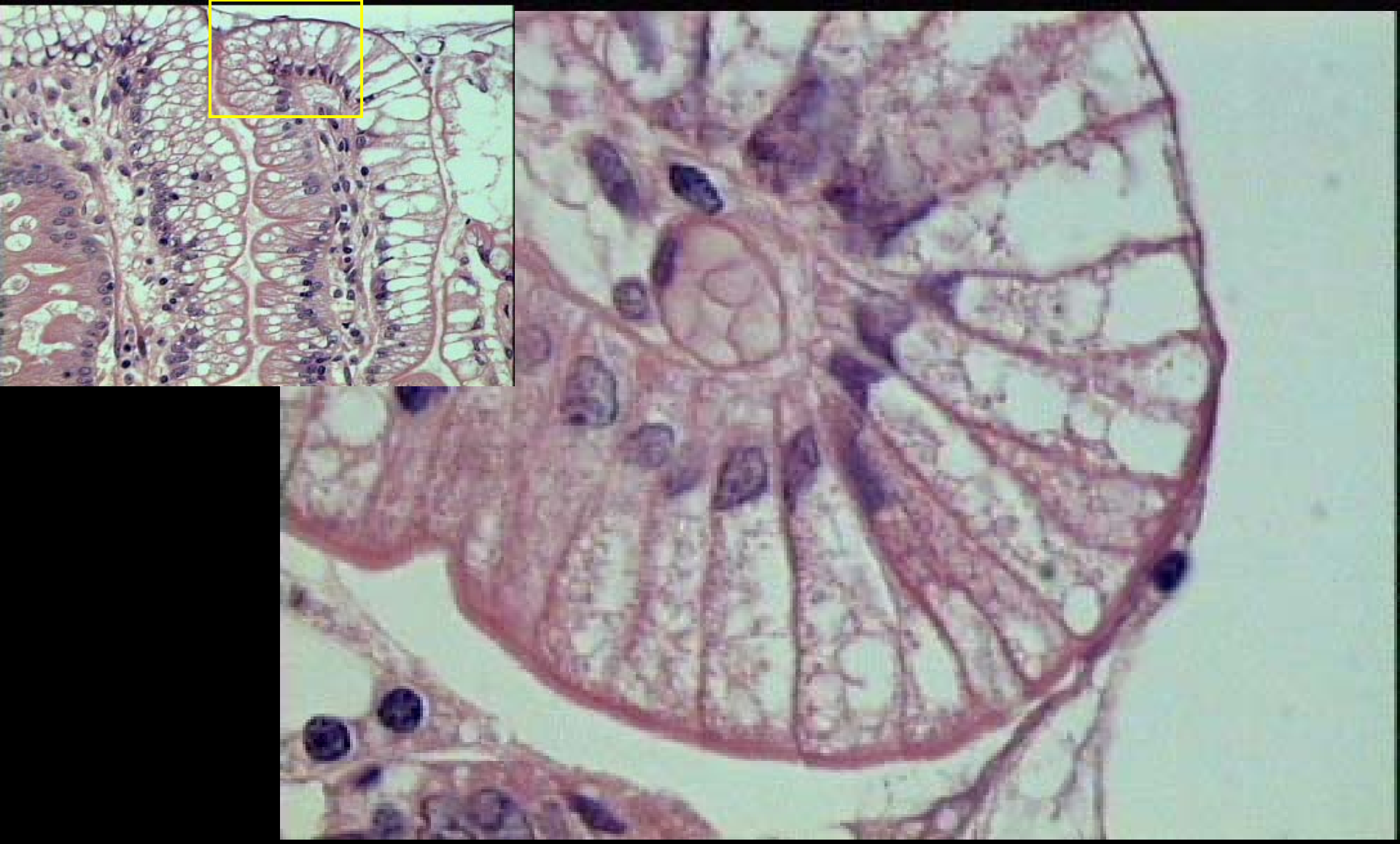
- Low power view, villi and crypts are regular.

MAIN MENU

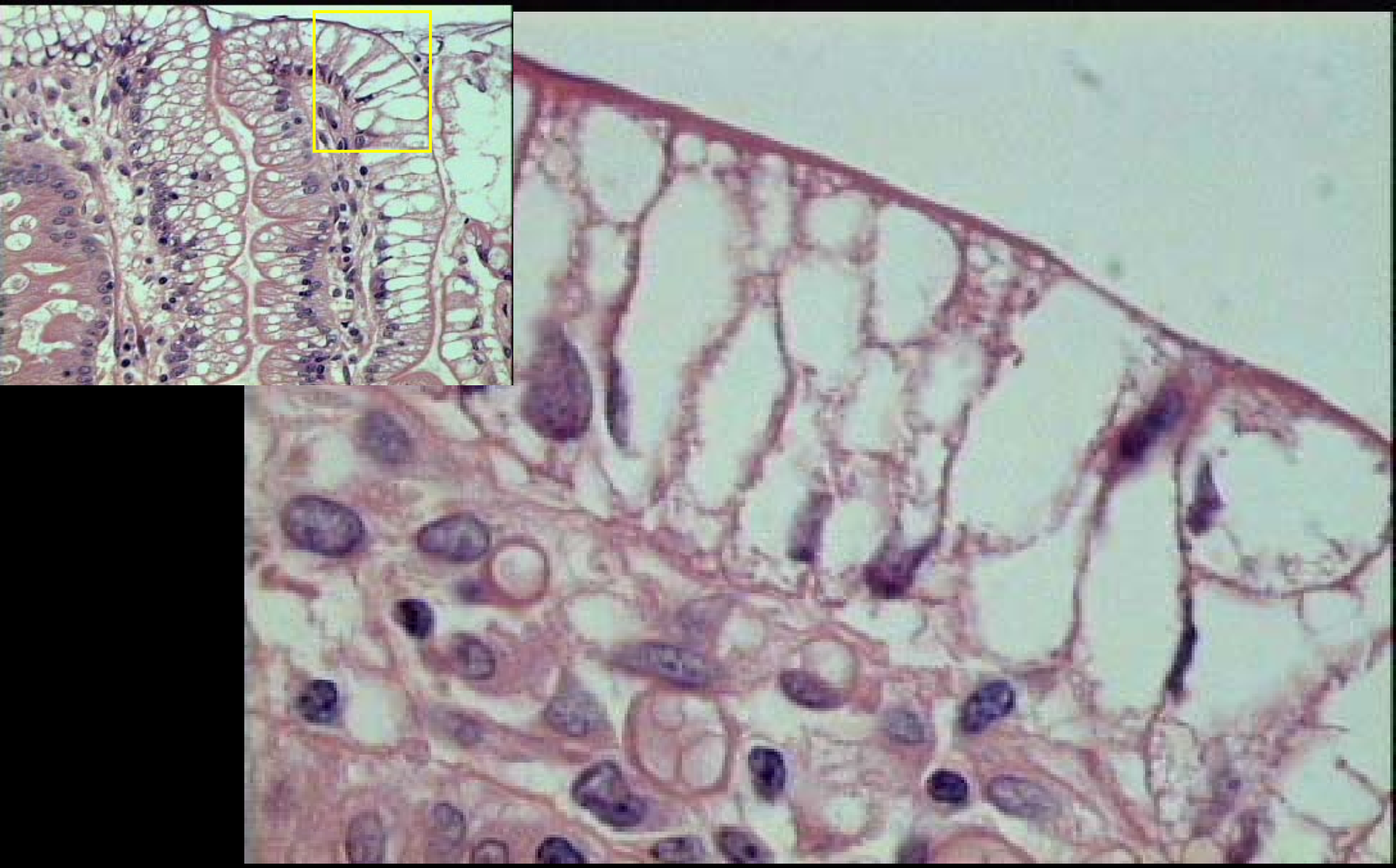
QUIT



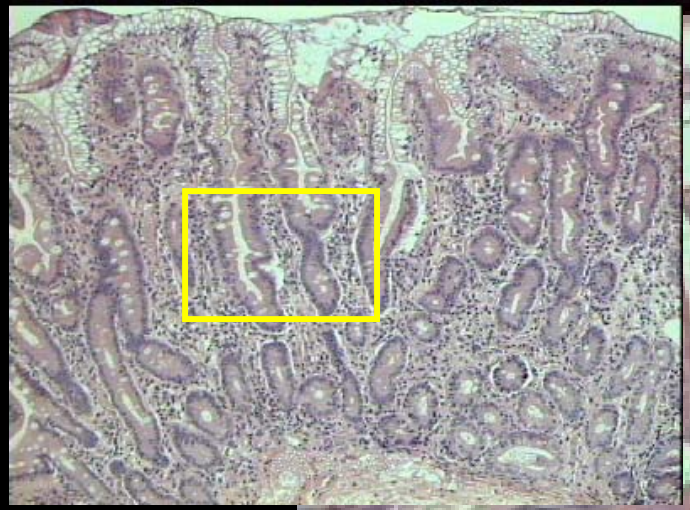
- Tip of villi, are lined by a columnar epithelium, having a clear cytoplasm.



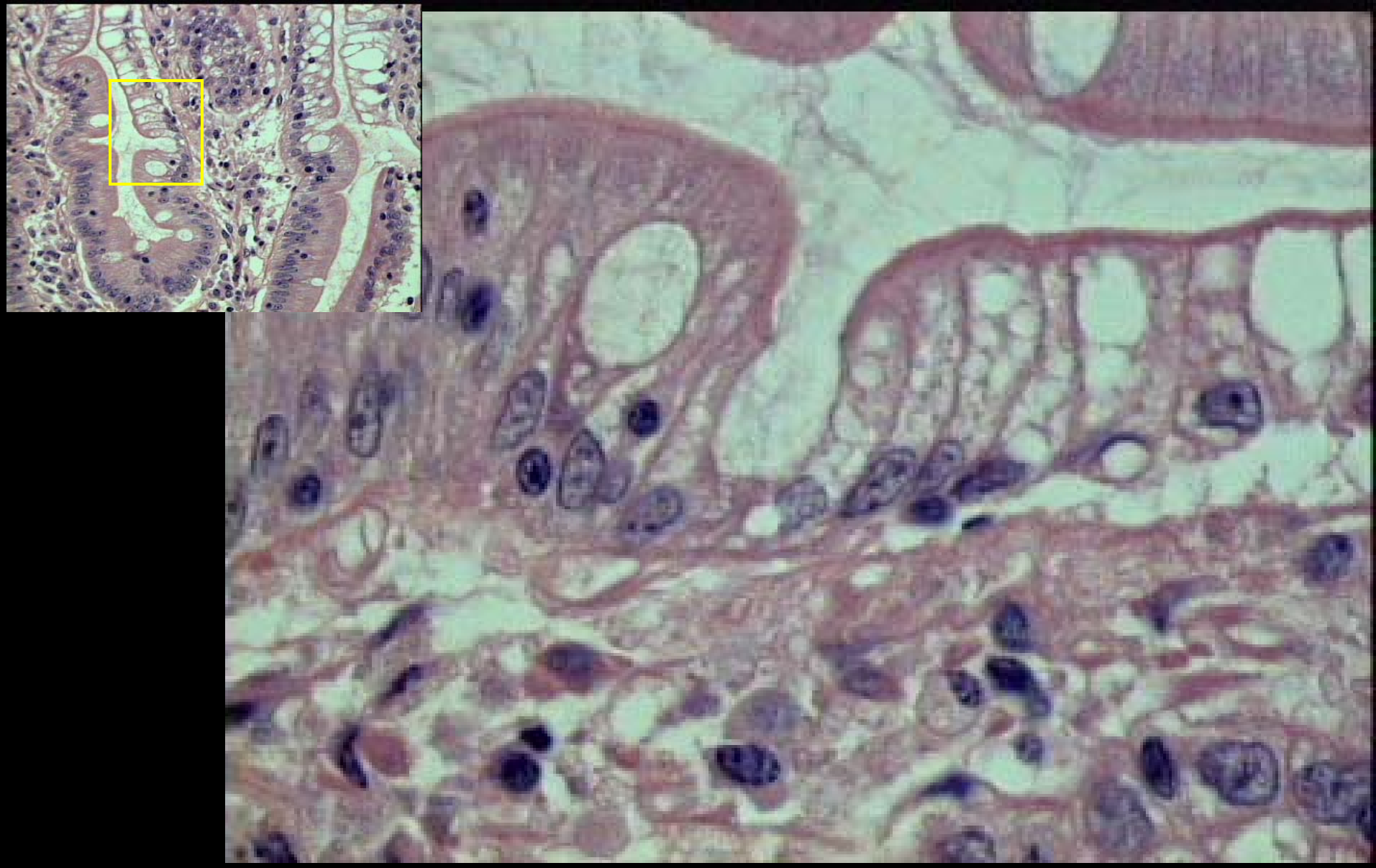
- High power view, enterocytes are stuffed with clear vacuoles.



- Closer view of the intestinal epithelium



- The epithelium gradually becomes normal towards the crypts.



- Closer view on the previous field: clear cytoplasmic vacuoles, are negative to special stains in the processed tissues.

DIAGNOSIS :

- Enterocytic lesions with preserved intestinal villous architecture consistent with abetalipoproteinemia.

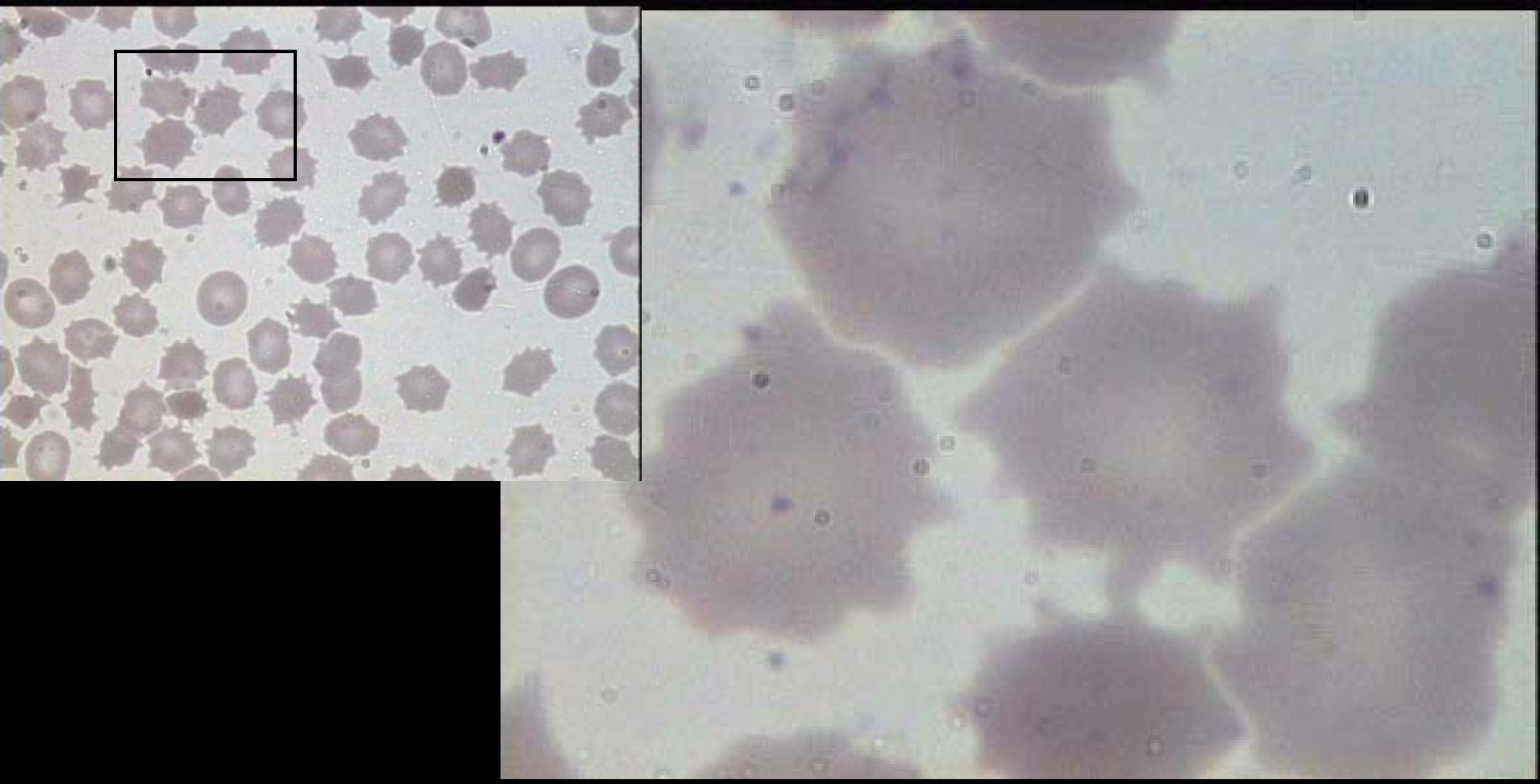
Abetalipoproteinemia

- is a rare group of inherited disorders of childhood characterized by an absence or a deficiency of apoprotein B, which is required to synthesize and transport very low density lipoproteins and chylomicrons. This results in normal absorptive cell uptake of fatty acids but inability to synthesize them into chylomicrons. The absorptive cells become stuffed with fat . Ultimately, fat is unable to enter the absorptive cells and steatorrhea ensues.

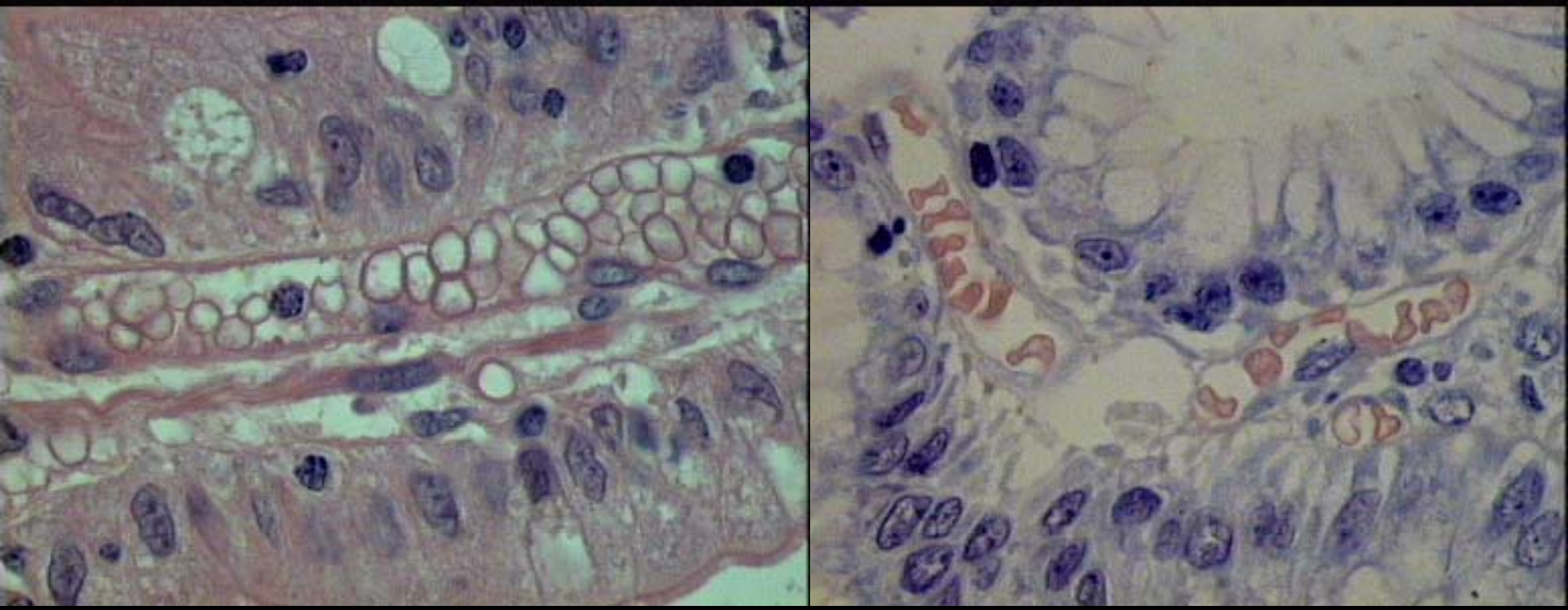
PATHOGENESIS

- The mechanism of chylomicron formation and secretion in the gut is complex. Following lipolysis of dietary fat in the gut lumen, there is passive diffusion of fatty acids into the endoplasmic reticulum of enterocytes. Here chylomicron assembly occurs in the presence of phospholipids, cholesterol, and a number of apoproteins. The chylomicrons are then assembled in the Golgi apparatus and extruded by exocytosis into the intercellular spaces or intestinal lymphatics. To date, abnormalities have been described in apoprotein production and from a defect in the final assembly of chylomicrons in the mechanism of exocytosis.
- The small bowel mucosa has a characteristic appearance. The upper two-thirds of the villi contain epithelial cells that appear vacuolated and clear-staining.

- There is one case report of a patient with a generalized neutral lipid disorder in whom there were jejunal changes similar to abetalipoproteinemia. However, this patient also had lipid accumulation in other parts of the gastrointestinal tract, such as the gastric glands, and in many other tissues, such as liver, striated muscle, and granulocytes.
- Another potential source of diagnostic confusion arises when an infant with suspected abetalipoproteinemia has a biopsy taken shortly after feeding. In the postfeeding state with a meal containing fat, vacuoles of absorbed fat may normally be seen in villous tip epithelium. This confusion can usually be resolved easily by taking an appropriate history and by avoiding feeding the infant for at least 6 hours prior to biopsy. Vacuolar change is also found in severe mucosal lesions, for example, celiac sprue. In these cases, the morphologic features of a severe mucosal lesion are clearly seen and should not pose any diagnostic problem.



- The other manifestations of this disorder are retinitis pigmentosa, **acanthocytosis** of red blood cells, ataxia, and mental retardation. Acanthocytosis noted in smear of the peripheral blood.



- Variation in size and form of RBC in the vessels noted in the duodenal biopsy (left) are due to fixative agents, are also observed in biopsies without acanthocytosis (right, Giemsa stain)