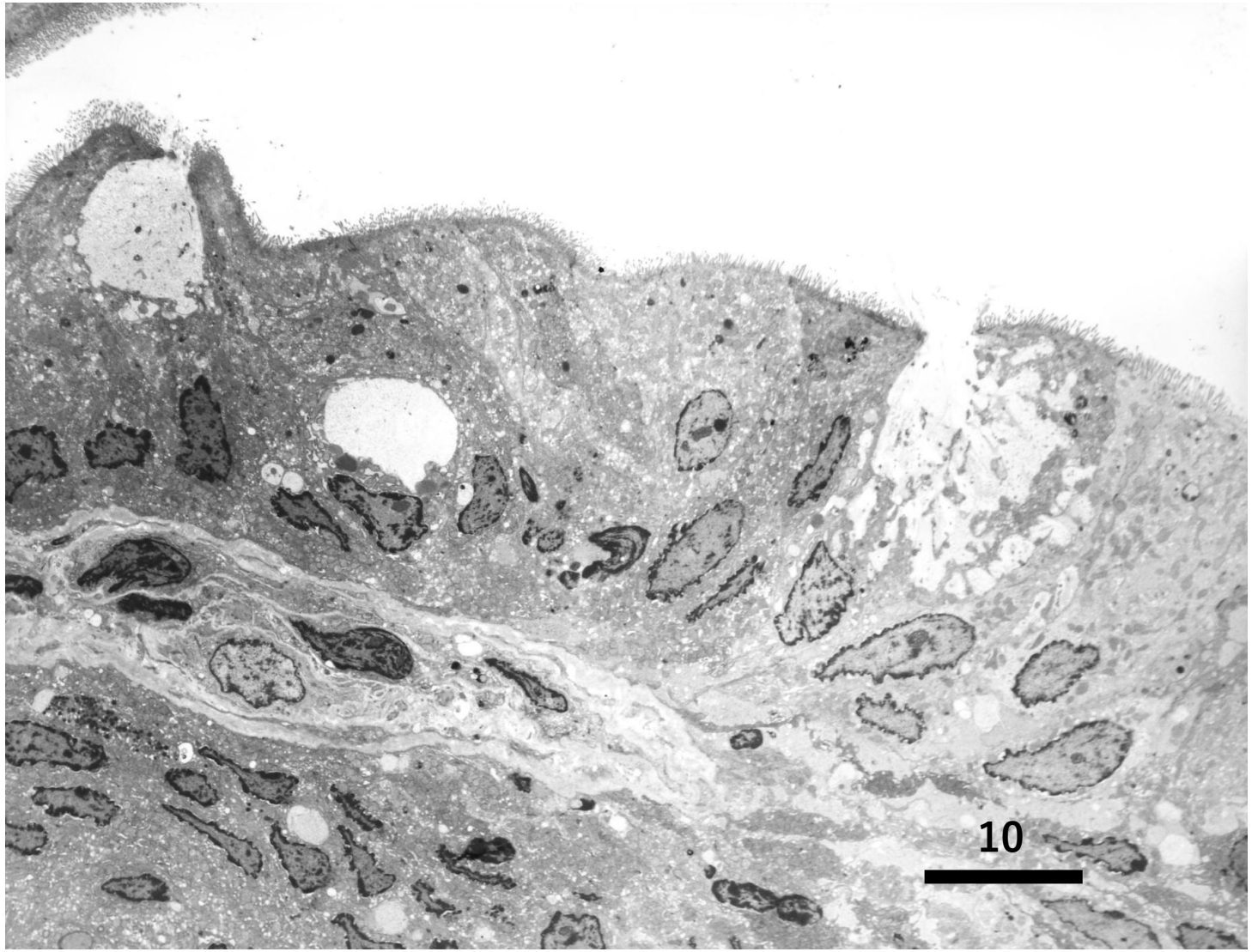
Microvillus inclusion disease

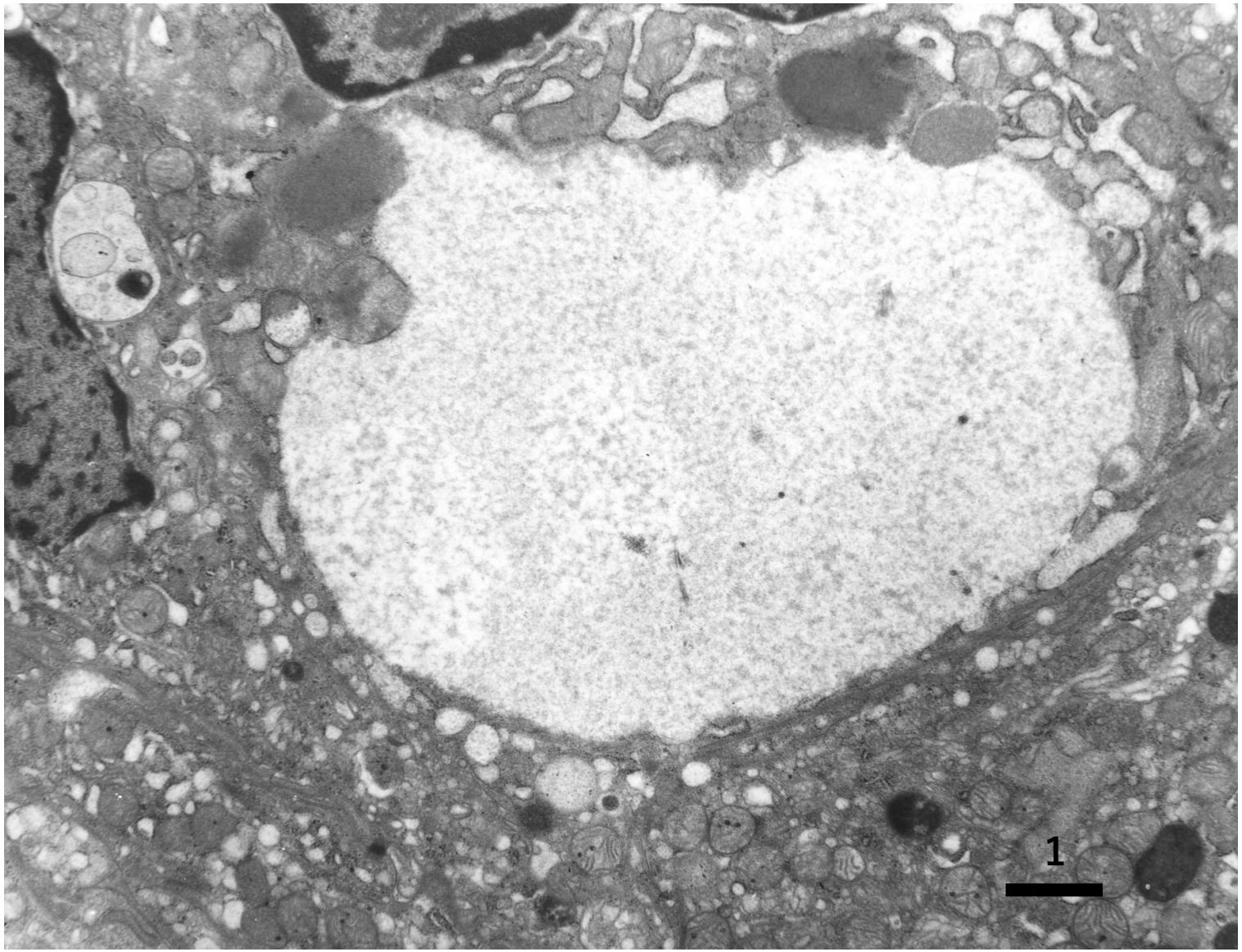
Microvillus inclusion disease (Davidson's disease) is a genetic disorder of the small intestine, inherited with an autosomal recessive trait, involving the MYO5B gene. Intractable diarrhea starts in the first few days of life, resulting in metabolic acidosis and severe dehydration. Biopsy features resemble those of celiac sprue, but intraepithelial lymphocytic infiltration is not observed. Apical microvillus atrophy and intracytoplasmic invagination of microvilli are noted. Ultrastructural study is essential for the diagnosis.

Ref.: Halac U, et al. Microvillous inclusion disease: how to improve the prognosis of a severe congenital enterocyte disorder. J Pediatr Gastroenterol Nutr 2011; 52(4): 460-465. doi: 10.1097/MPG.0b013e3181fb4559

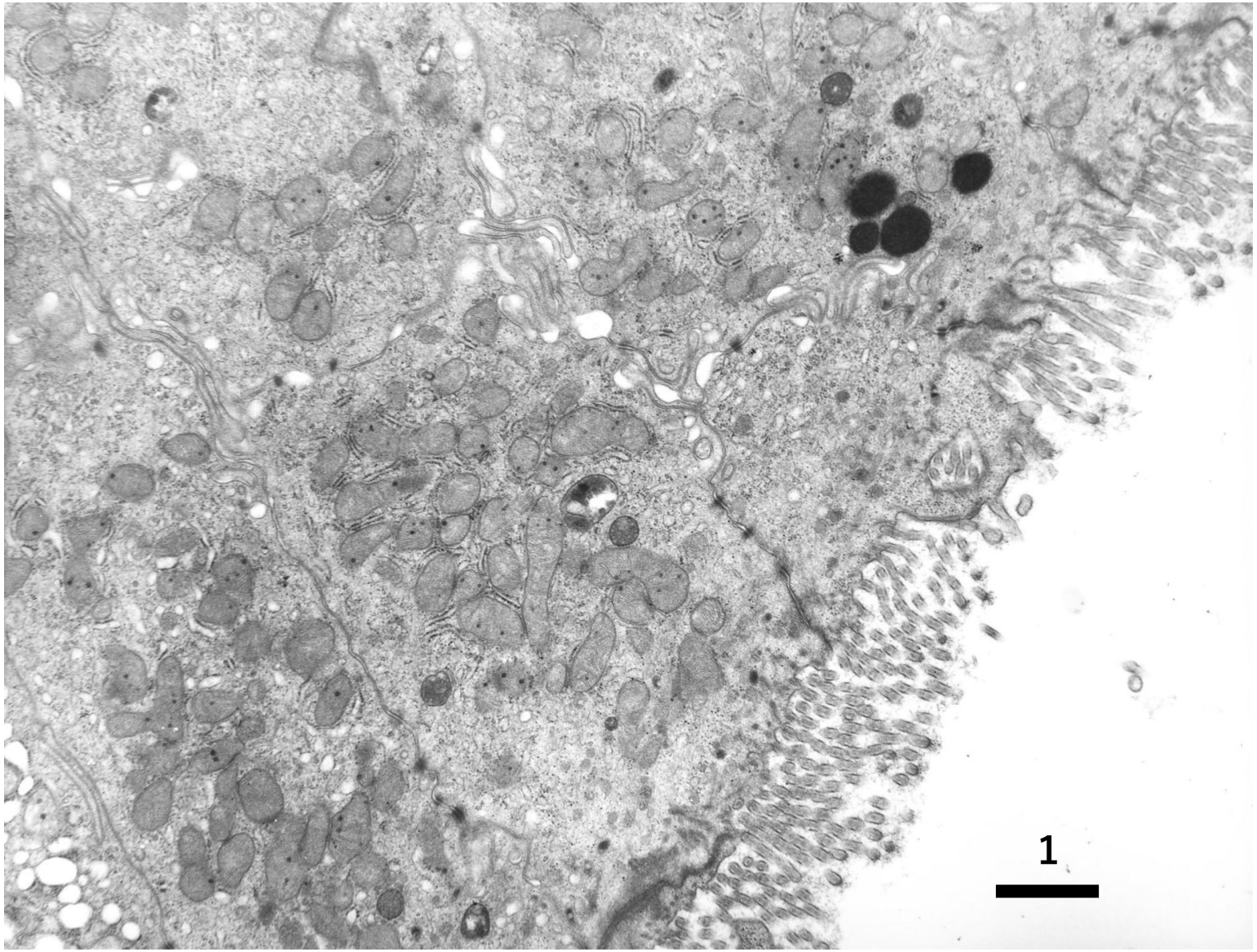
Brief clinical history:
Duodenal biopsy was taken from a 5-year-old boy. Soon after birth, he was diagnosed as familial intrahepatic cholestasis with hyperbilirubinemia and white stool. He showed growth retardation due to malabsorption syndrome. The possibility of microvillus inclusion disease was suspected. Similar clinical features are also seen in his young brother.



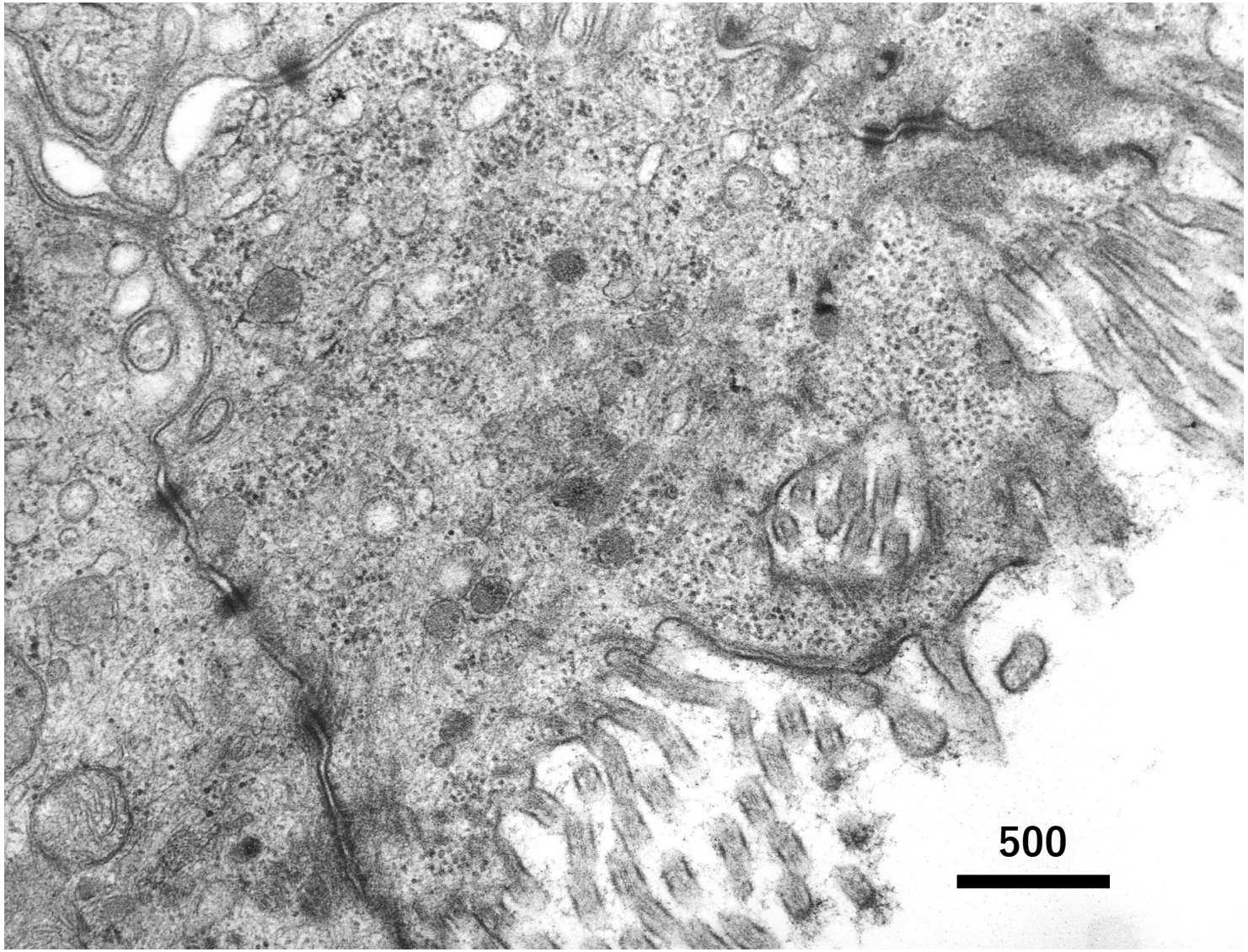
Ultrastructure of microvillus inclusion disease (duodenal biopsy from a 5-year-old boy with malabsorption syndrome). The microvilli on the apical surface are poorly developed. Cytoplasmic vacuole formation is focally associated. EM-1



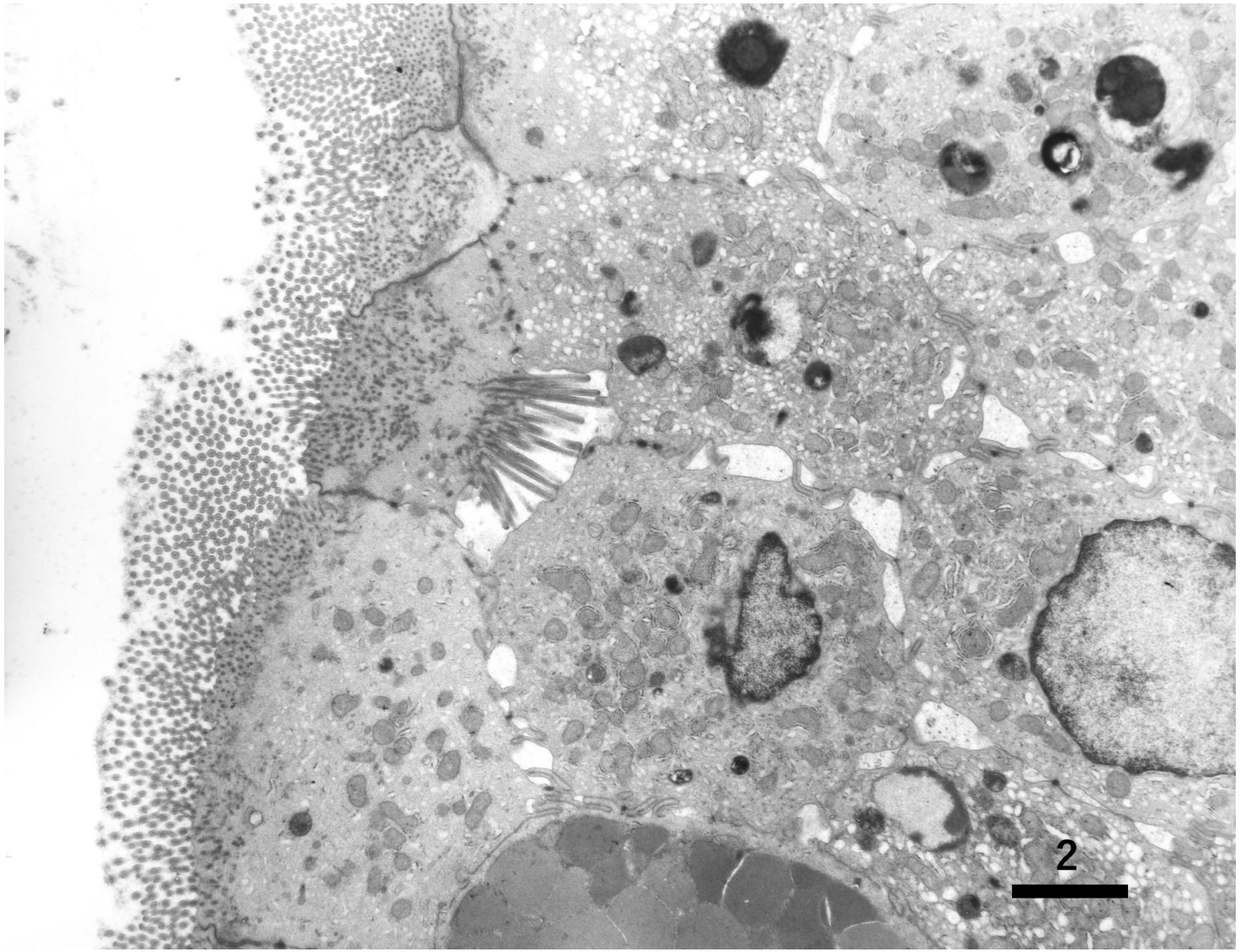
Ultrastructure of microvillus inclusion disease (duodenal biopsy from a 5-year-old boy with malabsorption syndrome). Cytoplasmic vacuole formation is focally associated in the absorptive columnar cell. EM-2



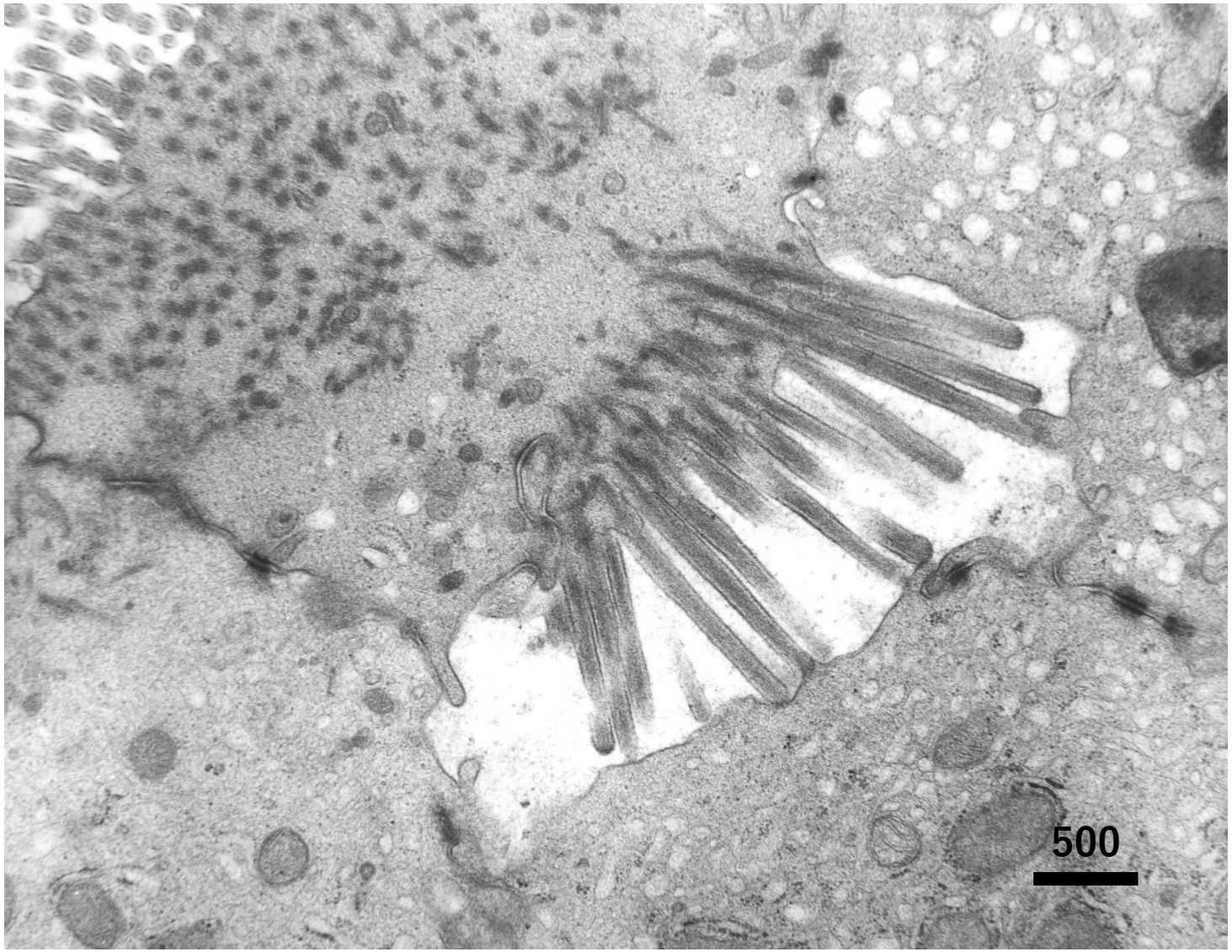
Ultrastructure of microvillus inclusion disease (duodenal biopsy from a 5-year-old boy with malabsorption syndrome). A microvillus inclusion is focally observed just beneath the apical surface. EM-3



Ultrastructure of microvillus inclusion disease (duodenal biopsy from a 5-year-old boy with malabsorption syndrome). A microvillus inclusion is focally observed just beneath the apical surface. EM-4



Ultrastructure of microvillus inclusion disease (duodenal biopsy from a 5-year-old boy with malabsorption syndrome). A microvillus inclusion (inversion) is focally observed just beneath the apical surface. EM-5



Ultrastructure of microvillus inclusion disease (duodenal biopsy from a 5-year-old boy with malabsorption syndrome). A microvillus inclusion (inversion) is focally observed just beneath the apical surface. EM-6