Chronic intestinal pseudo-obstruction in Leigh syndrome (mitochondriopathy)

Chronic intestinal pseudo-obstruction (CIPO) is characterized by signs and symptoms of intestinal obstruction without anatomic lesions. Primary (congenital) CIPO is subclassified into myopathic and neuropathic types. There are intrinsic abnormalities of the smooth muscle or peripheral nerve in the gastrointestinal tract, resulting in weakened or unsynchronized peristalsis. Secondary CIPO is usually caused by such underlying systemic, metabolic or organic conditions as autoimmune disorders, endocrine disorders, neurological disorders, malignancies and certain infections. Mitochondriopathy also causes secondary CIPO.

Leigh syndrome (subacute necrotizing encephalomyelopathy) is a neurodegenerative disease manifesting in infancy or early childhood. On MRI, symmetrical lesions are seen in the basal ganglia or brain stem. Clinically, rapid deterioration of cognitive and motor functions is noted. Leigh syndrome belongs to mitochondriopathy with mutations in mitochondrial and nuclear genes encoding components of the oxidative phosphorylation system. Dysfunctions in pyruvate dehydrogenase complex or coenzyme Q10 metabolism may also be associated. Here, a pediatric case of Leigh syndrome manifesting CIPO Is presented.

Ref.:-1 Zhu CZ, et al. Latest developments in chronic intestinal pseudo-obstruction. World J Clin Cases 2020; 8(23): 5852-5865. doi: 10.12998/wjcc.v8.i23.5852

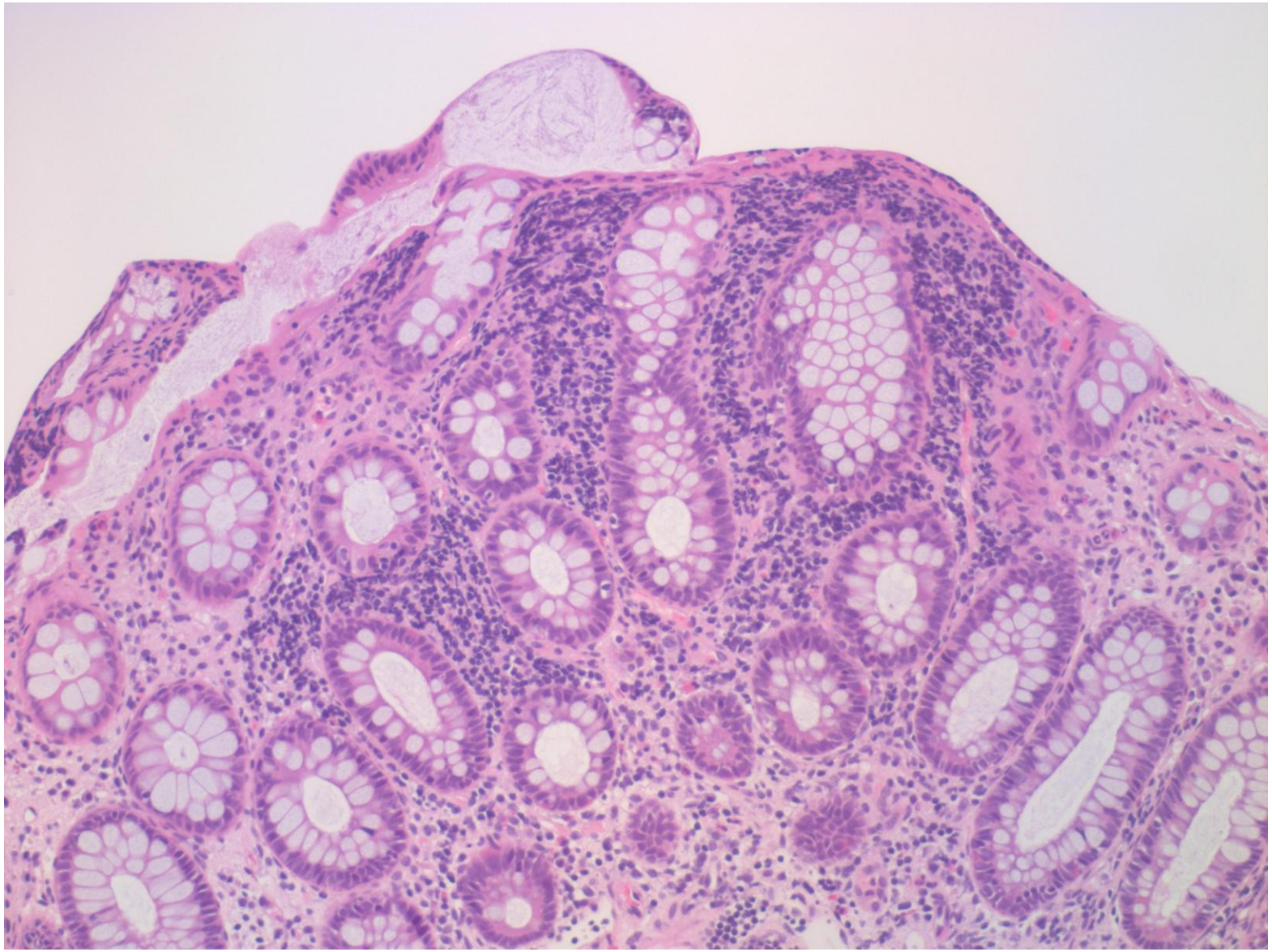
Ref.-2: Chang X, et al. A meta-analysis and systematic review of Leigh syndrome: clinical manifestations, respiratory chain enzyme complex deficiency, and gene mutations. Medicine (Baltimore) 2020; 99(5): e18634. doi: 10.1097/MD.0000000000018634

Case presentation of Leigh syndrome manifesting chronic intestinal pseudo-obstruction

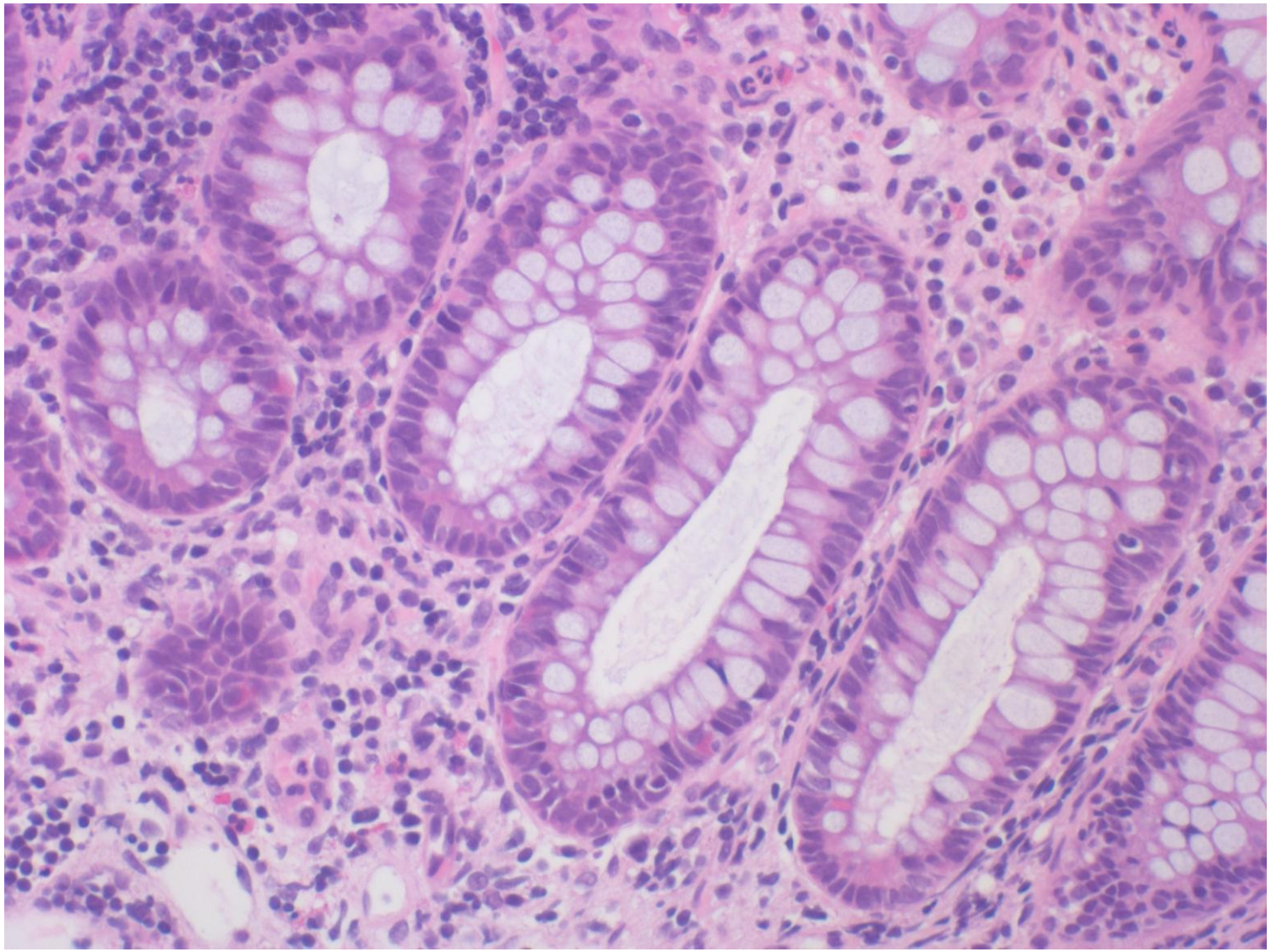
A 3-year-old girl acutely manifested ileus after persistent melena for 2 months. At emergency laparotomy, no anatomical obstructive lesion was found in the abdominal cavity. The diagnosis of chronic intestinal pseudo-obstruction was made.

She had been diagnosed as Leigh syndrome at the age of 1, based on developmental retardation with lactic acidosis. Mitochondrial DNA mutation (MT-ND3 variant: m.10158.T>C) was identified.

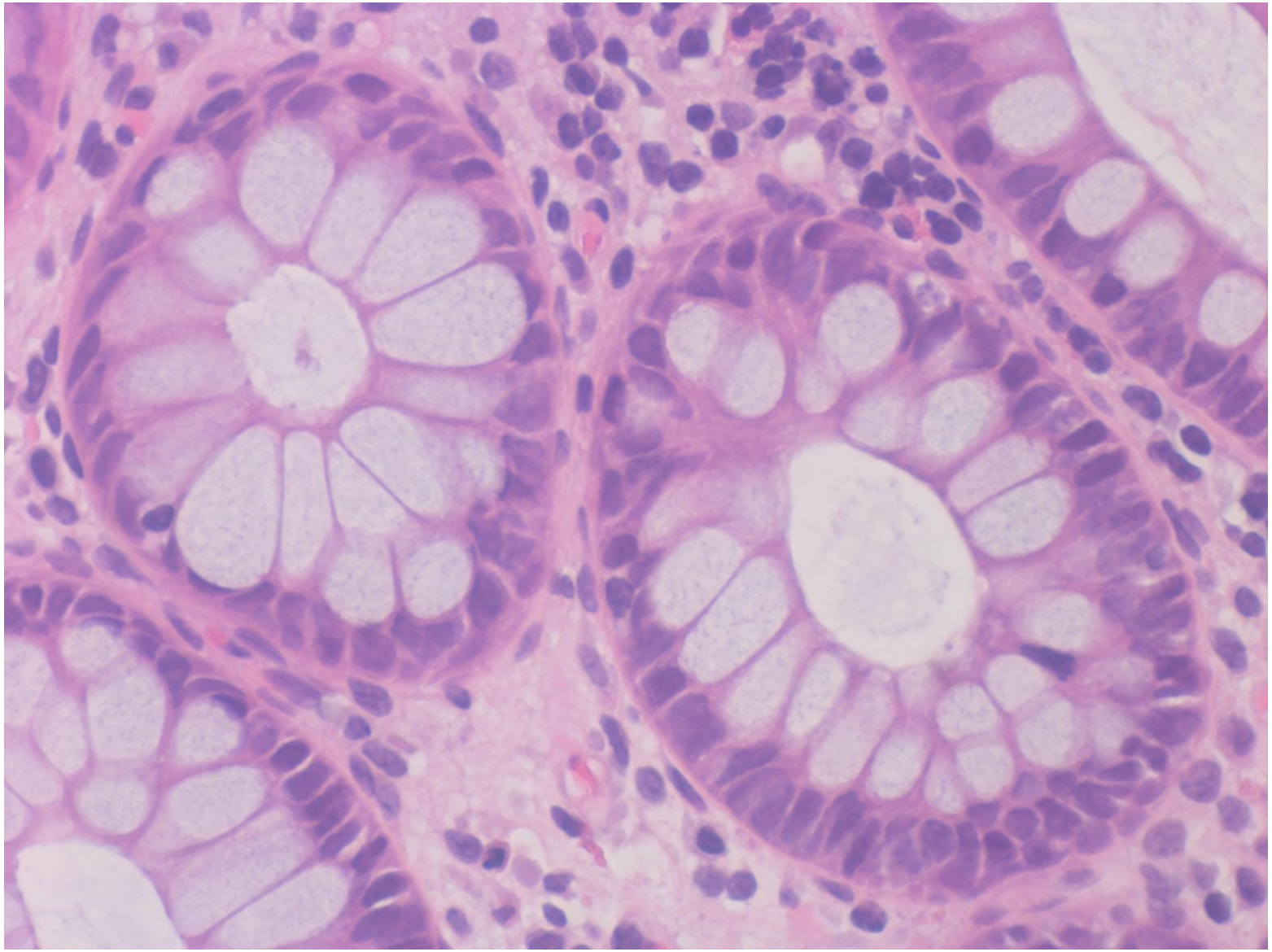
Endoscopic colonic biopsy was taken for evaluating mitochondrial abnormality at the EM level.



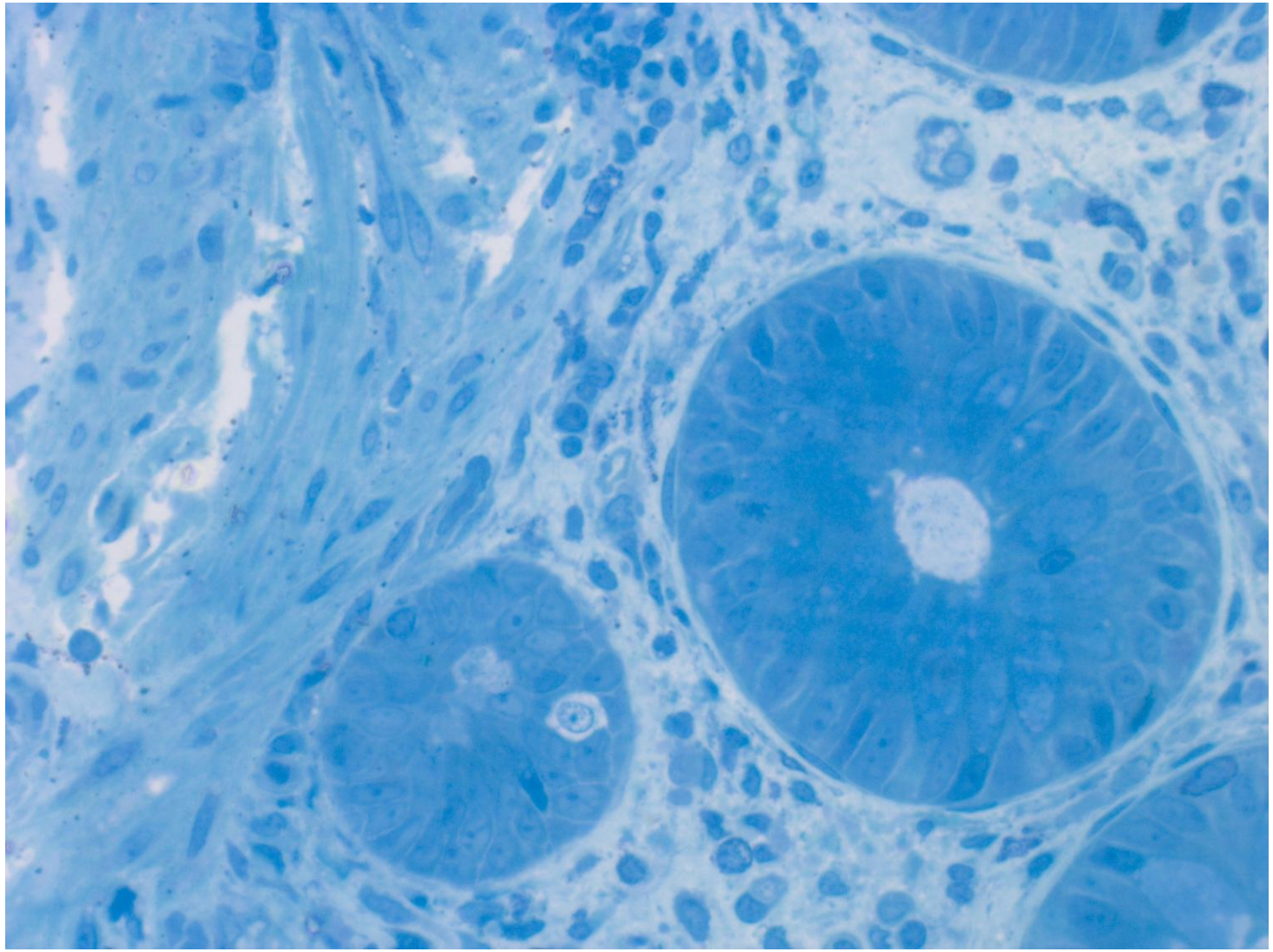
Colon biopsy from a 3-year-old girl with Leigh syndrome shows unremarkable change light microscopically. H&E-1



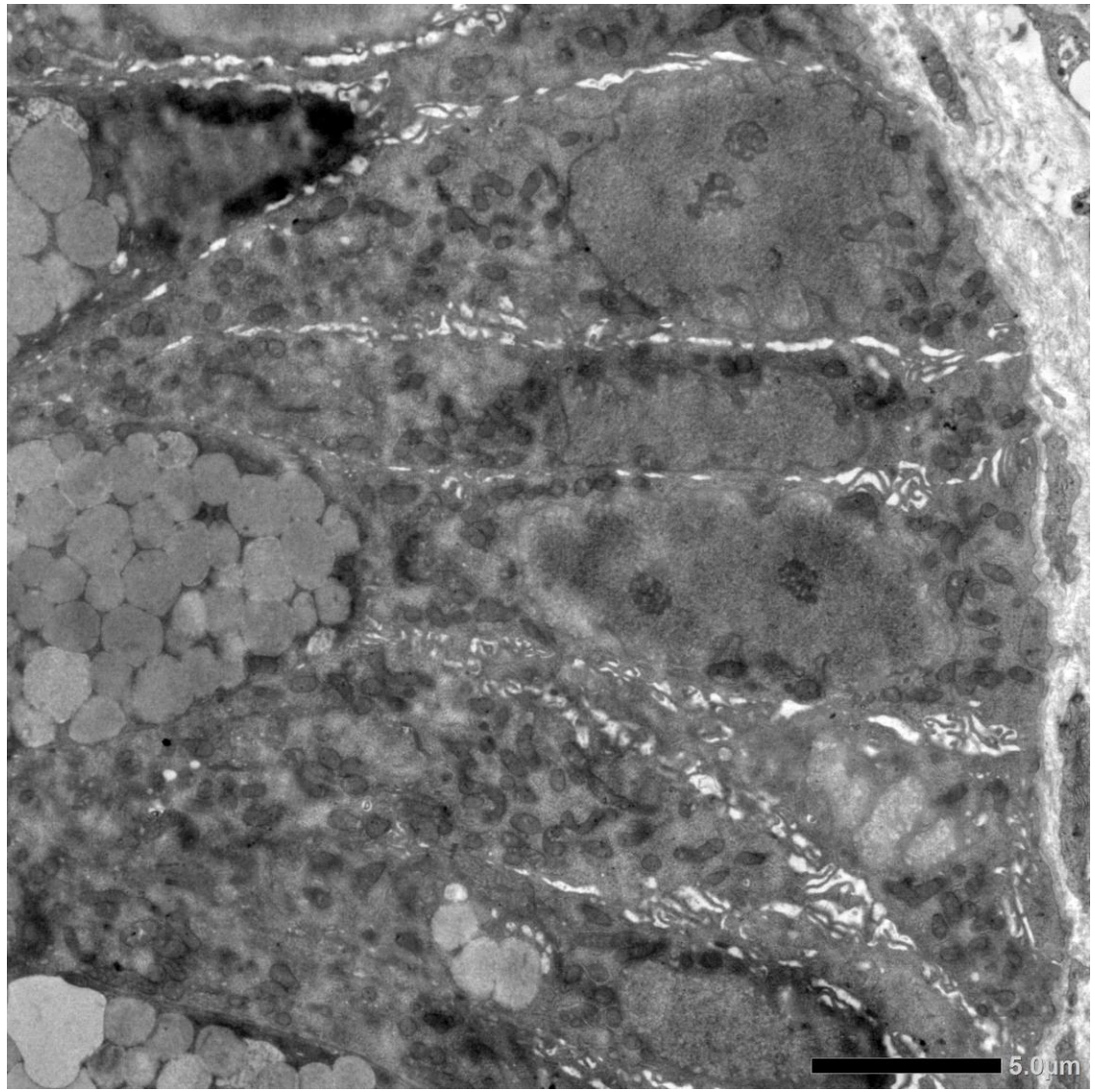
Chronic intestinal pseudo-obstruction complicated with Leigh syndrome in a 3-year-old girl. Biopsied colonic mucosa reveals little microscopic abnormalities. H&E-2



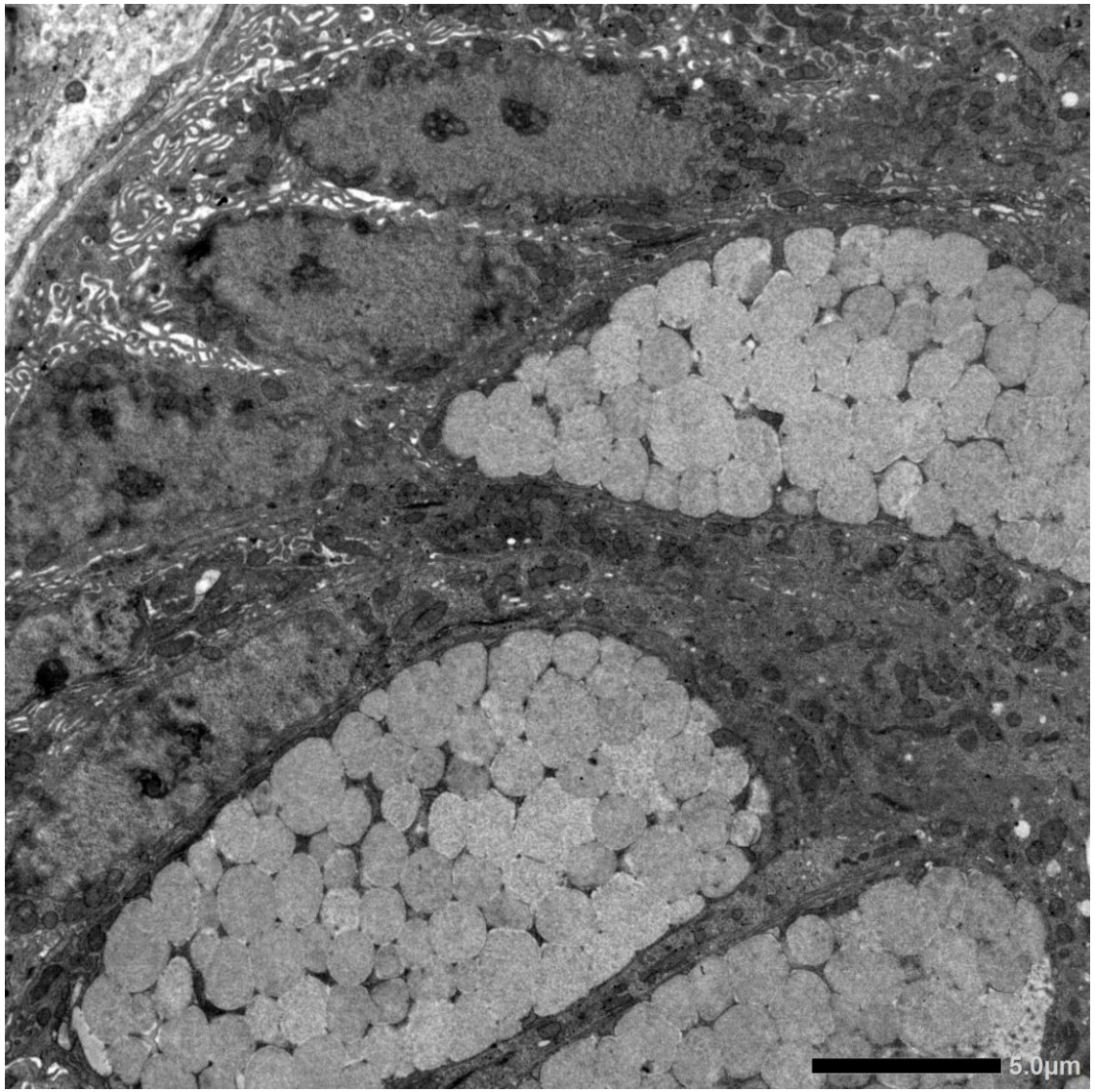
Colon biopsy from a 3-year-old girl with Leigh syndrome shows unremarkable change light microscopically. H&E-2



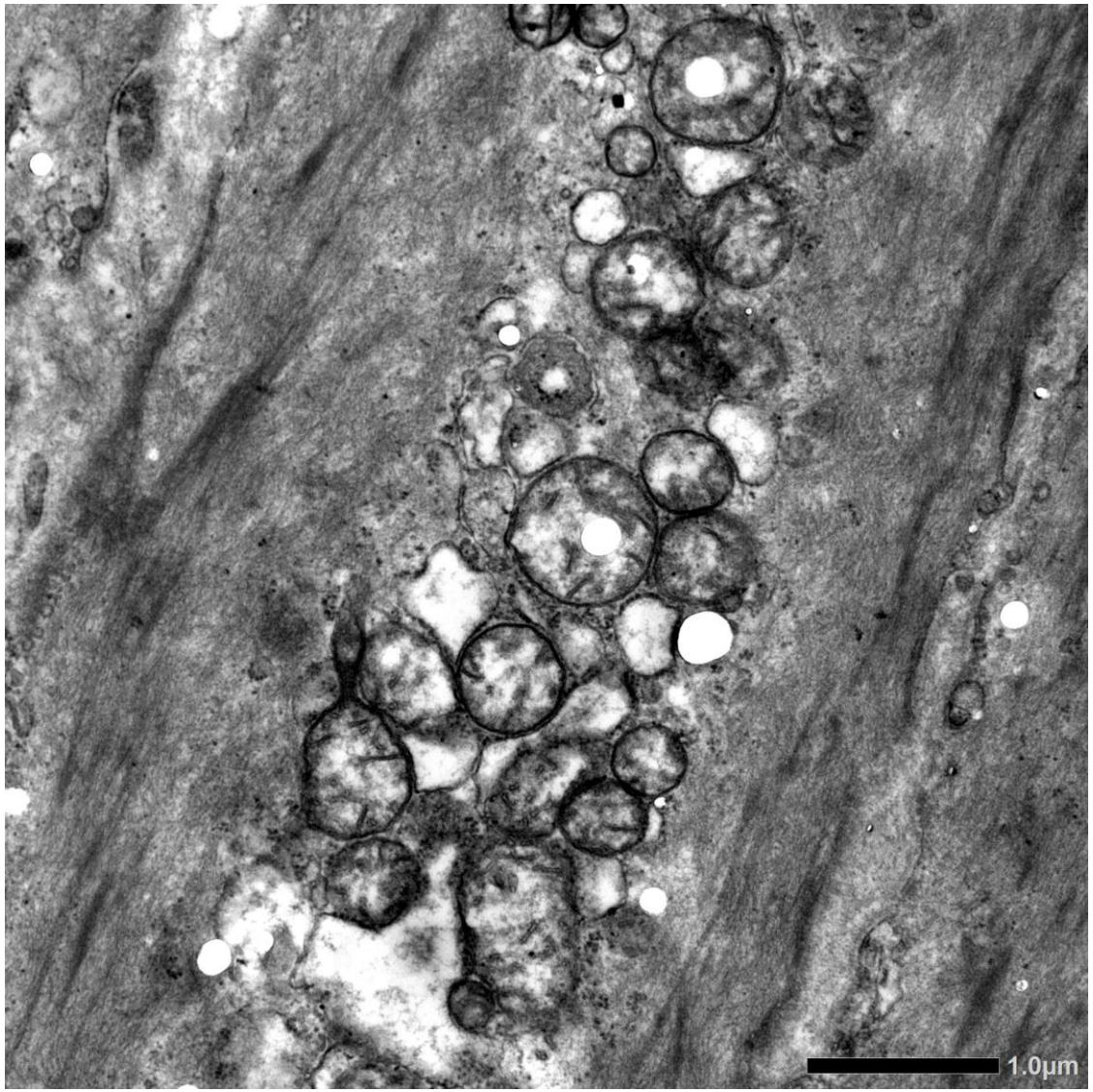
Colon biopsy from a 3-year-old girl with Leigh syndrome shows unremarkable change light microscopically. Thick section for EM study (toluidine blue)



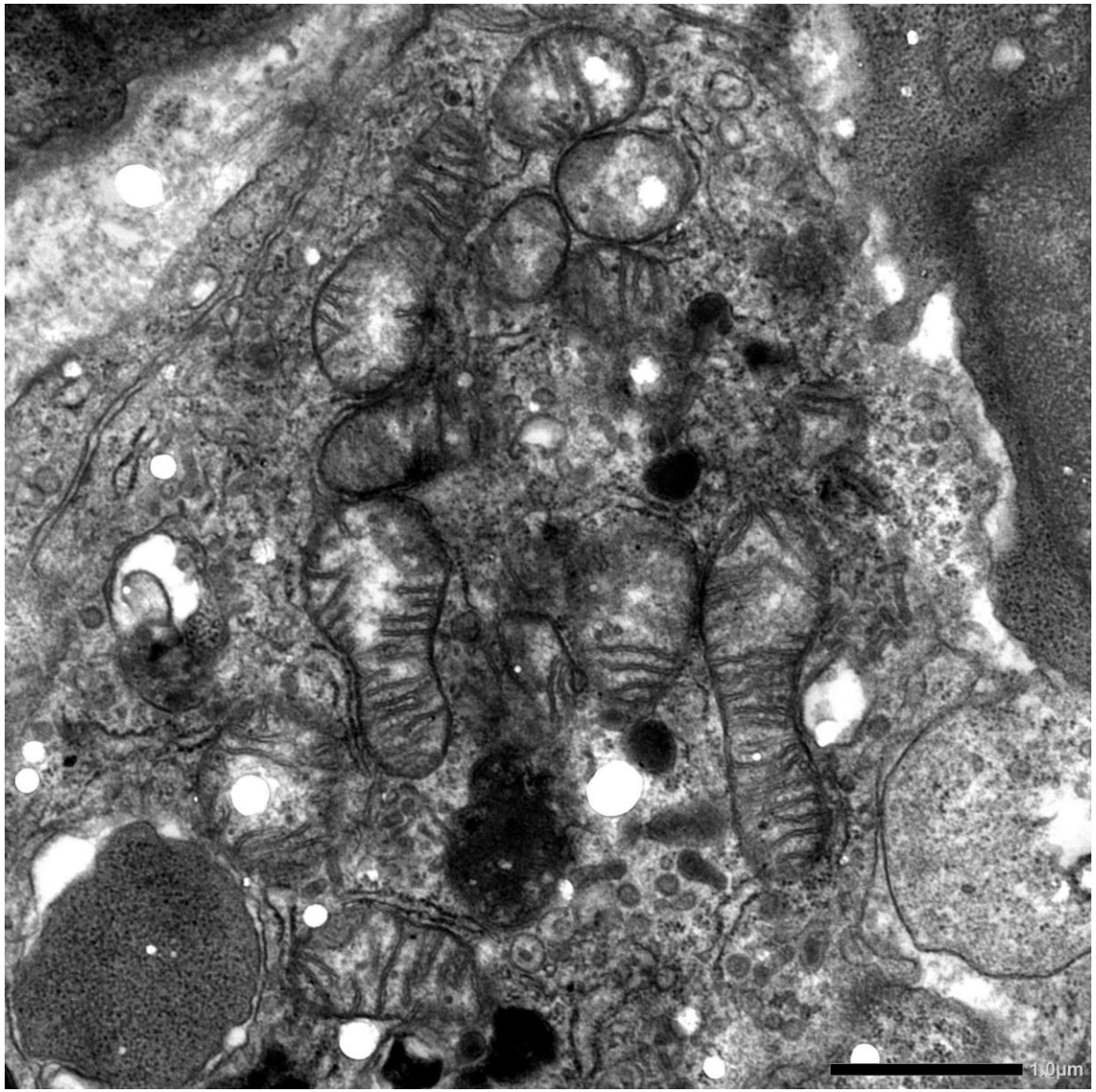
EM features of colonic mucosa. Mild increase of mitochondria is observed in the cytoplasm of the columnar cells. EM-1



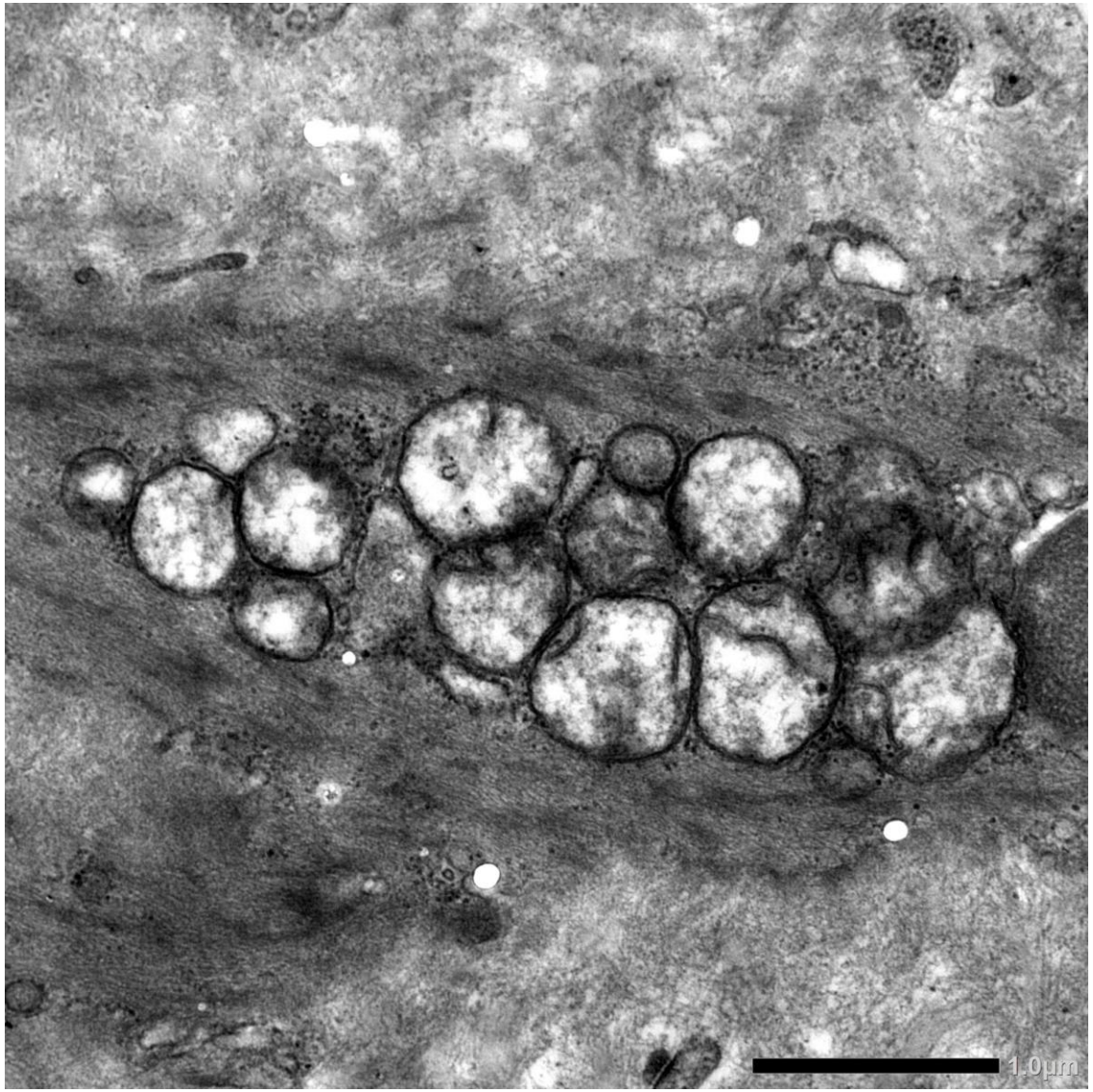
EM features of colonic mucosa. Mild increase of mitochondria is observed in the cytoplasm of the columnar cells, including goblet cells. EM-2



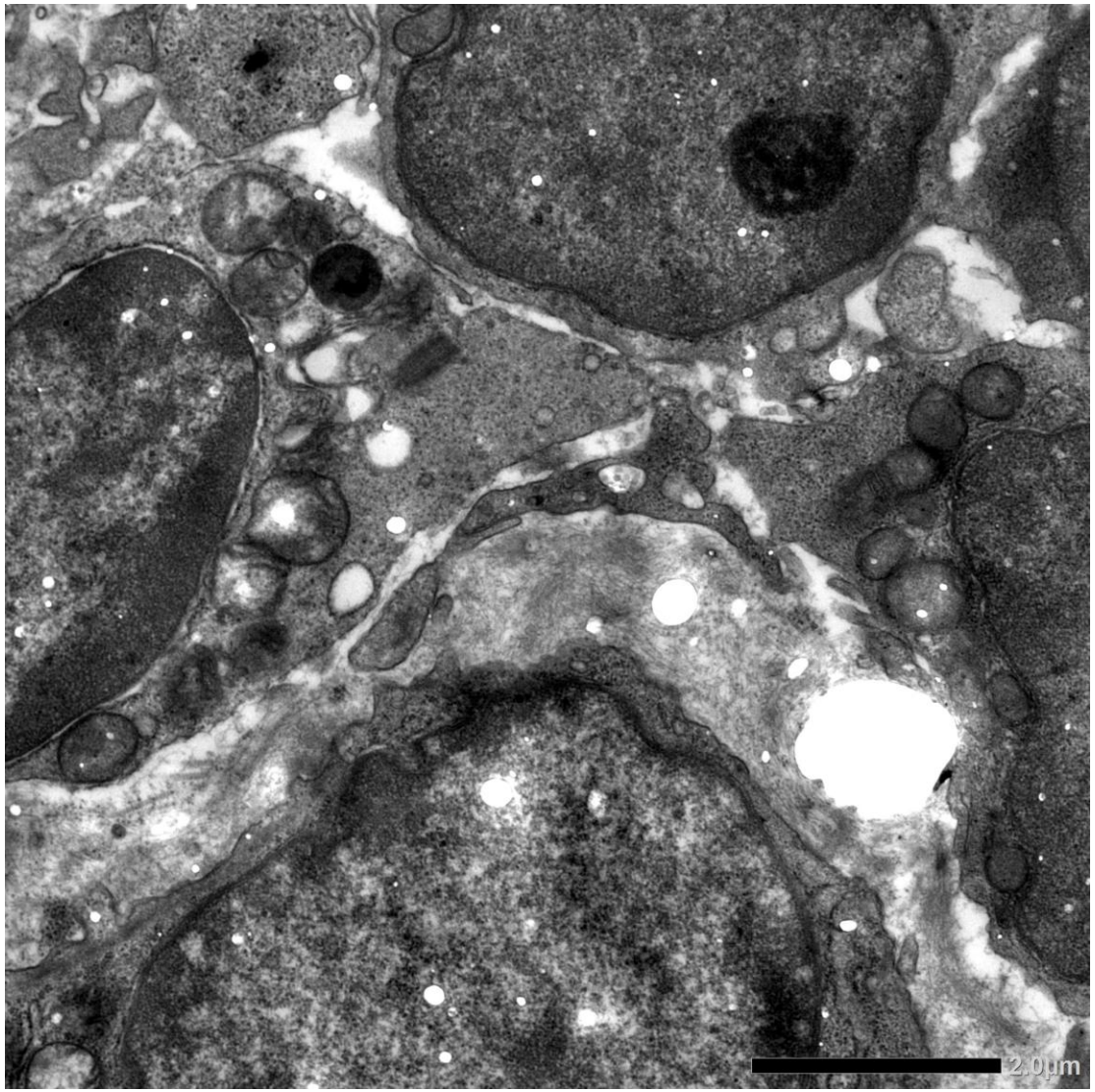
EM features of colonic mucosa. Mitochondria are clustered in the cytoplasm of the smooth muscle cells of the tunica muscularis mucosae. EM-3



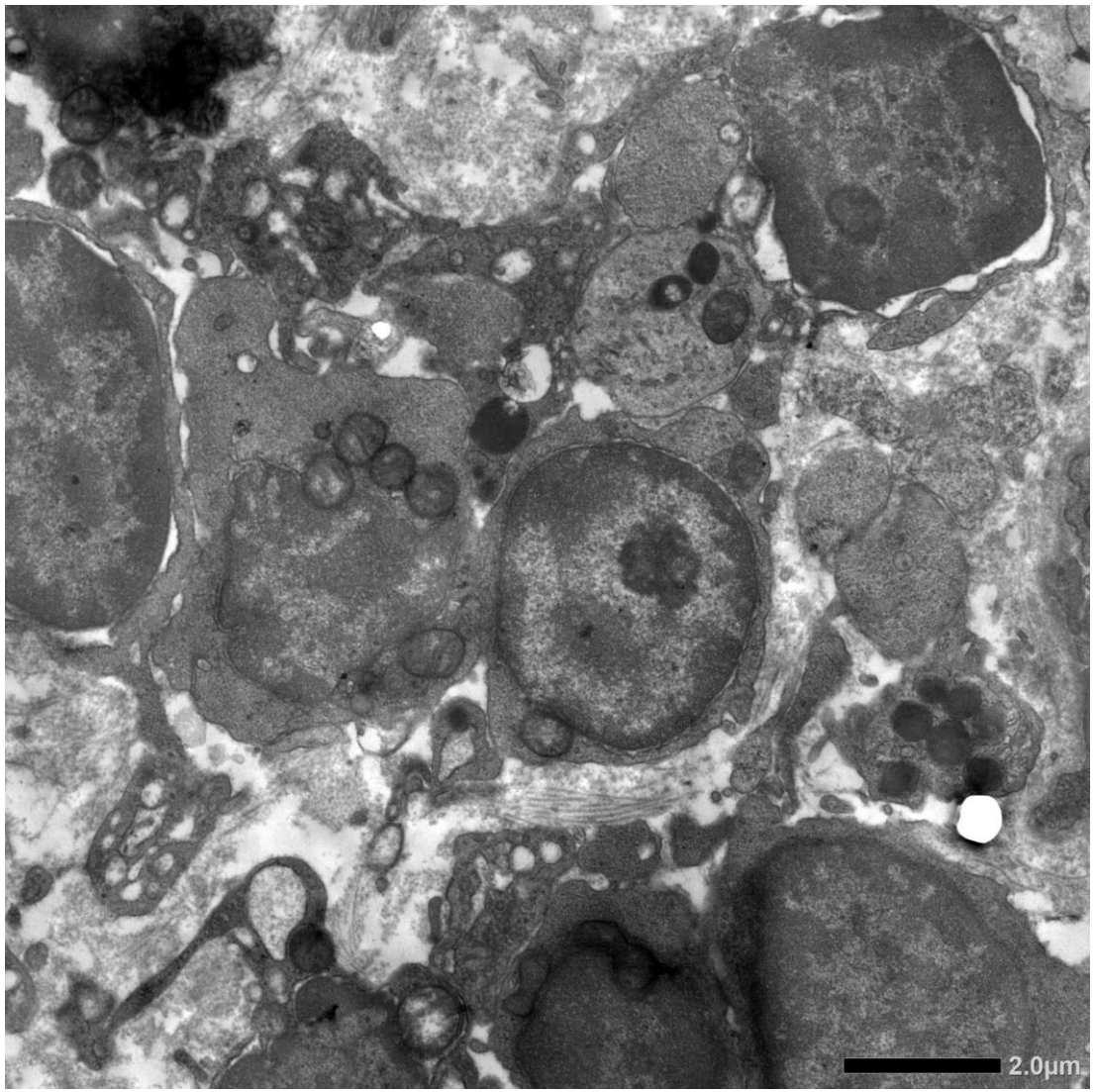
EM features of colonic mucosa. Mitochondria are clustered in the cytoplasm of the smooth muscle cells of the tunica muscularis mucosae. EM-4



EM features of colonic mucosa. Mitochondria are clustered in the cytoplasm of the lamina proprial myofibroblast. EM-5



EM features of colonic mucosa. Mitochondria seem to be mildly increased in the cytoplasm of the macrophages of the lamina muscularis mucosae. EM-6



Ultrastructure of the colonic lamina propria mucosae. Mitochondria are mildly increased in the cytoplasm of lymphocytes and macrophages. EM-6