Chronic intestinal pseudo-obstruction due to megacystis-microcolon-intestinal hypoperistalsis syndrome

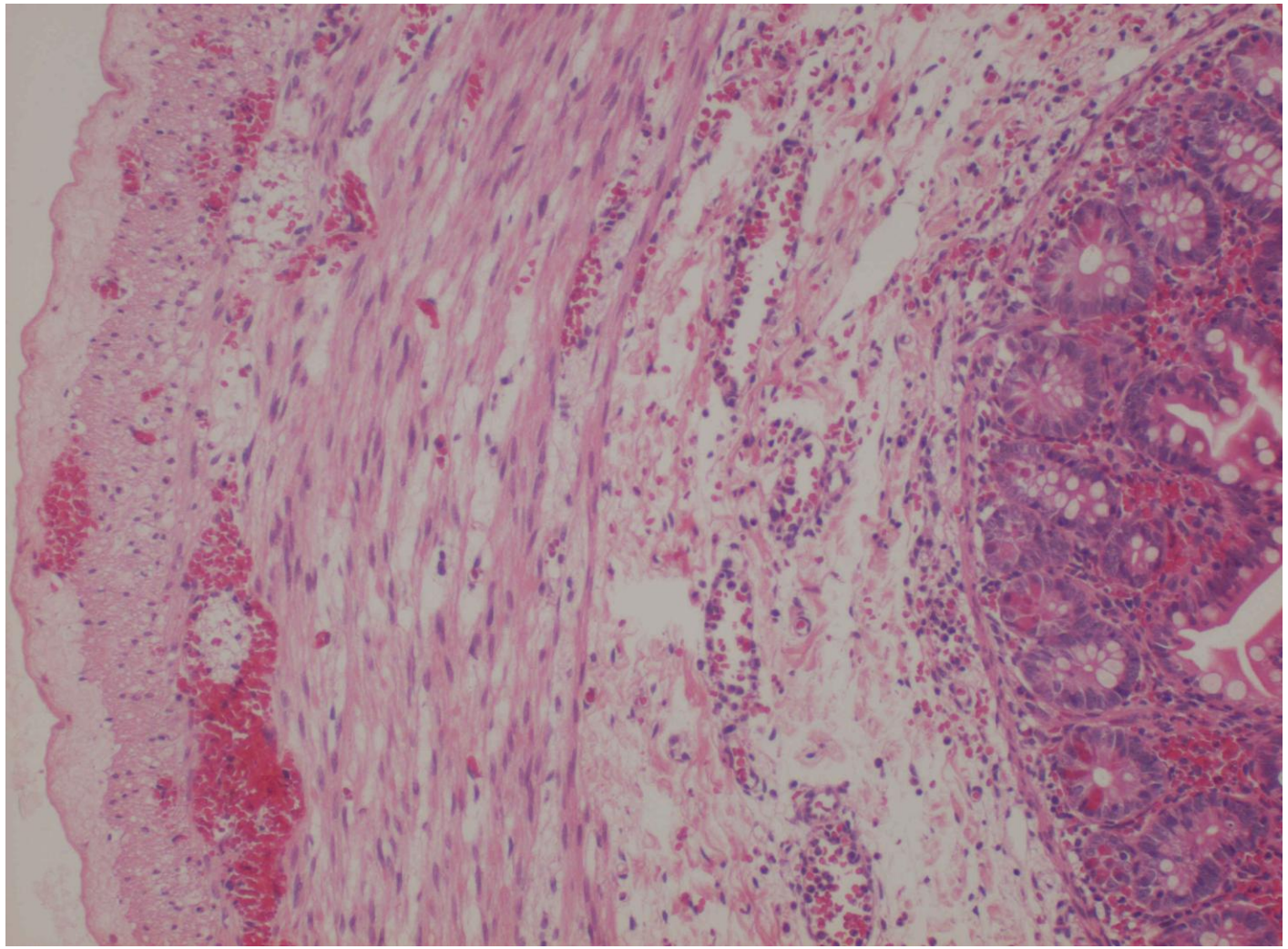
Megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS), a genetic disorder with autosomal dominant or recessive trait, is characterized by megacystis (bladder distention in the absence of mechanical obstruction), microcolon and intestinal hypoperistalsis (dysmotility). Infants and children with MMIHS have myopathic dysfunction of the urinary bladder. Gastrointestinal complication includes bowel obstruction in the absence of mechanical obstruction, known as chronic intestinal pseudo-obstruction (CIPO). Intestinal dysfunction ultimately leads to nutritional compromise and intestinal failure, resulting in the dependence on total parenteral nutrition. The prognosis is poor. Ultrastructural features of smooth muscle cells include vacuolar changes, excessive glycogen storage and markedly reduced contractile fibers displaced to the extreme periphery of the cells. A fundamental defect of glycogen-energy utilization is suggested.

Ref.-1: Ambartsumyan L. Megacystis-microcolon-intestinal hypoperistalsis syndrome overview. 2019. In: Adam MP, et al (eds). GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle. Available from: https://www.ncbi.nlm.nih.gov/books/NBK540960/

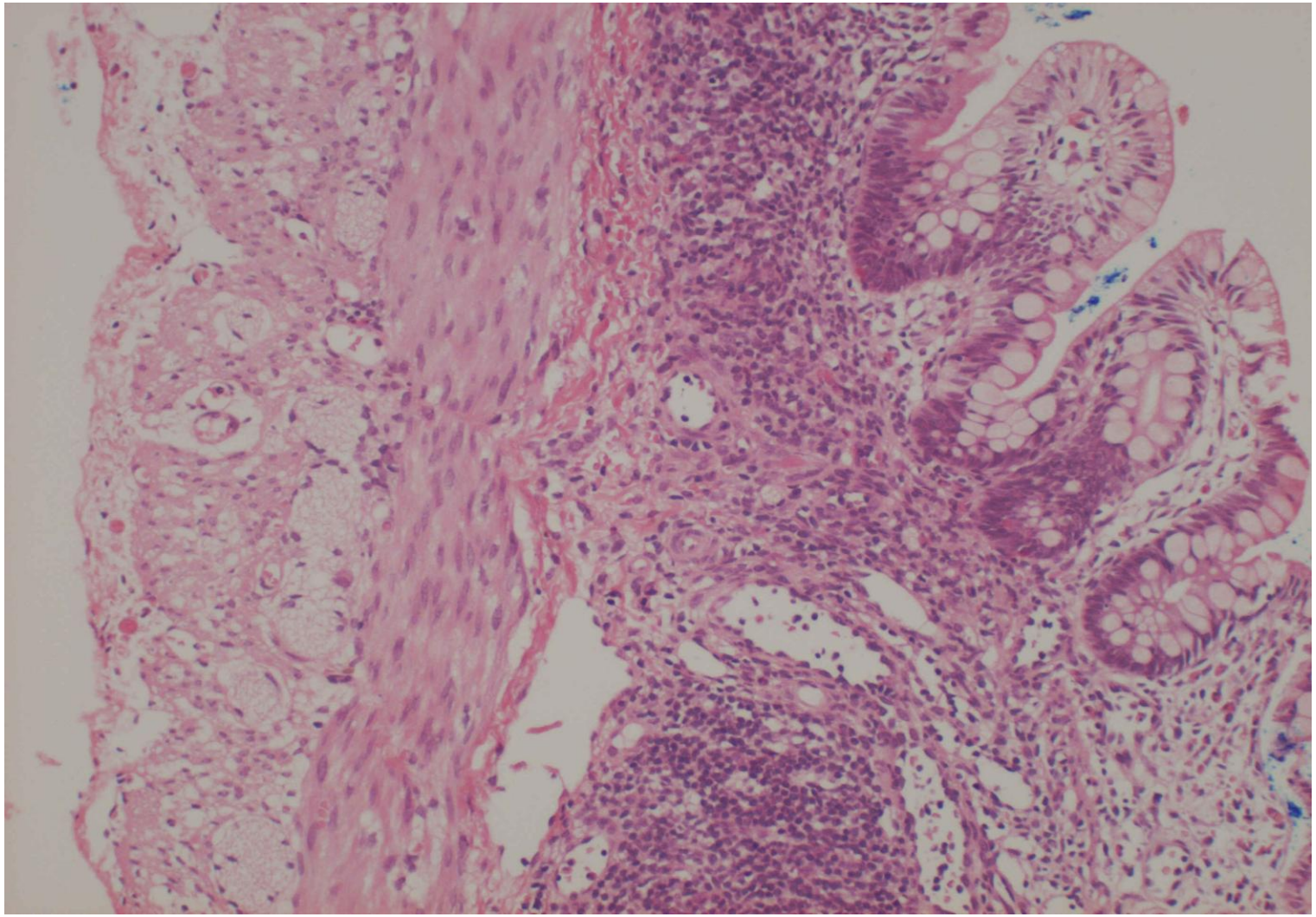
Ref.-2: Ciftci AO, et al. Megacystis microcolon intestinal hypoperistalsis syndrome: evidence of a primary myocellular defect of contractile fiber synthesis. J Pediatr Surg 1996; 31(12): 1706-1711. doi: 10.1016/s0022-3468(96)90058-5

Case presentation

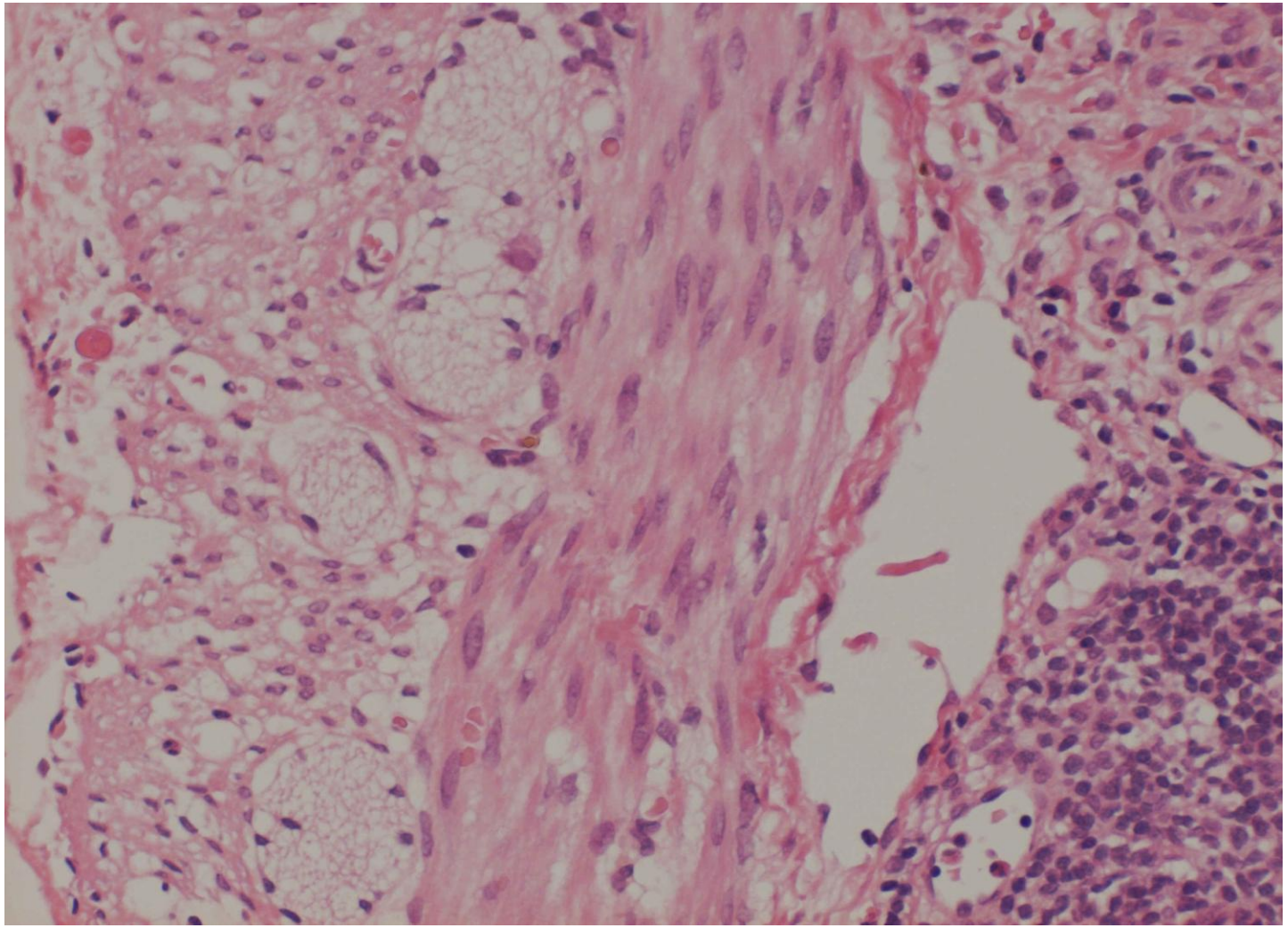
A 10-day-old female baby lacked meconium defecation, and emergency gastrostomy and colostomy were performed. The ileal tissue was partly sampled for microscopic and ultrastructural evaluations. Dilated urinary bladder had been identified *in utero*. Thus, megacystis-microcolon-intestinal hypoperistalsis syndrome was clinically suspected.



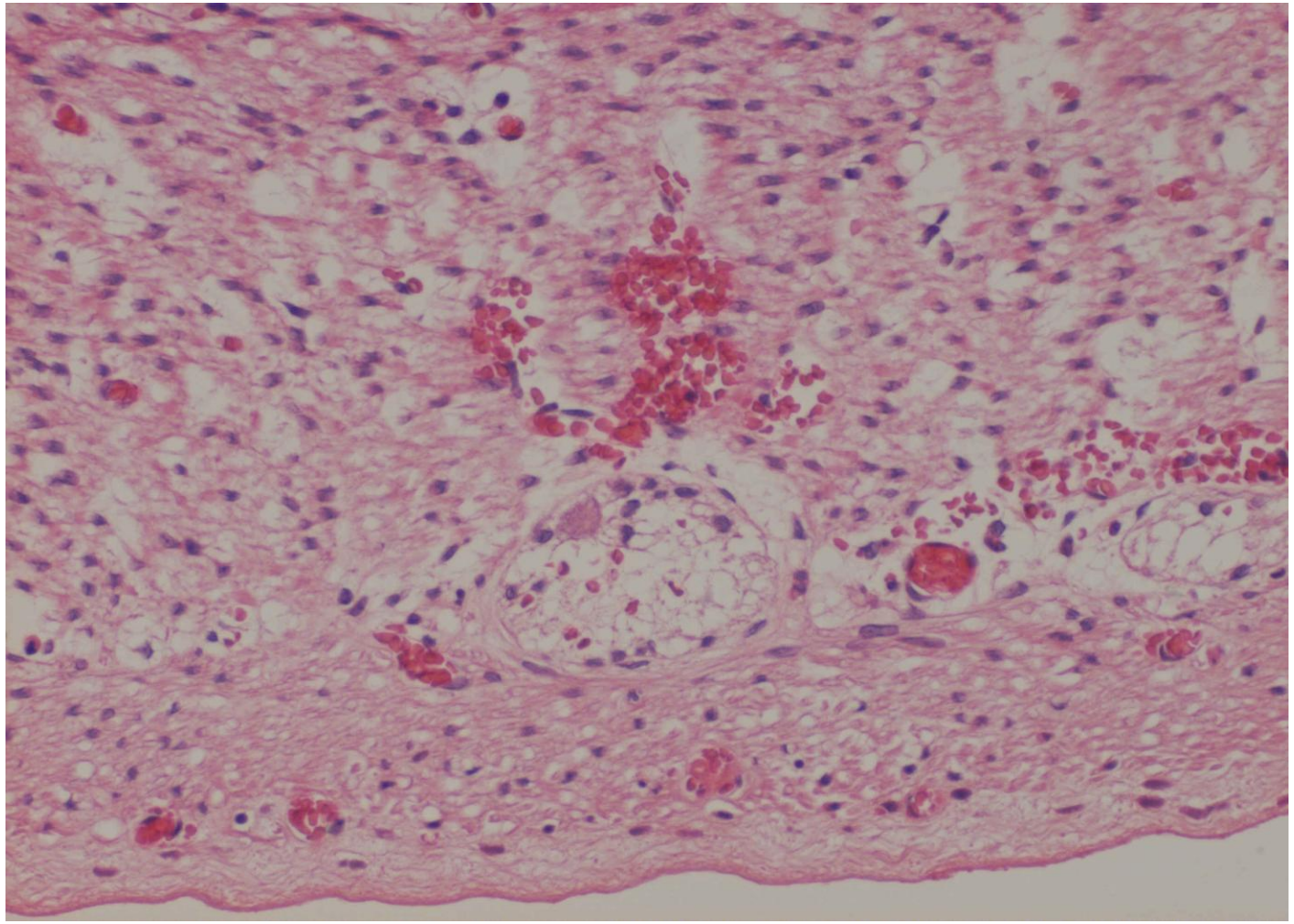
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. The ileal tissue was sampled during the emergency colostomy for chronic intestinal pseudo-obstruction. The proper muscle layer is thin and loosely arranged. The tunica muscularis mucosae is poorly developed. H&E-1



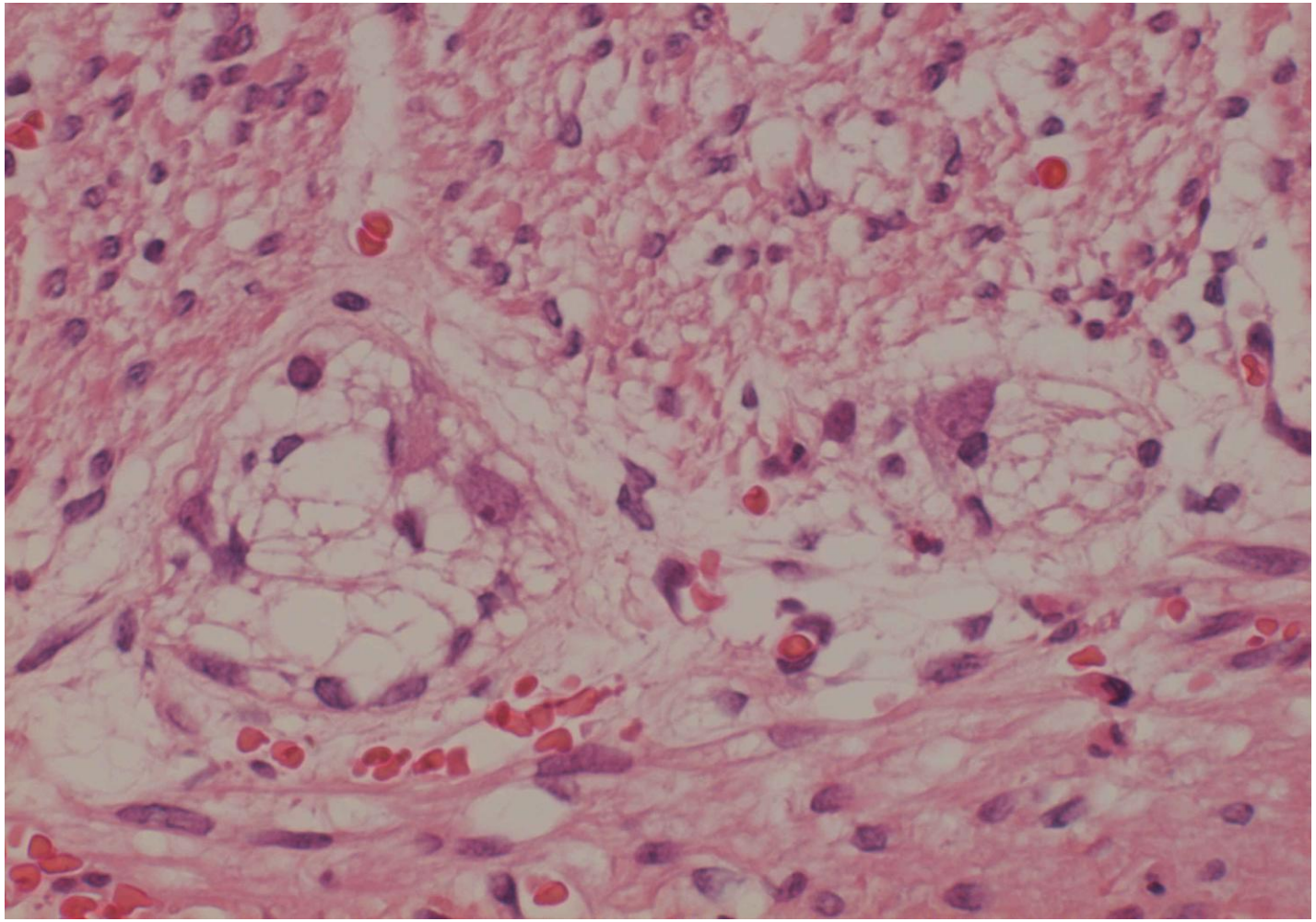
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. The ileal tissue was sampled during the emergency colostomy for chronic intestinal pseudo-obstruction. The proper muscle layer is thin and vacuolated. Auerbach's plexus appears intact. The tunica muscularis mucosae is poorly developed. H&E-2



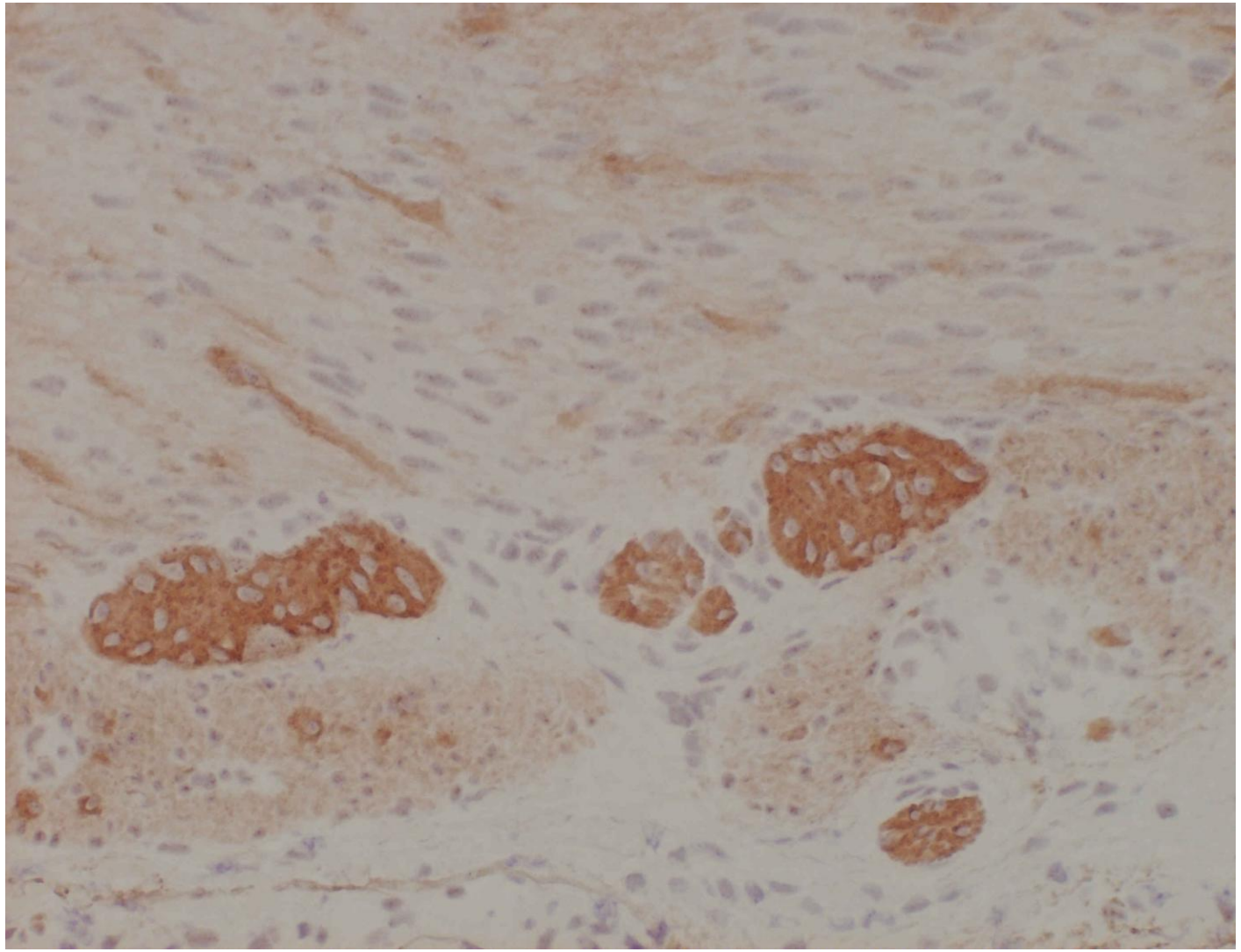
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. The ileal tissue was sampled during the emergency colostomy for chronic intestinal pseudo-obstruction. The proper muscle layer is thin and vacuolated. Auerbach’s plexus appears intact. H&E-3



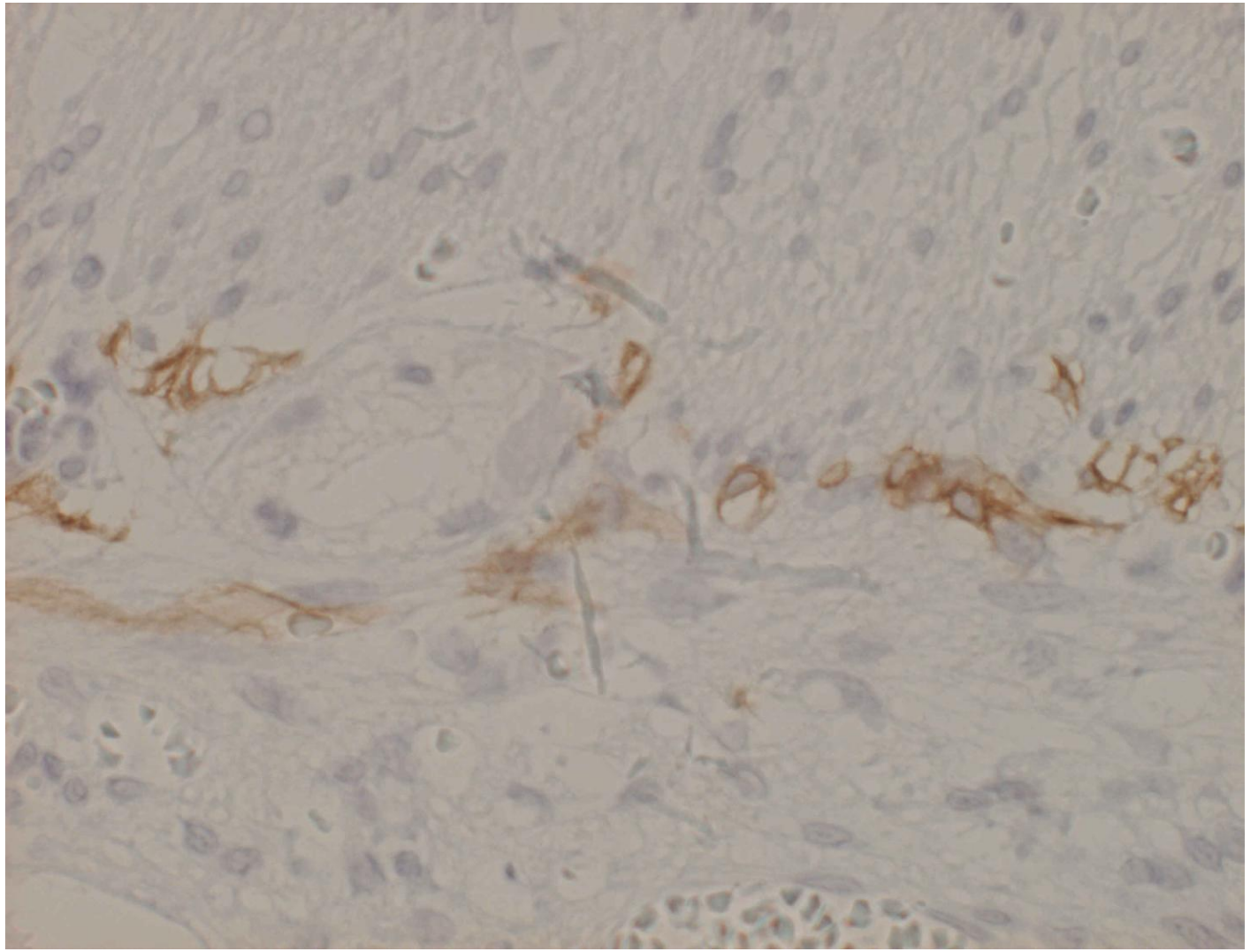
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. The ileal tissue was sampled during the emergency colostomy for chronic intestinal pseudo-obstruction. The proper muscle layer is thin and vacuolated. Auerbach's plexus appears intact. H&E-4



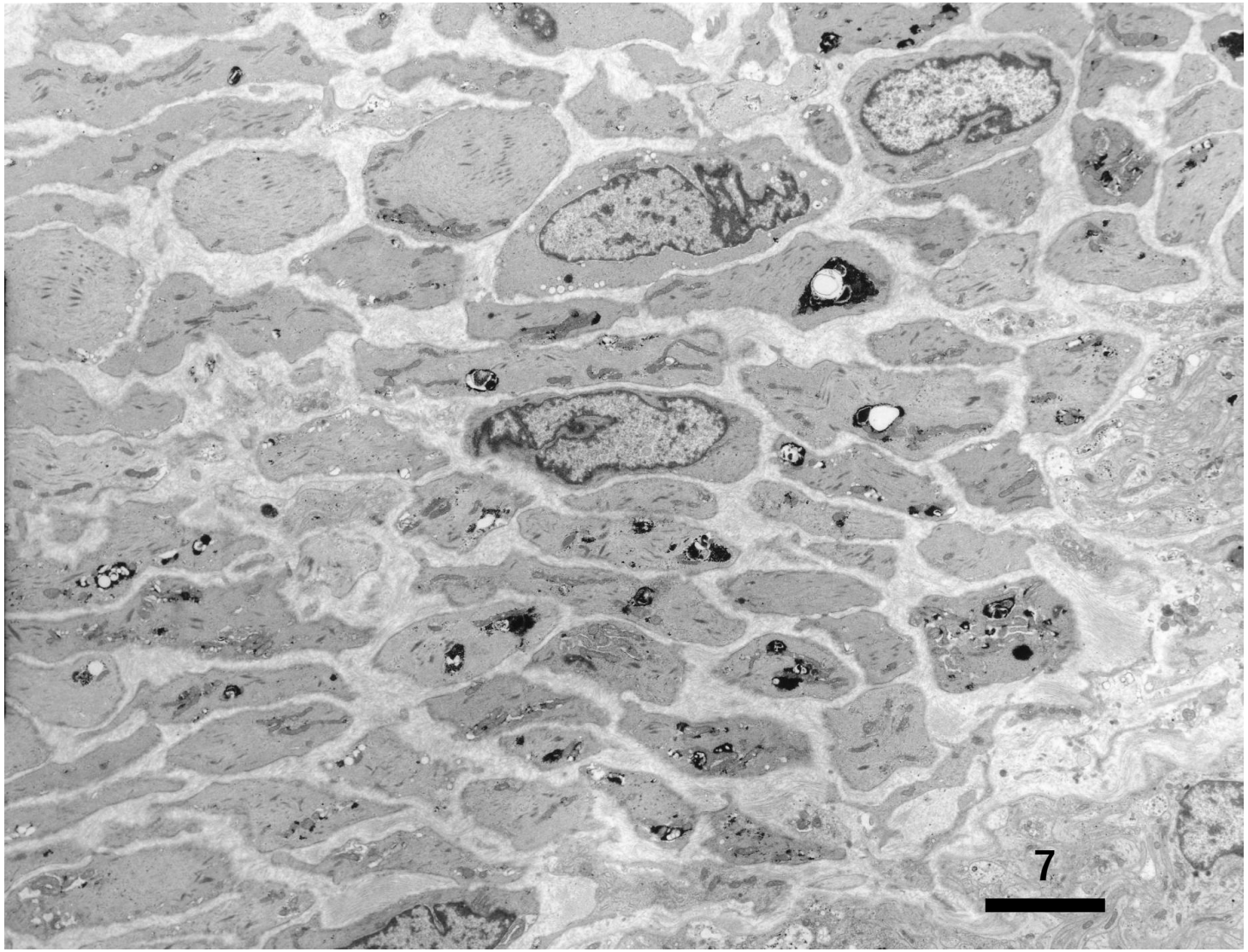
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. The ileal tissue was sampled during the emergency colostomy for chronic intestinal pseudo-obstruction. The proper muscle layer is thin and vacuolated. Auerbach's plexus appears intact. H&E-5



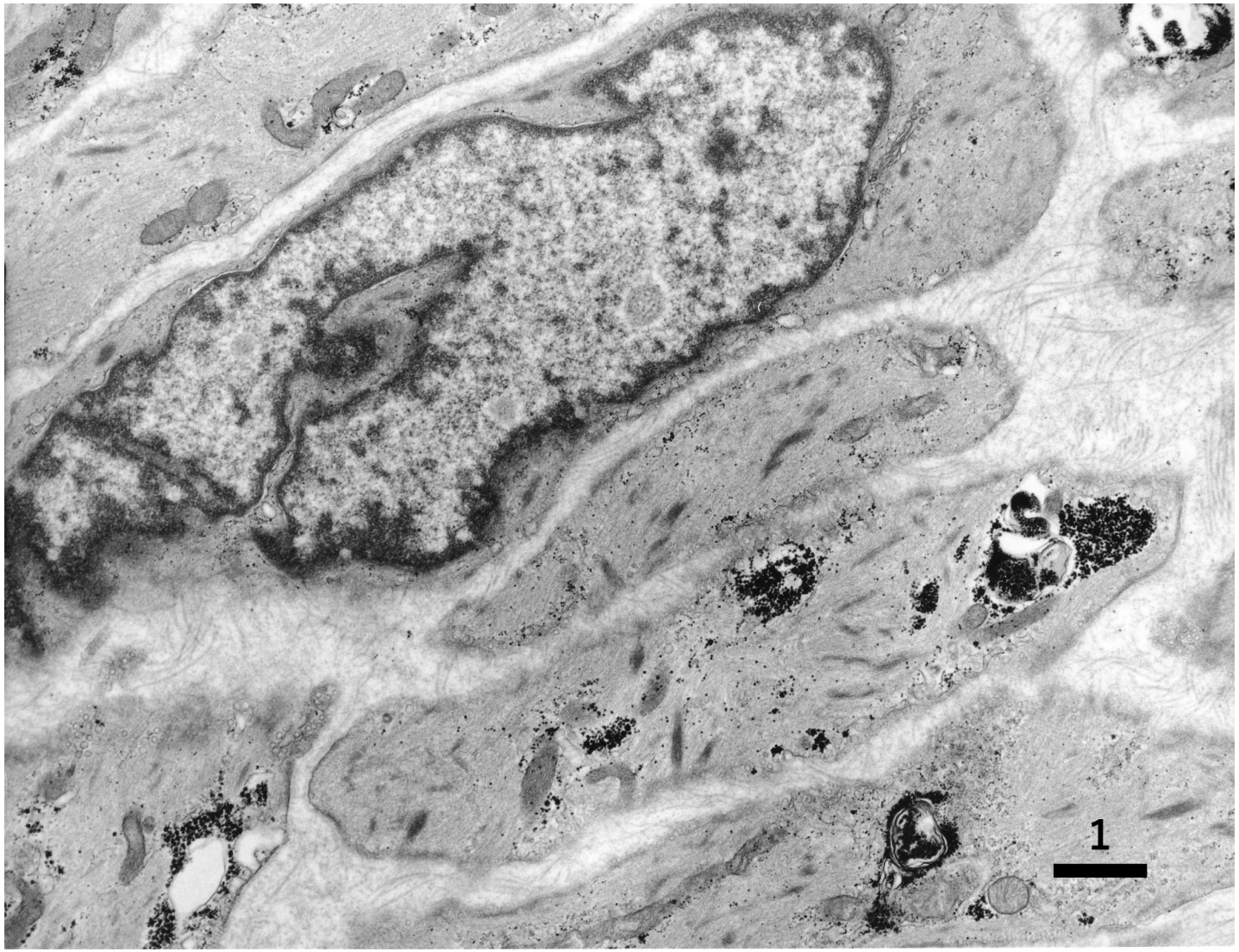
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. The ileal tissue was sampled during the emergency colostomy for chronic intestinal pseudo-obstruction. Auerbach's plexus with acetylcholine esterase immunoreactivity appears intact. Immunostaining for acetylcholine esterase



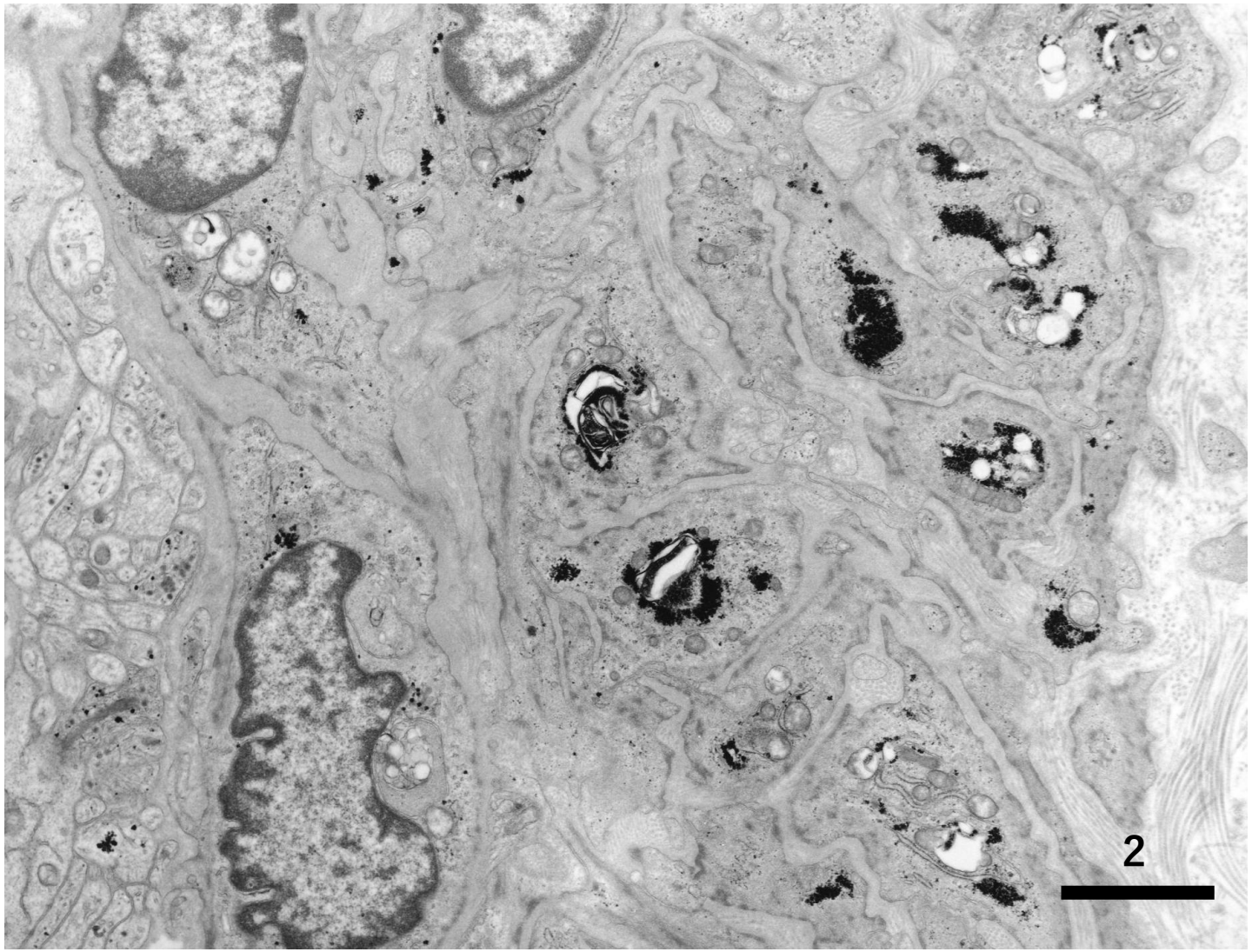
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. The ileal tissue was sampled during the emergency colostomy for chronic intestinal pseudo-obstruction. Cajal cells in the Auerbach's plexus with CD117 (c-kit) immunoreactivity appear intact. Immunostaining for c-kit



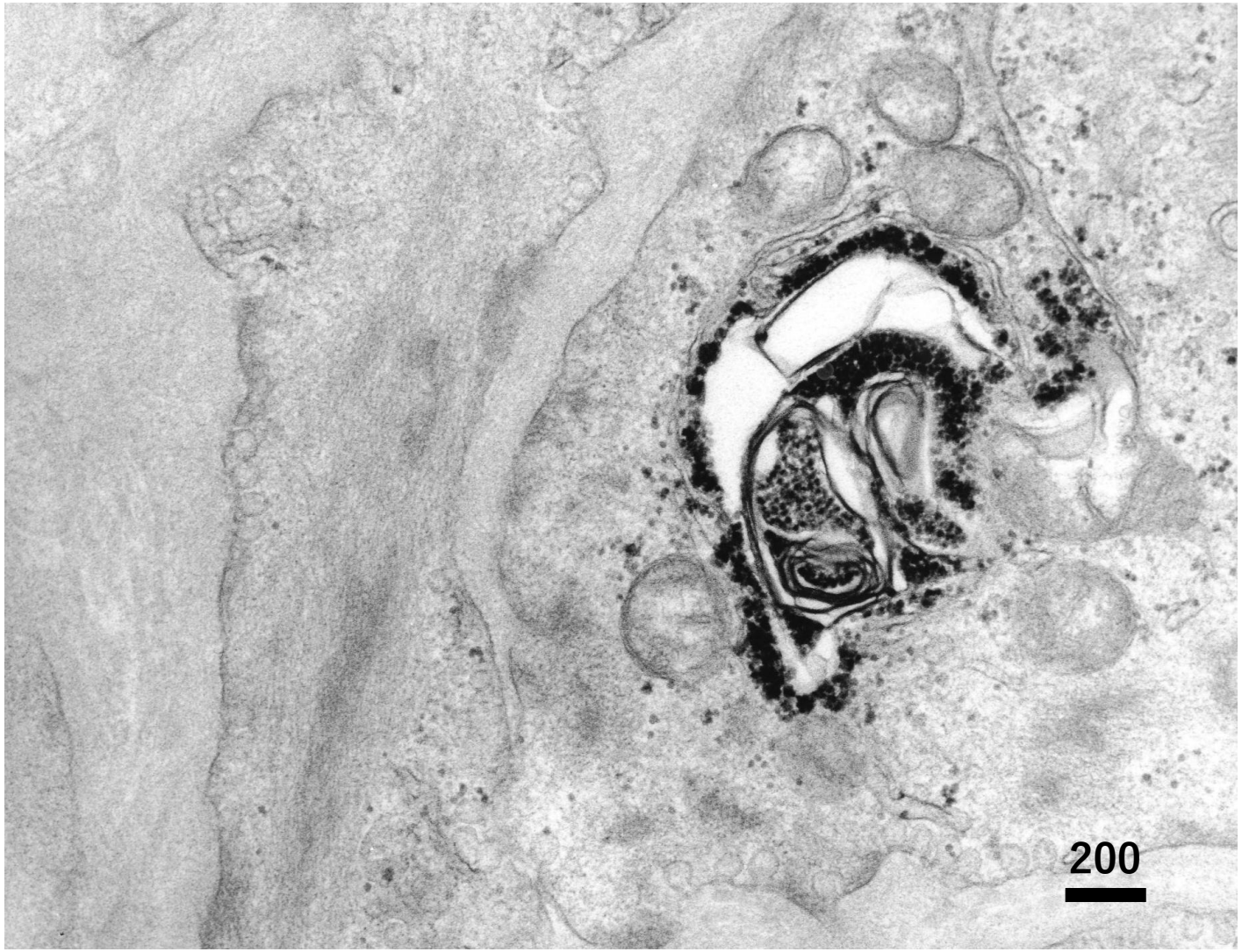
Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. Ultrastructure of the ileal proper muscle layer reveals smooth muscle cells with multifocal accumulation of electron-dense glycogen particles in the cytoplasm. EM-1



Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. Ultrastructure of the ileal proper muscle layer reveals smooth muscle cells with multifocal accumulation of electron-dense glycogen particles in the cytoplasm. Myofibrils with dense patch formation are sparse and displaced at the periphery of the cell. EM-2



Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. Ultrastructure of the ileal proper muscle layer reveals smooth muscle cells with multifocal accumulation of electron-dense glycogen particles in the cytoplasm. Myofibrils with dense patch formation are sparse and appears to be displaced at the periphery of the cell. EM-3



Megacystis-microcolon-intestinal hypoperistalsis syndrome in a 10-day-old girl. Ultrastructure of the ileal proper muscle layer reveals smooth muscle cells with multifocal accumulation of electron-dense glycogen particles in the cytoplasm. The glycogen particles surround membranous debris. The density of myofibrils is decreased. EM-4