

DIFFUSE INTESTINAL GANGLIONEUROMATOSIS OF THE ILEUM

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Key-word: Ganglioneuroma

Background: A 39-year-old man presented with nonspecific abdominal pain. He was known with neurofibromatosis type I, with history of café-au-lait spots, multiple cutaneous neurofibromas and osteoporosis. Findings on contrast-enhanced CT scan were rather nonspecific, but inflammatory small bowel disease was suspected.



Fig.

1A	2A
1B	2B

Work-up

Contrast-enhanced CT scan of the abdomen (Fig. 1) shows on transverse image at umbilical level (A) dilatation of the preterminal ileum over a length of 20-30 cm. Diffuse, asymmetric wall thickening of the mesenteric bowel wall of this segment (arrows). The terminal ileal loop appears normal. On reformatted image (coronal plane) (B) multiple nodules are observed in the mesentery adjacent to thickened wall (arrows).

CT enteroclysis (Fig. 2) demonstrates on transverse image at infra-umbilical level (A) the asymmetric aspect of the wall thickening in the preterminal ileum (arrows). Hypovascular tape-like infiltration of the mesenteric fat parallel to the bowel wall is noted. Reformatted image (coronal plane) (B) confirms the nodular infiltration of the mesentery (arrow).

Radiological diagnosis

Differential diagnosis in the presented case includes Whipple disease, nonspecific Crohn's disease and lymphoma or carcinoid tumor with diffuse infiltration of the ileum. Resection of the small bowel was performed. Pathologic examination revealed submucosal proliferation of neuroid cells and ganglion cells, with extension into the mucosa and serosa. These findings are diagnostic for *diffuse intestinal ganglioneuromatosis of the preterminal ileum*.

Discussion

Neurofibromatosis type I or von Recklinghausen disease is a genetic disorder, inherited in an autosomal dominant pattern, with incidence of approximately 1:3000. The disease presents with a broad spectrum of tumoral lesions, predominantly neuroectodermal or mesenchymal in origin, resulting from abnormal tumor suppression. These lesions may occur in a variety of organs and tissues. In 10-25% of patients, the gastro-intestinal tract is involved, the most common abdominal neoplasms in NF1 being neurofibromas. Ganglioneuromas are rare, benign tumors, that arise from sympathetic ganglia. They contain mature Schwann cells, ganglion cells and nerve fibres, and may appear anywhere along the parasympathic plexus and in the adrenal medulla. Rarely, they occur in the gastro-

intestinal tract. Ganglioneuromas of the bowel may present as solitary lesions, or as multiple polyps affecting the colon and the terminal ileum, termed ganglioneuromatosis polyposis. The polypoid form of ganglioneuromatosis has no proven association with neurofibromatosis or other syndromes. A third form, diffuse ganglioneuromatosis, is associated with syndromes such as neurofibromatosis type I and multiple endocrine neoplasia IIb (MEN IIb). It consists of hyperplasia of the myenteric plexus and infiltration of ganglioneuromatous tissue in the bowel wall. This leads to thickening of the bowel wall, sometimes with transmural extension and eventually stricture formation. In most reported cases, the large intestine is involved, but the terminal ileum and the appendix may also be affected. Affected patients may present with acute intestinal obstruction or nonspecific complaints due to motility disorders. Due to its rarity, diffuse intestinal ganglioneuromatosis is easily missed or misdiagnosed. When the terminal ileum is affected, radiographic findings may show circumferential bowel wall thickening, suggesting Crohn's disease or intestinal lymphoma. However, recognizing this entity is important, as the only therapeutic option is resection of the affected bowel. Furthermore, bowel involvement can be the first manifestation of neurofibromatosis. In these cases, a correct diagnosis has considerable implications for further management. In the presented case, the preterminal ileum was affected, while the terminal ileal loop and colon remained normal. Another interesting finding in this case are the mesenteric nodules. These were first considered enlarged lymph nodes, but pathological examination revealed they corresponded to ganglioneuromatous proliferations in the mesentery.

Bibliography

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