Ileal polyposis as manifestation of neurofibromatosis syndrome

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A 13-year-old girl presented with features of intestinal obstruction. At surgery, the terminal 25 cm of ileum, which was resected along with the right colon, showed plexiform neurofibromatosis of the serosa and mesentery, hyperplastic submucosal and myenteric nerve plexuses, and proliferation of neural tissue in the lamina propria, which manifested as diffuse polyposis of the ileal mucosa. The patient had a single inconspicuous external neurofibroma and a few café-au-lait spots on the back. [Indian J Gastroenterol 2006;25:159-160]

Von Recklinghausen’s disease or type I neurofibromatosis has variable clinical manifestations, but the most distinctive ones include café-au-lait spots, multiple neurofibromas and Lisch nodules (pigmented iris hamartomas).1 The café-au-lait spots usually manifest immediately after birth; gastrointestinal tract manifestations are seen in about 10%-25% of cases and occur in middle age.2,3

A 13-year-old girl, complaining of intermittent colicky right lower abdominal pain since one year, was admitted with abdominal pain and vomiting. An irregular ill-defined mass was palpated in the left lumbar region. Ultrasonography showed a few dilated fluid-filled bowel loops with normal peristalsis and thickened bowel wall in the left lumbar region and pelvis. CT scan in addition showed differential enhancement of serosa, muscularis propria and mucosa, suggestive of benign etiology, and maximum wall thickness of 2.5 cm. The clinical impression was intestinal obstruction due to tuberculosis or lymphoma.

At exploratory laparotomy, right hemicolecotomy was performed in view of an ileal and mesenteric mass involving the terminal 25 cm of the ileum with dilatation of the proximal bowel. The ileal specimen showed diffuse nodularity of the serosa and mesentery due to multiple white, glistening masses of varying sizes (1 cm to 6 cm). The ileal wall was thickened and the mucosa was carpeted with multiple, pinkish white, glistening filiform polyps (Fig 1a, 1b) measuring 0.2 cm to 0.4 cm in length. The proximal ileum was dilated. The ileo-cecal valve, cecum, appendix and ascending colon did not show any lesion.

Histology of the ileum showed marked distention of villi, proliferation of neural tissue and scattered ganglion cells in the lamina propria, abundant hypertrophy of submucosal and myenteric nerve plexuses with enlarged and dysplastic ganglion cells (Fig 1c), thickened muscle coat and multiple solitary and plexiform neurofibromas in the serosa and mesentery (Fig 1d). A diagnosis of neurofibromatosis and ganglioneuromatosis of the ileum was given. After this a single neurofibroma on the chest wall and two café-au-lait spots on the back were noted in the patient. On enquiry, the patient gave history of neurofibromatosis in the father and two maternal aunts.

Only a few reports of gastrointestinal involvement in von Recklinghausen’s disease exist. These patients may present with motility disorders, epigastric pain, dyspepsia, anemia, hematemesis, intussusception, volvulus, intestinal perforation or bowel obstruction.4 Gastrointestinal involvement may manifest in the form of hyperplasia of submucosal and myenteric plexuses, mucosal ganglioneuromatosis, gastrointestinal stromal tumors, and somatostatin-rich endocrine tumors of duodenum and peripancreatic region.1 Stromal tumors are the most well documented manifestation. They usually occur in middle age, and are usually benign. Neurofibromas, leiomyomas and schwannomas are common.

Neurofibromas have been found throughout the alimentary tract, most frequently in the stomach and jejunum.4 Morphologically, they can be solitary, diffuse or plexiform.4 Plexiform neurofibromas are pathognomonic of this disease. Occurrence of gastrointestinal neurofibromatosis in children is less common.5 Our patient manifested with diffuse polyposis of the mucosa. To our knowledge, such an appearance associated with neurofibromatosis has not been described in literature. Long-segment ileal neurofibromatosis as seen in the present case is also extremely uncommon, as opposed to the commoner pattern of involvement by localized neurofibroma.

References

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