Ileal Ganglioneuromatosis with Adenocarcinoma in a Patient with Multiple Neurofibromatosis

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Abstract Von Recklinghausen's disease or type 1 neurofibromatosis is an autosomal dominant disease characterized by mucocutaneous neurofibromas, café-au-lait spots and involves many organs including gastrointestinal tract. Gastrointestinal involvement in neurofibromatosis is uncommon and is seen 25% patients and nearly 5% of them are only symptomatic (Shekitka and Sobin, *Am J Surg Pathol* 18:250–257, 1994; Tomita et al., *Int J Colorectal Dis* 21: 89–91, 2006).

Keywords Ganglioneuroma · Neurofibromatosis

Introduction

A 44 year old patient with multiple neurofibromatosis presented with features of intestinal obstruction. CT scan featured a concentric thickening of terminal ileal loop with obstruction suspicious of ganglioneuromatosis intestine. Laparoscopic ileocaecal resection was performed and histopathology revealed ganglioneuromatosis with an area of adenocarcinoma involving terminal ileum.

Case Report

Von Recklinghausen's disease or type 1 neurofibromatosis is an autosomal dominant disease characterized by mucocutaneous neurofibromas, café-au-lait spots and involves

many organs including gastrointestinal tract. Gastrointestinal involvement in neurofibromatosis is uncommon and is seen 25% patients and nearly 5% of them are only symptomatic [1, 2].

A 44 year old male patient with multiple neurofibroma and café-au-lait spots presented with one week history of abdominal pain and distension. Clinical findings and X-ray findings were suggestive of intestinal obstruction. He had surgery for intracranial lesion 2 years before. CT scan revealed concentric homogenous non-enhancing thickening of terminal ileal loop with proximal bowel dilatation. In view of patient having multiple neurofibromas, ganglioneuromatosis was suspected. He required surgery as he did not respond to conservative regimen. Laparoscopy revealed thickened terminal ileum and mesentery and collapsed right colon with features of intestinal obstruction. Laparoscopic ileocaecal resection, mesenteric lymphadenectomy and end-to-side ileocolic anastomosis was performed.

Resected specimen revealed diffuse thickening of wall predominantly submucosal with multiple heaped up mucosal folds. An area of mucosal ulceration was noticed at the proximal end of bowel wall thickening. Histology revealed proliferation of ganglion cells and spindle cells predominantly involving the submucosa and mucosa along with hypertrophied nerve bundles in the mesentery suggestive of ganglioneuromatosis (Fig. 1). The ulcerated lesion was well differentiated adenocarcinoma involving subserosa with no lymph node involvement in the mesentery (pT3pN0).

Discussion

Gastrointestinal manifestations of multiple neurofibromatosis include 1). Hyperplasia of the submucosal and myenteric nerve plexuses 2). Ganglioneuromatosis and gastrointestinal

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Fig. 1 Proliferation of ganglion cells and spindle cells along with hypertrophied nerve bundles involving the submucosa and mucosa

stromal tumors and 3). Tumors in the duodenum and perampullary region [2, 3].

They present with abdominal pain, features of GI bleed, intussusception and bowel obstruction or perforation. Gastrointestinal ganglioneuromas may be divided into three groups: a). Polypoid ganglioneuroma with small sessile or pedunculated polyps, endoscopically indistinguishable from hyperplastic or adenomatous polyps b). Diffuse ganglioneuromatosis with lesions producing stricture-like segmental

thickening of the bowel c). Ganglioneuromatous polyposis with numerous polypoid lesions [2].

Association of ganglioneuroma with adenocarcinoma is reported with colon [2], however, small bowel ganglioneuroma with adenocarcinoma is extremely rare, though metastatic involvement is reported [4, 5]. In our patient association of adenocarcinoma with ganglioneuromatosis was not identified preoperatively, even in retrospect. Surgical resection is the treatment of choice in patients with multiple neurofibromatosis with symptomatic bowel involvement.

References

1. Shekitka KM, Sobin LH (1994) Ganglioneuromas of the gastrointestinal tract: relation to von Recklinghausen disease and other multiple tumor syndromes. *Am J Surg Pathol* 18:250–257
2. Tomita H, Miya K, Tanaka H, Shimokawa K (2006) Ganglioneuroma and adenocarcinoma associated with neurofibromatosis type1 in the colorectal region. *Int J Colorectal Dis* 21:89–91
3. Fuller CE, Williams GT (1991) Gastrointestinal manifestations of type 1 neurofibromatosis (von Recklinghausen's disease). *Histopathology* 19:1–11
4. Levy AD, Patel N, Abbott RM, Dow N, Miettinen M, Sobin LH (2004) Gastrointestinal stromal tumors in patients with neurofibromatosis: imaging features with clinicopathological correlation. *AJR* 183:1629–1636
5. Leslie A, Virjee JP, Moorghen M (1999) Plexiform neurofibroma of the small bowel infiltrated with metastatic adenocarcinoma. *Br J Radiol* 72:604–606