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Small Bowel Obstruction Due to Gastrointestinal Neurofibromas and Lipomas: A Case Report

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1. Abstract

Neurofibromatosis type 1 (NF-1), also known as Recklinghausen disease, is an autosomal dominant disorder most commonly recognized by cutaneous manifestations such as café-au-lait macules. Gastrointestinal involvement occurs in up to 25% of patients, although only a small proportion become symptomatic or require surgical intervention. We report a rare case of a 73-year-old woman with known intestinal NF-1 who presented with recurrent small bowel obstruction due to diffuse neurofibromas and lipomas. Computed tomography demonstrated extensive infiltration of the small bowel wall with associated intussusception. Surgical exploration confirmed multiple obstructing lesions, requiring segmental resections and primary anastomoses. This case highlights the rare but clinically significant gastrointestinal manifestations of NF-1 and the diagnostic and therapeutic challenges they present.

2. Keywords

Neurofibromatosis type 1, small bowel obstruction, intussusception, neurofibroma, gastrointestinal lipoma

3. Introduction

Neurofibromatosis type 1 (NF-1) is a common inherited neurocutaneous disorder with an estimated prevalence of 1 in 4,000 individuals. It is characterized by a wide spectrum of clinical manifestations, including café-au-lait spots, cutaneous neurofibromas, axillary freckling, and iris Lisch nodules. Diagnosis is based on established clinical criteria requiring at least two characteristic features.

Although NF-1 primarily affects the skin and nervous system, gastrointestinal involvement is reported in up to 25% of cases. The stomach and small intestine are the most frequently involved sites. Gastrointestinal manifestations include diffuse neurofibromatosis, ganglioneuromatosis, and solitary neurogenic tumors. Most lesions remain asymptomatic; however, a small subset of patients develop complications such as bleeding, obstruction, ulceration, or intussusception. Malignant transformation has been reported in approximately 10–15% of gastrointestinal lesions.

Lipomas are another benign but clinically relevant gastrointestinal tumor, accounting for about 5% of all gastrointestinal tumors. They are often submucosal, asymptomatic, and incidentally discovered; however, they may also cause obstruction or intussusception. When occurring alongside neurofibromas, they may complicate both diagnosis and management.

4. Case Presentation

A 73-year-old woman was admitted with a one-week history of progressive abdominal pain and symptoms consistent with intestinal obstruction. Her medical history was significant for intestinal NF-1, with five prior small bowel resections performed over the past decades due to recurrent intussusception and ileus caused by neurofibromas. Histopathological examination from previous surgeries consistently confirmed neurofibromatous lesions.

On current admission, abdominal computed tomography revealed diffuse and progressive tumor involvement of the small intestinal wall, consisting of multiple neurofibromas and lipomas. Several segments of the small bowel were markedly dilated, measuring up to 8.5 cm in diameter. Imaging also demonstrated an area of intussusception in the left iliac fossa, responsible for mechanical obstruction.

Given the patient's worsening clinical condition and radiological evidence of bowel obstruction, an emergency exploratory laparotomy was performed.

5. Operative Findings

Intraoperatively, two separate intussusception sites were identified: one located approximately 1.5 meters distal to the ligament of Treitz and another involving a previous ileocolic anastomosis. Multiple segments of the small intestine were affected by nodular lesions consistent with neurofibromas and lipomas.

Two segments of diseased bowel, each approximately 15 cm in length, were resected. Primary end-to-end anastomoses were successfully performed. No gross signs of malignancy or perforation were observed.

6. Postoperative Course

The patient initially recovered without complications. On postoperative day three, inflammatory markers increased, prompting initiation of broad-spectrum antibiotic therapy with

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cefuroxime and metronidazole. Clinical and laboratory parameters improved steadily, and antibiotics were discontinued on day six. The patient continued to recover uneventfully and tolerated oral intake.

Histopathological examination confirmed diffuse intestinal neurofibromatosis with multiple coexisting lipomas, consistent with the patient's known diagnosis.

7. Discussion

Gastrointestinal involvement in NF-1 is uncommon and often underrecognized. While most lesions remain clinically silent, symptomatic cases can present with serious complications such as obstruction, bleeding, and intussusception.

This case illustrates an unusual presentation of recurrent small bowel obstruction caused by extensive and diffuse involvement of both neurofibromas and lipomas. The patient's history of multiple prior resections reflects the chronic and recurrent nature of intestinal NF-1 in selected individuals.

Radiological diagnosis can be challenging because neurofibromas and lipomas may appear similar on imaging studies, and both can contribute to bowel wall thickening and luminal narrowing. In this case, computed tomography was essential in identifying the extent of disease and confirming the presence of intussusception.

Surgical intervention remains the primary treatment for symptomatic gastrointestinal NF-1. However, repeated resections should be approached cautiously, as diffuse disease increases the risk of recurrence. Conservative management is not appropriate in the setting of acute obstruction or ischemia.

This case also emphasizes an important clinical principle: benign gastrointestinal tumors can produce severe mechanical complications indistinguishable from malignant processes. Awareness of this possibility is essential when evaluating NF-1 patients with acute abdominal symptoms.

8. Conclusion

Gastrointestinal manifestations of NF-1, although rare, can lead to recurrent and clinically significant small bowel obstruction. The coexistence of neurofibromas and lipomas may contribute to intussusception and ileus, requiring surgical management. Careful long-term follow-up is necessary, as recurrence is common in patients with diffuse intestinal involvement.

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