Small Bowel Obstruction caused by NF-1

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Abstract

We report an unusual case of acute small bowel obstruction in a woman with Neurofibromatosis type 1 (NF1) caused by numerous gastrointestinal neurofibromas.

Introduction

- NF1 is classically characterized by cutaneous neurofibromas, flat hyperpigmented skins lesions (café-au-lait spots), tan-colored hamartomas of the iris (lisch nodules), optic gliomas, and freckles in the axillary and inguinal regions.
- Less commonly, NF1 is associated with a variety of gastrointestinal tumors including plexiform neurofibromas, mucosal ganglioneuromas, and gastrointestinal stromal tumors[1].
- Up to 25% of patients with NF1 are reported to have a gastrointestinal tumor of some kind, but it is estimated that only 5% of will develop gastrointestinal symptoms including bleeding, abdominal pain, abdominal distention, and constipation[1].
- Complications of the gastrointestinal tumors include perforations, hemorrhage, intussusception, and very rarely small bowel obstruction (SBO)[2].

Case Report

Case Report: Patient is a 26-year-old with a past medical history significant for Crohn's Disease who presented due to abdominal pain and hematochezia. Her Crohn's disease had been poorly controlled due to self-funding. Physical exam was notable for pain in the right upper quadrant. Labs were notable for Hgb 7.0, WBC 6.3, with CMP largely unremarkable. CT abdomen and pelvis with contrast showed high-grade small bowel obstruction with small bowel dilated up to 9 cm and severe active Crohn's disease with tethering of small bowel centrally. Patient had NG tube placed to wall suction. Colorectal surgery was consulted and performed laparotomy with ileocecectomy and end ileostomy formation. The terminal ileum and cecum were sent to pathology for further analysis. The pathology report showed "wall thickening showing extensive neural hyperplasia (ganglioneuromatosis) and plexiform neurofibromas. The features seen in the resection favor intestinal neurofibromatous proliferations (present throughout the bowel wall and in the mesentery) with secondary chronic mucosal changes. There is no transmural inflammation, granulomas or other features typically seen in Crohn's disease." After ileostomy creation, patient suffered from high ostomy output and had to be started on Loperamide.

Discussion

- Systematic review of the literature from 1972-2013 on acute intestinal obstruction due to NF1 identified only 25 cases of patients with NF1 who underwent laparotomy for acute small bowel obstruction[2].
- Histology was reported to show plexiform neurofibromas / neurofibroma (19), gastrointestinal stromal tumour (5) and adenocarcinoma (4)[2].

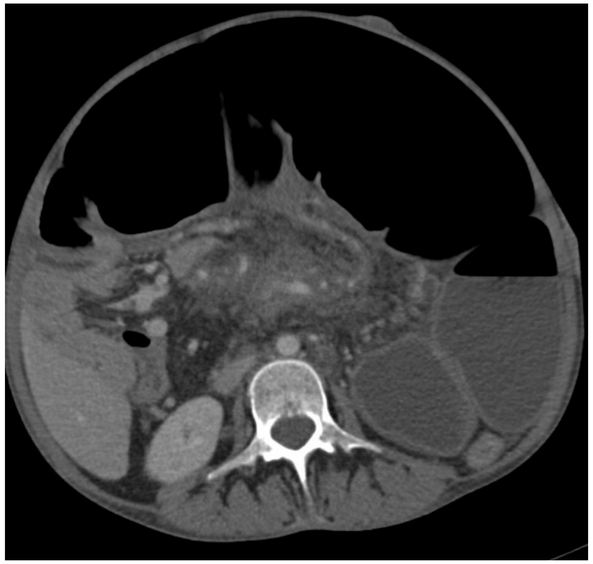

Figure 1: CT Abd/Pelvis showing high grade small bowel obstruction

References

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Disclosures: None