MEN2B-associated Chronic Intestinal Pseudo-obstruction

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Introduction

- Multiple endocrine neoplasia type 2B (MEN2B) is a rare genetic disorder characterized by medullary thyroid cancer and pheochromocytoma
- Gastrointestinal involvement is often observed with onset at birth
- Symptoms may manifest as constipation that can progress to megacolon – involvement of other portions of the GI tract are less commonly reported
- This patient with MEN2B presented with GI symptoms and was found to have chronic intestinal pseudo-obstruction (CIPO)

Case Description

- 23-year-old female with *MEN2B* presented to clinic for evaluation of abdominal pain, bloating, and loose stools
- Had chronic abdominal distension and episodes of abdominal pain since childhood
- 8 months prior to presentation, symptoms became severe and had required several hospitalizations for IV hydration

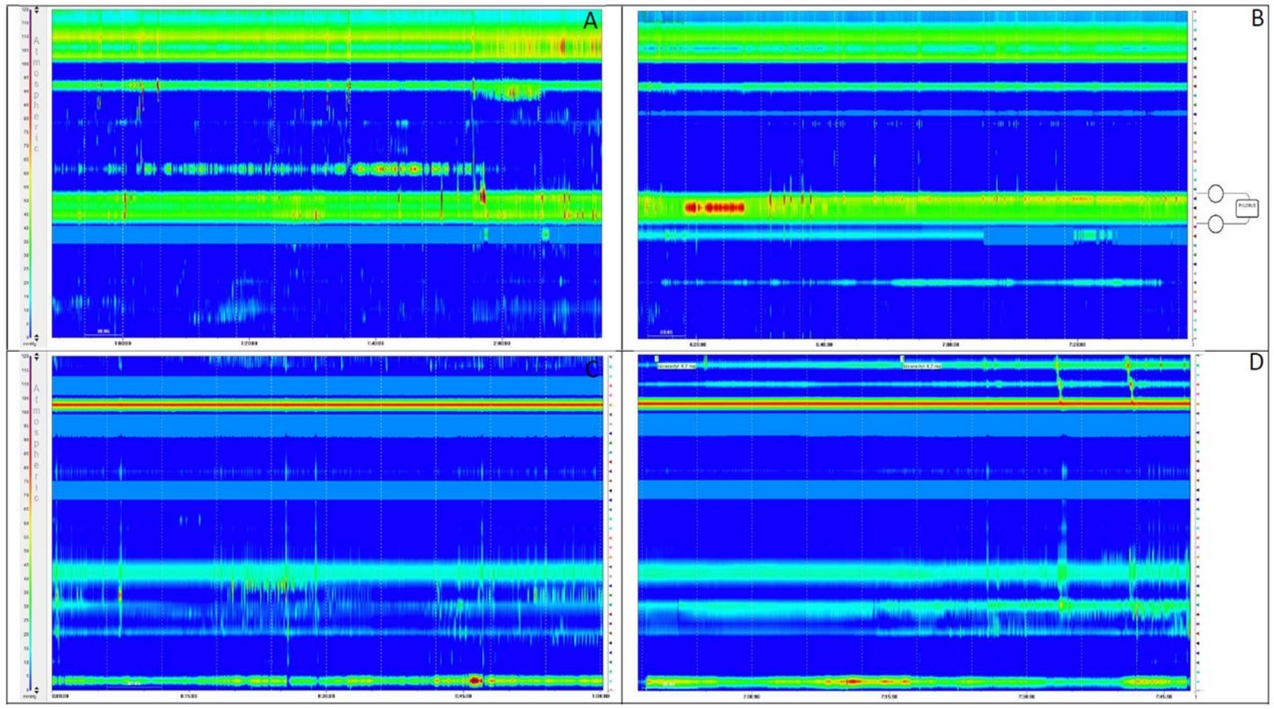
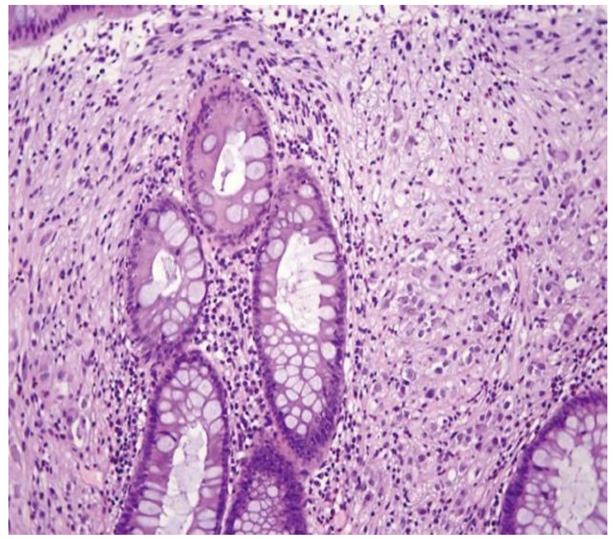


Figure 1: Antroduodenal manometry with notable absence of MMCIII in the fasted state (panel A), and lack of stimulation in response to octreotide (panel B) suggestive of neuropathic CIPO and consistent with the presumed pathology of diffuse intestinal ganglioneuromatosis. Colon manometry revealed no high-amplitude phasic contractions (panel C) with minimal response to bisacodyl stimulation (panel D).

- Vital signs were stable. Examination was notable for BMI 16, distended, tympanic abdomen with hyperactive bowel sounds, diffuse tenderness
- Labs were largely unremarkable. CT imaging showed marked distension of the entire colon and the small bowel without a definite transition point
- Extended gene analysis: heterozygous mutation in the RET gene, point mutation in exon 16 (M918/T) confirming MEN2B
- Endoscopic findings: normal upper endoscopy; gross dilation of the colon, and no stricture in the terminal ileum
- Antroduodenal manometry and colonic manometry (**Figure 1**) performed at the time of endoscopy were consistent with CIPO of neuropathic origin
- Started on prucalopride 2 mg daily with improvement of her symptoms



Diffuse intestinal ganglioneuromatosis
Bachiller MTH, Andrés JB, Pons F, et al. Diffuse intestinal ganglioneuromatosis an uncommon manifestation of Cowden syndrome. WJGO. 2013;5(2):34.

Discussion

- We present an unusual case of *MEN2B*-associated gastrointestinal involvement manifesting as CIPO
- Small intestinal dysmotility may need to be considered in the workup and management of gastrointestinal symptoms in *MEN2B* patients
- *MEN2B* classically presents with a pseudo-Hirschprung's type megacolon
- This case demonstrates that identification of chronic intestinal pseudo-obstruction, particularly in young patients, should consider evaluation for *MEN2B*

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