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Ganglioneuroma Masquerading as Rectal Polyp: Clinical Significance and Literature Review

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Abstract

Ganglioneuroma (GN) of colon is a rare tumor of gastrointestinal (GI) tract. GNs of GI tract are usually confined to colon particularly rectum. GNs are usually found incidentally but can be associated with MEN IIb, von Recklinghausen's neurofibromatosis or Cowden syndrome. A 21-year-old female with past medical history of bulimia, bipolar disorder and hypothyroidism, who presented with complaints of hematemesis and hematochezia. Her physical exam was unremarkable. She subsequently underwent an EGD and colonoscopy for further evaluation. EGD reveled mild gastritis. Colonoscopy revealed two 5 mm sessile polyps in the sigmoid colon and the rectum. Biopsy of the rectal polyp showed ganglioneuroma. This represents a case of polypoid ganglioneuromatosis. These polyps are endoscopically small sessile or pedunculated polyp indistinguishable from hyperplastic and adenomatous polyps. They are not associated with increased risk of colon cancer and don't require additional screening. On the other hand, ganglioneuromatous polyposis and diffuse ganglioneuromatosis can be associated with increased risk of colon and other cancers, and may require further work up.

Keywords: Ganglioneuroma; Gangliomatous polyposis; Diffuse ganglioneuromatosis; Gastrointestinal

Abbreviations: GN: Ganglioneuroma; GI: Gastrointestinal; GP: Gangliomatous Polyposis; DG: Diffuse Ganglioneuromatosis; CS: Cowden Syndrome; NCCN: National Comprehensive Cancer Network

Introduction

Ganglioneuroma (GN) of colon is a rare tumor of gastrointestinal (GI) tract, there are very few reported cases of this condition and incidence remains unknown. It is composed of ganglion cells, nerve fibers, Schwann cells and other supporting cells [1] GNs of GI tract are usually confined to colon particularly rectum but can be found in Upper GI tract [1,2]. GNs are usually found incidentally but can be associated with MEN IIb, von Reklinghausen's neurofibromatosis or Cowden syndrome [2-4].

Case Report

We present a case of 21-year-old female with past medical history of bulimia, bipolar disorder and hypothyroidism who presented with complaint of hematemesis and hematochezia. Her physical exam was unremarkable. She subsequently underwent an EGD and colonoscopy for further evaluation. EGD reveled mild gastritis. Colonoscopy revealed two 5 mm sessile polyps in the sigmoid colon and the biopsy of the rectal polyp showed ganglioneuroma. Biopsy of the sigmoid polyp showed non-specific changes. Image 1 shows the endoscopic view of the rectal polyp.

Discussion

GNs as mentioned in detail below are mainly divided into three categories.

1. Polypoid ganglioneuromatosis
2. Gangliomatous polyposis (GP)
3. Diffuse ganglioneuromatosis (DG) [1].

Endoscopically they are indistinguishable from adenomas or hyperplastic polyps [1]. Diagnosis is made by immunohistochemical staining which is positive for S-100, glial fibrillary acidic protein and vimentin (which stains spindle cell elements). Neuron specific enolase and neurofilament protein, confirm the presence of ganglion cells. Differentials are neurofibroma and Schwannoma but they don't contain ganglion cells [5].

Polypoid ganglioneuromatosis

Polypoid ganglioneuromatosis lesions are solitary or few in number, found incidentally on colonoscopy, endoscopically small sessile or pedunculated polyp indistinguishable from hyperplastic and adenomatous polyps. Patients are usually asymptomatic but may get symptomatic depending upon the lesion size.

They are not associated with increased risk of colon cancer and don't require additional screening [1].

Ganglioneuromatous polyposis (GP)

GP is characterized by multiple and often innumerable sessile or pedunculated mucosal or sub mucosal polypoid lesions [5-7]. Histologically there is greater variability in ganglionic, neural and supportive cell content and more demarcation when compared to polypoid ganglioneuroma but microscopically indistinguishable from polypoid GNs. GP can be associated with intestinal or extra intestinal manifestations like cutaneous lipomas, skin tags (achrochordon formation) [8], or with a syndrome such as Cowden syndrome (CS), juvenile polyposis, coexistent colonic adenoma or carcinoma and rarely von-Recklinghausen disease [1]. Patient can be symptomatic or asymptomatic and diagnosed incidentally.

Risk of colon cancer in this category is not firmly established and needs further evidence and reporting. Few cases of colon cancer have been reported in this subset of patients especially with Cowden syndrome (PTEN mutation) [3]. In one prospective study by Heald et al. [9] 13% patients with Cowden syndrome were associated with colorectal cancer [9], recent review suggests 16% lifetime risk of colon cancer. National Comprehensive cancer network (NCCN) recommends screening colonoscopy starting at the age of 40 and then every 2 years. Juvenile Polyposis (SMAD4 mutation) has also been associated with increased risk of colon cancer, which requires cancer screening starting from mid teens. NCCN recommends consideration of baseline esophagogastroduodenoscopy and colonoscopy at age 15 years then every year in patients with Juvenile polyposis. Several cases of non- Cowden Syndrome-related Ganglioneuromatous polyposis developing into adenocarcinoma have been reported, leading Kanter et al. [6] to suggest the condition to be considered premalignant. Discussion with patient about possible proctocolectomy can be considered on case-to-casebasis, as it is difficult to monitor for the development of an adenocarcinoma amongst innumerable polypoid lesions [4].

Diffuse ganglioneuromatosis (DG)

DG is characterized by poorly demarcated, mucosal or transmural infiltration of ganglioneuromatosis tissue with massive atypical proliferation of the myenteric plexus, often producing stricture like thickening of segments of bowel [4,6]. Most common symptoms reported are constipation or diarrhea [4]. DG may exist as an isolated finding or it is often observed as a component of von Recklinghausen's disease (NF1 gene mutation), MEN 2B (RET gene mutation), multiple intestinal neurofibromas or neurogenic sarcoma [1,4,10].

MEN-2B that results from mutation of RET proto-oncogene, is associated with modular thyroid cancer, pheochromocytoma, marfanoid habitus and mucocutaneous neuroma. Medullary carcinoma occur in 100% of patients and is aggressive and cause early metastasis so early diagnosis and surgery is essential [10,11]. Incidence of DG in patients with MEN 2B has been reported to approach 100 percent often antedating the characteristic medullary carcinoma, pheochromocytoma, and skeletal anomalies [10,11].

Chronic constipation or diarrhea or both are the most common symptoms associated with MEN 2B, these symptoms often begin in the first months of life, before the diagnosis of MEN 2B. Recognition of these symptoms can lead to early diagnosis if MEN 2B and potentially lifesaving thyroidectomy [4] von Recklinghausen syndrome is associated with neurofibrosarcoma.

We conclude that our patient had polypoid ganglioneuromatosis, which is not associated with increased risk of cancer. She didn't need any further follow up. Her symptoms may or may not be related to her polyps. It is important to differentiate this entity from others as we can avoid unnecessary surveillance colonoscopies in this subgroup of patients. On the other hand, ganglioneuromatous polyposis and diffuse ganglioneuromatosis can be associated with increased risk of colon and other cancers, and requires further work up.

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