Benign Colonic Schwann Cell Hamartoma in a Pediatric Patient with PTEN Hamartoma Tumor Syndrome (PHTS)

Naser Tolaymat MD
Lehigh Valley Health Network, Naser.Tolaymat@lvhn.org

Adam R. Paul DO
Lehigh Valley Health Network, Adam_R.Paul@lvhn.org

Published In/Presented At

Tolaymat, N. Paul, A. R. (2018, October 5). *Benign Colonic Schwann Cell Hamartoma in a Pediatric Patient with PTEN Hamartoma Tumor Syndrome (PHTS)*. poster Presented at: the (ACG) American College of Gastroenterology Annual Meeting, Philadelpgia, PA.

Benign Colonic Schwann Cell Hamartoma in a Pediatric Patient with PTEN Hamartoma Tumor Syndrome (PHTS)

Naser Tolaymat, MD, Adam Paul, DO
Children Hospital's at Lehigh Valley Health Network, Allentown, Pa.

INTRODUCTION

The phosphatase and tensin homolog on chromosome ten (PTEN) gene acts as a tumor suppressor through regulation of cell growth.[1] PTEN hamartoma syndrome (PHTS) represents a spectrum of disorders associated with germline mutations of PTEN tumor suppressor gene. These disorders include Cowden syndrome (CS), Bannayan-Riley-Ruvalcaba syndrome (BRRS), and Proteus syndrome (PS). These are rare conditions with prevalence estimated at 1 in 200,000. The GI polyps of the PHTS can show marked histologic variation displaying features of adenomatous, hamartomatous, and ganglioneuromatous polyps. Identification of PTEN mutation establishes diagnosis with certainty. The defining characteristic of PHTS is the development of hamartomas in multiple organ systems and an increase risk of malignancy including breast, thyroid, endometria, kidney, and possibly melanoma.[2] A recent systematic review found a high prevalence of colon polyposis with mixed histology, adenoma (40%), hyperplastic (43%), hamartomas (38%), inflammatory (25%) and ganglioneuroma (33%) polyps were found. More than 50% of patients have more than a single polyp histology.[3] There is increase prevalence of colorectal cancer in PHTS patients, the prevalence is estimated to be 13%, mean 47 (range 35–62) years.[3]

Schwannomas are neoplasms originating from Schwann cells, which are the cells forming nerve sheets. They generally involve peripheral nerves and rarely affect the gastrointestinal tract. A recent literature review reported a 96 patients with colorectal schwannoma, mean age 61.5 years. Three patients were 18 years or youngers; 14 year old with melena and CT revealing a sigmoid mass, an 18 year old with cecal intussusception, found to have a tumor measures 5 cm and an 18 Year old malignant Schwannoma arised from sigmoid colon.[4,5] Most schwannomas in adults are discovered incidentally on screening endoscopy or during abdominal imaging. Rectal bleeding and fecal occult blood were the most common presenting symptoms (28%), followed by abdominal pain (15%). However, asymptomatic patients account for 28% of all cases. In 3% of patients Schwannoma was malignant.[4]

One case report of choroidal schwannoma in a 10 year old with PHTS has been reported[6] in the literature.

CASE PRESENTATION

We report a case of a 12 year old male who presented with symptoms of painless rectal bleeding, and abdominal pain. He had normal CBC, negative stool infectious panel, Fecal Calprotectin was elevated at 708 ug/gm. His EGD reveled normal esophageal and gastric mucosa, duodenal mucosa appeared nodular but histology was normal. Colonoscopy revealed large number of small polyps in the colon sparing the ascending colon (figures 1–2). Several large polyps were present in distal sigmoid colon (figures 3–4). 7 polyps (5 large) were removed. Pathology was consistent with juvenile polyps.

Two weeks later he presented to the emergency department with rectal bleeding, he underwent a subsequent colonoscopy which revealed many polyps and few prominent polyps in sigmoid colon and rectum. A total of 8 polyps were removed and pathology was consistent with juvenile polyps. A small bowel fluoroscopy was ordered 1 month later and was reported to be normal.

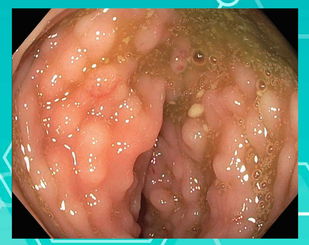

FIGURE 1

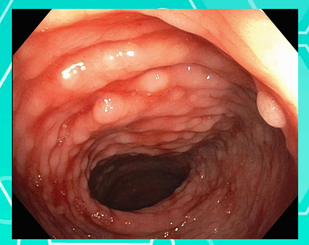

FIGURE 2

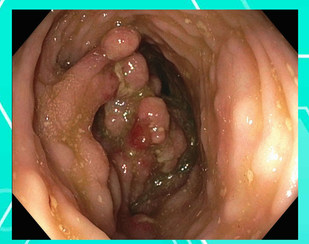

FIGURE 3

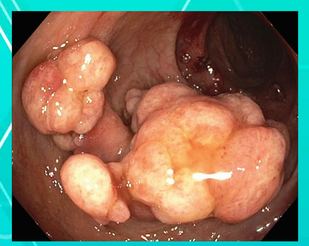

FIGURE 4

He underwent a subsequent colonoscopy and polypectomy 6 months later. A total of 7 polyps (one medium) were removed and again pathology was consistent with juvenile polyps except for one polyp was described as colonic mucosa with spindle cell proliferation, positive with immunohistochemical panel (S-100 protein), and compatible with Schwannoma.

He was followed up with a genetic consultation based on his presumptive clinical diagnosis of juvenile polyposis syndrome. He underwent genetic testing for BMPR1A and SMAD4 gene mutations but was negative. He subsequently underwent reflex testing with ColoNext panel that tested for 17 additional genes associated with colon polyps and/or increased risk of colon cancer. He was found to be positive for a pathogenic mutation in the PTEN gene, specifically p.R130Q (c.389G>A). Pathogenic mutations in the PTEN gene are associated with PTEN hamartoma tumor syndrome (PHTS) associated with the spectrum of disorders including CS, BRRS, and PS.

CONCLUSION

In conclusion this case report describes a pediatric patient originally diagnosed with JPS and found to have an isolated polyp with pathology consistent with benign schwann cell hamartoma. The patient was subsequently found to have PTEN mutation upon genetic testing. Although PHTS is described as having a high prevalence of colon polyposis with multiple histology types including hyperplastic, adenoma, hamartoma, ganglioneuroma, and inflammatory polyps it has never been described in the literature as being associated with mucosal neuromas. Our patient was found to have an isolated benign schwann cell hamartoma (mucosal neuroma). To our knowledge this is the first reported description of benign colonic schwann cell hamartoma in the pediatric population especially in the context of PHTS. In reported adult cases schwann cell hamartomas of the GI tract have been documented as occurring as isolated entities that are not associated with any genetic polyposis syndrome. It is unclear whether in our patient the benign schwann cell hamartoma is part of a newly described histological presentation of PHTS or whether it is an isolated finding in the setting of concurrent PHTS.

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