THE UNIVERSITY OF ARIZONA
COLLEGE OF MEDICINE TUCSON
Internal Medicine Residency

Mucosal Schwann Cell Hamartoma of the Sigmoid Colon in a Transgender Female to Male on Testosterone Therapy: A Case Report

Loveland, M. M.D., Alameri, A. M.D., Bach L. D.O.
University of Arizona College of Medicine - Tucson

Banner University Medicine

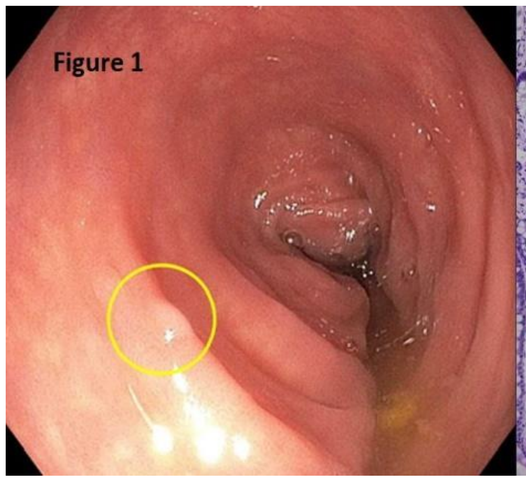
Introduction

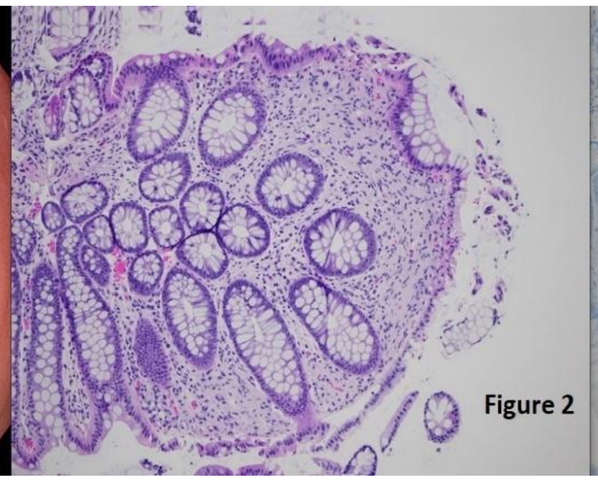
Mucosal Schwann cell hamartomas of the gastrointestinal tract are rare benign mesenchymal neoplasms, of which the exact incidence within the population is unknown. First described as a new entity in 2009 by Gibson and Hornick[1] in a retrospective case study examining pathology from 26 patients, it is suspected that the increase in incidence of colonic mucosal Schwann cell hamartomas is due to the increase of colorectal cancer screening. As of 2020, there have been 41 documented cases of Schwann cell hamartomas[2].

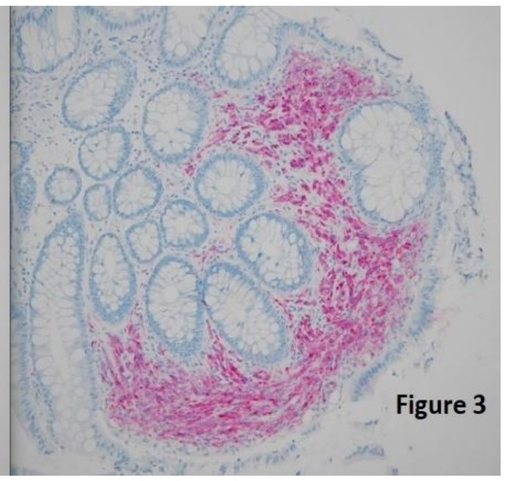
Case Presentation

A 63 year old transgender female to male on testosterone therapy for 10 years and history of adenomatous colon polyps 3 years prior presented for outpatient surveillance colonoscopy. The patient had no personal or family history of neurofibromatosis type 1 (NF1), Cowden syndrome, or multiple endocrine neoplasia (MEN) type 2b. Colonoscopy was notable for a 1mm sessile polyp in the sigmoid colon (Figure 1) and 4mm sessile ascending colon polyp. Review of the pathology demonstrated a tubular adenoma without dysplasia in the ascending colon, and in the sigmoid colon S-100 positive spindle cells with elongated nuclei and dense eosinophilic cytoplasm, consistent with a mucosal Schwann cell hamartoma (Figures 2 & 3).

Figures/Pictures







(Figure 1) Endoscopic finding of a 1 mm sigmoid colon polyp
(Figure 2) Histologic features show spindle cell proliferation, elongated nuclei, and eosinophilic cytoplasm under hematoxylin and eosin-stained sigmoid colonic mucosa. This is consistent with a mucosal Schwann cell hamartoma.
(Figure 3) Immunochemistry for S-100 spindle cells within the sigmoid colon. This stain further solidified the diagnosis of Schwann cell hamartoma.

Discussion

Our patient presented for routine surveillance colonoscopy and was found to have a rare benign mesenchymal neoplasm of the sigmoid colon, a Schwann cell hamartoma. Mucosal Schwann cell hamartomas are typically small, 1-8mm, primarily located in the left colon, have a female predominance and an average age at presentation of 62[3]. They are not associated with malignancy or inherited syndromes[1,4]. The histological differential diagnosis includes schwannoma, neurofibroma, mucosal neuroma, ganglioneuroma, ganglioneurmatosis, perineurioma and GIST, making accurate diagnosis key to avoid unnecessary treatments[4]. Interestingly, our patient found to have a mucosal Schwann cell hamartoma of the sigmoid colon is a transgender female to male on testosterone therapy, which to our knowledge has not previously been reported. Recent studies in animal models have suggested testosterone may promote the growth of colorectal adenomas[5]. The role of sex hormones in development of these neoplasms has yet to be explored.

References

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Conclusions

The exact prevalence of GI tract Schwann cell hamartomas within the general population is unknown. As transgenderism becomes more prevalent today, the role of gender confirming hormone therapy in colonic polyposis formation is poorly understood. This case report provides a useful framework to further explore the role that it may have in such disease processes.