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VIDEO CAPSULE ENDOSCOPY IN PATIENTS WITH MUIR-TORRE SYNDROME

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Introduction: Muir-Torre Syndrome (MTS) is a rare, primarily autosomal dominant disorder that is distinguished by having sebaceous skin malignancies in addition to visceral malignancies. The most common form of MTS is a variant of HNPCC. Our aim is to demonstrate the utilization of VCE in patients with MTS as the first line screening method.

Methods: Single center, retrospective chart review study of outpatients with MTS who underwent a video capsule endoscopy study between January 2006 and January 2016.

Results: Four patients, all women and mean age of 57 years old, with MTS underwent a video capsule endoscopy at our institution. In 75% of the patients, VCE detected polyps at a point in the small bowel which upper endoscopy and colonoscopy did not visualize. Two patients had large jejunal polyps, approximately 20mm in diameter. One patient had multiple 3-20mm sessile polyps from the duodenum to the ileum. On endoscopy, only one of the patients had a polyp detected and it was a 10mm polyp in the stomach which was positive for GIST. With regards to colonoscopy, 2 of the patients had critical findings of colonic mucosa with focal adenomatous changes and cryptitis (high grade dysplasia) and T1 poorly differentiated signet cell carcinoma respectively. Both of these patients underwent total colectomies. Cumulatively, all four of these patients have undergone 17 endoscopies/colonoscopies.

Conclusion: MTS is a disorder that needs to be monitored closely as patients have a high propensity of developing gastrointestinal malignancies. Current recommendations are colonoscopies annually starting at ages 20-25 and endoscopies with gastric antrum biopsies starting at ages 30-35. 75% of the patients had lesions in the small bowel that were only picked up by VCE. Failure to detect asymptomatic advanced lesions in the small bowel may have serious consequences. We therefore recommend pan-endoscopy in this rare syndrome.

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