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A Rare Case of Muir Torre Syndrome

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1. Abstract

Muir Torre Syndrome (MTS) is a rare autosomal dominant phenotypic variant of hereditary nonpolyposis colorectal carcinoma (HNPCC). It is characterized by the presence of at least one visceral malignancy and one sebaceous skin tumor. Patients have a strong family history of primary cancers and the Amsterdam criteria or the Mayo Muir Torre Risk Score are frequently used to diagnose it. We present a case of a 58 years old who initially presented with a request to repair two incisional hernias following a procedure at another institution. Past family history was remarkable for strong history of colon, lung, and skin cancer in his father and 3 brothers. On CT, the patient was found to have a mass in the ascending colon and the prostate suggesting primary colon carcinoma and prostate adenocarcinoma. The patient was found to have a 5cm lesion on his left upper back, which was excised, biopsied and found to be sebaceous carcinoma. Given the constellation of sebaceous skin and visceral tumors, and after the pathological study of the malignancies a diagnosis of Muir Torre Syndrome was made. Muir Torre Syndrome is a rare phenotypic variant of HNPCC associated with a poor prognosis. We present an interesting case of Muir Torre Syndrome that was diagnosed due to a detailed medical history and physical exam. This case highlights the need for thorough family history, physical examination, and screening for suspected malignancies.

2. Introduction

Muir Torre Syndrome (MTS) is a rare autosomal dominant phenotypic variant of hereditary nonpolyposis colorectal carcinoma (HNPCC). It is characterized by the presence of at least one visceral malignancy and one sebaceous skin tumor. Patients typically have a strong family history of primary cancers and the Mayo Muir Torre Risk Score (Table 1) os the Amsterdam criteria (Table 2) are frequently used to diagnose it. A 58-year-old male with past medical history of colonic perforation during colonoscopy at another institution that resulted in a laparotomy and segmental colon resection. He presented with abdominal pain, constipation, and urinary frequency at our general surgery clinic. The patient requested repair of an incisional umbilical hernia and a right lower quadrant hernia that had been present for the last three years. The patient was found to have a strong family history of cancer. His father and three brothers died at young ages from colon, lung, and skin cancer. On examination, he had significant distortion in the right lower quadrant with a 5 cm bulging and a second bulging near the umbilicus approximately 5cm. Both hernias were non reducible. Given the concern for his strong family history the patient was referred for colonoscopy. Biopsies obtained during colonoscopy were diagnostic for poorly differentiated adenocarcinoma in the ascending colon and sigmoid colon. Finding on the CT scan indicated a mass in the ascending colon suggesting primary colon carcinoma and enlarged mesenteric and iliac lymph nodes. Findings on the PET scan exhibited increased metabolic activity in the pericolonic lymph nodes in the ascending colon and bilateral pelvic lymphadenopathy. These findings were consistent for metastatic colon cancer. Genetic testing revealed a deletion in the MSH2 exon and a germline mutation in MUTYH. In subsequent clinic visits, the patient was found to have a 5cm lesion on his left upper back, which was excised, biopsied, and found to be sebaceous carcinoma. The patient stated that he had lost 8lbs recently and felt more fatigued than usual. The pathology from the sebaceous carcinoma returned CK5/6+, CK7+, AR+, and MLH1+. A diagnosis of Muir Torre was then made. The patient was also found to have prostate cancer with lymph node invasion, confirmed by biopsy and elevated prostate specific antigen (PSA) (PSA= 400). The prostate cancer was deemed non-operable and was treated with hormonal therapy. A subtotal colectomy with ileorectal anastomosis and small bowel resection was performed to manage the colon

carcinomas, and two incidental small bowel adenocarcinomas that were found at laparotomy. The patient was deemed a candidate for adjuvant chemotherapy, but after discussing with his family, he declined to pursue chemotherapy. Two and a half years following the subtotal colectomy, he entered hospice care and was lost to follow up.

Table 1: Mayo Muir Torre Risk Score. A score >2 is highly predictive of MTS.

Variable	Score	Patient Score
Age at diagnosis of first sebaceous neoplasm		
≥60years	0	
<60 years	1	1
Number of Sebaceous Neoplasms		
<2	0	0
≥2	2	
Personal history of HNPCC-related cancer		
No	0	
Yes	1	1
Family history of any HNPCC-related cancer		
No	0	
Yes	1	1
Total MTS risk score		3

Table 2: Amsterdam Criteria.

Amsterdam Criteria
Three or more relatives on the same side of the family with histologically verified Lynch-associated cancers, one of whom is a first-degree relative of the other two and in whom familial adenomatous polyposis (FAP) has been excluded
Lynch-associated cancers involving at least **two** generations.
One or more cancers diagnosed before the age of 50 years.

3. Discussion

MTS is a rare phenotypic variant of HNPCC, or Lynch Syndrome, consisting of at least 1 visceral and 1 cutaneous malignancy [1,2]. In 2004, there were only 200 total cases of MTS documented worldwide [3]. Alterations in the DNA mismatch repair genes MLH1, MLH2, and MSH2 results in microsatellite instability and the manifestations of malignancies [4-6]. The Mayo Muir Torre Risk score was developed to identify patients who are at high risk of being diagnosed with MTS [7]. Based on this risk score, our patient would receive 3 points and be placed at high risk for MTS (Table 1). Studies have shown that a surveillance interval of 1-2 years in families with Lynch Syndrome decreases the risk of developing colorectal carcinoma than with surveillance intervals of 2-3 years [8]. Genetic testing is an important component of the workup for these patients. In this case, genetic tests revealed a deletion in the MSH2 exon, which is common in MTS, and a germline mutation in the MUTYH. Mutations in MUTYH lead to abnormal protein production and function and are associated with an increased risk of developing colorectal carcinoma [9,10]. Additionally, immunohistochemistry (IHC) can be used to identify cases of MTS in suspected patients [11,12]. In this case, the visceral malignancies were identified and then the sebaceous carcinoma was discovered, keying in on MTS as the diagnosis. Sebaceous carcinomas themselves are rare and have high rates of recurrence and metastasis [13]. It is recommended that any patient with a sebaceous carcinoma and a family history of cancer be screened for internal malignancies [14]. The most common extracolonic malignancies seen in Lynch Syndrome are endometrial, ovarian, stomach, small bowel, and pancreatic/biliary tract tumors [15]. While prostate cancer is not a commonly seen malignancy in Lynch Syndrome, it has a clear etiology to the microsatellite instability seen in Lynch Syndrome [16, 17]. Males with Lynch Syndrome has approximately a five-fold increase in risk of developing prostate cancer compared to the general population [18]. In cases of Lynch Syndrome, screening colonoscopies are recommended every one to two years beginning at age 25 or five years before the earliest age of colon cancer in the family. Screening colonoscopies have been shown to decrease mortality in patients diagnosed with Lynch Syndrome [19]. Colonoscopies performed in Lynch Syndrome patients should include meticulous inspection and precise removal of all polyps, specifically polyps that are flat and located in the ascending colon [20]. Additionally, complete colonoscopies with adequate bowel preparation and chromoendoscopy use are associated with improved adenoma detection in patients with Lynch Syndrome [21]. As our patient had a family history of Lynch Syndrome, screening colonoscopies should have been started at age 25 and may have diagnosed his disease at an earlier age.

4. Conclusion

Muir Torre Syndrome is a rare phenotypic variant of HNPCC associated with a poor prognosis. We present a case of an interesting case of a male with Muir Torre Syndrome who was diagnosed due to a detailed medical history and physical exam. This case highlights the need for thorough family history, physical examination, and screening for suspected malignancies when interviewing our patients.

5. Lessons Learned

Further investigation of the skin following the presence of multiple visceral malignancies is imperative in the diagnosis of Muir Torre Syndrome.

References

1. John AM, Schwartz RA. Muir-Torre syndrome (MTS): An update and approach to diagnosis and management. J Am Acad Dermatol. 2016; 74: 558-566.
2. Muir EG, Bell AJY, Barlow KA. Multiple primary carcinomata of the colon, duodenum, and larynx associated with kerato-acanthomata of the face. Br J Surg. 1967; 54: 191-195.
3. Curry ML, Eng W, Lund K. Muir-Torre syndrome: role of the dermatopathologist in diagnosis. Am J Dermatopathol. 2004; 26: 217-221.
4. Honchel R, Halling KC, Schaid DJ. Microsatellite instability in Muir-Torre syndrome. Cancer Res. 1994; 54: 1159-1163.
5. Bapat B, Xia L, Madlensky L. The genetic basis of Muir-Torre syndrome includes the hMLH1 locus. Am J Hum Genet. 1996; 59: 736-739.
6. Godard V, Coulet F, Bernaudin JF. [Mutation in the MSH2 gene in Muir-Torre syndrome]. Ann Dermatol Venereol. 1999; 126: 600-603.
7. Roberts ME, Riegert-Johnson DL, Thomas BC. A clinical scoring system to identify patients with sebaceous neoplasms at risk for the Muir-Torre variant of Lynch syndrome. Genet Med. 2014; 16: 711-716.
8. Vasen HFA, Abdirahman M, Brohet R. One to 2-Year Surveillance Intervals Reduce Risk of Colorectal Cancer in Families With Lynch Syndrome. Gastroenterology 2010; 138: 2300-2306.
9. Win AK, Cleary SP, Dowty JG. Cancer risks for monoallelic MUTYH mutation carriers with a family history of colorectal cancer. Int J Cancer 2011; 129: 2256-2262.
10. Lubbe SJ, Di Bernardo MC, Chandler IP, Houlston RS. Clinical Implications of the Colorectal Cancer Risk Associated With MUTYH Mutation. JCO. 2009; 27: 3975-3980.
11. Kaliyappan K, Palanisamy M, Duraiyan J, Govindarajan R. Applications of immunohistochemistry. J Pharm Bioall Sci. 2012; 4: 307.
12. Torre K, Ricketts J, Dadras SS. Muir–Torre Syndrome: A Case Report in a Woman Without Personal Cancer History. The American Journal of Dermatopathology. 2019; 41: 55-59.
13. Kyllo RL, Brady KL, Hurst EA. Sebaceous carcinoma: review of the literature. Dermatol Surg. 2015; 41: 1-15.
14. Tsalis K, Blouhos K, Vasiliadis K. Sebaceous gland tumors and internal malignancy in the context of Muir-Torre syndrome. A case report and review of the literature. World J Surg Onc. 2006; 4: 8.
15. Koornstra JJ, Mourits MJ, Sijmons RH. Management of extracolonic tumours in patients with Lynch syndrome. The Lancet Oncology. 2009; 10: 400-408.
16. Soravia C, van der Klift H, Bründler M-A. Prostate cancer is part of the hereditary non-polyposis colorectal cancer (HNPCC) tumor spectrum: Prostate Cancer in HNPCC. Am J Med Genet. 2003; 121A: 159-162.
17. Bauer CM, Ray AM, Halstead-Nussloch BA. Hereditary prostate cancer as a feature of Lynch Syndrome. Familial Cancer. 2011; 10: 37-42.
18. Haraldsdottir S, Hampel H, Wei L. Prostate cancer incidence in males with Lynch syndrome. Genet Med .2014; 16: 553-557.
19. Engel C, Rahner N, Schulmann K. Efficacy of annual colonoscopic surveillance in individuals with hereditary nonpolyposis colorectal cancer. Clin Gastroenterol Hepatol. 2010; 8: 174-182.
20. Niv Y, Moeslein G, Vasen H. Quality of colonoscopy in Lynch syndrome. EIO 2014; 02: E252-E255.
21. Sánchez A, Roos VH, Navarro M. Quality of colonoscopy is associated with adenoma detection and post-colonoscopy colorectal cancer prevention in Lynch syndrome. Clinical Gastroenterology and Hepatology. 2020: S1542356520315147.