ORIGINAL RESEARCH PAPER
Volume - 13 | Issue - 08 | August - 2024 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

MUIR -TORRE SYNDROME – A RARE FAMILIAL COLONIC CANCER SYNDROME – A RADIOLOGICAL OVERVIEW

Radio-Diagnosis

Dr. Harikrushna G. Bhatt 3rd Year Resident Doctor, Department Of Radio-diagnosis, GCRI, BJ Medical College, Ahmedabad.

Dr. Ishan R. Maitreya* 2nd Year Resident Doctor, Department Of Radio-diagnosis, GCRI, BJ Medical College, Ahmedabad *Corresponding Author

Dr. Himanshu C. Soni Professor, Department Of Radio-diagnosis, GCRI, BJ Medical College, Ahmedabad.

ABSTRACT

Muir-Torre syndrome (MTS) is an autosomal dominant condition characterized by cutaneous neoplasms in addition to internal malignancies. Cutaneous manifestations consist of recurring sebaceous adenomas and keratoacanthomas. Visceral malignancies commonly observed include those affecting the colorectal, gynecological, and urological systems. This syndrome represents a variation of the hereditary non-polyposis colorectal carcinoma syndrome (HNPCC). The fundamental genetic etiology involves heritable mutations in the mismatch repair (MMR) genes, resulting in microsatellite instability (MSI) and an elevated susceptibility to malignancy development. Herein, we present a radiological case study of a 73-year-old male patient presented with bleeding per rectum.

KEYWORDS

Muir Torre Syndrome, Rectal Adenocarcinoma , sebaceous adenoma , multiple large bowel polypoidal lesions

INTRODUCTION

Muir-Torre syndrome (MTS) was initially delineated independently by Muir [1] in 1967 and Torre [2] in 1978. This condition represents a rare disorder characterized by the coexistence of one or more sebaceous gland neoplasms along with at least one internal malignancy. It is postulated to represent a phenotypic variation of Lynch syndrome or hereditary nonpolyposis colorectal carcinoma (HNPCC) due to their shared molecular pathogenic mechanisms. Both MTS and HNPCC syndrome result from mutations occurring in the DNA mismatch repair (MMR) genes, leading to the accumulation of genomic errors and subsequent cancer development [3,4]. Approximately two-thirds of MTS instances do not exhibit MMR defects and are classified within MTS II [5]. The mode of inheritance is autosomal dominant, although MTS II cases are more frequently observed as sporadic occurrences [6]. Sebaceous adenomas and keratoacanthomas are the most frequently observed cutaneous malignancies in this cohort [7]. The predominant visceral malignancies linked with Muir-Torre syndrome (MTS) are colorectal carcinomas.

Case Presentation

Presenting a case of a 73 years old male patient presented with lethargy, weakness and weight loss for a couple of months. On detailed questioning, the patient gave a history of bleeding per rectum for a few months. Patient was sent for radiological evaluation with clinical suspicion of colonic malignancy. On clinical examination multiple non ulcerative nodules in different parts of the body were noted. Patient underwent a CT scan and MRI, and other basic hematological investigation- as per the institute's protocol .

On CECT imaging of the abdomen and pelvis region, presence of heterogeneously enhancing asymmetrical circumferential wall thickening involving lower rectum and anorectal junction was noted. Lesion minimally extended into upper part of anal canal. Lesion showed extra serosal spread. Lesion involved bilateral levator ani muscles. Presence of few perilesional nodes were noted. Findings were suggestive of malignant involvement of lower rectum. (Figure 1C)CT scan also demonstrated few homogeneously mildly enhancing well defined soft tissue density lesions in subcutaneous plane in gluteal region and anterior abdominal wall. Largest one was noted at anterior abdominal wall. Possibility of benign cutaneous lesions – sebaceous adenoma appeared more likely. (Figure 1A, 1B)

MR images of the same patient showed thickening (hypointense on T1W, intermediate to hypointense on T2W & not suppressed on STIR with post contrast enhancement) involving lower rectum and adjacent anorectal junction and anal canal. (Figure 2). Lesion showed restriction on DWI. Significant perilesional fat stranding and few perilesional nodes were noted. Lesion extended into mesorectal fat on the left side. Lesion involved mesorectal fascia anteriorly. (EMVI Grade II present). Luminal narrowing & proximal dilatation was noted. MR images also demonstrated presence of multiple polypoidal lesion involving visualized upper rectum and sigmoid colon, average size of 6 mm, which appeared hypointense on T1W, hyperintense on T2W & showed homogeneous post-contrast enhancement (Figure 2A, 2C)

Figure 1:-Axial post contrast CT scan images of pelvic region

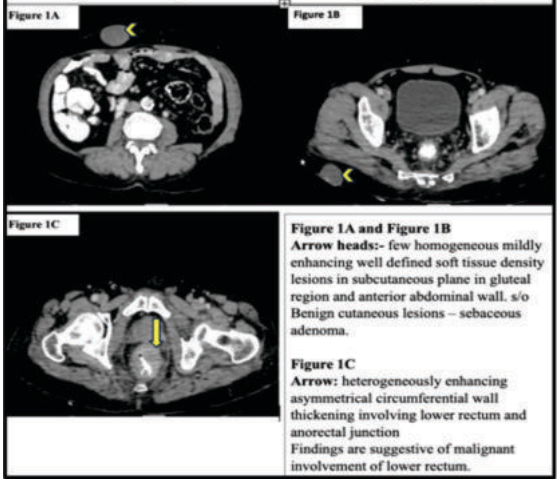
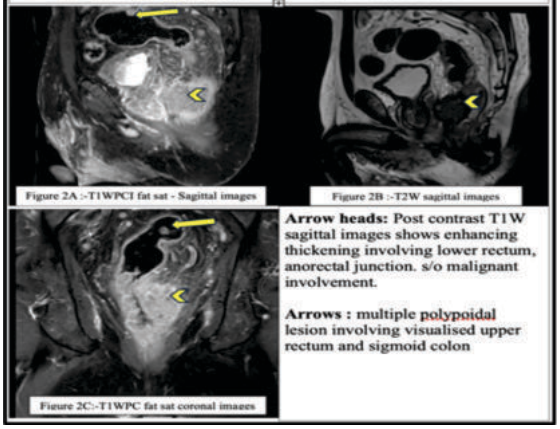


Figure 1A and Figure 1B
Arrow heads:- few homogeneous mildly enhancing well defined soft tissue density lesions in subcutaneous plane in gluteal region and anterior abdominal wall. s/o Benign cutaneous lesions – sebaceous adenoma.

Figure 1C
Arrow: heterogeneously enhancing asymmetrical circumferential wall thickening involving lower rectum and anorectal junction
Findings are suggestive of malignant involvement of lower rectum.

T2W sagittal images and T1W post contrast coronal and sagittal images of rectum



Arrow heads: Post contrast T1W sagittal images shows enhancing thickening involving lower rectum, anorectal junction. s/o malignant involvement.

Arrows : multiple polypoidal lesion involving visualised upper rectum and sigmoid colon

Biopsy from the rectal lesion -positive for Adenocarcinoma, Well differentiated type arising from a villous adenoma

DISCUSSION

Clinically, the diagnosis of Muir-Torre syndrome (MTS) is established through the identification of a minimum of one sebaceous gland tumor (such as adenoma, epithelioma, carcinoma, cystic sebaceous tumor, or keratoacanthoma with sebaceous differentiation) concomitant with at least one primary visceral malignancy. Alternatively, the diagnosis of MTS may be confirmed in cases where the individual presents multiple keratoacanthomas alongside numerous internal malignancies and a familial background of MTS. The visceral malignancies associated with MTS predominantly affect the colon in both genders, with fifty-one percent of patients developing at least one colorectal carcinoma which occurs approximately ten years earlier than in the general population, while endometrial cancer is prevalent among females. Internal malignancies in MTS cases are commonly multiple, characterized by low grade features and exhibit a relatively low propensity for metastasis.

Imaging plays a crucial role in early detection of the internal malignancy. CT scan helps to rule out distant spread of the disease and MR helps to evaluate the local spread of the disease. In our case, we suggested a possibility of the hereditary colonic cancer syndrome- "Muir Torre Syndrome" on the basis of the clinical and imaging findings.

CONCLUSION

Muir-Torre syndrome, a seldom encountered autosomal dominant disorder linked to Lynch syndrome, is correlated with mutations in the DNA mismatch repair (MMR) genes, notably MSH2. The diagnostic criteria entails the existence of at least one sebaceous malignancy alongside at least one visceral malignancy. Imaging plays a crucial role in early diagnosis and evaluation of exact extent of the disease.

REFERENCES

1. Muir EG, Bell AJ, Barlow KA: Multiple primary carcinomata of the colon, duodenum, and larynx associated with kerato-acanthomata of the face. Br J Surg. 1967, 54:191-5. 10.1002/bjs.1800540309
2. Torre D: Multiple sebaceous tumors. Arch Dermatol. 1968, 98:549-51.
3. Abbas O, Mahalingam M: Cutaneous sebaceous neoplasms as markers of Muir-Torre syndrome: a diagnostic algorithm. J Cutan Pathol. 2009, 36:613-9. 10.1111/j.1600-0560.2009.01248.x
4. Machin P, Catasus L, Pons C, et al.: Microsatellite instability and immunostaining for MSH-2 and MLH-1 in cutaneous and internal tumors from patients with the Muir-Torre syndrome. J Cutan Pathol. 2002, 29:415-20. 10.1034/j.1600-0560.2002.290705.x
5. Perera S, Ramyar L, Mitri A, et al.: A novel complex mutation in MSH2 contributes to both Muir-Torre and Lynch Syndrome. J Hum Genet. 2010, 55:37-41. 10.1038/jhg.2009.119
6. Ponti G, Ponz de Leon M: Muir-Torre syndrome. Lancet Oncol. 2005, 6:980-7. 10.1016/S1470-2045(05)70465-4
7. John AM, Schwartz RA: Muir-Torre syndrome (MTS): an update and approach to diagnosis and management. J Am Acad Dermatol. 2016,74:558-66. 10.1016/j.jaad.2015.09.074