Check for updates

OPEN ACCESS

DOI: 10.46903/gjms/23.1.Special.1816

CASE REPORT

AN UNUSUAL PRESENTATION OF WHIPPLE'S DISEASE: A YOUNG MALE PRESENTING WITH CHRONIC DIARRHEA

Rashid Ali[1], Madiha Ashraf[2], Asma Sikander[1], Nageen Naz[1], Amema Hafeez[3], Maira Jabbar Chaudhary[3], Fariha Sahrish[3], Mahrukh Afzal[1], Alia Ahmad[4], Mehwish Khan Niazi[3], Sara Masood Cheema[3], Rabia Afzal[3]

Departments of Gasteroenterology, Sir Ganga Ram Hospital, Lahore, [2]Tehsil Head quarter Hospital, Safdrabad, Sheikhupura, [3]Azra Naheed Medical College, Superior University, Lahore, [4]The university of Child Health sciences and the Children's Hospital, Lahore,

ABSTRACT

WD is a rare, multisystem, chronic infection caused by the Tropherima whipplei primarily involving the small intestine. The patient presents with diarrhea, weight loss, abdominal pain, and fever. The series of endoscopic evaluation, histopathology, and PCR assays are essential, however, histopathologically, the presence of sheets of foamy macrophages is the key suspicion leading to diagnosis. An 18-year-old boy presented with severe abdominal pain accompanied by severe diarrhea with episodes of intermittent constipation. He underwent a series of gastro-endoscopic and colonoscopic examinations. The tissue blocks after reviewing showed sheets of macrophages in lamina propria suggesting a provisional diagnosis of Whipple's disease. The patient was treated by injection of Oxidil 2g OD for two weeks and Septran DS BD. This treatment led to the improvement of symptoms i.e., diarrhea and weight loss. In the resource-limited setting, where repetition of tissue block review leads to exhaustion and the economic burden of repetitive endoscopic procedures on patients is laborious. The presence of foamy macrophages plays a pivotal role in patient management, ultimately leading to symptomatic improvement.

Keywords: Diarrhea; Foamy Macrophages; Malabsorption; Whipple's disease.

Cite as: Ali R, Ashraf M, Sikander A, Naz N, Hafeez A, Chaudhary MJ, et al. An unusual presentation of whipple's disease: a young male presenting with chronic diarrhea [case report]. Gomal J Med Sci 2025 Jan-Mar;23(1) Special:187-9. https://doi.org/1046903/gjms/23.1.Special.1816

INTRODUCTION

Malabsorption syndrome is a disease affecting the small intestine usually, due to defective absorption through microvilli or defective digestive enzymes, or indigestion.[1] There are many causes of malabsorption including celiac disease (CD), food allergies, Lactose intolerance (LI), and Whipple's disease (WD).[2] WD was rendered as intestinal lipodystrophy in 1907. WD is a rare, multisystem, chronic infection caused by the actinomycete Tropherima whipplei (T. whipplei).[3] WD primarily involves the small intestinal tract, causing diarrhea, weight loss, abdominal pain, and fever. However, extra-intestinal sites include joints, the musculoskeletal system, cardiac valves, the brain, eyes, lungs, and skin.[4] In 1961 the organism T. whipplei was detected in the electron microscope. Later on, a specific polymerase chain reaction (PCR) assay targeting T. whipplei 16S ribosomal RNA (rRNA) genes was detected.[5] Currently, it is detected by immunohistochemical staining and various diagnostic PCR assays.[6] The series of endoscopic evaluation, histopathology, and PCR assays are essential in the evaluation of WD, however, histopathologically, the presence of sheets of foamy macrophages is the most reliable diagnostic method.[7]

Corresponding Author:

Dr. Fariha Sahrish
Assistant Professor, Department of Pathology
Azra Naheed Medical College, Superior University
Lahore, Pakistan

E-mail: awan.fariha44@gmail.com

Date Submitted: 21-10-2024
Date Revised: 13-03-2025
Date Accepted: 25-03-2025

CASE REPORT

An 18-year-old boy was referred to our Gastroenterological Unit at Karimia Medical Complex on March 2024 due to non-shifting and non-radiating severe abdominal pain accompanied by severe diarrhea. There was a positive history of intermittent constipation with alternate loose stools. No complaints of fever, fatigue, and arthralgia were provided. Abdominal palpation was unremarkable. No cardiovascular, or neurologi-

cal deficits were found. His past medical history was completely unremarkable. His complete blood picture showed normal hemoglobin, increased platelet count, and lymphocytic leukocytosis. His C-reactive protein (CRP) was positive. The rest of the baseline investigations were within normal limits. His USG abdomen showed no specific pathology; however, 29 mm-sized mesenteric and para-aortic lymph nodes were detected and biopsied. The histopathology report showed reactive lymphoid hyperplasia only.

His upper GI endoscopy done on 3rd April 2024 showed edematous inflamed mucosa in cardia, fundus, body, and antrum of the stomach aligned with histopathological findings of chronic gastritis. The duodenum showed edematous mucosa in the cap and descending segment, (Fig 1A-B) however, histopathological findings showed complete villous atrophy and mild inflammation in lamina propria consistent with celiac disease. His colonoscopic examination showed a deformed ileocecal valve and an impression of IBD was made, however, Fecal Calprotectin turned out negative. In May 2024, the patient was given a gluten challenge for 3 months afterward but no clinical improvement in diarrhea was noted. In keeping with the clinical scenario, the tissue blocks were reviewed again by two different consultant histopathologists as CD was not suspected clinically. The in-house; and Agha Khan Histopathologist reported sheets of macrophages in lamina propria suggesting Whipple's disease. (Fig. 1C-F). Unfortunately, due to the repetition of review, special stains i.e., PAS and PASD were unable to perform due to block exhaustion. After the histopathology report, the patient was managed in the line of WD. The patient was treated by injection of Oxidil 2g OD for two weeks and Septran DS BD. This treatment led to the improvement of symptoms i.e., diarrhea and weight loss.

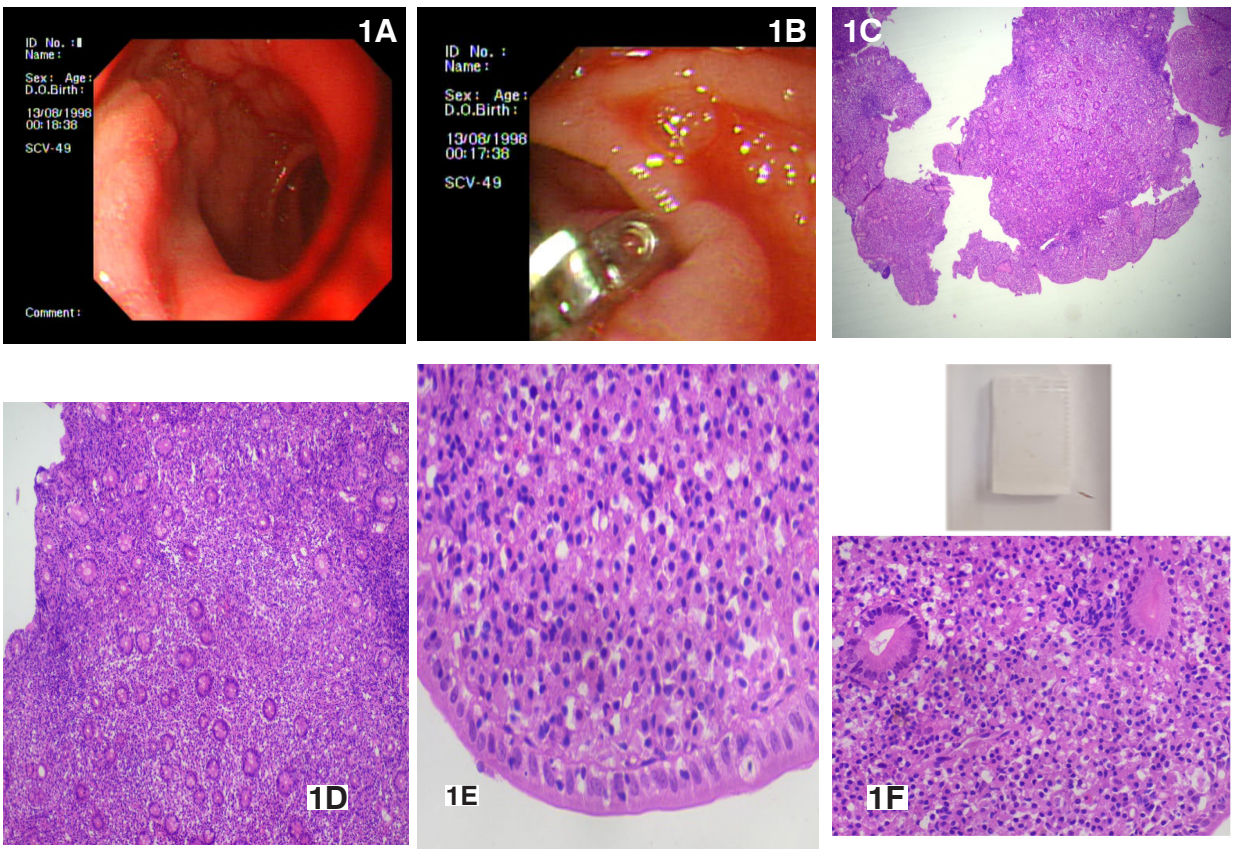

Fig 1A-F Endoscopic images and H&E stained tissue sections of small bowel at 20x and 40X

Fig 1A-B: Endoscopic images of small bowel mucosa show villous flattening and mildly inflamed edematous mucosa **Fig 1C-D:** H&E stained tissue section shows a panoramic view of multiple fragments of small bowel mucosa with expansion of lamina propria, marked villous flattening (arrow) with no increased intraepithelial lymphocytes and widely spaced intestinal glands (10x). **Fig 1E and F:** A high power examination of H&E stained tissue section show villous flattening, marked infiltration of foamy macrophages, and widely spaced small bowel glands. At places few epithelioid cells are also noted. (40x) (Highlighted by arrows). An insert image shows exhausted block with no tissue remaining.

DISCUSSION

WD is a rare multisystem disease which presents in the middle age group. This substantial age discrepancy between our case and the reported data highlights a rare and unusual occurrence warranting strong clinical suspicion of WD. In our index case, the patient was notably younger at 15 years-old compared to the other authors who reported middle-aged patients. Rey R.M. et al. reported 46 years, Crews N.R. reported 52 years, and Günther U. et al. found a mean age of 55 years.[7,4,6] De Francesco V. et al. and Ruggiero E. et al documented patients in 6th and 7th decades.[2,3]

The literature review indicates a male preponderance. The index case also a male patient, similarly reported by De Francesco V. et al., Ruggiero E. et al., and Crews N.R.et al.[2-4] However, female patients are also reported by Rezk A. et al. and Rey R.M.et al.[5,7] The pattern of gastrointestinal and rheumatological involvement was a consistently noted across studies. In our study, the patient presented with abdominal pain, diarrhea, malabsorption, and weight loss, similarly reported by Günther U. et al., Crews N.R., Nasim A. et al, Chandra S et al and Batista M et al. [6,4,8,10] In addition, Rezk A et al. reported arthritis symptoms with RA factor negativity apart from gastrointestinal symptoms.[5] Nasim A et al. reported Whipple's disease presented with diarrhea in transplant recipients.[8]

Whipple's disease is a multisystem disease and it is rarely present as only gastrointestinal disease. In the gastrointestinal tract it commonly affects small bowel as reported in the index cases aligned with Crews N.R. et al.[6] In contrast, Rezk A. et al. reported histiocytes containing periodic acid-Schiff (PAS)-positive globules in the mesenteric lymph node.[5]

REFERENCES

1. Kucharz EJ, Kramza J, Grosicka A, Pieczyrak R. Clinical manifestations of Whipple's disease mimicking rheumatic disorders. Reumatologia. 2021;59(2):104-10. https://doi.org/10.5114/reum.2021.105418
2. De Francesco V, Corsi F, Pennella A, Bellesia A, Fiorini G, Vaira D, et al. Whipple's disease: case report and review of the literature. J Gastrointest Liver Dis. 2018;27(3). https://doi.org/10.15403/jgld.2014.1121.273.fra
3. Cappellini A, Minerba P, Maimaris S, Biagi F. Whipple's disease: A rare disease that can be spotted by many doctors. Eur J Intern Med. 2024;121:25-9. https://doi.org/10.1016/j.ejim.2023.12.009
4. Crews NR, Cawcutt KA, Pritt BS, Patel R, Virk A. Diagnostic approach for classic compared with localized Whipple disease. Open Forum Infect Dis. 2018;7(5):ofy136. https://doi.org/10.1093/ofid/ofy136
5. Rey RM, Orozco LA, Marrugo K, López R, Pérez-Riveros ED, De la Hoz-Valle J, et al. Whipple disease diagnosed by enteroscopy: first case report in Colombia of an underdiagnosed disease and literature review. BMC Gastroenterol. 2020;20:1-5. https://doi.org/10.1186/s12876-020-01302-2
6. Nasim A, Dodani SK, Badlani S, Babar ZU, Shakil S, Mubarak M, et al. Whipple's disease in renal transplant recipients: Management experience of seven cases from Pakistan. Transpl Infect Dis. 2022;24(1):e13769. https://doi.org/10.1111/tid.13769
7. Melas N, Amin R, Gyllemark P, Younes AH, Almer S. Whipple's disease: the great masquerader-a high level of suspicion is the key to diagnosis. BMC Gastroenterol. 2021;21:1-8. https://doi.org/10.1186/s12876-021-01664-1
8. Chandra S, Bledsoe AC. A case of chronic diarrhea with weight loss. Gastroenterology. 2022;163(6):e20-2. https://doi.org/10.1053/j.gastro.2022.07.015
9. Batista M, Santos ML, Silva C, Pereira G, Alves G, Cotter J. Whipple's disease: a case report. Cureus. 2023;15(6):e39963. https://doi.org/10.7759/cureus.39963
10. Masood M, Channa RH, Butt N. Frequency and risk factors of malabsorption in patients presenting at tertiary care hospital, Karachi. Pak J Med Sci. 2024;40(3Pt-II):482. https://doi.org/10.12669/pjms.40.3.7957

CONFLICT OF INTEREST
Authors declare no conflict of interest.
GRANT SUPPORT AND FINANCIAL DISCLOSURE
None declared.

AUTHORS' CONTRIBUTION

The following authors have made substantial contributions to the manuscript as under:

Conception or Design:	RA, MA, AS,
Acquisition, Analysis or Interpretation of Data:	RA, MA, AS, NN, AH, MJC, FS, MA, AA
Manuscript Writing & Approval:	RA, MA, AS, MA, AA, MKN, SMC, RA

All the authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.