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Cronkhite-Canada Syndrome Presenting as Chronic Diarrhea with Ectodermal Changes: A Case Report

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ABSTRACT

Cronkhite-Canada syndrome (CCS) is a rare, non-inherited gastrointestinal polyposis syndrome characterized by diffuse hamartomatous polyps and ectodermal changes. We report a case of a 51-year-old woman presenting with chronic diarrhea, loss of taste, nail dystrophy, and alopecia, subsequently diagnosed with CCS. Early recognition and immunosuppressive therapy led to clinical and biochemical improvement.

INTRODUCTION

Cronkhite-Canada syndrome is an uncommon, sporadic disorder first described in 1955, with fewer than 500 cases reported worldwide. It typically presents in middle-aged to elderly individuals with gastrointestinal symptoms and characteristic ectodermal manifestations, including alopecia, nail dystrophy, and skin hyperpigmentation. The disease is associated with protein-losing enteropathy, leading to malnutrition and anemia. The etiology is unclear but thought to involve autoimmune mechanisms.

CASE DESCRIPTION

A 51-year-old woman presented with chronic, non-bloody diarrhea for six months, associated with loss of taste, brittle nails, and loss of eyebrow hair. She also reported generalized weakness and pedal swelling. On examination, she was pale, had bilateral pedal edema, loss of eyebrows, nail dystrophy involving both hands and feet, and diffuse hyperpigmentation of the hands Figure 1.

Investigations revealed hemoglobin 8.6 g/dL, mean corpuscular volume (MCV) 69 fL, peripheral smear showing microcytic hypochromic picture, and transferrin saturation 4% suggestive of severe iron deficiency anemia. Total protein was 4.3 g/dL, albumin 1.8 g/dL, and urine analysis showed no proteinuria.

Upper gastrointestinal endoscopy revealed diffuse gastric antral polyposis with mucosal edema, extending into the second part of the duodenum. Colonoscopy demonstrated diffuse mucosal edema and multiple hamartomatous polyps

throughout the colon and terminal ileum Figure 2. Histopathology from all sites showed hamartomatous polyps with edematous lamina propria and cystically dilated glands, consistent with Cronkhite-Canada syndrome Figure 3.

The patient was started on oral corticosteroids with a tapering regimen and azathioprine as a steroid-sparing agent. Supportive management included nutritional and iron supplementation. After three months of therapy, diarrhea subsided, taste sensation improved, and ectodermal changes partially regressed. Hemoglobin improved to 10 g/dL, total protein to 5.4 g/dL, and albumin to 3.1 g/dL.

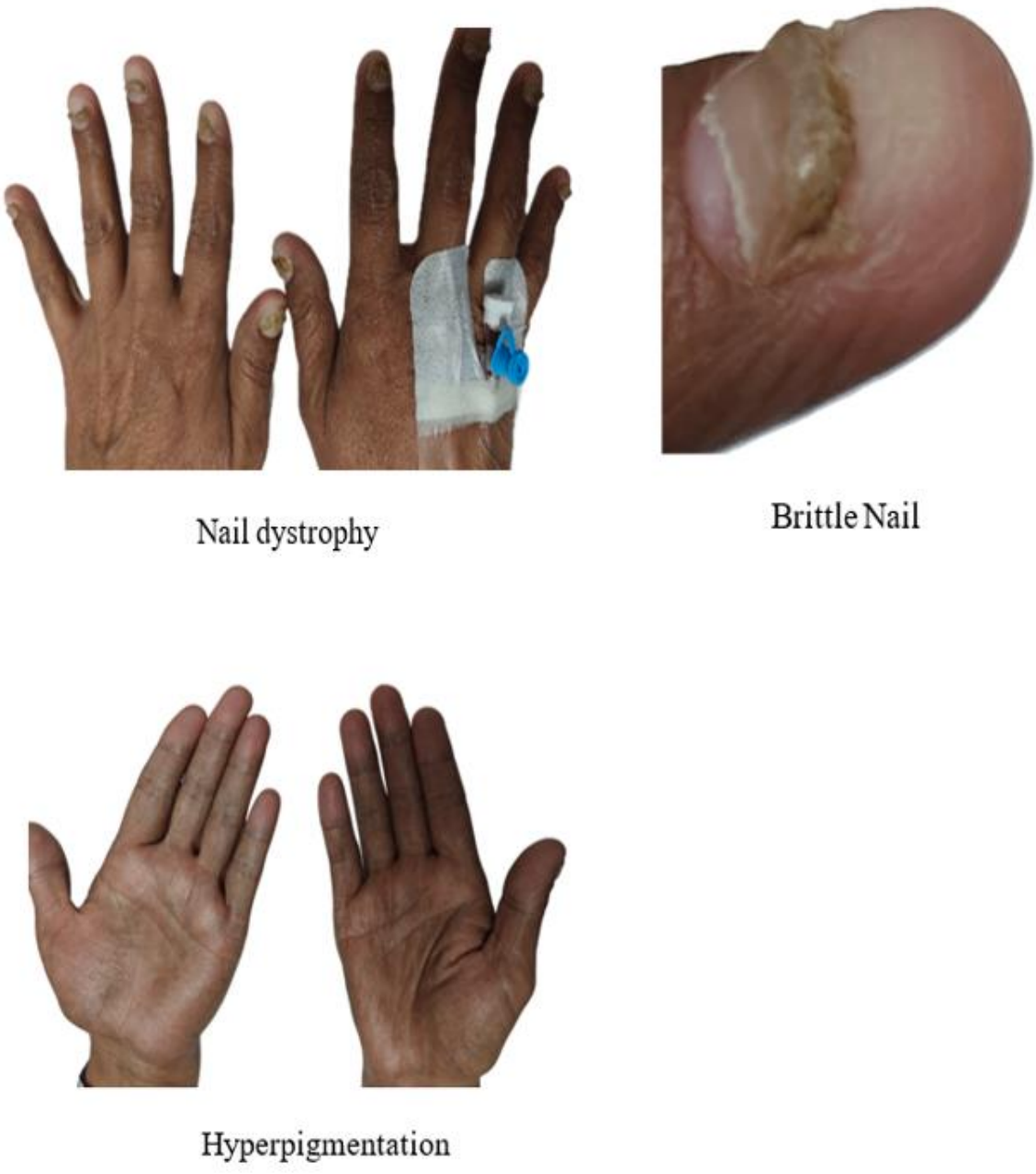
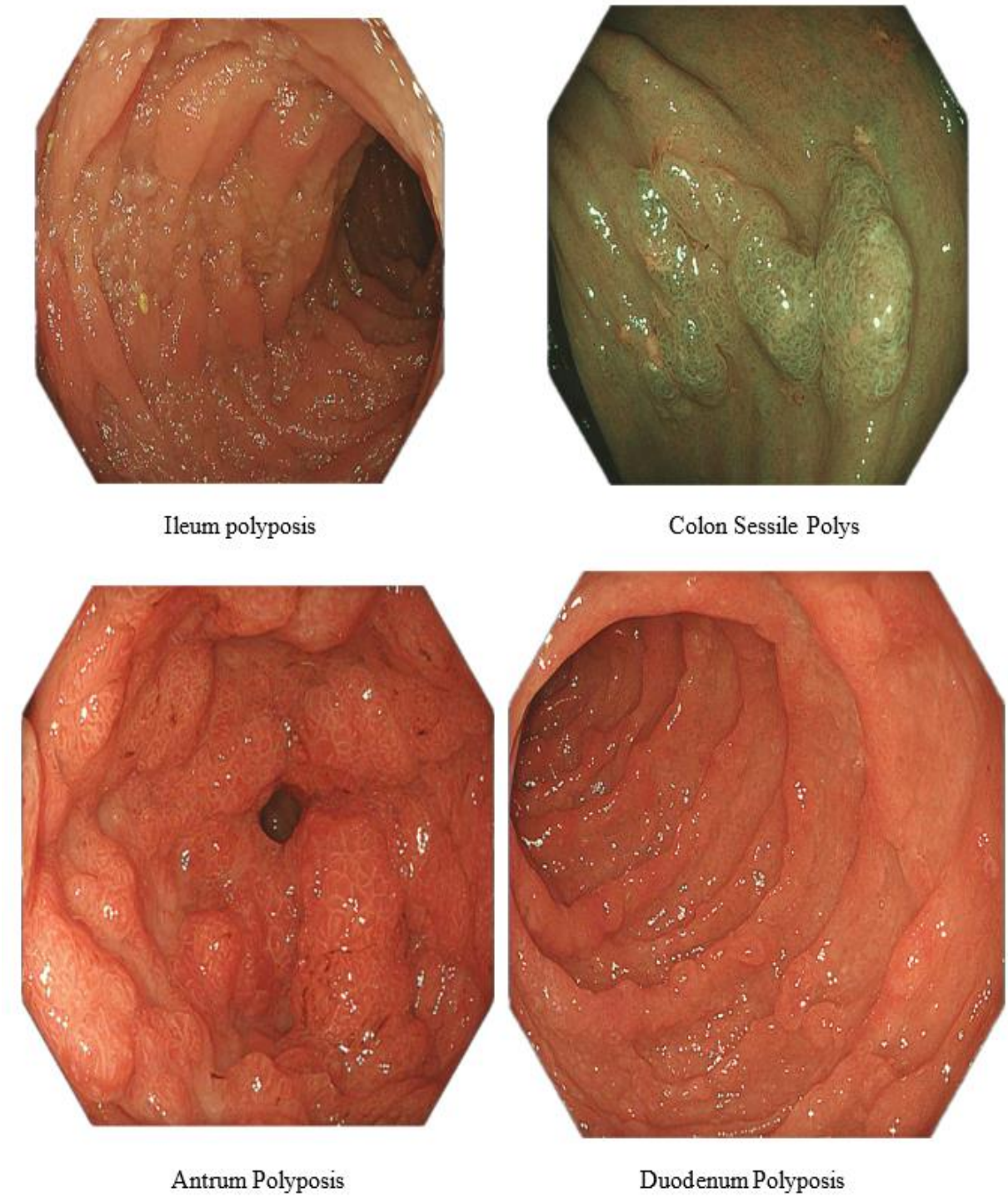


Figure 1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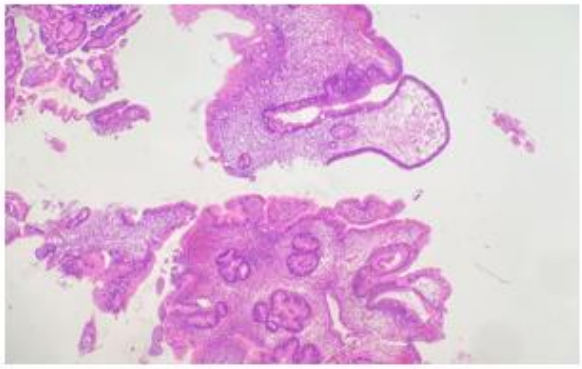
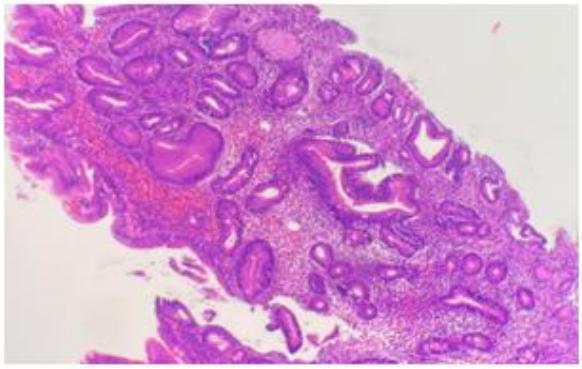
Colon Sessile Polys
Antrum Polyposis
Duodenum Polyposis

Figure 2



Antrum



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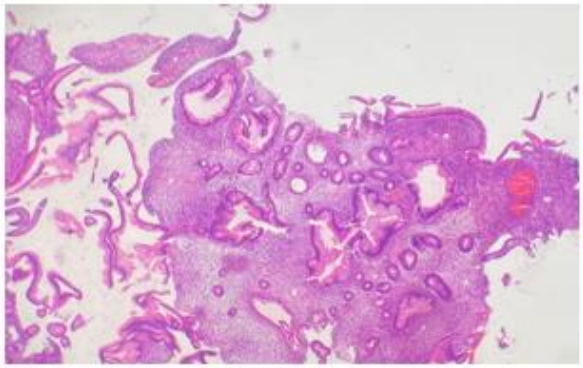
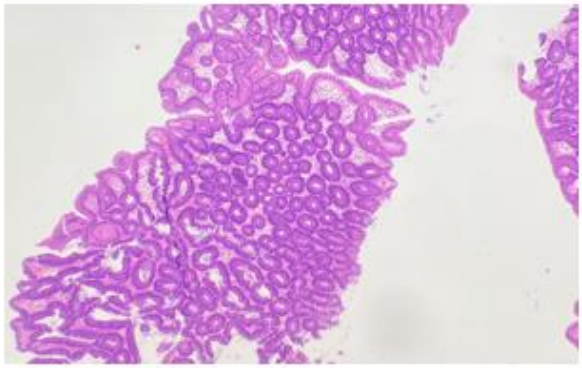


Terminal Ileum



Colon

DISCUSSION

This case underscores the importance of recognizing CCS in patients presenting with chronic diarrhea and ectodermal abnormalities. Prompt diagnosis and initiation of immunosuppressive therapy, coupled with nutritional support, can significantly improve both clinical outcomes and quality of life. Our report adds to the limited literature on CCS and emphasizes the need for awareness among clinicians, given its rarity and potential for severe complications.

Long-term surveillance is recommended because malignant transformation has been reported in approximately 9-15% of patients, particularly in gastric and colonic polyps [1,2]. Endoscopic follow-up at 6-12 month intervals, along with careful monitoring of nutritional status and laboratory parameters, is advised. Relapse can occur, and early detection is key to preventing severe complications.

Although the exact pathogenesis remains unclear, autoimmune mechanisms are strongly suspected. Supporting this, most patients respond favorably to immunosuppressive therapy, including corticosteroids and steroid-sparing agents such as azathioprine or cyclosporine [2,3]. Nutritional supplementation, including protein and micronutrients, is

essential due to chronic malabsorption. In our case, combined therapy resulted in clinical improvement, normalization of hemoglobin, and partial regression of ectodermal changes.

Patients typically present with chronic diarrhea, weight loss, malnutrition, and protein-losing enteropathy, often accompanied by iron deficiency anemia and hypoalbuminemia, as seen in our patient [2]. Laboratory findings generally reveal microcytic anemia, low serum albumin, and hypoproteinemia without evidence of renal or hepatic protein loss. The diagnosis is confirmed via endoscopy and histopathology, which reveal multiple hamartomatous polyps with edematous lamina propria and cystically dilated glands, helping to differentiate CCS from other polyposis syndromes such as juvenile polyposis, Peutz–Jeghers syndrome, and Cowden syndrome [3].

Cronkhite–Canada syndrome (CCS) is an extremely rare, nonhereditary gastrointestinal polyposis disorder with multisystem involvement. Since its first description in 1955, fewer than 500 cases have been reported worldwide, with a higher prevalence in Japan [1]. The syndrome is characterized by diffuse hamartomatous polyps, predominantly affecting the stomach, small intestine, and colon, along with distinctive ectodermal manifestations including alopecia, onychodystrophy, and skin hyperpigmentation. These features, when present together with gastrointestinal symptoms, are highly suggestive of CCS.

CONCLUSION

This case highlights the importance of recognizing Cronkhite–Canada syndrome in patients with chronic diarrhea and ectodermal changes. Early diagnosis and immunosuppressive therapy can significantly improve clinical and biochemical outcomes.

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