CASE REPORT

DOI: 10.4274/cmj.galenos.2025.42713
J Cau Med Sci 2025;3(1):10-11

Good Syndrome Associated Enteropathy: A Case Report

Narmin Naghizada[1], Tuğba Tolu[2], Haluk Tarık Kani[2,3]

[1]Marmara University Faculty of Medicine, Department of Internal Medicine, İstanbul, Turkey
[2]Marmara University Faculty of Medicine, Department of Gastroenterology, İstanbul, Turkey
[3]Marmara University Faculty of Medicine, Institute of Gastroenterology, İstanbul, Turkey

ABSTRACT

Good syndrome (GS) is a rare, late-onset primary immunodeficiency disorder characterized by the presence of thymoma and hypogammaglobulinemia, with no familial inheritance. Immunological abnormalities in GS include defects in both humoral and cellular immunity. We present the case of a patient with GS who developed chronic diarrhea. Unlike other reported cases of GS-associated enteropathy in the literature, this case exhibited similarities to common variable immunodeficiency-related enteropathy. The patient's chronic diarrhea was unresponsive to intravenous immunoglobulin therapy but showed significant improvement with steroids and infliximab. This case highlights the potential for atypical presentations of GS-associated enteropathy and underscores the importance of considering alternative therapeutic strategies, including immunomodulatory agents, in refractory cases.

Keywords: Colitis, Good syndrome, Infliximab

INTRODUCTION

Good syndrome (GS) is a primary immunodeficiency disorder characterized by the association of thymoma and hypogammaglobulinemia [1]. Although it was classified as a subset of common variable immunodeficiency (CVID) in 2005 [2], GS is considered a distinct late-onset primary immunodeficiency with low peripheral B-cell counts, adult onset (typically between ages 40-60), and no familial history [3]. The International Union of Immunological Societies and the World Health Organization have acknowledged it as a separate clinical entity [4].

CASE REPORT

A 54-year-old male presented to the gastroenterology outpatient clinic with persistent diarrhea. The patient reported persistent diarrhea for 3 months; up to 10 bowel movements per day, without blood or mucus. Additionally, he lost 25 kg in the last year. In his medical history, he had a thymectomy two years ago due to thymoma. In physical examination, he was dehydrated; other than that, his physical examination was normal. The patient was hospitalized with these findings.

Initial laboratory tests: white blood cell: 8100/mL, neutrophils: 6800/mL, lymphocytes: 900/mL, hemoglobin: 11.4 g/dL, platelets: 456,000/mL, creatinine: 0.43 mg/dL, aspartate aminotransferase: 13 U/L, alanine aminotransferase: 12 U/L, albumin: 28 g/L, total protein: 43 g/L, procalcitonin: 0.17 mg/L, C-reactive protein: 95 mg/L. There was no pathological finding in ileo-colonoscopy, and multiple biopsies were performed from each segment of the terminal ileum and colon.

Laboratory results showed significantly low immunoglobulin (Ig) levels: IgG 1.19 g/L, IgM <0.2 g/L, and IgA <0.1 g/L. Isohemagglutinin testing revealed anti A-IgM negativity and anti B-IgM positivity. Hypogammaglobulinemia, the presence of thymoma, and recurrent infections led to a diagnosis of GS. Intravenous immunoglobulin (IVIG) therapy was scheduled to be administered every 21 days.

Hypoxemia (oxygen saturation: 85%) and tachypnea (respiratory rate: 32/min) developed on the 7th day of hospitalization. A chest computed tomography scan revealed bilateral ground-glass opacities. Sputum cultures and a respiratory viral panel were sent for analysis. Opportunistic infections, including tuberculosis, Pneumocystis jirovecii pneumonia (PCP), and cytomegalovirus (CMV)

Address for Correspondence: Haluk Tarık Kani MD, Marmara University Faculty of Medicine, Department of Gastroenterology; Marmara University Faculty of Medicine, Institute of Gastroenterology, İstanbul, Turkey
E-mail: tarik.kani@marmara.edu.tr **ORCID ID:** 0000-0003-0042-9256
Received: 21.01.2025 **Accepted:** 11.04.2025 **Publication Date:** 30.04.2025

Cite this article as: Naghizada N, Tolu T, Kani HK. Good syndrome associated enteropathy: a case report. J Cau Med Sci. 2025;3(1):10-11

pneumonia, were considered in the differential diagnosis. He was started on intravenous trimethoprim/sulfamethoxazole (15 mg/kg) and methylprednisolone for suspected PCP. However, the bronchoalveolar lavage (BAL) PCP test result was negative, and the empirical treatment was discontinued. HIV and tuberculosis tests were negative. Plasma CMV polymerase chain reaction (PCR) was 1,096 copies/mL, while BAL CMV PCR showed 299,168 copies/mL; confirming CMV pneumonia. Additionally, the colonoscopy revealed CMV colitis. Intravenous ganciclovir (5 mg/kg twice daily) was initiated, resulting in symptom improvement. After this, the patient was transitioned to oral valganciclovir for maintenance therapy. The patient was discharged and continued his follow-up in the immunology outpatient clinic.

Four months later, the patient presented with diarrhea again (15-20 bowel movements per day). Stool culture, Giardia, Entamoeba histolytica, Clostridium difficile antigen, and parasites in stool were negative; and repeat colonoscopy ruled out CMV colitis. New biopsies showed findings (such as a reduction in plasma cells and eosinophilia) consistent with GS-associated enteropathy. Oral methylprednisolone (32 mg) was started, resulting in temporary symptom relief. However, diarrhea recurred (8-10 episodes daily) as the steroid dose was tapered. Stool testing revealed Clostridium difficile antigen positivity. The patient was treated with oral vancomycin (125 mg four times daily) and metronidazole (500 mg three times daily), which together resolved his symptoms. Despite this, the diarrhea relapsed upon further steroid tapering.

Despite initial improvements, the patient experienced a relapse of colitis upon further steroid tapering. Following a consultation with the Allergy and Immunology Department, intravenous infliximab therapy (5 mg/kg every 8 weeks) was initiated. This resulted in complete remission, and the patient was referred for follow-up in both gastroenterology and immunology clinics.

DISCUSSION

Enteropathy is a common manifestation in primary immunodeficiency disorders. It can present with chronic diarrhea, malabsorption, growth delay, iron deficiency anemia, and other symptoms of malnutrition. The most common causes are celiac disease (26.2%), Immunodysregulation Polyendocrinopathy Enteropathy X-linked syndrome (20.7%), autoimmune enteropathy (6.4%), and CVID-associated enteropathy (5.8%) [5].

A recent study reviewed on GS cases analyzing 225 patients was published between November 2020 and October 2022, among them, 22 (9.8%) had non-infectious gastrointestinal involvement, including inflammatory bowel disease (n=4), multiorgan autoimmunity involving the gastrointestinal system (n=3), celiac disease (n=1), collagenous colitis (n=1), autoimmune enteropathy (n=1), and non-granulomatous colitis (n=1). Chronic diarrhea (90.9%) and significant weight loss (50.0%) were the most common clinical findings [6].

Clinical improvement was observed in 11 patients with IVIG, and two patients improved following thymectomy [6].

In contrast, our case demonstrated no response to IVIG but achieved remission with infliximab, similar to the treatment approach for CVID-associated enteropathy [7].

GS remains a rare clinical entity, with no established consensus regarding its treatment or pathophysiology. Chronic diarrhea is a common symptom of this syndrome. Once infectious causes are excluded, further research and case series are essential to better understand immune-mediated enteropathies associated with GS.

Ethics

Informed Consent: A written informed consent has been granted from the patient.

Footnotes

Authorship Contributions

Surgical and Medical Practices: H.T.K., Concept: N.N., T.T., H.T.K., Design: N.N., T.T., H.T.K., Data Collection or Processing: N.N., T.T., H.T.K., Analysis or Interpretation: N.N., T.T., H.T.K., Literature Search: N.N., T.T., H.T.K., Writing: N.N., T.T., H.T.K.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study received no financial support.

REFERENCES

1. Kelesidis T, Yang O. Good's syndrome remains a mystery after 55 years: a systematic review of the scientific evidence. Clin Immunol. 2010;135:347-63.
2. Bonilla FA, Khan DA, Ballas ZK, Chinen J, Frank MM, et al. Practice parameter for the diagnosis and management of primary immunodeficiency. J Allergy Clin Immunol. 2015;136:1186-205.e1-78.
3. Malphettes M, Gérard L, Galicier L, Boutboul D, Asli B, et al. Good syndrome: an adult-onset immunodeficiency remarkable for its high incidence of invasive infections and autoimmune complications. Clin Infect Dis. 2015;61:e13-9.
4. Primary immunodeficiency diseases. Report of an IUIS Scientific Committee. International Union of Immunological Societies. Clin Exp Immunol. 1999;118(Suppl 1):1-28.
5. Chung H, Zheng B, Chen B, Wang A, Kong XF. Enteropathy in primary immunodeficiency diseases: a systematic review of cases. Am J Gastroenterol. 2022;117:e1111-e3.
6. Wang C, Shi Y. Non-infectious gastrointestinal manifestations of Good syndrome. Eur J Intern Med. 2023;109:123-4.
7. Malamut G, Verkarre V, Suarez F, Viallard JF, Lascaux AS, et al. The enteropathy associated with common variable immunodeficiency: the delineated frontiers with celiac disease. Am J Gastroenterol. 2010;105:2262-75.