Research Square

Preprints are preliminary reports that have not undergone peer review.
They should not be considered conclusive, used to inform clinical practice,
or referenced by the media as validated information.

Severe Chronic Enteropathy Associated with SLCO2A1 Mutation in a Male Child Complicated by Extensive Jejunal Strictures and Perforation: A Case Report

Yuyoung Oh
Seoul National University Children's Hospital
Hyun-Young Kim
spkhy02@snu.ac.kr

Seoul National University Children's Hospital

Case Report

Keywords:

Posted Date: March 22nd, 2026

DOI: https://doi.org/10.21203/rs.3.rs-8843708/v1

License: This work is licensed under a Creative Commons Attribution 4.0 International License.
Read Full License

Additional Declarations: No competing interests reported.

Abstract

Background

Chronic enteropathy associated with SLCO2A1 (CEAS) is a rare hereditary disorder characterized by multiple small-bowel ulcerations caused by impaired prostaglandin transport. The disease predominantly affects female patients and usually follows an indolent course. Severe complications requiring surgical intervention are uncommon, particularly in male patients.

Case presentation

We report a male child with genetically confirmed CEAS who developed progressive jejunal strictures complicated by capsule retention and bowel perforation. Initially diagnosed at 6 years of age after capsule endoscopy revealed multiple small-bowel erosions, the patient underwent stricturoplasty at 10 years due to capsule retention. At 14 years of age, he presented with worsening abdominal pain, and imaging demonstrated a jejunal microperforation. Exploratory laparotomy revealed multiple tight strictures involving approximately 1 meter of jejunum with an associated perforation, necessitating segmental resection and primary anastomosis. Histopathologic examination showed submucosal fibrosis, ischemic mucosal changes, and acute serositis. The patient recovered uneventfully and remained clinically stable at follow-up.

Conclusions

This case illustrates a rare and aggressive phenotype of CEAS in a male patient, highlighting the potential for progressive stricturing disease and life-threatening complications. Early recognition, cautious use of capsule endoscopy, and close surveillance are essential in managing patients with SLCO2A1-associated enteropathy.

Background

Chronic enteropathy associated with SLCO2A1 (CEAS), also known as chronic nonspecific multiple ulcers of the small intestine, is a rare autosomal recessive disorder caused by loss-of-function mutations in the SLCO2A1 gene, which encodes a prostaglandin transporter [1,2]. Impaired prostaglandin transport leads to chronic mucosal injury, resulting in multiple small-bowel ulcerations, anemia, hypoalbuminemia, and growth retardation.

CEAS predominantly affects female patients and typically presents during childhood or adolescence with a relatively indolent clinical course [3,4]. Most patients are managed conservatively, and severe complications such as obstruction, perforation, or the need for extensive bowel resection are considered uncommon. Reports of aggressive disease phenotypes in male patients are particularly rare.

Here, we describe a male child with genetically confirmed CEAS who developed progressive jejunal strictures complicated by capsule retention and small-bowel perforation, ultimately requiring segmental resection. This case expands the clinical spectrum of CEAS and underscores the potential severity of the disease.

Case presentation

A 6-year-old boy presented with chronic anemia, hypoalbuminemia, and growth failure, with both height and weight below the 3rd percentile for age. Capsule endoscopy revealed multiple erosive lesions throughout the small intestine. Genetic testing identified a homozygous SLCO2A1 mutation (c.940+1G>A), confirming the diagnosis of chronic enteropathy associated with SLCO2A1.

At 10 years of age, the patient developed progressive abdominal pain and small-bowel obstruction due to capsule retention. Surgical intervention with small-bowel stricturoplasty and removal of the retained capsule was performed.

At 14 years of age, he presented again with worsening abdominal pain. Computed tomography demonstrated focal small-bowel wall thickening with suspected microperforation. Exploratory laparotomy revealed multiple tight strictures involving approximately 1 meter of jejunum, along with a localized perforation site (Fig. 1A, B). Segmental resection of the affected jejunum with primary anastomosis was performed.

Gross examination showed multiple areas of luminal narrowing and ulceration. Histopathologic analysis demonstrated marked submucosal fibrosis, ischemic mucosal changes, hemorrhage, and acute serositis, consistent with advanced CEAS. The postoperative course was uneventful, and the patient was discharged in stable condition. At follow-up, he remained clinically stable without recurrent obstructive symptoms.

Discussion and conclusions

CEAS is a rare enteropathy characterized by chronic ulceration of the small intestine resulting from defective prostaglandin transport [1–4]. While most reported cases describe a relatively indolent disease course, our patient exhibited a markedly aggressive phenotype with progressive strictures, capsule retention, and bowel perforation.

The notable features of this case include the patient's male sex and the extent of jejunal involvement requiring segmental resection. Previous studies have suggested a female predominance and relatively mild clinical progression in CEAS; however, this case demonstrates that severe disease with life-threatening complications can occur, even in childhood [3,8].

Surgical intervention in CEAS is generally avoided when possible, as repeated resections may predispose patients to short bowel syndrome. Nevertheless, in cases complicated by obstruction or perforation, timely surgical management is unavoidable. This case underscores the importance of close clinical monitoring, cautious use of capsule endoscopy due to the risk of retention [7], and early consideration of surgical intervention in patients with progressive stricturing disease.

In conclusion, this case expands the phenotypic spectrum of CEAS and highlights that male patients may develop severe, progressive disease. Awareness of this possibility is essential for pediatric

surgeons and gastroenterologists involved in the care of patients with SLCO2A1-associated enteropathy.

Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

Written informed consent was obtained from the patient's parents for publication of this case report and accompanying images.

Author contributions

Y.O. collected clinical data and drafted the manuscript. H.-Y.K. supervised the study and critically revised the manuscript. All authors read and approved the final manuscript.

Funding

The authors received no specific funding for this work.

Availability of data and materials

Data supporting the findings of this study are available from the corresponding author upon reasonable request.

Competing interests

The authors declare that they have no competing interests.

References

1. Umeno J, Hisamatsu T, Esaki M, et al. A hereditary enteropathy caused by mutations in the SLCO2A1 gene, encoding a prostaglandin transporter. *Gastroenterology*. 2015;148:152–163.
2. Hosoe N, Kobayashi T, Umezawa S, et al. Chronic enteropathy associated with SLCO2A1 gene mutation: a newly recognized disease entity. *Gut*. 2017;66:1055–1064.
3. Hisamatsu T, Umeno J, Matsumoto T, et al. Clinical characteristics of chronic enteropathy associated with SLCO2A1 gene mutation. *Inflamm Bowel Dis*. 2016;22:588–597.
4. Nakase H, Uchino M, Shinzaki S, et al. Small intestinal ulcerations caused by SLCO2A1 mutations: clinical features and outcomes. *J Crohns Colitis*. 2018;12:1223–1230.
5. Matsumoto T, Iida M, Kimura Y, et al. Chronic nonspecific multiple ulcers of the small intestine unrelated to NSAIDs. *Gut*. 2006;55:777–782.
6. Ohmiya N, Nakamura M, Takenaka H, et al. Capsule endoscopy for the diagnosis of chronic nonspecific multiple ulcers of the small intestine. *Endoscopy*. 2011;43:246–254.
7. Yamada A, Komaki Y, Komaki F, et al. Risk of capsule retention in small bowel strictures. *Gastrointest Endosc*. 2017;86:1084–1093.

8. Kusaka T, Umezawa S, Hosoe N, et al. Surgical management of chronic enteropathy associated with SLCO2A1 mutation. *World J Gastroenterol*. 2019;25:4586–4595.

Figures



Figure 1

Intraoperative findings of chronic enteropathy associated with SLCO2A1 mutation.

ALaparotomy reveals extensive jejunal involvement with multiple strictures and marked bowel wall thickening over a long segment of small intestine.
B A focal jejunal perforation is identified within the strictured segment, necessitating segmental resection.