Familial jejun-ileoal diverticulitis: A case report and review of the literature

Jeffrey S. Barton a,⁎, Amit B. Karmur b, Jennifer F. Preston a, Brett C. Sheppard a

a Oregon Health and Science University, Department of Surgery
b Western University of Health Sciences, College of Osteopathic Medicine of the Pacific, United States

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ABSTRACT

INTRODUCTION: Jejuno-ileoal diverticulitis (JID) is a rare entity, presenting with symptoms of failure to thrive, abdominal pain, obstruction, bleeding, and acute or chronic perforation with associated pneumoperitoneum. Currently no specific genetic abnormality has been identified that leads to JID. Treatment is based on control of symptoms associated with the disease.

PRESENTATION OF CASE: We describe a familial cohort of patients with JID, with associated symptoms of chronic pneumoperitoneum, including a proposed genetic inheritance pattern and pedigree. In addition, we will describe the operative treatment of one family member’s JID and chronic pneumoperitoneum.

DISCUSSION: While JID is rare, this familial cohort demonstrates a pattern of inheritance most consistent with autosomal dominance. The pathology demonstrates true diverticula, unlike most previous descriptions of JID. The index patient was successfully treated by minimally invasive surgery.

CONCLUSION: Familial JID is a rare entity, without an identified genetic abnormality. Treatment of chronic symptoms currently focuses on non-operative management. While most case reports involve individual patients, this cohort may possess a genetic mutation with an autosomal dominant pattern of inheritance. Further study into patients with JID may reveal an underlying genetic abnormality associated with development of the disease.

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1. Introduction

Jejuno-ileoal diverticulitis (JID) is a rare pathologic finding of the small bowel, affecting an estimated 0.06–1.3% of the population. The disease is thought to have a pathophysiology similar to that of colonic diverticulitis, wherein chronically elevated intraluminal pressures lead to the formation of false diverticula.

To the best of our knowledge, only two series in the literature have documented JID occurring within families. Andersen et al. describe a family of eight that underwent barium contrast imaging, of whom six were diagnosed with either duodenal or jejuno-ileoal diverticula or both and five of the six presenting with symptoms. The authors noted concomitant autoimmune diseases in four family members, including rheumatoid arthritis, non-viral hepatitis, ulcerative colitis and thyroiditis, and suggested the JID may be a consequence of the underlying systemic disease. Koch and Schoon proposed an autosomal dominant inheritance pattern based on a family consisting of a mother and two of her four children with symptomatic JID.

Despite the description of multiple family members, neither of the above reports described pathologic specimens from the patients discussed. The purpose of the present report is to describe a family of patients with JID, to discuss the operative management and pathology findings of one family member, and to propose an alternative hypothesis as to the pathophysiology of JID.

2. Presentation of case

A 74-year old man was initially seen in the General Surgery clinic with a five-year history of chronic pneumoperitoneum in the setting of JID. At the time of referral, the patient complained of abdominal pain, nausea, intermittent diarrhea, a 10 lbs weight loss in the last 6 months and increased abdominal distension. The patient had a personal history notable for celiac disease, but no other connective tissue disorders. On examination, the patient was cachectic, with a taut and tympanic abdomen.

The patient’s family history was significant for an extensive history of JID (Fig. 1). His paternal grandfather had episodic abdominal cramping, and his mother had episodic severe abdominal pain with associated distension but neither had a medical diagnosis of JID prior to their deaths. Of ten children, six had radiographically or
pathologically confirmed JID, with the oldest affected sister having undergone small bowel resection for perforation. Additionally, his eldest brother had eight children, including a pair of monozygotic twin daughters who were both affected by radiographically confirmed JID.

The patient underwent an elective laparoscopic exploration. He was found to have pneumoperitoneum, ascites and extensive diverticulosis throughout the jejunum. The proximal jejunum was found to have dense diverticulosis, with scattered diverticula in the distal jejunum. We performed a laparoscopic resection of the proximal 100 cm of jejunum, with eight additional stapled diverticulectomies in the distal jejunum (Fig. 2a). The specimen was sent for pathology, where H&E staining demonstrated both muscularis mucosa and muscularis propria, consistent with true diverticula (Fig. 2b). Additional staining for desmin re-demonstrated the presence of muscularis mucosa and muscularis propria (Fig. 2c and d).

Six months after the index operation, the patient had resolution of his symptoms of distension, pneumoperitoneum and abdominal pain. He was gaining weight and his pre-albumin and albumin are now within normal limits. He was discharged from the clinic with as-needed follow-up.

3. Discussion

Two case series exist describing familial cohorts of JID, implicating concomitant autoimmune disease and autosomal dominant inheritance respectively as possible causes of the disease.\(^1,3\) The present case series demonstrates multiple family members with celiac disease and JID. The inheritance pattern observed is most consistent with an autosomal dominant trait. Based on history obtained from the living family members, either of the paternal grandfather or mother may have had symptomatic JID, although the authors suspect the mother is a more likely culprit based on the symptoms described. The presence of monozygotic twins with the disease is further suggestive of a genetic predisposition.

While JID has previously been considered a disease of false diverticula,\(^2\) the present pathology specimen clearly demonstrates all layers of the bowel consistent with true diverticula. This appears to represent a distinct pathophysiology from the previously proposed cause, where high intraluminal pressure leads to mucosal and submucosal extrusion through the muscularis propria. Rather, this disease process appears more consistent with that described by Krishamurthy et al.,\(^4\) wherein true diverticula contain fibrotic tissue within the muscularis propria.

Non-operative therapy remains a primary method of treatment of JID, as the majority of patients are asymptomatic.\(^5\) Despite this, complications such as bleeding, failure to thrive or perforation...
with associated sepsis may necessitate surgical intervention. The present case demonstrates that a minimally invasive approach can be safely and effectively employed for enterectomy and diverticulectomy in the treatment of complicated JID.

4. Conclusions

JID remains a rare entity. Here we present a family of patients with the disease, with an apparent autosomal dominant pattern suggestive of a genetic inheritance. Unlike most descriptions of JID, this series of patients showed a disease of true diverticula, involving all layers of the bowel. While the inheritance and pathophysiology of this disease appears different, surgical therapy is only necessary in the setting of complicated disease. Additionally, we show here that minimally invasive management of the disease is feasible by enterectomy and diverticulectomy.

Conflict of interest

None.

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None.

Ethical approval

This was ethically approved.

Author contributions

Jeffrey S. Barton – Data collection, data analysis and interpretation, writing the paper.
Amit B. Karmur – Data collection.
Jennifer F. Preston – Data collection.
Brett C. Sheppard – Study concept, data interpretation, writing the paper.

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