

# Case Report of Appendiceal Torsion in Ehlers-Danlos Syndrome: A Rare Phenomenon in a Rare Disease



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## Introduction

Appendiceal torsion or volvulus is a rare phenomenon. The first case of appendiceal torsion was reported in 1918<sup>1</sup> and since then, only 30-some cases have been reported in the literature. Both primary and secondary forms of appendiceal torsion exist. Secondary appendiceal torsion is caused by a distinct pathology such as carcinoid tumor making the appendix susceptible to torsion.<sup>2</sup> We present the first case of appendiceal torsion in Ehlers-Danlos syndrome.

## Case Presentation

Patient is a 36-year-old Caucasian male with past medical history of Ehlers-Danlos syndrome who presented to the emergency department with abdominal pain for one week. Pain was initially epigastric but migrated to his right lower quadrant. He endorsed nausea but no vomiting or fevers.

The CT scan showed a dilated appendix (up to 1.4 cm) with mucosal thickening, adjacent fat stranding and edema. There was no mention of appendiceal torsion.

He was taken to the operating room for a standard laparoscopic appendectomy. The distal appendix was swollen with erythema and inflamed mesoappendix (Figure 1). In the mid-appendix there was 270° twisting of the mesentery and appendix (Figure 2, Figure 3). The appendix was grossly normal proximal to the torsion. Using a laparoscopic stapler we divided the appendix off the cecum and divided the mesoappendix. The appendix was removed and sent to pathology.

## Discussion

Appendiceal torsion is clinically indistinguishable from acute appendicitis and this condition is often diagnosed during the operation. Similar to skin hyperelasticity and joint hyperlaxity, ligamentous laxity seen in EDS may make these patients more prone to torsion of abdominal organs. In our patient, primary torsion of the appendix may have been the initial event leading to ischemia and ultimately appendicitis. Alternatively, the tip appendicitis may have led to inflammation, edema and a subsequent lead point for appendiceal torsion.

Ehlers-Danlos syndrome is an infrequent disease affecting roughly 1 in 5000 people.<sup>3</sup> EDS is a connective tissue disorder that can be manifested in multiple organ systems. Classically, it is described in patients with hyperflexible joints and hyperextensible skin due to mutations in Type V collagen.<sup>4</sup> Gastrointestinal manifestations include hiatal hernia, rectocele, rectal prolapse, and diverticular disease.

Surgeries in patients with EDS are associated with significant complications, including poor wound healing, dehiscence and intraoperative bleeding.<sup>5</sup> Preoperative recommendations for the management of classic EDS patients include cardiology assessment with echocardiogram to assess for aortic root dilation and mitral valve prolapse. Blood type and crossmatch testing should be performed prior to surgery. In addition, the anesthesiology team must be informed of EDS diagnosis due to reported cases of cervical spine and mandibular instability, and skin trauma during intubation.<sup>6</sup> During the operation, careful handling of tissue to avoid bleeding and overstretching is advised. Many patients seek the help of facial and plastic surgeons to help amend and close large wounds.

## Conclusion

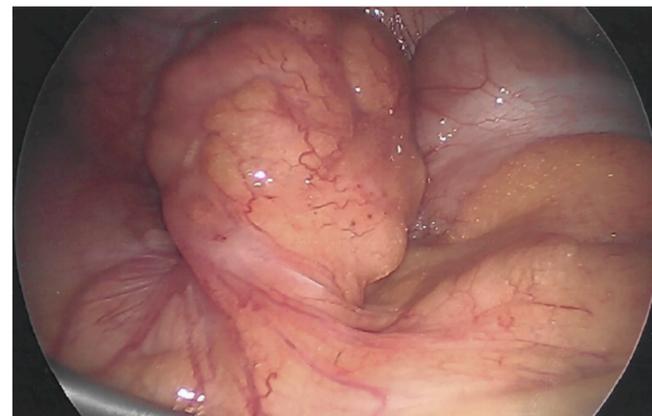
To our knowledge, **this is the first case report of an appendiceal torsion in a patient with Ehlers-Danlos syndrome.** This case serves as evidence of another manifestation of EDS. The ligamentous hypermobility of EDS may have predisposed our patient to appendiceal torsion and acute appendicitis. This is a rare phenomenon and may be underdiagnosed in EDS patients.

## References

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**Figure 1.**  
Inflammation at the tip of the appendix



**Figure 2.**  
270° twisting of the mid-appendix and mesoappendix from medial to anterior position



**Figure 3.**  
Appendiceal torsion