Case Report

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Radiology Section

Desmoid Fibromatosis Presenting with Gastrointestinal Manifestations in a Patient with Ehlers-Danlos Syndrome: A Case Report

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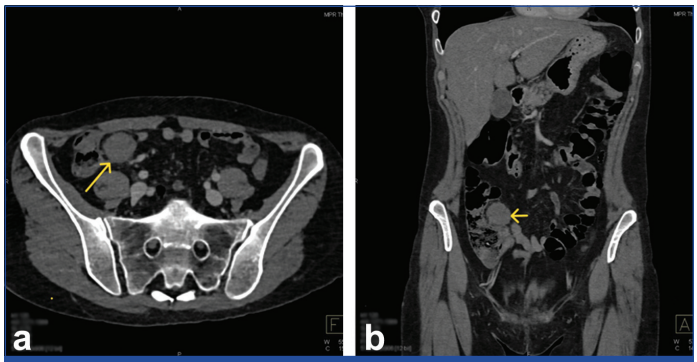
ABSTRACT

Ehlers-Danlos syndrome comprises 13 hereditary connective tissue disorders associated with skin hyperelasticity, joint hypermobility, atrophic scarring, and blood vessel fragility. Hypermobile EDS, the most common subtype, has no clear genetic or molecular source. When presented with wide-ranging clinical symptoms, it requires effective clinical diagnosis. A 31-year-old female with hypermobile EDS underwent an abdominal Computed Tomography (CT) scan displaying a 2.9 cm soft-tissue mass possibly arising from the terminal ileum, which was revealed to be a desmoid fibromatosis. It is believed that Gastrointestinal (GI) manifestations may be associated with EDS, specifically the hypermobile subtype. The patient demonstrated manifestations commonly seen throughout the literature on hypermobile EDS, including abdominal pain, nausea, and diarrhoea. With symptoms worsening, a follow-up CT disclosed an enlargement of the ileocecal desmoid tumour, measuring 5.1 cm. Because all the subtypes of EDS share features such as tissue fragility, joint hypermobility, and skin hyperextensibility, and many of the manifestations are insufficient to establish a diagnosis, imaging modalities are essential for proper identification and patient management. Given the patient's medical history and the involvement of EDS and desmoid tumours with connective tissue, there is a strong linkage between desmoid fibromatosis and the patient's hypermobile Ehlers-Danlos diagnosis. The available literature does not clearly attribute such a tumour to the syndrome. This patient's symptoms represent a possible association between hypermobile EDS and desmoid fibromatosis, warranting future research in this area. The patient is now stable and was recently discharged.

Keywords: Computed tomography, Hereditary connective tissue disorders, Hypermobility, Postural orthostatic tachycardia syndrome, Terminal ileum

CASE REPORT

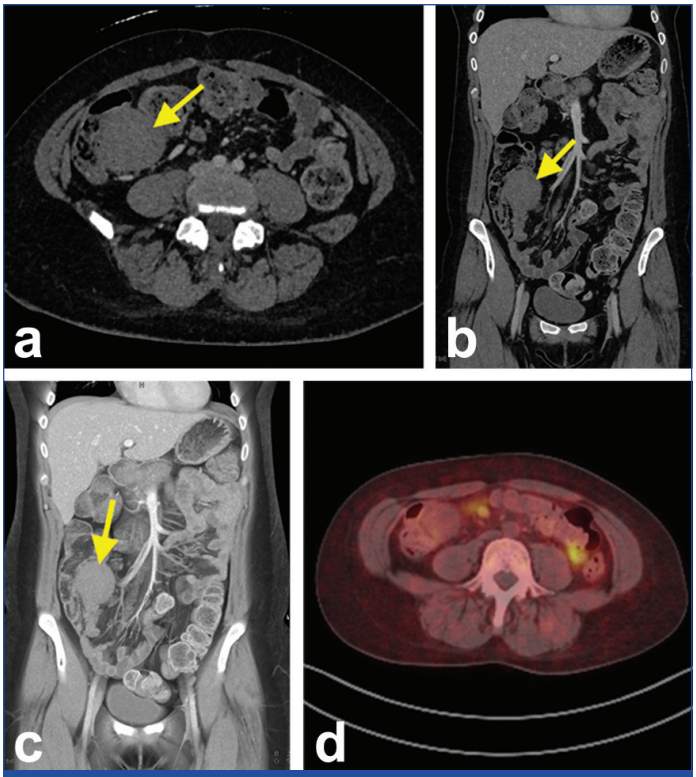
A 31-year-old female with a past history of microscopic colitis, eosinophilic oesophagitis, mast cell activation syndrome, Postural Orthostatic Tachycardia Syndrome (POTS), dysthymia, and hypermobile EDS presented to her Primary Care Physician (PCP) following intense, chronic diffuse abdominal pain, nausea, and watery diarrhoea. The patient reported a history of approximately five episodes of these symptoms beginning three to four years prior, when she was hospitalised for severe abdominal pain and diarrhoea. The patient revealed that she had been experiencing constant, migratory abdominal cramps and watery diarrhoea over the previous year, with acute worsening of symptoms within the past two weeks leading to episodes of syncope. She characterised her recent abdominal pain as crampy, sharp, worse with eating, and radiating from the middle to the right-side. Since the progression of symptoms, she has had diarrhoea six to seven times daily. These events resulted in a 40-pound unintentional weight loss, with 10 pounds lost within the prior two weeks. Given the compilation of symptoms and the development of bilateral tremors in her upper extremities, her PCP recommended her to the emergency department for further evaluation. An ensuing abdominal Computed Tomography (CT) scan displayed a 2.9 cm soft-tissue mass in the right lower quadrant, which abutted and possibly arose from the terminal ileum, and focal enhancing lesions in hepatic segments 8 and 6, representing haemangiomas [Table/Fig-1a,b]. Differential considerations for this mass included small bowel carcinoid tumour, mesenteric mass, GI stromal tumour, or lymphadenopathy, with the highest concern for gastric neuroendocrine tumour considering the patient's acute and chronic symptoms. Consequently, the patient underwent a percutaneous CT-guided coaxial core needle biopsy, the results of which revealed low-grade spindle cell proliferation supporting the clinical diagnosis of desmoid fibromatosis. Given the low-risk of morbidity and nonthreatening nature of this desmoid tumour, oncology recommended monitoring with imaging. Six months after the initial CT, due to the recurrence and worsening of the patient's abdominal pain, a follow-up CT was conducted, revealing enlargement of the ileocecal desmoid tumour now measuring 5.1 cm, with unchanged enhancing lesions in the liver [Table/Fig-2a-d]. Due to this growth, the patient underwent a laparoscopic right hemicolectomy. Following the procedure, her abdominal pain improved, and follow-up scans six months postoperation showed no evidence of disease recurrence [Table/Fig-3a,b].



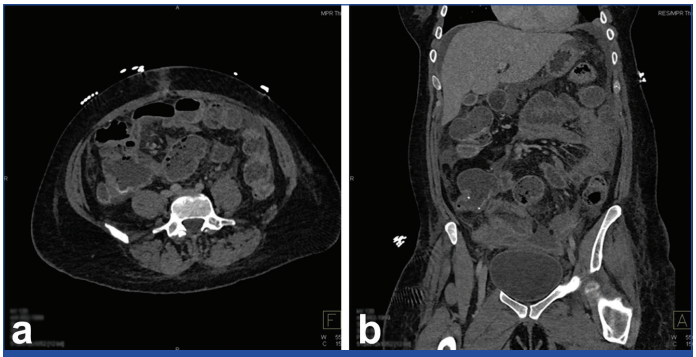
[Table/Fig-1]: a,b) Initial CT scan in patient with abdominal pain showing evidence of a 2.9 cm mass (arrows) emerging from the terminal ileum. The mass is well-defined and soft-tissue in density. No evidence of calcification or significant enhancement was seen.

DISCUSSION

Ehlers-Danlos syndrome (EDS) is a group of genetic connective tissue disorders stemming from abnormalities in collagen processing,



[Table/Fig-2]: Follow-up scan six months after the initial scan. a) 5.1 cm mass in right lower quadrant (arrow) intimately related to the distal ileum and cecum does not show significant enhancement; b) Coronal view from venous phase imaging demonstrates the well-defined mass (arrow) without clear planes from cecum and terminal ileum; c) Coronal projection of volume-rendered views shows solid mildly enhancing mass (arrow) at the border of the ileum and cecum; d) PET/CT at level of mass shows no evidence of increased uptake.



[Table/Fig-3]: Axial (a) and coronal (b) Images on this postoperative scan demonstrate no evdience of residual tumour. Postoperative changes including midly dilated bowel and minimal stranding by surgical bed noted.

production, and structure [1,2]. There are 13 EDS classifications, with each group having a unique set of features; however, joint hypermobility, tissue fragility, and skin hyperflexibility are all common characteristics among all EDS subtypes. The overall prevalence of EDS ranges from approximately 1 in 5,000 to 1 in 100,000, with each group varying in frequency. Ultimately, hypermobile EDS (hEDS), which implicates an autosomal dominant inheritance pattern but has no known associated gene mutation, is considered the most common subtype with an estimated prevalence of 1 in 3,100-5,000 [3,4]. The diagnosis of hEDS, considered the least severe form of EDS, is clinical, given the absence of a decisive genetic aetiology for which to test in most patients [5]. However, a definitive diagnosis is complicated due to the broad clinical presentations associated with hEDS, which may result in misdiagnosis or improper patient management [6]. Furthermore, as of recently, GI manifestations such as abdominal pain, reflux disease, diarrhoea, constipation, gastritis, diverticulitis, and rectal evacuation disorder have been identified as possible symptoms associated with hEDS [6,7]. Affecting the skin, bones, and blood vessels among numerous other tissues and organs [1], clinical manifestations of EDS include hypermobility of the joints, skin hyperelasticity, blood vessel fragility, and atrophic scarring [8]. Hypermobile EDS is inherited in an autosomal dominant pattern, with the responsible genes not having yet been identified, making it imperative to develop an accurate clinical diagnosis. The diagnosis of hEDS requires the concurrent presence of these three criteria: 1) generalised joint hypermobility; 2) proof of syndromic features, musculoskeletal issues, and/or family history; and 3) eliminating alternative diagnosis [8]. Other features such as functional GI disorders have been described in cases of hEDS but are not considered specific enough to be incorporated into the formal diagnostic criteria [9].

This report focuses on a 31-year-old female with a past history of mast cell activation syndrome, microscopic colitis, POTS, eosinophilic oesophagitis, dysthymia, and hEDS who presented to her local hospital following a prolonged bout of abdominal pain, nausea, and diarrhoea. A study by Leganger J et al., comparing EDS versus a control cohort found that GI co-morbidities including hernias and functional disorders were more common among those with EDS [10]. More specifically, patients with hEDS are more susceptible to GI structural and functional afflictions such as rectal prolapse, ptosis of internal organs, diaphragmatic hernias, intestinal intussusceptions, dyspepsia, chronic constipation, gastroesophageal reflux, and irritable bowel disease [11]. In a retrospective study by Nelson AD et al., performed at Mayo Clinic in which patients diagnosed with hEDS and other EDS subtypes were analysed to assess the syndrome's association with GI manifestations, it was revealed that 86.5% of those diagnosed with EDS and GI symptoms were female with a mean age of 29.6 years [6]. Of the 687 patients diagnosed with EDS, 378 (56%) had GI manifestations, with 271 (71.7%) of those individuals having hEDS. Approximately 58% of patients diagnosed with hEDS reported GI symptoms, with abdominal pain being the most common symptom among all EDS subtypes and affecting 56.1% of those with the hypermobile subtype. Other symptoms included nausea, heartburn, constipation, and diarrhoea. The 31-year-old female in this report, who experienced these symptoms along with the others identified above, underwent an abdominal CT scan. Imaging displayed a 2.9 cm soft-tissue mass possibly emerging from the terminal ileum, which was revealed to be a desmoid fibromatosis, a non cancerous growth that arises in the connective tissue [12]. Subsequently, given the worsening of symptoms, a follow-up CT disclosed an enlargement of the ileocecal desmoid tumour, which now measured 5.1 cm. All subtypes of EDS share features such as tissue fragility, joint hypermobility, and skin hyperextensibility [3]. However, these manifestations are insufficient to establish an absolute diagnosis, and imaging modalities are essential for proper identification as well as patient management. The most common CT with contrast abdominal findings in patients with hEDS includes renal abnormalities, obstruction of the small bowel, peritoneal hernia, acute colitis, pancreatitis, and appendicitis [8]. Additionally, diverticulitis, diverticulosis, hepatic cyst/mass, and diaphragmatic hernia have been detected in CTs with and without contrast in individuals with hEDS [8,13].

CONCLUSION(S)

Available literature shows a clear correlation among patients with EDS, specifically the hypermobile subtype, and GI manifestations. This case presentation discusses a patient with hEDS that demonstrated manifestations commonly seen throughout the literature, including abdominal pain, nausea, and diarrhoea. However, this patient was also found to have an ileocecal desmoid fibromatosis. Given the patient's medical history and the involvement of EDS and desmoid tumours with connective tissue, a strong linkage is suspected between the desmoid fibromatosis being examined and the patient's diagnosis of hEDS. Unfortunately, there is not sufficient literature available to incontrovertibly attribute the tumour to EDS. Future research should study this association.

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