Spontaneous Diaphragmatic Rupture in Hypermobile Type Ehlers-Danlos Syndrome

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1. Introduction

Ehlers-Danlos Syndrome (EDS) refers to a spectrum of inherited connective tissue disorders affecting the skin, joints, and blood vessels. Abnormal collagen formation or deficiency leads to excess joint mobility, cutaneous fragility, and poor wound healing. The hypermobile type (EDS type III) primarily presents with hypermobile joints and mild skin manifestations. We present a case of spontaneous diaphragmatic rupture in a patient with EDS type III.

2. Case Description

A 27-year-old woman with EDS type III, depression, gastroesophageal reflux, and hypothyroid disease was transferred to our institution with a four-day history of left chest and left upper quadrant abdominal pain. Prior to transfer, she underwent treatment for pneumonia. A thoracentesis for a left parapneumonic effusion was attempted; however, upon obtaining blood, a thoracic CT scan (Figure 1) was performed showing a diaphragmatic rupture with splenic herniation. On arrival, the patient was tachycardic and tachypneic, and she complained of severe left-sided pain with worsening shortness of breath. The patient was taken to the operating room. Upon entering the abdomen, the hernia was visualized with spleen and omentum incarcerating into a 6 × 4 cm defect in the posterolateral left hemidiaphragm. After reduction of the hernia and evacuation of blood in the left hemithorax, the diaphragmatic defect was repaired primarily with large, nonabsorbable (#1 Prolene) interrupted figure of eight sutures. Additionally, a thoracostomy tube was placed to ensure adequate reexpansion of the lung and evacuation of the pleural space. Afterwards, the patient was extubated without difficulty. The patient had an uneventful recovery with gradual improvement in her pain. The thoracostomy tube was removed on postoperative day 5, and she was discharged home the next day.

3. Discussion

EDS is a group of inherited disorders characterized primarily by hyperextensible skin and hypermobile joints, caused by abnormal collagen forms or density. The incidence is estimated at 1 in 5,000 births. Classification is based primarily upon expression of tissue involvement, mode of inheritance, and chemical analysis. Classic (EDS type I, 40%; EDS type II,
Diaphragmatic and/or chest wall destruction, in either running or interrupted fashion. In the setting of diaphragm repair with permanent suture (size 0, #1) is advocated for better exposure. The management of potential complications (perforation, strangulation, and necrosis of the herniated organs) can depend on the presentation (classical, hypermobile, and vascular type) but share some common elements. With the collagen defects, all have poor and delayed wound healing with potential for atrophic scar formation. Additionally, the tissue may not hold suture well, especially with joint related surgeries. The vascular type has potential life-limiting issues with aneurysm formation and difficulty in repair due to tissue friability. Furthermore, an increased risk in pregnancy related complications, including uterine rupture, has been seen with vascular type EDS. Bleeding disorders are not uncommon due to tissue friability, including easy bruising/mucosal bleeding, despite normal coagulation factors/platelet function.

While this is one of only a handful of cases presented worldwide, it is also the first ever reported case of spontaneous diaphragmatic rupture in a patient with EDS type III [5–9]. It is important to include diaphragmatic rupture in the differential diagnosis for patients with connective tissue disease and acute onset tachypnea and pain, as this complication has the potential for significant morbidity without prompt surgical intervention.

**Conflicts of Interest**

The authors declare that they have no conflicts of interest.

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