Case Report

Ehlers-Danlos Syndrome Presenting as Airway Obstruction and Hoarseness

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Abstract

Ehlers-Danlos syndrome is an autosomal dominant genetic defect in connective tissue synthesis and structure. The genetic disorder is classified into six major types based on clinical signs and symptoms as well as genetic testing. We describe the case of a 42-year-old female who presents with dyspnea and hoarseness. After an anterior glottic web was discovered, the attempts to remove the web created excess scarring. She was referred to a geneticist and diagnosed with classic Ehlers-Danlos syndrome and showed signs of vascular type. Genetic disorders should be considered in differentials when poor healing or abnormal scarring occurs post surgery.

Keywords

Ehlers-Danlos, dyspnea, hoarseness, excessive airway scarring, glottic web

1. Introduction

Ehlers-Danlos syndrome (EDS) is described as an autosomal dominant genetic defect in connective tissue synthesis and structure. EDS was first described in the 4th century BC by Hippocrates who noted nomads with joint laxity and “burn-like scars” [1]. Physician Frederick Parkes Weber officially named the disorder in 1936 after two dermatologists who had previously described the signs and symptoms: Ehlers (1901) and Danlos (1908) [2]. EDS has since been researched and grouped into “types” based on clinical signs and symptoms [3].

Often, EDS is diagnosed due to joint hypermobility, skin extensibility, and friable tissues. However, due to the wide range of signs and symptoms in the various types of EDS, the diagnosis is often missed or delayed. Although the estimated incidence of EDS is 1:5,000, it may be higher due to the amount of patients with mild disease who do not seek clinical assistance [4].

We describe an unusual case of excess scarring in a female patient who originally presented complaining of dyspnea and hoarseness.

2. Case report

A 42-year-old Caucasian female initially presented to the office with progressive dyspnea and a long history of a hoarse voice. Prior to her scheduled visit, she had developed a spontaneous small bowel perforation secondary to adhesions, which required urgent surgery. Due to concern about her airway, a tracheostomy was placed at the time of discharge.

At the first appointment, the patient weighed 85 lbs. Additionally, she was noted to have considerable nasal obstruction bilaterally. Severe collapse of the middle third of her nose, a deviated nasal septum, and severe airway obstruction were noted. During the first evaluation of her larynx with a rigid and flexible scope attached to a videostrobe, it was noted that she had a significant anterior glottis web that involved 50% of her vocal cords, which resulted in decreased abduction of both vocal cords (Figure 1).

After a weight gain of 9 lbs, the patient underwent a microdirect laryngoscopy with repair of the anterior glottic web and appeared to be healing well. Six weeks later at a follow-up appointment, the patient presented with scarring and webbing of the anterior commissure (Figure 2).
to develop postoperative scar hypertrophy and spontaneous skin keloid-like lesions [10, 11].

The scarring and webbing found in our patient initially had no known etiology. The vestibular stenosis in her nose, the anterior glottic web, and the prior admission to the hospital for bowel adhesions were suspicious for a genetic healing disorder in retrospect. The excessive scarring post-op on our patient’s anterior glottis was also attributed to her EDS. We suggest that unexplained abnormalities in healing should prompt consideration of a genetic disorder.

**Conflict of interest** The authors declare that they have no conflict of interest.

**References**


