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Tracheobronchomalacia in a Down Syndrome Patient: A Fatal Anomaly

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Abstract

Down syndrome is a common chromosomal abnormality associated with many different structural anomalies. Tracheomalacia results in tracheal collapse when increased airflow is demanded due to the flaccidity of the tracheal support cartilage. Sufficient research and data are currently not available on airway malformations associated with Down syndrome, including tracheomalacia. In this case report, we discuss a Down syndrome patient with pneumonia found to have narrowing of his airway which resulted in prolonged hospital stay and difficulty in management. In most children with Down syndrome, tracheomalacia, tracheobronchomalacia, and bronchomalacia are all under diagnosed, yet these anomalies are more commonly missed in adults with Down syndrome. This can result from the patient's presentation, which tends to be non-specific, and unless a specific obstruction is considered, it is unlikely that such forms of narrowing will be discovered. Severe adult tracheomalacia is a severe condition that can be difficult to treat, especially when associated with an airway infection that may result in increased mortality rates.

Keywords: Down syndrome; Airway; Respiratory; Tracheomalacia; Bronchomalacia

Introduction

Tracheomalacia and even bronchomalacia occur when there is insufficient cartilage that allows maintenance of airway patency during respirations. It is characterized by increased expiratory collapse due to the destruction or decline in the elastic fibers of the trachea by the actual reduction in the integrity of tracheal cartilage [1]. This results in tracheal collapse during expiration. Surgical treatment can include tracheostomy [1], stent placement [2, 3], or even external tracheal stabilization [4]. Tracheomalacia can be a common cause of persistent wheezing in infancy, however, can be a rare finding in adults including those with Down syndrome, which makes it a dangerous disease to manage. It results in high mortality rates, especially when associated with airway infections.

Adult airway malacia may be classified into congenital or acquired forms, including those resulting from chest trauma, tracheostomy, inflammation, chronic irritation, malignancy or mechanical anatomical factors [5]. One of these congenital forms includes Down syndrome; however, it is unusual to diagnose it in an adult. Although there is some association with Down syndrome, it remains a rare anomaly that is under diagnosed.

Case Report

In this article, we discuss the case of a 52-year-old male with a past medical history of Down syndrome, epilepsy, ischemic stroke with residual right-sided hemiparesis, and multiple prior admissions for pneumonia who presented to the emergency room in respiratory distress with a productive cough and fever of 101 °F. As the patient is non-verbal due to history of ischemic stroke, information was obtained from family members. As per the patient's family, the patient had difficulty swallowing his puree diet and developed increasingly worsening congestion 2 days prior to admission. There were no prior signs of dysphagia, sleep disturbances, or findings suggestive of airway malacia in the past. The patient was unable to tolerate his oral medications, resulting in several episodes of vomiting. The patient did not have any history of smoking, alcohol or drug abuse, and was cared for by his family. Upon physical examination, he was found to have a pulse of 102, BP 100/60, temperature 99.2 °F, respiratory rate 22, and saturation of 97% on 3 L nasal cannula. Significant findings showed a patient in moderate respiratory distress with bilateral wheezing and rhonchi to auscultation.

Laboratory data were significant for a white blood cell count of 12.2 and a pro-BNP of 243.9. Chest X-ray demon-

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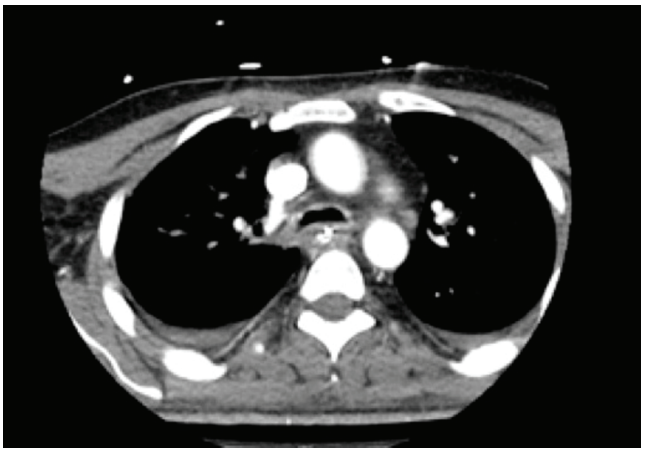

Figure 1. Neck CT demonstrating tracheal narrowing.

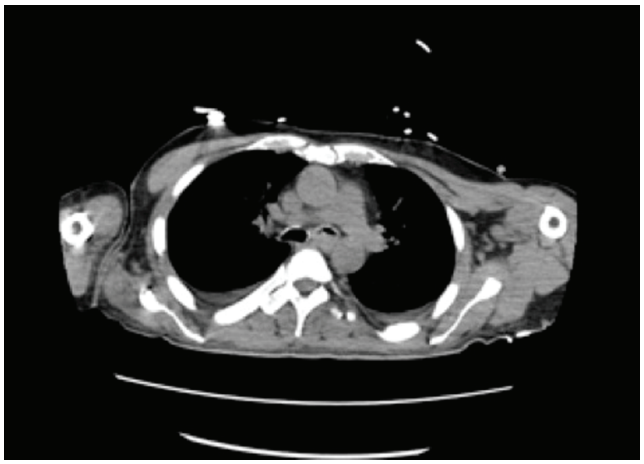

Figure 2. CT chest illustrating tracheal and left main bronchus narrowing.

strated persistent bilateral infiltrates suggestive of pneumonia. Patient was admitted to the critical care unit and started on duo-nebulizer treatment, azithromycin, and ceftriaxone. The patient had a breakthrough seizure despite prophylaxis with keppra and was intubated for airway protection. Sputum cultures returned positive for *Proteus mirabilis* and *E. coli*. While on antibiotics, he continued to have thick purulent secretions. CT scan of the neck (Fig. 1) and chest (Fig. 2) were conducted to rule out any foreign objects or signs of obstruction.

CT scan of the chest illustrated diffuse parenchymal opacities representing pneumonia. However, it also showed a tracheal abnormality signifying a narrowed lumen that may be representative of tracheomalacia. The trachea and left main bronchus were both narrowed (Fig. 1). There was a posterior impression on the trachea for a length of 4 cm above the carina and extending into the left main bronchus. A repeat CT of the neck confirmed tracheomalacia with tracheal collapse during expiration (Fig. 2). As a result of the tracheomalacia and severity of his pneumonia, the patient was unable to be successfully weaned from the ventilator and his ICU stay was prolonged as there was failure to liberate him from the ventilator. A tracheostomy was performed for adequate suctioning of the airway and to manage his secretions. Thereafter, the patient showed significant improvement along with the completion of antibiotic treatment.

Discussion

Such observations found in a patient with Down syndrome brought into discussion whether this anomaly is common in adult patients with such a condition. Severe tracheomalacia and tracheobronchomalacia are increasingly recognized as a cause for respiratory failure in the intensive care unit and prolonged hospital stay; however, the diagnosis is often overlooked, as chest radiography appears normal, and the role of invasive diagnostic testing for this diagnosis is not well illustrated [6]. Down syndrome is seen in the literature to be linked to narrowing of the trachea especially in newborns but is rarely diagnosed in adults. As seen in this patient, it results in a poor cough mechanism, increased secretions, and pneumonia, which can prolong a hospital stay and have an increased mortality rate if not diagnosed early.

Other similar findings that have been associated with Down syndrome include but are not limited to macroglossia, narrowed nasopharynx, adenotonsillar hypertrophy, flattened mid-face, low tone (hypotonia) of upper airway muscles causing dysphagia, tracheal stenosis, and even subglottic stenosis. There is no clear explanation of the presence of airway malacia in Down syndrome patients due to lack of research and published data. Symptomatic atlanto-axial instability is considered to be one explanation that is estimated to occur in 1-2% of patients with Down syndrome. It may result in irreversible spinal cord damage and it has also been documented that Down syndrome patients have a greater number of osseous anomalies of the upper cervical spine than do age- and sex-matched healthy children [7]. Laxity of the transverse atlantal ligament has been thought to be the primary cause of the instability [8].

Although the techniques and criteria for the diagnosis of tracheomalacia are not standardized, CT and bronchoscopy are the usual diagnostic tools implemented in prior published studies [5, 9]. In regards to the degree of airway stenosis, a narrowing of less than 50% is considered within normal limits, 50-75% as mild, while 75-90% is moderate and 91-100% indicates severe malacia [3, 5].

Surgery is considered the first line treatment in tracheomalacia or tracheobronchomalacia. Stent placement may be beneficial for symptom palliation in patients in which surgery is contraindicated or refused [9]. Stents are also beneficial both for symptom improvement by stabilizing the airway and for the determination of patients who will benefit from further surgical intervention [9]. Intratracheal stents are divided into two major types consisting of silicone stents and shape-memory alloy stents. Silicone stents are easily implanted and removed but are associated with several complications including infection, expectoration, and tendency to undergo migration, and restenosis [6]. Tracheal stent placement may be useful in improving ventilator dependence; however, as there is a risk with stent placement due to excessive laxity of tracheal sup-

portive tissue, sometimes tracheoplasty may be considered a better mode of intervention [6]. There still remains uncertainty whether these risk factors are increased in Down syndrome patients in comparison to the general population.

In cases of prolonged respiratory failure, such as the one presented here, a tracheostomy was a beneficial and successful form of management to improve the patient's condition. Ethical challenges in surgical interventions, when there may be insufficient caregivers in older patients, need to be taken into consideration when discussing treatment options with family members. Weighing the risks and benefits is an important concern as most patients who require surgery may need assisted living or to be placed in a long-term care facility. However, due to the benefits seen with treatment, especially in life-threatening conditions such as ventilator dependent respiratory failure, the options may be limited.

There remains a lack of literature describing causes of this anomaly and methods to improve its diagnosis. Down syndrome is one condition associated with narrowing of the trachea, and as in this case even the bronchus. Awareness of this complication in adults is not well known and thus can result in a prolonged hospital stay and increased risk of complications. However, unlike most cases of Down syndrome where airway narrowing is found in infants with persistent wheezing, it is unusual for it to be found in adulthood. By raising awareness of its presence in patients with such a condition, it can improve the management of such patients and can increase their overall survival rate. Diagnostic guidelines and even the options of standardized treatment are various but not well established, especially in patients with congenital conditions such as Down syndrome. With early diagnosis and rapid management, the hospital stay and prevention of further complications can be prevented.

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Competing Interests

There are no competing or conflicting interests associated with this article.

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