



Dystrophic Epidermolysis Bullosa with Laryngotracheal Involvement: Abscess on the Vocal Cords

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Abstract

Background: Dystrophic Epidermolysis Bullosa (DEB) is a rare genodermatosis characterized by blister formation as a result of separation of the skin from underlying tissue. The mucous membranes can be involved in DEB. Laryngotracheal involvement is rare.

Case Report: A 4-year old girl with episodes of respiratory problems over a long period of time was admitted to hospital. She had severe asthmatic attacks without complete recovery while using an inhaled bronchodilator together with inhaled steroid continuously. Her family was living in Iraq. Vital signs were stable. The patient had blisters with erosion and scar formation on the skin, especially at the pressure sites on the patient's extremities. Her toenails and fingernails were thickened and dystrophic. The patient had no respiratory distress on examination, however, inspiratory stridor was determined instead of expiratory wheezing. Laryngoscopic examination was performed in surgery and an inflammatory abscess was detected on the vocal cords.

Conclusion: A patient with an abscess on the vocal cords because of infected blisters due to DEB is presented.

Keywords: Children; Childhood; Dystrophic epidermolysis bullosa

Introduction

Epidermolysis Bullosa (EB) is a rare hereditary disease characterized by widespread blistering of the skin and mucous membranes. In patients suffering from EB, trauma or friction causes separation of the skin from underlying tissue resulting in consecutive painful blisters, scarifications, contractures, and pseudosyndactyly. The prevalence of EB is reported as 11.07 cases in United States, 54 cases in Norway, 7.8 cases in Japan, 15.4 cases in Italy, 10.3 cases in Australia and 9.6 cases in Croatia per one million live births [1]. EB of the skin results from genetic defects in molecules related to adhesion with loss of adhesion resulting in blister formation. These may occur anywhere on the body but most commonly appear at friction and minor trauma sites such as the feet and hands. In some subtypes, blisters may also occur on internal organs, such as the esophagus and respiratory tract. The mode of inheritance may be autosomal dominant or recessive but all forms of dystrophic EB (DEB) result from mutations in the gene encoding type VII collagen, COL7A1 [2]. EB can be described as a genodermatosis. There are three major types of EB based on the site of blister formation within the skin: EB Simplex (EBS), Junctional EB (JEB) and DEB. EBS results from mutations within either the keratin 5 or keratin 14 gene. JEB-Herlitz (JEB-H) is caused by severe mutations within any of the three genes which encode for laminin-332. A minority of non-Herlitz JEB (JEB-nH) have mutations within the gene encoding for type XVII collagen. However, in DEB, there are no mutations hotspots within the COL7A1 gene. Every family with DEB has its own unique type of mutation. If there is a mutations known in the family, It would facilitate in DNA mutational analysis, especially for prenatal diagnoses using specimens obtained from chorionic villi [3]. The site of blister formation within skin in DEB is the lamina densa and upper dermis. Mild pseudosyndactyly or flexion contractures of the fingers are present in 6% of those with autosomal dominant DEB. The frequency of Dominant DEB (DDEB) was 68%, with 19% of uncertain inheritance and 13% of recessive DEB (RDEB) [4]. The prognosis depends on the subtype of the disease. Infants and children with inherited EB, particularly those with JEB, are at significant risk of death as a result of complications [5]. Cases with EBS and DDEB have normal life expectancies with significant morbidity. Diagnostic tests are biopsy of a blister on the skin or mucous membranes which involves Immunofluorescence Antigen Mapping (IFM) and/or Transmission Electron Microscopy (TEM) [3,6]. The diagnosis of EB is most obvious for physicians experienced in dermatology. However, some cases will need differential diagnosis. Any blistering disorder in the newborn period may mimic EB. These include

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Figure 1: Blisters with erosion and scar formation on the skin.

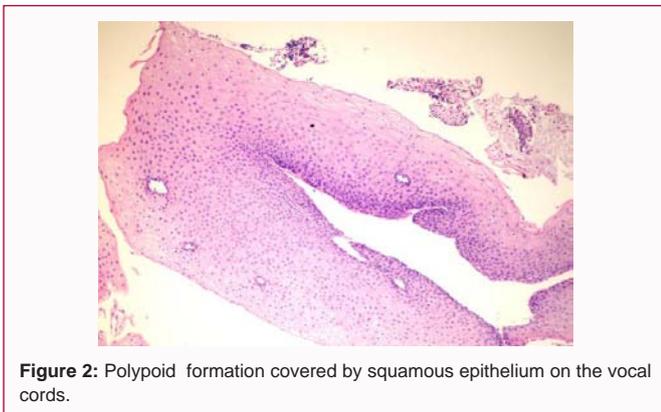


Figure 2: Polypoid formation covered by squamous epithelium on the vocal cords.

Herpes simplex virus, epidermolytic ichthyosis, bullous impetigo and incontinentia pigmenti. Acquired disorders for differential diagnosis are immunobullous disorders, EB acquisita, Linear IgA dermatosis, bullous pemphigoid, cicatricial pemphigoid, pemphigus, infectious diseases, *Herpes simplex*, *staphylococcal* scalded skin syndrome and bullous impetigo [3].

Currently, there is no cure for EB. The most important issues in basic care are prevention of blistering and secondary infection of blisters. Band-aids, bandages, or tape should not be used on the skin. Skin fragility is characteristically worsened in warm weather. Patients should avoid exposure to heat and humidity. If possible, air conditioned environments should be recommended. Hand deformities can be prevented by meticulous nightly wrapping of the digits. If not performed, surgical procedures will be necessary. There are some experimental approaches; *Ex Vivo* replacement for autosomal recessive EB, transplantation of allogeneic fibroblasts for RDEB, transplantation of bone marrow-derived stem cells or infusion of recombinant protein (i.e., type VII collagen for RDEB) [3]. The mucous membranes can also be involved in EB. The highest risk of upper airway strictures and obstruction occurs in JEB-H. Recurrent esophageal blistering and erosions can lead to stricture formation [3,7,8]. However, laryngotracheal involvement is rare in EB. This case report presents involvement of the vocal cords in EB in a 4-year old girl complaining about recurrent respiratory distress.

Case Presentation

The patient had been experiencing episodes of respiratory problems for a long time and occasionally had severe asthmatic attacks without complete recovery. She was using an inhaled bronchodilator

together with inhaled steroid continuously. In her family history, her maternal uncle had the same findings since birth. She was the fourth children of the same parents and other children in the family did not have any problems. Her family was living in Iraq and she was admitted to a different hospital for respiratory distress which never completely resolved. The patient was 4-years old when admitted to the pediatric clinic in Baskent University Hospital, Istanbul (Turkey). An examination revealed stable vital signs. The recurrent respiratory distress, skin fragility and nail abnormalities were major complaints. On examination, her weight was 15,500g and height was 101 cm. Failure to thrive and growth retardation were not determined. On inspection, the patient had blisters with erosion and scar formation on the skin, especially on the pressure sites of her extremities (Figure 1). In the case presented here, blister scars were commonly observed on the feet and hands at pressure sites of friction and minor trauma. She had significant dental decay in the mouth. Her toenails and fingernails were thickened and dystrophic. Milia were not present. The patient had no respiratory distress on examination; however, inspiratory stridor was determined instead of expiratory wheezing. She had used inhaled bronchodilator and inhaled steroids continuously for a long time. She may have complained of long-standing stridor, but not asthmatic attacks. She had minor lymphadenopathies. On auscultation, cardiac pansystolic murmur was determined. An atrial septal defect was diagnosed by a cardiologist. Chest radiography was completed, blood tests including CBC, BUN, creatinine, AST, ALT and urinalysis were evaluated as normal. She did not have anemia.

She was admitted to our hospital for laryngoscopic examination with anesthesia. The mucous membrane in the mouth and oropharynx were normal. Laryngoscopic examination was performed by an otolaryngologist in surgery in order to check the supraglottic structures, vocal cords or trachea in terms of separation of the mucous membranes from underlying tissue with minor trauma. The fragility of mucous membranes can make laryngoscopic examination risky because shedding by the blisters might be expected to produce upper airway obstruction or strictures of upper airways. The clinicians were aware of the risks. Similar lesions to those on the skin were observed in the larynx and an inflammatory abscess was identified on the vocal cords. Infected materials and tissue in the blister were aspirated and sent to the pathology department. Two hours later, severe respiratory distress suddenly occurred because of upper airway obstruction due to shedding of blisters. She was administered oxygen therapy for two hours, recovered with coughing attacks, was monitored for one whole day and then discharged. She was monitored for one week before returning to her country. Pathologic evaluation revealed blister cleavage below the basal lamina and subepithelial separation (Figure 2). Supportive therapies, future risks and genetic basis of this disease were explained to her parents. Genetic analysis could not be performed.

Discussion

EB is a rare hereditary disease involving the skin. However, certain types of EB can also affect the respiratory system. Involvement of the oropharynx is more common than laryngotracheal involvement. Even if mucous membrane involvement is extensive, airway obstruction does not contribute to mortality in these patients. Recurrent respiratory distress has been wrongly interpreted as asthmatic attacks in some children with EB. A child suffering from inspiratory stridor was diagnosed with a blister abscess on the vocal cords in EB and is presented as a rare case in our paper.

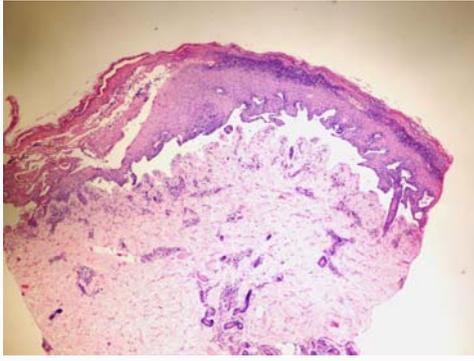


Figure 3: Hyperkeratosis, hypergranulosis and subepidermal separation in skin biopsy.

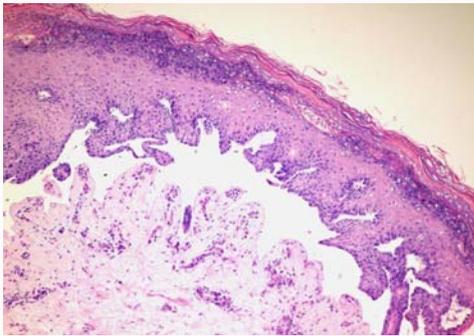


Figure 4: A larger image of skin biopsy.

Collagen VII anchoring fibrils are a component of the airway basal membrane. They are an integral part of the supracellular anchoring network that attaches the epithelium to the extracellular matrix. Reduction of collagen VII in EB is associated with sloughing of epithelium [9]. This means that respiratory tract involvement can be found in certain types of EB. Lesions can be detected on the mucosa of the respiratory tract from the oral cavity to the bronchi. The oral cavity and oropharynx are involved more frequently than the hypopharynx, larynx and trachea. Involvement of laryngeal and tracheal mucosa is generally associated with increased morbidity and mortality, complications and therapeutic difficulties [10]. These are more common in JEB and DEB than in EBS [5]. JEB may involve the upper airway, especially in JEB-H. Granulation tissue which is a pathognomonic finding in JEB-H was not present in our patient. In DDEB, blistering is often mild and limited to hands, feet, knees, and elbows, but nonetheless heals with scarring. Dystrophic nails are common and may be the only manifestation of DDEB [11]. In our case, toenails and fingernails were thickened and dystrophic. Failure to thrive, growth retardation and severe anemia which are characteristic features of RDEB and JEB-H were not determined in our patient [3]. The patient was diagnosed as having dominant DEB on the basis of clinical, familial and histological findings). The diagnosis was supported by biopsy (Figure 3 and 4).

Laryngotracheal involvement should be considered for EB in any child presenting with symptoms of respiratory distress and laryngoscopic examination should be considered and performed in the operating room by an otolaryngologist. However, clinicians should be aware of the risks of shedding by the blisters due to the endoscopy.

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Verbal permission was obtained from the family to publish pictures of their child.

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