EPIDERMOLYSIS BULLOSA ACQUISITA, A TONGUE TWISTER… LITERALLY!

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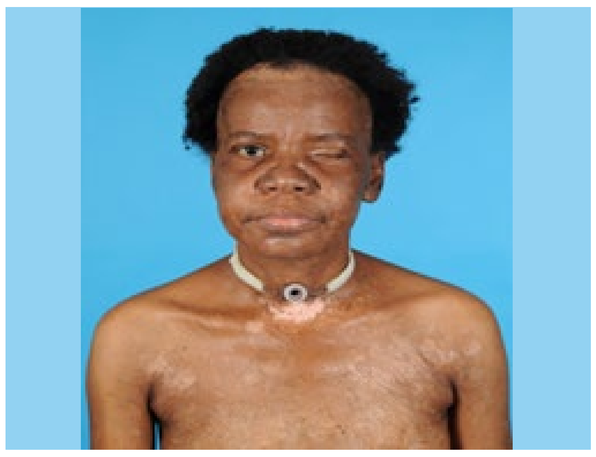
INTRODUCTION

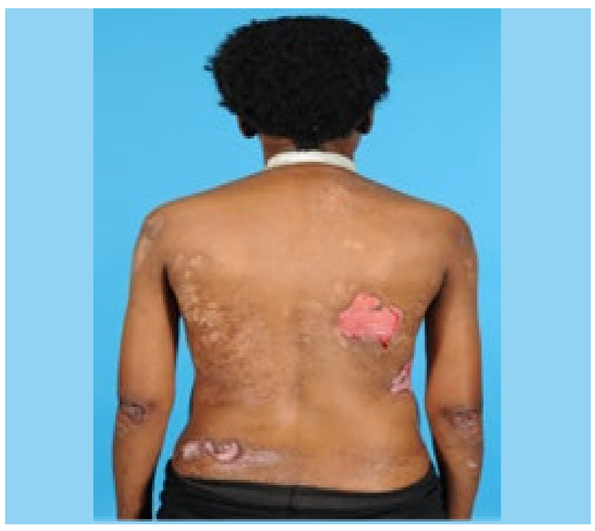
Epidermolysis bullosa acquisita (EBA) is a rare, acquired subepidermal, mucocutaneous blistering autoimmune disease. It is due to the production of Immunoglobulin G (IgG) against type-VII collagen, a major component of anchoring fibrils in the basement membrane zone of dermo-epidermal junction of the stratified squamous epithelium. This results in loss of dermo-epidermal adhesion manifesting as vesicles and bullae in the skin, and mucosal erosion which may include the oral cavity, nasal cavity, eyes, pharynx, larynx and oesophagus.

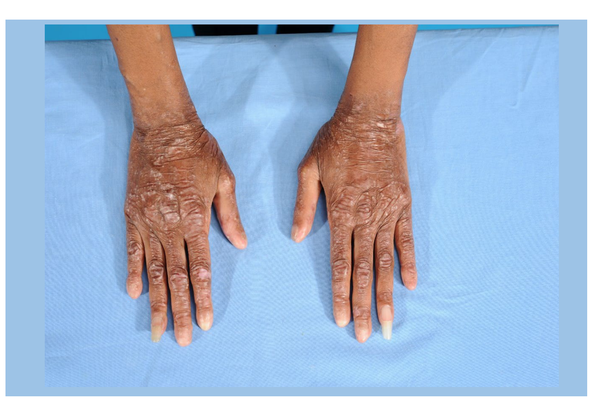
CASE REPORT

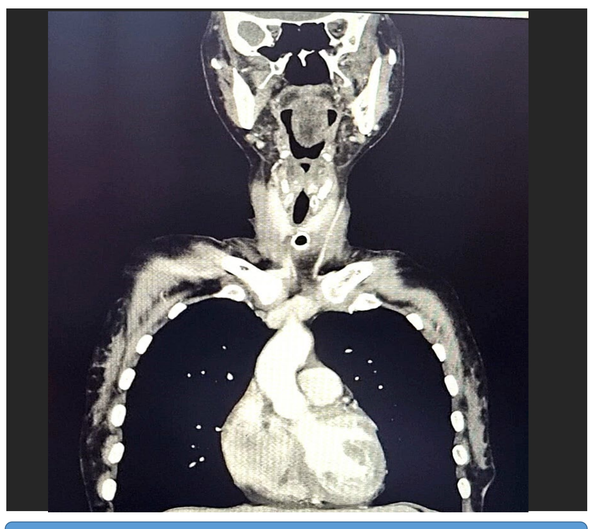
A 42-year-old female patient was referred from Dermatology clinic with a 3- month history of progressive hoarseness, dysphagia, and difficulty in breathing. These symptoms were preceded by bullous eruptions on the torso, limbs and face, with left symblepharon. The patient had a background history of Retroviral Disease, on Highly Active Antiretroviral Therapy.

Clinically the patient had bullous skin lesions, milia and hypopigmented patches. She also presented in respiratory distress and had an inspiratory stridor. Fibreoptic scope revealed nasal crusts, and synechiae, erosive oropharyngeal lesions, and supraglottic stenosis. Emergency tracheostomy and direct laryngoscopy & biopsy were done. The glottis, subglottis and trachea had no lesions. Histopathology report of epidermolysis bullosa acquisita was obtained.

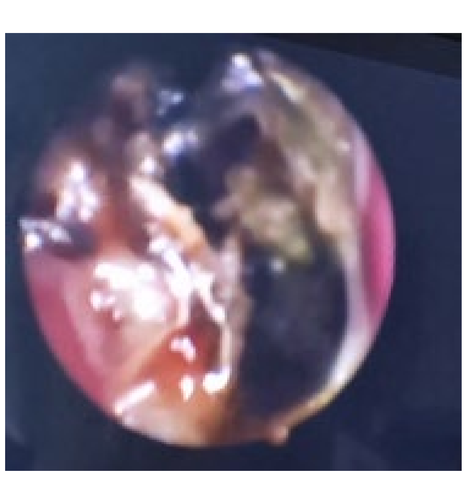




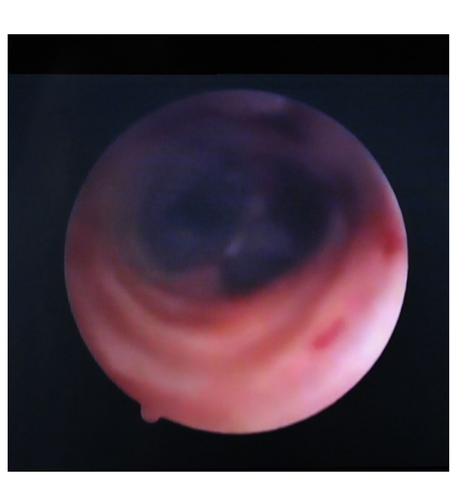




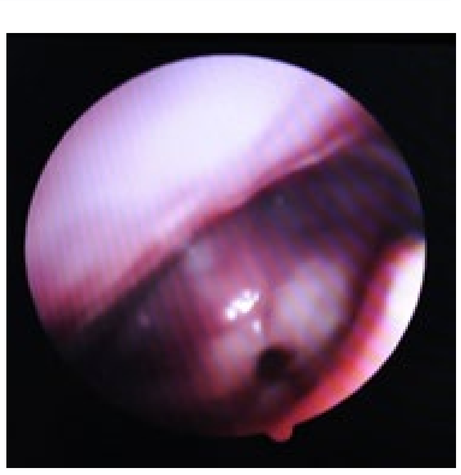
Coronal CT, supraglottic stenosis



Nasal crusts and synechiae



trachea



Supraglottic stenosis

DISCUSSION

EBA is an orphan autoimmune disease with an incidence of 0.08- 0.5/million. There is no sex predilection, and it occurs across all age groups (peak 4^{th} – 5^{th} DOL) Cutaneous disease is most commonly reported, with mucosal involvement being infrequently reported. Laryngeal involvement is uncommon. There are two main clinical forms, mechanobullous and inflammatory EBA.

Due to the rarity of EBA, there is a paucity of literature, current management recommendation is limited to case series, reports and expert opinion:

Diagnosis is based on clinicopathological correlation (clinical features-skin fragility, blister formation, scarring and milia. Mucosal involvement: cicatricial lesions of the nose, mouth, genitals and anus, oesophagus, larynx, trachea, bronchi, and conjunctiva. Histopathology- subepidermal cleavage, oedema, neutrophils, eosinophils and lymphocytes. Direct immunofluorescence- linear deposits of IgG and C3 at the BMZ. Immune electron microscopy- Gold standard,shows autoantibodies below lamina densa. Salt-split skin. Indirect immunofluorescence- circulating autoantibodies. ELISA).

Differential diagnosis: Porphyria cutanea tarda, bullous pemphigoid, mucous membrane pemphigoid, bullous SLE.

Treatment is multidisciplinary, includes, prevention of new lesions and cicatrization (which is irreversible), early diagnosis, medical therapy- *1^{st} line-* immunomodulators: Dapsone, Colchicine. Systemic corticosteroids and intravenous immunoglobulin, for severe disease. *2^{nd} line- systemic steroids,* immunosuppressants (mycophenolate mofetil, azathioprine, cyclosporin) for severe disease. 3^{rd} line- rituximab. Surgery (including tracheostomy for upper airway obstruction, oculoplastic surgery, removal of synechiae, esophageal dilation, or gastrostomy, if there is severe stenosis).

CONCLUSION

EBA is a chronic disease with high morbidity, potential for severe complications, and adverse impact on quality of life. A high index of suspicion is warranted in a patient presenting with mucocutaneous bullous lesions and upper airway obstruction.

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