

Epidermolysis Bullosa Acquisita in a patient with End Stage Renal Disease: Case study

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Abstract

Epidermolysis Bullosa Acquisita (EBA) is a rare disorder characterised by sub-epithelial mucocutaneous blistering. Its presentation may be in the form of inflammatory bullous eruption or non-inflammatory tense bullae, milia, and scarring. EBA is an immune disorder mediated with antibodies against type VII collagen, which is a major component of basement membrane zones of the skin and mucosa. Adults are more commonly affected; however, a few paediatric cases have also been reported. Risk factors for its development are still under study. Very few cases are seen of EBA in patients with End Stage Renal Disease and its association with this chronic illness is still under study. This case report presents the case of a patient whose lesions were initially misinterpreted for Calciphylaxis, but histological diagnosis revealed inflammatory EBA.

Keywords: Epidermolysis bullosa acquisita, End stage renal disease, Inflammatory bullous eruption.

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Introduction

EBA is a rare disorder with unknown prevalence. Antibodies against type VII collagen found in basement membrane of the skin and mucosa are usually present. Genetics (HLA-DR2) may play a role in the pathogenesis of EBA.^{1,2} The disease usually has a very long course and complications such as blindness, oesophageal strictures, and joint contractures may occur.³ Hands, feet, elbows, knees, and lower back are most commonly involved.⁴ The blisters often heal with scarring and small epidermal inclusion cysts referred to as milia.⁴

Case Report

A 52-year-old male, previously hypertensive and on maintenance dialysis, presented to the outpatient department of Shaikh Zayed Hospital, Lahore, in February 2020 with complaints of multiple wounds and small ulcers

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on both his hands and feet as shown in figures 1 and 2. He had been put on renal replacement therapy five years back because of end stage kidney disease secondary to bilateral small kidneys. Cause of renal failure was suggestive of chronic glomerulonephritis as evident by the documented history of body swelling and proteinuria previously. His PCR was positive for hepatitis C, although no signs of decompensation were observed and thus he was put on Direct Acting Anti-Viral (DAAV) therapy in the form of Sofosbuvir and Declatsvir. After a month of treatment with DAAV he developed life-threatening anaemia and his antivirals had to be discontinued. He is currently having thrice weekly four-hour haemodialysis sessions through right arm brachiocephalic AVF with history of left brachiocephalic fistula failure.

Two months ago, lesions developed at the fingertips of the hands and feet progressing to the involvement of legs as well. These lesions initially were just erythematous lesions with foul smelling discharge, although some lesions were followed by crusting and, later on, a black eschar formed over them as shown in figures 1 and 2.

On the basis of examination, a diagnosis of calciphylaxis was reached at. So, necessary lab investigations i.e. calcium, phosphate, intact PTH, MIBI scan and Doppler ultrasound of both upper limbs were done. His calcium phosphate product was elevated up to 65, with intact PTH of 598.8pg/ml and MIBI scan showed no evidence of hyper-functioning parathyroid gland. His Doppler ultrasound showed left upper limb thrombus formation in Basilic vein which was extending up to cubital fossa. Fistulogram



Figure-1: Multiple hypo and hyper pigmented skin lesion on dorsum of hands. Lesions with crusting and super-imposed black eschar.



Figure-2: Multiple hypo and hyper pigmented skin lesions on dorsum of foot. Lesions with crusting and super-imposed black eschar.

revealed normal study of the right AVF. Histological diagnosis was planned in collaboration with the dermatology department and specimen was taken from a lesion over the lateral aspect of the foot, which revealed epidermolysis bullosa acquisita which was inflammatory in nature. He is currently on oral Prednisolone 5mg once daily, and Cinacalcet to reduce his parathyroid hormone levels along with antiplatelet therapy.

Discussion

Epidermolysis bullosa acquisita (EBA) is a rare autoimmune sub epithelial blistering disorder.⁵ Clinical presentation is usually in the form of skin fragility and bullae formation having association with various systemic diseases. Diagnosis can be made by history, examination, investigations and by histological study of the lesion. Histology may reveal vacuolation along the dermo-epidermal junction with surrounding oedema at the early stages and may lead to dermal fibrosis at the later stages of the disease. Immunofluorescence shows linear deposits of IgD and complements, however, small amount of IgA, Factor B and properdin can also be deposited. Management includes general measures of skin care and medications like systemic steroids, Colchicine, Dapsone, and others for refractory disease.^{6,7}

A case report of EBA showed 79-year-old male resident of the Northern Mariana Islands who presented with multiple blistering skin lesions.⁸ Likewise, a 54-year-old Caucasian male was diagnosed with EBA who presented for the evaluation of a mucocutaneous blistering eruption that had evolved over a period of three years. Both these patients showed partial response to systemic corticosteroids.⁹ In Pakistan, there are 20 reported families with epidermolysis bullosa and one case was discovered to have EBA. Another case of EBA in Pakistan was reported of a 46-year-old male who had a history of three months of pruritic blistering eruptions at the upper back, scalp and neck, elbows, and knees.¹⁰

Conclusion

Though rare, EBA is notorious for its prolonged course; this

patient had it for around two years. This patient has been put on oral Prednisolone. Follow-up of the patient revealed mild improvement in the lesions with no evidence of steroids induced toxicity. Hence, steroids were continued with advice of regular follow up. So, early initiation of systemic steroids in inflammatory and a combination of Colchicine plus Dapsone in case of non-inflammatory variety can decrease its severity.

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Consent: consent of the patient for publishing the case report was taken.

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