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Case Report

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Novel case of ANCA Negative Pauci-immune Crescentic Glomerulonephritis in Recessive Dystrophic Epidermolysis Bullosa

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Abstract

We report a 24-year-old male with Recessive Dystrophic Epidermolysis Bullosa (RDEB) who presented with acute kidney injury, proteinuria and hematuria. Serological workup was negative. Renal biopsy revealed crescentic Glomerulonephritis (GN) without evidence of immune complexes, vasculitis, or amyloid deposits. This is the first report of Anti-Neutrophil Cytoplasmic Antibodies (ANCA) negative pauci-immune crescentic GN associated with RDEB. Management with immunosuppressive medications can be challenging in patients with RDEB due to high risk of skin infection. In our case, his renal function and proteinuria improved with pulsed methylprednisolone followed by careful tapering of oral corticosteroid.

Keywords: Acute kidney injury; Crescentic glomerulonephritis; Epidermolysis bullosa; Proteinuria

Background

Recessive dystrophic epidermolysis bullosa (RDEB) affects approximately 2.5 per million live births in the United States [1]. It is a rare genetic skin disease due to a genetic mutation in COL7A1, causing a type VII collagen defect and is characterized by the presence of extremely fragile skin and recurrent blister formation [1]. Kidney diseases have been associated with RDEB. There have been fewer than ten reported cases of renal dysfunctions in patients with RDEB. These cases include post- infectious Glomerulonephritis (GN), secondary renal amyloidosis, IgA nephropathy, membranoproliferative GN, and genitourinary tract stricture. Given the rarity of this disorder, information on RDEB associated-kidney diseases, their management and prognosis remain scarce. To our knowledge, this is the first case report of ANCA negative pauci-immune crescentic GN in a patient with RDEB and its successful management.

Case Report

A 24-year-old male with recessive dystrophic epidermolysis bullosa diagnosed in infancy was referred for proteinuria. His past medical history included multiple skin infections. There was no significant family history for skin or kidney disease. Physical examination revealed ectropion of eyelids and mitten deformity of hands and feet. His entire body had well-healed scars and multiple blisters without evidence of active infection. Laboratory findings showed serum creatinine (SCr) 1.2mg/dl, urinalysis showed 3+ protein, 4-10 WBC and >50 RBC per HPF. Urine Protein to Creatinine Ratio (UPCR) was 5.1. Serological work up showed negative antinuclear antibody, double-stranded DNA. HIV, Hepatitis B and C serology, Rapid Plasma Reagin (RPR), Anti-Neutrophil Cytoplasmic Antibodies (ANCA), Anti-Glomerular Basement Membrane (GBM) antibody, Anti-Streptolysin O antibody, SPEP, and UPEP were all negative. Complement levels were normal. Kidney ultrasound showed normal sized kidneys with increased echogenicity.

A kidney biopsy was performed to determine the cause of this patient's proteinuria and impaired renal function. Crescentic GN with cellular crescents was observed in at least 4 viable glomeruli; 10 out of 19 glomeruli were globally sclerotic. There were mild interstitial fibrosis and tubular atrophy. A linear pattern of IgG was inconclusive due to high background staining. Weak mesangial C3 was present without evidence of other immune complexes, vasculitis or amyloid. No deposits were seen by electron microscopy. The biopsy diagnosis was ANCA negative pauci-immune crescentic GN. The patient was treated with pulsed dose intravenous methylprednisolone sodium succinate 1g/day x 3 days followed by oral prednisone taper over the course of four months

and enalapril. He was closely followed by nephrology and dermatology clinics during his treatment. His renal function and proteinuria improved after four months (SCr 0.92mg/dl, UPCR 2.15). During treatment, there was no development of skin infection or sepsis (Figures 1-4).

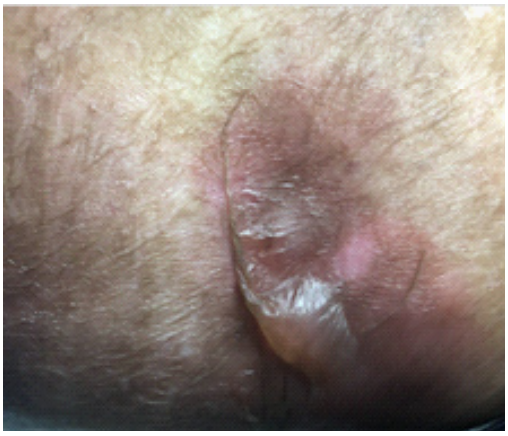

Figure 1: Bullous skin lesion.

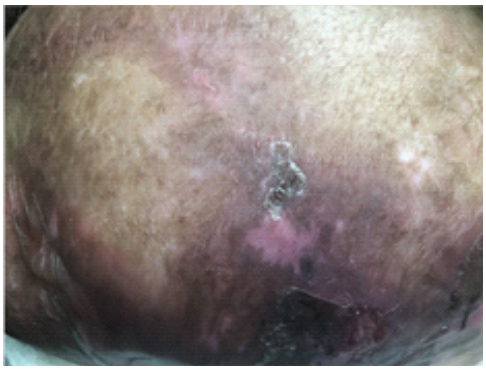

Figure 2: Scarring of ruptured blisters.

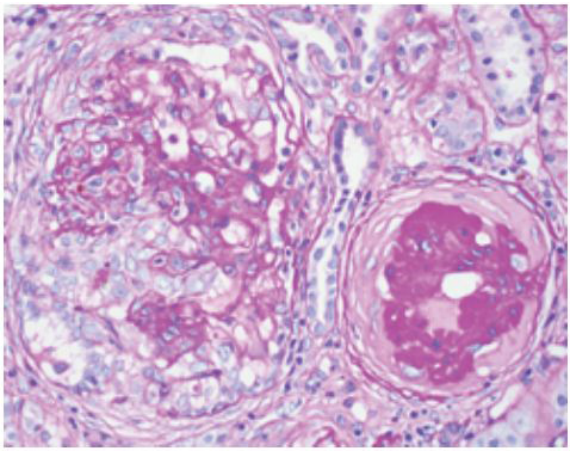

Figure 3: Light microscopy showed crescentic glomerulonephritis.

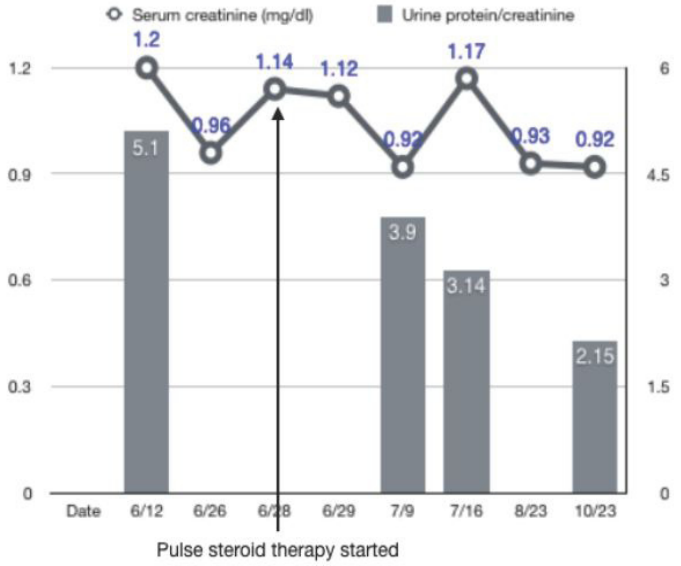

Figure 4: Serum creatinine and urine protein to creatinine ratio trends.

Discussion

Epidermolysis Bullosa (EB) is a heterogeneous group of diseases inherited in an autosomal dominant or an autosomal recessive manner, characterized by fragility of the skin and mucosa. There are four major Epidermolysis Bullosa (EB) types: EB simplex, junctional EB, dystrophic EB, and the Kindler syndromes [2]. Dystrophic Epidermolysis Bullosa (RDEB) is a rare autosomal recessive disease caused by a lack of expression of type VII collagen. Renal dysfunctions have been sporadically reported in patients with RDEB. The kidneys are believed to be injured in several ways. Chronic ureteral blister formation and strictures may lead to hydronephrosis. Glomerulonephritis may also develop in these patients. Previously reported glomerular disorders associated with RDEB include secondary renal amyloidosis and immune complex-medicated glomerulonephritis such as post- infectious glomerulonephritis, IgA nephropathy and membranoproliferative GN [3-6]. These glomerular disorders are thought to develop in relation to chronic inflammations and repeated skin infections [7]. Our patient is the first reported case of ANCA negative pauci-immune crescentic GN in patients with recessive dystrophic epidermolysis bullosa. Management of GN in these patients can be particularly challenging due to high rates of skin infection associated with RDEB, which can be further exacerbated by immunosuppressive therapy. This case illustrates that good short-term renal and patient outcome can be achieved with careful dosing of immunosuppressive medication while closely monitoring for the development of skin infection during treatment.

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