

# RENAL AND URINARY TRACT INVOLVEMENT IN PATIENTS WITH EPIDERMOLYSIS BULLOSA

N. Yildiz, N. Cicek Deniz, Ruslan Asadov<sup>1</sup>, İ. Gökçe, AD. Yucelten<sup>2</sup>, H. Alpay

Marmara University Medical Faculty, Department of Pediatric Nephrology, Radiology<sup>1</sup> and Dermatology<sup>2</sup>

## Introduction

Epidermolysis bullosa (EB) is a rare inherited disorder characterized by bullous skin lesions and fragility to minor traumas, and may present various systemic involvement. Blisters and vesicles may occur at mucosal surfaces of respiratory, genitourinary and gastrointestinal systems. Several renal complications including IgA nephropathy, amyloidosis, postinfectious glomerulonephritis, hereditary nephritis, and upper and lower urinary tract obstruction have been reported in these patients with EB. We aimed to evaluate renal and urinary tract involvement of patients with EB.

## Material and Method

We evaluated 13 patients with EB. Serum samples for creatinine, cystatin C, sodium, potassium, phosphorus and urine samples for urinary beta-2 microglobulin, microalbumin, sodium, phosphorus and creatinine were obtained. Fractional excretion of sodium (FeNa), tubular reabsorption of phosphorus (TRP) and glomerular filtration rate (GFR) were calculated. Urinary system ultrasonography was performed. We examined glomerular and tubular functions of patients with EB.

Table 1

	Gender	Age (year)	Serum Creatinine (mg/dl)	Serum Cystatin C (mg/dl)	GFR ml/min/1.73 m <sup>2</sup>	Um.a./cr (mg/gr)	Ubeta2/cr (micgr/gr)	TRP (%)	FeNa (%)
Patient 1	M	17.7	0.76	0.9	135	7.7	77	92	0.88
Patient 2	M	11	0.39	0.6	101	11.2	40	82	0.3
Patient 3	M	4.7	0.34	0.68	189	11.1	50	97	0.25
Patient 4	M	14.5	0.4	0.81	286	7.1	83	93	0.04
Patient 5	M	20.2	0.29	0.75	136	16.5	390	93	0.63
Patient 6	F	28.7	0.54	0.77	172	6.78	49.2	90	0.04
Patient 7	M	14.2	0.48	0.8	191	9.6	98	91	0.43
Patient 8	M	4.5	0.26	0.68	205	75	102	95	0.26
Patient 9	M	4.5	0.22	0.77	250	11	302	90	0.07
Patient 10	M	6	0.32	0.71	180	8.2	250	92	0.02
Patient 11	F	20.7	6	-	3.7	1	5000	32	25.6
Patient 12	F	22	0.31	0.6	265	9.1	2332	92.5	0.75
Patient 13	M	1.6	0.23	0.75	198	25.3	65.8	98	0.17

## Results

The mean age of the patients were 13.10±8.28.7) years. Mean serum creatinine, cystatin C were 0.81±1.67 mg/dl, 0.73±0.08 mg/dl and respectively and were normal in all patients except one. Mean urinary beta-2 microglobulin/creatinine was 679.9±1436 µg/g and it was high in three patients. Mean microalbumin/creatinine was 15.35±18.8 µg/g and it was normal in all patients. TRP was low in all patients. One patient has high FeNa and low TRP (Table 1).

Renal involvement was present in two patients. The first patient was a five year-old male with hydronephrosis who has bullous lesions within the bulbar and urethra leading partial obstruction on cystoscopy. The second patient was a 21 year-old female with nephrotic proteinuria. Renal biopsy of the first patient revealed focal segmental sclerosis. Her renal function deteriorated progressively and end stage renal disease developed. She is on continuous peritoneal dialysis therapy for 15 months. Twelve patients had normal renal functions and eleven patients had abnormal ultrasonography findings. One patient with renal failure had small kidneys with increased echogenicity and one patient who had bullous lesions in bladder and prostatic urethra had bilateral hydronephrosis.

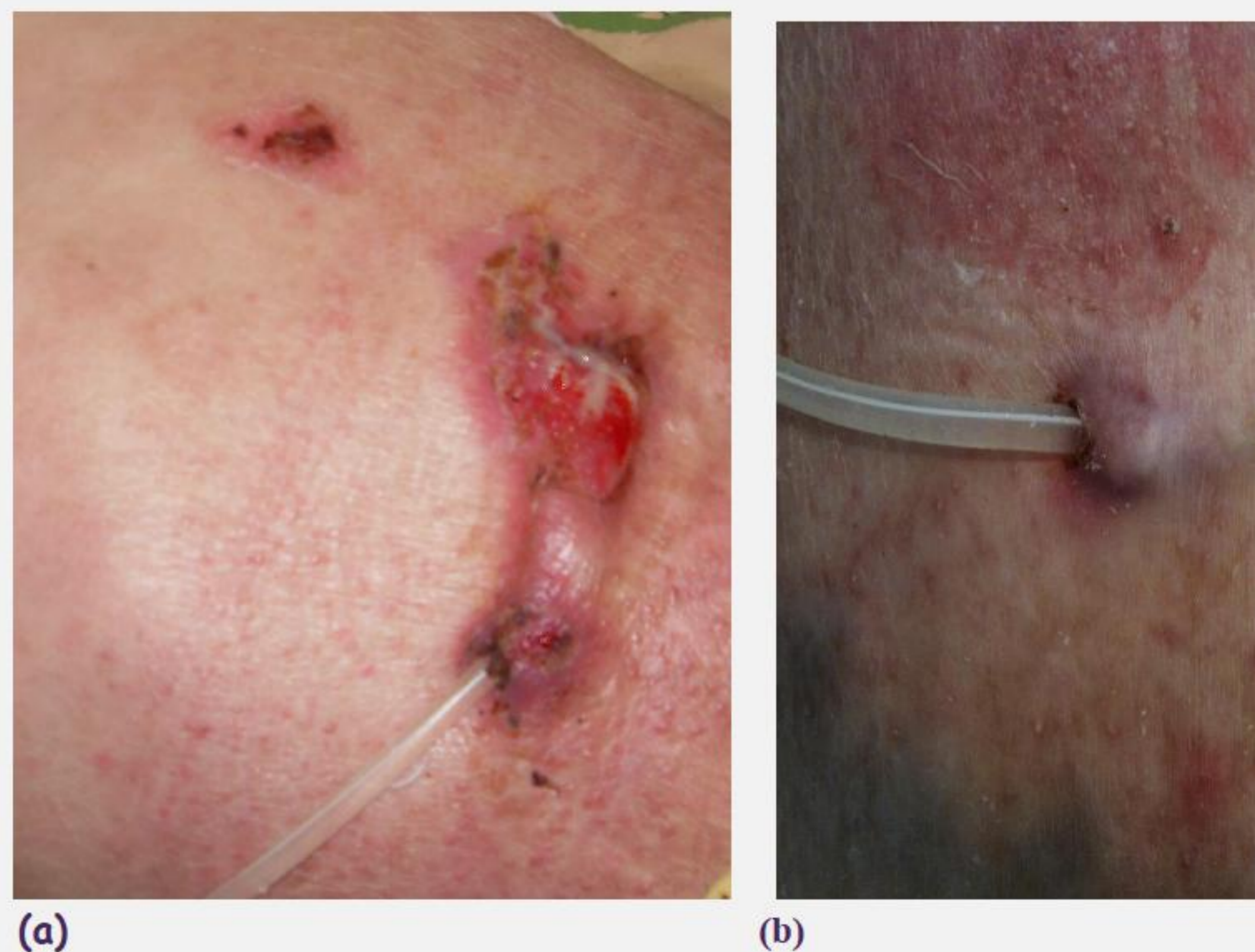


Figure 1: Peritoneal catheter postoperative, on 10 day (a) and on 1 month (b)

## Conclusions

We recommend that evaluation of renal and urinary tract involvement of patients with EB of any subtype should be included in the routine evaluation of the disease.

