Turk J Gastroenterol *2014; 25: 450-1*

A very rare cause of diarrhea: Epidermolysis bullosa

To the Editor,

Many gastrointestinal and systemic diseases may cause chronic diarrhea. The evaluation and management of diarrhea can be complex (1). Prolonged diarrhea sometimes can cause life-threatening complications and also death. The management of chronic diarrhea depends on the underlying disease. We present a very rare cause of chronic diarrhea that can be confused with gluten enteropathy and treated with causative drug of diarrhea.

A 29-year-old man presented with non-bloody, watery diarrhea for 6 months. He had a congenital disease, epidermolysis bullosa (EB), characterized by recurrent cutaneous blisters, multiple erosions, scars, and phalanx atrophy (Figure 1). His past medical history had an operation for squamous cell skin cancer, severe skin bacterial infection many times, and being on hemodialysis treatment for 1 year.

Physical examination revealed growth retardation (BMI: 11 kg/cm^2) and multiple blisters on the skin. Severe anemia (hemoglobin: 6.8 g/dL), high creatinine level (3.7 gr/dL), and hypoalbuminemia (albumin: 2.7 gr/dL) were detected. Colonoscopy and stool studies were unremarkable. Antibodies for gluten disease were negative. Gastroscopy showed mucosal blisters in the esophagus and scalloped folds and micronodular and granular mucosa (Figure 2) of the duodenum, resembling gluten enteropathy. Infiltration of the lamina propria with eosinophilic amorphous deposits and amyloid-associated (AA) deposits dyed with congo red were detected on pathologic examination of

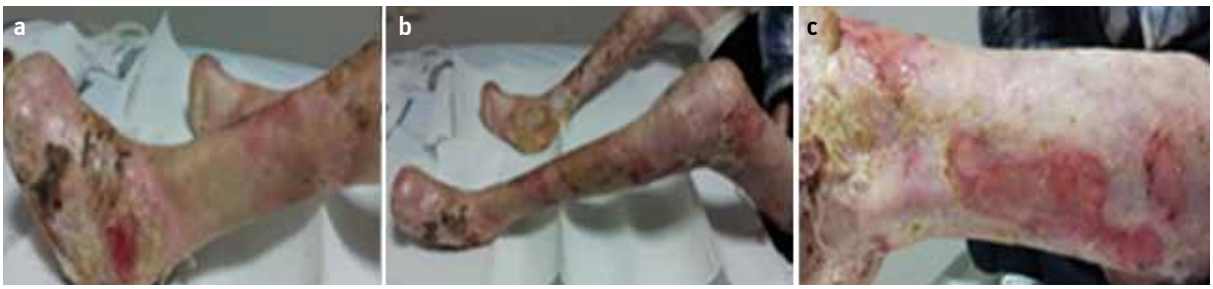

Figure 1. a-c. Prominent cutaneous blisters, multiple erosions, scars, and phalanx dystrophy.

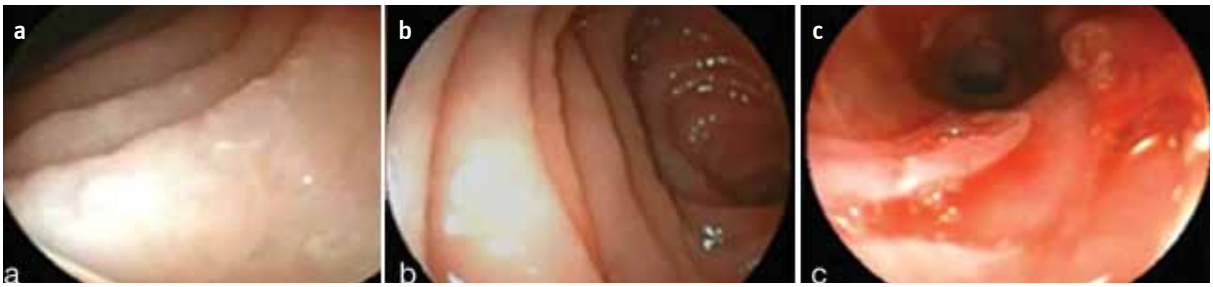

Figure 2. a-c. Upper gastrointestinal endoscopic view of the duodenum, scalloped duodenal folds, and micronodular and granular appearance of the mucosa **(a, b).** Mucosal stripping and blister formation affecting the esophageal mucosa **(c).**

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Received: 12.1.2014 **Accepted:** 7.2.2014

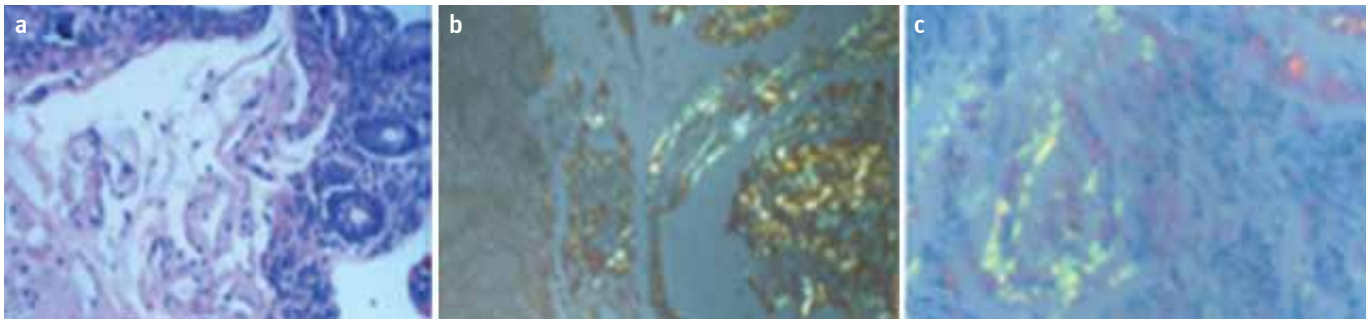

Figure 3. a-c. Diffuse infiltration by eosinophilic amorphous deposits into the lamina propria with crystal violet dye **(a).** "Prominent green highlights" with congo red dye in polarized light, indicating prominent mucosal amyloid deposition **(b, c).**

the duodenal biopsy (Figure 3). Colchicine was started at 1.5 mg/d. The complaint of diarrhea improved within 2 months. Also, serum creatinine level decreased to 1.9 gr/dL.

Epidermolysis bullosa is rare inherited genetic and debilitating disorder of the skin and mucosa, described by blister formation, which arises from an abnormal response of mechanical trauma and dystrophic changes. Three forms are described: simplex, junctional, and dystrophic (2). The dystrophic form, the most severe clinical form, is associated with severe cutaneous and extracutaneous complications due to extensive scarring and recurrent infections. Dystrophic epidermolysis bullosa (DEB) results from mutations of type VII collagen, a major component of anchoring fibrils holding the epidermis and dermis together. In the course of the disease, severe skin infections, skin squamous cell carcinoma, malabsorption, and secondary systemic amyloidosis (SSA) can develop (3).

Secondary systemic amyloidosis may develop in various chronic inflammatory disorders, but it is a rare complication of DEB (4). The postulated mechanism is recurrent skin infections; so, it causes overproduction of the amyloidogenic precursor. Colchicine is an effective medication of DEB-related amyloid nephropathy (5). Colchicine should be the first choice in this case because of the benefits that have been shown in a few cases (4). This case is the second report that "clinical symptom improvement" is shown with colchicine in diarrhea of DEB-related secondary amyloidosis.

Ethics Committee Approval: N/A.

Informed Consent: N/A.

Peer-review: Externally peer-reviewed.

Author contributions: Concept - T.A., G.D.; Design - T.A., G.D.; Supervision - Y.Ü., S.A.; Resource - T.A., G.D.; Materials - T.A.; Data Collection&/or Processing - T.A., G.D.; Analysis&/or Interpretation - T.A., G.D.; Literature Search - T.A.; Writing - T.A., G.D.; Critical Reviews - Y.Ü., S.A.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

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