A man with macroglossia and hemorrhagic bulla

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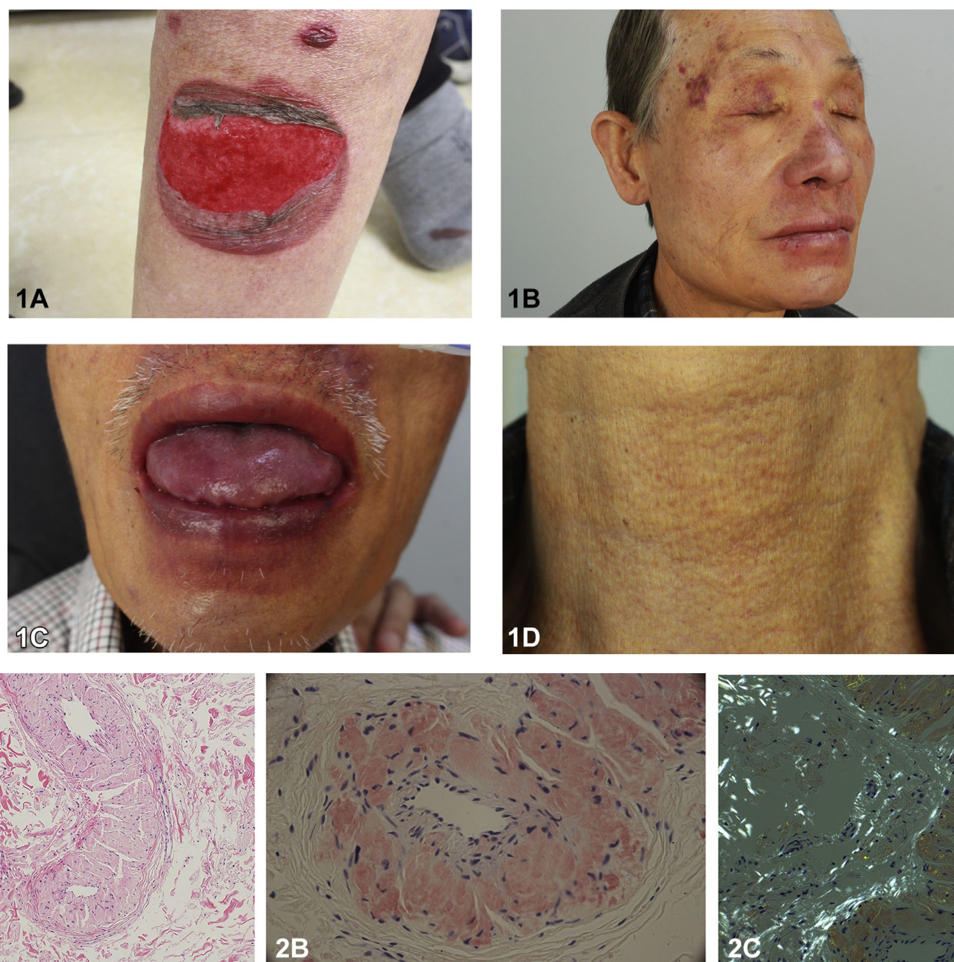

A 79-year-old man presented with ruptured large bulla on right shin (Fig 1, *A*). He had a recent history of heart failure and consulted us for wound care. On clinical examination, multiple purpuric patches based on yellowish thickened skin were found on his face, and macroglossia was observed (Fig 1, *B* and *C*). Laboratory evaluation found elevation of serum-free lambda chain. A punch biopsy specimen was obtained from the neck (Fig 1, *D*) and special staining was performed. The results are shown in Fig 2. Immunofluorescence study for the bullous skin lesion found no deposition.

Question 1: What kind of histochemical stain was performed for diagnosis?

A. Periodic Acid–Schiff (PAS)

B. Masson trichrome

C. Fontana-Masson

D. Alkaline Congo red

E. Oil red O

Answers:

A. PAS — Incorrect. PAS stain is useful for glycogen, neutral mucopolysaccharides, and fungal infection.

B. Masson trichrome — Incorrect. Masson trichrome stains collagen.

C. Fontana-Masson — Incorrect. Fontana-Masson stain is performed in pigmentary disorders.

D. Alkaline Congo red — Correct. Amyloid is stained with Congo red and shows birefringence under polarized microscope.[1,2] In Fig 2, the vessel wall is thickened and shows fragmented appearance, and eosinophilic amorphous materials are found in vessel wall. These materials show apple-green birefringence under polarized light. (**A**, Hematoxylin-eosin stain; **B**, Congo red stain; original magnifications: **A**, ×100; **B**, ×400; **C**, ×200.)

E. Oil red O — Incorrect. Oil red O stains lipids into red.

Question 2: What is the most likely diagnosis?

A. Hypothyroidism

B. Bullous pemphigoid

C. Amyloid light chain (AL) amyloidosis

D. Pseudoxanthoma elasticum

E. Dermatomyositis

Answers:

A. Hypothyroidism — Incorrect. Macroglossia can be found in hypothyroidism, but other findings are not.

B. Bullous pemphigoid — Incorrect. Immunofluorescence study did not show immune component deposition.

C. AL amyloidosis — Correct. Certain clinical features such as macroglossia and periorbital ecchymoses are very strongly suggestive of AL amyloidosis.[1,3]

D. Pseudoxanthoma elasticum — Incorrect. Small, yellowish papules and retinal symptoms are characteristics of pseudoxanthoma elasticum patients.

E. Dermatomyositis — Incorrect. Dermatomyositis is characterized by inflammation of the muscles and the skin. Macroglossia is not associated.

Question 3: Rarely, what can be accompanied with this disease?

A. Scleroderma

B. Multiple myeloma

C. Lymphoma

D. Myxedema

E. Inflammatory arthritis

Answers:

A. Scleroderma — Incorrect. Scleroderma can be found usually in IgG monoclonal gammopathy, occasionally myeloma.

B. Multiple myeloma — Correct. A degree of amyloid deposition is seen in up to 15% of patients with myeloma.[4,5]

C. Lymphoma — Incorrect. Pseudolymphoma like Kimura disease has been associated with lichen amyloidosis, but lymphoma is not.

D. Myxedema — Incorrect. Most patients with scleromyxedema have a monoclonal paraproteinemia, typically designated as monoclonal gammopathy of undetermined significance.

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E. Inflammatory arthritis — Incorrect. Inflammatory arthritis is associated with systemic amyloid A amyloidosis.

Abbreviations used:

AL: amyloid light chain
PAS: periodic acid—Schiff

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