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Macroglossia and amyloid deposition of the tongue

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Abstract

Amyloidosis encompasses a group of disorders involving extracellular deposition of an abnormally folded protein. Light chain amyloidosis is a common type of systemic amyloidosis and is typically associated with an underlying plasma cell neoplasm. Here we describe an uncommon presentation of light chain amyloidosis.

Keywords: Macroglossia; Light chain amyloidosis; Amyloid tongue deposition; Bortezomib.

Case description

A 69-year-old man was found to have hyperproteinemia on routine labs, followed by a monoclonal protein spike on urine immunofixation. At that time he declined bone marrow aspiration and biopsy. Six years later, an amyloid-related gastrointestinal polyp was found on routine colonoscopy. By this time he had developed renal failure requiring hemodialysis, congestive heart failure, and macroglossia with characteristic tongue lesions (Figure 1).

An incisional biopsy of the tongue revealed amyloidosis. Bone marrow biopsy showed 7-9% plasma cells with positive amyloid staining by Congo red, consistent with a diagnosis of light chain amyloidosis. He experienced notable improvement in tongue appearance after two years of treatment with the proteasome inhibitor bortezomib [1,2] (Figure 2). Amyloidosis encompasses a group of disorders involving extracellular deposition of amyloid, an abnormally folded protein. Light chain amyloidosis is a systemic disorder most often affecting the renal and cardiovascular systems, as illustrated in this case. Macroglossia is pathognomonic of light chain amyloidosis but only found in 10% of patients with the disease [3]. In some cases it may be the only presenting symptom, highlighting the importance of clinician awareness.



Figure 1: Initial presentation of macroglossia with tongue lesions prior to biopsy.

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Figure 2: Improvement in tongue appearance after two years of treatment with bortezomib for light chain amyloidosis.

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