

Macroglossia: amyloidosis misdiagnosed as angio-oedema

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DESCRIPTION

A 74-year-old Hispanic woman was admitted to our institution, with swelling of the tongue and lips.

She had arrived in the USA 1 week before admission. In her native country, she had been diagnosed with angio-oedema and treated for more than 9 months, with no improvement. The records from the institution, located in the patient's country, were never obtained. The patient's daughter confirmed that amyloidosis was never considered and biopsy was never obtained, and that she had decided to bring her mother to the USA for better care. Physical examination of the patient's head revealed waxy papules on both eyelids. Her tongue was mobile, soft and severely enlarged (figure 1). The lips and oral commissure had multiple ecchymosis. Skin examination showed multiple denuded bullae and petechia in dorsal hands, and bilateral groin and axilla (figure 2). The patient had multiple neck and axillary lymphadenopathies, and bilateral lower extremity oedema with multiple haematomas. Cardiopulmonary and abdominal examination was normal.

A biopsy of the tongue showed a dense stromal collagenous matrix with deposition of amyloid, and



Figure 2 Denuded bullae and ecchymosis in the axilla.

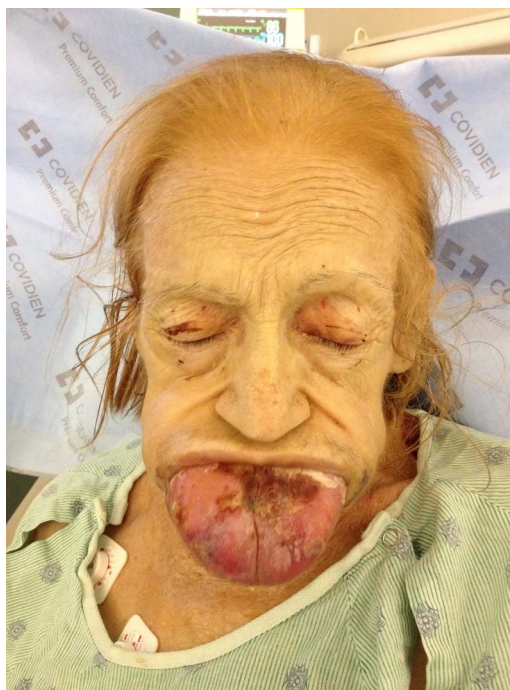


Figure 1 Macroglossia and periorbital purpuric lesion.

a Congo red stain was positive for the characteristic apple-green birefringence of amyloid under polarised light. The results of urine immunofixation suggested amyloid light-chain amyloidosis. Lambda light chain disease, staged as Mayo stage II.^{1 2} An echocardiogram showed left ventricular hypertrophy, biatrial enlargement and thickening of mitral valve leaflets, all consistent with cardiac amyloidosis. ECG showed left anterior fascicular block and low-voltage QRS. CT of the brain showed focal dilation of the right ophthalmic vein with mass effect on the right optic nerve sheath complex and superior rectus muscle.

The hospital course was characterised by hypoxaemia with emergent tracheostomy, pneumonia with Gram-negative bacteria and septic shock, cardiac arrest and anoxic brain injury. The patient died 28 days after admission.

Amyloidosis has a very poor prognosis, with median survival as short as 4–6 months when detected in an advanced stage, or when found with cardiac and hepatic involvement. In our patient, early diagnosis and treatment could probably have prevented other organ involvement. Early diagnosed patients with limited organ involvement can expect a median survival over 5 years.³



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Learning points

- ▶ A combination of macroglossia and periorbital purpura is highly specific for the diagnosis of amyloidosis.⁴
- ▶ A high degree of suspicion is needed to diagnose amyloidosis in a patient with macroglossia, haemorrhagic tendency, lymphadenopathy, low voltage and ventricular concentric thickening in the ECG.
- ▶ Early diagnosis of amyloid light-chain amyloidosis is critical for effective treatment suppressing the production of monoclonal free light chains before irreversible organ damage occurs.

Competing interests None declared.

Patient consent Obtained.

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