Amyloidosis: a rare cause of tongue swelling

An 84-year-old woman presented in August 2010 because of tongue swelling for 2 months. Her medical history comprised hypertension, gout, ischaemic heart disease, and sick sinus syndrome. Her speech was a bit slurred since she noted tongue swelling. Though she could not close her mouth completely due to the mass (Fig 1), she denied any breathing difficulty. The tongue was grossly enlarged with multiple rubbery nodules but no ulceration over its surface. Smaller similar nodules were also present on the lips (Fig 2),but there were no palpable cervical lymph nodes. What are the differential diagnoses?

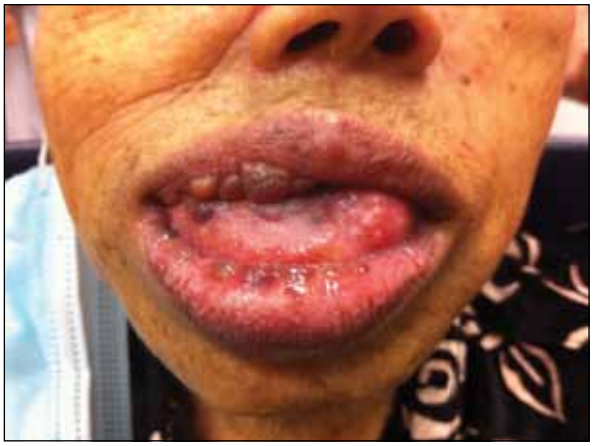

FIG 1. The patient cannot close her mouth completely due to the macroglossia

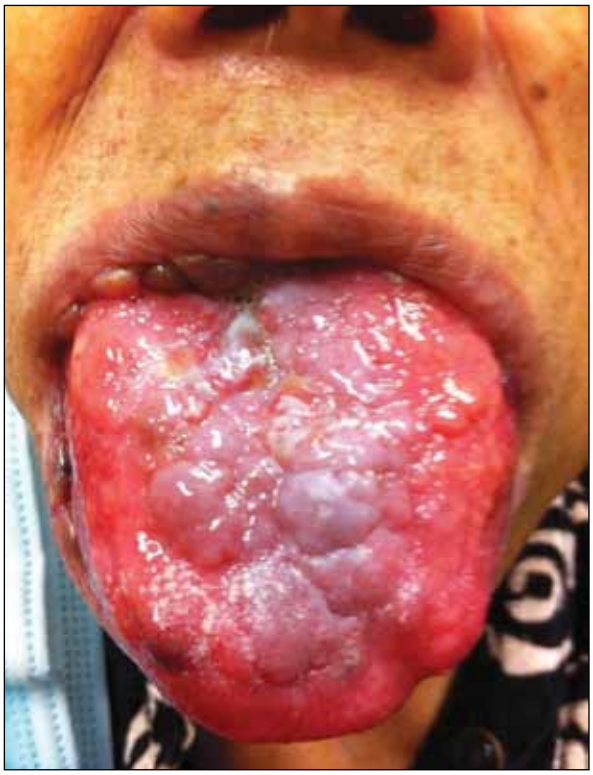

FIG 2. The tongue is grossly enlarged with multiple nodules with smaller nodules on the lips

Vascular or lymphatic malformation can present as a diffuse tongue mass, suggested by a bluish hue or overlying vesicles. Tuberculosis, hypothyroidism, acromegaly, and idiopathic muscular hypertrophy manifest clinically as macroglossia, for which the diagnosis is aided by the medical history and other physical signs. Moreover, the nodular appearance of the macroglossia suggests other pathologies, such as granular sell tumour, neurogenic tumour, lipoma, fibroma, minor salivary gland tumour, and amyloidosis. Imaging might not be able to discriminate among these lesions and definite diagnosis often required biopsy.

Magnetic resonance imaging (MRI) was performed and showed diffuse lingual enlargement without focal signal intensity, no contrast enhancement, and airway was not compressed. Incisional biopsy of the tongue swelling under local anaesthesia yielded aggregates of eosinophilic amorphous material within muscles. Histopathologically, the material was salmon-red after Congo red staining, and yielded apple-green birefringence under polarised light. Thus the diagnosis of amyloidosis was confirmed.

There was no paraproteinaemia but urine Bence-Jones protein was detected with a positive Kappa light chain. The erythrocyte sedimentation rate (ESR) was 102 mm/h and C-reactive protein level 150 mg/L, but all other blood tests yielded nil abnormal. Computed tomography of the thorax and abdomen did not reveal any abnormality. The patient declined further invasive investigations (including bone marrow biopsy) and debulking surgery for the macroglossia, but agreed to out-patient follow-up.

Amyloidosis refers to a spectrum of disorders characterised by deposition of insoluble extracellular proteins in beta-pleated sheets.[1] Diagnosis depends on demonstration of extracellular proteins that appear salmon-red on Congo red staining and apple-green birefringence under polarised light. Accumulation of excessive amyloid within tissues can be clinically deleterious.

Amyloidosis can be classified as localised or systemic. Systemic amyloidosis is further sub-divided into primary and secondary types. The former usually exists in the setting of immune disorders entailing plasma cell dyscrasias.[1] Secondary amyloidosis occurs in various chronic inflammatory or infective

diseases (rheumatoid arthritis, tuberculosis), or as a hereditary or familial disorder.[1]

Evaluation of patients with systemic amyloidosis may encompass testing liver and renal function, urine for Bence-Jones protein, serum for paraprotein and markers of the connective tissue disease, as well as imaging (echocardiogram), and even abdominal fat aspiration biopsy.[2]

Macroglossia can be the first or only manifestation for amyloidosis. Our patient with apparently localised lingual amyloidosis probably had a plasma cell dyscrasia (indicated by presence of Bence-Jones protein in urine). Moreover, most patients with tongue involvement have systemic amyloidosis,[3] though solitary lingual amyloidosis has been reported.[4,5] Distinguishing solitary from systemic tongue amyloidosis is pivotal to offering a prognosis and therapy. The former usually has a good prognosis and recurrence is rare following excision.[1] Conversely, most patients diagnosed with systemic amyloidosis die within a year.[4] When associated with plasma cell dyscrasia, therapy should target the systemic disease.[1] Milder forms of plasma cell dyscrasias (eg monoclonal gammopathy of unknown significance) do not deserve treatment as most patients are asymptomatic and the prognosis is not altered. For multiple myeloma, chemotherapy is started if the patient is deemed able to tolerate the treatment. Though peripheral stem cell transplantation is more effective than chemotherapy alone, it should only be contemplated in patients aged younger than 70 years.[1] When the multiple myeloma respond poorly to therapy, excision should only be contemplated if the tongue lesion imposes significant oral or respiratory distress.

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