

Case Report

A Macroglossia Case with Lingual Amyloidosis

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Abstract

The etiology of macroglossia has a wide spectrum. One rare cause of macroglossia is amyloid infiltration of the tongue. This infiltration generally occurs through systemic amyloidosis. Primary systemic amyloidosis is an abnormal protein infiltration of the extracellular space without a systemic disease, except in the cases of myeloma and hematological disorders. This abnormal protein is produced by the malignant plasma cells of myeloma, which produce excessive amounts of monoclonal kappa and lambda immunoglobulin light chains. Localized infiltration is much more rare. Presently described is the case of a 79-year-old female patient who sought treatment at the outpatient clinic for macroglossia due to amyloid infiltration.

Keywords: Amyloidosis, macroglossia, histopathology

Primary systemic amyloidosis is an abnormal protein infiltration in the extracellular space without a systemic disease except for myeloma and hematologic disorders. This abnormal protein is produced by the malign plasma cells of myeloma, which produce excessive amounts of monoclonal kappa and lambda immunoglobulin light chains. This infiltration may be localized or systemic.^[1]

There are five types of amyloidosis according to its etiology: 1) primary (56%), 2) secondary (8%), 3) localized (9%), 4) multiple myeloma-related (26%), and 5) hereditary-familial amyloidosis (1%).^[5] Amyloidosis is also classified in terms of being systemic or not in the following way: 1) primary systemic amyloidosis, 2) secondary systemic amyloidosis, and 3) localized amyloidosis. Survival is between 5 and 15 months in systemic amyloidosis, while the prognosis is excellent in the localized form.^[4] Oral findings in primary

amyloidosis are about 40%, and especially in the form of macroglossia and nodules in the lips and submandibular glands. Clinical dysphagia, speech disorders, and difficulties with chewing and mouth closure are the most frequently seen problems. In the late period the tongue becomes so big that the patient is unable to fit it in his or her mouth; in the terminal period blockage of the oropharynx and hypopharynx leads to obstructive sleep apnea.^[2, 3]

Case Report

A 79-year-old female patient sought treatment at our outpatient clinic for macroglossia (Fig. 1). In routine ENT physical examination we found macroglossia; no other pathologic findings in the head and neck region were observed. We learned from the patient's history that the size of the tongue began to increase 1.5 years ago. She had difficulties chewing and she could not use her dental prosthesis. We

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referred our patient to the internal medicine department and they diagnosed essential hypertension and chronic obstructive pulmonary disease. We performed MRI examination with a 1.5 Tesla Siemens (Germany) for the neck and mouth floor and found diffuse volume increase of the tongue characterized by extension to the extraoral region. T1A and T2A sequences and contrasted sequences showed no demarcation, indicating no heterogeneity or masses. Based on these findings, the patient must be investigated for diffuse infiltrative diseases like amyloidosis (Fig. 2). In histopathologic examination of the patient using hematoxylin and eosin sections, acellular eosinophilic material deposition was seen in the subepithelial region and especially between the muscle fibrils. In histochemical examina-

tion with crystal violet purplish and Congo red organophilic deposition was seen between the muscle fibrils (Fig 3).

The patient was referred to Cardiology, Nephrology and Internal Medicine to further investigate her systemic amyloidosis and to check for chronic infections like tuberculosis and osteomyelitis. Neither systemic amyloidosis nor chronic infection was found. For the treatment of macroglossia we recommended surgery to the patient, but she did not consent; we warned her about the possible risk for airway obstruction in the future.

Discussion

Different clinical types of amyloidosis can infiltrate multiple organs. The head and neck region is affected between 12% and 90% in amyloidosis. Laryngeal infiltration is usually localized, but lingual infiltration is generally secondary to a systemic disease.^[4-6] According to the affected site in the head and neck region, symptoms like hoarseness, congestion, odinophagia, mandibular deformity, dysphagia, airway obstruction by lingual infiltration, speech and chewing difficulties due to macroglossia, and taste disorders can be seen.^[4] Localized lingual infiltration is rare; most cases of lingual infiltration generally occur due to systemic amyloidosis. Since the size of the tongue increases in lingual infiltration, amyloidosis must be considered in the differential diagnosis of macroglossia. Other diseases that can cause macroglossia are tuberculosis, lymphangioma, hypothyroidism, acromegaly, idiopathic muscular hypertrophy, and Beckwith Wideman syndrome.^[4] Physical examination, patient history, and radiologic examinations are helpful, but definite diagnosis is achieved by histopathologic examination of biopsy material. In histopathologic examination routine hematoxylin and eosin dye and crystal violet and Congo red can be used.^[7] When amyloid infiltration in the tongue is diagnosed, systemic amyloidosis must be investigated by complete blood count, liver and kidney function tests, echocardiography for heart infiltration, serum and



Figure 1. An image of macroglossia. Note that the patient could not fit her tongue into the mouth.



Figure 2. Axial and sagittal MRI views of macroglossia.

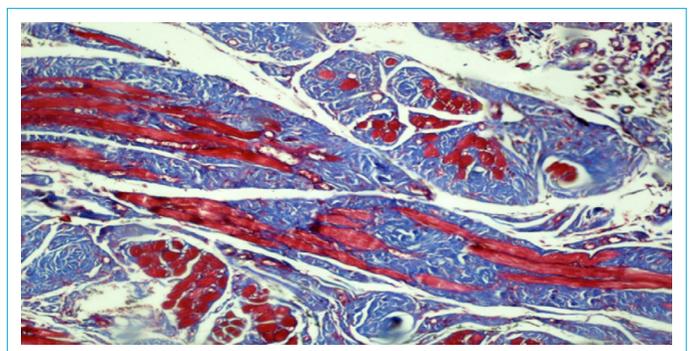


Figure 3. Histopathologic view (crystal violet x40); Note the amyloid collection in the subepithelial space between the muscle fibrils colored in violet.

urine Bence Jones proteins, and abdominal fat and rectum wall biopsies.^[4, 6]

In our case, the onset of macroglossia was between 1.5 and 2 years, but the prognosis of lingual infiltration cases due to systemic amyloidosis is between 5 and 15 months.^[4] Since the referral of the patient for systemic amyloidosis and the negative findings for chronic infections, we thought that our case was localized amyloidosis with lingual infiltration. The etiology of macroglossia has a wide spectrum and macroglossia can occur as a result of a systemic or local disease. Therefore, the reason for macroglossia must be investigated. Amyloidosis must be considered in the differential diagnosis. Lingual amyloidosis is a rare condition; however, once diagnosed, systemic amyloidosis must be investigated by complete clinical tests, including abdominal and rectal biopsies.

Disclosures

Informed Consent: Written informed consent was obtained from the patient who participated in this study.

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Conflict of Interest: None declared.

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