Just a Tongue Anomaly or Something More?

Macroglossia Isolada ou Algo Mais?

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A 54-year-old woman with a past history of breast and uterine cancer, appeared at the emergency room with a six-month history of fatigue, weight loss (> 12 kg), lower limbs swelling and dysphagia due to progressive tongue enlargement. ENT examination demonstrated macroglossia (Fig. 1-A), that was confirmed by computed tomography (CT) scan (Fig. 1-B). Laboratory tests complemented with image findings showed the presence of a restrictive cardiomyopathy and a nephrotic syndrome. The abdominal fat biopsy with immunofixation electrophoresis was positive for light-chain amyloidosis. The patient began chemotherapy treatment with bortezomib, dexamethasone and cyclophosphamide, completing four cycles of treatment with frank tongue reduction. The patient died 5 months after the diagnosis.

Tissue deposition of protein fibrils causes a group of rare diseases called amyloidosis, with light-chain amyloidosis (AL) being the most common type. Although it is considered to be a highly specific finding of AL, macroglossia is only found in about 10%-20% of cases. Tongue nodules, papules or ulcers can also appear in AL patients.[1] In addition to the head and neck region, it can affect important vital organs like the heart, kidneys or liver, potentially causing irreversible damage, and a poor overall prognosis. The diagnosis is usually made with biopsy, with Congo Red staining producing a pathognomonic birefringence green under polarized microscopy. The treatment includes chemotherapy and autologous stem cell transplant.[1,2]

An uncommon clinical finding like macroglossia can be present in tuberculosis, acromegaly, hypothyroidism and genetic syndromes, among others.[1,3] Amyloidosis is an important differential diagnosis to keep in mind, since an early diagnosis and timely treatment can improve the outcome of an otherwise poor prognosis disease.

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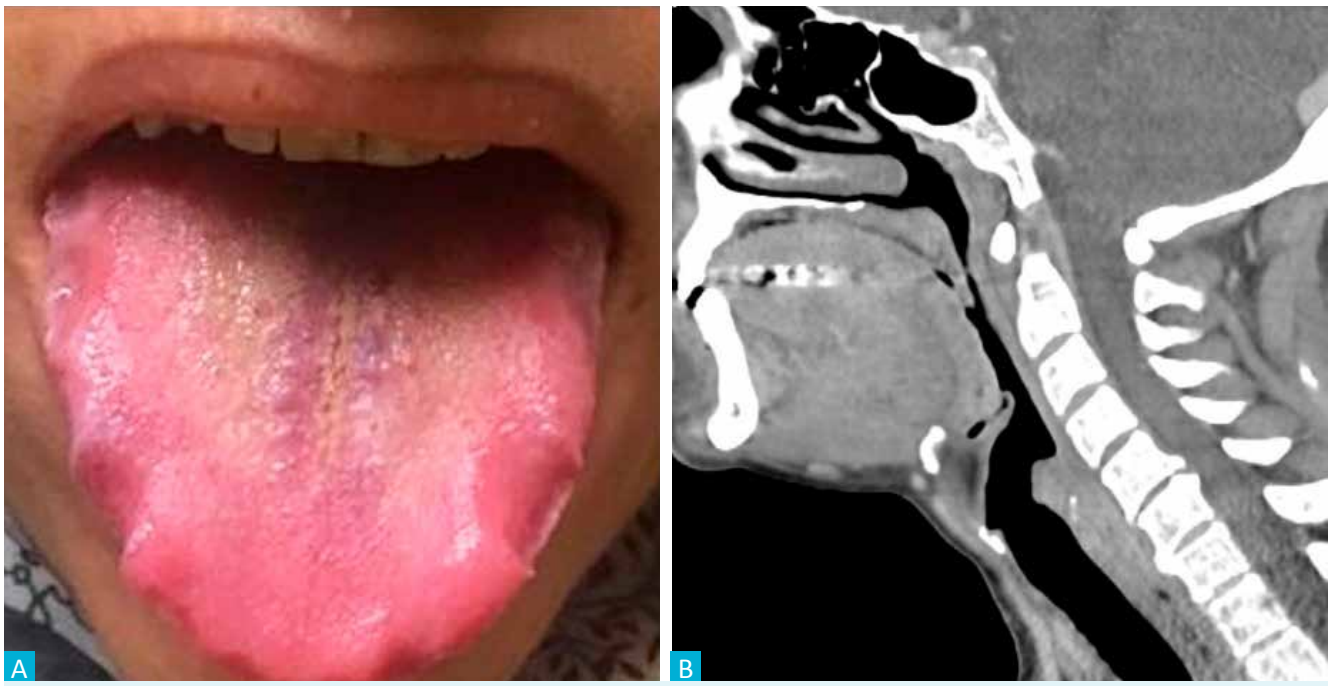

FIGURE 1. **(A)** Macroglossia with indentations from teeth; **(B)** Macroglossia on the head and neck CT, sagittal view.

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