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. 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.

An uncommon clinical finding like macroglossia can be present in tuberculosis, acromegaly, hypothyroidism and genetic syndromes, among others. 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.
AUTHORS’ CONTRIBUTION/CONTRIBUIÇÃO AUTORAL

CA: Elaboration of the manuscript
NM, LR and AC: Review of the manuscript

ETHICAL DISCLOSURES

CONFLICTS OF INTEREST: The authors have no conflicts of interest to declare.

FINANCING SUPPORT: This work has not received any contribution, grant or scholarship.

CONFIDENTIALITY OF DATA: The authors declare that they have followed the protocols of their work center on the publication of data from patients.

PATIENT CONSENT: Consent for publication was obtained.

PROVENANCE AND PEER REVIEW: Not commissioned; externally peer reviewed.

REFERENCES