Un signo de Amiloidosis

A sign of Amyloidosis

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ABSTRACT

The amyloidoses are a rare group of diseases that result from extracellular deposition of amyloid, a fibrillar material derived from various precursor proteins that self-assemble with highly ordered abnormal cross ß-sheet conformation. The authors present the case of a 76-year-old woman with anemia and worsening renal function, who develops intermittent nodular lesions on the tongue and hemorrhagic lesions on the oral mucosa. The etiological study allowed the diagnosis of light chain amyloidosis.

Keyword: macroglossia; monoclonal gammopathy; oral amyloidosis; pancytopenia; renal disease; Systemic amyloidosis.

RESUMEN

Las amiloidosis son un grupo poco común de enfermedades que resultan de la deposición extracelular de amiloide, un material fibrilar derivado de varias proteínas precursoras que se autoensamblan con una conformación de hoja ß cruzada anormal altamente ordenada. Los autores presentan el caso de una mujer de 76 años con anemia y deterioro de la función renal, que desarrolla lesiones nodulares intermitentes en la lengua y lesiones hemorrágicas en la mucosa oral. El estudio etiológico permitió el diagnóstico de amiloidosis de cadenas ligeras.

Palabras-clave: macroglosia; gammapatía monoclonal; amiloidosis oral; pancitopenia; enfermedad renal; amiloidosis sistémica.

CASE REPORT

A 76-year-old woman with a history of arterial hypertension, normochromic/normocytic anemia, and chronic kidney disease of unclear etiology. Admitted by asthenia, anorexia, and nausea. Blood workup revealed pancytopenia (hemoglobin 6.8 g/dL, leukocytes 3400/uL and platelets 34000/uL) and worsening renal function (creatinine 5.0 mg/dL). Additional tests revealed the low dosage of immunoglobulins (IgG 717, IgA 46 and IgM 29 mg/dL), proteinuria (3932 mg/24h) with the presence of urinary monoclonal lambda light chains. The bone marrow study showed the presence of monoclonal gammopathy of undetermined significance. Abdominal fat biopsy was performed, and the result was negative for the presence of amyloid substance. About 1 month after the onset of symptoms, the patient developed nodular lesions on the tongue (Figure 1 and 2 – arrows), with slight macroglossia and hemorrhagic lesions on the oral mucosa (Figure 2 – asterisk). The biopsy of the oral mucosa revealed positivity for Congo red, compatible with the presence of amyloid substance (Figure 3). This made the diagnosis of lambda light chain amyloidosis with renal, cardiac, gastrointestinal, soft tissue and autonomic involvement. This condition results from the deposition of monoclonal free light chains systemically due to monoclonal gammopathy, multiple myeloma, or, more rarely, B-cell lymphoma.1 It is a rare disease and owing to low specificity, prodromes are frequently misinterpreted and the diagnosis is often delayed.[1,2] Oral amyloidosis manifests with macroglossia or as yellowish-white nodules, widespread purple bulla-like masses, ecchymosis, papules, and ulcers, which frequently involves the mouth floor, gingival, and buccal mucosa.[4]

Figure 1

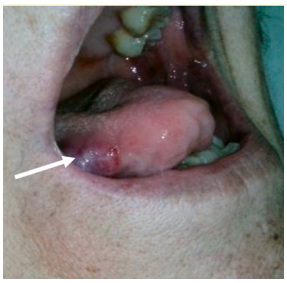


Figure 2

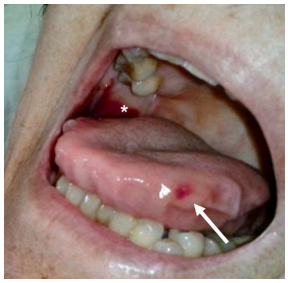
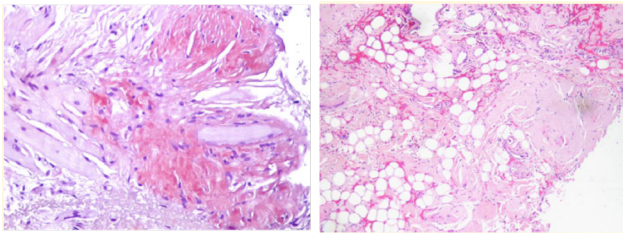


Figure 3



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Cómo citar este artículo: Figueiredo Albuquerque AL, Palavras MJ
A sign of Amyloidosis. Galicia Clin 2022; 83-2: 65-65
Recibido: 14/10/2021 ; Aceptado: 02/11/2021 // https://doi.org/10.22546/67/2683