Letters to the Editor

Exuberant macroglossia in a patient with primary systemic amyloidosis

Sirs,

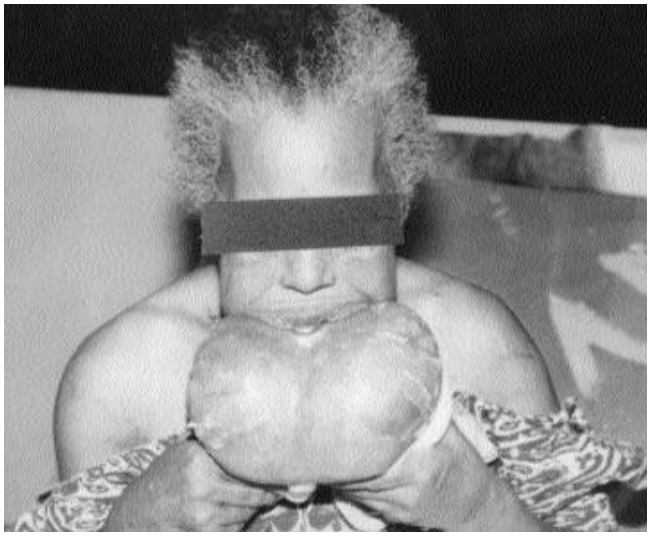
A 60-year-old woman was admitted to our hospital in November 1999 because of weakness and paresthesia in the right hand that had begun 2 years earlier. In 1998, she noted periorbital purpura and progressive enlargement of the tongue. There was no history of chronic inflamatory diseases such as rheumatoid arthritis, osteomyelitis, malaria or tuberculosis, as well as no history of amyloidosis.

On physical examination, there was bilateral periorbital purpura and submandibular swelling; extreme macroglossia that precluded closing the mouth and visualization of the throat was observed (Fig. 1). The Tinel sign was positive in the right superior limb. Cardiac and abdominal examination were normal. Selected laboratory investigations revealed a hemoglobin concentration of 10.5 g/dLand a normal leukocyte count; total serum protein and albumin were 6.9 g/dLand 4.3 g/dL, respectively. Renal functional impairment was not present and urinalysis was normal. Serum monoclonal paraprotein was detected by immunoelectrophoresis. X-ray studies of the chest and peripheral skeleton were normal. The electrocardiogram showed low voltage. The echocardiographic study was normal. Electromyography demonstrated prominent motor axonal neuropathy in the right superior limb. Tongue biopsy detected the presence of amyloid, and a bone marrow biopsy showed a normal number of plasma cells (4%). After the diagnosis surgery was performed, but the patient died of massive pulmonary embolism on the fifth post-operative day.

Systemic amyloidosis is a rare disorder that usually begins after the age of 40 and is associated with rapid progression, multisystemic involvement and short survival. The age-adjusted incidence of AL amyloidosis is estimated to be 5.1 to 12.8 per million person-years (1). In this type of amyloidosis, the deposits consist of monoclonal light chains produced by an indolent plasma cell dyscrasia in the bone marrow.The initial symptoms are frequently fatigue and weight loss, but the diagnosis is rarely made until symptoms or signs involving a particular organ appear (2). The median survival in a group of Mayo Clinic was 20 months, with a 5-year-survival of 20% (3).

As is often the case, in this patient an early diagnosis was not made because the initial complaints were unspecific. In the late stages of the disease, she developed periorbital purpura (racoon sign) and macroglossia; oral and skin involvement are important clues in diagnosing systemic amyloidosis. In the oral cavity, amyloid deposition may exhibit many forms. The best known oral finding is macroglossia, often associated with submandibular swelling (4). Periorbital purpura has been noted as the most characteristic form of cutaneous involvement in systemic amyloidosis (5).

Fig. 1. Patient suffering from exuberant macroglossia.



Autonomic and sensory neuropathy and carpal tunnel syndrome are relatively common features. The kidneys and heart – the organs most commonly involved in AL amiloidosis (6) – were not clinically affected in this case. In 90% of patients with AL amyloidosis, serum or urinary monoclonal immunoglobulins or light chains are detected by immunofixation electrophoresis. In most a clonal dominance of plasma cells is identified in bone marrow biopsy specimens (6). In this case, the monoclonal paraprotein detected by immunoelectrophoresis and a positive biopsy of the tongue established the diagnostic of ALamyloidosis, but the bone marrow biopsy did not reveal a plasma cell dyscrasia.

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Pulmonary granuloma, polyarthritis and antiphospholipids in common variable immunodeficiency: resolution after IVIG and the role of immunoglobulin A

Sirs,

Common variable immunodeficiency (CVI) is a heterogeneous immunodeficiency syndrome characterized by defective production of one or several immunoglobulin (Ig) isotypes, recurrent bacterial infections and an increased risk of autoimmune diseases, malignancies and granulomatous lesions (1, 2). The treatment of autoimmune diseases and granulomatous lesions is usually based on the steroids, but few report have shown the benefit of intravenous immunoglobulin (IVIG) (3-5). We report a case of CVI that seems original due to several points: its association antiphopholipid syndrome (APS), a favorable outcome of the granulomatous lesion with IVIG and a differential outcome according to the IVIG combination used.

A 28-year-old woman was referred to our center in October 2000 for relapse of a chronic polyarthritis. She had a medical history of recurrent sinusitis and pneumonia and in 1991 of post-infectious glomerulonephritis. Hypogammaglobulinemia was noticed at that time (4.6 g/L). In 1994 she was hospitalized for pulmonary embolism and iliac venous thrombosis revealing antiphospholipids [IgG 2 IU/ml; IgM 26 IU/ml (normal <20)], anticardiolipids (28 IU/ml; nor-