Case for diagnosis. Systemic light chain amyloidosis with cutaneous involvement*

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CASE REPORT

A 33-year-old female patient, previously healthy, presented with a four-month history of dyspnea on exertion, which evolved into orthopnea and dry cough. Concomitantly with the worsening of her clinical conditions, she developed periorbital, skin-colored infiltrated papules along with petechiae and ecchymoses (Figures 1, 2 and 3). A biopsy from the eyelid showed deposition of eosinophilic and amorphous material in the papillary dermis (Figure 4). Laboratory tests revealed mild elevations in GGT and ALP levels; an increased alpha-1 acid glycoprotein on serum protein electrophoresis; ANA and rheumatoid factor tests were negative. A myocardial MRI revealed a small pericardial effusion, mild bilateral pleural effusion and increased myocardial thickness, suggesting a restrictive and infiltrative heart disease. The patient went on hemodynamic instability with severe heart failure and decreased renal function unresponsive to treatment. She died shortly after the diagnosis.

Figure 1: Infiltrated, skin-colored papules on the eyelids

Figure 2: Close up of the previous figure showing petechiae

Figure 3: Evolution into ecchymoses

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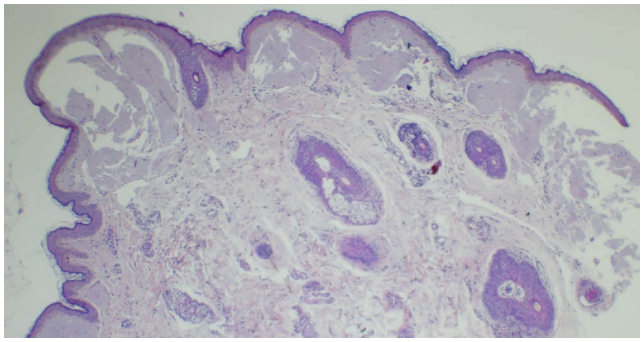

Figure 4: Deposit of eosinophilic and amorphous material in the papillary dermis (Hematoxylin and eosin, X40)

DISCUSSION

The amyloidoses comprise a group of rare diseases of unknown etiology in which defective proteins are deposited in the skin and other organs. They can be classified into systemic (generalized) or restricted (localized) to an organ, and subdivided according to the type of the deposited amyloid protein.[1] Systemic light chain amyloidosis (AL), also called primary systemic amyloidosis, has an estimated incidence of 10 people/million/year.[2]

Cutaneous manifestations are multiple, with a preference for skin folds, eyelids, retroauricular region, neck, axilla, umbilical and anogenital area.[3] Petechiae and ecchymoses are caused by the deposit of the protein in blood vessels. Bilateral eyelid ecchymosis, also known as raccoon eyes or raccoon sign, is a highly characteristic sign of amyloidosis, but can also be observed in cases of skull base fracture.[4] Protein infiltration into the dermis leads to the formation of yellowish nodules with a greasy surface. Macroglossia and periorbital purpura are described in about 15% of cases.[2] Carpal tunnel syndrome, alopecia, and sensitivity changes may also be found.

Biopsy is essential for the diagnosis and should be performed, whenever possible, in the affected organ. The aspiration of abdominal subcutaneous fat or bone marrow can be performed. Congo red staining is the gold standard for diagnosis,[5] demonstrating amyloid fibrils with apple-green birefringence. Systemic involvement, specific organ function, as well as cell dyscrasias or lymphomas should be investigated.[2,5,6]

There is no standard treatment for the condition. Treatment centers on suppressing the anomalous protein production and providing life support. Prognosis is reserved and cardiac involvement implies in worse evolution. Chemotherapy and transplantation of the affected organ, melphalan, and autologous stem cell transplantation have all been used with variable results.[1,2]

The unspecific clinical picture, the high mortality rate, difficulty in biopsying an internal organ, and the heterogeneous skin lesions highlight the role of a trained dermatologist and the importance of a skin biopsy for the accurate diagnosis of this entity. ❑

Abstract: Systemic light chain amiloydosis is a rare disease. Due to its typical cutaneous lesions, dermatologists play an essential role in its diagnosis. Clinical manifestations vary according to the affected organ and are often unspecific. Definitive diagnosis is achieved through biopsy. We report a patient with palpebral amyloidosis, typical bilateral ecchymoses and cardiac involvement, without plasma cell dyscrasia or lymphomas. The patient died shortly after the diagnosis.

Keywords: Amyloidosis; Amyloid; Plaque, amyloid; Ecchymosis; Purpura

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