Skin Rash, Dyspnea, Weight Loss and Leg Edema in a 59 Year Old Woman With Amyloidosis

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A 59 year old woman presented with a 3 year history of progressive nail atrophy and rash involving the hands, upper chest and face. One year prior to admission a skin biopsy performed elsewhere did not show any diagnostic findings. In the last 6 months she lost 10 kg in weight and noted bilateral leg edema. One month prior to admission she began to suffer from shortness of breath.

Physical examination revealed decreased breath sounds to the left lung with dullness to percussion, pitting leg edema, diffuse nails atrophy and blue vascular lesions on sun-exposed areas of skin [Figure 1]. Laboratory tests showed hypoalbuminemia (3.1 g/dl), proteinuria (500 mg/24 hr) and slightly elevated troponin TnI (0.5). Other laboratory test results were within normal limits and included complement levels, antinuclear factor, anti-DNA, rheumatoid factor, circulating anticoagulant, antineutrophil cytoplasmic antibodies, anticardiolipin, vitamin C levels, serology for human immunodeficiency virus, porphyrins in urine, and tumor markers. Chest X-ray demonstrated left pleural effusion. Thoracenthesis was performed and the pleural fluid was transudate. Cytology examination did not reveal malignant cells, and cultures were sterile. Further workup included gastroscopy and colonoscopy that showed mucosal petechiae and blood clots. Echocardiogram demonstrated moderate hypertrophy of the left ventricle with diastolic dysfunction. No signs of left ventricular hypertrophy were noted on electrocardiogram. Cardiac magnetic resonance imaging showed moderate left ventricular hypertrophy – a finding similar to the echocardiography. A tentative diagnosis of amyloidosis with cardiac involvement was suggested. To confirm the diagnosis a skin biopsy from a skin lesion was taken. Histological results were consistent with amyloidosis showing a typical picture of amyloid deposits in congo red stain [Figure 2].

Modern amyloid classification uses major fibril protein types [1]. The two most common forms of systemic amyloidosis are light-chain amyloidosis and reactive AA amyloidosis due to chronic inflammatory diseases [2]. Amyloidosis can involve various systems including the cardiovascular system, kidneys, nervous system, skin and lungs. In cardiac amyloidosis the most common presentations are congestive heart failure and conduction abnormalities [3]. An elevated troponin level is also one of the manifestations of cardiac amyloidosis and reflects the cardiac damage [4]. Patients with cardiac manifestations of amyloidosis have a poor prognosis with average survival of 6 months without therapy. Early diagnosis and treatment can improve the outcome [5]. Skin lesions can be the first

Figure 1. [A] Lesions consistent with petechiae and echymoses on upper chest skin. **[B]** Nail atrophy and similar lesions on hands

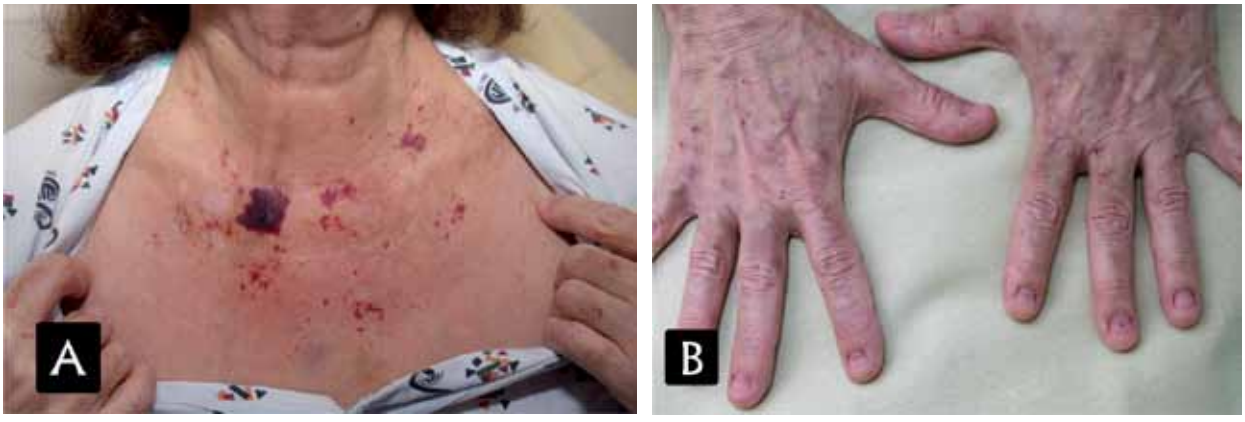
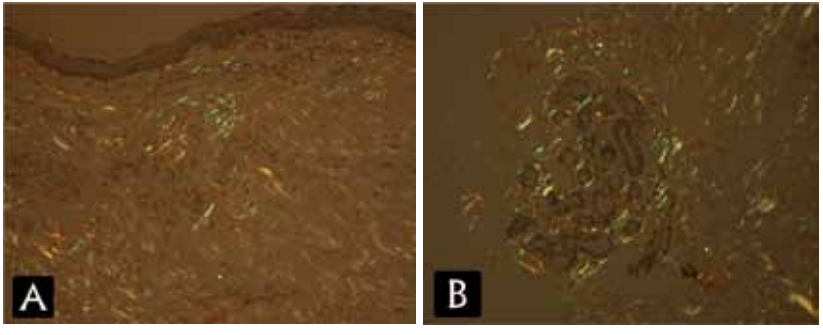


Figure 2. [A, B] Congo red stain demonstrates periadnexal , perineural and intravascular deposits of amyloid showing marked apple green birefringence staining on polarization microscopy



manifestation of systemic amyloidosis. Cutaneous manifestations include crusted nodules on the face and extremities as well as purpura and echymoses. As in the above presented patient, skin rash can be a manifestation of a disease that causes also congestive heart failure.

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