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A Rare Case of Bilateral Conjunctival Amyloidosis in Association with Systemic Involvement

Lina El Mekkoudi*, Salma Janati

Department of Internal Medicine, International Hospital Cheikh Zaid, Abulcasis University, Rabat, Morocco

***Corresponding author:** Lina El Mekkoudi
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Abstract | **Original Research Article**

Conjunctival amyloidosis is a very rare condition, mostly occurring as a local deposition of amyloid and very rarely in association with systemic involvement. Our purpose is to present a rare case of bilateral conjunctival amyloidosis with systemic involvement. A 58-year-old gentleman presented with bilateral eyelid ecchymosis and chemosis for past 2 years. Ocular examination found in both eyes a yellow-pink diffuse mass extending from the inferior bulbar conjunctiva to the lower fornix. Examination of the fundus revealed a single retinal cotton-wool spot. CT scan showed hypertrophy and infiltration of the orbital fat. Biopsies of conjunctival mucosa, bone marrow, perirenal fat, stomach, duodenal, and colic mucosa showed amyloid deposits. Immunofixation of blood samples revealed monoclonal gammopathy. Thus, patients with conjunctival amyloidosis should be examined comprehensively and regularly for systemic amyloidosis because of its poor life prognosis.

Keywords: Conjunctival amyloidosis, Systemic amyloidosis.

INTRODUCTION

Amyloidosis is a heterogeneous group of diseases characterized by extracellular amyloid deposits in different organs [1], it is associated with disturbance of organ function and causes a wide variety of clinical syndromes that are classified according to the respective fibril protein precursor [2]. The gold standard of amyloid detection is the demonstration of apple-green birefringence on congo red staining [3].

Classification has changed from primary and secondary amyloidosis to light chain amyloidosis and amyloid A protein amyloidosis [4]. Conjunctival amyloidosis is a rare manifestation often not clinically suspected [4, 5]. In à review of 2,455 conjunctival lesions submitted to a pathology laboratory, only five patients (0.002%) were found to have conjunctival amyloidosis [6]. In addition, amyloidosis of the conjunctiva is often localised with no other associations [5, 7]. We present a rare case of bilateral conjunctival amyloidosis with systemic involvement.

CASE REPORT

A 58-year-old gentleman with a history of cigarette smoking, presented with bilateral eyelid ecchymosis increasing spontaneously or with the effort, and chemosis that have a salmon-colored for past 2 years. He also reported Pinch hematoma at the neck and upper limbs (figure 1) associated with generalized weakness since 1 year.

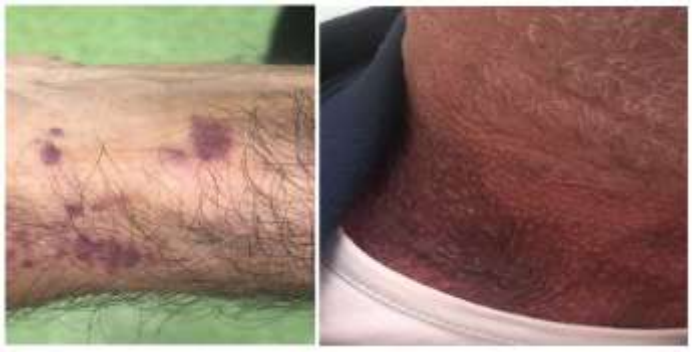

Fig-1: Pinch hematoma at the neck and upper limbs

There was no history suggestive of eye infections, trauma, chronic systemic illness, or familial involvement. Ocular examination found in both eyes a yellow-pink diffuse mass extending from the inferior bulbar conjunctiva to the lower fornix with areas of waxy-yellow condensations (figure 2).

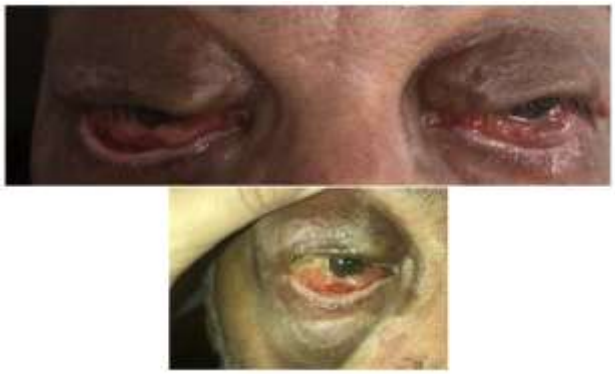

Fig-2: External photos showing bilateral yellow-pink conjunctival mass in the lower fornices

Slit-lamp examinations showed that the anterior chamber and the lens were clear in both eyes, intraocular pressure was normal. Examination of the fundus revealed a single retinal cotton-wool spot without retinal hemorrhage. CT scan showed hypertrophy and infiltration of the orbital fat.

Physical examination of the abdomen was unremarkable, neurologic examination reveals no focal deficits. As a conjonctival amyloidosis was suspected, a conjunctival biopsy was performed and the results were negative for Congo red staining.

Biopsies of conjunctival mucosa, perirenal fat, stomach, duodenal, and colic mucosa were redone, Histopathological examinations of these biopsies showed amyloid deposits around adipocytes ans focally in the wall of small blood vessels, congo-red positive material showed apple-green birefringence and dichroism with polarization microsopy. Minor salivary glands were uninvolved. Bone marrow biopsy also contain amyloid deposits with a clonal population of plasma cells monotypic kappa light chain (< 10%), multiple myeloma was ruled out. Del 17p was detected by FISH in bone marrow.

A complete systemic evaluation was carried out, immunofixation electrophoresis detected M-protein peak (IgG kappa) in a focal region of the gammaglobulin zone (9.7 g/l), the difference between involved and uninvolved circulating free light chain (dFLC) was 40 mg/l, complete blood count examination was normal, 24 hour urine examination was negative of albumin, urinary Bence Jones protein was absent, liver and kidney function tests were normal, factor X was within the normal range. Electromyography showed Axonal sensorimotor neuropathy affecting the lower limbs. On cardiovascular investigations, echocardiogram was normal, magnetic resonance imaging was refused by the patient, relevant blood tests revealed elevated NT Pro BNP (659 pg/ml) with normal cardiac troponin (0.033 ng/ml).

A diagnosis of systemic amyloidosis was made (Mayo Clinic stage II). Our patient was treated with chemotherapy according to VCD protocol (Velcade, Cyclophosphamide, Dexamethasone), the re-evaluation after 3 cycles has demonstrated a partial hematological response (dFLC: 6.35 mg/l), but the worsening of cardiac amyloidosis (NT Pro BNP : 1125 pg/ml, cardiac troponin : 0.06 ng/ml, Mayo Clinic stage III).

DISCUSSION

Conjunctival amyloidosis is very rare. An earlier pathological study of 2,455 cases of conjunctival lesions showed that conjunctival amyloidosis was diagnosed in only 5 patients (0.002%) [6]. Clinical presentation of conjunctival amyloidosis is extremely variable, leading to other presumption diagnoses such as inflammation or benign and malignant tumors of the conjunctiva [8, 9].

Conjunctival amyloidosis, predominantly affecting middle-aged adults, usually begins at the fornices, spreading to the bulbar and palpebral conjunctiva [8]. In our patient, 58-year-old, a bilateral diffuse pink-yellow infiltration was found extending from the inferior bulbar conjunctiva to the lower fornix. Associated spontaneous conjunctival hemorrhages are a common finding that may be explained by the fact that amyloid infiltration of vessel walls induces rigidity and disruption of conjunctival vessels [8].

In past studies, most of the conjunctival amyloidosis was found to be localized amyloidosis [10, 11]. Thus, conjunctival amyloidosis accompanied by systemic amyloidosis is extremely rare. However, systemic amyloidosis has to be excluded because it is a life-threatening disease, and a complete systemic evaluation should be performed.

Furthermore, testing for monoclonal protein population should be determined by serum and urine protein electrophoresis and immunofixation [12].

In our patient, systemic investigations are performed to rule out systemic disease, a tissue biopsy of conjunctival mucosa was taken to confirm the diagnosis, the sampling was also performed in perirenal fat, stomach, duodenal, and colic mucosa which confirmed the diagnosis of systemic amyloidosis. Immunohistochemical studies of bone marrow biopsies showed an increased population of plasma cells with the amyloidogenic over-expression of the subunit kappa. In addition, immunofixation of blood samples revealed monoclonal gammopathy.

CONCLUSION

Although the vast majority of patients with primary conjunctival deposits have no positive systemic findings, a systemic involvement should always be ruled out because of it is a life-threatening disease.

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