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## Case Report

# Retinitis pigmentosa associated with systemic light chain amyloidosis (AL amyloidosis)

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## ABSTRACT

Retinitis pigmentosa (RP) or hereditary retinal dystrophy is a rare disease that can be isolated (non-syndromic RP) or associated with other systemic signs (syndromic RP). Kidney damage is exceptionally reported in patients with RP, particularly in syndromic forms. Association with renal amyloidosis remains unusual with only one reported case of RP and hereditary gelsolin amyloidosis due to a G654A gelsolin mutation defining the new syndrome of Ardalan-Shoja-Kiuru. Apart from this publication, no case associating RP and AL amyloidosis has been found.

We report an original case of renal damage revealing kappa-type systemic light chains amyloidosis (AL amyloidosis) in 35-year-old man with sporadic RP. Our observation is, to our knowledge, the first to report this association.

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## 1. INTRODUCTION

Described for the first time as a clinico-pathological entity in 1853, and named "retinitis pigmentosa" (RP) in 1857 [1], the hereditary retinal dystrophy is a heterogeneous group of disorders having in common the genetic origin and the progressive loss of eye photoreceptor and epithelial pigment function [2, 3]. It is a rare disease with an overall prevalence in the general population estimated at 1/3000 to 1/5000 [1-3]. It therefore represents the most frequent hereditary dystrophy of the retina [3] and one of the most

common causes of visual impairment all ages combined [1, 2].

RP can be isolated (non-syndromic RP) [4] or associated with other systemic signs (syndromic RP) [2, 3, 5]. In the majority of cases this disease is inherited with more than 90 causal mutations and multiple patterns of inheritance: autosomal recessive, autosomal dominant and X-linked recessive [1-5]. These mutations can be identified in 30-80% of cases; the remaining cases are called "sporadic" [6].

Kidney damage is exceptionally reported in patients with RP, particularly in syndromic forms [6-8]. Association

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with renal amyloidosis remains unusual [9].

We report an original case of renal damage revealing systemic light chain amyloidosis in 35-year-old man with RP.

## 2. CASE REPORT

A 35-year-old Tunisian man with a history of progressive bilateral vision loss from the age of 22, was investigated for generalized edema on progressive onset. The somatic examination noted a white, soft, and painless edema predominantly on the lower limbs, a visual acuity of 7/10 on the right and 6/10 on the left, without other abnormalities.

The basic biological tests concluded in a pure nephrotic syndrome: protein at 50g/l, albumin at 20g/l, and 24-hour proteinuria at 3.8g. The other laboratory tests were within normal limits: hemoglobin, leukocytes, platelets, creatinine, calcemia, plasma ionogram, transaminases, muscle enzymes, thyroid hormones, blood sugar, erythrocyte sedimentation rate, C-reactive protein, and lipid parameters.

The specialized ophthalmologic examination concluded in bilateral retinitis pigmentosa, more severe on the left side.

The kidney biopsy showed renal amyloidosis and the typing was for a kappa AL amyloidosis.

Electrophoresis and immunoelectrophoresis of blood and urine proteins confirmed light chains kappa-type monoclonal gammopathy.

Sternal puncture, bone marrow biopsy, and bone marrow karyotype ruled out a possible underlying hematologic malignancy. Likewise tumor markers, immunological tests, and bacterial and viral serodiagnostics were negative eliminating possible cancers, connective tissue diseases, and progressive infections.

The biopsy of the accessory salivary glands also revealed the amyloid deposits confirming the systemic character of the amyloidosis. The electrocardiogram, trans-thoracic cardiac ultrasound, hepatic ultrasound, and thoraco-abdominal-pelvic computed tomography were without abnormalities.

Likewise, the ophthalmological examination as well as the kidney tests carried out on the parents and the siblings did not reveal any significant abnormalities.

Thus the diagnosis of sporadic RP associated with primary amyloidosis was retained. A treatment combining systemic corticosteroids and thalidomide was proposed but refused by the patient.

## 3. DISCUSSION

Retinitis pigmentosa, particularly in these syndromic forms, can be associated with several types of kidney disease defining rare genetic syndromes or diseases. The most common and best known of these syndromes/diseases are: Senior-Loken syndrome (RP and juvenile nephronophthisis), Alagille syndrome (RP and renal dysplasia), Bardet-Biedl syndrome (RP and renal hypoplasia, hydronephrosis, pyelonephritis, vesicourethral reflux, or glomerulonephritis), Alstrom syndrome (RP and progressive renal failure), Sensenbrenner syndrome (RP and interstitial nephritis), and Refsum's Disease (RP and tubular dysfunction or aminoaciduria) [7, 8, 10-13].

Immunoglobulin light chain amyloidosis (AL amyloidosis), formerly called primary amyloidosis, is the most common of systemic amyloidosis [14]. Renal involvement is very frequent during this type of amyloidosis and represents a major cause of death [14, 15].

The association of RP and renal amyloidosis has only been reported once in the world literature in 2007 [16]. It was a 25-year-old Iranian woman diagnosed with RP and hereditary gelsolin amyloidosis due to a G654A gelsolin mutation [16]. This association assumed a direct genetic link between these two disorders and defined a new syndrome: the Ardalan-Shoja-Kiuru syndrome [9, 16].

Apart from this publication, no case associating RP and AL amyloidosis has been found.

## 4. CONCLUSIONS

Our observation is, to our knowledge, the first to report the association of AL amyloidosis with retinitis pigmentosa. Further large-scale studies are needed to detect a possible pathogenesis link.

Investigation of renal function (creatinine and 24-hour proteinuria) is desirable in any patient with RP. Likewise, a specialized ophthalmologic examination seems useful in patient diagnosed with AL amyloidosis.

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