Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Nephrology

AA Amyloidosis Secondary to Gouty Arthritis in a Patient with Autosomal Dominant Polycystic Kidney Disease: A Case Report

El Aboudy Imane[1*], El Farouki Mohammed Reda[1], Naji Jamal-Eddine[1], Sina Mohammed[2], El hassani Mohammed[1]

[1]Nephrology Department, Military Hospital Moulay Ismail, Meknes, Morocco
[2]Pathology laboratory, Military Hospital Moulay Ismail, Meknes, Morocco

DOI: https://doi.org/10.36347/sjmcr.2025.v13i04.033 | **Received:** 13.02.2025 | **Accepted:** 18.03.2025 | **Published:** 26.04.2025

***Corresponding author:** El Aboudy Imane
Nephrology Department, Military Hospital Moulay Ismail, Meknes, Morocco

Abstract **Case Report**

AA amyloidosis is a rare complication of chronic inflammation. We present an unusual case of AA amyloidosis secondary to chronic gouty arthritis in a patient with autosomal dominant polycystic kidney disease (ADPKD). A 59-year-old man with a family history of ADPKD was referred for evaluation of massive proteinuria. Evaluation revealed AA amyloidosis, confirmed by accessory salivary gland biopsy, associated with chronic gouty arthritis and ADPKD. This case highlights the importance of investigating potential causes of amyloidosis in patients with ADPKD presenting with proteinuria, even in the presence of concomitant inflammatory conditions such as gout.

Keywords autosomal dominant polycystic kidney disease, AA amyloidosis, Gout, Proteinuria.

INTRODUCTION

Autosomal dominant polycystic kidney disease (ADPKD) is a hereditary disease caused by sequence variations in the *PKD1* or *PKD2* genes, in approximately 93% of cases, and characterized by the progressive development and growth of bilateral renal cysts [1].

Proteinuria is generally less than 1 g/day in ADPKD carriers [2]. However, detectable proteinuria and microalbuminuria are correlated with an increased total cyst volume and more severe kidney disease [1]. Massive or nephrotic proteinuria is not a common feature of ADPKD [3].

ADPKD is frequently associated with hyperuricemia and/or gout. This link has recently been established by the demonstration of epistatic interactions (interaction between genes where the phenotypic expression of one gene masks or interferes with the expression of a different gene or genes) between the expression of the *PKD2* gene, which is located near the *ABCG2* gene. The latter has been shown to be the most important variant in the genetic control of serum uric acid [4].

Although associations between ADPKD, gout, and AA amyloidosis have been described individually, the coexistence of these three conditions remains rare. Here, we report an unusual case of AA amyloidosis secondary to chronic gouty arthritis in a patient with ADPKD, diagnosed incidentally during the evaluation of proteinuria.

CASE PRESENTATION

A 59-year-old patient with no significant past medical history other than chronic smoking, which he quit 4 years prior, initially consulted a rheumatologist for inflammatory-type knee and heel pain evolving for more than two years, treated by self-medication with non-steroidal anti-inflammatory drugs. He was started on colchicine and febuxostat for suspected gouty arthritis. Joint ultrasound revealed an anechogenic effusion speckled with punctate hyperechogenic deposits, associated with a double contour sign, synovitis, and a positive Doppler signal. Polarized light microscopy of the fluid from the knee joint aspiration revealed monosodium urate microcrystals.

The patient was then referred to our department for investigation of massive proteinuria discovered during routine testing. His family history revealed ADPKD in his mother. He had no history of tuberculosis, pulmonary diseases, inflammatory bowel diseases, familial Mediterranean fever (FMF), or rheumatic diseases. He had no signs of macroscopic hematuria, urinary tract infection, rash, fever, or oral ulcers.

Physical examination revealed a normotensive (130/90 mmHg), afebrile patient with a swollen right knee and edematous feet. The rest of the physical examination was unremarkable.

Laboratory investigations revealed:

- Blood urea nitrogen: 0.62 g/L (N: 0.21-0.43 g/L)
- Serum creatinine: 26 mg/L (N: 5-12 mg/L), eGFR CKD-EPI 28 ml/min/1.73m2
- Serum uric acid: 71 mg/L (N: 23-61 mg/L)
- Proteinuria: 6 g/24h
- Serum total protein: 67 g/L (N: 64-83 g/L)
- Serum albumin: 39 g/L (N: 34-45 g/L)
- Complement fractions C3 and C4: normal
- Immunological workup (antinuclear antibodies, anti-double-stranded DNA antibodies): negative
- Rheumatoid factor: normal
- Serum protein electrophoresis: normal. Serum protein immunofixation: normal
- Hepatitis B and C serologies: negative

The remaining laboratory tests were unremarkable.

Given the massive proteinuria and the family history of ADPKD, a renal ultrasound was performed, showing enlarged kidneys with multiple bilateral cysts, confirming the diagnosis of ADPKD.

Due to the persistence of proteinuria despite gout treatment and in the absence of other obvious etiologies, an accessory salivary gland biopsy was performed. Histological examination revealed the presence of amorphous deposits staining with Congo red (**Figure 1,2**), exhibiting apple-green birefringence under polarized light **(Figure 3**), consistent with amyloidosis. Immunohistochemical typing confirmed the presence of anti-SAA antibodies, suggesting secondary AA amyloidosis.

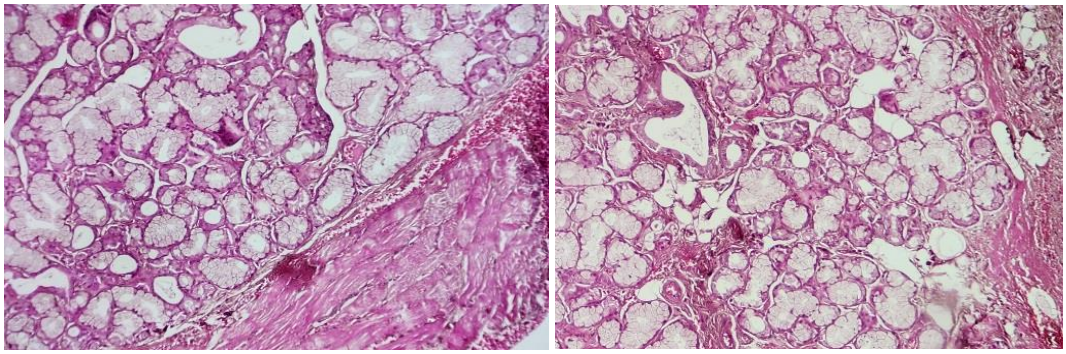

Figure 1,2: Accessory salivary gland biopsy showing amorphous deposits staining with Congo red

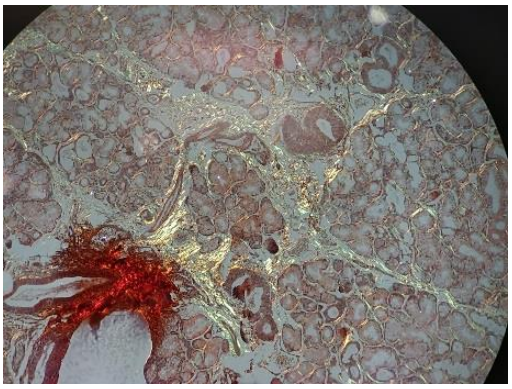

Figure 3: Accessory salivary gland biopsy showing apple-green birefringence of amorphous deposits under polarized light

Differential diagnosis:

Initial differential diagnoses included:

- Primary glomerulonephritis: This diagnosis was considered due to the massive proteinuria. However, the absence of hematuria, nephritic syndrome, and the presence of a family history of ADPKD made this diagnosis less likely. Furthermore, the chronic inflammatory context secondary to gout, and the histology ruled out this etiology.
- AL amyloidosis (light chain amyloidosis): The absence of monoclonal gammopathy on serum protein electrophoresis and immunofixation, as well as the positive immunohistochemical typing for SAA, made this diagnosis unlikely.
- Glomerulopathies associated with ADPKD: Patients with ADPKD may develop secondary

glomerulopathies, including focal segmental glomerulosclerosis [5]. However, these glomerulopathies typically do not exhibit amyloid deposits.

DISCUSSION

This case illustrates a rare association between ADPKD, gouty arthritis, and AA amyloidosis.

AA amyloidosis is a form of secondary amyloidosis resulting from the deposition of amyloid fibrils derived from serum amyloid A protein (SAA), an acute-phase protein produced in response to chronic inflammation [6]. The most common precursor diseases of AA amyloidosis, in the general population, are chronic inflammatory diseases such as rheumatoid arthritis, ankylosing spondylitis, and chronic infections [7].

Although gouty arthritis is among the 4 diseases strongly associated with AA amyloidosis [7], it remains an underestimated cause due to the small number of cases reported in the literature or lack of awareness of this association [8]. It should be noted that a strong association of a disease with AA amyloidosis does not necessarily indicate a frequent association in clinical practice (e.g., familial Mediterranean fever versus gout) [7]. In our case, chronic gouty arthritis, evolving for more than two years and insufficiently controlled, likely acted as the main inflammatory factor contributing to the development of AA amyloidosis.

ADPKD is among the unclear associations with AA amyloidosis but remains a potential candidate for a strong association [7]. Polycystic kidneys have an increased susceptibility to infections [1]. Amyloidosis can be seen during the disease due to recurrent cyst infections that stimulate a chronic inflammatory response [9-11].

Our patient had no history of chronic renal cyst infection or other chronic inflammatory diseases known to cause secondary amyloidosis, except for unmanaged gouty arthritis evolving for more than 2 years.

The diagnosis of AA amyloidosis secondary to gouty arthritis favored by ADPKD was then retained given the young age of the patient and the absence of other risk factors associated with gout.

CONCLUSION

This rare case of AA amyloidosis secondary to gouty arthritis in a patient with ADPKD highlights the need for increased vigilance and thorough evaluation in patients presenting with unusual combinations of conditions.

Massive proteinuria in any patient with ADPKD should prompt us to search for associated glomerular diseases, primary or secondary, given that there are underestimated associations of diseases such as AA amyloidosis that can be secondary to gouty arthritis.

A multidisciplinary approach is essential to optimize outcomes in these patients.

Conflicts of interest: The authors declare that they have no conflicts of interest related to this article.

Funding: No funding was received for the completion of this article.

REFERENCES

1. Bharathi V. Autosomal Dominant Polycystic Kidney Disease (ADPKD). Reddy, MD. Published January 1, 2023.
2. Simon HB, Thompson GJ. Congenital renal polycystic disease. JAMA. 1955;159:657.
3. Hiura T, Yamazaki H, Saeki T, et al. Nephrotic syndrome and IgA nephropathy in polycystic kidney disease. Clin Exp Nephrol. 2006;10:136-9.
4. Chalès G, Bardin T, Richette P. Hyperuricémies non goutteuses. EMC - Endocrinologie-Nutrition. 2022;33(3):1-13.
5. Chapman AB, Johnson AM, Gabow PA, Schrier RW. Overt proteinuria and microalbuminuria in autosomal dominant polycystic kidney disease. J Am Soc Nephrol. 1994;5:1349-54.
6. Papa R, Lachmann HJ. Secondary, AA, Amyloidosis. Rheum Dis Clin N Am 2018;44(4):585-03
7. Brunger AF, Wechalekar AD, Hawkins PN, Hazenberg BP, Hegenbart U, … Lachmann HJ. Causes of AA amyloidosis: a systematic review. Amyloid. 2020;27(1):1-12.
8. Gromova MA, Tsurko VV. Gout and AA-Amyloidosis: A Case-Based Review. Mediterr J Rheumatol. 2020;32(1):74-80.
9. Yenigun EC, Ertugrul DT, Ozturk MA, Ersoy FF, Turkmen K, Sever MS. Coexistence of Autosomal Dominant Polycystic Kidney Disease and Amyloidosis in a Patient with Nephrotic-range Proteinuria. Int J Nephrol. 2014;2014:243-5.
10. Kamimura H, Tsuchiya K, Honda K, et al. Secondary systemic amyloidosis associated with frequently infected hepatic cysts in a patient with autosomal dominant polycystic kidney disease. Clin Nephrol. 2003;59:485-8.
11. Tsuchiya Y, Ubara Y, Suwabe T, et al. AA-amyloidosis in Autosomal Dominant Polycystic Kidney Disease Caused by Chronic Cyst Infections Lasting for 30 years. Intern Med. 2013;52:791-4.