differential diagnosis, particularly if family or drug history is negative.

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Anti-Phospholipase A2 Receptor Antibody Testing to the Rescue

Abstract

Serological testing for M-type anti-phospholipase A2 receptor antibodies (antiPLA2R Ab) has abrogated the need for kidney biopsy to diagnose membranous nephropathy in the appropriate clinical setting. We report a case of a 63-year-old hypertensive male who presented with nephrotic syndrome associated with autosomal dominant polycystic kidney disease which was effectively diagnosed with the use of antiPLA2R Ab test. He achieved complete remission upon treatment with Rituximab. Hitherto, all anecdotal case reports of this uncommon association were diagnosed only by kidney biopsy. We highlight the usefulness of PLA2RAb especially in such cases of difficult-to-biopsy kidneys.

Keywords: *Membranous nephropathy, Nephrotic syndrome, PLA2R, Polycystic kidneys, Rituximab*

Introduction

Serological testing for M-type antiphospholipase A2 receptor antibodies (antiPLA2R Ab) has abrogated the need for kidney biopsy to diagnose membranous nephropathy in appropriate clinical settings.[1] The use of enzyme-linked immunosorbent assay testing for antiPLA2R Ab to diagnose and monitor response to treatment has been endorsed in the 2021 guideline by the Kidney Diseases Improving Global Outcomes.[2] We report a case of nephrotic syndrome associated with autosomal dominant polycystic kidney disease, which was effectively diagnosed and managed using an antiPLA2R Ab test.

Case Report

A 63-year-old man with a history of well-controlled hypertension for the past 5 years presented to the Nephrology services of our hospital with complaints of progressive pedal edema over 1 month. There were no other symptoms. On examination, the patient had bilateral pedal edema and blood pressure was 130/80 mmHg. The rest of the systemic examination was normal except for a ballotable kidney on the left side.

Investigations showed Hemoglobin of 14.2 g/dL, serum creatinine of 1.2 mg/dL, HbA1c 5.9%, fasting total

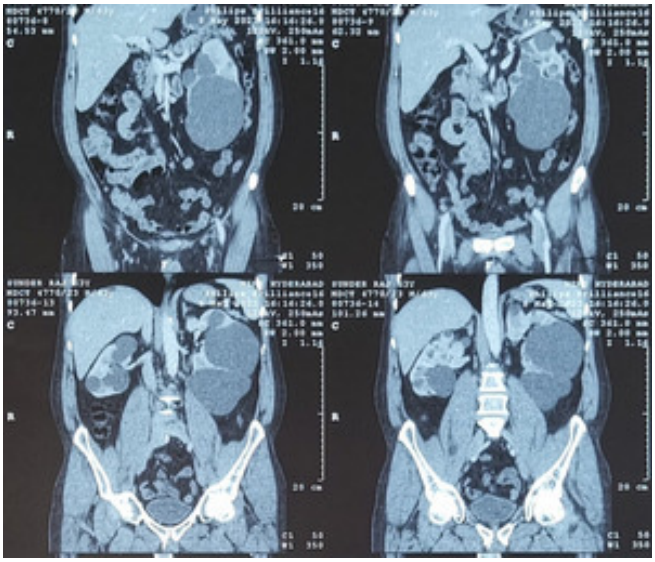

Figure 1: Contrast-enhanced computed tomography scan showing bilateral asymmetrical polycystic kidneys.

cholesterol of 362 mg/dl, total protein of 5.9 g/dL, and serum albumin of 2.7 g/dL. A urine examination showed 4+ albumin, nil RBC, and 1-2 pus cells. A spot urine protein-creatinine-ratio was 7.71 and 24-hour urinary proteins were 6,200 mg/day. Hepatitis B surface antigen, anti-Hepatitis C antibodies, and HIV antibodies were negative. Anti-nuclear factor antibodies were negative.

Ultrasonography revealed bilateral polycystic kidneys with a complex cyst in the lower pole of the left kidney. A contrast-enhanced computed tomography of the abdomen showed the right kidney at 15.0 × 7.7 cm and the left kidney at 23.4 × 10.6 cm. The lower pole of the left kidney showed a Bosniak IIF cyst of 12.2 × 13.0 × 13.0 cm [Figure 1]. There were few simple hepatic cysts. Other age-appropriate cancer screening (serum total PSA, chest X-Ray, stool occult blood testing was done; the patient was not willing for a colonoscopy) were also negative. An FDG-PET computed tomography scan was done which did not show any significant tracer uptake.

Serum PLA2R ab level was 78 IU/mL and a diagnosis of primary membranous nephropathy associated with Autosomal dominant polycystic kidney disease (ADPKD) was made. He had no known family history of ADPKD or renal failure and his immediate kin were advised to undergo screening ultrasound. His only daughter, 31 years old, was found to have bilateral kidney cysts and liver cysts.

He was treated with Rituximab at a dose of 1 g, 2 weeks apart, and followed up. At 3 months, his PLA2R was 23 IU/mL, serum albumin 3.2 g/dL, and spot urine PCR 2.8. At 6 months, the PLA2R was negative and serum albumin 3.7 g/dL and spot urine PCR 0.5—a complete remission was achieved [Table 1]. The patient is asymptomatic and is on angiotensin receptor blocker telmisartan for his hypertension and febuxostat for hyperuricemia and is on follow-up.

Table 1: Showing trend of investigation reports during the disease course

Parameter	At diagnosis	At 3 months	At 6 months
Serum PLA2R*	78 IU/mL	23 IU/mL	5 IU/mL
Proteinuria (UPCR)#	7.71	2.80	0.51
Serum Albumin	2.7 g/dL	3.2 g/dL	3.7 g/dL

*Antiphospholipase A2 Receptor antibody. #Urine Protein-Creatinine-Ratio

Discussion

Although anecdotal case reports of nephrotic syndrome in patients with ADPKD are documented,[3] with various histologies like focal segmental glomerulosclerosis, IgA nephropathy, diffuse proliferative glomerulonephritis, and membranous nephropathy,[4] all the cases had undergone an open/laparoscopic kidney biopsy and carried a tissue diagnosis. This is probably because all such case reports were reported prior to 2009—before the diagnosis of the target antigen in membranous nephropathy.[5]

To our knowledge, this is the first case of this uncommon association diagnosed and managed without the need for a kidney biopsy. We highlight the usefulness of PLA2RAb especially in such cases of difficult-to-biopsy kidneys so that the patient care can be delivered uncompromised without the need and risks of an invasive procedure.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent.

Conflicts of interest: There are no conflicts of interest.

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GALNT3 Mutation in Hyperphosphatemic Familial Tumoral Calcinosis – Novel Etiology of Secondary Amyloidosis

Abstract

Tumoral calcinosis is a rare syndrome characterized by calcium salt deposition in different periarticular soft tissue regions. We report this case of tumoral calcinosis with history of persistent soft tissue calcifications for over three decades. He presented with nephrotic syndrome and kidney biopsy revealed secondary amyloidosis. Genetic evaluation revealed GALNT3 mutation and diagnosis of hyperphosphatemic familial tumoral calcinosis was made. With this case report, we want to reiterate the need to consider tumoral calcinosis in secondary amyloidosis differentials and the pivotal role of genetic workup in chronic soft tissue calcifications.

Keywords: *Amyloidosis, Hyperphosphatemia, Mutation, Secondary amyloid, Soft tissue calcification, Tumoral calcinosis*

Introduction

Tumoral calcinosis (TC), a rare clinical syndrome characterized by periarticular soft tissue calcifications, presents a diagnostic challenge due to its varied manifestations.[1,2] We present a case of amyloidosis secondary to long-standing soft tissue calcification, eventually diagnosed to be hyperphosphatemic familial TC. To the best of our knowledge, this is the first reported biopsy-proven case of secondary amyloidosis presenting as nephrotic syndrome attributable to primary TC.

Case Report

A 39-year-old male, born out of a consanguineous marriage, first noticed a lump at his left hip at the age of 10 years. An excision biopsy revealed fibro-collagenous tissue with multiple calcific deposits with lymphoplasmacytic infiltrates, and he was diagnosed to have TC based on radiological and histopathological findings. No family history of similar illness was present. He was found to have hyperphosphatemia with normal serum calcium levels. On further evaluation, his 25-hydroxy vitamin D was found to be 44 nmol/L, intact parathormone (iPTH) 5 pg/mL, and FGF-23 13 RU/mL (reference: 0–150). His blood sugar and renal function were within normal range. He was started on acetazolamide, but his serum phosphorus levels remained high (ranging 5–7 mg/dL). For the next 30 years, he continued to develop soft-tissue calcification in different body areas. He subsequently underwent corrective surgeries and remained on a low-phosphate diet, acetazolamide, and aluminum hydroxide.

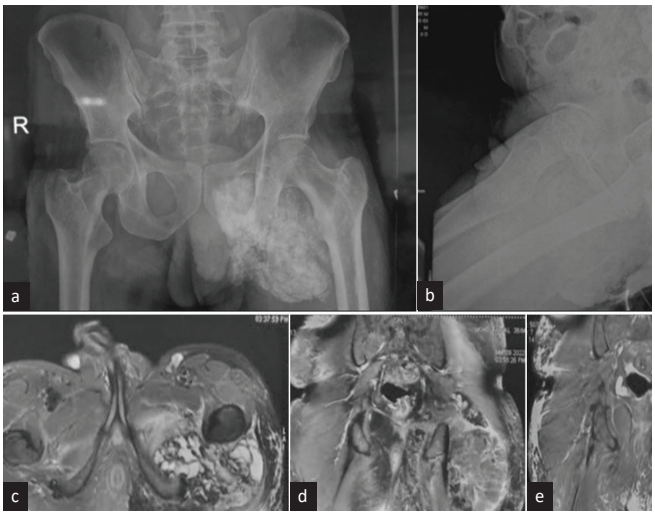

Figure 1: (a) X-ray pelvis anteroposterior and (b) lateral view depicting heterogenous fluffy opacity involving soft tissues of left side of inferior aspect of left hemi-pelvis and medial aspect of upper thigh, overlying ischial bones, extending up to left side hemi-scrotum. Magnetic resonance imaging (MRI) Pelvis with (c) contrast axial and (d) and (e) coronal view depicts large multilobulated septate cystic, partly solid soft tissue mass in proximal thigh & adjacent buttock surrounding posterior aspect of intertrochanteric and proximal shaft of femur. Septate cystic area was hypointense in T1, and hypo to hyperintense in T2 with multiple air fluid level showing dependent calcium. There was post-contrast patchy peripheral enhancement of the mass.

He was presented to our department with pedal and facial swelling for 1 week, along with frothuria. There