

AL AMYLOIDOSIS MASQUERADING AS OVARIAN MALIGNANCY: A CASE REPORT

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INTRODUCTION

Immunoglobulin light chain amyloidosis is a rare clonal plasma cell disorder in which misfolded immunoglobulin light chains deposit in organs such as the heart, kidneys, liver, gastrointestinal tract, and peripheral nerves, causing progressive multiorgan dysfunction.

Amyloidosis can mimic malignancy because its clinical and imaging features may appear tumor-like. Gynecologic involvement is rare but can affect the cervix, uterus, ovaries, fallopian tubes, or adnexa.

CA-125 is a well-established tumor marker for ovarian cancer, but it is not tumor-specific and may be elevated in several benign and inflammatory conditions. Systemic amyloidosis can lead to massive pleural effusions and ascites, resulting in secondary elevation of CA-125 and a clinical picture that may mimic advanced ovarian malignancy.

We report a case of 45-year-old female who presented with lower abdominal discomfort and ascites. Imaging showed bulky ovaries without a discrete mass and raised CA-125 level, raising strong suspicion of ovarian malignancy.

CASE SUMMARY

We present a case of a 45-year-old female from Punjab, Pakistan, whose medical history began approximately five months prior to her initial presentation at our institute, when she started experiencing persistent lower abdominal discomfort and abdominal distension.

She presented to our walk-in clinic on November 9, 2021, and a CT scan of the abdomen and pelvis revealed bulky ovaries with no evidence of discrete ovarian mass and small-volume abdominopelvic ascites (Figure 1). Her initial tumor markers were concerning, with a CA-125 level of 275 U/mL, leading to a strong suspicion of ovarian malignancy. Other tumor markers, including CEA of 4.96 ng/mL and AFP of 2.04 IU/mL were within normal range.

On November 18, 2021, an MRI of the pelvis confirmed a bulky left ovary but no evidence of a measurable mass or diffusion restriction. Simultaneously, a CT of the thorax identified minimal left pleural effusion. An ultrasound-guided ascitic tap was performed on November 23, 2021, which drained 500 mL of fluid; however, the cytology showed only a mixed inflammatory infiltrate with no evidence of malignancy. Her repeat CA-125 level rose to 1024 U/mL within 2 weeks of her initial presentation.

After a multidisciplinary team (MDT) discussion on December 1, 2021, her management was started on an ovarian malignancy protocol. A diagnostic laparoscopy and debulking surgery were planned. As part of the pre-op workup, she underwent upper and lower GI endoscopies on December 23, 2021, which revealed only small, benign rectal polyps.

On January 3, 2022, the anesthesia team noted generalized anasarca (edema), a mild pleural effusion, and a very low serum albumin of 1.54 g/dL. By January 4, 2022, nephrology evaluation identified nephrotic syndrome and chronic kidney disease (CKD) stage 3a, characterized by massive 4+ proteinuria, serum creatinine 1.59 mg/dL, and eGFR of 35. Due to these severely deranged parameters and the high risk of anesthesia, her planned debulking surgery was deferred.

Her albumin to creatinine ratio was 12.878 mg/mg, protein to creatinine ratio 21.23 mg/mg, ESR 103 mm/1st hour, CRP 13 mg/L. She had negative ANA, p-ANCA, c-ANCA, anti-ds-DNA antibodies, and anti-PLA2R. An ultrasound-guided renal biopsy was performed on January 12, 2022.

On January 18, 2022, laboratory results revealed a kappa to lambda ratio of 1.16 (1.17 - 2.93), and Serum Protein Electrophoresis (SPEP) revealed a monoclonal protein band (1.9 g/L) in the gamma region. The renal biopsy results soon followed, with the glomeruli showing marked mesangial expansion with eosinophilic acellular homogenous deposits (Congo Red stain positive, JMS negative, PAS weak positive) confirming the diagnosis of amyloidosis (Fig 2, Fig 3, Fig 4). Serum immunotyping detected an IgG-Lambda band.

On January 26, 2022, a final MDT review concluded that the patient's initial presentation, including the raised CA-125 (which had peaked at 1024 U/mL) and bulky ovaries, was likely secondary to systemic immunoglobulin light chain amyloidosis rather than primary ovarian cancer. Having confirmed the diagnosis of Immunoglobulin-related Amyloidosis (AL Amyloidosis) with renal involvement and nephrotic syndrome, the patient was informed on January 28, 2022, that she would need specialized hematologic treatment.

CASE SUMMARY (CONTINUED)

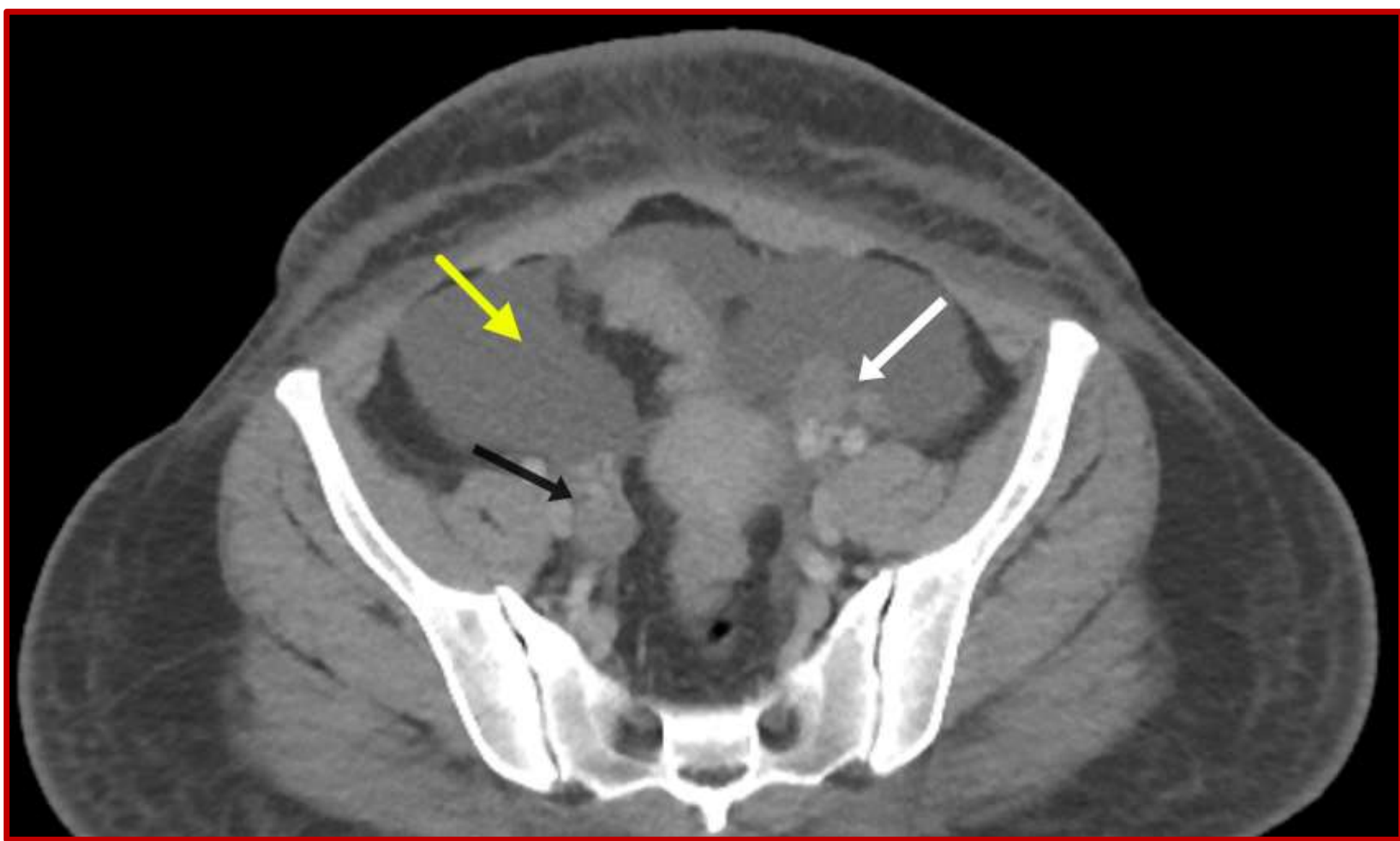


Fig 1: CT abdomen/pelvis: slightly bulky right (black arrow) and left (white arrow) ovaries with ascites (yellow arrow).

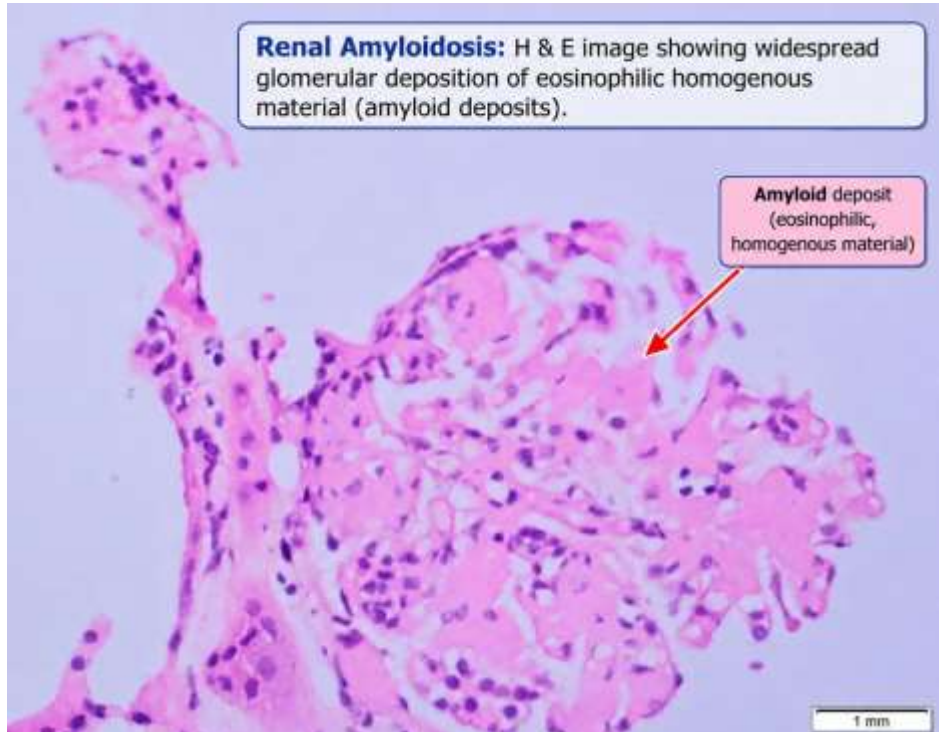


Fig 2: H&E 40x: mesangial expansion with eosinophilic homogeneous deposits; thick, hyalinized basement membranes.

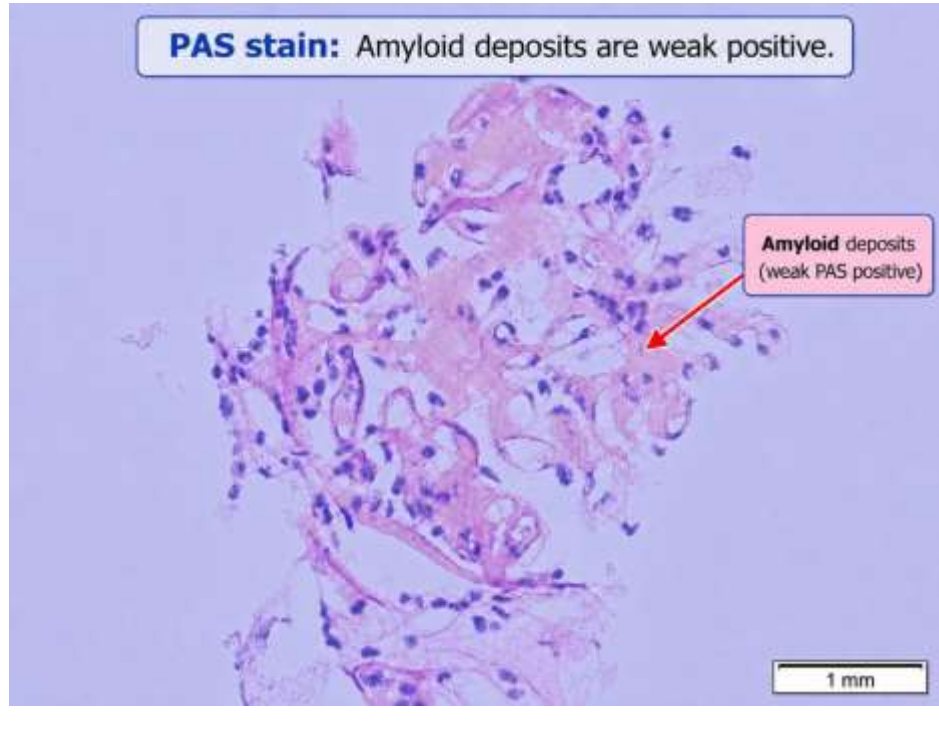


Fig 3: PAS stain: weakly positive.



Fig 4: Jones methenamine silver stain: negative.

Renal biopsy stains
Congo red: POSITIVE
PAS: weakly positive
JMS: negative
Serum: IgG-lambda

TABLE 1. CHRONOLOGICAL SUMMARY OF MAJOR EVENTS

Date	Event/Investigation	Key Findings/Outcome
09-11-2021	Initial Walk-in Assessment & CT scan	Abdominal distension, ascites and bulky ovaries
17-11-2021	Surgical Oncology Clinic Assessment	Raised CA-125 (257 U/mL). Workup for ovarian CA started
18-11-2021	MRI Pelvis & CT Thorax	Bulky ovaries (no mass) and minimal pleural effusion
23-11-2021	Ascitic fluid aspiration	Cytology revealed mixed inflammatory cells comprising lymphocytes, neutrophils and histiocytes (benign)
01-12-2021	MDT Discussion	CA-125 further increased to 1024 U/ml. Plan for diagnostic laparoscopy and debulking surgery
23-12-2021	OGD & Colonoscopy	Benign rectal polyps with no evidence of GI malignancy
03-01-2022	Pre-op Anesthesia assessment	Ver low albumin (1.54 g/dL) with ascites and pedal edema
04-01-2022	Nephrology evaluation	CKD Stage 3a and Nephrotic Syndrome identified. Surgery was deferred and further workup initiated.
12-01-2022	Renal Biopsy	Renal biopsy performed
21-01-2022	Nephrology Follow up	Low Kappa / lambda ratio and SPEP showing a monoclonal spike in the gamma region. Renal biopsy positive for amyloidosis involving all glomeruli and focally medullary vessels, confirming Immunoglobulin related Amyloidosis (AL amyloidosis)
27-01-2022	Second MDT discussion	Ovarian CA was ruled out, and patient was referred to hematology
07-02-2022	Lymphoma MDT & Hematology Clinic	Diagnosis of AL amyloidosis was explained and referred outside for definitive treatment.

DISCUSSION

- This case underscores a rare but clinically significant diagnostic pitfall in which systemic AL amyloidosis closely simulated advanced ovarian malignancy.
- The lack of a discrete ovarian mass on cross-sectional imaging and negative ascitic cytology were discordant with uncomplicated ovarian cancer. Subsequent development of severe hypoalbuminemia, anasarca, and heavy proteinuria instead pointed toward another systemic disorder.
- The persistently elevated CA-125 level in our patient remained a major factor directing the diagnostic consideration toward advanced ovarian malignancy.
- CA-125 is released by mesothelial cells whenever the peritoneal or pleural surface is stretched by fluid, regardless of cause. Because no validated CA-125 cutoff exists for non-malignant settings, extremely high values, as in our patient, do not by themselves confirm an ovarian primary.
- The decisive diagnostic step in this case was the recognition of nephrotic syndrome, followed by renal biopsy and monoclonal protein assessment, which demonstrated amyloid deposition with IgG-lambda restriction and confirmed AL amyloidosis with renal involvement.
- The histopathologic diagnosis of amyloidosis remains anchored in Congo red staining with characteristic apple-green birefringence under polarized light.
- Once amyloid is identified, subtyping is essential, since management differs fundamentally between AL, AA, ATTR, and hereditary forms.
- Viewed together, this body of literature situates the present case within a recognized, if underappreciated, category of diagnostic mimicry: a rare clonal disorder producing organ dysfunction and serosal fluid shifts that, through a shared final common pathway of mesothelial CA-125 release, reproduces the biochemical signature of ovarian malignancy.
- Careful clinicopathologic correlation, rather than tumor-marker level alone, is essential, because this distinction determines prognosis, the need for hematologic therapy, and the extent to which radical oncologic surgery may be avoidable.

CONCLUSION

This case highlights systemic AL amyloidosis as a rare but important mimic of ovarian malignancy and should be considered in a patient with unexplained proteinuria or hypoalbuminemia, to avoid unnecessary surgery and allow timely, organ-appropriate treatment.

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