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**V Edition of the Clinical Cases Contest on**

**non-surgical clinical management of Kidney Stones**

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**Title:** Management of Renal Lithiasis in a Patient with Distal Renal Tubular Acidosis and Horseshoe Kidney: A Case Report

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**Key words (3 to 6):** Horseshoe kidney, distal renal tubular acidosis, hypercalciuria, hypocitraturia

## 1. Abstract (no longer than 150 words).

**Objective**: To present a rare and interesting case along with its diagnostic and therapeutic management.

**Material and Methods**: A 62-year-old male patient with renal lithiasis was included. Subsequently, complementary tests, therapeutic management, and preventive strategies along with follow-up over two years were performed.

**Results**: This is a case of distal renal tubular acidosis (dRTA) in a patient with a horseshoe kidney. After a percutaneous nephrolithotomy for lithiasis, preventive treatment with thiazides, potassium citrate, and phytate compounds was initiated, along with periodic 24-hour urine monitoring and imaging studies that showed no recurrence of stones after two years.

**Conclusions**: Metabolic studies in high-risk patients are useful for etiological diagnosis in stone formation. The use of potassium citrate and phytate compounds can be effective in preventing recurrence of lithiasis in patients with dRTA.

## 2. Introduction

Horseshoe kidney is the most common form of renal fusion, with an estimated incidence of 1 in 400 people. It is characterized by the fusion of the kidneys via a parenchymal or fibrous isthmus, which prevents correct rotation, resulting in anteriorly oriented renal pelvises and alterations in calyceal and ureteral disposition. Although it is usually asymptomatic, the incidence of renal stones in patients with horseshoe kidney is estimated at 35%. In these cases, lithiasis is typically associated with impaired pyeloureteral drainage (1).

However, it is important to conduct a metabolic study to identify the correct etiology of stone formation. Though rare, distal renal tubular acidosis (dRTA) can also be associated with horseshoe kidney. dRTA is a rare disorder (incidence 1 in 100,000 people) characterized by impaired ammonium (NH4) excretion at the

collecting duct level, leading to systemic metabolic acidosis and alkaline urine pH. dRTA is associated with a higher incidence of renal lithiasis due to secondary urinary hydroelectrolytic abnormalities (hypocitraturia and hypercalciuria), and in severe cases, nephrocalcinosis. In adults, dRTA can be challenging to diagnose, as it is typically identified in childhood, and in older individuals, the presentation is usually milder, with fewer analytical repercussions (2). This case report details the diagnosis, treatment, and follow-up of a complex and rare case of renal lithiasis in a patient with distal renal tubular acidosis associated with a horseshoe kidney.

## 3. Clinical Case description

### a. Patient information / Medical records

We present the case of a 62-year-old male with no relevant personal or family history, who presented to the clinic after an incidental finding of a 2 cm hypodense lesion suggestive of left renal lithiasis on an abdominal X-ray, in a horseshoe kidney.

### b. Diagnostic support studies and results

- **Blood Chemistry**: Normal, with no urinary pH abnormalities.
- **Computed tomography** (Image 1): The imaging findings prompted consideration of various therapeutic options, ultimately leading to a percutaneous nephrolithotomy with complete removal of the renal stones (Image 2).

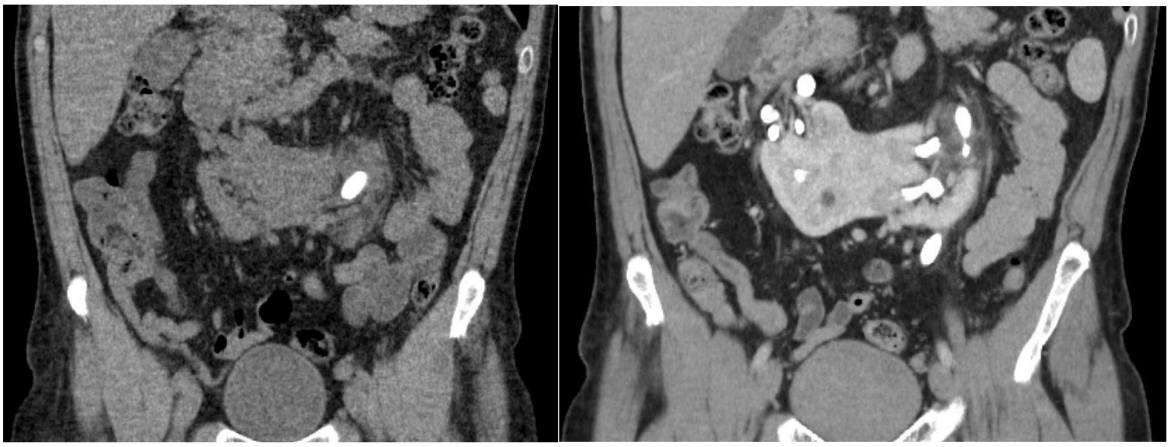

**Image 1.** *Computed tomography showing a 23 x 16mm stone with a density of 1065 HU.*

- **Stone Analysis**: The extracted stone was composed of 70% calcium oxalate dihydrate and 30% calcium phosphate (apatite carbonate).

First laboratory results:

- **Blood Chemistry**: Creatinine 0.58 mg/dL, GFR 100 mL/min, Uric acid 2.8 mg/dL, Albumin 4.8 g/dL, Calcium 9.0 mg/dL, Phosphorus 3.4 mg/dL, Magnesium 1.78 mg/dL, Chloride 102 mEq/L, Sodium 143 mEq/L, Potassium 4.4 mEq/L.
- **Hormones**: PTH 88.5 pg/mL, Vitamin D-25 Hydroxylase 11.7 ng/mL.
- **Urine Sediment**: Density 1.020 g/L, pH 6.5, Negative glucose, ketones, urobilinogen, bilirubin, proteins, nitrites, leukocytes, with traces of hematuria.
- **24 hour urine sample**: Diuresis 900 mL, Creatinine 2022 mg, Uric acid 382 mg, Calcium 249 mg, Phosphorus 1100 mg, Magnesium 132 mg, Chloride 125 mEq, Sodium 106 mEq, Potassium 46 mEq.

Second laboratory results (post-treatment with vitamin D):

- **Blood Chemistry**: Creatinine 0.62 mg/dL, GFR 98 mL/min, Uric acid 3 mg/dL, Albumin 4.5 g/dL, Alkaline Phosphatase 120 U/L, Calcium 8.9 mg/dL, Phosphorus 3.6 mg/dL, Magnesium 1.90 mg/dL, Chloride 100 mEq/L, Sodium 141 mEq/L, Potassium 4.1 mEq/L.
- **Hormones**: PTH 65 pg/mL, Vitamin D-25 Hydroxylase 25 ng/mL.
- **Urine Sediment**: Density 1.000 g/L, pH 7, Negative glucose, ketones, urobilinogen, bilirubin, proteins, nitrites, leukocytes, traces of hematuria.
- **24 hour Urine sample**: Diuresis 1000 mL, Creatinine 1400 mg, Uric acid 392 mg, Calcium 244 mg, Oxalate 19 mg, Phosphorus 1100 mg, Magnesium 131 mg, Chloride 125 mEq, Sodium 106 mEq, Potassium 46 mEq, Citrate 105 mg.

Furosemide/Fludrocortisone Test: Urinary sediment with pH 5.7 and venous blood gas pH 7.36 HCO3 23 mEq/L.

### c. Diagnosis

Given the patient's anatomical predisposition for lithiasis and the calcium phosphate composition of the stones, a metabolic study was indicated. The patient showed elevated PTH levels with low Vitamin D levels, without significant ionic disturbances but with hyperphosphaturia and alkaline urine pH. A differential diagnosis between primary hyperparathyroidism and Vitamin D deficiency was made. Vitamin D treatment was initiated for one month, and a follow-up analysis revealed correction of Vitamin D-25 hydroxyase levels and normalization of PTH. However, an elevated alkaline phosphatase level persisted, indicating bone resorption. A follow-up 24-hour urine sample showed persistent phosphorus excretion, hypocitraturia, and an alkaline urine pH. The presence of elevated bone resorption markers, an alkaline urine pH, hypocitraturia, and calcium phosphate stones warranted further investigation for distal renal tubular acidosis (dRTA), which was confirmed by a furosemide/fludrocortisone test. Given the absence of concurrent metabolic acidosis, this case was classified as **incomplete dRTA.**

### d. Treatment

The patient underwent a percutaneous nephrolithotomy with complete removal of the large stone (Image 2). A metabolic study led to recommendations for lifestyle modifications and preventive treatment to avoid future stone formation. The patient was prescribed potassium citrate with Lit-Control pH Up® (1 tablet every 12 hours), phytate with Lit-Control pH Balance® (1 tablet every 12 hours), and hydrochlorothiazide (12.50 mg every 24 hours).

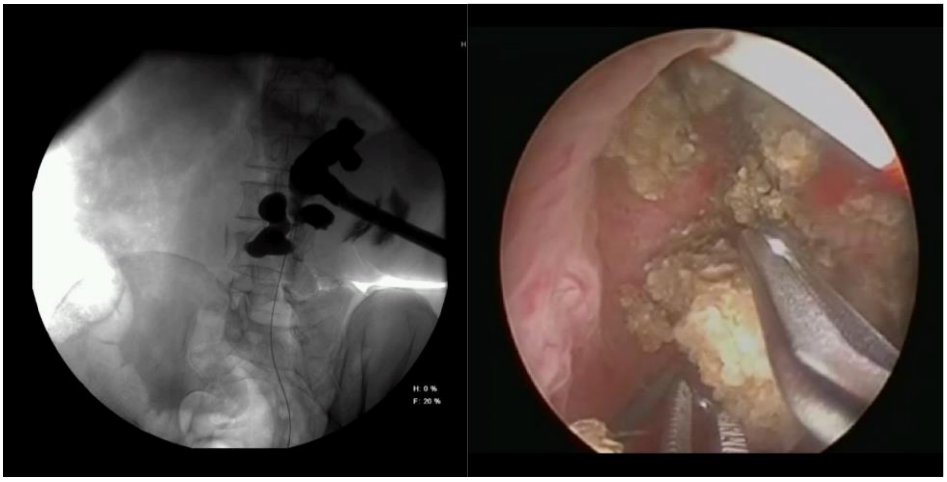

**Image 2.** *Percutaneous nephrolithotomy in a patient with a horseshoe kidney.*

### e. Evolution and progress

The patient was followed up every 3 months during the first year and every 6 months during the second year. He used the myLit-Control App® to improve adherence to both the medical treatment and dietary supplementation. A follow-up 24-hour urine test after 8 weeks showed improved hypocitraturia. A densitometry scan showed osteopenia, and the patient continued Vitamin D treatment initiated at the beginning of the study. The patient remained asymptomatic, without episodes of urinary tract infections, and imaging studies showed no recurrence of stones (Image 3).

### f. Clinical results

Despite being at high risk for stone formation due to anatomical abnormalities and dRTA, the patient remained disease-free after 2 years of follow-up with excellent adherence to therapy. The preventive management strategy successfully controlled stone recurrence.

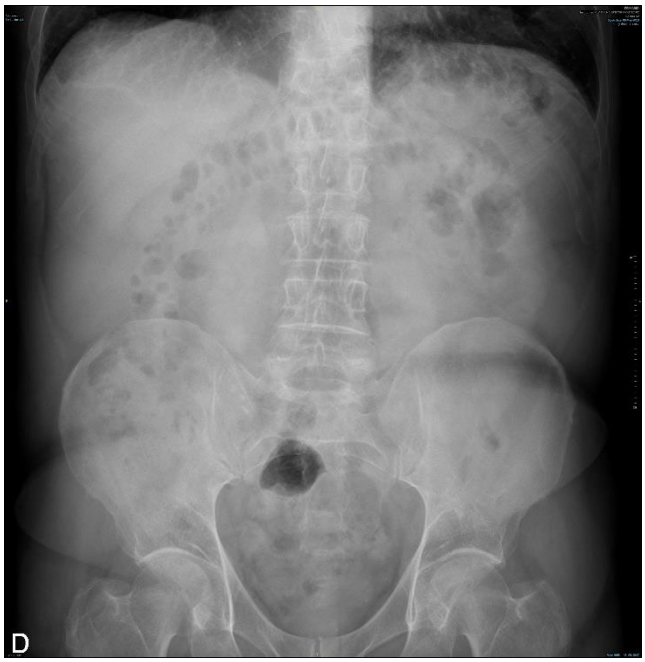


**Image 3.** *Abdominal X-ray after 2 years of follow-up.*

## 4. Discussion

Distal Renal Tubular Acidosis (RTA) is a rare condition but should be considered in stone-forming patients, particularly when calcium phosphate (apatite carbonate) components are present. It is associated with persistently alkaline urine pH and a tendency toward metabolic acidosis, although in incomplete forms, metabolic acidosis may not be present. In these patients, it is important to assess the association with bone resorption by performing a densitometry scan, given the condition's tendency to acidify blood pH (2).

Treatment focuses on reducing calciuria and increasing urinary citrate to lower the risk of stone formation. Potassium citrate formulations are of particular interest (3, 4), especially when combined with a thiazide diuretic to reduce calciuria, as thiazides are associated with hypocitraturia and hypokalemia (5), similar to RTA itself. Additionally, phytate has been shown to reduce the crystallization of both oxalate and calcium phosphate salts in urine (6), while also providing a protective effect against osteoporosis and secondary calciuria due to bone resorption (7). For this reason, a recent article recommended its use in treating incomplete distal renal tubular acidosis (8).

In recent years, with the development of new technologies, the use of mobile applications has been

investigated and shown to be a promising tool for improving adherence to chronic medication regimens (9), with evidence supporting its applicability in stone disease (10).

The presented case is of particular interest as it underscores the importance of metabolic control and its effectiveness even in patients with a predisposition to lithiasis due to both anatomical and metabolic abnormalities.

## 5. Conclusions and recommendations

Metabolic workup in high-risk patients is essential, as it can guide the diagnosis of systemic conditions and the most appropriate therapeutic approach. The case highlights the efficacy of combining potassium citrate and phytate to reduce recurrence risk and prevent stone formation in patients with distal renal tubular acidosis, even when compounded by anatomical predispositions

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