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Hypokalemic Paralysis in a Young Man With Autoimmune Polyglandular Syndrome and Distal Renal Tubular Acidosis: A Diagnostic Dilemma Between Autoimmune and Genetic Etiologies

Namaa Hassan*[1], Dr. Hussameldin Hassan[2]

[1] East-West University, Tbilisi, Georgia
[2] Department of Nephrology and Transplantation, Alhada Armed Forces Hospital,Taif, Kingdom of Saudi Arabia

Abstract
A 21-year-old man presented to the emergency department with acute generalized weakness and paralysis. Investigations revealed severe hypokalemia (serum potassium 1.7 mmol/L), non-anion gap metabolic acidosis, and alkaline urine, consistent with distal renal tubular acidosis (dRTA). His past medical history included primary hypoparathyroidism, type 2 diabetes mellitus, and childhood-onset sensorineural hearing loss. The urine potassium-to-creatinine ratio was elevated, and the patient had no evidence of gastrointestinal loss or diuretic use. His clinical picture raised suspicion of autoimmune polyglandular syndrome or genetic syndromes such as HDR (Barakat) syndrome. He responded to IV potassium and oral bicarbonate therapy. This case highlights the importance of identifying syndromic causes of hypokalemia and metabolic acidosis and explores the diagnostic overlap between autoimmune and genetic causes of dRTA.

Keywords: Distal renal tubular acidosis, Hypokalemia, HDR syndrome (Barakat syndrome), Hypoparathyroidism, Sensorineural hearing loss

Background

Distal renal tubular acidosis (dRTA) is a rare cause of non-anion gap metabolic acidosis and hypokalemia. When it presents with systemic features like endocrinopathy or hearing loss, clinicians must consider syndromic diagnoses. Autoimmune polyglandular syndromes (APS), Sjögren's syndrome, and genetic disorders such as HDR (hypoparathyroidism, deafness, renal anomalies) syndrome may overlap clinically. This case demonstrates how identifying these associations changes both diagnosis and long-term care.

Case Presentation

A 21-year-old university student presented with acute generalized weakness and inability to stand, while at home with his family. This episode resembled prior attacks occurring over three years, all associated with severe hypokalemia and rapid recovery after potassium replacement. He had a history of:

- Primary hypoparathyroidism since childhood (on calcium and vitamin D)
- Sensorineural hearing loss, requiring hearing aids since early age
- Type 2 diabetes mellitus, diagnosed three years prior, managed with metformin
- Photophobia and dry eyes, without dry mouth or dental caries

He reported non-compliance with potassium chloride (taking 1 tab/day vs prescribed 6 tabs/day). No family history of similar illness, hearing impairment, or endocrinopathies. He was otherwise cognitively normal but had academic delays due to frequent hospitalizations.

On examination:

- Short stature (150 cm), normal BMI, dry skin/tongue
- BP 120/70 mmHg, HR 75 bpm, no postural drop
- ENT, CVS, respiratory and abdominal exams were unremarkable
- No rash, ulcers, or joint signs

Investigations

- Serum potassium: 1.7 mmol/L
- ABG: pH 7.39, HCO_3^- 13 mmol/L, PCO_2 22 mmHg → mixed high and normal anion gap metabolic acidosis
- Anion gap: 16; Delta ratio: 4/11 → suggests combined high AG + non-AG acidosis
- Urine pH: >5.5
- Urine potassium: 56 mmol/L; urine K/Cr ratio = 28 → renal potassium wasting
- ECG: Prominent U waves
- KUB ultrasound/X-ray: No nephrocalcinosis or structural renal anomalies

Differential Diagnosis

- Autoimmune dRTA: possibly part of APS or Sjögren's syndrome
- Hereditary dRTA: autosomal recessive forms can be associated with SNHL
- HDR Syndrome (Barakat): Triad of hypoparathyroidism, deafness, and renal defects
- Gitelman/Bartter syndromes: Unlikely due to metabolic acidosis
- Thyrotoxic periodic paralysis: No hyperthyroidism; unlikely

Treatment

Immediate:

- Cardiac monitoring
- IV potassium chloride in dextrose-free solution

Maintenance:

- Potassium chloride 1200 mg PO TID
- Potassium citrate 600 mg PO TID
- Sodium bicarbonate 1 g PO TID
- Magnesium oxide

Outcome and Follow-up

The patient responded to IV potassium with resolution of weakness. He was discharged with oral potassium and bicarbonate therapy. He declined further inpatient evaluation due to pending university exams. Outpatient referral was arranged for endocrinology, nephrology, and possible genetic testing. His case remains under multidisciplinary follow-up.

Discussion

This case illustrates a complex interplay between renal electrolyte disturbance and systemic disease. dRTA can present with severe hypokalemia leading to paralysis. The patient's biochemical findings—non-anion gap metabolic acidosis, inappropriately alkaline urine, positive urine potassium loss—support a diagnosis of dRTA.

The association with primary hypoparathyroidism, sensorineural deafness, and dry eyes raised the possibility of both autoimmune polyglandular syndrome (APS) and HDR syndrome. APS type 1, though rare, is a plausible cause when multiple endocrine and autoimmune features are present. Sjögren's syndrome is a more common autoimmune cause of dRTA, though less likely given his age and male sex.

HDR syndrome, caused by mutations in GATA3, can present with the triad of hypoparathyroidism, deafness, and renal anomalies. While our patient had no obvious nephropathy on imaging, genetic testing may still be warranted.

Due to his refusal to remain for full workup, this case remains a diagnostic dilemma, but highlights the need for syndromic thinking in cases of unexplained metabolic abnormalities.

Learning Points

- Distal RTA can cause life-threatening hypokalemia and may present as flaccid paralysis.
- The presence of metabolic acidosis with a positive urine anion gap and high urine pH confirms dRTA.
- Autoimmune and genetic syndromes must be considered in young patients with systemic features.
- Management includes potassium and alkali replacement, along with screening for underlying systemic disease.
- Patient education and long-term follow-up are essential in managing chronic dRTA and preventing recurrence.

Patient's Perspective

"It was terrifying to suddenly not be able to stand. I've lived with this for years without knowing the real cause. Now that I understand it might be something more than just low potassium, I want to stay on treatment and finish my studies."

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