Distal renal tubular acidosis: Results of an online survey

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OBJECTIVES

Defining the clinical features of patients with inherited distal Renal Tubular Acidosis (dRTA) with respect to

1. Age at presentation
2. Complications
3. Treatment
4. Long-term outcome
## METHODS

1. Paediatric and Adult Nephrologists were contacted through European professional organizations
2. Collected demographic, clinical, genetic and biochemical data via online questionnaire
3. Patients were included if data provided was sufficient and consistent with diagnosis of primary dRTA
4. Physicians were re-contacted if necessary to complete data or resolve questions
A total of 393 responses were received
- 3 misdiagnosed (no acidosis)
- 59 with incomplete information

Total of 331 cases (52% female) for final analysis
- Age range: 0-70 years, with 83 (25%) >18 years
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• 198 (60%) had genetic screening (SLC4A1, ATP6V1B1, ATP6V0A4)

• Mutation identification in 168 (85%)
Age

- Median age at last follow-up: 11 years
Treatment

• A total of 34 different alkali preparations were used
• 84 (25%): Bicarbonate, 132 (40%) citrate and 113 (34%) with both
Long-term outcome: Height
Long-term outcome: eGFR

- Mean eGFR (Cockcroft-Gault) in adults: 94 ml/min/1.73m²
- 29 (52%) patients >20 years with CKD stage ≥ 2 (9 stage 3, 1 stage 4)
- Compares to 33% CKD stage ≥ 2 in NHANES III
Nephrocalcinosis/lithiasis

**Nephrocalcinosis**

- ATP6V1B1: 89%
- ATP6V0A4: 98%
- SLC4A1: 91%
- Unknown: 81%

**Lithiasis**

- ATP6V1B1: 80%
- ATP6V0A4: 80%
- SLC4A1: 53%
- Unknown: 80%
Hearing

RESULTS (6)

- Hearing loss
  - No
  - Yes

- Hearing aids
  - No
  - Yes

- Cochlear implants
  - No
  - Yes

- Hearing aids prescription (age, years)

- Mutations:
  - ATP6V1B1
  - ATP6V0A4
  - SLC26A1
  - Unknown

Graphs a), b), c), and d) illustrate the distribution of hearing impairment and the use of hearing aids and cochlear implants across different mutations.
1. Largest cohort of patients with primary dRTA
2. Treatment adequacy as judged by plasma bicarbonate and normocalciuria is achieved in most patients
3. Treatment dose decreases with age
4. Adult height is slightly impaired, but in the normal range in 91%
5. Half of adult patients have decreased eGFR.
6. Hearing loss is most severe in the group of patients with B1 subunit mutations.