Case report

Renal tubular acidosis with nerve deafness

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A child with renal tubular acidosis (RTA) and sensorineural deafness is described. This association represents a subtype of RTA and is inherited as an autosomal recessive trait. Late diagnosis of deafness occurred despite frequent follow-up, indicating the need for continual expert audiological assessment in all children with complex renal disease.

INTRODUCTION

Renal tubular acidosis (RTA) is a non-uraemic clinical syndrome of disordered renal acidification. In the classic distal RTA (Type 1), subjects have hyperchloraemic acidosis, hyponatraemia and hypokalaemia. The urinary pH remains high (pH > 6.2), even in the presence of systemic acidosis. Although the defect is permanent, the prognosis is good when the diagnosis, with correct management, is established early in order to prevent nephrocalcinosis and secondary renal damage. Inheritance of the classic distal RTA (Type 1) is autosomal dominant. Several subtypes of distal RTA have been described such as incomplete distal RTA, distal RTA with bicarbonate wasting, transient distal RTA in infants and distal RTA with nerve deafness. We describe a patient, to our knowledge the first case from Ireland, with distal renal tubular acidosis and bilateral sensorineural deafness.

CASE HISTORY

JC was born to non-consanguineous parents following an uncomplicated pregnancy, by normal delivery at term, weighing 4.1kg. There was no family history of genetic disease or congenital abnormalities. At one month, he became dehydrated, refusing feeds and vomiting. Investigations showed hypokalaemia and hyperchloraemic metabolic acidosis. The diagnosis of renal tubular acidosis was suspected and potassium and bicarbonate replacement therapy commenced. During his first two years he required frequent hospital admissions for recurrent dehydration, vomiting, acidosis and electrolyte imbalance precipitated by minor illnesses. Delay in speech was recognised from an early age. A bilateral flat sensorineural hearing loss with thresholds of 60db was diagnosed at three years (Free Field performance test). No hearing defect was suspected at routine health visitor assessment at eight months. At four years, while receiving bicarbonate...
(1 mmol/kg/day) and potassium (2 mmol/kg/day) he was noted to have marked polydipsia and polyuria (>3 litres/day). The only clinical abnormalities found were short stature — height 88.8 cm (3rd centile), weight 12.4 kg (3rd centile) and deafness.

Initial investigations showed hypokalaemia (K 2.4 mmol/l), hyperchloraemia (Cl 117 mmol/l), a partially compensated metabolic acidosis (pH 7.31, pCO₂ 27 mmHg, bicarbonate 16.2 mmol/l, base excess 11.8 mmol/l) and an alkaline urine (pH 7.4). Serum sodium, urea and creatinine were normal. After fluid restriction, urine osmolality was 148 mosmol/kg and desmopressin administration did not reduce urine output or increase urine osmolality. Radiologically, there was bilateral nephrocalcinosis but no evidence of rickets. Urinary excretion of calcium was elevated, 2.79 mmol/24 hours (9 mg/kg/day). Chromatography showed no excess aminoaciduria.

He was discharged on an increased dose of sodium bicarbonate (5 mmol/kg/day) but still required potassium supplements (2 mmol/kg/day). After one month, the urinary output had decreased to 1.3 l/day (mean of over three days). Serum electrolytes and acid base balance were normal and urinary calcium excretion had fallen to normal, 0.52 mmol/24 hours (1.7 mg/kg/day).

At the most recent review the high requirement for bicarbonate and potassium has continued (six years). The audiogram shows a 'ski slope' pattern with profound hearing loss (Figure). He now attends a school for the deaf and is making satisfactory progress. We believe this is the first report of this association in Ireland.

![FIGURE.](image)

Pure tone audiogram. Open circles – right ear; closed circles – left ear.
DISCUSSION

Our patient's initial presentation was typical of distal renal tubular acidosis. With the subsequent recognition of bilateral sensorineural deafness, it became clear that he had the subtype of RTA with deafness which is recognised as a distinct entity with an autosomal recessive mode of inheritance. The associated deafness may occur at birth or later in childhood. The late diagnosis of deafness in our patient and in other reports emphasises the need for early and repeated expert audiological assessment in children with RTA. Use of new techniques such as brain stem auditory evoked responses may be of help with these children.

Although the initial presentation of this case was typical, some features of the subsequent course were unexpected. Firstly, potassium supplements are not usually required to maintain normokalaemia when acidosis is corrected. In our patient their withdrawal caused a precipitous fall in serum potassium despite full bicarbonate replacement. Secondly, nephrocalcinosis developed despite bicarbonate replacement in the normal dosage of 1 – 3 mmol/kg/day from one month of age. Increasing bicarbonate therapy to 5 mmol/kg/day resulted in a reduction in urinary output, a fall in urinary calcium excretion to optimum levels (< 2 mg/kg/day), and catch-up growth (height velocity 9.6 cm/year).

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