

Audiological correlates of renal tubular acidosis - a case report

Abstract

Renal Tubular Acidosis (RTA) refers to a condition where the kidneys do not appropriately eliminate acids from the blood into the urine resulting in acid remnants in the blood. It could be caused either by dysfunction of distal renal tubules or proximal renal tubules. Homeostasis of the cells, cochlear fluids and generation of endocochlear potential are important factors which enable normal cochlear function. Hearing loss can be caused by disturbances in cochlear homeostasis due to certain gene mutations. In this report, a child with proximal Renal Tubular Acidosis who was evaluated and diagnosed as having bilateral sensorineural hearing loss is documented. This report aims to highlight the importance of early diagnosis of hearing loss and provide appropriate intervention to reduce the impact of hearing loss on the child's language, speech, education and social life.

Keywords: renal tubular acidosis, homeostasis, electrolytes, sensorineural hearing loss

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Abbreviations: RTA, renal tubular acidosis; pRTA, proximal renal tubular acidosis; dRTA, distal renal tubular acidosis: ATPase, adenosine tri phosphatase

Introduction

Acidosis occurs when acid builds up, or when bicarbonate (a base) is lost leading to electrolyte imbalances in the blood resulting in disruption of normal functioning of certain cells. Renal Tubular Acidosis (RTA) refers to a condition where the kidneys do not appropriately eliminate acids from the blood into the urine thus leaving traces of acid in the blood. RTA defines a class of disorders in which excretion of hydrogen ions or re absorption of filtered bicarbonate ions are impaired, leading to chronic metabolic acidosis with a normal anion gap. Hyperchloremia is usually present, and secondary dysfunction may involve other electrolytes, such as Potassium (frequently) and Calcium.

RTA could be caused either by dysfunction of distal renal tubules or proximal renal tubules. Primary distal renal tubular acidosis (dRTA)¹ is characterized by hyperchloremic metabolic acidosis due to failure in proton excretion, variably severe nephrocalcinosis and/or nephrolithiasis associated with hypercalciuria and hypocitraturia. Mutations of ATP6V1B1 or ATPV0A4 genes, causes sensorineural hearing loss in renal tubular acidosis.² The former is associated with early onset of hearing loss and the latter with late onset.

dRTA is inherited as either an autosomal dominant or autosomal recessive trait. The autosomal recessive inherited syndrome of distal renal tubular acidosis and sensorineural hearing loss may present in one of two distinct fashions. The rare adolescent form is characterized by mild renal tubular acidosis, mild to moderate sensorineural hearing loss, and otherwise normal growth and development. The more common infantile type typically presents in the first year of life with failure to thrive, acidosis, and a more severe sensorineural hearing loss.^{3,4}

Proximal RTA (pRTA) is caused by a failure of the proximal renal tubules to reabsorb filtered bicarbonate effectively, which results in bicarbonate wastage and acidemia. Since the distal renal tubular function is unaffected, the effects of acidemia are less severe than that seen in dRTA⁵. pRTA may be rarely present in isolation or more

commonly associated with Fanconi syndrome, which is characterized by phosphaturia, glycosuria, amino aciduria, uricosuria and tubular proteinuria. As a result of phosphate wasting, and abnormal Vitamin D metabolism, rickets or osteomalacia is seen as the main feature of this syndrome.

Homeostasis of the cells, cochlear fluids and generation of endocochlear potential are important factors which enable normal cochlear function hence regulation of pH in the cochlea is important to prevent hearing loss.⁶ It has been reported in literature that mutation of certain genes which alter the homeostasis are known to cause hearing loss in humans. The cochlea contains endolymph whose pH depends on HCO₃⁻ and H⁺ secretion and Carbonic Anhydrase activity.⁷ Prolonged effects of cochlear acidosis may reduce the energy metabolism and increase free radical generation and activate the innate immune system.⁸ Potassium ion is crucial for the generation of endocochlear potential and calcium concentration in the endolymph is critical for normal cochlear function. RTA with hearing loss is caused by mutations in the genes which are responsible for production of proteins, which are sub units of the large protein complex - Vacuolar H⁺ -ATPase (V-ATPase). These complexes are found in the inner ear and nephrons. These act as pumps to move H⁺ across membranes. Acids donate protons to other molecules. Hence the movement of protons is crucial in regulating the pH of cells. Thus mutations in the ATP6V1B1 or ATP6V0A4 gene cause impaired functioning of the protein complex V-ATPase, subsequently resulting in inability to maintain pH of blood and cochlear fluid.

Prevalence

Renal tubular acidosis with deafness is a rare disorder.

Case presentation

A three and a half year old girl born of a consanguineous marriage was brought with the complaint of being non-responsive to her name and inability to speak age appropriately. At the age of four and a half months, when the child was being treated for dehydration and failure to thrive, investigations revealed the presence of RTA Type 2. Consequently, due to more serious medical concerns, the hearing loss was overlooked, though the parents confirmed the manifestation of hearing loss at four and a half months of age.

Sibling history

The child reported here, is the second of three children, born. The first child, born prematurely, died at the age of two and a half years. The cause of death was not established. The third child, a one and a half year old boy, was diagnosed with the same condition that is RTA Type 2 at the age of eight months. The child was brought a week after the older sibling was evaluated for hearing loss, and was diagnosed as having profound hearing loss.

Audiological evaluations

Behavioral observation Audiometry revealed responses to tones at 85 dBHL. Immittance Audiometry revealed 'A' type of tympanogram with absence of stapedial reflexes. Distortion Product Otoacoustic Emissions were absent in both ears. Auditory Brainstem evoked Response to click stimulus confirmed the presence of bilateral profound hearing loss. Subsequent evaluation was done following parental observation of a change in response to sounds after a period of five months. The child's behavioral thresholds at low frequencies in the left ear were slightly better than those in the high frequencies. Conditioned Audiometry revealed profound hearing loss in the right ear and moderately severe to severe sensorineural hearing loss in the Left ear. Following the diagnosis, hearing aids were prescribed after performing real ear measurements and noting the behavioral thresholds. Since the child was benefiting from hearing aids, she was recommended to use them for audiological habilitation.

Speech and Language evaluation showed age appropriate receptive and expressive gestural age on the Gestural scale. Cognition was delayed by four months.

Discussion

RTA can be inherited as an autosomal recessive or autosomal dominant, or acquired. The significance of genetic testing and counseling cannot be underemphasized. RTA can also be drug-induced. Some of the drugs that could induce RTA are anticonvulsants, such as Valproic acid, certain anti-retroviral drugs, anti cancer drugs such as Cisplatin. Hence patients who are administered these drugs should be closely monitored through periodic hearing evaluations. Sensorineural hearing loss is one of the common features associated with RTA. Hearing loss could be manifested at the time of diagnosis of RTA or at a later stage. Since hearing loss is progressive in cases of RTA, patients diagnosed with this condition, should undergo a series of Audiological evaluation at regular intervals. Children diagnosed with pRTA require administration of alkali to prevent growth retardation. Alkali, potassium phosphate and Vitamin D are given to reduce bone demineralization. In infants and children having hearing loss associated with RTA, hearing aids used along with auditory habilitation, can improve the speech and language development.

This report highlights the importance of early diagnosis of hearing loss and appropriate intervention to reduce the impact of hearing loss on the child's language, speech, education and social life.

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Conflicts of interest

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