Renal Tubular Acidosis in Down Syndrome: A Case Report

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ABSTRACT

Down syndrome (DS) is often associated with serious cardiac malformations, so renal involvement in these patients is infrequently studied. The objective of this report was to draw attention to renal abnormalities and their potential progression to chronic renal failure in patients of Down syndrome. We present a case report of a 4-month-old female patient of Down syndrome, who was diagnosed with renal tubular acidosis. In conclusion, renal abnormalities in patients of DS should be considered, so as to maintain effective renal functions.

Key Words: *Down's syndrome, Renal tubular acidosis*

Introduction

Down syndrome (DS) is one of the most frequently observed hereditary cause of disability in learning and is reported in 1 of 920 live births. DS can affect several body systems; however, involvement of kidneys is occasional. About 3.5–21.4% cases of DS are associated with occurrence of kidney and urinary tract abnormalities.1

Primary distal renal tubular acidosis (dRTA) is an infrequent inherited ailment that disturbs the capacity of the kidneys to eliminate acid from the blood, leading to metabolic acidosis. It affects males and females alike, however, the actual number of people who have this disorder is not known. Rare disorders like primary distal renal tubular acidosis are frequently either undiagnosed or incorrectly diagnosed, therefore making it hard to estimate their actual frequency in the population.2

Case Report

A 4-month-old girl presented to ANTH with the presenting complaints of failure to thrive, and intermittent vomiting for the last three months. Since the age of one month, she had been hospitalized several times due to vomiting, which would subside after antibiotic therapy. She was delivered at a local hospital, at full term by spontaneous vaginal delivery and cried easily after birth. Her birth weight was 2.5 kg with no antenatal, natal or postnatal complications, however, she showed delayed milestones and neck holding was absent till four months of age.

Patient had characteristic features of Down syndrome, including typical facies with upward slanting eyes, a depressed nasal bridge, low muscle tone and a Simian crease. A typical 'machinery' murmur was heard on chest auscultation and echocardiography revealed a small patent ductus arteriosus (PDA).

Physical examination at the time of admission revealed a visibly sick, toxic and wasted baby girl with signs of dehydration. On admission, weight was 2.4 kg which was significantly less than that of a 4-month-old baby. Her length was 55 cm. There was generalized wasting of muscles, however, no jaundice, cyanosis, or clubbing was seen.

Initial laboratory studies revealed a red blood cell count of

4.08 million/mm3, WBC count of 16,330/mm3, and a platelet count of 2,76,000/mm3. Her C-Reactive Protein was raised,

serum sodium was 141 mEq/L, potassium 4.1 mEq/L and chloride 119 mEq/L. Her serum urea level was 123 mg/dl, uric acid, 12 mg/dl and creatinine, 1mg/dl. Serum ammonia and lactate were within normal limits. After the correction of hydration status, renal function tests were repeated and were found to be normal.

Arterial blood gas levels showed metabolic acidosis despite adequate hydration (pH= 7.26, HCO3 = 8 mEq/L) and a fixed anion gap. Urine pH was 7.5 and showed no bacterial growth on culture. Urine anion gap was 33.3. Renal ultrasound revealed bilateral nephrolithiasis.

Renal tubular acidosis was diagnosed on the basis of fixed anion gap, metabolic acidosis, nephrolithiasis, alkaline urinary pH, increased urinary anion gap and failure to thrive.

The child's vitals and hydration status were monitored. Bicarbonate was given at the rate of 2-4 mEq/kg/24hr. The baby started gaining weight and was discharged at 3.5 kg.

Discussion

Down syndrome is among the most frequently observed inherited cause of learning disabilities in children. While the incidence of kidney and urinary tract involvement in DS is quite uncommon, even so monitoring of such patients for renal involvement should be performed.1 Previous studies have suggested that early renal ultrasound studies may be valuable in detecting many of these anomalies and may be helpful for early diagnosis and management to decrease morbidity in individuals suffering from Down syndrome.3

Patients with DS present quite often with hereditary urogenital disorders. There are also various studies relating a syndrome comprising of hypercalcemia, hypercalciuria, medullary calcinosis, and renal failure in patients.4

In Distal Renal Tubular Acidosis (dRTA) also known as Classic RTA or Type 1 RTA, the capability to make the urine acidic is absent due to an error in the elimination of ammonium and H+ ions by certain specialized cells of the collecting duct called the alpha-intercalated cells.5

Information regarding the prevalence of this condition in the general population is not confirmed, however, a study conducted in Thailand, showed that the prevalence of an endemic form of dRTA was 2.8%.6 The genetic forms of dRTA are more prevalent in areas of high consanguinity (Arabianpeninsula and North Africa) whereas, acquired dRTA has been reported more frequently in Western countries.7 Inherited forms of dRTA often make their clinical appearance felt during infancy or childhood. In dRTA, patients may show grave failure to thrive, nephrocalcinosis, severe metabolic acidosis or only minor clinical symptoms, like mild metabolic acidosis, chance detection of renal stones, etc.8

In children, dRTA is usually observed as a prime occurrence. Main clinical findings are diminished growth, polyuria, hypercalciuria, nephrocalcinosis, lithiasis, and K+ depletion. Nephrocalcinosis may advance to chronic renal failure.9 In addition to promoting chronic kidney disease (CKD) advancement, metabolic acidosis has been identified as a reason of resistance to Insulin, reduced secretion of growth and thyroid hormones, protein breakdown leading to muscle wasting, demineralization of bones, increased β2-microglobulin accumulation and elevated mortality rates.10

Clinically, dRTA in pediatric patients usually present with polyuria, constipation, breathing problems, renal stones and failure to thrive.11 Although literature has described occurrence of isolated renal tubular acidosis, hardly any cases have been reported in literature that have described renal tubular acidosis in association with DS.

Conclusion

The present study highlights the fact that every case of Down syndrome should be treated individually and all body systems should be assessed in detail. Moreover, all patients suffering from RTA should be cautiously assessed to avert adverse problems, reveal a possibly correctable disorder, and prevent its advancement to chronic renal disease.

References

1. Kute VB, Vanikar AV, Shah PR, Gumber MR, Patel HV, Engineer DP, et al. Down Syndrome with End- Stage Renal Disease. IJCB. 2013; 28(4): 429-32.

2. Bajpai A, Bagga A, Hari P, Bardia A, Mantan M. Long-term outcome in children with primary distal renal tubular acidosis. Indian pediatr. 2005; 42(4): 321-8.

3. Blazek N, Volansky R. Children with Down syndrome had increased risk for renal, urinary tract anomalies. Infectious Diseases in Children. 2009; 22(11): 16.

4. Kosmadakis G, Smirloglou D, Gobou A, Draganis T,

Michail S. Hemodialysis treatment on an adult patient with down syndrome associated with ectopic right kidney chronic obstructive nephropathy and secondary amyloidosis. SJKDT. 2013; 24(2): 322.

5. Escobar L, Mejía N, Gil H, Santos F. Distal renal tubular acidosis: a hereditary disease with an inadequate urinary H+ excretion. Nefrología (English

Edition). 2013; 33(3): 289-96.

6. Vasquez-Rios G, Westrich DJ, Philip I, Edwards JC, Shieh S. Distal renal tubular acidosis and severe hypokalemia: a case report and review of the literature. J.Med.Case Rep. 2019; 13(1): 103.

7. El Mouzan MI, Al Salloum AA, Al Herbish AS, Qurachi MM, Al Omar AA. Consanguinity and major genetic disorders in Saudi children: a community-based cross-sectional study. Ann Saudi med. 2008; 28(3): 169-73.

8. Mohebbi N, Wagner CA. Pathophysiology, diagnosis and treatment of inherited distal renal tubular acidosis. J.Nephrol. 2018; 31(4): 511-22.

9. Soriano JR. Renal tubular acidosis: the clinical entity.

JASN. 2002; 13(8): 2160-70.

10. Dhondup T, Qian Q. Electrolyte and acid-base disorders in chronic kidney disease and end-stage kidney failure. Blood purif. 2017; 43(1-3): 179-88.

11. Ranawaka R, Dayasiri K, Gamage M. A child with distal (type 1) renal tubular acidosis presenting with progressive gross motor developmental regression and acute paralysis. BMC research notes. 2017; 10(1):618.