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A Case Report on Proximal Renal Tubular Acidosis with Ocular Findings

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Abstract: ***To report a rare case of proximal renal tubular acidosis associated with characteristic ocular findings such as band shaped keratopathy and cataract, in an eleven - year - old female child presented with hyperchloremic normal anion gap metabolic acidosis, developmental delay, stunted growth, bilateral globus pallidum calcifications and dental anomalies. This case report highlights the importance of early and regular ophthalmological evaluation in patients with renal tubular acidosis for better prognosis and management.***

Keywords: proximal renal tubular acidosis, band keratopathy, cataract

1. Introduction

Renal tubular acidosis is clinically characterized by a normal anion gap, hyperchloremic metabolic acidosis, and associated failure to thrive secondary to growth failure.1 It can be classified as type 1 – distal, type 2 - proximal, type 3 – a mixed type that exhibits both impaired proximal HCO_3^- reabsorption and impaired distal acidification, and type 4 – a heterogeneous group of disorders that is characterized by low urine NH_4^+, caused by hyperkalaemia or by aldosterone deficiency.1 These disorders can be primary, from genetic defects in tubular transport mechanisms, or secondary to systemic diseases and adverse drug reactions.1

Proximal renal tubular acidosis is caused by a reduction in bicarbonate reabsorption at the proximal tubules, resulting in low renal bicarbonate threshold. It can present as a part of Fanconi's syndrome or as isolated which is further divided into three types: autosomal recessive proximal renal tubular acidosis with ocular abnormalities, autosomal recessive proximal renal tubular acidosis with osteopetrosis and cerebral calcification, and autosomal dominant proximal renal tubular acidosis. Autosomal recessive proximal renal tubular acidosis with ocular abnormalities is the commonest form of isolated and inherited proximal RTA. Ocular abnormalities in this disorder include band keratopathy, glaucoma and cataracts.1 NBC - 1 gene and SLCA - 4 gene are found out to be associated with this disease. Loss - of - function mutations in the gene that codifies the NBC - 1, the *SLC4A4* gene, were first identified in two Japanese subjects with proximal RTA associated with cataracts, glaucoma and band keratopathy.2 SLC4A4 gene mutation is characteristically associated with isolated proximal renal tubular acidosis cases with ocular abnormalities.3, 4 Treatment is based on correcting the acidosis, symptomatic measures and when possible, treating the underlying cause. The percentage of patients with renal tubular acidosis presenting with ocular features varies, but is relatively uncommon.

2. Case Description

A 11 - year - old female child was referred to the Ophthalmology department. She was diagnosed with proximal renal tubular acidosis and presented with short stature, delay in developmental milestones, dental enamel defects (Figure 1a) and bilateral ocular disease (cataract, raised IOP and band keratopathy). A comprehensive ophthalmic examination was performed, and detailed ocular and systemic medical histories were taken to understand the full clinical spectrum.

Child was born out of a consanguineous marriage at term via LSCS with a weight of 2.5kgs. Immunization history was present till one and a half years with no relevant antenatal history or NICU admission. BCG scar was present.

Developmental delay was noted in communication, writing/reading and gross motor skills. Child's height was 90cm and weight 23kg. History of similar illness in a sibling (13 - year - old male) was noted. There was no associated history of any drug intake.

Laboratory investigations indicated a urine pH of 5.5. Urine analysis showed presence of glucose and protein. Arterial blood gas revealed a normal anion gap metabolic acidosis with low potassium levels (Table 1a).

Complete blood picture and blood culture were done, which were found to be within normal limits (Table1b). Abdominal ultrasound was done which was unremarkable for any findings. CT brain showed calcifications in bilateral globus pallidus (Figure 1b).

Figure 1 (a): Diagnostic image showing dental enamel defects.

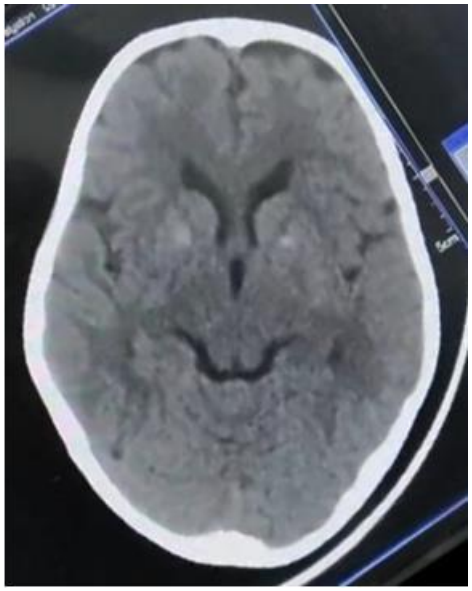

Figure 1 (b): CT Brain showing calcifications in bilateral globus pallidus

Table 1 (a): Lab investigations

Blood gas values	
pH	7.132
pCO_2	17.9 mmHg
$cHCO_3^-$ (P, st) $_c$	8.3 mmol/L
$cHCO_3^-$ (P) $_c$	5.7 mmol/L
pO_2	67.8 mmHg
Oximetry values	
Hct_c	26.60%
ctHb	8.6 g/dL
sO_2	90.80%
Electrolyte values	
cK^+	2.9 mmol/L
cNa^+	142 mmol/L
cCa^{2+}	1.02 mmol/L
cCl^-	128 mmol/L

Table 1 (b): Complete blood picture and culture

Hemoglobin	10 g%
Platelet count	1.56 lakh/cumm
WBC	8600 cells/cumm
Differential count	
Eosinophils	4%
Lymphocytes	28%
Neutrophils	68%
Blood culture and sensitivity	No growth

Complete ocular examination was done. Best corrected visual acuity of child was perception of light in both eyes. Corneal diameter was measured and found to be within normal limits (10.2mm). Intra ocular pressure was checked by digital palpation and found to be hard. Patient underwent a complete slit lamp examination and was found to have band shaped keratopathy in both eyes which was more in left eye (figure 2). A cataractous lens was found in right eye. Lens details were not visible in left eye. Fundus examination was limited due to lens and corneal opacities in both eyes. B scan showed vitreous degenerations in both eyes. Child was given topical lubricants and advised to follow up regularly. Keratoplasty with cataract surgery was recommended after explaining extremely poor prognosis and high chances for recurrence.

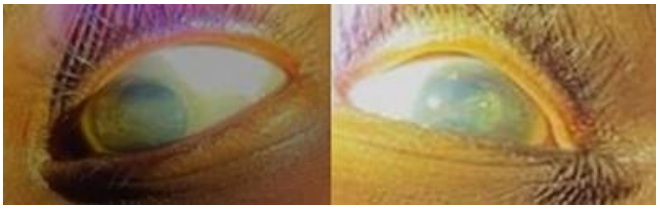

Figure 2: Left eye and right eye showing band shaped keratopathy

3. Discussion

Proximal renal tubular acidosis, a disorder characterized by the inability of proximal renal tubules to reabsorb bicarbonate, is often associated with various ocular findings. This defect leads to bicarbonate wastage and results in normal anion gap metabolic acidosis. This condition can be inherited as an autosomal recessive disorder associated with mutations in the sodium - bicarbonate cotransporter gene.

In this case, a hyperchloremic normal anion gap metabolic acidosis with a urinary pH of 5.5 associated with ocular abnormalities such as band shaped keratopathy, cataract, growth retardation, dental enamel defects and calcifications in bilateral globus pallidus was consistent with a diagnosis of proximal renal tubular acidosis. History of similar illness in the family and consanguineous marriage pointed towards an inherited pattern of disease.

The ocular manifestations of proximal renal tubular acidosis are diverse and can affect multiple structures within the eye. In this patient, slit - lamp examination revealed bilateral band keratopathy, similar to previous case reports on proximal renal tubular acidosis with ocular involvement.[4, 5] In addition to band keratopathy, patient had cataract. There is a potential association between proximal renal tubular acidosis and cataract formation, possibly due to metabolic disturbances or electrolyte imbalances affecting lens transparency and integrity. Further research is needed to study the exact mechanisms and management strategies for cataract in this disorder.

Prognosis after surgery is extremely poor in this case as the ocular abnormalities are at an advanced stage and it point towards the importance of early and regular ophthalmologic examination in cases of tubular acidosis

4. Conclusion

This case report presented a patient with ocular findings such as band keratopathy and cataract in a case of proximal renal tubular acidosis. Band keratopathy can lead to visual disturbances and corneal erosions which signifies the importance of early detection and management of ocular complications in this disorder. There are very few reported cases of renal tubular acidosis with ocular findings. The management of ocular manifestations in patients with proximal renal tubular acidosis involves a multidisciplinary approach, including collaboration between pediatricians, ophthalmologists and nephrologists. Treatment strategies aim to address the underlying metabolic abnormalities, control systemic manifestations and treat ocular symptoms. In this patient, management consisted of symptomatic measures like

alkali therapy to correct metabolic acidosis. Patient was given topical lubricants and advised keratoplasty with cataract surgery after explaining poor prognosis. This case report shows the importance of early and regular ophthalmic examination for better prognosis and treatment in cases of renal tubular acidosis with ocular findings.

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