Abstract Type : Oral presentation
Abstract Submission No.: A-0363
Abstract Topic : Pediatric Nephrology + Inherited Kidney Disease

Unraveling Genetically Proven Renal Tubular Acidosis: A Case Series Highlighting Rickets As A Common Thread.

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Case Study : Renal Tubular Acidosis (RTA) is a group of inherited and acquired tubular disorders characterised by reduced bicarbonate reabsorption or decreased hydrogen ion excretion. Although glomerular function and renal function test are normal, hyperchloremic metabolic acidosis with a normal anion gap is the hallmark. RTA can be broadly divided into three categories (I, II, and IV). Clinical picture differs based on the disease severity and laboratory findings, suggesting either proximal (pRTA) or distal RTA (dRTA). In infants and young children failure to thrive, polyuria, polydipsia, rickets, hypokalemia, and nephrocalcinosis/ nephrolithiasis are common manifestations of primary RTA. Rickets is often overlooked. Misdiagnosis can lead to inappropriate treatment with ineffective agents. Unless there is severe acidosis and/or hypophosphatemia, as in Fanconi syndrome, rickets is more likely in distal RTA and very rarely in proximal RTA. In such situations, genetic testing enables prompt diagnosis and effective treatment leading to clinical and radiological recovery. This case series analyses 11 children with rare forms of RTA, of whom 10 had genetically confirmed diagnosis (4 with dRTA and 6 with pRTA). Of these, 8 children had rickets as the most common clinical presentation with consistent bony abnormalities. Subsequent investigation showed normal anion gap metabolic acidosis, hypokalemia, and hyperchloremia, as well as low serum vitamin D levels. dRTA and pRTA were confirmed by follow-up diagnostic workup. Alkalisation helps to maintain bicarbonate levels in children especially with dRTA along with supportive measures, however genetic analysis is crucial to diagnose the subtype and determine the underlying etiology to offer the targeted treatment.

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Table 1: Genetic mutations and their association with rickets in RTA.

	Gene defect	No.	Rickets
pRTA	SLC2A2	2	+
	CTNS	2	+
	CLC-5	1	+
	NBCel	1	+
dRTA	ATP6V1B1	2	+
	ATP6V0A4	1	-
	WDR72	1	-