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FANCONI SYNDROME SECONDARY TO CYSTINOSIS IN YOUNG MALE: A CASE REPORT

Samina Bibi[1✉], Mian Sameer Ahmed[1], Bakht Babar[1], Tahir Ghaffar[1], Azizul Hasan Aamir[1], Sana Aziz[2]

1: Department of Diabetes and Endocrinology, Medical Teaching Institution Hayatabad Medical Complex, Peshawar, Pakistan
2: Department of Oncology, Medical Teaching Institution Hayatabad Medical Complex, Peshawar, Pakistan
Email✉: saminakhan940@gmail.com

ABSTRACT

INTRODUCTION: Nephropathic cystinosis is a rarely occurring inherited metabolic disorder, leading to Fanconi syndrome, progressive renal failure and a range of extra-renal manifestations including endocrinopathies. There is no definitive cure for Cystinosis. Never the less, early introduction of cysteamine treatment and other supportive measures can adequately retard the occurrence of complications and improve prognosis.

CASE DESCRIPTION: An 18-year-old boy who presented with complaints of abdominal pain, vomiting, polyuria and episodes of periodic paralysis for the last 01 year to the Endocrine Unit, Medical Teaching Institution Hayatabad Medical Complex, and Peshawar, Pakistan. He was lean and wasted with a BMI of 12.9kg/m^2 (weight=32.59kg, height=159cm) having frontal bossing and splaying of bones at wrist joint with normal to low blood pressure. Investigations revealed hypokalemia, alkaline urine and metabolic acidosis on arterial blood gas analysis. Consequently, his symptoms were attributed to underlying renal tubular acidosis. Later, slit lamp examination revealed Cysteine crystals in the cornea confirming the diagnosis of Cystinosis as the underlying cause. He was started on supportive therapy including potassium chloride and citrate supplements, intravenous fluids, calcium and vitamin D supplements and Indomethacin. Senior nephrologist and nutritionist were also taken on board regarding his management. Follow up at 02 months was suggestive of marked symptomatic and biochemical improvement.

CONCLUSION: Data regarding the definitive cure for Cystinosis is scarce. Never the less, early introduction of cysteamine therapy and other supportive measures can adequately retard the occurrence of complications and improvise prognosis.

KEYWORDS: Hypokalemia (MeSH); Acidosis (MeSH); Proteinuria (MeSH); Hypercalciuria (MeSH); Fanconi Syndrome (MeSH); Cystine (MeSH); Cystinosis (MeSH).

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CONFLICT OF INTEREST

Authors declared no conflict of interest

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KMUJ web address: www.kmuj.kmu.edu.pk
Email address: kmuj@kmu.edu.pk