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Original Research Paper

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LIMITED TREATMENT OPTIONS FOR PRIMARY HYPEROXALURIA WITH RENAL INSUFFICIENCY

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ABSTRACT Primary hyperoxaluria (PH) is a rare autosomal recessive metabolic disorder in which the serum levels of oxalate increase because of overproduction.The renal tubule is the primary target of the oxalate deposit, which damages the kidney and leads to cause ESRD. Here, we present a 54-year-old man with a terminal kidney disease; which is dependent on hemodialysis and is likely due to type 2 or 3 PH. Renal insufficiency is uncommon to be found in PH patients. With exceedingly high levels of serum oxalate (70 μmol/L), this patient had few treatment options available for his rare condition.This report details a unique introduction to a rare condition where renal biopsy was instrumental to reach the diagnosis.

KEYWORDS : Hyperoxaluria, Renal insufficiency, Renal Biopsy

BACKGROUND:-

Primary hyperoxaluria type 1, 2 and 3 are rare autosomal recessive disorders that involve errors in glyoxylate and oxalate metabolism [1].In these disorders, serum levels of oxalate increase as a result of over-production.The renal tubule is the primary target of the oxalate deposit, which damages the organ. It is estimated that the incidence of PH is 1 out of 58,000.Type 1 is the most common form and easily found , representing about 80 per cent of cases, while Categories 2 and 3 each represent approximately 10 per cent of cases [2]. Renal failure is a rare occurrence in PH patients. By comparison, there is an increased incidence of secondary hyperoxaluria that leads to ESRD in the setting of gastric bypass surgery and enteric disorders such as chrons disease, ulcerative colitis and short bowel syndrome. Additional cases of enteric free secondary hyperoxaluria are reported in cases of excessive ingestion of vitamin C.We have a 54-year-old man with ESRD because of type 2 or type 3 PH.

CASE:-

The patient was a 54-year-old man who had terminal kidney disease (Cr 4.9) that depended on regular dialysis. The etiology of his renal impairment was not clear with the absence of obvious risk factors. His first symptoms were nausea, vomiting, fever and a weight loss of 20 pounds. There was no history of enteric disorders, malabsorption or vitamin supplement usage. He had serum creatine levels of 4.91 mg/dl with an BUN of 81. Renal biopsy indicated oxalate nephropathy (Fig. 1) with tubular atrophy and interstitial fibrosis (Fig. 2). The patient was genetically tested for AGXT, the mutation observed in PH type 1 [1], and was negative. He is currently on a hypooxalate diet and taking pyridoxin (vitamin B6). However, plasma oxalate concentrations are still elevated at 70 μmol/L (normal range 1.3-3.1 μmol/L) [3]. In spite of great dialysis ampleness and treatment adherence, his interdisciplinary team concluded that his oxalate levels were so high that a kidney transplantation would be likely to have diminished survival.

DISCUSSION:-

In PH, there are insufficiencies of hepatic enzymes causing oxalate overproduction. Oxalate stores within the kidney tubules can lead to nephropathy and conceivable kidney failure. Genetic ponders aid the determination of PH, ordinarily appearing changes within the target qualities AGXT, GPHPR, and HOGA1 for sorts 1, 2, and 3, respectively. Negative testing for PH sort 1 drives us to accept our persistent has PH sort 2 or 3. Diagnosis of PH type 1 would make the patient a potential candidate for combined liver and kidney transplantation. Be that as it may, treatment for those with end-stage renal illness for PH type 2 and 3 is vague. Renal transplantation was assessed in spite of the fact that results are destitute [4]. Because of this, advanced hereditary testing for types 2 and 3 was conceded since it would not affect clinical decision-making. His care team is investigating the alternative of an extended donor kidney transplantation and usually, as of now, beneath evaluation. There's a lack of data within the literature to direct our clinical decision-making in this setting. The proportion of liver transplantation in expansion to kidney transplantation remains ambiguous. In any case, it is thought this may be curative, since GRHPR enzymatic movement within the liver is high [5], and recent case reports are promising [6]. This case presents a one of a kind demonstrative challenge where kidney biopsy was instrumental.

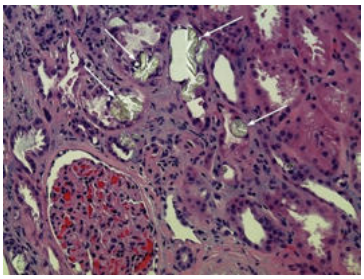

Figure1. Kidney biopsy specimen discoveries at 400× amplification with hematoxylin and eosin stain appearing oxalate deposition (arrows) and tubular decay

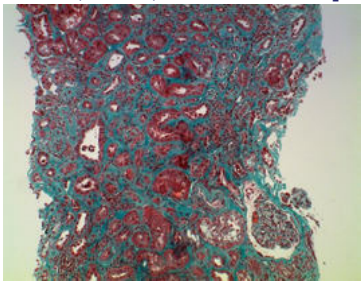

Figure2.Kidneybiopsy specimen discoveries at 400×amplification with trichome recolor illustrating diffuse

fibrosis.

Conflict of Interest Statement:-
The authors have no conflicts of interest to declare.
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