INBORN ERRORS OF METABOLISM - II

ROLE OF RENAL TRANSPLANTATION IN INBORN ERRORS OF METABOLISM

***Sangeetha G**

Abstract: *An enzyme deficiency or defect in a critical metabolic pathway of the human body causes inborn errors of metabolism. These disorders present with multisystem involvement. Renal manifestations can be in the form of tubular diseases like renal tubular acidosis, renal stone disease, hyperuricemia, proteinuria, glucosuria, acute kidney injury and chronic kidney disease. Necessary investigations for the renal manifestations must be a part of the routine follow-up for these children. Possible causes considered are the accumulation of intermediate toxic substrates, energy deficiency, deposition of crystalline substances and subsequent multi-organ impairment. Specific therapies with dietary modifications depending on the underlying condition, are essential steps in treating inborn errors of metabolism. Transplantation considerations in inborn errors of metabolism have unique challenges, as multisystem involvement is common. Isolated kidney, liver or combined kidney-liver transplantation might be needed depending on the underlying disease.*

Keywords: *Metabolic disorders, Fanconi syndrome, Kidney transplantation, Liver transplantation and combined liver-kidney transplantation.*

Points to Remember

- ***Various renal manifestations are common in children with inborn errors of metabolism.***
- ***Although children with Tyrosinemia often present with Fanconi syndrome, liver transplantation remains the preferred treatment.***
- ***Cysteamine therapy should be continued in children with cystinosis even after kidney transplantation.***
- ***Lumasiran is a specific RNA interference (RNAi) therapy for primary hyperoxaluria***
- ***Metabolic crises should be managed appropriately to prevent acute kidney injury in children with IEM.***
- ***Isolated kidney, liver or combined kidney-liver transplantation might be needed depending on the underlying disease.***

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* Senior Consultant,
Department of Pediatric Nephrology,
Kauvery Hospital, Chennai.
email : sangeethaperungo@gmail.com

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