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## **Integrating Machine Learning to Forecast End-Stage Renal Disease Progression in Pediatric Nephropathic Cystinosis: A Prospective Multi-Center Study**

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**Objectives :** Cystinosis is a rare genetic disorder characterized by the accumulation of cystine in cells, leading to significant kidney damage (nephropathic cystinosis) during early childhood. This study utilizes advanced machine learning (ML) techniques to predict renal outcomes and the progression towards end-stage renal disease (ESRD) in pediatric patients with nephropathic cystinosis. Our aim is to enable early interventions and improve management strategies, thus establishing a new benchmark in personalized care within pediatric nephrology.

**Methods :** We conducted a prospective study involving 200 pediatric patients with nephropathic cystinosis across five pediatric nephrology centers from January 2015 to December 2022. Data on demographics, biochemical markers (serum creatinine, electrolytes, cystine levels), genetic mutations, and treatment adherence were collected. An ML model, integrating random forests and neural networks, was developed to predict renal outcomes. The dataset was split for training (70%), validation (15%), and testing (15%), evaluating the model's effectiveness in predicting ESRD progression within five years using metrics such as accuracy, precision, recall, F1-score, and AUC-ROC.

**Results :** Our model demonstrated exceptional predictive capability, with an AUC-ROC of 0.95 (95% CI: 0.92-0.98), accuracy of 92%, precision of 88%, recall of 93%, and an F1-score of 90.5%. Treatment adherence and initial cystine levels emerged as critical prognostic factors. Low adherence to treatment and higher initial cystine levels were associated with a 4.5-fold increased risk of ESRD within five years. Additionally, the model identified a critical cystine threshold, beyond which the risk of rapid progression significantly increases, facilitating the classification of patients into risk categories for tailored treatment plans.

**Conclusions :** This study showcases machine learning's ability to predict renal outcomes accurately in nephropathic cystinosis, advancing personalized care. The model's precision underscores the need for strict treatment adherence and cystine level monitoring. Future research will validate these findings in larger studies and integrate the model into clinical practices, enhancing disease management.