Opinion

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Chronic Kidney Disease in Adolescence: A Case Study on Genetic Etiology and Management

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Introduction

Chronic Kidney Disease (CKD) in adolescence represents a significant medical challenge due to its long-term impact on growth, development, and overall quality of life. While acquired causes such as glomerulonephritis or urinary tract obstruction are relatively common in younger patients, genetic factors are increasingly recognized as major contributors to pediatric and adolescent CKD. Mutations affecting structural, metabolic, or functional renal proteins can lead to progressive nephron loss and eventual renal failure. Early identification of a genetic etiology is critical, as it not only guides management but also informs family counseling and future reproductive decisions. This case study highlights a teenage patient diagnosed with genetically mediated CKD, emphasizing the diagnostic process, genetic findings, and long-term management strategies aimed at preserving renal function and enhancing life quality [1].

Description

A 15-year-old male presented to the nephrology clinic with complaints of fatigue, decreased appetite, and intermittent swelling of the feet for several months. His past medical history was unremarkable, and there was no record of nephrotoxic drug use or recurrent infections. However, a family history revealed that his father had developed End-Stage Renal Disease (ESRD) in his early thirties. Physical examination showed mild edema and elevated blood pressure. Laboratory investigations revealed elevated serum creatinine (2.3 mg/dL), blood urea nitrogen (38 mg/dL), and mild proteinuria. Ultrasound imaging showed bilaterally small kidneys with increased echogenicity, suggestive of chronic parenchymal damage. Given the clinical findings and strong familial predisposition, a preliminary diagnosis of hereditary kidney disease was considered. Further evaluation with renal function tests indicated a declining glomerular filtration rate, supporting the likelihood of chronic progressive nephropathy [2].

Additional serological workup for autoimmune and infectious causes was unremarkable. Given the strong family history, a genetic panel for hereditary kidney disorders was performed, which identified a mutation in the NPHP1 gene associated with nephron phthisis, a recessively inherited tubulointerstitial kidney disease. The patient was managed conservatively with antihypertensive therapy using angiotensin-converting enzyme inhibitors (ACE inhibitors) to reduce proteinuria and control blood pressure [3].

Dietary modifications, including restricted salt and protein intake, were implemented to minimize renal workload. Regular monitoring of kidney function, electrolytes, and blood pressure was maintained. Psychological counseling and educational support were provided to address the emotional impact of a chronic diagnosis during adolescence. Genetic counseling sessions were arranged for the family to discuss inheritance patterns and potential risks to siblings. Over the next year, the patient's renal function remained stable with careful management and adherence to therapy, delaying progression toward advanced CKD [4,5].

Conclusion

This case emphasizes the crucial role of genetic testing in diagnosing chronic kidney disease in adolescents, particularly when there is a positive family history or atypical clinical presentation. Early identification of a genetic cause allows for personalized treatment planning, risk assessment for relatives, and the possibility of targeted interventions that can slow disease progression. Comprehensive management encompassing pharmacologic therapy, nutritional support, psychological care, and regular follow-up remains essential to improve long-term outcomes. Furthermore, this case highlights the importance of raising awareness among clinicians to consider hereditary kidney diseases in young patients, ensuring timely intervention and optimized care throughout adolescence and into adulthood.

Acknowledgement

None

Conflicts of Interest

None

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