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A Note on Hereditary Kidney Diseases in Adults

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Introduction

Approximately 10% to 15% of kidney disorders are hereditary or linked to genetic factors. Because genetic kidney disorders have no distinctive clinical symptoms and respond poorly to treatment, hereditary kidney illnesses account for around 30% of hospitalised children in the United States with chronic renal failure. Many genes are linked to hereditary kidney disease. In the diagnostic and prenatal diagnosis of hereditary kidney disorders, molecular genetic analysis is crucial. For than 30 years, our group has conducted research on inherited kidney illnesses. Our group's research efforts and major findings in hereditary renal disorders are summarised here. A significant number of hereditary kidney illnesses have a Mendelian inheritance, implying that the encoded proteins are required for renal function maintenance.

The discovery of genes implicated in kidney congenital abnormalities and familial variants of nephrotic syndrome has greatly aided our understanding of renal development and filtration barrier physiology. The characteristic phenotypic and clinical heterogeneity seen in monogenic versions of these illnesses will be the focus of this review. Also highlighted will be the significance of susceptibility genes in renal illnesses with a complicated inheritance.

About the study

For a subgroup of individuals with hereditary kidney disease, the introduction of a novel class of aminoglycosides with improved translational read through of nonsense mutations and lower toxicity offers a new treatment option. The kidney is an appropriate target for this technique because aminoglycosides are taken up and retained at a high intracellular concentration in the kidney. In this study, we look at the possibility of aminoglycoside read through therapy in a variety of hereditary kidney illnesses, as well as the therapeutic window for subclasses of each disease caused by nonsense mutations.

Polycystic kidney disease syndromes manifest in unique, as well as overlapping, clinical and hereditary patterns, and are caused by genetic abnormalities at separate loci. Because molecular diagnostics aren't yet a viable clinical tool for diagnosing most cystic kidney illnesses, doctors must depend on their clinical judgement and experience to identify these individuals.

The purpose of this publication is to discuss the genetic patterns, basic epidemiology, and phenotypic aspects of the most frequent cystic renal illnesses so that practising physicians are more aware of these diseases. Autosomal dominant polycystic kidney disease, autosomal recessive polycystic kidney disease, nephronopthisis-medullary cystic kidney disease complex, Bardet-Biedl syndrome, and oral-facial-digital syndrome type 1 will all be discussed in detail. Approximately 10% of adults and virtually all children who require renal replacement therapy have hereditary kidney disease. Our capacity to undertake genetic diagnostics has increased, and our understanding of renal and syndromic disorders has improved [1-5].

Conclusion

We discuss the genetics of renal illnesses in this article, including common monogenic diseases like polycystic kidney disease, Alport syndrome, and Fabry disease, as well as complicated disorders including kidney and urinary tract congenital abnormalities. We give the nephrologist a broad strategy for dealing with inherited illnesses, including a review of common genetic testing, a guide to genetic counselling, and reproductive choices for at-risk couples, such as prenatal diagnosis or pre-implantation genetic diagnosis

Conflict of Interest

The author declares that there is no conflict of interest associated with this paper.

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