

A Short Note on Kidney Diseases in Effected Patients

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Description

Approximately 10% to 15% of kidney disorders are hereditary or are linked to genetic factors. Hereditary kidney diseases account for approximately 30% of hospitalised children in the United States with chronic renal failure, owing to the lack of distinguishing clinical symptoms and poor treatment response. A number of genes have been linked to hereditary kidney disease. Molecular genetic analysis is critical in the diagnosis and prenatal diagnosis of hereditary kidney disorders. Our group has been studying inherited kidney diseases for over 30 years. The research efforts and major findings of our group in hereditary renal disorders are summarised here.

Mendelian inheritance is found in a significant number of hereditary kidney diseases, implying that the encoded proteins are required for renal function maintenance. The identification of genes involved in kidney congenital abnormalities and familial nephrotic syndrome variants has greatly aided our understanding of renal development and filtration barrier physiology. This review will concentrate on the distinct phenotypic and clinical heterogeneity seen in monogenic versions of these illnesses. The importance of susceptibility genes in renal diseases [1-3] with complicated inheritance will also be highlighted.

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

Polycystic kidney disease syndromes have distinct clinical and hereditary patterns that are caused by genetic abnormalities at different loci. Because molecular diagnostics are not yet a viable clinical tool for diagnosing most cystic kidney diseases, doctors must rely on clinical judgement and experience to identify these people. The goal of this publication is to educate practising physicians about the genetic patterns, basic epidemiology, and phenotypic aspects of the most common cystic renal illnesses. The following conditions will be discussed in depth: autosomal dominant polycystic kidney disease, autosomal recessive polycystic kidney disease, nephronophthisis-medullary

cystic kidney disease complex, Bardet-Biedl syndrome, and oral-facial-digital syndrome type 1.

Hereditary kidney disease affects approximately 10% of adults and virtually all children who require renal replacement therapy. Our ability to perform genetic diagnostics has grown, as has our understanding of renal and syndromic disorders [4,5]. In this article, we discuss the genetics of renal illnesses, including common monogenic diseases such as polycystic kidney disease, Alport syndrome, and Fabry disease, as well as more complicated disorders such as kidney and urinary tract congenital abnormalities. We provide a broad strategy for dealing with inherited illnesses to the nephrologist, including a review of common genetic testing, a guide to genetic counselling, and reproductive options for at-risk couples, such as prenatal diagnosis or pre-implantation genetic diagnosis.

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Conflict of Interest

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

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