Foreword

Genetic Advances in Childhood Nephrological Disorders

Patrick D. Brophy¹

¹ Division of Pediatric Nephrology, Dialysis & Transplantation, Stead Family Department of Pediatrics, Carver College of Medicine, University of Iowa, Iowa City, Iowa, United States

J Pediatr Genet 2016;5:1.

Pediatric nephrology, like other pediatric subspecialties, has a profound base in genetic diseases. A clear majority of pediatric chronic kidney disease and end-stage renal disease takes root in congenital abnormalities of the kidney and urinary tract (CAKUT). The discovery of and diversity in genetic etiologies underlying CAKUT is growing daily. Indeed, there is a growing recognition that rare and ultra-rare diseases, such as the nephrotic syndrome and atypical hemolytic-uremic syndrome, respectively, have foundations in genetic abnormalities. The genetic complexity of renal diseases is clear when considering human traits such as hypertension, which likely involve genetic/epigenetic underpinnings with environmental triggers for phenotypic expression. Almost every pediatrician will encounter patients with genetically determined renal disease at some point in their career. In fact, there is mounting evidence that even relatively common renal conditions such as acute kidney injury have a genetic determinant. After reading this special renal genetics issue, specialists should have a good perspective on how to recognize, diagnose, and initiate management of these patients when present in the pediatric clinic.

The selections that encompass this issue range from the common to the ultra-rare. The advancement and cost reductions of techniques available for diagnosing renal genetic conditions are mind-boggling. This has led to significant and almost daily advances in defining new genetic pathways that underlie renal disease states. The exciting revolution in our understanding of the genetic etiologies of urinary tract infections (UTIs) and vesicoureteral reflux is beginning to have profound impact on day-to-day medical management of these clinically relevant conditions. Indeed, this new information has also led to considerable disagreement on how best

to manage UTIs. The continued and significant contribution developmental biologists and geneticists are making to our understanding of renal development cannot be understated. These groups of scientists are paving the way for improved identification of proteomic and metabolomic biomarkers that will eventually allow us to not only predict renal disease development, but also identify potential pharmacologic pathways that will allow us to target specific renal diseases.

A recurrent theme in this issue is the rapidity with which genetic sequencing and technologic breakthroughs have revolutionized gene identification and genome diagnostics in the field of renal genetic diseases. Given the enormous heterogeneity in nearly all forms of CAKUT, this new technology allows us to screen gene panels, all coding DNA, or even whole genomes in single experiments. The evolving commercial genetic services available for diagnostic services will be both a great help for clinicians to provide patients and their relatives with a fast, early, and accurate diagnosis and prognosis, and a potential bane, if adequate interpretation and genetic counseling services are not available. This represents a slippery slope given the vast amounts of “big data informatics” that are being presented to clinicians. Having such data presented in actionable and digestible units should be a strong goal for all of those engaged in these endeavors.

Finally, genetic insights into the underlying causes of renal disorders provide a very solid foundation for the development of targeted and personalized treatments. The movement toward such precision medicine approaches is clear and will hopefully eventually lead to health care cost reductions by reducing the associated morbidity with and the number of investigations and generalized treatment approaches employed in today's medical management of renal disorders.

Address for correspondence
Patrick D. Brophy, MD, MHCDs,
Division of Pediatric Nephrology,
Dialysis & Transplantation, Stead
Family Department of Pediatrics,
Carver College of Medicine,
University of Iowa, 4037 Boyd Tower,
200 Hawkins Dr., Iowa City, IA,
United States
(e-mail: patrick-brophy@uiowa.edu).

Issue Theme Genetic Advances in
Childhood Nephrological
Disorders; Guest Editor: Patrick D.
Brophy, MD, MHCDs

Copyright © 2016 by Georg Thieme
Verlag KG, Stuttgart · New York

DOI <http://dx.doi.org/10.1055/s-0036-1571843>.
ISSN 2146-4596.