KDIGO Controversies Conference on
Autosomal Dominant Tubulointerstitial Kidney Disease (ADTKD)

September 10-11, 2014
Boston, USA

Kidney Disease: Improving Global Outcomes (KDIGO) is an international organization whose mission is to improve the care and outcomes of kidney disease patients worldwide by promoting coordination, collaboration, and integration of initiatives to develop and implement clinical practice guidelines. Periodically, KDIGO hosts conferences on topics of importance to patients with kidney disease. These conferences are designed to review the state of the art on a focused subject and to ask conference participants what needs to be done in this area to improve patient care and outcomes. Sometimes the recommendations from these conferences lead to KDIGO guideline efforts and other times they highlight areas for which additional research is needed to produce evidence that might lead to guidelines in the future.

Background

For decades, ill-defined autosomal dominant kidney diseases have been reported in which disorders originate from tubular cells that lead to tubular atrophy and interstitial fibrosis. Affected family members often display renal fibrosis in the biopsy and exhibit gradually declining renal function, with renal failure usually occurring between the third and sixth decade of life. These diseases are caused by mutations in at least four different genes: $UMOD$, $HNF1B$, $REN$ and as recently described, $MUC1$, but the clinical manifestations of each gene defect are largely indistinguishable.
Variable and inconsistent terminology has been developed to describe these diseases, including medullary cystic kidney disease (MCKD type 1 and 2). Such terminology, however, is misleading since renal cysts are neither a pathognomonic feature nor do they appear to be of pathophysiological relevance for disease progression. These dominantly inherited diseases are currently considered to be rare but the true incidence remains unknown. In fact, given the unspecific manifestation it appears highly likely that most cases have remained unrecognized. There is also robust evidence that variants in the genes causing these monogenic diseases contribute to the polygenic predisposition of other types of chronic kidney disease. Moreover, unraveling the molecular mechanisms of these disorders is likely to provide significant insight into the pathogenesis of tubulointerstitial fibrosis, a hallmark of virtually all types of kidney disease.

**Conference Overview**

The objective of this KDIGO conference is to gather a global panel of clinical and scientific experts who will establish the terminology and nomenclature for this group of diseases and address key issues related to their detection, diagnostic work-up, management and treatment. It is hoped that developing this classification and assessing our current state of knowledge will not only improve the characterization of these disorders but will also facilitate communication between researchers and inform clinicians of the evidence base for present treatment options. This conference will also put forth a summary of the outstanding knowledge gaps and propose a research agenda to better resolve standing controversial issues and help pave the way for future studies in this area.
Drs. Olivier Devuyst (University of Zurich, Switzerland) and Kai-Uwe Eckardt (University of Erlangen- Nürnberg, Germany) will co-chair this conference. The format of the conference will involve topical plenary session presentations followed by focused discussions on key clinical issues as outlined in the Appendix: Scope of Coverage. The conference output will include publication of a position statement that will help guide KDIGO and others on therapeutic management and future research.

APPENDIX: SCOPE OF COVERAGE

1. Definition and nomenclature
2. Diagnostic criteria and work-up
3. Is renal biopsy useful and if so, how to proceed with it?
4. Is urine analysis helpful to raise the suspicion or make the diagnosis?
5. Who should receive genetic testing?
6. How should genetic testing be performed?
7. What is the optimal management?
8. What is the optimal follow-up?