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Genetics in CKD – Where are We and Where are We going? Outcomes from the 2021 KDIGO Controversies Conference on Genetics in CKD

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Immense progress in discovering and understanding the genetic and genomic underpinnings of kidney disease over the past several decades have brought into closer focus the need for consensus and connection globally in this space. Resultantly, the KDIGO Genetics in CKD Controversies Conference in March 2021 was able to bring identify key areas of consensus across the spectrum of monogenic and complex kidney diseases whilst also identifying multiple areas of ongoing controversy, processes for improvement, and critically, priorities for implementation.

Firstly, an ongoing dialogue is indicated in regard to definitions, terminology and nomenclature. Moving to an ontology that embraces two-part names whilst providing better phenotypic and etiological definition is needed.

Secondly, a broader suite of data capture and analysis is indicated. This encompasses phenotyping, genomics, metadata, system & economic data, data sharing infrastructures & frameworks, representative & equitable datasets, and efforts to illuminate disease causal mechanisms.

Lastly, the gap between discovery and implementation remains wide even in spite of major efforts in recent years. Priorities include genomic resources in underrepresented populations, investigation of clinical applications of polygenic scores, core competences for kidney clinicians, development of genetic subspecialty training in nephrology, and a deeper analysis of utility, cost-effectiveness & patient outcomes.

The outcomes and findings of this Controversies Conferences are a key waypoint for the understanding and application of genetics in kidney medicine. Whilst pointing to and celebrating the substantial progress to date, they also now provide opportunity for the global kidney community to move forwards in a collaborative manner that will ultimately result in improved outcomes for patients and families.