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Diagnostic Yield of Kidney Disease Gene Panel Testing During Kidney Transplant Evaluation

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Objectives : Approximately 15% of patients with end-stage kidney disease (ESKD) do not have a primary renal disease diagnosis. Monogenic diseases are estimated to account for 10-15% of the overall prevalence of ESKD in adults. Since knowledge of the underlying kidney disease is crucial for ESKD management in the context of transplantation (KT), we conducted genetic testing using massively parallel sequencing (MPS) to access the diagnostic yield in patients on the KT waitlist.
Methods : A total of 109 patients, waitlisted for KT between January 2023 and December 2023 in a single transplant center, underwent MPS kidney disease gene panel testing, containing 282 genes. We classified the variants according to the classification of the American College of Medical Genetics and Genomics.
Results : Out of the 109 patients on the KT waitlist, 41.3% of patients (n=45) did not have a definite renal diagnosis, while 58.7% (n=64) had a known etiology of ESKD after a thorough clinical evaluation. Patients with a known etiology were subcategorized by their primary causes as hereditary (n=8, all of whom had autosomal dominant polycystic kidney disease) or non-hereditary (n=56). Among the 101 genetically analyzed patients with non-hereditary or without a definite renal diagnosis, 7.9% (n=8) were found to carry pathogenic or likely pathogenic variants. Of these 8 patients, 6 had mutations that constitute the primary cause of their ESKD, and one-third (2 out of 6) were found to carry COL4A4 mutations. In another 35.6% (n=36), we identified variants of unknown significance. MPS gene panel testing reduced the number of patients without a definite renal diagnosis from 45 to 42 and decreased the number in the non-hereditary category from 56 to 53. As a result, the ratio of hereditary cases increased to 12.8% from 7.3%.
Conclusions : MPS gene panel testing has benefits for defining precise renal diagnoses and unraveling the etiology of ESKD.