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Genetic Etiology of Kidney Disease in Korean Bio Big Data

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The clinical landscape of genetic kidney diseases is characterized by profound heterogeneity, where overlapping phenotypes often mask diverse genetic origins. This study utilizes the Korean Bio Big Data Pilot Project to evaluate the diagnostic utility of Whole Genome Sequencing (WGS) in a massive cohort of 9,963 individuals from 4,659 families. While the overall diagnostic yield across the entire project reached 31%, our specific analysis focused on 209 families with renal disorders, achieving a significant diagnostic rate of 24%. The most prevalent clinical presentations within this renal cohort included proteinuria, hematuria, and chronic kidney disease, alongside structural anomalies such as polycystic or multicystic kidney dysplasia and focal segmental glomerulosclerosis. By transitioning from conventional targeted panels—which are often limited by predefined gene sets and delayed updates—to a genome-wide approach, we successfully identified pathogenic variants that encompass single nucleotide variants and complex structural variations. These findings underscore that WGS not only enhances diagnostic yield for common manifestations like glomerular or cystic diseases but also enables the discovery of novel or Korean-specific variants through large-scale genomic comparisons. Furthermore, the integration of a continuous reanalysis framework ensures that previously unsolved cases can be resolved as genomic databases evolve. Ultimately, these results demonstrate that the Korean Bio Big Data initiative serves as a critical infrastructure for precision nephrology, bridging the gap between ambiguous clinical phenotypes and definitive genetic diagnoses to inform personalized management and genetic counseling.

Keywords: Big data, genome sequencing, renal disease