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Establishing a Clinician-Optimized Genetic Panel for Unknown CKD Etiology: Resolving Inter-Institutional Heterogeneity Through Expert Nephrologist Review

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Objectives : Genetic testing for kidney diseases is increasingly utilized in Korea; however, standardized gene panels remain lacking. Consequently, institutions have independently assembled panels—often by aggregating multiple phenotype-specific sub-panels—resulting in substantial variability in gene inclusion. We aimed to quantify inter-institutional heterogeneity in genetic kidney disease panels a clinician-oriented panel optimized for genetic etiology workup in unexplained CKD.
Methods : Gene lists collected from five Korean tertiary-care institutions, four literature-derived sources published in major nephrology journals, and one external commercial testing provider. After harmonizing gene symbols and removing duplicates, genes present in two or more sources but absent from a pre-existing 247-gene institutional panel were selected for expert review. Three nephrologists independently assessed kidney phenotype relevance and clinical applicability, with disagreements resolved by consensus.
Results : Across nine sources, 1,150 unique genes were identified. Panel size varied substantially across sources (117–773 genes per source), indicating marked heterogeneity in gene inclusion. Comparing any two sources, the proportion of genes present in both lists—among all genes present in either list—ranged from 7% to 71%. By recurrence, 771 genes appeared in ≥2 sources, 306 in ≥5, 83 in ≥8, and 28 were shared by all 9 sources. Among 903 candidate genes reviewed, 414 (45.8%) were recommended for inclusion, the others were excluded. The updated panel incorporated genes across diverse kidney disease categories, across CAKUT, cystic kidney disease, glomerular diseases, tubular disorders, thrombotic microangiopathy, and metabolic nephropathy, all with established primary renal phenotypes.
Conclusions : Genetic kidney disease panels in Korea show substantial inter-source heterogeneity in both size and gene content. Applying explicit criteria requiring direct primary renal phenotype, we curated a 661-gene panel from 1,150 candidates. This clinician-optimized resource provides a transparent, replicable framework for genetic diagnosis of unexplained CKD and supports efforts toward national panel standardization.