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Promoting Sustainable Kidney Health: The Asia-Pacific and Beyond

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Navigate Childhood Kidney Disease through Genotyping and Phenotyping

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Abstract (words 250) Background The kidney disease is subject to a wide range of phenotypes, many of which have a significant hereditable component. To delineate the genotype and phenotype spectrum of pediatric kidney disease, a multicenter registration system is currently being conducted based on Chinese Children Genetic Kidney Disease Database (CCGKDD). Methods All consecutive patients with kidney and urological disease were recruited from 2014 to 2020. Genetic analysis through exome sequencing was conducted for the families who had multiple affected individuals with nephropathy or clinical suspicion of a genetic kidney disease owing to early onset or extrarenal features. Results Genetic diagnosis was confirmed in 883 from 2256 (39.1%) patients from 23 provinces in China. The phenotypic profile showed the primary diagnosis including steroid resistant nephrotic syndrome (SRNS, 23.5%), glomerulonephritis (GN, 32.2%), congenital anomalies of the kidney and urinary tract (CAKUT, 21.2%), cystic renal disease (3.9%), renal calcinosis/stone (3.6%), tubulopathy (9.7%), and chronic kidney disease of unknown etiology (CKDu 5.8%). Pathogenic variants of 105 monogenetic disorders were identified. 10 distinct genomic disorders were established as pathogenic copy number variants (CNV) in 11 patients. The diagnostic yield differed by diagnostic subgroup, being highest in those with cystic renal disease (66.3%), followed by tubulopathy (58.4%), GN (57.7%), SRNS (29.2%), renal calcinosis /stone (29.3%), CAKUT (8.6%), and CKDu (43.5%). Reverse phenotyping permitted allowed correct identification in 40 cases with clinical reassessment and unexpected genetic findings. Conclusion Data sharing combined with genotype and phenotype based on national patient registry is pivotal in gaining knowledge on genetic kidney disease.

Keywords: chronic kidney disease (CKD), whole exome sequencing (WES), steroid-

resistant nephrotic syndrome (SRNS), congenital anomalies of the kidney and urinary tract (CAKUT), Nephronophthisis (NPHP)