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Next-generation sequencing identifies causative mutations of chronic kidney diseases

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Chronic kidney diseases (CKD) are the leading cause of end-stage kidney disease (ESKD) and incur a medical cost of more than 317 billion Won per year in Korea. Worldwide the most common diagnostic groups of renal disease that manifest before the age of 25 years are congenital anomalies of the kidneys and urinary tract (CAKUT), steroid-resistant nephrotic syndrome (SRNS), chronic glomerulonephritis and renal cystic ciliopathies, which together encompass >70% of early-onset CKD diagnoses. Recently, over 200 genes, that if mutated cause monogenic forms of these disorders have been identified. Next-generation sequencing (NGS) allows identification of the causative mutation in a high proportion (~20%) of individuals with early onset CKD. Molecular genetic diagnostics in early onset-CKD (before the age of 25 years) will, i) provide patients and families with a molecular genetic diagnosis, ii) generate new insights into diseases mechanisms, iii) allow etiology-based classification of patient cohorts for clinical studies and, iv) may have consequences for personalized treatment and prevention of CKD. Furthermore, the progress in high-throughput sequencing will ensure that additional CKD-causing genes will be detected in the near future.