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Promoting Sustainable Kidney Health: The Asia-Pacific and Beyond

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GWAS Studies in Chronic Diseases

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Genome-wide association studies (GWAS) are powerful tools for identifying genetic variants associated with diseases by scanning genomes of large populations. GWAS have revolutionized our understanding of the genetic basis of chronic diseases, uncovering thousands of associations and providing insights into disease mechanisms and potential therapeutic targets. Recent GWAS have significantly advanced our understanding of chronic diseases such as type 2 diabetes (T2D), hypertension, and chronic kidney disease (CKD). For T2D, large-scale studies have identified more than 600 risk loci, highlighting the roles of beta-cell function and insulin resistance. The latest blood pressure GWAS, encompassing over 1 million individuals, have revealed more than 2,000 variants in genes involved in renal sodium handling and vascular function. CKD GWAS have uncovered variants influencing kidney development, filtration, and injury response, emphasizing pathways related to cell signaling and immune regulation. Polygenic risk scores (PRS) aggregate the effects of many genetic variants to predict an individual's risk of developing a disease. PRS have shown promise in identifying individuals at high risk for T2D, hypertension, and CKD, enabling personalized prevention and early intervention strategies. Our preliminary data demonstrate that individuals with a high PRS for CKD not only have reduced renal function at baseline but are also more likely to experience an accelerated decline in renal function. This underscores the potential of PRS in monitoring kidney health and implementing preemptive measures in high-risk individuals to slow disease progression and improve outcomes. Despite their success, GWAS have limitations, including the inability to detect rare variants with large effects. The identified common variants often explain only a small fraction of the heritability of chronic diseases. There is a growing need for studies focusing on rare variants, which may provide deeper insights into the genetic architecture of chronic diseases and reveal new therapeutic targets.

Keywords: Genome-wide association study, common complex disease, polygenic risk score, precision medicine