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Session Name : Basic Research 1

Session Topic : Basic Research 1

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From Genome to Precision Nephrology: Finding the Answers

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The functional interpretation of genome-wide association studies (GWAS) is challenging due to the cell-type-dependent influences of genetic variants. Here, we generated comprehensive maps of expression quantitative trait loci (eQTLs) for 659 microdissected human kidney samples and identified cell-type-eQTLs by mapping interactions between cell type abundances and genotypes. By partitioning heritability using stratified linkage disequilibrium score regression to integrate GWAS with single-cell RNA sequencing and single-nucleus assay for transposase-accessible chromatin with high-throughput sequencing data, we prioritized proximal tubules for kidney function and endothelial cells and distal tubule segments for blood pressure pathogenesis. Bayesian colocalization analysis nominated more than 200 genes for kidney function and hypertension. Our study clarifies the mechanism of commonly used antihypertensive and renal-protective drugs and identifies drug repurposing opportunities for kidney disease.

Keywords: eQTL, GWAS, Causal gene, Cell type eQTL, CKD