Diagnostic mutations ID'd in chronic kidney disease patients

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(HealthDay)—About one-quarter of adults with chronic kidney disease

(CKD) of unknown cause or familial nephropathy or hypertension have diagnostic mutations, which can be identified with whole-exome sequencing (WES), according to a study published online Dec. 5 in the *Annals of Internal Medicine*.

Sneh Lata, Ph.D., from Columbia University in New York City, and colleagues examined the diagnostic utility of WES in a cohort of 92 adults with CKD of unknown cause or familial nephropathy or hypertension.

The researchers found that WES provided a diagnosis in 24 percent of patients, including nine probands with CKD of unknown cause encompassing 13 distinct genetic disorders. In two probands with tubulointerstitial fibrosis, loss-of-function mutations were identified in *PARN*; these findings extend the phenotype of *PARN* mutations to renal fibrosis. A pathogenic *BRCA2* mutation was identified in a proband who was diagnosed with breast cancer on follow-up. In most solved cases the results affected clinical management, including initiation of targeted surveillance, familial screening to guide selection of a donor for transplantation, and alterations in therapy.

"Whole-exome sequencing identified diagnostic mutations in a substantial number of adults with CKD of many causes," the authors write. "Further study of the utility of WES in the evaluation and care of patients with CKD in additional settings is warranted."

More information: Abstract/Full Text (subscription or payment may be required)

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