



*The following research summary was prepared by ASF's Research Director, B. André Weinstock, PhD.*

Knoers NVAM, van Eerde AM. [The Role of Genetic Testing in Adult CKD](#). *J Am Soc Nephrol*. Online 2024, May 6. DOI: 10.1681/ASN.000000000000401.

Savigne J. [Tips for Testing Adults With Suspected Genetic Kidney Disease](#). *Am J Kidney Dis*. 2024 Jun;83(6):816-824. DOI: 10.1053/j.ajkd.2023.10.011.

Two recent articles were published that discuss the benefits of genetic screening for Alport syndrome in patients with chronic kidney disease (CKD).

In 2018, a consensus was reached to define Alport syndrome based on the presence of a specific genetic mutation or mutations (genotype) rather than its symptoms (phenotype), such as proteinuria, hematuria, glomerular filtration rate, hearing impairment, etc.<sup>1</sup> Because the presence and severity of these symptoms vary so widely among patients and can also be impacted by other factors (like age, diet, medications, blood pressure, etc), it was not a reliable way to classify the disease. As such, the “gold standard” for Alport diagnosis has become genetic testing that specifically looks for variants in the COL4A3, COL4A4, and/or COL4A5 genes.

However, these recent articles report that genetic testing as a diagnostic tool is still significantly underutilized by nephrologists, particularly among nephrologists who treat adult CKD patients. In publishing these findings, the authors seek to educate the broader nephrology community about the significant benefits such testing can offer many patients, including increased understanding of their disease and its impact on their treatment options and future planning.

1. Kashtan CE, Ding J, Garosi G, et al. [Alport syndrome: a unified classification of genetic disorders of collagen IV  \$\alpha\$ 345: a position paper of the Alport Syndrome Classification Working Group](#). *Kidney Int*. 2018 May;93(5):1045-1051. DOI: 10.1016/j.kint.2017.12.018.