The following research summary was prepared by ASF's Volunteer Research Program Chair and Board of Directors member, B. André Weinstock, PhD, MSAS.

Solanki K, Hu Y, Moore B, et al. The Phenotypic Spectrum of COL4A3 Heterozygotes. *KI Reports.* In press 2023, July 25. **DOI**: https://doi.org/10.1016/j.ekir.2023.07.010 (open access).

The Geisinger Health System maintains a database of nearly 175,000 de-identified (made anonymous to protect patient privacy) genomes. From this unique database, Solanki et al. have confirmed that pathogenic COL4A3 variants are a common disease state, affecting about 1 in 100 individuals in the United States.

By the strictest definition, these are autosomal dominant Alport syndrome (ADAS) patients. However, unlike autosomal recessive (ARAS), X-linked (XLAS), and digenic Alport syndrome patients, the risk of kidney failure via primary glomerulosclerosis was not apparent until relative old age. Most of the corresponding de-identified health records showed life-long persistent proteinuria and hematuria.

Also, most of the corresponding patients were not correctly identified as having any kidney health risks, and virtually none had a proper diagnosis of ADAS. This in turn means that the majority of these patients are missing the potential benefit of treatment with the standard of care and educational awareness of their increased risk for kidney disease from non-genetic causes.