METHODS AND KITS FOR DETERMINING PREDISPOSITION TO DEVELOP KIDNEY DISEASES (Ramot)

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Many rare kidney disorders exhibit a monogenic, Mendelian pattern of inheritance. Population-based genetic studies have identified many genetic variants associated with an increased risk of developing common kidney diseases. Strongly associated variants have potential clinical uses as predictive markers and may advance our understanding of disease pathogenesis. These principles are elegantly illustrated by a region within chromosome 22q12 that has a strong association with common forms of kidney disease.

Researchers had identified DNA sequence variants in this locus that were highly associated with an increased prevalence of common chronic kidney diseases in people of African ancestry.

Initial research concentrated on MYH9 as the most likely candidate gene; however, population-based whole-genome analysis enabled two independent research teams to discover more strongly associated mutations in the neighboring APOL1 gene. The powerful evolutionary selection pressure of an infectious pathogen in West Africa favored the spread of APOL1 variants that protect against a lethal form of African sleeping sickness but are highly associated with an increased risk of kidney disease.

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Additional information can be provided upon request.

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