

# A Simple Genetic Test for Kidney Disease

Published date: Feb. 1, 2012

## Technology description

### Summary

This technology relates to methods of diagnosing a predisposition to diseases that cause chronic kidney disease (CKD) and end-stage kidney disease (ESKD). Variations in a gene, non-muscle myosin IIA (MYH9), are associated with 79% of the risk of focal segmental glomerulosclerosis (FSGS), the disease that causes ESKD, in African Americans with HIV, and 56% of African Americans as a whole. The variants are also associated with a 2-3 fold increased risk for end stage kidney disease (ESKD) associated with hypertension. The variations are also present among European Americans, however they are less common.

A simple genetic screening test has been developed that identifies single nucleotide polymorphisms (SNP) and haplotypes in the non-muscle myosin gene MYH9. These variants confer genetic risk for the following kidney diseases: FSGS, collapsing glomerulopathy, HIV-associated nephropathy, hypertensive kidney disease, sickle cell nephropathy, lupus nephropathy, and possibly other kidney diseases.

#### Market:

An estimated 26 millions have CKD, with impaired glomerular filtration rate and approximately 100,000 individuals in the United States develop ESKD every year. The lifetime risk for ESKD is 7.5% of individuals of African American descent and 2.1% for individuals of European descent. Early identification of individuals with MYH9 variants who are at increased risk for CKD might substantially reduce morbidity and mortality in this population, as impaired kidney function is associated with death from cardiovascular disease even in patients who do not progress to ESKD.

## Application area

Facilitate rigorous population (i.e. all individuals) screening for early kidney disease.

Screen individuals with hypertension, to identify individual who might benefit from more intensive therapy.

Screen kidney donors for MYH9 risk alleles to improve renal allograft survival.

Screen patients with sickle cell disease to identify those at increased risk for CKD.

Screen patients with lupus nephritis to identify those at increase risk for CKD.

Screen patients with HIV-1 infection to identify those at increased risk for kidney disease.

Screen patient with other kidney diseases, including idiopathic and secondary kidney disease, where MYH9 mutations may alter the propensity to develop kidney disease or the rate of progressive renal function decline.

Pharmaceutical agents might be developed that reverse the susceptibility phenotype, reducing propensity to CKD. These agents might alter non-muscle myosin IIA function or its interactions with critical molecular partners.

## Institution

[NIH - National Institutes of Health](#)

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