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# Persons of African and Hispanic heritage at higher risk of chronic kidney disease

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Chronic kidney disease affects millions in North America, with persons of African heritage being at a four-fold higher risk and those of Hispanic heritage having a two-fold higher risk compared to the rest of the population. An international study carried out by Dr. Karl Skorecki, from the Technion-Israel Institute of Technology and Rambam Medical Center in Haifa, and his team points to the APOL1 gene as involved in the increased risk of kidney disease in this high-risk population. The results are to be published online in Springer's journal *Human Genetics*.

Excitement has been growing in the scientific community worldwide, with an intense race to determine the genetic link responsible for the greatly increased risk many people of African heritage face for end stage kidney disease and the need for dialysis or transplantation. Identifying the correct gene that puts people at risk for progressive kidney disease is necessary to understand the underlying reason for the increased risk, and to be able to find strategies to prevent or slow down kidney failure.

Research studies in the past two years have focused on a particular gene, called MYH9, as being responsible for these population disparities, but no mutations could be identified which might account for the connection.

This led the authors of the study to look beyond MYH9, by computerized data mining of the recently released 1000 Genomes Project dataset. This data mining, combined with the results obtained from DNA analysis in 955 African Americans and Hispanic Americans

and 676 individuals from twelve populations residing in Africa, led the authors to specific genetic variations in the neighboring APOL1 gene as responsible for the greatly increased risk for kidney disease in persons of western African heritage.

A key element in identifying mutations in the APOL1 gene associated with kidney failure was the absence of these mutations in the 306 Ethiopian individuals included in the DNA analysis. Several of the authors of the study had already reported that Ethiopians are actually relatively protected from kidney disease and correspondingly do not have the mutations identified as associated with risk for kidney failure.

The authors conclude: "In addition to accounting for the risk previously attributed to MYH9 in persons of African heritage, these findings now set the stage for a new area of research relating the APOL family of genes to kidney disease more generally, and for discovery of numerous additional less frequent mutations in the APOL genes that might also perturb or modify kidney function in many population groups. The challenge in future studies will be to broaden the epidemiological, biological and medical relationship between variation in the APOL1 gene and the risk for an extended spectrum of kidney disease."

**More information:** Tzur S et al. (2010) Missense mutations in the APOL1 gene are highly associated with end stage kidney disease risk previously attributed to the MYH9 gene. Human Genetics. DOI 10.1007/s00439-010-0861-0

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