MedicalXpress

Studies identify genetic links to kidney disease, kidney failure

June 26 2010

Researchers at the Translational Genomics Research Institute (TGen) will make two presentations at this week's 70th Scientific Sessions of the American Diabetes Association, June 25-29, in Orlando, Florida.

One presentation describes a DNA study of American Indians in Arizona, in which a TGen-led team discovered a genetic biomarker with a significant association to kidney failure.

The study showed "the strongest evidence" for association with End Stage Renal Disease (ESRD), or kidney failure, in marker rs13315275, and also found evidence of some lesser associations between ESRD and four other markers.

"This study could someday lead to better treatment options for those patients suffering from diabetic kidney disease," said Dr. Johanna DiStefano, Director of TGen's Diabetes, Cardiovascular and Metabolic Diseases Division, and lead author of the study's abstract. "We are conducting ongoing studies to further investigate these markers, and potentially what they might mean for the development of new therapeutics."

All five biomarkers are genetic variants of the gene SUCNR1, which is located in a chromosomal region of the human genome identified as 3q24-q27. This region has been linked to diabetic nephropathy (DM), or diabetic kidney disease, in previous studies.

SUCNR1 is a receptor gene that acts on succinate in the kidneys to mediate the rennin-angiotensin system (RAS), a hormone system that helps control the body's blood pressure and fluid balance. High blood pressure can damage the heart, kidneys and exacerbate the harmful effects of diabetes.

Past studies have shown that diabetes is relatively high among Arizona's Native American communities.

In another TGen study presented Friday, June 25, at the ADA sessions, researchers initiated an investigation into the ways that the PVT1 gene impacts development of diabetic kidney disease.

Previous studies have shown an association between PVT1 and kidney failure in patients with diabetes, both autoimmune (type 1) and that most commonly caused by excessive weight, poor diet and lack of exercise (type 2).

In the new study, researchers found that PVT1 was expressed in mesangial cells, which are specialized cells around blood vessels in the kidneys, at a rate up to five times higher in conditions of high glucose (high blood sugar), compared to normal glucose levels. High blood sugar is a signature symptom of diabetes.

"These findings show that additional study of the role of PVT1 in diabetic kidney disease is well-justified," said Dr. Lucrecia Alvarez, a TGen Post-Doctoral Fellow and the first author of the study's abstract. Dr. Alvarez announced the findings during an oral presentation at the ADA sessions.

Provided by The Translational Genomics Research Institute