Researchers propose new ultrasound screening criteria for diagnosing polycystic kidney disease

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Modification of the current screening criteria are needed for diagnosing patients with autosomal dominant polycystic disease (ADPKD), according to a study appearing in the January 2009 issue of the *Journal of the American Society Nephrology* (*JASN*). The results suggest that some patients with a milder form of the disease may otherwise be misdiagnosed.

ADPKD, a genetic disorder characterized by the growth of numerous cysts in the kidneys, can lead to reduced kidney function and kidney failure. It is the most common inherited disorder of the kidney, occurring in approximately 1 in 500 births, and symptoms usually develop between the ages of 30 and 40 years.

Individuals at risk for developing ADPKD are commonly screened by imaging techniques such as ultrasound. Diagnosis can also be made with a genetic test that detects mutations in the two genes that cause the disease, called PKD1 and PKD2. However, gene-based diagnosis is expensive and detects definitive mutations in only 41% to 63% of cases. In many clinical settings, it is rarely performed.

Testing for ADPKD in individuals whose families have a history of the disease is important. Young people who know they are affected may be able to better preserve their kidney function through diet, life style modification and blood pressure control. Testing also can be used to

determine whether an at-risk individual can safely donate a kidney to a family member with the disease.

Ultrasound screening for ADPKD is based on diagnostic criteria developed to detect cases caused by mutations in the PKD1 gene. This form of the disease is more common and more severe than the form caused by mutations in the PKD2 gene. Therefore, there may be a need for different diagnostic criteria for patients with the milder, less common form of autosomal dominant polycystic disease.

To test whether this is the case, York Pei, MD, of the University of Toronto, in Ontario, Canada, and his colleagues performed kidney ultrasounds and genetic tests on 577 and 371 at-risk individuals from 58 PKD1 and 39 PKD2 families, respectively.

The researchers found that the ultrasound diagnostic criteria currently in use may misdiagnose individuals with mutations in the PKD2 gene. Therefore, the investigators designed new ultrasound criteria that could accurately detect the disease in individuals with mutations in either PKD1 or PKD2 gene. They determined that in families of unknown gene type, the presence of three or more kidney cysts is sufficient for establishing the diagnosis in individuals aged 15 to 39 years, two or more cysts in each kidney is sufficient for individuals aged 40 to 59 years, and four or more cysts in each kidney is required for subjects ≥60 years. Also, fewer than two kidney cysts in at-risk individuals aged ≥40 years is sufficient to exclude the disease. For at-risk individuals aged 30 to 39 years, the absence of any kidney cysts provides almost certainty that the disease is not present.

"For the first time we have derived a set of highly predictive criteria for ultrasound diagnosis of ADPKD that is suitable for test subjects with either gene type," said Pei. "These unified diagnostic criteria will be useful for genetic counseling and for evaluation of at-risk subjects as

living-related kidney donors for their affected relatives," he added.

Source: American Society of Nephrology