Gene mutations predict early, severe form of kidney disease

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The most common kidney disease passed down through families, autosomal dominant polycystic kidney disease (ADPKD) affects one in 400 to 1,000 individuals and is characterized by cysts on the kidneys. The condition slowly gets worse and leads to kidney failure.

Most ADPKD cases arise in adults, but some patients show severe symptoms of the disease in early childhood. New research indicates that these severely affected patients carry, in addition to expected inherited genetic defects, further mutations that aggravate the disease.The study, conducted by Carsten Bergmann, MD (Bioscientia Center for Human Genetics Ingelheim and RWTH Aachen University, in Germany) and his colleagues appears in an upcoming issue of the *Journal of the* American Society Nephrology (JASN), a publication of the American Society of Nephrology..

The researchers studied eight pedigrees in which the severely affected patients were the only family members who had gene mutations other than those that are known to cause ADPKD.

The analysis revealed that additional mutations worsen the disease and contribute to early and severe symptoms.

"Our findings are crucial for understanding polycystic kidney disease, and they help explain why one family member might be profoundly affected while another one only shows mild clinical features," said Dr. Bergmann. He added that similar events may occur in other diseases that

range in severity.

The results may help clinicians as they provide genetic counseling for ADPKD patients, as few know that their future children could develop an early, severe form of the disease. The findings might also help investigators design new therapies for ADPKD.

More information: The article, entitled "Mutations in Multiple PKD Genes May Explain Early and Severe Polycystic Kidney Disease," will appear online on Monday, October 24, 2011, doi: 10.1681/ASN.2010101080

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