ASN
THE AMERICAN SOCIETY OF NEPHROLOGY

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Nov. 1-3, 2002, ASN Media Room, Pennsylvania Convention Center, Room 302,
215-418-2366 (media room), 240-381-7513 (after hours)

IDENTIFICATION OF GENE PROVIDES UNDERSTANDING OF AUTOSOMAL RECESSIVE POLYCYSTIC KIDNEY

Philadelphia, PA (Nov. 1, 2002)— Researchers have identified the disease gene, PKHD1, that causes Autosomal Recessive Polycystic Kidney Disease (ARPKD), a severe form of Polycystic Kidney Disease (PKD). The findings were announced at American Society of Nephrology's (ASN) 35th Annual Renal Week Meeting in Philadelphia, PA.

Polycystic Kidney Disease is the most common life-threatening genetic disease and affects 600,000 children and adults in the United States and 12.5 million people worldwide. It is a condition in which a number of cysts grow and multiply in a patient's kidneys, causing the kidney mass to increase. The diseased kidneys may eventually stop functioning, causing end-stage renal disease and requiring dialysis and transplantation.

The most common form of PKD in children, Autosomal Recessive Polycystic Kidney Disease affects 1 in 20,000 live births. This disease often leads to death or significant complications, including loss of renal function and the progressive liver disorder, congenital hepatic fibrosis.

"The significance of identifying the ARPKD gene is extraordinary, because the gene will make it possible to predetermine a child's genetic predisposition to PKHD1," said the lead investigator of the Mayo group, Peter C. Harris, PhD.

"The gene's discovery gives us the tools to determine the function of the normal gene and how dysfunction results in the disease. With this insight, we can develop new drug and gene therapies and ultimately prevent future development of ARPKD," said Greg Germino, MD, director of the ARPKD Consortium at Johns Hopkins University.

(MORE)

IDENTIFICATION OF GENE

Seven years ago, scientists narrowed down the location of the disease gene to a region on chromosome 6, beginning the first step in identifying the gene. Earlier this year, two laboratory groups employed independent yet complimentary approaches to identify the PKHD1 gene.

The following poster presentations about the discovery of the ARPKD gene will be presented at 10:00 am on Friday, November 1 in Halls A and B of the Pennsylvania Convention Center: *Identification and Characterization of Mouse Pkhd1* [F-P0255], *Mutation Analysis of the ARPKD Gene* [F-P0222], and *Tissue-Specific Expression Patterns of Pkhd1 during Mouse Development* [F-P0259].

The Basic and Clinical Science Symposium "Polycystic Kidney Disease: New Genes and Mechanisms" will be presented at 10:00 am on Monday, November 4 in Room 105 of the Pennsylvania Convention Center.

As the largest nephrology meeting of its kind, Renal Week 2002 is expected to draw more than 11,000 nephrologists to reveal the latest findings in renal research and in the care of patients with kidney and related disorders. The ASN is a not-for-profit organization of 8,700 physicians and scientists dedicated to the study and practice of nephrology and committed to providing a forum for the promulgation of information regarding the latest research and clinical findings on kidney diseases.

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