



Press Release

Cause of Cystic Kidneys Explained

Freiburg graduate student identifies novel gene responsible for cystic kidney disease in children

Sylvia Hoff, a graduate student from the Spemann Graduate School of Biology and Medicine (SGBM), has identified a new gene that causes cystic kidneys in children and young adults. The work by the PhD student Sylvia Hoff and her international collaboration partners was published in the scientific journal *Nature Genetics*. The research group's results lead to the identification of novel insights into the molecular mechanism underlying NPH, which is a prerequisite for developing pharmacological targets and new therapies for children with nephronophthisis.

Nephronophthisis (NPH) is the most common inherited kidney disease that leads to renal failure in children. The kidneys of affected children develop cysts, and as there is no approved therapy yet, patients need dialysis and renal transplantation. In addition, NPH often affects other organs apart from the kidney, such as the eyes, the liver, or the brain.

The PhD student Sylvia Hoff, together with **Dr. Soeren Lienkamp** of the Nephrology Department at the Freiburg University Medical Center headed by Prof. Gerd Walz, analyzed the function of NPH proteins during early developmental processes. They found that the ANKS6 protein has functions similar to those of some of the known NPH proteins. In collaboration with research groups in France, USA, Denmark, Switzerland, Egypt, the Netherlands, and Germany, they succeeded in identifying mutations in the ANKS6 gene of children with NPH. This confirmed that ANKS6 is a novel NPH-disease gene. The patients suffered from early onset cystic kidney

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■ disease and structural heart abnormalities. Further analysis revealed that ANKS6 also forms a protein network with three other NPH proteins (INVS, NPHP3, and NEK8) at the cilium, a hair-like structure on the surface of many cells. The formation of this network is regulated by the enzyme HIF1AN. This is the first time that the assembly of NPH proteins has been described as a dynamic process. Thus, the finding sheds some light on how the binding of multiple NPH proteins can be regulated. This can serve as a basis for investigating the function of NPH protein groups in kidney cells, which will improve our understanding of the disease on the cellular level.

Publication:

ANKS6 is a central component of a nephronophthisis module linking NEK8 to INVS and NPHP3.

Hoff S, Halbritter J, Epting D, Frank V, Nguyen TM, van Reeuwijk J, Boehlke C, Schell C, Yasunaga T, Helmstädter M, Mergen M, Filhol E, Boldt K, Horn N, Ueffing M, Otto EA, Eisenberger T, Elting MW, van Wijk JA, Bockenhauer D, Sebire NJ, Rittig S, Vyberg M, Ring T, Pohl M, Pape L, Neuhaus TJ, Elshakhs NA, Koon SJ, Harris PC, Grahmmer F, Huber TB, Kuehn EW, Kramer-Zucker A, Bolz HJ, Roepman R, Saunier S, Walz G, Hildebrandt F, Bergmann C, Lienkamp SS.

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