

PKD2 gene

polycystin 2, transient receptor potential cation channel

Normal Function

The *PKD2* gene provides instructions for making a protein called polycystin-2. This protein is found in the kidneys before birth and in many adult tissues. Although its exact function is not well understood, polycystin-2 can be regulated by a larger, somewhat similar protein called polycystin-1.

Polycystin-2 likely functions as a channel spanning the cell membrane of kidney cells. In conjunction with polycystin-1, the channel transports positively charged atoms (ions), particularly calcium ions, into the cell. This influx of calcium ions triggers a cascade of chemical reactions inside the cell that may instruct the cell to undergo certain changes, such as maturing to take on specialized functions. Polycystin-1 and polycystin-2 likely work together to help regulate cell growth and division (proliferation), cell movement (migration), and interactions with other cells.

Polycystin-2 is also active in other parts of the cell, including cellular structures called primary cilia. Primary cilia are tiny, fingerlike projections that line the small tubes where urine is formed (renal tubules). Researchers believe that primary cilia sense the movement of fluid through these tubules, which appears to help maintain the tubules' size and structure. The interaction of polycystin-1 and polycystin-2 in renal tubules promotes the normal development and function of the kidneys.

Health Conditions Related to Genetic Changes

Polycystic kidney disease

More than 75 mutations in the *PKD2* gene have been identified in people with polycystic kidney disease. These mutations are responsible for about 15 percent of all cases of autosomal dominant polycystic kidney disease (ADPKD), which is the most common type of this disorder. Mutations in the *PKD2* gene include changes in single DNA building blocks (base pairs) and deletions or insertions of a small number of base pairs in the gene. Most *PKD2* mutations are predicted to result in the production of an abnormally small, nonfunctional version of the polycystin-2 protein. Although researchers are uncertain how a lack of polycystin-2 leads to the formation of cysts, it likely disrupts the protein's interaction with polycystin-1 and alters signaling within the cell and in primary cilia. As a result, cells lining the renal tubules may grow and divide

abnormally, leading to the growth of numerous cysts characteristic of polycystic kidney disease.

Other Names for This Gene

- APKD2
- Pc-2
- PC2
- PKD2_HUMAN
- PKD4
- polycystic kidney disease 2 (autosomal dominant)
- polycystin-2
- TRPP2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PKD2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5311\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5311[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PKD2%5BTIAB%5D%29+OR+%28polycystic+kidney+disease+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- POLYCYSTIN 2; PKD2 (<https://omim.org/entry/173910>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5311>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PKD2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PKD2[gene]))

References

- Al-Bhalal L, Akhtar M. Molecular basis of autosomal dominant polycystic kidney disease. *Adv Anat Pathol*. 2005 May;12(3):126-33. doi:10.1097/01.pap.

0000163959.29032.1f. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15900113>)

- Bissler JJ, Dixon BP. A mechanistic approach to inherited polycystic kidney disease. *Pediatr Nephrol*. 2005 May;20(5):558-66. doi: 10.1007/s00467-004-1665-z. Epub 2005 Feb 18. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15719257>)
- Boucher C, Sandford R. Autosomal dominant polycystic kidney disease (ADPKD, MIM 173900, PKD1 and PKD2 genes, protein products known as polycystin-1 and polycystin-2). *Eur J Hum Genet*. 2004 May;12(5):347-54. doi:10.1038/sj.ejhg.5201162. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14872199>)
- Grimm DH, Karihaloo A, Cai Y, Somlo S, Cantley LG, Caplan MJ. Polycystin-2 regulates proliferation and branching morphogenesis in kidney epithelial cells. *JBiol Chem*. 2006 Jan 6;281(1):137-44. doi: 10.1074/jbc.M507845200. Epub 2005 Nov 8. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/16278216>)
- Harris PC, Torres VE. Polycystic Kidney Disease, Autosomal Dominant. 2002 Jan 10 [updated 2022 Sep 29]. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. *GeneReviews*(R) [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1246/> Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20301424>)
- Lina F, Satlin LM. Polycystic kidney disease: the cilium as a common pathway in cystogenesis. *Curr Opin Pediatr*. 2004 Apr;16(2):171-6. doi:10.1097/00008480-200404000-00010. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15021197>)
- Nauli SM, Alenghat FJ, Luo Y, Williams E, Vassilev P, Li X, Elia AE, Lu W, Brown EM, Quinn SJ, Ingber DE, Zhou J. Polycystins 1 and 2 mediate mechanosensation in the primary cilium of kidney cells. *Nat Genet*. 2003 Feb;33(2):129-37. doi: 10.1038/ng1076. Epub 2003 Jan 6. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12514735>)
- Ong AC, Harris PC. Molecular pathogenesis of ADPKD: the polycystin complex gets complex. *Kidney Int*. 2005 Apr;67(4):1234-47. doi:10.1111/j.1523-1755.2005.00201.x. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15780076>)
- Wilson PD. Polycystic kidney disease. *N Engl J Med*. 2004 Jan 8;350(2):151-64. doi: 10.1056/NEJMra022161. No abstract available. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14711914>)

Genomic Location

The *PKD2* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

Last updated June 1, 2006