



PKHD1 gene

PKHD1, fibrocystin/polyductin

Normal Function

The *PKHD1* gene provides instructions for making a protein called fibrocystin (sometimes known as polyductin). This protein is present in fetal and adult kidney cells, and is also present at low levels in the liver and pancreas.

Fibrocystin spans the cell membrane of kidney cells, so that one end of the protein remains inside the cell and the other end projects from the outer surface of the cell. Based on its structure, fibrocystin may act as a receptor, interacting with molecules outside the cell and receiving signals that help the cell respond to its environment. This protein also may be involved in connecting cells together (adhesion), keeping cells apart (repulsion), and promoting the growth and division of cells (proliferation).

Fibrocystin is also found in cell 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 play an important role in maintaining the size and structure of these tubules; however, the function of fibrocystin in primary cilia remains unclear.

Health Conditions Related to Genetic Changes

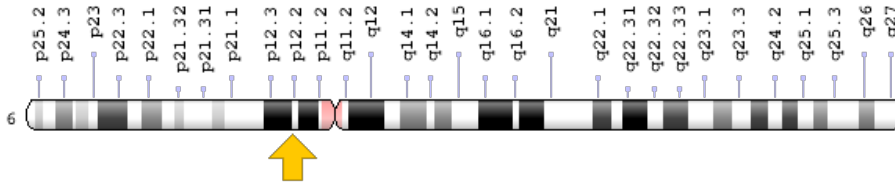
Polycystic kidney disease

More than 270 mutations in the *PKHD1* gene have been identified in people with polycystic kidney disease. These mutations cause autosomal recessive polycystic kidney disease (ARPKD), which is a severe type of the disorder that is usually evident at birth or in early infancy. *PKHD1* mutations include changes in single DNA building blocks (base pairs) and insertions or deletions of a small number of base pairs in the gene. These mutations disrupt the normal structure and function of the fibrocystin protein, or lead to the production of an abnormally small, nonfunctional version of the protein. Researchers have not determined how these genetic changes lead to the formation of numerous cysts characteristic of polycystic kidney disease.

Chromosomal Location

Cytogenetic Location: 6p12.3-p12.2, which is the short (p) arm of chromosome 6 between positions 12.3 and 12.2

Molecular Location: base pairs 51,614,685 to 52,087,625 on chromosome 6 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- FCYT
- fibrocystin
- PKHD1_HUMAN
- polycystic kidney and hepatic disease 1 (autosomal recessive)
- polyductin
- TIGM1
- tigmin

Additional Information & Resources

Clinical Information from GeneReviews

- Polycystic Kidney Disease, Autosomal Recessive
<https://www.ncbi.nlm.nih.gov/books/NBK1326>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PKHD1%5BTIAB%5D%29+OR+%28polycystic+kidney+and+hepatic+disease+1%5BTIAB%5D%29%29+OR+%28%28ARPKD%5BTIAB%5D%29+OR+%28FCYT%5BTIAB%5D%29+OR+%28fibrocystin%5BTIAB%5D%29+OR+%28polyductin%5BTIAB%5D%29+OR+%28TIGM1%5BTIAB%5D%29+OR+%28tigmin%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>

Catalog of Genes and Diseases from OMIM

- PKHD1 GENE
<http://omim.org/entry/606702>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_PKHD1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=PKHD1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:9016
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:5314>
- Mutation Database, Autosomal Recessive Polycystic Kidney Disease (Aachen University, Germany)
<http://www.humgen.rwth-aachen.de/>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/5314>
- UniProt
<https://www.uniprot.org/uniprot/P08F94>

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