

# Class work

# Nucleotide sequence database:




# DNA and RNA

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# https://www.ncbi.nlm.nih.gov/nucleotide/

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Nucleotide

Nucleotide

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## Nucleotide

The Nucleotide database is a collection of sequences from several sources, including GenBank, RefSeq, TPA and PDB. Genome, gene and transcript sequence data provide the foundation for biomedical research and discovery.

### Using Nucleotide

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[RefSeq FTP](#)

### Nucleotide Tools

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### Other Resources

[GenBank Home](#)

[RefSeq Home](#)

[Gene Home](#)

[SRA Home](#)

[INSDC](#)

Where possible, the sequences are annotated so that you can find the strings of sequences that may be functional.

# Example 5: Human PKD1

Search with Human PKD1 Gene.

Gene

Gene

Homo sapiens PKD1

Search

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Gene sources  
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Alternatively spliced  
Annotated genes  
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Tabular 20 per page Sort by Relevance Send to: Hide sidebar >>

GENE

Was this helpful?

PKD1 – polycystin 1, transient receptor potential channel interacting

Homo sapiens (human)

Also known as: PBP, PC1, Pc-1, TRPP1

Gene ID: 5310

RefSeq transcripts (10) RefSeq proteins (10) RefSeqGene (1) PubMed (316)

Orthologs Genome Data Viewer BLAST Download

RefSeq Sequences

Filters: Manage Filters

Results by taxon

Top Organisms Tree

Homo sapiens (102)

Cellulosimicrobium cellulans (4)

Bacteroides nordii CL02T12C05 (3)

Necator americanus (1)

Fonsecaea monophora (1)

All other taxa (11)

More...

Find related data

Database: Select

Find items

Search details

("Homo sapiens"[Organism] OR Homo sapiens[All Fields]) AND PKD1[All Fields]

Search

See more...

Search results

Items: 1 to 20 of 122

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Name/Gene ID	Description	Location	Aliases	MIM
<input type="checkbox"/> PKD1	polycystin 1, transient	Chromosome 16,	PBP, PC1, Pc-1, TRPP1	601313

# Example 5: Human PKD1

How many exons of Human PKD1?

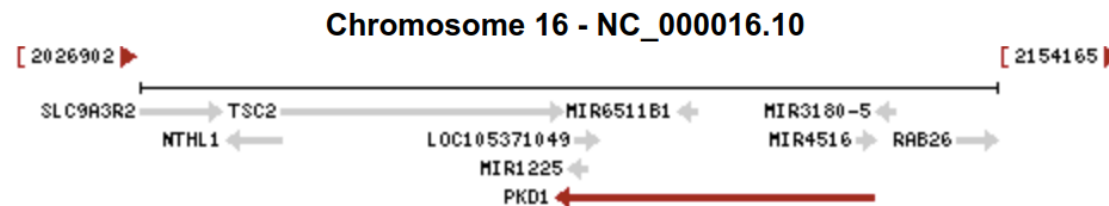
 **Genomic context**  

**Location:** 16p13.3

See PKD1 in [Genome Data Viewer](#)




**Exon count:** 51

Annotation release	Status	Assembly	Chr	Location
109.20210514	current	GRCh38.p13 ( <a href="#">GCF_000001405.39</a> )	16	NC_000016.10 (2088708..2135898, complement)
105.20201022	previous assembly	GRCh37.p13 ( <a href="#">GCF_000001405.25</a> )	16	NC_000016.9 (2138709..2185899, complement)



# Example 5: Human PKD1

How many transcripts of Human PKD1?

 Summary  

<b>Official Symbol</b>	PKD1 <small>provided by <a href="#">HGNC</a></small>
<b>Official Full Name</b>	polycystin 1, transient receptor potential channel interacting <small>provided by <a href="#">HGNC</a></small>
<b>Primary source</b>	<a href="#">HGNC:HGNC:9008</a>
<b>See related</b>	<a href="#">Ensembl:ENSG00000008710</a> <a href="#">MIM:601313</a>
<b>Gene type</b>	protein coding
<b>RefSeq status</b>	REVIEWED
<b>Organism</b>	<a href="#">Homo sapiens</a>
<b>Lineage</b>	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
<b>Also known as</b>	PBP; PC1; Pc-1; TRPP1
<b>Summary</b>	<p>This gene encodes a member of the polycystin protein family. The encoded glycoprotein contains a large N-terminal extracellular region, multiple transmembrane domains and a cytoplasmic C-tail. It is an integral membrane protein that functions as a regulator of calcium permeable cation channels and intracellular calcium homoeostasis. It is also involved in cell-cell/matrix interactions and may modulate G-protein-coupled signal-transduction pathways. It plays a role in renal tubular development, and mutations in this gene cause autosomal dominant polycystic kidney disease type 1 (ADPKD1). ADPKD1 is characterized by the growth of fluid-filled cysts that replace normal renal tissue and result in end-stage renal failure. Splice variants encoding different isoforms have been noted for this gene. Also, six pseudogenes, closely linked in a known duplicated region on chromosome 16p, have been described. [provided by RefSeq, Oct 2008]</p>
<b>Expression</b>	Ubiquitous expression in endometrium (RPKM 11.1), fat (RPKM 9.6) and 25 other tissues <a href="#">See more</a>
<b>Orthologs</b>	<a href="#">mouse</a> <a href="#">all</a>
<b>NEW</b>	Try the new <a href="#">Gene table</a>
	Try the new <a href="#">Transcript table</a>

# Example 5: Human PKD1

How many transcripts of Human PKD1?

NCBI Datasets

NCBI Datasets

Homepage

Genomes

Genes

Coronavirus genomes

Coronavirus proteins

Documentation

Quickstart guides

Command-line tools

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PKD1 Transcripts BETA

PKD1 — polycystin 1, transient receptor potential channel interacting (*Homo sapiens*)

10 transcripts

← SWITCH TO GENE VIEW

EDIT

DOWNLOAD

SELECT COLUMNS

<input type="checkbox"/>	Gene ID	Symbol	Transcript	Length (nt)	Protein	Length (aa)	Protein name	Isoform	Organism
<input type="checkbox"/>	5310	PKD1	NM_000296.4	14137	NP_000287.4	4302	polycystin-1	2 precursor	Homo sapiens
<input type="checkbox"/>	5310	PKD1	NM_001009944.3	14140	NP_001009944.3	4303	polycystin-1	1 precursor	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_024450299.1	13994	XP_024306067.1	4319	polycystin-1	X4	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_024450298.1	14066	XP_024306066.1	4343	polycystin-1	X1	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_024450300.1	14052	XP_024306068.1	4273	polycystin-1	X5	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_011522529.2	14163	XP_011520831.1	4320	polycystin-1	X3	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_011522528.3	14166	XP_011520830.1	4321	polycystin-1	X2	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_024450301.1	12283	XP_024306069.1	3635	polycystin-1	X6	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_005255370.3	11727	XP_005255427.1	3288	polycystin-1	X8	Homo sapiens
<input type="checkbox"/>	5310	PKD1	XM_011522537.2	11098	XP_011520839.1	3329	polycystin-1	X7	Homo sapiens

Rows per page

25

Showing transcripts for 1-1 of 1 Gene

|<

<

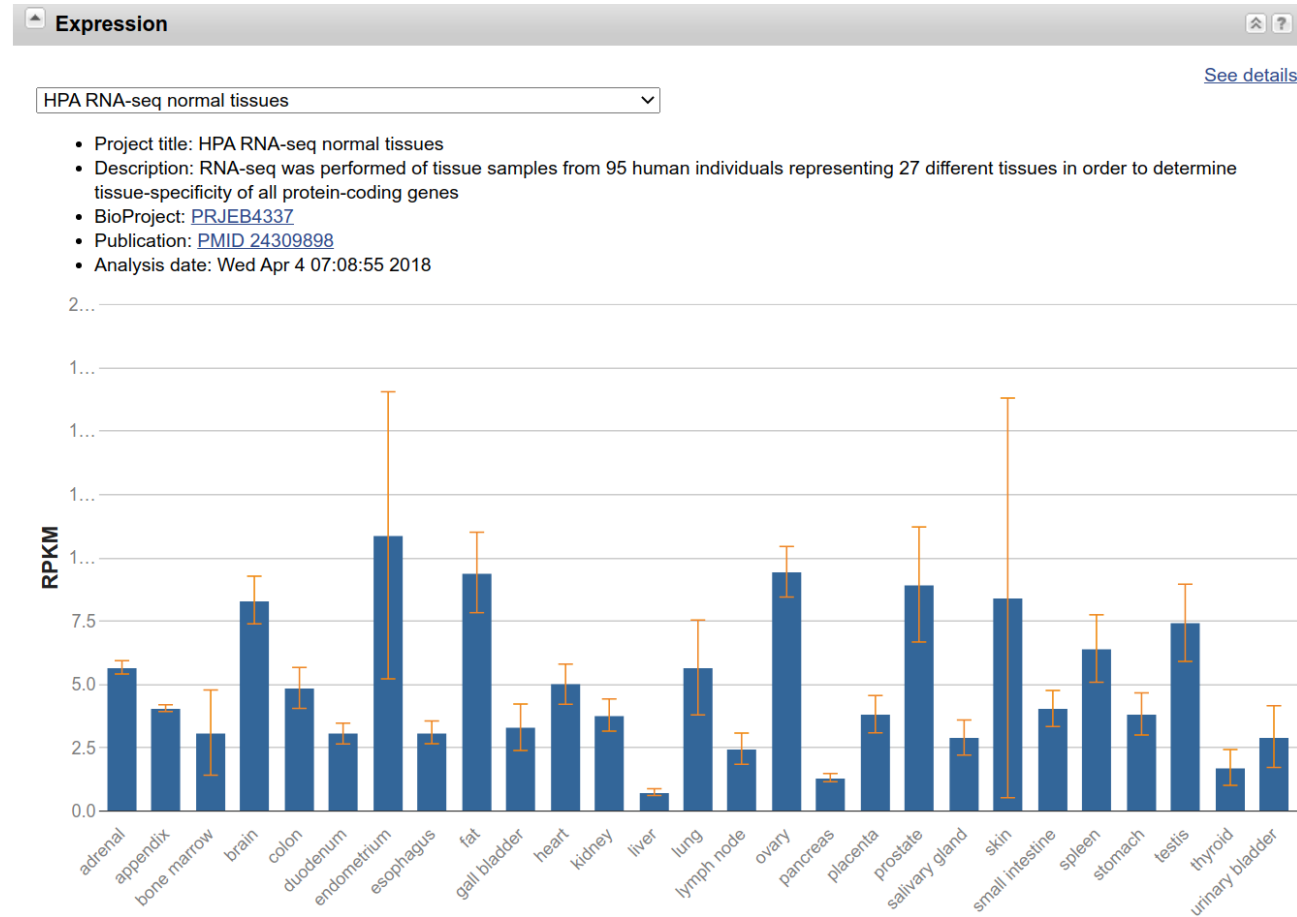
>

>|

<https://www.ncbi.nlm.nih.gov/gene/5310>

# Example 5: Human PKD1

Is Human PKD1 expressed in all the tissues?



# Example 5: Human PKD1

How many pseudogenes of Human PKD1?

**Summary** This gene encodes a member of the polycystin protein family. The encoded glycoprotein contains a large N-terminal extracellular region, multiple transmembrane domains and a cytoplasmic C-tail. It is an integral membrane protein that functions as a regulator of calcium permeable cation channels and intracellular calcium homoeostasis. It is also involved in cell-cell/matrix interactions and may modulate G-protein-coupled signal-transduction pathways. It plays a role in renal tubular development, and mutations in this gene cause autosomal dominant polycystic kidney disease type 1 (ADPKD1). ADPKD1 is characterized by the growth of fluid-filled cysts that replace normal renal tissue and result in end-stage renal failure. Splice variants encoding different isoforms have been noted for this gene. Also, six **pseudogenes**, closely linked in a known duplicated region on chromosome 16p, have been described. [provided by RefSeq, Oct 2008]

**Expression** Ubiquitous expression in endometrium (RPKM 11.1), fat (RPKM 9.6) and 25 other tissues [See more](#)

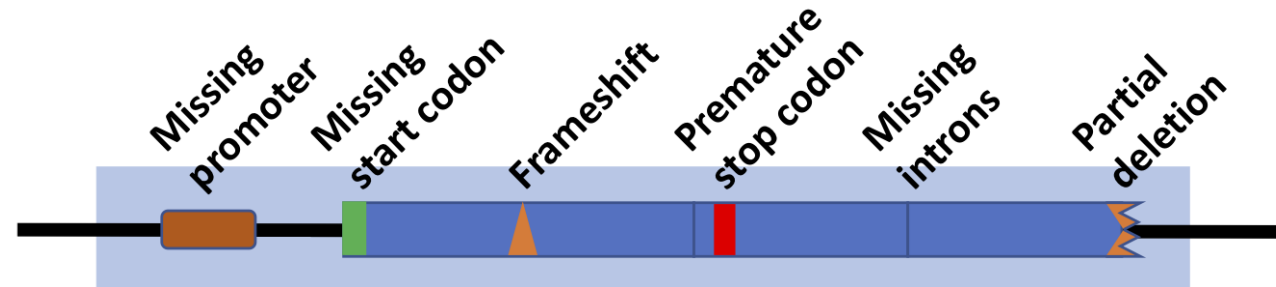


# Example 5: Human PKD1

## What is Pseudogenes?

- Pseudogenes are nonfunctional segments of DNA that resemble functional genes.
- Most arise as superfluous copies of functional genes, either directly by DNA duplication or indirectly by reverse transcription of an mRNA transcript.
- Pseudogenes are usually identified when genome sequence analysis finds gene-like sequences that lack regulatory sequences needed for transcription or translation, or whose coding sequences are obviously defective due to frameshifts or premature stop codons.

### Common defects of pseudogenes:



# Example 5: Human PKD1

How many pseudogenes of Human PKD1?

General gene information

Markers

Related pseudogene(s)  
10 found [Review record\(s\) in Gene](#)

Homology  
[Homologs of the PKD1 gene](#): The PKD1 gene is conserved in dog, cow, mouse, rat, chicken, zebrafish, and frog.  
[Orthologs from Annotation Pipeline](#): 399 organisms have orthologs with human gene PKD1  
[Orthologs](#)

Gene Ontology [Provided by GOA](#)

Function	Evidence Code	Pubs
<a href="#">enables Wnt-activated receptor activity</a>	IDA	<a href="#">PubMed</a>
<a href="#">contributes to calcium channel activity</a>	IDA	<a href="#">PubMed</a>
<a href="#">enables calcium channel activity</a>	ISS	
<a href="#">enables carbohydrate binding</a>	IEA	
<a href="#">enables protein binding</a>	IPI	<a href="#">PubMed</a>
<a href="#">enables protein domain specific binding</a>	IPI	<a href="#">PubMed</a>
<a href="#">enables protein kinase binding</a>	IPI	<a href="#">PubMed</a>

# Example 5: Human PKD1

How many pseudogenes of Human PKD1?

Gene

Gene

related\_functional\_gene\_5310[group]

Search

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Help

Gene sources

Genomic

Categories

Alternatively spliced

Annotated genes

Pseudogene

Sequence content

Ensembl

RefSeq

Status

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Tabular

20 per page

Sort by Relevance

Send to:

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Search results

Items: 10

Showing Current items.

Name/Gene ID	Description	Location	Aliases	MIM
<input type="checkbox"/> <a href="#">PKD1P1</a> ID: 339044	polycystin 1, transient receptor potential channel interacting pseudogene 1 [ <i>Homo sapiens</i> (human)]	Chromosome 16, NC_000016.10 (16317609..16350608)	HG1, NPIP	
<input type="checkbox"/> <a href="#">PKD1P6</a> ID: 353511	polycystin 1, transient receptor potential channel interacting pseudogene 6 [ <i>Homo sapiens</i> (human)]	Chromosome 16, NC_000016.10 (15125139..15154873, complement)	HG6	
<input type="checkbox"/> <a href="#">PKD1P6-NPIPP1</a> ID: 105369154	PKD1P6-NPIPP1 readthrough [ <i>Homo sapiens</i> (human)]	Chromosome 16, NC_000016.10 (15104723..15131601, complement)		

Filters: [Manage Filters](#)

Find related data

Database: Select

Find items

Search details

related\_functional\_gene\_5310[group] AND alive[prop]

Search

See more...

Recent activity

related\_functional\_gene\_5310[group] AND (alive[prop]) (10)

Turn Off Clear

[https://www.ncbi.nlm.nih.gov/gene/?Term=related\\_functional\\_gene\\_5310%5Bgroup%5D](https://www.ncbi.nlm.nih.gov/gene/?Term=related_functional_gene_5310%5Bgroup%5D)