Class work
Nucleotide sequence database: DNA and RNA

23/05/2024

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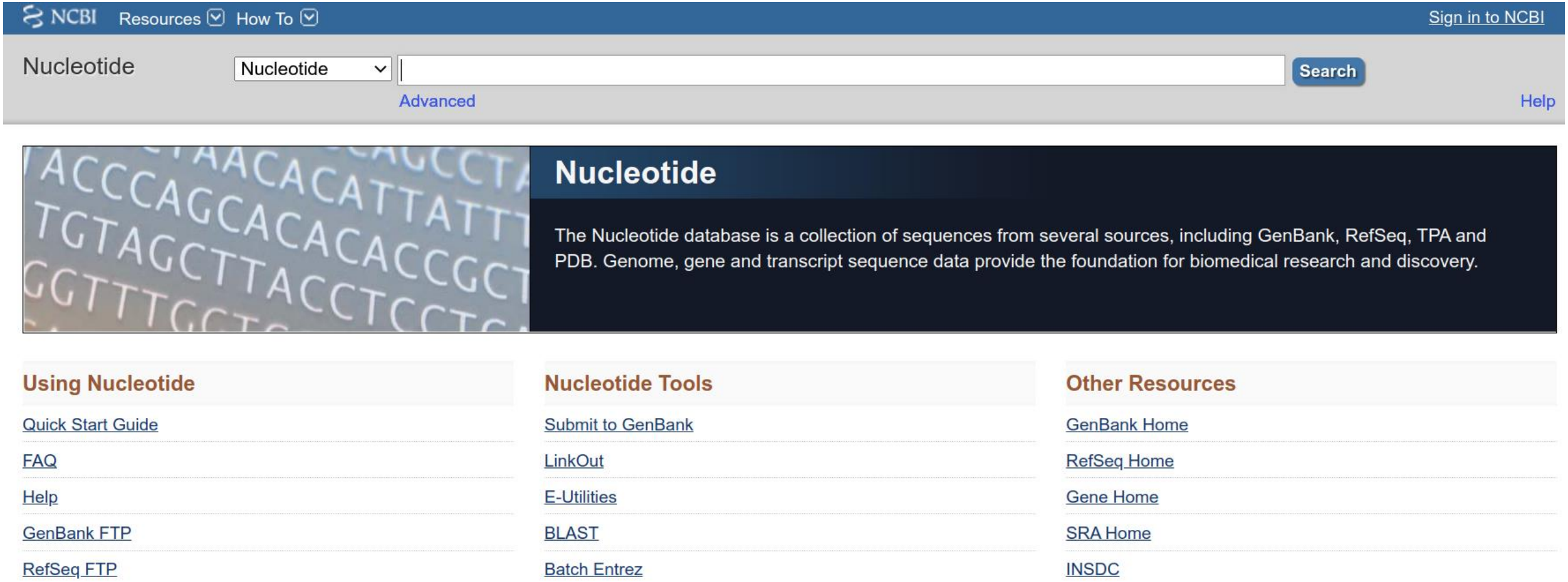
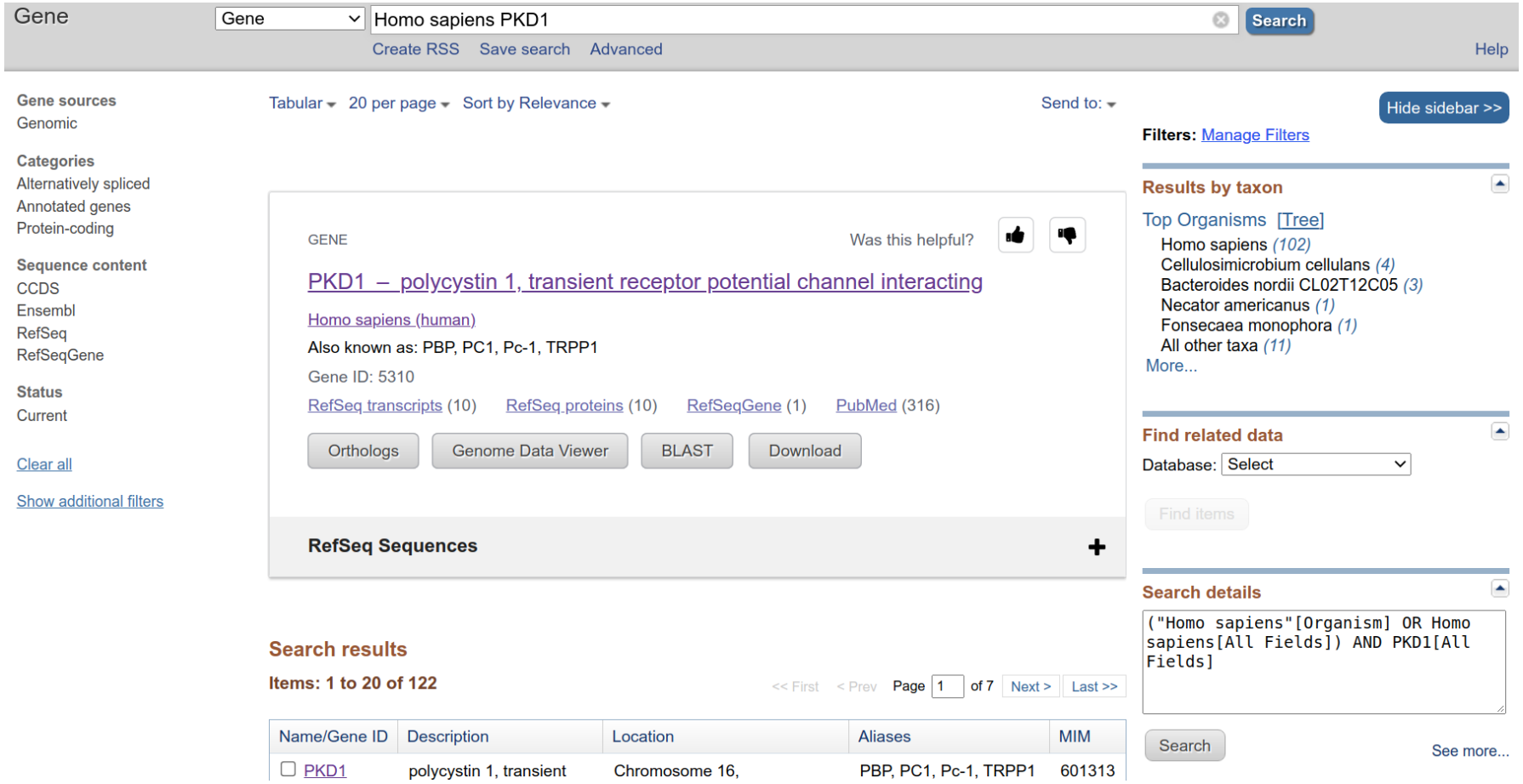
https://www.ncbi.nlm.nih.gov/nucleotide/



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.



Example 5: Human PKD1

How many exons of Human PKD1?

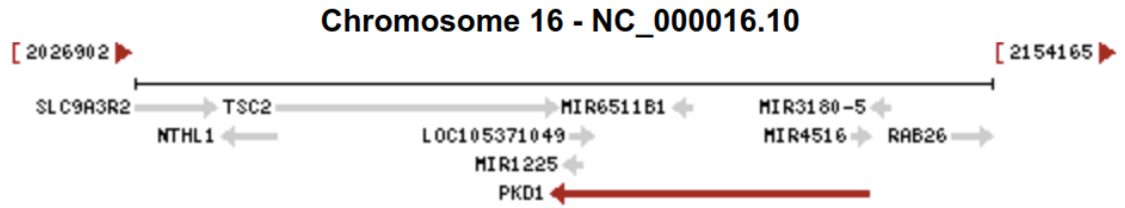
Genomic context

See PKD1 in Genome Data Viewer

Location: 16p13.3

Exon count: 51

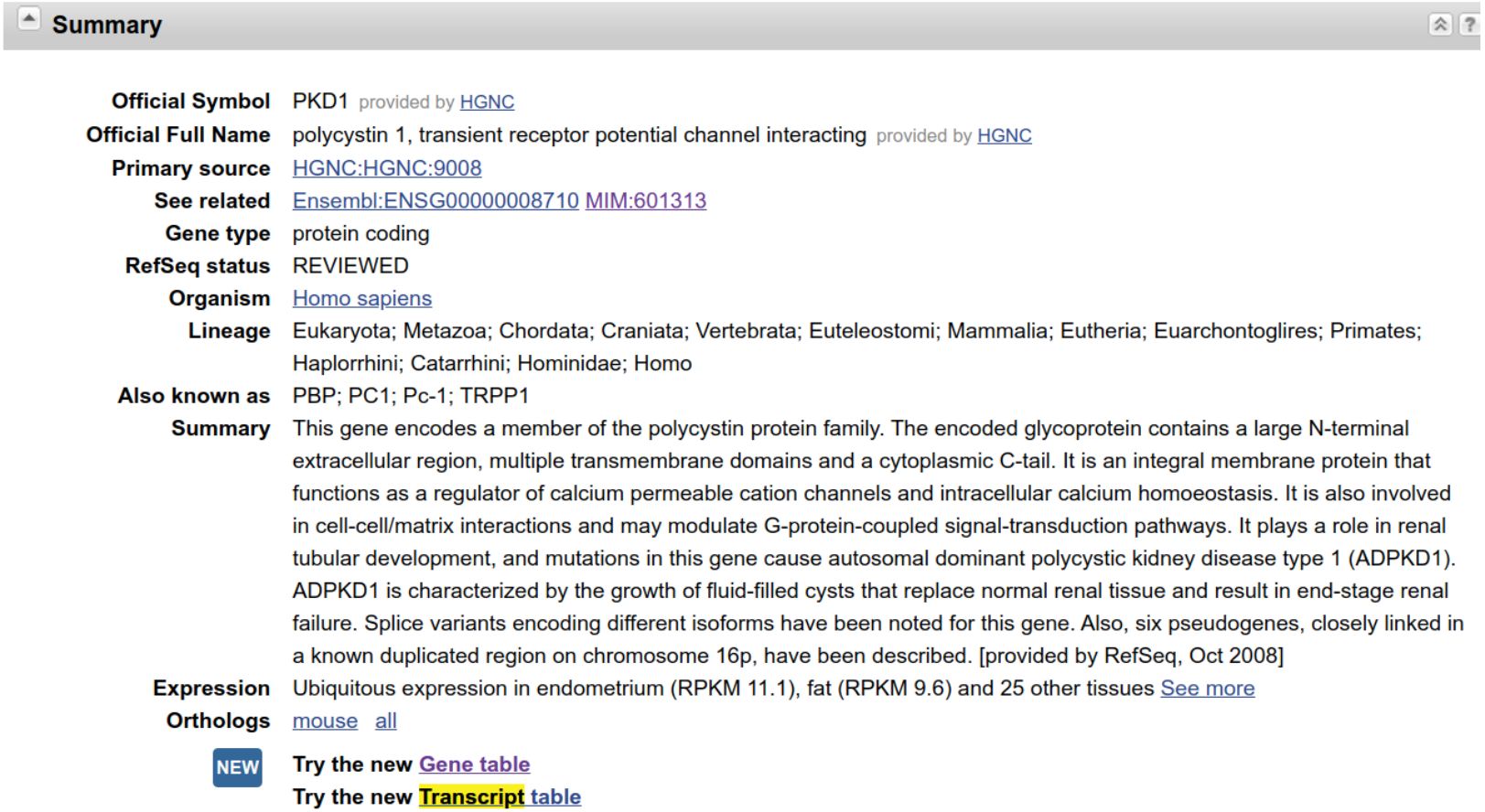
Annotation release	Status	Assembly	Chr	Location
109.20210514	current	GRCh38.p13 (GCF_000001405.39)	16	NC_000016.10 (2088708..2135898, complement)
105.20201022	previous assembly	GRCh37.p13 (GCF_000001405.25)	16	NC_000016.9 (2138709..2185899, complement)



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

Example 5: Human PKD1

How many transcripts of Human PKD1?



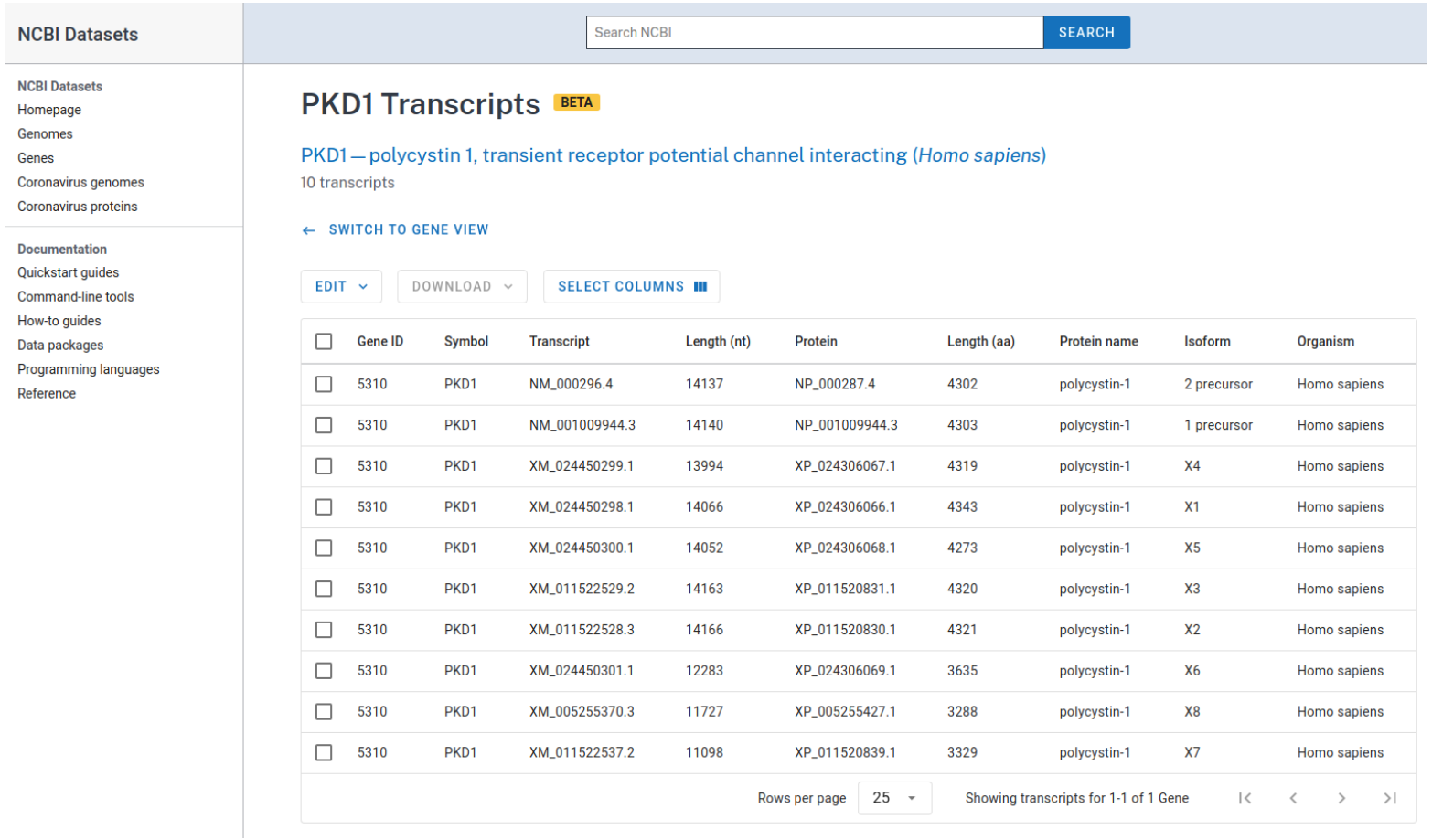
Summary

Official Symbol	PKD1 provided by HGNC
Official Full Name	polycystin 1, transient receptor potential channel interacting provided by HGNC
Primary source	HGNC:HGNC:9008
See related	Ensembl:ENSG00000008710 MIM:601313
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	PBP; PC1; Pc-1; TRPP1
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
Orthologs	mouse all
NEW	**Try the new Gene table**
	Try the new Transcript table

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

Example 5: Human PKD1

How many transcripts of Human PKD1?



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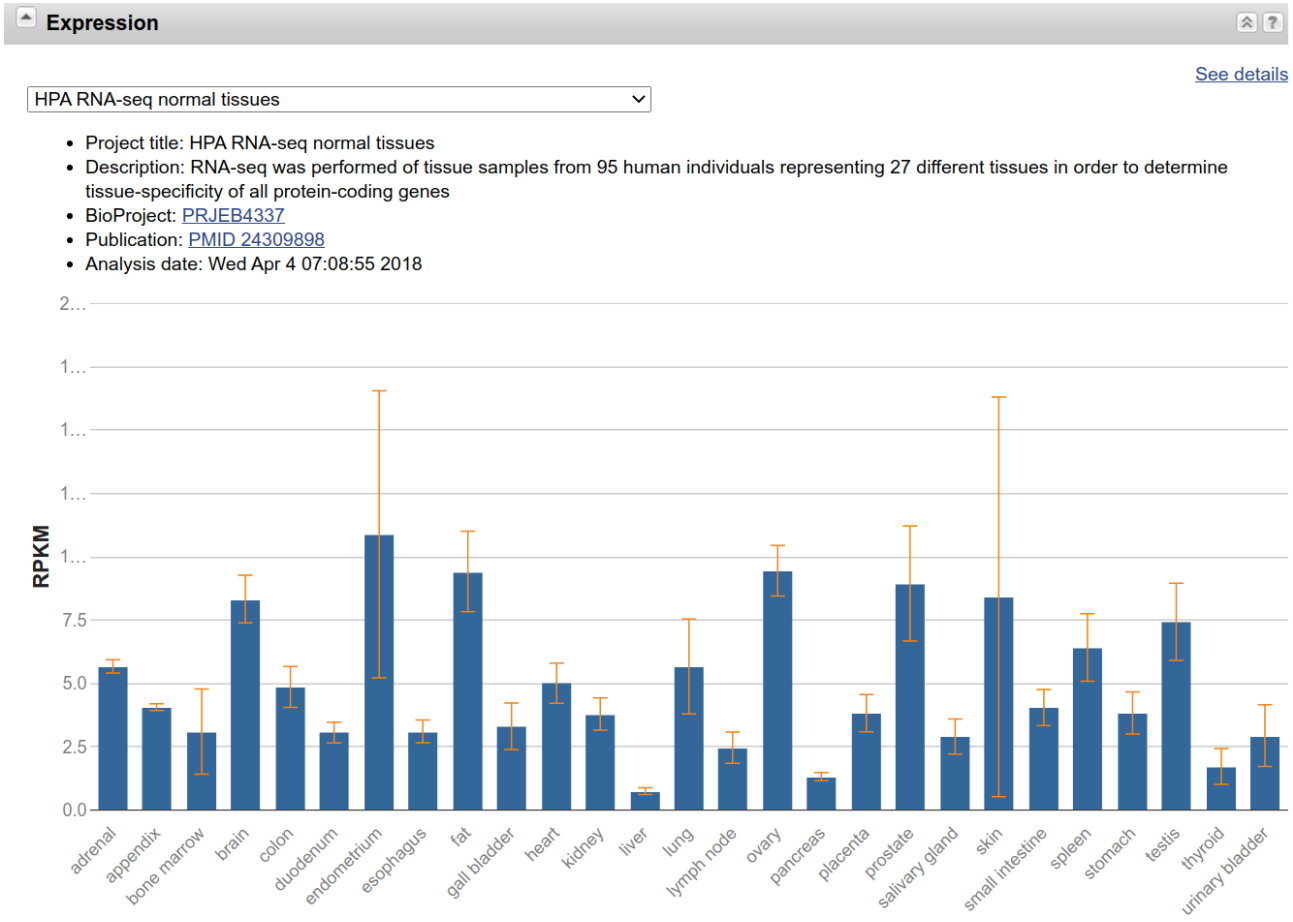
Example 5: Human PKD1

Is Human PKD1 expressed in all the tissues?



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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

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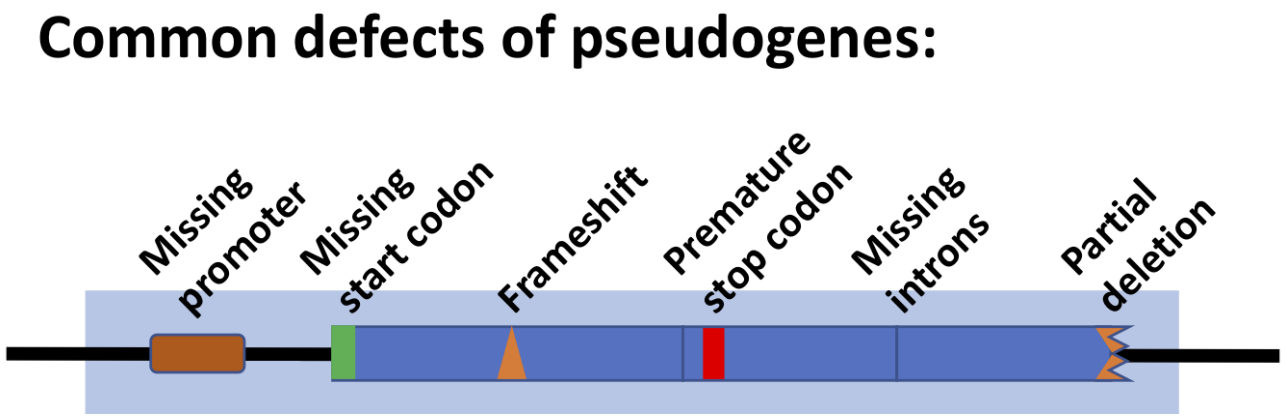
Example 5: Human PKD1

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.



https://en.wikipedia.org/wiki/Pseudogene

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
enables Wnt-activated receptor activity	IDA	PubMed
contributes_to calcium channel activity	IDA	PubMed
enables calcium channel activity	ISS	
enables carbohydrate binding	IEA	
enables protein binding	IPI	PubMed
enables protein domain specific binding	IPI	PubMed
enables protein kinase binding	IPI	PubMed

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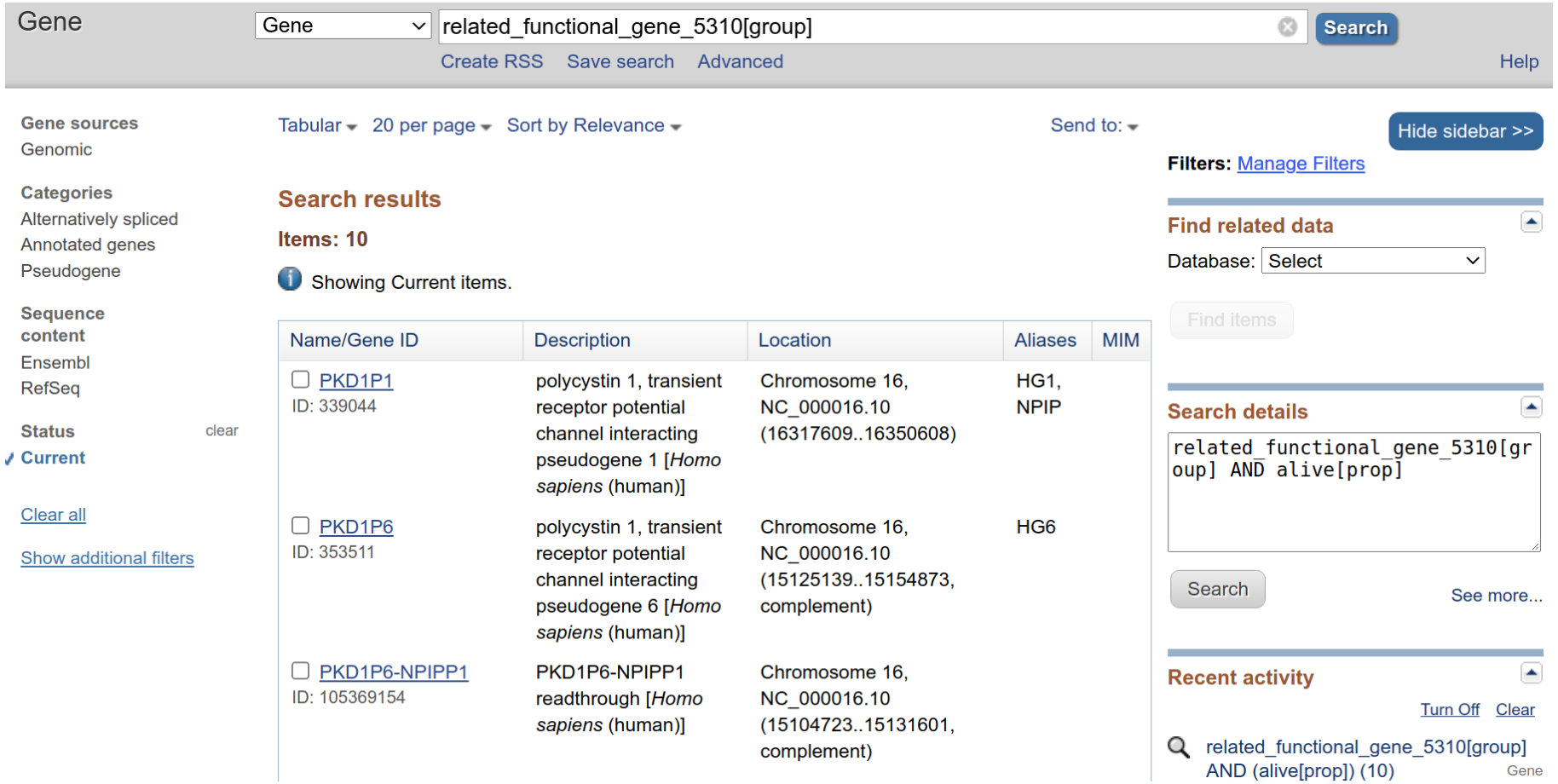

https://www.ncbi.nlm.nih.gov/gene/?Term=related_functional_gene_5310%5Bgroup%5D