

## Entrez Gene Quick Start

### An NCBI Mini-Course

NCBI's Entrez Gene provides gene-based information such as chromosome location, sequence, expression, structure, functional, and homology data. Each record represents a single gene from an organism. Entrez Gene includes organisms for which there is a RefSeq genome record.

In this course, we will learn how to obtain information about a human gene such as:

- mRNA, genomic, and protein sequence
- general gene and protein information
- homologs from other eukaryotes
- known SNPs, and whether the SNPs in the coding region alter the function of the protein product
- phenotypes associated with mutations
- protein structure

The course will also cover the advantages of Entrez Gene such as efficient searching options and availability of gene-specific information for all completely sequenced genomes, including bacteria and viruses.

The following handout includes the screen shots of the exercise demonstrated in the mini-course.

URL: <http://www.ncbi.nlm.nih.gov/Class/minicourses/entrezgene.html>

Course developed by Medha Bhagwat (bhagwat@ncbi.nlm.nih.gov)

## Problem 1

Retrieve human entries related to "prion protein" in Entrez Gene. Identify the gene for prion protein (PRNP). Name the map location of this gene on the human genome. What is the function of this protein? What are the alternate gene symbols? Name the phenotypes associated with the mutations in this gene.

Is the RefSeq mRNA record reviewed? How many alternatively spliced products have been annotated for the gene?

To obtain information about the homologs from other eukaryotes, click on the Homologene link. Change the Display option to "Alignment Scores". How great is the percent identity between the human and mouse proteins? View the alignment by clicking on the "Blast" link.

Go back to the Entrez Gene report. Identify the variations annotated on this gene by clicking on the geneView in dbSNP link. How many of them are nonsynonymous changes? To determine whether known SNPs in the coding region of a gene are associated with any phenotype, access the OMIM record by clicking on the "Yes" link under the OMIM column in the SNP report. Compare the nonsynonymous changes from the SNP report with the "ALLELIC VARIANTS" in the OMIM record. Are there any SNPs known to cause a change in the function of the prion protein?

Go back to the Entrez gene report. View the list of similar proteins through the "BL" link in the next to the protein NP\_000302. To view the site of mutation in the 3D structure, superimpose the protein sequence on the 3D-structure of human prion protein (use BL--3D-structure button--click on the first blue dot--Get 3D Structure Data). Identify and highlight the mutated residue on the 3D structure.

NCBI Entrez Gene

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All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy Books OMIM

Search Gene for prion protein Go Clear

Limits Preview/Index History Clipboard Details

Entrez Gene is a searchable database of genes, from RefSeq genomes, and defined by sequence and/or located in the NCBI Map Viewer

News New "has ccds" property added. News archives...

Sample Searches

Find genes by... Search text

free text human muscular dystrophy

partial name and multiple species transporter[title] AND ("Drosophila melanogaster"[organism] OR "Mus musculus"[organism])

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Search Gene for prion protein Preview Go Clear

Limits Preview/Index History Clipboard Details

- Enter terms and click Preview to see only the number of search results.
- To combine searches use # before search number, e.g., (#2 OR #3) AND asthma.

No history available

Add Term(s) to Query or View Index:

- Enter a term in the text box; use the pull-down menu to specify a search field.
- Click Preview to add terms to the query box and see the number of search results, or click Index to view terms within a field.

Organism human Preview Index

Click AND OR NOT to add a term to the query box.

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Search Gene for prion protein AND human[Organism] Go Clear Save Search

Limits Preview/Index History Clipboard Details

Display Summary Show 20 Send to

All: 51 Current Only: 39 Genes Genomes: 39 SNP GeneView: 38

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1: [TP53](#) Order cDNA clone, Links

Official Symbol TP53 and Name: tumor protein p53 (Li-Fraumeni syndrome) [*Homo sapiens*]  
 Other Aliases: LFS1, TRP53, p53  
 Other Designations: p53 tumor suppressor, tumor protein p53  
 Chromosome: 17; Location: 17p13.1  
 Annotation: Chromosome 17, NC\_000017.9 (7512464..7531642, complement)  
 MIM: 191170  
 GeneID: 7157

2: [PRNP](#) Order cDNA clone, Links

Official Symbol PRNP and Name: prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [*Homo sapiens*]  
 Other Aliases: ASCR, CD230, CJD, GSS, MGC26679, PRIP, PrP, PrP27-30, PrP33-35C, PrPc  
 Other Designations: CD230 antigen, major prion protein; prion protein; prion protein PrP; prion-related protein  
 Chromosome: 20; Location: 20p13  
 Annotation: Chromosome 20, NC\_000020.9 (4615069..4630234)  
 MIM: 176640  
 GeneID: 5621

3: [HSPA5](#) Order cDNA clone, Links

Official Symbol HSPA5 and Name: heat shock 70kDa protein 5 (glucose-regulated protein, 78kDa) [*Homo sapiens*]  
 Other Aliases: BIP, FLJ26106, GRP78, MIF2  
 Other Designations: Heat-shock 70kD protein-5 (glucose-regulated protein, 78kD); heat shock 70kD protein 5 (glucose-regulated protein, 78kD); heat shock 70kDa protein 5  
 Chromosome: 9; Location: 9q33-q34.1  
 Annotation: Chromosome 9, NC\_000009.10 (127036953..127043430, complement)  
 MIM: 138120  
 GeneID: 3309

NCBI Entrez Gene

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All: [Home](#) [Conduct a Search](#) [Genes](#) [Genomes](#) [SNP GeneView](#) 1

**1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)** [ Entrez Gene Home ]

*Homo sapiens* | updated 08-Jul-2007

GeneID: 5621

**Summary**

**Official Symbol** PRNP provided by HGNC

**Official Full Name** prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) provided by HGNC

**Primary source** HGNC:9449

**See related** Ensembl:ENSG00000171867; HPRD:01453; MIM:176640

**Gene type** protein coding

**RefSeq status** Reviewed

**Organism** *Homo sapiens*

**Lineage** Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominoidea; Homo

**Also known as** CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C

**Summary** The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Strausler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.

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**Links** Explain

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- OMIA
- OMIM
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- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy

**Genomic regions, transcripts, and products**

Go to [reference sequence details](#)

**NC\_000020.9**

[ 4615969 ] [ 4639234 ]

5' 3'

NP\_488231.2 NP\_488232.1 prnp/protein C0013038.1  
 NC\_153779.1 NC\_153780.1 prnp/protein C0013038.1

■ - coding region ■ - untranslated region

**Genomic context**

chromosome: 20; Location: 20p13

See PRNP in MapViewer

[ 4519639 ] [ 4669314 ]

RPL7AL2 RPS4L2 PRNP PRNT

**SNP: Genotype**

**SNP: GeneView**

Taxonomy

UniSTS

AceView

CCDS

Evidence Viewer

GDB

GeneTests for MIM: 176640

HGMD

HGNC

HPRD

KEGG

MGC

ModelMaker

PharmGKB

UniGene

LinkOut

**Entrez Gene Info**

**Feedback**

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Search: Gene for [Go] [Clear]

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Gene Table

Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia [ ]

updated 08-Jul-2007

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Lineage: Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Homnidae; Homo

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Search: Gene for [Go] [Clear]

Display: Gene Table Show: 20 Send to: [ ]

Gene Table

1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [ *Homo sapiens* ]

GeneID: 5621 updated 08-Jul-2007

RefSeq status: Reviewed total gene size: 15166 bp

Genomic regions, transcripts, and products

Go to reference sequence details

mRNA bp exons Protein aa exons

NM_000311.2	2468	2	NP_000309.1	254	1
NM_183079.1	2464	2	NP_898302.1	254	1

Exon information:

NM\_000311.2 length: 2468 bp, number of exons: 2

EXON	Coding EXON	INTRON			
coords	length	coords	length	coords	length
1-90	90 bp	91-12708	12698 bp		
12709-13166	2378 bp	12793-13166	762 bp		

NM\_183079.1 length: 2464 bp, number of exons: 2

EXON	Coding EXON	INTRON			
coords	length	coords	length	coords	length
1-86	86 bp	87-12708	12702 bp		

Resources

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SNP GeneView: 1

**Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial** Entrez Gene Home

updated 08-Jul-2007

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PubMed (GeneRIF)

SNP

SNP: Genotype

SNP: GeneView

Taxonomy

UniSTS

AcView

NC\_000020.9

[4639234]

[4639234]

NP\_013422.1 prnp protein 500913022.1

NP\_000302.1 prnp protein 500913022.1

untranslated region

mRNA	bp	exons	Protein	aa	exons
NM_000311.2	2468	2	NP_000302.1	254	1
NM_183073.1	2464	2	NP_338302.1	254	1

Genomic context

chromosome: 20; Location: 20p13

See PRNP in MapViewer

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Related Articles in PubMed

PubMed links

GeneRIFs: Gene References Into Function

What's a GeneRIF?

1. Norwegian sisters with late onset Creutzfeldt-Jakob disease caused by the E200K mutation.
2. Finds polymorphism at codon 129 of PRNP gene implicated in the development of CJD in Greek population. Met/Val allele frequencies and genotype distribution examined in 345 individuals. Genotypes Met/Met 50%, Met/Val 39% and Val/Val 11% were observed.
3. Increased plasma PrP(C) reflects an endogenous increase in expression in acute stroke-affected brain tissue.
4. review focuses on transfusion-transmission of variant Creutzfeldt-Jakob disease by red cell preparations
5. Combined molecular, biochemical, and single living polarized cell imaging characterizations suggest that hPrP(C) is selectively targeted to the apical side of Madin-Darby canine kidney (MDCKII) and of intestinal epithelia (Caco2) cells.
6. Oxidative stress might be an influence that leads to substantial structural conversions of PrP in vivo.
7. prion protein does not require other Bcl-2 family proteins to protect against Bax-mediated cell death
8. This study suggested that polymorphism at position -101 in the regulatory region of PRNP may be a risk factor for sCJD among codon 129 heterozygotes.
9. Equilibrium binding and kinetics of FRET show that the PRNP binding to the oligonucleotides and their bending occur simultaneously.
10. analysis of experimentally derived constraints for high-resolution structural models of PrP amyloid fibrils

Submit: [New GeneRIF](#) [Correction](#)

**HIV-1 protein interactions**

**Protein Interaction**  
 1: Tat HIV-1 Tat binds to a stem-loop structure in the mRNA of prion protein (PrP) that is similar to HIV-1 TAR RNA and infection of astrocytes with HIV-1 results in an increased level of PrP mRNA, suggesting Tat upregulates PrP expression [PubMed](#)  
[Go to the HIV-1, Human Protein Interaction Database](#)

**Interactions**

Product	Interactant	Other Gene	Complex	Source	Pubs
NP_000302.1	NP_001155.1	APBB1		HPRD	PubMed
NP_000302.1	NP_052759.2	CLSTN1		HPRD	PubMed
NP_000302.1	NP_001822.2	CLU		HPRD	PubMed
NP_000302.1	NP_001834.2	CNTN1		HPRD	PubMed
PrPc interacts with CSNK2A1 (CK2 alpha). This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2A1 (CK2 alpha).					
NP_000302.1	NP_001886.1	CSNK2A1		BIND	PubMed
PrPc interacts with CSNK2A2 (CK2 alpha prime). This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2A2 (CK2 alpha prime).					
NP_000302.1	NP_001887.1	CSNK2A2		BIND	PubMed
PrPc interacts with CSNK2B (CK2 beta) albeit weakly. This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2B (CK2 beta).					
NP_000302.1	NP_001311.3	CSNK2B		BIND	PubMed
NP_000302.1	NP_004399.1	DNM1		HPRD	PubMed
NP_000302.1	NP_570629.1	DPP6		HPRD	PubMed
NP_000302.1	NP_002077.1	GRB2		HPRD	PubMed
PrPc interacts with HSPA5 (BiP).					
NP_000302.1	NP_005338.1	HSPA5		BIND	PubMed
PrPc interacts with HSPD1 (Hsp60). This interaction was modeled on a demonstrated interaction between hamster PrPc and human HSPD1 (Hsp60).					
NP_000302.1	NP_002147.2	HSPD1		BIND	PubMed
NP_000302.1	NP_000416.1	L1CAM		HPRD	PubMed

**General gene information**

**Markers**

**WI-18738(e-PCR)**  
 Links: [UniSTS:1017](#)  
 Alternate names: HSA.55; RH57301; STS-D00015

**SGC44304(e-PCR)**  
 Links: [UniSTS:2335](#)  
 Alternate names: EST498946; RH57429

**D20S1014(e-PCR)**  
 Links: [UniSTS:21619](#)  
 Alternate names: G00-677-676; GDB:120720; GDB:677676; RH14068; RH63750; SHGC-12813; stSG10911; UTR-03221; WI-7784

**RH71030(e-PCR)**  
 Links: [UniSTS:34672](#)  
 Alternate names: GDB:177793; stSG20232

**RH47809(e-PCR)**  
 Links: [UniSTS:38471](#)  
 Alternate name: stSG28721

**RH70248(e-PCR)**  
 Links: [UniSTS:43453](#)  
 Alternate name: T27631

**Phenotypes**

## Phenotypes

Creutzfeldt-Jakob disease  
[MIM: 123400](#)  
Gerstmann-Straussler disease  
[MIM: 137440](#)  
Huntington disease-like 1  
[MIM: 603218](#)  
Insomnia, fatal familial  
[MIM: 600072](#)  
Prion disease with protracted course  
[MIM: 606688](#)

## Pathways

KEGG pathway: Neurodegenerative Disorders  
[01510](#)  
KEGG pathway: Prion disease  
[05060](#)

## Homology

Mouse, Rat  
[Map Viewer](#)

## GeneOntology

Provided by [GOA](#)

Function	Evidence
<a href="#">GPI anchor binding</a>	IEA
<a href="#">copper ion binding</a>	TAS <a href="#">Pubmed</a>
<a href="#">microtubule binding</a>	IDA <a href="#">Pubmed</a>
<a href="#">protein binding</a>	IEA

Process	Evidence
<a href="#">copper ion homeostasis</a>	NAS <a href="#">Pubmed</a>
<a href="#">metabolic process</a>	TAS <a href="#">Pubmed</a>
<a href="#">response to oxidative stress</a>	ISS

Component	Evidence
<a href="#">Golgi apparatus</a>	ISS
<a href="#">cytoplasm</a>	TAS <a href="#">Pubmed</a>
<a href="#">endoplasmic reticulum</a>	ISS
<a href="#">extrinsic to membrane</a>	TAS <a href="#">Pubmed</a>
<a href="#">lipid raft</a>	ISS

**General protein information** 

**Names**  
 prion protein  
 CD230 antigen  
 prion protein PrP  
 major prion protein  
 prion-related protein

**NCBI Reference Sequences (RefSeq)** 

**RefSeqs maintained independently of Annotated Genomes**

These reference sequences exist independently of genome builds: [Explain](#)

**mRNA and Protein(s)**

1.	<a href="#">NM_000311.3</a> - <a href="#">NP_000302.1</a>	<b>prion protein preproprotein</b>
	Description	Transcript Variant: This variant (1) represents the longest transcript. Variants 1-5 encode the same protein.
	Source sequence(s)	<a href="#">AW452130</a> , <a href="#">BC022532</a> , <a href="#">DA297032</a> , <a href="#">M13899</a>
	Consensus CDS	<a href="#">CCDS13080.1</a>
	Conserved Domains (1)	<a href="#">summary</a>
		<a href="#">smart00157</a> Location:23-230 Blast Score:546 PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Strausler syndrome and bovine spongiform encephalopathy
2.	<a href="#">NM_001080121.1</a> - <a href="#">NP_001073590.1</a>	<b>prion protein preproprotein</b>
	Description	Transcript Variant: This variant (3) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
	Source sequence(s)	<a href="#">AW452130</a> , <a href="#">BC022532</a> , <a href="#">BP251427</a> , <a href="#">DA122620</a> , <a href="#">M13899</a>
	Conserved Domains (1)	<a href="#">summary</a>
		<a href="#">smart00157</a> Location:23-230 Blast Score:546 PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Strausler syndrome and bovine spongiform encephalopathy
3.	<a href="#">NM_001080122.1</a> - <a href="#">NP_001073591.1</a>	<b>prion protein preproprotein</b>
	Description	Transcript Variant: This variant (4) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
	Source sequence(s)	<a href="#">AW452130</a> , <a href="#">BC022532</a> , <a href="#">B1669189</a> , <a href="#">DA297032</a> , <a href="#">M13899</a>
4.	<a href="#">NM_001080123.1</a> - <a href="#">NP_001073592.1</a>	<b>prion protein preproprotein</b>
	Description	Transcript Variant: This variant (5) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
	Source sequence(s)	<a href="#">BC022532</a> , <a href="#">D8461478</a> , <a href="#">M13899</a>
	Conserved Domains (1)	<a href="#">summary</a>
		<a href="#">smart00157</a> Location:23-230 Blast Score:546 PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Strausler syndrome and bovine spongiform encephalopathy
5.	<a href="#">NM_183079.2</a> - <a href="#">NP_898902.1</a>	<b>prion protein preproprotein</b>
	Description	Transcript Variant: This variant (2) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.

**RefSeqs of Annotated Genomes: Build 36.2**

The following sections contain reference sequences that belong to a specific genome build: [Explain](#)

**Reference assembly**

**Genomic**

1.	<a href="#">NC_000020.9</a>	<b>Reference assembly</b>
	Range	4615069..4630234
	Download	<a href="#">GenBank</a> <a href="#">FASTA</a>
2.	<a href="#">NT_011387.8</a>	
	Range	4607069..4622234
	Download	<a href="#">GenBank</a> <a href="#">FASTA</a>

**Alternate assembly (based on Celera assembly)**

**Genomic**

1.	<a href="#">AC_000063.1</a>	<b>Alternate assembly (based on Celera assembly)</b>
	Range	4736784..4751948
	Download	<a href="#">GenBank</a> <a href="#">FASTA</a>
2.	<a href="#">NW_927317.1</a>	
	Range	4593960..4609124
	Download	<a href="#">GenBank</a> <a href="#">FASTA</a>

**Related Sequences** 

Nucleotide	Protein
Genomic <a href="#">AF030575.1</a>	<a href="#">AAC05365.1</a>
Genomic <a href="#">AF076976.1</a>	<a href="#">AAD46098.1</a>
Genomic <a href="#">AF085477.2</a>	<a href="#">AAC62750.2</a>
Genomic <a href="#">AF315723.1</a>	None
Genomic <a href="#">AL133396.2</a>	<a href="#">CAB75503.1</a>
	<a href="#">CAI19053.1</a>
Genomic <a href="#">AY219882.1</a>	<a href="#">AAO83635.1</a>
Genomic <a href="#">AY219883.1</a>	<a href="#">AAO83636.1</a>
Genomic <a href="#">AY458651.1</a>	<a href="#">AAR21603.1</a>
Genomic <a href="#">CH471133.3</a>	<a href="#">EAX10449.1</a>
	<a href="#">EAX10450.1</a>
Genomic <a href="#">DQ408531.1</a>	<a href="#">ABD63004.1</a>
Genomic <a href="#">DQ894502.2</a>	<a href="#">ABM85428.1</a>
Genomic <a href="#">M81929.1</a>	<a href="#">AAB59442.1</a>
Genomic <a href="#">M81930.1</a>	<a href="#">AAB59443.1</a>
Genomic <a href="#">S71208.1</a>	<a href="#">AAB20521.1</a>

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All: 1 Current Only: 1 Genes Genomes: 1 SNP GeneView: 1

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*Homo sapiens* updated 08-Jul-2007

GeneID: 5621

**Summary**

**Official Symbol** PRNP provided by [HGNC](#)

**Official Full Name** prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) provided by [HGNC](#)

**Primary source** [HGNC:9449](#)

**See related** [Ensembl:ENSG00000171867](#); [HPRD:01453](#); [MIM:176640](#)

**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*

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**Genomic regions, transcripts, and products**

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- CCDS

NCBI HomoloGene Discover Homologs

Search HomoloGene for [ ] Go Clear

Display Alignment Scores Show 20 Send to

All: 1 Fungi: 0 Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota

H.sapiens	PRNP	prion protein (p27-30) (Creutzfeldt-Jakob ...
P.troglodytes	PRNP	prion protein (p27-30) (Creutzfeldt-Jakob ...
C.familiaris	PrP	prion protein (p27-30) (Creutzfeldt-Jakob ...
M.musculus	Prnp	prion protein
R.norvegicus	Prnp	prion protein
G.gallus	PRNP	prion protein (p27-30)

NCBI HomoloGene Discover Homologs

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Limits Preview/Index History Clipboard Details

Display Alignment Scores Show 20 Send to

All: 1 Fungi: 0 Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota Download, Link

### Alignment Scores

Species	Gene	aa%ID	nt%ID	D	Ka/Ks	Knr/Knc	
<b>H.sapiens PRNP</b>							
vs. P.troglodytes	PRNP	99.2	99.2	0.008	0.138	0.548	<a href="#">Blast</a>
vs. C.familiaris	PrP	87.7	87.3	0.139	0.126	0.282	<a href="#">Blast</a>
vs. M.musculus	Prnp	90.1	85.3	0.163	0.077	0.342	<a href="#">Blast</a>
vs. R.norvegicus	Prnp	89.7	86.8	0.145	0.090	0.260	<a href="#">Blast</a>
vs. G.gallus	LOC396452	47.1	57.4	0.631	0.399	0.878	<a href="#">Blast</a>
<b>P.troglodytes PRNP</b>							
vs. H.sapiens	PRNP	99.2	99.2	0.008	0.138	0.548	<a href="#">Blast</a>
vs. C.familiaris	PrP	87.7	87.3	0.139	0.126	0.253	<a href="#">Blast</a>
vs. M.musculus	Prnp	90.1	85.2	0.165	0.075	0.307	<a href="#">Blast</a>
vs. R.norvegicus	Prnp	89.7	86.7	0.146	0.088	0.229	<a href="#">Blast</a>
vs. G.gallus	LOC396452	47.1	56.9	0.642	0.351	0.882	<a href="#">Blast</a>
<b>C.familiaris PrP</b>							
vs. H.sapiens	PRNP	87.7	87.3	0.139	0.126	0.282	<a href="#">Blast</a>
vs. P.troglodytes	PRNP	87.7	87.3	0.139	0.126	0.253	<a href="#">Blast</a>
vs. M.musculus	Prnp	86.9	82.5	0.200	0.092	0.335	<a href="#">Blast</a>

NCBI Blast 2 Sequences results

PubMed Entrez BLAST OMIM Taxonomy Structure

BLAST 2 SEQUENCES RESULTS VERSION BLAST 2.2.16 [Mar-25-2007]

Matrix: BLOSUM62 gap open: 11 gap extension: 1  
 x\_dropoff: 0 expect: 10.0000 wordsize: 3 Filter View option: Standard  
 Masking character option: X for protein, n for nucleotide Masking color option: Black  
 Show CDS translation Align

**Sequence 1:** [gi|24335270|ref|NP\\_898902.1|](#)prion protein preproprotein [Homo sapiens] >[gi|4506113|ref|NP\\_000302.1|](#)prion protein preproprotein [Homo sapiens] >[gi|122056623|ref|NP\\_001073590.1|](#)prion protein preproprotein [Homo sapiens] >[gi|122056625|ref|NP\\_001073591.1|](#)prion protein preproprotein [Homo sapiens] >[gi|122056628|ref|NP\\_001073592.1|](#)prion protein preproprotein [Homo sapiens] >[gi|130912|sp|P04156|PRIO\\_HUMAN](#) Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR) (CD230 antigen) >[gi|190468|gb|AAA60182.1|](#)prion protein >[gi|6996155|...](#)  
 Length = 253 (1 .. 253)

**Sequence 2:** [gi|13173473|ref|NP\\_035300.1|](#)prion protein [Mus musculus] >[gi|130914|sp|P04925|PRIO\\_MOUSE](#) Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen) >[gi|200529|gb|AAA39997.1|](#)prion protein >[gi|2865215|gb|AAC02804.1|](#)short incubation prion protein Prnpa [Mus musculus] >[gi|13879449|gb|AAH06703.1|](#)Prion protein [Mus musculus] >[gi|71060019|emb|CAJ18553.1|](#)Prnp [Mus musculus] >[gi|74182795|dbj|BAE34724.1|](#)unnamed protein product [Mus musculus] >[gi|74186646|dbj|BAE34788.1|](#)unnamed protein product [Mus musculus] >[gi|7419279755|.....](#)  
 Length = 254 (1 .. 254)

**NOTE:** Bitscore and expect value are calculated based on the size of the nr database.

NCBI Entrez Gene

Search: Gene for [ ] Go Clear

Display: Full Report Show 20 Send to [ ]

All: 1 Current Only: 1 Genes Genomes: 1 SNP GeneView: 1

**1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [ Homo sapiens ]**  
 GeneID: 5621 updated 08-Jul-2007

**Summary**

**Official Symbol** PRNP provided by HGNC

**Official Full Name** prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) provided by HGNC

**Primary source** HGNC:9449

**See related** Ensembl: ENSG00000171867; HPRD:01453; MIM:176640

**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** CID; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C

**Summary** The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Strausler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.

**Genomic regions, transcripts, and products**

Go to [reference sequence details](#)

NC\_000020.9

Entrez Gene Home

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- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- CCDS
- Ensembl
- Evidence Viewer
- GDB

NCBI Single Nucleotide Polymorphism

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

Search Entrez SNP for [ ] Go

**BUILD 127**

SNP linked to Gene [PRNP](#)(geneID:5621) Via Contig Annotation

Send rs# on all gene models to Batch Query Download all rs# to file. GENE GENOTYPE REPORT

**Gene Model (mRNA alignment) information from genome sequence**

Total gene model (contig mRNA transcript): 4

mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	List SNP
NM_000311	plus strand	NP_000302	forward	NT_011387	reference	<- currently shown
NM_000311	plus strand	NP_000302	forward	NW_927317	Celera	<a href="#">View snp on GeneModel</a>
NM_183079	plus strand	NP_898902	forward	NT_011387	reference	<a href="#">View snp on GeneModel</a>
NM_183079	plus strand	NP_898902	forward	NW_927317	Celera	<a href="#">View snp on GeneModel</a>

in gene region  cSNP  has frequency  double hit  haplotype tagged refresh

gene model	Contig Label	Contig	mRNA	protein	mRNA orientation	transcript	snp count
(contig mRNA transcript):	reference	NT_011387	NM_000311	NP_000302	forward	plus strand 22, coding	

Region	Contig position	mRNA pos	dbSNP rs#	Heterozygosity	Validation	3D	OMIM	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos
exon_1	4619867	101						start codon				1
exon_2	4619942	176	<a href="#">rs11538755</a>	N.D.				nonsynonymous	A	Thr [T]	1	26
				N.D.				contig reference	C	Pro [P]	1	26
	4619949	183	<a href="#">rs11538762</a>	N.D.				nonsynonymous	A	His [H]	2	28
				N.D.				contig reference	C	Pro [P]	2	28
	4619975	209	<a href="#">rs11538763</a>	N.D.				synonymous	A	Arg [R]	1	37
				N.D.				contig reference	C	Arg [R]	1	37

4620378	612	<a href="#">rs16990018</a>	0.046		Yes	nonsynonymous	G	Ser [S]	2	171
			0.046		Yes Yes	contig reference	A	Asn [N]	2	171
4620399	633	<a href="#">rs11538766</a>	N.D.		Yes	nonsynonymous	T	Val [V]	2	178
			N.D.		Yes	contig reference	A	Asp [D]	2	178
4620405	639	<a href="#">rs11538767</a>	N.D.		Yes	nonsynonymous	C	Ala [A]	2	180
			N.D.		Yes	contig reference	T	Val [V]	2	180
4620464	698	<a href="#">rs28933385</a>	N.D.		Yes	nonsynonymous	A	Lys [K]	1	200
			N.D.		Yes Yes	contig reference	G	Glu [E]	1	200
4620521	755	<a href="#">rs1800014</a>	N.D.		Yes	nonsynonymous	A	Lys [K]	1	219
			N.D.		Yes	contig reference	G	Glu [E]	1	219
4620538	772	<a href="#">rs6052773</a>	N.D.		Yes	synonymous	T	Ala [A]	3	224
			N.D.		Yes	contig reference	C	Ala [A]	3	224
4620545	779	<a href="#">rs17852079</a>	N.D.		Yes	nonsynonymous	A	Lys [K]	1	227
			N.D.		Yes	contig reference	C	Gln [Q]	1	227

**.0006 CREUTZFELDT-JAKOB DISEASE [PRNP, GLU200LYS] dbSNP**  
 FATAL FAMILIAL INSOMNIA, INCLUDED

**MIM \*176640**  
 Description  
 Cloning  
 Gene Structure  
 Mapping  
 Gene Function  
 Molecular Genetics  
 Genotype/Phenotyp  
 Correlations  
 Population Genetics  
 Animal Model  
 History  
 Allelic Variants  
 • View List  
 See Also  
 References  
 Contributors

In 2 patients with Creutzfeldt-Jakob disease ([123400](#)) from the same family, [Goldgaber et al. \(1989\)](#) identified a G-to-A transition in the PRNP gene, resulting in a glu200-to-lys (E200K) substitution.

Studying an unusual cluster of cases of CJD in rural Slovakia, [Goldfarb et al. \(1990\)](#) found the E200K mutation in all 11 tested cases of 'focal CJD,' in 12 of 40 healthy first-degree relatives, and in 6 of 23 other relatives. By contrast, no extrafocal cases or their relatives had the mutation; nor did any unrelated individuals within or outside the cluster regions. One of the healthy individuals with the E200K mutation was the 75-year-old mother of one of the patients. The unusually high incidence of CJD in the Orava and Lucenec regions of Slovakia appeared to be of recent origin. [Goldfarb et al. \(1990\)](#) interpreted this as indicating that the mutation is a necessary, but not sufficient, factor in the disease. Another factor such as scrapie-infected sheep was proposed.

[Mitrova et al. \(1990\)](#) described the familial occurrence of 3 definite and 2 possible cases of CJD with temporal and spatial separation in the area of focal CJD accumulation in Slovakia. The incubation period appeared to be about 51 years, judging by the interval between the death of the affected mother and the clinical onset in the first affected child. Affected offspring tended to die at the same time, not at the same age. Due to separation of the affected children, a possible common exposure to CJD infection was limited to approximately 7 years during their childhood.

Population Genetics  
 Animal Model  
 History  
 Allelic Variants  
 • View List  
 See Also  
 References  
 Contributors  
 Creation Date  
 Edit History

**\*176640**  
**PRION PROTEIN; PRNP**

**ALLELIC VARIANTS**  
**(selected examples)**

- [0001 CREUTZFELDT-JAKOB DISEASE \[PRNP, EXTRA OCTAPEPTIDE CODING REPEATS\]](#)  
 GERSTMANN-STRAUSSLER DISEASE, INCLUDED  
 HUNTINGTON DISEASE-LIKE 1, INCLUDED
- [0002 GERSTMANN-STRAUSSLER DISEASE \[PRNP, PRO102LEU\]](#)
- [0003 REMOVED FROM DATABASE](#)
- [0004 GERSTMANN-STRAUSSLER DISEASE \[PRNP, ALA117VAL\]](#)
- [0005 PRION DISEASE, SUSCEPTIBILITY TO \[PRNP, MET129VAL\] dbSNP](#)  
 ALZHEIMER DISEASE, EARLY-ONSET, SUSCEPTIBILITY TO, INCLUDED  
 APHASIA, PRIMARY PROGRESSIVE, SUSCEPTIBILITY TO, INCLUDED
- [0006 CREUTZFELDT-JAKOB DISEASE \[PRNP, GLU200LYS\] dbSNP](#)  
 FATAL FAMILIAL INSOMNIA, INCLUDED
- [0007 CREUTZFELDT-JAKOB DISEASE \[PRNP, ASP178ASN AND MET129VAL\]](#)
- [0008 REMOVED FROM DATABASE](#)
- [0009 REMOVED FROM DATABASE](#)
- [0010 FATAL FAMILIAL INSOMNIA \[PRNP, ASP178ASN AND MET129\]](#)
- [0011 GERSTMANN-STRAUSSLER DISEASE \[PRNP, PHE198SER\]](#)
- [0012 GERSTMANN-STRAUSSLER DISEASE \[PRNP, GLN217ARG\]](#)
- [0013 REMOVED FROM DATABASE](#)
- [0014 CREUTZFELDT-JAKOB DISEASE \[PRNP, VAL210ILE\]](#)
- [0015 GERSTMANN-STRAUSSLER DISEASE \[PRNP, PRO105LEU\]](#)
- [0016 CREUTZFELDT-JAKOB DISEASE \[PRNP, VAL180ILE\]](#)
- [0017 CREUTZFELDT-JAKOB DISEASE \[PRNP, MET232ARG\]](#)  
 DEMENTIA, LEWY BODY, INCLUDED

Entrez Gene  
 Nomenclature  
 RefSeq  
 GenBank  
 Protein  
 UniGene

LinkOut  
 HGVS  
 HGMD  
 OAD

NCBI OMIM Online Mendelian Inheritance in Man Johns Hopkins University

All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy OMIM

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All: 1

**\*176640**  
**PRION PROTEIN; PRNP**

Alternative titles; symbols

PRP  
 PRION-RELATED PROTEIN; PRIP

Gene map locus [20pter-p12](#)

GeneTests Links

- Books
- Gene
- GEO Profiles
- HomoloGene
- OMIA
- Free in PMC
- Gene Genotype
- GeneView in dbSNP
- UniGene

repeats. This gene is found on chromosome 20, approximately 20-kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Strausler disease, Huntington disease-like 1, and kuru. Two transcript variants encoding the same protein have been found for this gene.

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

NC\_000020.9

Genomic context

chromosome: 20; Location: 20p13

See PRNP in MapViewer

Bibliography

Related Articles in Pubmed

PubMed links

GeneRIFs: Gene References Into Function [What's a GeneRIF?](#)

1. elevated plasma PrP(C) levels in renal disease were observed, showing that plasma PrP (C) is not a specific marker of neurological disease or Creutzfeldt-Jakob disease
2. the polymorphism at residue 129 does not change efficiency of conversion to beta-PrP conformation or affect binding of copper ions, but in a partially denatured

Nucleotide  
 OMIA  
 OMIM  
 Full text in PMC  
 Probe  
 Protein  
 PubMed  
 PubMed (GeneRIF)  
 SNP  
 SNP: Genotype  
 SNP: GeneView  
 Taxonomy  
 UniSTS  
 AceView  
 CCDS  
 Evidence Viewer  
 GDB  
 GeneTests for MIM: 176640  
 HGMD  
 HGNC  
 HPRD  
 KEGG  
 MGC  
 ModelMaker  
 PharmGKB  
 UniGene  
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NCBI

BLAST Protein Structure PubMed Taxonomy  
Genome Nucleotide 3D-Domains Books Help

Query: gi4506113 prion protein preproprotein [Homo sapiens]  
Matching gi: 40075008, 17908793, 3999540, 4000210, 5993964, 6996155, 20251002, 10053355, 11079226, 21506484, 83353219, 46095329, 111961006, 112019514, 130912, 190468, 53934492, 62774377, 15108368, 34335270, 122056623, 122056625, 122056628, 31674692, 33695389, 1828059, 3996152, 11963085, 119630855

Hide identical Best hits Common Tree Taxonomy Report 3D structures CDD-Search Glist Run BLAST

200 BLAST hits to 55 unique species [Sort by taxonomy proximity](#)

Archaea 0 Bacteria 192 Metazoa 0 Fungi 0 Plants 0 Viruses 0 Other Eukaryotae

Keep only [ ] Cut-Off 100 Select Reset New search by GI: 4506113 Go

253 aa

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
<a href="#">Conserved Domain Database hits</a>				
1435	31	CBT8503	4996155	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1435	31	AAQ21693	11079226	prion protein [Homo sapiens]
1435	31	AA880162	46095329	prion protein [Homo sapiens]
1435	31	P04156	130912	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR) (CD230 antigen)
1435	31	AAA60182	190468	prion protein
1435	31	NP_898902	34335270	prion protein preproprotein [Homo sapiens]
1435	31	NP_001...	122056623	prion protein preproprotein [Homo sapiens]
1435	31	NP_001...	122056625	prion protein preproprotein [Homo sapiens]
1435	31	NP_001...	122056628	prion protein preproprotein [Homo sapiens]
1435	31	EAX10449	119630854	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1435	31	EAX10450	119630855	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1432	29	AAQ50089	474359	prion protein
1432	29	P40282	730390	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen)
1432	1	AAK42953	61367107	prion protein [synthetic construct]
1432	1	AAV38282	54695820	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1432	1	AAK37089	60834334	prion protein [synthetic construct]
1431	1	ABM62244	123980830	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1431	1	ABM65428	123995653	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1431	31	CAG46869	49457097	PRNP [Homo sapiens]
1431	31	AAH12844	15277486	Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa

NCBI

BLAST Protein Structure PubMed Taxonomy  
Genome Nucleotide 3D-Domains Books Help

Query: gi4506113 prion protein preproprotein [Homo sapiens]  
Matching gi: 40075008, 17908793, 3999540, 4000210, 5993964, 6996155, 20251002, 10053355, 11079226, 21506484, 83353219, 111961006, 112019514, 130912, 190468, 53934492, 62774377, 15108368, 34335270, 122056623, 122056625, 122056628, 31674692, 33695389, 1828059, 3996152, 119630854, 119630855

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Hide identical Best hits Common Tree Taxonomy Report 3D structures CDD-Search Glist Run BLAST

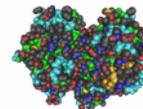
51 BLAST hits to 14 unique species [Sort by taxonomy proximity](#)

Archaea 0 Bacteria 49 Metazoa 0 Fungi 0 Plants 0 Viruses 0 Other Eukaryotae

Keep only [ ] Cut-Off 100 Select Reset New search by GI: 4506113 Go

253 aa

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
<a href="#">Conserved Domain Database hits</a>				
1203	*	1QL2A	6730487	Chain A, Human Prion Protein
1203	*	1QLXA	6730485	Chain A, Human Prion Protein
1136	*	1DX1A	9955174	Chain A, Bovine Prion Protein Residues 23-230
1136	*	1DX0A	9955173	Chain A, Bovine Prion Protein Residues 23-230
771	*	1QN1A	6730489	Chain A, Human Prion Protein Fragment 90-230
771	*	1QNOA	6730488	Chain A, Human Prion Protein Fragment 90-230
768	*	1F07A	10835618	Chain A, Human Prion Protein Mutant E200K Fragment 90-231
768	*	1FCA	10835617	Chain A, Human Prion Protein (Mutant E200K) Fragment 90-231
698	*	1B10A	6729981	Chain A, Solution Nmr Structure Of Recombinant Syrian Hamster Prion Protein Rprp(90-231) , 25 Struct
678	*	2FJ3A	122920186	unnamed protein product [Oryctolagus cuniculus]
616	*	1TFXA	50513788	Chain A, Ovine Recombinant Prp(114-234), Arq Variant In Complex With The Fab Of The Vrq14 Antibody
604	*	1QNSA	6730491	Chain A, Human Prion Protein Fragment 121-230
604	*	1QNS2A	6730490	Chain A, Human Prion Protein Fragment 121-230
597	*	1I4HA	20150089	Chain A, Crystal Structure Of The Human Prion Protein Reveals A Mechanism For Oligomerization
594	*	1DWZA	9955172	Chain A, Bovine Prion Protein Fragment 121-230
594	*	1DWYA	9955171	Chain A, Bovine Prion Protein Fragment 121-230
583	*	1HOLA	28373307	Chain A, Human Prion Protein 121-230 M166E221C
576	*	1Y2SA	60594516	Chain A, Ovine Prion Protein Variant R168
569	*	1XYUA	60594486	Chain A, Solution Structure Of The Sheep Prion Protein With Polymorphism H168
563	*	1HJNA	33356987	Chain A, Human Prion Protein At Ph 7.0
563	*	1HJMA	33356986	Chain A, Human Prion Protein At Ph 7.0
563	*	1XYWA	60594487	Chain A, Elk Prion Protein
560	*	1E1WA	9955179	Chain A, Human Prion Protein Variant R220K
560	*	1E1UA	9955178	Chain A, Human Prion Protein Variant R220K



**Query:** prion protein preproprotein [Homo sapiens]  
[gi: [4506113](#)]

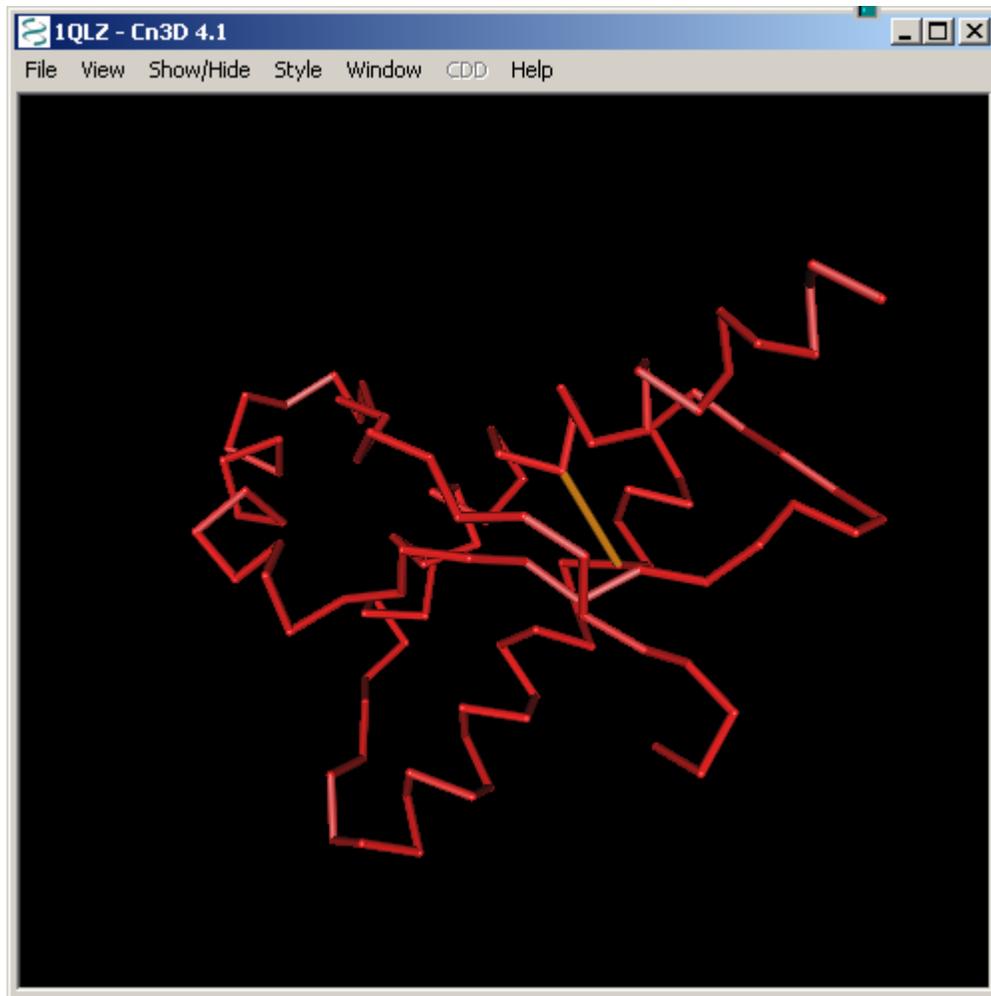
**Structure:** 1QLZ Chain A, Human Prion Protein

**Reference:** [[MIMDB](#)] [[PubMed](#)]

to:  (To display structure, download [Cn3D](#))

**E-value = 1e-131, Bit score = 468, Aligned length = 208, Sequence Identity = 100%**

		10	20	30	40	50	60	70	80	
		.....*	.....*	.....*	.....*	.....*	.....*	.....*	.....*	
<a href="#">gi_4506113</a>	23	KKRPKPGGWNTGGSRYPGQGS	PGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP							102
<a href="#">1QLZ_A</a>	3	KKRPKPGGWNTGGSRYPGQGS	PGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP							82
		90	100	110	120	130	140	150	160	
		.....*	.....*	.....*	.....*	.....*	.....*	.....*	.....*	
<a href="#">gi_4506113</a>	103	SKPKTNMKHMAGAAAAGAVVGG	LGGYMLGSAMSRPIIHFGSDYEDRYRENMMHRYPNQVYYRPMDEYSNQNNFVHDCVNI							182
<a href="#">1QLZ_A</a>	83	SKPKTNMKHMAGAAAAGAVVGG	LGGYMLGSAMSRPIIHFGSDYEDRYRENMMHRYPNQVYYRPMDEYSNQNNFVHDCVNI							162
		170	180	190	200					
		.....*	.....*	.....*	.....*	.....*	.....*	.....*	.....*	
<a href="#">gi_4506113</a>	183	TIKQHTVTTTTKGENFTETDV	KMMERVVEQMCITQYERESQAYYQRGS							230
<a href="#">1QLZ_A</a>	163	TIKQHTVTTTTKGENFTETDV	KMMERVVEQMCITQYERESQAYYQRGS							210



1QLX - Sequence/Alignment Viewer

View Edit Mouse Mode Unaligned Justification Imports

1QLX A	LGGYMLGSAMSRPIIHFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNIITIKQHTVTTTTKGENFTETDVKMME
gi 4506113	LGGYMLGSAMSRPIIHFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNIITIKQHTVTTTTKGENFTETDVKMME

## Problem 2

Retrieve human entries related to "colon cancer" in [Entrez Gene](#). Identify the gene MLH1. Name the map location of this gene on the human genome. What is the function of this protein? What are the alternate gene symbols? Name the phenotypes associated with the mutations in this gene.

Is the RefSeq mRNA record reviewed? How many alternatively spliced products have been annotated for the gene?

To obtain information about the homologs from other eukaryotes, click on the Homologene link. Change the Display option to "Alignment Scores". How great is the percent identity between the human and mouse proteins? View the alignment by clicking on the "Blast" link.

Go back to the Entrez Gene report. Identify the variations annotated on this gene by clicking on the geneView in dbSNP link. How many of them are nonsynonymous changes? To determine whether known SNPs in the coding region of a gene are associated with any phenotype, access the OMIM record by clicking on the "Yes" link under the OMIM column in the SNP report. Compare the nonsynonymous changes from the SNP report with the "ALLELIC VARIANTS" in the OMIM record. Are there any SNPs known to cause a change in the function of the MLH1 protein?

Go back to the Entrez gene report. View the list of similar proteins through the "BL" link in the next to the protein NP\_000240. To view the sites of mutations in the 3D structure, superimpose the protein sequence on the 3D-structure of E.coli multL protein 1BKNB (use BL--3D-structure button--click on the second blue dot--Get 3D Structure Data). Identify and highlight the amino acid corresponding to the human MLH1 isoleucine 32 on the 3D structure. What is the amino acid at this position in the E.coli protein? Based on this information, do you think the I32V mutation in the human protein will alter its function? Confirm your findings through the OMIM record for MLH1.