

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.

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Search Gene for prion protein Go Clear

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

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- 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:

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Organism Preview Index

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

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Display Summary Show 20 Send to

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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

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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
 [*Homo sapiens*]
 GeneID: 5621 updated 07-Oct-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 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.

Genomic regions, transcripts, and products

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

NC_000020.9

■ = coding region ■ = untranslated region

Genomic context

chromosome: 20; Location: 20p13

See PRNP in MapViewer

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SNP: Genotype
 SNP: GeneView
 Taxonomy
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 AceView
 CCDS
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NIH cDNA clone links

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OMIM Links

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Probe Links

Protein Links

Exon

NP_000302.1 length: 253 aa, number of exons: 2

NP_000302.1

EXON Coding EXON INTRON

coords length coords length

1-90 90 bp 91-12788 12698 bp

12789-15466 2678 bp 12789-12860 762 bp

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OMIA

OMIM

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Probe

Protein

PubMed (GeneRIF)

SNP

SNP: Genotype

SNP: GeneView

Taxonomy

UNISTs

AceView

CCDS

Ensembl

Evidence Viewer

GDB

GeneTests for MIM: 176640

HGMD

HGNC

Genomic context

chromosome: 20; Location: 20p13

See PRNP in MapViewer

[4519639] RPL7RL2 [4669314]

RPL7RL2 RPS4L2 PRNP PRNP PRNT

Bibliography

Related Articles in PubMed

PubMed links

GeneRIFs: Gene References Into Function

What's a GeneRIF?

1. analysis of species-specific differences in the intermediate states of human and Syrian hamster prion protein detected by high pressure NMR spectroscopy
2. A South African family had a progressive dementia and atypical pathology associated with kuru-like prion protein plaques. The original mutation in this family occurred on a PRNP allele encoding a 1-octapeptide repeat deletion polymorphism.
3. We found that rPrP fibrils but not alpha-rPrP or soluble beta-sheet rich oligomers caused degeneration of neuronal processes. Degeneration of processes was accompanied by a collapse of microtubules and aggregation of cytoskeletal proteins.
4. Prion protein gene MM genotype increases late-onset Alzheimer's disease risk in Polish population
5. human brain Pp(2)C interacts with selectins in a manner that is distinct from interactions in peripheral tissues; alternations in these interactions may have pathological consequences
6. This is the first publication of data that support the hypothesis that the common methionine/valine polymorphism at codon 129 of the PRNP gene may modify the susceptibility of women to mild temporal lobe epilepsy.
7. A novel three extra-repeat (72 bp) insertion within the octapeptide-coding region was identified in a Chinese family.
8. the PRNP polymorphism is more common in the Korean than in the Japanese population
9. plasmin cleaves Pp(2)C in vitro and the liberated NH(2)-terminal fragment accelerates plasminogen activation

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

HGMD

HGNC

HPRD

KEGG

MGC

ModelMaker

PharmGKB

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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

Description	Product	Interactant	Other Gene	Complex	Source	Pubs
	NP_000302.1	NP_001155.1	APBB1		HPRD	PubMed
	NP_000302.1	NP_055759.2	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	HIF1A		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

Genotypes

[See PRNP SNP GeneView Report](#)
[See PRNP SNP Genotype Report](#)

Phenotypes

Phenotypes

Creutzfeldt-Jakob disease
[MIM: 123400](#)
Gerstmann-Strausler 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
GPI anchor binding	IEA
copper ion binding	TAS PubMed
microtubule binding	IDA PubMed
protein binding	IPI PubMed

Process	Evidence
cellular copper ion homeostasis	NAS PubMed
metabolic process	TAS PubMed
response to oxidative stress	ISS

Component	Evidence
Golgi apparatus	ISS
cytoplasm	TAS PubMed
endoplasmic reticulum	ISS
extrinsic to membrane	TAS PubMed
lipid raft	ISS
membrane	IEA
plasma membrane	ISS

General protein information



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. NM_000311.3–NP_000302.1 prion protein preproprotein	
Description	Transcript Variant: This variant (1) represents the longest transcript. Variants 1-5 encode the same protein.
Source sequence(s)	AW452130 , BC022532 , DA297032 , M13899
Consensus CDS	CCDS13080.1
Conserved Domains (1)	summary
	smart00157 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-Straussler syndrome and bovine spongiform encephalopathy.
2. NM_001080121.1–NP_001073590.1 prion protein preproprotein	
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)	AW452130 , BC022532 , BP251427 , DA122620 , M13899
Conserved Domains (1)	summary
	smart00157 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-Straussler syndrome and bovine spongiform encephalopathy.
3. NM_001080122.1–NP_001073591.1 prion protein preproprotein	
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)	AW452130 , BC022532 , BI669189 , DA297032 , M13899
Conserved Domains (1)	summary
	smart00157 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-Straussler syndrome and bovine spongiform encephalopathy.
4. NM_001080123.1–NP_001073592.1 prion protein preproprotein	
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)	BC022532 , DB461478 , M13899
Conserved Domains (1)	summary
	smart00157 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-Straussler syndrome and bovine spongiform encephalopathy.
5. NM_183079.2–NP_898902.1 prion protein preproprotein	
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.
Source sequence(s)	AW452130 , AY008282 , BC022532 , DA122620
Consensus CDS	CCDS13080.1
Conserved Domains (1)	summary
	smart00157 PRP; Major prion protein: The prion protein is a major component of scrapie-associated fibrils in

Reference assembly

Genomic

1. **NC_000020.9 Reference assembly**
Range 4615069..4630234
Download [GenBank](#) [FASTA](#)
2. **NT_011387.8**
Range 4607069..4622234
Download [GenBank](#) [FASTA](#)

Alternate assembly (based on Celera assembly)

Genomic

1. **AC_000063.1 Alternate assembly (based on Celera assembly)**
Range 4736784..4751948
Download [GenBank](#) [FASTA](#)
2. **NW_927317.1**
Range 4593960..4609124
Download [GenBank](#) [FASTA](#)

Related Sequences



Nucleotide	Protein
Genomic AF030575.1	AAC05365.1
Genomic AF076976.1	AAD46098.1
Genomic AF085477.2	AAC62750.2
Genomic AF315723.1	None
Genomic AL133396.2	CAB75503.1
	CAI19053.1
Genomic AY219882.1	AAO83635.1
Genomic AY219883.1	AAO83636.1
Genomic AY458651.1	AAR21603.1
Genomic CH471133.3	FAX10449.1
	FAX10450.1
Genomic DQ408531.1	ABD63004.1
Genomic M81929.1	AAB59442.1
Genomic M81930.1	AAB59443.1
Genomic S71208.1	AAB20521.1
Genomic S71210.1	AAB20522.1
Genomic S71212.1	AAB20523.1
Genomic S79978.1	AAB35416.1
Genomic S80539.1	AAB21334.1
Genomic S80732.1	AAB50648.2
Genomic S80743.1	AAB50649.2
Genomic S83341.1	AAB50777.1
Genomic U29185.1	AAC78725.1
Genomic X83416.1	CAA58442.1

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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
 [*Homo sapiens*]
 GeneID: 5621 updated 07-Oct-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 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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Camelus taurus **Pmp** prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)

Mus musculus **Pmp** prion protein

Rattus norvegicus **Pmp** prion protein

Gallus gallus **PRNP** prion protein (p27-30)

Proteins Proteins used in sequence comparisons and their conserved domain architectures.

- NP_898902.1 253 aa
- NP_001009093.1 253 aa
- XP_542906.2 257 aa
- NP_035300.1 254 aa
- NP_036763.1 254 aa
- NP_990796.1 267 aa

NCBI HomoloGene Discover Homologs

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All: 1 Fungi: 0 Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota

Alignment Scores

Species	Gene	Symbol	Identity (%)		Substitution Rates ¹			Blast
			Protein	DNA	d	d _N /d _S	d _{NR} /d _{NC}	
Homo sapiens PRNP								
vs. Pan troglodytes	PRNP		99.2	99.2	0.008	0.138	0.548	Blast
vs. Canis lupus familiaris	PrP		87.7	87.3	0.139	0.126	0.282	Blast
vs. Mus musculus	Pmp		90.1	85.3	0.163	0.077	0.342	Blast
vs. Rattus norvegicus	Pmp		89.7	86.8	0.145	0.090	0.260	Blast
vs. Gallus gallus	PRNP		47.1	57.4	0.631	0.399	0.878	Blast
Pan troglodytes PRNP								
vs. Homo sapiens	PRNP		99.2	99.2	0.008	0.138	0.548	Blast
vs. Canis lupus familiaris	PrP		87.7	87.3	0.139	0.126	0.253	Blast
vs. Mus musculus	Pmp		90.1	85.2	0.165	0.075	0.307	Blast
vs. Rattus norvegicus	Pmp		89.7	86.7	0.146	0.088	0.229	Blast
vs. Gallus gallus	PRNP		47.1	56.9	0.642	0.351	0.882	Blast
Canis lupus familiaris PrP								
vs. Homo sapiens	PRNP		87.7	87.3	0.139	0.126	0.282	Blast

NCBI Blast 2 Sequences results

BLAST 2 SEQUENCES RESULTS VERSION BLASTP 2.2.17 [Aug-26-2007]

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|34335270|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|69961... Length = 253 (1 .. 253)

Sequence 2: gi|13173473|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 Prmpa [Mus musculus] >gi|13879449|gb|AAH06703.1| Prion protein [Mus musculus] >gi|71060019|emb|CAJ18553.1| Pmp [Mus musculus] >gi|74182795|dbj|BAE34724.1| unnamed protein product [Mus musculus] >gi|74186646|dbj|BAE34788.1| unnamed protein product [Mus musculus] >gi|74192797... Length = 254 (1 .. 254)

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

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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
 [*Homo sapiens*]
 GeneID: 5621 updated 07-Oct-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 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.

Genomic regions, transcripts, and products

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NCBI Single Nucleotide Polymorphism

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

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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	View snp on GeneModel
NM_183079	plus strand	NP_898902	forward	NT_011387	reference	View snp on GeneModel
NM_183079	plus strand	NP_898902	forward	NW_927317	Celera	View snp on GeneModel

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#	Hetero-cluster id	Hetero-zygosity	Validation	3D	OMIM	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos
exon_1	4619867	101							start codon				1
exon_2	4619942	176	rs11538755		N.D.				nonsynonymous	A	Thr [T]	1	26
					N.D.				contig reference	C	Pro [P]	1	26
	4619949	183	rs11538762		N.D.				nonsynonymous	A	His [H]	2	28
					N.D.				contig reference	C	Pro [P]	2	28
	4619975	209	rs11538763		N.D.				synonymous	A	Arg [R]	1	37
					N.D.				contig reference	C	Arg [R]	1	37

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

Online Mendelian Inheritance in Man

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***176640**

PRION PROTEIN; PRNP

Alternative titles; symbols

PRP
PRION-RELATED PROTEIN; PRIP

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

TEXT

DESCRIPTION

The PRNP gene encodes the prion protein, which has been implicated in various types of transmissible neurodegenerative spongiform encephalopathies. The human prion diseases occur in inherited, acquired, and sporadic forms. Approximately 15% are inherited and associated with coding mutations in the PRNP gene. Inherited prion diseases include familial Creutzfeldt-Jakob disease (CJD, [123400](#)), Gerstmann-Strausler disease (GSD, [137440](#)), and fatal familial insomnia (FFI, [600072](#)). Acquired prion diseases include iatrogenic CJD, kuru ([245300](#)), variant CJD (vCJD) in humans, scrapie in sheep, and bovine spongiform encephalopathy (BSE) in cattle. Prion diseases are also referred to as transmissible spongiform encephalopathies (TSE). Variant CJD is believed to be acquired from cattle infected with BSE. However, the majority of human cases of prion disease occur as sporadic CJD (sCJD) ([Collinge et al., 1996](#); [Parchi et al., 2000](#); [Hill et al., 2003](#)).

CLONING

[Oesch et al. \(1985\)](#) isolated a cDNA clone corresponding to a pathogenic PrP fragment from a scrapie-infected hamster brain cDNA library. Southern blotting with PrP cDNA revealed a single gene with the same restriction patterns in normal and scrapie-infected brain DNA. A single PrP-related gene was also detected in murine and human DNA. Proteinase K digestion yielded PrP 27-30 in infected brain extract, but completely degraded the PrP-related protein in normal brain extract.

[Kretzschmar et al. \(1986\)](#) isolated a PRNP cDNA from a human retina cDNA library. The 253-amino acid protein shared 90% amino acid sequence identity with the hamster protein. Northern blot analysis detected a 2.5-kb mRNA in a variety of human neuroectodermal cell lines.

[Basler et al. \(1986\)](#) determined that the pathogenic PrP protein in scrapie and normal cellular PrP are encoded by the same gene. The PrP coding sequence encodes an amino-terminal signal peptide. The primary structure of PrP encoded by the gene of a healthy animal did not differ from that encoded by a cDNA from a scrapie-infected animal, suggesting that the

Entrez Gene

Nomenclature

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***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

NCBI

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PRION PROTEIN; PRNP

Alternative titles; symbols

PRP
PRION-RELATED PROTEIN; PRIP

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

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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-Straussler disease, Huntington disease-like 1, and kuru. Two transcript variants encoding the same protein have been found for this gene.

- Nucleotide
- OMIA
- OMIM
- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
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- HPRD
- KEGG
- MGC
- ModelMaker
- PharmGKB
- UniGene
- LinkOut

Genomic regions, transcripts, and products

Go to [reference sequence details](#)



- Links
- FASTA
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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

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

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

Matching gi: [15108368](#), [122056623](#), [122056625](#), [122056628](#), [1828059](#), [3996152](#), [3999510](#), [4000210](#), [336](#), [5389](#), [34335270](#), [130912](#), [190468](#), [111961006](#), [112019514](#), [31674692](#), [119630854](#), [119630855](#), [62774377](#), [83553219](#), [46095329](#), [155100459](#), [53934492](#), [10053355](#), [11079226](#), [17908793](#), [20251002](#), [21506484](#), [590](#), [964](#), [6996155](#), [40075008](#)

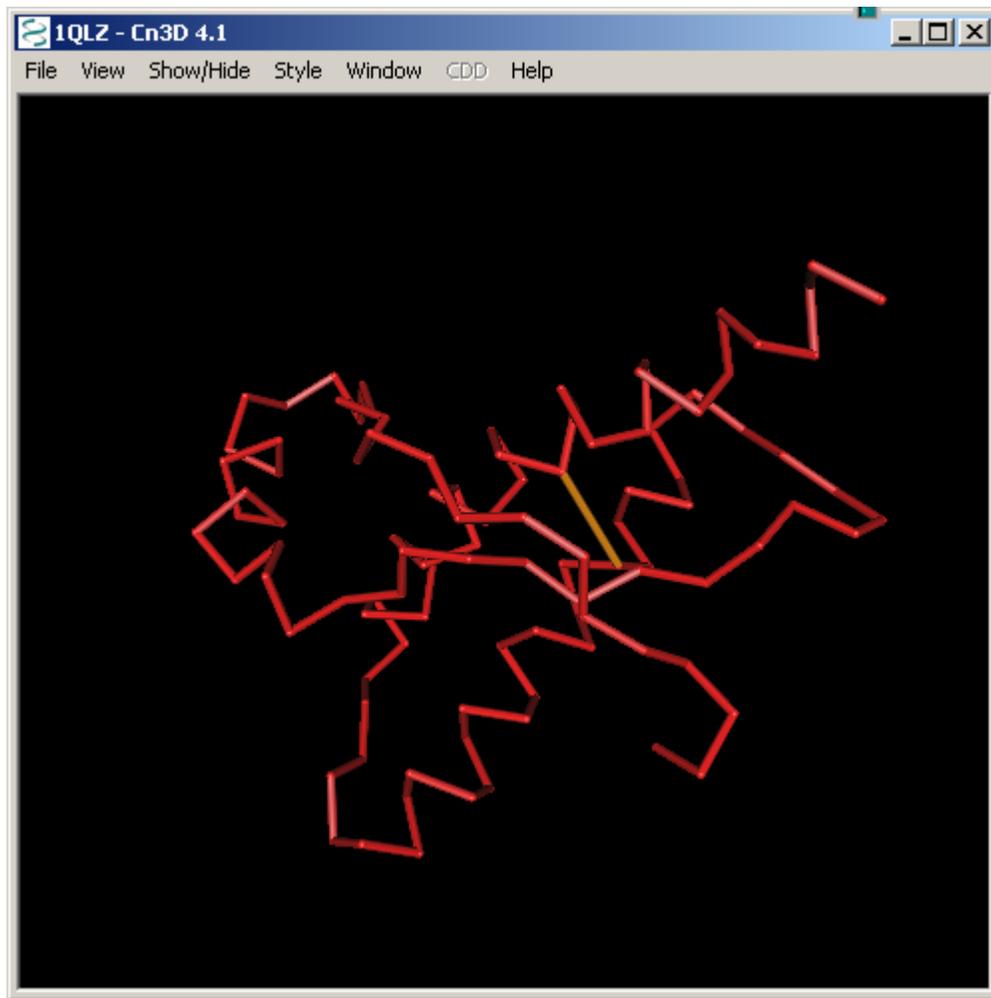
Hide identical
Best hits
Common Tree
Taxonomy Report
3D structures
CDD-Search
GI list
Run BLAST

200 BLAST hits to 52 unique species [Sort by taxonomy proximity](#)

Archaebacteria
Bacteria
192 Metazoa
Fungi
Plants
Viruses
Other Eukaryotes

Keep only: Cut-Off: Select New search by GI:

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
Conserved Domain Database hits				
1435	31	NP_001...	122056623	prion protein preproprotein [Homo sapiens]
1435	31	NP_001...	122056628	prion protein preproprotein [Homo sapiens]
1435	31	NP_001...	122056625	prion protein preproprotein [Homo sapiens]
1435	31	NP_898902	34939370	prion protein preproprotein [Homo sapiens]
1435	31	P04156	130912	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR) (CD230 antigen)
1435	31	AAH60182	190468	prion protein
1435	31	EAX10449	119630854	prion protein (p27-30) (Creutzfeld-Jakob disease, Gerstmann-Straussler-Scheinker syndrome, fatal fam
1435	31	EAX10450	119630855	prion protein (p27-30) (Creutzfeld-Jakob disease, Gerstmann-Straussler-Scheinker syndrome, fatal fam
1435	31	AAS80162	46095329	prion protein [Homo sapiens]
1435	31	AAG21693	11079226	prion protein [Homo sapiens]
1435	31	CAB75503	6996155	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Straussler-Scheinker syndrome, fatal fa
1432	29	AAC50089	474359	prion protein
1432	29	P40252	730390	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen)
1432	1	AAK42953	61967107	prion protein [synthetic construct]
1432	1	AAV32822	54695820	prion protein (p27-30) (Creutzfeld-Jakob disease, Gerstmann-Straussler-Scheinker syndrome, fatal fam
1432	1	AAV37058	60834334	prion protein [synthetic construct]
1431	1	ABM82244	123980830	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Straussler-Scheinker syndrome, fatal fa
1431	1	ABM85428	123985653	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Straussler-Scheinker syndrome, fatal fa
1431	31	CAG46869	49457097	PRNP [Homo sapiens]
1431	31	AAH12844	15277486	Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Straussler-Scheinker syndrome, fatal fa
1430	31	ABD63004	89160954	prion protein PrP [Homo sapiens]
1427	29	AAC50085	474351	prion protein
1427	27	AAC50088	474357	prion protein
1427	27	AAC50086	474378	prion protein



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?