

## 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

Entrez Gene is the successor to LocusLink. 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

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 ccids" property added. [News archives...](#)

Sample Searches

Find genes by...	Search text
free text	<a href="#">human muscular dystrophy</a>
partial name and multiple species	<a href="#">transporter[title] AND ("Drosophila melanogaster"[organism] OR "Mus musculus"[organism])</a>

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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  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: 48 Current Only: 36 Genes Genomes: 36 SNP GeneView: 36

Items 1 - 20 of 48 Page 1 of 3 Next

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  
**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  
**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, HSPA5, GRP78, HSP70

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Limits Preview/Index History Clipboard Details

Display Full Report Show 5 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 11-Mar-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** [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-Straussler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.

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

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- OMIA
- OMIM
- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView

Full Report Summary Brief ASN.1 XML Gene Table Gene Table UI List LinkOut Books Links Conserved Domain Links Genome Links GENSAT Links GEO Profile Links HomoloGene Links Nucleotide Links NIH cDNA clone links OMIA Links OMIM Links BioAssay Links PMC Links

Display: Gene Table Show: 5 Send to: [dropdown]

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 11-Mar-2007

RefSeq status: Reviewed  
total gene size: 15166 bp

**Entrez Gene Home**

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- Evidence Viewer
- GDB
- GeneTests for MIM: 176640
- HGMD
- HGNC
- HPRD
- KEGG
- MGC
- ModelMaker
- PharmGKB
- UniGene
- LinkOut

**Genomic regions, transcripts, and products** ?

Go to [reference sequence details](#)

mRNA	bp	exons	Protein	aa	exons
<a href="#">NM_000311.2</a>	2468	2	<a href="#">NP_000302.1</a>	254	1
<a href="#">NM_183079.1</a>	2464	2	<a href="#">NP_898902.1</a>	254	1

**Exon information:**

[NM\\_000311.2](#) length: 2468 bp, number of exons: 2

[NP\\_000302.1](#) length: 253 aa, number of exons: 1

EXON	length	Coding EXON	length	INTRON	length
<a href="#">1 - 90</a>	90 bp	<a href="#">91 - 12788</a>	12698 bp		
<a href="#">12789 - 15166</a>	2378 bp	<a href="#">12799 - 13560</a>	762 bp		

[NM\\_183079.1](#) length: 2464 bp, number of exons: 2

[NP\\_898902.1](#) length: 253 aa, number of exons: 1

EXON	length	Coding EXON	length	INTRON	length
<a href="#">1 - 86</a>	86 bp	<a href="#">87 - 12788</a>	12702 bp		
<a href="#">12789 - 15166</a>	2378 bp	<a href="#">12799 - 13560</a>	762 bp		

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- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP

**Genomic regions, transcripts, and products** ?

Go to [reference sequence details](#)

**Genomic context** ↑ ?

chromosome: 20; Location: 20p13 See PRNP in MapViewer

[HGMD](#)  
[HGNC](#)  
[HPRD](#)  
[KEGG](#)  
[MGC](#)  
[ModelMaker](#)  
[PharmGKB](#)  
[UniGene](#)  
[LinkOut](#)

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Related Articles in PubMed

[PubMed](#) links

GeneRIFs: Gene References Into Function What's a GeneRIF?

1. results suggest that the PRNP genetic variants are not associated with the risk for Alzheimer's disease in Korean population
2. first study that provides experimental evidence supporting the hypothesis that there might be silent prions lying dormant in normal human brains
3. 1-OPRD (one octapeptide-repeat deletion) homozygosity or heterozygosity exists in several gastric cancer cell lines, e.g. MKN28 and KatoIII are homozygous for 1-OPRD, and SGC7901 and BGC-823 are heterozygous for 1-OPRD.
4. Polymorphic microsatellite sites within 148 kb of the human prion gene complex, including the genes PRNP, PRND and PRNT, were analysed together with the Codon129 variants regarding 50 Creutzfeldt-Jakob Disease patients and 46 non-diseased control persons.
5. The highest affinity copper (II)-binding modes cause self-association of both peptides, suggesting a role for copper (II) in controlling prion protein self-association in vivo
6. there is a link between ER stress and the formation of cytosolic PrP isoforms potentially endowed with novel signaling or cytotoxic functions
7. prion protein may act as an inhibitor of microtubule assembly by inducing formation of stable tubulin oligomers
8. At pH 5.5 the prion protein binds 2 Cu<sup>2+</sup> ions. All 6 histidines of the unfolded N-terminal domain & the N-terminal amino

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** ↑ ?

Description .....	Product	Interactant	Other Gene	Complex	Source	Pubs
	NP_000302.1	<a href="#">NP_001155.1</a>	<a href="#">APBB1</a>		<a href="#">HPRD</a>	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_055759.2</a>	<a href="#">CLSTN1</a>		<a href="#">HPRD</a>	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_001822.2</a>	<a href="#">CLU</a>		<a href="#">HPRD</a>	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_001834.2</a>	<a href="#">CNTN1</a>		<a href="#">HPRD</a>	<a href="#">PubMed</a>
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	<a href="#">NP_001886.1</a>	<a href="#">CSNK2A1</a>		<a href="#">BIND</a>	<a href="#">PubMed</a>
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	<a href="#">NP_001887.1</a>	<a href="#">CSNK2A2</a>		<a href="#">BIND</a>	<a href="#">PubMed</a>
PrPc interacts with CSNK2B (CK2 beta) albeit weakly. This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2B (CK2 beta).						

**General gene information**  

**Markers**

**WI-19738(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**

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
<a href="#">plasma membrane</a>	ISS

**General protein information**  

**Names**

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

NCBI Entrez Gene

Search: Gene for [ ] Go Clear

Display: Full Report Show: 5 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 05-Feb-2007

**Summary**

**Official Symbol** PRNP provided by HGNC

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**Primary source** HGNC:9449

**See related** 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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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** Download, Links

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	LOC396452	prion protein (p27-30) (Creutzfeldt-Jakob ...



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*Discover Homologs*

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**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>


**Blast 2 Sequences results**

[PubMed](#)
[Entrez](#)
[BLAST](#)
[OMIM](#)
[Taxonomy](#)
[Structure](#)

**BLAST 2 SEQUENCES RESULTS VERSION BLASTP 2.2.14 [May-07-2006]**

Matrix:  gap open:  gap extension:

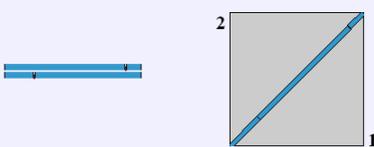
x\_dropoff:  expect:  wordsize:    View option

Masking character option  for protein, n for nucleotide  Masking color option

Show CDS translation

**Sequence 1:** [gi|34335270|ref|NP\\_898902.1|](#) prion protein preproprotein [Homo sapiens] > [gi|4506113|ref|NP\\_000302.1|](#) 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|emb|CAB75503.1|](#) PRNP [Homo sapiens] > [gi|11079226|gb|AAG21693.1|](#) prion protein [Homo sapiens] > [gi|46095329|gb|AAS80162.1|](#) prion protein [Homo sapiens]  
 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|74192797...](#)  
 Length = 254 (1 .. 254)



Limits Preview/Index History Clipboard Details

Display: Full Report Show: 5 Send to:

All: 1 Current Only 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 05-Feb-2007

Entrez Gene Home

**Table Of Contents**

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- Genomic context
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- Interactions
- General gene information
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**Links** Explain

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- Nucleotide
- OMIA
- OMIM
- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView

**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** [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

Go to [reference sequence details](#)

NCBI Single Nucleotide Polymorphism

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search Entrez SNP for [ ] Go

**SNP linked to Gene (geneID:5621)**

SNP are linked from gene [PRNP](#) via the following methods:

[Contig Annotation](#) [GenBank\(mrna\) Mapping](#)

Send all rs# 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	snp list
<a href="#">NM_000311</a>	plus strand	<a href="#">NP_000302</a>	forward	<a href="#">NT_011387</a>	reference	currently shown
<a href="#">NM_000311</a>	plus strand	<a href="#">NP_000302</a>	forward	<a href="#">NW_927317</a>	Celera	<a href="#">view</a>
<a href="#">NM_183079</a>	plus strand	<a href="#">NP_898902</a>	forward	<a href="#">NT_011387</a>	reference	<a href="#">view</a>
<a href="#">NM_183079</a>	plus strand	<a href="#">NP_898902</a>	forward	<a href="#">NW_927317</a>	Celera	<a href="#">view</a>

in gene region  cSNP  has frequency  double hit  haplotype tagged

gene model	Contig Label	Contig	mRNA	protein	mRNA orientation	transcript	snp count
(contig mRNA transcript):	reference	<a href="#">NT_011387</a>	<a href="#">NM_000311</a>	<a href="#">NP_000302</a>	forward	plus strand 21, coding	

**Color Legend**

Region	Contig position	mRNA pos	dbSNP rs# cluster id	Heterozygosity	Validation	3D	OMIM	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos
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

SEARCH

			N.D.		Yes		contig reference	A	Asp [D]	2	178
	4620405	<a href="#">rs11538767</a>	N.D.		Yes		nonsynonymous	C	Ala [A]	2	180
			N.D.		Yes		contig reference	T	Val [V]	2	180
	4620464	<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	<a href="#">rs1800014</a>	N.D.		Yes		nonsynonymous	A	Lys [K]	1	219
			N.D.		Yes		contig reference	G	Glu [E]	1	219
	4620538	<a href="#">rs6052773</a>	N.D.		Yes		synonymous	T	Ala [A]	3	224
			N.D.		Yes		contig reference	C	Ala [A]	3	224
	4620545	<a href="#">rs17852079</a>	N.D.		Yes		nonsynonymous	A	Lys [K]	1	227
			N.D.		Yes		contig reference	C	Gln [Q]	1	227
	4620625	<a href="#">rs11538759</a>	N.D.				synonymous	G	Gly [G]	3	253
			N.D.				contig reference	A	Gly [G]	3	253


**.0006 CREUTZFELDT-JAKOB DISEASE [PRNP, GLU200LYS] dbSNP**

FATAL FAMILIAL INSOMNIA, INCLUDED

**MIM \*176640**  
 Description  
 Cloning  
 Gene Structure  
 Mapping  
 Gene Function  
 Molecular Genetics  
 Genotype/Phenotype  
 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. ☹

Molecular Genetics  
 Genotype/Phenotype Correlations  
 Population Genetics  
 Animal Model  
 History  
 Allelic Variants  
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• Gene map

Entrez Gene  
 N Nomenclature  
 R RefSeq  
 G GenBank  
 P Protein  
 U UniGene

LinkOut  
 HGVS  
 HGMD

All: 1 OMIM dbSNP: 1 OMIM UniSTS: 0

**\*176640** GeneTests, Links  
**PRION PROTEIN; PRNP**

**ALLELIC VARIANTS**  
 (selected examples)

- [0001 CREUTZFELDT-JAKOB DISEASE](#) [PRNP, EXTRA OCTAPEPTIDE CODING REPEATS]
- [0002 GERSTMANN-STRAUSSLER DISEASE](#) [PRNP, PRO102LEU]
- [0003 REMOVED FROM DATABASE](#)
- [0004 GERSTMANN-STRAUSSLER DISEASE](#) [PRNP, ALA117VAL]
- [0005 PRION DISEASE, SUSCEPTIBILITY TO](#) [PRNP, MET129VAL] **dbSNP**
- [0006 CREUTZFELDT-JAKOB DISEASE](#) [PRNP, GLU200LYS] **dbSNP**
- [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]
- [0018 SPONGIFORM ENCEPHALOPATHY WITH NEUROPSYCHIATRIC FEATURES](#) [PRNP, ASN171SER ] **dbSNP**
- [0019 CREUTZFELDT-JAKOB DISEASE, PROTECTION AGAINST](#) [PRNP, GLU219LYS]
- [0020 REMOVED FROM DATABASE](#)

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OMIM  
 Online Mendelian Inheritance in Man

Johns Hopkins University

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All: 1

**\*176640** GeneTests, Links  
**PRION PROTEIN; PRNP**

*Alternative titles; symbols*

**PRP**  
**PRION-RELATED PROTEIN; PRIP**

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

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-Straussler 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](#)

**Genomic context**

chromosome: 20; Location: 20p13

**Bibliography**

Related Articles in Pubmed

PubMed links

GeneRIFs: Gene References Into Function

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

**Entrez Gene Info**

**Feedback**

**Subscriptions**

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  
LinkOut

NCBI

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

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

Matching gi: [112019514](#), [111961006](#), [83353219](#), [62774377](#), [53934492](#), [46095329](#), [40075008](#), [34335270](#), [33695389](#), [31674692](#), [21506484](#), [20251002](#), [17908793](#), [15108368](#), [11079226](#), [10053355](#), [6996155](#), [5993964](#), [400020](#), [3999540](#), [3996152](#), [1828059](#), [190468](#), [130912](#)

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200 BLAST hits to 48 unique species [Sort by taxonomy proximity](#)

Archaea Bacteria 194 Metazoa Fungi Plants Viruses Other Eukaryotae

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

253 aa

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
<b>Conserved Domain Database hits</b>				
1435	31	AAS80162	46095329	prion protein [Homo sapiens]
1435	31	NP_898902	34335270	prion protein preproprotein [Homo sapiens]
1435	31	AAG21693	11079226	prion protein [Homo sapiens]
1435	31	CAB75503	6996155	PRNP [Homo sapiens]
1435	31	AAA60182	190468	prion protein
1435	31	P04156	130912	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR)
1432	29	P40252	730390	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230)
1432	29	AAC50089	474359	prion protein
1432	1	AAX37089	60834334	prion protein [synthetic construct]
1432	1	AAX42953	61367107	prion protein [synthetic construct]
1432	1	AAV38282	54695820	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strau)
1431	31	CAG46869	49457097	PRNP [Homo sapiens]
1431	31	AHH12844	15277486	Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Stra)
1430	31	ABD63004	89160954	prion protein PrP [Homo sapiens]
1427	31	AHH22532	18490397	Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Stra)
1427	29	NP_001...	57114055	prion protein [Pan troglodytes]
1427	29	AAV74306	56122310	prion protein [Pan troglodytes]
1427	29	P61768	48429057	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230)
1427	27	P61767	48429056	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230)

NCBI

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

Query: gi:4506113 prion protein preproprotein [Homo sapiens]  
 Matching gi: 112019514, 111961006, 83353219, 62774377, 53934492, 46095329, 40075008, 34335270, 33695389, 31674692, 21506484, 20251002, 17908793, 15108368, 11079226, 10053355, 6996155, 5993964, 4000210, 3999540, 3996152, 1828059, 190468, 130912

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43 BLAST hits to 12 unique species [Sort by taxonomy proximity](#)

0 Archaea 0 Bacteria 43 Metazoa 0 Fungi 0 Plants 0 Viruses 0 Other Eukaryotae

Keep only  Cut-Off 100   New search by GI: 4506113

253 aa

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
<a href="#">Conserved Domain Database hits</a>				
1203	0	1QLZA	6730487	Chain A, Human Prion Protein
1203	0	1QLXA	6730485	Chain A, Human Prion Protein
1136	0	1DX1A	9955174	Chain A, Bovine Prion Protein Residues 23-230
1136	0	1DX0A	9955173	Chain A, Bovine Prion Protein Residues 23-230
771	0	1QM1A	6730489	Chain A, Human Prion Protein Fragment 90-230
771	0	1QMOA	6730488	Chain A, Human Prion Protein Fragment 90-230
768	0	1FO7A	10835618	Chain A, Human Prion Protein Mutant E200k Fragment 90-231
768	0	1FKCA	10835617	Chain A, Human Prion Protein (Mutant E200k) Fragment 90-231
698	0	1B10A	6729981	Chain A, Solution Nmr Structure Of Recombinant Syrian Hamster Prion Protein
604	0	1QM3A	6730491	Chain A, Human Prion Protein Fragment 121-230
604	0	1QM2A	6730490	Chain A, Human Prion Protein Fragment 121-230
587	0	1I4MA	20150089	Chain A, Crystal Structure Of The Human Prion Protein Reveals A M
584	0	1DWZA	9955172	Chain A, Bovine Prion Protein Fragment 121-230
584	0	1DWYA	9955171	Chain A, Bovine Prion Protein Fragment 121-230
583	0	1H0LA	28373307	Chain A, Human Prion Protein 121-230 M166cE221C
576	0	1Y2SA	60594516	Chain A, Ovine Prion Protein Variant R168
569	0	1XYUA	60594486	Chain A, Solution Structure Of The Sheep Prion Protein With Polym

NCBI

# Related Structures

HOME SEARCH SITE MAP PubMed Blast Entrez Structure Help

Query: prion protein preproprotein [Homo sapiens]  
[gi: 4506113]

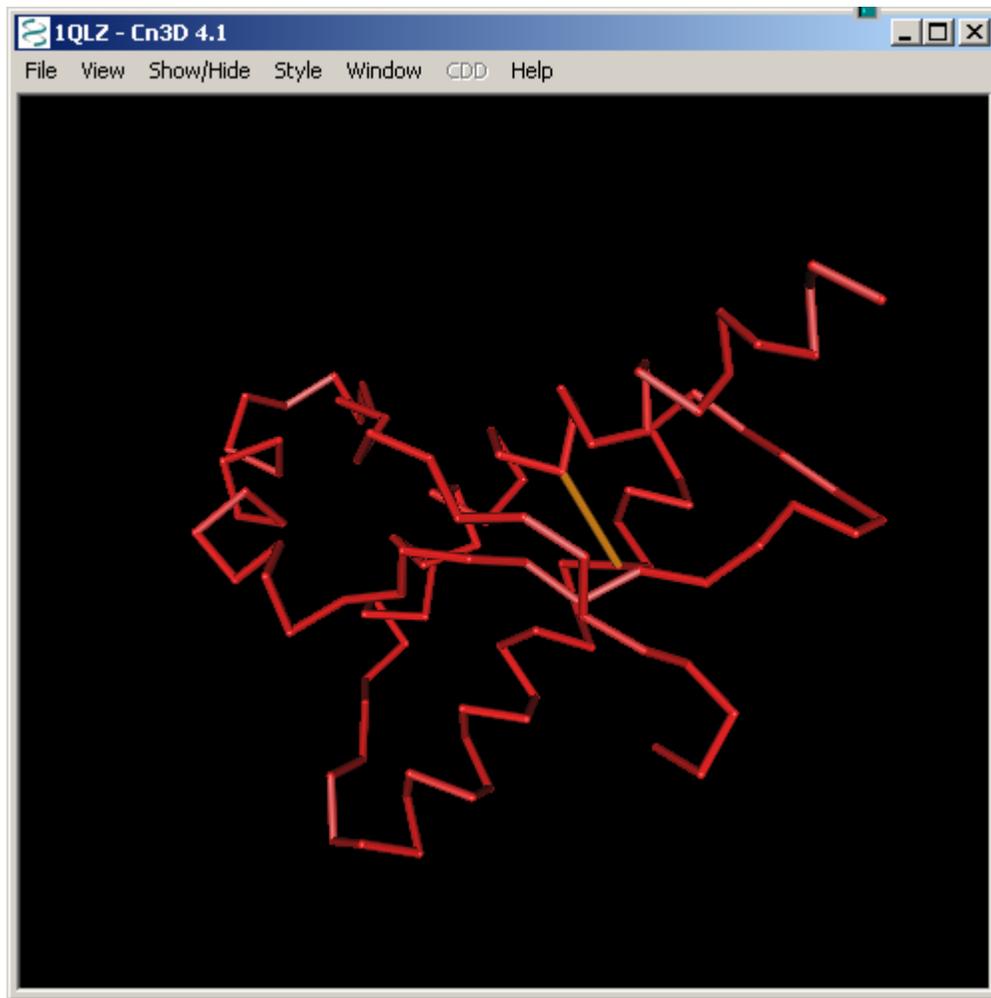
Structure: 1QLZ Chain A, Human Prion Protein

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
gi_4506113	23	KKRKPKGGWNTGGSRYPGQSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP	102						
1QLZ_A	3	KKRKPKGGWNTGGSRYPGQSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP	82						
		90	100	110	120	130	140	150	160
gi_4506113	103	SKPKTNMKHMAGAAAAGAVVGGGLGGYMLGSAMSRP I I HFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNI	182						
1QLZ_A	83	SKPKTNMKHMAGAAAAGAVVGGGLGGYMLGSAMSRP I I HFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNI	162						
		170	180	190	200				
gi_4506113	183	TIKQHTVTTTTKGENFTE TDVKMMERVVEQMCITQYERESQAYYQRGS	230						
1QLZ_A	163	TIKQHTVTTTTKGENFTE TDVKMMERVVEQMCITQYERESQAYYQRGS	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.