

## 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 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

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Display Summary Show 20 Send to

All: 52 Current Only: 40 Genes Genomes: 40 SNP GeneView: 39

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1: TP53 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 Location: 17p13.1 Chromosome: 17 Annotation: NC\_000017.9 (7531641..7512463, complement) MIM: 191170 GeneID: 7157

2: PRNP 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 Location: 20p13 Chromosome: 20 Annotation: NC\_000020.9 (4615068..4630233) MIM: 176640 GeneID: 5621

3: HSPA5 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 Location: 9q33-q34.1 Chromosome: 9 Annotation: NC\_000009.10 (127043429..127036952, complement) MIM: 138120 GeneID: 3309

NCBI Entrez Gene

Search Gene for [ ] Go Clear

Limits: Preview/Index History Clipboard Details

Display: Full Report Show 5 Send to

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

GeneID: 5621 updated 22-Apr-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-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](#)

**Genomic regions, transcripts, and products**

Go to [reference sequence details](#)

■ coding region ■ untranslated region

**Genomic context**

chromosome: 20; Location: 20p13

See PRNP in MapViewer

**Entrez Gene Info**

**Feedback**

**Subscriptions**

- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- CCDS
- Evidence Viewer
- GDB
- GeneTests for MIM: 176640
- HGMD
- HGNC
- HPRD
- KEGG
- MGC
- ModelMaker
- PharmGKB
- UniGene
- LinkOut

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1: ASN.1 Hom XML

Gene Gene Table updated 22-Apr-2007

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

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Search Gene for [ ] Go Clear

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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) [ [Entrez Gene Home](#) ]

Homo sapiens ]

GeneID: 5621 updated 22-Apr-2007

RefSeq status: Reviewed

total gene size: 15166 bp

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

mRNA

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	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	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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- PubMed
- PubMed (GeneRIF)
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Search: Gene for [Go] [Clear]

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Display: Gene Table Show: 5 Send to

All: 1 Summary Brief ASN.1 XML Gene Table UJ List LinkOut Books Links Conserved Domain Links

Gene Table

RefSeq total

Genes

Genome Links GENSAT Links GEO Profile Links HomoloGene Links Nucleotide Links NIH cDNA clone links OMIA Links OMIM Links BioAssay Links PMC Links

Genomes: 1 SNP GeneView: 1

30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [Entrez Gene Home] updated 22-Apr-2007

Links: Order cDNA clone Books Conserved Domains Genome GEO Profiles HomoloGene Map Viewer Nucleotide OMIA OMIM Full text in PMC Probe Protein PubMed PubMed (GeneRIF) SNP

and products

NC\_000020\_9

[4619609] 5' [4639234] 3'

NP\_088382.1 prnp protein CC813989.1

NP\_835922.1 prnp protein CC813989.1

■ - coding region ■ - untranslated region

PharmGKB UniGene LinkOut

Entrez Gene Info Feedback Subscriptions

**Genomic context** [?] [↑]

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

[4519609] RPL79L2 [4669314] PRNP PRND PRNT

**Bibliography** [?] [↑]

Related Articles in PubMed

PubMed links

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

1. 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.
2. Oxidative stress might be an influence that leads to substantial structural conversions of PrP in vivo.
3. prion protein does not require other Bcl-2 family proteins to protect against Bax-mediated cell death
4. 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.
5. Equilibrium binding and kinetics of FRET show that the PRNP binding to the oligonucleotides and their bending occur simultaneously.
6. analysis of experimentally derived constraints for high-resolution structural models of PrP amyloid fibrils
7. p53-dependent staurosporine-induced caspase-3 activation is affected by the C-terminal products of cellular prion protein processing, C1 and C2
8. According to PrP gene polymorphism and PrP type, 18 cases were classified as MM1-type, two as MV1-type, two as MM2-type and one as MM1 + 2-type sporadic Creutzfeldt-Jakob disease.

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-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)**

- 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](#)

<a href="#">smart00157</a>	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
Location:23-230	
Blast Score:546	
- 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,RP251427,DA122620,M13899](#)

Conserved Domains (1): [summary](#)

<a href="#">smart00157</a>	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
Location:23-230	
Blast Score:546	
- 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,RI669189,DA297032,M13899](#)
- 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](#)

<a href="#">smart00157</a>	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
Location:23-230	
Blast Score:546	
- 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.

**RefSeqs of Annotated Genomes: Build 36.2**

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

**Reference assembly**

**Genomic**

- NC\_000020.9 Reference assembly**

Range: 4615069..4630234

Download: [GenBank](#) [FASTA](#)
- NT\_011387.8**

Range: 4607069..4622234

Download: [GenBank](#) [FASTA](#)

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

**Genomic**

- AC\_000063.1 Alternate assembly (based on Celera assembly)**

Range: 4736784..4751948

Download: [GenBank](#) [FASTA](#)
- NW\_927317.1**

Range: 4593960..4609124

Download: [GenBank](#) [FASTA](#)

**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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Search Gene for [Go] [Clear]

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GeneID: 5621 updated 22-Apr-2007

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**Genomic regions, transcripts, and products**

Go to [reference sequence details](#)

NC\_000020.9

[4615869] 5' [4638294] 3'

ML\_428711.2  
ML\_153872.1

NP\_488702.1 prnp protein CC011288.1  
NP\_538921.1 prnp protein CC011288.1

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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	Pmp	prion protein
R.norvegicus	Pmp	prion protein
G.gallus	PRNP	prion protein (p27-30)

About Entrez

HomoloGene

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Genome Resources

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- Mus musculus



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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**

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[Entrez](#)
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[OMIM](#)
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**BLAST 2 SEQUENCES RESULTS VERSION BLASTP 2.2.14 [May-07-2006]**

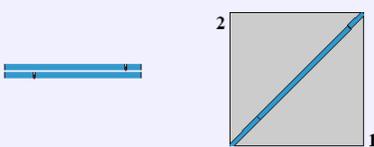
Matrix:  gap open:  gap extension:   
 x\_dropoff:  expect:  wordsize:    View option

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)



NCBI Entrez Gene

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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 22-Apr-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** HPPD:01453; MIM:176640

**Gene type** protein coding

**RefSeq status** Reviewed

**Organism** [Homo sapiens](#)

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

**Also known as** CJD; GSS; PrP; ASCR; PrP<sup>Sc</sup>; PrP<sup>C</sup>; 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](#)

Entrez Gene Home

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- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AcView
- CCDS
- Evidence Viewer

NCBI Single Nucleotide Polymorphism

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

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	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	<a href="#">rs16990018</a>	0.046			Yes	nonsynonymous	G	Ser [S]	2	171
			0.046			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	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

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

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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See PRNP in MapViewer

Links

- FASTA
- GENPEPT
- Blink
- Conserved Domains

NCBI

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

Query: gi:4506113 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 Eukaryotes

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

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
<u>Conserved Domain Database hits</u>				
1435	31	CBT5503	4996155	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1435	31	AAQ21693	11079226	prion protein [Homo sapiens]
1435	31	AA580162	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	EA010449	119630854	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1435	31	EA010450	119630855	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1432	29	AA050089	474359	prion protein
1432	29	P40282	130390	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen)
1432	1	AA042953	61367107	prion protein [synthetic construct]
1432	1	AA038282	54695820	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1432	1	AA037089	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: gi:4506113 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, 119630854, 119630855

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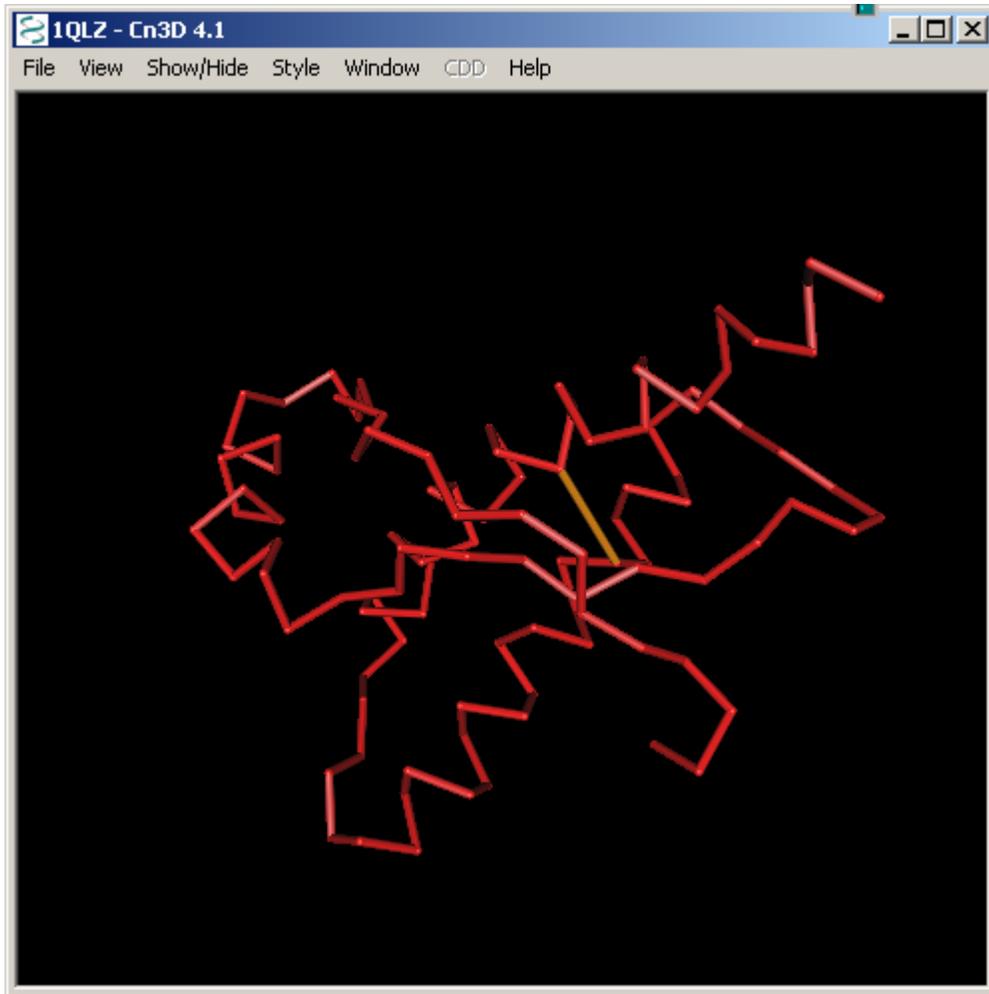
46 BLAST hits to 14 unique species Sort by taxonomy proximity

Archaea 0 Bacteria 44 Metazoa 0 Fungi 0 Plants 0 Viruses 0 Other Eukaryotes

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

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
<u>Conserved Domain Database hits</u>				
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	•	1QM1A	6730489	Chain A, Human Prion Protein Fragment 90-230
771	•	1QMOA	6730488	Chain A, Human Prion Protein Fragment 90-230
768	•	1FO7A	10835618	Chain A, Human Prion Protein Mutant E200k Fragment 90-231
768	•	1FRCA	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]
604	•	1QM3A	6730491	Chain A, Human Prion Protein Fragment 121-230
604	•	1QM2A	6730490	Chain A, Human Prion Protein Fragment 121-230
587	•	1LHMA	20150089	Chain A, Crystal Structure Of The Human Prion Protein Reveals A Mechanism For Oligomerization
584	•	1DN7A	9955172	Chain A, Bovine Prion Protein Fragment 121-230
584	•	1DN6A	9955171	Chain A, Bovine Prion Protein Fragment 121-230
583	•	1HOLA	28373307	Chain A, Human Prion Protein 121-230 M166cE221c
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





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.