

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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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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- 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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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](#)

RPL7R2 [4519659] RPS4L2 [4519659] PRNP [4615969] PRNP [4639234] PRNP [4649514]

SNP: Genotype
 SNP: GeneView
 Taxonomy
 UniSTS
 AceView
 CCDS
 Evidence Viewer
 GDB
 GeneTests for MIM: 176640
 HGMD
 HGNC
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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [Homo sapiens]

updated 07-Oct-2007

provided by HGNC

0171867; HPRD:01453; MIM:176640

Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Hominidae; Homo

CR; PRIP; PrPC; CD230; MGC26679; PrP27-30; PrP33-35C

encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to form beta-sheet-like structures. The encoded protein contains a highly unstable region of five tandem repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a highly and structurally similar protein to the one encoded by this gene. Mutations in the repeat

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- EST
- Nucleotide
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Search: Gene for [Go] [Clear]

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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 20-Oct-2007

RefSeq status: Reviewed total gene size: 15166 bp

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

NC_000020.9

■ - coding region ■ - untranslated region

mRNA	bp	exons	Protein	aa	exons
NM_000311.2	2468	2	NP_000302.1	253	1
NM_183079.1	2464	2	NP_898902.1	253	1

Exon information:

NM_000311.2 length: 2468 bp, number of exons: 2

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

EXON	Coding EXON	INTRON			
coords	length	coords	length	coords	length
1 - 90	90 bp	91 - 12788	12698 bp		
12789 - 15166	2378 bp	12789 - 13560	762 bp		

NM_183079.1 length: 2464 bp, number of exons: 2

NP_898902.1 length: 253 aa, number of exons: 1

EXON	Coding EXON	INTRON			
coords	length	coords	length	coords	length
1 - 86	86 bp	87 - 12788	12702 bp		

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- Probe
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- PubMed (GeneRIF)
- SNP
- SNP: Genotype
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- Taxonomy
- UniSTS
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- Ensembl
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GeneID: 5621 updated 07-Oct-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
NM_000311.2	2468	2	NP_000302.1	253	1
NM_183073.1	2464	2	NP_898902.1	253	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
1-90	90 bp			91-12788	12698 bp

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mRNA

mRNA	bp	exons	Protein	aa	exons
NM_000311.2	2468	2	NP_000302.1	253	1
NM_183073.1	2464	2	NP_898902.1	253	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
1-90	90 bp			91-12788	12698 bp

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. 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 PrP(C) interacts with selectins in a manner that is distinct from interactions in peripheral tissues; alterations 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 PrP(c) in vitro and the liberated NH(2)-terminal fragment accelerates plasminogen activation

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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	ITC6M		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-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
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																
<p>Names</p> <ul style="list-style-type: none"> prion protein CD230 antigen prion protein PrP major prion protein prion-related protein 																
NCBI Reference Sequences (RefSeq)																
<p>RefSeqs maintained independently of Annotated Genomes</p> <p>These reference sequences exist independently of genome builds. Explain</p>																
mRNA and Protein(s)																
<p>1. NM_000311.3–NP_000302.1 prion protein preproprotein</p> <table border="1"> <tr> <td>Description</td> <td colspan="2">Transcript Variant: This variant (1) represents the longest transcript. Variants 1-5 encode the same protein.</td> </tr> <tr> <td>Source sequence(s)</td> <td colspan="2">AW452130,BC022532,DA297032,M13899</td> </tr> <tr> <td>Consensus CDS</td> <td colspan="2">CCDS13080.1</td> </tr> <tr> <td>Conserved Domains (1)</td> <td>summary</td> <td></td> </tr> <tr> <td></td> <td>smart00157 Location:23–230 Blast Score:546</td> <td>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.</td> </tr> </table>		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.
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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.														
<p>4. NM_001080123.1–NP_001073592.1 prion protein preproprotein</p> <table border="1"> <tr> <td>Description</td> <td colspan="2">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.</td> </tr> <tr> <td>Source sequence(s)</td> <td colspan="2">BC022532,DB461478,M13899</td> </tr> <tr> <td>Conserved Domains (1)</td> <td>summary</td> <td></td> </tr> <tr> <td></td> <td>smart00157 Location:23–230 Blast Score:546</td> <td>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.</td> </tr> </table>		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.			
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.														
<p>5. NM_183079.2–NP_898902.1 prion protein preproprotein</p> <table border="1"> <tr> <td>Description</td> <td colspan="2">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.</td> </tr> <tr> <td>Source sequence(s)</td> <td colspan="2">AW452130,AY008282,BC022532,DA122620</td> </tr> <tr> <td>Consensus CDS</td> <td colspan="2">CCDS13080.1</td> </tr> <tr> <td>Conserved Domains (1)</td> <td>summary</td> <td></td> </tr> <tr> <td></td> <td>smart00157</td> <td>PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in</td> </tr> </table>		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
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
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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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Canis lupus familiaris PrP 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

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All: 1 Fungi: 0 Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota

Alignment Scores

Species	Gene	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; Haplorhini; 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](#)

NC_000020_9

4615669 4630234

NM_002021.2 NP_002021.1 prnp protein CCDS1388v1
 NM_103972v1 NP_009902v1 prnp protein CCDS1388v1

■ - coding region ■ - untranslated region

Genomic context

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- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- CCDS
- Ensembl
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SNP linked to Gene [PRNP](#) (geneID:5621) Via Contig Annotation

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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_000311.3	plus strand	NP_000302.1	forward	NT_011387.8	reference	<- currently shown
NM_000311.3	plus strand	NP_000302.1	forward	NW_927317.1	Celera	View snp on GeneModel
NM_183079.2	plus strand	NP_898902.1	forward	NT_011387.8	reference	View snp on GeneModel
NM_183079.2	plus strand	NP_898902.1	forward	NW_927317.1	Celera	View snp on GeneModel

in gene region cSNP has frequency double hit refresh

gene model	Contig Label	Contig	mRNA	protein	mRNA orientation	transcript	snp count
(contig mRNA transcript):	reference	NT_011387.8	NM_000311.3	NP_000302.1	forward	plus strand	22, coding



Region	Contig position	mRNA pos	dbSNP rs# cluster id	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.				missense	A	Thr [T]	1	26
				N.D.				contig reference	C	Pro [P]	1	26
	4619949	183	rs11538762	N.D.				missense	A	His [H]	2	28

				N.D.			Yes	contig reference	C	Pro [P]	3	165
	4620378	612	rs16990018	0.039	H	Yes	Yes	missense	G	Ser [S]	2	171
				0.039	H	Yes	Yes	contig reference	A	Asn [N]	2	171
	4620399	633	rs11538766	N.D.	H	Yes	Yes	missense	T	Val [V]	2	178
				N.D.	H	Yes	Yes	contig reference	A	Asp [D]	2	178
	4620405	639	rs11538767	N.D.		Yes	Yes	missense	C	Ala [A]	2	180
				N.D.		Yes	Yes	contig reference	T	Val [V]	2	180
	4620464	698	rs28933385	N.D.		Yes	Yes	missense	A	Lys [K]	1	200
				N.D.		Yes	Yes	contig reference	G	Glu [E]	1	200
	4620521	755	rs1800014	N.D.		Yes	Yes	missense	A	Lys [K]	1	219
				N.D.		Yes	Yes	contig reference	G	Glu [E]	1	219
	4620538	772	rs6052773	N.D.	H	Yes	Yes	synonymous	T	Ala [A]	3	224
				N.D.	H	Yes	Yes	contig reference	C	Ala [A]	3	224
	4620545	779	rs17852079	N.D.		Yes	Yes	missense	A	Lys [K]	1	227
				N.D.		Yes	Yes	contig reference	C	Gln [Q]	1	227
	4620625	859	rs11538759	N.D.				synonymous	G	Gly [G]	3	253
				N.D.				contig reference	A	Gly [G]	3	253

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***176640** GeneTests, Link

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-Straussler 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.

[Kretschmar 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.

[Bader 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, RefSeq, GenBank, Protein, UniGene

LinkOut: HGVS, HGMD, GAD

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

***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

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***176640**
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.

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

NC_000020.9

4415169 4438234

5' 3'

NC_000020.9 NC_000020.9

■ - coding region ■ - untranslated region

Genomic context

chromosome: 20; Location: 20p13

4519639 4469324

RPL17A2 RPS4L2 PRNP PRND PRNT

See PRNP in MapViewer

Links

PROTEIN LINKS

- FASTA
- GENPEPT
- Blink
- Conserved Domains

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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SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
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	NP_898902	34335270	prion protein preproprotein [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	FAV10449	119630854	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fam
1435	31	FAV10450	119630855	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fam
1435	31	AA80162	46095329	prion protein [Homo sapiens]
1435	31	AA821693	11079226	prion protein [Homo sapiens]
1435	31	CAB75503	6996155	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1432	29	AAC50089	474359	prion protein
1432	29	F40252	730390	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen)
1432	1	AAV42953	61367107	prion protein [synthetic construct]
1432	1	AAV38282	54695820	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fam
1432	1	AAV37089	60834334	prion protein [synthetic construct]
1431	1	ABM92244	123980830	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1431	1	ABM95428	123995653	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa
1431	31	CA04698	49457097	PrP ^{Sc} [Homo sapiens]
1431	31	AAH12844	15277486	Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-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	474375	prion protein

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, 593964, 6996155, 20251002, 10053355, 11079226, 46095329, 21506484, 83353219, 111961006, 112019514, 130912, 190468, 53934492, 62774377, 15108368, 34335270, 122056623, 122056625, 122056628, 31674692, 33695389, 1828059, 3996152, 119630854, 119630855

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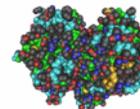
Hide identical Best hits Common Tree Taxonomy Report 3D structures CDD-Search G list Run BLAST

51 BLAST hits to 14 unique species Sort by taxonomy proximity

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

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

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
1203	*	1QL7A	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	*	1QH1A	6730489	Chain A, Human Prion Protein Fragment 90-230
771	*	1QH0A	6730488	Chain A, Human Prion Protein Fragment 90-230
768	*	1FO7A	10835618	Chain A, Human Prion Protein Mutant E200k Fragment 90-231
768	*	1FKCA	10835617	Chain A, Human Prion Protein (Mutant E200k) Fragment 90-231
698	*	1B10A	6729981	Chain A, Solution Nmr Structure Of Recombinant Syrian Hamster Prion Protein Rprp (90-231) , 25 Struct
678	*	2FJ3A	122920186	unnamed protein product [Oryctolagus cuniculus]
616	*	1TFXA	50513788	Chain A, Ovine Recombinant Prp(114-234), Arq Variant In Complex With The Fab Of The Vrq14 Antibody
604	*	1QH3A	6730491	Chain A, Human Prion Protein Fragment 121-230
604	*	1QH2A	6730490	Chain A, Human Prion Protein Fragment 121-230
587	*	1I4HA	20150069	Chain A, Crystal Structure Of The Human Prion Protein Reveals A Mechanism For Oligomerization
584	*	1DWZA	9955172	Chain A, Bovine Prion Protein Fragment 121-230
584	*	1DWYA	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	60594466	Chain A, Solution Structure Of The Sheep Prion Protein With Polymorphism H168
563	*	1HJNA	33356987	Chain A, Human Prion Protein At Ph 7.0
563	*	1HJMA	33356986	Chain A, Human Prion Protein At Ph 7.0
563	*	1XYWA	60594487	Chain A, Elk Prion Protein
560	*	1E1WA	9955179	Chain A, Human Prion Protein Variant R220k
560	*	1E1UA	9955178	Chain A, Human Prion Protein Variant R220k



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

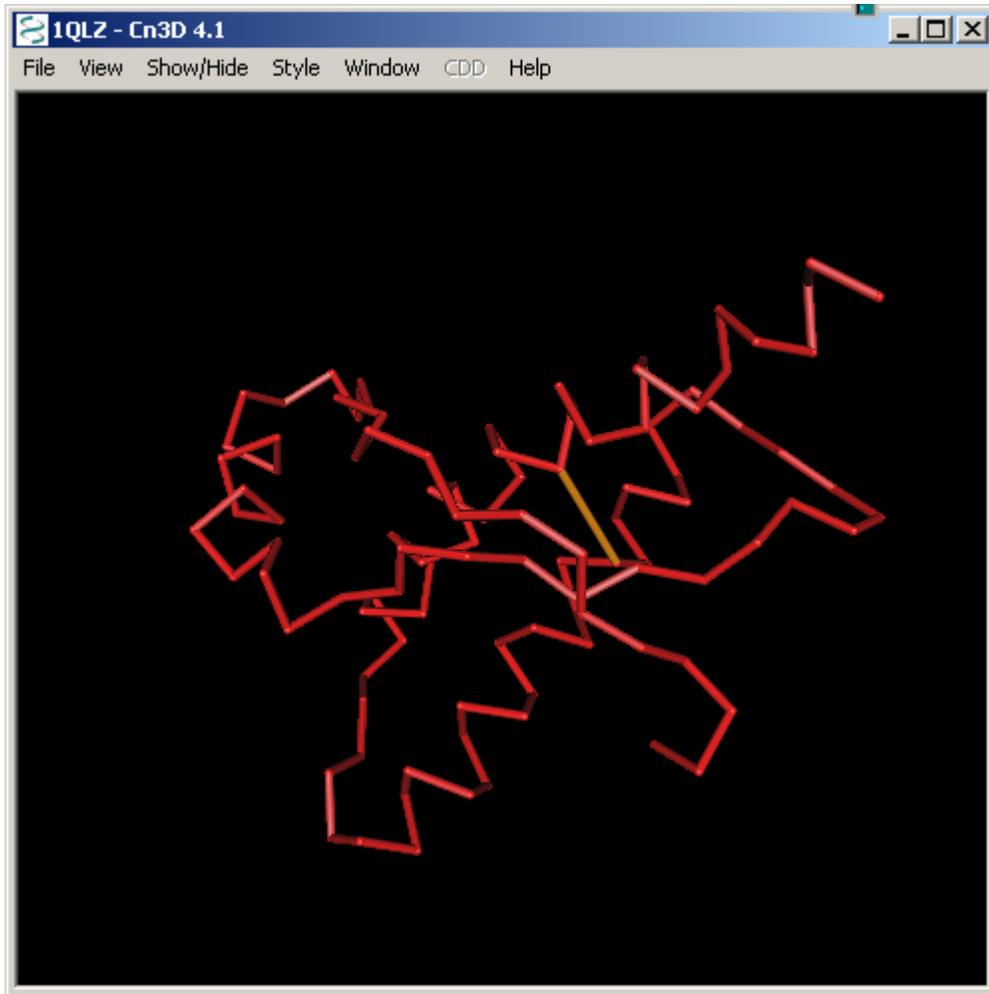
Structure: 1QLZ Chain A, Human Prion Protein

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

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

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

		10	20	30	40	50	60	70	80	
gi_4506113	23	KKRPKPGGWNTGGSRYPGQGS	PGGNRYPPQGGGGWGP	HGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP	102					
1QLZ_A	3	KKRPKPGGWNTGGSRYPGQGS	PGGNRYPPQGGGGWGP	HGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP	82					
		90	100	110	120	130	140	150	160	
gi_4506113	103	SKPKTNMKHMAGAAAAGAVVGG	LGGYMLGSAMSRPIIHFGSDYEDRYREN	MHRYPNQVYYRPMDEYSNQNNFVHDCVNI	182					
1QLZ_A	83	SKPKTNMKHMAGAAAAGAVVGG	LGGYMLGSAMSRPIIHFGSDYEDRYREN	MHRYPNQVYYRPMDEYSNQNNFVHDCVNI	162					
		170	180	190	200					
gi_4506113	183	TIKQHTVTTTTKGENFTETDV	KMMERVVEQMCITQYERESQAYYORGS	230						
1QLZ_A	163	TIKQHTVTTTTKGENFTETDV	KMMERVVEQMCITQYERESQAYYORGS	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?