

## 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 missense (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 icon under the "Clinically Associated" column in the SNP report. Compare the missense 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. To view the site of mutation in the 3D structure, superimpose the protein sequence on the 3D-structure of the human prion protein. Select the GENPEPT link for NP\_000302 under the section "Genomic Region, Transcripts and products". Then select "Related Structure" from the Links menu, click on the first arrow representing the related structure and then on the "Get 3D-structure data" button. Identify and highlight the residue corresponding to the mutation site the 3D structure.

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National Library of Medicine National Institutes of Health

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<b>PubMed Central:</b> free, full text journal articles	<b>OMIM:</b> online Mendelian Inheritance in Man
<b>Site Search:</b> NCBI web and FTP sites	<b>OMIA:</b> online Mendelian Inheritance in Animals

<b>CoreNucleotide:</b> Core subset of nucleotide sequence records	<b>dbGaP:</b> genotype and phenotype
<b>EST:</b> Expressed Sequence Tag records	<b>UniGene:</b> gene-oriented clusters of transcript sequences
<b>GSS:</b> Genome Survey Sequence records	<b>CDD:</b> conserved protein domain database
<b>Protein:</b> sequence database	<b>3D Domains:</b> domains from Entrez Structure
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<b>Taxonomy:</b> organisms in GenBank	<b>GEO Profiles:</b> expression and molecular abundance profiles
<b>SNP:</b> single nucleotide polymorphism	<b>GEO DataSets:</b> experimental sets of GEO data
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<b>Homologous Gene:</b> eukaryotic homology groups	<b>PubChem BioAssay:</b> bioactivity screens of chemical substances
<b>GENSAT:</b> gene expression atlas of mouse central nervous system	<b>PubChem Compound:</b> unique small molecule chemical structures
<b>Probe:</b> sequence-specific reagents	<b>PubChem Substance:</b> deposited chemical substance records
<b>Genome Project:</b> genome project information	<b>Protein Clusters:</b> a collection of related protein sequences

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Search Gene for prion protein Go Clear

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

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**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"[orgn] OR "Mus musculus"[orgn])</a>

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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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Search: Gene for prion protein AND human[organism] Save Search

Display: Summary Show 20 Send to

All: 53 Current Only: 41 Genes Genomes: 41 SNP GeneView: 40

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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  
 Chromosome: 17; Location: 17p13.1  
 Annotation: Chromosome 17, NC\_000017.9 (7512464..7531642, 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  
 Chromosome: 20; Location: 20p13  
 Annotation: Chromosome 20, NC\_000020.9 (4615069..4630234)  
 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  
 Chromosome: 9; Location: 9q33-q34.1  
 Annotation: Chromosome 9, NC\_000009.10 (127036953..127043430, complement)  
 MIM: 138120

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Search: Gene for

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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 28-Oct-2007

**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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Genomic regions, transcripts, and products

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Genomic context

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

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SNP GeneView: 1

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

updated 28-Oct-2007

provided by [HGNC](#)

30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial

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 region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial

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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 17-Mar-2008  
**RefSeq status:** Reviewed  
 total gene size: 15438 bp

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mRNA	bp	exons	Protein	aa	exons
<a href="#">NM_000311.3</a>	2740	2	<a href="#">NP_000302.1</a>	253	1
<a href="#">NM_001080121.1</a>	2735	2	<a href="#">NP_001073590.1</a>	253	1
<a href="#">NM_183079.2</a>	2736	2	<a href="#">NP_098902.1</a>	253	1
<a href="#">NM_001080122.1</a>	2731	2	<a href="#">NP_001073591.1</a>	253	1
<a href="#">NM_001080123.1</a>	2604	2	<a href="#">NP_001073592.1</a>	253	1

**Exon information:**  
[NM\\_000311.3](#) length: 2740 bp, number of exons: 2  
[NP\\_000302.1](#) length: 253 aa, number of exons: 1

EXON	coords	length	Coding EXON	coords	length	INTRON	coords	length
1	1 - 362	362 bp	363 - 13060	12698 bp				
	13061 - 15438	2378 bp	13071 - 13832	762 bp				

[NM\\_001080121.1](#) length: 2735 bp, number of exons: 2  
[NP\\_001073590.1](#) length: 253 aa, number of exons: 1

EXON	coords	length	Coding EXON	coords	length	INTRON	coords	length
1	1 - 362	362 bp	363 - 13065	12703 bp				
	13066 - 15438	2373 bp	13071 - 13832	762 bp				

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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-PrP 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; alternations in these interactions may have pathological consequences
6. This is the first publication of data that support the hypothesis that the common methionine/valine polymorphism at codon 129 of the PRNP gene may modify the susceptibility of women to mild temporal lobe epilepsy.
7. A novel three extra-repeat (72 bp) insertion within the octapeptide-coding region was identified in a Chinese family.
8. the PRNP polymorphism is more common in the Korean than in the Japanese population
9. plasmin cleaves PrP(c) in vitro and the liberated NH(2)-terminal fragment accelerates plasminogen activation

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>	APBB1		HPRD	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_055759.2</a>	CLSTN1		HPRD	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_001822.2</a>	CLU		HPRD	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_001834.2</a>	CNTN1		HPRD	<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>	CSNK2A1		BIND	<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>	CSNK2A2		BIND	<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).	NP_000302.1	<a href="#">NP_001311.3</a>	CSNK2B		BIND	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_004399.1</a>	DNM1		HPRD	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_570629.1</a>	DPP6		HPRD	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_002077.1</a>	GRB2		HPRD	<a href="#">PubMed</a>
PrPc interacts with HSPA5 (BIP).	NP_000302.1	<a href="#">NP_005338.1</a>	HSPA5		BIND	<a href="#">PubMed</a>
PrPc interacts with HSPD1 (Hsp60). This interaction was modeled on a demonstrated interaction between hamster PrPc and human HSPD1 (Hsp60).	NP_000302.1	<a href="#">NP_002147.2</a>	HSPD1		BIND	<a href="#">PubMed</a>
	NP_000302.1	<a href="#">NP_000416.1</a>	ITCAM		HPRD	<a href="#">PubMed</a>

**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
<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>	IPI <a href="#">PubMed</a>

Process	Evidence
<a href="#">cellular 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="#">membrane</a>	IEA
<a href="#">plasma membrane</a>	ISS

**General protein information** ↑ ?

General protein information	
<b>Names</b>	prion protein CD230 antigen prion protein PrP major prion protein prion-related protein
NCBI Reference Sequences (RefSeq)	
<b>RefSeqs maintained independently of Annotated Genomes</b> These reference sequences exist independently of genome builds. <a href="#">Explain</a>	
mRNA and Protein(s)	
1. <b>NM_000311.3--NP_000302.1 prion protein preproprotein</b>	
Description	Transcript Variant: This variant (1) represents the longest transcript. Variants 1-5 encode the same protein.
Source sequence(s)	<a href="#">AW452130,BC022532,DA297032,M13899</a>
Consensus CDS	<a href="#">CCDS13080.1</a>
Conserved Domains (1)	<a href="#">summary</a>
	<a href="#">smart00157</a> Location:23-230 Blast Score:546 PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.
2. <b>NM_001080121.1--NP_001073590.1 prion protein preproprotein</b>	
Description	Transcript Variant: This variant (3) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	<a href="#">AW452130,BC022532,BP251427,DA122620,M13899</a>
Conserved Domains (1)	<a href="#">summary</a>
	<a href="#">smart00157</a> Location:23-230 Blast Score:546 PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.
3. <b>NM_001080122.1--NP_001073591.1 prion protein preproprotein</b>	
Description	Transcript Variant: This variant (4) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	<a href="#">AW452130,BC022532,BI669189,DA297032,M13899</a>
Conserved Domains (1)	<a href="#">summary</a>
	<a href="#">smart00157</a> Location:23-230 Blast Score:546 PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.
4. <b>NM_001080123.1--NP_001073592.1 prion protein preproprotein</b>	
Description	Transcript Variant: This variant (5) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	<a href="#">BC022532,DB461478,M13899</a>
Conserved Domains (1)	<a href="#">summary</a>
	<a href="#">smart00157</a> Location:23-230 Blast Score:546 PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.
5. <b>NM_183079.2--NP_898902.1 prion protein preproprotein</b>	
Description	Transcript Variant: This variant (2) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	<a href="#">AW452130,AY008282,BC022532,DA122620</a>
Consensus CDS	<a href="#">CCDS13080.1</a>
Conserved Domains (1)	<a href="#">summary</a>
	<a href="#">smart00157</a> PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in

## Reference assembly

### Genomic

1. **NC\_000020.9 Reference assembly**  
Range 4615069..4630234  
Download [GenBank](#) [FASTA](#)
2. **NT\_011387.8**  
Range 4607069..4622234  
Download [GenBank](#) [FASTA](#)

## Alternate assembly (based on Celera assembly)

### Genomic

1. **AC\_000063.1 Alternate assembly (based on Celera assembly)**  
Range 4736784..4751948  
Download [GenBank](#) [FASTA](#)
2. **NW\_927317.1**  
Range 4593960..4609124  
Download [GenBank](#) [FASTA](#)

## Related Sequences



Nucleotide	Protein
Genomic <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="#">FAX10449.1</a>
	<a href="#">FAX10450.1</a>
Genomic <a href="#">DQ408531.1</a>	<a href="#">ABD63004.1</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>
Genomic <a href="#">S71210.1</a>	<a href="#">AAB20522.1</a>
Genomic <a href="#">S71212.1</a>	<a href="#">AAB20523.1</a>
Genomic <a href="#">S79978.1</a>	<a href="#">AAB35416.1</a>
Genomic <a href="#">S80539.1</a>	<a href="#">AAB21334.1</a>
Genomic <a href="#">S80732.1</a>	<a href="#">AAB50648.2</a>
Genomic <a href="#">S80743.1</a>	<a href="#">AAB50649.2</a>
Genomic <a href="#">S83341.1</a>	<a href="#">AAB50777.1</a>
Genomic <a href="#">U29185.1</a>	<a href="#">AAC78725.1</a>
Genomic <a href="#">X83416.1</a>	<a href="#">CAA58442.1</a>

NCBI Entrez Gene

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) [Homo sapiens]

GeneID: 5621 updated 28-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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1: H Alignment Scores

Multiple Alignment

ASN.1

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EST Links

GSS Links

Nucleotide Links

OMIA Links

OMIM Links

PMC Links

**Canis lupus familiaris** PRNP 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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Display: Alignment Scores Show: 0 Send to: [ ]

All: 1 Fungi: 0 Mammals: 0

Alignment Scores

Species	Gene	Symbol	Identity (%)		Substitution Rates <sup>1</sup>			Blast
			Protein	DNA	d	d <sub>N</sub> /d <sub>S</sub>	d <sub>NS</sub> /d <sub>NC</sub>	
<b>Homo sapiens</b>								
		<b>PRNP</b>						
vs. Pan troglodytes	CD230		99.2	99.2	0.008	0.138	0.548	Blast
vs. Canis lupus familiaris	PRNP		87.7	87.3	0.139	0.126	0.282	Blast
vs. Bos taurus	MGC140197		92.0	88.0	0.130	0.064	0.348	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
<b>Pan troglodytes</b>								
		<b>CD230</b>						
vs. Homo sapiens	PRNP		99.2	99.2	0.008	0.138	0.548	Blast
vs. Canis lupus familiaris	PRNP		87.7	87.3	0.139	0.126	0.253	Blast
vs. Bos taurus	MGC140197		92.0	87.9	0.132	0.063	0.300	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
<b>Canis lupus familiaris</b>								
		<b>PRNP</b>						
vs. Homo sapiens	PRNP		87.7	87.3	0.139	0.126	0.282	Blast
vs. Pan troglodytes	CD230		87.7	87.3	0.139	0.126	0.253	Blast
vs. Bos taurus	MGC140197		91.4	87.3	0.139	0.068	0.259	Blast
vs. Mus musculus	Pmp		86.9	82.5	0.200	0.092	0.335	Blast
vs. Rattus norvegicus	Pmp		86.9	83.6	0.185	0.099	0.300	Blast
vs. Gallus gallus	PRNP		48.9	55.6	0.673	0.338	0.908	Blast
<b>Bos taurus</b>								
		<b>MGC140197</b>						

NCBI Blast 2 Sequences results

PubMed Entrez BLAST OMIM Taxonomy Structure

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|CA118553.1| Pmp \[Mus musculus\]](#) >[gi|74182795|db|BAE34724.1| unnamed protein product \[Mus musculus\]](#) >[gi|74186646|db|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) [ Entrez Gene Home ]

Homo sapiens ]

GeneID: 5621 updated 17-Mar-2008

Summary

**Official Symbol** PRNP provided by HGNC

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

**Primary source** HGNC:9449

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

**Gene type** protein coding

**RefSeq status** Reviewed

**Organism** Homo sapiens

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

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

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

**Genomic regions, transcripts, and products**

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- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AcView
- CCDS
- Ensembl
- Evidence Viewer

NCBI Single Nucleotide Polymorphism

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search for SNP on NCBI Reference Assembly

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**BUILD 128**  
Have a question about dbSNP? Try searching the SNP FAQ Archive!

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_000311.3	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	<a href="#">View snp on GeneModel</a>
NM_183079.2	plus strand	NP_898902.1	forward	NT_011387.8	reference	<a href="#">View snp on GeneModel</a>
NM_183079.2	plus strand	NP_898902.1	forward	NW_927317.1	Celera	<a href="#">View snp on GeneModel</a>

show rare variation  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	

Color Legend

Region	Contig position	mRNA pos	dbSNP rs#	Heterozygosity	Validation	3D	Clinically Associated	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
								conhiz				

4620399	633	<a href="#">rs11538766</a>	N.D.	H	Yes	missense	T	Val [V]	2	178
			N.D.	H	Yes	conhg reference	A	Asp [D]	2	178
4620405	639	<a href="#">rs11538767</a>	N.D.		Yes	missense	C	Ala [A]	2	180
			N.D.		Yes	conhg reference	T	Val [V]	2	180
4620464	698	<a href="#">rs28933385</a>	N.D.		Yes	missense	A	Lys [K]	1	200
			N.D.		Yes	conhg reference	G	Glu [E]	1	200
4620521	755	<a href="#">rs1800014</a>	N.D.		Yes	missense	A	Lys [K]	1	219
			N.D.		Yes	conhg reference	G	Glu [E]	1	219
4620538	772	<a href="#">rs6052773</a>	N.D.	H	Yes	synonymous	T	Ala [A]	3	224
			N.D.	H	Yes	conhg reference	C	Ala [A]	3	224
4620545	779	<a href="#">rs17852079</a>	N.D.		Yes	missense	A	Lys [K]	1	227
			N.D.		Yes	conhg reference	C	Gln [Q]	1	227
4620625	859	<a href="#">rs11538759</a>	N.D.			synonymous	G	Gly [G]	3	253
			N.D.			conhg reference	A	Gly [G]	3	253





Online Mendelian Inheritance in Man



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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.

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

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Entrez Gene  
 M Nomenclature  
 R RefSeq  
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LinkOut  
 HGVS  
 HGMD  
 GAD

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

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

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

NCBI

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  
 Creation Date  
 Edit History

• Gene map

**OMIM**  
 Online Mendelian Inheritance in Man

Johns Hopkins University

All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy OMIM

Search OMIM for Go Clear

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Display Detailed Show 20 Send to

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

**Genomic regions, transcripts, and products**

Go to [reference sequence details](#) [Try our new Sequence Viewer](#)

Genomic context  
 chromosome: 20; Location: 20p13

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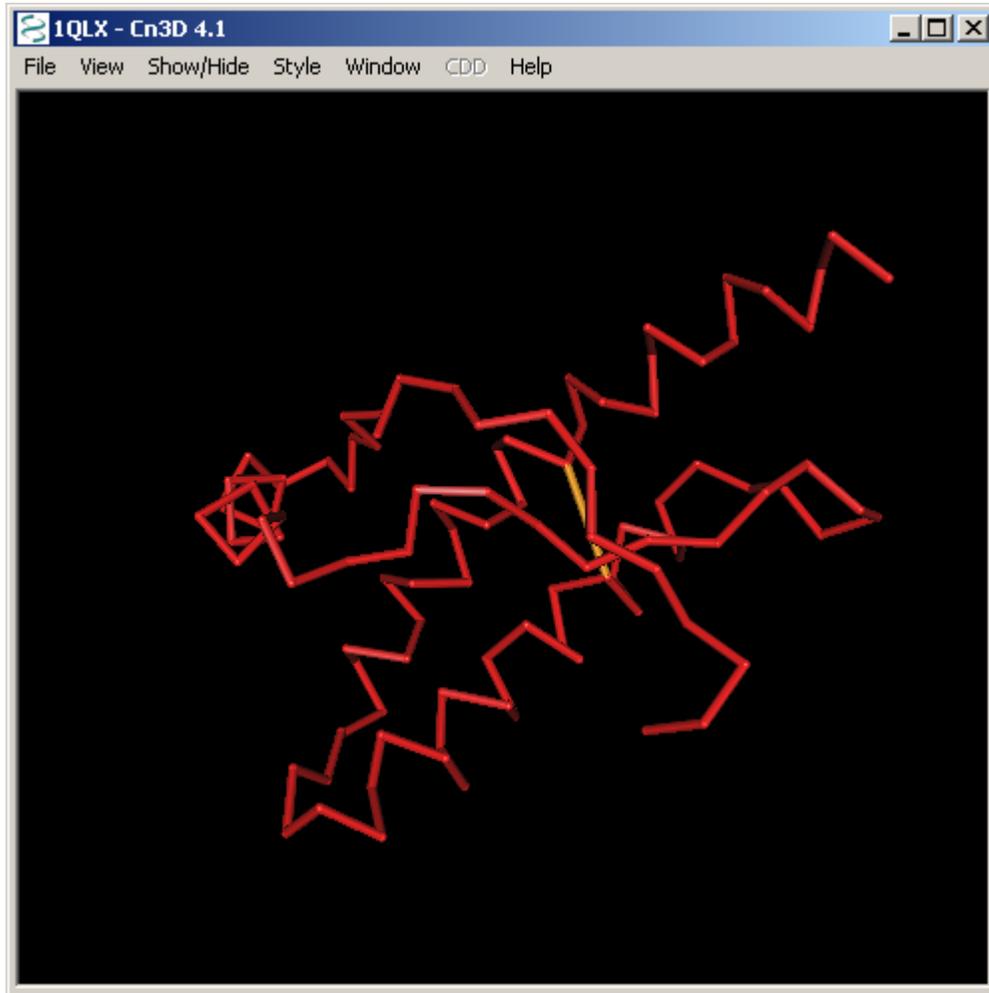
Range: from  to  Features:  SNP  CDD  HPRD

1: NP\_000302. Reports prion protein pre...[gi:4506113] BLink, Conserved Domains, Links

[Comment](#) [Features](#) [Sequence](#)

LOCUS NP\_000302 253 aa linear PRI 16-MAR-2008  
 DEFINITION prion protein preproprotein [Homo sapiens].  
 ACCESSION NP\_000302  
 VERSION NP\_000302.1 GI:4506113  
 DBSOURCE REFSEQ: accession [NM\\_000311.3](#)  
 KEYWORDS .  
 SOURCE Homo sapiens (human)  
 ORGANISM [Homo sapiens](#)  
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;  
 Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;  
 Catarrhini; Hominidae; Homo.  
 REFERENCE  
 1 (residues 1 to 253)  
 AUTHORS Poleggi, A., Bizzarro, A., Acciarri, A., Antonino, P., Bagnoli, S.,  
 Cellini, E., Forno, G.D., Giannattasio, C., Lauria, A., Matera, M.G.,  
 Nacmias, B., Puopolo, H., Seripa, D., Sorbi, S., Wekstein, D.R.,  
 Focchini, M. and Masullo, C.  
 TITLE Codon 129 polymorphism of prion protein gene in sporadic  
 Alzheimer's disease  
 JOURNAL Eur. J. Neurol. 15 (2), 173-178 (2008)  
 PUBMED 18217885  
 REMARK GeneRIF: This study does not support a role of PRNP polymorphism as  
 a susceptibility factor for Alzheimer's Disease.  
 REFERENCE  
 2 (residues 1 to 253)  
 AUTHORS Sakudo, A., Nakamura, I., Tsuji, S. and Ikuta, K.  
 TITLE GPI-anchorless human prion protein is secreted and glycosylated but  
 lacks superoxide dismutase activity  
 JOURNAL Int. J. Mol. Med. 21 (2), 217-222 (2008)  
 PUBMED 18204788  
 REMARK GeneRIF: The glycosylphosphatidylinositol-anchor site, but not  
 glycosylation, appears to be essential for the secretion of PrP.  
 REFERENCE  
 3 (residues 1 to 253)  
 AUTHORS Nadifi, S., Slassi, I., Hachimi, K.M., Gazzaz, B., Bellayou, H.,





IQLX - Sequence/Alignment Viewer  
View Edit Mouse Mode Unaligned Justification Imports

IQLX_A	DYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYY
gi 4506113	DYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYY

## 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 missense (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. To view the site of mutation in the 3D structure, superimpose the protein sequence on the 3D-structure of E.coli multL protein. Select the GENPEPT link for NP\_000240 under the section "Genomic Region, Transcripts and products". Then select "Related Structure" from the Links menu, click on the first arrow representing the related structure and then on the "Get 3D-structure data" button. 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?