

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.

Entrez Gene

Search Gene for prion protein Preview Go Clear current records only

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.

Entrez Gene

Search Gene for prion protein AND human[Organism] Preview Go Clear Save Search current records only

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.
- Click on query # to add to strategy

Search	Most Recent Queries	Time	Result
#14	Search prion protein AND human[Organism]	15:36:16	24

Entrez Gene

Search Gene for prion protein AND human[organism] Go Clear Save Search

Limits Preview/Index History Clipboard Details

Display Summary Show 20 Send to

All: 46 Current Only: 34 Genes Genomes: 34 SNP GeneView: 34

Items 1 - 20 of 46 Page 1 of 3 Next

1: [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, CID, GSS, MGCC26679, PRIP, PrP, PrP27-30, PrP33-35C, PrPc
Other Designations: CD230 antigen; major prion protein; prion protein; prion protein (p27-30) (Creutzfeld-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia); prion protein PrP; prion-related protein
Chromosome: 20; **Location:** 20p13
MIM: 176640
GeneID: 5621

2: [HSPD1](#) Order cDNA clone, Links
Official Symbol: HSPD1 **and Name:** heat shock 60kDa protein 1 (chaperonin) [*Homo sapiens*]
Other Aliases: CPN60, GROEL, HSP60, HSP65, HuCHA60, SPG13
Other Designations: P60 lymphocyte protein; chaperonin; heat shock 60kD protein 1 (chaperonin); heat shock protein 60; heat shock protein 65; mitochondrial matrix protein P1; short heat shock protein 60 Hsp60s1; spastic paraplegia 13 (autosomal dominant)
Chromosome: 2; **Location:** 2q33.1
MIM: 118190
GeneID: 3329

3: [HSPA5](#) Order cDNA clone, Links
Official Symbol: HSPA5 **and Name:** heat shock 70kDa protein 5 (glucose-regulated protein, 78kDa) [*Homo sapiens*]

NCBI Entrez Gene

Search Gene for [] Go Clear

Limits PreviewIndex History Clipboard Details

Display Full Report Show 5 Send to

All: 1 Current Only 1 Genes Genomes: 1 SNP GeneView 1

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

GeneID: 5621 updated 25-Sep-2006

Summary

Official Symbol PRNP

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

Primary source HGNC:9449

See related HPRD:01453; MIM:176640

Gene type protein coding

RefSeq status Reviewed

Organism [Homo sapiens](#)

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

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

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

Genomic regions, transcripts, and products

Entrez Gene Home

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Gene Table

NCBI Entrez Gene

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 25-Sep-2006
 RefSeq status: Reviewed
 total gene size: 15166 bp

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

mRNA bp exons Protein aa exons

NM_000311.2	2468	2	NP_000302.1	254	1
NM_183079.1	2464	2	NP_898902.1	254	1

Exon information:
 NM_000311.2 length: 2468 bp, number of exons: 2
 NP_000302.1 length: 254 aa, number of exons: 1

EXON	length	Coding EXON	length	INTRON	length
1 - 90	90 bp			91 - 12788	12698 bp

Links: Order cDNA clone, Books, Conserved Domains, Genome, GEO Profiles, HomoloGene, Map Viewer, Nucleotide, OMA, 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

NCBI Entrez Gene

Search Gene for [Go] [Clear]

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Display Gene Table Show 5 Send to

All: 1 Full Report Summary Brief ASN.1 XML Gene Table UI List LinkOut Books Links Conserved Domain Links RefSeq Genome Links total GENSAT Links

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Links: Order cDNA clone, Books, Conserved Domains, Genome

Genomic context

chromosome: 20; Location: 20p13

See PRNP in MapViewer

Bibliography

Related Articles in Pubmed

PubMed links

GeneRIFs: Gene References Into Function

What's a GeneRIF?

1. elevated plasma PrP(C) levels in renal disease were observed, showing that plasma PrP(C) is not a specific marker of neurological disease or Creutzfeldt-Jakob disease
2. the polymorphism at residue 129 does not change efficiency of conversion to beta-PrP conformation or affect binding of copper ions, but in a partially denatured conformation, it has a profound influence on ability of the protein to form amyloid fibrils
3. fetal and perinatal cellular prion protein (PrPC) expression in the forebrain; early expression of PrPC in the axonal field may suggest a specific role for this molecule in axonal growth during development
4. neuropathology and the characteristics of the PrPC associated with the H197D mutation in Gerstmann-Strausler-Scheinker syndrome

Links: UniSTS, AceView, CCDS, Evidence Viewer, GDB, GeneTests for MIM: 176640, HGMD, HGNC, HPRD, KEGG, MGC, ModelMaker, PharmGKB, UniGene, LinkOut

Entrez Gene Info

Feedback

Subscriptions

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

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

- 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](#)

Homology

Mouse, Rat
[Map Viewer](#)

GeneOntology Provided by [GOA](#)

Function	Evidence
GPI anchor binding	IEA
copper ion binding	TAS Pubmed
microtubule binding	IDA Pubmed

Process	Evidence
copper ion homeostasis	NAS Pubmed
metabolism	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
plasma membrane	ISS

General protein information ↑ ?

Names

- prion protein
- CD230 antigen
- prion protein PrP

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All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy Books OMIM

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*] Entrez Gene Home

GeneID: 5621 updated 25-Sep-2006 Table Of Contents

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See related	HPRD:01453 ; MIM:176640
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RefSeq status	Reviewed
Organism	Homo sapiens
Lineage	<i>Eukaryota</i> ; <i>Metazoa</i> ; <i>Chordata</i> ; <i>Craniata</i> ; <i>Vertebrata</i> ; <i>Euteleostomi</i> ; <i>Mammalia</i> ; <i>Eutheria</i> ; <i>Euarchontoglires</i> ; <i>Primates</i> ; <i>Haplorrhini</i> ; <i>Catarrhini</i> ; <i>Hominidae</i> ; <i>Homo</i>
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, Gerstmann-Strausler-Scheinker syndrome, and fatal familial insomnia.

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- OMIM
- Full Text via PubMed

NCBI HomoloGene Discover Homologs

Search HomoloGene for [] Go Clear

Limits **Preview/Index** History Clipboard Details

Display Alignment Scores Show 20 Send to []

All: 1 Fungi: 0 Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota Download, Links

H.sapiens	PRNP	prion protein (p27-30) (Creutzfeldt-Jakob ...
P.troglodytes	PRNP	prion protein (p27-30) (Creutzfeldt-Jakob ...
C.familiaris	PrP	prion protein (p27-30) (Creutzfeldt-Jakob ...
M.musculus	Prnp	prion protein
R.norvegicus	Prnp	prion protein
G.gallus	LOC396452	prion protein (p27-30) (Creutzfeldt-Jakob ...

NCBI HomoloGene Discover Homologs

Search HomoloGene for [] Go Clear

Limits Preview/Index **History** Clipboard Details

Display Alignment Scores Show 20 Send to []

All: 1 Fungi: 0 Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota Download, Link

Alignment Scores

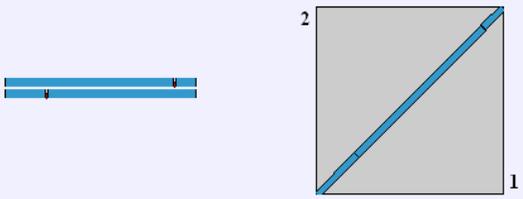
Species	Gene	aa%ID	nt%ID	D	Ka/Ks	Knr/Knc	
H.sapiens PRNP							
vs. P.troglodytes	PRNP	99.2	99.2	0.008	0.138	0.548	Blast
vs. C.familiaris	PrP	87.7	87.3	0.139	0.126	0.282	Blast
vs. M.musculus	Prnp	90.1	85.3	0.163	0.077	0.342	Blast
vs. R.norvegicus	Prnp	89.7	86.8	0.145	0.090	0.260	Blast
vs. G.gallus	LOC396452	47.1	57.4	0.631	0.399	0.878	Blast
P.troglodytes PRNP							
vs. H.sapiens	PRNP	99.2	99.2	0.008	0.138	0.548	Blast
vs. C.familiaris	PrP	87.7	87.3	0.139	0.126	0.253	Blast
vs. M.musculus	Prnp	90.1	85.2	0.165	0.075	0.307	Blast
vs. R.norvegicus	Prnp	89.7	86.7	0.146	0.088	0.229	Blast
vs. G.gallus	LOC396452	47.1	56.9	0.642	0.351	0.882	Blast
C.familiaris PrP							
vs. H.sapiens	PRNP	87.7	87.3	0.139	0.126	0.282	Blast
vs. P.troglodytes	PRNP	87.7	87.3	0.139	0.126	0.253	Blast
vs. M.musculus	Prnp	86.9	82.5	0.200	0.092	0.335	Blast

BLAST 2 SEQUENCES RESULTS VERSION BLASTP 2.2.14 [Apr-09-2006]

Matrix: gap open: gap extension:
 x_dropoff: expect: wordsize: [Filter](#) View option:
 Masking character option: Masking color option:
 Show CDS translation

Sequence 1: [gi4506113|ref|NP_000302.1|](#) prion protein preproprotein [Homo sapiens] >[gi34335270|ref|NP_898902.1|](#) prion protein preproprotein [Homo sapiens] >[gi46095329|gb|AAS80162.1|](#) prion protein [Homo sapiens] >[gi6996155|emb|CAB75503.1|](#) PRNP [Homo sapiens] >[gi11079226|gb|AAG21693.1|](#) prion protein [Homo sapiens] >[gi130912|sp|P04156|PRIO_HUMAN](#) Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR) (CD230 antigen) >[gi190468|gb|AAA60182.1|](#) prion protein
 Length = 253 (1 .. 253)

Sequence 2: [gi13173473|ref|NP_035300.1|](#) prion protein [Mus musculus] >[gi2865215|gb|AAC02804.1|](#) short incubation prion protein Prmpa [Mus musculus] >[gi71060019|emb|CAJ18553.1|](#) Prnp [Mus musculus] >[gi74192797|dbj|BAE34911.1|](#) unnamed protein product [Mus musculus] >[gi74194091|dbj|BAE36950.1|](#) unnamed protein product [Mus musculus] >[gi74213636|dbj|BAE35622.1|](#) unnamed protein product [Mus musculus] >[gi74186646|dbj|BAE34788.1|](#) unnamed protein product [Mus musculus] >[gi74182795|dbj|BAE34724.1|](#) unnamed protein product [Mus musculus] >[gi13879449|gb|AAH06703.1|](#) Prion protein [Mus musculus] >[gi130914|sp|P04925|PRIO_MOUSE](#) Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen) >[gi200529|gb|AAA39997.1|](#) prion protein
 Length = 254 (1 .. 254)



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



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- Protein
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- PubMed (GeneRIF)
- SNP
 - SNP: Genotype
 - SNP: GeneView

Genomic regions, transcripts, and products

Go to [reference sequence details](#)



NCBI Single Nucleotide Polymorphism

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

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Have a question about dbSNP? Try searching the SNP FAQ Archive

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dbSNP Handbook
Overview
RefSNP Summary Info
Database Schema
Database Dictionary **NEW**
Database Changes **NEW**
Genotype Schema
Data Formats
Heterozygosity
Computation

SEARCH

SNP linked to Gene (geneID:5621)

SNP are linked from gene [PRNP](#) via the following methods:
[Contig Annotation](#) [GenBank\(mrna\) Mapping](#)

Send all rs# to Batch Query Download all rs# to file: [GENE GENOTYPE REPORT](#)

Gene Model (mRNA alignment) information from genome sequence ↑

Total gene model (contig mRNA transcript): 4

mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	snp list
NM_000311	plus strand	NP_000302	forward	NT_011387	reference	currently shown
NM_000311	plus strand	NP_000302	forward	NW_927317	Celera	view
NM_183079	plus strand	NP_898902	forward	NT_011387	reference	view
NM_183079	plus strand	NP_898902	forward	NW_927317	Celera	view

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 21, coding	

Region	Contig position	mRNA pos	dbSNP rs#	Heterozygosity	Validation	3D	OMIM	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos
exon_2	4619942	176	rs11538755	N.D.				nonsynonymous	A	Thr [T]	1	26
				N.D.				contig reference	C	Pro [P]	1	26

				N.D.				contig reference	A	Asp [D]	2	178
	4620405		rs11538767	N.D.				nonsynonymous	C	Ala [A]	2	180
				N.D.				contig reference	T	Val [V]	2	180
	4620464		rs28933385	N.D.				nonsynonymous	A	Lys [K]	1	200
				N.D.				contig reference	G	Glu [E]	1	200
	4620521		rs1800014	N.D.				nonsynonymous	A	Lys [K]	1	219
				N.D.				contig reference	G	Glu [E]	1	219
	4620538		rs6052773	N.D.				synonymous	T	Ala [A]	3	224
				N.D.				contig reference	C	Ala [A]	3	224
	4620545		rs17852079	N.D.				nonsynonymous	A	Lys [K]	1	227
				N.D.				contig reference	C	Gln [Q]	1	227
	4620625		rs11538759	N.D.				synonymous	G	Gly [G]	3	253
				N.D.				contig reference	A	Gly [G]	3	253

NCBI

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

FATAL FAMILIAL INSOMNIA, INCLUDED

MIM *176640

Description
Cloning
Gene Structure
Mapping
Gene Function
Molecular Genetics
Genotype/Phenotype
Correlations
Population Genetics
Animal Model
History
Allelic Variants
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See Also
References
Contributors

In 2 patients with Creutzfeldt-Jakob disease (123400) from the same family, Goldgaber et al. (1989) identified a G-to-A transition in the PRNP gene, resulting in a glu200-to-lys (E200K) substitution.

Studying an unusual cluster of cases of CJD in rural Slovakia, Goldfarb et al. (1990) found the E200K mutation in all 11 tested cases of 'focal CJD,' in 12 of 40 healthy first-degree relatives, and in 6 of 23 other relatives. By contrast, no extrafocal cases or their relatives had the mutation; nor did any unrelated individuals within or outside the cluster regions. One of the healthy individuals with the E200K mutation was the 75-year-old mother of one of the patients. The unusually high incidence of CJD in the Orava and Lucenec regions of Slovakia appeared to be of recent origin. Goldfarb et al. (1990) interpreted this as indicating that the mutation is a necessary, but not sufficient, factor in the disease. Another factor such as scrapie-infected sheep was proposed.

Mitrova et al. (1990) described the familial occurrence of 3 definite and 2 possible cases of CJD with temporal and spatial separation in the area of focal CJD accumulation in Slovakia. The incubation period appeared to be about 51 years, judging by the interval between the death of the affected mother and the clinical onset in the first affected child. Affected offspring tended to die at the same time, not at the same age. Due to separation of the affected children, a possible common exposure to CJD infection was limited to approximately 7 years during their childhood.

Molecular Genetics
Genotype/Phenotype
Correlations
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Gene map

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

LinkOut
H HGVs
G HGMD

All: 1 OMIM dbSNP: 1 OMIM UniSTS: 0

***176640** GeneTests, Links

PRION PROTEIN; PRNP

ALLELIC VARIANTS
(selected examples)

- 0001 CREUTZFELDT-JAKOB DISEASE [PRNP, EXTRA OCTAPEPTIDE CODING REPEATS]
- 0002 GERSTMANN-STRAUSSLER DISEASE [PRNP, PRO102LEU]
- 0003 REMOVED FROM DATABASE
- 0004 GERSTMANN-STRAUSSLER DISEASE [PRNP, ALA117VAL]
- 0005 PRION DISEASE, SUSCEPTIBILITY TO [PRNP, MET129VAL] dbSNP
- 0006 CREUTZFELDT-JAKOB DISEASE [PRNP, GLU200LYS] dbSNP
- 0007 CREUTZFELDT-JAKOB DISEASE [PRNP, ASP178ASN AND MET129VAL]
- 0008 REMOVED FROM DATABASE
- 0009 REMOVED FROM DATABASE
- 0010 FATAL FAMILIAL INSOMNIA [PRNP, ASP178ASN AND MET129]
- 0011 GERSTMANN-STRAUSSLER DISEASE [PRNP, PHE198SER]
- 0012 GERSTMANN-STRAUSSLER DISEASE [PRNP, GLN217ARG]
- 0013 REMOVED FROM DATABASE
- 0014 CREUTZFELDT-JAKOB DISEASE [PRNP, VAL210ILE]
- 0015 GERSTMANN-STRAUSSLER DISEASE [PRNP, PRO105LEU]
- 0016 CREUTZFELDT-JAKOB DISEASE [PRNP, VAL180ILE]
- 0017 CREUTZFELDT-JAKOB DISEASE [PRNP, MET232ARG]
- 0018 SPONGIFORM ENCEPHALOPATHY WITH NEUROPSYCHIATRIC FEATURES [PRNP, ASN171SER] dbSNP
- 0019 CREUTZFELDT-JAKOB DISEASE, PROTECTION AGAINST [PRNP, GLU219LYS]
- 0020 REMOVED FROM DATABASE

NCBI 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

repeats. This gene is found on chromosome 20, approximately 20 kb 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

4415049 5' 4459234 3'

NP_418314.2 NP_418315.1

■ - coding region ■ - untranslated region

Genomic context

chromosome: 20; Location: 20p13

RPL7L2 RPL4L2 PRNP PRND PRNT

See PRNP in MapViewer

Bibliography

Related Articles in Pubmed

PubMed links

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

1. elevated plasma PrP(C) levels in renal disease were observed, showing that plasma PrP (C) is not a specific marker of neurological disease or Creutzfeldt-Jakob disease
2. the polymorphism at residue 129 does not change efficiency of conversion to beta-PrP conformation or affect binding of copper ions, but in a partially denatured

Nucleotide
OMIA
OMIM
Full text in PMC
Probe
Protein
PubMed
PubMed (GeneRIF)
SNP
SNP: Genotype
SNP: GeneView
Taxonomy
UniSTS
AceView
CCDS
Evidence Viewer
GDB
GeneTests for MIM: 176640
HGMD
HGNC
HPRD
KEGG
MGC
ModelMaker
PharmGKB
UniGene
LinkOut

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► Feedback
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NCBI

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

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

Hide identical Best hits Common Tree Taxonomy Report 3D structures CDD-Search Glist RunBLAST

200 BLAST hits to 48 unique species Sort by taxonomy proximity

Archaea Bacteria 194 Metazoa Fungi Plants Viruses Other Eukaryotae

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

253 aa

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
Conserved Domain Database hits				
1435	31	AAS80162	46095329	prion protein [Homo sapiens]
1435	31	NP_898902	34335270	prion protein preproprotein [Homo sapiens]
1435	31	AAG21693	11079226	prion protein [Homo sapiens]
1435	31	CAB75503	6996155	PRNP [Homo sapiens]
1435	31	AAA60182	190468	prion protein
1435	31	P04156	130912	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR)
1432	29	P40252	730390	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230)
1432	29	AAC50089	474359	prion protein
1432	1	AAV37089	60834334	prion protein [synthetic construct]
1432	1	AAV42953	61367107	prion protein [synthetic construct]
1432	1	AAV38282	54695820	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strau
1431	31	CAG46869	49457097	PRNP [Homo sapiens]
1431	31	AAH12844	15277486	Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Stra
1430	31	ABD63004	89160954	prion protein PrP [Homo sapiens]
1427	31	AAH22532	18490397	Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Stra
1427	29	NP_001...	57114055	prion protein [Pan troglodytes]
1427	29	AAV74306	56122310	prion protein [Pan troglodytes]
1427	29	P61768	48429057	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230)
1427	27	P61767	48429056	Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230)

NCBI

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

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

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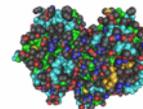
43 BLAST hits to 12 unique species Sort by taxonomy proximity

Archaea Bacteria 43 Metazoa Fungi Plants Viruses Other Eukaryotae

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

253 aa

SCORE	E	ACCESSION	GI	PROTEIN DESCRIPTION
Conserved Domain Database hits				
1203	•	IQLZA	6730487	Chain A, Human Prion Protein
1203	•	IQLXA	6730485	Chain A, Human Prion Protein
1136	•	IDX1A	9955174	Chain A, Bovine Prion Protein Residues 23-230
1136	•	IDX0A	9955173	Chain A, Bovine Prion Protein Residues 23-230
771	•	IQM1A	6730489	Chain A, Human Prion Protein Fragment 90-230
771	•	IQMOA	6730488	Chain A, Human Prion Protein Fragment 90-230
768	•	IFO7A	10835618	Chain A, Human Prion Protein Mutant E200k Fragment 90-231
768	•	IFKCA	10835617	Chain A, Human Prion Protein (Mutant E200k) Fragment 90-231
698	•	IB10A	6729981	Chain A, Solution Nmr Structure Of Recombinant Syrian Hamster Prion
604	•	IQM3A	6730491	Chain A, Human Prion Protein Fragment 121-230
604	•	IQM2A	6730490	Chain A, Human Prion Protein Fragment 121-230
587	•	II4MA	20150089	Chain A, Crystal Structure Of The Human Prion Protein Reveals A M
584	•	IDWZA	9955172	Chain A, Bovine Prion Protein Fragment 121-230
584	•	IDWYA	9955171	Chain A, Bovine Prion Protein Fragment 121-230
583	•	IH0LA	28373307	Chain A, Human Prion Protein 121-230 M166cE221C
576	•	IY2SA	60594516	Chain A, Ovine Prion Protein Variant R168
569	•	IXYUA	60594486	Chain A, Solution Structure Of The Sheep Prion Protein With Polym



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

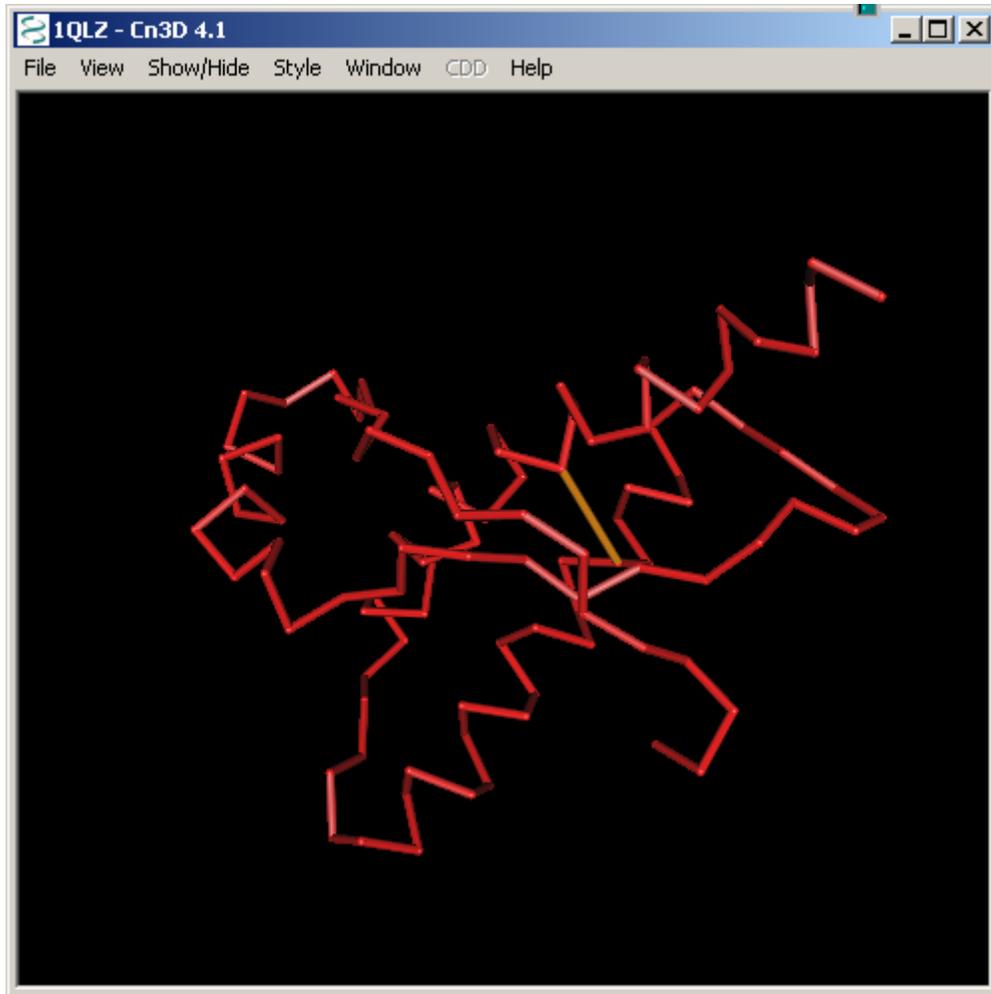
Structure: 1QLZ Chain A, Human Prion Protein

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	PGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP							102
1QLZ_A	3	KKRPKPGGWNTGGSRYPGQGS	PGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP							82
		90	100	110	120	130	140	150	160	
	*********
gi_4506113	103	SKPKTNMKHMAGAAAAGAVVGG	LGGYHLGSAMSRPIIHFGSDYEDRYRENMMHRYPNQVYYRPMDEYSNQNNFVHDCVNI							182
1QLZ_A	83	SKPKTNMKHMAGAAAAGAVVGG	LGGYHLGSAMSRPIIHFGSDYEDRYRENMMHRYPNQVYYRPMDEYSNQNNFVHDCVNI							162
		170	180	190	200					
	*********
gi_4506113	183	TIKQHTVTTTTKGENFTETDV	KMMERVVEQMCITQYERESQAYYQRGS			230				
1QLZ_A	163	TIKQHTVTTTTKGENFTETDV	KMMERVVEQMCITQYERESQAYYQRGS			210				



1QLX - Sequence/Alignment Viewer

View Edit Mouse Mode Unaligned Justification Imports

1QLX A	LGGYMLGSAMSRPIIHFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNIITIKQHTVTTTTKGENFTETDVKMME
gi 4506113	LGGYMLGSAMSRPIIHFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNIITIKQHTVTTTTKGENFTETDVKMME

Problem 2

Retrieve human entries related to "colon cancer" in [Entrez Gene](#). Identify the gene MLH1. Name the map location of this gene on the human genome. What is the function of this protein? What are the alternate gene symbols? Name the phenotypes associated with the mutations in this gene.

Is the RefSeq mRNA record reviewed? How many alternatively spliced products have been annotated for the gene?

To obtain information about the homologs from other eukaryotes, click on the Homologene link. Change the Display option to "Alignment Scores". How great is the percent identity between the human and mouse proteins? View the alignment by clicking on the "Blast" link.

Go back to the Entrez Gene report. Identify the variations annotated on this gene by clicking on the geneView in dbSNP link. How many of them are nonsynonymous changes? To determine whether known SNPs in the coding region of a gene are associated with any phenotype, access the OMIM record by clicking on the "Yes" link under the OMIM column in the SNP report. Compare the nonsynonymous changes from the SNP report with the "ALLELIC VARIANTS" in the OMIM record. Are there any SNPs known to cause a change in the function of the MLH1 protein?

Go back to the Entrez gene report. View the list of similar proteins through the "BL" link in the next to the protein NP_000240. To view the sites of mutations in the 3D structure, superimpose the protein sequence on the 3D-structure of E.coli multL protein 1BKNB (use BL--3D-structure button--click on the second blue dot--Get 3D Structure Data). Identify and highlight the amino acid corresponding to the human MLH1 isoleucine 32 on the 3D structure. What is the amino acid at this position in the E.coli protein? Based on this information, do you think the I32V mutation in the human protein will alter its function? Confirm your findings through the OMIM record for MLH1.