Mini Courses NCBI Resources one course at a time

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.

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for prion protein Preview Go Clear

Limits | Preview/Index | History | Clipboard | Details

- Enter terms and click Preview to see only the number of search results.
- To combine searches use # before search number, e.g., (#2 OR #3) AND asthma.

No history available

Add Term(s) to Query or View Index:

- Enter a term in the text box, use the pull-down menu to specify a search field.
- Click Preview to add terms to the query box and see the number of search results, or click Index to view terms within a field.

Organism human Preview Index

Click AND OR NOT to add a term to the query box.

Entrez Gene: Home, About, FAQ, Help, Gene Handbook, Statistics, Downloads (FTP)

Mailing Lists: Gene, RefSeq

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for prion protein Preview Go Clear

Limits | Preview/Index | History | Clipboard | Details

- Enter terms and click Preview to see only the number of search results.
- To combine searches use # before search number, e.g., (#2 OR #3) AND asthma.
- To save search indefinitely, click query # and select Save in My NCBI.
- To combine searches use #search, e.g., #2 AND #3 or click query # for more options.

Search	Most Recent Queries	Time	Result
#25	Search **prion protein**	13:21:17	273

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.

All Fields Preview Index

Click AND OR NOT to add a term to the query box.

Entrez Gene: Home, About, FAQ, Help, Gene Handbook, Statistics, Downloads (FTP)

Mailing Lists: Gene, RefSeq

Feedback

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for prion protein Go Clear Save Search

Limits | Preview/Index | History | Clipboard | Details

Display Summary Show 20 Send to

All: 273 | Current Only: 239 | Genes Genomes: 220 | SNP GeneView: 78

Items 1 - 20 of 273 Page 1 of 14 Next

1: tat Links
Tat [*Human immunodeficiency virus 1*]
Other Aliases: HIV1gp5
GeneID: 155871

2: PRNP Order cDNA clone, Links
Official Symbol: PRNP **and Name:** prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [*Homo sapiens*]
Other Aliases: ASCR, CD230, CJD, GSS, MGC26679, PRIP, PrP, PrP27-30, PrP33-35C, PrPc
Other Designations: CD230 antigen; major prion protein; prion protein; prion protein PrP; prion-related protein
Chromosome: 20; **Location:** 20p13
MIM: 176640
GeneID: 5621

3: PRNP Order cDNA clone, Links
prion protein [*Bos taurus*]
Other Aliases: MGC140197, PrP
Other Designations: polymorphic; prion protein PrP; prion protein precursor PrP
Chromosome: 13; **Location:** 13q17
GeneID: 281427

Entrez Gene: Home, About, FAQ, Help, Gene Handbook, Statistics, Downloads (FTP)

Mailing Lists: Gene, RefSeq

Feedback: Help Desk, Corrections, About GeneRIFs

Related Sites: BLAST, Entrez Genome, Genome Projects, Genomic Biology, GEO

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for Go Clear

Limits | Preview/Index | History | Clipboard | Details

Display Full Report Show 5 Send to

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

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

GeneID: 5621 updated 05-Feb-2007

Summary

Official Symbol	PRNP provided by HGNC
Official Full Name	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) provided by HGNC
Primary source	HGNC:9449
See related	HPRD:01453; MIM:176640
Gene type	protein coding
RefSeq status	Reviewed
Organism	*Homo sapiens*
Lineage	*Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo*
Also known as	CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C
Summary	The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Straussler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.

Entrez Gene Home

Table Of Contents
- Summary
- Genomic regions, transcripts...
- Genomic context
- Bibliography
- HIV-1 protein interactions
- Interactions
- General gene information
- General protein information
- Reference Sequences
- Related Sequences
- Additional Links

Links Explain
- Order cDNA clone
- Books
- Conserved Domains
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- OMIA
- OMIM
- Full text in PMC
- Probe
- Protein
- PubMed

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for Go Clear

Limits | Preview/Index | History | Clipboard | Details

Display Gene Table Show 5 Send to

Full Report
Summary
Brief
ASN.1
XML
Gene Table
UI List
LinkOut
Books Links
Conserved Domain Links
Genome Links
GENSAT Links
GEO Profile Links
HomoloGene Links
Nucleotide Links
NIH cDNA clone links
OMIA Links
OMIM Links
BioAssay Links
PMC Links
Probe Links
Protein Links
PubMed Links
PubMed (GeneRIF) Links
SNP Links
Gene Genotype Links
Taxonomy Links
UniGene Links
UniSTS Links

Entrez Gene Home

Table Of Contents
- Summary
- Genomic regions, transcripts...
- Genomic context
- Bibliography
- HIV-1 protein interactions
- Interactions
- General gene information
- General protein information
- Reference Sequences
- Related Sequences
- Additional Links

Links Explain
- Order cDNA clone
- Books
- Conserved Domains
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- OMIA
- OMIM
- Full text in PMC
- Probe

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for Go Clear

Limits | Preview/Index | History | Clipboard | Details

Display Gene Table Show 5 Send to

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

1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
[*Homo sapiens*]
GeneID: 5621 updated 05-Feb-2007
RefSeq status: Reviewed
total gene size: 15166 bp

Genomic regions, transcripts, and products

Go to reference sequence details

NC_000020.9

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: 253 aa, number of exons: 1

EXON coords	length	Coding EXON coords	length	INTRON coords	length
1 - 90	90 bp			91 - 12788	12698 bp
12789 - 15166	2378 bp	12799 - 13560	762 bp		

NM_183079.1 length: 2464 bp, number of exons: 2
NP_898902.1 length: 253 aa, number of exons: 1

EXON coords	length	Coding EXON coords	length	INTRON coords	length
1 - 86	86 bp			87 - 12788	12702 bp
12789 - 15166	2378 bp	12799 - 13560	762 bp		

Entrez Gene Home

Explain

Links
Order cDNA clone
Books
Conserved Domains
Genome
GEO Profiles
HomoloGene
Map Viewer
Nucleotide
OMIA
OMIM
Full text in PMC
Probe
Protein
PubMed
PubMed (GeneRIF)
SNP
SNP: Genotype
SNP: GeneView
Taxonomy
UniSTS
AceView
CCDS
Evidence Viewer
GDB
GeneTests for MIM: 176640
HGMD
HGNC
HPRD
KEGG
MGC
ModelMaker
PharmGKB
UniGene
LinkOut

Entrez Gene Info

Feedback

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for Go Clear

Limits | Preview/Index | History | Clipboard | Details

Display Gene Table Show 5 Send to

Full Report
Summary
Brief
ASN.1
XML
Gene Table
UI List
LinkOut
Books Links
Conserved Domain Links
Genome Links

All: 1 | Genes Genomes: 1 | SNP GeneView: 1

7-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)

updated 05-Feb-2007

Entrez Gene Home

Explain

Links
Order cDNA clone
Books
Conserved Domains
Genome

Genomic context

chromosome: 20; Location: 20p13

See PRNP in MapViewer

HGMD
HGNC
HPRD
KEGG
MGC
ModelMaker
PharmGKB
UniGene
LinkOut

▼ Entrez Gene Info

▶ Feedback

▼ Subscriptions

Bibliography

Related Articles in PubMed

PubMed links

GeneRIFs: Gene References Into Function

What's a GeneRIF?

1. results suggest that the PRNP genetic variants are not associated with the risk for Alzheimer's disease in Korean population
2. first study that provides experimental evidence supporting the hypothesis that there might be silent prions lying dormant in normal human brains
3. 1-OPRD (one octapeptide-repeat deletion) homozygosity or heterozygosity exists in several gastric cancer cell lines, e.g. MKN28 and KatoIII are homozygous for 1-OPRD, and SGC7901 and BGC-823 are heterozygous for 1-OPRD.
4. Polymorphic microsatellite sites within 148 kb of the human prion gene complex, including the genes PRNP, PRND and PRNT, were analysed together with the Codon129 variants regarding 50 Creutzfeldt-Jakob Disease patients and 46 non-diseased control persons.
5. The highest affinity copper (II)-binding modes cause self-association of both peptides, suggesting a role for copper (II) in controlling prion protein self-association in vivo
6. there is a link between ER stress and the formation of cytosolic PrP isoforms potentially endowed with novel signaling or cytotoxic functions
7. prion protein may act as an inhibitor of microtubule assembly by inducing formation of stable tubulin oligomers
8. At pH 5.5 the prion protein binds 2 Cu2+ ions. All 6 histidines of the unfolded N-terminal domain & the N-terminal amine

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	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).					

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

Entrez Gene: PRNP prion protein (p27-30) (Creutzfeld...

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	IEA	

Process	Evidence	
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	
plasma membrane	ISS	

General protein information

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

NCBI Entrez Gene

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | PMC | Taxonomy | Books | OMIM

Search Gene for Go Clear

Limits | Preview/Index | History | Clipboard | Details

Display Full Report Show 5 Send to

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

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

GeneID: 5621 updated 05-Feb-2007

Summary

Official Symbol	PRNP provided by HGNC
Official Full Name	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) provided by HGNC
Primary source	HGNC:9449
See related	HPRD:01453; MIM:176640
Gene type	protein coding
RefSeq status	Reviewed
Organism	*Homo sapiens*
Lineage	*Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo*
Also known as	CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C
Summary	The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Straussler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.

Entrez Gene Home

Table Of Contents
- Summary
- Genomic regions, transcripts...
- Genomic context
- Bibliography
- HIV-1 protein interactions
- Interactions
- General gene information
- General protein information
- Reference Sequences
- Related Sequences
- Additional Links

Links Explain
- Order cDNA clone
- Books
- Conserved Domains
- Genome
- GEO Profiles
- HomoloGene
- Map Viewer
- Nucleotide
- OMIA
- OMIM
- Full text in PMC
- Probe
- Protein
- PubMed

NCBI HomoloGene *Discover Homologs* Help

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | Map Viewer | Gene | UniGene | OMIM

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 ...

About Entrez

HomoloGene
- Home
- Query Tips
- Build Procedure
- FTP Site

Genome Resources
- Homo sapiens
- Mus musculus
- Rattus norvegicus
- Danio rerio

NCBI HomoloGene Discover Homologs

Help

My NCBI [Sign In] [Register]

All Databases | PubMed | Nucleotide | Protein | Genome | Structure | Map Viewer | Gene | UniGene | OMIM

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

NCBI **Blast 2 Sequences results**

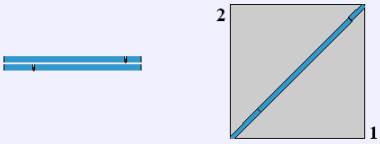
PubMed | Entrez | BLAST | OMIM | Taxonomy | Structure

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

Matrix BLOSUM62 gap open: 11 gap extension: 1
x_dropoff: 50 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|ref|NP_898902.1|prion protein preproprotein [Homo sapiens] >gi|4506113|ref|NP_000302.1| prion protein preproprotein [Homo sapiens] >gi|130912|sp|P04156|PRIO_HUMAN Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR) (CD230 antigen) >gi|190468|gb|AAA60182.1| prion protein >gi|6996155|emb|CAB75503.1| PRNP [Homo sapiens] >gi|11079226|gb|AAG21693.1| prion protein [Homo sapiens] >gi|46095329|gb|AAS80162.1| prion protein [Homo sapiens]
Length = 253 (1 .. 253)

Sequence 2: gi|13173473|ref|NP_035300.1|prion protein [Mus musculus] >gi|130914|sp|P04925|PRIO_MOUSE Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen) >gi|200529|gb|AAA39997.1| prion protein >gi|2865215|gb|AAC02804.1| short incubation prion protein Prmpa [Mus musculus] >gi|13879449|gb|AAH06703.1| Prion protein [Mus musculus] >gi|71060019|emb|CAJ18553.1| Prnp [Mus musculus] >gi|74182795|dbj|BAE34724.1| unnamed protein product [Mus musculus] >gi|74186646|dbj|BAE34788.1| unnamed protein product [Mus musculus] >gi|74192797...
Length = 254 (1 .. 254)



Limits | Preview/Index | History | Clipboard | Details

Display Full Report | Show 5 | Send to

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

1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
[*Homo sapiens*]
GeneID: 5621 updated 05-Feb-2007

Summary

Official Symbol	PRNP (provided by HGNC)
Official Full Name	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) (provided by HGNC)
Primary source	HGNC:9449
See related	HPRD:01453; MIM:176640
Gene type	protein coding
RefSeq status	Reviewed
Organism	*Homo sapiens*
Lineage	*Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo*
Also known as	CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C
Summary	The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Straussler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.

Genomic regions, transcripts, and products

Go to reference sequence details

Entrez Gene Home

Table Of Contents
Summary
Genomic regions, transcripts...
Genomic context
Bibliography
HIV-1 protein interactions
Interactions
General gene information
General protein information
Reference Sequences
Related Sequences
Additional Links

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

NCBI **Single Nucleotide Polymorphism**

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

Search Entrez SNP for Go

BUILD 126

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

Go

GENERAL
Contact Us
dbSNP Homepage
SNP Science Primer
Announcements
dbSNP Summary
FTP Download
Build History

SNP SUBMISSION
How to Submit
Handle Request

DOCUMENTATION
Build Release Note
Search FAQ Archive
updated 2006
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 mRNA transcript):	Contig Label	Contig	mrna	protein	mrna orientation	transcript	snp count
	reference	NT_011387	NM_000311	NP_000302	forward	plus strand 21, coding	

Color Legend

Region	Contig position	mRNA pos	dbSNP rs# cluster id	Hetero-zygosity	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.		Yes		contig reference	A	Asp [D]	2	178
	4620405	rs11538767	N.D.		Yes		nonsynonymous	C	Ala [A]	2	180
			N.D.		Yes		contig reference	T	Val [V]	2	180
	4620464	rs28933385	N.D.		Yes		nonsynonymous	A	Lys [K]	1	200
			N.D.		Yes	Yes	contig reference	G	Glu [E]	1	200
	4620521	rs1800014	N.D.		Yes		nonsynonymous	A	Lys [K]	1	219
			N.D.		Yes		contig reference	G	Glu [E]	1	219
	4620538	rs6052773	N.D.		Yes		synonymous	T	Ala [A]	3	224
			N.D.		Yes		contig reference	C	Ala [A]	3	224
	4620545	rs17852079	N.D.		Yes		nonsynonymous	A	Lys [K]	1	227
			N.D.		Yes		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

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

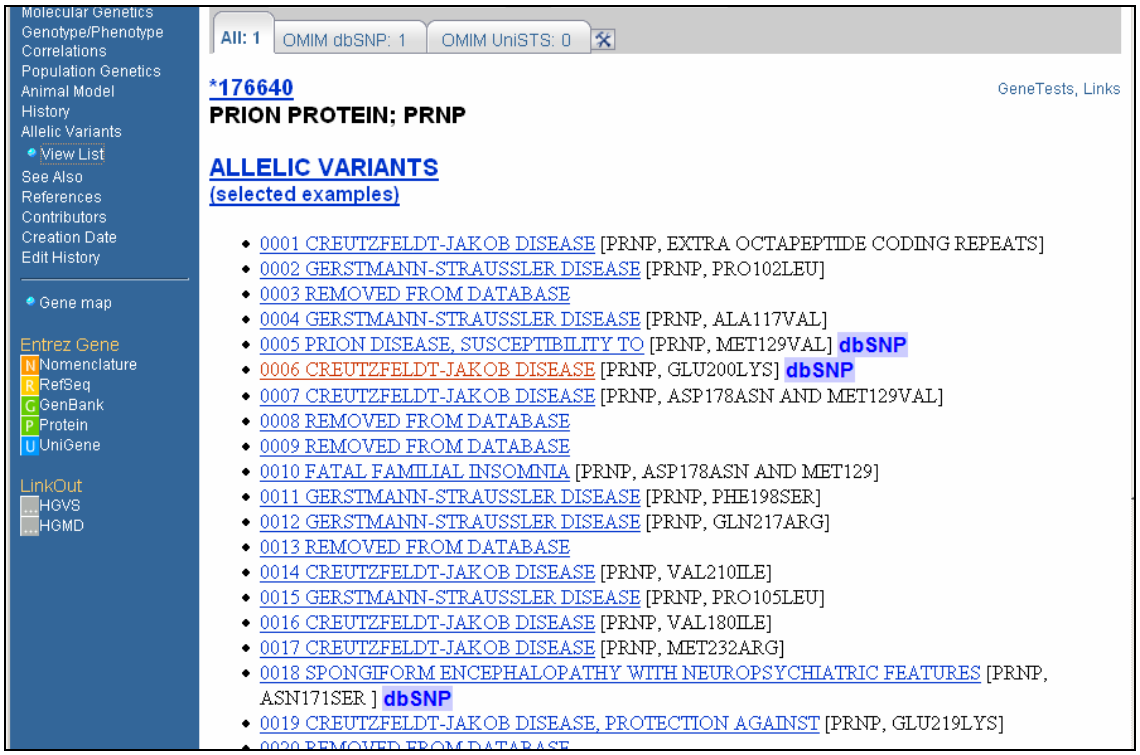
.0006 CREUTZFELDT-JAKOB DISEASE [PRNP, GLU200LYS] dbSNP

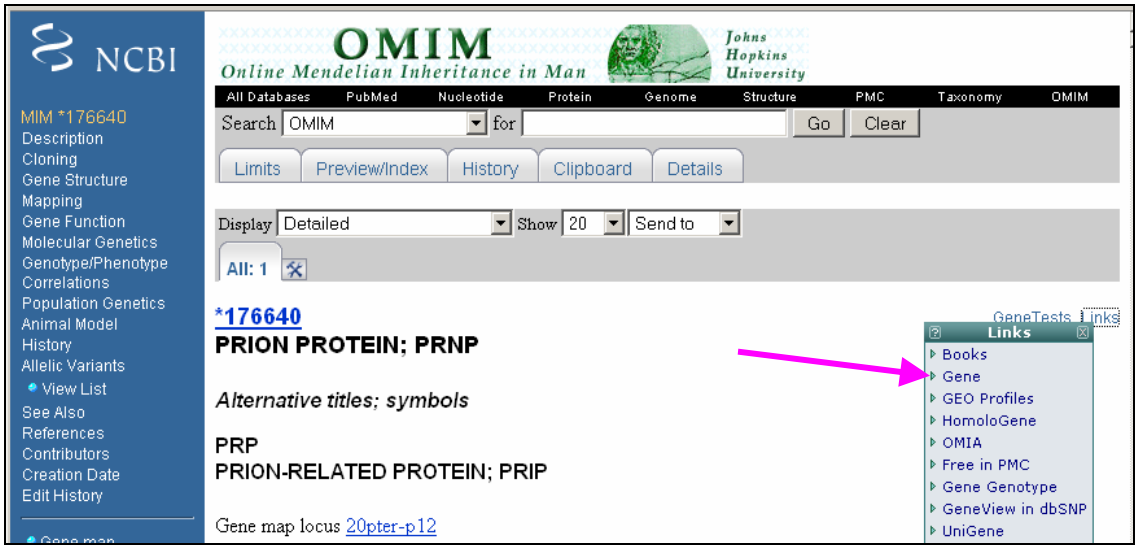
FATAL FAMILIAL INSOMNIA, INCLUDED

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

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

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


Molecular Genetics
Genotype/Phenotype
Correlations
Population Genetics
Animal Model
History
Allelic Variants
View List
See Also
References
Contributors
Creation Date
Edit History
Gene map
Entrez Gene
Nomenclature
RefSeq
GenBank
Protein
UniGene
LinkOut
HGVS
HGMD
All: 1
OMIM dbSNP: 1
OMIM UniSTS: 0
*176640
GeneTests, Links
PRION PROTEIN; PRNP
ALLELIC VARIANTS
(selected examples)
0001 CREUTZFELDT-JAKOB DISEASE [PRNP, EXTRA OCTAPEPTIDE CODING REPEATS]
0002 GERSTMANN-STRAUSSLER DISEASE [PRNP, PRO102LEU]
0003 REMOVED FROM DATABASE
0004 GERSTMANN-STRAUSSLER DISEASE [PRNP, ALA117VAL]
0005 PRION DISEASE, SUSCEPTIBILITY TO [PRNP, MET129VAL] dbSNP
0006 CREUTZFELDT-JAKOB DISEASE [PRNP, GLU200LYS] dbSNP
0007 CREUTZFELDT-JAKOB DISEASE [PRNP, ASP178ASN AND MET129VAL]
0008 REMOVED FROM DATABASE
0009 REMOVED FROM DATABASE
0010 FATAL FAMILIAL INSOMNIA [PRNP, ASP178ASN AND MET129]
0011 GERSTMANN-STRAUSSLER DISEASE [PRNP, PHE198SER]
0012 GERSTMANN-STRAUSSLER DISEASE [PRNP, GLN217ARG]
0013 REMOVED FROM DATABASE
0014 CREUTZFELDT-JAKOB DISEASE [PRNP, VAL210ILE]
0015 GERSTMANN-STRAUSSLER DISEASE [PRNP, PRO105LEU]
0016 CREUTZFELDT-JAKOB DISEASE [PRNP, VAL180ILE]
0017 CREUTZFELDT-JAKOB DISEASE [PRNP, MET232ARG]
0018 SPONGIFORM ENCEPHALOPATHY WITH NEUROPSYCHIATRIC FEATURES [PRNP, ASN171SER] dbSNP
0019 CREUTZFELDT-JAKOB DISEASE, PROTECTION AGAINST [PRNP, GLU219LYS]


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
Limits
Preview/Index
History
Clipboard
Details
Display Detailed
Show 20
Send to
All: 1
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
*176640
PRION PROTEIN; PRNP
Alternative titles; symbols
PRP
PRION-RELATED PROTEIN; PRIP
Gene map locus 20pter-p12
GeneTests, Links
Links
Books
Gene
GEO Profiles
HomoloGene
OMIA
Free in PMC
Gene Genotype
GeneView in dbSNP
UniGene

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

Links
PROTEIN LINKS
- FASTA
- GENPEPT
- Blink
- Conserved Domains

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

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

▼ **Entrez Gene Info**
▶ **Feedback**
▼ **Subscriptions**

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 | GI list | Run BLAST

200 BLAST hits to 48 unique species Sort by taxonomy proximity

0 Archaea | 0 Bacteria | 194 Metazoa | 0 Fungi | 0 Plants | 0 Viruses | 0 Other Eukaryotae

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

253 aa

SCORE	P	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	AAX37089	60834334	prion protein [synthetic construct]
1432	1	AAX42953	61367107	prion protein [synthetic construct]
1432	1	AAV38282	54695820	prion protein (p27-30) (Creutzfeld-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

Get Cn3D Now!

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

43 BLAST hits to 12 unique species Sort by taxonomy proximity

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

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

253 aa

SCORE	P	ACCESSION	GI	PROTEIN DESCRIPTION
Conserved Domain Database hits				
1203	•	1QLZA	6730487	Chain A, Human Prion Protein
1203	•	1QLXA	6730485	Chain A, Human Prion Protein
1136	•	1DX1A	9955174	Chain A, Bovine Prion Protein Residues 23-230
1136	•	1DX0A	9955173	Chain A, Bovine Prion Protein Residues 23-230
771	•	1QM1A	6730489	Chain A, Human Prion Protein Fragment 90-230
771	•	1QM0A	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 Pri
604	•	1QM3A	6730491	Chain A, Human Prion Protein Fragment 121-230
604	•	1QM2A	6730490	Chain A, Human Prion Protein Fragment 121-230
587	•	1I4MA	20150089	Chain A, Crystal Structure Of The Human Prion Protein Reveals A M
584	•	1DWZA	9955172	Chain A, Bovine Prion Protein Fragment 121-230
584	•	1DWYA	9955171	Chain A, Bovine Prion Protein Fragment 121-230
583	•	1H0LA	28373307	Chain A, Human Prion Protein 121-230 M166cE221C
576	•	1Y2SA	60594516	Chain A, Ovine Prion Protein Variant R168
569	•	1XYUA	60594486	Chain A, Solution Structure Of The Sheep Prion Protein With Polym

NCBI

Related Structures

HOME | SEARCH | SITE MAP | PubMed | Blast | Entrez Structure | Help

Query: prion protein preproprotein [Homo sapiens] [gi: 4506113]
Structure: 1QLZ Chain A, Human Prion Protein
Reference: [MMDB] [PubMed]

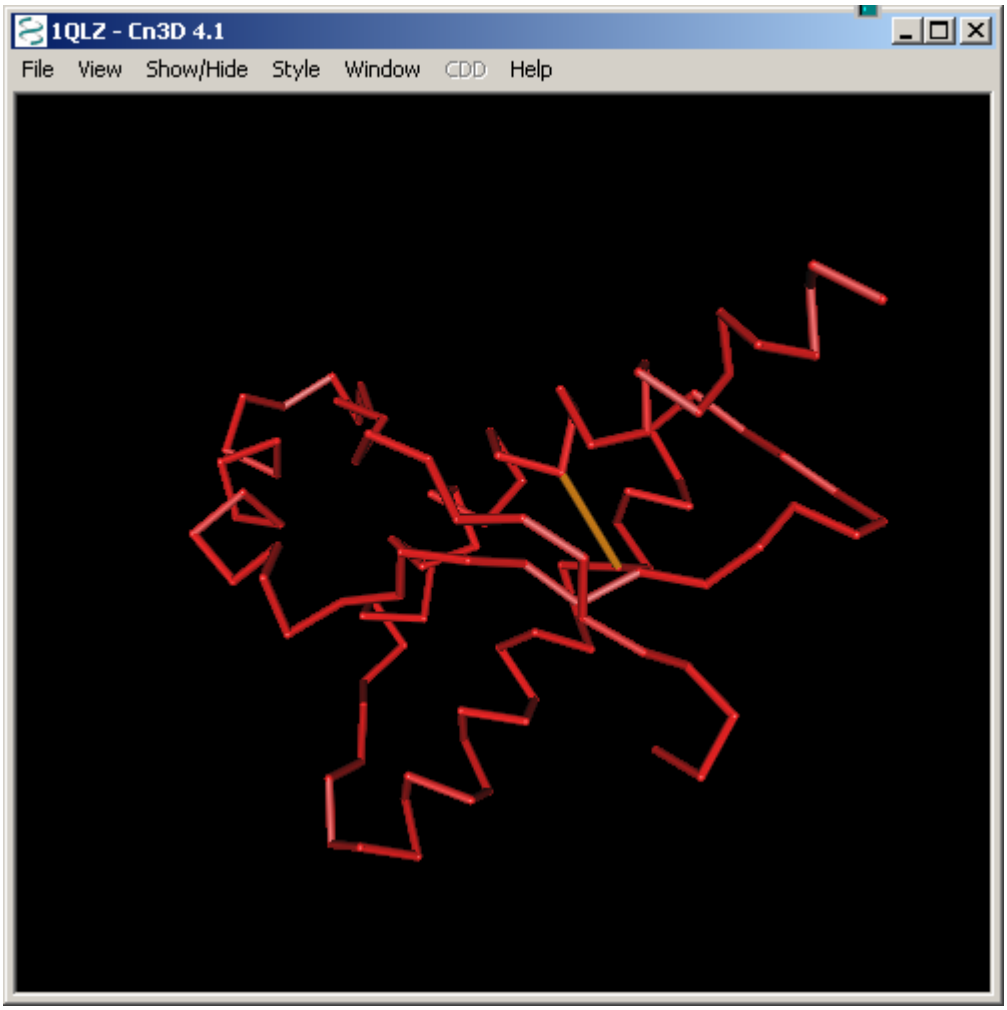
Get 3D Structure data | to: View in Cn3D | *(To display structure, download Cn3D)*

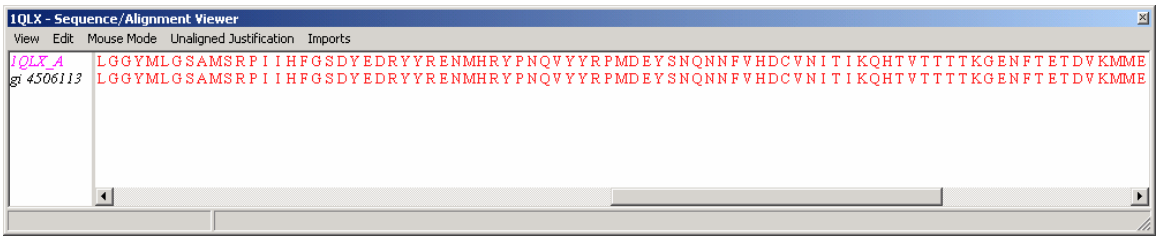
E-value = 1e-131, Bit score = 468, Aligned length = 208, Sequence Identity = 100%

```
                        10        20        30        40        50        60        70        80
                ....*....|....*....|....*....|....*....|....*....|....*....|....*....|....*....|
gi 4506113   23 KKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP 102
1QLZ A        3 KKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP 82

                        90       100       110       120       130       140       150       160
                ....*....|....*....|....*....|....*....|....*....|....*....|....*....|....*....|
gi 4506113  103 SKPKTNMKHMAGAAAAGAVVGGLGGYMLGSAMSRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNI 182
1QLZ A       83 SKPKTNMKHMAGAAAAGAVVGGLGGYMLGSAMSRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNI 162

                       170       180       190       200
                ....*....|....*....|....*....|....*....|....*...
gi 4506113  183 TIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYYQRGS 230
1QLZ A      163 TIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYYQRGS 210
```


1QLZ - Cn3D 4.1
File View Show/Hide Style Window CDD Help


1QLX - Sequence/Alignment Viewer
View Edit Mouse Mode Unaligned Justification Imports
1QLX_A LGGYMLGSAMSRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMME
gi 4506113 LGGYMLGSAMSRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMME

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 hightlight 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.