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.

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Search Gene for prion protein Preview Go Clear ☑ current records only

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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.
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Organism human Preview Index

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

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

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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.
- Click on query # to add to strategy

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

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☐ 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, CJD, GSS, MGC26679, 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*]

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

GeneID: 5621 Primary source: HGNC:9449 updated 12-Aug-2006

Summary

Official Symbol: PRNP **and Name**: prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) **provided by** HUGO Gene Nomenclature Committee

See related: HPRD:01453, MIM:176640

Gene type: protein coding

Gene name: PRNP

Gene description: prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)

RefSeq status: Reviewed

Organism: *Homo sapiens*

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

Gene aliases: 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

RefSeq below

NC_000020.9

[4615069 ▶ [4638234 ▶

5' 3'

NM_000311.2 NP_000302 preproprotein CCDS13080.1

NM_183079.1 NP_898902 preproprotein CCDS13080.1

■ - coding region ■ - untranslated region

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☐ 1: P 27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndr nnia) [*Homo sapiens*]

GeneID TC:9449 updated 12-Aug-2006

Sum

Officia prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fan UGO Gene Nomenclature Committee

See rel 6640

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GeneID: 5621 Primary source: HGNC:9449 updated 07-Aug-2006

RefSeq status: Reviewed

total gene size: 15166 bp

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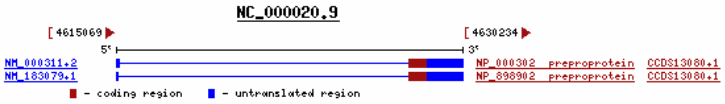
Genomic regions, transcripts, and products



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		Coding EXON		INTRON	
coords	length	coords	length	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: 254 aa, number of exons: 1

EXON	Coding EXON	INTRON

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☐ 1: P 27-30) (Creutzfeld-Jakob disease, Gerstmann-Strausler-Scheinker syndr mnia) [*Homo sapiens*]

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Genomic context

See PRNP in MapViewer

chromosome: 20; **Location:** 20p13

Bibliography

Gene References into Function (GeneRIF): **Submit**

PubMed links

GeneRIFs:

1. Sequencing the PRNP gene in Caco-2 and HT-29 parental and clonal cell lines, revealed that these cells have a distinct polymorphism at codon 129 and PrPc is expressed in lipid rafts. PubMed
2. Finds polymorphism at codon 129 of prion protein gene confers similar genetic susceptibility to vCJD in Brazilian populations as in European and Asian countries. PubMed
3. the presence of Prnp variant alleles was found not to be significantly associated to cognitive performance of patients with mesial temporal lobe epilepsy with hippocampal sclerosis PubMed
4. protease-resistant prion protein (PrPres) in spontaneous lymphoreticular tumors of mice infected PubMed

95. Polymorphism of the codon 129 of the prion protein (PrP) gene and neuropathology of cerebral ageing. PubMed

96. a specific prion protein fragment has a role in disulfide stability by retarding the rate of fibril formation PubMed

97. fibrillogenesis of PrP (195-213) PubMed

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

Interactions

Description .....

PRNP 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

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:677676; RH14068; RH63750; SHGC-12813; UTR-03221; WI-7784; gdb:120720; stSG10911
RH71030(e-PCR) (Links: UniSTS:34672)
Alternate names: GDB:177793; stSG20232
RH70248(e-PCR) (Links: UniSTS:43453)
Alternate name: T27631
RH71032(e-PCR) (Links: UniSTS:58320)
Alternate names: GDB:513003; stSG20379
GDB:181561(e-PCR) (Links: UniSTS:155287)
GDB:607655(e-PCR) (Links: UniSTS:158256)
PRNP-2(e-PCR) (Links: UniSTS:254000)
PMC136957P1(e-PCR) (Links: UniSTS:270809)

GeneOntology

Provided by GOA

Function	Evidence	
copper ion binding	TAS	PubMed
microtubule binding	IDA	PubMed
Process		
copper ion homeostasis	NAS	PubMed
metabolism	TAS	PubMed
response to oxidative stress	ISS	
Component		
Golgi apparatus	ISS	
endoplasmic reticulum	ISS	
extrinsic to membrane	TAS	PubMed
lipid raft	ISS	

Homology:
Mouse, Rat
Map Viewer

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

General protein information

Names: prion protein
CD230 antigen; prion protein PrP; major prion protein; prion-related protein; prion protein (p27-30) (Creutzfeld-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)

NCBI Reference Sequences (RefSeq)

mRNA Sequence NM_000311
Transcriptional Variant
Transcript Variant: This variant (1) represents the longer transcript. Variants 1 and 2 encode the same protein.
Source Sequence BC022532,BG397054,M13899
Product NP_000302 prion protein preproprotein
Consensus CDS (CCDS) CCDS13080.1 (preliminary)
Conserved Domains (1) summary

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1: **PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)** [*Homo sapiens*]
GeneID: 5621 Primary source: HGNC:9449 updated 12-Aug-2006

Summary

Official Symbol: PRNP **and Name:** prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) **provided by** HUGO Gene Nomenclature Committee
See related: HPRD:01453, MIM:176640
Gene type: protein coding
Gene name: PRNP
Gene description: prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
RefSeq status: Reviewed
Organism: *Homo sapiens*
Lineage: *Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires;*

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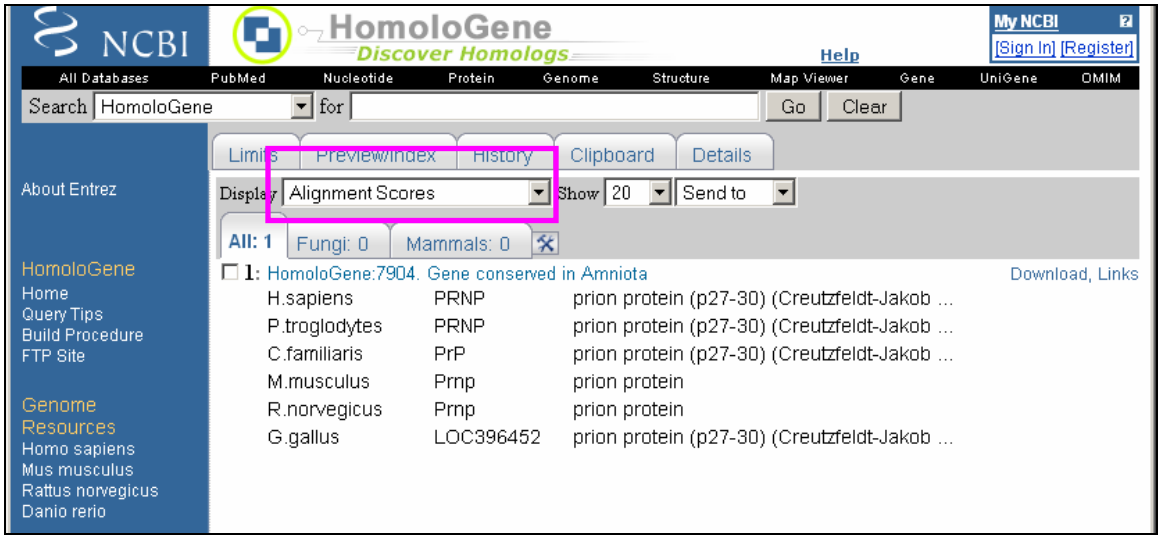
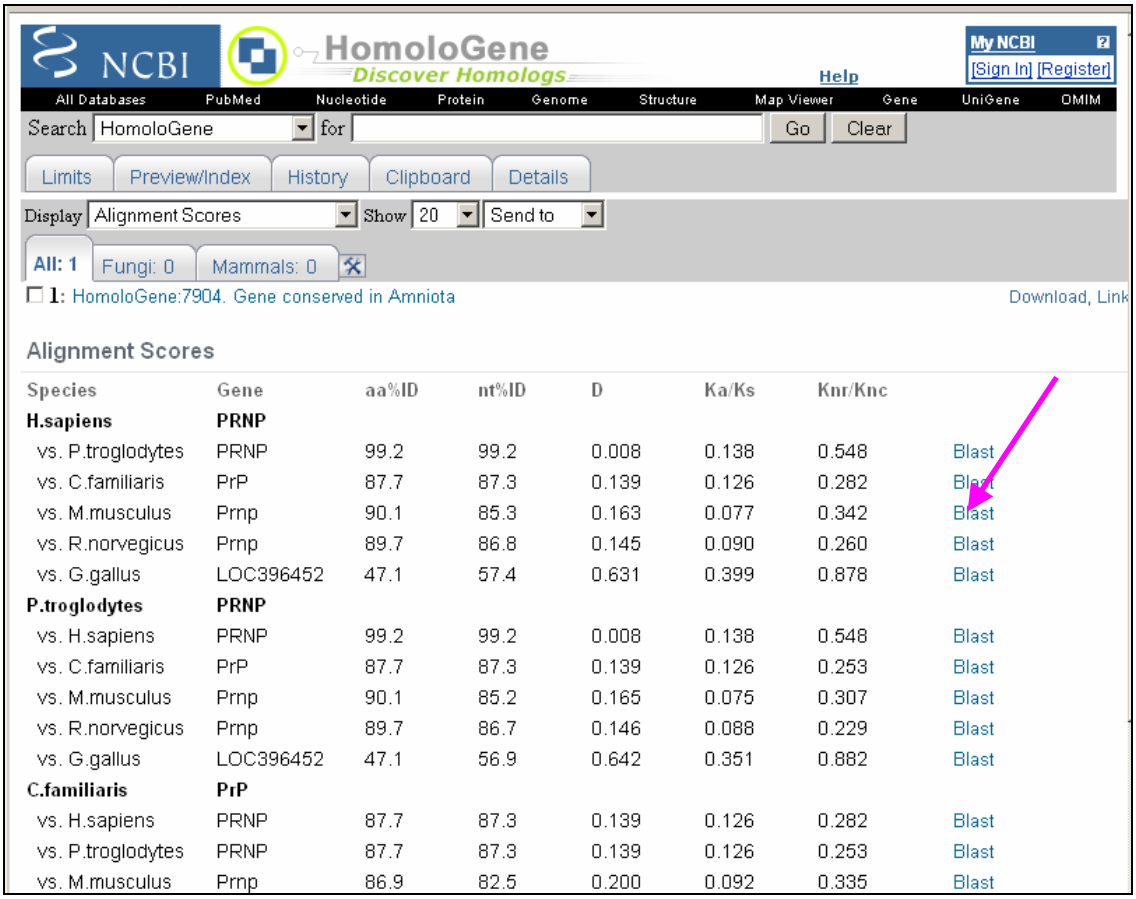




Species	Gene	aa%ID	nt%ID	D	Ka/Ks	Knr/Knc	
H.sapiens	**PRNP**						
vs. P.troglodytes	PRNP	99.2	99.2	0.008	0.138	0.548	Blast
vs. C.familiaris	PrP	87.7	87.3	0.139	0.126	0.282	Blast
vs. M.musculus	Prnp	90.1	85.3	0.163	0.077	0.342	Blast
vs. R.norvegicus	Prnp	89.7	86.8	0.145	0.090	0.260	Blast
vs. G.gallus	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**

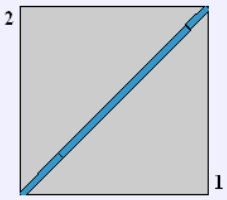
PubMed Entrez BLAST OMIM Taxonomy Structure

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

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|4506113|ref|NP_000302.1|prion protein preproprotein [Homo sapiens] >gi|34335270|ref|NP_898902.1| prion protein preproprotein [Homo sapiens] >gi|46095329|gb|AAS80162.1| prion protein [Homo sapiens] >gi|6996155|emb|CAB75503.1| PRNP [Homo sapiens] >gi|11079226|gb|AAG21693.1| prion protein [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
Length = 253 (1 .. 253)

Sequence 2: gi|13173473|ref|NP_035300.1|prion protein [Mus musculus] >gi|2865215|gb|AAC02804.1| short incubation prion protein Prnpa [Mus musculus] >gi|71060019|emb|CAJ18553.1| Prnp [Mus musculus] >gi|74192797|dbj|BAE34911.1| unnamed protein product [Mus musculus] >gi|74194091|dbj|BAE36950.1| unnamed protein product [Mus musculus] >gi|74213636|dbj|BAE35622.1| unnamed protein product [Mus musculus] >gi|74186646|dbj|BAE34788.1| unnamed protein product [Mus musculus] >gi|74182795|dbj|BAE34724.1| unnamed protein product [Mus musculus] >gi|13879449|gb|AAH06703.1| 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
Length = 254 (1 .. 254)



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

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

GeneID: 5621 Primary source: HGNC:9449 updated 07-Aug-2006

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- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- CCDS
- Evidence Viewer
- GDB

Summary

Official Symbol: PRNP **and Name:** prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) **provided by** HUGO Gene Nomenclature Committee
See related: HPRD:01453, MIM:176640
Gene type: protein coding
Gene name: PRNP
Gene description: prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
RefSeq status: Reviewed
Organism: *Homo sapiens*
Lineage: *Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo*
Gene aliases: 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

RefSeq below

NCBI **Single Nucleotide Polymorphism**

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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

OMIM
Online Mendelian Inheritance in Man
Johns Hopkins University

PubMed Nucleotide Protein Genome Structure PMC Taxonomy OMIM

Search OMIM for Go Clear
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Display Detailed Show: 20 Send to Text

MIM *176640
Text
Gene Structure
Mapping
Molecular Genetics
Genotype/Phenotype
Correlations
Population Genetics
Animal Model
History
Allelic Variants
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Contributors
Creation Date
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Gene map

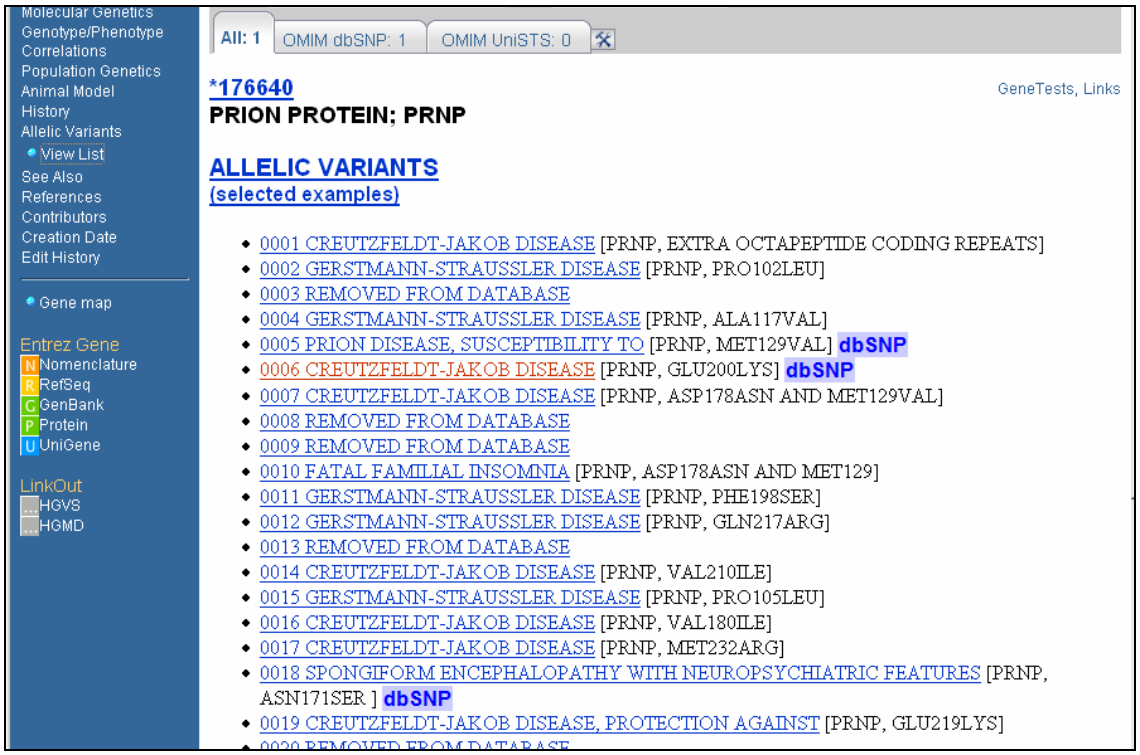
*176640 GeneTests,

PRION PROTEIN; PRNP

Alternative titles; symbols

PRP
PRION-RELATED PROTEIN; PRIP

Gene map locus 20pter-p12


Molecular Genetics
Genotype/Phenotype
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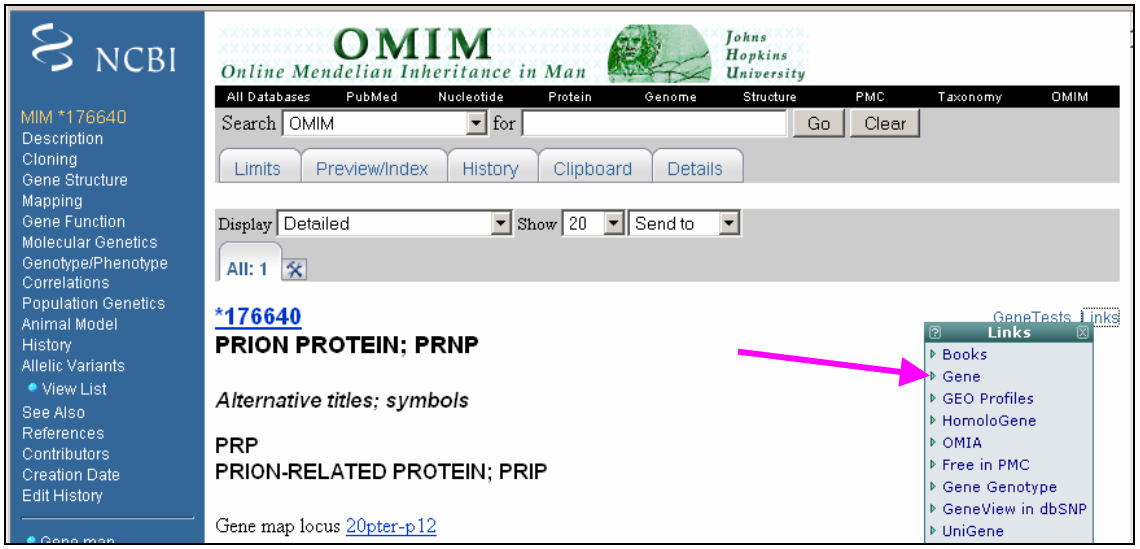
Gene map
Entrez Gene
Nomenclature
RefSeq
GenBank
Protein
UniGene
LinkOut
HGVS
HGMD
All: 1
OMIM dbSNP: 1
OMIM UniSTS: 0
*176640
GeneTests, Links
PRION PROTEIN; PRNP
ALLELIC VARIANTS
(selected examples)
0001 CREUTZFELDT-JAKOB DISEASE [PRNP, EXTRA OCTAPEPTIDE CODING REPEATS]
0002 GERSTMANN-STRAUSSLER DISEASE [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
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All Databases
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PRION-RELATED PROTEIN; PRIP
Gene map locus 20pter-p12
GeneTests, Links
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Books
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GEO Profiles
HomoloGene
OMIA
Free in PMC
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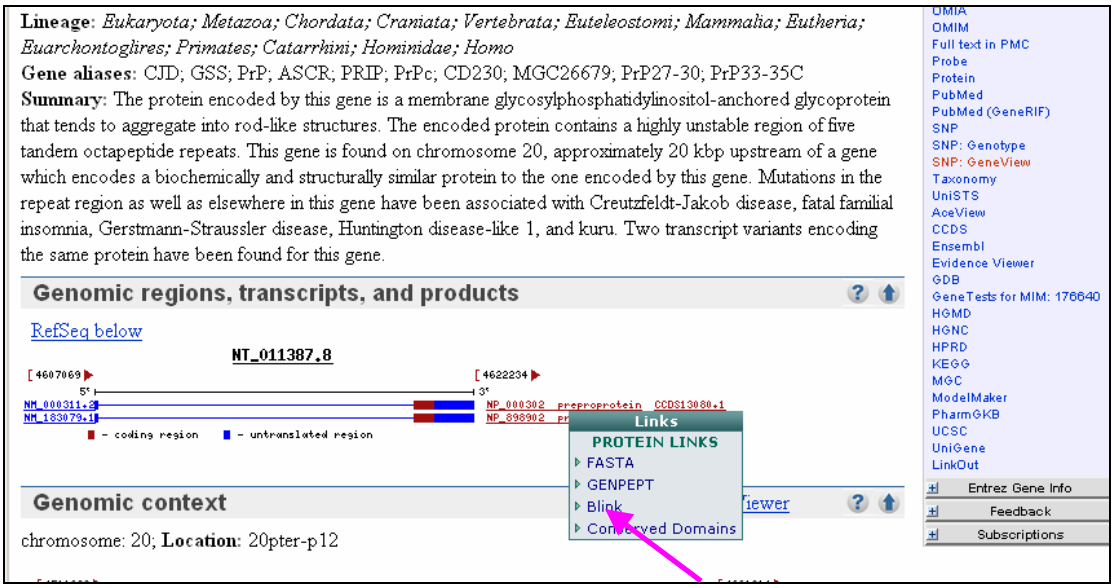
UniGene


Lineage: Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Catarrhini; Hominidae; Homo
Gene aliases: 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
RefSeq below
NT_011387.8
Links
PROTEIN LINKS
FASTA
GENPEPT
Blink
Conserved Domains
Genomic context
chromosome: 20; Location: 20pter-p12
OMIA
OMIM
Full text in PMC
Probe
Protein
PubMed
PubMed (GeneRIF)
SNP
SNP: Genotype
SNP: GeneView
Taxonomy
UniSTS
AceView
CCDS
Ensembl
Evidence Viewer
GDB
GeneTests for MIM: 176640
HGMD
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HPRD
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ModelMaker
PharmGKB
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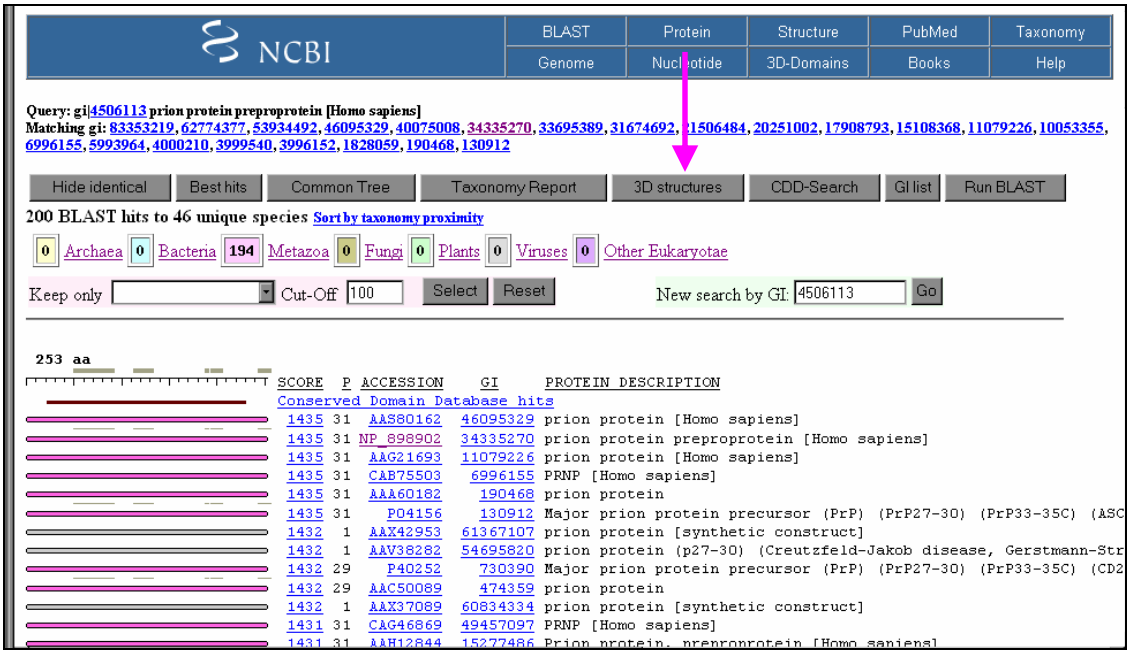
NCBI
BLAST
Protein
Structure
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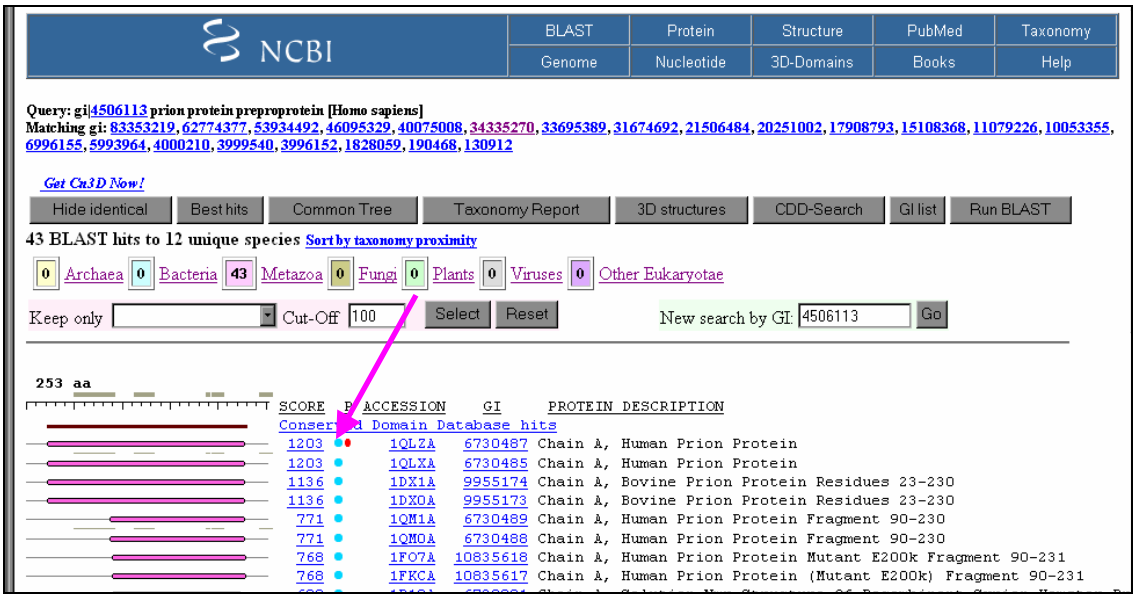
Books
Help
Query: gi|4506113 prion protein preprotein [Homo sapiens]
Hide identical
Best hits
Common Tree
Taxonomy Report
3D structures
CDD-Search
GI list
Run BLAST
200 BLAST hits to 46 unique species Sort by taxonomy proximity
Archaea
Bacteria
Metazoa
Fungi
Plants
Viruses
Other Eukaryotae
Keep only
Cut-Off
Select
Reset
New search by GI:
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) (ASC
1432 1 AAX42953 61367107 prion protein [synthetic construct]
1432 1 AAV38282 54695820 prion protein (p27-30) (Creutzfeld-Jakob disease, Gerstmann-Str
1432 29 P40252 730390 Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD2
1432 29 AAC50089 474359 prion protein
1432 1 AAX37089 60834334 prion protein [synthetic construct]
1431 31 CAG46869 49457097 PRNP [Homo sapiens]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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
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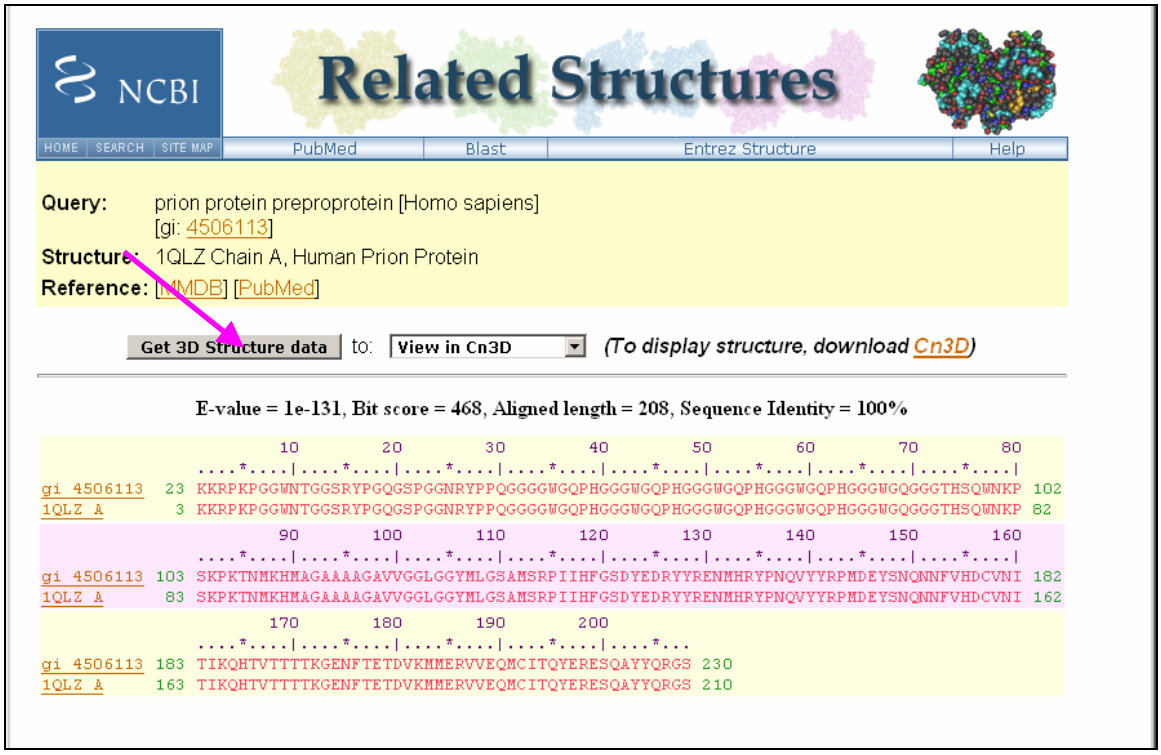
1203 1QLXA 6730485 Chain A, Human Prion Protein
1136 1DX1A 9955174 Chain A, Bovine Prion Protein Residues 23-230
1136 1DX0A 9955173 Chain A, Bovine Prion Protein Residues 23-230
771 1QM1A 6730489 Chain A, Human Prion Protein Fragment 90-230
771 1QM0A 6730488 Chain A, Human Prion Protein Fragment 90-230
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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SEARCH
SITE MAP
PubMed
Blast
Entrez Structure
Help
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[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)
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1QLZ A 3 KKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP 82
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gi 4506113 183 TIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYYQRGS 230
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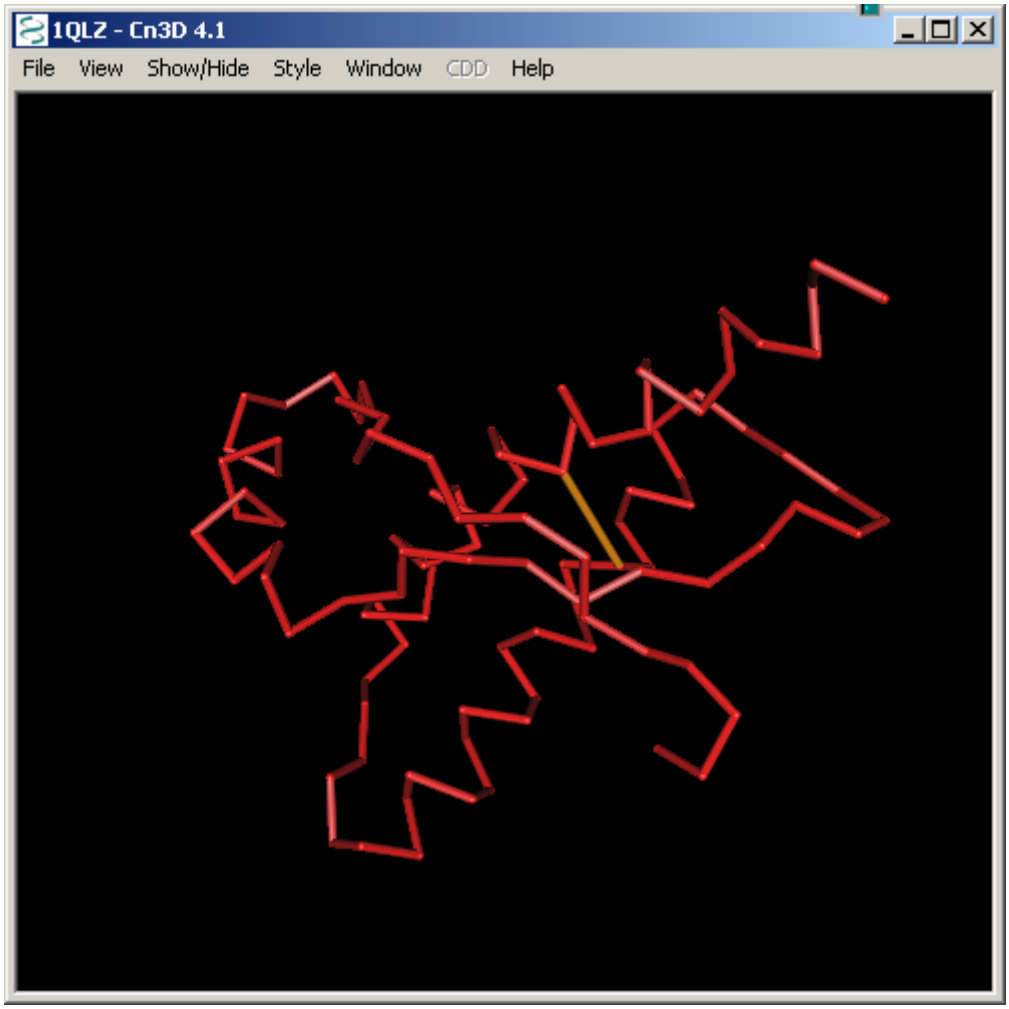
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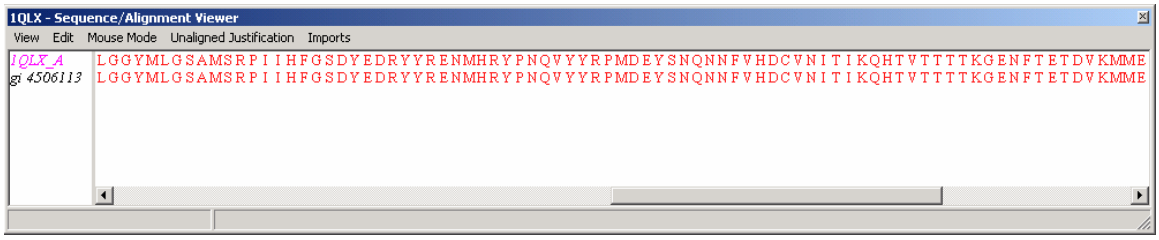
File View Show/Hide Style Window CDD Help


1QLX - Sequence/Alignment Viewer
View Edit Mouse Mode Unaligned Justification Imports
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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.