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

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

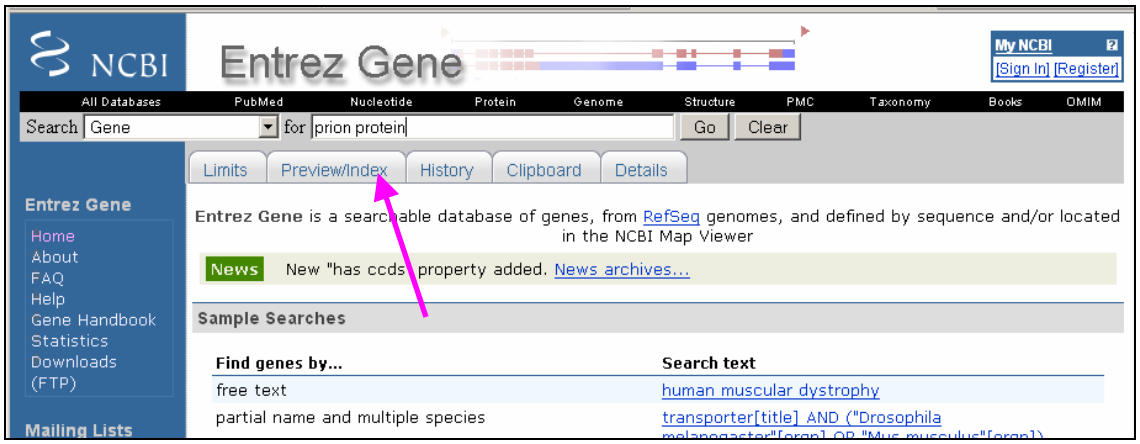
Retrieve human entries related to "prion protein" in Entrez Gene. Identify the gene for prion protein (PRNP). Name the map location of this gene on the human genome. What is the function of this protein? What are the alternate gene symbols? Name the phenotypes associated with the mutations in this gene.

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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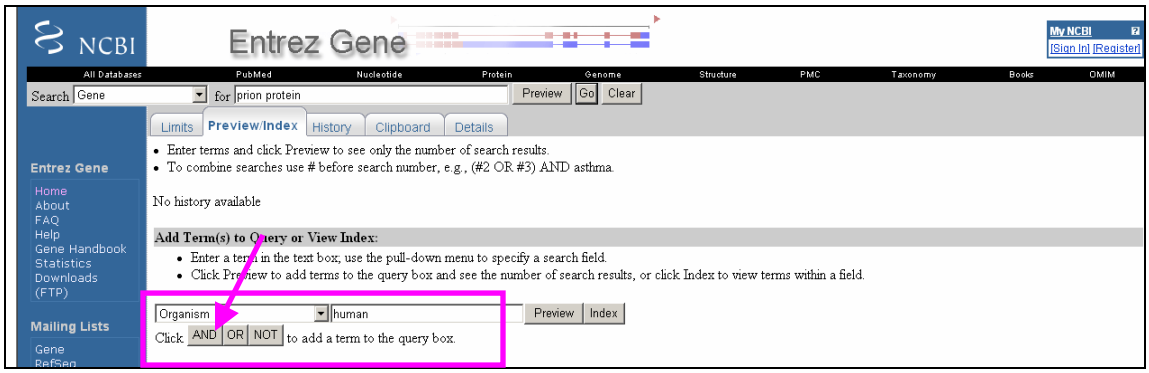
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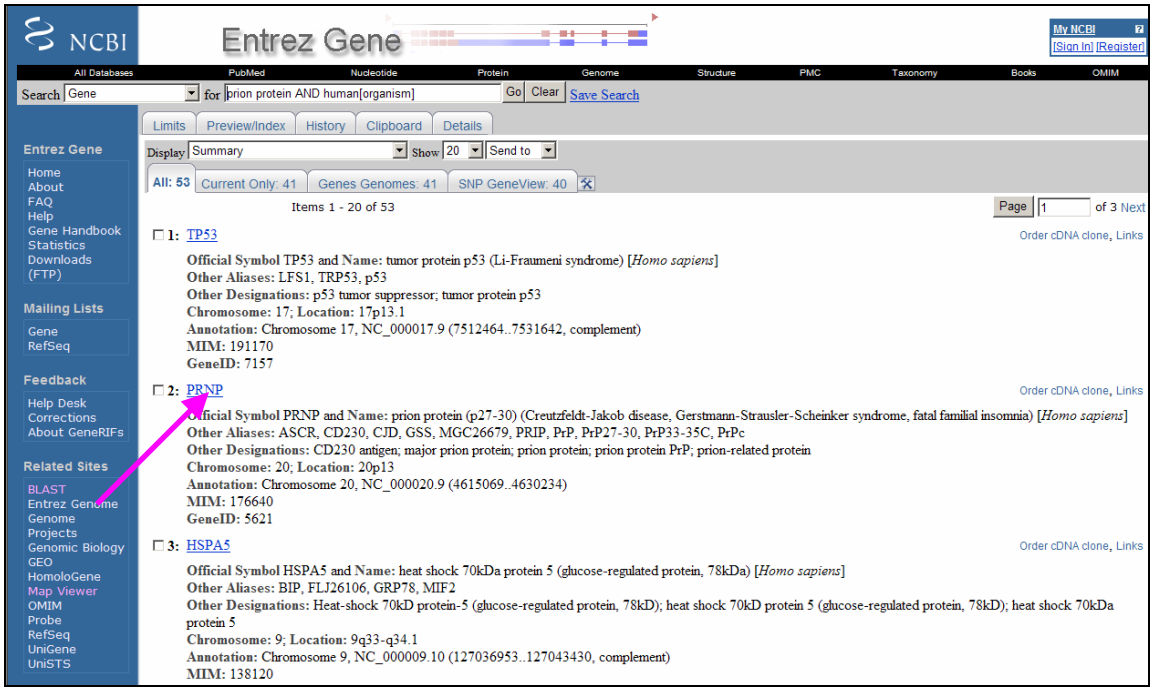
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HomoloGene
Map Viewer
OMIM
Probe
RefSeq
UniGene
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1: TP53
Order cDNA clone, Links
Official Symbol TP53 and Name: tumor protein p53 (Li-Fraumeni syndrome) [Homo sapiens]
Other Aliases: LFS1, TRP53, p53
Other Designations: p53 tumor suppressor; tumor protein p53
Chromosome: 17; Location: 17p13.1
Annotation: Chromosome 17, NC_000017.9 (7512464..7531642, complement)
MIM: 191170
GeneID: 7157
2: PRNP
Order cDNA clone, Links
Official Symbol PRNP and Name: prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [Homo sapiens]
Other Aliases: ASCR, CD230, CJD, GSS, MGC26679, PRIP, PrP, PrP27-30, PrP33-35C, PrPc
Other Designations: CD230 antigen; major prion protein; prion protein; prion protein PrP; prion-related protein
Chromosome: 20; Location: 20p13
Annotation: Chromosome 20, NC_000020.9 (4615069..4630234)
MIM: 176640
GeneID: 5621
3: HSPA5
Order cDNA clone, Links
Official Symbol HSPA5 and Name: heat shock 70kDa protein 5 (glucose-regulated protein, 78kDa) [Homo sapiens]
Other Aliases: BIP, FLJ26106, GRP78, MIF2
Other Designations: Heat-shock 70kD protein-5 (glucose-regulated protein, 78kD); heat shock 70kD protein 5 (glucose-regulated protein, 78kD); heat shock 70kDa protein 5
Chromosome: 9; Location: 9q33-q34.1
Annotation: Chromosome 9, NC_000009.10 (127036953..127043430, complement)
MIM: 138120

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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
[*Homo sapiens*]
GeneID: 5621 updated 28-Oct-2007

Summary

Official Symbol	PRNP provided by HGNC
Official Full Name	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) provided by HGNC
Primary source	HGNC:9449
See related	Ensembl:ENSG00000171867; HPRD:01453; MIM:176640
Gene type	protein coding
RefSeq status	Reviewed
Organism	*Homo sapiens*
Lineage	*Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo*
Also known as	CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C
Summary	The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Straussler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.

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- Protein

Genomic regions, transcripts, and products

Go to reference sequence details

NC_000020.9

[4615069] [4630234]

NM_000311.2 NM_183079.1 NP_000302.1 preproprotein CCDS13880.1 NP_898902.1 preproprotein CCDS13880.1

- coding region - untranslated region

Genomic context

chromosome: 20; Location: 20p13 See PRNP in MapViewer

[4519639] [4669314]

RPL7AL2 RPS4L2 PRNP PRND PRNT

- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- CCDS
- Evidence Viewer
- GDB
- GeneTests for MIM: 176640
- HGMD
- HGNC
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NIH cDNA clone links
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1: tzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
updated 28-Oct-2007

provided by HGNC

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

provided by HGNC

171867; HPRD:01453; MIM:176640

a; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires;
ini; Catarrhini; Hominidae; Homo

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

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-like structures. The encoded protein contains a highly unstable region of five tandem
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hically and structurally similar protein to the one encoded by this gene. Mutations in the repeat
region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial

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NCBI Entrez Gene

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

GeneID: 5621 updated 28-Oct-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	253	1
NM_183079.1	2464	2	NP_898902.1	253	1

Exon information:

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

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

EXON 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		

NCBI Entrez Gene

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OMIM Links
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PMC Links
Probe Links
Protein Links

Genomic context

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

See PRNP in MapViewer

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GeneRIFs: Gene References Into Function

What's a GeneRIF?

1. analysis of species-specific differences in the intermediate states of human and Syrian hamster prion protein detected by high pressure NMR spectroscopy
2. A South African family had a progressive dementia and atypical pathology associated with kuru-like prion protein plaques. The original mutation in this family occurred on a PRNP allele encoding a 1-octapeptide repeat deletion polymorphism.
3. We found that rPrP fibrils but not alpha-rPrP or soluble beta-sheet rich oligomers caused degeneration of neuronal processes. Degeneration of processes was accompanied by a collapse of microtubules and aggregation of cytoskeletal proteins.
4. Prion protein gene MM genotype increases late-onset Alzheimer's disease risk in Polish population
5. human brain PrP(C) interacts with selectins in a manner that is distinct from interactions in peripheral tissues; alternations in these interactions may have pathological consequences
6. This is the first publication of data that support the hypothesis that the common methionine/valine polymorphism at codon 129 of the PRNP gene may modify the susceptibility of women to mild temporal lobe epilepsy.
7. A novel three extra-repeat (72 bp) insertion within the octapeptide-coding region was identified in a Chinese family.
8. the PRNP polymorphism is more common in the Korean than in the Japanese population
9. plasmin cleaves PrP(c) in vitro and the liberated NH(2)-terminal fragment accelerates plasminogen activation

Submit: New GeneRIF Correction

HIV-1 protein interactions

Protein	Interaction	
1.Tat	HIV-1 Tat binds to a stem-loop structure in the mRNA of prion protein (PrP) that is similar to HIV-1 TAR RNA and infection of astrocytes with HIV-1 results in an increased level of PrP mRNA, suggesting Tat upregulates PrP expression	PubMed

Go to the HIV-1, Human Protein Interaction Database

Interactions

Description .....

Product	Interactant	Other Gene	Complex	Source	Pubs
NP_000302.1	NP_001155.1	APBB1		HPRD	PubM
NP_000302.1	NP_055759.2	CLSTN1		HPRD	PubM
NP_000302.1	NP_001822.2	CLU		HPRD	PubM
NP_000302.1	NP_001834.2	CNTN1		HPRD	PubM
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	PubM
PrPc interacts with CSNK2A2 (CK2 alpha prime). This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2A2 (CK2 alpl prime).					
NP_000302.1	NP_001887.1	CSNK2A2		BIND	PubM
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	PubM
NP_000302.1	NP_004399.1	DNM1		HPRD	PubM
NP_000302.1	NP_570629.1	DPP6		HPRD	PubM
NP_000302.1	NP_002077.1	GRB2		HPRD	PubM
PrPc interacts with HSPA5 (BiP).					
NP_000302.1	NP_005338.1	HSPA5		BIND	PubM
PrPc interacts with HSPD1 (Hsp60). This interaction was modeled on a demonstrated interaction between hamster PrPc and human HSPD1 (Hsp60).					
NP_000302.1	NP_002147.2	HSPD1		BIND	PubM
NP_000302.1	NP_000416.1	L1CAM		HPRD	PubM

General gene information

Markers

WI-18738(e-PCR)
Links: UniSTS:1017
Alternate names: HSA.55; RH57301; STS-D00015

SGC44304(e-PCR)
Links: UniSTS:2335
Alternate names: EST498946; RH57429

D20S1014(e-PCR)
Links: UniSTS:21619
Alternate names: G00-677-676; GDB:120720; GDB:677676; RH14068; RH63750; SHGC-12813; stSG10911; UTR-03221; WI-7784

RH71030(e-PCR)
Links: UniSTS:34672
Alternate names: GDB:177793; stSG20232

RH47809(e-PCR)
Links: UniSTS:38471
Alternate name: stSG28721

RH70248(e-PCR)
Links: UniSTS:43453
Alternate name: T27631

Genotypes

See PRNP SNP GeneView Report
See PRNP SNP Genotype Report

Phenotypes

Phenotypes

Creutzfeldt-Jakob disease
MIM: 123400
Gerstmann-Straussler disease
MIM: 137440
Huntington disease-like 1
MIM: 603218
Insomnia, fatal familial
MIM: 600072
Prion disease with protracted course
MIM: 606688

Pathways

KEGG pathway: Neurodegenerative Disorders
01510
KEGG pathway: Prion disease
05060

Homology

Mouse, Rat
Map Viewer

GeneOntology

Provided by GOA

Function	Evidence	
GPI anchor binding	IEA	
copper ion binding	TAS	PubMed
microtubule binding	IDA	PubMed
protein binding	IPI	PubMed

Process	Evidence	
cellular copper ion homeostasis	NAS	PubMed
metabolic process	TAS	PubMed
response to oxidative stress	ISS	

Component	Evidence	
Golgi apparatus	ISS	
cytoplasm	TAS	PubMed
endoplasmic reticulum	ISS	
extrinsic to membrane	TAS	PubMed
lipid raft	ISS	
membrane	IEA	
plasma membrane	ISS	

General protein information

General protein information

Names

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

NCBI Reference Sequences (RefSeq)

RefSeqs maintained independently of Annotated Genomes

These reference sequences exist independently of genome builds. Explain

mRNA and Protein(s)

1. **NM_000311.3→NP_000302.1 prion protein preproprotein**

Description	Transcript Variant: This variant (1) represents the longest transcript. Variants 1-5 encode the same protein.
Source sequence(s)	AW452130,BC022532,DA297032,M13899
Consensus CDS	CCDS13080.1
Conserved Domains (1)	summary
smart00157 Location:23–230 Blast Score:546	PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

2. **NM_001080121.1→NP_001073590.1 prion protein preproprotein**

Description	Transcript Variant: This variant (3) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	AW452130,BC022532,BP251427,DA122620,M13899
Conserved Domains (1)	summary
smart00157 Location:23–230 Blast Score:546	PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

3. **NM_001080122.1→NP_001073591.1 prion protein preproprotein**

Description	Transcript Variant: This variant (4) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	AW452130,BC022532,BI669189,DA297032,M13899
Conserved Domains (1)	summary
smart00157 Location:23–230 Blast Score:546	PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

4. **NM_001080123.1→NP_001073592.1 prion protein preproprotein**

Description	Transcript Variant: This variant (5) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	BC022532,DB461478,M13899
Conserved Domains (1)	summary
smart00157 Location:23–230 Blast Score:546	PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

5. **NM_183079.2→NP_898902.1 prion protein preproprotein**

Description	Transcript Variant: This variant (2) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
Source sequence(s)	AW452130,AY008282,BC022532,DA122620
Consensus CDS	CCDS13080.1
Conserved Domains (1)	summary
smart00157	PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in

Reference assembly

Genomic

1. **NC_000020.9 Reference assembly**
   Range 4615069..4630234
   Download GenBank FASTA
2. **NT_011387.8**
   Range 4607069..4622234
   Download GenBank FASTA

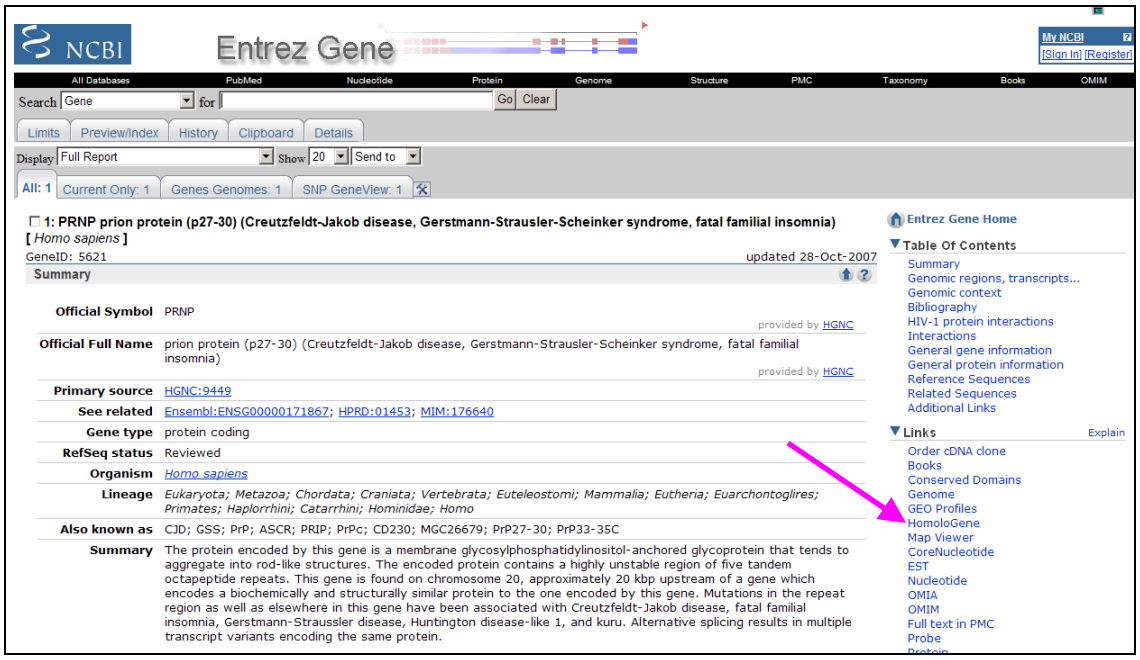
Alternate assembly (based on Celera assembly)

Genomic

1. **AC_000063.1 Alternate assembly (based on Celera assembly)**
   Range 4736784..4751948
   Download GenBank FASTA
2. **NW_927317.1**
   Range 4593960..4609124
   Download GenBank FASTA

Related Sequences

Nucleotide		Protein
Genomic	AF030575.1	AAC05365.1
Genomic	AF076976.1	AAD46098.1
Genomic	AF085477.2	AAC62750.2
Genomic	AF315723.1	None
Genomic	AL133396.2	CAB75503.1
		CAI19053.1
Genomic	AY219882.1	AAO83635.1
Genomic	AY219883.1	AAO83636.1
Genomic	AY458651.1	AAR21603.1
Genomic	CH471133.3	EAX10449.1
		EAX10450.1
Genomic	DQ408531.1	ABD63004.1
Genomic	M81929.1	AAB59442.1
Genomic	M81930.1	AAB59443.1
Genomic	S71208.1	AAB20521.1
Genomic	S71210.1	AAB20522.1
Genomic	S71212.1	AAB20523.1
Genomic	S79978.1	AAB35416.1
Genomic	S80539.1	AAB21334.1
Genomic	S80732.1	AAB50648.2
Genomic	S80743.1	AAB50649.2
Genomic	S83341.1	AAB50777.1
Genomic	U29185.1	AAC78725.1
Genomic	X83416.1	CAA58442.1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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
[Homo sapiens]
GeneID: 5621
updated 28-Oct-2007
Summary
Official Symbol PRNP
provided by HGNC
Official Full Name prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
provided by HGNC
Primary source HGNC:9449
See related Ensembl:ENSG00000171867; HPRD:01453; MIM:176640
Gene type protein coding
RefSeq status Reviewed
Organism Homo sapiens
Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C
Summary The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Straussler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein.
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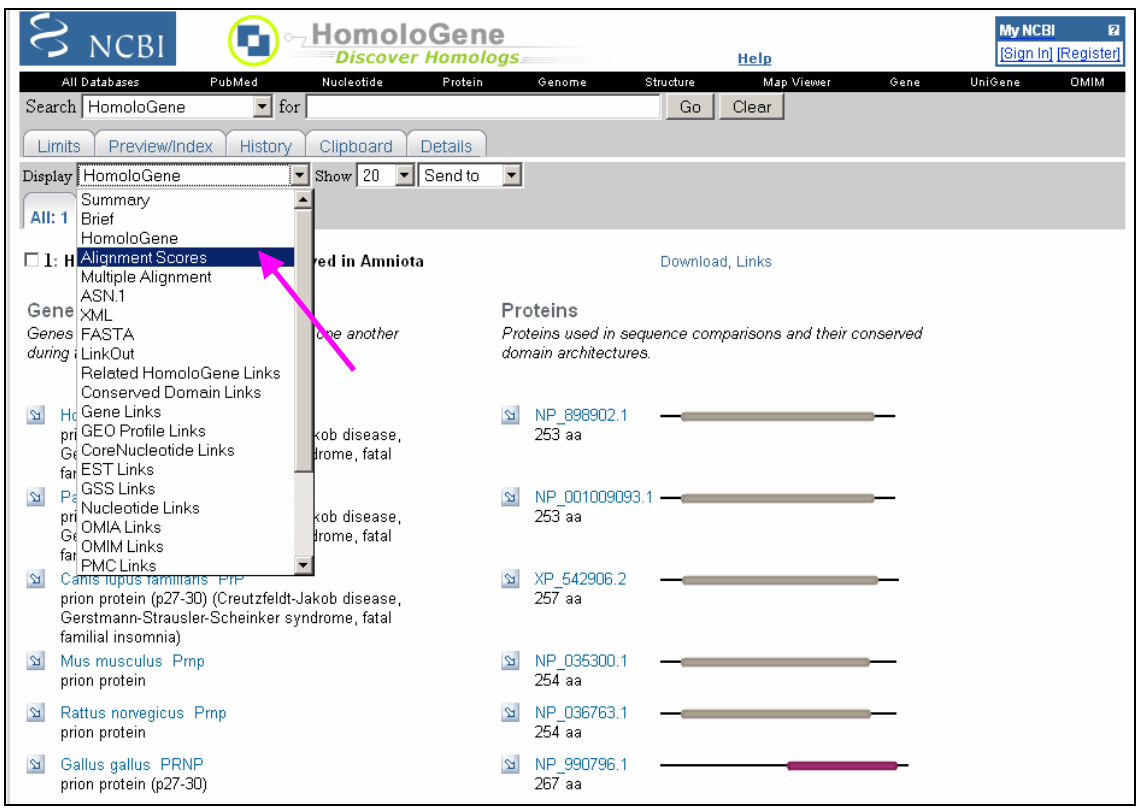
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Proteins
Proteins used in sequence comparisons and their conserved domain architectures.
NP_898902.1
253 aa
NP_001009093.1
253 aa
XP_542906.2
257 aa
prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
Mus musculus Prnp
prion protein
NP_035300.1
254 aa
Rattus norvegicus Prnp
prion protein
NP_036763.1
254 aa
Gallus gallus PRNP
prion protein (p27-30)
NP_990796.1
267 aa

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All: 1 | Fungi: 0 | Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota

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Alignment Scores

Gene		Identity (%)		Substitution Rates [1]			
Species	Symbol	Protein	DNA	d	d_N/d_S	d_{NR}/d_{NC}	
Homo sapiens	**PRNP**						
vs. Pan troglodytes	PRNP	99.2	99.2	0.008	0.138	0.548	Blast
vs. Canis lupus familiaris	PrP	87.7	87.3	0.139	0.126	0.282	Blast
vs. Mus musculus	Prnp	90.1	85.3	0.163	0.077	0.342	Blast
vs. Rattus norvegicus	Prnp	89.7	86.8	0.145	0.090	0.260	Blast
vs. Gallus gallus	PRNP	47.1	57.4	0.631	0.399	0.878	Blast
Pan troglodytes	**PRNP**						
vs. Homo sapiens	PRNP	99.2	99.2	0.008	0.138	0.548	Blast
vs. Canis lupus familiaris	PrP	87.7	87.3	0.139	0.126	0.253	Blast
vs. Mus musculus	Prnp	90.1	85.2	0.165	0.075	0.307	Blast
vs. Rattus norvegicus	Prnp	89.7	86.7	0.146	0.088	0.229	Blast
vs. Gallus gallus	PRNP	47.1	56.9	0.642	0.351	0.882	Blast
Canis lupus familiaris	**PrP**						
vs. Homo sapiens	PRNP	87.7	87.3	0.139	0.126	0.282	Blast

NCBI **Blast 2 Sequences results**

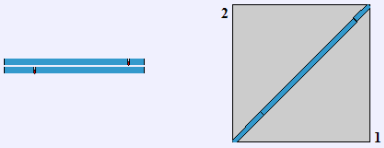
PubMed | Entrez | BLAST | OMIM | Taxonomy | Structure

BLAST 2 SEQUENCES RESULTS VERSION BLASTP 2.2.17 [Aug-26-2007]

Matrix BLOSUM62 gap open: 11 gap extension: 1
x_dropoff: 0 expect: 10.0000 wordsize: 3 Filter View option Standard
Masking character option X for protein, n for nucleotide Masking color option Black
Show CDS translation Align

Sequence 1: gi|34335270|prion protein preproprotein [Homo sapiens] >gi|4506113|ref|NP_000302.1| prion protein preproprotein [Homo sapiens] >gi|122056623|ref|NP_001073590.1| prion protein preproprotein [Homo sapiens] >gi|122056625|ref|NP_001073591.1| prion protein preproprotein [Homo sapiens] >gi|122056628|ref|NP_001073592.1| prion protein preproprotein [Homo sapiens] >gi|130912|sp|P04156|PRIO_HUMAN Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR) (CD230 antigen) >gi|190468|gb|AAA60182.1| prion protein >gi|69961...
Length = 253 (1 .. 253)

Sequence 2: gi|13173473|prion protein [Mus musculus] >gi|130914|sp|P04925|PRIO_MOUSE Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen) >gi|200529|gb|AAA39997.1| prion protein >gi|2865215|gb|AAC02804.1| short incubation prion protein Prnpa [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)



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

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

GeneID: 5621 updated 28-Oct-2007

Summary

Official Symbol	PRNP provided by HGNC
Official Full Name	prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) provided by HGNC
Primary source	HGNC:9449
See related	Ensembl:ENSG00000171867; HPRD:01453; MIM:176640
Gene type	protein coding
RefSeq status	Reviewed
Organism	*Homo sapiens*
Lineage	*Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo*
Also known as	CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C
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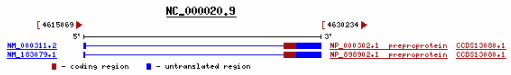
Genomic regions, transcripts, and products

Go to reference sequence details



Genomic context

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- SNP: GeneView
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NCBI **Single Nucleotide Polymorphism**

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SNP linked to Gene PRNP(geneID:5621) Via Contig Annotation

Send rs# on all gene models to Batch Query Download all rs# to file. GENE GENOTYPE REPORT

Gene Model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript): 4

mrna	transcript	protein	mrna orientation	Contig	Contig Label	List SNP
NM_000311.3	plus strand	NP_000302.1	forward	NT_011387.8	reference	<- currently shown
NM_000311.3	plus strand	NP_000302.1	forward	NW_927317.1	Celera	View snp on GeneModel
NM_183079.2	plus strand	NP_898902.1	forward	NT_011387.8	reference	View snp on GeneModel
NM_183079.2	plus strand	NP_898902.1	forward	NW_927317.1	Celera	View snp on GeneModel

in gene region cSNP has frequency double hit refresh

gene model (contig mRNA transcript):	Contig Label	Contig	mrna	protein	mrna orientation	transcript	snp count
	reference	NT_011387.8	NM_000311.3	NP_000302.1	forward	plus strand	22, 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_1	4619867	101						start codon				1
exon_2	4619942	176	rs11538755	N.D.				missense	A	Thr [T]	1	26
				N.D.				contig reference	C	Pro [P]	1	26
	4619949	183	rs11538762	N.D.				missense	A	His [H]	2	28
				N.D.			Yes	contig reference	C	Pro [P]	3	165
	4620378	612	rs16990018	0.039	H		Yes	missense	G	Ser [S]	2	171
				0.039	H		Yes	contig reference	A	Asn [N]	2	171
	4620399	633	rs11538766	N.D.	H		Yes	missense	T	Val [V]	2	178
				N.D.	H		Yes	contig reference	A	Asp [D]	2	178
	4620405	639	rs11538767	N.D.			Yes	missense	C	Ala [A]	2	180
				N.D.			Yes	contig reference	T	Val [V]	2	180
	4620464	698	rs28933385	N.D.			Yes	missense	A	Lys [K]	1	200
				N.D.			Yes	Yes contig reference	G	Glu [E]	1	200
	4620521	755	rs1800014	N.D.			Yes	missense	A	Lys [K]	1	219
				N.D.			Yes	contig reference	G	Glu [E]	1	219
	4620538	772	rs6052773	N.D.	H		Yes	synonymous	T	Ala [A]	3	224
				N.D.	H		Yes	contig reference	C	Ala [A]	3	224
	4620545	779	rs17852079	N.D.			Yes	missense	A	Lys [K]	1	227
				N.D.			Yes	contig reference	C	Gln [Q]	1	227
	4620625	859	rs11538759	N.D.				synonymous	G	Gly [G]	3	253
				N.D.				contig reference	A	Gly [G]	3	253

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***176640**
PRION PROTEIN; PRNP

Alternative titles; symbols

PRP
PRION-RELATED PROTEIN; PRIP

Gene map locus 20pter-p12

TEXT

DESCRIPTION

The PRNP gene encodes the prion protein, which has been implicated in various types of transmissible neurodegenerative spongiform encephalopathies. The human prion diseases occur in inherited, acquired, and sporadic forms. Approximately 15% are inherited and associated with coding mutations in the PRNP gene. Inherited prion diseases include familial Creutzfeldt-Jakob disease (CJD; 123400), Gerstmann-Straussler disease (GSD; 137440), and fatal familial insomnia (FFI; 600072). Acquired prion diseases include iatrogenic CJD, kuru (245300), variant CJD (vCJD) in humans, scrapie in sheep, and bovine spongiform encephalopathy (BSE) in cattle. Prion diseases are also referred to as transmissible spongiform encephalopathies (TSE). Variant CJD is believed to be acquired from cattle infected with BSE. However, the majority of human cases of prion disease occur as sporadic CJD (sCJD) (Collinge et al., 1996; Parchi et al., 2000; Hill et al., 2003).

CLONING

Oesch et al. (1985) isolated a cDNA clone corresponding to a pathogenic PrP fragment from a scrapie-infected hamster brain cDNA library. Southern blotting with PrP cDNA revealed a single gene with the same restriction patterns in normal and scrapie-infected brain DNA. A single PrP-related gene was also detected in murine and human DNA. Proteinase K digestion yielded PrP 27-30 in infected brain extract, but completely degraded the PrP-related protein in normal brain extract.

Kretzschmar et al. (1986) isolated a PRNP cDNA from a human retina cDNA library. The 253-amino acid protein shared 90% amino acid sequence identity with the hamster protein. Northern blot analysis detected a 2.5-kb mRNA in a variety of human neuroectodermal cell lines.

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

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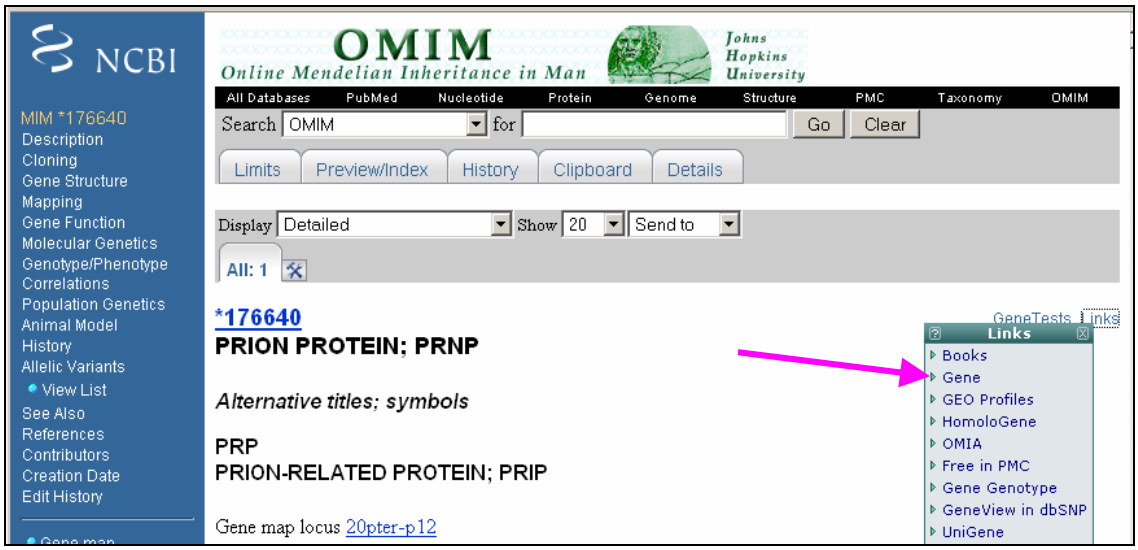
***176640**
PRION PROTEIN; PRNP

ALLELIC VARIANTS
(selected examples)

- **0001 CREUTZFELDT-JAKOB DISEASE [PRNP, EXTRA OCTAPEPTIDE CODING REPEATS]**
  GERSTMANN-STRAUSSLER DISEASE, INCLUDED
  HUNTINGTON DISEASE-LIKE 1, INCLUDED
- **0002 GERSTMANN-STRAUSSLER DISEASE [PRNP, PRO102LEU]**
- **0003 REMOVED FROM DATABASE**
- **0004 GERSTMANN-STRAUSSLER DISEASE [PRNP, ALA117VAL]**
- **0005 PRION DISEASE, SUSCEPTIBILITY TO [PRNP, MET129VAL] dbSNP**
  ALZHEIMER DISEASE, EARLY-ONSET, SUSCEPTIBILITY TO, INCLUDED
  APHASIA, PRIMARY PROGRESSIVE, SUSCEPTIBILITY TO, INCLUDED
- **0006 CREUTZFELDT-JAKOB DISEASE [PRNP, GLU200LYS] dbSNP**
  FATAL FAMILIAL INSOMNIA, INCLUDED
- **0007 CREUTZFELDT-JAKOB DISEASE [PRNP, ASP178ASN AND MET129VAL]**
- **0008 REMOVED FROM DATABASE**
- **0009 REMOVED FROM DATABASE**
- **0010 FATAL FAMILIAL INSOMNIA [PRNP, ASP178ASN AND MET129]**
- **0011 GERSTMANN-STRAUSSLER DISEASE [PRNP, PHE198SER]**
- **0012 GERSTMANN-STRAUSSLER DISEASE [PRNP, GLN217ARG]**
- **0013 REMOVED FROM DATABASE**
- **0014 CREUTZFELDT-JAKOB DISEASE [PRNP, VAL210ILE]**
- **0015 GERSTMANN-STRAUSSLER DISEASE [PRNP, PRO105LEU]**
- **0016 CREUTZFELDT-JAKOB DISEASE [PRNP, VAL180ILE]**
- **0017 CREUTZFELDT-JAKOB DISEASE [PRNP, MET232ARG]**
  DEMENTIA, LEWY BODY, INCLUDED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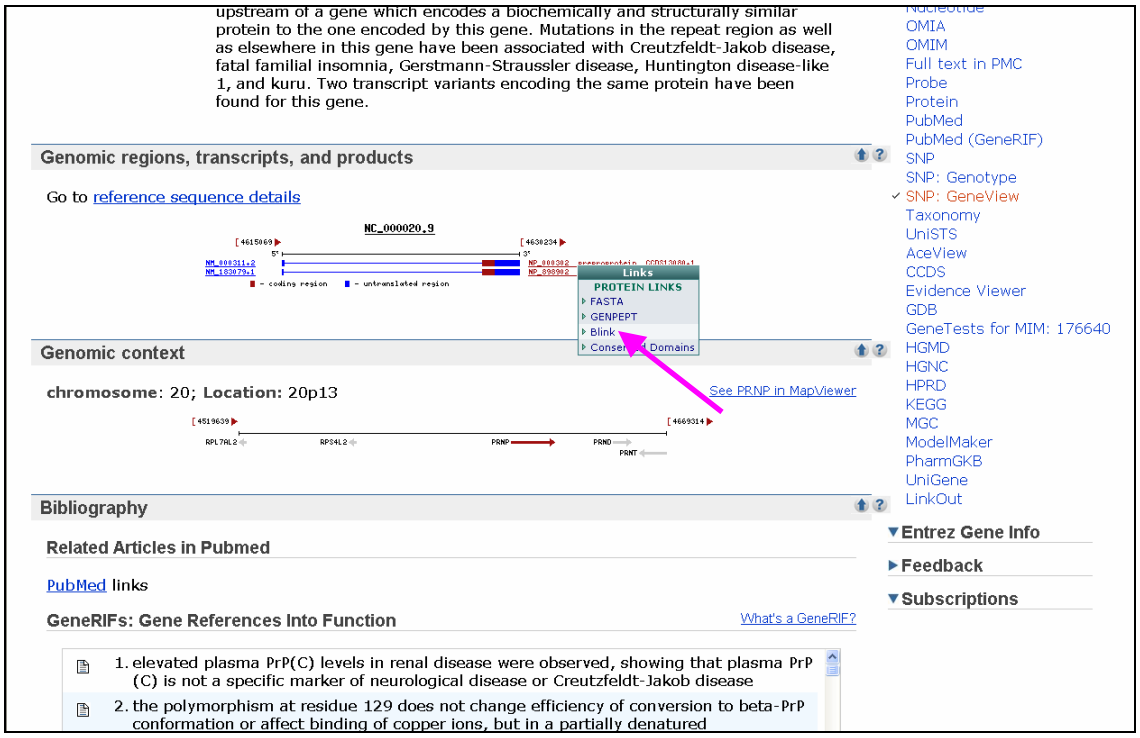
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chromosome: 20; Location: 20p13
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GeneRIFs: Gene References Into Function
What's a GeneRIF?
1. elevated plasma PrP(C) levels in renal disease were observed, showing that plasma PrP (C) is not a specific marker of neurological disease or Creutzfeldt-Jakob disease
2. the polymorphism at residue 129 does not change efficiency of conversion to beta-PrP conformation or affect binding of copper ions, but in a partially denatured
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Protein
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PubMed (GeneRIF)
SNP
SNP: Genotype
SNP: GeneView
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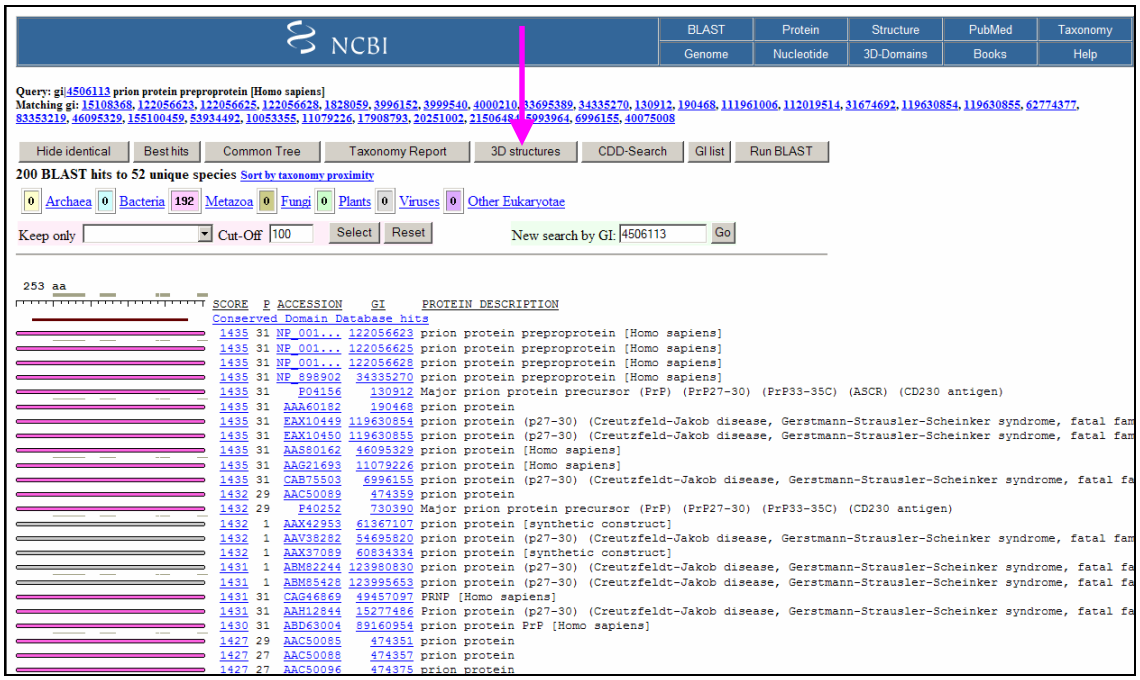
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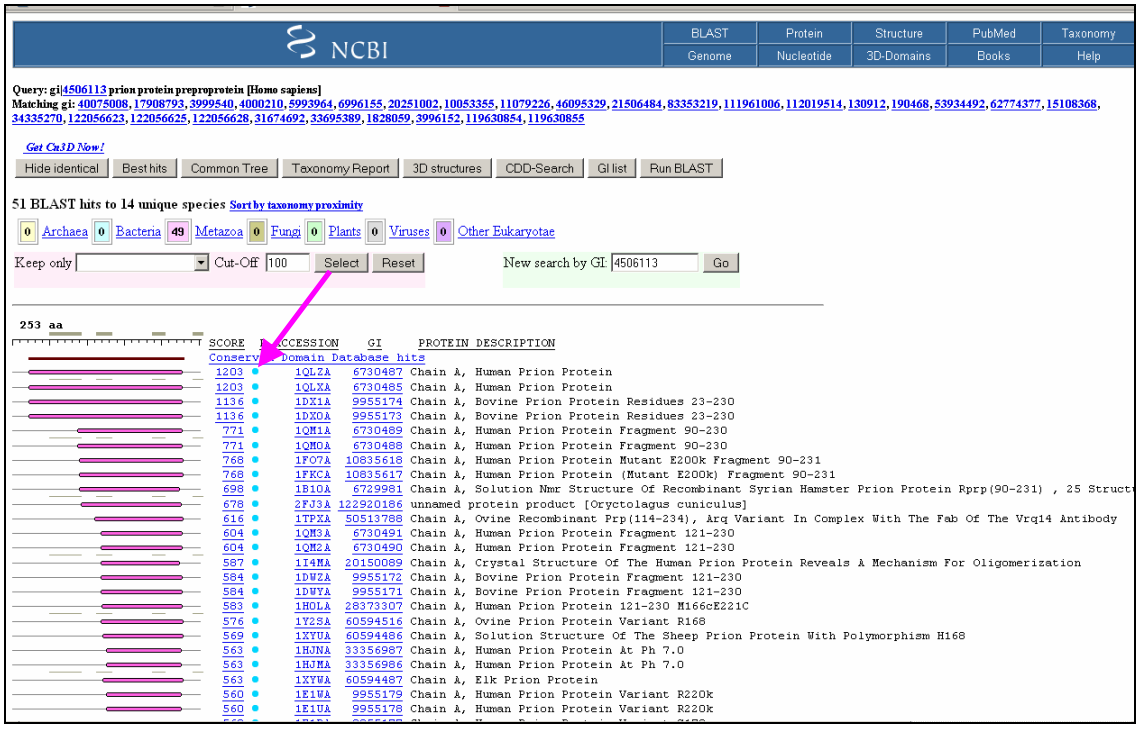
3D structures
CDD-Search
GI list
Run BLAST
200 BLAST hits to 52 unique species Sort by taxonomy proximity
Archaea
Bacteria
Metazoa
Fungi
Plants
Viruses
Other Eukaryotae
Keep only
Cut-Off
Select
Reset
New search by GI:
Go
253 aa
SCORE P ACCESSION GI PROTEIN DESCRIPTION
Conserved Domain Database hits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Run BLAST
51 BLAST hits to 14 unique species Sort by taxonomy proximity
Archaea
Bacteria
Metazoa
Fungi
Plants
Viruses
Other Eukaryotae
Keep only
Cut-Off
Select
Reset
New search by GI:
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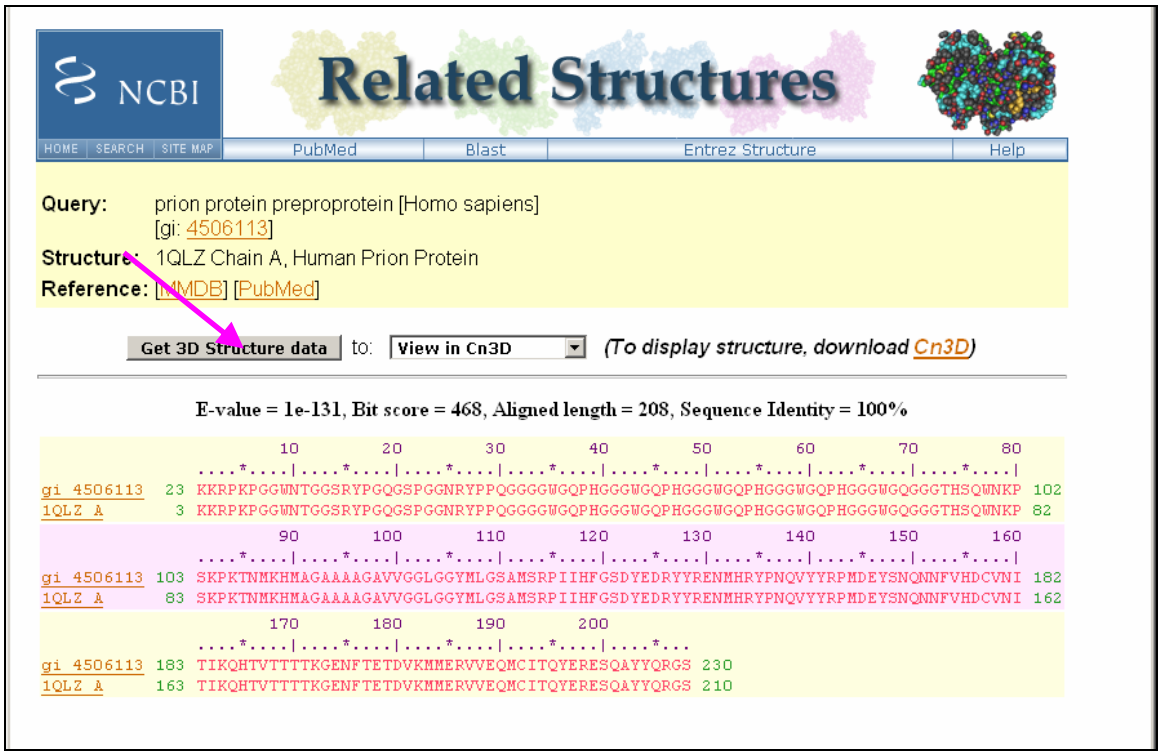
253 aa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Query: prion protein preproprotein [Homo sapiens]
[gi: 4506113]
Structure: 1QLZ Chain A, Human Prion Protein
Reference: [MMDB] [PubMed]
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E-value = 1e-131, Bit score = 468, Aligned length = 208, Sequence Identity = 100%
gi 4506113 23 KKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP 102
1QLZ A 3 KKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKP 82
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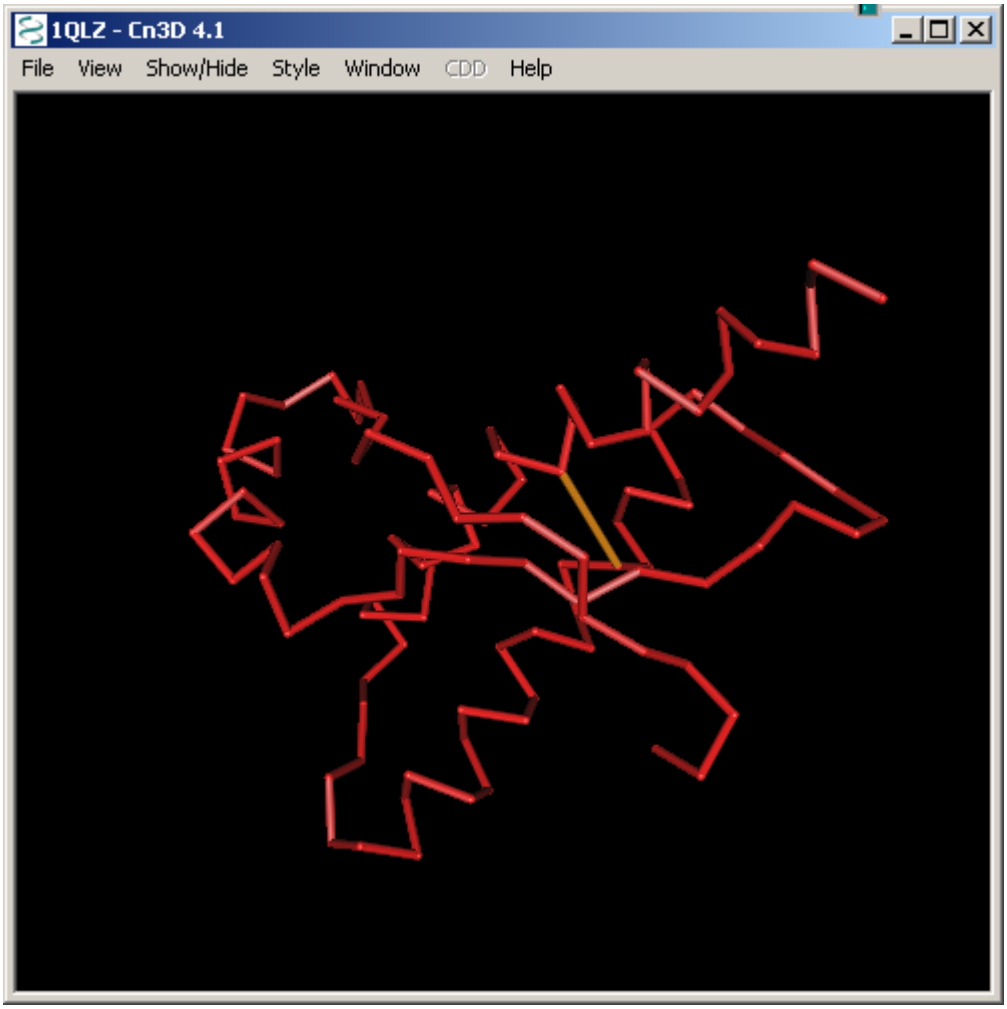
1QLZ A 83 SKPKTNMKHMAGAAAAGAVVGGLGGYMLGSAMSRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNI 162
gi 4506113 183 TIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYYQRGS 230
1QLZ A 163 TIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYYQRGS 210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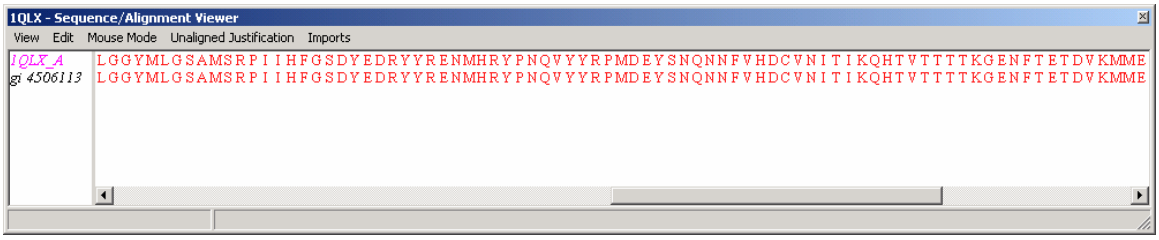
1QLX - Sequence/Alignment Viewer
View Edit Mouse Mode Unaligned Justification Imports
1QLX_A LGGYMLGSAMSRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMME
gi 4506113 LGGYMLGSAMSRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMME

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 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?