

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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Search Gene for prion protein Go Clear

Limits Preview/Index History Clipboard Details

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

News New "has ccds" property added. News archives...

Sample Searches

Find genes by... Search text

free text human muscular dystrophy

partial name and multiple species transporter[title] AND ("Drosophila melanogaster"[organism] OR "Mus musculus"[organism])

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

No history available

Add Term(s) to Query or View Index:

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

Organism 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] Go Clear Save Search

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Display Summary Show 20 Send to

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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
 GeneID: 3309

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All: Current only Gene names only SNP GeneView: 1

1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [*Homo sapiens*] updated 12-Aug-2007

GeneID: 5621

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; Homnidae; Homo

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

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

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- SNP: Genotype

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

NC_000020.9

Genomic context

chromosome: 20; Location: 20p13

[See PRNP in MapViewer](#)

SNP: Genotype

SNP: GeneView

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Gene Table

1: ASN.1

Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia [] updated 12-Aug-2007

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

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

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

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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [Homo sapiens] updated 12-Aug-2007

RefSeq status: Reviewed total gene size: 15166 bp

Genomic regions, transcripts, and products

Go to reference sequence details

NC_000020.9

461969 4639294

NM_000311.2 NM_183079.1 NP_000302.1 NP_898902.1

coding region untranslated region

| 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 | Coding EXON | INTRON | | | |
|-------------|-------------|-------------|----------|--------|--------|
| coords | length | coords | length | coords | length |
| 1-90 | 90 bp | 91-12728 | 12698 bp | | |
| 12729-15166 | 2378 bp | 12795-13565 | 762 bp | | |

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

NP_898902.1 length: 253 aa, number of exons: 1

| EXON | Coding EXON | INTRON | | | |
|-------------|-------------|-------------|----------|--------|--------|
| coords | length | coords | length | coords | length |
| 1-86 | 86 bp | 87-12788 | 12702 bp | | |
| 12789-15166 | 2378 bp | 12795-13565 | 762 bp | | |

NCBI Entrez Gene

Search Gene for [] Go Clear

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

updated 12-Aug-2007

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- UniGene
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EST NIH cDNA clone links

GSS Links

GSS NIH cDNA clone links

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 | length | Coding EXON | length | INTRON | length |
|------------|---------|-------------|--------|---------|---------|
| 1-90 | 90 bp | | | 91-1725 | 1268 bp |
| 1726-15166 | 2378 bp | 1729-11566 | 762 bp | | |

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

NP_898902.1 length: 253 aa, number of exons: 1

Genomic context

chromosome: 20; Location: 20p13

See PRNP in MapViewer

Entrez Gene Info

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Related Articles in PubMed

PubMed links

GeneRIFs: Gene References Into Function

What's a GeneRIF?

- Norwegian sisters with late onset Creutzfeldt-Jakob disease caused by the E200K mutation.
- Findings of polymorphism at codon 129 of PRNP gene implicated in the development of CJD in Greek population. Met/Val allele frequencies and genotype distribution examined in 348 individuals. Genotypes Met/Met 50%, Met/Val 39% and Val/Val 11% were observed.
- Increased plasma PIP(C) reflects an endogenous increase in expression in acute stroke-affected brain tissue.
- review focuses on transfusion-transmission of variant Creutzfeldt-Jakob disease by red cell preparations
- Combined molecular, biochemical, and single living polarized cell imaging characterizations suggest that hPIP(C) is selectively targeted to the apical side of Madin-Darby canine kidney (MDCKII) and of intestinal epithelia (Caco2) cells.
- Oxidative stress might be an influence that leads to substantial structural conversions of PrP in vivo.
- prion protein does not require other Bcl-2 family proteins to protect against Bax-mediated cell death
- This study suggested that polymorphism at position -101 in the regulatory region of PRNP may be a risk factor for sCJD among codon 129 heterozygotes.
- Equilibrium binding and kinetics of FRET show that the PRNP binding to the oligonucleotides and their bending occur simultaneously.
- analysis of experimentally derived constraints for high-resolution structural models of PrP amyloid fibrils

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

| Product | Interactant | Other Gene | Complex | Source | Pubs |
|--|-------------|------------|---------|--------|--------|
| NP_000302.1 | NP_001155.1 | APBB1 | | HPRD | PubMed |
| NP_000302.1 | NP_052759.2 | CLSTN1 | | HPRD | PubMed |
| NP_000302.1 | NP_001822.2 | CLU | | HPRD | PubMed |
| NP_000302.1 | NP_001834.2 | CNTN1 | | HPRD | PubMed |
| PrPc interacts with CSNK2A1 (CK2 alpha). This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2A1 (CK2 alpha). | | | | | |
| NP_000302.1 | NP_001886.1 | CSNK2A1 | | BIND | PubMed |
| PrPc interacts with CSNK2A2 (CK2 alpha prime). This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2A2 (CK2 alpha prime). | | | | | |
| NP_000302.1 | NP_001887.1 | CSNK2A2 | | BIND | PubMed |
| PrPc interacts with CSNK2B (CK2 beta) albeit weakly. This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2B (CK2 beta). | | | | | |
| NP_000302.1 | NP_001311.3 | CSNK2B | | BIND | PubMed |
| NP_000302.1 | NP_004399.1 | DNM1 | | HPRD | PubMed |
| NP_000302.1 | NP_570629.1 | DPP6 | | HPRD | PubMed |
| NP_000302.1 | NP_002077.1 | GRB2 | | HPRD | PubMed |
| PrPc interacts with HSPA5 (BiP). | | | | | |
| NP_000302.1 | NP_005338.1 | HSPA5 | | BIND | PubMed |
| 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 | PubMed |
| NP_000302.1 | NP_000416.1 | L1CAM | | HPRD | PubMed |

General gene information

Markers

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

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

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

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

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

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

Phenotypes

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 | 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 |

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)

- 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 | PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Strausler syndrome and bovine spongiform encephalopathy |
| Location:23-230 | |
| Blast Scores:546 | |
- 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 | PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Strausler syndrome and bovine spongiform encephalopathy |
| Location:23-230 | |
| Blast Scores:546 | |
- 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, B1669189, DA297032, M13899](#)

Conserved Domains (1): [summary](#)

| | |
|----------------------------|--|
| smart00157 | PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Strausler syndrome and bovine spongiform encephalopathy |
| Location:23-230 | |
| Blast Scores:546 | |
- 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, D8461478, M13899](#)

Conserved Domains (1): [summary](#)

| | |
|----------------------------|--|
| smart00157 | PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Strausler syndrome and bovine spongiform encephalopathy |
| Location:23-230 | |
| Blast Scores:546 | |
- 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.

RefSeqs of Annotated Genomes: Build 36.2

The following sections contain reference sequences that belong to a specific genome build: [Explain](#)

Reference assembly

Genomic

- NC_000020.9 Reference assembly**

Range: 4615069..4630234

Download: [GenBank](#) [FASTA](#)
- NT_011387.8**

Range: 4607069..4622234

Download: [GenBank](#) [FASTA](#)

Alternate assembly (based on Celera assembly)

Genomic

- AC_000063.1 Alternate assembly (based on Celera assembly)**

Range: 4736784..4751948

Download: [GenBank](#) [FASTA](#)
- 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 DQ894502.2 | ABM85428.1 |
| Genomic M81929.1 | AAB59442.1 |
| Genomic M81930.1 | AAB59443.1 |
| Genomic S71208.1 | AAB20521.1 |

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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [[Entrez Gene Home](#)]

Homo sapiens [[Table Of Contents](#)]

GeneID: 5621 updated 12-Aug-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*

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Genomic regions, transcripts, and products [[↑ ?](#)]

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Display Alignment Scores Show 20 Send to

All: 1 Fungi: 0 Mammals: 0

1: HomoloGene:7904. Gene conserved in Amniota

| | | |
|---------------|------|---|
| 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 | Pmp | prion protein |
| R.norvegicus | Pmp | prion protein |
| G.gallus | PRNP | prion protein (p27-30) |

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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 BLAST 2.2.16 [Mar-25-2007]

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|24335270|ref|NP_898902.1|](#)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|6996155|...](#)
 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 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|7419279755|.....](#)
 Length = 254 (1 .. 254)

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

NCBI Entrez Gene

Search: Gene for [] Go Clear

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1: PRNP prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia) [*Homo sapiens*]
 GeneID: 5621 updated 12-Aug-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: ENSG00000171067; HPRD:01453; MIM:176640

Gene type protein coding

RefSeq status Reviewed

Organism *Homo sapiens*

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

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

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

Genomic regions, transcripts, and products

Go to [reference sequence details](#)

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NCBI Single Nucleotide Polymorphism

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP

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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 | plus strand | NP_000302 | forward | NT_011387 | reference | <- currently shown |
| NM_000311 | plus strand | NP_000302 | forward | NW_927317 | Celera | View snp on GeneModel |
| NM_183079 | plus strand | NP_898902 | forward | NT_011387 | reference | View snp on GeneModel |
| NM_183079 | plus strand | NP_898902 | forward | NW_927317 | Celera | View snp on GeneModel |

in gene region cSNP has frequency double hit haplotype tagged refresh

| gene model | Contig Label | Contig | mRNA | protein | mRNA orientation | transcript | snp count |
|---------------------------|--------------|-----------|-----------|-----------|------------------|-------------|------------|
| (contig mRNA transcript): | reference | NT_011387 | NM_000311 | NP_000302 | forward | plus strand | 22, coding |

| Region | Contig position | mRNA pos | dbSNP rs# | Heterozygosity | 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. | | | | nonsynonymous | A | Thr [T] | 1 | 26 |
| | | | | N.D. | | | | contig reference | C | Pro [P] | 1 | 26 |
| | | 4619949 | 183 | rs11538762 | N.D. | | | nonsynonymous | A | His [H] | 2 | 28 |
| | | | | N.D. | | | | contig reference | C | Pro [P] | 2 | 28 |
| | | 4619975 | 209 | rs11538763 | N.D. | | | synonymous | A | Arg [R] | 1 | 37 |
| | | | | N.D. | | | contig reference | C | Arg [R] | 1 | 37 | |

| | | | | | | | | | | |
|---------|-----|----------------------------|-------|--|---------------------|------------------|---|---------|---|-----|
| 4620378 | 612 | rs16990018 | 0.046 | | Yes | nonsynonymous | G | Ser [S] | 2 | 171 |
| | | | 0.046 | | Yes | contig reference | A | Asn [N] | 2 | 171 |
| 4620399 | 633 | rs11538766 | N.D. | | Yes | nonsynonymous | T | Val [V] | 2 | 178 |
| | | | N.D. | | Yes | contig reference | A | Asp [D] | 2 | 178 |
| 4620405 | 639 | rs11538767 | N.D. | | Yes | nonsynonymous | C | Ala [A] | 2 | 180 |
| | | | N.D. | | Yes | contig reference | T | Val [V] | 2 | 180 |
| 4620464 | 698 | rs28933385 | N.D. | | Yes | nonsynonymous | A | Lys [K] | 1 | 200 |
| | | | N.D. | | Yes | contig reference | G | Glu [E] | 1 | 200 |
| 4620521 | 755 | rs1800014 | N.D. | | Yes | nonsynonymous | A | Lys [K] | 1 | 219 |
| | | | N.D. | | Yes | contig reference | G | Glu [E] | 1 | 219 |
| 4620538 | 772 | rs6052773 | N.D. | | Yes | synonymous | T | Ala [A] | 3 | 224 |
| | | | N.D. | | Yes | contig reference | C | Ala [A] | 3 | 224 |
| 4620545 | 779 | rs17852079 | N.D. | | Yes | nonsynonymous | A | Lys [K] | 1 | 227 |
| | | | N.D. | | Yes | contig reference | C | Gln [Q] | 1 | 227 |

Online Mendelian Inheritance in Man

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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-Strausler 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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Nomenclature

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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]
DEMENCIA, LEWY BODY, INCLUDED

NCBI

MIM *176640
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***176640**
PRION PROTEIN; PRNP

Alternative titles; symbols

PRP
PRION-RELATED PROTEIN; PRIP

Gene map locus [20pter-p12](#)

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- ▶ 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-Strausler disease, Huntington disease-like 1, and kuru. Two transcript variants encoding the same protein have been found for this gene.

- Nucleotide
- OMIA
- OMIM
- Full text in PMC
- Probe
- Protein
- PubMed
- PubMed (GeneRIF)
- SNP
- SNP: Genotype
- SNP: GeneView
- Taxonomy
- UniSTS
- AceView
- CCDS
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- HGMD
- HGNC
- KEGG
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Genomic regions, transcripts, and products

Go to [reference sequence details](#)



- Links
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- 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

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

Query: gi:4506113 prion protein preproprotein [Homo sapiens]

Matching gi: 40075008, 17908793, 3999540, 4000210, 5993964, 6996155, 20251002, 10053355, 11079226, 21506484, 83353219, 46095329, 111961006, 112019514, 130912, 190468, 53934492, 62774377, 15108368, 34335270, 122056623, 122056625, 122056628, 31674692, 33695389, 1828059, 3996152, 11963085, 11963085

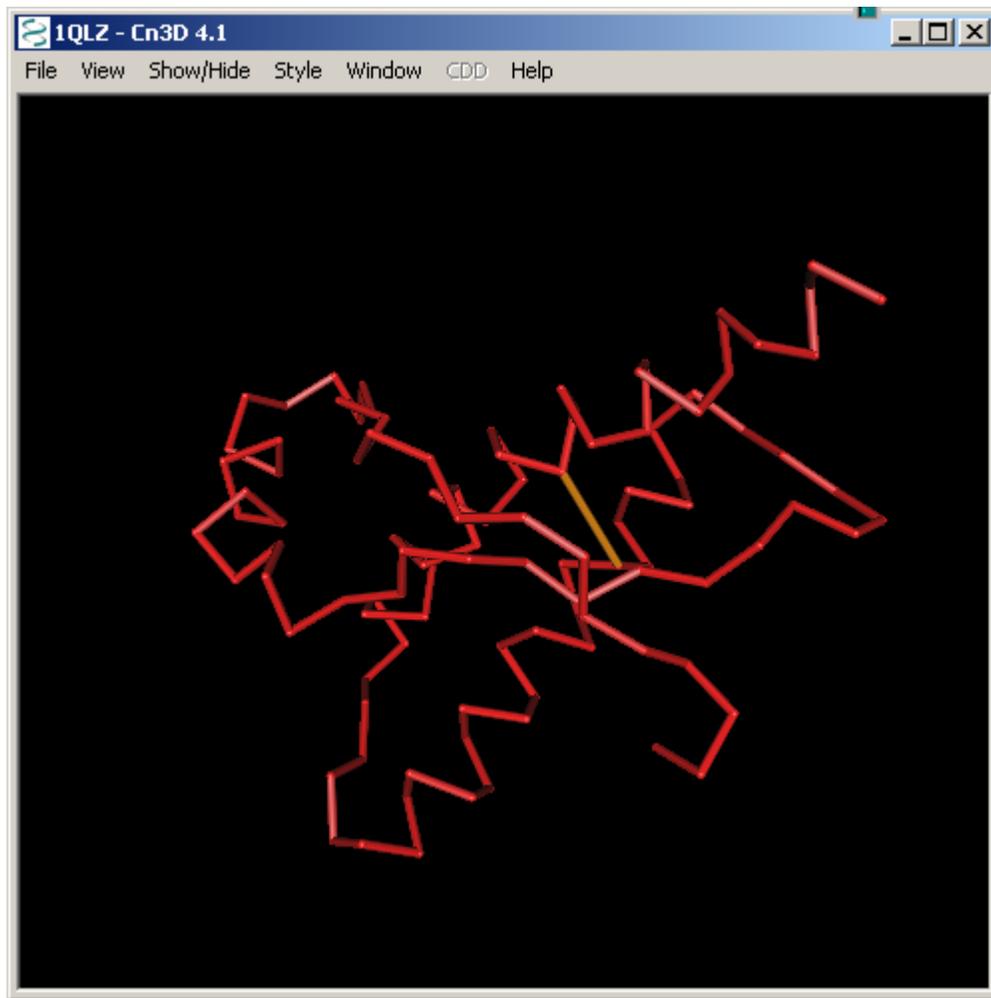
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0 Archaea | 0 Bacteria | 192 Metazoa | 0 Fungi | 0 Plants | 0 Viruses | 0 Other Eukaryotes

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

| SCORE | E | ACCESSION | GI | PROTEIN DESCRIPTION |
|---------------------------------------|----|-----------|-----------|---|
| Conserved Domain Database hits | | | | |
| 1435 | 31 | CAB75503 | 6996155 | prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa |
| 1435 | 31 | AAG21693 | 11079226 | prion protein [Homo sapiens] |
| 1435 | 31 | AAS80162 | 46095329 | prion protein [Homo sapiens] |
| 1435 | 31 | P04156 | 130912 | Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (ASCR) (CD230 antigen) |
| 1435 | 31 | AAR60182 | 190468 | prion protein |
| 1435 | 31 | NP_898902 | 34335270 | prion protein preproprotein [Homo sapiens] |
| 1435 | 31 | NP_001... | 122056623 | prion protein preproprotein [Homo sapiens] |
| 1435 | 31 | NP_001... | 122056625 | prion protein preproprotein [Homo sapiens] |
| 1435 | 31 | NP_001... | 122056628 | prion protein preproprotein [Homo sapiens] |
| 1435 | 31 | FXN10449 | 11963085 | prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa |
| 1435 | 31 | FXN10450 | 11963085 | prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa |
| 1432 | 29 | AAC50089 | 474359 | prion protein |
| 1432 | 29 | P40252 | 730390 | Major prion protein precursor (PrP) (PrP27-30) (PrP33-35C) (CD230 antigen) |
| 1432 | 1 | AAK42953 | 61367107 | prion protein [synthetic construct] |
| 1432 | 1 | AAV38282 | 54695820 | prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa |
| 1432 | 1 | AAK37089 | 60834334 | prion protein [synthetic construct] |
| 1431 | 1 | ABM82244 | 123980830 | prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa |
| 1431 | 1 | ABM85428 | 123995653 | prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa |
| 1431 | 31 | CAG46869 | 49457097 | PRNP [Homo sapiens] |
| 1431 | 31 | AAN12944 | 15277466 | Prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal fa |



1QLX - Sequence/Alignment Viewer

View Edit Mouse Mode Unaligned Justification Imports

| | |
|------------|---|
| 1QLX_A | LGGYMLGSAMSRPIIHFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNIITIKQHTVTTTTKGENFTETDVKMME |
| gi 4506113 | LGGYMLGSAMSRPIIHFGSDYEDRYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNIITIKQHTVTTTTKGENFTETDVKMME |

Problem 2

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

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

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

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

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