

| Project | Nucleotide | Protein | Genome | Structure | PopSet | Taxonomy | OMIM |
|--|------------|----------------|-------------|-----------|--------|----------|------|
| | | | | Go | Clear | | |
| Genome sequencing projects statistics | | | | | | | |
| Organism | Complete | Draft assembly | In progress | total | | | |
| Prokaryotes | 597 | 397 | 492 | 1486 | | | |
| Archaea | 47 | 3 | 30 | 80 | | | |
| Bacteria | 550 | 394 | 462 | 1406 | | | |
| Eukaryotes | 23 | 131 | 184 | 338 | | | |
| Animals | 4 | 54 | 89 | 147 | | | |
| Mammals | 2 | 22 | 25 | 49 | | | |
| Birds | | 1 | 2 | 3 | | | |
| Fishes | | 3 | 6 | 9 | | | |
| Insects | 1 | 19 | 20 | 40 | | | |
| Flatworms | | 1 | 3 | 4 | | | |
| Roundworms | 1 | 3 | 13 | 17 | | | |
| Amphibians | | | 2 | 2 | | | |
| Reptiles | | | 2 | 2 | | | |
| Other animals | | 6 | 19 | 25 | | | |
| Plants | 3 | 3 | 34 | 40 | | | |
| Land plants | 2 | 2 | 27 | 31 | | | |
| Green Algae | 1 | 1 | 7 | 9 | | | |
| Fungi | 10 | 53 | 30 | 93 | | | |
| Ascomycetes | 8 | 46 | 21 | 75 | | | |
| Basidiomycetes | 1 | 5 | 5 | 11 | | | |
| Other fungi | 1 | 2 | 4 | 7 | | | |
| Protists | 6 | 19 | 27 | 52 | | | |
| Apicomplexans | 1 | 10 | 6 | 17 | | | |
| Kinetoplasts | 1 | 2 | 6 | 9 | | | |
| Other protists | 4 | 7 | 14 | 25 | | | |
| total: | 620 | 528 | 676 | 1824 | | | |

Summary pages

| | |
|--------------------|-------------|
| Entrez Gene | 4683 |
| HGED Symbol Report | NBS1 |
| KEGG gene | 4683 |
| euGenes | HUgn0004683 |
| CGAP | 25812 |
| SOURCE | 4683 |

Genomic resources

| | | | |
|----------------------|---|---|---|
| Genomic sequences | NCBI EBI DBP AB013139.1 | NCBI EBI DBP AF069291.1 | |
| GDB | 9598211 | | |
| GenAtlas | NBS1 | | |
| UCSC Genome Browser | chr8:91014739-91066075 | | |
| Vista Genome Browser | Align sequence to [17 other genomes] | | |
| AceView | NBS1 | | |
| dbSNP | rs14448 rs769420 rs3026268 rs3358 | rs769414 rs1063054 rs3087624 rs16786 | rs769416 rs2735383 rs3780123 More... |

Transcripts

| | | | |
|----------------|---|---|---|
| RefSeq | NM_002485 | | |
| cDNA sequences | NCBI EBI DBP AF051334 | NCBI EBI DBP AF058696 | NCBI EBI DBP AK001017 |
| TIGR | NCBI EBI DBP BC005293 | NCBI EBI DBP BC040519 | NCBI EBI DBP BX640816 |
| | THC16941 | | |

Protein sequences

| | |
|---------|-------------------|
| TrEMBL | O60934 |
| GenPept | AAC39732 AAC39752 |

Protein structure and domains

| | | | |
|------------------|-----------|-----------|-----------|
| InterPro domains | IPR001357 | IPR000253 | IPR008984 |
| Pfam | PF00533 | PF00498 | |
| ProDom | IPR001357 | IPR000253 | IPR008984 |
| PROSITE | PS50006 | | |

Protein function and disease links

| | |
|------|---------------|
| OMIM | 602667 251260 |
|------|---------------|

Networks and pathways

| | |
|---------|------|
| PubGene | NBS1 |
|---------|------|

Literature Links

| | |
|--------|--------------------|
| PubMed | 24 PubMed articles |
|--------|--------------------|

UCSC Genome Bioinformatics

[Genomes](#) - [Blat](#) - [Tables](#) - [Gene Sorter](#) - [PCR](#) - [VisiGene](#) - [Proteome](#) - [FAQ](#) - [Help](#)

[Genome
Browser](#)

[ENCODE](#)

[Blat](#)

[Table
Browser](#)

[Gene Sorter](#)

[In Silico PCR](#)

[VisiGene](#)

[Proteome
Browser](#)

About the UCSC Genome Bioinformatics Site

This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides a portal to the ENCODE project.

We encourage you to explore these sequences with our tools. The Genome Browser zooms and scrolls over chromosomes, showing the work of annotators worldwide. The Gene Sorter shows expression, homology and other information on groups of genes that can be related in many ways. Blat quickly maps your sequence to the genome. The Table Browser provides convenient access to the underlying database. VisiGene lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns.

News

[News Archives ►](#)

To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the [genome-announce](#) mailing list.

Human (*Homo sapiens*) Genome Browser Gateway

The UCSC Genome Browser was created by the [Genome Bioinformatics Group of UC Santa Cruz](#).
Software Copyright (c) The Regents of the University of California. All rights reserved.

| clade | genome | assembly | position or search term | image width | |
|------------|--------|----------|-------------------------|-------------|---------------------------------------|
| Vertebrate | Human | May 2004 | sickle cell | 620 | <input type="button" value="submit"/> |

[Click here to reset](#) the browser user interface settings to their defaults.

[manage custom tracks](#)

[configure tracks and display](#)

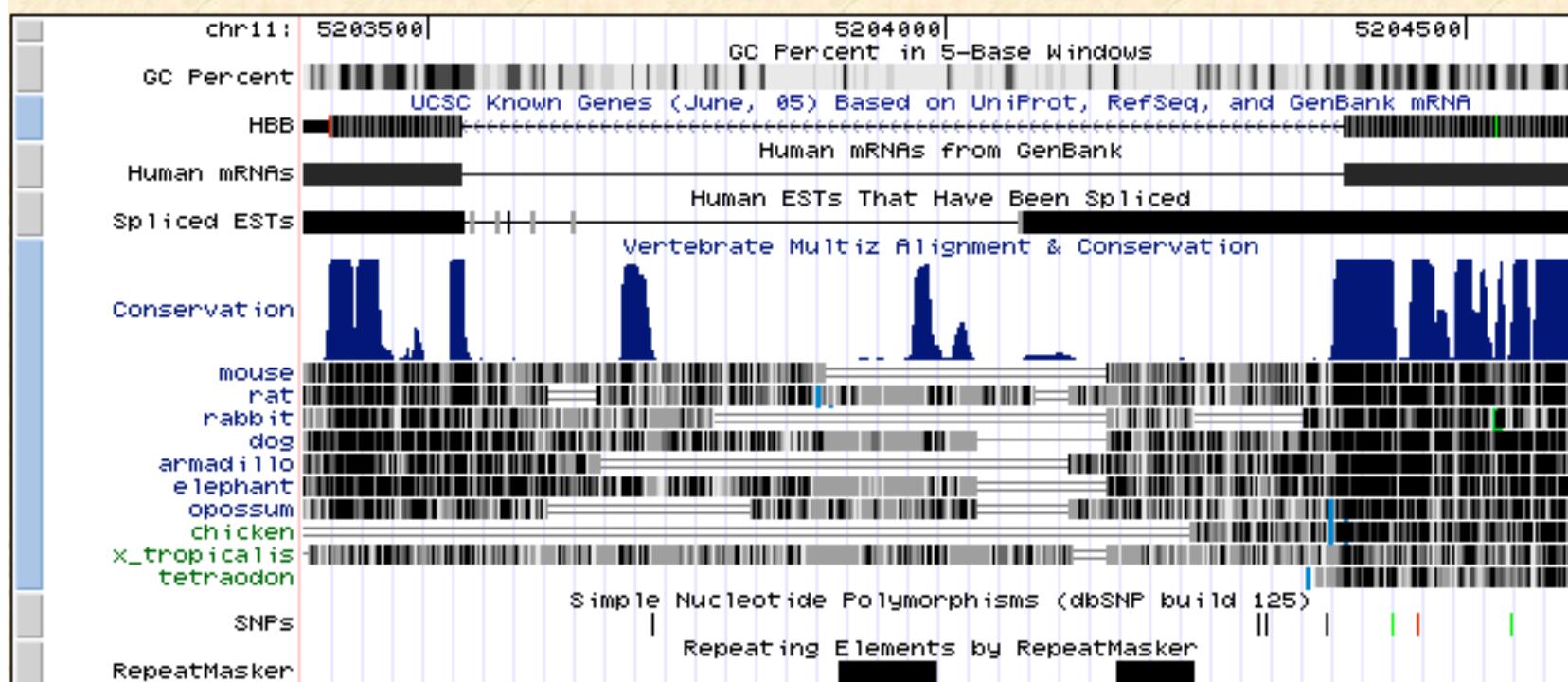
[clear position](#)

Sample position queries

A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, or a cytological band, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the [User's Guide](#) for more information.

| Request: | Genome Browser Response: |
|---------------------|--|
| chr7 | Displays all of chromosome 7 |
| 20p13 | Displays region for band p13 on chr 20 |
| chr3:1-1000000 | Displays first million bases of chr 3, counting from p arm telomere |
| chr3:1000000+2000 | Displays a region of chr3 that spans 2000 bases, starting with position 1000000 |
| D16S3046 | Displays region around STS marker D16S3046 from the Genethon/Marshfield maps. Includes 100,000 bases on each side as well. |
| RH18061;RH80175 | Displays region between STS markers RH18061;RH80175. Includes 100,000 bases on each side as well. This syntax may also be used for other range queries, such as between cytobands and uniquely-determined ESTs, mRNAs, refSeqs, etc. |
| AA205474 | Displays region of EST with GenBank accession AA205474 in BRCA1 cancer gene on chr 17 |
| AC008101 | Displays region of clone with GenBank accession AC008101 |
| AF083811 | Displays region of mRNA with GenBank accession number AF083811 |
| PRNP | Displays region of genome with HUGO Gene Nomenclature Committee identifier PRNP |
| NM_017414 | Displays the region of genome with RefSeq identifier NM_017414 |
| NP_059110 | Displays the region of genome with protein accession number NP_059110 |
| pseudogene | Lists transcribed pseudogenes, but not cDNAs |
| mRNA | Lists mRNAs for caudal homeobox genes |
| homeobox caudal | Lists many zinc finger mRNAs |
| zinc finger | Lists only kruppel-like zinc fingers |
| kruppel zinc finger | Lists candidate genes associated with Huntington's disease |
| huntington | Lists mRNAs deposited by scientist named Zahler |
| zahler | Lists mRNAs deposited by co-author J.E. Evans |
| Evans,J.E. | |

position/search chr11:5,203,380-5,204,607 [jump](#) [clear](#) size 1,228 bp. [configure](#)



[default tracks](#)[hide all](#)[manage custom tracks](#)[configure](#)[refresh](#)

Use drop down controls below and press refresh to alter tracks displayed.

Tracks with lots of items will automatically be displayed in more compact modes.

Custom Tracks

[Base Position](#)[newSnp](#)[full](#)[dense](#)

Mapping and Sequencing Tracks

[Chromosome Band](#)[STS Markers](#)[FISH Clones](#)[Recomb Rate](#)[Map Contigs](#)[hide](#)[hide](#)[hide](#)[hide](#)[hide](#)[Assembly](#)[Gap](#)[Coverage](#)[BAC End Pairs](#)[Fosmid End Pairs](#)[hide](#)[hide](#)[hide](#)[hide](#)[hide](#)[GC Percent](#)[WSSD](#)[Short Match](#)[Restr Enzymes](#)[Blat Sequence](#)[hide](#)[Duplication](#)[hide](#)[hide](#)[hide](#)

Phenotype and Disease Associations

[RGD QTL](#)[Locus Variants](#)[hide](#)[hide](#)

Genes and Gene Prediction Tracks

[Known Genes](#)[CCDS](#)[RefSeq Genes](#)[Other RefSeq](#)[MGC Genes](#)[pack](#)[hide](#)[pack](#)[hide](#)[hide](#)[hide](#)[Vega](#)[Ensembl Genes](#)[AccView Genes](#)[ECgene Genes](#)[dense](#)[Pseudogenes](#)[full](#)[hide](#)[hide](#)[squish](#)[hide](#)[hide](#)[hide](#)[hide](#)[pack](#)[SGP Genes](#)[Geneid Genes](#)[Genscan Genes](#)[Exoniphy](#)[full](#)[hide](#)[hide](#)[hide](#)[hide](#)

Display mode:

Filter: red green blue exclude include **Combination Logic:** and or

accession:

BE256422

author:

library:

tissue:

cell:

keyword:

gene:

product:

description:

Color track by bases:

position/search chrX:100,081,640-100,081,670

jump

clear

size 31 bp.

configure

chrX (q22.1) p22.2 21.1 12 Xq21.1 Xq23 24 Xq25 Xq28

chrX: 100081645] 100081650] 100081655] 100081660] 100081665]

CXorf34 SNP

UCSC Known Genes (June, 05) Based on UniProt, RefSeq, and GenBank mRNA

| | | | | | | | | | | | | |
|----------------|---|---|---|---|---|---|---|---|---|---|---|--|
| AL832849 | R | A | D | E | V | A | Q | E | L | L | L | |
| BC007526 | R | A | D | E | V | A | Q | E | L | L | L | |
| CXorf34 | R | A | D | E | V | A | Q | E | L | L | L | |

Human ESTs That Have Been Spliced

BE256422 C Vertebrate Multiz Alignment & Conservation

Conservation

Gaps

| | | | | | | | | | | | | | | | | | | | | | | | | | |
|--------------|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| human | A | G | A | A | C | G | T | A | G | G | A | G | T | G | A | C | G | R | G | A | T | T | T | A | |
| chimp | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | G | A | . | . | C | G |
| rhesus | . | . | . | . | . | . | . | . | . | . | A | . | . | . | . | . | . | . | . | G | G | . | . | C | G |
| mouse | G | . | . | C | . | . | . | . | . | . | A | . | . | . | . | . | . | . | . | G | G | . | . | C | G |
| rat | G | . | . | C | . | . | . | . | . | A | . | . | . | . | . | . | . | . | G | G | . | . | C | G | |
| rabbit | G | . | . | C | . | . | . | . | . | A | . | . | . | . | . | . | . | . | G | G | . | . | C | G | |
| dog | G | . | . | C | . | . | . | . | . | A | C | . | G | . | . | . | . | A | . | G | . | . | G | | |
| cow | G | . | C | C | . | . | . | . | . | A | C | . | G | . | . | . | . | A | . | G | . | . | G | | |
| armadillo | G | . | T | C | . | . | . | . | . | A | . | . | T | . | - | - | - | - | A | . | G | . | . | G | |
| elephant | G | . | C | . | . | . | . | . | . | A | . | . | G | . | . | . | . | A | . | G | . | . | G | | |
| tenrec | G | . | T | . | . | . | . | . | . | A | . | C | G | T | A | . | . | A | . | G | T | . | C | G | |
| opossum | . | A | . | T | . | . | . | . | . | A | . | C | . | . | A | . | G | . | A | . | G | T | A | C | |
| chicken | G | . | C | . | . | . | . | . | . | A | . | A | . | . | A | . | A | . | C | A | . | G | T | A | |
| x_tropicalis | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = | = |
| zebrafish | G | A | . | C | . | . | C | . | . | A | . | C | . | . | A | . | G | . | C | A | . | G | T | C | G |
| tetraodon | . | A | . | C | . | . | C | . | . | A | . | C | . | . | A | . | G | . | C | C | . | C | C | G | |
| fugu | . | A | . | C | . | . | C | . | . | A | . | C | . | . | A | . | G | . | C | C | . | C | C | G | |

Simple Nucleotide Polymorphisms (dbSNP build 125)
Duplications of >1000 Bases of Non-RepeatMasked Sequence

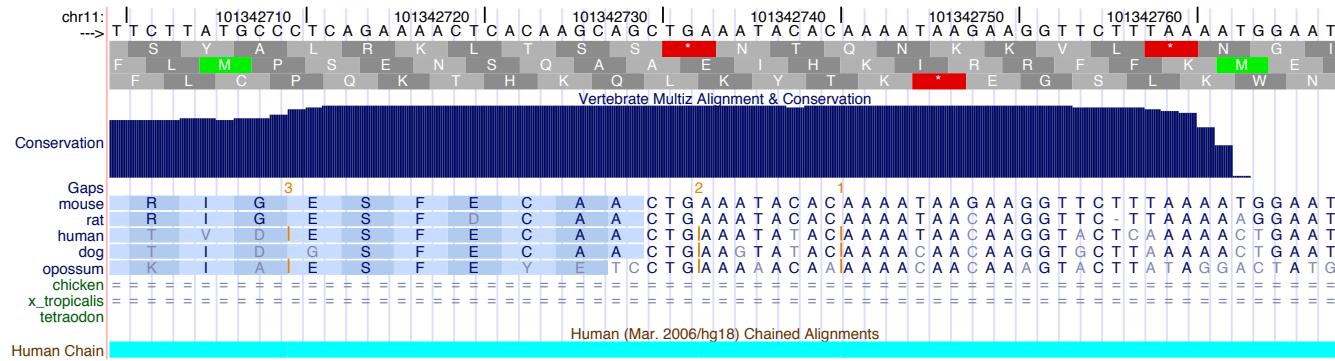
Structural Variation

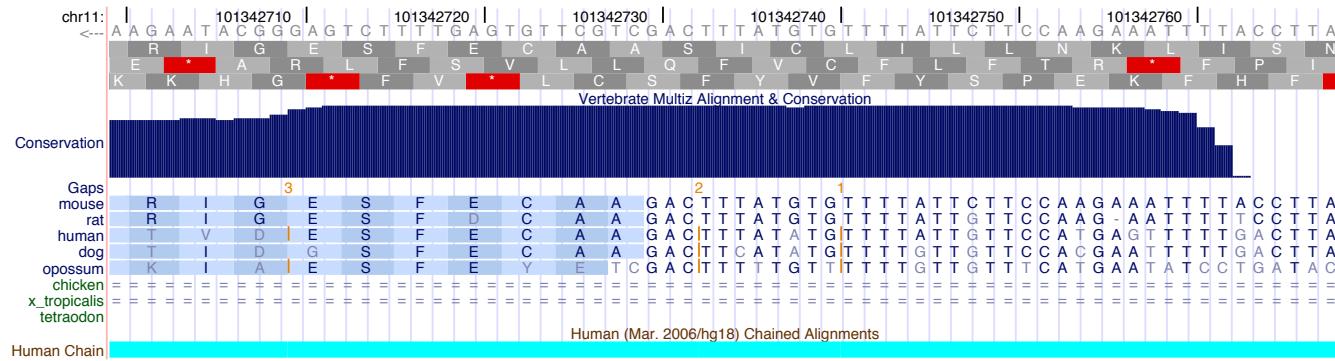
Segmental Dups

Sharp CNPs
Iafrate CNPs
Sebat CNPs
Tuzun Fossids
McCarroll Dels
Conrad Dels
Hinds Dels

Repeating Elements by RepeatMasker

SINE
LINE
LTR





Human Gene HBB Description and Page Index

Description: beta globin

Alternate Gene Symbols: AF117710, AF181989, AF349114, AY136510, AY509193, BC007075, CR536530, M25113, V00497

CCDS: [CCDS7753.1](#)

Representative Refseq: [NM_000518](#) **Protein:** [P68871](#) (aka HBB_HUMAN)

RefSeq Summary: The alpha (HBA) and beta (HBB) loci determine the structure of the 2 types of polypeptide chains in adult hemoglobin, Hb A. The normal adult hemoglobin tetramer consists of two alpha chains and two beta chains. Mutant beta globin causes sickle cell anemia. Absence of beta chain causes beta-zero-thalassemia. Reduced amounts of detectable beta globin causes beta-plus-thalassemia. The order of the genes in the beta-globin cluster is 5'-epsilon -- gamma-G -- gamma-A -- delta -- beta--3'. Publication Note: This RefSeq record includes a subset of the publications that are available for this gene. Please see the Entrez Gene record to access additional publications.

Position: chr11:5203272-5204877

Strand: -

Genomic Size: 1606

Exon Count: 3

| Page Index | Quick Links | UniProt Comments | Sequence | Microarray | RNA Structure |
|-------------------|---------------|------------------|-------------------|------------|---------------|
| Protein Structure | Other Species | GO Annotations | mRNA Descriptions | Pathways | Methods |

Quick Links to Tools and Databases

| | | | | | |
|-----------------|----------------|------------------|--------------|-----------|-------------------|
| Gene Sorter | Genome Browser | Proteome Browser | Table Schema | VisiGene | Allen Brain Atlas |
| CGAP | Ensembl | Entrez Gene | ExonPrimer | GeneCards | GeneLynx |
| H-INV | HGNC | HPRD | Jackson Labs | OMIM | PubMed |
| Stanford SOURCE | UniProt | Gepis Tissue | | | |

Comments and Description Text from UniProt (Swiss-Prot/TrEMBL)

ID: [HBB_HUMAN](#)

DESCRIPTION: Hemoglobin beta subunit (Hemoglobin beta chain) (Beta-globin).

FUNCTION: Involved in oxygen transport from the lung to the various peripheral tissues.

SUBUNIT: Heterotetramer of two alpha chains and two beta chains in adult hemoglobin A (HbA).

TISSUE SPECIFICITY: Red blood cells.

171 images match



Mouse Hoxa9



Mouse Hoxa10



Mouse Hoxa10



Mouse Hoxa11



source: [Mahoney Lab](#) source: [MGI](#) Reference: [Mouse Brain Organization Revealed Through Direct Genome-Scale TF Expression Analysis](#).

Year: 2004 Contributors: Gray P.A.,Fu H.,Luo P.,Zhao Q.,Yu J.,Ferrari A.,Tenzen T.,Yuk D.I.,Tsung E.F.,Cai Z.,Alberta J.A.,Cheng L.P.,Liu Y.,Stenman J.M.,Valerius M.T.,Billings N.,Kim H.A.,Greenberg M.E.,McMahon A.P.,Rowitch D.H.,Stiles C.D.,Ma Q..

Gene: [Hoxa9](#) Probe: [RNA from primers](#) GenBank: [AB005458](#)

Organism: Mus musculus Sex: n/a Strain: C57BL Genotype: wild type

Stage: 10.5 day old embryo (Theiler 17) Body Part: whole

Expression: central nervous system(0.17) Section Type: whole mount

Acknowledgements: Thanks to Paul Gray for transferring the images.

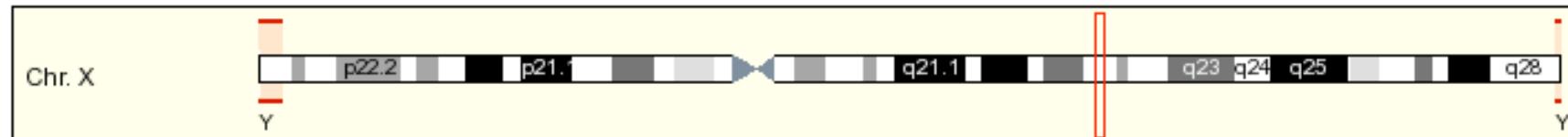
Home UCSC Human Gene Sorter

genome Human assembly May 2004 search CXorf34 Go!

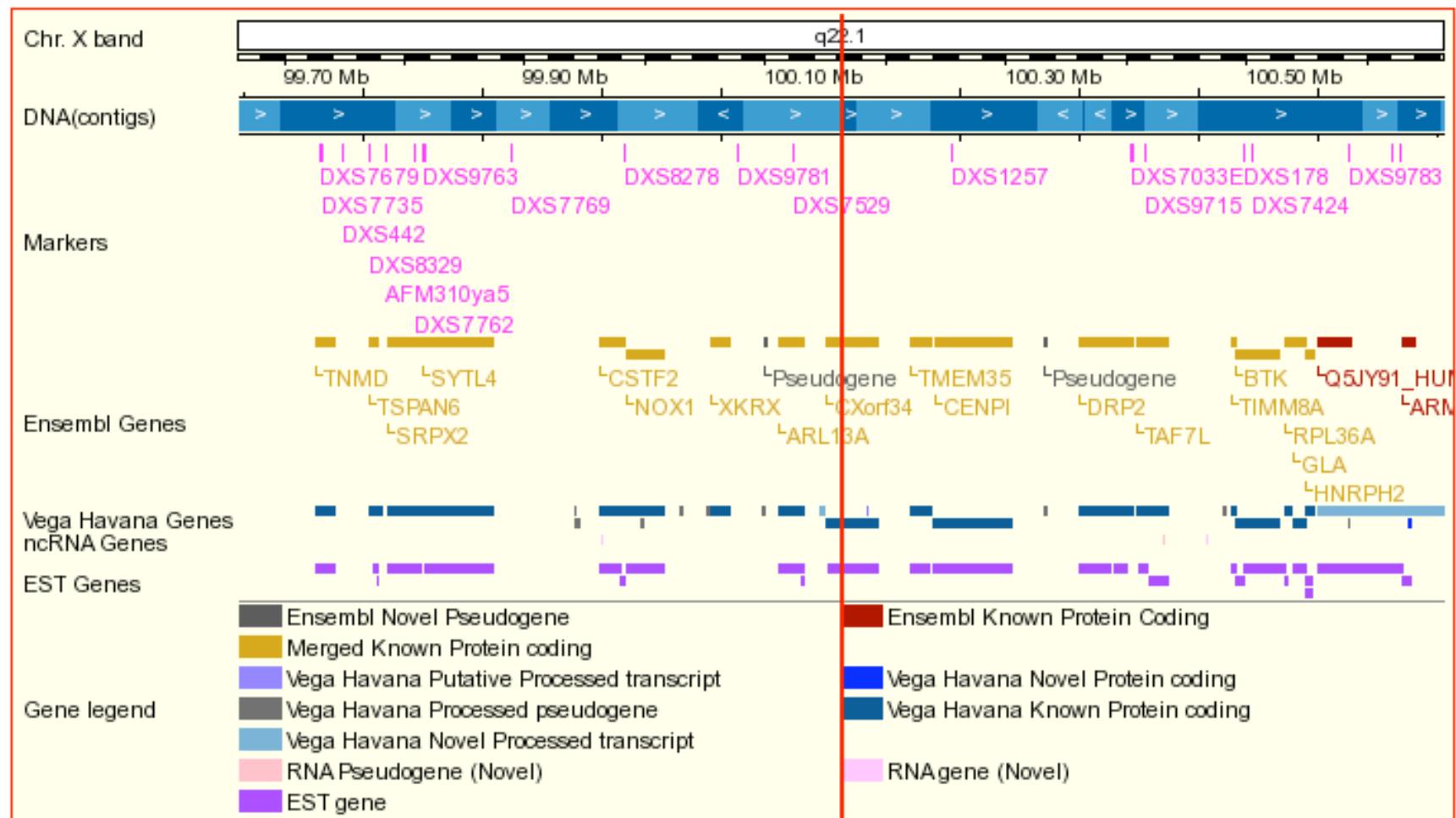
sort by Protein Homology - BLASTP configure filter (now off) display 50 output sequence text

| # | Name | fetal brain | whole brain | amygdala | thymus | bone marrow | PB-CD4+ Tcells | skin | pancreatic islets | adipocyte | heart | lung | kidney | liver | ovary | testis | BLASTP E-Value | Genome Position | Description |
|---|----------|-------------|-------------|----------|--------|-------------|----------------|------|-------------------|-----------|-------|------|--------|------------------|--|--------|----------------|-----------------|-------------|
| 1 | CXorf34 | | | | | | | | | | | | 0 | chrX 100,091,954 | hypothetical protein LOC79979 | | | | |
| 2 | CR592601 | | | | | | | | | | | | 1e-75 | chr22 18,476,857 | HpaII tiny fragments locus 9c protein. | | | | |

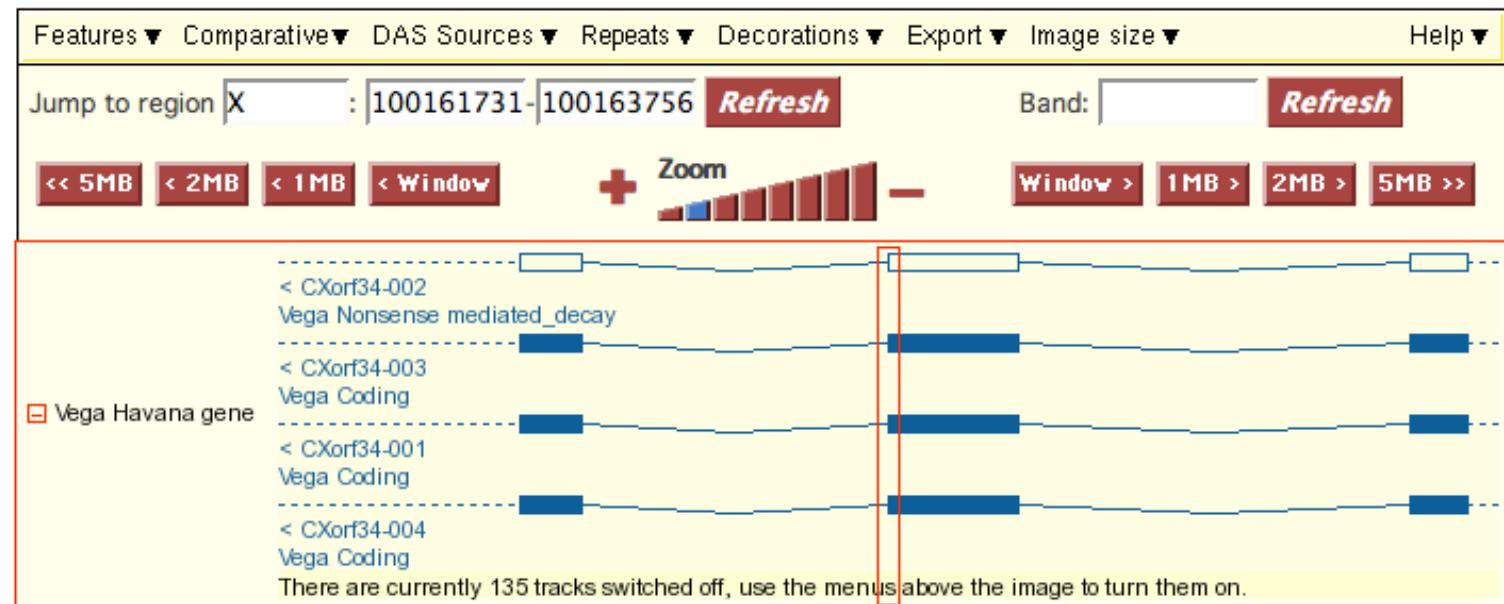
Chromosome X



Overview

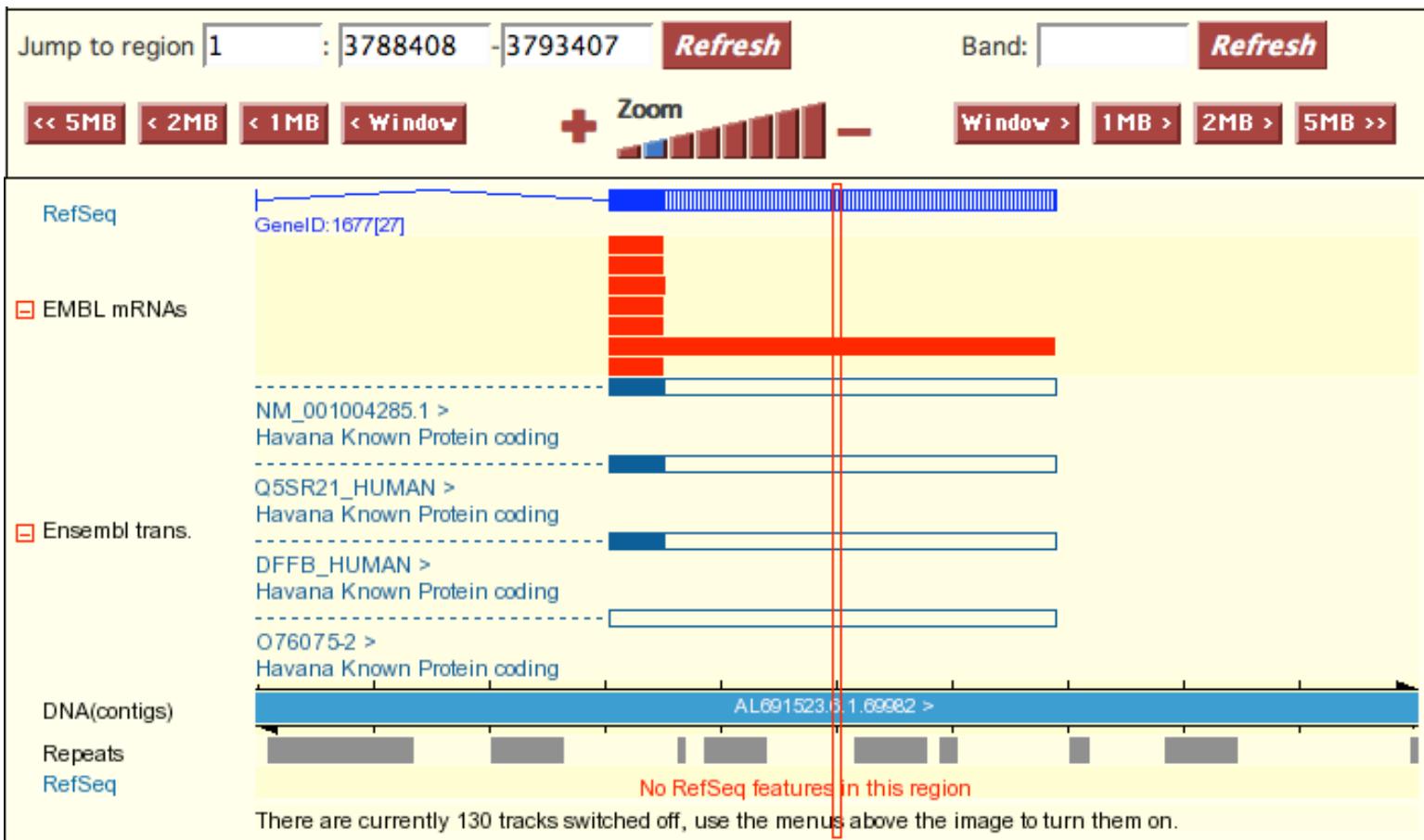


Detailed view

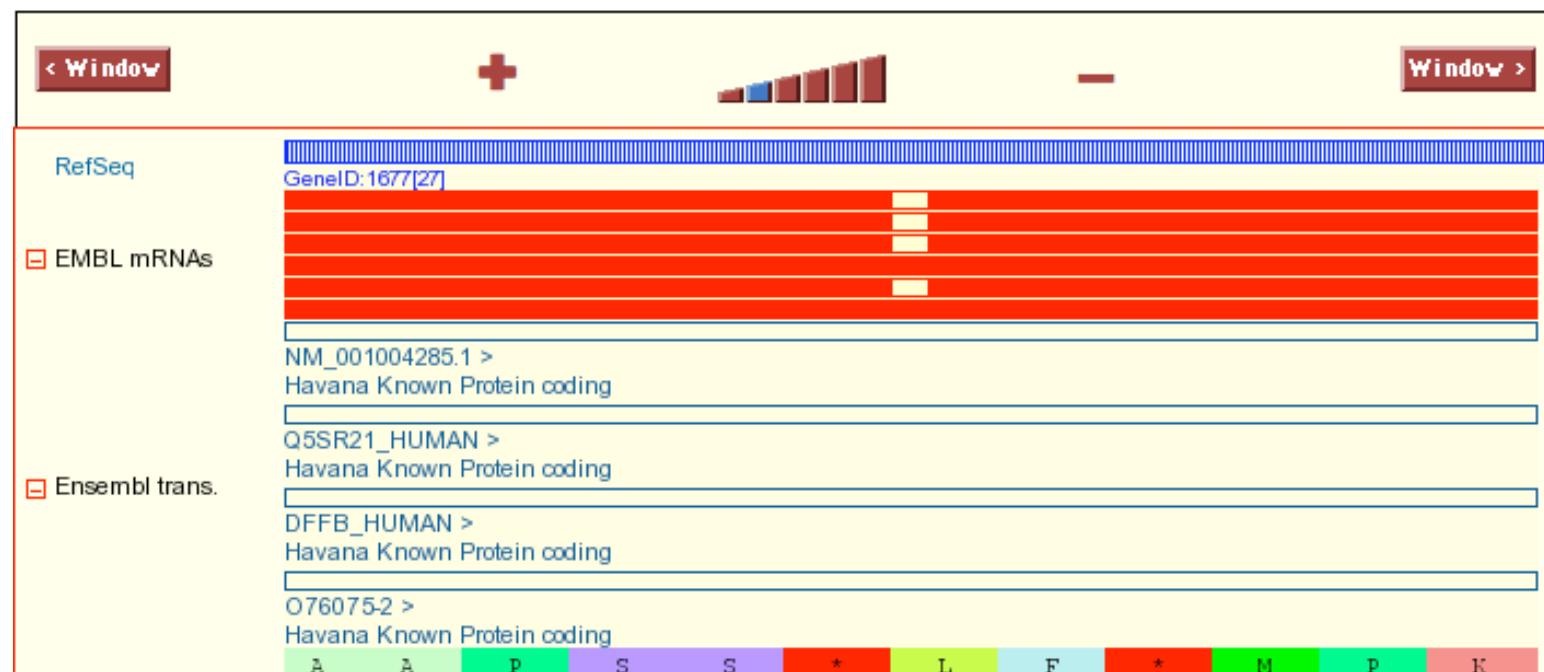


Basepair view





Basepair view



Jump to region X : 100161153-100163178

Refresh

Band:

Refresh

<< 5MB < 2MB < 1MB < Window

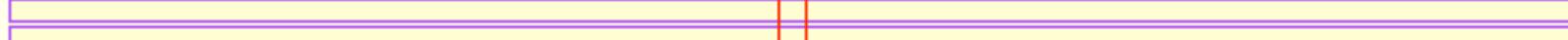


Window > 1MB > 2MB > 5MB >>

OMIM Morbid map

No OMIM Morbid map features in this region

□ EST (ex.)

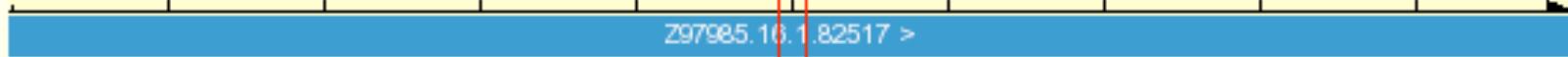


□ EMBL mRNAs



DNA(contigs)

Z97985.16.1.82517 >



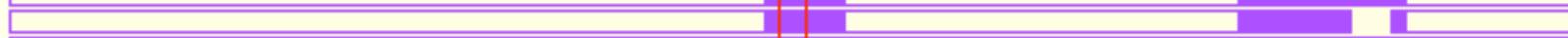
□ Human RefSeqs



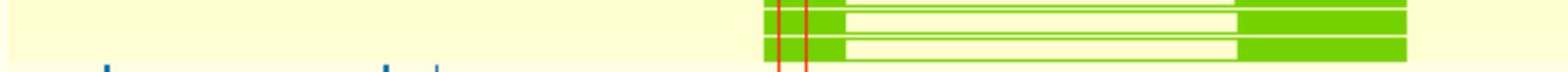
□ EMBL mRNAs



□ EST (ex.)



□ Human cDNAs



SNPs

Repeats

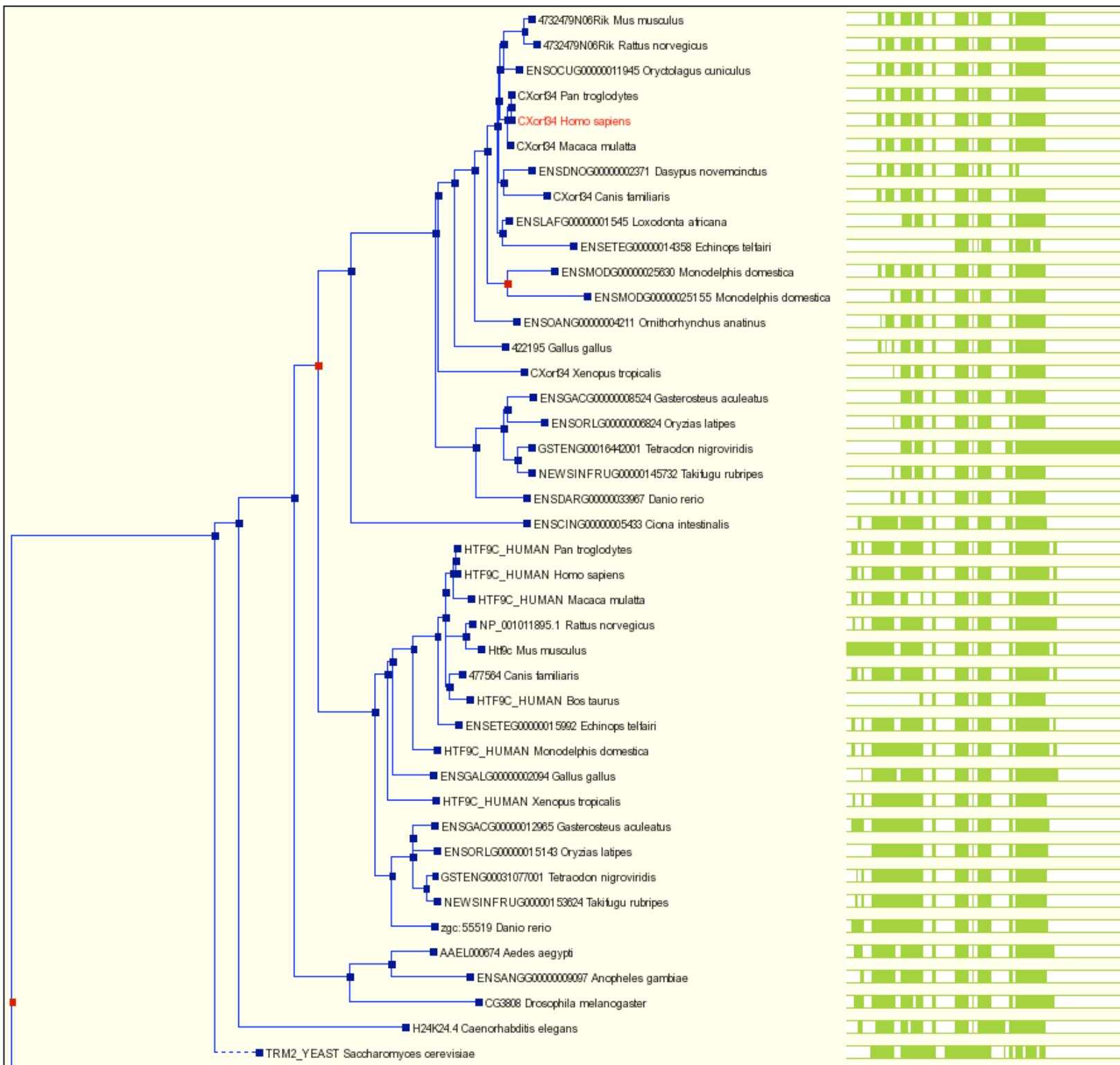
CXorf34snp

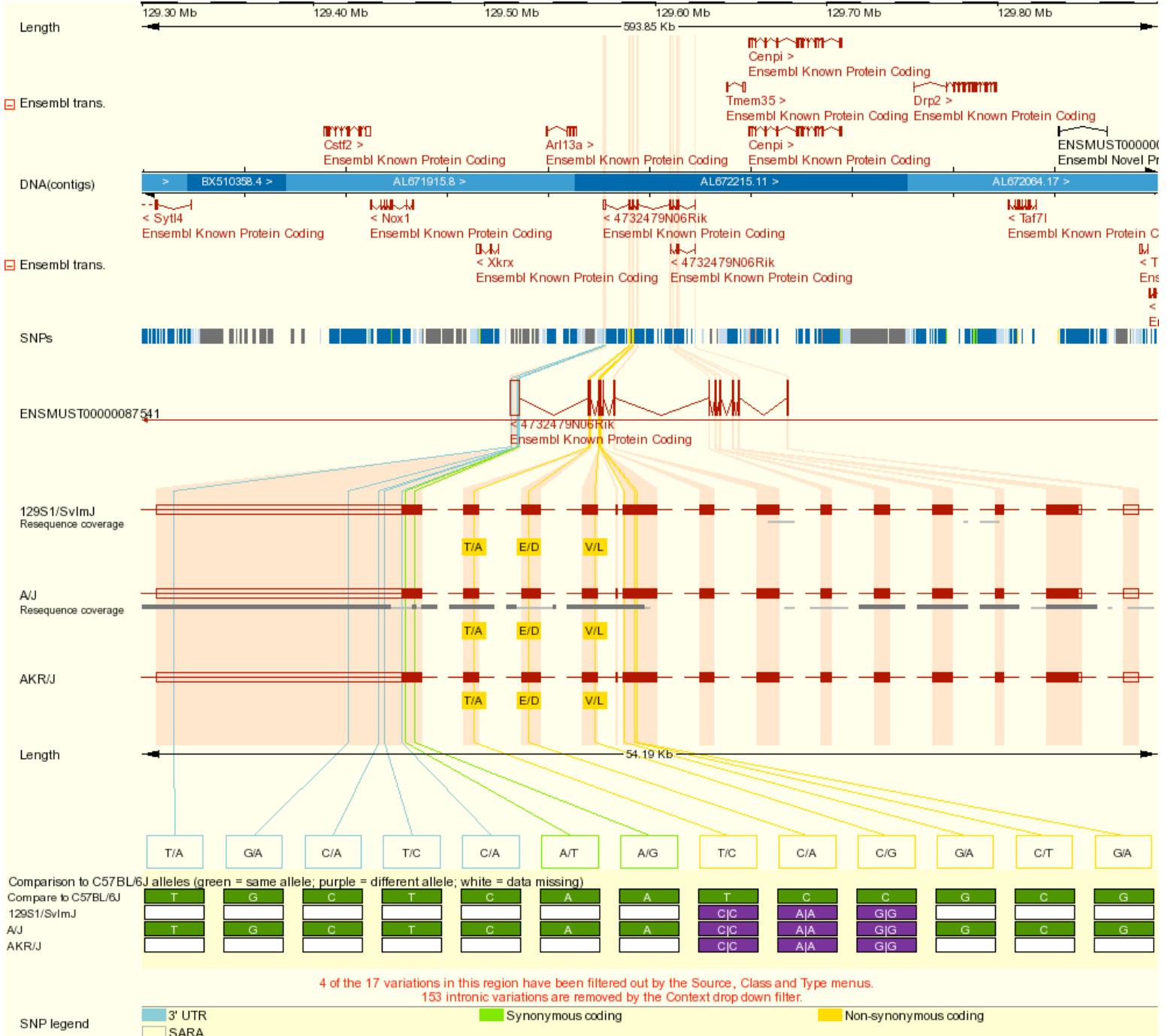
OMIM Morbid map

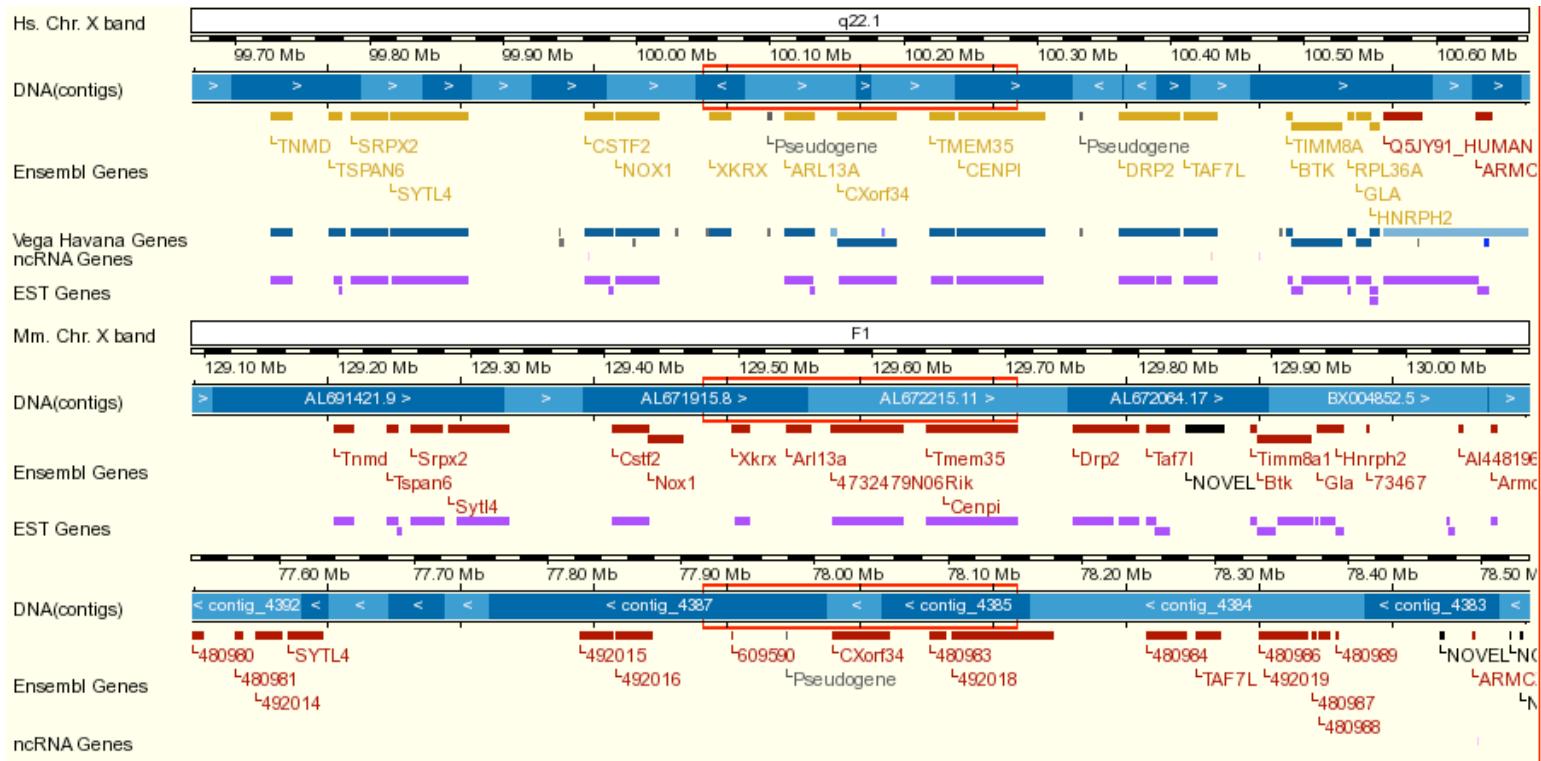
No OMIM Morbid map features in this region

SegDup WashU

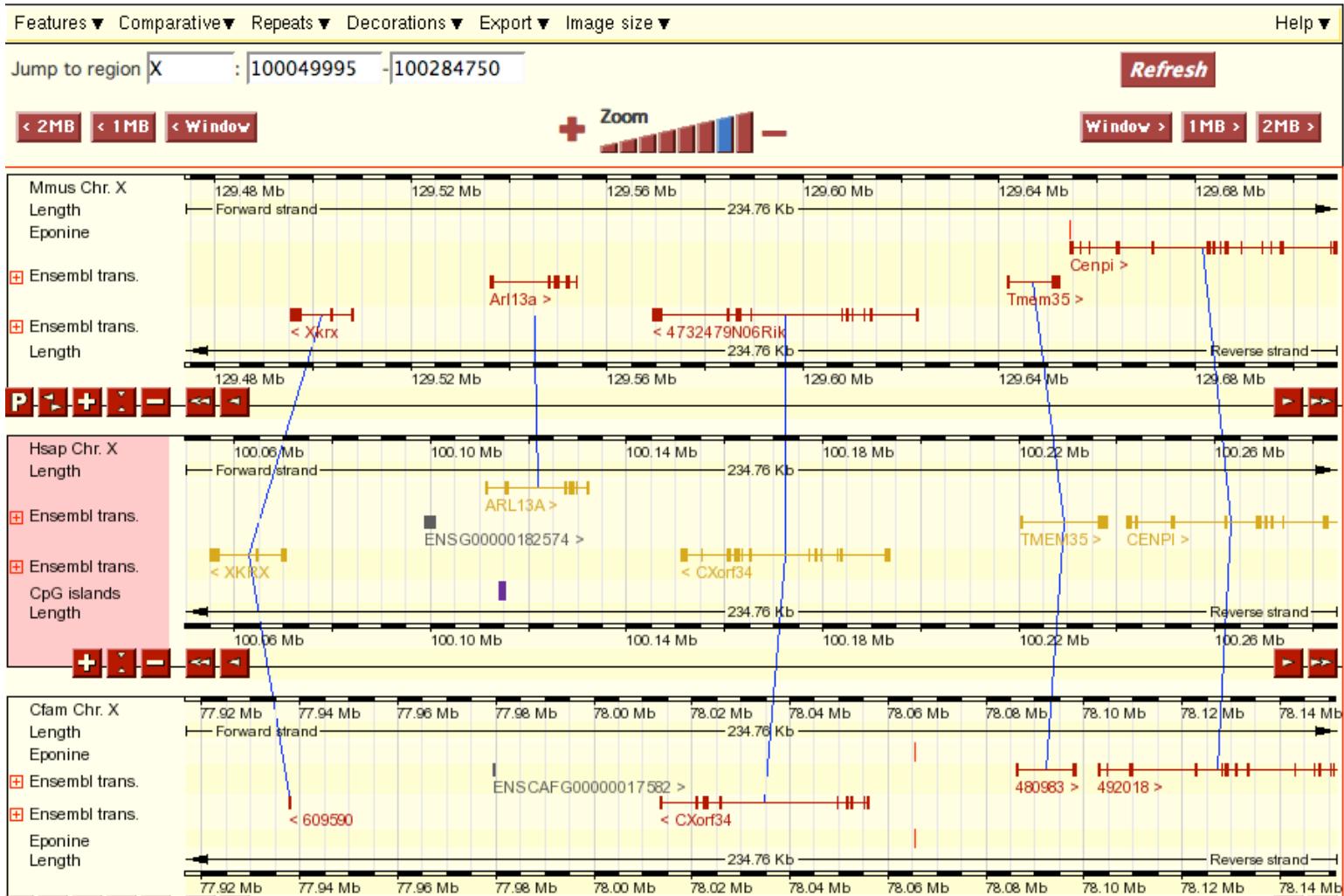
No SegDup WashU features in this region







Ailed View



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Help & Documentation

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- [What's New](#)
- [About Ensembl](#)
- [Downloading data](#)
- [Displaying your own data](#)
- [Ensembl software](#)

Select a species

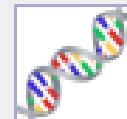
- [Mammals](#)

Search Ensembl

Search: for

e.g. mouse chromosome 2 or X:10000..20000 or human gene BRCA2

Ensembl tools



[Start a sequence search](#) →
Search Ensembl for nucleotide
and peptide sequences with
BLAST and SSAHA.



[Mine Ensembl with
BioMart](#) →
Cross-reference Ensembl
datasets with BioMart, a
powerful data-mining tool.

Ensembl 42

Preliminary species

Popular genomes



Homo sapiens

NCBI 36 | Vega



Mus musculus

NCBI mm36 | Vega



Danio rerio

Enter the Query Sequence

Either Paste sequences (max 30 sequences) in FASTA or plain text:

```
GTCCTTGGGATTGAATTGTTGGAGCAGGCAC TGGAGGATGCAAGATGGACTGCAGCCTT
```

Or Upload a file containing one or more FASTA sequences

[Browse...](#)

Or Enter a sequence ID or accession (EMBL, UniProt, RefSeq)

[Retrieve](#)

Or Enter an existing ticket ID:

[Retrieve](#)

- dna queries
- peptide queries

Select the databases to search against

Select species:

Use 'ctrl' key to select multiple species

Gallus_gallus
Gasterosteus_aculeatus
Homo_sapiens

- dna database
- peptide database

Genomic sequence

Known Consensus CDS Peptides (CCDS peptides)

Select the Search Tool

BLASTN

SSAHA2

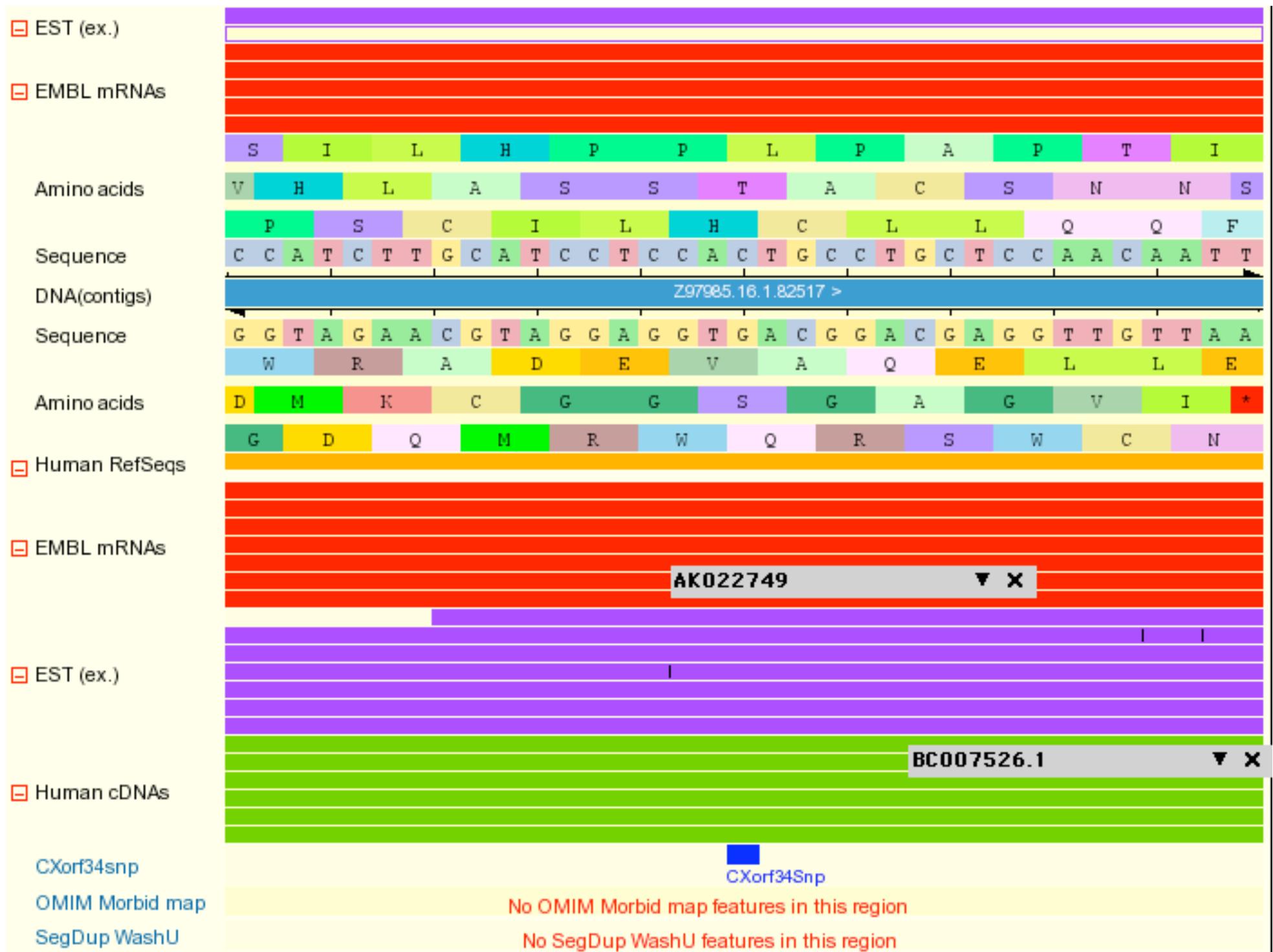
TBLASTX

[configure▶](#)

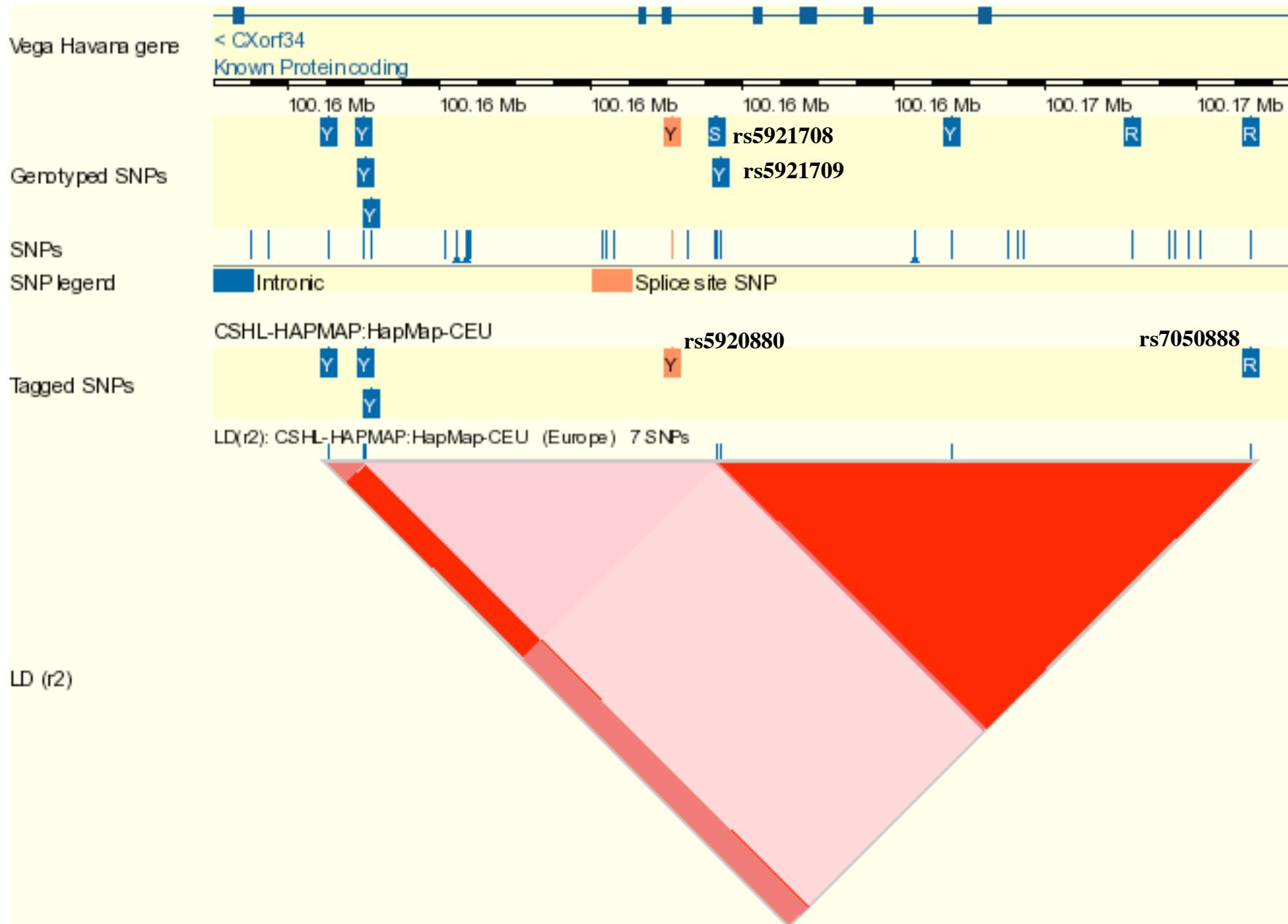
RUN▶

Search sensitivity:

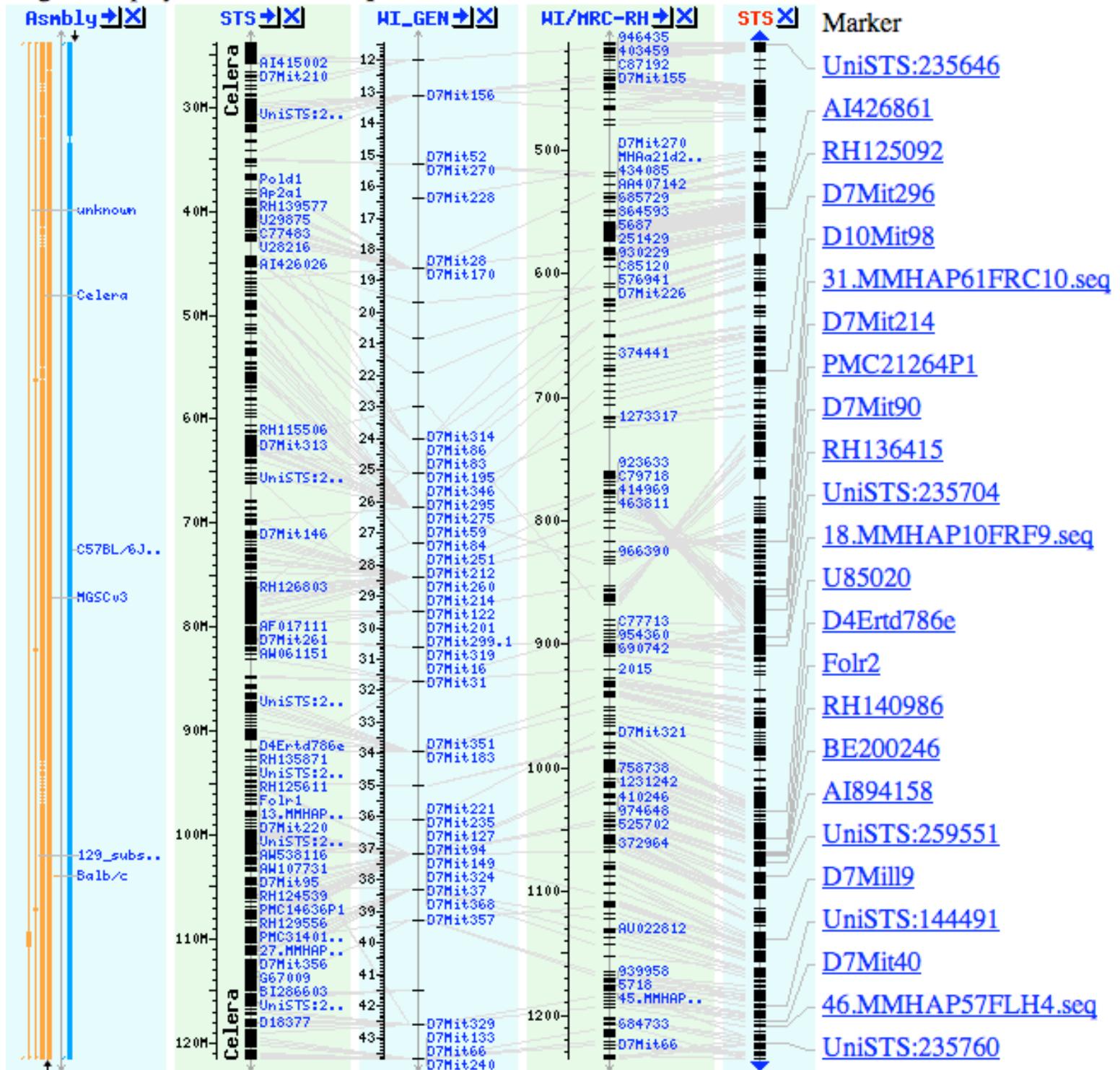
Near-exact matches



| | | | | | | | | | | | | | | | | | | | | | |
|--------------------|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Homo sapiens | A | T | C | T | T | G | C | A | T | C | C | T | C | C | A | C | T | G | C | C | T |
| Mus musculus | A | C | C | T | G | G | C | A | T | C | C | T | C | T | A | C | T | G | C | C | T |
| Canis familiaris | A | C | C | T | G | G | C | A | T | C | C | T | C | T | A | C | T | G | C | C | T |
| Monodelphis do ... | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . |
| Rattus norvegicus | A | C | C | T | G | G | C | A | T | C | C | T | C | T | A | C | T | G | C | C | T |
| Bos taurus | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . | . |
| Gallus gallus | A | C | C | T | G | G | C | A | T | C | C | T | C | T | A | T | T | G | C | T | T |
| Pan troglodytes | a | t | c | t | t | g | c | a | t | c | c | t | c | c | a | c | t | g | c | c | t |
| Macaca mulatta | A | T | C | T | T | G | C | A | T | C | C | T | C | C | A | C | T | G | C | C | T |



Region Displayed: 30M-120M bp





NCBI Map Viewer

Genomic Biology | Genome | Taxonomy | Entrez | BLAST | Help

Search - Select Group or Organism -

for

Go!

Click on the organism name to go to the genome view

Vertebrates

Mammals

- [BLAST](#) *Bos taurus* (cow) Build 3.1
- [BLAST](#) *Bos taurus* (cow) Build 2.1
- [BLAST](#) *Canis familiaris* (dog)
- [BLAST](#) *Felis catus* (cat)
- [BLAST](#) *Homo sapiens* (human) Build 36
- [BLAST](#) *Homo sapiens* (human) Build 35
- [BLAST](#) *Macaca mulatta* (rhesus macaque)
- [BLAST](#) *Mus musculus* (mouse) Build 36
- [BLAST](#) *Mus musculus* (mouse) Build 35
- [BLAST](#) *Ovis aries* (sheep)
- [BLAST](#) *Pan troglodytes* (chimpanzee)
- [BLAST](#) *Rattus norvegicus* (rat)
- [BLAST](#) *Sus scrofa* (pig)
- ...

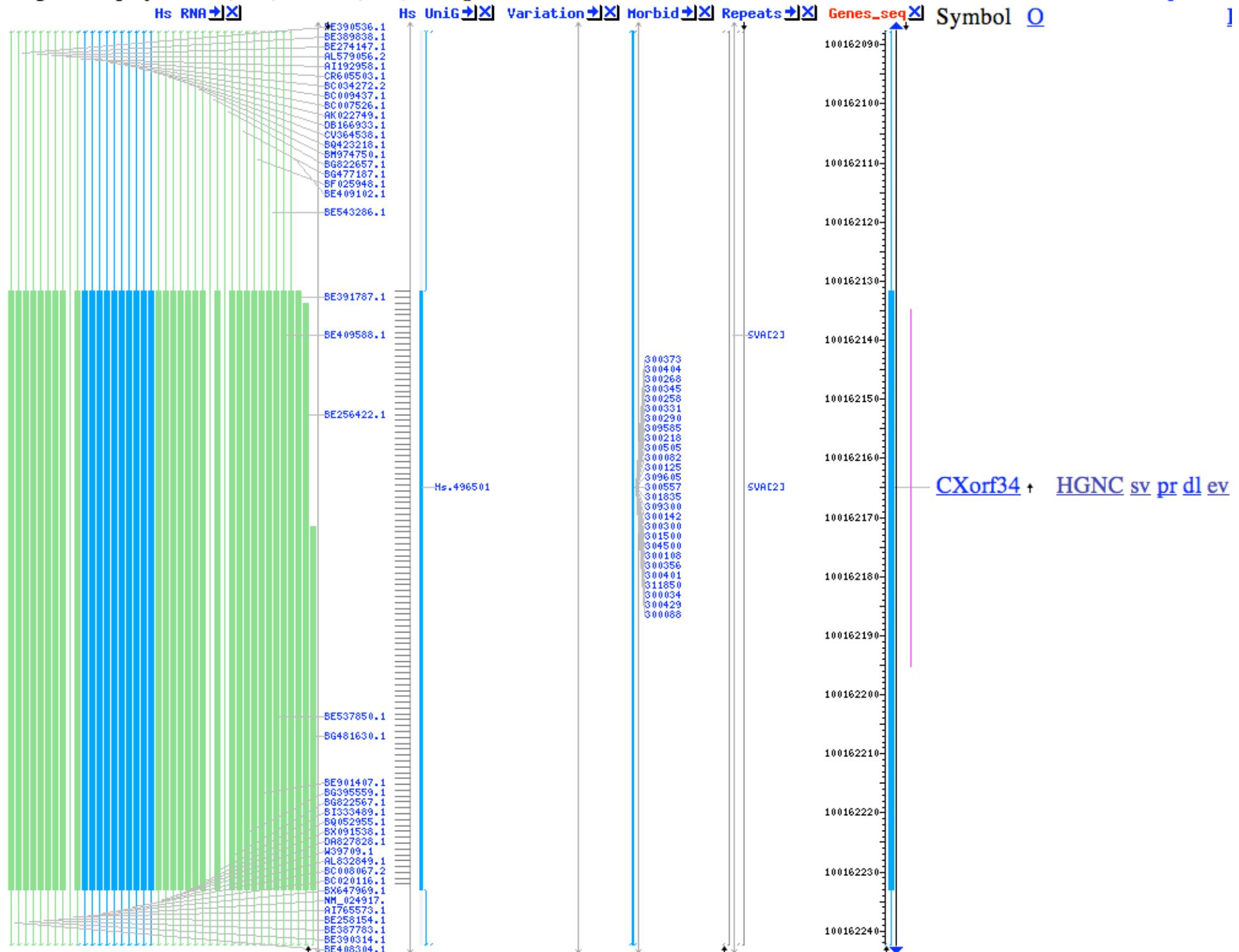
[Switch to Graphical View](#)

Plants [BLAST](#) [Search all plant maps](#)

- [BLAST](#) *Aegilops tauschii*
- [BLAST](#) *Aegilops umbellulata*
- [BLAST](#) *Allium cepa* (onion)
- [BLAST](#) *Arabidopsis thaliana* (thale cress)
- [BLAST](#) *Avena sativa* (oat)
- [BLAST](#) *Beta vulgaris* (beet)
- [BLAST](#) *Brassica juncea* (Indian mustard)
- [BLAST](#) *Brassica napus* (oilseed rape)
- [BLAST](#) *Brassica nigra* (black mustard)
- [BLAST](#) *Brassica oleracea*
- [BLAST](#) *Brassica rapa* (field mustard)
- [BLAST](#) *Capsicum annuum*
- [BLAST](#) *Eragrostis tef* (tef)
- [BLAST](#) *Glycine max* (soybean)
- [BLAST](#) *Hordeum vulgare* (barley)

Region Displayed: 100,162,088-100,162,242 bp

[Download/View Sequence](#)





Phenotypic Allele Detail

Your Input Welcome

| Allele | Symbol: Brca1^{tm1Arge} Name: targeted mutation 1, Argiris Efstratiadis ID: MGI:1930613 | | | | | | | | | | | | | | | | | | | | | | | | | | |
|--|---|--------------------|----------|--|---------------------|--------------------|---|-------------------------------|--|---|-------------------------------|--|--|---|--|--|-------------------------------|--|--|--------------------------------|--|-------|--|---------------------|--------------------|---|-------------------------------|
| Synonyms | Brca1^{ex2} | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Allele details | Allele Type: Targeted (knock-out) Strain of Origin: 129S/SvEv-Gpi ^c ES Cell Line: CCE/EK.CCE ES Cell Line Strain: 129S/SvEv-Gpi ^c Mutation: Disruption caused by insertion of vector Replacement of exon 2, encoding part of the RING finger motif, and part of the flanking introns with a neomycin cassette. (J:40594) Gene Expression in Brca1^{tm1Arge} mutants (3 assay results) International Mouse Strain Resource: (Search for IMSR strains with Brca1 mutations) References and Additional Notes: (See Below) | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Gene information | Symbol: Brca1 Name: breast cancer 1 Chromosome: 11 Genetic Position: 60.5 cM, cytoband D Genome Coordinates: Chr11:101304854-101368045 bp, - strand (From VEGA annotation of NCBI Build 36) Human Ortholog: BRCA1 | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Phenotypes | <p style="text-align: center;">Phenotypic details for all genotypes that include at least one Brca1^{tm1Arge} allele</p> <table border="1"> <thead> <tr> <th rowspan="2">Phenotype</th> <th colspan="2">Genotype</th> </tr> <tr> <th>Allelic Composition</th> <th>Genetic Background</th> </tr> </thead> <tbody> <tr> <td>Go To Brca1^{tm1Arge}/Brca1^{tm1Arge}</td><td>Involves: 129S/SvEv * C57BL/6</td><td></td></tr> <tr> <td>Go To Brca1^{tm1Arge}/Brca1⁺</td><td>Involves: 129S/SvEv * C57BL/6</td><td></td></tr> <tr> <td>Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} Trp53^{tm1Tyr}/Trp53^{tm1Tyr}</td><td>Involves: 129S/SvEv * 129S2/SvPas * C57BL/6</td><td></td></tr> <tr> <td>Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} Brca2^{tm1Arge}/Brca2^{tm1Arge}</td><td>Involves: 129S/SvEv * C57BL/6</td><td></td></tr> <tr> <td>Go To Bard1^{tm1Thl}/Bard1^{tm1Thl} Brca1^{tm1Arge}/Brca1^{tm1Arge}</td><td>Involves: 129S/SvEv * 129S1/Sv</td><td></td></tr> <tr> <td colspan="2"><hr/></td></tr> <tr> <th>Allelic Composition</th><th>Genetic Background</th></tr> <tr> <td>Brca1^{tm1Arge}/Brca1^{tm1Arge}</td><td>Involves: 129S/SvEv * C57BL/6</td></tr> </tbody> </table> <p>lethality/embryonic-perinatal embryonic lethality before turning of embryo (J:40594) <input type="radio"/> E5.5 -E8.5</p> <p>embryogenesis absent mesoderm (J:40594) abnormal embryonic tissue morphology (J:40594) <input type="radio"/> many empty decidua found; some decidua contained only giant cells and extraembryonic membranes, and others also contained some embryonic remnants abnormal egg cylinder morphology (J:40594) <input type="radio"/> at e5.5, embryonic tissue, when present, appears as small and short egg cylinders compared to controls; no clear division between embryonic and extraembryonic tissue is apparent <input type="radio"/> at e6.5, embryos, when present, are half the size of controls and were two-layered cylinders <input type="radio"/> at e6.5, mutants lack amniotic folds and the primary giant cells are prominent and large <input type="radio"/> at e7.5, embryos are small egg cylinders without amniotic folds and embryonic mesoderm is not visible; some extraembryonic tissue development appears normal <input type="radio"/> at E8.5, remaining embryos are variable in shape and achieved developmental stage; some consist of only extraembryonic tissue, some have rudimentary embryonic epithelium with no mesoderm, while others appear to have developed further to the heart beat stage, although the embryo is severely reduced in size <input type="radio"/> at E9.5, remaining embryos are variable in shape and achieved developmental stage absent amniotic folds (J:40594) embryonic growth retardation (J:40594)</p> | Phenotype | Genotype | | Allelic Composition | Genetic Background | Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} | Involves: 129S/SvEv * C57BL/6 | | Go To Brca1^{tm1Arge}/Brca1⁺ | Involves: 129S/SvEv * C57BL/6 | | Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} Trp53^{tm1Tyr}/Trp53^{tm1Tyr} | Involves: 129S/SvEv * 129S2/SvPas * C57BL/6 | | Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} Brca2^{tm1Arge}/Brca2^{tm1Arge} | Involves: 129S/SvEv * C57BL/6 | | Go To Bard1^{tm1Thl}/Bard1^{tm1Thl} Brca1^{tm1Arge}/Brca1^{tm1Arge} | Involves: 129S/SvEv * 129S1/Sv | | <hr/> | | Allelic Composition | Genetic Background | Brca1^{tm1Arge}/Brca1^{tm1Arge} | Involves: 129S/SvEv * C57BL/6 |
| Phenotype | Genotype | | | | | | | | | | | | | | | | | | | | | | | | | | |
| | Allelic Composition | Genetic Background | | | | | | | | | | | | | | | | | | | | | | | | | |
| Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} | Involves: 129S/SvEv * C57BL/6 | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Go To Brca1^{tm1Arge}/Brca1⁺ | Involves: 129S/SvEv * C57BL/6 | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} Trp53^{tm1Tyr}/Trp53^{tm1Tyr} | Involves: 129S/SvEv * 129S2/SvPas * C57BL/6 | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Go To Brca1^{tm1Arge}/Brca1^{tm1Arge} Brca2^{tm1Arge}/Brca2^{tm1Arge} | Involves: 129S/SvEv * C57BL/6 | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Go To Bard1^{tm1Thl}/Bard1^{tm1Thl} Brca1^{tm1Arge}/Brca1^{tm1Arge} | Involves: 129S/SvEv * 129S1/Sv | | | | | | | | | | | | | | | | | | | | | | | | | | |
| <hr/> | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Allelic Composition | Genetic Background | | | | | | | | | | | | | | | | | | | | | | | | | | |
| Brca1^{tm1Arge}/Brca1^{tm1Arge} | Involves: 129S/SvEv * C57BL/6 | | | | | | | | | | | | | | | | | | | | | | | | | | |

» Dataset:
Homo sapiens genes
(NCBI36)
» Attributes (Features)
Ensembl Gene ID
Ensembl Transcript ID
RefSeq DNA ID
Chromosome Name
Start Position (bp)
End Position (bp)
Strand
» Filters
Refseq DNA ID(s): [ID-list specified]

» Dataset:
[None Selected]

Features SNPs
 Structures Sequences

⊕ REGION:

⊖ GENE:

Ensembl Attributes

- | | |
|---|---|
| <input checked="" type="checkbox"/> Ensembl Gene ID | <input type="checkbox"/> Ensembl Peptide length |
| <input checked="" type="checkbox"/> Ensembl Transcript ID | <input type="checkbox"/> Transcript count |
| <input type="checkbox"/> Ensembl Peptide ID | <input type="checkbox"/> % GC content |
| <input type="checkbox"/> External Gene ID | <input type="checkbox"/> Description |
| <input type="checkbox"/> External Gene DB | <input type="checkbox"/> Biotype |
| <input type="checkbox"/> Ensembl CDS length | <input type="checkbox"/> Source |
| <input type="checkbox"/> Ensembl cDNA length | <input type="checkbox"/> Status |

GO Attributes

- | | |
|---|---|
| <input type="checkbox"/> GO ID | <input type="checkbox"/> GO evidence code |
| <input type="checkbox"/> GO description | |

External References (max 3)

- | | |
|--------------------------------------|--|
| <input type="checkbox"/> CCDS ID | <input type="checkbox"/> RefSeq Predicted DNA ID |
| <input type="checkbox"/> Codelink ID | <input type="checkbox"/> RefSeq Peptide ID |
| <input type="checkbox"/> EMBL ID | <input type="checkbox"/> Rfam ID |

| Ensembl Gene ID | Ensembl Transcript ID | RefSeq DNA ID | Chromosome Name | Start Position (bp) | End Position (bp) | Strand |
|---------------------------------|---------------------------------|---------------|--------------------|---------------------------|---------------------------|--------|
| ENSG00000113525 | ENST00000231454 | NM_000879 | 5 | 131905035 | 131907113 | -1 |
| ENSG00000100296 | ENST00000358079 | NM_001002879 | 22 | 28231868 | 28279703 | -1 |
| ENSG00000172531 | ENST00000312989 | NM_001008709 | 11 | 66922228 | 66925978 | -1 |
| ENSG00000204075 | ENST00000372866 | NM_001008739 | 6 | 42966199 | 42966504 | -1 |
| ENSG00000146955 | ENST00000275874 | NM_001008749 | 7 | 139753916 | 139772419 | 1 |
| ENSG00000163806 | ENST00000296122 | NM_001008779 | 2 | 28828118 | 28926981 | 1 |
| ENSG00000171649 | ENST00000307468 | NM_001010879 | 19 | 62787320 | 62795570 | 1 |
| ENSG00000105146 | ENST00000302804 | NM_001015879 | 19 | 62434240 | 62438727 | 1 |
| ENSG00000127241 | ENST00000337774 | NM_001879 | 3 | 188418632 | 188492446 | -1 |
| ENSG00000003402 | ENST00000309955 | NM_003879 | 2 | 201689135 | 201737246 | 1 |

| Human Ens. Gene ID | Mouse Ens Gene ID | Mouse chromosome |
|--------------------|--------------------|------------------|
| ENSG00000205070 | ENSMUSG00000069038 | Y |
| ENSG00000102144 | ENSMUSG00000066632 | 12 |
| ENSG00000174028 | ENSMUSG00000029672 | 6 |
| ENSG00000123130 | ENSMUSG00000047565 | 15 |

Table Browser

Use this program to retrieve the data associated with a track in text format, to calculate intersections between tracks, and to retrieve DNA sequence covered by a track. See [Using the Table Browser](#) for a description of the controls in this form. For more complex queries, you may want to use our [public MySQL server](#). Refer to the [Credits](#) page for the list of contributors and usage restrictions associated with these data.

clade: Vertebrate genome: Human assembly: Mar. 2006

group: Variation and Repeats track: SNPs

table:.snp126 [describe table schema](#)

region: genome position chrX:151073054-151383976 [lookup](#)

identifiers (names/accessions): [paste list](#) [upload list](#)

filter: [create](#)

intersection: [create](#)

correlation: [create](#)

output format: all fields from selected table

output file: (leave blank to keep output in browser)

file type returned: plain text gzip compressed

[get output](#) [summary/statistics](#)

Schema for SNPs - Simple Nucleotide Polymorphisms (dbSNP build 126)

Database: hg18 **Primary Table:**.snp126 **Row Count:** 12,351,941

Description: Polymorphism data from dbSnp database or genotyping arrays

| field | example | SQL type | description |
|------------|--------------|--|--|
| bin | 585 | smallint(5) unsigned | Indexing field to speed chromosome range queries. |
| chrom | chr10_random | varchar(15) | Reference sequence chromosome or scaffold |
| chromStart | 490 | int(10) unsigned | Start position in chrom |
| chromEnd | 491 | int(10) unsigned | End position in chrom |
| name | rs28694205 | varchar(15) | Reference SNP identifier or Affy SNP name |
| score | 0 | smallint(5) unsigned | Not used |
| strand | + | enum('?', '+', '-') | Which DNA strand contains the observed alleles |
| refNCBI | A | blob | Reference genomic from dbSNP |
| refUCSC | A | blob | Reference genomic from nib lookup |
| observed | A/T | varchar(255) | The sequences of the observed alleles from rs-fasta files |
| molType | genomic | enum('unknown', 'genomic', 'cDNA') | Sample type from exemplar ss |
| class | single | enum('unknown', 'single', 'in-del', 'het', 'microsatellite', 'named', 'no var', 'mixed', 'mnp', 'insertion', 'deletion') | The class of variant (simple, insertion, deletion, range, etc.) |
| valid | unknown | set('unknown', 'by-cluster', 'by-frequency', 'by-submitter', 'by-2hit-2allele', 'by-hapmap') | The validation status of the SNP |
| avHet | 0 | float | The average heterozygosity from all observations |
| avHetSE | 0 | float | The Standard Error for the average heterozygosity |
| func | intron | set('unknown', 'locus', 'coding', 'coding-synon', 'coding-nonsynon', 'untranslated', 'intron', 'splice-site', 'cds-reference') | The functional category of the SNP (coding-synon, coding-nonsynon, intron, etc.) |
| locType | exact | enum('unknown', 'range', 'exact', 'between', 'rangeInsertion', 'rangeSubstitution', 'rangeDeletion') | How the variant affects the reference sequence |
| weight | 1 | int(10) unsigned | The quality of the alignment |

Filter on Fields from hg18.snp126

| | | | | |
|------------|------|---------|---|-----|
| bin | is | ignored | * | AND |
| chrom | does | match | * | AND |
| chromStart | is | ignored | * | AND |
| chromEnd | is | ignored | * | AND |
| name | does | match | * | AND |
| score | is | ignored | * | AND |
| strand | does | match | * | AND |
| refNCBI | does | match | * | AND |
| refUCSC | does | match | * | AND |
| observed | does | match | * | AND |
| molType | does | match | * | AND |
| class | does | match | * | AND |
| valid | does | include | * | AND |
| avHet | is | ignored | * | AND |
| avHetSE | is | ignored | * | AND |
| func | does | include | * | AND |
| locType | does | match | * | AND |
| weight | is | ignored | * | AND |

AND Free-form query:

Lift Genome Annotations

This tool converts genome coordinates and genome annotation files between assemblies. The input data can be pasted into the text box, or uploaded from a file.

Original Genome:

Human

Original Assembly:

May 2004

New Genome:

Human

New Assembly:

Mar. 2006

Minimum ratio of bases that must remap:

0.95

Minimum chain size in target:

0

Minimum hit size in query:

0

Allow multiple output regions:

Min ratio of alignment blocks/exons that must map:

1

If thickStart/thickEnd is not mapped, use the closest mapped base:

For descriptions of the supported data formats, see the bottom of this page.

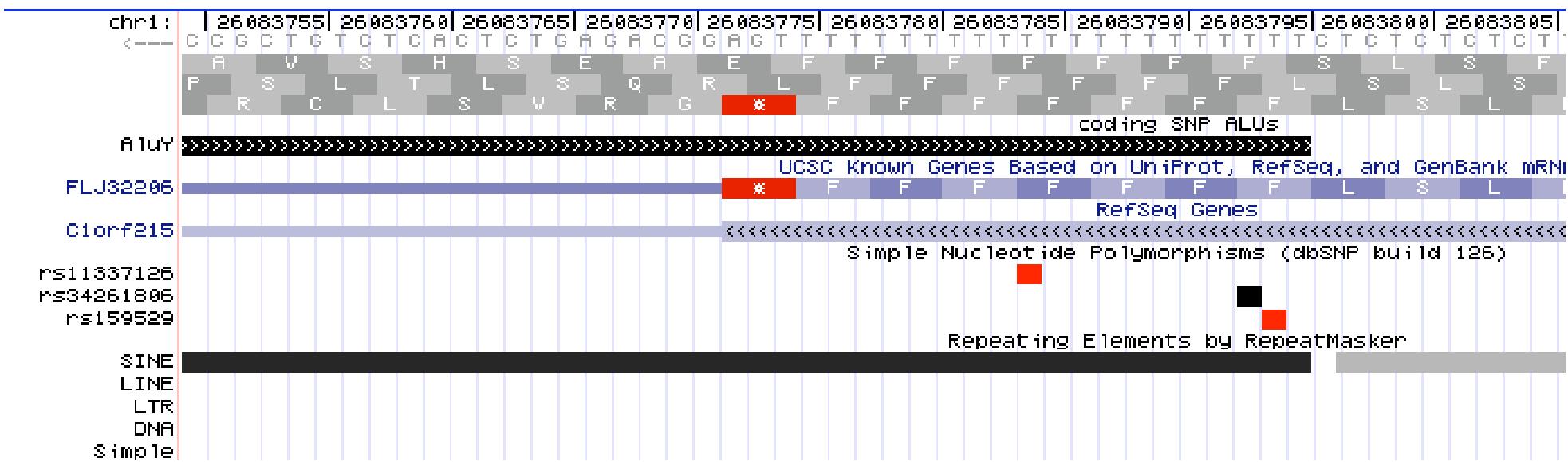
Data Format: BED

Paste in data:

```
chr1 164495305 164495447  
chr1 65011673 65011804  
chr1 179175367 179175462  
chr1 6289274 6312880 BF920027
```

```
x93828 0 +  
CV354059 0 +  
AA906903 0 -  
0 -
```

Submit



Tools

[Get Data](#)
[Get ENCODE Data](#)

[ENCODE Tools](#)
[Text Manipulation](#)

[Filter and Sort](#)

[Join, Subtract and Group](#)

- [Join two Queries](#) side by side on a specified field
- [Compare two Queries](#) to find common or distinct rows
- [Subtract Whole Query](#) from another query
- [Group data by a column](#) and perform aggregate operation on other columns.

[Convert Formats](#)

[Extract Features](#)

[Fetch Sequences](#)

[Fetch Alignments](#)

[Get Genomic Scores](#)

[Operate on Genomic Intervals](#)

[Statistics](#)

[Graph/Display Data](#)

[Evolution: HyPhy](#)

[EMBOSS](#)



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Download Galaxy brochure [here](#).

Unsequenced Genomes of the World | November 2007



Spectacled Flying Fox (*Pteropus conspicillatus*) | Queensland, Australia

Galaxy is for Biologists

Use this site to access popular sources of data like the UCSC Table Browser. Run analyses right on the spot using a variety of integrated tools. Your results are always available and can be easily shared with others. Just [watch](#) how.

Galaxy is for Developers

Galaxy is an easy-to-use, open-source, scalable framework for tool and data integration. Stop wasting time writing interfaces and get your tools used by biologists! Galaxy includes everything you need to get started, so [download](#) and start [integrating](#)!

History ([options](#))

[refresh](#) | [collapse all](#)

3: shortExons

2,444 regions, format: bed, database: hg18

Info: Filtering with c7 < 10, [save](#) | display at UCSC [main](#)

| 1 | 2 | 3 |
|------|---------|---------|
| chr1 | 7440 | 7444 |
| chr1 | 853117 | 853124 |
| chr1 | 855852 | 855859 |
| chr1 | 886796 | 886799 |
| chr1 | 1209148 | 1209150 |
| chr1 | 1212414 | 1212419 |

2: ensExonWithLength

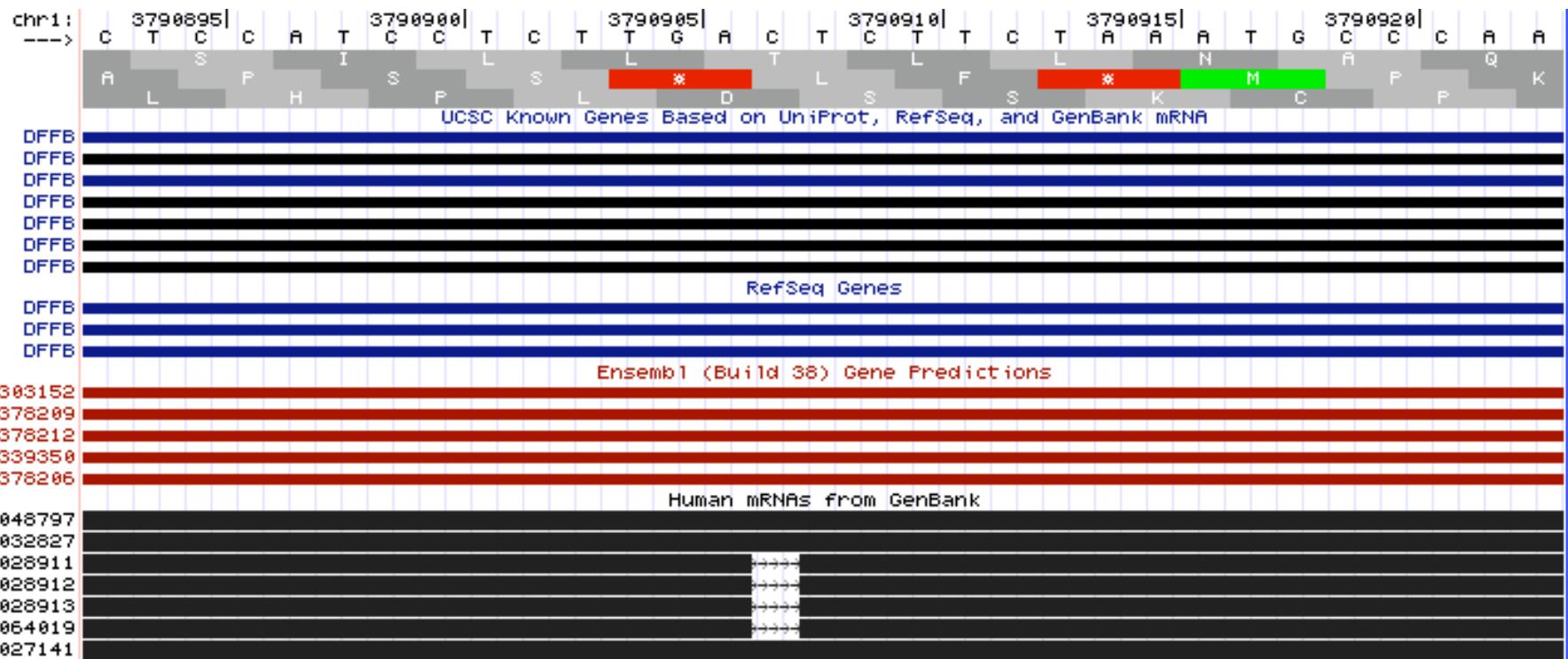
490,526 regions, format: bed, database: hg18

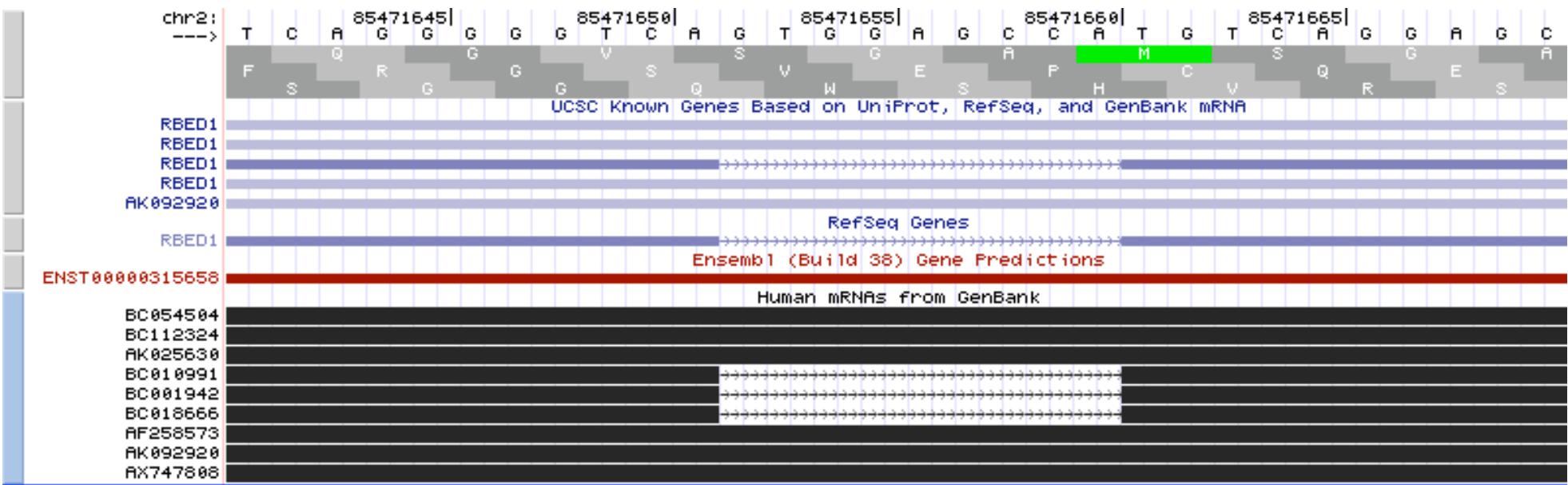
Info: Creating column 7 with expression c3-c2

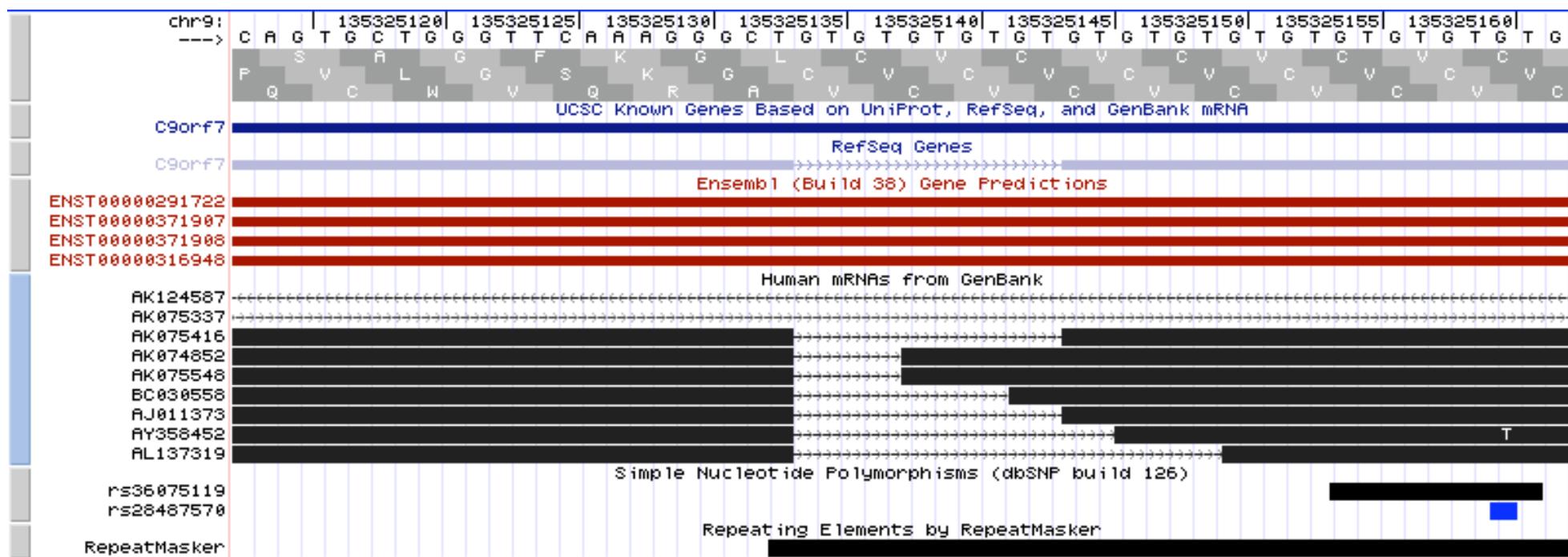
[save](#) | display at UCSC [main](#)

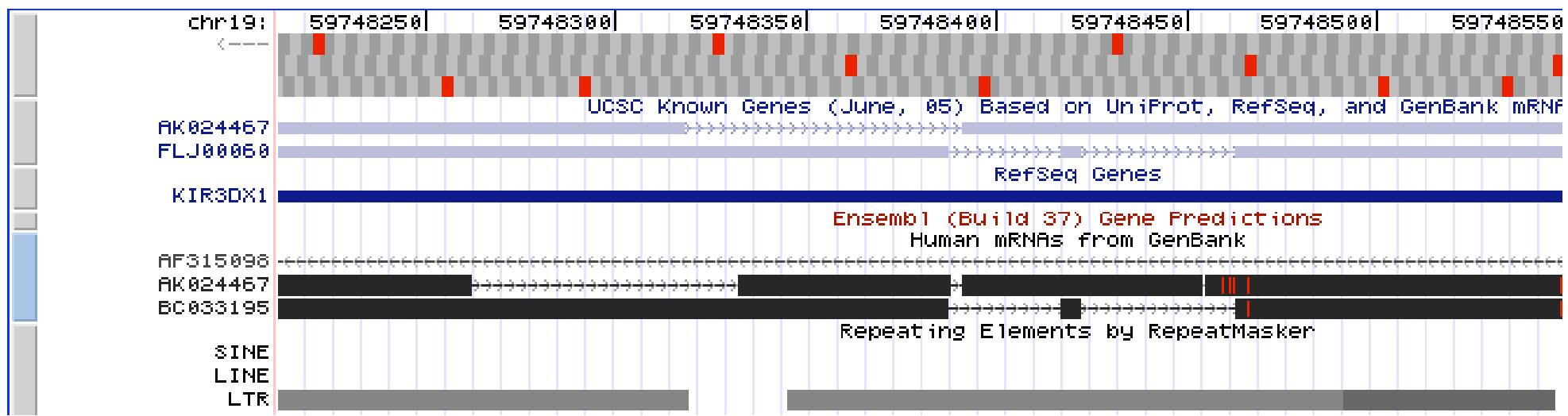
| 1 | 2 | 3 | 4 |
|------|------|------|------------------------|
| chr1 | 4273 | 4391 | ENST00000326632_exon_0 |
| chr1 | 4870 | 4901 | ENST00000326632_exon_1 |
| chr1 | 5658 | 5764 | ENST00000326632_exon_2 |
| chr1 | 5766 | 5810 | ENST00000326632_exon_3 |
| chr1 | 6469 | 6608 | ENST00000326632_exon_4 |
| chr1 | 6610 | 6628 | ENST00000326632_exon_5 |











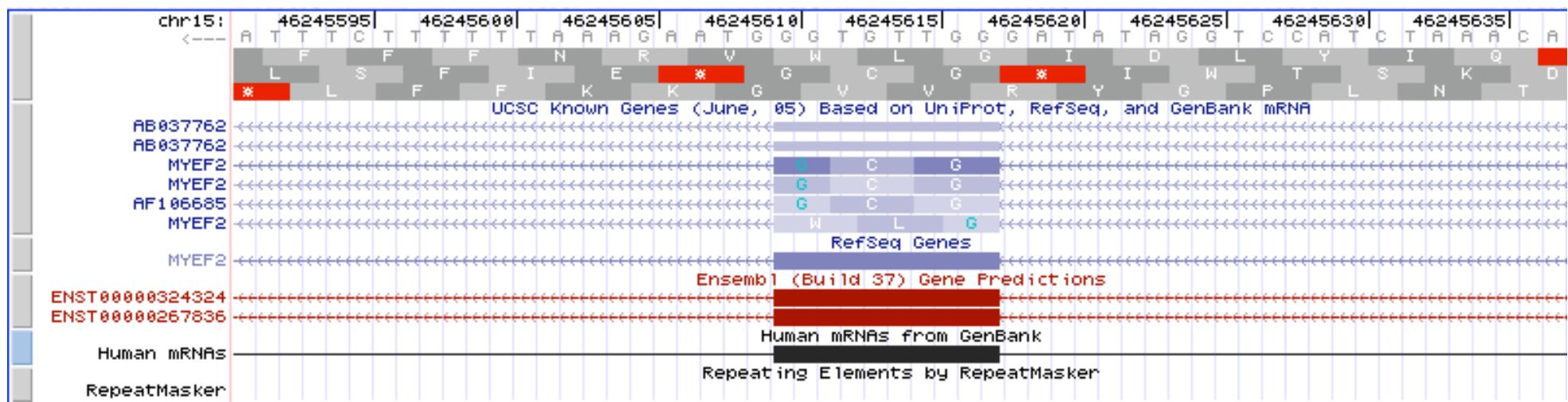


Table Browser

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clade: Vertebrate genome: Human assembly: Mar. 2006

group: All Tables database: proteome

table: proteome.spDisease

identif

- proteome.pepMolWtDist
- proteome.pepMwAa

filter:

- proteome.pepPi
- proteome.pepPiDist
- proteome.pepResDist

output

- proteome.pfamDesc
- proteome.pfamXref

output

- proteome.sfAssign
- proteome.sfDes

file type

- get or
- proteome.spDisease
- proteome.spOldNew

To reser

- proteome.spOrganism
- proteome.spReactomeEvent
- proteome.spReactomeID
- proteome.spSecondaryID
- proteome.spVariant

Using

- proteome.spXref2
- proteome.spXref3

This se

- proteome.swlInterPro

informa

- proteome.tableDescriptions

describe table schema

list upload list

table

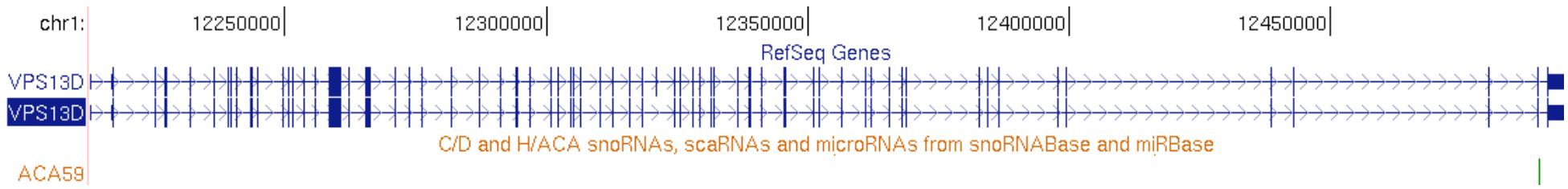
(leave blank to keep output in browser)

zip compressed

custom tracks), [click here](#).

the descriptions of the Table Browser controls. For more information see the [Table Browser User's Guide](#).





bioperl-1.5.2
bioperl-1.5.2::Bio
bioperl-1.5.2::Bio

SearchUtils
SearchWriterI
Seg
Segment
Segment
SegmentI
Seq

SeqAnalysisParser
SeqAnalysisParser
SeqAnalysisParser
SeqBuilder

SeqDiff

SeqFactory

SeqFastaSpeedFac

SeqFeature

SeqFeatureI

SeqHound
Search

SeqI
SacI

SeqI
SeqII

SeqI
SeqIO

SeqIO SeqPattern

SeqStats

SeqUtils

SeqVersi

SeqWithQu

Socorro

Summary

Bio::Seq - Sequence object, with features

Package variables

No package variables defined.

Included modules

Bio::Annotation::Collection

Bio::PrimarySeq

Inherit

Bio::AnnotatableI **Bio::DescribableI** **Bio::FeatureHolderI** **Bio::IdentifiableI** **Bio::F**

Synopsis

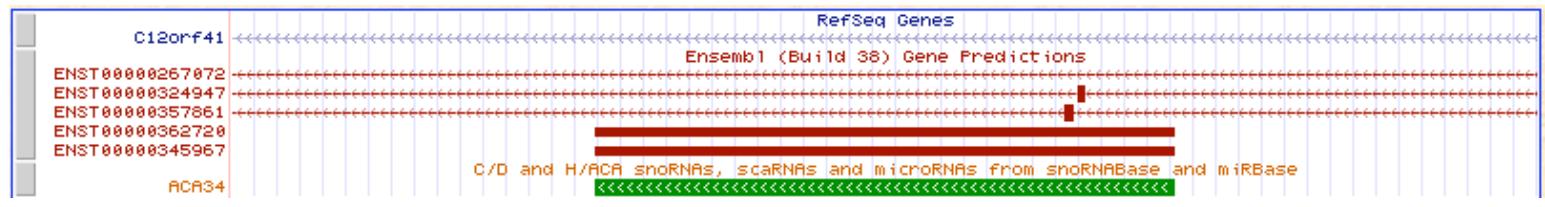
OR select a class from the list:

| | |
|--|----------------------------|
| Bio::DB::GFF::Aggregator::ucsc_genscan | UCSC genscan aggregator |
| Bio::Tools::Genscan | Results of one Genscan run |

sort by method ▾

methods for **Bio::Tools::Genscan**

| | | | |
|----------------------------------|--|--|--|
| next_feature | Bio::Tools::Genscan | A Bio::Tools::Prediction::Gene object. | while(\$gene = \$genscan->next_feature()) |
| next_prediction | Bio::Tools::Genscan | A Bio::Tools::Prediction::Gene object. | while(\$gene = \$genscan->next_prediction) |
| noclose | Bio::Root::IO | value of noclose (a scalar) | \$obj->noclose(\$newval) |
| otherwise | Bio::Root::Root | not documented | not documented |
| parse | Bio::Tools::AnalysisResult | not documented | not documented |
| qualify | Bio::Tools::Genscan | not documented | not documented |
| qualify_to_ref | Bio::Tools::Genscan | not documented | not documented |
| rmmtree | Bio::Root::IO | number of files successfully deleted | Bio::Root::IO->rmmtree(\$dirname); |
| stack_trace | Bio::Root::RootI | array containing a reference of arrays | @stack_array_ref= \$self->stack_trace |
| stack_trace_dump | Bio::Root::RootI | not documented | not documented |
| tempdir | Bio::Root::IO | The name of a new temporary directory | my (\$tempdir) = File::Tempdir->new(CLEANUP => |



position/search | chr4:158,477,294-158,477,353

jump

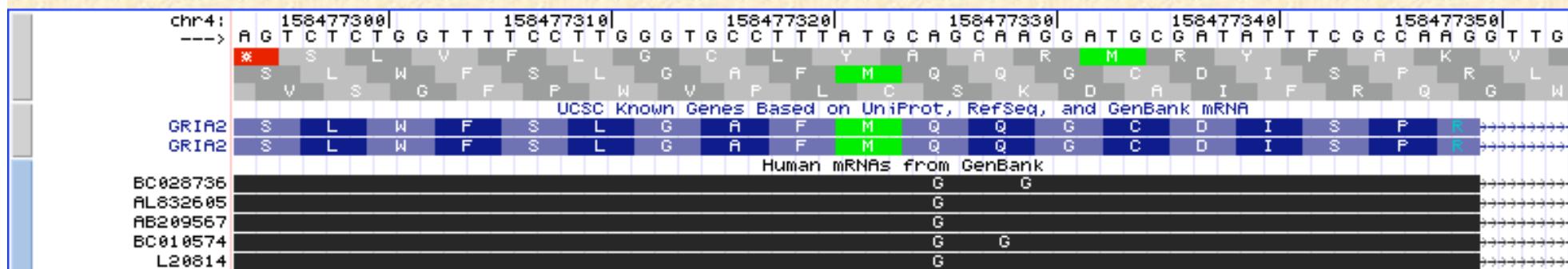
clear

size 60 bp.

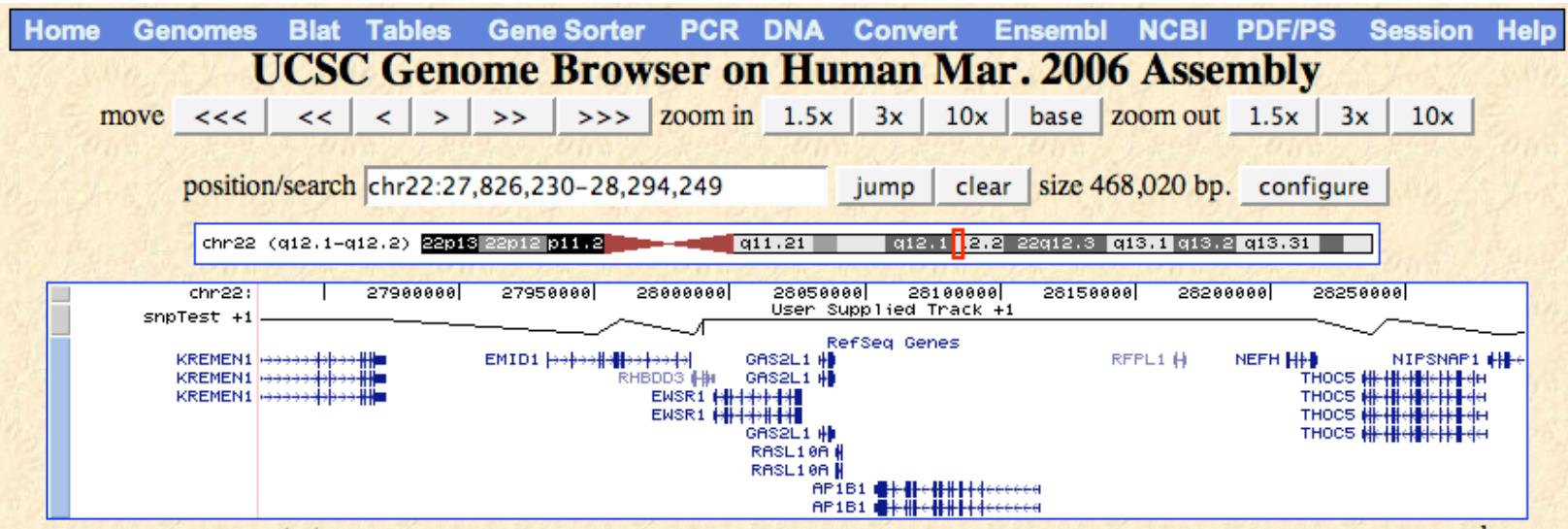
configure

chr4 (q32.1) 1413 q12

24q25q26 26.6



snpTest +1
28.1 Mb in
154 regions > 0.3
[chr22 14.4M to 14.7M](#)
[chr22 14.8M to 15.8M](#)
[chr22 15.8M to 15.9M](#)
[chr22 16.0M to 16.0M](#)
[chr22 16.0M to 16.0M](#)
[chr22 16.3M to 16.4M](#)
[chr22 16.5M to 16.6M](#)
[chr22 16.7M to 17.0M](#)
[chr22 17.0M to 17.1M](#)
[chr22 17.1M to 17.2M](#)
[chr22 17.2M to 17.3M](#)
[chr22 17.3M to 17.3M](#)
[chr22 17.4M to 17.4M](#)
[chr22 17.4M to 17.5M](#)
[chr22 17.5M to 17.5M](#)





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Abundance Profile

434 clusters retrieved.

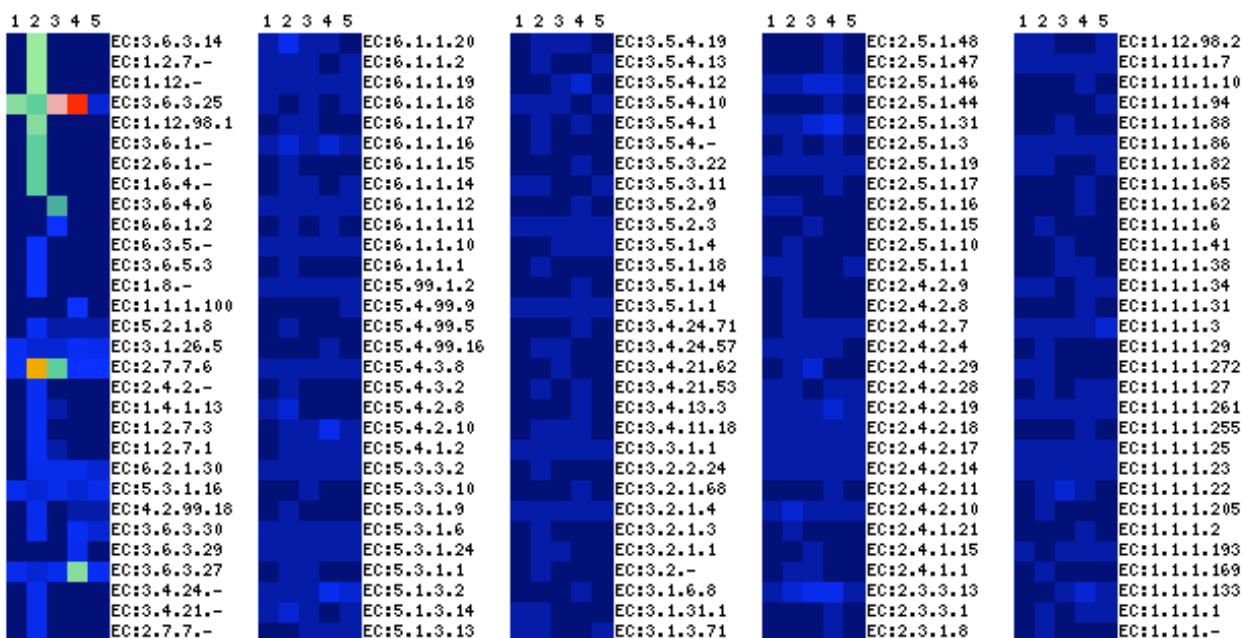
Mouse over labels to see additional information.

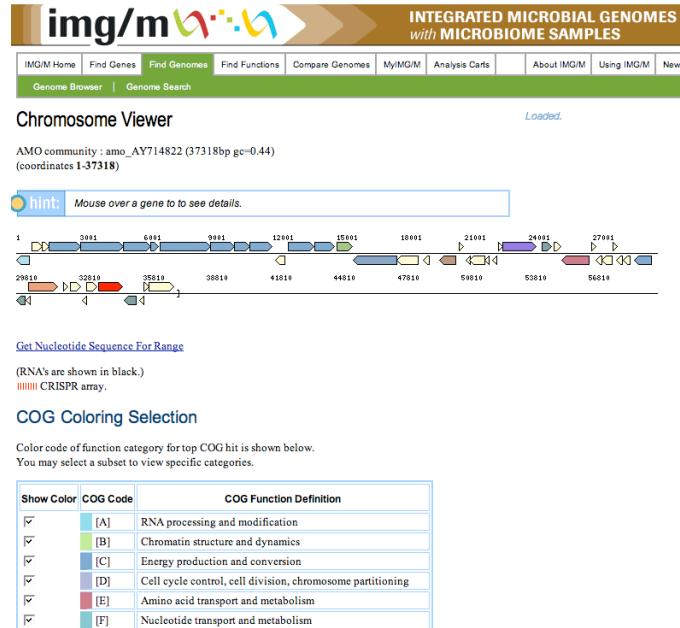
Clicking on the column number will sort rows for that column in descending gene count order.

Clicking on row cluster ID will add the cluster to the appropriate analysis cart (if cart is supported).

Mouse over heat map to see gene counts. Clicking on gene count will take you to the gene list.

- 1 - [Methanocaldococcus jannaschii DSM 2661](#)
- 2 - [Methanococcus maripaludis S2](#)
- 3 - [Methanosaeta thermophila PT](#)
- 4 - [Methanosarcina acetivorans C2A](#)
- 5 - [Methanothermobacter thermautotrophicus Delta H](#)





- 1 - [Mouse Gut Community lean1](#)
- 2 - [Mouse Gut Community lean2](#)
- 3 - [Mouse Gut Community ob1](#)
- 4 - [Mouse Gut Community ob2](#)

