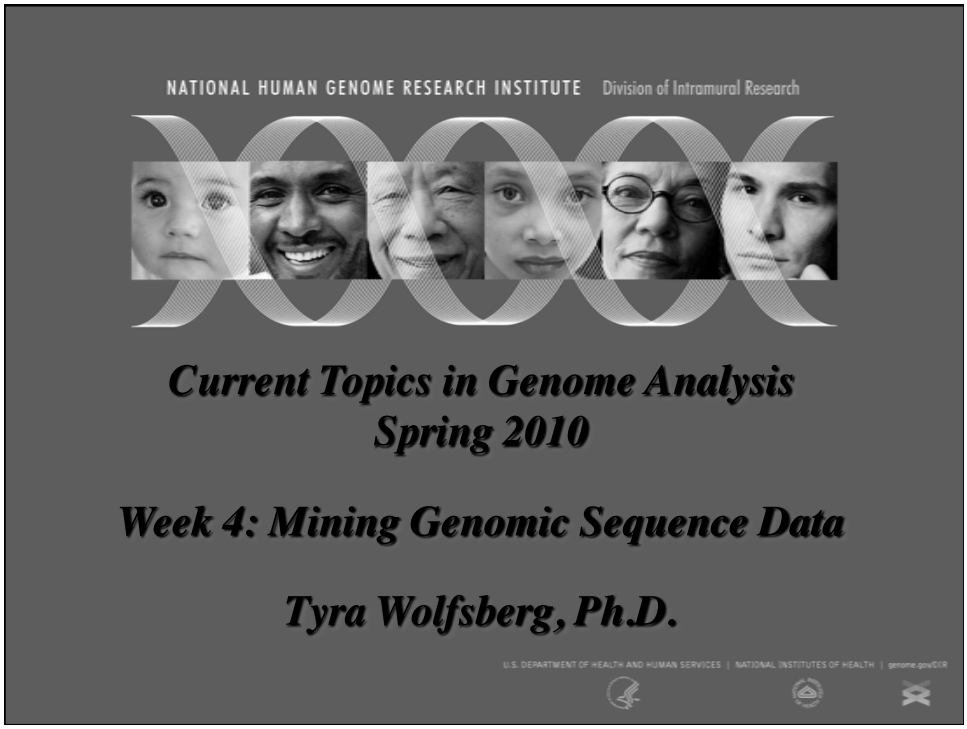


Accessing the public genome sequence data

UCSC's Genome Browser ("Golden Path")
http://genome.ucsc.edu

Ensembl
http://www.ensembl.org

NCBI's Map Viewer
http://www.ncbi.nlm.nih.gov/mapview/

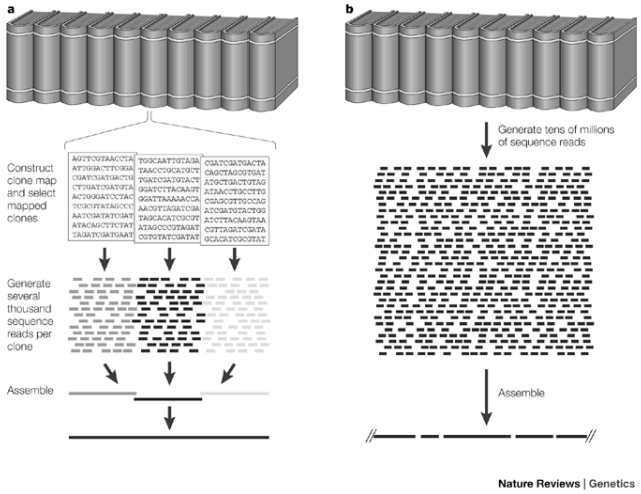
Types of data integrated in genome browsers

- Same starting material for all genome browsers: genomic sequence
- Annotations calculated independently by each genome browser
  - Genes
    - RefSeq mRNAs (non-redundant)
    - GenBank mRNAs (redundant)
    - ESTs
    - Gene predictions
  - SNPs
  - Homologous sequences from other organisms
  - STSs

Overview of genome sequencing strategies

Clone-by-clone shotgun sequencing

Whole-genome shotgun sequencing



Green ED. Strategies for the systematic sequencing of complex genomes. Nat Rev Genet. 2001. 2:573-83.

Genome Sequence Assemblies

- Complex algorithms needed to incorporate all sequence data
- Assemblies updated periodically as new sequence becomes available
  - Mouse and human genomes assembled by NCBI
  - Other genomes assembled by sequencing centers or consortia
- Assemblies not updated concurrently by the three Genome Browsers
  - "Pre-release" assemblies and annotations available at
    - UCSC: *http://genome-test.cse.ucsc.edu/*
    - pre!Ensembl: *http://pre.ensembl.org/*
  - UCSC and Ensembl provide archive of all genome assemblies and annotations; NCBI provides only limited archive
- IF YOU ARE COMPARING DATA FROM DIFFERENT GENOME BROWSERS, MAKE SURE YOU ARE LOOKING AT THE SAME VERSION OF THE ASSEMBLY

Genome Assembly Versions

	Same assembly?	UCSC	NCBI	Ensembl
Human	Yes	Feb 2009/hg19/ GRCh37	Build 37.1	GRCh37
Mouse	Yes	July 2007/mm9/Build 37	Build 37.1	Build 37
Dog	Yes	May 2005 /canFam 2.0	Build 2.1/ CanFam 2.0	CanFam 2.0
Zebrafish	NO	Dec 2008/danRer6/ Zv8	Zv7/build 3.1	Zv8
Rhesus	Yes	Jan 2006/rheMac2/ v. 1.0, Mmul_051212	Build 1.1/ v.1.0, Mmul_051 212	Mmul_1

NCBI Reference Sequences (RefSeqs)

- Non-redundant collection of richly annotated DNA, RNA, and protein sequences from diverse taxa
- Each RefSeq represents a single, naturally occurring molecule from one organism

	derived from GenBank submissions	model reference sequences produced by NCBI's Genome Annotation project
mRNA	NM_123456	XM_123456
protein	NP_123456	XP_123456
non-coding transcripts	NR_123456	XR_123456

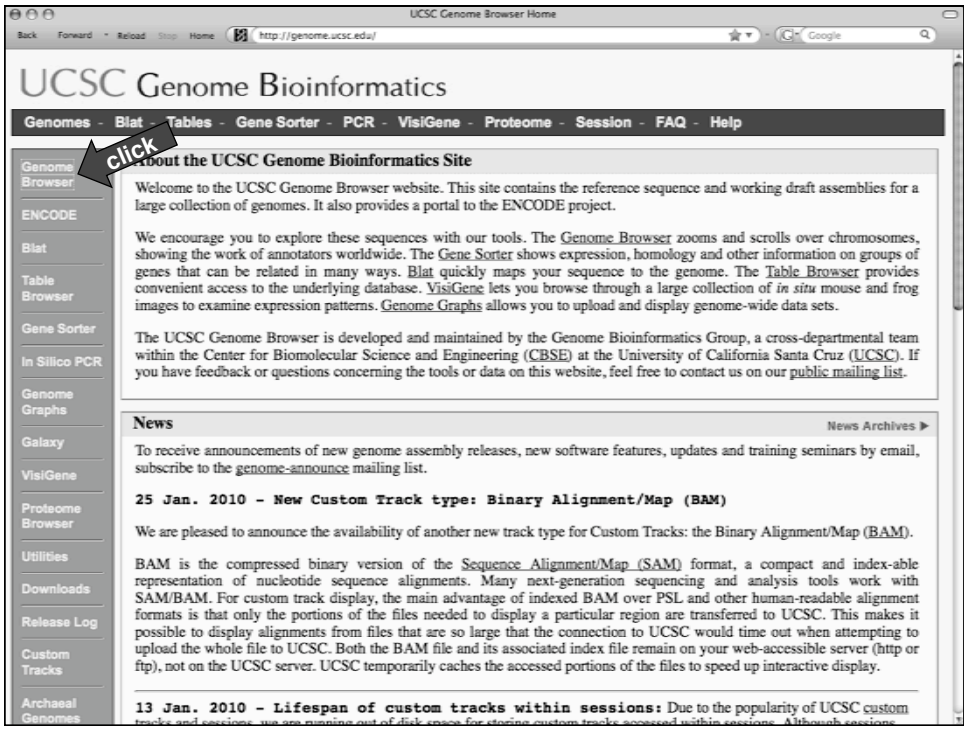
http://www.ncbi.nlm.nih.gov/RefSeq/key.html

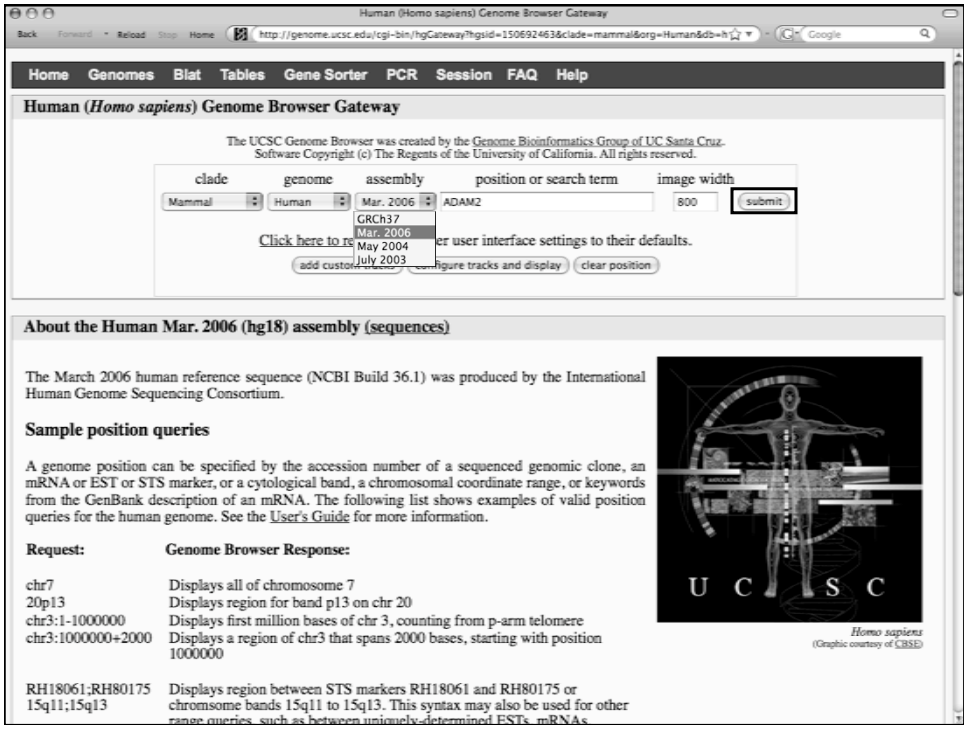
```
LOCUS       NM_001101               1852 bp    mRNA    linear   PRI 27-DEC-2009
DEFINITION  Homo sapiens actin, beta (ACTB), mRNA.
ACCESSION   NM_001101
VERSION     NM_001101.3  GI:168480144
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
            Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE   1  (bases 1 to 1852)
  AUTHORS   Yamaguchi,H., Shiraishi,M., Fukami,K., Tanabe,A., Ikeda-Matsuo,Y.,
            Naito,Y. and Sasaki,Y.
  TITLE     MARCKS regulates lamellipodia formation induced by IGF-I via
            association with PIP2 and beta-actin at membrane microdomains
  JOURNAL   J. Cell. Physiol. 220 (3), 748-755 (2009)
   PUBMED   19475567

COMMENT     REVIEWED REFSEQ: This record has been curated by NCBI staff. The
            reference sequence was derived from AK130157.1 and BC009636.1.
            On Feb 22, 2008 this sequence version replaced gi:5016088.

            Summary: This gene encodes one of six different actin proteins.
            Actins are highly conserved proteins that are involved in cell
            motility, structure, and integrity. This actin is a major
            constituent of the contractile apparatus and one of the two
            nonmuscle cytoskeletal actins. [provided by RefSeq].

            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.
            COMPLETENESS: complete on the 3' end.

     CDS             85..1212
                     /gene="ACTB"
                     /gene_synonym="PS1TP5BP1"
                     /note="beta cytoskeletal actin; PS1TP5-binding protein 1;
                     actin, cytoplasmic 1"
                     /codon_start=1
                     /product="beta actin"
                     /protein_id="NP_001092.1"
                     /db_xref="GI:4501885"
                     /db_xref="CCDS:CCDS5341.1"
                     /db_xref="GeneID:60"
                     /db_xref="HGNC:132"
                     /db_xref="HPRD:00032"
                     /db_xref="MIM:102630"
                     /translation="MDDDIAALVVDNGSGMCKAGFAGDDAPRAVFPSIVGRPRHQGVM
                     VGMGQKDSYVGDEAQSKRGILTLKYPIEHGIVTNWDDMEKIWHHTFYNELRVAPEEHP
                     VLLTEAPLNPKANREKMTQIMFETFNTPAMYVAIQAVLSLYASGRTTGIVMDSGDGVT
                     HTVPIYEGYALPHAILRLDLAGRDLTDYLMKILTERGYSFTTTAEREIVRDIKEKLCY
                     VALDFEQEMATAASSSSLEKSYELPDGQVITIGNERFRCPEALFQPSFLGMESCGIHE
                     TTFNSIMKCDVDIRKDLYANTVLSGGTTMYPGIADRMQKEITALAPSTMKIKIIAPPE
                     RKYSVWIGGSILASLSTFQQMWISKQEYDESGPSIVHRKCF"

ORIGIN
        1 accgccgaga ccgcgtccgc cccgcgagca cagagcctcg cctttgccga tccgccgccc
       61 gtccacaccc gccgccagct caccatggat gatgatatcg ccgcgctcgt cgtcgacaac
      121 ggctccggca tgtgcaaggc cggcttcgcg ggcgacgatg ccccccgggc cgtcttcccc
```

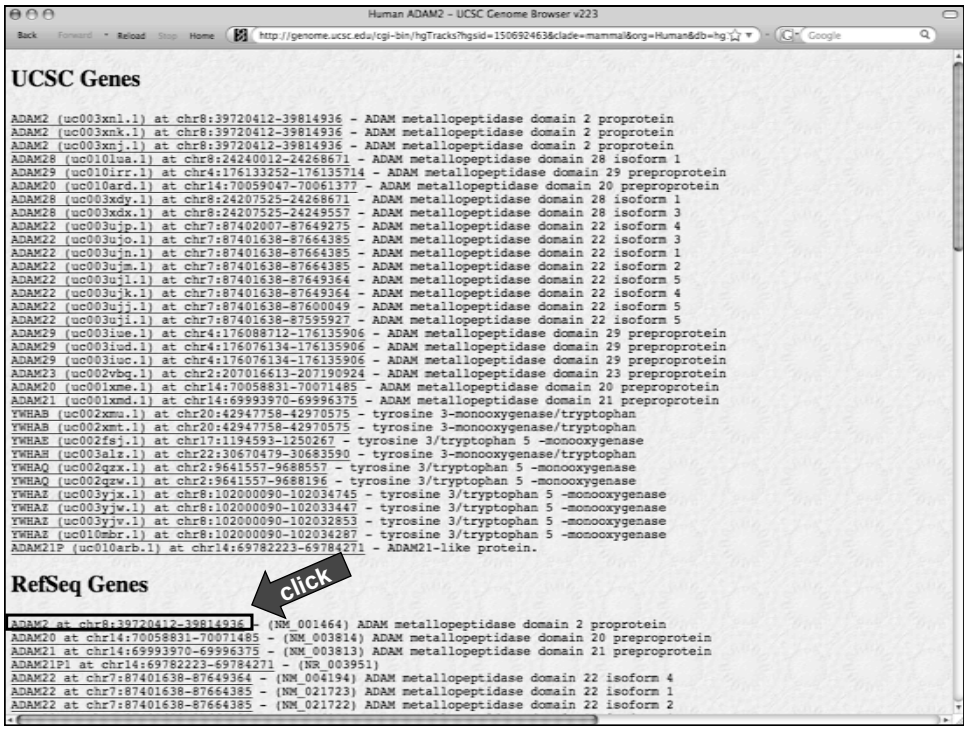
Beta actin mRNA RefSeq

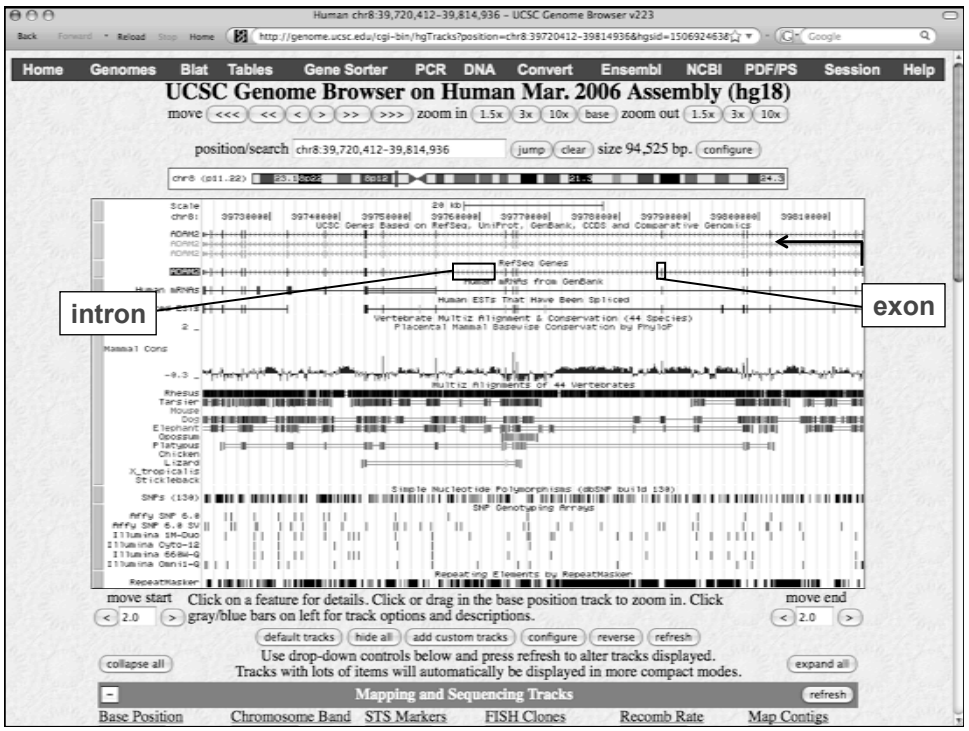
UCSC

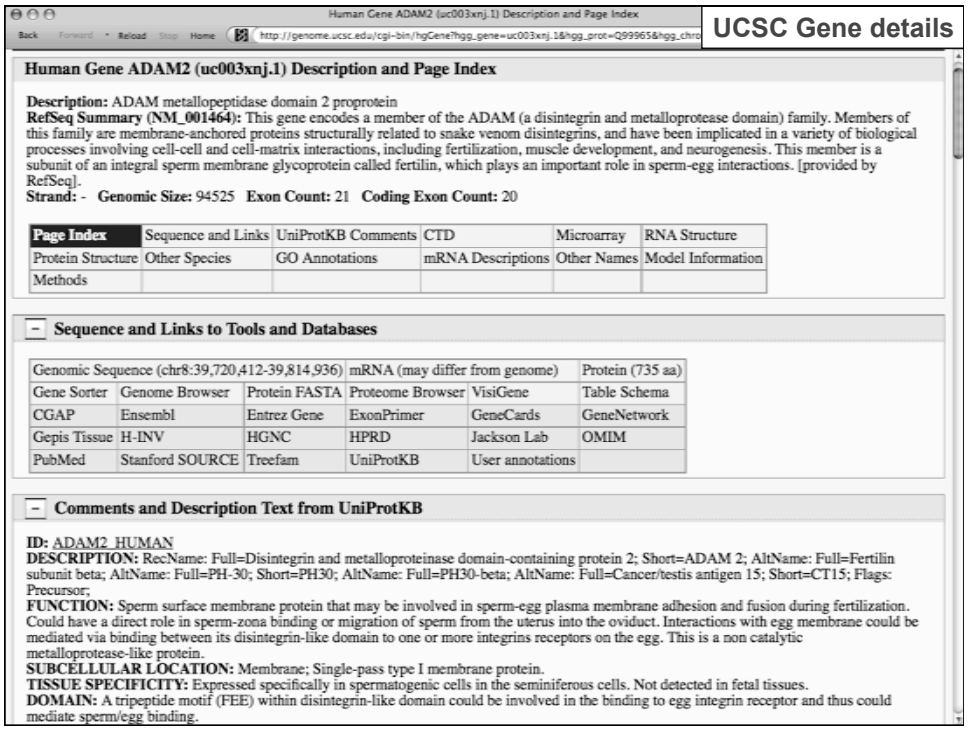
View a region in the genome by querying with a gene symbol

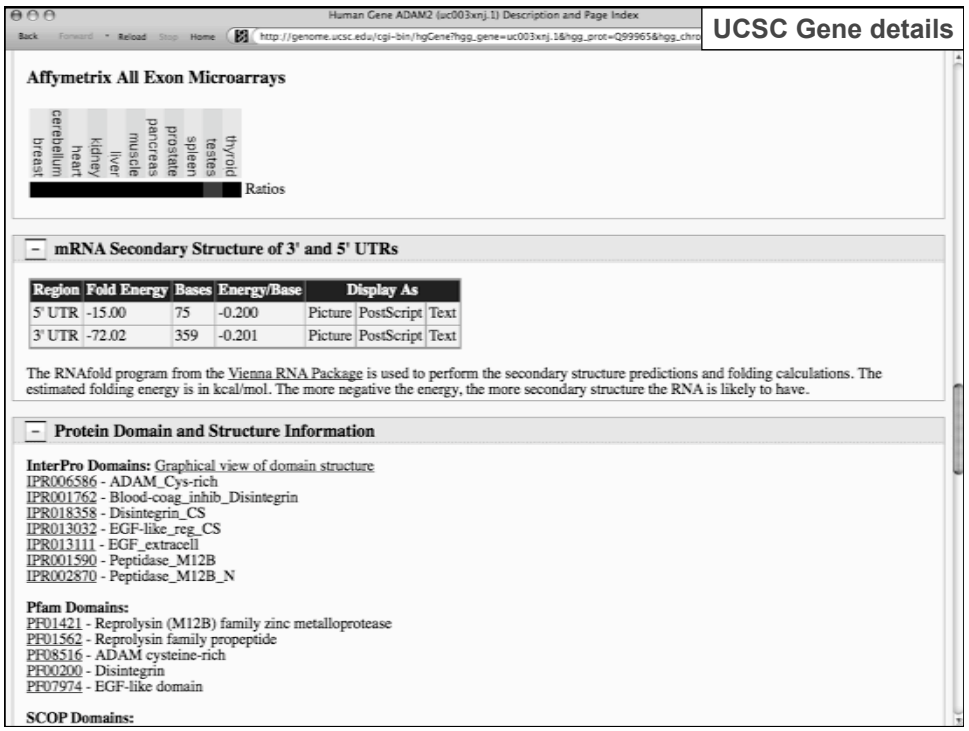


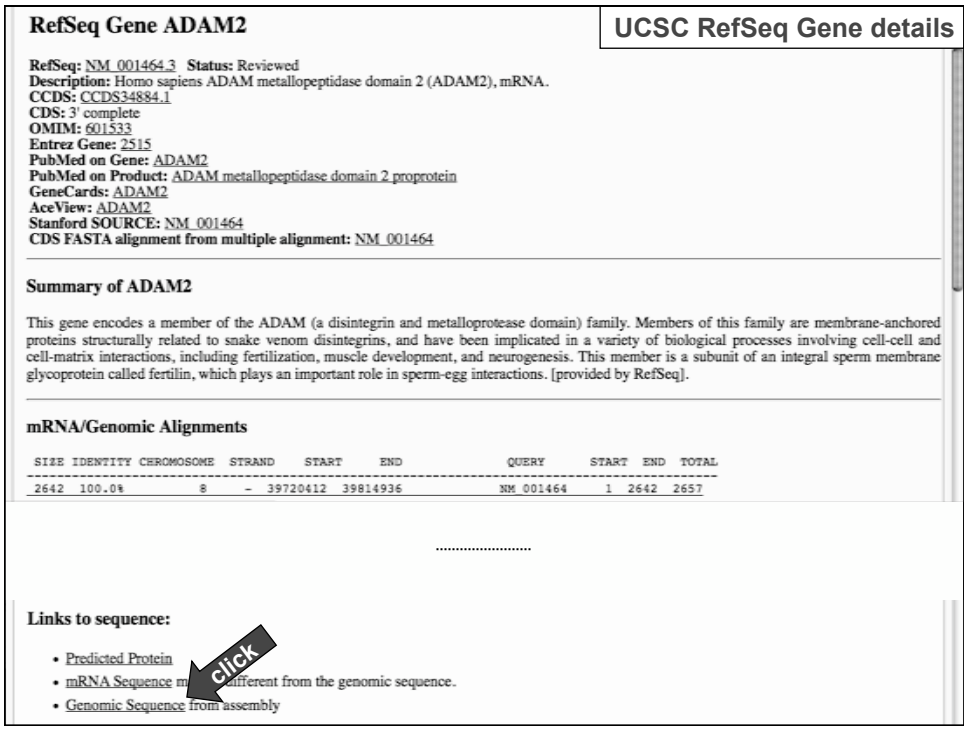

Human (Homo sapiens) Genome Browser Gateway
Home Genomes Blat Tables Gene Sorter PCR Session FAQ Help
Human (Homo sapiens) Genome Browser Gateway
The UCSC Genome Browser was created by the Genome Bioinformatics Group of UC Santa Cruz.
clade genome assembly position or search term image width
Mammal Human Mar. 2006 ADAM2 800 submit
GRCh37
Mar. 2006
May 2004
July 2003
add custom tracks configure tracks and display clear position
About the Human Mar. 2006 (hg18) assembly (sequences)
The March 2006 human reference sequence (NCBI Build 36.1) was produced by the International Human Genome Sequencing Consortium.
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
RH18061;RH80175 Displays region between STS markers RH18061 and RH80175 or
15q11;15q13 chromsome bands 15q11 to 15q13. This syntax may also be used for other
U C S C
Homo sapiens

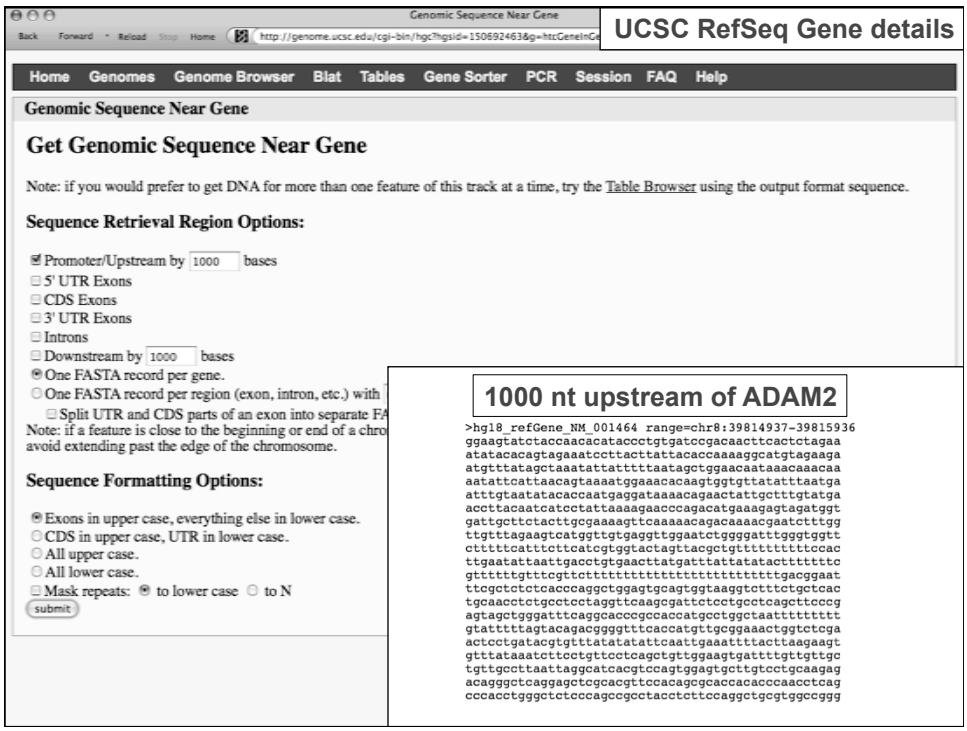

Human ADAM2 – UCSC Genome Browser v223
UCSC Genes
ADAM2 (uc003xnl.1) at chr8:39720412-39814936 - ADAM metallopeptidase domain 2 proprotein
ADAM2 (uc003xnk.1) at chr8:39720412-39814936 - ADAM metallopeptidase domain 2 proprotein
ADAM2 (uc003xnj.1) at chr8:39720412-39814936 - ADAM metallopeptidase domain 2 proprotein
ADAM28 (uc010lua.1) at chr8:24240012-24268671 - ADAM metallopeptidase domain 28 isoform 1
ADAM29 (uc010irr.1) at chr4:176133252-176135714 - ADAM metallopeptidase domain 29 preproprotein
ADAM20 (uc010ard.1) at chr14:70059047-70061377 - ADAM metallopeptidase domain 20 preproprotein
ADAM28 (uc003xdy.1) at chr8:24207525-24268671 - ADAM metallopeptidase domain 28 isoform 1
ADAM28 (uc003xdx.1) at chr8:24207525-24249557 - ADAM metallopeptidase domain 28 isoform 3
ADAM22 (uc003ujp.1) at chr7:87402007-87649275 - ADAM metallopeptidase domain 22 isoform 4
ADAM22 (uc003ujo.1) at chr7:87401638-87664385 - ADAM metallopeptidase domain 22 isoform 3
ADAM22 (uc003ujn.1) at chr7:87401638-87664385 - ADAM metallopeptidase domain 22 isoform 1
ADAM22 (uc003ujm.1) at chr7:87401638-87664385 - ADAM metallopeptidase domain 22 isoform 2
ADAM22 (uc003ujl.1) at chr7:87401638-87649364 - ADAM metallopeptidase domain 22 isoform 5
ADAM22 (uc003ujk.1) at chr7:87401638-87649364 - ADAM metallopeptidase domain 22 isoform 4
ADAM22 (uc003ujj.1) at chr7:87401638-87600049 - ADAM metallopeptidase domain 22 isoform 5
ADAM22 (uc003uji.1) at chr7:87401638-87595927 - ADAM metallopeptidase domain 22 isoform 5
ADAM29 (uc003iue.1) at chr4:176088712-176135906 - ADAM metallopeptidase domain 29 preproprotein
ADAM29 (uc003iud.1) at chr4:176076134-176135906 - ADAM metallopeptidase domain 29 preproprotein
ADAM29 (uc003iuc.1) at chr4:176076134-176135906 - ADAM metallopeptidase domain 29 preproprotein
ADAM23 (uc002vbq.1) at chr2:207016613-207190924 - ADAM metallopeptidase domain 23 preproprotein
ADAM20 (uc001xme.1) at chr14:70058831-70071485 - ADAM metallopeptidase domain 20 preproprotein
ADAM21 (uc001xmd.1) at chr14:69993970-69996375 - ADAM metallopeptidase domain 21 preproprotein
YWHAB (uc002xmu.1) at chr20:42947758-42970575 - tyrosine 3-monooxygenase/tryptophan
YWHAB (uc002xmt.1) at chr20:42947758-42970575 - tyrosine 3-monooxygenase/tryptophan
YWHAE (uc002fsj.1) at chr17:1194593-1250267 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAH (uc003alz.1) at chr22:30670479-30683590 - tyrosine 3-monooxygenase/tryptophan
YWHAQ (uc002qzx.1) at chr2:9641557-9688557 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAQ (uc002qzw.1) at chr2:9641557-9688196 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAZ (uc003yjx.1) at chr8:102000090-102034745 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAZ (uc003yjw.1) at chr8:102000090-102033447 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAZ (uc003yjv.1) at chr8:102000090-102032853 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAZ (uc010mbr.1) at chr8:102000090-102034287 - tyrosine 3/tryptophan 5 -monooxygenase
ADAM21P (uc010arb.1) at chr14:69782223-69784271 - ADAM21-like protein.
RefSeq Genes
click
ADAM2 at chr8:39720412-39814936 - (NM_001464) ADAM metallopeptidase domain 2 proprotein
ADAM20 at chr14:70058831-70071485 - (NM_003814) ADAM metallopeptidase domain 20 preproprotein
ADAM21 at chr14:69993970-69996375 - (NM_003813) ADAM metallopeptidase domain 21 preproprotein
ADAM21P1 at chr14:69782223-69784271 - (NR_003951)
ADAM22 at chr7:87401638-87649364 - (NM_004194) ADAM metallopeptidase domain 22 isoform 4
ADAM22 at chr7:87401638-87664385 - (NM_021723) ADAM metallopeptidase domain 22 isoform 1
ADAM22 at chr7:87401638-87664385 - (NM_021722) ADAM metallopeptidase domain 22 isoform 2


Human chr8:39,720,412-39,814,936 - UCSC Genome Browser v223
Home Genomes Blat Tables Gene Sorter PCR DNA Convert Ensembl NCBI PDF/PS Session Help
UCSC Genome Browser on Human Mar. 2006 Assembly (hg18)
intron
exon
Click on a feature for details. Click or drag in the base position track to zoom in. Click gray/blue bars on left for track options and descriptions.
Mapping and Sequencing Tracks
Base Position Chromosome Band STS Markers FISH Clones Recomb Rate Map Contigs


UCSC Gene details
Human Gene ADAM2 (uc003xnj.1) Description and Page Index
Description: ADAM metallopeptidase domain 2 proprotein
RefSeq Summary (NM_001464): This gene encodes a member of the ADAM (a disintegrin and metalloprotease domain) family. Members of this family are membrane-anchored proteins structurally related to snake venom disintegrins, and have been implicated in a variety of biological processes involving cell-cell and cell-matrix interactions, including fertilization, muscle development, and neurogenesis. This member is a subunit of an integral sperm membrane glycoprotein called fertilin, which plays an important role in sperm-egg interactions. [provided by RefSeq].
Strand: - Genomic Size: 94525 Exon Count: 21 Coding Exon Count: 20
Page Index | Sequence and Links | UniProtKB Comments | CTD | Microarray | RNA Structure
Protein Structure | Other Species | GO Annotations | mRNA Descriptions | Other Names | Model Information
Methods
Sequence and Links to Tools and Databases
Genomic Sequence (chr8:39,720,412-39,814,936) | mRNA (may differ from genome) | Protein (735 aa)
Gene Sorter | Genome Browser | Protein FASTA | Proteome Browser | VisiGene | Table Schema
CGAP | Ensembl | Entrez Gene | ExonPrimer | GeneCards | GeneNetwork
Gepis Tissue | H-INV | HGNC | HPRD | Jackson Lab | OMIM
PubMed | Stanford SOURCE | Treefam | UniProtKB | User annotations
Comments and Description Text from UniProtKB
ID: ADAM2_HUMAN
DESCRIPTION: RecName: Full=Disintegrin and metalloproteinase domain-containing protein 2; Short=ADAM 2; AltName: Full=Fertilin subunit beta; AltName: Full=PH-30; Short=PH30; AltName: Full=PH30-beta; AltName: Full=Cancer/testis antigen 15; Short=CT15; Flags: Precursor;
FUNCTION: Sperm surface membrane protein that may be involved in sperm-egg plasma membrane adhesion and fusion during fertilization. Could have a direct role in sperm-zona binding or migration of sperm from the uterus into the oviduct. Interactions with egg membrane could be mediated via binding between its disintegrin-like domain to one or more integrins receptors on the egg. This is a non catalytic metalloprotease-like protein.
SUBCELLULAR LOCATION: Membrane; Single-pass type I membrane protein.
TISSUE SPECIFICITY: Expressed specifically in spermatogenic cells in the seminiferous cells. Not detected in fetal tissues.
DOMAIN: A tripeptide motif (FEE) within disintegrin-like domain could be involved in the binding to egg integrin receptor and thus could mediate sperm/egg binding.

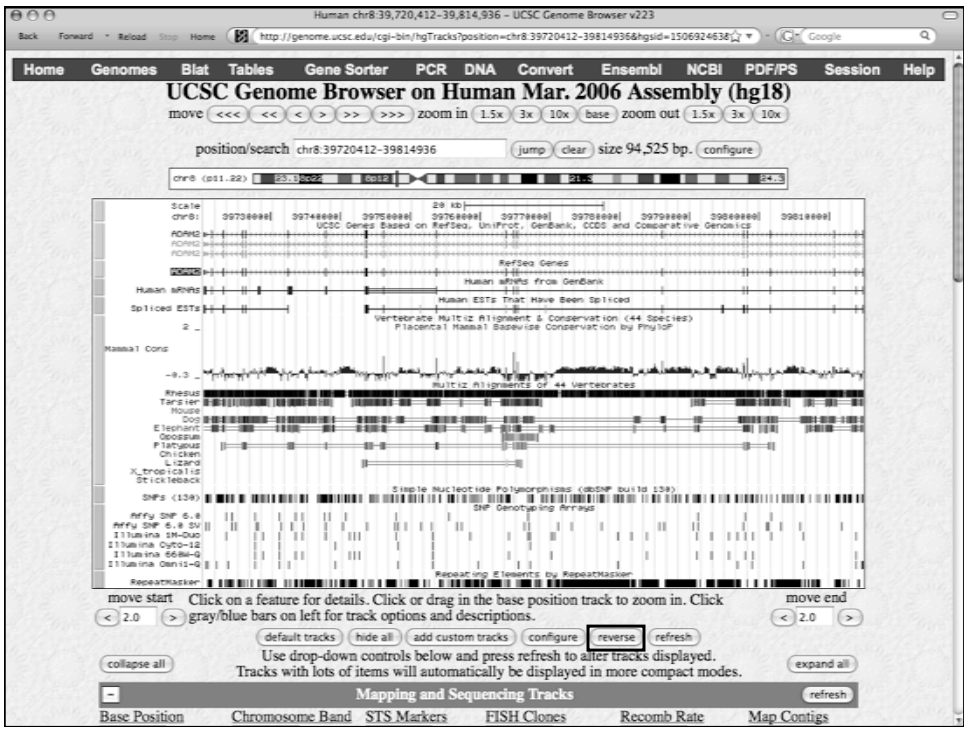

Human Gene ADAM2 (uc003xnj.1) Description and Page Index
UCSC Gene details
Affymetrix All Exon Microarrays
breast
cerebellum
heart
kidney
liver
muscle
pancreas
prostate
spleen
testes
thyroid
Ratios
mRNA Secondary Structure of 3' and 5' UTRs
Region Fold Energy Bases Energy/Base Display As
5' UTR -15.00 75 -0.200 Picture PostScript Text
3' UTR -72.02 359 -0.201 Picture PostScript Text
The RNAfold program from the Vienna RNA Package is used to perform the secondary structure predictions and folding calculations. The estimated folding energy is in kcal/mol. The more negative the energy, the more secondary structure the RNA is likely to have.
Protein Domain and Structure Information
InterPro Domains: Graphical view of domain structure
IPR006586 - ADAM_Cys-rich
IPR001762 - Blood-coag_inhib_Disintegrin
IPR018358 - Disintegrin_CS
IPR013032 - EGF-like_reg_CS
IPR013111 - EGF_extracell
IPR001590 - Peptidase_M12B
IPR002870 - Peptidase_M12B_N
Pfam Domains:
PF01421 - Reprolysin (M12B) family zinc metalloprotease
PF01562 - Reprolysin family propeptide
PF08516 - ADAM cysteine-rich
PF00200 - Disintegrin
PF07974 - EGF-like domain
SCOP Domains:

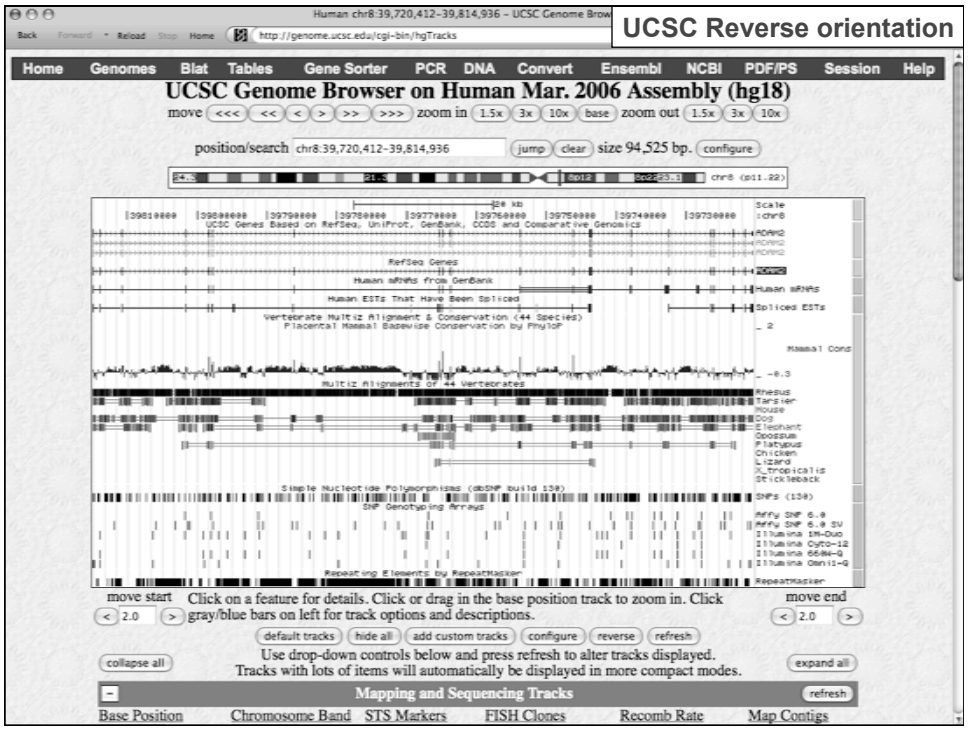

RefSeq Gene ADAM2
UCSC RefSeq Gene details
RefSeq: NM_001464.3 Status: Reviewed
Description: Homo sapiens ADAM metallopeptidase domain 2 (ADAM2), mRNA.
CCDS: CCDS34884.1
CDS: 3' complete
OMIM: 601533
Entrez Gene: 2515
PubMed on Gene: ADAM2
PubMed on Product: ADAM metallopeptidase domain 2 proprotein
GeneCards: ADAM2
AceView: ADAM2
Stanford SOURCE: NM_001464
CDS FASTA alignment from multiple alignment: NM_001464
Summary of ADAM2
This gene encodes a member of the ADAM (a disintegrin and metalloprotease domain) family. Members of this family are membrane-anchored proteins structurally related to snake venom disintegrins, and have been implicated in a variety of biological processes involving cell-cell and cell-matrix interactions, including fertilization, muscle development, and neurogenesis. This member is a subunit of an integral sperm membrane glycoprotein called fertilin, which plays an important role in sperm-egg interactions. [provided by RefSeq].
mRNA/Genomic Alignments
SIZE IDENTITY CHROMOSOME STRAND START END QUERY START END TOTAL
2642 100.0% 8 - 39720412 39814936 NM_001464 1 2642 2657
Links to sequence:
Predicted Protein
mRNA Sequence
Genomic Sequence from assembly
click

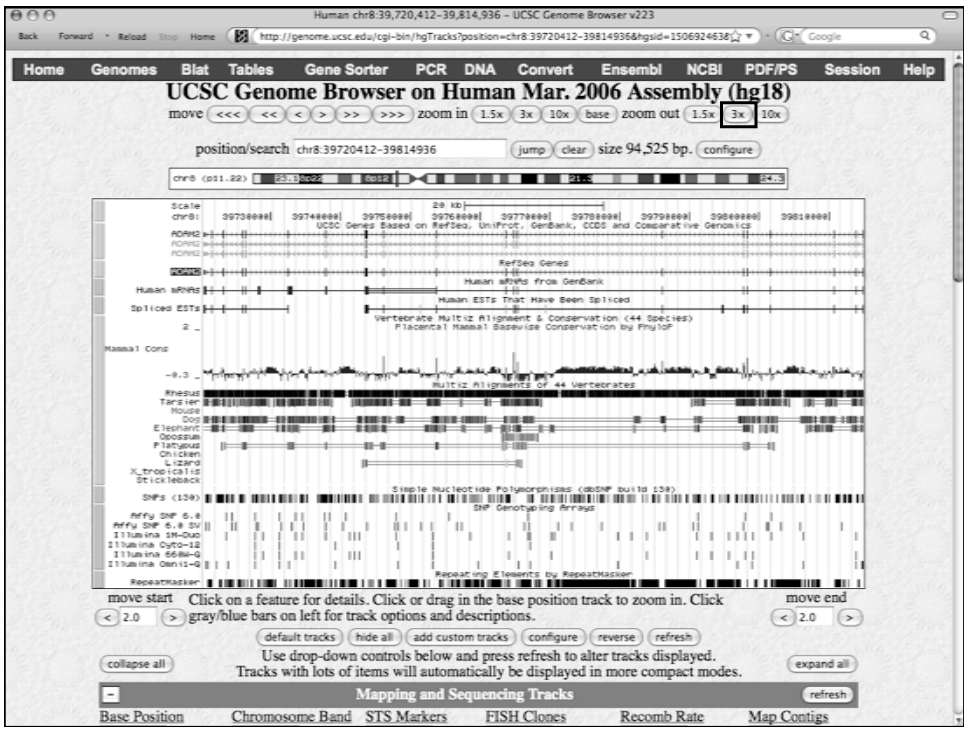


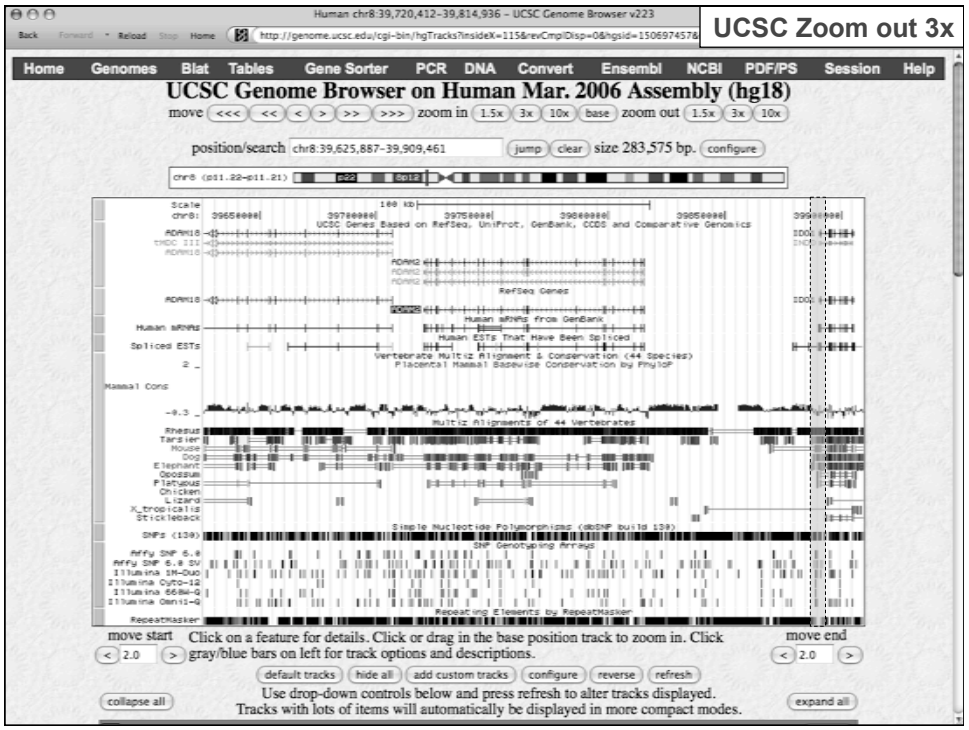
UCSC

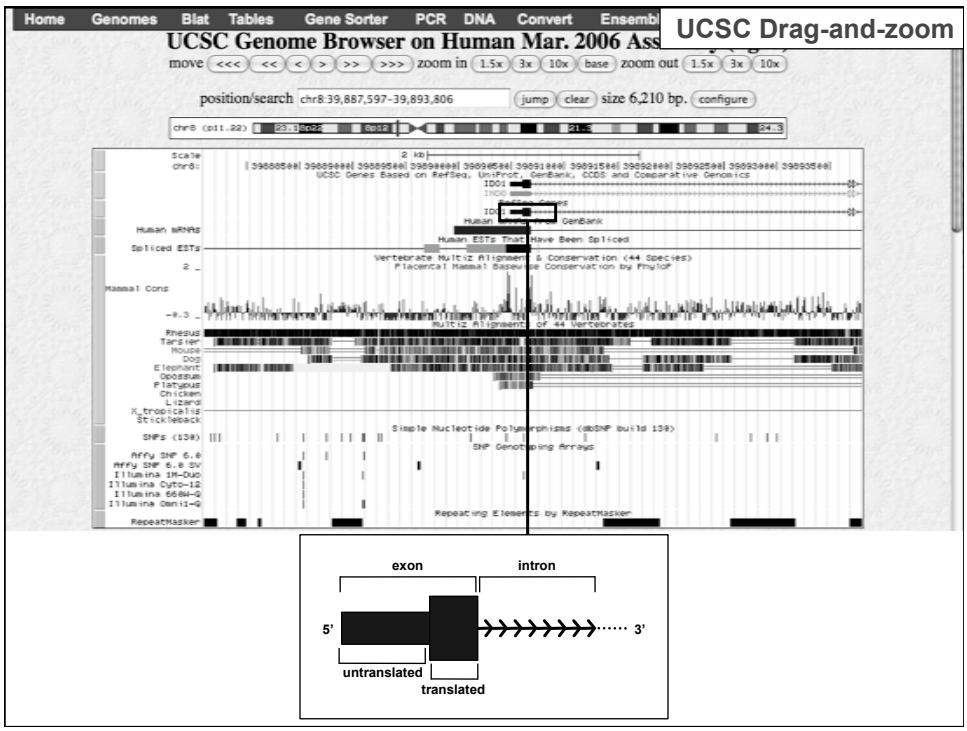
Navigating around the Genome Browser


Human chr8:39,720,412-39,814,936 - UCSC Genome Browser v223
Home Genomes Blat Tables Gene Sorter PCR DNA Convert Ensembl NCBI PDF/PS Session Help
UCSC Genome Browser on Human Mar. 2006 Assembly (hg18)
move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x
position/search chr8:39720412-39814936 jump clear size 94,525 bp. configure
RefSeq Genes
Human mRNAs from GenBank
Human ESTs That Have Been Spliced
Vertebrate Multiz Alignment & Conservation (44 Species)
Placental Mammal Basewise Conservation by PhyloP
Multiz Alignments of 44 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP build 130)
SNP Genotyping Arrays
Repeating Elements by RepeatMasker
move start Click on a feature for details. Click or drag in the base position track to zoom in. Click gray/blue bars on left for track options and descriptions. move end
default tracks hide all add custom tracks configure reverse 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.
collapse all expand all
Mapping and Sequencing Tracks refresh
Base Position Chromosome Band STS Markers FISH Clones Recomb Rate Map Contigs


UCSC Reverse orientation
Home Genomes Blat Tables Gene Sorter PCR DNA Convert Ensembl NCBI PDF/PS Session Help
UCSC Genome Browser on Human Mar. 2006 Assembly (hg18)
position/search chr8:39,720,412-39,814,936 jump clear size 94,525 bp. configure
move start Click on a feature for details. Click or drag in the base position track to zoom in. Click gray/blue bars on left for track options and descriptions. move end
default tracks hide all add custom tracks configure reverse 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.
collapse all expand all
Mapping and Sequencing Tracks refresh
Base Position Chromosome Band STS Markers FISH Clones Recomb Rate Map Contigs


Human chr8:39,720,412-39,814,936 – UCSC Genome Browser v223
Home Genomes Blat Tables Gene Sorter PCR DNA Convert Ensembl NCBI PDF/PS Session Help
UCSC Genome Browser on Human Mar. 2006 Assembly (hg18)
move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x
position/search chr8:39720412-39814936 jump clear size 94,525 bp. configure
RefSeq Genes
Human mRNAs from GenBank
Human ESTs That Have Been Spliced
Vertebrate Multiz Alignment & Conservation (44 Species)
Placental Mammal Basewise Conservation by PhyloP
Multiz Alignments of 44 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP build 130)
SNP Genotyping Arrays
Repeating Elements by RepeatMasker
move start Click on a feature for details. Click or drag in the base position track to zoom in. Click gray/blue bars on left for track options and descriptions. move end
default tracks hide all add custom tracks configure reverse 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.
collapse all expand all
Mapping and Sequencing Tracks refresh
Base Position Chromosome Band STS Markers FISH Clones Recomb Rate Map Contigs

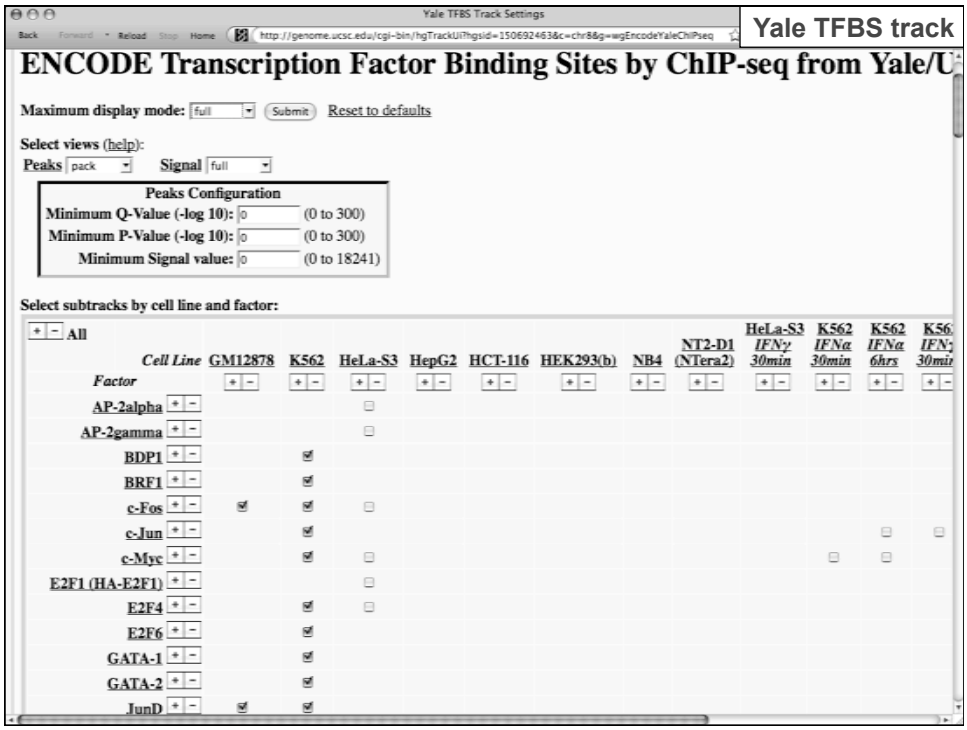

UCSC Zoom out 3x
Human chr8:39,720,412-39,814,936 – UCSC Genome Browser v223
Home Genomes Blat Tables Gene Sorter PCR DNA Convert Ensembl NCBI PDF/PS Session Help
UCSC Genome Browser on Human Mar. 2006 Assembly (hg18)
move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x
position/search chr8:39,625,887-39,909,461 jump clear size 283,575 bp. configure
RefSeq Genes
Human mRNAs from GenBank
Human ESTs That Have Been Spliced
Vertebrate Multiz Alignment & Conservation (44 Species)
Placental Mammal Basewise Conservation by PhyloP
Multiz Alignments of 44 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP build 130)
SNP Genotyping Arrays
Repeating Elements by RepeatMasker
move start Click on a feature for details. Click or drag in the base position track to zoom in. Click gray/blue bars on left for track options and descriptions. move end
default tracks hide all add custom tracks configure reverse 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.
collapse all expand all

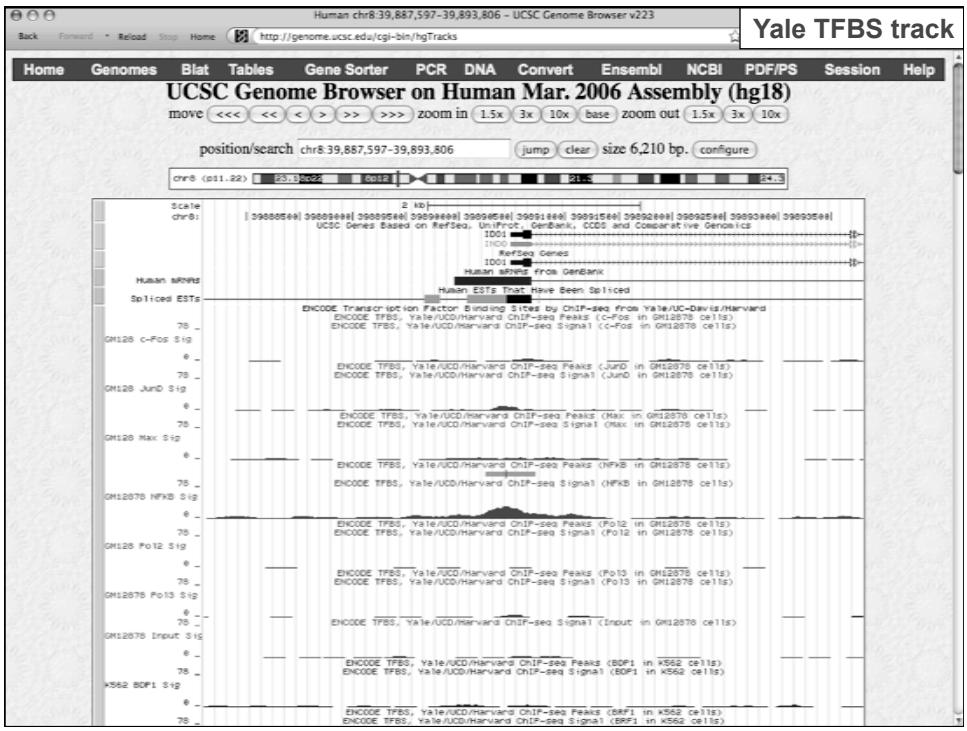


UCSC

Add a track to the Genome Browser


UCSC track selection
Genes and Gene Prediction Tracks
mRNA and EST Tracks
Expression
Regulation
Comparative Genomics
click
Yale TFBS
hide
dense
squish
pack
full

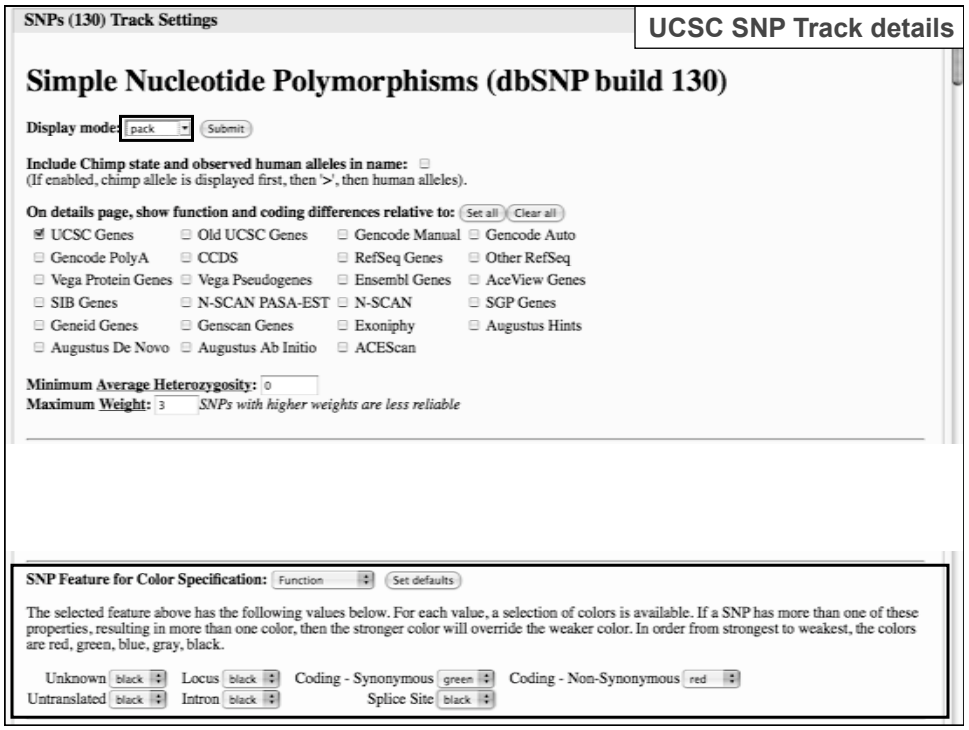

Yale TFBS track
ENCODE Transcription Factor Binding Sites by ChIP-seq from Yale/U
Maximum display mode: full
Submit
Reset to defaults
Select views (help):
Peaks pack
Signal full
Peaks Configuration
Minimum Q-Value (-log 10): 0 (0 to 300)
Minimum P-Value (-log 10): 0 (0 to 300)
Minimum Signal value: 0 (0 to 18241)
Select subtracks by cell line and factor:
Cell Line
Factor
GM12878
K562
HeLa-S3
HepG2
HCT-116
HEK293(b)
NB4
NT2-D1 (NTera2)
HeLa-S3 IFNγ 30min
K562 IFNα 30min
K562 IFNα 6hrs
AP-2alpha
AP-2gamma
BDP1
BRF1
c-Fos
c-Jun
c-Myc
E2F1 (HA-E2F1)
E2F4
E2F6
GATA-1
GATA-2
JunD

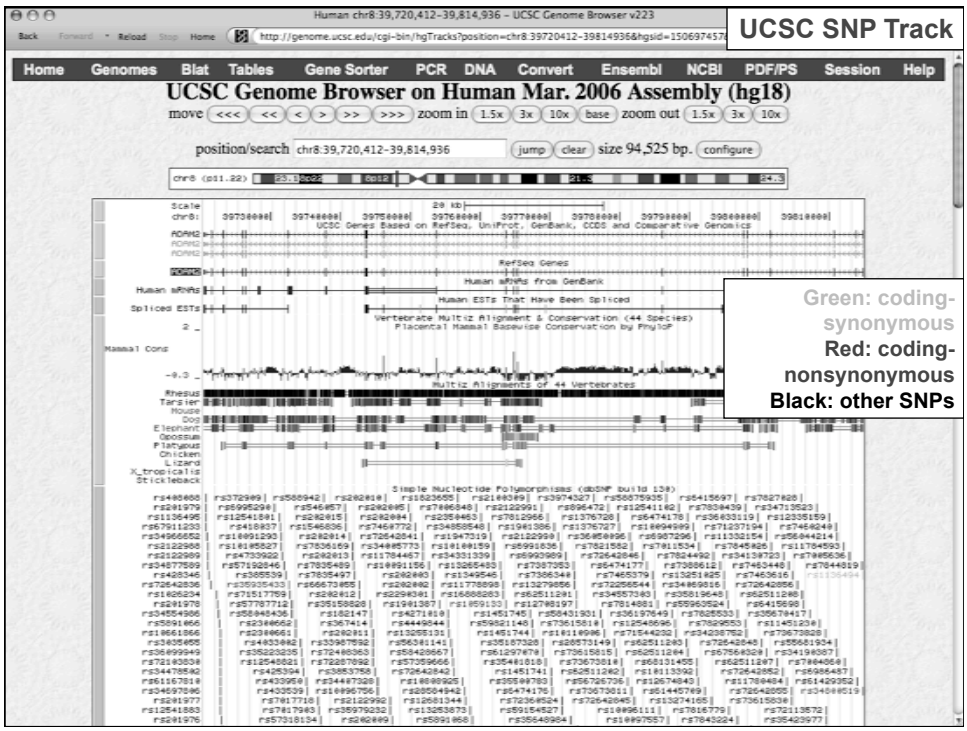


UCSC

Change the color of items in a track

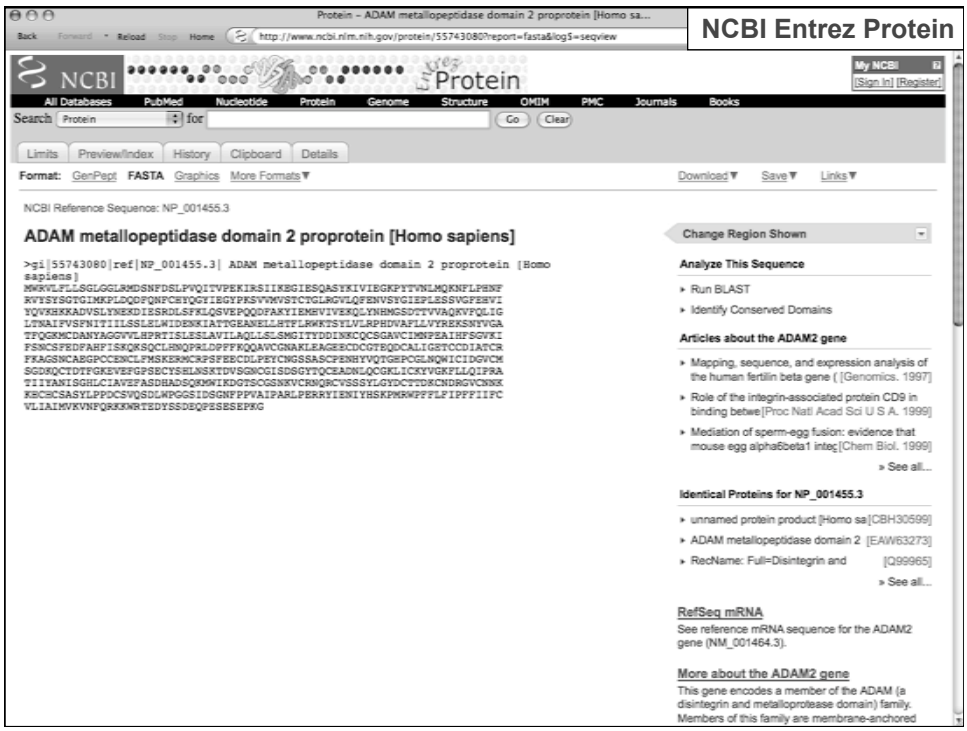

Human chr8:39,887,597-39,893,806 – UCSC Genome Browser v223
Lamprey Net
Medaka Chain
Medaka Net
Stickleback Chain
Stickleback Net
Fugu Chain
Fugu Net
Tetraodon Chain
Tetraodon Net
Tetraodon Ecores
Zebrafish Chain
Zebrafish Net
X. tropicalis Chain
X. tropicalis Net
Zebra finch Chain
Zebra finch Net
Lizard Chain
Lizard Net
Platypus Chain
Platypus Net
Chimp Chain/Net
Orangutan Chain/Net
Rhesus Chain/Net
Guinea Pig Chain
Guinea Pig Net
Rat Chain
Rat Net
Marmoset Chain/Net
Mouse Chain
Mouse Net
Dog Chain/Net
Cat Chain/Net
Horse Chain/Net
Cow Chain/Net
Opossum Chain/Net
Chicken Chain/Net
Variation and Repeats
refresh
click
SNPs (130)
SNPs (129)
SNPs (128)
SNPs (126)
SNP Arrays
HGDP Allele Freq
HGDP Smoothd FST
HGDP Hetrzygsty
HGDP iHS
HGDP XP-EHH
HapMap SNPs
HapMap LD Phased
Tajima's D SNPs
Tajima's D
DGV Struct Var
HGSV Discordant
Segmental Dups
Structural Var
Exapted Repeats
RepeatMasker
RepMask 3.2.7
Interrupted Rpts
Intr Rpts 3.2.7
Simple Repeats
Microsatellite
Self Chain
Genome Variants
Pilot ENCODE Regions and Genes
Pilot ENCODE Transcription
Pilot ENCODE Chromatin Immunoprecipitation
Pilot ENCODE Chromatin Structure
Pilot ENCODE Comparative Genomics and Variation

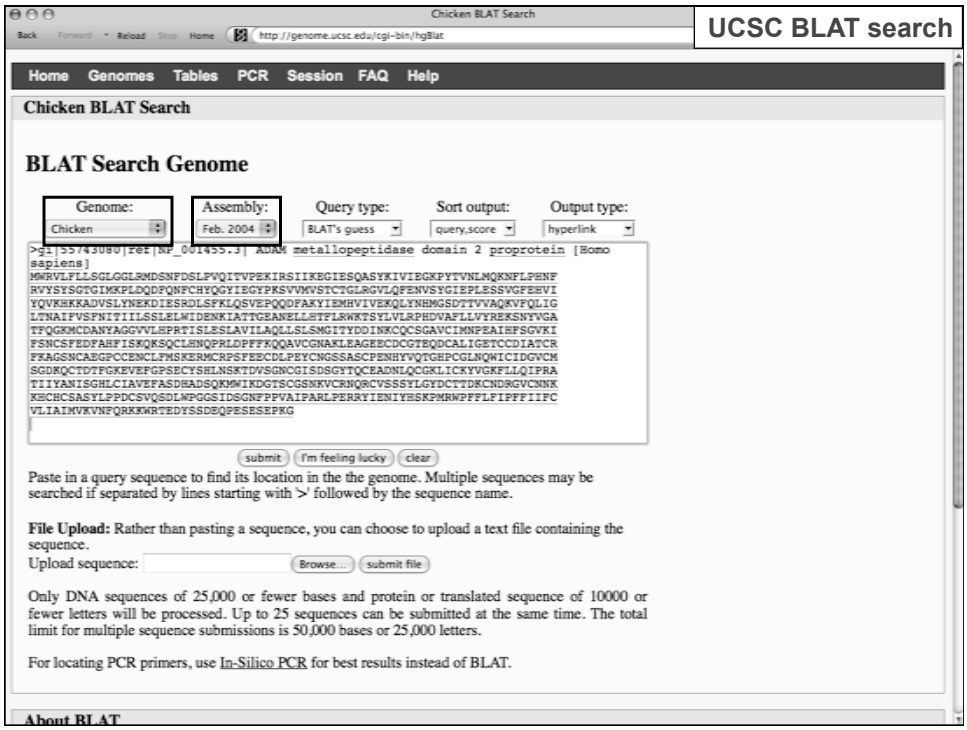

SNPs (130) Track Settings
UCSC SNP Track details
Simple Nucleotide Polymorphisms (dbSNP build 130)
Display mode: pack Submit
Include Chimp state and observed human alleles in name:
(If enabled, chimp allele is displayed first, then '>', then human alleles).
On details page, show function and coding differences relative to: Set all Clear all
UCSC Genes
Old UCSC Genes
Gencode Manual
Gencode Auto
Gencode PolyA
CCDS
RefSeq Genes
Other RefSeq
Vega Protein Genes
Vega Pseudogenes
Ensembl Genes
AceView Genes
SIB Genes
N-SCAN PASA-EST
N-SCAN
SGP Genes
Geneid Genes
Genscan Genes
Exoniphy
Augustus Hints
Augustus De Novo
Augustus Ab Initio
ACEScan
Minimum Average Heterozygosity:
Maximum Weight: SNPs with higher weights are less reliable
SNP Feature for Color Specification: Function Set defaults
The selected feature above has the following values below. For each value, a selection of colors is available. If a SNP has more than one of these properties, resulting in more than one color, then the stronger color will override the weaker color. In order from strongest to weakest, the colors are red, green, blue, gray, black.
Unknown black
Locus black
Coding - Synonymous green
Coding - Non-Synonymous red
Untranslated black
Intron black
Splice Site black

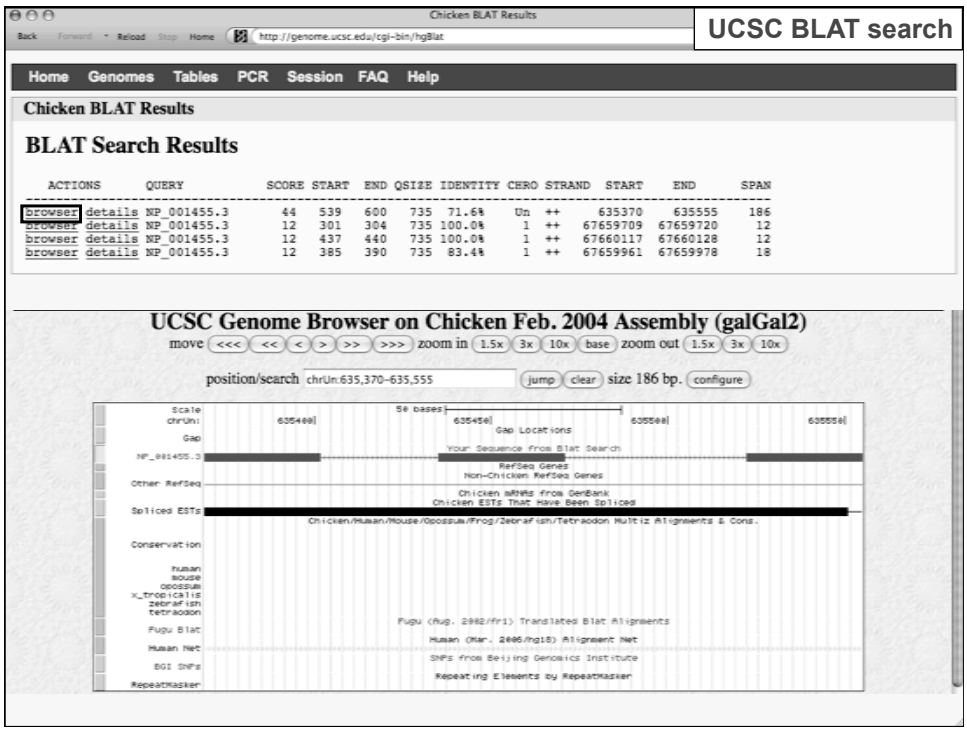


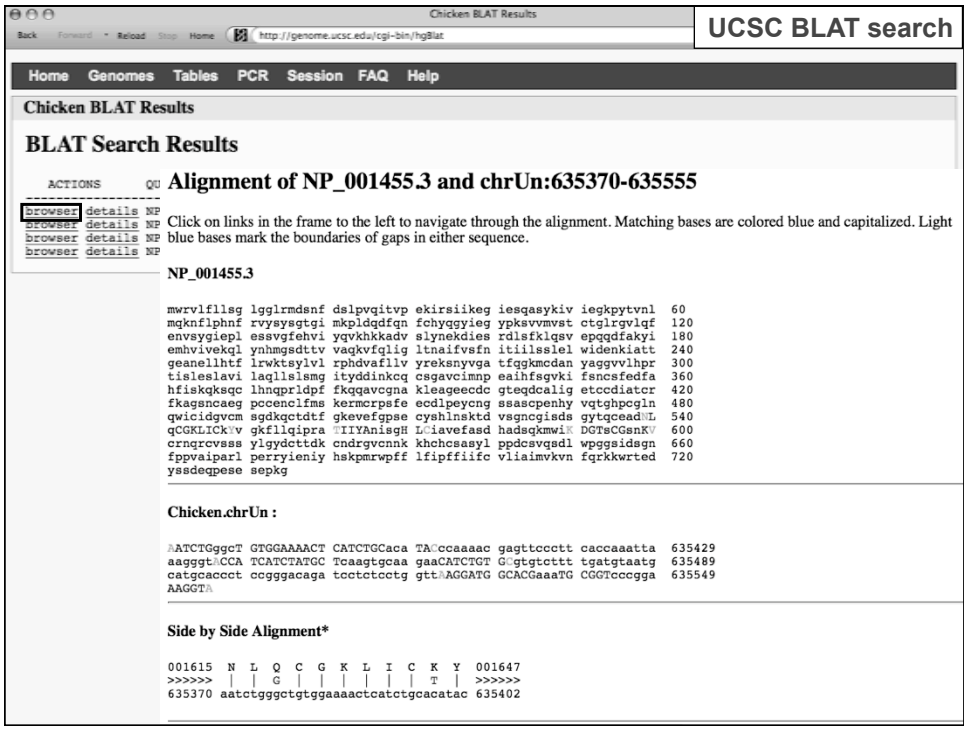
UCSC

Find a chicken homolog of a human protein


NCBI Entrez Protein
Protein – ADAM metallopeptidase domain 2 proprotein [Homo sa...
http://www.ncbi.nlm.nih.gov/protein/55743080?report=fasta&log$=seqview
NCBI
Protein
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search Protein for Go Clear
Limits Preview/Index History Clipboard Details
Format: GenPept FASTA Graphics More Formats
Download Save Links
NCBI Reference Sequence: NP_001455.3
ADAM metallopeptidase domain 2 proprotein [Homo sapiens]
>gi|55743080|ref|NP_001455.3| ADAM metallopeptidase domain 2 proprotein [Homo sapiens]
MWRVLFLLSGLGGLRMDSNFDSLPVQITVPEKIRSIIKEGIESQASYKIVIEGKPYTVNLMQKNFLPHNF
RVYSYSGTGIMKPLDQDFQNFCHYQGYIEGYPKSVVMVSTCTGLRGVLQFENVSYGIEPLSSSVGFEHVI
YQVKHKKADVSLYNEKDIESRDLSFKLQSVEPQQDFAKYIEMHVIVEKQLYNHMGSDTTVVAQKVFQLIG
LTNAIFVSFNITIILSSLELWIDENKIATTGEANELLSTFLRWKTSYLVLRPHDVAFLLVYREKSNYVGA
TFQGKMCDANYAGGVVLHPRTISLESLAVILAQLLSLSMGITYDDINKCQCSGAVCIMNPEAIEFSGVKI
FSNCSFEDFAHFISKQKSQCLHNQPRLDPFFKQQAVCGNAKLEAGEECDCGTEQDCALIGETCCDIATCR
FKAGSNCAEGPCCENCLFMSKERMCRPSFEECDLPEYCNGSSASCPENHYVQTGHPCGLNQWICIDGVCM
SGDSQCTDTFGKEVEFGPSECYSHLNSKTDVSGNCGISDSGYTQCEADNLQCGKLICKYVGKFLLQIPRA
TIIYANISGHLCIAVEFASDHADSQKMWIKDGTSCGSNKVCRNQRCVSSSYLGYDCTTDKCNDRGVCNNK
KHCHCSASYLPPDCSVQSDLWPGGSIDSGNFPPVAIPARLPERRYIENIYHSKPMRWPFFLFIPFFIIFC
VLIAIMVKVNFQRKKWRTEDYSSDEQPESESEPKG
Change Region Shown
Analyze This Sequence
Run BLAST
Identify Conserved Domains
Articles about the ADAM2 gene
Mapping, sequence, and expression analysis of the human fertilin beta gene ([Genomics. 1997]
Role of the integrin-associated protein CD9 in binding betwe [Proc Natl Acad Sci U S A. 1999]
Mediation of sperm-egg fusion: evidence that mouse egg alpha6beta1 intec [Chem Biol. 1999]
» See all...
Identical Proteins for NP_001455.3
unnamed protein product [Homo sa [CBH30599]
ADAM metallopeptidase domain 2 [EAW63273]
RecName: Full=Disintegrin and [Q99965]
» See all...
RefSeq mRNA
See reference mRNA sequence for the ADAM2 gene (NM_001464.3).
More about the ADAM2 gene
This gene encodes a member of the ADAM (a disintegrin and metalloprotease domain) family. Members of this family are membrane-anchored


UCSC BLAT search
Chicken BLAT Search
http://genome.ucsc.edu/cgi-bin/hgBlat
Home Genomes Tables PCR Session FAQ Help
Chicken BLAT Search
BLAT Search Genome
Genome: Chicken
Assembly: Feb. 2004
Query type: BLAT's guess
Sort output: query,score
Output type: hyperlink
>gi|55743080|ref|NP_001455.3| ADAM metallopeptidase domain 2 proprotein [Homo sapiens]
MWRVLFLLSGLGGLRMDSNFDSLPVQITVPEKIRSIIKEGIESQASYKIVIEGKPYTVNLMQKNFLPHNF
RVYSYSGTGIMKPLDQDFQNFCHYQGYIEGYPKSVVMVSTCTGLRGVLQFENVSYGIEPLSSSVGFEHVI
YQVKHKKADVSLYNEKDIESRDLSFKLQSVEPQQDFAKYIEMHVIVEKQLYNHMGSDTTVVAQKVFQLIG
LTNAIFVSFNITIILSSLELWIDENKIATTGEANELLSTFLRWKTSYLVLRPHDVAFLLVYREKSNYVGA
TFQGKMCDANYAGGVVLHPRTISLESLAVILAQLLSLSMGITYDDINKCQCSGAVCIMNPEAIEFSGVKI
FSNCSFEDFAHFISKQKSQCLHNQPRLDPFFKQQAVCGNAKLEAGEECDCGTEQDCALIGETCCDIATCR
FKAGSNCAEGPCCENCLFMSKERMCRPSFEECDLPEYCNGSSASCPENHYVQTGHPCGLNQWICIDGVCM
SGDKQCTDTFGKEVEFGPSECYSHLNSKTDVSGNCGISDSGYTQCEADNLQCGKLICKYVGKFLLQIPRA
TIIYANISGHLCIAVEFASDHADSQKMWIKDGTSCGSNKVCRNQRCVSSSYLGYDCTTDKCNDRGVCNNK
KHCHCSASYLPPDCSVQSDLWPGGSIDSGNFPPVAIPARLPERRYIENIYHSKPMRWPFFLFIPFFIIFC
VLIAIMVKVNFQRKKWRTEDYSSDEQPESESEPKG
submit I'm feeling lucky clear
Paste in a query sequence to find its location in the the genome. Multiple sequences may be searched if separated by lines starting with '>' followed by the sequence name.
File Upload: Rather than pasting a sequence, you can choose to upload a text file containing the sequence.
Upload sequence: Browse... submit file
Only DNA sequences of 25,000 or fewer bases and protein or translated sequence of 10000 or fewer letters will be processed. Up to 25 sequences can be submitted at the same time. The total limit for multiple sequence submissions is 50,000 bases or 25,000 letters.
For locating PCR primers, use In-Silico PCR for best results instead of BLAT.
About BLAT


Chicken BLAT Results
http://genome.ucsc.edu/cgi-bin/hgBlat
UCSC BLAT search
Home Genomes Tables PCR Session FAQ Help
Chicken BLAT Results
BLAT Search Results
ACTIONS QUERY SCORE START END QSIZE IDENTITY CHRO STRAND START END SPAN
browser details NP_001455.3 44 539 600 735 71.6% Un ++ 635370 635555 186
browser details NP_001455.3 12 301 304 735 100.0% 1 ++ 67659709 67659720 12
browser details NP_001455.3 12 437 440 735 100.0% 1 ++ 67660117 67660128 12
browser details NP_001455.3 12 385 390 735 83.4% 1 ++ 67659961 67659978 18
UCSC Genome Browser on Chicken Feb. 2004 Assembly (galGal2)
move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x
position/search chrUn:635,370-635,555 jump clear size 186 bp. configure
Gap Locations
Your Sequence from Blat Search
RefSeq Genes
Non-Chicken RefSeq Genes
Chicken mRNAs from GenBank
Chicken ESTs That Have Been Spliced
Chicken/Human/Mouse/Opossum/Frog/Zebrafish/Tetraodon Multiz Alignments & Cons.
Fugu (Aug. 2002/fr1) Translated Blat Alignments
Human (Mar. 2006/hg18) Alignment Net
SNPs from Beijing Genomics Institute
Repeating Elements by RepeatMasker


Chicken BLAT Results
http://genome.ucsc.edu/cgi-bin/hgBlat
UCSC BLAT search
Home Genomes Tables PCR Session FAQ Help
Chicken BLAT Results
BLAT Search Results
Alignment of NP_001455.3 and chrUn:635370-635555
Click on links in the frame to the left to navigate through the alignment. Matching bases are colored blue and capitalized. Light blue bases mark the boundaries of gaps in either sequence.
NP_001455.3
mwrvlfllsg lgglrmdsnf dslpvqitvp ekirsiikeg iesqasykiv iegkpytvnl 60
mqknflphnf rvysysgtgi mkpldqdfqn fchyqgyieg ypksvvmvst ctglrgvlgf 120
envsygiepl essvgfehvi yqvkhkkadv slynekdies rdlsfklqsv epqqdfakyi 180
emhvivekql ynhmgsdttv vaqkvfqlig ltnaifvsfn itiilssiel widenkiatt 240
geanellhtf lrwktsylvl rphdvafllv yreksnyvga tfqgkmcdan yagqvvlhpr 300
tisleslavi laqllslsmg ityddinkcq csgavcimnp eaihfsgvki fsncsfedfa 360
hfiskqksqc lhnqprldpf fkqqavcgna kleageecdc gteqdcalig etccdiatcr 420
fkagsncaeg pccenclfms kermcrpsfe ecdlpeycng ssascpenhy vqtghpcgln 480
qwicidgvcm sgdkqctdtf gkevefgpse cyshlnsktd vsgncgisds gytqceadNL 540
qCGKLICkYv gkfllqipra TIIYAnisgH LCiavefasd hadsqkmwiK DGTsCGsnKV 600
crnqrcvsss ylgydcttdk cndrgvcnnk khchcsasyl ppdcsvqsdl wpggsidsgn 660
fppvaiparl perryieniy hskpmrwpff lfipffiifc vliaimvkvn fqrkkwrted 720
yssdeqpese sepkg
Chicken.chrUn :
AATCTGggcT GTGGAAAACT CATCTGCaca TACccaaaac gagttccctt caccaaatta 635429
aagggtACCA TCATCTATGC Tcaagtgcaa gaaCATCTGT GCgtgtcttt tgatgtaatg 635489
catgcaccct ccgggacaga tcctctcctg gttAAGGATG GCACGaaaTG CGGTcccgga 635549
AAGGTA
Side by Side Alignment*
001615 N L Q C G K L I C K Y 001647
>>>>>> | | G | | | | | | T | >>>>>>
635370 aatctgggctgtggaaaactcatctgcacatac 635402

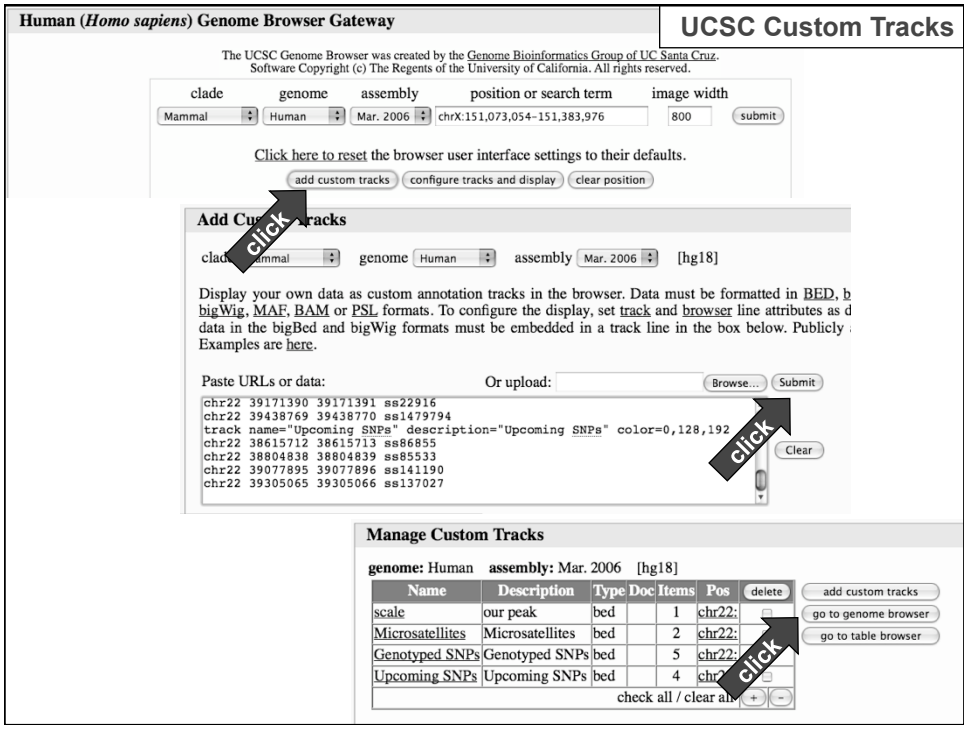
UCSC

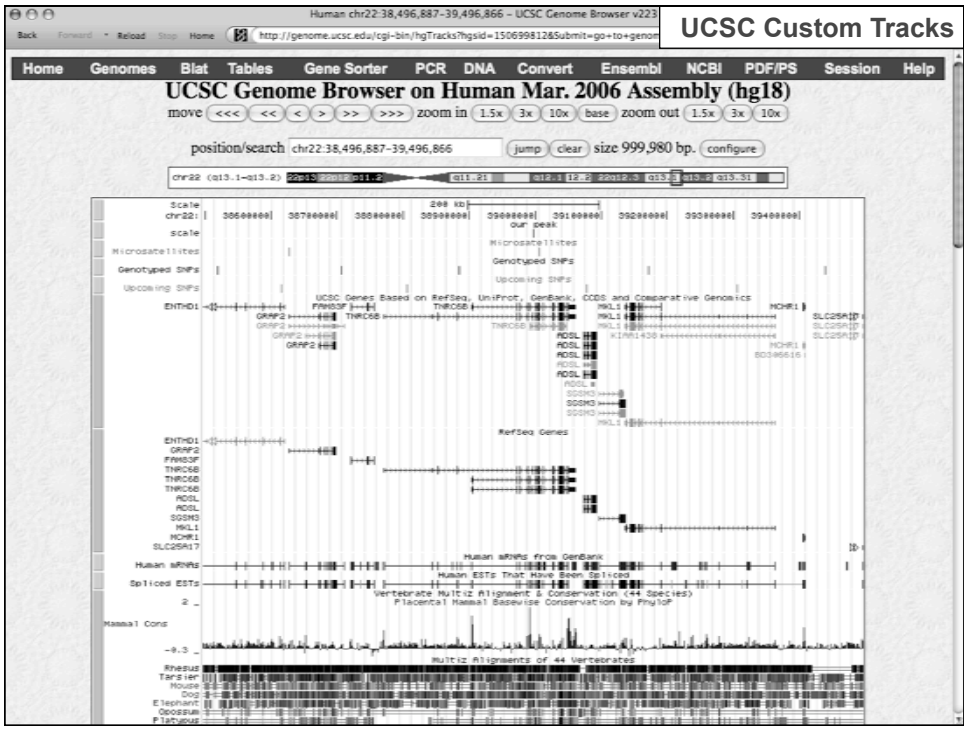
Add your own custom tracks

UCSC custom track format

```
browser position chr22:38496887-39496866
browser hide cytoBand
browser hide stsMap
browser hide gap
browser hide clonePos
browser full refGene
browser dense mrna
track name="scale" description="our peak"
chr22 38996887 38996888 peak
track name="Microsatellites" description="Microsatellites" color=0,128,0
chr22 38627059 38627060 D22S276
chr22 39005417 39005418 D22S307
track name="Genotyped SNPs" description="Genotyped SNPs" color=0,0,255
chr22 38518342 38518343 ss146131
chr22 38705963 38705964 ss2941443
chr22 38884157 38884158 ss141110
chr22 39171390 39171391 ss22916
chr22 39438769 39438770 ss1479794
track name="Upcoming SNPs" description="Upcoming SNPs" color=0,128,192
chr22 38615712 38615713 ss86855
chr22 38804838 38804839 ss85533
chr22 39077895 39077896 ss141190
chr22 39305065 39305066 ss137027
```

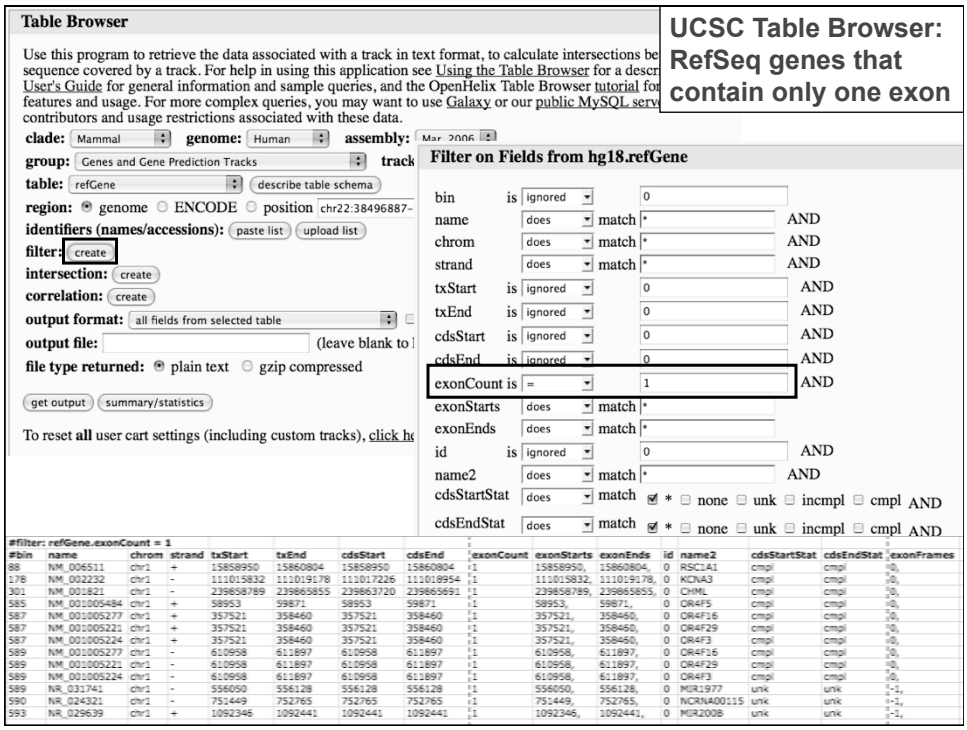
http://research.nhgri.nih.gov/teaching/custom_tracks.shtml


Human (*Homo sapiens*) Genome Browser Gateway
UCSC Custom Tracks
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
Mammal
Human
Mar. 2006
chrX:151,073,054-151,383,976
800
submit
Click here to reset the browser user interface settings to their defaults.
add custom tracks
configure tracks and display
clear position
click
Add Custom Tracks
genome
Human
assembly
Mar. 2006
[hg18]
Display your own data as custom annotation tracks in the browser. Data must be formatted in BED, bigWig, MAF, BAM or PSL formats. To configure the display, set track and browser line attributes as data in the bigBed and bigWig formats must be embedded in a track line in the box below. Publicly Examples are here.
Paste URLs or data:
Or upload:
Browse...
Submit
chr22 39171390 39171391 ss22916
chr22 39438769 39438770 ss1479794
track name="Upcoming SNPs" description="Upcoming SNPs" color=0,128,192
chr22 38615712 38615713 ss86855
chr22 38804838 38804839 ss85533
chr22 39077895 39077896 ss141190
chr22 39305065 39305066 ss137027
click
Clear
Manage Custom Tracks
genome: Human assembly: Mar. 2006 [hg18]
Name
Description
Type
Doc
Items
Pos
delete
scale
our peak
bed
1
chr22:
Microsatellites
Microsatellites
bed
2
chr22:
Genotyped SNPs
Genotyped SNPs
bed
5
chr22:
Upcoming SNPs
Upcoming SNPs
bed
4
chr2
check all / clear all
add custom tracks
go to genome browser
go to table browser
click


UCSC Custom Tracks
Home
Genomes
Blat
Tables
Gene Sorter
PCR
DNA
Convert
Ensembl
NCBI
PDF/PS
Session
Help
UCSC Genome Browser on Human Mar. 2006 Assembly (hg18)
move
zoom in
zoom out
position/search
chr22:38,496,887-39,496,866
jump
clear
size 999,980 bp.
configure
our peak
Microsatellites
Genotyped SNPs
Upcoming SNPs
UCSC Genes Based on RefSeq, UniProt, GenBank, CCDS and Comparative Genomics
RefSeq Genes
Human mRNAs from GenBank
Spliced ESTs
Vertebrate Multiz Alignment & Conservation (44 Species)
Placental Mammal Basewise Conservation by PhyloP
Multiz Alignments of 44 Vertebrates

UCSC Table Browser

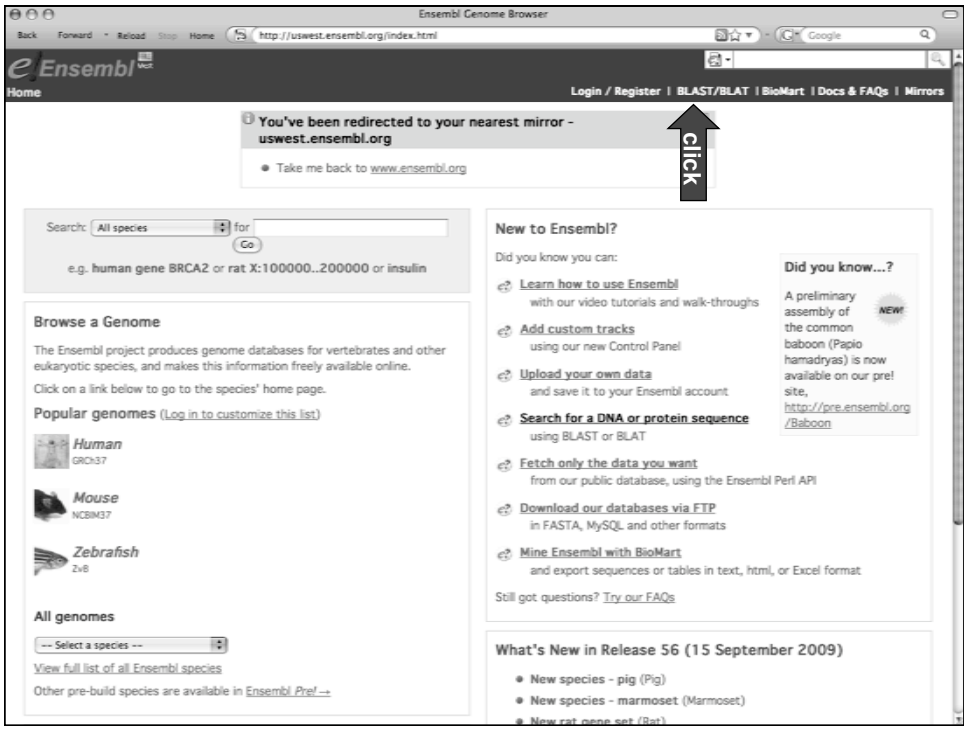
- Download track in text format
- Retrieve DNA sequence covered by a track
- Calculate intersections between tracks and view in the Genome Browser.
  - List all SNPs in a gene
- Filter track data based on certain criteria
  - Show all RefSeq genes that contain only one exon

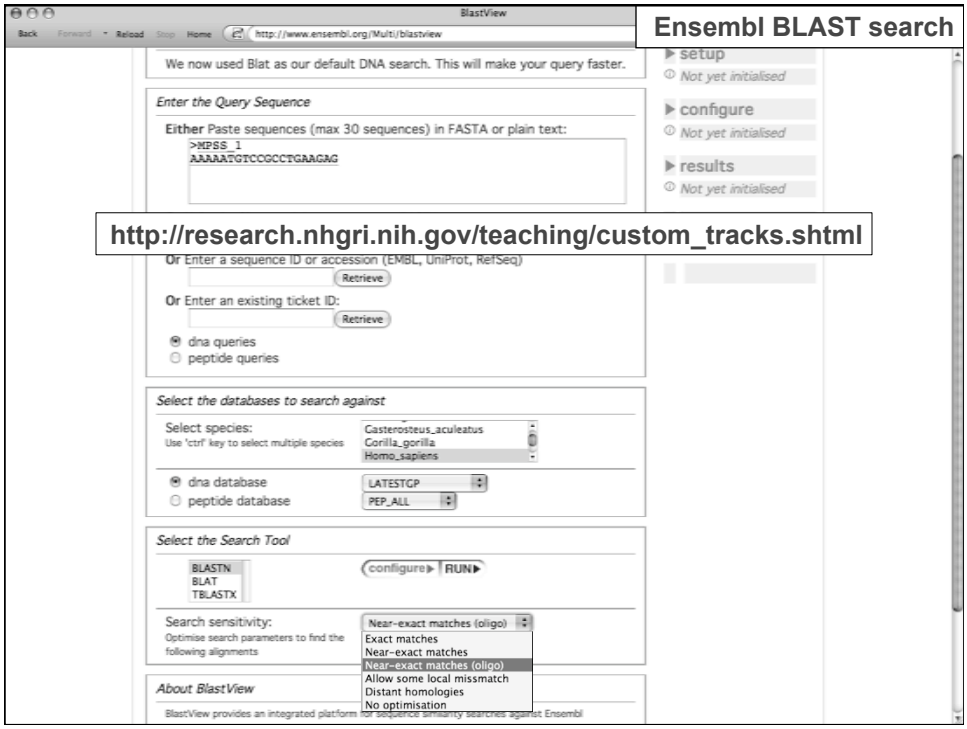


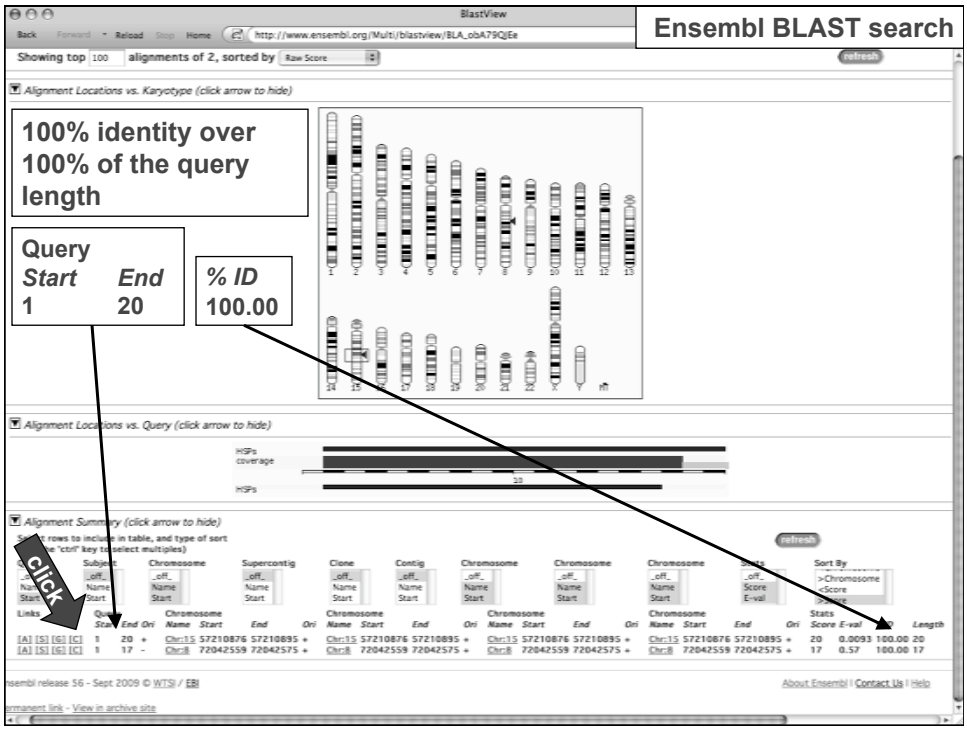
UCSC Table Browser: RefSeq genes that contain only one exon

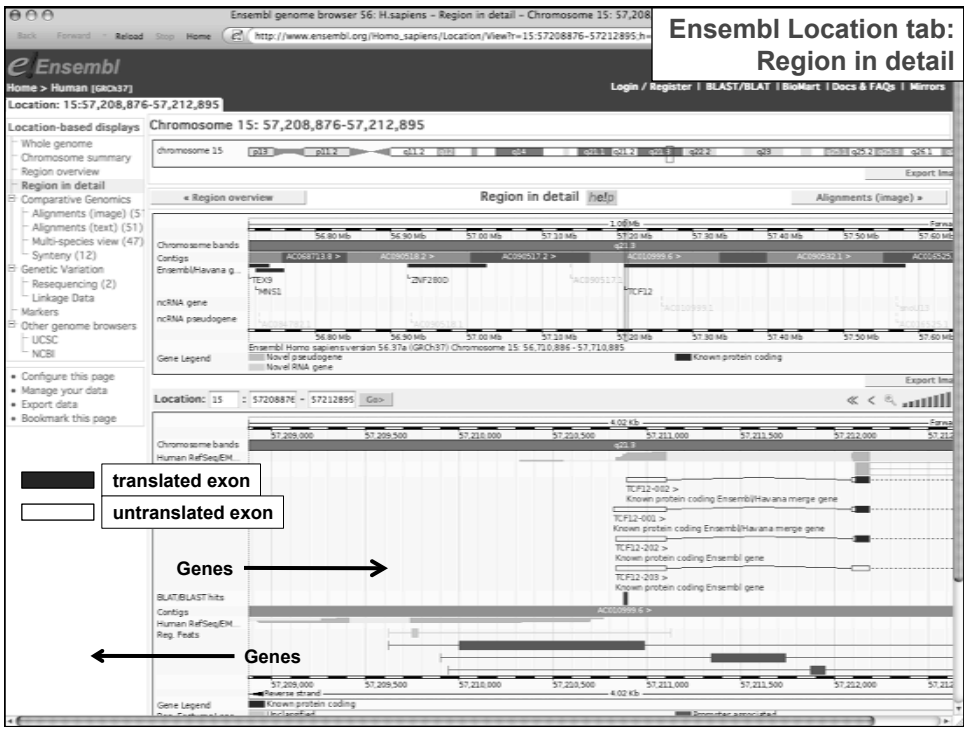
Ensembl

Identify genes that overlap with an oligo tag



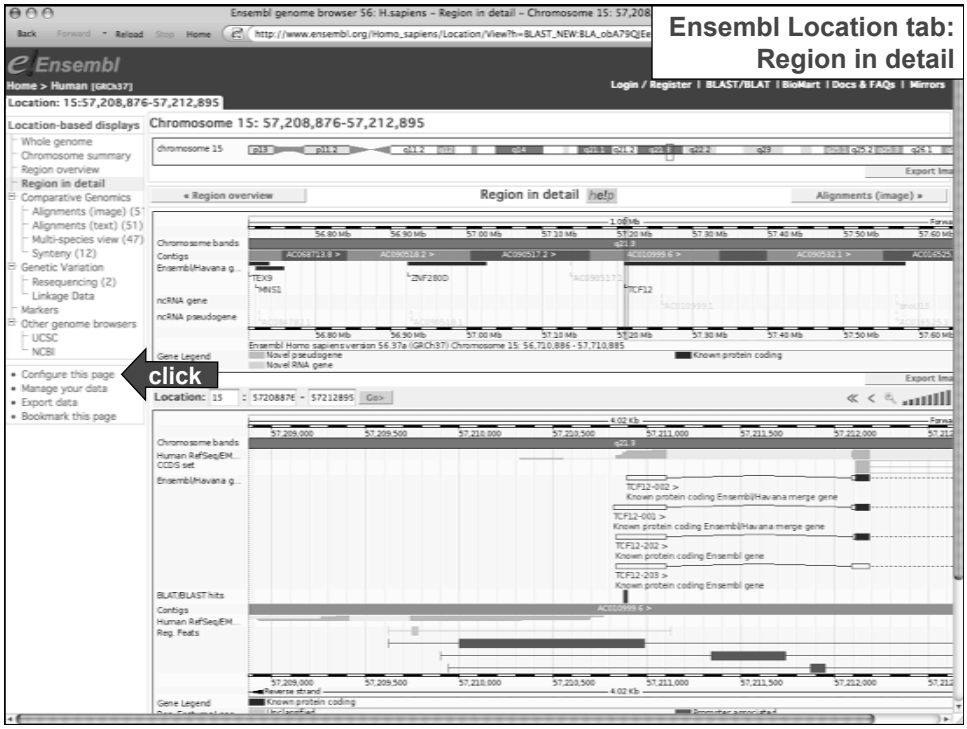

Ensembl BLAST search
http://research.nhgri.nih.gov/teaching/custom_tracks.shtml

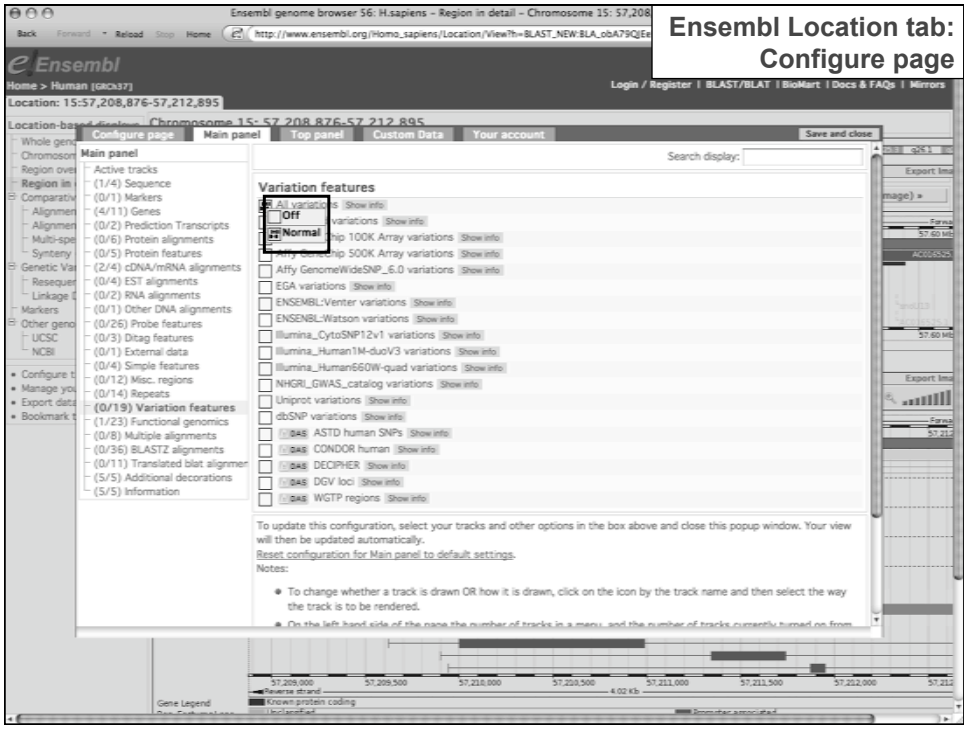

Ensembl BLAST search
100% identity over 100% of the query length
Query
Start
End
1
20
% ID
100.00
click

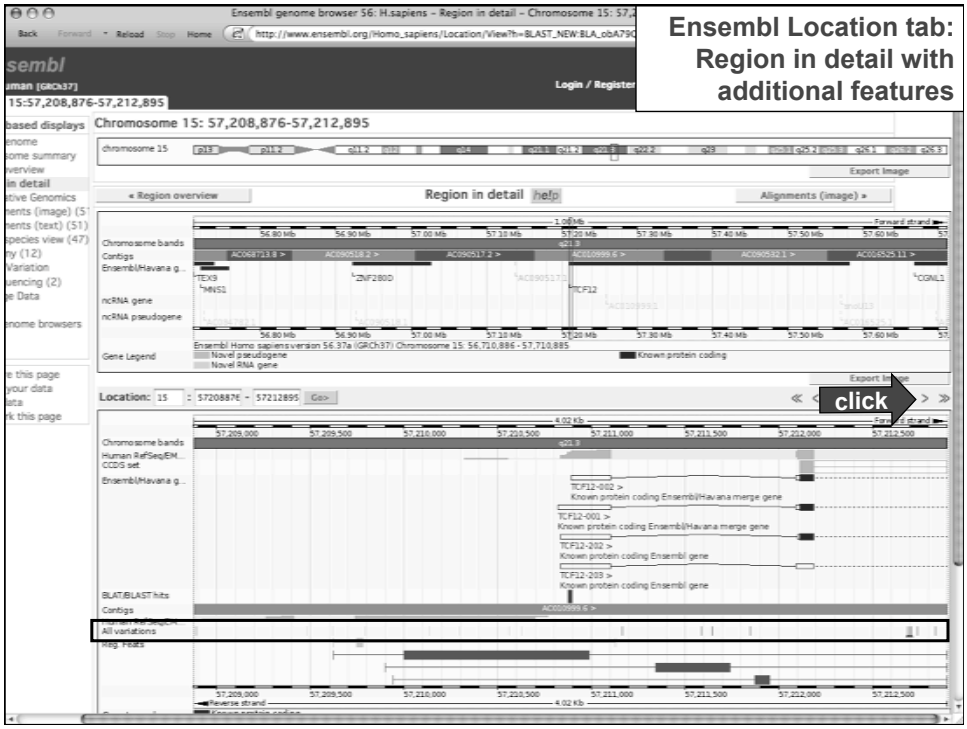


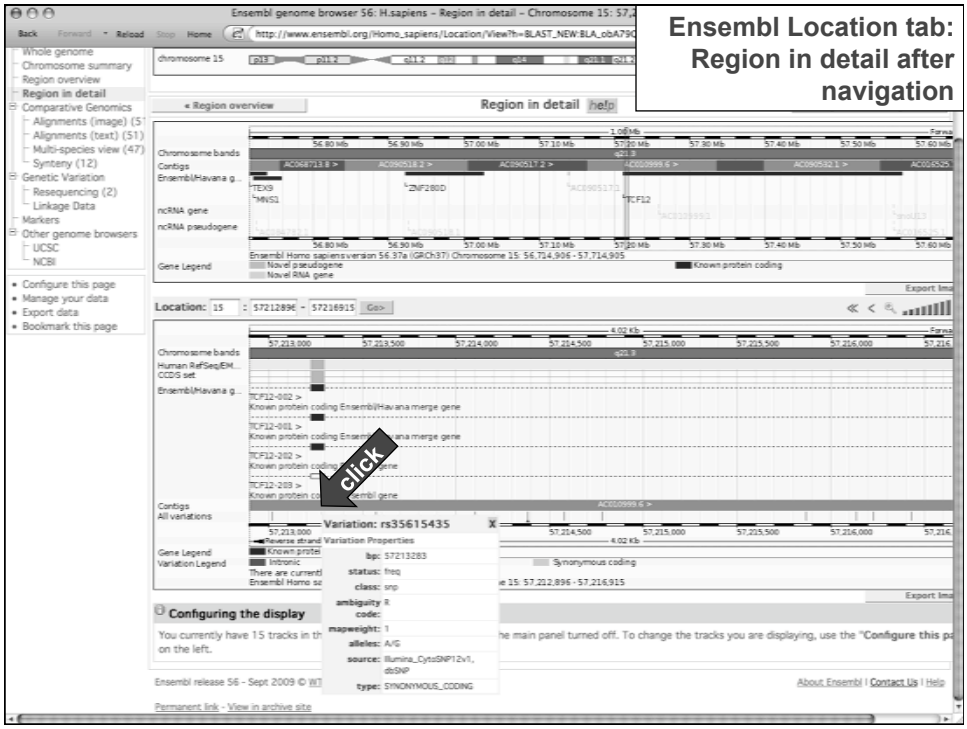
Ensembl

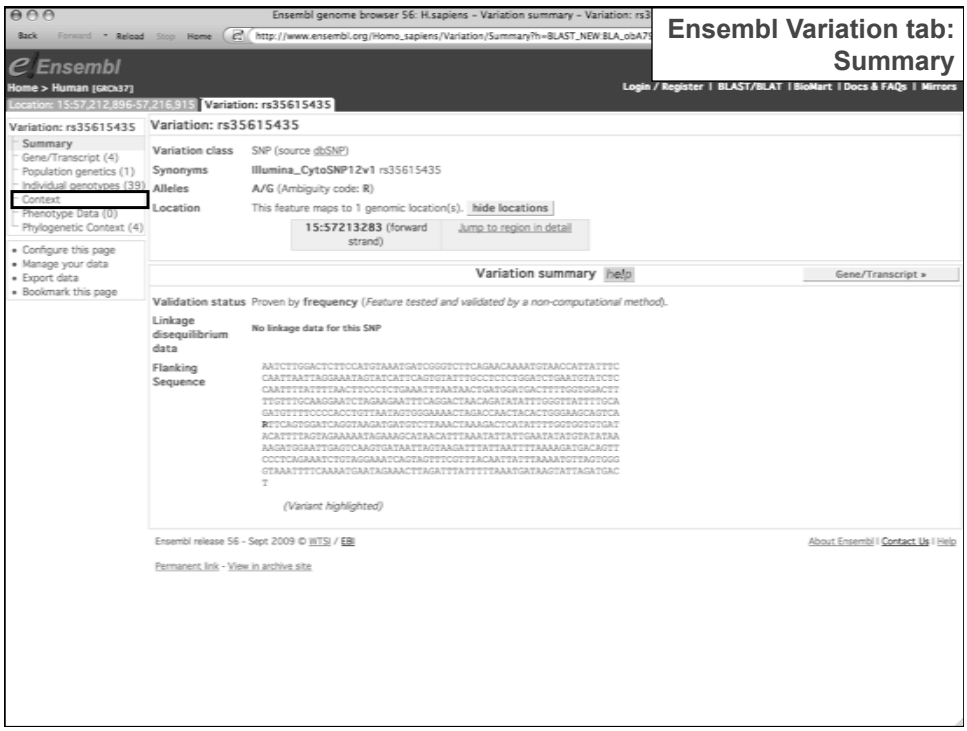
Location tab


Ensembl Location tab:
Region in detail
click


Ensembl Location tab:
Configure page


Ensembl Location tab:
Region in detail with
additional features
click


Ensembl Location tab:
Region in detail after
navigation
click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Variation: rs35615435
Variation: rs35615435
Summary
Gene/Transcript (4)
Population genetics (1)
Individual genotypes (39)
Context
Phenotype Data (0)
Phylogenetic Context (4)
Configure this page
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Variation: rs35615435
Variation class SNP (source dbSNP)
Synonyms Illumina_CytoSNP12v1 rs35615435
Alleles A/G (Ambiguity code: R)
Location This feature maps to 1 genomic location(s). hide locations
15:57213283 (forward strand)
Jump to region in detail
Variation summary help
Gene/Transcript »
Validation status Proven by frequency (Feature tested and validated by a non-computational method).
Linkage disequilibrium data No linkage data for this SNP
Flanking Sequence
(Variant highlighted)
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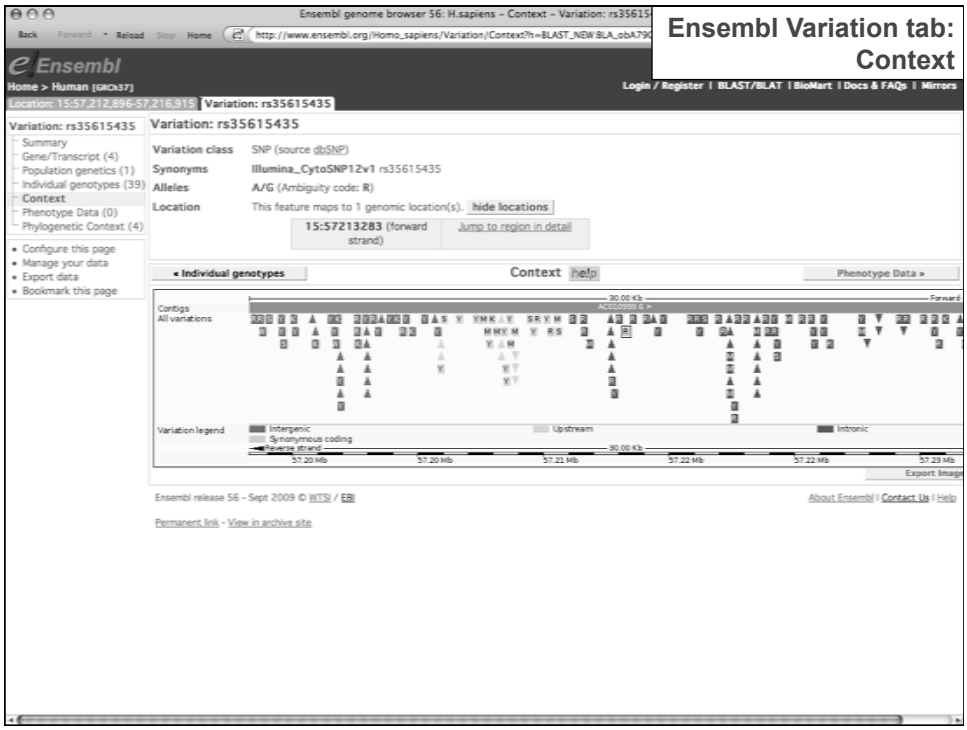
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15:57213283 (forward strand)
Jump to region in detail
« Individual genotypes
Context help
Phenotype Data »
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All variations
Variation legend
Intergenic
Synonymous coding
Reverse strand
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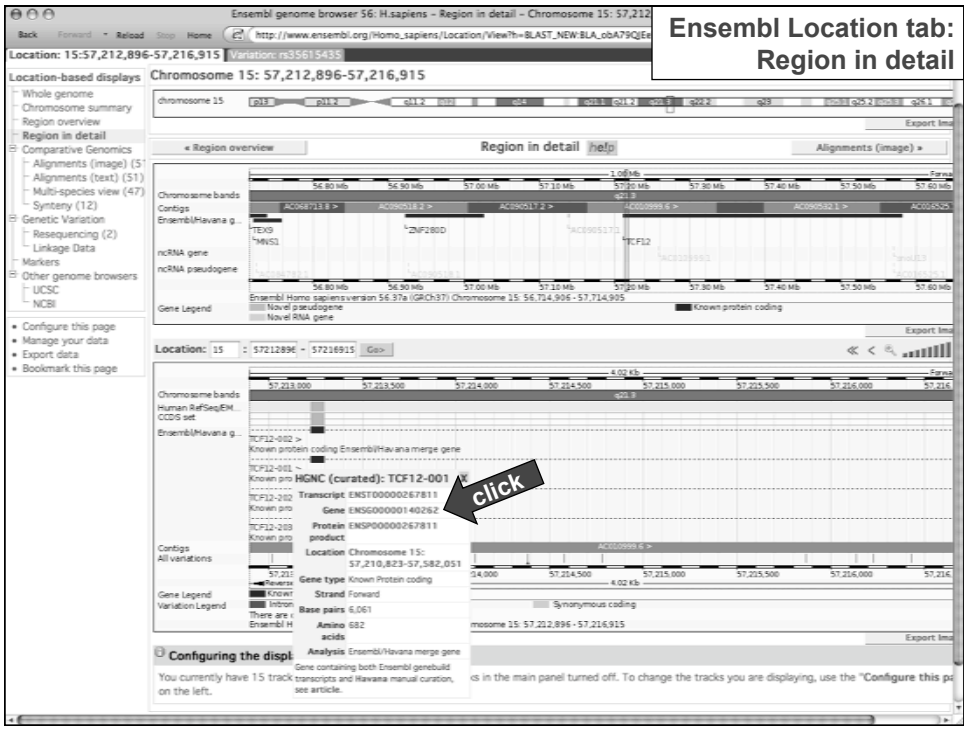
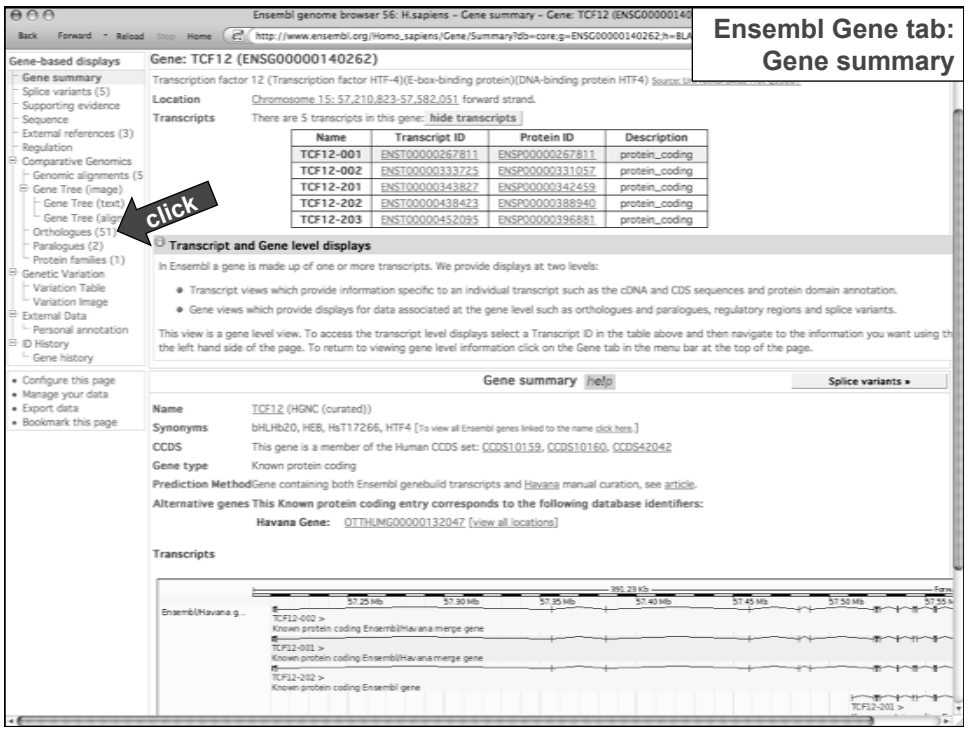
Gene tab

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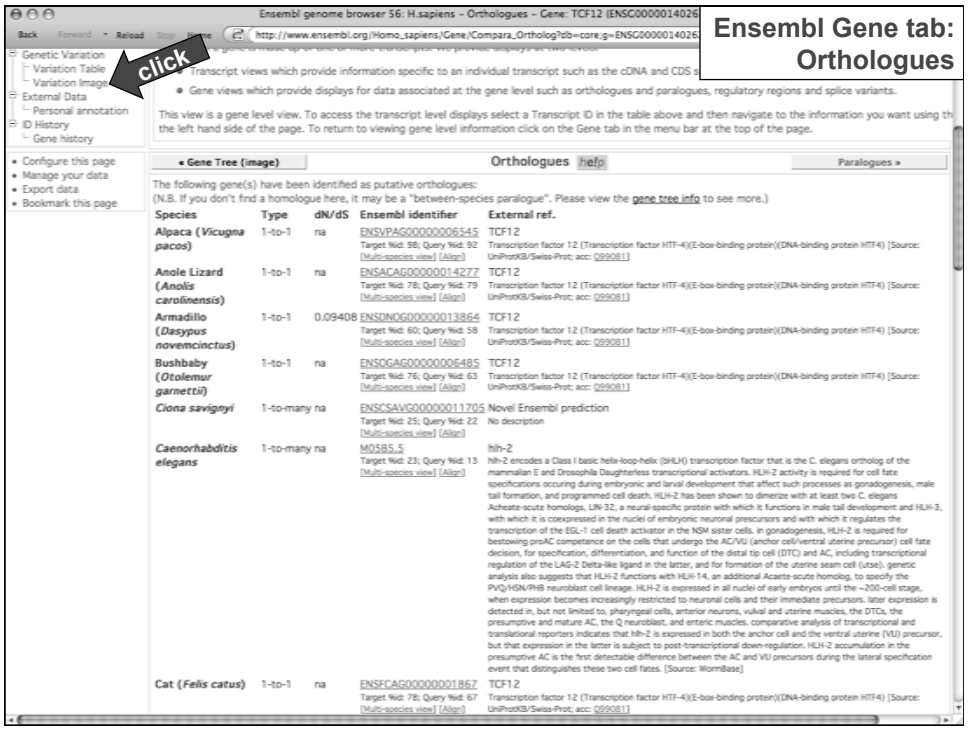
Gene: TCF12 (ENSG00000140262)
Gene summary
Transcript and Gene level displays


Ensembl Gene tab:
Orthologues
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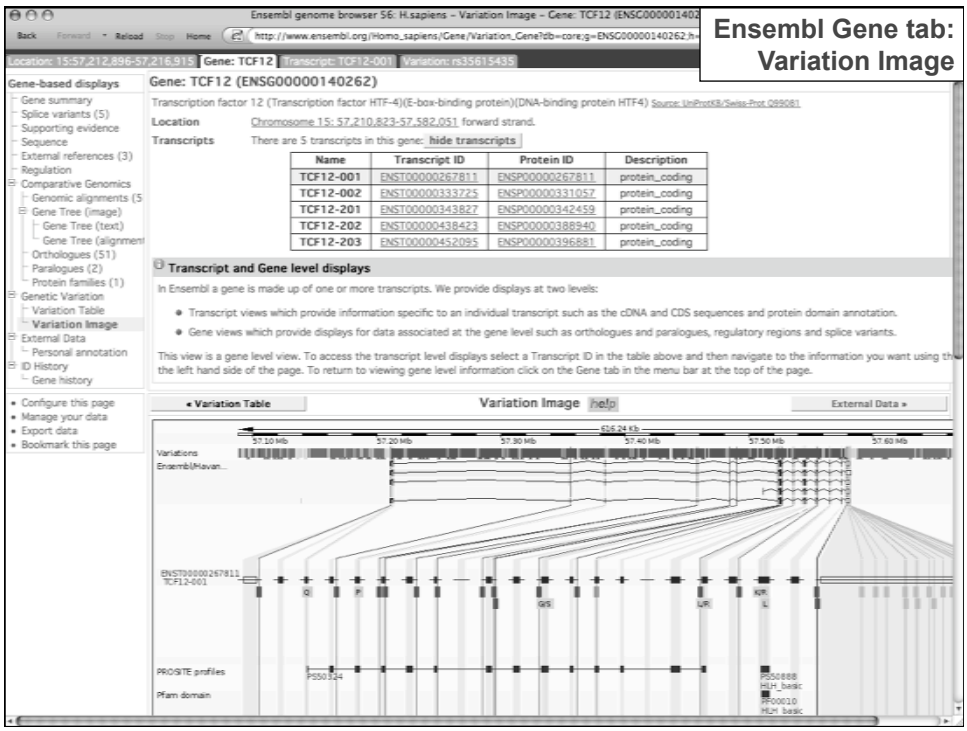
Orthologues
Gene Tree (image)
Paralogues »



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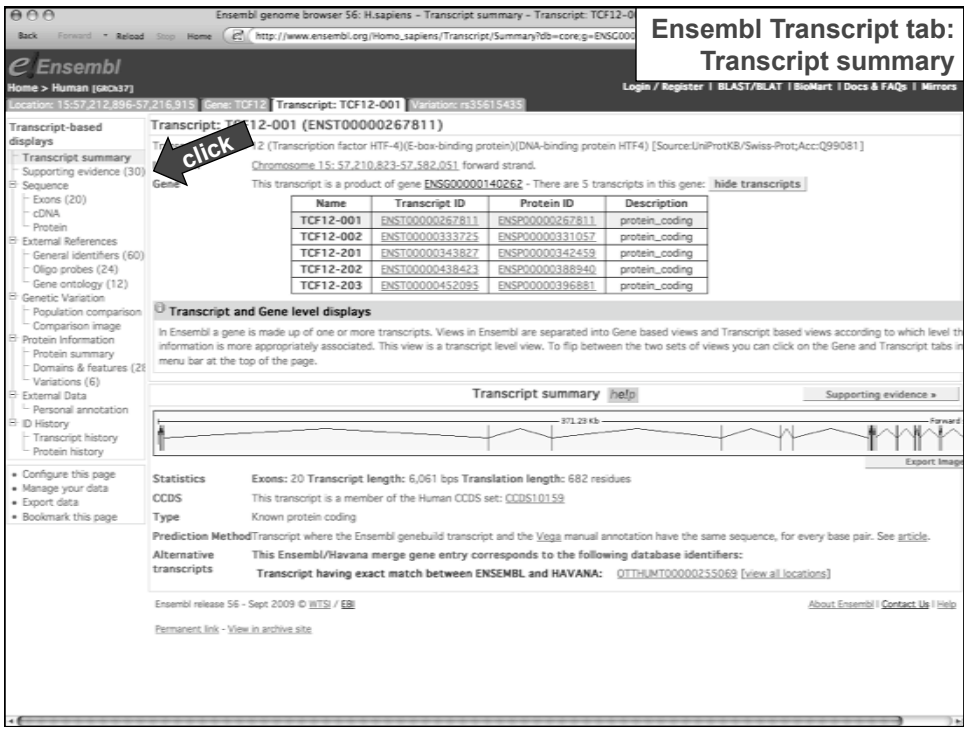
Transcript tab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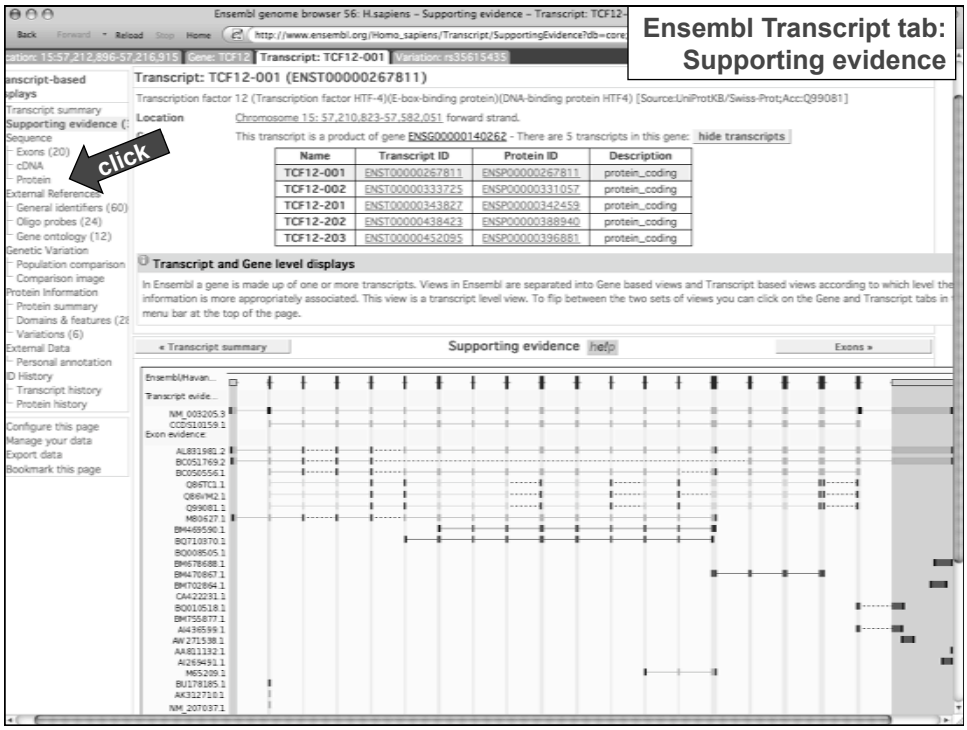
Transcript summary
Transcript and Gene level displays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Supporting evidence
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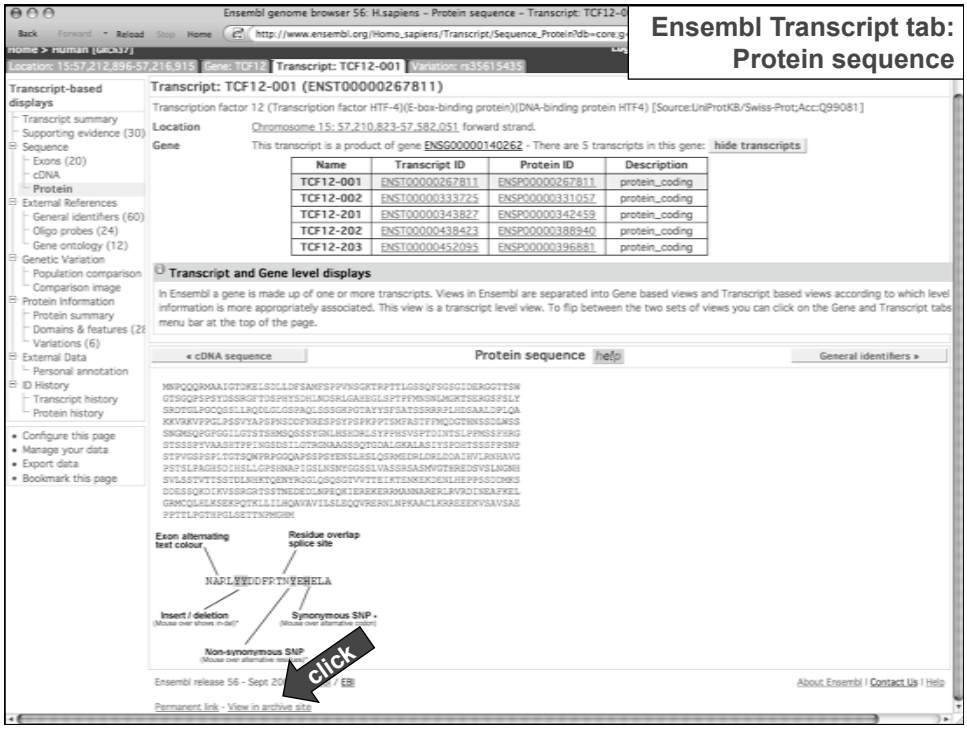
Supporting evidence
Transcript and Gene level displays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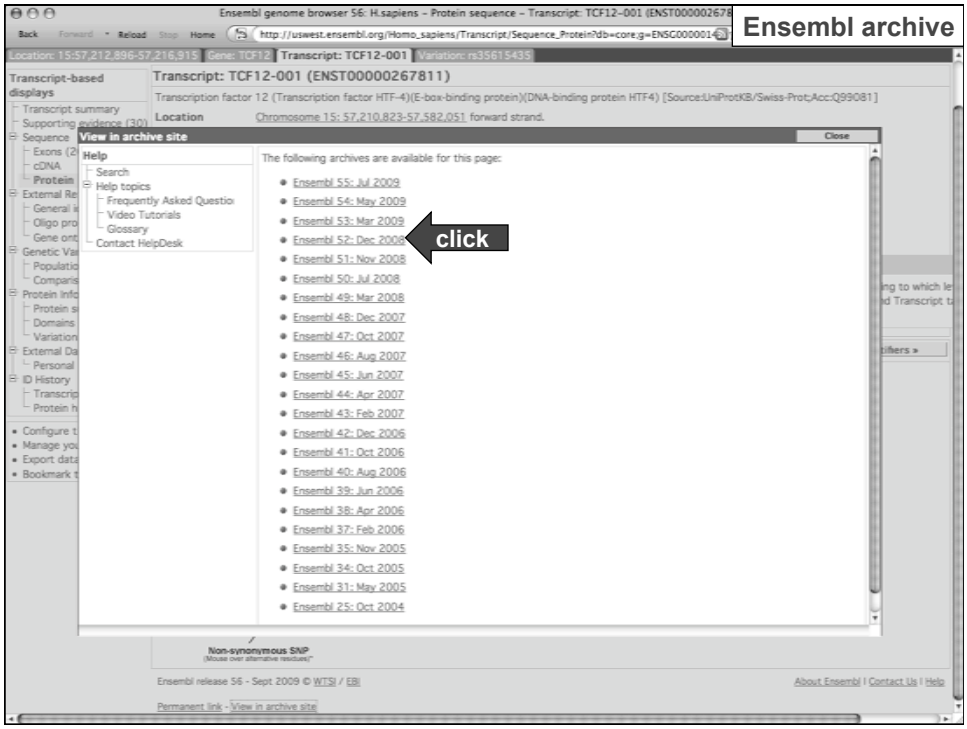
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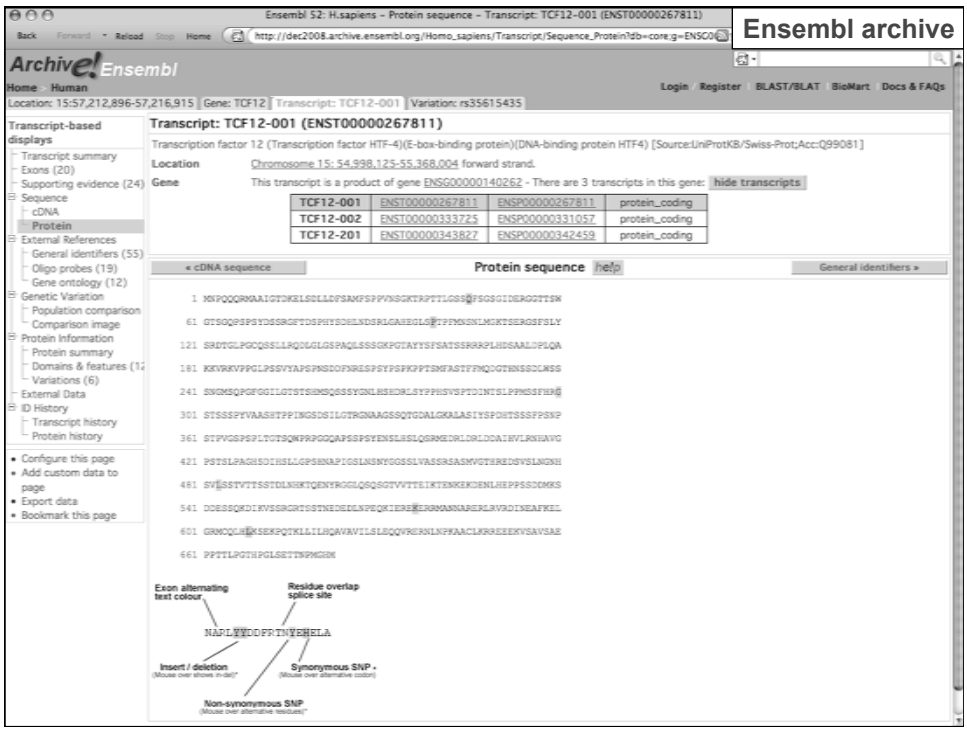

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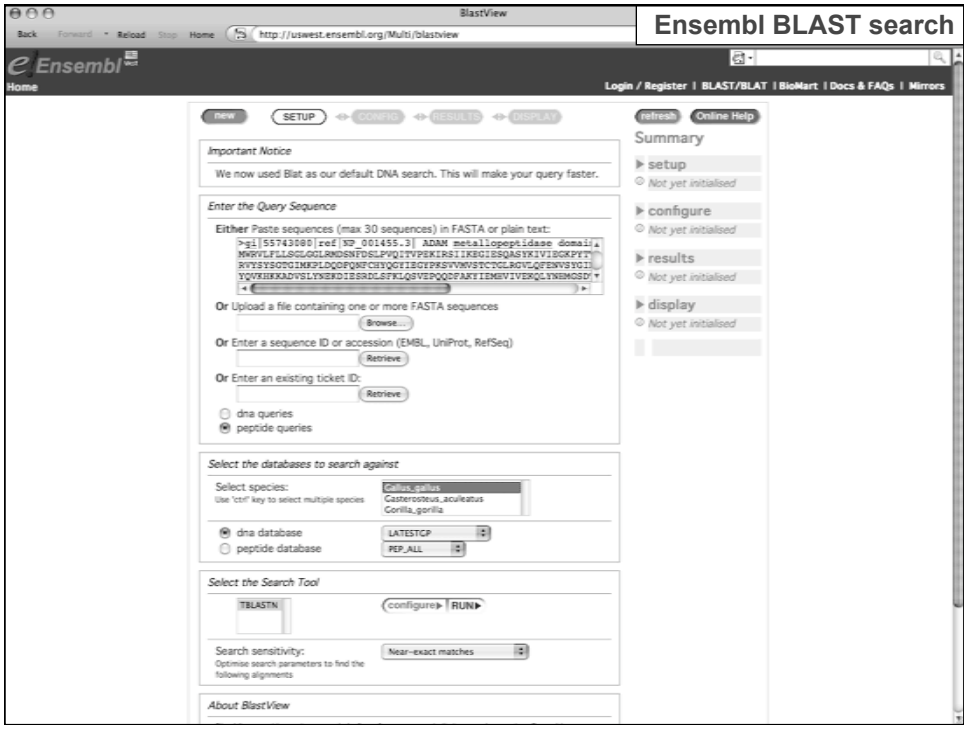
Find a chicken homolog of a human protein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results
Not yet initialised
display
Not yet initialised
Important Notice
We now used Blat as our default DNA search. This will make your query faster.
Enter the Query Sequence
Either Paste sequences (max 30 sequences) in FASTA or plain text:
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
Gorilla_gorilla
dna database
peptide database
LATESTCP
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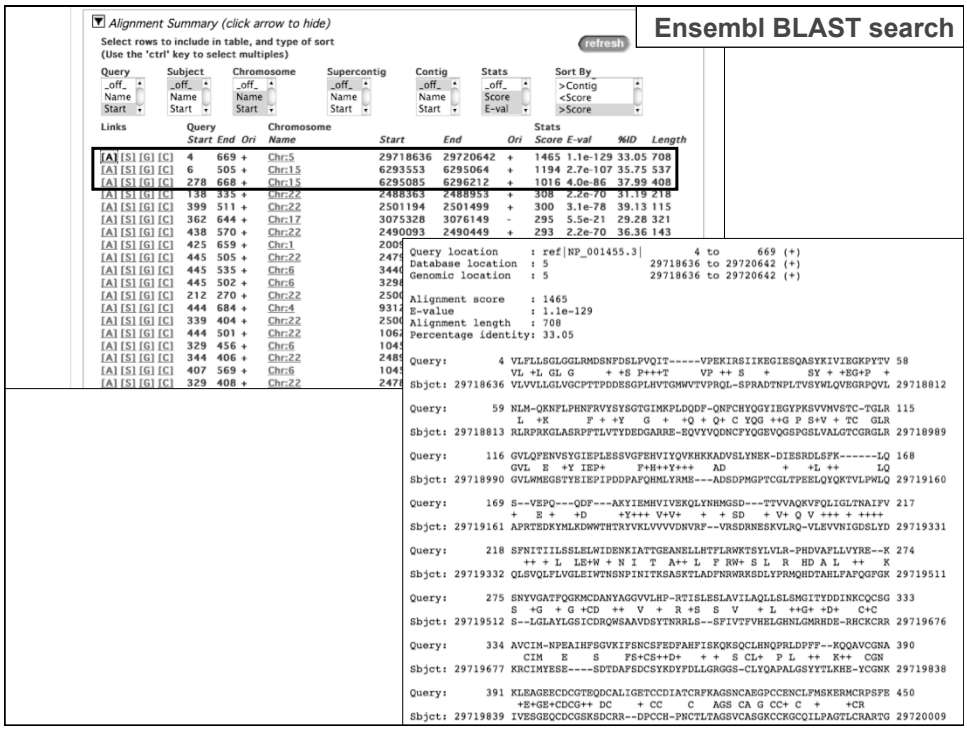
Select the Search Tool
TBLASTN
configure
RUN
Search sensitivity:
Optimise search parameters to find the following alignments
Near-exact matches
About BlastView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Alignment Summary (click arrow to hide)
Select rows to include in table, and type of sort
(Use the 'ctrl' key to select multiples)
refresh
Query
Subject
Chromosome
Supercontig
Contig
Stats
Sort By
Links
Query Start End Ori
Chromosome Name
Start End Ori
Stats Score E-val %ID Length
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[A] [S] [G] [C] 6 505 + Chr:15 6293553 6295064 + 1194 2.7e-107 35.75 537
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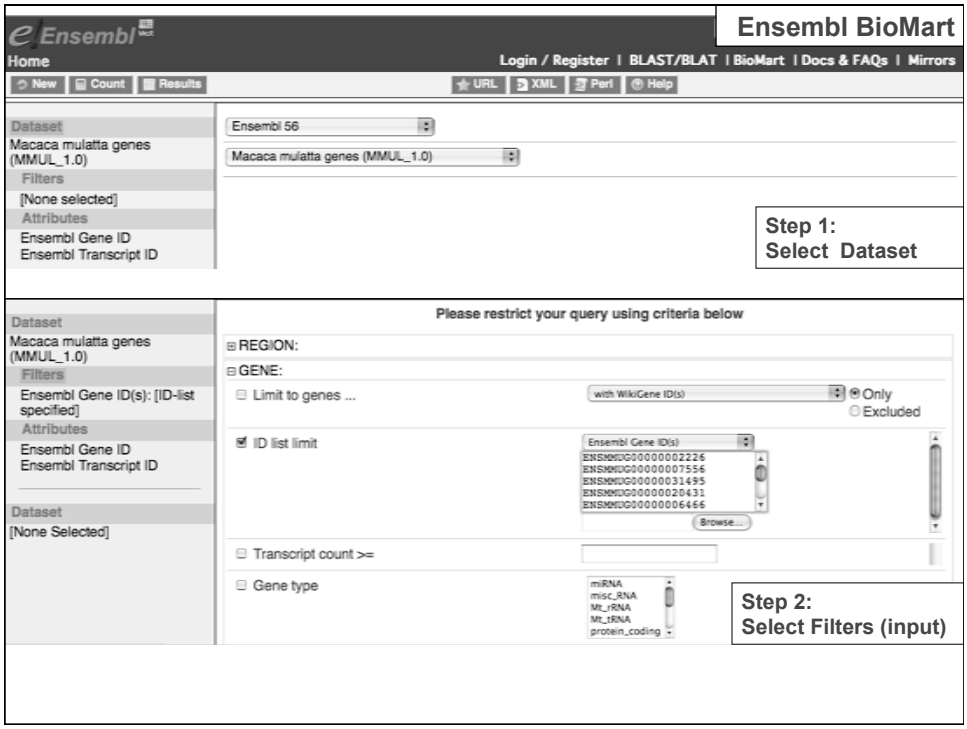
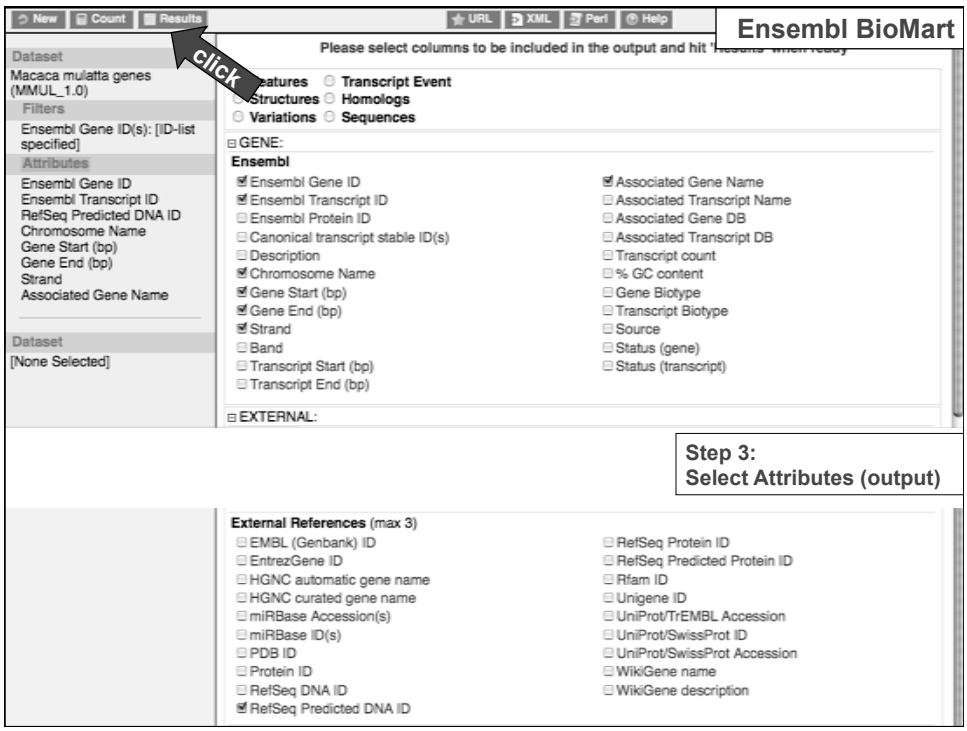
Ensembl

Using BioMart to cross-reference data from different sources

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Macaca mulatta genes (MMUL_1.0)
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Ensembl Transcript ID
RefSeq Predicted DNA ID
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Gene End (bp)
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Associated Gene Name
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Sequences
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Canonical transcript stable ID(s)
Description
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Status (transcript)
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External References (max 3)
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EntrezGene ID
HGNC automatic gene name
HGNC curated gene name
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PDB ID
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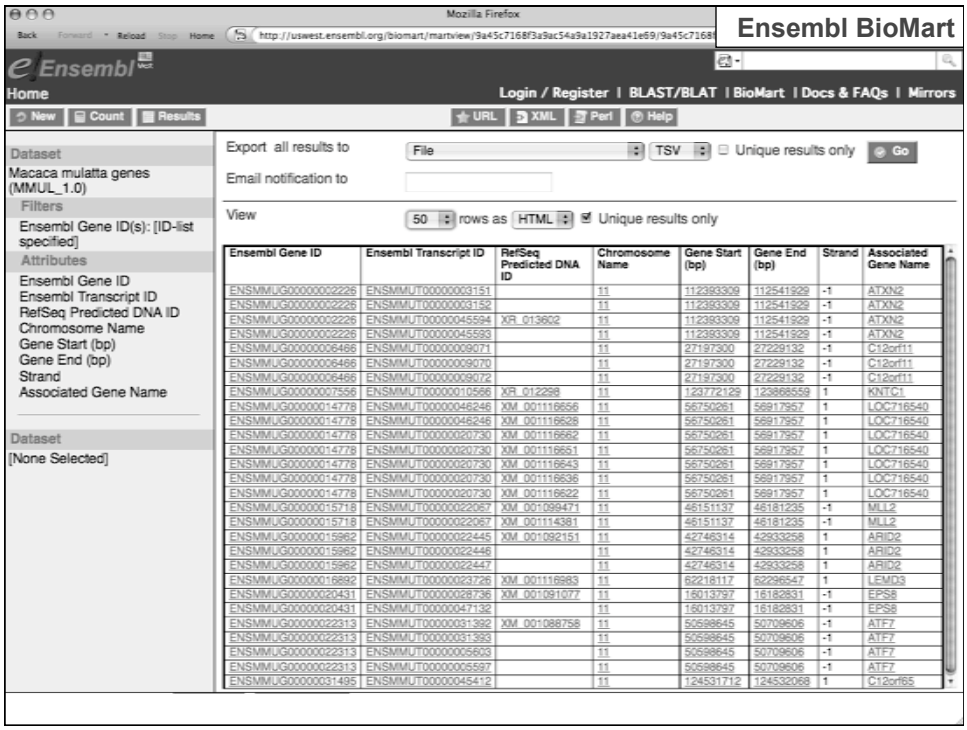
WikiGene description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Macaca mulatta genes (MMUL_1.0)
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Chromosome Name
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Gene End (bp)
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Associated Gene Name
Dataset
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Gene End (bp)
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Associated Gene Name
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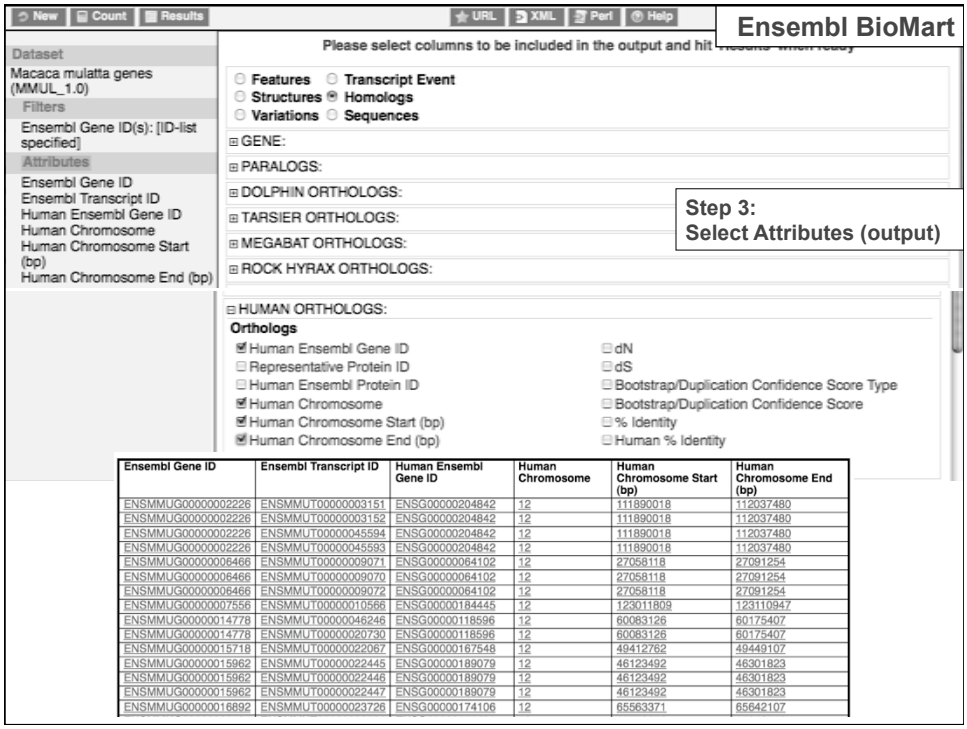

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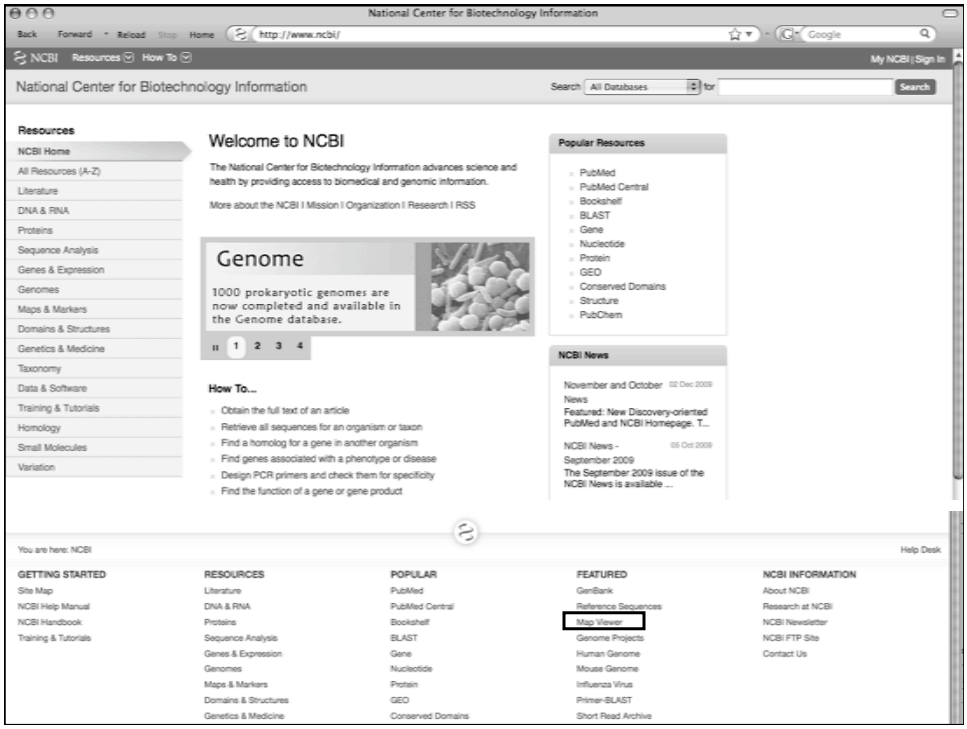
NCBI

View a genomic region between two SNPs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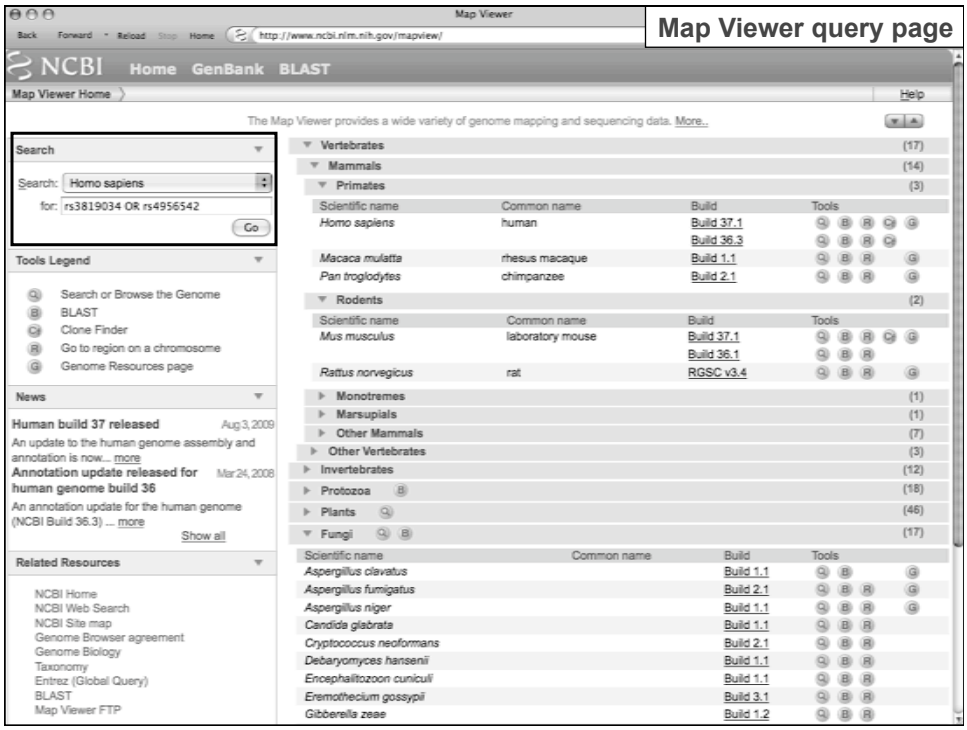
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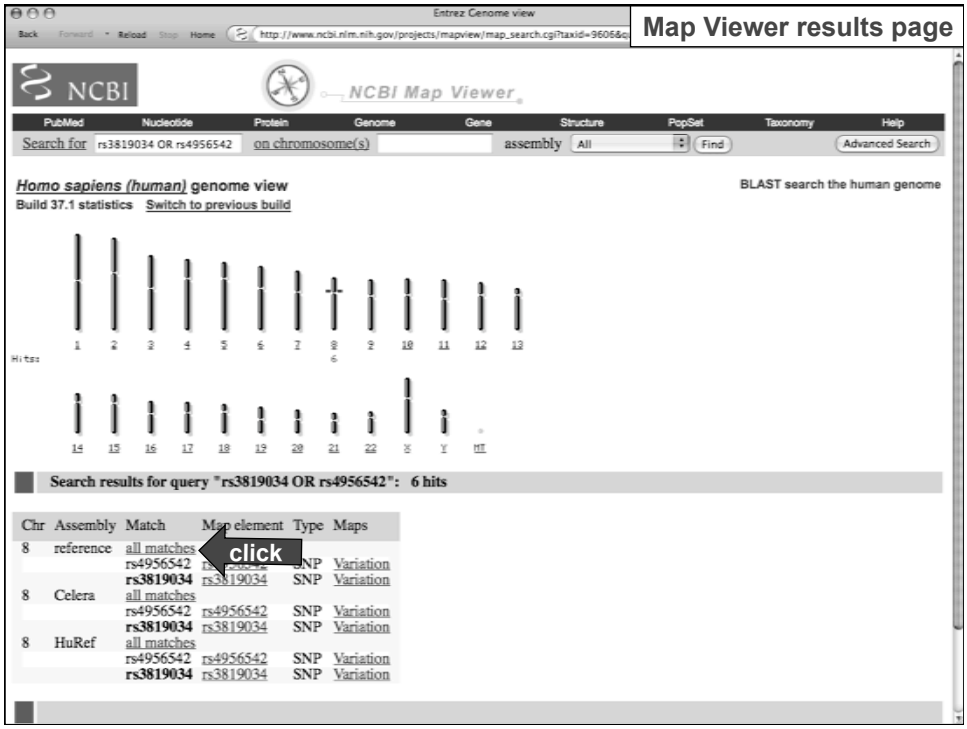
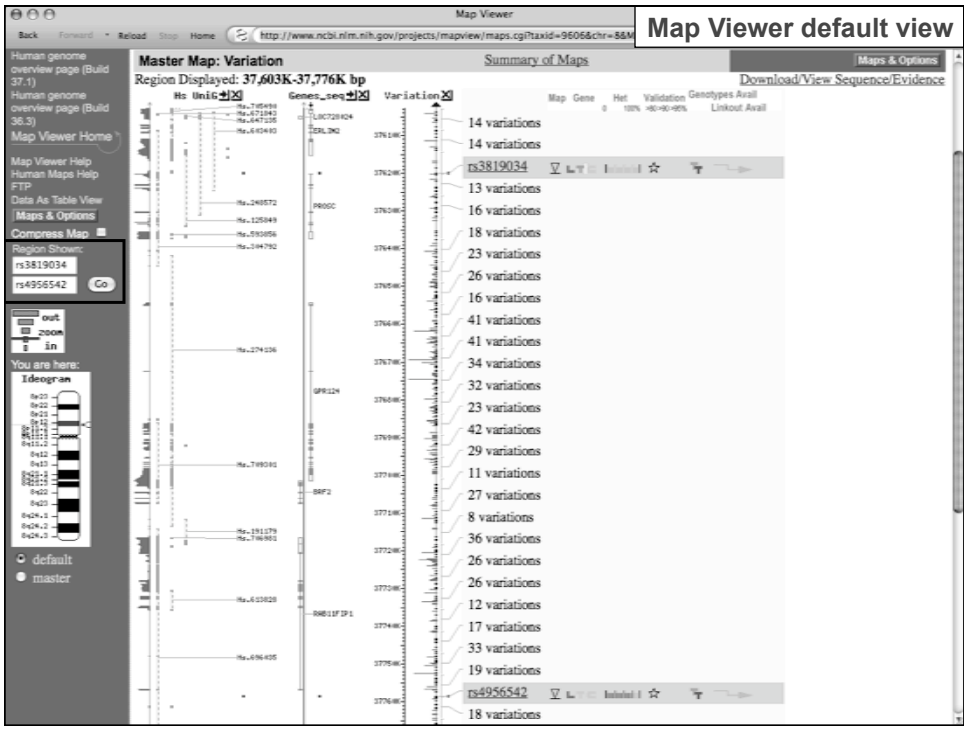
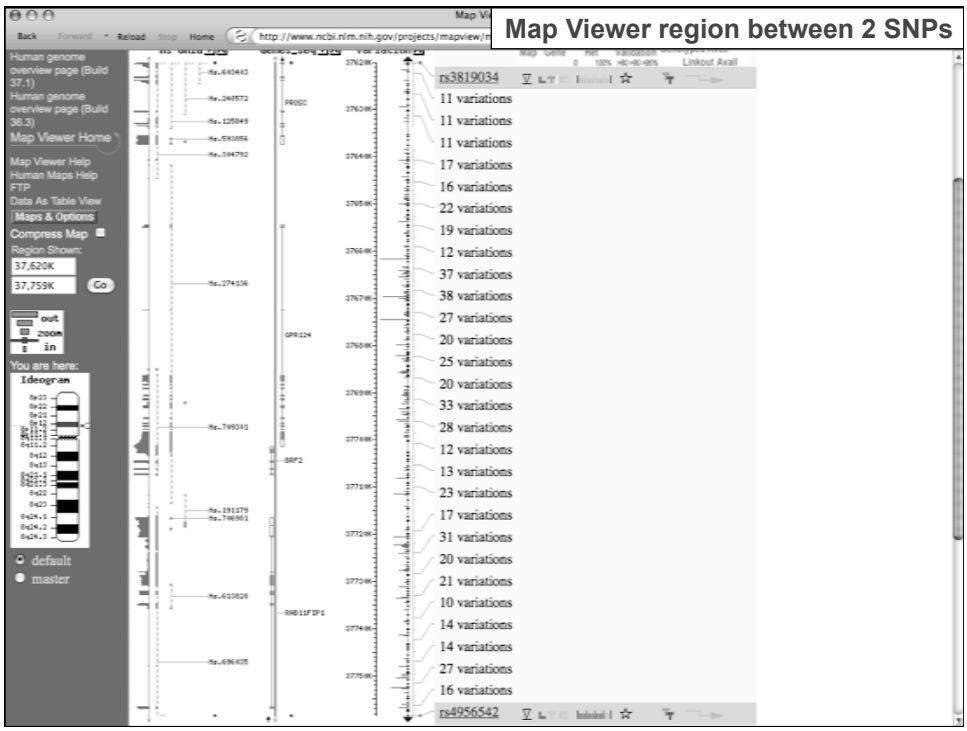
Help
The Map Viewer provides a wide variety of genome mapping and sequencing data. More..
Search
Search: Homo sapiens
for: rs3819034 OR rs4956542
Go
Tools Legend
Search or Browse the Genome
BLAST
Clone Finder
Go to region on a chromosome
Genome Resources page
News
Human build 37 released Aug 3, 2009
An update to the human genome assembly and annotation is now... more
Annotation update released for human genome build 36 Mar 24, 2008
An annotation update for the human genome (NCBI Build 36.3) ... more
Show all
Related Resources
NCBI Home
NCBI Web Search
NCBI Site map
Genome Browser agreement
Genome Biology
Taxonomy
Entrez (Global Query)
BLAST
Map Viewer FTP
Vertebrates (17)
Mammals (14)
Primates (3)
Scientific name | Common name | Build | Tools
Homo sapiens | human | Build 37.1 | Build 36.3
Macaca mulatta | rhesus macaque | Build 1.1
Pan troglodytes | chimpanzee | Build 2.1
Rodents (2)
Mus musculus | laboratory mouse | Build 37.1 | Build 36.1
Rattus norvegicus | rat | RGSC v3.4
Monotremes (1)
Marsupials (1)
Other Mammals (7)
Other Vertebrates (3)
Invertebrates (12)
Protozoa (18)
Plants (46)
Fungi (17)
Aspergillus clavatus | Build 1.1
Aspergillus fumigatus | Build 2.1
Aspergillus niger | Build 1.1
Candida glabrata | Build 1.1
Cryptococcus neoformans | Build 2.1
Debaryomyces hansenii | Build 1.1
Encephalitozoon cuniculi | Build 1.1
Eremothecium gossypii | Build 3.1
Gibberella zeae | Build 1.2


Map Viewer results page
Entrez Genome view
NCBI
NCBI Map Viewer
PubMed Nucleotide Protein Genome Gene Structure PopSet Taxonomy Help
Search for rs3819034 OR rs4956542 on chromosome(s) assembly All Find Advanced Search
Homo sapiens (human) genome view
Build 37.1 statistics Switch to previous build
BLAST search the human genome
Search results for query "rs3819034 OR rs4956542": 6 hits
Chr Assembly Match Map element Type Maps
8 reference all matches
rs4956542 rs4956542 SNP Variation
rs3819034 rs3819034 SNP Variation
8 Celera all matches
rs4956542 rs4956542 SNP Variation
rs3819034 rs3819034 SNP Variation
8 HuRef all matches
rs4956542 rs4956542 SNP Variation
rs3819034 rs3819034 SNP Variation
click

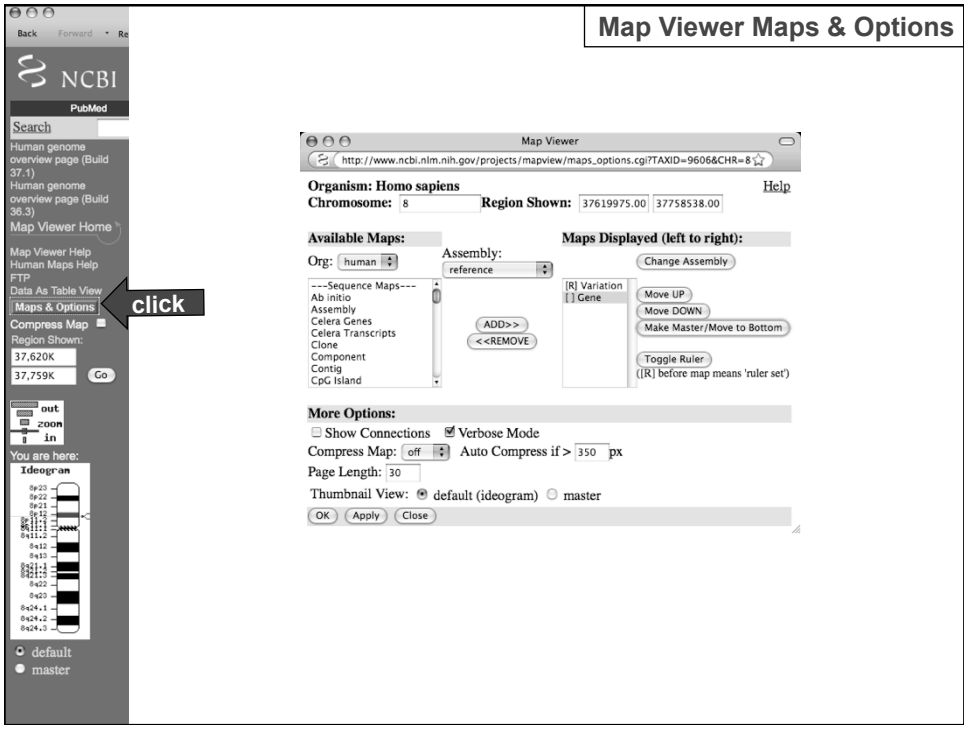

Map Viewer default view
Map Viewer
Master Map: Variation
Summary of Maps
Region Displayed: 37,603K-37,776K bp
Download/View Sequence/Evidence
Maps & Options
Human genome overview page (Build 37.1)
Human genome overview page (Build 36.3)
Map Viewer Home
Map Viewer Help
Human Maps Help
FTP
Data As Table View
Compress Map
Region Shown:
rs3819034
rs4956542
Go
out
zoom
in
You are here:
Ideogram
default
master
14 variations
14 variations
rs3819034
13 variations
16 variations
18 variations
23 variations
26 variations
16 variations
41 variations
41 variations
34 variations
32 variations
23 variations
42 variations
29 variations
11 variations
27 variations
8 variations
36 variations
26 variations
26 variations
12 variations
17 variations
33 variations
19 variations
rs4956542
18 variations

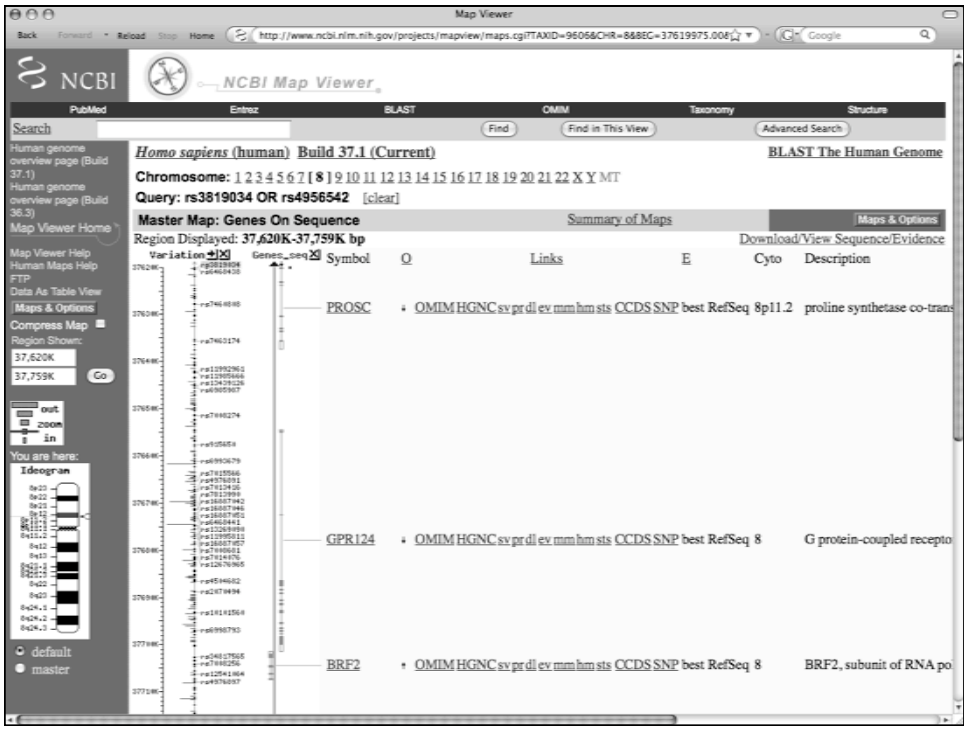


NCBI

Change the maps displayed on the Map Viewer

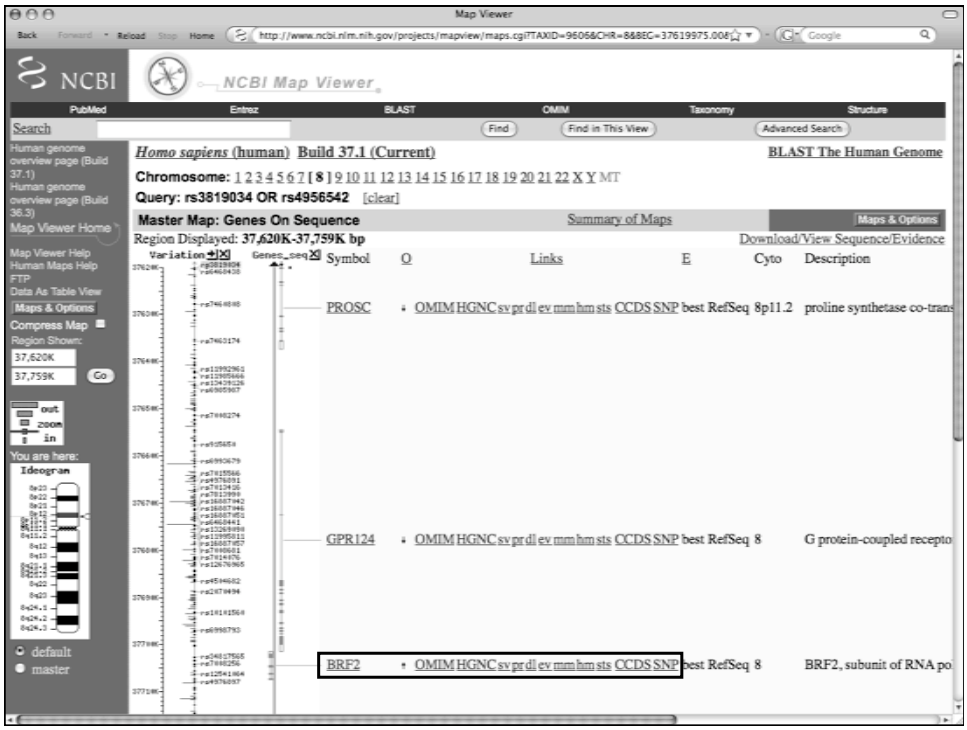
Map Viewer Maps & Options

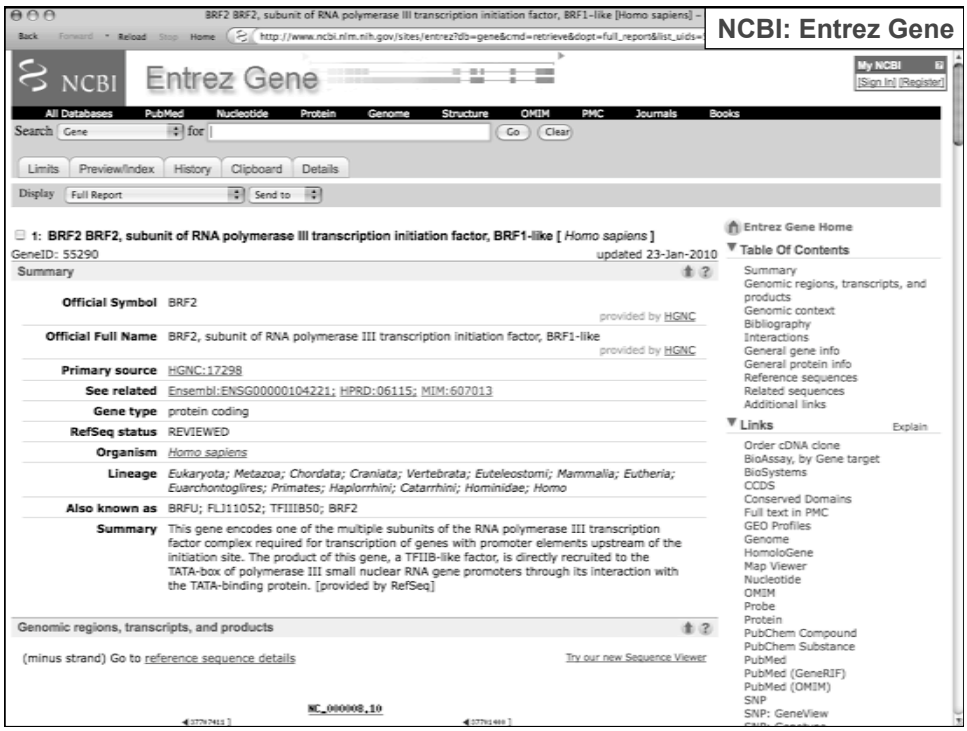


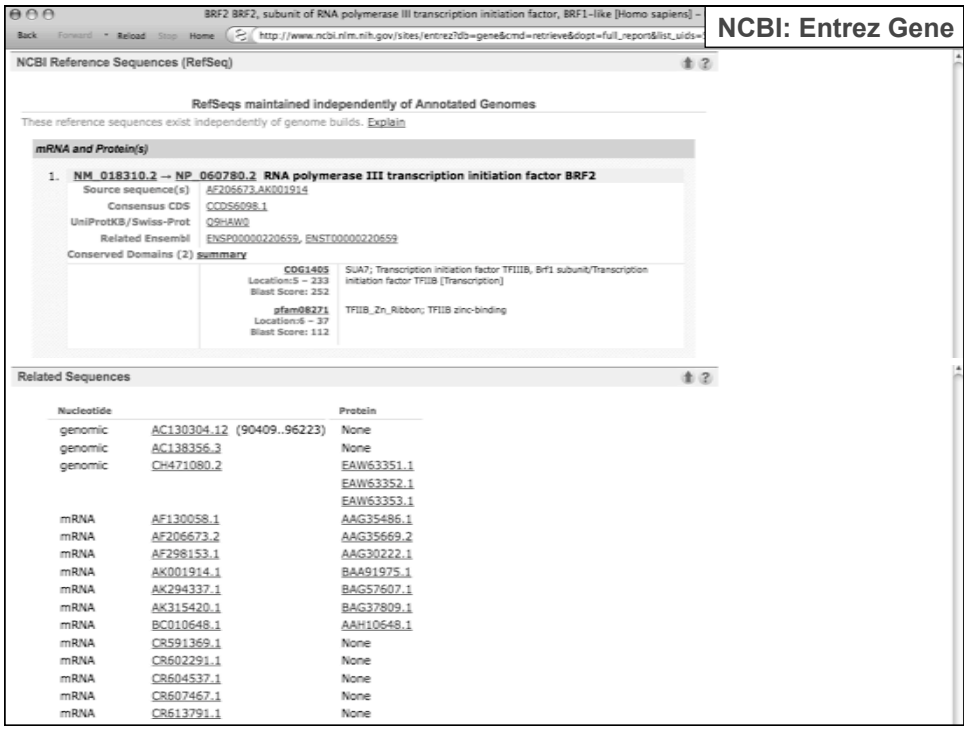


NCBI

View additional information about a gene




NCBI: Entrez Gene
NCBI
Entrez Gene
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search Gene for
Limits Preview/Index History Clipboard Details
Display Full Report Send to
1: BRF2 BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like [Homo sapiens]
GeneID: 55290 updated 23-Jan-2010
Summary
Official Symbol BRF2 provided by HGNC
Official Full Name BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like provided by HGNC
Primary source HGNC:17298
See related Ensembl:ENSG00000104221; HPRD:06115; MIM:607013
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 BRFU; FLJ11052; TFIIIB50; BRF2
Summary This gene encodes one of the multiple subunits of the RNA polymerase III transcription factor complex required for transcription of genes with promoter elements upstream of the initiation site. The product of this gene, a TFIIB-like factor, is directly recruited to the TATA-box of polymerase III small nuclear RNA gene promoters through its interaction with the TATA-binding protein. [provided by RefSeq]
Genomic regions, transcripts, and products
(minus strand) Go to reference sequence details
Try our new Sequence Viewer
NC_000008.10
Entrez Gene Home
Table Of Contents
Summary
Genomic regions, transcripts, and products
Genomic context
Bibliography
Interactions
General gene info
General protein info
Reference sequences
Related sequences
Additional links
Links
Explain
Order cDNA clone
BioAssay, by Gene target
BioSystems
CCDS
Conserved Domains
Full text in PMC
GEO Profiles
Genome
HomoloGene
Map Viewer
Nucleotide
OMIM
Probe
Protein
PubChem Compound
PubChem Substance
PubMed
PubMed (GeneRIF)
PubMed (OMIM)
SNP
SNP: GeneView


NCBI: Entrez Gene
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_018310.2 → NP_060780.2 RNA polymerase III transcription initiation factor BRF2
Source sequence(s) AF205673,AK001914
Consensus CDS CCDS6098.1
UniProtKB/Swiss-Prot Q9HAW0
Related Ensembl ENSP00000220659, ENST00000220659
Conserved Domains (2) summary
COG1405 Location:5 – 233 Blast Score: 252 SUA7; Transcription initiation factor TFIIIB, Brf1 subunit/Transcription initiation factor TFIIB [Transcription]
pfam08271 Location:6 – 37 Blast Score: 112 TFIIB_Zn_Ribbon; TFIIB zinc-binding
Related Sequences
Nucleotide Protein
genomic AC130304.12 (90409..96223) None
genomic AC138356.3 None
genomic CH471080.2 EAW63351.1
EAW63352.1
EAW63353.1
mRNA AF130058.1 AAG35486.1
mRNA AF206673.2 AAG35669.2
mRNA AF298153.1 AAG30222.1
mRNA AK001914.1 BAA91975.1
mRNA AK294337.1 BAG57607.1
mRNA AK315420.1 BAG37809.1
mRNA BC010648.1 AAH10648.1
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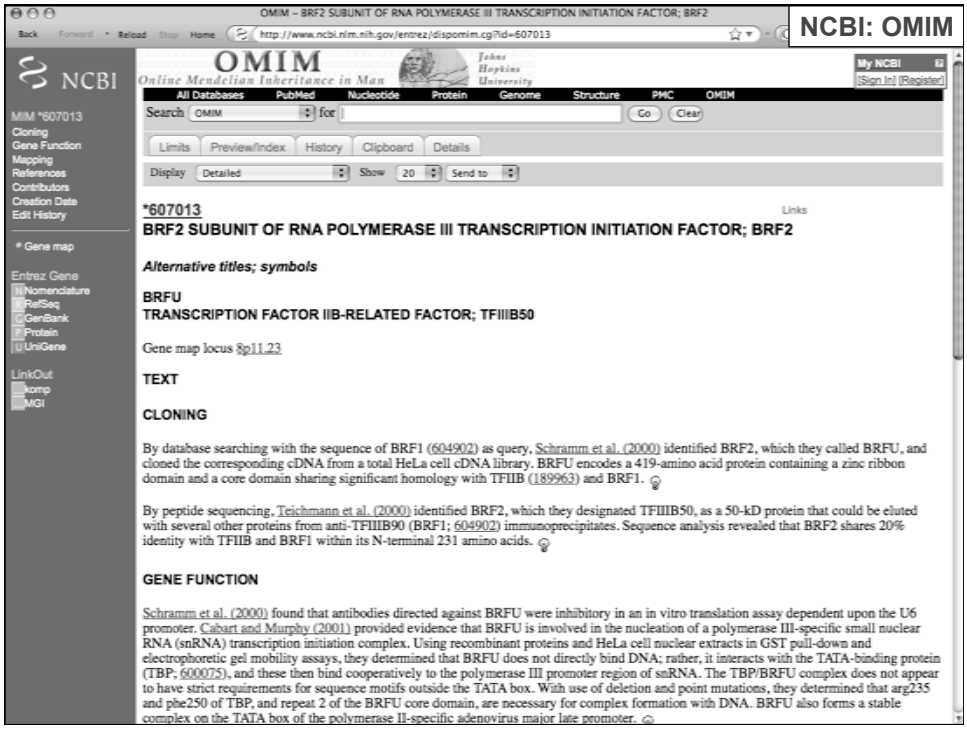
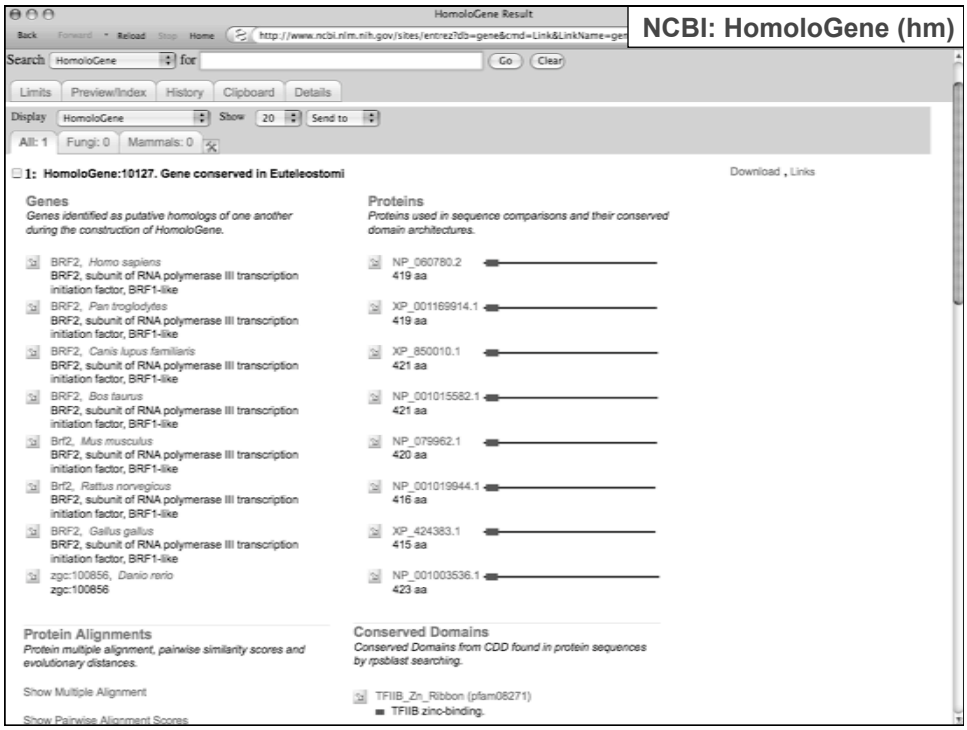
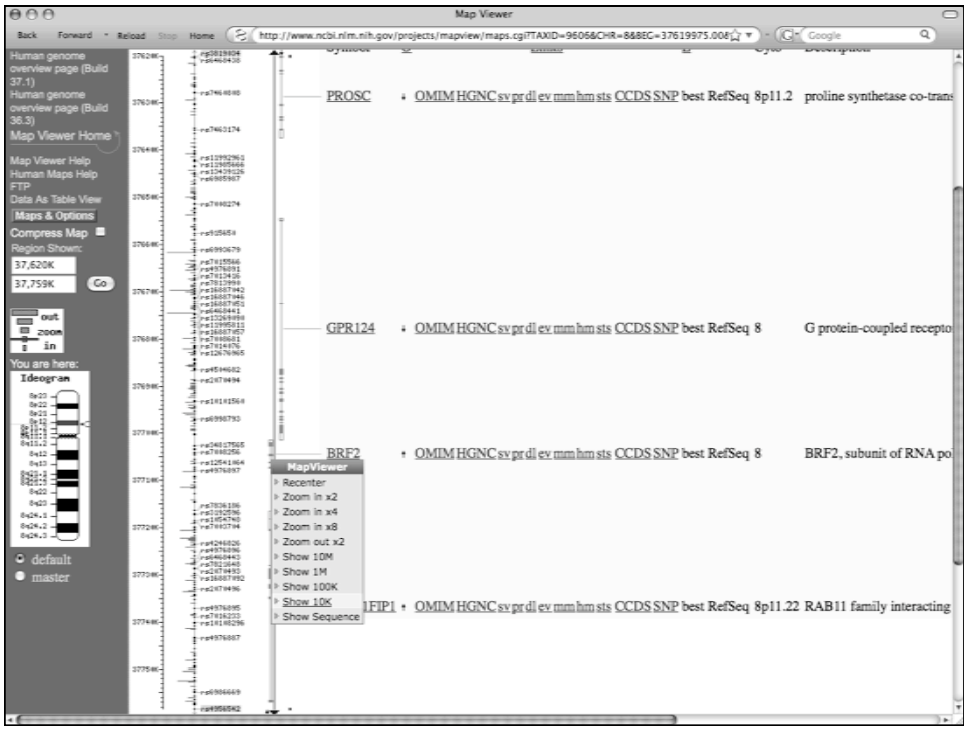
mRNA CR602291.1 None
mRNA CR604537.1 None
mRNA CR607467.1 None
mRNA CR613791.1 None

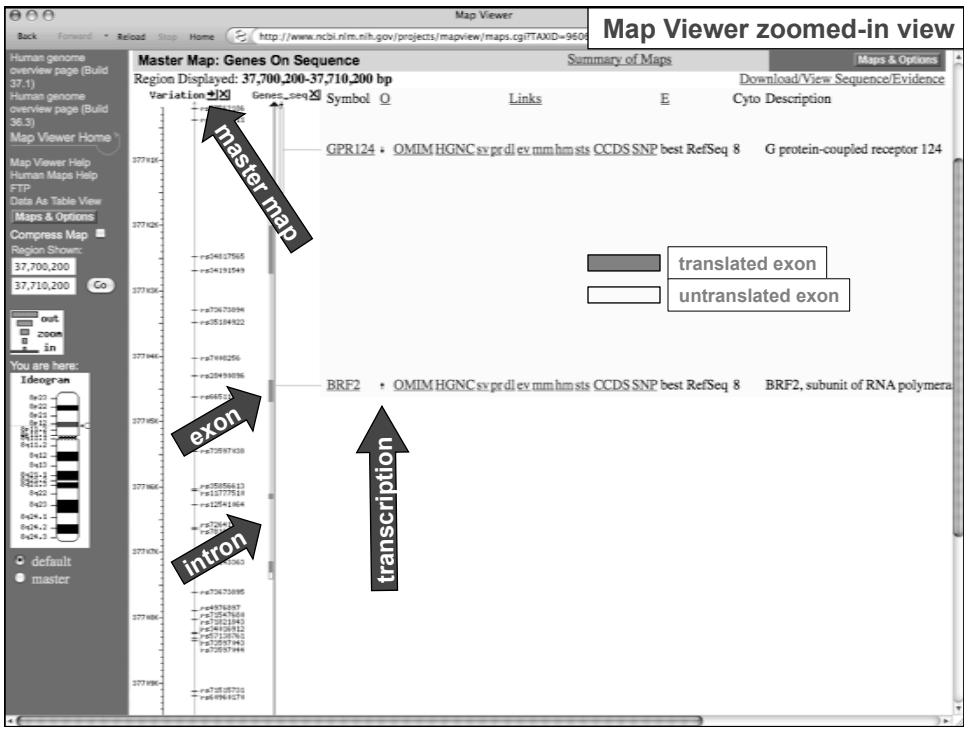

NCBI: OMIM
OMIM – BRF2 SUBUNIT OF RNA POLYMERASE III TRANSCRIPTION INITIATION FACTOR; BRF2
http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=607013
NCBI
OMIM
Online Mendelian Inheritance in Man
Johns Hopkins University
All Databases PubMed Nucleotide Protein Genome Structure PMC OMIM
Search OMIM for Go Clear
Limits Preview/Index History Clipboard Details
Display Detailed Show 20 Send to
MIM *607013
Cloning
Gene Function
Mapping
References
Contributors
Creation Date
Edit History
* Gene map
Entrez Gene
Nomenclature
RefSeq
GenBank
Protein
UniGene
LinkOut
komp
MGI
*607013
Links
BRF2 SUBUNIT OF RNA POLYMERASE III TRANSCRIPTION INITIATION FACTOR; BRF2
Alternative titles; symbols
BRFU
TRANSCRIPTION FACTOR IIB-RELATED FACTOR; TFIIIB50
Gene map locus 8p11.23
TEXT
CLONING
By database searching with the sequence of BRF1 (604902) as query, Schramm et al. (2000) identified BRF2, which they called BRFU, and cloned the corresponding cDNA from a total HeLa cell cDNA library. BRFU encodes a 419-amino acid protein containing a zinc ribbon domain and a core domain sharing significant homology with TFIIB (189963) and BRF1.
By peptide sequencing, Teichmann et al. (2000) identified BRF2, which they designated TFIIIB50, as a 50-kD protein that could be eluted with several other proteins from anti-TFIIIB90 (BRF1; 604902) immunoprecipitates. Sequence analysis revealed that BRF2 shares 20% identity with TFIIB and BRF1 within its N-terminal 231 amino acids.
GENE FUNCTION
Schramm et al. (2000) found that antibodies directed against BRFU were inhibitory in an in vitro translation assay dependent upon the U6 promoter. Cabart and Murphy (2001) provided evidence that BRFU is involved in the nucleation of a polymerase III-specific small nuclear RNA (snRNA) transcription initiation complex. Using recombinant proteins and HeLa cell nuclear extracts in GST pull-down and electrophoretic gel mobility assays, they determined that BRFU does not directly bind DNA; rather, it interacts with the TATA-binding protein (TBP; 600075), and these then bind cooperatively to the polymerase III promoter region of snRNA. The TBP/BRFU complex does not appear to have strict requirements for sequence motifs outside the TATA box. With use of deletion and point mutations, they determined that arg235 and phe250 of TBP, and repeat 2 of the BRFU core domain, are necessary for complex formation with DNA. BRFU also forms a stable complex on the TATA box of the polymerase II-specific adenovirus major late promoter.

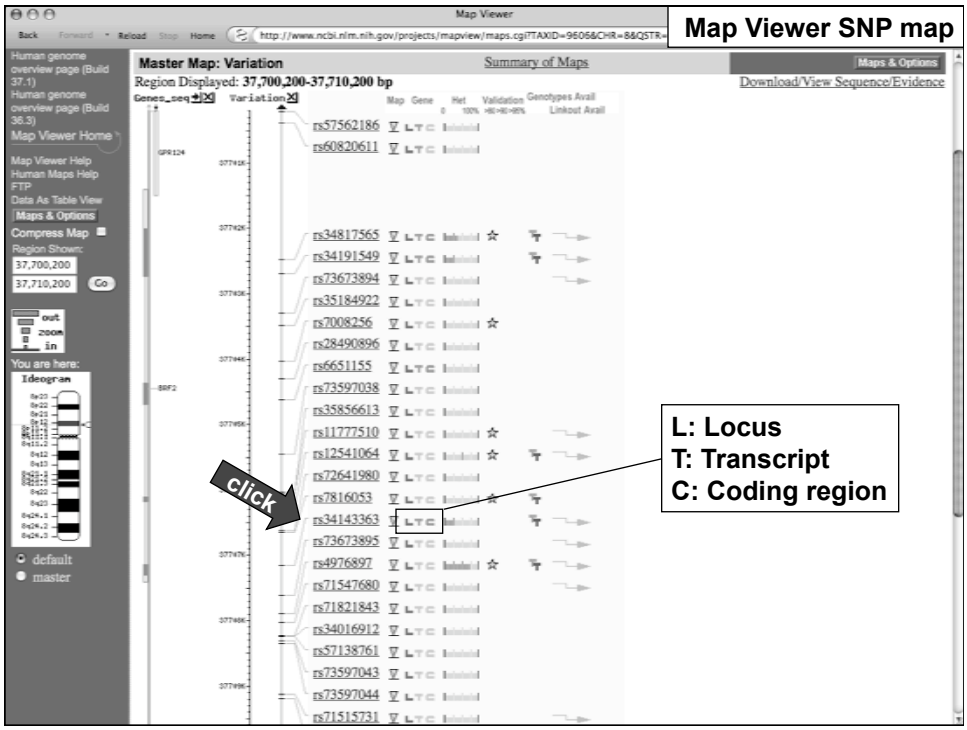

NCBI: HomoloGene (hm)
HomoloGene Result
Search HomoloGene for Go Clear
Limits Preview/Index History Clipboard Details
Display HomoloGene Show 20 Send to
All: 1 Fungi: 0 Mammals: 0
1: HomoloGene:10127. Gene conserved in Euteleostomi
Download , Links
Genes
Genes identified as putative homologs of one another during the construction of HomoloGene.
Proteins
Proteins used in sequence comparisons and their conserved domain architectures.
BRF2, Homo sapiens
BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
NP_060780.2
419 aa
BRF2, Pan troglodytes
BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
XP_001169914.1
419 aa
BRF2, Canis lupus familiaris
BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
XP_850010.1
421 aa
BRF2, Bos taurus
BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
NP_001015582.1
421 aa
Brf2, Mus musculus
BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
NP_079962.1
420 aa
Brf2, Rattus norvegicus
BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
NP_001019944.1
416 aa
BRF2, Gallus gallus
BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
XP_424383.1
415 aa
zgc:100856, Danio rerio
zgc:100856
NP_001003536.1
423 aa
Protein Alignments
Protein multiple alignment, pairwise similarity scores and evolutionary distances.
Conserved Domains
Conserved Domains from CDD found in protein sequences by rpsblast searching.
Show Multiple Alignment
Show Pairwise Alignment Scores
TFIIB_Zn_Ribbon (pfam08271)
TFIIB zinc-binding.

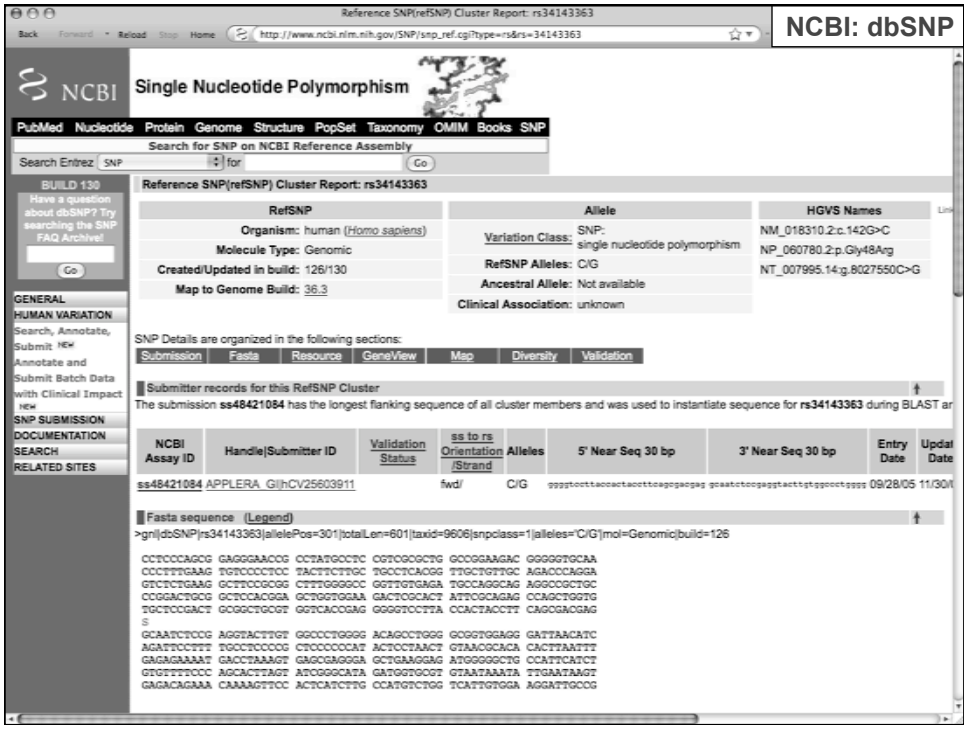
NCBI

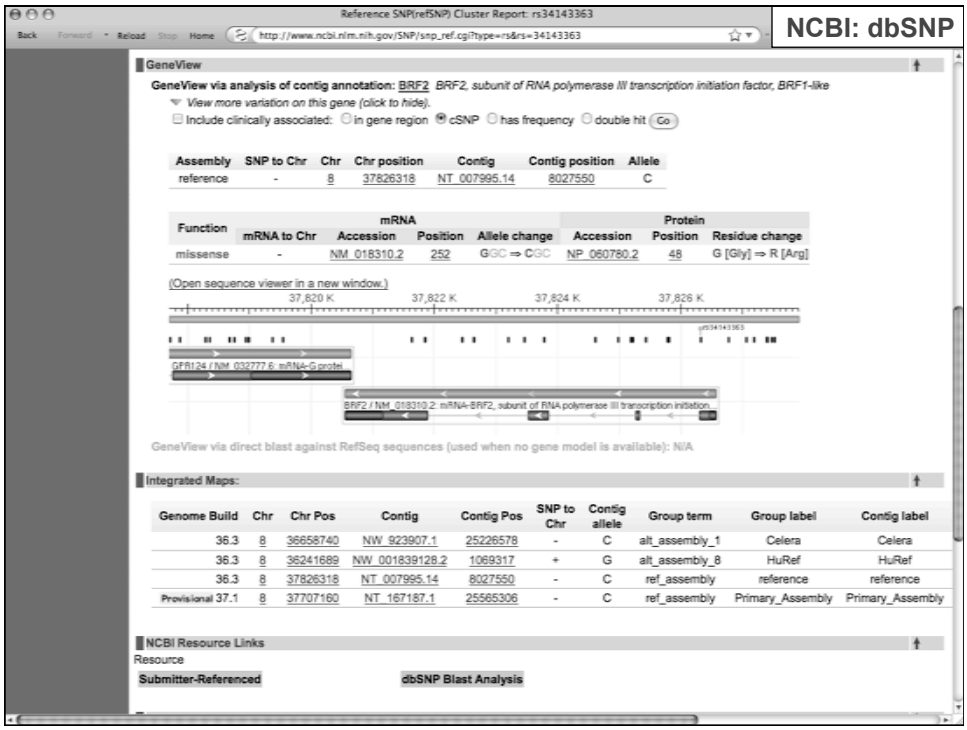
Zoom in to view greater detail




Map Viewer zoomed-in view
Master Map: Genes On Sequence
Region Displayed: 37,700,200-37,710,200 bp
Summary of Maps
Download/View Sequence/Evidence
master map
translated exon
untranslated exon
exon
intron
transcription
GPR124
BRF2


Map Viewer SNP map
Master Map: Variation
Region Displayed: 37,700,200-37,710,200 bp
Summary of Maps
Download/View Sequence/Evidence
click
L: Locus
T: Transcript
C: Coding region

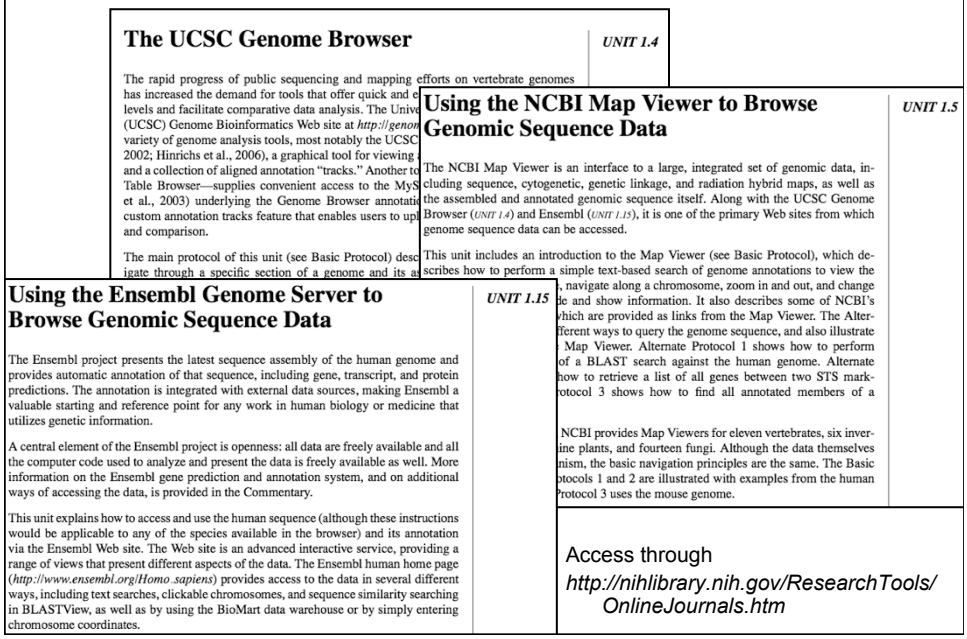

NCBI: dbSNP
Reference SNP(refSNP) Cluster Report: rs34143363
Single Nucleotide Polymorphism
PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books SNP
Search for SNP on NCBI Reference Assembly
Reference SNP(refSNP) Cluster Report: rs34143363
RefSNP
Organism: human (Homo sapiens)
Molecule Type: Genomic
Created/Updated in build: 126/130
Map to Genome Build: 36.3
Allele
Variation Class: SNP: single nucleotide polymorphism
RefSNP Alleles: C/G
Ancestral Allele: Not available
Clinical Association: unknown
HGVS Names
NM_018310.2:c.142G>C
NP_060780.2:p.Gly48Arg
NT_007995.14:g.8027550C>G
SNP Details are organized in the following sections:
Submission Fasta Resource GeneView Map Diversity Validation
Submitter records for this RefSNP Cluster
Fasta sequence (Legend)


NCBI: dbSNP
Reference SNP(refSNP) Cluster Report: rs34143363
GeneView
GeneView via analysis of contig annotation: BRF2 BRF2, subunit of RNA polymerase III transcription initiation factor, BRF1-like
Assembly SNP to Chr Chr Chr position Contig Contig position Allele
reference - 8 37826318 NT_007995.14 8027550 C
Function mRNA to Chr mRNA Accession mRNA Position mRNA Allele change Protein Accession Protein Position Protein Residue change
missense - NM_018310.2 252 GGC ⇒ CGC NP_060780.2 48 G [Gly] ⇒ R [Arg]
(Open sequence viewer in a new window.)
Integrated Maps:
Genome Build Chr Chr Pos Contig Contig Pos SNP to Chr Contig allele Group term Group label Contig label
36.3 8 36658740 NW_923907.1 25226578 - C alt_assembly_1 Celera Celera
36.3 8 36241689 NW_001839128.2 1069317 + G alt_assembly_8 HuRef HuRef
36.3 8 37826318 NT_007995.14 8027550 - C ref_assembly reference reference
Provisional 37.1 8 37707160 NT_167187.1 25565306 - C ref_assembly Primary_Assembly Primary_Assembly
NCBI Resource Links
Resource
Submitter-Referenced
dbSNP Blast Analysis

Additional resources

- UCSC Human Genome Browser User Guide
  http://genome.ucsc.edu/goldenPath/help/
- Ensembl Tutorials and Worked Examples
  http://www.ensembl.org/info/website/tutorials/
- NCBI MapViewer Help
  http://www.ncbi.nlm.nih.gov/mapview/static/MapViewerHelp.html

Current Protocols in Bioinformatics



The UCSC Genome Browser *UNIT 1.4*

The rapid progress of public sequencing and mapping efforts on vertebrate genomes has increased the demand for tools that offer quick and e levels and facilitate comparative data analysis. The Unive (UCSC) Genome Bioinformatics Web site at *http://genom* variety of genome analysis tools, most notably the UCSC 2002; Hinrichs et al., 2006), a graphical tool for viewing and a collection of aligned annotation "tracks." Another to Table Browser—supplies convenient access to the MyS et al., 2003) underlying the Genome Browser annotati custom annotation tracks feature that enables users to up and comparison.

The main protocol of this unit (see Basic Protocol) desc igate through a specific section of a genome and its as

Using the NCBI Map Viewer to Browse Genomic Sequence Data *UNIT 1.5*

The NCBI Map Viewer is an interface to a large, integrated set of genomic data, including sequence, cytogenetic, genetic linkage, and radiation hybrid maps, as well as the assembled and annotated genomic sequence itself. Along with the UCSC Genome Browser (*UNIT 1.4*) and Ensembl (*UNIT 1.15*), it is one of the primary Web sites from which genome sequence data can be accessed.

This unit includes an introduction to the Map Viewer (see Basic Protocol), which describes how to perform a simple text-based search of genome annotations to view the , navigate along a chromosome, zoom in and out, and change de and show information. It also describes some of NCBI's which are provided as links from the Map Viewer. The Alter- ferent ways to query the genome sequence, and also illustrate Map Viewer. Alternate Protocol 1 shows how to perform of a BLAST search against the human genome. Alternate how to retrieve a list of all genes between two STS mark- rotocol 3 shows how to find all annotated members of a

NCBI provides Map Viewers for eleven vertebrates, six inver- ine plants, and fourteen fungi. Although the data themselves nism, the basic navigation principles are the same. The Basic otocols 1 and 2 are illustrated with examples from the human Protocol 3 uses the mouse genome.

Using the Ensembl Genome Server to Browse Genomic Sequence Data *UNIT 1.15*

The Ensembl project presents the latest sequence assembly of the human genome and provides automatic annotation of that sequence, including gene, transcript, and protein predictions. The annotation is integrated with external data sources, making Ensembl a valuable starting and reference point for any work in human biology or medicine that utilizes genetic information.

A central element of the Ensembl project is openness: all data are freely available and all the computer code used to analyze and present the data is freely available as well. More information on the Ensembl gene prediction and annotation system, and on additional ways of accessing the data, is provided in the Commentary.

This unit explains how to access and use the human sequence (although these instructions would be applicable to any of the species available in the browser) and its annotation via the Ensembl Web site. The Web site is an advanced interactive service, providing a range of views that present different aspects of the data. The Ensembl human home page (*http://www.ensembl.org/Homo_sapiens*) provides access to the data in several different ways, including text searches, clickable chromosomes, and sequence similarity searching in BLASTView, as well as by using the BioMart data warehouse or by simply entering chromosome coordinates.

Access through
http://nihlibrary.nih.gov/ResearchTools/OnlineJournals.htm