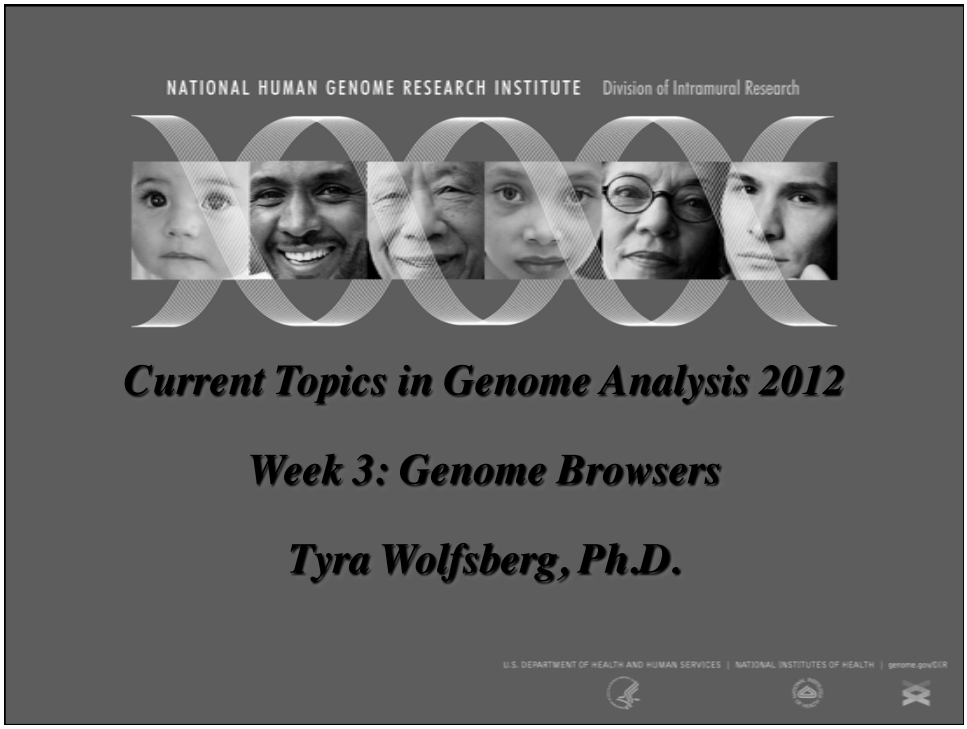
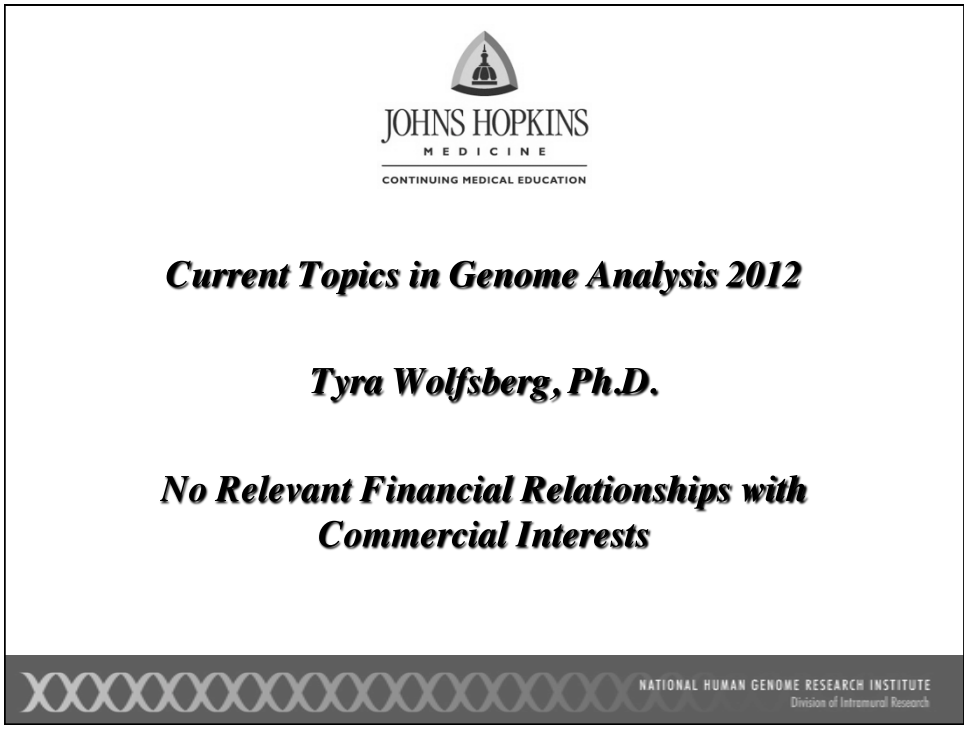

NATIONAL HUMAN GENOME RESEARCH INSTITUTE Division of Intramural Research
Current Topics in Genome Analysis 2012
Week 3: Genome Browsers
Tyra Wolfsberg, Ph.D.
U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES | NATIONAL INSTITUTES OF HEALTH | genome.gov/DIR


JOHNS HOPKINS
MEDICINE
CONTINUING MEDICAL EDUCATION
Current Topics in Genome Analysis 2012
Tyra Wolfsberg, Ph.D.
No Relevant Financial Relationships with Commercial Interests
NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research

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/

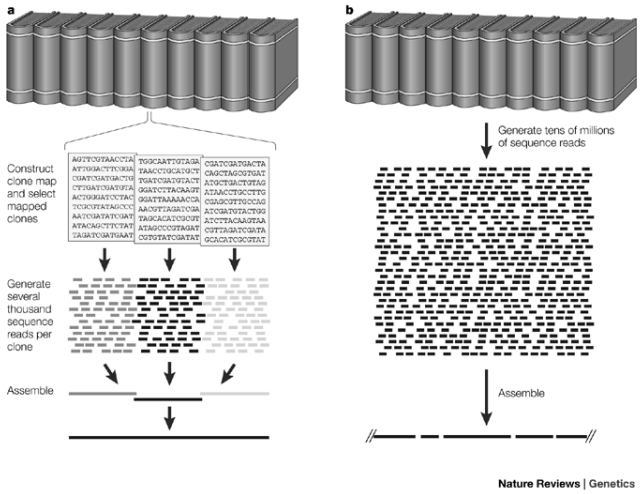
Types of data integrated in genome browsers

- 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
  - Non-coding functional elements

Overview of genome sequencing strategies

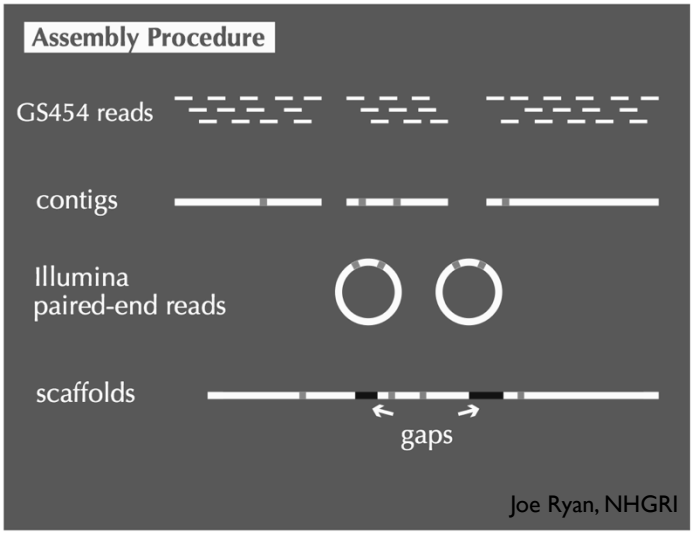
Clone-by-clone shotgun sequencing

Whole-genome shotgun sequencing



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

Next Generation genome assemblies



Genome Sequence Assemblies

- Complex algorithms needed to incorporate all sequence data
- Assemblies updated periodically as new sequence becomes available
  - Mouse, human, and zebrafish (future) genomes assembled by the Genome Reference Consortium (GRC)
  - 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-preview.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/GRCh37/ hg19	Build 37.3	GRCh37
Mouse	Yes	July 2007 (NCBI37/ mm9)	Build 37.2	NCBIM37
Dog	NO	May 2005 (Broad/ canFam2) canFam3 at *genome-preview.cse.ucsc.edu*	Build 3.1/ CanFam 3.1	BROAD2; CanFam 2.0
Zebrafish	Yes	Jul. 2010 (Zv9/ danRer7)	Zv9	Zv9

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.

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                     /gene_synonym="PS1TP5BP1"
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                     actin, cytoplasmic 1"
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                     HTVPIYEGYALPHAILRLDLAGRDLTDYLMKILTERGYSFTTTAEREIVRDIKEKLCY
                     VALDFEQEMATAASSSSLEKSYELPDGQVITIGNERFRCPEALFQPSFLGMESCGIHE
                     TTFNSIMKCDVDIRKDLYANTVLSGGTTMYPGIADRMQKEITALAPSTMKIKIIAPPE
                     RKYSVWIGGSILASLSTFQQMWISKQEYDESGPSIVHRKCF"

ORIGIN
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       61 gtccacaccc gccgccagct caccatggat gatgatatcg ccgcgctcgt cgtcgacaac
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```

Beta actin mRNA RefSeq

UCSC

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

http://genome.ucsc.edu

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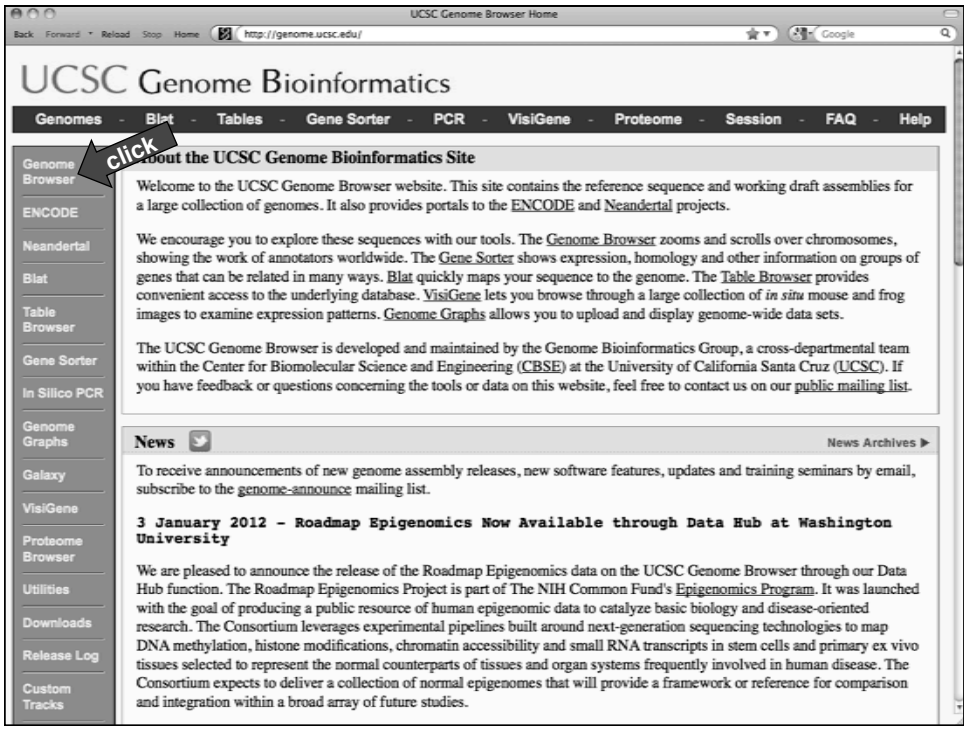
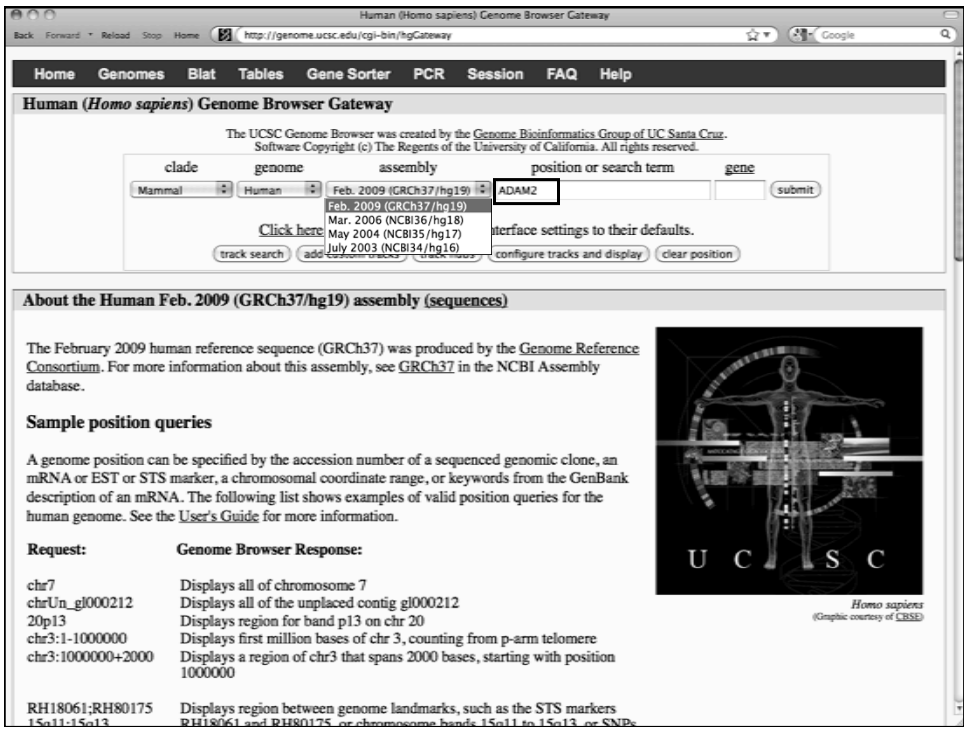
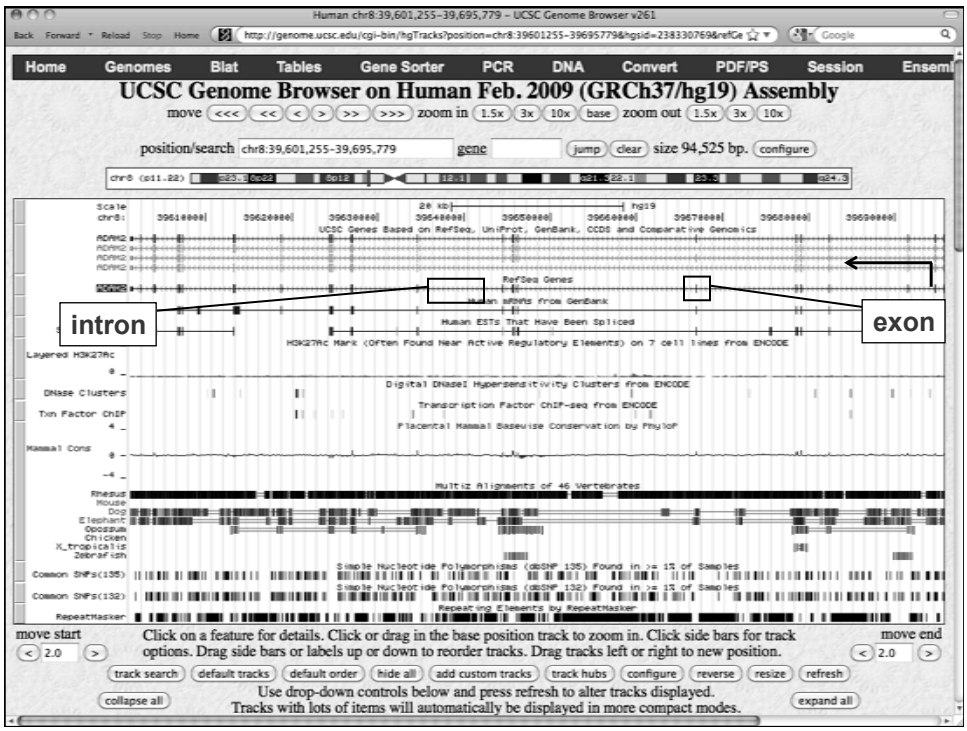
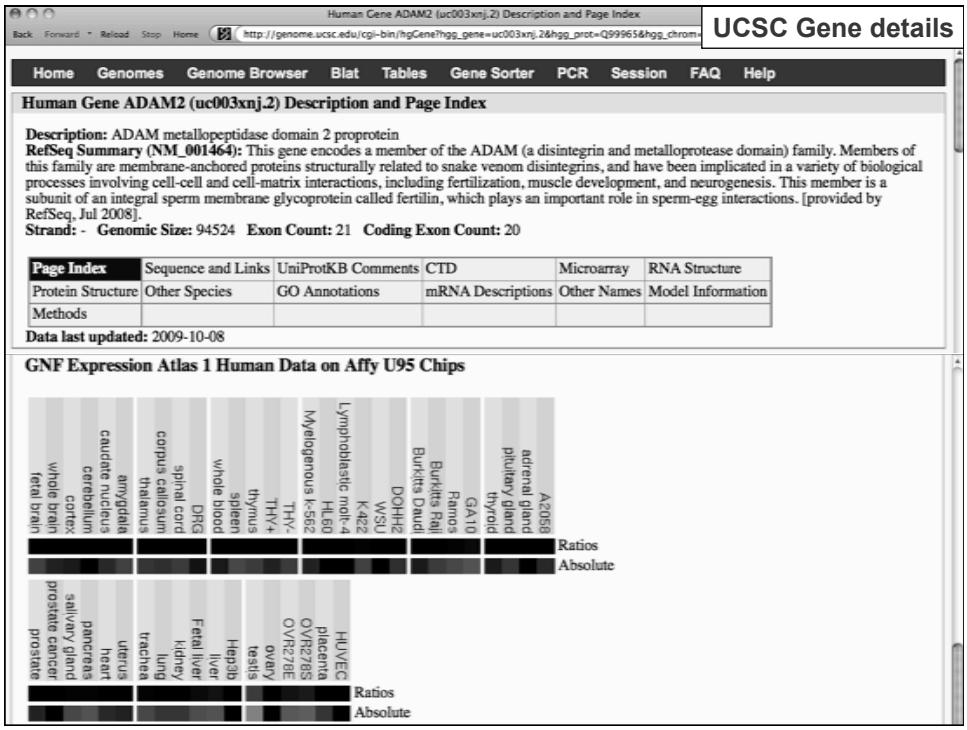
Division of Intramural Research

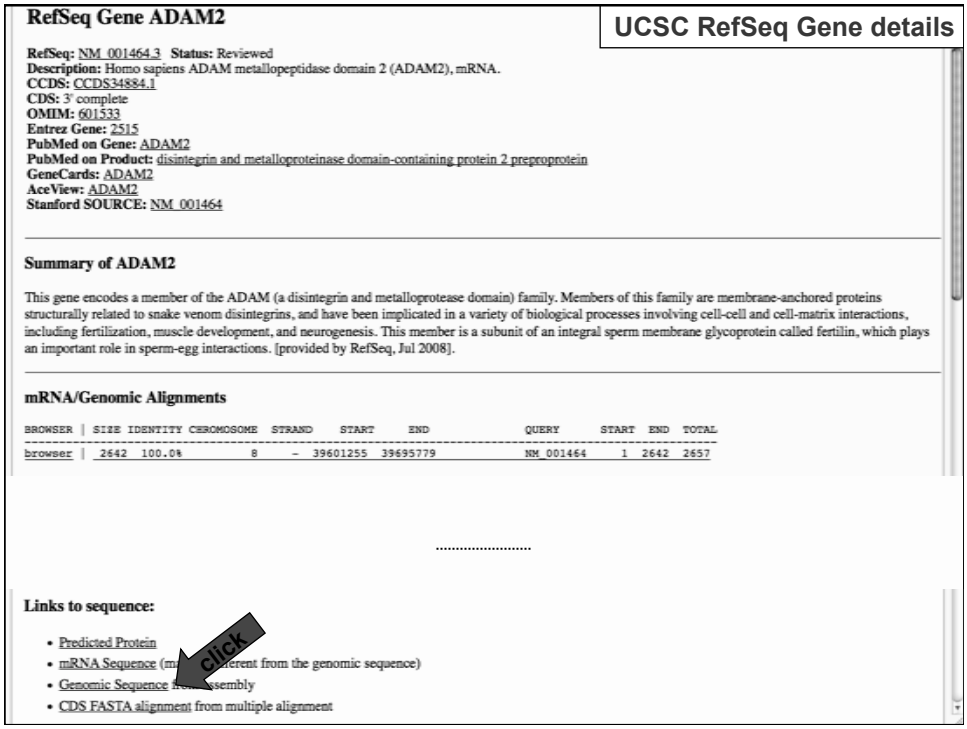


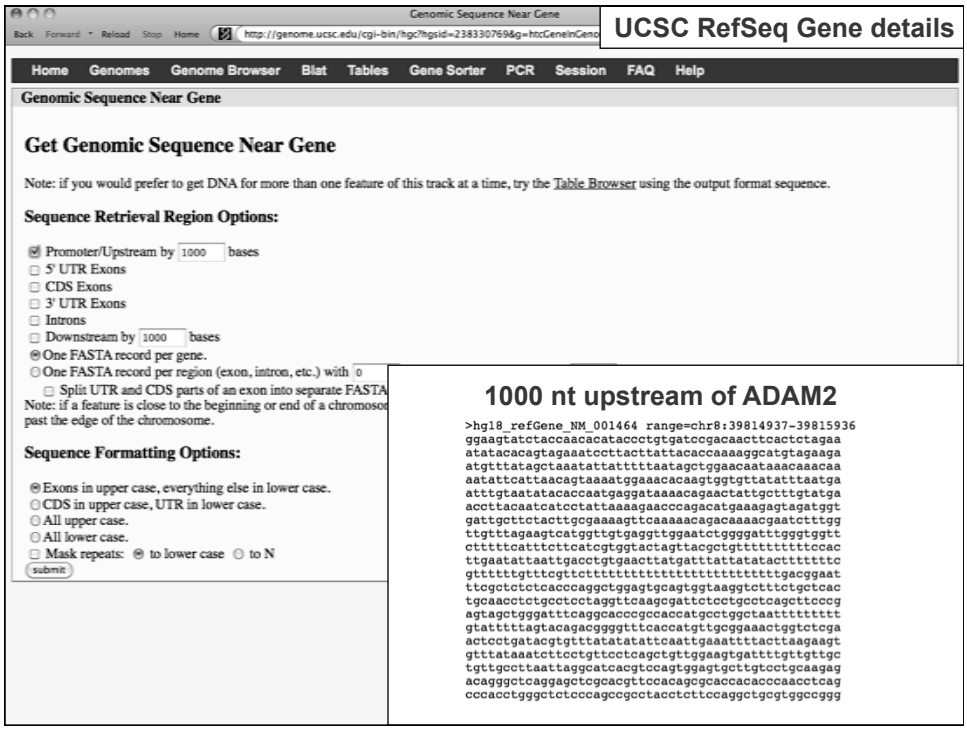

Human (Homo sapiens) Genome Browser Gateway
http://genome.ucsc.edu/cgi-bin/hgGateway
Home Genomes Blat Tables Gene Sorter PCR Session FAQ Help
Human (Homo sapiens) Genome Browser Gateway
The UCSC Genome Browser was created by the Genome Bioinformatics Group of UC Santa Cruz.
Software Copyright (c) The Regents of the University of California. All rights reserved.
clade genome assembly position or search term gene
Mammal Human Feb. 2009 (GRCh37/hg19) ADAM2 submit
Feb. 2009 (GRCh37/hg19)
Mar. 2006 (NCBI36/hg18)
May 2004 (NCBI35/hg17)
July 2003 (NCBI34/hg16)
Click here
track search
configure tracks and display
clear position
About the Human Feb. 2009 (GRCh37/hg19) assembly (sequences)
The February 2009 human reference sequence (GRCh37) was produced by the Genome Reference Consortium. For more information about this assembly, see GRCh37 in the NCBI Assembly database.
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, 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
chrUn_gl000212 Displays all of the unplaced contig gl000212
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 genome landmarks, such as the STS markers
U C S C
Homo sapiens
(Graphic courtesy of CBSE)


Human ADAM2 – UCSC Genome Browser v261
http://genome.ucsc.edu/cgi-bin/hgTracks?hgHubConnect.destUrl=..%2Fcgi-bin%2FhgTracks&clade=mamm
UCSC Genes
ADAM2 (uc011lck.1) at chr8:39601256-39695779 - ADAM metallopeptidase domain 2 proprotein
ADAM2 (uc003xnl.2) at chr8:39601256-39695779 - ADAM metallopeptidase domain 2 proprotein
ADAM2 (uc003xnk.2) at chr8:39601256-39695779 - ADAM metallopeptidase domain 2 proprotein
ADAM2 (uc003xnj.2) at chr8:39601256-39695779 - ADAM metallopeptidase domain 2 proprotein
ADAM28 (uc011laa.1) at chr8:24151580-24212725 - Homo sapiens metalloprotease disintegrin cysteine-rich protein, transmembran
ADAM28 (uc011kzz.1) at chr8:24151580-24193610 - SubName: Full=cDNA FLJ60418, highly similar to ADAM 28 (EC 3.4.24.-) (Adisin
ADAM29 (uc011cki.1) at chr4:175839509-175899330 - ADAM metallopeptidase domain 29 preproprotein
adam23 (uc010ziv.1) at chr2:207310031-207482677 - Homo sapiens mRNA for MDC3, complete cds.
ADAM28 (uc010lua.2) at chr8:24184067-24212725 - ADAM metallopeptidase domain 28 isoform 1
ADAM29 (uc010irr.2) at chr4:175839509-175899330 - ADAM metallopeptidase domain 29 preproprotein
ADAM28 (uc003xdy.2) at chr8:24151580-24212725 - ADAM metallopeptidase domain 28 isoform 1
ADAM28 (uc003xdx.2) at chr8:24151580-24193610 - ADAM metallopeptidase domain 28 isoform 3
ADAM22 (uc003ujp.1) at chr7:87564071-87811339 - ADAM metallopeptidase domain 22 isoform 4
ADAM22 (uc003ujo.2) at chr7:87563702-87826447 - ADAM metallopeptidase domain 22 isoform 3
ADAM22 (uc003ujn.2) at chr7:87563702-87826447 - ADAM metallopeptidase domain 22 isoform 1
ADAM22 (uc003ujm.2) at chr7:87563702-87826447 - ADAM metallopeptidase domain 22 isoform 2
ADAM22 (uc003ujl.1) at chr7:87563702-87811428 - ADAM metallopeptidase domain 22 isoform 5
ADAM22 (uc003ujk.1) at chr7:87563702-87811428 - ADAM metallopeptidase domain 22 isoform 4
ADAM22 (uc003ujj.1) at chr7:87563702-87762113 - ADAM metallopeptidase domain 22 isoform 5
ADAM22 (uc003uji.1) at chr7:87563702-87757991 - ADAM metallopeptidase domain 22 isoform 5
ADAM29 (uc003iud.2) at chr4:175839509-175899330 - ADAM metallopeptidase domain 29 preproprotein
ADAM29 (uc003iuc.2) at chr4:175839509-175899330 - ADAM metallopeptidase domain 29 preproprotein
ADAM23 (uc002vbq.2) at chr2:207308368-207482677 - ADAM metallopeptidase domain 23 preproprotein
ADAM20 (uc001xme.2) at chr14:70989079-71001732 - ADAM metallopeptidase domain 20 preproprotein
ADAM21 (uc001xmd.2) at chr14:70924217-70926621 - ADAM metallopeptidase domain 21 preproprotein
YWHAB (uc002xmu.2) at chr20:43514344-43537160 - tyrosine 3-monooxygenase/tryptophan
YWHAB (uc002xmt.2) at chr20:43514344-43537160 - tyrosine 3-monooxygenase/tryptophan
YWHAQ (uc002qzx.2) at chr2:9724107-9771106 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAQ (uc002qzw.2) at chr2:9724107-9770745 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAE (uc002fsj.2) at chr17:1247836-1303556 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAH (uc003alz.2) at chr22:32340479-32353589 - tyrosine 3-monooxygenase/tryptophan
YWHAE (uc011lhf.1) at chr8:101930804-101963560 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAE (uc011lhe.1) at chr8:101930804-101962799 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAE (uc010mbr.2) at chr8:101930804-101965221 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAE (uc003yjx.2) at chr8:101930804-101965623 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAE (uc003yjw.2) at chr8:101930804-101964357 - tyrosine 3/tryptophan 5 -monooxygenase
YWHAE (uc003yjv.2) at chr8:101930804-101963560 - tyrosine 3/tryptophan 5 -monooxygenase
ADAM21P (uc010ttg.1) at chr14:70712471-70714518 - SubName: Full=ADAM21-like protein;
RefSeq Genes
click
ADAM2 at chr8:39601255-39695779 - (NM_001464) disintegrin and metalloproteinase domain-containing protein 2 preproprotein
ADAM20 at chr14:70989078-71001732 - (NM_003814) disintegrin and metalloproteinase domain-containing protein 20 preproprotein
ADAM21 at chr14:70918874-70926622 - (NM_003813) disintegrin and metalloproteinase domain-containing protein 21 preproprotein


Human chr8:39,601,255-39,695,779 – UCSC Genome Browser v261
Home
Genomes
Blat
Tables
Gene Sorter
PCR
DNA
Convert
PDF/PS
Session
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
intron
exon


UCSC Gene details
Human Gene ADAM2 (uc003xnj.2) Description and Page Index
Home
Genomes
Genome Browser
Blat
Tables
Gene Sorter
PCR
Session
FAQ
Help
Human Gene ADAM2 (uc003xnj.2) 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, Jul 2008].
Strand: - Genomic Size: 94524 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
Data last updated: 2009-10-08
GNF Expression Atlas 1 Human Data on Affy U95 Chips
Ratios
Absolute


UCSC RefSeq Gene details
RefSeq Gene ADAM2
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: disintegrin and metalloproteinase domain-containing protein 2 preproprotein
GeneCards: ADAM2
AceView: ADAM2
Stanford SOURCE: 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, Jul 2008].
mRNA/Genomic Alignments
BROWSER | SIZE IDENTITY CHROMOSOME STRAND START END QUERY START END TOTAL
browser | 2642 100.0% 8 - 39601255 39695779 NM_001464 1 2642 2657
Links to sequence:
Predicted Protein
mRNA Sequence
Genomic Sequence
CDS FASTA alignment from multiple alignment
click


UCSC RefSeq Gene details
Genomic Sequence Near Gene
Home Genomes Genome Browser Blat Tables Gene Sorter PCR Session FAQ Help
Genomic Sequence Near Gene
Get Genomic Sequence Near Gene
Note: if you would prefer to get DNA for more than one feature of this track at a time, try the Table Browser using the output format sequence.
Sequence Retrieval Region Options:
Promoter/Upstream by 1000 bases
5' UTR Exons
CDS Exons
3' UTR Exons
Introns
Downstream by 1000 bases
One FASTA record per gene.
One FASTA record per region (exon, intron, etc.) with 0
Split UTR and CDS parts of an exon into separate FASTA
Note: if a feature is close to the beginning or end of a chromoso
past the edge of the chromosome.
Sequence Formatting Options:
Exons in upper case, everything else in lower case.
CDS in upper case, UTR in lower case.
All upper case.
All lower case.
Mask repeats: to lower case to N
submit
1000 nt upstream of ADAM2
>hg18_refGene_NM_001464 range=chr8:39814937-39815936
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UCSC

Navigating around the Genome Browser

http://genome.ucsc.edu

NATIONAL HUMAN GENOME RESEARCH INSTITUTE
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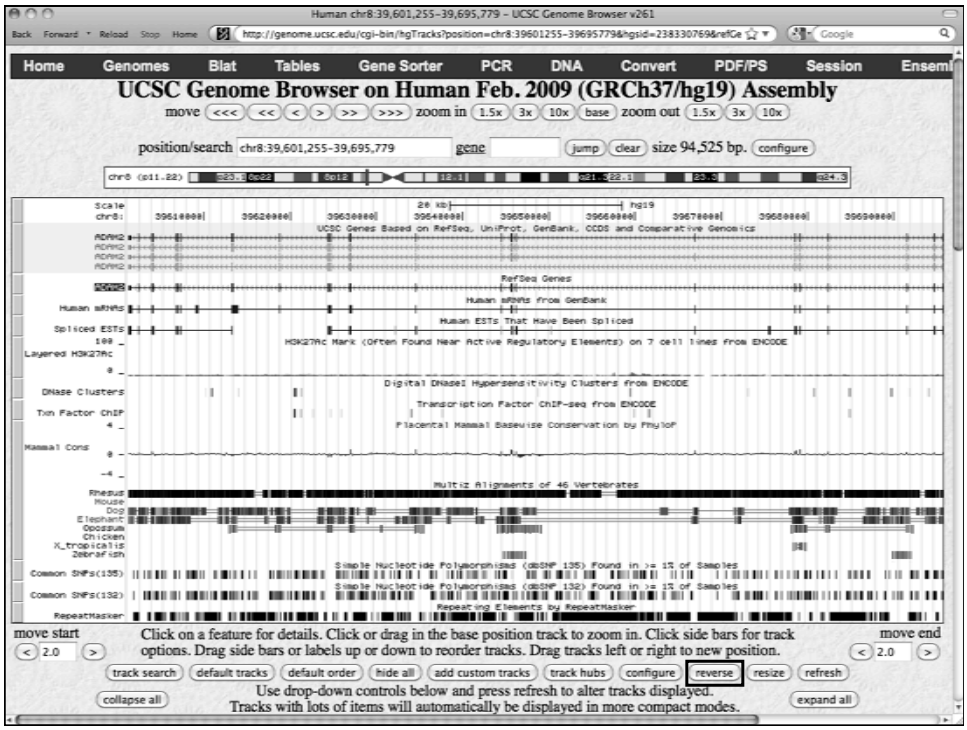
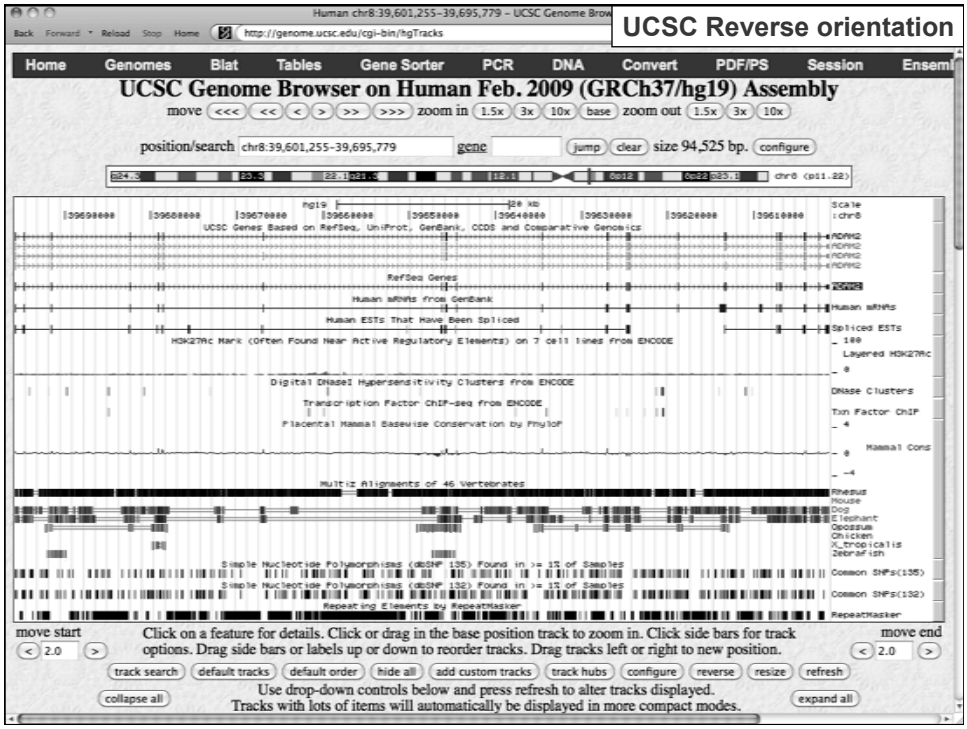
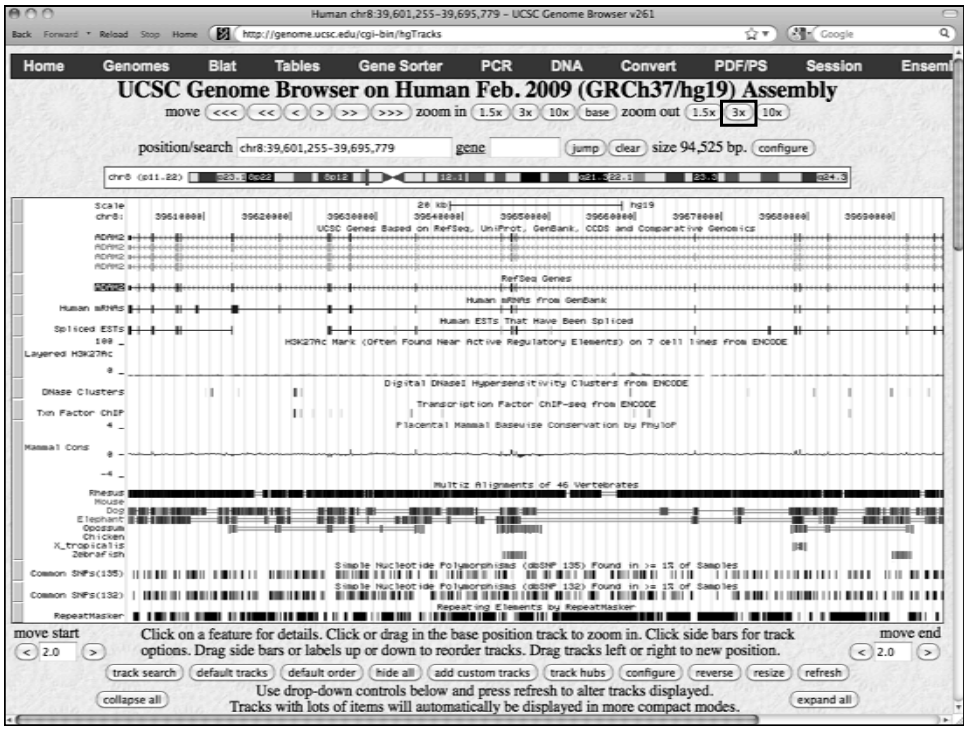



UCSC Reverse orientation
Home
Genomes
Blat
Tables
Gene Sorter
PCR
DNA
Convert
PDF/PS
Session
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
position/search chr8:39,601,255-39,695,779
size 94,525 bp.
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.
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.


Home
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PDF/PS
Session
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
position/search chr8:39,601,255-39,695,779
size 94,525 bp.
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.
Use drop-down controls below and press refresh to alter tracks displayed.
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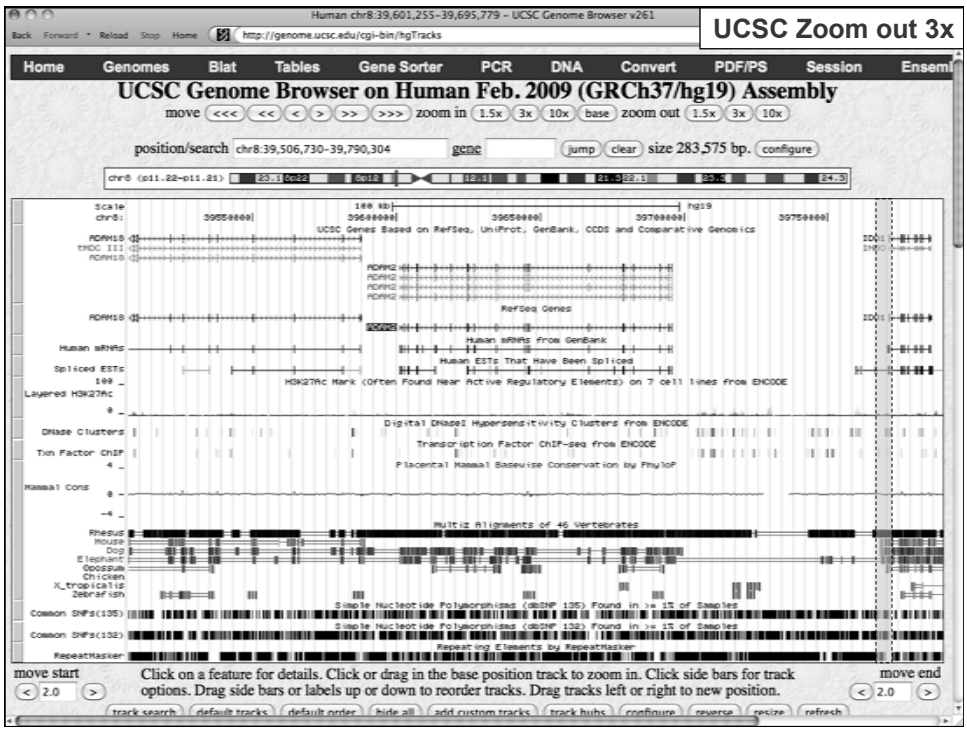
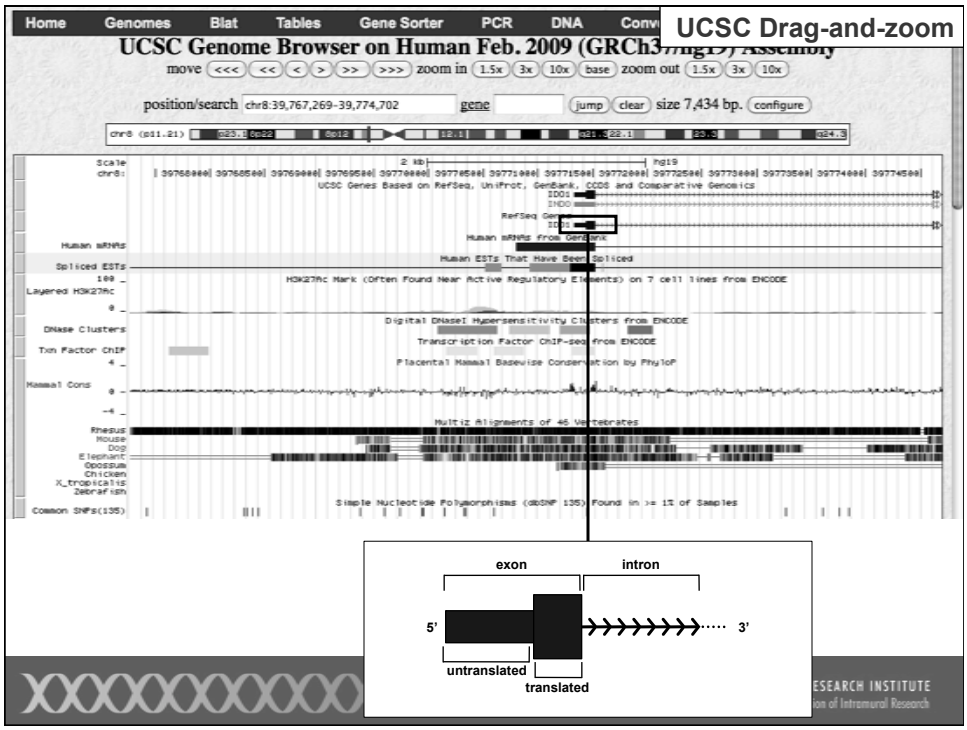

UCSC Zoom out 3x
Human chr8:39,601,255-39,695,779 - UCSC Genome Browser v261
Home
Genomes
Blat
Tables
Gene Sorter
PCR
DNA
Convert
PDF/PS
Session
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
move
zoom in
zoom out
position/search chr8:39,506,730-39,790,304
gene
jump
clear
size 283,575 bp.
configure
UCSC Genes Based on RefSeq, UniProt, GenBank, CCDS and Comparative Genomics
RefSeq Genes
Human mRNAs from GenBank
Human ESTs That Have Been Spliced
H3K27Ac Mark (Often Found Near Active Regulatory Elements) on 7 cell lines from ENCODE
Digital DNaseI Hypersensitivity Clusters from ENCODE
Transcription Factor ChIP-seq from ENCODE
Placental Mammal Basewise Conservation by PhyloP
Multiz Alignments of 46 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP 135) Found in >= 1% of Samples
Simple Nucleotide Polymorphisms (dbSNP 132) Found in >= 1% of Samples
Repeating Elements by RepeatMasker
move start
move end
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.

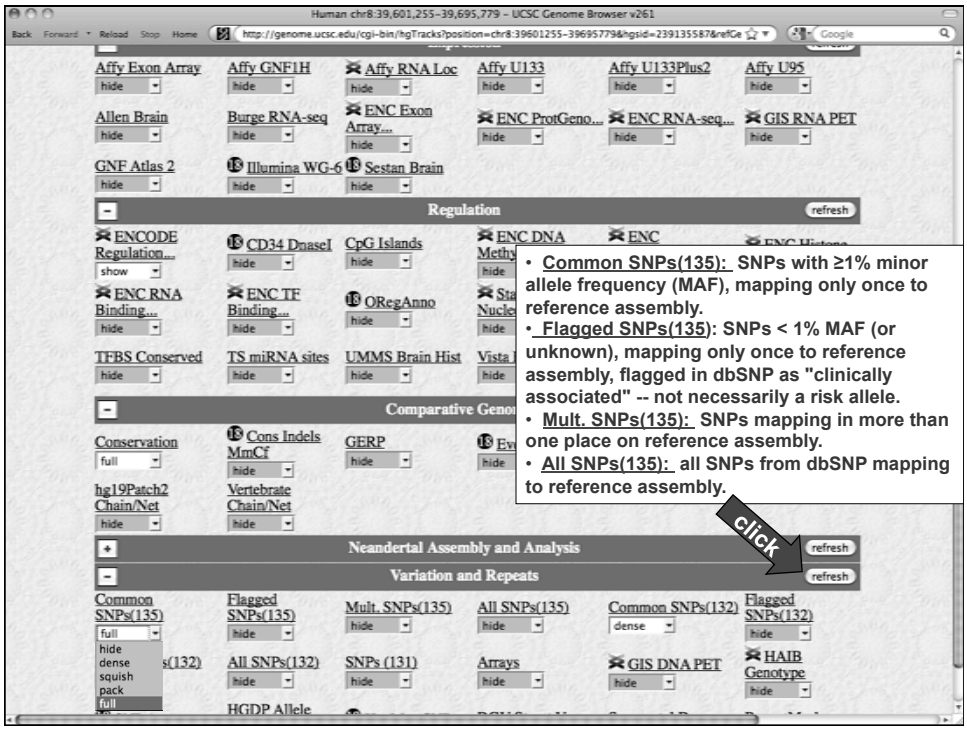

UCSC Drag-and-zoom
Home
Genomes
Blat
Tables
Gene Sorter
PCR
DNA
move
zoom in
zoom out
position/search chr8:39,767,269-39,774,702
gene
jump
clear
size 7,434 bp.
configure
exon
intron
5'
3'
untranslated
translated

UCSC

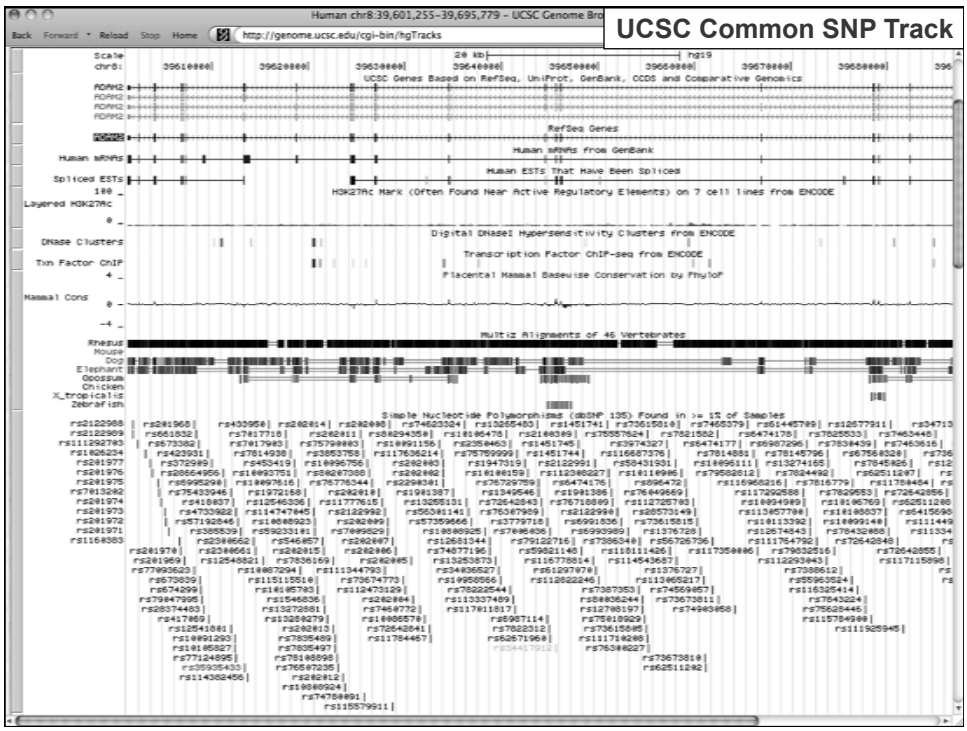
Add a track to the Genome Browser

http://genome.ucsc.edu

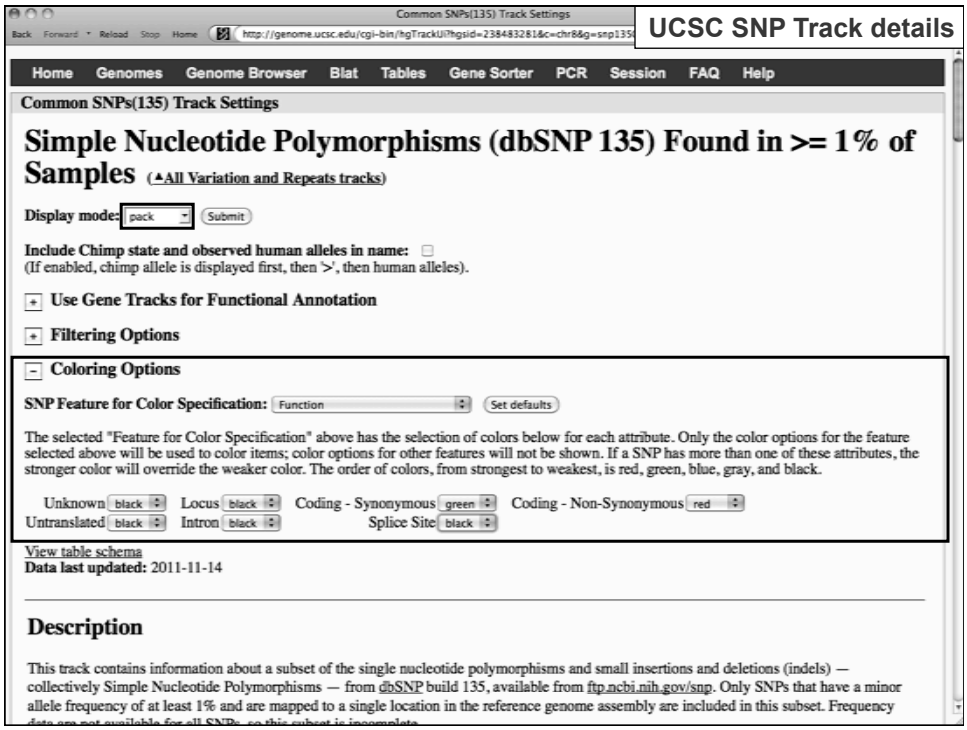
NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research

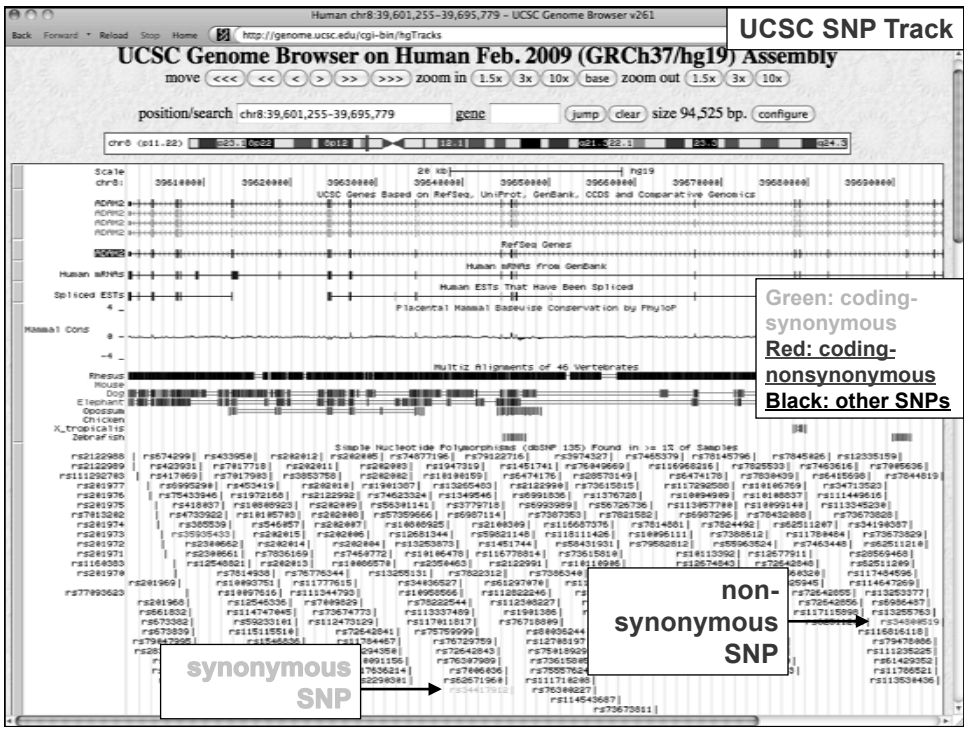


UCSC Common SNP Track



UCSC SNP Track details

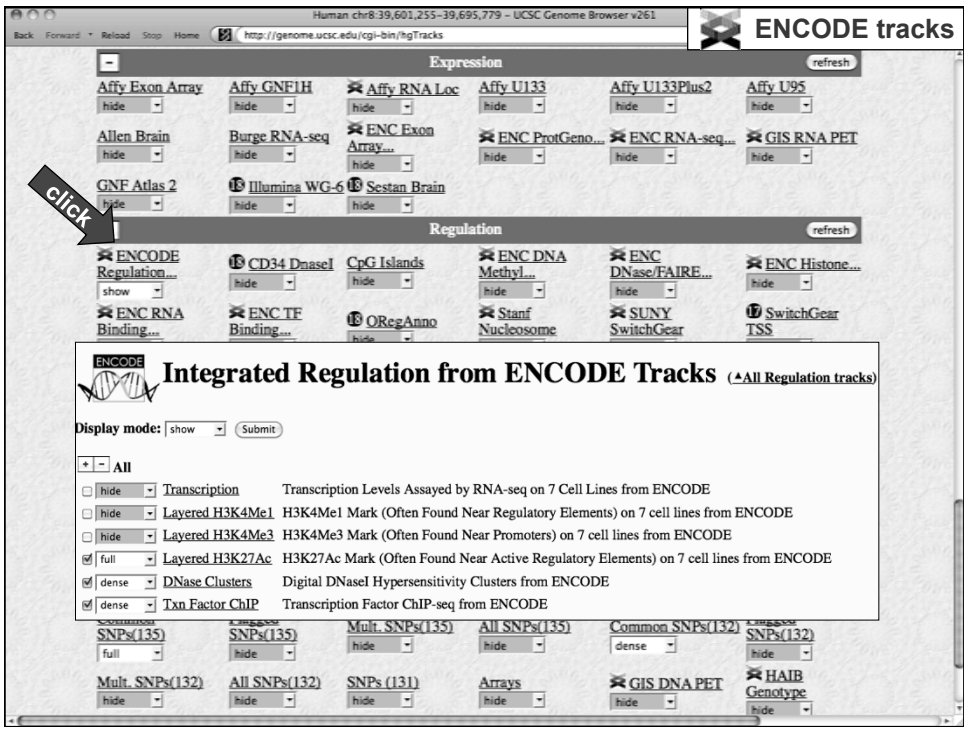


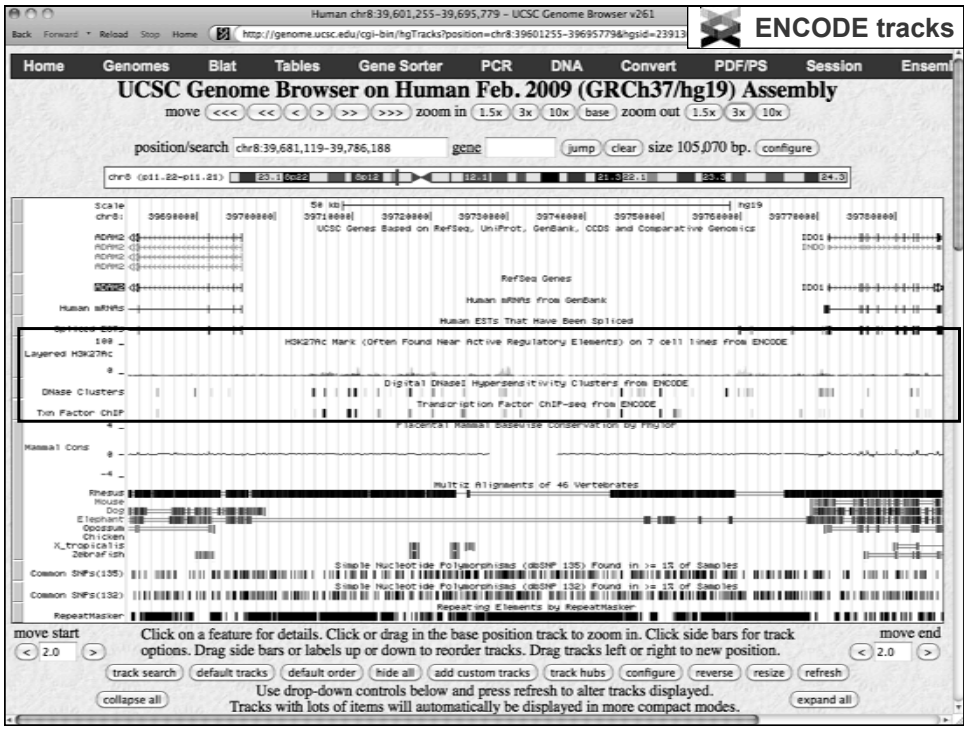


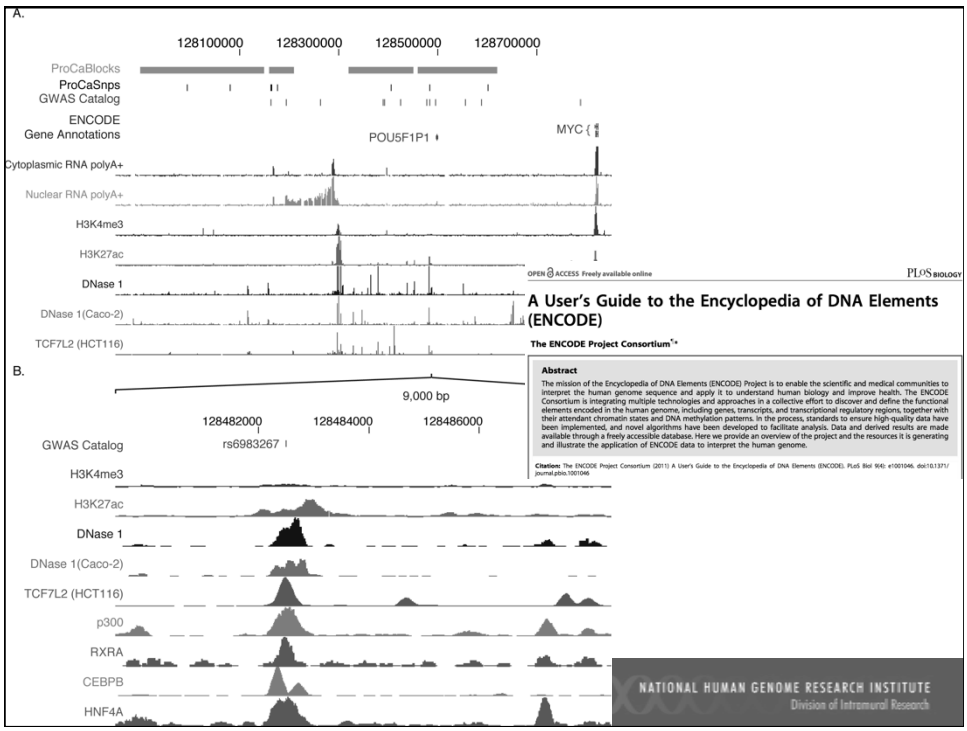
UCSC

ENCODE tracks

http://genome.ucsc.edu


ENCODE tracks
click
Expression
Regulation
Integrated Regulation from ENCODE Tracks
Display mode:
Transcription
Transcription Levels Assayed by RNA-seq on 7 Cell Lines from ENCODE
Layered H3K4Me1
H3K4Me1 Mark (Often Found Near Regulatory Elements) on 7 cell lines from ENCODE
Layered H3K4Me3
H3K4Me3 Mark (Often Found Near Promoters) on 7 cell lines from ENCODE
Layered H3K27Ac
H3K27Ac Mark (Often Found Near Active Regulatory Elements) on 7 cell lines from ENCODE
DNase Clusters
Digital DNaseI Hypersensitivity Clusters from ENCODE
Txn Factor ChIP
Transcription Factor ChIP-seq from ENCODE

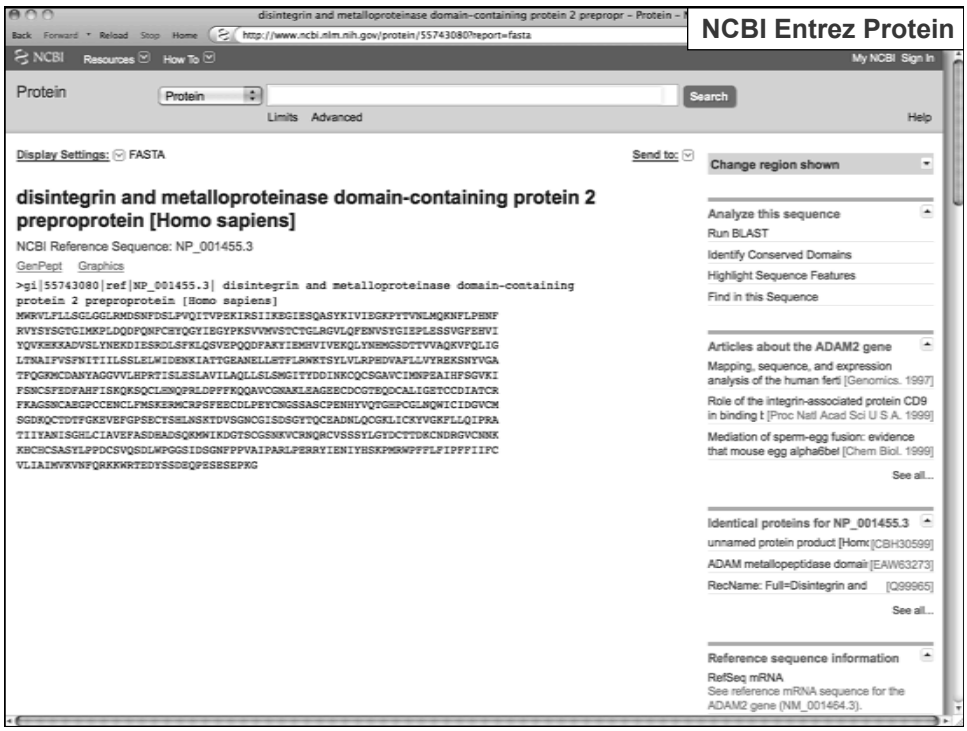

ENCODE tracks
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
position/search chr8:39,681,119-39,786,188
size 105,070 bp.
Layered H3K27Ac
DNase Clusters
Txn Factor ChIP
Common SNPs(135)
Common SNPs(132)
RepeatMasker

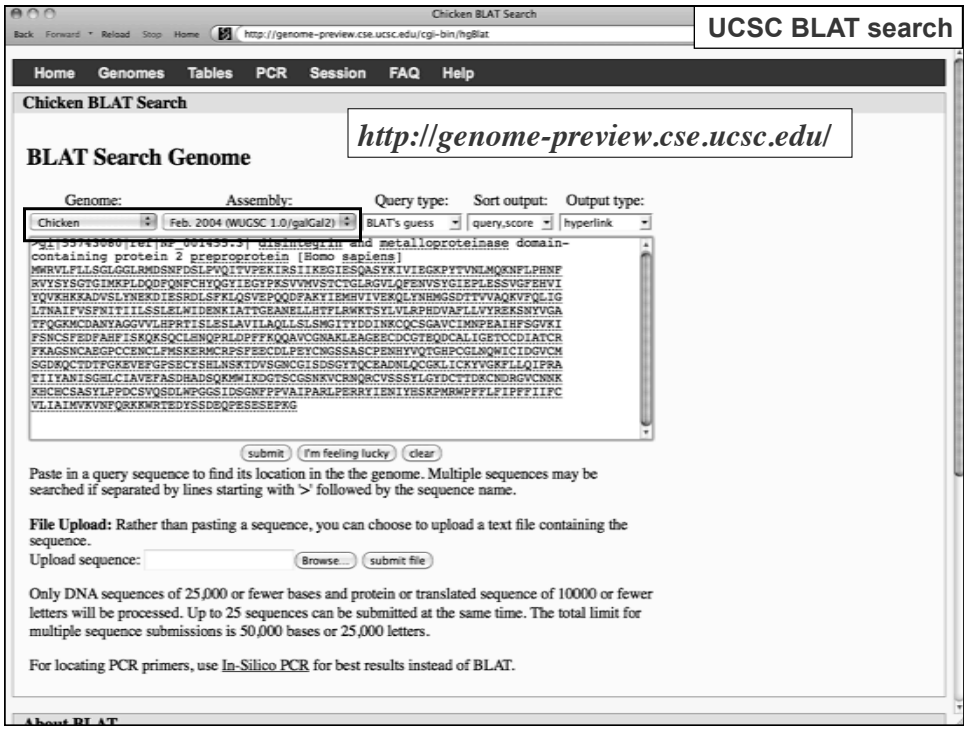


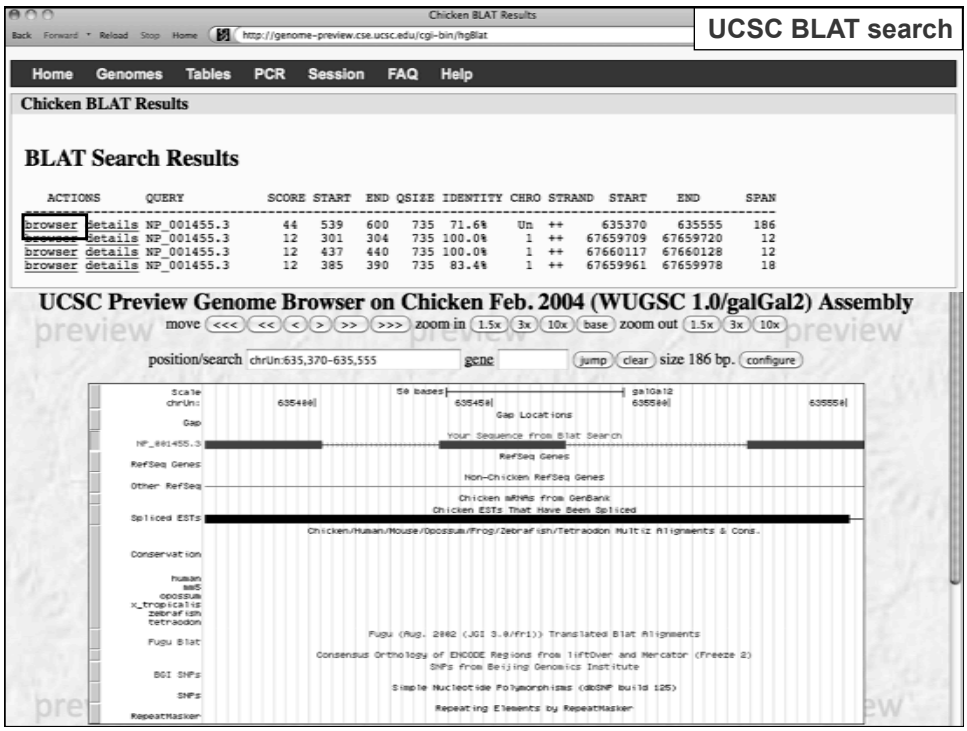
UCSC

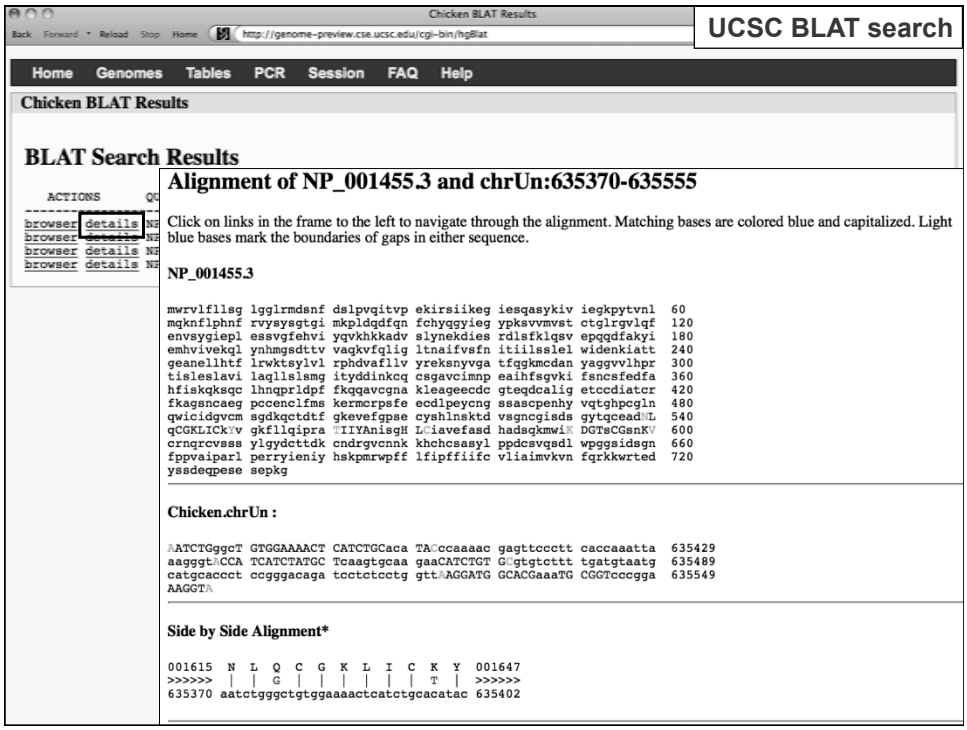
Find a chicken homolog of a human protein

http://genome.ucsc.edu


NCBI Entrez Protein
disintegrin and metalloproteinase domain-containing protein 2 preprotein [Homo sapiens]
NCBI Reference Sequence: NP_001455.3


UCSC BLAT search
http://genome-preview.cse.ucsc.edu/
Chicken BLAT Search
BLAT Search Genome


UCSC BLAT search
Chicken BLAT Results
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 Preview Genome Browser on Chicken Feb. 2004 (WUGSC 1.0/galGal2) Assembly
position/search chrUn:635,370-635,555
RefSeq Genes
Chicken mRNAs from GenBank
Chicken ESTs That Have Been Spliced
Gap Locations
Your Sequence from Blat Search
Non-Chicken RefSeq Genes
Conservation
Fugu Blat
BGI SNPs
SNPs
RepeatMasker


UCSC BLAT search
Chicken BLAT Results
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
Chicken.chrUn :
Side by Side Alignment*
001615 N L Q C G K L I C K Y 001647
635370 aatctgggctgtggaaaactcatctgcacatac 635402

UCSC

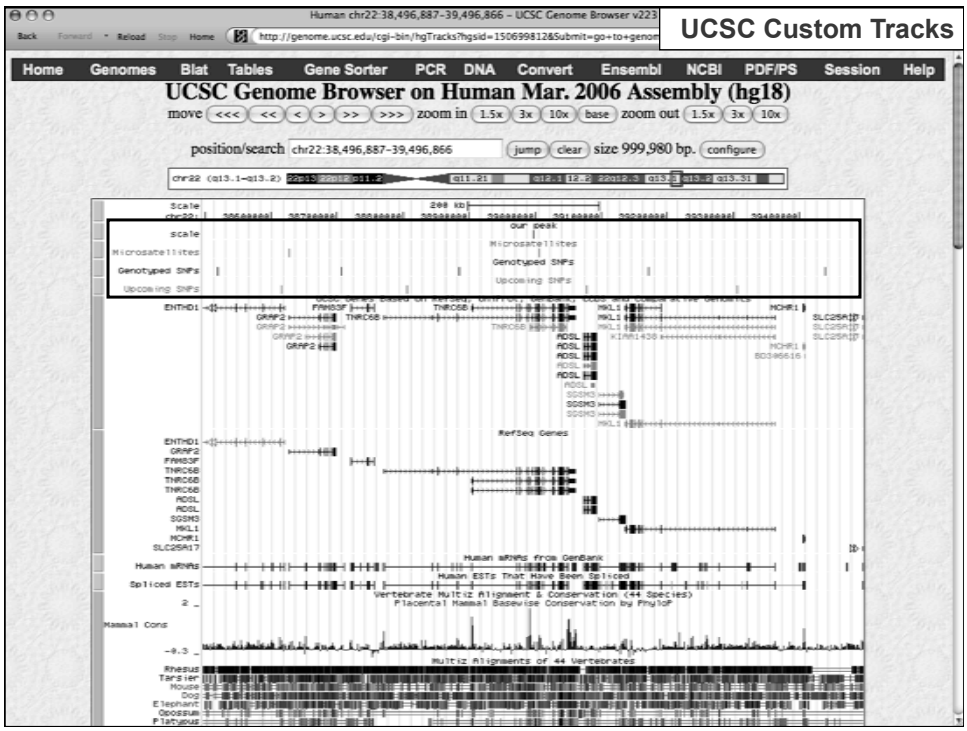
Add your own custom tracks

http://genome.ucsc.edu

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



Types of UCSC custom tracks

- Upload annotation data from your computer
  - Only viewable on the machine from which they were uploaded
  - Discarded after 48 hours
- Post annotation data to your Web site
  - URL, with link to Genome Browser, can be shared with anyone
  - Never discarded
- Create a Session to configure your browser with specific track combinations, including custom tracks
  - Can be shared or non-shared
  - Session persists for 4 months; custom tracks for 48 hours
- Contribute your tracks to the UCSC Genome Browser

http://genome.ucsc.edu/goldenPath/help/hgTracksHelp.html#CustomTracks
http://genome.ucsc.edu/goldenPath/help/hgSessionHelp.html

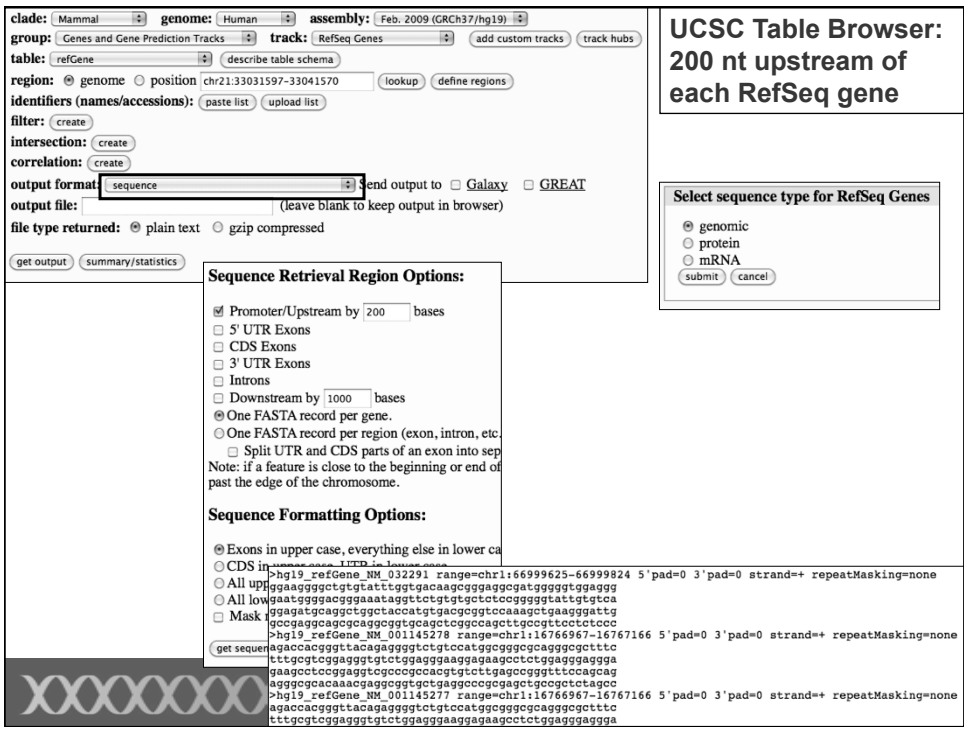
UCSC

Table Browser

http://genome.ucsc.edu

UCSC Table Browser

- Download track in text format
- Retrieve DNA sequence covered by a track
  - Get sequence 1 Kb upstream of each RefSeq gene
- 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

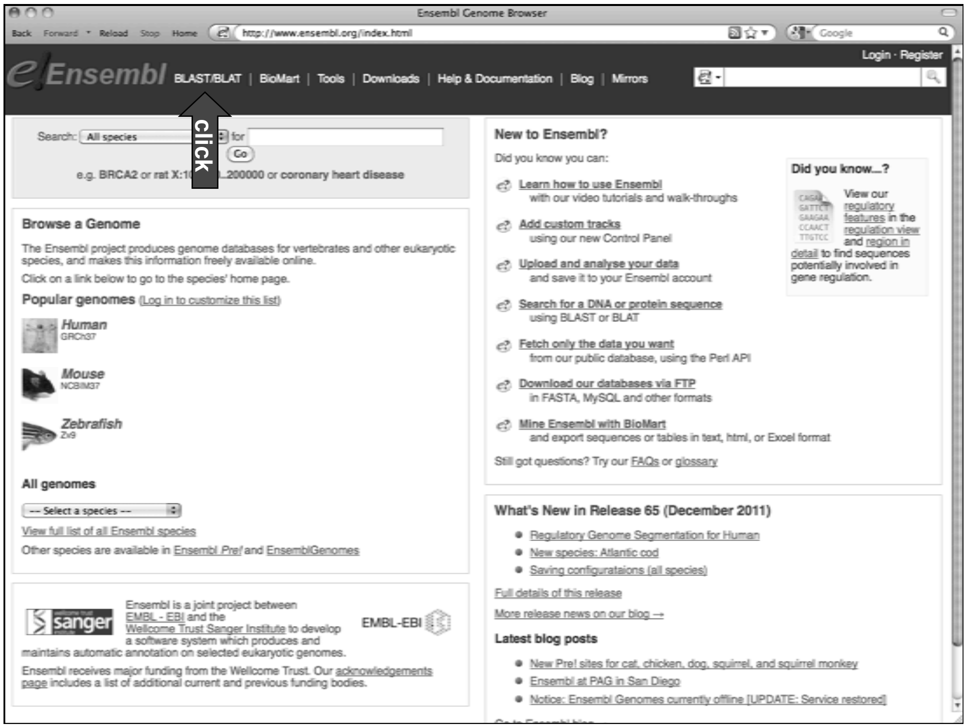


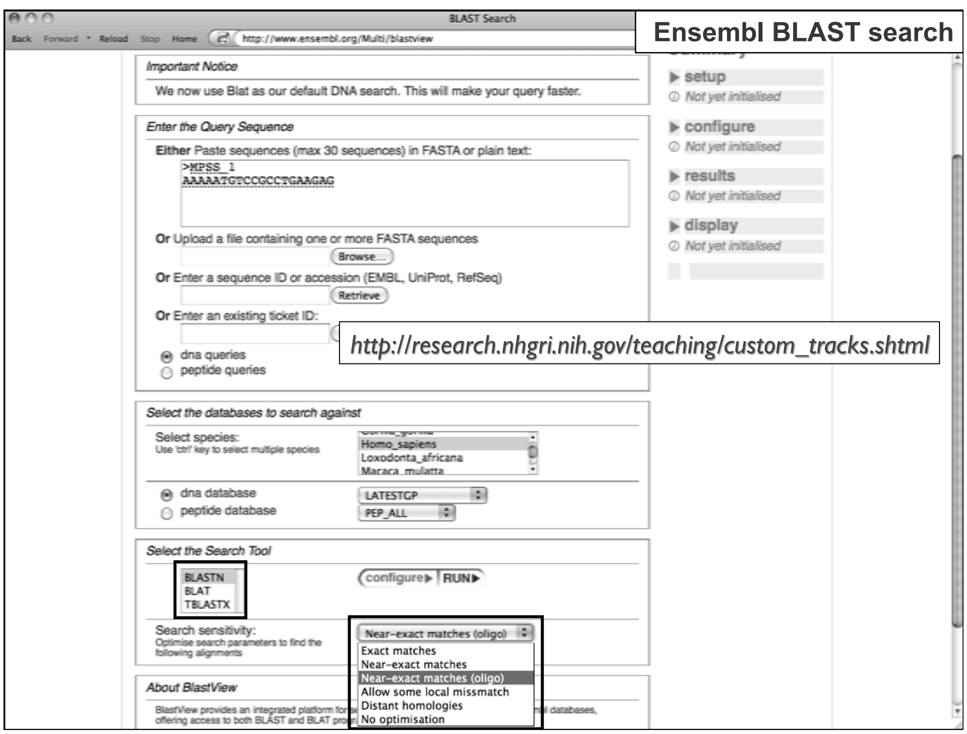
UCSC Table Browser: 200 nt upstream of each RefSeq gene

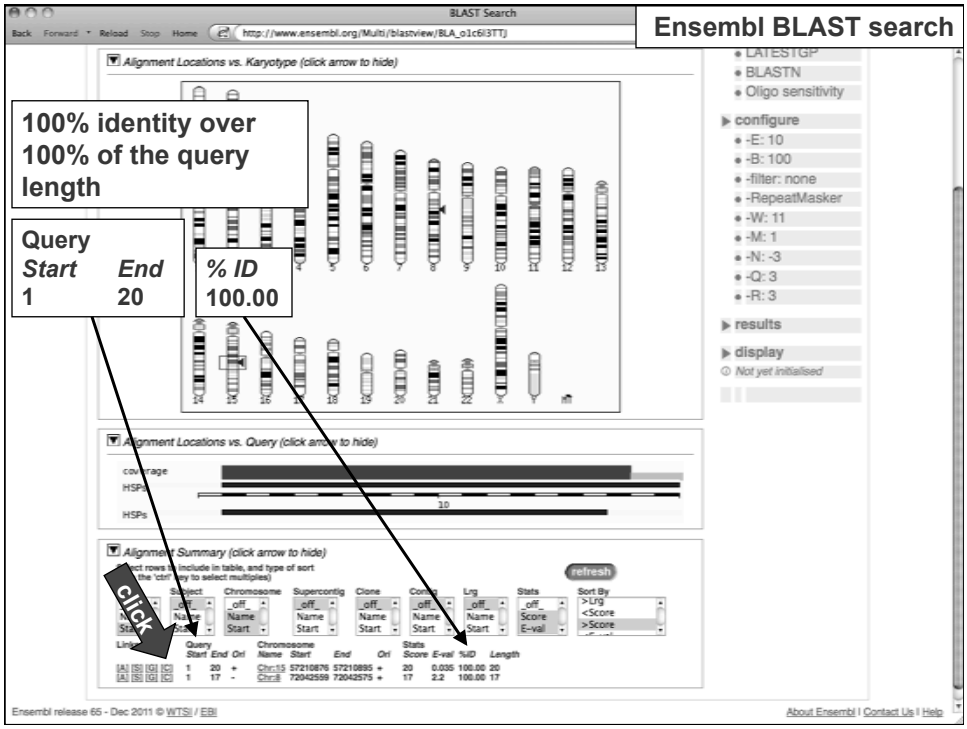
Ensembl

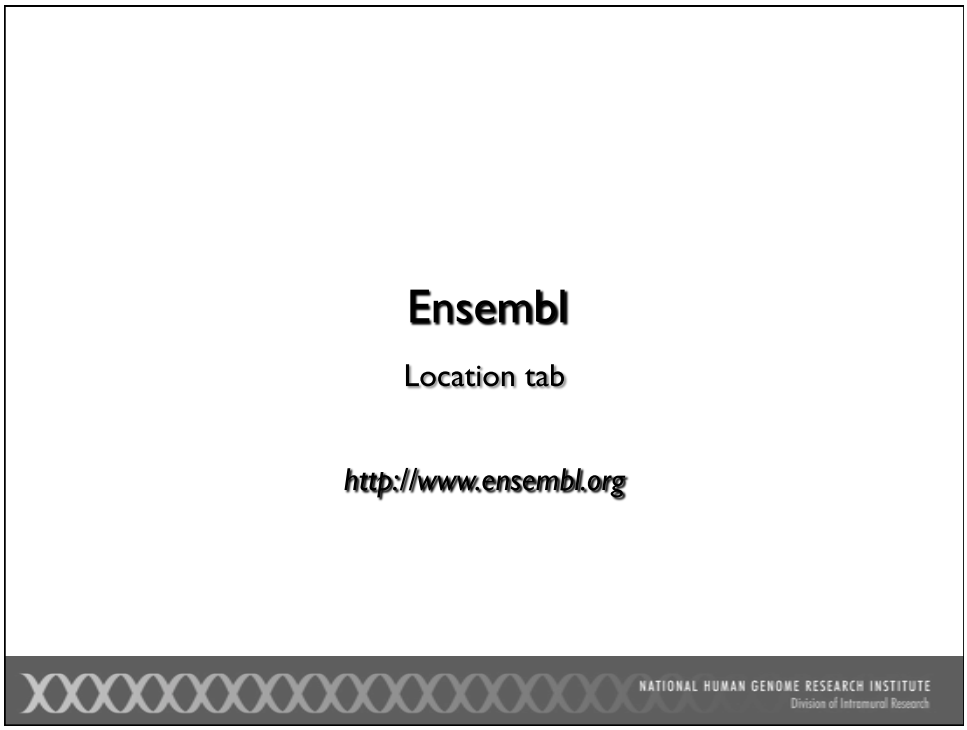
Identify genes that overlap with an oligo tag

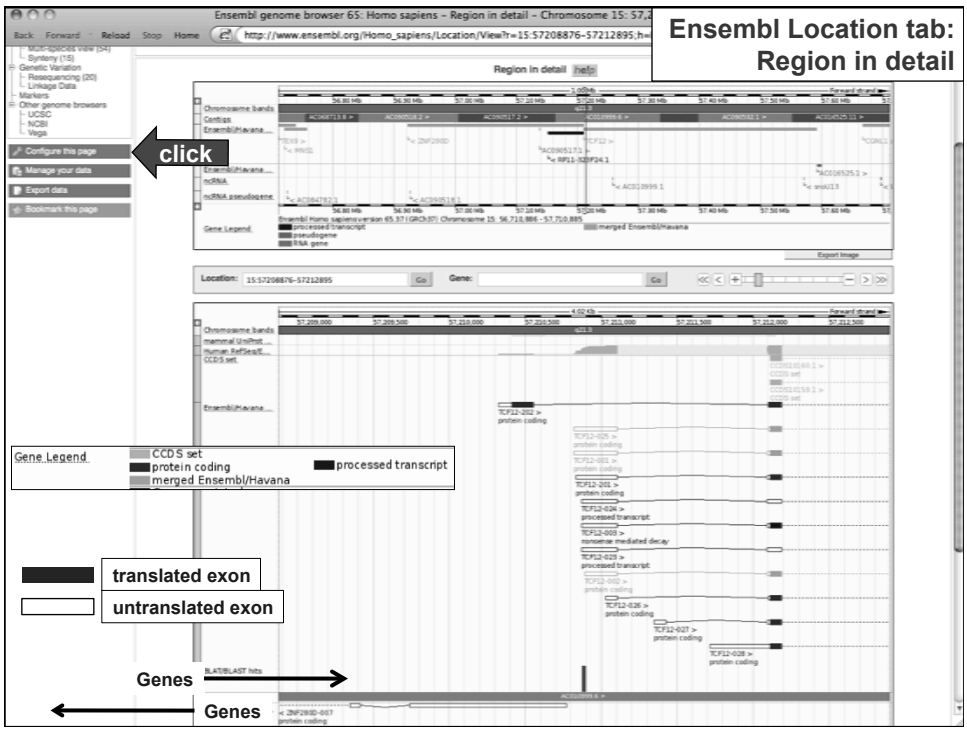
http://www.ensembl.org

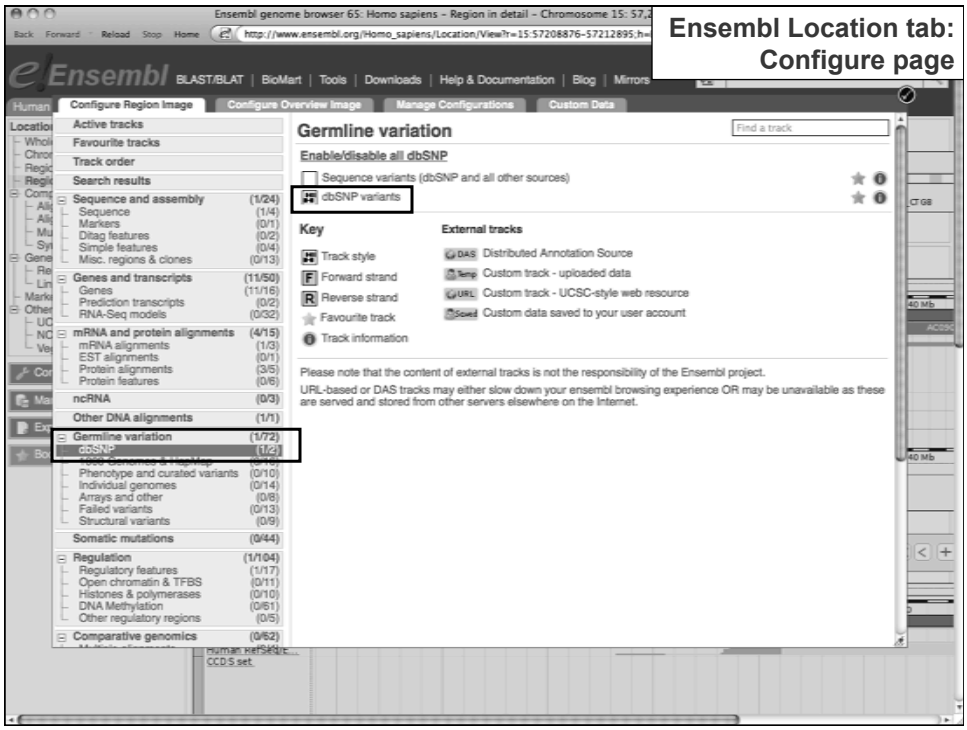

Ensembl Genome Browser
http://www.ensembl.org/index.html
Ensembl
BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog | Mirrors
Login · Register
click
Browse a Genome
Popular genomes
Human
Mouse
Zebrafish
All genomes
New to Ensembl?
Did you know...?
What's New in Release 65 (December 2011)
Latest blog posts


BLAST Search
http://www.ensembl.org/Multi/blastview
Ensembl BLAST search
Important Notice
We now use Blat as our default DNA search. This will make your query faster.
Enter the Query Sequence
>MPSS_1
AAAAATGTCCGCCTGAAGAG
http://research.nhgri.nih.gov/teaching/custom_tracks.shtml
Select the databases to search against
Homo_sapiens
Select the Search Tool
BLASTN
BLAT
TBLASTX
Search sensitivity:
Near-exact matches (oligo)
Exact matches
Near-exact matches
Near-exact matches (oligo)
Allow some local missmatch
Distant homologies
No optimisation
setup
configure
results
display

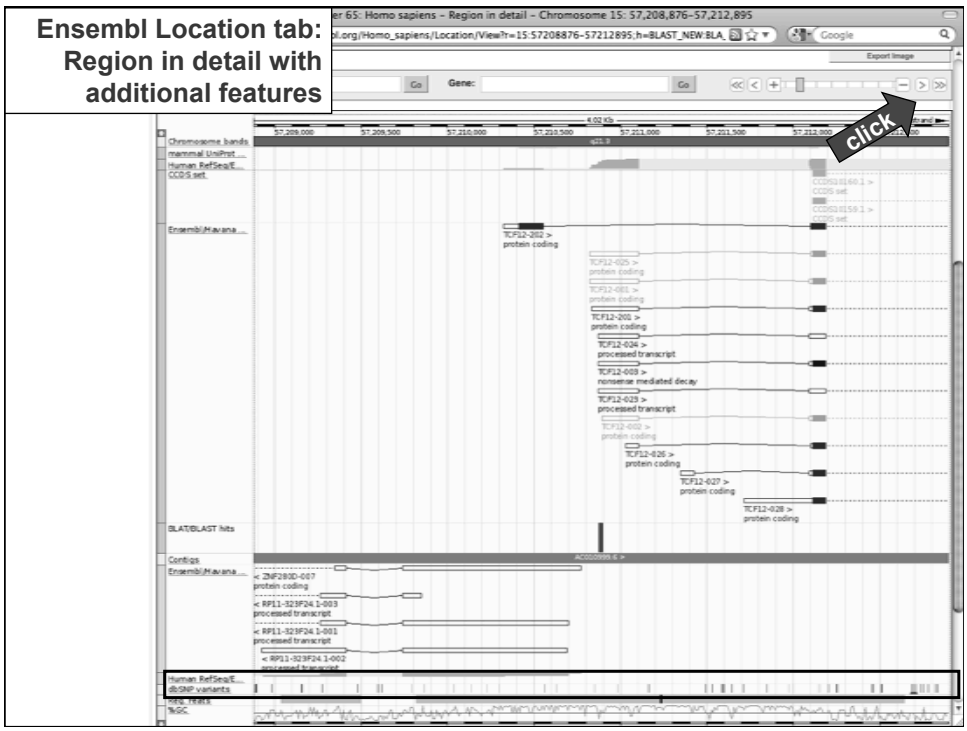

Ensembl BLAST search
100% identity over 100% of the query length
Query
Start
1
End
20
% ID
100.00
click
BLAST Search
Alignment Locations vs. Karyotype (click arrow to hide)
Alignment Locations vs. Query (click arrow to hide)
Alignment Summary (click arrow to hide)
refresh
BLASTN
Oligo sensitivity
configure
-E: 10
-B: 100
-filter: none
-RepeatMasker
-W: 11
-M: 1
-N: -3
-Q: 3
-R: 3
results
display
Not yet initialised


Ensembl
Location tab
http://www.ensembl.org
NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research

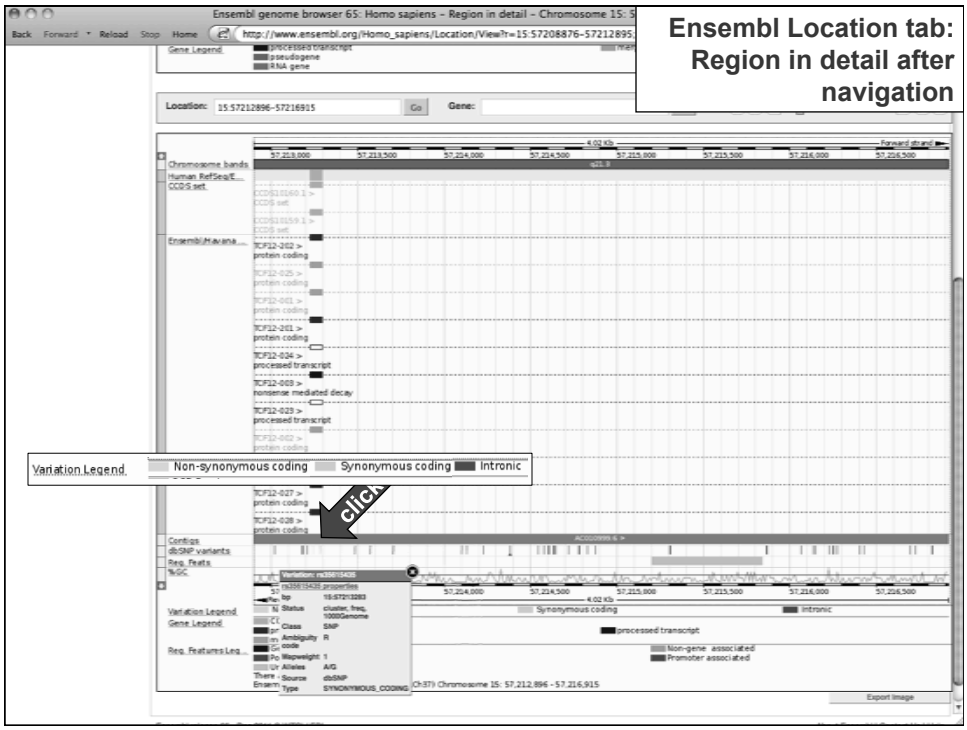

Ensembl Location tab:
Region in detail
click
Gene Legend
CCDS set
protein coding
merged Ensembl/Havana
processed transcript
translated exon
untranslated exon
Genes
Genes


Ensembl Location tab:
Configure page
Germline variation
Enable/disable all dbSNP
Sequence variants (dbSNP and all other sources)
dbSNP variants
Key
External tracks
Track style
Forward strand
Reverse strand
Favourite track
Track information
Distributed Annotation Source
Custom track - uploaded data
Custom track - UCSC-style web resource
Custom data saved to your user account
Please note that the content of external tracks is not the responsibility of the Ensembl project.
URL-based or DAS tracks may either slow down your ensembl browsing experience OR may be unavailable as these are served and stored from other servers elsewhere on the Internet.

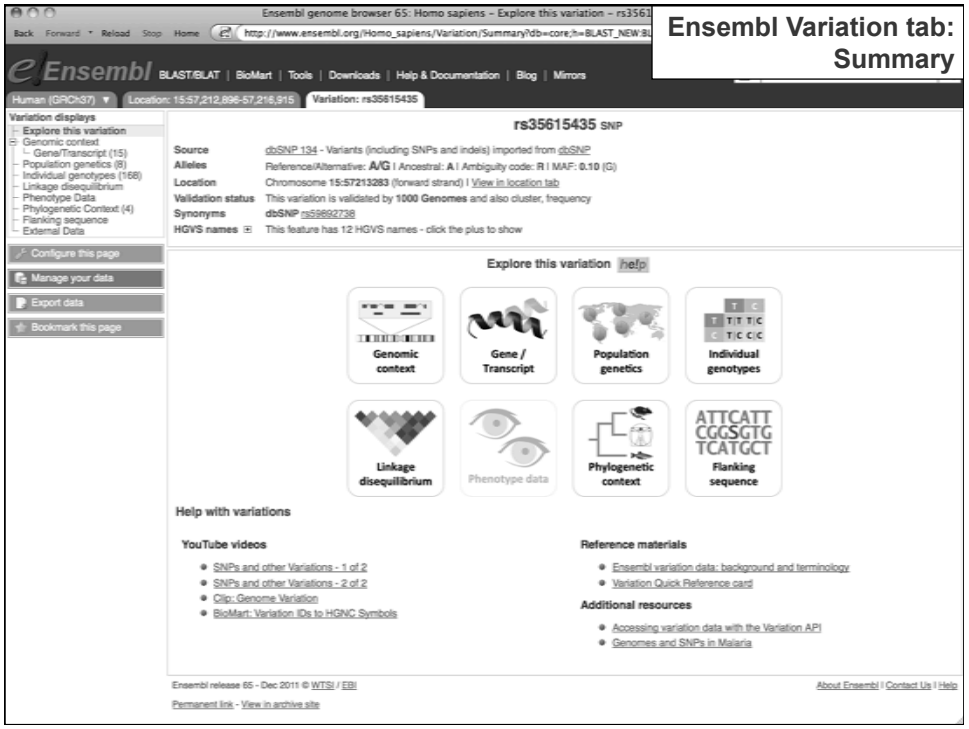
Ensembl Location tab: Region in detail with additional features



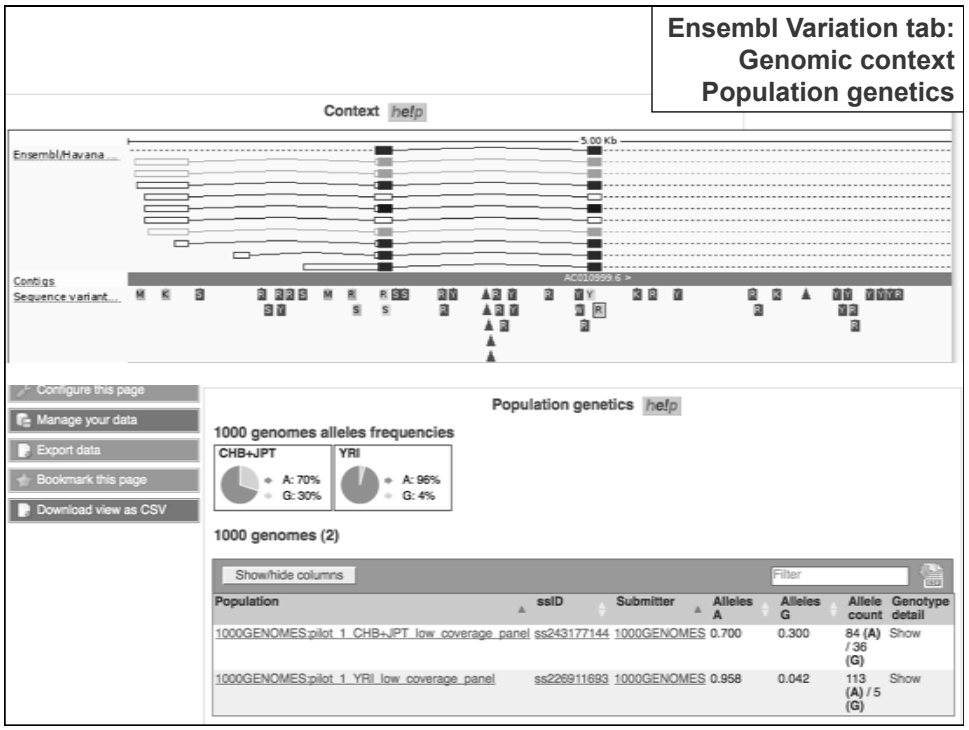
Ensembl Location tab: Region in detail after navigation



Ensembl Variation tab: Summary



Ensembl Variation tab: Genomic context Population genetics



Ensembl

Gene tab

http://www.ensembl.org

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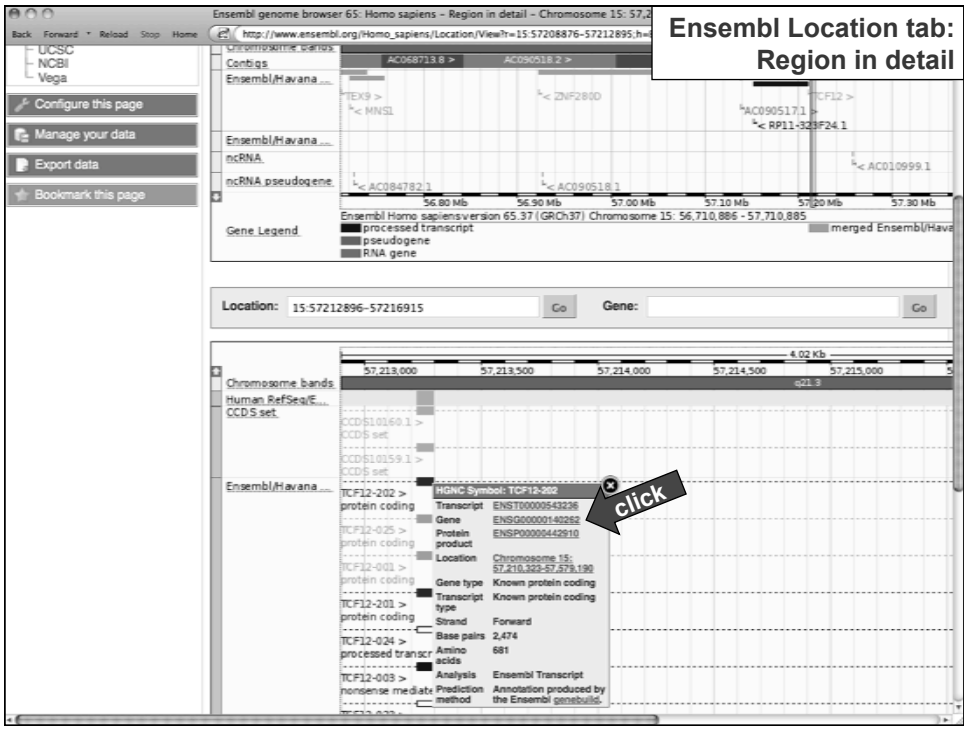
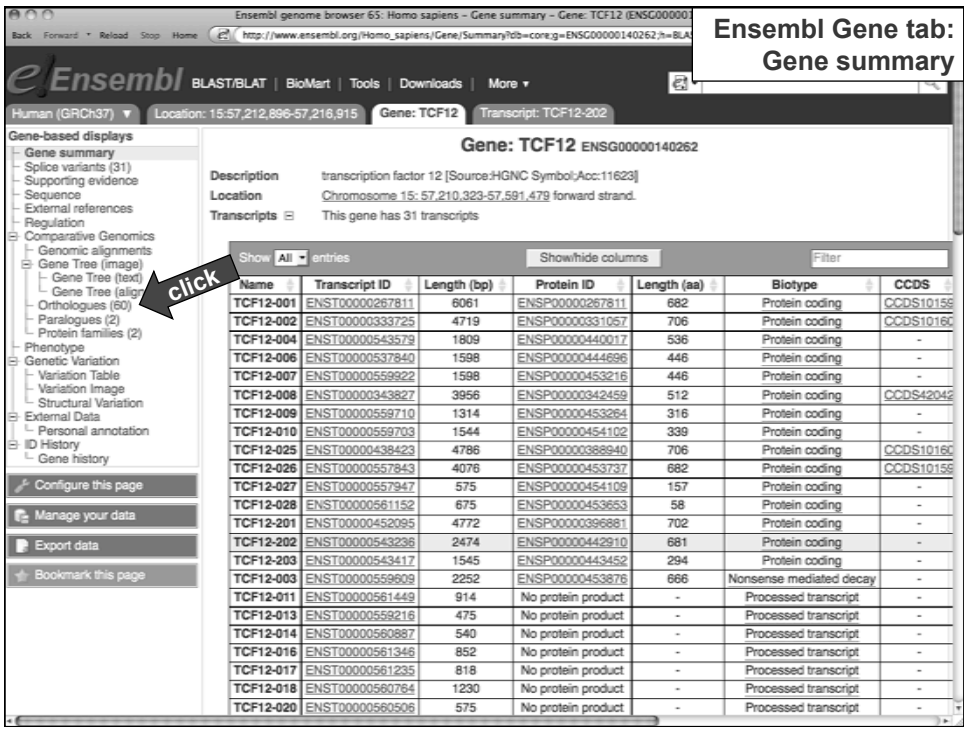
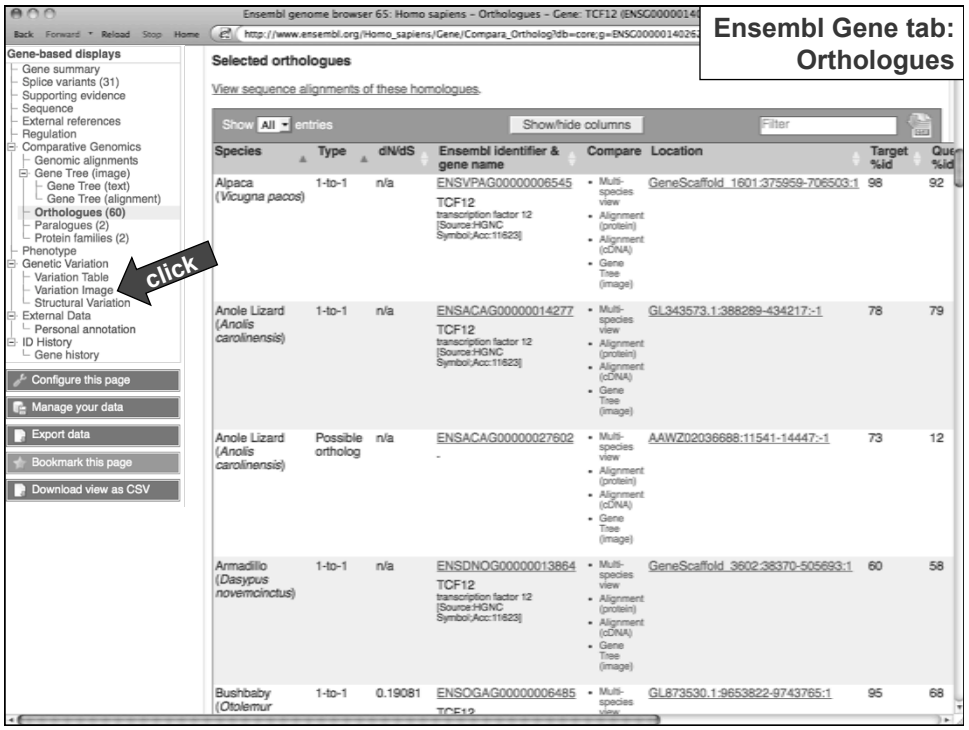
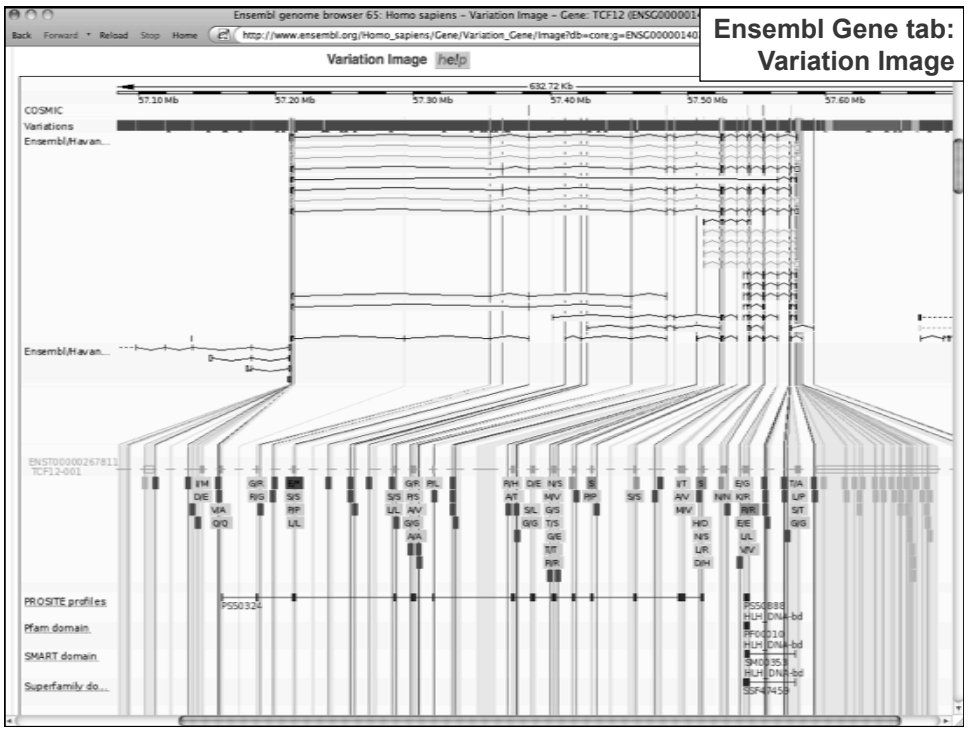




Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
TCF12-001	ENST00000267811	6061	ENSP00000267811	682	Protein coding	CCDS10159
TCF12-002	ENST00000333725	4719	ENSP00000331057	706	Protein coding	CCDS10160
TCF12-004	ENST00000543579	1809	ENSP00000440017	536	Protein coding	-
TCF12-006	ENST00000537840	1598	ENSP00000444696	446	Protein coding	-
TCF12-007	ENST00000559922	1598	ENSP00000453216	446	Protein coding	-
TCF12-008	ENST00000343827	3956	ENSP00000342459	512	Protein coding	CCDS42042
TCF12-009	ENST00000559710	1314	ENSP00000453264	316	Protein coding	-
TCF12-010	ENST00000559703	1544	ENSP00000454102	339	Protein coding	-
TCF12-025	ENST00000438423	4786	ENSP00000388940	706	Protein coding	CCDS10160
TCF12-026	ENST00000557843	4076	ENSP00000453737	682	Protein coding	CCDS10159
TCF12-027	ENST00000557947	575	ENSP00000454109	157	Protein coding	-
TCF12-028	ENST00000561152	675	ENSP00000453653	58	Protein coding	-
TCF12-201	ENST00000452095	4772	ENSP00000396881	702	Protein coding	-
TCF12-202	ENST00000543236	2474	ENSP00000442910	681	Protein coding	-
TCF12-203	ENST00000543417	1545	ENSP00000443452	294	Protein coding	-
TCF12-003	ENST00000559609	2252	ENSP00000453876	666	Nonsense mediated decay	-
TCF12-011	ENST00000561449	914	No protein product	-	Processed transcript	-
TCF12-013	ENST00000559216	475	No protein product	-	Processed transcript	-
TCF12-014	ENST00000560887	540	No protein product	-	Processed transcript	-
TCF12-016	ENST00000561346	852	No protein product	-	Processed transcript	-
TCF12-017	ENST00000561235	818	No protein product	-	Processed transcript	-
TCF12-018	ENST00000560764	1230	No protein product	-	Processed transcript	-
TCF12-020	ENST00000560506	575	No protein product	-	Processed transcript	-



Species	Type	dN/dS	Ensembl identifier & gene name	Compare	Location	Target %id	Query %id
Alpaca (*Vicugna pacos*)	1-to-1	n/a	ENSVPAG00000006545 TCF12 transcription factor 12 [Source:HGNC Symbol;Acc:11623]	Multi-species view, Alignment (protein), Alignment (cDNA), Gene Tree (image)	GeneScaffold_1601:375959-706503:1	98	92
Anole Lizard (*Anolis carolinensis*)	1-to-1	n/a	ENSACAG00000014277 TCF12 transcription factor 12 [Source:HGNC Symbol;Acc:11623]	Multi-species view, Alignment (protein), Alignment (cDNA), Gene Tree (image)	GL343573.1:388289-434217:-1	78	79
Anole Lizard (*Anolis carolinensis*)	Possible ortholog	n/a	ENSACAG00000027602 -	Multi-species view, Alignment (protein), Alignment (cDNA), Gene Tree (image)	AAWZ02036688:11541-14447:-1	73	12
Armadillo (*Dasypus novemcinctus*)	1-to-1	n/a	ENSDNOG00000013864 TCF12 transcription factor 12 [Source:HGNC Symbol;Acc:11623]	Multi-species view, Alignment (protein), Alignment (cDNA), Gene Tree (image)	GeneScaffold_3602:38370-505693:1	60	58
Bushbaby (*Otolemur*	1-to-1	0.19081	ENSOGAG00000006485 TCF12	Multi-species view	GL873530.1:9653822-9743765:1	95	68

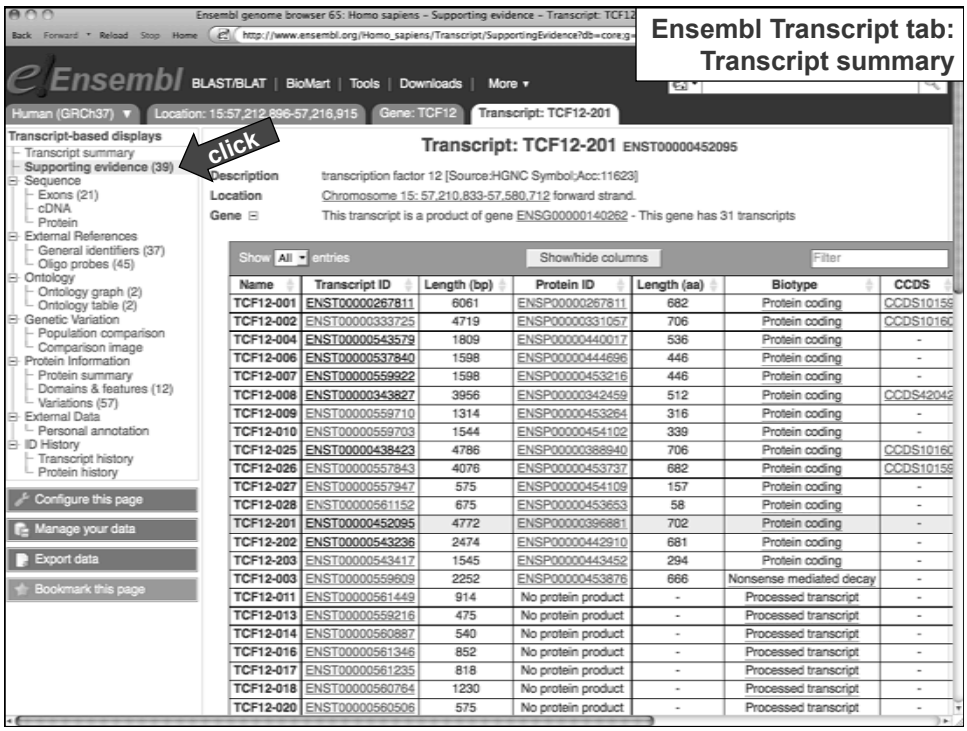


Ensembl Gene tab: Variation Image

Ensembl

Transcript tab

http://www.ensembl.org



Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
TCF12-001	ENST00000267811	6061	ENSP00000267811	682	Protein coding	CCDS10155
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TCF12-006	ENST00000537840	1598	ENSP00000444696	446	Protein coding	-
TCF12-007	ENST00000559922	1598	ENSP00000453216	446	Protein coding	-
TCF12-008	ENST00000343827	3956	ENSP00000342459	512	Protein coding	CCDS42042
TCF12-009	ENST00000559710	1314	ENSP00000453264	316	Protein coding	-
TCF12-010	ENST00000559703	1544	ENSP00000454102	339	Protein coding	-
TCF12-025	ENST00000438423	4786	ENSP00000388940	706	Protein coding	CCDS10160
TCF12-026	ENST00000557843	4076	ENSP00000453737	682	Protein coding	CCDS10155
TCF12-027	ENST00000557947	575	ENSP00000454109	157	Protein coding	-
TCF12-028	ENST00000561152	675	ENSP00000453653	58	Protein coding	-
TCF12-201	ENST00000452095	4772	ENSP00000396881	702	Protein coding	-
TCF12-202	ENST00000543236	2474	ENSP00000442910	681	Protein coding	-
TCF12-203	ENST00000543417	1545	ENSP00000443452	294	Protein coding	-
TCF12-003	ENST00000559609	2252	ENSP00000453876	666	Nonsense mediated decay	-
TCF12-011	ENST00000561449	914	No protein product	-	Processed transcript	-
TCF12-013	ENST00000559216	475	No protein product	-	Processed transcript	-
TCF12-014	ENST00000560887	540	No protein product	-	Processed transcript	-
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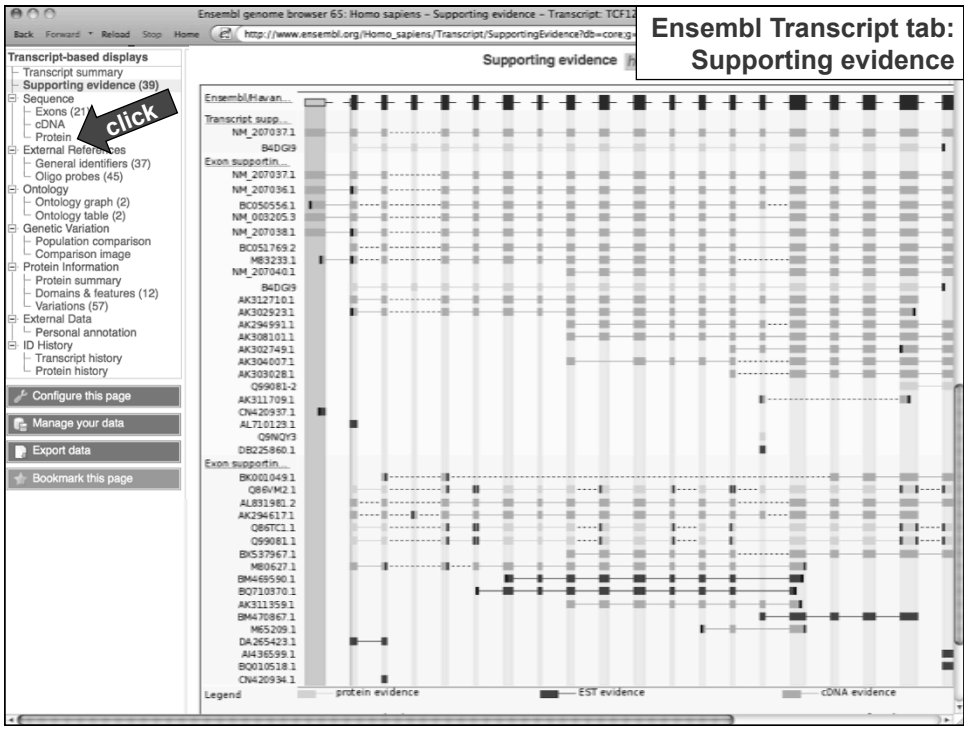

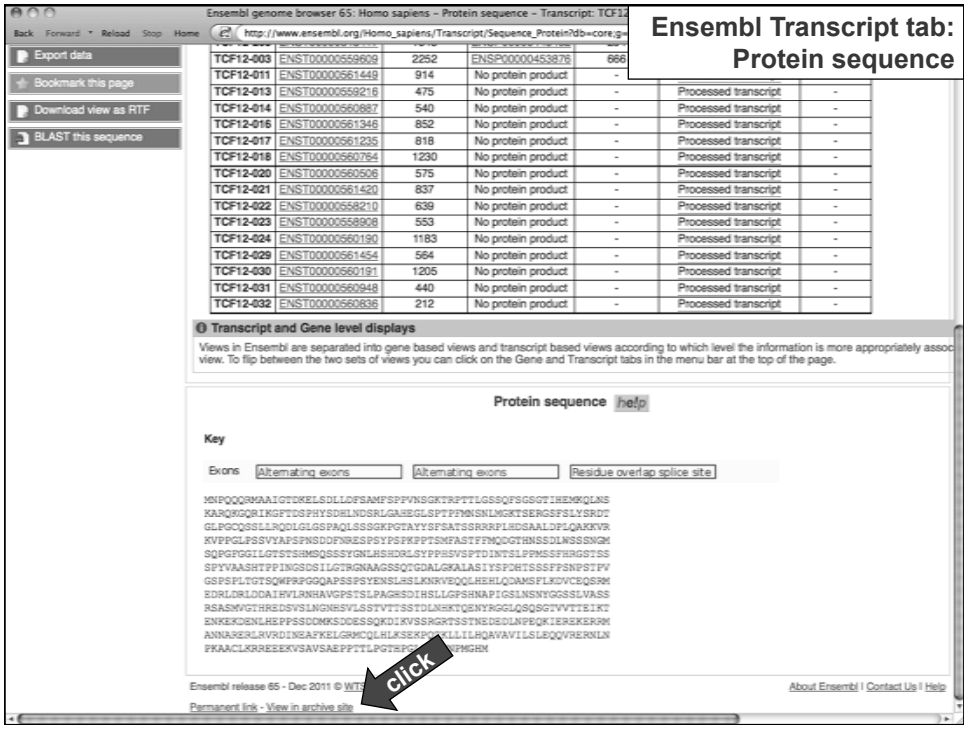
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Protein sequence
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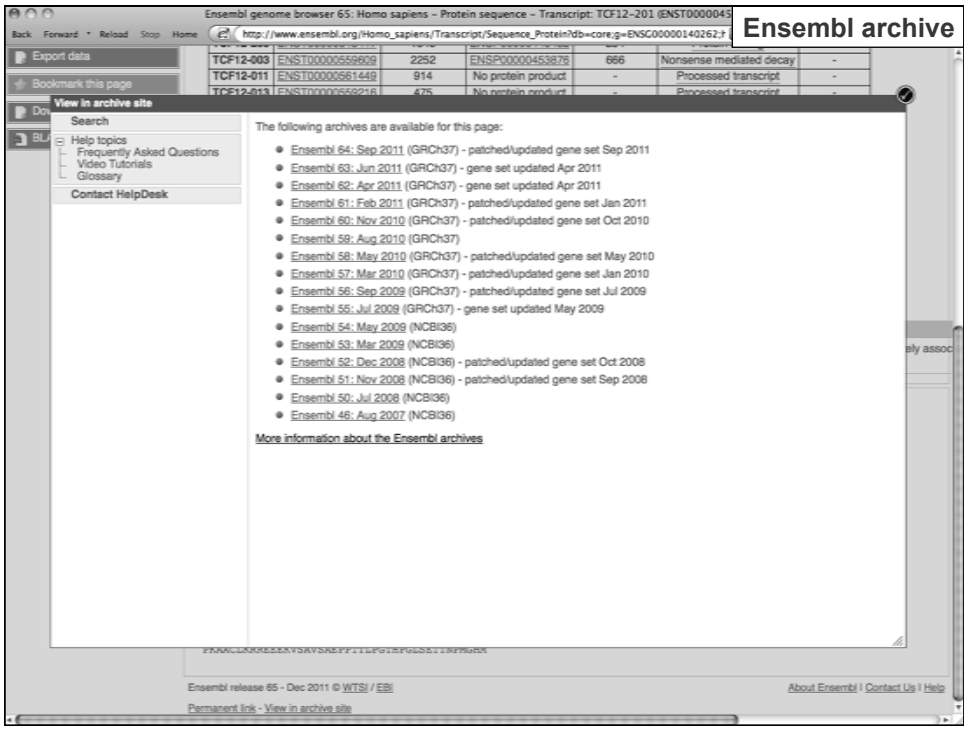
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Ensembl 64: Sep 2011 (GRCh37) - patched/updated gene set Sep 2011
Ensembl 63: Jun 2011 (GRCh37) - gene set updated Apr 2011
Ensembl 62: Apr 2011 (GRCh37) - gene set updated Apr 2011
Ensembl 61: Feb 2011 (GRCh37) - patched/updated gene set Jan 2011
Ensembl 60: Nov 2010 (GRCh37) - patched/updated gene set Oct 2010
Ensembl 59: Aug 2010 (GRCh37)
Ensembl 58: May 2010 (GRCh37) - patched/updated gene set May 2010
Ensembl 57: Mar 2010 (GRCh37) - patched/updated gene set Jan 2010
Ensembl 56: Sep 2009 (GRCh37) - patched/updated gene set Jul 2009
Ensembl 55: Jul 2009 (GRCh37) - gene set updated May 2009
Ensembl 54: May 2009 (NCBI36)
Ensembl 53: Mar 2009 (NCBI36)
Ensembl 52: Dec 2008 (NCBI36) - patched/updated gene set Oct 2008
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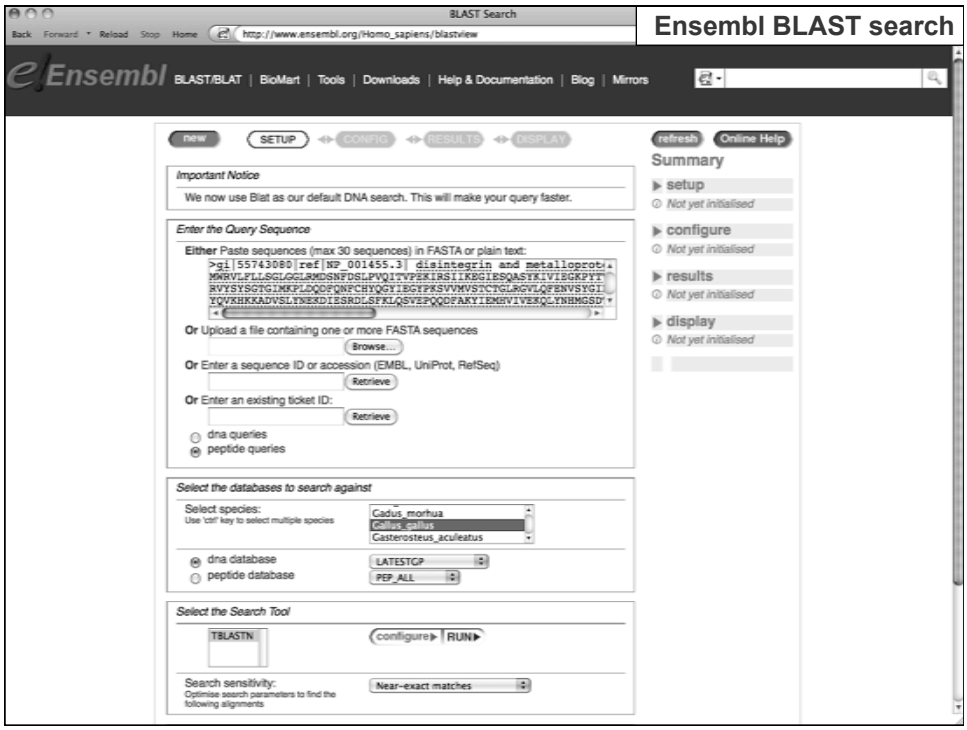

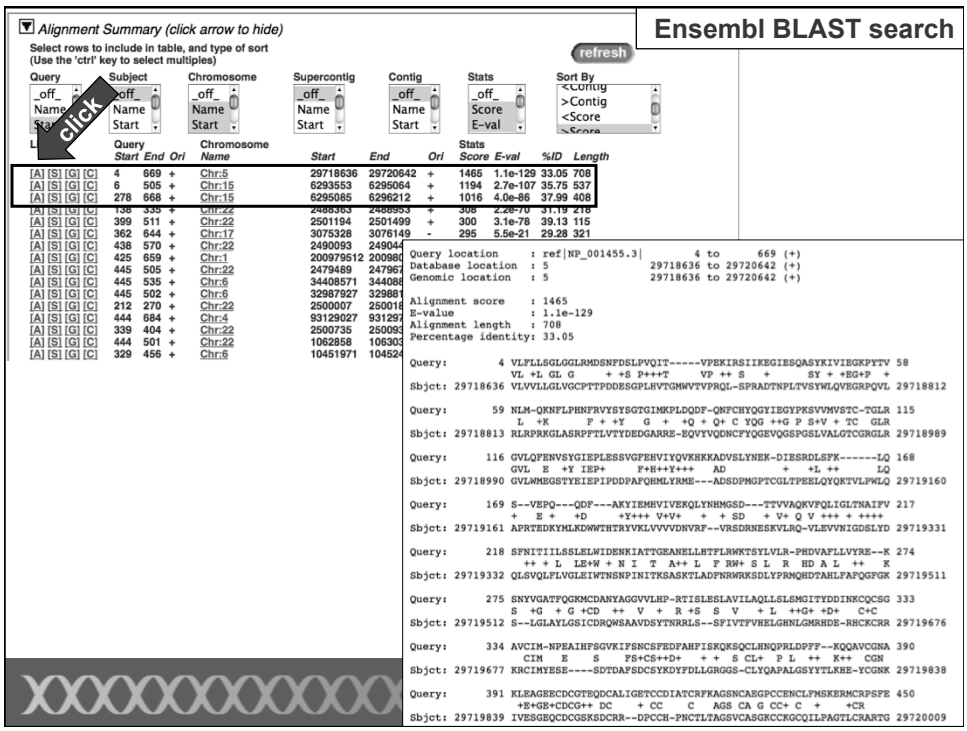

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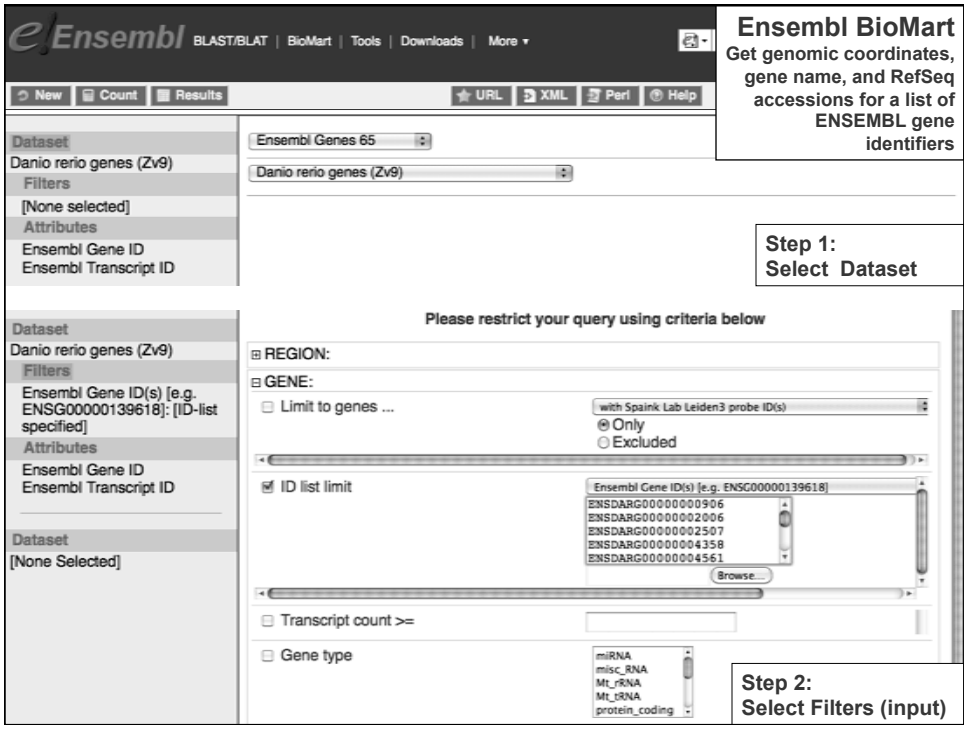
Ensembl BLAST/BLAT | BioMart | Tools | Downloads | More
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Danio rerio genes (Zv9)
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Ensembl Genes 65
Danio rerio genes (Zv9)
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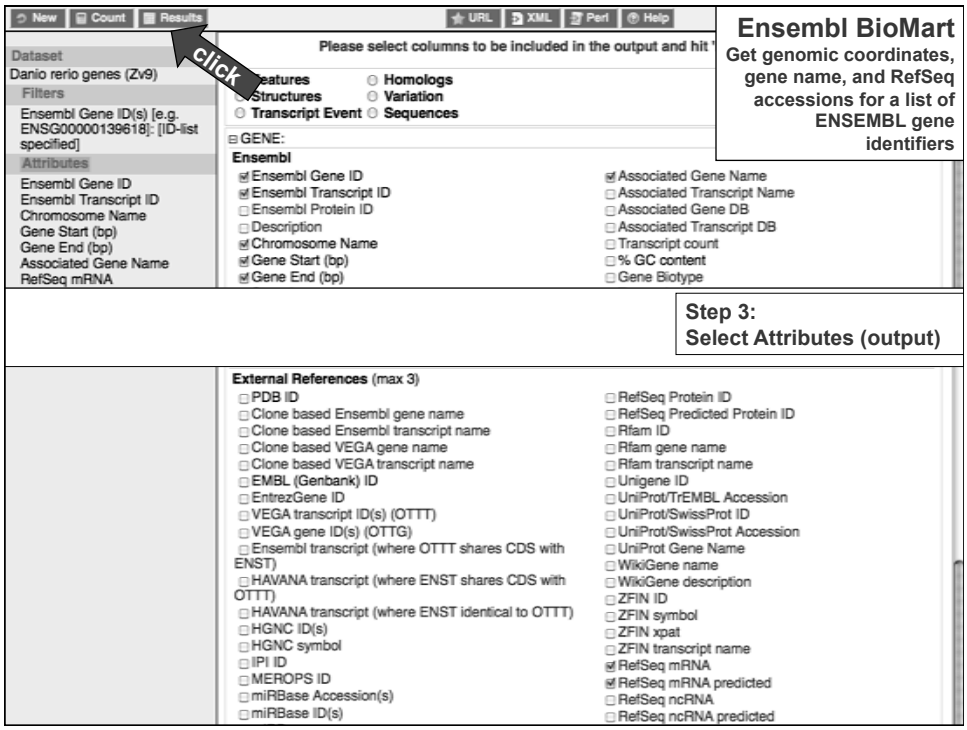
New Count Results URL XML Perl Help
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VEGA gene ID(s) (OTTG)
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HAVANA transcript (where ENST shares CDS with OTTT)
HAVANA transcript (where ENST identical to OTTT)
HGNC ID(s)
HGNC symbol
IPI ID
MEROPS ID
miRBase Accession(s)
miRBase ID(s)
RefSeq Protein ID
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Rfam transcript name
Unigene ID
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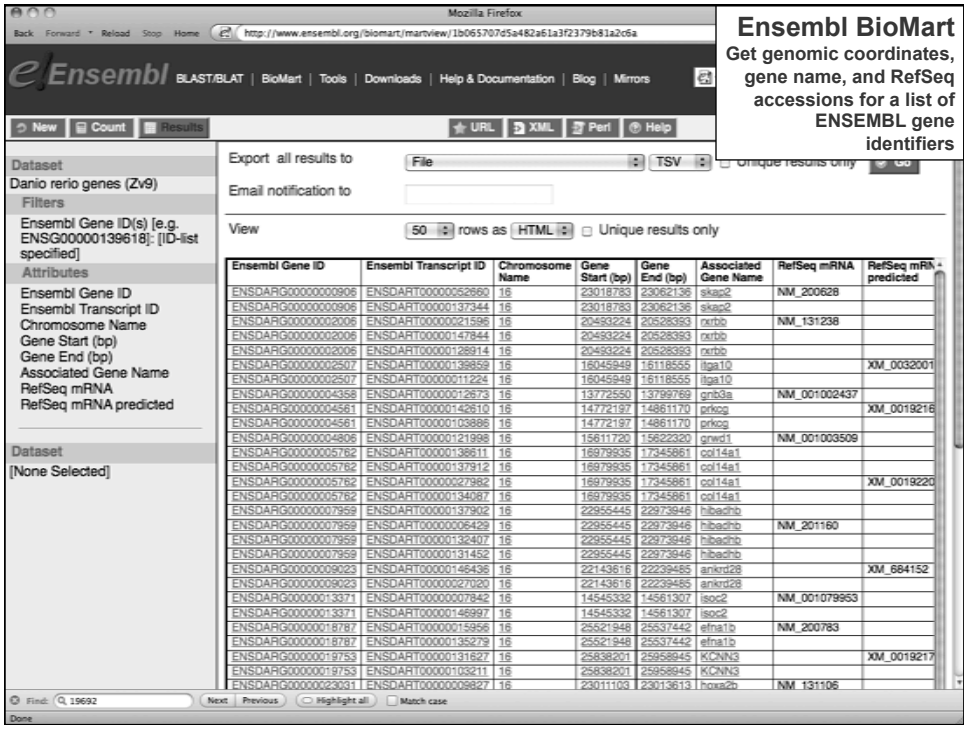
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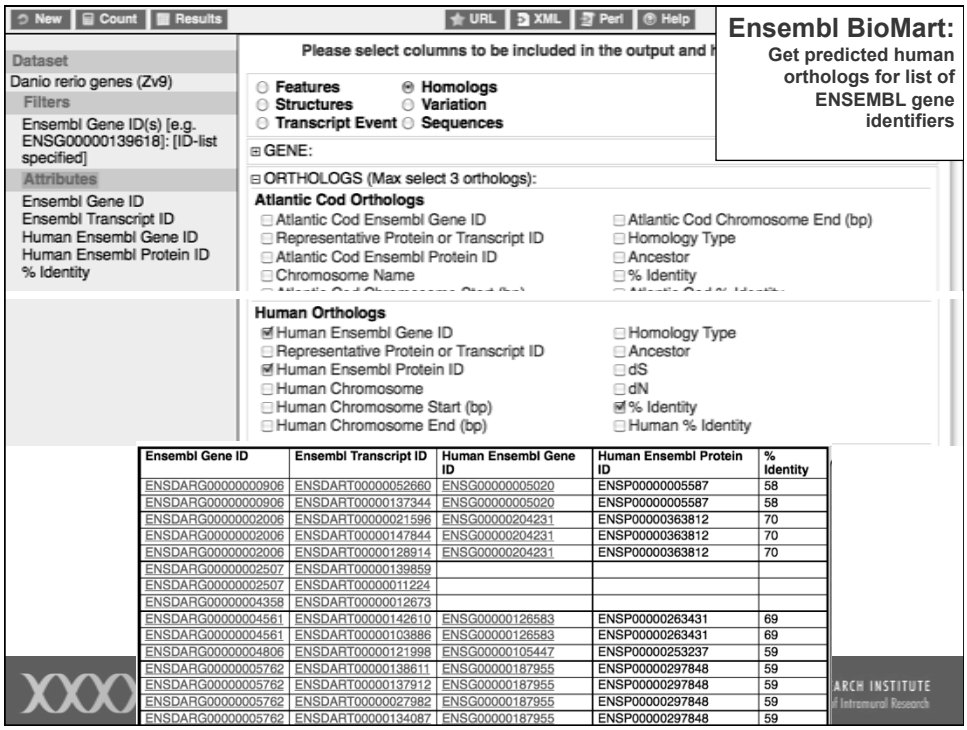
Ensembl BioMart
Get genomic coordinates, gene name, and RefSeq accessions for a list of ENSEMBL gene identifiers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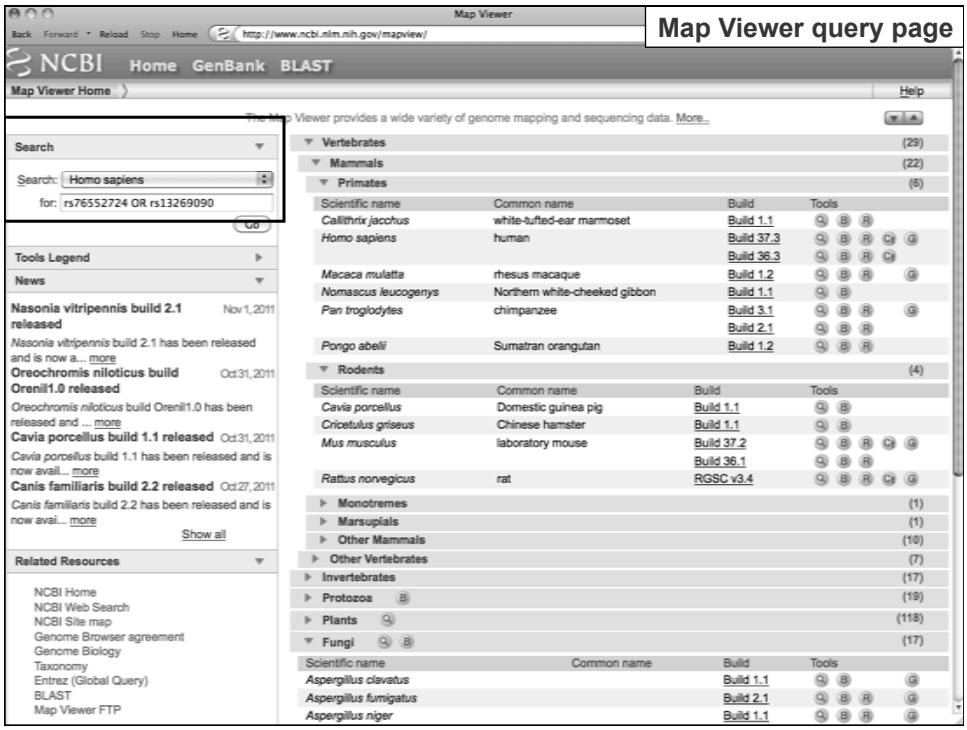

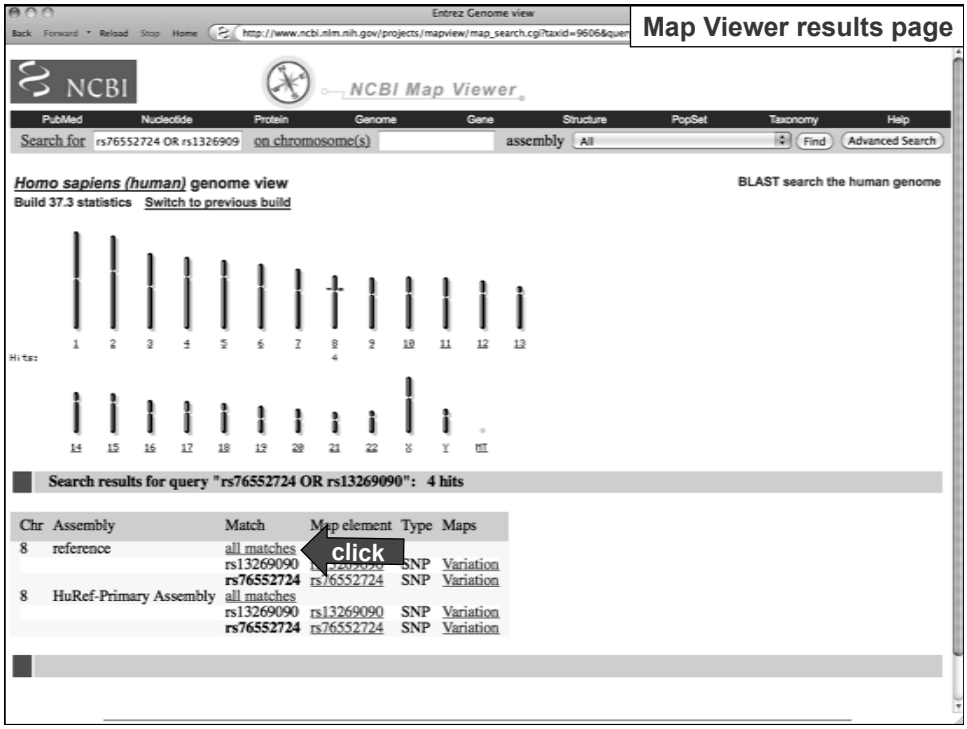
Map Viewer results page
Entrez Genome view
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PubMed Nucleotide Protein Genome Gene Structure PopSet Taxonomy Help
Search for rs76552724 OR rs1326909 on chromosome(s) assembly All Find Advanced Search
Homo sapiens (human) genome view
Build 37.3 statistics Switch to previous build
BLAST search the human genome
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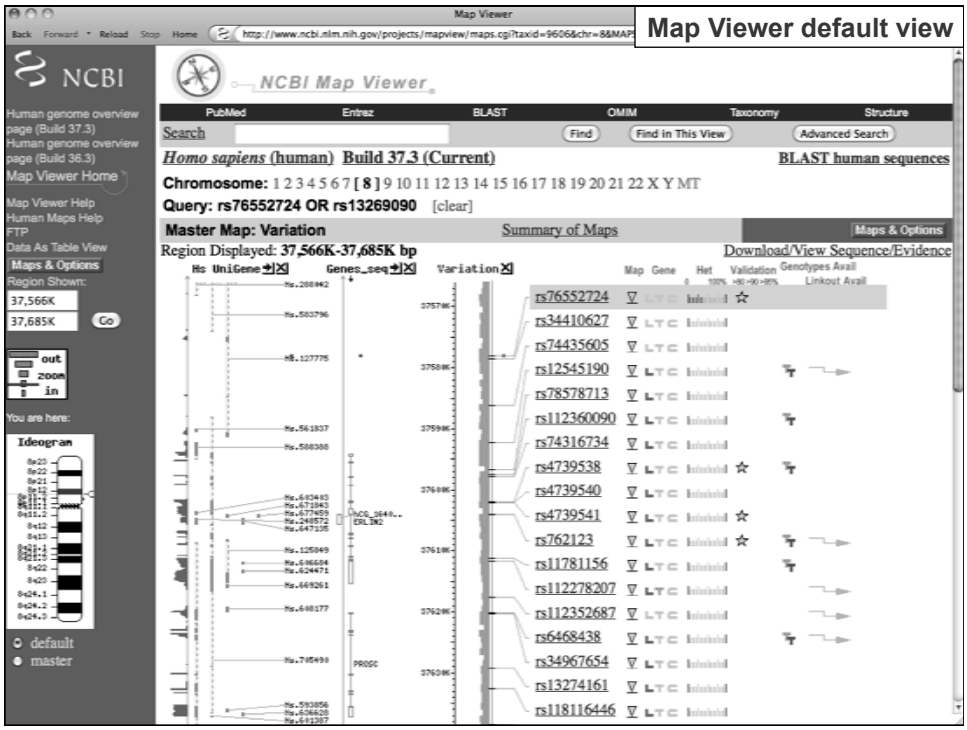
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BLAST human sequences
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Master Map: Variation
Summary of Maps
Maps & Options
Region Displayed: 37,566K-37,685K bp
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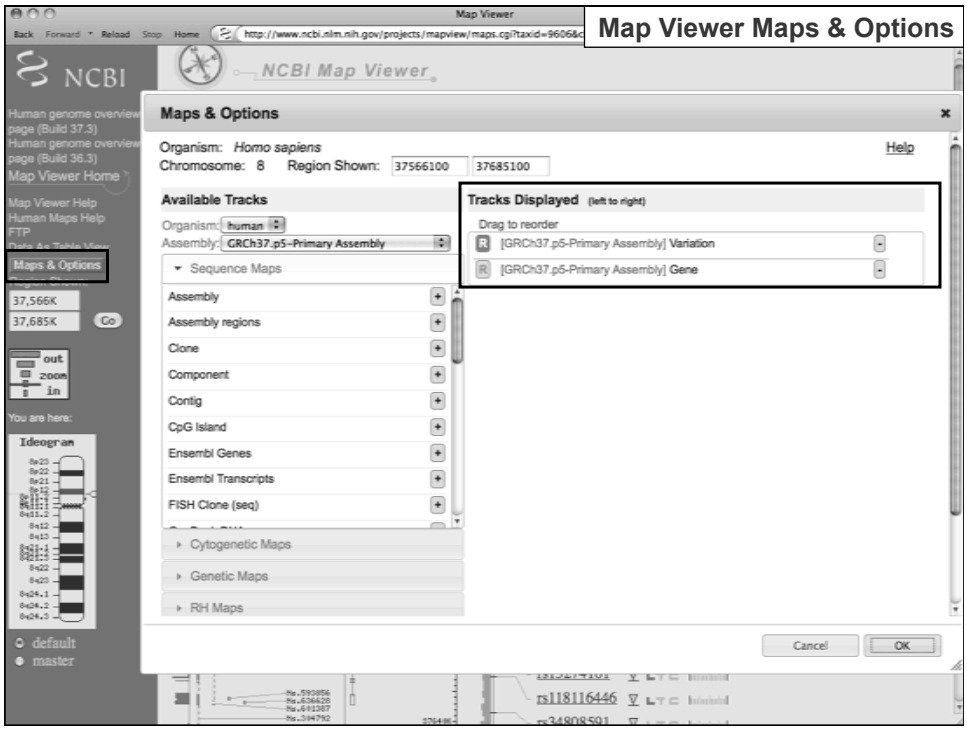

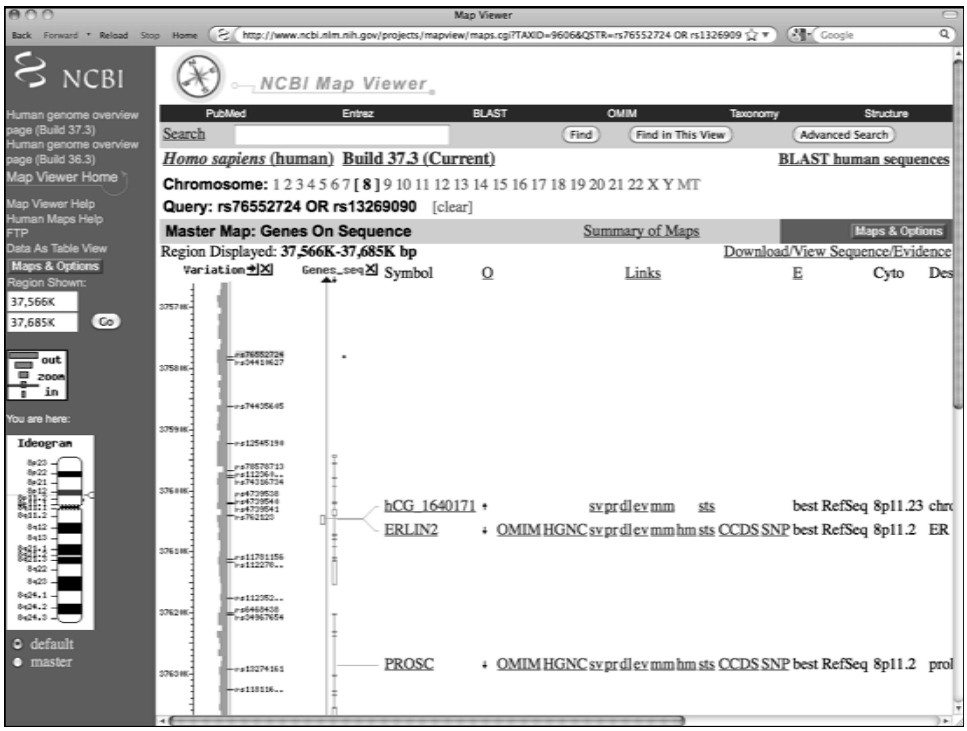

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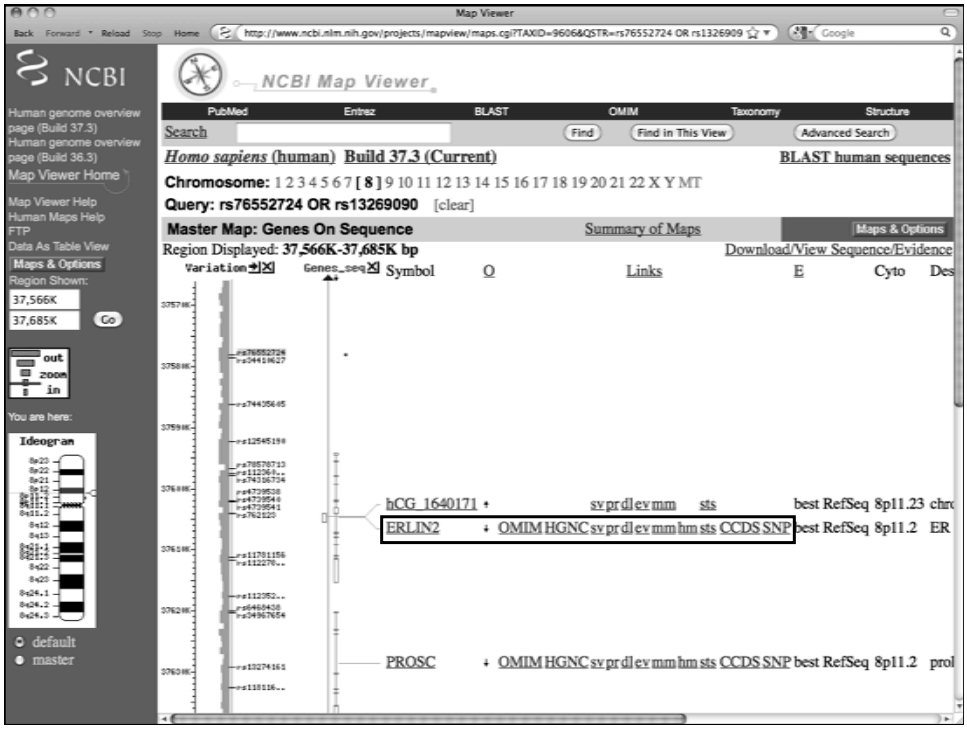
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BLAST human sequences
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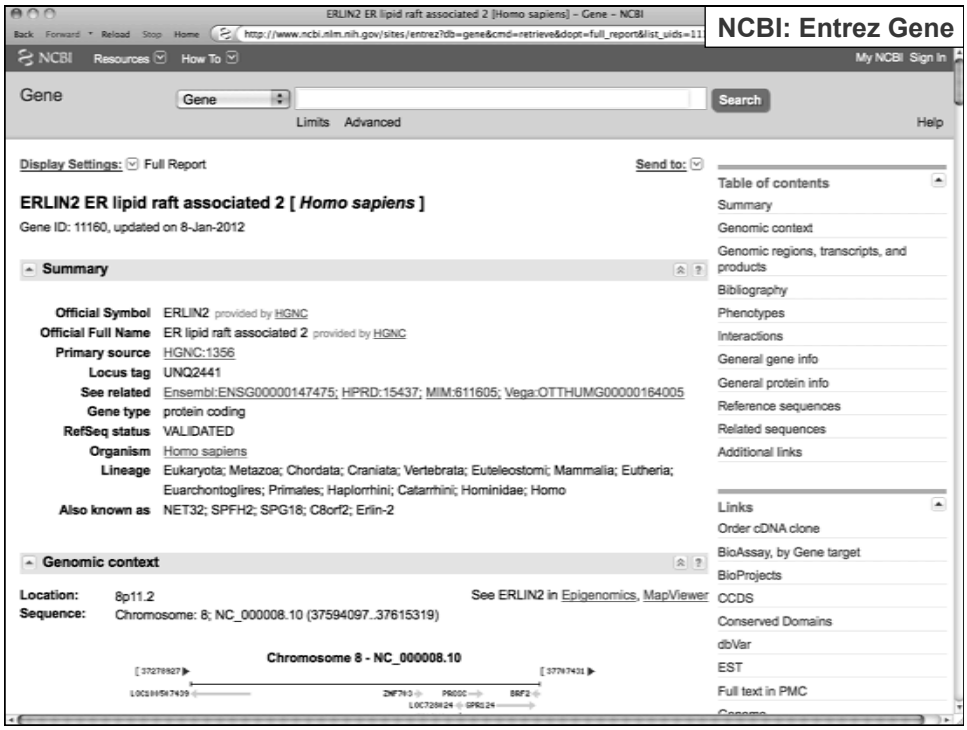
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Gene ID: 11160, updated on 8-Jan-2012
Summary
Official Symbol ERLIN2 provided by HGNC
Official Full Name ER lipid raft associated 2 provided by HGNC
Primary source HGNC:1356
Locus tag UNQ2441
See related Ensembl:ENSG00000147475; HPRD:15437; MIM:611605; Vega:OTTHUMG00000164005
Gene type protein coding
RefSeq status VALIDATED
Organism Homo sapiens
Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as NET32; SPFH2; SPG18; C8orf2; Erlin-2
Genomic context
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Sequence: Chromosome: 8; NC_000008.10 (37594097..37615319)
See ERLIN2 in Epigenomics, MapViewer
Chromosome 8 - NC_000008.10
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Additional links
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CCDS
Conserved Domains
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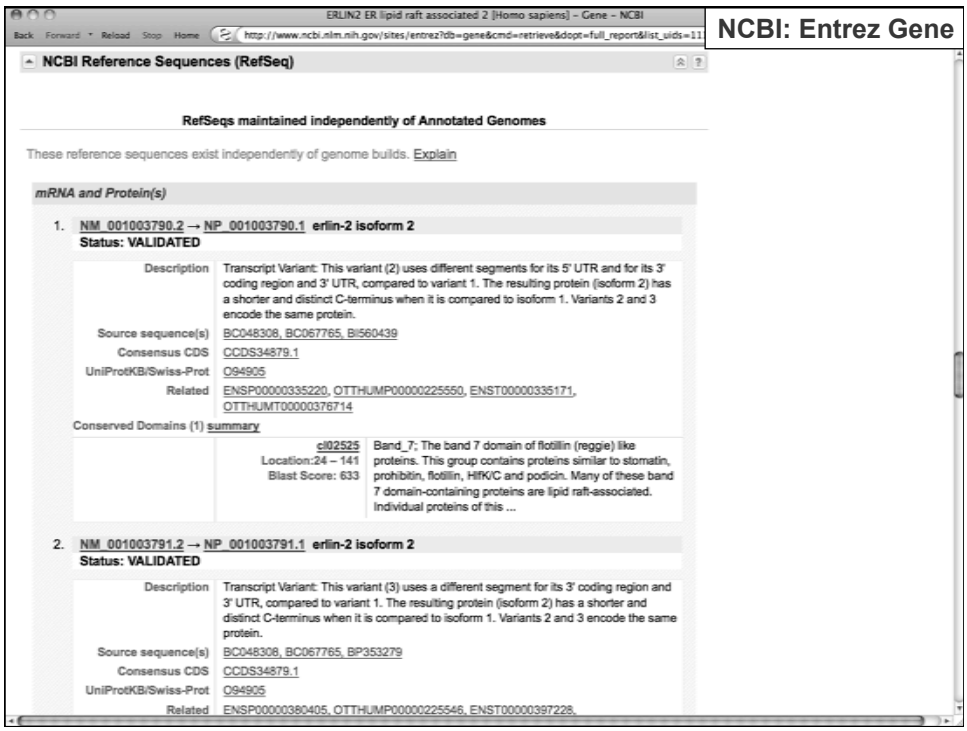
Full text in PMC

NCBI: Entrez Gene



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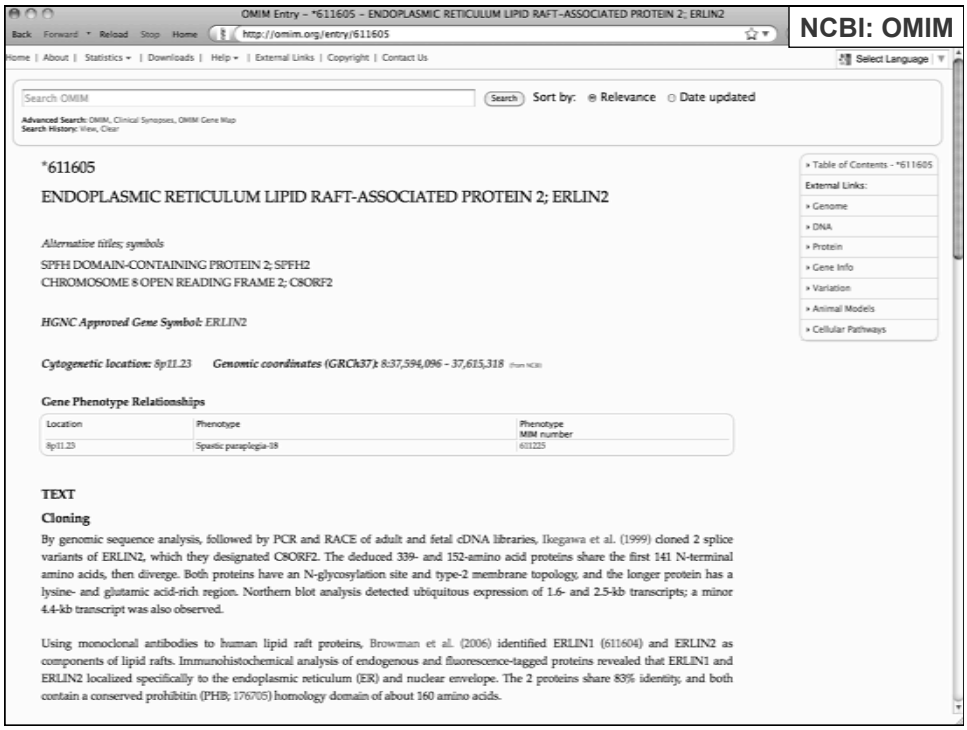

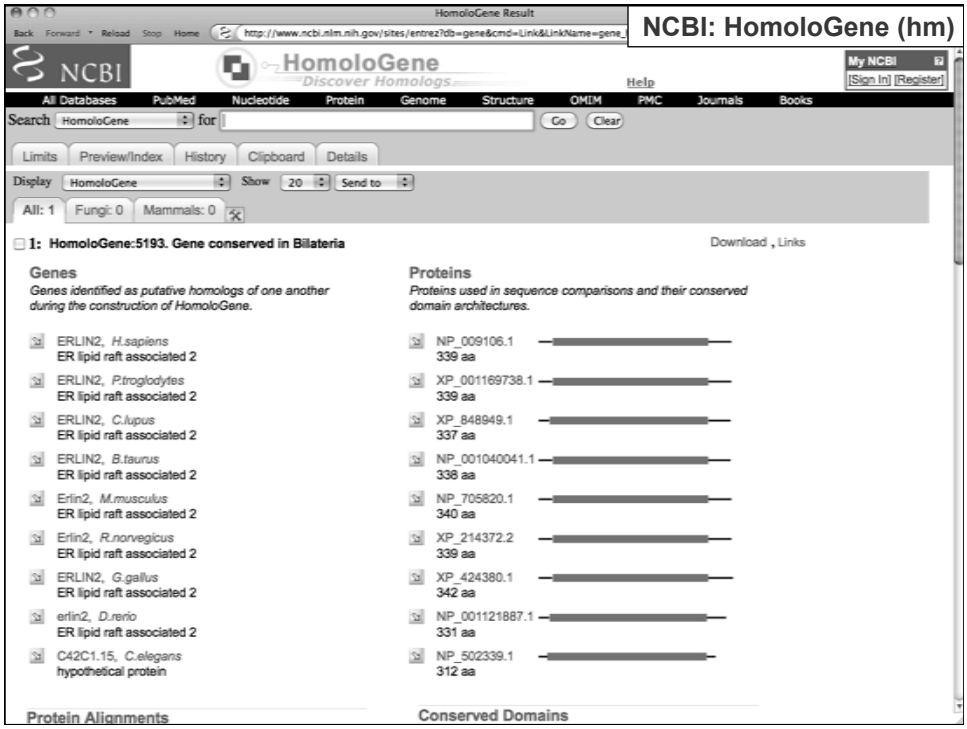
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All: 1 Fungi: 0 Mammals: 0
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Genes
Genes identified as putative homologs of one another during the construction of HomoloGene.
Proteins
Proteins used in sequence comparisons and their conserved domain architectures.
ERLIN2, H.sapiens ER lipid raft associated 2
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XP_001169738.1 339 aa
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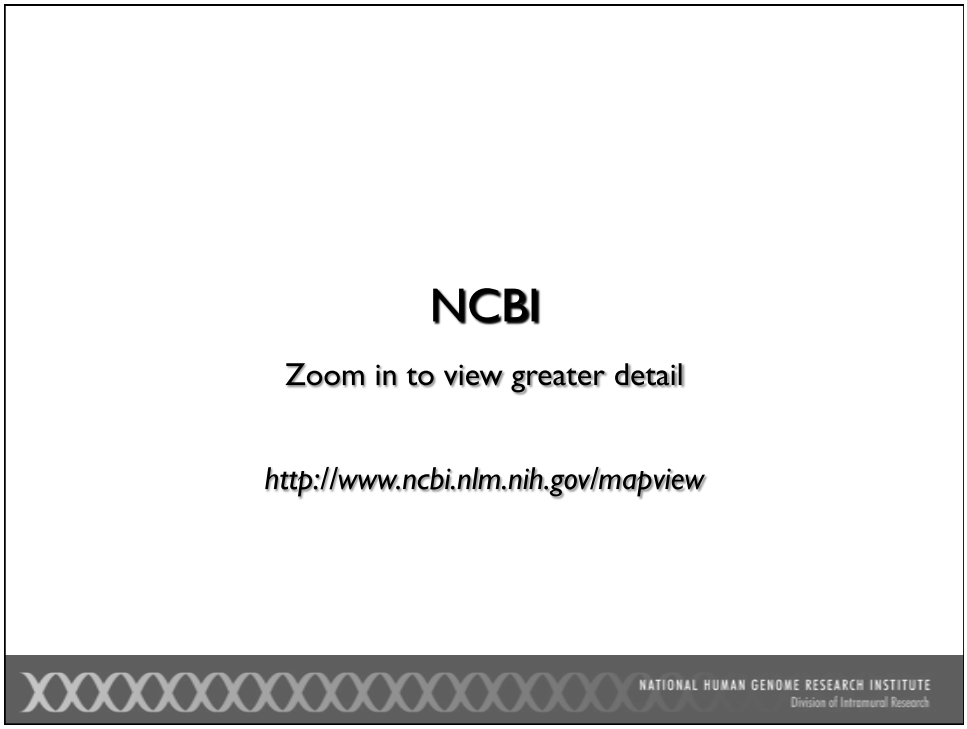
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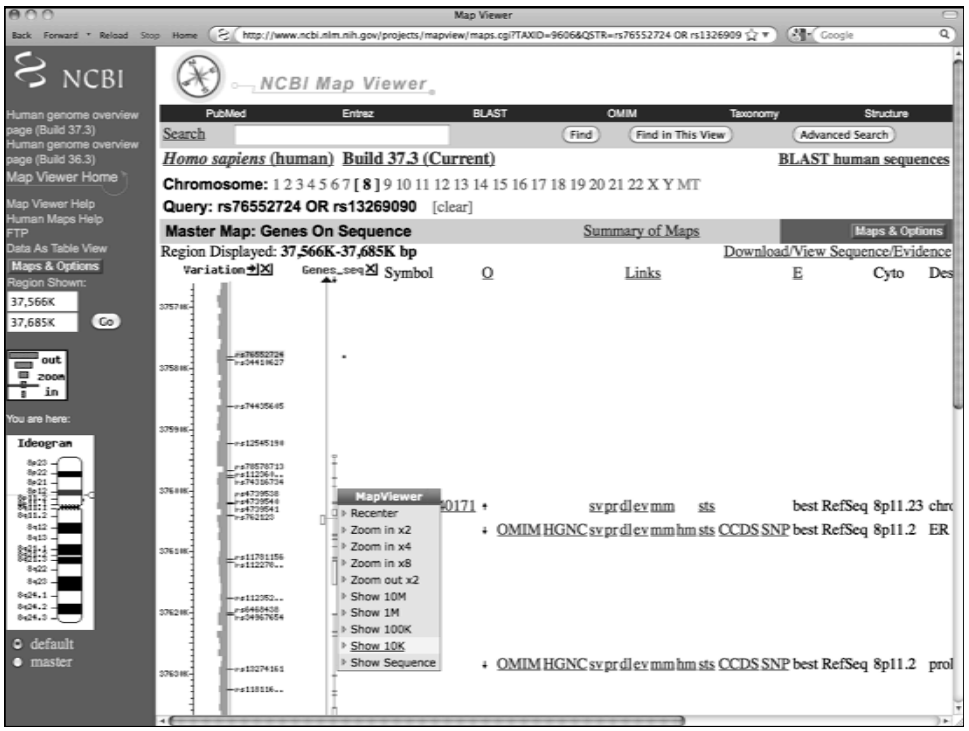
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Master Map: Genes On Sequence
Summary of Maps
Maps & Options
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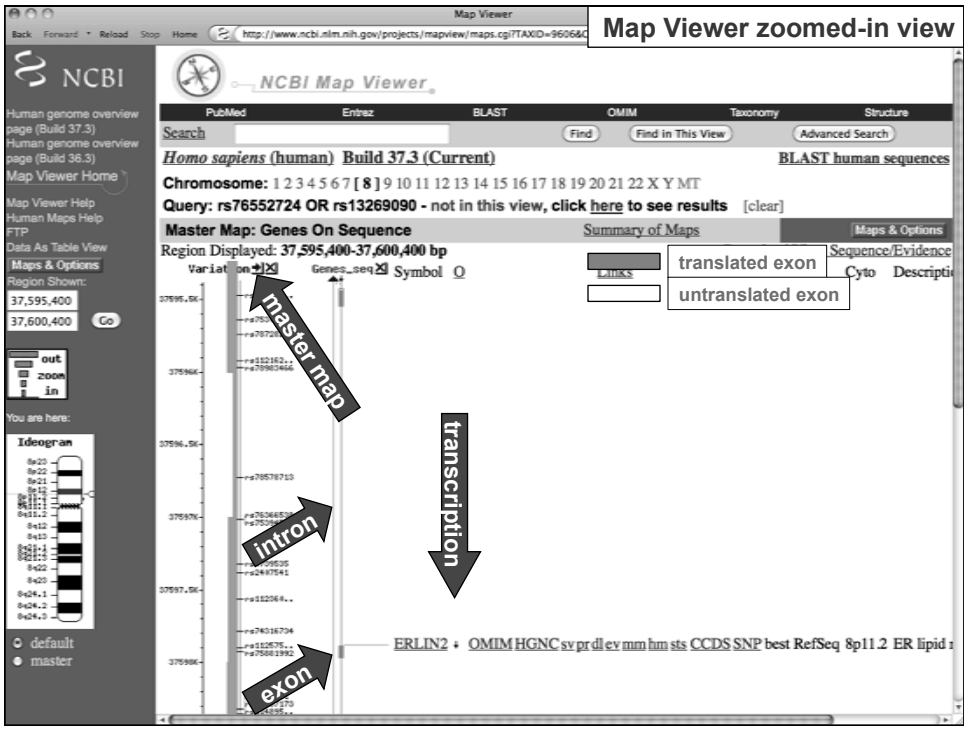
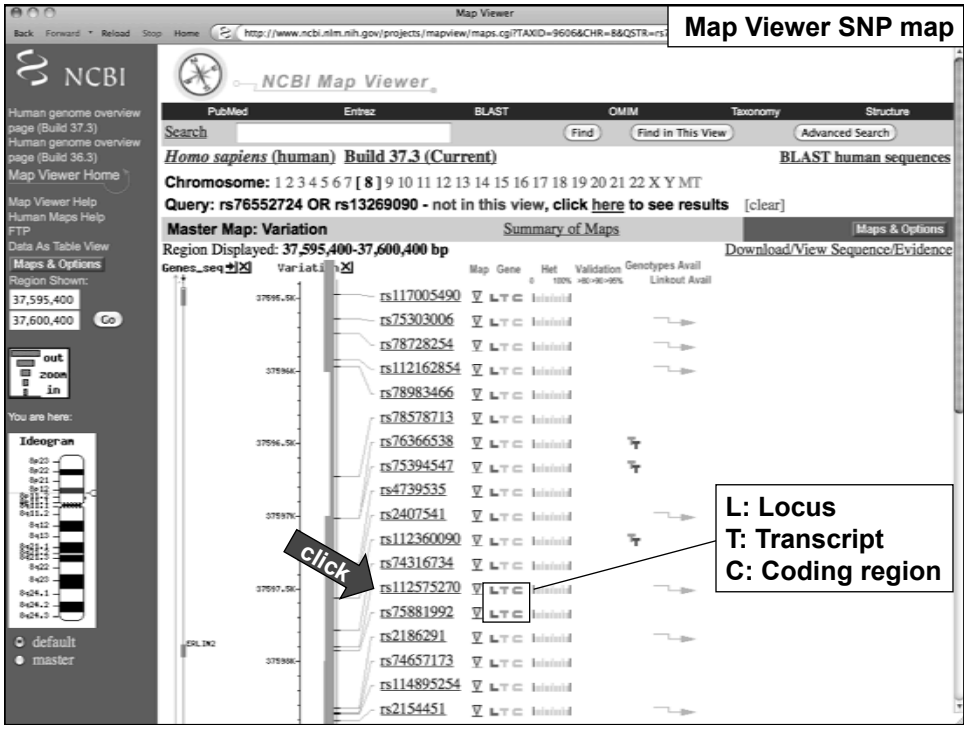
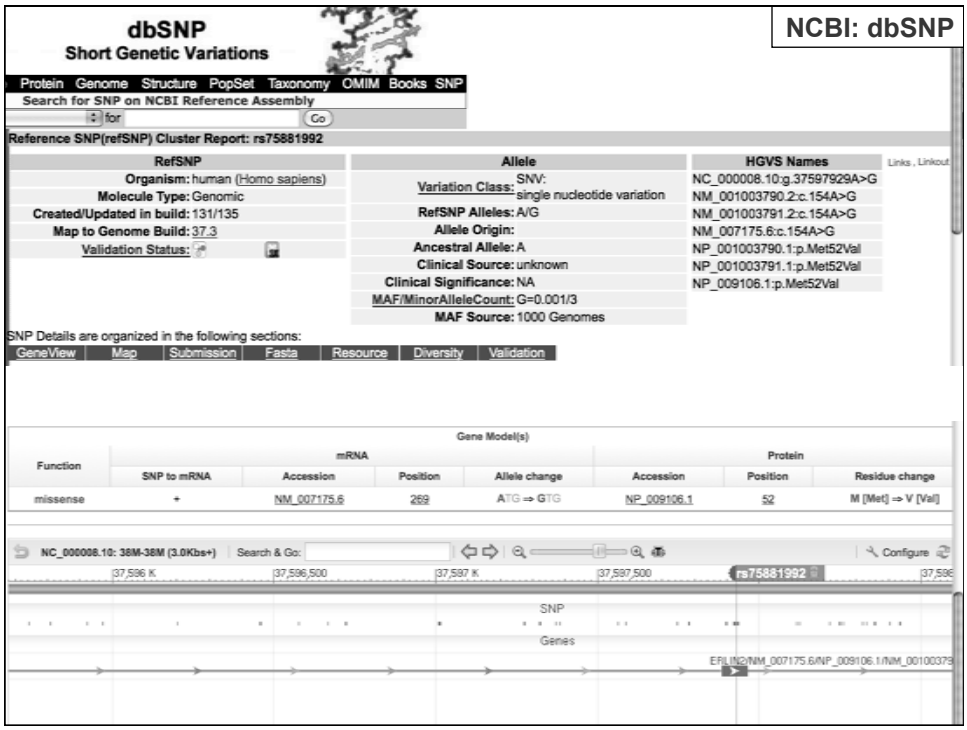
37,685K
Go
out
zoom
in
You are here:
Ideogram
default
master


Map Viewer zoomed-in view
NCBI
NCBI Map Viewer
Homo sapiens (human) Build 37.3 (Current)
Chromosome: 1 2 3 4 5 6 7 [8] 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y MT
Query: rs76552724 OR rs13269090 - not in this view, click here to see results [clear]
Master Map: Genes On Sequence
Region Displayed: 37,595,400-37,600,400 bp
translated exon
untranslated exon
master map
transcription
intron
exon
ERLIN2
Region Shown:
37,595,400
37,600,400
Go
Ideogram
default
master

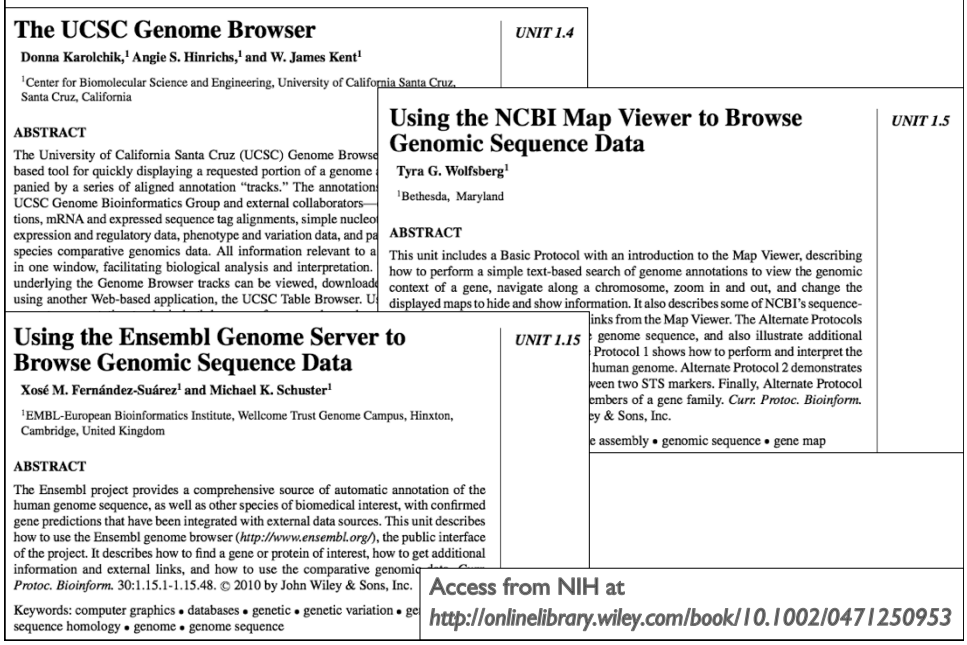

Map Viewer SNP map
NCBI
NCBI Map Viewer
Human genome overview page (Build 37.3)
Human genome overview page (Build 36.3)
Map Viewer Home
Map Viewer Help
Human Maps Help
FTP
Data As Table View
Maps & Options
Region Shown:
37,595,400
37,600,400
Go
out
zoom
in
You are here:
Ideogram
default
master
PubMed
Entrez
BLAST
OMIM
Taxonomy
Structure
Search
Find
Find in This View
Advanced Search
Homo sapiens (human) Build 37.3 (Current)
BLAST human sequences
Chromosome: 1 2 3 4 5 6 7 [8] 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y MT
Query: rs76552724 OR rs13269090 - not in this view, click here to see results
[clear]
Master Map: Variation
Summary of Maps
Maps & Options
Region Displayed: 37,595,400-37,600,400 bp
Download/View Sequence/Evidence
Map Gene Het Validation Genotypes Avail Linkout Avail
rs117005490
rs75303006
rs78728254
rs112162854
rs78983466
rs78578713
rs76366538
rs75394547
rs4739535
rs2407541
rs112360090
rs74316734
rs112575270
rs75881992
rs2186291
rs74657173
rs114895254
rs2154451
click
L: Locus
T: Transcript
C: Coding region


NCBI: dbSNP
dbSNP
Short Genetic Variations
Protein Genome Structure PopSet Taxonomy OMIM Books SNP
Search for SNP on NCBI Reference Assembly
for
Go
Reference SNP(refSNP) Cluster Report: rs75881992
RefSNP
Organism: human (Homo sapiens)
Molecule Type: Genomic
Created/Updated in build: 131/135
Map to Genome Build: 37.3
Validation Status:
Allele
Variation Class: SNV: single nucleotide variation
RefSNP Alleles: A/G
Allele Origin:
Ancestral Allele: A
Clinical Source: unknown
Clinical Significance: NA
MAF/MinorAlleleCount: G=0.001/3
MAF Source: 1000 Genomes
HGVS Names
NC_000008.10:g.37597929A>G
NM_001003790.2:c.154A>G
NM_001003791.2:c.154A>G
NM_007175.6:c.154A>G
NP_001003790.1:p.Met52Val
NP_001003791.1:p.Met52Val
NP_009106.1:p.Met52Val
Links , Linkout
SNP Details are organized in the following sections:
GeneView Map Submission Fasta Resource Diversity Validation
Gene Model(s)
Function
mRNA
Protein
SNP to mRNA
Accession
Position
Allele change
Accession
Position
Residue change
missense
+
NM_007175.6
269
ATG ⇒ GTG
NP_009106.1
52
M [Met] ⇒ V [Val]
NC_000008.10: 36M-38M (3.0Kbs+)
Search & Go:
Configure
37,596 K
37,596,500
37,597 K
37,597,500
rs75881992
SNP
Genes
EFLN2/NM_007175.6/NP_009106.1/NM_00100379

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

Donna Karolchik,[1] Angie S. Hinrichs,[1] and W. James Kent[1]

[1]Center for Biomolecular Science and Engineering, University of California Santa Cruz, Santa Cruz, California

ABSTRACT

The University of California Santa Cruz (UCSC) Genome Browse based tool for quickly displaying a requested portion of a genome panied by a series of aligned annotation "tracks." The annotation UCSC Genome Bioinformatics Group and external collaborators— tions, mRNA and expressed sequence tag alignments, simple nucleo expression and regulatory data, phenotype and variation data, and pa species comparative genomics data. All information relevant to a in one window, facilitating biological analysis and interpretation. underlying the Genome Browser tracks can be viewed, downloade using another Web-based application, the UCSC Table Browser. U

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

Tyra G. Wolfsberg[1]

[1]Bethesda, Maryland

ABSTRACT

This unit includes a Basic Protocol with an introduction to the Map Viewer, describing how to perform a simple text-based search of genome annotations to view the genomic context of a gene, navigate along a chromosome, zoom in and out, and change the displayed maps to hide and show information. It also describes some of NCBI's sequence- inks from the Map Viewer. The Alternate Protocols genome sequence, and also illustrate additional Protocol 1 shows how to perform and interpret the human genome. Alternate Protocol 2 demonstrates ween two STS markers. Finally, Alternate Protocol embers of a gene family. *Curr. Protoc. Bioinform.* ey & Sons, Inc.

e assembly • genomic sequence • gene map

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

Xosé M. Fernández-Suárez[1] and Michael K. Schuster[1]

[1]EMBL-European Bioinformatics Institute, Wellcome Trust Genome Campus, Hinxton, Cambridge, United Kingdom

ABSTRACT

The Ensembl project provides a comprehensive source of automatic annotation of the human genome sequence, as well as other species of biomedical interest, with confirmed gene predictions that have been integrated with external data sources. This unit describes how to use the Ensembl genome browser (*http://www.ensembl.org/*), the public interface of the project. It describes how to find a gene or protein of interest, how to get additional information and external links, and how to use the comparative genomic

Protoc. Bioinform. 30:1.15.1-1.15.48. © 2010 by John Wiley & Sons, Inc.

Keywords: computer graphics • databases • genetic • genetic variation • ge sequence homology • genome • genome sequence

Access from NIH at
http://onlinelibrary.wiley.com/book/10.1002/0471250953