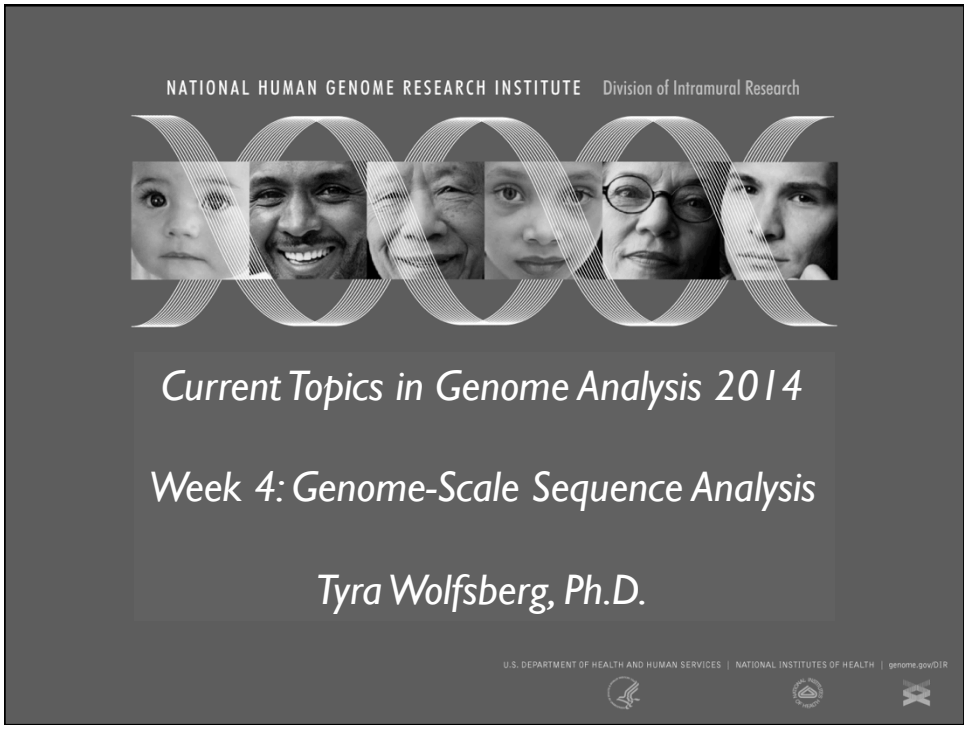
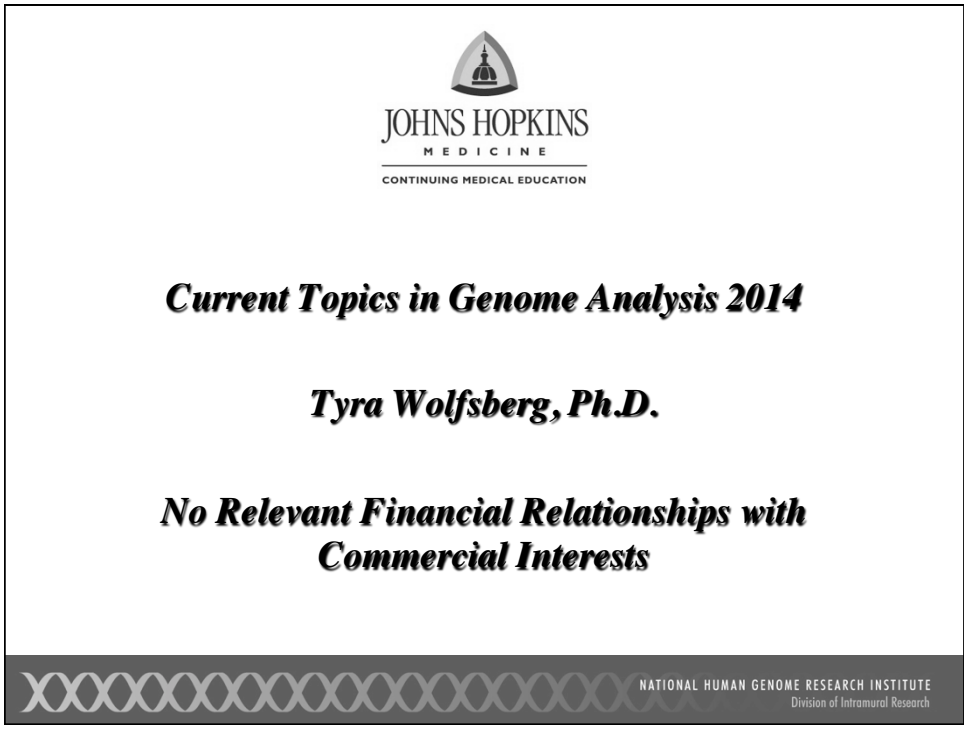

NATIONAL HUMAN GENOME RESEARCH INSTITUTE Division of Intramural Research
Current Topics in Genome Analysis 2014
Week 4: Genome-Scale Sequence Analysis
Tyra Wolfsberg, Ph.D.
U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES | NATIONAL INSTITUTES OF HEALTH | genome.gov/DIR


JOHNS HOPKINS
MEDICINE
CONTINUING MEDICAL EDUCATION
Current Topics in Genome Analysis 2014
Tyra Wolfsberg, Ph.D.
No Relevant Financial Relationships with Commercial Interests
NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research

Graphical Genome Browsers

- UCSC Genome Browser
  http://genome.ucsc.edu
- Ensembl
  http://www.ensembl.org

Web-based access to genome data

- BioMart
  http://www.ensembl.org/biomart
- Galaxy
  https://usegalaxy.org

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

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
- Updated assemblies not available immediately in the Genome Browsers
  - "Pre-release" assemblies and annotations
    - UCSC: *http://genome-preview.cse.ucsc.edu/*
    - pre!Ensembl: *http://pre.ensembl.org/*
  - UCSC and Ensembl provide archive of old assemblies
- IF YOU ARE COMPARING DATA FROM DIFFERENT GENOME BROWSERS, MAKE SURE YOU ARE LOOKING AT THE SAME VERSION OF THE ASSEMBLY

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GRCh38 human genome assembly

- First new human genome assembly in 4 years
  - Released in December, 2013
  - Fixes sequences and misassembled regions
  - Fills or reduces gaps
- 261 alternate loci
  - Many from LRC/KIR area of chr19 and MHC region on chr6
  - Example: chr6_GL000250v2_alt
- Preliminary browser (hg38) available at UCSC
- Posted on pre!Ensembl later in March
- UCSC's liftOver converts coordinates between assembles

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UCSC

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

http://genome.ucsc.edu

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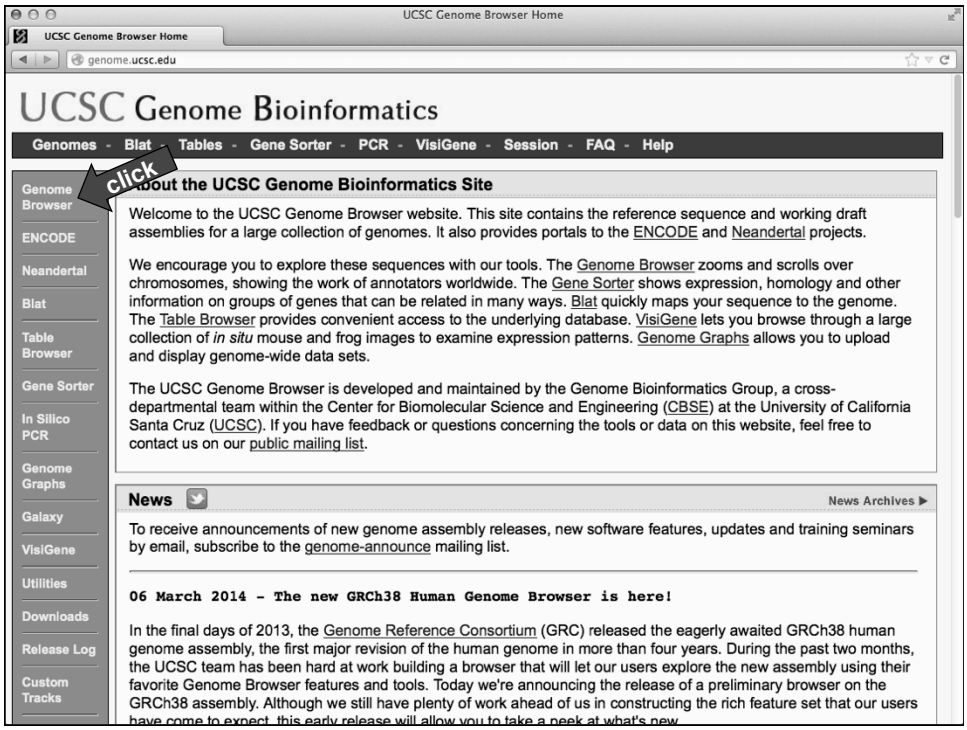

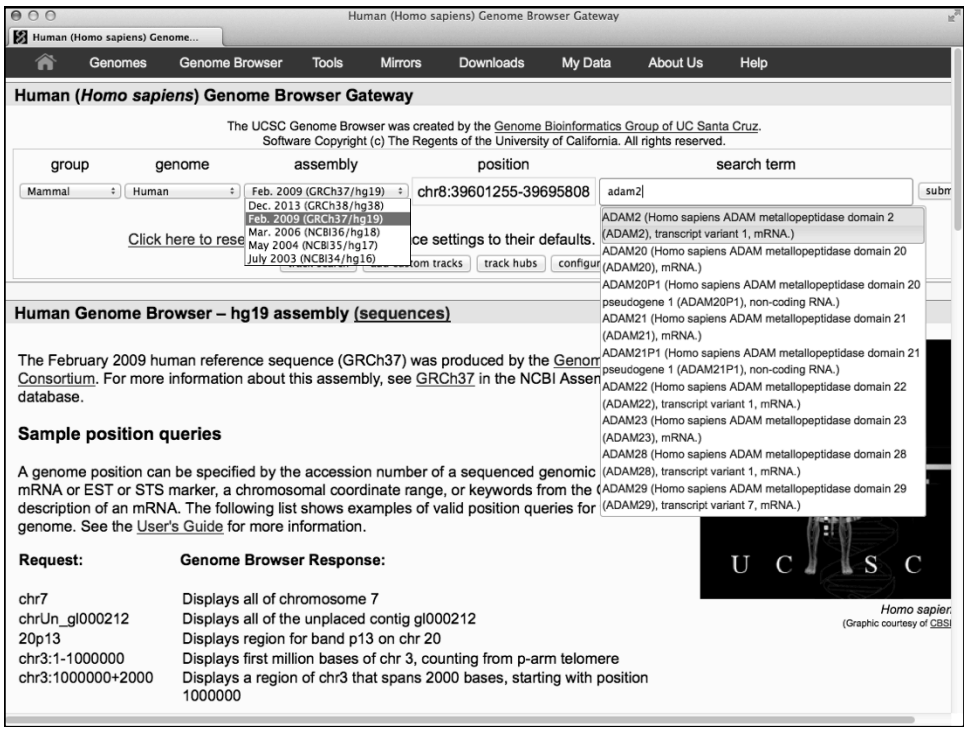
Human (Homo sapiens) Genome Browser Gateway
Genomes
Genome Browser
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Mirrors
Downloads
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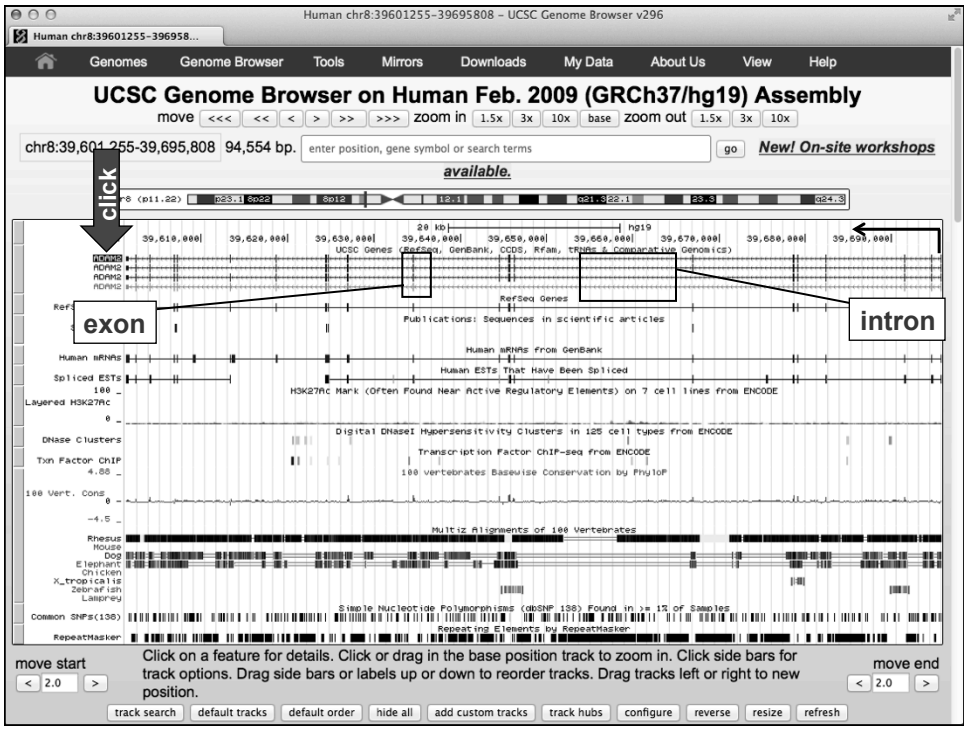
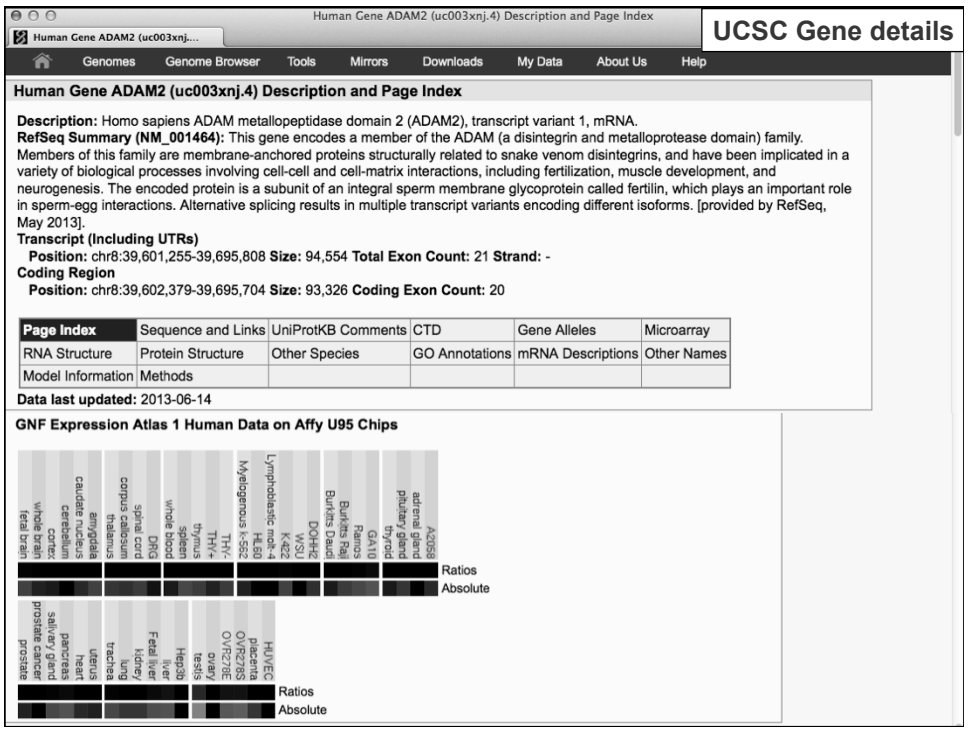
About Us
Help
Human (Homo sapiens) Genome Browser Gateway
The UCSC Genome Browser was created by the Genome Bioinformatics Group of UC Santa Cruz.
Software Copyright (c) The Regents of the University of California. All rights reserved.
group
genome
assembly
position
search term
Mammal
Human
Feb. 2009 (GRCh37/hg19)
chr8:39601255-39695808
adam2
Dec. 2013 (GRCh38/hg38)
Feb. 2009 (GRCh37/hg19)
Mar. 2006 (NCBI36/hg18)
May 2004 (NCBI35/hg17)
July 2003 (NCBI34/hg16)
ADAM2 (Homo sapiens ADAM metallopeptidase domain 2 (ADAM2), transcript variant 1, mRNA.)
ADAM20 (Homo sapiens ADAM metallopeptidase domain 20 (ADAM20), mRNA.)
ADAM20P1 (Homo sapiens ADAM metallopeptidase domain 20 pseudogene 1 (ADAM20P1), non-coding RNA.)
ADAM21 (Homo sapiens ADAM metallopeptidase domain 21 (ADAM21), mRNA.)
ADAM21P1 (Homo sapiens ADAM metallopeptidase domain 21 pseudogene 1 (ADAM21P1), non-coding RNA.)
ADAM22 (Homo sapiens ADAM metallopeptidase domain 22 (ADAM22), transcript variant 1, mRNA.)
ADAM23 (Homo sapiens ADAM metallopeptidase domain 23 (ADAM23), mRNA.)
ADAM28 (Homo sapiens ADAM metallopeptidase domain 28 (ADAM28), transcript variant 1, mRNA.)
ADAM29 (Homo sapiens ADAM metallopeptidase domain 29 (ADAM29), transcript variant 7, mRNA.)
Click here to rese
ce settings to their defaults.
track hubs
Human Genome Browser – hg19 assembly (sequences)
The February 2009 human reference sequence (GRCh37) was produced by the Genom
Consortium. For more information about this assembly, see GRCh37 in the NCBI Assen
database.
Sample position queries
A genome position can be specified by the accession number of a sequenced genomic
mRNA or EST or STS marker, a chromosomal coordinate range, or keywords from the
description of an mRNA. The following list shows examples of valid position queries for
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
U C S C
Homo sapien
(Graphic courtesy of CBSI


Human chr8:39601255-39695808 – UCSC Genome Browser v296
Genomes
Genome Browser
Tools
Mirrors
Downloads
My Data
About Us
View
Help
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
move
zoom in
zoom out
chr8:39,601,255-39,695,808
94,554 bp.
enter position, gene symbol or search terms
go
New! On-site workshops available.
click
exon
intron
UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)
RefSeq Genes
Publications: Sequences in scientific articles
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 in 125 cell types from ENCODE
Transcription Factor ChIP-seq from ENCODE
100 vertebrates Basewise Conservation by PhyloP
Multiz Alignments of 100 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP 138) 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.
track search
default tracks
default order
hide all
add custom tracks
track hubs
configure
reverse
resize
refresh


UCSC Gene details
Human Gene ADAM2 (uc003xnj.4) Description and Page Index
Genomes
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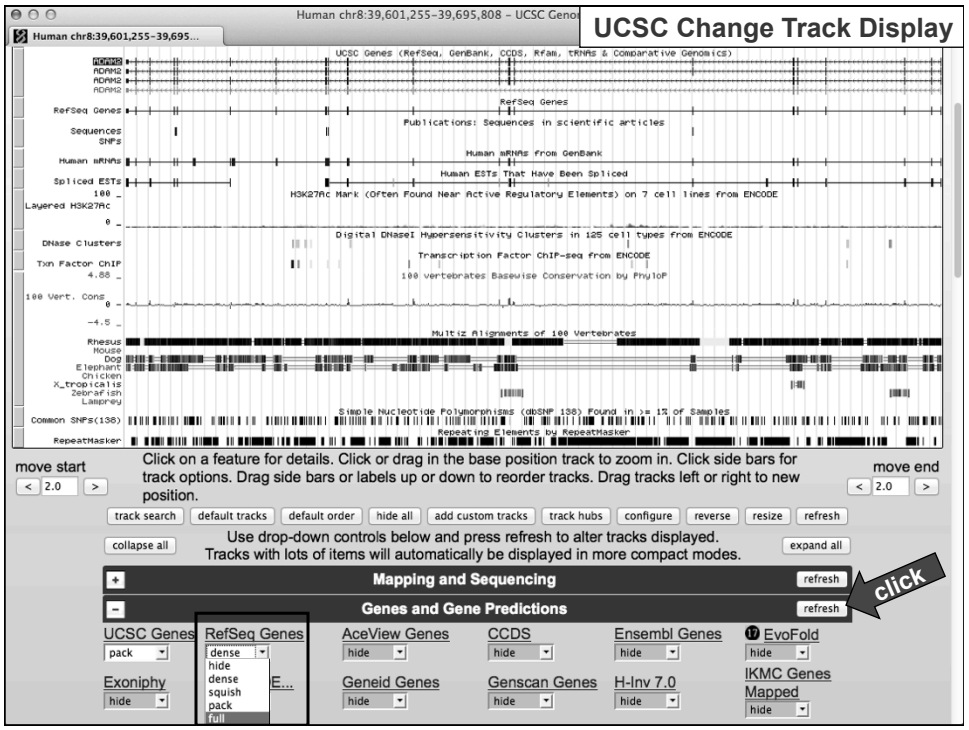
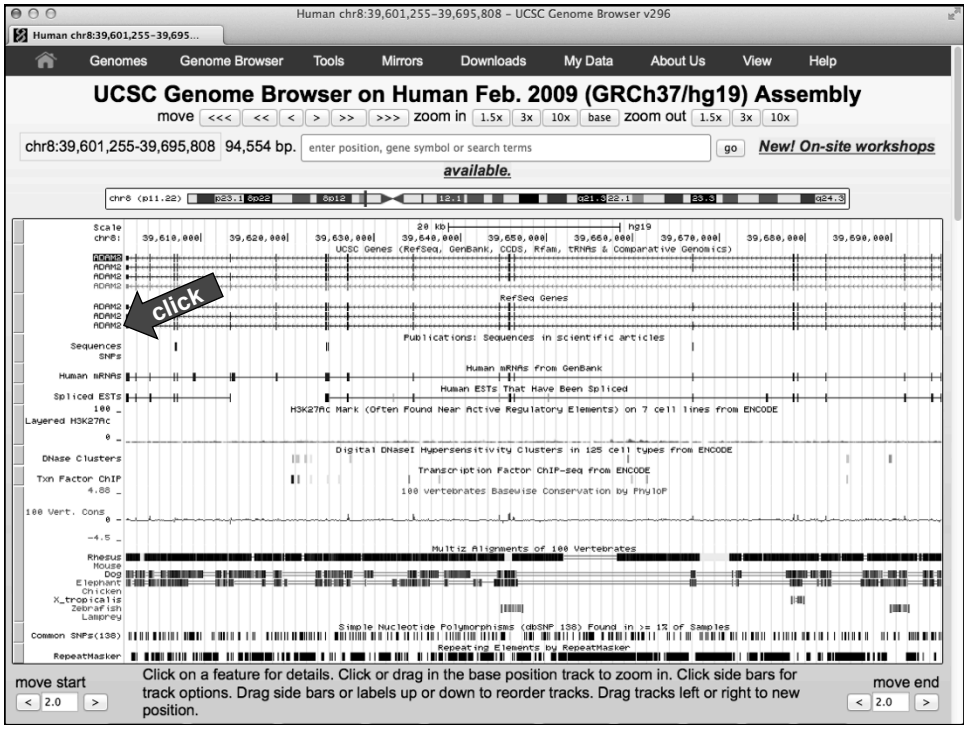
Downloads
My Data
About Us
Help
Human Gene ADAM2 (uc003xnj.4) Description and Page Index
Description: Homo sapiens ADAM metallopeptidase domain 2 (ADAM2), transcript variant 1, mRNA.
RefSeq Summary (NM_001464): This gene encodes a member of the ADAM (a disintegrin and metalloprotease domain) family. 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. The encoded protein is a subunit of an integral sperm membrane glycoprotein called fertilin, which plays an important role in sperm-egg interactions. Alternative splicing results in multiple transcript variants encoding different isoforms. [provided by RefSeq, May 2013].
Transcript (Including UTRs)
Position: chr8:39,601,255-39,695,808 Size: 94,554 Total Exon Count: 21 Strand: -
Coding Region
Position: chr8:39,602,379-39,695,704 Size: 93,326 Coding Exon Count: 20
Page Index
Sequence and Links
UniProtKB Comments
CTD
Gene Alleles
Microarray
RNA Structure
Protein Structure
Other Species
GO Annotations
mRNA Descriptions
Other Names
Model Information
Methods
Data last updated: 2013-06-14
GNF Expression Atlas 1 Human Data on Affy U95 Chips
Ratios
Absolute
Ratios
Absolute


UCSC Change Track Display
UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)
RefSeq Genes
Publications: Sequences in scientific articles
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 in 125 cell types from ENCODE
Transcription Factor ChIP-seq from ENCODE
100 vertebrates Basewise Conservation by PhyloP
Multiz Alignments of 100 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP 138) 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.
track search
default tracks
default order
hide all
add custom tracks
track hubs
configure
reverse
resize
refresh
collapse all
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.
expand all
Mapping and Sequencing
Genes and Gene Predictions
click
UCSC Genes
RefSeq Genes
AceView Genes
CCDS
Ensembl Genes
EvoFold
Exoniphy
Geneid Genes
Genscan Genes
H-Inv 7.0
IKMC Genes Mapped
hide
dense
squish
pack
full


Human chr8:39,601,255-39,695,808 - UCSC Genome Browser v296
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UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
move
zoom in
zoom out
chr8:39,601,255-39,695,808
94,554 bp.
go
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click
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Publications: Sequences in scientific articles
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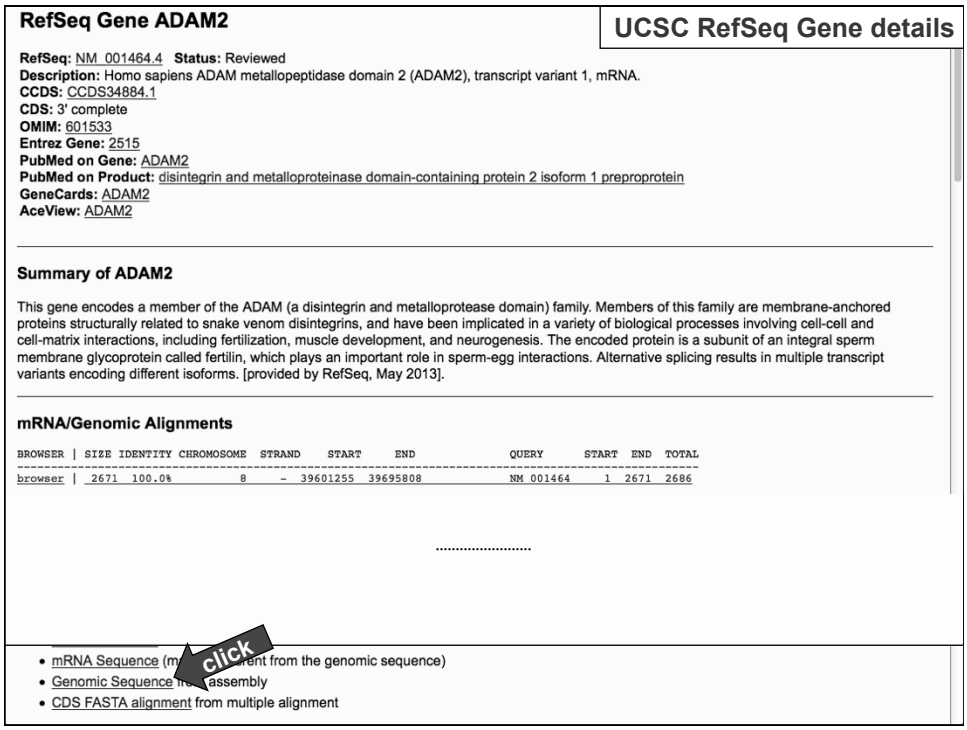
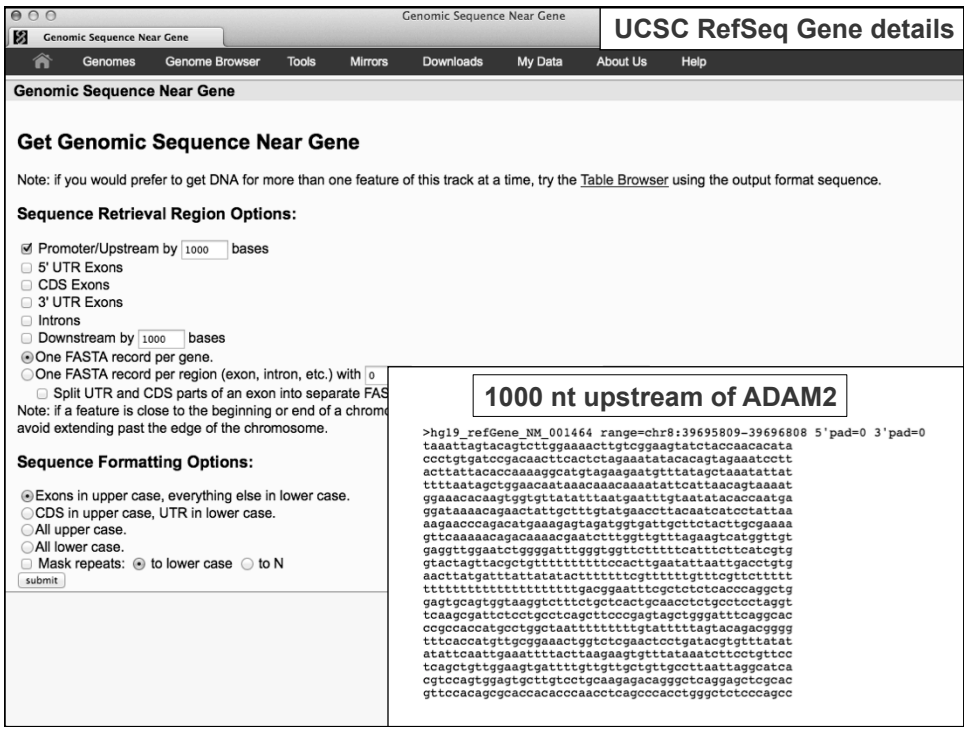
move start
move end
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.


RefSeq Gene ADAM2
UCSC RefSeq Gene details
RefSeq: NM_001464.4 Status: Reviewed
Description: Homo sapiens ADAM metallopeptidase domain 2 (ADAM2), transcript variant 1, 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 isoform 1 preproprotein
GeneCards: ADAM2
AceView: ADAM2
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. The encoded protein is a subunit of an integral sperm membrane glycoprotein called fertilin, which plays an important role in sperm-egg interactions. Alternative splicing results in multiple transcript variants encoding different isoforms. [provided by RefSeq, May 2013].
mRNA/Genomic Alignments
BROWSER | SIZE IDENTITY CHROMOSOME STRAND START END QUERY START END TOTAL
browser | 2671 100.0% 8 - 39601255 39695808 NM_001464 1 2671 2686
click
mRNA Sequence
Genomic Sequence
CDS FASTA alignment from multiple alignment


Genomic Sequence Near Gene
Genomic Sequence Near Gene
UCSC RefSeq Gene details
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About Us
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 FAS
Note: if a feature is close to the beginning or end of a chrom
avoid extending 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
>hg19_refGene_NM_001464 range=chr8:39695809-39696808 5'pad=0 3'pad=0
taaattagtacagtcttggaaaacttgtcggaagtatctaccaacacata
ccctgtgatccgacaacttcactctagaaatatacacagtagaaatcctt
acttattacaccaaaaggcatgtagaagaatgtttatagctaaatattat
ttttaatagctggaacaataaacaaacaaaatattcattaacagtaaaat
ggaaacacaagtggtgttatatttaatgaatttgtaatatacaccaatga
ggataaaacagaactattgctttgtatgaaccttacaatcatcctattaa
aagaacccagacatgaaagagtagatggtgattgcttctacttgcgaaaa
gttcaaaaacagacaaaacgaatctttggttgtttagaagtcatggttgt
gaggttggaatctggggatttgggtggttcttttttcatttcttcatcgtg
gtactagttacgctgtttttttttccacttgaatattaattgacctgtg
aacttatgatttattatatacttttttcgtttttgtttcgttcttttt
tttttttttttttttttttgacggaatttcgctctctcacccaggctg
gagtgcagtggtaaggtctttctgctcactgcaacctctgcctcctaggt
tcaagcgattctcctgcctcagcttcccgagtagctgggatttcaggcac
ccgccaccatgcctggctaatttttttttgtatttttagtacagacgggg
tttcaccatgttgcggaaactggtctcgaactcctgatacgtgtttatat
atattcaattgaaatttacttaagaagtgtttataaatcttcctgttcc
tcagctgttggaagtgattttgttgttgctgttgccttaattaggcatca
cgtccagtggagtgcttgtcctgcaagagacagggctcaggagctcgcac
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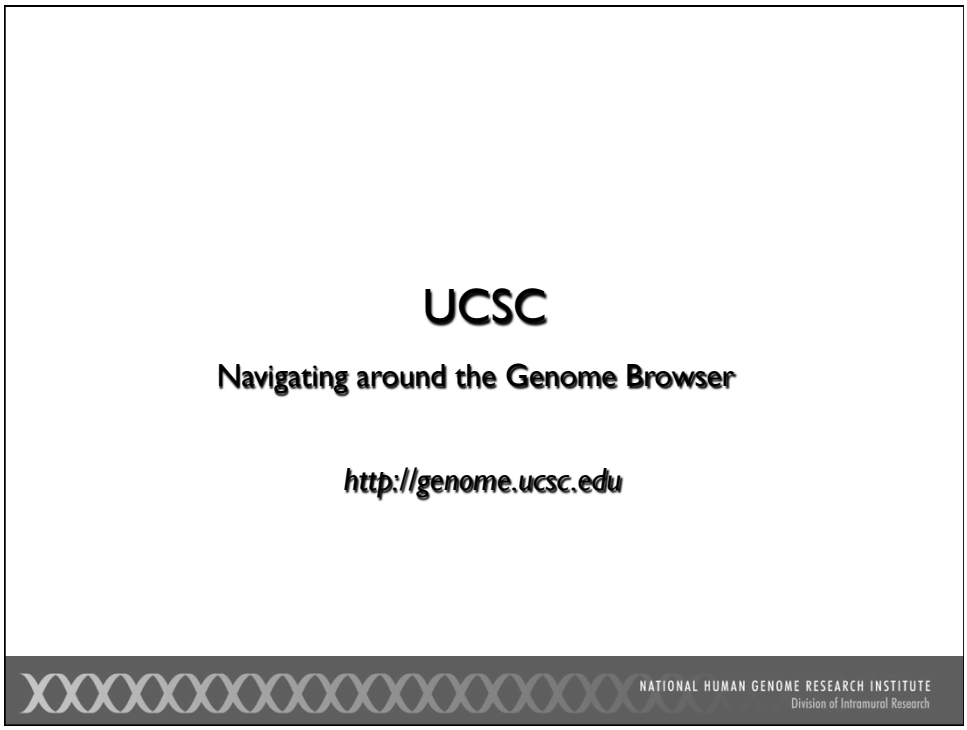
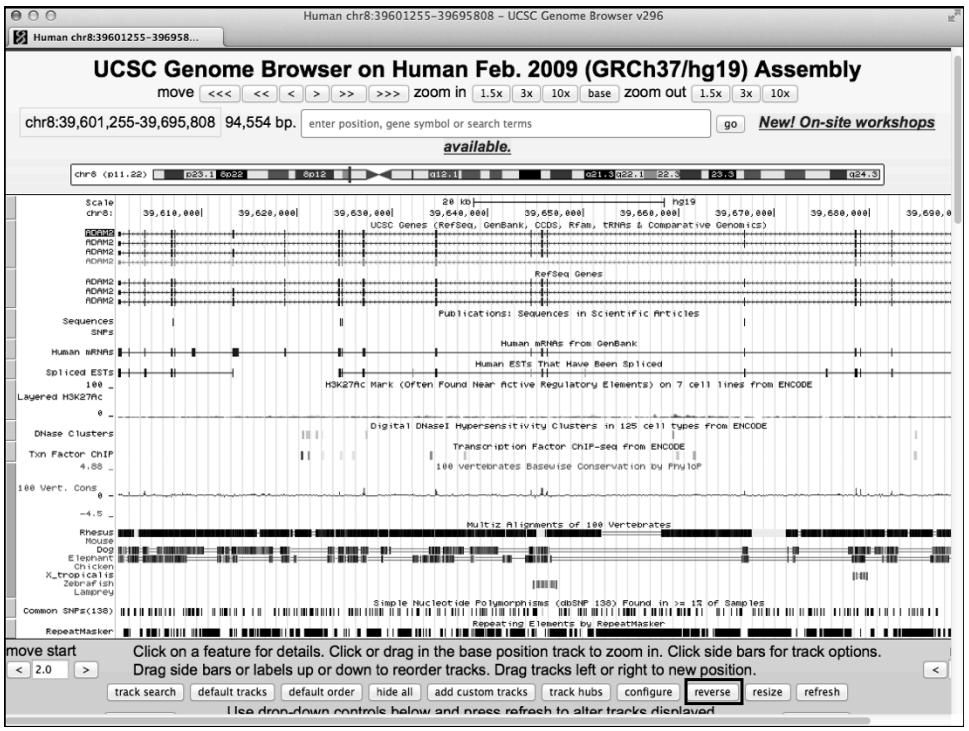

UCSC
Navigating around the Genome Browser
http://genome.ucsc.edu
NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research


Human chr8:39601255-39695808 - UCSC Genome Browser v296
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
move zoom in zoom out
chr8:39,601,255-39,695,808 94,554 bp.
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RefSeq Genes
Publications: Sequences in Scientific Articles
Human mRNAs from GenBank
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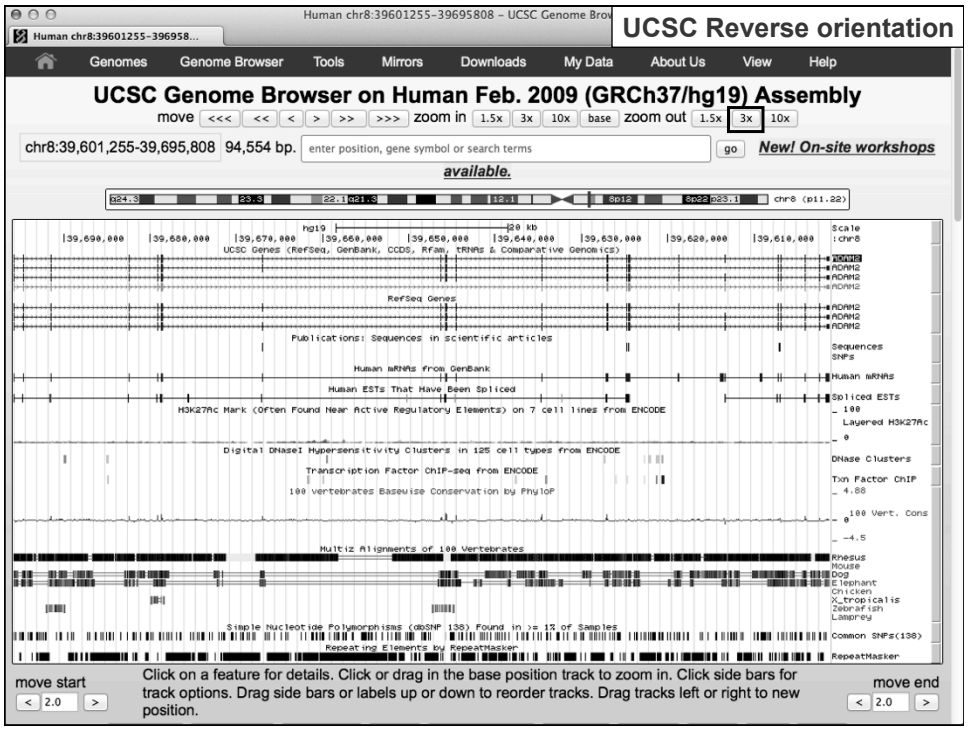
100 vertebrates Basewise Conservation by PhyloP
Multiz Alignments of 100 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples
Repeating Elements by RepeatMasker
move start
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options.
Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.
track search default tracks default order hide all add custom tracks track hubs configure reverse resize refresh


UCSC Reverse orientation
Genomes Genome Browser Tools Mirrors Downloads My Data About Us View Help
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
chr8:39,601,255-39,695,808 94,554 bp.
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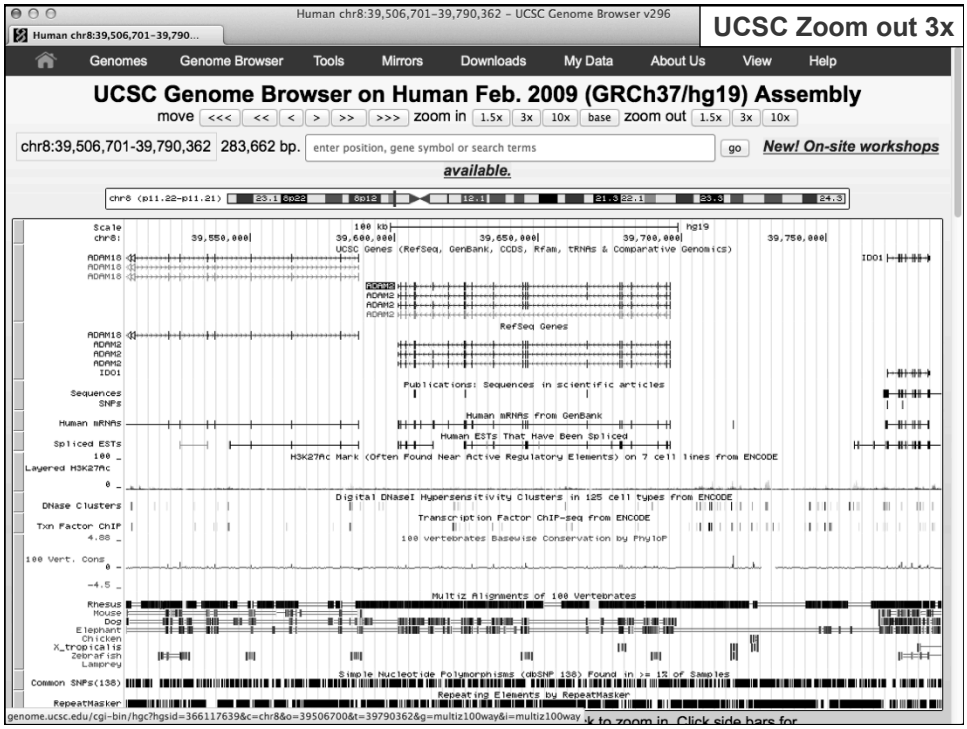
move start
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position.
move end


UCSC Zoom out 3x
Human chr8:39,506,701-39,790,362 – UCSC Genome Browser v296
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UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
move
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chr8:39,506,701-39,790,362
283,662 bp.
enter position, gene symbol or search terms
go
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100 vertebrates Basewise Conservation by PhyloP
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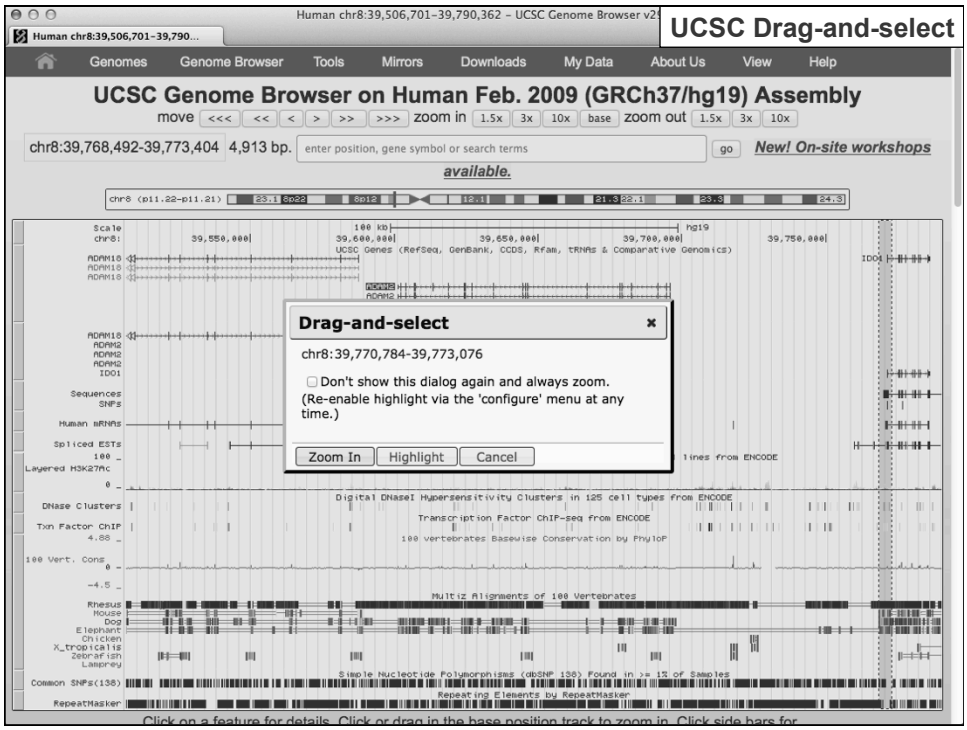
Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples
Repeating Elements by RepeatMasker


UCSC Drag-and-select
Human chr8:39,506,701-39,790,362 – UCSC Genome Browser v25
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UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
move
zoom in
zoom out
chr8:39,768,492-39,773,404
4,913 bp.
enter position, gene symbol or search terms
go
New! On-site workshops
available.
Drag-and-select
chr8:39,770,784-39,773,076
Don't show this dialog again and always zoom.
(Re-enable highlight via the 'configure' menu at any time.)
Zoom In
Highlight
Cancel
UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)
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Transcription Factor ChIP-seq from ENCODE
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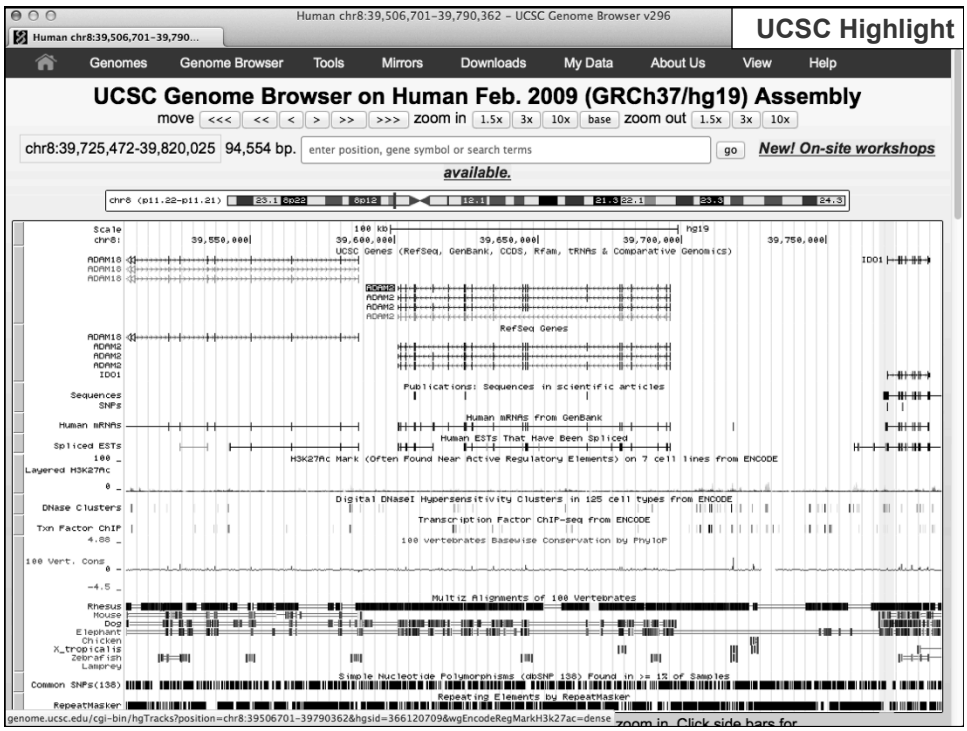
Multiz Alignments of 100 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples
Repeating Elements by RepeatMasker
Click on a feature for details. Click or drag in the base position track to zoom in. Click side bars for


UCSC Highlight
Human chr8:39,506,701-39,790,362 - UCSC Genome Browser v296
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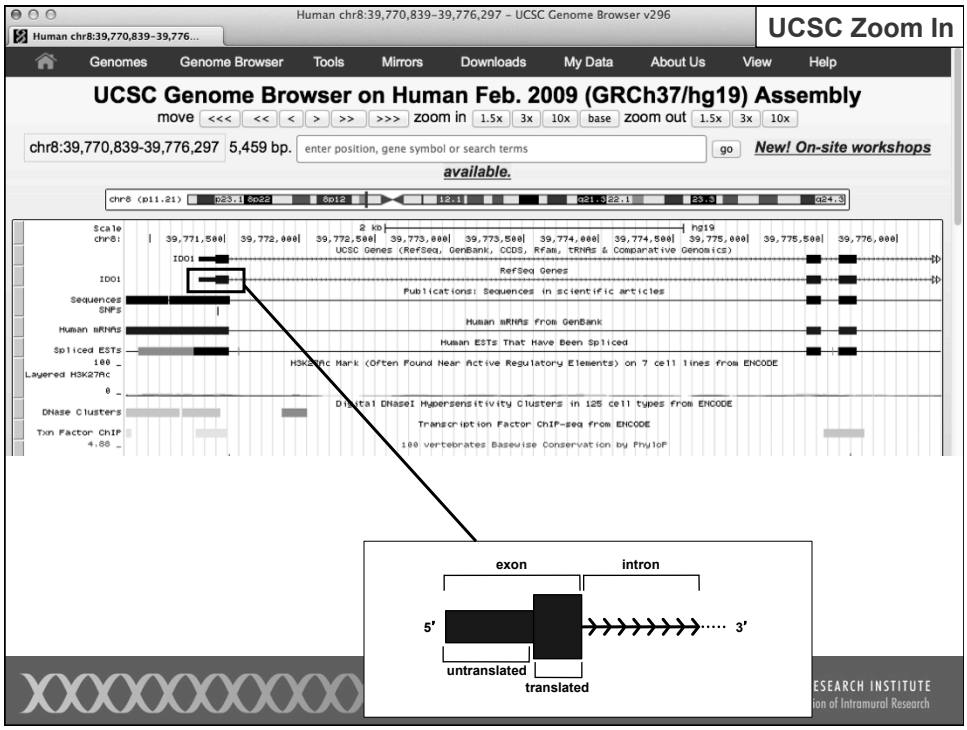
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
chr8:39,725,472-39,820,025
94,554 bp.
New! On-site workshops
available.


UCSC Zoom In
Human chr8:39,770,839-39,776,297 - UCSC Genome Browser v296
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Genome Browser
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Downloads
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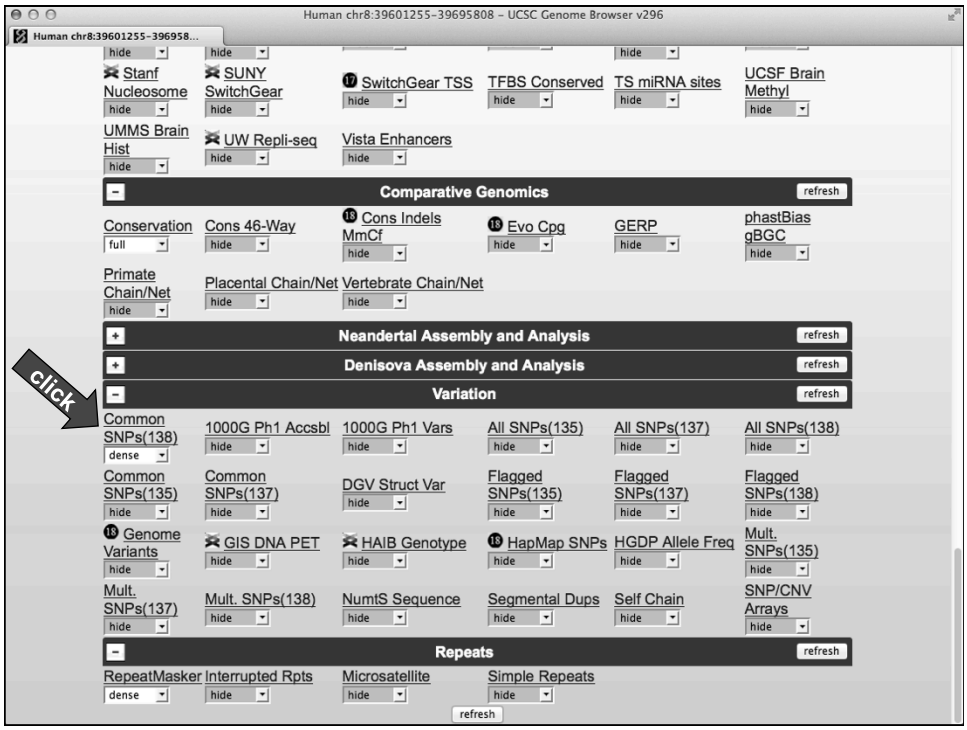
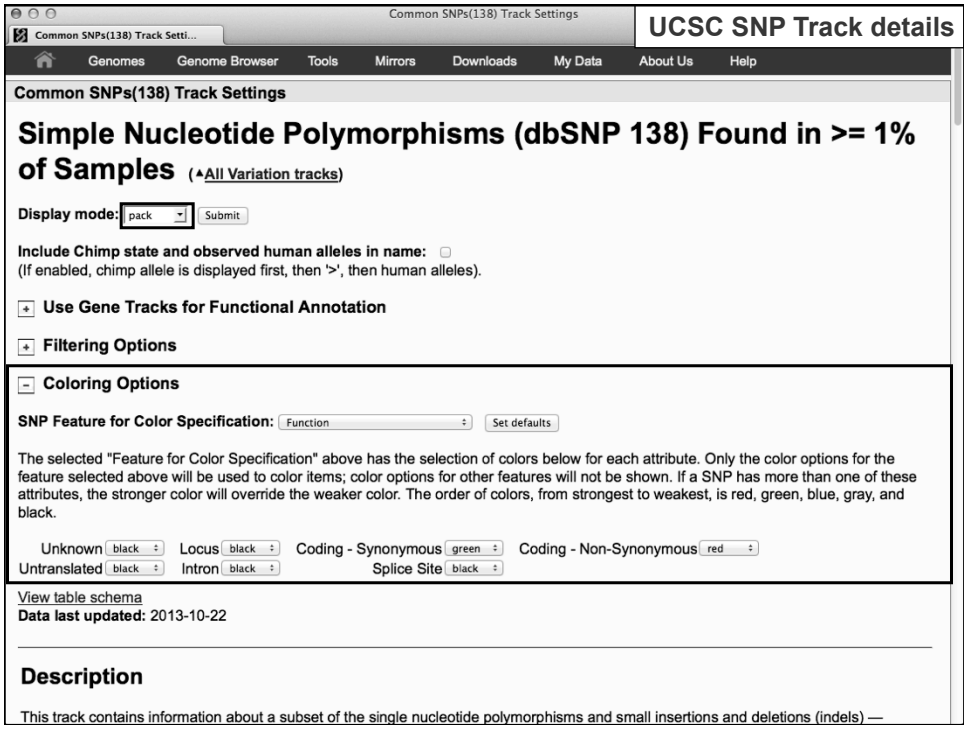
About Us
View
Help
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
chr8:39,770,839-39,776,297
5,459 bp.
New! On-site workshops
available.
exon
intron
5'
3'
untranslated
translated

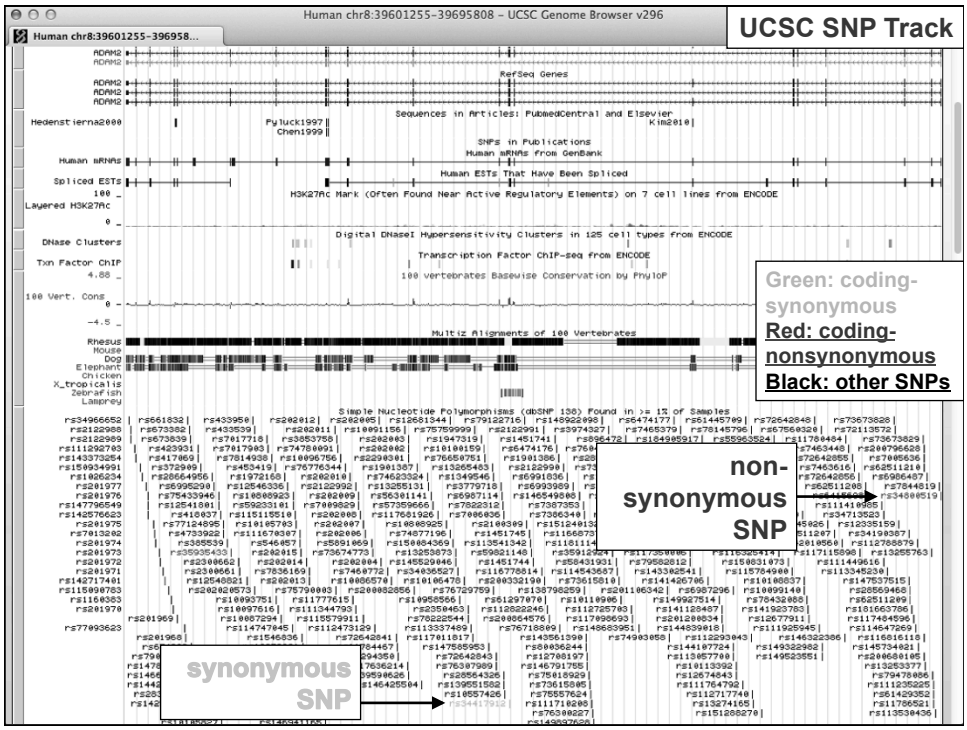
UCSC

Configure Track on the Genome Browser

http://genome.ucsc.edu




UCSC SNP Track details
Common SNPs(138) Track Settings
Genomes
Genome Browser
Tools
Mirrors
Downloads
My Data
About Us
Help
Common SNPs(138) Track Settings
Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples
(▲All Variation tracks)
Display mode: pack Submit
Include Chimp state and observed human alleles in name:
(If enabled, chimp allele is displayed first, then '>', then human alleles).
Use Gene Tracks for Functional Annotation
Filtering Options
Coloring Options
SNP Feature for Color Specification: Function Set defaults
The selected "Feature for Color Specification" above has the selection of colors below for each attribute. Only the color options for the feature selected above will be used to color items; color options for other features will not be shown. If a SNP has more than one of these attributes, the stronger color will override the weaker color. The order of colors, from strongest to weakest, is red, green, blue, gray, and black.
Unknown black Locus black Coding - Synonymous green Coding - Non-Synonymous red
Untranslated black Intron black Splice Site black
View table schema
Data last updated: 2013-10-22
Description
This track contains information about a subset of the single nucleotide polymorphisms and small insertions and deletions (indels) —


UCSC SNP Track
Human chr8:39601255-39695808 – UCSC Genome Browser v296
RefSeq Genes
Sequences in Articles: PubmedCentral and Elsevier
SNPs in Publications
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 in 125 cell types from ENCODE
Transcription Factor ChIP-seq from ENCODE
100 vertebrates Basewise Conservation by PhyloP
Multiz Alignments of 100 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples
Green: coding-synonymous
Red: coding-nonsynonymous
Black: other SNPs
non-synonymous SNP
synonymous SNP

UCSC

ENCODE tracks

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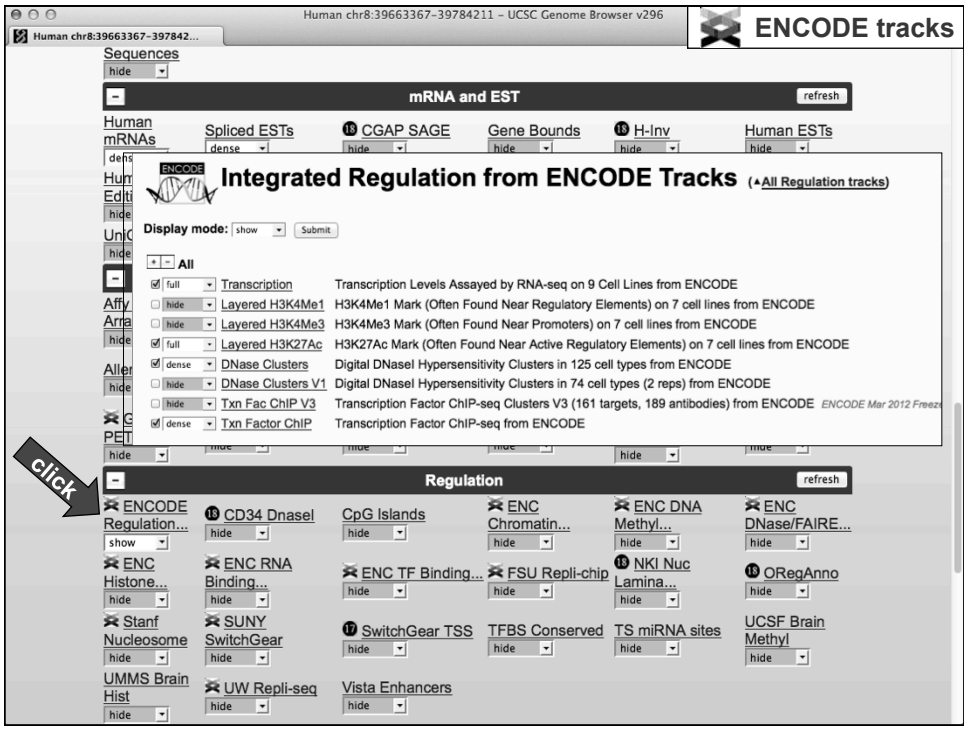

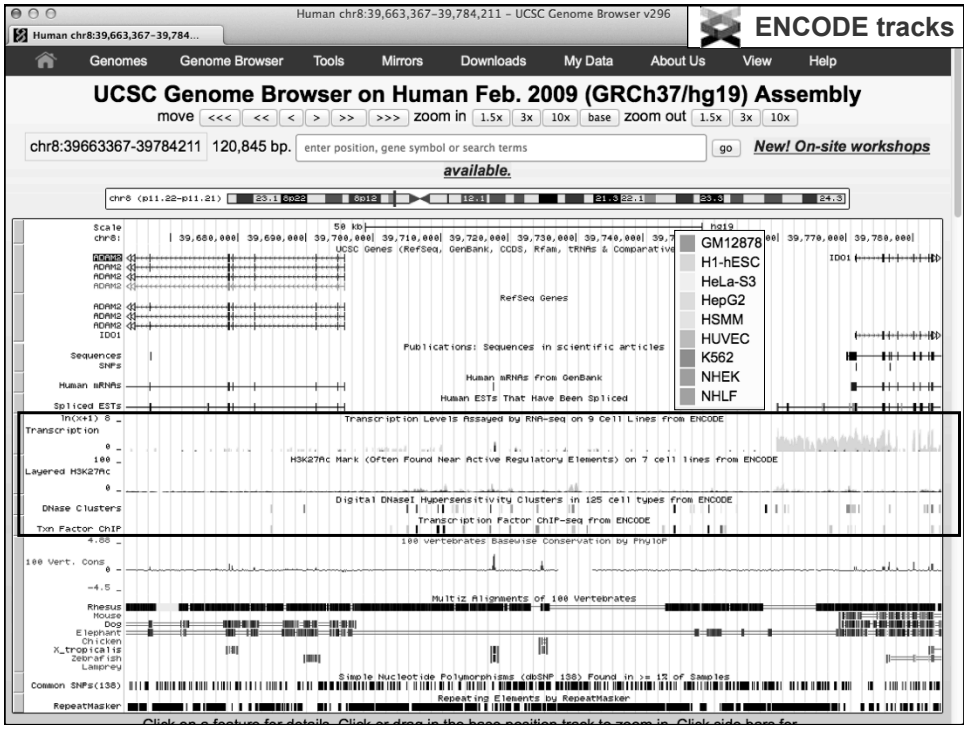
Human chr8:39,663,367-39,784,211 - UCSC Genome Browser v296
ENCODE tracks
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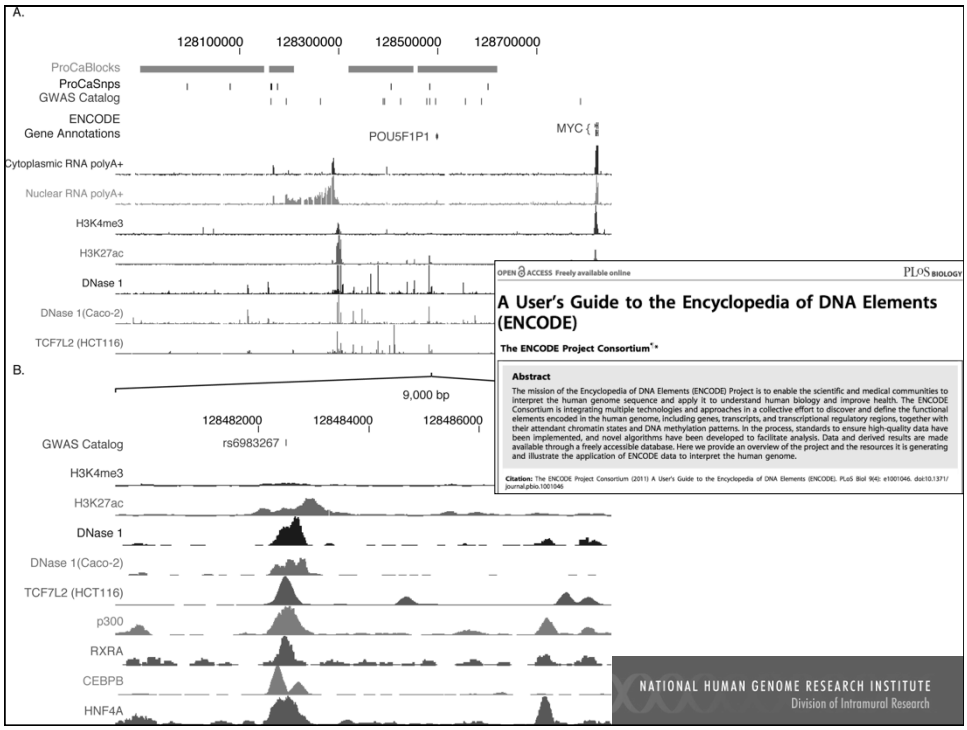
UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly
chr8:39663367-39784211
120,845 bp.
New! On-site workshops
available.
GM12878
H1-hESC
HeLa-S3
HepG2
HSMM
HUVEC
K562
NHEK
NHLF
RefSeq Genes
Publications: Sequences in scientific articles
Human mRNAs from GenBank
Human ESTs That Have Been Spliced
Transcription Levels Assayed by RNA-seq on 9 Cell Lines from ENCODE
H3K27Ac Mark (Often Found Near Active Regulatory Elements) on 7 cell lines from ENCODE
Digital DNaseI Hypersensitivity Clusters in 125 cell types from ENCODE
Transcription Factor ChIP-seq from ENCODE
100 vertebrates Basewise Conservation by PhyloP
Multiz Alignments of 100 Vertebrates
Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples
Repeating Elements by RepeatMasker


A.
ProCaBlocks
ProCaSnps
GWAS Catalog
ENCODE
Gene Annotations
POU5F1P1
MYC
Cytoplasmic RNA polyA+
Nuclear RNA polyA+
H3K4me3
H3K27ac
DNase 1
DNase 1(Caco-2)
TCF7L2 (HCT116)
B.
9,000 bp
GWAS Catalog
rs6983267
p300
RXRA
CEBPB
HNF4A
A User's Guide to the Encyclopedia of DNA Elements (ENCODE)
The ENCODE Project Consortium
Abstract
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UCSC

Find a chicken homolog of a human protein

http://genome.ucsc.edu

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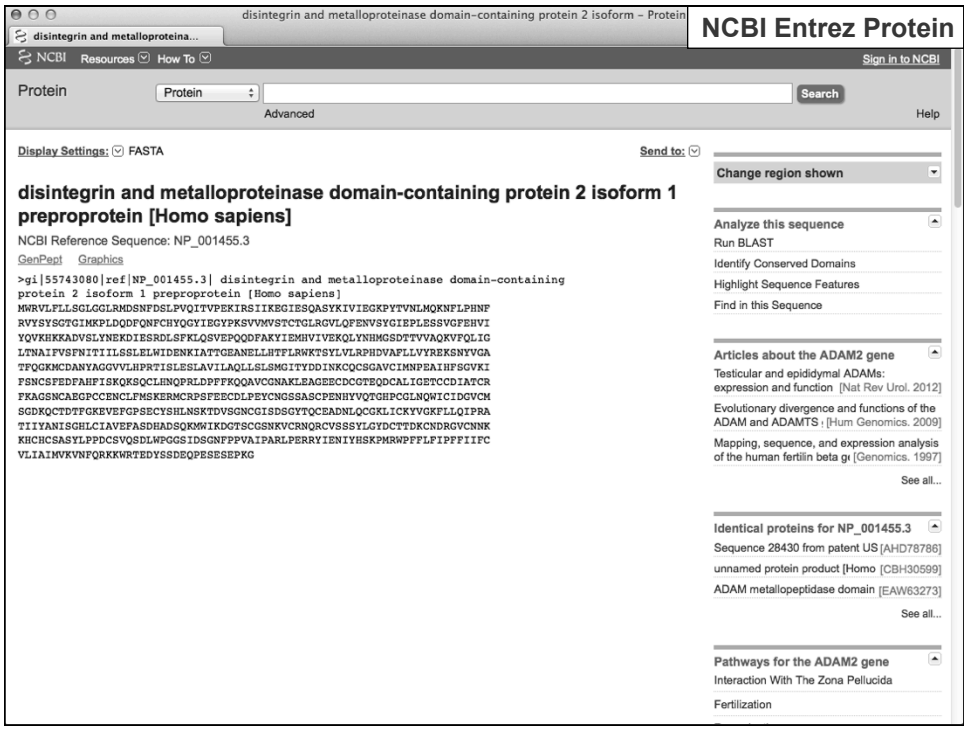
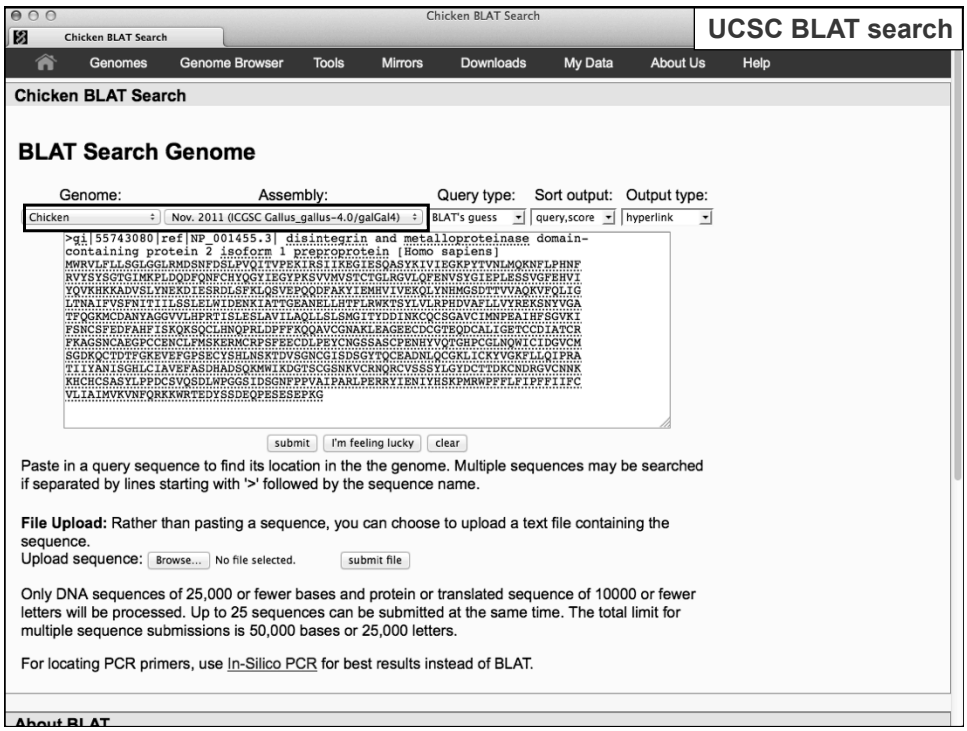
NCBI Entrez Protein

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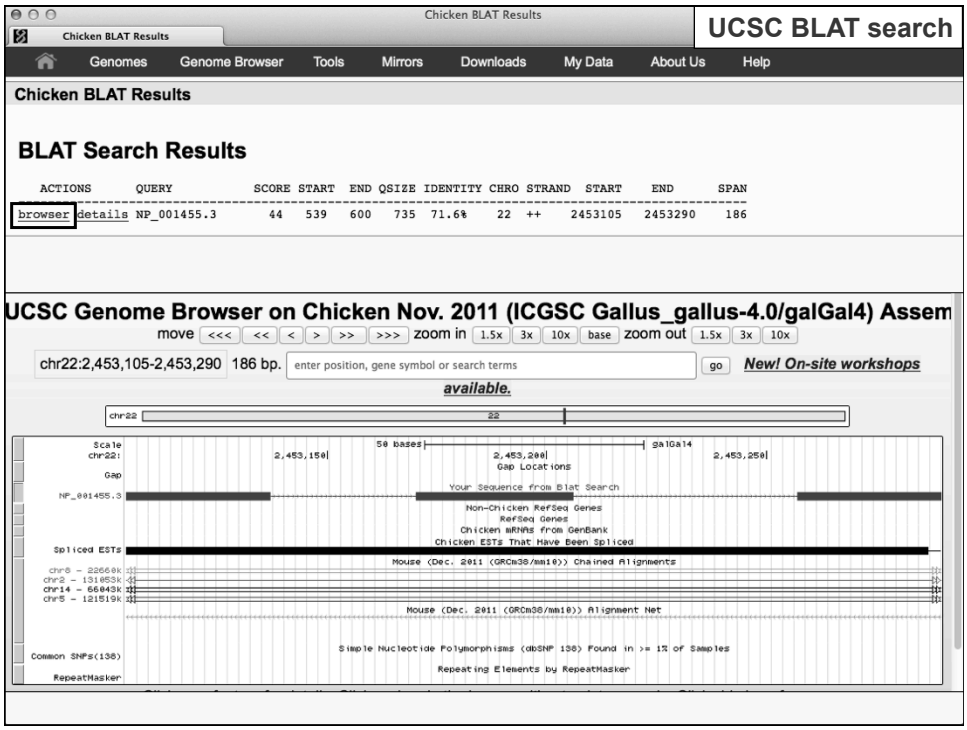
Help
Chicken BLAT Search
BLAT Search Genome
Genome:
Assembly:
Query type:
Sort output:
Output type:
Chicken
Nov. 2011 (ICGSC Gallus_gallus-4.0/galGal4)
BLAT's guess
query,score
hyperlink
>gi|55743080|ref|NP_001455.3| disintegrin and metalloproteinase domain-containing protein 2 isoform 1 preproprotein [Homo sapiens]
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...
No file selected.
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.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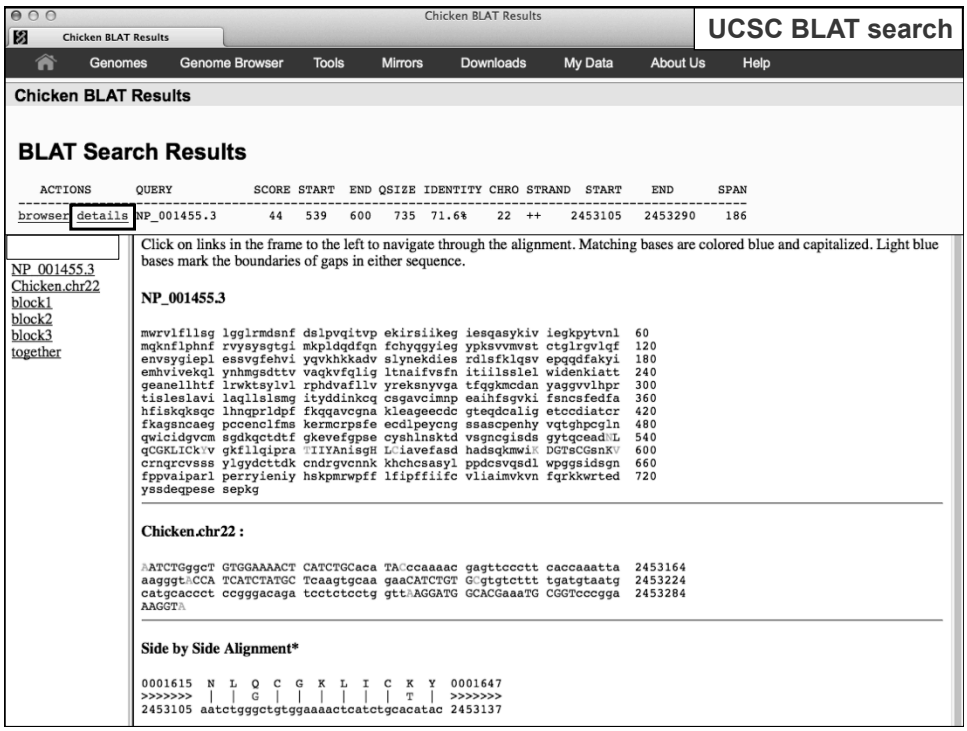
Chicken BLAT Results
BLAT Search Results
ACTIONS QUERY SCORE START END QSIZE IDENTITY CHRO STRAND START END SPAN
browser details NP_001455.3 44 539 600 735 71.6% 22 ++ 2453105 2453290 186
UCSC Genome Browser on Chicken Nov. 2011 (ICGSC Gallus_gallus-4.0/galGal4) Assem
move
zoom in
zoom out
chr22:2,453,105-2,453,290 186 bp.
enter position, gene symbol or search terms
go
New! On-site workshops available.
Gap Locations
Your Sequence from Blat Search
NP_001455.3
Non-Chicken RefSeq Genes
RefSeq Genes
Chicken mRNAs from GenBank
Chicken ESTs That Have Been Spliced
Spliced ESTs
Mouse (Dec. 2011 (GRCm38/mm10)) Chained Alignments
Mouse (Dec. 2011 (GRCm38/mm10)) Alignment Net
Simple Nucleotide Polymorphisms (dbSNP 138) Found in >= 1% of Samples
Repeating Elements by RepeatMasker
Common SNPs(138)
RepeatMasker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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% 22 ++ 2453105 2453290 186
NP_001455.3
Chicken.chr22
block1
block2
block3
together
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 ctglrgvlqf 120
envsygiepl essvgfehvi yqvkhkkadv slynekdies rdlsfklqsv epqqdfakyi 180
emhvivekql ynhmgsdttv vaqkvfqlig ltnaifvsfn itiilsslel widenkiatt 240
geanellhtf lrwktsylvl rphdvafllv yreksnyvga tfqgkmcdan yaggvvlhpr 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 gytgceadNL 540
qCGKLICkv gkfllqipra KIIYAnisgH LKiavefasd hadsqkmwiK DGTsCGsnKV 600
crnqrcvsss ylgydcttdk cndrgvcnnk khchcsasyl ppdcsvqsdl wpggsidsgn 660
fppvaiparl perryieniy hskpmrwpff lfipffiifc vliaimvkvn fqrkkwrted 720
yssdeqpese sepkg
Chicken.chr22 :
AATCTGggcT GTGGAAAACT CATCTGCaca TACccaaaac gagttccctt caccaaatta 2453164
aagggtACCA TCATCTATGC Tcaagtgcaa gaaCATCTGT GCgtgtcttt tgatgtaatg 2453224
catgcaccct ccgggacaga tcctctcctg gttAAGGATG GCACGaaaTG CGGTcccgga 2453284
AAGGTA
Side by Side Alignment*
0001615 N L Q C G K L I C K Y 0001647
>>>>>>> | | G | | | | | T | >>>>>>>
2453105 aatctgggctgtggaaaactcatctgcacatac 2453137


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Add your own custom tracks
http://genome.ucsc.edu
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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

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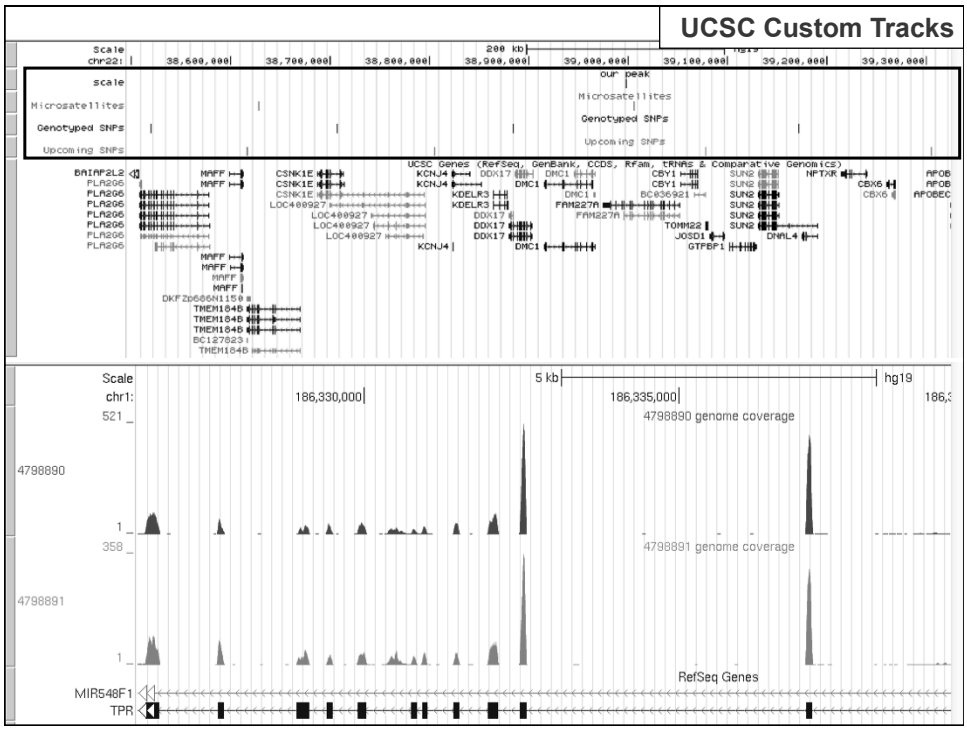

Types of UCSC custom tracks

- **Upload annotation data from your computer**
  - Tracks viewable only from your computer
  - Discarded after 48 hours
- **Post annotation data to your Web site**
  - Tracks can be shared with anyone
  - Never discarded
- **Create a Session with specific track combinations, including custom tracks**
  - Session can be shared or non-shared
  - Session persists for 4 months; custom tracks for 48 hours

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

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Table Browser

http://genome.ucsc.edu

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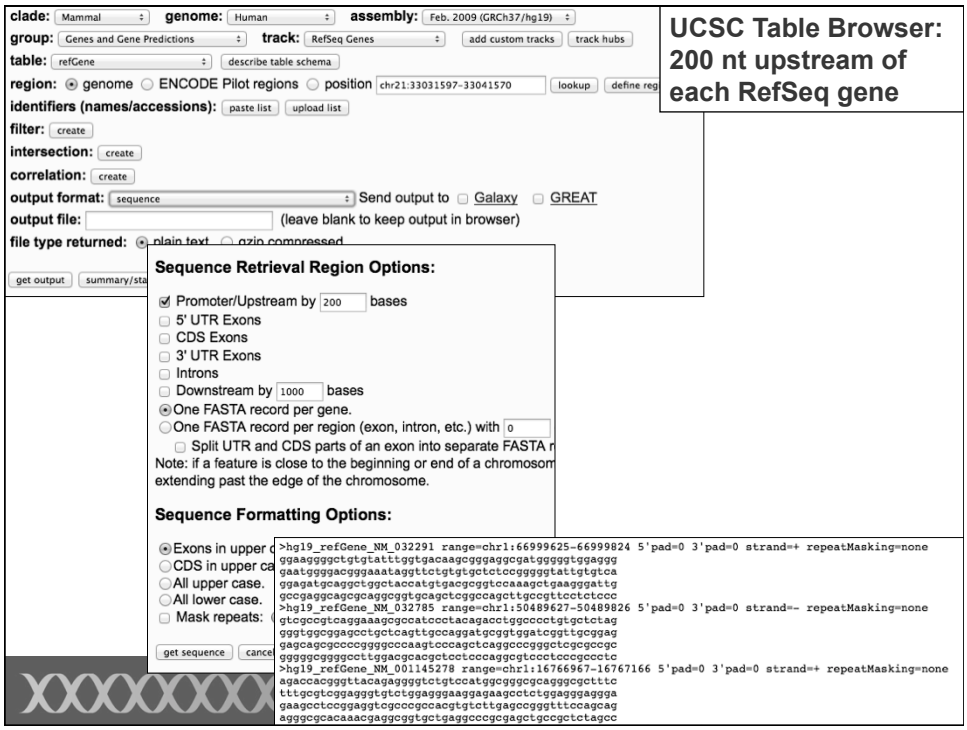
UCSC Table Browser

- Download track in text format or create custom tracks
- Retrieve DNA sequence
  - Get sequence 200 nt upstream of each RefSeq gene
- Calculate intersections between tracks
  - List all SNPs in a RefSeq 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

Variant Effect Predictor (VEP)

http://www.ensembl.org

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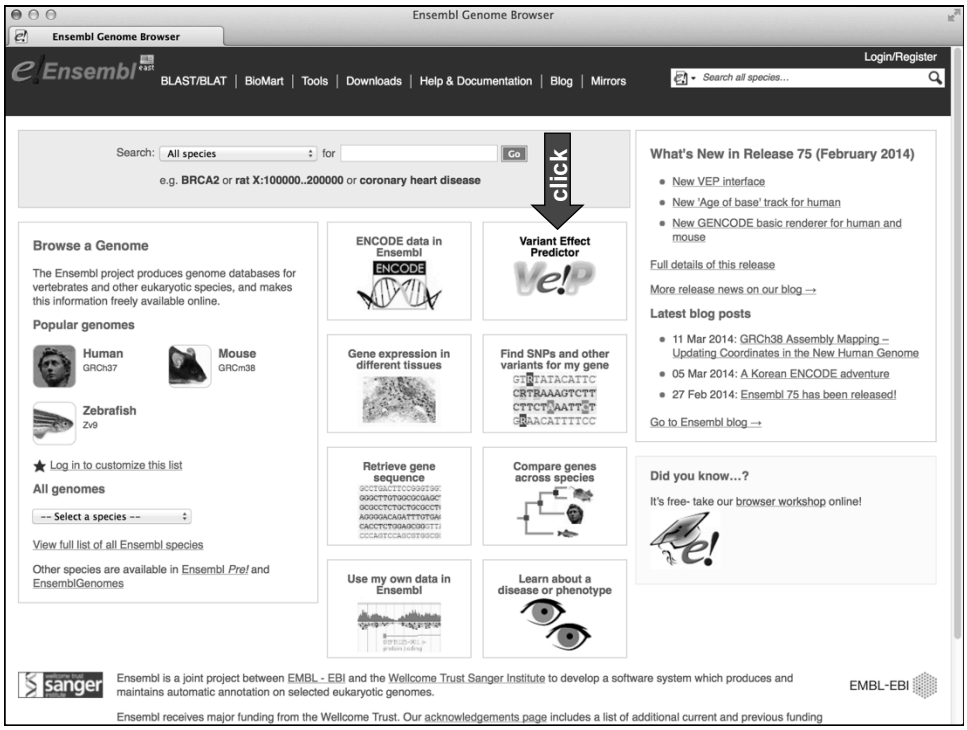

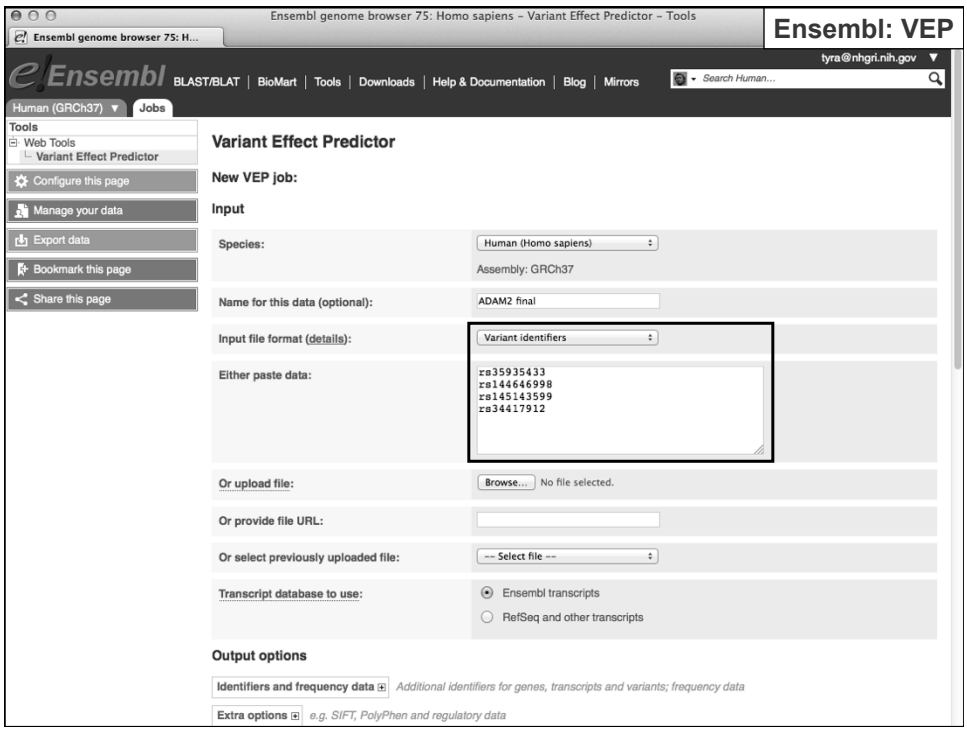
Ensembl: VEP
Ensembl genome browser 75: Homo sapiens - Variant Effect Predictor - Tools
tyra@nhgri.nih.gov
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Variant Effect Predictor
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Manage your data
Export data
Bookmark this page
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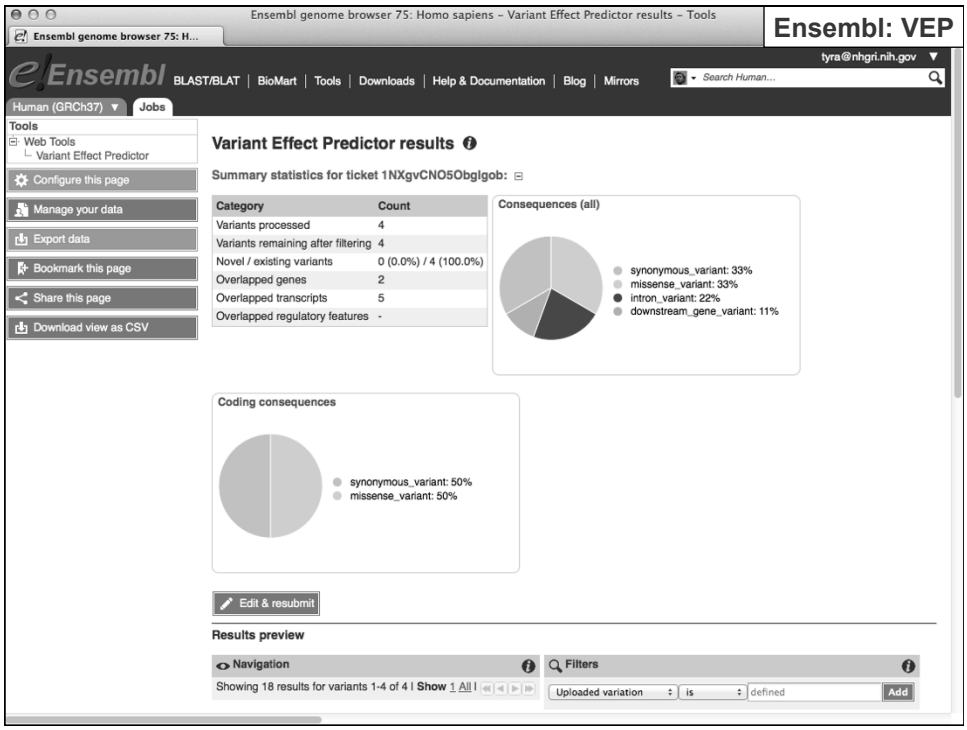
Variant Effect Predictor
New VEP job:
Input
Species:
Human (Homo sapiens)
Assembly: GRCh37
Name for this data (optional):
ADAM2 final
Input file format (details):
Variant identifiers
Either paste data:
rs35935433
rs144646998
rs145143599
rs34417912
Or upload file:
Browse...
No file selected.
Or provide file URL:
Or select previously uploaded file:
-- Select file --
Transcript database to use:
Ensembl transcripts
RefSeq and other transcripts
Output options
Identifiers and frequency data
Additional identifiers for genes, transcripts and variants; frequency data
Extra options
e.g. SIFT, PolyPhen and regulatory data


Ensembl: VEP
Ensembl genome browser 75: Homo sapiens - Variant Effect Predictor results - Tools
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Manage your data
Export data
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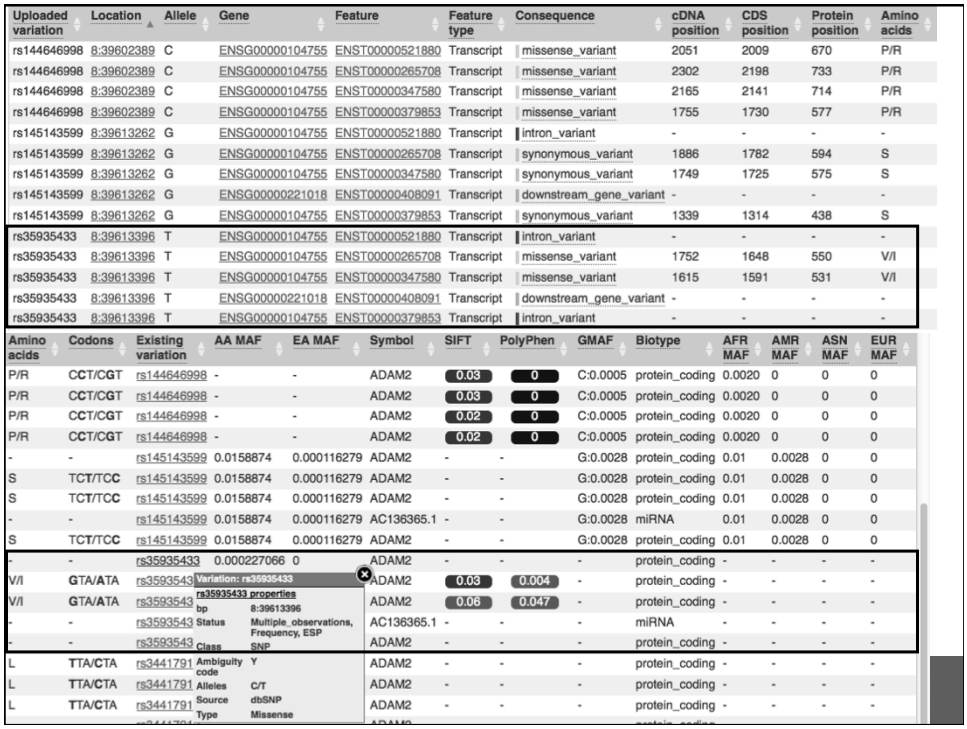
Download view as CSV
Variant Effect Predictor results
Summary statistics for ticket 1NXgvCNO5Obglgob:
Category
Count
Variants processed
4
Variants remaining after filtering
4
Novel / existing variants
0 (0.0%) / 4 (100.0%)
Overlapped genes
2
Overlapped transcripts
5
Overlapped regulatory features
-
Consequences (all)
synonymous_variant: 33%
missense_variant: 33%
intron_variant: 22%
downstream_gene_variant: 11%
Coding consequences
synonymous_variant: 50%
missense_variant: 50%
Edit & resubmit
Results preview
Navigation
Showing 18 results for variants 1-4 of 4 | Show 1 All |
Filters
Uploaded variation
is
defined
Add



Uploaded variation	Location	Allele	Gene	Feature	Feature type	Consequence	cDNA position	CDS position	Protein position	Amino acids
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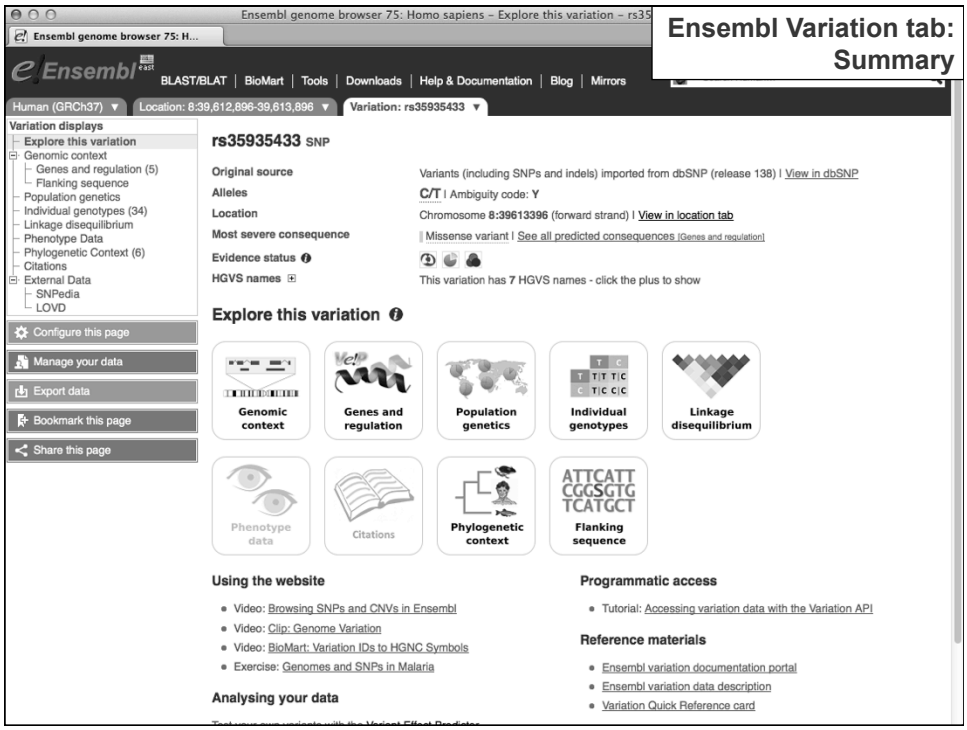
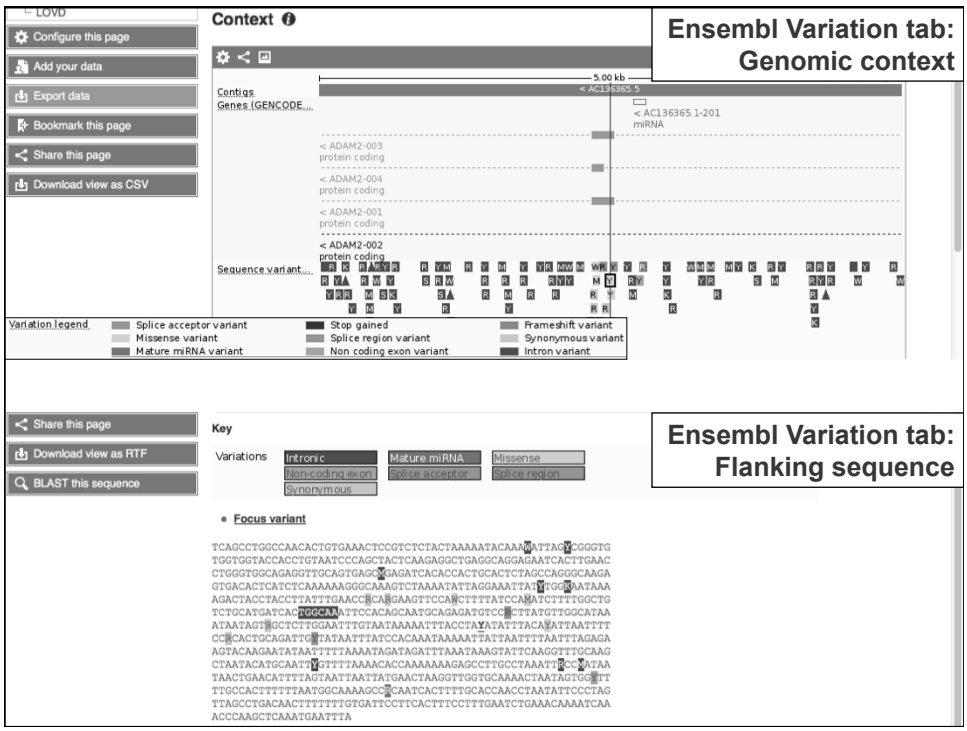
rs144646998	8:39602389	C	ENSG00000104755	ENST00000265708	Transcript	missense_variant	2302	2198	733	P/R
rs144646998	8:39602389	C	ENSG00000104755	ENST00000347580	Transcript	missense_variant	2165	2141	714	P/R
rs144646998	8:39602389	C	ENSG00000104755	ENST00000379853	Transcript	missense_variant	1755	1730	577	P/R
rs145143599	8:39613262	G	ENSG00000104755	ENST00000521880	Transcript	intron_variant	-	-	-	-
rs145143599	8:39613262	G	ENSG00000104755	ENST00000265708	Transcript	synonymous_variant	1886	1782	594	S
rs145143599	8:39613262	G	ENSG00000104755	ENST00000347580	Transcript	synonymous_variant	1749	1725	575	S
rs145143599	8:39613262	G	ENSG00000221018	ENST00000408091	Transcript	downstream_gene_variant	-	-	-	-
rs145143599	8:39613262	G	ENSG00000104755	ENST00000379853	Transcript	synonymous_variant	1339	1314	438	S
rs35935433	8:39613396	T	ENSG00000104755	ENST00000521880	Transcript	intron_variant	-	-	-	-
rs35935433	8:39613396	T	ENSG00000104755	ENST00000265708	Transcript	missense_variant	1752	1648	550	V/I
rs35935433	8:39613396	T	ENSG00000104755	ENST00000347580	Transcript	missense_variant	1615	1591	531	V/I
rs35935433	8:39613396	T	ENSG00000221018	ENST00000408091	Transcript	downstream_gene_variant	-	-	-	-
rs35935433	8:39613396	T	ENSG00000104755	ENST00000379853	Transcript	intron_variant	-	-	-	-

Amino acids	Codons	Existing variation	AA MAF	EA MAF	Symbol	SIFT	PolyPhen	GMAF	Biotype	AFR MAF	AMR MAF	ASN MAF	EUR MAF
P/R	CCT/CGT	rs144646998	-	-	ADAM2	0.03	0	C:0.0005	protein_coding	0.0020	0	0	0
P/R	CCT/CGT	rs144646998	-	-	ADAM2	0.03	0	C:0.0005	protein_coding	0.0020	0	0	0
P/R	CCT/CGT	rs144646998	-	-	ADAM2	0.02	0	C:0.0005	protein_coding	0.0020	0	0	0
P/R	CCT/CGT	rs144646998	-	-	ADAM2	0.02	0	C:0.0005	protein_coding	0.0020	0	0	0
-	-	rs145143599	0.0158874	0.000116279	ADAM2	-	-	G:0.0028	protein_coding	0.01	0.0028	0	0
S	TCT/TCC	rs145143599	0.0158874	0.000116279	ADAM2	-	-	G:0.0028	protein_coding	0.01	0.0028	0	0
S	TCT/TCC	rs145143599	0.0158874	0.000116279	ADAM2	-	-	G:0.0028	protein_coding	0.01	0.0028	0	0
-	-	rs145143599	0.0158874	0.000116279	AC136365.1	-	-	G:0.0028	miRNA	0.01	0.0028	0	0
S	TCT/TCC	rs145143599	0.0158874	0.000116279	ADAM2	-	-	G:0.0028	protein_coding	0.01	0.0028	0	0
-	-	rs35935433	0.000227066	0	ADAM2	-	-	-	protein_coding	-	-	-	-
V/I	GTA/ATA	rs3593543	Variation: rs35935433		ADAM2	0.03	0.004	-	protein_coding	-	-	-	-
V/I	GTA/ATA	rs3593543	rs35935433 properties		ADAM2	0.06	0.047	-	protein_coding	-	-	-	-
-	-	rs3593543	bp 8:39613396		AC136365.1	-	-	-	miRNA	-	-	-	-
-	-	rs3593543	Status Multiple_observations, Frequency, ESP		ADAM2	-	-	-	protein_coding	-	-	-	-
-	-	rs3593543	Class SNP		ADAM2	-	-	-	protein_coding	-	-	-	-
L	TTA/CTA	rs3441791	Ambiguity code Y		ADAM2	-	-	-	protein_coding	-	-	-	-
L	TTA/CTA	rs3441791	Alleles C/T		ADAM2	-	-	-	protein_coding	-	-	-	-
L	TTA/CTA	rs3441791	Source dbSNP		ADAM2	-	-	-	protein_coding	-	-	-	-
			Type Missense										



Ensembl Variation tab: Summary



Ensembl

Location tab

http://www.ensembl.org

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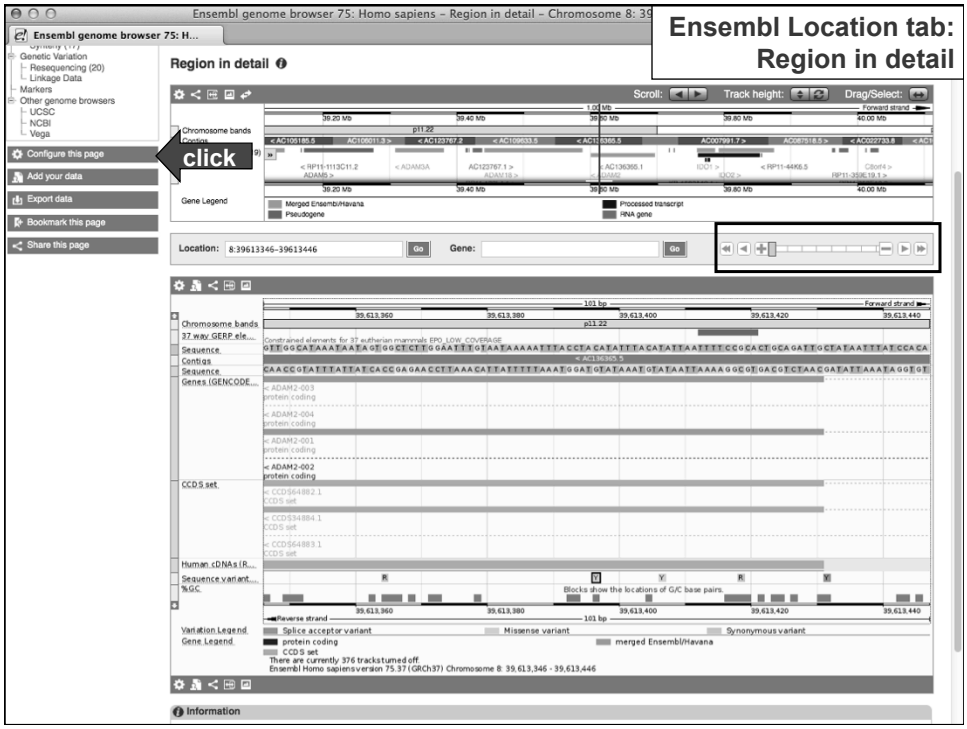
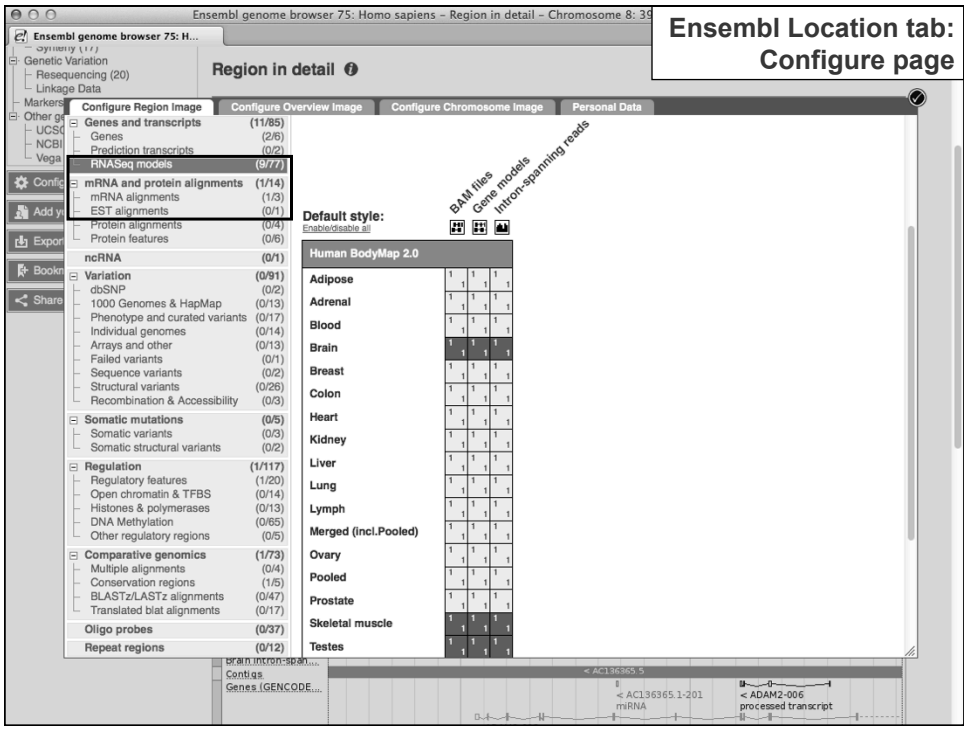
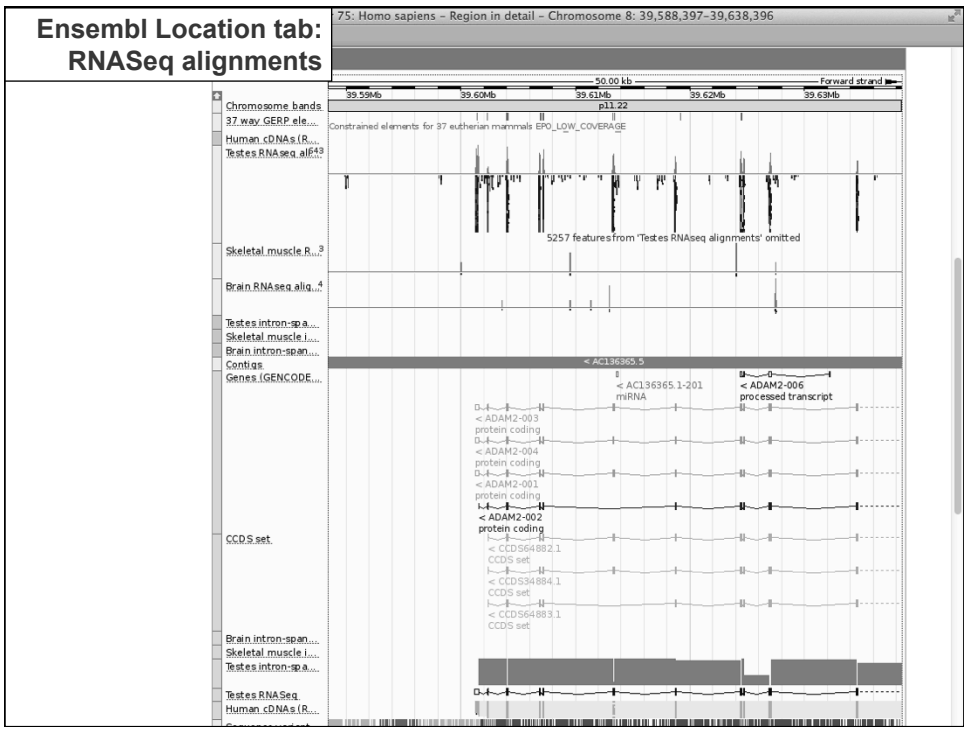

Ensembl Location tab:
Region in detail
Ensembl genome browser 75: Homo sapiens – Region in detail – Chromosome 8: 39
Region in detail
click
Configure this page
Add your data
Export data
Bookmark this page
Share this page
Location: 8:39613346-39613446
Gene:
Go
Scroll:
Track height:
Drag/Select:
Chromosome bands
Gene Legend
Merged Ensembl/Havana
Pseudogene
Processed transcript
RNA gene
Sequence
Contigs
Genes (GENCODE...
CCDS set
Human cDNAs (R...
Sequence variant...
%GC
Blocks show the locations of G/C base pairs.
Variation Legend
Splice acceptor variant
Missense variant
Synonymous variant
Gene Legend
protein coding
merged Ensembl/Havana
CCDS set
There are currently 376 tracks turned off.
Ensembl Homo sapiens version 75.37 (GRCh37) Chromosome 8: 39,613,346 - 39,613,446
Information


Ensembl Location tab:
Configure page
Region in detail
Configure Region Image
Configure Overview Image
Configure Chromosome Image
Personal Data
Genes and transcripts (11/85)
Genes (2/6)
Prediction transcripts (0/2)
RNASeq models (0/77)
mRNA and protein alignments (1/14)
mRNA alignments (1/3)
EST alignments (0/1)
Protein alignments (0/4)
Protein features (0/6)
ncRNA (0/1)
Variation (0/91)
dbSNP (0/2)
1000 Genomes & HapMap (0/13)
Phenotype and curated variants (0/17)
Individual genomes (0/14)
Arrays and other (0/13)
Failed variants (0/1)
Sequence variants (0/2)
Structural variants (0/26)
Recombination & Accessibility (0/3)
Somatic mutations (0/5)
Somatic variants (0/3)
Somatic structural variants (0/2)
Regulation (1/117)
Regulatory features (1/20)
Open chromatin & TFBS (0/14)
Histones & polymerases (0/13)
DNA Methylation (0/65)
Other regulatory regions (0/5)
Comparative genomics (1/73)
Multiple alignments (0/4)
Conservation regions (1/5)
BLASTz/LASTz alignments (0/47)
Translated blat alignments (0/17)
Oligo probes (0/37)
Repeat regions (0/12)
BAM files
Gene models
Intron-spanning reads
Default style:
Enable/disable all
Human BodyMap 2.0
Adipose
Adrenal
Blood
Brain
Breast
Colon
Heart
Kidney
Liver
Lung
Lymph
Merged (incl.Pooled)
Ovary
Pooled
Prostate
Skeletal muscle
Testes
Contigs
Genes (GENCODE...
< AC136365.5
< AC136365.1-201
miRNA
< ADAM2-006
processed transcript

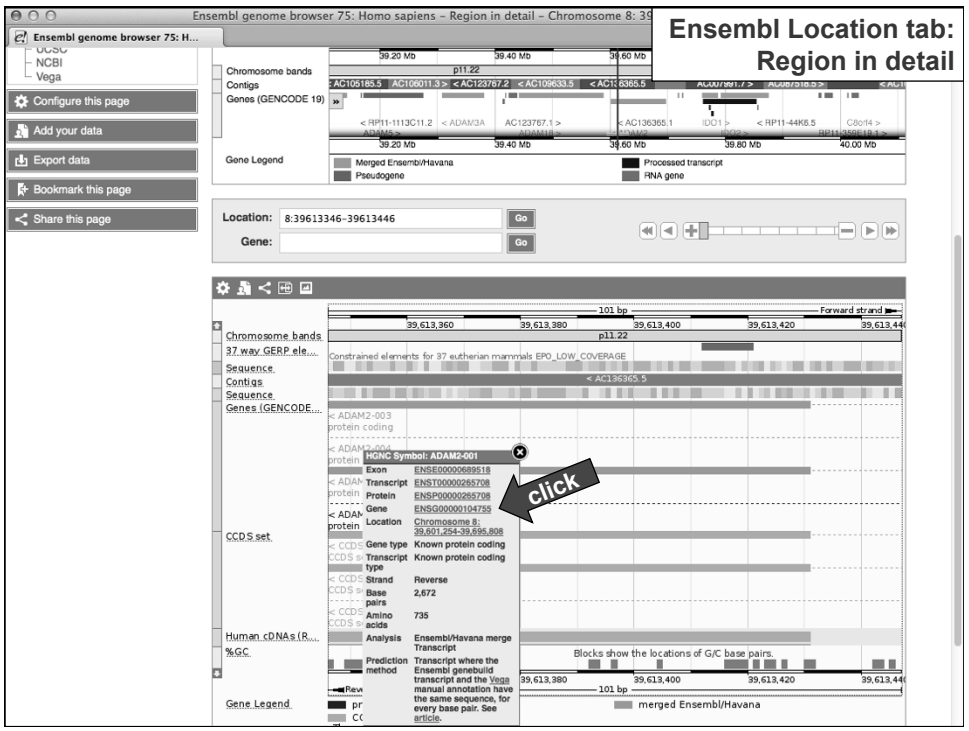
Ensembl Location tab: RNASeq alignments

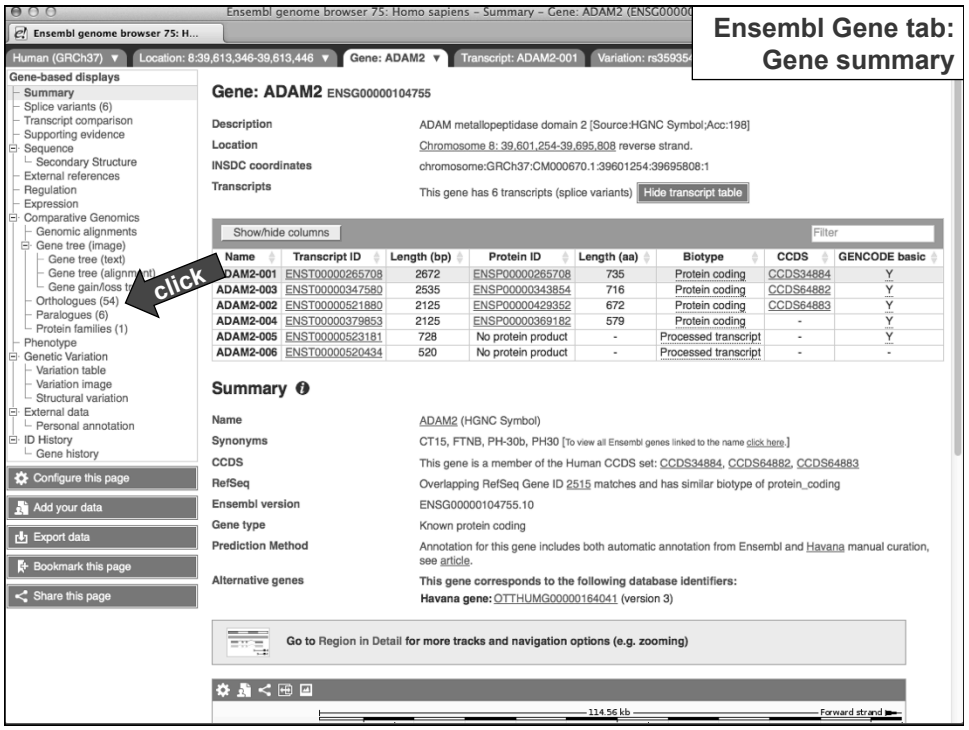


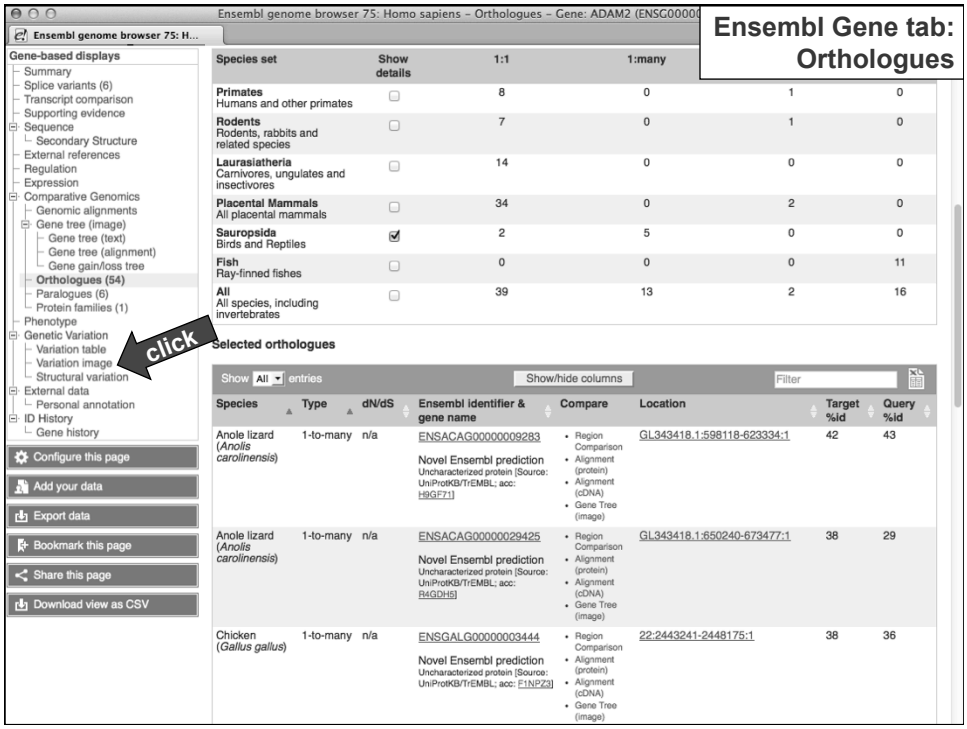
Ensembl

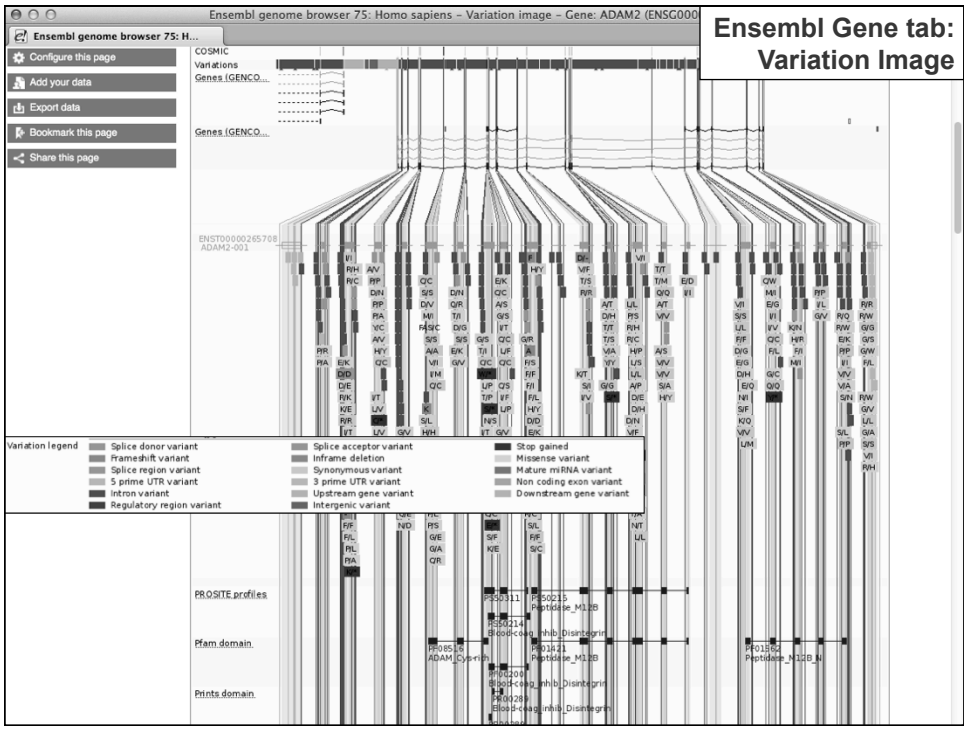
Gene tab

http://www.ensembl.org


Ensembl Location tab:
Region in detail
Ensembl genome browser 75: Homo sapiens – Region in detail – Chromosome 8: 39
Ensembl genome browser 75: H...
NCBI
Vega
Configure this page
Add your data
Export data
Bookmark this page
Share this page
Chromosome bands
Contigs
Genes (GENCODE 19)
Gene Legend
Merged Ensembl/Havana
Pseudogene
Processed transcript
RNA gene
Location: 8:39613346-39613446
Gene:
Go
Chromosome bands
37 way GERP ele...
Sequence
Contigs
Sequence
Genes (GENCODE...
Constrained elements for 37 eutherian mammals EPO_LOW_COVERAGE
HGNC Symbol: ADAM2-001
Exon
Transcript
Protein
Gene
Location
Gene type
Transcript type
Strand
Base pairs
Amino acids
Analysis
Prediction method
click
CCDS set
Human cDNAs (R...
%GC
Blocks show the locations of G/C base pairs.
merged Ensembl/Havana


Ensembl Gene tab:
Gene summary
Ensembl genome browser 75: Homo sapiens – Summary – Gene: ADAM2
Human (GRCh37)
Location: 8:39,613,346-39,613,448
Gene: ADAM2
Transcript: ADAM2-001
Gene-based displays
Summary
Splice variants (6)
Transcript comparison
Supporting evidence
Sequence
Secondary Structure
External references
Regulation
Expression
Comparative Genomics
Genomic alignments
Gene tree (image)
Gene tree (text)
Orthologues (54)
Paralogues (6)
Protein families (1)
Phenotype
Genetic Variation
Variation table
Variation image
Structural variation
External data
Personal annotation
ID History
Gene history
click
Gene: ADAM2 ENSG00000104755
Description
ADAM metallopeptidase domain 2 [Source:HGNC Symbol;Acc:198]
Location
Chromosome 8: 39,601,254-39,695,808 reverse strand.
INSDC coordinates
chromosome:GRCh37:CM000670.1:39601254:39695808:1
Transcripts
This gene has 6 transcripts (splice variants)
Hide transcript table
Show/hide columns
Filter
Name | Transcript ID | Length (bp) | Protein ID | Length (aa) | Biotype | CCDS | GENCODE basic
ADAM2-001 | ENST00000265708 | 2672 | ENSP00000265708 | 735 | Protein coding | CCDS34884 | Y
ADAM2-003 | ENST00000347580 | 2535 | ENSP00000343854 | 716 | Protein coding | CCDS64882 | Y
ADAM2-002 | ENST00000521880 | 2125 | ENSP00000429352 | 672 | Protein coding | CCDS64883 | Y
ADAM2-004 | ENST00000379853 | 2125 | ENSP00000369182 | 579 | Protein coding | - | Y
ADAM2-005 | ENST00000523181 | 728 | No protein product | - | Processed transcript | - | Y
ADAM2-006 | ENST00000520434 | 520 | No protein product | - | Processed transcript | - | -
Summary
Name
ADAM2 (HGNC Symbol)
Synonyms
CT15, FTNB, PH-30b, PH30 [To view all Ensembl genes linked to the name click here.]
CCDS
This gene is a member of the Human CCDS set: CCDS34884, CCDS64882, CCDS64883
RefSeq
Overlapping RefSeq Gene ID 2515 matches and has similar biotype of protein_coding
Ensembl version
ENSG00000104755.10
Gene type
Known protein coding
Prediction Method
Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see article.
Alternative genes
This gene corresponds to the following database identifiers:
Havana gene: OTTHUMG00000164041 (version 3)
Go to Region in Detail for more tracks and navigation options (e.g. zooming)
Configure this page
Add your data
Export data
Bookmark this page
Share this page


Ensembl Gene tab:
Orthologues
Ensembl genome browser 75: Homo sapiens – Orthologues – Gene: ADAM2 (ENSG00000
Gene-based displays
Summary
Splice variants (6)
Transcript comparison
Supporting evidence
Sequence
Secondary Structure
External references
Regulation
Expression
Comparative Genomics
Genomic alignments
Gene tree (image)
Gene tree (text)
Gene tree (alignment)
Gene gain/loss tree
Orthologues (54)
Paralogues (6)
Protein families (1)
Phenotype
Genetic Variation
Variation table
Variation image
Structural variation
External data
Personal annotation
ID History
Gene history
click
Configure this page
Add your data
Export data
Bookmark this page
Share this page
Download view as CSV
Species set
Show details
1:1
1:many
Primates
Humans and other primates
Rodents
Rodents, rabbits and related species
Laurasiatheria
Carnivores, ungulates and insectivores
Placental Mammals
All placental mammals
Sauropsida
Birds and Reptiles
Fish
Ray-finned fishes
All
All species, including invertebrates
Selected orthologues
Show All entries
Show/hide columns
Filter
Species
Type
dN/dS
Ensembl identifier & gene name
Compare
Location
Target %id
Query %id
Anole lizard (Anolis carolinensis)
1-to-many
n/a
ENSACAG00000009283
Novel Ensembl prediction
Uncharacterized protein [Source: UniProtKB/TrEMBL; acc: H9GF71]
Region Comparison
Alignment (protein)
Alignment (cDNA)
Gene Tree (image)
GL343418.1:598118-623334:1
42
43
Anole lizard (Anolis carolinensis)
1-to-many
n/a
ENSACAG00000029425
Novel Ensembl prediction
Uncharacterized protein [Source: UniProtKB/TrEMBL; acc: R4GDH5]
GL343418.1:650240-673477:1
38
29
Chicken (Gallus gallus)
1-to-many
n/a
ENSGALG00000003444
Novel Ensembl prediction
Uncharacterized protein [Source: UniProtKB/TrEMBL; acc: F1NPZ3]
22:2443241-2448175:1
38
36


Ensembl Gene tab:
Variation Image
Ensembl genome browser 75: Homo sapiens – Variation image – Gene: ADAM2 (ENSG000
Configure this page
Add your data
Export data
Bookmark this page
Share this page
COSMIC Variations
Genes (GENCO...
Genes (GENCO...
ENST00000265708
ADAM2-001
Variation legend
Splice donor variant
Frameshift variant
Splice region variant
5 prime UTR variant
Intron variant
Regulatory region variant
Splice acceptor variant
Inframe deletion
Synonymous variant
3 prime UTR variant
Upstream gene variant
Intergenic variant
Stop gained
Missense variant
Mature miRNA variant
Non coding exon variant
Downstream gene variant
PROSITE profiles
Pfam domain
Prints domain

Ensembl

Transcript tab

http://www.ensembl.org

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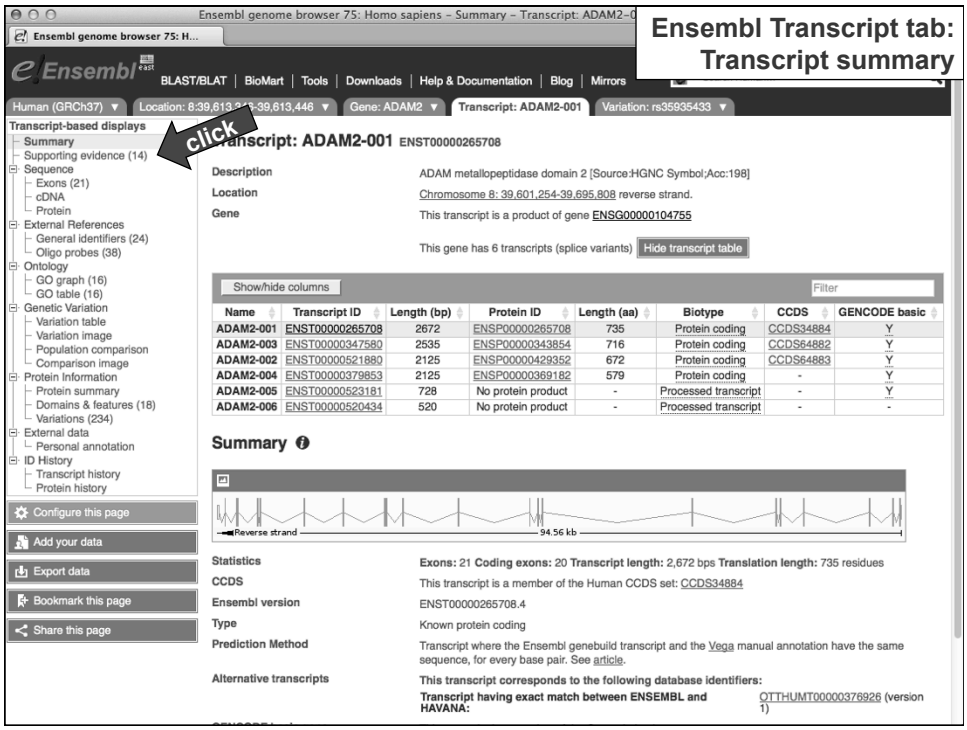
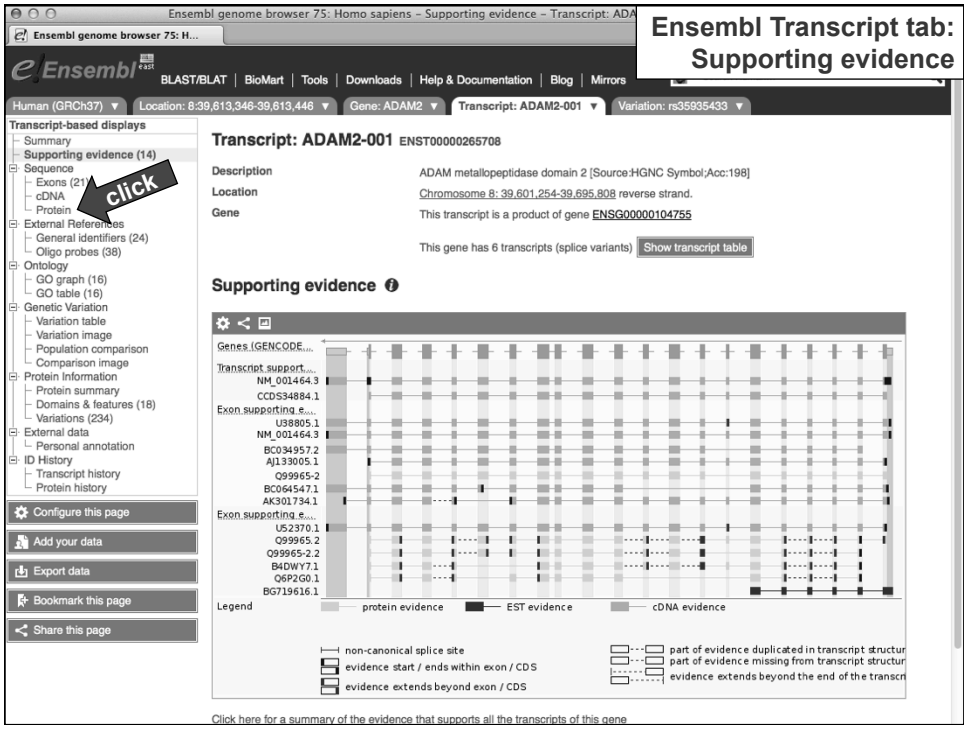
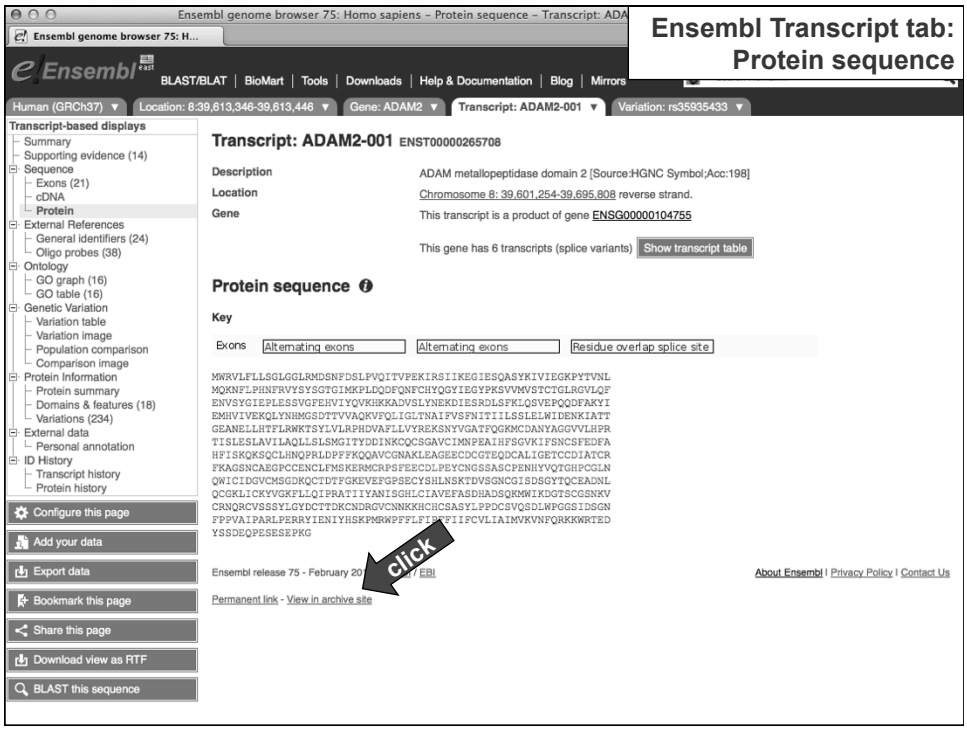
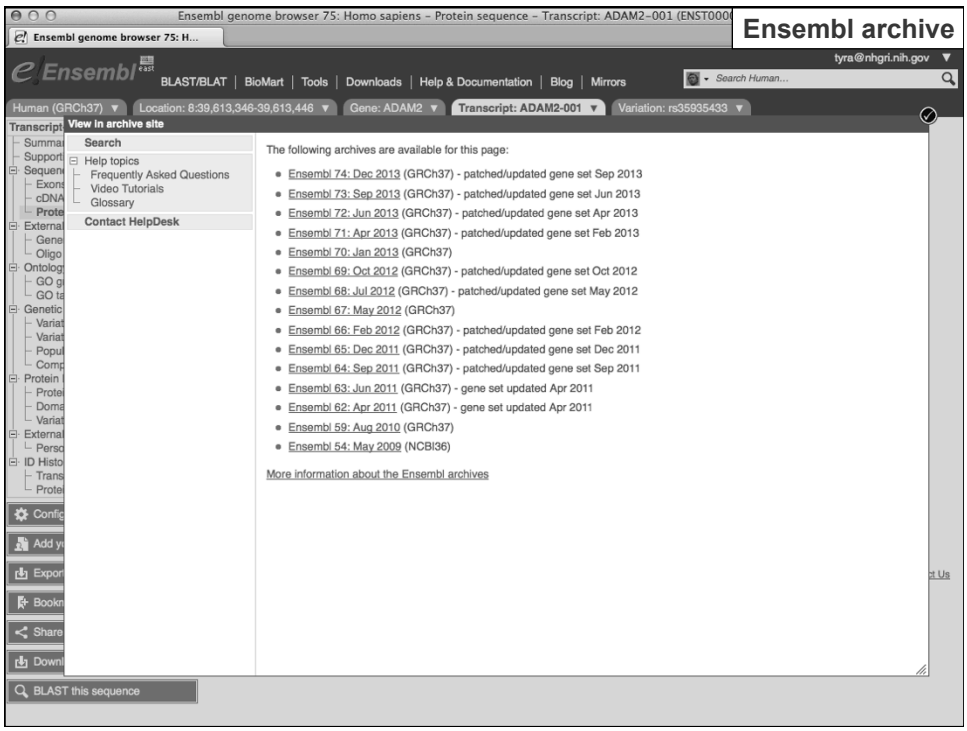



Ensembl Transcript tab:
Supporting evidence
Ensembl genome browser 75: Homo sapiens – Supporting evidence – Transcript: ADA
BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog | Mirrors
Human (GRCh37) | Location: 8:39,613,346-39,613,446 | Gene: ADAM2 | Transcript: ADAM2-001 | Variation: rs35935433
Transcript-based displays
Summary
Supporting evidence (14)
Sequence
Exons (21)
cDNA
Protein
External References
General identifiers (24)
Oligo probes (38)
Ontology
GO graph (16)
GO table (16)
Genetic Variation
Variation table
Variation image
Population comparison
Comparison image
Protein Information
Protein summary
Domains & features (18)
Variations (234)
External data
Personal annotation
ID History
Transcript history
Protein history
click
Configure this page
Add your data
Export data
Bookmark this page
Share this page
Transcript: ADAM2-001 ENST00000265708
Description
ADAM metallopeptidase domain 2 [Source:HGNC Symbol;Acc:198]
Location
Chromosome 8: 39,601,254-39,695,808 reverse strand.
Gene
This transcript is a product of gene ENSG00000104755
This gene has 6 transcripts (splice variants)
Show transcript table
Supporting evidence
Genes (GENCODE...
Transcript support...
NM_001464.3
CCDS34884.1
Exon supporting e...
U38805.1
NM_001464.3
BC034957.2
AJ133005.1
Q99965-2
BC064547.1
AK301734.1
Exon supporting e...
U52370.1
Q99965.2
Q99965-2.2
B4DWY7.1
Q6P2G0.1
BG719616.1
Legend
protein evidence
EST evidence
cDNA evidence
non-canonical splice site
evidence start / ends within exon / CDS
evidence extends beyond exon / CDS
part of evidence duplicated in transcript structur
part of evidence missing from transcript structur
evidence extends beyond the end of the transcri
Click here for a summary of the evidence that supports all the transcripts of this gene

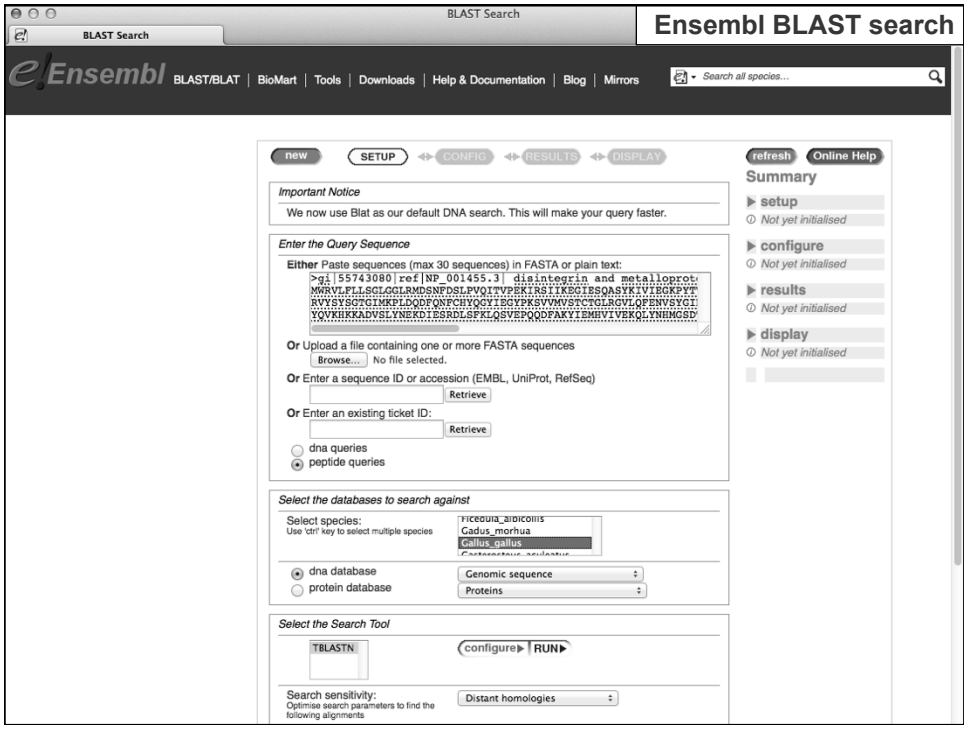

Ensembl Transcript tab:
Protein sequence
Ensembl genome browser 75: Homo sapiens – Protein sequence – Transcript: ADA
BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog | Mirrors
Human (GRCh37) | Location: 8:39,613,346-39,613,446 | Gene: ADAM2 | Transcript: ADAM2-001 | Variation: rs35935433
Transcript-based displays
Summary
Supporting evidence (14)
Sequence
Exons (21)
cDNA
Protein
External References
General identifiers (24)
Oligo probes (38)
Ontology
GO graph (16)
GO table (16)
Genetic Variation
Variation table
Variation image
Population comparison
Comparison image
Protein Information
Protein summary
Domains & features (18)
Variations (234)
External data
Personal annotation
ID History
Transcript history
Protein history
Configure this page
Add your data
Export data
Bookmark this page
Share this page
Download view as RTF
BLAST this sequence
Transcript: ADAM2-001 ENST00000265708
Description
ADAM metallopeptidase domain 2 [Source:HGNC Symbol;Acc:198]
Location
Chromosome 8: 39,601,254-39,695,808 reverse strand.
Gene
This transcript is a product of gene ENSG00000104755
This gene has 6 transcripts (splice variants)
Show transcript table
Protein sequence
Key
Exons
Alternating exons
Alternating exons
Residue overlap splice site
MWRVLFLLSGLGGLRMDSNFDSLPVQITVPEKIRSIIKEGIESQASYKIVIEGKPYTVNL
MQKNFLPHNFRVYSYSGTGIMKPLDQDFQNFCHYQGYIEGYPKSVVMVSTCTGLRGVLQF
ENVSYGIEPLESSVGFEHVIYQVKHKKADVSLYNEKDIESRDLSFKLQSVEPQQDFAKYI
EMHVIVEKQLYNHMGSDTTVVAQKVFQLIGLTNAIFVSFNITIILSSLELWIDENKIATT
GEANELLHTFLRWKTSYLVLRPHDVAFLLVYREKSNYVGATFQGKMCDANYAGGVVLHPR
TISLESLAVILAQLLSLSMGITYDDINKCQCSGAVCIMNPEAIHFSGVKIFSNCSFEDFA
HFISKQKSQCLHNQPRLDPFFKQQAVCGNAKLEAGEECDCGTEQDCALIGETCCDIATCR
FKAGSNCAEGPCCENCLFMSKERMCRPSFEECDLPEYCNGSSASCPENHYVQTGHPCGLN
QWICIDGVCMSGDKQCTDTFGKEVEFGPSECYSHLNSKTDVSGNCGISDSGYTQCEADNL
QCGKLICKYVGKFLLQIPRATIIYANISGHLCIAVEFASDHADSQKMWIKDGTSCGSNKV
CRNQRCVSSSYLGYDCTTDKCNDRGVCNNKKHCHCSASYLPPDCSVQSDLWPGGSIDSGN
FPPVAIPARLPERRYIENIYHSKPMRWPFFLFIWFIIPCVLIAIMVKVNFQRKKWRTED
YSSDEQPESESEPKG
click
Ensembl release 75 - February 20
EBI
About Ensembl | Privacy Policy | Contact Us
Permanent link - View in archive site

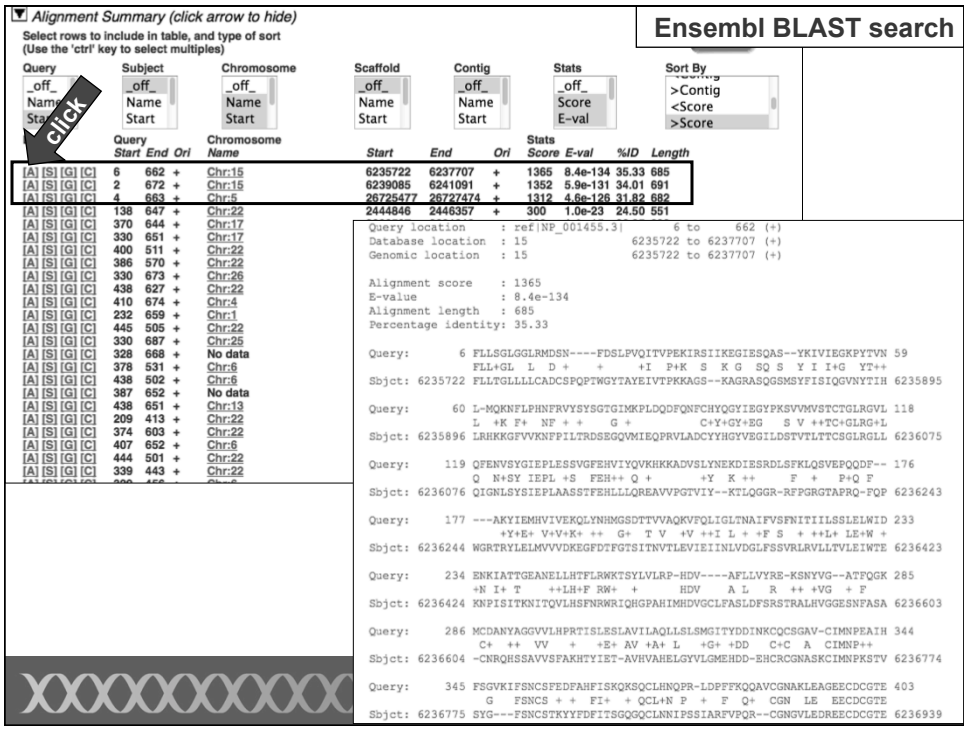


Ensembl

Find a chicken homolog of a human protein

http://www.ensembl.org


Ensembl BLAST search
BLAST Search
Ensembl
BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog | Mirrors
Search all species...
new
SETUP
CONFIG
RESULTS
DISPLAY
refresh
Online Help
Summary
setup
Not yet initialised
configure
Not yet initialised
results
Not yet initialised
display
Not yet initialised
Important Notice
We now use 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:
>gi|55743080|ref|NP_001455.3| disintegrin and metalloprot
Or Upload a file containing one or more FASTA sequences
Browse...
No file selected.
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
Gadus_morhua
Gallus_gallus
dna database
protein database
Genomic sequence
Proteins
Select the Search Tool
TBLASTN
configure
RUN
Search sensitivity:
Optimise search parameters to find the following alignments
Distant homologies

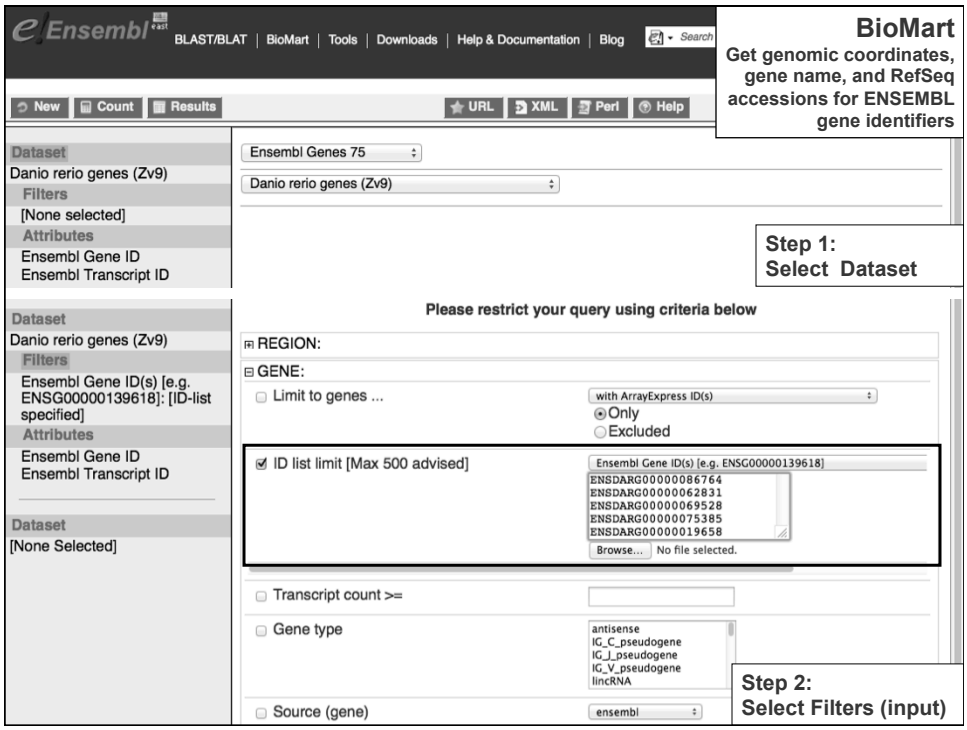

Ensembl BLAST search
Alignment Summary (click arrow to hide)
Select rows to include in table, and type of sort
(Use the 'ctrl' key to select multiples)
Query
Subject
Chromosome
Scaffold
Contig
Stats
Sort By
>Contig
<Score
>Score
click
Query Start End Ori
Chromosome Name
Start End Ori
Stats Score E-val %ID Length
[A] [S] [G] [C] 6 662 + Chr:15 6235722 6237707 + 1365 8.4e-134 35.33 685
[A] [S] [G] [C] 2 672 + Chr:15 6239085 6241091 + 1352 5.9e-131 34.01 691
[A] [S] [G] [C] 4 663 + Chr:5 26725477 26727474 + 1312 4.6e-126 31.82 682
[A] [S] [G] [C] 138 647 + Chr:22 2444846 2446357 + 300 1.0e-23 24.50 551
Query location : ref|NP_001455.3| 6 to 662 (+)
Database location : 15 6235722 to 6237707 (+)
Genomic location : 15 6235722 to 6237707 (+)
Alignment score : 1365
E-value : 8.4e-134
Alignment length : 685
Percentage identity: 35.33
Query: 6 FLLSGLGGLRMDSN----FDSLPVQITVPEKIRSIIKEGIESQAS--YKIVIEGKPYTVN 59
Sbjct: 6235722 FLLTGLLLLCADCSPQPTWGYTAYEIVTPKKAGS--KAGRASQGSMSYFISIQGVNYTIH 6235895
Query: 60 L-MQKNFLPHNFRVYSYSGTGIMKPLDQDFQNFCHYQGYIEGYPKSVVMVSTCTGLRGVL 118
Sbjct: 6235896 LRHKKGFVVKNFPILTRDSEGQVMIEQPRVLADCYYHGYVEGILDSTVTLTTCSGLRGLL 6236075
Query: 119 QFENVSYGIEPLESSVGFEHVIYQVKHKKADVSLYNEKDIESRDLSFKLQSVEPQQDF-- 176
Sbjct: 6236076 QIGNLSYSIEPLAASSTFEHLLLQREAVVPGTVIY--KTLQGGR-RFPGRGTAPRQ-FQP 6236243
Query: 177 ---AKYIEMHVIVEKQLYNHMGSDTTVVAQKVFQLIGLTNAIFVSFNITIILSSLELWID 233
Sbjct: 6236244 WGRTRYLELMVVVDKEGFDTFGTSITNVTLEVIEIINLVDGLFSSVRLRVLLTVLEIWTE 6236423
Query: 234 ENKIATTGEANELLHTFLRWKTSYLVLRP-HDV----AFLLVYRE-KSNYVG--ATFQGK 285
Sbjct: 6236424 KNPISITKNITQVLHSFNRWRIQHGPAHIMHDVGCLFASLDFSRSTRALHVGGESNFASA 6236603
Query: 286 MCDANYAGGVVLHPRTISLESLAVILAQLLSLSMGITYDDINKCQCSGAV-CIMNPEAIH 344
Sbjct: 6236604 -CNRQHSSAVVSFAKHTYIET-AVHVAHELGYVLGMEHDD-EHCRCGNASKCIMNPKSTV 6236774
Query: 345 FSGVKIFSNCSFEDFAHFISKQKSQCLHNQPR-LDPFFKQQAVCGNAKLEAGEECDCGTE 403
Sbjct: 6236775 SYG---FSNCSTKYYFDFITSGQGQCLNNIPSSIARFVPQR--CGNGVLEDREECDCGTE 6236939

BioMart

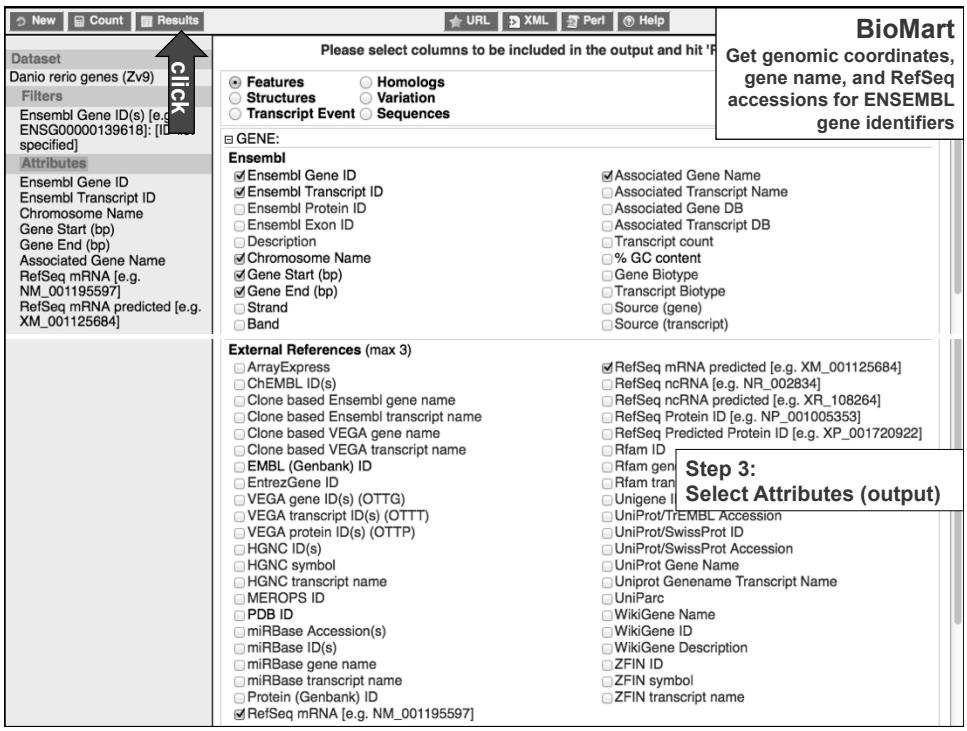
Cross-reference data from different sources

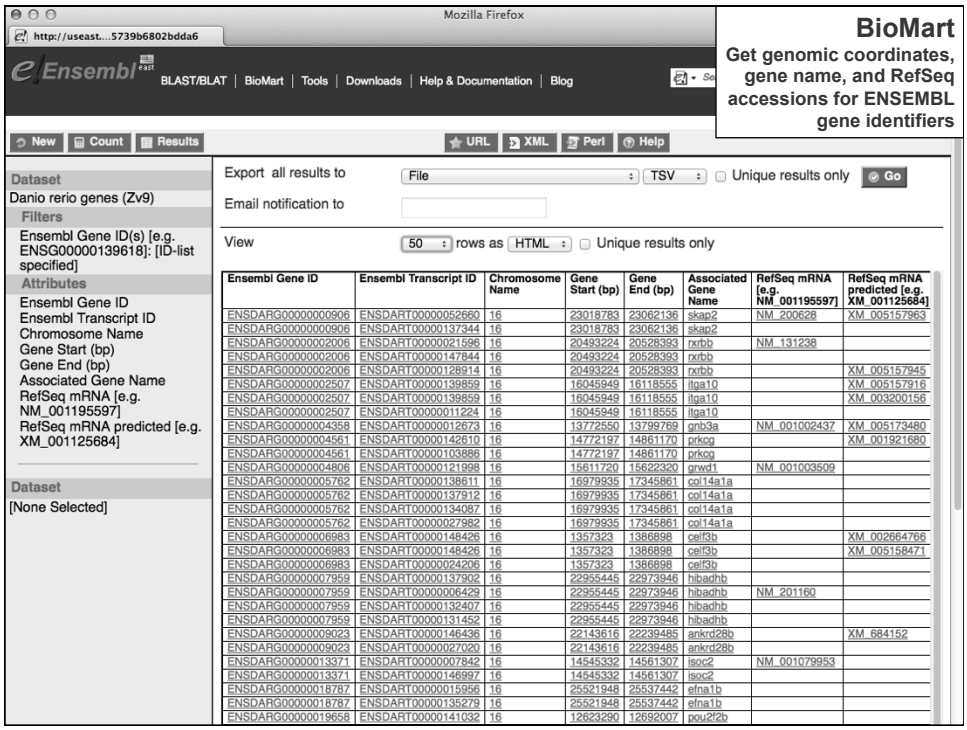
http://www.ensembl.org/biomart

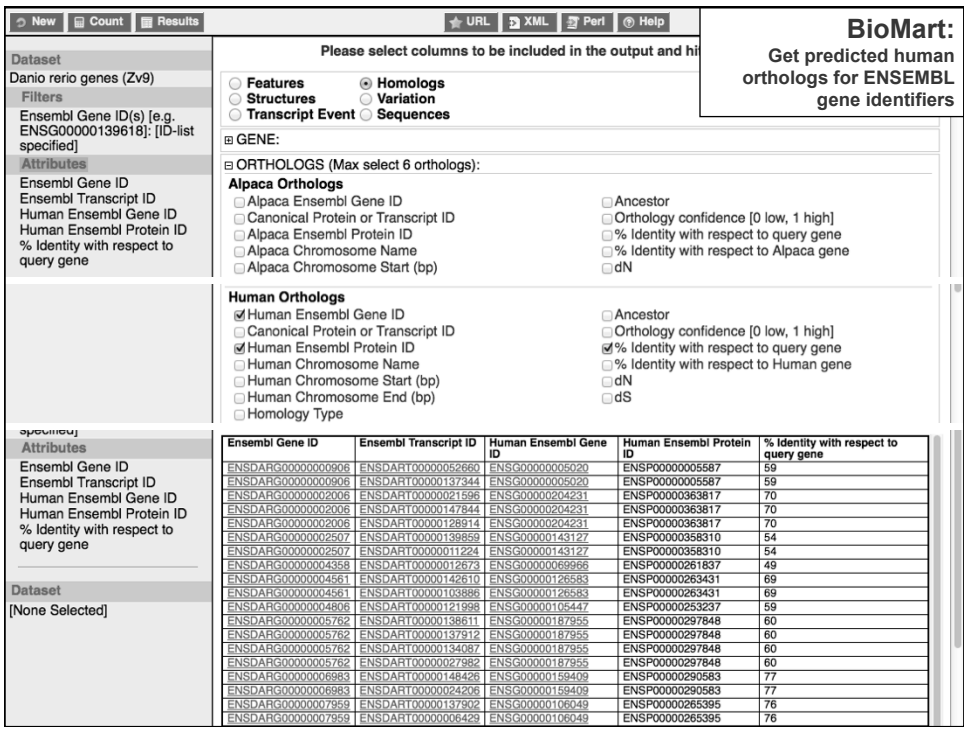
NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research



BioMart
Get genomic coordinates, gene name, and RefSeq accessions for ENSEMBL gene identifiers


New
Count
Results
URL
XML
Perl
Help
click
Dataset
Danio rerio genes (Zv9)
Filters
Ensembl Gene ID(s) [e.g. ENSG00000139618]: [ID-list specified]
Attributes
Ensembl Gene ID
Ensembl Transcript ID
Chromosome Name
Gene Start (bp)
Gene End (bp)
Associated Gene Name
RefSeq mRNA [e.g. NM_001195597]
RefSeq mRNA predicted [e.g. XM_001125684]
Please select columns to be included in the output and hit '
Features
Homologs
Structures
Variation
Transcript Event
Sequences
GENE:
Ensembl
Ensembl Gene ID
Ensembl Transcript ID
Ensembl Protein ID
Ensembl Exon ID
Description
Chromosome Name
Gene Start (bp)
Gene End (bp)
Strand
Band
Associated Gene Name
Associated Transcript Name
Associated Gene DB
Associated Transcript DB
Transcript count
% GC content
Gene Biotype
Transcript Biotype
Source (gene)
Source (transcript)
External References (max 3)
ArrayExpress
ChEMBL ID(s)
Clone based Ensembl gene name
Clone based Ensembl transcript name
Clone based VEGA gene name
Clone based VEGA transcript name
EMBL (Genbank) ID
EntrezGene ID
VEGA gene ID(s) (OTTG)
VEGA transcript ID(s) (OTTT)
VEGA protein ID(s) (OTTP)
HGNC ID(s)
HGNC symbol
HGNC transcript name
MEROPS ID
PDB ID
miRBase Accession(s)
miRBase ID(s)
miRBase gene name
miRBase transcript name
Protein (Genbank) ID
RefSeq mRNA [e.g. NM_001195597]
RefSeq mRNA predicted [e.g. XM_001125684]
RefSeq ncRNA [e.g. NR_002834]
RefSeq ncRNA predicted [e.g. XR_108264]
RefSeq Protein ID [e.g. NP_001005353]
RefSeq Predicted Protein ID [e.g. XP_001720922]
Rfam ID
UniProt/TrEMBL Accession
UniProt/SwissProt ID
UniProt/SwissProt Accession
UniProt Gene Name
Uniprot Genename Transcript Name
UniParc
WikiGene Name
WikiGene ID
WikiGene Description
ZFIN ID
ZFIN symbol
ZFIN transcript name
BioMart
Get genomic coordinates, gene name, and RefSeq accessions for ENSEMBL gene identifiers
Step 3:
Select Attributes (output)


Mozilla Firefox
http://useast....5739b6802bdda6
Ensembl
BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog
New
Count
Results
URL
XML
Perl
Help
Dataset
Danio rerio genes (Zv9)
Filters
Ensembl Gene ID(s) [e.g. ENSG00000139618]: [ID-list specified]
Attributes
Ensembl Gene ID
Ensembl Transcript ID
Chromosome Name
Gene Start (bp)
Gene End (bp)
Associated Gene Name
RefSeq mRNA [e.g. NM_001195597]
RefSeq mRNA predicted [e.g. XM_001125684]
Dataset
[None Selected]
Export all results to
File
TSV
Unique results only
Go
Email notification to
View
50
rows as
HTML
Unique results only
Ensembl Gene ID | Ensembl Transcript ID | Chromosome Name | Gene Start (bp) | Gene End (bp) | Associated Gene Name | RefSeq mRNA [e.g. NM_001195597] | RefSeq mRNA predicted [e.g. XM_001125684]
ENSDARG00000000906 | ENSDART00000052660 | 16 | 23018783 | 23062136 | skap2 | NM_200628 | XM_005157963
ENSDARG00000000906 | ENSDART00000137344 | 16 | 23018783 | 23062136 | skap2
ENSDARG00000002006 | ENSDART00000021596 | 16 | 20493224 | 20528393 | rxrbb | NM_131238
ENSDARG00000002006 | ENSDART00000147844 | 16 | 20493224 | 20528393 | rxrbb
ENSDARG00000002006 | ENSDART00000128914 | 16 | 20493224 | 20528393 | rxrbb | | XM_005157945
ENSDARG00000002507 | ENSDART00000139859 | 16 | 16045949 | 16118555 | itga10 | | XM_005157916
ENSDARG00000002507 | ENSDART00000139859 | 16 | 16045949 | 16118555 | itga10 | | XM_003200156
ENSDARG00000002507 | ENSDART00000011224 | 16 | 16045949 | 16118555 | itga10
ENSDARG00000004358 | ENSDART00000012673 | 16 | 13772550 | 13799769 | gnb3a | NM_001002437 | XM_005173480
ENSDARG00000004561 | ENSDART00000142610 | 16 | 14772197 | 14861170 | prkcg | | XM_001921680
ENSDARG00000004561 | ENSDART00000103886 | 16 | 14772197 | 14861170 | prkcg
ENSDARG00000004806 | ENSDART00000121998 | 16 | 15611720 | 15622320 | grwd1 | NM_001003509
ENSDARG00000005762 | ENSDART00000138611 | 16 | 16979935 | 17345861 | col14a1a
ENSDARG00000005762 | ENSDART00000137912 | 16 | 16979935 | 17345861 | col14a1a
ENSDARG00000005762 | ENSDART00000134087 | 16 | 16979935 | 17345861 | col14a1a
ENSDARG00000005762 | ENSDART00000027982 | 16 | 16979935 | 17345861 | col14a1a
ENSDARG00000006983 | ENSDART00000148426 | 16 | 1357323 | 1386898 | celf3b | | XM_002664766
ENSDARG00000006983 | ENSDART00000148426 | 16 | 1357323 | 1386898 | celf3b | | XM_005158471
ENSDARG00000006983 | ENSDART00000024206 | 16 | 1357323 | 1386898 | celf3b
ENSDARG00000007959 | ENSDART00000137902 | 16 | 22955445 | 22973946 | hibadhb
ENSDARG00000007959 | ENSDART00000006429 | 16 | 22955445 | 22973946 | hibadhb | NM_201160
ENSDARG00000007959 | ENSDART00000132407 | 16 | 22955445 | 22973946 | hibadhb
ENSDARG00000007959 | ENSDART00000131452 | 16 | 22955445 | 22973946 | hibadhb
ENSDARG00000009023 | ENSDART00000146436 | 16 | 22143616 | 22239485 | ankrd28b | | XM_684152
ENSDARG00000009023 | ENSDART00000027020 | 16 | 22143616 | 22239485 | ankrd28b
ENSDARG00000013371 | ENSDART00000007842 | 16 | 14545332 | 14561307 | lsoc2 | NM_001079953
ENSDARG00000013371 | ENSDART00000146997 | 16 | 14545332 | 14561307 | lsoc2
ENSDARG00000018787 | ENSDART00000015956 | 16 | 25521948 | 25537442 | efna1b
ENSDARG00000018787 | ENSDART00000135279 | 16 | 25521948 | 25537442 | efna1b
ENSDARG00000019658 | ENSDART00000141032 | 16 | 12623290 | 12692007 | pou2f2b
BioMart
Get genomic coordinates, gene name, and RefSeq accessions for ENSEMBL gene identifiers

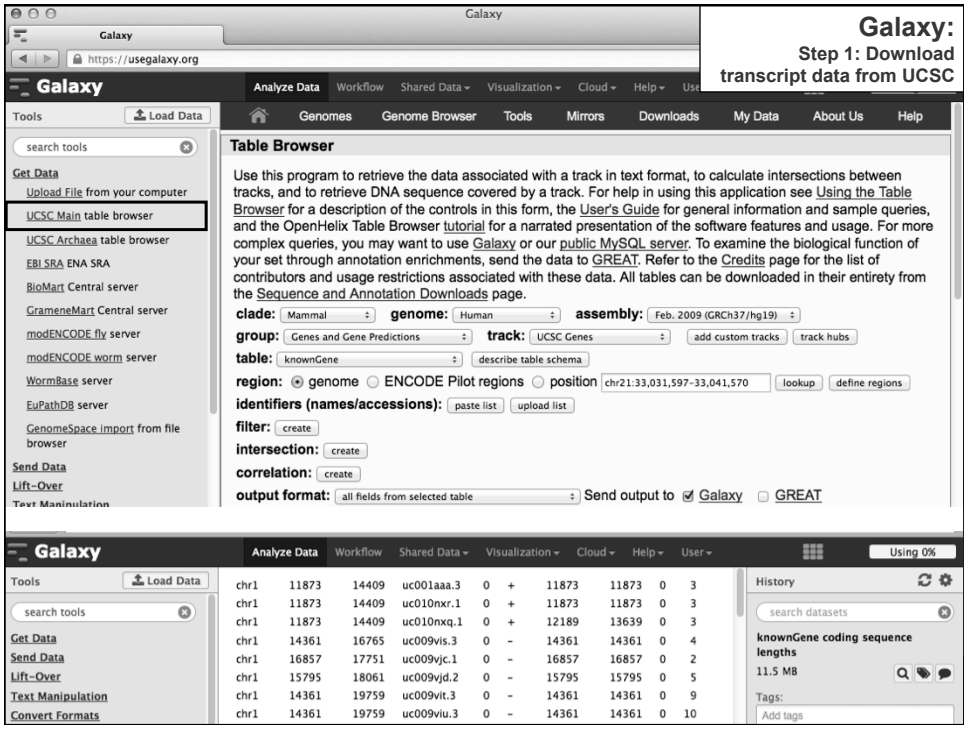


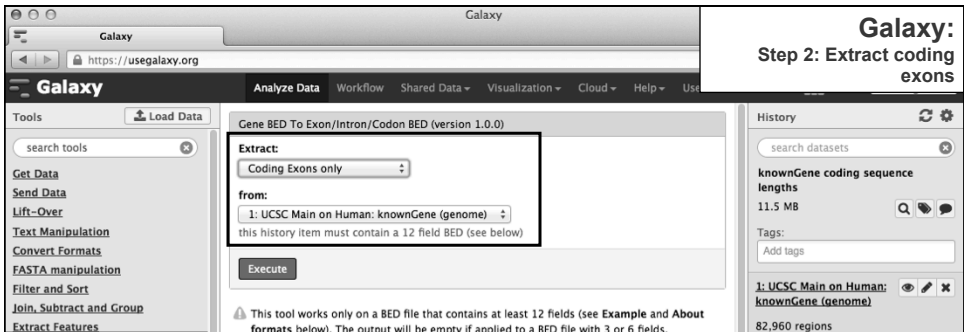
BioMart:
Get predicted human orthologs for ENSEMBL gene identifiers

Galaxy

knownGene coding sequence lengths

https://usegalaxy.org/


Galaxy:
Step 1: Download transcript data from UCSC
Table Browser
Use this program to retrieve the data associated with a track in text format, to calculate intersections between tracks, and to retrieve DNA sequence covered by a track. For help in using this application see Using the Table Browser for a description of the controls in this form, the User's Guide for general information and sample queries, and the OpenHelix Table Browser tutorial for a narrated presentation of the software features and usage. For more complex queries, you may want to use Galaxy or our public MySQL server. To examine the biological function of your set through annotation enrichments, send the data to GREAT. Refer to the Credits page for the list of contributors and usage restrictions associated with these data. All tables can be downloaded in their entirety from the Sequence and Annotation Downloads page.
clade: Mammal genome: Human assembly: Feb. 2009 (GRCh37/hg19)
group: Genes and Gene Predictions track: UCSC Genes
table: knownGene
region: genome ENCODE Pilot regions position chr21:33,031,597-33,041,570
identifiers (names/accessions):
filter:
intersection:
correlation:
output format: all fields from selected table Send output to Galaxy GREAT

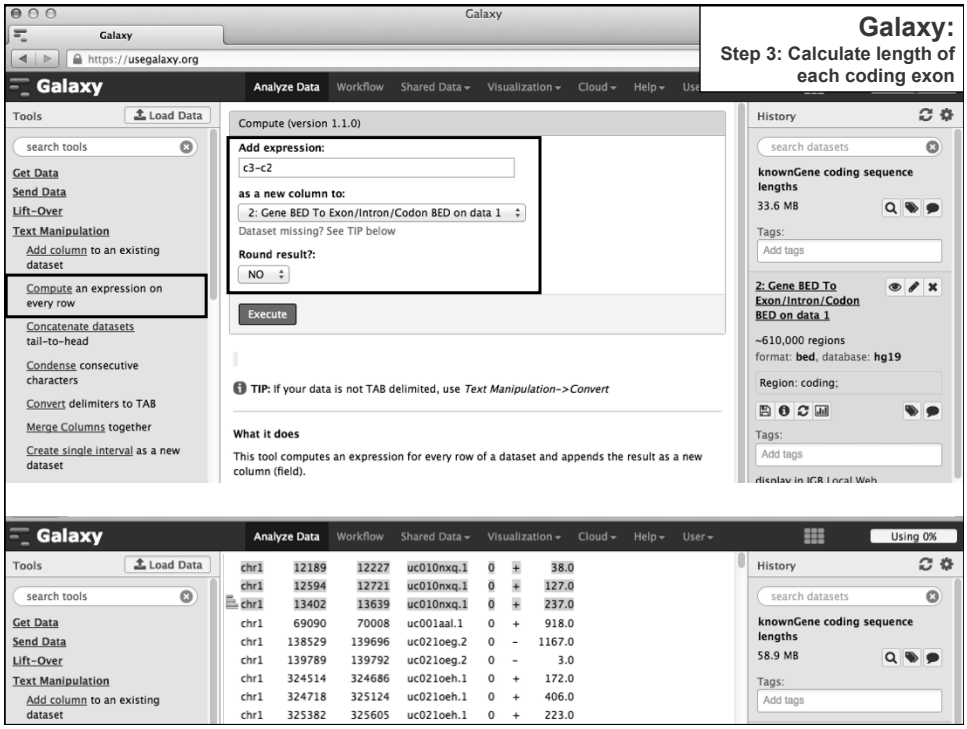

knownGene coding sequence lengths
11.5 MB

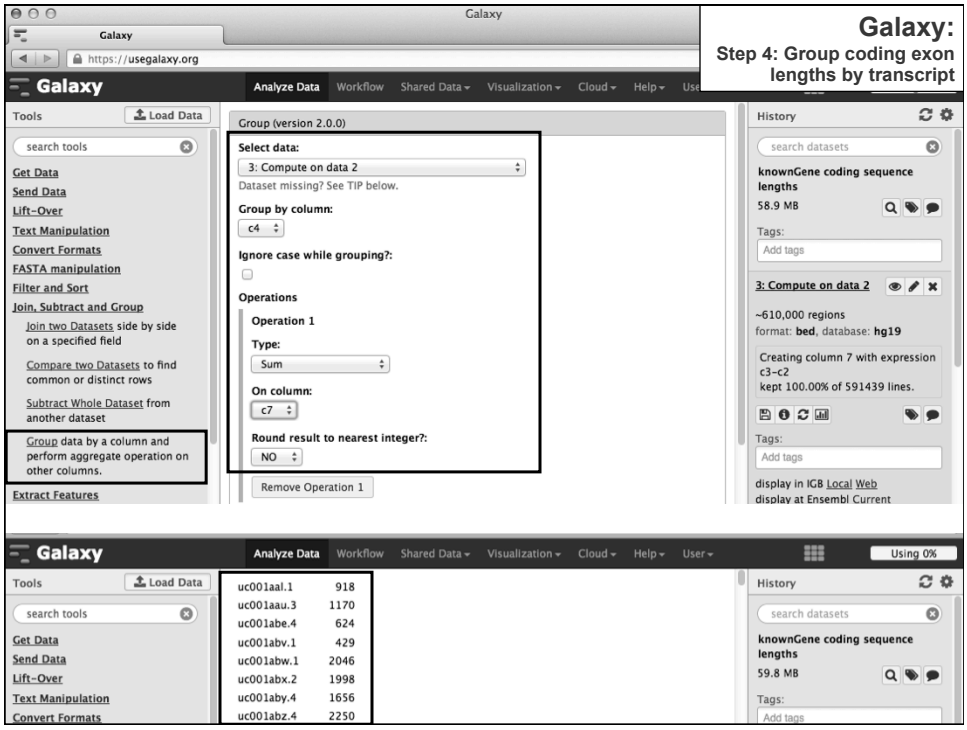
chr1 11873 14409 uc001aaa.3 0 + 11873 11873 0 3
chr1 11873 14409 uc010nxr.1 0 + 11873 11873 0 3
chr1 11873 14409 uc010nxq.1 0 + 12189 13639 0 3
chr1 14361 16765 uc009vis.3 0 - 14361 14361 0 4
chr1 16857 17751 uc009vjc.1 0 - 16857 16857 0 2
chr1 15795 18061 uc009vjd.2 0 - 15795 15795 0 5
chr1 14361 19759 uc009vit.3 0 - 14361 14361 0 9
chr1 14361 19759 uc009viu.3 0 - 14361 14361 0 10

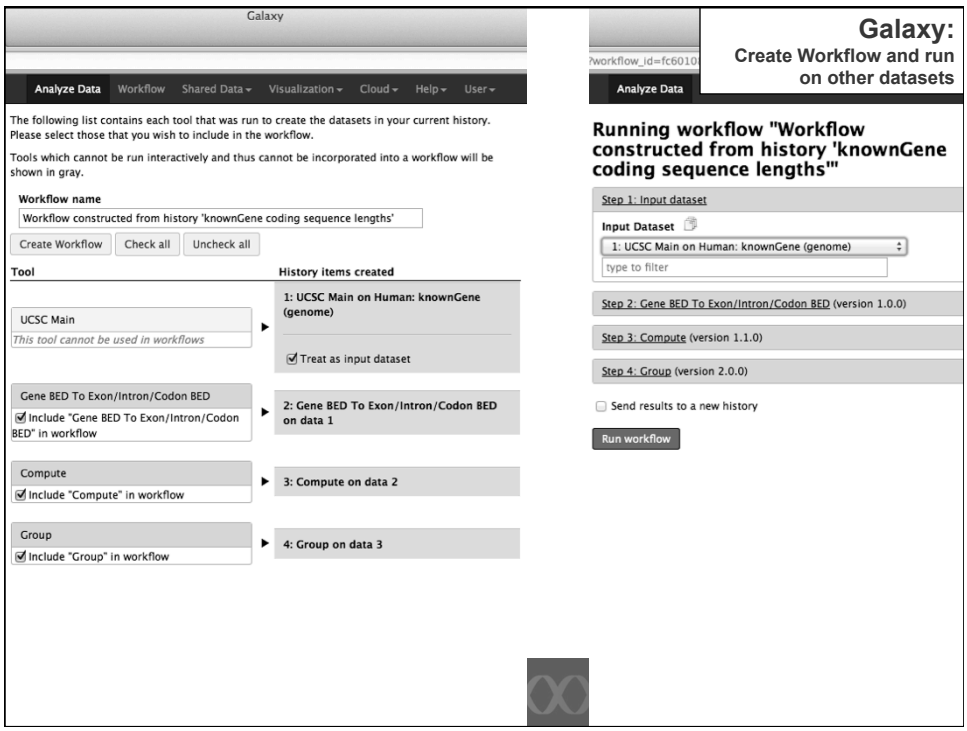
Galaxy:
Step 2: Extract coding exons
Gene BED To Exon/Intron/Codon BED (version 1.0.0)
Extract:
Coding Exons only
from:
1: UCSC Main on Human: knownGene (genome)
this history item must contain a 12 field BED (see below)
Execute
This tool works only on a BED file that contains at least 12 fields (see Example and About formats below). The output will be empty if applied to a BED file with 3 or 6 fields.
What it does
BED format can be used to represent a single gene in just one line, which contains the information about exons, coding sequence location (CDS), and positions of untranslated regions (UTRs). This tool unpacks this information by converting a single line describing a gene into a collection of lines representing individual exons, introns, UTRs, etc.
1: UCSC Main on Human: knownGene (genome)
82,960 regions
format: bed, database: hg19
2: Gene BED To Exon/Intron/Codon BED on data 1

knownGene coding sequence lengths
33.6 MB

chr1 12189 12227 uc010nxq.1 0 +
chr1 12594 12721 uc010nxq.1 0 +
chr1 13402 13639 uc010nxq.1 0 +
chr1 69090 70008 uc001aal.1 0 +
chr1 138529 139696 uc021oeg.2 0 -
chr1 139789 139792 uc021oeg.2 0 -
chr1 324514 324686 uc021oeh.1 0 +
chr1 324718 325124 uc021oeh.1 0 +
chr1 325382 325605 uc021oeh.1 0 +
chr1 324342 324345 uc009vjk.2 0 +
chr1 324438 325605 uc009vjk.2 0 +
chr1 327745 328213 uc021oei.1 0 +


Galaxy:
Step 3: Calculate length of each coding exon
Galaxy
https://usegalaxy.org
Analyze Data
Workflow
Shared Data
Visualization
Cloud
Help
Tools
Load Data
search tools
Get Data
Send Data
Lift-Over
Text Manipulation
Add column to an existing dataset
Compute an expression on every row
Concatenate datasets tail-to-head
Condense consecutive characters
Convert delimiters to TAB
Merge Columns together
Create single interval as a new dataset
Compute (version 1.1.0)
Add expression:
c3-c2
as a new column to:
2: Gene BED To Exon/Intron/Codon BED on data 1
Dataset missing? See TIP below
Round result?:
NO
Execute
TIP: If your data is not TAB delimited, use Text Manipulation->Convert
What it does
This tool computes an expression for every row of a dataset and appends the result as a new column (field).
History
search datasets
knownGene coding sequence lengths
33.6 MB
Tags:
Add tags
2: Gene BED To Exon/Intron/Codon BED on data 1
~610,000 regions
format: bed, database: hg19
Region: coding;
Using 0%
chr1 12189 12227 uc010nxq.1 0 + 38.0
chr1 12594 12721 uc010nxq.1 0 + 127.0
chr1 13402 13639 uc010nxq.1 0 + 237.0
chr1 69090 70008 uc001aal.1 0 + 918.0
chr1 138529 139696 uc021oeg.2 0 - 1167.0
chr1 139789 139792 uc021oeg.2 0 - 3.0
chr1 324514 324686 uc021oeh.1 0 + 172.0
chr1 324718 325124 uc021oeh.1 0 + 406.0
chr1 325382 325605 uc021oeh.1 0 + 223.0
58.9 MB


Galaxy:
Step 4: Group coding exon lengths by transcript
Galaxy
https://usegalaxy.org
Analyze Data
Workflow
Shared Data
Visualization
Cloud
Help
Tools
Load Data
search tools
Get Data
Send Data
Lift-Over
Text Manipulation
Convert Formats
FASTA manipulation
Filter and Sort
Join, Subtract and Group
Join two Datasets side by side on a specified field
Compare two Datasets to find common or distinct rows
Subtract Whole Dataset from another dataset
Group data by a column and perform aggregate operation on other columns.
Extract Features
Group (version 2.0.0)
Select data:
3: Compute on data 2
Dataset missing? See TIP below.
Group by column:
c4
Ignore case while grouping?:
Operations
Operation 1
Type:
Sum
On column:
c7
Round result to nearest integer?:
NO
Remove Operation 1
History
search datasets
knownGene coding sequence lengths
58.9 MB
Tags:
Add tags
3: Compute on data 2
~610,000 regions
format: bed, database: hg19
Creating column 7 with expression c3-c2
kept 100.00% of 591439 lines.
display in IGB Local Web
display at Ensembl Current
Using 0%
uc001aal.1 918
uc001aau.3 1170
uc001abe.4 624
uc001abv.1 429
uc001abw.1 2046
uc001abx.2 1998
uc001aby.4 1656
uc001abz.4 2250
59.8 MB



Galaxy NGS tools

- Quality control and manipulation
  - FASTQC
- Mapping
  - Bowtie
- Peak Calling
  - MACS
- RNA-seq
  - TopHat
  - CuffLinks

NGS TOOLBOX BETA
Phenotype Association
NGS: QC and manipulation
NGS: Mapping
NGS: SAM Tools
NGS: GATK Tools (beta)
NGS: Peak Calling
NGS: RNA-seq
NGS: Picard (beta)
NGS: Variant Analysis
snpEff
BEDTools
EMBOSS

Additional resources

- **UCSC Genome Browser User Guide**
  http://genome.ucsc.edu/goldenPath/help/
- **Ensembl Tutorials and Worked Examples**
  http://www.ensembl.org/info/website/tutorials/
- **Galaxy Support**
  https://wiki.galaxyproject.org/Support/

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 Calif
Santa Cruz, California

ABSTRACT

The University of California Santa Cruz (UCSC) Genome Browse
based tool for quickly displaying a requested portion of a genor
companied by a series of aligned annotation "tracks." The annotati
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 r
one window, facilitating biological analysis and interpretation. The
derlying the Genome Browser tracks can be viewed, downloaded, ar
another Web-based application, the UCSC Table Browser. Users

Using Galaxy to Perform Large-Scale Interactive Data Analyses

UNIT 10.5

Jennifer Hillman-Jackson,[1] Dave Clements,[2] Daniel Blankenberg,[1] James Taylor,[2] Anton Nekrutenko,[1] and Galaxy Team[1,2]

[1]Penn State University, University Park, Pennsylvania
[2]Emory University, Atlanta, Georgia

ABSTRACT

Innovations in biomedical research technologies continue to provide experimental biol-
ogists with novel and increasingly large genomic and high-throughput data resources to
be analyzed. As creating and obtaining data has become easier, the key decision faced
by many researchers is a practical one: where and how should an analysis be performed?
up and use is riddled with complexities outside of
e authors believe that Galaxy provides a powerful
on and analysis in an intuitive Web application,
nformatics tools previously only available to com-
based environments. We will demonstrate through
cols how Galaxy specifically brings together (1)
sources, for example, UCSC's Eukaryote and
om tools (wrapped Unix functions, format stan-
tions), and 3rd-party analysis tools. *Curr. Protoc.*
by John Wiley & Sons, Inc.

mics • genomic alignments •

Using the Ensembl Genome Server to Browse Genomic Sequence Data

UNIT 1.15

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

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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