


NATIONAL HUMAN GENOME RESEARCH INSTITUTE *Division of Intramural Research*




Current Topics in Genome Analysis 2012

Week 3: Genome Browsers

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U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES | NATIONAL INSTITUTES OF HEALTH | genome.gov/DIR



Current Topics in Genome Analysis 2012

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*No Relevant Financial Relationships with
Commercial Interests*



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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

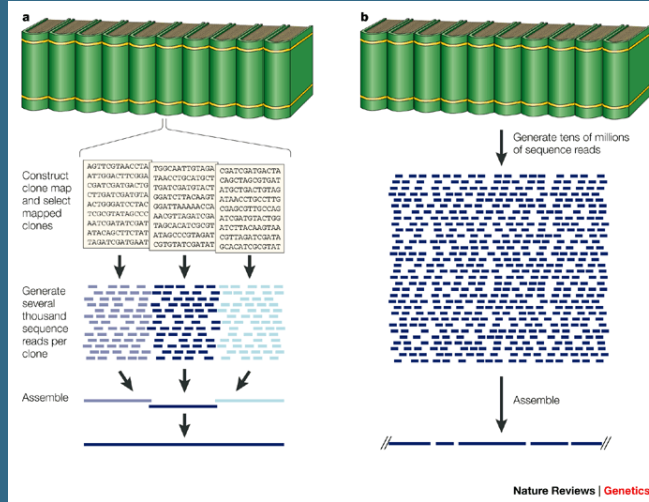
- Same starting material for all genome browsers:
genomic sequence
- Annotations calculated independently by each genome browser
 - Genes
 - RefSeq mRNAs (non-redundant)
 - GenBank mRNAs (redundant)
 - ESTs
 - Gene predictions
 - SNPs
 - Non-coding functional elements



Overview of genome sequencing strategies

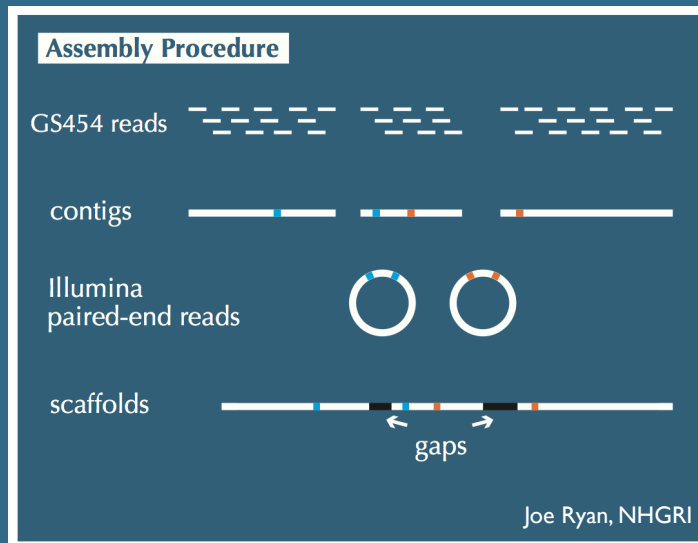
Clone-by-clone shotgun sequencing

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., Shiraiishi,M., Fukami,K., Tanabe,A., Ikeda-Matsuo,Y.,
            Naito,Y. and Sasaki,Y.
TITLE      MARCKS regulates lamellipodia formation induced by IGF-I via
            association with PIP2 and beta-actin at membrane microdomains
JOURNAL    J. Cell. Physiol. 220 (3), 748-755 (2009)
PUBMED    19475567

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

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

            Publication Note: This RefSeq record includes a subset of the
            publications that are available for this gene. Please see the
            Entrez Gene record to access additional publications.
            COMPLETENESS: complete on the 3' end.

CDS       85..1212
            /gene="ACTB"
            /gene_synonym="PS1TP5BP1"
            /note="beta cytoskeletal actin; PS1TP5-binding protein 1;
            actin, cytoplasmic 1"
            /codon_start=1
            /product="beta actin"
            /protein_id="NP_001092.1"
            /db_xref="GI:4501885"
            /db_xref="CCDS:CCDS341.1"
            /db_xref="GeneID:610"
            /db_xref="HGNC:132"
            /db_xref="HPRD:00032"
            /db_xref="MIM:102630"
            /translation="MDDIALVVDNGSGMCKAGFAGDDAPRAVFPISIVCRPRHQGVN
            VGMGQKDSVVGDEAQSCKGILITLKYPIEIGLVTNDDMEKIWHFTFYNELRVAPPEHP
            VLLTEAPLNPKANREKMTQIMFTFNTFAMYVAIQAVLSLYASGRTGIVMDSGDGVT
            HTVPIYEGVALPHAIRLDLAGRDLDYLMKILTERGYSFTTAREIVRDIKELICY
            VALDFQEMTAAGSSLEKSYELPQGVITIGNERFCPEALPQPSFLGMSCCGHE
            TTFNSIMKCDVDIRKDLVANTVLSGGTTPYGIADRMQKEITLALPSTMKIKIITAPE
            RRYSVNIGGSILASLSTFQQMMLSKQEYDESQPSIVHRKCF"

ORIGIN     1 accgcgagca cgcgctcgc ccgcgagca cagagctcg cctttgccg tcgcgcgcc
            61 gtccacacc gccgcagct caccatgat gatgatatg ccgcgctcg ctcgcacac
            121 gctcccgca tgtgcaagg cggtctcgc ggcgacgat cccccggc cgtctccccc
    
```

Beta actin mRNA RefSeq

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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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The image shows a blue background with the UCSC logo in yellow. Below the logo, the text 'View a region in the genome by querying with a gene symbol' is displayed in white. The URL 'http://genome.ucsc.edu' is shown in yellow. At the bottom, there is a white DNA double helix graphic and the text 'NATIONAL HUMAN GENOME RESEARCH INSTITUTE Division of Intramural Research'.

UCSC Genome Bioinformatics

Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Proteome - Session - FAQ - Help

click

About the UCSC Genome Bioinformatics Site

Welcome to the UCSC Genome Browser website. This site contains the reference sequence and working draft assemblies for a large collection of genomes. It also provides portals to the [ENCODE](#) and [Neandertal](#) projects.

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

The UCSC Genome Browser is developed and maintained by the Genome Bioinformatics Group, a cross-departmental team within the Center for Biomolecular Science and Engineering (CBSE) at the University of California Santa Cruz (UCSC). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our [public mailing list](#).

News News Archives ▶

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

3 January 2012 - Roadmap Epigenomics Now Available through Data Hub at Washington University

We are pleased to announce the release of the Roadmap Epigenomics data on the UCSC Genome Browser through our Data Hub function. The Roadmap Epigenomics Project is part of The NIH Common Fund's [Epigenomics Program](#). It was launched with the goal of producing a public resource of human epigenomic data to catalyze basic biology and disease-oriented research. The Consortium leverages experimental pipelines built around next-generation sequencing technologies to map DNA methylation, histone modifications, chromatin accessibility and small RNA transcripts in stem cells and primary ex vivo tissues selected to represent the normal counterparts of tissues and organ systems frequently involved in human disease. The Consortium expects to deliver a collection of normal epigenomes that will provide a framework or reference for comparison and integration within a broad array of future studies.

The screenshot shows a web browser window with the UCSC Genome Bioinformatics website. The browser address bar shows 'http://genome.ucsc.edu/'. The website has a blue header with the title 'UCSC Genome Bioinformatics' and a navigation menu with links: 'Genomes', 'Blat', 'Tables', 'Gene Sorter', 'PCR', 'VisiGene', 'Proteome', 'Session', 'FAQ', and 'Help'. A sidebar on the left contains a list of tools: 'Genome Browser', 'ENCODE', 'Neandertal', 'Blat', 'Table Browser', 'Gene Sorter', 'In Silico PCR', 'Genome Graphs', 'Galaxy', 'VisiGene', 'Proteome Browser', 'Utilities', 'Downloads', 'Release Log', and 'Custom Tracks'. A red arrow points to the 'Genome Browser' link in the sidebar with the word 'click'. The main content area has a heading 'About the UCSC Genome Bioinformatics Site' and a welcome message. Below this is a 'News' section with a Twitter icon and a link to 'News Archives'. The news item is dated '3 January 2012' and is titled 'Roadmap Epigenomics Now Available through Data Hub at Washington University'. The text of the news item describes the release of Roadmap Epigenomics data and the goals of the project.

RefSeq Gene ADAM2
UCSC RefSeq Gene details

RefSeq: [NM_001464.3](#) **Status:** Reviewed
Description: Homo sapiens ADAM metalloproteinase 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](#)
AcView: [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](#) (mRNA derived from the genomic sequence)
- [Genomic Sequence](#) from assembly
- [CDS FASTA alignment](#) from multiple alignment

Genomic Sequence Near Gene
UCSC RefSeq Gene details

[Back](#) [Forward](#) [Reload](#) [Stop](#) [Home](#) [http://genome.ucsc.edu/cgi-bin/hgChgtsid=238330769&g=htcGeneInGeno](#)

[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 bases
 5' UTR Exons
 CDS Exons
 3' UTR Exons
 Introns
 Downstream by bases
 One FASTA record per gene.
 One FASTA record per region (exon, intron, etc.) with
 Split UTR and CDS parts of an exon into separate FASTA

Note: if a feature is close to the beginning or end of a chromosome 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

1000 nt upstream of ADAM2


```

>hg18_refGene_NM_001464 range=chr8:39814937-39815936
ggagatctaccacacataacctgtgatccgacaactcactctagaa
ataacacagtgaatccttacttattacacaaaaggcatgagaaga
atggttatagctaaatatttttaatagctggaacaataaacaaca
aatatctatcaacgtaaaatggaacacaaagtggttatattatga
attgtaataacacaaatgaggataaacagaactattgctttagatga
acctacaatcactctataaaagaaccagacatgaaagatagatggt
gattgcttctacttgcaaaaagtcaaaaacagacaaaacgaatcttgg
ttggttagaagtcattggtgaggttgaatctgggattgggtggtt
cttttcaattctcatctggtactagtaeagctgttttttttccac
ttgaatattatgaactgtgaactatgatttatataactttttc
gtttttgtttctttttttttttttttttttttttttttttttttttc
ttgctctcaccagctggagtgagtgagtgagtgagtgagtgagtgag
tgaacctctgctcctaggttcaagcattctctgctcagctcccg
agtgtgggatttcaaggacccgccaccatgctggtaatttttttt
gtatttttagtacagcggggtttccaccattgctggaaactggtctega
acctctgacagcgggtttatataattcaattgaaatcttacttaagaag
gtttataaattctctgctcctcagctgttgaagtgattttgtgtgc
tgttgccttaattagcactcagctcagtgagtgctgctcctcgaag
acagggctaggagctgcaagcttcaacagcaccacacccaacctcag
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```

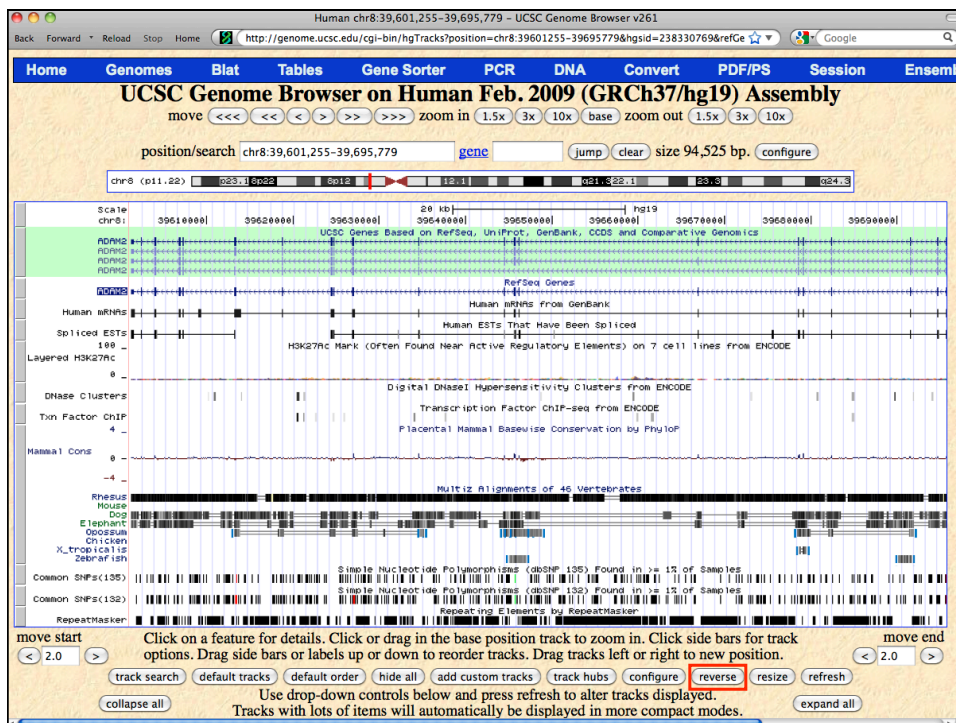


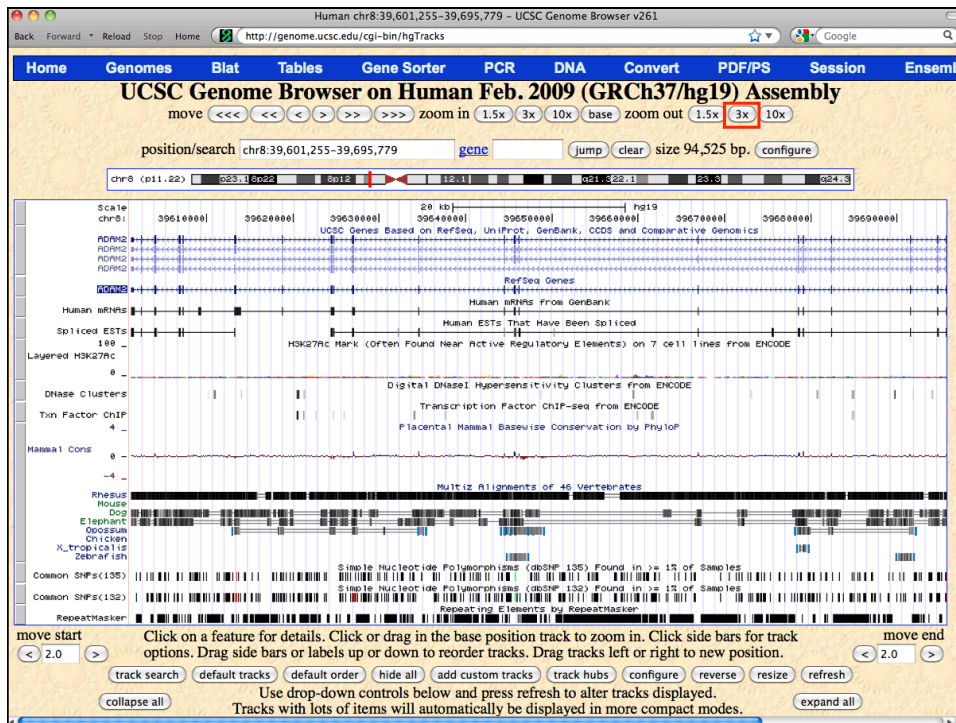
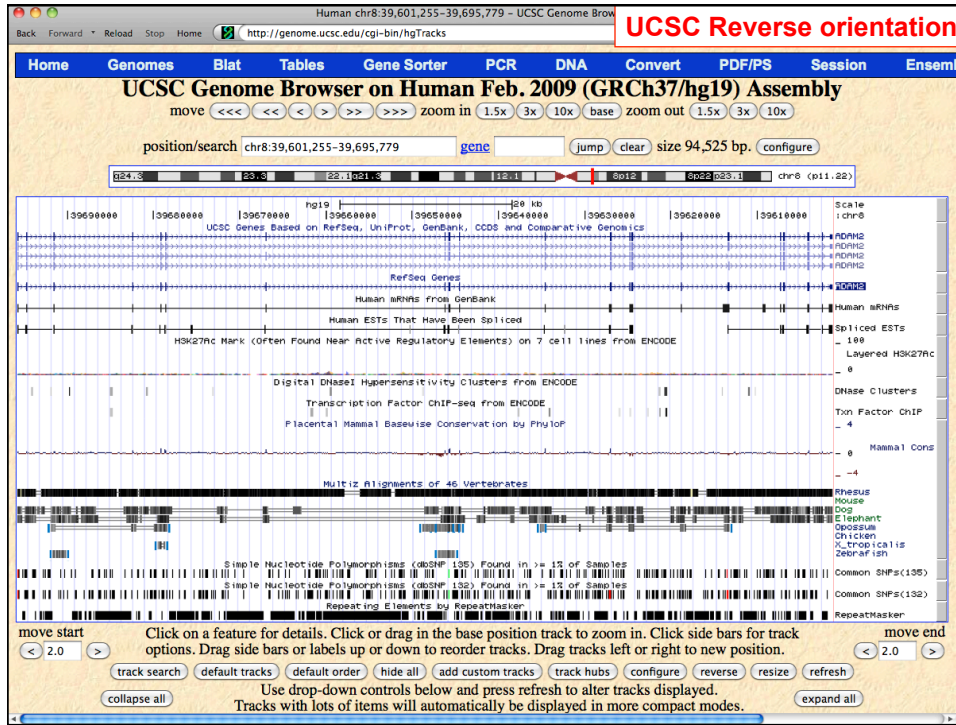
Navigating around the Genome Browser

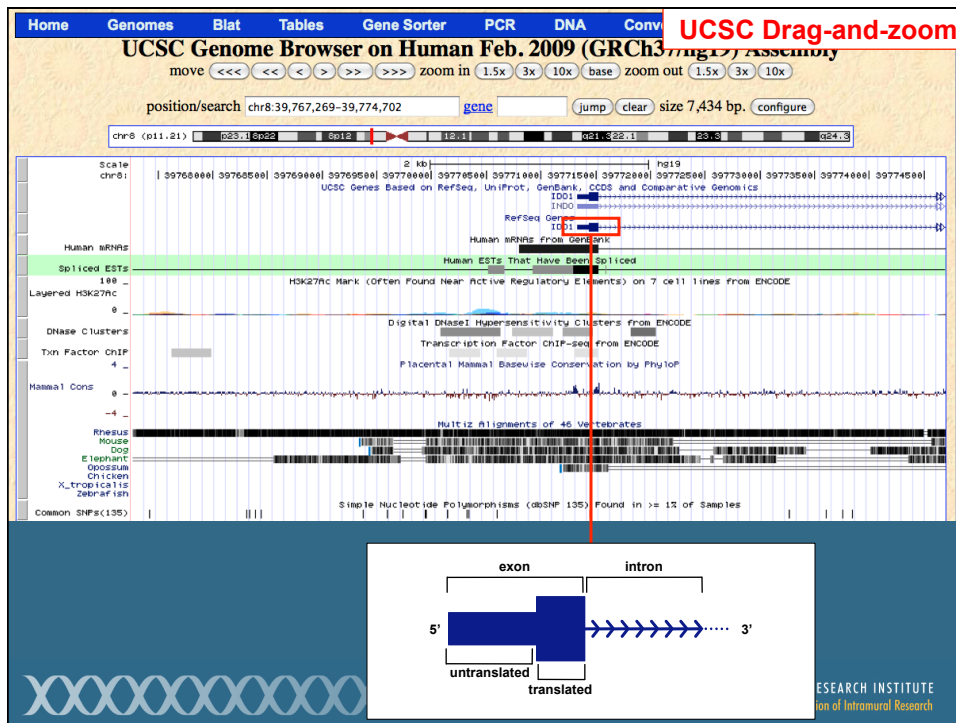
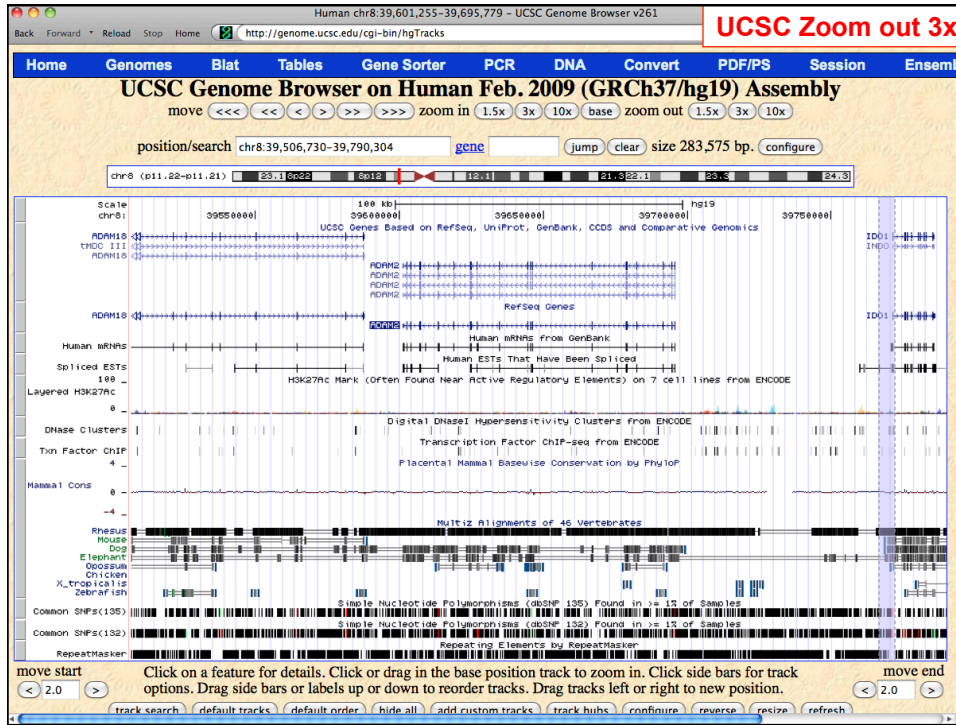
<http://genome.ucsc.edu>



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Add a track to the Genome Browser

<http://genome.ucsc.edu>

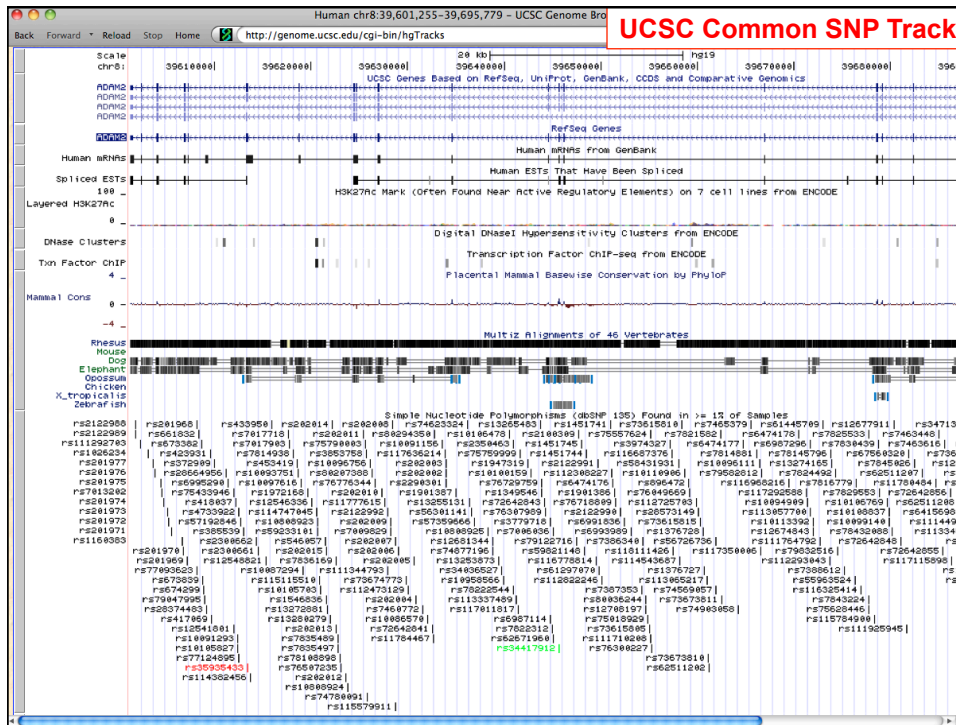
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The screenshot shows the UCSC Genome Browser interface for Human chromosome 8 (hg19). The tracks are organized into several categories:

- Regulation:** Includes tracks for ENCODE Regulation, ENCODE TF Binding, ENCODE RNA Binding, ENCODE DNA Methylation, ENCODE Histone, CpG Islands, ORegAnno, and Vista.
- Comparative Genomics:** Includes Conservation, Cons Indels, GERP, and Evid.
- Neanderthal Assembly and Analysis:** A track for comparing Neanderthal and human genomes.
- Variation and Repeats:** Includes tracks for Common SNPs(135), Flagged SNPs(135), Mult. SNPs(135), All SNPs(135), Common SNPs(132), Flagged SNPs(132), All SNPs(132), SNPs (131), Arrays, GIS DNA PET, HAIB Genotype, and HGDP Allele.

A red callout box with a 'click' arrow points to the 'refresh' button of the 'Common SNPs(135)' track. The text in the callout box defines the categories:

- Common SNPs(135):** SNPs with $\geq 1\%$ minor allele frequency (MAF), mapping only once to reference assembly.
- Flagged SNPs(135):** SNPs $< 1\%$ MAF (or unknown), mapping only once to reference assembly, flagged in dbSNP as "clinically associated" -- not necessarily a risk allele.
- Mult. SNPs(135):** SNPs mapping in more than one place on reference assembly.
- All SNPs(135):** all SNPs from dbSNP mapping to reference assembly.



UCSC SNP Track details

Common SNPs(135) Track Settings

Home Genomes Genome Browser Blat Tables Gene Sorter PCR Session FAQ Help

Common SNPs(135) Track Settings

Simple Nucleotide Polymorphisms (dbSNP 135) Found in >= 1% of Samples (▲ All Variation and Repeats tracks)

Display mode:

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:

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	<input type="text" value="black"/>	Locus	<input type="text" value="black"/>	Coding - Synonymous	<input type="text" value="green"/>	Coding - Non-Synonymous	<input type="text" value="red"/>
Untranslated	<input type="text" value="black"/>	Intron	<input type="text" value="black"/>	Splice Site	<input type="text" value="black"/>		

[View table schema](#)
 Data last updated: 2011-11-14

Description

This track contains information about a subset of the single nucleotide polymorphisms and small insertions and deletions (indels) — collectively Simple Nucleotide Polymorphisms — from dbSNP build 135, available from ftp.ncbi.nih.gov/snp. Only SNPs that have a minor allele frequency of at least 1% and are mapped to a single location in the reference genome assembly are included in this subset. Frequency data are not available for all SNPs, so this subset is incomplete.

Human chr8:39,601,255-39,695,779 - UCSC Genome Browser v261

ENCORE tracks

Expression refresh

Affy Exon Array hide
 Affy GNF1H hide
 Affy RNA Loc hide
 Affy U133 hide
 Affy U133Plus2 hide
 Affy U95 hide
 Allen Brain hide
 Burge RNA-seq hide
 ENC Exon Array... hide
 ENC ProtGeno... hide
 ENC RNA-seq... hide
 GIS RNA PET hide
 GNF Atlas 2 hide
 Illumina WG-6 hide
 Seston Brain hide

Regulation refresh

ENCORE Regulation... show
 CD34 DNaseI hide
 CpG Islands hide
 ENC DNA Methyl... hide
 ENC DNase/FAIRE... hide
 ENC Histone... hide
 ENC RNA Binding... hide
 ENC TF Binding... hide
 ORegAnno hide
 Stanf Nucleosome hide
 SUNY SwitchGear hide
 SwitchGear TSS hide

Integrated Regulation from ENCODE Tracks (All Regulation tracks)

Display mode: show Submit

+- All

hide Transcription Transcription Levels Assayed by RNA-seq on 7 Cell Lines from ENCODE

hide Layered H3K4Me1 H3K4Me1 Mark (Often Found Near Regulatory Elements) on 7 cell lines from ENCODE

hide Layered H3K4Me3 H3K4Me3 Mark (Often Found Near Promoters) on 7 cell lines from ENCODE

full Layered H3K27Ac H3K27Ac Mark (Often Found Near Active Regulatory Elements) on 7 cell lines from ENCODE

dense DNase Clusters Digital DNaseI Hypersensitivity Clusters from ENCODE

dense Txn Factor ChIP Transcription Factor ChIP-seq from ENCODE

SNPs(135) hide
 Mult. SNPs(135) hide
 All SNPs(135) hide
 Common SNPs(132) dense
 SNPs(132) hide
 Mult. SNPs(132) hide
 All SNPs(132) hide
 SNPs(131) hide
 Arrays hide
 GIS DNA PET hide
 HAIB Genotype hide

Human chr8:39,601,255-39,695,779 - UCSC Genome Browser v261

ENCORE tracks

Home Genomes Blat Tables Gene Sorter PCR DNA Convert PDF/PS Session Ensembl

UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x

position/search chr8:39,681,119-39,786,188 gene jump clear size 105,070 bp. configure

chr8 (p11.22-p11.21) 23.1 23.2 23.3 23.4 23.5 23.6 23.7 23.8 23.9 24.0

Scale chr8: 39690000 39700000 39710000 39720000 39730000 39740000 39750000 39760000 39770000 39780000

UCSC Genes Based on RefSeq, UniProt, GenBank, CCDS and Comparative Genomics

RefSeq Genes

Human mRNAs From GenBank

Human ESTs That Have Been Spliced

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

PhyloP Conservation by PhyloP

Multiple Alignments of 46 Vertebrates

Rhesus Mouse Dog Elephant Chimpanzee Chicken X_TROB1c2112 Zebrafish

Common SNPs(135) Simple Nucleotide Polymorphisms (SNPs) Found in >= 1% of Samples

Common SNPs(132) Simple Nucleotide Polymorphisms (SNPs) Found in >= 1% of Samples

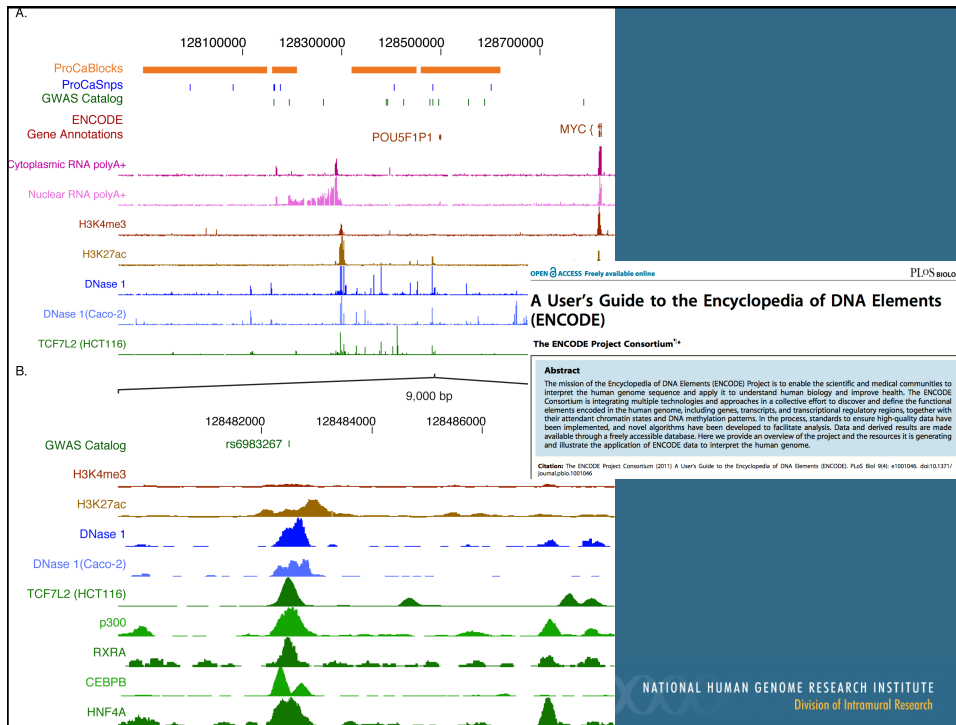
RepeatMasker Repeating Elements by RepeatMasker

move start < 2.0 > 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 < 2.0 >

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. expand all

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



UCSC

Find a chicken homolog of a human protein

<http://genome.ucsc.edu>

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The screenshot shows the NCBI Entrez Protein interface. The browser address bar displays the URL: <http://www.ncbi.nlm.nih.gov/protein/55743080?report=fasta>. The page title is "disintegrin and metalloproteinase domain-containing protein 2 preproprotein [Homo sapiens]". The NCBI Reference Sequence is NP_001455.3. The protein sequence is displayed in a monospaced font, starting with >gi|55743080|ref|NP_001455.3| disintegrin and metalloproteinase domain-containing protein 2 preproprotein [Homo sapiens]. The sequence is: MWKLVLLSLGGLRMDNFDLSPVQITVPEKIRSIKEGIESQASYKIVIEGKPYTVNLMQKNLPHNF RYVYSYSGTGMKPLDQDFQNFCHYQGYIEGPKSVVMVSTCTGLRGVLFQFENVSYGIEPLESSVGFVHVI YQVYHKKADVSLYNEKDIERSDLSPKLSQVPEQDFAKYIEMHVIKQLYNHMGSDTTVAQKVFQILG LTNALFVSNITIISSLELWIDENKIATGEANELLHTFLRWKTSYLVLRPHDVAFLVYREKSNYVGA TFGKMCNDANYAGGVVLPRTISLES LAVILAQLLSLMSGITYDDINKCQCSGAVCIMNPEALHFGVSKI FSNCSFEDFAHFISKQKSCLEHNPRLDFFKQAVCGNARLEAGEECDCGTEQDCALIGETCCDIATCR FKAGSNCAEGPCCENCLFMSKERMRCPSEFCDLPEYCNSSASCENHNVQTHPCGLNQWICIDGVCMS GDQKQCTDTFGKEVEFGSECYSHLNSKTDVSGNCGISDSGYTQCEADNLQCKLICKYVKGKFLQIPRA TIIYANISGHLCAVEFASDHADSQKMWIKDGTSCSNKVCNRQCVSSSYLYGDCDTPDKNDRGVGNK KHCHCSASYLPDCVQSDLWPGGSI DSGNFPVAIPARLPERRYIENIYHSPMRWPFLLI PFFLIIFC VLIATMVKVNFQRKWRTEYSSDEQPESESEPKG. On the right side, there are several sections: "Analyze this sequence" with options for Run BLAST, Identify Conserved Domains, Highlight Sequence Features, and Find in this Sequence; "Articles about the ADAM2 gene" with links to "Mapping, sequence, and expression analysis of the human fert [Genomics. 1997]", "Role of the integrin-associated protein CD9 in binding t [Proc Natl Acad Sci U S A. 1999]", and "Mediation of sperm-egg fusion: evidence that mouse egg alpha6beta1 [Chem Biol. 1999]"; "Identical proteins for NP_001455.3" with links to "unnamed protein product [Homr [CBH30599]", "ADAM metalloproteinase domain [EAW63273]", and "RecName: Full=Disintegrin and [Q99965]"; and "Reference sequence information" with a link to "RefSeq mRNA" and a note: "See reference mRNA sequence for the ADAM2 gene (NM_001464.3)."

The screenshot shows the UCSC BLAT search interface. The browser address bar displays the URL: <http://genome-preview.cse.ucsc.edu/cgi-bin/hgBlat>. The page title is "Chicken BLAT Search". The main heading is "BLAT Search Genome". Below the heading, there are several dropdown menus: "Genome:" set to "Chicken", "Assembly:" set to "Feb. 2004 (WUCSC 1.0/galGal2)", "Query type:" set to "BLAT's guess", "Sort output:" set to "query.score", and "Output type:" set to "hyperlink". A text area contains the same protein sequence as in the first screenshot. Below the text area are buttons for "submit", "I'm feeling lucky", and "clear". Below the buttons, there is a paragraph of instructions: "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." Below this is a section for "File Upload:" with a note: "Rather than pasting a sequence, you can choose to upload a text file containing the sequence." and a "Browse..." button. Below that is a note: "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." At the bottom, there is a note: "For locating PCR primers, use [In-Silico PCR](#) for best results instead of BLAT."

Chicken BLAT Results

UCSC BLAT search

Home Genomes Tables PCR Session FAQ Help

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

move <<< << < > >> >>> zoom in 1.5x 3x 10x (base) zoom out 1.5x 3x 10x preview

position/search chrUn:635,370-635,555 size 186 bp.

Chicken BLAT Results

UCSC BLAT search

Home Genomes Tables PCR Session FAQ Help

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

```

mvrvfllsg lgglrmdanf dslpvqitvp ekirsiikeg iesqasykiv iegkpytvnl 60
mqknlphnf rvyesygtgi mkpldqdfgn fchygyieg ypksvmvst ctglrgvlqf 120
envvyglepl esvvgfehvi ygvkhkadv slynekies rdlsfkisgv epqgdfakyi 180
emhivkekql ynhmsdttv vagkvfqlig ltnaifvsn itiilslel widenkiatt 240
geanelhft lrwktaylvi rphdvafllv yreksnyvga tfqgkmedan yagvvlhpr 300
tisleslavi laqlslsmg ityddinkq csgavcimp eaihfgsvki fscnscfedfa 360
hfiskqkqsc lhnqprldpf fkqgavcna kleageecd gteqdcailg etccdiatr 420
fkagsnaeag pccencflms kermcrpsfe ecdlpeycng ssascpenhy vqtghpcgin 480
qvicidgvcm sgdqgetdf gkevefpgse cyhlnsktd vsnecjisda gytqceadNL 540
qGKLiCKiVv gkflqipra TIIVAnisgH LLaavefaad hadsqkmwIX DGTsCGenKV 600
crngqvssss ylygdcttdk cndrgvcnk khchosasyl pdcsvqsd1 wpggsidsqn 660
fppvaipar1 perryieniy hskpmrwpff lfipffiihc vliaimvkn fgrkkwrtd 720
yssdeqpepe sepgk
    
```

Chicken.chrUn :


```

AATCTGggcT GTGGAAAACt CATCTGcaca TAcccaaaac gaggtcocct caccaaatTA 635429
aagggtACCA TCACTATAGC Tcaagtcaaa gaACATCTGT Gcgtgtettt tgatgtaatg 635489
catgcaacct ceggacaga tctctcctg gttAGGATG GCACGaaatG CGGTcccgga 635549
AAGGTA
    
```

Side by Side Alignment*


```

001615 N L Q C G K L I C K Y 001647
>>>>> | | G | | | | | | T | >>>>>
635370 aatctgggctgtggaaaactcatctgcacatac 635402
    
```



Add your own custom tracks

<http://genome.ucsc.edu>




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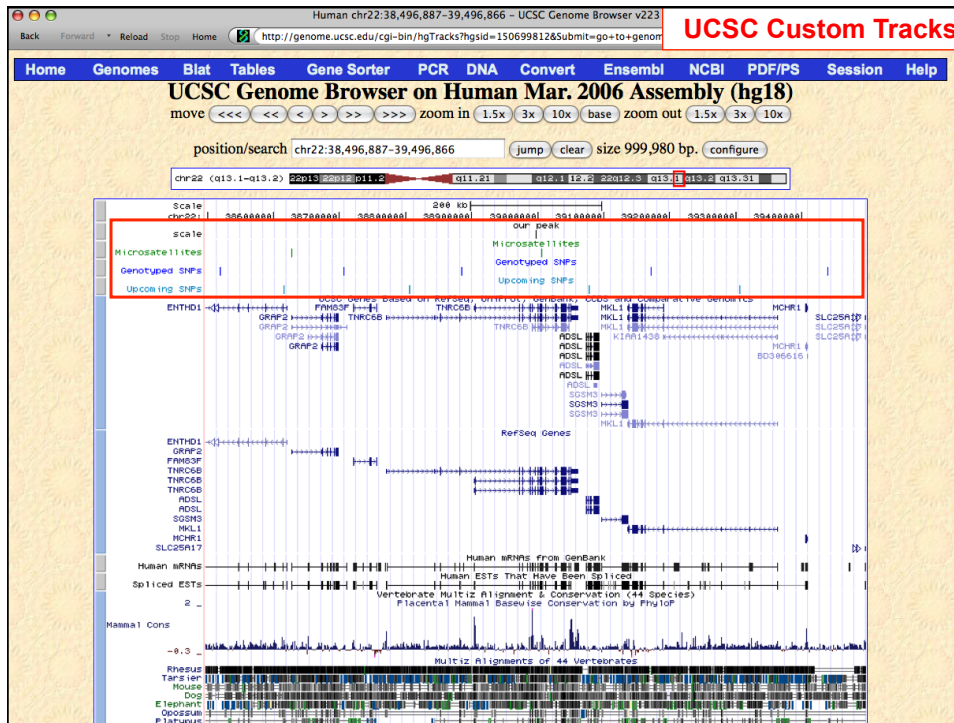
UCSC custom track format

```
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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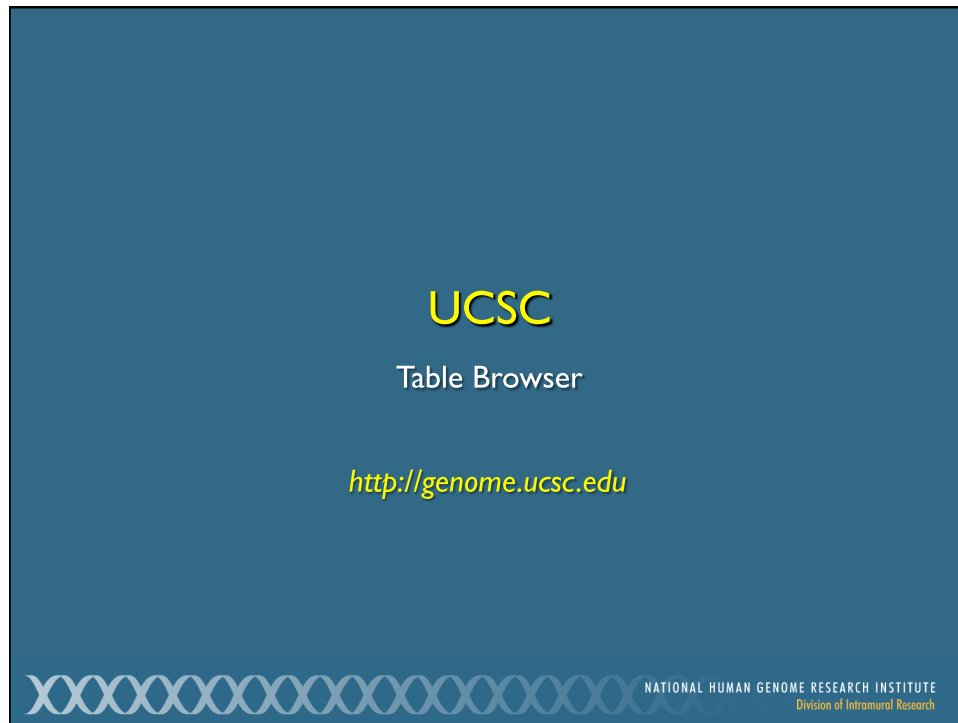


Types of UCSC custom tracks

- Upload annotation data from your computer
 - 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>

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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

Select sequence type for RefSeq Genes

- genomic
- protein
- mRNA

Sequence Retrieval Region Options:

- Promoter/Upstream by 200 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.)
- Split UTR and CDS parts of an exon into sep

Note: if a feature is close to the beginning or end of past the edge of the chromosome.

Sequence Formatting Options:

- Exons in upper case, everything else in lower ca
- CDS in
- All up
- All low
- Mask

```
hg19_refGene_NM_032291 range=chr1:66999625-66999824 5'pad=0 3'pad=0 strand=+ repeatMasking=none
hg19_refGene_NM_001145278 range=chr1:16766967-16767166 5'pad=0 3'pad=0 strand=+ repeatMasking=none
hg19_refGene_NM_001145277 range=chr1:16766967-16767166 5'pad=0 3'pad=0 strand=+ repeatMasking=none
```

Ensembl

Identify genes that overlap with an oligo tag

<http://www.ensembl.org>

Ensembl Genome Browser

Search: (All species) for (Go)
 e.g. BRCA2 or rat X:10...200000 or coronary heart disease

Browse a Genome

The Ensembl project produces genome databases for vertebrates and other eukaryotic species, and makes this information freely available online. Click on a link below to go to the species' home page.

Popular genomes (Log in to customize this list)

- Human (GRCh37)
- Mouse (NCBIM37)
- Zebrafish (Zv9)

All genomes

-- Select a species --

View full list of all Ensembl species

Other species are available in [Ensembl Pre!](#) and [EnsemblGenomes](#)

Ensembl is a joint project between EMBL-EBI and the Wellcome Trust Sanger Institute to develop a software system which produces and maintains automatic annotation on selected eukaryotic genomes. Ensembl receives major funding from the Wellcome Trust. Our [acknowledgements page](#) includes a list of additional current and previous funding bodies.

New to Ensembl?

Did you know you can:

- Learn how to use Ensembl with our video tutorials and walk-throughs
- Add custom tracks using our new Control Panel
- Upload and analyse your data and save it to your Ensembl account
- Search for a DNA or protein sequence using BLAST or BLAT
- Fetch only the data you want from our public database, using the Perl API
- Download our databases via FTP in FASTA, MySQL and other formats
- Mine Ensembl with BioMart and export sequences or tables in text, html, or Excel format

Still got questions? Try our [FAQs](#) or [glossary](#)

What's New in Release 65 (December 2011)

- Regulatory Genome Segmentation for Human
- New species: Atlantic cod
- Saving configurations (all species)

Full details of this release
 More release news on our blog -->

Latest blog posts

- New Pre! sites for cat, chicken, dog, squirrel, and squirrel monkey
- Ensembl at PAG in San Diego
- Notice: EnsemblGenomes currently offline [UPDATE: Service restored]

BLAST Search

Ensembl BLAST search

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:

```
>MESS_1
AAAATATGCGCCCTGAAGAG
```

Or Upload a file containing one or more FASTA sequences

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

Or Enter an existing ticket ID:

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

Select the databases to search against

Select species: Homo_sapiens, Loxodonta_africana, Macaca_mulatta

Select database: dna database, LATESTGP, PEP_ALL

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 mismatch, Distant homologies, No optimisation

Ensembl BLAST search

Alignment Locations vs. Karyotype (click arrow to hide)

100% identity over 100% of the query length

Query Start 1 End 20 % ID 100.00

Alignment Locations vs. Query (click arrow to hide)

Alignment Summary (click arrow to hide)

click

Subject	Chromosome	Supercontig	Clone	Contig	Lrg	Stats	Sort By
Name	Name	Name	Name	Name	Name	Score	<Score
Start	Start	Start	Start	Start	Start	E-val	>Score
Link	Query	Chromosome	Stats				
	Start	End	Ori	Name	Start	End	Score
	1	20	+	Chr:15	57210876	57210895	20
	1	17	-	Chr:2	79042858	79042875	2.2
							100.00
							20
							100.00
							17

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Ensembl

Location tab

<http://www.ensembl.org>


 NATIONAL HUMAN GENOME RESEARCH INSTITUTE
 Division of Intramural Research

Ensembl Location tab: Region in detail

click

translated exon
 untranslated exon

Genes

Gene Legend:

- CCDS set
- protein coding
- merged Ensembl/Havana
- processed transcript
- pseudogene
- RNA gene

Ensembl Location tab: Configure page

Germline variation

Enable/disable all dbSNP

Sequence variants (dbSNP and all other sources)

dbSNP variants

Key

External tracks

- DAS Distributed Annotation Source
- Temp Custom track - uploaded data
- URL Custom track - UCSC-style web resource
- Saved 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 genome browser 65: Homo sapiens - Region in detail - Chromosome 15: 57,208,876-57,212,895

Location: 15:57208876-57212895

Gene: []

Chromosome bands: Human RefSeq/CCDS set

Ensembl/Ensembl: TCF12-002 > protein coding, TCF12-025 > protein coding, TCF12-001 > protein coding, TCF12-201 > protein coding, TCF12-024 > processed transcript, TCF12-003 > nonsense mediated decay, TCF12-023 > processed transcript, TCF12-002 > protein coding, TCF12-026 > protein coding, TCF12-027 > protein coding, TCF12-028 > protein coding

BLAT/BLAST hits: AC010999.6 >

Contigs: Ensembl/Ensembl: < ZNF2800-007 > protein coding, < RP11-323F24.1-003 > processed transcript, < RP11-323F24.1-001 > processed transcript, < RP11-323F24.1-002 > processed transcript

Human RefSeq/CCDS set: dbSNP variants, %GC

Ensembl Location tab: Region in detail after navigation

Ensembl genome browser 65: Homo sapiens - Region in detail - Chromosome 15: 57,212,896-57,216,915

Location: 15:57212896-57216915

Gene: []

Chromosome bands: Human RefSeq/CCDS set

Ensembl/Ensembl: TCF12-027 > protein coding, TCF12-025 > protein coding, TCF12-001 > protein coding, TCF12-201 > protein coding, TCF12-024 > processed transcript, TCF12-003 > nonsense mediated decay, TCF12-023 > processed transcript, TCF12-002 > protein coding

Contigs: Ensembl/Ensembl: < ZNF2800-007 > protein coding, < RP11-323F24.1-003 > processed transcript, < RP11-323F24.1-001 > processed transcript, < RP11-323F24.1-002 > processed transcript

Human RefSeq/CCDS set: dbSNP variants, %GC

Variation Legend: Non-synonymous coding, Synonymous coding, Intronic

Variation: rs3611433

rs3611433 properties: chr, bp, cluster freq, 1000Genome

Gene Legend: processed transcript, pseudogene, RNA gene

Req. Features Leg.:

Gene Legend: CC, CP, Class, SNP, In, Antisense, In, CpG island, In, Repeat, Ur, Alleles

Req. Features Leg.:

There - Source Ensembl, Type SYNONYMOUS_CODING

Ch37: Chromosome 15: 57,212,896 - 57,216,915

Ensembl genome browser 65: Homo sapiens - Explore this variation - rs35615435

Ensembl Variation tab: Summary

Human (GRCh37) Location: 15:57,212,896-57,216,915 Variation: rs35615435

rs35615435 SNP

Source: dbSNP_134 - Variants (including SNPs and indels) imported from dbSNP
 Alleles: Reference/Alternative: A/G | Ancestral: A | Ambiguity code: R | MAF: 0.10 (G)
 Location: Chromosome 15:57213283 (forward strand) | View in location tab
 Validation status: This variation is validated by 1000 Genomes and also cluster, frequency
 Synonyms: dbSNP rs59892738
 HGVS names: This feature has 12 HGVS names - click the plus to show

Explore this variation help

Genomic context, Gene / Transcript, Population genetics, Individual genotypes, Linkage disequilibrium, Phenotype data, Phylogenetic context, Flanking sequence

Help with variations

YouTube videos: SNPs and other Variations - 1 of 2, SNPs and other Variations - 2 of 2, Clip: Genome Variation, BioMart: Variation IDs to HGNC Symbols

Reference materials: Ensembl variation data: background and terminology, Variation Quick Reference card

Additional resources: Accessing variation data with the Variation API, Genomes and SNPs in Malaria

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Ensembl Variation tab: Genomic context Population genetics

Context help

Ensembl/Havana... 5.00 Kb

Contigs
 Sequence variant...

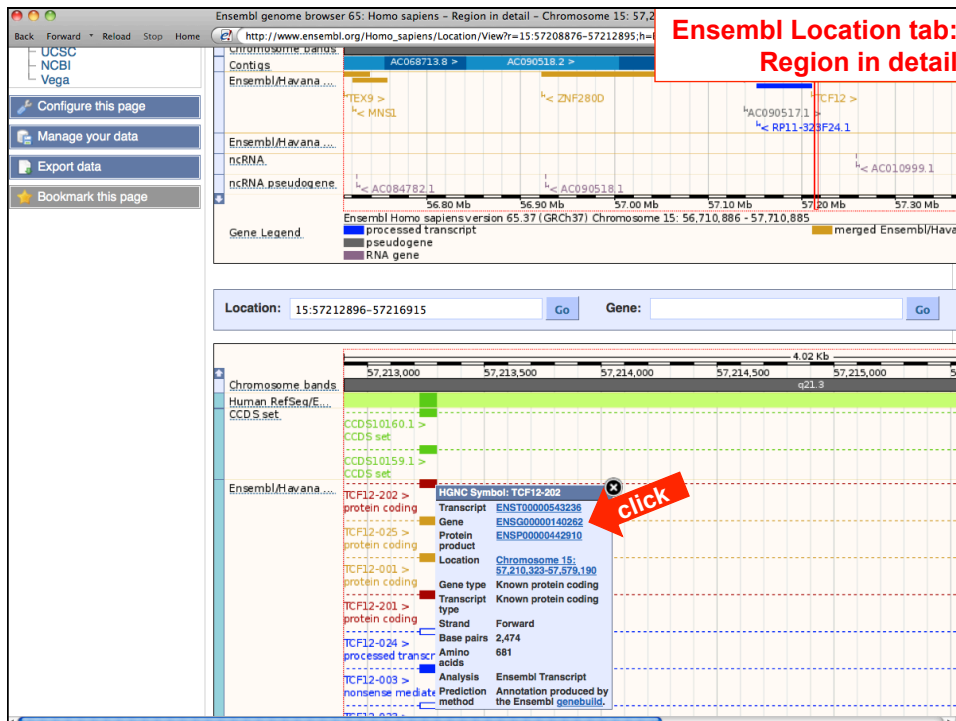
Population genetics help

1000 genomes alleles frequencies

CHB+JPT: A: 70%, G: 30%
 YRI: A: 96%, G: 4%

1000 genomes (2)

Population	ssID	Submitter	Alleles A	Alleles G	Allele count	Genotype detail
1000GENOMES:pilot_1_CHB+JPT_low_coverage_panel	ss243177144	1000GENOMES	0.700	0.300	84 (A) / 36 (G)	Show
1000GENOMES:pilot_1_YRI_low_coverage_panel	ss226911693	1000GENOMES	0.958	0.042	113 (A) / 5 (G)	Show



Ensembl genome browser 65: Homo sapiens - Gene summary - Gene: TCF12 (ENSG00000140262)

Ensembl BLAST/BLAT BioMart Tools Downloads More

Human (GRCh37) Location: 15:57,212,896-57,216,915 Gene: TCF12 Transcript: TCF12-202

Gene: TCF12 ENSG00000140262

Description transcription factor 12 [Source:HGNC Symbol;Acc:11623]
 Location Chromosome 15: 57,210,323-57,591,479 forward strand.
 Transcripts This gene has 31 transcripts

Gene-based displays
 Gene summary
 Splice variants (31)
 Supporting evidence
 Sequence
 External references
 Regulation
 Comparative Genomics
 Genomic alignments
 Gene Tree (image)
 Gene Tree (text)
 Gene Tree (alignment)
 Orthologues (60)
 Paralogs (2)
 Protein families (2)
 Phenotype
 Genetic Variation
 Variation Table
 Variation Image
 Structural Variation
 External Data
 Personal annotation
 ID History
 Gene history

Configure this page
 Manage your data
 Export data
 Bookmark this page

click

Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
TCF12-001	ENST00000267811	6061	ENSP00000267811	682	Protein coding	CCDS10158
TCF12-002	ENST00000333725	4719	ENSP00000331057	706	Protein coding	CCDS10160
TCF12-004	ENST00000543579	1809	ENSP00000440017	536	Protein coding	-
TCF12-006	ENST00000537840	1598	ENSP00000446966	446	Protein coding	-
TCF12-007	ENST00000559922	1598	ENSP00000453216	446	Protein coding	-
TCF12-008	ENST00000343827	3956	ENSP00000342459	512	Protein coding	CCDS42049
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	CCDS10158
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	ENST00000560784	1230	No protein product	-	Processed transcript	-
TCF12-020	ENST00000560506	575	No protein product	-	Processed transcript	-

Ensembl genome browser 65: Homo sapiens - Orthologues - Gene: TCF12 (ENSG00000140262)

Ensembl BLAST/BLAT BioMart Tools Downloads More

Human (GRCh37) Location: 15:57,212,896-57,216,915 Gene: TCF12 Transcript: TCF12-202

Gene: TCF12 ENSG00000140262

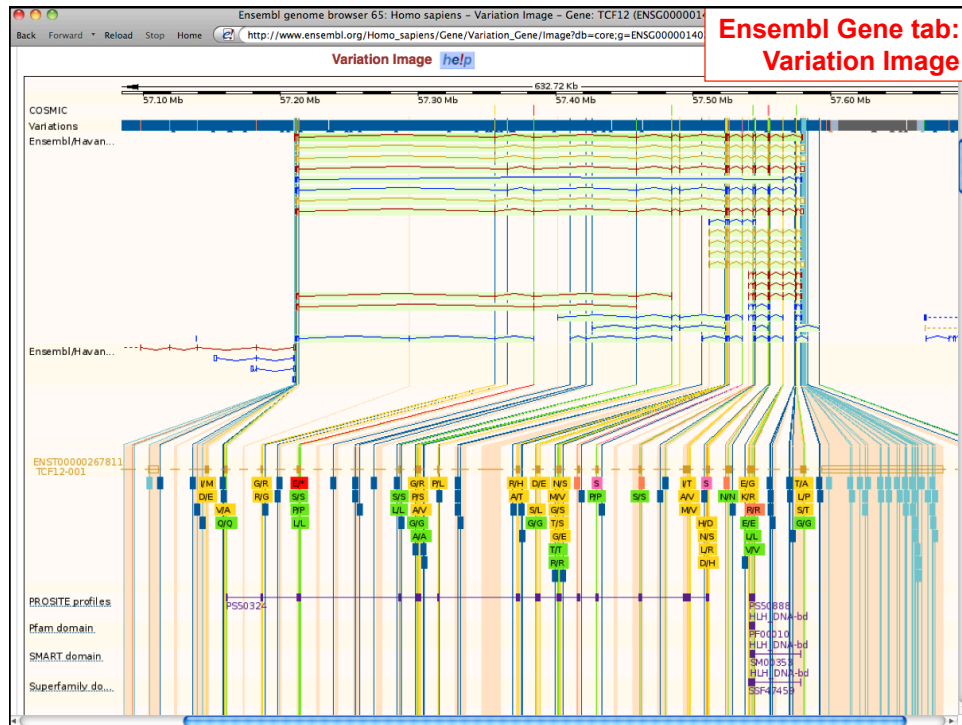
Selected orthologues

View sequence alignments of these homologues.

Gene-based displays
 Gene summary
 Splice variants (31)
 Supporting evidence
 Sequence
 External references
 Regulation
 Comparative Genomics
 Genomic alignments
 Gene Tree (image)
 Gene Tree (text)
 Gene Tree (alignment)
 Orthologues (60)
 Paralogs (2)
 Protein families (2)
 Phenotype
 Genetic Variation
 Variation Table
 Variation Image
 Structural Variation
 External Data
 Personal annotation
 ID History
 Gene history

click

Species	Type	dN/dS	Ensembl identifier & gene name	Compare	Location	Target %id	Query %id
Alpaca (<i>Vicugna pacos</i>)	1-to-1	n/a	ENSPVAG00000006545 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 (<i>Anolis carolinensis</i>)	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 (<i>Anolis carolinensis</i>)	Possible ortholog	n/a	ENSACAG00000027602	Multi-species view Alignment (protein) Alignment (cDNA) Gene Tree (image)	AAWZ02036688:11541-14447:-1	73	12
Armadillo (<i>Dasypus novemcinctus</i>)	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 (<i>Otolemur</i>)	1-to-1	0.19081	ENSOGAG00000006485 TCF12	Multi-species view	GL873530.1:9653822-9743765:1	95	68



Ensembl

Transcript tab

<http://www.ensembl.org>

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Ensembl genome browser 65: Homo sapiens - Supporting evidence - Transcript: TCF12-201

Ensembl BLAST/BLAT BioMart Tools Downloads More

Human (GRCh37) Location: 15:57,212,896-57,216,915 Gene: TCF12 Transcript: TCF12-201

Ensembl Transcript tab: Transcript summary

click

Transcript: TCF12-201 ENST00000452095

Description transcription factor 12 [Source:HGNC Symbol;Acc:11623]
 Location Chromosome 15: 57,210,833-57,580,712 forward strand.
 Gene This transcript is a product of gene ENSG00000140262 - This gene has 31 transcripts

Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
TCF12-001	ENST00000267811	6061	ENSP00000267811	682	Protein coding	CCDS10158
TCF12-002	ENST00000333725	4719	ENSP00000331057	706	Protein coding	CCDS10160
TCF12-004	ENST00000543579	1809	ENSP00000440017	536	Protein coding	-
TCF12-006	ENST00000537840	1598	ENSP00000446996	446	Protein coding	-
TCF12-007	ENST00000559922	1598	ENSP00000453216	446	Protein coding	-
TCF12-008	ENST00000343827	3956	ENSP00000342459	512	Protein coding	CCDS42049
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	CCDS10158
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	686	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	ENST00000560784	1230	No protein product	-	Processed transcript	-
TCF12-020	ENST00000560506	575	No protein product	-	Processed transcript	-

Ensembl genome browser 65: Homo sapiens - Supporting evidence - Transcript: TCF12-201

Ensembl BLAST/BLAT BioMart Tools Downloads More

Human (GRCh37) Location: 15:57,212,896-57,216,915 Gene: TCF12 Transcript: TCF12-201

Ensembl Transcript tab: Supporting evidence

click

Supporting evidence

Legend: protein evidence (yellow), EST evidence (purple), cDNA evidence (green)

Ensembl genome browser 65: Homo sapiens - Protein sequence - Transcript: TCF12

http://www.ensembl.org/Homo_sapiens/Transcript/Sequence_Protein?db=core;g=TCF12

Ensembl Transcript tab: Protein sequence

Transcript ID	ENST ID	Length	Protein product	Protein length	Transcript status
TCF12-003	ENST00000559609	2252	ENSP00000453878	666	-
TCF12-011	ENST00000561449	914	No protein product	-	-
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
TCF12-021	ENST00000561420	837	No protein product	-	Processed transcript
TCF12-022	ENST00000558210	639	No protein product	-	Processed transcript
TCF12-023	ENST00000558908	553	No protein product	-	Processed transcript
TCF12-024	ENST00000560190	1183	No protein product	-	Processed transcript
TCF12-029	ENST00000561454	564	No protein product	-	Processed transcript
TCF12-030	ENST00000560191	1205	No protein product	-	Processed transcript
TCF12-031	ENST00000560948	440	No protein product	-	Processed transcript
TCF12-032	ENST00000560836	212	No protein product	-	Processed transcript

Transcript and Gene level displays
 Views in Ensembl are separated into gene based views and transcript based views according to which level the information is more appropriately associated. To flip between the two sets of views you can click on the Gene and Transcript tabs in the menu bar at the top of the page.

Protein sequence [help](#)

Key

Exons Alternating exons Alternating exons Residue overlap splice site

```

MNPQQRMAALIGTDEKLSDDLDFSAMFSPVPVNSGKTRPTTLGSSQFSGSGTIEHMKQLNS
KARQKQRIKGFDTSPHYSDHLNDRSLGAHEGLSPTPFMNSNLGKTSERGSFSLYSDRT
GLPGCQSSLLRQDLGLGSPAQLSSGKPGTYYSFSAATSSRRRLHDSAALDPLQAKKVR
KVPFGLPSSVYAPSPNSCDFNRESFSYSPFRFPTMEASTFFMGDGHNSDLWSSNGM
SQPFSGILLQTSFHSMSQSSBYONLHSDRLSYPHVSPTINTSLPFMSFIRGSSSS
SPYVAASHTPPINGSDSLGTRGNAGSSQTDGALKALASISYDHTSSPSSPNSTPV
GSPFLTGTQWPRPQGGQAPSSPVSLENLSLHKNRVEQLHEHLQDAMSFLKDVCEQSRM
EDRLRLDDALHVLRNHVAVGPSTSLPAGHSDIHSLLGPHSNAPIGSLNNGYGGSSLVASS
RSASVGTTHRSDVSLNGNHSVLSSTVTTSSDLNHTQENYRGGLOSQSGVTVTEIKT
ENRDKDENLHEPPSSDMKSDDESQKDKVSSRGRTSSTNEDELDNPEKQIEREKRRM
ANNARELRVVDIENAFKELGRMCQLHLSKSEKQVLLLRQAVVILSLEQQVRENLN
PKAACLRREKESVAVSAEPPFTLPGTHRSGTINFGHM
    
```

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[Permanent link](#) - [View in archive site](#)

Click

Ensembl genome browser 65: Homo sapiens - Protein sequence - Transcript: TCF12-201 (ENST00000453878)

http://www.ensembl.org/Homo_sapiens/Transcript/Sequence_Protein?db=core;g=TCF12-201;transcript=ENST00000453878

Ensembl archive

Transcript ID	ENST ID	Length	Protein product	Protein length	Transcript status
TCF12-003	ENST00000559609	2252	ENSP00000453878	666	Nonsense mediated decay
TCF12-011	ENST00000561449	914	No protein product	-	Processed transcript
TCF12-013	ENST00000559216	475	No protein product	-	Processed transcript

View in archive site

The following archives are available for this page:


- [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
- [Ensembl 51: Nov 2008 \(NCBI36\)](#) - patched/updated gene set Sep 2008
- [Ensembl 50: Jul 2008 \(NCBI36\)](#)
- [Ensembl 46: Aug 2007 \(NCBI36\)](#)

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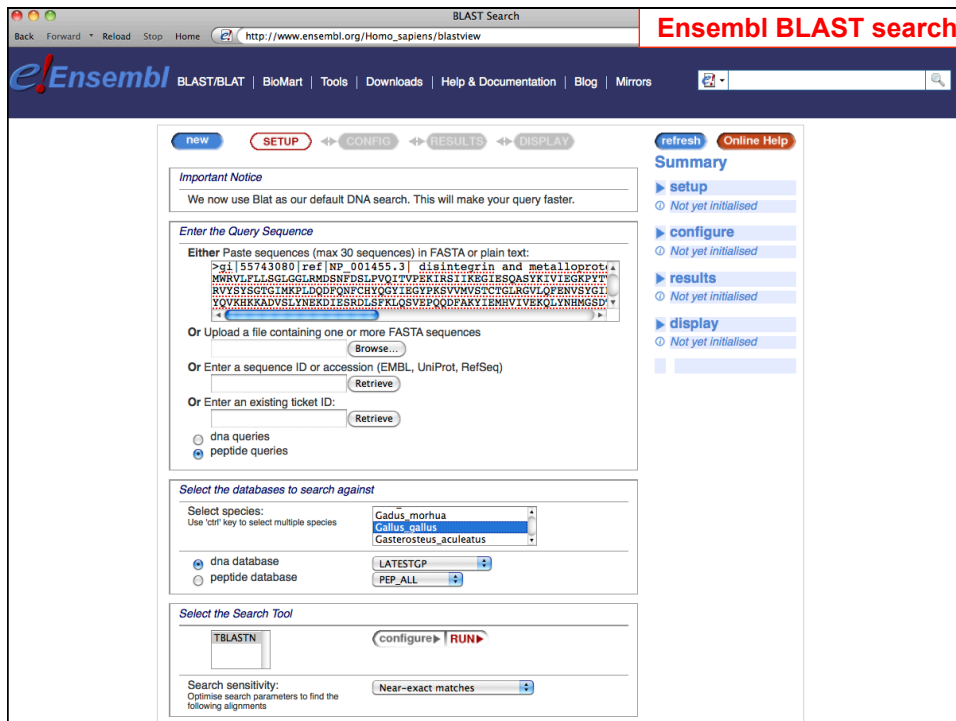


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Find a chicken homolog of a human protein

<http://www.ensembl.org>

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BLAST Search

Ensembl BLAST search

new SETUP CONFIG RESULTS DISPLAY refresh Online Help

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:

```
>g155743080|ref|NP_001455.3| diintegrin and metalloprot...
MWRVLFLLSLGGLRMDSNFDSLPPVQITVPEKIRSIKRGIESQASYKIVIEGKPYT
RVNYSFGGIMKPLDQDFQNFCHVQGITIGVFKSVVWVSTCTLAGVLFQFNVSYGI
YVYKREKADVSLYNEKDIISRDLSEKLSQVPEPODYAKYIEBHVIYVKOLYNHGSD*
```

Or Upload a file containing one or more FASTA sequences

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

Or Enter an existing ticket ID:

dna queries
 peptide queries

Select the databases to search against

Select species: Cadus morhua, Gallus gallus, Gasterosteus aculeatus

dna database
 peptide database

LATESTGP
PEP_ALL

Select the Search Tool

TBLASTN

Search sensitivity: Near-exact matches

Summary

- setup (Not yet initialised)
- configure (Not yet initialised)
- results (Not yet initialised)
- display (Not yet initialised)

▼ 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: Supercontig: Contig: Stats: Sort By:

Ensembl BLAST search

L	Query	Subject	Chromosome	Start	End	Ori	Score	E-val	%ID	Length				
[A]	[S]	[G]	[C]	4	669	+	Chr:9	2718636	29720642	+	1465	1.1e-129	33.05	708
[A]	[S]	[G]	[C]	8	505	+	Chr:15	6293553	6256064	+	1194	2.7e-107	35.75	537
[A]	[S]	[G]	[C]	278	668	+	Chr:15	6295085	6296212	+	1016	4.0e-86	37.99	408

Query location : ref|NP_001455.3| 4 to 669 (+)

Database location : 5 29718636 to 29720642 (+)

Genomic location : 5 29718636 to 29720642 (+)

Alignment score : 1465

E-value : 1.1e-129

Alignment length : 708

Percentage identity: 33.05

Query: 4 VFLFLGGLRLMDSNFDLSPVOIT----VPEKIRSIIEKIGESQASVKYIVIEGKPYTV 58

Sbjct: 29718636 VLVVLLGLVGCPTTDDSEGLHVGWVTVVPROL-SPRADTNPLTVSWLQVGRPQVL 29718812

Query: 59 NLM-QKNFLPHNFRVVSYSCTGIMKPLQDF-QNPHYQGYIEGYKSVVWVSTC-TGLR 115

Sbjct: 29718813 RLRPRKGLASRPFLLVTDDEGARRE-EQVYVDNCFYQGEVQSGPSGLVALGTCGRGLR 29718989

Query: 116 GVLQFENVSYGIEPLESSVGFHVIYQVHKKADVSLYNEK-DIESRDLSEK-----LQ 168

Sbjct: 29718990 GVLNMGSTYIEIPDDPAPQRMLYRME---ADSDPMGPTCGLTPEELQYQKTVLPWLQ 29719160

Query: 169 S--VEPQ---ODF---AKYIEMHVIKELYNHMGSD---TTVVAQKVFOLIGLINAIFV 217

Sbjct: 29719161 APRTEDRYMLKDWHTRYVYKLVVVVDNVRP--VRSDRNESKVLRLQ-VLEVVNIGDSLVD 29719331

Query: 218 SFNITILSSLELWIDENKIATTEANELLHTFLRWKTSYLVLVLR-PHDVAPLLVYRE--K 274

Sbjct: 29719332 QLSVQLFLVGLIWTNSNPNITKSASKTLADFNWRKSDLYPRMHDHTAHLFAPQGFQK 29719511

Query: 275 SNVYVATFGQKMCNDANYAGVLEP-RTISLES LAVILQLLSLMSGITDYDDINKQCQSG 333

Sbjct: 29719512 S--LGLAYLGSICDRQWSAAVDSYNNRRLS--SFIVTFVHELHNLGNRHDH-RHCKCR 29719676

Query: 334 AVCIM-NPEAIFHSGVKIFSNCSFDFARFIFSKQKSQLHNQPLRDPFF--KQAVCGNA 390

Sbjct: 29719677 KRCIMYSE----SDTDFSDCSYKDYFDLLGRGGS-CLYQAPALGSIYTLKRE-YCGNK 29719838

Query: 391 KLEAGEBCDGTGQDCALIGETCCDIATCRFRAGSNCAEGPCENCLFMSKERMCRPSFE 450

Sbjct: 29719839 IVBSGQDCDGSKSDCR--DPCCH-FNCTLTAGSVCASGKCKGQCLIPAGTLICRART 29720009

Ensembl

Using BioMart to cross-reference data from different sources

<http://www.ensembl.org>

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Ensembl BioMart
 Get genomic coordinates, gene name, and RefSeq accessions for a list of ENSEMBL gene identifiers

Step 1: Select Dataset

Step 2: Select Filters (input)

Please restrict your query using criteria below

REGION:

GENE:

Limit to genes ... with Spaink Lab Leiden3 probe ID(s)

Only
 Excluded

ID list limit

Ensembl Gene ID(s) [e.g. ENSG00000139618]

ENSDARG00000000906
 ENSDARG00000002006
 ENSDARG00000002507
 ENSDARG00000004358
 ENSDARG00000004561

Transcript count >=

Gene type

miRNA
 misc_RNA
 Mt_rRNA
 Mt_rRNA
 protein_coding

Ensembl BioMart
 Get genomic coordinates, gene name, and RefSeq accessions for a list of ENSEMBL gene identifiers

Step 3: Select Attributes (output)

Please select columns to be included in the output and hit

click

Ensembl BioMart

Ensembl

Ensembl Gene ID
 Ensembl Transcript ID
 Ensembl Protein ID
 Description
 Chromosome Name
 Gene Start (bp)
 Gene End (bp)
 Associated Gene Name
 Gene End (bp)

Associated Gene Name
 Associated Transcript Name
 Associated Gene DB
 Associated Transcript DB
 Transcript count
 % GC content
 Gene Biotype

External References (max 3)

PDB ID
 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 transcript ID(s) (OTTT)
 VEGA gene ID(s) (OTTG)
 Ensembl transcript (where OTTT shares CDS with ENST)
 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
 RefSeq Predicted Protein ID
 Rfam ID
 Rfam gene name
 Rfam transcript name
 Unigene ID
 UniProt/TrEMBL Accession
 UniProt/SwissProt ID
 UniProt/SwissProt Accession
 UniProt Gene Name
 WikiGene name
 WikiGene description
 ZFIN ID
 ZFIN symbol
 ZFIN xpat
 ZFIN transcript name
 RefSeq mRNA
 RefSeq mRNA predicted
 RefSeq ncRNA
 RefSeq ncRNA predicted

Mozilla Firefox
 http://www.ensembl.org/biomart/martview/1b065707d5a482a61a3f2379b81a2c6a

Ensembl BioMart
 Get genomic coordinates, gene name, and RefSeq accessions for a list of ENSEMBL gene identifiers

Export all results to: File TSV Unique results only

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	RefSeq mRNA predicted
ENSDARG00000000906	ENSDART000000052660	16	23018783	23062136	skap2	NM_200628	
ENSDARG00000000906	ENSDART00000137344	16	23018783	23062136	skap2		
ENSDARG00000002006	ENSDART00000021596	16	20493224	20528393	rxrb	NM_131238	
ENSDARG00000002006	ENSDART00000147844	16	20493224	20528393	rxrb		
ENSDARG00000002006	ENSDART00000128914	16	20493224	20528393	rxrb		
ENSDARG00000002507	ENSDART00000139859	16	16045949	16118555	lga10		XM_0032001
ENSDARG00000002507	ENSDART0000011224	16	16045949	16118555	lga10		
ENSDARG00000004358	ENSDART0000012673	16	13772550	13799769	grb3a	NM_001002437	
ENSDARG00000004561	ENSDART00000142610	16	14772197	14861170	prkca		XM_0019216
ENSDARG00000004561	ENSDART00000103886	16	14772197	14861170	prkca		
ENSDARG00000004806	ENSDART00000121998	16	15611720	15622320	grwd1	NM_001003509	
ENSDARG00000005782	ENSDART00000138611	16	16979935	17345861	col14a1		
ENSDARG00000005782	ENSDART00000137912	16	16979935	17345861	col14a1		
ENSDARG00000005782	ENSDART00000027982	16	16979935	17345861	col14a1		XM_0019220
ENSDARG00000005782	ENSDART00000134087	16	16979935	17345861	col14a1		
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		
ENSDARG00000009223	ENSDART00000146436	16	22143616	22239485	ankrd28		XM_684152
ENSDARG00000009223	ENSDART00000027020	16	22143616	22239485	ankrd28		
ENSDARG000000013371	ENSDART00000007842	16	14545332	14561307	isoc2	NM_001079953	
ENSDARG000000013371	ENSDART00000146997	16	14545332	14561307	isoc2		
ENSDARG000000018787	ENSDART00000119566	16	25521948	25537442	efna1b	NM_200783	
ENSDARG000000018787	ENSDART00000135279	16	25521948	25537442	efna1b		
ENSDARG000000019753	ENSDART00000131627	16	25838201	25958945	KCNN3		XM_0019217
ENSDARG000000019753	ENSDART00000103211	16	25838201	25958945	KCNN3		
ENSDARG000000023031	ENSDART00000009827	16	23011103	23013613	hoxa2b	NM_131106	

Find: 19692 Next Previous Highlight all Match case

Ensembl BioMart:
 Get predicted human orthologs for list of ENSEMBL gene identifiers

Please select columns to be included in the output and

Features
 Homologs
 Structures
 Variation
 Transcript Event
 Sequences

GENE:

ORTHOLOGS (Max select 3 orthologs):

Atlantic Cod Orthologs

Atlantic Cod Ensembl Gene ID
 Representative Protein or Transcript ID
 Atlantic Cod Ensembl Protein ID
 Chromosome Name
 Atlantic Cod Chromosome End (bp)
 Homology Type
 Ancestor
 % Identity

Human Orthologs

Human Ensembl Gene ID
 Representative Protein or Transcript ID
 Human Ensembl Protein ID
 Human Chromosome
 Human Chromosome Start (bp)
 Human Chromosome End (bp)
 Homology Type
 Ancestor
 dS
 dN
 % Identity
 Human % Identity

Ensembl Gene ID	Ensembl Transcript ID	Human Ensembl Gene ID	Human Ensembl Protein ID	% Identity
ENSDARG00000000906	ENSDART000000052660	ENSG00000005020	ENSP00000005587	58
ENSDARG00000000906	ENSDART00000137344	ENSG00000005020	ENSP00000005587	58
ENSDARG00000002006	ENSDART00000021596	ENSG00000204231	ENSP00000363812	70
ENSDARG00000002006	ENSDART00000147844	ENSG00000204231	ENSP00000363812	70
ENSDARG00000002006	ENSDART00000128914	ENSG00000204231	ENSP00000363812	70
ENSDARG00000002507	ENSDART00000139859			
ENSDARG00000002507	ENSDART0000011224			
ENSDARG00000004358	ENSDART0000012673			
ENSDARG00000004561	ENSDART00000142610	ENSG00000126583	ENSP00000263431	69
ENSDARG00000004561	ENSDART00000103886	ENSG00000126583	ENSP00000263431	69
ENSDARG00000004806	ENSDART00000121998	ENSG00000105447	ENSP00000253237	59
ENSDARG00000005782	ENSDART00000138611	ENSG00000187955	ENSP00000297848	59
ENSDARG00000005782	ENSDART00000137912	ENSG00000187955	ENSP00000297848	59
ENSDARG00000005782	ENSDART00000027982	ENSG00000187955	ENSP00000297848	59
ENSDARG00000005782	ENSDART00000134087	ENSG00000187955	ENSP00000297848	59

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View a genomic region between two SNPs

<http://www.ncbi.nlm.nih.gov/mapview>

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Map Viewer query page

The Map Viewer provides a wide variety of genome mapping and sequencing data. More...

Search
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 for:

Tools Legend
 News
Nasonia vitripennis build 2.1 released Nov 1, 2011
 Nasonia vitripennis build 2.1 has been released and is now a... [more](#)
Oreochromis niloticus build Orenil1.0 released Oct 31, 2011
 Oreochromis niloticus build Orenil1.0 has been released and ... [more](#)
Cavia porcellus build 1.1 released Oct 31, 2011
 Cavia porcellus build 1.1 has been released and is now avail... [more](#)
Canis familiaris build 2.2 released Oct 27, 2011
 Canis familiaris build 2.2 has been released and is now avai... [more](#)
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 Genome Browser agreement
 Genome Biology
 Taxonomy
 Entrez (Global Query)
 BLAST
 Map Viewer FTP

Scientific name	Common name	Build	Tools
<i>Callithrix jacchus</i>	white-tufted-ear marmoset	Build 1.1	Q B R
<i>Homo sapiens</i>	human	Build 37.3 Build 36.3	Q B R Cf G
<i>Macaca mulatta</i>	rhesus macaque	Build 1.2	Q B R Cf G
<i>Nomascus leucogenys</i>	Northern white-cheeked gibbon	Build 1.1	Q B
<i>Pan troglodytes</i>	chimpanzee	Build 3.1 Build 2.1	Q B R G
<i>Pongo abelii</i>	Sumatran orangutan	Build 1.2	Q B R

Scientific name	Common name	Build	Tools
<i>Cavia porcellus</i>	Domestic guinea pig	Build 1.1	Q B
<i>Cricetus griseus</i>	Chinese hamster	Build 1.1	Q B
<i>Mus musculus</i>	laboratory mouse	Build 37.2 Build 36.1	Q B R Cf G
<i>Rattus norvegicus</i>	rat	RGSC v3.4	Q B R Cf G

Scientific name	Common name	Build	Tools
<i>Aspergillus clavatus</i>		Build 1.1	Q B G
<i>Aspergillus fumigatus</i>		Build 2.1	Q B R G
<i>Aspergillus niger</i>		Build 1.1	Q B R G

Entrez Genome view
 http://www.ncbi.nlm.nih.gov/projects/mapview/map_search.cgi?taxid=9606&quer

Map Viewer results page

NCBI
 NCBI Map Viewer

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

Hits: 4

Search results for query "rs76552724 OR rs13269090": 4 hits

Chr	Assembly	Match	Map element	Type	Maps	
8	reference	all matches	rs13269090	SNP	Variation	
			rs76552724	rs76552724	SNP	Variation
8	HuRef-Primary Assembly	all matches	rs13269090	SNP	Variation	
			rs76552724	rs76552724	SNP	Variation

click

Map Viewer
 http://www.ncbi.nlm.nih.gov/projects/mapview/maps.cgi?taxid=9606&chr=8&MAP

Map Viewer default view

NCBI
 NCBI Map Viewer

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 [clear]

Master Map: Variation Summary of Maps Maps & Options

Region Displayed: 37,566K-37,685K bp Download/View Sequence/Evidence

Map	Gene	Het	Validation	Genotypes Avail	Linkout	Evidence
rs76552724	LTC	Individual	100%	>80-95%	★	
rs34410627	LTC	Individual				
rs74435605	LTC	Individual				
rs12545190	LTC	Individual				
rs78578713	LTC	Individual				
rs112360090	LTC	Individual				
rs74316734	LTC	Individual				
rs4739538	LTC	Individual			★	
rs4739540	LTC	Individual				
rs4739541	LTC	Individual			★	
rs762123	LTC	Individual			★	
rs11781156	LTC	Individual				
rs112278207	LTC	Individual				
rs112352687	LTC	Individual				
rs6468438	LTC	Individual				
rs34967654	LTC	Individual				
rs13274161	LTC	Individual				
rs118116446	LTC	Individual				

NCBI

Change the maps displayed on the Map Viewer

<http://www.ncbi.nlm.nih.gov/mapview>

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Map Viewer Maps & Options

Organism: *Homo sapiens*
Chromosome: 8 Region Shown: 37566100 37685100

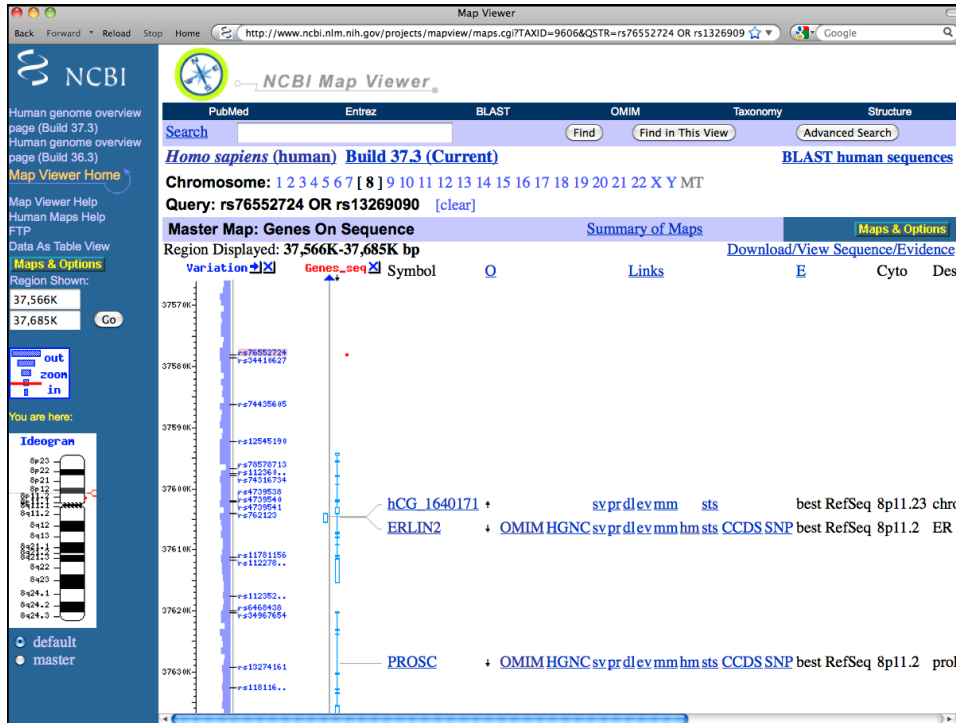
Available Tracks

- Organism: human
- Assembly: GRCh37.p5-Primary Assembly
- Sequence Maps
 - Assembly
 - Assembly regions
 - Clone
 - Component
 - Contig
 - CpG Island
 - Ensembl Genes
 - Ensembl Transcripts
 - FISH Clone (seq)
- Cytogenetic Maps
- Genetic Maps
- RH Maps

Tracks Displayed (left to right)

- [GRCh37.p5-Primary Assembly] Variation
- [GRCh37.p5-Primary Assembly] Gene

default
master

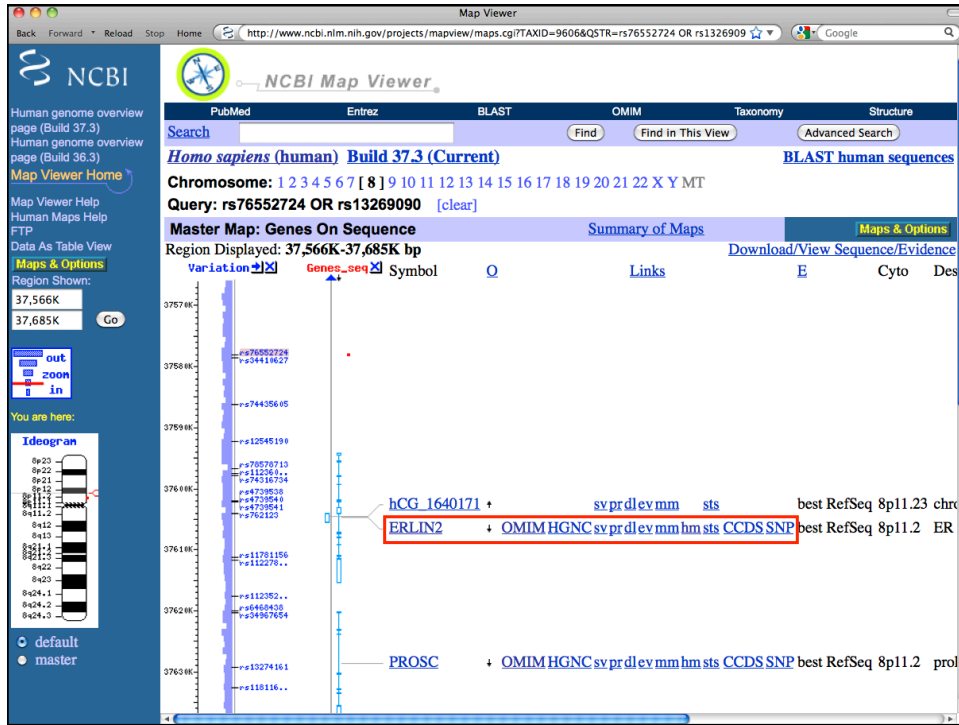


NCBI

View additional information about a gene

<http://www.ncbi.nlm.nih.gov/mapview>

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ERLIN2 ER lipid raft associated 2 [Homo sapiens] - Gene - NCBI

http://www.ncbi.nlm.nih.gov/sites/entrez?db=gene&cmd=retrieve&opt=full_report&list_uids=11

NCBI: Entrez Gene

NCBI Reference Sequences (RefSeq)

RefSeqs maintained independently of Annotated Genomes

These reference sequences exist independently of genome builds. [Explain](#)

mRNA and Protein(s)

1. **NM_001003790.2 → NP_001003790.1 erlin-2 isoform 2**
Status: VALIDATED

Description: Transcript Variant: This variant (2) uses different segments for its 5' UTR and for its 3' coding region and 3' UTR, compared to variant 1. The resulting protein (isoform 2) has a shorter and distinct C-terminus when it is compared to isoform 1. Variants 2 and 3 encode the same protein.

Source sequence(s): [BC048308](#), [BC067765](#), [BJ560439](#)

Consensus CDS: [CCDS34879.1](#)

UniProtKB/Swiss-Prot: [Q94905](#)

Related: [ENSP00000335220](#), [OTTHUMP00000225550](#), [ENST00000335171](#), [OTTHUMT00000376714](#)

Conserved Domains (1) [summary](#)

c102525	Band_7; The band 7 domain of flotillin (reggie) like proteins. This group contains proteins similar to stomatin, prohibitin, flotillin, Hfl/KC and podocin. Many of these band 7 domain-containing proteins are lipid raft-associated. Individual proteins of this ...
Location: 24 - 141	
Blast Score: 633	

2. **NM_001003791.2 → NP_001003791.1 erlin-2 isoform 2**
Status: VALIDATED

Description: Transcript Variant: This variant (3) uses a different segment for its 3' coding region and 3' UTR, compared to variant 1. The resulting protein (isoform 2) has a shorter and distinct C-terminus when it is compared to isoform 1. Variants 2 and 3 encode the same protein.

Source sequence(s): [BC048308](#), [BC067765](#), [BP353279](#)

Consensus CDS: [CCDS34879.1](#)

UniProtKB/Swiss-Prot: [Q94905](#)

Related: [ENSP00000380405](#), [OTTHUMP00000225546](#), [ENST00000397228](#)

OMIM Entry - *611605 - ENDOPLASMIC RETICULUM LIPID RAFT-ASSOCIATED PROTEIN 2; ERLIN2

http://omim.org/entry/611605

NCBI: OMIM

Search OMIM

Advanced Search: OMIM, Clinical Synopses, OMIM Gene Map

Sort by: Relevance Date updated

*611605

ENDOPLASMIC RETICULUM LIPID RAFT-ASSOCIATED PROTEIN 2; ERLIN2

Alternative titles; symbols
 SPFH DOMAIN-CONTAINING PROTEIN 2; SPFH2
 CHROMOSOME 8 OPEN READING FRAME 2; C8ORF2

HGNC Approved Gene Symbol: **ERLIN2**

Cytogenetic location: **8p11.23** Genomic coordinates (GRCh37): **8:37,594,096 - 37,615,318** (from NCBI)

Gene Phenotype Relationships

Location	Phenotype	Phenotype MIM number
8p11.23	Spastic paraplegia-18	611225

TEXT

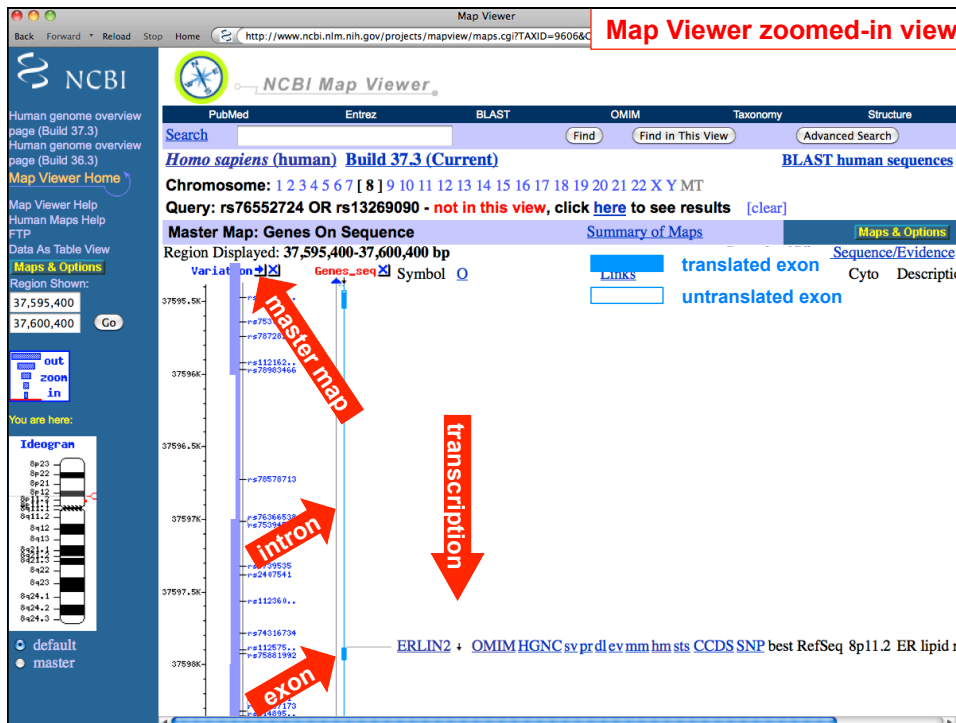
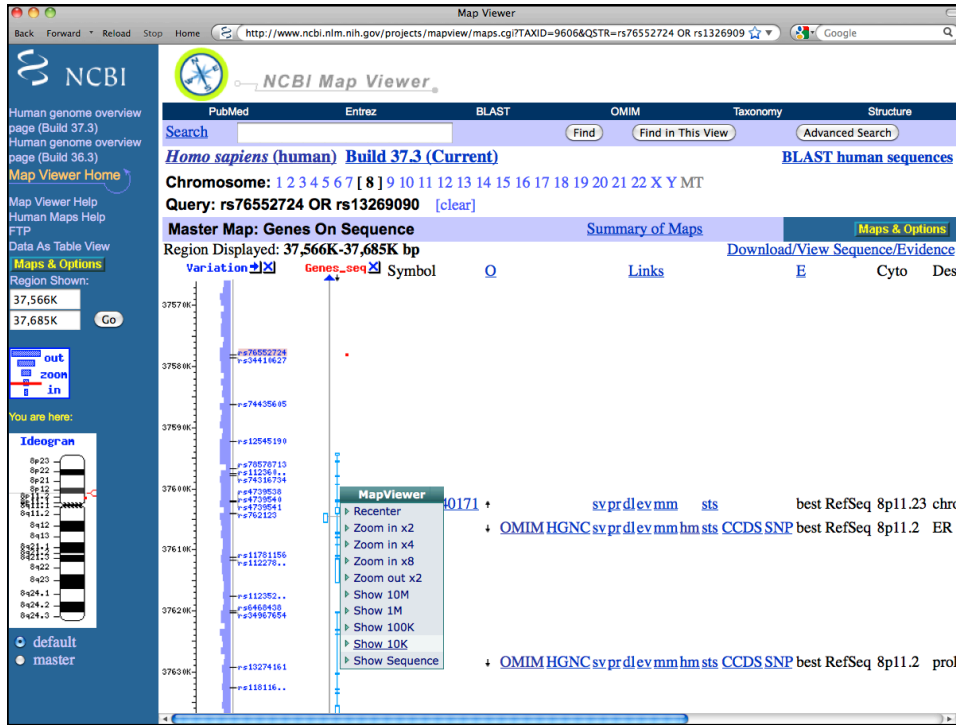
Cloning

By genomic sequence analysis, followed by PCR and RACE of adult and fetal cDNA libraries, [Ikegawa et al. \(1999\)](#) cloned 2 splice variants of ERLIN2, which they designated C8ORF2. The deduced 339- and 152-amino acid proteins share the first 141 N-terminal amino acids, then diverge. Both proteins have an N-glycosylation site and type-2 membrane topology, and the longer protein has a lysine- and glutamic acid-rich region. Northern blot analysis detected ubiquitous expression of 1.6- and 2.5-kb transcripts; a minor 4.4-kb transcript was also observed.

Using monoclonal antibodies to human lipid raft proteins, [Browman et al. \(2006\)](#) identified ERLIN1 (611604) and ERLIN2 as components of lipid rafts. Immunohistochemical analysis of endogenous and fluorescence-tagged proteins revealed that ERLIN1 and ERLIN2 localized specifically to the endoplasmic reticulum (ER) and nuclear envelope. The 2 proteins share 83% identity, and both contain a conserved prohibitin (PHB; 176705) homology domain of about 160 amino acids.

The screenshot shows the NCBI HomoloGene web interface. At the top, there is a search bar with 'HomoloGene' entered and a 'Go' button. Below the search bar, there are tabs for 'Limits', 'Preview/Index', 'History', 'Clipboard', and 'Details'. The 'Preview/Index' tab is selected. The main content area displays search results for 'HomoloGene:5193. Gene conserved in Bilateria'. It is divided into two columns: 'Genes' and 'Proteins'. The 'Genes' column lists various genes from different species, such as ERLIN2 from H. sapiens, P. troglodytes, C. lupus, B. taurus, M. musculus, R. norvegicus, G. gallus, and D. rerio, along with a hypothetical protein from C. elegans. The 'Proteins' column lists corresponding protein sequences with their accession numbers and lengths in amino acids (aa). At the bottom of the page, there are links for 'Protein Alignments' and 'Conserved Domains'.

The slide features the NCBI logo in yellow and white. Below the logo, the text reads 'Zoom in to view greater detail' in white. Underneath that, the URL 'http://www.ncbi.nlm.nih.gov/mapview' is displayed in yellow. At the bottom of the slide, there is a decorative graphic of a DNA double helix and the text 'NATIONAL HUMAN GENOME RESEARCH INSTITUTE' and 'Division of Intramural Research' in white.



Map Viewer SNP map

Map Viewer SNP map

NCBI Map Viewer

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: rs7652724 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

Genes_seq	Variation	Map	Gene	Het	Validation	Genotypes Avail	Linkout	Avail
	rs117005490	L	T	C	Inte			
	rs75303006	L	T	C	Inte			
	rs78728254	L	T	C	Inte			
	rs112162854	L	T	C	Inte			
	rs78983466	L	T	C	Inte			
	rs78578713	L	T	C	Inte			
	rs76366538	L	T	C	Inte			
	rs75394547	L	T	C	Inte			
	rs4739535	L	T	C	Inte			
	rs2407541	L	T	C	Inte			
	rs112360090	L	T	C	Inte			
	rs74316734	L	T	C	Inte			
	rs112575270	L	T	C	Inte			
	rs75881992	L	T	C	Inte			
	rs2186291	L	T	C	Inte			
	rs74657173	L	T	C	Inte			
	rs114895254	L	T	C	Inte			
	rs2154451	L	T	C	Inte			

**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

Reference SNP(refSNP) Cluster Report: rs75881992

RefSNP	Allele	HGVS Names
Organism: human (<i>Homo sapiens</i>)	Variation Class: SNV: single nucleotide variation	NC_000008.10:g.37597929A>G
Molecule Type: Genomic	RefSNP Alleles: A/G	NM_001003790.2:c.154A>G
Created/Updated in build: 131/135	Allele Origin:	NM_001003791.2:c.154A>G
Map to Genome Build: 37.3	Ancestral Allele: A	NP_001003790.1:p.Met52Val
Validation Status:	Clinical Source: unknown	NP_001003791.1:p.Met52Val
	Clinical Significance: NA	NP_009106.1:p.Met52Val
	MAF/MinorAlleleCount: G=0.001/3	
	MAF Source: 1000 Genomes	

SNP Details are organized in the following sections:
[GeneView](#) [Map](#) [Submission](#) [Fasta](#) [Resource](#) [Diversity](#) [Validation](#)

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: 38M-38M (3.0Kbs+) Search & Go:

37,596 K 37,596,500 37,597 K 37,597,500 rs75881992

SNP

Genes

ERLIN2/NM_007175.6/NP_009106.1/NM_001003790.2

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>



NATIONAL HUMAN GENOME RESEARCH INSTITUTE
Division of Intramural Research

Current Protocols in Bioinformatics

The UCSC Genome Browser

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

ABSTRACT

The University of California Santa Cruz (UCSC) Genome Browser is a Web-based tool for quickly displaying a requested portion of a genome, accompanied by a series of aligned annotation "tracks." The annotation tracks are provided by the UCSC Genome Bioinformatics Group and external collaborators—genomic features, mRNA and expressed sequence tag alignments, simple nucleotide sequence, and regulatory data, phenotype and variation data, and pairwise comparative genomics data. All information relevant to a genomic region is displayed in one window, facilitating biological analysis and interpretation. Underlying the Genome Browser tracks can be viewed, downloaded, and used in another Web-based application, the UCSC Table Browser. U

UNIT 1.4

Using the NCBI Map Viewer to Browse Genomic Sequence Data

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¹Bethesda, 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-

UNIT 1.5

Using the Ensembl Genome Server to Browse Genomic Sequence Data

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

UNIT 1.15

links from the Map Viewer. The Alternate Protocols describe how to view the genome sequence, and also illustrate additional protocols. Protocol 1 shows how to perform and interpret the human genome. Alternate Protocol 2 demonstrates how to view two STS markers. Finally, Alternate Protocol 3 describes how to view members of a gene family. *Curr. Protoc. Bioinform.* 30:1.15.1-1.15.48. © 2010 by John Wiley & Sons, Inc.

Keywords: genome assembly • genomic sequence • gene map

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

Access from NIH at

<http://onlinelibrary.wiley.com/book/10.1002/0471250953>