

UiO **Content of Biosciences**

University of Oslo

MBV4410/9410 Fall 2016 Bioinformatics for Molecular Biology





Use Unix locally

Windows: Cygwin (<u>https://www.cygwin.com/</u>)

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The /etc/group (and possibly /etc/passwd) files should be rebuilt. See the man pages for mkpasswd and mkgroup then, for example, run	
mkpasswd -1 [-d] > /etc/passwd mkgroup -1 [-d] > /etc/group	
Note that the -d switch is necessary for domain users.	
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Use Unix locally

Mac: Terminal, iTerm2 (<u>https://www.iterm2.com/</u>)

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Ensembl genome browser and database

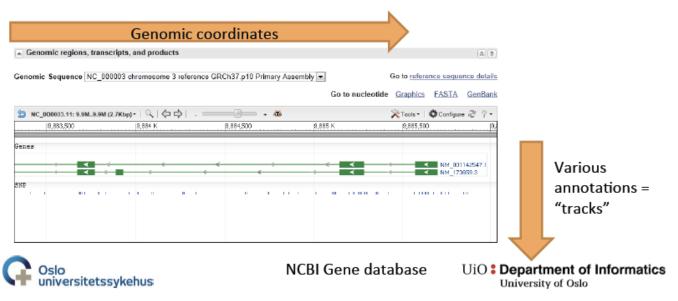


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Genome browsers

- · Graphical interface for genomic data
- Shows information from biological databases mapped onto genomic sequence



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Ensembl Genome Browser

- Joint project between EMBL-EBI and the Wellcome Trust Sanger Institute
- Central resource for studying genomes of vertebrates
 - Mainly chordates, but some few extra (e.g. C. elegans and S. cerevisiae)
 - Updated several times a year with new genome assemblies and new species
 - Annotations of genomes (*e.g.* genes and their splice variant, SNPs) added by the Ensembl pipeline
 - Automatic gene prediction (with or without experimental evidence) & some curator input



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Ensembl Genome Browser

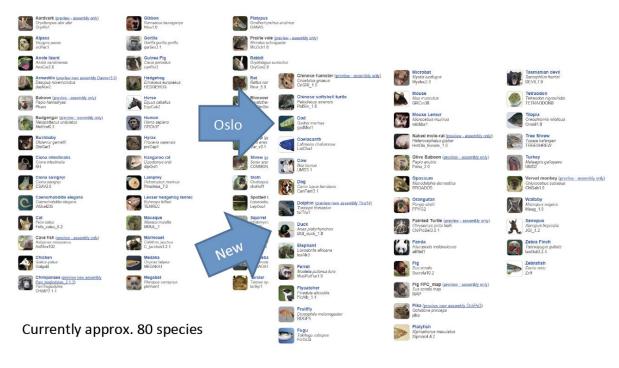
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Ensembl receives major funding from the We	illcome Trust. Our acknowledgen	nents page includes a list of addition	onal current and previous funding bodies.				
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Ensembl Genome Browser



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EnsemblGenomes

ent Species Working with communities FAQs	<u>@</u> ∙						
ant species working with communities FAUs	Bactena	Protists Fungi Plants Metazoa Vertebra					
Ensembl Genomes: Extending Ensembl across the taxonomic space.	- Agricultural-Omics Training Course						
Assembly Mapping	Register now for an upcoming EBI training course in Agricultural-Omics. For more details, please go to http://www.ebi.ac.uk/training/course/agricultural-omics posted 2013-10-03						
For genomes where Ensembl Genomes has provided older assembly versions in the past, assembly mappings are now available. These can be accessed using the Perl API or REST service, or via the assembly converter in the web interface. This is available for plants, metazoa, fungi and protists.	What's New in Release 20 (September 2013)						
Rice ganoma updated	The twentieth release of Ensembl Genomes features updates to version 73 of the Ensembl software across all divisions, and a number of new genomes added bringing the total number of genomes to 9225 (full	Have a question? Frequently Asked Questions (FAQs) are now available for all					
Improved resources for wheat genomes	list). Detailed notes can be found here. See the individual homepages for more information.	domains of Ensembl Genomes. Have a question? Check if it's been asked before! If there is a					
Four aquatic motazoan genomes	Ensembl Bacteria Ensembl Bacteria has been updated to include the latest versions	FAQ missing, contact us.					
CREST Ensembl Genomes REST Service	of 9.009 genomes (8.842 eubacteria and 247 archaea) from the INSDC archives. Cross-references to Fihea and MetaCyc have also been added, as have Enzyme Commission classifications. In addition, data from RegulonDE have been used to add operon and other regulatory features to E. coll K-12 MG1655.						
sembl Genemos is developed by EMBL-EBI and is powered by Ensembl software system for the analysis and utilization of genemic data. For datalis of our funding please click here.	Ensembl Fungi Two new plant pathogen genomes, Microbotryum violaceum and Blumene gramine. Cross-references to PHI- base wore added for plant pathogene.						
	Ensembl Metazoa Three metazoan species have updated assemblies and gene models in rr aphid, the western honey bee, and the purple sea urchin. The variation do updated to include ~7.5 million additional variants.						
	Ensembl Plants						
	The first assembly of the bread wheat genome. Triticum aestivum, from the IWGSC has been added in this						

• Bacteria, protists, fungi, plants and other metazoa

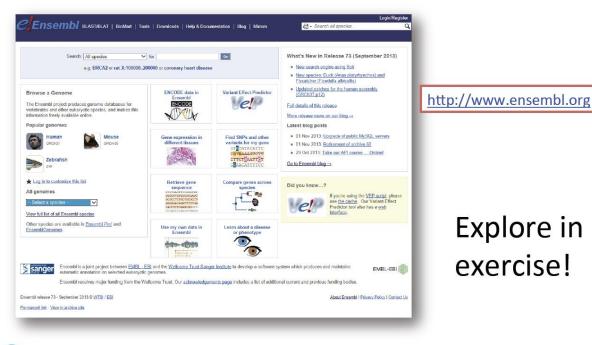
release. In addition we have loaded the latest assembly for Oryza sativa from IRGSP.



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Ensembl Genome Browser very brief demo

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UCSC Genome Browser



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UCSC Genome Browser

- Developed and maintained at the University of California, Santa Cruz (UCSC)
- Interactive website
- Access to genome sequence data from
 - Human genome
 - Latest assembly (GRCh38), the 2nd latest (GRCh37), but also earlier versions
 - Mouse, rat, and approx. 50 other mammals
 - Chicken, turkey, budgerigar, reptiles, frogs, and fishes
 - Insects, nematodes, S. cerevisiae and more
 - In total 91 species in 2014



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UCSC Genome Browser

BRIEFINGS IN BIOINFORMATICS. VOL 14. NO 2. 144–161 Advance Access published on 20 August 2012 doi:10.1093/bib/bbs038

The UCSC genome browser and associated tools

Robert M. Kuhn, David Haussler and W. James Kent

Submitted: 8th February 2012; Received (in revised form): 9th June 2012

Abstract

The UCSC Genome Browser (http://genome.ucsc.edu) is a graphical viewer for genomic data now in its 13th year. Since the early days of the Human Genome Project, it has presented an integrated view of genomic data of many kinds. Now home to assemblies for 58 organisms, the Browser presents visualization of annotations mapped to genomic coordinates. The ability to juxtapose annotations of many types facilitates inquiry-driven data mining. Gene predictions, mRNA alignments, epigenomic data from the ENCODE project, conservation scores from vertebrate whole-genome alignments and variation data may be viewed at any scale from a single base to an entire chromosome. The Browser also includes many other widely used tools, including BLAT, which is useful for alignments from high-throughput sequencing experiments. Private data uploaded as Custom Tracks and Data Hubs in many formats may be displayed alongside the rich compendium of precomputed data in the UCSC database. The Table Browser is a full-featured graphical interface, which allows querying, filtering and intersection of data tables. The Saved Session feature allows users to store and share customized views, enhancing the utility of the system for organizing multiple trains of thought. Binary Alignment/Map (BAM), Variant Call Format and the Personal Genome Single Nucleotide Polymorphisms (SNPs) data formats are useful for visualizing a large sequencing experiment (whole-genome or whole-exome), where the differences between the data set and the reference assembly may be displayed graphically. Support for high-throughput sequencing extends to compact, indexed data formats, such as BAM, bigBed and bigWig, allowing rapid visualization of large datasets from RNA-seq and ChIP-seq experiments via local hosting.



http://genome.ucsc.edu

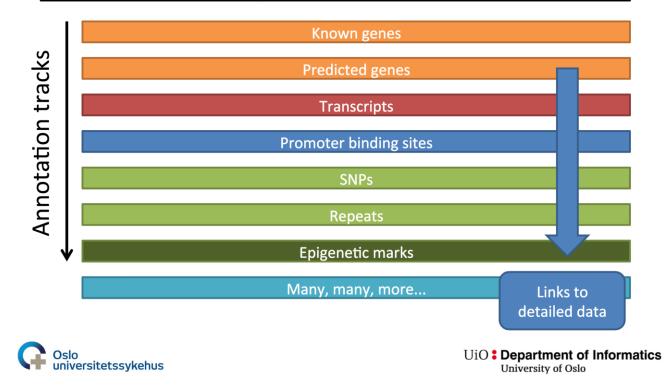


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UCSC Genome Browser

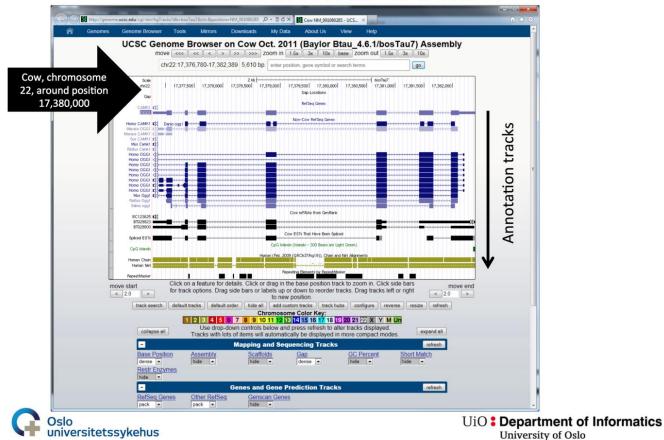
Reference genome, chromosome coordinates



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UCSC Genome Browser



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UCSC Genome Browser brief demo

http://genome.ucsc.edu/cgi-bin/hgTrac ,D - 🗄 C × 🔛 Human chr3:9,790,701-9,79... × Genomes Genome Browser Tools Mirrors My Data About Us UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly move <<< < < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x chr3 9,790,701-9,799,453 8,753 bp. qo ches (227.3) 🕴 202422 2024 20 227.31 (4.254.4.013)12.3 (227.3) Refleg Gene Reffeq Gene Mumiro amin Spliced ESTs TON ENCODE DNase Cluster: similar since o Farter Chit-see from ENCODE tin Factor Chil 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 < 2.0 > labels up or down to reorder tracks. Drag tracks left or right to new position. < 2.0 > track search default tracks default order hide all add custom tracks track hubs configure reverse resize Use drop-down controls below and press refresh to alter tracks displayed collapse all expand all Tracks with lots of items will automatically be displayed in more compact mode refresh Mapping and Sequencing Tracks B deCODE Recomb STS Markers FISH Clones Chromosome Band dense . hide hide hide hide 💌 hide -GRC Map Contigs Gap Publications ENCODE Pilot Map Contigs Assembly



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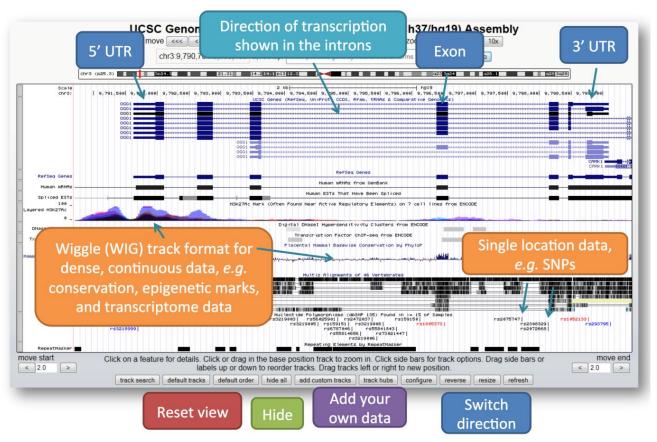
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Different kinds of data

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Help to investigate correct splicing?

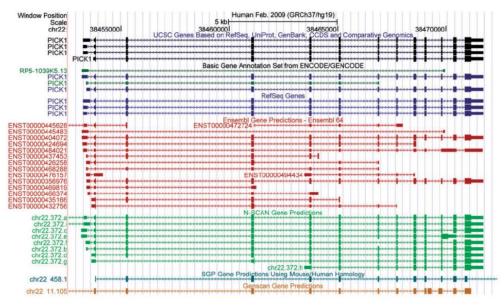


Figure 1: Screenshot of UCSC Genome Browser displaying human PICKI gene region on chr22 in hgl9 assembly. Different gene prediction algorithms predict different annotations in the region. By presenting multiple data sets of similar type, the user is able to more easily evaluate hypotheses. The different tracks often predict different 3'- and 5'-untranslated regions (half-height boxes on ends of annotations), coding regions (fullheight boxes), introns (thin line with transcription-direction arrows) or start and end coordinates. The differences may be used to establish a level of confidence in an annotation not obtained from any single method.



Kuhn et al. Brief. Bioinform. 14, 144 (2012) UiO : Department of Informatics University of Oslo

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ENCODE data in UCSC

Published online 30 October 2010

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ENCODE whole-genome data in the UCSC genome browser (2011 update)

Brian J. Raney^{1,*}, Melissa S. Cline¹, Kate R. Rosenbloom¹, Timothy R. Dreszer¹, Katrina Learned¹, Galt P. Barber¹, Laurence R. Meyer¹, Cricket A. Sloan¹, Venkat S. Mallad¹, Krishna M. Roskin¹, Bernard B. Suh¹, Angie S. Hinrichs¹, Hiram Clawson¹, Ann S. Zweig¹, Vanessa Kirkup¹, Pauline A. Fujita¹, Brooke Rhead¹, Kayla E. Smith¹, Andy Pohl¹, Robert M. Kuhn¹, Donna Karolchik¹, David Haussler^{1,2} and W. James Kent¹

¹Center for Biomolecular Science and Engineering, School of Engineering and ²Howard Hughes Medical Institute, University of California Santa Cruz (UCSC), Santa Cruz, CA 95064, USA

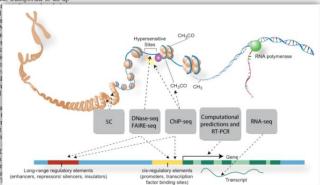
Received September 15, 2010; Accepted October 9, 2010

ABSTRACT

The ENCODE project is an international consortium with a goal of cataloguing all the functional elements in the human genome. The ENCODE Data Coordination Center (DCC) at the University of California, Santa Cruz serves as the central repository for ENCODE data. In this role, the DCC offers a collection of high-throughput, genome-wide data generated with technologies such as ChIP-Seq, RNA-Seq, DNA digestion and others. This data helps illuminate transcription factor-binding sites. histone marks, chromatin accessibility, DNA methylation, RNA expression, RNA binding and other cell-state indicators. It includes sequences with quality scores, alignments, signals calculated from the alignments, and in most cases, element or peak calls calculated from the signal data. Each data set is available for visualization and download via the UCSC Genome Browser (http://genome.ucsc.edu/). ENCODE data can also be retrieved using a metadata system that captures the experimental parameters of each assay. The ENCODE web portal at UCSC (http://encodeproject.org/) provides information about the ENCODE data and links for access.

into RNA, which might be spliced, transported to an appropriate cellular compartment proteins. This process is regulated DNA methylation, chromatin transcription factors to the DI factors to the RNA and RNA trai itable traits are determined as m regulation as differences in gene of The goal of the ENCODE proj tional elements in the human ge processes, through direct measur genomic technologies and detail ENCODE began with a pilot ph of the genome (3), and scaled analysis in 2007 The role of the ENCODE Da

The role of the ENCODE Da (DCC) is to organize and display) labs in the consortium, and to en specific quality standards when it Before a lab submits any data, th a data agreement that defines the and associated metadata. The I data to ensure consistency with loads the data onto a test server tion, and coordinates with the la into a consistent set of tracks. W the DCC Quality Assurance team performers servers or un-



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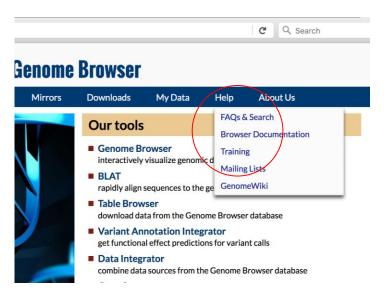
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http://genome.ucsc.edu/ENCODE/aboutScaleup.html

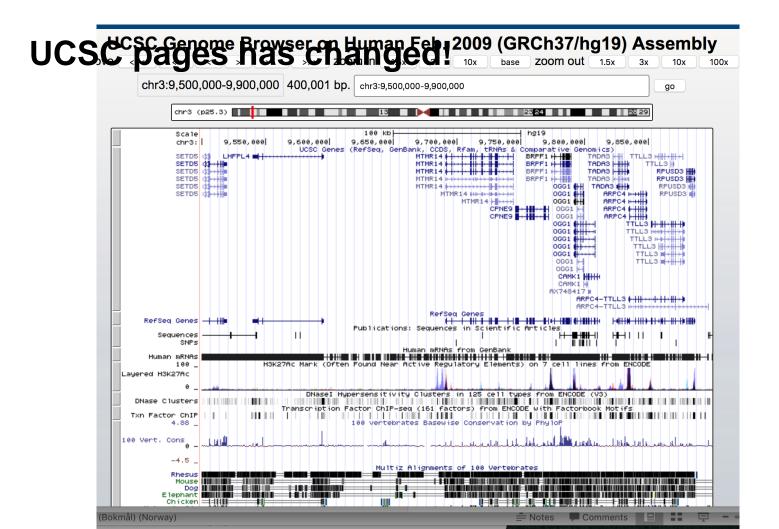
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UCSC pages has changed!





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Data las	t updated: 20	13-06-14	1			1	·				

Sequence and Links to Tools and Databases

Genomic Se	quence (chr3:9,7	745,510-9,771,592)	mRNA (may diffe	er from genome)	Protein (553 aa)
Gene Sorter	Genome Brows	er Protein FASTA	Table Schema	BioGPS	CGAP
Ensembl	Entrez Gene	ExonPrimer	GeneCards	GeneNetwork	Gepis Tissue
HGNC	HPRD	Lynx	MGI	MOPED	neXtProt
PubMed	Stanford SOUR	CE Treefam	UniProtKB		