MEDS5420 Lec13

Using the UCSC Genome Browser

March 1, 2023

Lead creators of UCSC genome browser: Jim Kent and David Haussler



Jim Kent's Web Page

I'm a research scientist at UCSC. I work primarily on web tools to help understand the human genome. Before becoming a bioinformatician I got a PhD in biology working with with the Zahler lab. Before that I wrote computer art and animation programs. I live in Santa Cruz, CA and have three children, Mira, Tisa, and Maia.

email Jim

Links to Stuff I Work On:

- · The UCSC Genome Browser also known as 'the golden path'.
- Parasol A Job Control System for Computer Clusters. It's fast and it's free.
- Papers PubMed index of my papers.
- <u>Presentations</u> slides from presentations. Sadly no voice-over.
 <u>The Intronerator</u> to look at C. elegans genes and splicing patterns.
- cis-Site Seeker Look for regulatory regions in RNA or DNA sequences with the Improbizer.
- Cross Species Alignments Program and some samples using the not yet famous WABA algorithm.
- Commands Catalog of command line driven programs.
- Source Code free for academic, non-profit or personal use.
- Executables commonly requested compiled versions also free for academic, non-profit and personal use.

Other Useful Links

- · PubMed to search the scientific literature.
- <u>Blast</u> homology searching.
- <u>Google Scholar</u> search scientific literature the Google way.
 <u>UCSC Schedule of Classes</u> er, when was that final?

https://cbse.soe.ucsc.edu/people/kent https://users.soe.ucsc.edu/~kent/

https://hausslergenomics.ucsc.edu/people/ david-haussler/

Lead creator of UCSC genome browser: Jim Kent

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Jim Kent



Director, UCSC Genome Browser Project

Research Scientist, Department of Biomolecular Engineering, UC

Jim Kent

Research Projects

Jim Kent directs the genome br assurance staff of the UCSC Ge created the computer program i of the human genome sequence sequencing centers worldwide a informatics associated with the Bioinformatics Group participate produce, assemble, and annota

UCSC Genome Browser

The UCSC Genome Browser primetazoan genome sequences. I wide annotation in a web-based

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RESEARCH TALKS

David Haussler is Scientific D California Santa Cruz (UCSC) Investigator of the Howard He (HHMI). Haussler uses mathe biology to study the genomes

understanding disease and evolution. As part of the Human Genor that published the first publicly available draft of the human genor several large-scale projects, including the Genome 10K Project, th Hub (CGHub), and the Global Alliance for Genomics and Health.

https://www.ibiology.org/speakers/davidhaussler/

Some useful capabilities of the browser

- Some resources:
 - Genome build files (fasta format)
 - Converting genome coordinates between versions (e.g. hg19 to hg38)
 - Gene annotation files
 - Convert gene IDs from different sources (GENCODE-refSeq)
 - Obtaining DNA sequences from regions of interest
 - In-silico tools
 - BLAT- fast alignment of short sequences
 - PCR
- Custom tracks and resources:
 - Uploading custom data or annotations
 - Storing them in a user profile
 - Sharing / viewing track hubs
 - Configure Display for optimal clarity (e.g. for making figures)

UCSC homepage



More tools...

Click on Genomes



More tools...

Available genomes and info



Click on the Genome Browser



Browser view



Browser view



Downloading Genomes and source code for utilities and browser



The source for the Genome Browser, Blat, liftOver and other utilities is free for non-profit academic research and for personal use. For information on commercial licensing, see the Genome Browser and Blat licensing requirements. The source and executables for several of these products can be downloaded or purchased from our online store.

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Mirroring the Genome Browser

Precompiled executable binaries are available for installing a local mirrored copy of the Genome Browser website on your web server, eliminating the need to compile the entire source tree.

- Mirror instructions
- Build instructions
- Genome Browser hgcentral tables
- Genome Browser source code downloads

If you encounter difficulties with slow download speeds, try using UDT Enabled Rsync (UDR), which improves the throughput of large data transfers over long distances. The 32-bit and 64-bit versions can be downloaded here.

Utilities

The utilities directory offers downloads of pre-compiled standalone binaries for:

LiftOver (which may also be accessed via the web version). The over.chain liftOver conversion files are located in the individual assembly download sections.

- Blat
- Other command-line utilities

All our command line tools can be obtained as pre-built binaries from our downloads server. The following command can be used to copy all command line utilities into a directory with the correct permission bits set:

\$ rsync -aP hgdownload.soe.ucsc.edu::genome/admin/exe/linux.x86_64/ ./

Note about 'permission denied' error when downloading with curl or wget:

In order for your computer to run a freshly downloaded utility, you will need to update the file system permissions to allow your operating system to run the program. To make utilities usable, download a tool and turn on its 'executable' bit:

\$ wget https://hgdownload.cse.ucsc.edu/admin/exe/linux.x86_64/liftOver

\$ chmod +x ./filePath/utility_name

Downloading Genomes

Human genome

Dec. 2013 (GRCh38/hg38)

- Genome sequence files and select annotations (2bit, GTF, GC-content, etc) ▼
 - Standard genome sequence files and select annotations (2bit, GTF, GC-content, etc)
 - Analysis set sequence files (See: What is the analysis set?)
- Sequence data by chromosome
- Annotations
 - SQL table dump annotations
 - Fileserver (bigBed, maf, fa, etc) annotations Also see Data Access
- SNP-masked fasta files
- LiftOver files
- Pairwise alignments
- Multiple alignments
- Patches
- Data archive

Downloading Genomes and more

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Introduction

The Dec. 2013 assembly of the human genome (GRCh38 Genome Reference Consortium Human Reference 38), is called hg38 at UCSC. This directory contains the genome as released by UCSC, selected annotation files and updates. The directory "genes/" contains GTF/GFF files for the main gene transcript sets.

For more information about this assembly, see these NCBI resources: http://www.ncbi.nlm.nih.gov/genome/51

- http://www.ncbi.nlm.nih.gov/genome/assembly/883148 http://www.ncbi.nlm.nih.gov/bioproject/31257

These files are used by the UCSC Genome Browser for display and analysis. If you want to do analysis and show it later on the browser, it is usually easiest to run your analysis on the UCSC hg38 file. For most users, this will be the file "latest/hg38.fa.gz" in this directory. However, if you need a genome file for alignment or variant calling, please read the section "Analysis set" below.

The sequences of the main chromosomes are identical to the genome files distributed by NCBI and the EBI, but the sequence names are different. For example, the name of chromosome 1 is called "chr1" at UCSC, "NC 000001.11" at NCBI, and "1" at the EBI. Also, the lowercasing in the files is not exactly identical, as UCSC, NCBI and EBI run Repeatmasker with slightly different settings.

The NCBI accession of the UCSC hg38 genome is GCA_000001405.15. The version that includes the updates for patch release 13 GRCh38.pl3 has the NCBI accession GCA_000001405.28.

Analysis set

The GRCh38 assembly contains more than just the chromosome sequences, but also a mitochondrial genome, unplaced seguences, centromeric seguences and alternates. To better capture variation in the human genome across the world it contains more copies of some loci than hg19. Some of these additions, like the EBV genome, are mostly relevant for genomic analysis, i.e. alignment. For an overview of the different types and reasons for the additions see https://software.broadinstitute.org/gatk/documentation/article?id=11010

This means that if you want to use the genome sequence for alignment and especially for variant calling, you should use the optimal genome file for your aligner. The genome file can make a big difference, especially for variant calling. In most cases, the authors of your alignment program will provide advice on which hg38 genome version to use and usually they recommend one of the files in our analysisSet/ directory, like the GATK link above. These special genome files sometimes remove the alternate sequences, sometimes they add decoys or change single nucleotides towards the major allele, but they never insert or delete sequences, so the annotation coordinates remain the same.

- for BWA see also https://lh3.github.io/2017/11/13/which-human-reference-genome-to-use

- for Novoalign see its manual at http://www.novocraft.com/userfiles/file/Novocraft.pdf - For Bowtie, see the different versions of the human genome that the Bowtie authors

provide: http://bowtie-bio.sourceforge.net/index.shtml

Also see analysisSet/README.txt for further details

Patches

Like hg19, hg38 has been updated with patches since its release in 2013. GRC patch releases do not change any previously existing sequences; they simply add small, new sequences for fix patches or alternate haplotypes that correspond to specific regions of the main chromosome sequences (see below). For most users, the patches are unlikely to make a difference and may complicate the analysis as they introduce more duplication. If you want a version of the genome without these complexities, look at the analysisSet/ subdirectory.

The initial/ subdirectory contains files for the initial release of GRCh38, which includes the original alternate sequences (261) and no fix sequences.

The pll/ subdirectory contains files for GRCh38.pll (patch release 11).

The p12/ subdirectory contains files for GRCh38.p12 (patch release 12).

The p13/ subdirectory contains files for GRCh38.p13 (patch release 13).

The "latest/" symbolic link points to the subdirectory for the most recent patch version

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| | Name | Last modifi | <u>ed</u> | <u>Size</u> | Description |
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| uted | Parent Directory | | | _ | |
| | analysisSet/ | 2021-10-07 | 16:48 | - | |
| | est.fa.gz | 2020-09-09 | 11:27 | 1.5G | |
| | est.fa.gz.md5 | 2020-09-09 | 11:27 | 44 | |
| | genes/ | 2021-12-09 | 03:03 | - | |
| | hg38.2bit | 2015-04-30 | 16:16 | 797M | |
| | hg38.agp.gz | 2014-01-15 | 20:55 | 842K | |
| | hg38.chrom.sizes | 2013-12-24 | 21:06 | 11K | |
| | hg38.chromAlias.txt | 2021-10-06 | 13:44 | 27K | |
| | hg38.chromFa.tar.gz | 2014-01-23 | 17:18 | 938M | |
| a | hg38.chromFaMasked.tar.gz | 2014-01-23 | 17:10 | 487M | |
| | <u>hg38.fa.align.gz</u> | 2014-01-08 | 23:43 | 2.4G | |
| | hg38.fa.gz | 2014-01-15 | 21:14 | 938M | |
| | hg38.fa.masked.gz | 2014-01-15 | 21:24 | 487M | |
| | hg38.fa.out.gz | 2014-01-15 | 20:56 | 172M | |
| | hg38.gc5Base.bw | 013-12-24 | 21:28 | 1.6G | |
| | hg38.gc5Base.wib | 2019-01-17 | 14:50 | 591M | |
| | <u>hg38.gc5Base.wig.gz</u> | 2010-01-17 | 14:50 | 11M | |
| · / | hg38.gc5Base.wigVarStep.gz | 2013 12-24 | 21:14 | 1.5G | |
| | hg38.trf.bed.gz | 2014-01-15 | 20:56 | 7.9M | |
| e-to-use | <u>initial/</u> | 2021-10-07 | 16:50 | - | |
| raft.pdf | <u>latest/</u> | 2021-10-7 | 16 : 57 | - | |
| | md5sum.txt | 2021-10-00 | 14:07 | 667 | |
| | <u>mrna.fa.gz</u> | 2020-09-09 | 1:08 | 372M | |
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| | <pre>upstream1000.fa.gz.md5</pre> | 2020-09-09 | 11:29 | 53 | |
| genome | <u>upstream2000.fa.gz</u> | 2020-09-09 | 11:29 | 20M | |
| genome | <pre>upstream2000.fa.gz.md5</pre> | 2020-09-09 | 11:29 | 53 | |
| | <u>upstream5000.fa.gz</u> | 2020-09-09 | 11:30 | 52M | |
| | <pre>upstream5000.fa.gz.md5</pre> | 2020-09-09 | 11:30 | 53 | |
| | <u>xenoMrna.fa.gz</u> | 2020-09-09 | 11:19 | 6.7G | |
| | <pre>xenoMrna.fa.gz.md5</pre> | 2020-09-09 | 11:19 | 49 | |
| | <u>xenoRefMrna.fa.gz</u> | 2020-09-09 | 11:27 | 255M | |
| | <pre>xenoRefMrna.fa.gz.md5</pre> | 2020-09-09 | 11:27 | 52 | |

Descriptions of file content are in the middle

Converting genome coordinates

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Command Line Tool

To lift genome annotations locally on Linux systems, download the liftOver executable and the appropriate chain file. Run liftOver with no arguments to see the usage message.

Downloading annotation tracks: gene lists



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Downloading DNA sequence from browser



Downloading DNA from browser

🕋 Genomes Genome Browser

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Get DNA in Window (hg38/Human)

Get DNA for

Position chr17:8,164,338-8,204,114

Note: This page retrieves genomic DNA for a single region. If you would prefer to get DNA for many items in a particular track, or get DNA with formatting options based on gene structure (introns, exons, UTRs, etc.), try using the <u>Table Browser</u> with the "sequence" output format. You can also use the <u>REST API</u> with the **/getData/sequence** endpoint function to extract sequence data with coordinates.

Sequence Retrieval Region Options:

```
Add o extra bases upstream (5') and o extra downstream (3') Options to get flanking DNA as well
```

Mirrors

Note: if a feature is close to the beginning or end of a chromosome and upstream/downstream bases are added, they may be truncated in order to avoid extending past the edge of the chromosome.

Sequence Formatting Options:

All upper case.

All lower case.

Mask repeats: • to lower case • to N

Options for case and masking of repeats

Reverse complement (get '-' strand sequence)

get DNA extended case/color options

Note: The "Mask repeats" option applies only to "get DNA", not to "extended case/color options".

Fasta output



>hg38 dna range=chr17:8164338-8204114 5'pad=0 3'pad=0 strand=+ repeatMasking=none TGGCTTCCCAGGGCCTCCGGGTGTGAGATCCTCCCCCACTGCAGTGCCCC ACCCGCTCCCCACAGAAGCCCCGGAGAGTGGCTCTGTCACCAGAGGTGTCA TGTGGGAACTCAGCCCAATCTAAGAGAAGATACTCTTGGCTTCCTCCCCC TCAGAGGAGCAGCCGCGTCCCTGGTCCTTGTGCACTGACAGTCCCGGGTG GCTCAGCCTATGTGATGAGAAAGAAAAGAGCAGCTTCCACGGGGGGTCTCA GATCATGCTGTGAGATGCTGGCTCCTGCTTTTTGGTCTTCAAATGATTCC TCTTTCTAGTCAGAGAACAAAGATGCCAAATTCCAGCCTTTTAAGTTCAA AACCCTCAGTGTCTGTCAGGGTATAAATAAGACATGGAGGATTGGAAGAG GCAGGCACAGGAAGCCCTTCCGCCCCAGTTGGACACCCTGCACTAACTG AGGGGAGCCTAGTGCAGAGCCAGACAAGACATTTGATGCTGGTTTGGGGG TGATGTGGAAGGGACAGAGCCCCAGATACACGGTCAAGGCCAGGGGTCGT GCTATTGCTATTGGATCGTGGGGGACGGCAGCAGCAGAGTAGAAATGAGGG CATGTGTGACCCACGGAGATGAAGGGCCCAGGAGGATTCATCTCAGAAAG AGGACTCTCATCCAGCAGCCCCTTCCCACCCCACCCACCTAGGCTGTGG CTCAGAGGAACTCCCCAAATTTAGATCAATGGTGGAGGTGGGAGAAGGAG TGGGATGACGGAGTTCACCTGGAAGTGACTGAGAGAATGTTTTCTGCAGG CAGGTAGAATGGAGACCCAAATAGAGGTAAGTTGAACAATAGAAAAATAA TGTATTTGCATAACTCAGATTTGGCTTCTACATGTACTGGCTTCGGGAAT TGGCCAATGATTTTCTAGGGCACACCCCCAAGACTCATTCAAAGGATGTA CCAGGCTGGAGTGCAATGGCGCGCATCTCTGCTCACTGCAAACTCCGCCTC CCAGGTTCACGCCATTCTCCTCCCTCAGCCTCCCGAGTAGCTGGGACTAC AGGCGCCCACCACCCGGCCCGGCTAATTTTTTGTATTTTAGTAGAGAGACA GGGTTTCACCGTGTTAGCCAGGGTGGTCTTGATCTCCTGACCCCGTGATC

Fast DNA search: BLAT

SANTA CRUZ Genomics Institute

Genome Browser

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Fast DNA search with BLAT

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BLAT on DNA is designed to quickly find sequences of 95% and greater similarity of length 25 bases or more. It may miss more divergent or shorter sequence alignments. It will find perfect sequence matches of 20 bases. BLAT on proteins finds sequences of 80% and greater similarity of length 20 amino acids or more. In practice DNA BLAT works well on primates, and protein BLAT on land vertebrates.

BLAT is not BLAST. DNA BLAT works by keeping an index of the entire genome in memory. The index consists of all overlapping 11-mers stepping by 5 except for those heavily involved in repeats. The index takes up about 2 gigabytes of RAM. RAM can be further reduced to less than 1 GB by increasing step size to 11. The genome itself is not kept in memory, allowing BLAT to deliver high performance on a reasonably priced Linux box. The index is used to find areas of probable homology, which are then loaded into memory for a detailed alignment. Protein BLAT works in a similar manner, except with 4-mers rather than 11-mers. The protein index takes a little more than 2 gigabytes.

BLAT was written by <u>Jim Kent</u>. Like most of Jim's software, interactive use on this web server is free to all. Sources and executables to run batch jobs on your own server are available free for academic, personal, and non-profit purposes. Non-exclusive commercial licenses are also available. See the <u>Kent Informatics</u> website for details.

For more information on the graphical version of BLAT, click the Help button on the top menu bar or see the Genome Browser FAQ.

Kent WJ. BLAT - the BLAST-like alignment tool. Genome Res. 2002 Apr;12(4):656-64. PMID: 11932250

BLAT Search Results Go back to ch Find Sta B DINA csearch with BLAT

Custom track name: blat hg38_dna

Custom track description: blat on hg38_dna

Build a custom track with these results

move start

< 2.0

| ACTIONS | QUERY | SCORE | START | END | QSIZE | IDENTITY | CHROM | STI | RAND START | END | SPAN |
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| browser details | dna | 400 | 1 | 400 | 400 | 100.0% | chr17 | + | 8164338 | 8164737 | 400 |
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| browser details | hg38_dna | 25 | 349 | 379 | 400 | 88.9% | chr1 | + | 247443829 | 247443858 | 30 |
| browser details | hg38_dna | 25 | 15 | 49 | 400 | 89.3% | chr1 | + | 204244803 | 204244836 | 34 |
| browser details | hg38_dna | 23 | 34 | 58 | 400 | 96.0% | chr14 | - | 20262748 | 20262772 | 25 |
| browser details | hg38_dna | 23 | 321 | 344 | 400 | 100.0% | chr1 | - | 59157007 | 59157032 | 26 |
| browser details | hg38_dna | 23 | 42 | 65 | 400 | 100.0% | chr14 | + | 72549910 | 72549939 | 30 |
| browser details | hg38_dna | 22 | 34 | 58 | 400 | 96.0% | chr6 | - | 166516416 | 166516441 | 26 |
| browser details | hq38_dna | 22 | 32 | 56 | 400 | 95.98 | chr13 | + | 51135725 | 51135753 | 29 |
| browser details | ng38_dna | 21 | 32 | 52 | 400 | 100.0% | chr10 | - | 5966101 | 5966121 | 21 |
| browser details | hg38_dna | 20 | 26 | 47 | 400 | 95.5% | chr18 | - | 44760280 | 44760301 | 22 |
| browser details | hg38_dna | 20 | 367 | 386 | 400 | 100.0% | chr15 | + | 93348003 | 93348022 | 20 |
| browser details | hg38_dna | 20 | 360 | 381 | 400 | 95.5% | chr11 | + | Alignment of hg38 dn | a and chr17.8164338-81 | 64737 |
| browser details | hg38_dna | 20 | 32 | 51 | 400 | 100.0% | chr1 | + | Click on links in the frame to the le | ft to navigate through the alignment M | atching bases in cDNA and gen |
| browser details | hg38_dna | 20 | 196 | 215 | 400 | 100.0% | chr1 | + | mark the boundaries of gaps in eith | er sequence (often splice sites). | and and outer in other and gen |
| browser details | hg38 dna | 20 | 32 | 51 | 400 | 100.0% | chr1 | + | cDNA hg38_dna | | |

Help ligging Tools UCSC Genome Browser on Human Dec. 2013 (GRCh38/hg38) Assembly move <<< < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x multi-region chr17:8,164,338-8,164,737 400 bp. gene, chro go chr17 (p13.1) p13.3513.217 13.1 17p12 17p11.2 17q11.2 17q12 21.2 q21.31 17a22 23.2 24.2 a24.3 a25.1 17a25.3 Scale chr17: 8,164,400 8,164,450 8,164,500 8,164,550 8,164,600 8,164,650 8,164,700 GENCODE V38 Repeating Elements by RepeatMasker LINE LTR DNA Satellite RNA Other Click on a feature for details. Click+shift+drag to zoom in. Click side bars for track options. Drag side bars or labels up or down to move end reorder tracks. Drag tracks left or right to new position. Press "?" for keyboard shortcuts. < 2.0 > track search default tracks default order hide all add custom tracks track hubs co Use drop-down controls below and press refresh to alter tracks displayed. collapse al expand all Tracks with lots of items will automatically be displayed in more compact modes.

| 50 | TCCCCCACCC | GCTTCCTCCC | ACAGGAACCA | TCTAAGACAA | CTTGATC |
|-----|-------------|------------|------------|------------|----------|
| 100 | GCAGTGCCCC | CTCCCCCACT | GTGTGAGATC | GGGCCTCCGG | SCTTCCCA |
| 150 | AGAGGTGTCA | CTCTGTCACC | CGGAGAGTGG | CACAGAAGCC | CCGCTCCC |
| 200 | GTGTGCTGGG | CAGGGAATGT | GGTGAGTGAG | GTCTGTGGGA | PCCCAGCT |
| 250 | TTCCTCCCCC | TACTCTTGGC | TAAGAGAAGA | CAGCCCAATC | FGGGAACT |
| 300 | GTCCCGGGTG | TGCACTGACA | CTGGTCCTTG | AGCCGCGTCC | AGAGGAGC |
| 350 | GGGGGGTCTCA | CAGCTTCCAC | AAGAAAAGAG | TGTGATGAGA | PCAGCCTA |
| 400 | AAATGATTCC | TTTGGTCTTC | GCTCCTGCTT | TGAGATGCTG | CATGCTG |
| | | | | | |

Genomic chr17 :

| aaagcaagc | ctcaagggtg | caggaggcat | tttaggegag | gecetttgge | 8164287 | |
|-----------|------------|------------|------------|------------|---------|--|
| ctggggtgg | gggtgcttac | tacatccctg | gaaggtgcag | ggaggetgat | 8164337 | |
| GCCTTGATC | TCTAAGACAA | ACAGGAACCA | GCTTCCTCCC | TCCCCCACCC | 8164387 | |
| GGCTTCCCA | GGGCCTCCGG | GTGTGAGATC | CTCCCCCACT | GCAGTGCCCC | 8164437 | |
| CCCGCTCCC | CACAGAAGCC | CGGAGAGTGG | CTCTGTCACC | AGAGGTGTCA | 8164487 | |
| TTCCCAGCT | GTCTGTGGGA | GGTGAGTGAG | CAGGGAATGT | GTGTGCTGGG | 8164537 | |
| GTGGGAACT | CAGCCCAATC | TAAGAGAAGA | TACTCTTGGC | TTCCTCCCCC | 8164587 | |
| CAGAGGAGC | AGCCGCGTCC | CTGGTCCTTG | TGCACTGACA | GTCCCGGGTG | 8164637 | |
| CTCAGCCTA | TGTGATGAGA | AAGAAAAGAG | CAGCTTCCAC | GGGGGTCTCA | 8164687 | |
| ATCATGCTG | TGAGATGCTG | GCTCCTGCTT | TTTGGTCTTC | AAATGATTCC | 8164737 | |
| ctttctagt | cagagaacaa | agatgccaaa | ttccagcctt | ttaagttcaa | 8164787 | |
| accetcagt | gtctgtcagg | gtataaataa | gacatggagg | attggaagag | 8164837 | |
| | | | | | | |

Cido by Cido Allemmont

Your Sequence From Blat Search" Matched region shown as track in browser

are colored blue and capitalized. Light

Custom tracks

| L | JCSC Gen move <<< | ome Brow | ser on H | om in 1.5x 3 | c. 2013 (C x 10x base 2 | SRCh38/h | g38) As | sembly | |
|--|-------------------------------------|--|----------------------------------|--|---|------------------------|--------------|--------------------|---------------|
| | multi-region chr1 | 7:8,164,338-8,16 | 54,737 400 b | p. gene, chromos | ome range, or other p | position, see examp | es | go <u>examples</u> | |
| chr17 (p13.1) | 013.3p13.2 <mark>17p</mark> 13.1 | 17p12 17p11.2 | 17q1 | 1.2 17q12 | 21.2 q21.31 | 17q22 | 23.2 24. | 2 q24.3 q25.1 | 17q25.3 |
| Scale chr17: | 8,164,400 | 8,164,4 | 100 bas 150 8 | ses 8,164,500 Reference Assem Your Se | 8,164,550 bly Fix Patch Sequence from Blat | 8,164,60 search | 38 0 - 1 | 8,164,650 | 8,164,700 |
| hg38_dna | | | | | CENCODE V28 | | | | |
| SINE LINE LTR DNA | | | | Repeating | Elements by Repe | eatMasker | | | |
| Simple Low Complexity Satellite RNA | | | | | | | | | |
| Other Unknown | | | | | | | | | |
| ve startClick on a2.0>reorder tra | feature for deta cks. Drag track | ils. Click+shift+d s left or right to n | rag to zoom ir ew position. F | n. Click side ba Press "?" for ke | rs for track opti byboard shortcu | ions. Drag side ts. | bars or lab | els up or down | to move < 2.0 |
| | track search | default tracks defa | ult order hide a | all add custom tr | acks track hubs | configure rev | resize | refresh | |
| collapse all | | Use drop | o-down contro | ow and p | ress refresh to | alter tracks dis | played. | expa | ind all |

| Custom tracks: uploading |
|--|
| r Genomes Genome Browser Tools Mirrors Downloads My Data Projects Help About Us |
| Add Custom Tracks |
| clade (Mammal +) genome (Human +) assembly (Dec. 2013 (GRCh38/hg38) +) |
| Display your own data as custom annotation tracks in the browser. Data must be formatted in bigBed, bigBarChart, bigChain, bigCenePred, bigInteract, bigLolly, bigMaf, bigPsl, bigWig, BAM, barChart, VCF, BED, BED detail, bedGraph, broadPeak, CRAM, GFF, GTF, hic, interact, MAF, narrowPeak, Personal Genome SNP, PSL, or WIG formats. |
| e e considered just of URL to the file, without a "track" line, for bigBed, bigWig, bigGenePred, BAM and VCF. To configure the display bet track and browser line attributes as described in the <u>User's Guide</u>. Examples are <u>here</u>. If you do not have web-accessible data storage available, please see the <u>Hosting</u> section of the Track Hub Help documentation. |
| Please note a much more efficient to bad data is to use Track Hubs, which are loaded from the Track Hubs Portal found in the menu under My Data. |
| Paste URLs or data: Or uproad. Choose File no file selected Submit Upload |
| Clear |
| Optional track documentation: Or upload: Choose File no file selected |
| Clear |
| Click here for an H [*] ocument template that may be used for Genome Browser track descriptions. |
| rojects Help About Us |
| Manage Custom Tracks |
| genome: Human assembly: Dec. 2013 (GRCh38/hq38) [hq38] |
| Name Description Type Doc Items Pos delete view in Genome Browser Igo |
| ATF1_ChIP_10mil ATF1_ChIP_10mil_bedGraph bedGraph 8443869 chr11: add custom tracks |
| ATF1_ChIP_bed ATF1_ChIP_bed bed 7252908 chr1: |
| Managing Custom Tracka |
| |

This section provides a brief description of the columns in custom track management table. For more details about managing custom tracks, see the Genome Browser User's Guide.

- Name a hyperlink to the update page where you can edit your track data.
- Description the value of the "description" attribute from the track line, if present. If no description is included in the input file, this field contains the track name.
- Type the track type, determined by the Browser based on the format of the data.
- Doc displays "Y" (Yes) if a description page has been uploaded for the track; otherwise the field is blank.
- Items the number of data items in the custom track file. An item count is not displayed for tracks lacking individual items (e.g. wiggle format data).
- Pos the default chromosomal position defined by the track file in either the browser line "position" attribute or the first data line. Clicking this link opens the Genome Browser or Table Browser at the specified position (note: only the chromosome name is shown in this column). The Pos column remains blank if the track lacks individual items (e.g. wiggle format data) and the browser line "position" attribute hasn't been set.

Custom tracks: saving and sharing

| | Â | Genomes | Genome Browser | ools Mirre | ors | Downlo | ads | My Data | Projects | He | lp | About Us |
|---|---------|-----------|-------------------------|--------------|------|---------|---------------|--------------|---------------|----|--------|----------|
| N | lanage | Custom | Tracks | | | | | Custom Tra | cks | | | |
| | aenome: | Human | assembly: Dec. 2013 (Gl | RCh38/ha38) | [ha: | 381 | | My Session | s | | | |
| | Na | ame | Description | Туре | Doc | Items | Pos | Track Hubs | | | | ♦) go |
| | ATF1_C | hIP_10mil | ATF1_ChIP_10mil_bedGr | aph bedGraph | ٦ | 8443869 | <u>chr11:</u> | Track Collec | ction Builder | , | tracks | |
| | ATF1_C | hIP_bed | ATF1_ChIP_bed | bed | | 7252908 | chr1: | Public Sess | ions | | | |
| | | | | | | | | | | | | |

Managing Custom Tracks

This section provides a brief description of the columns in custom track management table. For more details about managing custom tracks, see the Genome Browser User's Guide.



23

Save current

as:

(leave file blank to get output in browser window)

Cruising the genome: Displays and Tracks



Track Display:

| < 2.0 > | Click on a feature for detai reorder tracks. Drag tracks | Is. Click+shift+drag to zoom in. Click side bars for track options. Drag side bars or labels e left or right to new position. Press "?" for keyboard shortcuts. | up or down to n |
|-----------|---|--|-----------------|
| | track search det | fault tracks default order hide all manage custom tracks track hubs configure reverse resize | refresh |
| | collapse all | Use drop-down controls below and press refresh to alter tracks displayed. Tracks with lots of items will automatically be displayed in more compact modes. | expand all |
| | • | Custom Tracks | refresh |
| | • | Mapping and Sequencing | refresh |
| | • | Genes and Gene Predictions | refresh |
| | • | Phenotype and Literature | refresh |
| | • | COVID-19 | refresh |
| Available | • | Single Cell RNA-seq | refresh |
| display | • | mRNA and EST | refresh |
| tracks | • | Expression | refresh |
| tracks | • | Regulation | refresh |
| | • | Comparative Genomics | refresh |
| | • | Variation | refresh |
| | • | Repeats | refresh |
| | | refresh | |

Track Display:



26

Track Display:



Configure Display



Configure Display Options

| 1 | ñ | Genomes | Genome Browser | Tools | Mirrors | Downloads | My Data | Projects | Help | About U |
|-----|---------|---------------|--------------------------|--------------|--------------|------------------|----------------|----------|------|---------|
| Cor | nfigu | re Image | | | | | | | | |
| sut | bmit | | | | | | | | | |
| ima | age w | idth: | 1000 | pixels | | | | | | |
| lab | el are | a width: | 20 | characters | | | | | | |
| tex | t size: | | 12 ᅌ | | | | | | | |
| fon | ıt: | He | Ivetica 🗘 | | | | | | | |
| sty | le: | | Normal | | | | | | | |
| | Displa | ay chromoso | ome ideogram above | e main grap | hic | | | | | |
| | Show | light blue ve | ertical guidelines, or | light red ve | rtical windo | ow separators ir | n multi-region | view | | |
| | Displa | y labels to t | the left of items in tra | acks | | | | | | |
| | Displa | ay descriptio | n above each track | | | | | | | |
| | Show | track contro | ols under main grap | nic | | | | | | |
| | Next/p | previous iter | n navigation | | | | | | | |
| | Next/p | previous exc | on navigation | | | | | | | |
| | Show | exon numb | ers | | | | | | | |
| | Enabl | e highlight v | with drag-and-select | (if uncheck | ed, drag-ar | nd-select always | s zooms to se | lection) | | |
| | | | | | | | | | | |

Configure Tracks on UCSC Genome Browser: Human Dec. 2013 (GRCh38/hg38)

Tracks: track search hide all show all default Groups: collapse all expand all Control track and group visibility more selectively below.

| Custom Tracks | | | | | hide all | show all | default | submit |
|--|---------|------|----------|---------|-----------|----------|---------|--------|
| ATF1_ChIP_10mil | (full 🗘 |) A' | TF1_ChIP | _10mil_ | _bedGraph | | | |
| ATF1_ChIP_bed | hide 🗘 |) A' | TF1_ChIP | _bed | | | | |
| Mapping and Sequencing | | | | | hide all | show all | default | submit |
| Genes and Gene Predictions | | | | | hide all | show all | default | submit |
| Phenotype and Literature | | | | | hide all | show all | default | submit |
| + COVID-19 | | | | | hide all | show all | default | submit |
| • Single Cell RNA-seq | | | | | hide all | show all | default | submit |
| + mRNA and EST | | | | | hide all | show all | default | submit |
| + Expression | | | | | hide all | show all | default | submit |
| + Regulation | | | | | hide all | show all | default | submit |
| Comparative Genomics | | | | | hide all | show all | default | submit |
| + Variation | | | | | hide all | show all | default | submit |
| + Repeats | | | | | hide all | show all | default | submit |

Configure Display: Clarity

| submit | | | |
|-------------------|---------------------|----------|---|
| image width: | | 600 | pixels |
| label area width: | | 20 | characters |
| text size: | | 14 ᅌ | |
| font: | Helvetica | ٥ | |
| style: | Normal | \$ | |
| Display chrom | osome ideogran | n abov | e main graphic |
| Show light blu | e vertical guideli | ines, or | light red vertical window separators in multi-region view |
| Display labels | to the left of iter | ns in tr | acks |
| Display descri | ption above eac | h track | |
| Show track co | ntrols under mai | in grap | hic |
| Next/previous | item navigation | | |
| Next/previous | exon navigation | | |
| Show exon nu | mbers | | |
| Enable highlig | ht with drag-and | I-select | (if unchecked, drag-and-select always zooms to selection) |
| | | | |

Configure Tracks on UCSC Genome Browser: Human Dec. 2013 (GRCh38/hg38)

Tracks: track search hide all show all default Groups: collapse all expand all Control track and group visibility more selectively below.

| Custom Tracks | | hide all | show all | default sui | bmit |
|-----------------|--------|--------------------------|----------|-------------|------|
| ATF1_ChIP_10mil | full 🛊 | ATF1_ChIP_10mil_bedGraph | | | |
| ATF1_ChIP_bed | hide 🗘 | ATF1_ChIP_bed | | | |
| | | | | | |

UCSC Genome Browser on Human Dec. 2013 (GRCh38/hg38) Assembly



Configure Display: track order and options



Configure Display: track order and options

| ATF1_ChIP_10mil Track Settings |
|---|
| ATF1_ChIP_10mil_bedGraph (<u>All Custom Tracks</u>) |
| Display mode: full Submit Remove custom track Update custom ack Ex. Lower height |
| Type of graph: bar 🗘 |
| Track height: 128 pixels (range: 11 to 128) |
| Data view scaling: auto-scale to data view Always include zero: ON Ex. |
| Vertical viewing range: min: 0 max: 1000 (range: 0 to 1000) |
| Transform function: Transform data points by: NONE |
| Windowing function: (maximum +) Smoothing window: OFF +) pixe |
| Negate values: |
| Draw y indicator lines: at y = 0.0: OFF + at y = 0 |
| Graph configuration help |
| View table schema Pata last undated at UCSC: 2022-02-01 |
| |
| |

These are all track line settings that can be set when you upload data. You can upload without any settings and then modify them here

Track hubs: view public data



Track hubs: view public data

| | | | 10015 | - Millions- | -Dominoutio | - my Data | | Thorp | |
|---------------------------------|------------------|--|----------------------------------|---|---|--|--|-------------------|--|
| k Data I | Hubs | | | | | | | | |
| Public H | lubs | My Hubs | Hub De | velopment | | | | | |
| Frack da show up Frack Hu | ta hub in the | s are collections of hub's own blue ba created and maint | externa r track g ained by | l tracks th proup unde | at can be add r the browser | ed to the U graphic. Fo | CSC Genome or more infor | Browse mation, | er. Click Connect to attach a hul including <u>where to host your tra</u> |
| The list l | helow o | can be filtered on w | vords in | the hub de | escription pag | es or by as | semblies | | |
| Search t | erms: | e.g. methylation | | | As | sembly: e. | g. hg38 | Searc | h Public Hubs |
| Display | | Hub Name | De | escription | | | As: Click | c to conn | ect and browse directly |
| Conne | ct | ALFA Hub | NC for | BI's Allele F variants in | requency Aggre dbGaP studies. | gator (ALFA) | allele frequen | cy <u>hg</u> | <u>19, hg38</u> |
| Conne | ct | Bird Alignment (363 species) | Bir | d Alignment | (363 species) | | | [+ |] Gallus_gallus, Acanthisitta_chloris |
| Conne | ct | Bird assemblies | Bir | d genome a | ssemblies | | | [+ |] GCF_000699105.1, GCF_00069896 |
| Conne | ct | Blueprint Hub | Blu | eprint Epige | enomics Data H | ub | | <u>hg</u> | <u>38</u> |
| Conne | ct | BrainEpigenomeHub | DN ex sai | IA methylati pression dat mples | on, chromatin a a from NeuN flo | ccessibility, a w-sorted hu | and gene man brain | hg | 19 |
| Conne | ct | Breast Cancer IncRNA | A Bre | east Cancer | IncRNA PMID 2 | 5236012 | | hg | <u>19</u> |
| Conne | ct | Broad Improved Cani Annotation v1 | ne Bro | oad Institute | e CanFam3 Imp | roved Annota | tion Data v1 | <u>ca</u> | nFam3 |
| Conne | ct | C_elegans_isolates | C. | elegans wild | isolate assemb | oly hub | | [+ |] CB4856Princeton_JR-contig |
| Conne | ct | CADD | CA | DD Track fo | r v1.3 to v1.6 | | | hg | <u>19, hg38</u> |
| Conne | ct | Cancer Genomics Tra | TC Im Pej CIV | GA and ICG mune Epito ptides Datat /IC, MyCano | C Cancer Mutati pes Database (I pase, Dienstmar cerGenome.org, | ons, TCGA E EDB), Cance 1n Variant/Ca OncoKB | xpression, r Immunity ncer database | , <u>hg</u> | <u>19</u> |
| Conne | ct | CEMT (CEEHRC) | Epi | igenomic Da | ta tracks from | 3CGSC, Vanc | ouver, B.C. | hg | <u>38, hg19</u> |
| Conne | ct | CESAR Gene Mapping | js Hu | man Exons | mapped by CES | AR | | [+ <u>ca</u> l |] <u>bosTau7</u> , <u>ailMel1</u> , <u>allMis1</u> , <u>anoCar2</u> , <u>Jac3</u> |
| Conne | ct | ChIP-seq data track HUBs from MSC cells from GSE79815 | Ch dri dei (10 | IP-seq data ves adipoge rived mesen).1074/jbc.I | from the public nic and osteoge chymal stem ce 1116.736538) f | ation "Epiger nic differenti Ils" rom GEO rec | netic plasticity ation of marro ord GSE79815 | w- <u>mr</u> | <u>n9</u> |
| Conne | ct | Coloc segments | Co | localized se | gments of huma | n genome fo | r Roadmap cel | l <u>hg</u> | 19 |
| Conne | ct | Cotney Lab Human Craniofacial Epigenor | Hu nics UC | man Embry romatin Sta onn Health | onic Craniofacia te Segmentatio | l Tissue Epig ns from the (| enomic Data a Cotney Lab at | nd <u>hg</u> | <u>19, mm9, hg38</u> |
| Conne | ct | Cotney Lab Human | Hu | man Embry | onic Heart Tissu | e Epigenomi | c and | hg | <u>19, hq38</u> |

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Track hubs: view public data



ENCODE data is available from several organisms



Encyclopedia of DNA Elements at UCSC 2003 - 2012

| Human Data at UCSC | About | |
|-----------------------------|--|---|
| Experiment Matrix Search | The Encyclopedia of DNA Elements (ENCODE) Consortium is an im Research Institute (<u>NHGRI</u>). The goal of ENCODE is to build a comp act at the protein and RNA levels, and regulatory elements that cont | ernational collaboration of research groups funded by the National Human Genome orehensive parts list of functional elements in the human genome, including elements that rol cells and circumstances in which a gene is active. |
| Genome Browser (hg19) | UCSC coordinated data for the ENCODE Consortium from its incept production in 2012. All data produced by ENCODE investigators and Genome browser and database. Explore ENCODE data using the in | ion in 2003 (Pilot phase) to the end of the first 5 year phase of whole-genome data the results of ENCODE analysis projects from this period are hosted in the UCSC are greely the left menu bar. <i>All ENCODE data at UCSC are freely</i> |
| Experiment List | avaliable for download and analysis. | |
| Cell Types | ENCODE results from 2013 and later are available from the ENC ENCODE data from the first production phase, additional ENCODE | ODE Project Portal , <u>encodeproject.org</u> . The ENCODE Project Portal also hosts access tools, and ENCODE project pages including up-to-date information about data |
| Mouse Data at UCSC | releases, publications, and upcoming tutorials. | |
| Downloads | | |
| Experiment Matrix | Explore ENCODE data at UCSC | View ENCODE data in the UCSC Genome Browser |
| Search | ENCODE Experiment Matrix (2007-2012) +++ | UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly |
| Genome Browser (mm9) | A control of the second | |
| Experiment List | | Marrier Ma Arrier Marrier Marr |
| Cell Types | | |
| Metadata Terms | | |
| Registered Variables | | |
| Antibodies | | |

Experiment matrix

Preloaded browser tracks

ENCODE data tracks

| tracks. Drag tracks left or right | |
|--|----------------------------|
| to new position. Press "?" for | |
| Keyboard Snortcuts. | refresh |
| Use drop-down controls below and press refresh to alter tracks displayed | ellesi |
| Tracks with lots of items will automatically be displayed in more compact modes. | expand all |
| - Custom Tracks | refresh |
| ATF1_ChIP_10mil ATF1_ChIP_bed | |
| full \$ | |
| Cotney Lab Human Craniofacial Epigenomics | onnect refresh |
| Mapping and Sequencing | refresh |
| Genes and Gene Predictions | refresh |
| Phenotype and Literature | refresh |
| COVID-19 | refresh |
| * Single Cell RNA-seq | refresh |
| • mRNA and EST | refresh |
| Expression | refresh |
| - Regulation | refresh |
| ENCODE cCREs hide Hide Hide | Hi-C and cro-C de \$ |
| New JASPAR Transcription ORegAnno RefSeq Func Elems Factors ORegAnno RefSeq Func Elems | |
| hide | |
| Comparative Genomics | refresh |
| Variation | refresh |
| + Repeats | refresh |
| | 37 |

ENCODE data browser



ENCODE experiment matrix

| | Assays | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
|---|---------------|-----------|----------|-------------|---------|---------|--------|--------------------|---------|---------------------|------------|-----|-------------|-----|---------|--------|-------|-------|-------------|---------------|-------------------|---------|----|-------|--------|-------|-------|---------|-------------|---------|--------|
| search for: tracks files | A Methylation | hyl Array | hyl RRBS | n Chromatin | Ise-DGF | bas-asi | RE-seq | A Binding Proteins | Gene ST | Tiling Array | Validation | bas | A Profiling | SE | n Array | A-chip | A-PET | 4-seq | all RNA-seq | IS & Histones | P-seq view matrix | er | | A-PET | nbined | A-PET | otype | leosome | teogenomics | li-chip | li-seq |
| Cell Types | N | Met | Met | ð | DNa | DNa | FAII | R | RIP | RIP | RIP | RIP | N. | CAC | Exo | RN | RN | RN | Smi | H | Chi | ð | 50 | Chl | Con | DN | Gen | Nuc | Pro | Rep | Rep |
| Tier 1 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| GM12878 | 6 | 1 | 1 | | | 2 | 1 | | 7 | 4 | | 4 | | 6 | 2 | 6 | 2 | 12 | 5 | | 133 | | 2 | | 2 | 3 | 1 | 1 | 5 | | 1 |
| H1-hESC | • | 1 | 1 | | | 2 | 1 | | 3 | | | | | 4 | 1 | | 1 | 10 | 3 | | 91 | | 1 | | 2 | | 1 | | 2 | 1 | |
| K562 | | 1 | 1 | | 3 | 16 | 3 | | 6 | 4 | | 4 | | 9 | 7 | 9 | 6 | 17 | 7 | | 224 | | 2 | 2 | 2 | 3 | 1 | 1 | 6 | | 1 |
| Tier 2 | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| A549 | | 1 | 1 | | 1 | 2 | 1 | | | | | | | 3 | 2 | | 3 | 10 | 9 | | 87 | | | | | | 1 | | | | |
| CD20+ | • | | | | | | | | | | | | | 1 | | | | 2 | 1 | | 4 | | | | | | | | | | |
| CD20+_RO01778 | • | | | | 1 | 1 | | | | | | | | | | | | | | | 2 | | | | | | | | | | |
| CD20+_RO01794 | • | | | | | 1 | | | | | | | | | | | | | | | 5 | | | | | | | | | | |
| H1-neurons | • | | | | | | | | | | | | | | | | | | 3 | | 4 | | | | | | | | 1 | | |
| HeLa-S3 | | 1 | 1 | | | 3 | 3 | | 4 | | | | | 6 | 4 | | 3 | 8 | 3 | | 93 | | 1 | 1 | 2 | | 1 | | | 1 | 1 |
| HepG2 | | 1 | 1 | | 1 | 2 | 1 | 1 | 4 | | | | | 6 | 2 | 5 | 2 | 8 | 3 | | 114 | | 1 | | 2 | | 1 | | | | 1 |
| HUVEC | ۲ | 1 | | | 1 | 2 | 1 | - [| | | | 1 | | 5 | 2 | | 2 | 8 | 1 | | 36 | | | | 2 | | 1 | | | | 1 |
| IMR90 | • | 1 | 1 | | | 1 | | | | | | | | 3 | | | 3 | 4 | 9 | | 11 | | | | | | 1 | | | 1 | 1 |
| LHCN-M2 | | | | | 2 | 2 | | | | | | 1 | | | | | | 2 | | | | | | | | | | | | | |
| MCF-7 | | 1 | 3 | | | 8 | 3 | | | | | | | 3 | 7 | | 3 | 5 | 7 | | 49 | | 1 | 3 | | | 1 | | | | 1 |
| Monocytes-CD14+ | 0 | | | | | 1 | | | | | | | | 1 | | | | 2 | 1 | | | 1 | | | | | | | | | |
| Monocytes-CD14+_RO01746 | • | | | | 1 | 1 | | | | | | | | | | | | | | | 17 | | | | | | | | | | |
| SK-N-SH | | 1 | 1 | | | 1 | | | | | | | | 3 | | | 3 | 4 | 9 | | 34 | | | | | | | | | | 1 |
| 7 | | - | | 1 | | | | 1 | | - | | - | 12 | | | | - | | | | | 1. 1. L | | | - | | - | - | 0 | 0 (C) | |

Can view in browser or download raw and processed data

Resources / links

Browser:

- <u>http://www.sciencedirect.com/science/article/pii/</u>
 <u>S0888754308000451</u>
- <u>http://genome.ucsc.edu/training/vids/</u>
- <u>http://www.nature.com/scitable/ebooks/guide-to-the-ucsc-genome-browser-16569863</u>

ENCODE:

- <u>http://genome.ucsc.edu/ENCODE/index.html</u>
- <u>http://www.genome.gov/encode/</u>