# Visualizing ENCODE Data in the UCSC Genome Browser



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- Genomewiki: genomewiki.ucsc.edu
- Mailing list archives: genome.ucsc.edu/FAQ/
- Training page: genome.ucsc.edu/training.html
- Twitter Section @GenomeBrowser
- Tutorial videos: YouTube channel
- Open Helix: openhelix.com/ucsc

# Outline

- Basics: search, display, more info
- Tools for finding ENCODE data
- Annotating a BED file: RNAseq example
- Annotating a VCF file
- Track Hubs: What are they? How do I make one?
- Exercises

# **Basic Navigation: Main Display**

### genome.ucsc.edu/cgi-bin/hgTracks?db=hg19



# **Display Configuration**

• Visibility:

hide, dense, squish, pack, full



- Track ordering: drag and drop
- Drag and zoom/highlighting
- Configuration page
- Right click menu



# How to find more info



# More info: Track Description

Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us
UCSC G	enes Track S	Settings						
UCS Com	C Gen parativ	es (RefSe ve Genon	eq, Ge nics)	enBar (* <u>All Gen</u>	nk, CCD	S, Rfa	m, tF	RNAs &
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Show co	don numberi	ng: 🗆						
<u>View tabl</u> Data last	<u>e schema</u> updated: 20	13-06-14						

### Description

The UCSC Genes track is a set of gene predictions based on data from RefSeq, GenBank, CCDS, Rfam, and the <u>tRNA Genes</u> track. The track includes both protein-coding genes and non-coding RNA genes. Both types of genes can produce non-coding transcripts, but non-coding RNA genes do not produce protein-coding transcripts. This is a moderately conservative set of predictions. Transcripts of protein-coding genes require the support of one RefSeq RNA, or one GenBank RNA sequence plus at least one additional line of evidence. Transcripts of non-coding RNA genes require the support of one RefSeq RNA, or one Rfam or tRNA prediction. Compared to RefSeq, this gene set has generally about 10% more protein-coding genes, approximately four times as many putative non-coding genes, and about twice as many splice variants.

# More info: Item Description

 Image: Construction of the large subunit of RNA polymerase II and provide a link between transcription and pre-mRNA splicing. Alternatively spliced transcript variants have been described. [provided by RefSeq, Feb 2009].
 My Data
 Help
 About Us

Position: chr21:33,043,313-33,104,431 Size: 61,119 Total Exon Count: 20 Strand: -

#### **Coding Region**

Position: chr21:33,043,712-33,104,005 Size: 60,294 Coding Exon Count: 20

Page Index	Sequence and Links	UniProtKB Comments	CTD	Gene Alleles	RNA Structure				
Protein Structure	Other Species	GO Annotations	mRNA Descriptions	Other Names	Model Information				
Methods									

Data last updated: 2013-06-14

#### Sequence and Links to Tools and Databases

Genomic Sequ	ience (chr21:33,043	,313-33,104,431)	mRNA (may	differ from genome)	Protein (1147 aa)
Gene Sorter	Genome Browser	Protein FASTA	VisiGene	Table Schema	BioGPS
CGAP	Ensembl	Entrez Gene	ExonPrimer	GeneCards	GeneNetwork
Gepis Tissue	H-INV	HGNC	HPRD	Jackson Lab	Lynx
MOPED	neXtProt	OMIM	PubMed	Stanford SOURCE	UniProtKB

#### Comments and Description Text from UniProtKB

### ENCODE



# **ENCODE: Super-track Settings**

-	Regulation refresh									
ENCODE Regulation	B CD34 Dnasel	CpG Islands	ENC Chromatin hide	ENC DNA Methyl hide	ENC DNase/FAIRE hide					
ENC Histone	ENC RNA Binding	Show	FSU Repli-ch	hide	IS <u>NKI Nuc</u> Lamina hide ▼					
Genomes	Genome Browser	Tools Mirrors [	Downloads My Data	Help About Us						

#### ENC TF Binding Super-track Settings



- <u>UChicago TFBS</u>
   Transcription Factor Binding Sites by Epitope-Tag from ENCODE/UChicago
  - Open Chromatin TFBS by ChIP-seq from ENCODE/Open Chrom(UT Austin) ENCODE July 2011 Freeze
- UW CTCF Binding CTCF Binding Sites by ChIP-seq from ENCODE/University of Washington

#### Description

UTA TFBS

hide

hide

hide

# **ENCODE: Track Settings**



Uniform TFBS Track Settings

ENCODE Downloads Subtracks Description Contact

Transcription Factor ChIP-seq Uniform Peaks from ENCODE/Ana

Display mode:	dense	-	Submit	Cancel	Reset to defaults

Score range: min: 0 (0 to 1000)

#### Select subtracks by cell line and factor: (help)



### **ENCODE: Item Details**



#### MCF-7 (serum\_stimulated) TFBS Uniform Peaks of c-Myc from ENCODE/UT-A/Analysis

Position: chr17:7589391-7589530 Peak point: 7589461 Score: 688 Signal value: 107.768 Q-value (FDR): 4.212

View table: schema, downloads, metadata -

Go to Uniform TFBS track controls

Data version: ENCODE Mar 2012 Freeze Data last updated: 2013-04-12

### Description

This track represents a comprehensive set of human transcription factor binding sites based on ChIP-seq experiments generated by production in the ENCODE Consortium from the inception of the project in September 2007, through the March 2012 internal data freeze. The track represented by the trac

# **ENCODE** Tools

### UCSC Genome Bioinformatics

Genomes - Blat - Tables - Gene Sor	ter - PCR	<ul> <li>VisiGene</li> </ul>	- Session -	FAQ -	Help
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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 <u>ENCODE</u> data at UCSC (2003 to 2012) and to the <u>Neandertal</u> project. Download or purchase the Genome Browser source code, or the Genome Browser in a Box (<u>GBiB</u>) at our <u>online store</u>.

We encourage you to explore these sequences with our tools. The <u>Genome Browser</u> zooms and scrolls over chromosomes, showing the work of annotators worldwide. The <u>Gene Sorter</u> shows expression, homology and other information on groups of genes that can be related in many ways. <u>Blat</u> quickly maps your sequence to the genome. The <u>Table Browser</u> provides convenient access to the underlying database. <u>VisiGene</u> lets you browse through a large collection of *in situ* mouse and frog images to examine expression patterns. <u>Genome Graphs</u> 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 <u>UC Santa Cruz</u> <u>Genomics Institute</u> and the Center for Biomolecular Science and Engineering (<u>CBSE</u>) at the University of California Santa Cruz (<u>UCSC</u>). If you have feedback or questions concerning the tools or data on this website, feel free to contact us on our <u>public mailing list</u>.

The Genome Browser project team relies on public funding to support our work. Donations are welcome -- we have many more ideas than our funding supports! If you have ideas, drop a comment in our suggestion box.

#### News 💟 F

News Archives >

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To receive announcements of new genome assembly releases, new software features, updates and training seminars by email, subscribe to the genomeannounce mailing list. Please see our blog for posts about Genome Browser tools, features, projects and more.

#### 03 March 2015 - Use UDR for Quick Sizable Downloads

The UCSC Genome Browser is pleased to share a download protocol to use when downloading large sets of files from our download servers: **UDR (UDT Enabled Rsync)**. UDR utilizes rsync as the transport mechanism, but sends the data over the UDT protocol, which enables huge amounts of data to be downloaded efficiently over long distances.

Protocols like http, ftp and rsync can have a problem in that the further away the download source is from the user, the slower the speed becomes. Protocols like UDT/UDR allow for many UDP packets to be sent in batch, thus allowing for much higher transmit speeds over long distances. UDR will be especially useful for users who are downloading from places that are far away from California. The US East Coast and the international community will likely see much higher download speeds by using UDR rather than rsync, http or ftp. <u>Read more</u>.

Genomes

Microbial

Genome Browser

Ebola

Blat

Table Browser

Gene Sorter

In Silico PCR

Genome

Graphs

Galaxy

VisiGene

Utilities

Downloads

**Release Log** 

Custom Tracks

Cancer

Browser

# ENCODE





### genome.ucsc.edu/ENCODE/

### Encyclopedia of DNA Elements at UCSC 2003 - 2012

Human Data at UCSC	About	
Downloads Experiment Matrix	The <u>Encyclopedia of DNA Elements</u> (ENCODE) Consor funded by the National Human Genome Research Instit comprehensive parts list of functional elements in the hu RNA levels, and regulatory elements that control cells a	tium is an international collaboration of research groups ute ( <u>NHGRI</u> ). The goal of ENCODE is to build a uman genome, including elements that act at the protein and nd circumstances in which a gene is active.
Search Genome Browser (hg19)	UCSC coordinated data for the ENCODE Consortium from year phase of whole-genome data production in 2012. A ENCODE analysis projects from this period are hosted in ENCODE data using the image links below or via the left for down load and production.	om its inception in 2003 (Pilot phase) to the end of the first 5 All data produced by ENCODE investigators and the results of n the UCSC Genome browser and database. Explore ft menu bar. <i>All ENCODE data at UCSC are freely available</i>
Experiment List Cell Types Mouse Data at UCSC	for download and analysis. <b>ENCODE results from 2013 and later are available fr</b> ENCODE Project Portal also hosts ENCODE data from and ENCODE project pages including up-to-date inform tutorials.	om the ENCODE Project Portal, <u>encodeproject.org</u> . The the first production phase, additional ENCODE access tools, ation about data releases, publications, and upcoming
Downloads  Experiment	Explore ENCODE data at UCSC	View ENCODE data in the UCSC Genome Browser
Matrix Search Genome Browser (mm9)	ENCODE Experiment Matrix (2007-2012) >===	UCSC Genome Browser on Human Feb. 2009 (GRCh37/hg19) Assembly men and a man

# **ENCODE: Experiment Matrix**

	As	says																				
search for: <a>tracks</a> files	hylation	rray	RBS	romatin	GF	be	ba	ding Proteins	s ST	g Array	ation		filing		ay	_			A-seq	listones	view matrix	
Cell Types	DNA Met	Methyl A	Methyl R	Open Ch	DNase-D	DNase-se	FAIRE-se	RNA Bin	<b>RIP Gene</b>	RIP Tiling	<b>RIP Valid</b>	RIP-seq	<b>RNA Pro</b>	CAGE	Exon Arr	RNA-chip	RNA-PET	RNA-seq	Small RN	TFBS & I	ChIP-seq	Other
Tier 1								_					_							_		
GM12878	•	1	1			2	1		7	4		4		6	2	6	2	12	5		133	
H1-hESC	•	1	1			2	1		3					4	1		1	10	3		91	
K562	•	1	1		3	16	3		6	4		4		9	7	9	6	17	7		224	
Tier 2																						
A549	•	1	1		1	2	1							3	2		3	10	9		87	
CD20+	•													1				2	1		4	
CD20+_RO01778	•				1	1							-								2	
CD20+_RO01794	•					1															5	
H1-neurons	•																		3		4	
HeLa-S3	•	1	1			3	3		4					6	4		3	8	3		93	
Han C2						•								-	_	-	•	•	2			

# **ENCODE: ChIP-Seq Matrix**



# **ENCODE: Experiment Summary**



### ENCODE Experiment Summary hg19 2007 - 2012

search for: otracks files

Genome Annotation	
GENCODE	4
Mappability	10
ORChID	1
Total: 3	15

All Experiments	view matrix by cell line
5C	13
CAGE	78
ChIA-PET	8
ChIP-seq	1479
Combined	26
DNA-PET	6
DNase-DGF	56
DNase-seq	219
Exon Array	158
FAIRE-seq	37
Genotype	64
Methyl Array	62

ChIP-seq Experiments	view matrix by cell line
ARID3A	2
ATF1	1
ATF2	2
ATF3	7
BACH1	2
BATF	1
BCL11A	3
BCL3	3
BCLAF1	2
BDP1	2
BHLHE40	5
BRCA1	4

### **ENCODE: Track Search**

#### Search for Tracks in the Human Feb. 2009 (GRCh37/hg19) Assembly

Search Advanced		
Track Name:	contains	
and Description:	contains	
and Group:	is Any ᅌ	
and Data Format:	is Any	
		ENCODE terms
+ and Antibody or target prote	ein 📀 is among BRCA1 (A300-000A)	Antibody or target protein
+ and View - Peaks or Signals	s is among Any	
search clear cancel		

return to browser (12 of 12 selected)

÷. –	Visibility	Track Name	Sort: • by Relevance Alphabetically by Hierarchy
	pack 🛊	GM12878 BRCA1	GM12878 TFBS Uniform Peaks of BRCA1_(A300-000A) from ENCODE/Stanford/Analysis -
	pack 🛊	H1-hESC BRCA1	H1-hESC TFBS Uniform Peaks of BRCA1_(A300-000A) from ENCODE/Stanford/Analysis -
	pack 🛊	HeLa-S3 BRCA1	HeLa-S3 TFBS Uniform Peaks of BRCA1_(A300-000A) from ENCODE/Stanford/Analysis -
	pack 🛟	HepG2 BRCA Displa	ay track details 3S Uniform Peaks of BRCA1_(A300-000A) from ENCODE/Stanford/Analysis -
	pack 🛊	GM78 BRC1 lgM	GM12878 BRCA1 IgG-mus ChIP-seq Peaks from ENCODE/SYDH -
	full 🛊	GM78 BRC1 lgM	GM12878 BRCA1 IgG-mus ChIP-seq Signal from ENCODE/SYDH -
	pack 🛟	H1ES BRC1 lgR	H1-hESC BRCA1 IgG-rab ChIP-seq Peaks from ENCODE/SYDH -
	full 🛊	H1ES BRC1 lgR	H1-hESC BRCA1 IgG-rab ChIP-seq Signal from ENCODE/SYDH -
	pack 🛊	<u>HeLa BRC1 lgR</u>	HeLa-S3 BRCA1 IgG-rab ChIP-seq Peaks from ENCODE/SYDH -
	full 🛊	<u>HeLa BRC1 lgR</u>	HeLa-S3 BRCA1 IgG-rab ChIP-seq Signal from ENCODE/SYDH -
	pack 🛟	HEPG BRC1 lgR	HEPG2 BRCA1 IgG-rab ChIP-seq Peaks from ENCODE/SYDH -
	full A	HEPG BRC1 IdR	HEPG2 BBCA1 InG_rab ChIP-seq Signal from ENCODE/SVDH





Positional annotations. (ex. Regions w/: enriched ChIP-seq signal for TF binding,  $\Delta$ 'I methylation, splice jxns from RNA-seq)

wig(gle)

BED

Continuous signal data. # of reads (ex. DNase I HS and ChIP-seq signals)

BAM

Alignments of seq. reads, mapped to genome (ex. RNAseq alignments)

VCF

Variation data: SNPs, indels, Copy Number Variants, Structural Variants (ex. ExAC data)

### **Indexed** File Formats









wig(gle)



bigWig

BAM

VCF

# Indexed File Formats

- Only displayed portions of files
   transferred to UCSC
- Display large files (would time out)
- File + index on your web-accessible server (http, https, or ftp)
- Faster display
- More user control

Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us
Add Cu	stom Tracks							
clade Display y BAM, BE as descr box belo	Aammal C Your own data a D detail, Perso ibed in the Use w. Publicly ava	genome Human as custom annotational Genome SNF er's Guide. Data in t illable custom tracks	CIT tracks i VCF, broat he bigBed s are listed	assembly in the browse adPeak, narm , bigWig, BA d <u>here</u> . Exam	Feb. 2009 (GRCh37/h er. Data must be <u>owPeak</u> , or <u>PSL</u> in and VCF for nples are <u>here</u> .	e formatted in formats. To mats can be p	BED, big configure provided v	Bed, <u>bedGraph, GFF, GTF, WIG, bigWig, MAF,</u> the display, set <u>track</u> and <u>browser</u> line attributes ia only a URL or embedded in a track line in the

Submit

Or upload: Browse... No file selected.

Paste URLs or data:

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UC	CSC Genome	e Bioinfo	rmatics					
Home	- Genomes - Blat -	Tables - Gen	e Sorter - PCR	- VisiGene	- Session -	Help		

#### FAQ Table of Contents

This page contains responses to questions frequently asked by our user community and subscribers to the Genome Browser mailing

- Display Problems
- Assembly Releases and Versions
- Data and Downloads
- Genome Browser Tracks
- Custom Annotation Tracks
- Data File Formats
- Blat
- ENCODE Resources and EAO

### UCSC Genome Bioinformatics

Home - Genomes - Blat - Tables - Gene Sorter - PCR - VisiGene - Session - Help

#### Frequently Asked Questions: Data File Formats

**General formats:** 

- Axt format
- BAM format
- BED format
- BED detail format
- bedGraph format
- bigBed format
- bigGenePred table format
- bigWig format

#### ENCODE-specific formats:

- ENCODE broadPeak format
- ENCODE gappedPeak format
- ENCODE narrowPeak format
- ENCODE pairedTagAlign format
- ENCODE peptideMapping format
- ENCODE RNA elements format
- ENCODE tagAlign format

### File Formats www.encodeproject.org/help/file-formats/





	Help <del>-</del>	Search ENCODE Q Sign in
	Getting started REST API	
Common File Formats	File formats Tutorials	he ENCODE Consortium
Overview	Contact	

### 

The ENCODE consortium uses several file formats to store, display, and disseminate data:

- FASTQ: a text-based format for storing nucleotide sequences (reads) and their quality scores. [1]
- BAM: The Sequence Alignment/Mapping (SAM) format is a text-based format for storing read alignments against reference sequences and it is interconvertible with the binary BAM format. [2]
- bigWig: The bigWig format is an indexed binary format for rapid display of continuous and dense data in the UCSC Genome Browser.
- bigBed: The bigBed format is also an indexed binary format for rapid display of annotation items such as a linked collection of exons or the binding peaks of a transcription factor.

These file formats were originally designed to be generic and flexible. As the ENCODE consortium is a collaborative effort, the consortium has made several specifications on the file formats to facilitate data archival, presentation, and distribution, as well as integrative analysis on the data. The consortium considers FASTQ as the basic file format for archival purpose and thus the FASTQ format's specifications aim to preserve the raw

### **Custom Tracks**

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	group	genome	assembly		pos	ition		sea	arch term		
Mar	mmal ᅌ	Human ᅌ	Feb. 2009 (GRCh37/hg	19) ᅌ (	chr17:7,583,	734-7,595,1 <sup>.</sup>	19 enter p	osition, gene	symbol or search	n terms	submit
	<u>Click</u>	<u>chere to reset</u> the	track search add	rface so	acks track hu	ir defaults.	More or	n <b>-site wor</b> splay	<mark>kshops ava</mark>	<u>ailable!</u>	
Â	Genomes	Genome Browser	Tools Mirro	ors I	Downloads	My Data	View	Help	About Us		
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	Scale chr17: TP53 TP53 TP53 TP53	7,585,000  7 	5 Kb - ,586,000  7,587,000	7,588,0 Bas	000  7,589,000  ic Gene Annotati -199F11.2	7,590,000  on Set from GEN 	7,591,000 CODE Version	 7,592,000  19 WRAP53 WRAP53	hg19 7,593,000	7,594,000	7,595,000
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### **Custom Tracks**

### genome.ucsc.edu/cgi-bin/hgCustom

Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us
Add Cu	stom Tracks							
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BED fo additio optiona If your The fire	ormat provides a nal optional fiel al fields is bindin data set is BEE st three require	a flexible way to de ds. The number of ng: lower-numbered D-like, but it is very d BED fields are:	fine the da fields per l d fields mu large and y	ata lines tha line must be ust always b you would li	t are displayed i e consistent thro e populated if h ke to keep it on	n an annotati ughout any s igher-numbe your own se	ion track. E ingle set o red fields a rver, you si	BED lines have three required fields and nine f data in an annotation track. The order of the are used. hould use the <u>bigBed</u> data format.
1. <b>c</b>	hrom - The na	me of the chromoso	ome (e.g. o	chr3, chrY, d	chr2_random) o	r scaffold (e.g	g. scaffold1	0671).

- 2. chromStart The starting position of the feature in the chromosome or scaffold. The first base in a chromosome is numbered 0.
- 3. chromEnd The ending position of the feature in the chromosome or scaffold. The chromEnd base is not included in the display of the feature. For example, the first 100 bases of a chromosome are defined as chromStart=0, chromEnd=100, and span the bases numbered 0-99.

The 9 additional optional BED fields are:

4 name - Defines the name of the RED line. This lead is displayed to the left of the RED line in the Conome Browser window when the track is

### **Custom Tracks**

### genome.ucsc.edu/cgi-bin/hgCustom

Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us	
Add Cu	ustom Tracks								
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# Annotating your data: BED





### Data Integrator genome.ucsc.edu/cgi-bin/hgIntegrator

Data Integrator Undo Red	0			
Select Genome Assembly a	nd Region			
groupgenomeMammalImage: Human	Sembly (GRCh37/hg19)			
position or search term Chr21:330	31597-33041570			
Configure Data Sources				
SYDH_TFBS View table sche	ma			×
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Add Data Source				
track group       Genes and Gene Predictions       View table schema	GENCODE Genes V19	view Genes ᅌ	subtrack Basic (wgEncodeGencodeBasicV19)	<b>C</b>
get more data: track hubs custom tracks				
Output Options				
Choose fields				
Get output				

Data		do Redo	
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Confi	✓ chromEnd ✓ name	End position in chromosome Name of item.	
ţsy ţgi	score	Score (0-1000) + or - for strand	× ×
Add I	Basic Set all Clear all		
Genes	☑ name □ chrom	Name of gene (usually transcript_id from GTF) Reference sequence chromosome or scaffold	
get n track	☐ strand ☐ txStart	+ or - for strand Transcription start position	
Outp	□ txEnd □ cdsStart	Transcription end position Coding region start	
Choce	cdsEnd	Coding region end Number of exons	
Get o	<ul> <li>exonStarts</li> <li>exonEnds</li> </ul>	Exon start positions Exon end positions	
Usin	<ul> <li>score</li> <li>name2</li> <li>aduStartStat</li> </ul>	score Alternate name (e.g. gene_id from GTF)	
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Data Integrator Undo Redo		
Select Genome Assembly and Region		
group     genome     assembly       Mammal     Image: Human     Feb. 2009 (GRCh37/hg19)     Image: Comparison of the second seco		
region to annotate         position or search term         Chr21:33031597-33041570		
Configure Data Sources		
SYDH_TFBS <u>View table schema</u> GENCODE Genes V19 - Basic <u>View table schema</u>		× ×
Add Data Source		
track group       track       view       subtrack         Genes and Gene Predictions       Image: GENCODE Genes V19       Image: Genes       Image: Genes       Image: Genes Code G	0	
Output Options		
Choose fields Get output		
Using the Data Integrator		

#ct_SYDHTFBS_4733.chrom ct_SYDHTFBS_4733.chromStart							
ct_SYDH	TFBS_4733.ch	romEnd ct	_SYDHTFBS_4	733.name	ct_SYDHTFBS_4733.score		
wgEncodeGencodeBasicV19.name wgEncodeGencodeBasicV19.pame2							
chr21	33031473	33032186	•	608	ENST00000449339.1		
AP00025	3.1						
chr21	33031473	33032186	•	608	ENST00000270142.6	SOD1	
chr21	33031473	33032186	•	608	ENST00000389995.4	SOD1	
chr21	33031473	33032186	•	608	ENST00000470944.1	SOD1	

# Annotating your VCF file

- 1. Make a VCF custom track
- 2. Go to the Variant Annotation Integrator
- 3. Choose your track
- 4. Add annotations

# **Remotely Hosted Custom Tracks**

- Put data file (bigBed/bigWig/BAM/VCF, etc) in internet accessible location
- Must have: 1. track info, 2. bigDataUrl
- VCF example:

```
track type=vcfTabix
name="VCF_Example"
description="VCF Ex. 1: 1000 Genomes phase 1
interim SNVs"
bigDataUrl=
http://hgwdev.cse.ucsc.edu/~pauline/presentations/
vcfExample.vcf.gz
```

# Variant Annotation Integrator

- Upload pgSnp or VCF custom track
- Associate UCSC annotations with your uploaded variant calls
- Add dbSNP info if dbSNP identifier found
- Select custom track and VAI options

# Variant Annotation Integrator

### Tools

### Variant Annotation Integrator



# Variant Annotation Integrator

### genome.ucsc.edu/cgi-bin/hgVai

Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us					
Variant	Variant Annotation Integrator												
Select	Genome As	ssembly and R	egion										
clade	genor	ne assem	nbly										
Mammal	Human	n ᅌ Feb. 20	09 (GRCh37/hg	19) ᅌ									
region to annotate genome													
Select Variants If you have more than one custom track or hub track in pgSnp or VCF format, please select the one you wish to annotate:													
variants	COLE X. 1: 100	0 Genomes phase 1 inter	im SNVs 🙁										

maximum number of variants to be processed: 10	0,000 🗘	
--	---------	--

manage custom tracks [track hubs] To reset all user cart settings (including custom tracks), click here.

#### Select Genes

The gene predictions selected here will be used to determine the effect of each variant on genes, for example intronic, missense, splice site, interg

#### Select Regulatory Annotations

The annotations in this section provide predicted regulatory regions based on various experimental data. When a variant overlaps an annotation s consequence term regulatory region variant will be assigned. Follow the links to description pages that explain how each dataset was constructed significant portion of the genome and it may be desirable to filter these annotations by cell type and/or score in order to avoid an overabundance of the second dataset.

# Track Data Hubs

- Remotely hosted
- Data persistence
- File formats: bigBED, bigWig, BAM, VCF
- Track organization: groups, supertracks
- multiWigs
- Assembly hubs

# Track Hubs





### Track Hubs

#### Human (Homo sapiens) Genome Browser Gateway



# Track Hubs

### genome.ucsc.edu/cgi-bin/hgHubConnect



Track data hubs are collections of external tracks that can be imported into the UCSC Genome Browser. Hub tracks show up under the label bar on the main browser page, as well as on the configure page. For more information, see the <u>User's Guide</u>. To import a public hu "Connect" button below.

#### NOTE: Because Track Hubs are created and maintained by external sources, UCSC is not responsible for their content.



IMassMed H3K4me3 ChIP-seq data for

### My Hubs genome.ucsc.edu/cgi-bin/hgHubConnect







Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us
Frack D	ata Hubs							

Track data hubs are collections of external tracks that can be imported into the UCSC Genome Browser. Hub tracks show up under the hub's own blue label bar on the main browser page, as well as on the configure page. For more information, see the <u>User's Guide</u>. To import a public hub click its "Connect" button below.

NOTE: Because Track Hubs are created and maintained by external sources, UCSC is not responsible for their content.

Public Hubs	My Hubs	
URL:	Add Hub	
	No Unlisted Track Hubs	
Contact genome@	soe.ucsc.edu to add a public hub.	

# Make Your Own Track Hub

### You will need:

- Data (compressed binary index formats: bigBed, bigWig, BAM, VCF)
- Text files to define properties of the track hub
- Internet-enabled web/ftp server
- Assembly Hubs: a twoBit sequence file

# Track Hubs

### genome.ucsc.edu/cgi-bin/hgHubConnect







myHub/ - directory containing track hub files



hub.txt - a short description of hub properties



genomes.txt - list of genome assemblies included



hg19/ - directory of data for the hg19 human assembly



Data files! BAM, bigBed, bigWig, VCF

### An Example Assembly Hub

### An Arabidopsis hub:

### http://genome-test.cse.ucsc.edu/ ~pauline/hubs/Plants/hub.txt



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- Bob Kuhn Associate Director, Outreach co-PI
- Donna Karolchik, Ann Zweig Project Management

### Engineering

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THE ORIGINAL AUTHORITY ON QUESTIONING AUTHORITY

UC Santa Cruz Genomics Institute

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**National Cancer Institute (NCI)** 

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Eunice Kennedy Shriver National Institute of Child Health and Human Development



UC Santa Cruz Genomics Institute

### genome.ucsc.edu

# **THANK YOU!**

UC Santa Cruz Genomics Institute

### Exercises

- 1. Load example BED and VCF tracks via url
- 2. Look at custom track data by pasting url into a web browser.
- 3. Annotate the TFBS custom track using the Data Integrator.
- 4. Annotate the VCF custom track using the Variant Annotation Integrator.

# **Exercise** 1

### Load example BED and VCF tracks via url

- 1. Go to the Custom tracks menu
  - My Data -> Custom Tracks
- 2. Input this url: <u>http://bit.ly/customtracks</u> (note that you must include the "http" part of this url or you will get an error) and click [submit].
- 3. Click the [Go to genome browser] button.
- 4. Once in the main Browser, jump to this position:
  - chr21:33,034,804-33,037,719
- 5. See if you can drag your 2 custom tracks to the top of the display

### **Exercise 2** Exploring your BED and VCF tracks

- 1. Now that you have 2 custom tracks loaded, take a look at the data by pasting that same url into a web browser:
- 2. These custom tracks are actually data copied from some existing tracks, see if you can find them, turn them on, and observe that the original tracks and custom tracks look the same in the browser:
  - <u>Track 1 (BED format)</u>: Group (Regulation), Super Track (ENC TF Binding), Track (SYDH TFBS)
  - <u>Track 2 (VCF format)</u>: Group (Variation), Track (1000G Ph1 Vars)
- 3. Navigate to this position for best comparison (esp. for the VCF track): chr21:33,034,804-33,037,719

### **Exercise 3**

### Annotate your BED with the Data Integrator

- 1. Go to the Data Integrator
- 2. Once there select:
  - 1. Region to annotate: chr21:33031597-33041570
  - 2. Add data source: group (custom tracks), track (SYDH...) [click add]
- 3. Now choose which annotations you want to add by [add]ing more tracks to the list ex:
  - Find the genes that overlap with your regions: group (Genes and Gene Prediction), track (GENCODE V19), view (Genes), subtrack (Basic) [add]
  - 2. Find the SNPs that overlap with your regions: group (Variation), track (Common SNPs) [add]

Choose which fields to include in your output:

Output options -> Choose fields [Done] -> [get output]

### **Exercise** 4

Annotate your VCF with the Variant Annotation Integrator

- 1. Go to the Variant Annotation Integrator
  - Tools -> V.A.I.
- 2. Select Variants:
  - Variants: "VCF Ex. 1..."
- 3. Now choose which annotations you want to add:
  - To determine which gene regions your variants fall into, select a gene track (Select Genes = "Basic Gene Annotation Set... GENCODE")
  - Add regulatory annotations: Under "Select Regulatory Annotations" click the "+" button to choose which TFs to include (or select none to include all binding sites)

# **Bonus Material!**

# Where to search genome.ucsc.edu/cgi-bin/hgGateway

Genon	nes Geno	Genome Browser		Mirrors	Downloads	My Data	Help	About Us		
(Homo	sapiens) G	ienome Br	owser Ga	ateway						
		The UCSC ( Softw	Genome Brov /are Copyrigh	wser was creat It (c) The Rege	ted by the <u>Genome</u> ents of the Universi	Bioinformatics (	Group of UC All rights rese	<u>Santa Cruz</u> . erved.		
group	roup genome		assemb	bly	positi	on	search term			
nmal	O Human	Feb	. 2009 (GRCh3	7/hg19) ᅌ C	hr21:33,031,59	7-33,041,570	brc			submit
	Click here to	reset the br	OWSET USE	r interface s add custom t	ettings to their	defaults. M	ore on-sit	t <mark>e workshops</mark> a	available!	

#### Genome Browser – hg19 assembly <u>(sequences)</u>

ruary 2009 human reference sequence (GRCh37) was produced by the <u>Genome Reference</u> um. For more information about this assembly, see <u>GRCh37</u> in the NCBI Assembly database.

### position queries

a nacition can be aposified by the accession number of a paquaneed conomic alone an



# Where to search genome.ucsc.edu/cgi-bin/hgGateway

Genomes	nomes Genome Browser Tools Mirrors Downloads My		My Data	Help	About Us				
(Homo sa	apiens) Genome Br	owser Ga	teway						
	The UCSC ( Softw	Genome Brow vare Copyright	ser was creat (c) The Rege	ted by the <u>Genome I</u> ents of the University	Bioinformatics ( of California. /	Group of UC All rights rese	<u>Santa Cruz</u> . erved.		
group	genome	assembl	у	positio	n		search term		
mmal ᅌ	Human 📀 Feb	. 2009 (GRCh37	/hg19) ᅌ C	hr21:33,031,597	-33,041,570	brc			submit
<u>Cli</u>	<u>ck here to reset</u> the br	rowser user track search	interface s add custom t	ettings to their d	efaults. <u>M</u> configure tract	BRCA1 (Ho onset (BRC BRCA2 (Ho s onset (BRC	omo sapiens breast cancer 1, ear CA1), transcript variant 2, mRNA. omo sapiens breast cancer 2, ear CA2), mRNA.)	rly ) rly	
Genome	Browser – hg19 as	sembly <u>(s</u>	equences	s <u>)</u>		BRCC3 (He containing transcript v	omo sapiens BRCA1/BRCA2- complex, subunit 3 (BRCC3), rariant 1, mRNA.)	j	

bruary 2009 human reference sequence (GRCh37) was produced by the <u>Genome Reference</u> <u>ium</u>. For more information about this assembly, see <u>GRCh37</u> in the NCBI Assembly database.

### e position queries



# Where to search: Main Browser genome.ucsc.edu/cgi-bin/hgTracks



# **Public Hubs**





### Track Hubs

#### Human (Homo sapiens) Genome Browser Gateway



# Where to search genome.ucsc.edu/cgi-bin/hgHubConnect

Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us	
Track D	ata Hubs								

Track data hubs are collections of external tracks that can be imported into the UCSC Genome Browser. Hub tracks show up under own blue label bar on the main browser page, as well as on the configure page. For more information, see the <u>User's Guide</u>. To imp hub click its "Connect" button below.

NOTE: Because Track Hubs are created and maintained by external sources, UCSC is not responsible for their content.

Public Hubs	My Hubs	
Enter search ter	ms to find in public track hub description pages:	
		Search Public Hubs

Clicking Connect redirects to the gateway page of the selected hub's default assembly.

Display	Hub Name	Description	Assemblies
Connect	Roadmap Epigenomics Data Complete Collection at Wash U VizHub	Roadmap Epigenomics Human Epigenome Atlas Data Complete Collection, VizHub at Washington University in St. Louis	hg19
	Cancer genome polyA site &	An in-depth map of polyadenylation	1 . 10

### Track search

Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us	
Homo sapi	<i>ens</i> ) Genome B	rowser Ga	iteway					
	The UCSC Soft	Genome Brow ware Copyright	/ser was crea t (c) The Reg	ted by the <u>Genome</u> ents of the University	Bioinformatics Gr	oup of UC	<u>Santa Cruz</u> . rved.	
group	genome	assemb	ly	positio	n		search ter	m
mal ᅌ H	luman ᅌ Fe	b. 2009 (GRCh3)	7/hg19) ᅌ 🕻	chr21:33,031,597	-33,041,570	brc		submit
<u>Click</u>	<u>here to reset</u> the t	track search	interface s	settings to their d	efaults. <u>Mo</u> configure tracks	<b>re on-site</b> and display	e workshops	available!
Genomes	Genome Brow	ser Tool	s Mirro	ors Downloa	ds 🛛 My Da	ata V	/iew Help	About Us
UC: r17:41,196,3	move <<< <	e Brow < < > 81,189 bp.	ser on	Human Fo	eb. 2 Sess 3x 10x Trac ch terms Cust	sions k Hubs tom Tracks	3 	) Assembly 0x 100x 1 site workshops a
	Scale chr17 (q21.:	31) 13 41,210,000	1 17p12 17p1	1.2 q11.2 17q1 20 kb 00  41,230,000  Basic Gene Annota	2 41,240,000  tion Set from G	17q22     hg: 41,250,000 ENCODE Vers	24.325	.1 q25.3 41,270,000
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### Track search

Â	Genomes	Genome	e Browser	Tools	Mirrors	Downloads	My Data	Help	About Us			
Search for Tracks in the Human Feb. 2009 (GRCh37/hg19) Assembly												
[												
Sea	rch	Advanced										
search clear cancel												

#### About Track Search

Search for terms in track names, descriptions, groups, and ENCODE metadata. If multiple terms are entered, only tracks with all terms will be part of the results. more help

### Track search

	Â	Genomes	Genome Browser	Tools	Mirrors	Downloads	My Data	Help	About Us
S	earch fo	or Tracks i	n the Human Fe	eb. 2009 (C	GRCh37/hg	19) Assembl	у		
	Searc	ch Adv	vanced						
		Trac	k Name: co	ntains Methy	/lation				
		and Desc	cription: co	ntains					
		and Grou	ıp:	is Any			٥		
		and Data	Format:	is Any					<u> </u>
				o is ama					ENCODE terms
	+ an	Cell, tissu	ie or DNA sample	s and	H1-hESC	}	Cell, ti	ssue or DN	IA sample
	+ an	d Antibody	or target protein	ᅌ is amo	ng Any			Antibody o	r target protein
	search	n clear d	cancel						
Г		a lh illtr	Treak Norra						
Ľ			GM12878 GI	M12878 Me	thylation 450	K Bead Array f			
		ide 🗾	H1-hESC H	I-hESC Met	hylation 450	K Bead Array f	rom ENCODE	E/HAIB -	
(	Return to	Browser ((	) of 2 selected)						
-									

#### **About Track Search**