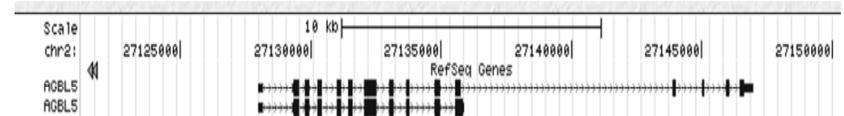




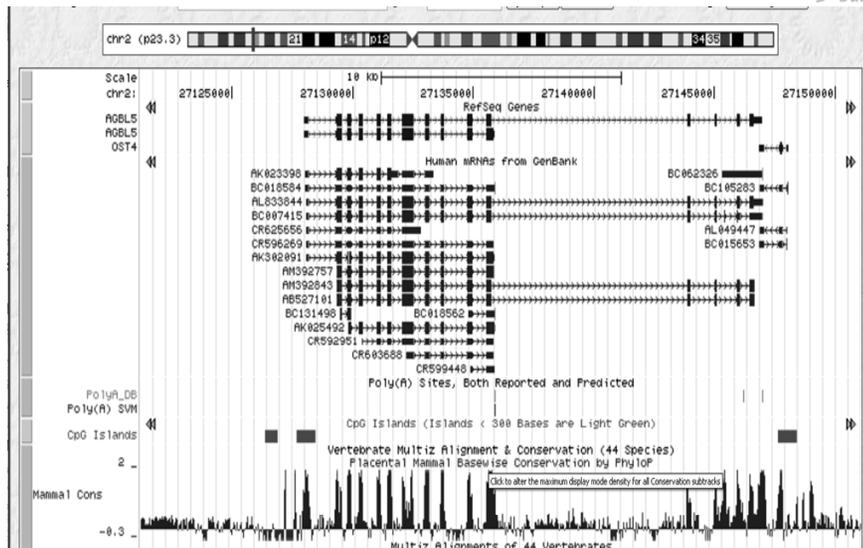
UCSC Genome Browser

Bingbing Yuan
May 20, 2010

How many isoforms AGLB5 has?



Comprehensive View



Genome Browser



- Ensembl Browser: <http://www.ensembl.org/index.html>
 - BLAST/BLAT: low similarity
 - Genetic Variation
- UCSC Browser: <http://genome.ucsc.edu/>
 - Easy to use
 - lots of tracks
- GBrowse: <http://gmod.org/wiki/Gbrowse>
 - private data
 - long setup & maintenance time
- IGV: <http://www.broadinstitute.org/igv/>
 - easy navigation
 - good for large data analysis

Ensembl genome browser 56: H.sapiens - Variation Image - Gene: TCF12 (ENSG00000140262)

Location: 15:57,210,896-57,216,911 Gene: TCF12 Transcript: TCF12-001 Variation: n35815435

Gene-based displays: Gene summary, Splice variants (5), Supporting evidence, Sequence, External references (3), Regulation, Comparative Genomics, Genomic alignments (5), Gene Tree (tree), Gene Tree (alignment), Orthologues (51), Paralogues (2), Protein families (13), Genetic Variation, Variation Table, Variation Image, External Data, Personal annotation, ID History, Configure this page, Manage your data, Export data, Bookmark this page.

Gene: TCF12 (ENSG00000140262)
Transcription factor 12 (Transcription factor HTF-4)(E-box-binding protein)(DNA-binding protein HTF4) [Source: UniProtKB/Swiss-Prot: Q95041]

Location: Chromosome 15: 57,210,823-57,582,051, forward strand.

Transcripts: There are 3 transcripts in this gene: [hide transcripts](#)

Name	Transcript ID	Protein ID	Description
TCF12-001	ENST00000267811	ENSP00000267811	protein_coding
TCF12-002	ENST00000333725	ENSP00000331057	protein_coding
TCF12-201	ENST00000343827	ENSP00000343450	protein_coding
TCF12-202	ENST00000438423	ENSP00000388840	protein_coding
TCF12-203	ENST00000452095	ENSP00000396881	protein_coding

Transcript and Gene level displays

In Ensembl a gene is made up of one or more transcripts. We provide displays at two levels:

- Transcript views which provide information specific to an individual transcript such as the cDNA and CDS sequences and protein domain annotation.
- Gene views which provide displays for data associated at the gene level such as orthologues and paralogs, regulatory regions and splice variants.

This view is a gene level view. To access the transcript level displays select a Transcript ID in the table above and then navigate to the information you want using the left hand side of the page. To return to viewing gene level information click on the Gene tab at the top of the page.

Variation Table Variation Image External Data

Variations: EnsemblVariation

INSITE profiles: P50014, P50015, P50016, P50017, P50018, P50019, P50020, P50021, P50022, P50023, P50024, P50025, P50026, P50027, P50028, P50029, P50030, P50031, P50032, P50033, P50034, P50035, P50036, P50037, P50038, P50039, P50040, P50041, P50042, P50043, P50044, P50045, P50046, P50047, P50048, P50049, P50050, P50051, P50052, P50053, P50054, P50055, P50056, P50057, P50058, P50059, P50060, P50061, P50062, P50063, P50064, P50065, P50066, P50067, P50068, P50069, P50070, P50071, P50072, P50073, P50074, P50075, P50076, P50077, P50078, P50079, P50080, P50081, P50082, P50083, P50084, P50085, P50086, P50087, P50088, P50089, P50090, P50091, P50092, P50093, P50094, P50095, P50096, P50097, P50098, P50099, P50100, P50101, P50102, P50103, P50104, P50105, P50106, P50107, P50108, P50109, P50110, P50111, P50112, P50113, P50114, P50115, P50116, P50117, P50118, P50119, P50120, P50121, P50122, P50123, P50124, P50125, P50126, P50127, P50128, P50129, P50130, P50131, P50132, P50133, P50134, P50135, P50136, 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P51637, P51638, P51639, P516

Navigate →

Drag-and-zoom →

Genome Viewer

Configure →

Tracks (group of data)

UCSC Genome Browser on Human Mar. 2006 (NCBI36/hg18) Assembly

UCSC Genes, Old UCSC Genes, All Events, RefSeq Genes, CDS, RefSeq Genes, pack, hide, alt, UCSC Genes, Old UCSC Genes, All Events, RefSeq Genes, CDS, RefSeq Genes, pack, hide, alt, Other RefSeq, MGC Genes, Other RefSeq Clusters, TransMap, Vega Genes, Ensembl Genes, hide, alt, AceView Genes, SIF Genes, N-SCAN, CONTRAST, SOP Genes, hide, alt, Genes on Genes, ExonOnly, AUGUSTAL, RNA Genes, ACES, hide, alt, ncRNA, Post-Seq Genes

mRNA and EST Tracks

Expression

Regulation

Comparative Genomics

Variation and Repeats

Plot ENCODE Regions and Genes

Plot ENCODE Transcription

Plot ENCODE Chromatin Immunoprecipitation

Plot ENCODE Chromatin Structure

Plot ENCODE Comparative Genomics and Variation

9

Home Genomes Genome Browser Blat Tables Gene Sorter

Configure Image

submit

image width: 800 pixels

label area width: 17 characters

text size: 8

- Display chromosome ideogram above main graphic
- Show light blue vertical guidelines
- Display labels to the left of items in tracks
- Display description above each track
- Show track controls under main graphic
- Next/previous item navigation
- Next/previous exon navigation
- Enable track re-ordering
- Enable advanced javascript features

← ⏪ ⏩

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UCSC Genes, Old UCSC Genes, All Events, RefSeq Genes, CDS, RefSeq Genes, pack, hide, alt, UCSC Genes, Old UCSC Genes, All Events, RefSeq Genes, CDS, RefSeq Genes, pack, hide, alt, Other RefSeq, MGC Genes, Other RefSeq Clusters, TransMap, Vega Genes, Ensembl Genes, hide, alt, AceView Genes, SIF Genes, N-SCAN, CONTRAST, SOP Genes, hide, alt, Genes on Genes, ExonOnly, AUGUSTAL, RNA Genes, ACES, hide, alt, ncRNA, Post-Seq Genes

mRNA and EST Tracks

Expression

Regulation

Comparative Genomics

Variation and Repeats

Plot ENCODE Regions and Genes

Plot ENCODE Transcription

Plot ENCODE Chromatin Immunoprecipitation

Plot ENCODE Chromatin Structure

Plot ENCODE Comparative Genomics and Variation

Track Mode

11

mode of an individual annotation track:

Hide: the track is not displayed at all.

Dense: the track is displayed with all features collapsed into a single line.

Squish: the track is displayed with each annotation feature shown separately, but at 50% the height of full mode. Features are unlabeled.

Pack: the track is displayed with each annotation feature shown separately and labeled.

Full: the track is displayed with each annotation feature on a separate line.

Human ESTs

Human ESTs Including Unspliced

Human ESTs Including Unspliced

Human ESTs Including Unspliced

12



Track Setting

Neanderthal Assembly and Analysis refresh

Variation and Repeats refresh

Common Cell CNV GIS DNA PET SNPs (130) SNPs (129) SNPs (128) SNPs (126)

hide hide dense hide hide hide hide

SNP Arrays HGDP Allele Freq HGDP Smoothd FST HGDP Hetzygsty HGDP iHS HGDP XP-EHH

dense hide [No data-chrX] [No data-chrX] hide [No data-chrX]

HapMap SNPs HapMap LD Phased Tajima's D SNPs Tajima's D HGSV Discordant Segmental Dups

hide [No data-chrX] hide hide hide hide

Structural Var Exapted Repeats RepeatMasker RepMask 3.2.7 Interrupted Rpts Intr Rpts 3.2.7

hide hide dense hide hide hide

Simple Repeats Microsatellite Self Chain Genome Variants

hide hide hide hide

Pilot ENCODE Regions and Genes refresh

Pilot ENCODE Transcription refresh

13

Simple Nucleotide Polymorphisms (dbSNP build 130)



Display mode:

Include Chimp state and observed human alleles in name:
(If enabled, chimp allele is displayed first, then "?", then human alleles)

On details page, show function and coding differences relative to:

- UCSC Genes Old UCSC Genes Gencode Manual Gencode Auto
 Gencode PolyA CCDS RefSeq Genes Other RefSeq
 Vega
 SIB C
 Gene
 Augu

Minimum
Maximum

SNP Feature for Color Specification:

The selected feature above has the following values below. For each value, a selection of colors is available. If a SNP has more than one of these properties, resulting in more than one color, then the stronger color will override the weaker color. In order from strongest to weakest, the colors are red, green, blue, gray, black.

Unknown: Locus: Coding - Synonymous: Coding - Non-Synonymous:
 Untranslated: Intron: Splice Site:

[View table schema](#)

Data last updated: 2009-08-18

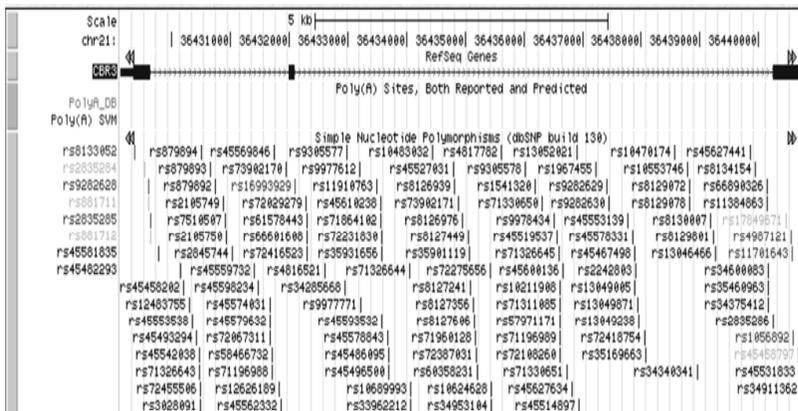
Description

This track contains information about single nucleotide polymorphisms and small insertions and deletions (indels) — collectively Simple Nucleotide Polymorphisms — from dbSNP build 130, available from <ftp.ncbi.nih.gov/snp>.

Interpreting and Configuring the Graphical Display

Variants are shown as single tick marks at most zoom levels. When viewing the track at or near base-level resolution, the displayed width of the SNP corresponds to the width of the variant in the reference sequence. Insertions are indicated by a single tick mark displayed between two nucleotides, single nucleotide polymorphisms are displayed as the width of a single base, and multiple nucleotide variants are represented by a block that spans two or more bases.

14



15

Simple Nucleotide Polymorphisms (dbSNP build 130)

dbSNP build 130 rs4987121

dbSNP: rs4987121
Position: chr21:36440549-36440549
Band: 21a22.12

Gen:
View:
Sun:
Str:
Obs:
Ref:
Chi:
Ora:
Ma:
Class: single
Validation: by-cluster,by-frequency,by-hapmap
Function: missense
Molecule Type: genomic
Average Heterozygosity: 0.015 +/- 0.084
Weight: 1

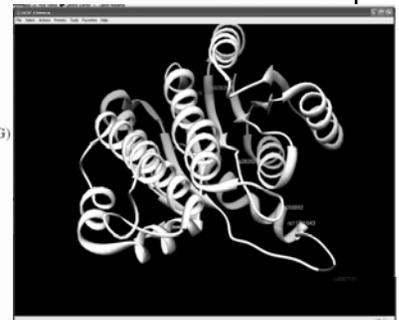
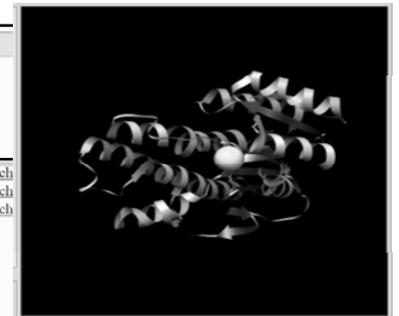
Coding annotations by dbSNP:
NM_001236: missense M (ATG) -> L (TTG)

UCSC's predicted function relative to selected gene tracks:
UCSC Genes AB004851 (uc002yvf.1) intron
UCSC Genes CBR3 (uc002yve.1) missense M (ATG) -> L (TTG)
UCSC Genes BC047014 (uc002yvd.1) intron
UCSC Genes CR594732 (uc002yvc.1) intron

HapMap SNP

Mappings to PDB protein structures

[Chimera help](#)



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Home Genomes Blat Tables Gene Sorter Session FAQ Help

UCSC In-Silico PCR

Genome: Assembly: Target: Forward Primer: Reverse Primer:

Max Product Size: Min Perfect Match: Min Good Match: Flip Reverse Primer:

UCSC In-Silico PCR Genome browser

```
>chr22:34304505+34304954 450bp TAACAGATTGATGATGCATGAAATGGG CCATGAGTGGCTCCTAAAGCAGCTGC
TtACAGATTGATGATGCATGAAATGGGgggtggccagggtgggggggga
gactgcagagaaggcagggtggttcataacaagcttctgctgccca
tatgacagctgaagtttccaggggctgatggtgagccagtgagggtaa
taacaagaacctcctagagaacctcctccttaagatataaaataa
gacttgcgtctgtaaggatggattatcctattgagaattctgtta
tccagaatggottaacccacaatgctgaaaagtgtgtaacgtaataoaa
agcaagctcctcctcagacagagaacaccagcctcaccaggaagcaaa
aaattggcttcaactttaaagtgaaatccagaccagatgacagagctcc
aagcacttgcctcagctccacGCAGCTGCTTAGGAGCCACTCATGgG
```

Primer Melting Temperatures

Forward: 66.7 C taacagattgatgcatgcatgaaatggg
Reverse: 73.8 C cccatgagtggtccttaagcagctgc
The temperature calculations are done assuming 50 mM salt and 50 nM annealing oligo concentration. The code to calculate the melting temp comes from Primer3.



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

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

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

position/search chr22:34,304,505-34,304,954 gene jump clear size 450 bp. configure

chr22 (at2.3)

Encyclopedia of DNA Elements (ENCODE) Consortium
 Founded by National Human Genome Research Institute (NHGRI)



Expression

<input type="checkbox"/> Affy Exon	<input type="checkbox"/> Affy GNF1H	<input checked="" type="checkbox"/> Affy RNA Loc	<input type="checkbox"/> Affy U133	<input type="checkbox"/> Affy U133Plus2	<input type="checkbox"/> Affy U95
<input type="checkbox"/> Agilent Array	<input type="checkbox"/> Allen Brain	<input type="checkbox"/> Bertone Yale	<input checked="" type="checkbox"/> Caltech RNA-seq	<input checked="" type="checkbox"/> CSHL Long RNA-seq	<input checked="" type="checkbox"/> CSHL Sm RNA-seq
<input type="checkbox"/> GIS PET RNA	<input type="checkbox"/> ONE Atlas 2	<input type="checkbox"/> ONE Ratio	<input checked="" type="checkbox"/> Helicos RNA-seq	<input type="checkbox"/> Illumina WG-6	<input type="checkbox"/> RIKEN CAGE Loc
<input type="checkbox"/> Sentin Brain	<input checked="" type="checkbox"/> UW Affy Exon	<input type="checkbox"/> Affy Tss			

Regulation

<input checked="" type="checkbox"/> Broad Histone	<input type="checkbox"/> ChIP-Seq	<input type="checkbox"/> EIO/PCV LNAS	<input type="checkbox"/> Epigenome TSS	<input type="checkbox"/> FROTH	<input type="checkbox"/> GIS ChIP-PET
<input checked="" type="checkbox"/> JHAR Methylation	<input type="checkbox"/> HAIB Methylation	<input type="checkbox"/> HAIB TFBS	<input type="checkbox"/> NHGRI Bi-Pro	<input type="checkbox"/> NHGRI SRE	<input type="checkbox"/> Open Chromatin
<input type="checkbox"/> OReg Anno	<input checked="" type="checkbox"/> SUNY RBP	<input type="checkbox"/> SwitchGear TSS	<input type="checkbox"/> TFBS Conserved	<input type="checkbox"/> TSS mRNA sites	<input type="checkbox"/> UW DNase-Seq
<input checked="" type="checkbox"/> UW Histone	<input type="checkbox"/> Vista Enhancer	<input type="checkbox"/> Yale TFBS	<input type="checkbox"/> TX Reg Potential	<input type="checkbox"/> FOX2 CLIP-seq	<input type="checkbox"/> LUCISD
<input type="checkbox"/> NIE Nbc Lamma	<input type="checkbox"/> Nucleosome Occupancy	<input type="checkbox"/> Upreals ChIP			

Comparative Genomics

Neanderthal Assembly and Analysis

Variation and Repeats



Broad Histone Track Settings

ENCODE Histone Modifications by Broad Institute ChIP-seq

Maximum display mode:

Select views (help): Signal Full

Signal Configuration

Type of graph: Track height: pixels (range: 16 to 100)
 Vertical viewing range: min: max: (range: 0 to 15647)
 Data view scaling: Use vertical viewing range setting Always include zero: OFF
 Transform function: Transform data points by:
 Windowing function: Smoothing window: pixels
 Draw y indicator lines: at y = 0.0: OFF at y = 0: OFF

Select subtracks by cell line and antibody:

Antibody	GM12878	HL-MESC	HepG2	HMEC	HSM	HUVEC	K562	NHEK	NHLF	Cell Line	Antibody
CTCF	<input checked="" type="checkbox"/>	<input type="checkbox"/>	CTCF								
H3K4me1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K4me1	
H3K4me2	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K4me2	
H3K4me3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K4me3	
H3K9ac	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K9ac	
H3K9me1	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K9me1	
H3K27ac	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K27ac	
H3K27me3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K27me3	
H3K36me3	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	H3K36me3	



List subtracks: only selected/visible all

Cell Line ¹	Antibody ²	Views ³	Restricted Until
<input type="checkbox"/> GM12878	CTCF	Peaks	ENCODE Histone Mods, Broad ChIP-seq Peaks (CTCF, GM12878) ... schema 2009-10-05
<input type="checkbox"/> GM12878	CTCF	Signal	ENCODE Histone Mods, Broad ChIP-seq Signal (CTCF, GM12878) ... schema 2009-10-05
<input type="checkbox"/> GM12878	H3K4me1	Peaks	ENCODE Histone Mods, Broad ChIP-seq Peaks (H3K4me1, GM12878) ... schema 2009-10-05
<input type="checkbox"/> GM12878	H3K4me1	Signal	ENCODE Histone Mods, Broad ChIP-seq Signal (H3K4me1, GM12878) ... schema 2009-10-05
<input type="checkbox"/> GM12878	H3K4me2	Peaks	ENCODE Histone Mods, Broad ChIP-seq Peaks (H3K4me2, GM12878) ... schema 2009-10-05

<input type="checkbox"/> NHLF	H4K20me1	Signal	ENCODE Histone Mods, Broad ChIP-seq Signal (H4K20me1, NHLF) ... schema 2010-06-28
<input type="checkbox"/> NHLF	Input Control	Signal	ENCODE Histone Mods, Broad ChIP-seq Signal (NHLF control) ... schema 2010-06-29

24 of 177 selected

Submit

Downloads

Data version: through the ENCODE Jan 2010 Freeze

RESTRICTED

until	File	Size	Submitted	Details
2009-10-05	wgEncodeBroadChIPseqPeaksGm12878CtcfBroadPeak.gz	363K	2009-01-05	cell=GM12878; dataType=ChIPSeq; antibody=CTCF; lab=Broad; type=Peaks
2009-10-05	wgEncodeBroadChIPseqRawDataRep1Gm12878CtcfFastq.gz	774M	2009-01-05	cell=GM12878; dataType=ChIPSeq; antibody=CTCF; lab=Broad; type=Signal
2009-10-05	wgEncodeBroadChIPseqRawDataRep2Gm12878CtcfFastq.gz	517M	2009-01-05	cell=GM12878; dataType=ChIPSeq; antibody=CTCF; lab=Broad; type=Signal

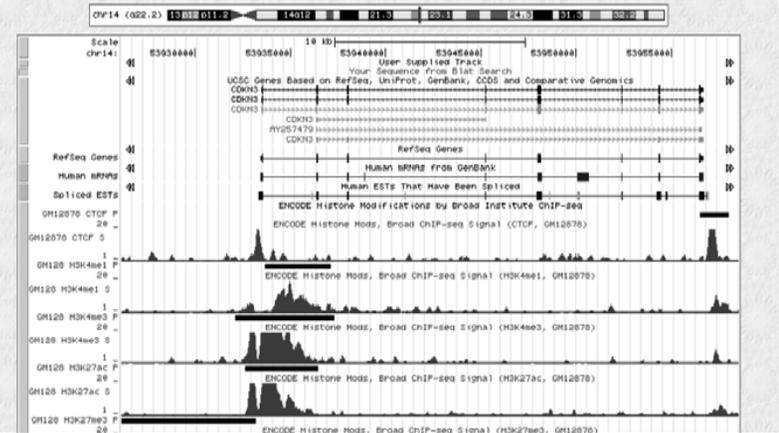
25



UCSC Genome Browser on Human Mar. 2006 (NCBI36/hg18) Assembly

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

position/search chr14:53,926,128-53,958,540 gene jump clear size 32,413 bp configure



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References



OpenHelix: <http://www.openhelix.com/ucsc/>

MIT library: [Bioinformatics Tutorial Series](http://libguides.mit.edu/bits)
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Tyra Wolfsberg, Ph.D. [Current Topics in Genome Analysis 2010](#)
<http://www.youtube.com/watch?v=7BN0T7AQqmY>

[ENCODE whole-genome data in the UCSC Genome Browser](#). *Nucleic Acids Res.* 2010 Jan;38(Database issue):D620-5. Epub 2009 Nov 17.

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[The UCSC Genome Browser Database: 2008 update](#). *Nucleic Acids Res.* 2008 Jan;36:D773-9.

UCSC Genome Browser Wiki site:
http://genomewiki.ucsc.edu/index.php/Main_Page

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