

BMI206 UCSC browser lab

TUTORIAL

UCSC BROWSER (<http://genome.ucsc.edu>)

Genomes: Allows you to view your region/gene/SNP/BAC etc. of interest in your genome of interest.

BLAT: Allows you to search where a sequence hits the genome.

DNA: Provides you with the DNA sequence of the window you are looking at.

Tables: Generates tables based on your specifications. For more info on how to use Tables go to:

<http://genome.ucsc.edu/goldenPath/help/hgTablesHelp.html>

PCR: Does an electronic PCR on the genome with primers that you input.

Convert: Converts genomic coordinates between different assemblies and genomes.

Add Custom Tracks: Allows you to insert your own custom track. To insert custom tracks and learn more on how to generate custom tracks go to:

<http://genome.ucsc.edu/cgi-bin/hgCustom?hgHubConnect.destUrl=.%2Fcgi-bin%2FhgTracks&clade=mammal&org=Human&db=hg19&position=chr4%3A123%2C747%2C863-123%2C819%2C390&hgt.suggest=&hgt.suggestTrack=knownGene&hgsid=233853131&hgt.newJQuery=1>

Tracks covered

-UCSC genes: <http://genome.ucsc.edu/cgi-bin/hgTrackUi?hgsid=233846161&c=chr4&g=knownGene>

-RefSeq genes: <http://genome.ucsc.edu/cgi-bin/hgTrackUi?hgsid=233846161&c=chr4&g=refGene>

-Conservation: <http://genome.ucsc.edu/cgi-bin/hgTrackUi?hgsid=233846161&c=chr4&g=cons46way>

-Repeat masker: <http://genome.ucsc.edu/cgi-bin/hgTrackUi?hgsid=233846161&c=chr4&g=rmsk>

-Common SNPs(135): <http://genome.ucsc.edu/cgi-bin/hgTrackUi?hgsid=233846161&c=chr4&g=snp135Common>

For more help and tutorials on the UCSC browser see the following:

Genome browser user guide: <http://genome.ucsc.edu/goldenPath/help/hgTracksHelp.html>

Genome browser intro tutorial: <http://www.openhelix.com/cgi/tutorialInfo.cgi?id=27>

Custom tracks & table tutorial: <http://www.openhelix.com/cgi/tutorialInfo.cgi?id=28>

Other websites covered

OMIM (<http://www.ncbi.nlm.nih.gov/omim>): Compendium of human genes and genetic phenotypes.

dbSNP (<http://www.ncbi.nlm.nih.gov/projects/SNP>): Compendium of human SNPs.

LAB EXERCISES

Coding Sequence

Sonic Hedgehog (*SHH*)

1. Where did the name for this gene come from (hint: RefSeq track)?
2. What human diseases are caused by mutations in *SHH*? What's its OMIM number?
3. What chemicals interact with *SHH*?
4. What is the PDB code that ModBase predicts its structure for?
5. What is the 'Molecular Function' Gene Ontology (GO) term for *SHH*?
6. To what other genes does the *SHH* mRNA sequence have similarity to?

PROBLEMS 7 & 8 ARE HOMEWORK:

7. Make a table of the genomic coordinates of *SHH* 'RefSeq genes' exons.
8. Make a track that shows the regions overlapping between the *SHH* 'RefSeq' gene and human mRNAs.

Noncoding Sequences

FORWARD: TCATTGTAATTCATAGACCATGCTG
REVERSE: AAAGTTAGAAGATCAAACGAAAGAGG

9. What is the genomic region that these primers pick up?
10. What would be the genomic region in the March 2006 (hg18) assembly?
11. Is this sequence evolutionarily conserved?
12. How many repeats are within this sequence and what are they?
13. How many SNPs are in this region and what's their name?
14. What is the minor allele frequency of this SNP/s?

PROBLEMS 15 & 16 ARE HOMEWORK:

15. Does the sequence overlap an EnhancerFinder predicted brain enhancer? What is its score? (You will need to make a custom track from the brain BED file of EnhancerFinder predictions from Data File S1 in Erwin et al. Note: this file is in hg19 coordinates.)
16. Was this sequence tested for enhancer activity by the VISTA Enhancer Browser project? If so, in which tissues is it active?