

UCSC genome browser

Tutorial 9

Sequence data

Variants

Annotations

Expression data

Genes

Regulation

Different
Organisms

Genome Browsers

(Just a few) examples of genome browsers

- UCSC Genome Browser - <http://genome.ucsc.edu/>



- Ensembl - <http://www.ensembl.org/index.html>



- NCBI - <https://www.ncbi.nlm.nih.gov/>



UCSC Genome Browser



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Genome Browser

UCSC - overview

- A website that consists of a of free on-line tools that can be used to browse, analyze, and query genomic data. It is a display engine for genomic annotations.
- Many of the annotation tracks are **submitted by scientists** worldwide; others are computed by the UCSC Genome Bioinformatics group from **publicly available sequence data**.
- You can also upload your own data.



Genomes

Genome Browser

Tools

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Our tools

- [Genome Browser](#)
interactively visualize genomic data
- [COVID-19 Research](#)
use the SARS-CoV-2 genome browser and explore coronavirus datasets
- [BLAT](#)
rapidly align sequences to the genome
- [Table Browser](#)
download data from the Genome Browser database
- [Variant Annotation Integrator](#)
get functional effect predictions for variant calls
- [Data Integrator](#)
combine data sources from the Genome Browser database
- [Genome Browser in a Box \(GBiB\)](#)
run the Genome Browser on your laptop or server
- [In-Silico PCR](#)
rapidly align PCR primer pairs to the genome
- [LiftOver](#)
convert genome coordinates between assemblies
- [Track Hubs](#)
import and view external data tracks
- [REST API](#)
returns data in JSON format

[More tools...](#)



Genomes

Genome Browser

Tools

Mirrors

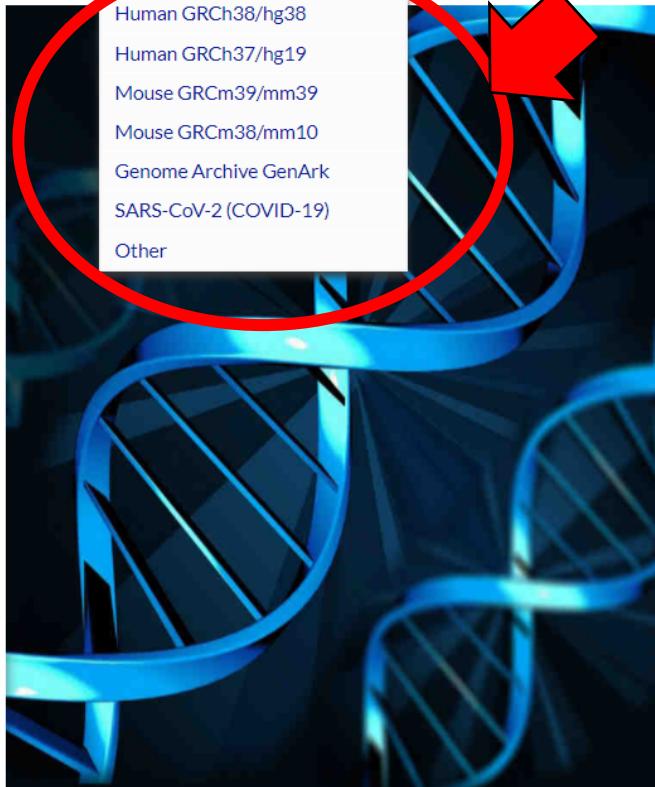
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Our tools

- **Genome Browser**
interactively visualize genomic data
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use the SARS-CoV-2 genome browser and explore coronavirus datasets
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run the Genome Browser on your laptop or server
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rapidly align PCR primer pairs to the genome
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convert genome coordinates between assemblies
- **Track Hubs**
import and view external data tracks
- **REST API**
returns data in JSON format

[More tools...](#)

- The genomes of many organisms are constantly updating.
 - Note that the reference genome and its annotation must be from the same version.

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Genome Browser Gateway



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Browse>Select Species

POPULAR SPECIES



Human



Mouse



Rat



Fruitfly



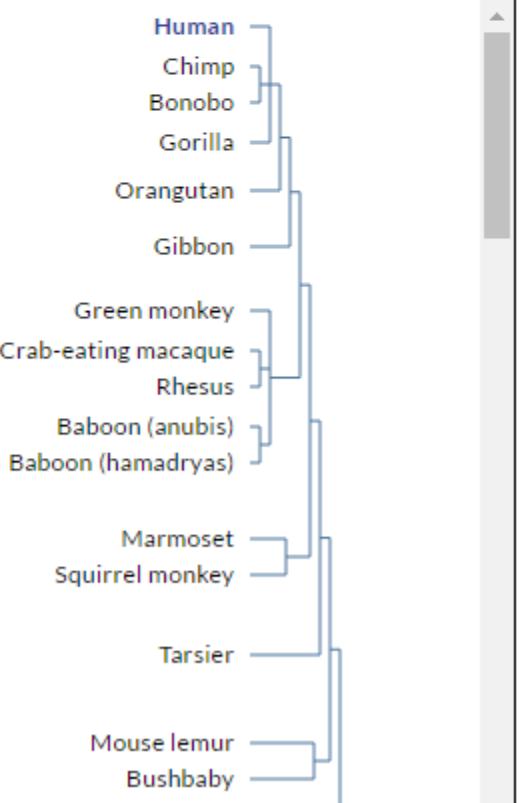
Worm



Yeast

Enter species or common name

REPRESENTED SPECIES



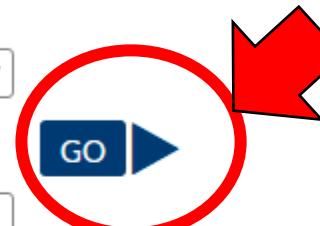
Find Position

Human Assembly

Dec. 2013 (GRCh38/hg38)



GO



Position/Search Term

Enter position, gene symbol or search terms

Current position: chr9:133,252,000-133,280,861



Human Genome Browser - hg38 assembly

UCSC Genome Browser assembly ID: hg38

Sequencing/Assembly provider ID: GRCh38 Genome Reference Consortium

Assembly date: Dec. 2013

GenBank accession ID: GCA_000001305.2

NCBI Genome information: NCBI genome/51 (Homo sapiens)

NCBI Assembly information: NCBI assembly/883148 (GRCh38/GCA_000)

BioProject information: NCBI Bioproject: 31257

Search the assembly:

- **By position or search term:** Use the "position or search term" box to enter a genomic position, gene name, marker names; or keywords from the GenBank description of an mRNA or protein.
- **By gene name:** Type a gene name into the "search term" box, choose a species, and click "Search".
- **By track type:** Click the "track search" button to find Genome Browser tracks.

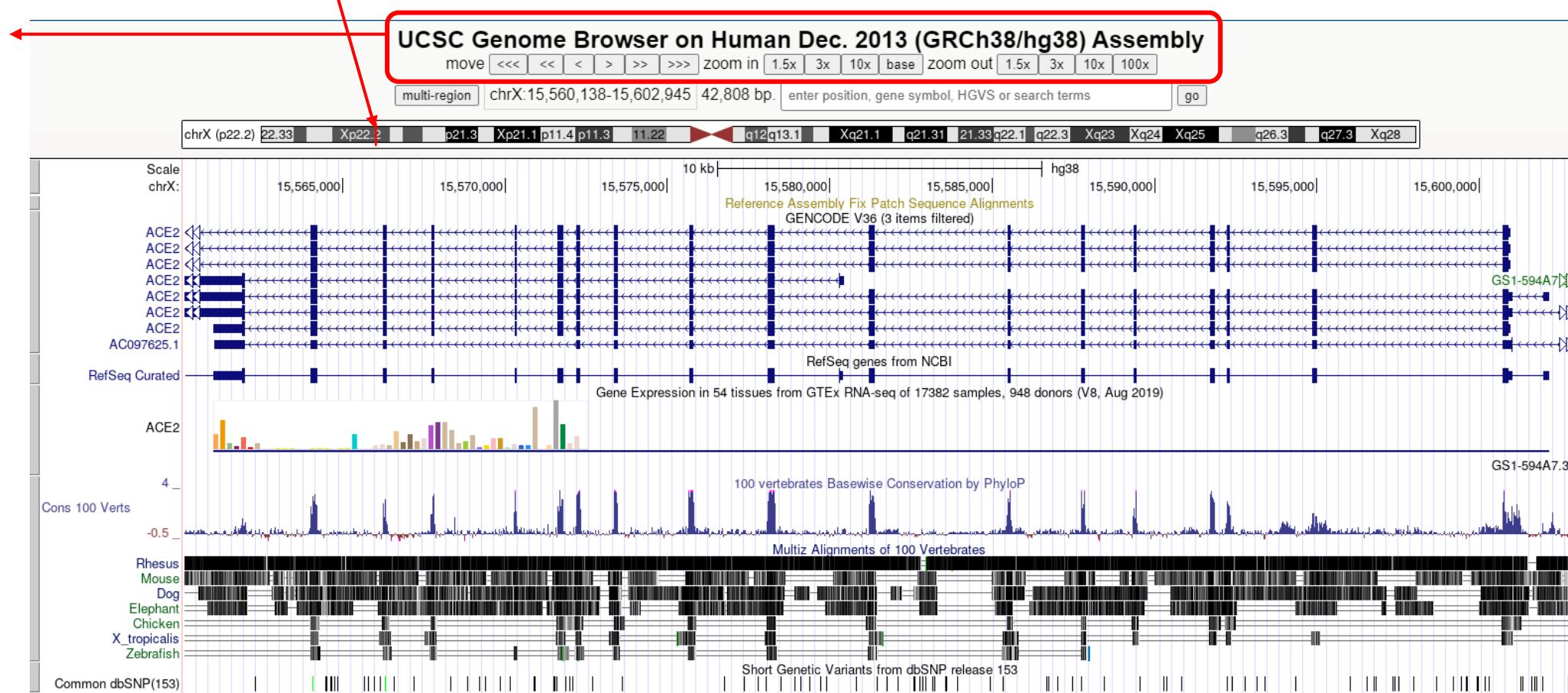
Download sequence and annotation data:

- **Using rsync (recommended)**

What can we view in the browser

Navigation in the sequence

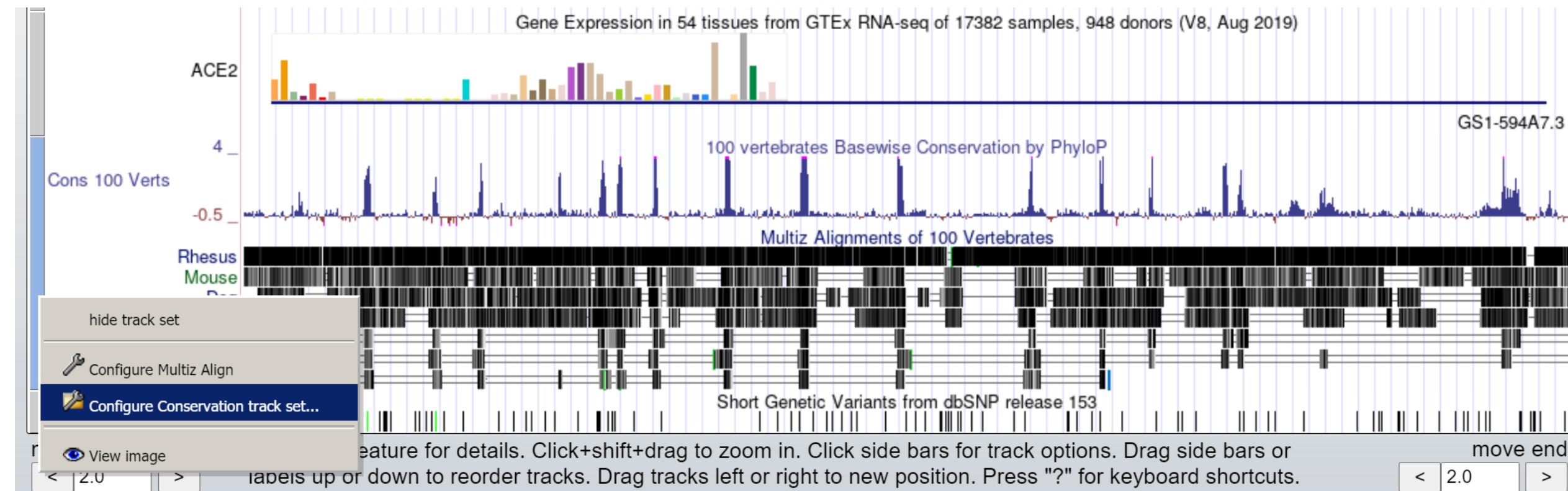
Position in the chromosome



Gene expression
Conservations
compared to
other organisms

SNPs

What can we view in the browser



Conservation Track Settings

[Subtracks](#) [Description](#)

Vertebrate Multiz Alignment & Conservation (100 Species) ([^All Comparative Genomics tracks](#))

Maximum display mode: full [Submit](#) [Cancel](#) [Reset to defaults](#)

Select views ([Help](#)):

[Multiz Alignments](#) pack [Baseswise Conservation \(phyloP\)](#) full [Element Conservation \(phastCons\)](#) hide [Conserved Elements](#) hide

Multiz Alignments Configuration

Species selection: [+](#) [-](#) [Defaults](#)

Primate [+](#) [-](#)

chimp gorilla orangutan gibbon rhesus
 crab-eating macaque baboon green monkey marmoset squirrel monkey
 bushbaby

Euarchontoglires [+](#) [-](#)

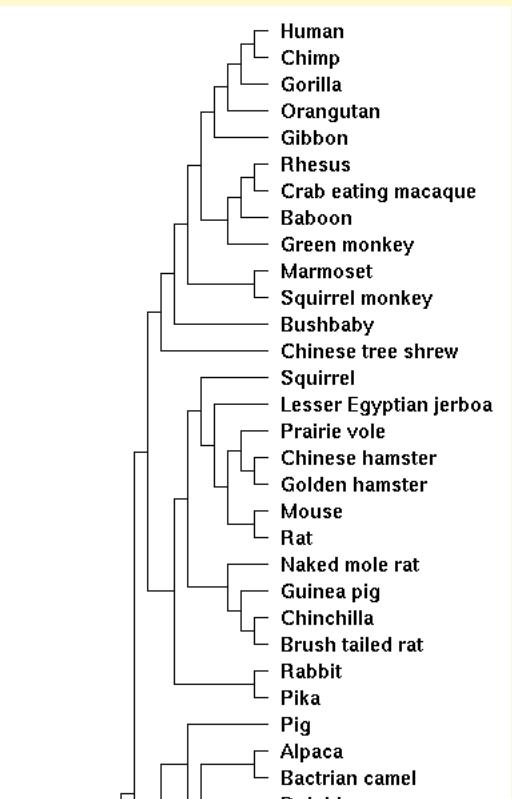
chinese tree shrew squirrel lesser Egyptian jerboa prairie vole chinese hamster
 golden hamster mouse rat naked mole-rat guinea pig
 chinchilla brush-tailed rat rabbit pika

Laurasiatheria [+](#) [-](#)

pig alpaca bactrian camel dolphin killer whale
 tibetan antelope cow sheep domestic goat horse
 white rhinoceros cat dog ferret panda
 pacific walrus weddell seal black flying-fox megabat david's myotis (bat)
 microbat big brown bat hedgehog shrew star-nosed mole

Afrotheria [+](#) [-](#)

elephant cape elephant shrew manatee cape golden mole tenrec
 aardvark



What can we view in the browser

At the bottom of the browser, you can find many more tracks that you can view along the genome.

For details. Click+shift+drag to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag track labels to move them. "?" for keyboard shortcuts.

track search default tracks default order hide all add custom tracks track hubs configure reverse resize refresh

collapse all expand 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.

+	Mapping and Sequencing	refresh
+	Genes and Gene Predictions	refresh
+	Phenotype and Literature	refresh
+	COVID-19	refresh
+	mRNA and EST	refresh
+	Expression	refresh
+	Regulation	refresh
+	Comparative Genomics	refresh
+	Variation	refresh
-	Repeats	refresh

Zoom in

Click and drag
in the ruler
area

The screenshot shows the UCSC Genome Browser interface for the Human Dec. 2013 (GRCh38/hg38) Assembly. A red arrow points from the text "Click and drag in the ruler area" to the top ruler of the genome track. A blue box highlights the "Drag-and-select" dialog box, which is overlaid on the browser window. The dialog box contains the following text:

- Hold **Shift+drag** to show this dialog
- Hold **Alt+drag** to add a highlight
- Hold **Ctrl+drag** (Windows) or **Cmd+drag** (Mac) to zoom
- To cancel, press **Esc** anytime or drag mouse outside image
- Highlight the current position with **h** then **m**
- Clear all highlights with View - Clear Highlights or **h** then **c**

Highlight color: #aaedff [Reset](#)

Don't show this again and always zoom with shift.
Re-enable via 'View - Configure Browser' (c then f)

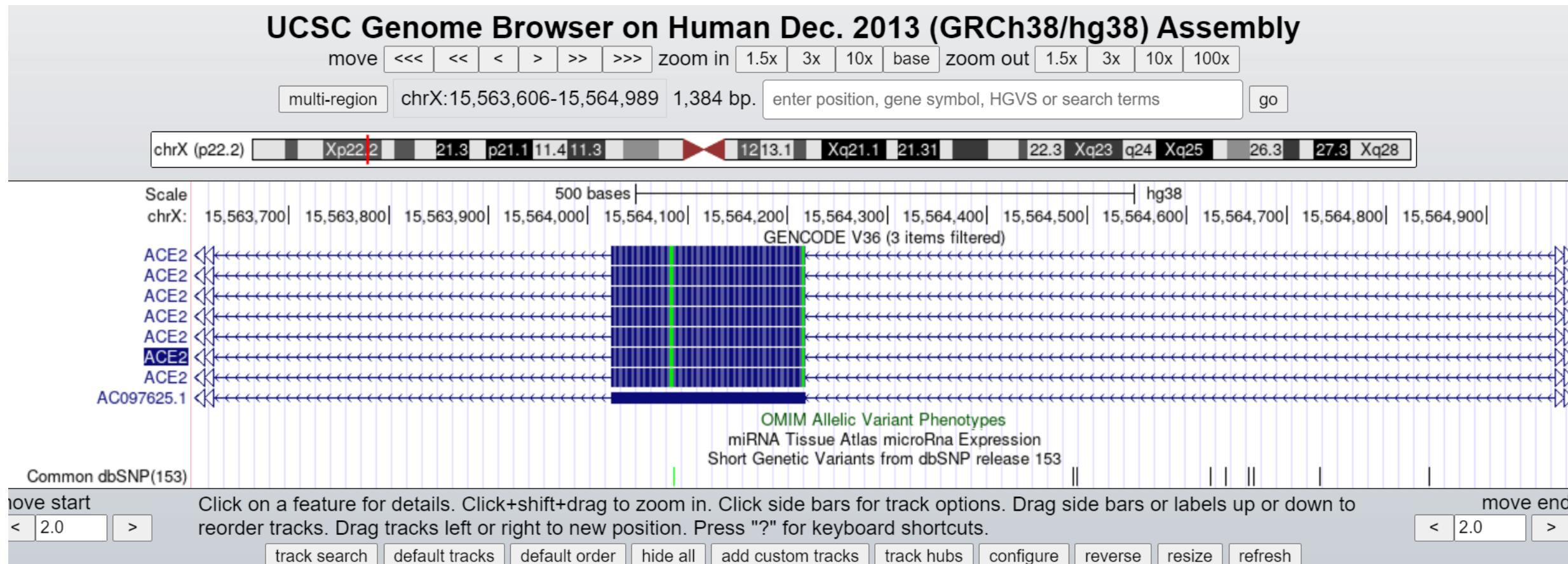
Selected chromosome position: chrX:15563606-15564989

Buttons at the bottom of the dialog: **Zoom In**, **Single Highlight**, **Add Highlight**, **Cancel**.

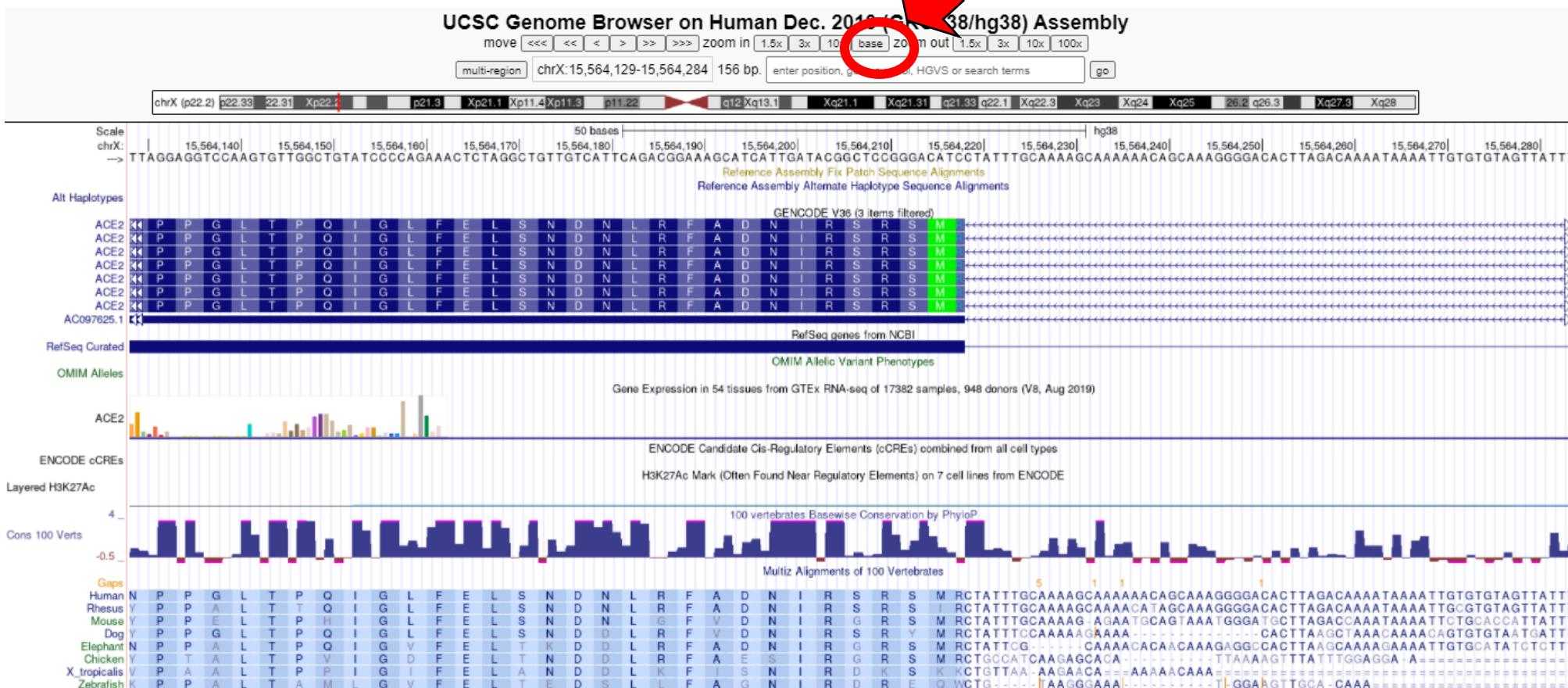
Below the dialog, a note says: Click on a feature for details. Click+shift+drag to zoom in. Click side bars for track options. Drag side bars or labels up or down to reorder tracks. Drag tracks left or right to new position. Press "?" for keyboard shortcuts.

At the very bottom, there are buttons for: track search, default tracks, default order, hide all, add custom tracks, track hubs, configure, reverse, resize, and refresh.

You can zoom in to view the base-pair level



You can zoom in to view the base-pair level



DNA sequence

Protein sequences

Conservation at the base level

Click on the gene for more information

[!\[\]\(fc5cbba58387ad7b76029814c568029c_img.jpg\) Genomes](#) [Genome Browser](#) [Tools](#) [Mirrors](#) [Downloads](#) [My Data](#) [Projects](#) [Help](#) [About Us](#)

Human Gene ACE2 (ENST00000677282.1) Description and Page Index

Description: angiotensin I converting enzyme 2 (from HGNC ACE2)

RefSeq Summary (NM_001371415): The protein encoded by this gene belongs to the angiotensin-converting enzyme family of dipeptidyl carboxydiptidases and has considerable homology to human angiotensin 1 converting enzyme. This secreted protein catalyzes the cleavage of angiotensin I into angiotensin 1-9, and angiotensin II into the vasodilator angiotensin 1-7. The organ- and cell-specific expression of this gene suggests that it may play a role in the regulation of cardiovascular and renal function, as well as fertility. In addition, the encoded protein is a functional receptor for the spike glycoprotein of the human coronavirus HCoV-NL63 and the human severe acute respiratory syndrome coronaviruses, SARS-CoV and SARS-CoV-2 (COVID-19 virus). [provided by RefSeq, Mar 2020].

Gencode Transcript: ENST00000677282.1

Gencode Gene: ENSG00000130234.12

Transcript (Including UTRs)

Position: hg38 chrX:15,557,039-15,580,438 **Size:** 23,400 **Total Exon Count:** 11 **Strand:** -

Coding Region

Position: hg38 chrX:15,561,905-15,580,312 **Size:** 18,408 **Coding Exon Count:** 11

Page Index	Sequence and Links	MalaCards	CTD	RNA-Seq Expression	Microarray Expression
RNA Structure	Other Species	mRNA Descriptions	Pathways	Other Names	Methods

Data last updated at UCSC: 2021-01-14 15:32:12

Sequence and Links to Tools and Databases

Genomic Sequence (chrX:15,557,039-15,580,438)	mRNA (may differ from genome)	Protein (459 aa)
Gene Sorter	Genome Browser	Other Species FASTA
CGAP	Ensembl	ExonPrimer
MGI	PubMed	Wikipedia

Click on the gene for more information

MalaCards Disease Associations

MalaCards Gene Search: [ACE2](#)

Diseases sorted by gene-association score: [severe acute respiratory syndrome](#) (28), [neurogenic hypertension](#) (18), [hartnup disorder](#) (14), [tetanus neonatorum](#) (9), [internal hemorrhoid](#) (9), [intracranial aneurysm](#) (8), [posterior urethral valves](#) (6), [hypertension, essential](#) (2), [myocardial infarction](#) (1)

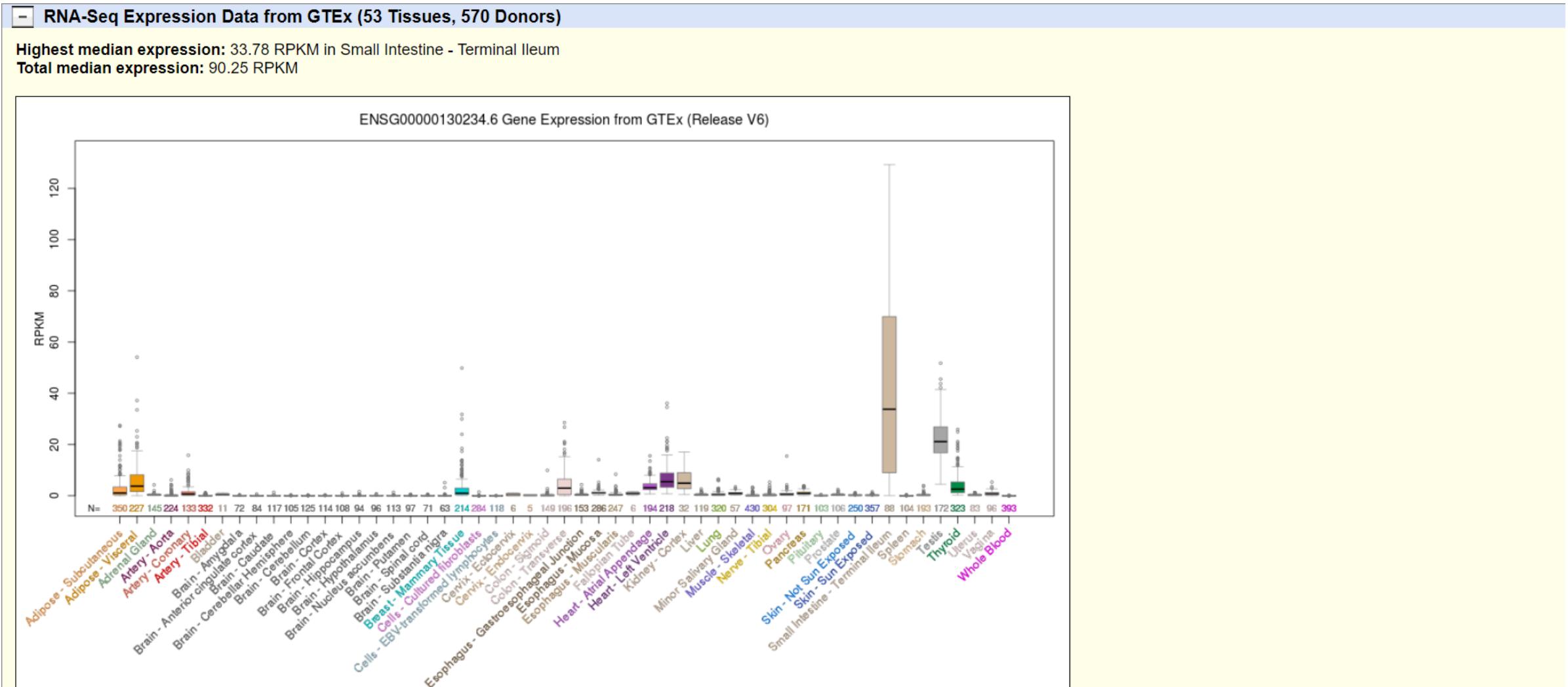
Comparative Toxicogenomics Database (CTD)

The following chemicals interact with this gene

- [D019327](#) Copper Sulfate
- [D007545](#) Isoproterenol
- [D013311](#) Streptozocin
- [C111118](#) 2',3,3',4',5-pentachloro-4-hydroxybiphenyl
- [D015124](#) 8-Bromo Cyclic Adenosine Monophosphate
- [D001205](#) Ascorbic Acid
- [D001623](#) Betamethasone
- [D019256](#) Cadmium Chloride
- [D016572](#) Cyclosporine
- [C556151](#) DX600 peptide

more ... [click here to view the complete list](#)

Click on the gene for more information



Looking for a specific gene

Genome Browser Gateway

Mirrors Downloads My Data Help About Us

Find Position

Human Assembly
Dec. 2013 (GRCh38/hg38)

Yeast

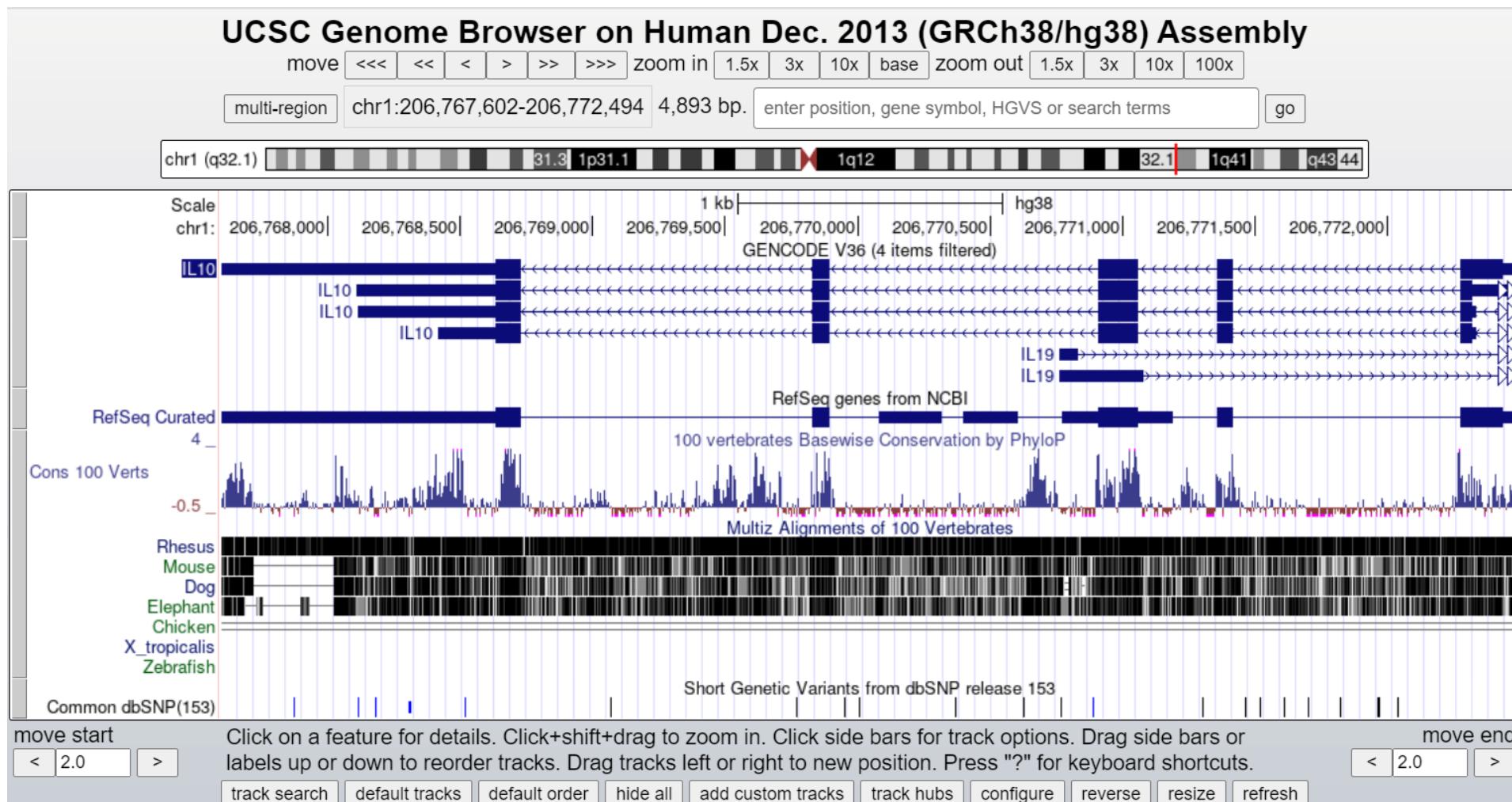
Position/Search Term
IL10

Current position: chr9:133,236,213-133,296

Known Genes

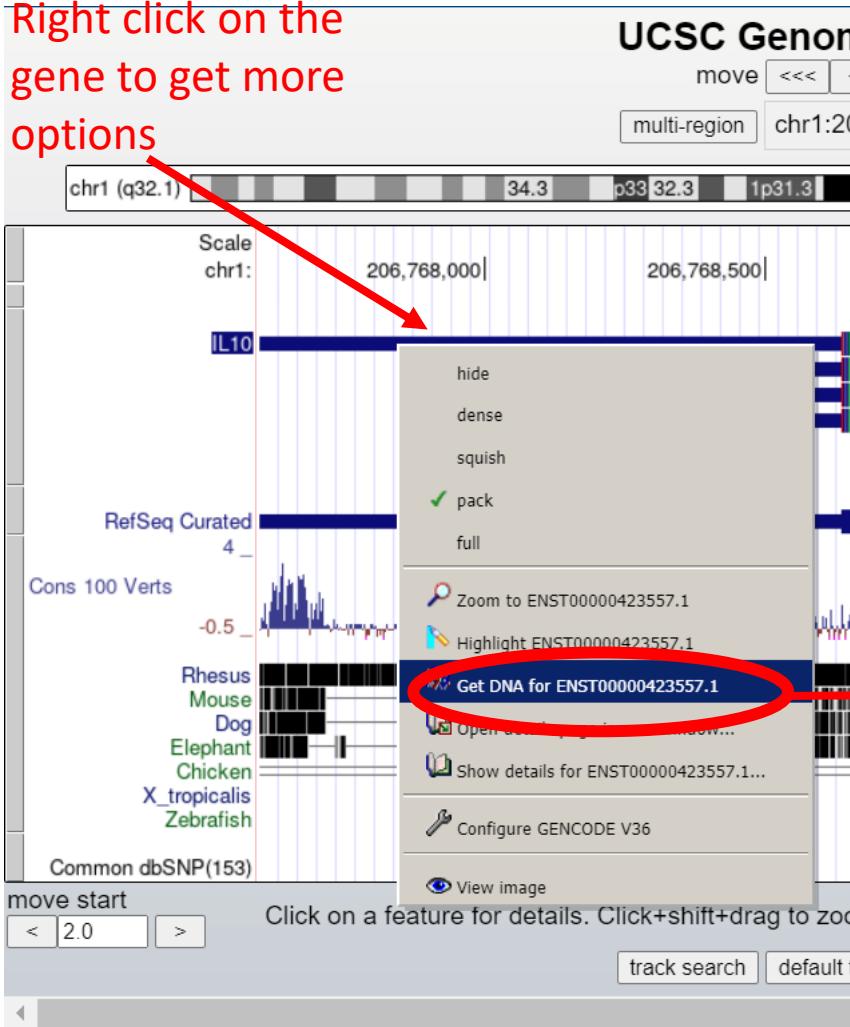
IL10 (uc001hen.2) at chr1:206767602-206772494 - Homo sapiens interleukin 10 (IL10), mRNA. (from RefSeq NM_000572)
IL10 (uc057oxt.1) at chr1:206769621-206771189 - interleukin 10 (from HGNC IL10)
IL10 (uc057oxs.1) at chr1:206768695-206771199 - interleukin 10 (from HGNC IL10)
TIGIT (uc062mgn.1) at chr3:114291102-114310288 - Binds with high affinity to the poliovirus receptor (PVR) which c
IL20 (uc031vvlv.2) at chr1:206865354-206869223 - Proinflammatory and angiogenic cytokine that may be involved in ep
IL20 (uc009xby.4) at chr1:206865809-206869217 - Proinflammatory and angiogenic cytokine that may be involved in ep
IL24 (uc001hew.3) at chr1:206897444-206904139 - Homo sapiens interleukin 24 (IL24), transcript variant 4, mRNA. (fr
IL24 (uc001hex.2) at chr1:206897443-206904139 - Homo sapiens interleukin 24 (IL24), transcript variant 5, mRNA. (fr
IL24 (uc001heu.3) at chr1:206897443-206903480 - Homo sapiens interleukin 24 (IL24), transcript variant 3, mRNA. (fr
IL24 (uc001hes.3) at chr1:206897444-206903865 - Homo sapiens interleukin 24 (IL24), transcript variant 1, mRNA. (fr
IL20 (uc001her.4) at chr1:206865809-206868914 - Homo sapiens interleukin 20 (IL20), mRNA. (from RefSeq NM_018724)
IL19 (uc001heo.3) at chr1:206798870-206842981 - Homo sapiens interleukin 19 (IL19), transcript variant 1, mRNA. (fr
IL19 (uc001hep.4) at chr1:206828877-206842622 - Homo sapiens interleukin 19 (IL19), transcript variant 2, mRNA. (fr
IL26 (uc001stx.2) at chr12:68201351-68225821 - Homo sapiens interleukin 26 (IL26), mRNA. (from RefSeq NM_018402)
TSC22D3 (uc065apd.1) at chrX:107714370-107777067 - Protects T-cells from IL2 deprivation-induced apoptosis through
TSC22D3 (uc004enj.4) at chrX:107713221-107775985 - Protects T-cells from IL2 deprivation-induced apoptosis through
TSC22D3 (uc004eni.4) at chrX:107713224-107775988 - Protects T-cells from IL2 deprivation-induced apoptosis through
SOCS3 (uc002jvl.4) at chr17:78356778-78360077 - Homo sapiens suppressor of cytokine signaling 3 (SOCS3), mRNA. (fr
CD101 (uc057jsu.1) at chr1:117001750-117034250 - Plays a role as inhibitor of T-cells proliferation induced by CD3
CD101 (uc010oxc.3) at chr1:117001812-117036417 - Homo sapiens CD101 molecule (CD101), transcript variant 1, mRNA. (fr
LAIR1 (uc032idt.2) at chr19:54351384-54364957 - Functions as an inhibitory receptor that plays a constitutive nega
LAIR1 (uc032idu.2) at chr19:543514675-54361866 - Functions as an inhibitory receptor that plays a constitutive nega

Viewing IL10 on the browser



Retrieving sequence

Right click on the gene to get more options



Fasta format

>hg38_dna range=chr1:206767603-206772494 5'pad=0 3'pad=0 strand
GATGTGAATAAGATACATTATTATTCAAAATAAAATCTGCTATGAAG
ACAGACAAACAATGTAACATTCCCAGAGGAATTGAATAAATTCTAGTTAA
AACTGAGTTCTATTAGAACCAATTATTATTTTATTTTTATTTTTGAGAC
AGAGTCTTGCTCTGTCAACCAGGCTGGAGTACAGGGGCATGATATCAGCT
CACTGCAACTCCATCTCTGGGTTCAAGCAATTCTCTTGCTCAGCCTC
CCAAGTAGCTGGGATTACAGGTGCGGCCACCATGCCCGCTAATTNTTG
TATTTTTAGTAGAGACGGGTTTACCATGTTGACCAGGCTGGTAGGAA
CTCTGACCTCAAGTGTACCCACCCGCCCTCAGCCTCCCAAAGTGTGGGAT
TACAGGCGTGAGCCACCGCGCCCGGCTAGAACCAAATTAGGTTGTTAT
AAATAACAAGCTGGCCACAGCTTCAGAACATGAAGTGGTTGGGAATGAG
GTTAGGGGAATCCCTCGAGACACTGGAAGGTGAATTAAATCATCAAAGGG
GCTCCCTGGTTCTCTTCTTAAGAGTATTGTAGCAGTTAGGAAGCCCCA
AGCCCAGAGACAAGATAAAATTAGAGGGAGGTCAAGGGAAAACAGCTCAACA
GCTAGAAAAGCTGGTCAAGGCTGGGAATGAAAGCTTCTGTTGGCTCCCCA
AGAACATTTTTTCTCCCTTATGTAACTTATAATTATCTAAACAC
TCAAATACCATAGTGTGTCACCCATGGAAACAGCTAAAAACAGGTGAA
ATAATAAAATTGAAAAAAATTATAATATTGGGCTTCTTCTAAATCGT
TCACAGAGAAGCTCAGTAAATAATAGAAATGGGGTTGAGGTATCAGAG

Get DNA in Window (hg38/Human)

Get DNA for

Position chr1:206,767,603-206,772,494

Note: This page retrieves genomic DNA for a single region. If you a particular track, or get DNA with formatting options based on ge using the Table Browser with the "sequence" output format.

Sequence Retrieval Region Options:

Add 0 extra bases upstream (5') and 0 extra downstream (3')

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:

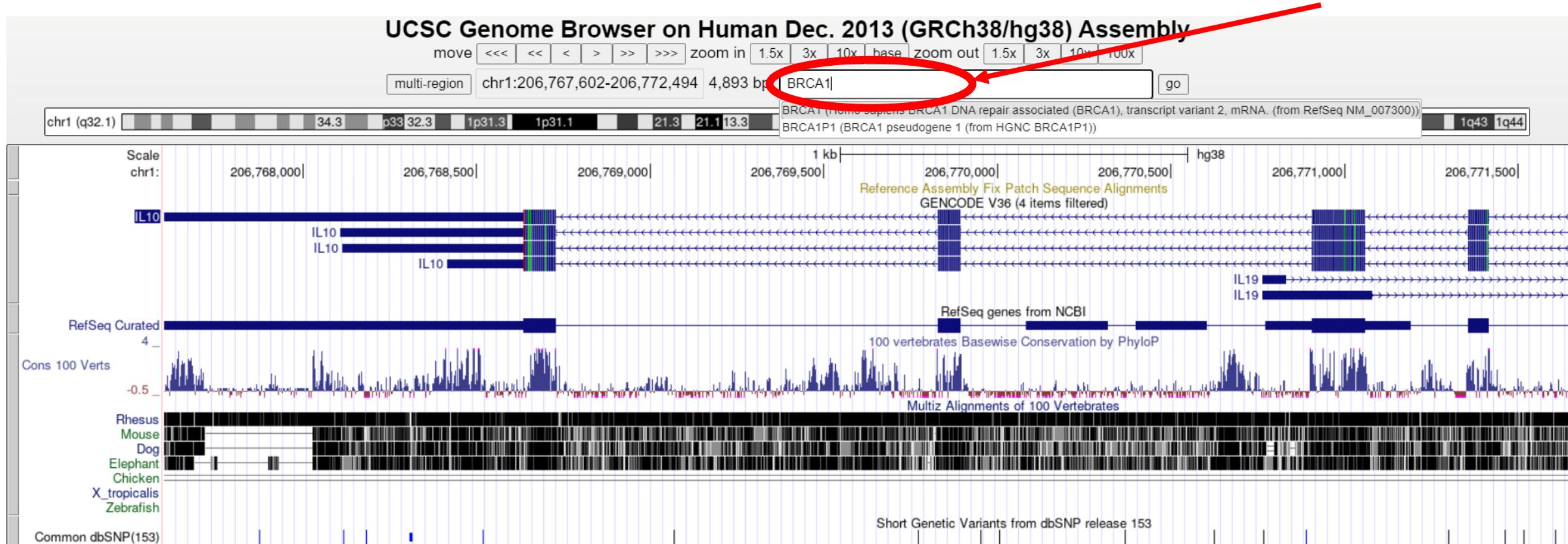
All upper case.
 All lower case.
 Mask repeats: to lower case to N
 Reverse complement (get '-' strand sequence)

get DNA extended case/color options

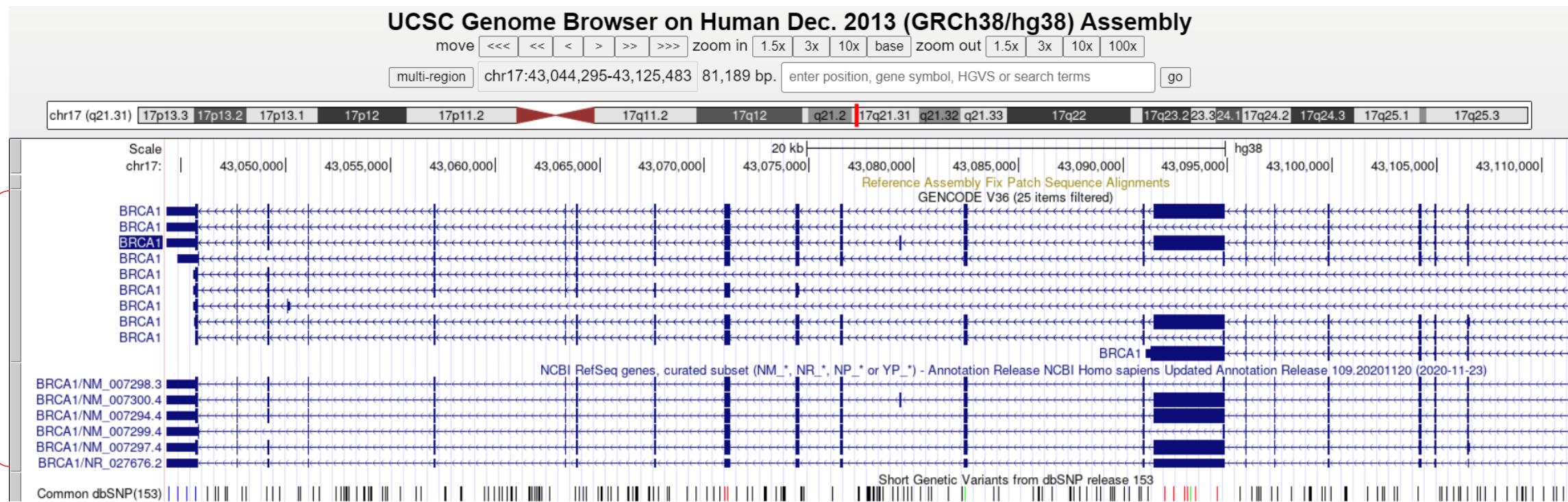
Note: The "Mask repeats" option applies only to "get DNA", not to "extended case/color options".

Another example: BRCA1

You can also query
a gene from here



Another example: BRCA1



Known variants and association to disease



Online Mendelian Inheritance in Man®
An Online Catalog of Human Genes and
Genetic Disorders
Updated May 28, 2021

<http://www.omim.org/>

The screenshot shows a complex web interface for OMIM. At the top, there are several dropdown menus for hiding or showing different gene and variant databases, such as Geneid Genes, P12 Genscan Genes, Non-coding RNA..., Old UCSC Genes, SGP Genes, SIB Genes, 19 IKMC Genes Mapped, LRG Transcripts, MANE select v0.92, P12 MGC Genes, P12 Pfam in UCSC Gene, RetroGenes V9, UniProt, and others. Below these are sections for "Phenotype and Literature" (OMIM Alleles, New CADD..., Coriell CNVs, COSMIC Regions, Updated HGMD Variants, LOVD Variants, OMIM Cyto Loci, ClinGen, Development Delay, Gene Interactions, OMIM Genes, ClinVar Variants, Deprecated ClinGen CNVs, GeneReviews, GWAS Catalog, SNPedia, TCGA Pan-Cancer), "COVID-19" (COVID GWAS v4, COVID GWAS v3, Rare Harmful Vars), "mRNA and EST" (P12 Human ESTs, P12 Human mRNAs, P12 Other ESTs, P12 Other mRNAs, SIB Alt-Splicing, P12 Spliced ESTs), and "Expression". A red box highlights the OMIM logo and the URL at the bottom left of the interface.

OMIM® is a continuously updated catalog of human genes and genetic disorders and traits, with particular focus on the molecular relationship between genetic variation and phenotypic expression.

Known variants and association to disease

Refresh to get the tracks at the relevant observation mode in browser



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Updated May 28, 2021

<http://www.omim.org/>

The screenshot shows the OMIM browser interface with several tracks listed on the right side:

- Geneid Genes
- P12 Genscan Genes
- Non-coding RNA...
- Old UCSC Genes
- SGP Genes
- SIB Genes
- 19 IKMC Genes Mapped
- LRG Transcripts
- MANE select v0.92
- P12 MGC Genes
- P12 ORFeome Clones
- P12 Other RefSeq
- TransMap V5...
- P12 UCSC Alt Events
- UniProt
- ClinGen
- Gene Interactions
- OMIM Genes
- SNPedia
- Cancer Gene Expr...
- Development Delay.
- LOVD Variants
- OMIM Cyto Loci
- Variants in Papers...
- ClinVar Variants
- GeneReviews
- GWAS Catalog
- TCGA Pan-Cancer
- Deprecated ClinGen
- CNVs
- COSMIC Regions
- IMD
- UnProt Variants
- COVID-19
- COVID GWAS v4
- COVID GWAS v3
- Rare Harmful Vars
- mRNA and EST
- P12 Human ESTs
- P12 Human mRNAs
- P12 Other ESTs
- P12 Other mRNAs
- SIB Alt-Splicing
- P12 Spliced ESTs

A red arrow points from the text "Refresh to get the tracks at the relevant observation mode in browser" to the "refresh" button located at the bottom right of the interface.

Known variants and association to disease



Known variants and association to disease

Refresh to get the tracks at the relevant observation mode in browser



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Genetic Disorders
Updated May 28, 2021

<http://www.omim.org/>

The screenshot shows a genomic browser interface with a header bar containing various tracks and a central panel labeled "Phenotype and Literature".

Header Tracks (left to right):

- Non-coding RNA... (hide dropdown)
- Old UCSC Genes (hide dropdown)
- P12 ORFeome Clones (hide dropdown)
- P12 Other RefSeq (hide dropdown)
- P12 Pfam in UCSC Gene (hide dropdown)
- RetroGenes V9 (hide dropdown)

Header Tracks (left to right):

- SGP Genes (hide dropdown)
- SIB Genes (hide dropdown)
- TransMap V5... (hide dropdown)
- P12 UCSC Alt Events (hide dropdown)
- UniProt (hide dropdown)

Central Panel:

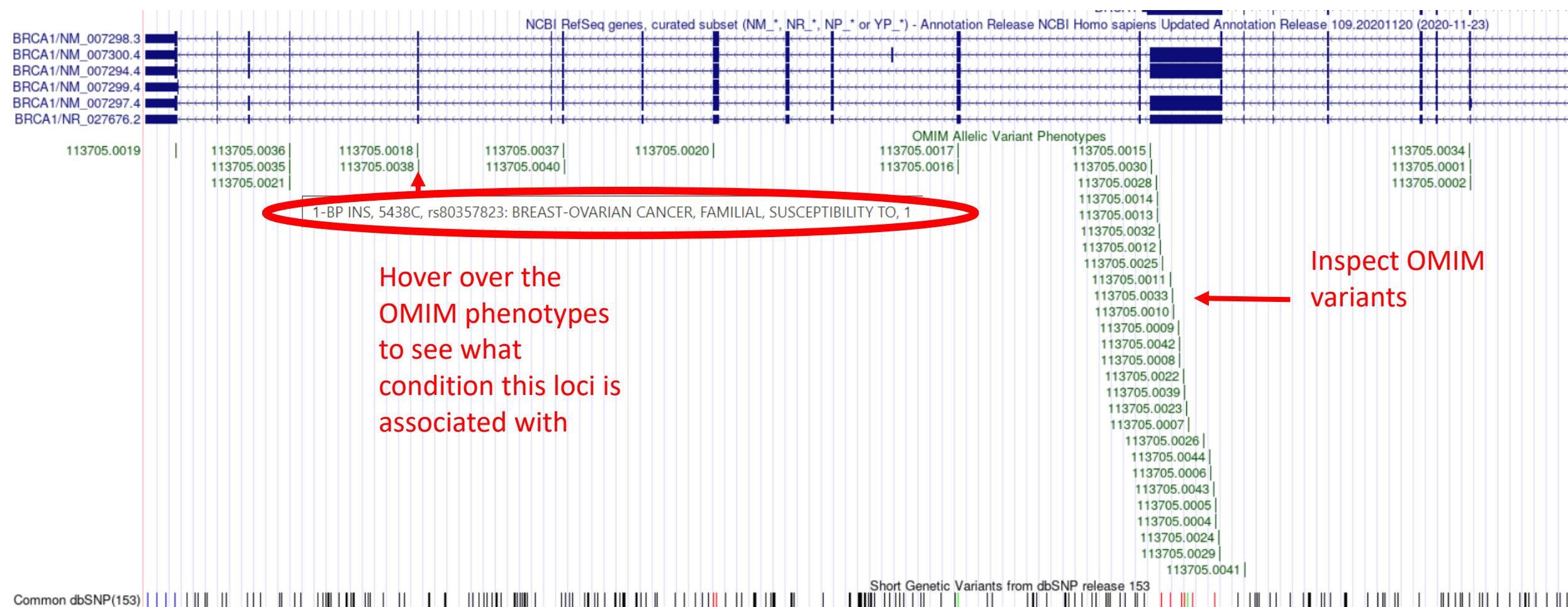
Phenotype and Literature

Category	Track	Action
OMIM Alleles	New CADD...	hide dropdown
	Cancer Gene Expr...	hide dropdown
CNVs	OMIM Cyto Loci	hide dropdown
	Development Delay	hide dropdown
COSMIC Regions	Gene Interactions	hide dropdown
	GeneReviews	hide dropdown
LOVD Variants	SNPedia	hide dropdown
	TCGA Pan-Cancer	hide dropdown
OMIM Genes	ClinGen	hide dropdown
	ClinVar Variants	hide dropdown
UniProt Variants	Variants in Papers...	hide dropdown
	GWAS Catalog	hide dropdown

Bottom Panel:

COVID-19

Known variants and association to disease



Click on a variant to find more information

OMIM Allelic Variant Phenotypes (113705.0016)

OMIM Allelic Variant: [113705.0016](#) BREAST-OVARIAN CANCER, FAMILIAL, SUSCEPTIBILITY TO, 1

OMIM: [113705](#): Breast cancer-1 gene

Amino Acid Replacement: ARG1443TER

dbSNP/ClinVar: [rs41293455](#)

Position: [chr17:43082434-43082434](#)

Band: 17q21.31

Genomic Size: 1

[View table schema](#)

[Go to OMIM Alleles track controls](#)

Data last updated at UCSC: 2021-05-26

Description

NOTE:

OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions. Further, please be sure to click through to [omim.org](#) for the very latest, as they are continually updating data.

NOTE ABOUT DOWNLOADS:

OMIM is the property of Johns Hopkins University and is not available for download or mirroring by any third party without their permission. Please see [OMIM](#) for downloads.



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***113705**

Table of Contents

Title

Gene-Phenotype Relationships

Text

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Cloning and Expression

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Gene Function

Molecular Genetics

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Evolution

Animal Model

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Table View

See Also

References

Contributors

Creation Date

Edit History

*** 113705**

BREAST CANCER 1 GENE; BRCA1

*HGNC Approved Gene Symbol: BRCA1**Cytogenetic location: 17q21.31 Genomic coordinates (GRCh38): 17:43,044,294-43,125,363 (from NCBI)*

Gene-Phenotype Relationships

Location	Phenotype	Clinical Synopses	Phenotype MIM number	Inheritance	Phenotype mapping key
17q21.31	[Breast-ovarian cancer, familial, 1]		604370	AD, Mu	3
	[Pancreatic cancer, susceptibility to, 4]		614320		3
	Fanconi anemia, complementation group S		617883	AR	3

PheneGene Graphics ▾

TEXT

▼ Description

BRCA1 plays critical roles in DNA repair, cell cycle checkpoint control, and maintenance of genomic stability. BRCA1 forms several distinct complexes through association with different adaptor proteins, and each complex forms in a mutually exclusive manner (Wang et al., 2009).

▼ External Links

► Genome

► DNA

► Protein

► Gene Info

► Clinical Resources

▼ Variation

1000 Genome

ClinVar

gnomAD

GWAS Catalog

GWAS Central

HGMD

HGVS

► Locus Specific DBs

NHLBI EVS

PharmgKB

► Animal Models

► Cell Lines

► Cellular Pathways

Click on a variant to find more information

OMIM Allelic Variant Phenotypes (113705.0016)

OMIM Allelic Variant: [113705.0016](#) BREAST-OVARIAN CANCER, FAMILIAL, SUSCEPTIBILITY TO, 1

OMIM: [113705](#): Breast cancer 1 gene

Amino Acid Replacement: ARG1443TER

dbSNP/ClinVar: [rs41293455](#)

Position: [chr17:43082434-43082434](#)

Band: 17q21.31

Genomic Size: 1

[View table schema](#)

[Go to OMIM Alleles track controls](#)

Data last updated at UCSC: 2021-05-26

Description

NOTE:

OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions.

Further, please be sure to click through to [omim.org](#) for the very latest, as they are continually updating data.

NOTE ABOUT DOWNLOADS:

OMIM is the property of Johns Hopkins University and is not available for download or mirroring by any third party without their permission. Please see [OMIM](#) for downloads.



Search OMIM...



Options ▾

***113705**[Table of Contents](#)[Title](#)[Gene-Phenotype Relationships](#)[Text](#)[Description](#)[Cloning and Expression](#)[Gene Structure](#)[Mapping](#)[Gene Function](#)[Molecular Genetics](#)[Genotype/Phenotype Correlations](#)[Evolution](#)[Animal Model](#)[Allelic Variants](#)[Table View](#)[See Also](#)[References](#)[Contributors](#)[Creation Date](#)[Edit History](#)

Friedman et al. (1994) studied 63 breast cancer patients and 10 ovarian cancer patients in 10 families with cancer linked to chromosome 17q21 (604370). Simard et al. (1994) studied 30 Canadian families with breast and/or ovarian cancer for germline mutations in the coding region of the BRCA1 candidate gene. In both studies, a 4-bp (TCAA) deletion in exon 11 at position 4184, leading to a premature termination codon at position 1364 and a truncated protein, was identified. [+](#)

.0016 BREAST-OVARIAN CANCER, FAMILIAL, SUSCEPTIBILITY TO, 1BRCA1, ARG1443TER [rs41293455](#) [gnomAD:rs41293455](#) [RCV000515235...](#)

Castilla et al. (1994) studied 50 probands with a family history of breast and/or ovarian cancer (604370) for germline mutations in the coding region of the BRCA1 candidate gene. They identified a C-to-T substitution at position 4446 of the BRCA1 gene, leading to a premature termination codon in place of arginine-1443 and a truncated protein. [+](#)

.0017 BREAST-OVARIAN CANCER, FAMILIAL, SUSCEPTIBILITY TO, 1BRCA1, ARG1443GLY [rs41293455](#) [gnomAD:rs41293455](#) [RCV000427206...](#)

Castilla et al. (1994) studied 50 probands with a family history of breast and/or ovarian cancer (604370) for germline mutations in the coding region of the BRCA1 candidate gene. They identified a C-to-G transition at position 4446, changing arginine-1443 to glycine. [+](#)

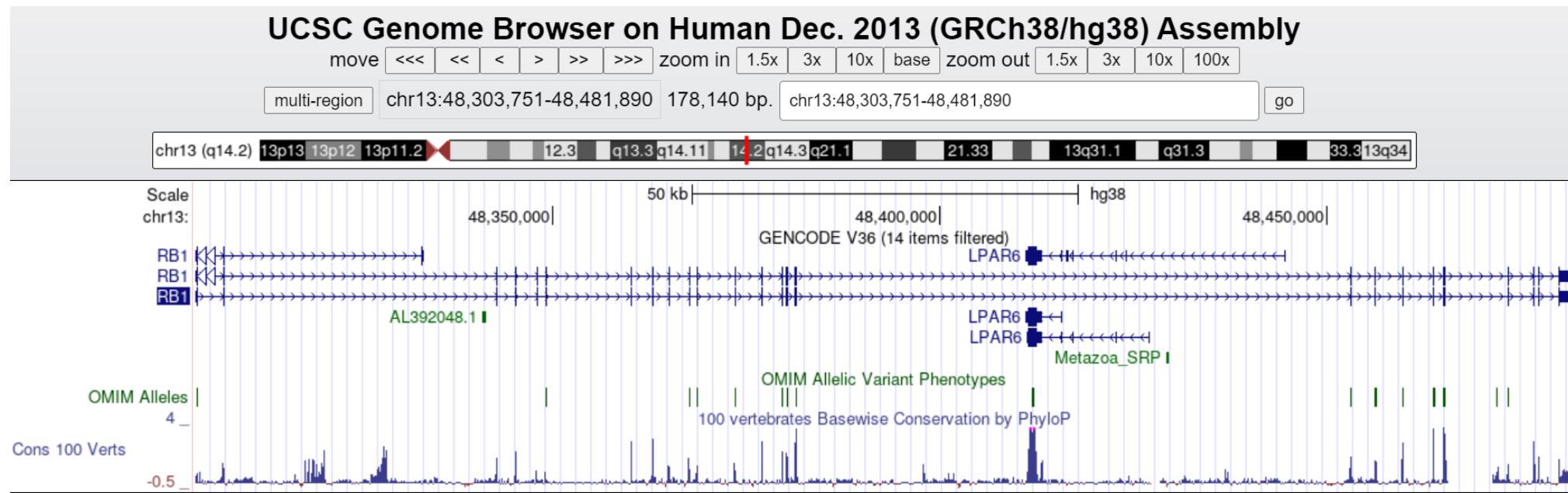
.0018 BREAST-OVARIAN CANCER, FAMILIAL, SUSCEPTIBILITY TO, 1

PANCREATIC CANCER, SUSCEPTIBILITY TO, INCLUDED

External Links[▶ Genome](#)[▶ DNA](#)[▶ Protein](#)[▶ Gene Info](#)[▶ Clinical Resources](#)**▼ Variation**[1000 Genome](#)[ClinVar](#)[gnomAD](#)[GWAS Catalog](#)[GWAS Central](#)[HGMD](#)[HGVS](#)**▶ Locus Specific DBs**[NHLBI EVS](#)[PharmgKB](#)[▶ Animal Models](#)[▶ Cell Lines](#)[▶ Cellular Pathways](#)

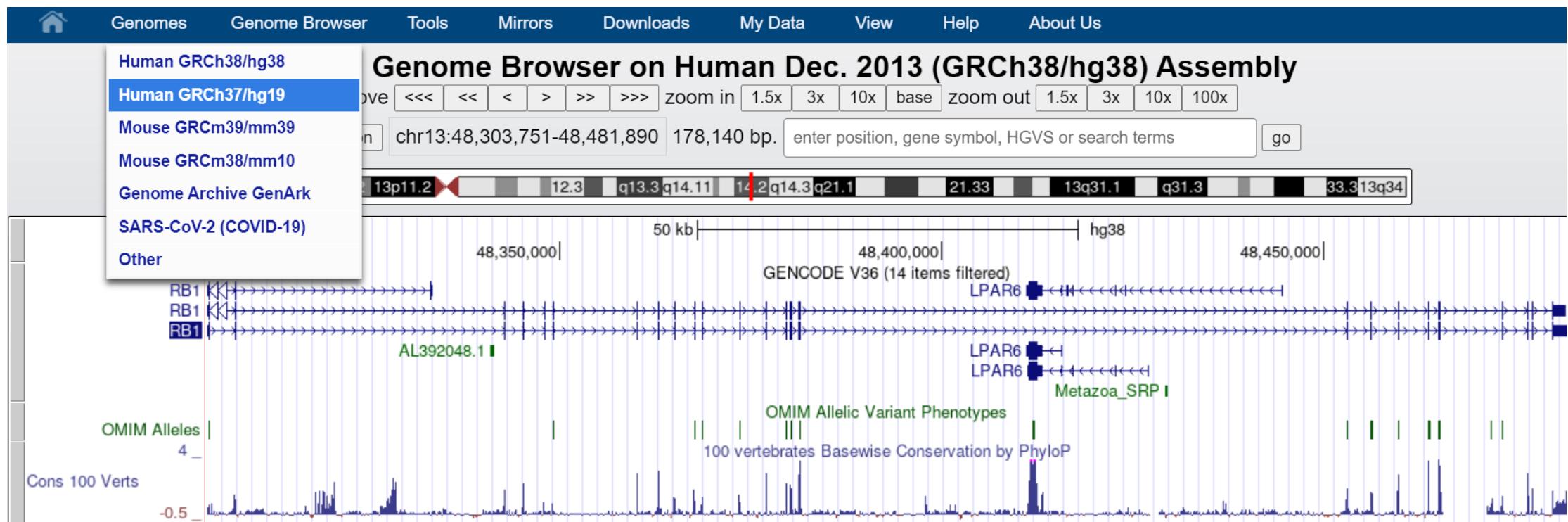
Why it's important to use the correct genome assembly ?

chr13:48,303,751-48,481,890



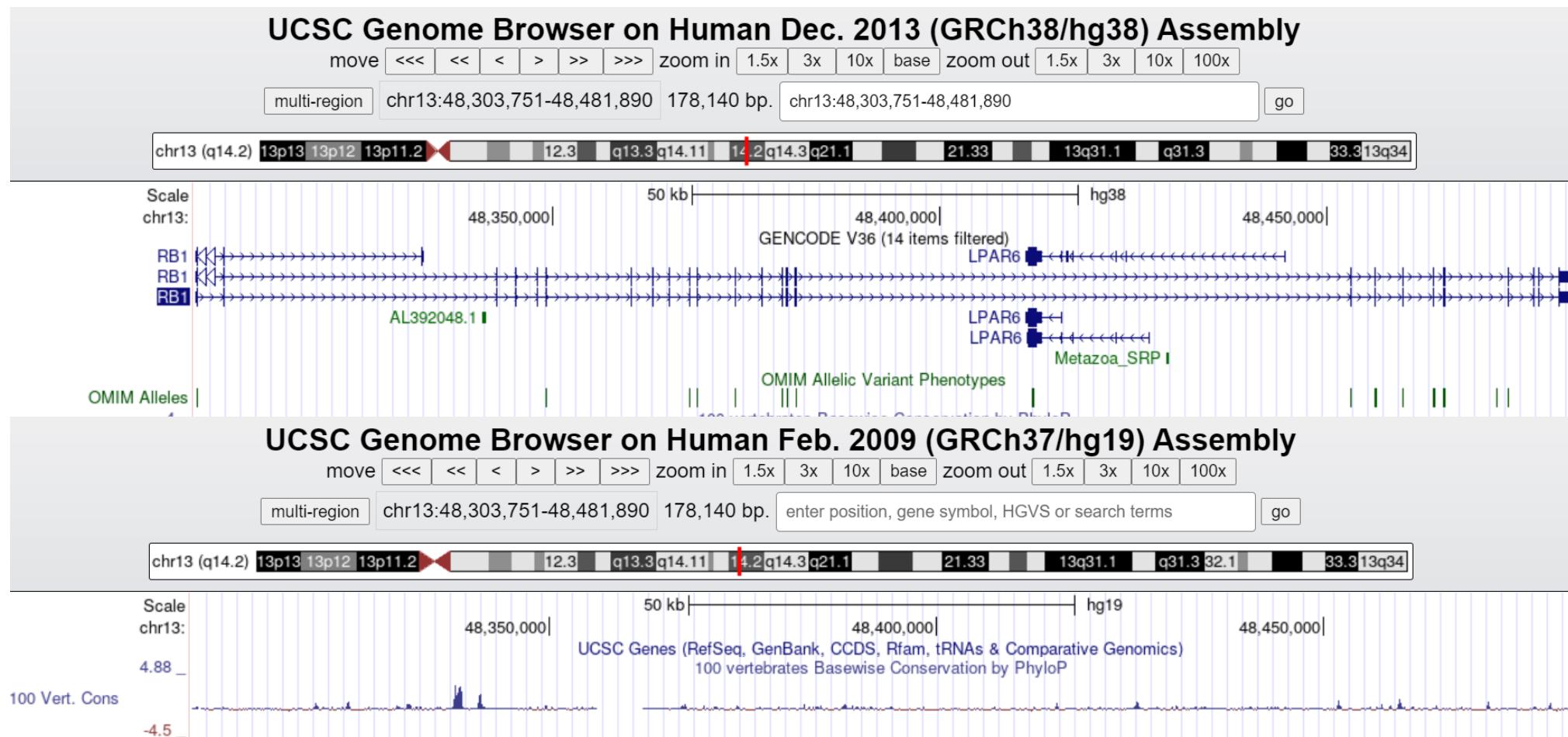
Why it's important to use the correct genome assembly ?

chr13:48,303,751-48,481,890



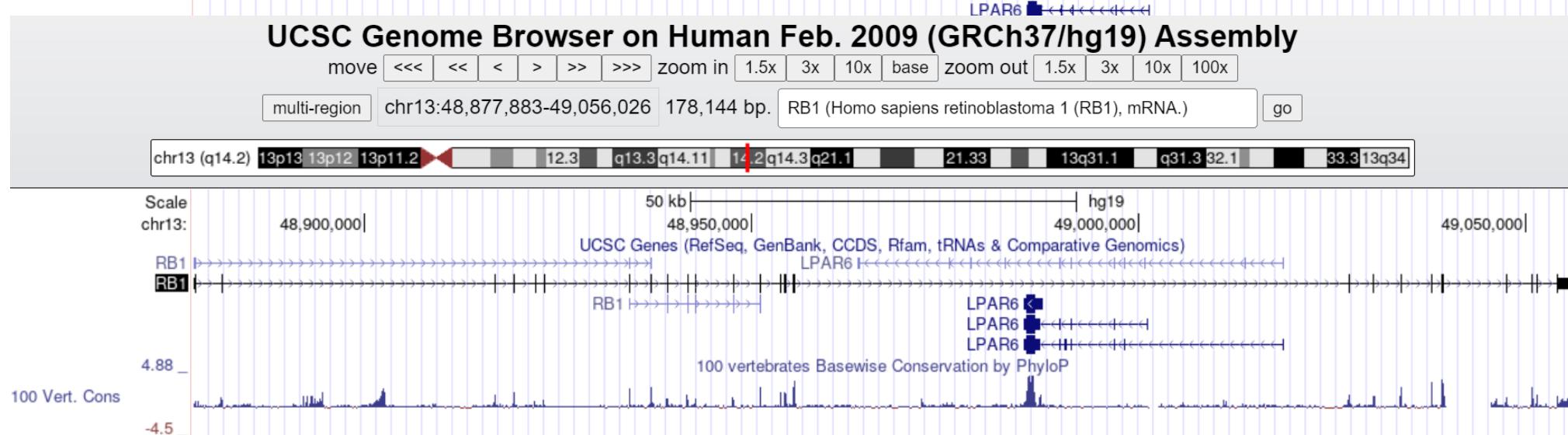
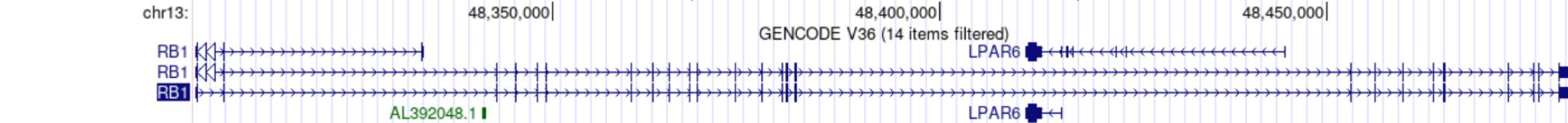
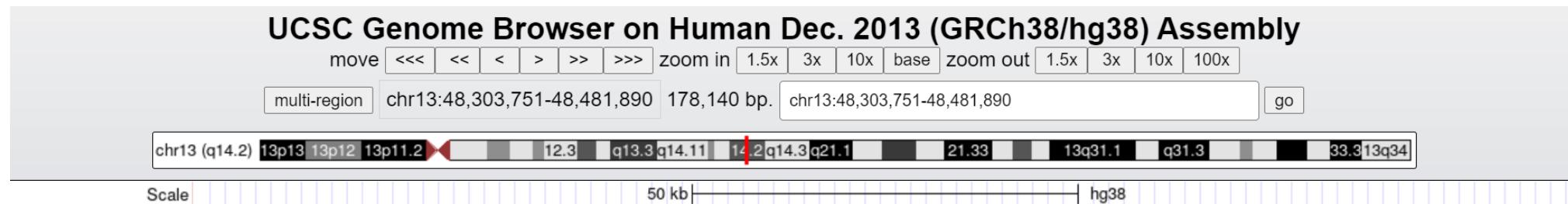
Why it's important to use the correct genome assembly ?

chr13:48,303,751-48,481,890



Why it's important to use the correct genome assembly?

chr13:48,303,751-48,481,890



Exons in different transcripts

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

move <<< << < > >> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

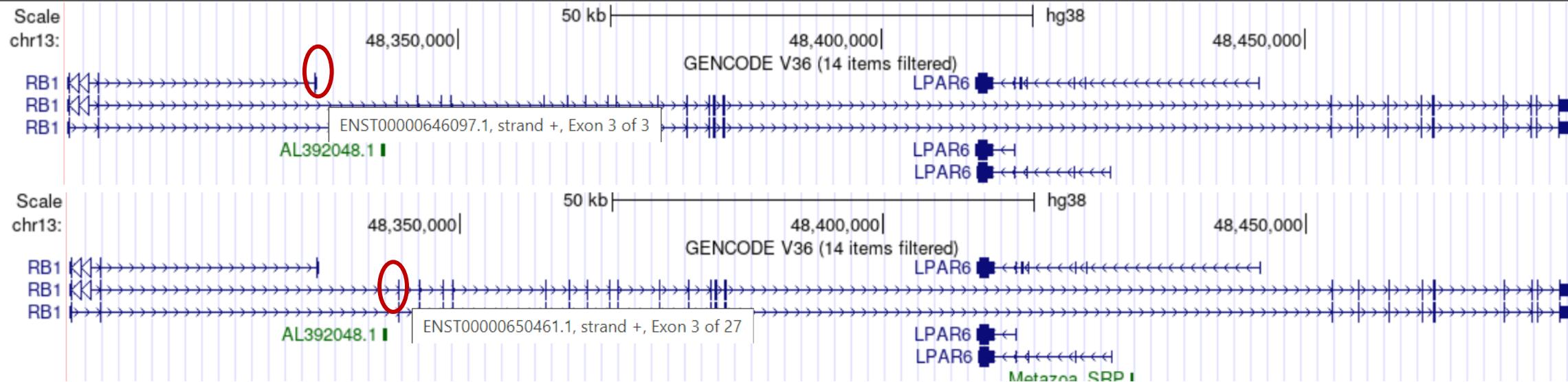
multi-region

chr13:48,303,751-48,481,890 178,140 bp.

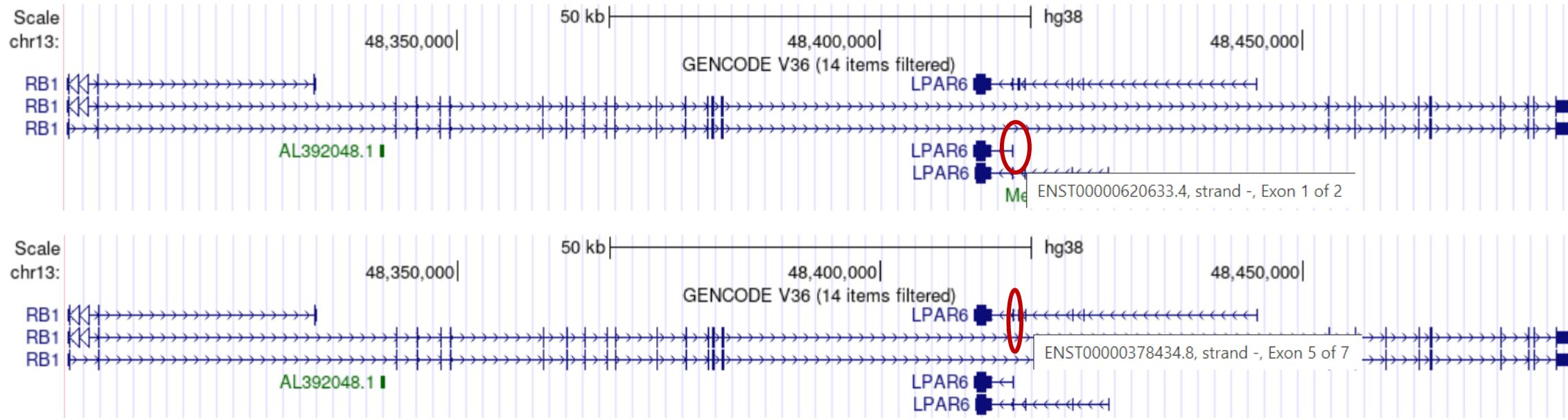
enter position, gene symbol, HGVS or search terms

go

chr13 (q14.2) 13p13 13p12 13p11.2 12.3 q13.3 q14.11 14.2 q14.3 q21.1 21.33 13q31.1 q31.3 33.3 13q34



Exons in different transcripts



miRNA track

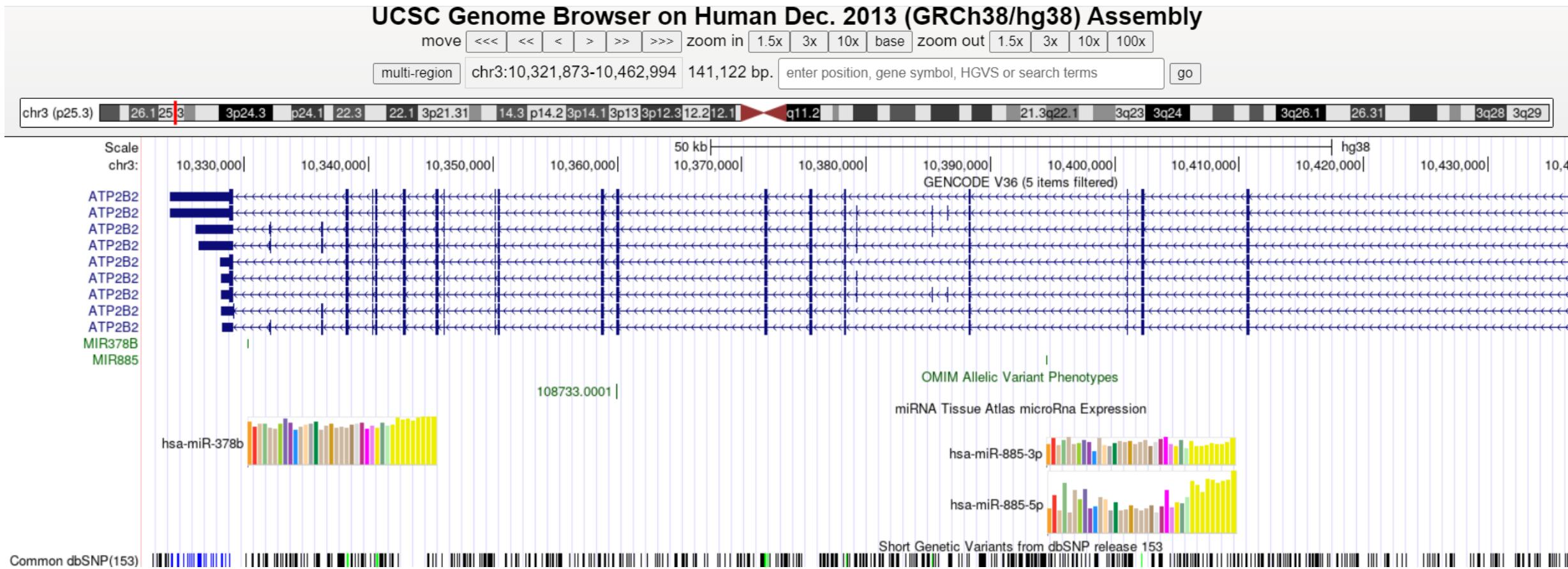
MicroRNAs (miRNAs) are a class of non-coding RNAs that play important roles in regulating gene expression.

In most cases, miRNAs interact with the 3' untranslated region (3' UTR) of target mRNAs to induce mRNA degradation and translational repression.

The screenshot shows the 'miRNA track' in the UCSC Genome Browser. The interface is organized into several horizontal panels, each containing a list of genomic tracks with dropdown menus for hiding or showing them.

- Phenotype and Literature:** Includes tracks for OMIM Alleles, New CADD..., Cancer Gene Expr..., ClinGen, Deprecated ClinGen CNVs, ClinVar Variants, Coriell CNVs, COSMIC Regions, Development Delay, Gene Interactions, GeneReviews, HGMD Variants, LOVD Variants, OMIM Cyto Loci, OMIM Genes, SNPedia, UniProt Variants, Variants in Papers..., and TCGA Pan-Cancer.
- COVID-19:** Includes COVID GWAS v4, COVID GWAS v3, and Rare Harmful Vars.
- mRNA and EST:** Includes P12 Human ESTs, P12 Human mRNAs, P12 Other ESTs, P12 Other mRNAs, SIB Alt-Splicing, and P12 Spliced ESTs.
- Expression:** Includes GTEx Gene V8, Affy GNF1H, Affy U133, Affy U95, EPDnew Promoters, GNF Atlas 2, P12 GTeX Gene, GTeX Transcript, GWIPS-viz Riboseq, and miRNA Tissue Atlas.
- Regulation:** Includes ENCODE cCREs, P12 ENCODE Regulation..., P12 CpG Islands..., Hi-C and Micro-C, and ORegAnno. A dropdown menu for regulation visualization is open, showing options: full (selected), hide, dense, squish, pack, and full.
- Comparative Genomics:** Includes RefSeq Func Elems.

miRNA track



Extracting genomic data using the table browser

The screenshot shows the UCSC Genome Browser Gateway homepage. At the top, there is a logo for the University of California Santa Cruz Genomics Institute and the UCSC logo. Below the header, a navigation bar includes links for Home, Genomes, Genome Browser, Tools, Mirrors, Downloads, My Data, Projects, Help, and About. A dropdown menu is open over the 'Tools' link, listing various options: Blat, In-Silico PCR, Table Browser, LiftOver, Gene Sorter, Variant Annotation Integrator, Data Integrator, Genome Graphs, Gene Interactions, and Other Tools. The 'Table Browser' option is highlighted with a red circle. To the right of the dropdown, there is a search bar labeled 'Position' with a dropdown menu set to 'Assembly: hg38 (GRCh38/hg38)'. Below the search bar is a 'Search Term' input field containing the value 'position: chr3:10,328,102-10,449,543'. A blue 'GO' button is next to the search term. On the left side of the page, there is a section titled 'Browse/Select Species' with a 'POPULAR SPECIES' list featuring icons for Human, Mouse, Rat, Zebrafish, and Fruitfly. Below this is a text input field for 'Enter species, common name or assembly ID'. Further down, there is a link 'Can't find a genome assembly?'. On the far left, there is a vertical bar with a human icon and a phylogenetic tree diagram showing the relationships between Human, Chimp, Bonobo, Gorilla, Orangutan, and Gibbon.

UNIVERSITY OF CALIFORNIA
SANTA CRUZ Genomics Institute

UCSC Genome Browser Gateway

Home Genomes Genome Browser Tools Mirrors Downloads My Data Projects Help About

Browse>Select Species

POPULAR SPECIES

Human Mouse Rat Zebrafish Fruitfly

Enter species, common name or assembly ID

Can't find a genome assembly?

REPRESENTED SPECIES

Human Chimp Bonobo Gorilla Orangutan Gibbon

Position

Assembly: hg38 (GRCh38/hg38)

Search Term

position, gene symbol or search terms

position: chr3:10,328,102-10,449,543

GO

UCSC Genome Browser - hg38 assembly

Genome Browser assembly ID: hg38

Sequencing/Assembly provider ID: Genome Reference Consortium

Assembly date: Dec. 2013 initial release; Dec. 2017 patch release

Assembly accession: GCA_000001405.27

NCBI Genome ID: 51 (Homo sapiens (human))

NCBI Assembly ID: 5800238 (GRCh38.p12, GCA_000001405.27)

BioProject ID: PRJNA31257

Table Browser

Table Browser

Use this program to retrieve the data associated with a track in text format, to calculate intersections between tracks, and to examine the biological function of your set through annotation enrichments. Send the data to [GREAT](#). Refer to the [User's Guide](#) for general information and sample usage.

clade: Mammal **group:** Mammal **table:** Deuterostome **region:** Insect **identifier:** Other **filter:** Viruses

genome: Human **assembly:** Dec. 2013 (GRCh38/hg38) **Predictions** **track:** GENCODE V36 **describe table schema** **position:** chr3:10,328,102-10,449,543 **lookup** **define regions**

sessions:

intersection:

output format: all fields from selected table **Send output to:** Galaxy GREAT

output file: (leave blank to keep output in browser)

file type returned: plain text gzip compressed

To reset **all** user cart settings (including custom tracks), [click here](#).

Table Browser

Table Browser

Use this program to retrieve the data associated with a track in text format, to calculate intersections between tracks, and [Table Browser](#) for a description of the controls in this form, and the [User's Guide](#) for general information and sample queries. To examine the biological function of your set through annotation enrichments, send the data to [GREAT](#). Refer to the [Credits](#) page to see who helped develop this tool. You can also download the data in their entirety from the [Sequence and Annotation Downloads](#) page.

clade: Mammal genome: Human assembly: Dec. 2013 (GRCh38/hg38)

group: Genes and Gene Predictions track: GENCODE V36

table: Genes and Gene Predictions assembly: Dec. 2013 (GRCh38/hg38)

region: 3:10,328,102-10,449,543 lookup define regions

identifier: paste list upload list

filter: Expression regulation

intersection: Comparative Genomics

output: Variation repeats

output: All Tracks All Tables

file type: table Send output to Galaxy GREAT
 (leave blank to keep output in browser)
gzip compressed

get output summary/statistics

To reset **all** user cart settings (including custom tracks), [click here](#).

Table Browser

Table Browser

Use this program to retrieve the data associated with a track in text format, to calculate intersections between tracks, and to examine the biological function of your set through annotation enrichments. Send the data to [GREAT](#). Refer to the [Table Browser](#) for a description of the controls in this form, and the [User's Guide](#) for general information and instructions.

be downloaded in their entirety from the [Sequence and Annotation Downloads](#) page.

clade: Mammal **genome:** Human **assembly:** Dec. 2013 (GRCh38/hg38)

group: Genes and Gene Predictions **track:** GENCODE V36

table: knownGene **describe table schema**

region: genome position chr3:10,328,100

identifiers (names/accessions): [paste list](#)

filter: [create](#)

intersection: [create](#)

output format: all fields from selected table

output file:

file type returned: plain text gzip compressed

[get output](#) [summary/statistics](#)

To reset **all** user cart settings (including custom tracks)

Using the Table Browser

This section provides brief line-by-line descriptions of the Table Browser controls. For more information on using the Table Browser, refer to the [Table Browser](#).

The **track:** dropdown menu is currently set to "GENCODE V36". Other options available in the dropdown include:

- GENCODE V36 (selected)
- NCBI RefSeq
- TransMap Ensembl
- TransMap RefGene
- TransMap RNA
- TransMap ESTs
- All GENCODE V37
- All GENCODE V36
- All GENCODE V35
- All GENCODE V34
- All GENCODE V33
- All GENCODE V32
- All GENCODE V31
- All GENCODE V29
- All GENCODE V28
- All GENCODE V27
- All GENCODE V26
- All GENCODE V25
- All GENCODE V24
- All GENCODE V23

add custom tracks **track hubs**

define regions

[Galaxy](#) [GREAT](#)

[in browser](#)

Table Browser

Table Browser

Use this program to retrieve the data associated with a track in text format, to calculate intersections between tracks, and to retrieve DNA sequence covered by a track. For help in using this application see [Using the Table Browser](#) for a description of the controls in this form, and the [User's Guide](#) for general information and sample queries. For more complex queries, you may want to use [Galaxy](#) or our [public MySQL server](#). To examine the biological function of your set through annotation enrichments, send the data to [GREAT](#). Refer to the [Credits](#) page for the list of contributors and usage restrictions associated with these data. All tables can be downloaded in their entirety from the [Sequence and Annotation Downloads](#) page.

clade: Mammal genome: Human assembly: Dec. 2013 (GRCh38/hg38)

group: Genes and Gene Predictions track: Gencode V36 add custom tracks track hubs

table: knownGene **describe table schema**

region: genome position chr3:10394489-10394562 lookup define regions

identifiers (names/accessions): [paste list](#) [upload list](#)

filter: [create](#)

intersection: [create](#)

output format: all fields from selected table Send output to Galaxy GREAT

output file: (leave blank to keep output in browser)

file type returned: plain text gzip compressed

[get output](#) [summary/statistics](#)

To reset **all** user cart settings (including custom tracks), [click here](#).

Table Browser

Schema for GENCODE V36 - GENCODE V36

Database: hg38 Primary Table: knownGene Data last updated: 2021-01-14

Big Bed File: /gbdb/hg38/gencode/gencodeV36.bb

Item Count: 232,184

Format description: GENCODE bigGenePred

field	example	description
chrom	chr1	Reference sequence chromosome or scaffold
chromStart	166022214	Start position in chromosome
chromEnd	166023027	End position in chromosome
name	ENST00000422100.1	Ensembl ID
score	0	Score (0-1000)
strand	+	+ or - for strand
thickStart	166022214	Start of where display should be thick (start codon)
thickEnd	166022214	End of where display should be thick (stop codon)
reserved	16724991	RGB value (use R,G,B string in input file)
blockCount	1	Number of blocks
blockSizes	813,	Comma separated list of block sizes
chromStarts	0,	Start positions relative to chromStart
name2	uc286ezv.1	UCSC Genes ID
cdsStartStat	none	Status of CDS start annotation (none, unknown, incomplete, or complete)
cdsEndStat	none	Status of CDS end annotation (none, unknown, incomplete, or complete)
exonFrames	-1,	Exon frame {0,1,2}, or -1 if no frame for exon
type	none	Transcript type
geneName	RPS3AP10	Gene Symbol
geneName2	none	UniProt display ID
geneType	none	Gene type
transcriptClass	pseudo	Transcript Class
source	havana_homo_sapiens	Source of transcript (from gencodeTranscriptSource)
transcriptType	processed_pseudogene	BioType of transcript (from gencodeAttrs)
tag	basic,pseudo_consens	symbolic tags (from gencodeTags)
level	1	support level, tsl1 is strongest support, tsl5 weakest, NA means not analyzed (from gencodeTranscriptionSupportLevel)
tier	canonical,basic,all	Transcript Tier

Table Browser

Table Browser

Use this program to retrieve the data associated with a track in text format, to calculate intersections between tracks, and to retrieve DNA sequence covered by a track. For help in using this application see [Using the Table Browser](#) for a description of the controls in this form, and the [User's Guide](#) for general information and sample queries. For more complex queries, you may want to use [Galaxy](#) or our [public MySQL server](#). To examine the biological function of your set through annotation enrichments, send the data to [GREAT](#). Refer to the [Credits](#) page for the list of contributors and usage restrictions associated with these data. All tables can be downloaded in their entirety from the [Sequence and Annotation Downloads](#) page.

clade: Mammal genome: Human assembly: Dec. 2013 (GRCh38/hg38)

group: Genes and Gene Predictions track: Gencode V36 [add custom tracks](#) [track hubs](#)

table: knownGene [describe table schema](#)

region: genome position chr3:10394489-10394562 [lookup](#) [define regions](#)

identifiers (names/accessions): [paste list](#) [upload list](#)

filter: [create](#)

intersection: [create](#)

output format: all fields from selected table [Send output to](#) Galaxy GREAT

output file: (leave blank to keep output in browser)

file type returned: plain text gzip compressed

[get output](#) [summary/statistics](#)

To reset **all** user cart settings (including custom tracks), [click here](#).

Table Browser

```
#chrom chromStart chromEnd name score strand thickStart thickEnd reserved blockCount blockSizes chromStarts name2
cdsStartStat cdsEndStat exonFrames type geneName geneName2 geneType transcriptClass source transcriptType tag level tier
chr1 11868 14409 ENST00000456328.2 0 + 11868 11868 16724991 3 359,109,1189, 0,744,1352, uc286dmu.1 none none
-1,-1,-1, none DDX11L1 none none pseudo havana_homo_sapiens processed_transcript basic 2 canonical,basic,all
chr1 12009 13670 ENST00000450305.2 0 + 12009 12009 16724991 6 48,49,85,78,154,218, 0,169,603,965,1211,1443, uc286dmv.1
none none -1,-1,-1,-1,-1,-1, none DDX11L1 none none pseudo havana_homo_sapiens transcribed_unprocessed_pseudogene basic 2 basic,all
chr1 14403 29570 ENST00000488147.1 0 - 14403 14403 16724991 11 98,34,152,159,198,136,137,147,99,154,37,
0,601,1392,2203,2454,2829,3202,3511,3864,10334,15130, uc286dmw.1 none none -1,-1,-1,-1,-1,-1,-1,-1,-1,-1,-1, none WASH7P none none pseudo
havana_homo_sapiens unprocessed_pseudogene basic 2 canonical,basic,all
chr1 17368 17436 ENST00000619216.1 0 - 17368 17368 25600 1 68, 0, uc031tla.1 none none -1, none MIR6859-1
none none nonCoding ncRNA_homo_sapiens miRNA basic 3 canonical,basic,all
chr1 29553 31097 ENST00000473358.1 0 + 29553 29553 25600 3 486,104,122, 0,1010,1422, uc057aty.1 none none -1,-1,-1,
none MIR1302-2HG none none nonCoding havana_homo_sapiens lncRNA basic,dotter_confirmed,ncRNA_host,not_best_in_genome_evidence 2 canonical,basic,all
```

The results can be saved as a file and analyzed using a script.