

Genomic visualization

**BROWSERS:
UCSC - ENSEMBL - IGV**

UCSC Genome Browser



UCSC: University of California Santa Cruz

Human Genome Sequencing Project (July 2000)

UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute

UCSC Genome Browser

Our tools

- **Genome Browser** interactively visualize genomic data
- **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
- **Gene Sorter** find genes that are similar by expression and other metrics
- **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
- **VisiGene** interactively view *in situ* images of mouse and frog

More tools...

Vertebrate and model species

Browse>Select Species

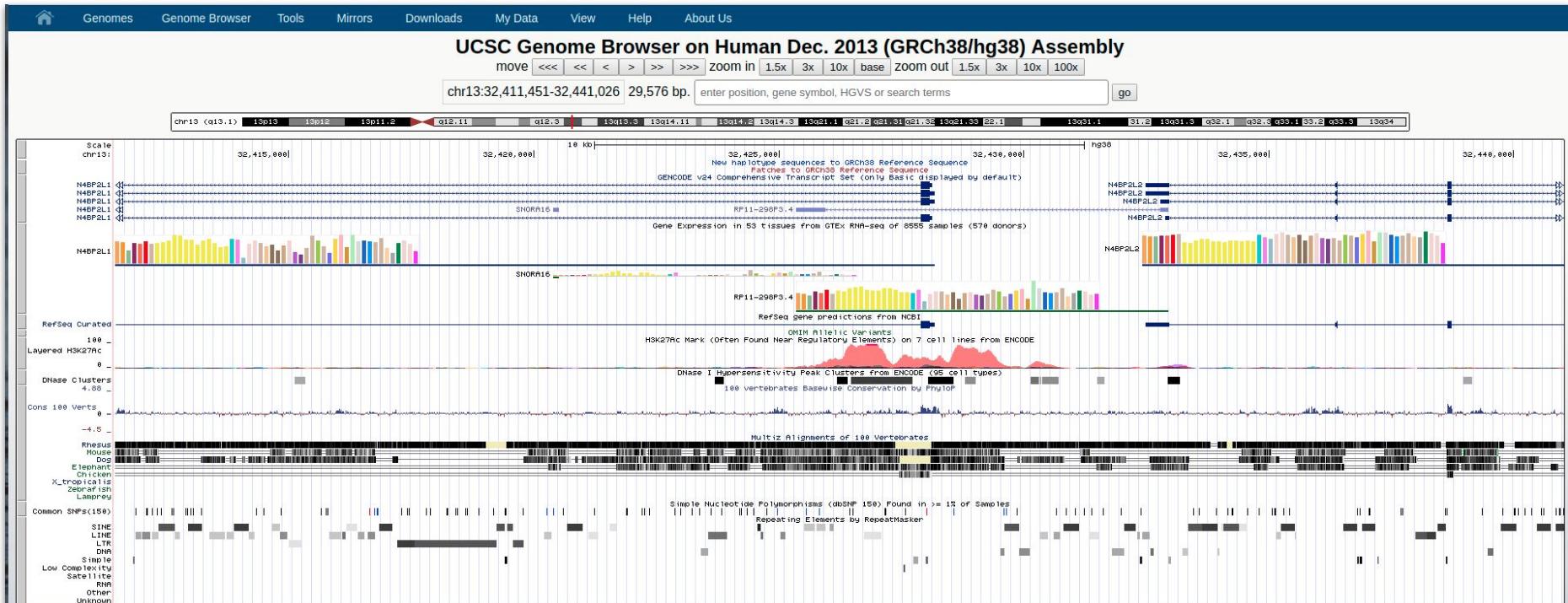
POPULAR SPECIES

Human	Mouse	Rat	Fruitfly	Worm	Yeast
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Enter species or common name

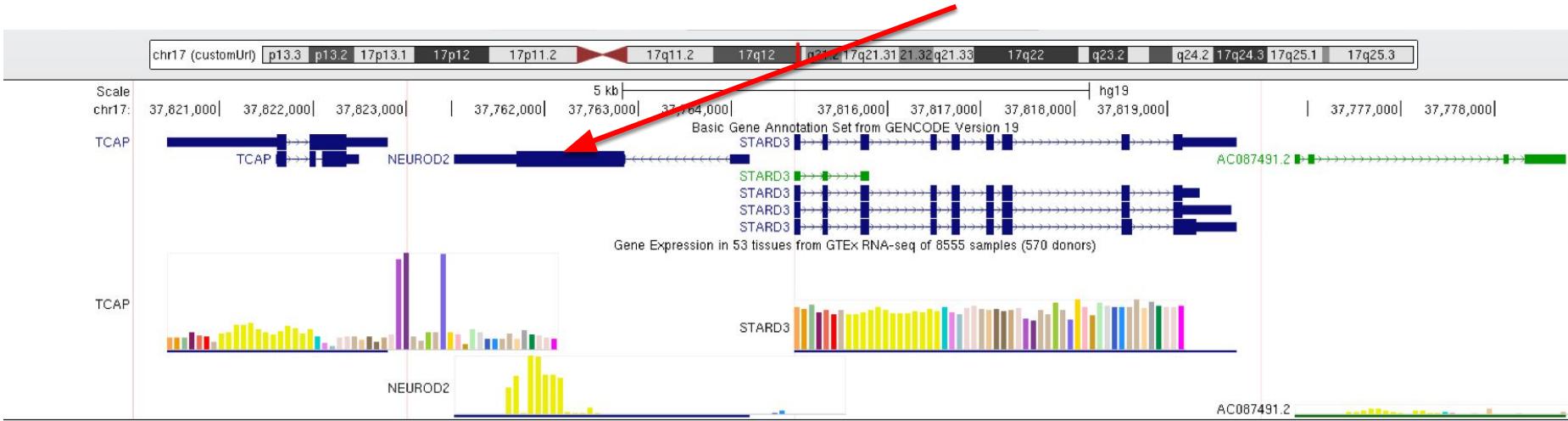
<https://genome.ucsc.edu/>

UCSC: Genome Browser



Many available tracks, highly customizable and constantly updated

UCSC: Datasets

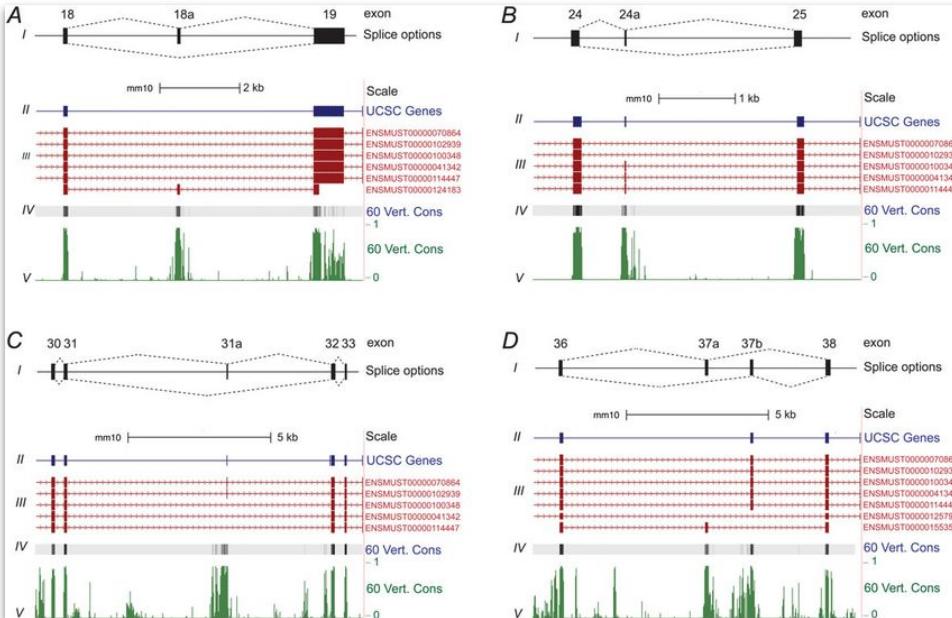


- Many datasets are easily imported to the Browser, i.e.:

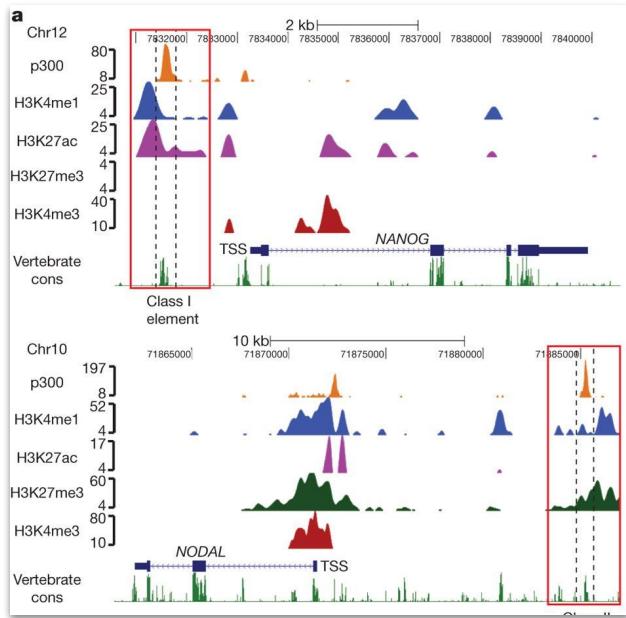
(G)ENCODE Project, GTEx, 1000 Genomes Project, Clinical variants (ClinVar), etc.

UCSC: Publication images

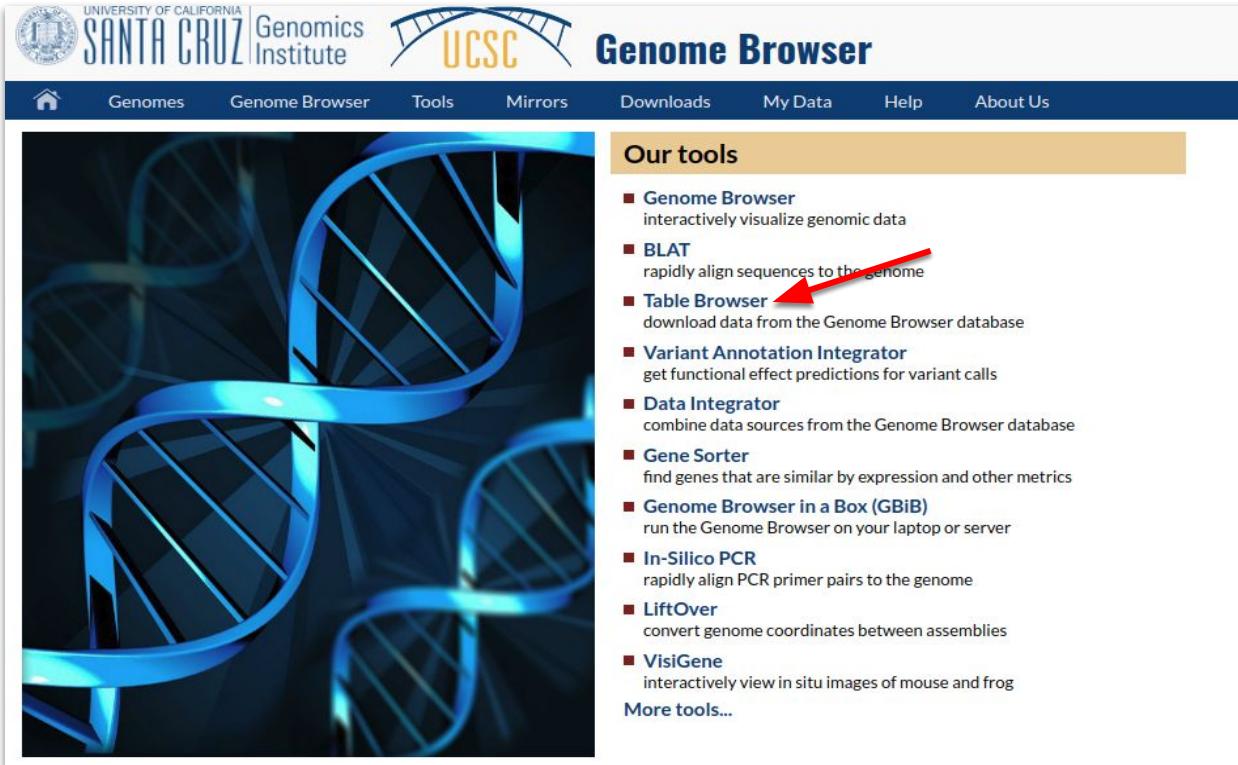
Splicing conservation



Methylation markers



UCSC: Table Browser



The image shows the UCSC Genome Browser homepage. The header includes the University of California Santa Cruz Genomics Institute logo and the UCSC logo. The main navigation menu has links for Home, Genomes, Genome Browser, Tools, Mirrors, Downloads, My Data, Help, and About Us. A large blue DNA helix graphic is on the left. On the right, there's a sidebar titled "Our tools" with a list of various bioinformatics tools. A red arrow points to the "Table Browser" entry in the list.

Our tools

- **Genome Browser**
interactively visualize genomic data
- **BLAT**
rapidly align sequences to the genome
- **Table Browser** (arrow)
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
- **Gene Sorter**
find genes that are similar by expression and other metrics
- **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
- **VisiGene**
interactively view *in situ* images of mouse and frog

[More tools...](#)

UCSC: Table Browser

Genomes Genome Browser Tools Mirrors Downloads My Data Help About Us

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 tool, the [User's Guide](#) for general information and sample queries, and the OpenHelix Table Browser [tutorial](#) for a narrated presentation of the software features and usage. 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](#). Send data to [GenomeSpace](#) for use with diverse computational tools. 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: Variation track: Common SNPs(150) add custom tracks track hubs

table: snp150Common describe table schema

region: genome position chr1:1102837-11267747 lookup define regions

identifiers (names/accessions): paste list upload list

filter: create

intersection: create

correlation: create

output format: GTF - gene transfer format (limited) Send output to Galaxy GREAT GenomeSpace

output file: danRe11.gtf (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](#).

Using the Table Browser

This section provides brief line-by-line descriptions of the Table Browser controls. For more information on using this program, see the [Table Browser User's Guide](#).

- **clade:** Specifies which clade the organism is in.
- **genome:** Specifies which organism data to use.
- **assembly:** Specifies which version of the organism's genome sequence to use.
- **group:** Selects the type of tracks to be displayed in the track list. The options correspond to the track groupings shown in the Genome Browser. Select 'All Tracks' for an alphabetical list of all available tracks in all groups. Select 'All Tables' to see all tables including those not associated with a track.
- **database:** (with "All Tables" group option) Determines which database should be used for options in table menu.
- **track:** Selects the annotation track data to work with. This list displays all tracks belonging to the group specified in the group list. Some tracks are not available when the region is set to genome due to the data provider's restrictions on sharing.
- **table:** Selects the SQL table data to use. This list shows all tables associated with the track specified in the track list. Some tables may be unavailable due to the data provider's restrictions on sharing.
- **describe table schema:** Displays schema information for the tables associated with the selected track.
- **region:** Restricts the query to a particular chromosome or region. Select genome to apply the query to the entire genome (not available for certain tracks with restrictions on data sharing). In some Human assemblies, you may select ENCODE to examine only the ENCODE Pilot regions. To limit the query to a specific position, type a chromosome name, e.g. chrX, or a chromosome coordinate range, such as chrX:100000-200000, or a gene name or other id in the text box. You can select multiple genomic regions by clicking the "define regions" button and entering up to 1,000 regions in a 3- or 4-field [BED](#) file format.
- **lookup:** Press this button after typing in a gene name or other id in the position text box to look up the chromosome position
- **identifiers (selected tracks only):** Restricts the output to table data that match a list of identifiers, for instance RefSeq accessions for the RefSeq track. If no identifiers are entered, all table data within the specified region will be displayed.

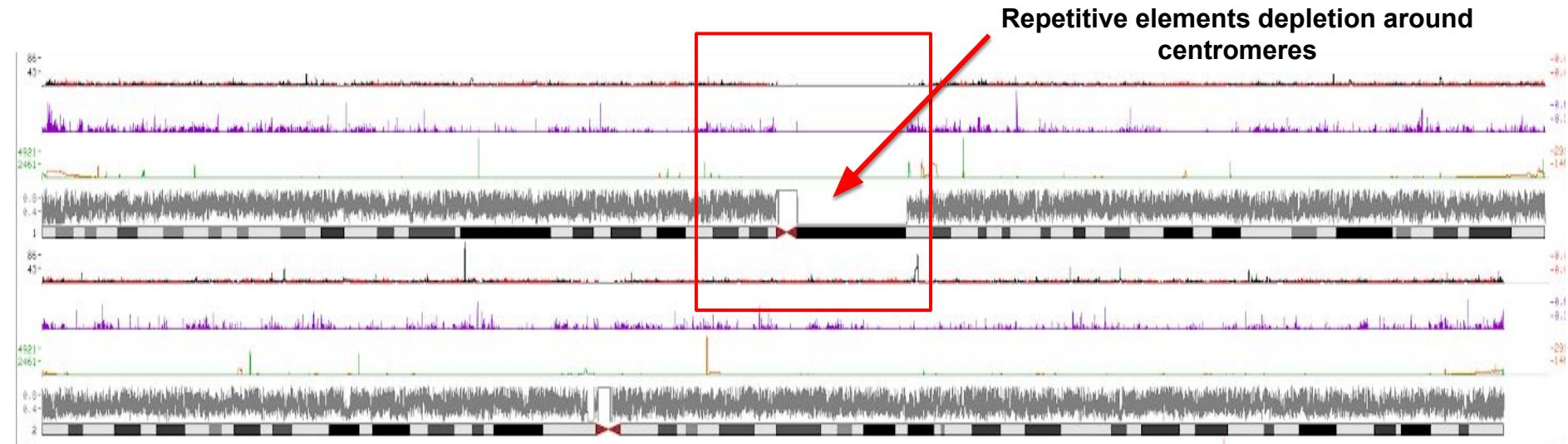
Highly customizable tracks with the table browser

UCSC: Genome Graphs



Visualization of all the chromosomes together with customizable tracks

UCSC: Customization example



Genes, CpG Islands, Repetitive elements, SNPs and many others

UCSC: Free tutorials and training



Genomes Genome Browser Tools Mirrors Downloads My Data Help About Us

Getting Help with the UCSC Genome Browser

[Host a Genome Browser Workshop!](#) [Map](#)

Do you suspect that there is a lot more to the Genome Browser than you have been using? Timeslots are available to host a Genome Browser workshop at your institution. Thanks to the funding support of NHGRI, we are able to offer hands-on Genome Browser training onsite at your institution at very affordable prices, tailored to your audience's level of expertise. [Host](#)

For more information or to submit a request to host a workshop, please visit our [sign-up page](#).

Can't host? Catch one of our training sessions in your area:

October 17, 2018	ASHG Annual Meeting, San Diego, CA. FREE workshop. See us Wednesday 7:15 am [sic] - 8:45. Room 5AB Convention Center
October 18, 2018	ASHG Annual Meeting, San Diego, CA. Part of Data visualization workshop. Thursday 2:00 pm - 3:30. Room 5AB Convention Center
October 30-31, 2018	University of Cincinnati Medical School Libraries, Cincinnati, OH. Contact
November 13-14, 2018	UCLA Institute for Quantitative and Computational Biology, Los Angeles, CA. Contact
future:	Sao Paulo; City of Hope; Plant and Animal Genomes; Arizona State; Graz, Austria; ESHG, Gothenberg, Sweden.

Online training and tutorials

Our video tutorials address some common questions we've gathered from our [mailing list](#). Along the way we try to show you interesting features of the Browser you may not have found on your own.

Visit our [YouTube channel](#) or use the links below.

Video tutorials

- [Controlling visibility of data tracks in the Browser.](#) [[transcript](#)]
- [Using the isPCR tool \(isPCR\) in the UCSC Genome Browser.](#) [[transcript](#)]
- [dbSNP resources in the UCSC Genome Browser database.](#) [[transcript](#)]
- [Using the UCSC Genome Browser Data Integrator.](#) [[transcript](#)]
- [Finding a list of genes in a region.](#) [[transcript](#)]
- [Finding exon numbers.](#) [[transcript](#)]
- [Finding all SNPs in a gene.](#) [[transcript](#)]
- [Finding SNPs upstream from a gene.](#) [[transcript](#)]

Video tutorials

- [Find which tables belong to a data track.](#) [[transcript](#)]
- [Identifying codon numbers in a gene.](#) [[transcript](#)]
- [Obtaining exon coordinates and sequences.](#) [[transcript](#)]
- [Exon-only display mode \(Multi-Region View\).](#) [[transcript](#)]
- [Viewing alternate haplotypes \(Multi-Region View\).](#) [[transcript](#)]
- Multi-Region: View **discontinuous regions** in the Browser. [[transcript](#)]
- How-to: [Genome Browser in the Cloud.](#)
- How-to: [Genome Browser Gateway.](#)

<https://genome.ucsc.edu/training/>

ENSEMBL Browser



ENSEMBL (v94)

e!Ensembl BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog

Login/Register

Search all species...

Tools

BioMart > Export custom datasets from Ensembl with this data-mining tool

BLAST/BLAT > Search our genomes for your DNA or protein sequence

Variant Effect Predictor > Analyse your own variants and predict the functional consequences of known and unknown variants

Search

All species for Go

e.g. BRCA2 or rat 5:62797383-63627669 or rs699 or coronary heart disease

All genomes

-- Select a species --

[View full list of all Ensembl species](#)
[Edit your favourites](#)

Favourite genomes

 **Human**
GRCh38.p12
[Still using GRCh37?](#)

 **Mouse**
GRCm38.p6

 **Zebrafish**
GRCz11

Ensembl is a genome browser for vertebrate genomes that supports research in comparative genomics, evolution, sequence variation and transcriptional regulation. Ensembl annotate genes, computes multiple alignments, predicts regulatory function and collects disease data. Ensembl tools include BLAST, BLAT, BioMart and the Variant Effect Predictor (VEP) for all supported species.

Ensembl Release 94 (October 2018)

- New fish: 38 new and updated fish genomes
- GENCODE update 29 for human and M19 for mouse
- Additional pathogenicity predictors for missense variants
- New transcription factor binding motifs from SELEX
- Gene trees using HMMs

[More release news](#) on our blog

Other news from our blog

- 08 Oct 2018: [Ensembl & Friends at ASHG](#)
- 03 Oct 2018: [Ensembl 94 is out!](#)
- 28 Sep 2018: [Cool things the VEP can do: variant prioritisation with G2P](#)

<https://www.ensembl.org/>

ENSEMBL Browser: BRCA2

[e!Ensembl](#) BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog

Login/Register

Human (GRCh38.p12) ▾

Location: 13:32,313,623-32,401,805 Gene: BRCA2 Jobs ▾

Location-based displays

- Whole genome
- Chromosome summary
- Region overview
- **Region in detail**
- Comparative Genomics
 - Synteny
 - Alignments (image)
 - Alignments (text)
 - Region Comparison
- Genetic Variation
 - Variant table
 - Resequencing
 - Linkage Data
 - Markers
- Other genome browsers
 - UCSC
 - NCBI
 - Ensembl GRCh37

Configure this page

Custom tracks

Export data

Share this page

Bookmark this page

Chromosome 13: 32,313,623-32,401,805

Assembly exceptions. Chr. 13 p13 p11.2 q12.3 q13.3 q14.11 q14.2 q14.3 q21.1 q21.33 q31.1 q31.3 q34

Region in detail

1.00 Mb

1.00 Mb

Chromosome bands

Contigs

Genes (Comprehensive set from GENCODE 29)

Regulatory Build

Gene Legend

Regulation Legend

Location: 13:32313779-32401961 Go Gene: Go

Forward strand

RF00017 > < FRY-AS1
< AC002525.1
EEF1DP3 >

AL137143.8 > AL137143.8 > AL138692.26 > AL44_212.9 > AL137247.14 > AL353665.13 > Z84467.1 > Z75889.1 > AL138820.11

CAR1L
BRCA2 >

< RF00190 ATP8A2P2 >
< AL137247.1 < N4BP2L2-IT2
IF1IP1 > < N4BP2L2
< N4BP2L1

PDS5B >

AL138820.1 >
< RNY1P4

merged Ensembl/Havana
pseudogene

processed transcript
RNA gene

CTCF
Open Chromatin
Promoter Flank

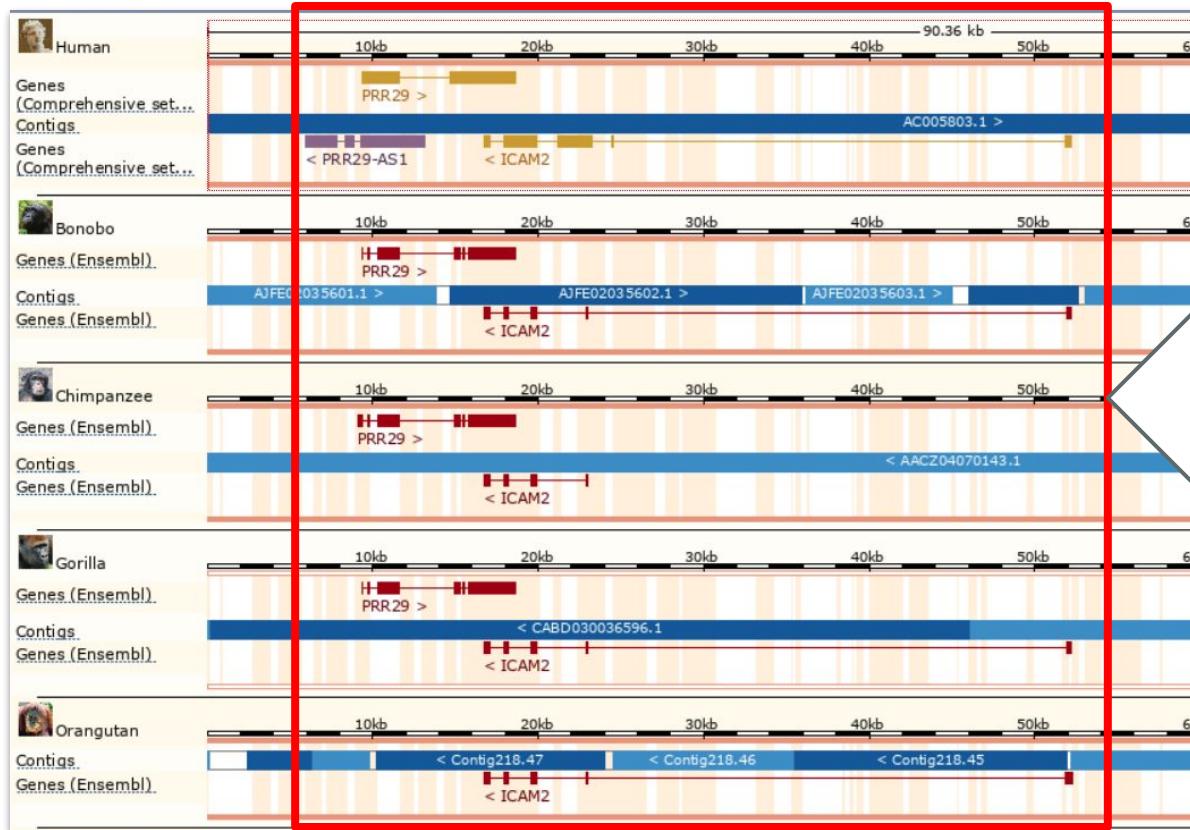
Enhancer
Promoter
Transcription Factor Binding Site

ENSEMBL: BioMart

The screenshot shows the ENSEMBL BioMart search interface. On the left, there's a sidebar with sections for 'Dataset' (Human genes (GRCh38.p12)), 'Filters' (None selected), and 'Attributes' (Gene stable ID, Transcript stable ID). Below that is another 'Dataset' section with [None Selected]. At the top, there are navigation links: New, Count, Results, URL, XML, Perl, and Help. The main area has a heading 'Please restrict your query using criteria below' followed by a note '(If filter values are truncated in any lists, hover over the list item to see the full text)'. It lists several filter categories with checkboxes: REGION, GENE, PHENOTYPE, GENE ONTOLOGY, MULTI SPECIES COMPARISONS, PROTEIN DOMAINS AND FAMILIES, and VARIANT.

Equivalent to the table browser from UCSC, highly customizable tracks with lots of information

ENSEMBL: Synteny in great apes

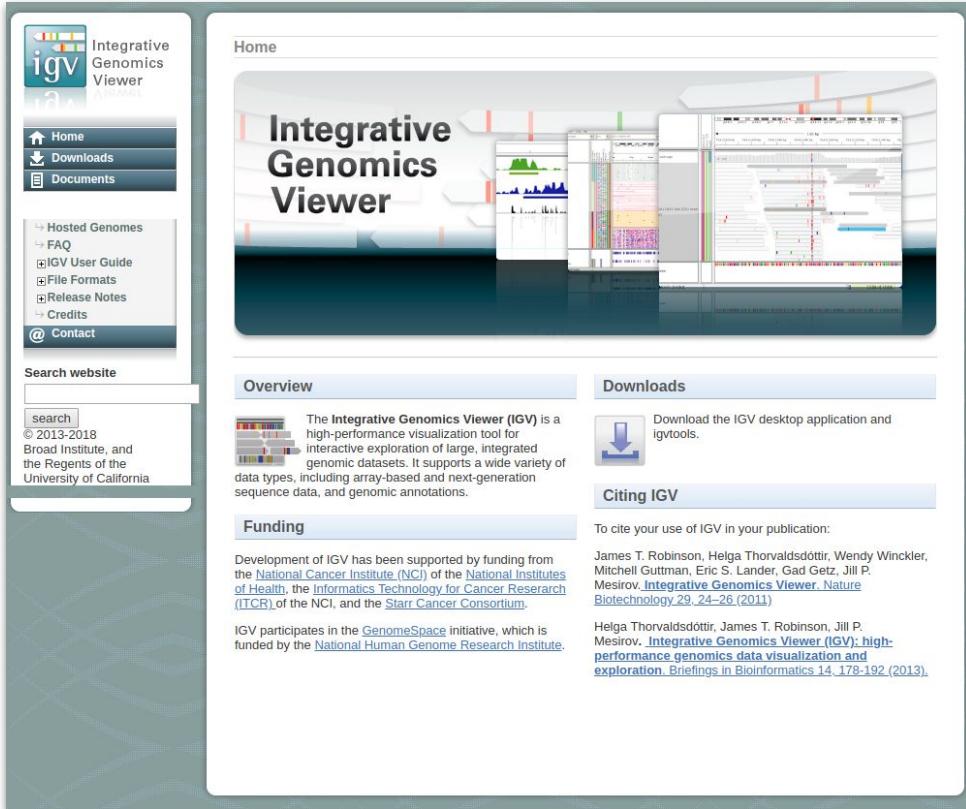


Syntetic block

Integrative Genomics Viewer



Integrative Genomics Viewer (IGV)

The screenshot shows the homepage of the IGV website. At the top left is the IGV logo. The main header reads "Home" and "Integrative Genomics Viewer". Below the header is a large image showing a screenshot of the IGV software interface with multiple tracks of genomic data. To the left of the main content area is a sidebar with links to "Hosted Genomes", "FAQ", "IGV User Guide", "File Formats", "Release Notes", "Credits", and "Contact". Below the sidebar is a search bar labeled "Search website" with a "search" button. At the bottom left is a footer containing copyright information: "© 2013–2018 Broad Institute, and the Regents of the University of California".

- Java-based genomic browser
- Supports a wide variety of NGS data
- Genomic annotation tracks
- Easy to use
- Flexible

<http://software.broadinstitute.org/software/igv/userguide>

IGV supports different data formats

- ChIP-Seq, RNA-Seq (TDF)
- Copy number (CN, SNP)
- LOH data (LOH)
- Mutation data (MUT)
- RNAi data (GCT format)
- Sequence alignment data (SAM/BAM)
- GWAS data (GWAS)
- Any numeric data (IGV, TAB, WIG)

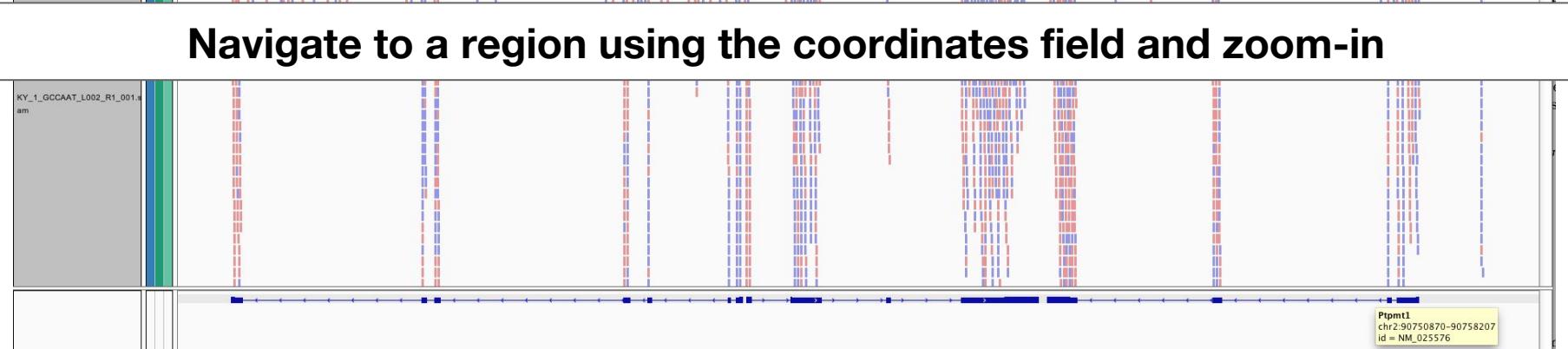
BAM	IGV
BED	LOH
BedGraph	MAF
bigBed	MUT
bigWig	narrowPeak
chrom.sizes	PSL
CN	RES
Cytoband	RNA Structure
FASTA	SAM
GCT	SNP
CRAM	TAB
genePred	VCF
GFF/GTF	WIG
GWAS	

Custom File Formats

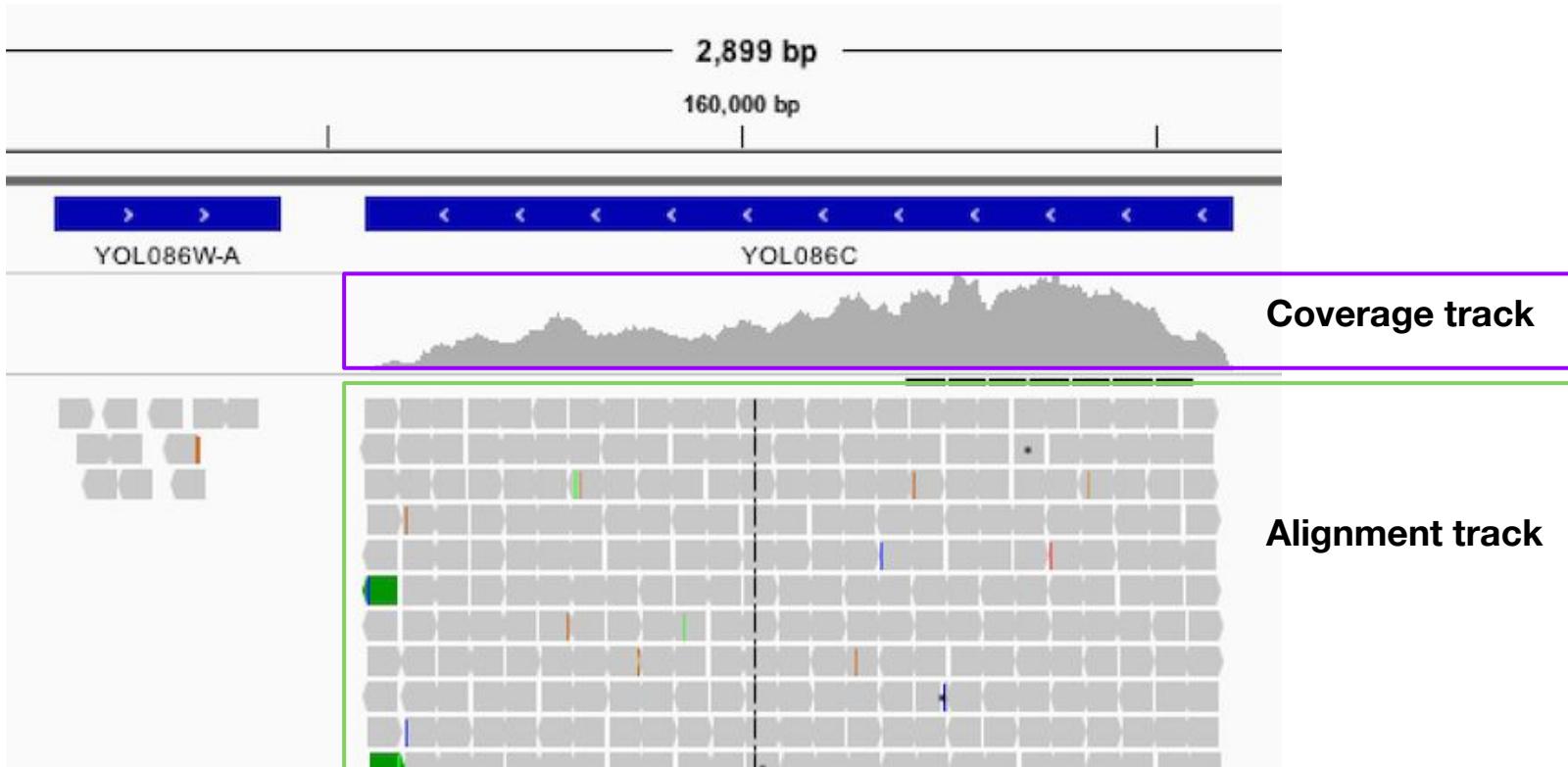
IGV display of reads and coverage



Navigate to a region using the coordinates field and zoom-in



IGV display of reads and coverage



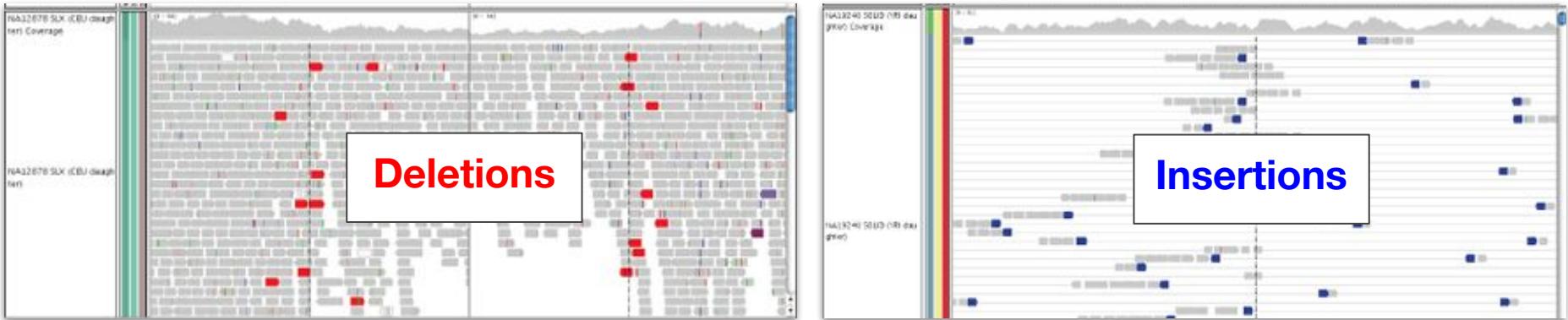
Interpreting pairs of reads

Category	Illumina	
LR		LR = Normal reads → <u>Left and right</u> of the DNA fragment
LL		LL, RR = Inversion of the sequenced DNA fragment
RR		
RL		RL = Duplication or translocation of the DNA fragment

All relative to the alignment with the reference genome

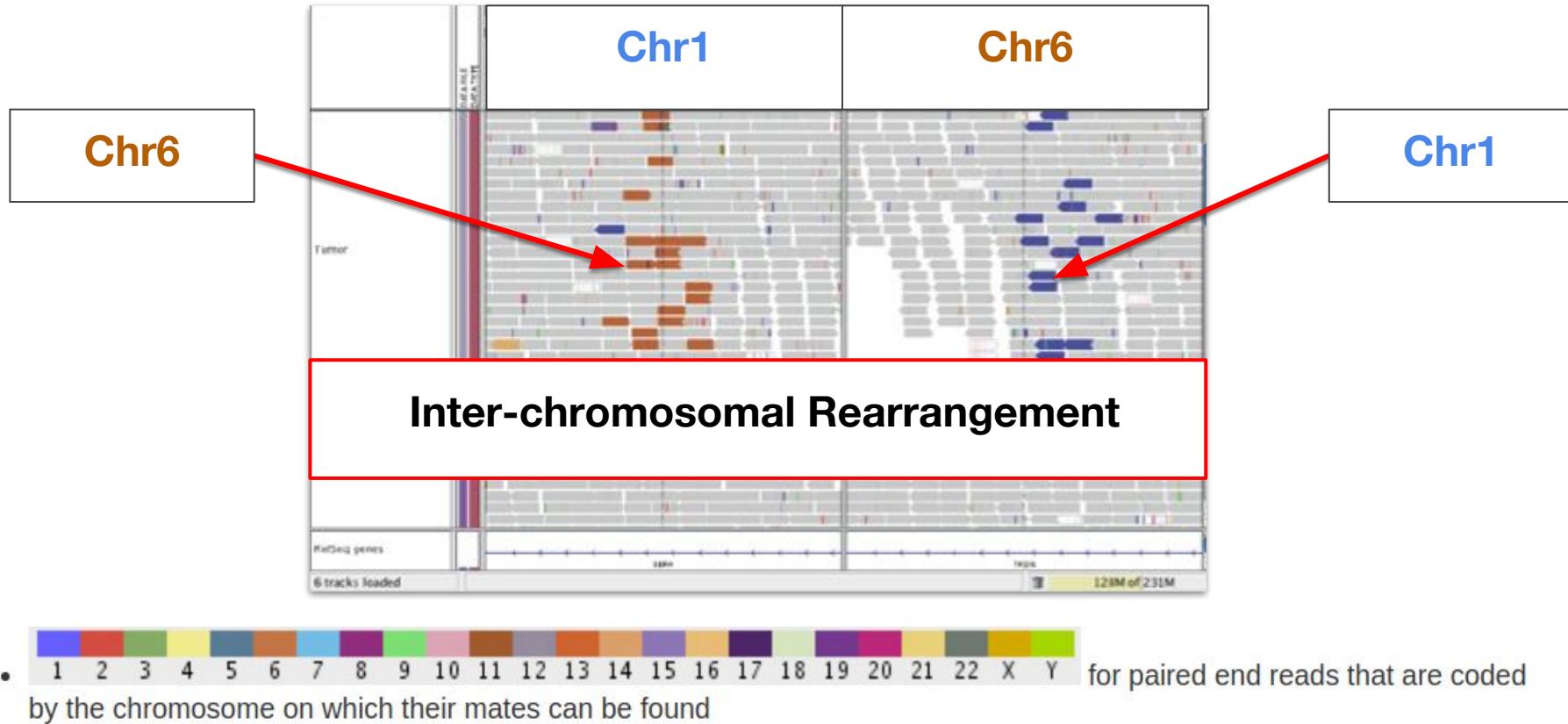
Interpreting read colours (I)

- **DELETION** for an inferred insert size that is larger than expected (deletion)
- **INSERTION** for an inferred insert size that is smaller than expected (insertion)

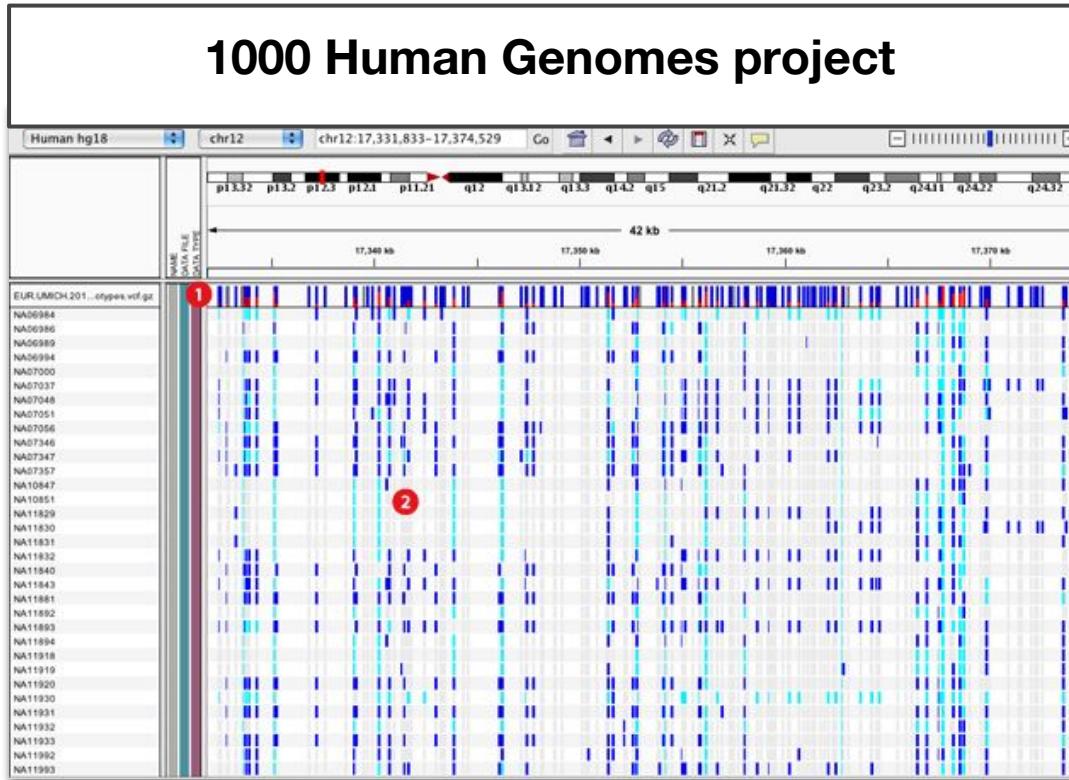


- for paired end reads that are coded by the chromosome on which their mates can be found

Interpreting read colours (II)



Allele frequencies

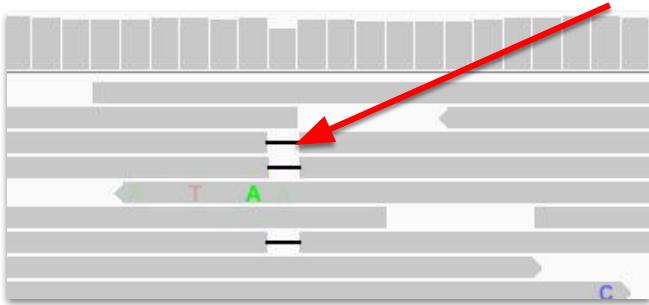


Allele fractions for a single locus

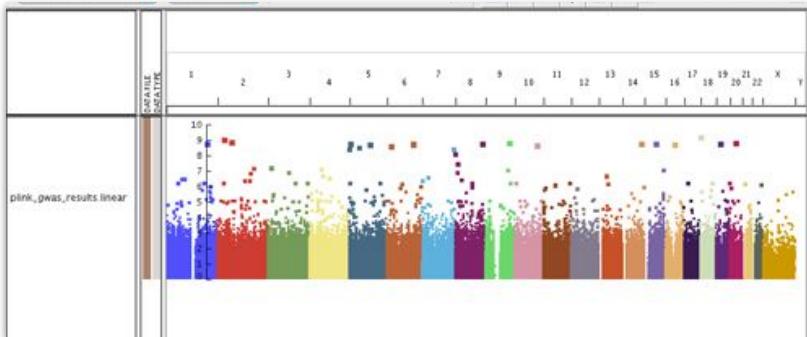
- **Blue:** Heterozygous
- **Cyan:** Homozygous
- **Gray:** Reference
- **Transparent:** Filtered entries

Other features

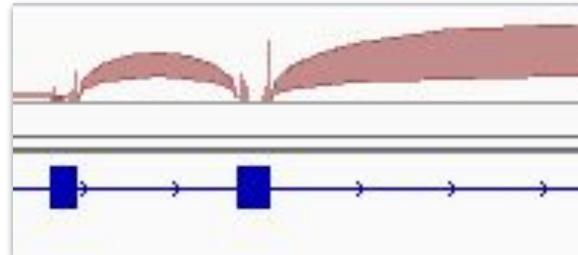
Gaps



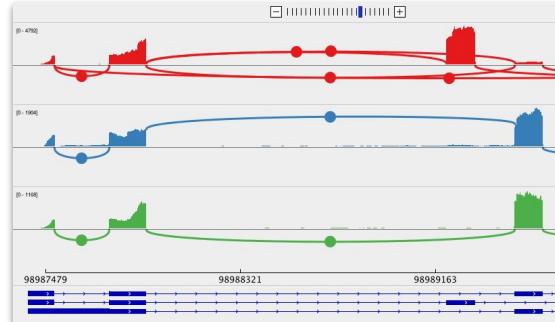
GWAS data (Manhattan plots)



Splicing junctions

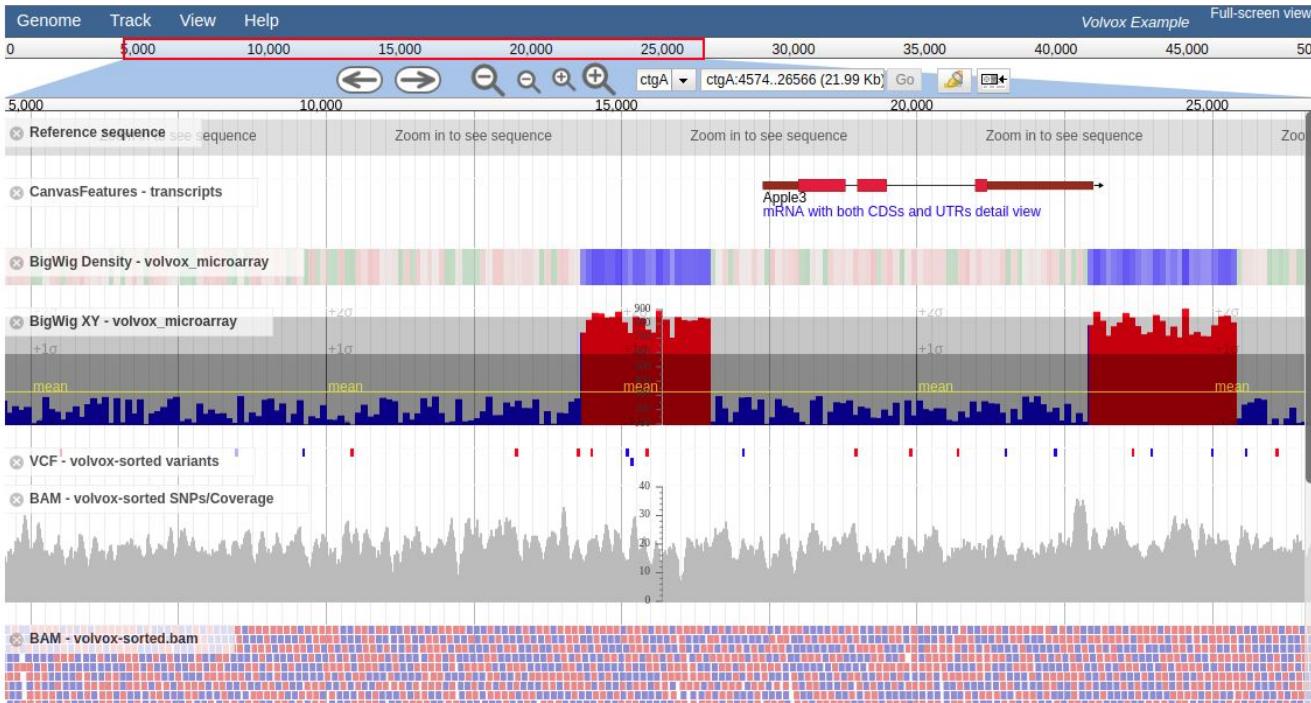


Sashimi plots



**Many other alternatives
to visualize genomes**

JBrowse Genome Browser

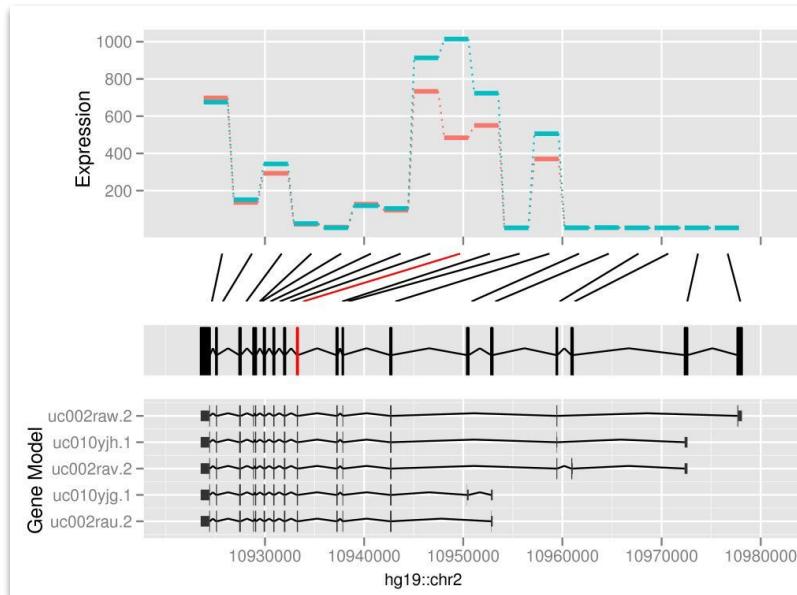


<https://jbrowse.org/>

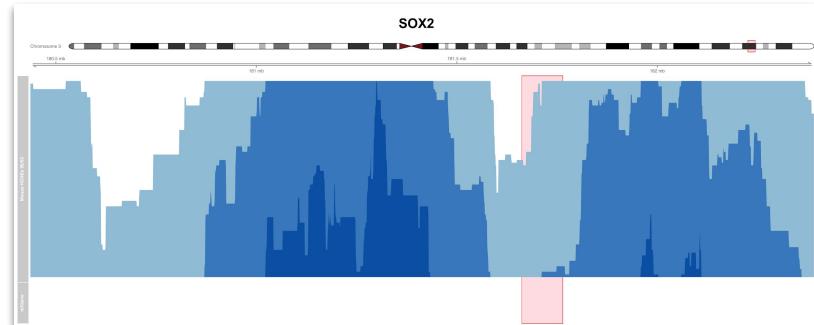
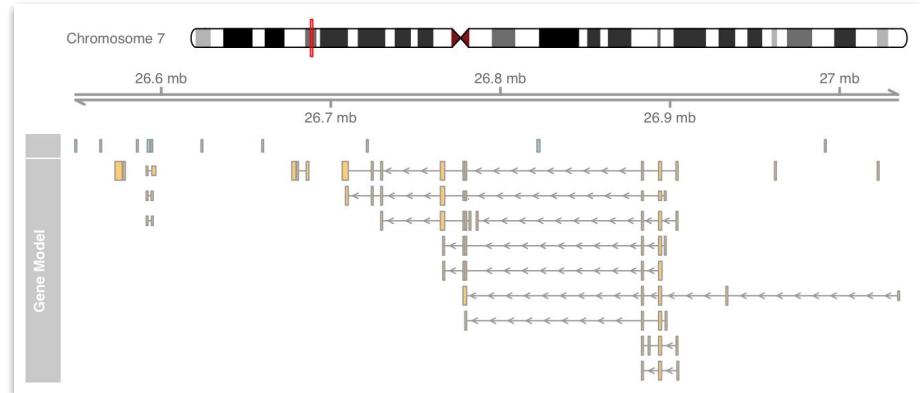
D3 Genome Browser in Python



ggbio:

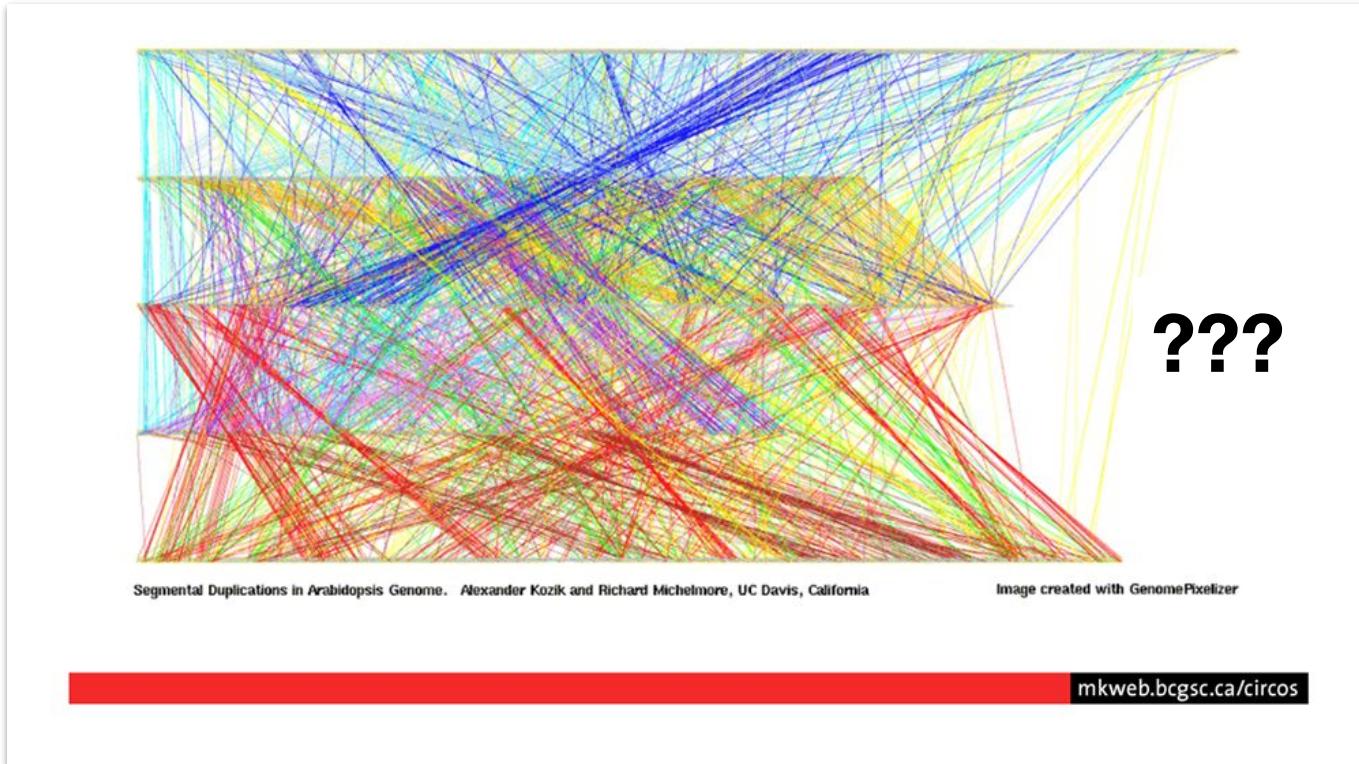


Gviz:



...and many others.

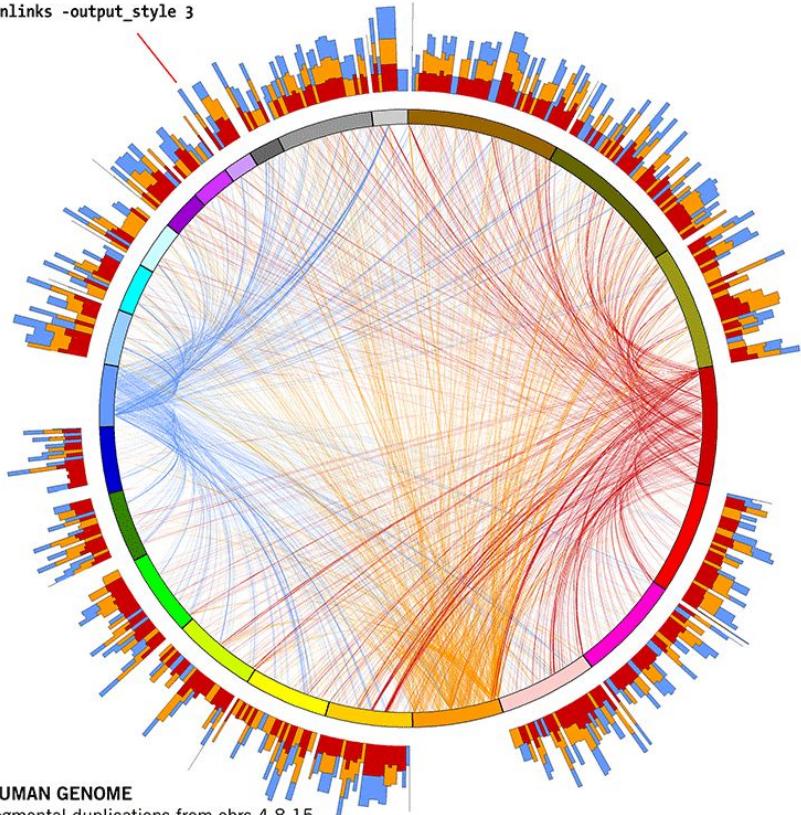
There are times when linear doesn't work



Circles to the rescue

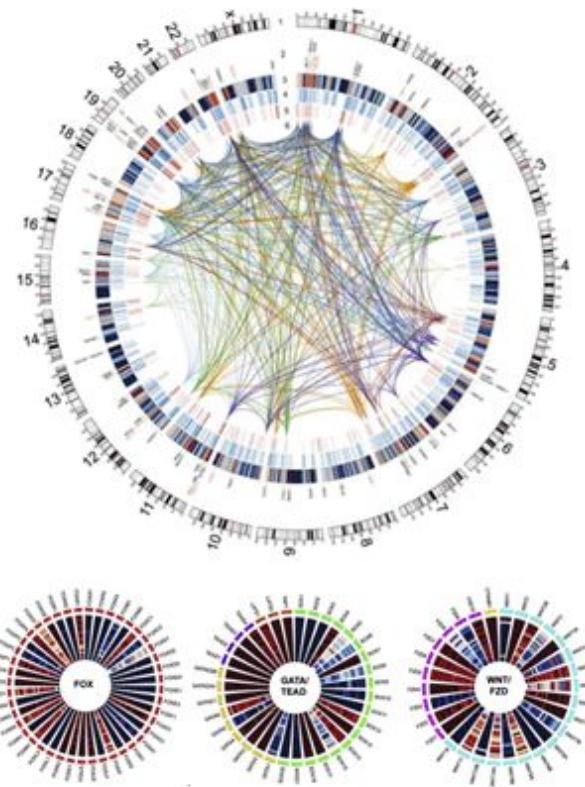


binlinks -output_style 3



HUMAN GENOME

segmental duplications from chrs 4,8,15
(repeats >1kb with >90% identity)



<http://circos.ca/>

Hands-on session

Please, go to: https://github.com/carlalbc/URPP_tutorials/