

Introduction to the Integrative Genomics Viewer (IGV)

Did you follow the instructions in the pre-workshop fact sheet?

If not, go to **www.igv.org/workshops/ASHG2016**

1. Download the workshop data: **Exercises.zip**
2. If you do not have IGV installed, see: **DownloadAndLaunch.pdf**
3. If you have not used IGV before, see: **BasicsExercise.pdf**

Introduction to the Integrative Genomics Viewer (IGV)

Jim Robinson, UCSD
Helga Thorvaldsdóttir, Broad Institute

ASHG Annual Meeting,
Vancouver, October 2016

Disclosure for:

ASHG Interactive Workshop:
Introduction to the
Integrative Genomics Viewer (IGV)

No relevant conflicts to disclose

The logo consists of the letters 'IGV' in a bold, white, sans-serif font, centered within a dark gray square.

IGV

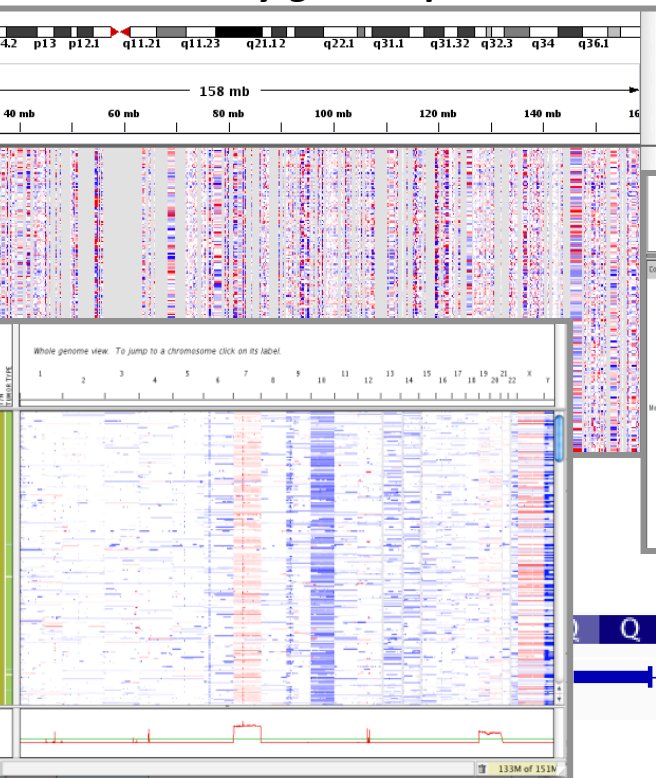
**Interactive
visual exploration of
integrated
genomic data**

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IGV

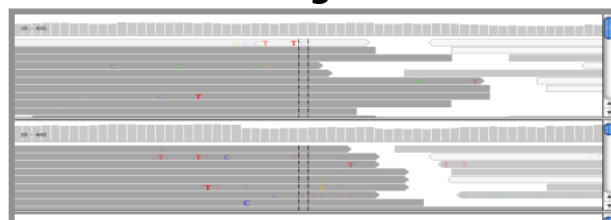
**Interactive
visual exploration of
integrated
genomic data**

Microarray gene expression



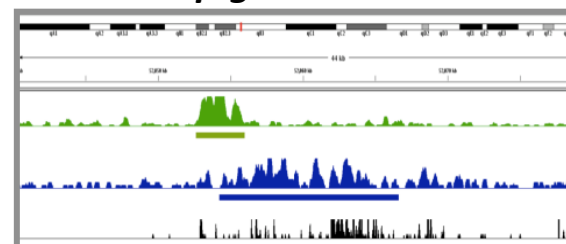
Copy number

NGS alignments

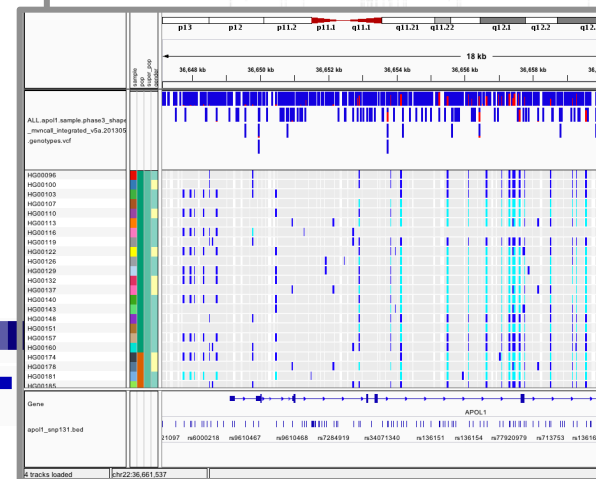


RNA-seq

Epigenomics



Gene annotations



Variants and genotypes

IGV data sources: File formats

- IGV supports many different file formats.

- [BAM](#)
- [BED](#)
- [BedGraph](#)
- [bigBed](#)
- [bigWig](#)
- [Birdsuite Files](#)
- [broadPeak](#)
- [CBS](#)
- [CN](#)
- [Custom File Formats](#)
- [Cytoband](#)
- [FASTA](#)
- [GCT](#)
- [genePred](#)
- [GFF/GTF](#)
- [GISTIC](#)
- [Goby](#)
- [GWAS](#)
- [IGV](#)
- [LOH](#)
- [MAF \(Multiple Alignment Format\)](#)
- [MAF \(Mutation Annotation Format\)](#)
- [Merged BAM File](#)
- [MUT](#)
- [narrowPeak](#)
- [PSL](#)
- [SAM](#)
- [Sample Info \(Attributes\) file](#)
- [SEG](#)
- [SNP](#)
- [TAB](#)
- [TDF](#)
- [Track Line](#)
- [Type Line](#)
- [VCF](#)
- [WIG](#)
- [chrom.sizes](#)

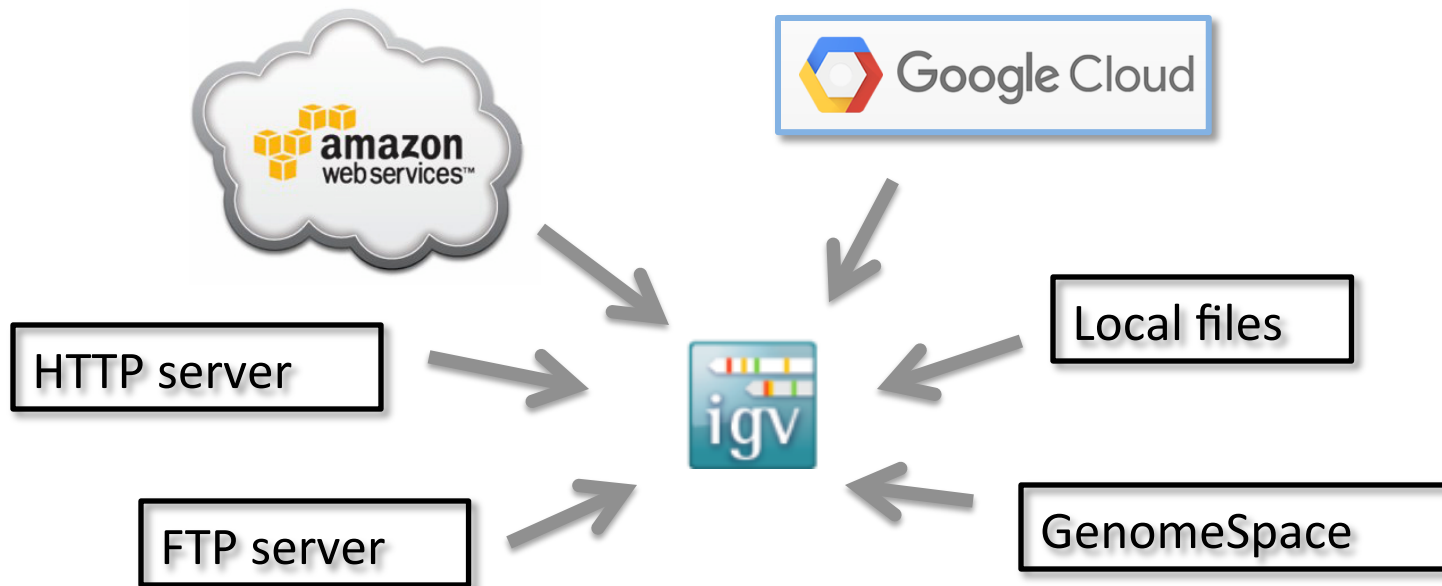
- For more info see: [*www.broadinstitute.org/igv/FileFormats*](http://www.broadinstitute.org/igv/FileFormats)

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IGV

**Interactive
visual exploration of
integrated
genomic data**

IGV data sources



- View **local** files without uploading.
- View **remote** files without downloading whole dataset.

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IGV

Interactive

visual exploration of

integrated

genomic data

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IGV

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IGV

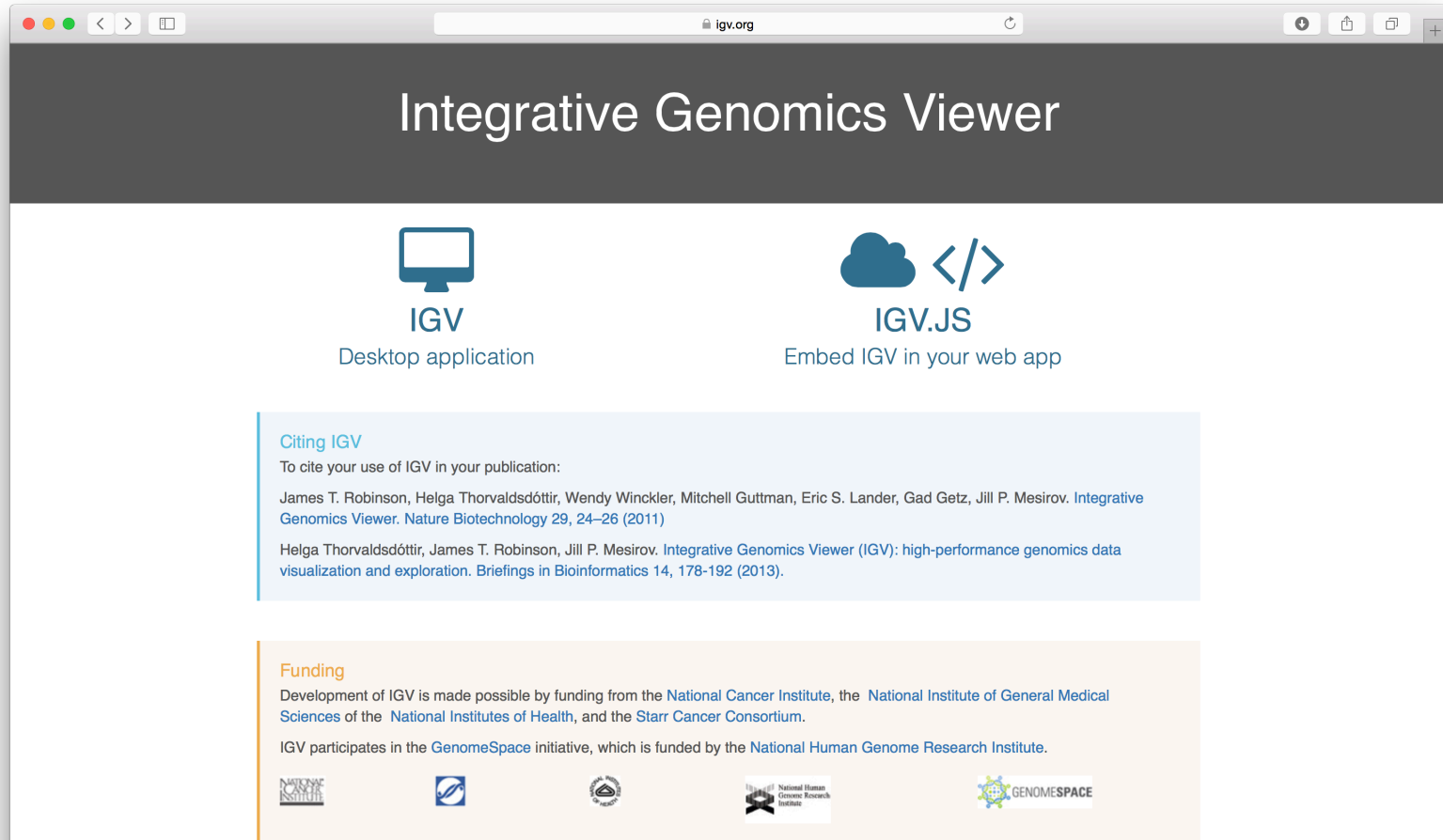
Interactive

visual exploration of

integrated

genomic data

www.igv.org



The screenshot shows a web browser window with the URL `igv.org`. The page has a dark header with the title "Integrative Genomics Viewer". Below the header, there are two main options: "IGV Desktop application" with a monitor icon, and "IGV.JS" with a cloud and code icon, labeled "Embed IGV in your web app". The page also features a "Citing IGV" section with a light blue background, providing citation information for publications. At the bottom, there is a "Funding" section with a light orange background, listing the organizations that fund the project, including the National Cancer Institute, the National Institutes of Health, the Starr Cancer Consortium, and the National Human Genome Research Institute. Logos for these organizations and the GENOMESPACE initiative are displayed at the bottom of the funding section.

Integrative Genomics Viewer



IGV
Desktop application



IGV.JS
Embed IGV in your web app

Citing IGV

To cite your use of IGV in your publication:

James T. Robinson, Helga Thorvaldsdóttir, Wendy Winckler, Mitchell Guttman, Eric S. Lander, Gad Getz, Jill P. Mesirov. [Integrative Genomics Viewer](#). *Nature Biotechnology* 29, 24–26 (2011)

Helga Thorvaldsdóttir, James T. Robinson, Jill P. Mesirov. *Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration*. *Briefings in Bioinformatics* 14, 178-192 (2013).

Funding

Development of IGV is made possible by funding from the [National Cancer Institute](#), the [National Institute of General Medical Sciences](#) of the [National Institutes of Health](#), and the [Starr Cancer Consortium](#).

IGV participates in the [GenomeSpace](#) initiative, which is funded by the [National Human Genome Research Institute](#).



www.igv.org

The screenshot shows the IGV website homepage. At the top, the URL 'igv.org' is in the browser's address bar. The main heading is 'Integrative Genomics Viewer'. Below this, there are two primary options: 'Desktop application' (highlighted with a red circle) and 'IGV.JS'. The 'Desktop application' section features a computer monitor icon and the text 'IGV Desktop application'. The 'IGV.JS' section features a cloud and code icon and the text 'IGV.JS Embed IGV in your web app'. Below these are two informational sections: 'Citing IGV' with citation information and 'Funding' with details about the project's financial support. At the bottom, there are logos for the National Cancer Institute, National Institutes of Health, Starr Cancer Consortium, National Human Genome Research Institute, and GENOMESPACE.

Integrative Genomics Viewer

Desktop application


IGV
Desktop application


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www.broadinstitute.org/igv



Integrative
Genomics
Viewer



Home



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Hosted Genomes

FAQ

IGV User Guide

File Formats

Release Notes

IGV for iPad

Credits

Contact

Search website

search



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Home

Integrative Genomics Viewer



Overview



The **Integrative Genomics Viewer (IGV)** is a high-performance visualization tool for interactive exploration of large, integrated genomic datasets. It supports a wide variety of data types, including array-based and next-generation sequence data, and genomic annotations.

Downloads



Download the IGV desktop application and igvtools.

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IGV participates in the [GenomeSpace](#) initiative, which is funded by the [National Human Genome Research Institute](#).

Citing IGV

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James T. Robinson, Helga Thorvaldsdóttir, Wendy Winckler, Mitchell Guttman, Eric S. Lander, Gad Getz, Jill P. Mesirov. [Integrative Genomics Viewer \(IGV\): high-performance genomics data visualization and exploration](#). *Nature Biotechnology* 29, 24–26 (2011)

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www.igv.org

Integrative Genomics Viewer



IGV
Desktop application



IGV.JS
Embed IGV in your web app

**JavaScript
version**

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Integrative Genomics Viewer

Desktop application


IGV
Desktop application


IGV.JS
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Using IGV: The Basics

Review of pre-workshop exercise

Download and launch IGV

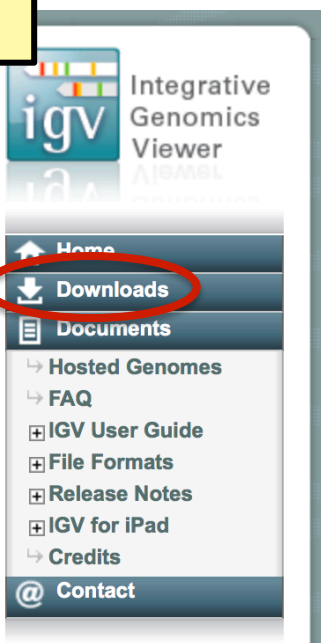
1 Go to www.igv.org and click on



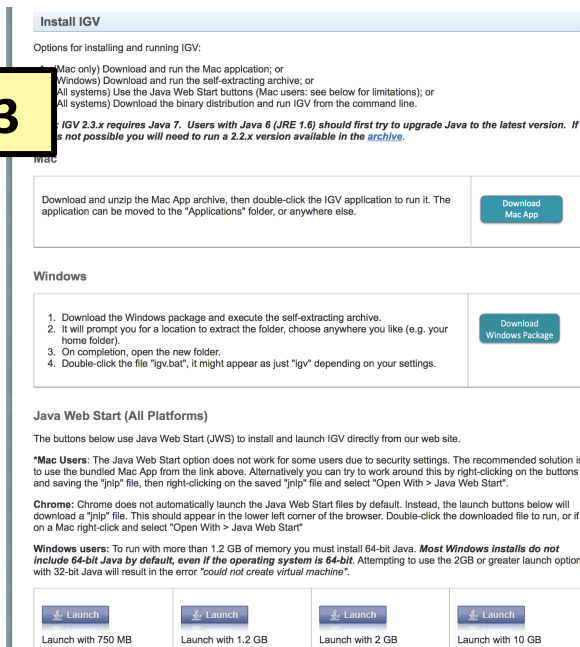
IGV

Desktop application

2



3



Download
Mac App

or

Download
Windows Package

or

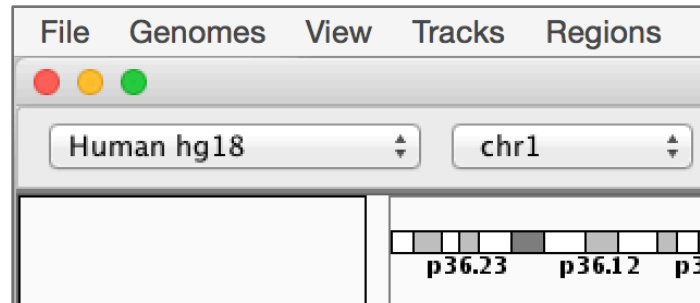
Launch

Select the reference genome

4

The genome drop-down menu is in the upper left corner

Select **Human hg18**

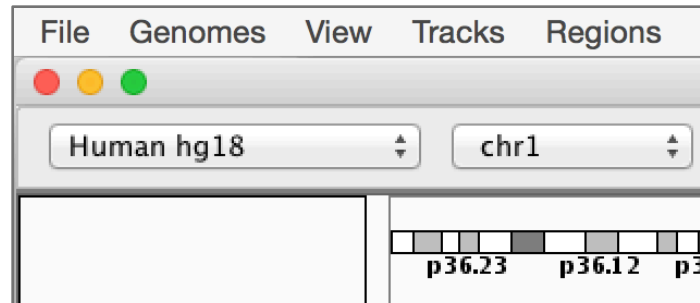


Select the reference genome

4

The genome drop-down menu is in the upper left corner

Select **Human hg18**



!

THERE'S MORE TO REFERENCE GENOMES

The genome menu is short when you first install IGV.

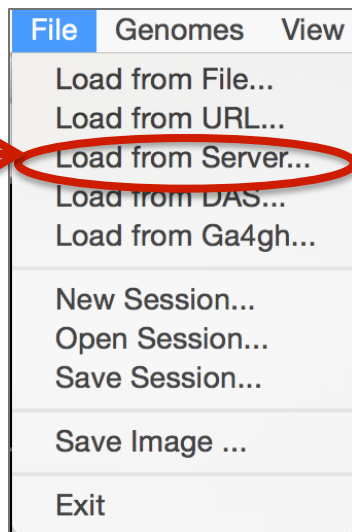
But IGV **hosts dozens of genomes** and allows you to **load other genomes** too.

More about that later

Load data

5

select

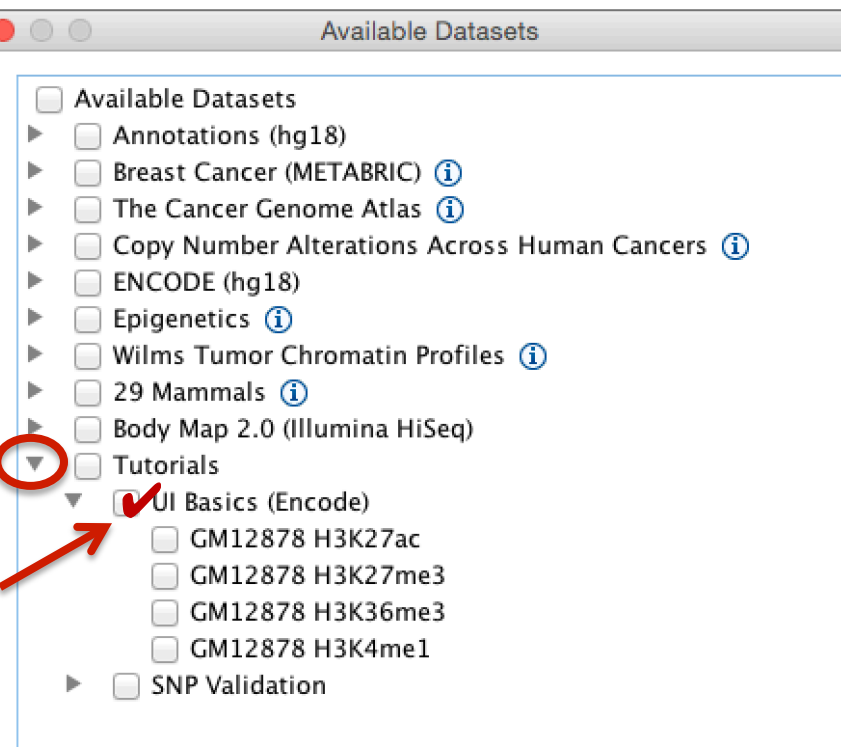


6

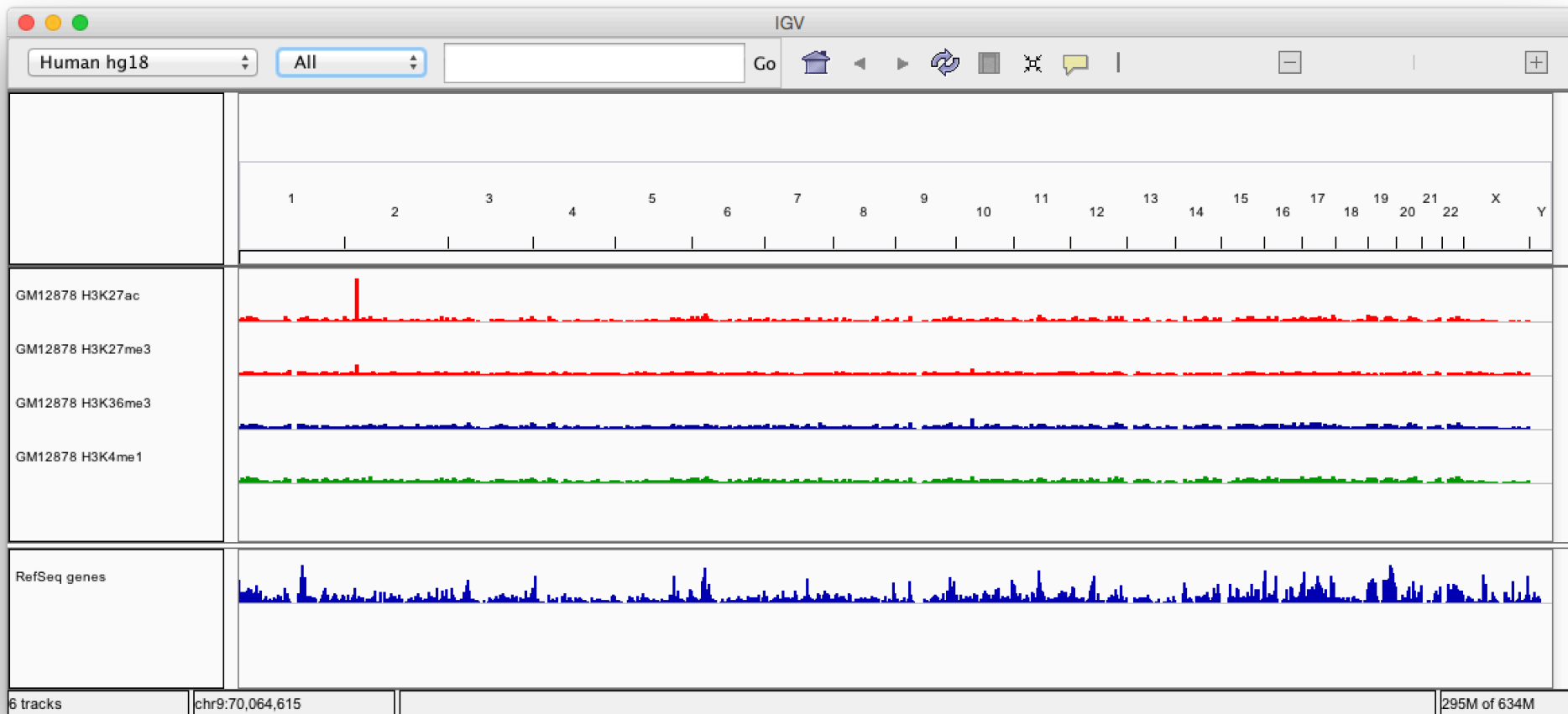
open

and

select



Load data



Navigate

Zoom in / out

From whole genome view
to base pair resolution

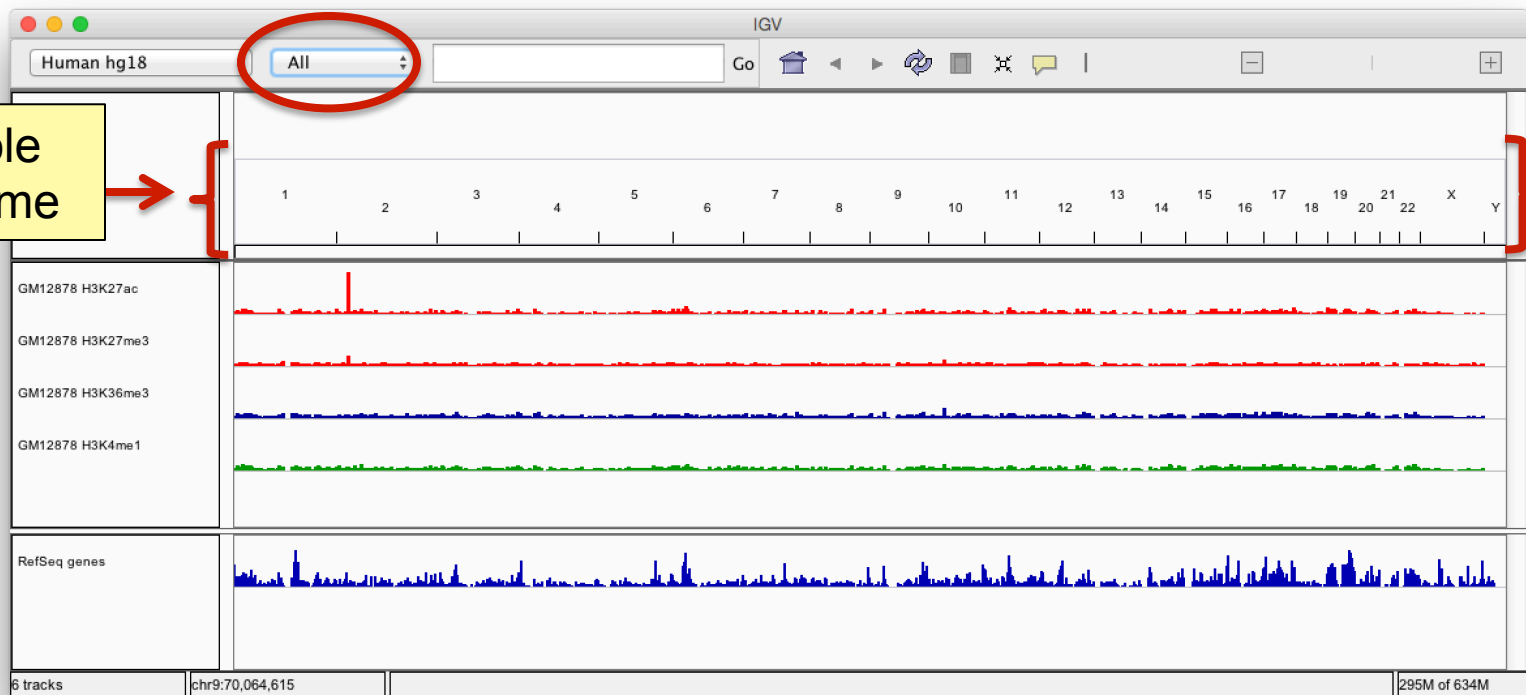
Scroll / pan

View neighboring regions

Jump to locus

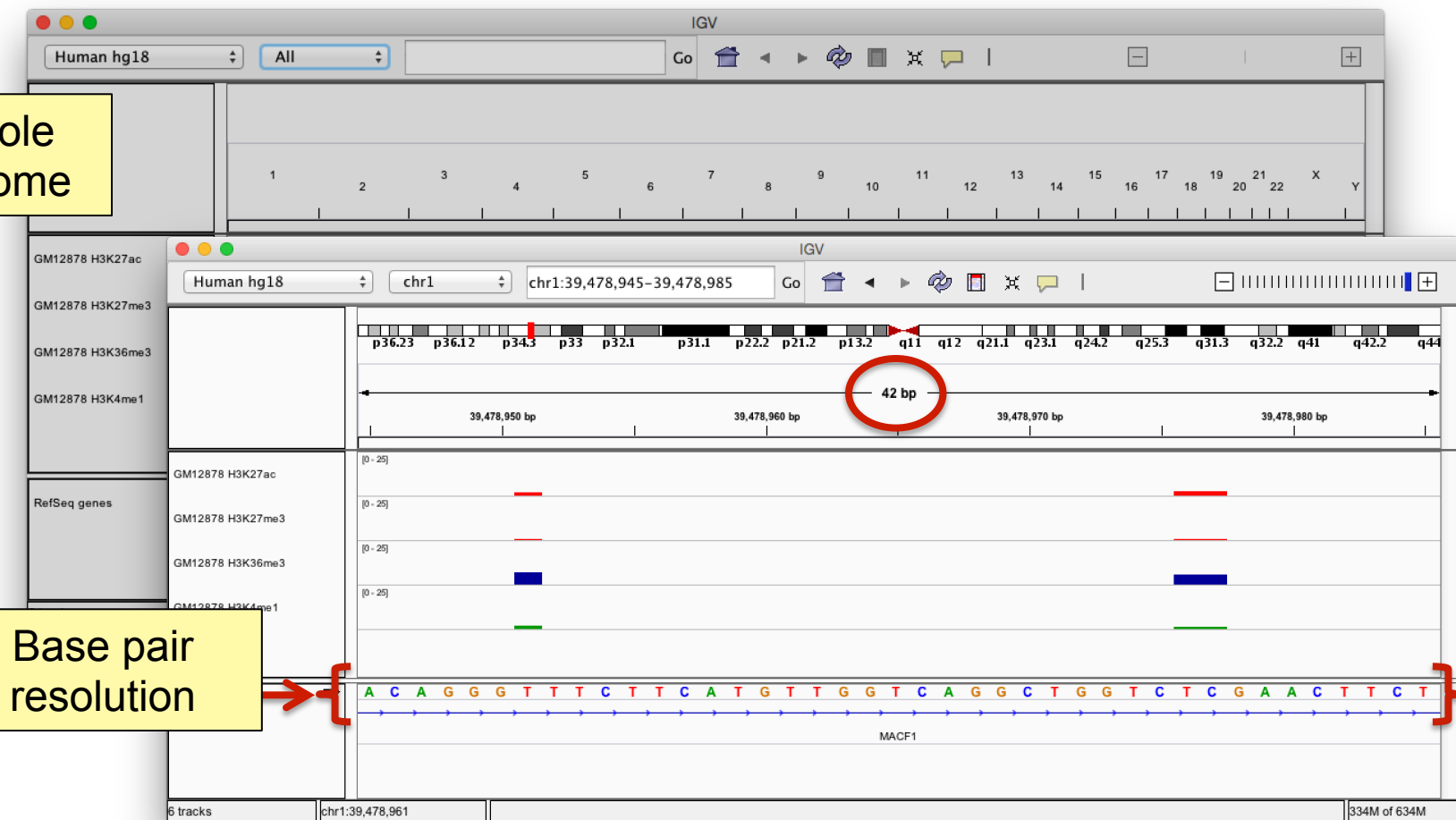
Enter coordinates or name

Navigate



Navigate

Whole
genome

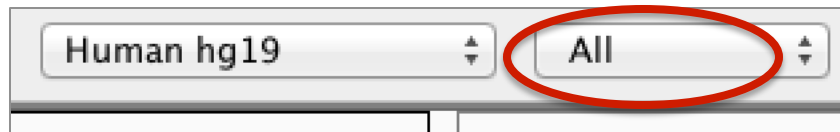


Base pair
resolution

Navigate

View whole
genome

select



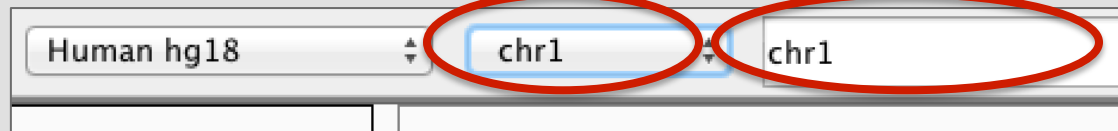
A screenshot of a web interface. On the left, a dropdown menu displays 'Human hg19'. To its right, another dropdown menu shows 'All' and is circled in red. A red arrow points from the 'select' label above to this red circle.

or click



View whole
chromosome

select

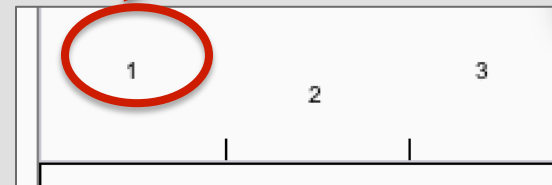


A screenshot of a web interface. On the left, a dropdown menu displays 'Human hg18'. To its right, a dropdown menu shows 'chr1' and is circled in red. A red arrow points from the 'select' label above to this red circle.

or enter

chr1

or click

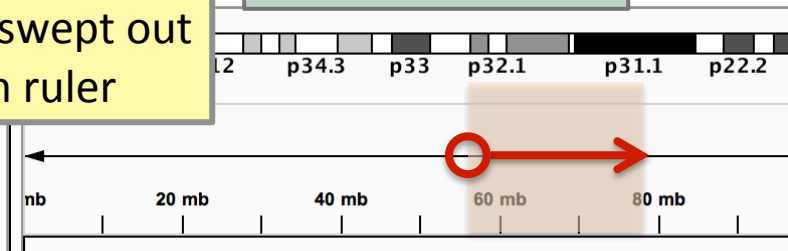


A screenshot of a web interface showing a list of numbers: 1, 2, and 3. The number '1' is circled in red. A red arrow points from the 'click' label above to this red circle.

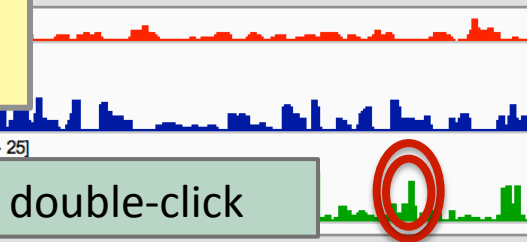
Navigate

Zoom in to
area swept out
in ruler

click & drag



Zoom in on area
of interest



double-click

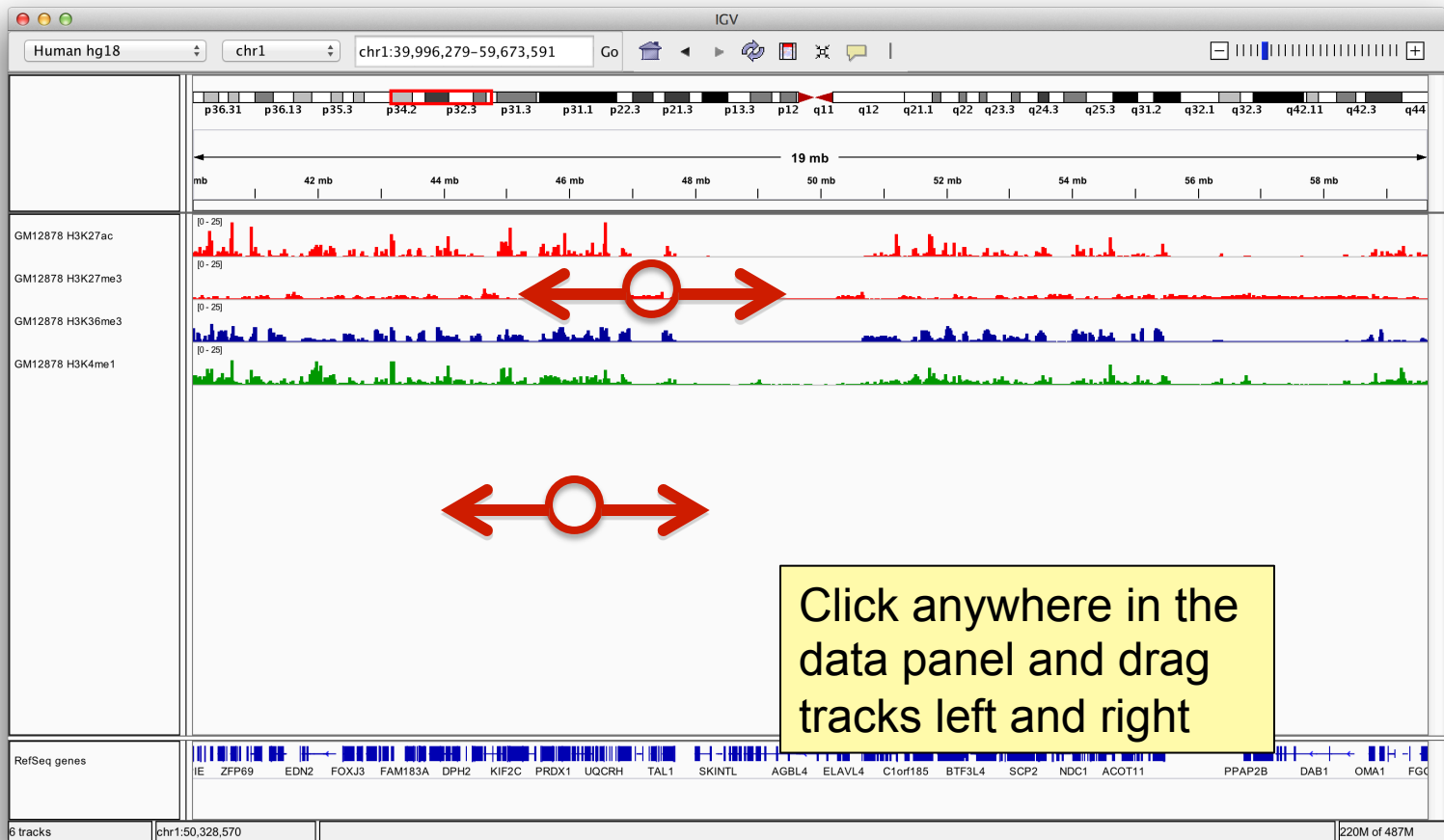
Zoom out

alt-click

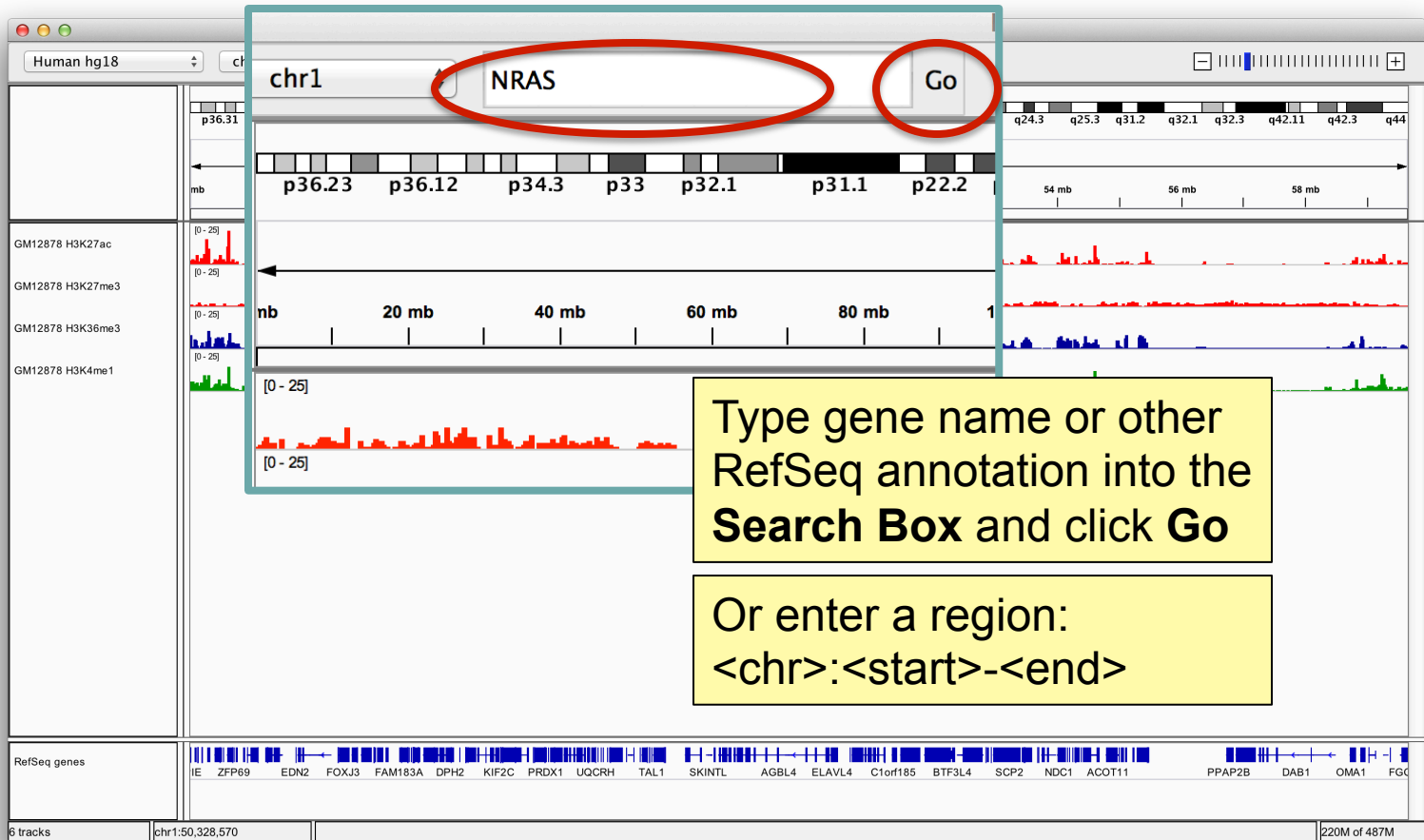
Zoom in or out using the
“railroad track”



Navigate

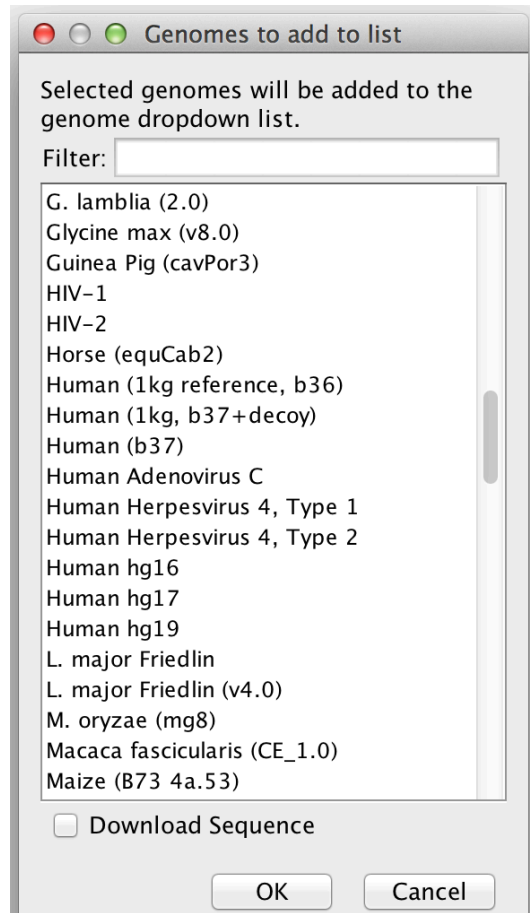
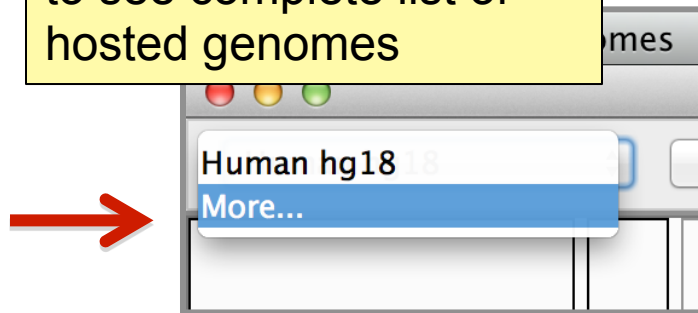


Navigate



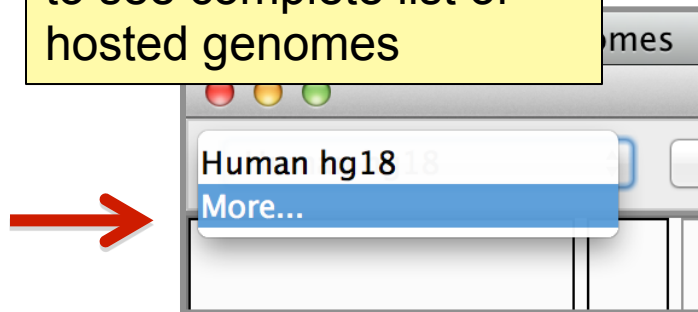
More options for reference genomes

1. Click on **More** in the genome dropdown menu to see complete list of hosted genomes

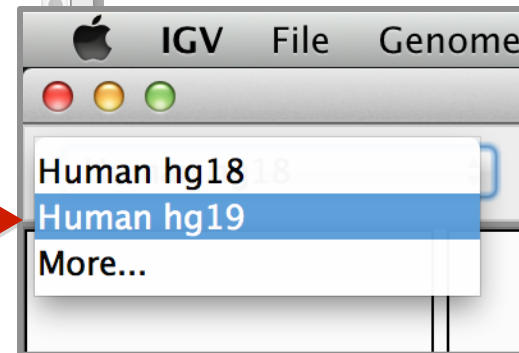
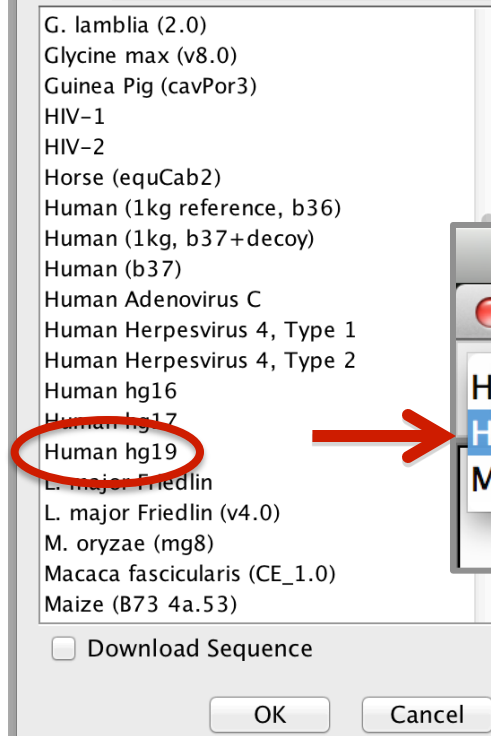


More options for reference genomes

1. Click on **More** in the genome dropdown menu to see complete list of hosted genomes

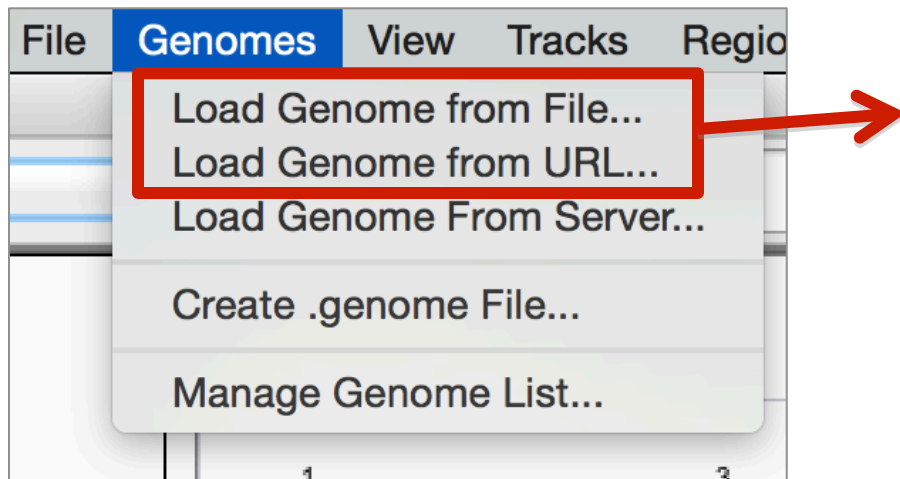


2. Select entry from the list to load genome and add to the dropdown menu



More options for reference genomes

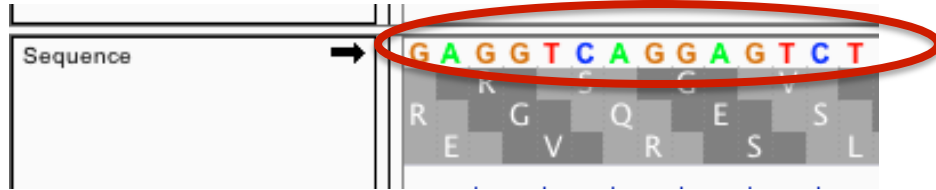
Genomes menu



Load From File or
Load From URL
Load genome from
indexed FASTA file

Reference sequence

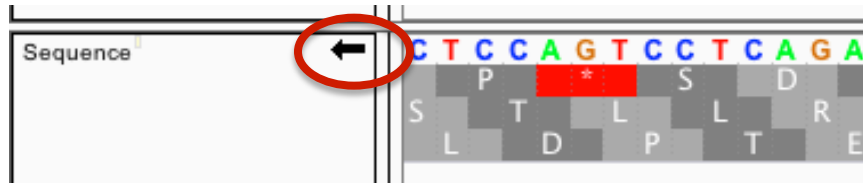
Click anywhere on the sequence to see a 3 frame translation.



By default the sequence for the forward strand is shown.



Click the arrow on the left to reverse the strand.



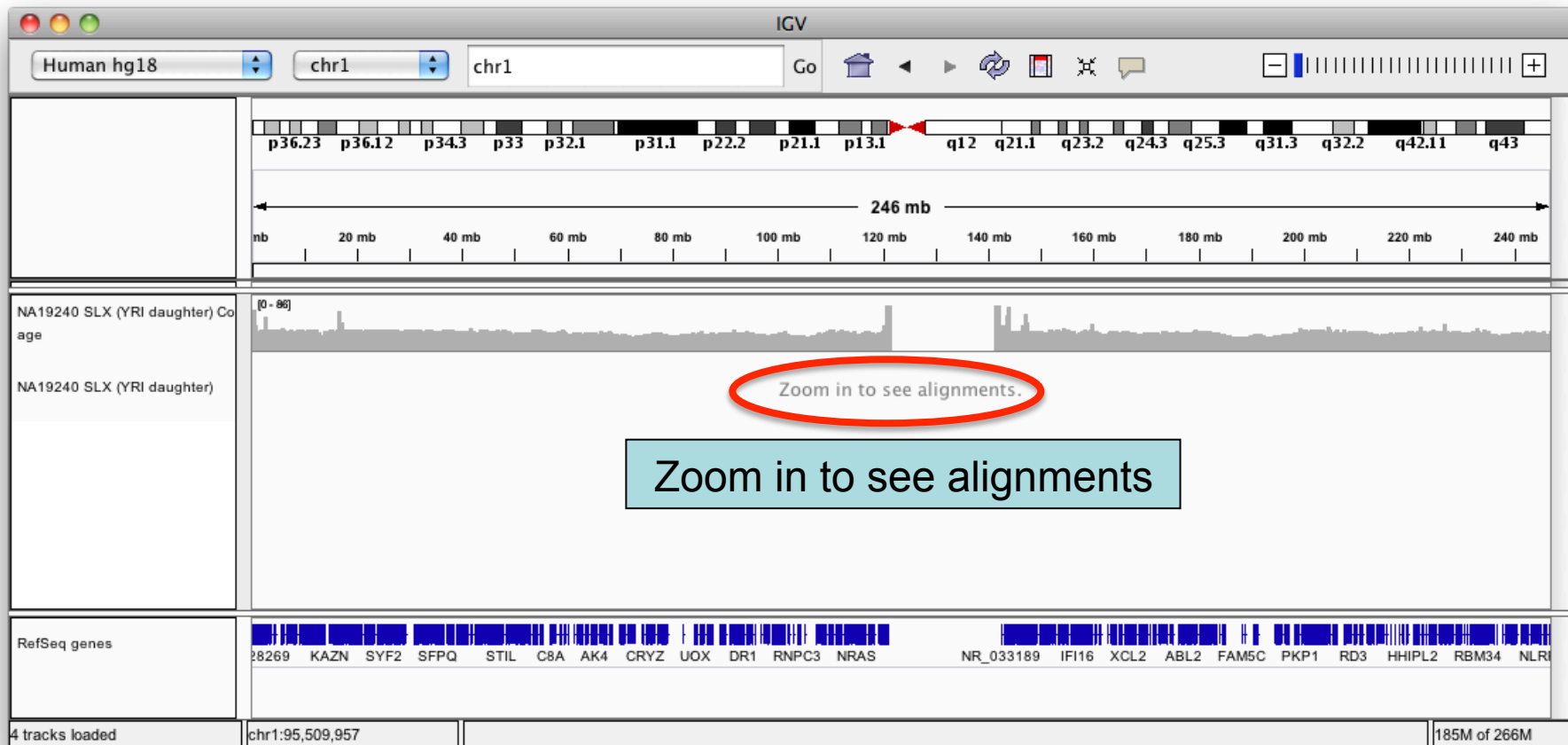
Viewing multiple regions



Viewing Next Generation Sequencing (NGS) Data

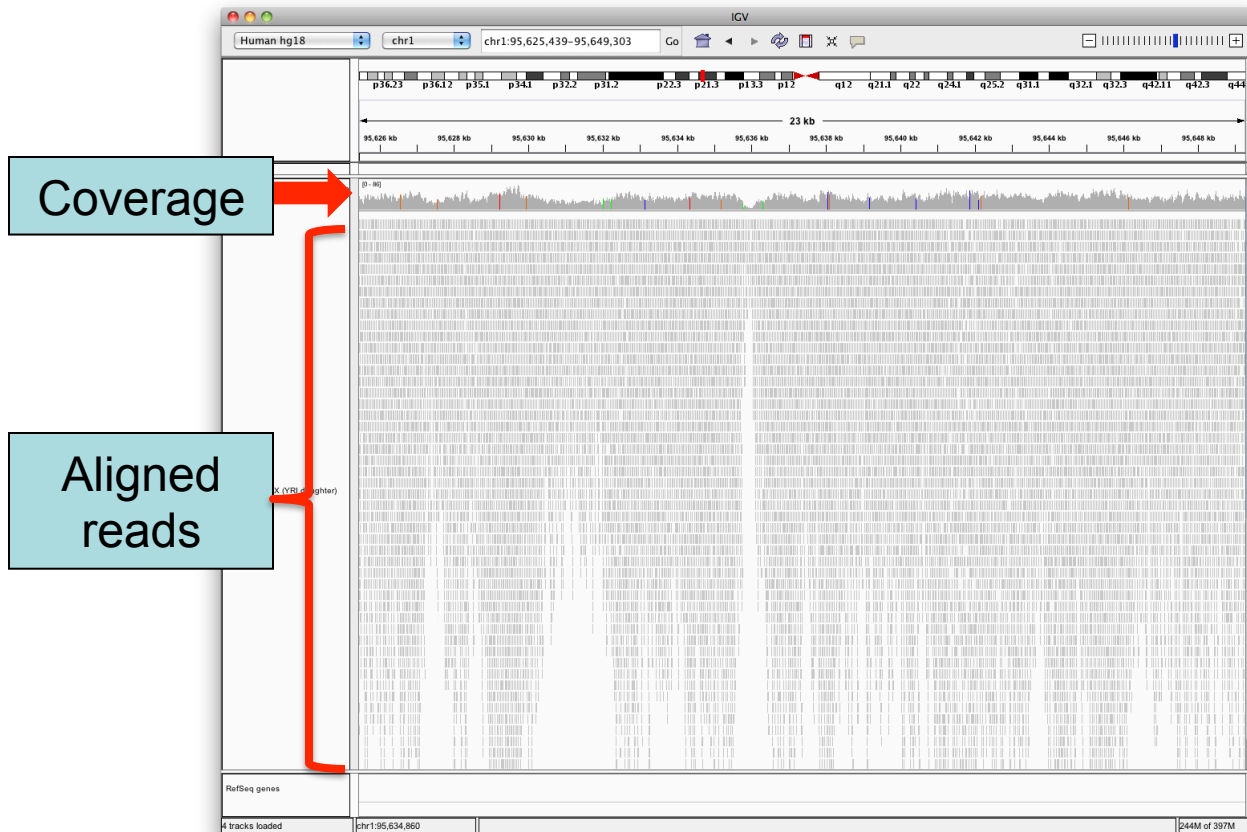
Viewing alignments

Whole chromosome view



Viewing alignments

Zoom in to view alignments



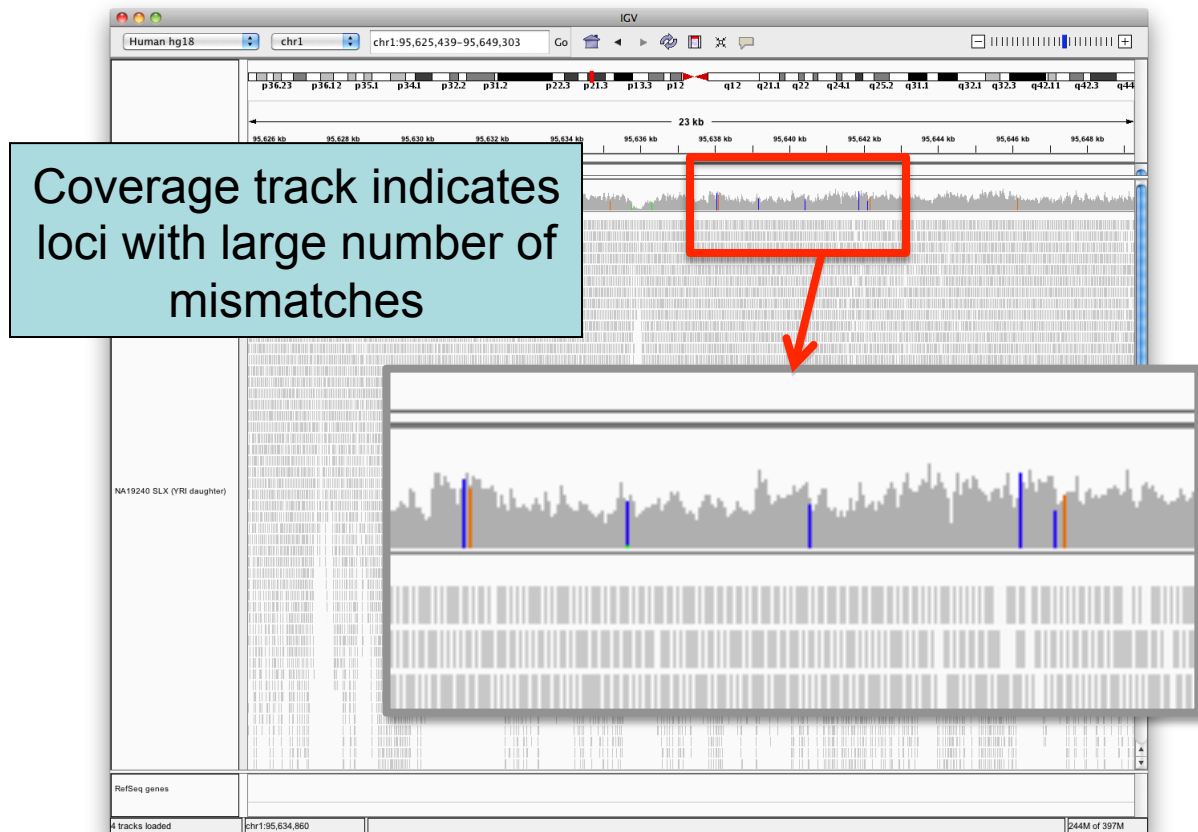
Viewing alignments

Zoom in to see more detail

Bases that do not match the reference sequence are highlighted by color



Viewing alignments

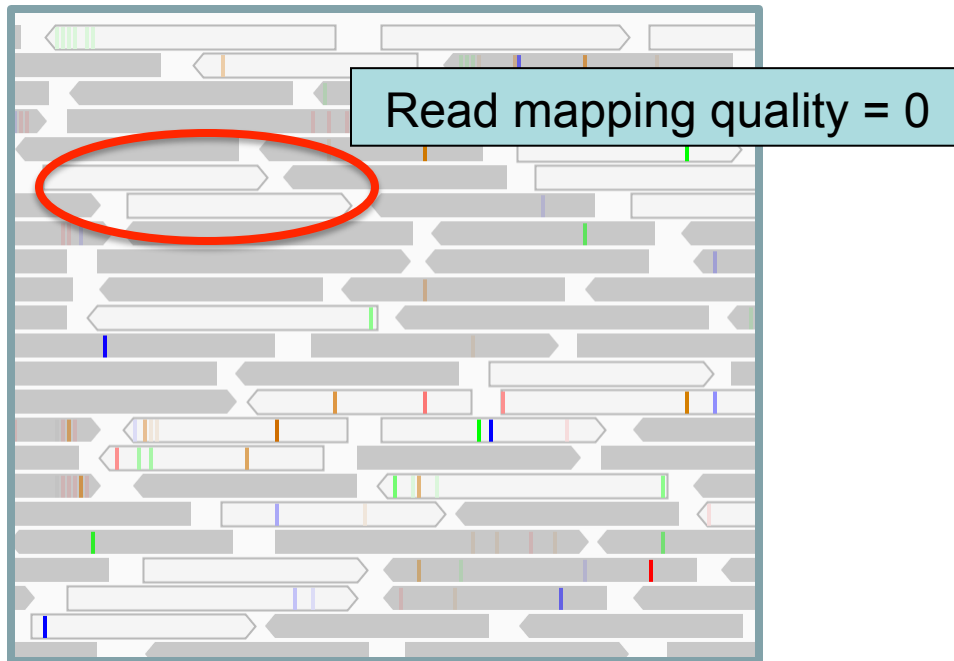


Viewing alignments

Zoom in to see more detail

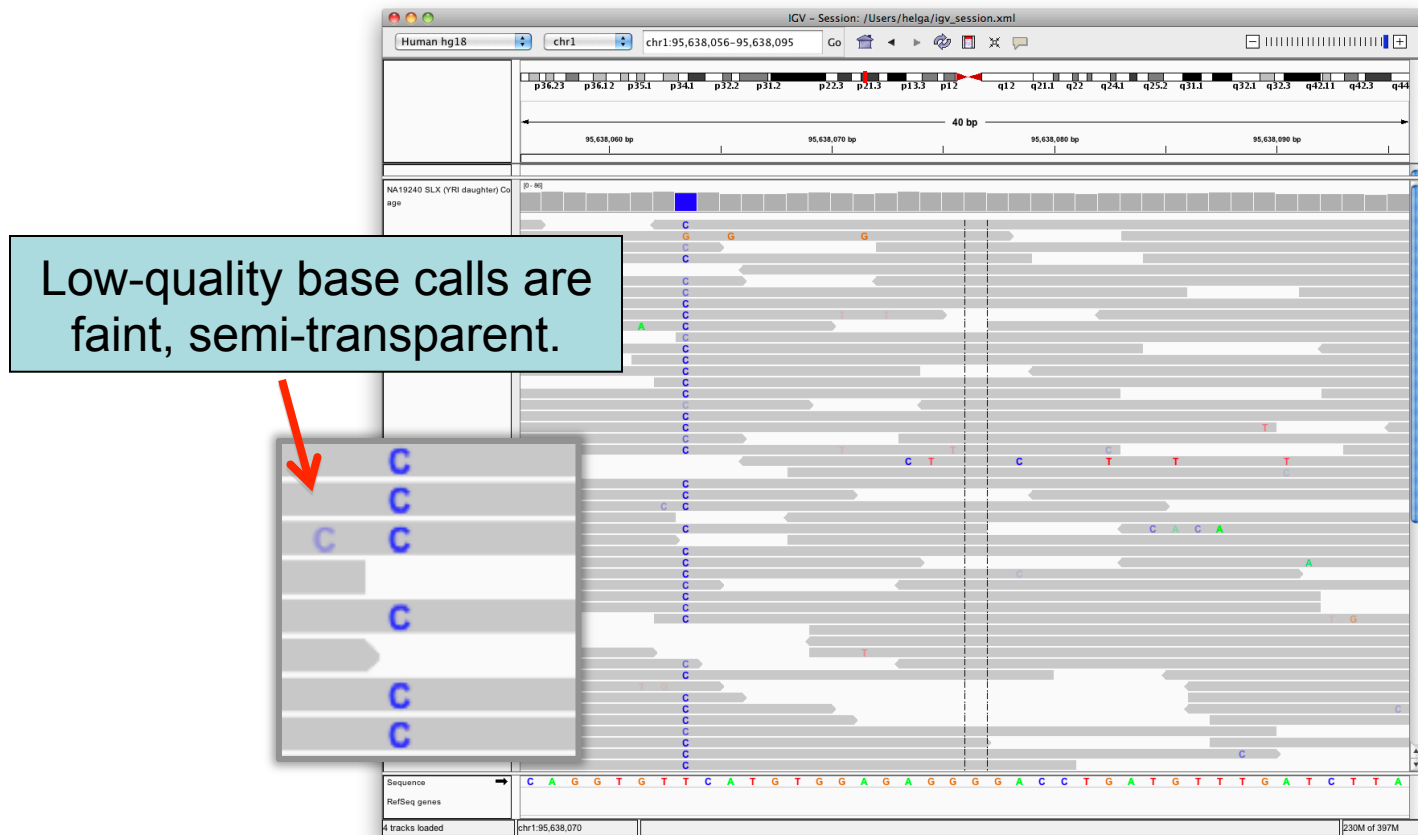


Viewing alignments



Viewing alignments

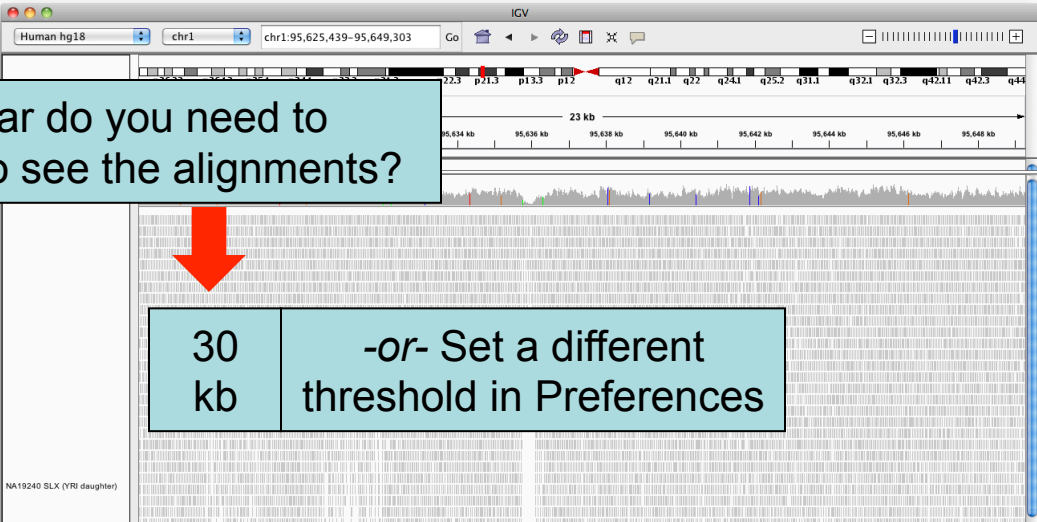
Zoom in to see more detail



Viewing alignments

Zoom in to view alignments

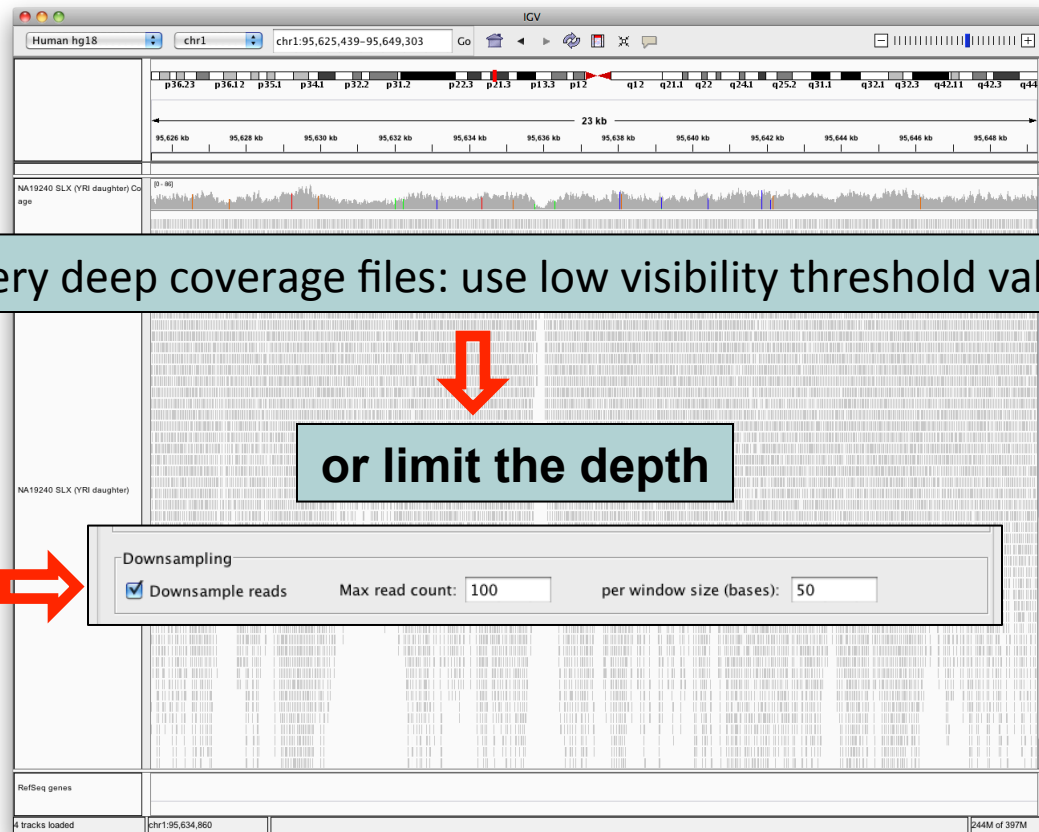
How far do you need to zoom in to see the alignments?



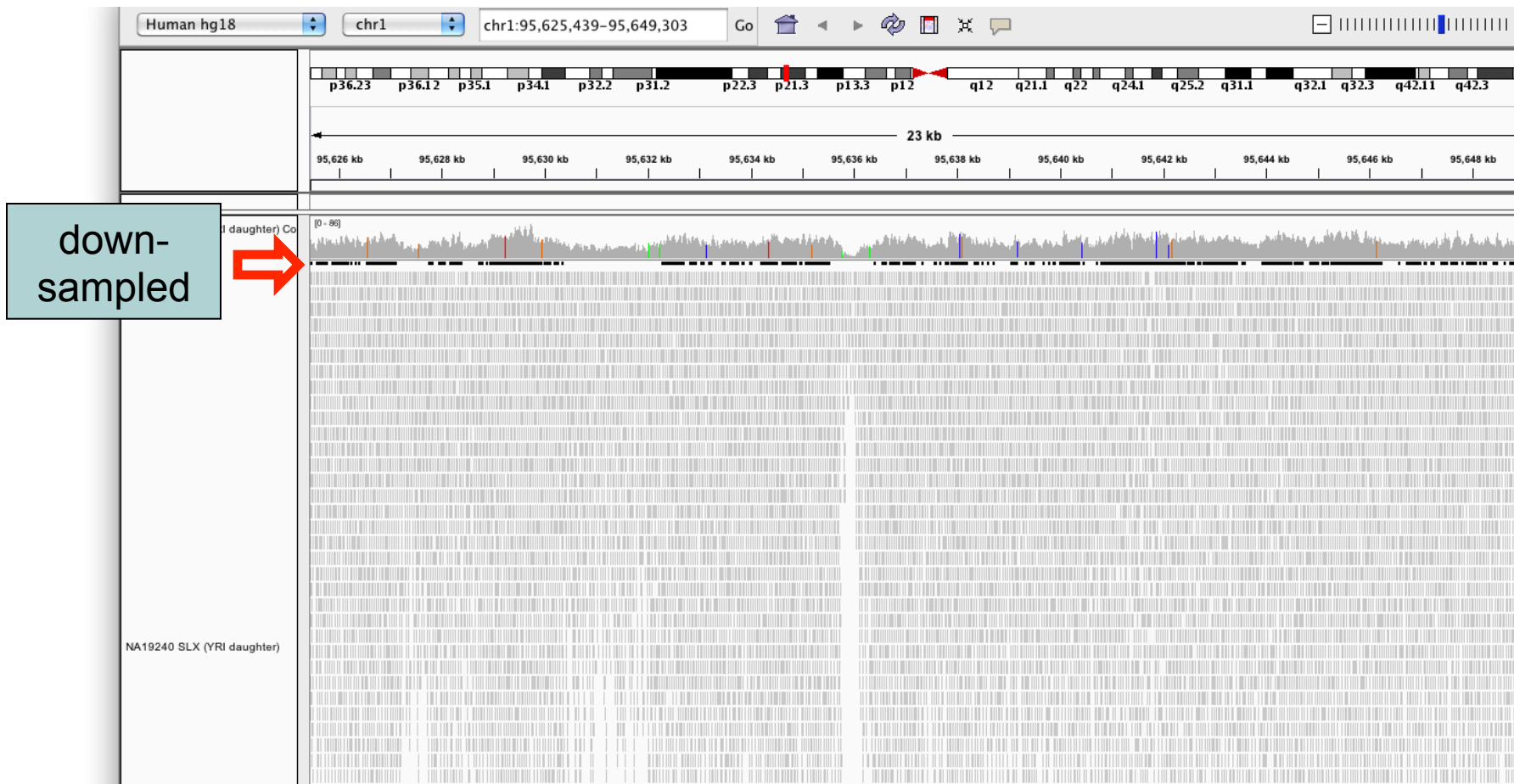
30 kb	-or- Set a different threshold in Preferences
----------	--

- Higher value ➔ requires more memory
- Low coverage files ➔ ok to use higher value
- Very deep coverage files ➔ use lower value

Viewing alignments



Viewing alignments



Viewing SNPs: Exercise

Hands-on

- Load alignments from whole genome sequencing
- View sites where SNPs were called
- Sort and color to highlight patterns

Data for today's exercises:

www.igv.org/workshops/ASHG2016/Exercises.zip

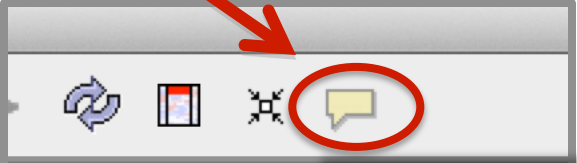
Instructions for downloading and launching IGV:

www.igv.org/workshops/ASHG2016/DownloadAndLaunch.pdf

Viewing SNPs: Exercise

To modify the info popup behavior:
Click yellow balloon icon in toolbar

For a larger data panel:
Click & drag the window divider



A screenshot of a genomic viewer toolbar. A red arrow points to a yellow speech bubble icon, which is circled in red. Other icons include a refresh symbol, a window icon, and a crosshair icon.

Sample = NA12878
Read group = ERR001757

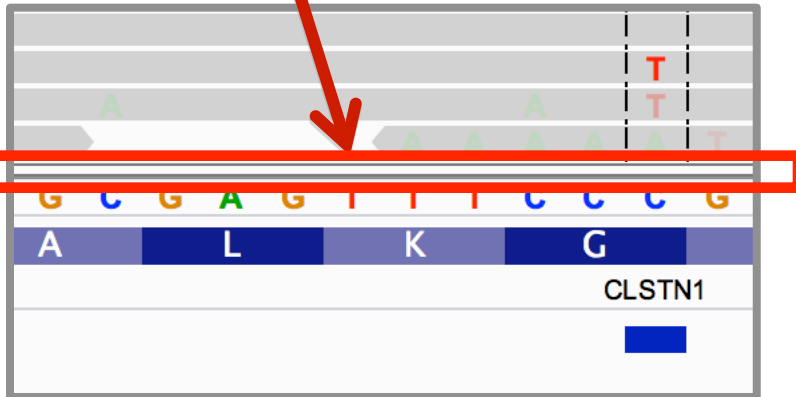
Read name = IL10_385:6:180:155:338
Location = chr1:9,718,629
Alignment start = 9,718,628 (-)
Cigar = 36M
Mapped = yes
Mapping quality = 70

Base = A
Base phred quality = 6

Pair start = chr1:9718480 (+)
Pair is mapped = yes
Insert size = -184
Pair orientation = F2R1

First in pair

H0 = 0
H1 = 1
MF = 18
RG = ERR001757
NM = 1
UQ = 4
AS = 25



Viewing SNPs: www.igv.org/workshops/ASHG2016/ExInstr.pdf

1 Initialize exercise: Click **File > Open Session**. Go to the folder where you downloaded the workshop data and select **snps / snp_session.xml**.

2 Go to first putative SNP locus: Type in the search box and click **Go**.

3

- Click & drag on alignments to put the column of red T's between the centerlines.
- Right-click on alignments, and select **Sort alignments by > base**.



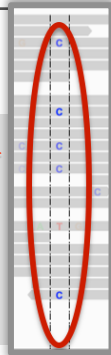
4 Does this look like a true SNP? For more info, mouse over the red & blue bar in the coverage track.



5 Go to next putative SNP locus: Type in the search box and click **Go**.

6

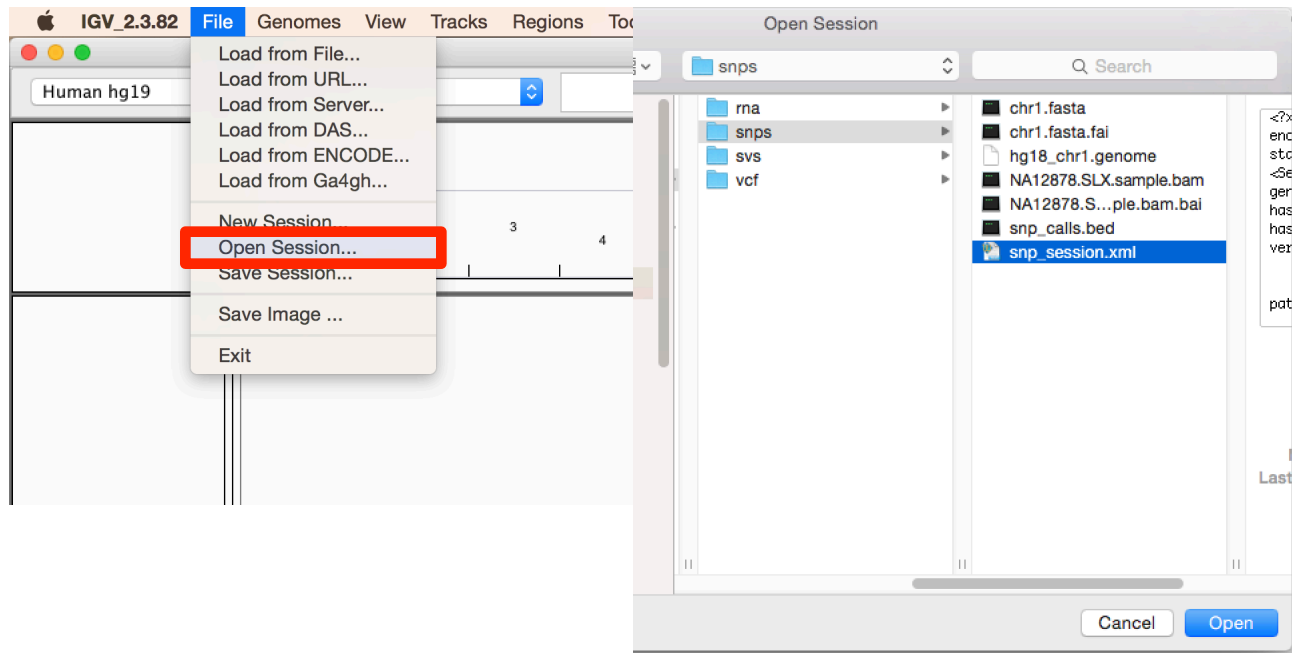
- Click & drag on alignments to position the 5 blue C's between the centerlines.
- Right-click on alignments, and deselect **Shade base by > quality**.
 - + select **Color alignments by > read strand**.
 - + select **Sort alignments by > read strand**.



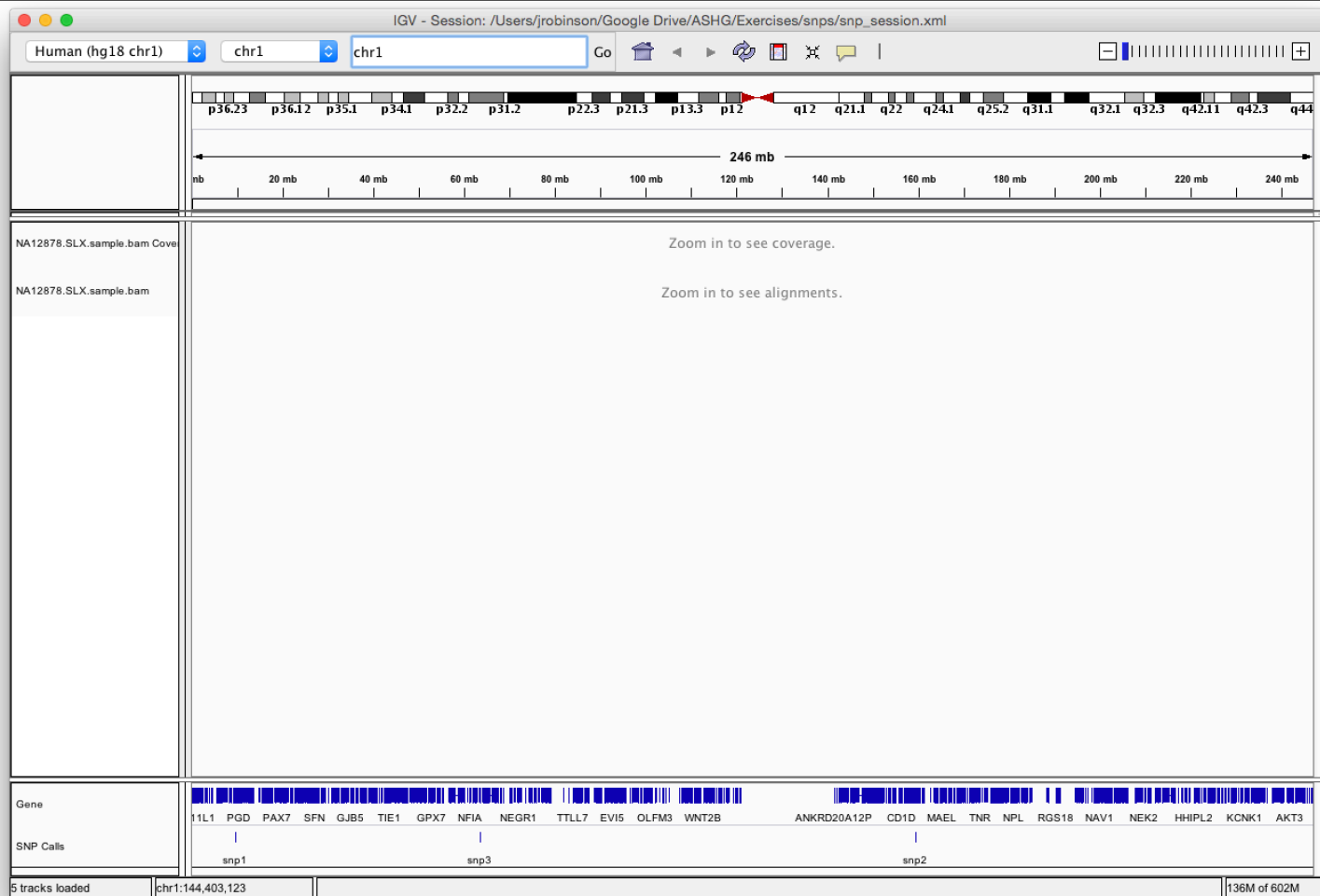
7 Does this look like a true SNP? Note this was not from a strand preserving library.

Viewing SNPs: Initialize the exercise

- Click **File > Open Session**
- Navigate to the folder where you downloaded the workshop data – and select **snps / snp_session.xml**



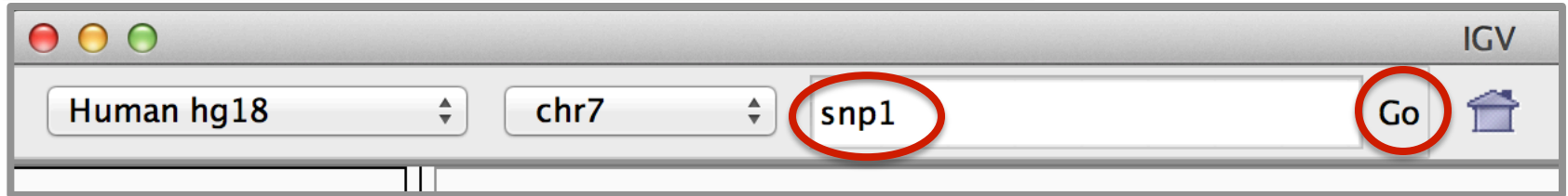
Viewing SNPs



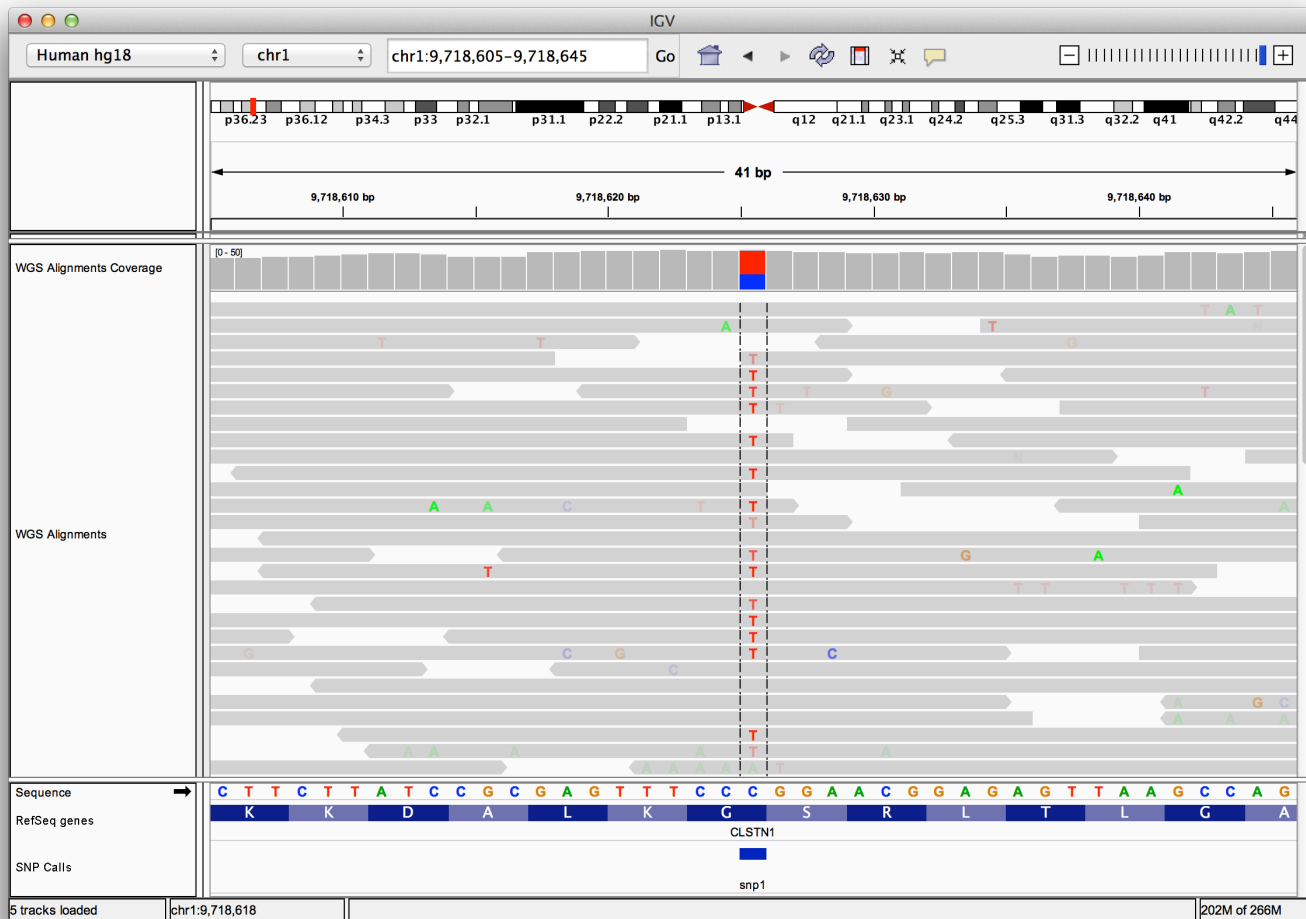
Viewing SNPs

- Navigate to first putative SNP locus

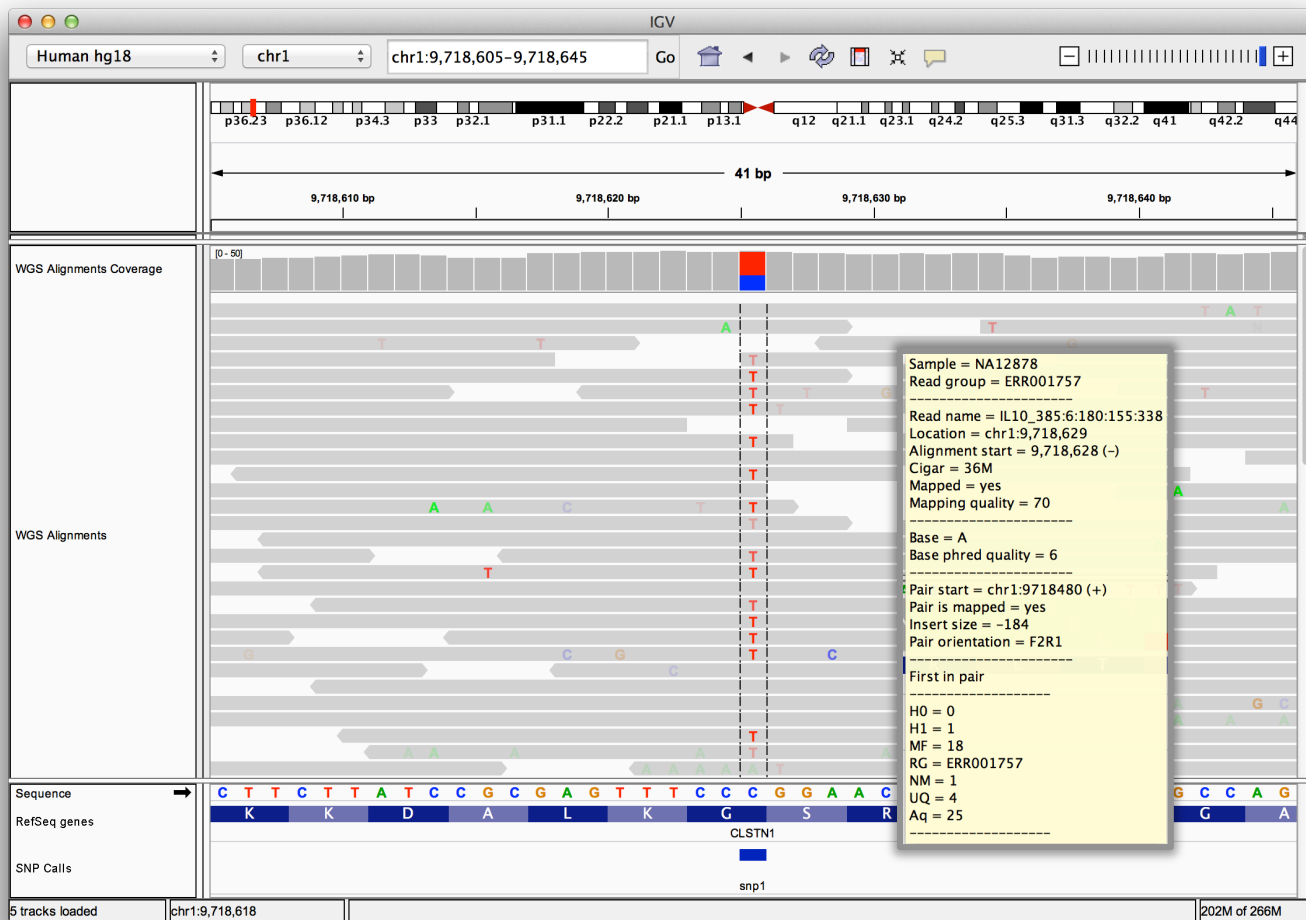
Type “snp1” in the **Search Box** and click **Go**



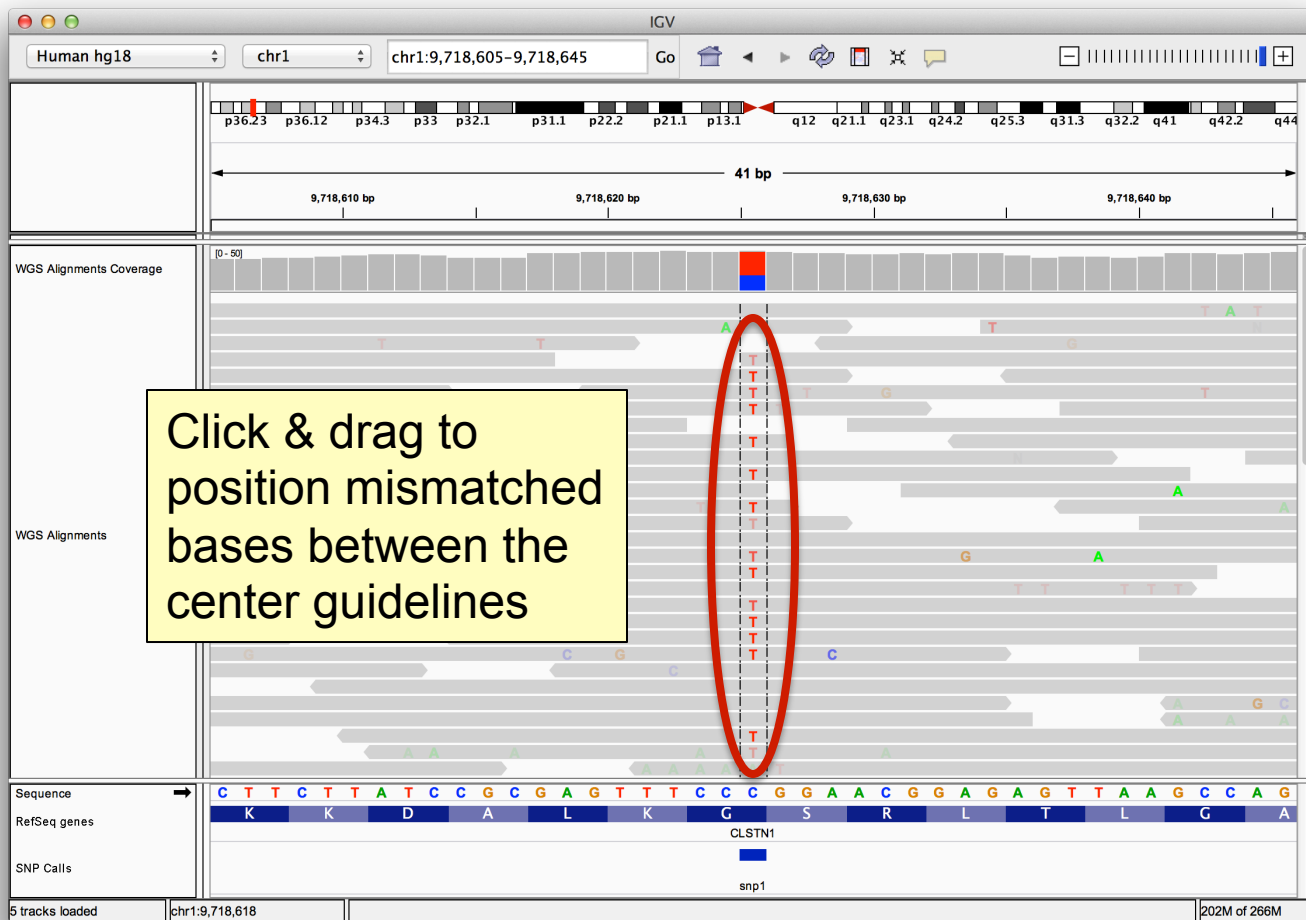
Viewing SNPs



Viewing SNPs



Viewing SNPs



Viewing SNPs

IGV

Human hg18 chr1 chr1:9,718,605-9,718,645 Go

WGS Alignments

- Rename Track...
- Copy read details to clipboard
- Group alignments by
- Sort alignments by
- Color alignments by
- ✓ Shade base by quality
- ✓ Show mismatched bases
- Show all bases

- start location
- read strand
- first-of-pair strand
- base
- mapping quality
- sample
- read group
- insert size
- chromosome of mate
- tag

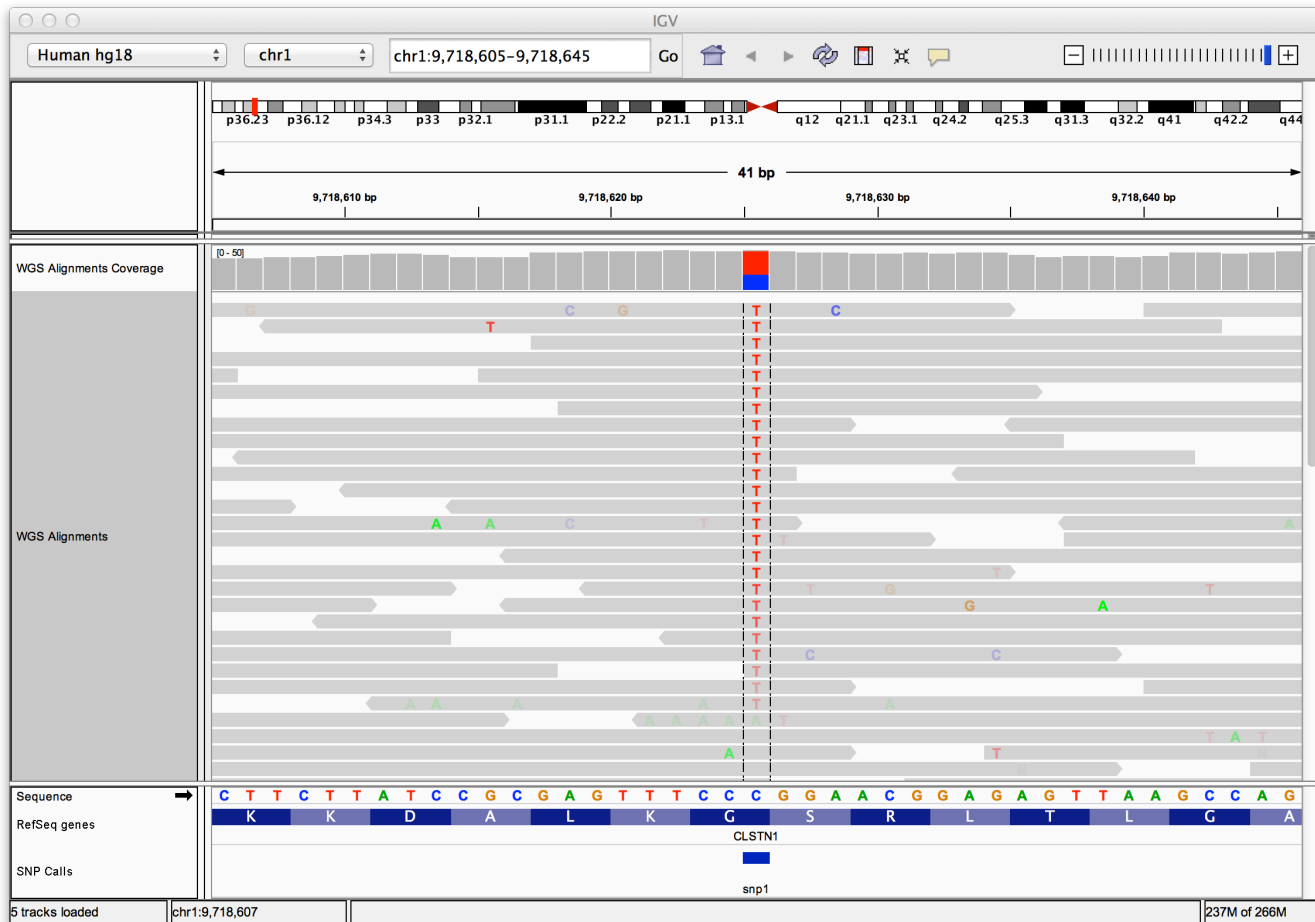
Right-click on alignments and select **Sort alignments by > base**

On Mac:
Right-click = Control-click

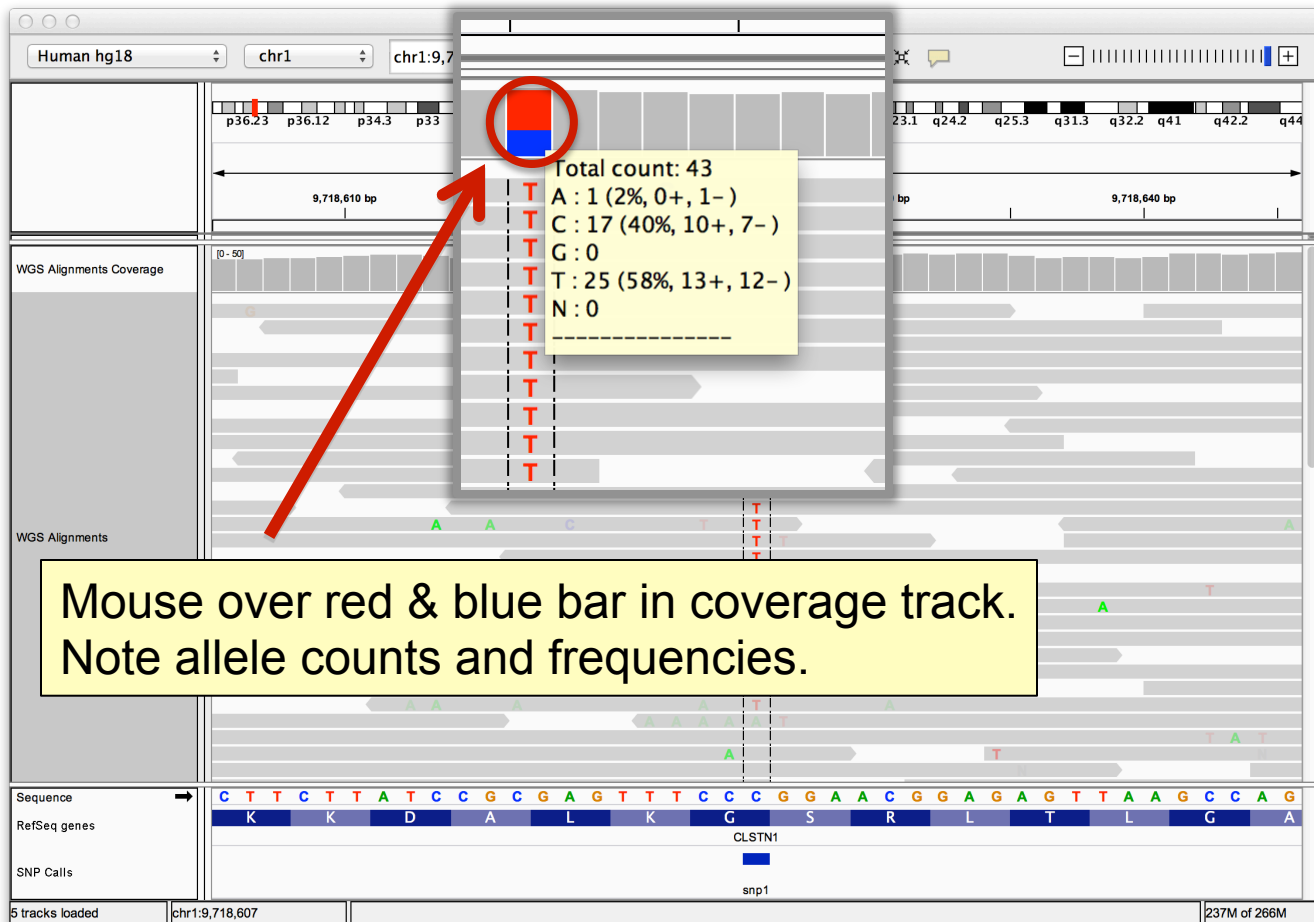
Sequence
RefSeq genes
SNP Calls

5 tracks loaded chr1:9,718,618 202M of 266M

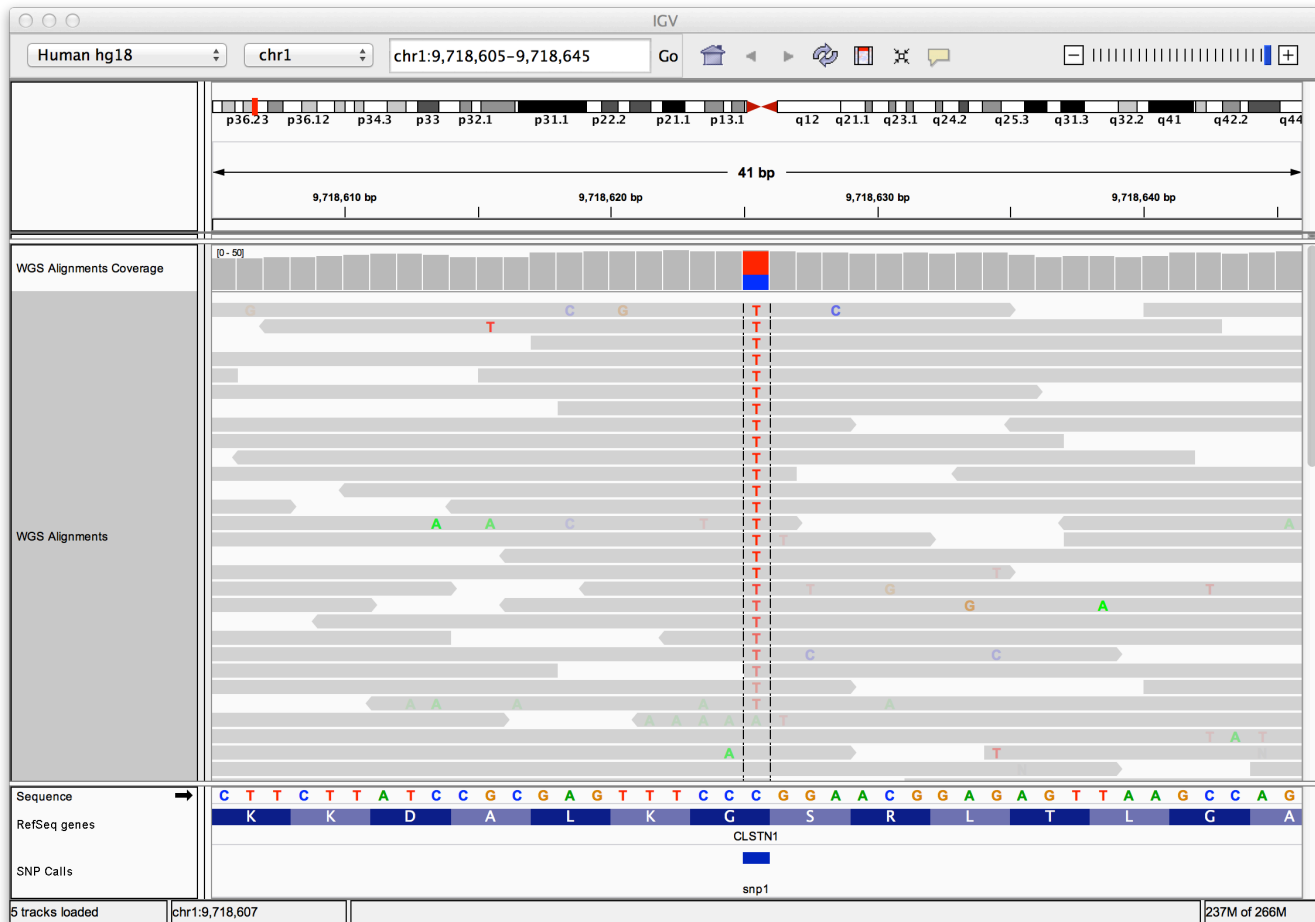
Viewing SNPs



Viewing SNPs



Viewing SNPs



Viewing SNPs

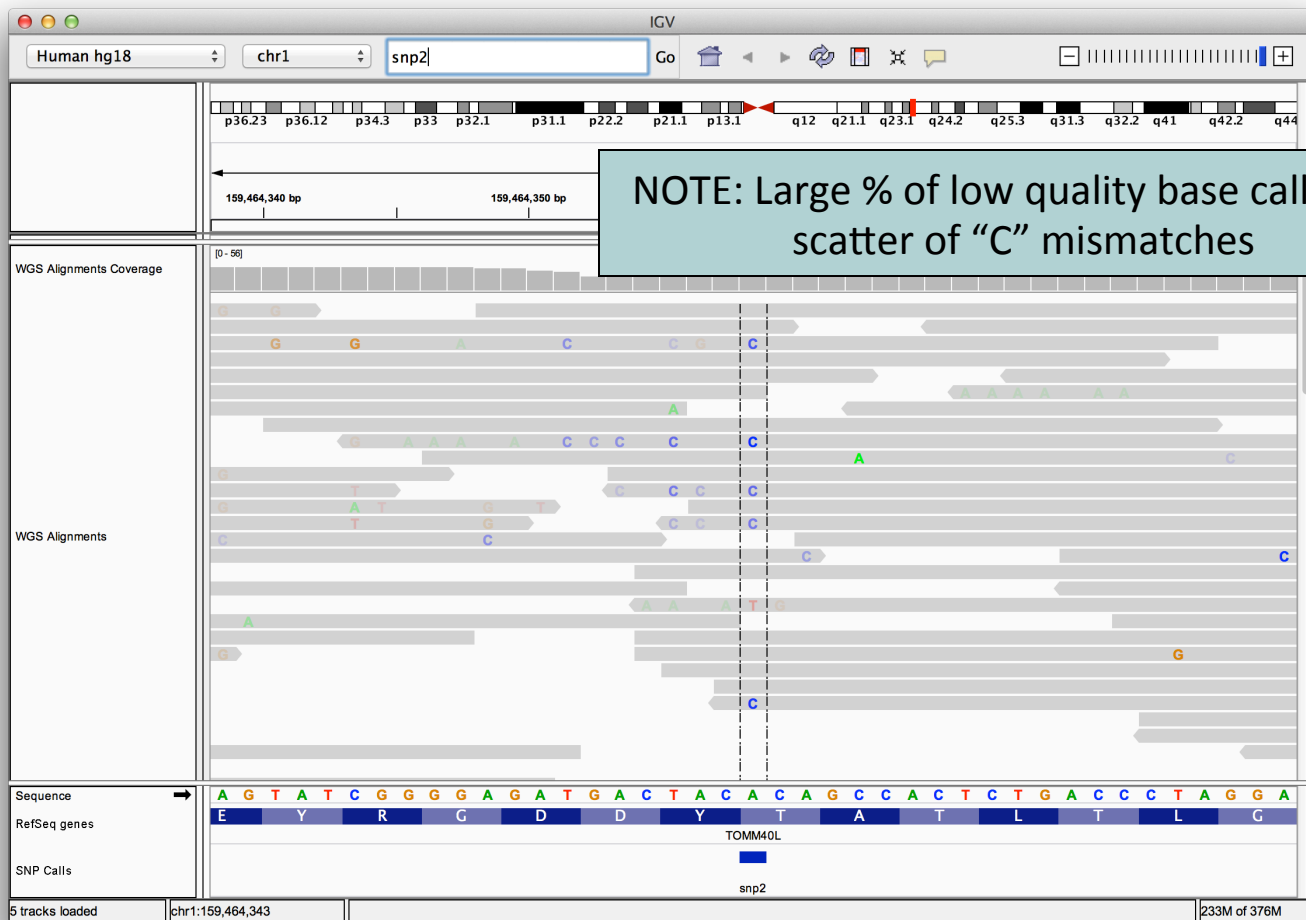
Type "snp2" in the **Search Box** and click **Go**

The screenshot displays the IGV interface with the following components:

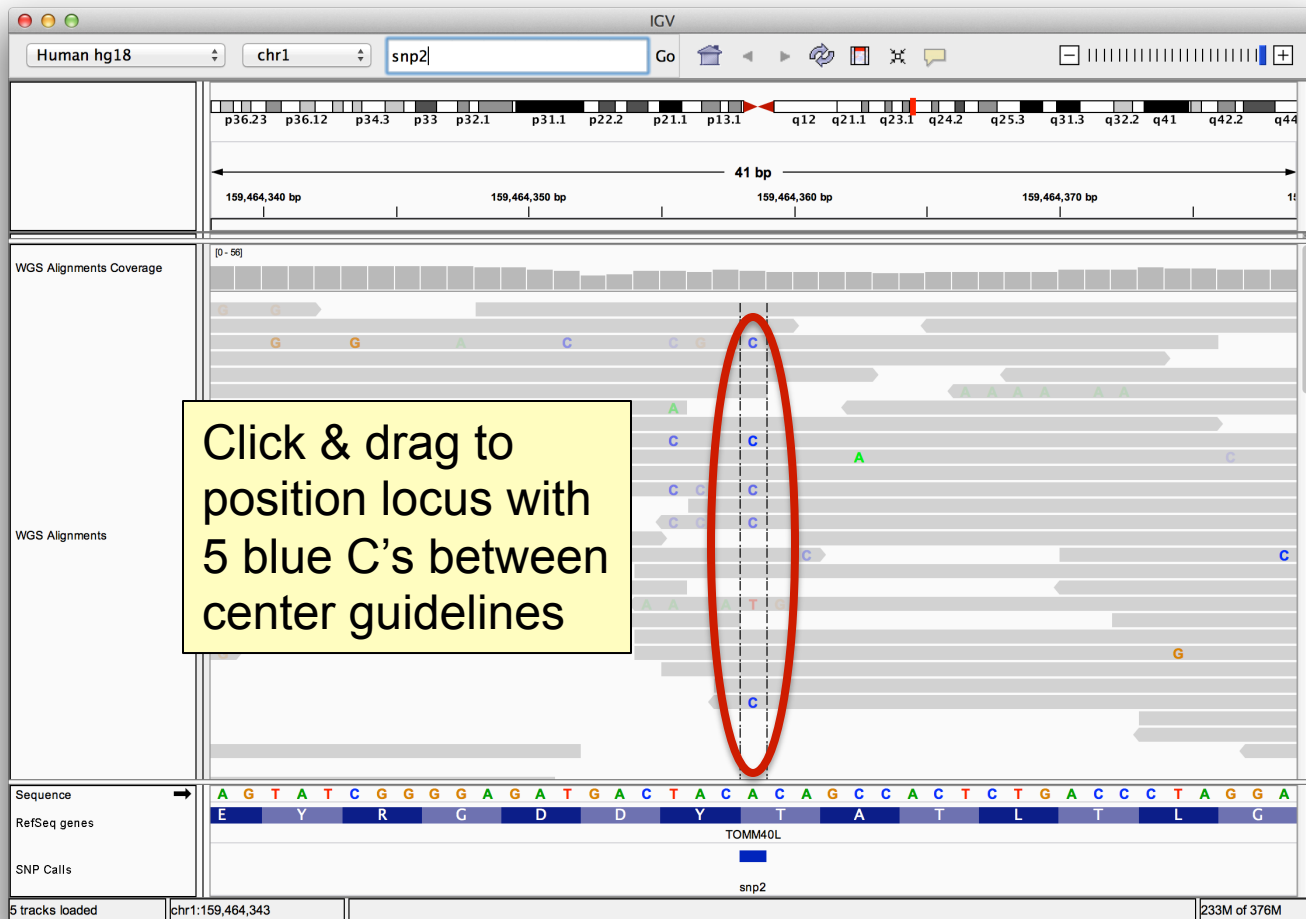
- Search Bar:** Contains the text "snp2" and a "Go" button.
- Genome Browser:** Shows a genomic track with a red arrow pointing to a specific location on chromosome 1 (chr1).
- WGS Alignments Coverage:** A bar chart showing the coverage of the WGS alignments.
- WGS Alignments:** A track showing individual sequencing reads with colored letters (A, G, C, T) indicating mismatches from the reference.
- Sequence:** A track showing the reference sequence (A, G, T, A, T, C, G, G, G, G, A, G, A, T, G, A, C, T, A, C, A, C, A, G, C, C, A, C, T, C, T, G, A, C, C, C, T, A, G, G, A).
- RefSeq genes:** A track showing the gene model for the region, including the gene TOMM40L.
- SNP Calls:** A track showing the specific SNP position (snp2) and its call (A).

5 tracks loaded | chr1:159,464,343 | 233M of 376M

Viewing SNPs



Viewing SNPs



Viewing SNPs

Human hg18 chr1 snp2 Go

WGS Alignments Coverage

WGS Alignments

Sequence RefSeq genes SNP Calls

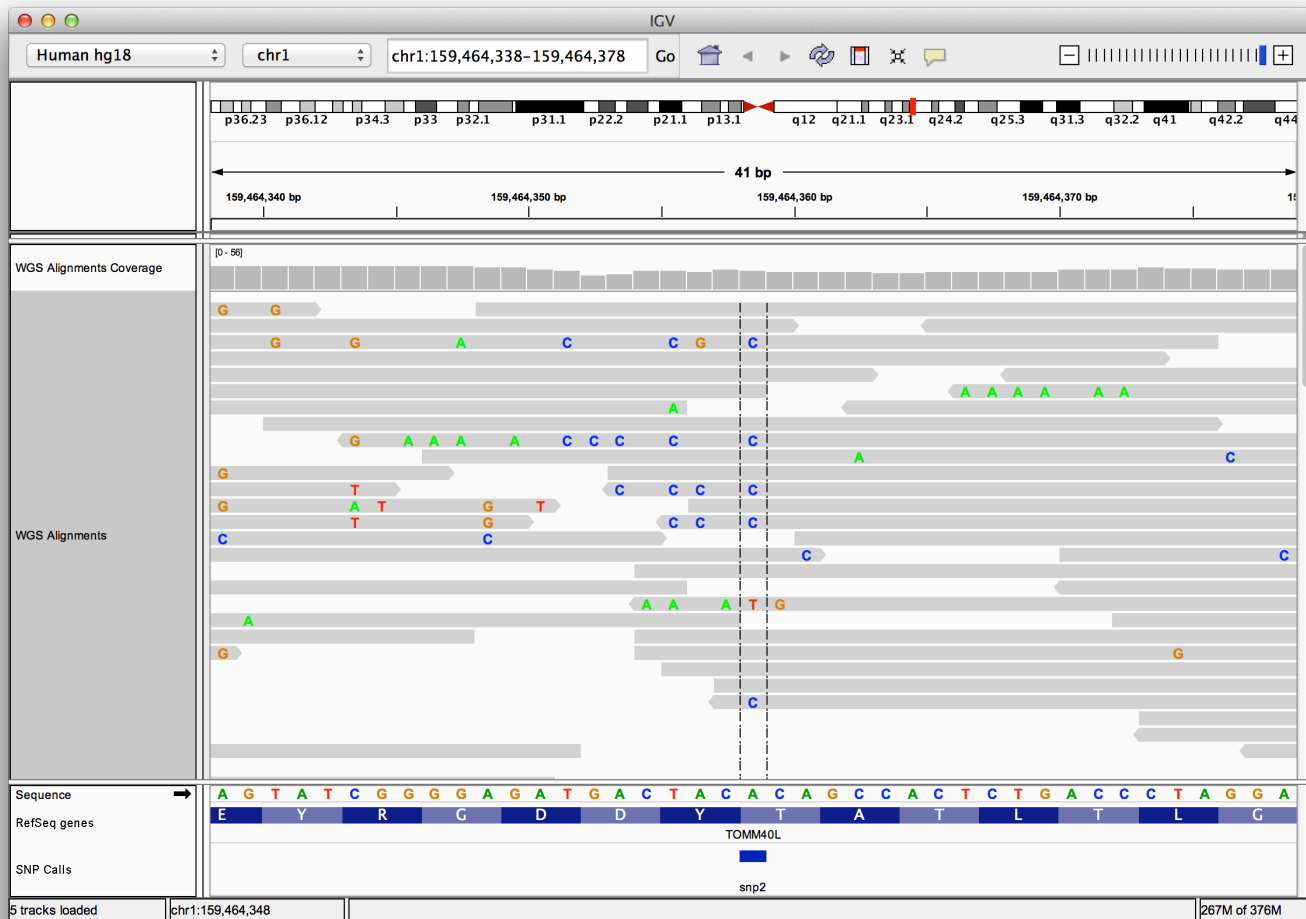
5 tracks loaded chr1:159,464,343 233M of 376M

WGS Alignments

- Rename Track...
- Copy read details to clipboard
- Group alignments by ▶
- Sort alignments by ▶
- Color alignments by ▶
- ☒ Shade base by quality
- ☒ Show mismatched bases
- Show all bases
- View as pairs
- Go to mate
- View mate region in split screen
- Set insert size options ...
- Re-pack alignments

Right-click on alignments and deselect **Shade base by quality**

Viewing SNPs



Viewing SNPs

The screenshot displays a genomic browser interface with the following components:

- Top Bar:** Human hg18, chr1, snp2
- WGS Alignments Coverage:** A track showing coverage with a red arrow pointing to the context menu.
- WGS Alignments:** A track showing individual sequencing reads with bases colored by quality.
- Sequence:** A track showing the reference sequence: A G T A T C G G G G A G A T G A C T A C A C A G C C A C T C T G A C C C T A G G A.
- RefSeq genes:** A track showing the gene TOMM40L.
- SNP Calls:** A track showing the SNP call for snp2.

A context menu is open over the WGS Alignments track, titled "WGS Alignments". The menu options are:

- Rename Track...
- Copy read details to clipboard
- Group alignments by
- Sort alignments by** (highlighted with a red arrow)
- Color alignments by
- ✓ Shade base by quality
- ✓ Show mismatched bases
- Show all bases
- View as pairs
- Go to mate
- View mate region in split screen

A secondary menu is open for "Sort alignments by", listing the following options:

- start location
- read strand** (highlighted with a red arrow)
- first-of-pair strand
- base
- mapping quality
- sample
- read group
- insert size
- chromosome of mate
- tag

Right-click on alignments and select Sort alignments by > read strand

5 tracks loaded | chr1:159,464,343 | 233M of 376M

Viewing SNPs

The screenshot displays a genomic browser interface with the following components:

- Top Bar:** Human hg18, chr1, and a search box containing 'snp2'.
- WGS Alignments Coverage:** A track showing read coverage with a red arrow pointing to the 'Color alignments by' option in the context menu.
- WGS Alignments:** A track showing individual sequencing reads with bases colored by strand.
- Sequence:** A track showing the reference sequence: A G T A T C G G G G A G A T G A C T A C A C G C C A C T C T G A C C C T A G G A.
- RefSeq genes:** A track showing the gene structure for TOMM40L.
- SNP Calls:** A track showing the SNP call for snp2.

A context menu is open over the WGS Alignments track, showing the following options:

- WGS Alignments
- Rename Track...
- Copy read details to clipboard
- Group alignments by
- Sort alignments by
- Color alignments by** (selected)
- Shade base by quality
- Show mismatched bases
- Show all bases
- View as pairs
- Go to mate
- View mate region in split screen
- Set insert size options ...

The 'Color alignments by' submenu is open, showing the following options:

- no color
- insert size
- pair orientation
- insert size and pair orientation
- read strand** (selected)
- first-of-pair strand
- read group
- sample
- tag
- bisulfite mode

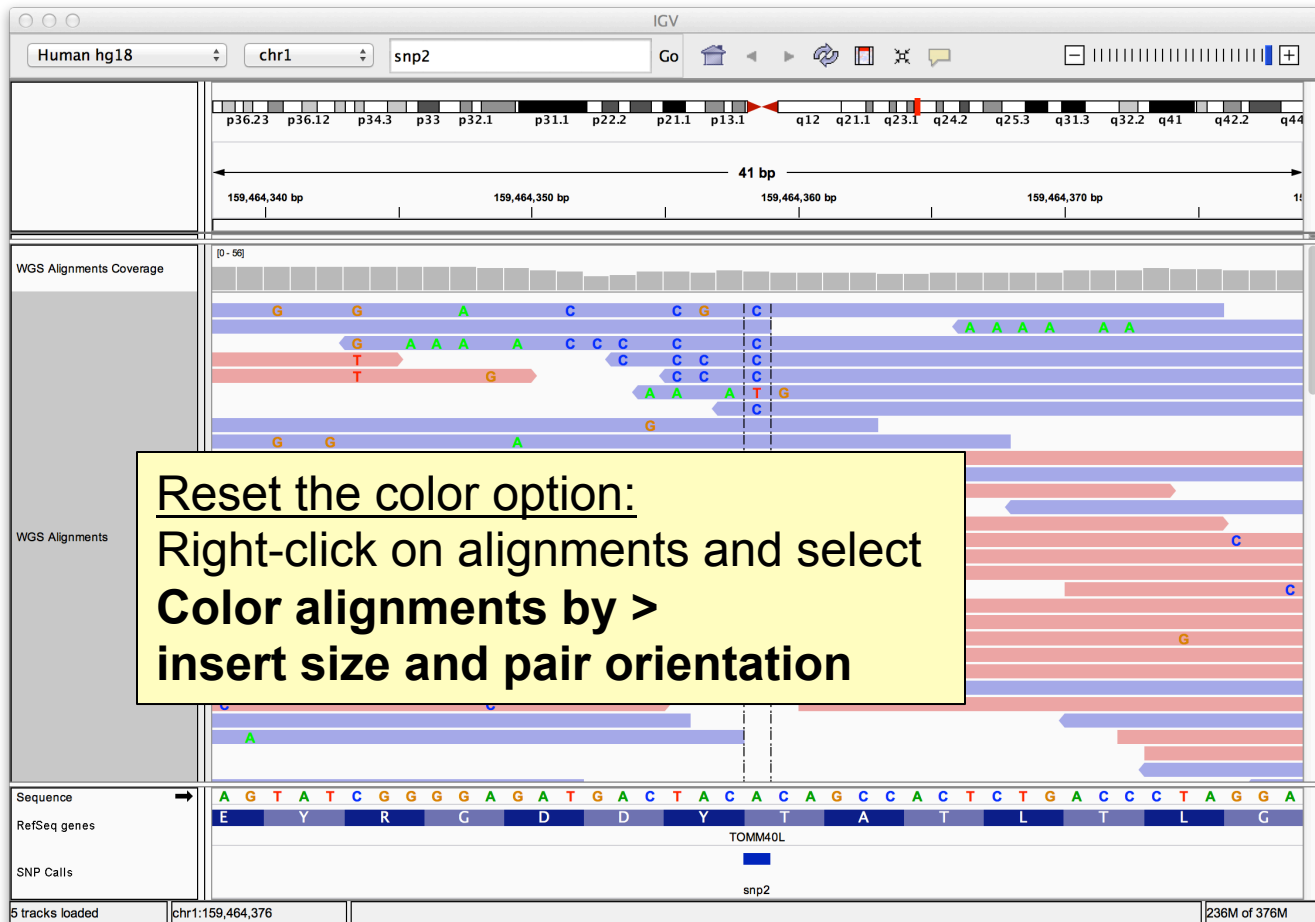
Right-click on alignments and select Color alignments by > read strand

5 tracks loaded | chr1:159,464,343 | 233M of 376M

Viewing SNPs

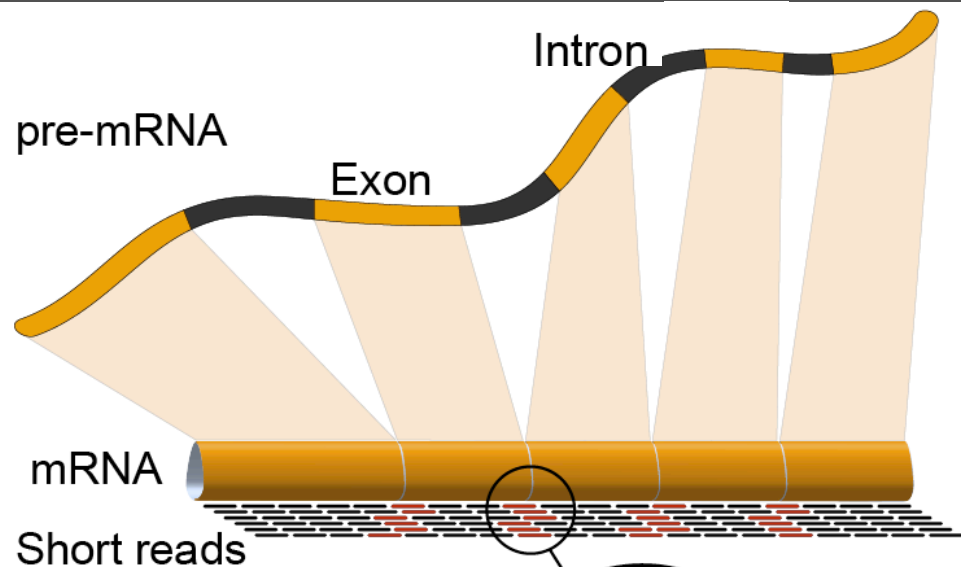


Viewing SNPs



Viewing RNA-seq Data

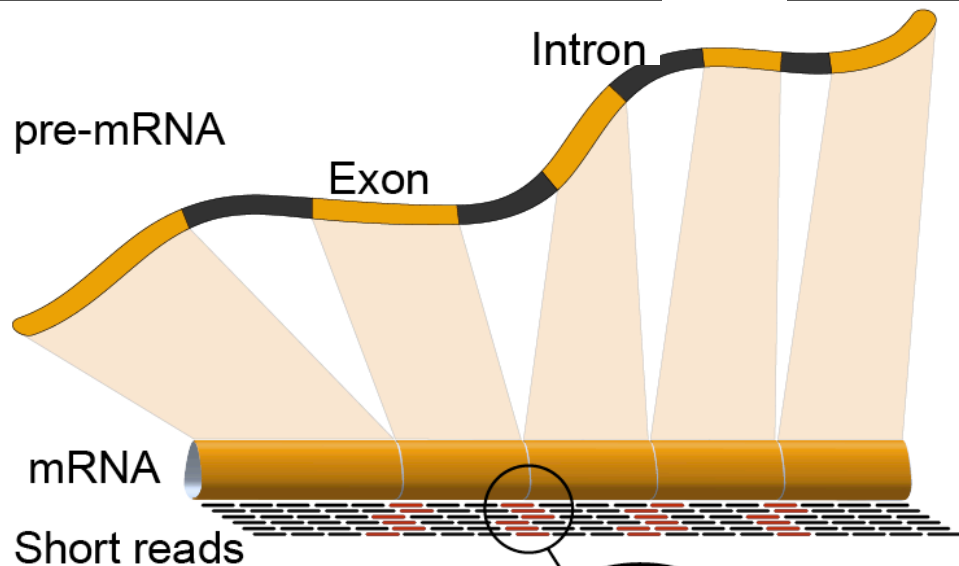
RNA-seq



Short read is split by
intron when aligning
to reference Genome

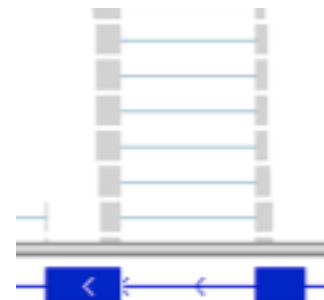
http://en.wikipedia.org/wiki/RNA-Seq#Gene_expression

RNA-seq



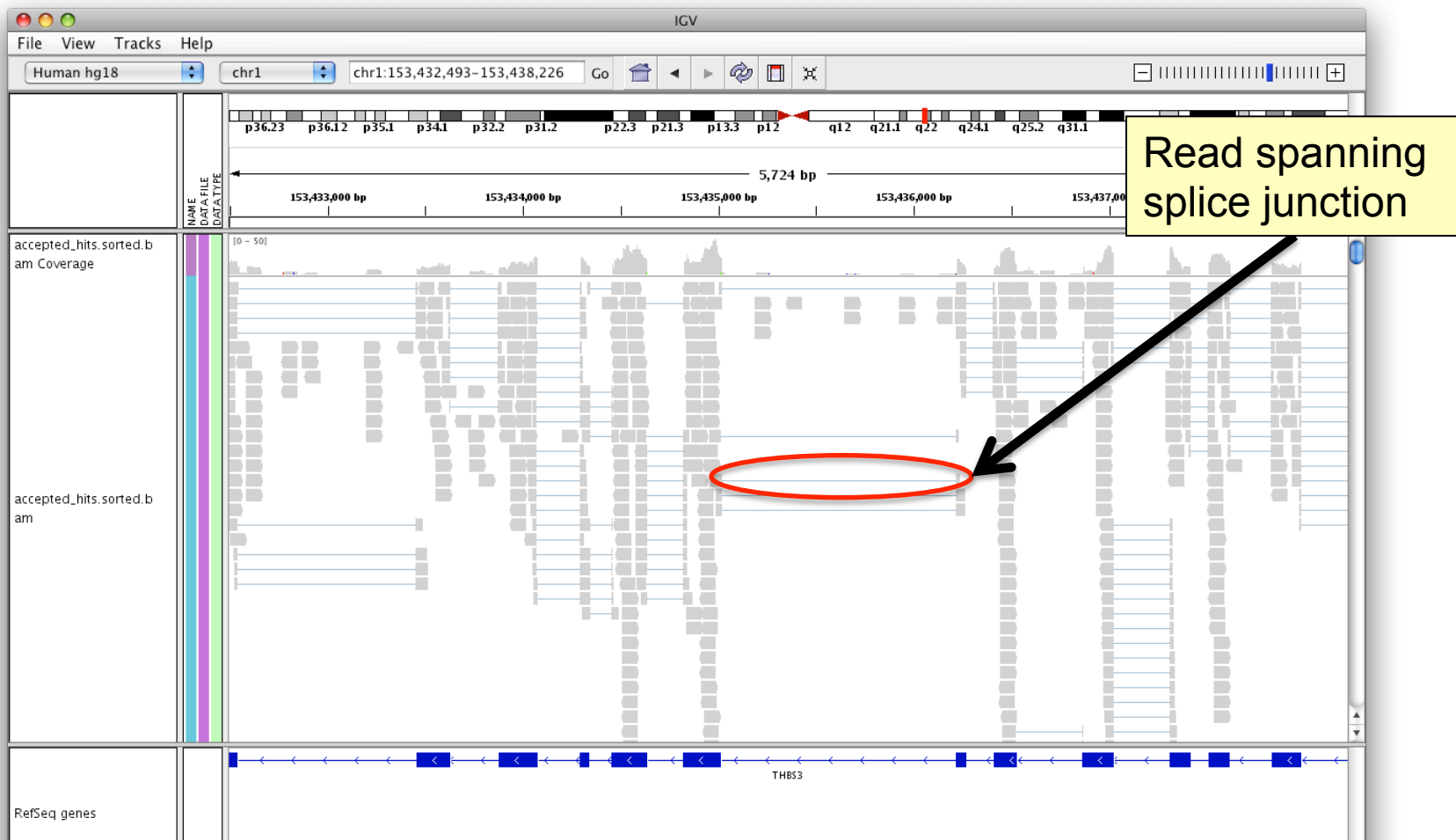
Short reads

Short read is split by
intron when aligning
to reference Genome



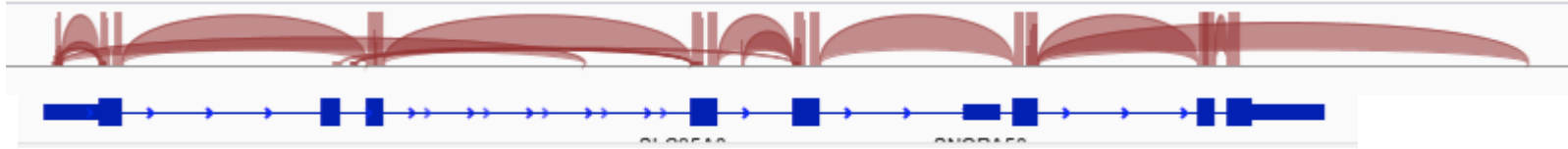
http://en.wikipedia.org/wiki/RNA-Seq#Gene_expression

RNA-seq alignments



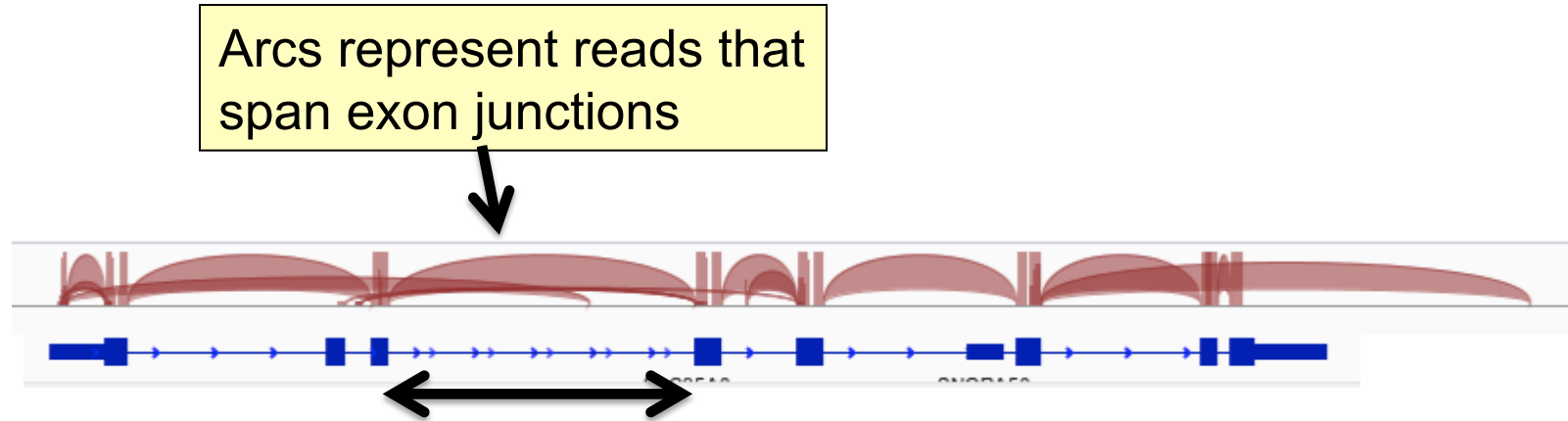
RNA-seq alignments

- Splice junction track



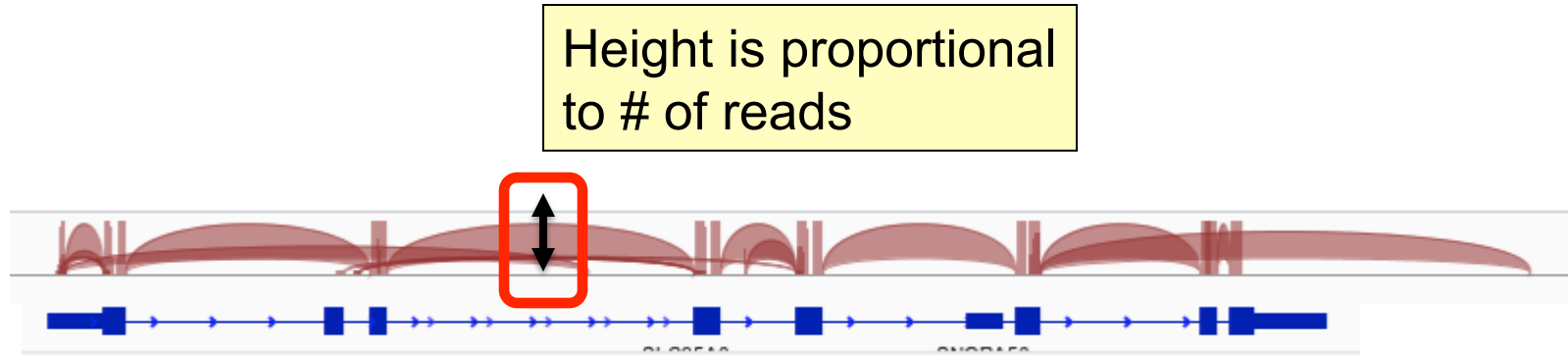
RNA-seq alignments

- Splice junction track



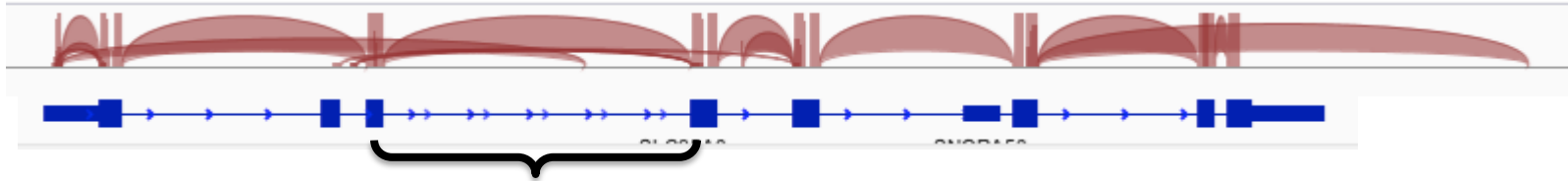
RNA-seq alignments

- Splice junction track



RNA-seq alignments

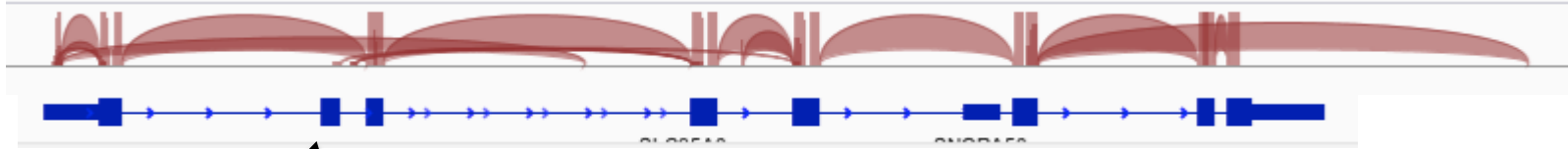
- Splice junction track



Many reads connecting
these exons

RNA-seq alignments

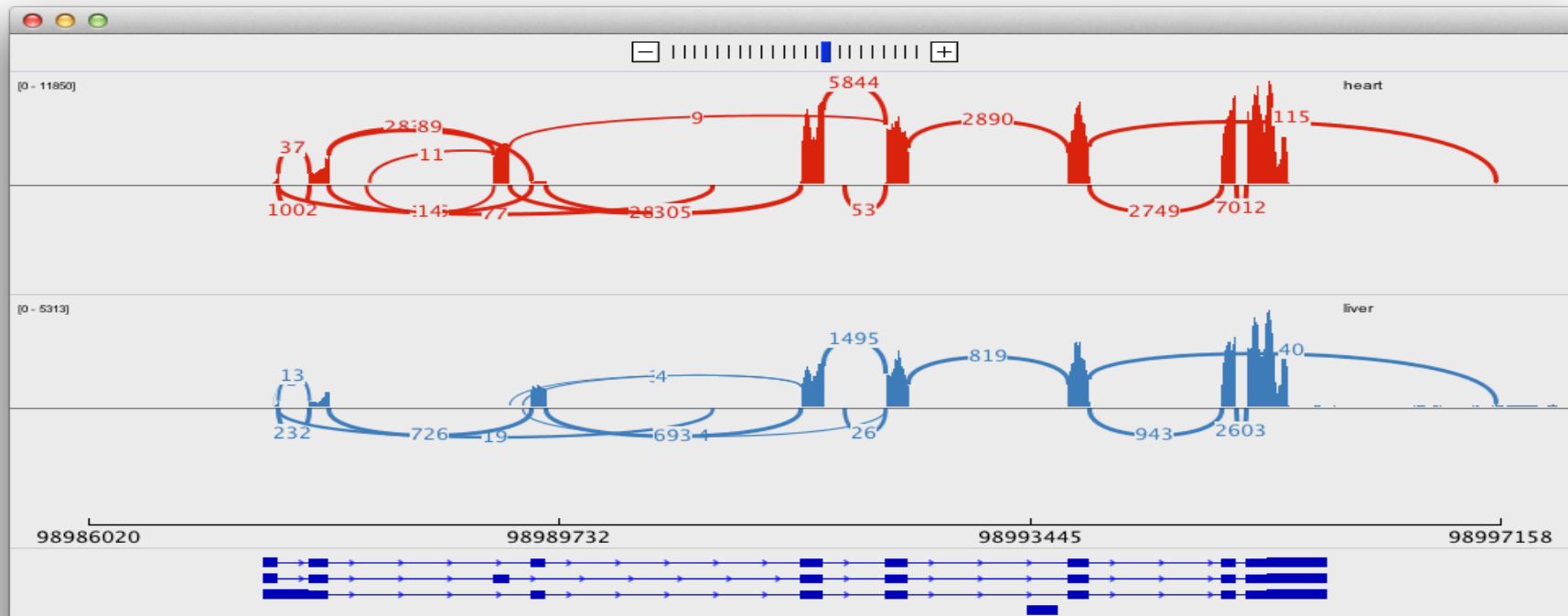
- Splice junction track



Relatively few
spanning to this exon

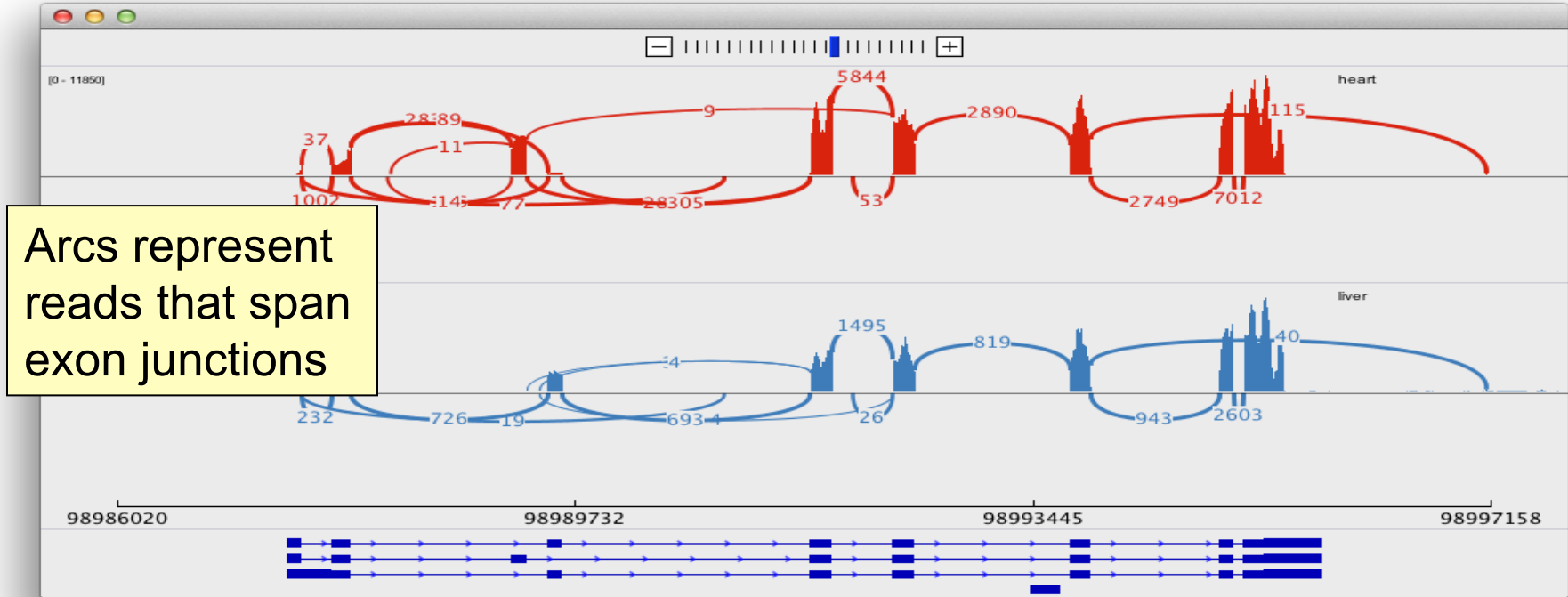
RNA-seq alignments

Sashimi plot



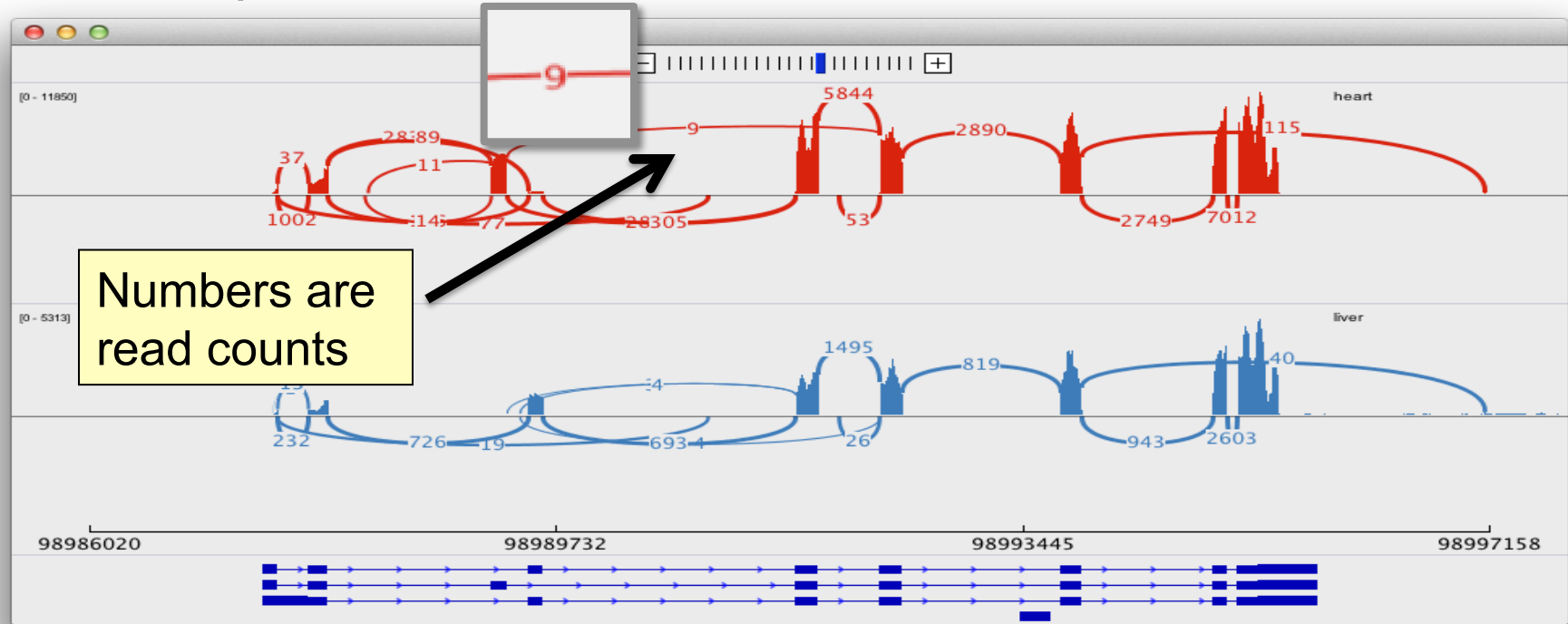
RNA-seq alignments

Sashimi plot

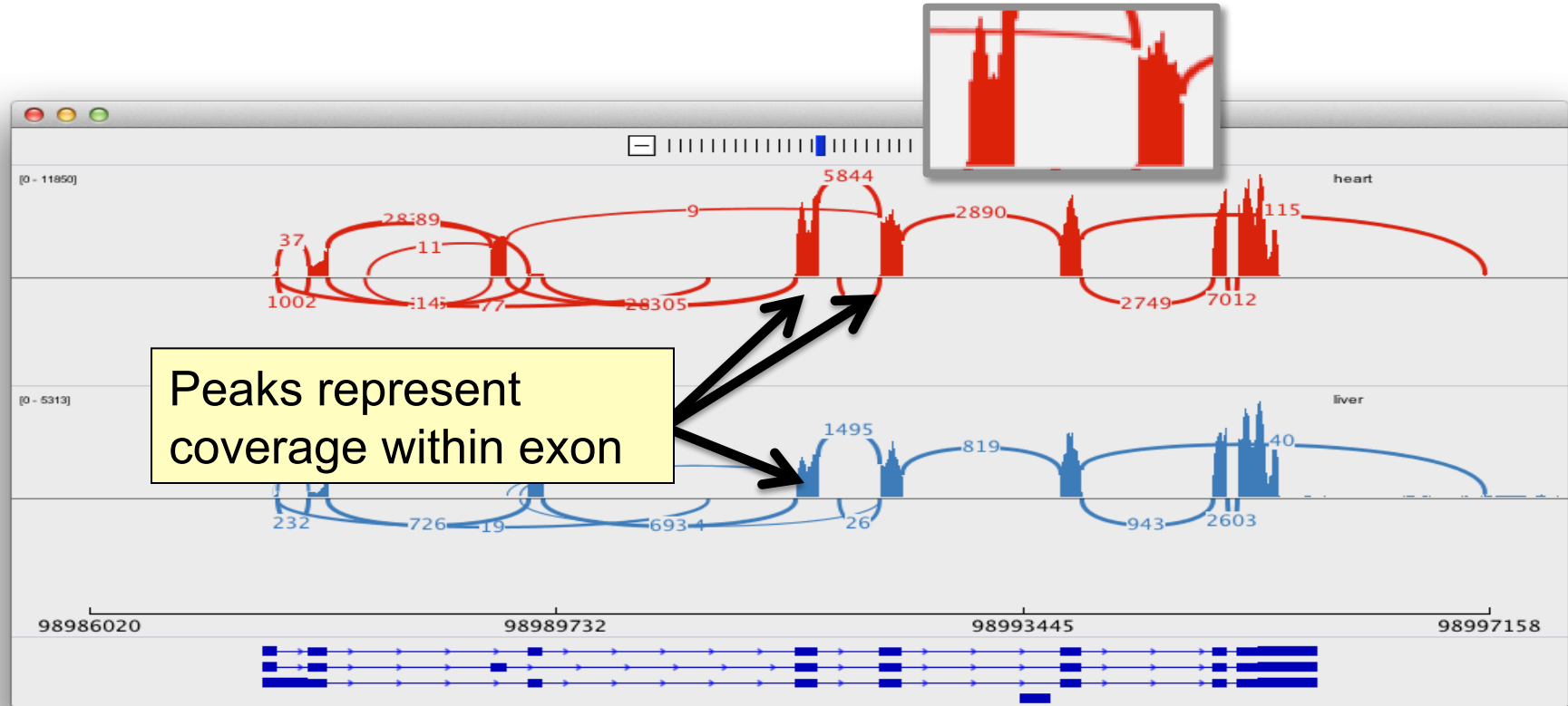


RNA-seq alignments

Sashimi plot



RNA-seq alignments



Sashimi plot

Viewing RNA splicing with Sashimi Plots

Reference:

Katz et al.

***Quantitative visualization of alternative exon expression
from RNA-seq data***

Bioinformatics (2015)

RNA-seq: Exercise

Hands-on

- Examine tissue-specific alternative splicing.
- Data: Illumina BodyMap 2.0

http://portals.broadinstitute.org/genome_bio/human_lincrnas/

RNA-seq: Exercise

Steps

1. Enable Splice Junction track in IGV Preferences
2. Load BodyMap alignments
3. View splice junction track
4. View Sashimi plot

RNA-seq: www.igv.org/workshops/ASHG2016/ExInstr.pdf

1 Initialize exercise:

- Click **View > Preferences**, select the **Alignments** tab, check **Splice Junction Track**, and click **OK**.
- Click **File > Open Session**. Go to the folder where you downloaded the workshop data and select **rna / rna_session.xml**.

2

Observe the two samples: Heart and Liver.

Each sample has a coverage track, splice junction track, and the RNA-seq reads.

3

- Right click on the Genes track, and select **Squished** to display the isoforms.
- Zoom in to get a closer view of the first 4 exons of the gene.

4

Observe evidence of alternative splicing:

Note differences in the heart vs. liver samples and how they compare to the gene isoforms.

5

- Right click on the data (reads or junction track) and select **Sashimi Plot**.
- In the track selection window that pops up, make sure both tracks are selected.

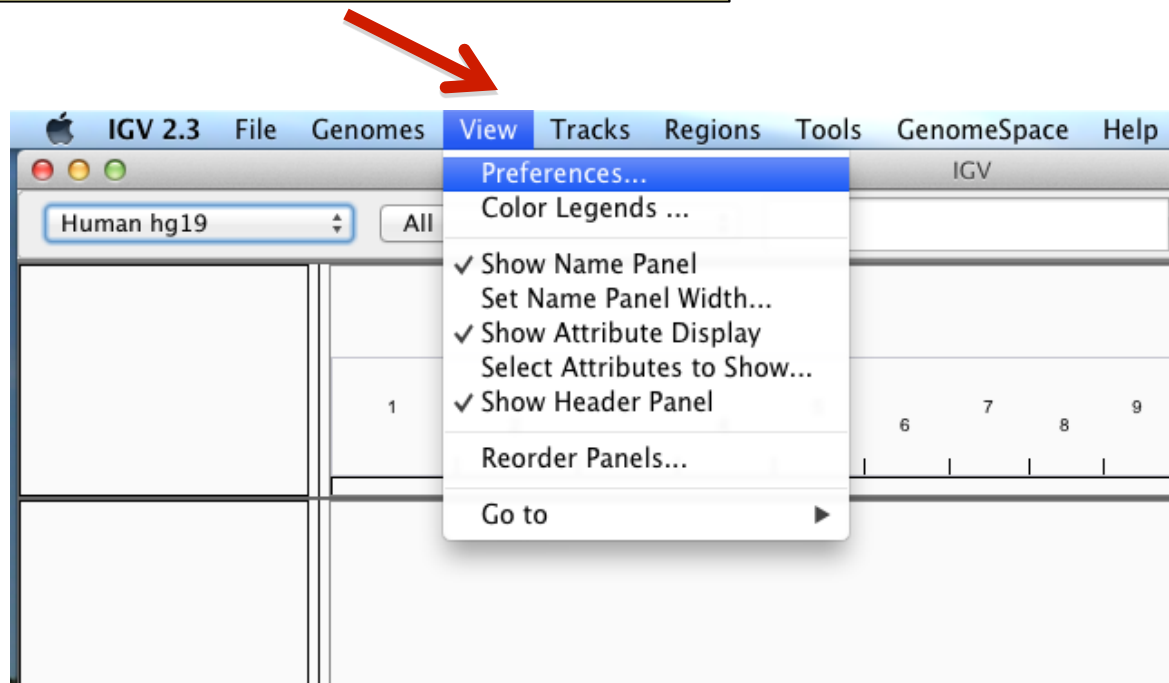
6

Sashimi Plot:

The Sashimi Plot pops up in a separate window.

Note it is interactive: Zoom in and out. Pan. Right-click to set options.

Select **View > Preferences...**



Click **Alignments** tab

The screenshot shows the IGV (Integrative Genomics Viewer) interface. The 'Alignments' tab is selected and highlighted with a red box and an arrow. A yellow callout box points to this tab with the text 'Click Alignments tab'. The interface displays a genomic track for chromosome 12, specifically the region around 98,986,192. The track shows a single read alignment. The 'Alignments' tab is active, and the 'On initial load show' section is checked for 'Alignment Track', 'Coverage Track', and 'Splice Junction Track'. The 'Alignment Track Options' section includes settings for visibility range threshold (30 kb), downsampling reads (max read count: 100, per window size: 50), shading mismatched bases by quality (5 to 20), flagging insertions larger than 1 base, mapping quality threshold (0), and hidden SAM tags (MD,SA). The 'Coverage Track Options' section includes a coverage allele-fraction threshold (0.2) and a checked option for 'Quality weight allele fraction'. The 'Splice Junction Track Options' section includes a checked option for 'Show flanking regions' with a minimum flanking width of 0 and a minimum junction coverage of 1. The 'Insert Size Options' section includes a checked option for 'Compute' with a minimum percentile of 0.5 and a maximum percentile of 99.5. The 'OK' and 'Cancel' buttons are visible at the bottom right of the dialog.

IGV

Variant Calls Alignments Probes Proxy Ion Torrent Advanced

On initial load show: ☒ Alignment Track ☒ Coverage Track ☒ Splice Junction Track

Alignment Track Options

Visibility range threshold (kb): 30 *Range at which alignments become visible*

☒ Downsample reads Max read count: 100 per window size (bases): 50

☒ Shade mismatched bases by quality: 5 to 20

☐ Flag insertions larger than: 1 bases

Mapping quality threshold: 0

Hidden SAM tags: MD,SA

☒ Filter duplicate reads ☐ Flag unmapped pairs

☒ Filter vendor failed reads ☐ Show soft-clipped bases

☐ Filter secondary alignments ☒ Show center line

☐ Filter supplementary alignments

Coverage Track Options

Coverage allele-fraction threshold: 0.2 ☒ Quality weight allele fraction

Splice Junction Track Options

☒ Show flanking regions Min flanking width: 0 Min junction coverage: 1

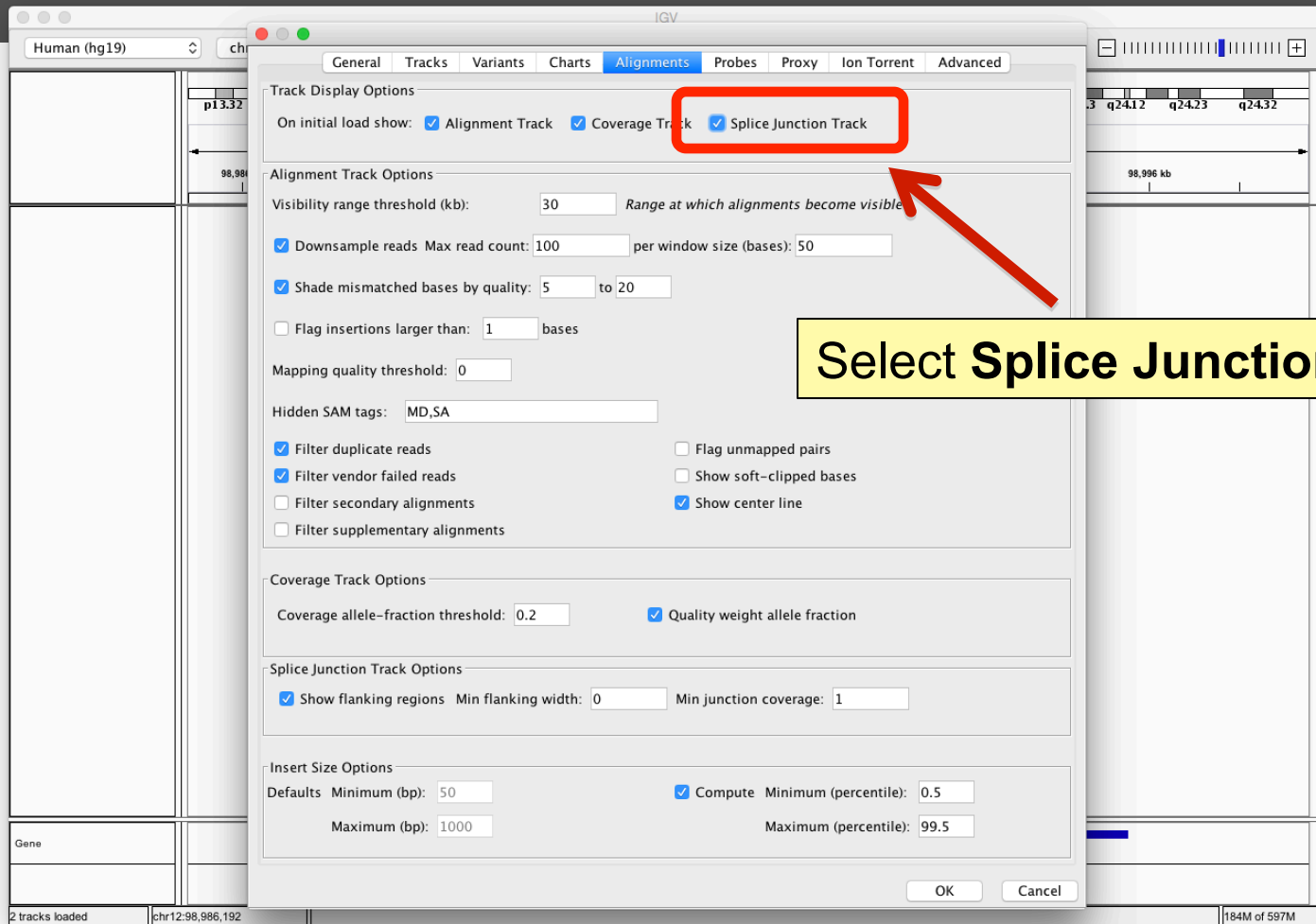
Insert Size Options

Defaults Minimum (bp): 50 ☒ Compute Minimum (percentile): 0.5

Maximum (bp): 1000 Maximum (percentile): 99.5

OK Cancel

2 tracks loaded chr12:98,986,192 184M of 597M



IGV

Human (hg19) chr12

General Tracks Variants Charts **Alignments** Probes Proxy Ion Torrent Advanced

Track Display Options

On initial load show: ☒ Alignment Track ☒ Coverage Track ☒ Splice Junction Track

Alignment Track Options

Visibility range threshold (kb): 30 *Range at which alignments become visible*

☒ Downsample reads Max read count: 100 per window size (bases): 50

☒ Shade mismatched bases by quality: 5 to 20

☐ Flag insertions larger than: 1 bases

Mapping quality threshold: 0

Hidden SAM tags: MD,SA

☒ Filter duplicate reads ☐ Flag unmapped pairs

☒ Filter vendor failed reads ☐ Show soft-clipped bases

☐ Filter secondary alignments ☒ Show center line

☐ Filter supplementary alignments

Coverage Track Options

Coverage allele-fraction threshold: 0.2 ☒ Quality weight allele fraction

Splice Junction Track Options

☒ Show flanking regions Min flanking width: 0 Min junction coverage: 1

Minimum (percentile): 0.5

Maximum (bp): 1000

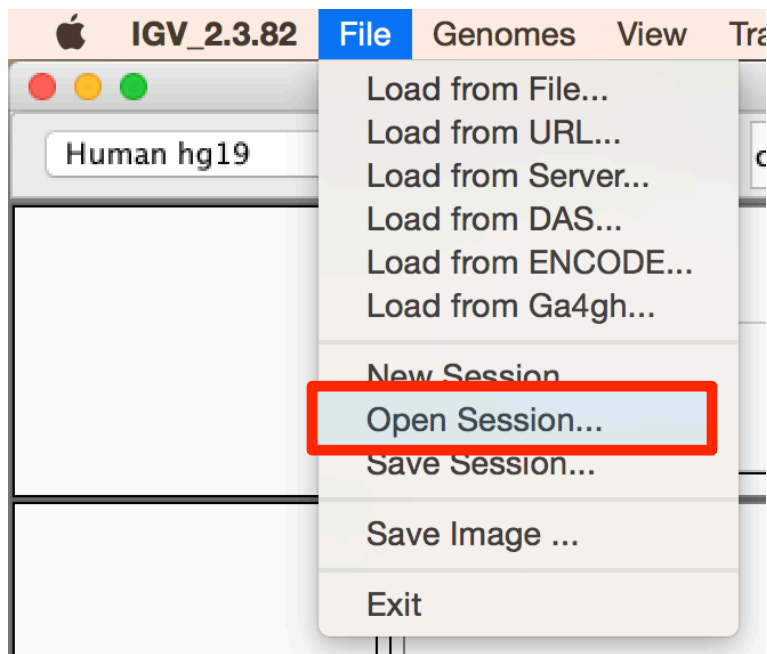
Maximum (percentile): 99.5

Click **OK** to save changes

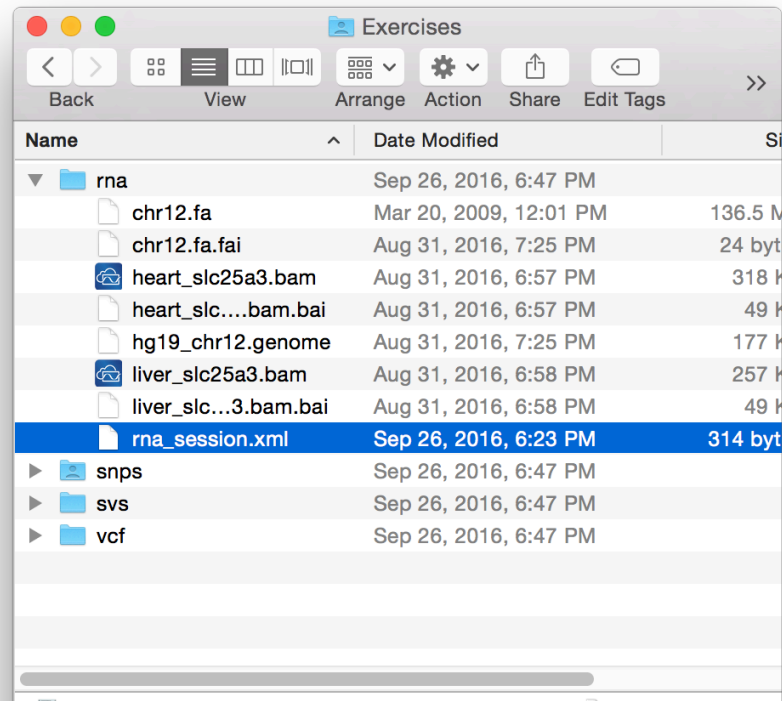
OK Cancel

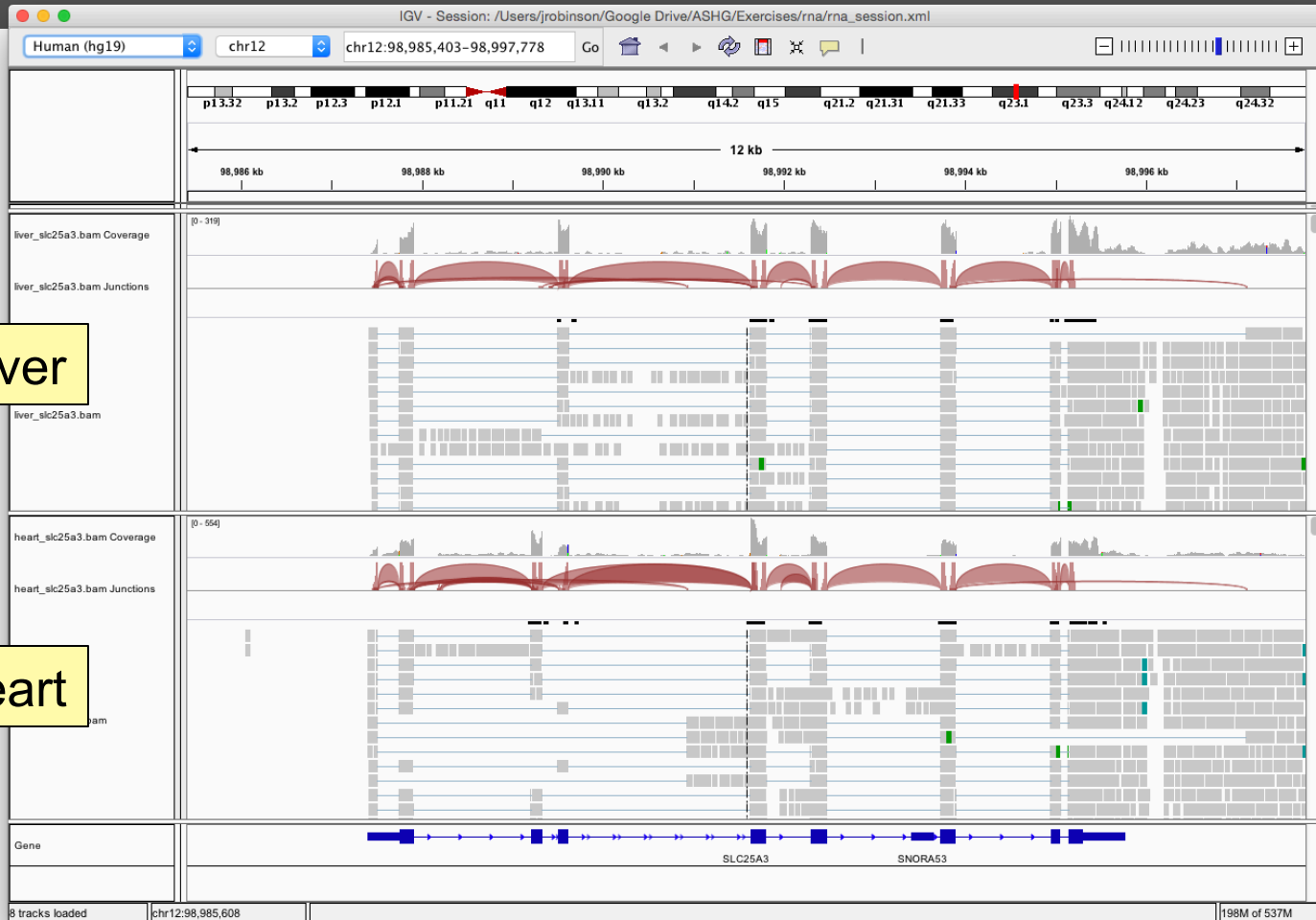
2 tracks loaded chr12:98,986,192 184M of 597M

- Click **File > Open Session**



- Navigate to the folder where you downloaded the workshop data, and select **rna / rna_session.xml**





Liver

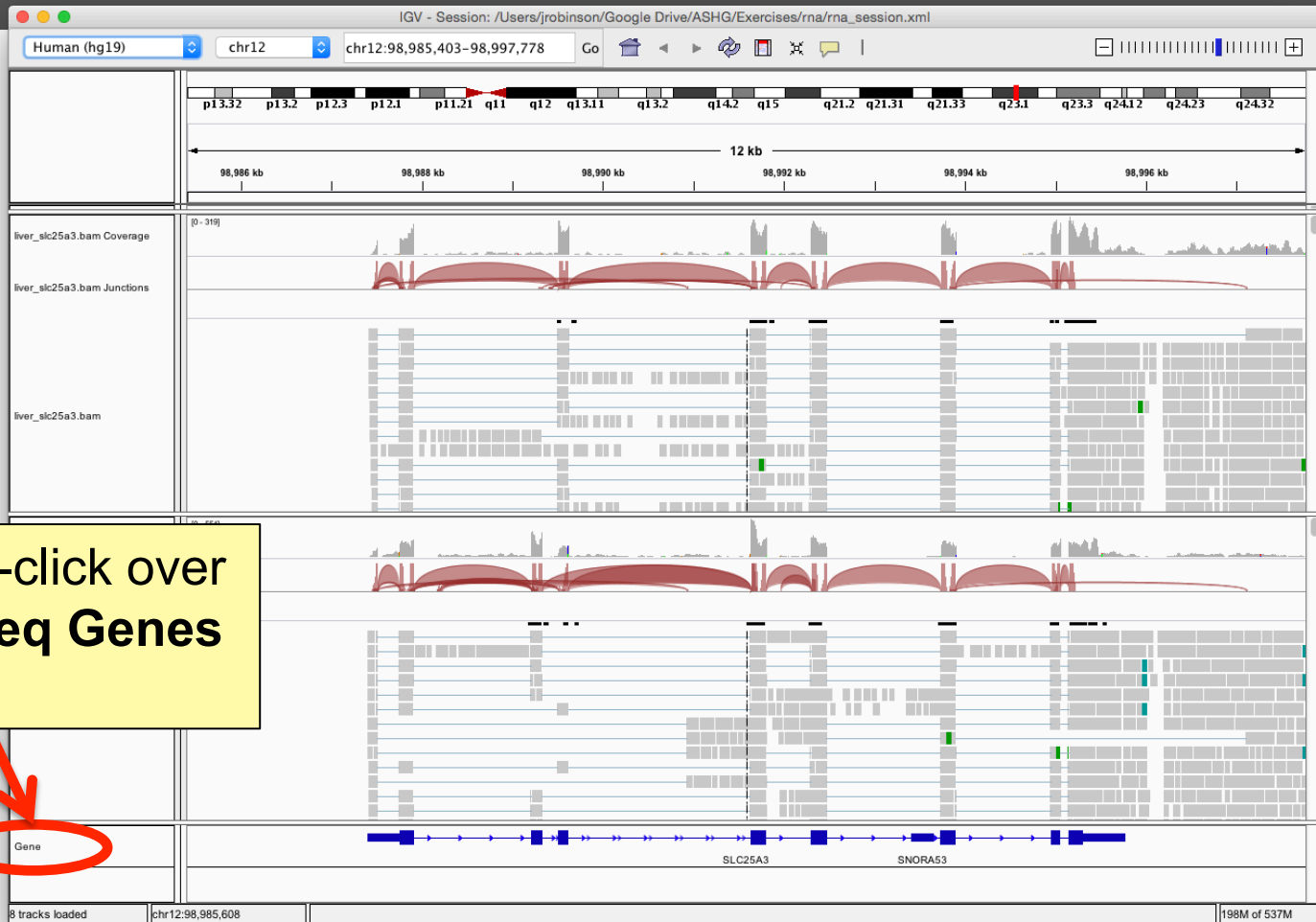
Heart

Coverage

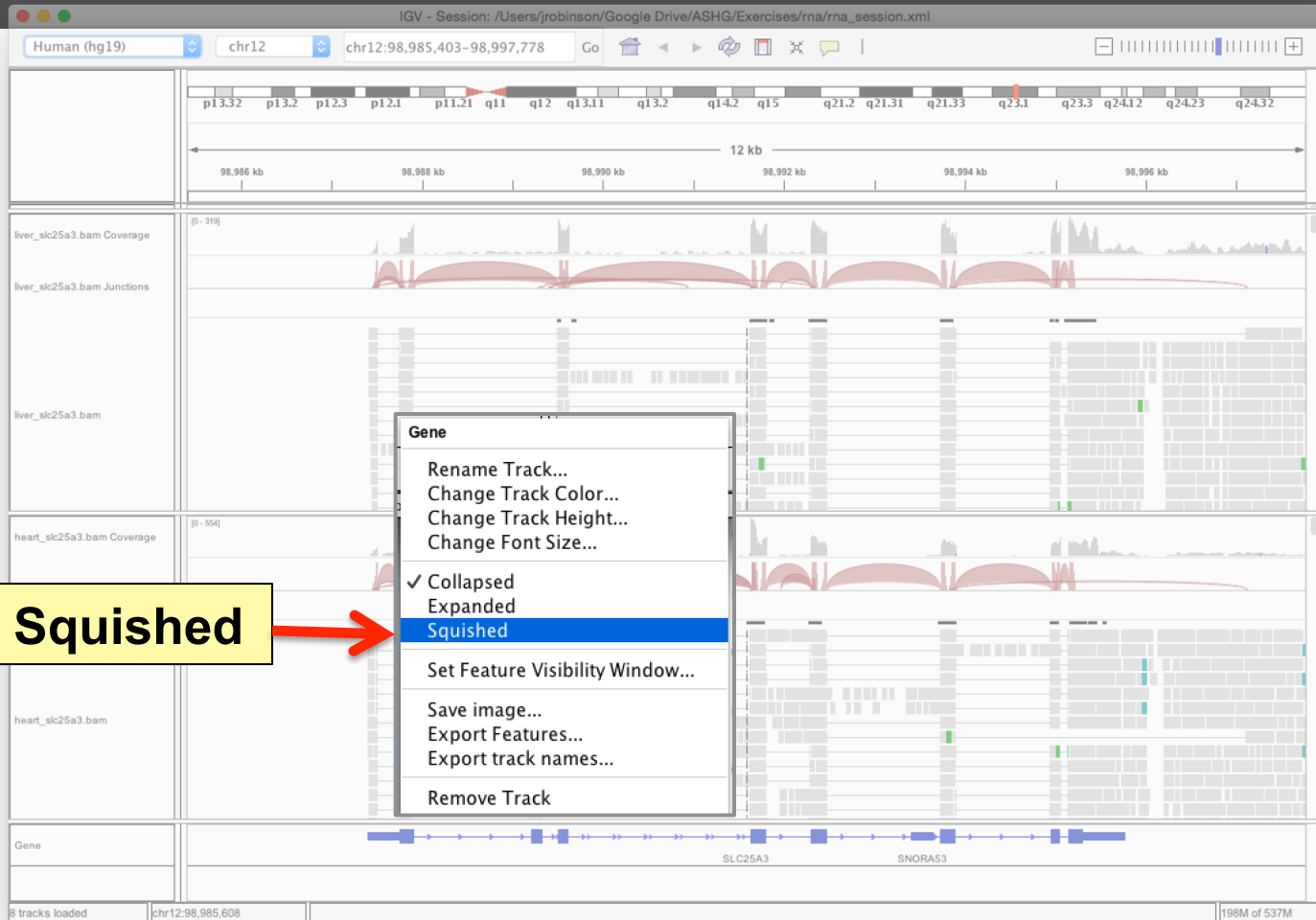


Junction Coverage

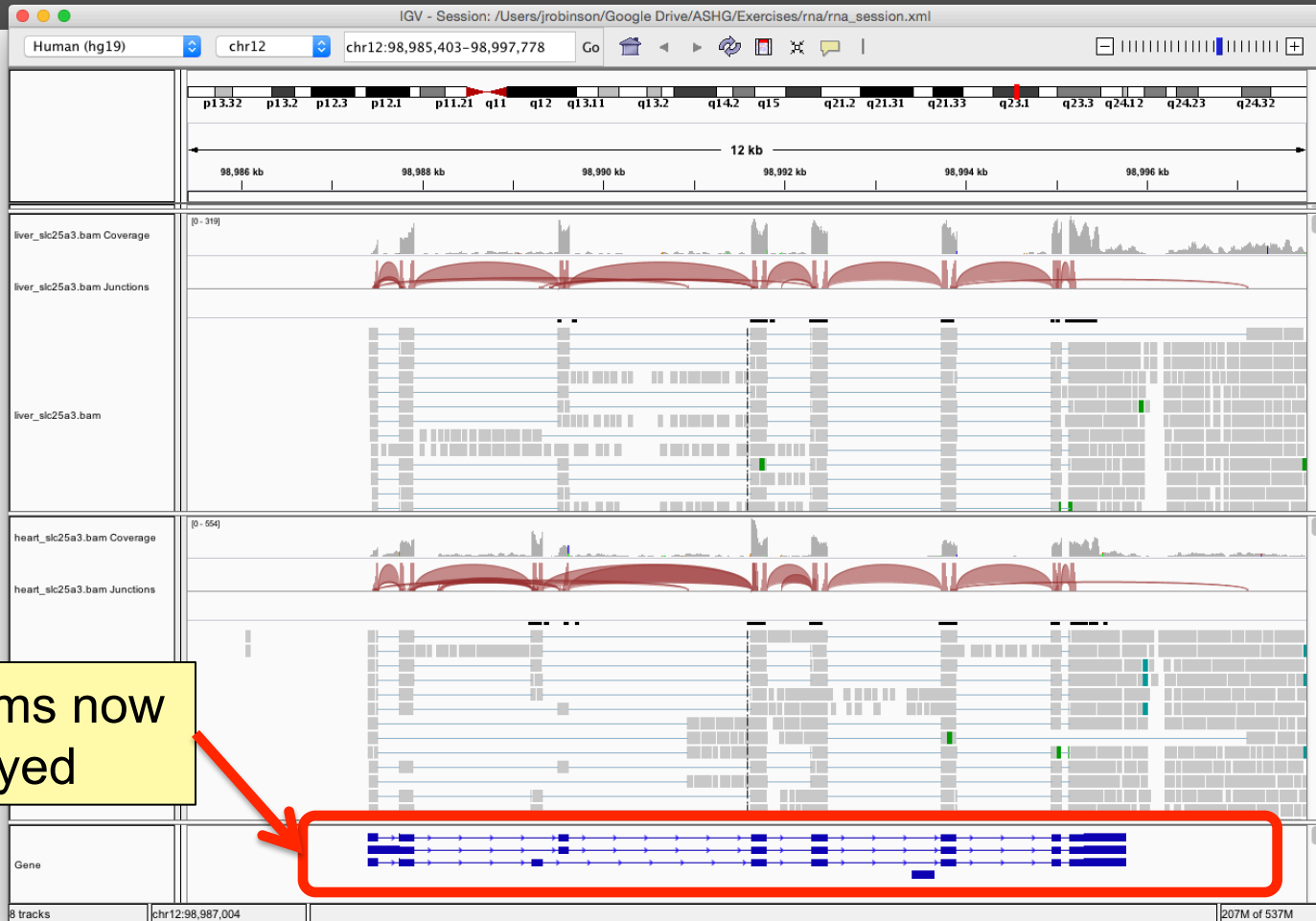




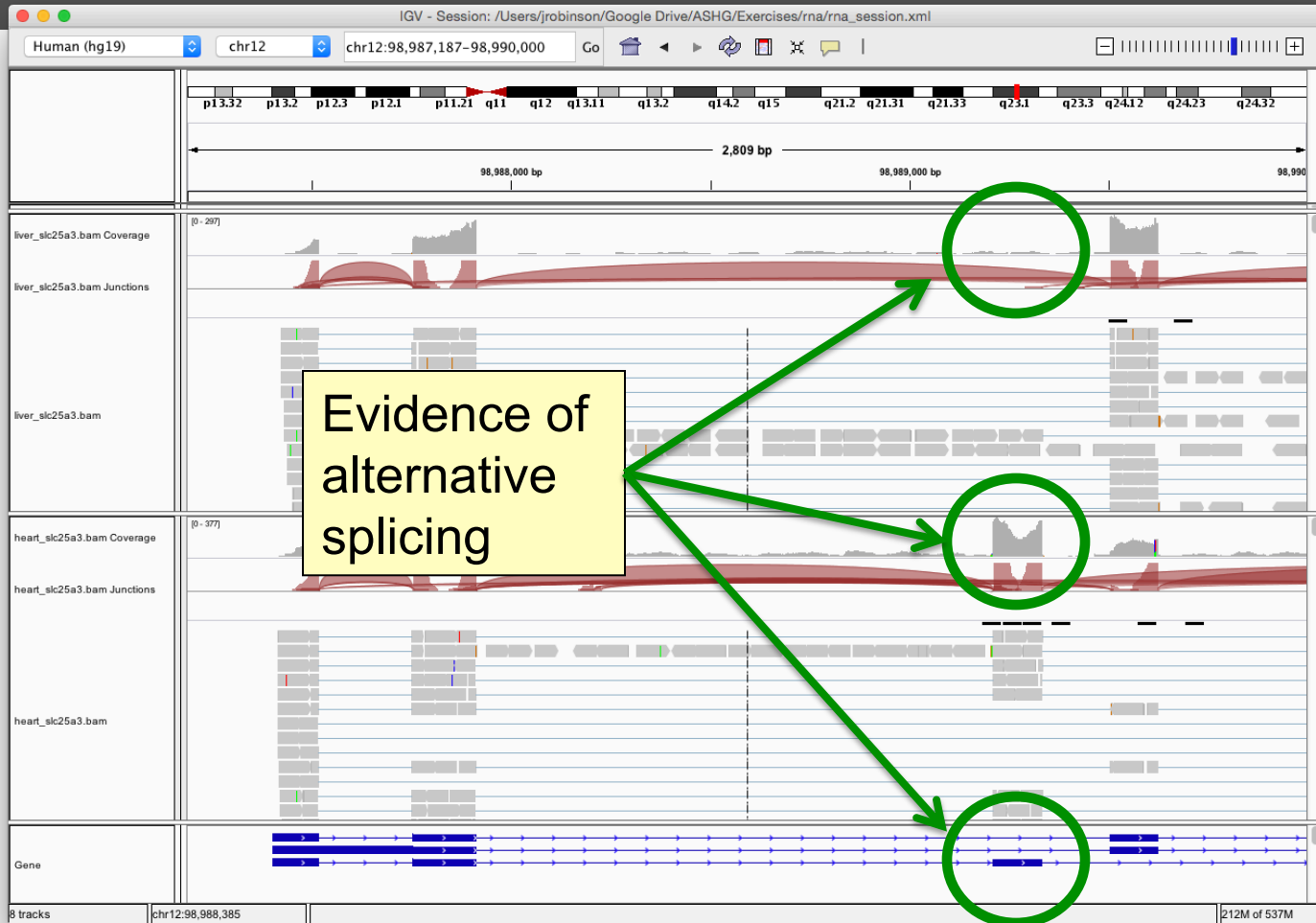
Right-click over
RefSeq Genes
track



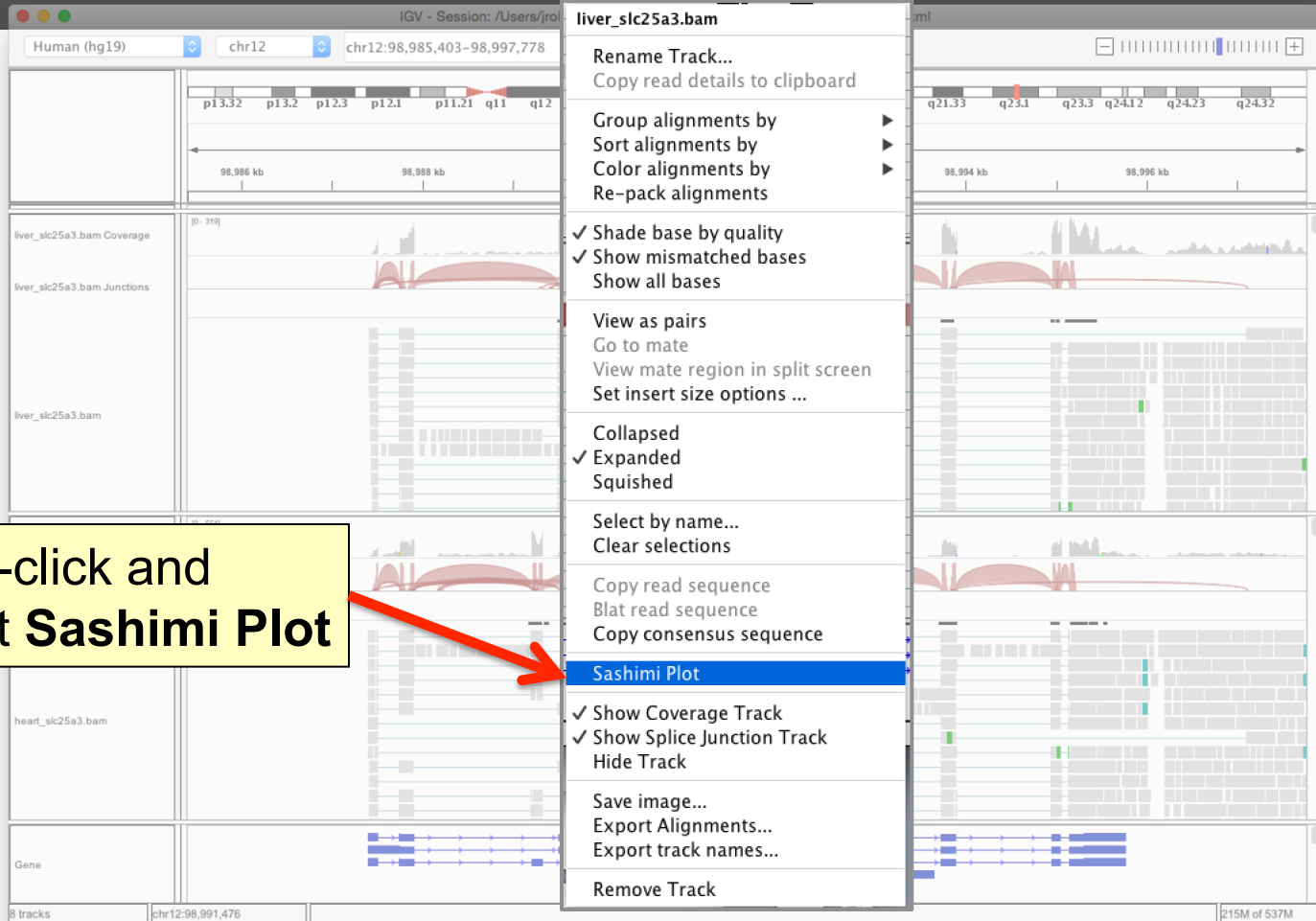
Select **Squished**

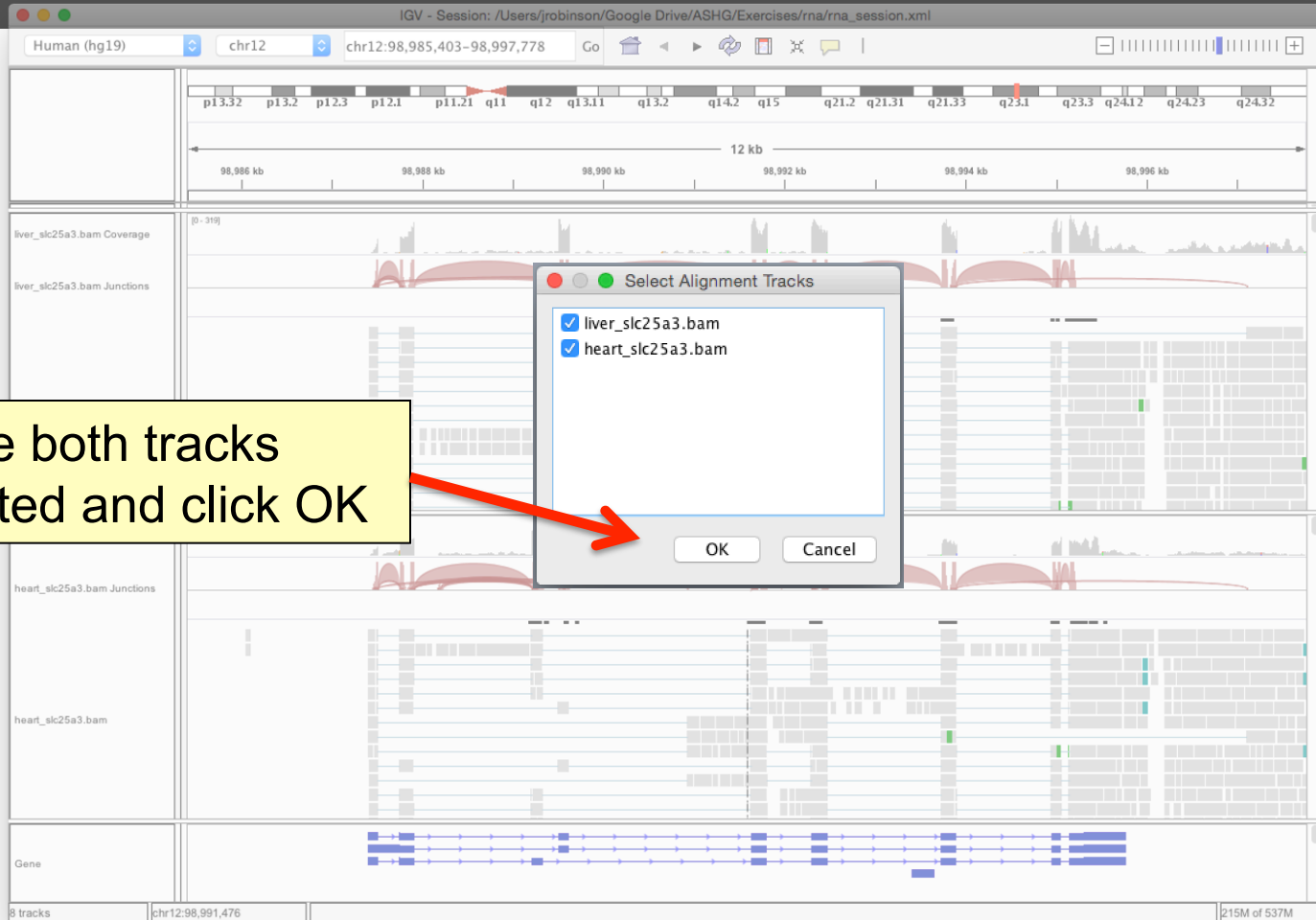


Isoforms now
displayed

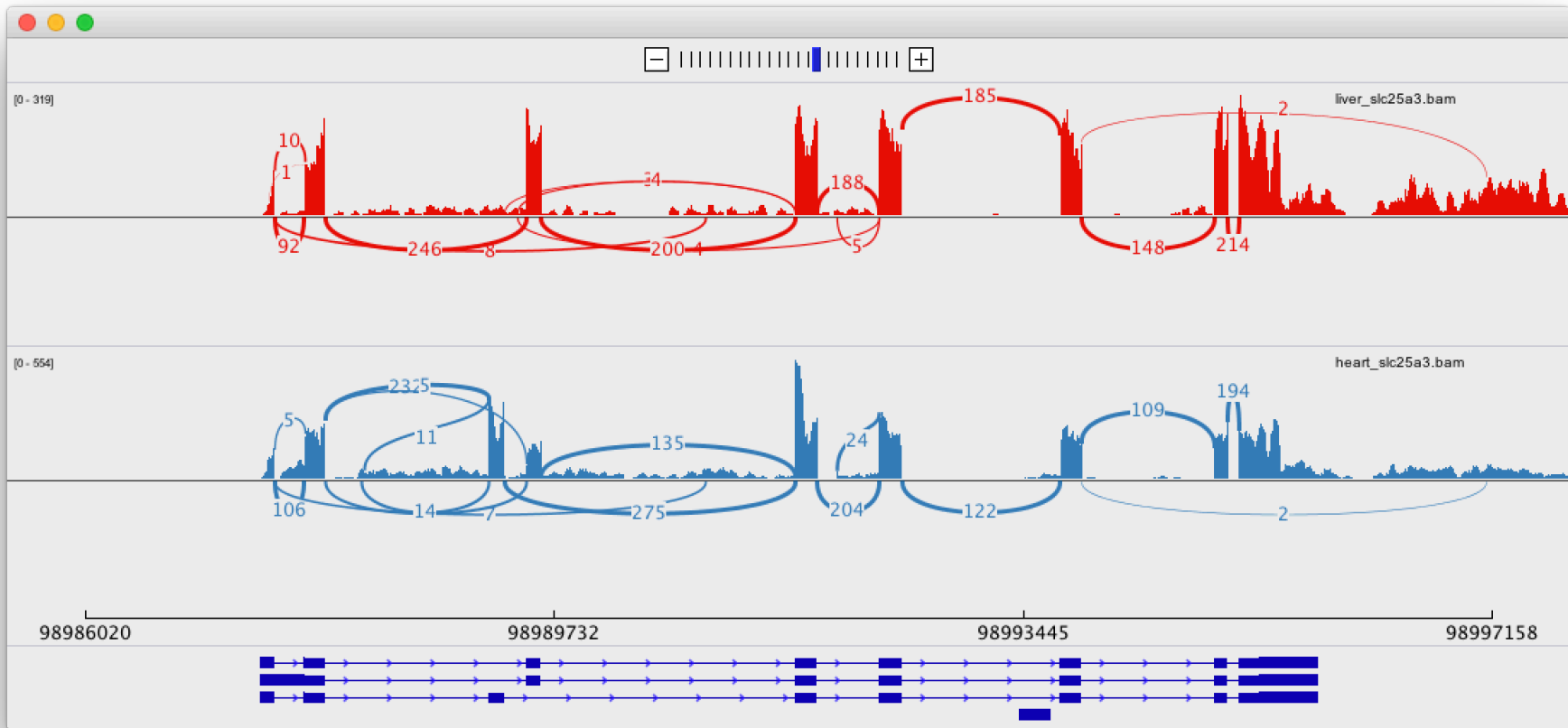


Right-click and
select **Sashimi Plot**





Leave both tracks
selected and click OK

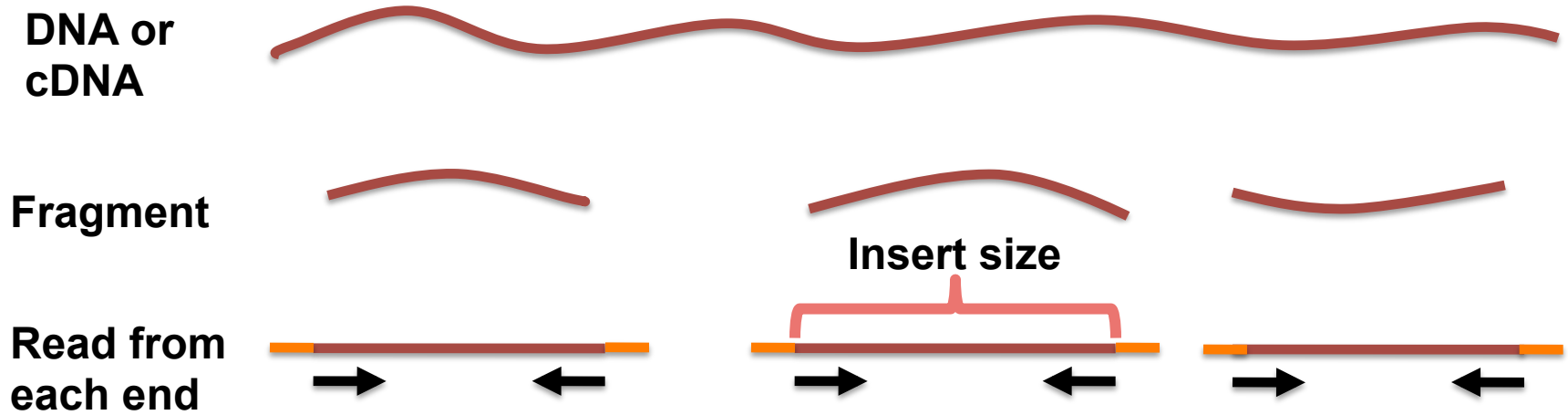


Viewing Structural Events

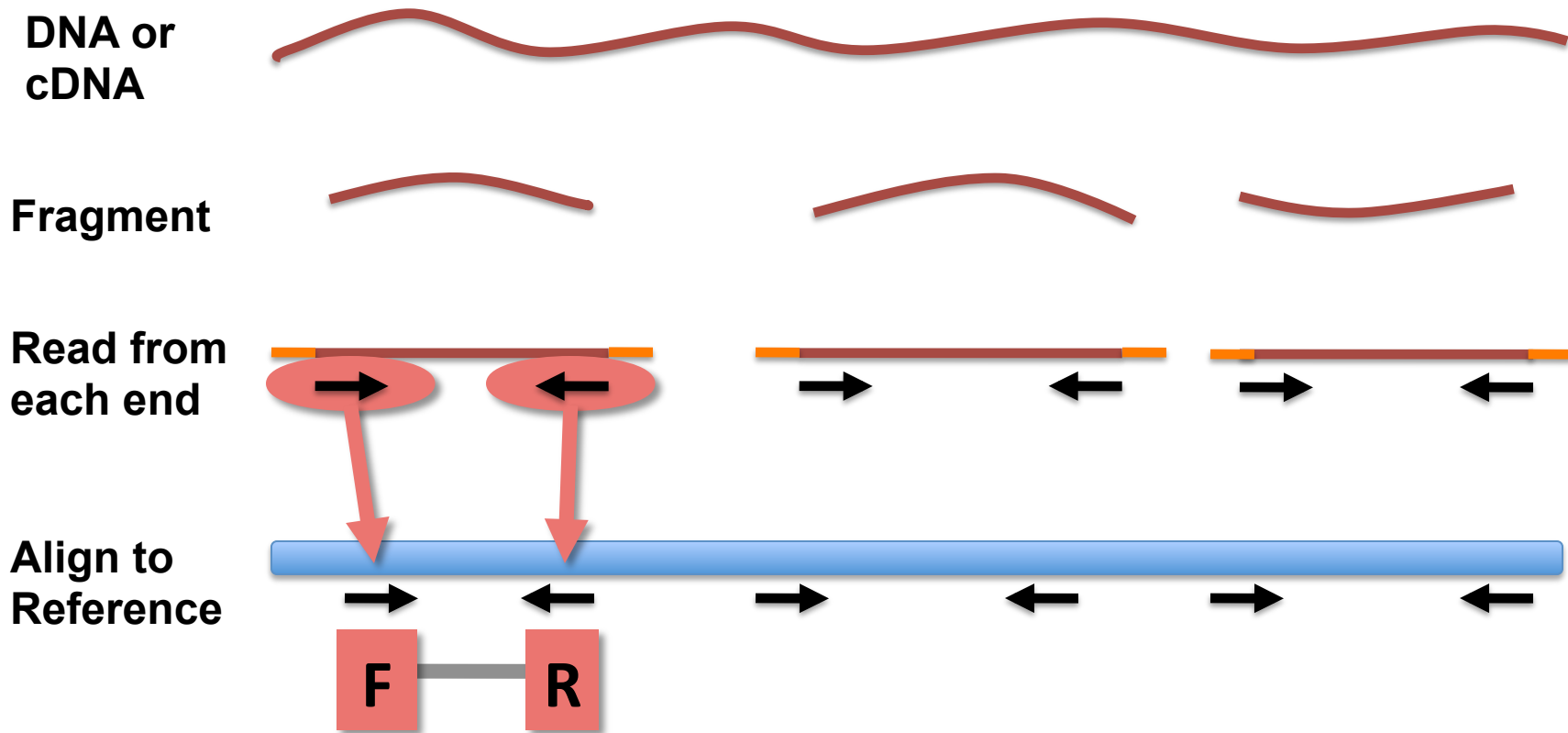
Structural events

- Structural events include deletions, duplications, translocations, and inversions.
- Paired reads can yield evidence of structural events.
- Alignment coloring options help highlight these events based on:
 - Inferred insert size (template length)
 - Pair orientation (relative strand of pair)

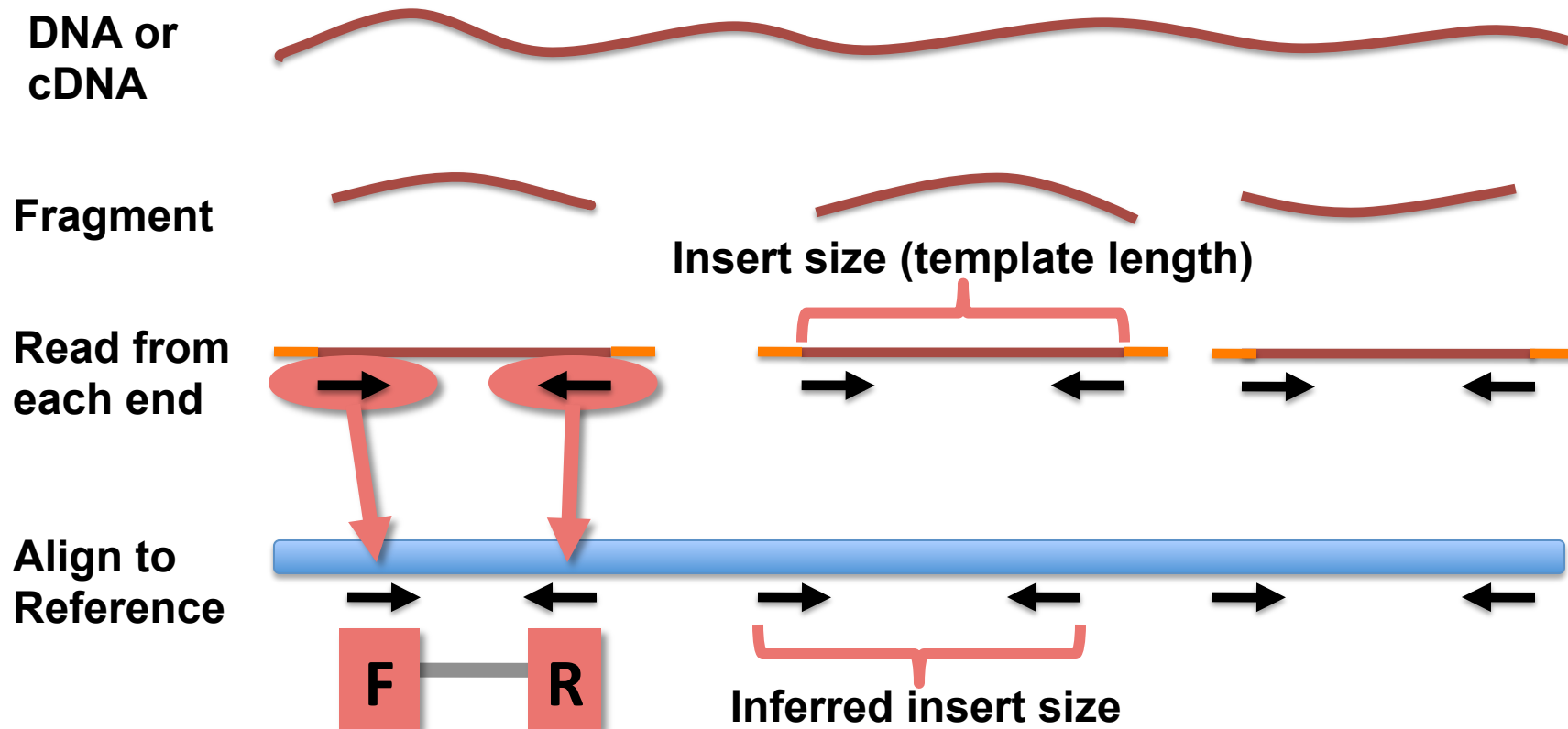
Paired-end sequencing



Paired-end sequencing



Paired-end sequencing



Interpreting Insert Size

Interpreting inferred insert size

The “inferred insert size” can be used to detect evidence of structural variants, including

- Deletions
- Insertions
- Inter-chromosomal rearrangements

Deletion

What is the effect of a deletion on inferred insert size?

Deletion

**Reference
Genome**



Deletion

**Reference
Genome**



Subject



Deletion

**Reference
Genome**



Subject



Deletion

**Reference
Genome**



Subject



Deletion

**Reference
Genome**



Subject

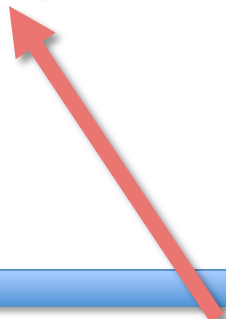


Deletion

**Reference
Genome**

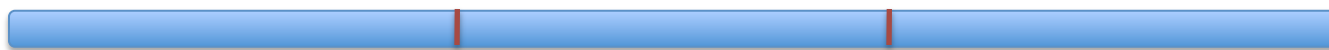


Subject



Deletion

**Reference
Genome**



Subject



Deletion

**Reference
Genome**



Inferred insert size

Subject



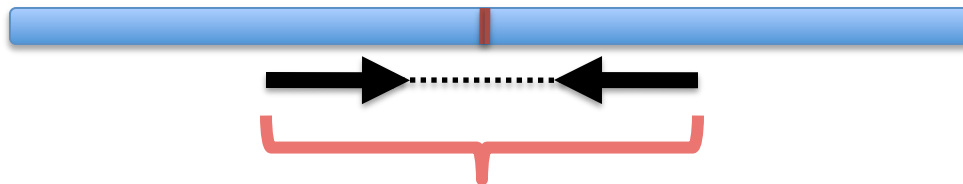
Deletion

**Reference
Genome**



Inferred insert size

Subject

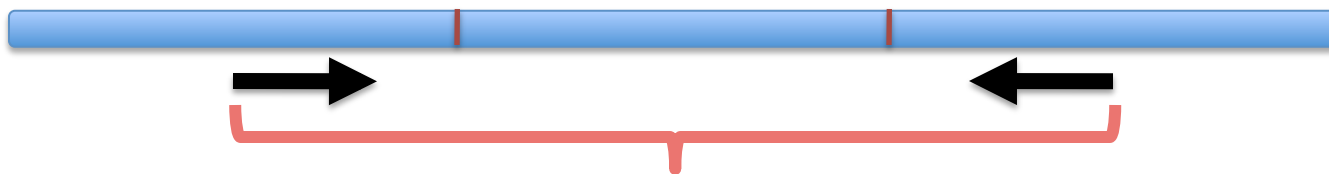


Expected insert size

Deletion

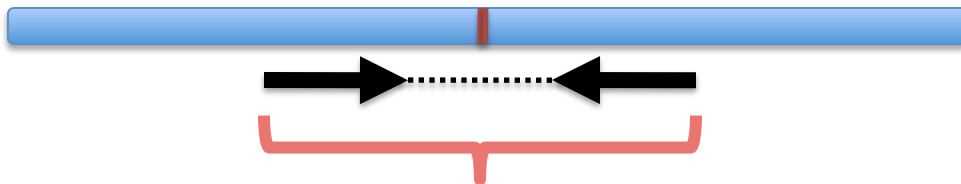
Inferred insert size is greater than expected value

Reference
Genome



Inferred insert size

Subject



Expected insert size

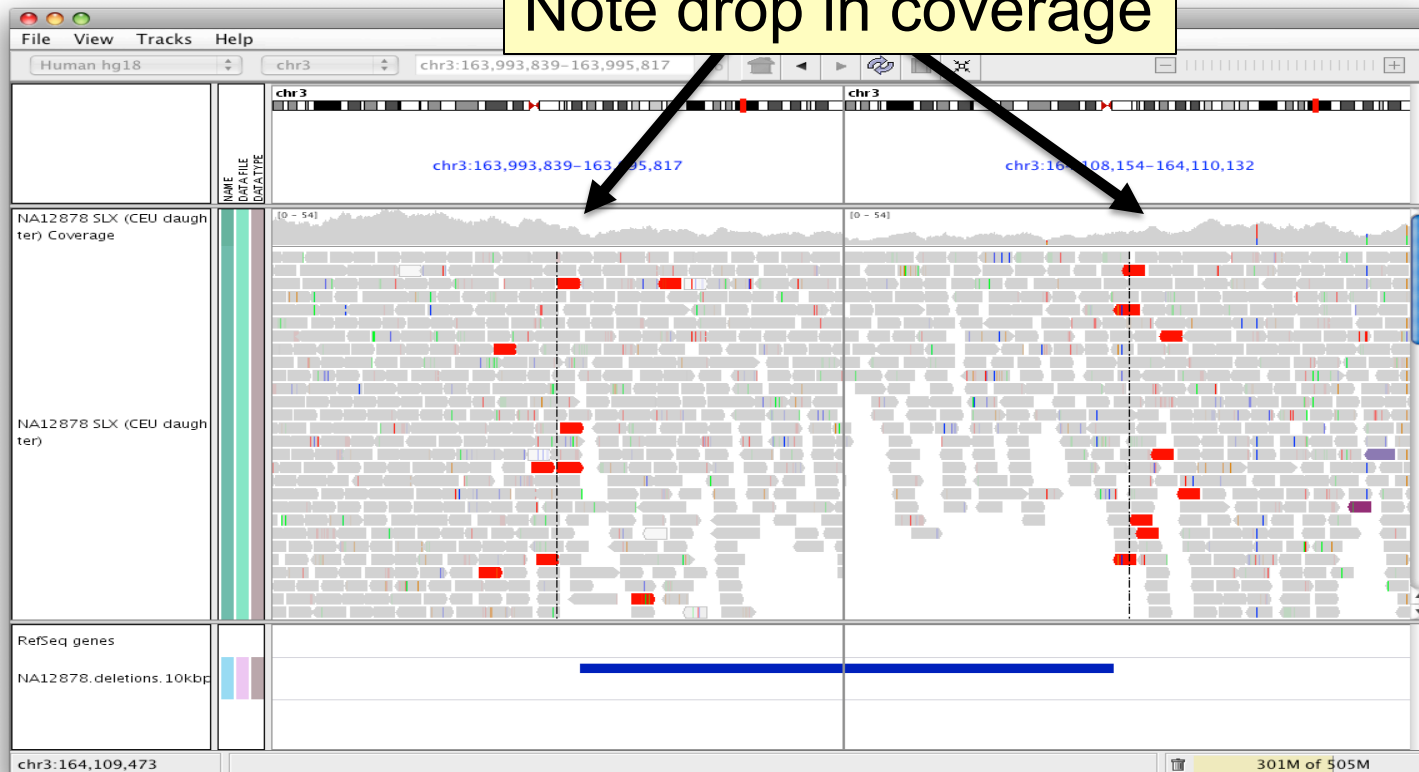
Deletion

Pairs with larger than expected insert size are colored red.



Deletion

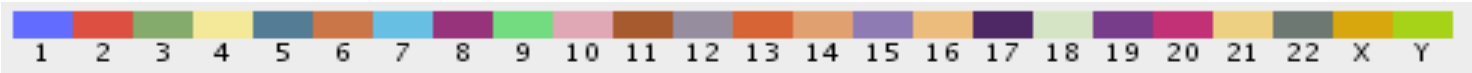
Note drop in coverage



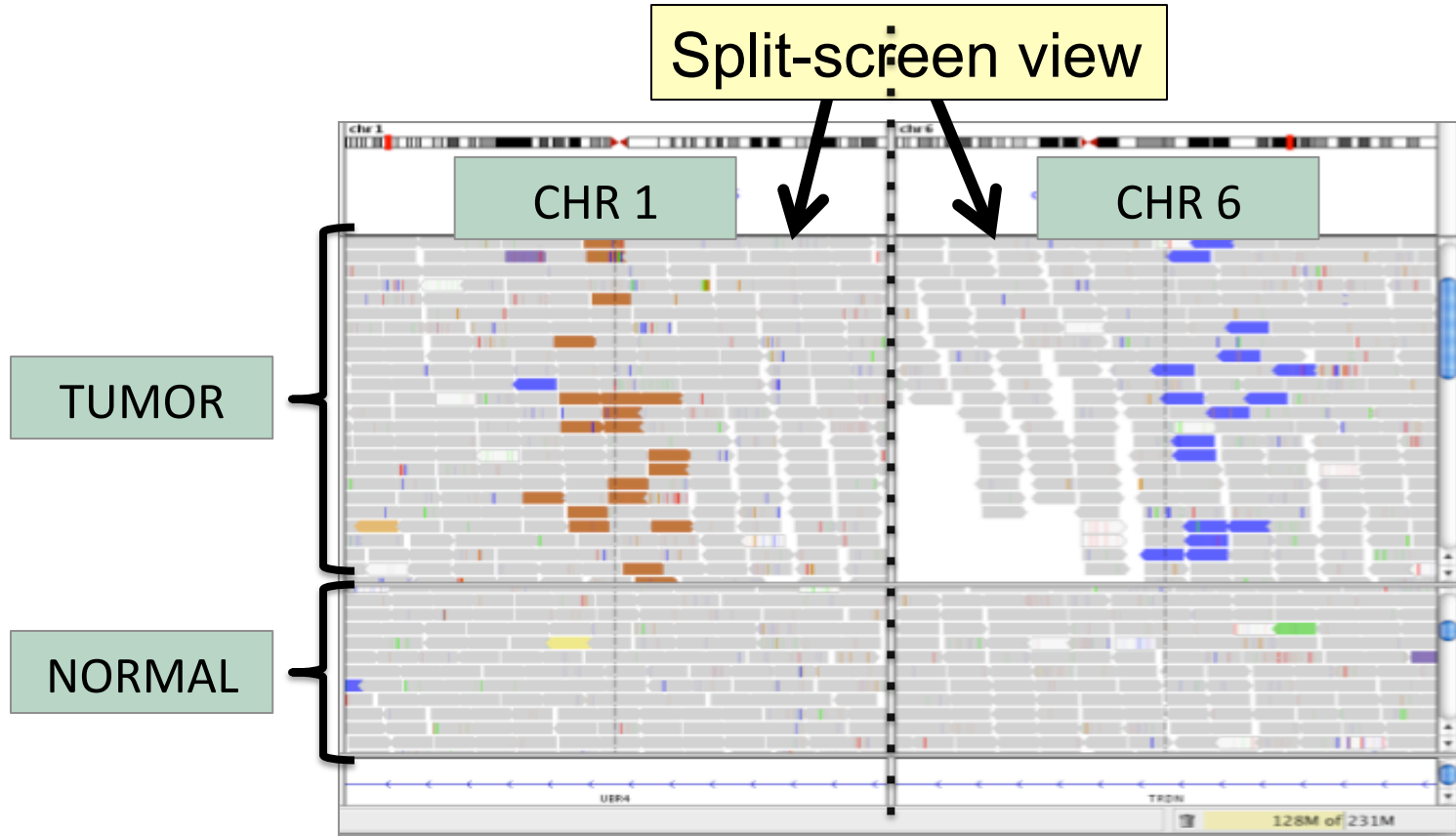
Insert size color scheme

- Smaller than expected insert size: 
- Larger than expected insert size: 
- Pairs on different chromosomes:

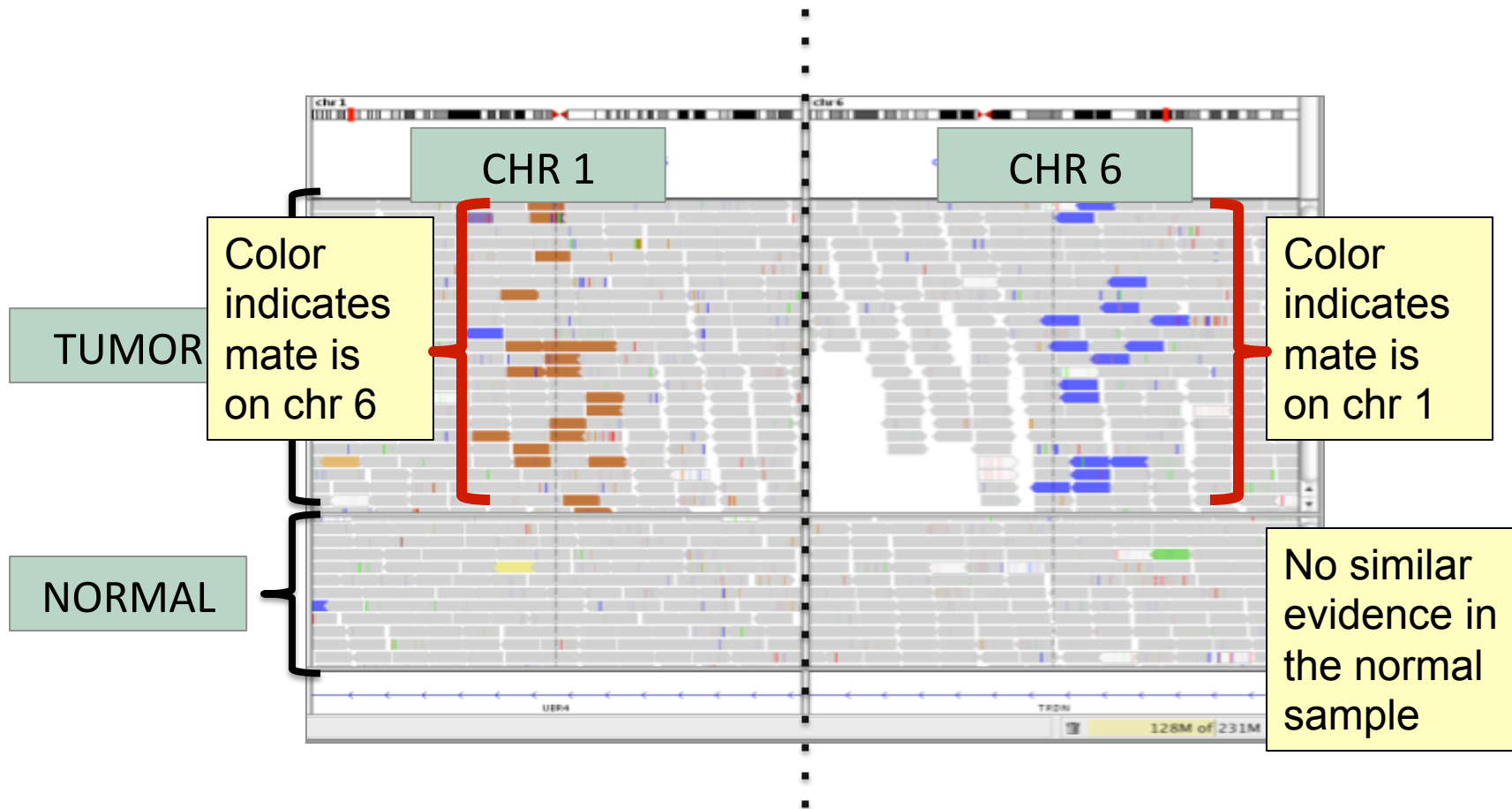
Each end colored by chromosome of its mate



Rearrangement



Rearrangement



Interpreting Pair Orientations

Interpreting pair orientations

Orientation of paired reads can reveal evidence of structural events, including:

- Inversions
- Duplications
- Translocations

Inversion

**Reference
Genome**



Inversion

**Reference
Genome**

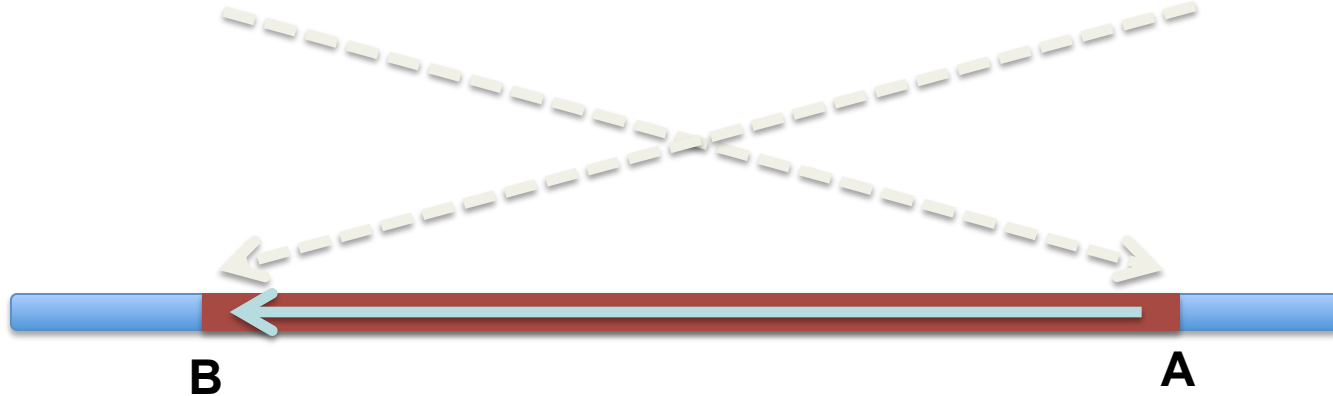


Inversion

**Reference
Genome**



Subject



Inversion

**Reference
Genome**



Subject



Inversion

Reference
Genome



Subject

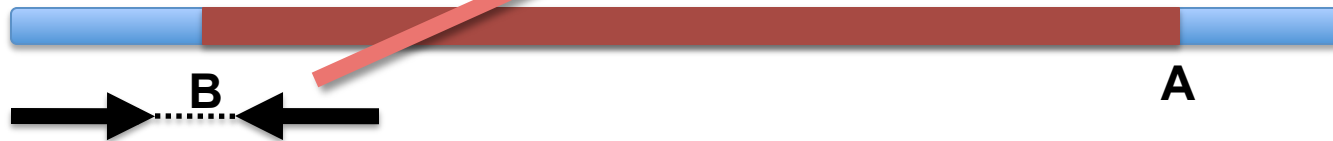


Inversion

Reference
Genome



Subject



Inversion

Reference
Genome



Subject



Inversion

Reference
Genome

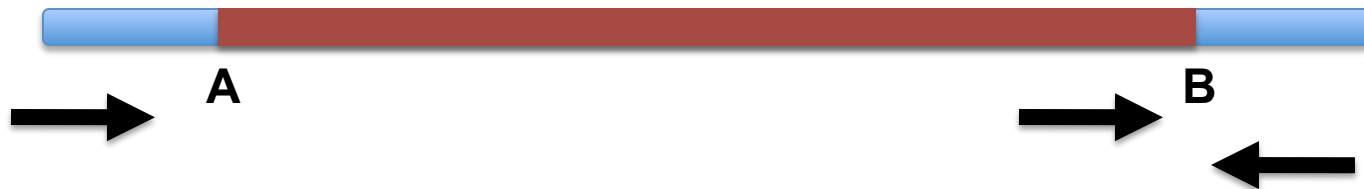


Subject

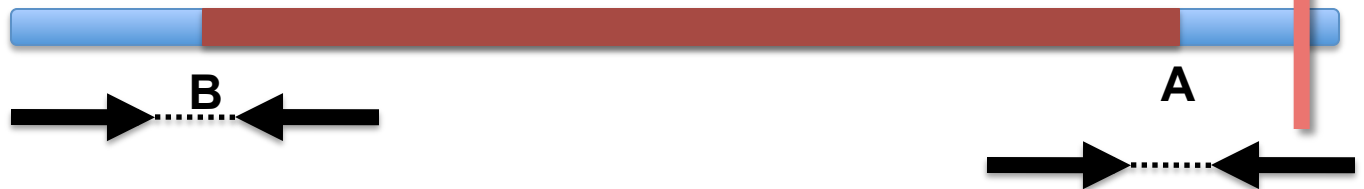


Inversion

Reference
Genome

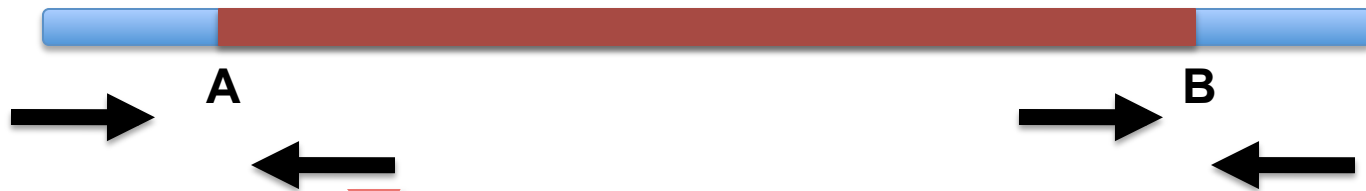


Subject



Inversion

Reference
Genome



Subject



Inversion

Reference
Genome



Subject



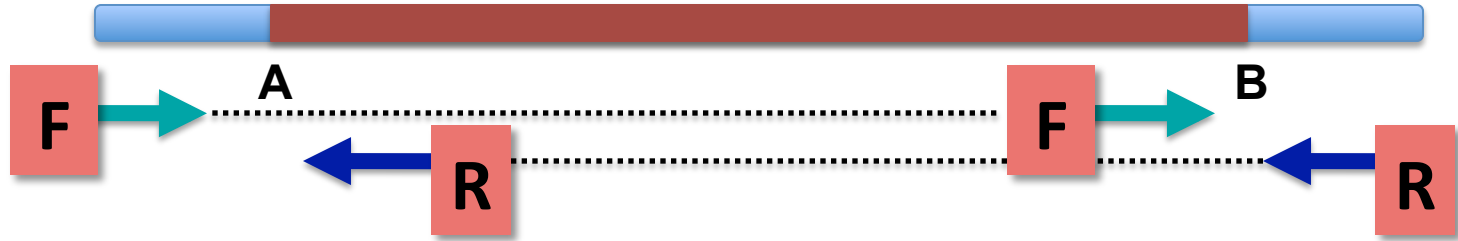
Inversion

Reference
Genome

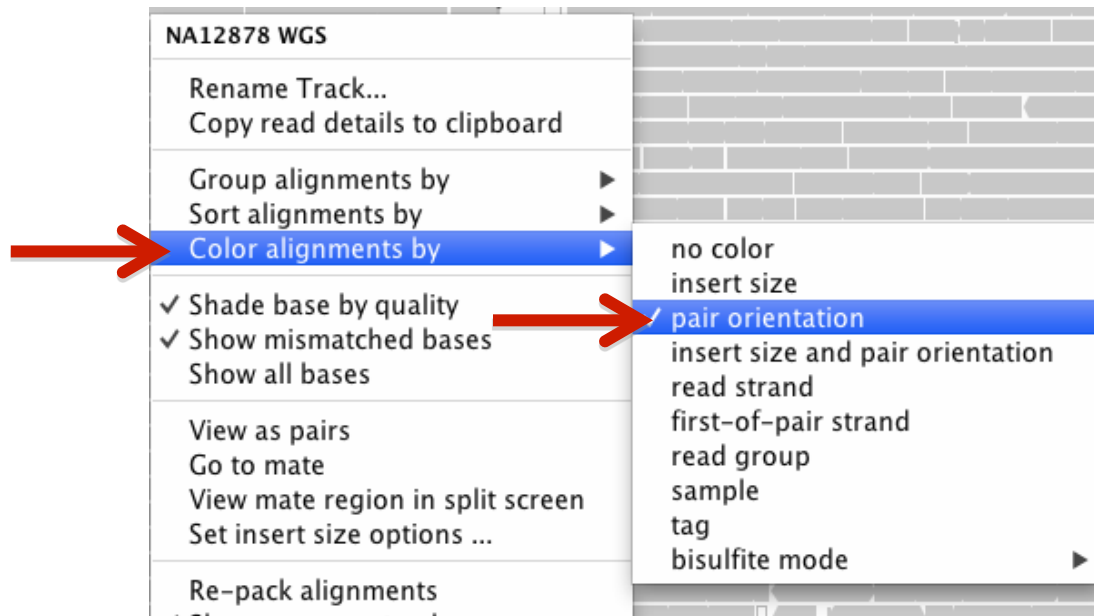


Inversion

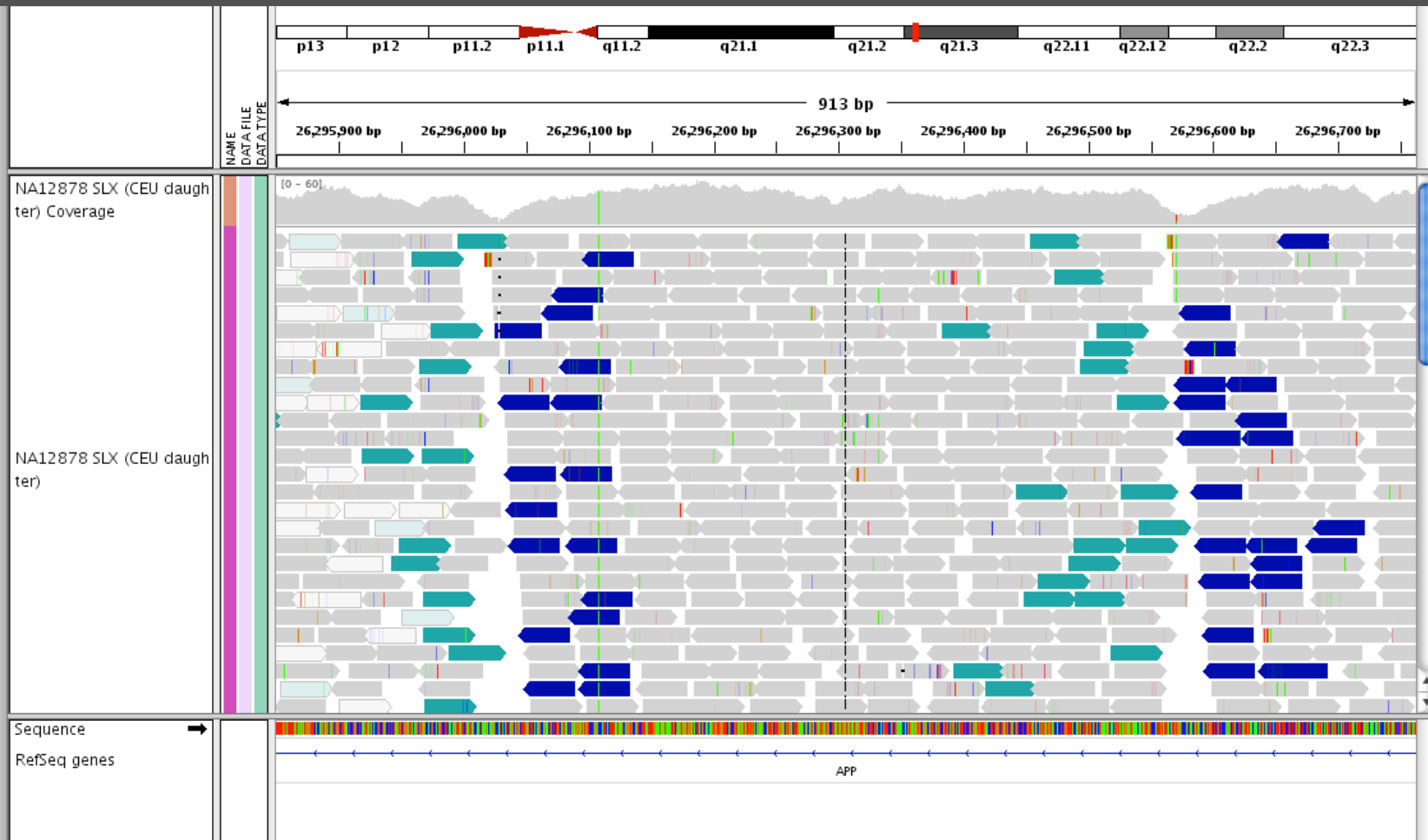
Reference
Genome



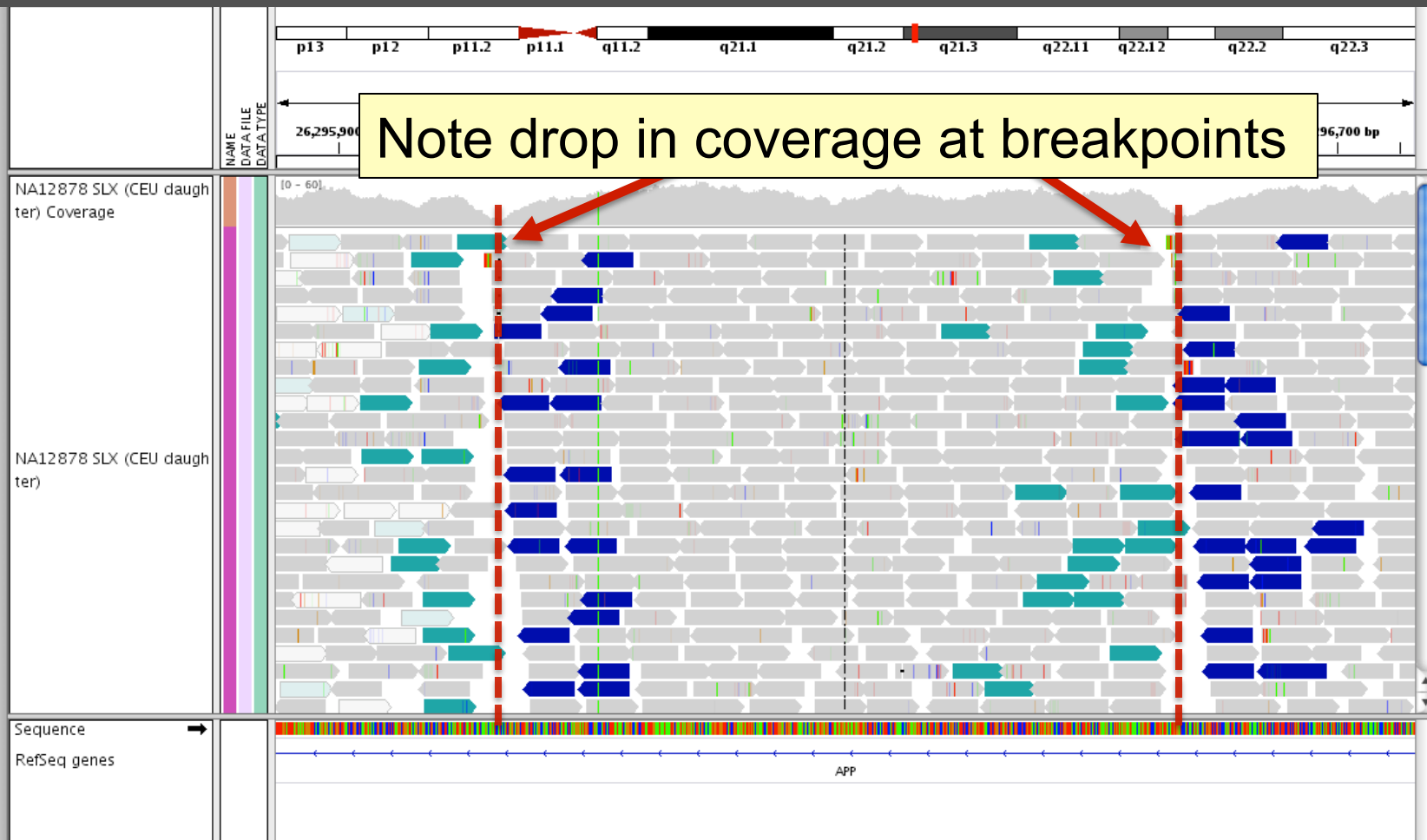
Color by pair orientation



Inversion



Inversion



Hands on

Use color and viewing options to examine an inversion

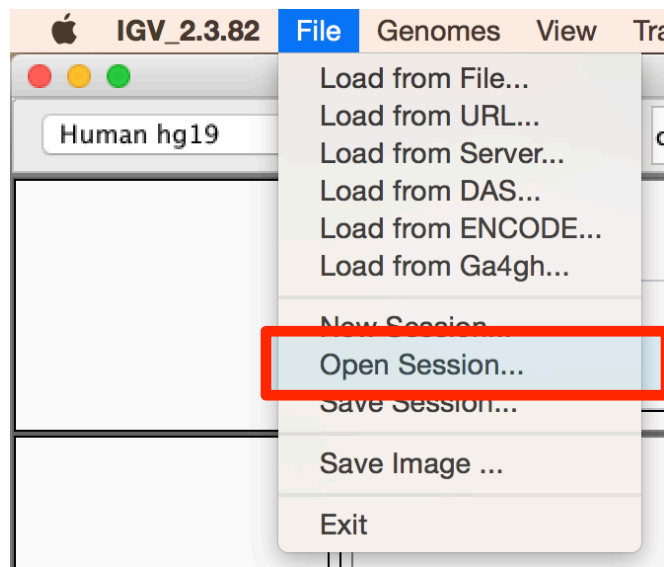
Data from 1000 Genomes

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

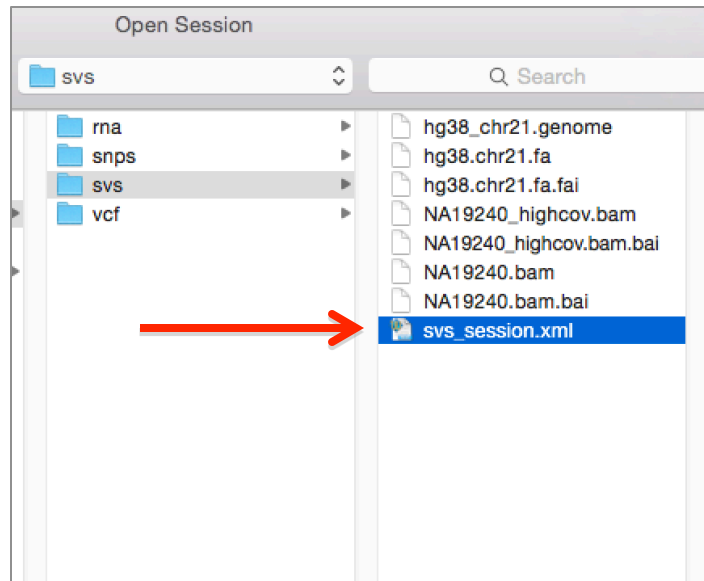
Structural events: igv.org/workshops/ASHG2016/ExInstr.pdf

- 1 **Initialize exercise:** – Click **File > Open Session**. Go to the folder where you downloaded the workshop data and select **svs / svs_session.xml**.
- 2 Observe colorful alignments. By default color mode = By insert size and pair orientation
- 3
 - Right-click over alignments and select **Color alignments by > insert size**.
Observe: changes in color.
 - Right-click over alignments and select **Color alignments by > pair orientation**.
Observe: most of the initial colorful alignments are now lit up again.
 - Right-click over alignments and select **View as pairs**.
- 4 **Observe evidence of inversion:** High % of forward-forward pairs and reverse-reverse pairs at the same locus

Click **File > Open Session**



Navigate to the folder where you downloaded the workshop data, and select **svs / svs_session.xml**



Human (hg38 chr21) chr21 chr21:26,000,000–26,004,000

NA19240.bam

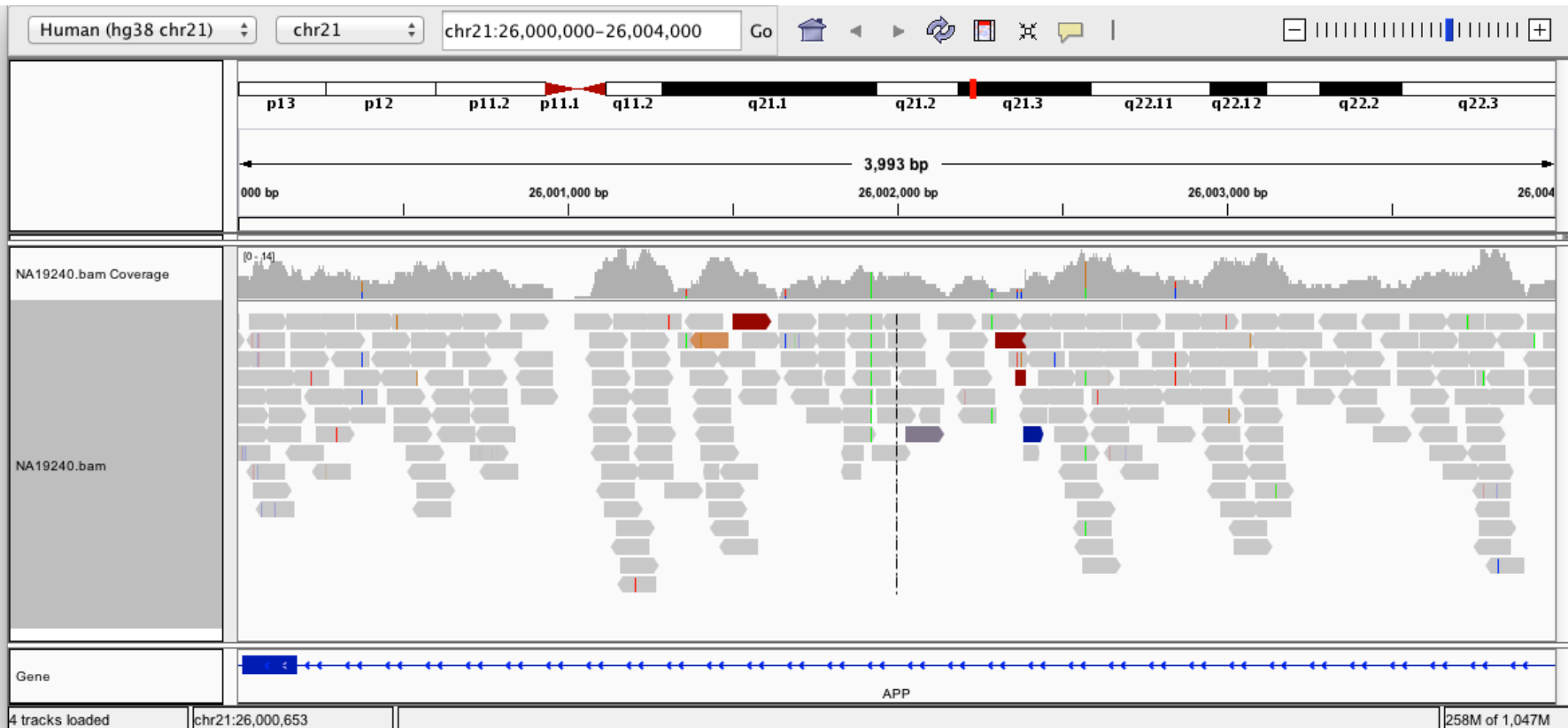
- Rename Track...
- Copy read details to clipboard
- Group alignments by
- Sort alignments by
- Color alignments by
 - no color
 - insert size
 - pair orientation
 - ✓ insert size and pair orientation
 - read strand
 - first-of-pair strand
 - read group
 - sample
 - library
 - tag
 - bisulfite mode
- Re-pack alignments
- ✓ Shade base by quality
- ✓ Show mismatched bases
- Show all bases
- View as pairs
- Go to mate
- View mate region in split screen
- Set insert size options ...
- Collapsed
- ✓ Expanded
- Squished
- Select by name...
- Clear selections
- Copy read sequence
- Blat read sequence
- Copy consensus sequence
- Sashimi Plot
- ✓ Show Coverage Track
- Show Splice Junction Track
- Hide Track
- Save image...
- Export Alignments...
- Export track names...
- Remove Track

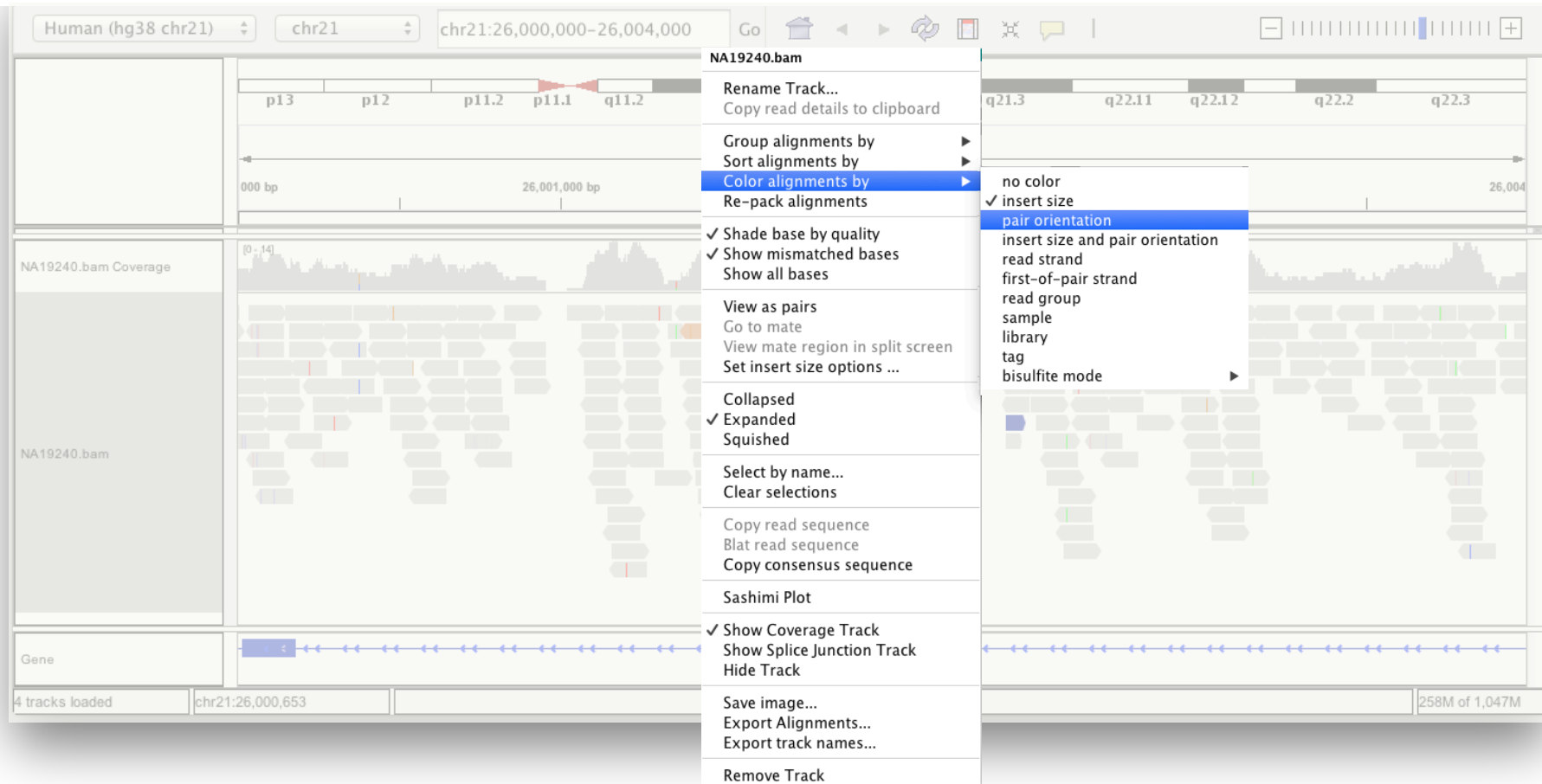
NA19240.bam Coverage

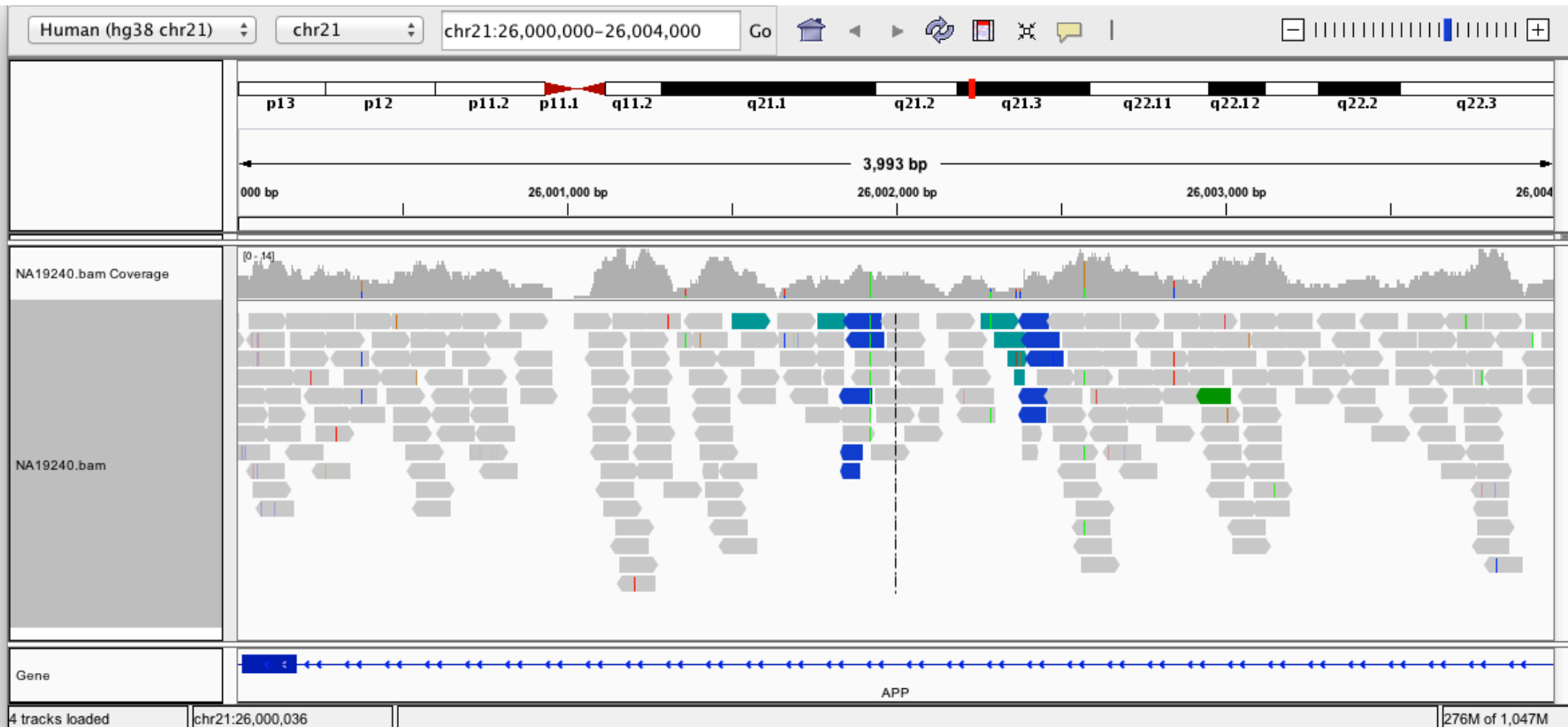
NA19240.bam

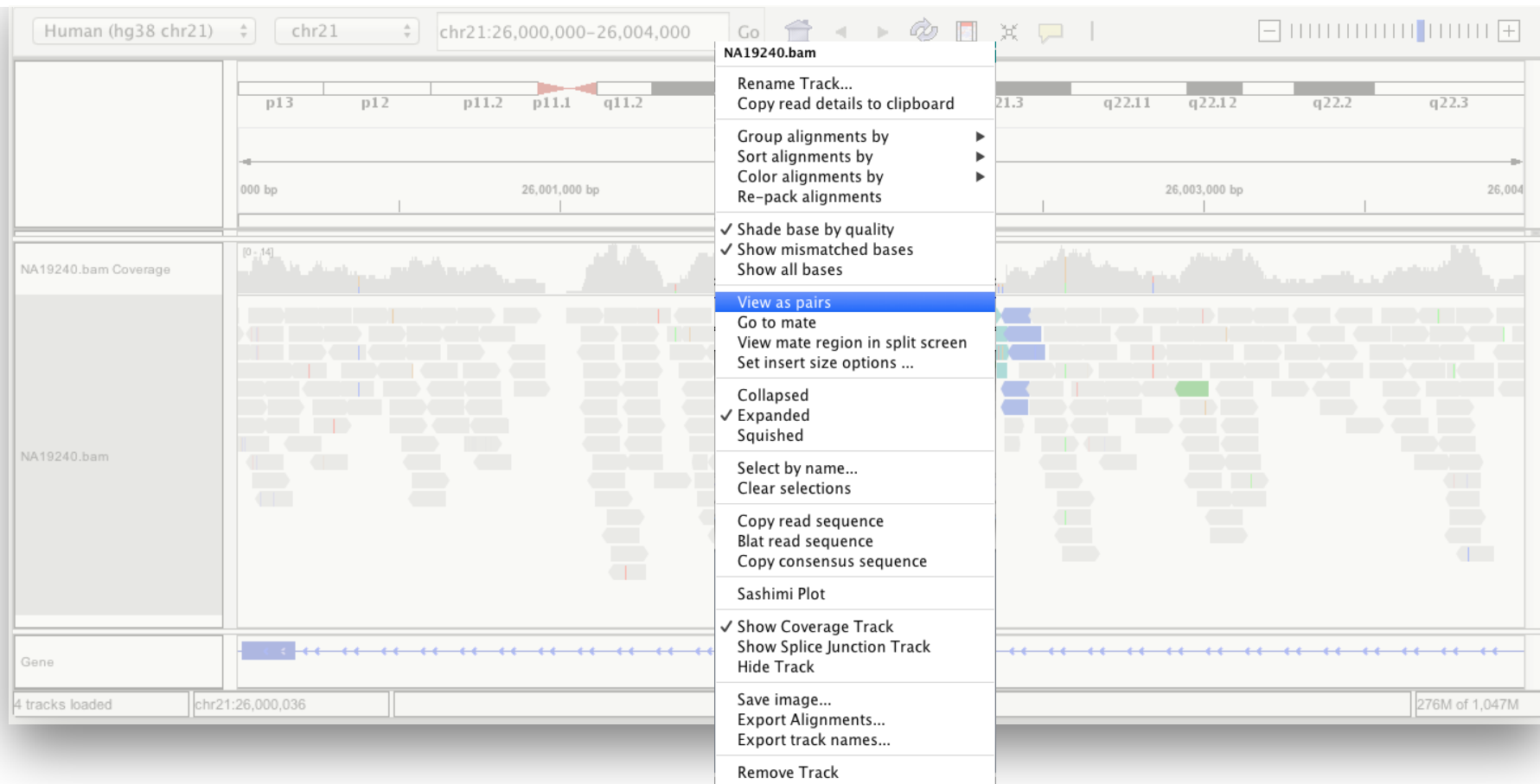
Gene

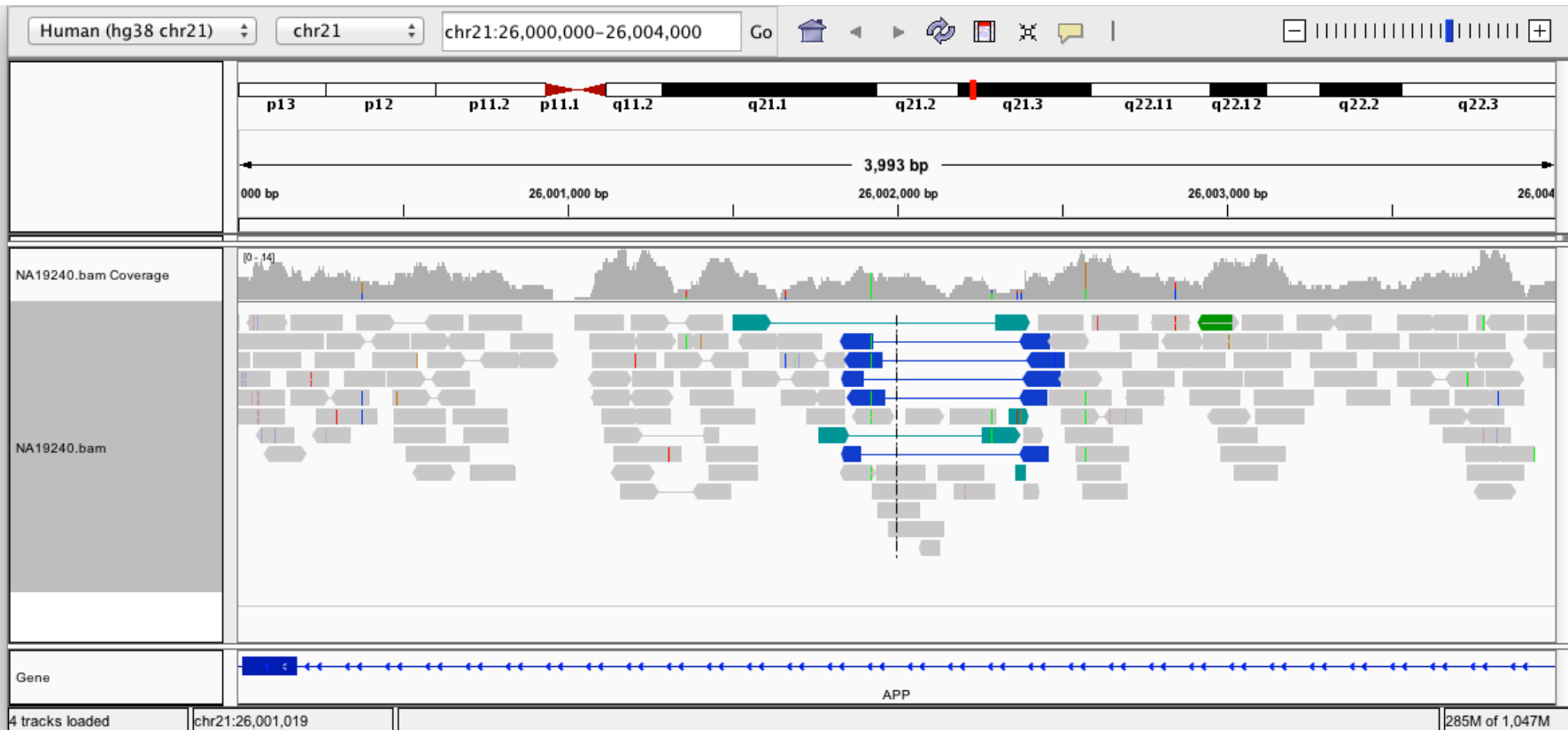
4 tracks loaded chr21:26,000,828 235M of 1,047M











Viewing Structural Events



Third Gen Sequencing

- Applications
 - structural variants, long-range haplotype phasing, de novo assembly, ...
- Long reads:
 - ~10,000 bp up to ~100,000 bp
- High raw error rate in individual reads
 - (high consensus accuracy with sufficient coverage)
- Platforms:
 - PacBio, Oxford Nanopore, 10X, Illumina Tru-Seq, ...

IGV Enhancements

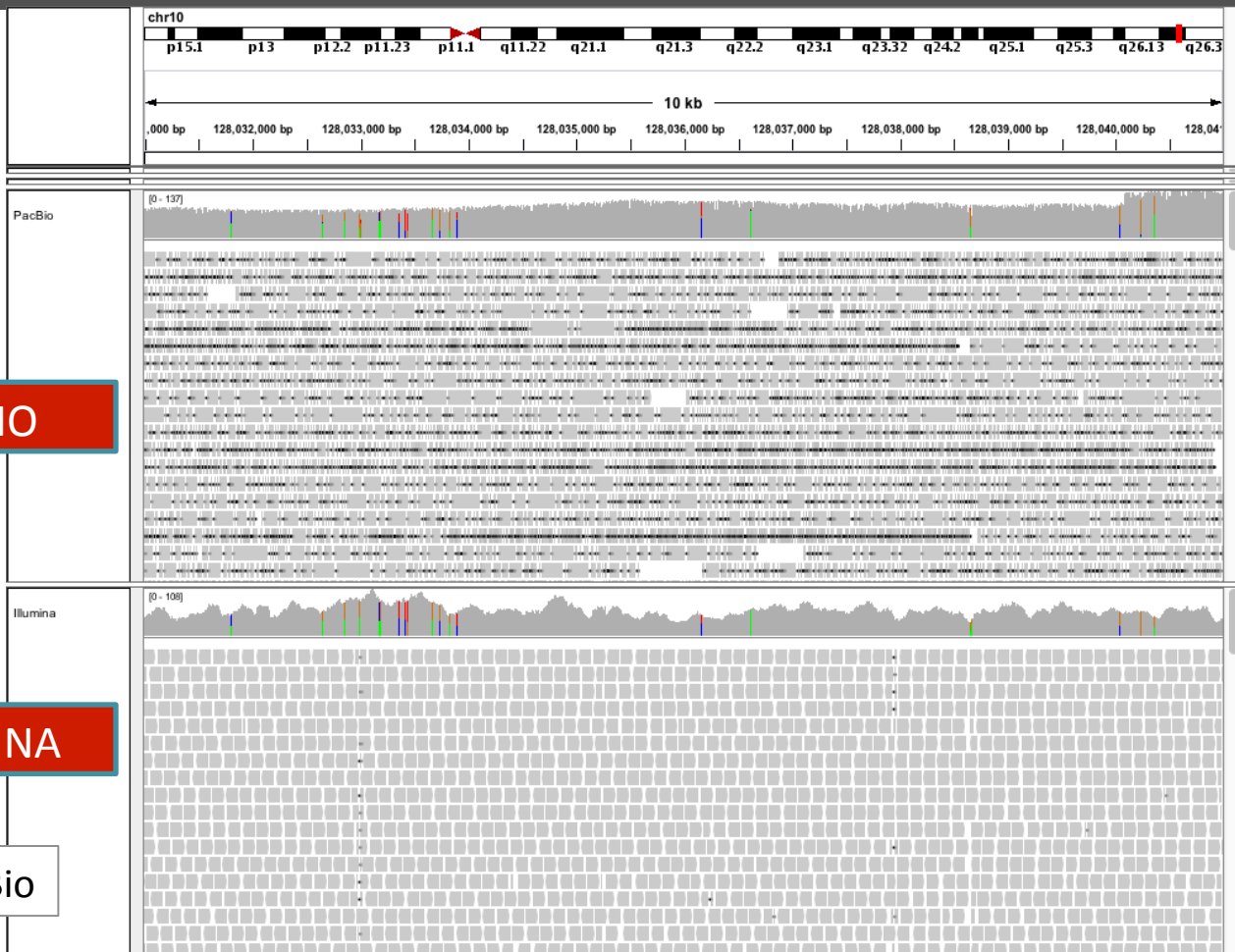
- Filtering raw errors to reveal consensus variants
- Extended support for split alignments
 - Linking split reads
 - Support for “SA” tag
- Support for barcoded alignments (10X, Illumina Tru-seq)
- Improved performance for wide range views (100kb - 500kb)

Filtering raw errors – small indels

PACBIO

ILLUMINA

Aaron Wenger, PacBio

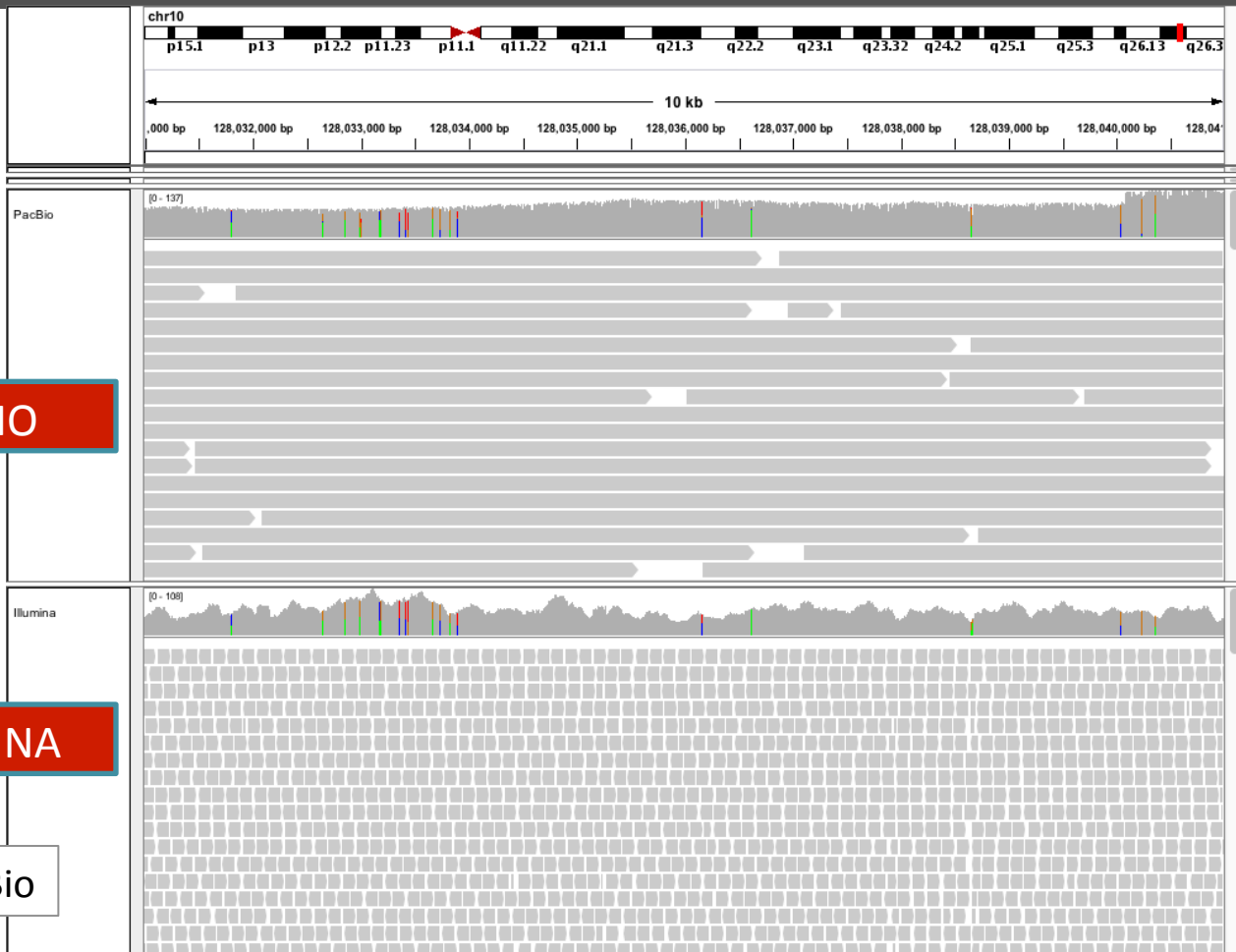


Filtering raw errors - indels

PACBIO

ILLUMINA

Aaron Wenger, PacBio



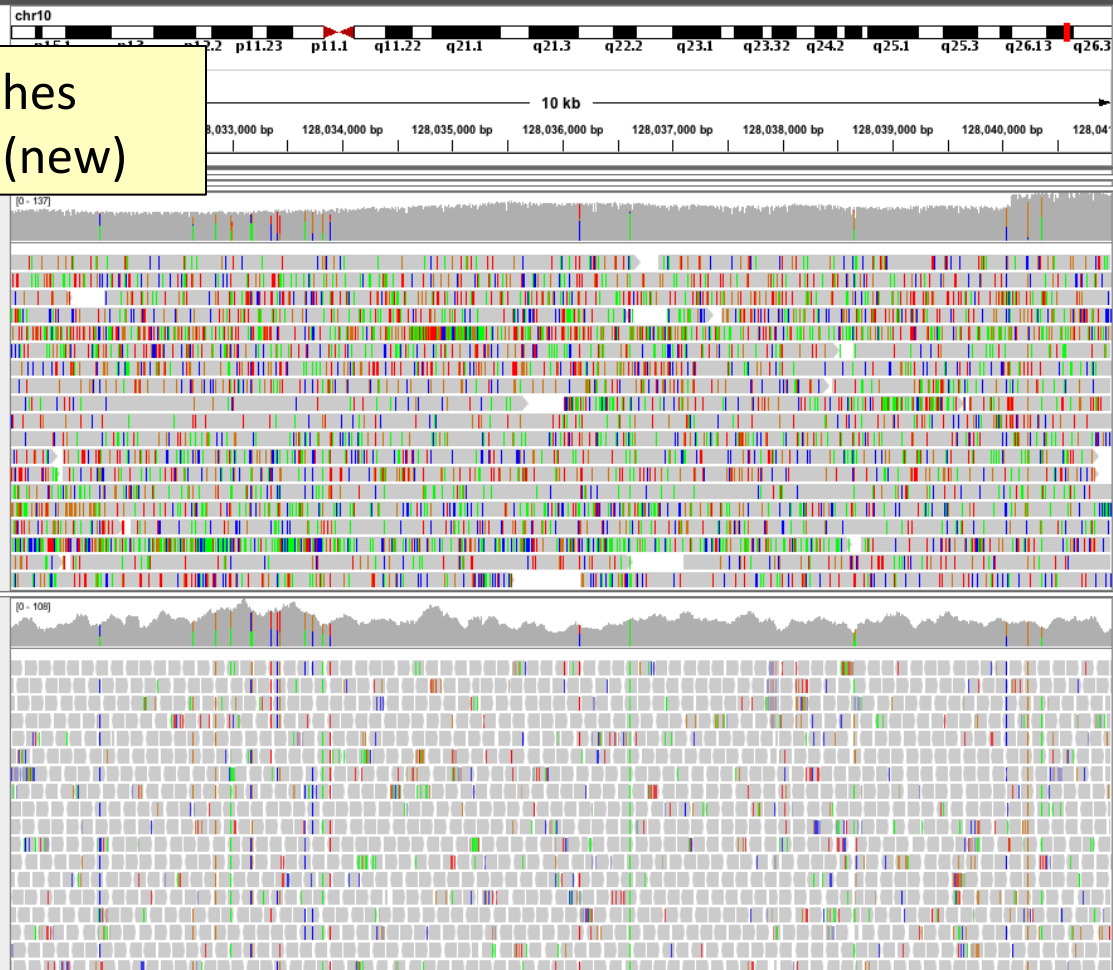
Filtering raw errors - nucleotides

Nucleotide mismatches
shown at low zoom (new)

PACBIO

ILLUMINA

Aaron Wenger, PacBio



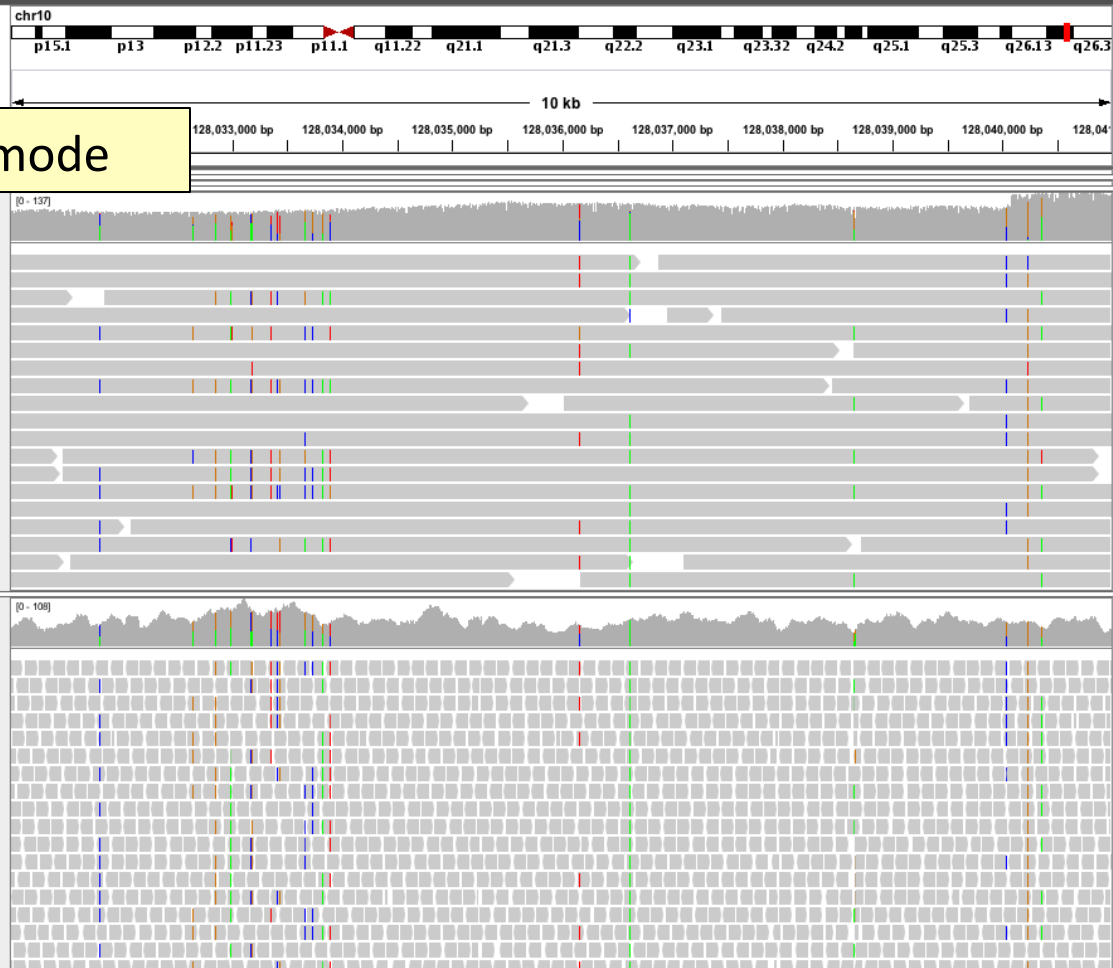
Filtering raw errors - nucleotides

“Quick consensus” mode

PACBIO

ILLUMINA

Aaron Wenger, PacBio

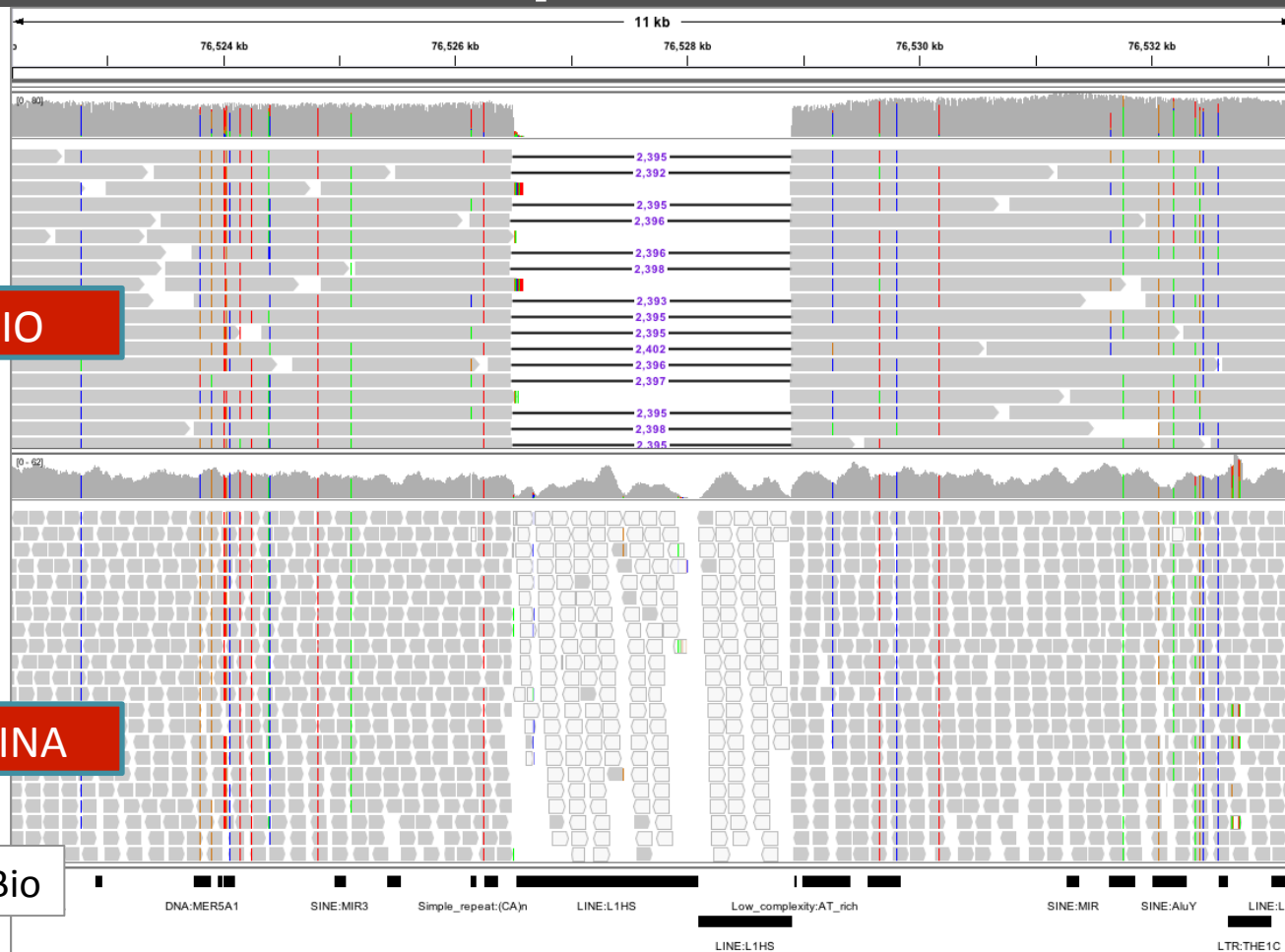


Structural variant example - deletion

PACBIO

ILLUMINA

Aaron Wenger, PacBio



3rd gen support enabled in preferences

View > Preferences
Alignments tab

Purple insertion labels

Hide small indels

Only show consistent mismatches

The screenshot shows the 'Alignments' tab in a preferences window. The 'Alignments' tab is highlighted with a red circle. The 'Track Display Options' section has 'Alignment Track' and 'Coverage Track' checked, and 'Splice Junction Track' unchecked. The 'Alignment Track Options' section includes: 'Visibility range threshold (kb): 500', 'Downsample reads' checked with 'Max read count: 2000' and 'per window size (bases): 50', 'Shade mismatched bases by quality: 5 to 20', 'Mapping quality threshold: 20', 'Label indels > 5 bases' checked, 'Hide indels < 50 bases' checked, 'Hide indels < 1 pixels' unchecked, 'Filter duplicate reads' unchecked, 'Filter vendor failed reads' checked, 'Filter secondary alignments' checked, 'Show center line' unchecked, 'Flag unmapped pairs' unchecked, 'Show soft-clipped bases' unchecked, 'Quick consensus mode' checked, and 'Filter supplementary alignments' checked. The 'Hidden SAM tags' field contains 'MD,SA,XA'. The 'Coverage Track Options' section has 'Coverage allele-fraction threshold: 0.25' and 'Quality weight allele fraction' unchecked. The 'Splice Junction Track Options' section is partially visible at the bottom.

General Tracks Variants Charts **Alignments** Probes Proxy IonTorrent Advanced

Track Display Options

On initial load show: ☒ Alignment Track ☒ Coverage Track ☐ Splice Junction Track

Alignment Track Options

Visibility range threshold (kb): 500 *Range at which alignments become visible*

☒ Downsample reads Max read count: 2000 per window size (bases): 50

☒ Shade mismatched bases by quality: 5 to 20

Mapping quality threshold: 20

☒ Label indels > 5 bases

☒ Hide indels < 50 bases

☐ Hide indels < 1 pixels

☐ Filter duplicate reads

☒ Filter vendor failed reads

☒ Filter secondary alignments

☐ Show center line

☐ Flag unmapped pairs

☐ Show soft-clipped bases

☒ Quick consensus mode

☒ Filter supplementary alignments

Hidden SAM tags: MD,SA,XA

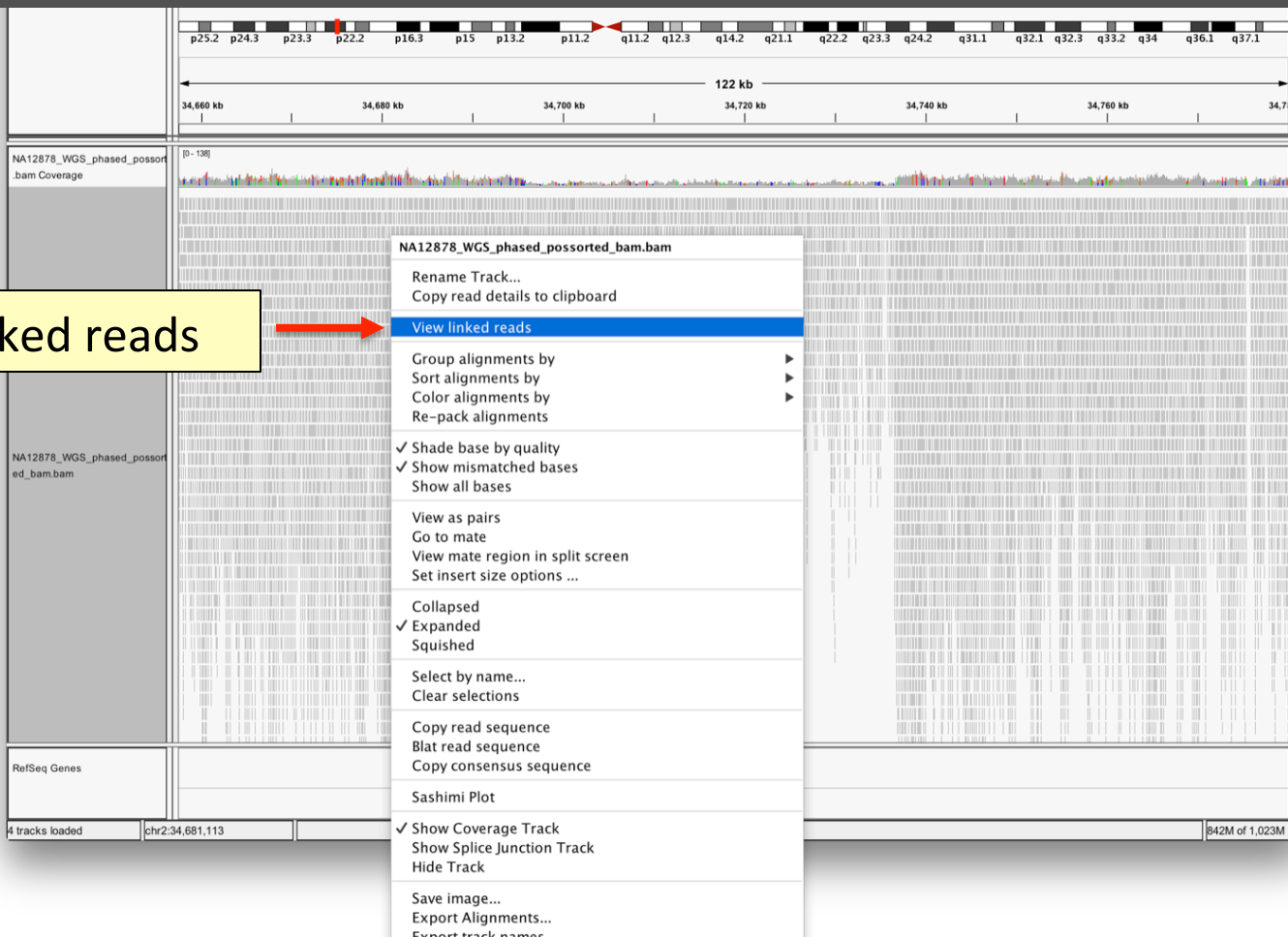
Coverage Track Options

Coverage allele-fraction threshold: 0.25 ☐ Quality weight allele fraction

Splice Junction Track Options

10X

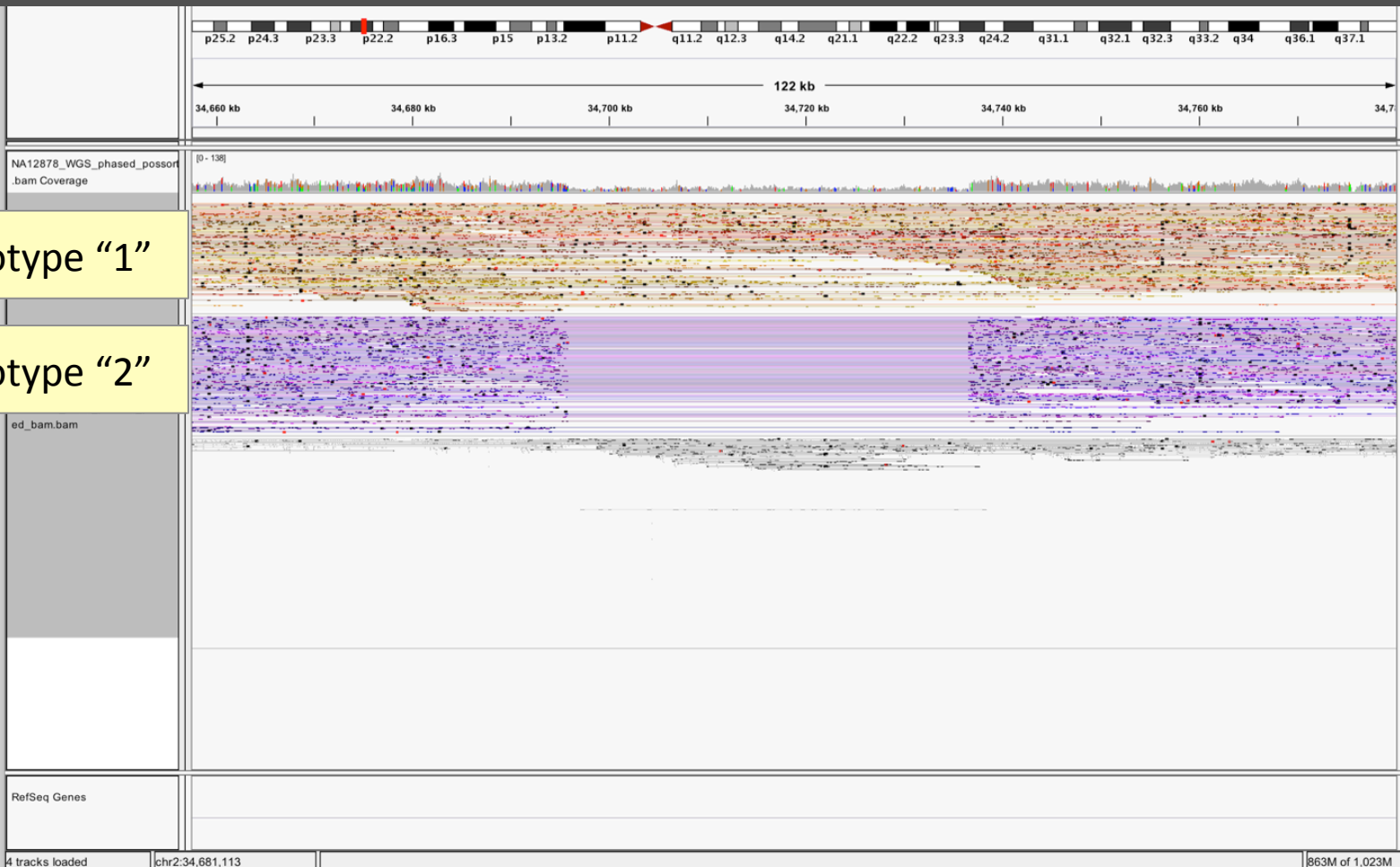
View linked reads



10X

Haplotype "1"

Haplotype "2"



Availability

- Features available now in development version of IGV
broadinstitute.org/igv/download_snapshot

Viewing Variants

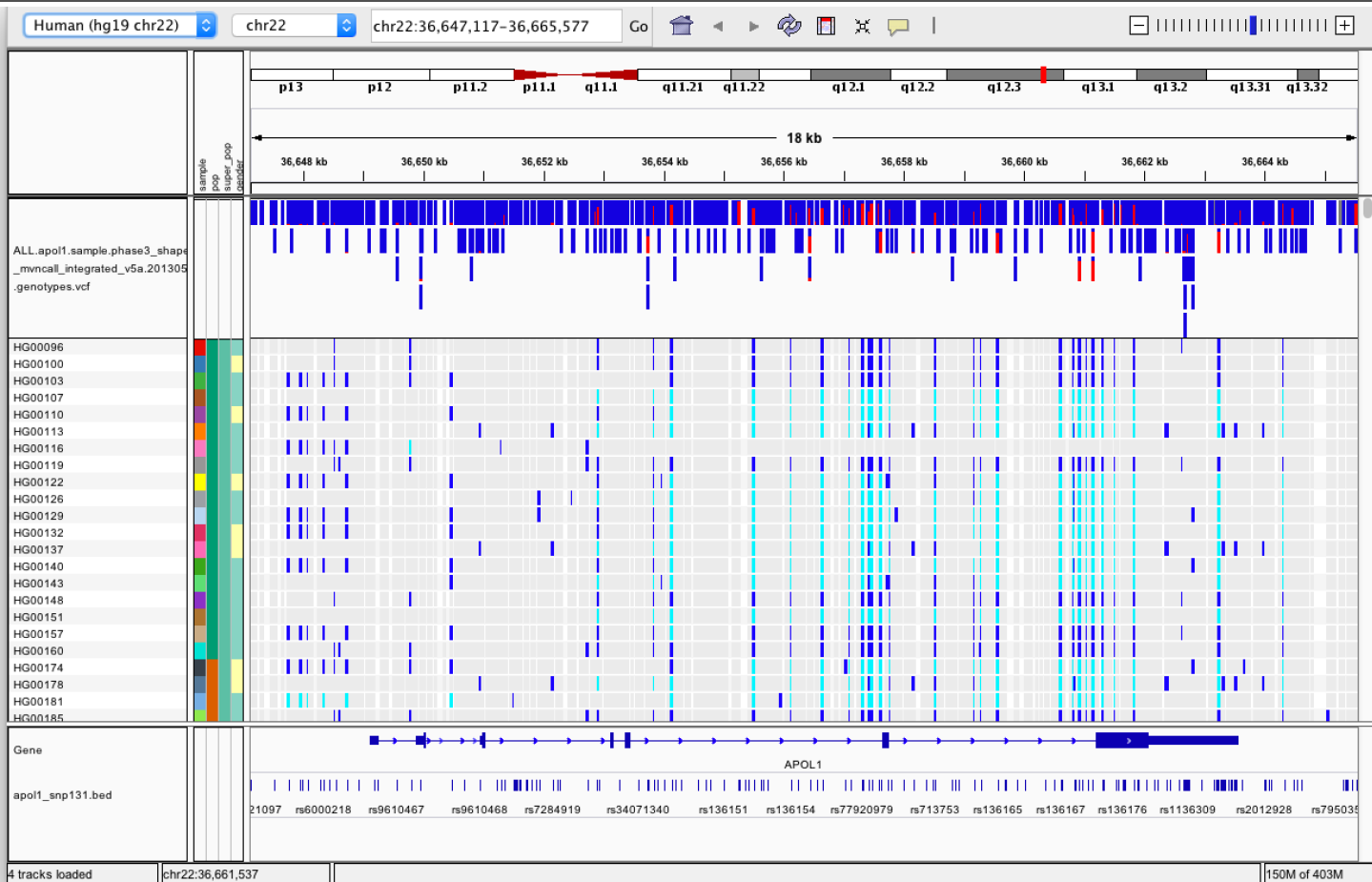
Viewing variants (VCF files)

Variant Call Format (VCF):

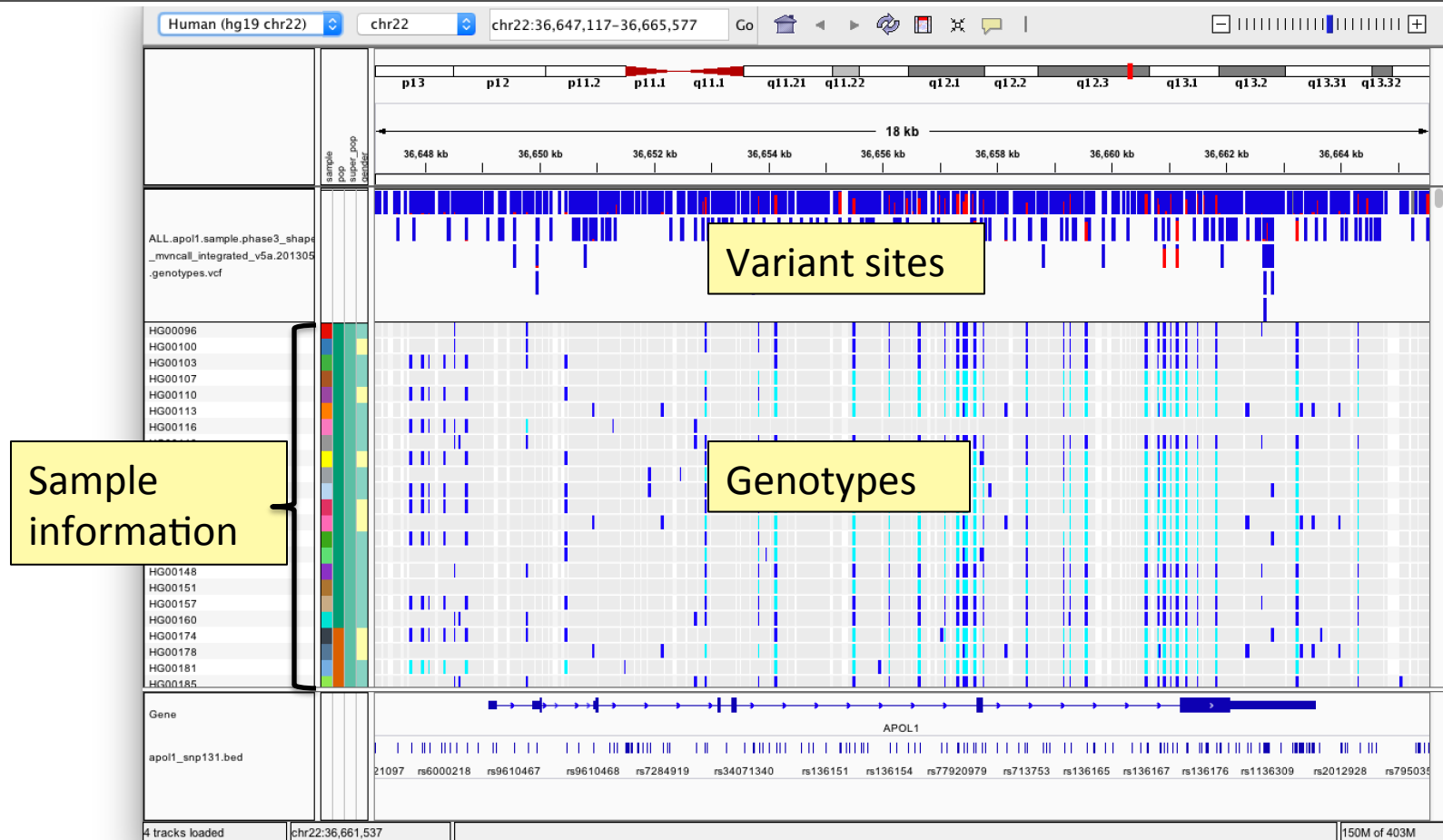
File format for encoding variant sites and genotypes

<http://www.internationalgenome.org/wiki/Analysis/variant-call-format>

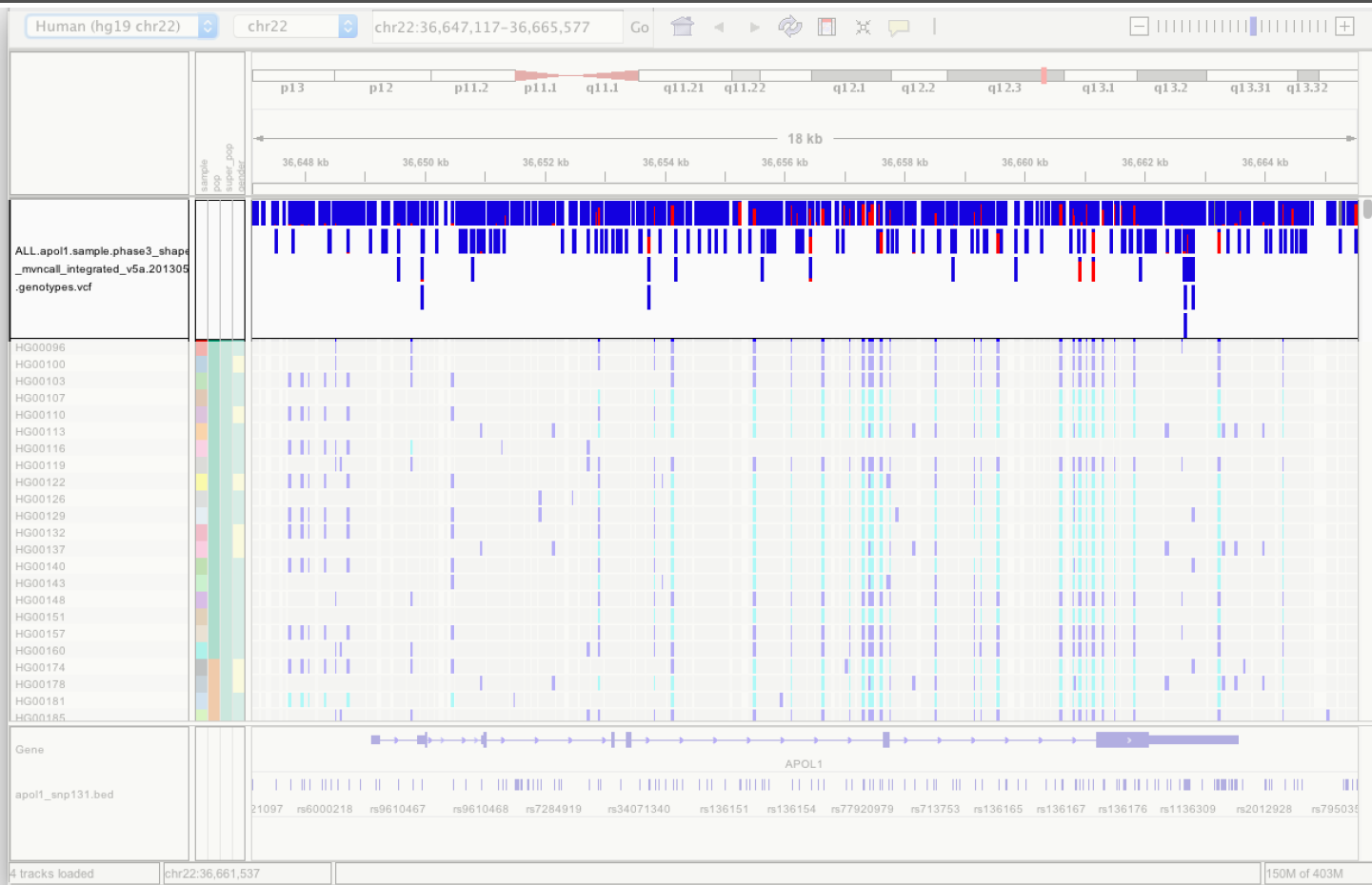
Viewing variants (VCF files)



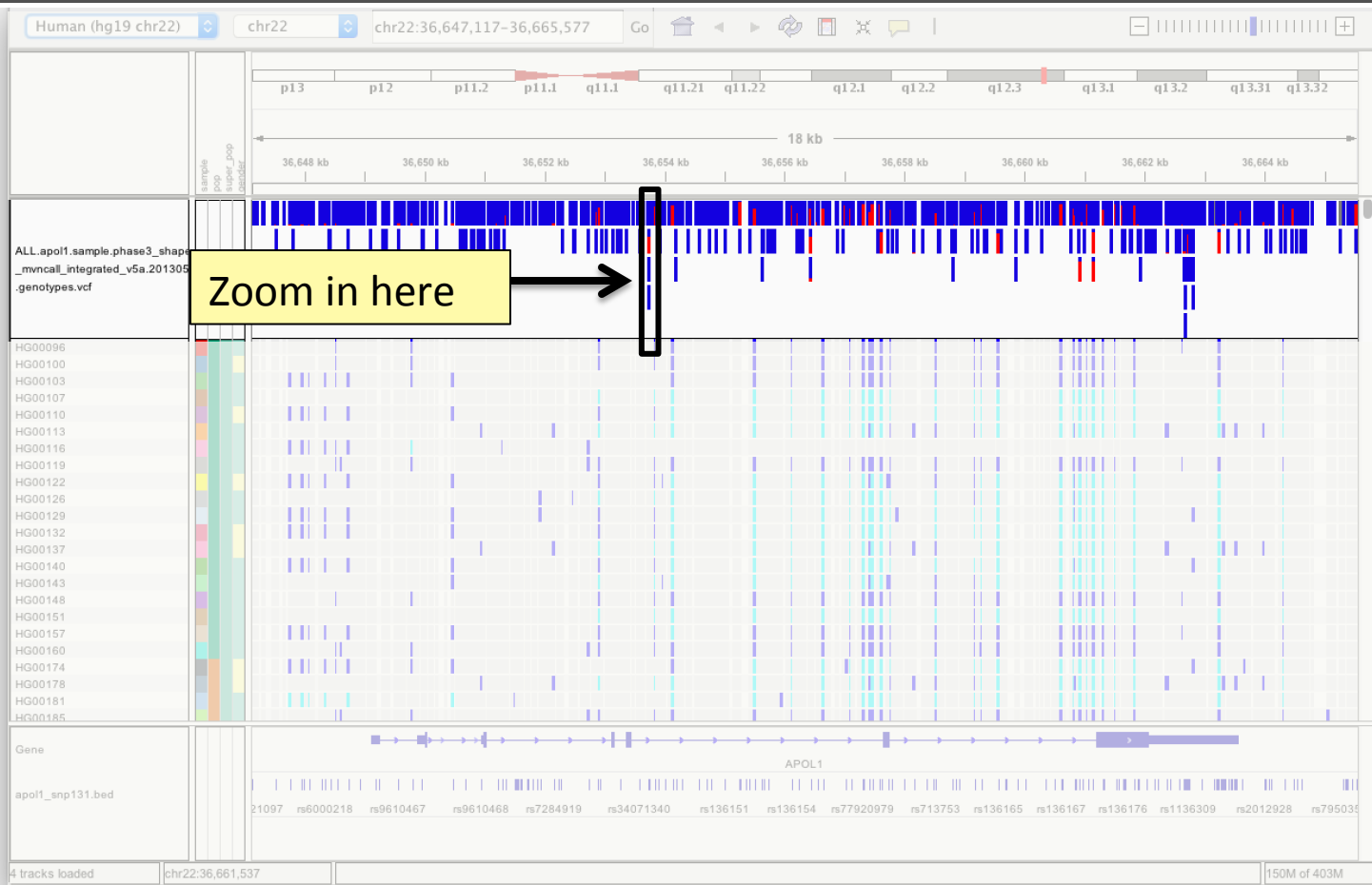
Viewing variants (VCF files)



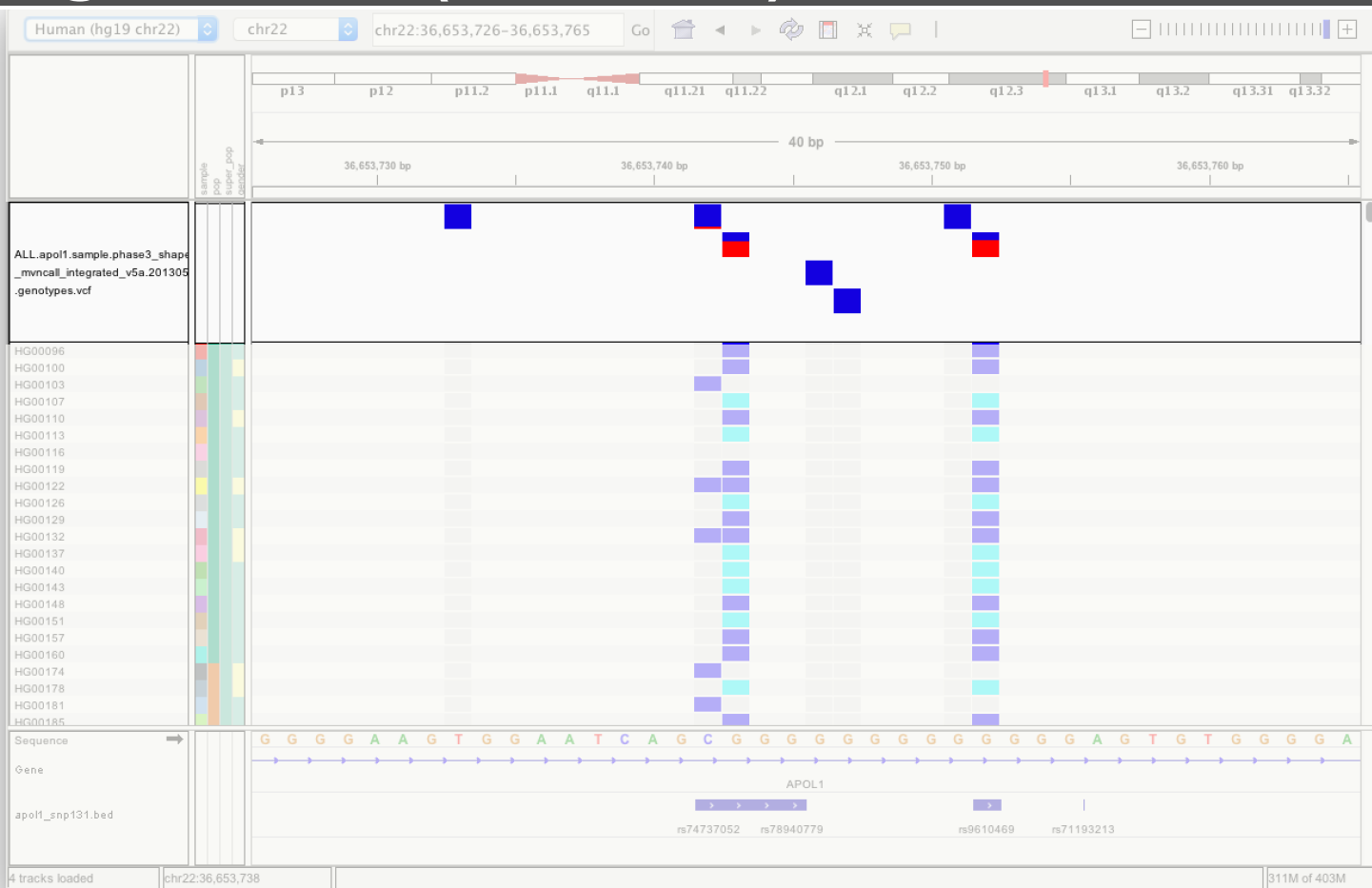
Viewing variants (VCF files)



Viewing variants (VCF files)



Viewing variants (VCF files)

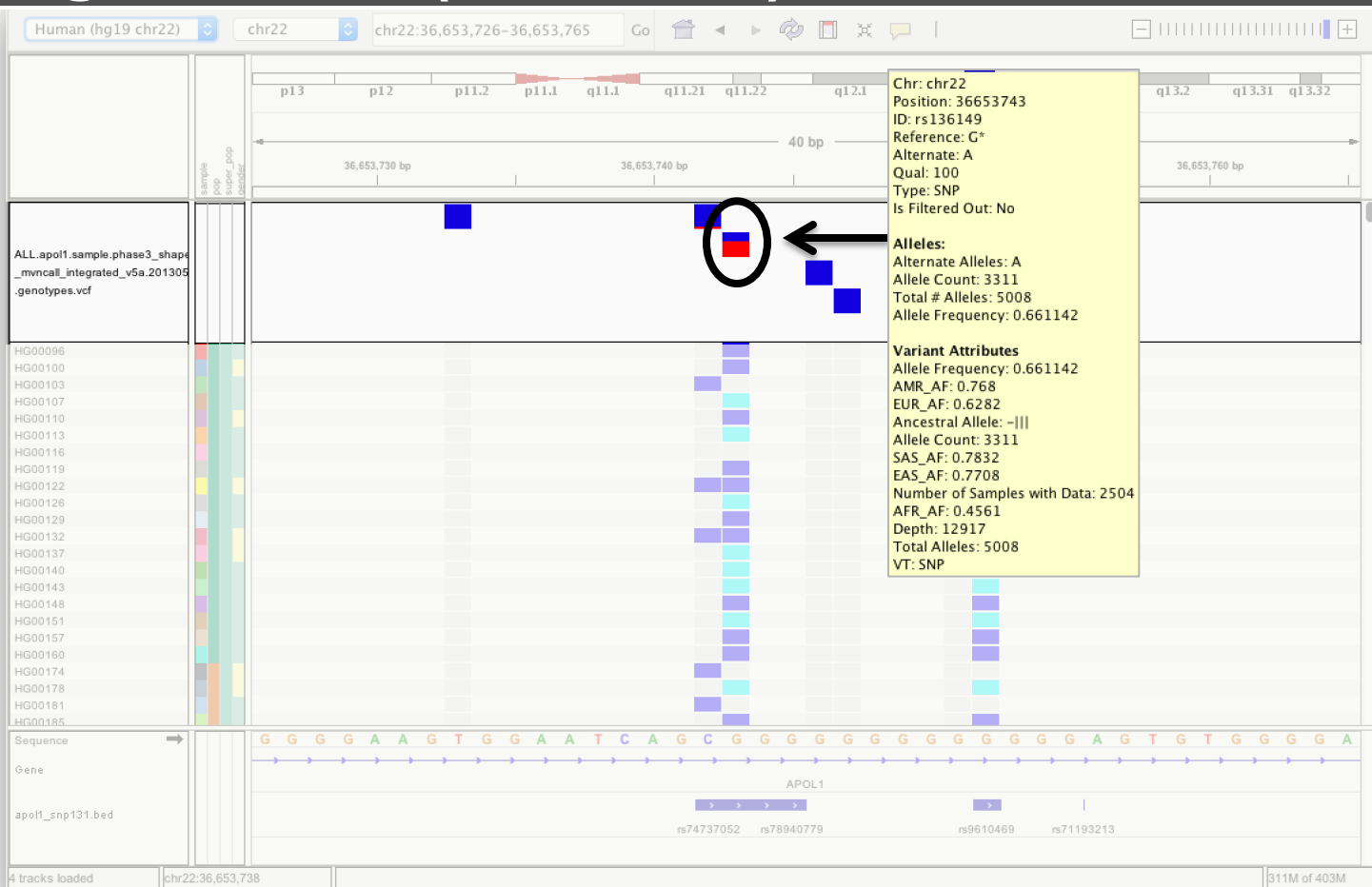


Viewing variants (VCF files)

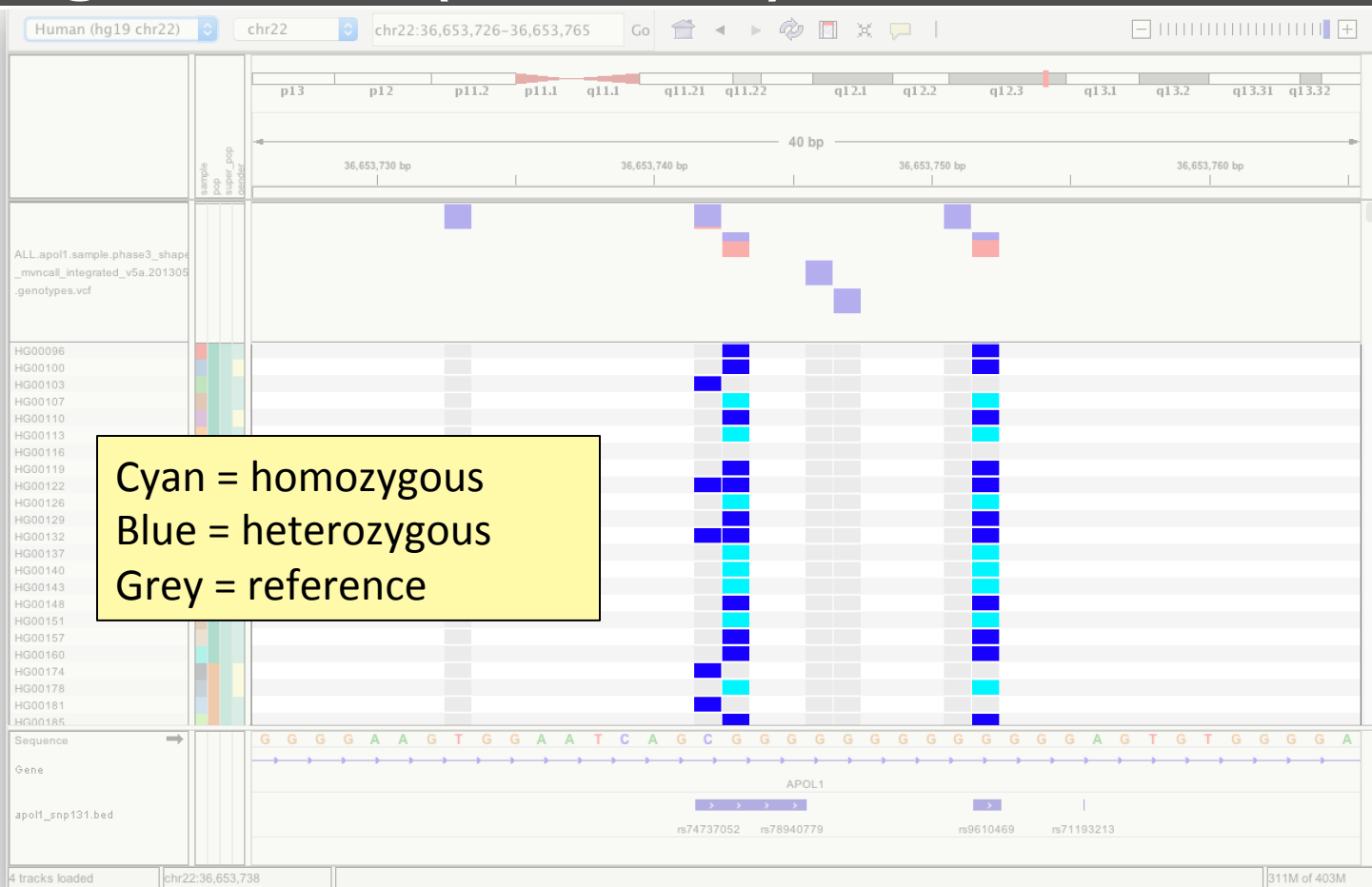
Red = alternate allele
Blue = reference allele



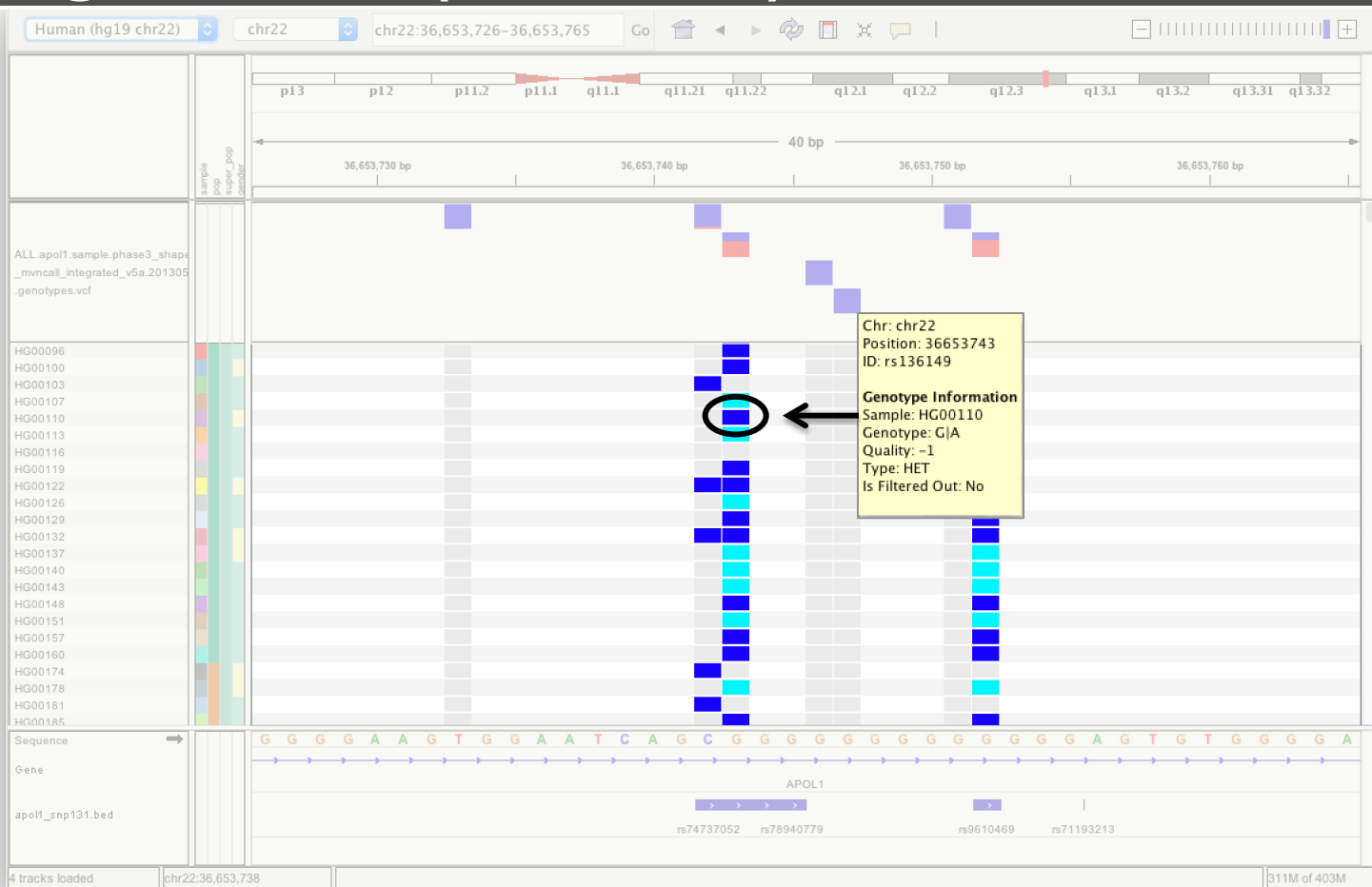
Viewing variants (VCF files)



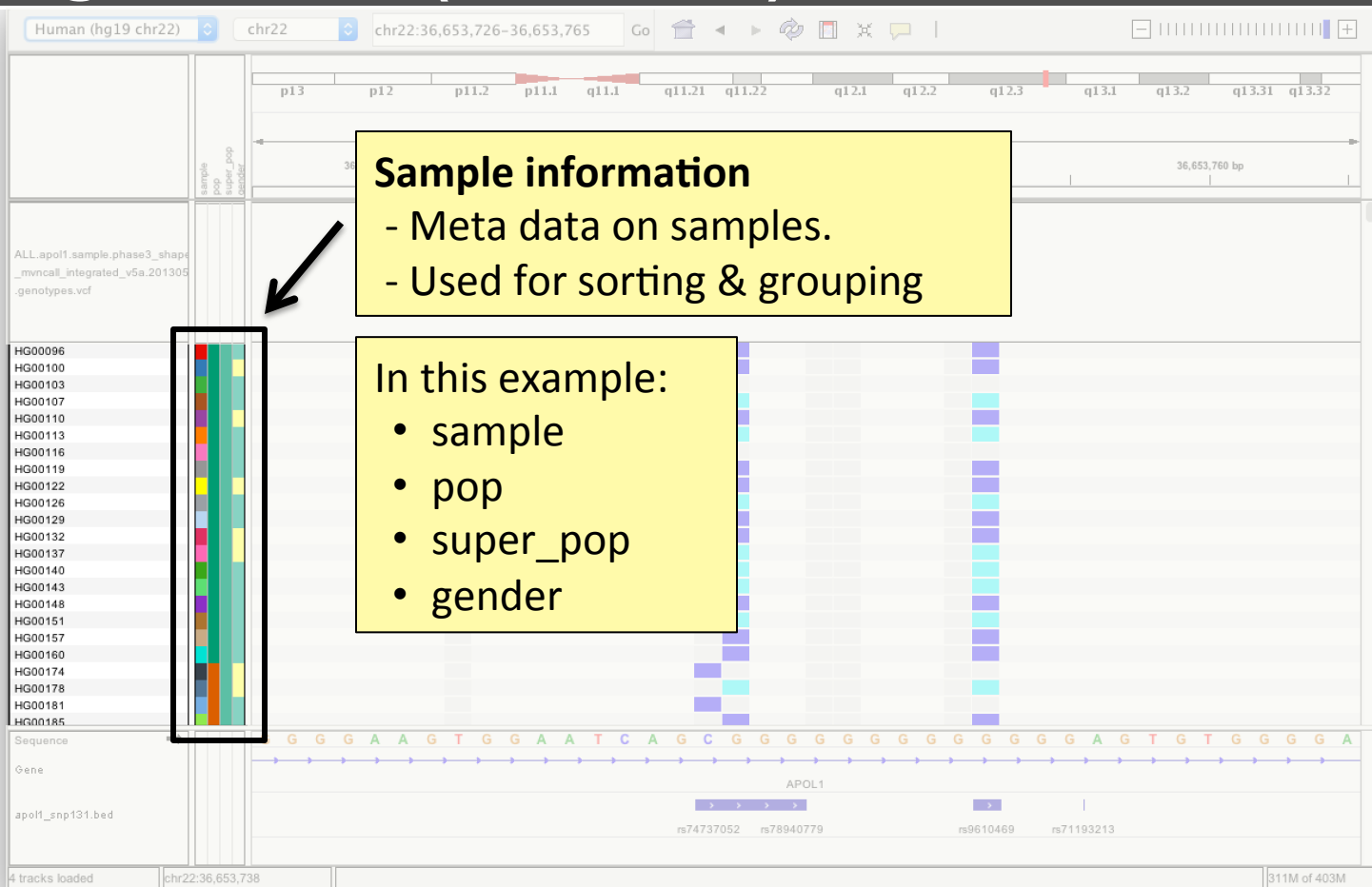
Viewing variants (VCF files)



Viewing variants (VCF files)



Viewing variants (VCF files)



Sample information

Sample information is defined in an auxiliary tab-delimited file.

- First **row** defines column names
- First **column** contains sample names (must match VCF sample names)

Example:

sample	pop	super_pop	gender
HG00096	GBR	EUR	male
HG00097	GBR	EUR	female
HG00099	GBR	EUR	female
HG00100	GBR	EUR	female
HG00101	GBR	EUR	male
HG00102	GBR	EUR	female
HG00103	GBR	EUR	male
HG00105	GBR	EUR	male

Hands on: Viewing variants (VCF file)

Use sample information to examine a variant that segregates by population.

Reference:

Giulio Genovese et al.

Association of Trypanolytic ApoL1 Variants with Kidney Disease in African Americans,

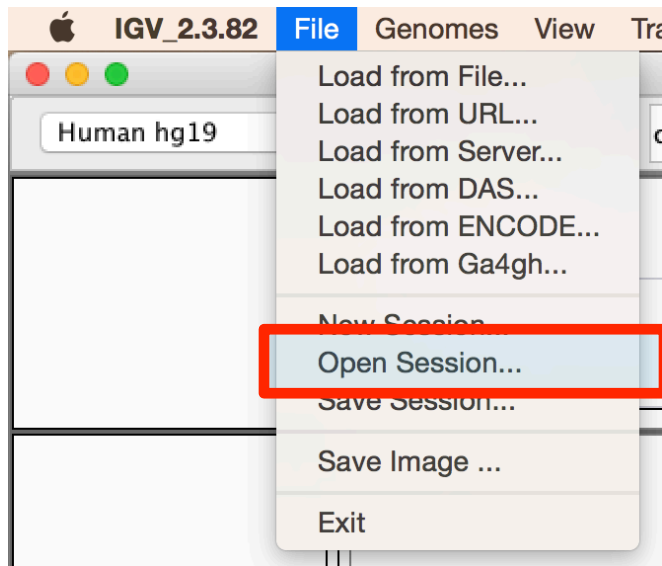
Science 329, 841 (2010); DOI: 10.1126/science.1193032

Viewing variants: www.igv.org/workshops/ASHG2016/ExInstr.pdf

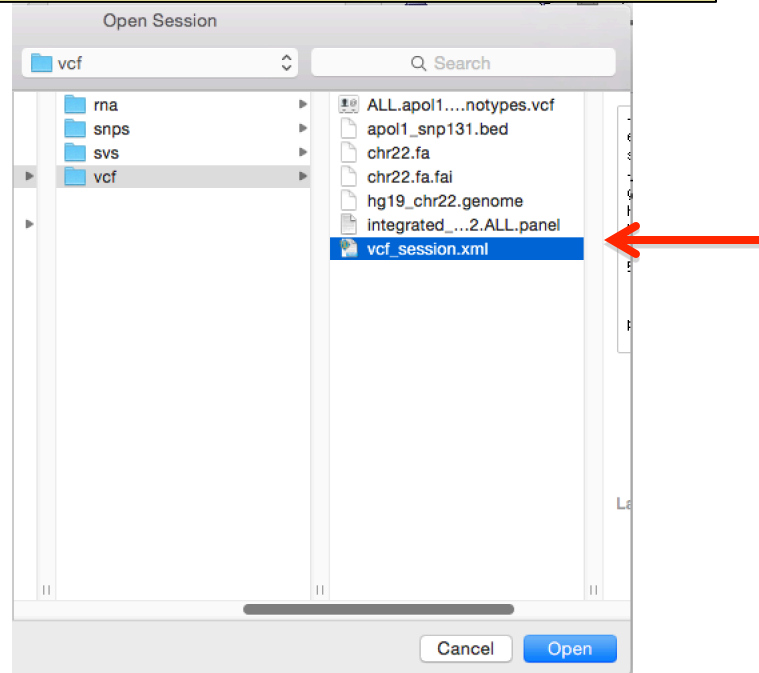
- 1 Initialize exercise:** – Click **File > Open Session**. Go to the folder where you downloaded the workshop data and select **vcf / vcf_session.xml**.
- 2** Observe the two data panels: Variant sites (upper panel) and Genotypes (lower panel).
- 3**
 - Type APOL1:S342G in the search box and click **Go**.
 - Right-click over genotypes and select **Display Mode > Squished**.
 - Right-click over genotypes and select **Group By**, and select attribute **super_pop**.
 - Scroll down to see all the groups
- 4 Observe differences between groups:** Variants not present in some population groups and prevalent in others.

Hands on: Viewing variants (VCF file)

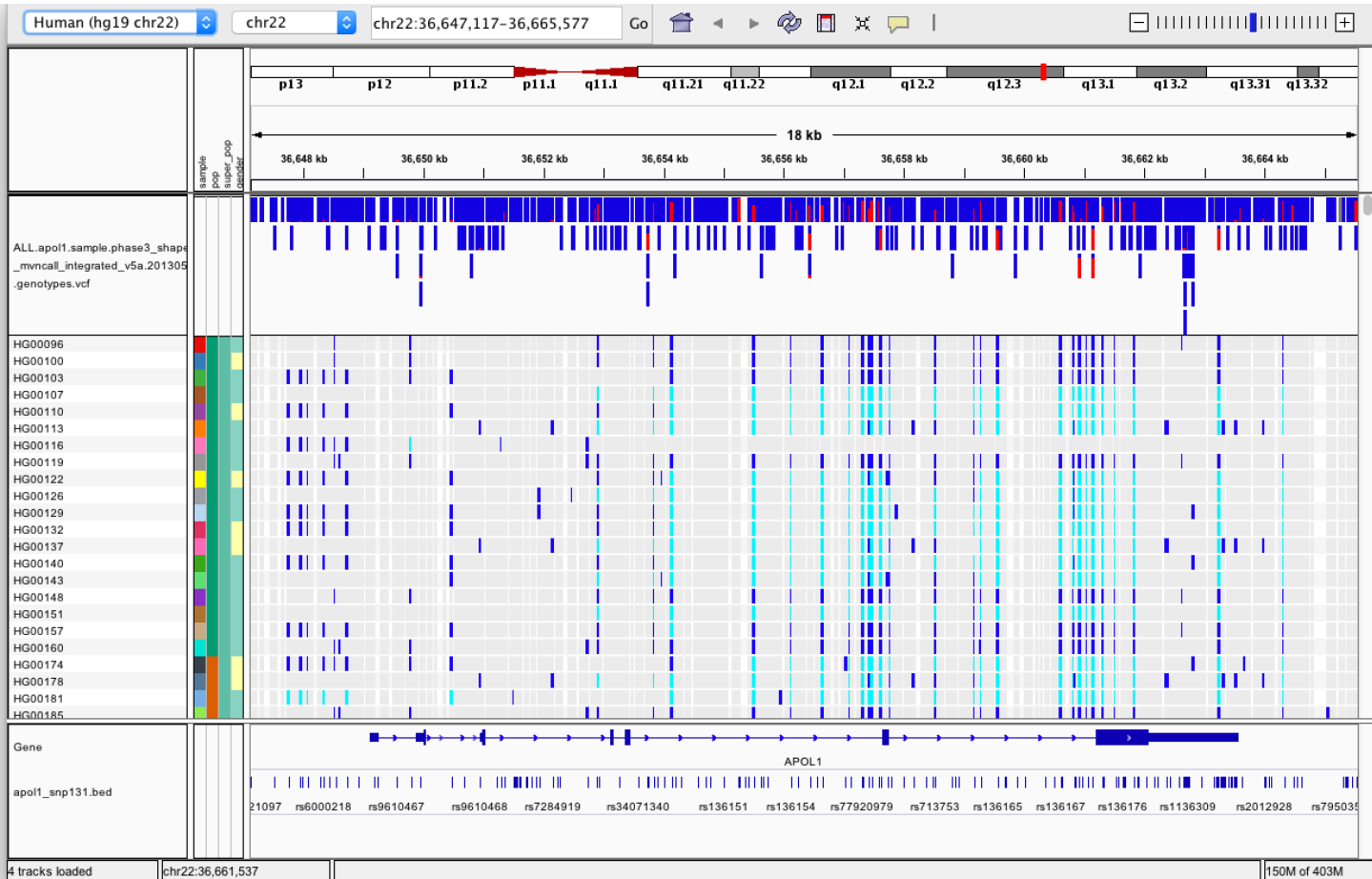
Click **File > Open Session**



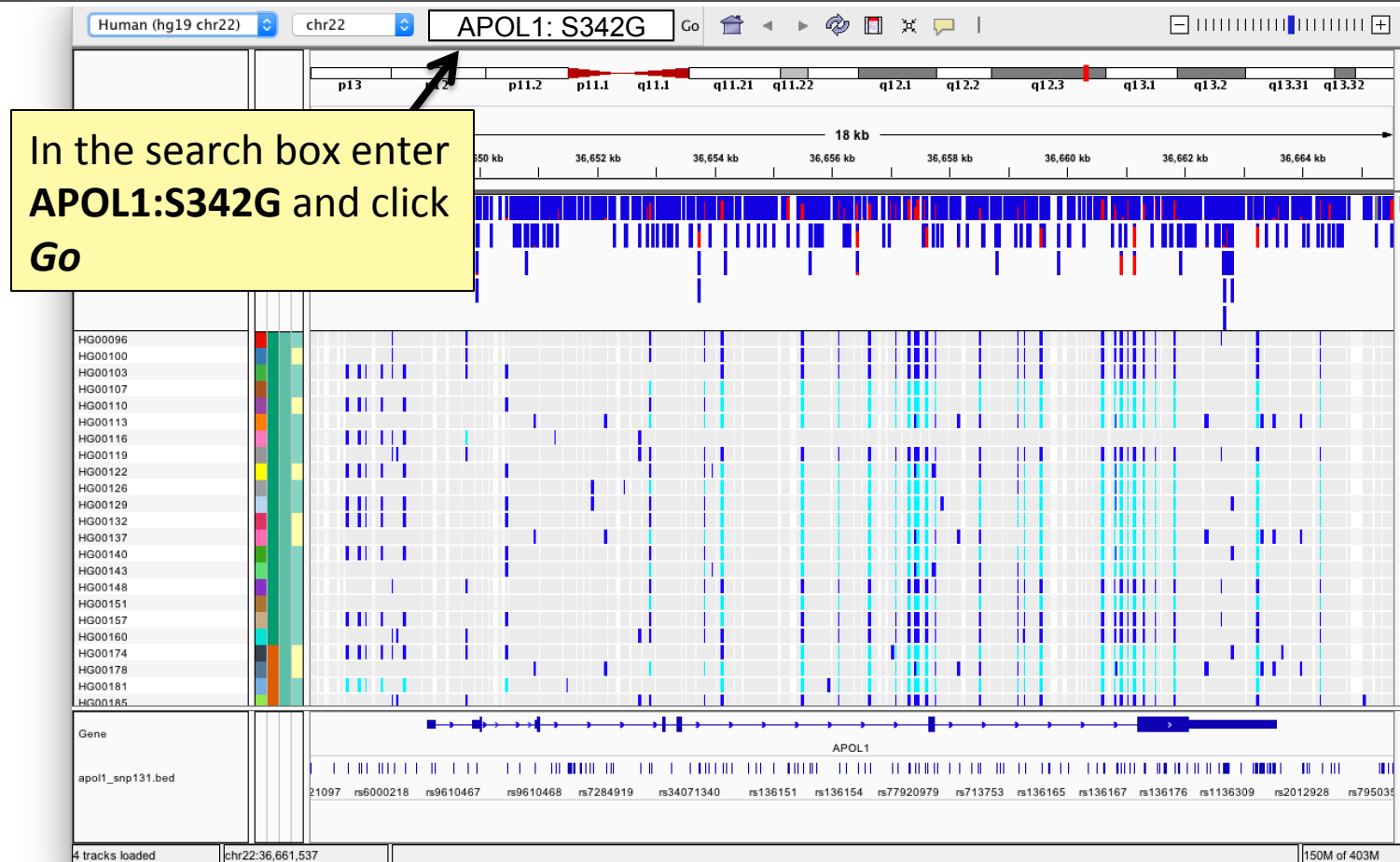
Navigate to the folder where you downloaded the workshop data, and select **vcf / vcf_session.xml**



Hands on: Viewing variants (VCF file)



Hands on: Viewing variants (VCF file)



Hands on: Viewing variants (VCF file)



Hands on: Viewing variants (VCF file)

Human (hg19 chr22) chr22 chr22:36,661,887-36,661,927 Go

41 bp

36,661,890 bp 36,661,900 bp 36,661,910 bp 36,661,920 bp

ALL.apol1.sample.phase3_shape2_mvncall_integrated_v5a.20130502.genotypes.vcf

HG00096
HG00100
HG00103
HG00107
HG00110

Right-click over genotypes and select **Display Mode > Squished**

ALL.apol1.sample.phase3_shape2_mvncall_integrated_v5a.20130502.genotypes.vcf

- Rename Track...
- Change Font Size...
- Color By
 - ✓ Allele Frequency
 - Allele Fraction
- Sort By Genotype
- Sort By Sample Name
- Sort By Depth
- Sort By Quality
- Group By...
- Display Mode
 - Collapsed
 - Squished**
 - Expanded
- Change Squished Row Height...
- Suppress Filtered Sites
- Set Feature Visibility Window...
- Save image...
- Export Features...
- Export track names...
- Remove Track

Sequence →

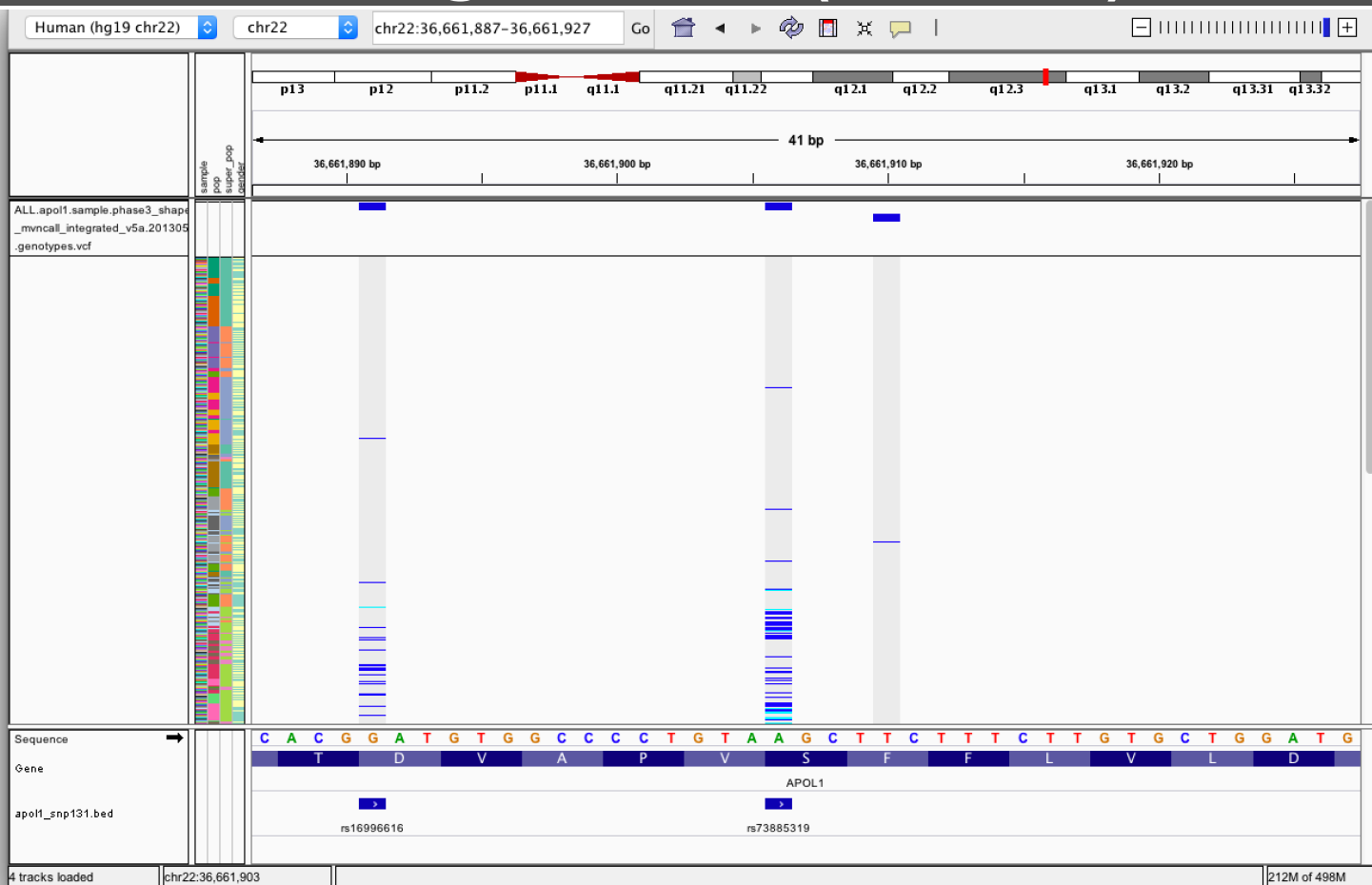
Gene

apol1_snp131.bed

rs16996616

4 tracks loaded chr22:36,661,907 197M of 498M

Hands on: Viewing variants (VCF file)



Hands on: Viewing variants (VCF file)

The screenshot displays a genomic browser interface. At the top, the browser is set to Human (hg19 chr22) and shows a specific region on chromosome 22: chr22:36,661,887-36,661,927. The main view includes a cytogenetic band map (p13 to q13.32) and a scale bar indicating a 41 bp region. Below this, a track for 'ALL.apo1.sample.phase3_shapeit2_mvncall_integrated_v5a.20130502.genotypes.vcf' is visible. A right-click context menu is open over the genotypes track, showing options such as 'Rename Track...', 'Change Font Size...', 'Color By' (with 'Allele Frequency' and 'Allele Fraction' selected), 'Sort By' (with 'Sort By Genotype' selected), 'Group By...', 'Display Mode' (with 'Collapsed' and 'Squished' selected), 'Change Squished Row Height...', 'Suppress Filtered Sites', 'Set Feature Visibility Window...', 'Save image...', 'Export Features...', 'Export track names...', and 'Remove Track'. A yellow callout box with the text 'Right-click over genotypes and select **Group By**' has an arrow pointing to the 'Group By...' option in the menu. The bottom of the browser shows a sequence track with the sequence 'C A C G G A T G T G G C C C C T G' and a gene track with the gene 'apo1_snp131.bed'.

Human (hg19 chr22) chr22 chr22:36,661,887-36,661,927 Go

p13 p12 p11.2 p11.1 q11.1 q11.21 q11.22 q12.1 q12.2 q12.3 q13.1 q13.2 q13.31 q13.32

36,661,890 bp 36,661,900 bp 36,661,910 bp 36,661,920 bp

41 bp

ALL.apo1.sample.phase3_shapeit2_mvncall_integrated_v5a.20130502.genotypes.vcf

Right-click over genotypes and select **Group By**

ALL.apo1.sample.phase3_shapeit2_mvncall_integrated_v5a.20130502.genotypes.vcf

- Rename Track...
- Change Font Size...
- Color By
 - ✓ Allele Frequency
 - Allele Fraction
- Sort By Genotype
- Sort By Sample Name
- Sort By Depth
- Sort By Quality
- Group By...
- Display Mode
 - Collapsed
 - ✓ Squished
 - Expanded
- Change Squished Row Height...
- Suppress Filtered Sites
- Set Feature Visibility Window...
- Save image...
- Export Features...
- Export track names...
- Remove Track

Sequence → C A C G G A T G T G G C C C C T G

Gene T D V A P

apo1_snp131.bed

rs16996616

4 tracks loaded chr22:36,661,903 212M of 498M

Hands on: Viewing variants (VCF file)

The screenshot displays a genomic browser interface for Human (hg19 chr22). The top navigation bar shows the chromosome (chr22) and a specific genomic region (chr22:36,661,887-36,661,927). Below the navigation bar, a track labeled 'sample pop super_pop gender' is visible. A 'Group By Attribute' dialog box is open, showing a list of attributes: 'None', 'sample', 'pop', 'super_pop', and 'gender'. The 'super_pop' attribute is selected, indicated by a blue highlight and a checkmark. A yellow callout box with the text 'Select attribute super_pop' and an arrow points to the 'super_pop' option in the dialog. The main view shows a genomic track with a 41 bp scale bar and a sequence alignment track. The sequence alignment track shows the sequence 'C A C G G A T G T G G C C C C T G T A A G C T T C T T G T G C T G G A T G' and the gene 'APOL1'. The track also shows variant positions (rs16996616 and rs73885319) and a track labeled 'apoli_snp131.bed'.

Human (hg19 chr22) chr22 chr22:36,661,887-36,661,927 Go

sample pop super_pop gender

41 bp

36,661,890 bp 36,661,900 bp 36,661,910 bp 36,661,920 bp

ALL.apo1f.sample.phase3_shape_mvncai_integrated_v5a.201305.genotypes.vcf

Group By Attribute

Group tracks by:

- ✓ None
- sample
- pop
- super_pop
- gender

Select attribute **super_pop**

Sequence →

Gene

apoli_snp131.bed

C A C G G A T G T G G C C C C T G T A A G C T T C T T G T G C T G G A T G

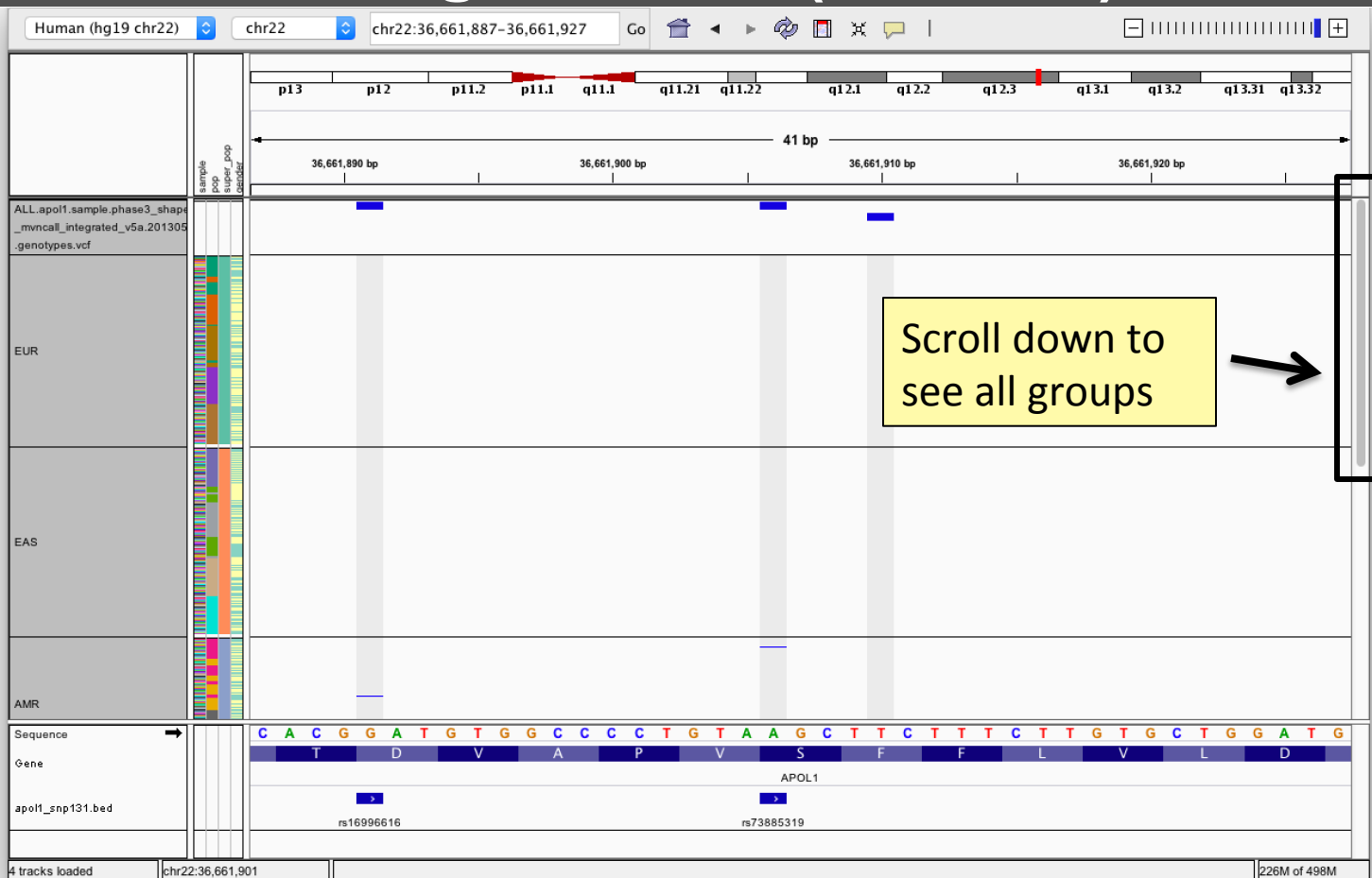
T D V A P V S F F L V L D

APOL1

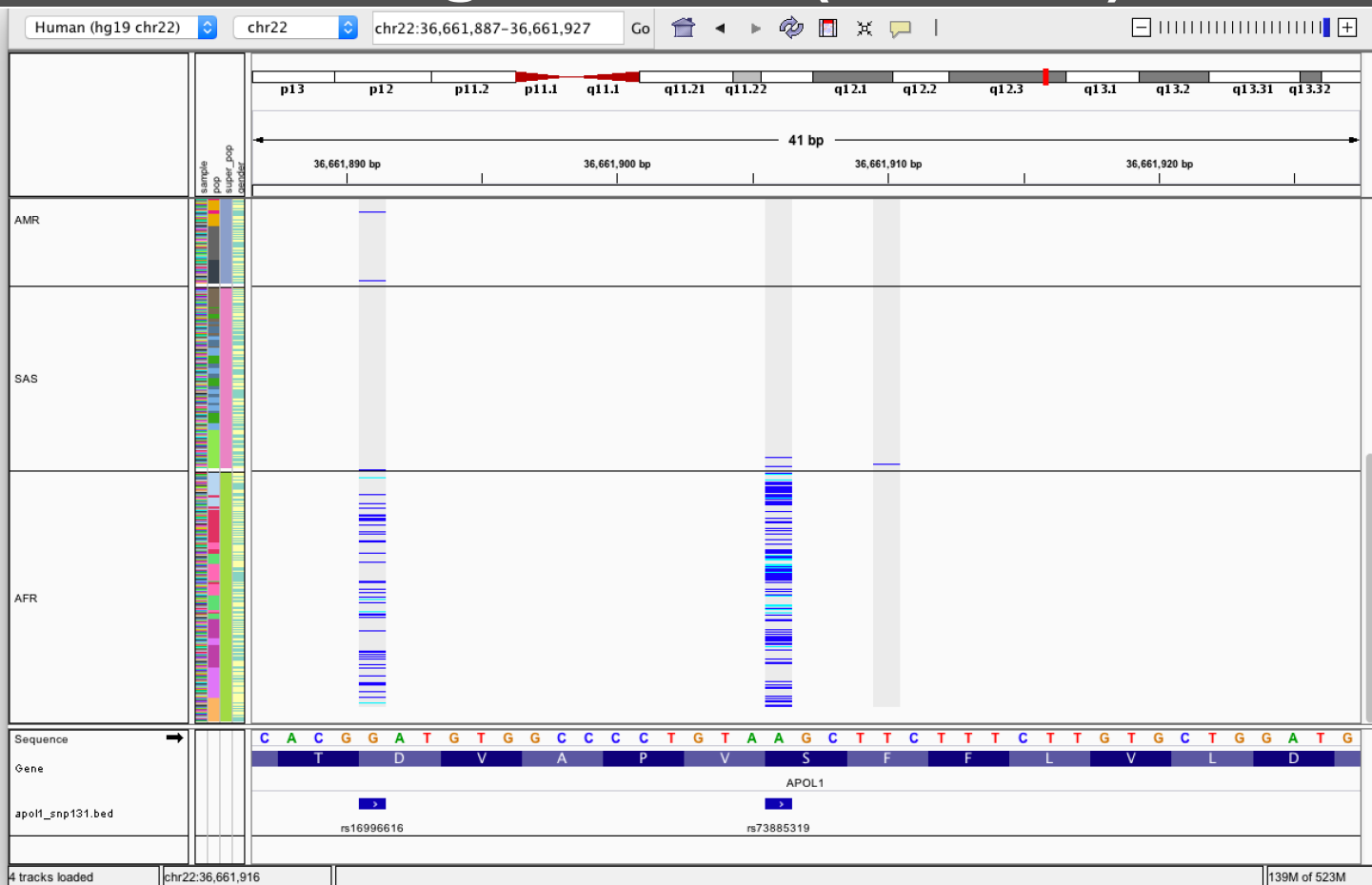
rs16996616 rs73885319

4 tracks loaded chr22:36,661,903 212M of 498M

Hands on: Viewing variants (VCF file)



Hands on: Viewing variants (VCF file)



Tools

Tools

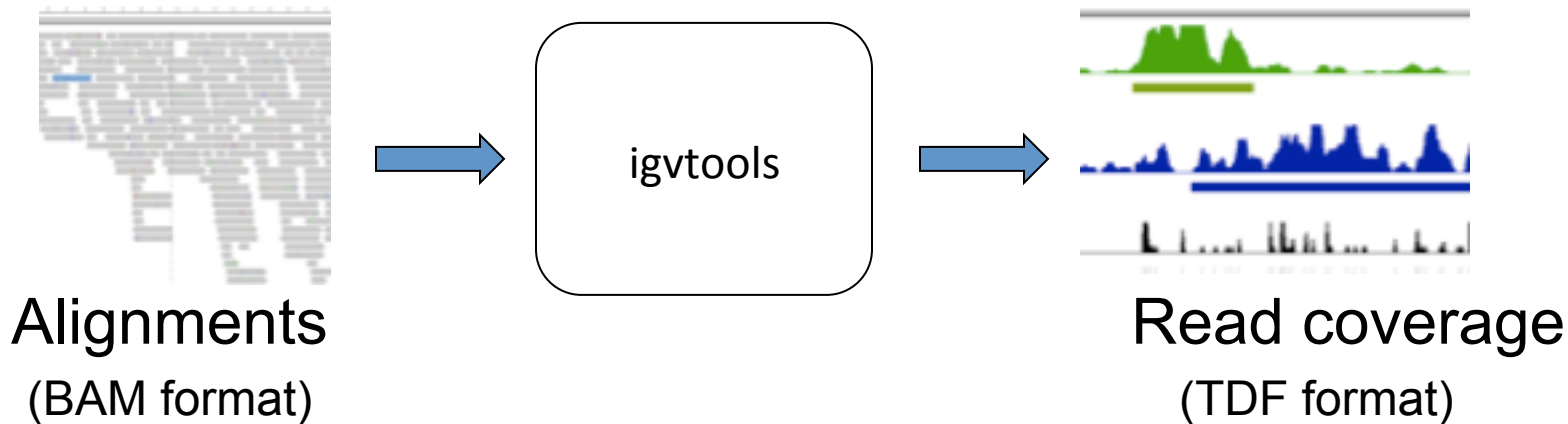
- Pre-processing data with ***igvtools***
- Combining numeric data tracks
- BLAT

A set of utilities for preparing large files for efficient display.

count	– Computes alignment coverage from BAM files – Produces TDF or WIG files
toTDF	– Converts sorted data file to binary tiled data (TDF) – Supported file formats: WIG, bedGraph
sort	– Sorts file by genomic start position. – Supported file formats: BED, GFF, GTF, PSL, VCF
index	– Creates index for large genomic annotation files. – Supported file formats: BED, GFF, GTF, PSL, VCF

igvtools: count

count transforms alignment files to read coverage files in TDF or WIG format.



igvtools

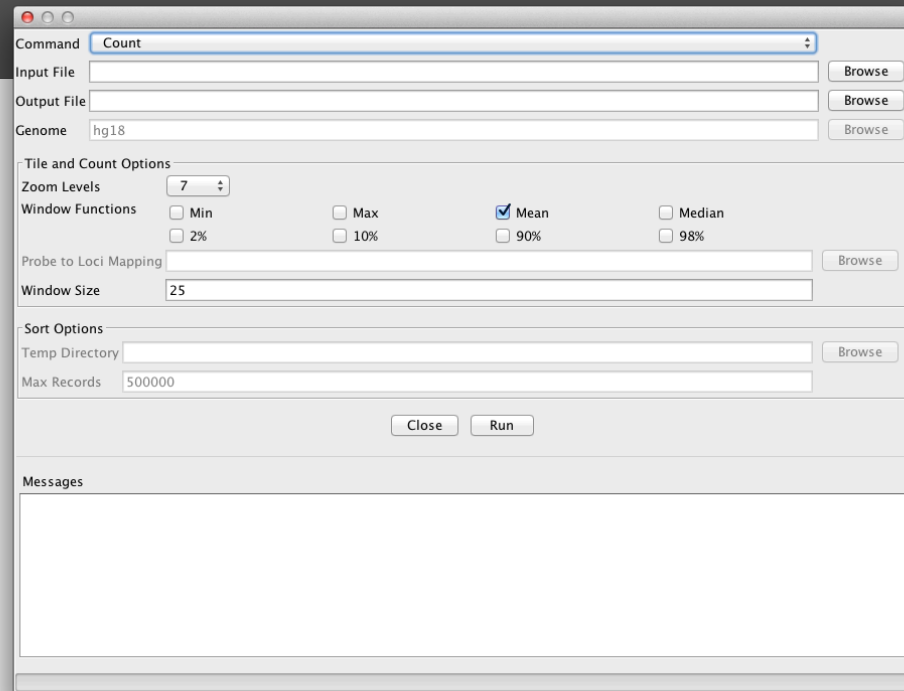
(A) Launch from the IGV menu

Tools > Run igvtools...

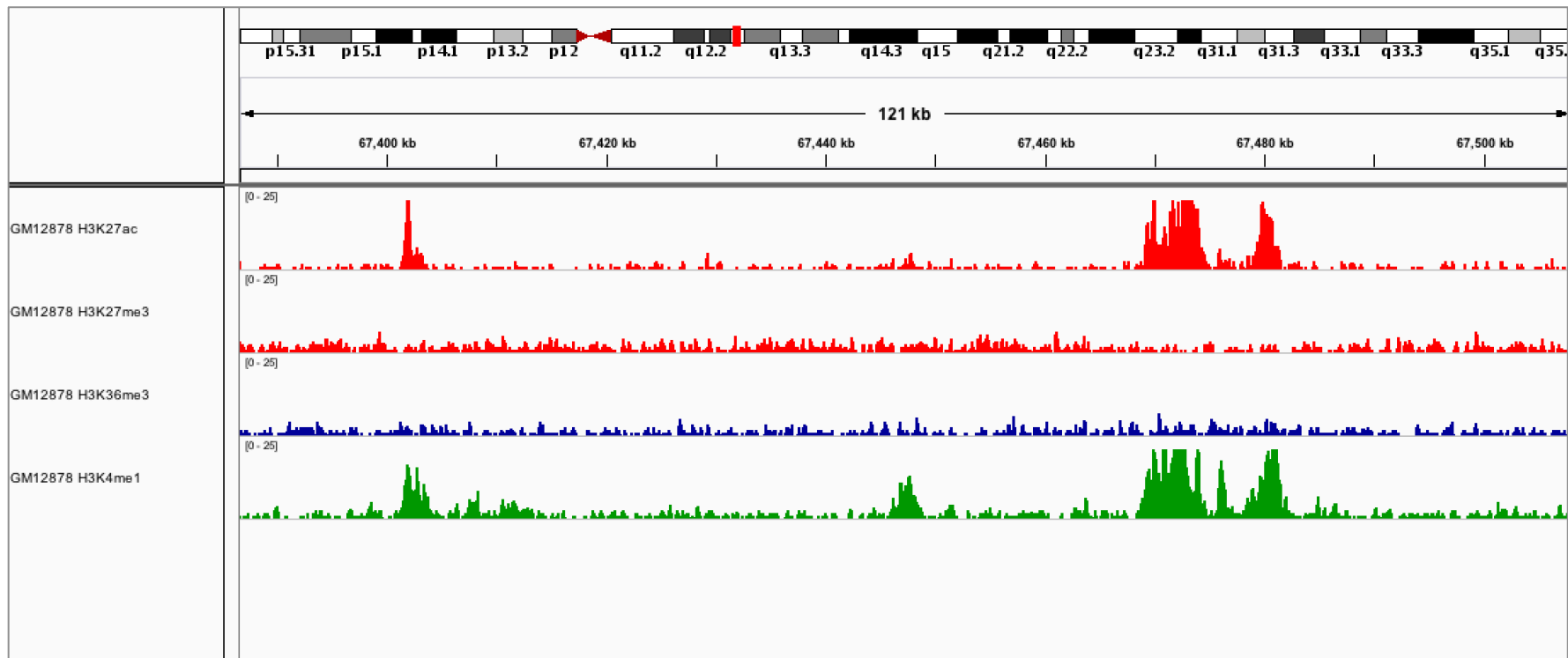
or

(B) Run from the command line

```
[GM31D-693:~] jrobinso%  
[GM31D-693:~] jrobinso% igvtools count NA12878.bam NA12878.bam.tdf hg18
```



Combining numeric data tracks



Combining numeric data tracks

Tools

Run Batch Script...

Run igvtools...

Find Motif...

BLAT ...

Combine Data Tracks

Gitools Heatmaps ▶

BEDTools ▶

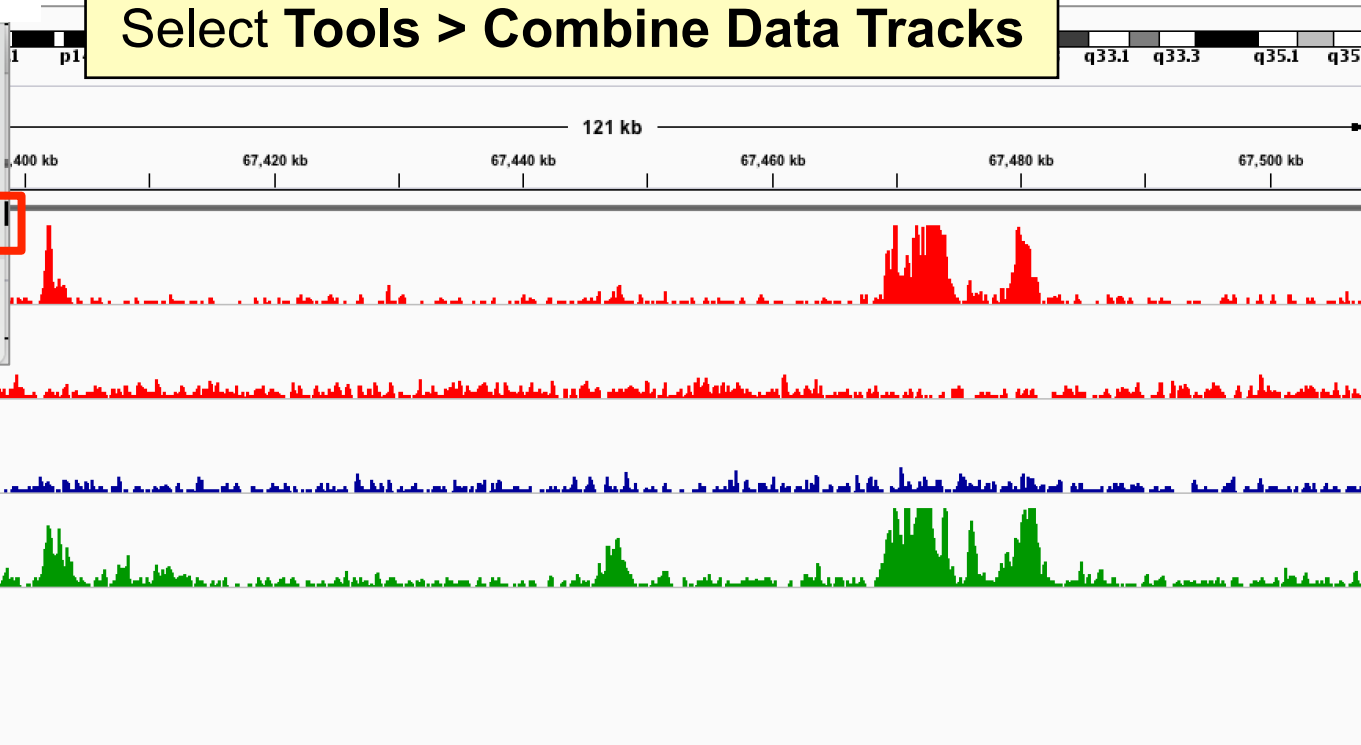
Select Tools > Combine Data Tracks

GM12878 H3K36me3

GM12878 H3K4me1

[0 - 25]

[0 - 25]



Combining numeric data tracks

1. Select two tracks from the dropdown menus

2. Select operation:

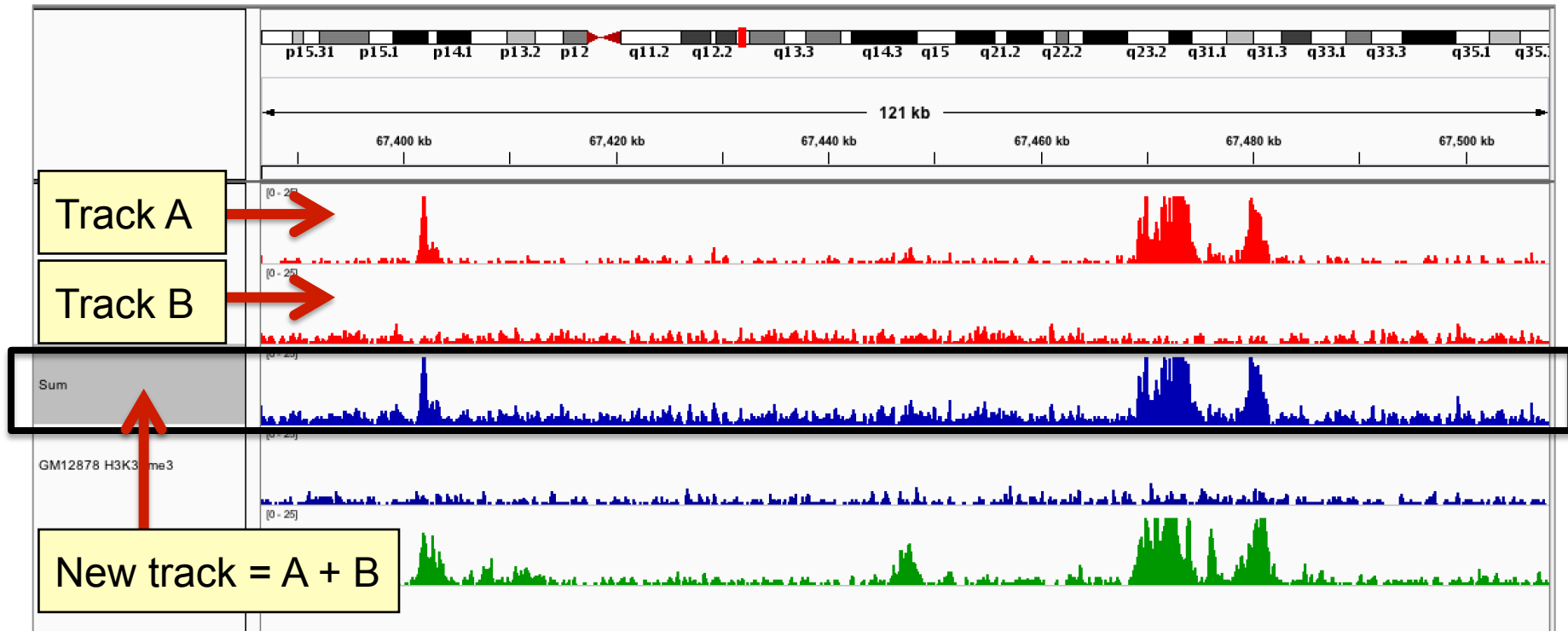
- Add
- Subtract
- Multiply
- Divide

3. Name the new combined track

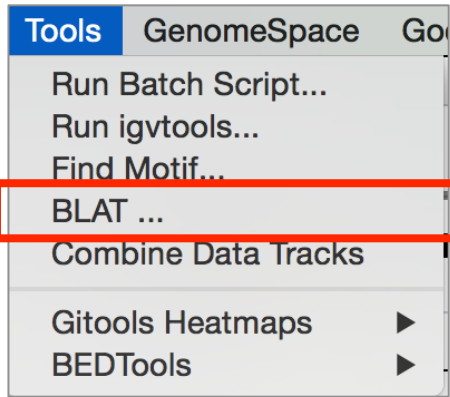
The image shows a 'Combine tracks' dialog box with the following elements and annotations:

- Track A:** A dropdown menu showing 'GM12878 H3K27ac'. A red arrow points from the first instruction box to this dropdown.
- Operation:** A dropdown menu showing 'Add'. A red arrow points from the second instruction box to this dropdown.
- Track B:** A dropdown menu showing 'GM12878 H3K27me3'. A red arrow points from the first instruction box to this dropdown.
- Result Track Name:** A text input field containing 'Sum'. A red arrow points from the third instruction box to this field.
- Buttons:** 'OK' and 'Cancel' buttons at the bottom.

Combining numeric data tracks

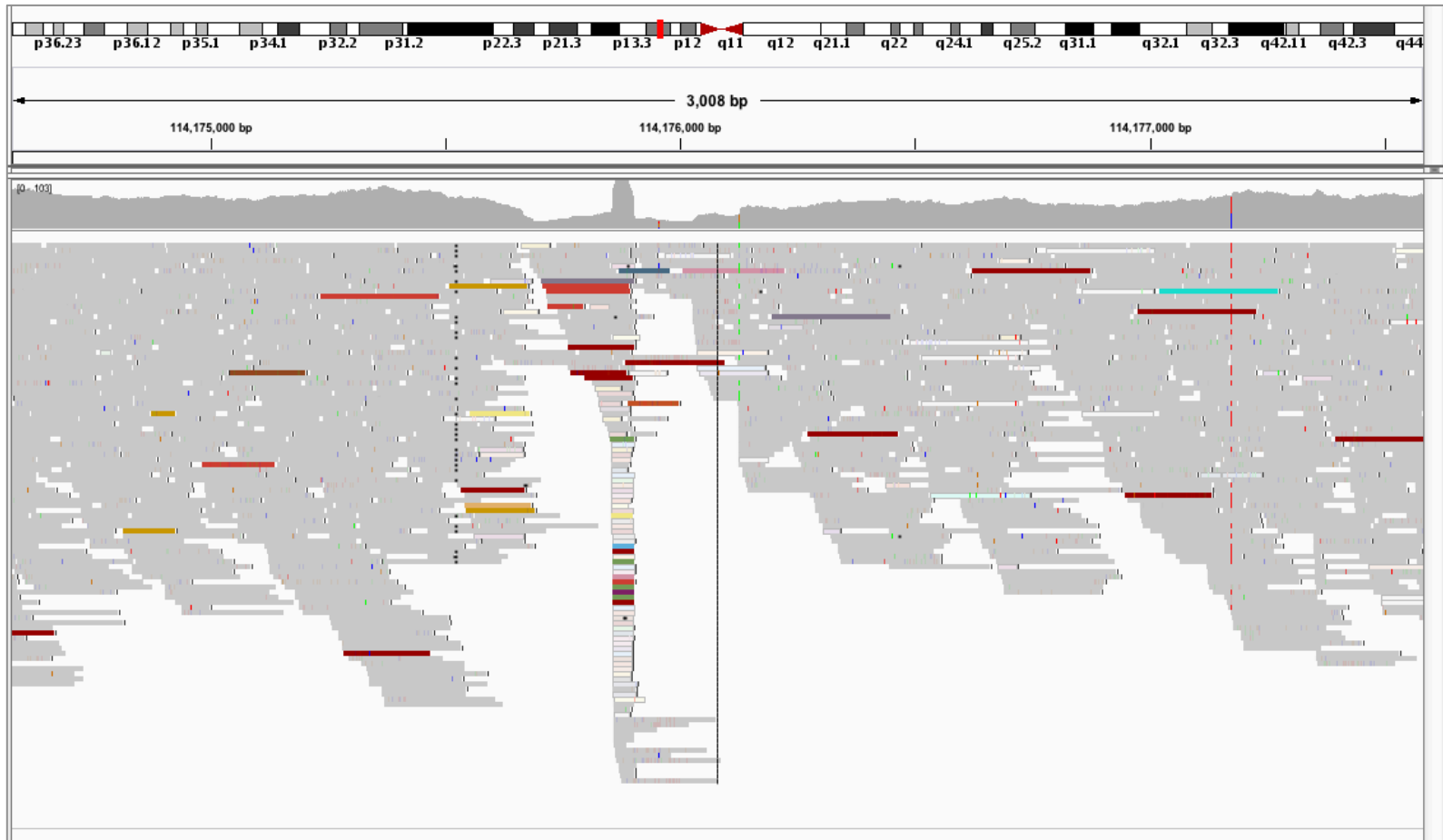


BLAT

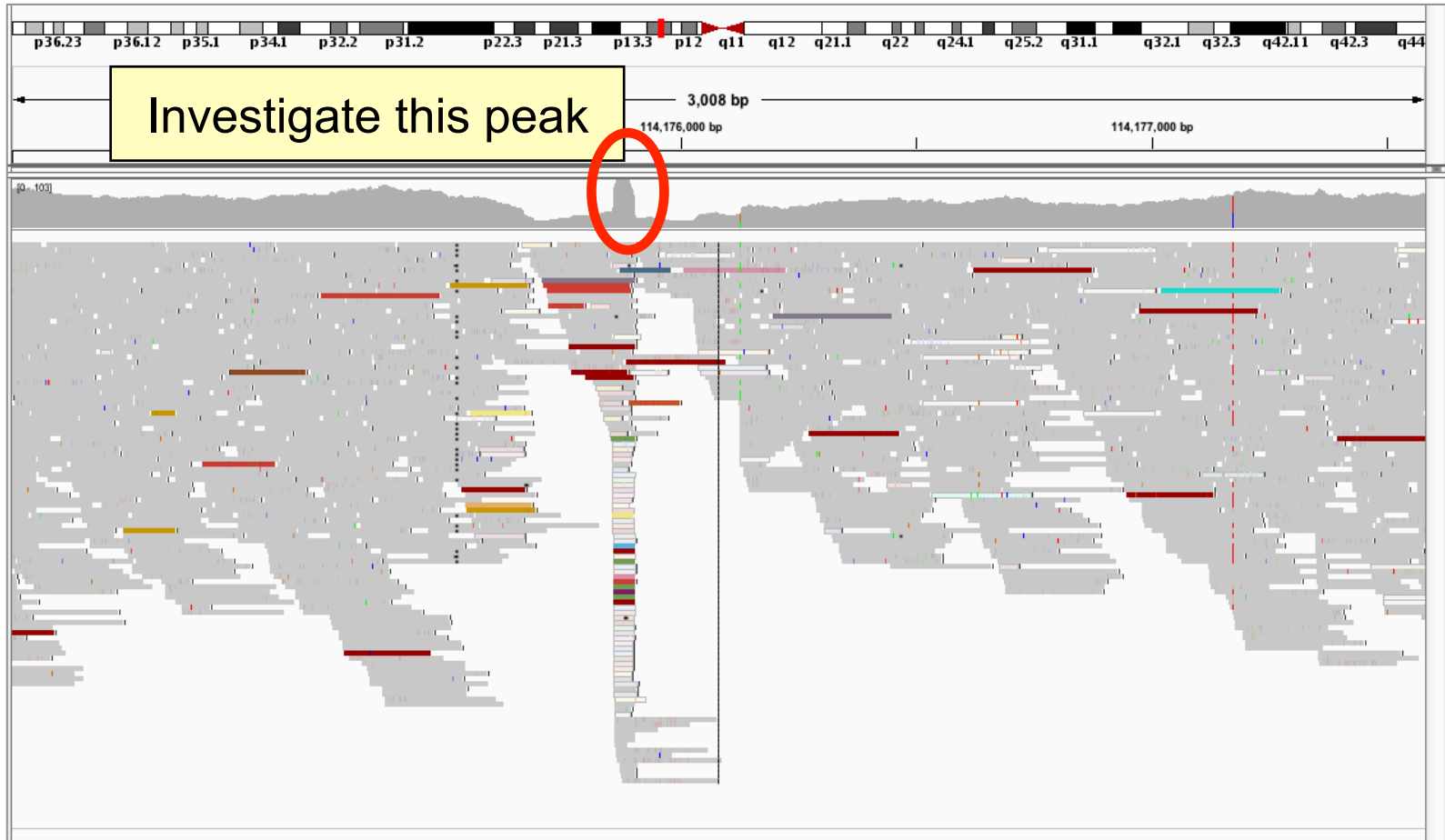


- Use a BLAT server to do search for alternative alignments for
 - any feature or sequence read, or
 - user-entered sequence.
- User-specified server or UCSC (volume limits apply).

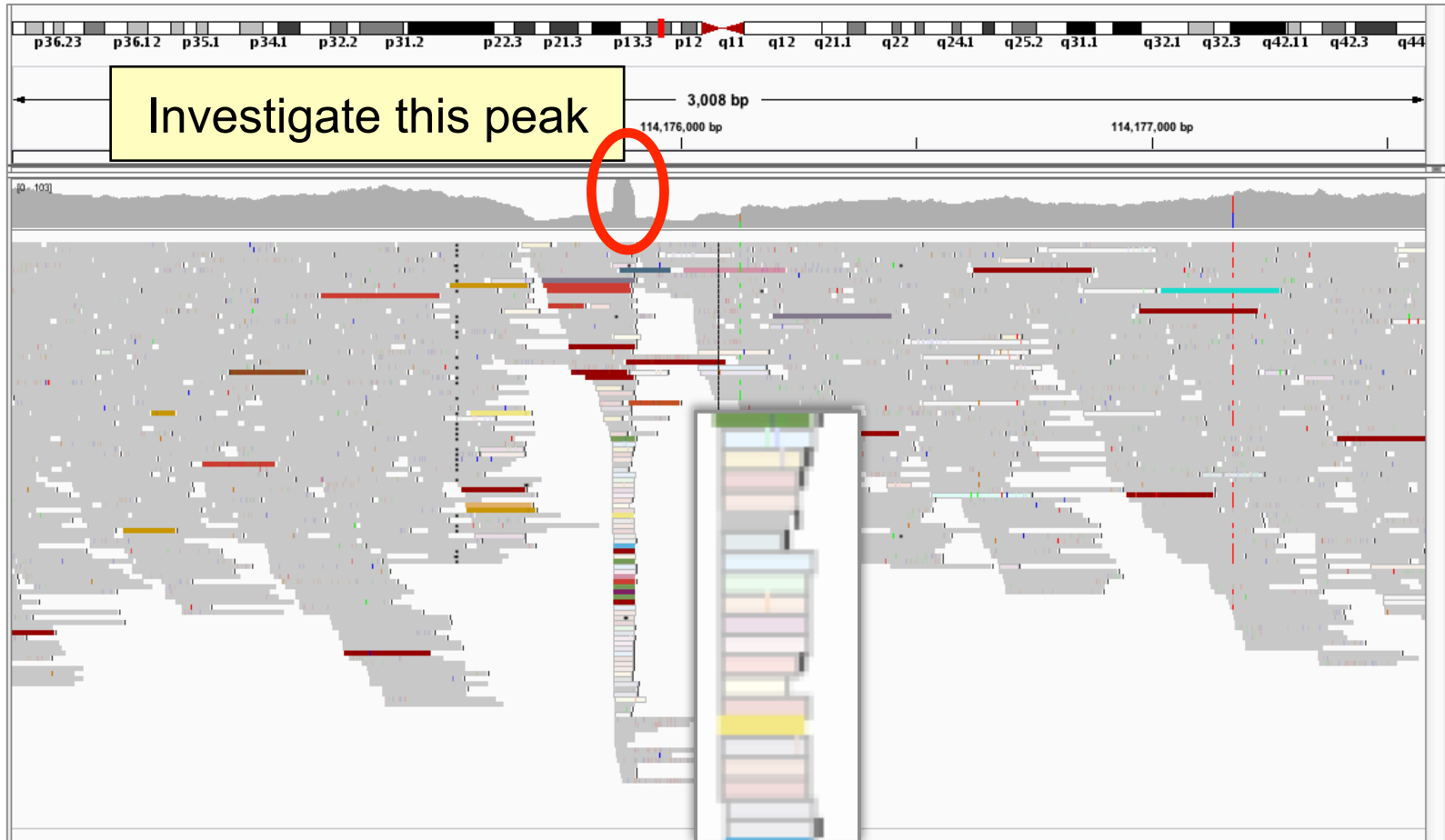
BLAT example



BLAT example

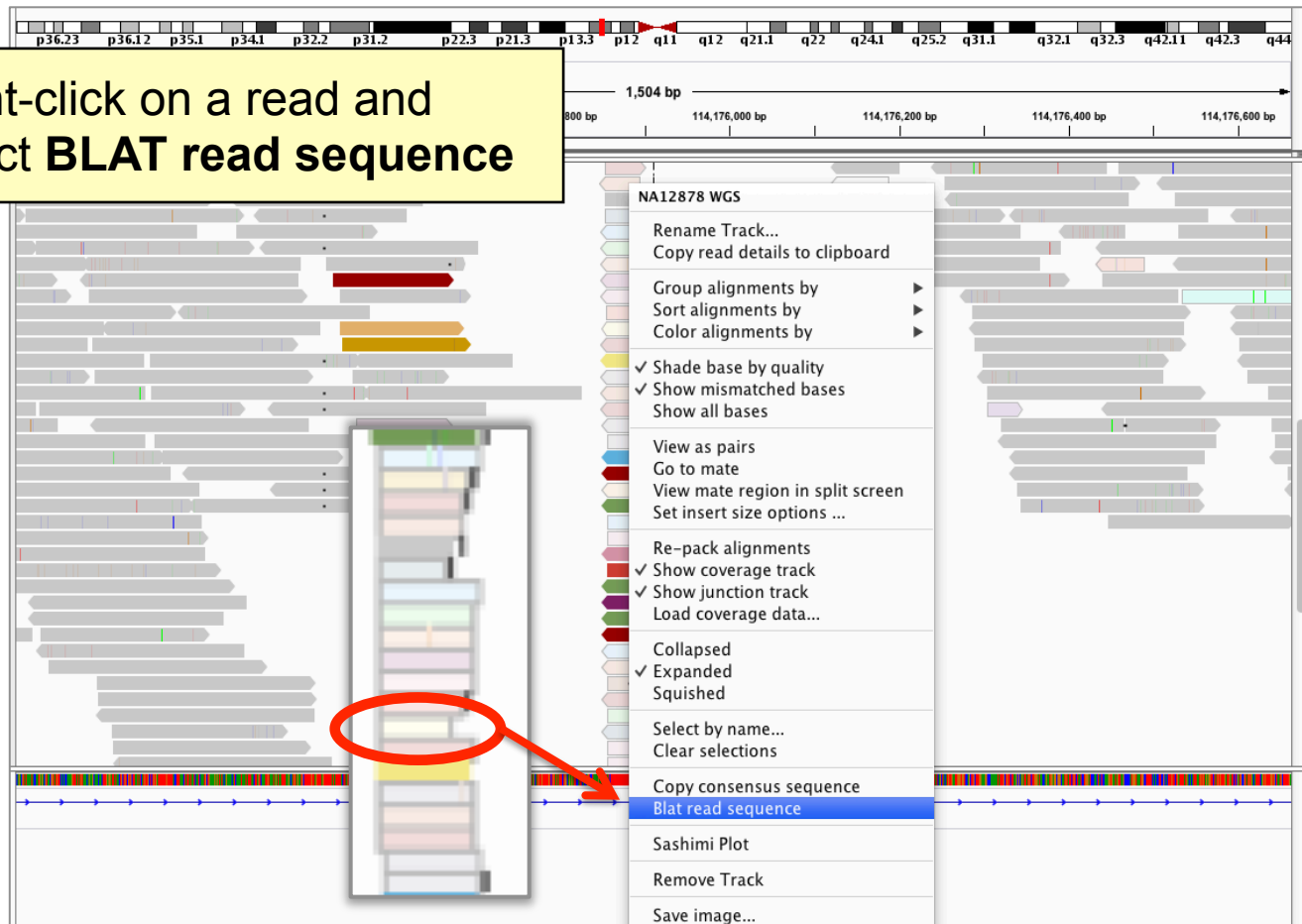


BLAT example



BLAT example

Right-click on a read and select **BLAT read sequence**



BLAT example

File												
BLAT result for query sequence: TAATGCACACGAACTCCATTAGGTATTCATATGGGTTGAAAAGGGGGGTTAACTTGGCCTAAATAGAAAATGTCTTCTCATCATTTTTTT												
Click on a row to go to alignment												
chr	start	end	strand	score	match	mis-match	rep. match	N's	Q gap co...	Q gap ba...	T gap co...	T gap ba...
chr6	1552157	1552307	-	536	142	8	0	0	0	0	0	0
chr1	401076...	401078...	-	492	147	19	0	0	2	9	3	16
chr2	715213...	715215...	+	456	137	22	0	0	0	0	1	1
chr13	525933...	525935...	+	444	135	23	0	0	0	0	1	1
chr17	5468054	5468211	+	440	128	16	0	0	1	12	1	13
chr10	933987...	933989...	+	436	132	22	0	0	0	0	1	1
chr15	956086...	956088...	-	428	129	20	0	0	0	0	2	4
chr5	166236...	166238...	+	424	126	18	0	0	0	0	2	4
chr3	102995...	102996...	+	424	124	15	0	0	1	5	2	9
chr17	708930...	708932...	-	420	130	24	0	0	0	0	1	1
chr12	571299...	571301...	-	420	135	27	0	0	1	1	2	4
chr5	665088...	665089...	+	420	136	31	0	0	0	0	0	0
chr9	915725	915886	+	416	127	21	0	0	0	0	2	13
chr2	223622...	223622...	+	400	120	17	0	0	1	11	2	13
chr19	583386...	583388...	-	396	124	24	0	0	0	0	1	1
chr16	574045...	574047...	+	392	118	16	0	0	2	29	2	8
chr1	171497...	171497...	+	388	114	16	0	0	0	0	1	1
chr10	101042...	101042...	-	384	111	13	0	0	0	0	2	7
chr2	228547...	228547...	+	384	116	19	0	0	0	0	1	1
chr1	202874...	202875...	+	384	120	23	0	0	0	0	1	1
chr1	549438...	549439...	+	384	119	22	0	0	0	0	1	1
chr12	570020...	570021...	-	380	113	17	0	0	0	0	1	1
chr4	878470...	878471...	+	376	119	22	0	0	0	0	3	6
chr2	145618...	145619...	+	376	110	11	0	0	3	38	2	40
chr1	7404808	7404937	+	372	109	15	0	0	0	0	1	5
chr1	247194	247194	-	368	106	12	0	0	0	0	2	6

BLAT example

Many alternative alignments
with similar scores

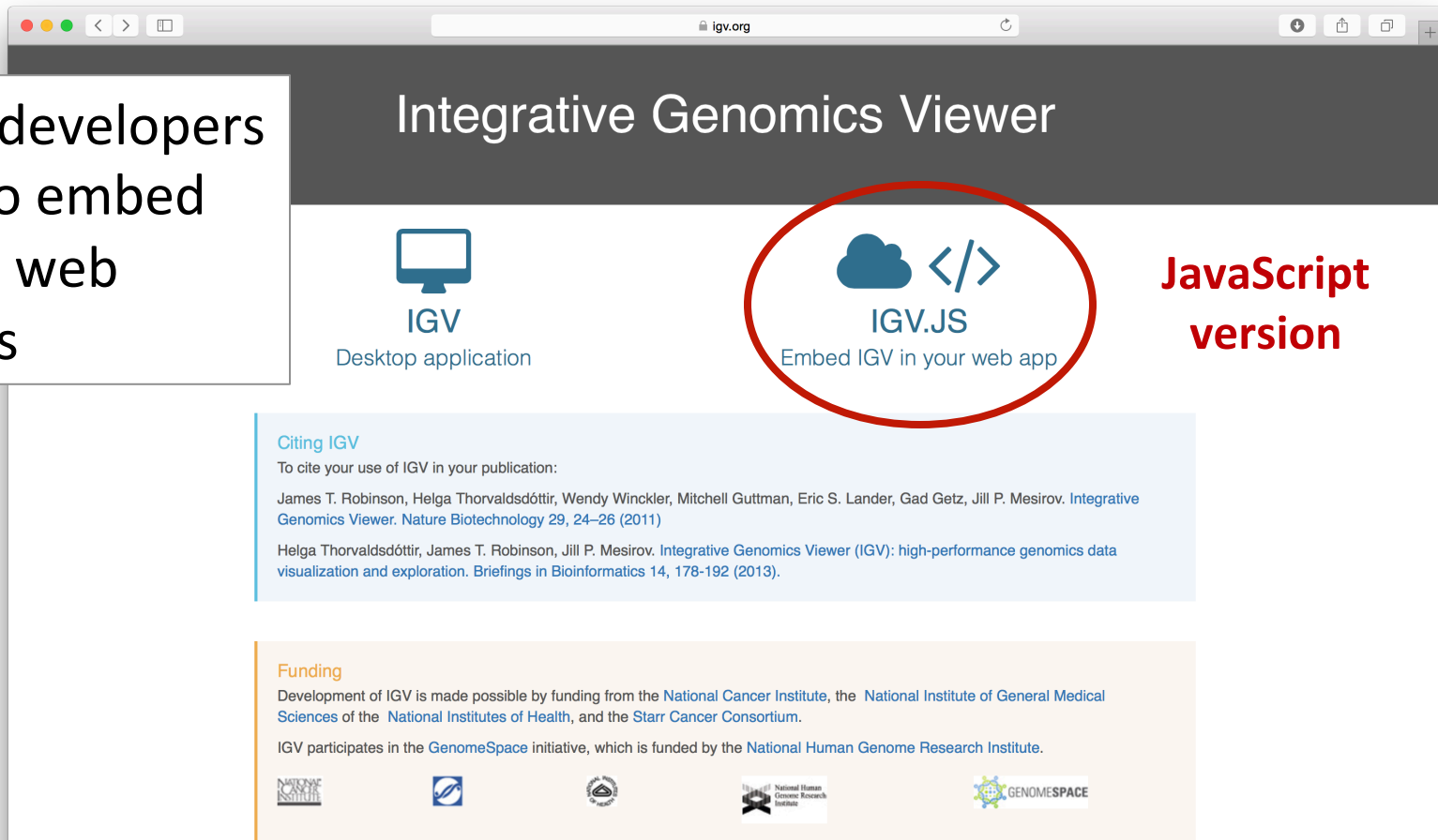
GTGAAAAGGGGGGTAACTTGGCCTAAATAGAAAATGTCTTCTCATCATTTTTTT

chr	start	end	strand	score	match	mis-match	rep. match	N's	Q gap co...	Q gap ba...	T gap co...	T gap ba...
chr6	1552157	1552307	-	536	142	8	0	0	0	0	0	0
chr1	401076...	401078...	-	492	147	19	0	0	2	9	3	16
chr2	715213...	715215...	+	456	137	22	0	0	0	0	1	1
chr13	525933...	525935...	+	444	135	23	0	0	0	0	1	1
chr17	5468054	5468211	+	440	128	16	0	0	1	12	1	13
chr10	933987...	933989...	+	436	132	22	0	0	0	0	1	1
chr15	956086...	956088...	-	428	129	20	0	0	0	0	2	4
chr5	166236...	166238...	+	424	126	18	0	0	0	0	2	4
chr3	102995...	102996...	+	424	124	15	0	0	1	5	2	9
chr17	708930...	708932...	-	420	130	24	0	0	0	0	1	1
chr12	571299...	571301...	-	420	135	27	0	0	1	1	2	4
chr5	665088...	665089...	+	420	136	31	0	0	0	0	0	0
chr9	915725	915886	+	416	127	21	0	0	0	0	2	13
chr2	223622...	223622...	+	400	120	17	0	0	1	11	2	13
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chr16	574045...	574047...	+	392	118	16	0	0	2	29	2	8
chr1	171497...	171497...	+	388	114	16	0	0	0	0	1	1
chr10	101042...	101042...	-	384	111	13	0	0	0	0	2	7
chr2	228547...	228547...	+	384	116	19	0	0	0	0	1	1
chr1	202874...	202875...	+	384	120	23	0	0	0	0	1	1
chr1	549438...	549439...	+	384	119	22	0	0	0	0	1	1
chr12	570020...	570021...	-	380	113	17	0	0	0	0	1	1
chr4	878470...	878471...	+	376	119	22	0	0	0	0	3	6
chr2	145618...	145619...	+	376	110	11	0	0	3	38	2	40
chr1	7404808	7404937	+	372	109	15	0	0	0	0	1	5
chr1	247184	247184	-	368	106	12	0	0	0	0	2	5

igv.js

www.igv.org

igv.js is for developers who wish to embed IGV in their web applications



Integrative Genomics Viewer


IGV
Desktop application


IGV.JS
Embed IGV in your web app

JavaScript version

Citing IGV

To cite your use of IGV in your publication:

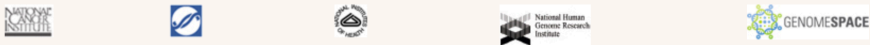
James T. Robinson, Helga Thorvaldsdóttir, Wendy Winckler, Mitchell Guttman, Eric S. Lander, Gad Getz, Jill P. Mesirov. [Integrative Genomics Viewer](#). *Nature Biotechnology* 29, 24–26 (2011)

Helga Thorvaldsdóttir, James T. Robinson, Jill P. Mesirov. *Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration*. *Briefings in Bioinformatics* 14, 178-192 (2013).

Funding

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IGV participates in the [GenomeSpace](#) initiative, which is funded by the [National Human Genome Research Institute](#).





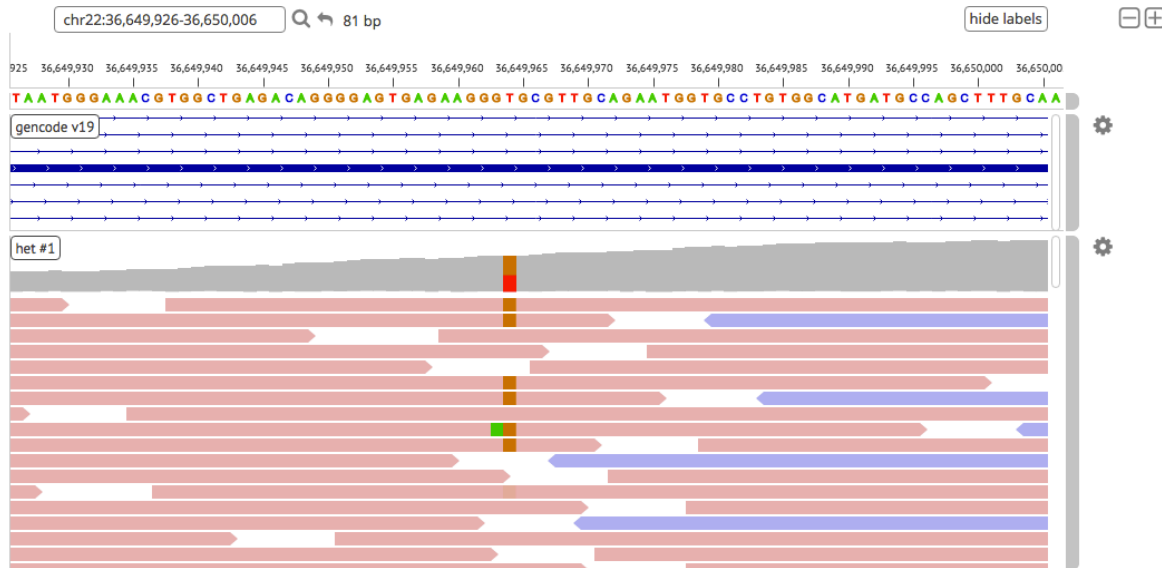
Example of web application that uses igv.js:

ExAc browser from the Exome Aggregation Consortium (exac.broadinstitute.org)

Read Data

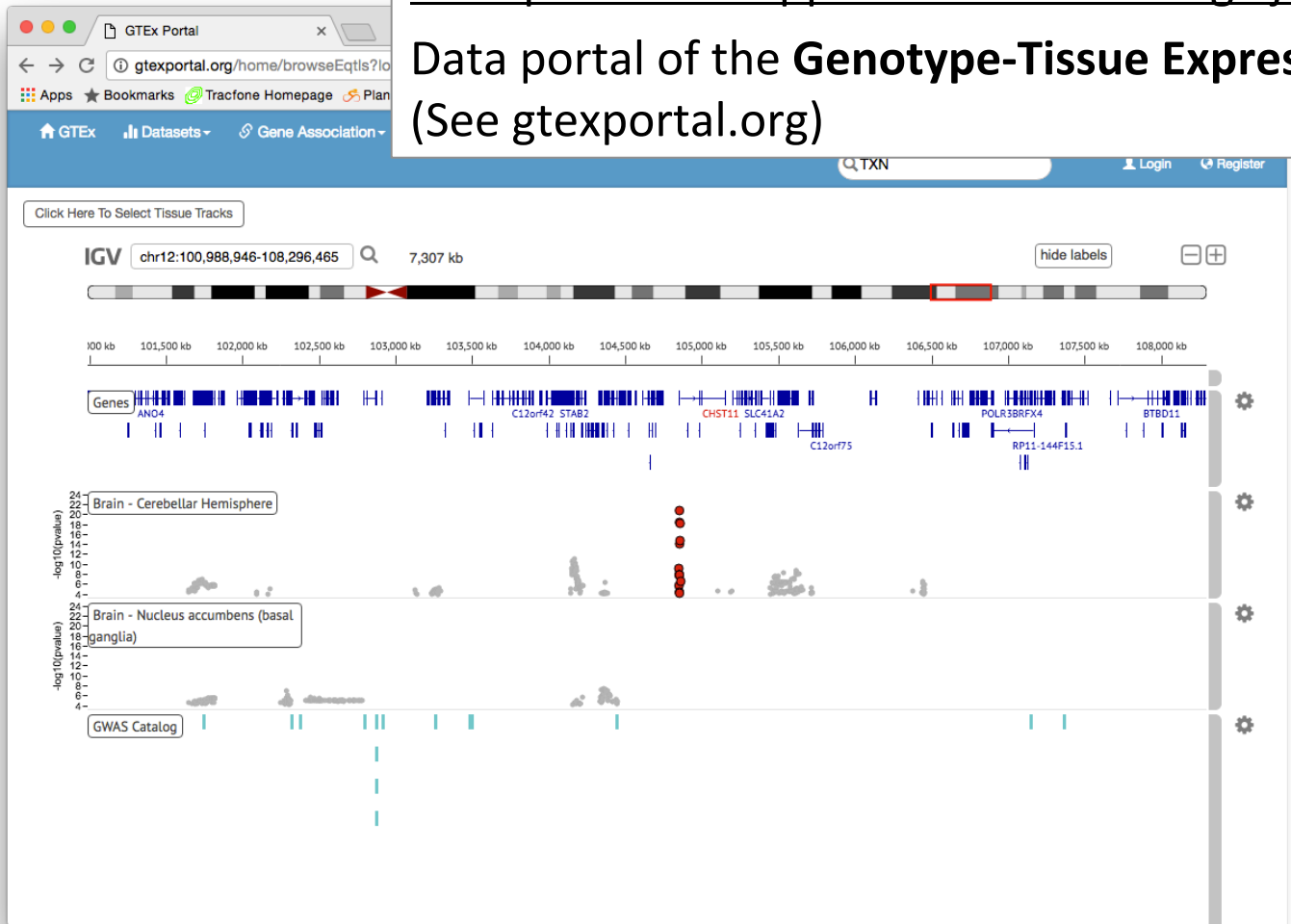
This interactive [IGV.js](#) visualization shows reads that went into calling this variant.

Note: These are reassembled reads produced by [GATK HaplotypeCaller --bamOutput](#) so they accurately represent what HaplotypeCaller was seeing when it called this variant.



Example of web application that uses igv.js:

Data portal of the **Genotype-Tissue Expression (GTEx)** project
(See gtexportal.org)



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- National Cancer Institute (NCI) (www.cancer.gov)
- NCI's Information Technology for Cancer Research (ITCR) (itcr.nci.nih.gov)
- Starr Cancer Consortium (www.starrcancer.org)
- Stand Up 2 Cancer
- National Institute of General Medical Sciences (NIGMS) (www.nigms.nih.gov)
- IGV participates in GenomeSpace (www.genomespace.org), which is funded by the National Human Genome Research Institute (NHGRI) (www.genome.gov).

For further information and help

Workshop slides and data: www.igv.org/workshops/ASHG2016

IGV web site: www.igv.org

User forum: groups.google.com/group/igv-help

Source code: github.com/igvteam

Please cite your use of IGV

Robinson et al.

Integrative Genomics Viewer. Nature Biotechnology (2011).

Thorvaldsdóttir, Robinson, and Mesirov.

Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration. Briefings in Bioinformatics (2012).