

Introduction to the Integrative Genomics Viewer (IGV)

Jim Robinson
UC San Diego
April 2017



Acknowledgments

Development of IGV has been made possible by funding from:

- 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)
- And
 - 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).

The logo for the Integrative Genomics Viewer (IGV) consists of the letters 'IGV' in a bold, white, sans-serif font, centered within a dark gray square.

IGV

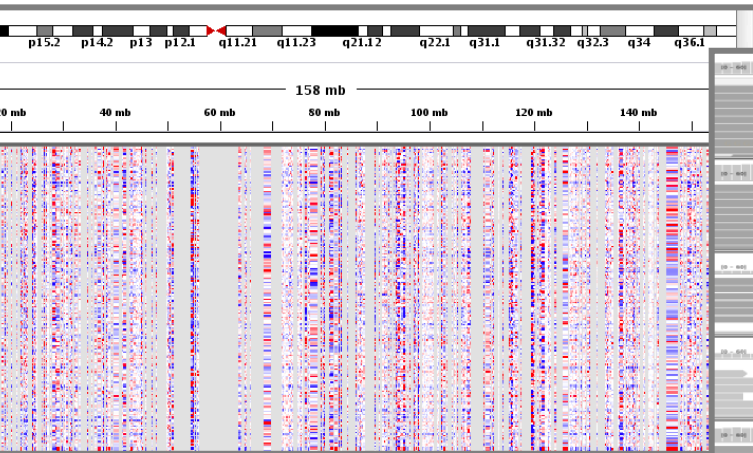
**Interactive
visual exploration of
integrated
genomic data**

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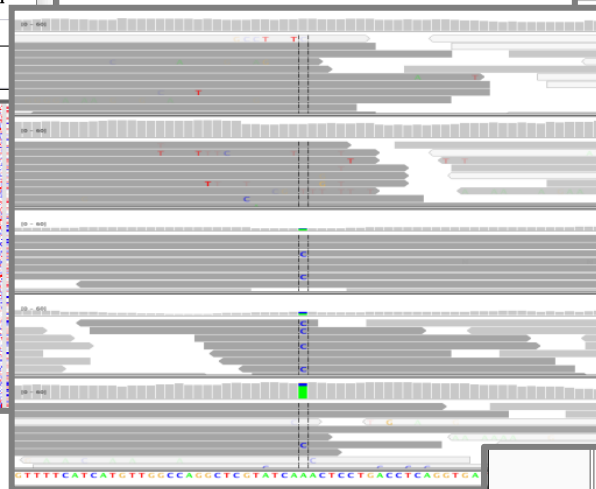
IGV

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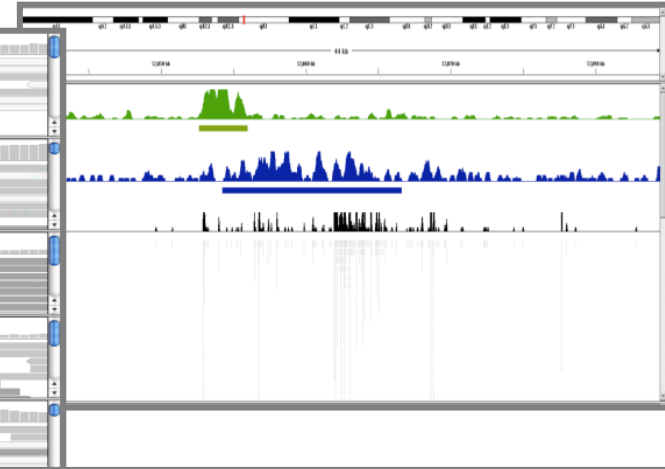
Microarray gene expression



NGS alignments



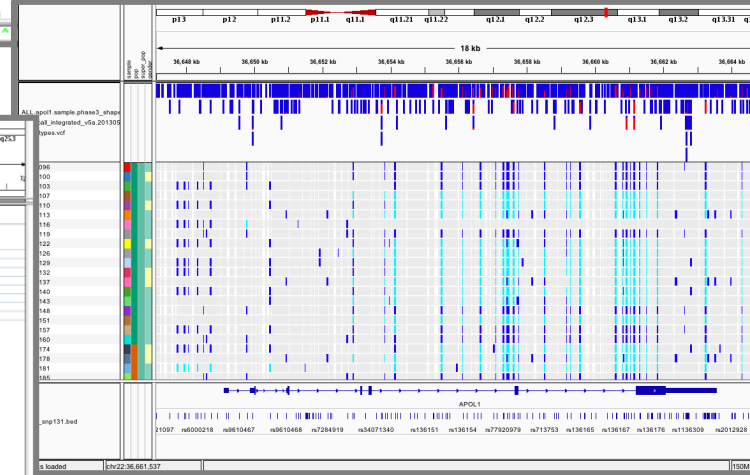
Epigenomics



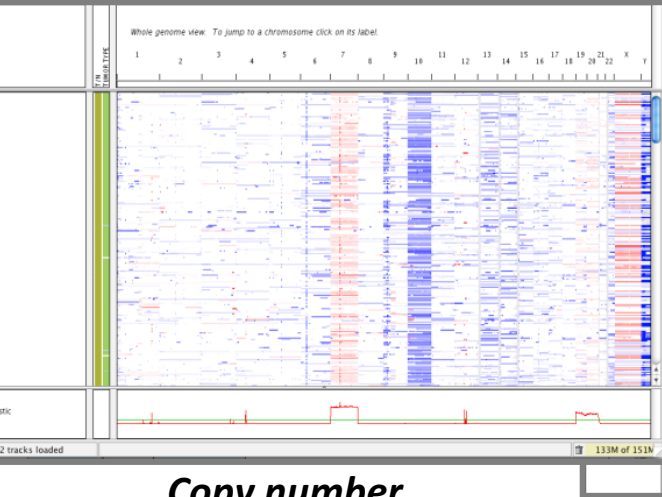
RNA-seq



Variants and genotypes



Copy number



Gene annotations



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](#)

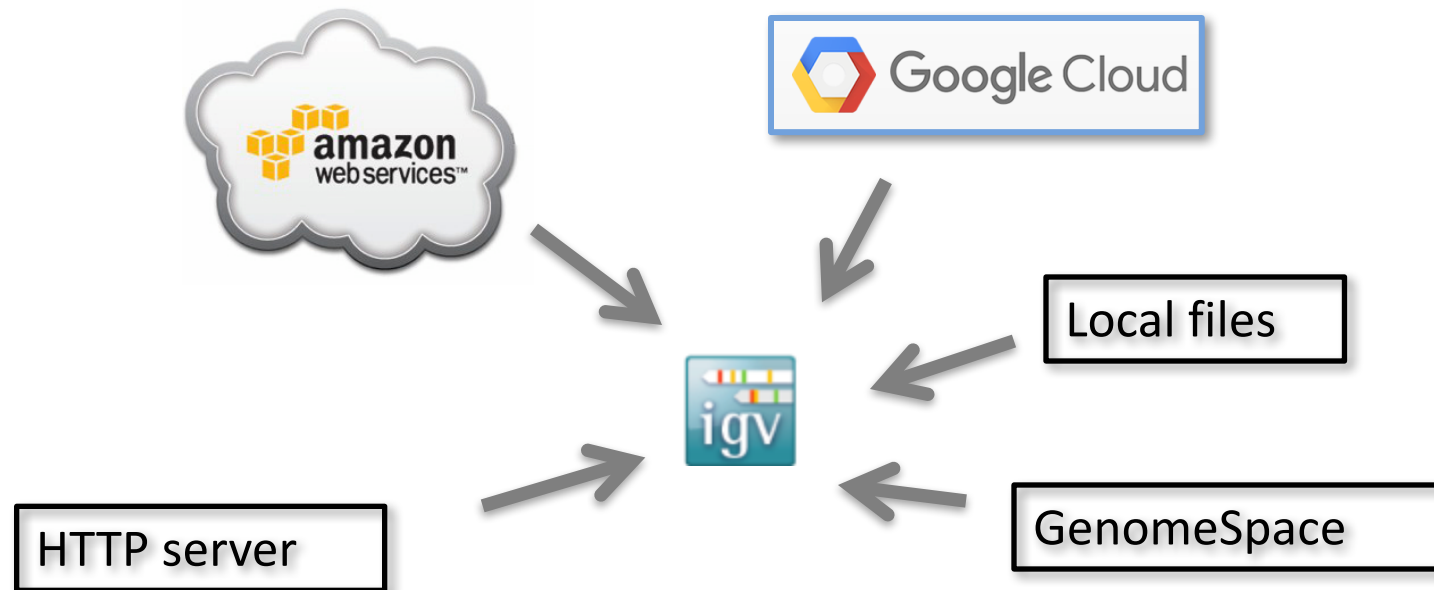
For more info see: [*www.broadinstitute.org/igv/FileFormats*](http://www.broadinstitute.org/igv/FileFormats)

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IGV

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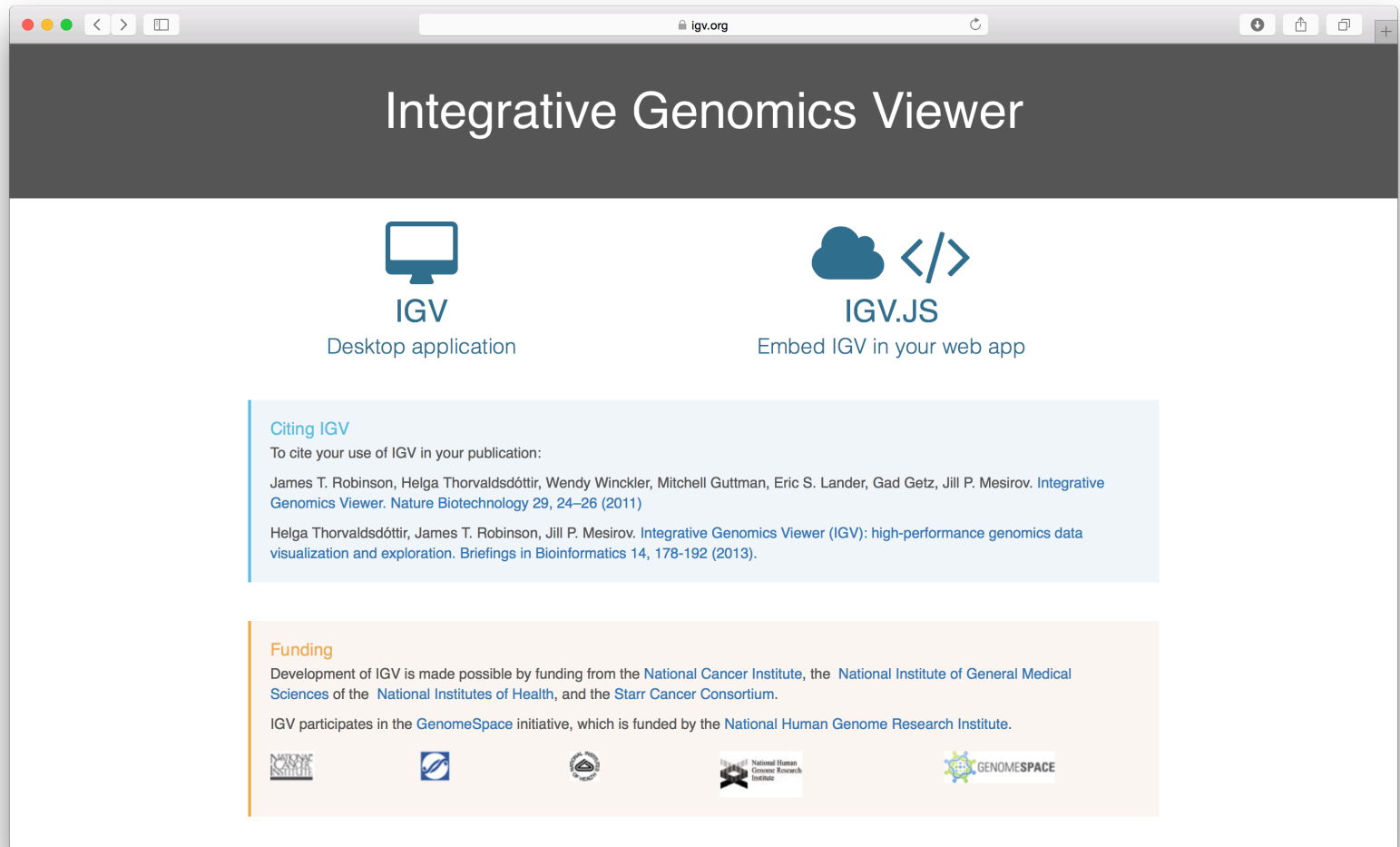
IGV data sources



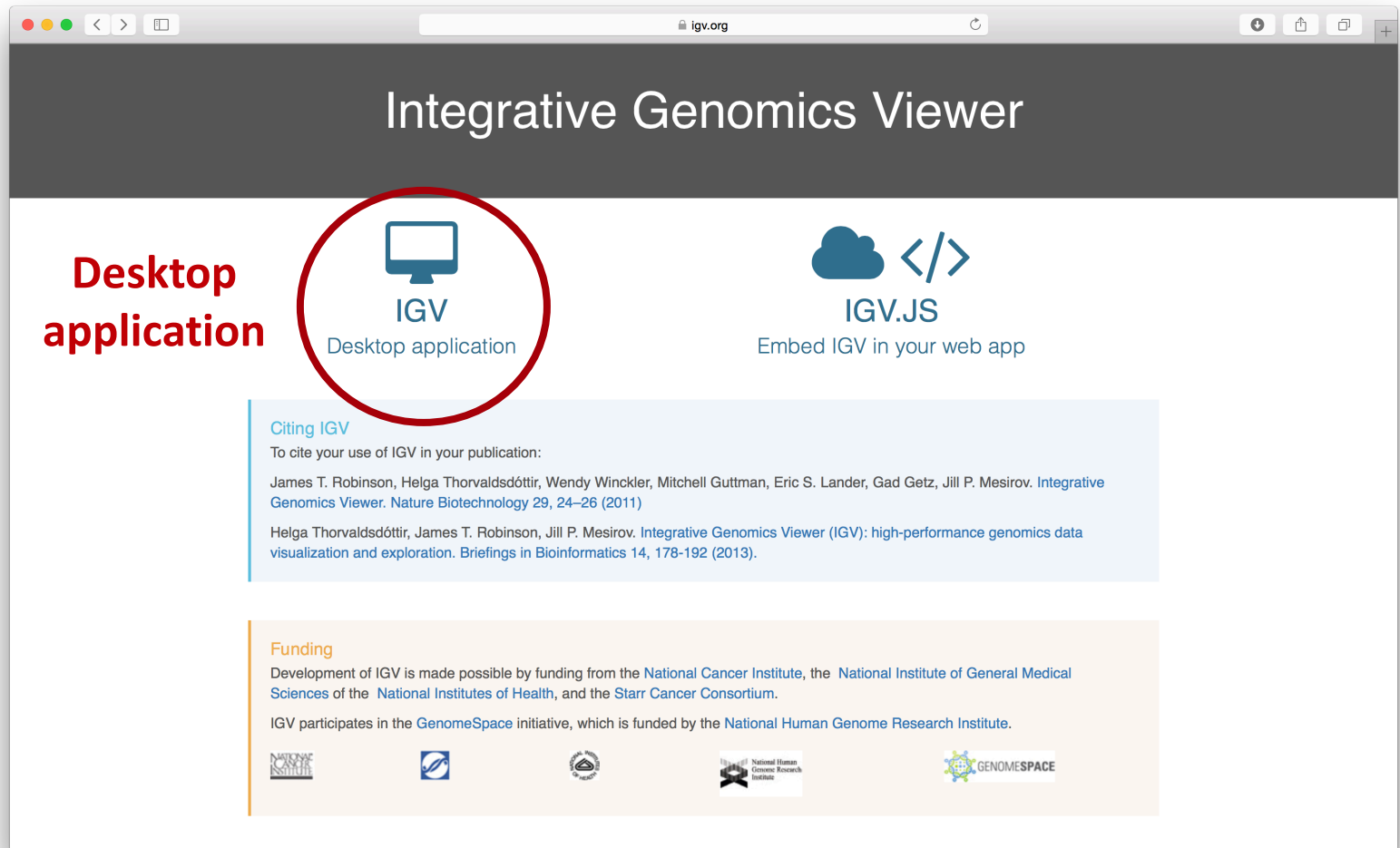
View **local** files without uploading

View **remote** files without downloading whole dataset

www.igv.org



www.igv.org



www.broadinstitute.org/igv



Integrative
Genomics
Viewer

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[Downloads](#)
[Documents](#)
 Hosted Genomes
 FAQ
 IGV User Guide
 File Formats
 Release Notes
 IGV for iPad
 Credits
[@ Contact](#)

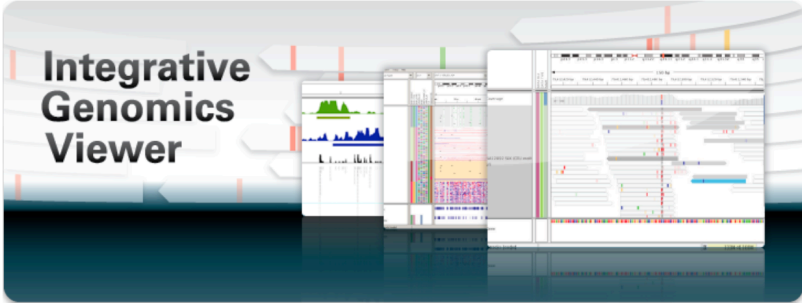
Search website



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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.

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](#).



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 \(IGV\): high-performance genomics data visualization and exploration](#). [Briefings in Bioinformatics](#) 14, 178-192 (2013).

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).

www.broadinstitute.org/igv

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IGV is freely available under an MIT open-source license.

Install IGV

Options for installing and running IGV:

1. (Mac only) Download and run the Mac application; or
2. (Windows) Download and run the self-extracting archive; or
3. (All systems) Use the Java Web Start buttons (Mac users: see below for limitations); or
4. (All systems) Download the binary distribution and run IGV from the command line.

Note: IGV 2.3.x requires Java 7. Users with Java 6 (JRE 1.6) should first try to upgrade Java to the latest version. If this is not possible you will need to run a 2.2.x version available in the [archive](#).

Mac

Download and unzip the Mac App archive, then double-click the IGV application to run it. The application can be moved to the "Applications" folder, or anywhere else.

Download
Mac App

Download
Mac App

or

Download
Windows Package

Windows

NOTE: for 64-bit windows only. For 32-bit windows use the 750MB launch button below.

1. Download the Windows package and execute the self-extracting archive.
2. It will prompt you for a location to extract the folder, choose anywhere you like (e.g. your home folder).
3. On completion, open the new folder.
4. Double-click the file "igv.bat", it might appear as just "igv" depending on your settings.

Download
Windows Package

Java Web Start (All Platforms)

The buttons below use Java Web Start (JWS) to install and launch IGV directly from our web site.

***Mac Users:** The Java Web Start option does not work for some users due to security settings. The recommended solution is to use the bundled Mac App from the link above. Alternatively you can try to work around this by right-clicking on the buttons and saving the ".jnlp" file, then right-clicking on the saved ".jnlp" file and select "Open With > Java Web Start".

Chrome: Chrome does not automatically launch the Java Web Start files by default. Instead, the launch buttons below will download a ".jnlp" file. This should appear in the lower left corner of the browser. Double-click the downloaded file to run, or if on a Mac right-click and select "Open With > Java Web Start".

Windows users: To run with more than 1.2 GB of memory you must install 64-bit Java. **Most Windows installs do not include 64-bit Java by default, even if the operating system is 64-bit.** Attempting to use the 2GB or greater launch options with 32-bit Java will result in the error "could not create virtual machine".



Launch with 750 MB



Launch with 1.2 GB

Maximum usable memory
for Windows OS with 32-
bit Java.



Launch with 2 GB

Maximum usable memory
for 32-bit MacOS.



Launch with 10 GB

For large memory
machines with 64-bit Java.



Launch

Java Web Start

or

Download
Binary Distribution

Binary Distribution

Download and unzip the binary distribution archive in a folder of your choosing. IGV is launched from a command prompt -- follow instructions in the "readme" file. To launch igv on Mac or Linux platforms use the shell script "igv.sh". On Windows use "igv.bat".

Download
Binary Distribution

Install IGV

Options for installing and running IGV:

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Launch

Launch with 750 MB

Launch

Launch with 1.2 GB

Maximum usable memory
for Windows OS with 32-
bit Java.

Launch

Launch with 2 GB

Maximum usable memory
for 32-bit MacOS.

Launch

Launch with 10 GB

For large memory
machines with 64-bit Java.

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Binary Distribution

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Java Web Start

or

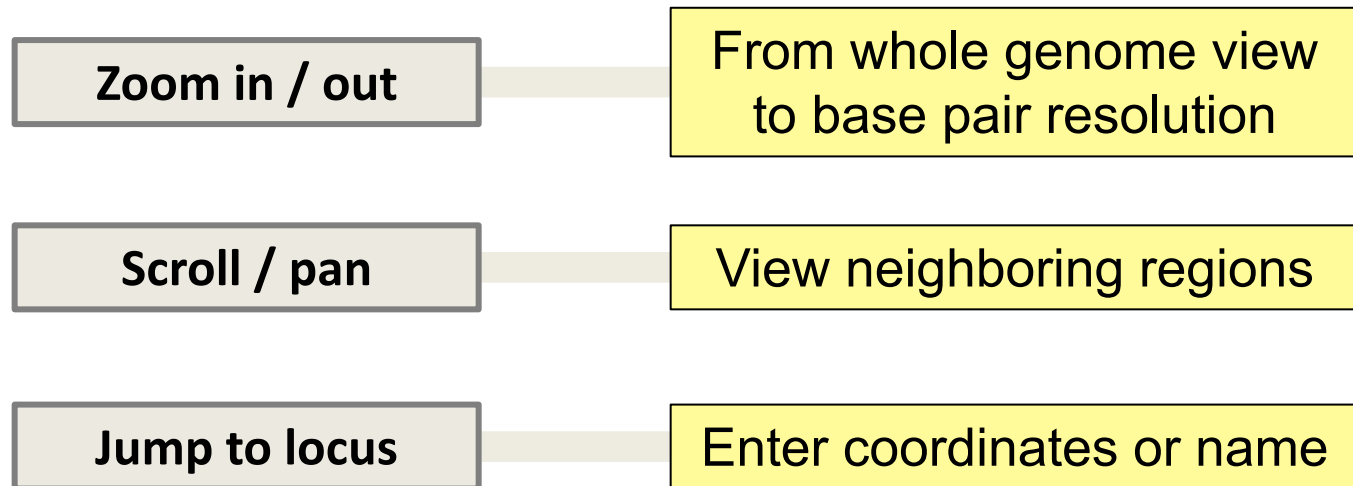
Download
Binary Distribution

Using IGV – The Basics

IGV user interface basics

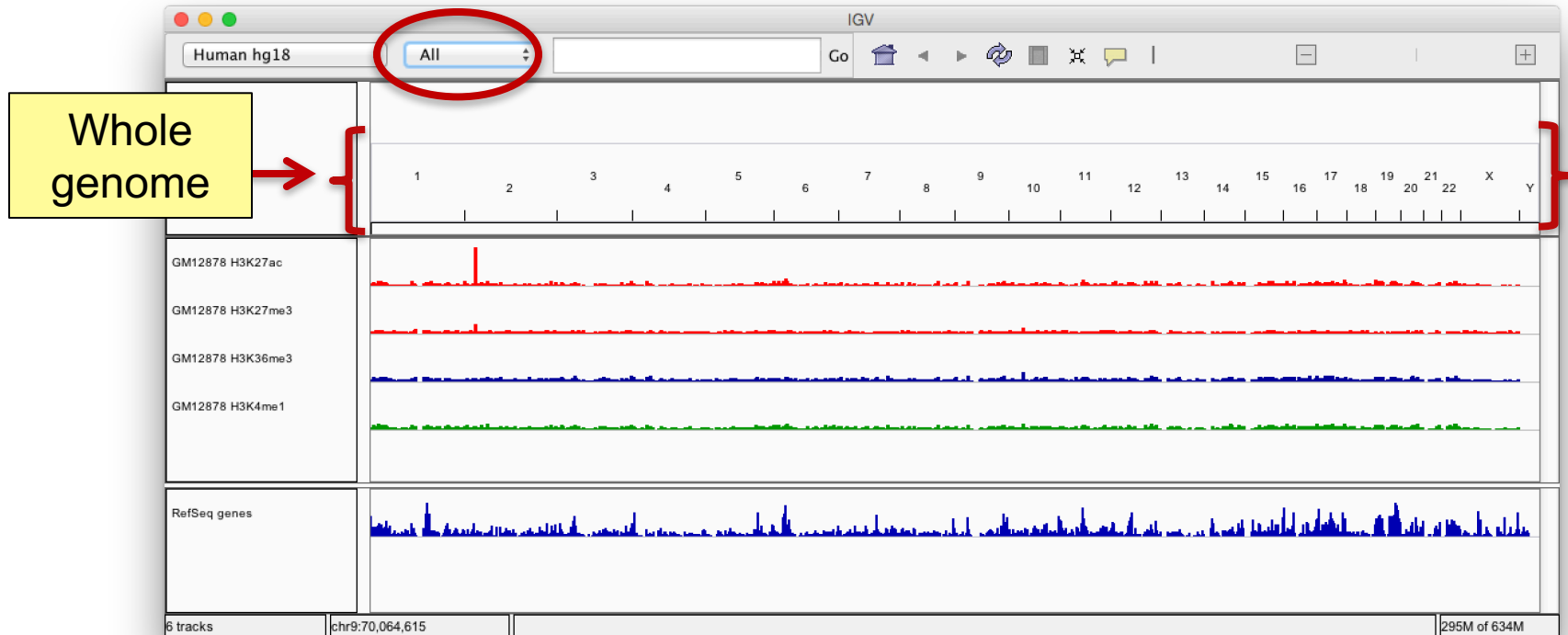
Hands-on exercise

- Select reference genome
- Load data from IGV hosted data server
- Navigate



IGV user interface basics

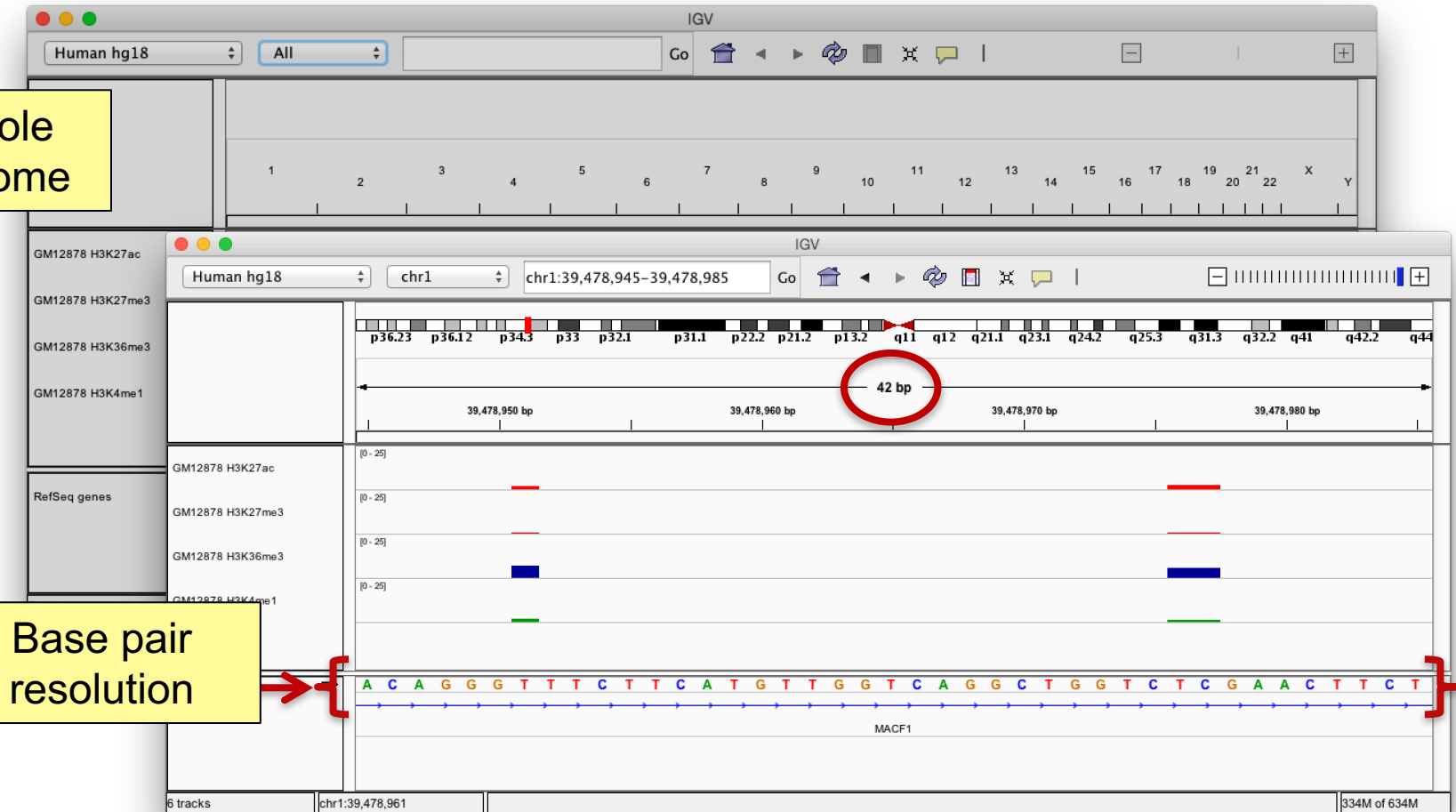
Hands-on exercise



IGV user interface basics

Hands-on exercise

Whole
genome



Base pair
resolution

IGV user interface basics

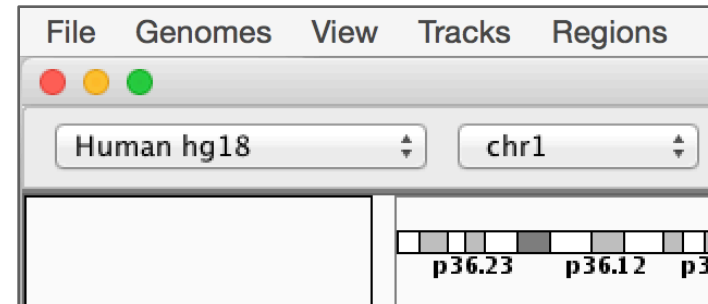
Hands-on exercise

See handout

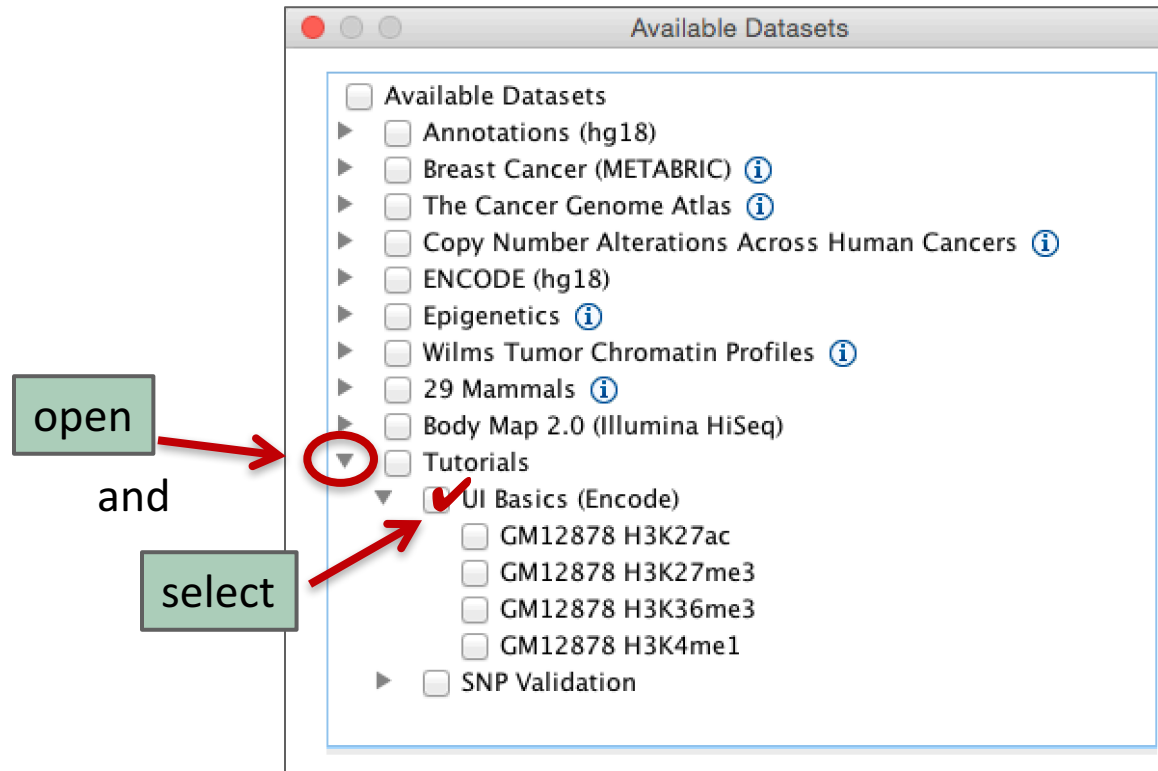
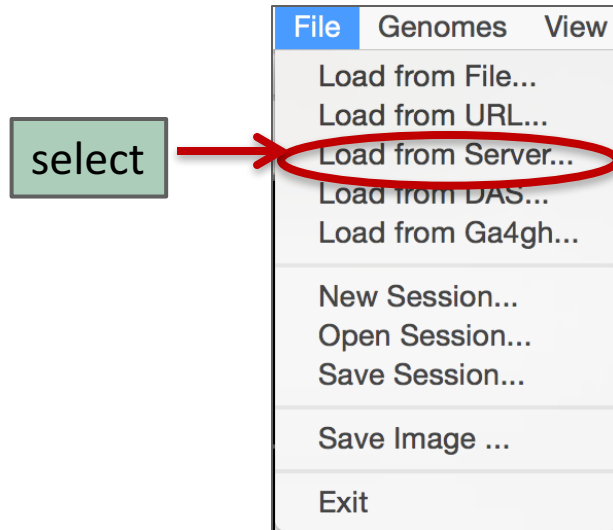
Select the reference genome

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

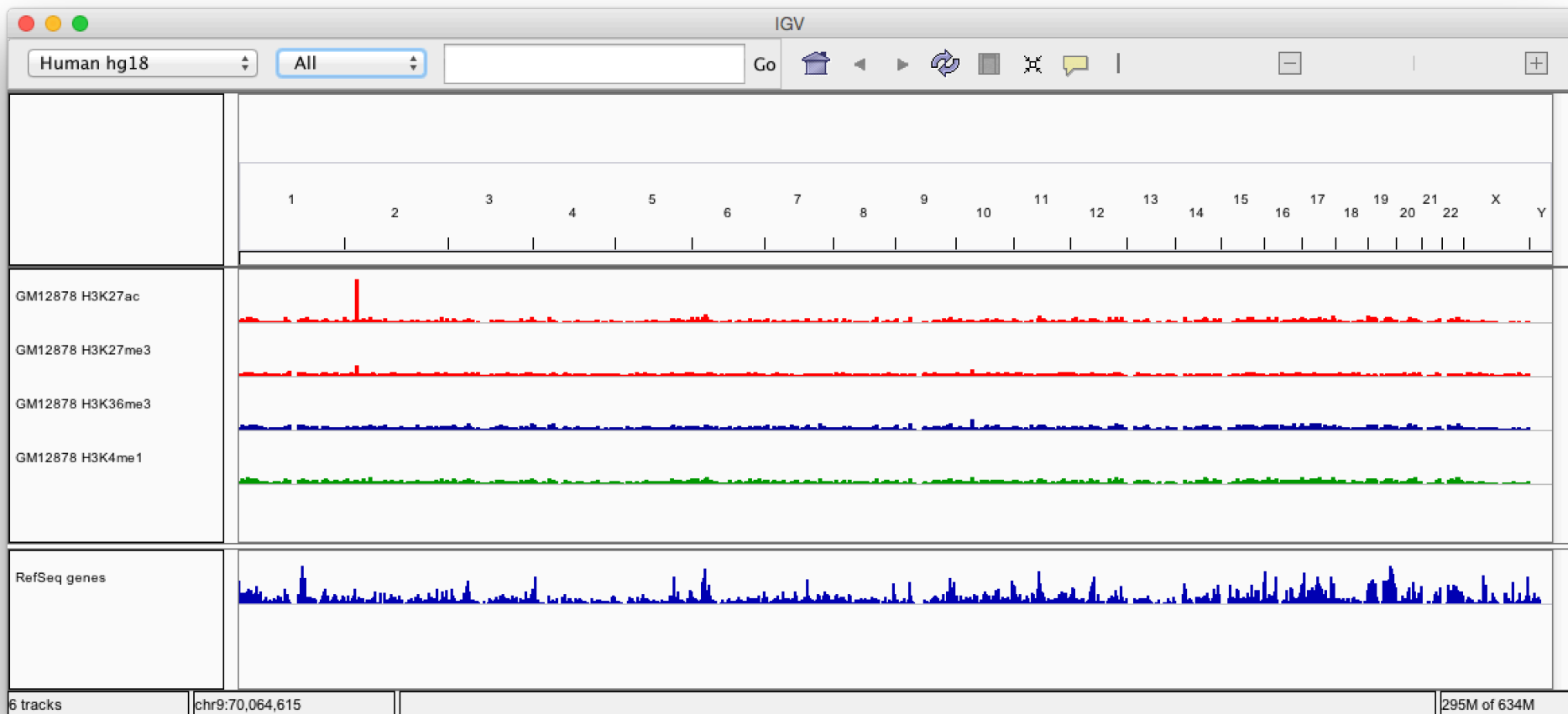
Select **Human hg18**



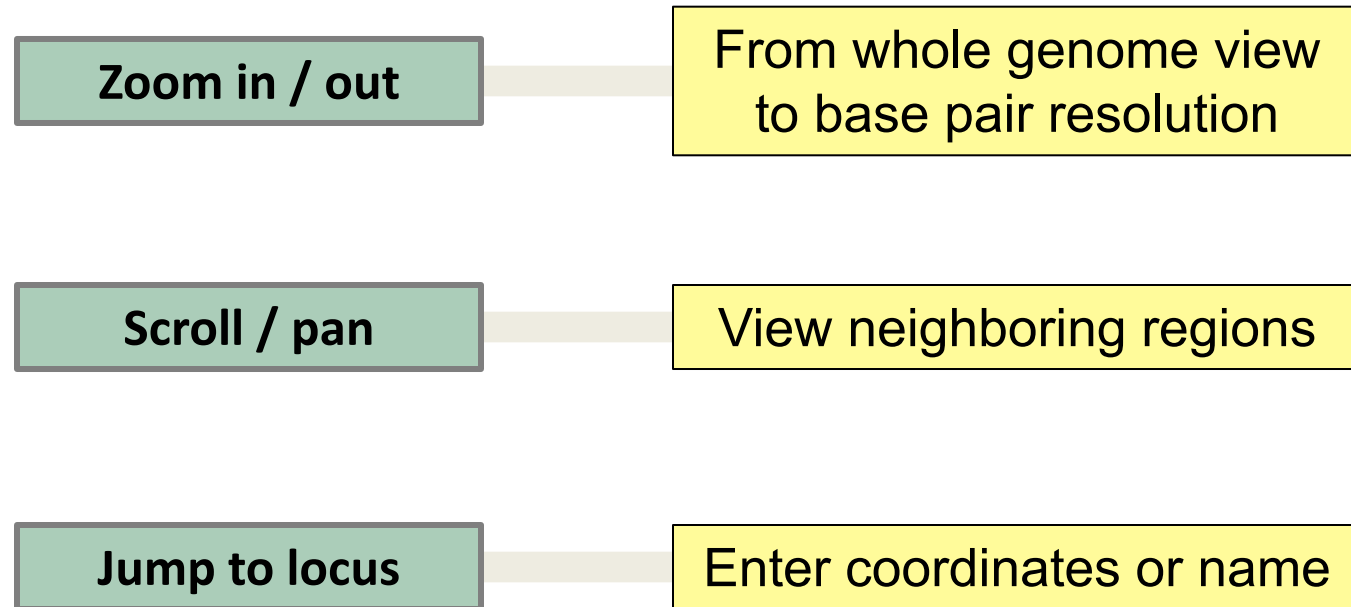
Load data



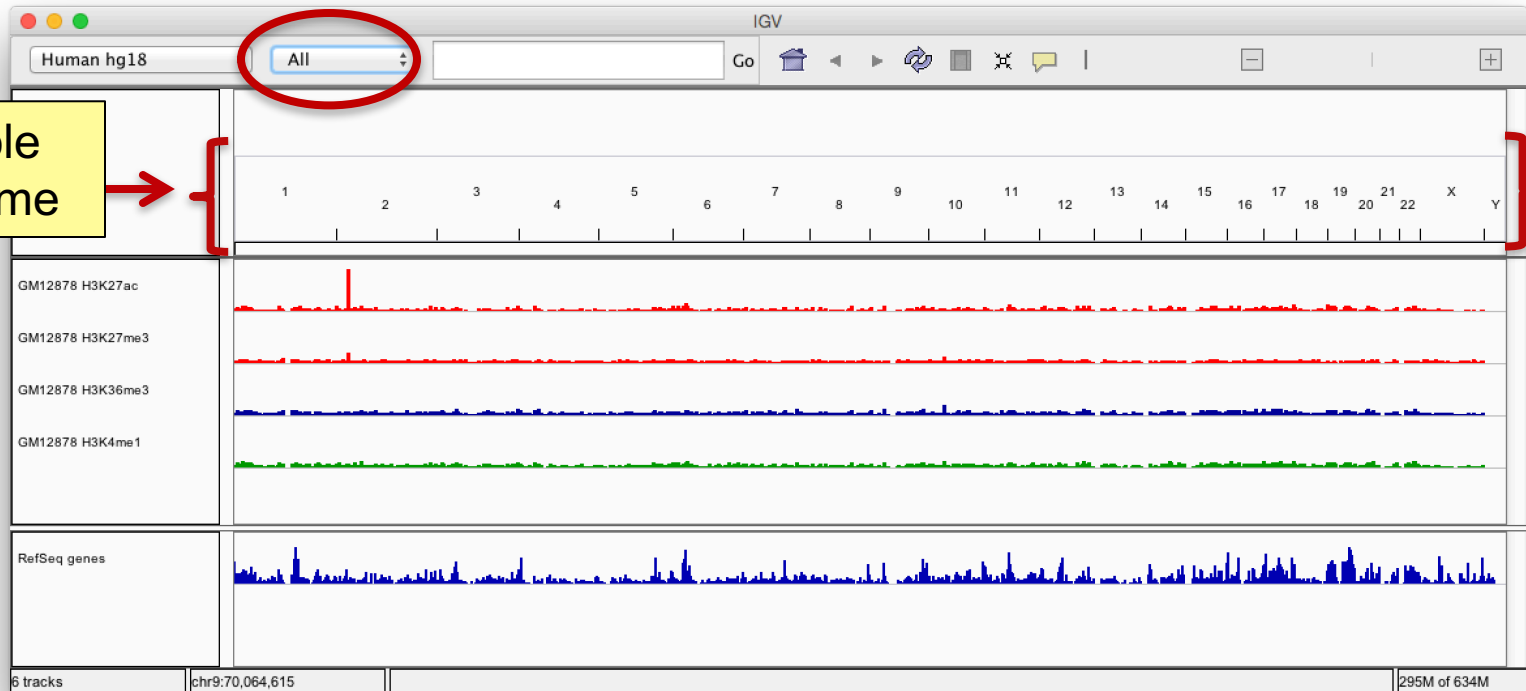
Load data



Navigate



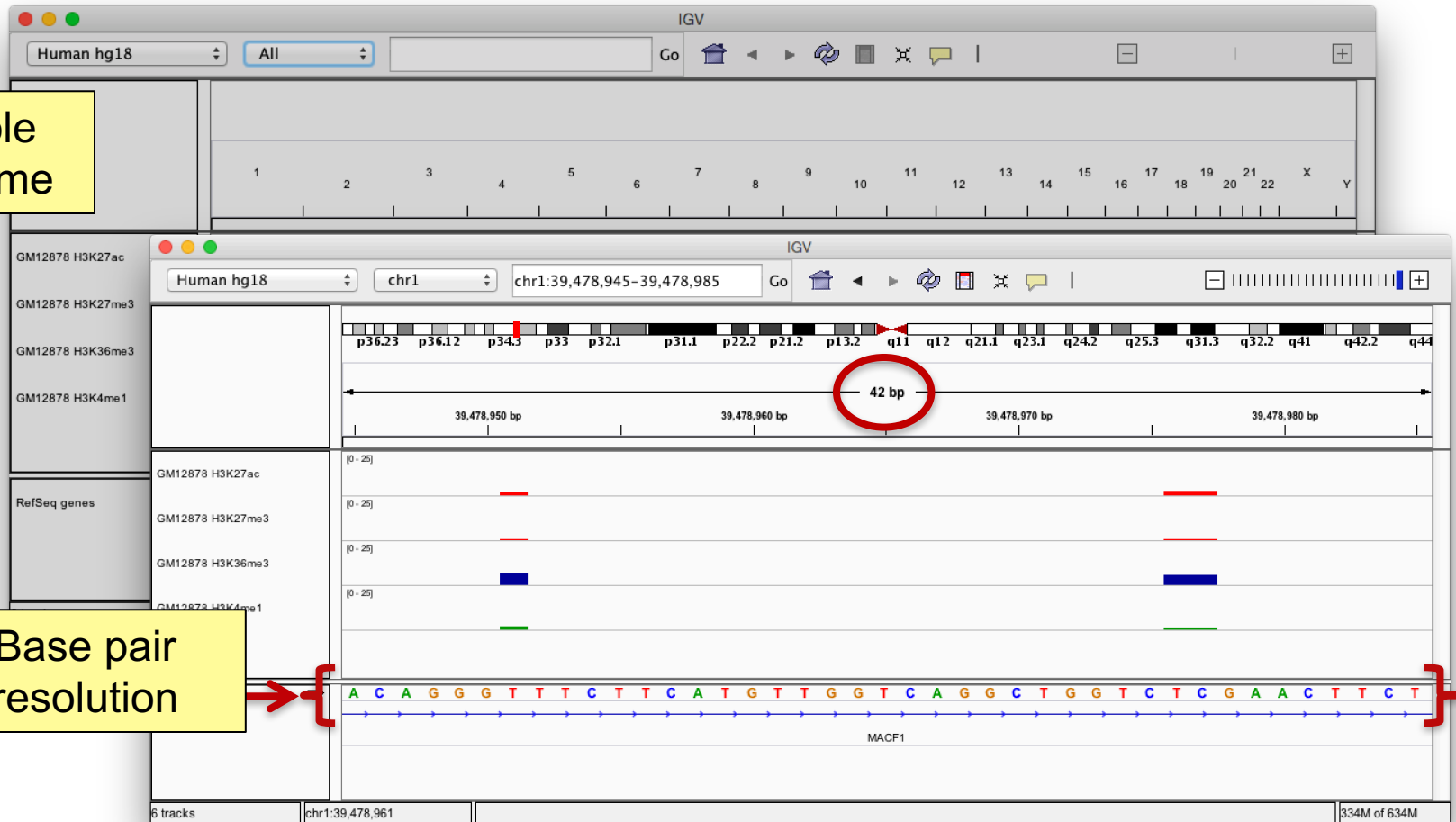
Navigate



Navigate

Whole
genome

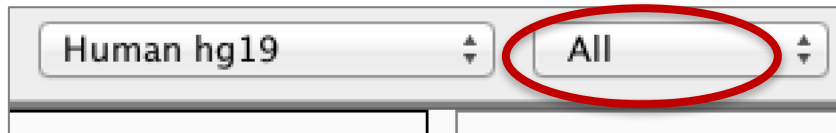
Base pair
resolution



Navigate

View whole
genome

select

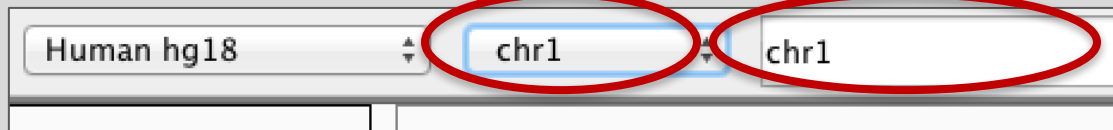


or click



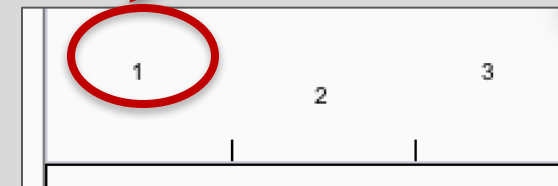
View whole
chromosome

select



or enter

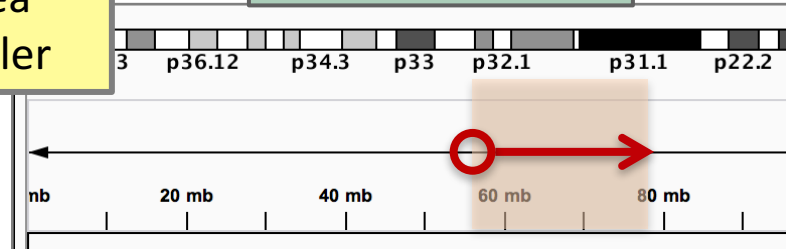
or click



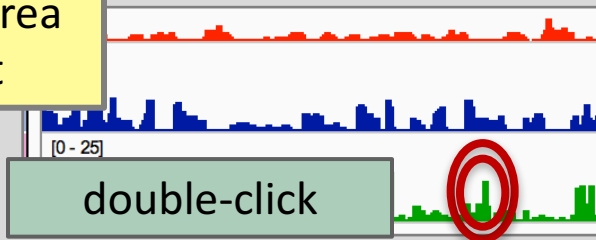
Navigate

Zoom in to area swept out in ruler

click & drag



Zoom in on area of interest



double-click

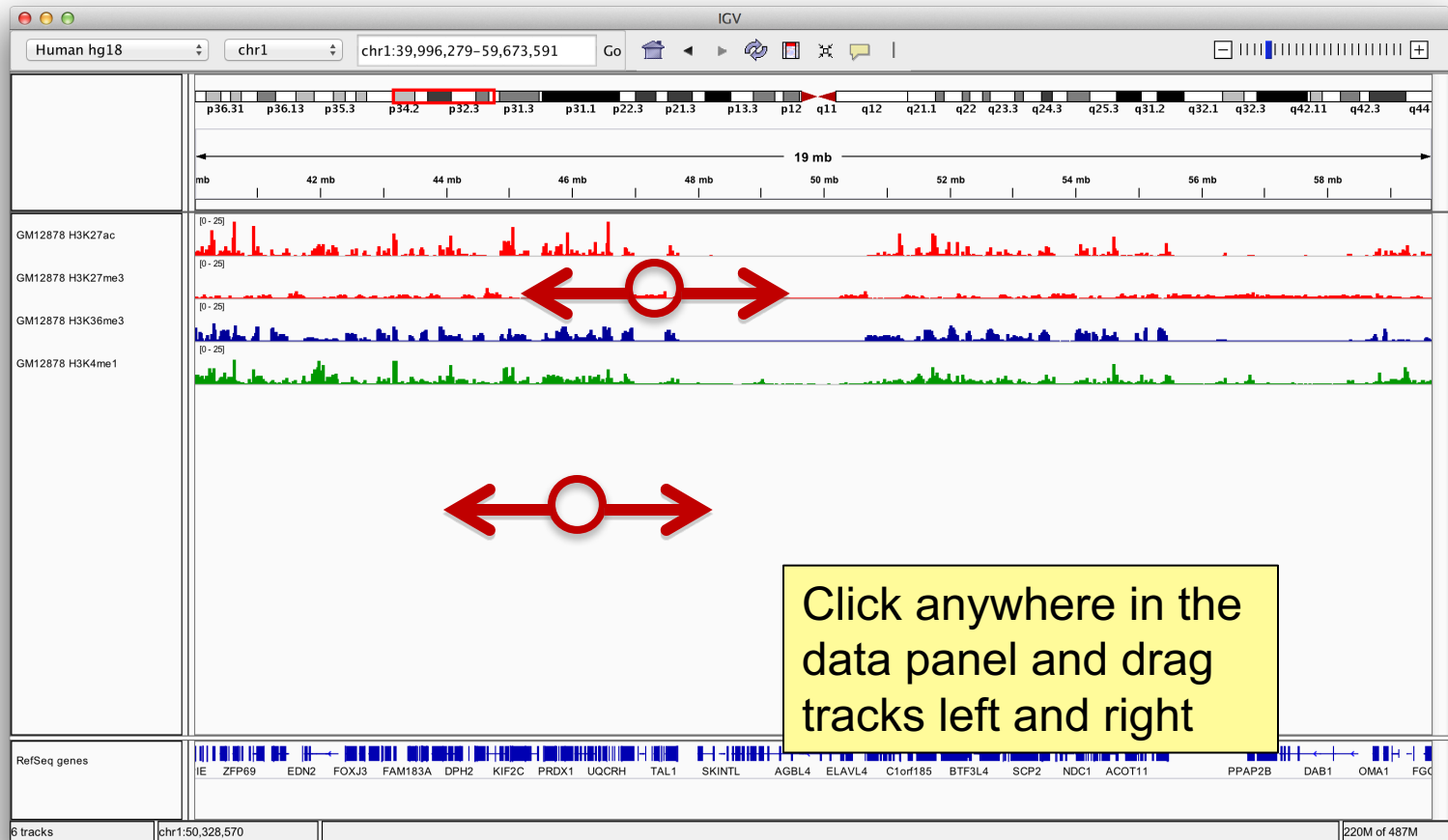
Zoom out

alt-click

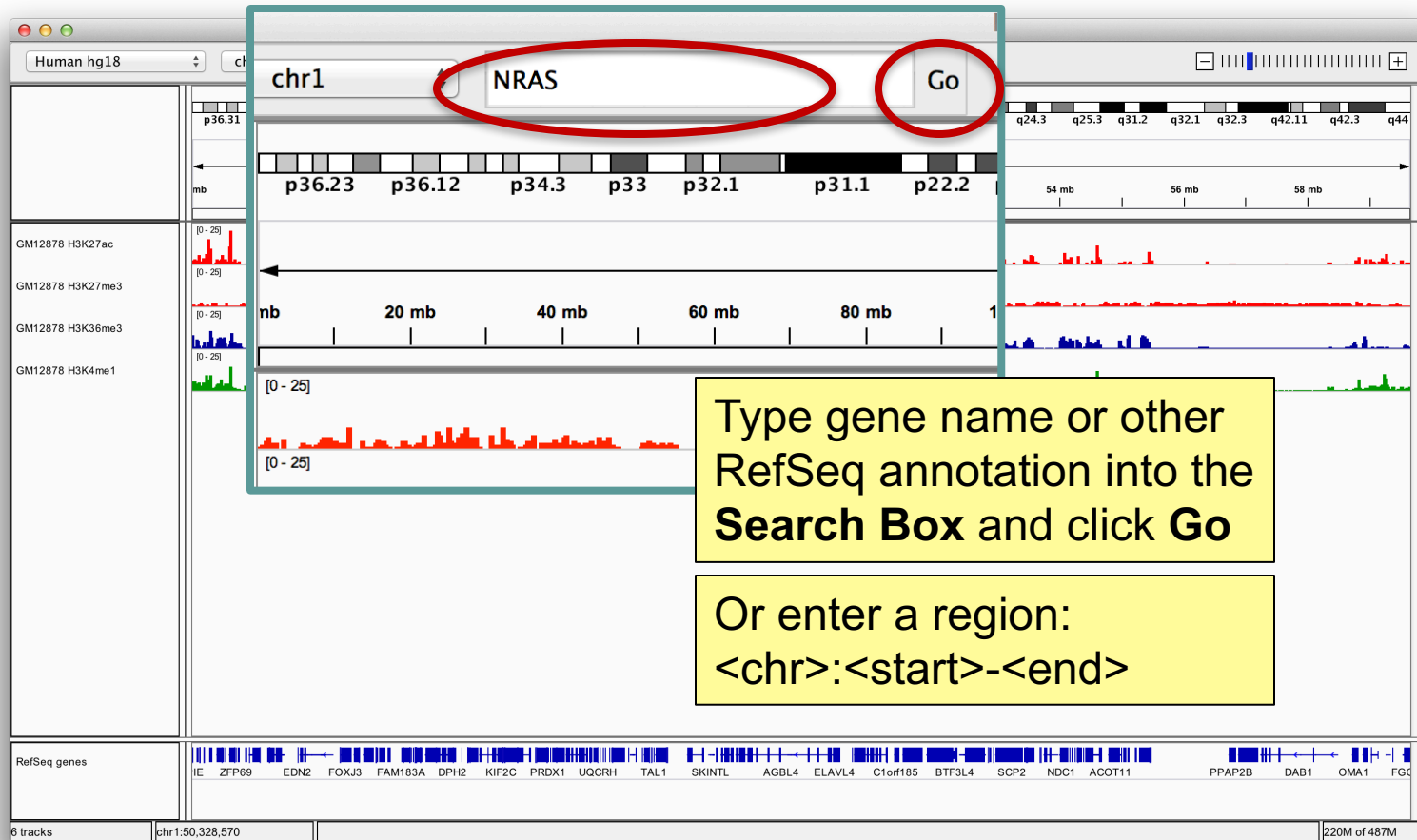
Zoom in or out using the "railroad track"



Navigate

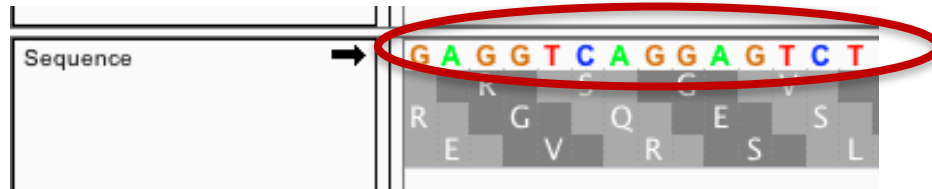


Navigate



Reference sequence

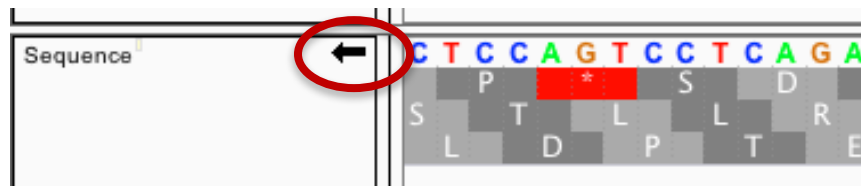
Click anywhere on the sequence to see a 3 frame translation.



By default the sequence for the forward strand is shown.



Click the arrow on the left to reverse the strand.



More basics – loading data

Other common ways of loading data

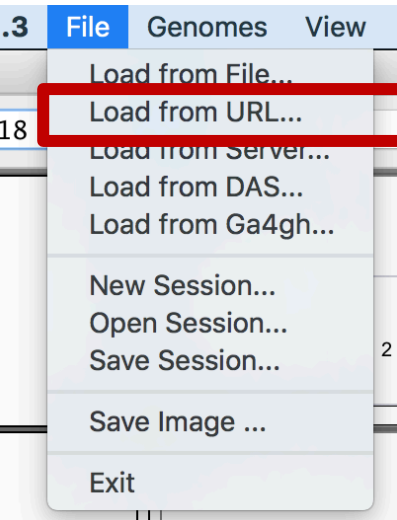
- Files > Load from File
- Files > Load from URL

More basics – loading data

Other common ways of loading data

- Files > Load from File
- Files > Load from URL

Try loading from URL:



1) Select *Files > Load from URL*

2) Enter

<http://www.igv.org/workshops/March2017/signal.tdf>

3) Click “OK”

OR

1) In a web browser, navigate to

<http://www.igv.org/workshops/March2017>

2) Drag the file ***signal.tdf*** and drop it onto the IGV window

More basics – track options

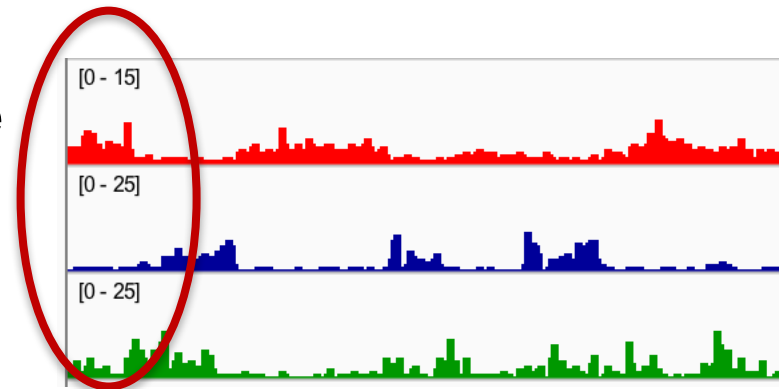
Right-click popup menu = track specific options menu

Common options

- Change track name, color, height
- Remove track

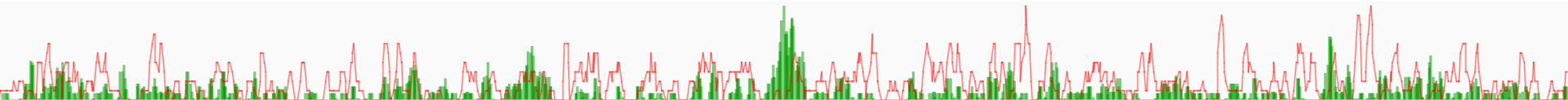
For quantitative data tracks: **Data Range**

- Set to an explicit range
- Enable log scale
- Enable autoscale
- Select multiple tracks and enable “group” autoscale



For quantitative data tracks: **Data Display:**

- Change between bar chart, line plot, point plot, heatmap
- Select multiple tracks and enable “overlay tracks”



More basics – track options

Right-click popup menu = track specific options menu

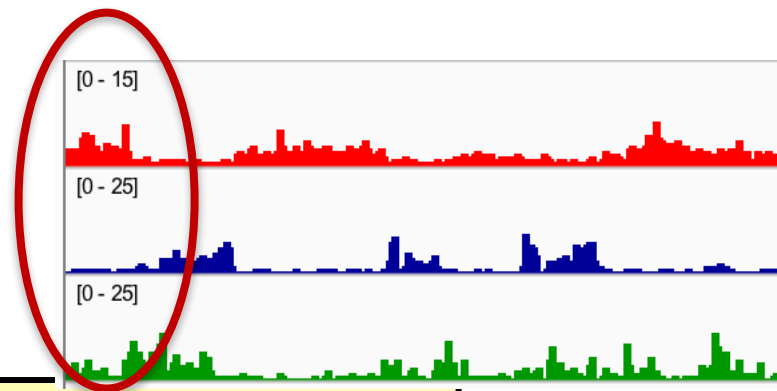
Common options

- Change track name, color, height.
- Remove track

For quantitative data tracks: **Data Range**

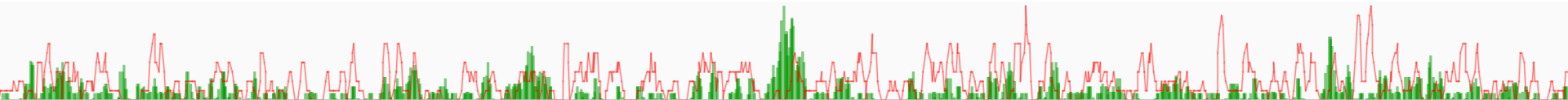
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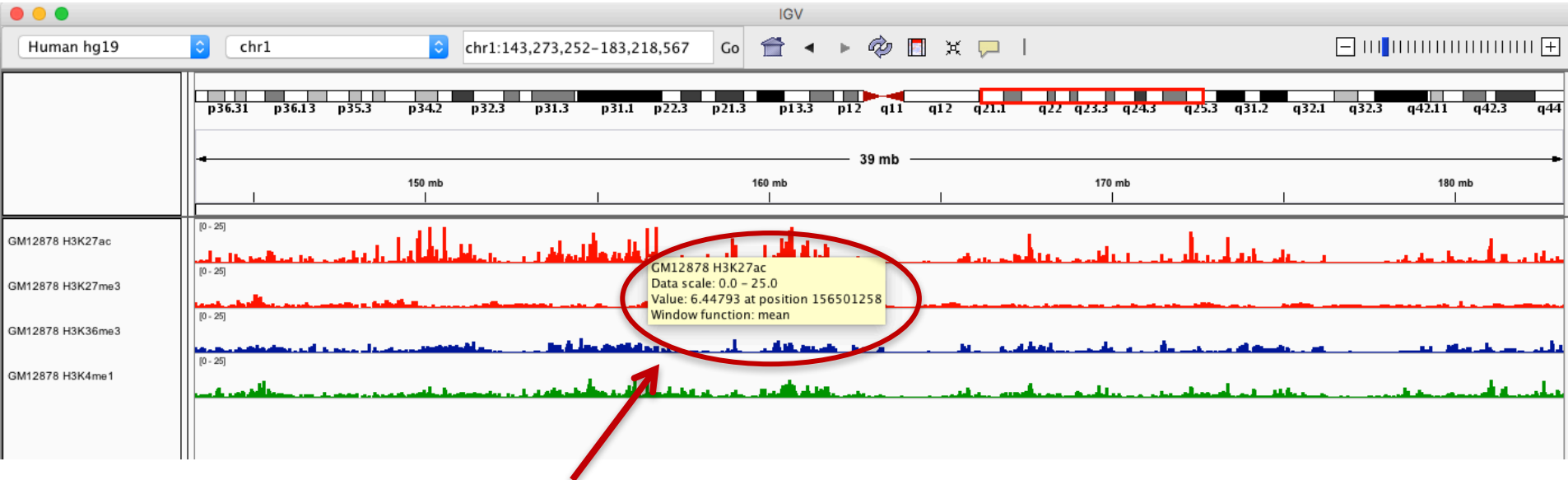


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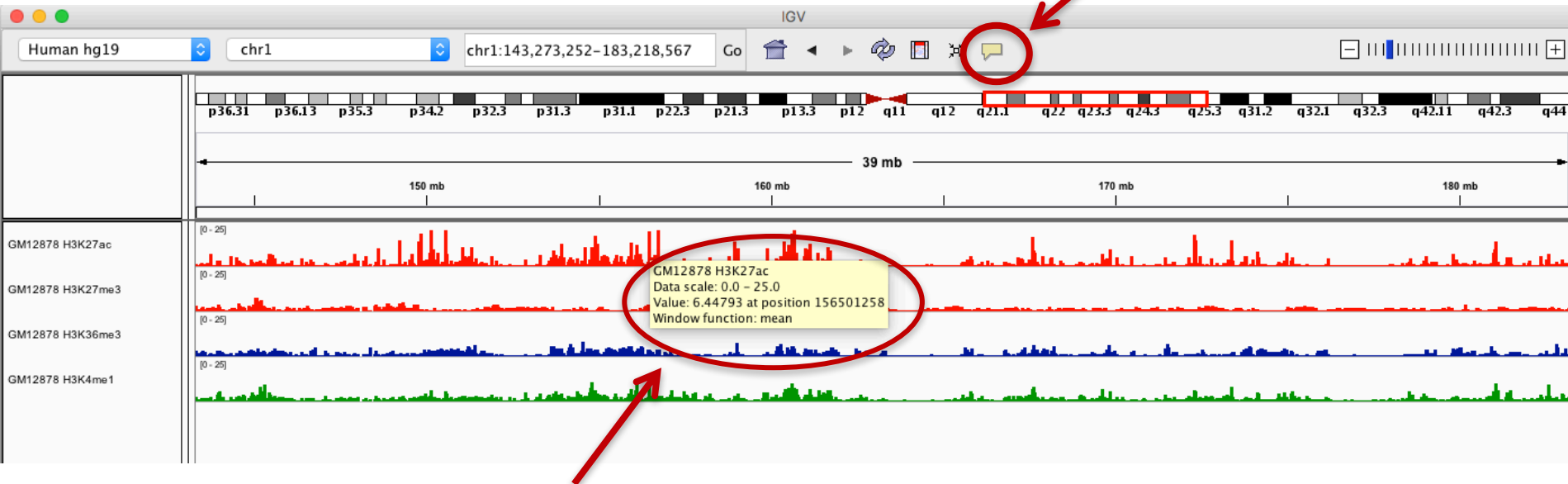
More basics – data details popup



By default, a “data detail” window pops up as the the cursor hovers over a data element in a track

More basics – data details popup

Change the popup behavior by clicking on the yellow balloon icon.
Options are (1) ***On hover*** (2) ***On click*** (3) ***Never***



By default, a “data detail” window pops up as the the cursor hovers over a data element in a track

More basics – the *igv* folder

In your home folder, IGV creates a folder named **igv** to store:

igv.log file

- Logs IGV activity and error messages
- If you send us a bug report or help question, we may ask for a copy of this file
- To start a fresh log, you can delete or rename this file (but not while IGV is running)

prefs.properties file

- Stores all the preference you set in *View > Preferences* so they are remembered across IGV sessions
- You can reset all preferences by deleting or renaming this file (but not while IGV is running)

genomes folder

- Stores info about the genomes you've loaded – for the *genomes dropdown menu*.

Viewing Multiple Regions

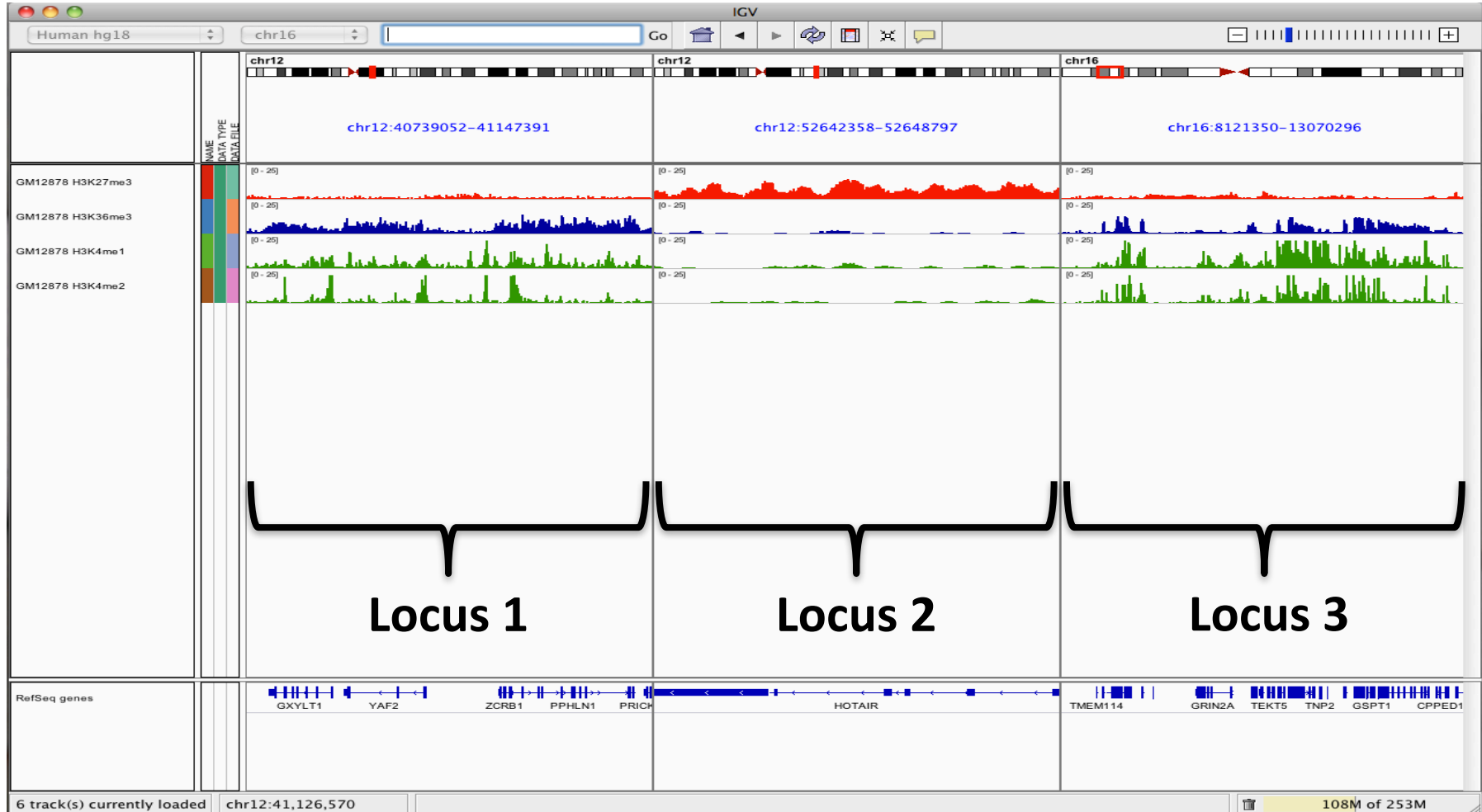
Viewing multiple regions

multi-locus or split-screen view



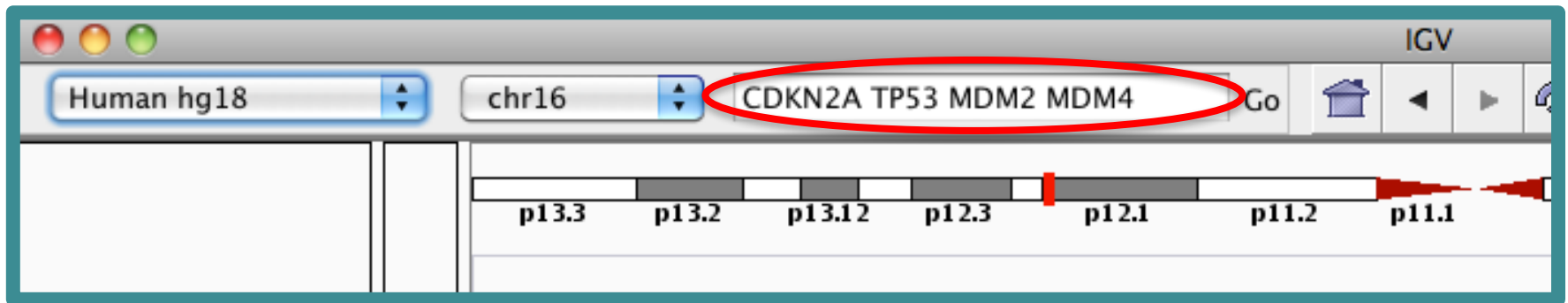
Viewing multiple regions

multi-locus or split-screen view



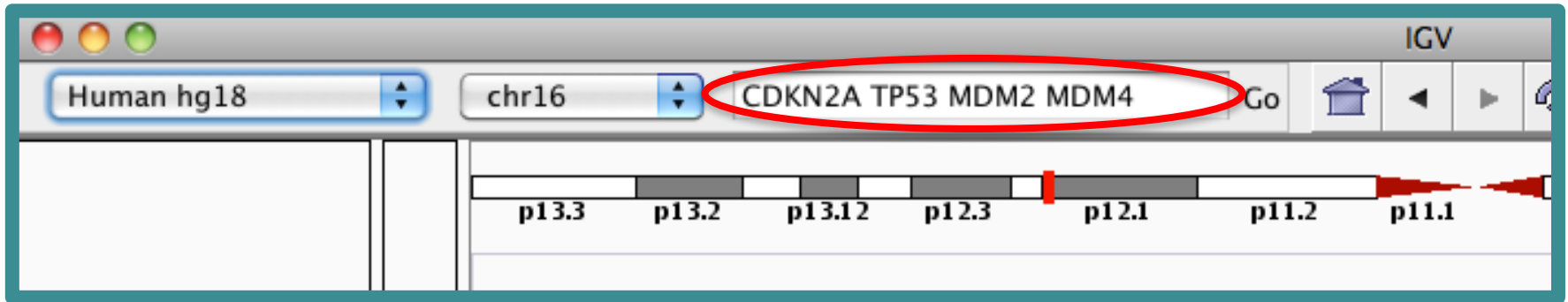
Multiple regions – search box

Enter multiple loci or features in the search box



Multiple regions – search box

Enter multiple loci or features in the search box



For a more persistent list of regions, use ***Regions of Interest***

Multiple regions – “regions of interest” tools

For a more persistent list of regions, use *Regions of Interest*

To **define** regions:

(1) Use the Region of Interest tool

- Click on the tool icon in the tool bar
- Click twice in the data panel to specify the region boundaries



Multiple regions – “regions of interest” tools

For a more persistent list of regions, use *Regions of Interest*

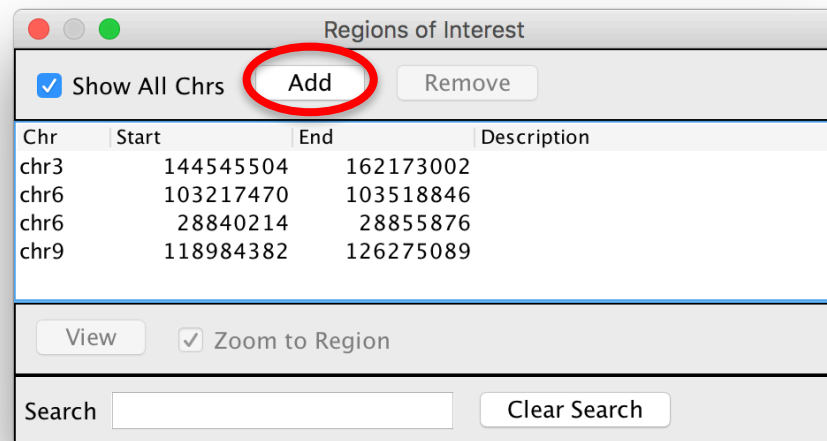
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(2) Use the Region Navigator

- Navigate to the new region
- Select *Regions > Region Navigator*
- Click **Add** to create a region from the current view



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- Select *Regions > Region Navigator*
- Click **Add** to create a region from the current view

(3) **Import a file** of pre-defined regions

- Prepare a .BED file containing the region definitions
- Select *Regions > Import Regions*

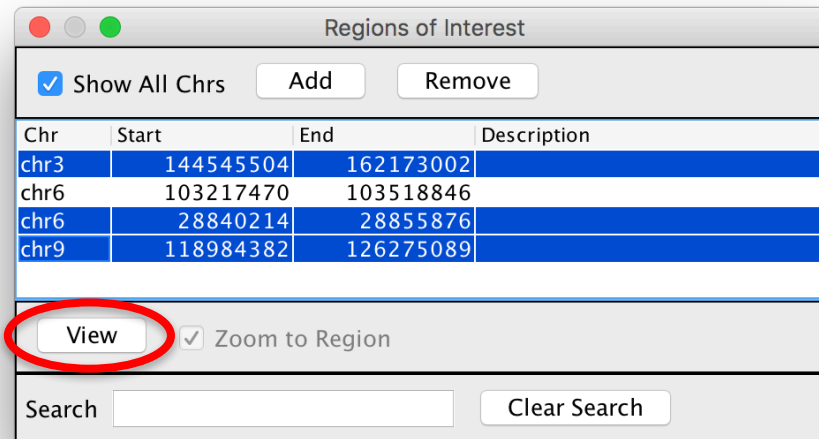
Multiple regions – “regions of interest” tools

For a more persistent list of regions, use *Regions of Interest*

To **view** multiple regions of interest you defined:

Use the Region Navigator

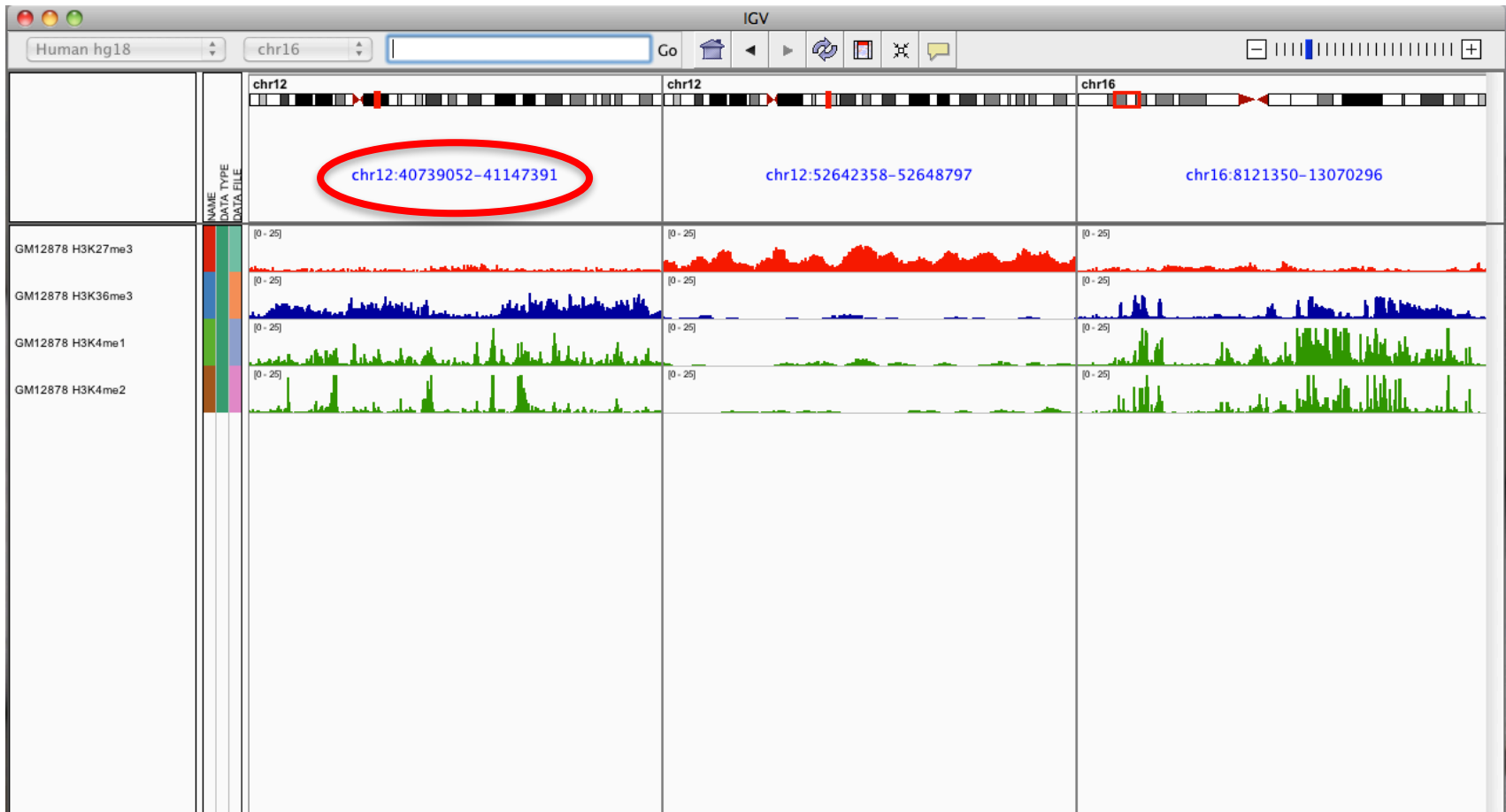
- Select *Regions > Region Navigator*
- Select 2 or more regions
- Click **View**



Viewing multiple regions

To go back to the standard, single-region view:

- *double-click* on a region label – or –
- *right-click* and select **Switch to standard view**

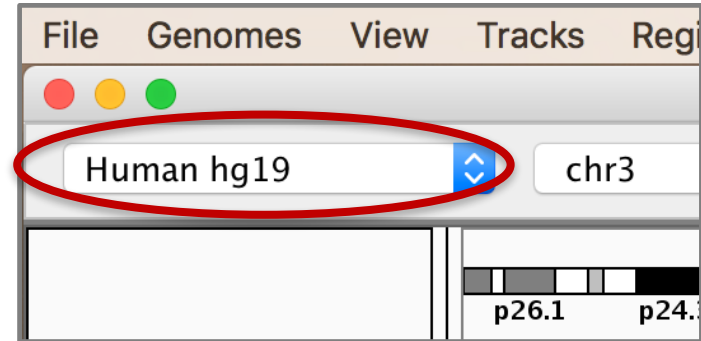


more about loading

Reference Genomes

Genome menu

The genome drop-down menu is in the upper left corner of the IGV window.



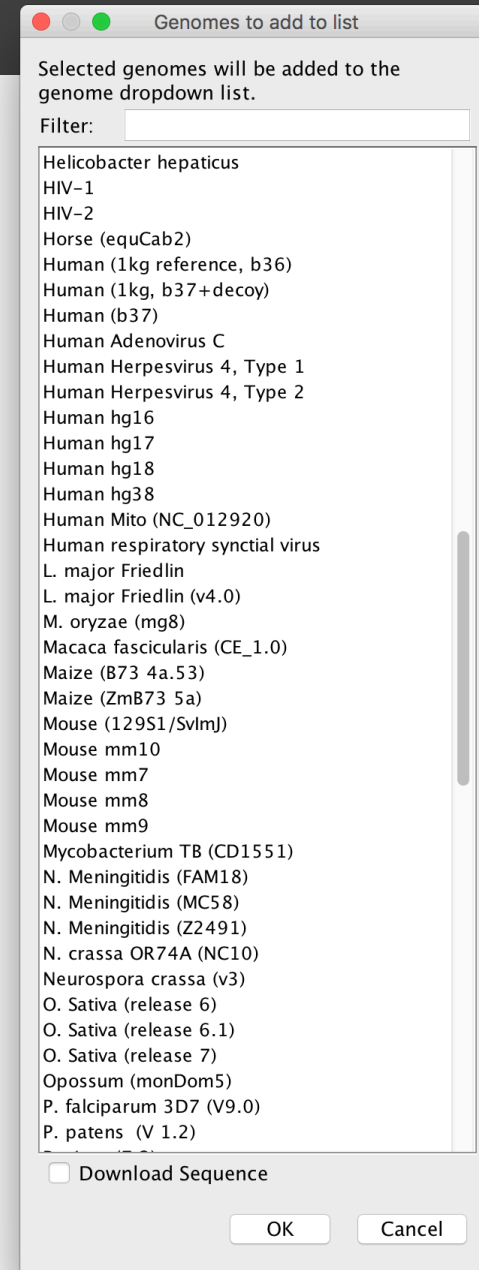
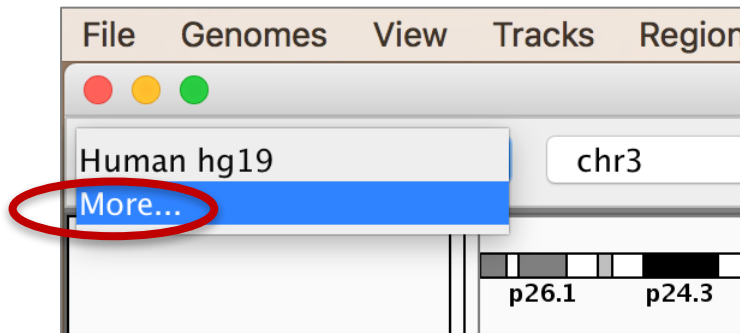
The genome menu is short when you first install IGV.

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

More hosted genomes

To get more genomes from the IGV server:

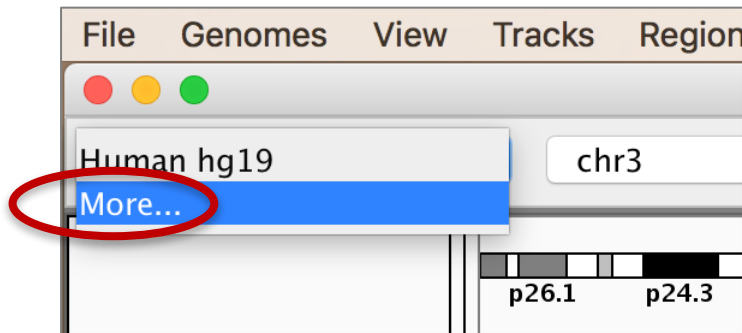
- (1) Select **More** from the genome menu to see the complete list



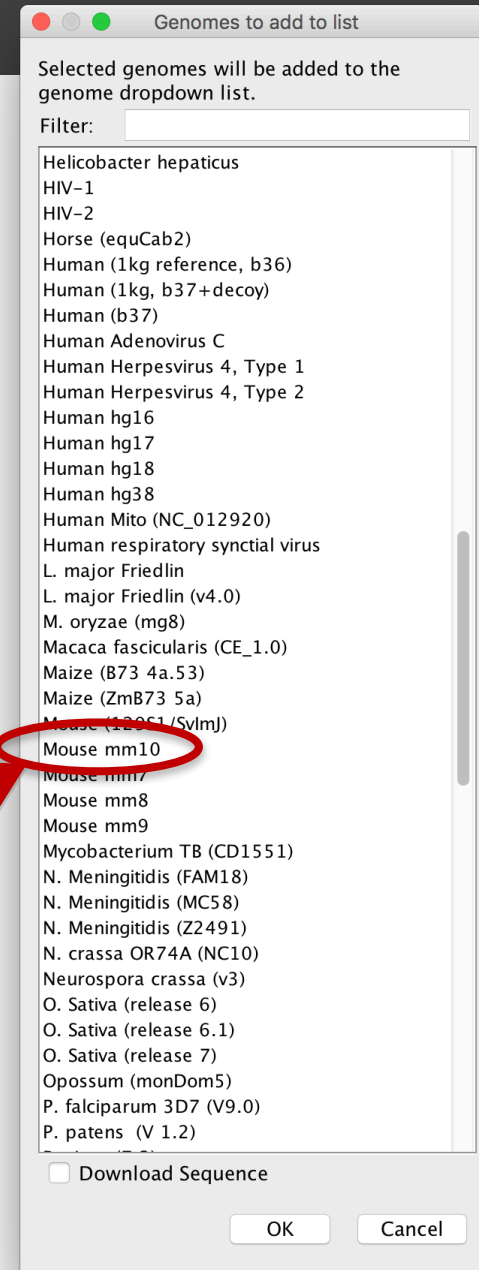
More hosted genomes

To get more genomes from the IGV server:

- (1) Select **More** from the genome menu to see the complete list



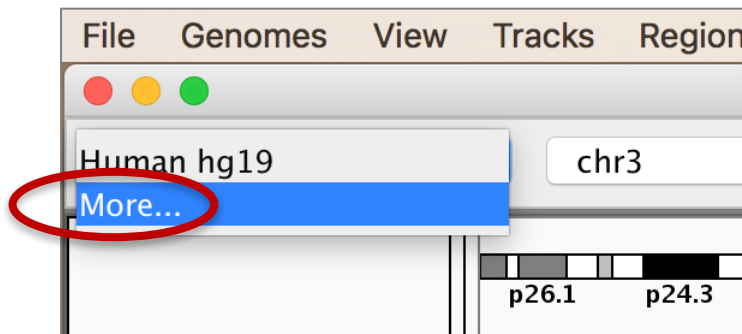
- (2) Click on entry in the list, e.g. **Mouse mm10**



More hosted genomes

To get more genomes from the IGV server:

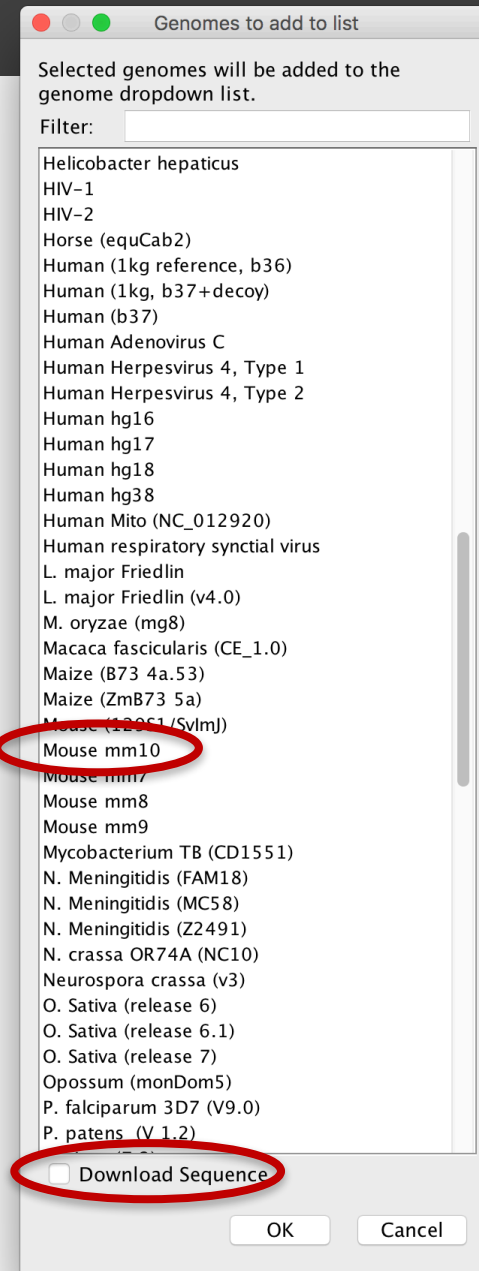
- (1) Select **More** from the genome menu to see the complete list



- (2) Click on entry in the list, e.g. **Mouse mm10**

- (3) Optionally select **Download Sequence**

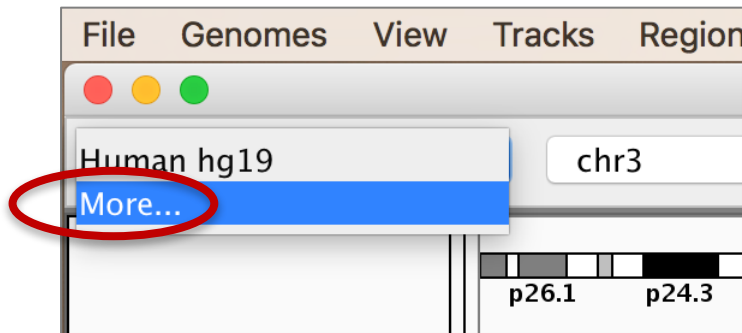
Do **not** download the sequence today!



More hosted genomes

To get more genomes from the IGV server:

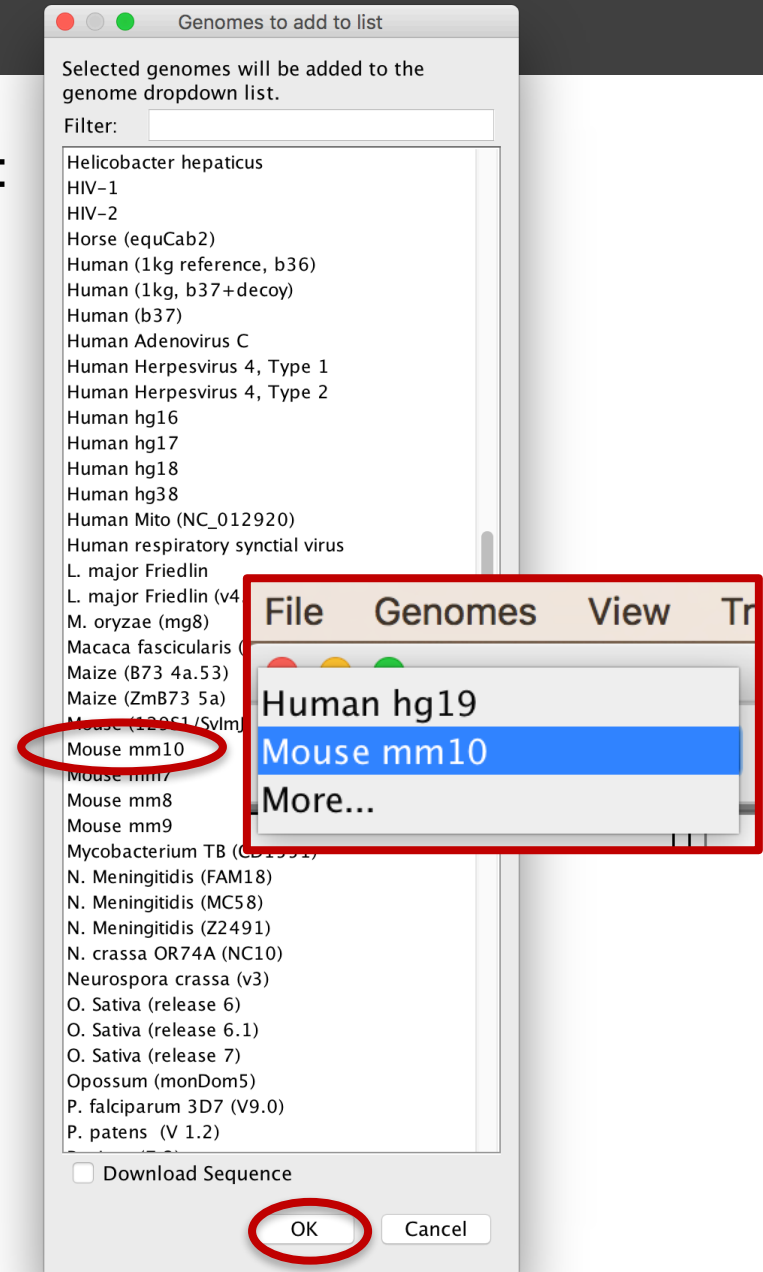
(1) Select **More** from the genome menu to see the complete list



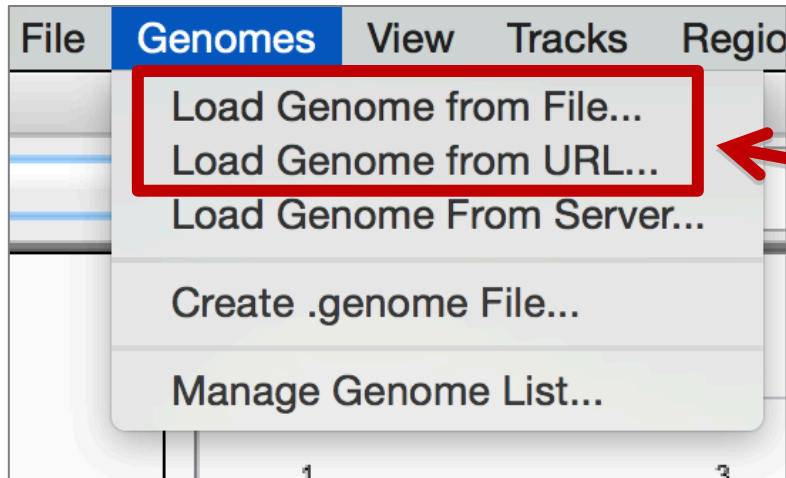
(2) Click on entry in the list, e.g. **Mouse mm10**

(3) Optionally select **Download Sequence**

(4) Click **OK**

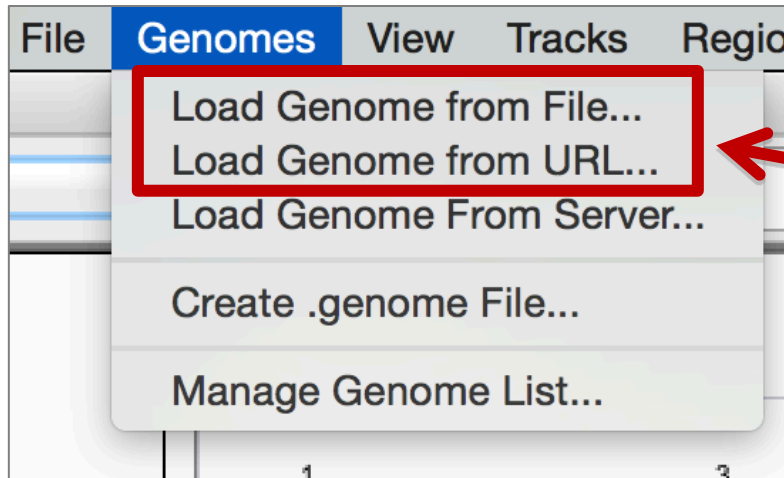


Loading other genomes



Use **Genomes** menu
to load genome from
an indexed FASTA file

Loading other genomes



Use **Genomes** menu to load genome from an indexed FASTA file

- (1) Use *samtools faidx* command to create .fai index file
See github.com/samtools/samtools for info about samtools
- (2) If no index file found, IGV will try to create the index upon load.

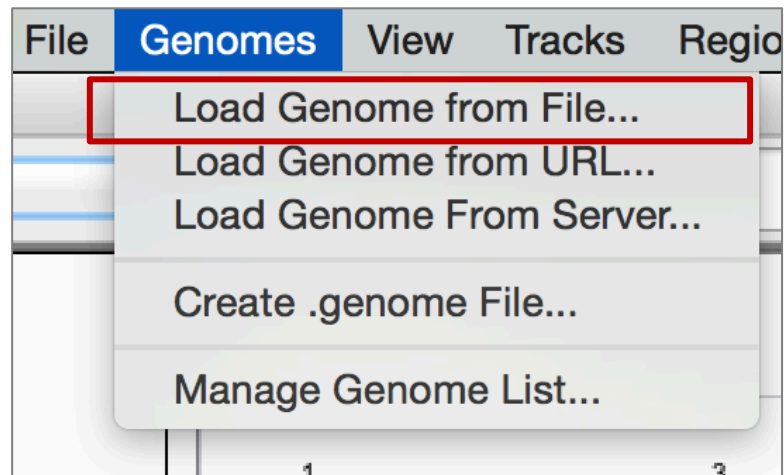
Load genome from file

Hands-on exercise

See handout

Load genome from file

Select **Genomes** > *Load Genome from File* and open **.fasta** file.



Load gene annotation from file

Select ***Files*** > *Load from File*

and open annotation file (e.g. BED, or GTF)

Load gene annotation from file

Select **Files** > *Load from File*

and open annotation file (e.g. BED, or GTF)

[Home](#) [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 form, 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: **genome:** **assembly:**

group: **track:**

table:

region: ☒ genome ☐ ENCODE Pilot regions ☐ position

identifiers (names/accessions):

filter:

intersection:

output format: ☐ [Galaxy](#) ☐ [GREAT](#) ☐ [GenomeSpace](#)

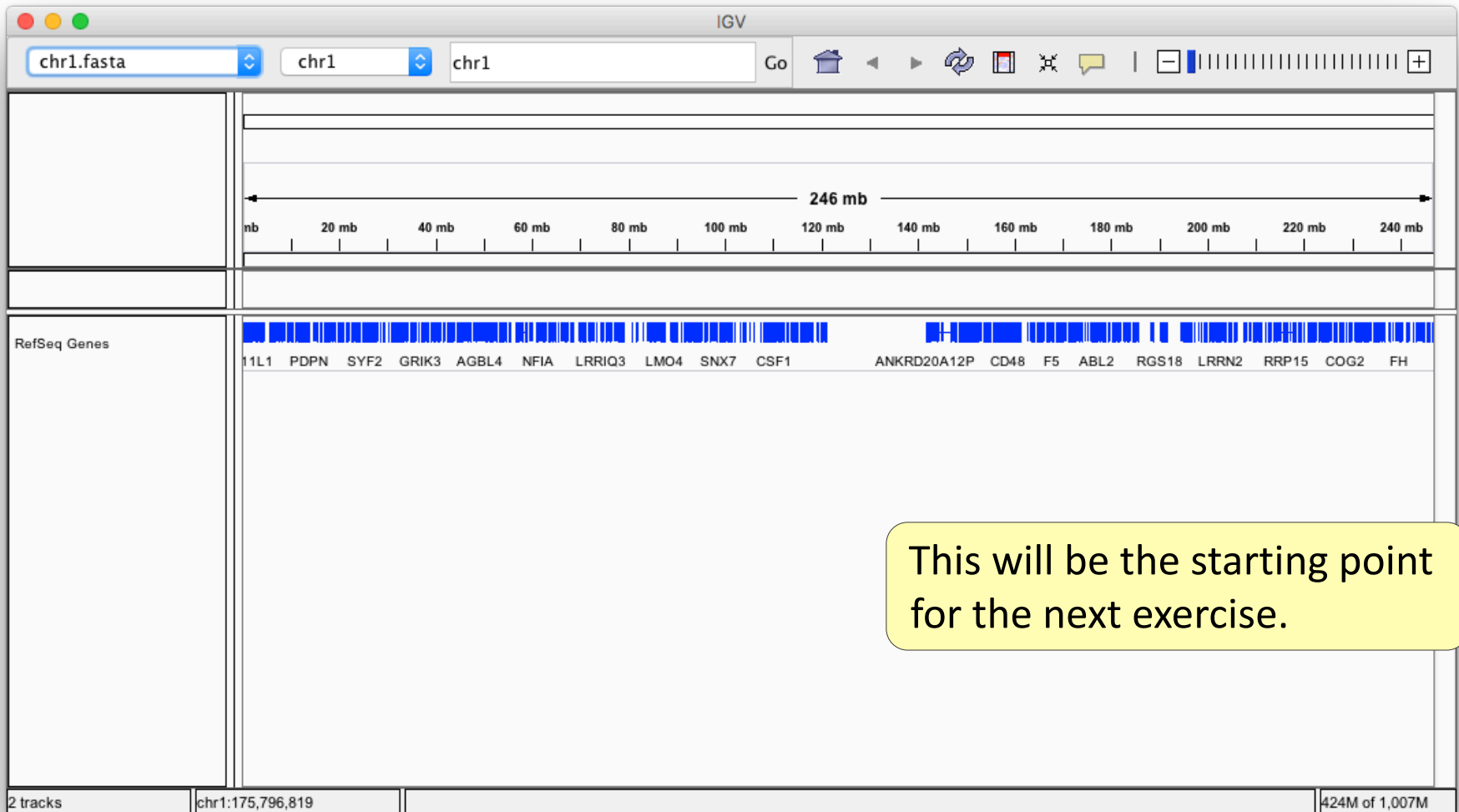
output file: (leave blank to keep output in browser)

file type returned: ☒ plain text ☐ gzip compressed

<http://genome.ucsc.edu/cgi-bin/hgTables>

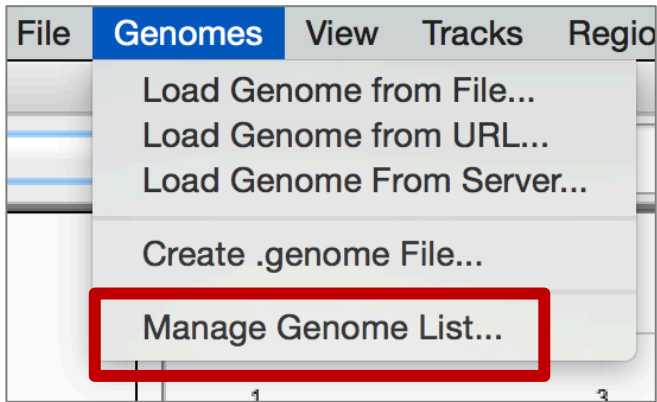
Load genome from file

Hands-on exercise



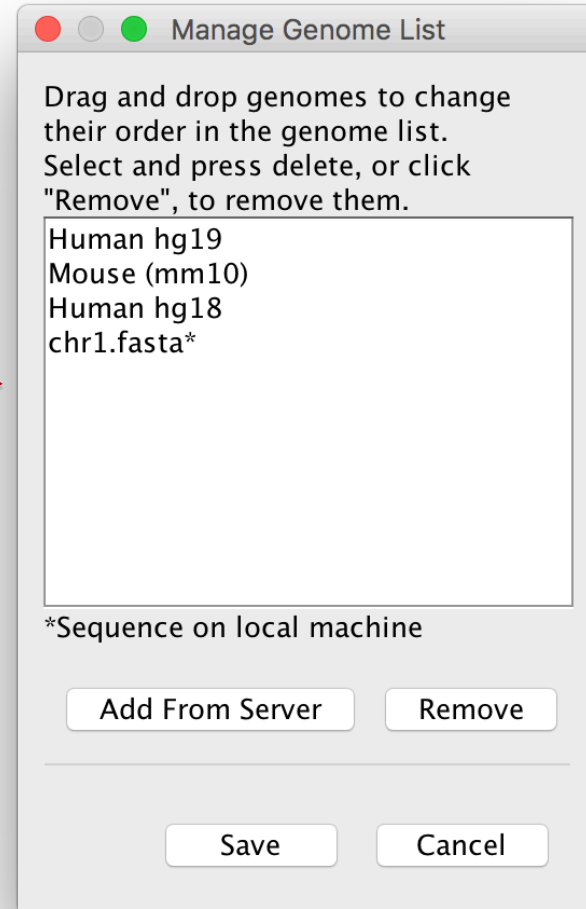
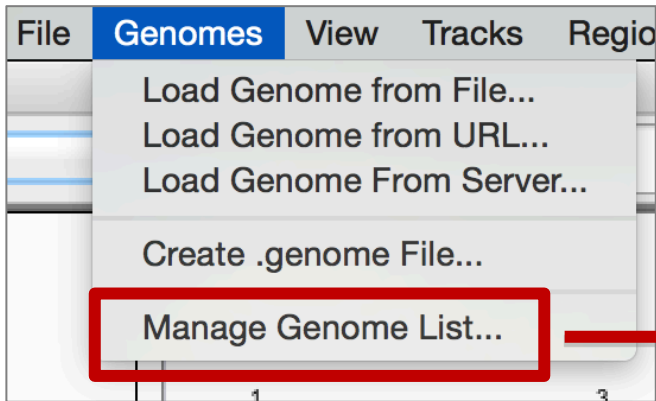
Removing genome from list

Genomes > Manage Genome List



Removing genome from list

Genomes > Manage Genome List

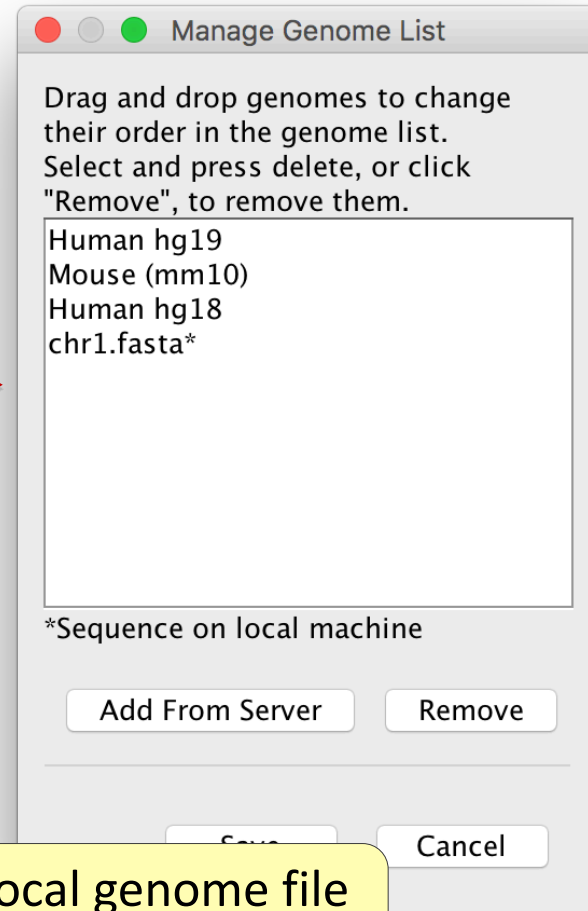
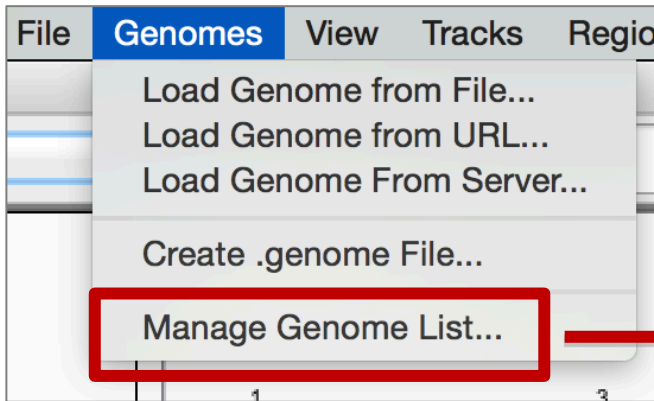


- Remove genomes from list
- Rearrange list
- Add to list

Do **not** remove chr1.fasta now.
We're using it for the next exercise.

Removing genome from list

Genomes > Manage Genome List



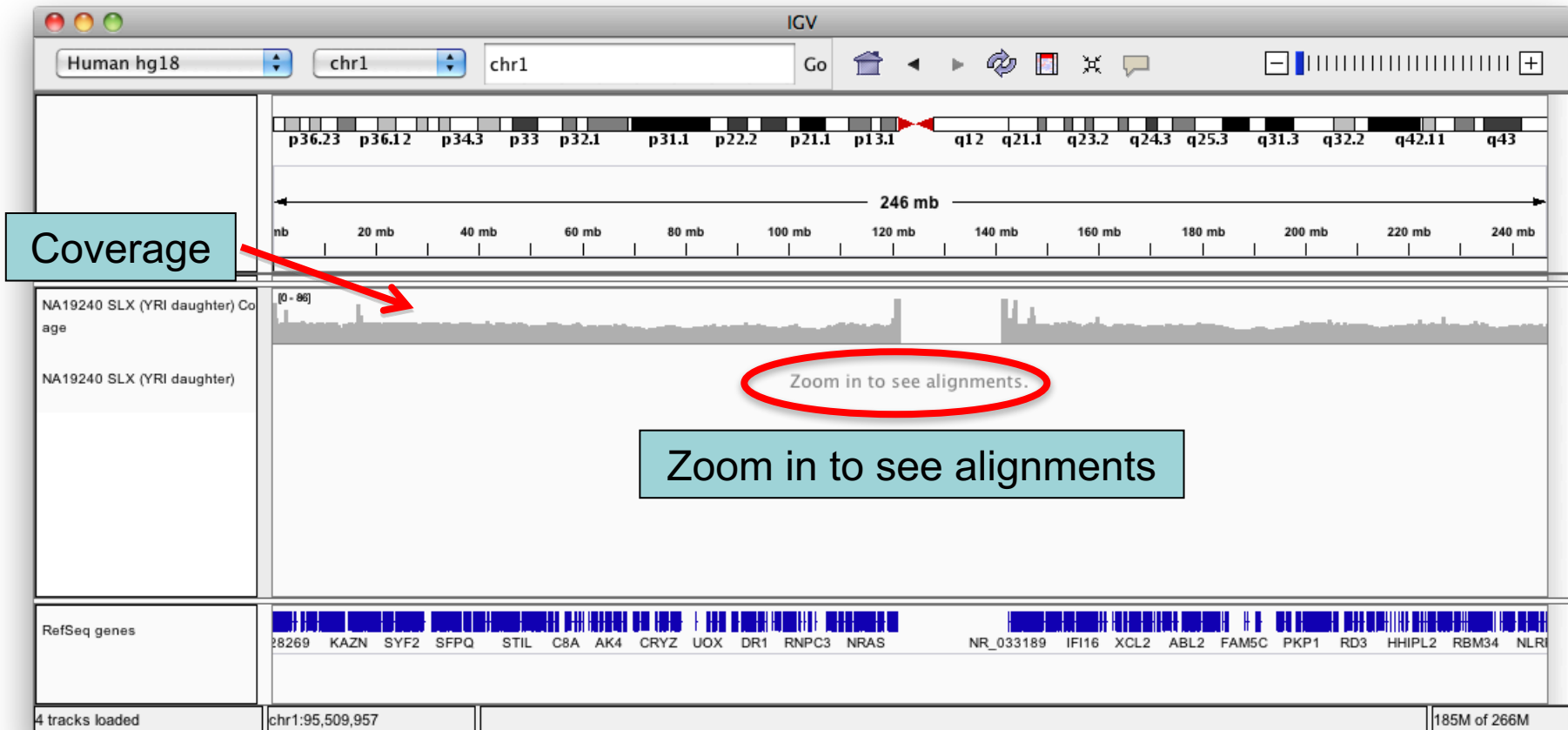
- Remove genomes from list
- Rearrange list
- Add to list

Note: IGV can get confused if you delete a local genome file from the file system but not from the IGV menu

Viewing Next Generation Sequencing (NGS) Data

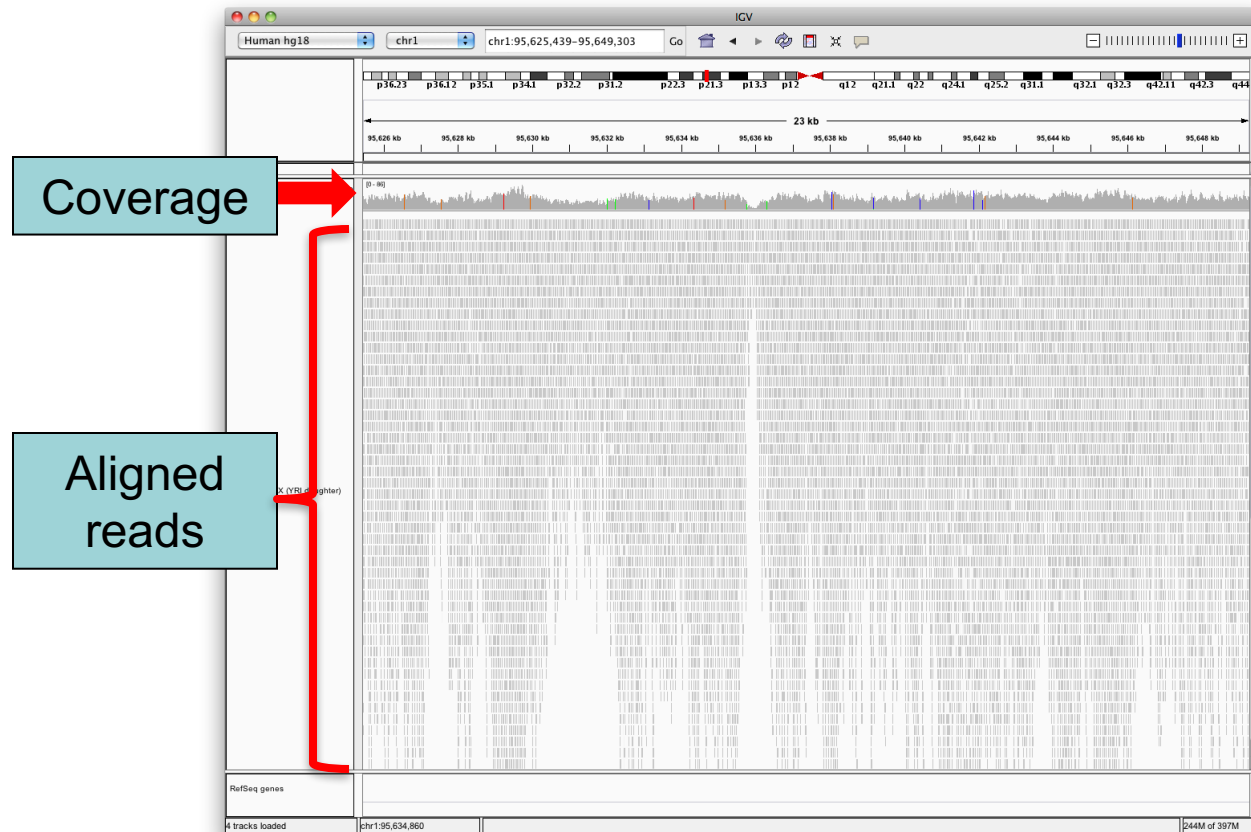
Viewing alignments

Whole chromosome view



Viewing alignments

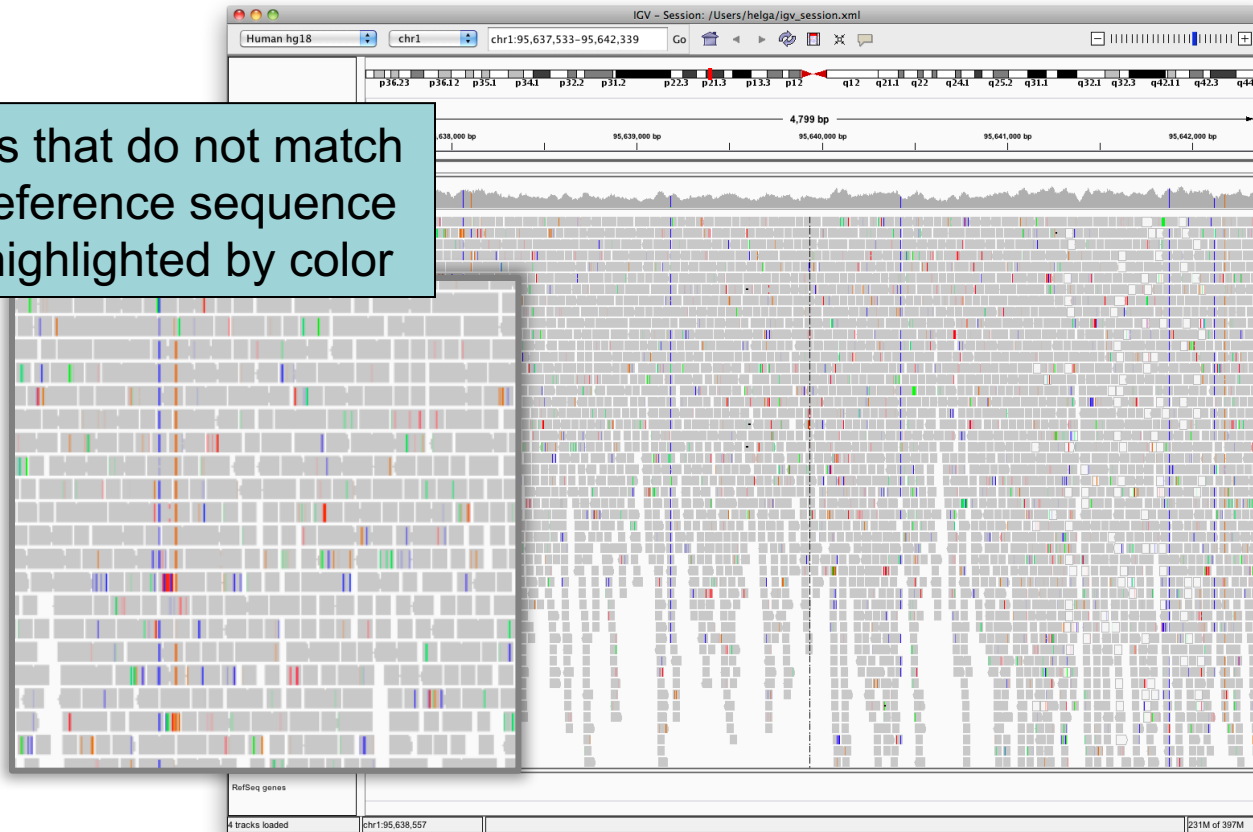
Zoom in to view alignments



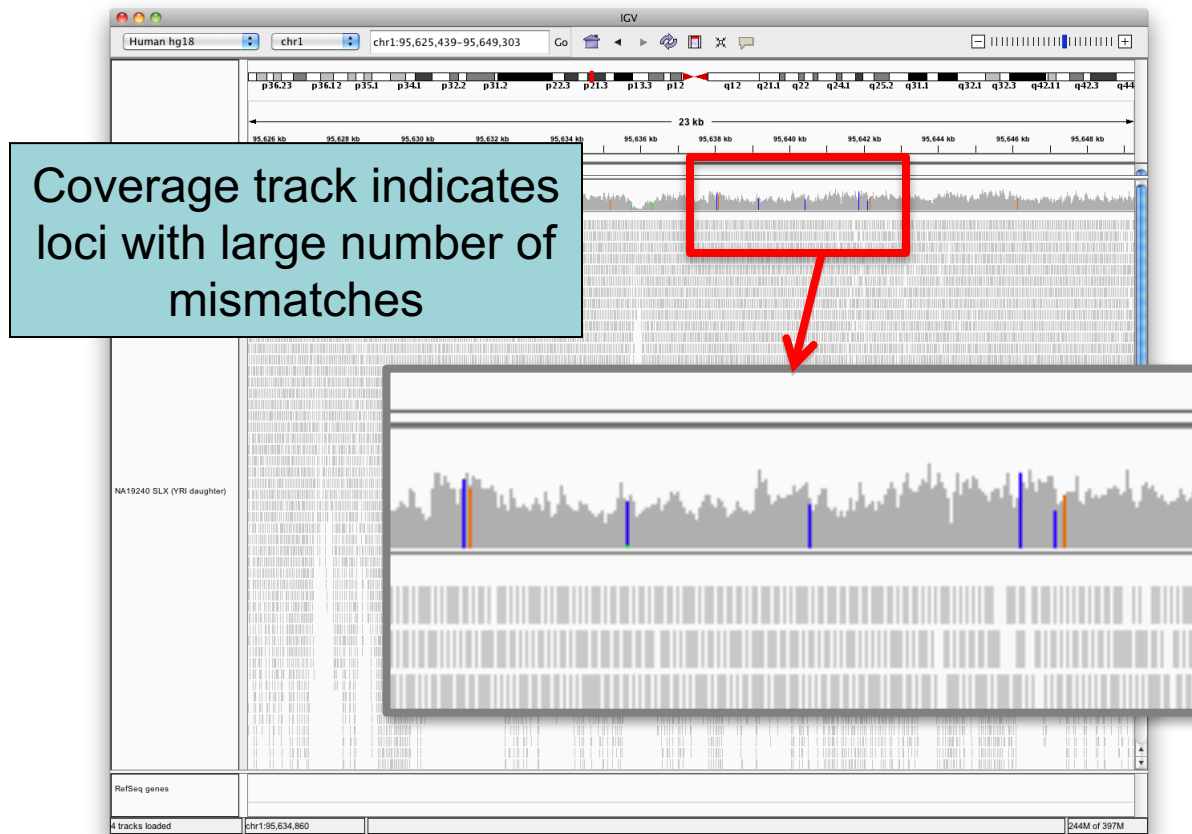
Viewing alignments

Zoom in to see more detail

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



Viewing alignments

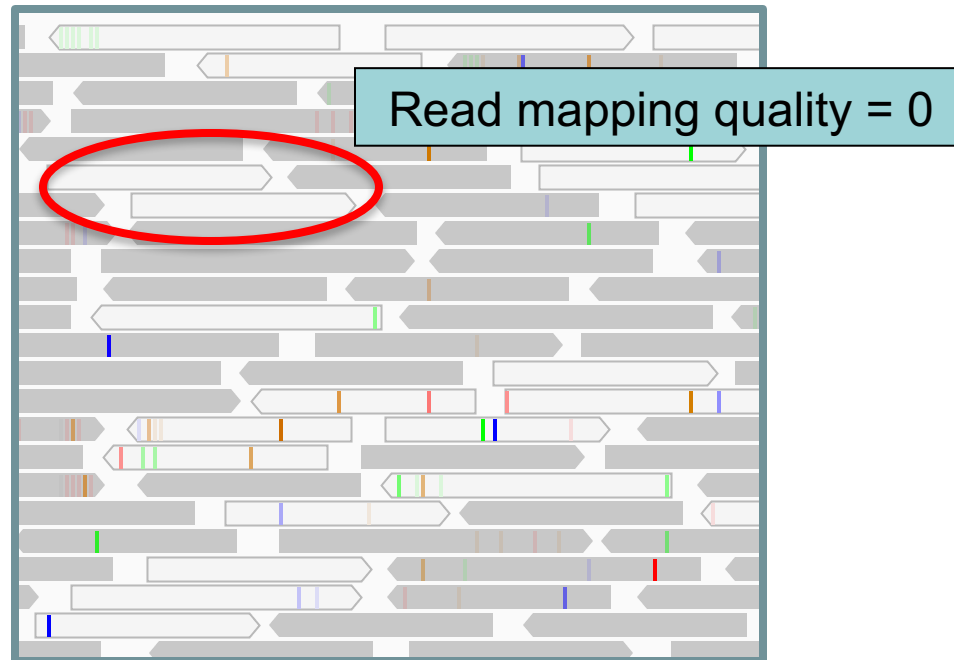


Viewing alignments

Zoom in to see more detail

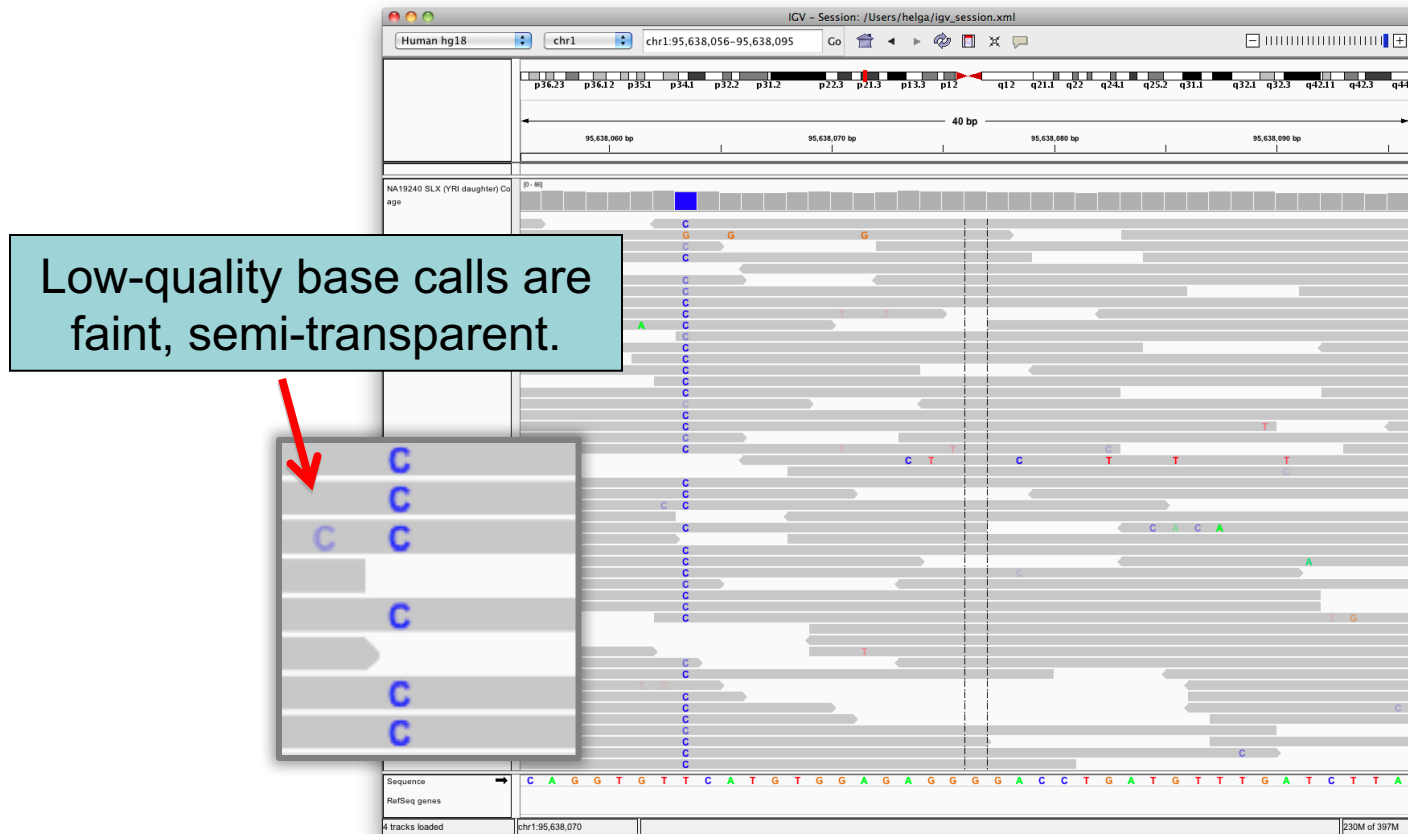


Viewing alignments



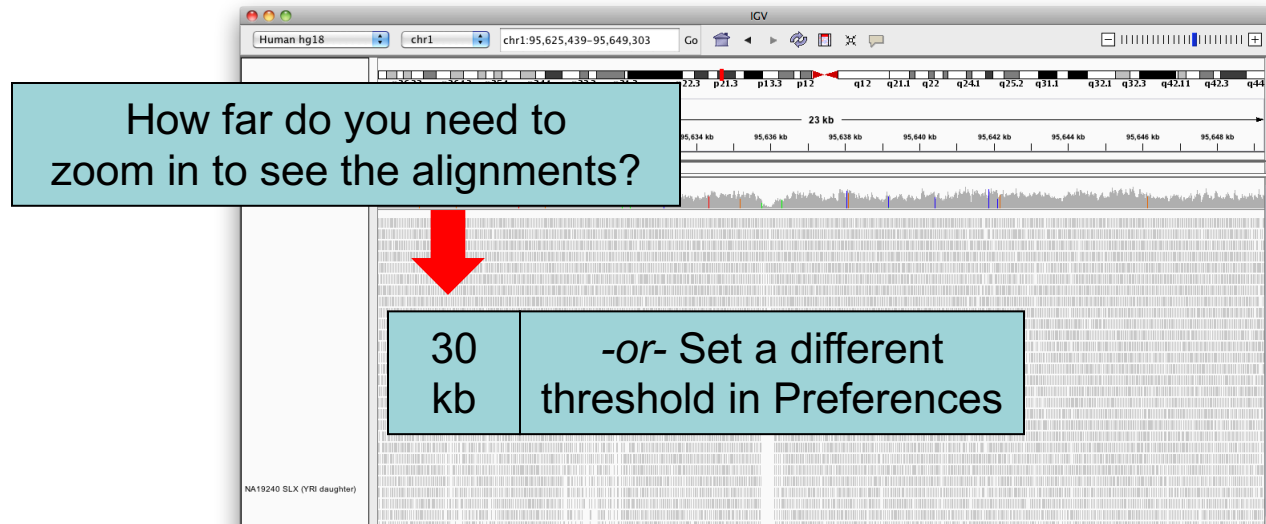
Viewing alignments

Zoom in to see more detail



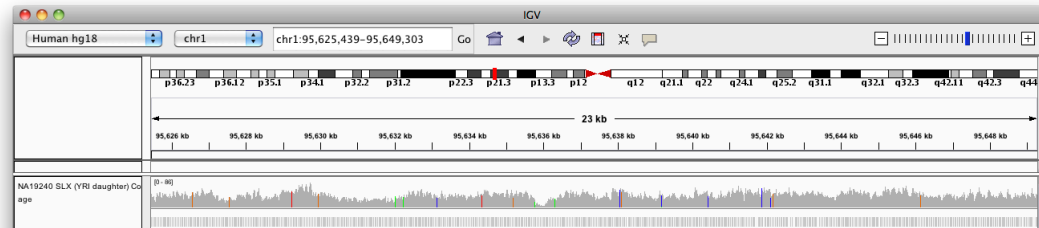
Viewing alignments

Zoom in to view alignments



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

Viewing alignments



For very deep coverage files: use low visibility threshold value

or limit the depth

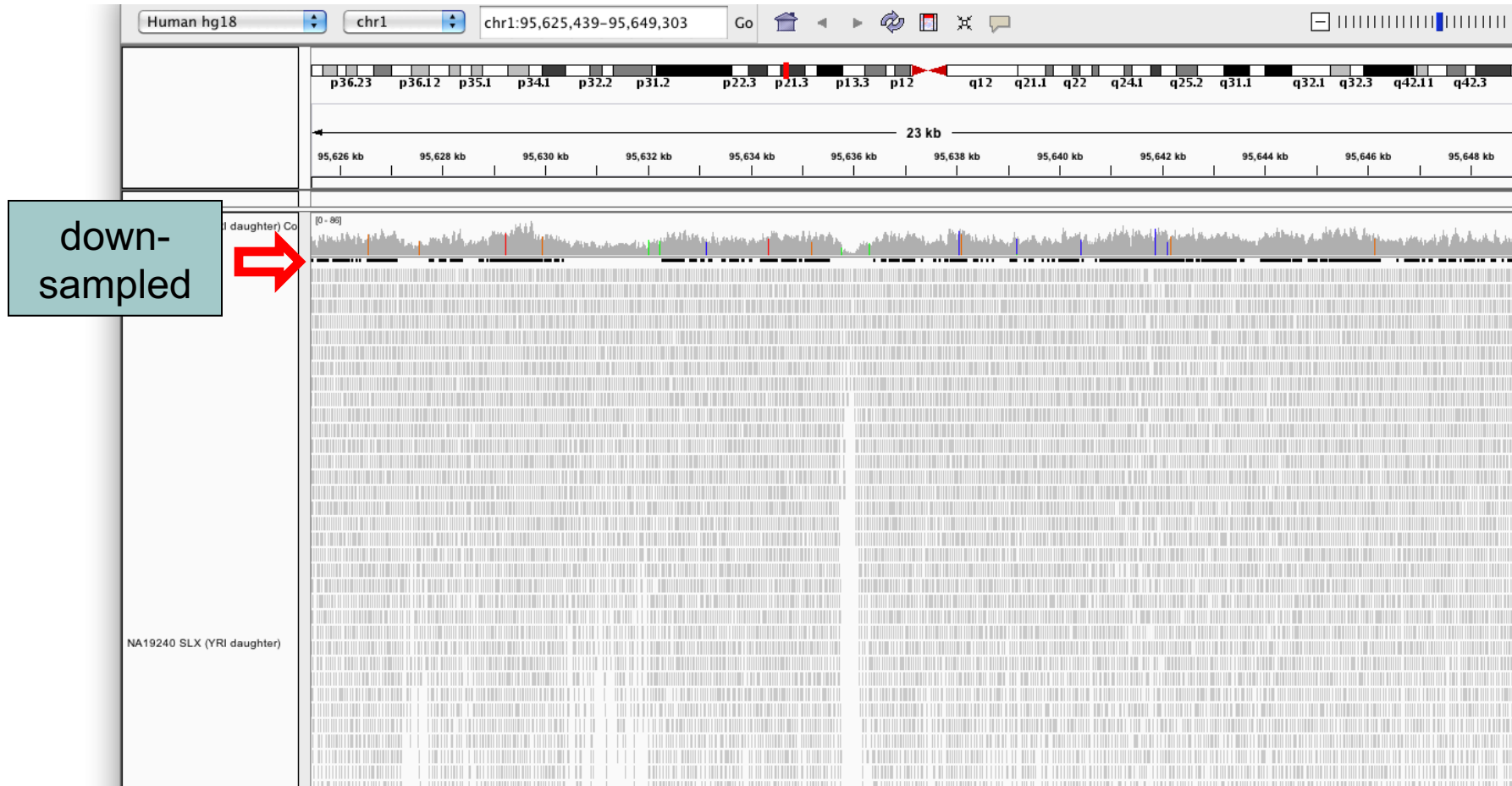
Downsampling

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

RefSeq genes

4 tracks loaded chr1:95,634,860 244M of 397M

Viewing alignments



Viewing SNPs

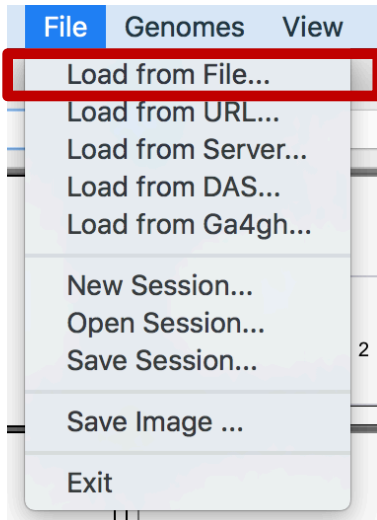
Hands-on exercise

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

Follow along

Viewing SNPs

Hands-on exercise



Click *File > Load from File*

Navigate to IGV folder that was pre-installed on the computer, and then to **Data / snps**

Open

NA12878.SLX.sample.bam

and

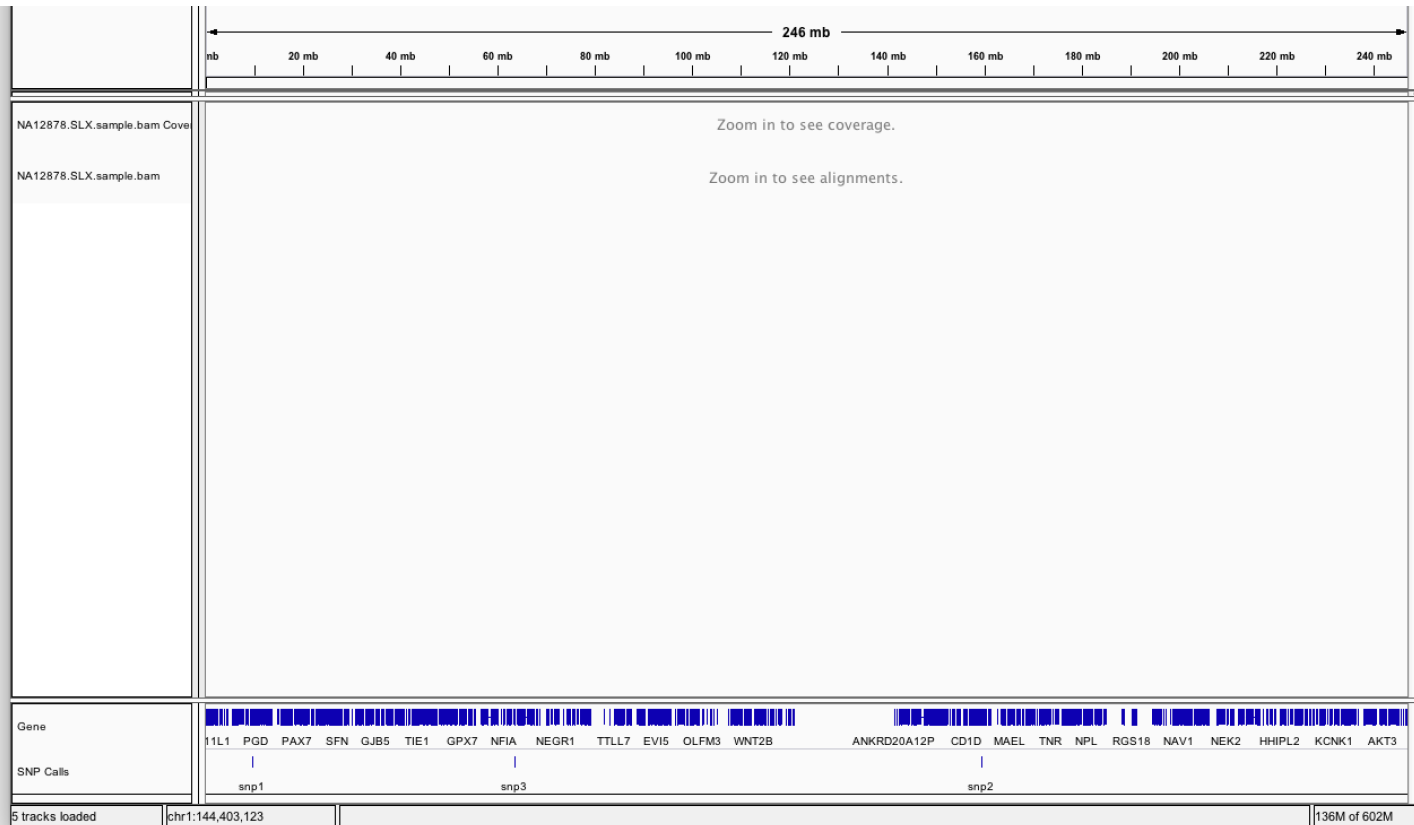
snp_calls.bed

not the .bai file

Note: This assumes you successfully loaded the genome in the previous exercise.

Viewing SNPs

Hands-on exercise

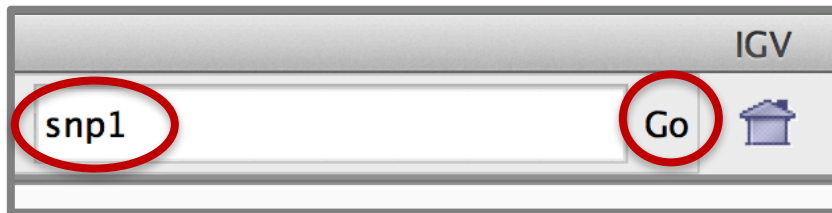


Viewing SNPs

Hands-on exercise

Navigate to first putative SNP locus

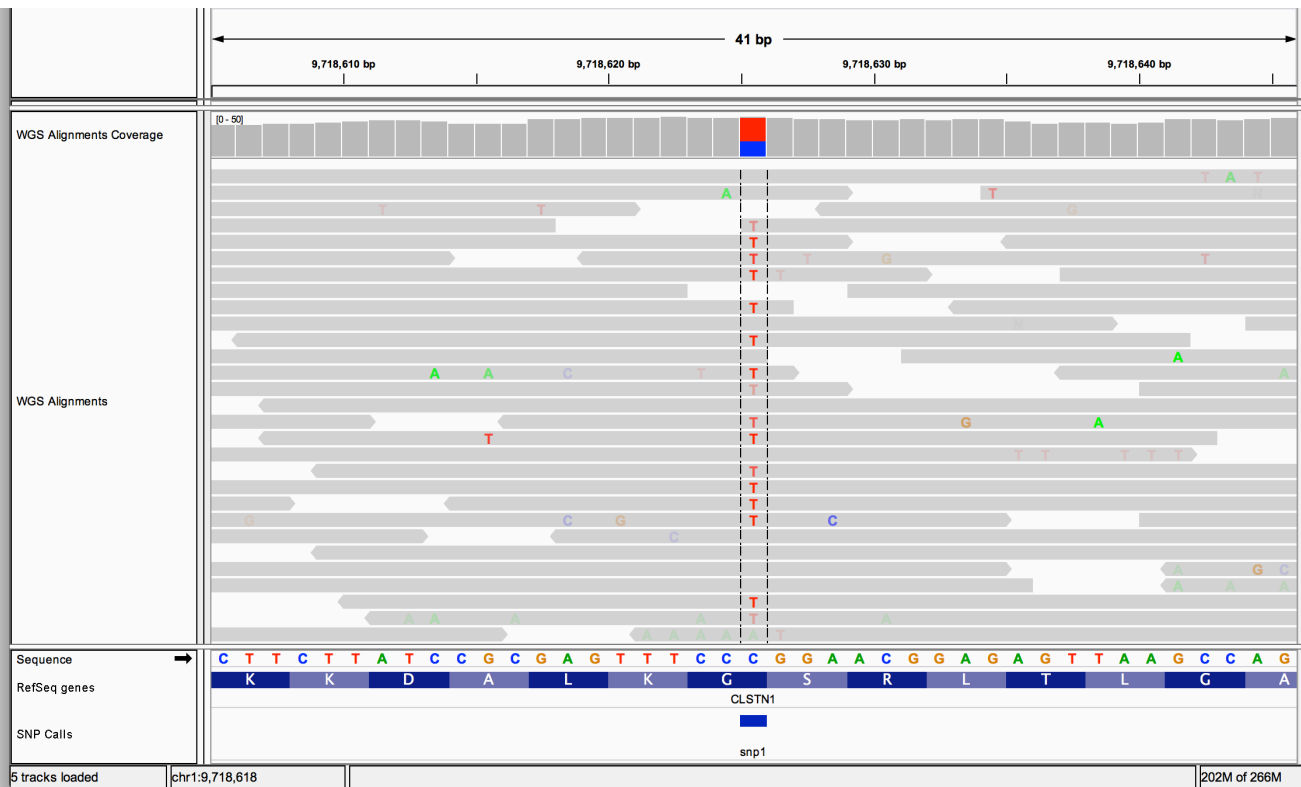
Type “snp1” in the **Search Box** and click **Go**



A screenshot of the IGV (Integrative Genomics Viewer) search interface. The window has a title bar with 'IGV' on the right. Below the title bar is a search bar containing the text 'snp1'. To the right of the search bar is a 'Go' button and a home icon. Both the search bar and the 'Go' button are circled in red.

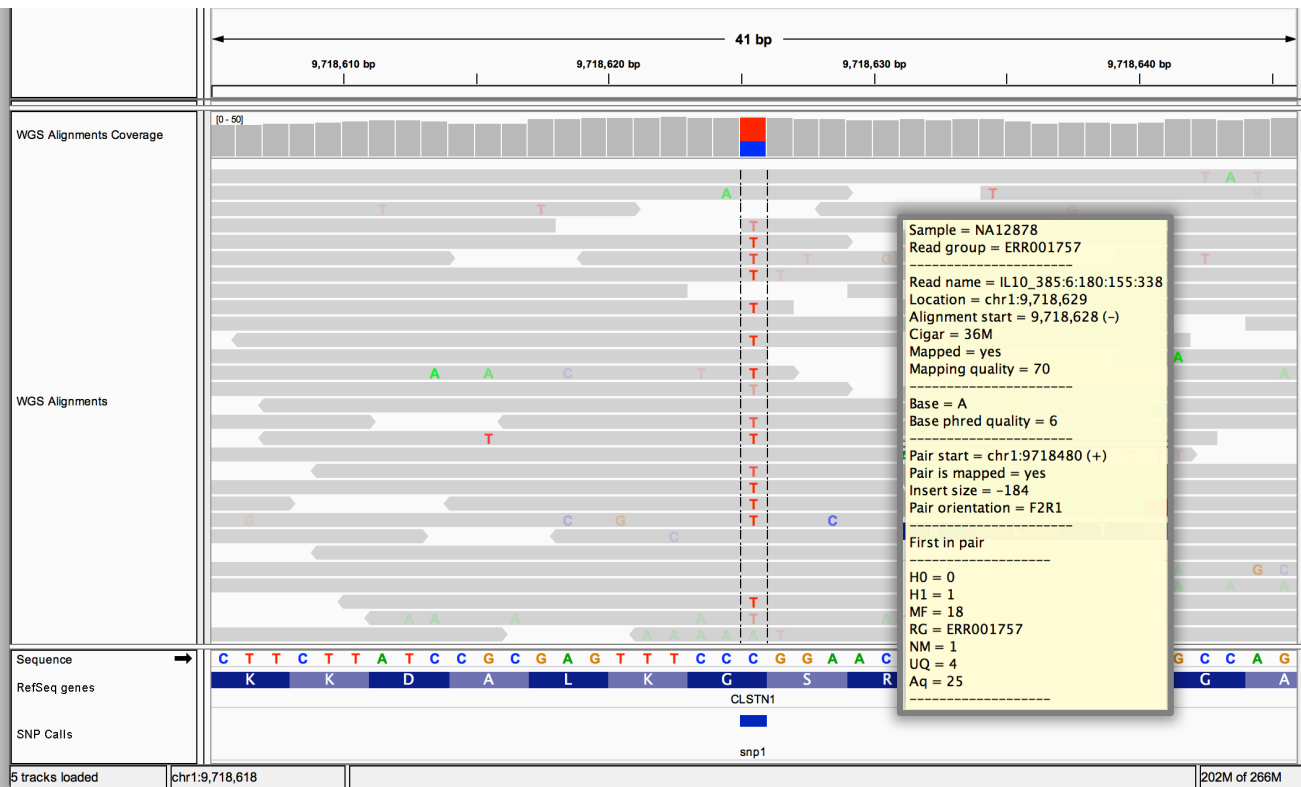
Viewing SNPs

Hands-on exercise



Viewing SNPs

Hands-on exercise

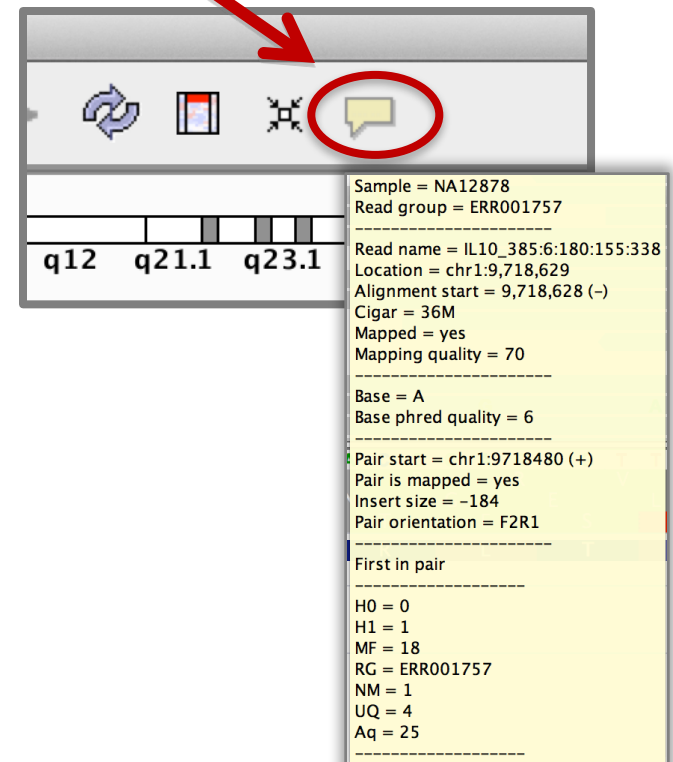
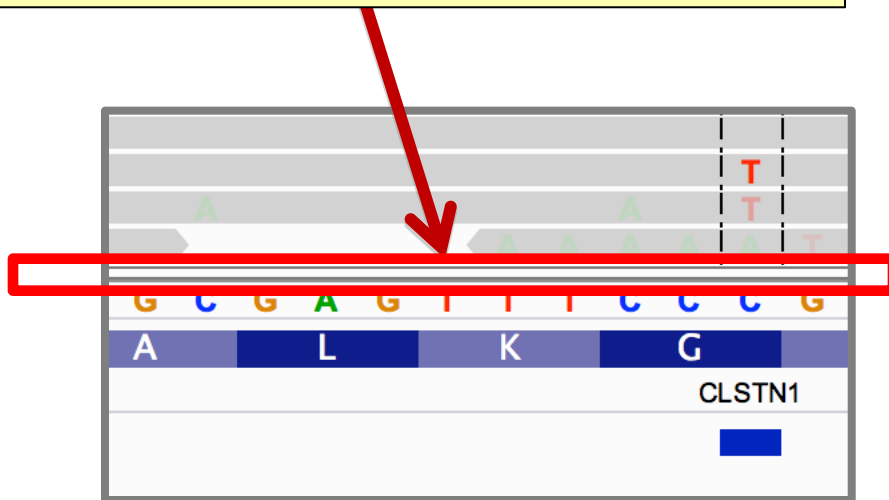


Viewing SNPs

Hands-on exercise

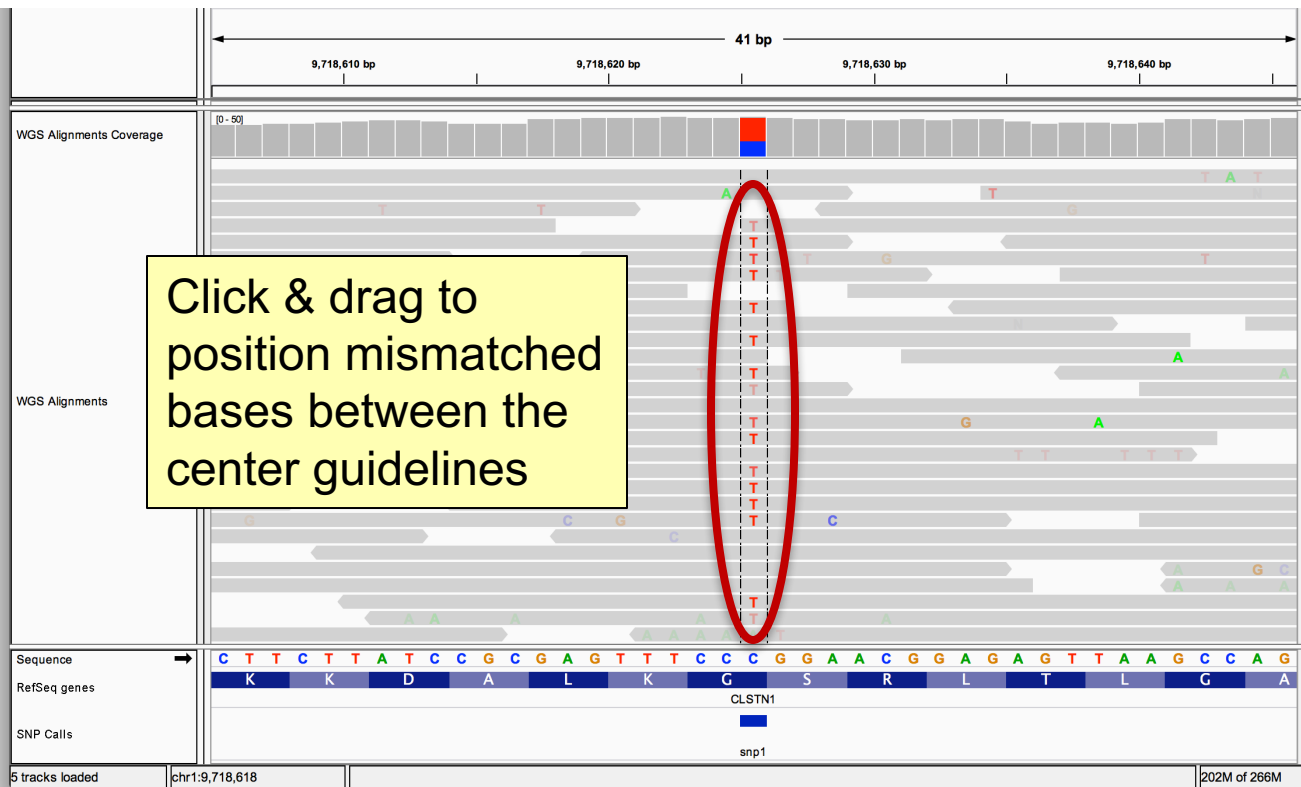
To modify the info popup behavior:
Click yellow balloon icon in toolbar

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



Viewing SNPs

Hands-on exercise



Viewing SNPs

Hands-on exercise

The screenshot displays a genomic browser interface with several tracks. The top track is 'WGS Alignments Coverage' with a scale of [0 - 50]. Below it is the 'WGS' track showing individual sequencing reads. A red arrow points to the 'WGS Alignments' context menu, which is open. The menu options are: 'Rename Track...', 'Copy read details to clipboard', 'Group alignments by', 'Sort alignments by', 'Color alignments by', 'Shade base by quality' (checked), 'Show mismatched bases' (checked), and 'Show all bases'. A second red arrow points to the 'base' option in the 'Sort alignments by' submenu. The background tracks include 'Sequence' (showing the reference sequence: C T T C T T A T C C G C G A G T T T C C G G A A C G G A G A G T T A A G C C A G), 'RefSeq genes' (showing the gene 'CLSTN1'), and 'SNP Calls' (showing a single SNP 'snp1'). The bottom status bar indicates '5 tracks loaded', 'chr1:9,718,618', and '202M of 266M'.

WGS Alignments

- Rename Track...
- Copy read details to clipboard
- Group alignments by
- Sort alignments by**
 - start location
 - read strand
 - first-of-pair strand
 - base**
 - mapping quality
 - sample
 - read group
 - insert size
 - chromosome of mate
 - tag
- Color alignments by
- ✓ Shade base by quality
- ✓ Show mismatched bases
- Show all bases

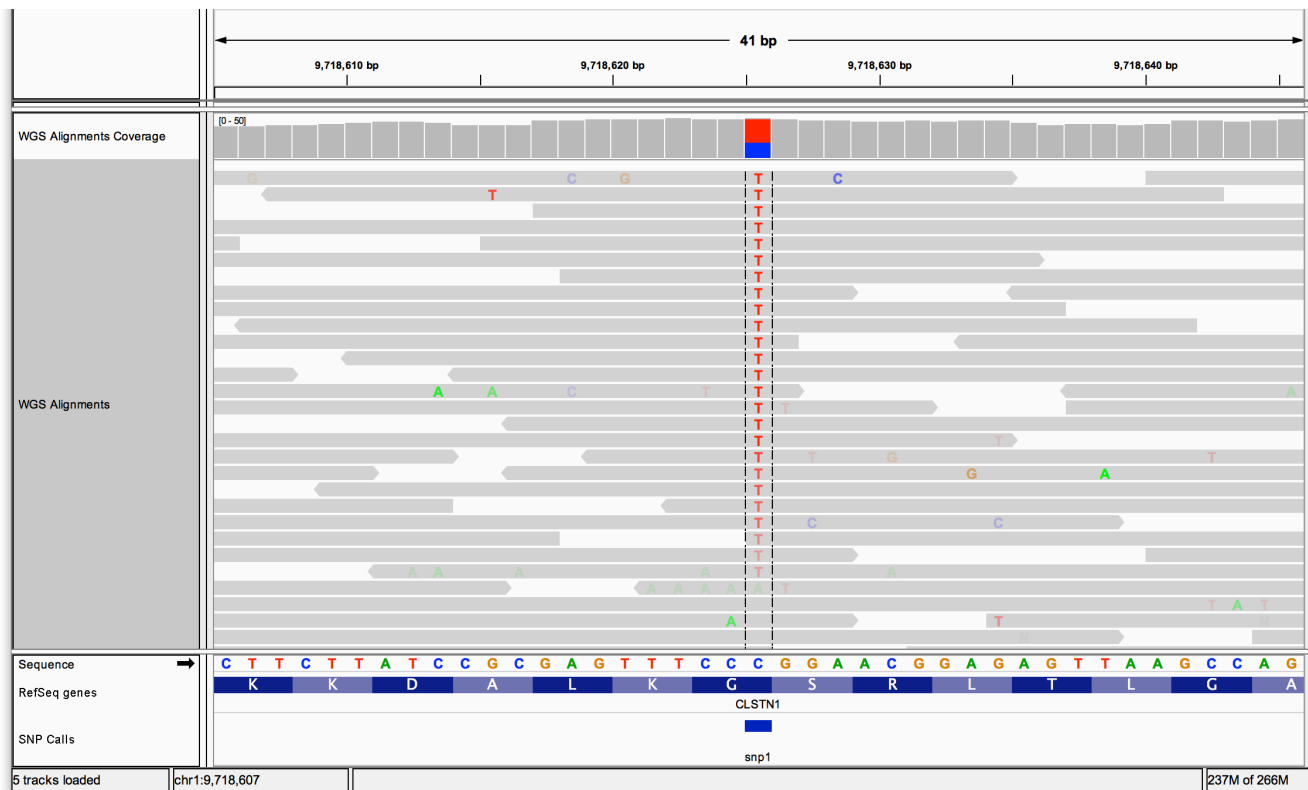
**Right-click on alignments and select
Sort alignments by > base**

**On Mac:
Right-click = Control-click**

Sequence → C T T C T T A T C C G C G A G T T T C C G G A A C G G A G A G T T A A G C C A G
RefSeq genes K K D A L K G S R L T L G A
CLSTN1
SNP Calls snp1
5 tracks loaded | chr1:9,718,618 | 202M of 266M

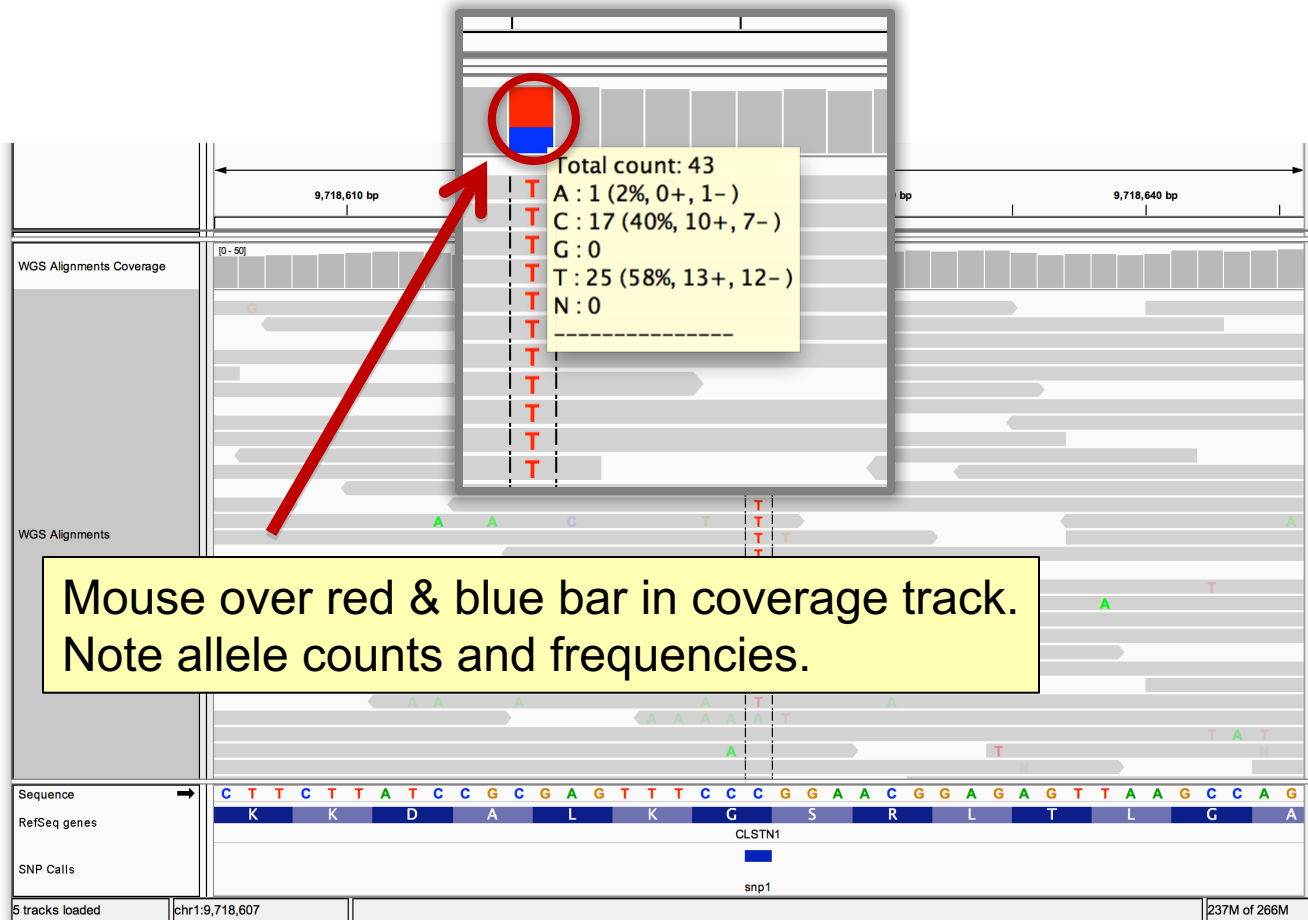
Viewing SNPs

Hands-on exercise



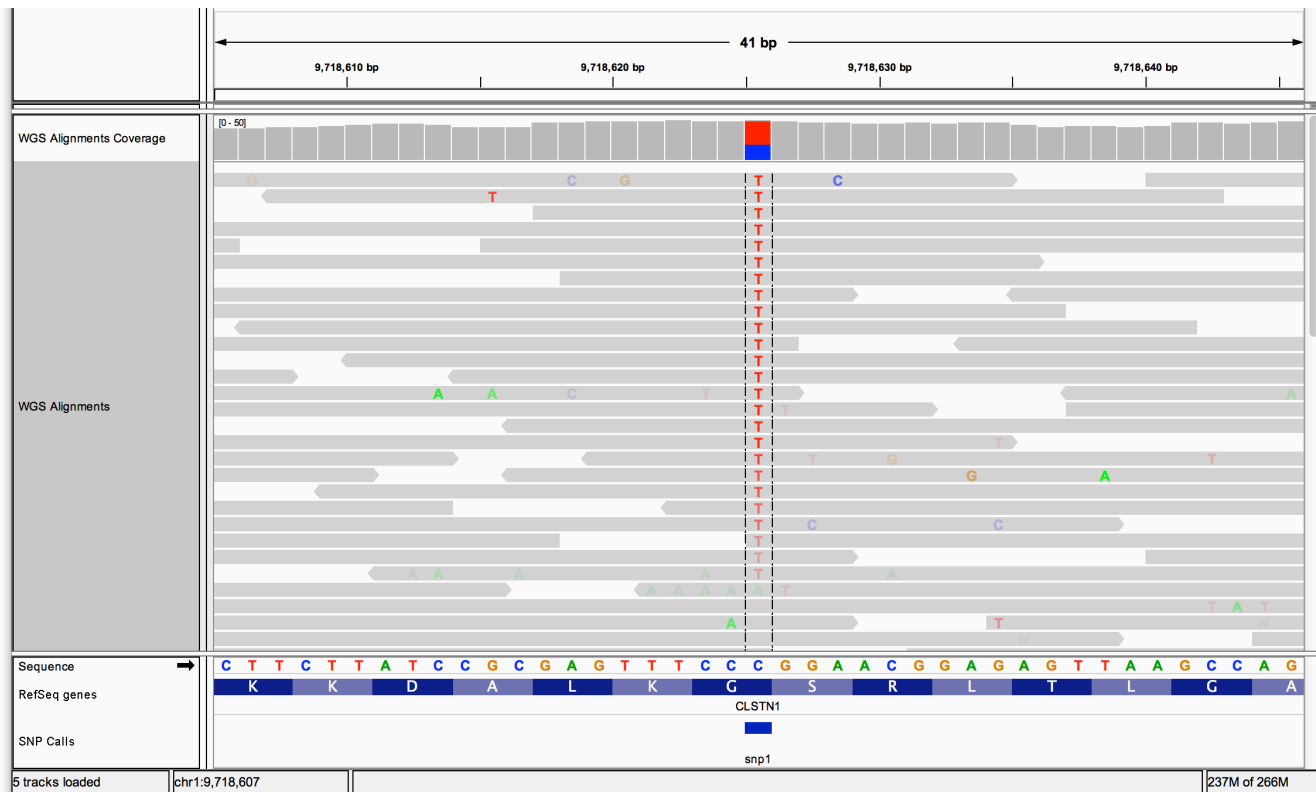
Viewing SNPs

Hands-on exercise



Viewing SNPs

Hands-on exercise

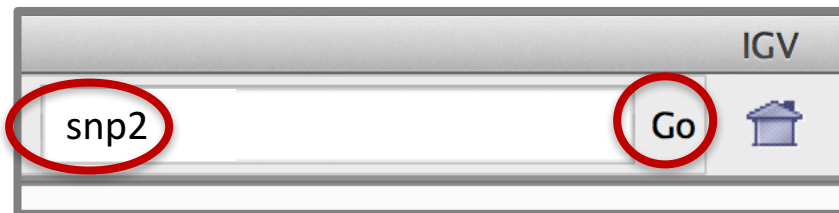


Viewing SNPs

Hands-on exercise

Navigate to next putative SNP locus

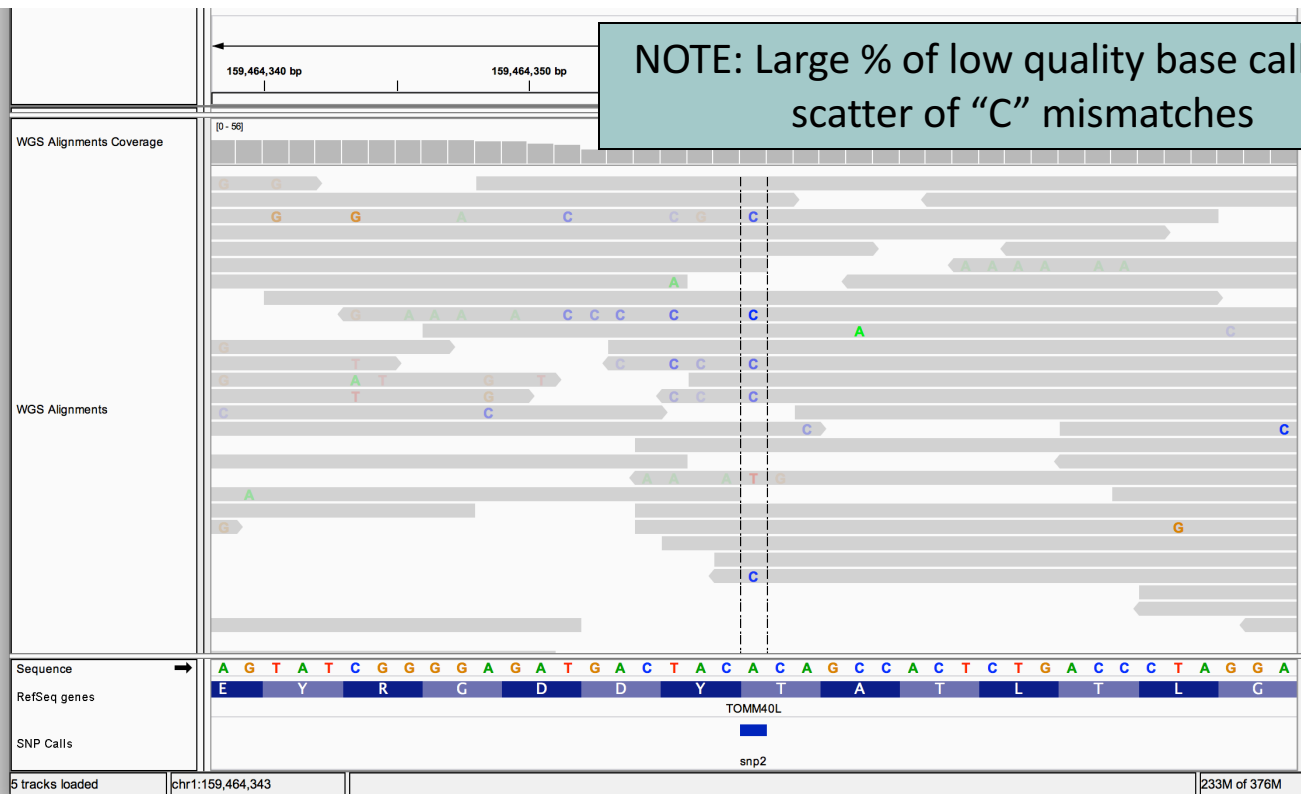
Type “snp2” in the **Search Box** and click **Go**



The screenshot shows the IGV (Integrative Genomics Viewer) search interface. It features a search bar with the text "snp2" entered, a "Go" button, and a home icon. The text "IGV" is visible in the top right corner of the interface.

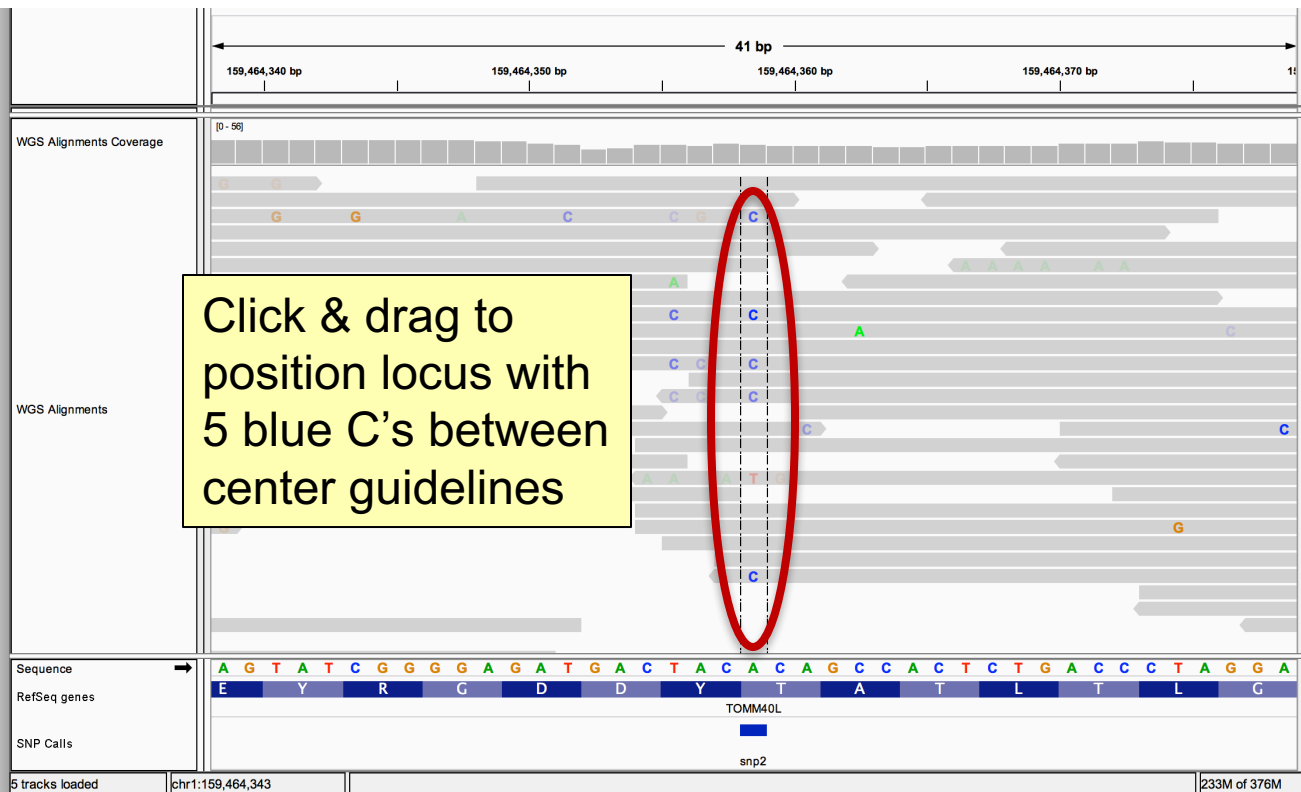
Viewing SNPs

Hands-on exercise



Viewing SNPs

Hands-on exercise



Viewing SNPs

Hands-on exercise

The screenshot displays a genomic browser interface with several tracks. The top track shows a genomic region from 159,464,340 bp to 159,464,360 bp, with a 41 bp scale bar. Below this is the 'WGS Alignments Coverage' track, followed by the 'WGS Alignments' track. A red arrow points to a right-click context menu that is open over the alignments. The menu options are:

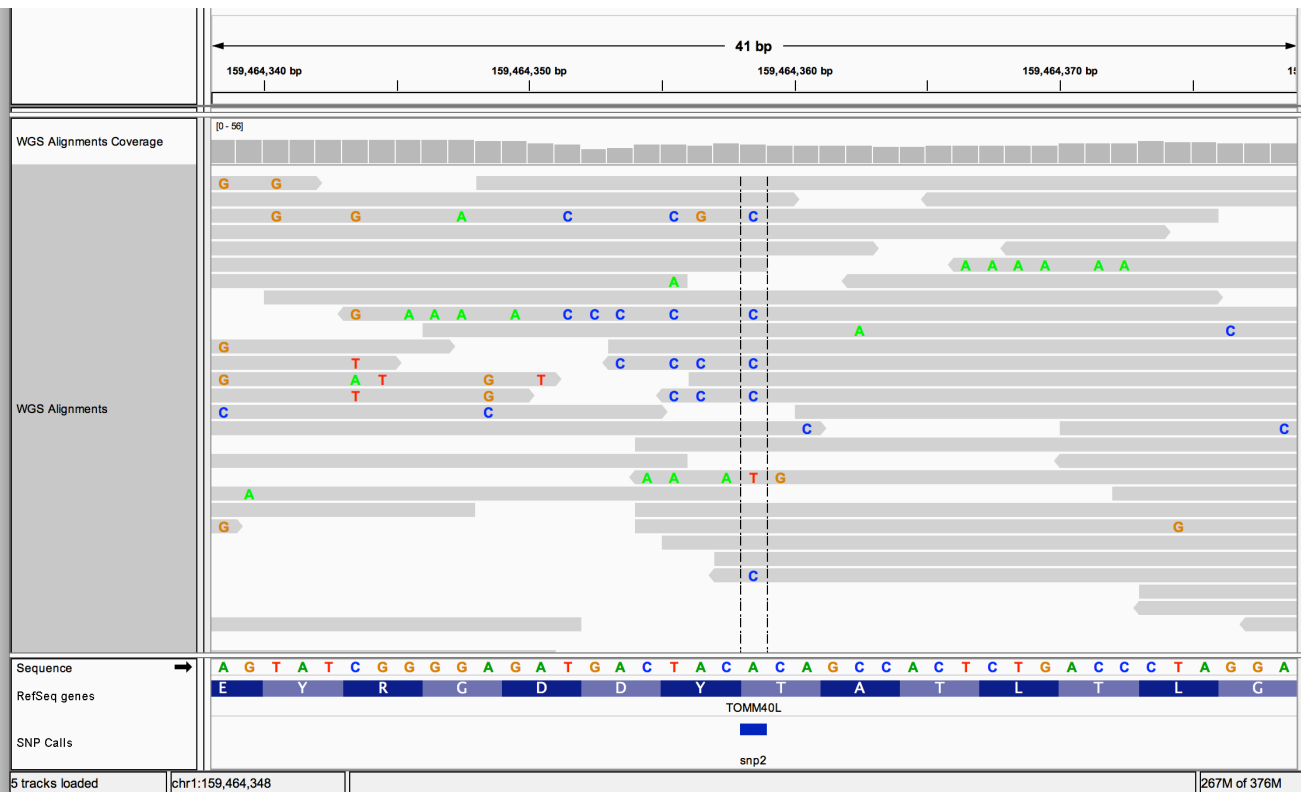
- WGS Alignments
- Rename Track...
- Copy read details to clipboard
- Group alignments by ▶
- Sort alignments by ▶
- Color alignments by ▶
- ☒ Shade base by quality
- ☒ Show mismatched bases
- Show all bases
- View as pairs
- Go to mate
- View mate region in split screen
- Set insert size options ...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

A yellow callout box with black text is overlaid on the bottom left of the alignment track, stating: "Right-click on alignments and deselect **Shade base by quality**".

At the bottom of the browser, the 'Sequence' track shows the reference sequence: A G T A T C G G G A G A T G A C T A C A C A G G C C A C T C T G A C C C T A G G A. Below this, the 'RefSeq genes' track shows the gene 'TOMM40L'. The 'SNP Calls' track shows a single SNP call 'snp2'. The bottom status bar indicates '5 tracks loaded', 'chr1:159,464,343', and '233M of 376M'.

Viewing SNPs

Hands-on exercise



Viewing SNPs

Hands-on exercise

The screenshot displays a genomic browser interface with several tracks. The 'WGS Alignments Coverage' track shows a bar chart of coverage. The 'WGS Alignments' track shows individual sequencing reads. A right-click context menu is open over the alignments, with the 'Sort alignments by' option selected. A secondary menu is visible, showing 'read strand' as the chosen sorting criterion. Red arrows indicate the sequence of clicks. A yellow callout box provides the instruction: 'Right-click on alignments and select Sort alignments by > read strand'. The bottom of the interface shows the reference sequence, RefSeq genes (TOMM40L), and SNP calls (snp2).

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

Sort alignments by

- 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 > read strand

Sequence
RefSeq genes
SNP Calls

5 tracks loaded | chr1:159,464,343 | TOMM40L | snp2 | 233M of 376M

Viewing SNPs

Hands-on exercise

The screenshot displays a genomic browser interface with several tracks. The 'WGS Alignments Coverage' track is highlighted, and a right-click context menu is open over it. The menu options are:

- 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 ...

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

- no color
- insert size
- pair orientation
- ✓ insert size and pair orientation
- read strand**
- first-of-pair strand
- read group
- sample
- tag
- bisulfite mode ▶

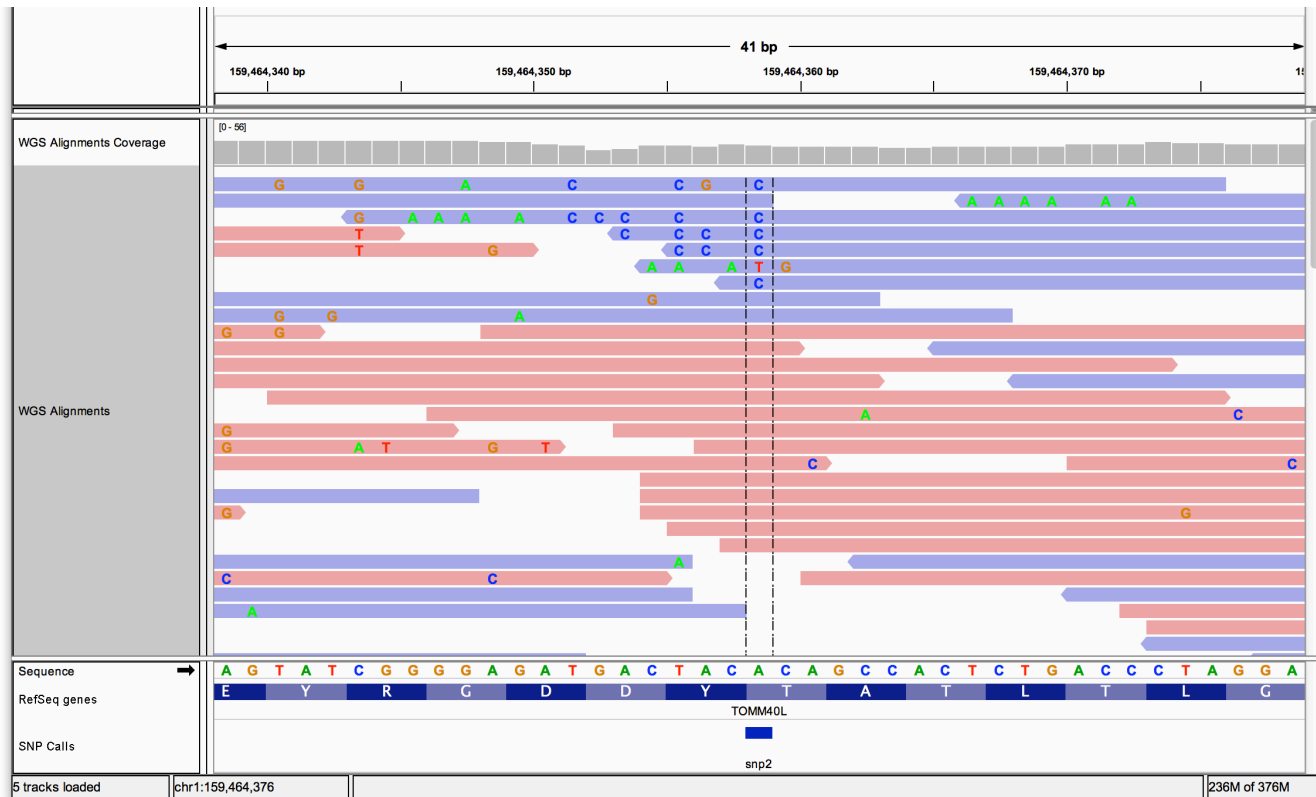
Red arrows indicate the sequence of clicks: from the 'WGS Alignments Coverage' track to 'Color alignments by', and then to 'read strand'.

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

The background interface includes tracks for 'WGS Alignments Coverage', 'WGS Alignments', 'Sequence', 'RefSeq genes', and 'SNP Calls'. The 'Sequence' track shows 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. The 'RefSeq genes' track shows the gene: TOMM40L. The 'SNP Calls' track shows a single SNP: snp2. The bottom status bar indicates '5 tracks loaded', 'chr1:159,464,343', and '233M of 376M'.

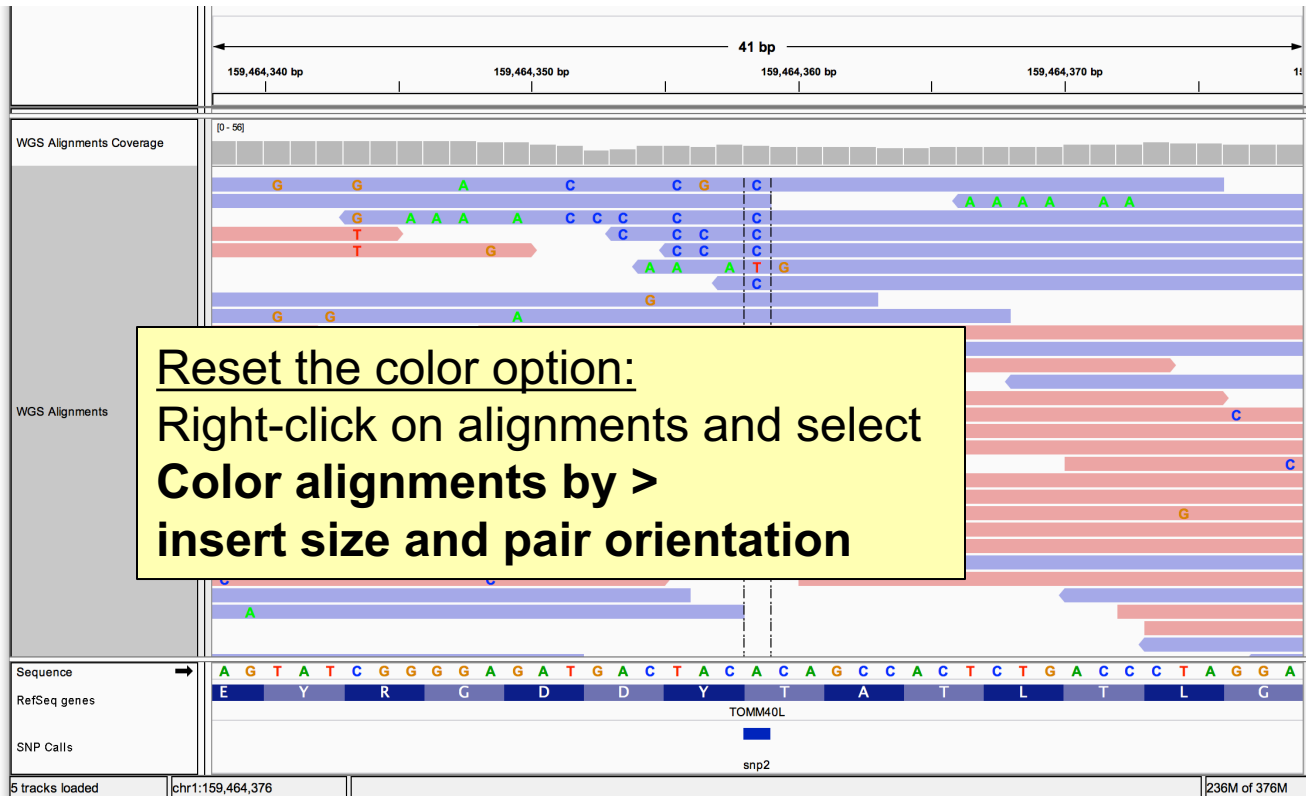
Viewing SNPs

Hands-on exercise



Viewing SNPs

Hands-on exercise



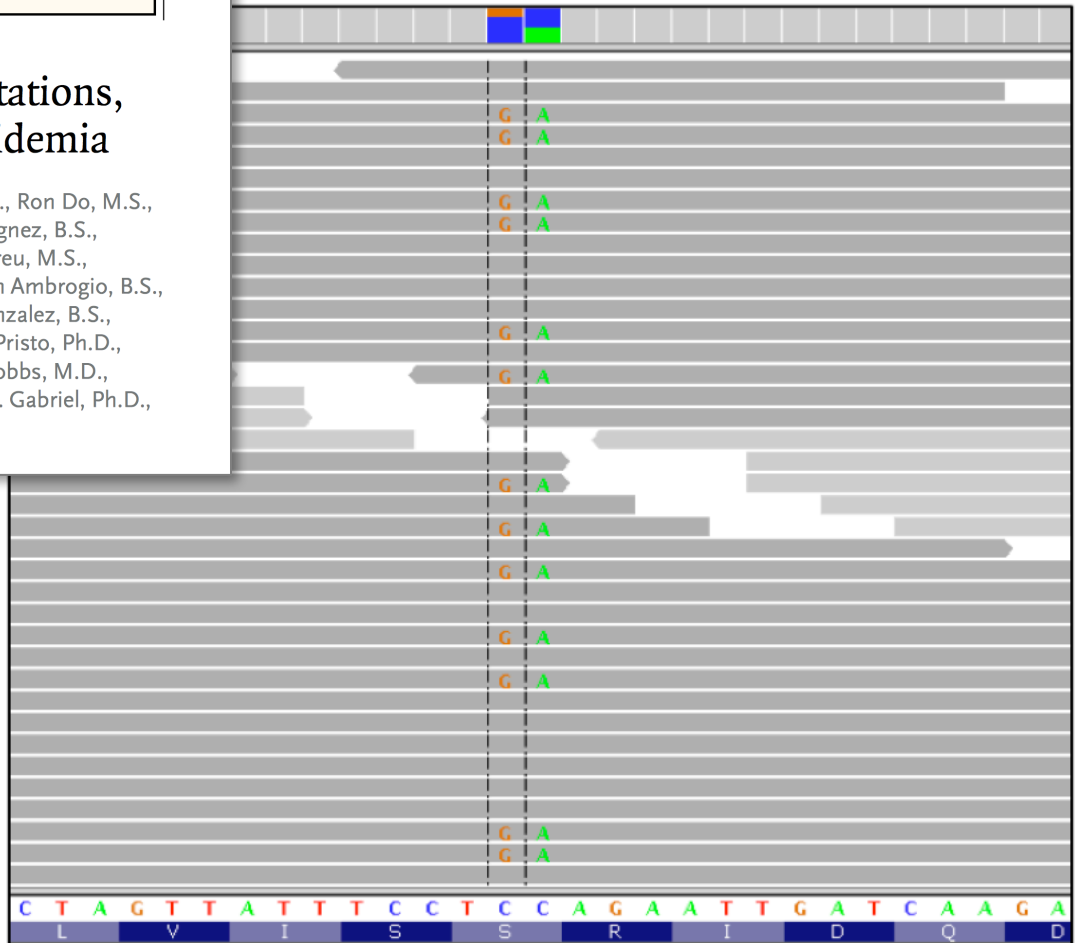
Viewing SNPs

The NEW ENGLAND JOURNAL of MEDICINE

BRIEF REPORT

Exome Sequencing, *ANGPTL3* Mutations, and Familial Combined Hypolipidemia

Kiran Musunuru, M.D., Ph.D., M.P.H., James P. Pirruccello, B.S., Ron Do, M.S.,
Gina M. Peloso, M.S., Candace Guiducci, B.S., Carrie Sougnéz, B.S.,
Kiran V. Garimella, M.S., Sheila Fisher, M.L.A., Justin Abreu, M.S.,
Andrew J. Barry, B.S., Tim Fennell, B.S., Eric Banks, Ph.D., Lauren Ambrogio, B.S.,
Kristian Cibulskis, B.S., Andrew Kernysky, Ph.D., Elena Gonzalez, B.S.,
Nicholas Rudzicz, M.S., James C. Engert, Ph.D., Mark A. DePristo, Ph.D.,
Mark J. Daly, Ph.D., Jonathan C. Cohen, Ph.D., Helen H. Hobbs, M.D.,
David Altshuler, M.D., Ph.D., Gustav Schonfeld, M.D., Stacey B. Gabriel, Ph.D.,
Pin Yue, Ph.D., and Sekar Kathiresan, M.D.



Viewing SNPs



BED file used to define “bookmarks” in SNP exercise

snp_calls.bed

track name="SNP Calls"			
1	9718624	9718625	snp1
1	159464357	159464358	snp2
1	63650225	63650226	snp3

igvtools

igvtools

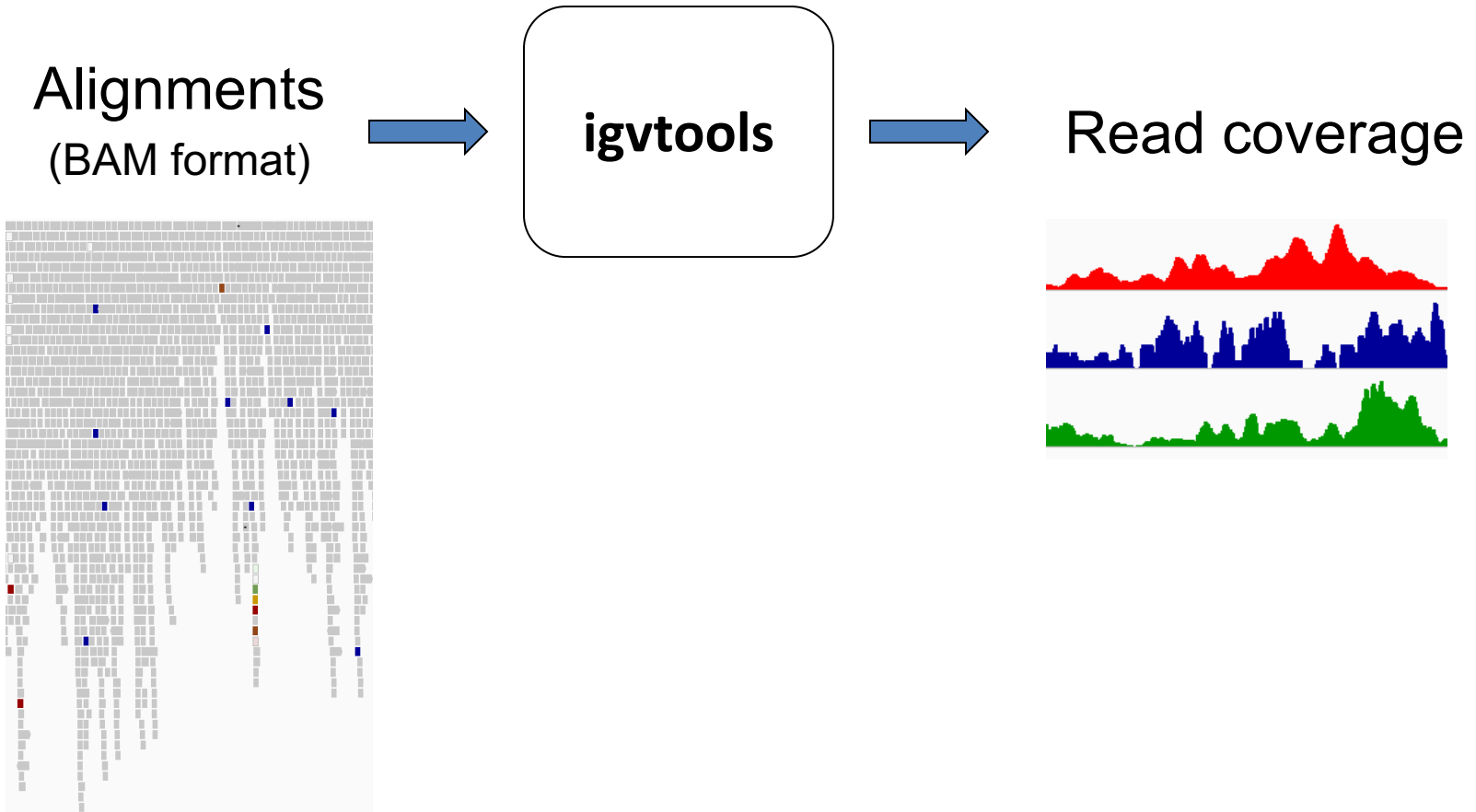
A set of utilities for preparing large files for efficient display.

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, SAM, BAM, VCF
index	– Creates index for large genomic annotation files and alignments. – Supported file formats: BED, GFF, GTF, PSL, SAM, BAM, VCF

igvtools: count

count

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



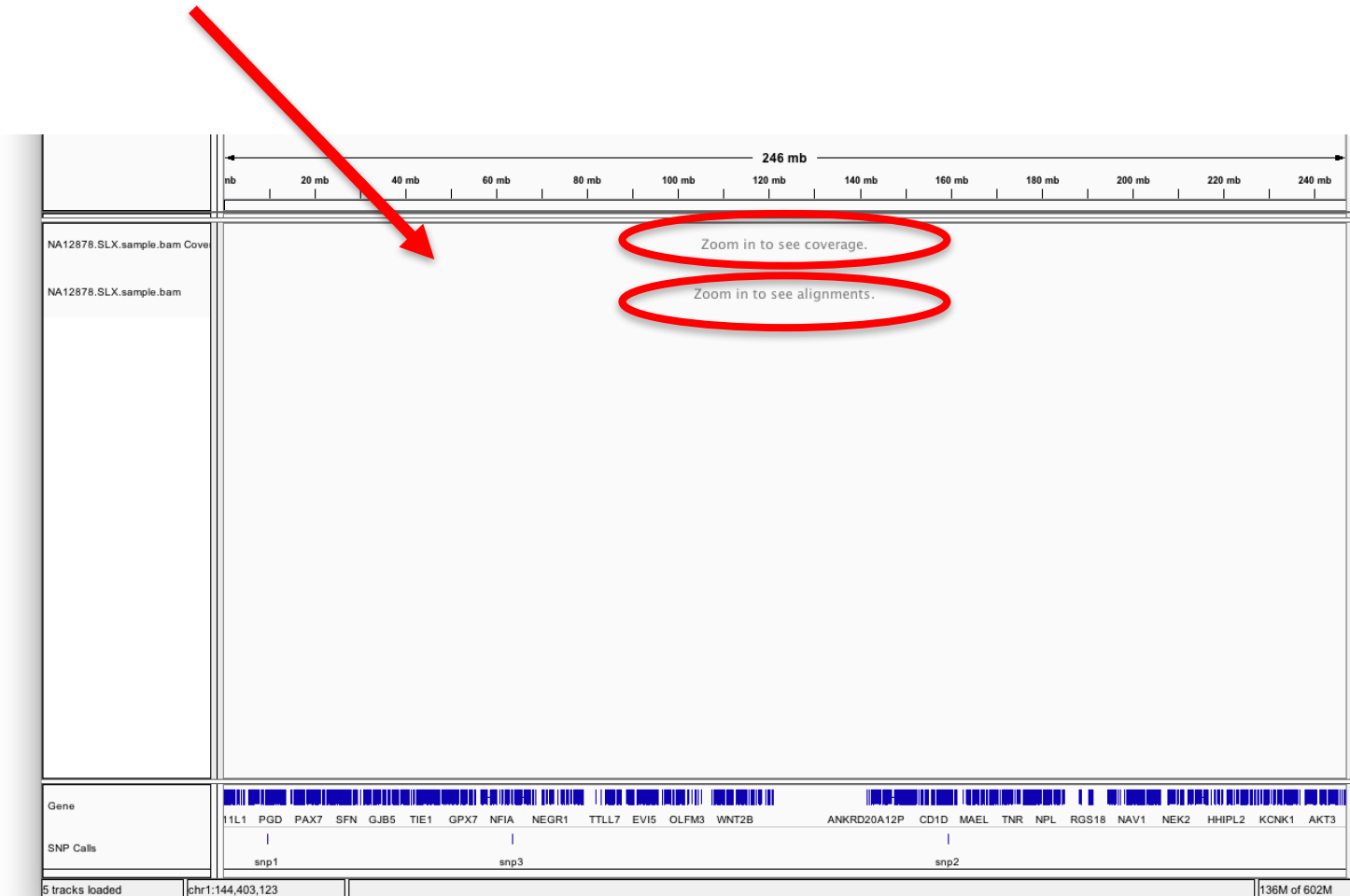
Coverage track

Coverage track visible before alignments



Coverage track

In our hands-on exercise:
Coverage track was **not** visible before alignments



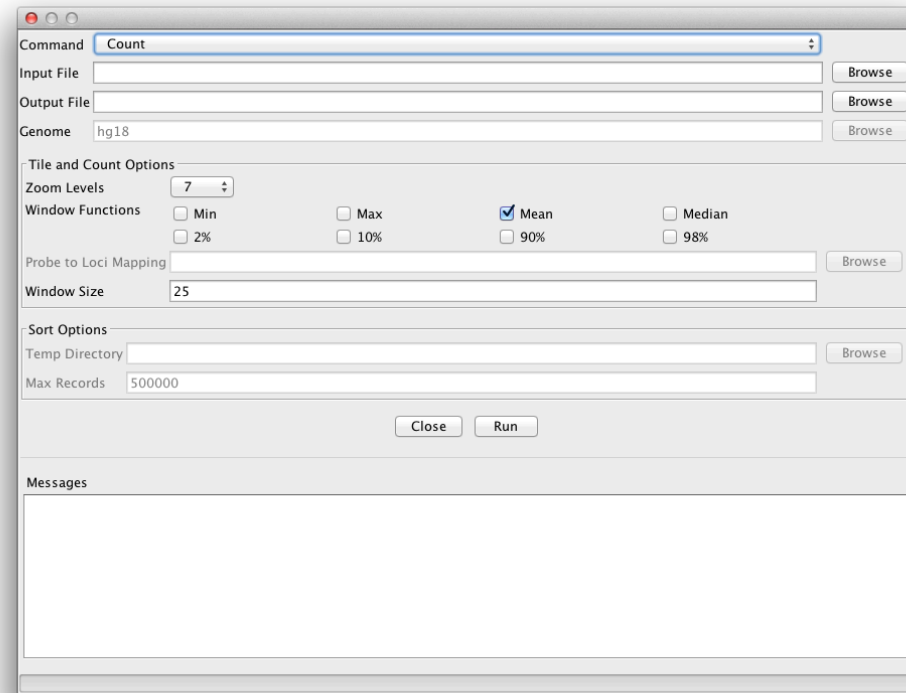
igvtools

(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
```



Pre-compute coverage track

Hands-on exercise

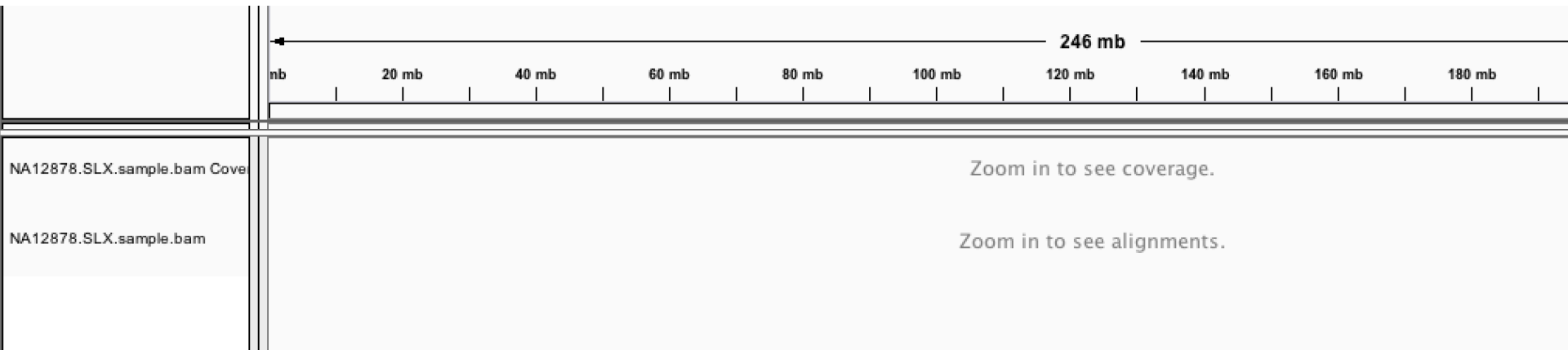
- Use ***igvtools*** to compute the coverage for a .bam file
- Associate the new file with the coverage track

Follow along

Coverage track

Hands-on exercise

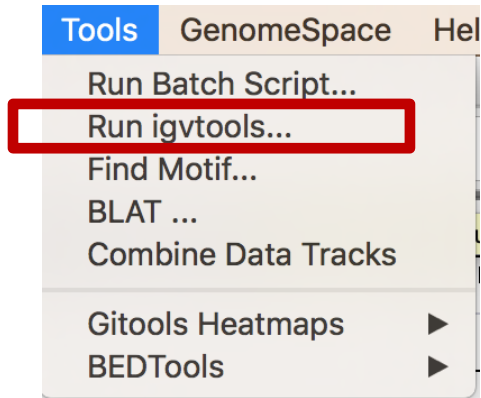
Click on the left-most tick on the zoom ruler to zoom all the way out



Pre-compute coverage track

Hands-on exercise

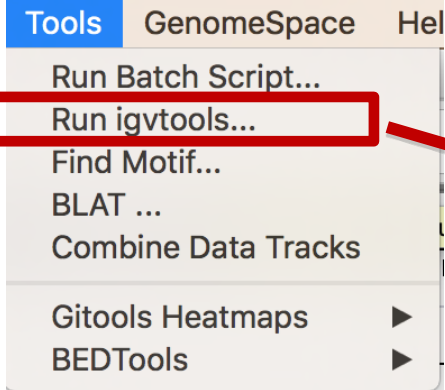
Click **Tools** > **Run igvtools**



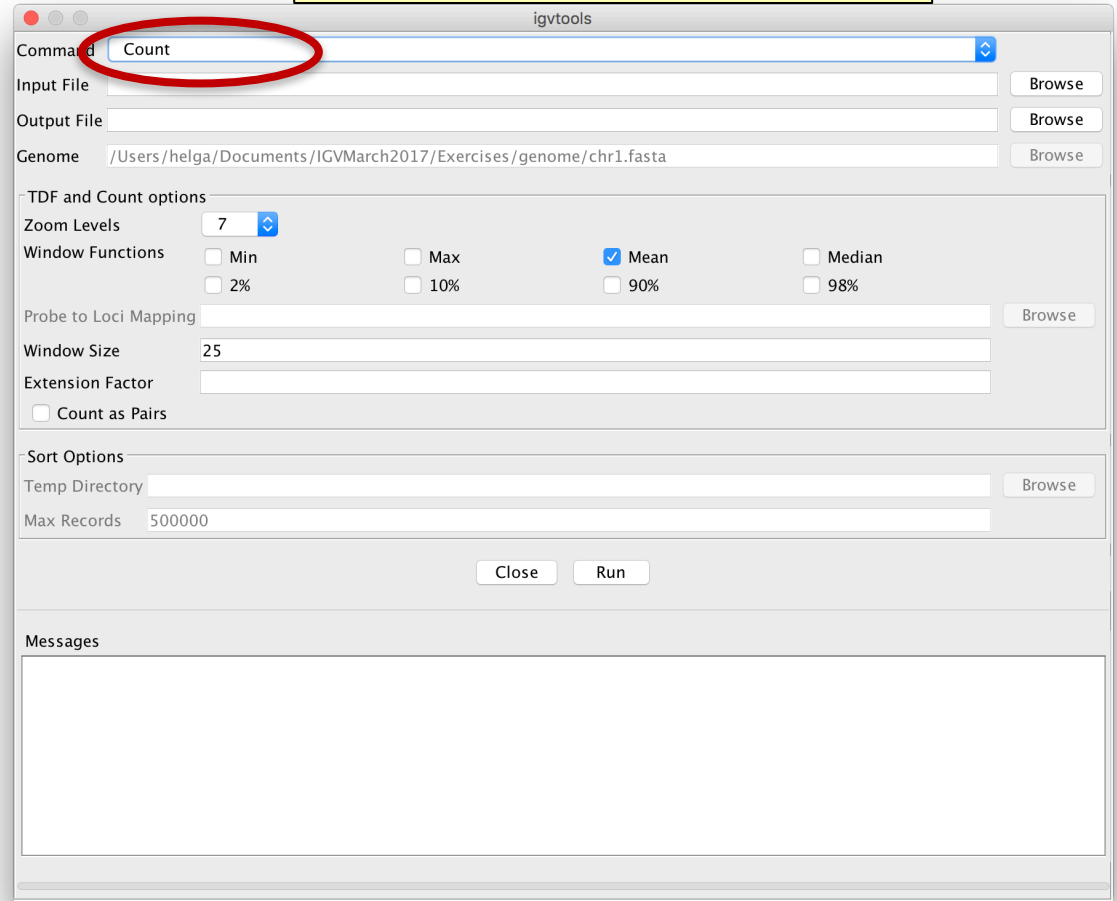
Pre-compute coverage track

Hands-on exercise

Click **Tools** > **Run igvtools**



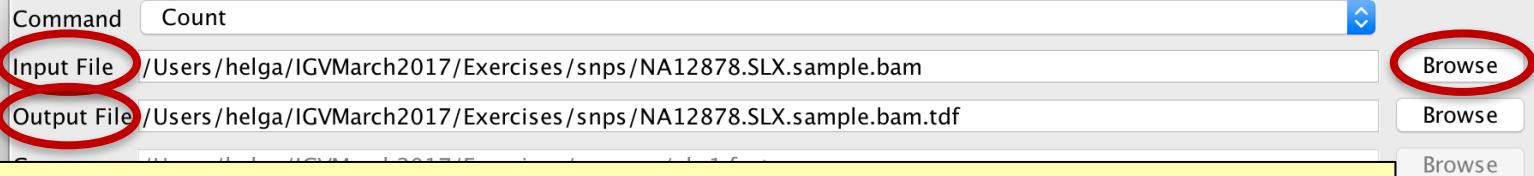
Select the **Count** command from the dropdown menu



Pre-compute coverage track

Hands-on exercise

1. Input file: Browse to the *Data / snps_morning* folder and select the **.bam** file

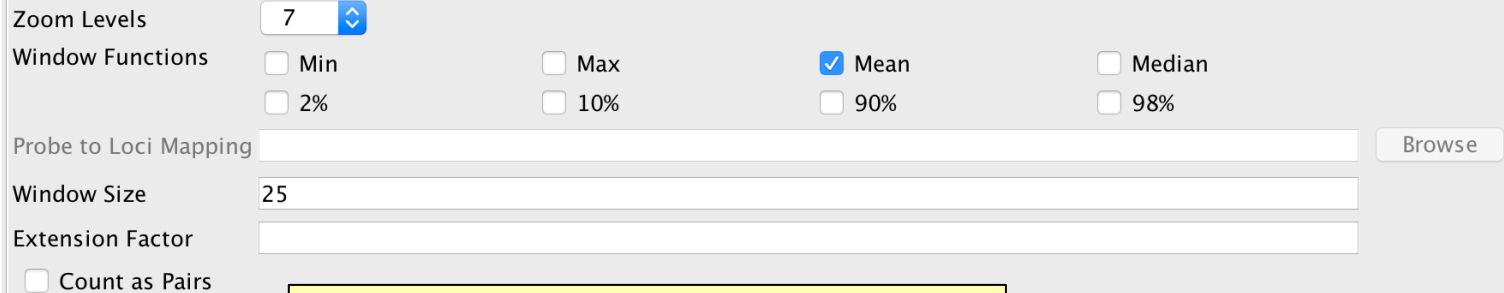


Command Count

Input File

Output File

2. Output file: Defaults to same folder, and same name + .tdf suffix



Zoom Levels

Window Functions ☐ Min ☐ Max ☒ Mean ☐ Median

☐ 2% ☐ 10% ☐ 90% ☐ 98%

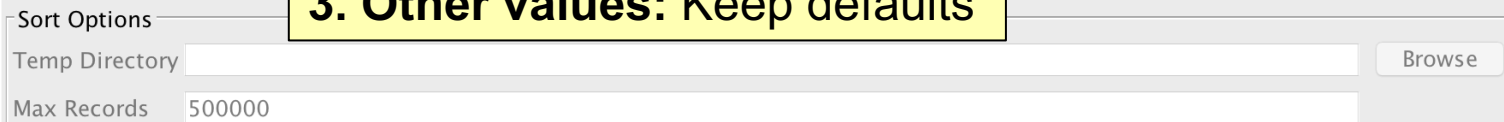
Probe to Loci Mapping

Window Size

Extension Factor

☐ Count as Pairs

3. Other values: Keep defaults



Sort Options

Temp Directory

Max Records

4. Click *Run*

Messages

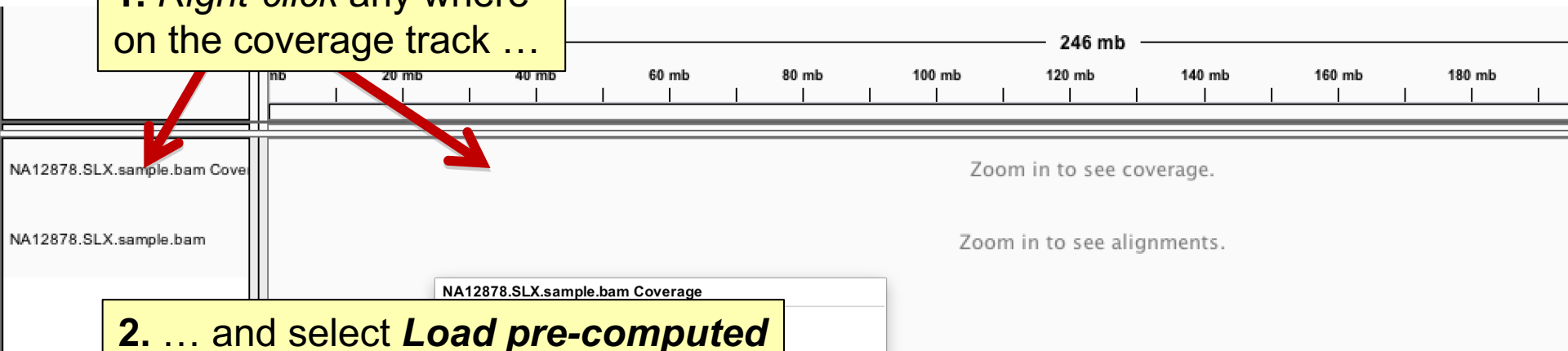
100.0%
Done

5. Close window after you see “Done” in the **Messages area**

Coverage track

Hands-on exercise

1. Right-click anywhere on the coverage track ...



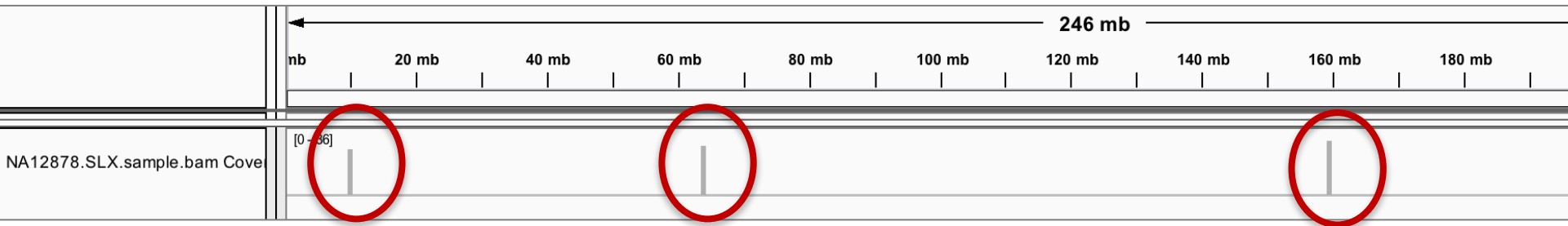
2. ... and select **Load pre-computed coverage data** from the popup menu

- Set Data Range...
- Log scale
- ☒ Autoscale
- ☒ Show Data Range
- Set allele frequency threshold...
- Load pre-computed coverage data...**
- Copy Details to Clipboard
- ☒ Show Alignment Track
- Show Splice Junction Track
- Hide Track
- Save image...
- Export track names...

3. Browse to the **Data / snps_morning** folder and select the file **NA12878.SLX.sample.bam.tdf**

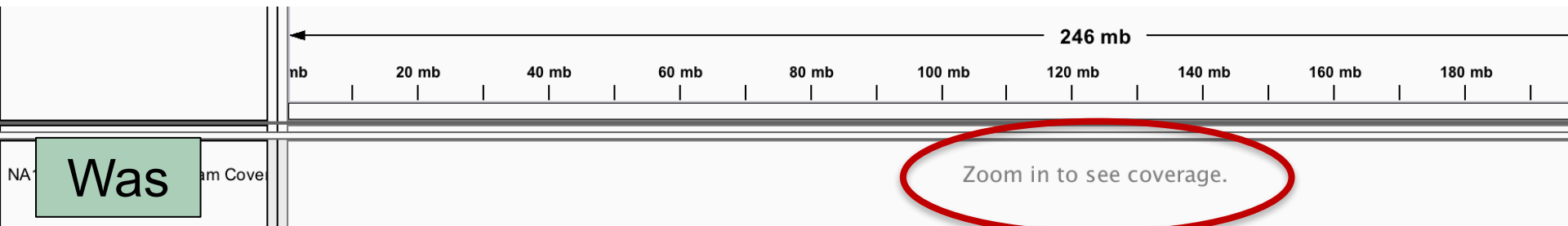
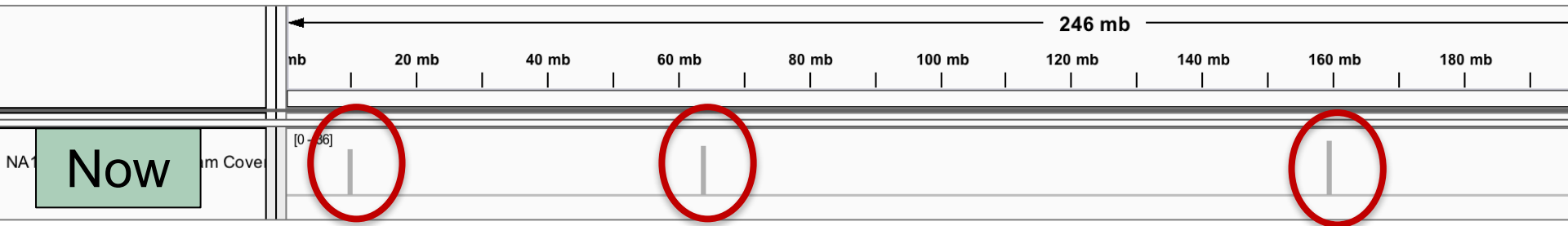
Coverage track

Hands-on exercise



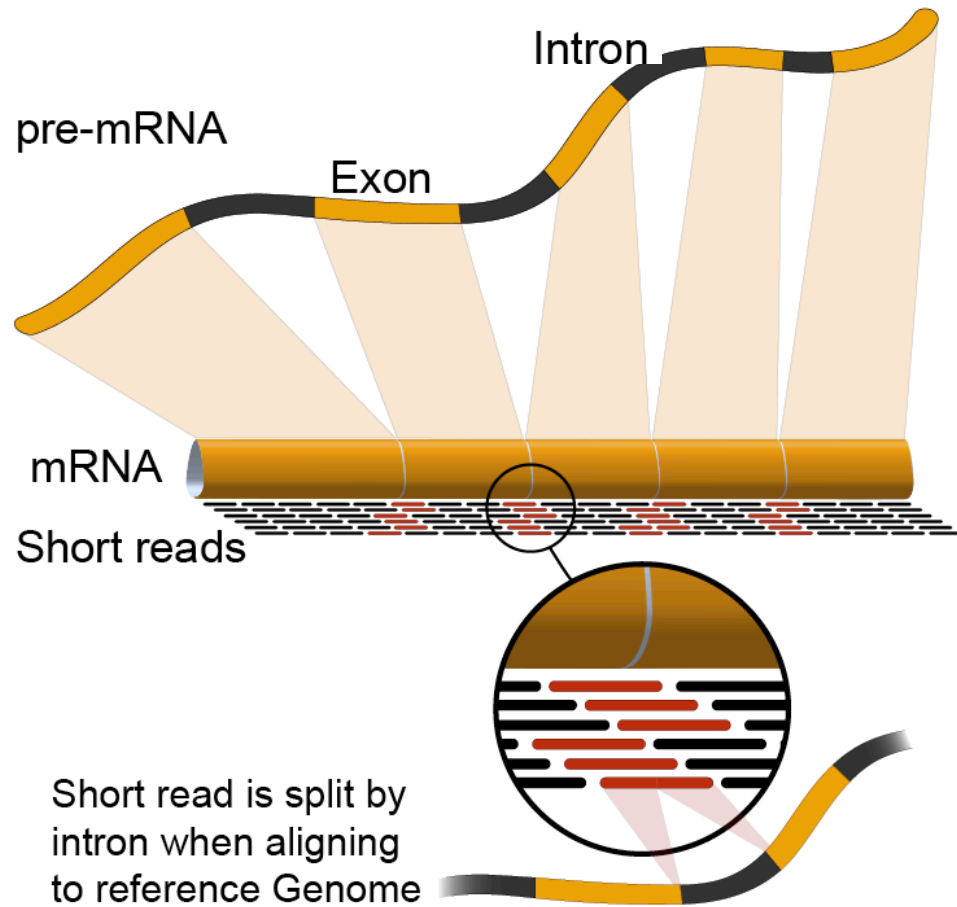
Coverage track

Hands-on exercise

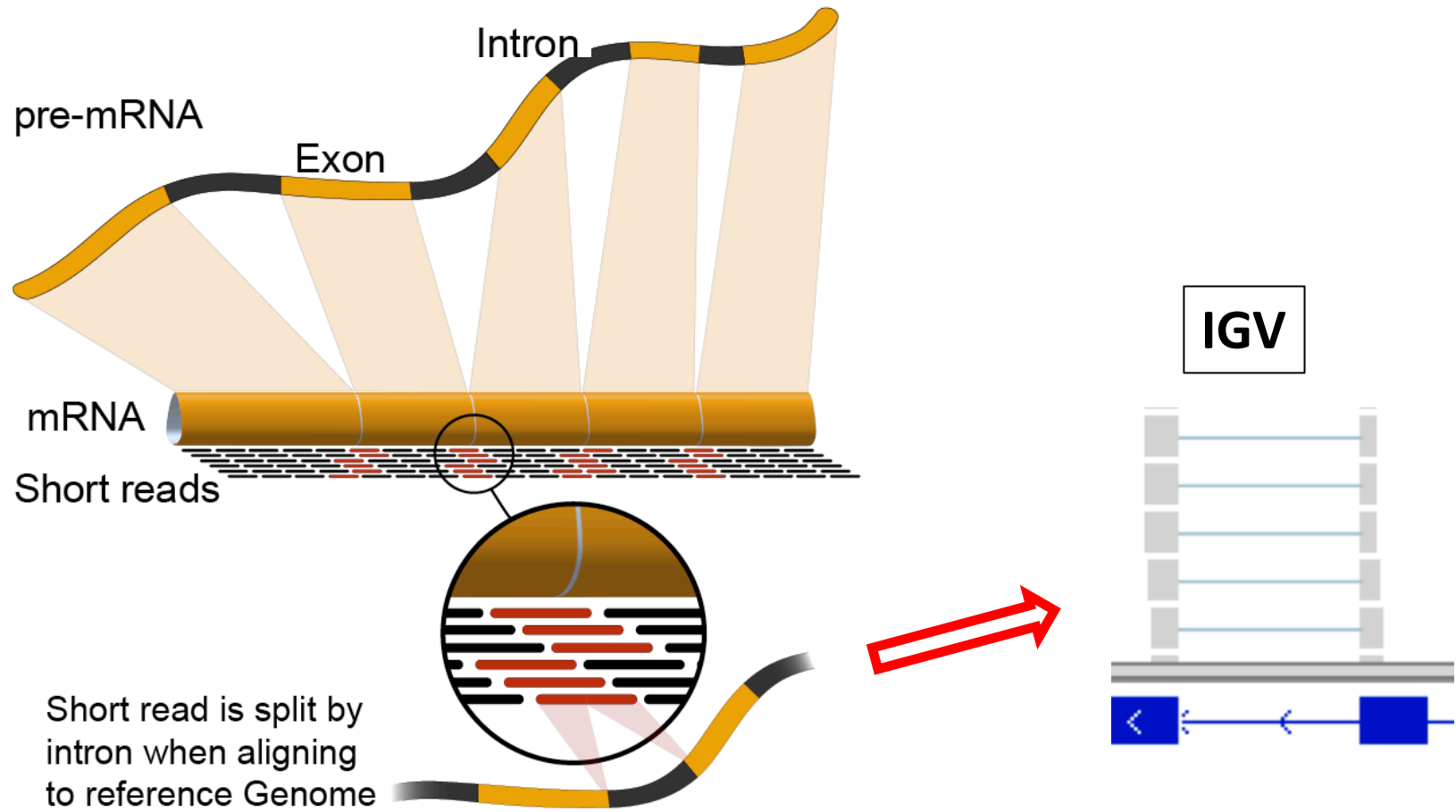


Viewing RNA-seq Data

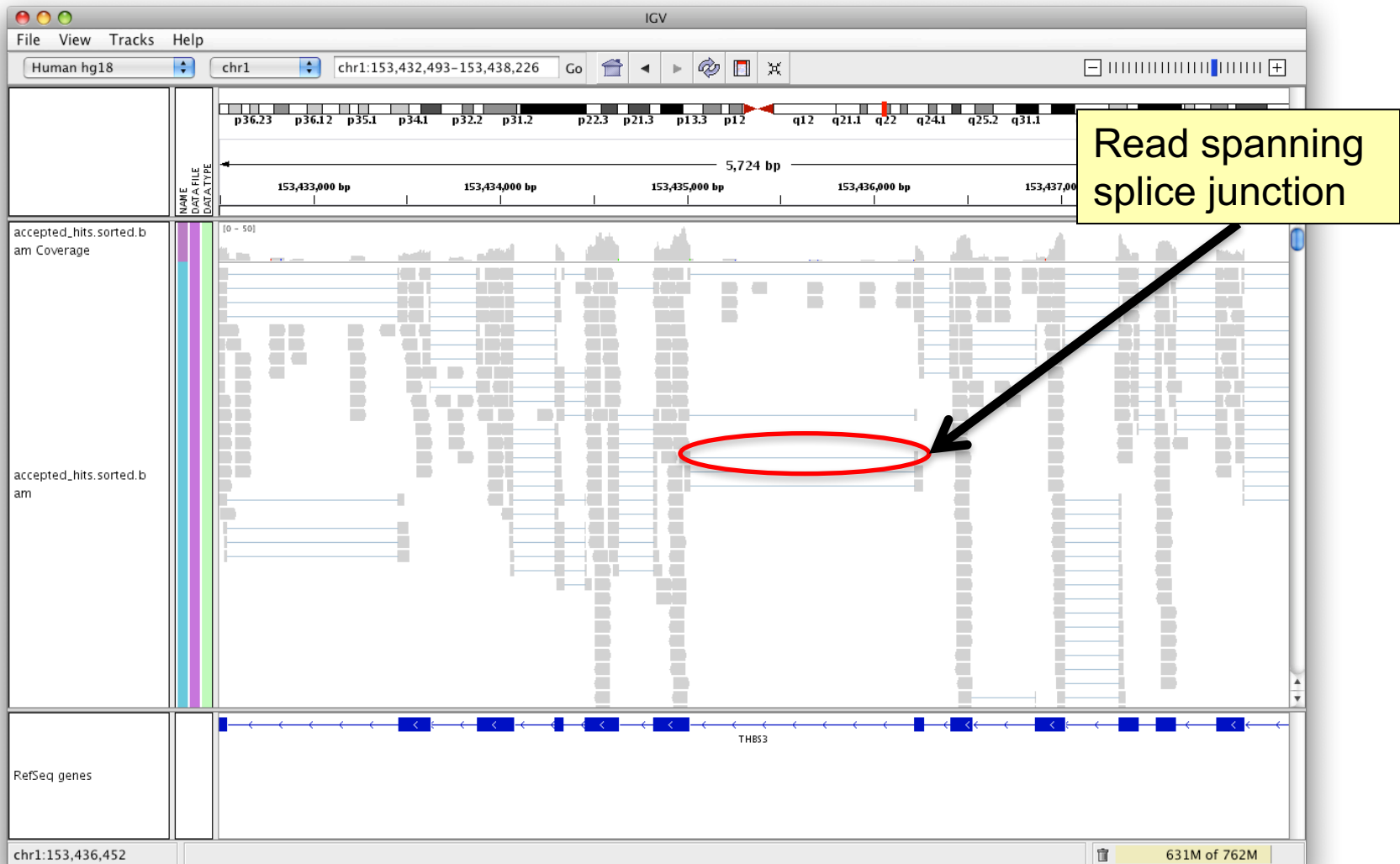
RNA-seq



RNA-seq

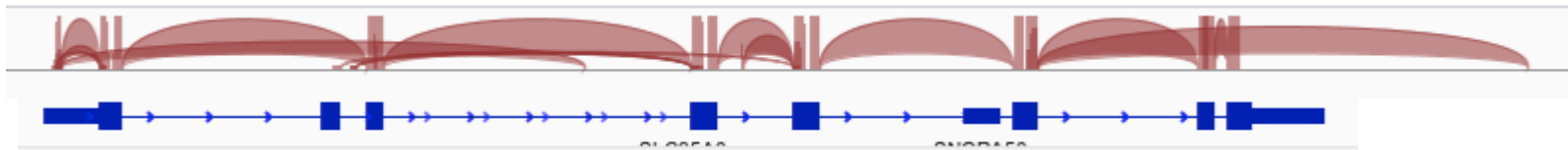


RNA-seq alignments



RNA-seq alignments

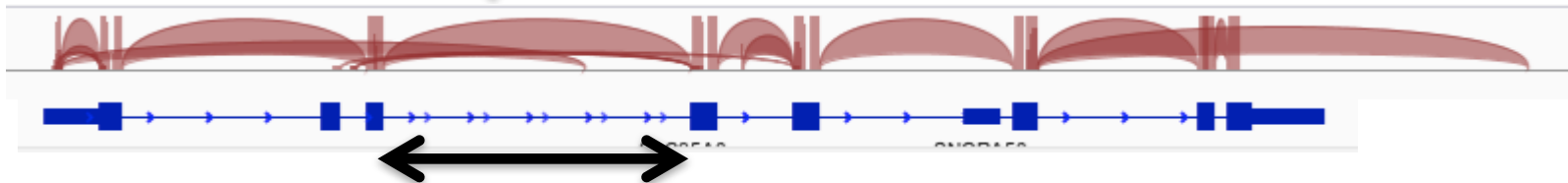
Splice junction track



RNA-seq alignments

Splice junction track

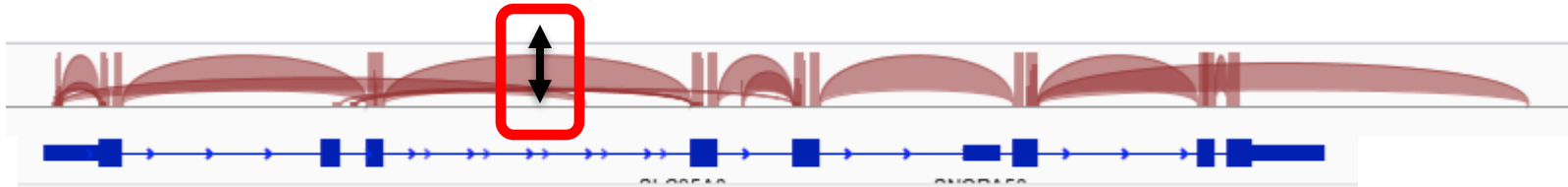
Arcs represent reads that span exon junctions



RNA-seq alignments

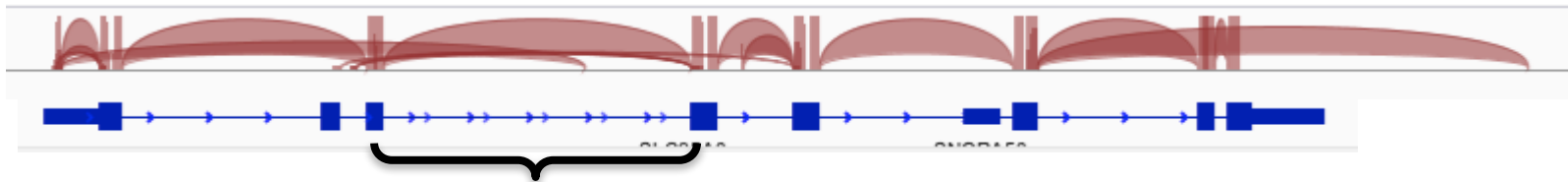
Splice junction track

Height is proportional
to number of reads



RNA-seq alignments

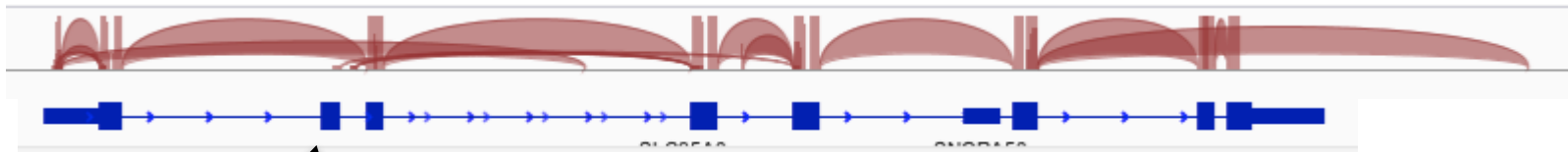
Splice junction track



Many reads connecting
these exons

RNA-seq alignments

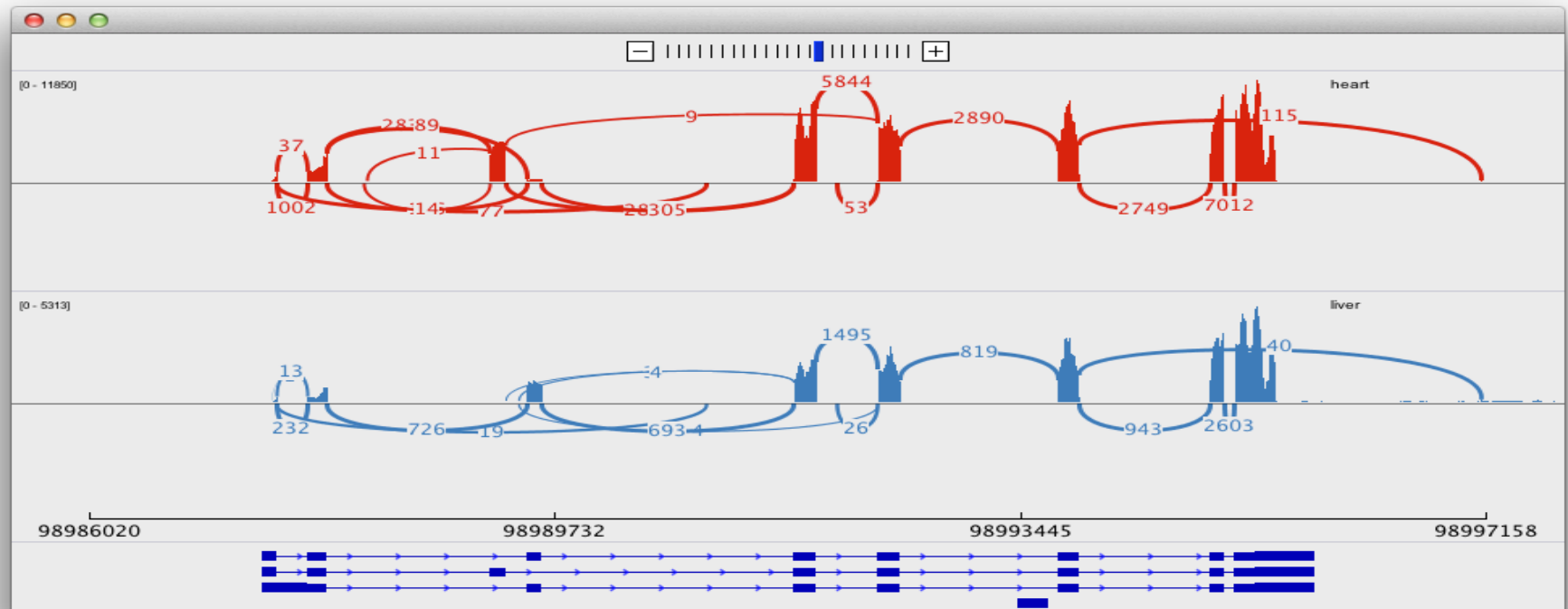
Splice junction track



Relatively few
spanning to this exon

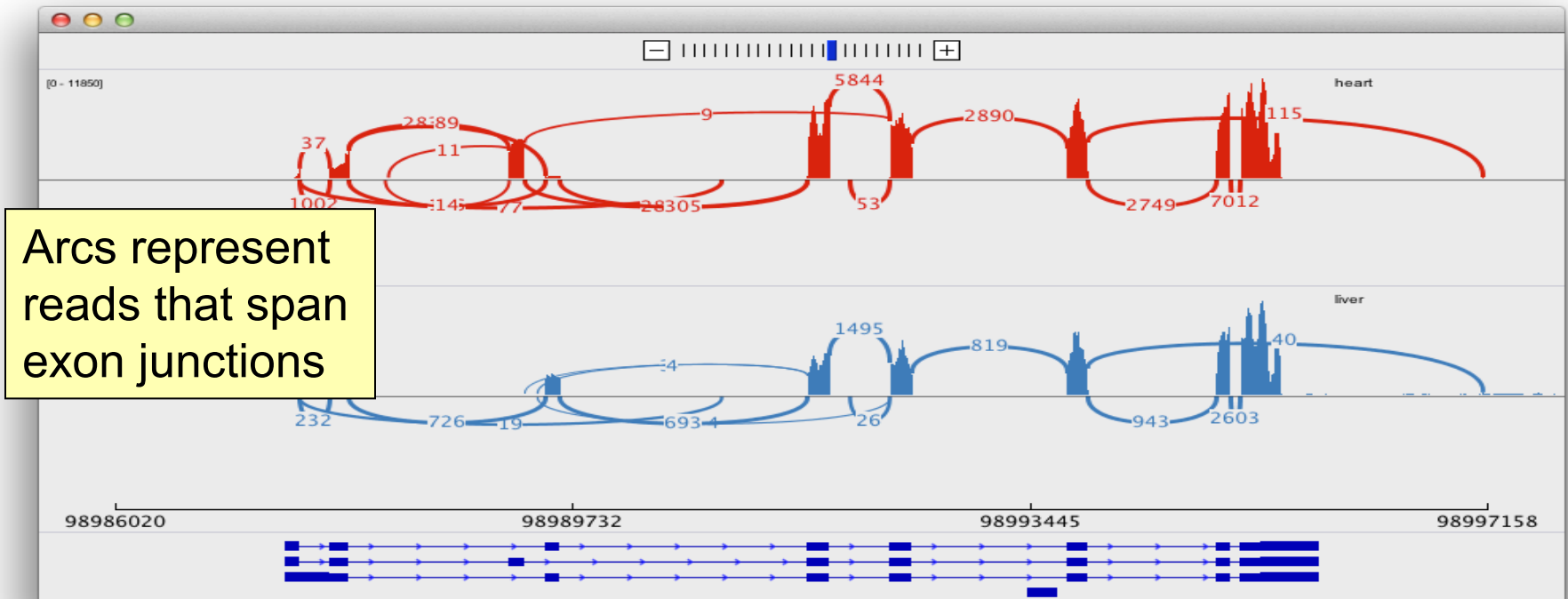
RNA-seq alignments

Sashimi Plot



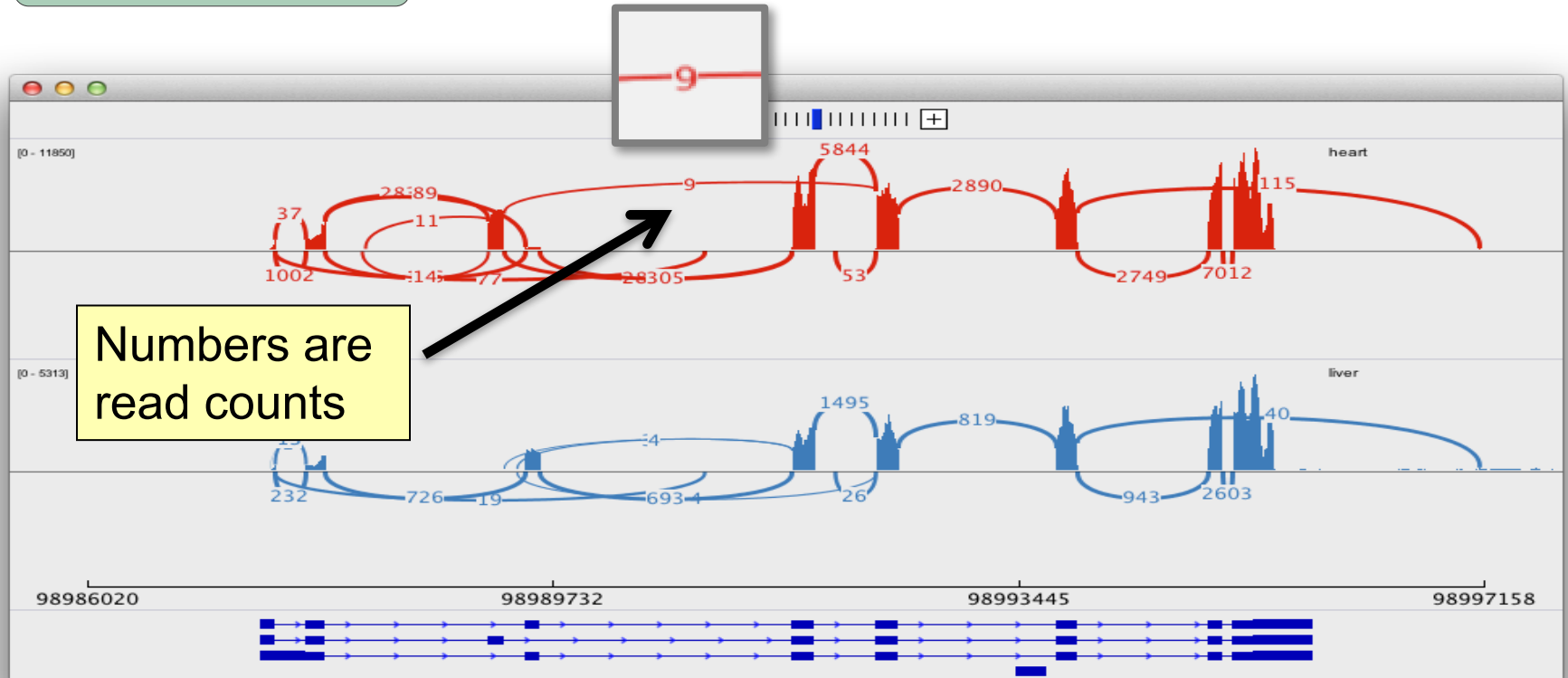
RNA-seq alignments

Sashimi Plot



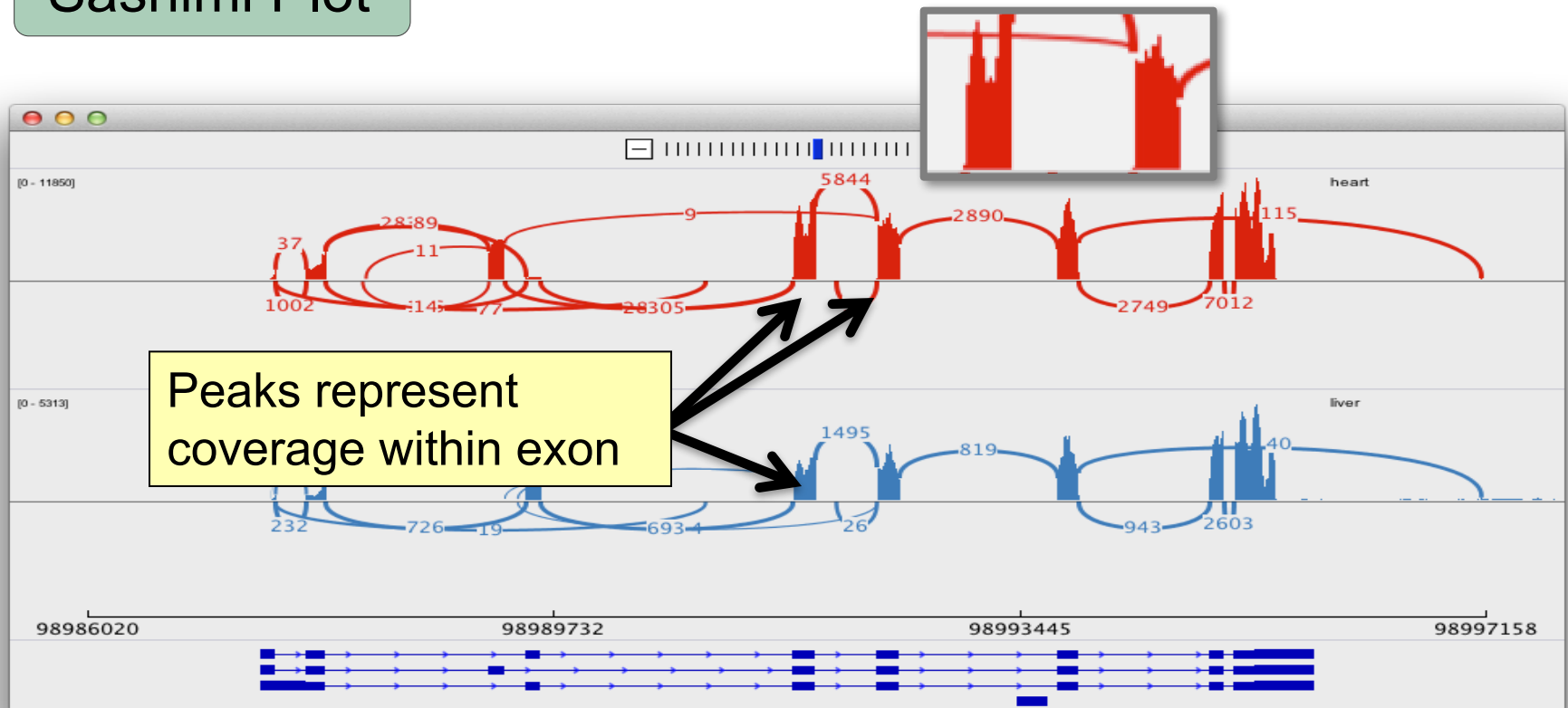
RNA-seq alignments

Sashimi Plot



RNA-seq alignments

Sashimi Plot



Sashimi plot

Viewing RNA splicing with Sashimi Plots

Reference:

Katz et al.

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

Bioinformatics (2015)

Viewing RNA-seq data

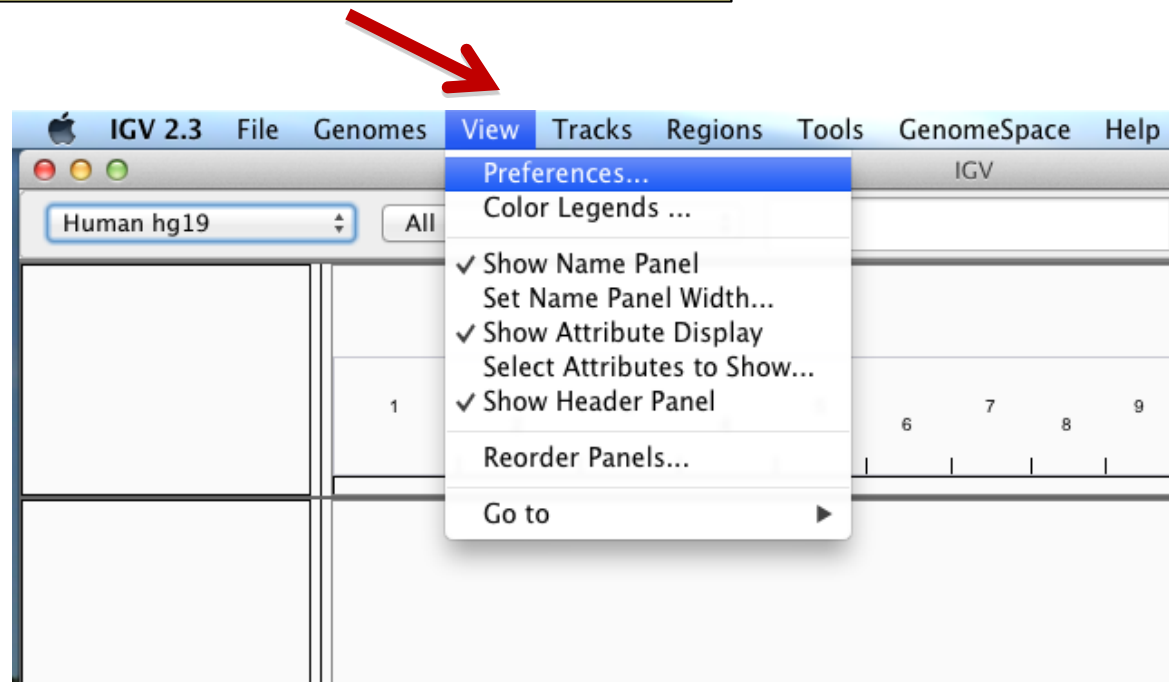
Hands-on exercise

Examine tissue-specific alternative splicing.

Data: Illumina BodyMap 2.0

See handout

Select **View > Preferences...**



Click **Alignments** tab

The screenshot shows the IGV application window. The top navigation bar includes tabs for Variants, Alignments, Proxies, Ion Torrent, and Advanced. The 'Alignments' tab is selected and highlighted with a red rectangle. A red arrow points from a yellow callout box containing the text 'Click Alignments tab' to this tab. The main settings dialog box is open, displaying various options for the Alignment Track, Coverage Track, and Splice Junction Track. The Alignment Track Options section includes checkboxes for 'Downsample reads', 'Shade mismatched bases by quality', 'Flag insertions larger than', 'Filter duplicate reads', 'Filter vendor failed reads', 'Filter secondary alignments', 'Filter supplementary alignments', 'Flag unmapped pairs', 'Show soft-clipped bases', and 'Show center line'. The Coverage Track Options section includes a checkbox for 'Quality weight allele fraction'. The Splice Junction Track Options section includes a checkbox for 'Show flanking regions'. The Insert Size Options section includes checkboxes for 'Compute' and 'Maximum (percentile)'. The background shows a genomic track with a gene model and a coverage plot.

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

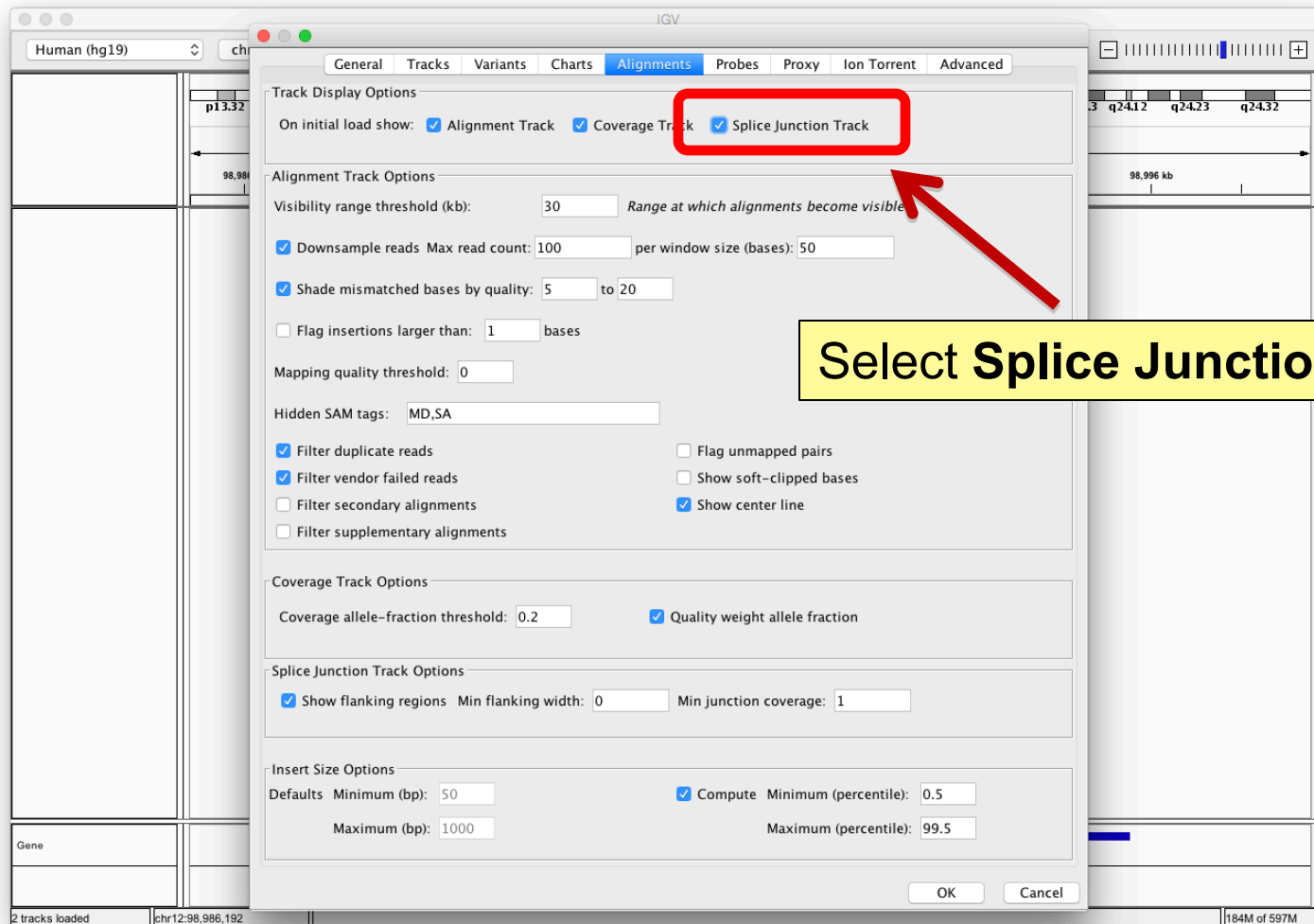
☒ Show flanking regions Min flanking width: 0 Min junction coverage: 1

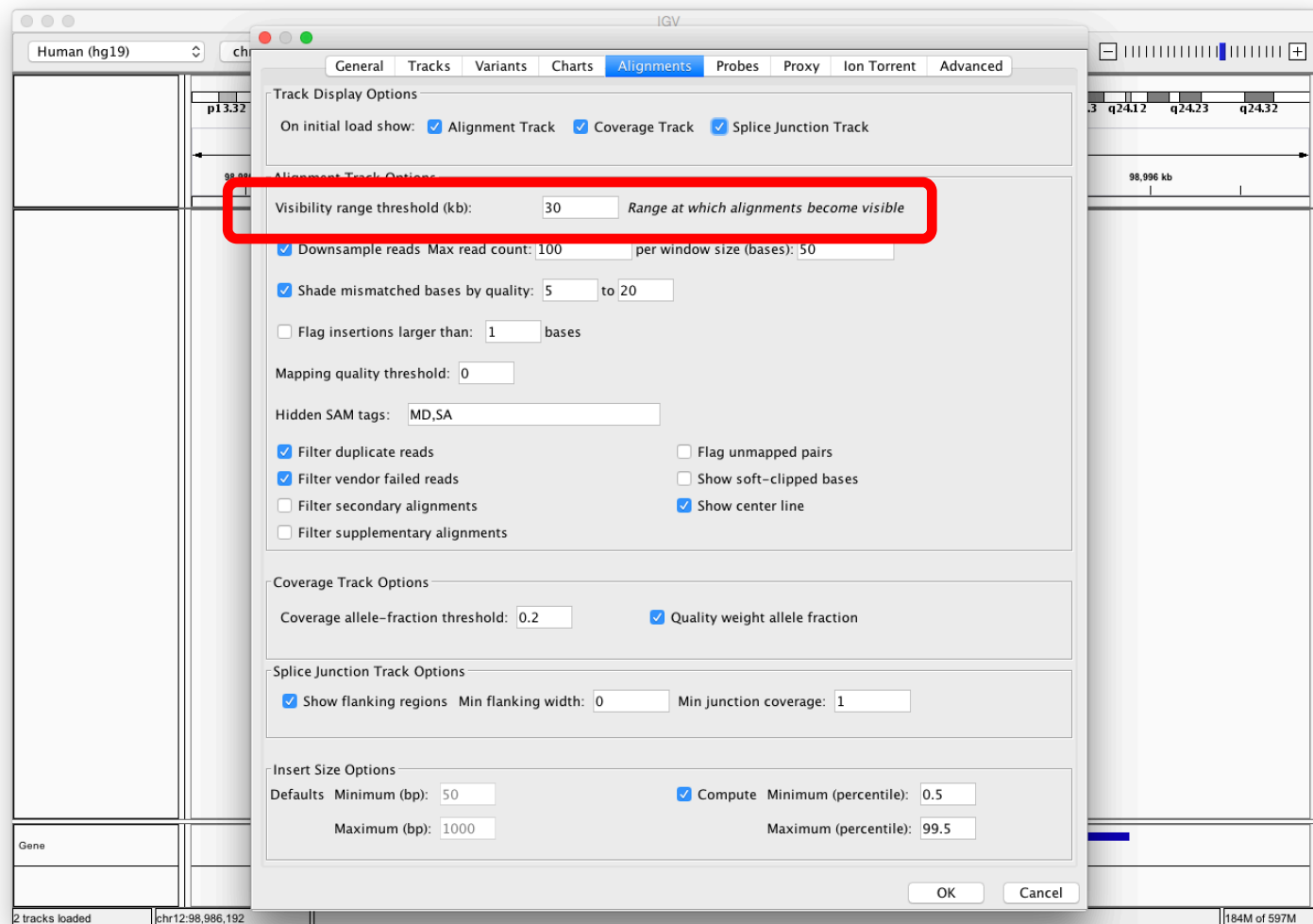
Insert Size Options

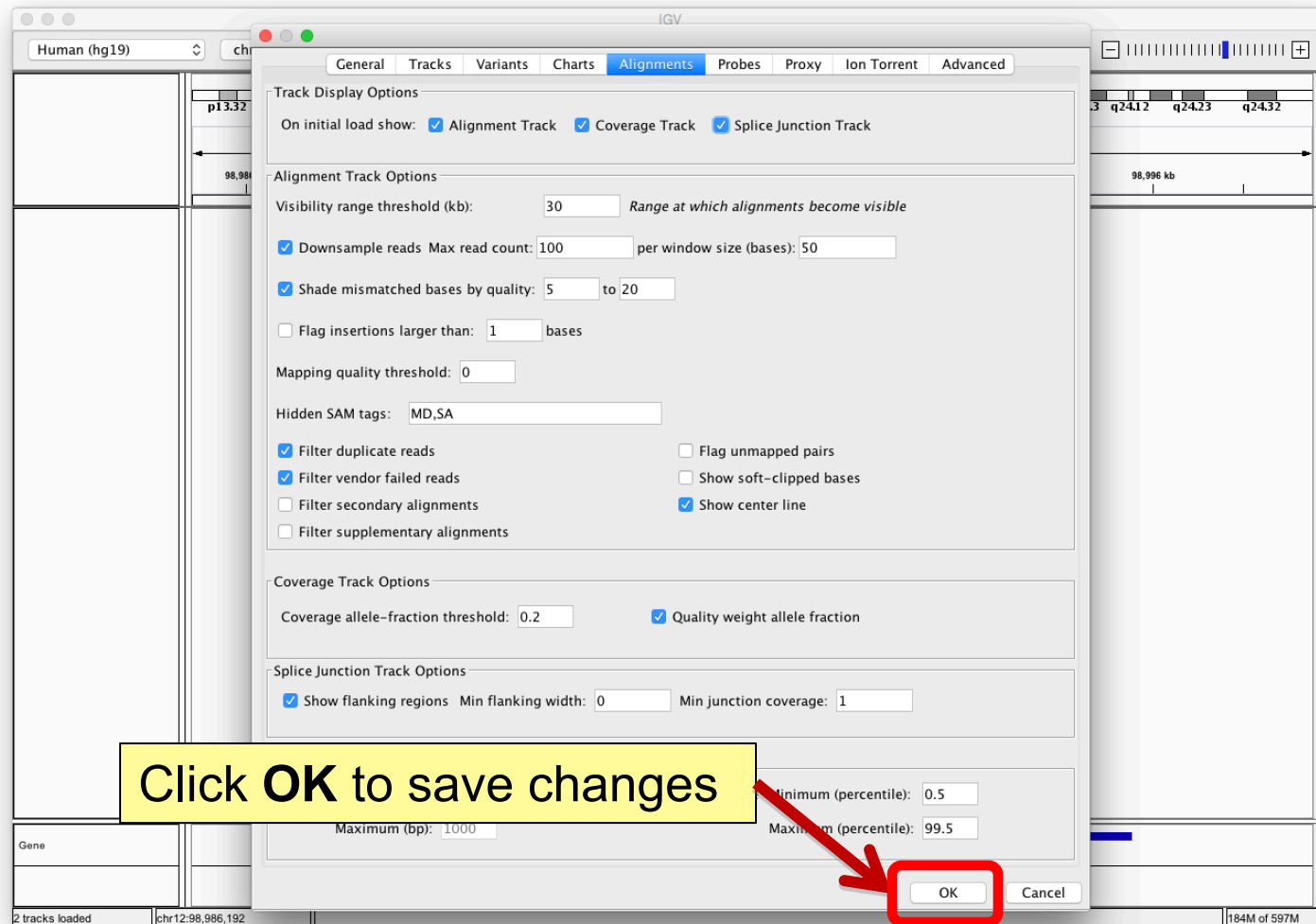
Defaults Minimum (bp): 50 ☒ Compute Minimum (percentile): 0.5

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

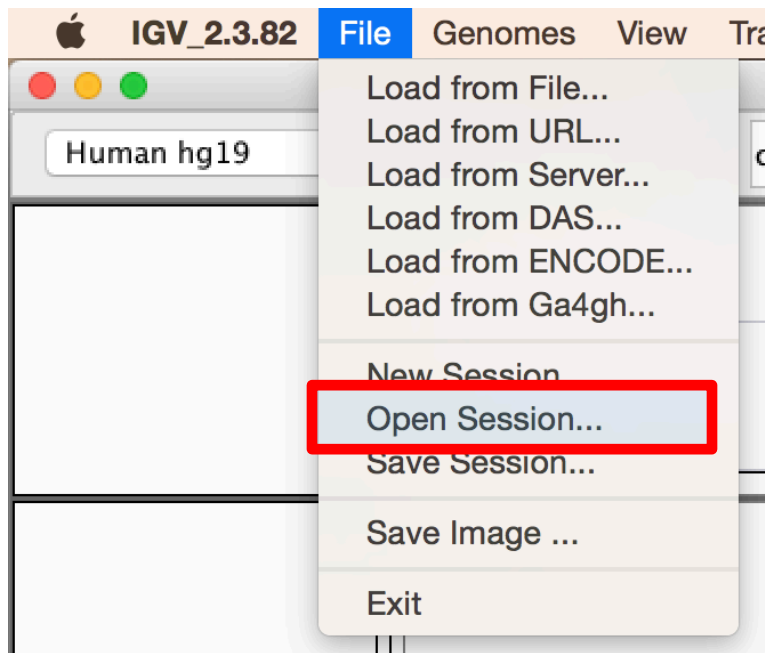
OK Cancel



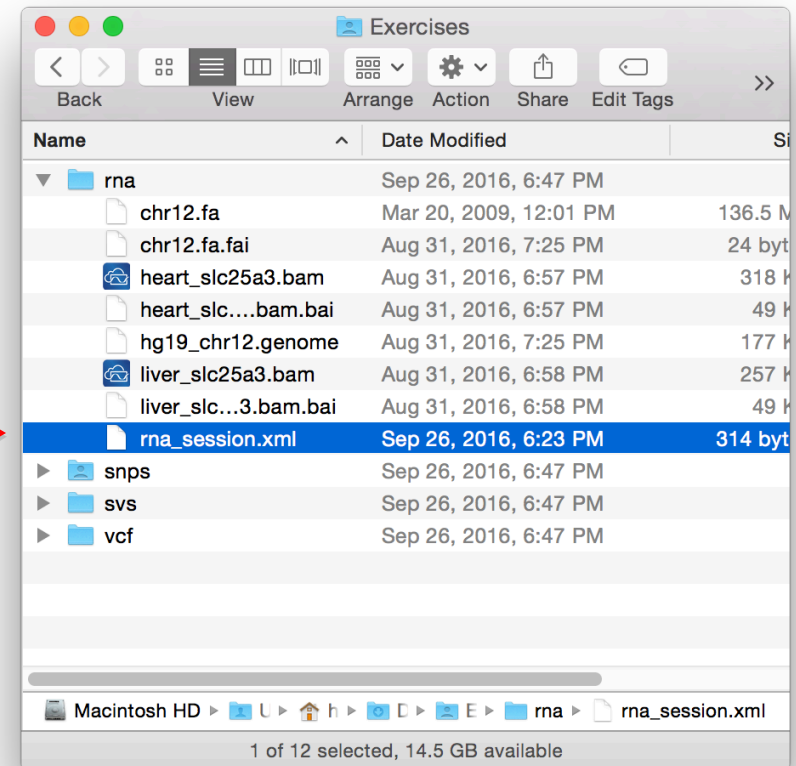




- Click **File > Open Session**

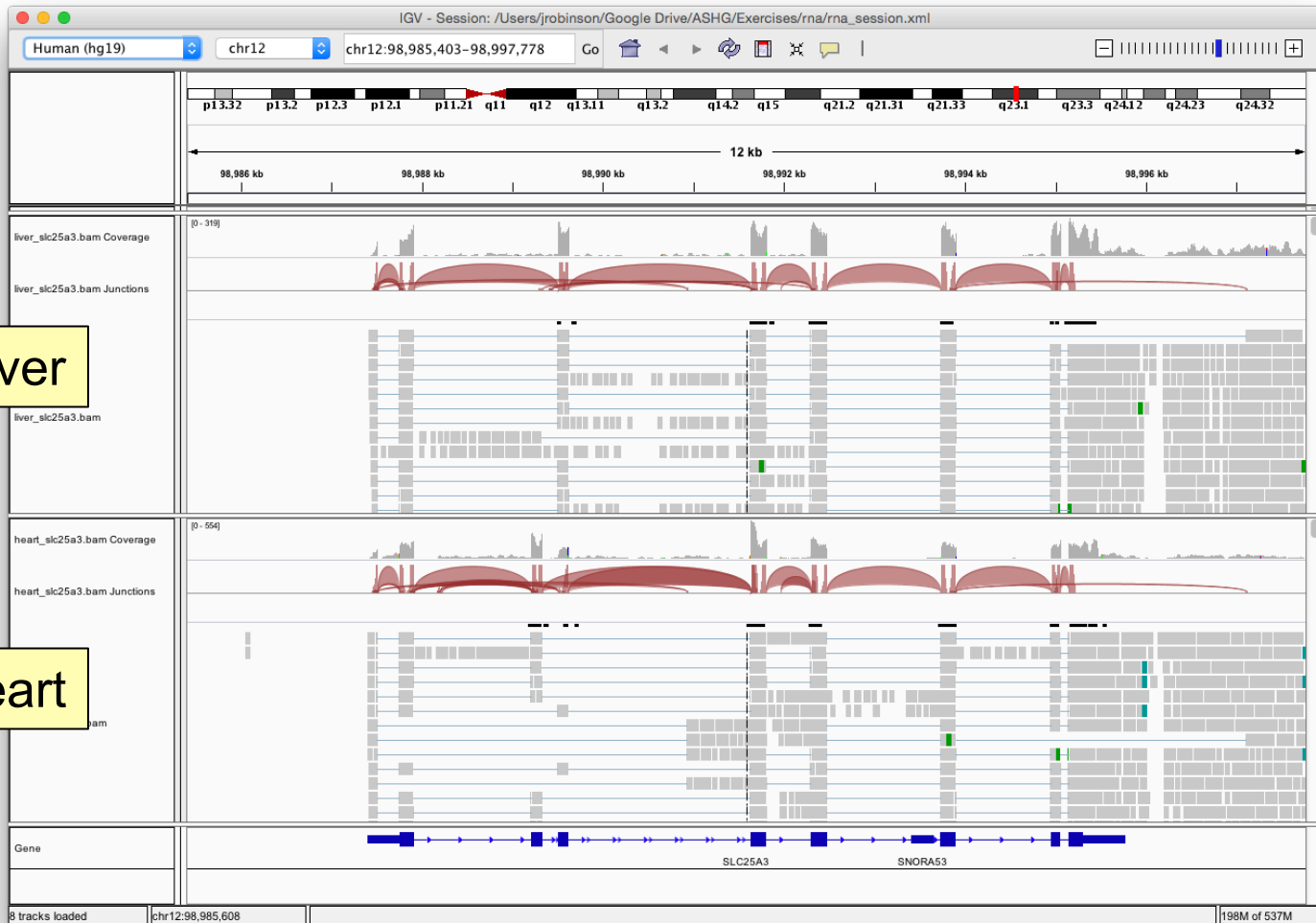


- Navigate to the workshop data folder, and select **rna / rna_session.xml**



Viewing RNA-seq data

Hands-on exercise



Viewing RNA-seq data

Hands-on exercise

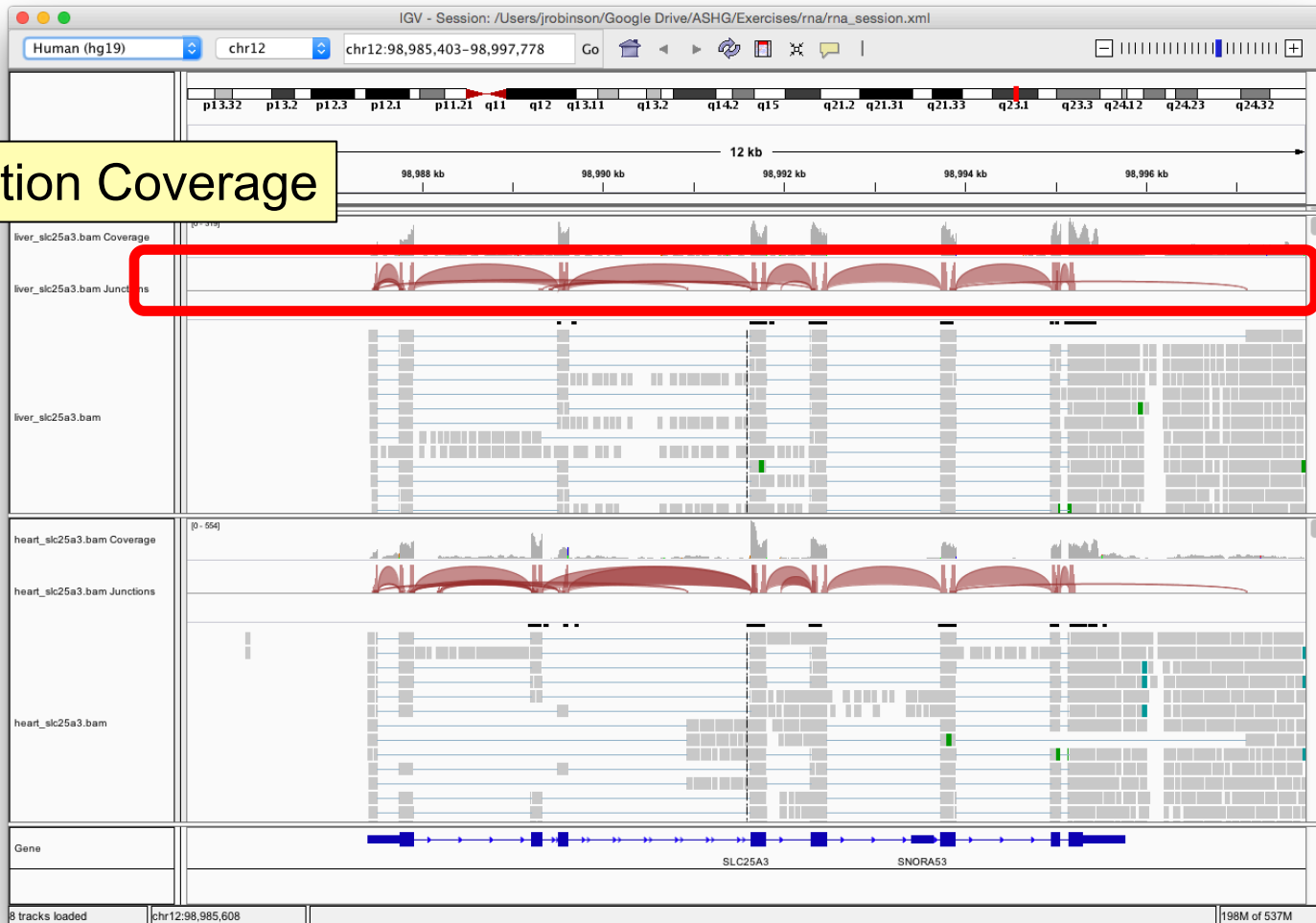
Coverage



Viewing RNA-seq data

Hands-on exercise

Junction Coverage



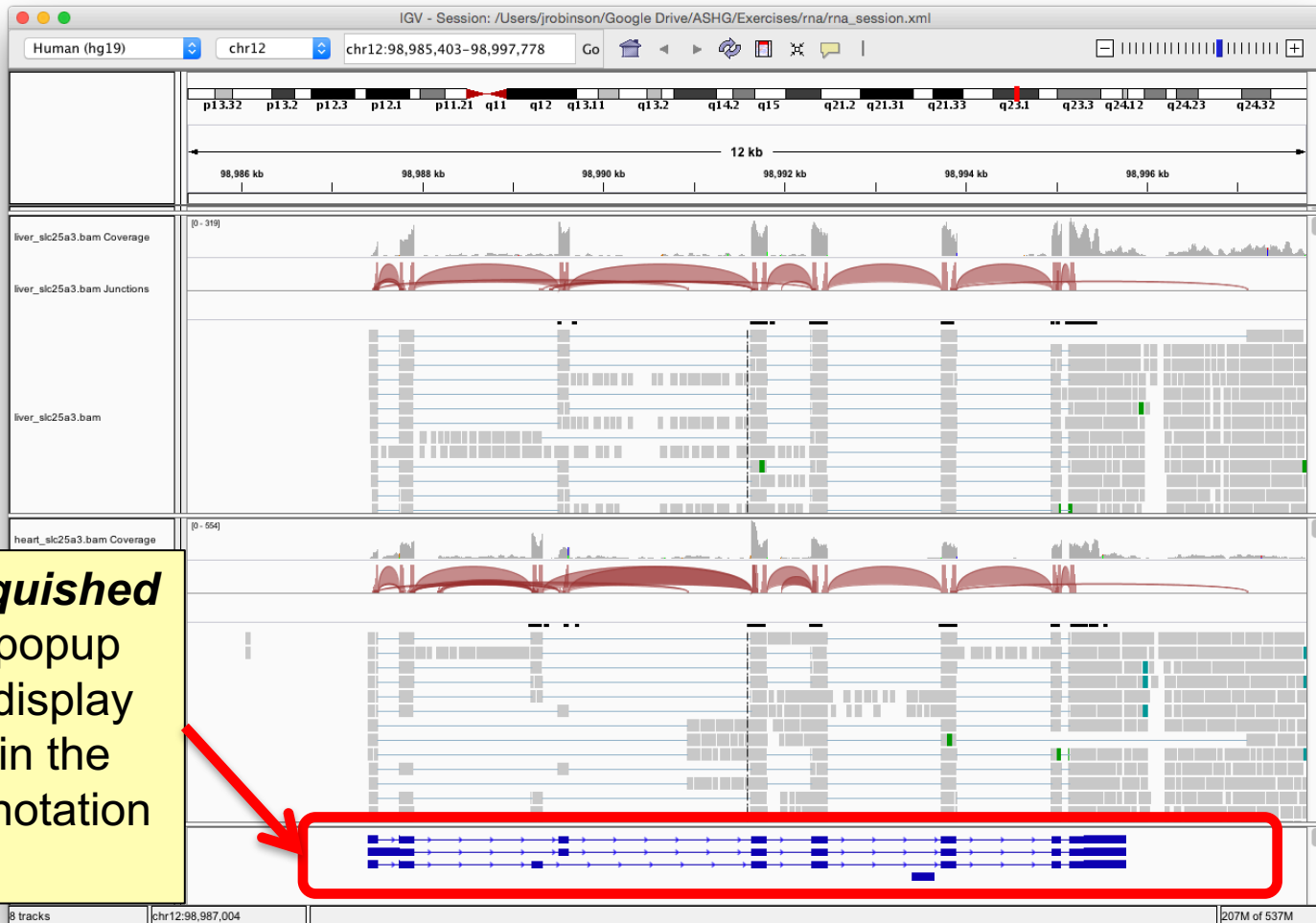
Viewing RNA-seq data

Hands-on exercise



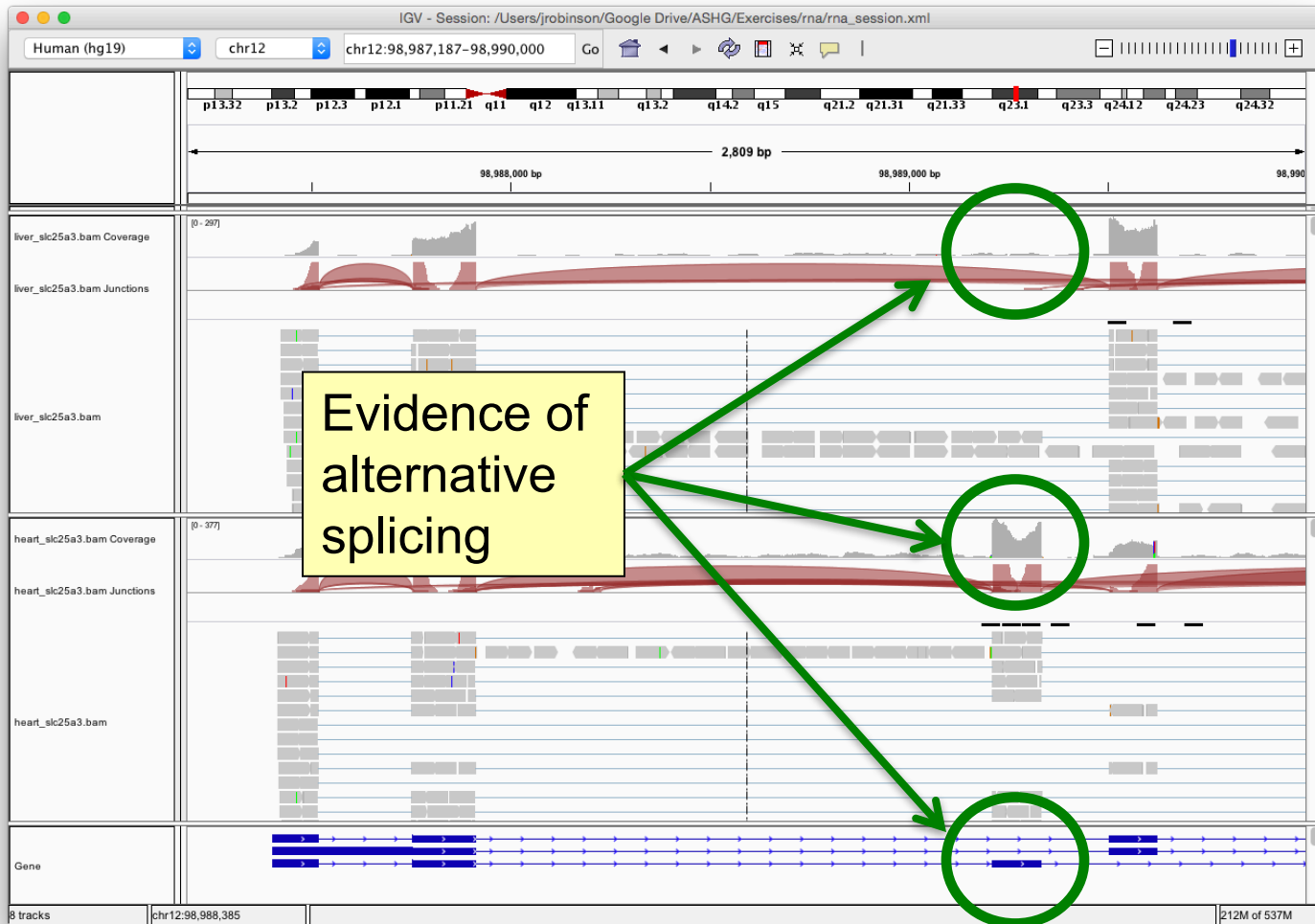
Viewing RNA-seq data

Hands-on exercise

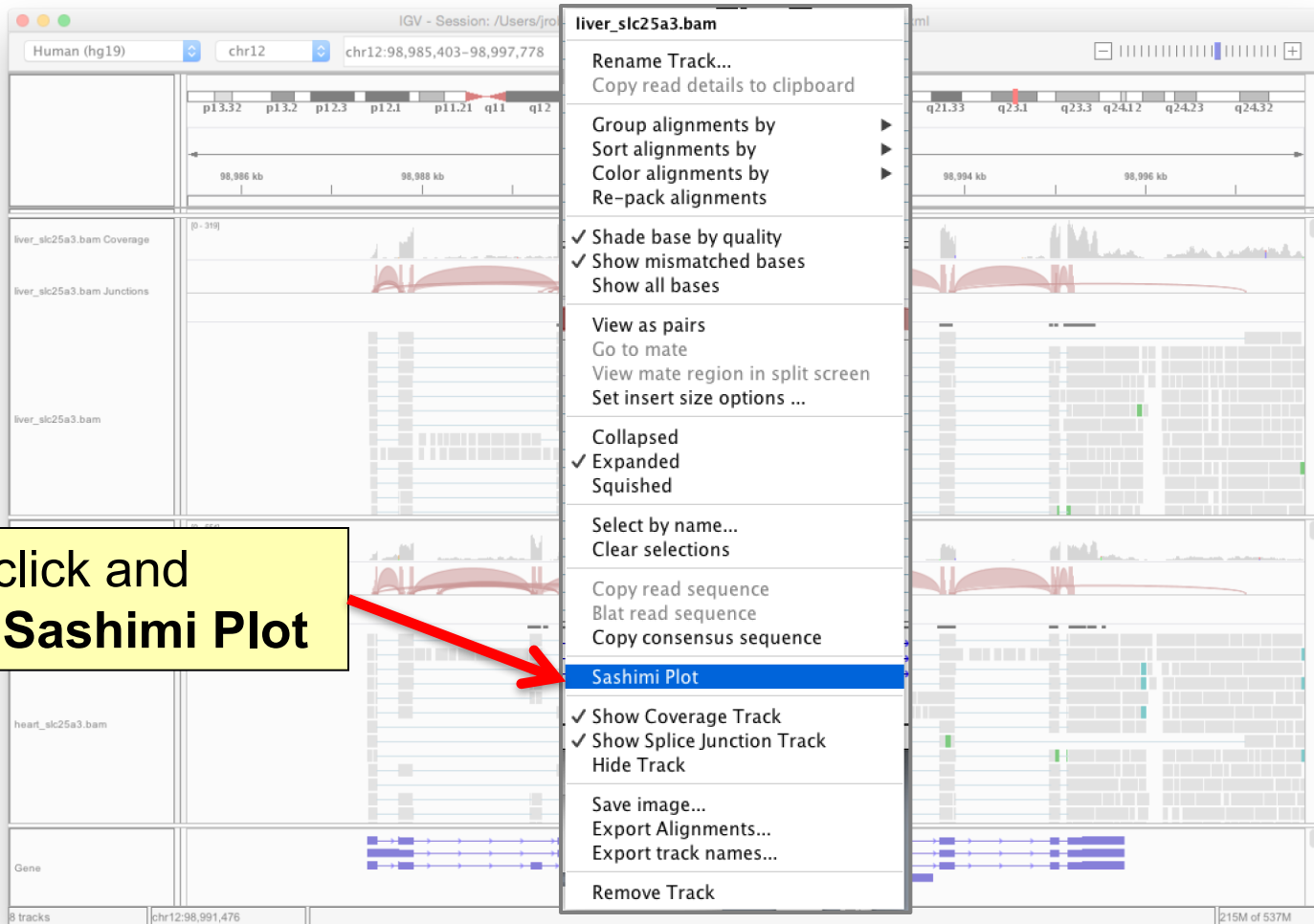


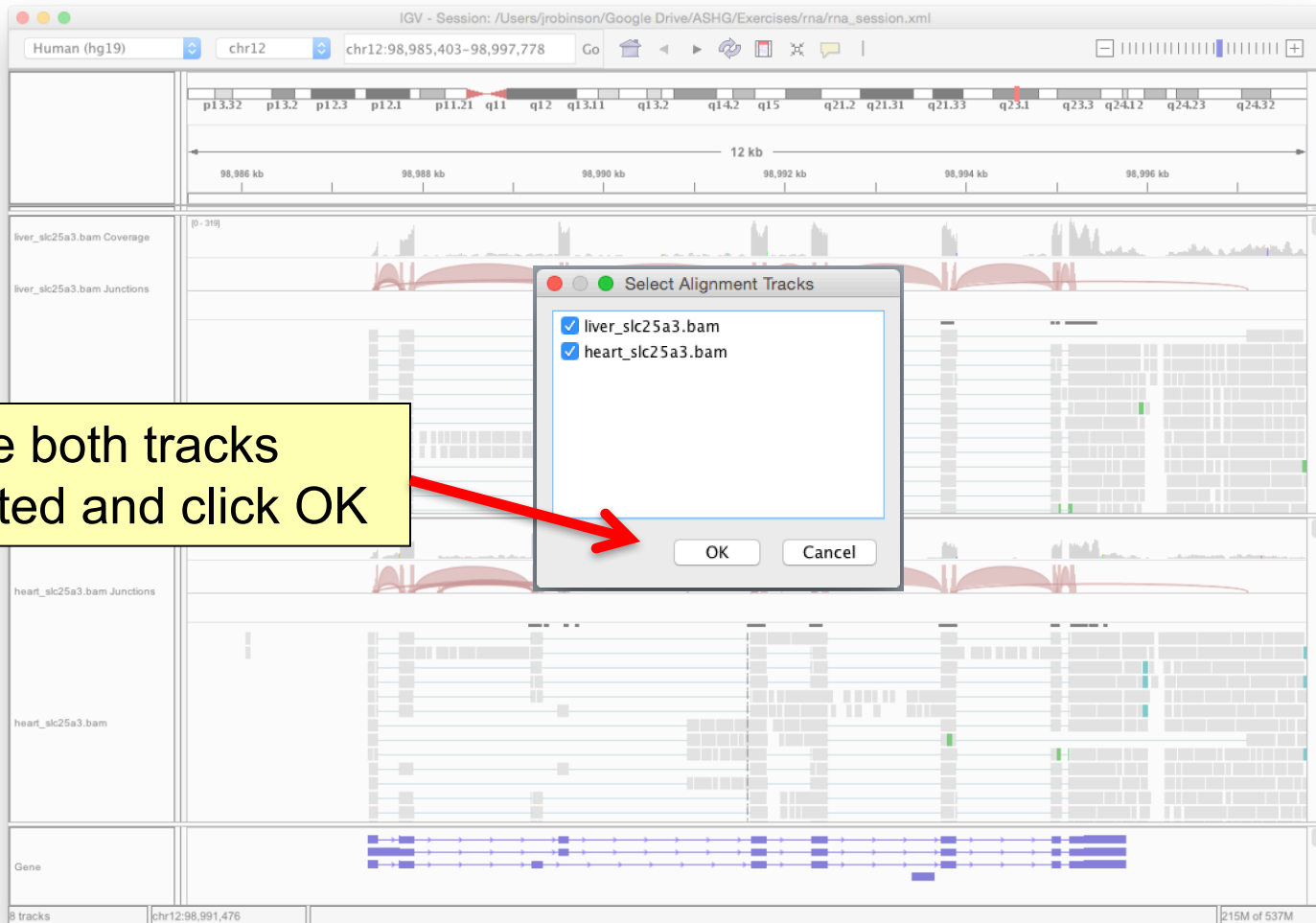
Viewing RNA-seq data

Hands-on exercise



Right-click and
select **Sashimi Plot**



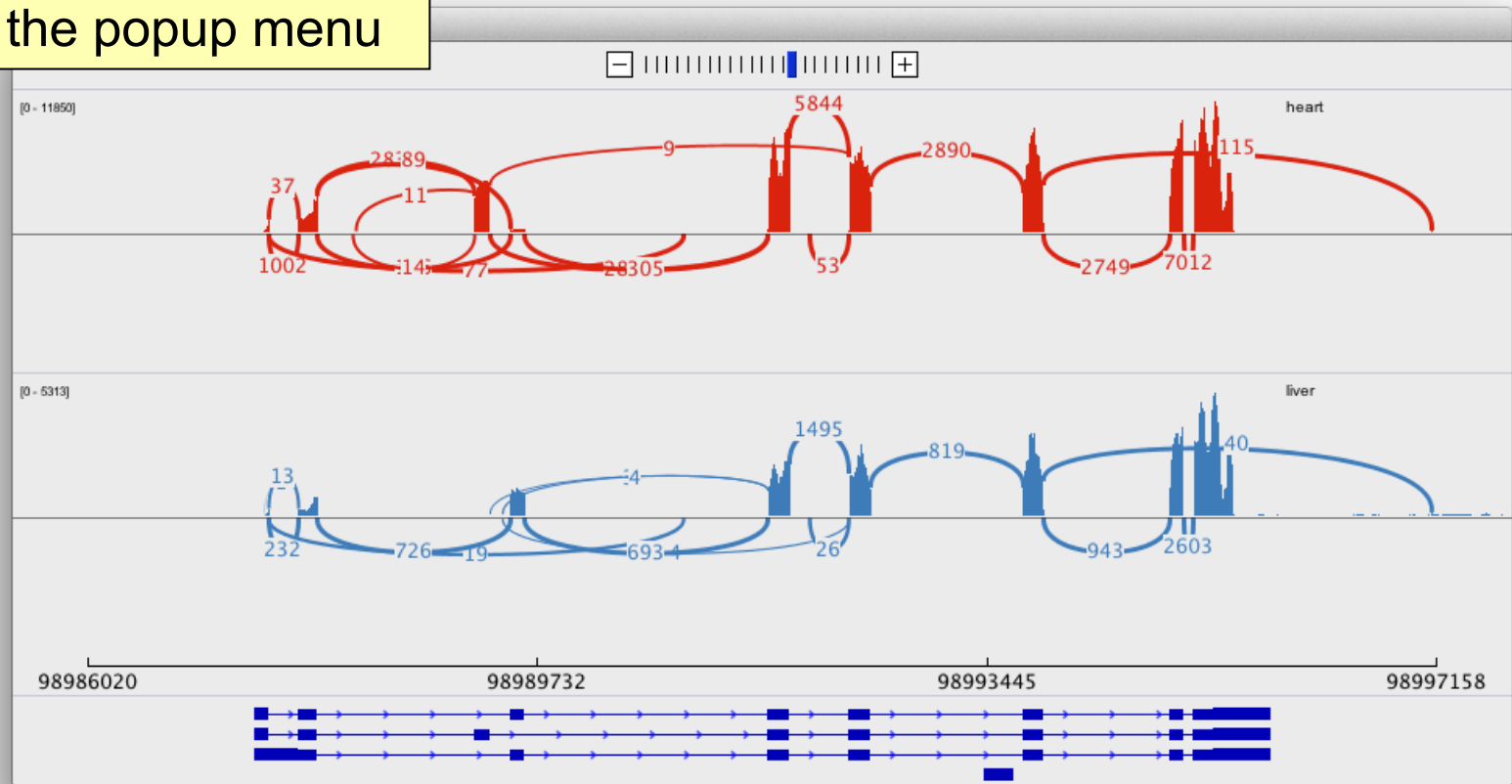


Leave both tracks
selected and click OK

Viewing RNA-seq data

Hands-on exercise

Launch **Sashimi Plot**
from the popup menu



Viewing RNA-seq data

Hands-on exercise

Filter and zoom in



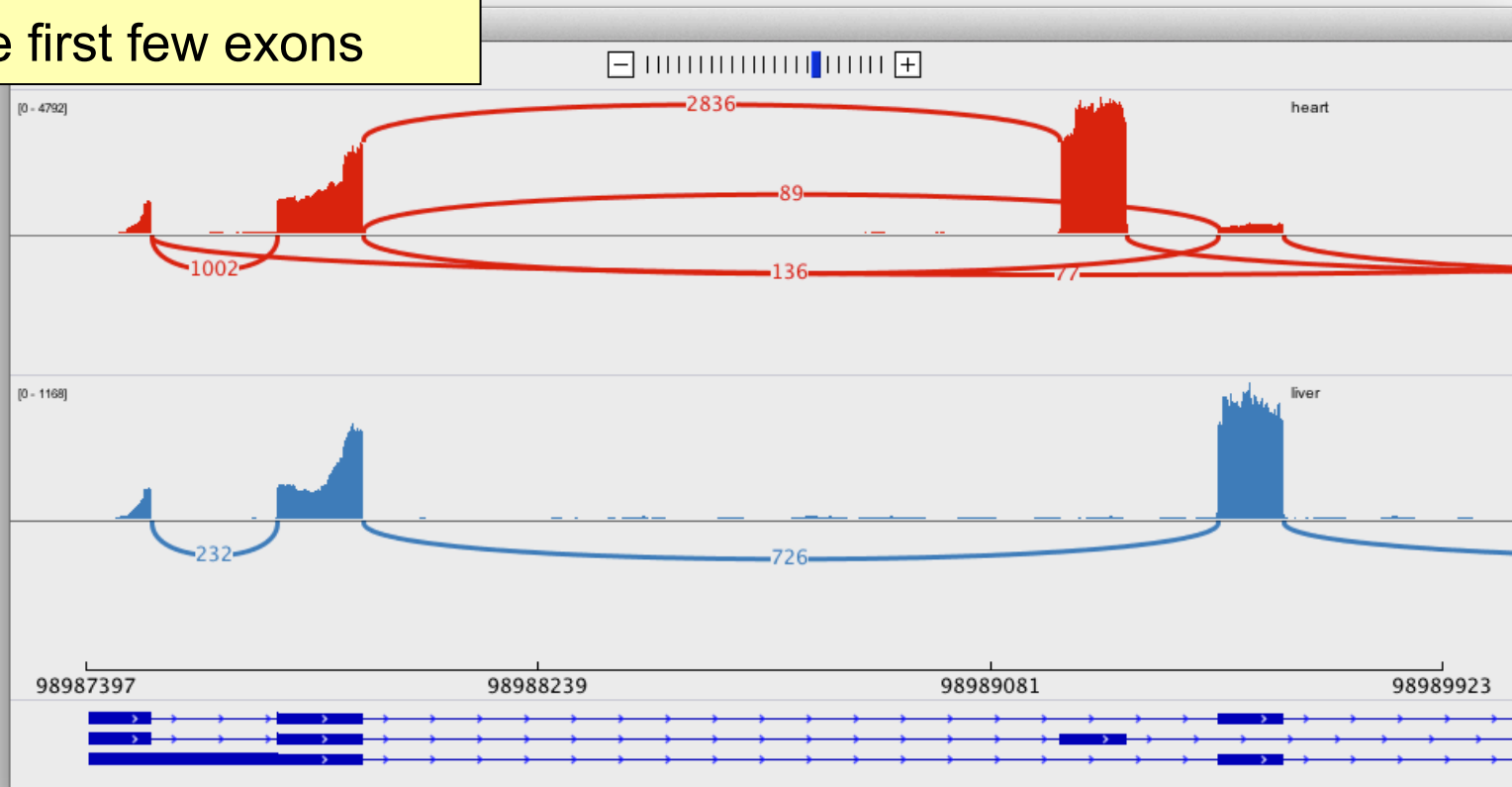
Set Junction Coverage Min = 50



Viewing RNA-seq data

Hands-on exercise

Filter and zoom in on
the first few exons



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

DNA or
cDNA



fragment



read from
each end



align to
reference



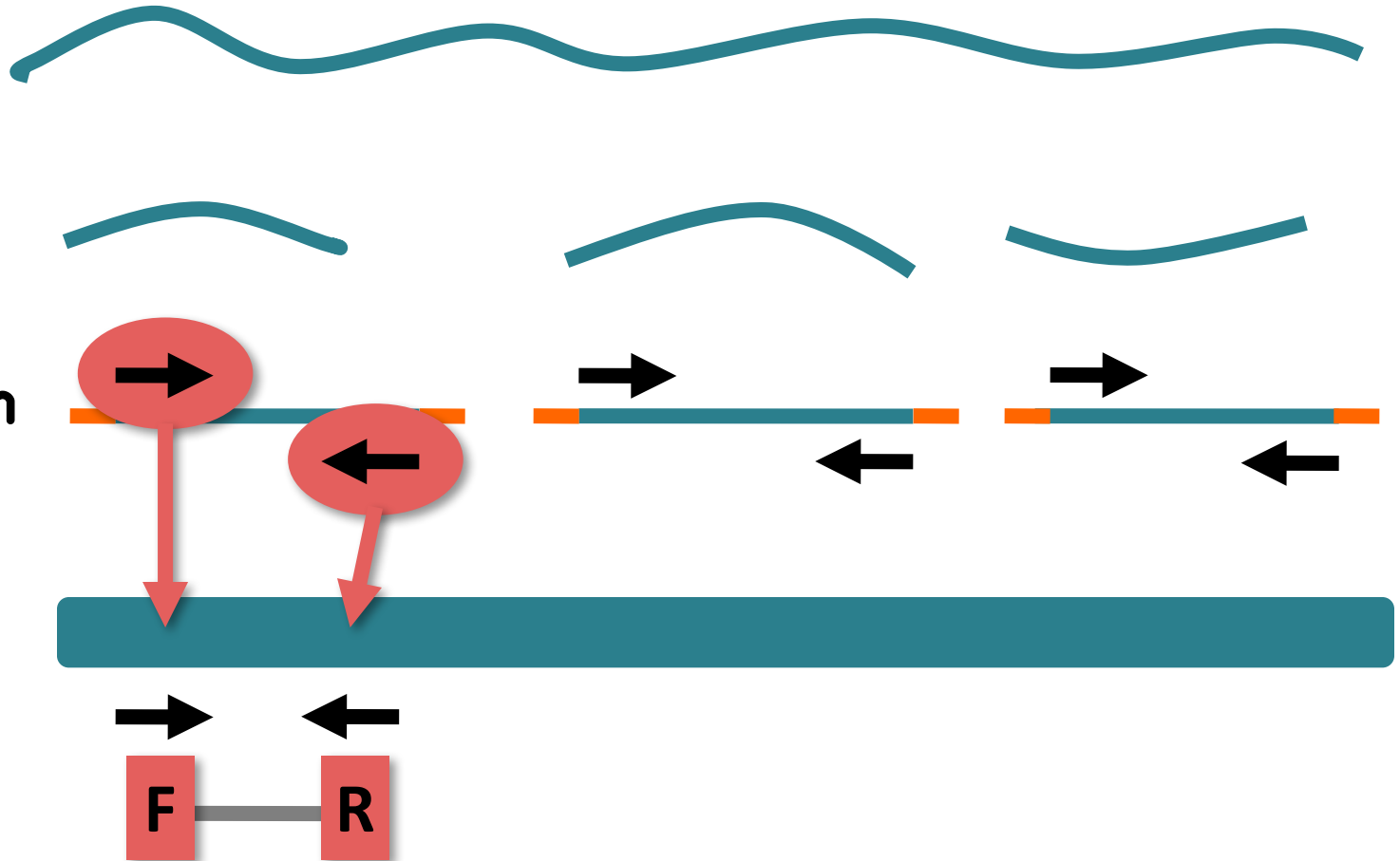
Paired-end sequencing

DNA or
cDNA

fragment

read from
each end

align to
reference



Paired-end sequencing

DNA or
cDNA



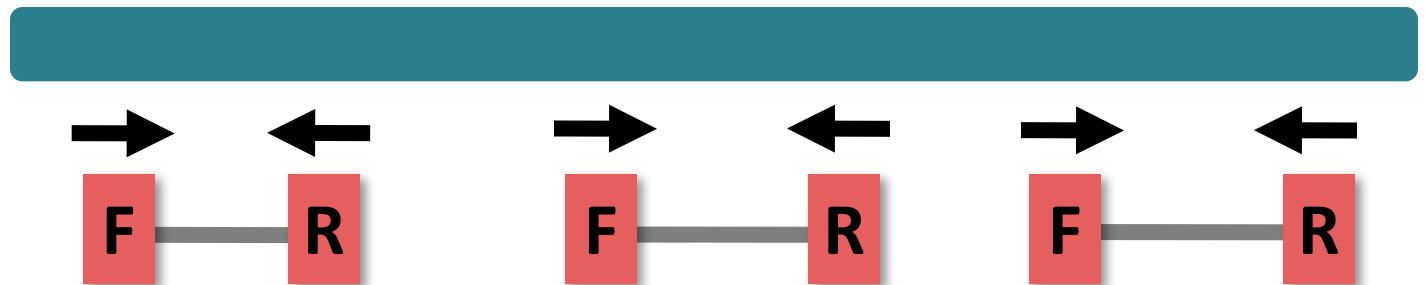
fragment



read from
each end



align to
reference



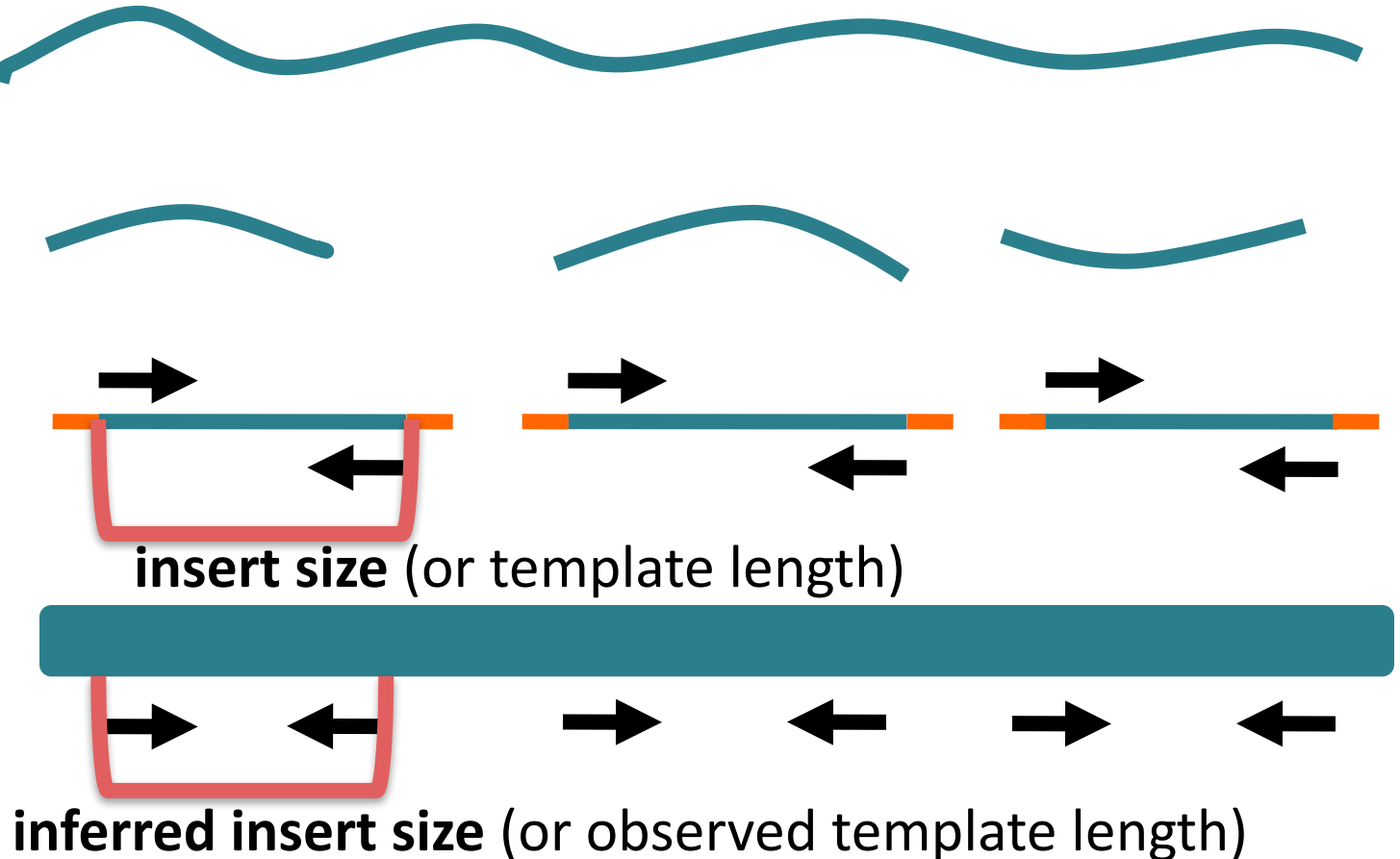
Paired-end sequencing

DNA or
cDNA

fragment

read from
each end

align to
reference



Interpreting inferred insert size

Differences between *insert size* and *inferred insert size** can be used to detect evidence of structural variants, including

- Deletions
- Insertions
- Inter-chromosomal rearrangements

* or *template length* and *observed template length*

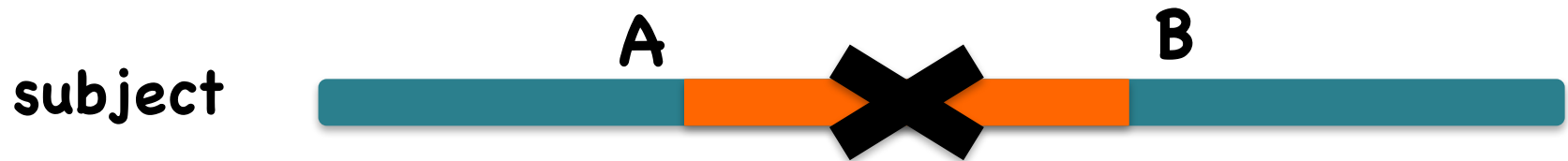
Interpreting inferred insert size

Differences between *insert size* and *inferred insert size** can be used to detect evidence of structural variants, including

- Deletions
- Insertions
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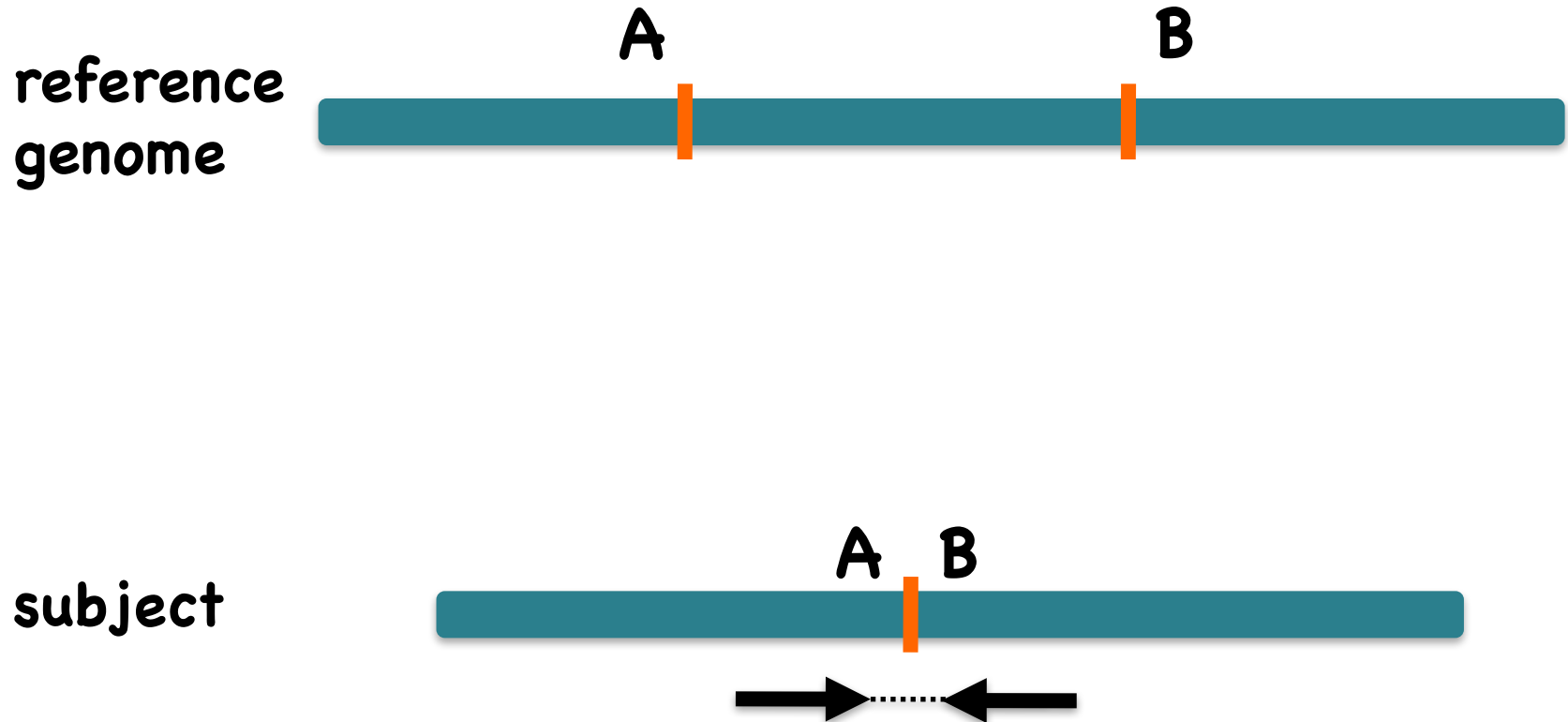
Deletion



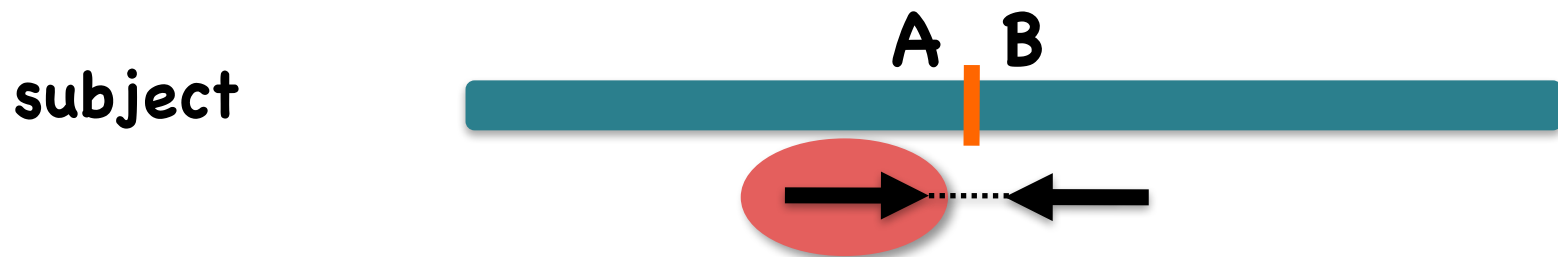
Deletion



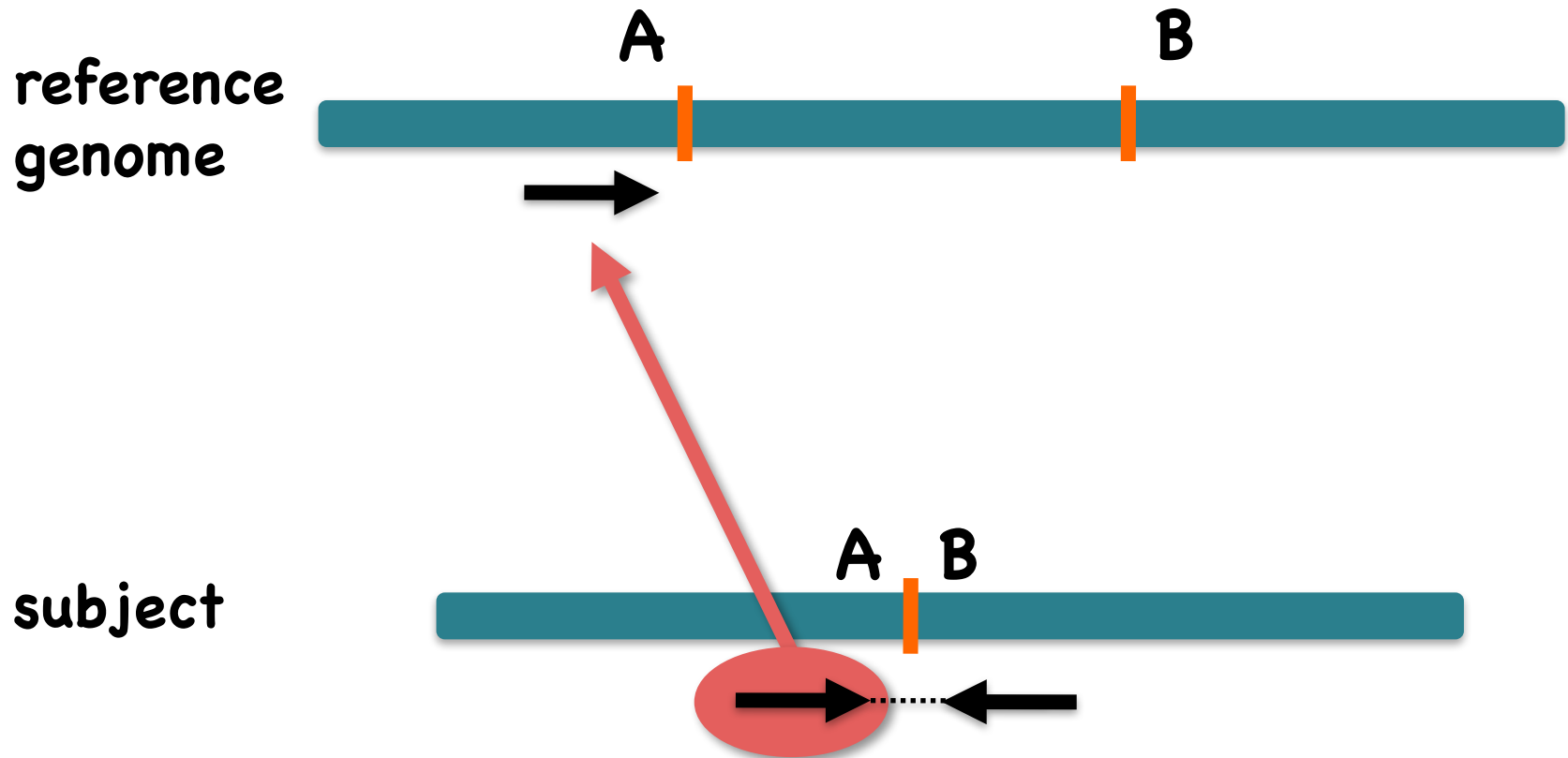
Deletion



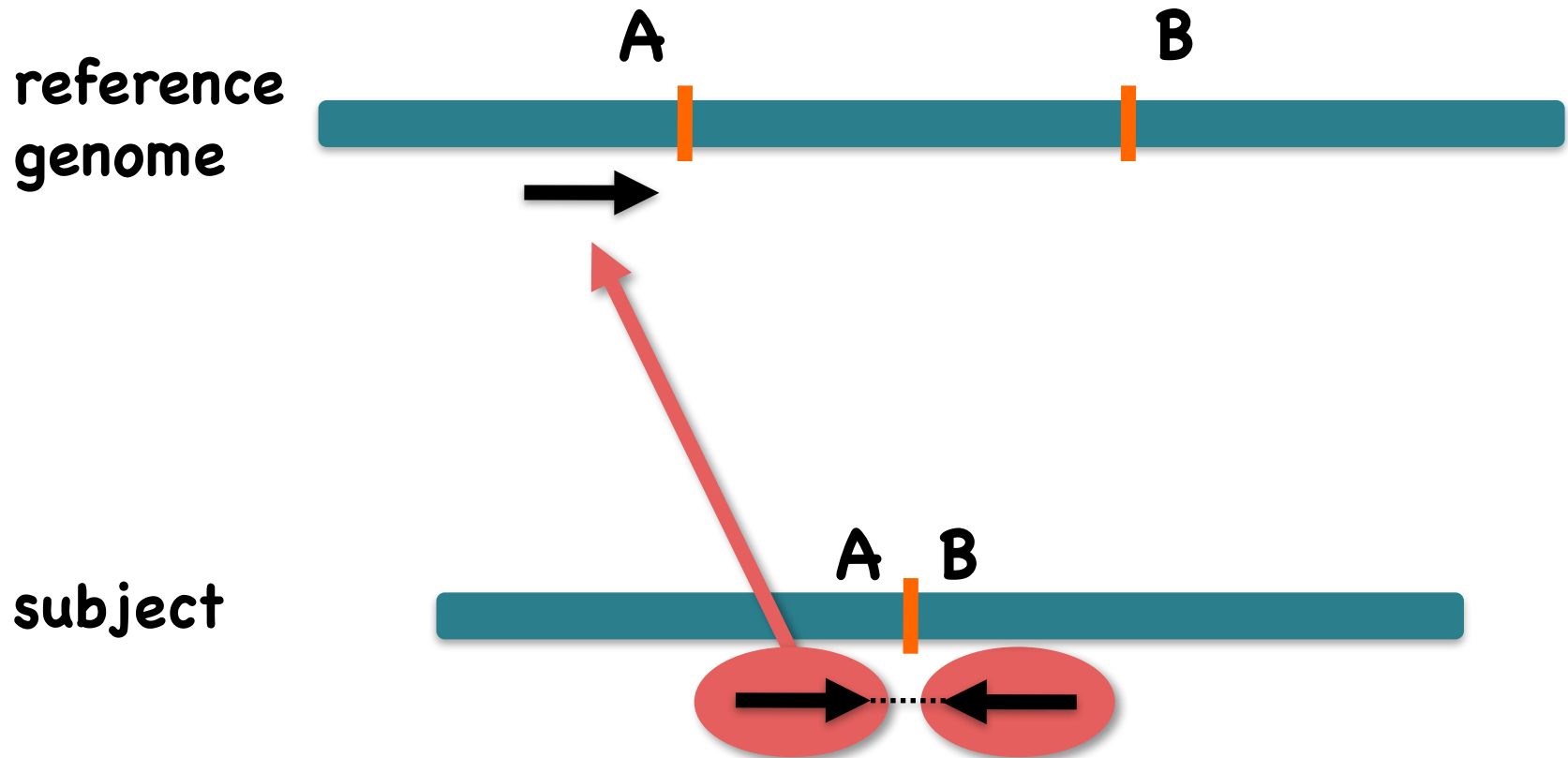
Deletion



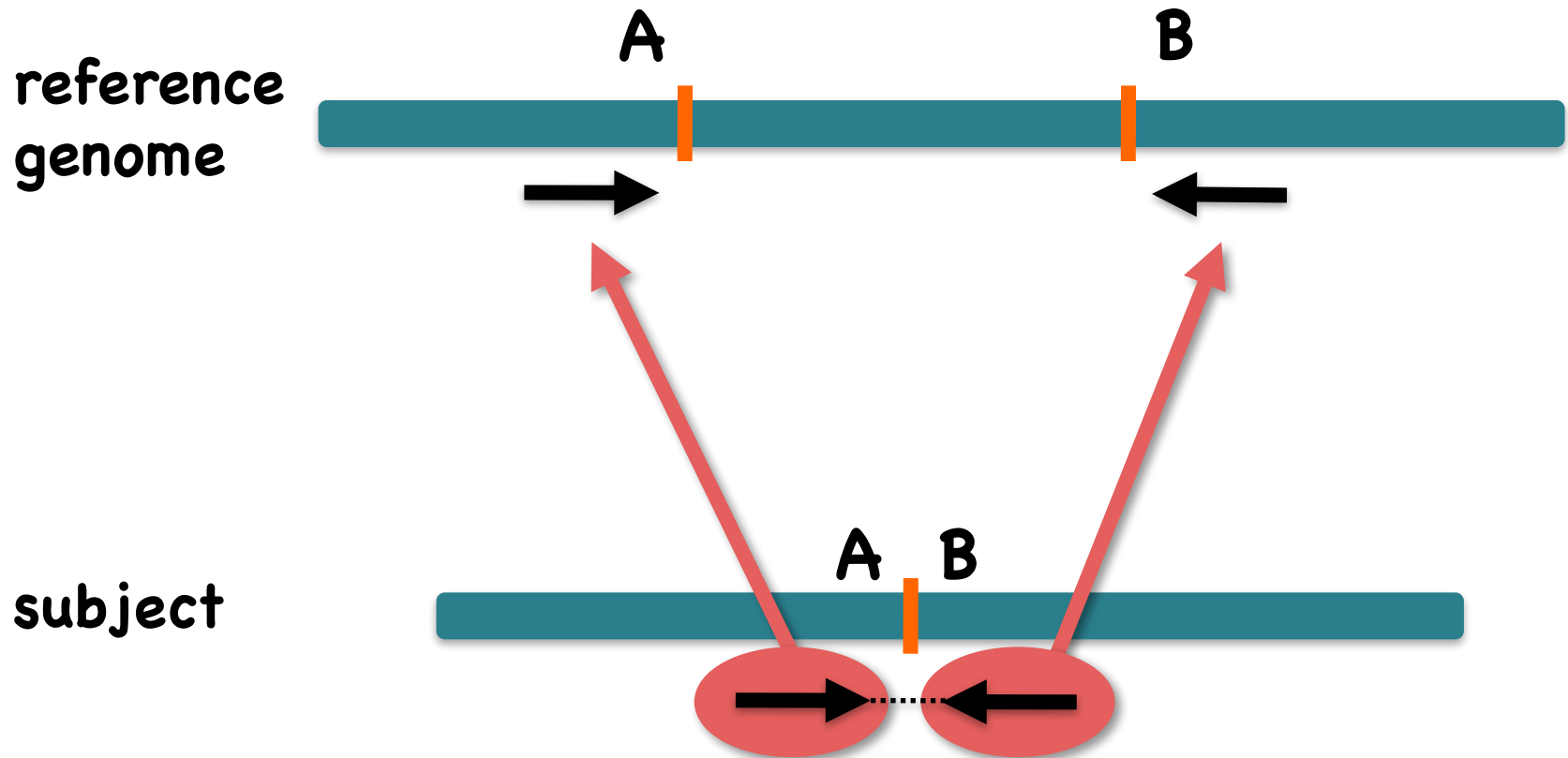
Deletion



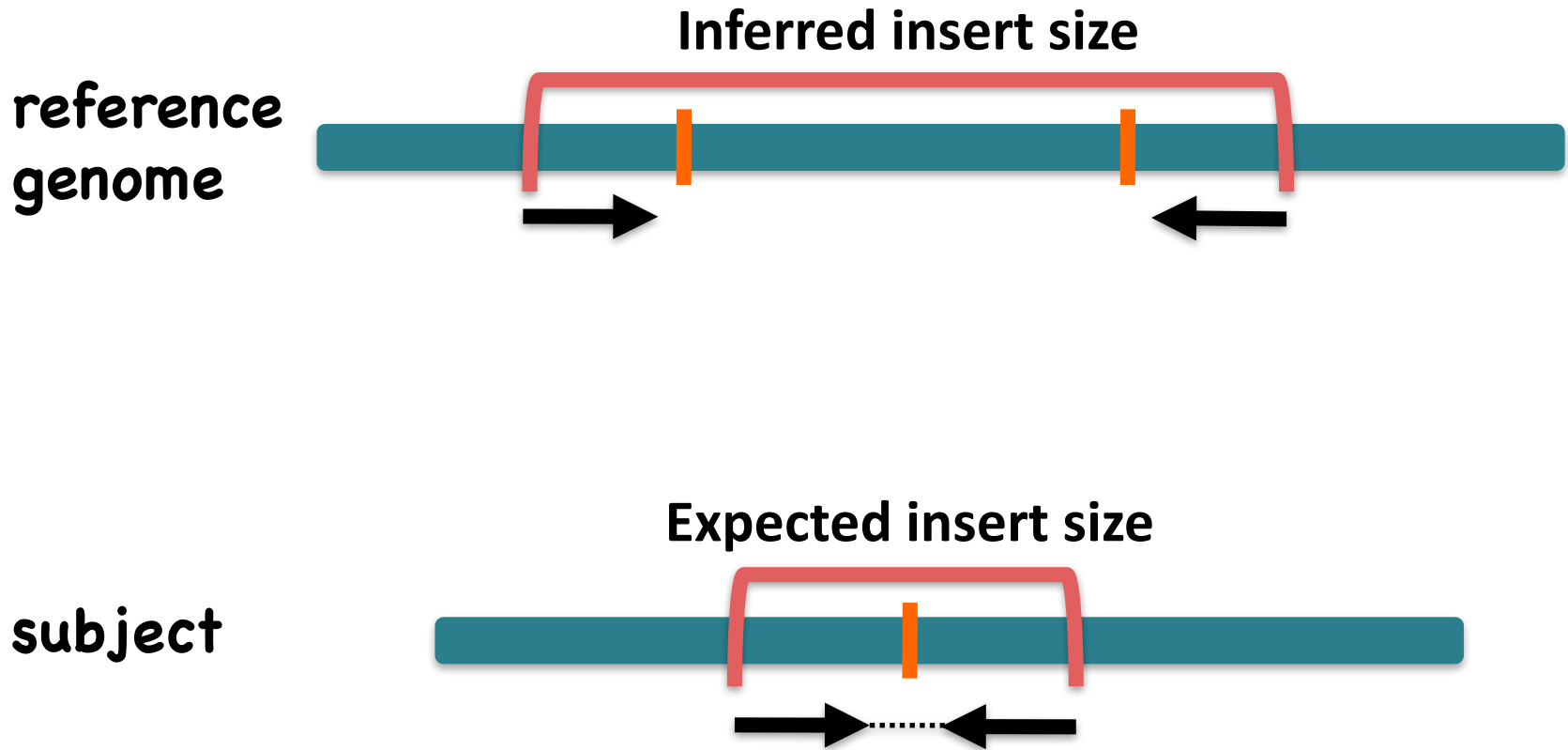
Deletion



Deletion



Deletion



Inferred insert size is greater than expected value

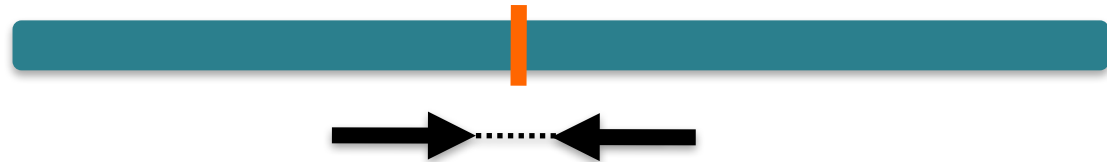
Deletion

Pairs with larger than expected insert size are colored red

**reference
genome**

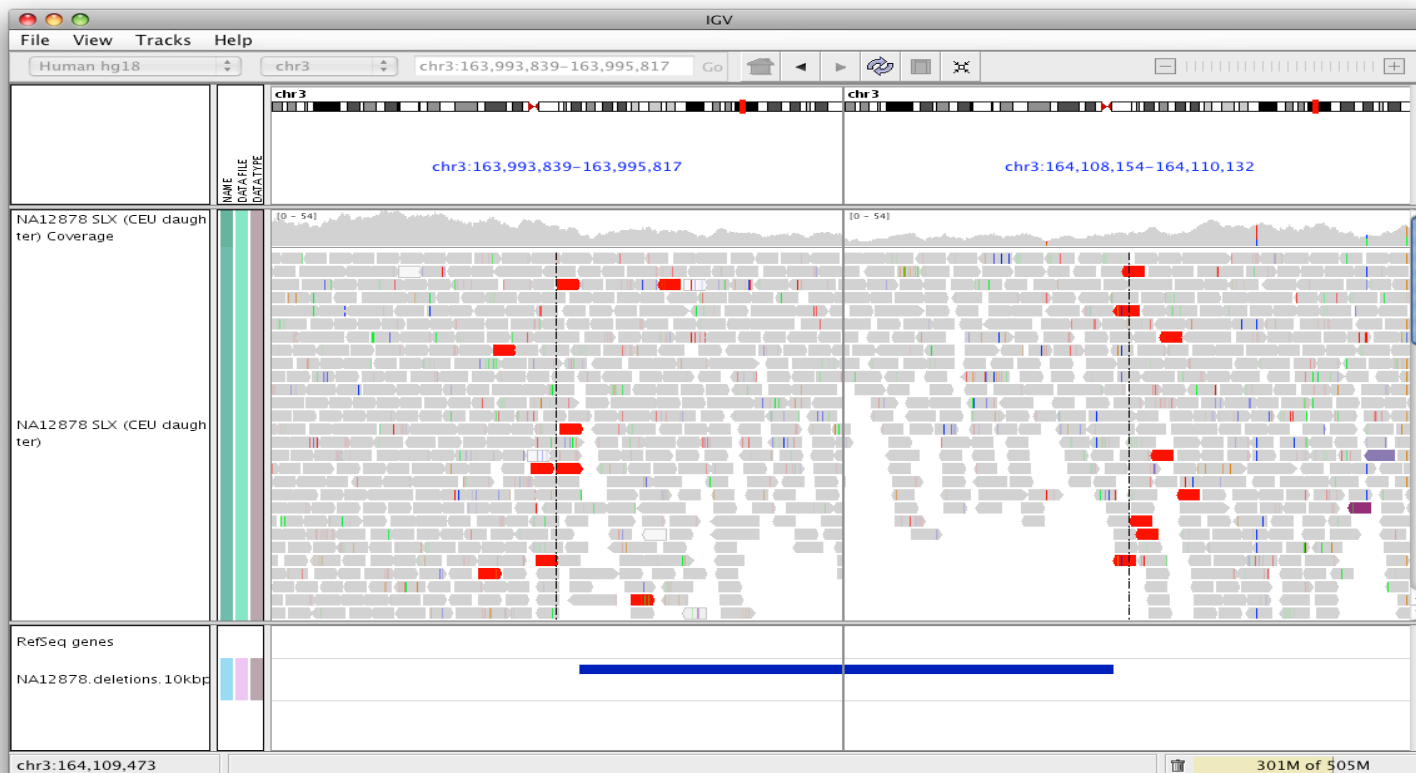


subject

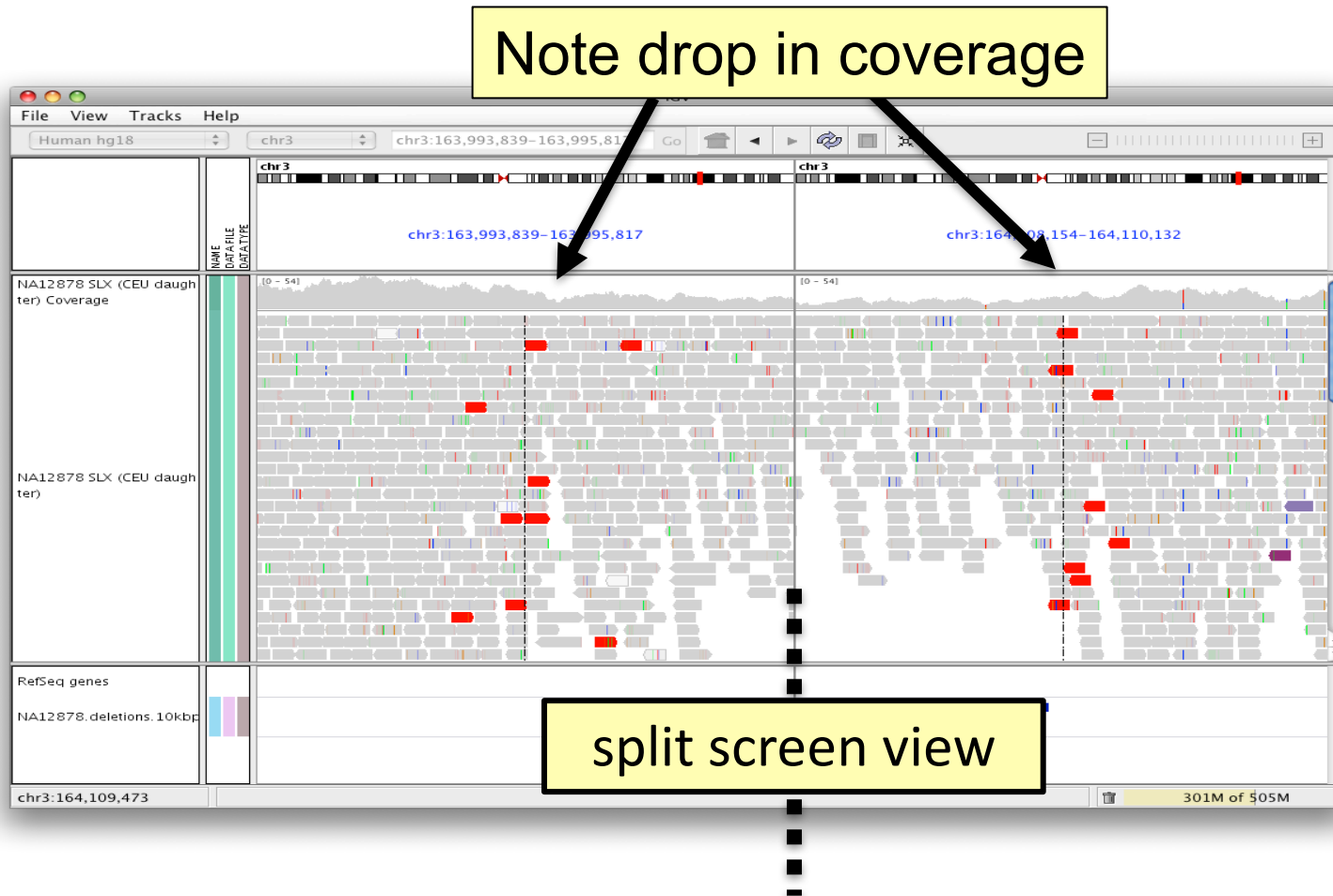


Deletion

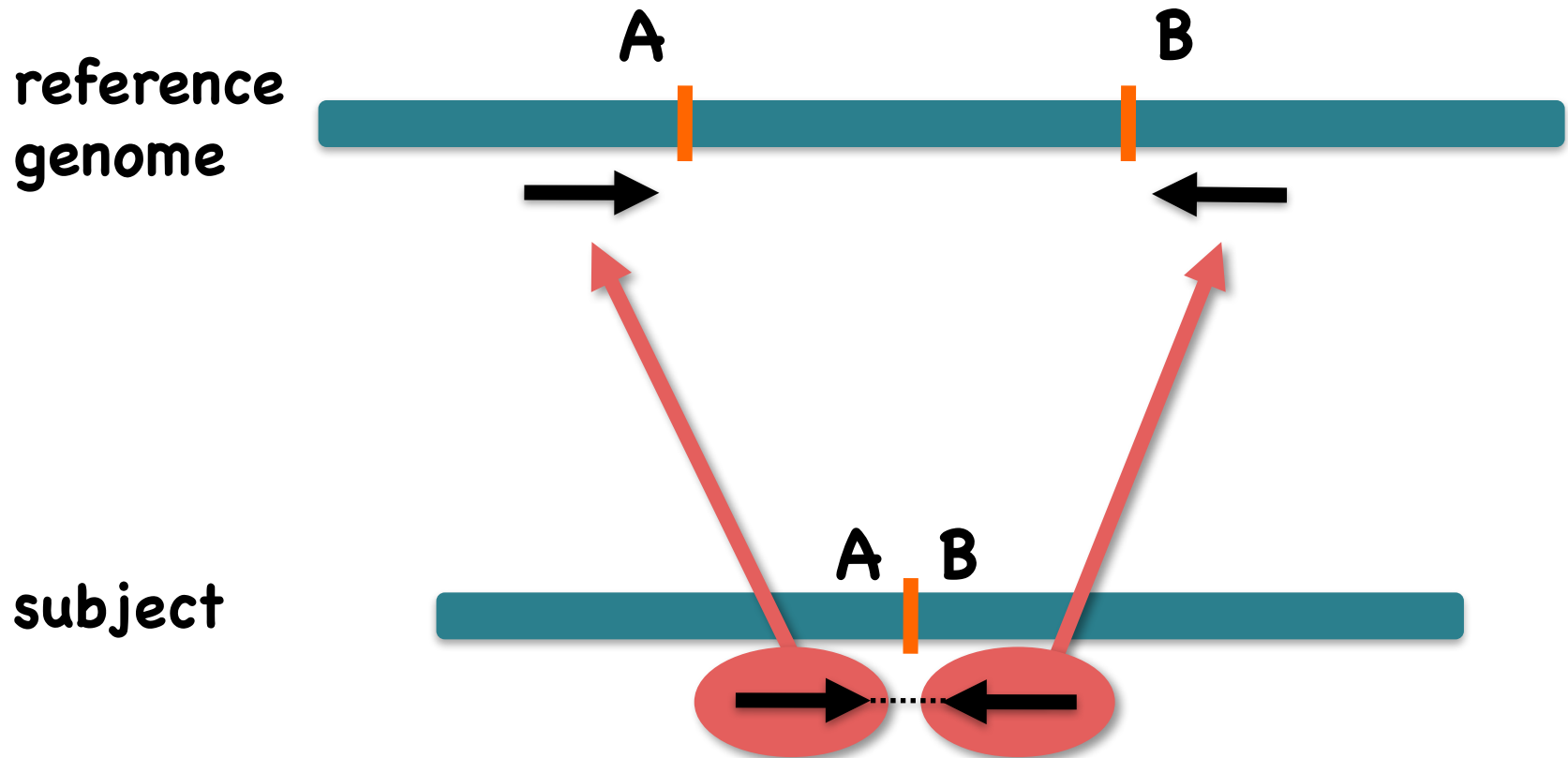
Pairs with larger than expected insert size are colored red



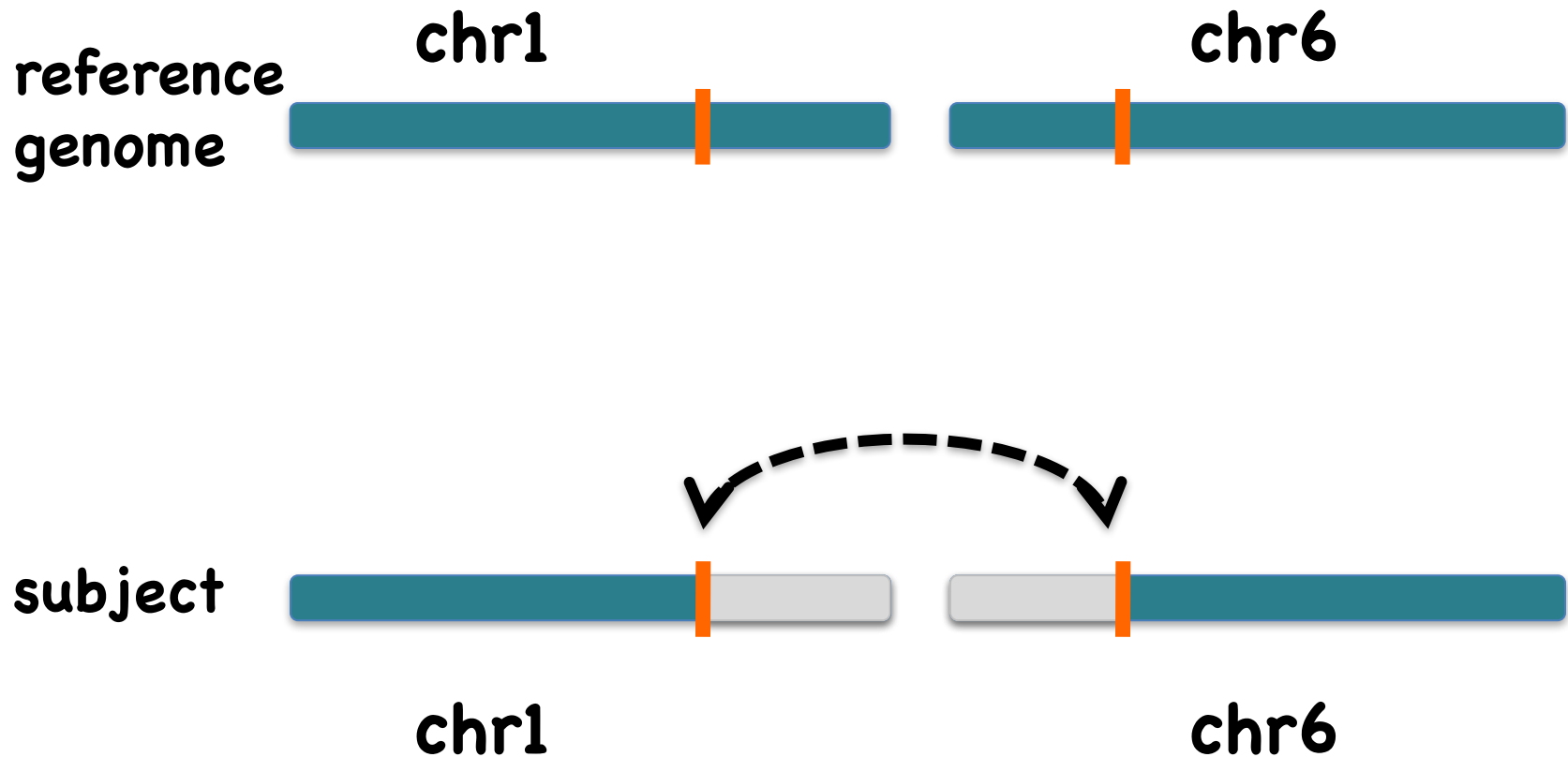
Deletion



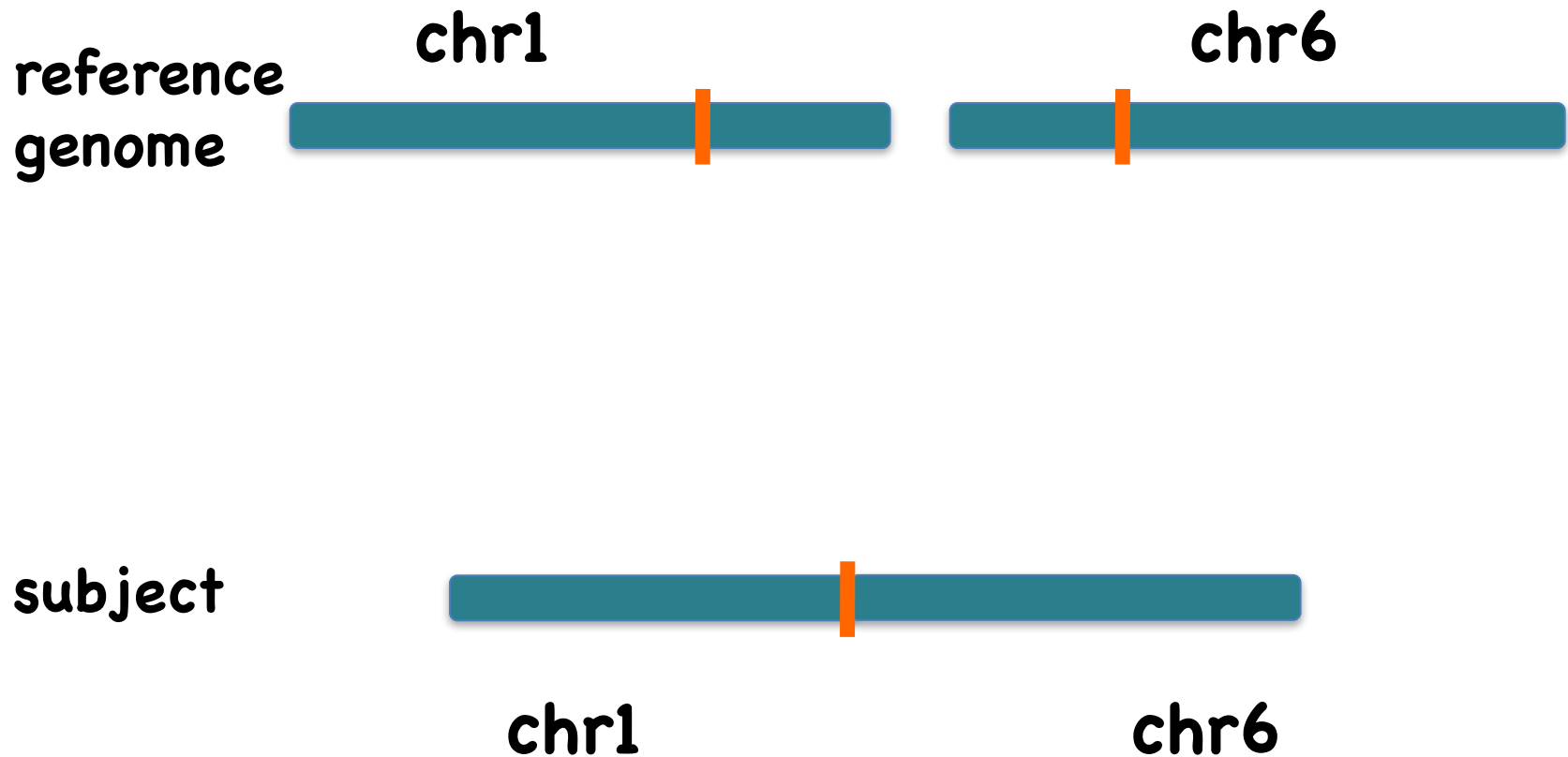
Deletion



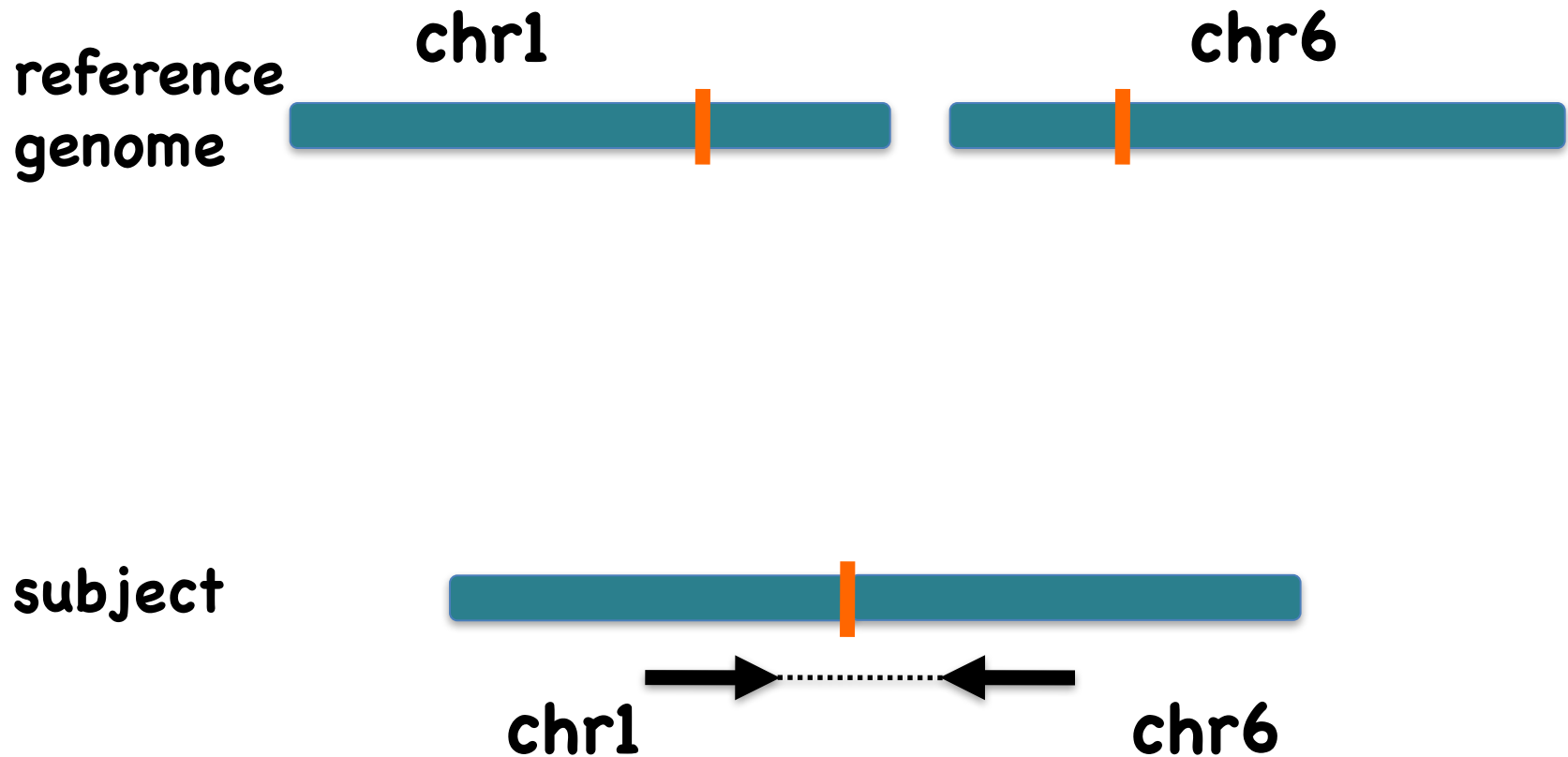
Inter-chromosomal fusion



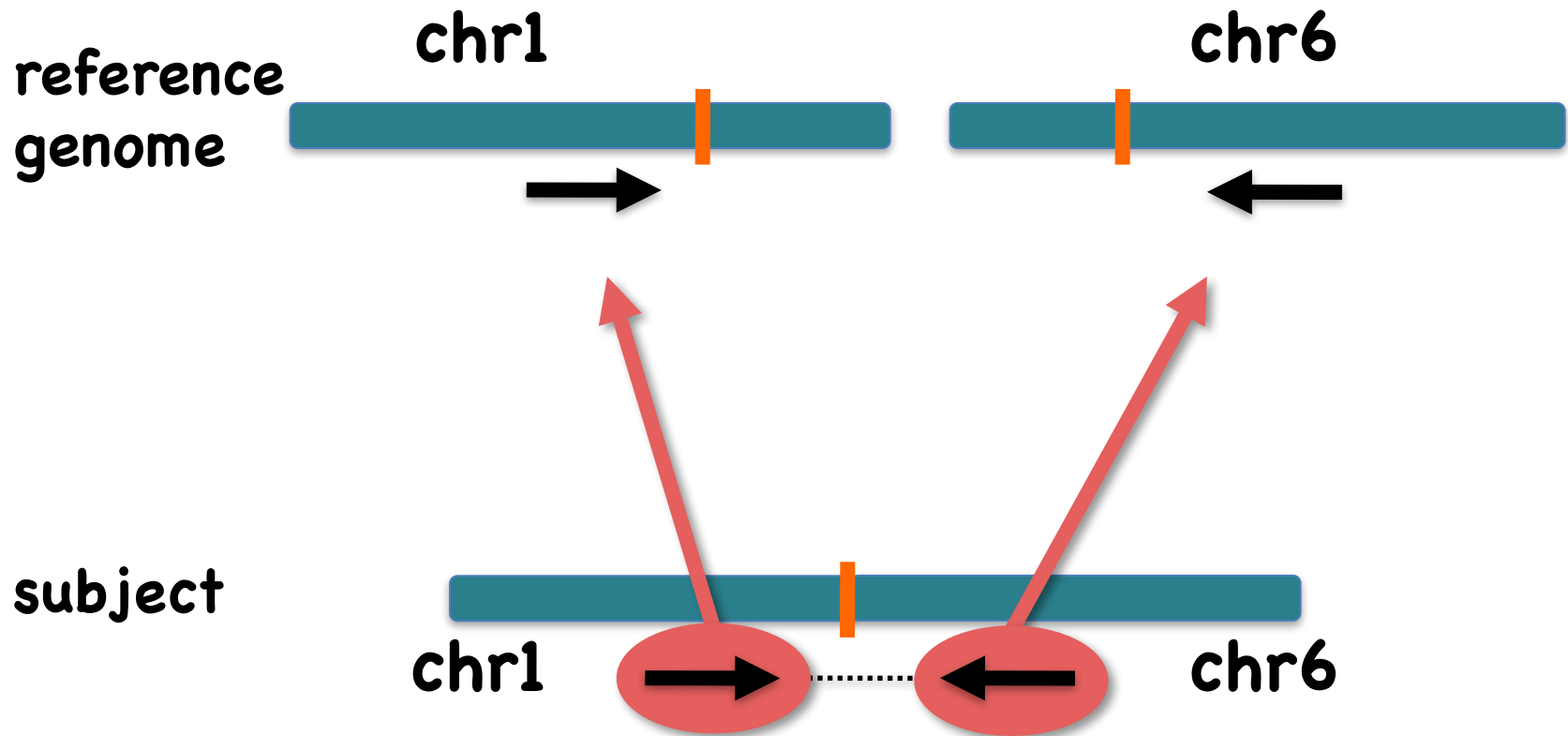
Inter-chromosomal fusion



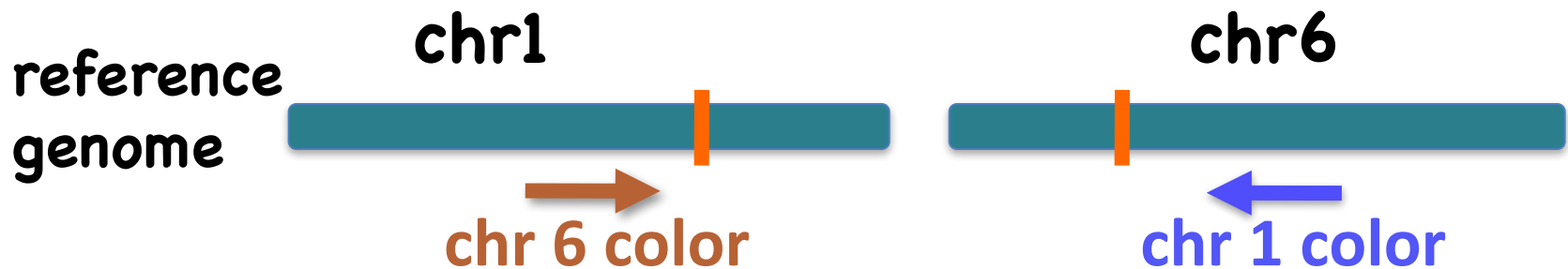
Inter-chromosomal fusion



Inter-chromosomal fusion



Inter-chromosomal fusion

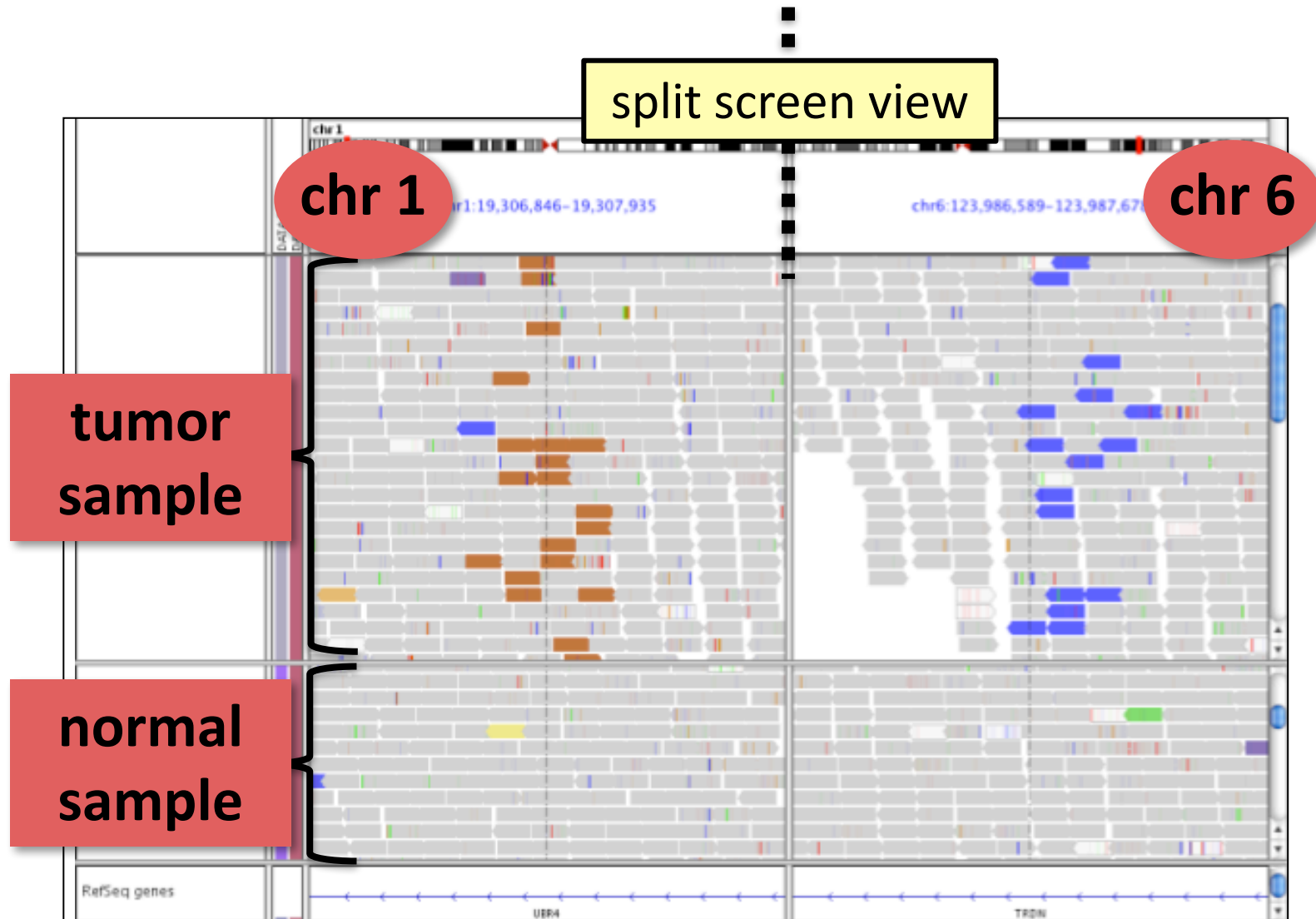


Each read is colored by the chromosome of its mate

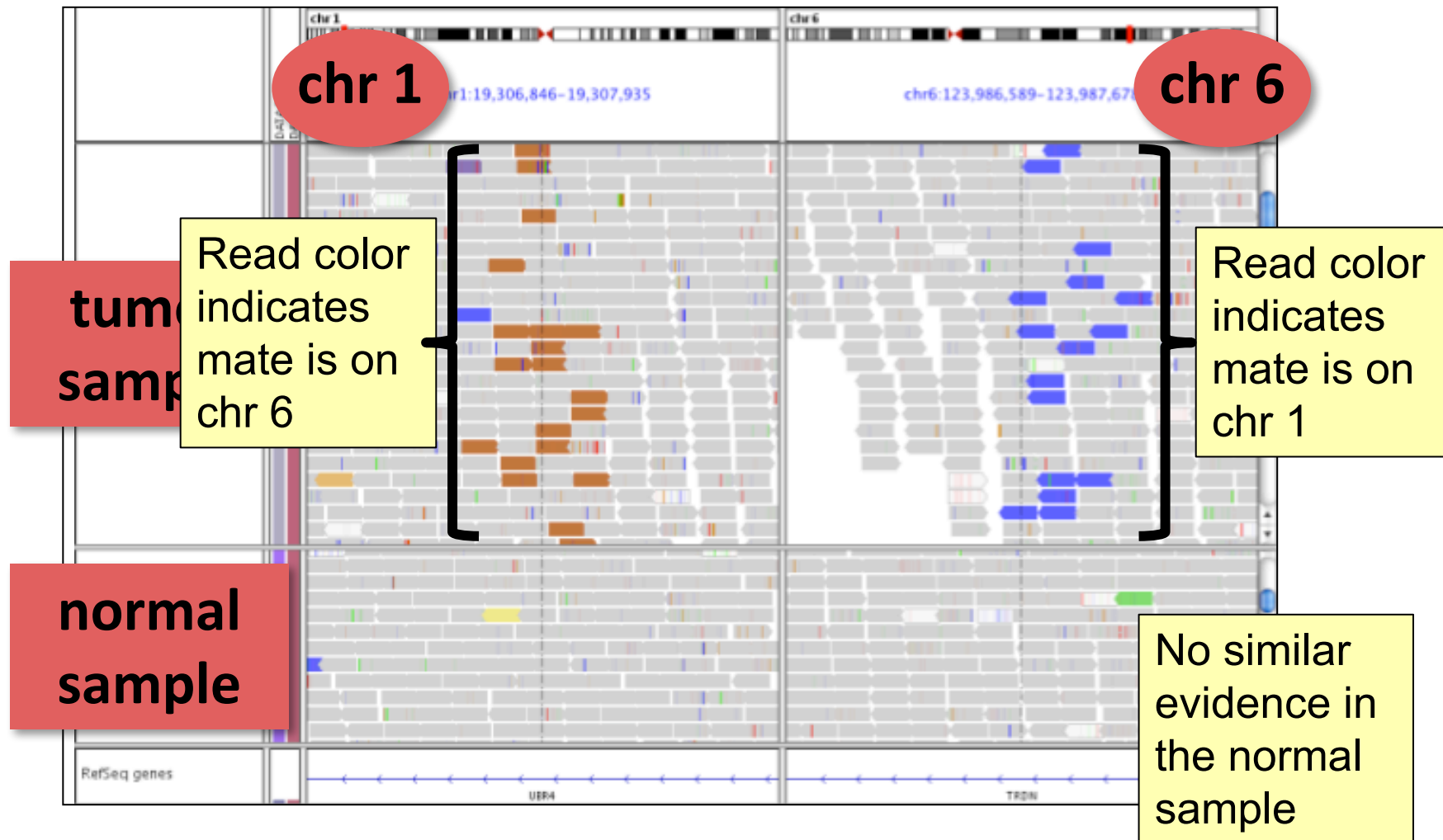
Chromosome colors:



Inter-chromosomal fusion



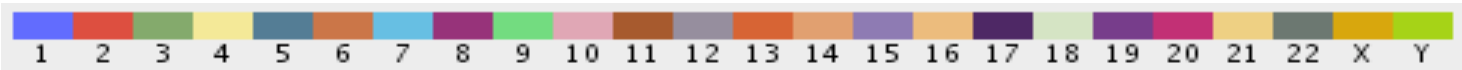
Inter-chromosomal fusion



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



Interpreting pair orientations

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

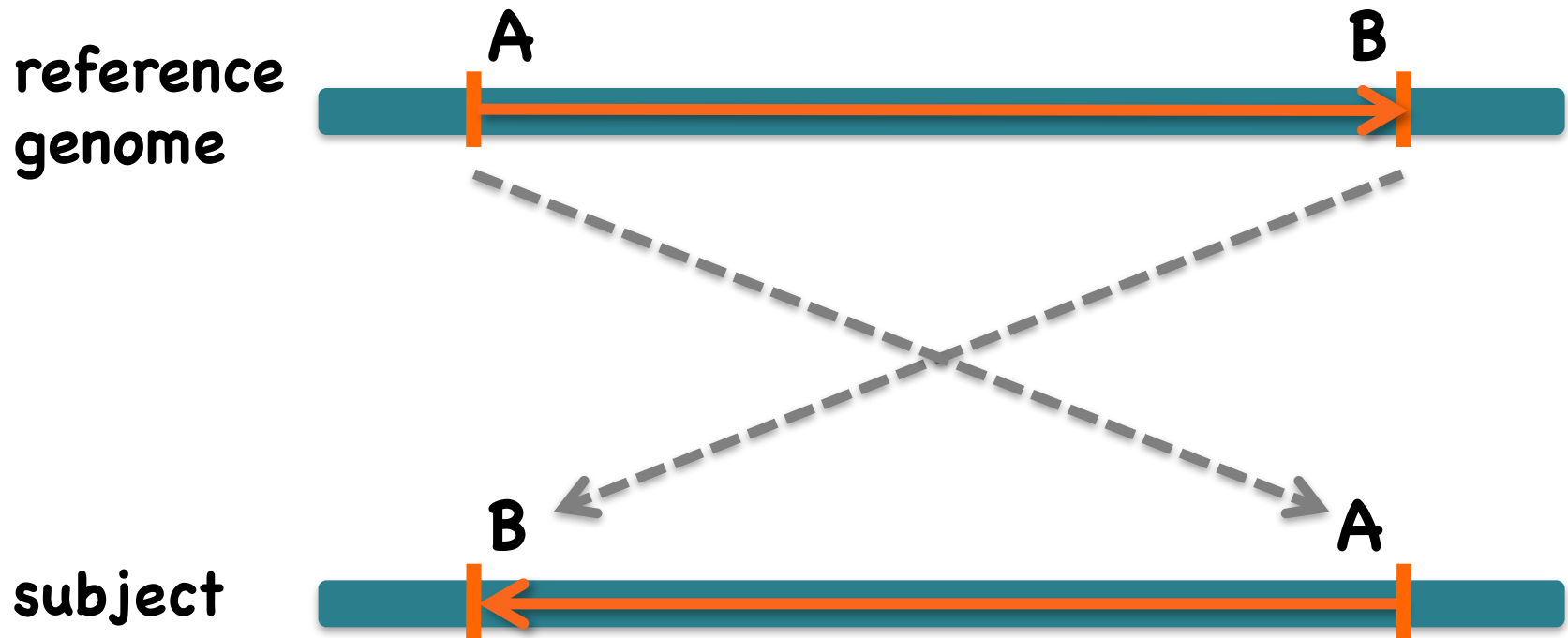
- Inversions
- Duplications
- Translocations

Interpreting pair orientations

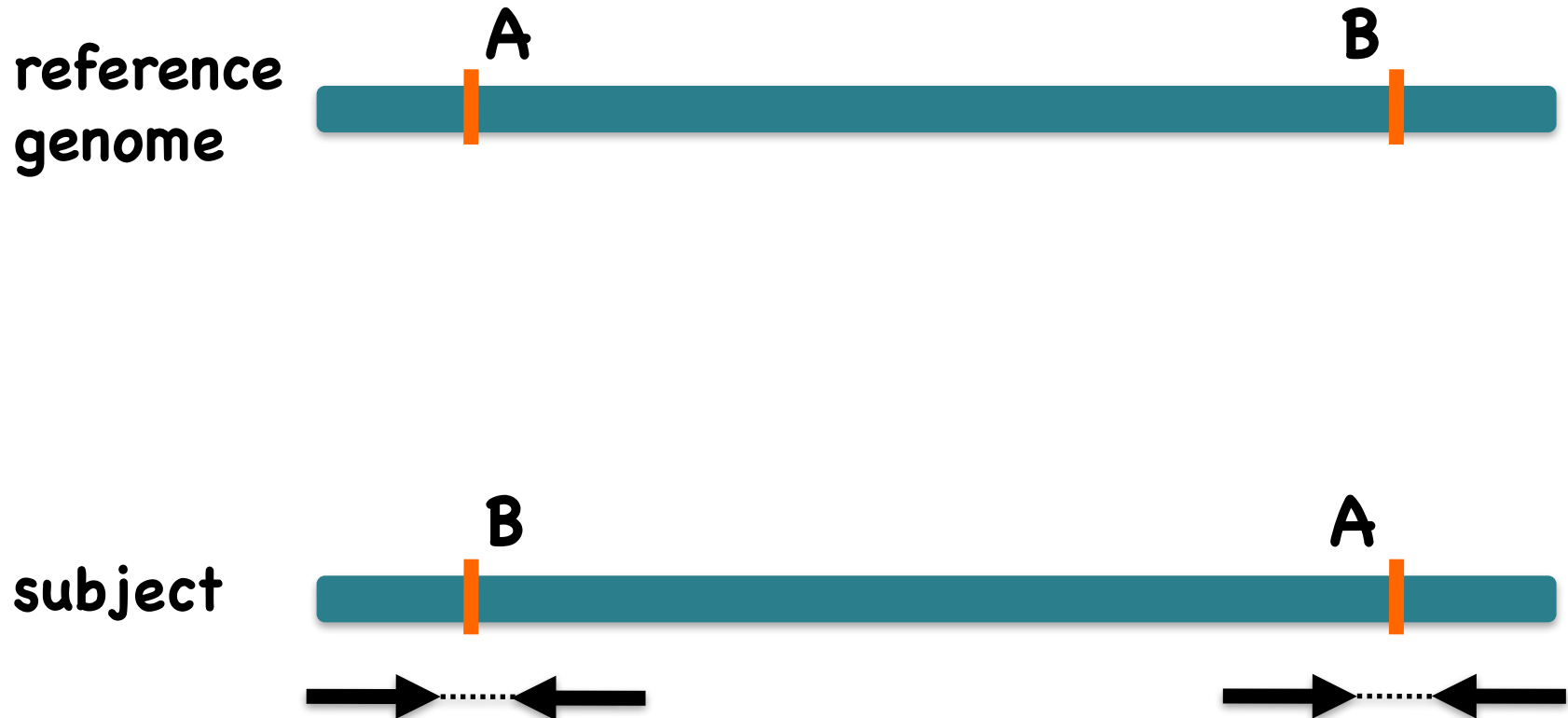
Orientation of paired reads can reveal evidence of structural events, including:

- Inversions
- Duplications
- Translocations

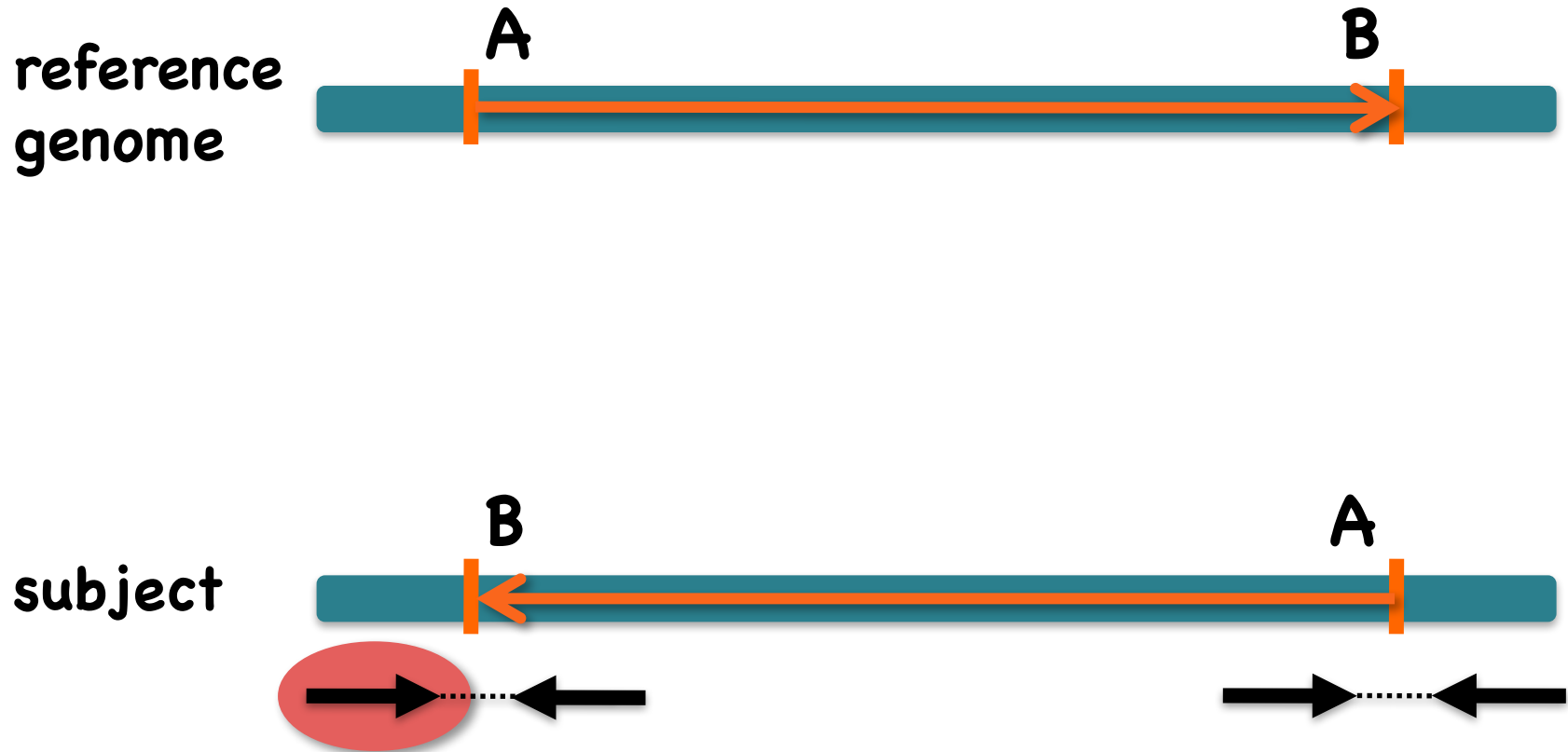
Inversion



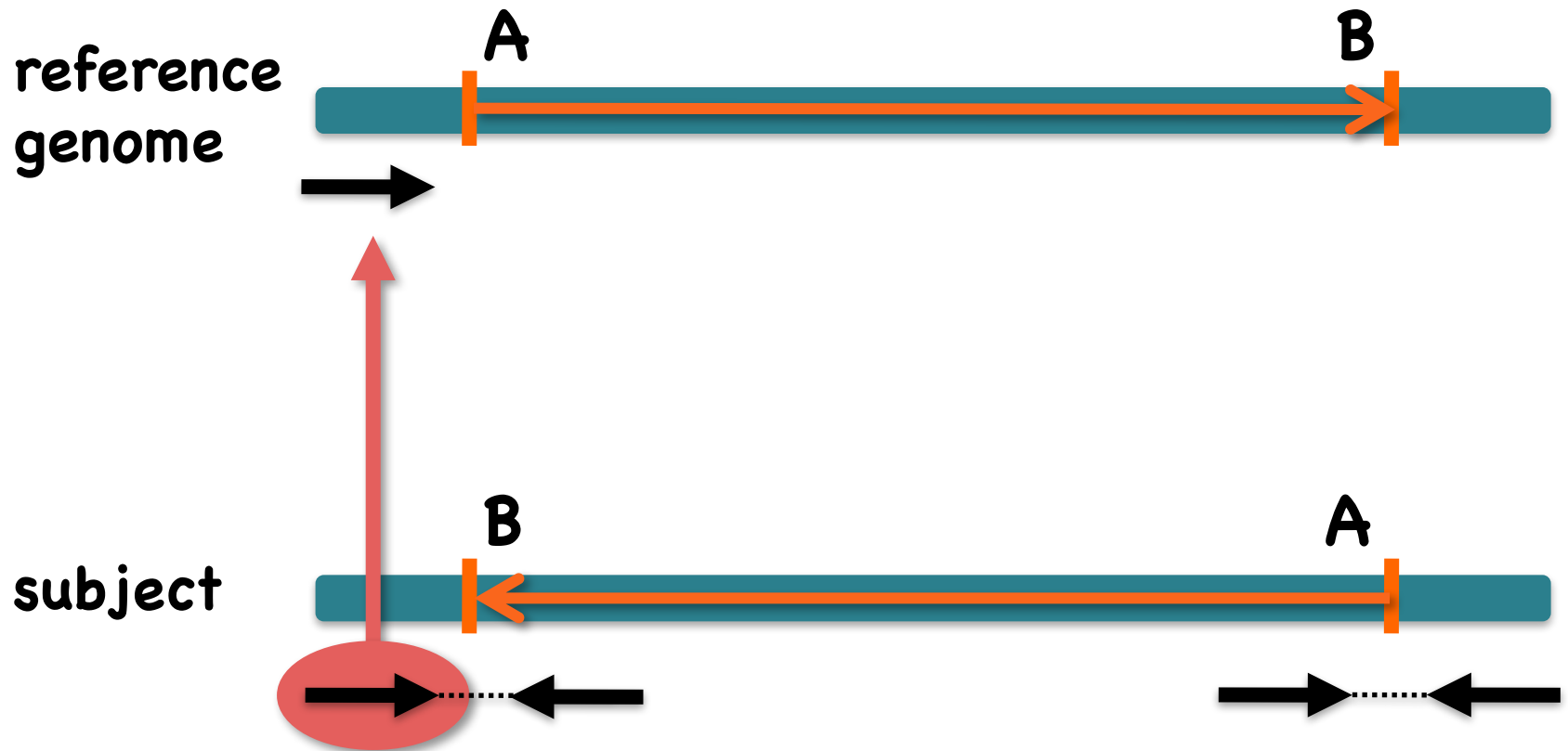
Inversion



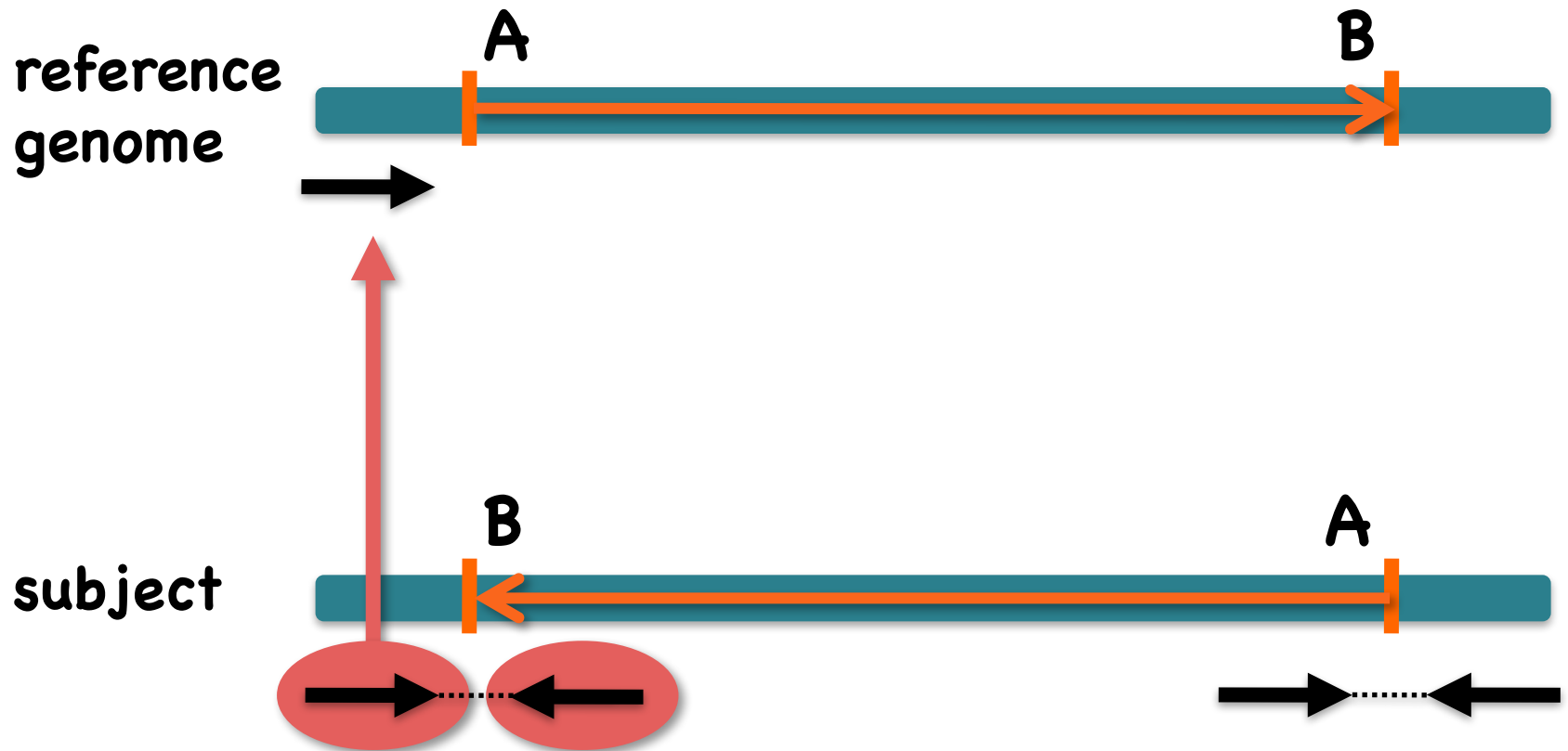
Inversion



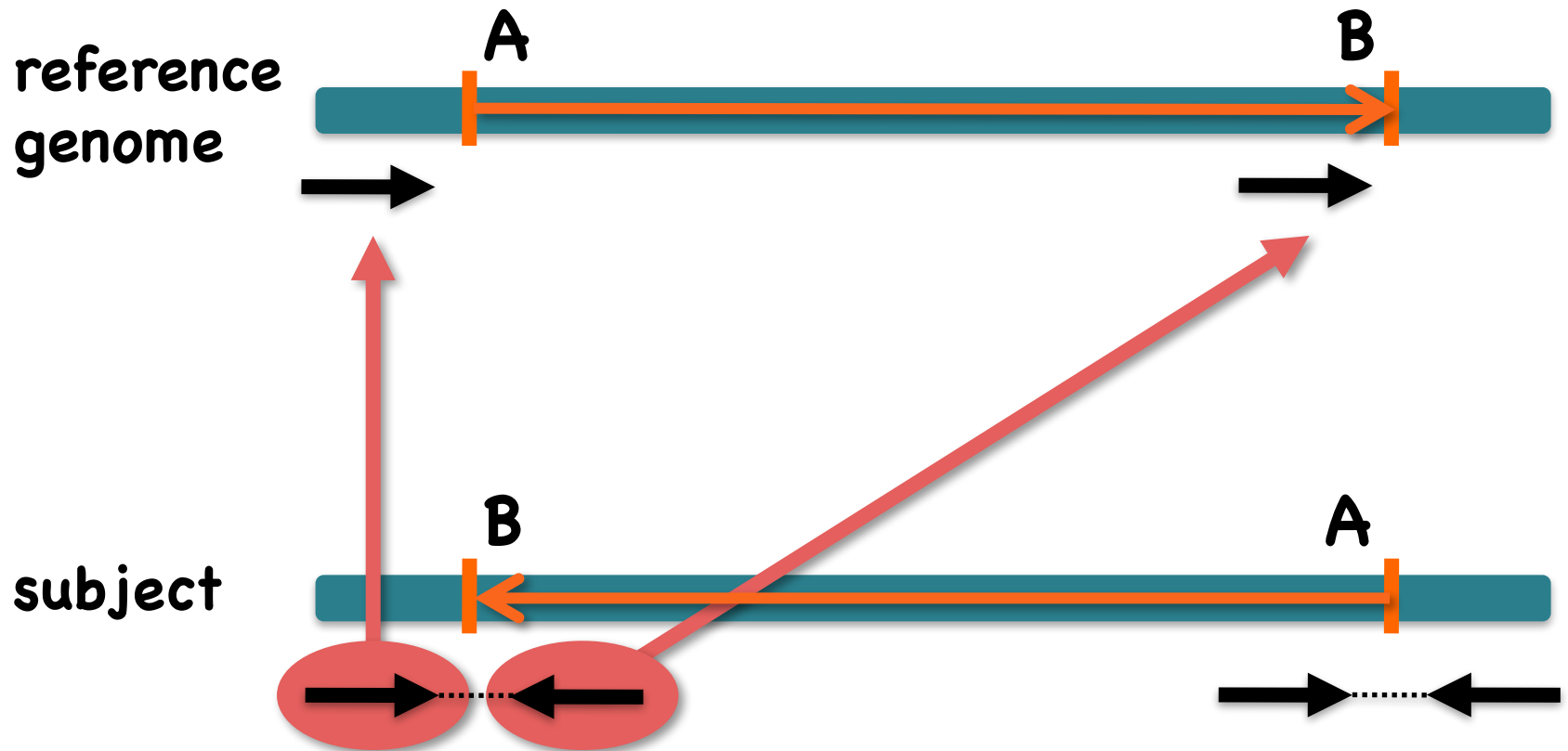
Inversion



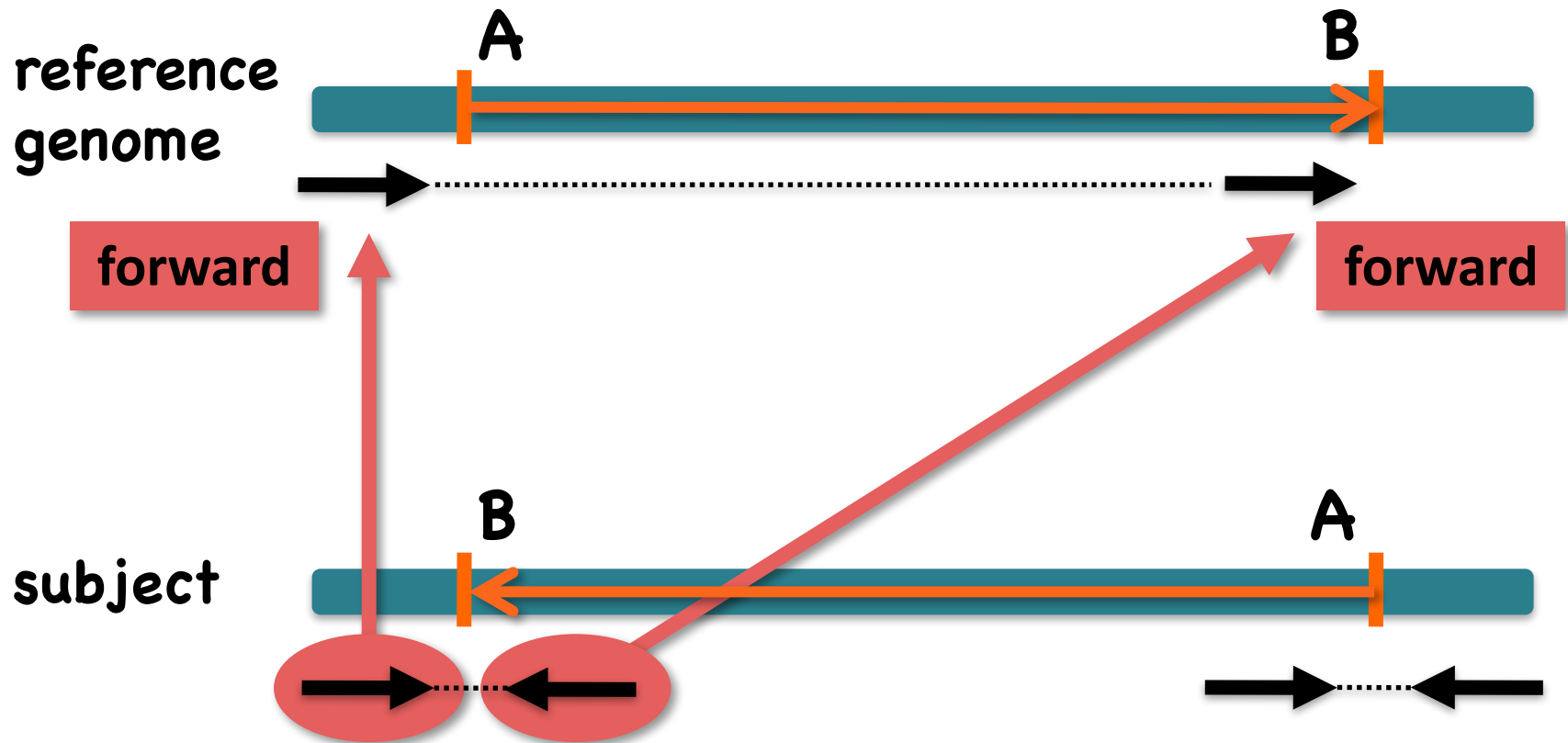
Inversion



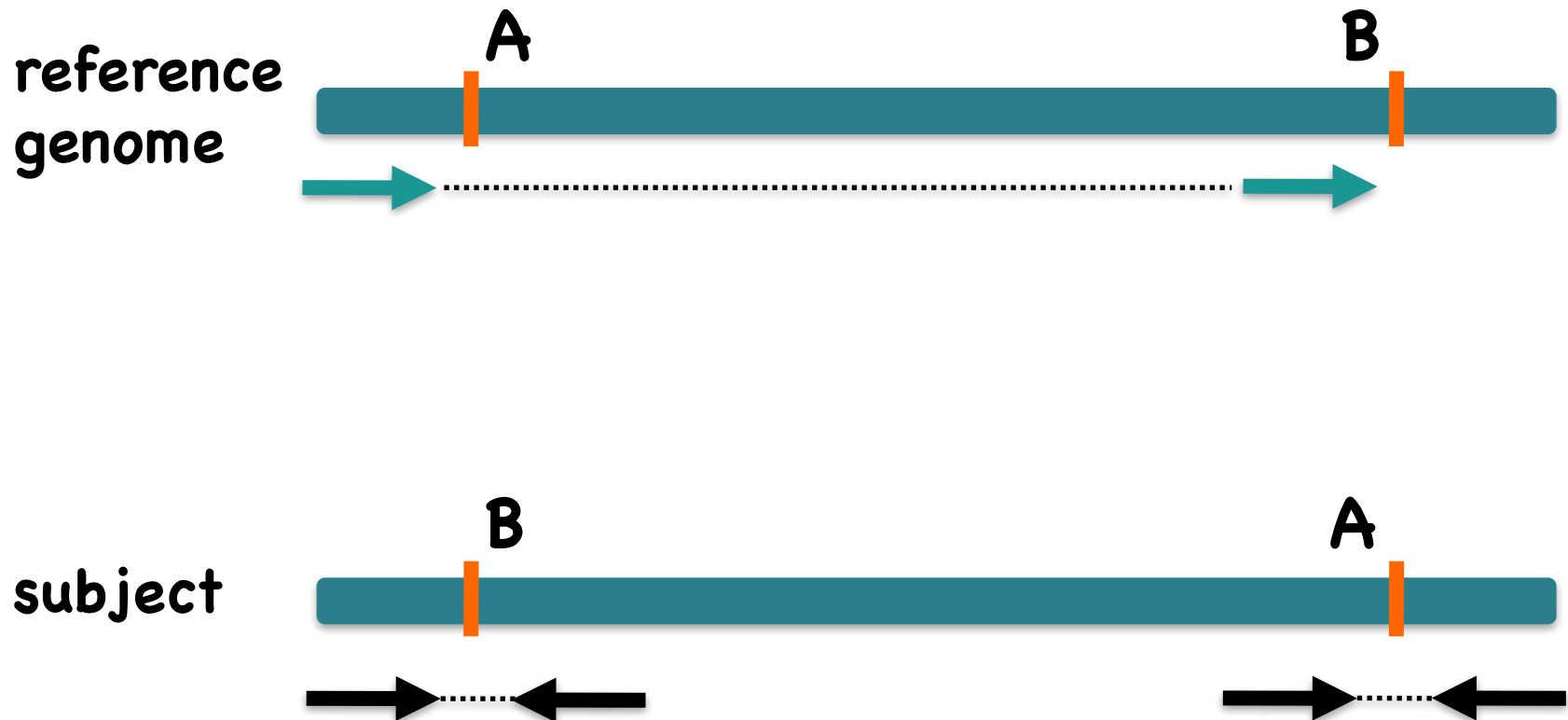
Inversion



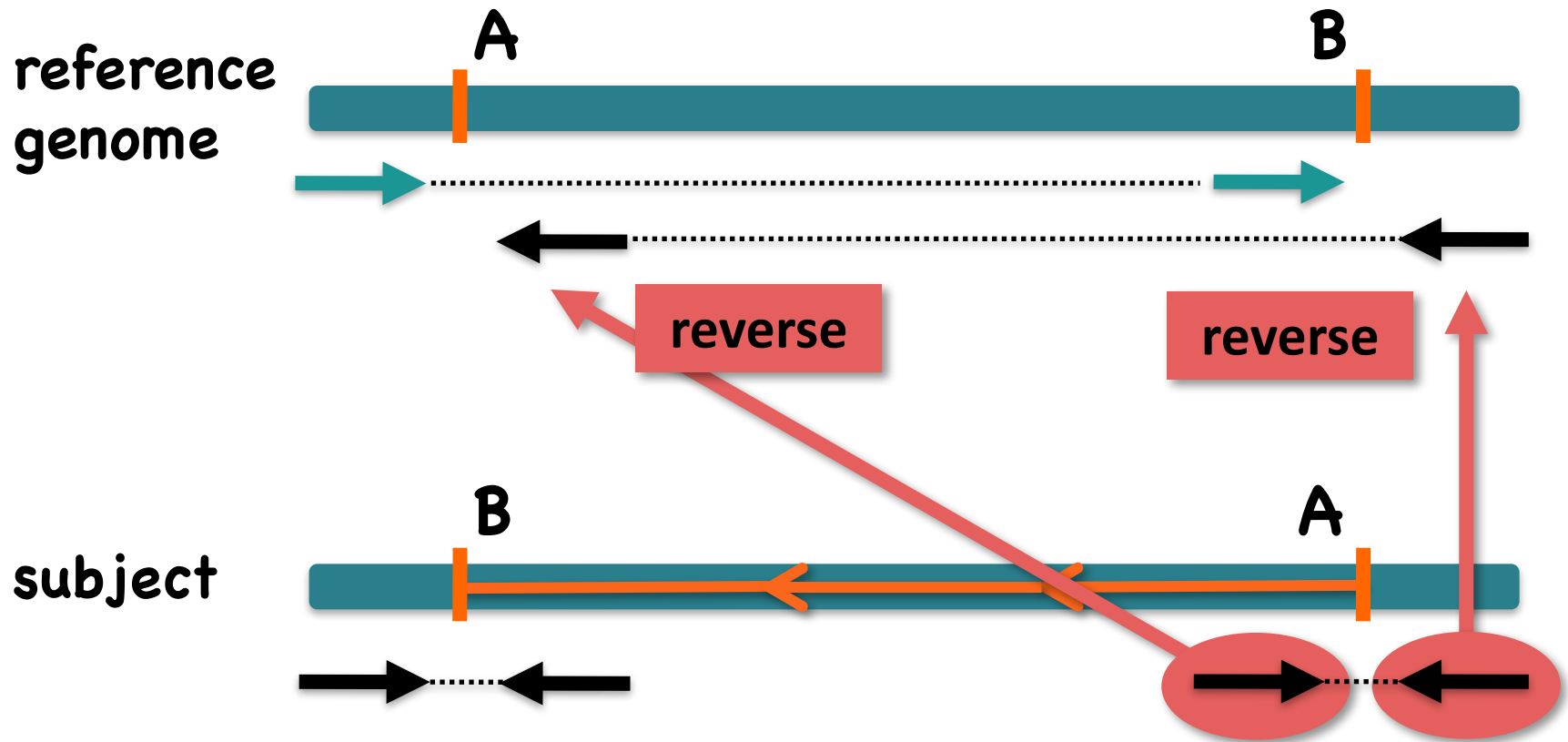
Inversion



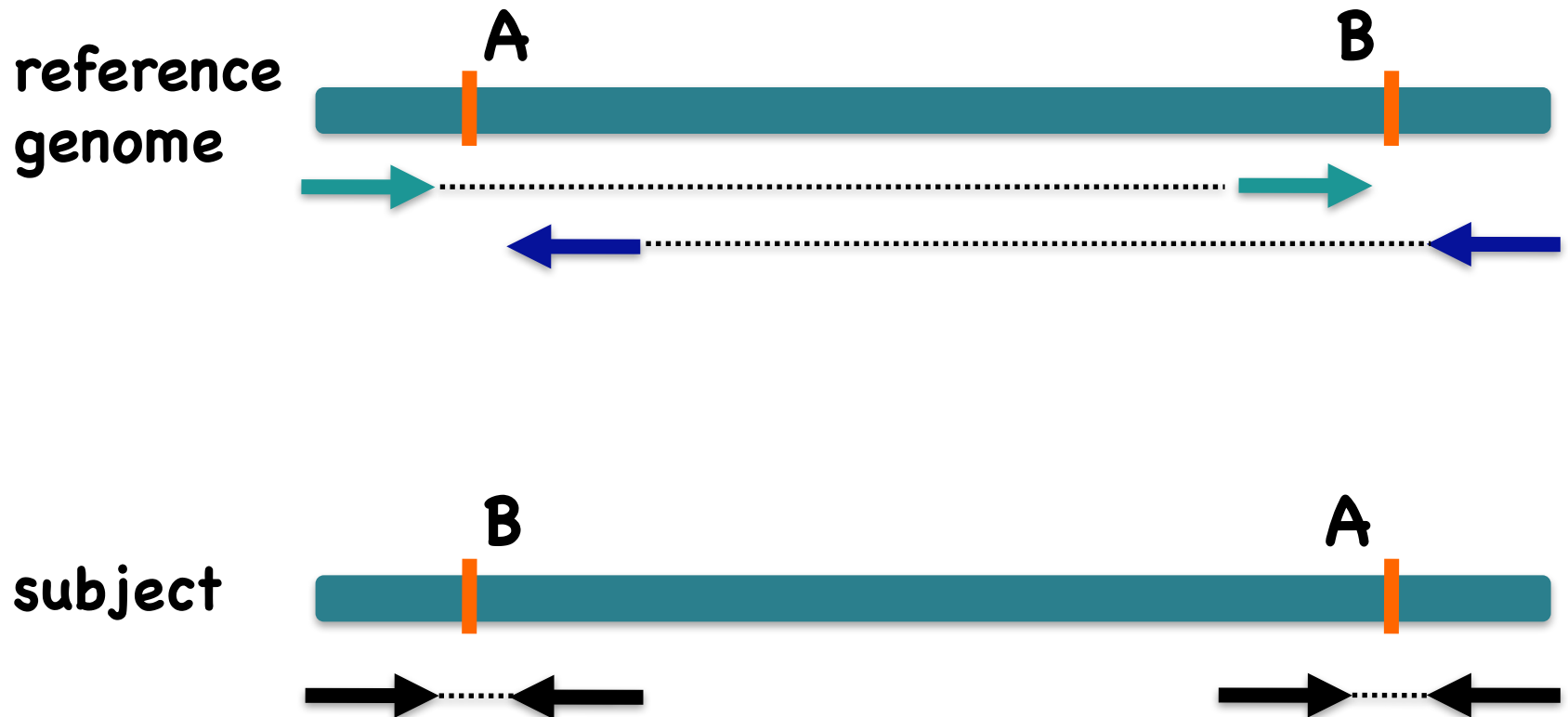
Inversion



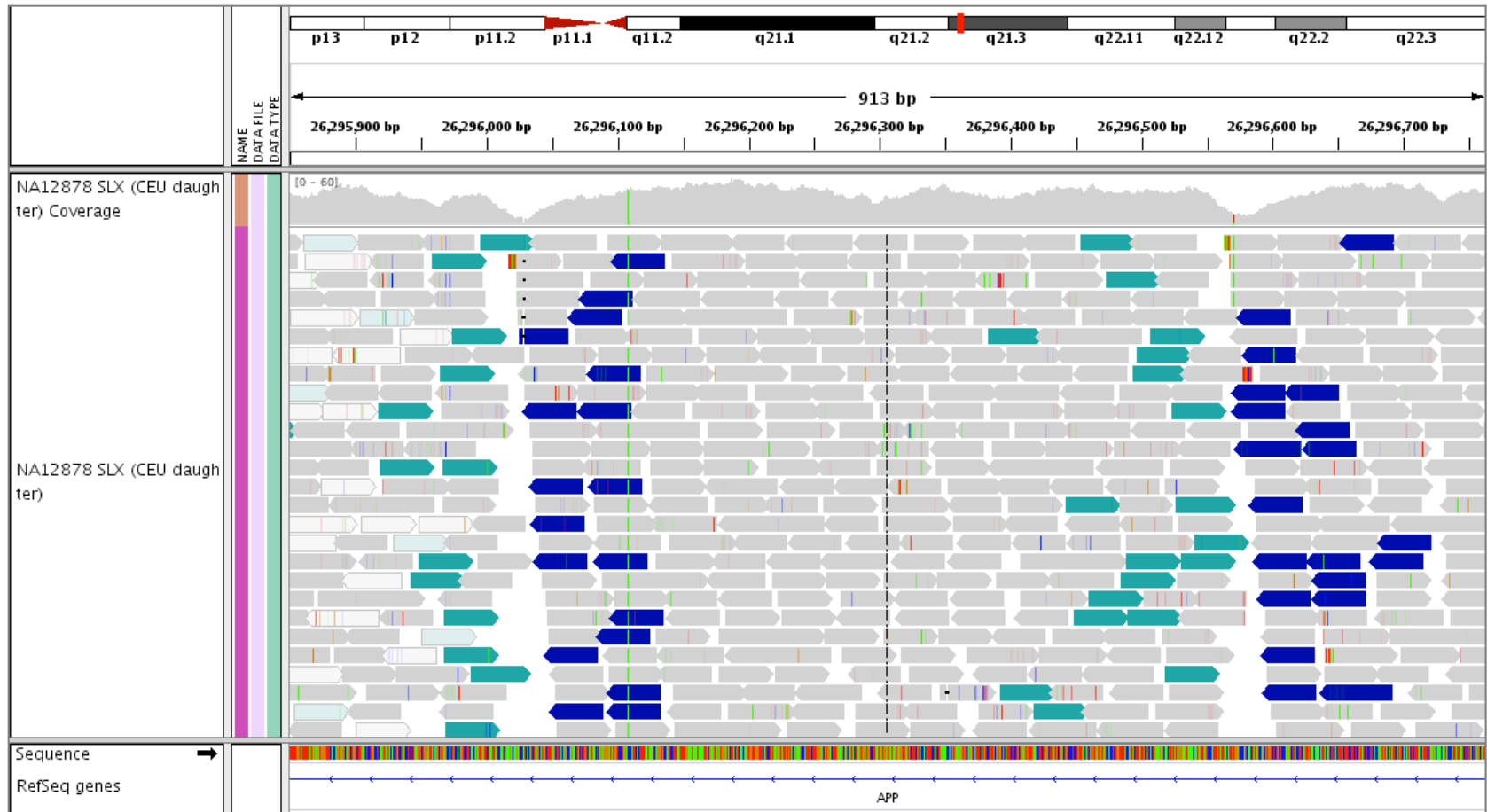
Inversion



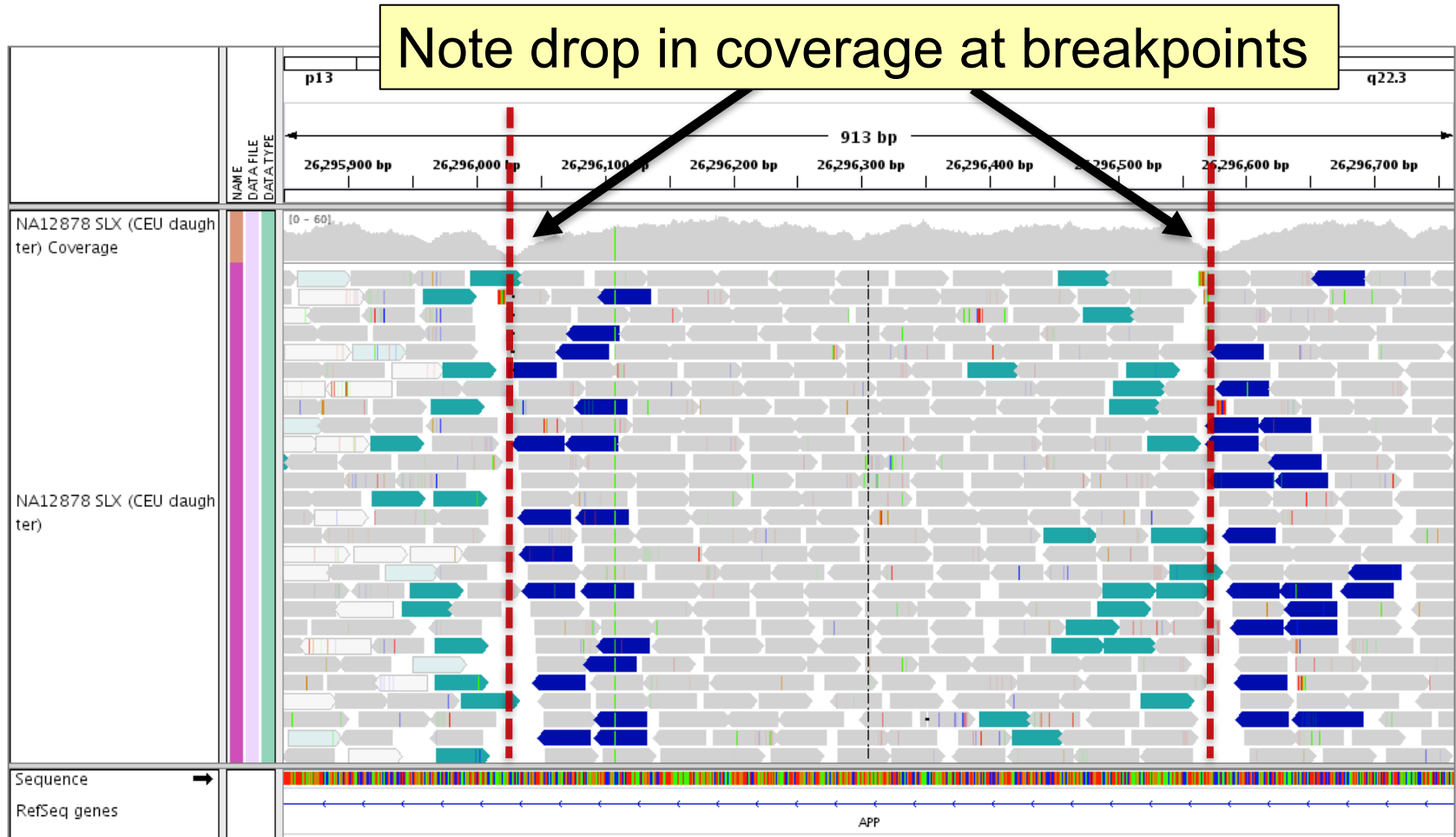
Inversion



Inversion



Inversion



Viewing structural events

Hands-on exercise

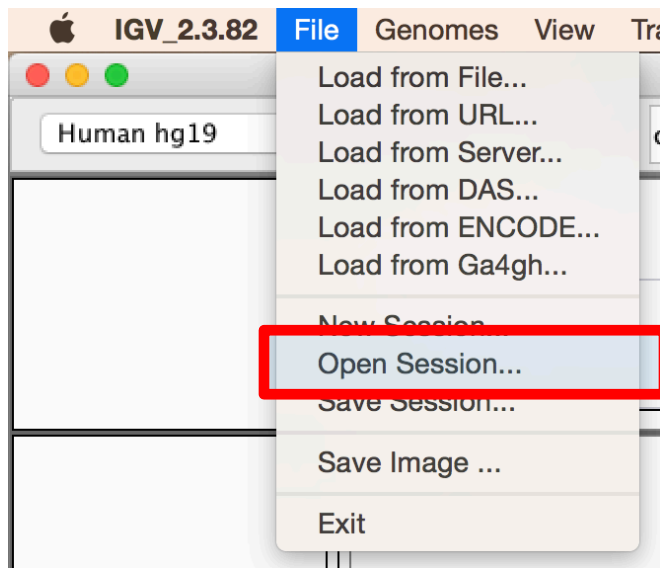
- Use color and viewing options to examine an inversion
- Data from 1000 Genomes

Follow along

Viewing structural events

Hands-on exercise

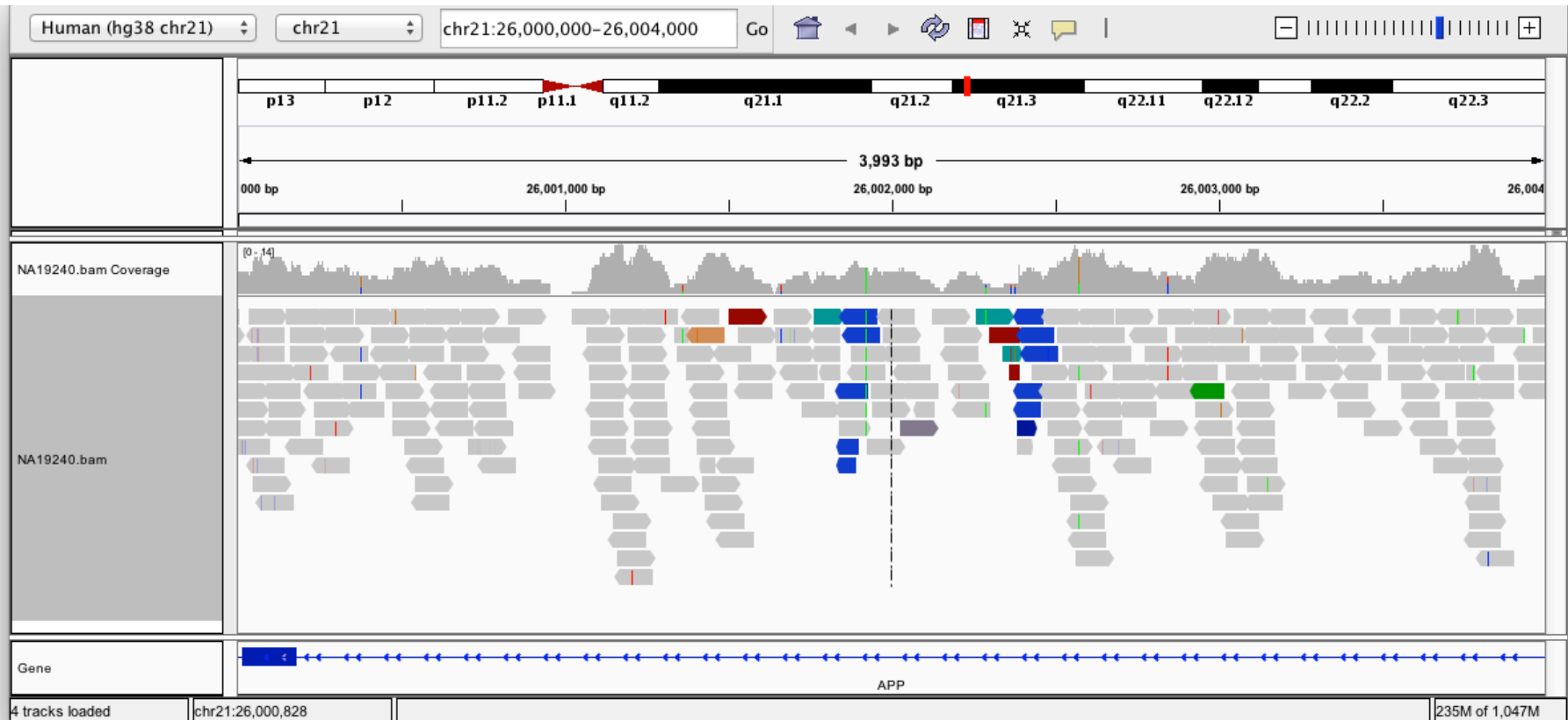
Click **File > Open Session**



Navigate to the folder IGV workshop folder / **Data / sv**s and select file **svs_session.xml**

Viewing structural events

Hands-on exercise



Viewing structural events

Hands-on exercise

Right-click on the alignments and select
Color alignments by > pair orientation

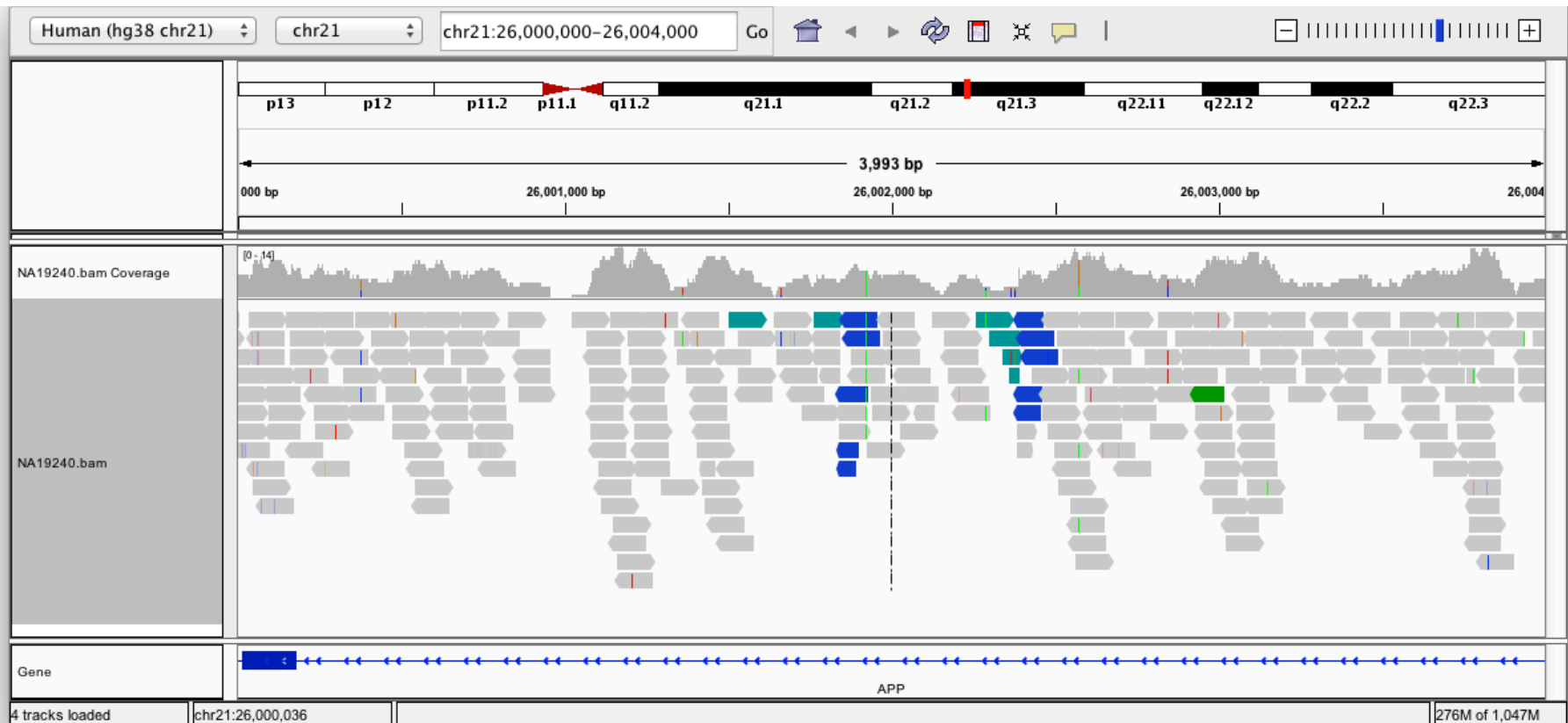
The screenshot displays a genomic viewer interface with a right-click context menu open over a track of alignments. The menu options are as follows:

- Rename Track...
- Copy read details to clipboard
- Group alignments by
- Sort alignments by
- Color alignments by** (highlighted)
 - no color
 - insert size** (checked)
 - pair orientation** (highlighted)
 - 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

The background interface shows a genomic track with a coverage plot at the top, a track of alignments in the middle, and a gene track at the bottom. The alignment track is labeled with coordinates p13, p12, p11.2, p11.1, q11.2, q21.3, q22.11, q22.12, q22.2, and q22.3. The gene track shows a gene structure with exons and introns. The status bar at the bottom indicates 4 tracks loaded, chr21:26,000,653, and 258M of 1,047M.

Viewing structural events

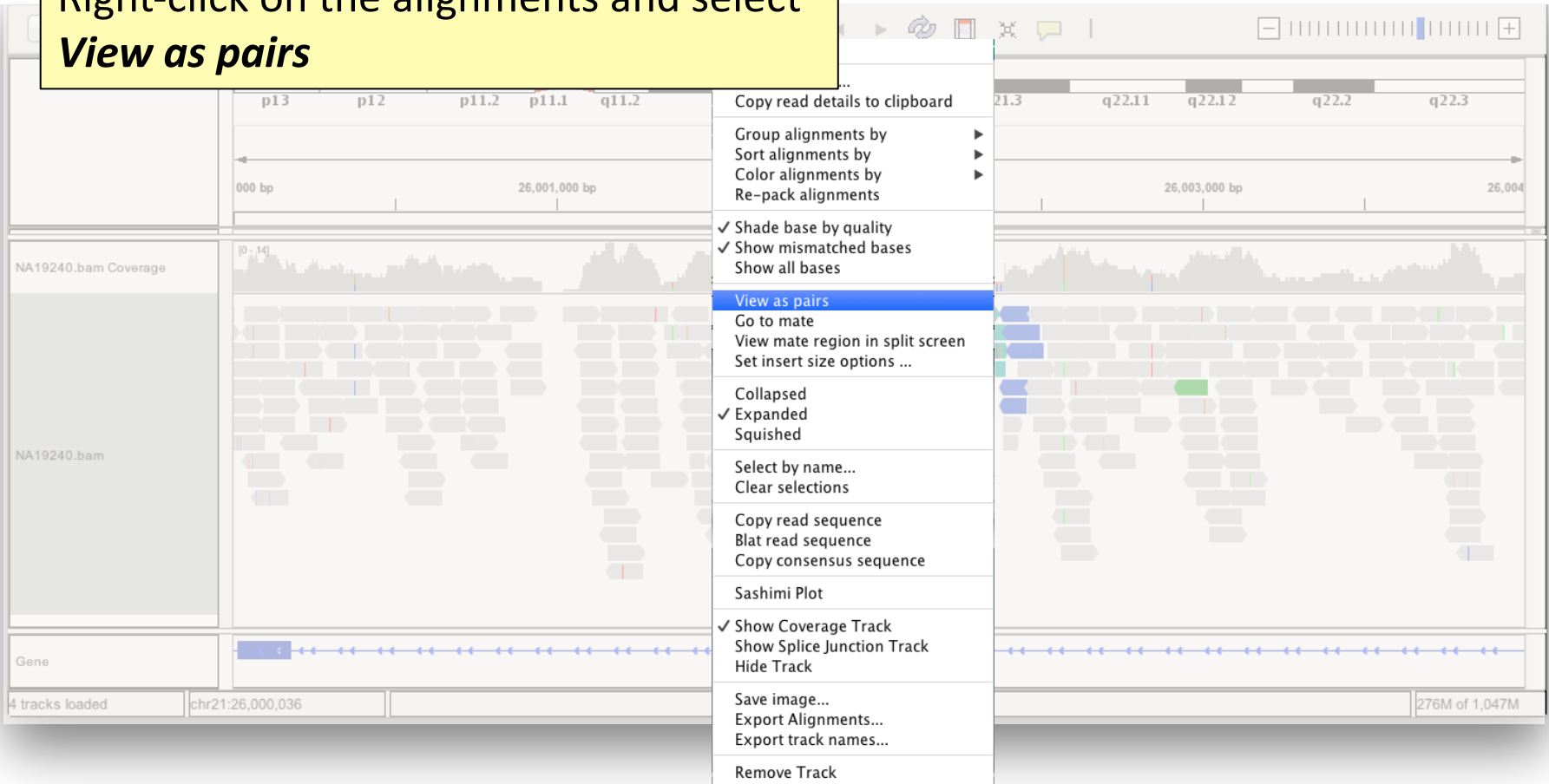
Hands-on exercise



Viewing structural events

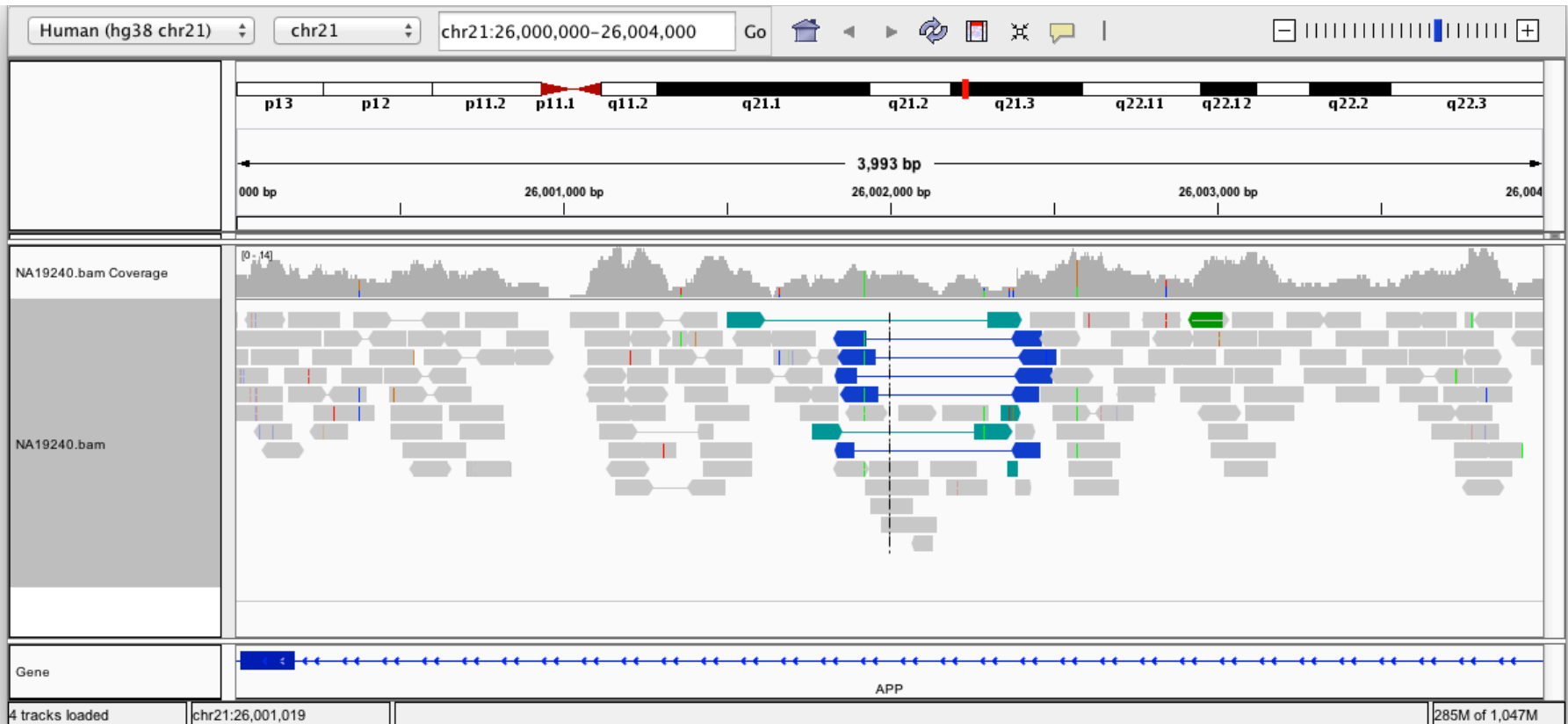
Hands-on exercise

Right-click on the alignments and select
View as pairs



Viewing structural events

Hands-on exercise



Viewing Structural Events



Availability

Features available in **IGV 3.0 Beta**

See **Other Versions** on the IGV Downloads page

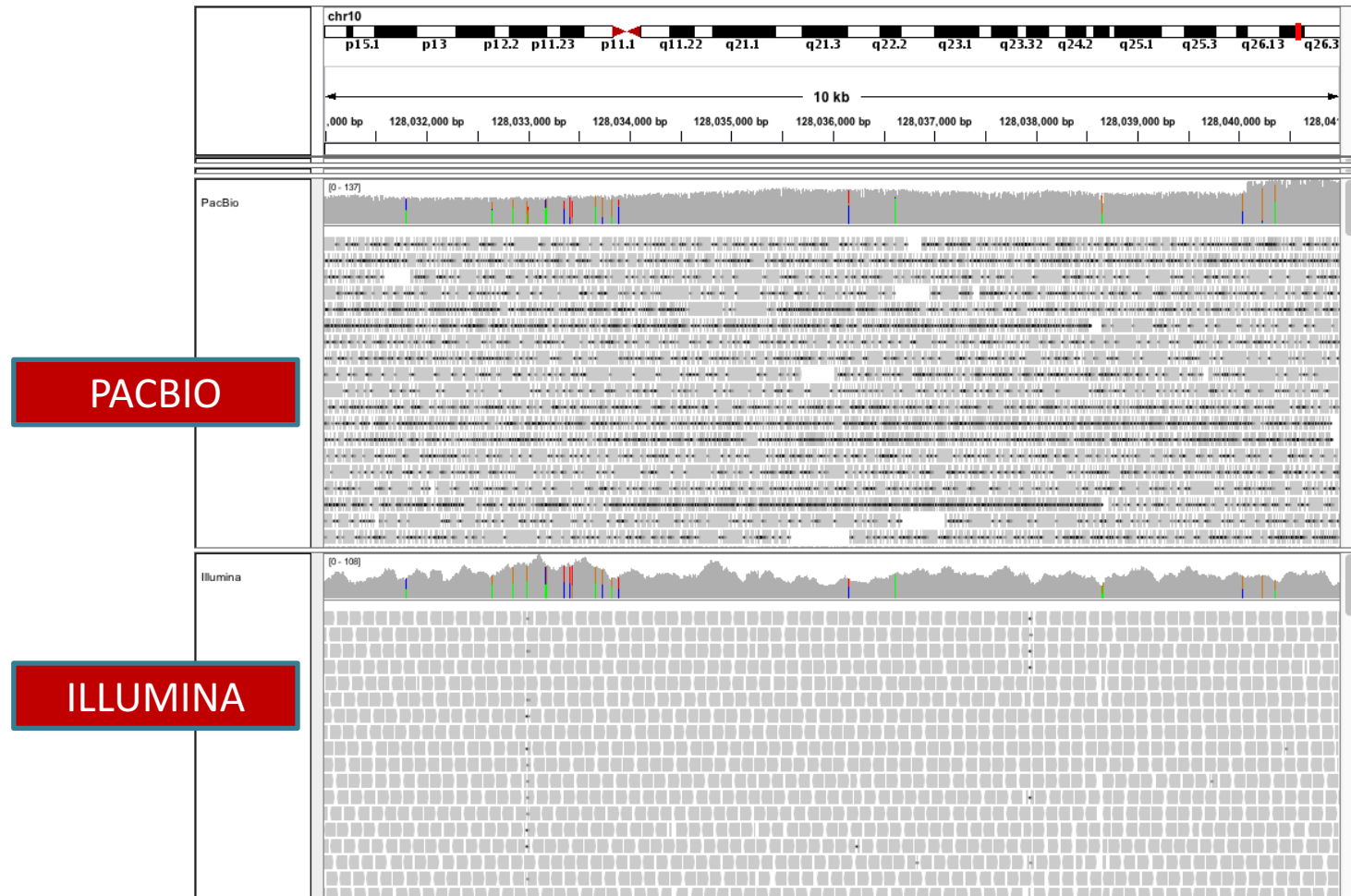
Third Gen Sequencing

- Long reads:
~10,000 bp up to ~100,00 bp
- High raw error rate in individual reads
(high consensus accuracy with sufficient coverage)
- Platforms:
 - PacBio, Oxford Nanopore

IGV Enhancements

- Filtering raw errors to reveal consensus variants
- Extended support for split alignments
 - Linking split reads
 - Support for “SA” tag

Filtering raw errors – small indels

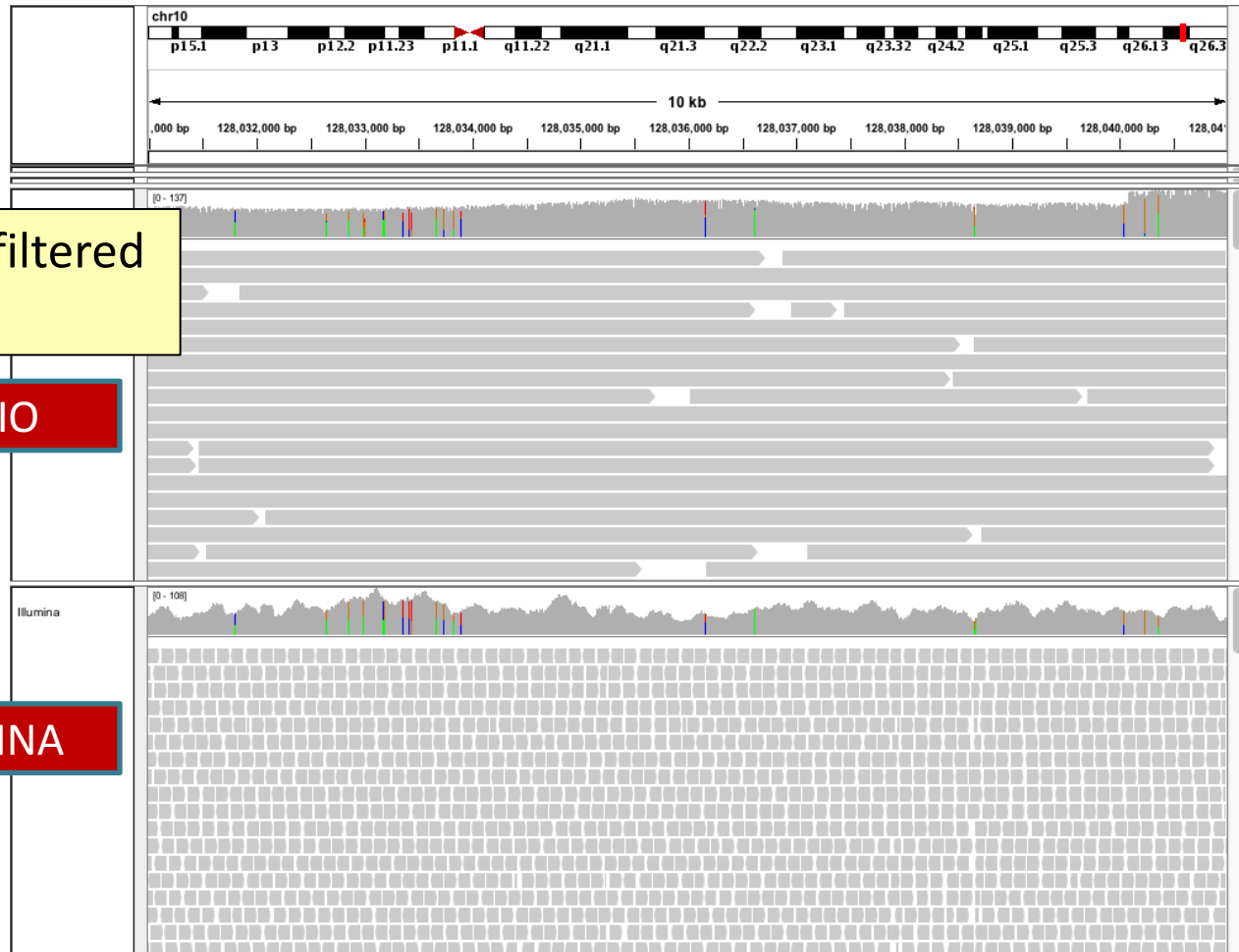


Filtering raw errors - indels

Small indels filtered
from view

PACBIO

ILLUMINA



Filtering raw errors - nucleotides



Filtering raw errors - nucleotides

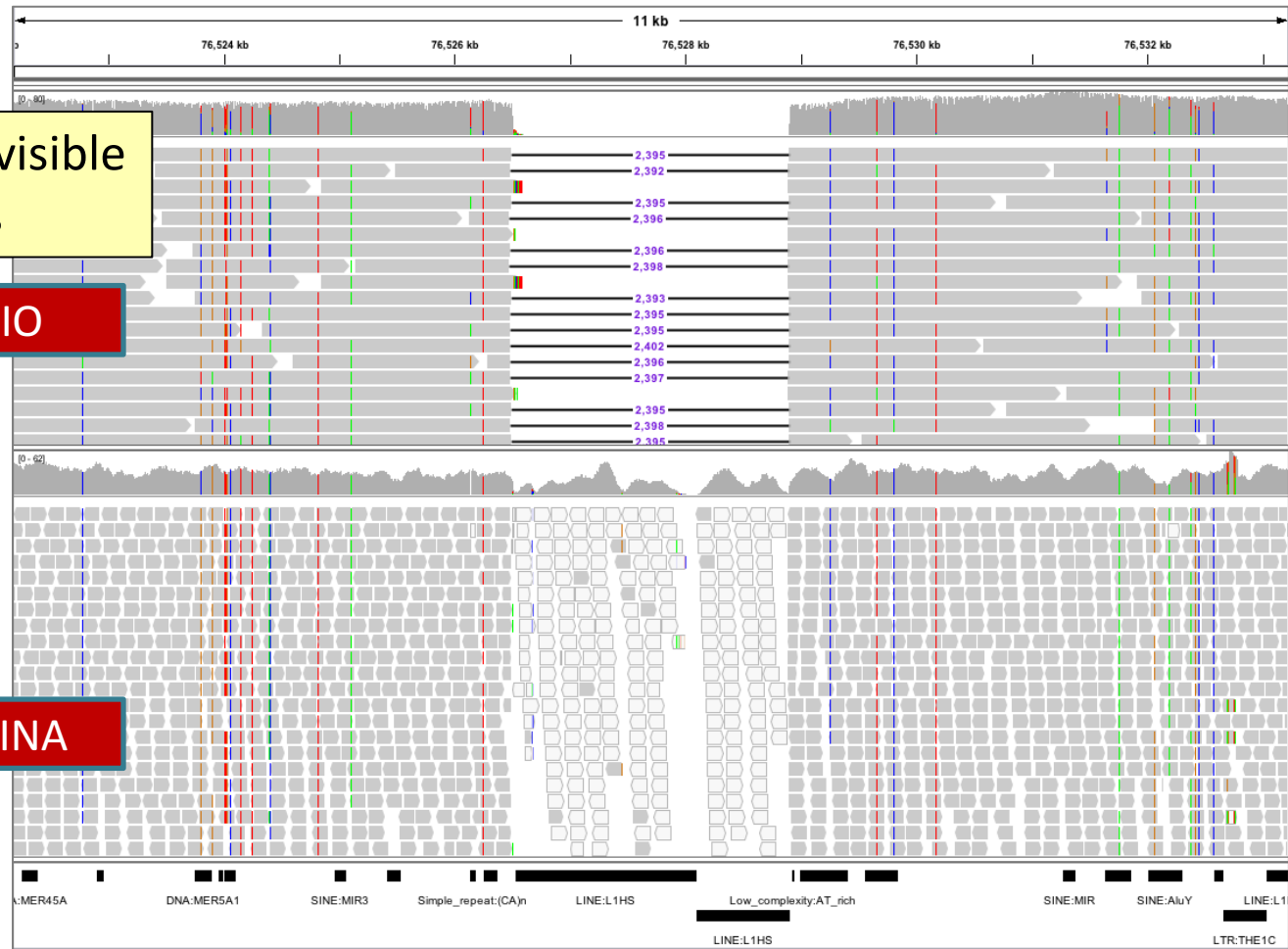


Structural variant example - deletion

Deletion clearly visible
in the long reads

PACBIO

ILLUMINA



View > Preferences

Alignments tab

Purple insertion labels

Hide small indels

Only show consistent mismatches

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

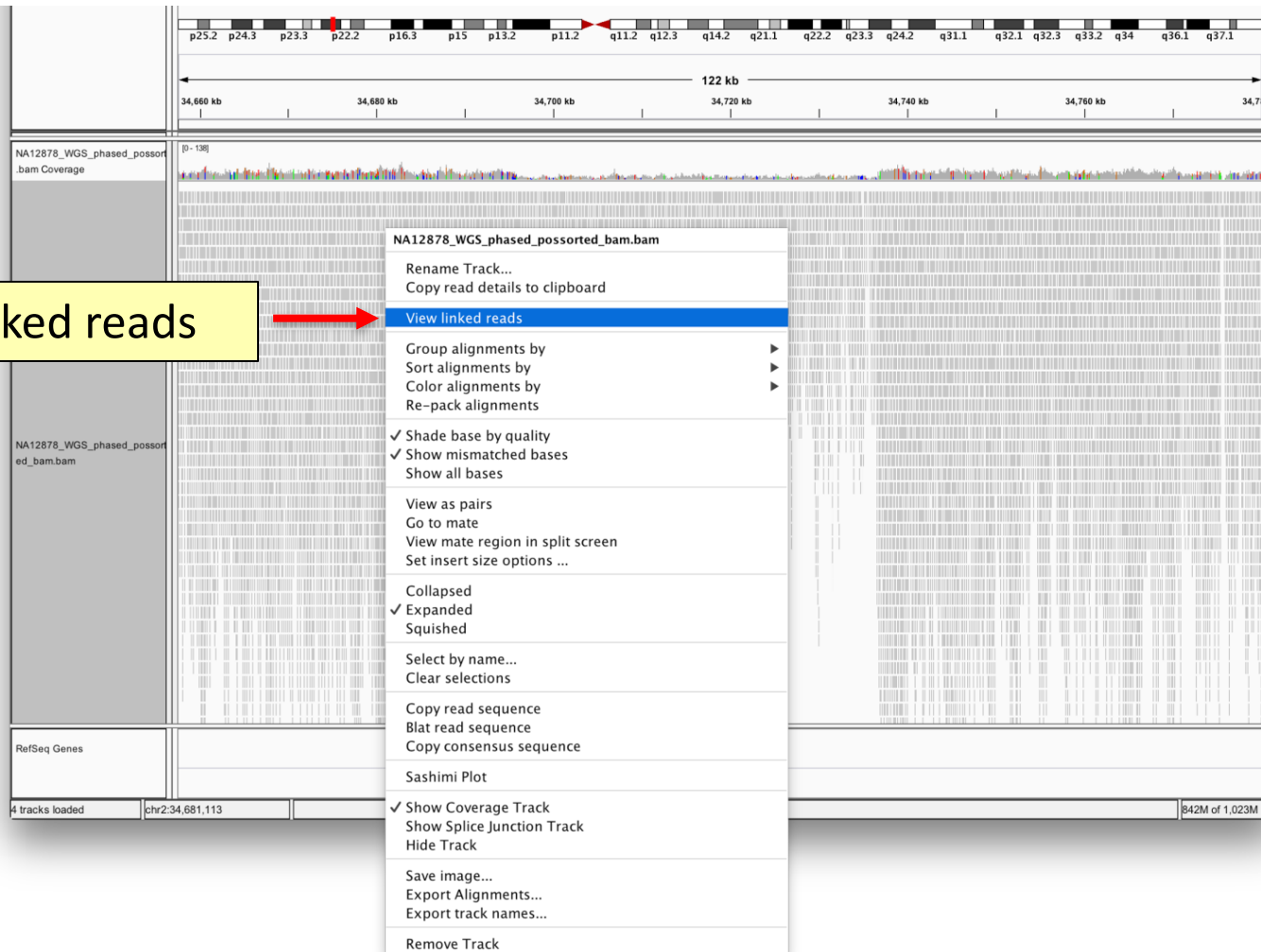
Coverage allele-fraction threshold: 0.25 ☐ Quality weight allele fraction

Splice Junction Track Options

- Long molecules are separated into compartments.
All molecules in same compartment share a barcode.
- Barcodes can be used to reconstruct molecules from short read alignments (Illumina)

IGV enhancements for 10X

View linked reads



IGV enhancements for 10X



Viewing Variants

Variant Call Format (VCF):

File format for encoding variant sites and genotypes

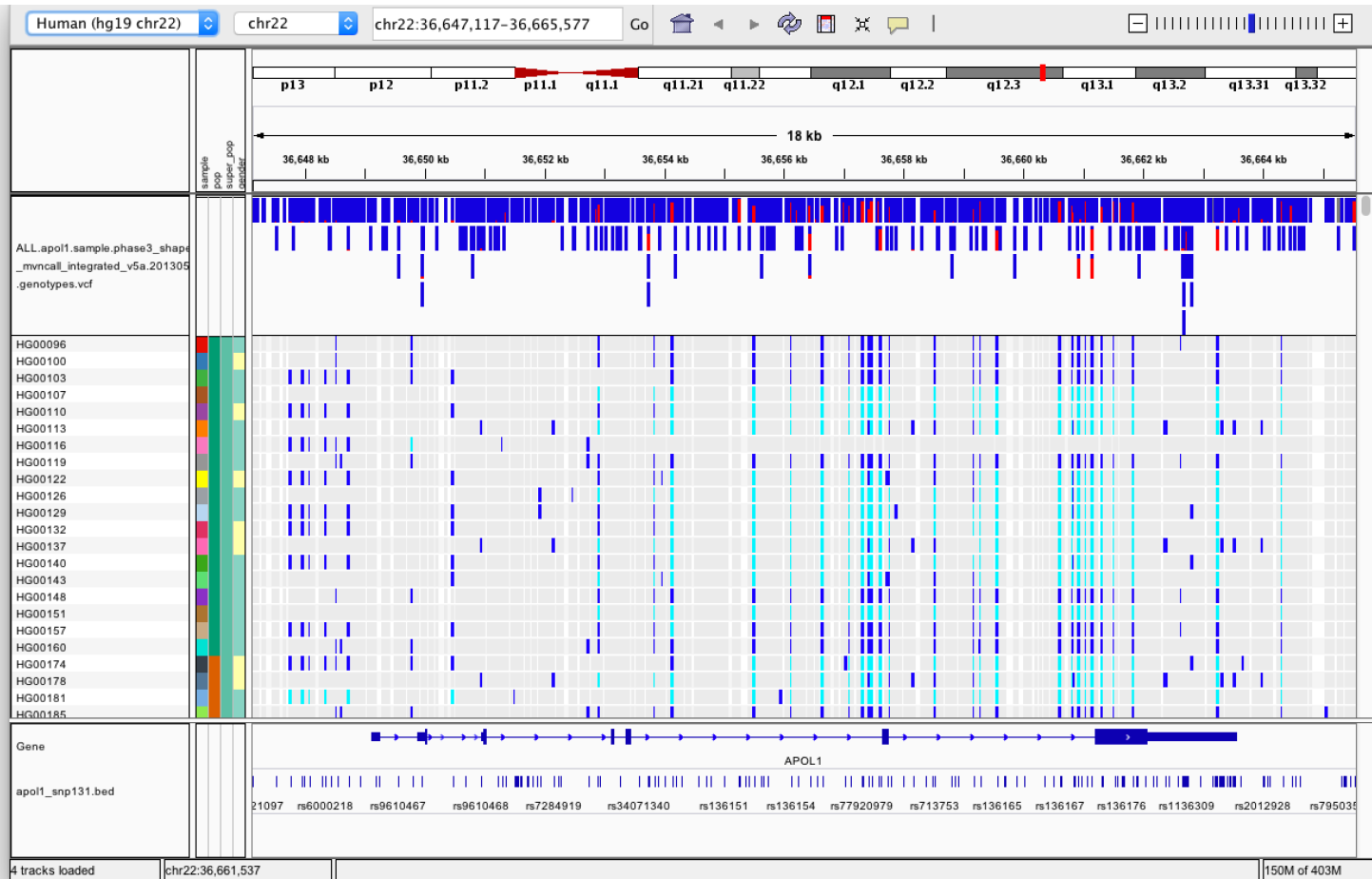
The VCF specification is now maintained by the

Global Alliance for Genomics and Health (GA4GH)

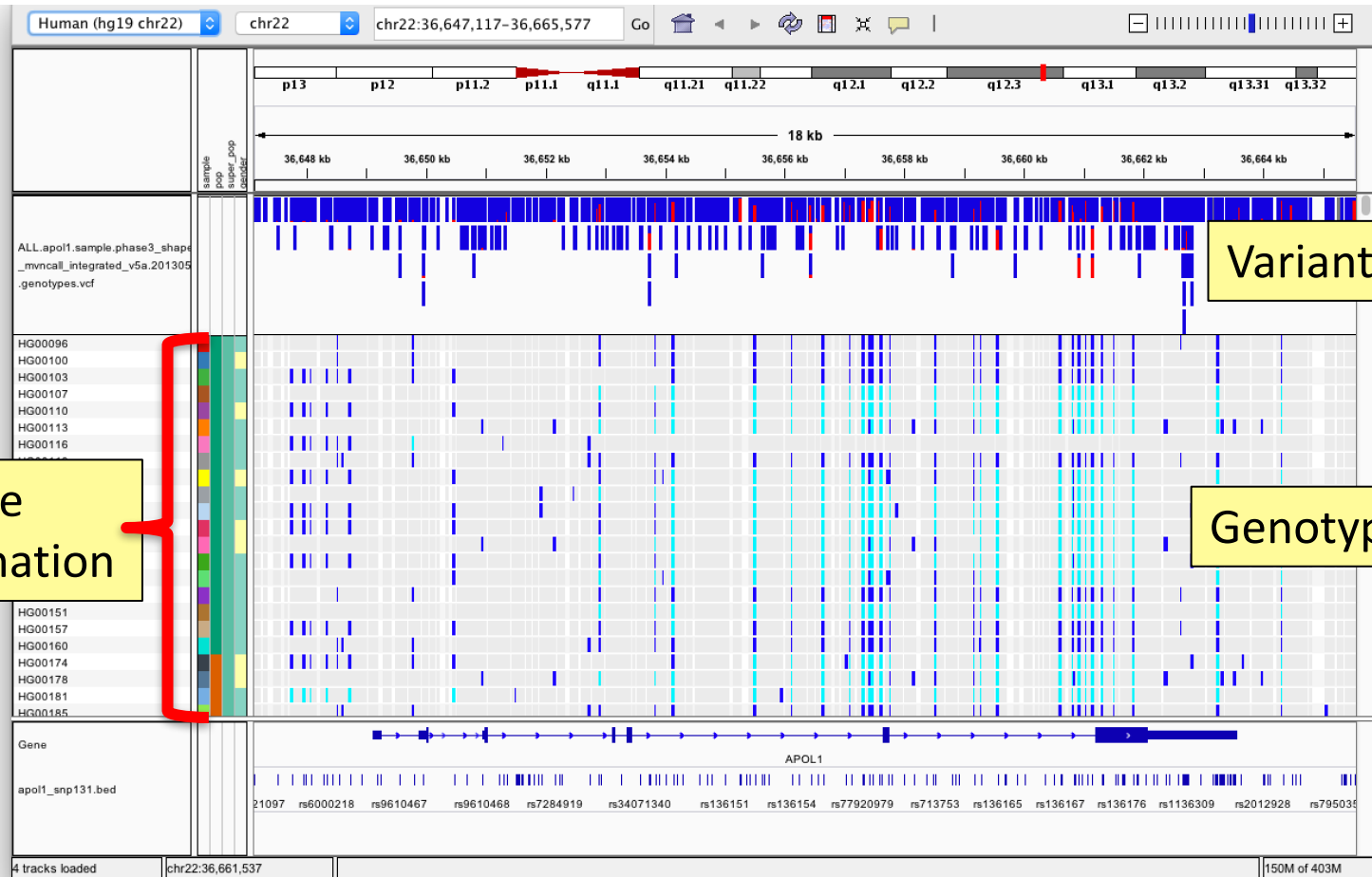
→ *Data Working Group* → *File Formats Task Team*

(<http://ga4gh.org/#/fileformats-team>)

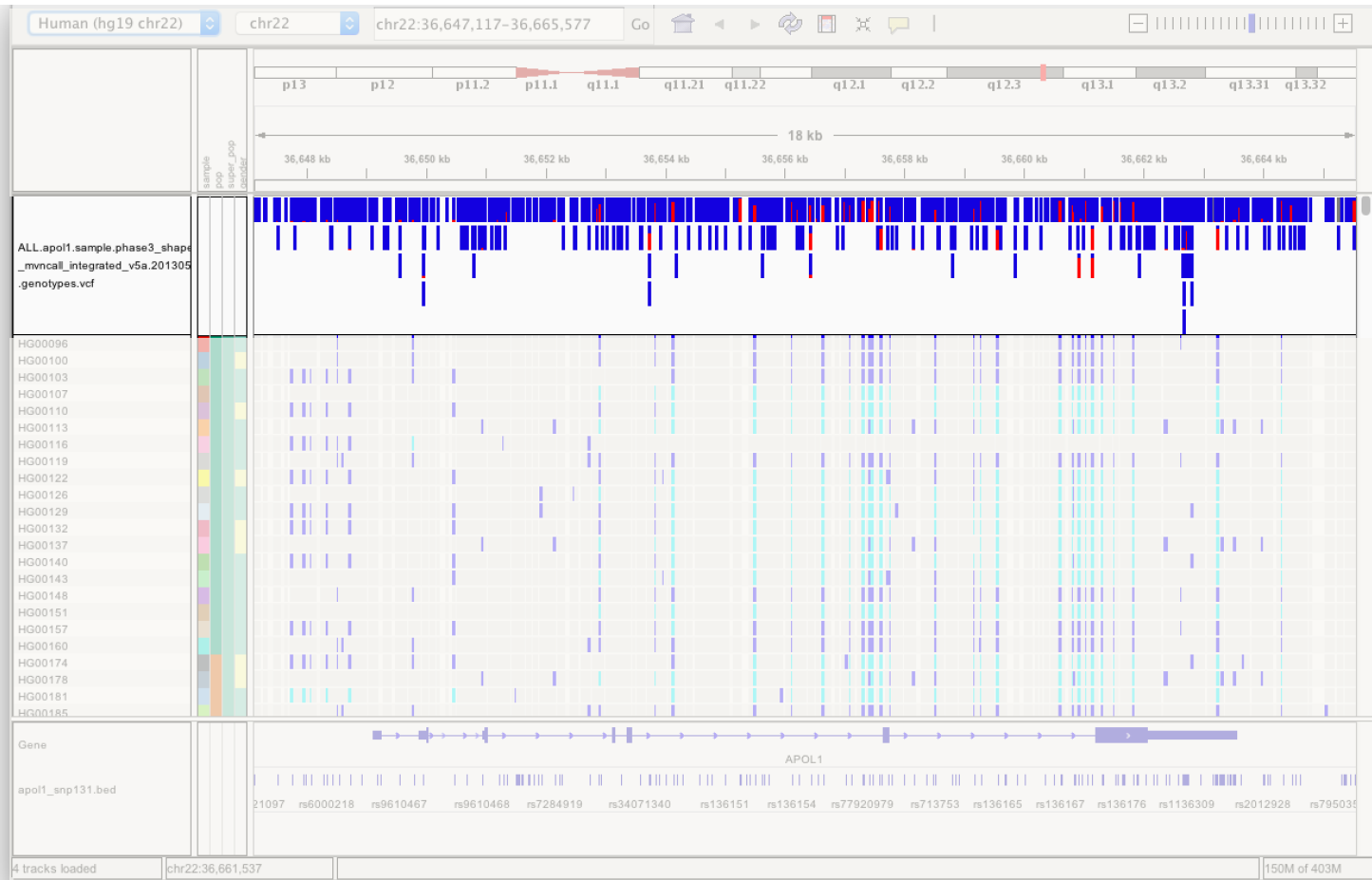
Viewing variants (VCF file)



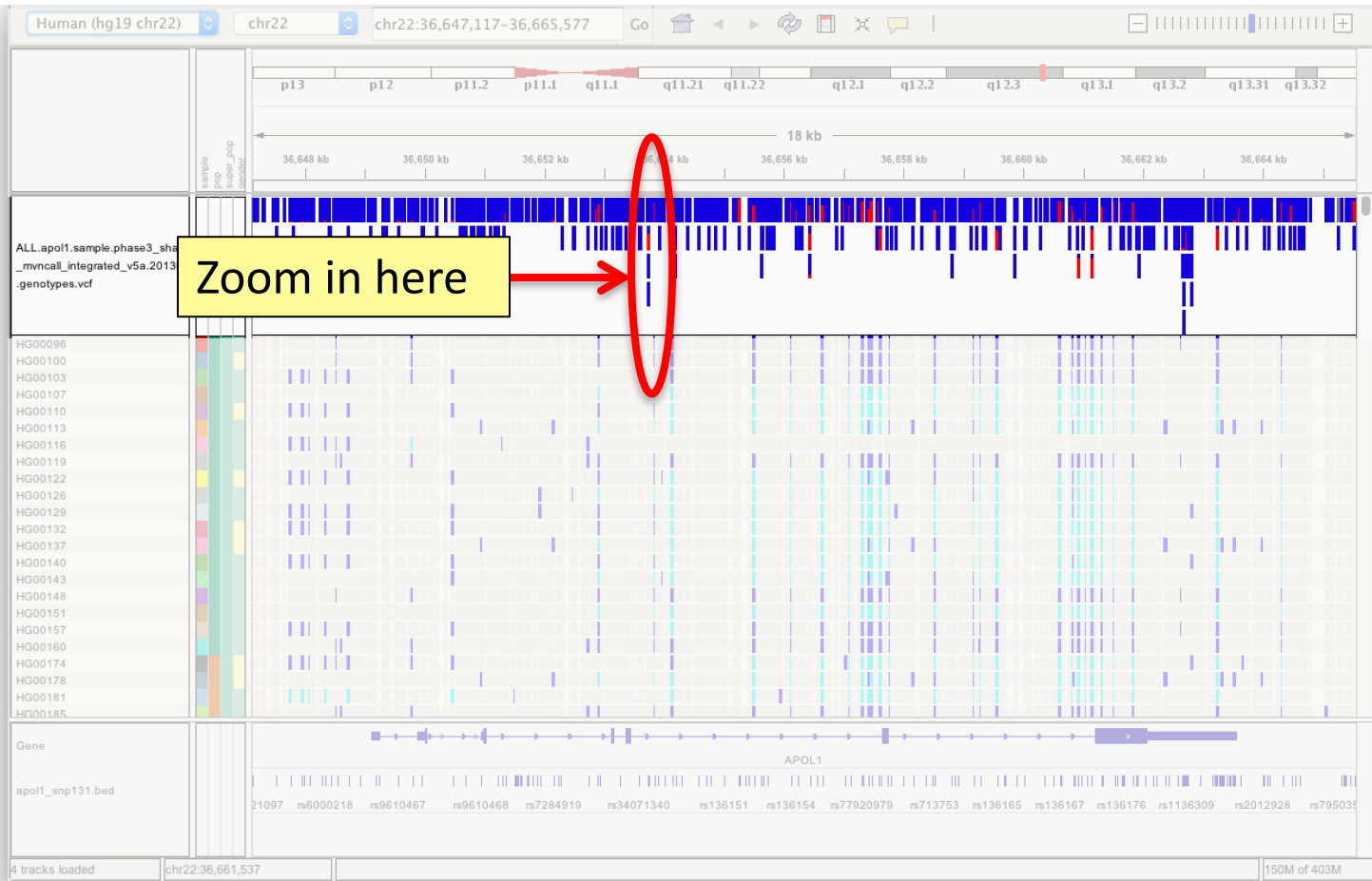
Viewing variants (VCF file)



Viewing variants (VCF file)

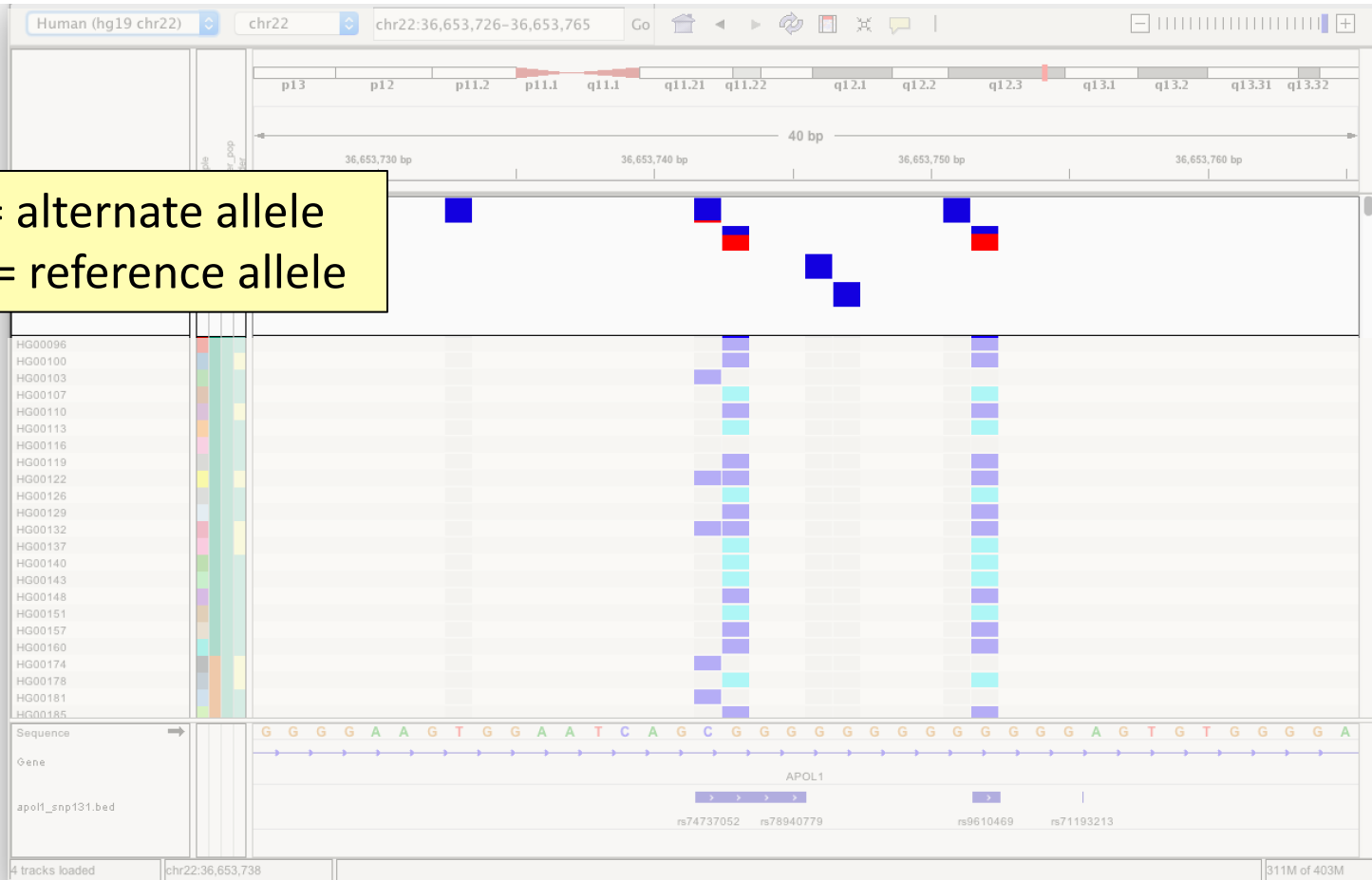


Viewing variants (VCF file)

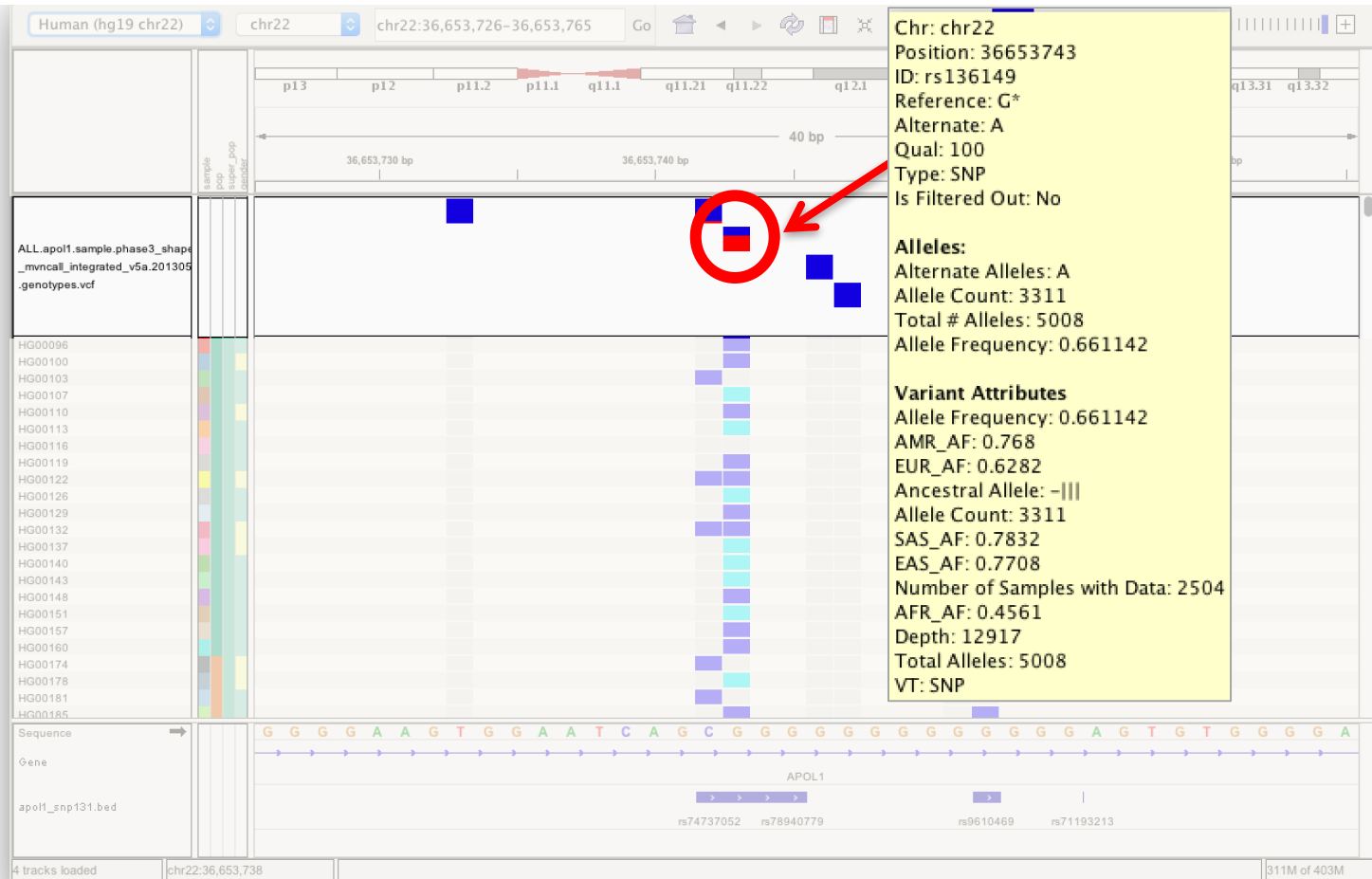


Viewing variants (VCF file)

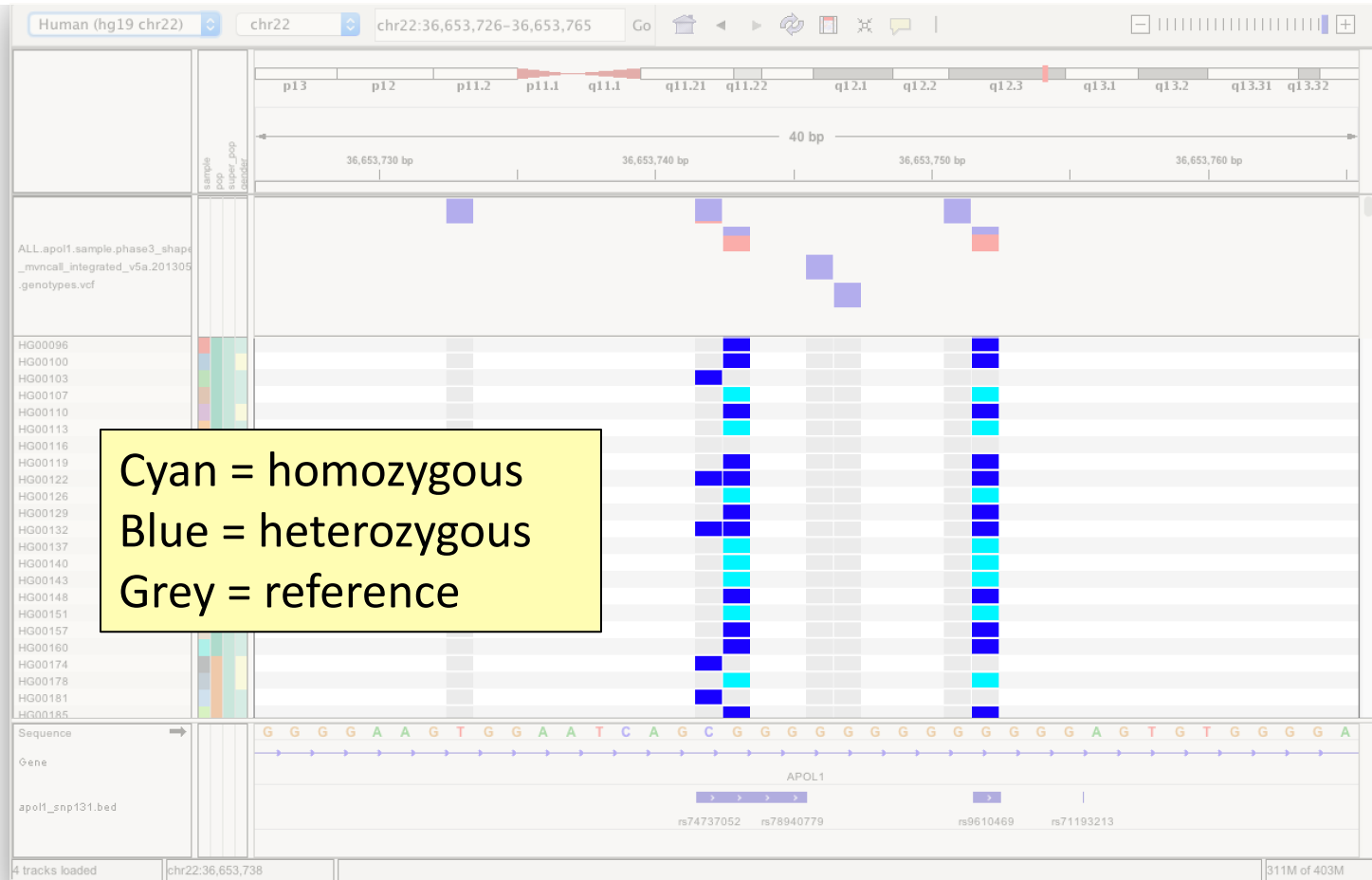
Red = alternate allele
Blue = reference allele



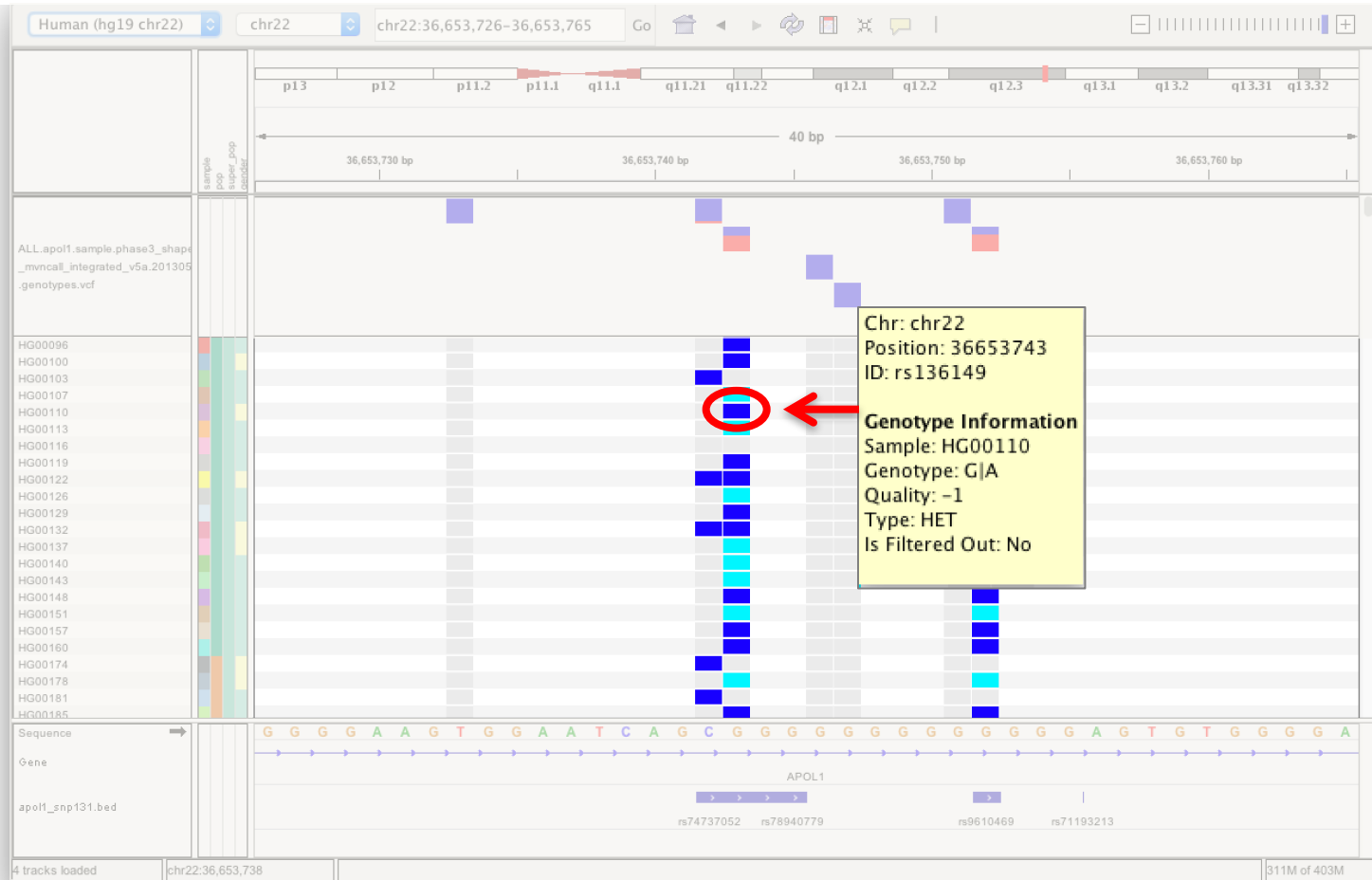
Viewing variants (VCF file)



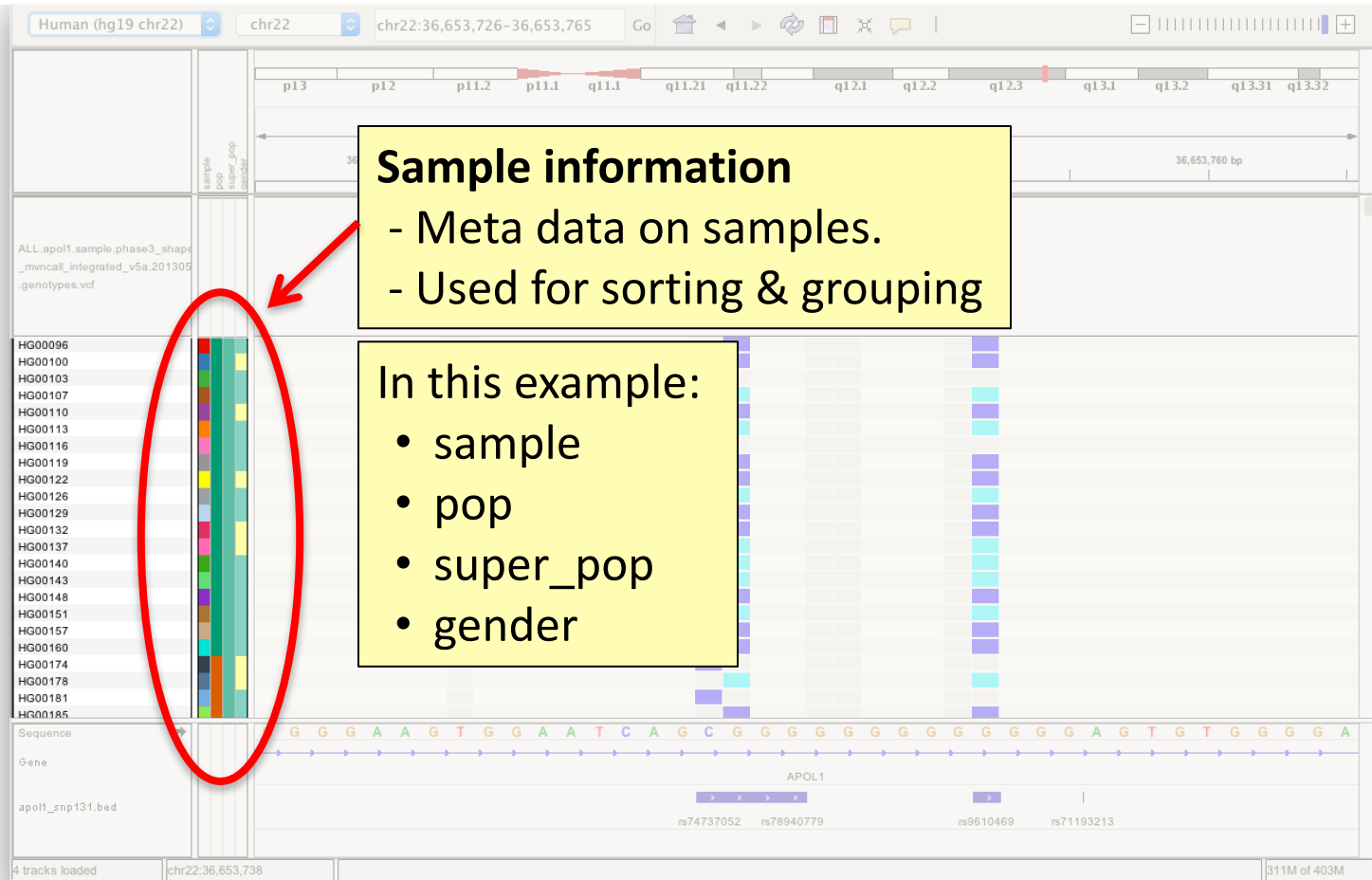
Viewing variants (VCF file)



Viewing variants (VCF file)



Viewing variants – sample information



Viewing variants – sample information

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

- First **row** defines the meta data 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

VCF sample names

Meta data column names

Viewing variants (VCF file)

Hands-on exercise

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

Reference:

Giulio Genovese et al.

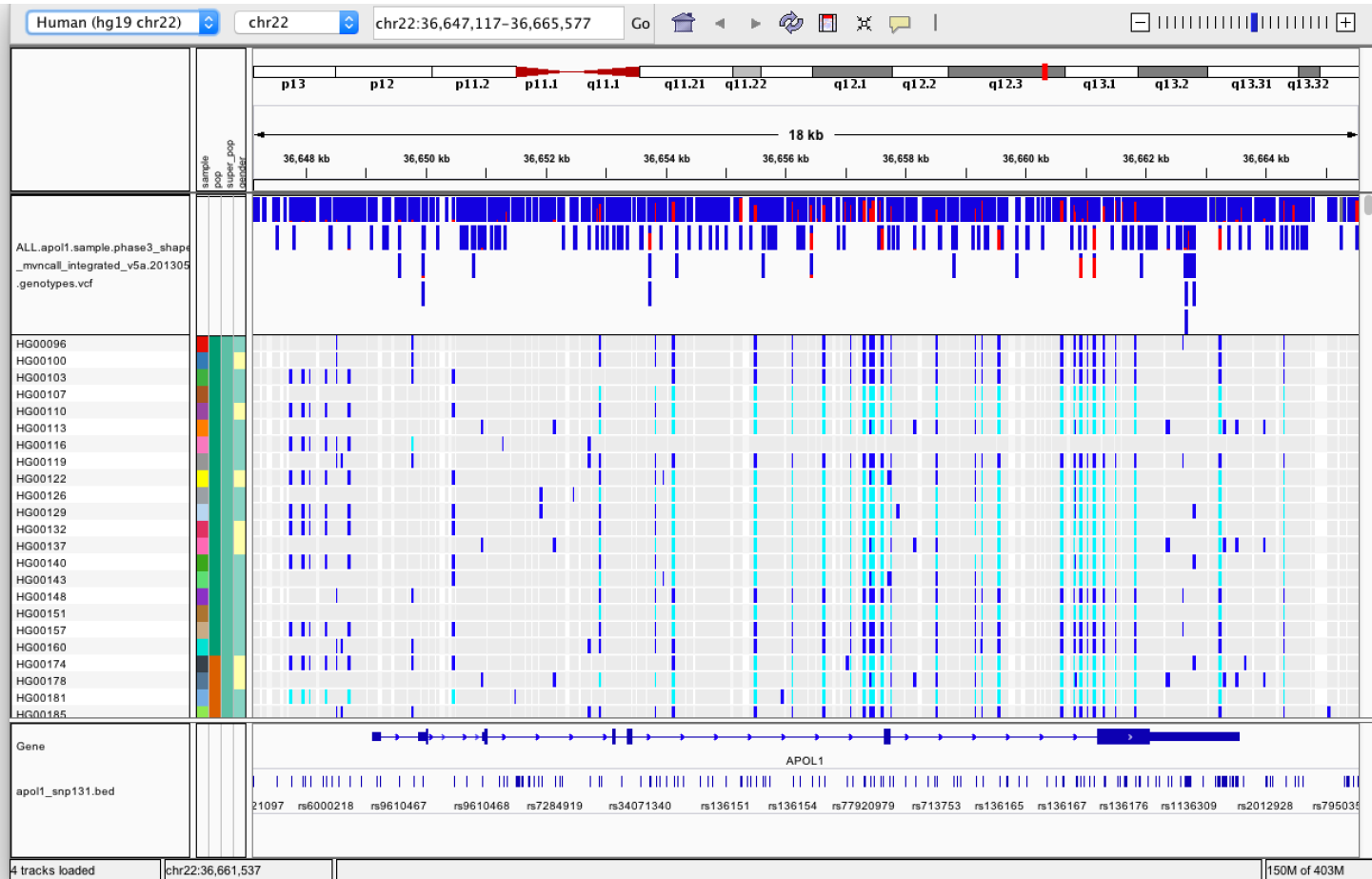
Association of Trypanolytic ApoL1 Variants with Kidney Disease in African Americans,

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

See handout

Viewing variants (VCF file)

Hands-on exercise



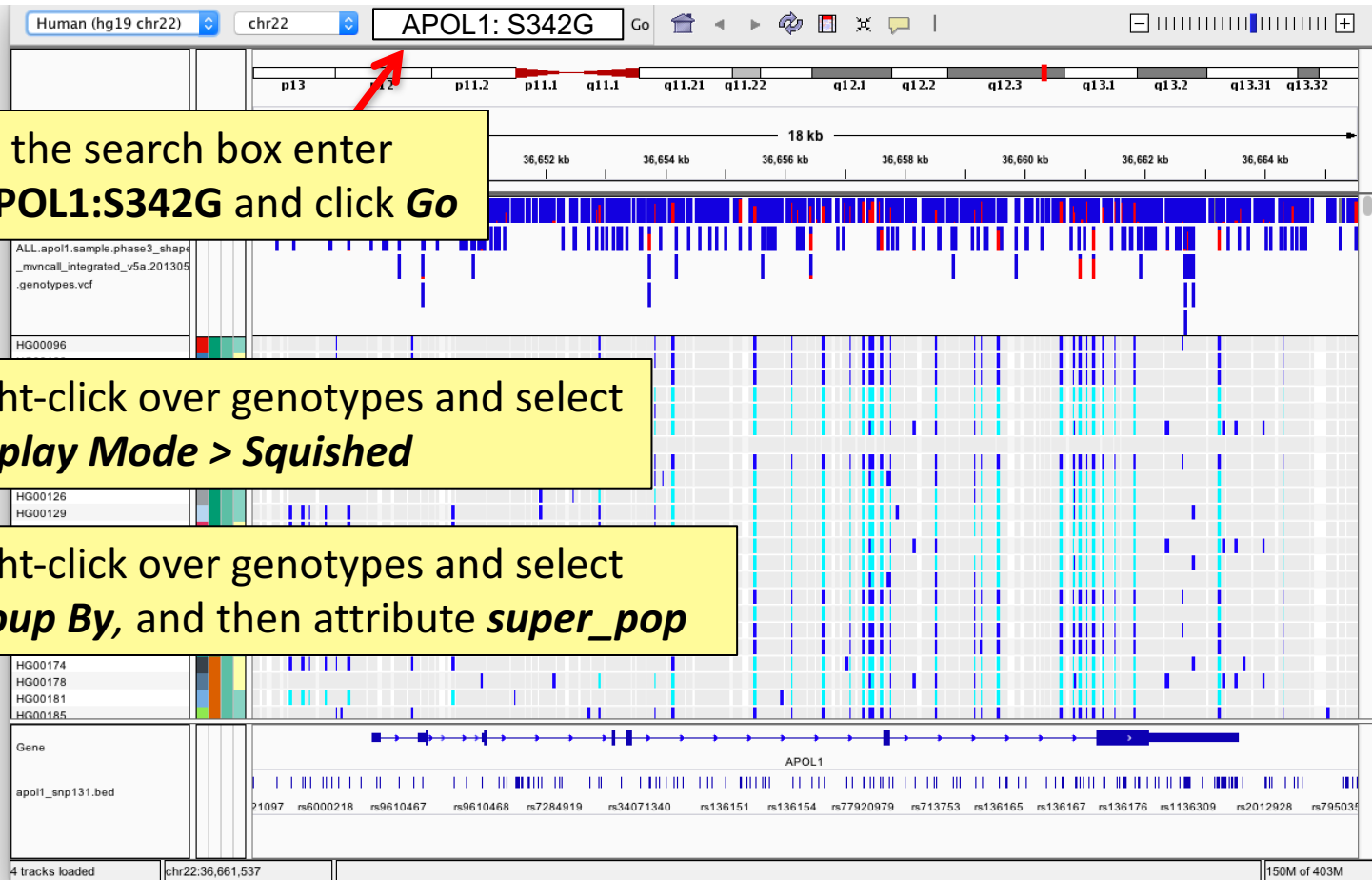
Viewing variants (VCF file)

Hands-on exercise

In the search box enter
APOL1:S342G and click **Go**

Right-click over genotypes and select
Display Mode > Squished

Right-click over genotypes and select
Group By, and then attribute **super_pop**



Viewing variants (VCF file)

Hands-on exercise



Viewing variants (VCF file)

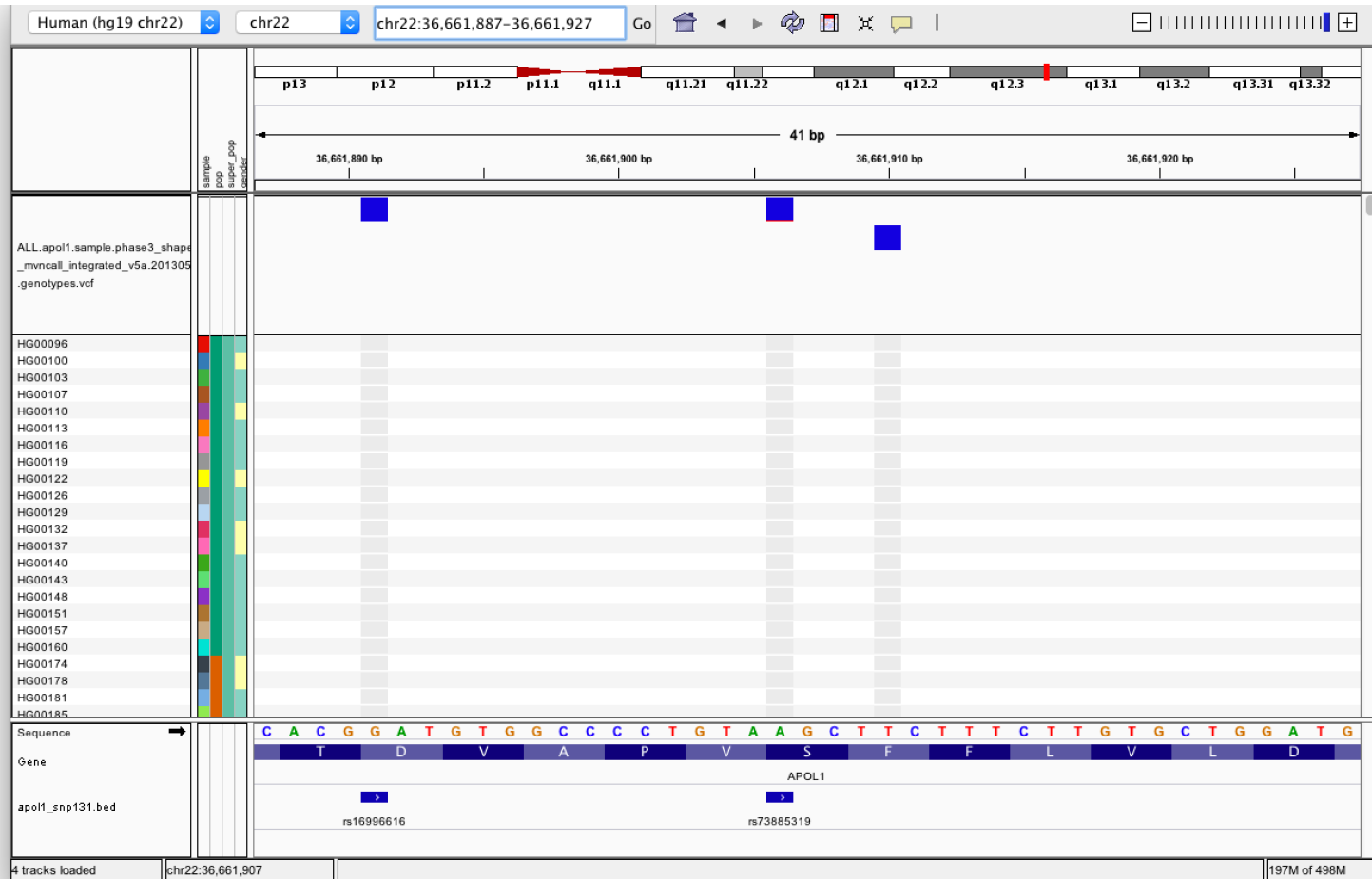
Hands-on exercise

In the search box enter
APOL1:S342G and click **Go**



Viewing variants (VCF file)

Hands-on exercise



Viewing variants (VCF file)

Hands-on exercise

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

Genomic browser tracks:

- chr22:36,661,887-36,661,927 (41 bp)
- ALL.apol1.sample.phase3_shapeit2_mvncall_integrated_v5a.201305.genotypes.vcf
- HG00096
- HG00100
- HG00103
- HG00107
- HG00110
- HG00137
- HG00140
- HG00143
- HG00148
- HG00151
- HG00157
- HG00160
- HG00174
- HG00178
- HG00181
- HG00185

Sequence: C A C G G A T G T G G C C C T

Gene: apol1_snp131.bed

Variant: rs16996616

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

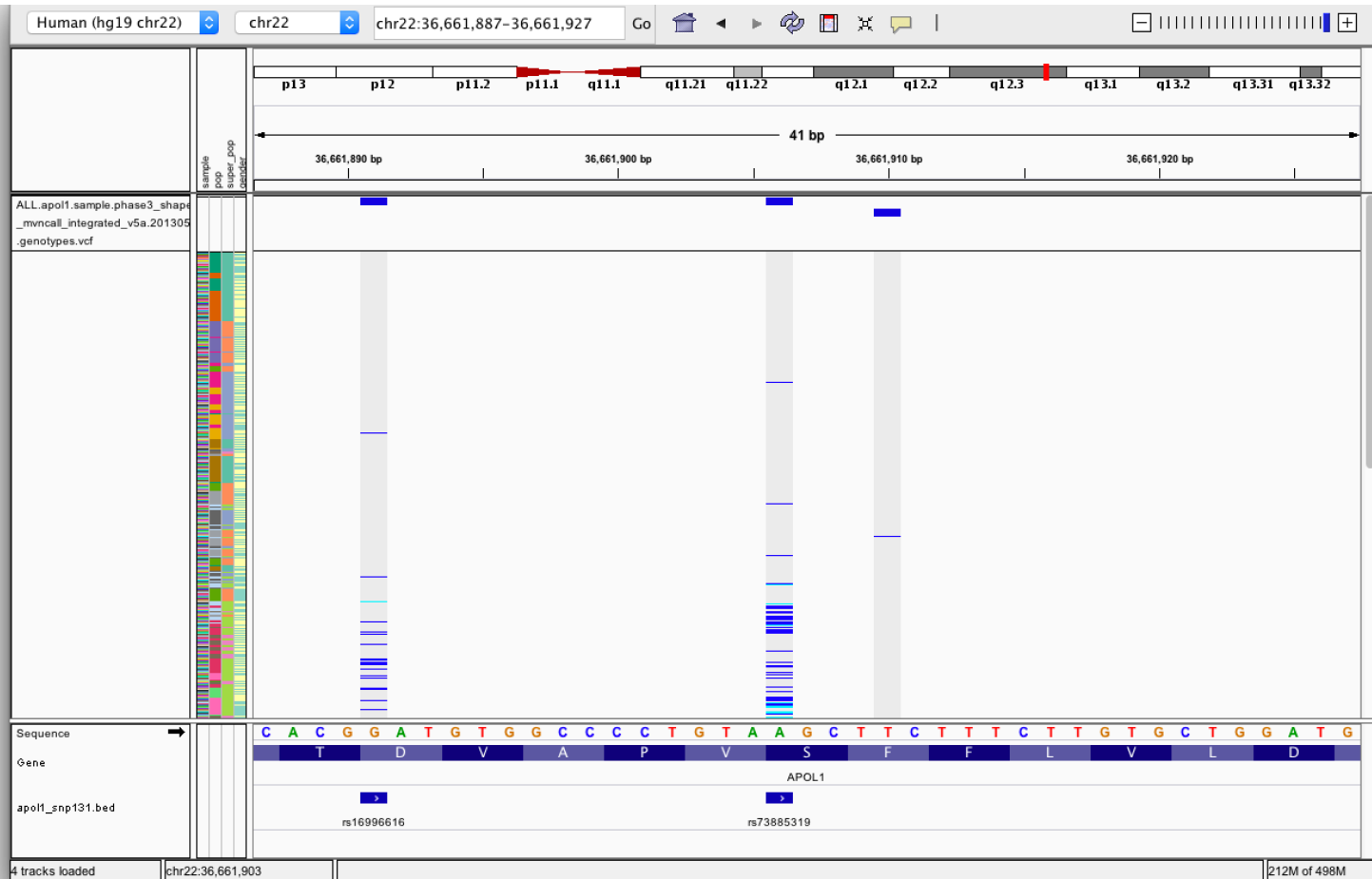
Right-click over genotypes and select **Display Mode > Squished**

Context menu options:

- Rename Track...
- Change Font Size...
- Color By
 - ✓ Allele Frequency
 - Allele Fraction
- Sort By
 - Genotype
 - Sample Name
 - Depth
 - 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

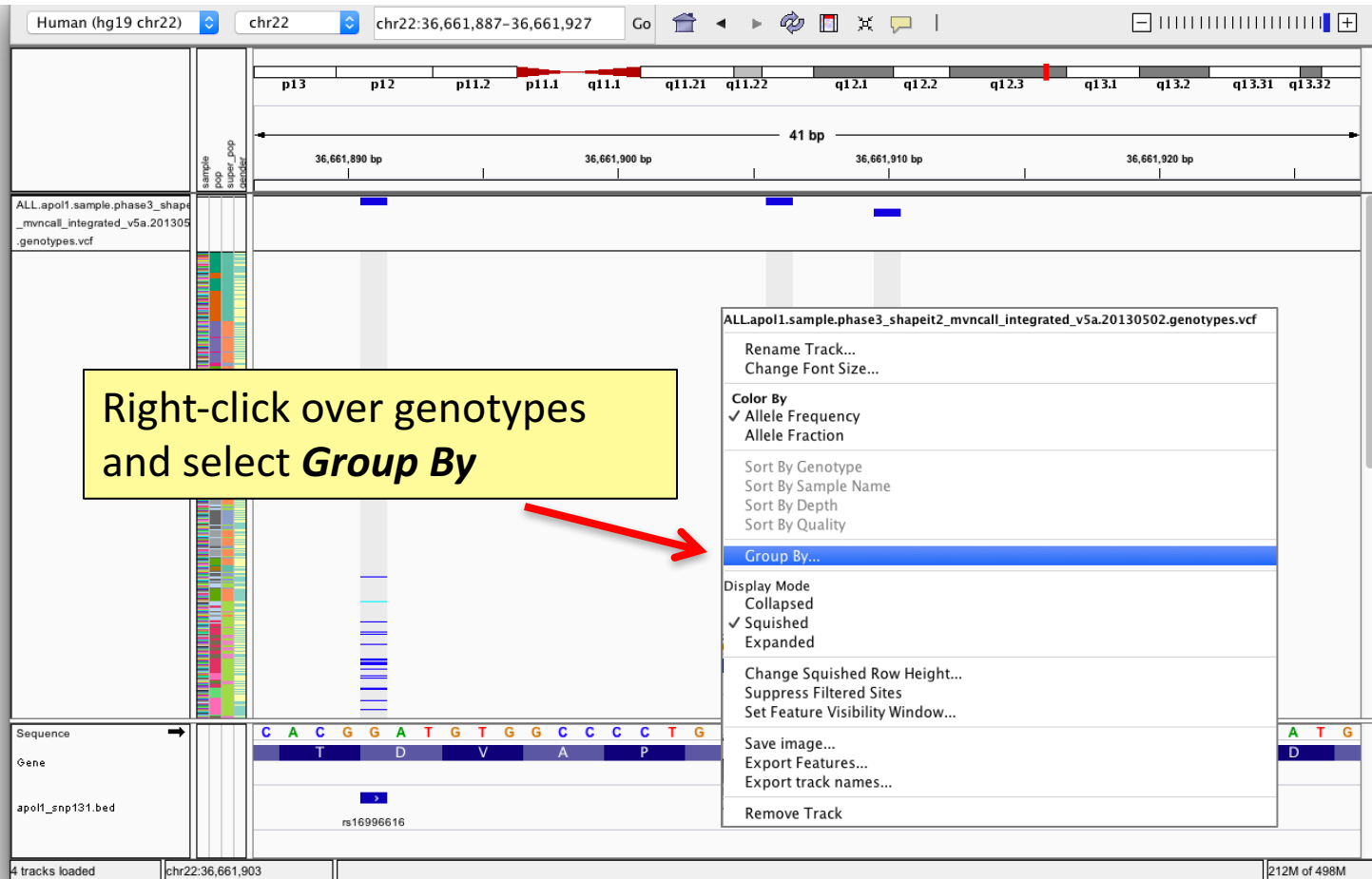
Viewing variants (VCF file)

Hands-on exercise



Viewing variants (VCF file)

Hands-on exercise



Viewing variants (VCF file)

Hands-on exercise

The screenshot shows a genomic browser interface for Human (hg19 chr22). The main view displays a genomic track for chr22:36,661,887-36,661,927. The track is divided into bands representing different populations: p13, p12, p11.2, p11.1, q11.1, q11.21, q11.22, q12.1, q12.2, q12.3, q13.1, q13.2, q13.31, and q13.32. A 41 bp scale bar is shown above the track. Below the track, a list of variants is displayed, including rs16996616 and rs73885319. 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. A yellow callout box with the text 'Select attribute super_pop' and a red arrow points to the 'super_pop' option in the dialog.

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

ALL.apo1f.sample.phase3_shape_mvnca1_integrated_v5a.201305.genotypes.vcf

Group By Attribute

Group tracks by:

- ✓ None
- sample
- pop
- super_pop**
- gender

Select attribute **super_pop**

Sequence

Gene

apo1f_snp131.bed

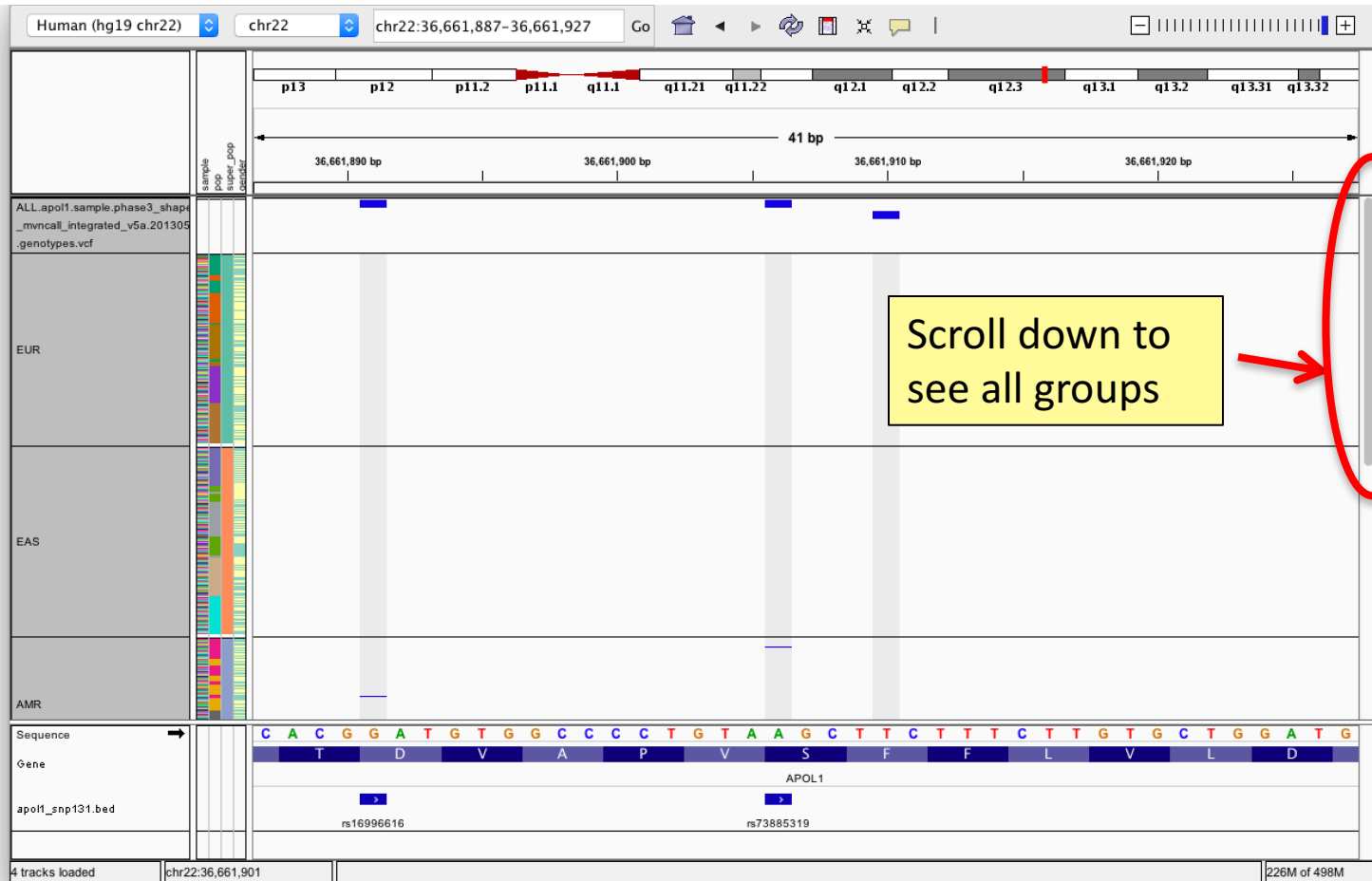
APOL1

rs16996616 rs73885319

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

Viewing variants (VCF file)

Hands-on exercise



Viewing variants (VCF file)

Hands-on exercise

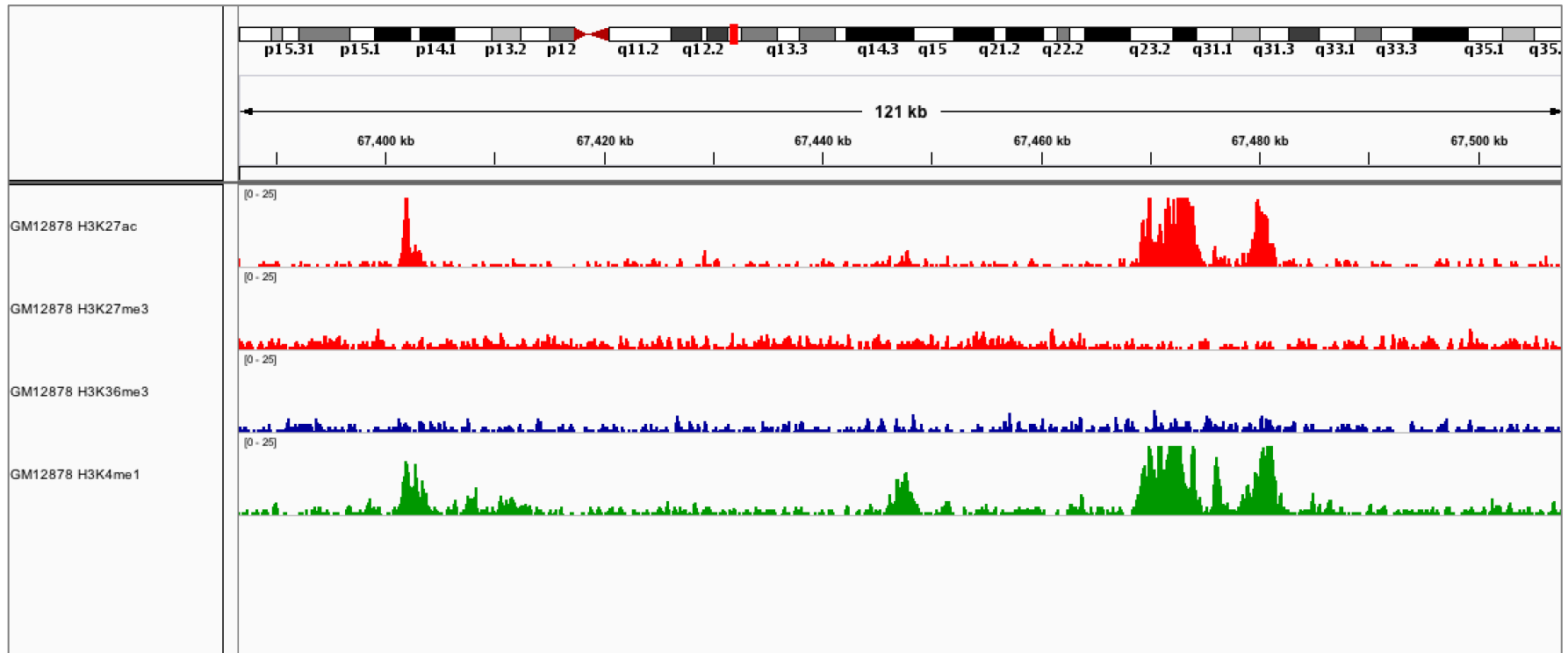


Other Tools

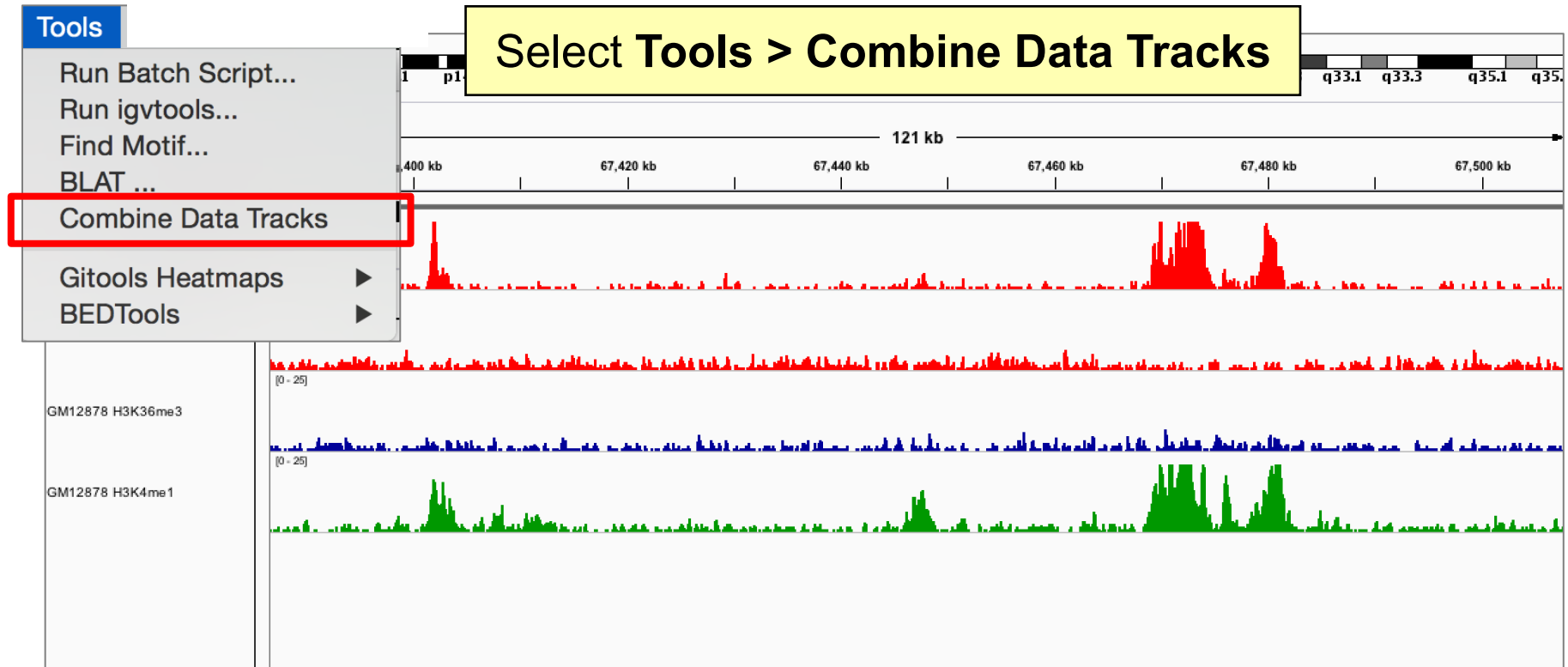
Tools

- Combining numeric data tracks
- BLAT

Combining numeric data tracks



Combining numeric data tracks



Combining numeric data tracks

1. Select two tracks from the dropdown menus

2. Select the operation:

- Add
- Subtract
- Multiply
- Divide

3. Name the new combined track

The screenshot shows a 'Combine tracks' dialog box. It has three dropdown menus: 'Track A' (selected: GM12878 H3K27ac), 'Operation' (selected: Add), and 'Track B' (selected: GM12878 H3K27me3). Below these is a text field for 'Result Track Name' containing the text 'Sum'. At the bottom are 'OK' and 'Cancel' buttons. Three yellow callout boxes with black borders and red arrows point to the 'Track A' dropdown, the 'Operation' dropdown, and the 'Result Track Name' text field.

Combine tracks

Track A: GM12878 H3K27ac

Operation: Add

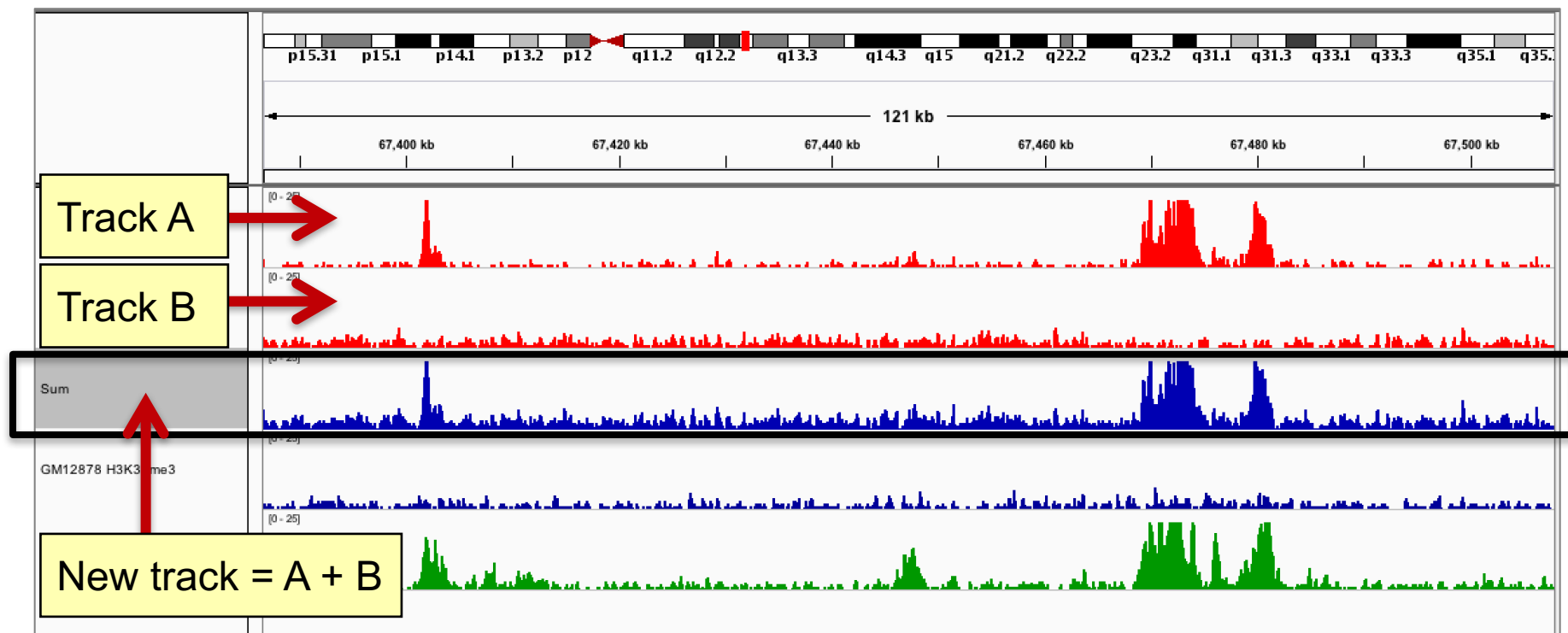
Track B: GM12878 H3K27me3

Result Track Name

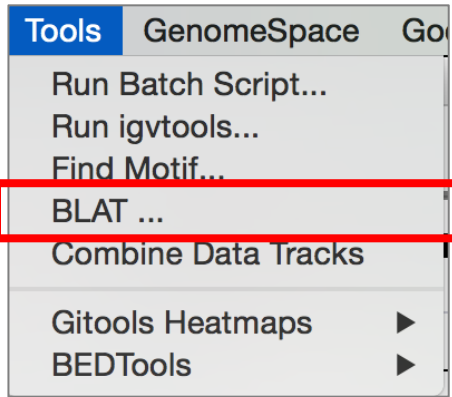
Sum

OK Cancel

Combining numeric data tracks

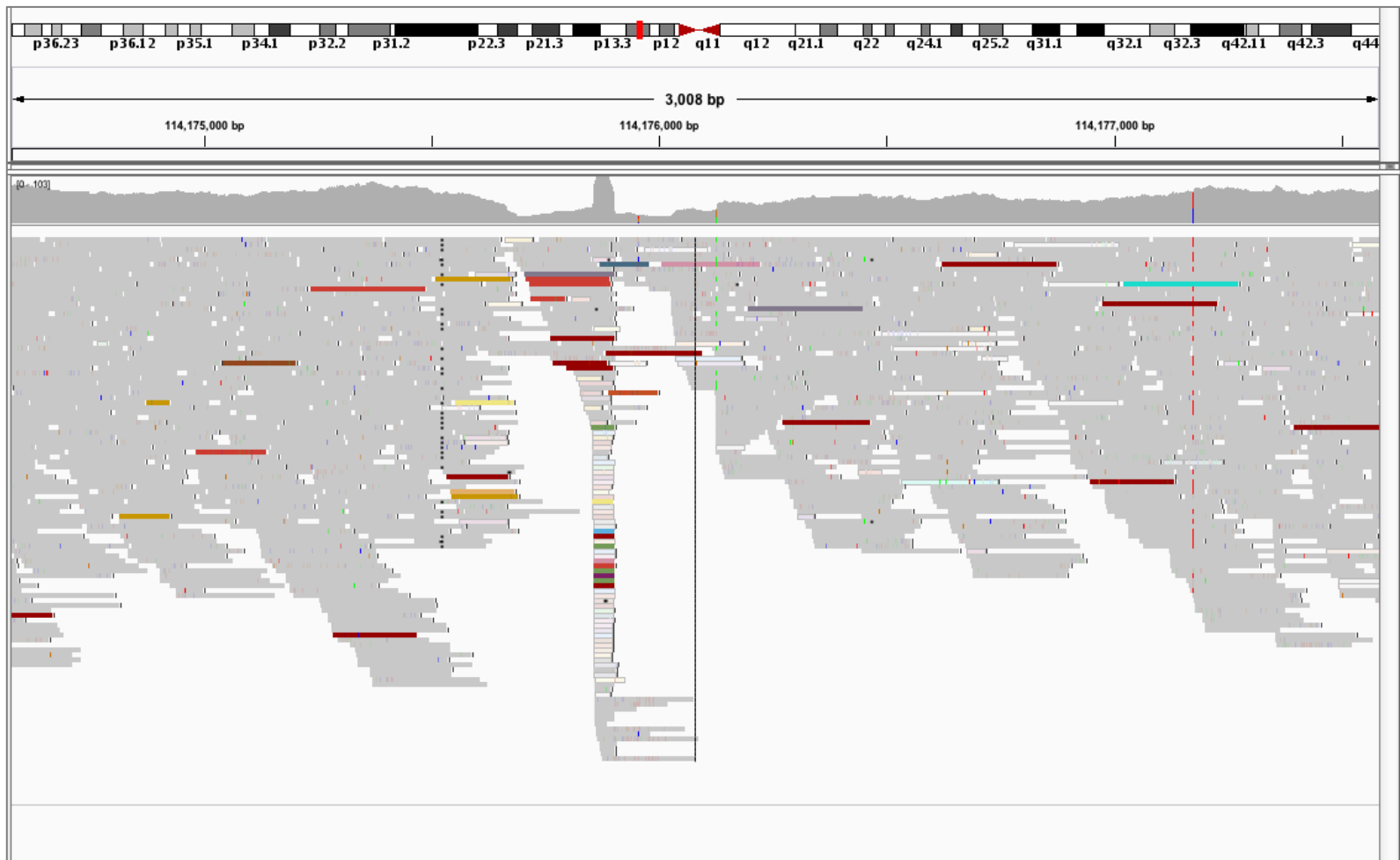


BLAT

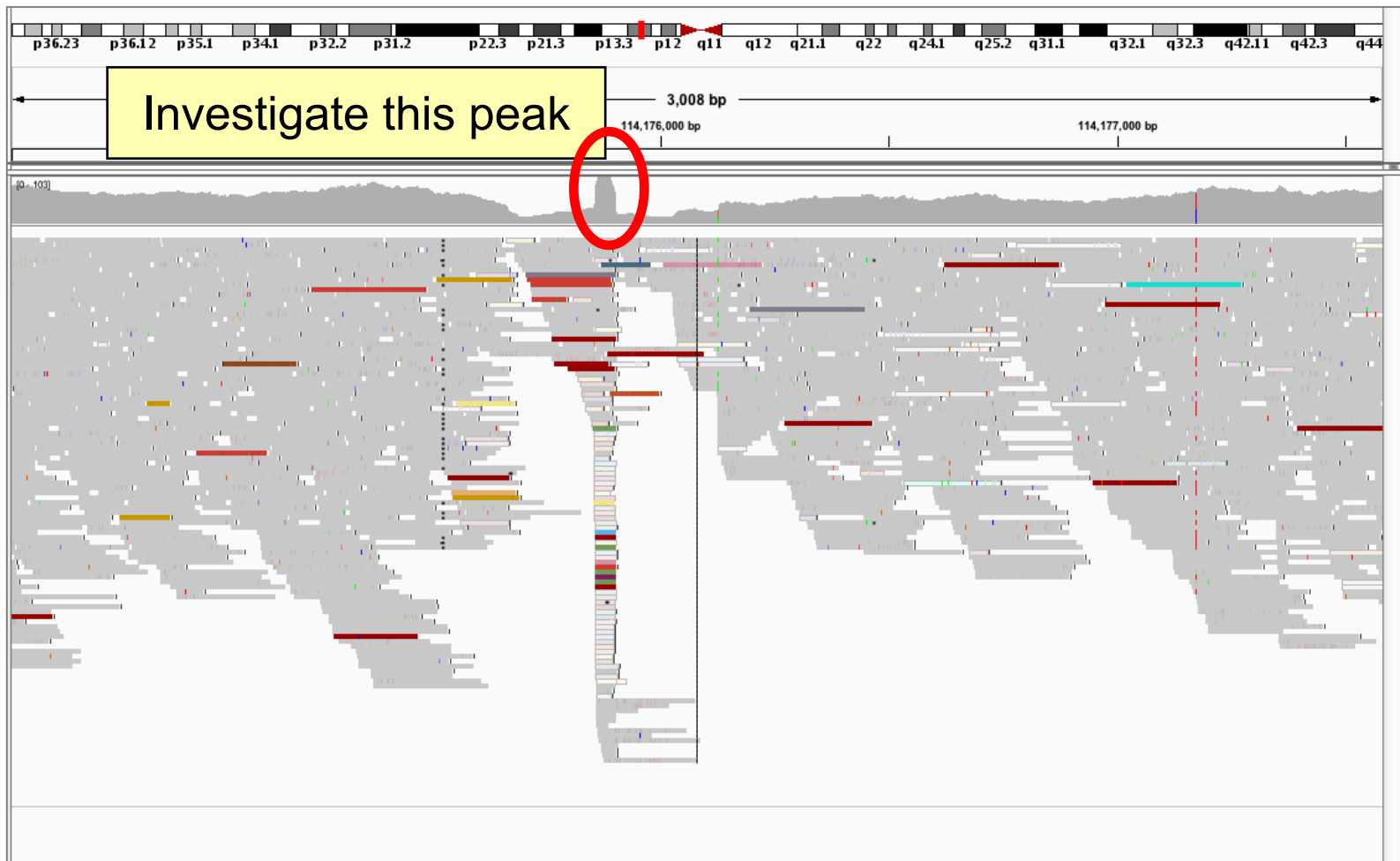


- Use a BLAT server to do search for alternative alignments for
 - any feature or sequence read, or
 - user-entered sequence.
- User-specified server or UCSC (volume limits apply).

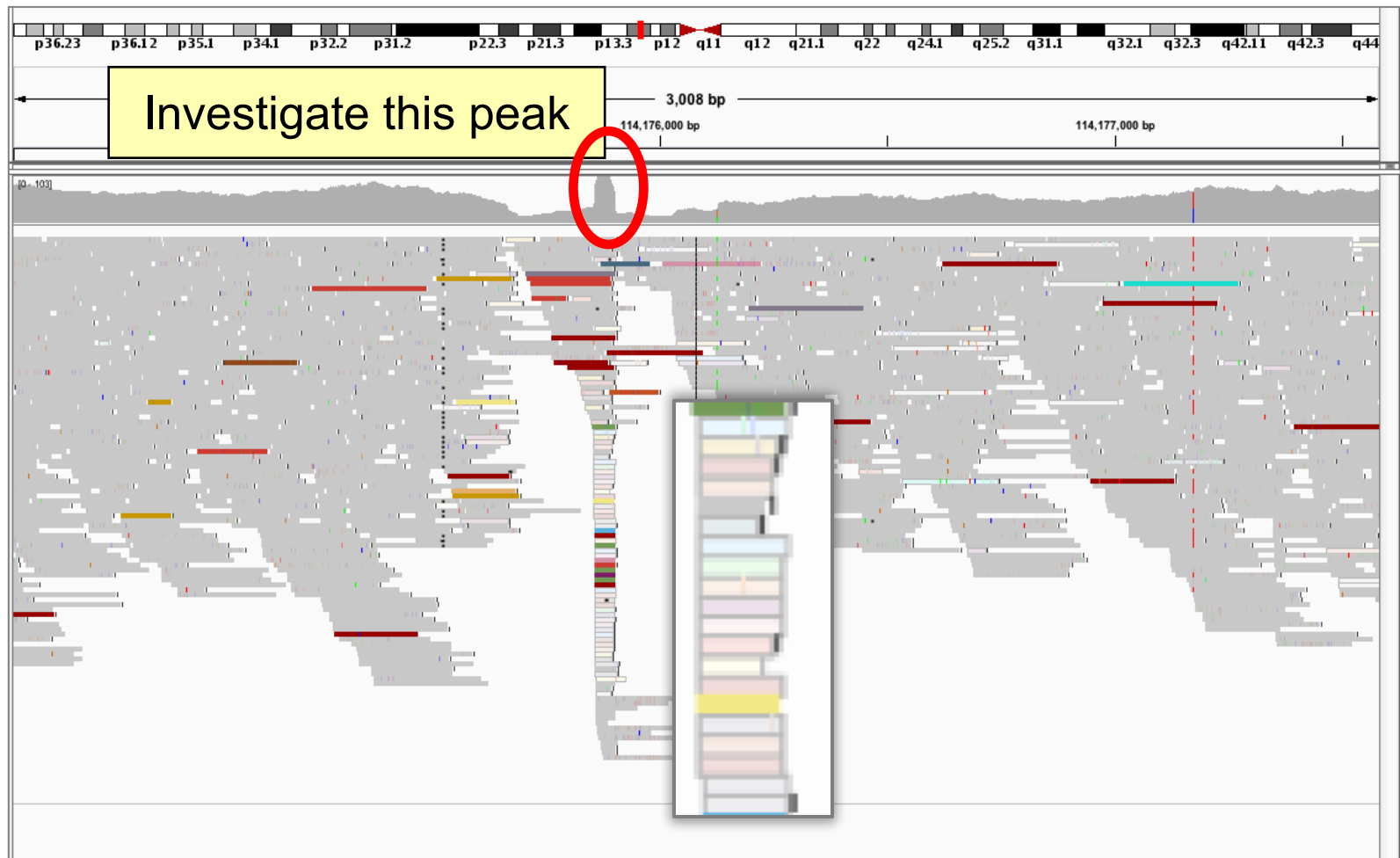
BLAT example



BLAT example

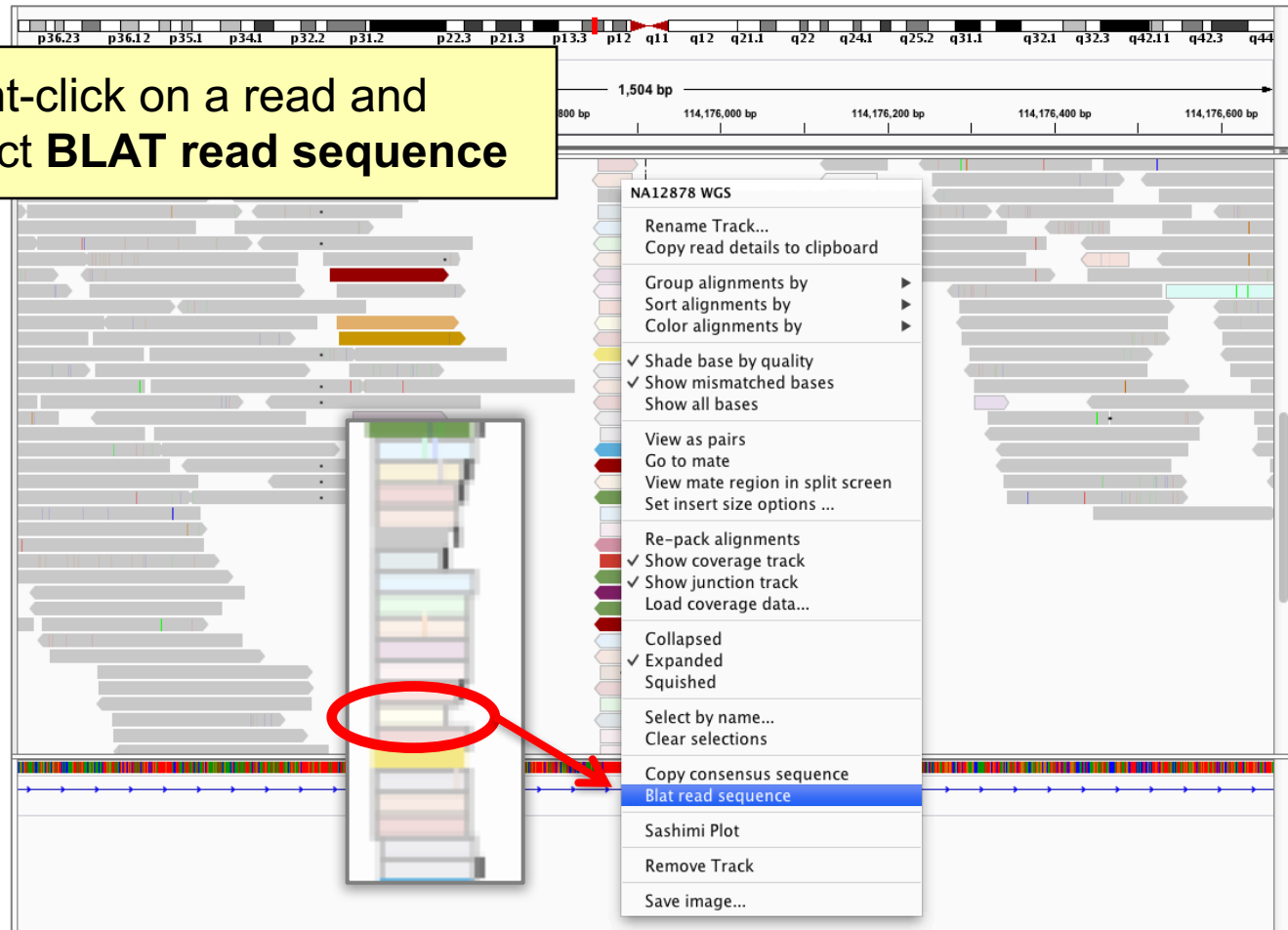


BLAT example



BLAT example

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



BLAT example

File													
BLAT result for query sequence: TAATGCACACGAACTCCATTAGGTATTCATATGGGTTGAAAAGGGGGGTAACTTGGCCTAAATAGAAAATGTCTTCTCATCATTTTTTT													
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	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	
chrX	147458...	147458...	-	356	108	17	0	0	1	35	1	36	

BLAT example

Many alternative alignments
with similar scores

TTGAAAAGGGGGGTAACTTGGCCTAAATAGAAAATGTCTTCTCATCATTTTTT

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
chrX	147458...	147458...	-	356	108	17	0	0	1	35	1	36

Closing

For further information and help

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.

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