

Visualization of a structural
variant on igv web app

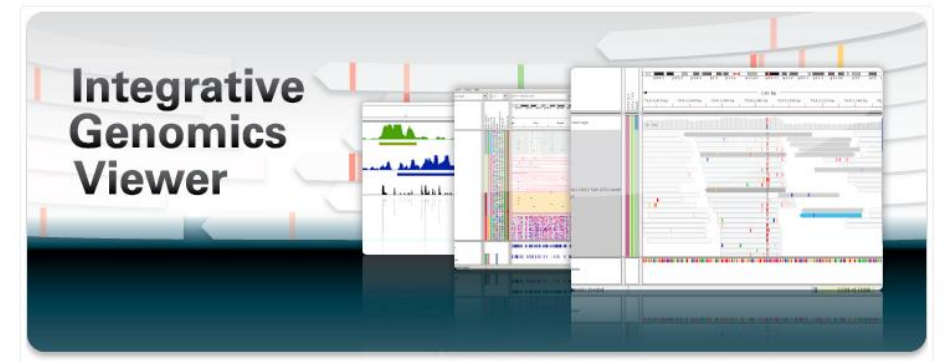
Visualization of evidence of a structural variant from aligned reads using igv web app

Integrative Genomics Viewer (IGV) is a high-performance, easy-to-use, interactive tool for the visual exploration of genomic data.

IGV is available in multiple forms, including:

- the original IGV - a Java desktop application,
- IGV-Web - a web application,
- igv.js - a JavaScript component that can be embedded in web pages (for developers)

We are going to use the web application : <https://igv.org/app/>



Dataset

Sample : NA19240

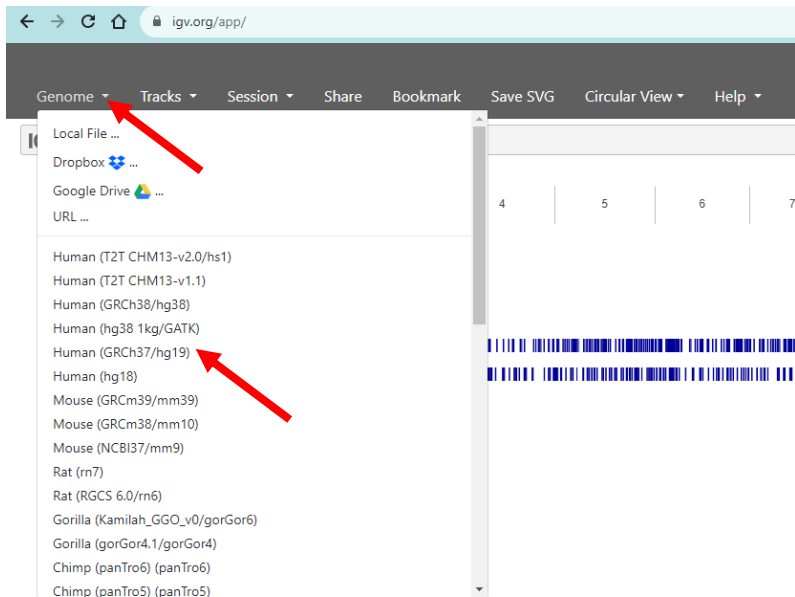
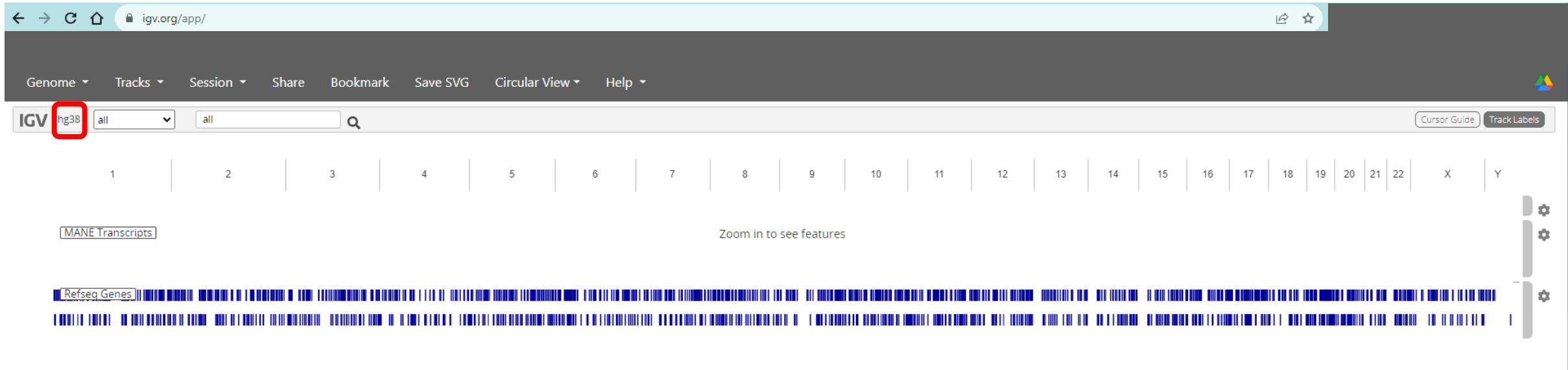
Data type : aligned reads in bam format restricted to a chromosome 21 region

Human reference sequence version : hg19

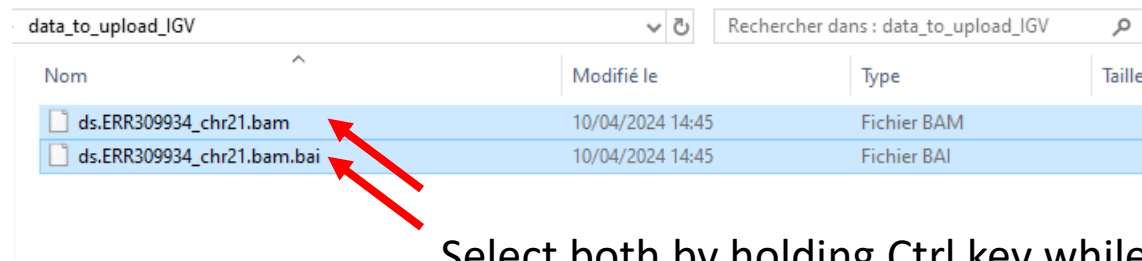
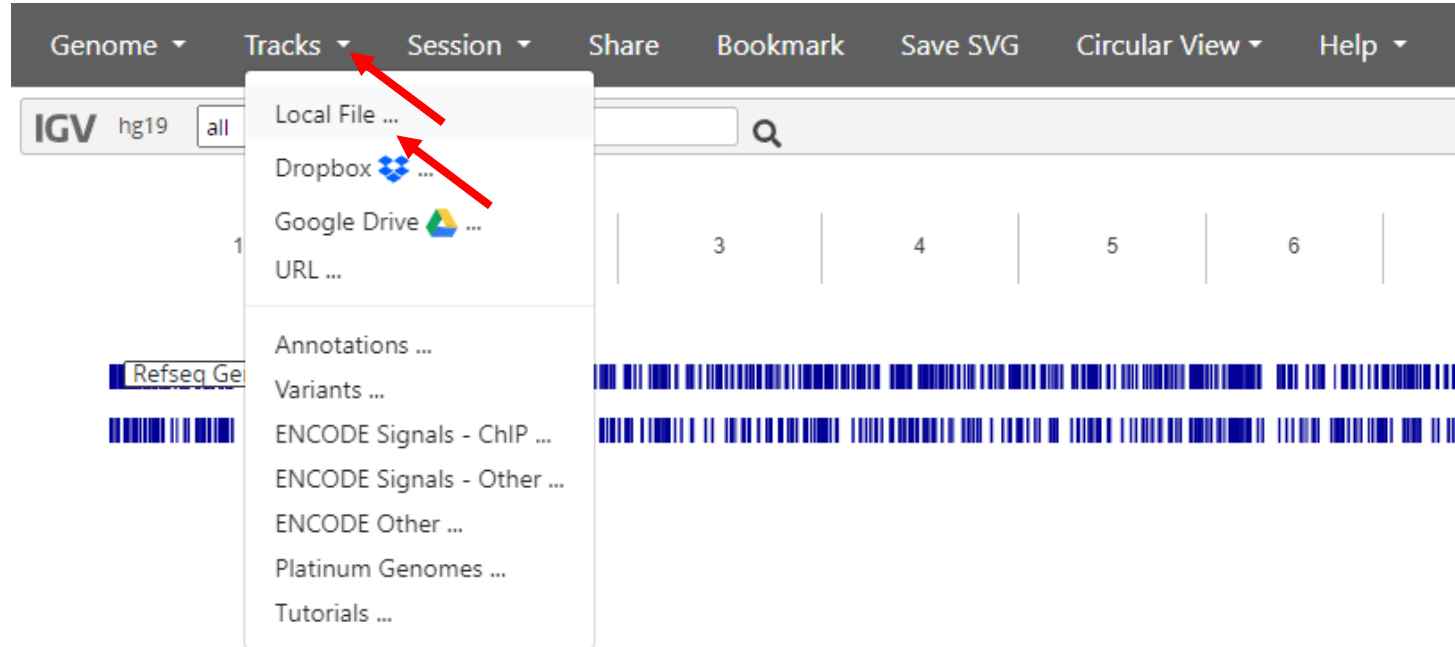
bam filename : ds.ERR309934_chr21.bam

bam index filename : ds.ERR309934_chr21.bam.bai

1. Choose the right version of reference genome

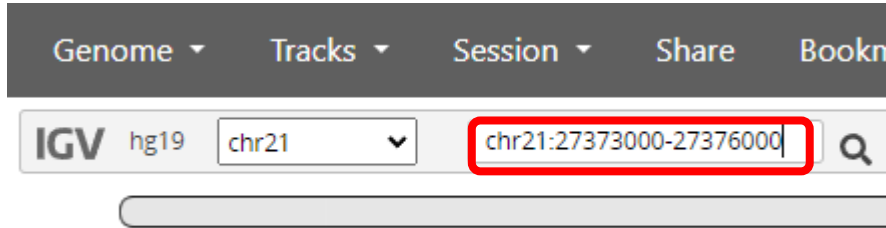


2. upload our data : both bam and bai files have to be simultaneously selected

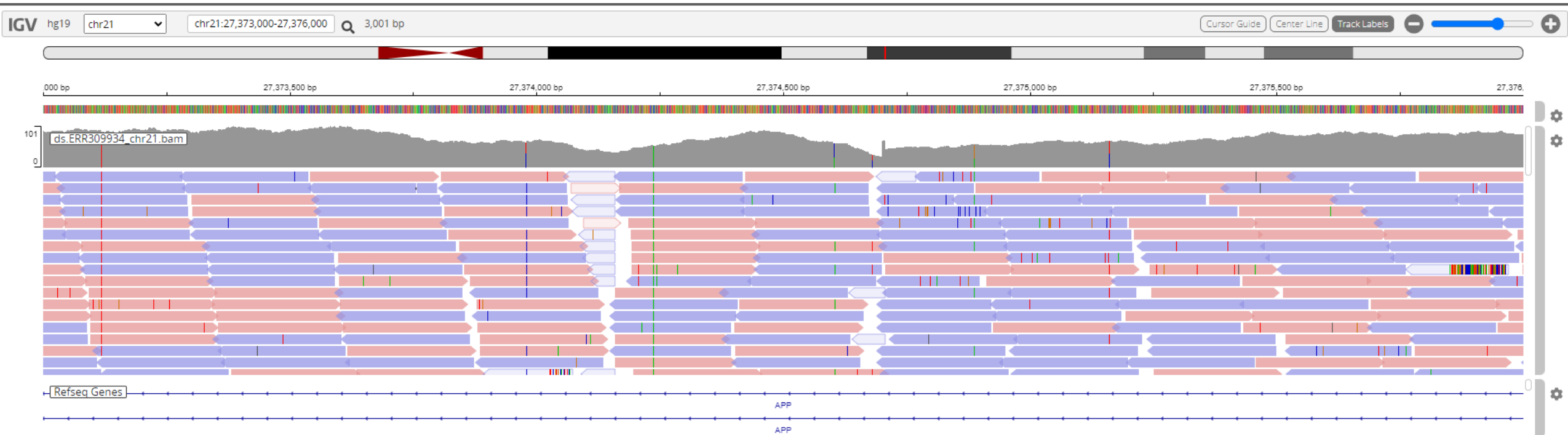


Select both by holding Ctrl key while selecting the two files


3. Specify region of interest : chr21 : 27372300-27372600



Carefully follow the nomenclature « chr : start – end »

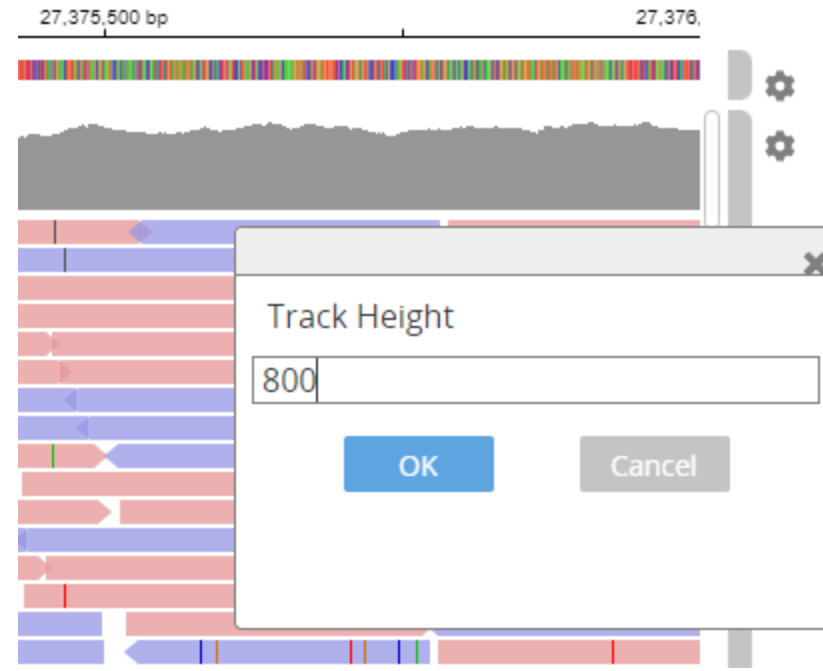


4. Set the parameters of the track



A screenshot of a genomic track interface. The track shows a reference sequence at the top, followed by a multi-colored alignment track, and a grey coverage track below it. A context menu is open over the alignment track, listing various options. Red arrows point to specific menu items: 'Set track height', 'pair orientation', and 'View as pairs'. The menu items include:

- Set track name
- Set track height
- Set track color
- Unset track color
- Set data range
- Autoscale
- Color by:
 - read strand
 - first-of-pair strand
 - pair orientation
 - insert size (TLEN)
 - pair orientation & insert size (TLEN)
 - tag
- Show Coverage
- Show Alignments
- Show all bases
- Show mismatches
- Show insertions
- Show soft clips
- View as pairs
- Add discordant pairs to circular view
- Display mode:
 - expand
 - squish
- Set visibility window
- Remove track



A screenshot of the same genomic track interface. A dialog box titled 'Track Height' is open, showing a text input field with the value '800' and 'OK' and 'Cancel' buttons. The track below shows the alignment track with red and blue bars representing read pairs.

Set the track height to
800

+

Set « Color by » to « pair orientation »

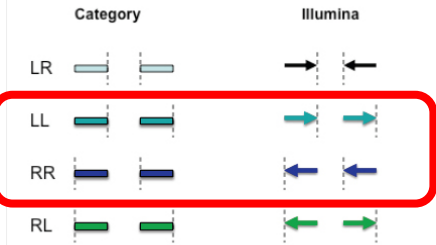
+

Set « View as pairs »

5. Observe the read pair orientations.

You can right-click exactly at this position and sort by base

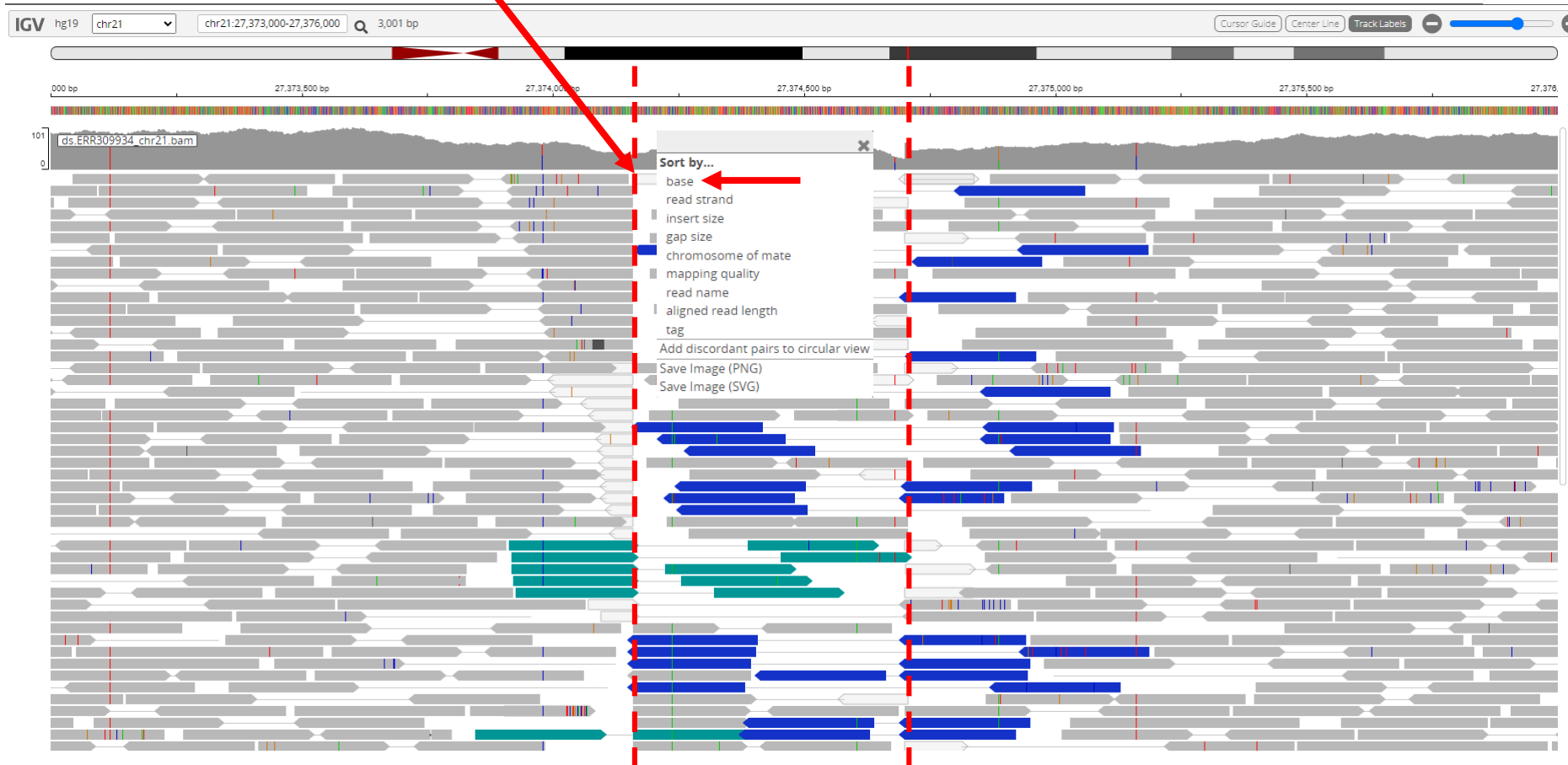
Interpretation of read pair orientations



LR Normal reads.
The reads are left and right (respectively) of the unsequenced part of the sequenced DNA fragment when aligned back to the reference genome.

LL,RR Implies inversion in sequenced DNA with respect to reference.

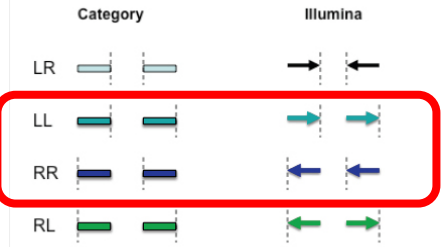
RL Implies duplication or translocation with respect to reference.



5. Observe the read pair orientations.

What kind of Structural Variant can it be ?

Interpretation of read pair orientations



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The reads are left and right (respectively) of the unsequenced part of the sequenced DNA fragment when aligned back to the reference genome.

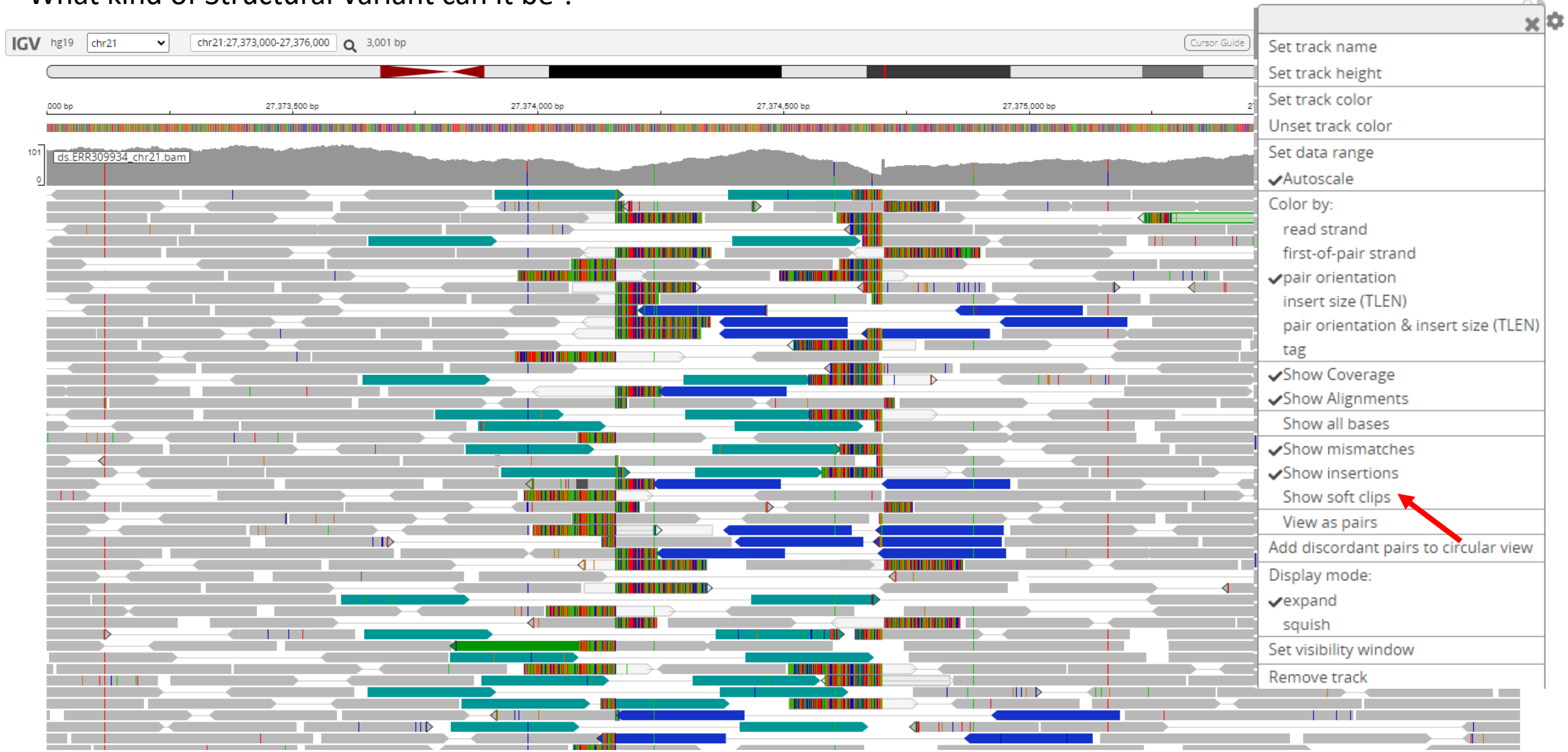
LL,RR Implies inversion in sequenced DNA with respect to reference.

RL Implies duplication or translocation with respect to reference.



6. Add other evidence by showing soft clipped bases (split reads)

What kind of Structural Variant can it be ?



CONCLUSION : both pair orientations and split reads are evidences of an inversion between positions chr21:27,374,147 and chr21:27,374,700

