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 tu use the web application : https://igv.org/app/

Dataset

Sample : NA19240

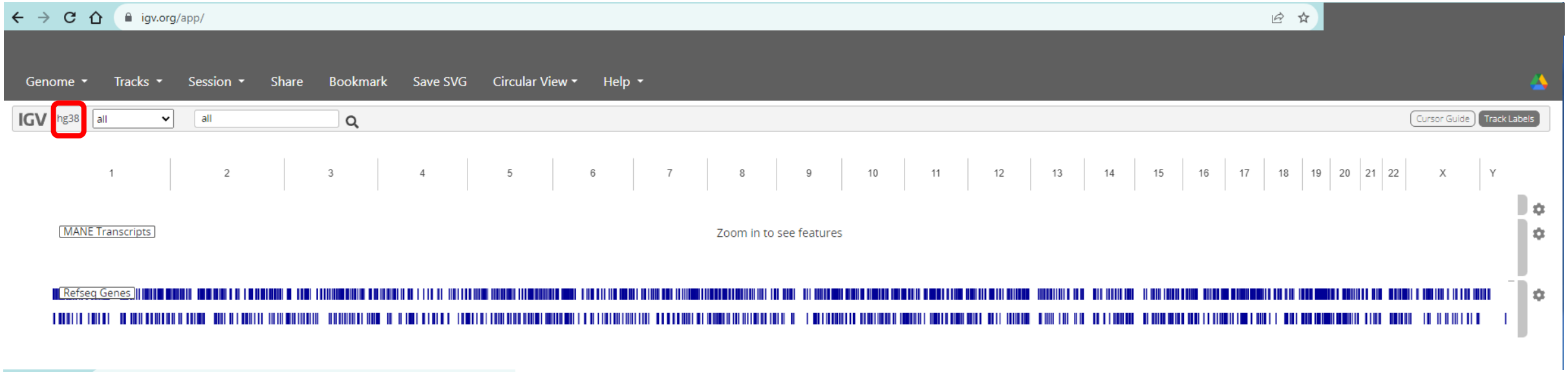
Data type : aligned reads in bam format restricted to a chromosome 21 region

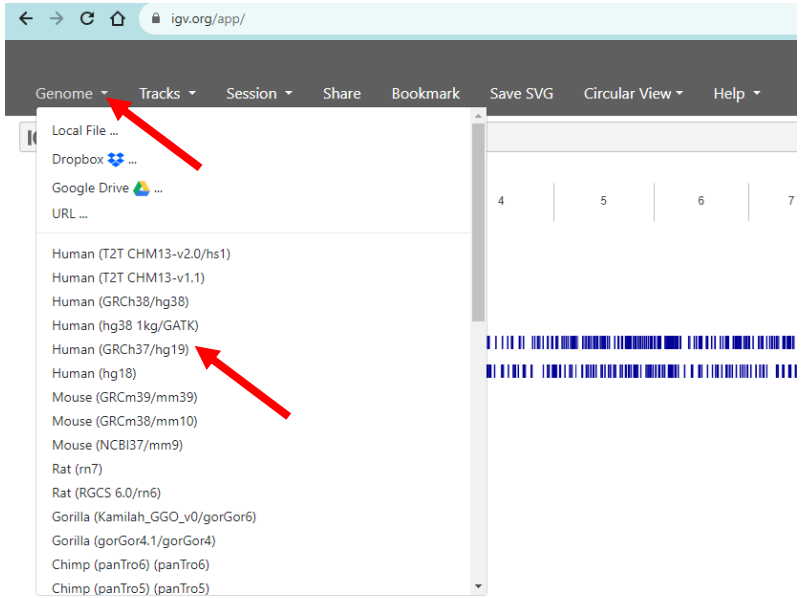
Human reference sequence version : hg19

bam filename : ds.ERR309934_chr21.bam

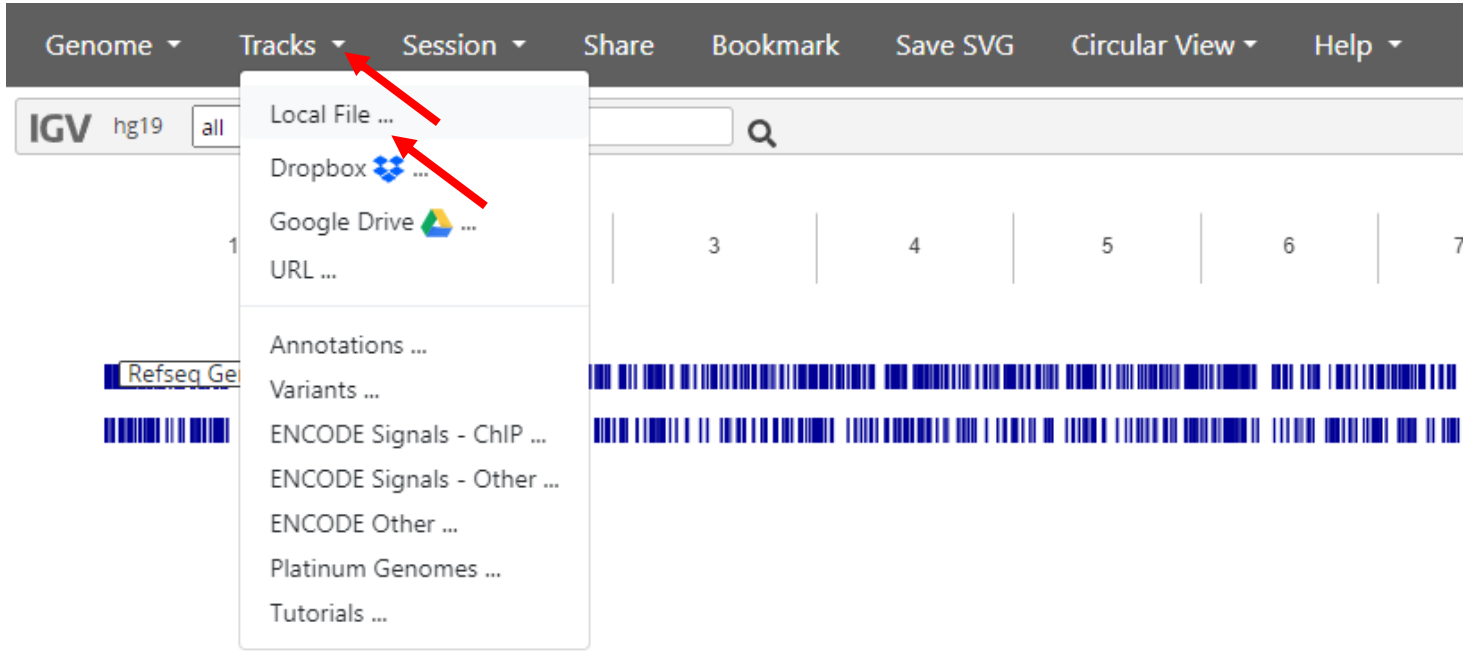
bam index filename : ds.ERR309934_chr21.bam.bai

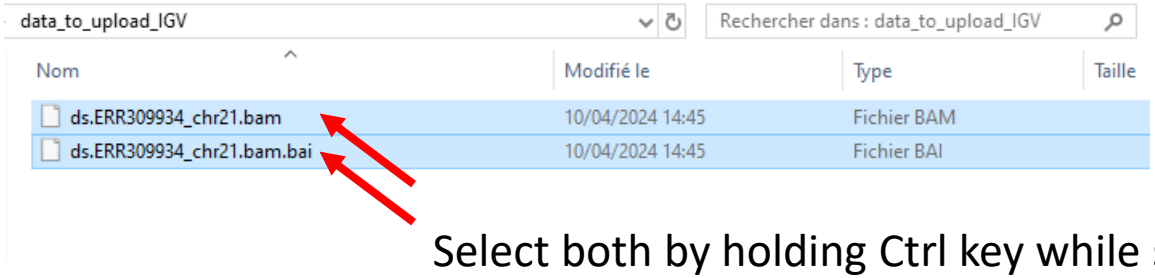
1. Choose the right version of reference genome





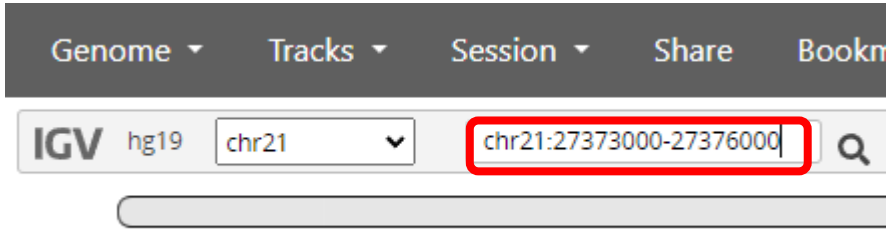
2. upload our data : both bam and bai files have to be simultaneously selected



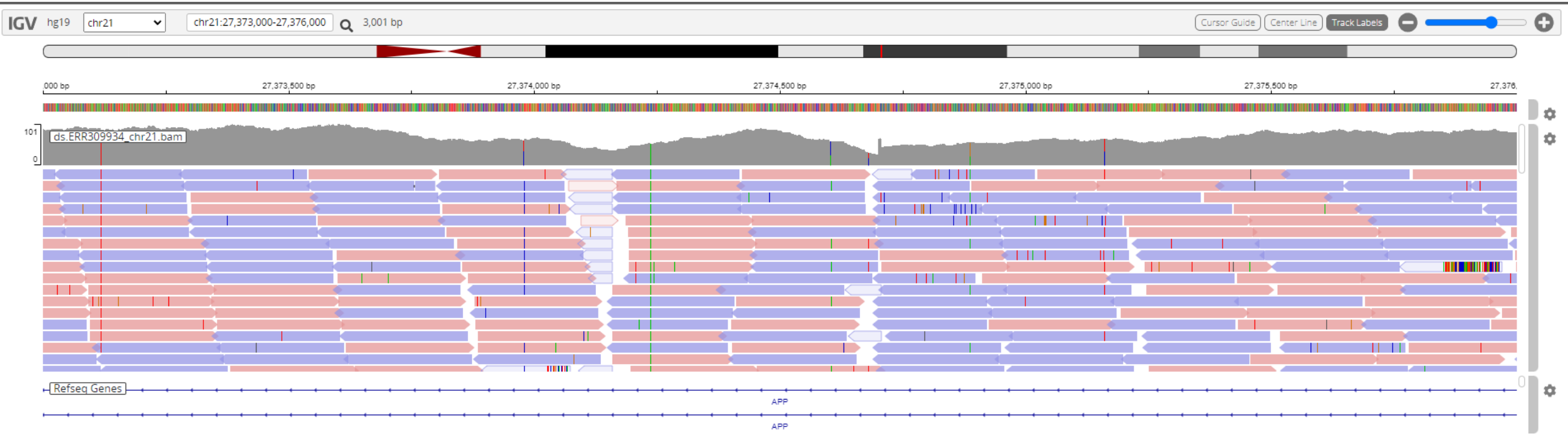


Select both by holding Ctrl key while selecting the two files

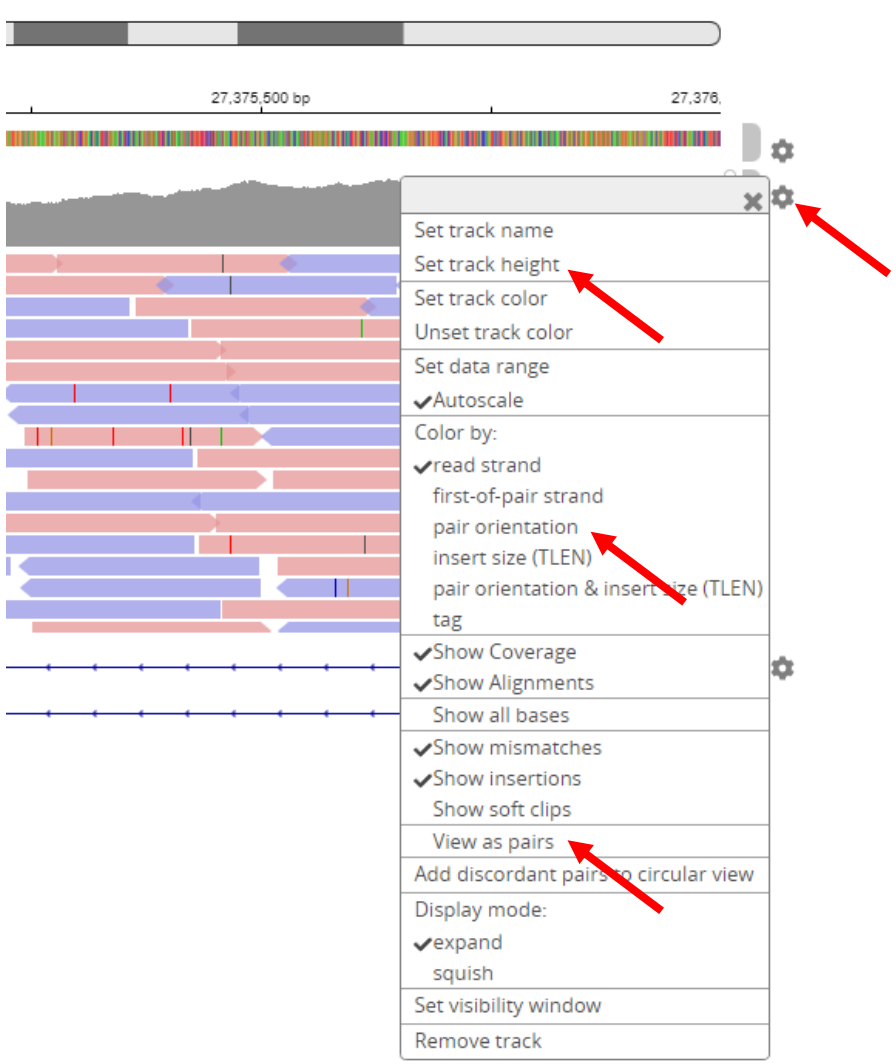
3. Specify region of interest : **chr21 : 27372300-27372600**

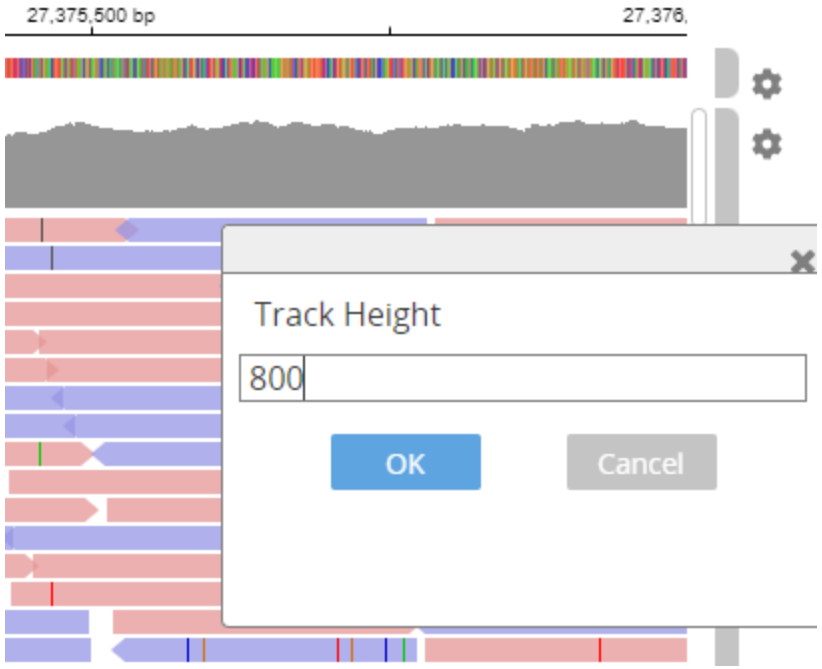


Carefully follow the nomeclature « chr : start – end »



4. Set the parameters of the track





Set the track height to 800

+

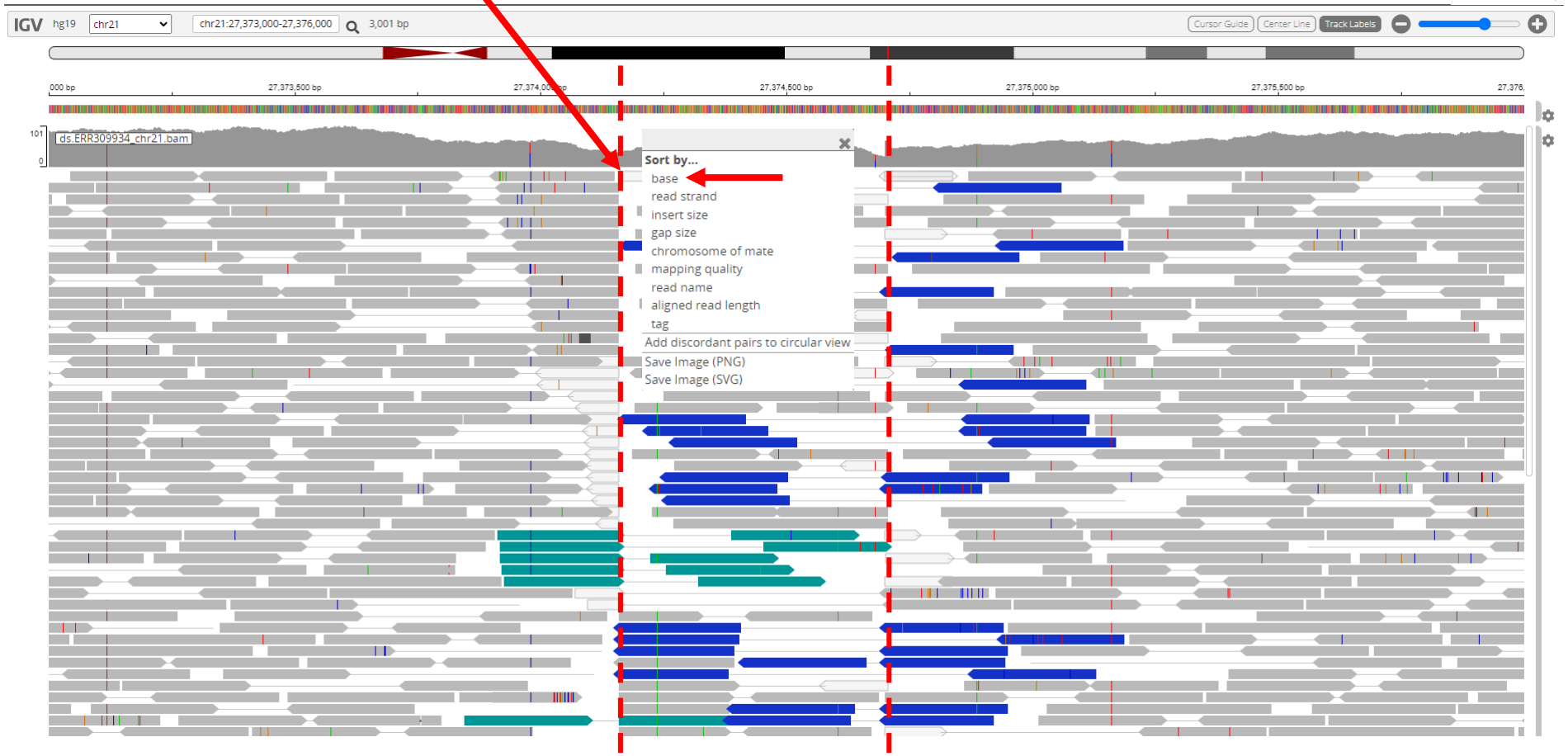
Set « Color by » to « pair orientation »

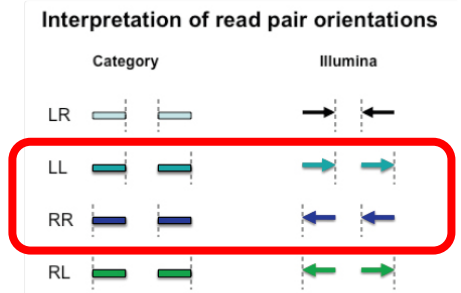
+

Set « View as pairs »

5. Observe the read pair orientations.

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

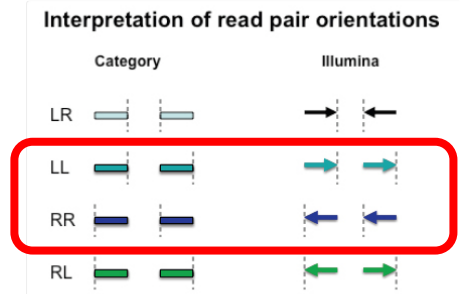




LR	Normal reads. The reads are left and right (respectively) of the unsequenced part of the sequenced DNA fragment when aligned back to the reference genome.
LL,RR	Implies inversion in sequenced DNA with respect to reference.
RL	Implies duplication or translocation with respect to reference.

5. Observe the read pair orientations.

What kind of Structural Variant can it be ?



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

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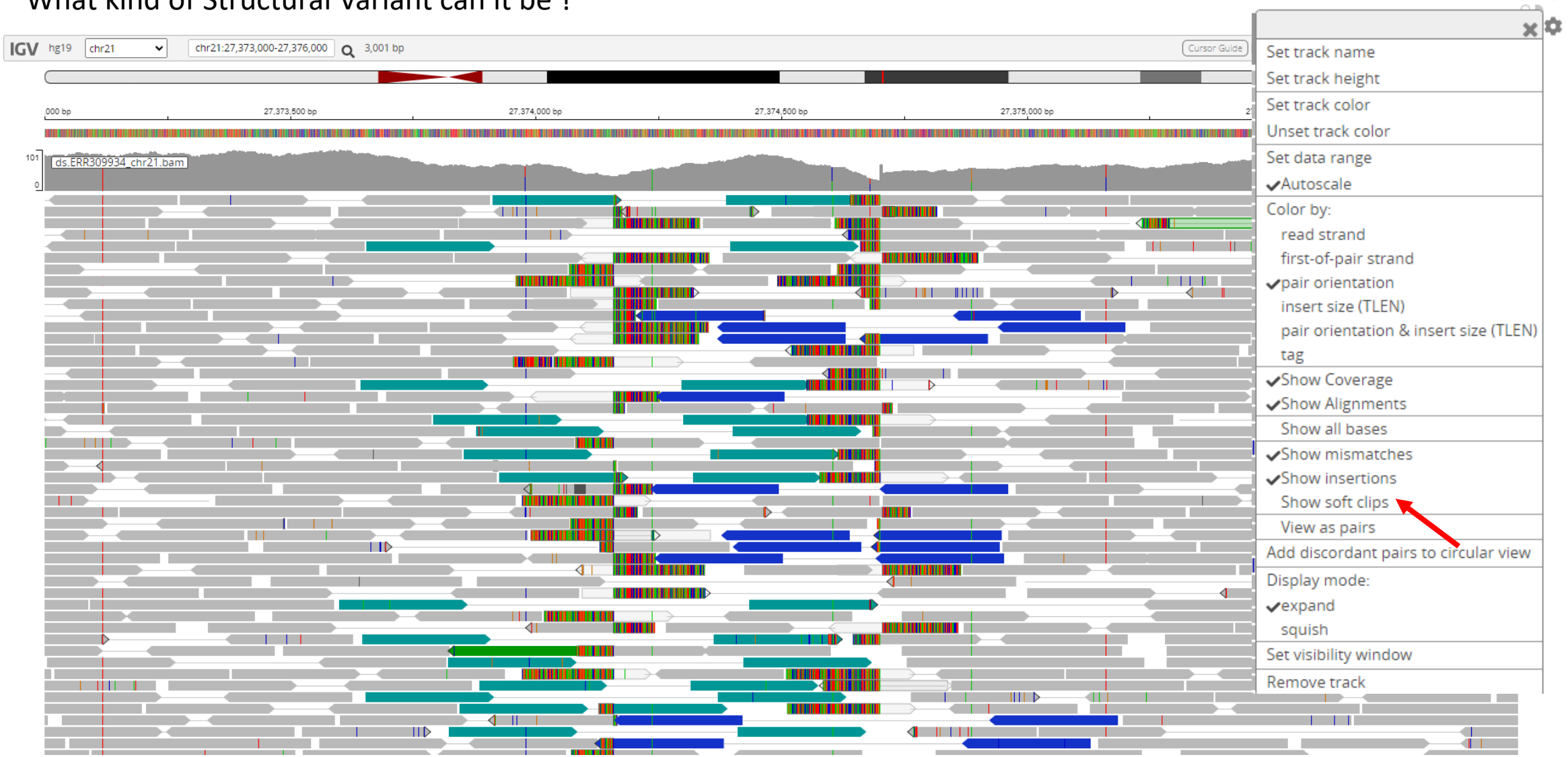

CONCLUSION : both pair orientations and split reads are evidences of an inversion between positions chr21:27,374,147 and chr21:27,374,700