

Visualization of mapped reads

Integrative Genomics Viewer (IGV)

Edinburgh Genomics

Edinburgh, UK

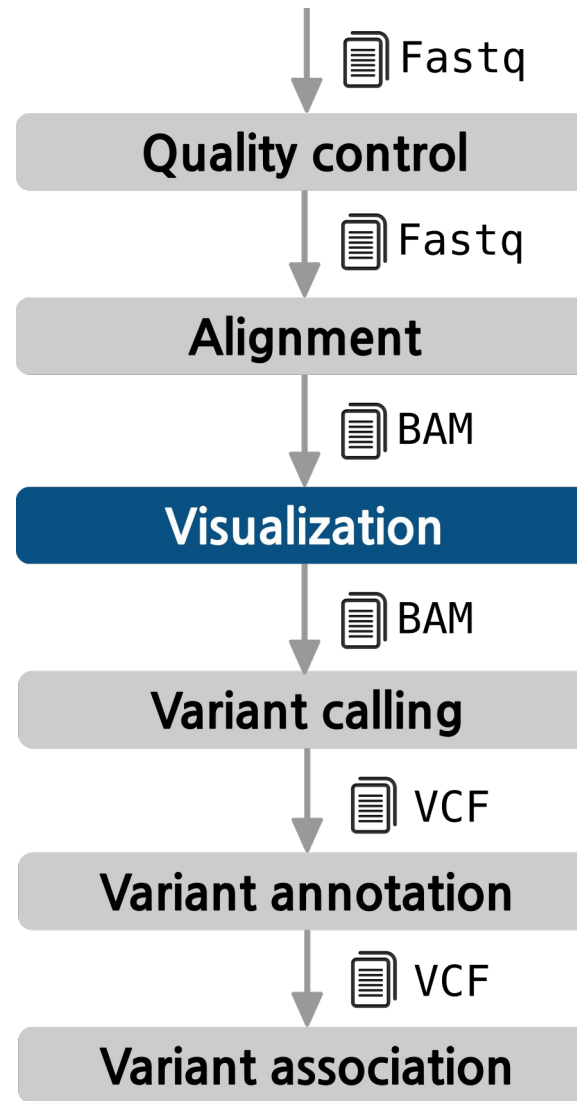
22nd October 2015

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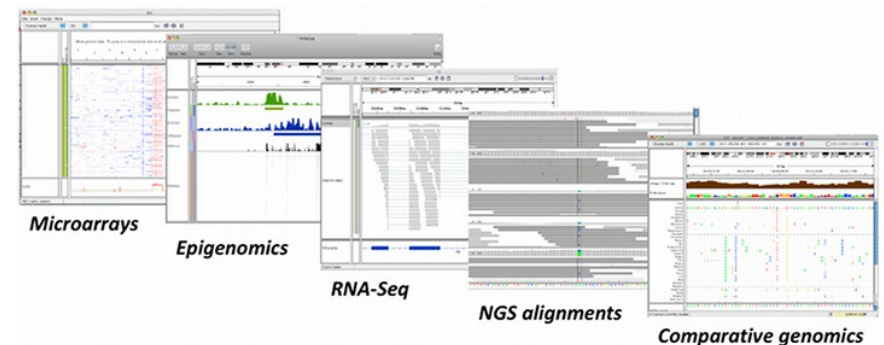
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The pipeline



Introduction

- **Large** quantities of genomic **data** (NGS, array based methods, etc)
- **Human interpretation** and judgment using visualization can help complex biological relationships
- Integrative Genomics Viewer (**IGV**)
 - **Integrate** different data types simultaneously
 - View **large datasets** easily
 - Faster navigation or browsing
 - Runs **locally** on your desktop
 - Used by large-scale projects
 - Open source and **freely available**



Helga Thorvaldsdóttir, James T. Robinson, and Jill P. Mesirov
Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration
Brief Bioinform (2013) 14 (2): 178-192

Data types

- Any data related to **genome coordinates**
- **Sample** annotations or attributes
- **Genome** annotations

Recommended file formats

| Source data | Recommended File Formats |
|-------------------------|---|
| Sequence alignment data | SAM (must be sorted/indexed) BAM (must be indexed) |
| Genome annotations | GFF or GFF3 format BED format |
| Variant data | VCF |
| Any numeric data | IGV format, TAB format WIG format |
| Gene expression data | GCT format RES format |

Indexing a BAM file

- BAM format: Binary **SAM** file → Reduces disk space and time
- BAM/SAM files need to be **indexed** (using **samtools**) → SAM files will be sorted by start position and indexed
- Index files must reside in the **same directory** as the BAM or SAM file

Indexing BAM file

```
samtools index example.bam
```

Registration and download

1. Be sure that **Java 6 or later** is installed on your machine
2. Go to the IGV website:

<http://www.broadinstitute.org/igv/home>

3. Click **Downloads** at the left panel
4. Click to register and fill the form

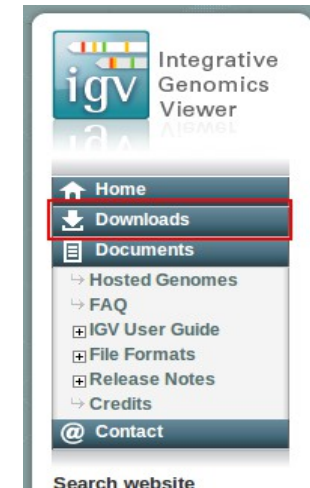
Log In

To use IGV, registration is required.
[Click here](#) to register.

If you have already registered for IGV please enter your registration email address below.

email address:

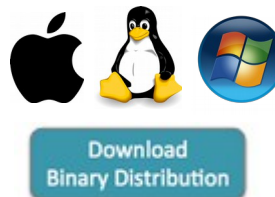
Login



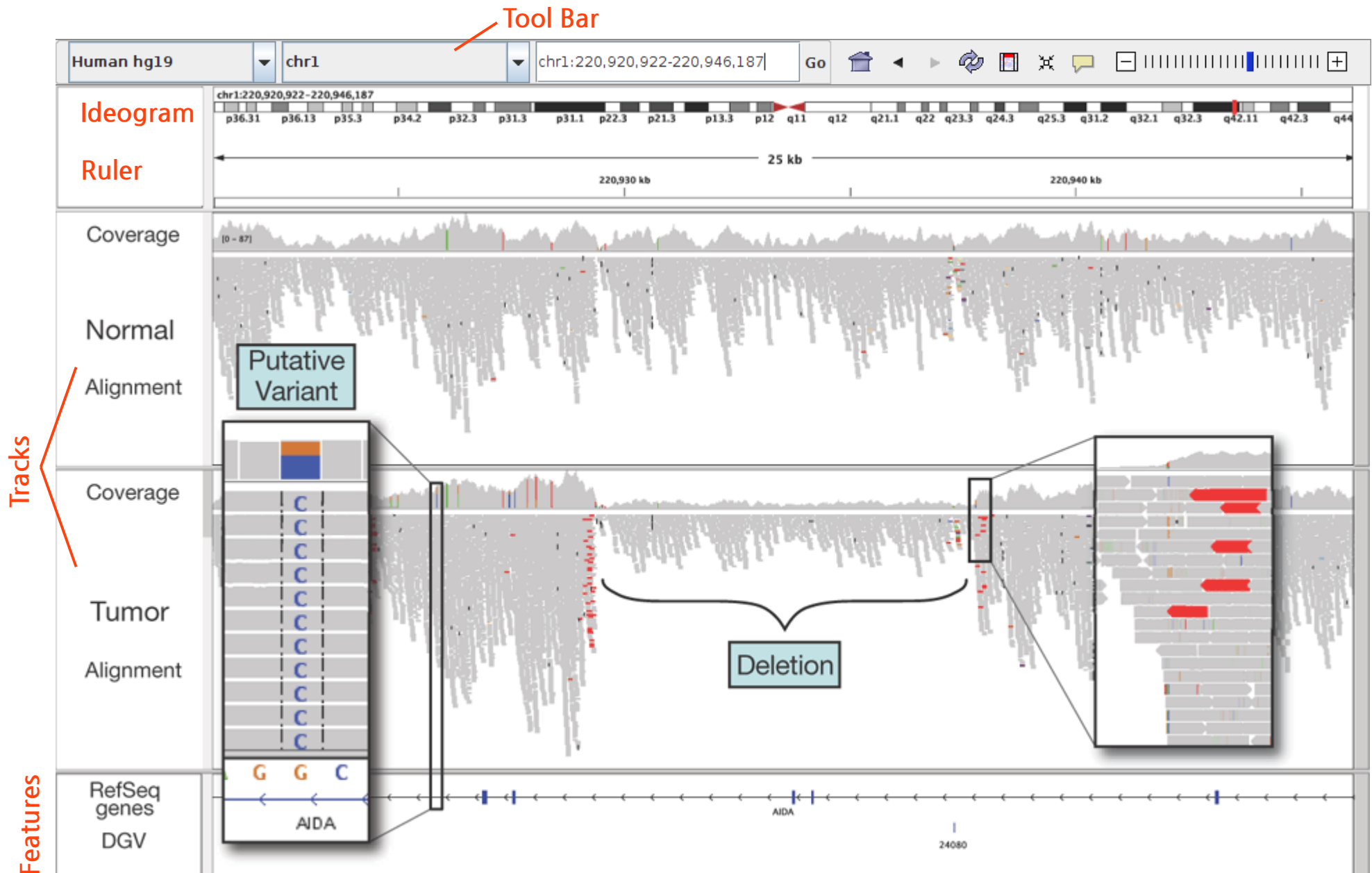
5. Download the most suitable file for your system



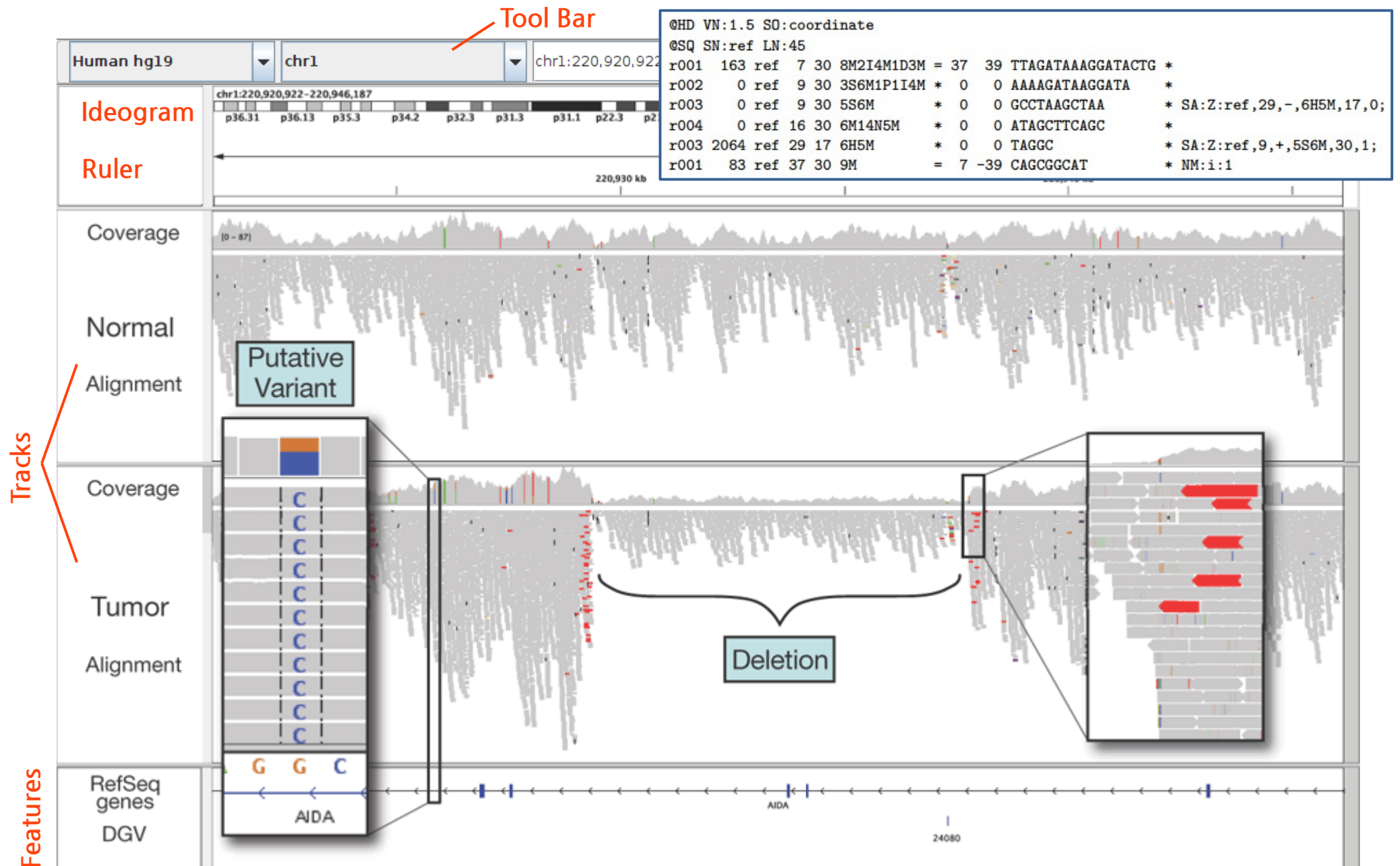
* Requires Java 7



IGV interface



IGV interface



Hands on!

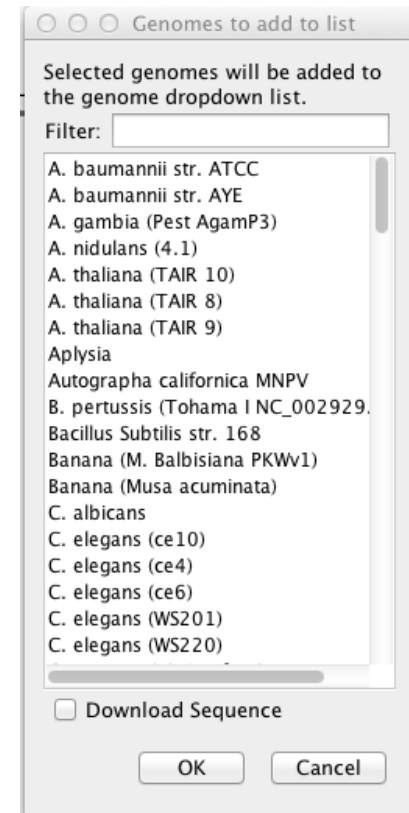
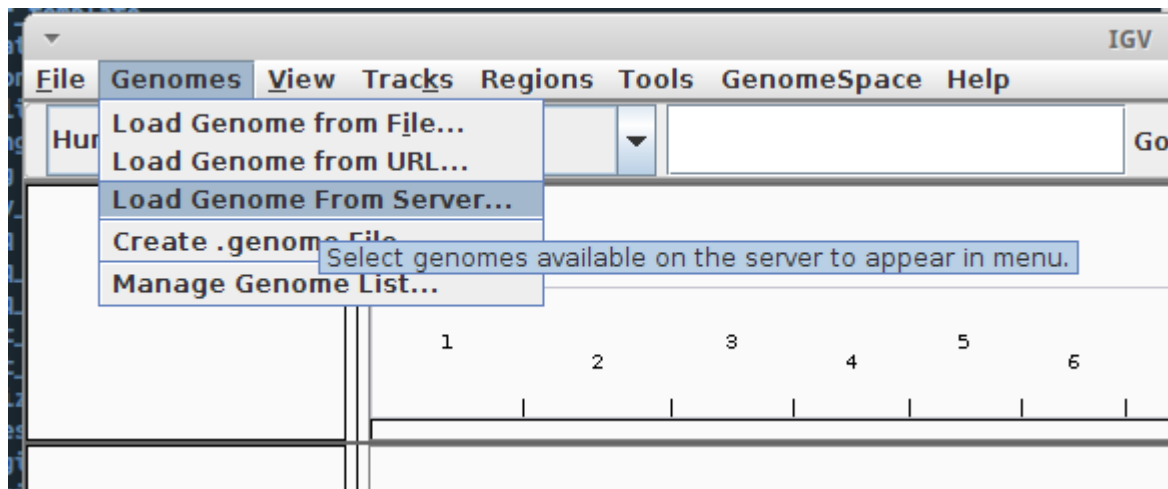
Download genome

Current version of IGV has "Human hg18"

IGV provides a number of genomes that are hosted on a server at the Broad Institute

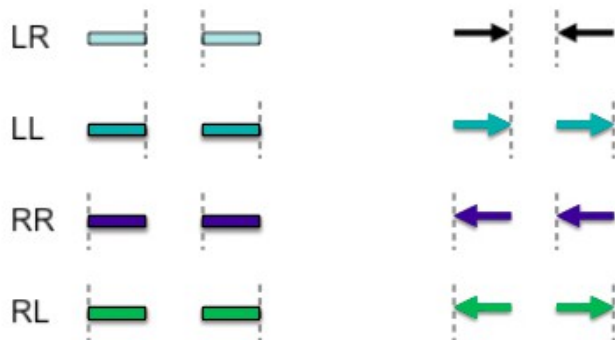
List of genomes hosted: <http://www.broadinstitute.org/software/igv/Genomes>

- Genomes → Load genome from server...
Select Human hg19



IGV: interpreting colored reads

Read pair orientation



- 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.

Insert size

Larger than expected (Deletion)



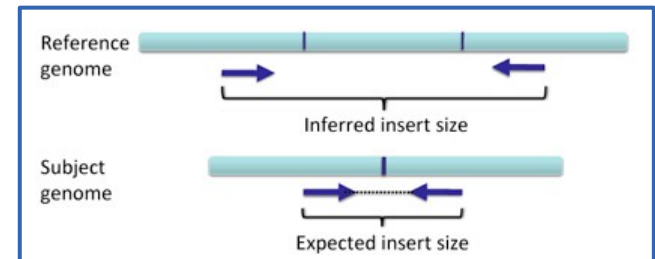
Smaller than expected (Insertion)



Mate of paired end reads that map to other chromosomes



Deletion



Insertion

