Introduction to NGS Visualization with the Integrative Genomics Viewer (IGV)





Integrative Genomics Viewer (IGV)



Desktop application for the interactive visual exploration of integrated genomic datasets





Features



With IGV you can...

- Explore large genomic datasets with an intuitive, easy-to-use interface.
- Integrate multiple data types with clinical and other sample information.
- View data from multiple sources:
 - local, remote, and "cloud-based".









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



Using IGV: The Basics





Using IGV: the basics



Hands-on exercise

- Launch IGV
- Select a reference genome
- Load data
- Navigate through the data





http://www.broadinstitute.org/igv





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 → FAQ ➡ IGV User Guide ➡ File Formats ➡ Release Notes 	Email Organization	
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Integrative Genomics

Viewer



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Select Human hg18

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Load data



Select File > Load from Server...





Load data





Screen layout

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Integrative Genomics

Viewer

1gv

Screen layout





Screen layout





Integrative Genomics





File formats and track types



- The file format defines the track type.
- The track type determines the display options



File formats and track types

- The **file format** defines the track type.
- The **track type** determines the display options
- IGV supports many different file formats.
 - BAM
 - BED
 - BedGraph
 - bigBed
 - bigWig
 - Birdsuite Files
 - broadPeak
 - CBS
 - CN
 - Cufflinks Files
 - Custom File Formats

 Merged BAM File
 - Cytoband
 - FASTA

- GCT
- genePred
- GFF
- GISTIC
- Goby
- GWAS
- IGV
- LOH
- MAF (Multiple Alignment Format)
- MAF (Mutation Annotation Format)
- MUT
- narrowPeak

- PSL
- RES
- SAM
 - Sample Information
- SEG
- SNP
- ТАВ
- TDF
- Track Line
- Type Line
- VCF
- WIG
- For current list see: www.broadinstitute.org/igv/FileFormats





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Navigate

X chr1:27,078,527

6 tracks





146M of 304M

Integrative





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Integrative Genomics















Navigate Genomics Viewer $\Theta \Theta \Theta$ IGV +Go 👚 🔺 🕨 Human hg18 chr1 \$ chr1:115,046,608-115,063,038 -p36.23 p36.12 p34.3 p33 p32.1 p31.1 p22.2 p21.2 p13.2 q32.2 q41 q44 q25.3 q31.3 q42.2 EU 115,048 kb 115,050 kb 115,052 kb 115,054 kb NAME DATA DATA [0 - 25] GM12878 H3K27ac [0 - 25] GM12878 H3K27me3 115,060 kb 115,062 kb [0 - 25] GM12878 H3K36me3 [0 - 25] GM12878 H3K4me1 Click on the last tick on the "railroad track" to zoom in to maximum resolution CSDE1 RefSeq genes NRAS chr1:115,051,417 148M of 317M 6 tracks



Integrative




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.





Genome annotation track







Annotation display mode



1. Features are drawn in a single row, by default

2. Expand the track using the popup menu





Annotation display mode



3. For a compact view of all variants use "Squished"











Integrative Genomics Viewer

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• Search box

Enter multiple loci or features in the search box

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• Regions > Gene Lists...

Select from a number of pre-defined gene lists, or Create your own persistent list





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

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

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Viewing NGS Data







Whole chromosome view

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NA19240 SLX (YRI daughter)			Zoom in to see alig	gnments.	
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4 tracks loaded	chr1:95,509,957				185M of 266M





Zoom in to view alignments





Coverage track now has more detail





Zoom in to see more detail





Zoom in to see more detail





Zoom in to see more detail













- Higher value (larger region) → requires more memory
- Low coverage files \rightarrow ok to use higher value
- Very deep coverage files → use lower value











Hands-on exercise

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















Select File > Load from Server...











É	IGV	File	Genomes	View	Tracks	Regions	Tools	GenomeSpace	Help
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Type "snp1" in the Search Box and click Go



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Integrative Genomics





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Integrative Genomics Viewer



Integrative Genomics

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Integrative Genomics Viewer

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Integrative Genomics





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Integrative Genomics Viewer

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Viewing SNPs

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Integrative Genomics

Viewer

igv

Viewing SNPs

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Integrative Genomics

Viewing Structural Events









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









Interpreting Insert Size





Interpreting inferred insert size



The "inferred insert size" can be used to detect structural variants, including:

- Deletions
- Insertions
- Inter-chromosomal rearrangements: (Undefined insert size)







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







Reference Genome







Reference Genome

Subject















Subject











Subject





Deletion







Deletion

















Inferred insert size is > expected value





Deletion







Deletion







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

Each end colored by chromosome of its mate











Rearrangement







Rearrangement







Interpreting Pair Orientations





Interpreting pair orientations



Orientation of paired reads can reveal structural events, including:

- inversions
- duplications
- translocations

Orientation is defined in terms of

- read strand, left vs right, and
- read order, first vs second







Reference genome

















































Integrative
Inversion









Inversion





"Left" side pair





"Right" side pair



Color by pair orientation



NA12878 WGS	
Rename Track Copy read details to clipboard	
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View as pairs Go to mate View mate region in split screen Set insert size options	first-of-pair strand read group sample tag bisulfite mode
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Inversion

















Hands-on exercise

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

http://www.illumina.com/science/data_library.ilmn











• Step 1: Tune settings for RNA.





Select View > Preferences					
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Integrative Genomics Viewer

Click Alignments tab

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Integrative Genomics Viewer





































































Viewing RNA splicing with Sashimi Plots

Reference: Katz Y, Wang ET, Silterra J, Schwartz S, Wong B, Mesirov JP, Airoldi EM, Burge, CB. *Sashimi plots: Quantitative visualization of RNA sequencing read alignments.* arXiv:1306.3466 [qbio.GN], 2013









Heart










































igvtools





igvtools



A set of utilities for preparing files for efficient display.

toTDF	 Converts sorted data file to a binary tiled data file (TDF). Supported file formats: .wig, .cn, .snp, .igv, .gct
count	 Computes average alignment or feature density over a specified window size across the genome. Supported file formats: .sam, .bam, .aligned, .sorted.txt, .bed
sort	 Sorts file by genomic start position. Supported file formats: .cn, .igv, .sam, .aligned, .bed.
index	 Creates an index file for alignment or feature file. Supported file formats: .sam, .aligned, .sorted.txt, .bed



igvtools



- Can be launched from the IGV user interface *File > Run igvtools...*
- Or run from the command line

Command Count					÷
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Genome hg19					Browse
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The **toTDF** utility converts large ASCII data files into tiled data format (.tdf) files.

TDF files have the following advantages:

- Data is indexed for efficient retrieval.
- Data is preprocessed for zoomed out views.
- TDF files are web friendly large data files can be shared over the web. Only small slices of the file are actually transferred as needed.



igvtools count



The **count** command is used to transform alignment files to read density TDF files, e.g. for ChIP-Seq, RNA-Seq, and similar alignment counting experiments.



Alignments

Alignments in bam/sam, .aligned, or bed format

Read Density

TDF format, indexed and optimized for fast retrieval at multiple resolution scales



igvtools sort



- Sorts IGV-supported genomic formats by start position.
- The index command requires sorted files.

Example:

igvtools sort -m 1000000 –t ~/myTmpDir inputFile.sam outputFile.sorted.sam

• Uses combination of memory and disk to handle large files.

-m = maximum # of lines to hold in memory. When this number is exceeded a temporary file is created.

-t = directory used to create temporary files during sorting.





Creates an index file for viewing large files in bed, gff, or vcf formats. An index is optional for bed or gff files, but required for vcf files.

An alternative indexing tool is "tabix". Tabix both compresses and indexes genomic files. IGV can read either type of index (igvtools or tabix).

Example: igvtools index myFeatures.bed

The index file must remain in the same directory as the input file





Hands-on exercise

• Compute alignment coverage from a BAM file using igvtools count command.

Data source

Illumina BodyMap





Download data files required for this exercise from: <u>ftp://ftp.broadinstitute.org/pub/igv/CSH_2013/files.zip</u>

Files included in the zip: heart.bodyMap.bam heart.bodyMap.bam.bai sacCer3.fa (used in <u>next</u> exercise)



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Integrative Genomics

Viewer

1gv



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Integrative Genomics

Viewer

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IGV doesn't host the genome you need?

Use any genome you want, if you have the sequence in FASTA format.

Optionally, package genome annotations with the sequence.





Hands-on exercise







Integrative Genomics



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Integrative Genomics
Acknowledgments



IGV Team

Jim Robinson, Jacob Silterra, Helga Thorvaldsdóttir, Jill Mesirov (PI)

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- IGV participates in GenomeSpace http://genomespace.org/, which is funded by the the National Human Genome Research Institute (NHGRI) http://www.genome.gov/



For further information and help:

http://www.broadinstitute.org/igv

http://groups.google.com/group/igv-help

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