

基生研ゲノムインフォマティクス・トレーニングコース 2017春  
RNA-seq入門 – NGSの基礎からde novo解析まで –  
実践編：RNA-seq解析パイプライン  
2017.03.09–2017.03.10

# NGS基本ツールIGV

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生物機能解析センター  
山口勝司

## データ可視化ツール・IGVの紹介・実習

The screenshot shows the IGV website homepage. At the top, there's a navigation bar with links to Home, Downloads, Documents, Hosted Genomes, FAQ, IGV User Guide, File Formats, Release Notes, IGV for iPad, and Contact. Below this is a search bar and the Broad Institute logo. The main content area features a large image of the IGV interface with the text 'Integrative Genomics Viewer'. To the left of this image is an 'Overview' section describing IGV as a high-performance visualization tool for interactive exploration of large, integrated genomic datasets. To the right is a 'Citing IGV' section with a list of publications. Below the main image is a 'Downloads' section with a download button and a 'Funding' section with text about the tool's development and funding sources. At the bottom, there are logos for the National Cancer Institute, National Institutes of Health, and the Genomics Research Institute.

Home

Integrative Genomics Viewer

Home  
Downloads  
Documents  
Hosted Genomes  
FAQ  
IGV User Guide  
File Formats  
Release Notes  
IGV for iPad  
Contact

search  
Broad Home  
Cancer Program  
BROAD  
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Search website

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.

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. *Nature Biotechnology* 29, 24–26 (2011)  
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).

Downloads

Please [register](#) to download IGV. After registering, you can log in at any time using your email address.

Funding

Development of IGV is made possible by funding from the National Cancer Institute, the National Institutes of Health, the National Cancer Research Institute, and the Sloan Cancer Consortium.  
IGV participates in the GenomeSpace initiative, which is funded by the National Human Genome Research Institute.

<https://www.broadinstitute.org/igv/>

# なぜIGVを取り上げるか

## データ可視化ツール

- ・自分のパソコン(ローカル環境)にインストールして使うタイプ
- ・サーバーに構築して、ネットワークで使うタイプ

### The JBrowse Genome Browser

JBrowse is a fast, embeddable genome browser built completely with JavaScript and HTML5, with optional run-once data formatting tools written in Perl.

### Featured Post

[Exploring structural variation using JBrowse](#) by Richard Pinkers

Latest Release – [JBrowse 1.11.6](#)



コミュニティに広く利用、あるいはウェブ公開を目的とするには良いが、ネットワーク・情報セキュリティの高度な知識も要求される。

より大容量なデータに対応できる。

管理者的な人がいて、その人がやってくれるなら、これも良いが。

もっとお手軽なものとしてIGVを紹介

## 可視化ツールに求められるものは何か

膨大なデータを如何に直感的に理解できるようにするか  
sortや絞り込みができる表データと対比双壁

- ・配列、GC ratio、遺伝子情報
- ・遺伝子発現情報
- ・SNPの位置情報・頻度情報
- ・様々なデータの精度情報

レファレンス配列 / gene model / gene annotationとNGSデータを並べて比較  
複数のデータセットを並べて比較

色々なデータ(variant, 発現, ChIP, BSseq等々)を、様々なスケールで  
比較・統合的に解釈できるようにしたい

ゲノムviewerに自分のデータを乗せ、  
統合的直感的に解釈できること

# 可視化ツールをどう選ぶか

選択の基準

genome data viewing に求められるもの  
取捨選択の基準

1. 無料 / 有料 / 基本無料
2. 個人的レベルの使用 / コミュニティレベルの使用
3. 見るだけ/自分から色々工夫
4. アクセスのしやすさ・使いやすさ  
導入に必要なコンピュータスペック  
マニュアルは分かりやすいか  
情報の多さ  
利用の簡便さ  
使っている人が近くににいるか

## Integrative Genomics Viewer(IGV)

### お手軽ツール

- ・アカデミックウェアで無料
- ・コミュニティでの利用者が多いから、情報も多い
- ・javaのプログラムなので、オールプラットフォーム対応
- ・マニュアルは親切、サンプルデータがある
- ・WEBサーバーではなく、PCレベルでできる
- ・データ閲覧環境の共有が可能

誰もが簡単に使えるものが良い。



- Home
- Downloads
- Documents
- Hosted Genomes
- FAQ
- IGV User Guide**
- File Formats
- Release Notes
- IGV for iPad
- Credits
- Contact

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**Overview**

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Helga Thorvaldsdottir, James T. Robinson, Jill P. Mesirov, *Integrative Genomics Viewer (IGV): high-performance genomics data visualization and exploration*. *Briefings in Bioinformatics* 14, 178–192 (2013).

**Downloads**

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IGV participates in the GenomeSpace initiative, which is funded by the National Human Genome Research Institute.



- Home
- Downloads
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- IGV User Guide**
- User Interface
- Starting IGV
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- Loading Data and Attributes
- Viewing Data
- Viewing Alignments
- Gene List View
- Regions of Interest
- Sample Attributes
- Sorting, Grouping, and Filtering
- Saving and Restoring Sessions
- Server Configuration
- External Control of IGV
- igvtools
- Motif Finder
- File Formats
- Release Notes
- IGV for iPad
- Credits
- Contact

#### Home » IGV User Guide

### IGV User Guide

This guide describes the Integrative Genomics Viewer (IGV).

- To start IGV, go to the IGV downloads page: <http://www.broadinstitute.org/igv/download>.

Look at a printer-friendly HTML version of the whole User Guide.

- User Interface
  - Starting IGV
  - Navigating
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  - Loading Data and Attributes
  - Viewing Data
  - Viewing Alignments
  - Gene List View
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  - Sample Attributes
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  - Saving and Restoring Sessions
  - Server Configuration
  - External Control of IGV
  - igvtools

[User Interface »](#)





## Downloads

## Integrative Genomics Viewer (IGV) (Version 2.3)

## Install IGV

### Options for installing and running JGV:

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 (GV from the command line).

**Note:** JGV 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

## Windows

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 "gv.bat", it might appear as just "gv" 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 "jnp" file, then right-clicking on the saved "jnp" file and select "Open With > Java Web Start".

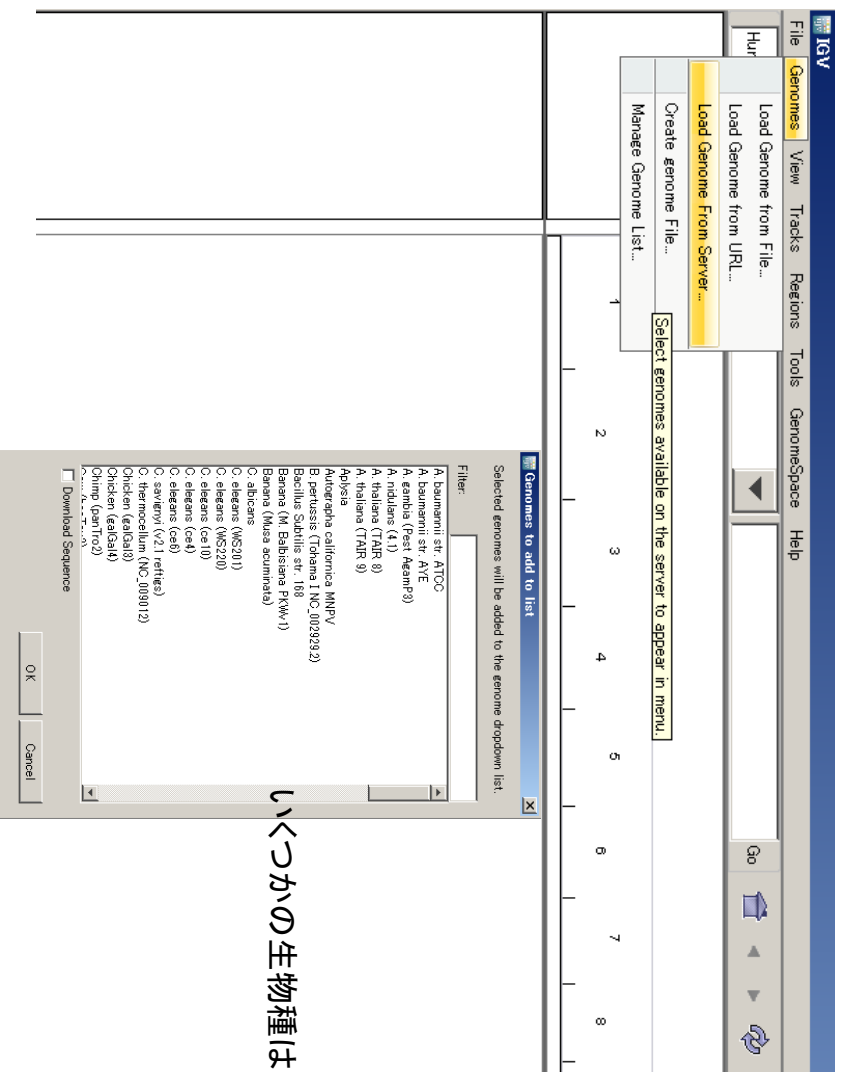
Chrome: Chrome does not automatically launch the Java Web Start files by default. Instead, the launch buttons below will download a "jnp" file. This should appear in the lower left corner of the browser. Double-click the downloaded file to run, or if on a Mac right-click and select "Open With > Java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

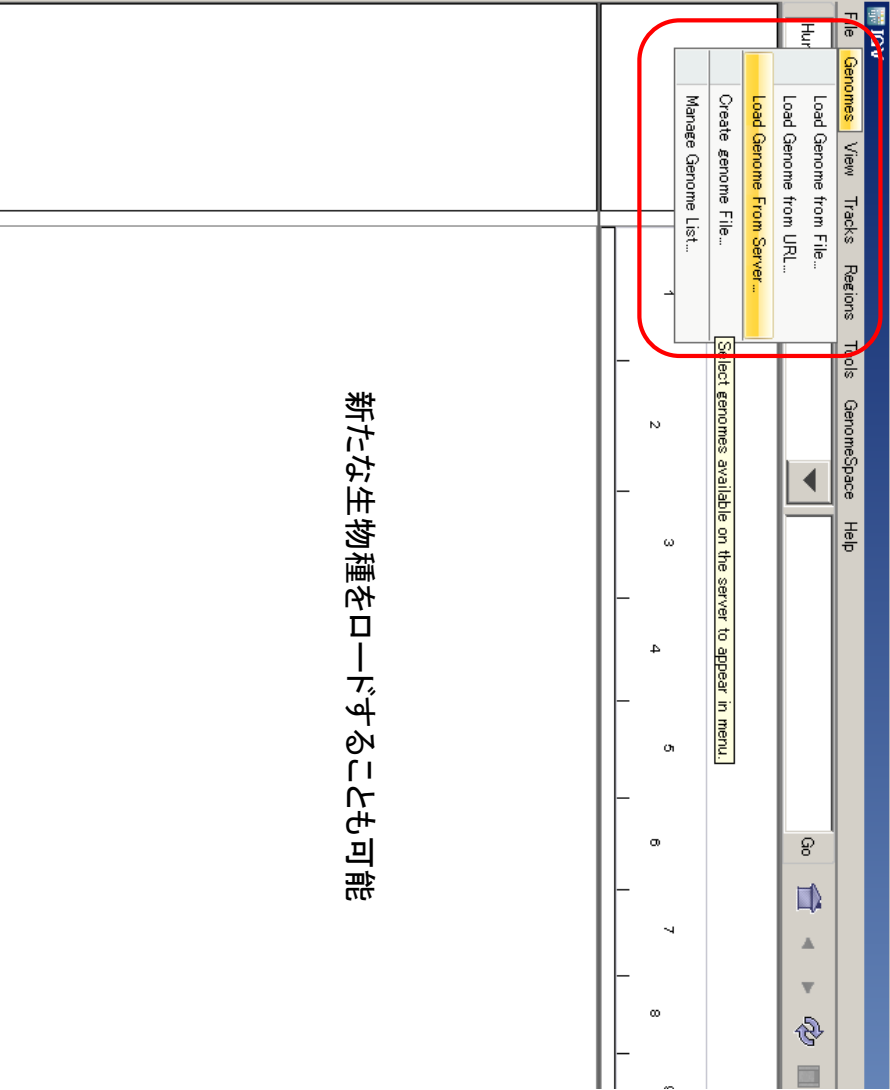
<p> <b>Launch</b></p> <p>Launch with 750 MB</p>	<p> <b>Launch</b></p> <p>Launch with 1.2 GB</p> <p>Maximum usable memory for Windows OS with 32-bit Java</p>	<p> <b>Launch</b></p> <p>Launch with 2 GB</p> <p>Maximum usable memory for 32-bit Mac OS</p>	<p> <b>Launch</b></p> <p>Launch with 10 GB</p> <p>For large memory machines with 64-bit Java</p>
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## 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

いくつかの生物種は登録されている



新たな生物種をロードすることも可能

ゲノムViewerなので次世代DNAシーケンサーのデータに限定されない。  
マイクロアレイの結果や、ゲノムアノテーションの情報も随時表示できる。

対応するファイル形式に応じて、表示方法が決まる。

File Formats

- File Extension Identifies Format
- Recommended File Formats
- BAM
- BED
- BedGraph
- bigBed
- bigWig
- Birdsuite Files
- broadPeak
- CBS
- CN
- Custom File Formats
- Cytoband
- FASTA
- GCT
- genePred
- gtfGtf
- gistic
- GxWx
- GWAS
- IGV
- LOH
- MAF (Multiple Alignment Format)
- MAF (Mutation Annotation Format)
- Merged BAM File
- MUT
- narrowPeak
- PSL
- RES
- SAM
- Sample Information
- SEG
- SNP
- TAB
- TDF
- Track Line
- Type Line
- VCF
- VIG
- WIG
- cytrom sizes

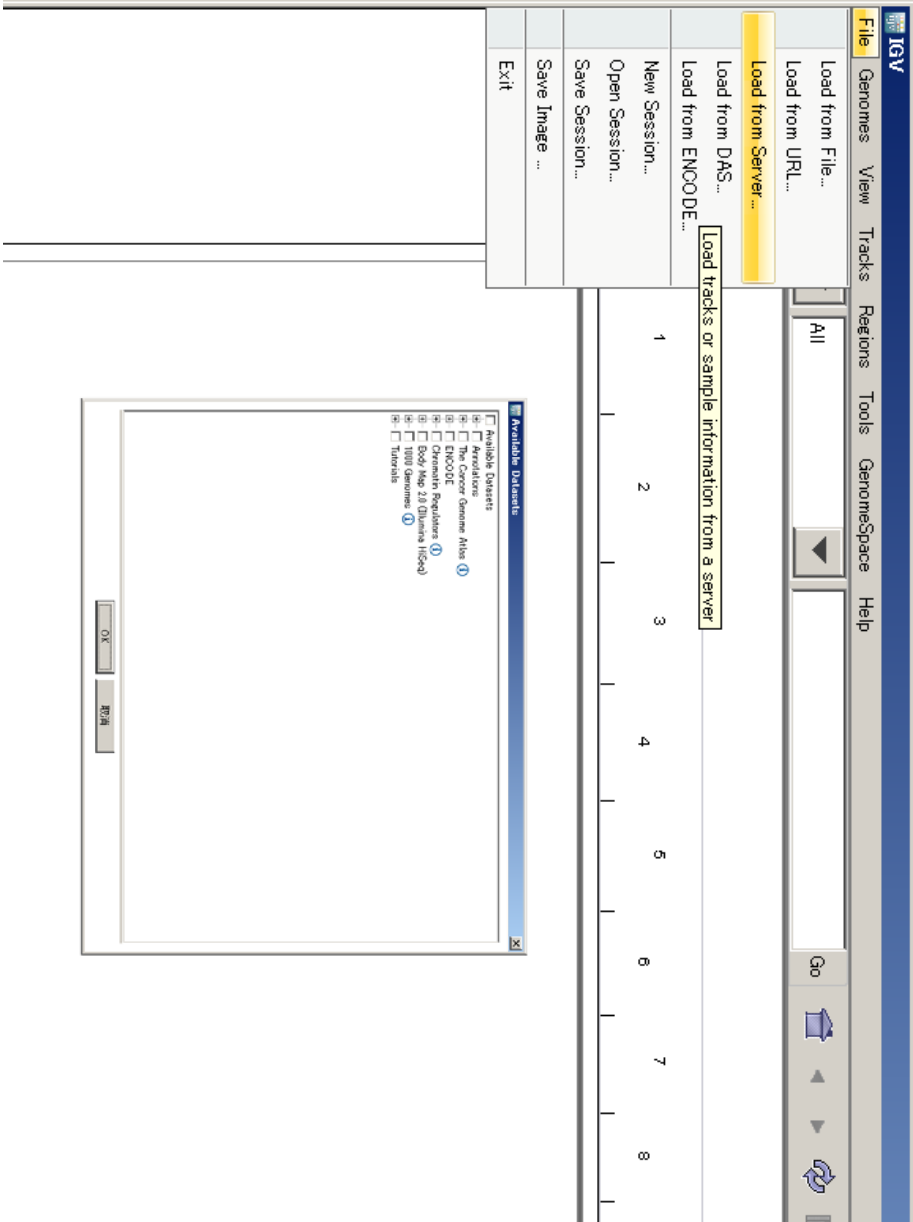
File Formats

IGV supports a number of different file formats for experimental data and genome annotations. For a complete list of supported formats see <http://www.broadinstitute.org/igv/FileFormats>. The following table shows the recommended file formats for a number of common data types.

Source Data	Recommended File Formats
ChIP-Seq, RNA-Seq	WIG, TDF
Copy number	CN, SNP, TDF, canary_calls (Birdsuite)
Gene expression data	GCT, RES, TDF
Genome annotations	GFF, BED, GTF, PSL, UCSC table format
GIS TIC data	GIS TIC
LOH data	LOH, TDF
Mutation data	MUT, MAF
Variant calls	VCF
RNAi data	GCT
Segmented data	SEG, CBS
Sequence alignment data	BAM, SAM, PSL
Any numeric data	IGV, WIG, TDF
Sample metatadata	Tab-delimited sample info file



# 公開情報のviewerとして



## その他の便利機能

### セッションの保存

表示しているデータの読み込み状況を、それごと保存。  
セッションをロードすることで、意図した画面を表示できる。  
データセットが揃っていること、フォルダー構造が同一である必要がある。

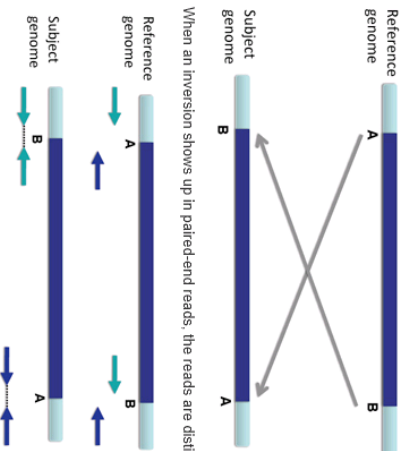
### バッチ処理

重要領域の画面スナップショットを自動で取ったりできる。

```
new
load myfile.bam
snapshotDirectory mySnapshotDirectory
genome hg18
goto chr1:65,289,335-65,309,335
sort position
collapse
snapshot
goto chr1:113,144,120-113,164,120
collapse
snapshot
```

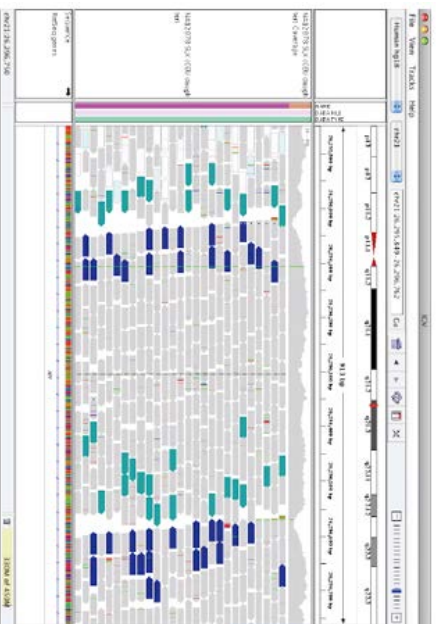
## Inversions

An inversion is a large section of DNA that is reversed in the subject genome compared to the reference genome.



When an inversion shows up in paired-end reads, the reads are distinctively variant from the reference genome.

This appears in IGV as shown below.






## Interpreting Color by Insert Size

The inferred insert size can be used to detect structural variants, such as:

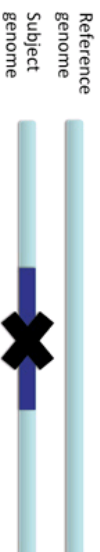
- deletions
- insertions
- inter-chromosomal rearrangements

IGV uses color coding to flag anomalous insert sizes. When you select Color alignments>by insert size in the popup menu, the default coloring scheme is:

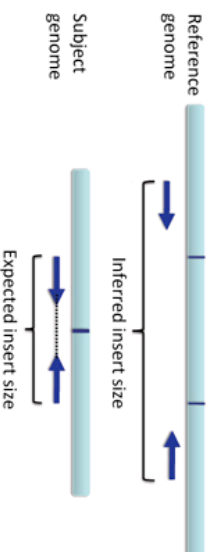
-  for an insert that is larger than expected
-  for an insert that is smaller than expected
-  for paired end reads that are coded by the chromosome on which their mates can be found

## Deletions

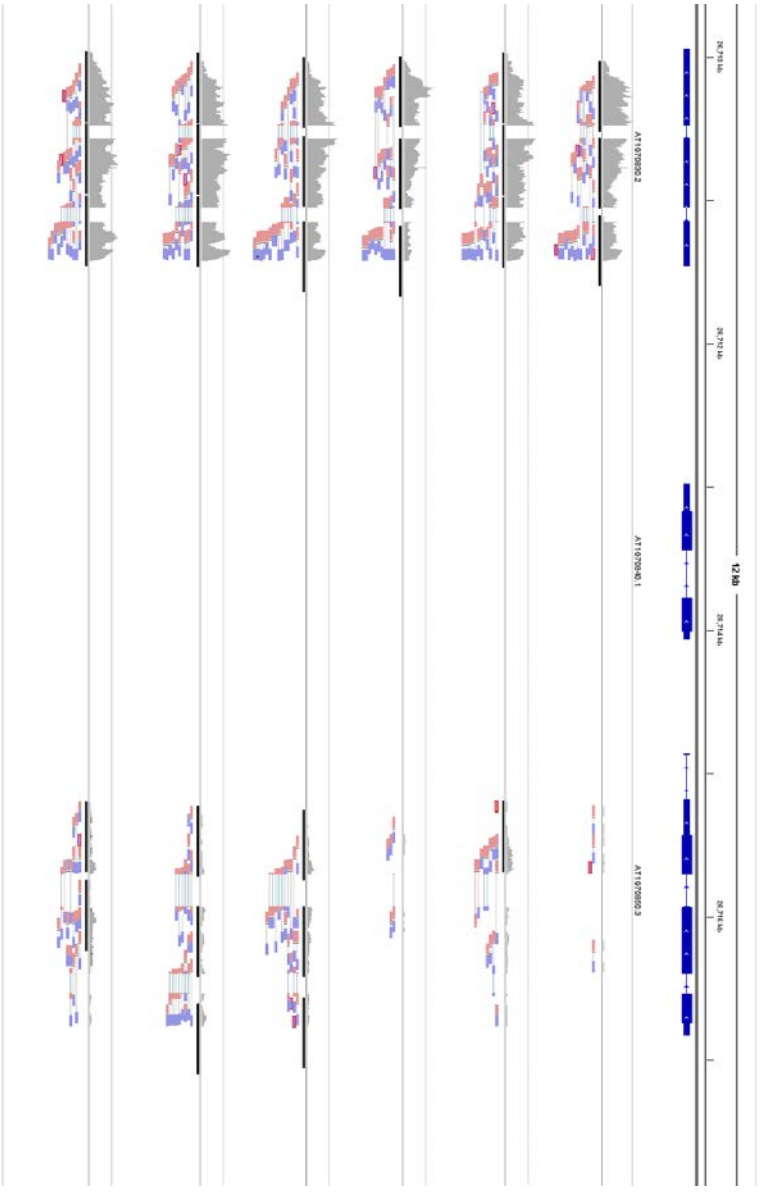
A deletion is a large section of DNA that is absent in the subject genome compared to the reference genome.



The "expected" insert size is the insert size obtained in sequencing the subject genome. The "inferred" insert size is the insert size that would result in the reference genome, assuming the same pair of reads.

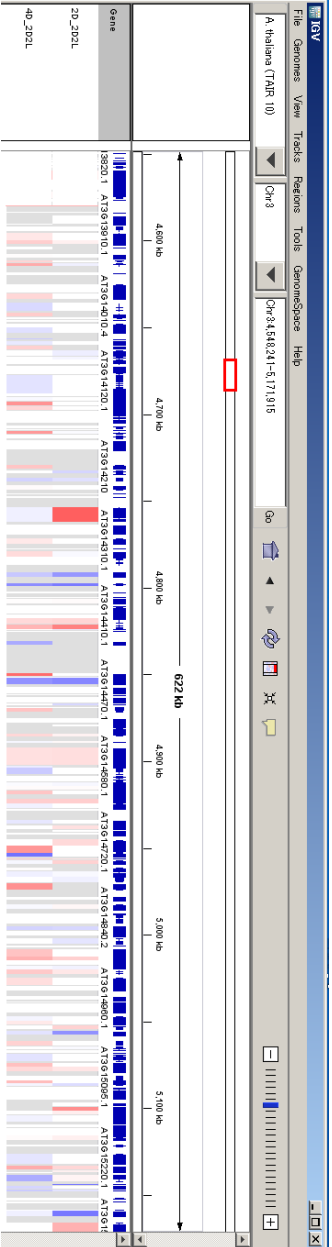


# RNA-Seqのデータ表示させる



#	Name	Description	2D_2D2L	4D_2D2L
1	ANAC001	@Chr1:3630-5899	-2.60184	-2.60956
2	DC11	@Chr1:23145-33153	-0.742675	-1.5642
3	MIR38A	@Chr1:23145-33153	0	0
4	AT1G01073	@Chr1:44676-44787	0	0
5	IOD18	@Chr1:52238-54692	-1.93871	-1.13128
6	AT1G01115	@Chr1:56623-56740	0	0
7	GIF2	@Chr1:72338-74737	-0.251287	-0.616679
8	AT1G01180	@Chr1:75582-76758	0.45929	-0.809567
9	AT1G01210	@Chr1:88897-89745	1.6964	0.857196
10	FRGP	@Chr1:91375-95651	-0.174589	0.725947
11	AT1G01240	@Chr1:99893-101834	-0.226384	-0.936641
12	AT1G01260	@Chr1:108945-111609	-0.161848	0.315699
13	CYP703A2	@Chr1:11262-113947	0	0
14	CNX3	@Chr1:114285-116108	0.111249	-0.551359
15	AT1G01300	@Chr1:116942-118764	-0.68348	0.108578

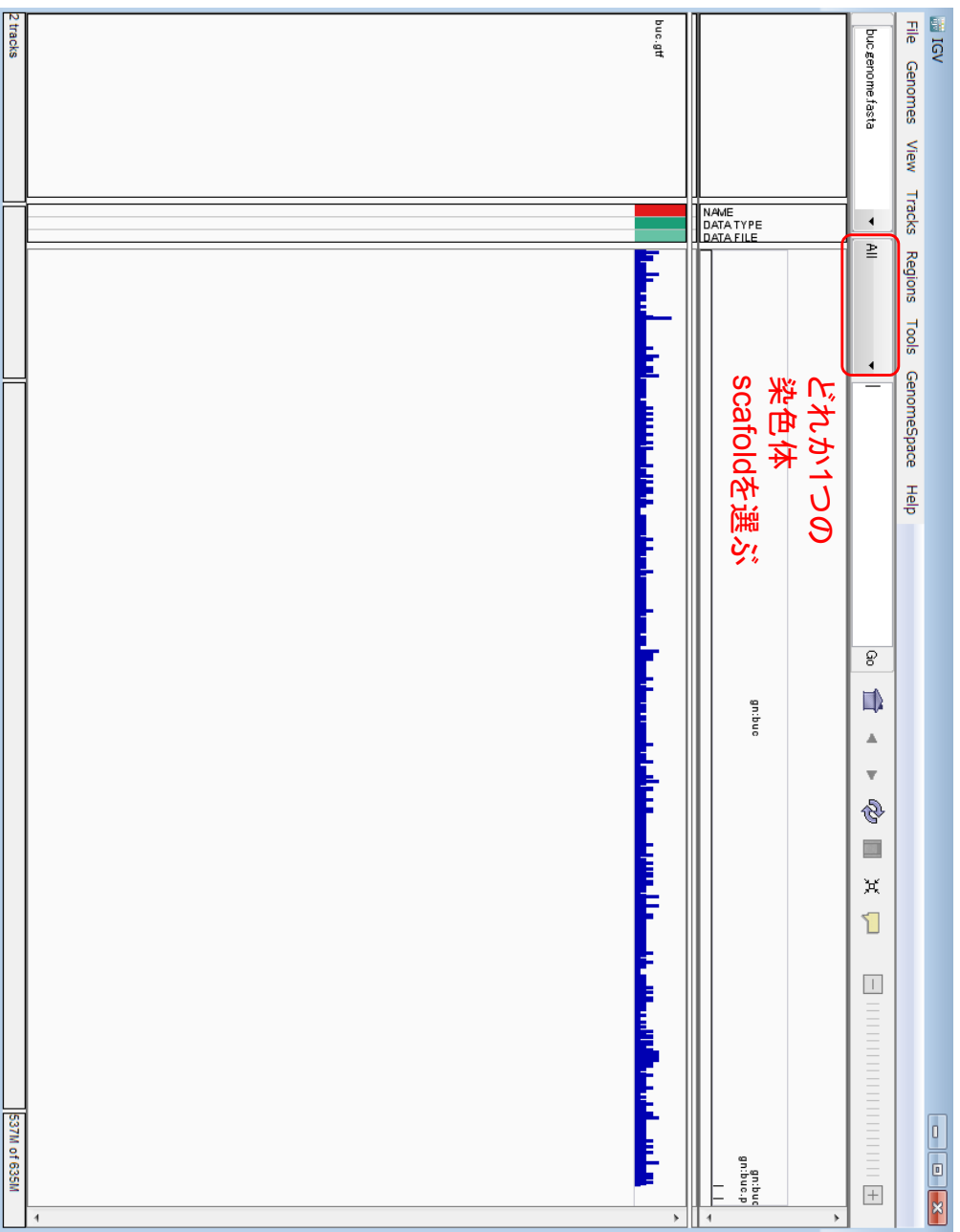
# GCTファイルでgene ローカスの発現情報を図示



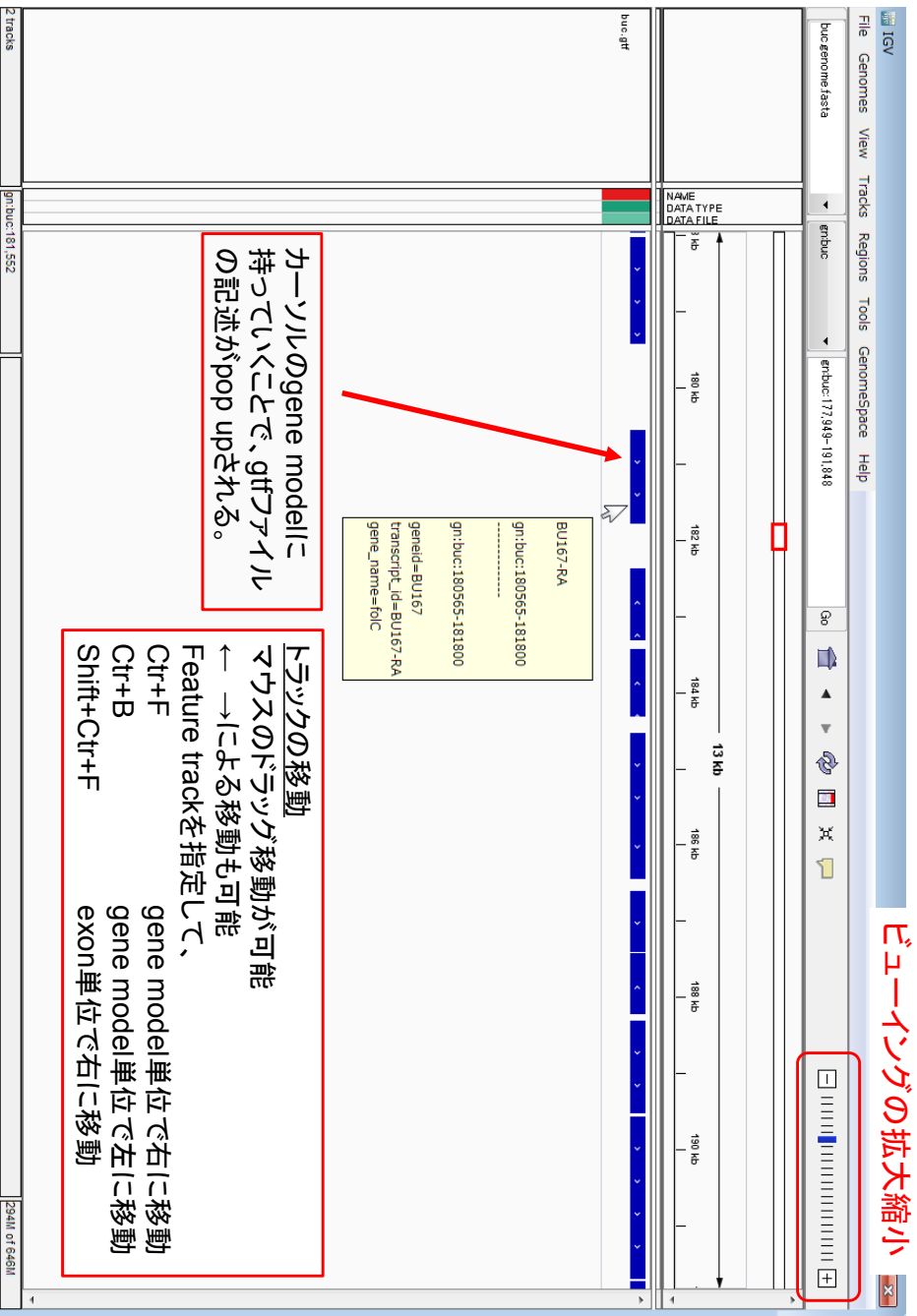
```
buc.genome.fasta
buc.gtf
buc.cg.wig
illumina_ex_B2_Read_bowtie2.mate.sort.bam
illumina_ex_B2_Read_bowtie2.mate.sort.bam.bai
illumina_ex_B4_Read_bowtie2.mate.sort.bam
illumina_ex_B4_Read_bowtie2.mate.sort.bam.bai
```







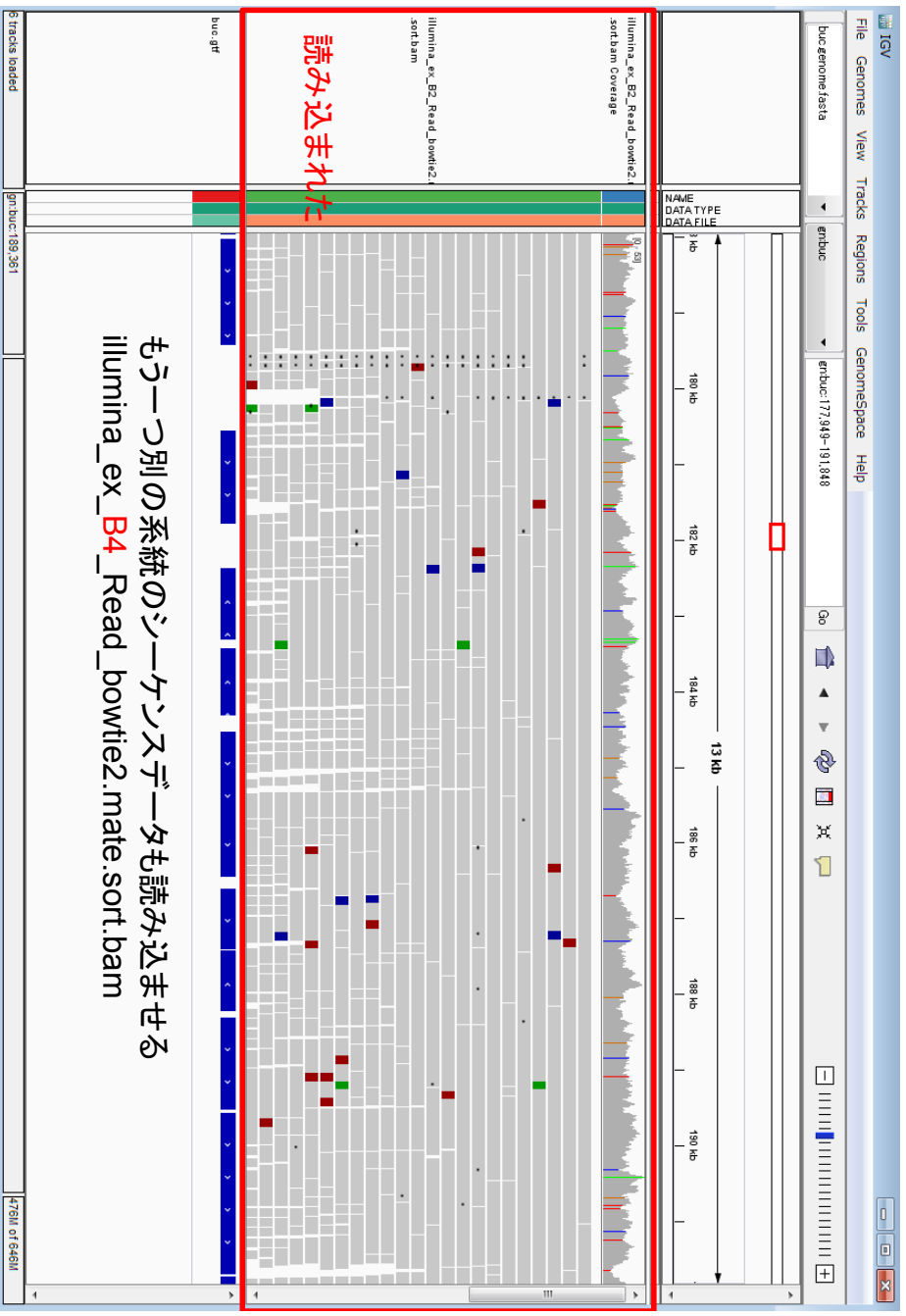
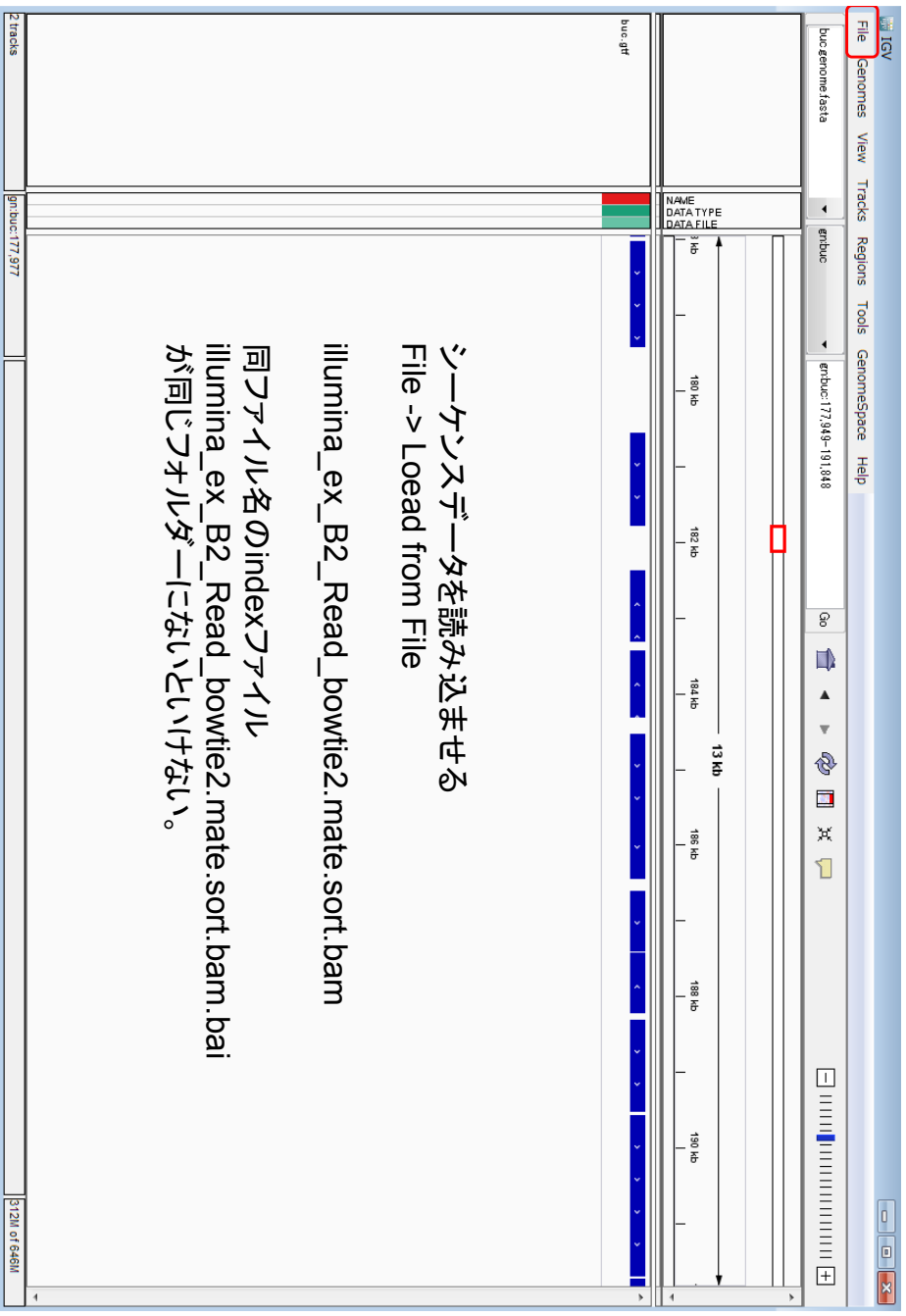
どれか1つの  
染色体  
scaffoldを選ぶ



ビューイングの拡大縮小

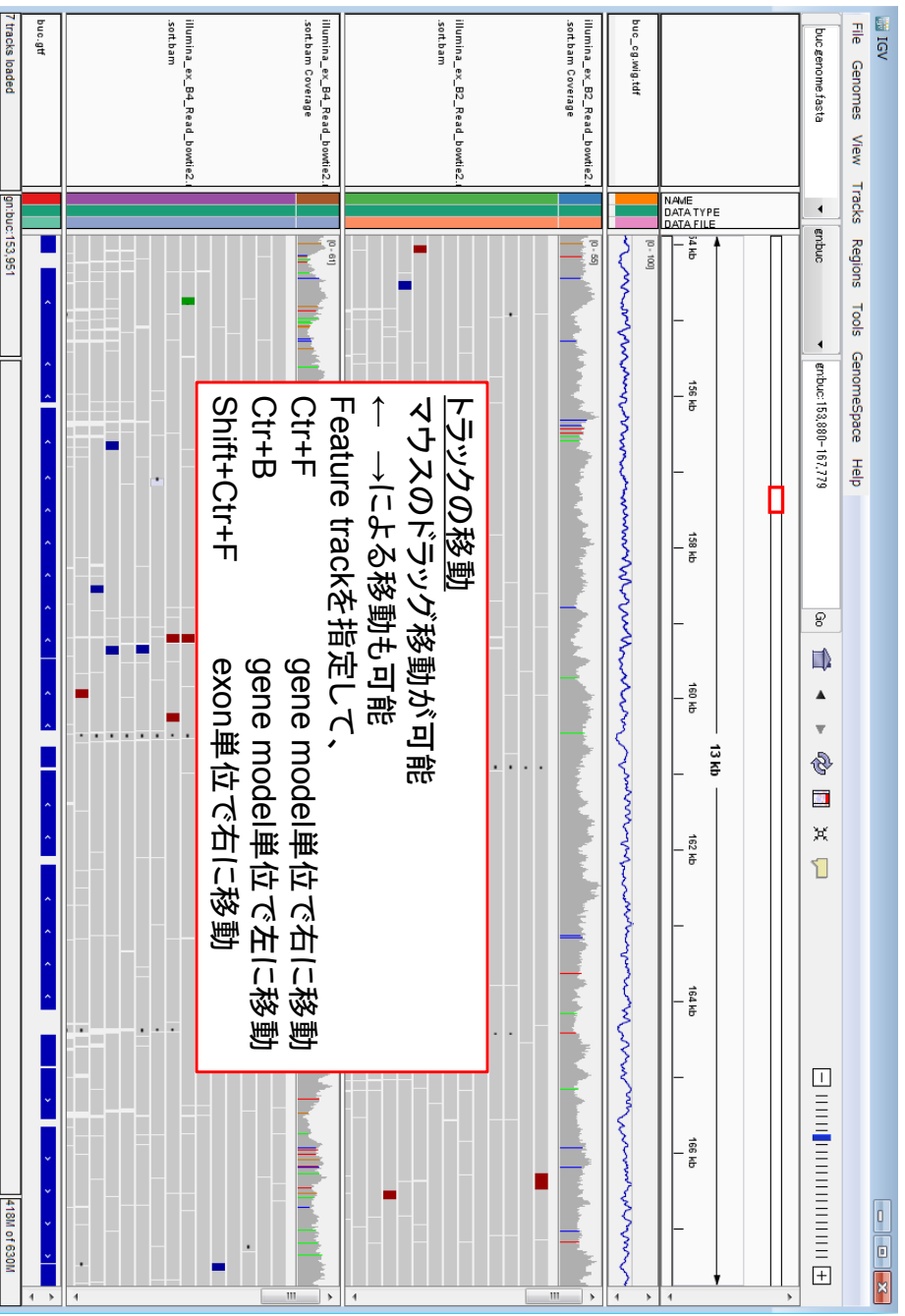
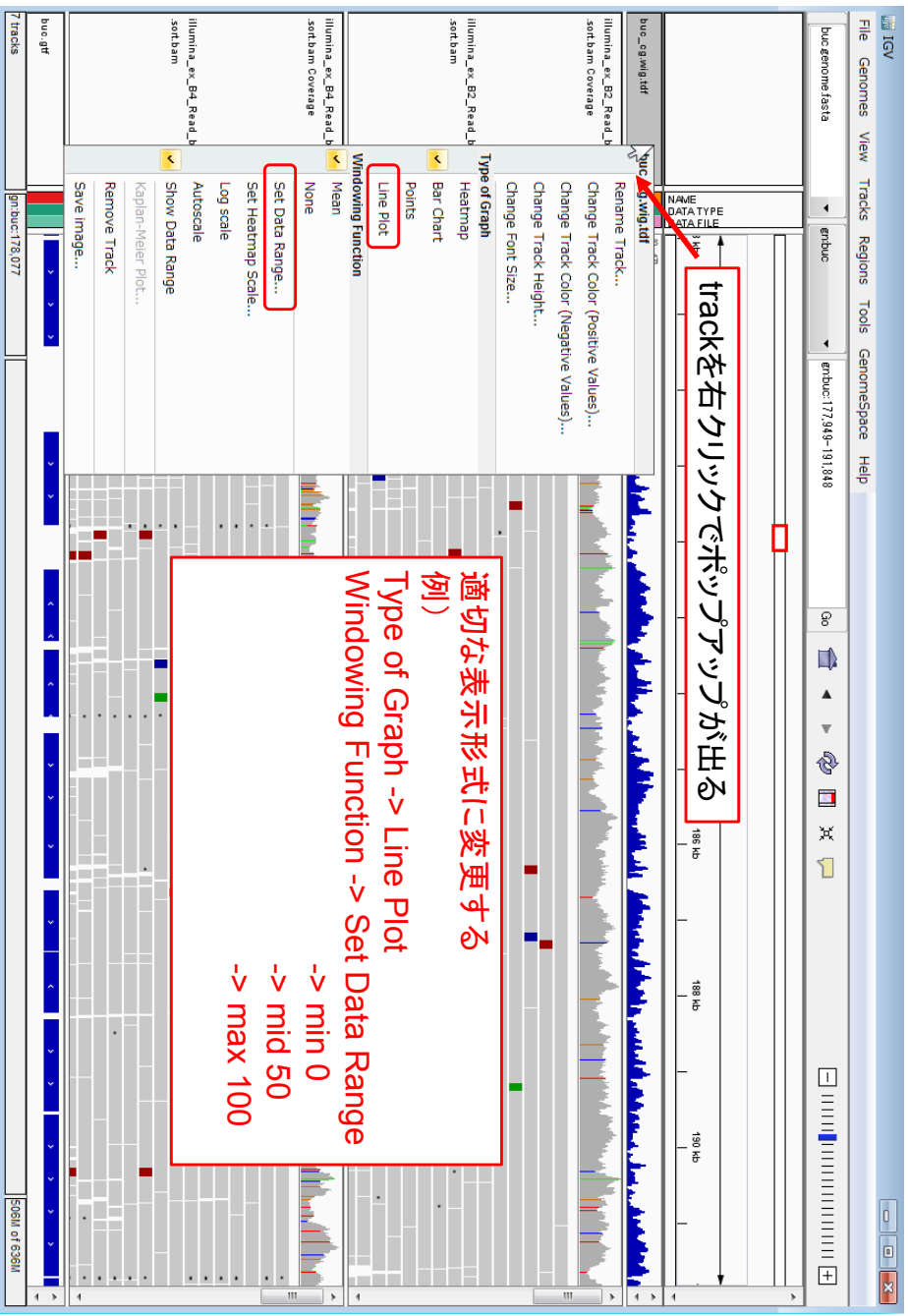
カーソルのgene modelに  
持っていくことで、gtfファイル  
の記述がpop upされる。

トラックの移動  
マウスのドラッグ移動が可能  
← →による移動も可能  
Feature trackを指定して、  
Ctrl+F  
Ctrl+B  
Shift+Ctrl+F  
gene model単位で右に移動  
gene model単位で左に移動  
exon単位で右に移動









**トラックの移動**  
マウスのドラッグ移動が可能  
← →による移動も可能  
Feature trackを指定して、  
Ctrl+F  
Ctrl+B  
Shift+Ctrl+F  
gene model単位で右に移動  
gene model単位で左に移動  
exon単位で右に移動







# IGV紹介のまとめ

可視化ツールとして十分な機能を持つ

- ・無料
- ・比較的簡単・お手軽
- ・自分で見るためにも良し、人に見せるためにも良し
- ・利用範囲は次世代DNAシーケンサーに限定しない  
広くゲノミクスの解析に有用

ごく一部のみの機能を紹介しました。  
ウェブサイトを見ながら復習をお勧めします。