

# NGS基本ツールIGV

基礎生物学研究所  
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. On the left is a sidebar with navigation links: Home, Downloads, Documents, Hosted Genomes, FAQ, IGV User Guide, File Formats, Release Notes, Credits, and Contact. Below these is a search bar and copyright information for 2013-2018. The main content area features a large banner image of the IGV interface. Below the banner are sections for Overview, Downloads, Funding, and Citing IGV. The Overview section describes IGV as a high-performance visualization tool. The Downloads section provides a link to download the desktop application. The Funding section lists the National Cancer Institute and the National Institutes of Health as funders. The Citing IGV section provides citation information for publications.

**Home**

**Integrative Genomics Viewer**

**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.

**Downloads**

Download the IGV desktop application and igvtools.

**Funding**

Development of IGV has been supported by funding from the [National Cancer Institute \(NCI\)](#) of the [National Institutes of Health](#), the [Informatics Technology for Cancer Research \(ITCR\)](#) of the NCI, and the [Starr Cancer Consortium](#).

IGV participates in the [GenomeSpace](#) initiative, which is funded by the [National Human Genome Research Institute](#).

**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 \(IGV\): high-performance genomics data visualization and exploration. \*Briefings in Bioinformatics\* 14, 178-192 \(2013\).](#)

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

# なぜIGVを取り上げるか

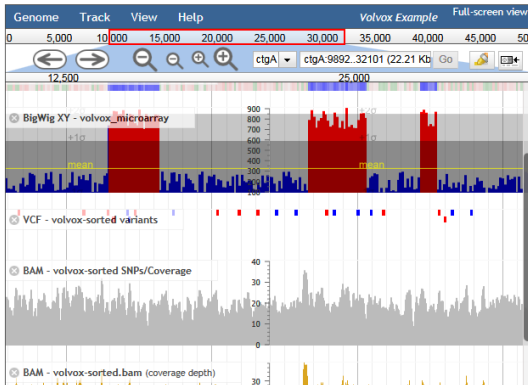
## データ可視化ツール

- ・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、ネットワーク上で使うタイプ

### 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.

Latest Release – [JBrowse 1.14.2](#)



後者はコミュニティで広く利用、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

多様なデジタル情報

- ・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
- ・ChIP-seq, RAD-seq, BS-seq...

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

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、  
統合的直感的に比較・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 / コミュニティーレベルの使用
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
- ・コミュニティー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、サンプルデータのある
- ・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

誰もが簡便に使えるものが良い。



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- IGV User Guide
- File Formats
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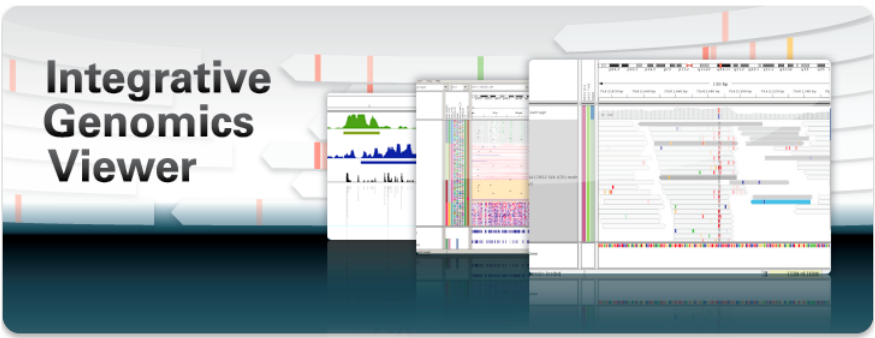
---

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Home



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NATURE BIOTECHNOLOGY | OPINION AND COMMENT | CORRESPONDENCE

## Integrative genomics viewer

James T Robinson, Helga Thorvaldsdóttir, Wendy Winckler, Mitchell Guttman, Eric S Lander, Gad Getz & Jill P Mesirov

**Affiliations** | **Corresponding authors**

*Nature Biotechnology* **29**, 24–26 (2011) | doi:10.1038/nbt.1754  
Published online 10 January 2011

To the Editor:

Rapid improvements in sequencing and array-based platforms are resulting in a flood of diverse genome-wide data, including data from exome and whole-genome sequencing, epigenetic surveys, expression profiling of coding and noncoding RNAs, single nucleotide polymorphism (SNP) and copy number profiling, and functional assays. Analysis of these large, diverse data sets holds the promise of a more comprehensive understanding of the genome and its relation to human disease. Experienced and knowledgeable human review is an essential component of this process, complementing computational approaches. This calls for efficient and intuitive visualization tools able to scale to very large data sets and to flexibly integrate multiple data types, including clinical data. However, the sheer volume and scope of data pose a significant challenge to the development of such tools.

**Journal home**

**Current issue**

**For authors**

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**Ramalingaswami Re-Entry Fellowship**  
Ministry of Science & Technology, Government of India



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- Navigating
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- Viewing the Reference Genome
- Loading Data and Attributes
- Viewing Data
- Viewing Alignments
- Viewing Variants
- Gene List View
- Regions of Interest
- Sample Attributes
- Sorting, Grouping, and Filtering
- Saving and Restoring Sessions
- Server Configuration
- igvtools
- Motif Finder
- BLAT search
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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>

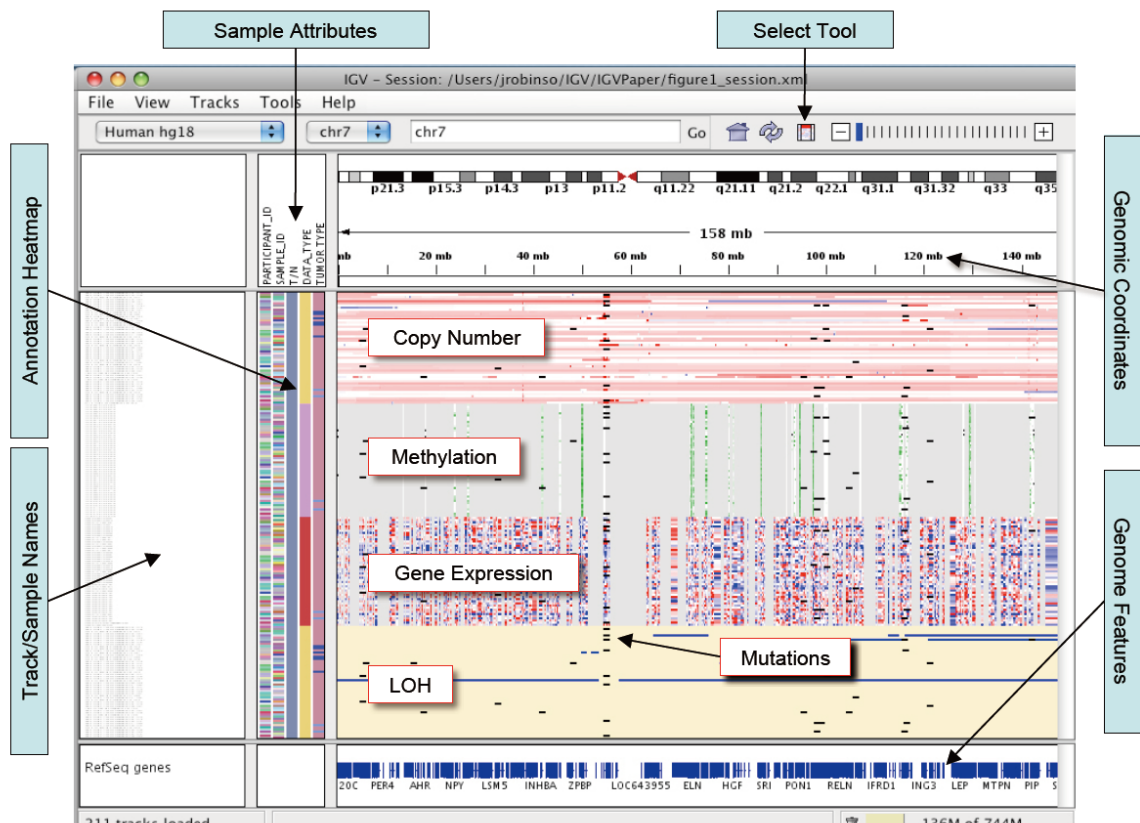
[Look at a printer-friendly HTML version of the whole User Guide.](#)

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[User Interface >](#)





Nature Biotech. 29:24–26 (2011) Supplement figureからの抜粋



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## Downloads

### Integrative Genomics Viewer - IGV 2.4

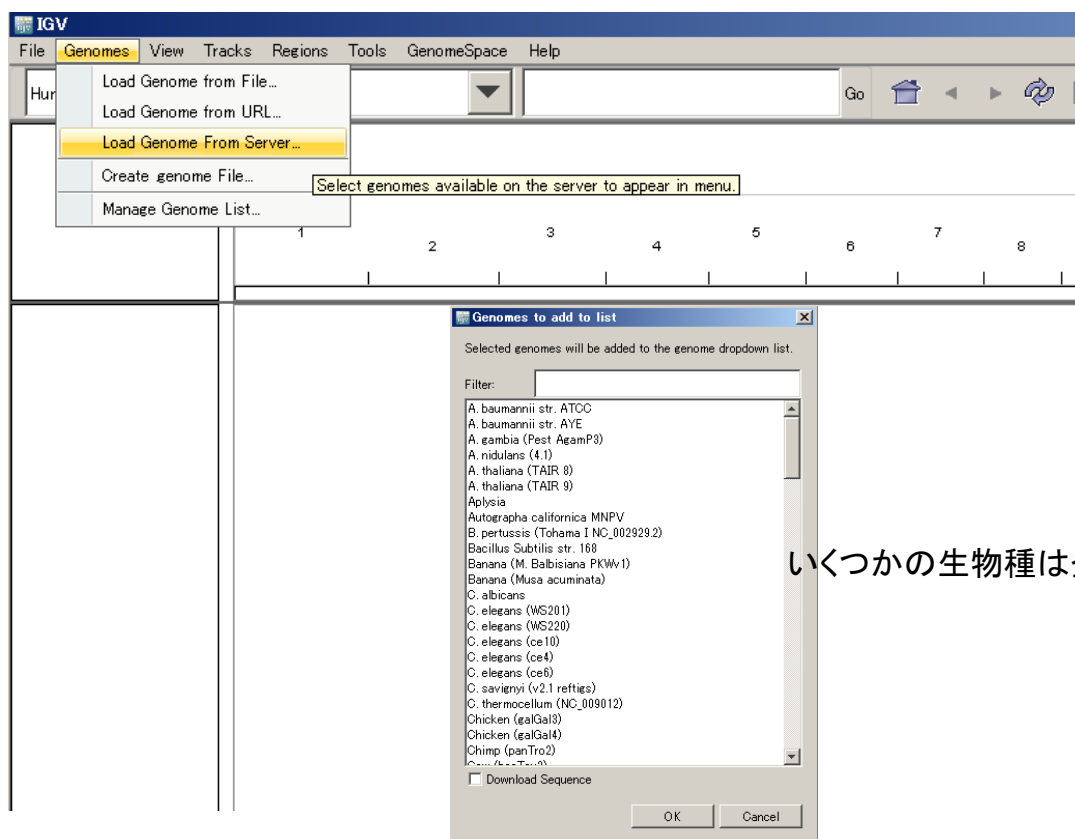
#### Install IGV

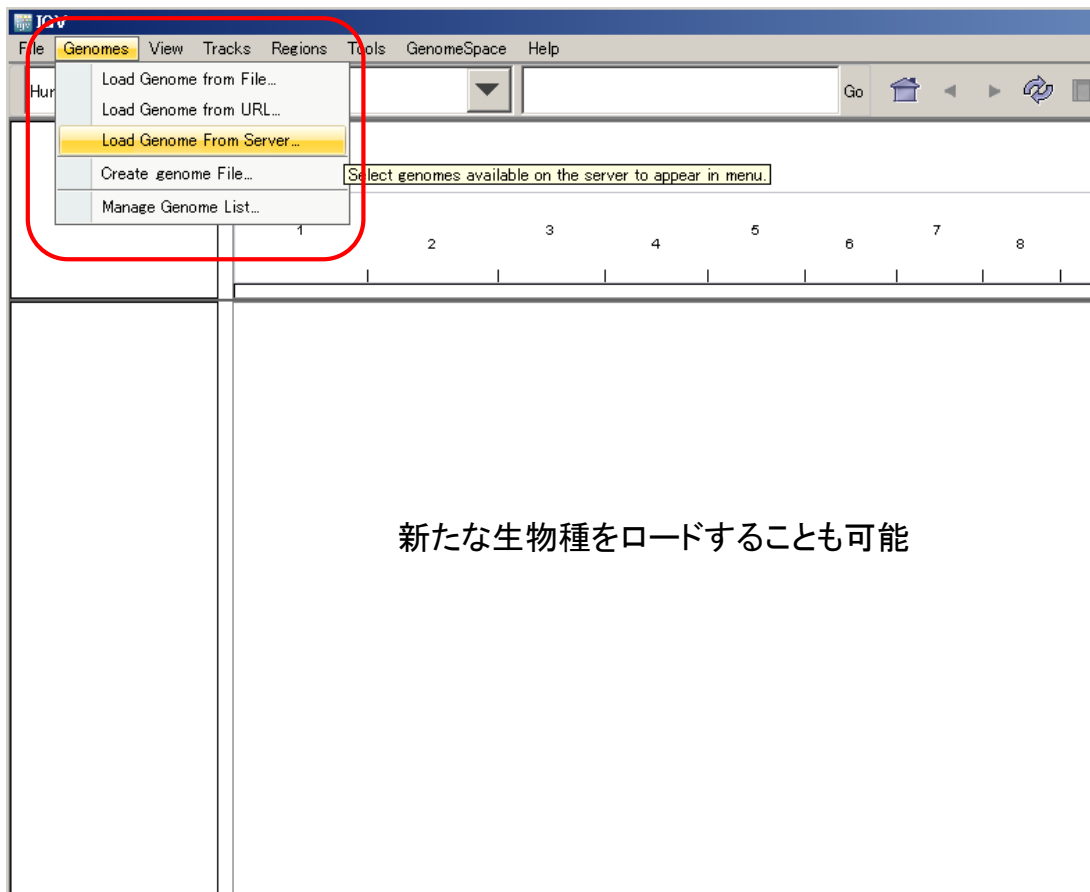
**NOTE: IGV 2.4.x requires Java 8. Java versions 9 and above are currently not supported.**

Use one of the following 4 options to install and run the current version of IGV.

1.  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.
2.  Download and unzip the **Windows Zip Archive**, then double-click the *igv.bat* file to start IGV. A black console window will appear, followed by the IGV application. **Note:** Windows users with **high resolution screens** should use this version -- it includes a modified Java executable for use with high-resolution screens.
3.  Download and unzip the **Binary Distribution archive**. IGV is launched from a command prompt -- follow the instructions in the *readme* file. To launch IGV on Mac or Linux use the shell script *igv.sh*. On Windows use *igv.bat*.
4. Click on one of the *Launch* buttons below to download a .jnlp file and execute the file using **Java Web Start (JWS)**.
  - **Mac users:** If you are notified of security errors that prevent launching IGV, try the following:
    - Right-click on the downloaded .jnlp file; select *Open With > Java Web Start*; dismiss the warnings.
    - After IGV has been run this way at least once from the .jnlp file, you can double-click on the file to launch.
  - **Windows users:** To run with more than 1.2 GB of memory on Windows 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*".

 Launch with 750 MB	 Launch with 1.2 GB (Max usable memory for Windows with 32-bit Java)	 Launch with 2 GB (Max usable memory for 32-bit MacOS)	 Launch with 10 GB (Only for large memory machines with 64-bit Java)
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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
- Chemical Reactivity Probing Profiles
- chrom.sizes
- CN
- Custom File Formats
- Cytoband
- FASTA
- GCT
- CRAM
- genePred
- GFF/GTF
- GISTIC
- Goby
- GWAS
- IGV
- LOH
- MAF (Multiple Alignment Format)
- MAF (Mutation Annotation Format)
- Merged BAM File
- MUT
- narrowPeak
- PSL
- RES
- RNA Secondary Structure Formats
- SAM
- Sample Info (Attributes) file
- SEG
- SNP
- TAB
- TDF
- Track Line
- Type Line
- VCF
- WIG

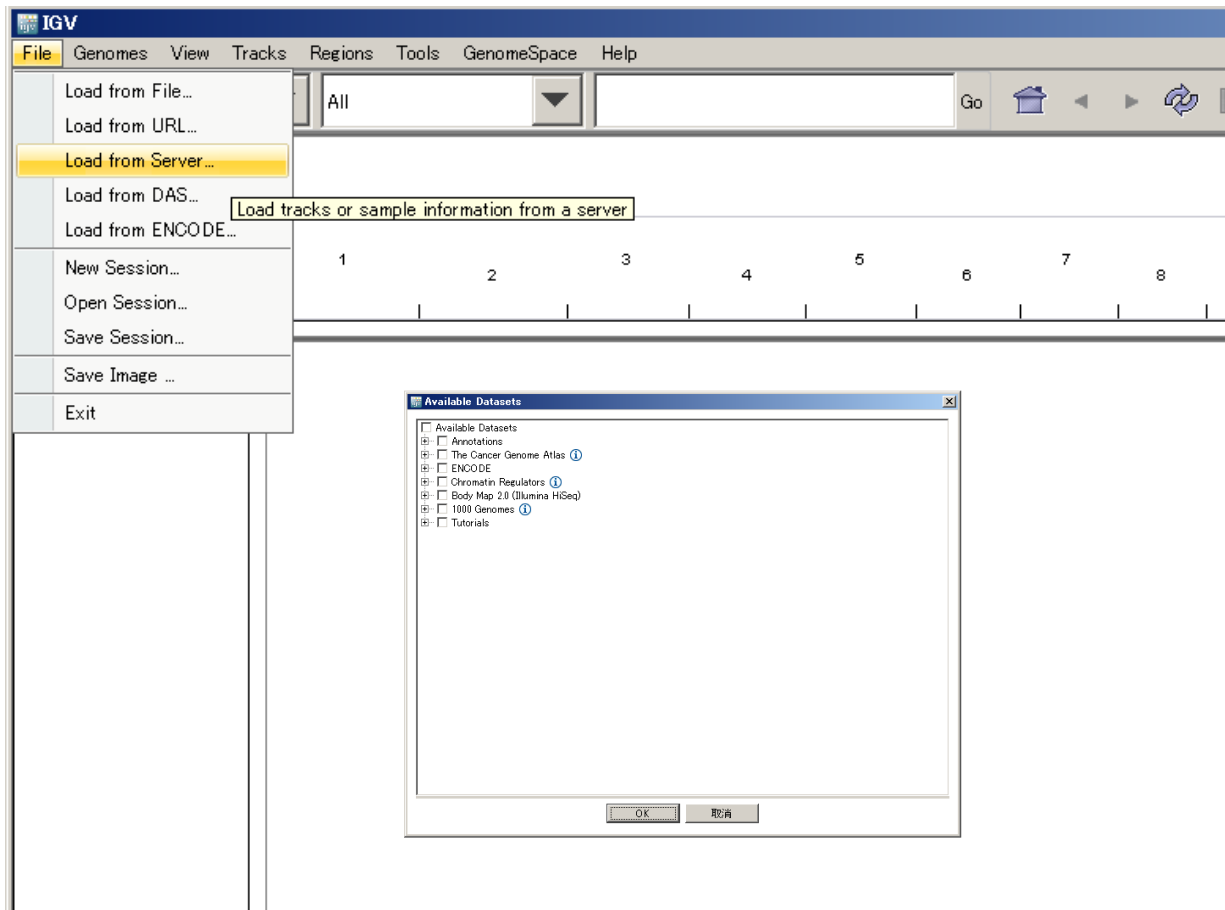
#### File Formats

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
GISTIC data	GISTIC
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。

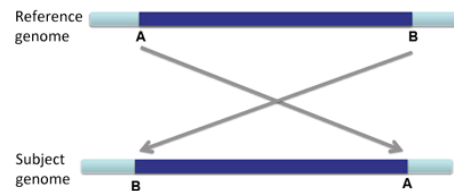
### バッチ処理

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。

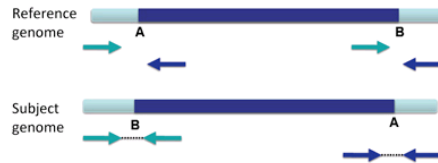
```
new
load myfile.bam
snapshotDirectory mySnapshotDirectory
genome hg18
goto chr1:65,289,335-65,309,335
sort position
collapse
snapshot
goto chr1:113,144,120-113,164,120
sort base
collapse
snapshot
```

## Inversions

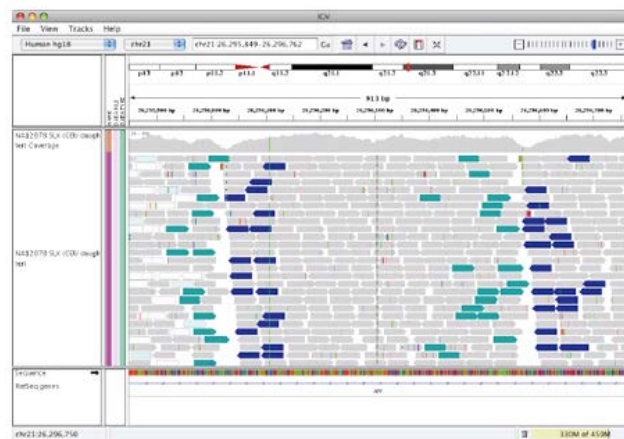
An inversion is a large section of DNA that is reversed in the subject genome compared to the reference genome.



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:

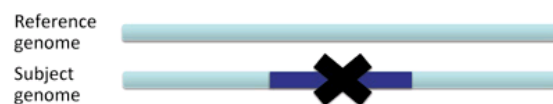
- deletions
- insertions
- inter-chromosomal rearrangements

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:

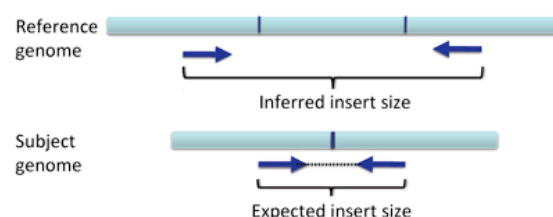
-  for an insert that is larger than expected
-  for an insert that is smaller than expected
-  for paired end reads that are coded by the chromosome on which their mates can be found

## Deletions

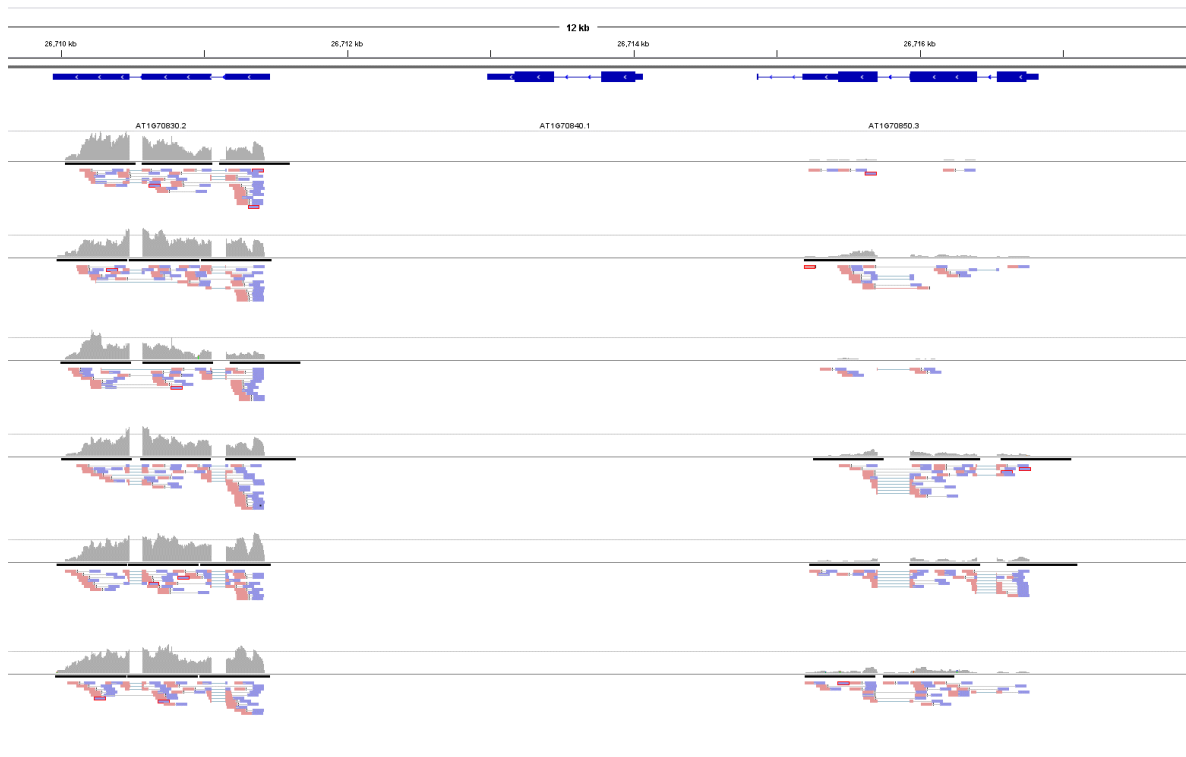
A deletion is a large section of DNA that is absent in the subject genome compared to the reference genome.



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.

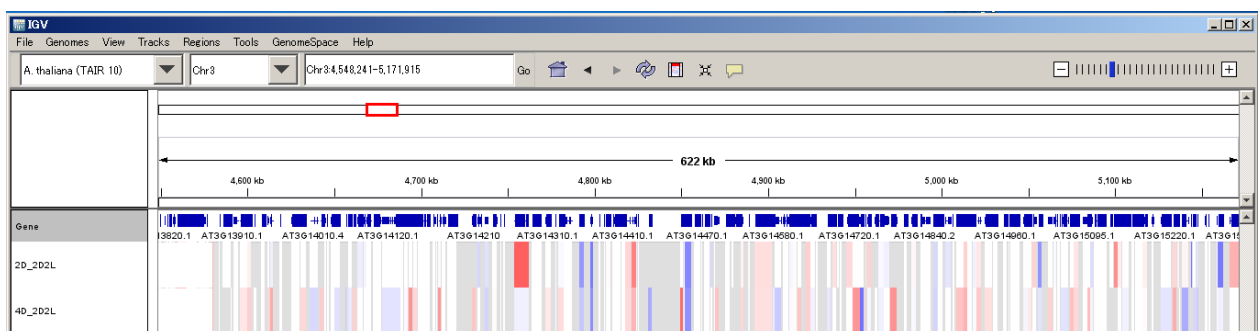


# RNA-Seqのデータ表示させる

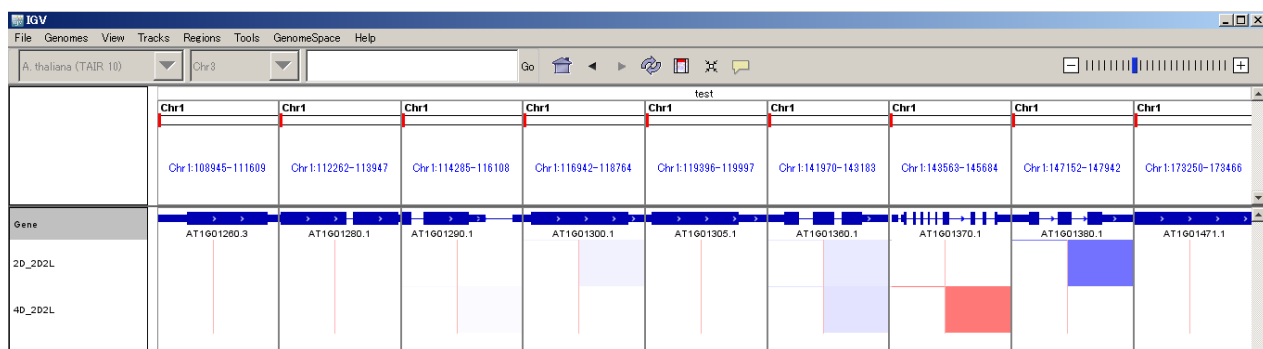
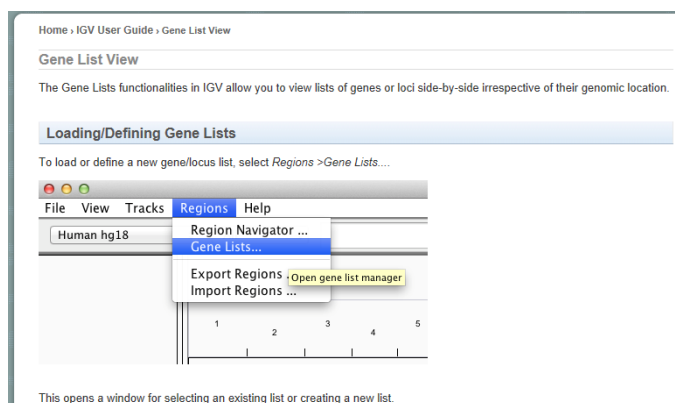


## GCTファイルでgene ローカスの発現情報を図示

#				
#	Name	Description	2D_2D2L	4D_2D2L
	ANAC001	@Chr1:3630-5899	-2.60184	-2.60956
	DCL1	@Chr1:23145-33153	-0.742675	-1.5642
	MIR838A	@Chr1:23145-33153	0	0
	AT1G01073	@Chr1:44676-44787	0	0
	IQD18	@Chr1:52238-54692	-1.93871	-1.13128
	AT1G01115	@Chr1:56623-56740	0	0
	GIF2	@Chr1:72338-74737	-0.251287	-0.616679
	AT1G01180	@Chr1:75582-76758	0.45929	-0.809567
	AT1G01210	@Chr1:88897-89745	1.6964	0.857196
	FKGP	@Chr1:91375-95651	-0.174589	0.725947
	AT1G01240	@Chr1:99893-101834	-0.226384	-0.936641
	AT1G01260	@Chr1:108945-111609	-0.161848	0.315699
	CYP703A2	@Chr1:112262-113947	0	0
	CNX3	@Chr1:114285-116108	0.111249	-0.551359
	AT1G01300	@Chr1:116942-118764	-0.68348	0.108578



Gene listを定義して  
サンプルごと  
条件ごと  
の発現・発現変動を  
カラーマップできる



## IGV実習

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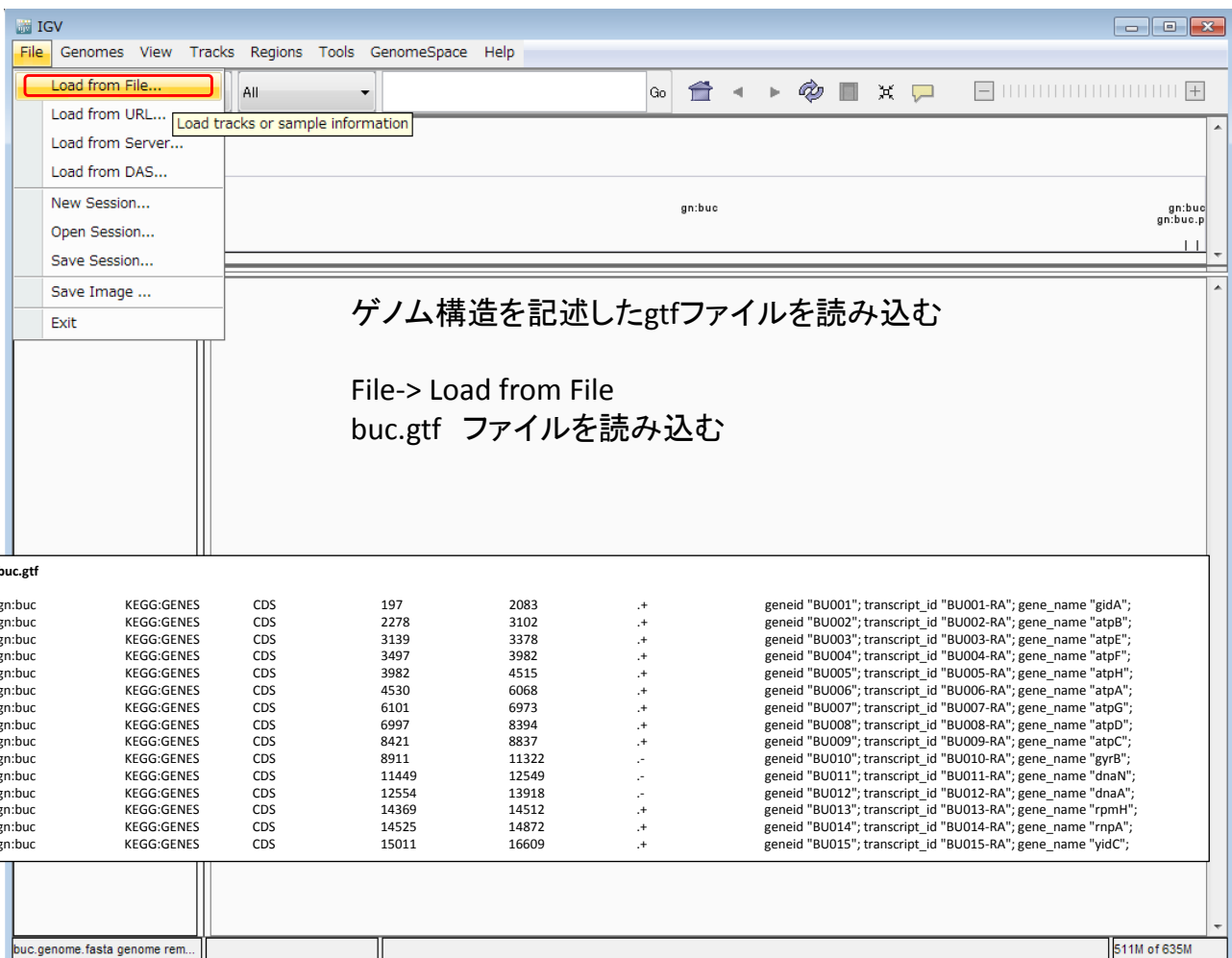
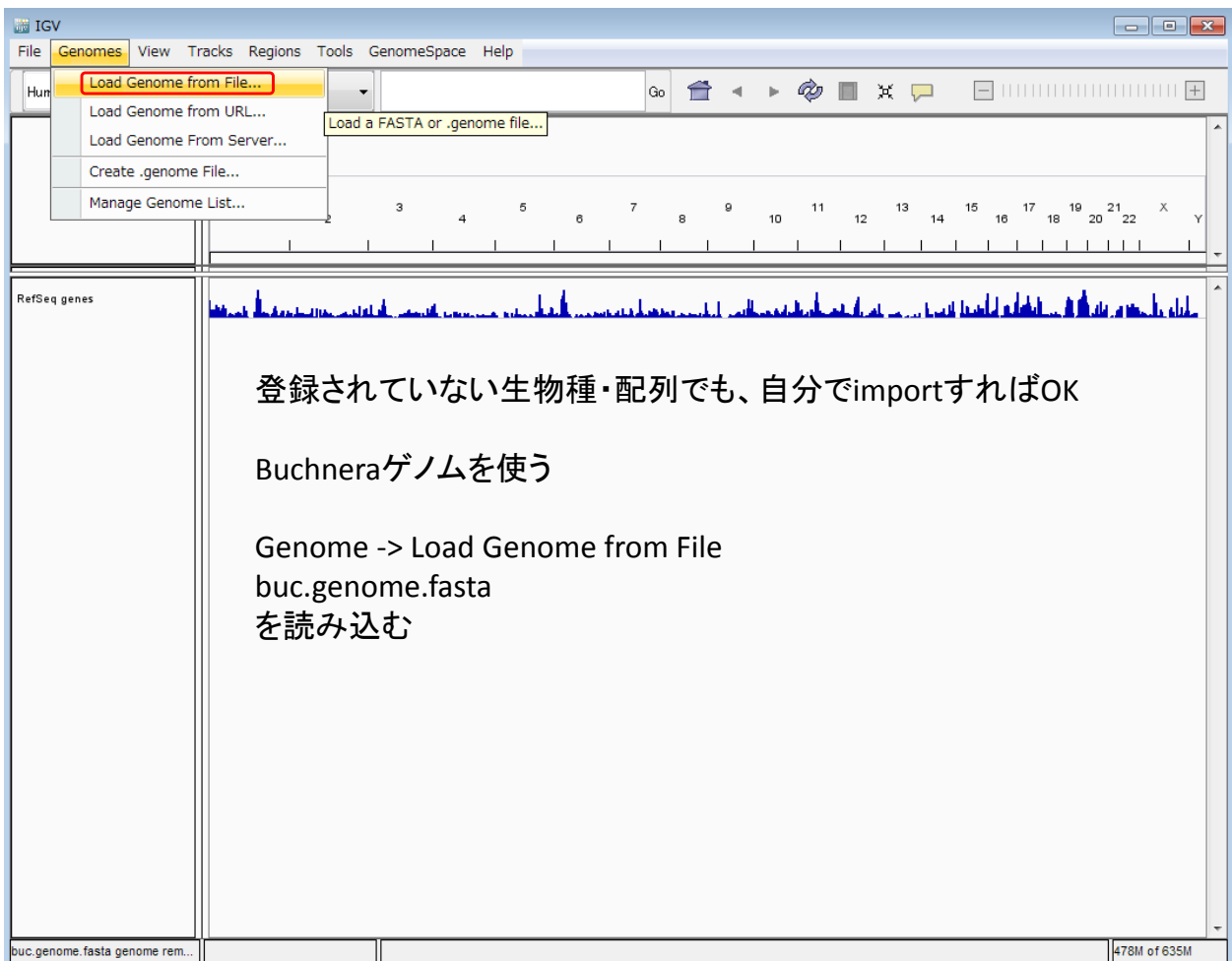
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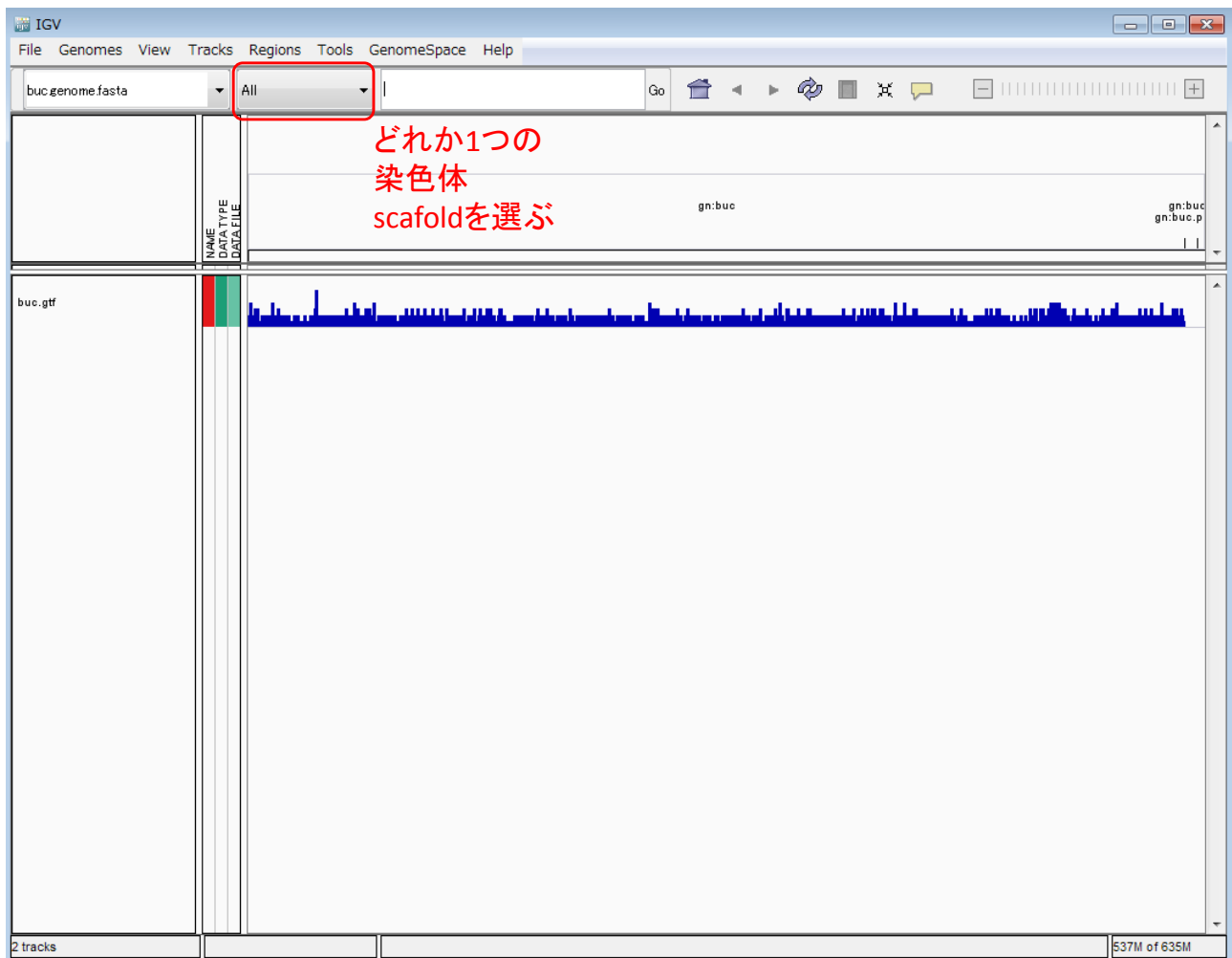
IGVの使用法を学ぶと共に  
先のファイルフォーマットも  
確認しよう

以下のファイルを確認

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



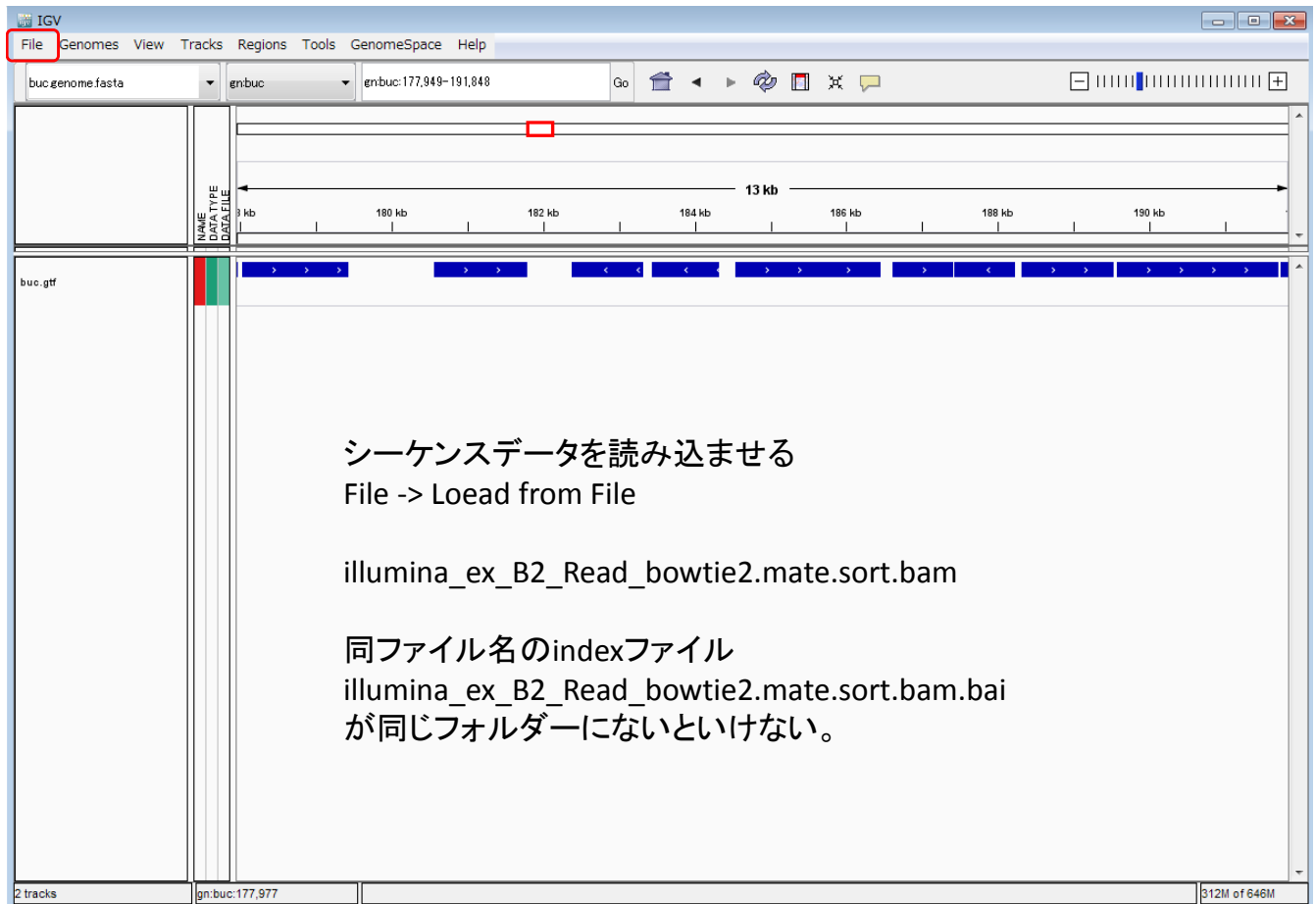




ビューイングの拡大縮小

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。

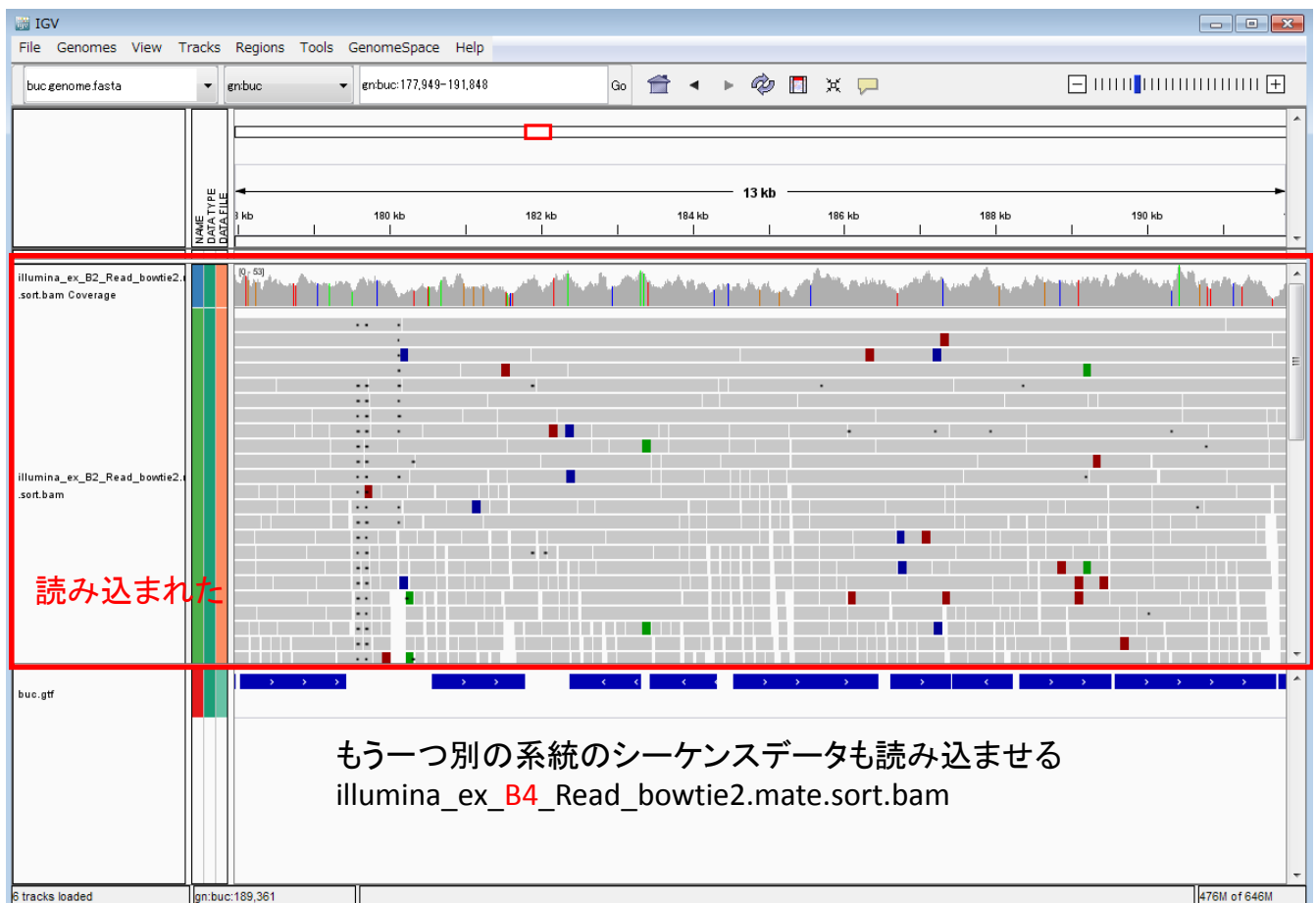
**トラックの移動**  
マウスのドラッグ移動が可能  
← →による移動も可能  
Feature trackを指定して、  
Ctrl+F gene model単位で右に移動  
Ctrl+B gene model単位で左に移動  
Shift+Ctrl+F exon単位で右に移動



シーケンスデータを読み込ませる  
File -> Load from File

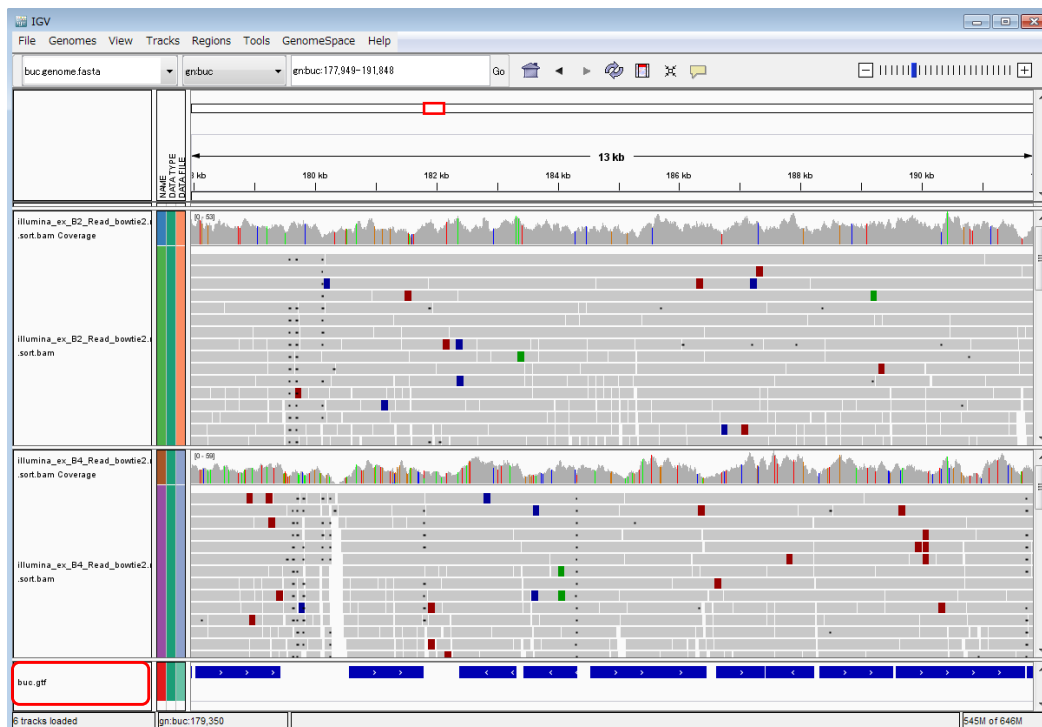
illumina\_ex\_B2\_Read\_bowtie2.mate.sort.bam

同ファイル名のindexファイル  
illumina\_ex\_B2\_Read\_bowtie2.mate.sort.bam.bai  
が同じフォルダーにないといけない。



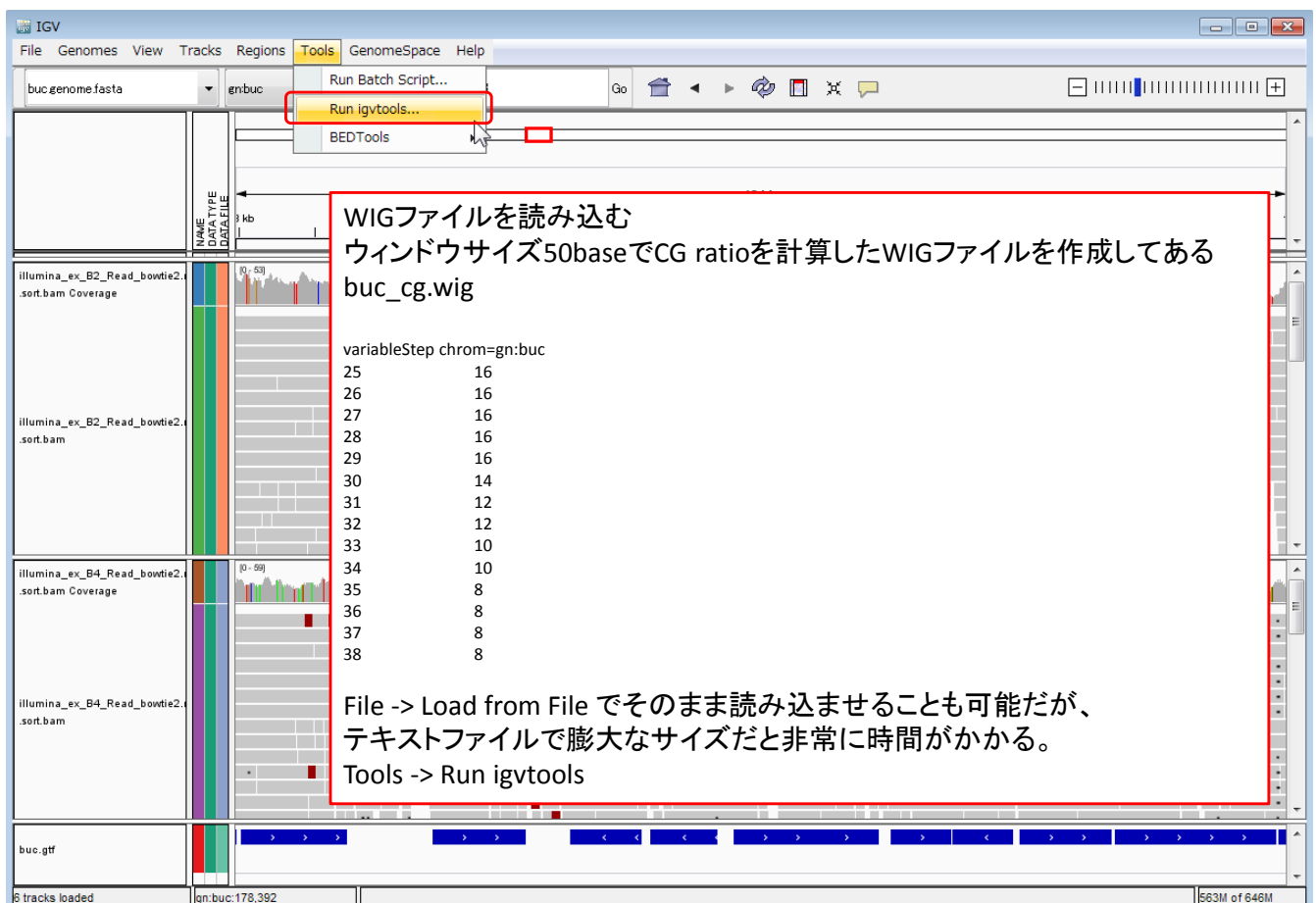
読み込まれた

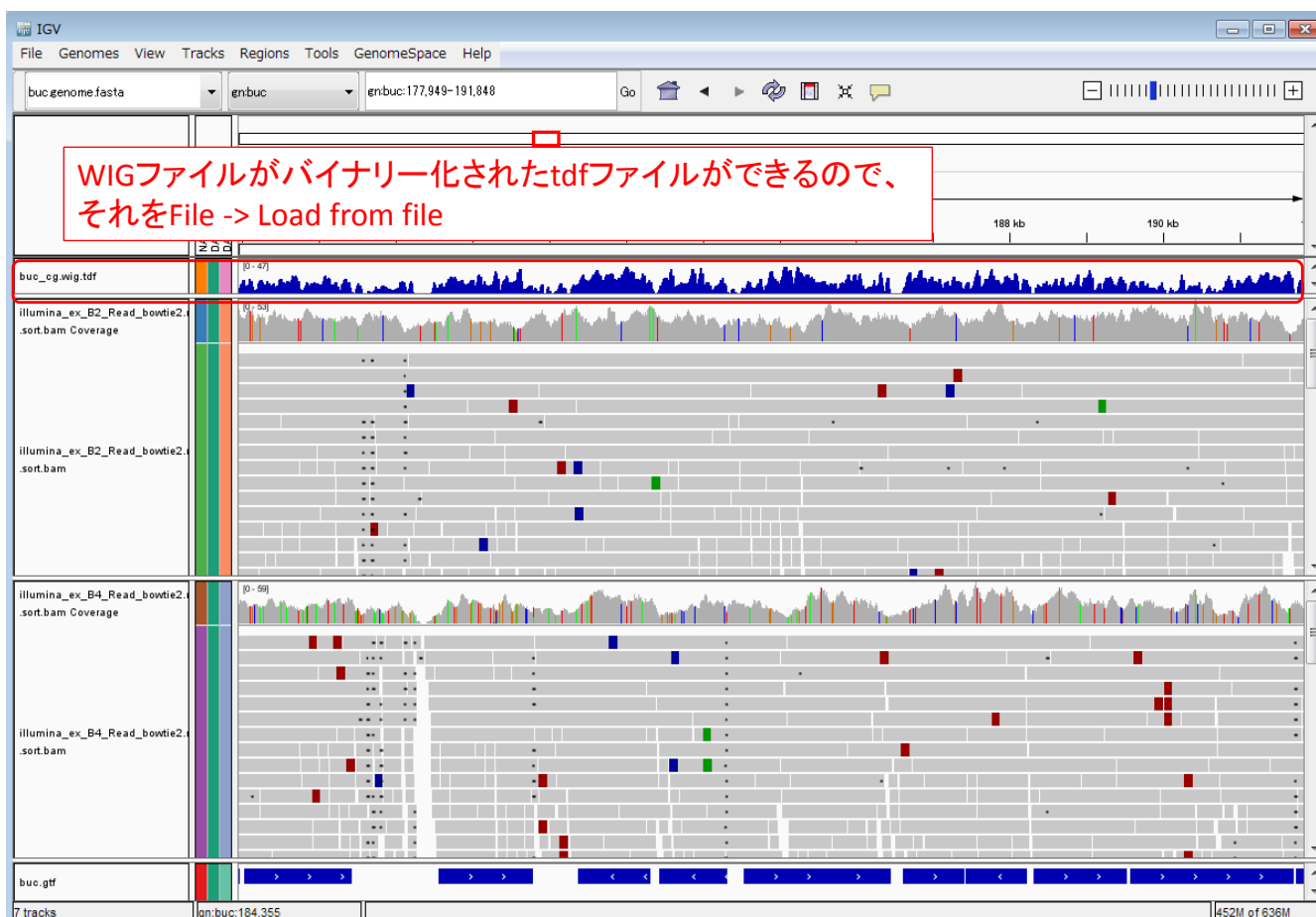
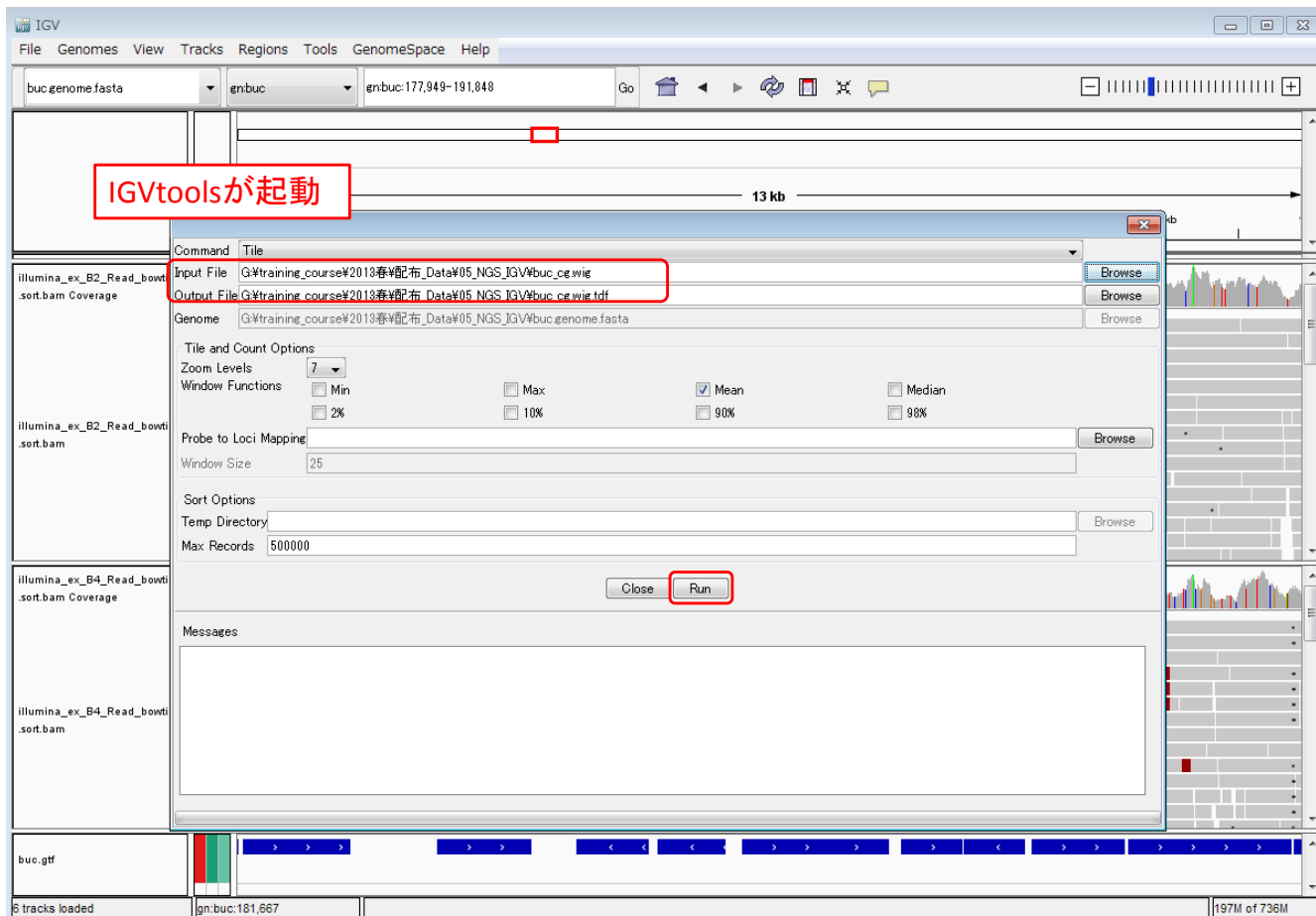
もう一つ別の系統のシーケンスデータも読み込ませる  
illumina\_ex\_B4\_Read\_bowtie2.mate.sort.bam

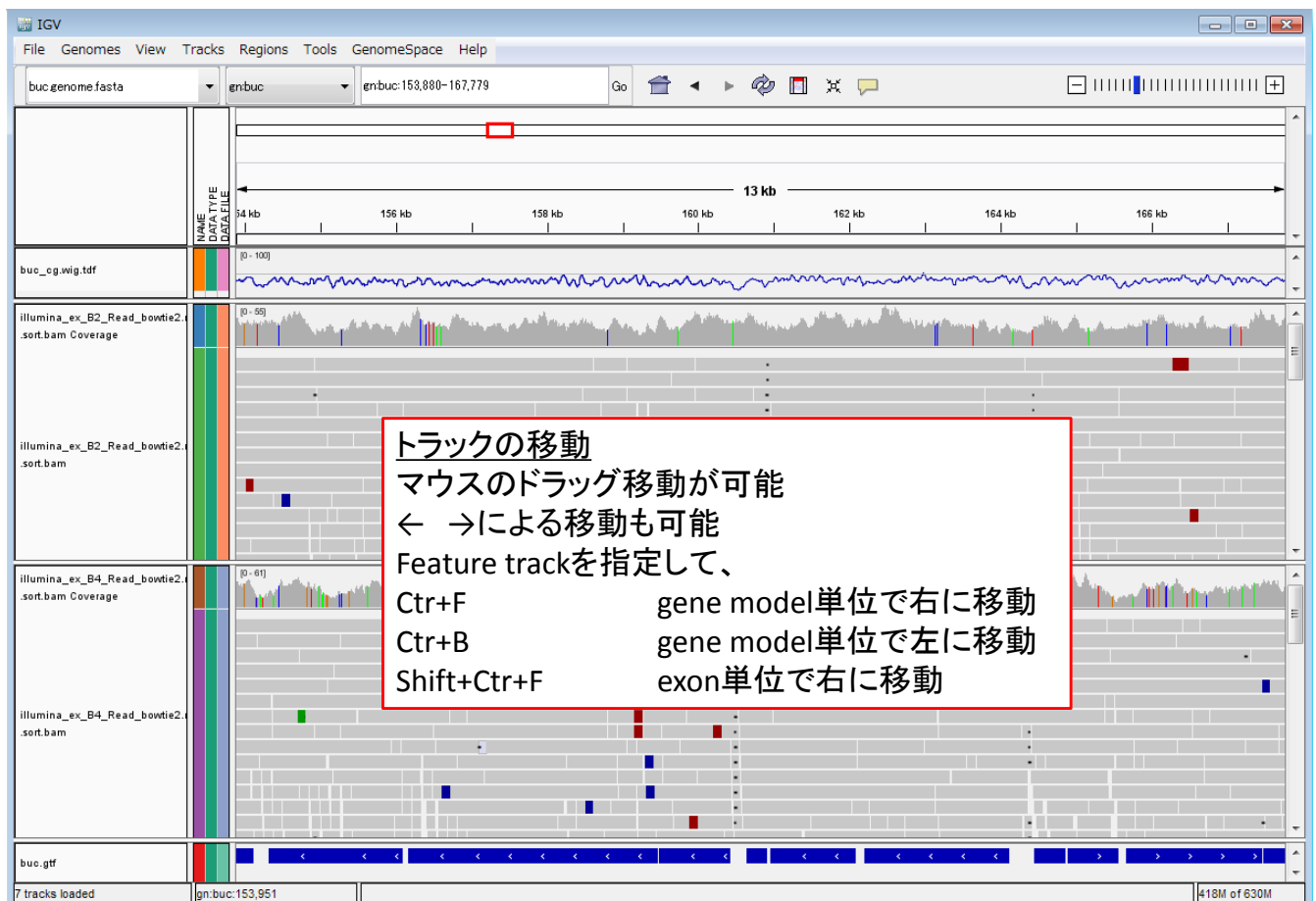
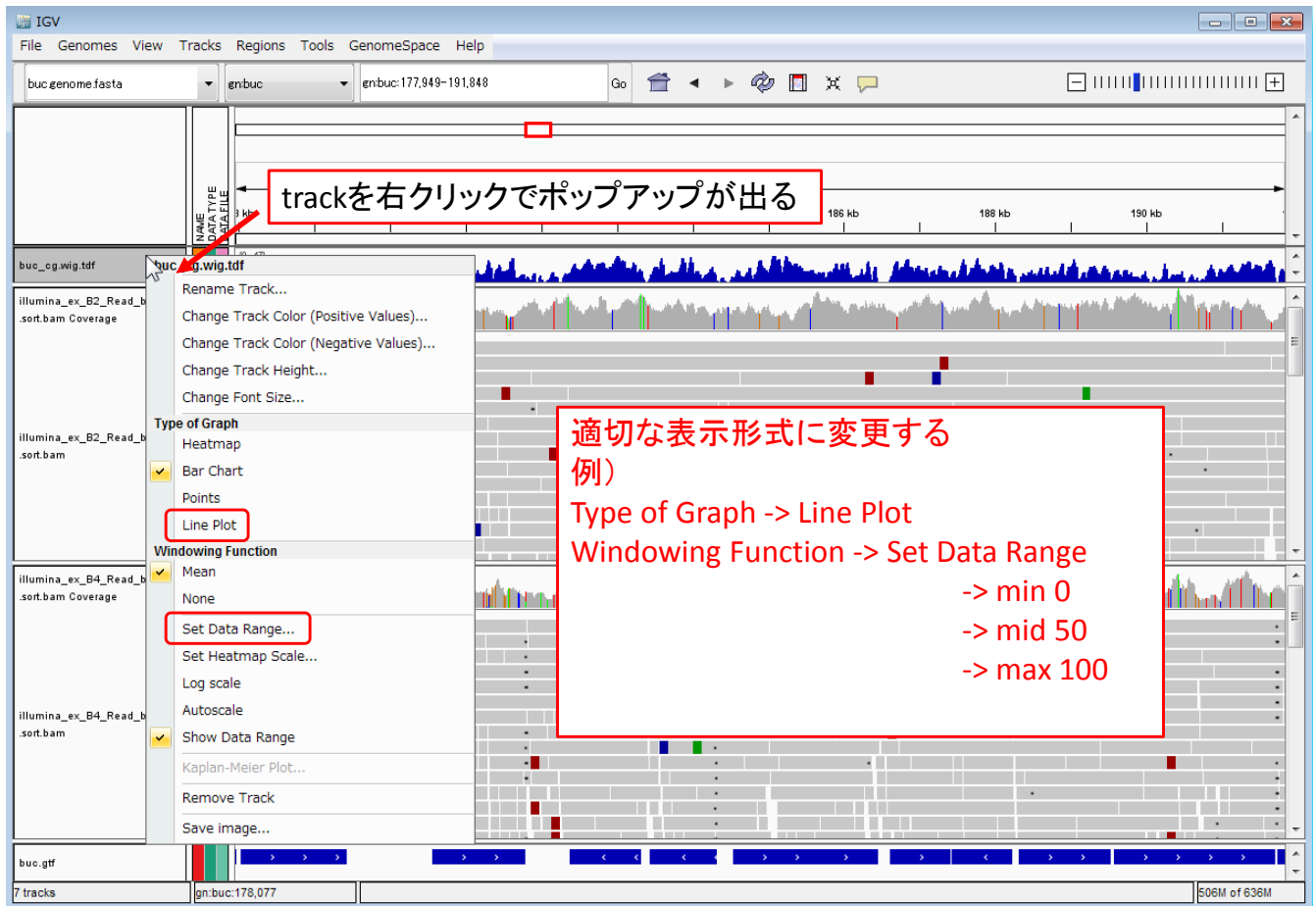


こういう所をクリックしたまま移動させることで幅を変更

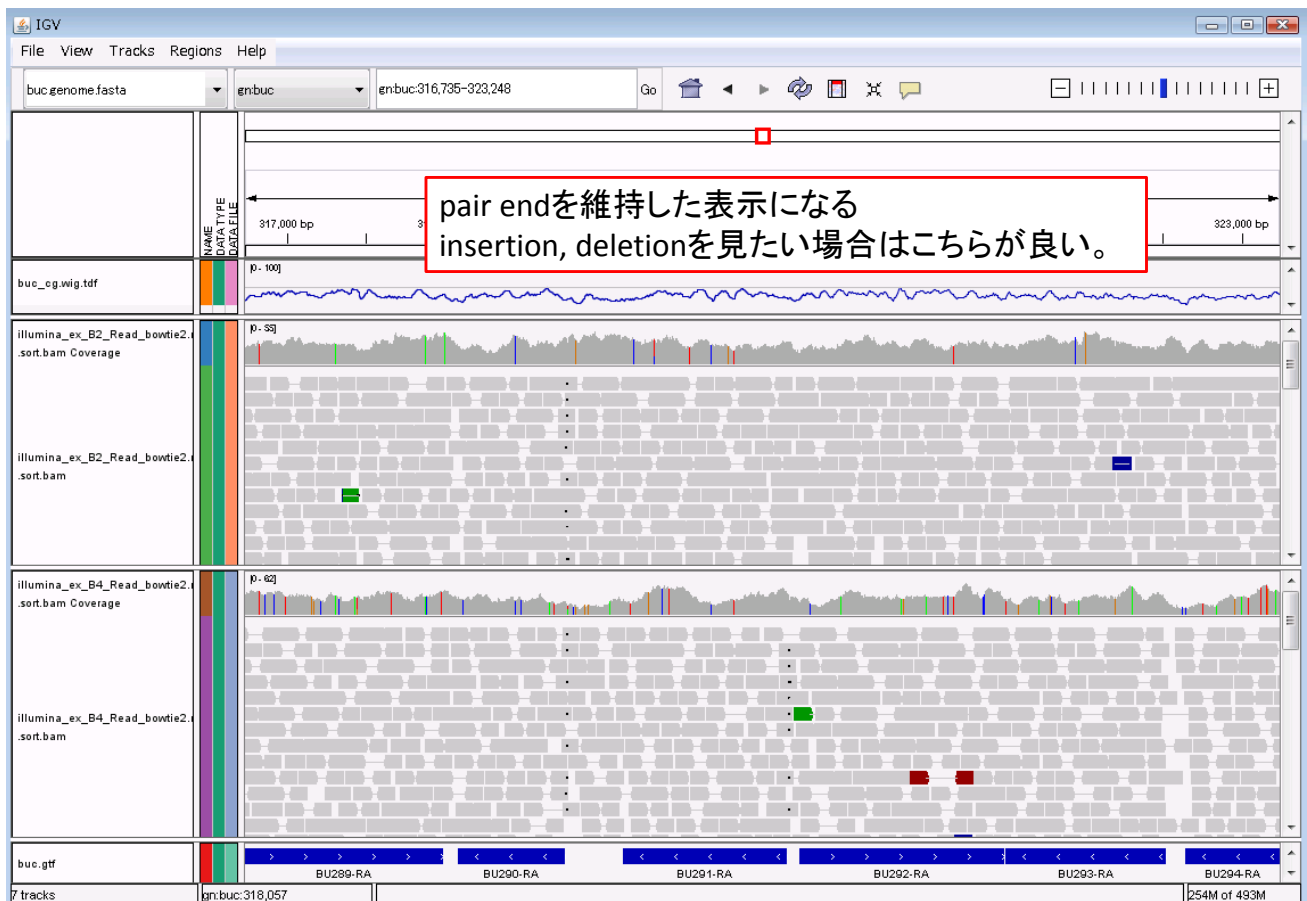
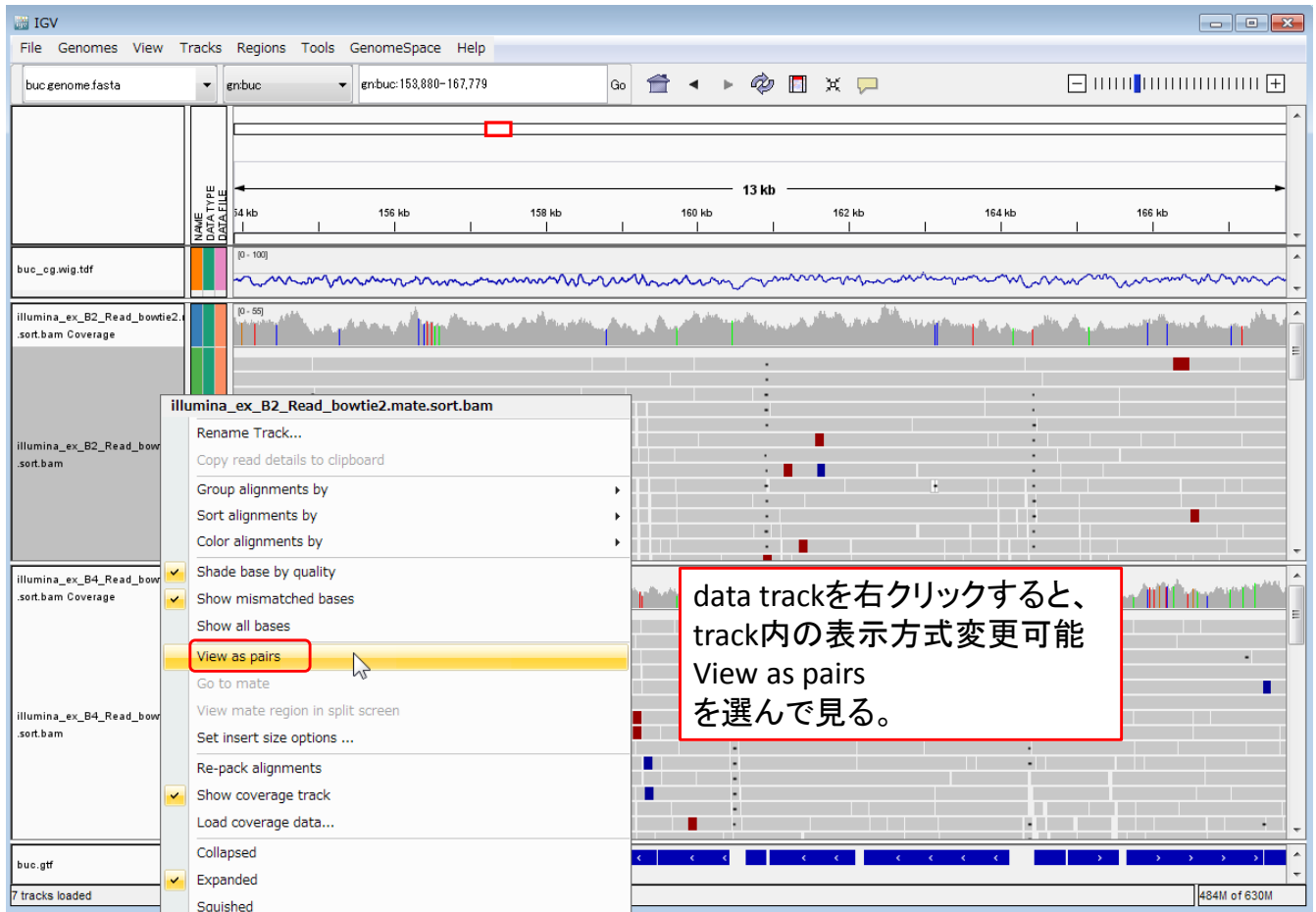
各Trackはドラッグアンドドロップで移動できる

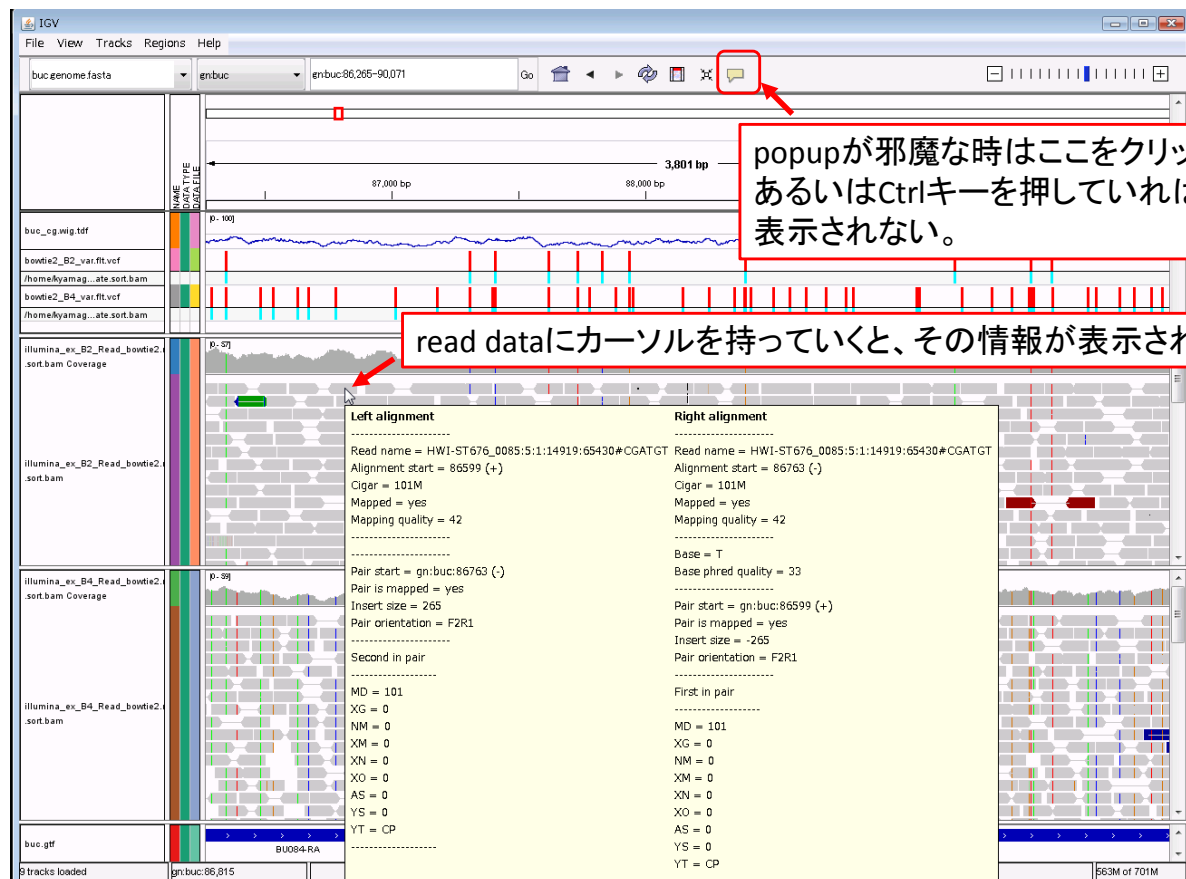
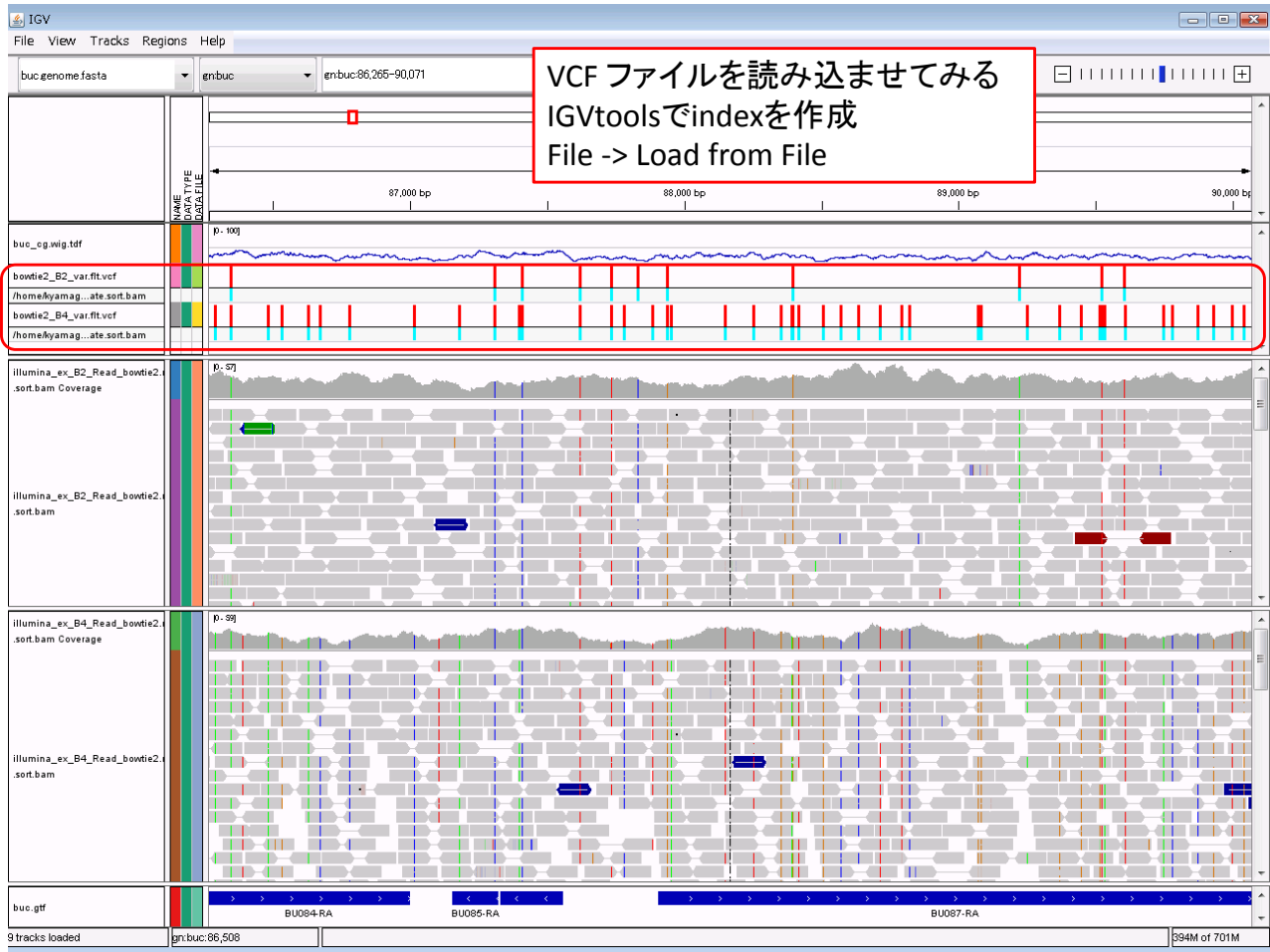












# IGV紹介のまとめ

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
- ・自分で見るとためにもよし、人に見せるためにもよし
- ・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。