

NGS基本ツールIGV

基礎生物学的研究所
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, IGV for iPad, Credits, and Contact. Below these is a search bar and the Broad Institute logo. The main content area has 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 includes a registration link. The Funding section lists funding sources like the National Cancer Institute and the National Institutes of Health. The Citing IGV section provides citation information for publications. At the bottom, the URL <https://www.broadinstitute.org/igv/> is displayed in red.

Integrative Genomics Viewer

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

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 Institute of General Medical Sciences](#) of the [National Institutes of Health](#), 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](#). *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).

<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

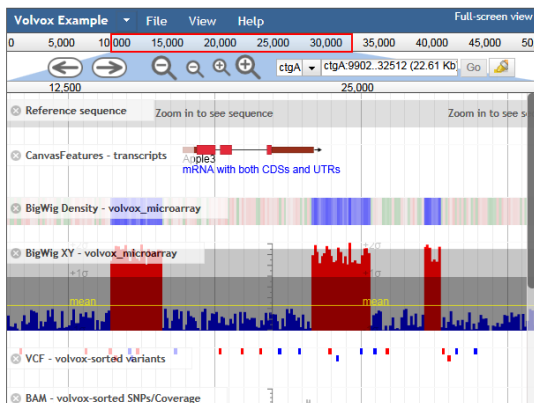
The JBrowse Genome Browser

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 Finkers

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。



Integrative Genomics Viewer

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

Search website

search

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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.

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Integrative Genomics Viewer

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

Search website


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Home



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Integrative Genomics Viewer

- Home
- Downloads
- Documents
 - Hosted Genomes
 - FAQ
 - IGV User Guide**
 - User Interface
 - Starting IGV
 - Navigating
 - Loading a Genome
 - 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
 - External Control of IGV
 - igvtools
 - Motif Finder
 - File Formats
 - Release Notes
 - IGV for iPad
 - Credits
- Contact

Home > IGV User Guide

IGV User Guide

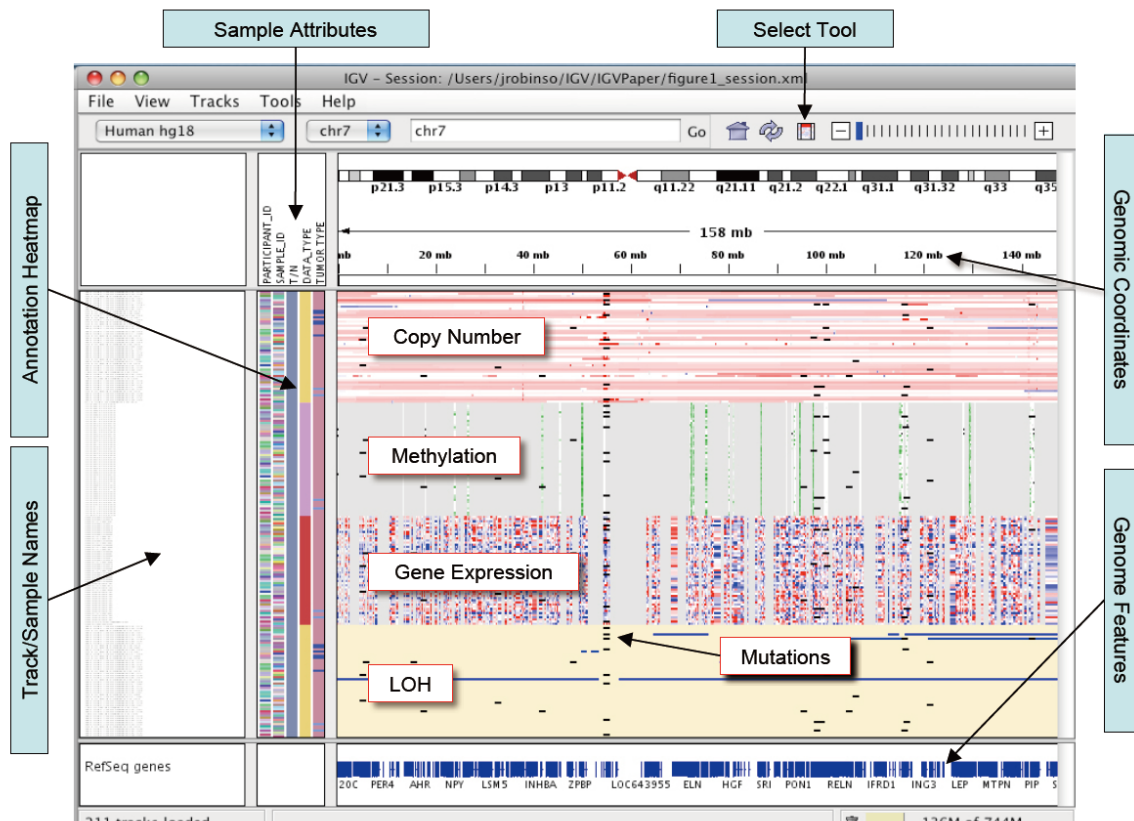
This guide describes the Integrative Genomics Viewer (IGV).

- 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](#)
- [Loading a Genome](#)
- [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](#)
- [External Control of IGV](#)
- [Motif Finder](#)
- [igvtools](#)

[User Interface >](#)



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



Integrative Genomics Viewer

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

Search website

search

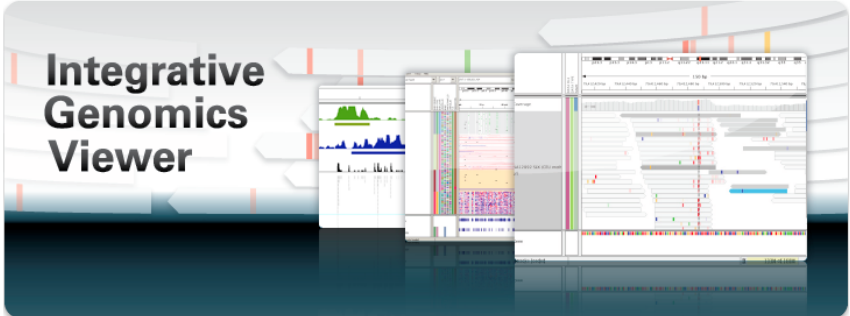
[Broad Home](#)

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Home



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レジストレーションが必要

Downloads

Integrative Genomics Viewer (IGV) (Version 2.3)

Install IGV

Options for installing and running IGV:

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 IGV from the command line.

Note: IGV 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 "igv.bat", it might appear as just "igv" 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 "jnlp" file, then right-clicking on the saved "jnlp" 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 "jnlp" 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"

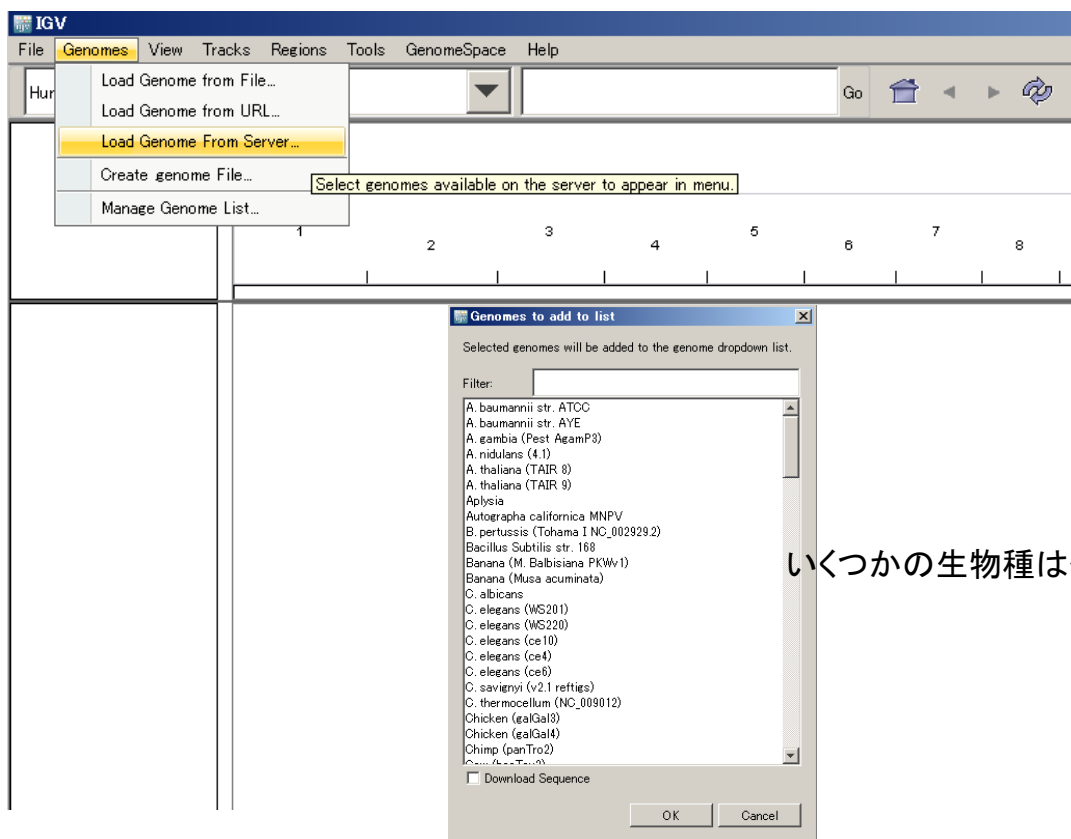
Windows users: To run with more than 1.2 GB of memory you must install 64-bit Java. **Most Windows installs do not include 64-bit Java by default, even if the operating system is 64-bit.** Attempting to use the 2GB or greater launch options with 32-bit Java will result in the error "could not create virtual machine".

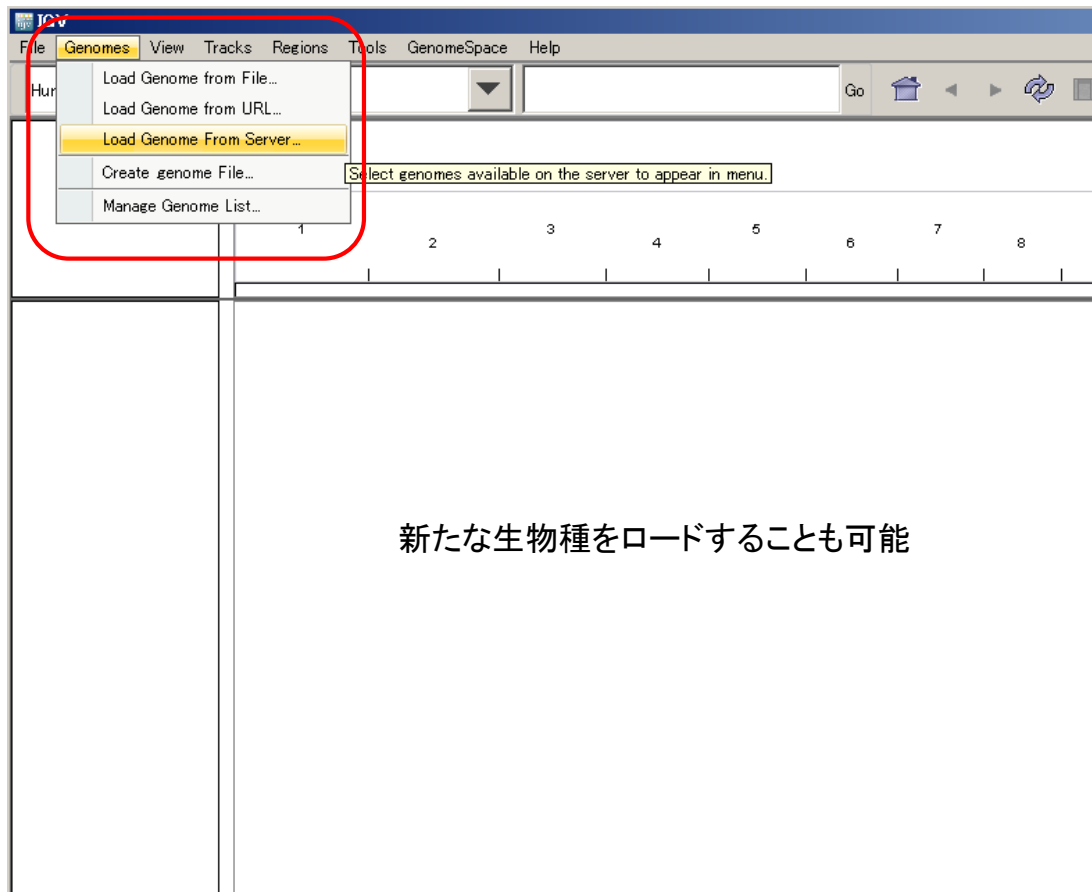
 Launch with 750 MB	 Launch with 1.2 GB Maximum usable memory for Windows OS with 32-bit Java.	 Launch with 2 GB Maximum usable memory for 32-bit MacOS.	 Launch with 10 GB For large memory machines with 64-bit Java.
---	---	---	---

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





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

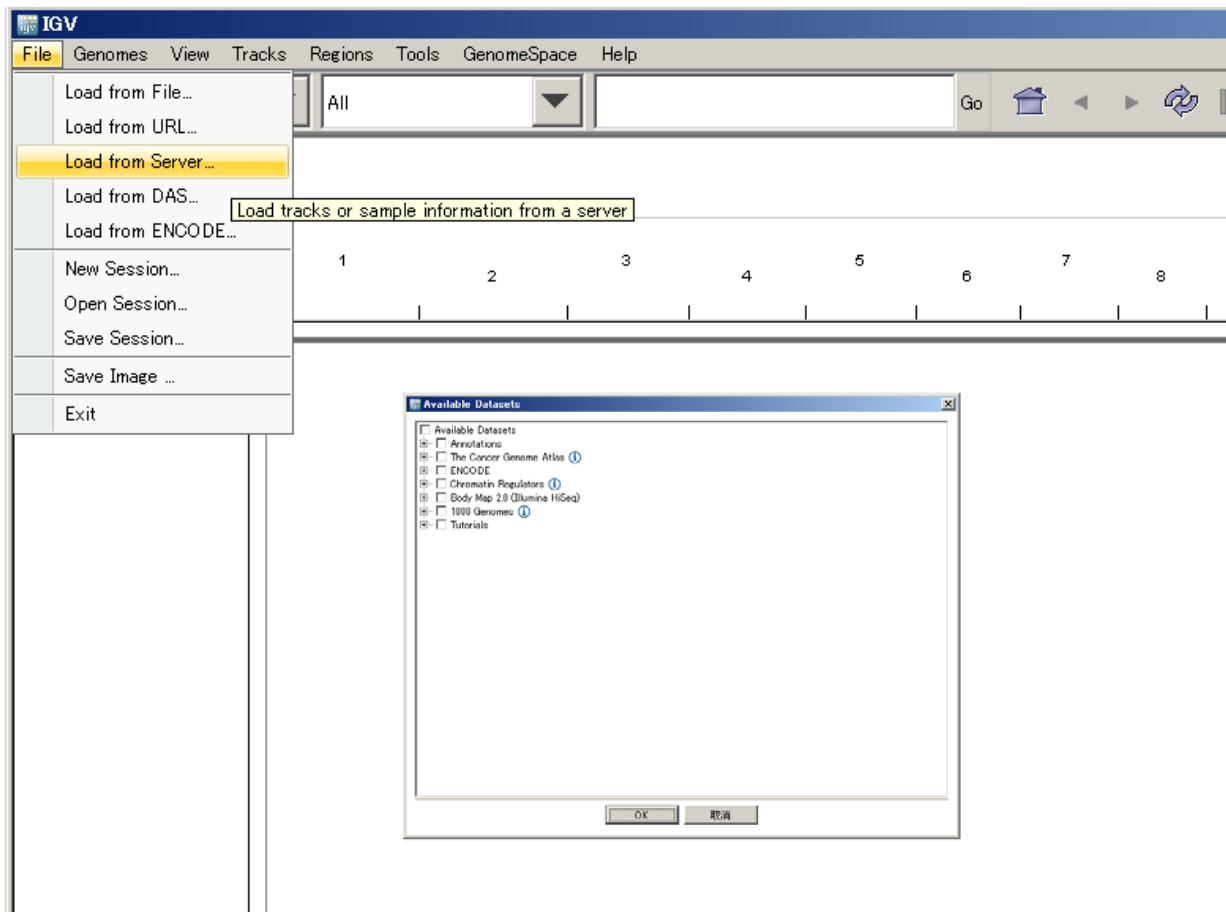
- [File Extension Identifies Format](#)
- [Recommended File Formats](#)
- BAM
- BED
- BedGraph
- bigBed
- bigWig
- Birdsuite Files
- broadPeak
- CBS
- CN
- Custom File Formats
- Cytoband
- FASTA
- GCT
- genePred
- GFF/GTF
- GISTIC
- Goby
- GWAS
- IGV
- LOH
- MAF (Multiple Alignment Format)
- MAF (Mutation Annotation Format)
- Merged BAM File
- MUT
- narrowPeak
- PSL
- RES
- SAM
- Sample Information
- SEG
- SNP
- TAB
- TDF
- Track Line
- Type Line
- VCF
- WIG
- chrom.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
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 metadata	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。

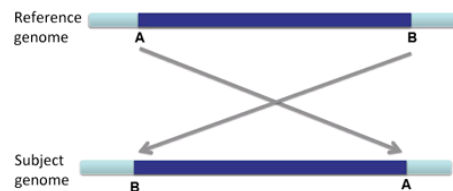
バッチ処理

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。

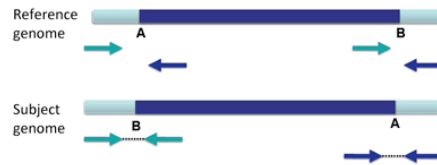
```
new
load myfile.bam
snapshotDirectory mySnapshotDirectory
genome hg18
goto chr1:65,289,335-65,309,335
sort position
collapse
snapshot
goto chr1:113,144,120-113,164,120
sort base
collapse
snapshot
```

Inversions

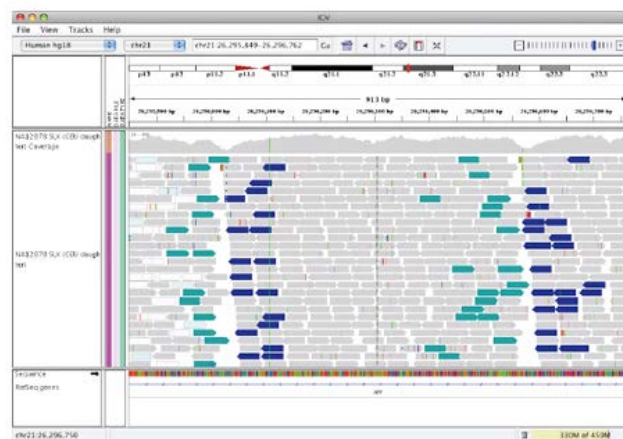
An inversion is a large section of DNA that is reversed in the subject genome compared to the reference genome.



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:

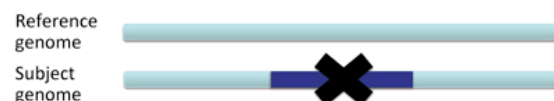
- deletions
- insertions
- inter-chromosomal rearrangements

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:

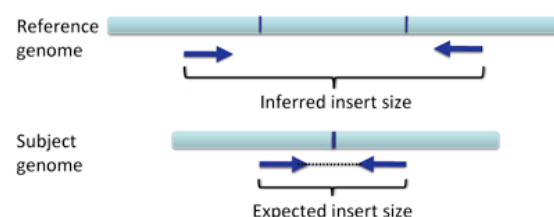
-  for an insert that is larger than expected
-  for an insert that is smaller than expected
-  for paired end reads that are coded by the chromosome on which their mates can be found

Deletions

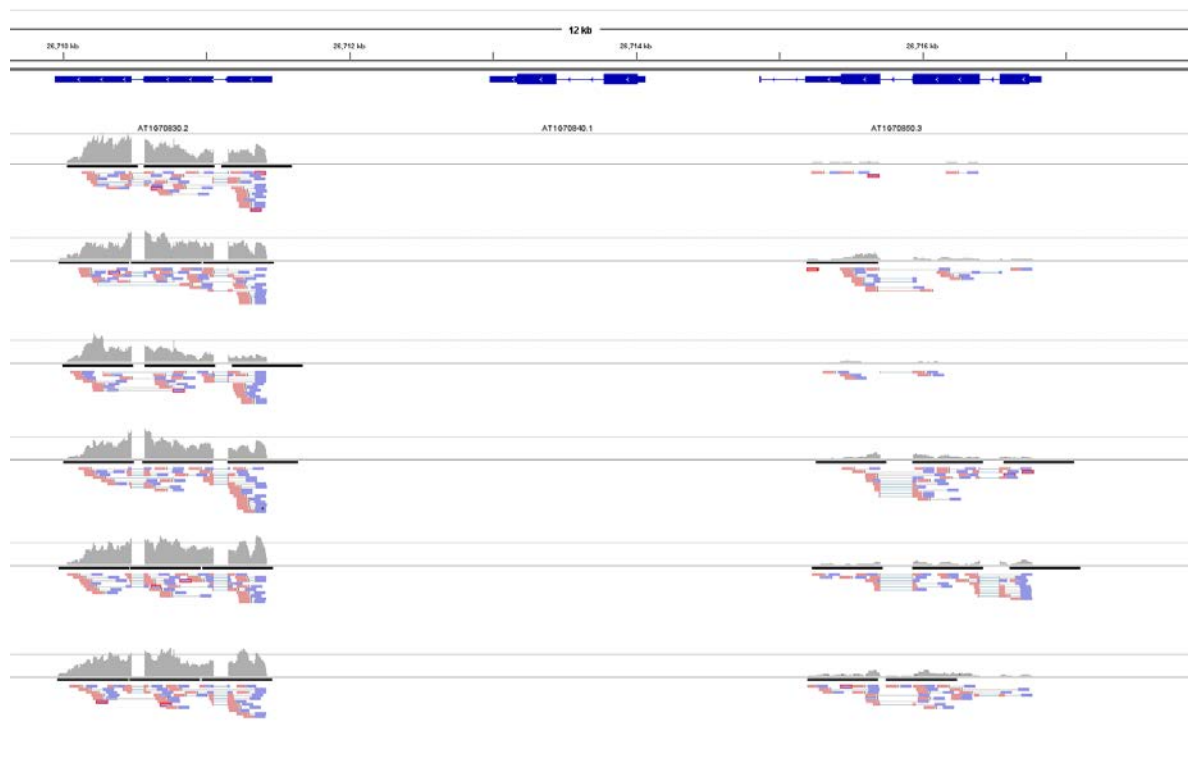
A deletion is a large section of DNA that is absent in the subject genome compared to the reference genome.



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.

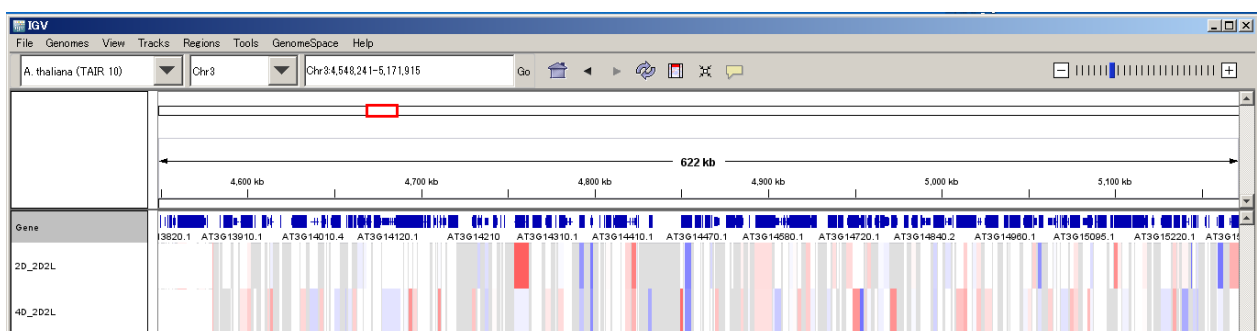


RNA-Seqのデータ表示させる

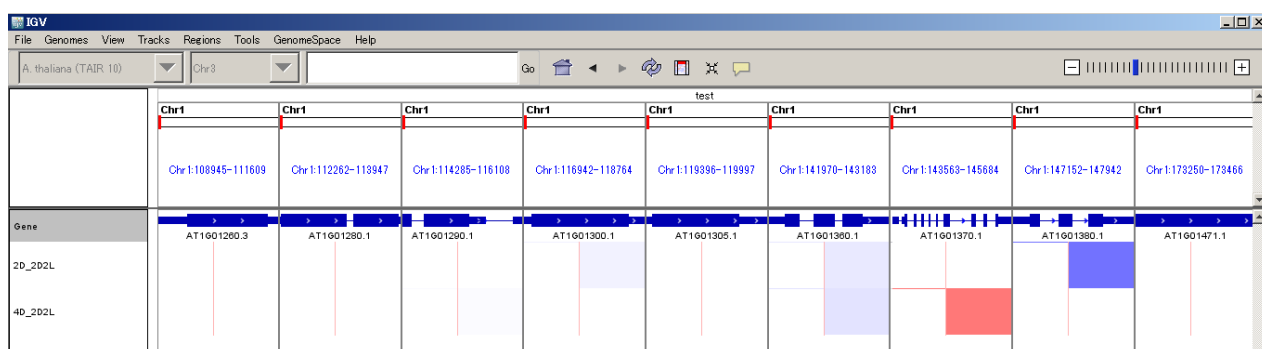
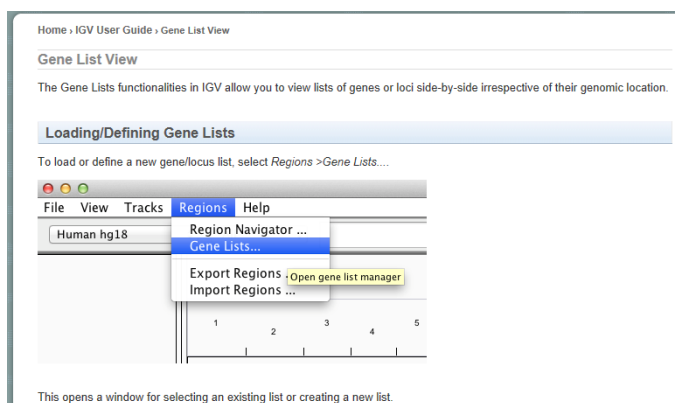


GCTファイルでgene ローカスの発現情報を図示

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

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Home > Downloads

Downloads

Integrative Genomics Viewer (Version 2.3)

IGV can either be downloaded on to the local machine, or launched via Java Webstart. As described below, the Java Webstart option is not recommended for Mac users.

Downloads

Mac users: Download and unzip the following archive, then double-click the IGV application to run it. The application can be moved to the "Applications" folder, or anywhere else.

- [IGV_2.3.31.app.zip](#)

Windows and Linux users: Download and unzip the archive in a folder of your choosing. IGV is launched from a command prompt, follow instructions in the "readme" file. Windows users, use the "igv.bat". On Linux, use "igv.sh".

- [IGV_2.3.31.zip](#)

Java Webstart

The buttons below use Java Webstart (JWS) to install and launch IGV directly from our website.

Mac Users: The Java Webstart (JWS) option is not recommended for Mac users. Using it requires that you set Gatekeeper security to its lowest level, and its possible that even this will not be enough. If you do use the JWS buttons below on MacOS Java 7 is required.

Chrome: Chrome does not automatically launch the Java Webstart 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.

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".

Launch	Launch	Launch	Launch
Launch with 750 MB	Launch with 1.2 GB	Launch with 2 GB	Launch with 10 GB
	Maximum usable memory for Windows OS with 32-bit Java.	Maximum usable memory for 32-bit MacOS.	For large memory machines with 64-bit Java.

[Development Snapshot Build](#). Latest development snapshot, built at least nightly.

[Archived Versions](#)

igvtools

Utilities for preprocessing data files.

- [igvtools_2.3.31.zip](#)

Source Code

Source code repository is hosted at GitHub:

- <https://github.com/broadinstitute/IGV/>

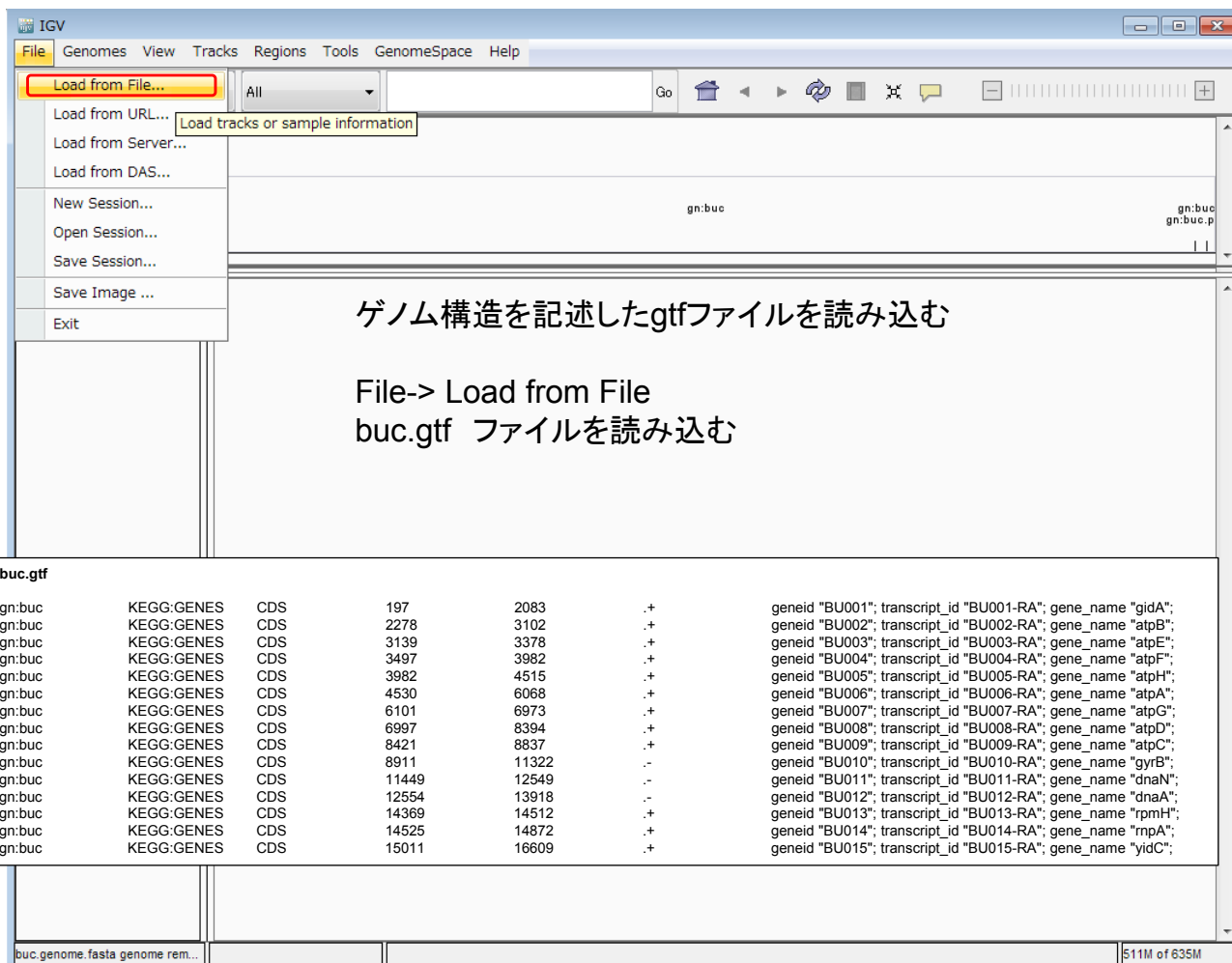
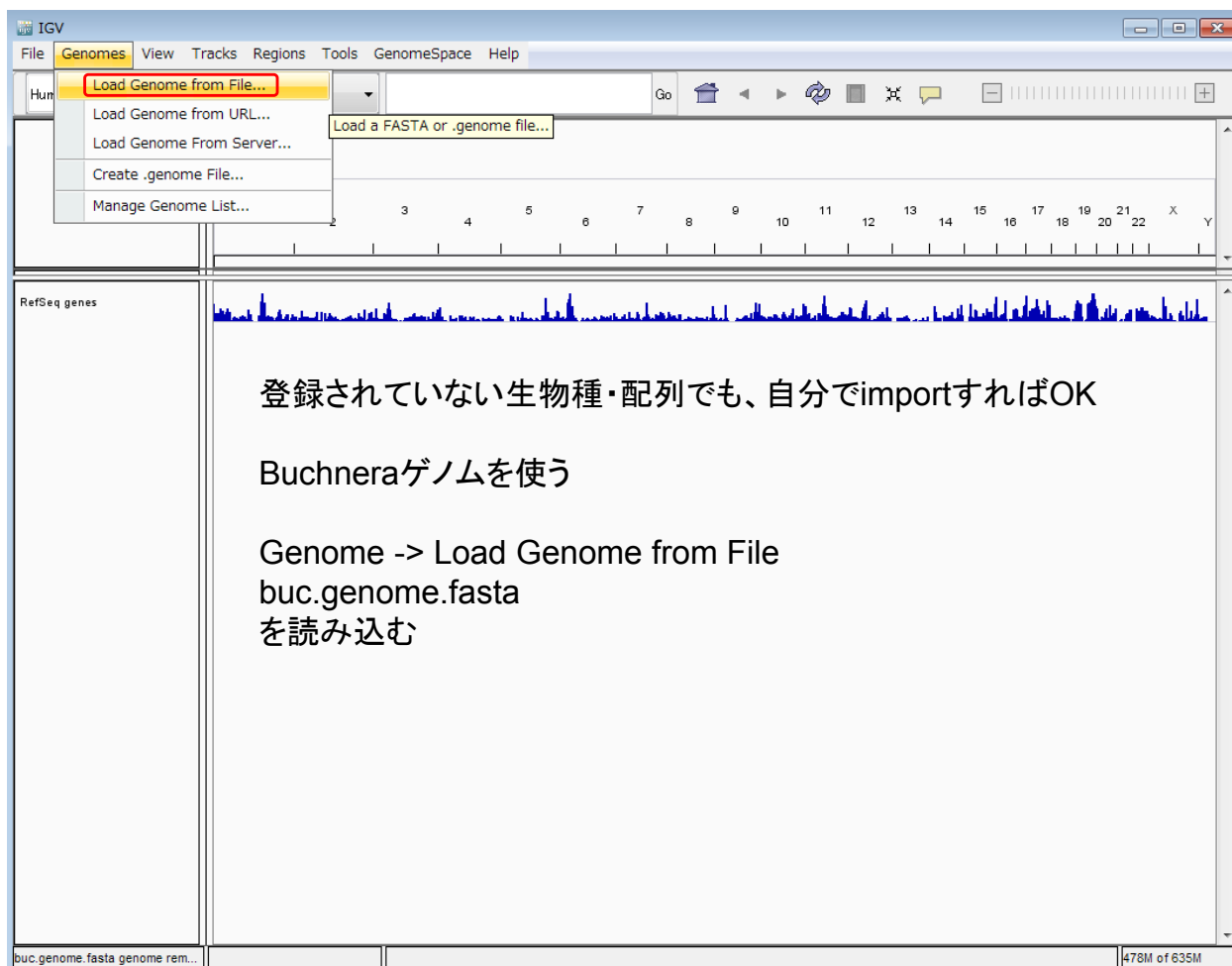
Source distribution archive:

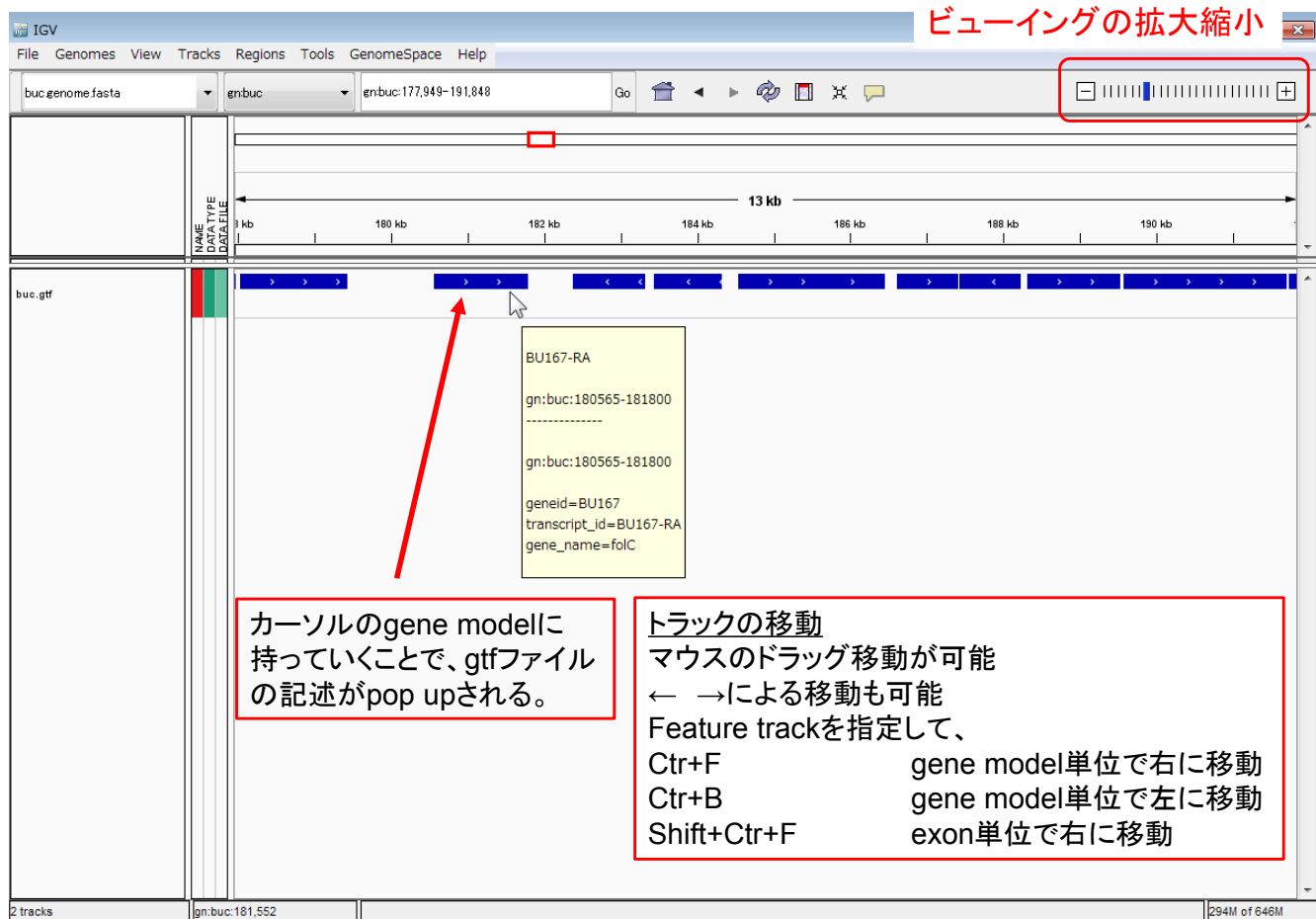
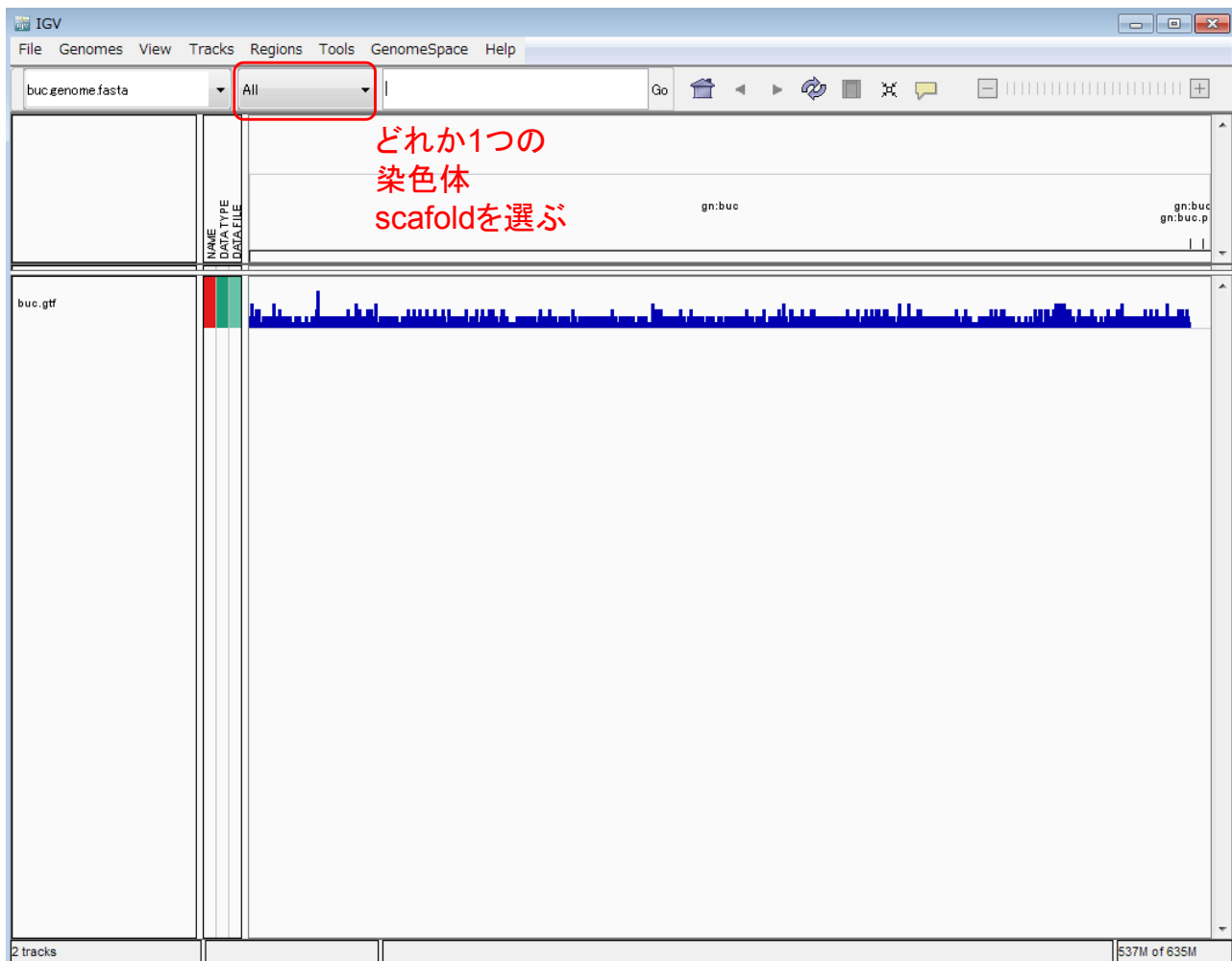
- [v2.3.31.zip](#)

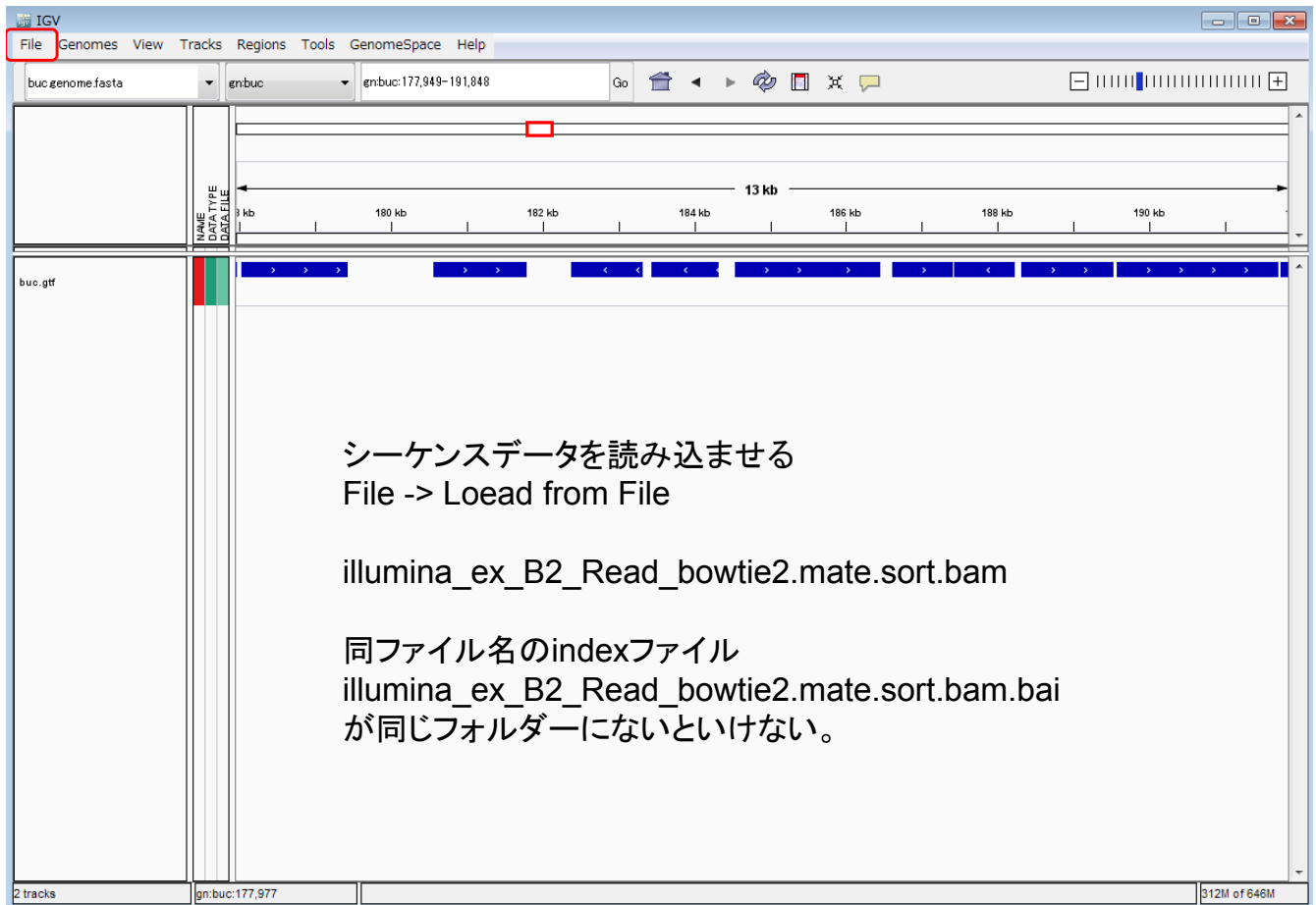
IGVの使用法を学ぶと共に
先のファイルフォーマットも
確認しよう

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
```



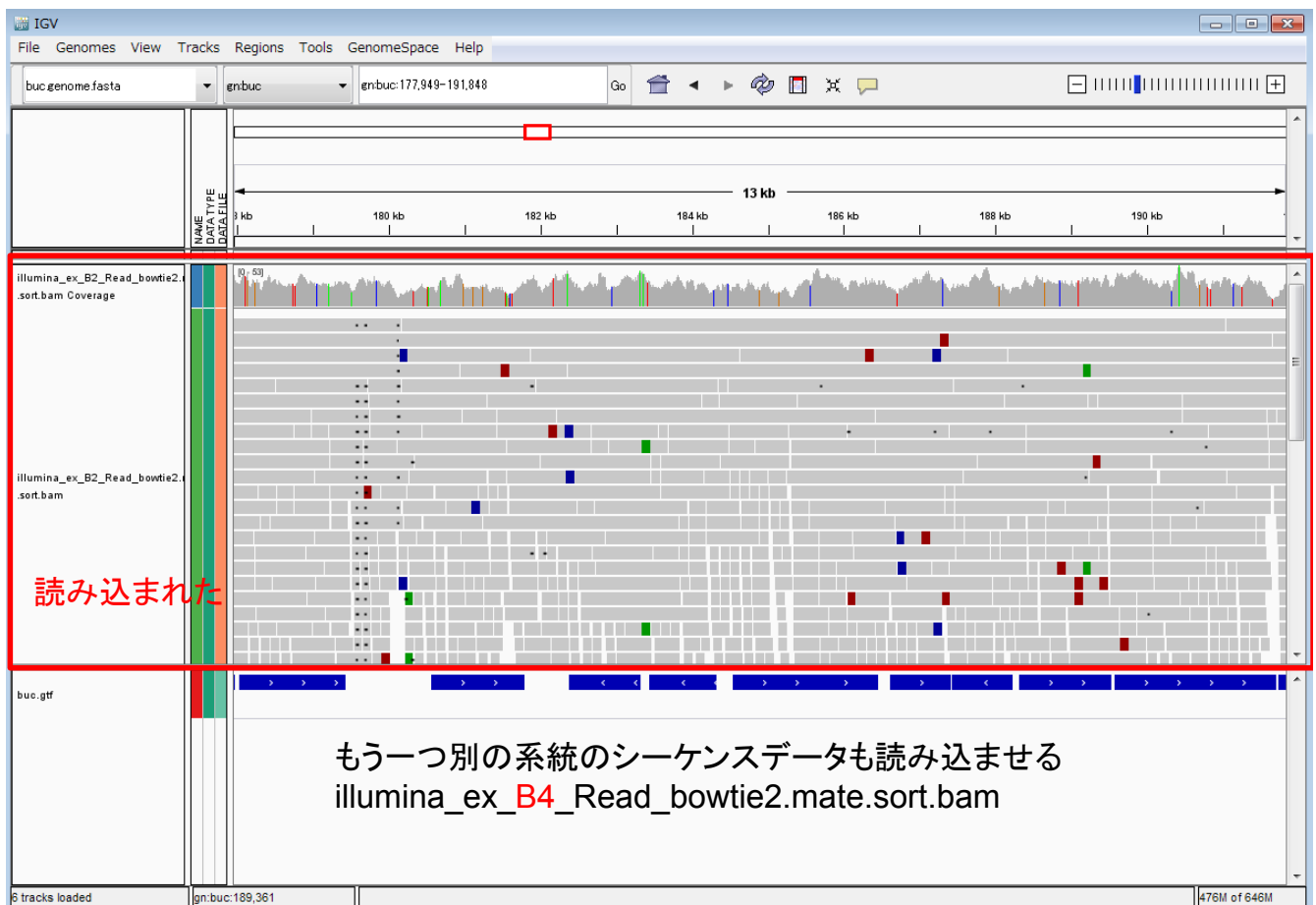




シーケンスデータを読み込ませる
File -> Load from File

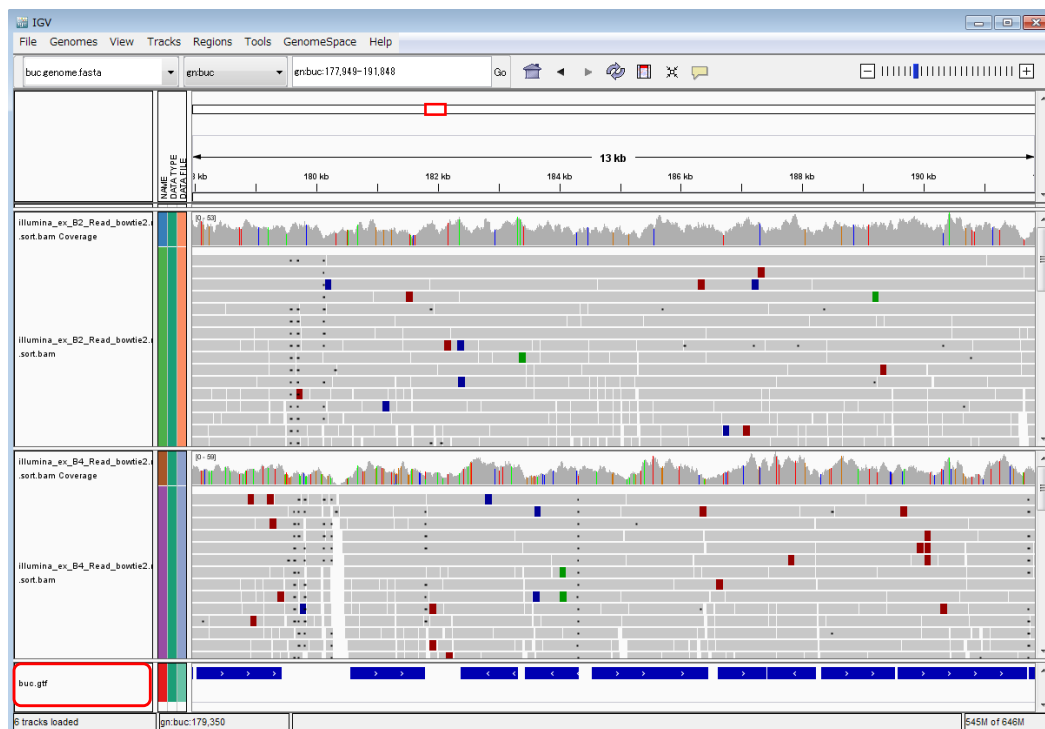
illumina_ex_B2_Read_bowtie2.mate.sort.bam

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。



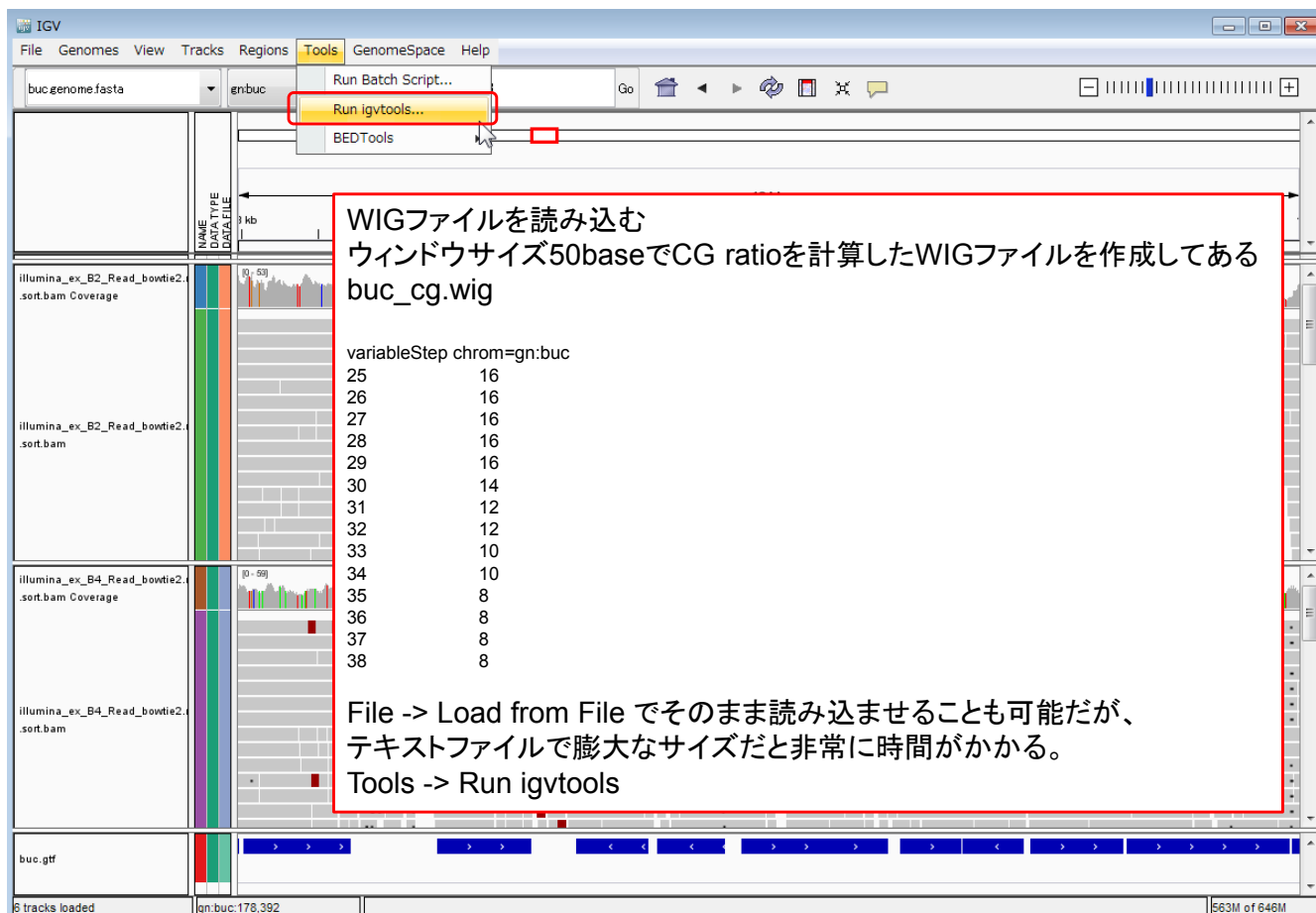
読み込まれた

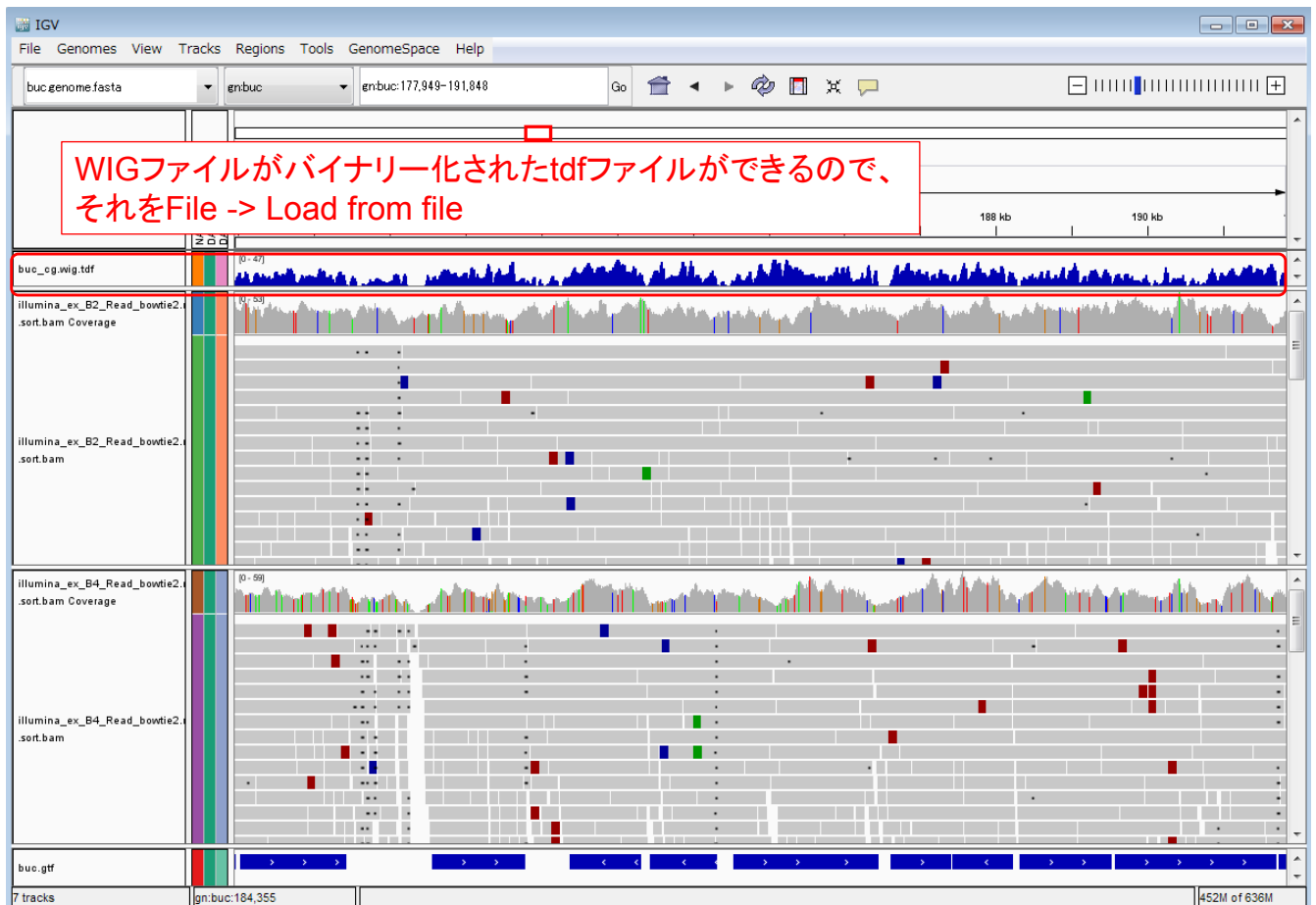
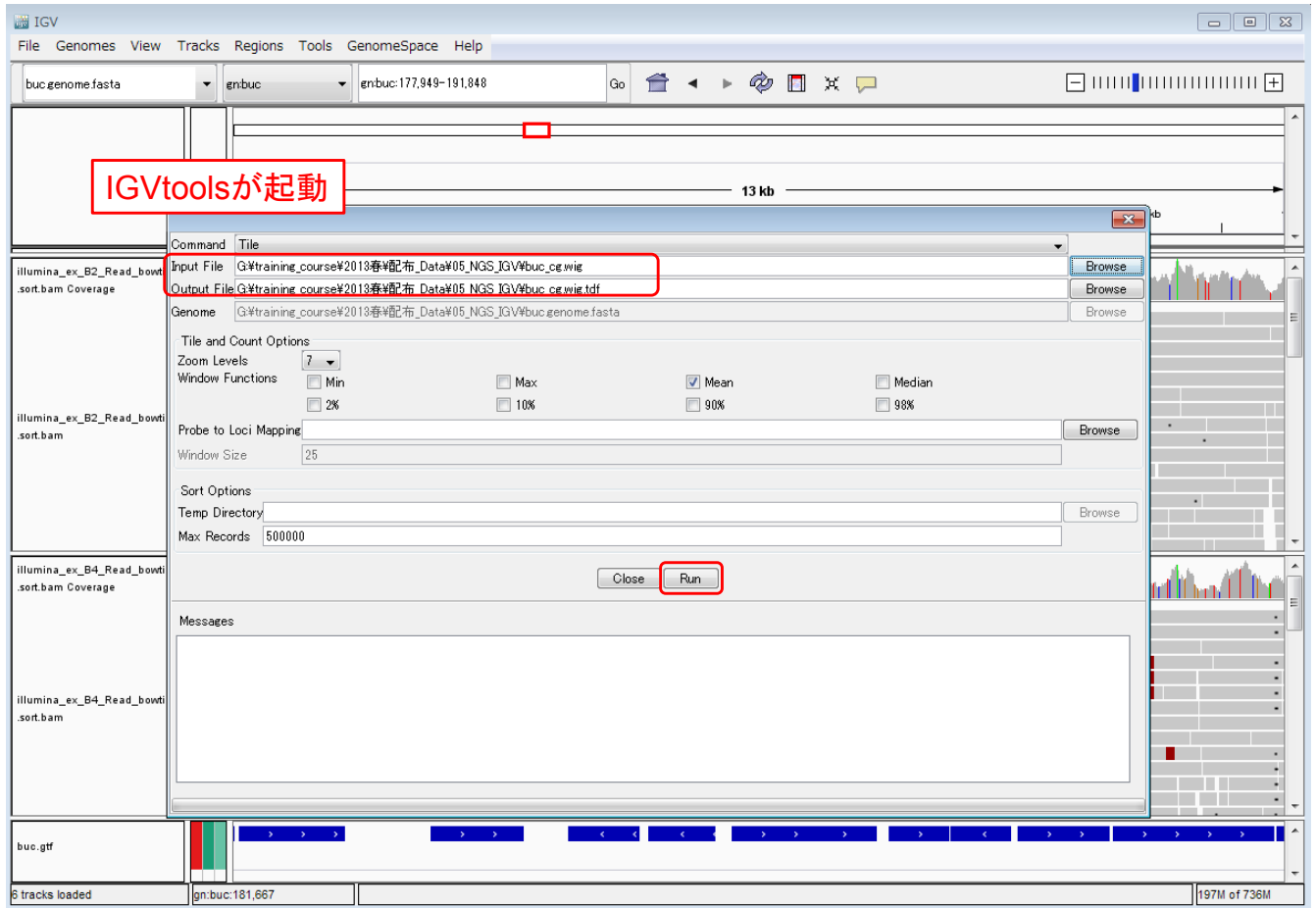
もう一つ別の系統のシーケンスデータも読み込ませる
illumina_ex_B4_Read_bowtie2.mate.sort.bam

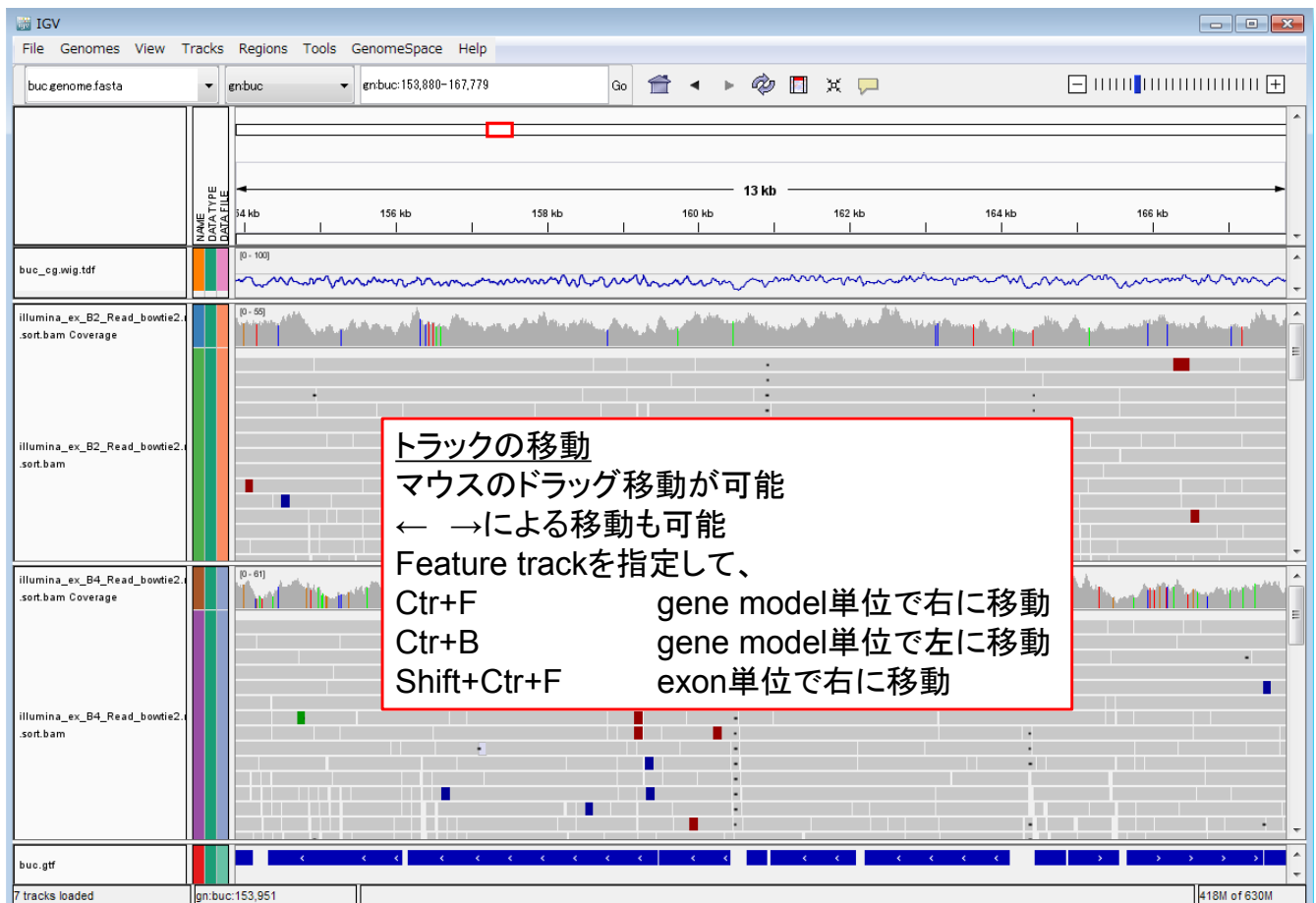
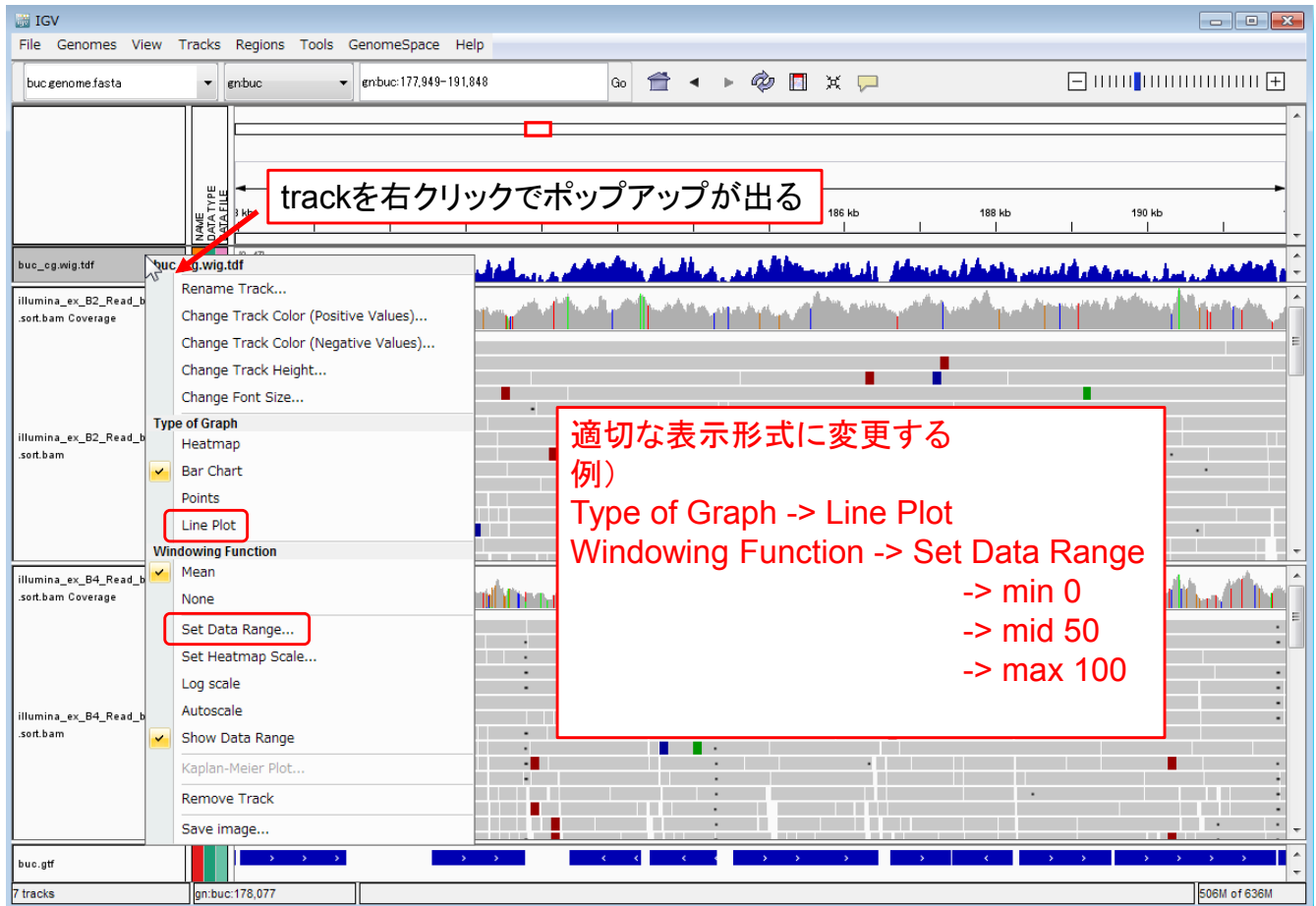


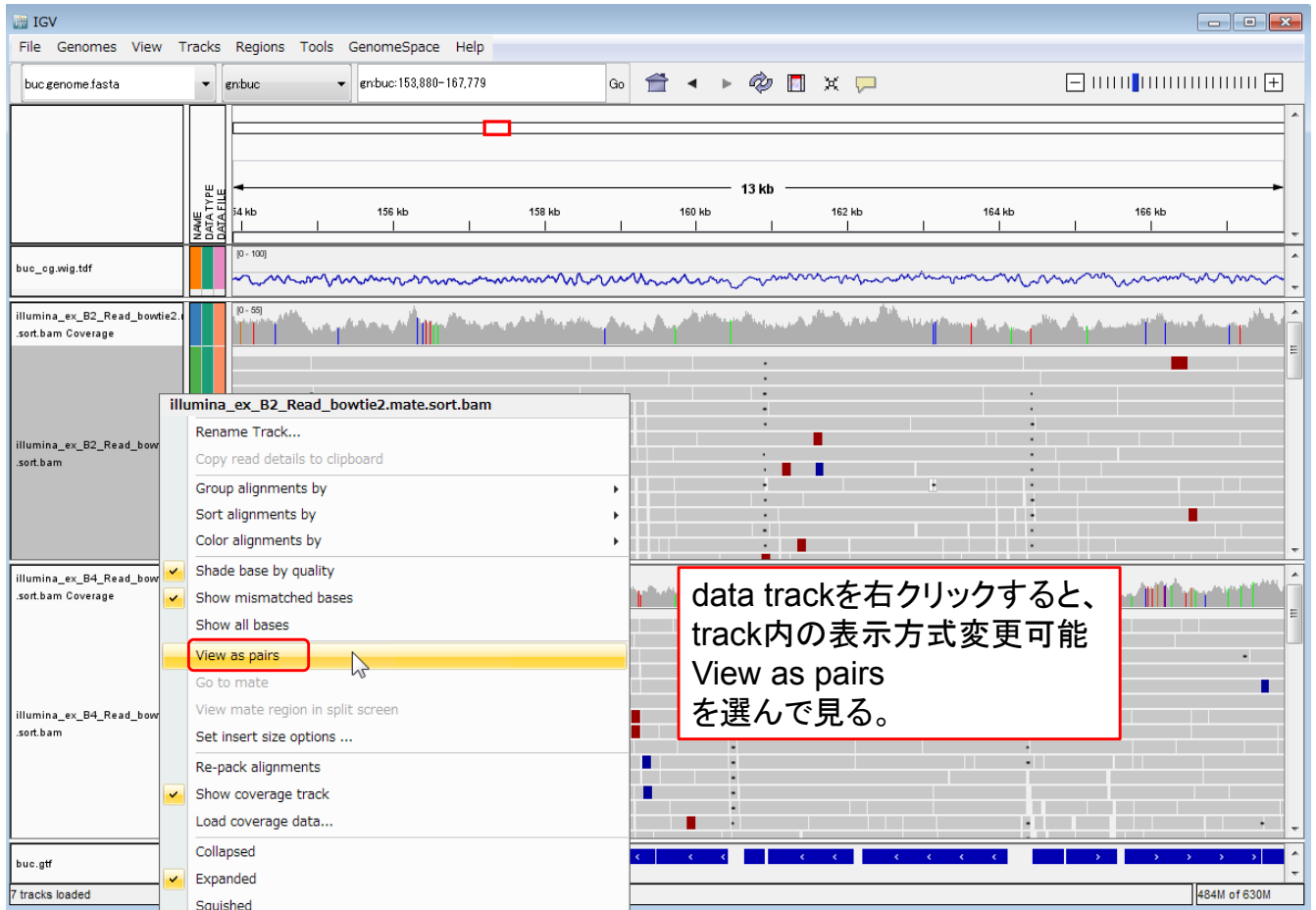
こういう所をクリックしたまま移動させることで幅を変更

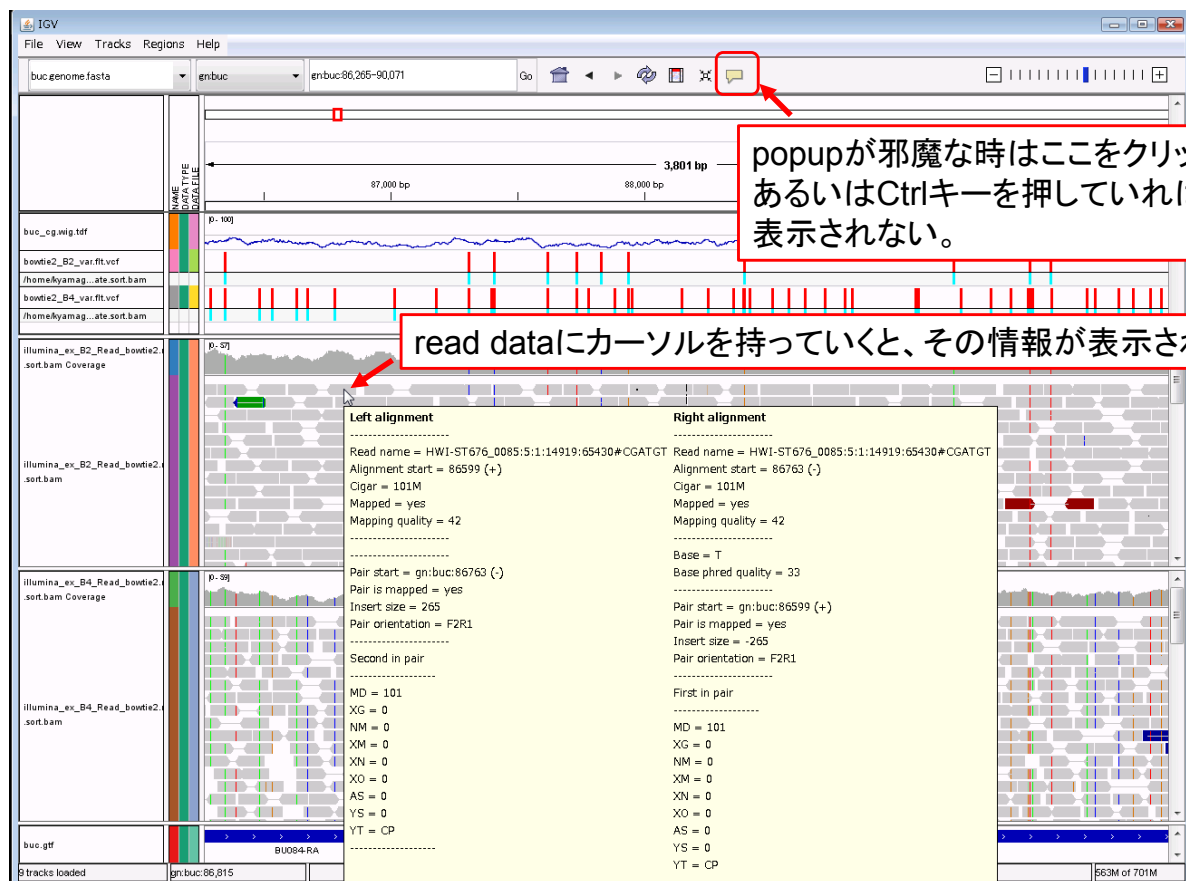
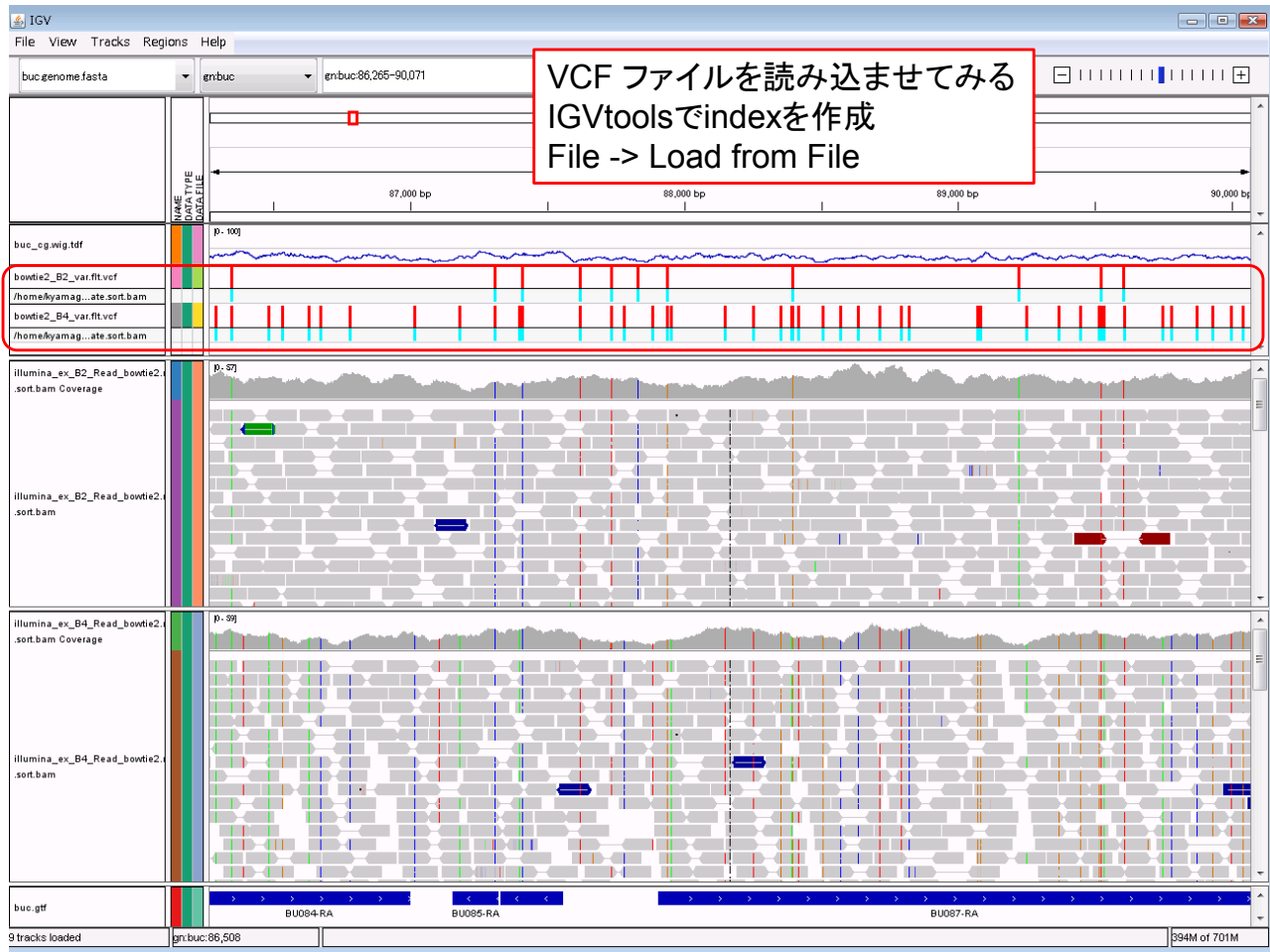
各Trackはドラッグアンドドロップで移動できる











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。