

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

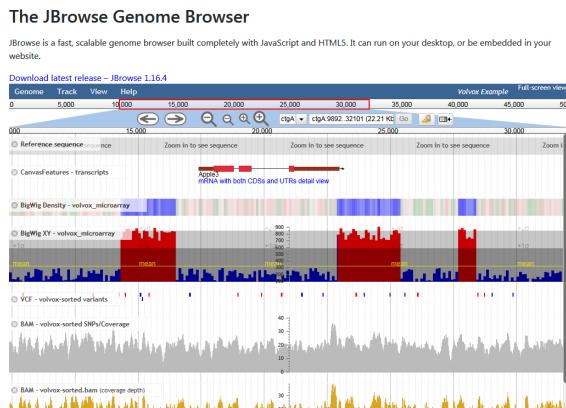
The screenshot shows the homepage of the Integrative Genomics Viewer (IGV) website. The left sidebar contains links for Home, Downloads, Documents (including IGV User Guide, Tutorial Videos, File Formats, Hosted Genomes, FAQ, Release Notes, Credits, and Contact), and a search bar. The main content area features a large banner image of the IGV software interface, which displays multiple tracks of genomic data. Below the banner, there are sections for Overview, Citing IGV, Download IGV, and Funding. The Overview section describes IGV as a high-performance visualization tool for exploring genomic datasets. The Citing IGV section provides citation information. The Download IGV section offers links to download the desktop application and web components. The Funding section notes the support from the National Cancer Institute (NCI) and other organizations.

<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



後者はコミュニティーで広く利用、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璧

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ため、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

誰もが簡単に使えるものが良い。

The screenshot shows the IGV website. On the left is a sidebar with a logo, navigation links (Home, Downloads, Documents, etc.), and a search bar. The main content area features a large banner with the text "Integrative Genomics Viewer" and a screenshot of the software interface. Below the banner are sections for "Overview", "Citing IGV" (with a red box highlighting it), "Download IGV", and "Funding".

Citing IGV

To cite your use of IGV in your publication, please reference one or more of:

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). (Free PMC article [here](#)).

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).

James T. Robinson, Helga Thorvaldsdóttir, Aaron M. Wenger, Ahmet Zehir, Jill P. Mesirov. [Variant Review with the Integrative Genomics Viewer \(IGV\)](#). *Cancer Research* 77(21) 31–34 (2017).

The screenshot shows a journal article from Nature Biotechnology. The header includes the journal title, a login link, a search bar, and navigation links for previous and next abstracts. The main content area has a green background with a circuit board graphic. It displays the article title, authors, date, and a summary. To the right are columns for journal navigation, a call to action for subscriptions, citation links, science jobs, and faculty positions.

Abstract

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

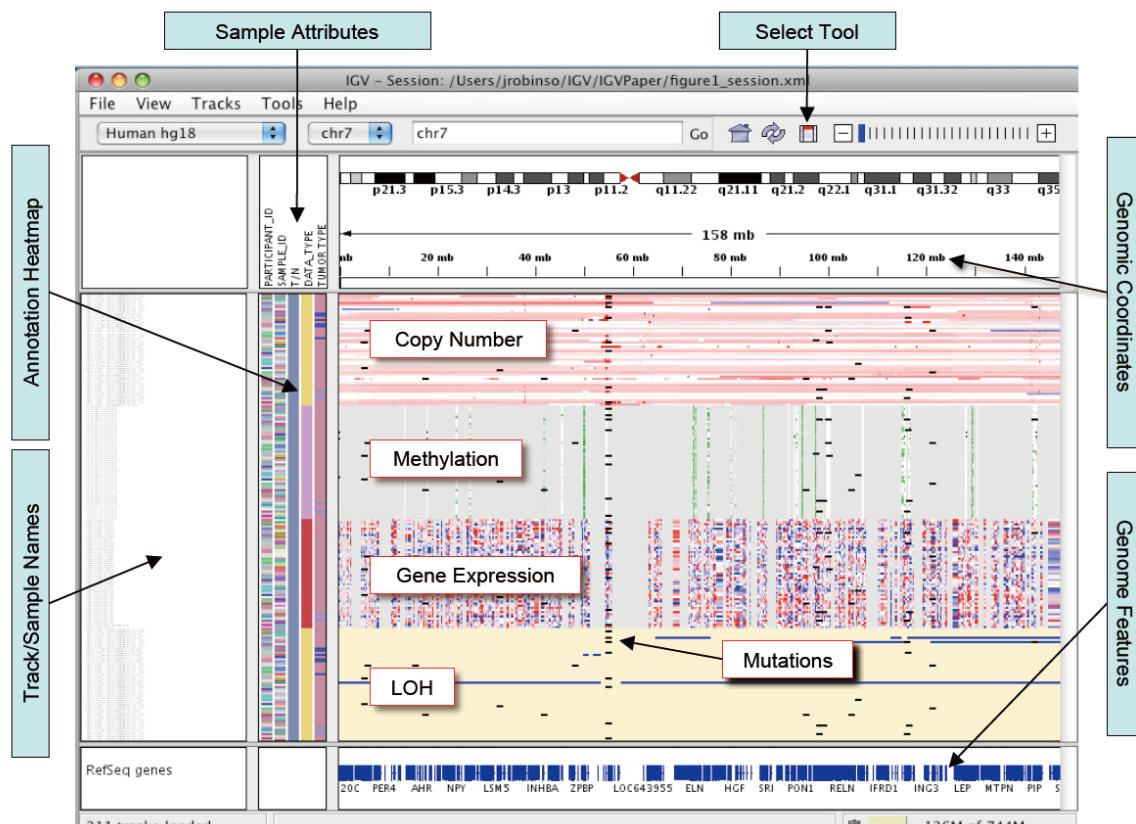
To the Editor:

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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Citations to this article
Crossref (10) Scopus (12) Web of Science (0)

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Faculty Position
Harvard Medical School
Ramalingaswami Re-Entry Fellowship
Ministry of Science & Technology, Government of India



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

IGV Integrative Genomics Viewer

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Search website

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Home

Integrative Genomics Viewer

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.

IGV is available in multiple forms, including:

- the original IGV - a Java desktop application,
- IGV-Web - a web application,
- igv.js - a JavaScript component that can be embedded in web pages ([for developers](#))

This site is focused on the IGV desktop application. See <https://igv.org> for links to all forms of IGV.

Download IGV

Download the IGV desktop application and igvtools.

Note that the IGV-Web application at <https://igv.org/app> runs in a web browser and requires no downloads. Click on the Help link in the app for more information.

Citing IGV

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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). (Free PMC article [here](#)).

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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](#).



Integrative Genomics Viewer
ALMEL

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- [Documents](#)

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 - ⊕ Viewing Data
 - ⊕ Viewing Alignments
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 - ⊕ Release Notes
 - ↳ 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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[User Interface >](#)



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Home



Overview

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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

Did you know that there is also an IGV web application that runs only in a web browser, does not use Java, and requires no downloads? See <https://igv.org/app>. Click on the Help link in the app for more information about using IGV-Web.

Install IGV 2.5.x



IGV Mac App

Download and unzip the Mac App Archive, then double-click the IGV application to run it. You can move the app to the Applications folder, or anywhere else.



IGV for Windows

Download and run the installer. An IGV shortcut will be created on the Desktop; double-click it to run the application.



IGV for Linux

Download and unzip the Archive. See the downloaded *readme.txt* for further instructions.



IGV and igvtools to run on the command line (all platforms)

Download and unzip the Archive. **Requires Java 11**. See the downloaded *readme.txt* and *igvtools_readme.txt* for further instructions.

Other IGV Versions

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

[Archived Versions](#), Old releases going back to IGV 2.0.1

If you are looking for the *IGV-Web* application, see <https://igv.org/app>

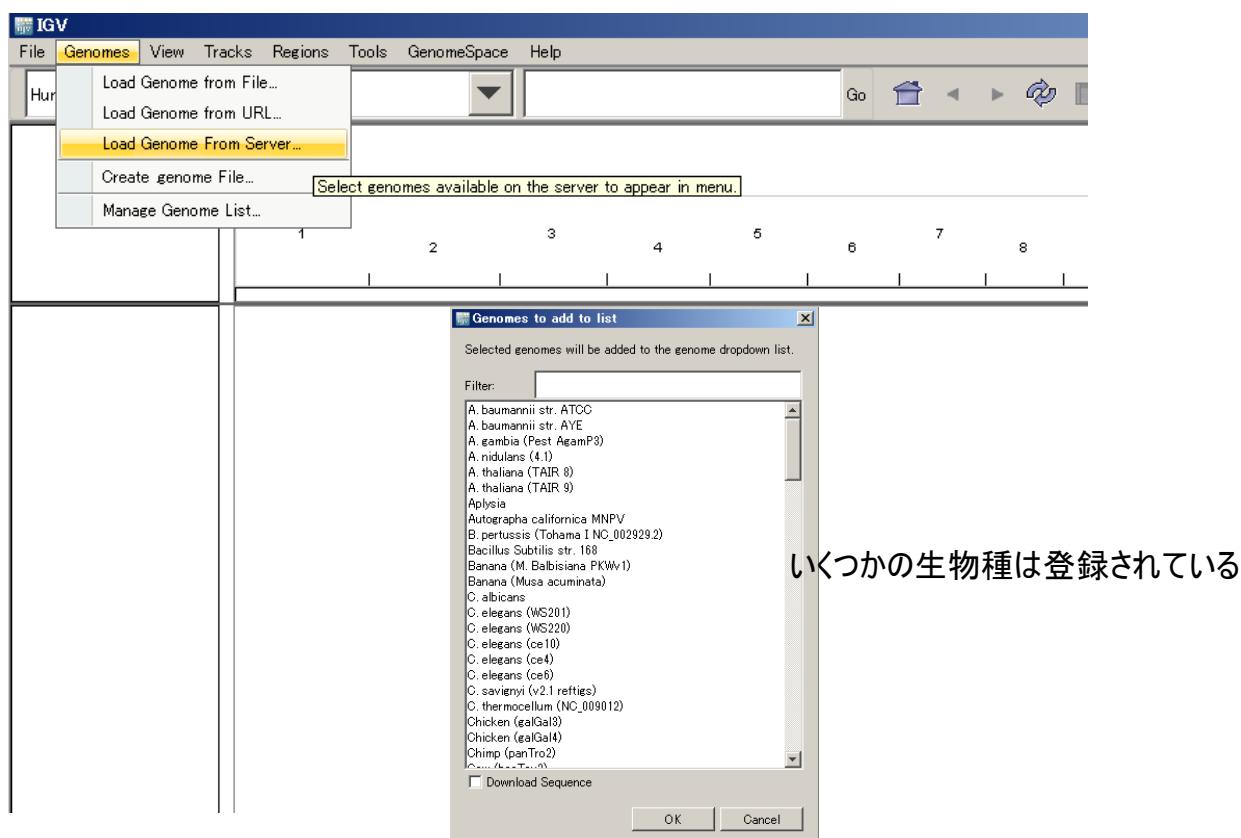
If you are a developer looking for information about the embeddable *igv.js* component, see <https://github.com/gvteam/igv.js>

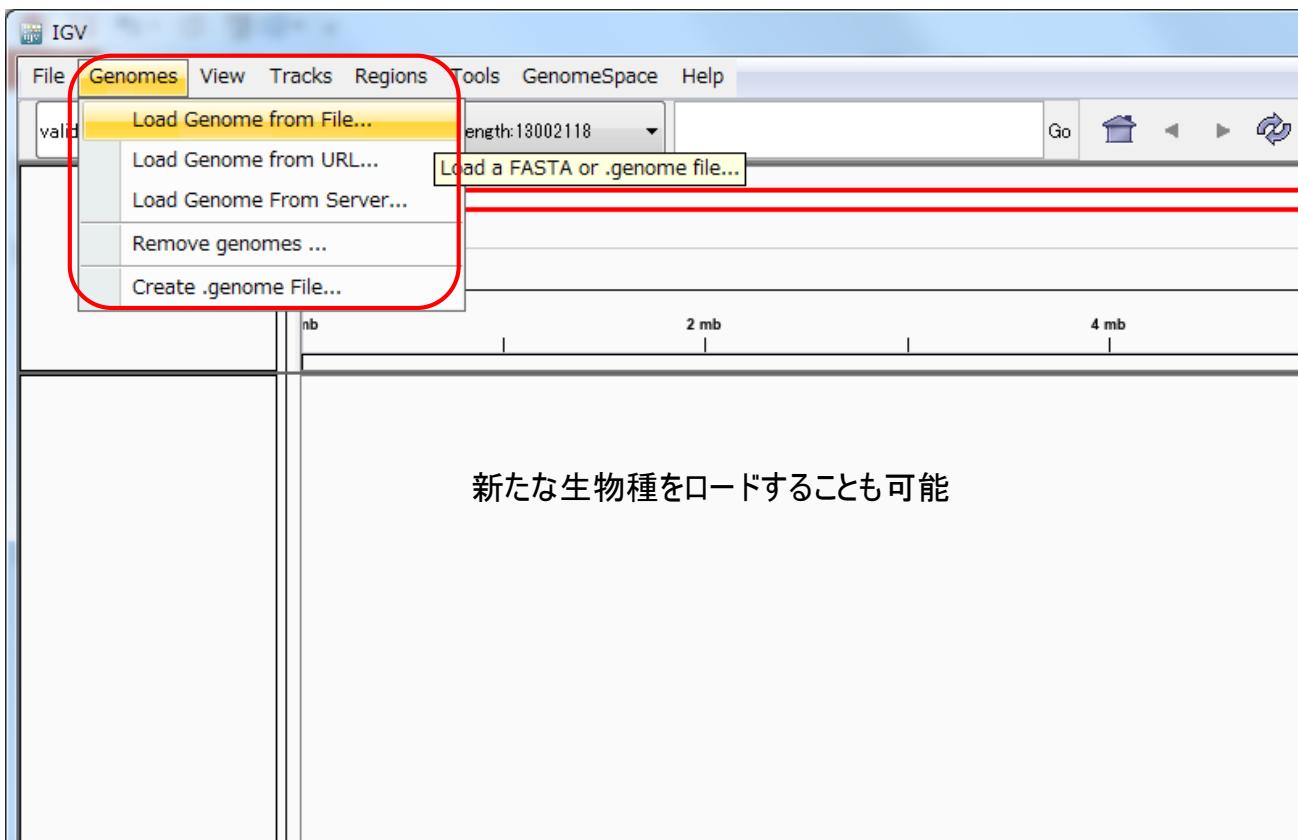
Source Code

The source code repository is hosted on GitHub at <https://github.com/gvteam/igv/>

License

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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。

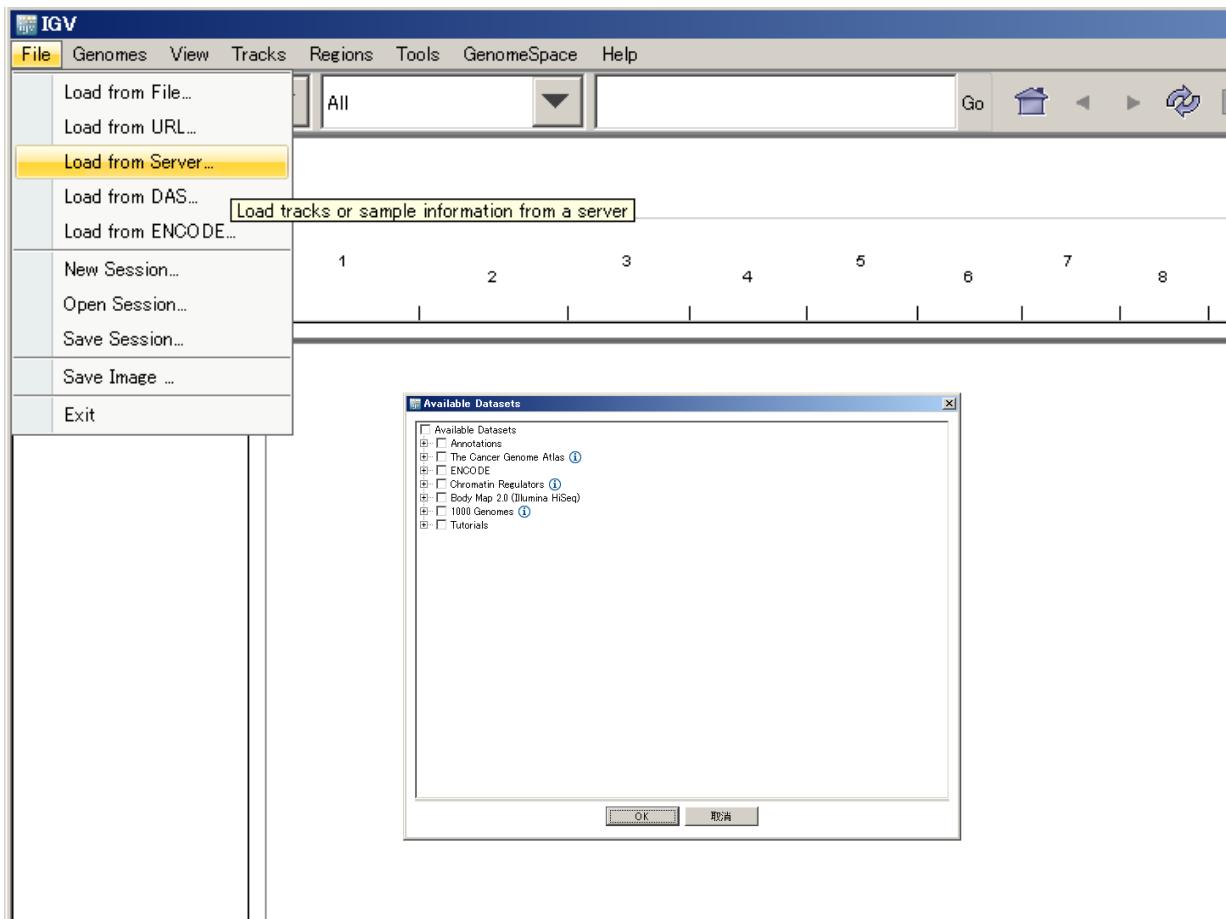
File Formats
▪ File Extension Identifies Format
▪ Recommended File Formats
▪ BAM
▪ BED
▪ BedGraph
▪ bigBed
▪ bigWig
▪ Birdsuite Files
▪ broadPeak
▪ CBS
▪ Chemical Reactivity Probing Profiles
▪ chrom.sizes
▪ CN
▪ Custom File Formats
▪ Cytoband
▪ FASTA
▪ GCT
▪ CRAM
▪ genePred
▪ GFF/GTF
▪ GISTIC
▪ Goby
▪ 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 metadatadata	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。

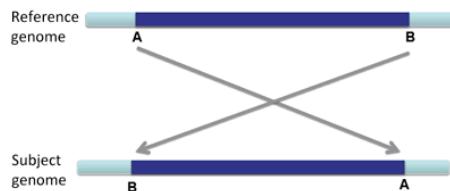
バッチ処理

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。

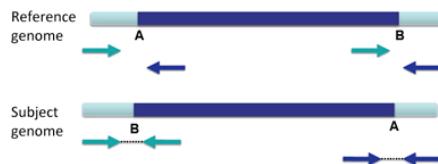
```
new
load myfile.bam
snapshotDirectory mySnapshotDirectory
genome hg18
goto chr1:65,289,335-65,309,335
sort position
collapse
snapshot
goto chr1:113,144,120-113,164,120
sort base
collapse
snapshot
```

Inversions

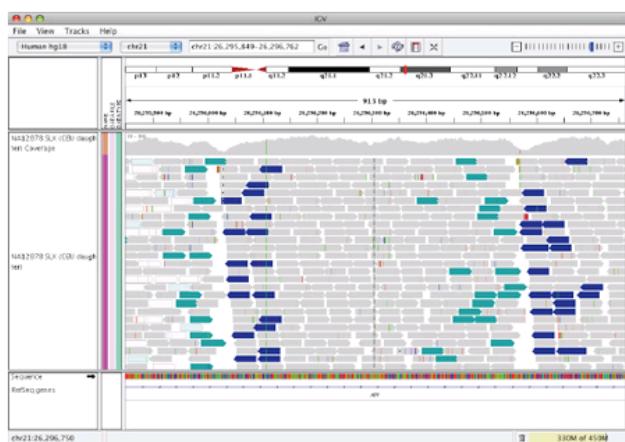
An inversion is a large section of DNA that is reversed in the subject genome compared to the reference genome.



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:

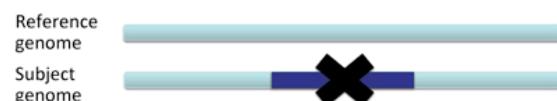
- deletions
- insertions
- inter-chromosomal rearrangements

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:

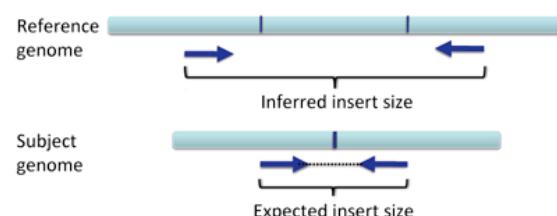
- Red arrow for an insert that is larger than expected
- Blue arrow for an insert that is smaller than expected
- Chromosome color bar for paired end reads that are coded by the chromosome on which their mates can be found

Deletions

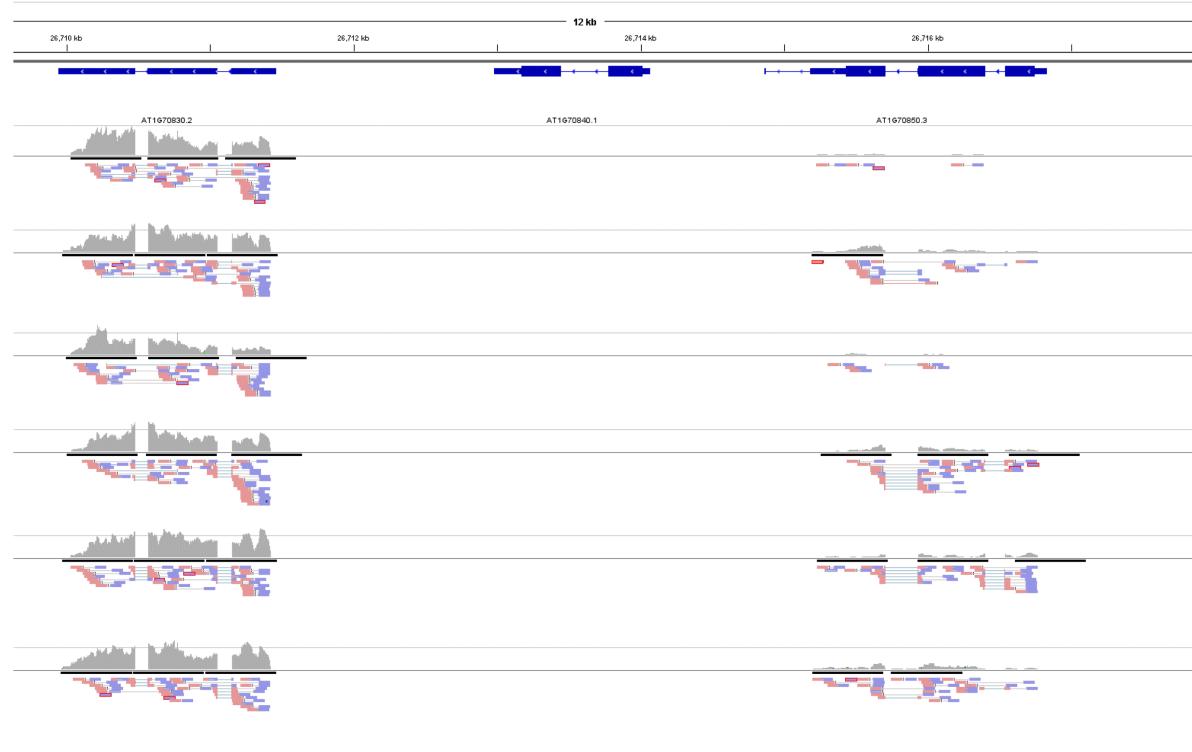
A deletion is a large section of DNA that is absent in the subject genome compared to the reference genome.



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.

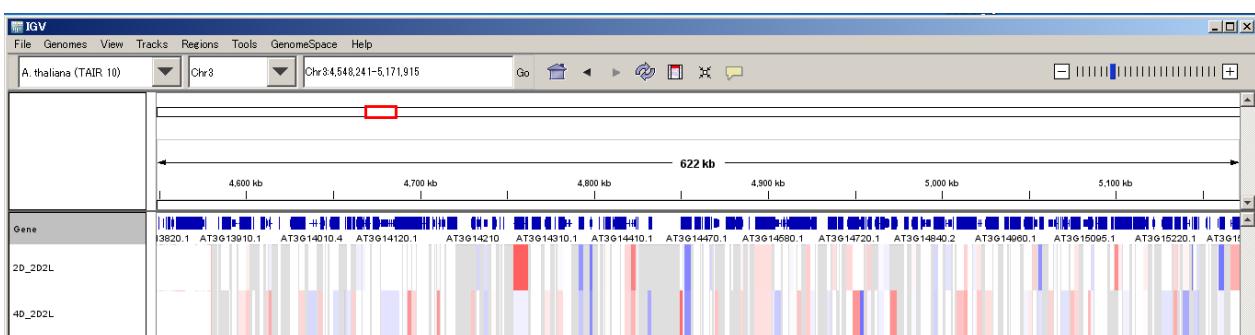


RNA-Seqのデータ表示させる



GCTファイルでgene ローラスの発現情報を図示

#	#	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



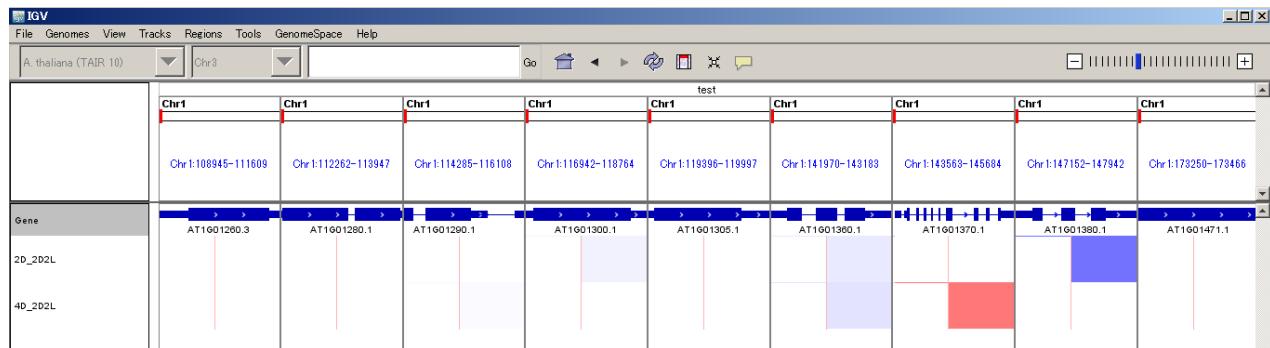
Gene listを定義して
サンプルごと
条件ごと
の発現・発現変動を
カラーマップできる

The Gene Lists functionalities in IGV allow you to view lists of genes or loci side-by-side irrespective of their genomic location.

Loading/Defining Gene Lists

To load or define a new gene/locus list, select Regions >Gene Lists....

This opens a window for selecting an existing list or creating a new list.



IGV実習

Home > Downloads

Downloads

Did you know that there is also an IGV web application that runs only in a web browser, does not use Java, and requires no downloads? See <https://igv.org/app>. Click on the Help link in the app for more information about using IGV-Web.

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IGV and igvtools to run on the command line (all platforms)

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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

Other IGV Versions

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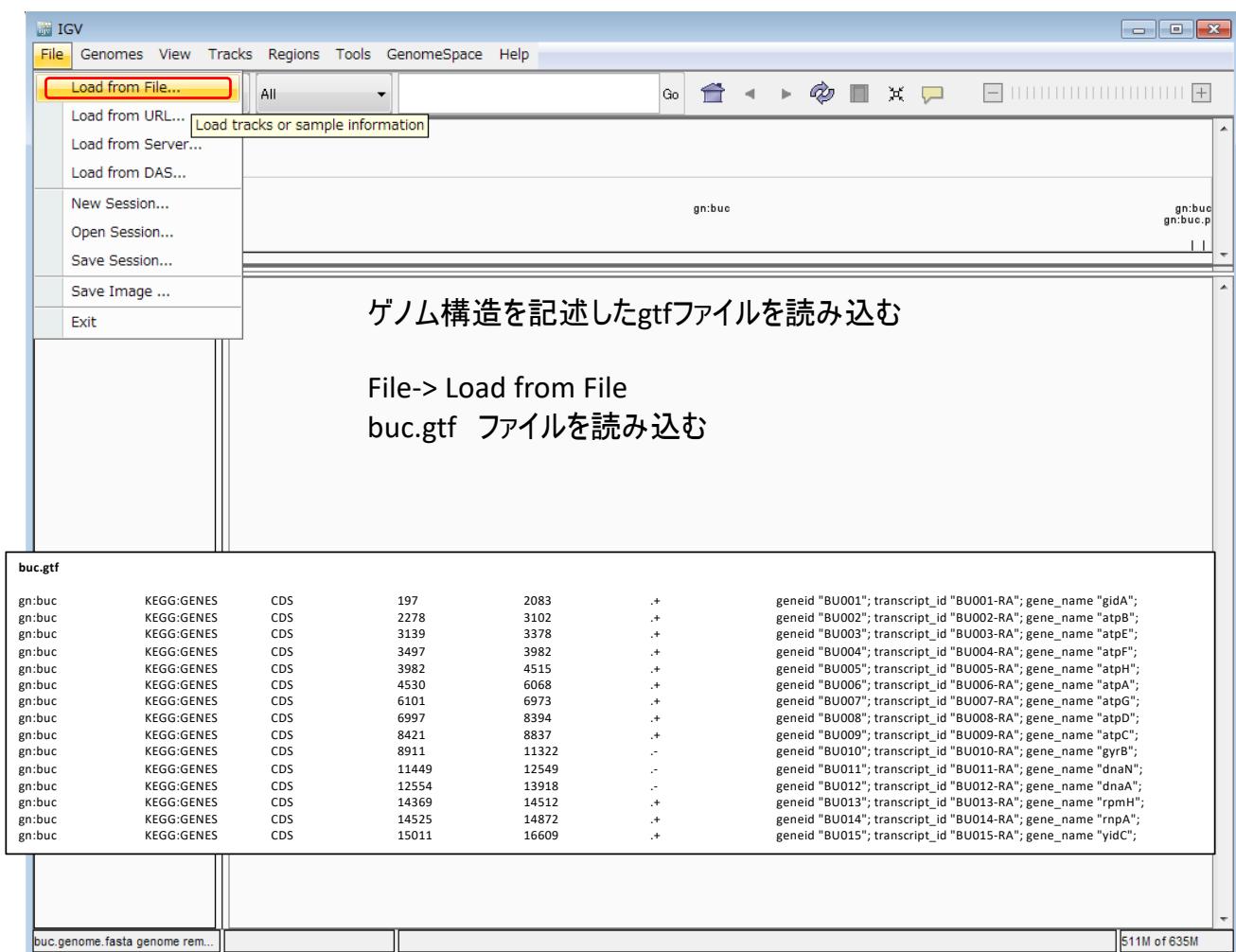
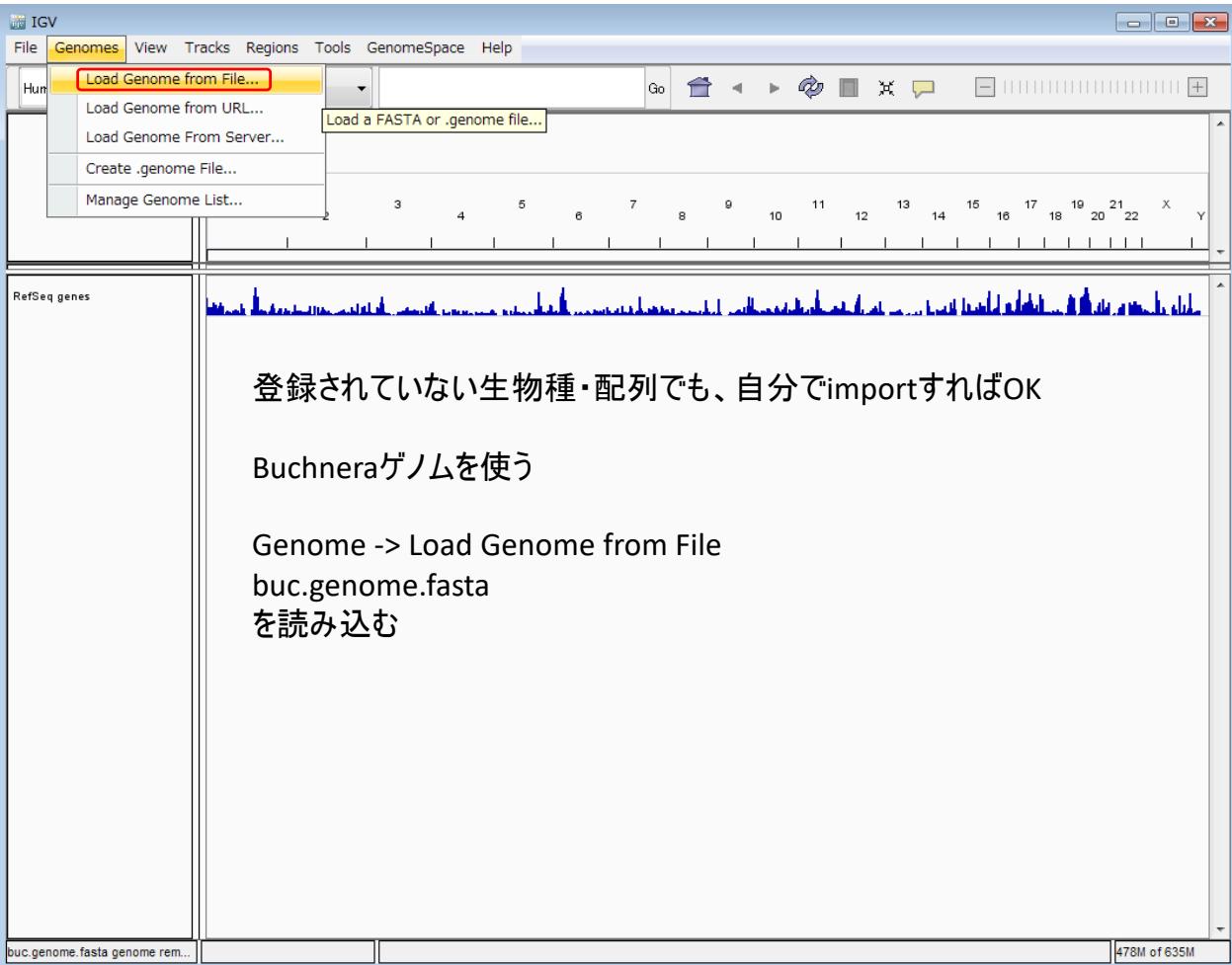
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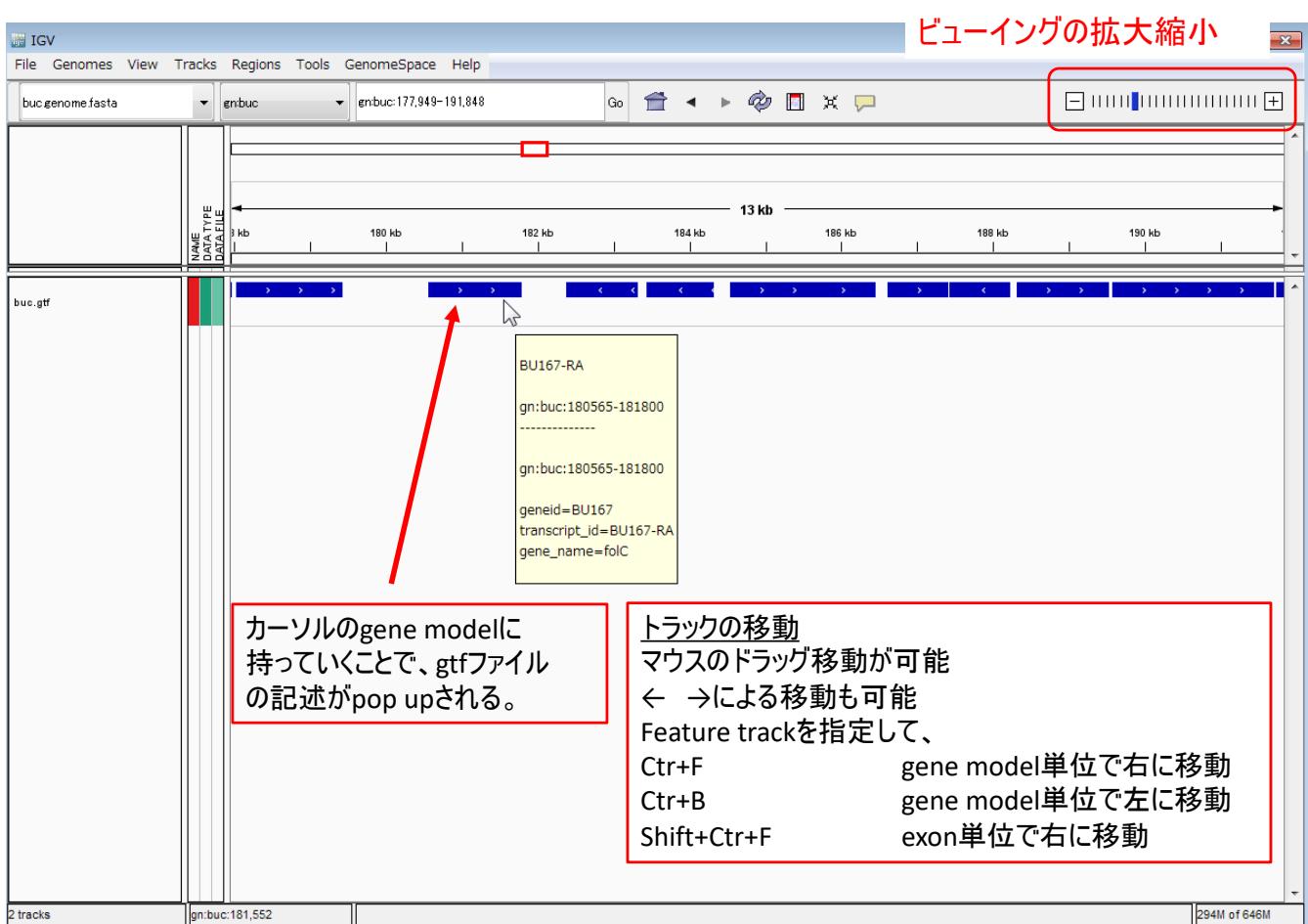
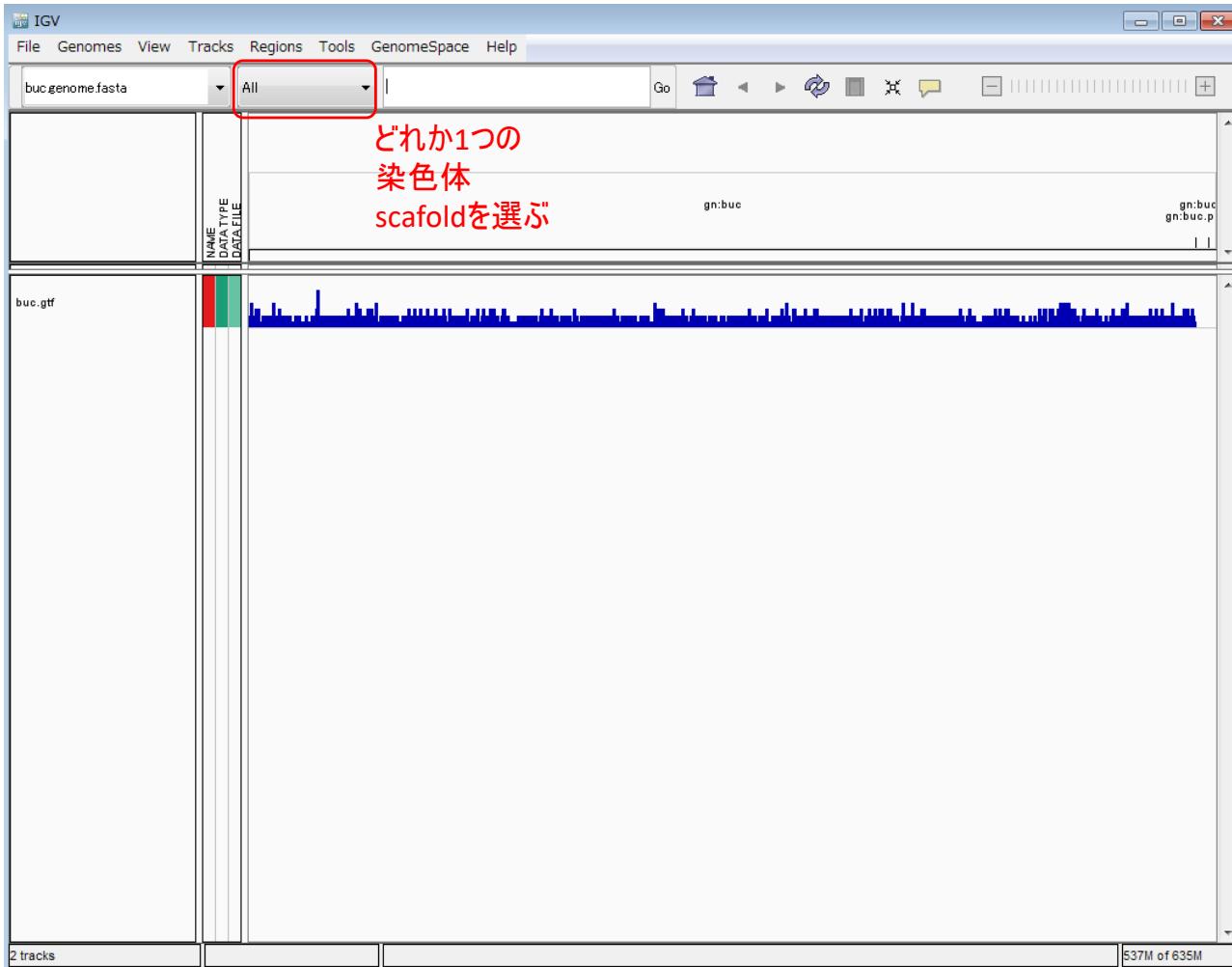
Source Code

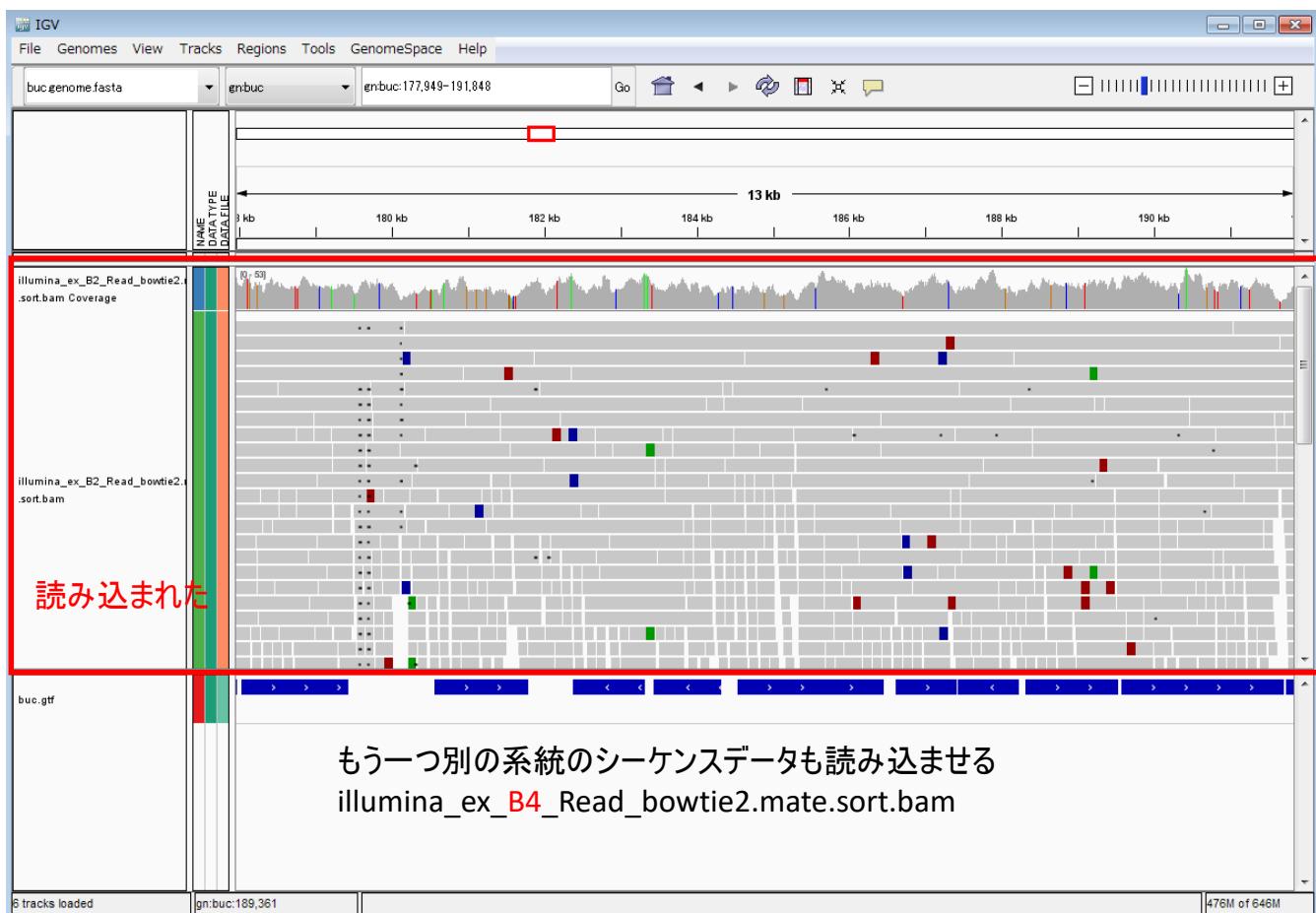
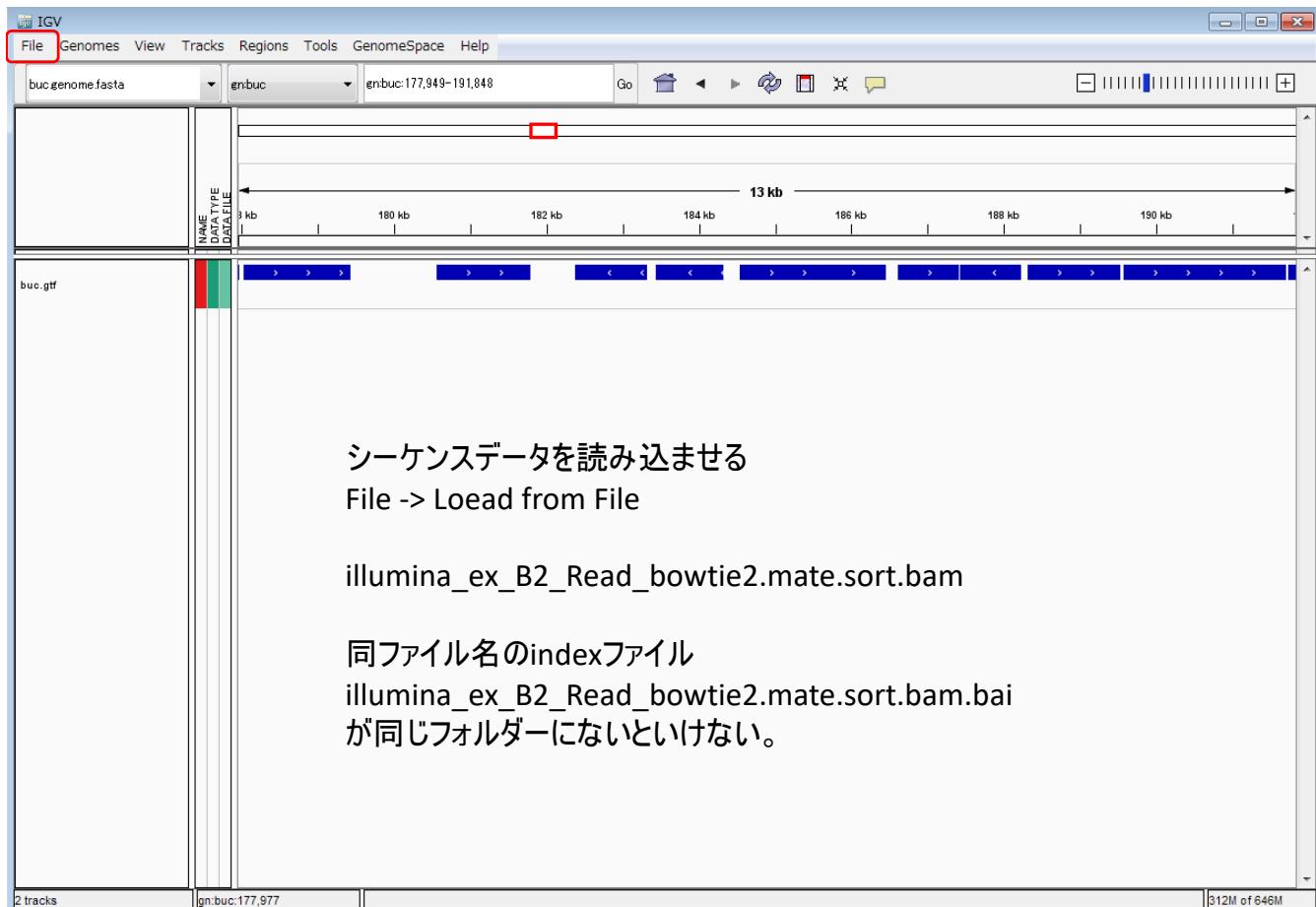
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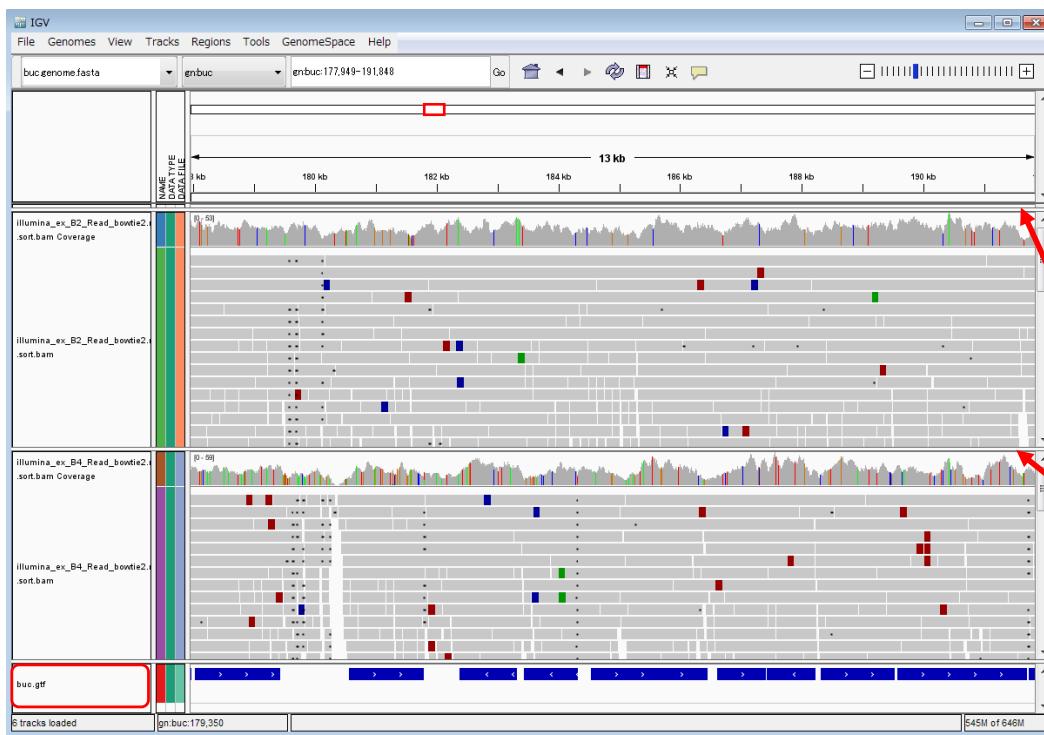
License

Permission to use this work is granted under the [MIT License](#)

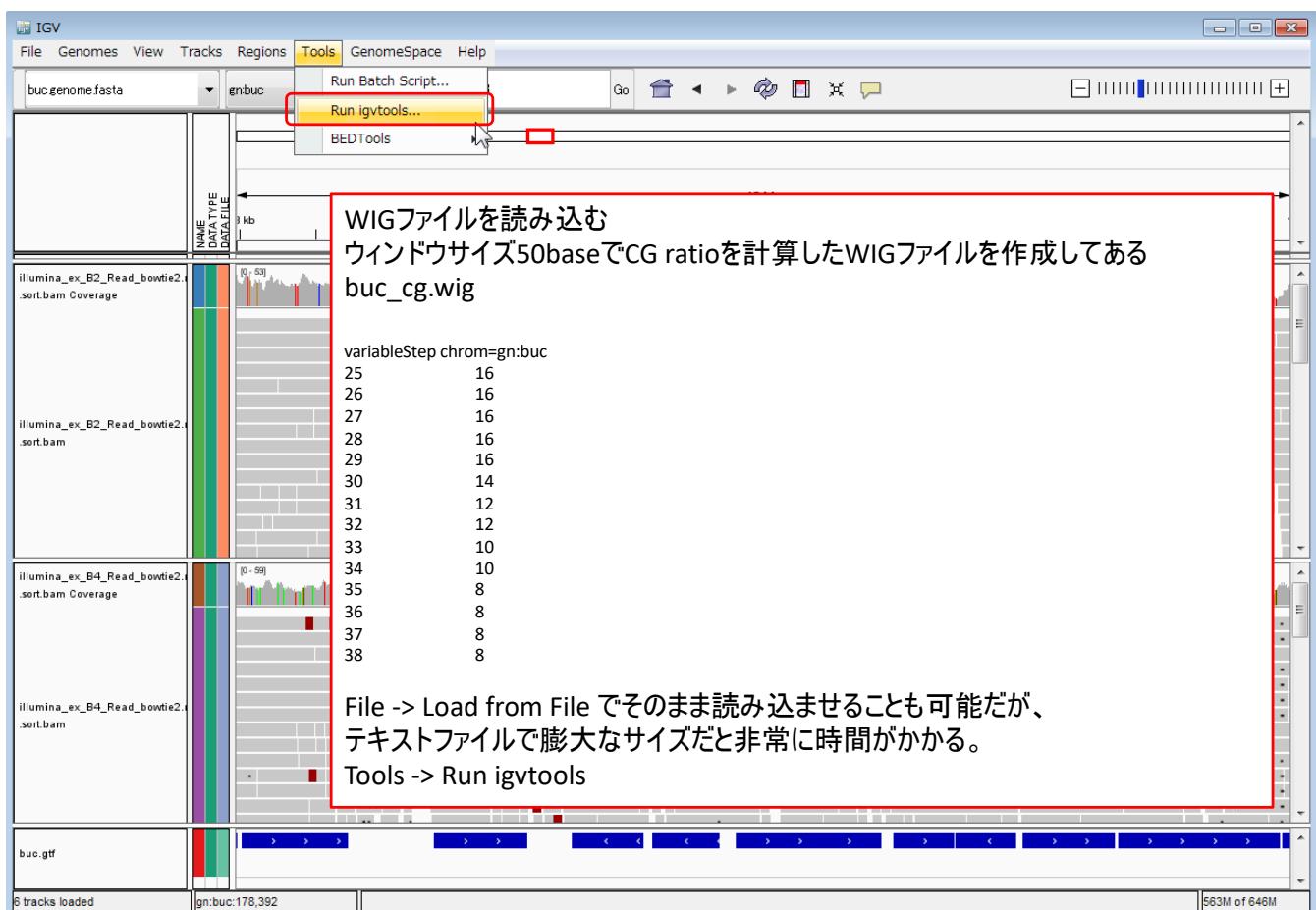


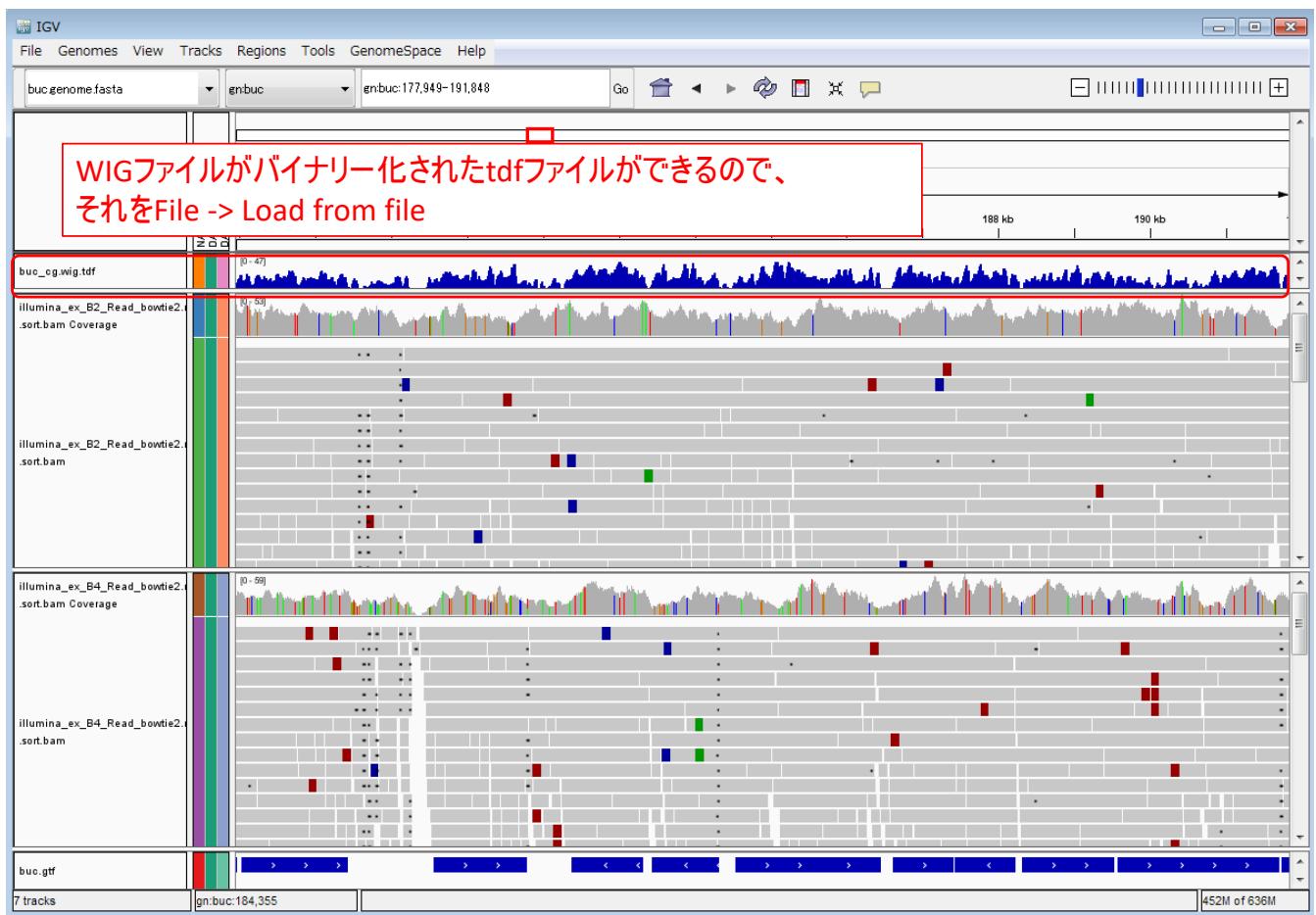
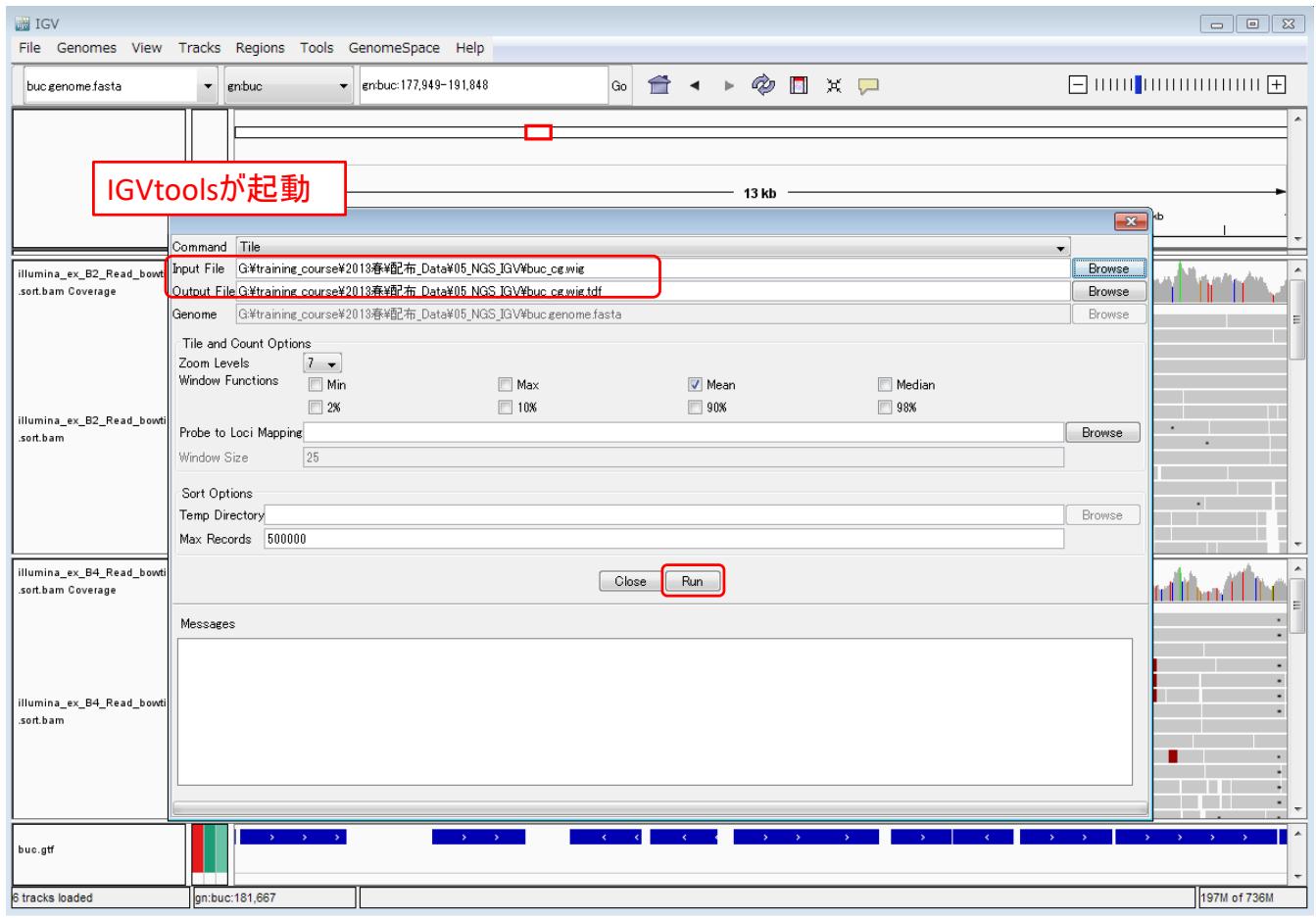


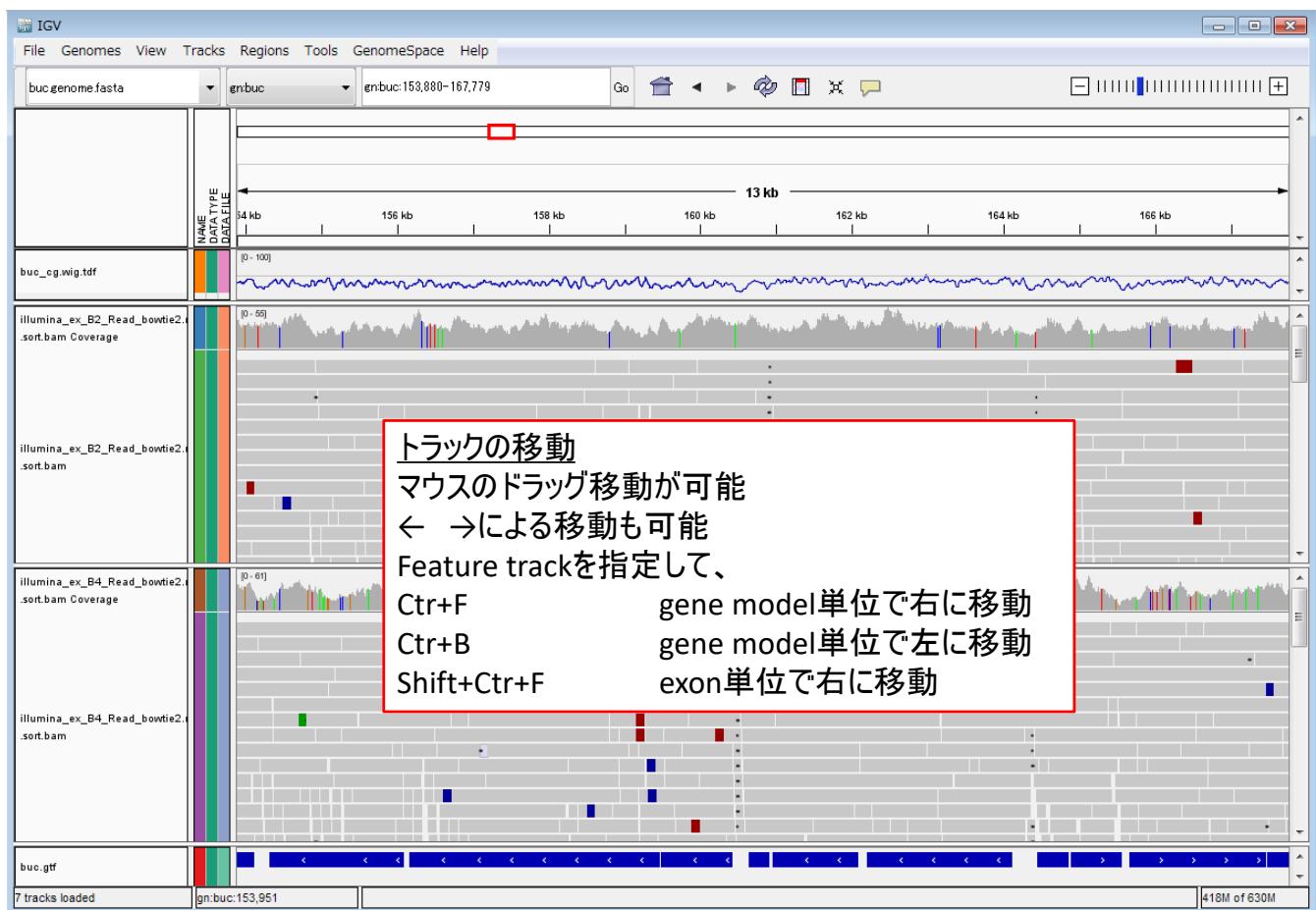
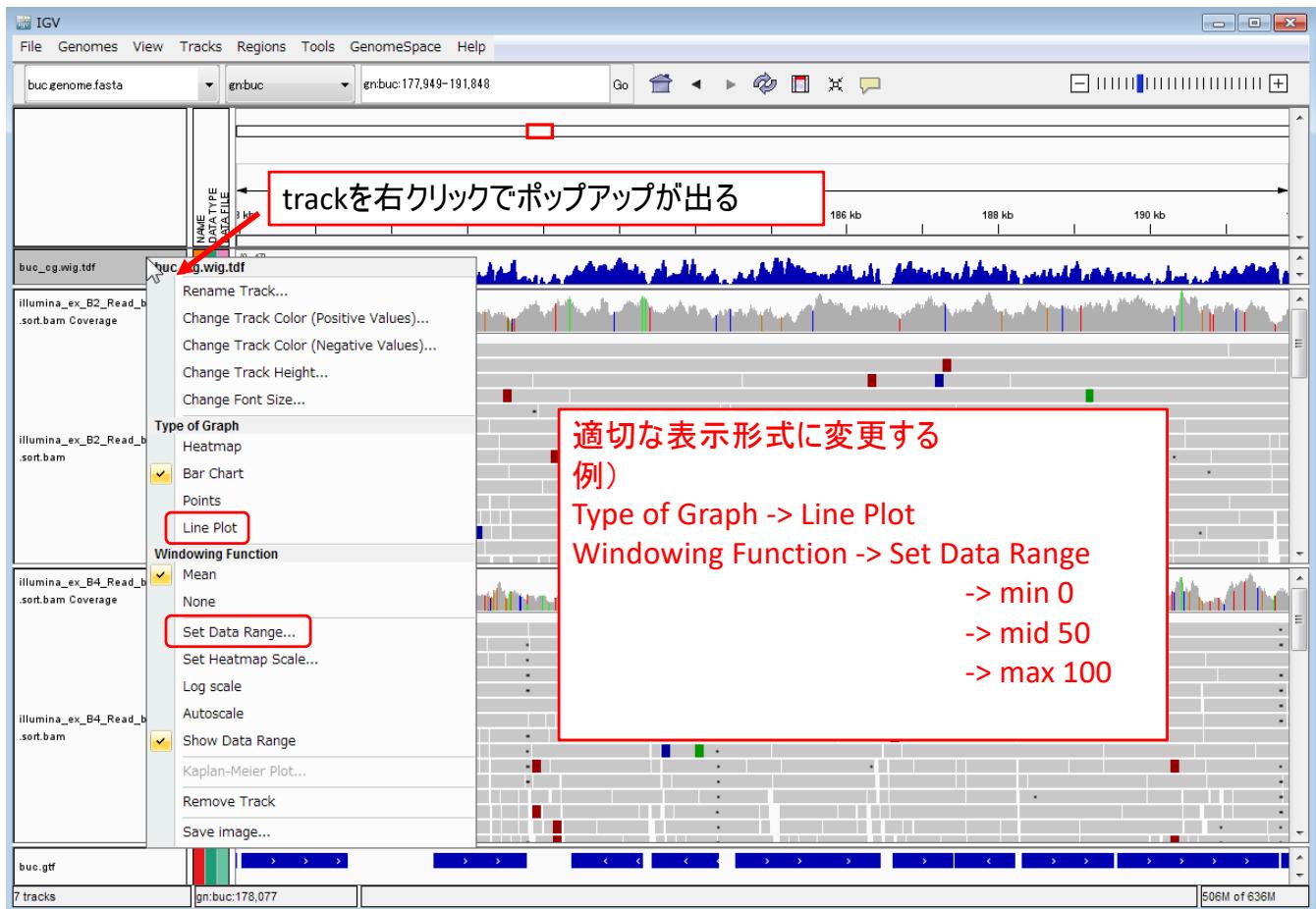


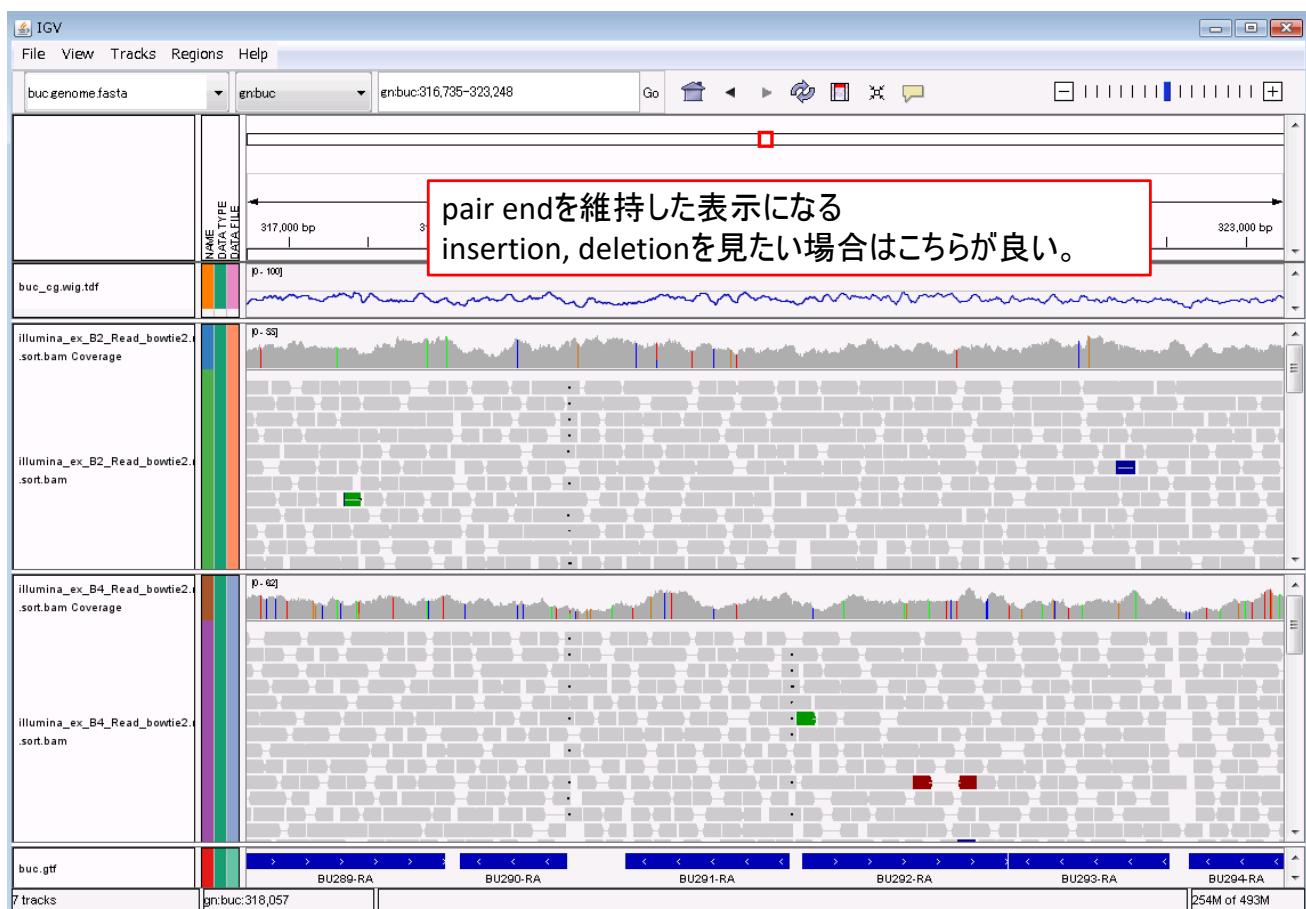
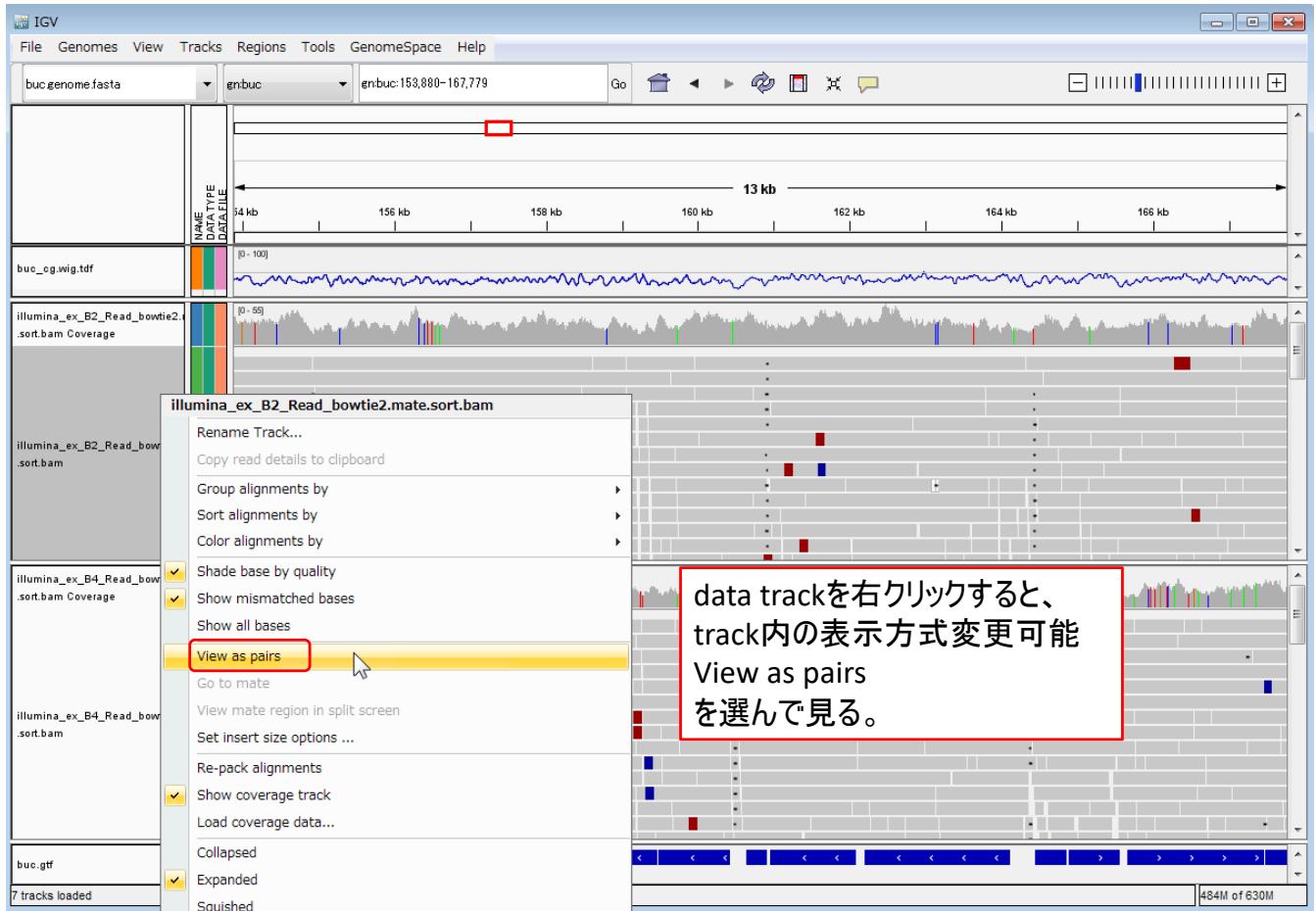


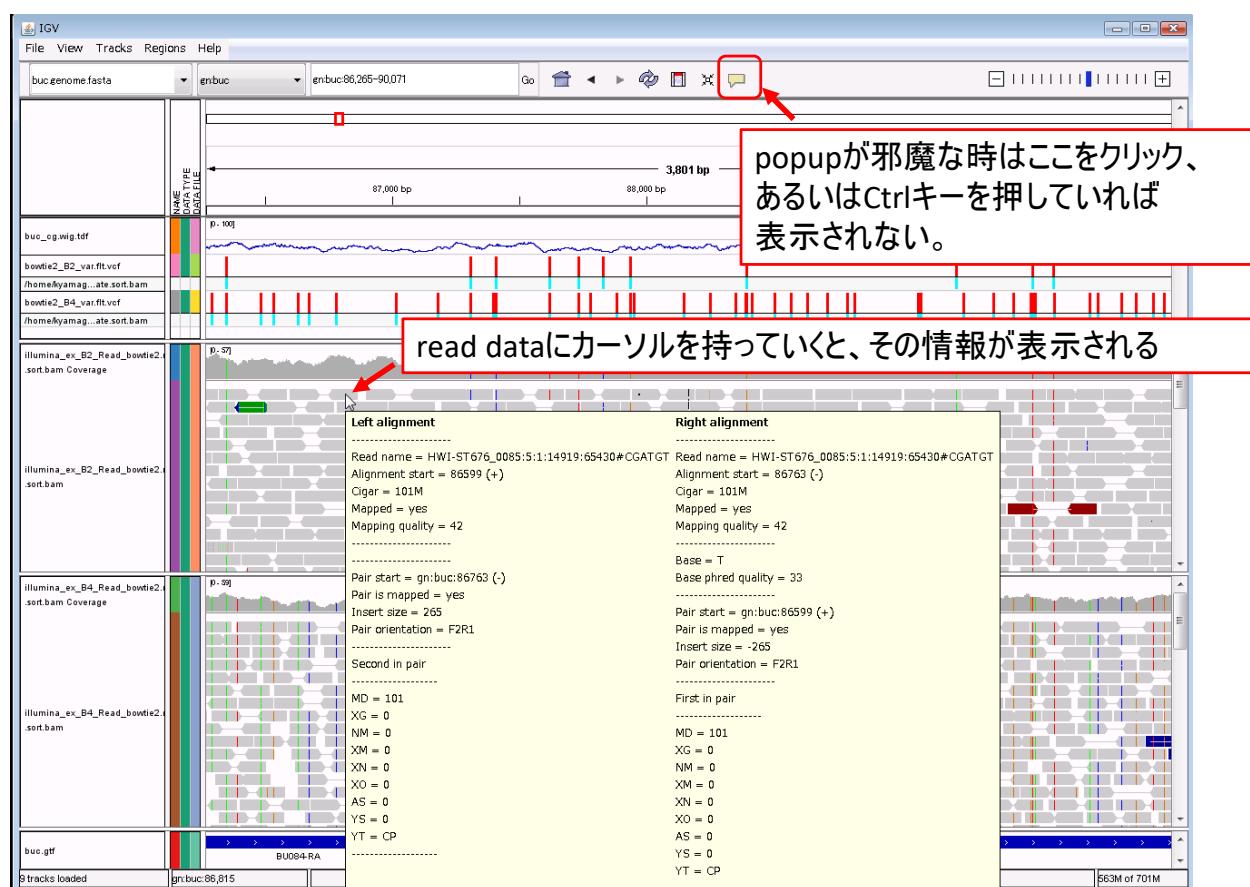
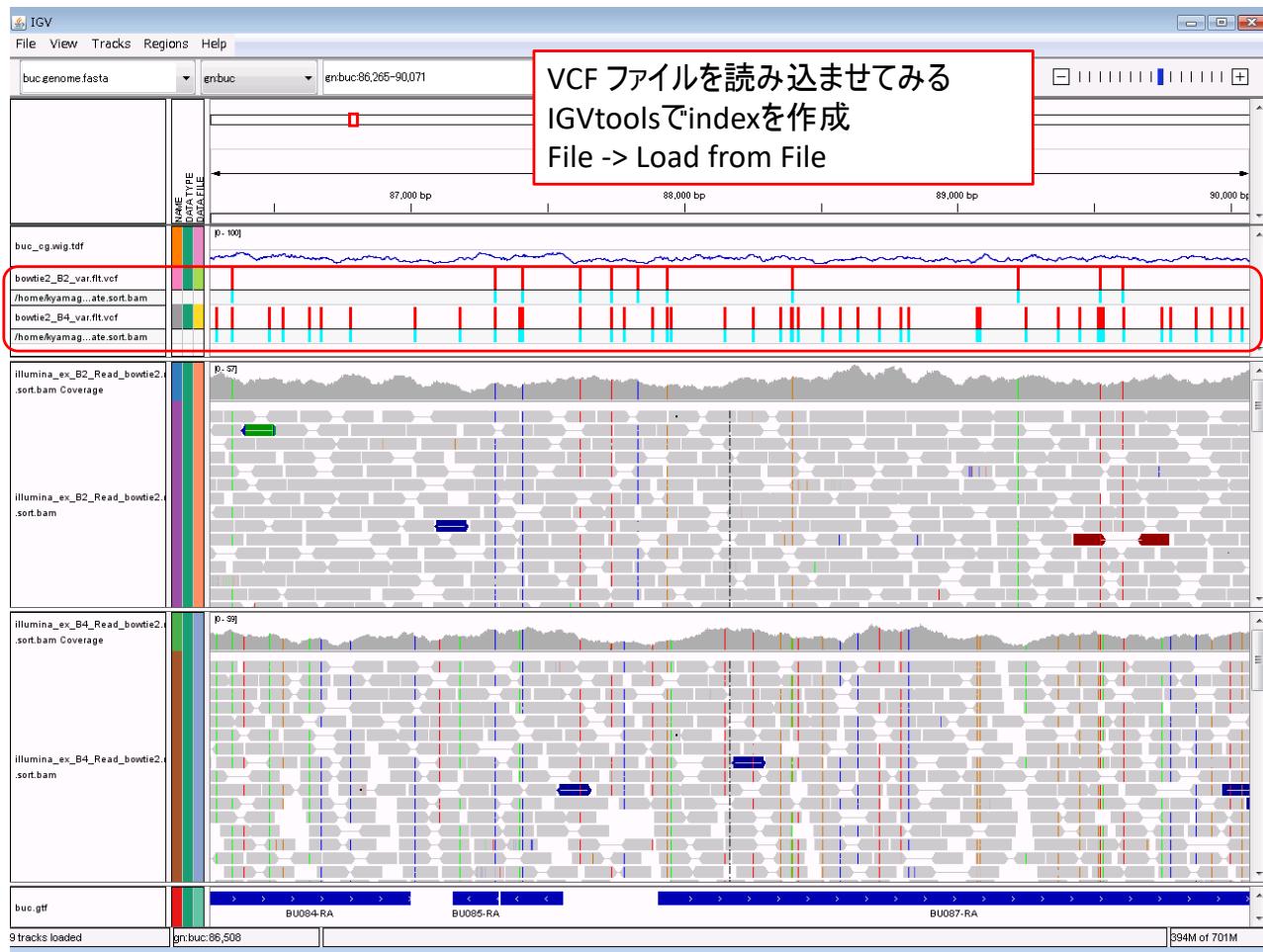
各Trackはドラッグアンドドロップで移動できる











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。