

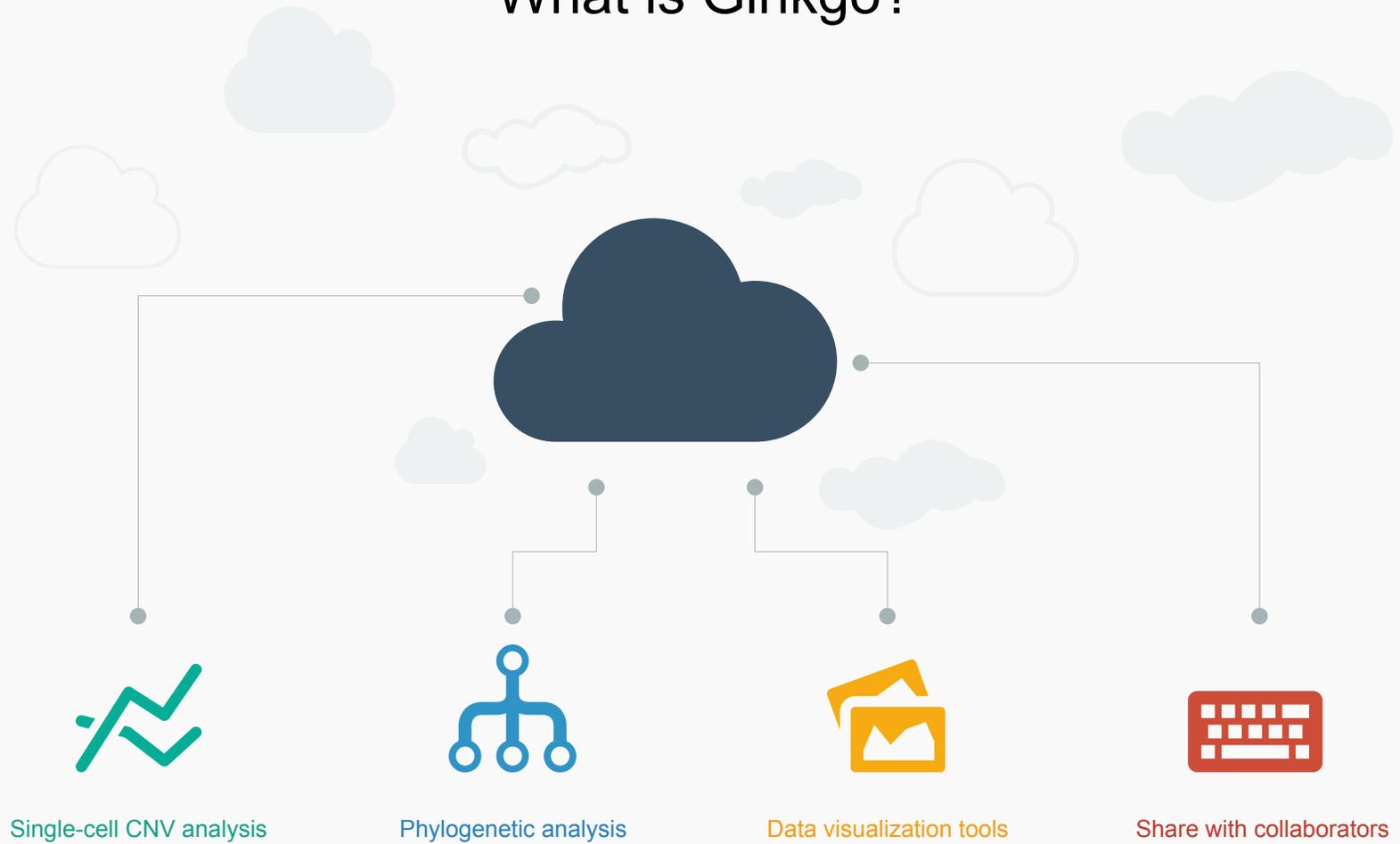


SINGLE-CELL COPY-NUMBER ANALYSIS

## **VIZBI 2015 WORKSHOP**

Robert Aboukhalil  
raboukha@cshl.edu

# What is Ginkgo?



# Outline

Experimental  
approaches

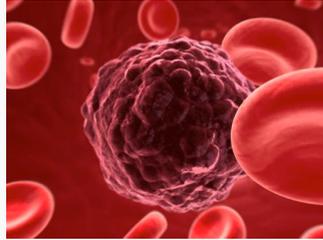
Computational  
approaches for  
CNV analysis

Demo

Hands-on



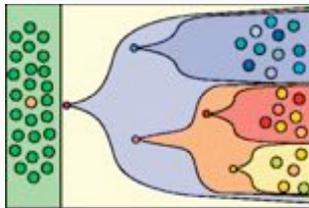
# Single-cell sequencing



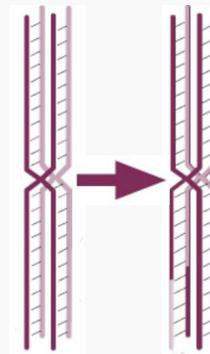
Circulating tumor cells



Neuronal mosaicism



Clonal evolution in tumors



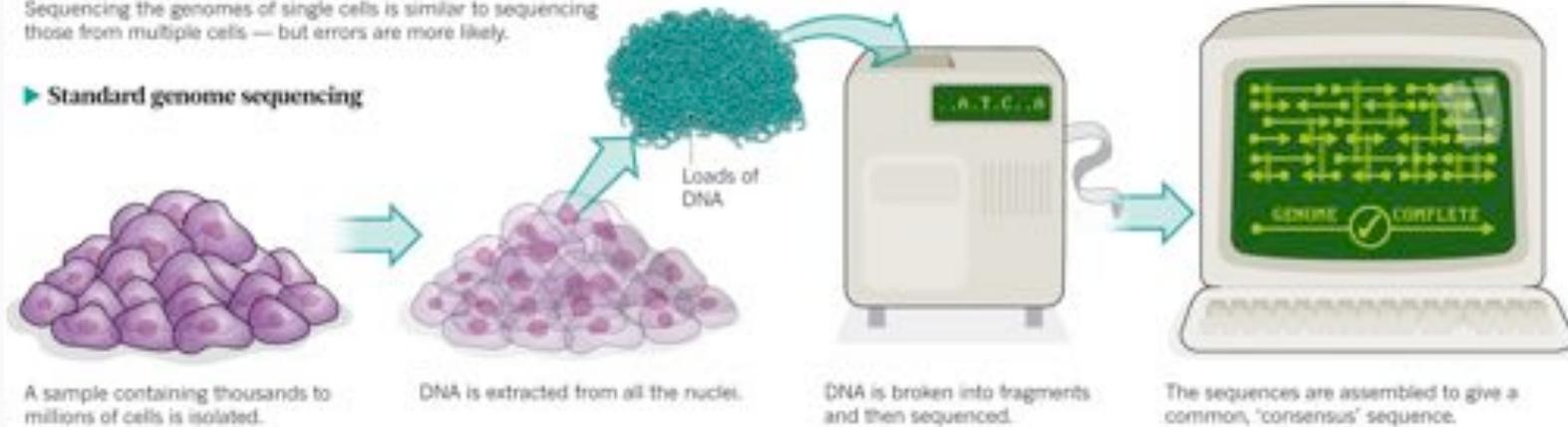
Recombination/  
crossover in germ cells

# Single-cell vs. bulk sequencing

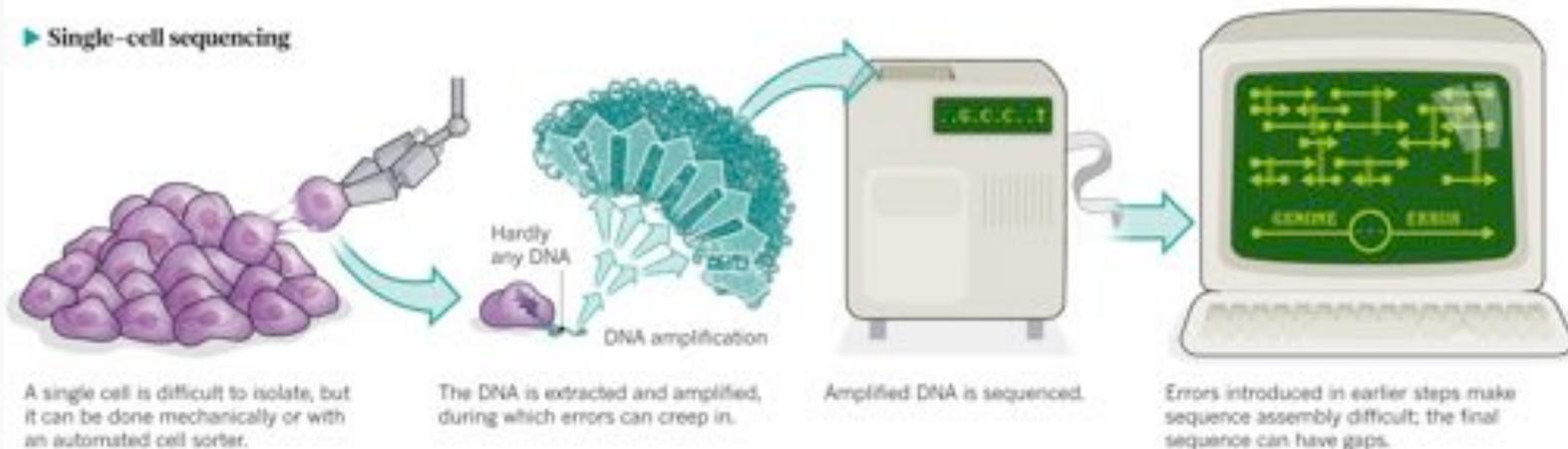
## ONE GENOME FROM MANY

Sequencing the genomes of single cells is similar to sequencing those from multiple cells — but errors are more likely.

### ► Standard genome sequencing



### ► Single-cell sequencing

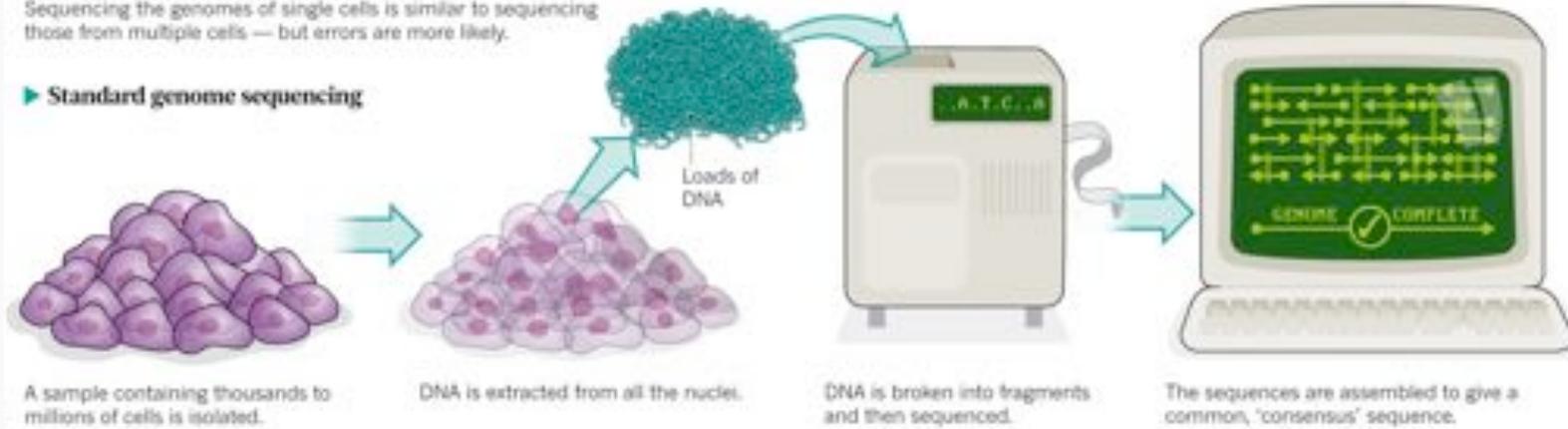


# Single-cell vs. bulk sequencing

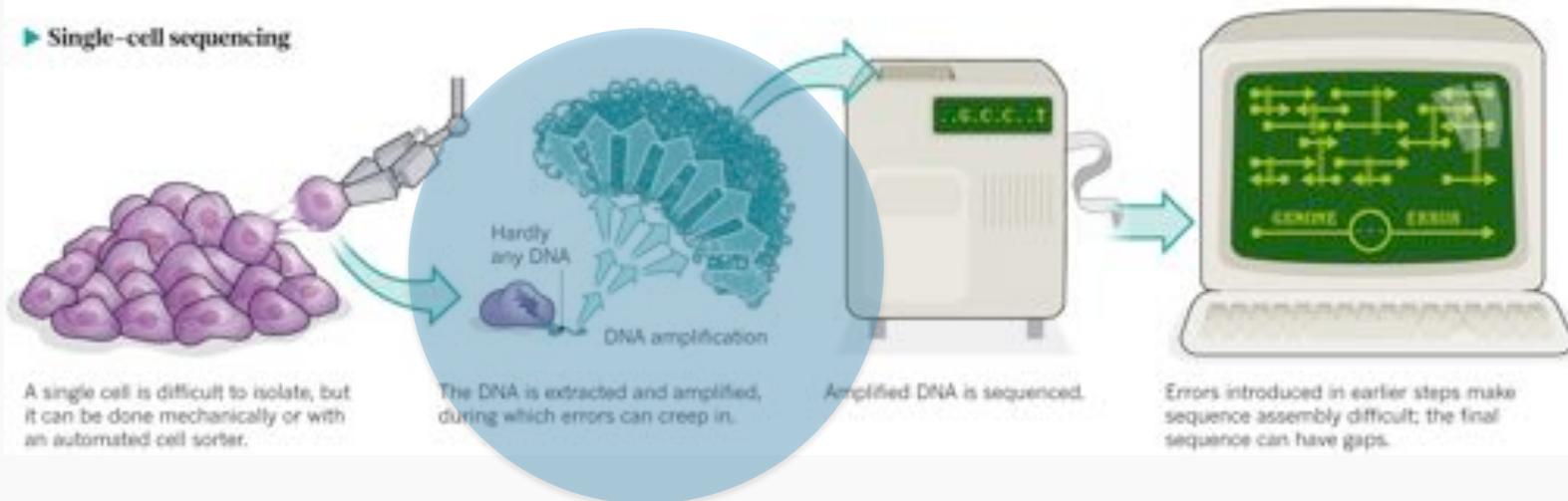
## ONE GENOME FROM MANY

Sequencing the genomes of single cells is similar to sequencing those from multiple cells — but errors are more likely.

### ► Standard genome sequencing



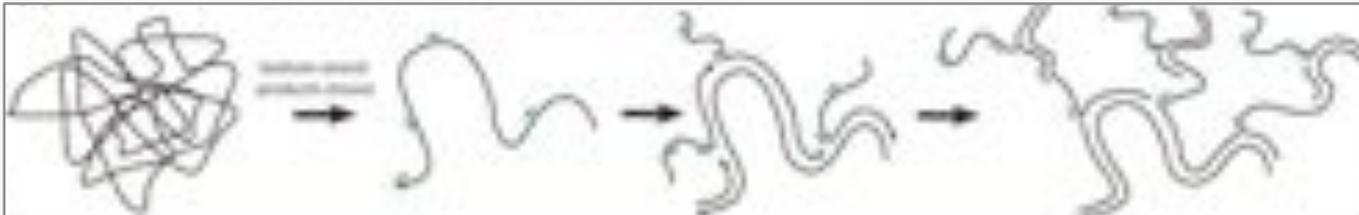
### ► Single-cell sequencing



# Whole Genome Amplification (WGA) methods



DOP-PCR (Degenerate Oligonucleotide Primed PCR)



MDA (Multiple Displacement Amplification)



MALBAC (Multiple Annealing and Looping Based Amplification Cycles)

# Whole Genome Amplification (WGA) methods

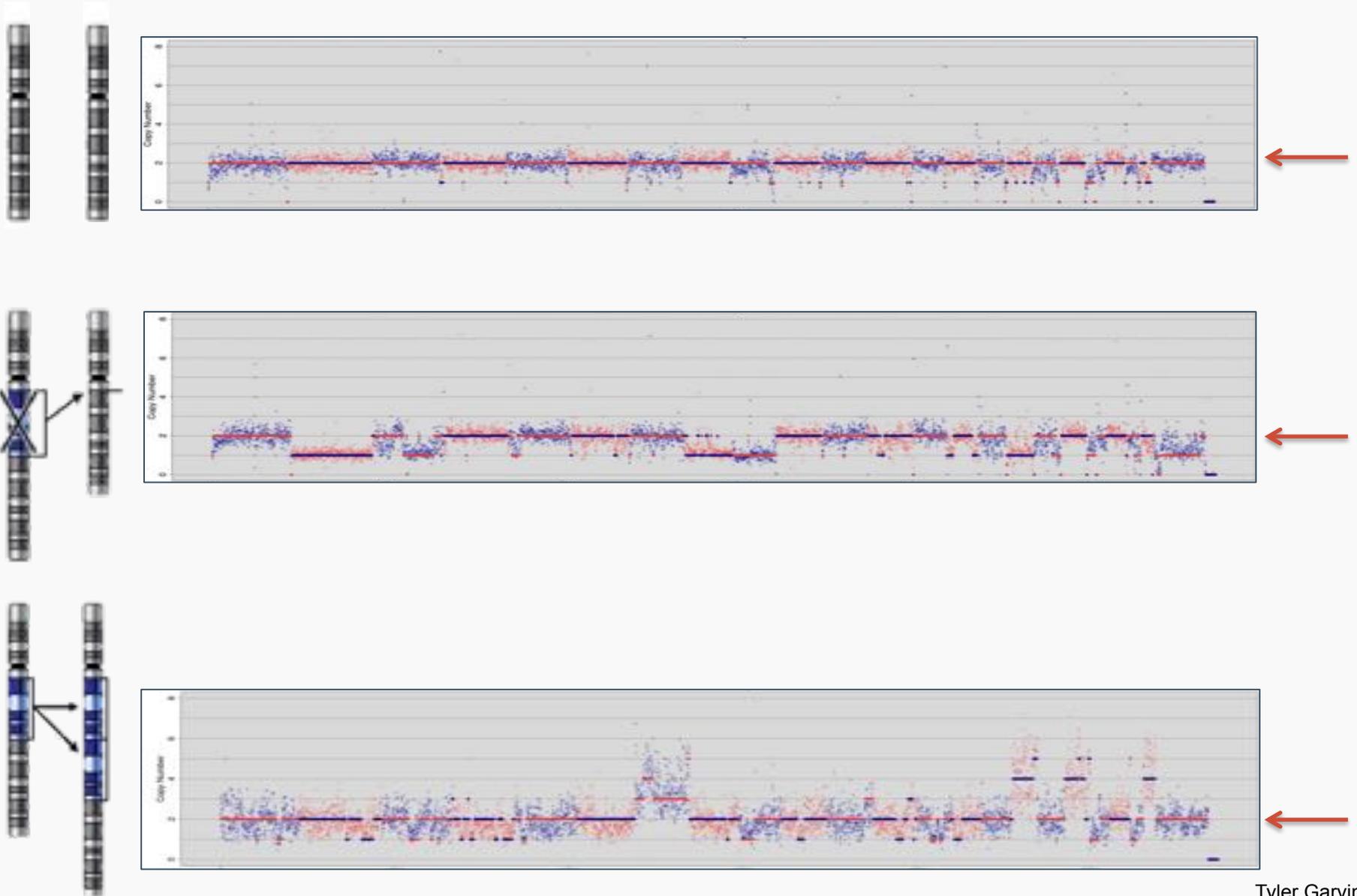
The image displays three Whole Genome Amplification (WGA) methods, each represented by a schematic diagram and a label:

- DOP-PCR (Degenerate Oligonucleotide-primed PCR)**: Shown as a tangled network of lines.
- MDA (Multiple Displacement Amplification)**: Shown as a tangled network of lines.
- MALBAC (Multiple Annealing and Looping Based Amplification Cycles)**: Shown as a sequence of steps from a tangled network to a single strand.

The central bioRxiv preprint card contains the following information:

- Logos**: CSH Cold Spring Harbor Laboratory and bioRxiv beta THE PREPRINT SERVER FOR BIOLOGY.
- Text**: New Results [bit.ly/1EkCCOO](https://bit.ly/1EkCCOO)
- Title**: **Interactive analysis and quality assessment of single-cell copy-number variations**
- Authors**: Tyler Garvin, Robert Aboukhalil, Jude Kendall, Timour Baslan, Gurinder S Atwal, James Hicks, Michael Wigler, Michael Schatz
- DOI**: <http://dx.doi.org/10.1101/011346>

# Copy-number variant analysis



# Copy-number variant analysis



---

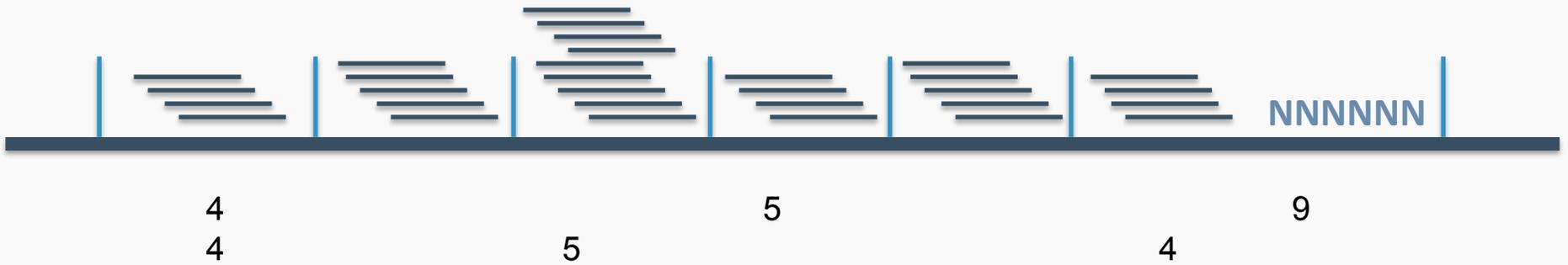
Low coverage allows us to study copy-number variants

# Copy-number variant analysis

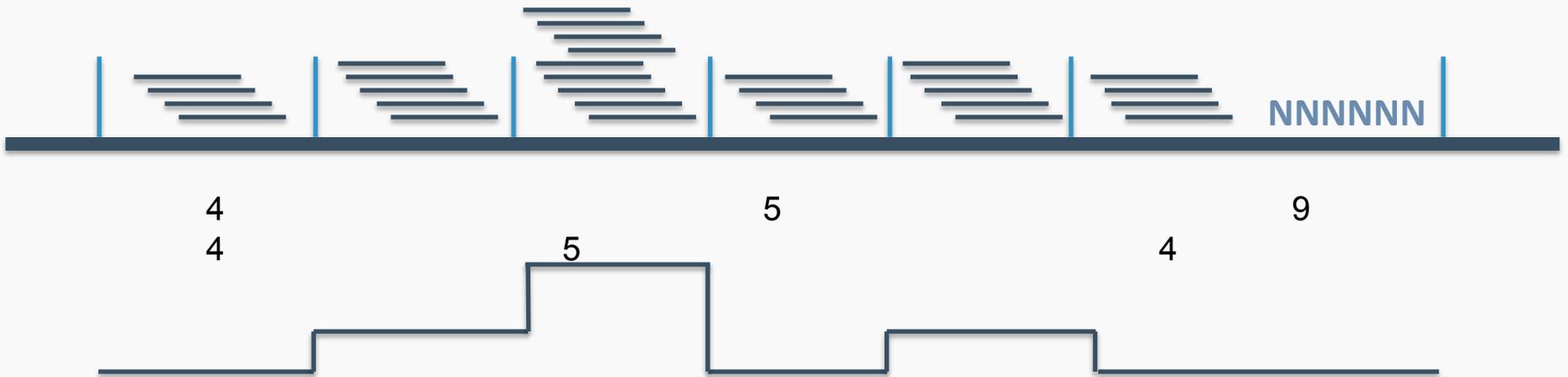


Divide genome into “bins” with ~50 – 100 reads / bin

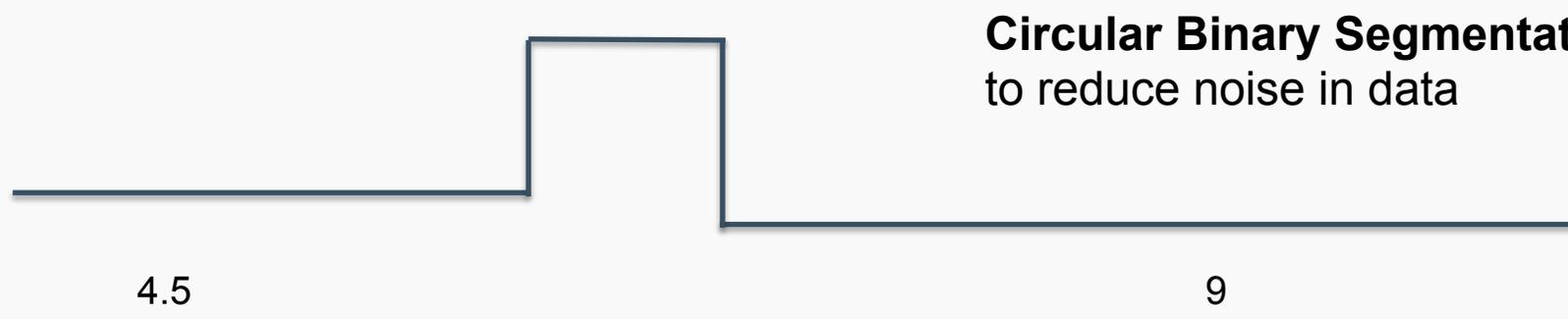
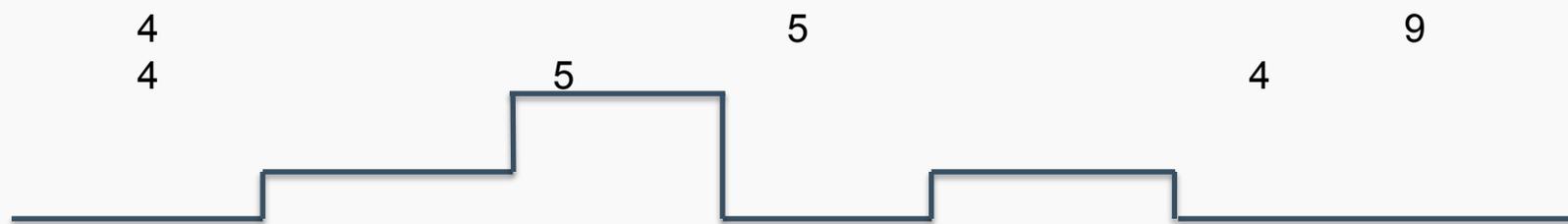
# Copy-number variant analysis



# Copy-number variant analysis



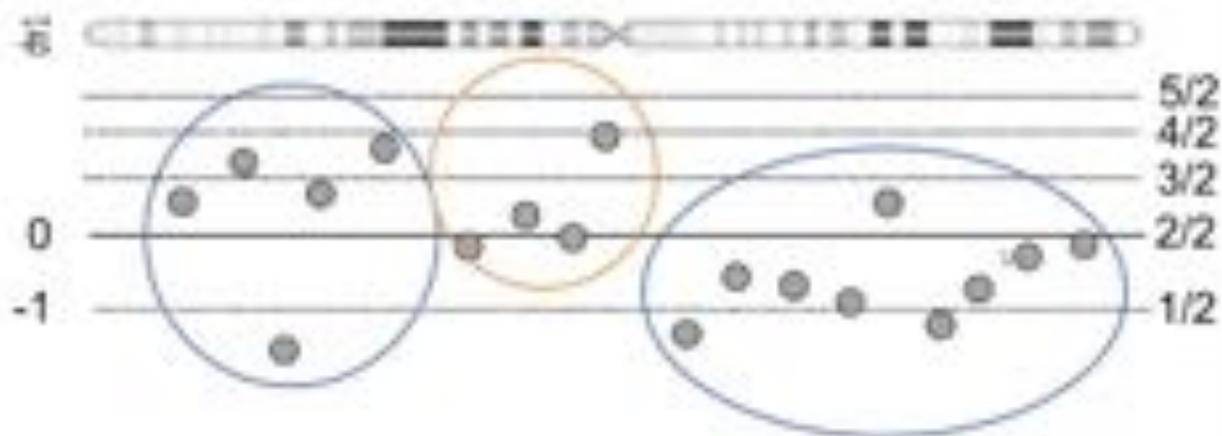
# Copy-number variant analysis



**Circular Binary Segmentation (CBS)**  
to reduce noise in data

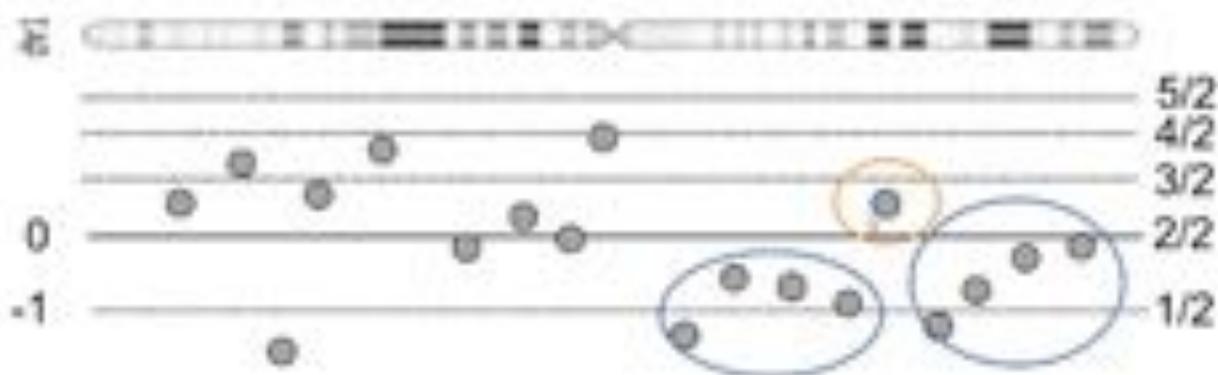
## Circular Binary Segmentation

- Recursively divide up the genome until identify segments that have probe distribution different than neighbors



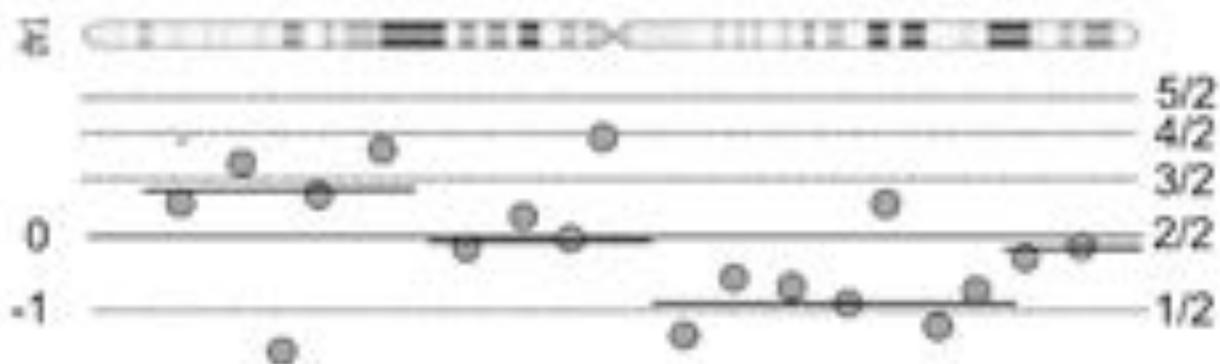
## Circular Binary Segmentation

- Recursively divide up the genome until identify segments that have probe distribution different than neighbors

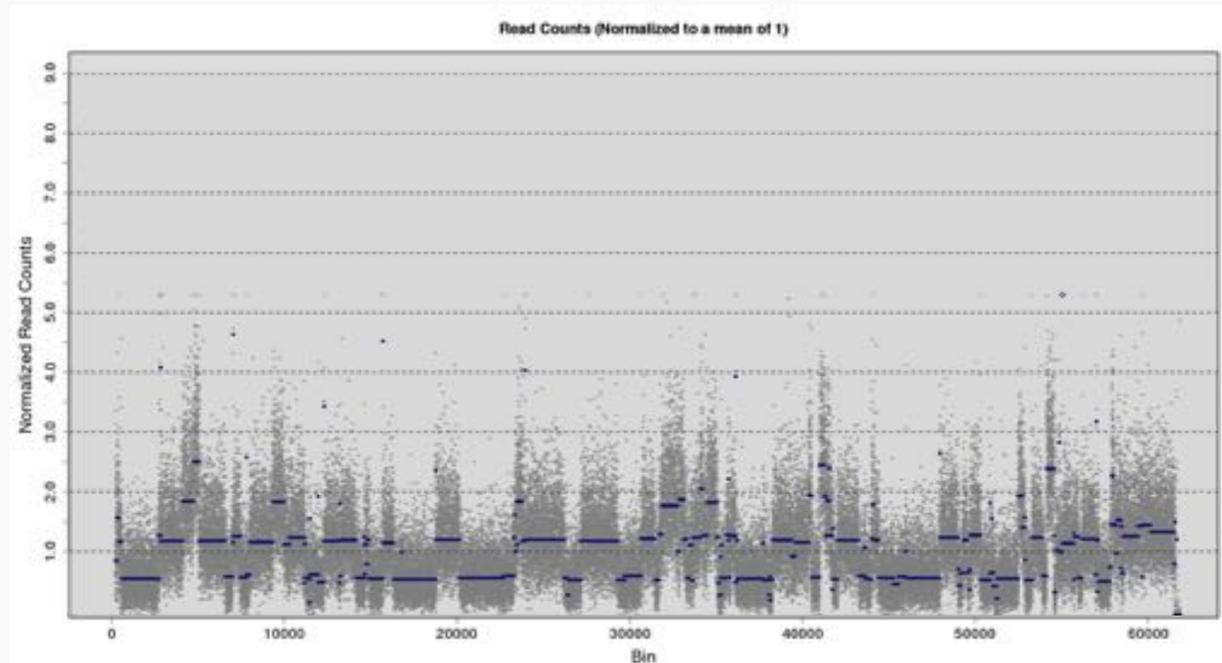


## Circular Binary Segmentation

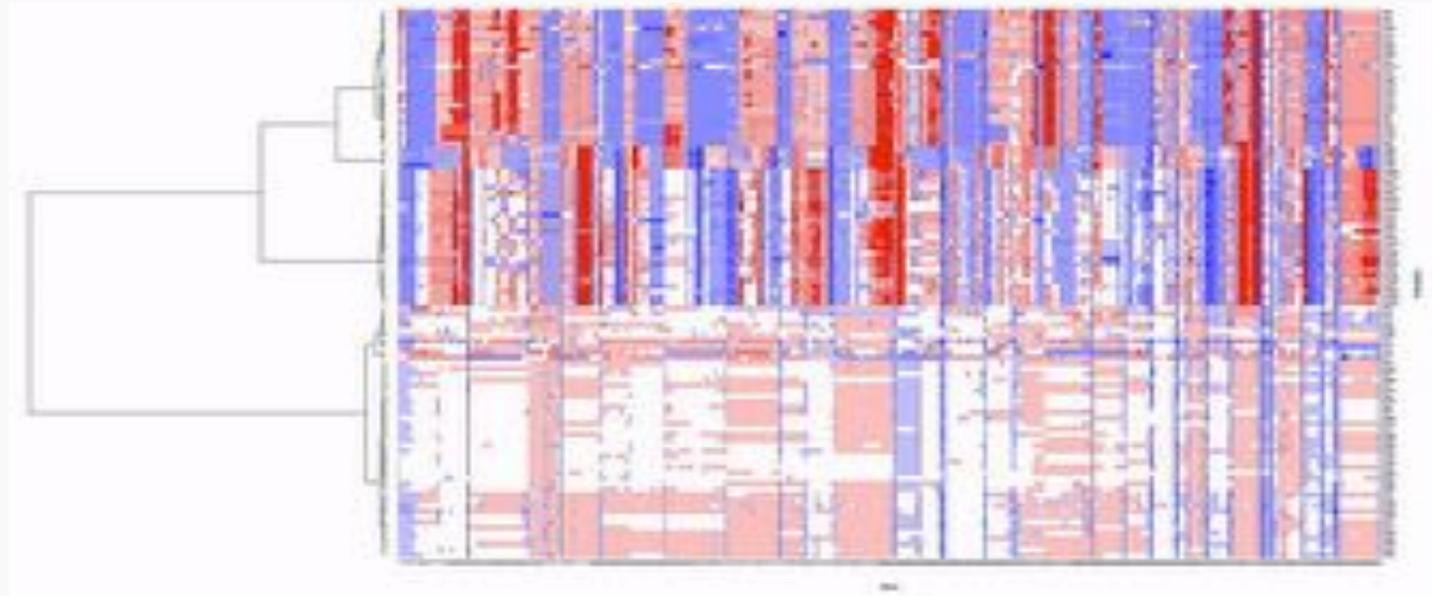
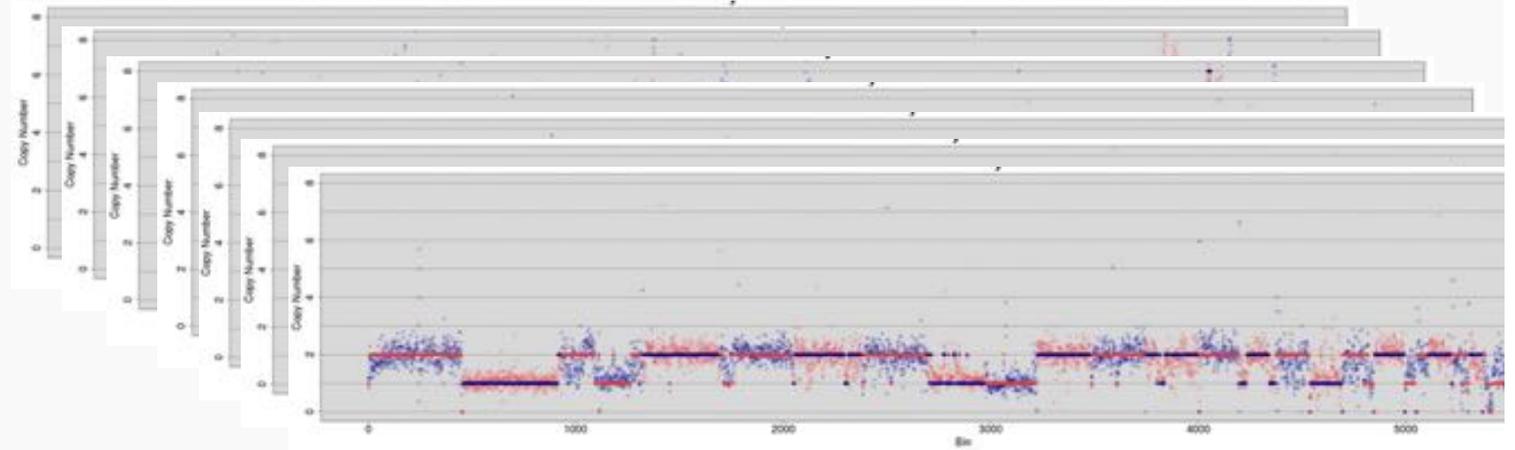
- Recursively divide up the genome until identify segments that have probe distribution different than neighbors



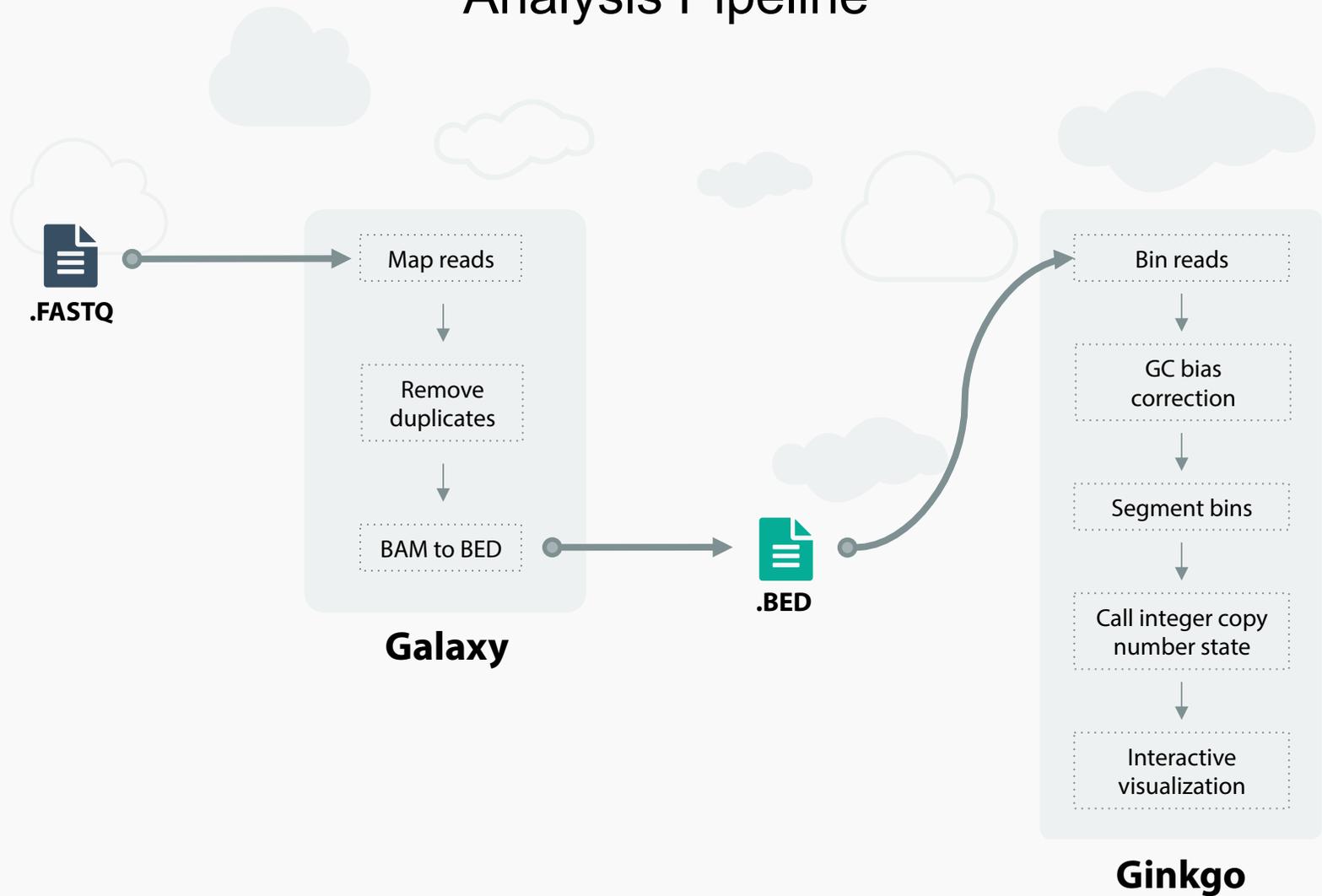
# Copy-number variant analysis



# Copy-number variant analysis



# Analysis Pipeline



# Galaxy intro



# Demo – Galaxy Pipeline

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Using 0 tools

Tools

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- Join, Subtract and Cross
- NCA, QC and manipulation
- NCA Missing
- NCA, BAM Tools
- NCA Picard
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- BIGTools
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- Motif Tools
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- NCA SAM Tools

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The very first tutorial you need

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Confirm password:

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- sniffIT
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- Graph/Display Data
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Get Data

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NGS Mapping

LRGE Map short reads against reference sequence

Workflows

• All workflows

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SRR054622

The image shows a screenshot of a web browser displaying the Galaxy interface for the European Nucleotide Archive (ENA). The browser's address bar shows the URL <https://usegalaxy.org>. The main content area features the ENA logo and a search bar containing the accession number **SRR054622**. Below the search bar, there are navigation tabs: Home, Search & Browse, Submit & Update, About ENA, and Support. A message below the tabs reads: "Please subscribe to our announce mailing list from [enahelp@ebi.ac.uk](mailto:enahelp@ebi.ac.uk) to receive alerts about ENA services." The footer contains a grid of menu items for Services, Research, Training, Industry, and About us, along with contact information for EMBL-EBI.

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NCBI Mapping

Look up short reads against reference sequence

Workflows

- All workflows



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**Read: SRR054622**

Illumina Genome Analyzer sequencing: Single Cells from Breast Tumor T10

View: [JSON](#) Send feedback to [Download: \[JSON\]\(#\)](#)

Submitting Centre	Run Date	Platform	Model	Read Count	Base Count
Cold Spring Harbor Laboratory		ILLUMINA	Illumina Genome Analyzer	2,799,858	131,913,184

Library Layout	Library Strategy	Library Source	Library Selection	Library Name
SCOLE	WGS	GENOMIC	RANDOM	SC.WGS.LIB

Broker Name  
NCBI

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This table contains the files for run SRR054622

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Get Data  
BLAST DNA SMA

NGS Mapping  
LRR2 Map short reads against reference sequence

Workflows  
• All workflows

### Read: SRR054622

Human Genome Analyzer sequencing; Single Cells from Breast Tumor T10

View: XML Send Feedback   
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Submitting Centre	Run Date	Platform	Model	Read Count	Base Count
Cold Spring Harbor Laboratory		ILLUMINA	Human Genome Analyzer	2,789,858	133,913,184

Library Layout	Library Strategy	Library Source	Library Selection	Library Name
SINGLE	WGS	GENOMIC	RANDOM	SC-WGA-LIB

Broker Name  
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Showing results 1 - 1 of 1 results

Study accession	Secondary study accession	Sample accession	Secondary sample accession	Experiment accession	Run accession	Tax ID	Scientific name	Instrument model	Library layout	Fastq files (R1)	Fastq files (R2)
SRR002535	SRR002535	SAMN0004736	SRR074087	SRR021400	SRR054622	9606	Homo sapiens	Human Genome Analyzer	SINGLE	File 1	File 1

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- 1. SRA DNA SRA

NCBI Mapping

1. SRA Map short reads against reference sequence

Workflows

- All workflows

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1 shown

0 bytes

Annotation

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1. 1. SRA, SRR054622

2. File: ftp://ftp.sra.ehp.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz

A job has been successfully added to the queue – resulting in the following dataset:

- 1. 1. SRA, SRR054622 File: ftp://ftp.sra.ehp.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz

You can check the status of queued jobs and view the resulting data by refreshing the History pane. When the job has been run the status will change from "queued" to "finished" if completed successfully or "error" if problems were encountered.

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https://usegalaxy.org

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- Get Data
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- Edit and Sort
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- NGS BAM Tools
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- Fetch Alignments
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- snEff
- BEITools
- Genome Diversity
- EMBOSS
- Regional Variation
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- Microarray analysis
- Metatools
- NGS TOOLBOX BETA

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Try Galaxy on the Cloud

Now you can host a personal Galaxy within the cloud browser.

Tweets

PyPi Recent Updates @pyup\_updates in planemo 0.6.0: Command-line utilities to assist in building tools for the Galaxy project #Pip-Galaxy - bit.ly/100CkYI 11 Retweeted by Galaxy Project

Galaxy Project @galaxyproject in #usegalaxy Histories now support advanced searching and offer multiple other improvements. [wiki.galaxyproject.org/Histories](http://wiki.galaxyproject.org/Histories) [go.twitter.com/1PBNYMSQ](https://twitter.com/1PBNYMSQ) 81 Show Photo

Yannick Worms @yannick\_worms 12 Mar Fun genome Bioinformatics #job in

History

search histories

Unnamed history

1 shown

0 items

Annotation

Click here to edit annotation

1. ERI.MA.SRR05462

2. File:ftp://ftp.ebi.ac.uk/pub/ena/trace/SRR054/SRR054622/SRR054622.fastq.gz

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**Galaxy 101**  
Start small  
The very first tutorial you need

Tweets

PyPI Recent Updates @py\_updates in  
planemo 0.6.0: Command-line utilities to assist in building tools for the Galaxy project #fhs-galaxytr ... bit.ly/100CkYI  
13 Retweeted by Galaxy Project  
Retweet

Galaxy Project @galaxyproject in  
#usegalaxy Histories now support advanced searching and offer multiple other improvements. [wiki.galaxyproject.org/Histories](http://wiki.galaxyproject.org/Histories) [go.twitter.com/1JRN7NSQ](https://twitter.com/1JRN7NSQ)  
@ Show Photo

Yannick Worms @yannick... 12 Mar  
Fun genome Bioinformatics #job in  
Tweet to @galaxyproject

History

search histories

Unnamed history  
1 shown

0 items

Annotation  
Click here to edit annotation

1. EBLMA, SRR05462  
2. EBL, flp., Thauria, et al.  
3. HGI, Tasa, SRR054, SRR054622,  
SRR054622.fasta.gz

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 Retweet
- Galaxy Project** @galaxyproject in #usegalaxy Histories now support advanced searching and offer multiple other improvements. [wiki.galaxyproject.org/Histories](http://wiki.galaxyproject.org/Histories) [go.twitter.com/LJRN1Y6SQ](https://twitter.com/LJRN1Y6SQ) 81 Show Photo
- Yannick Worms** @yannick\_worms 12 Mar Fun genome Bioinformatics #job in  
 Tweet to @galaxyproject

History

search histories

Unnamed history

1 shown

394.8 MB

Annotation

Click here to edit annotation

LJRN.SMA.SRR054622.1

IN.PBI.LT.A.STR40LAC.uk

Zval.L.Taxa.SRR054.SRR054622.SM

R054622.fastq.gz

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- Motif Tools
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Workflows Convert Formats Database Annotations

### Edit Attributes

Name:  
EBI SRA: SRR054622 File: ftp://ftp.sra.eric.edu

Info:

Annotation / Notes:

Add an annotation or notes to a dataset; annotations are available when a history is viewed.

Database / Build:  
----- Additional Species Are Below -----

Save

Auto-detect

This will inspect the dataset and attempt to correct the above column values if they are not accurate.

History

search histories

Unnamed history  
1 shown  
194.8 MB

Annotation:  
Click here to edit annotation

1. EBI SRA: SRR054622  
File: ftp://ftp.sra.eric.edu  
wb:vcf;Type: SRR054: SRR054622;  
SRR054622.fastq.gz

https://usegalaxy.org/tracktools/tracktools.html?toolId=galaxy\_toolbox\_beta

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Attributes | Convert Format | **Display** | Recalculate

### Change data type

New Type:

String

This will change the datatype of the existing dataset but not modify its contents. Use this if Galaxy has incorrectly guessed the type of your dataset.

Save

History

search datasets

Unnamed history

1 shown

394.8 MB

Annotation

Click here to full annotation

1. EN 356, 388054622

File: /gal/Toolshed/UCSC/ensembl/356/388054622/388054622.fa.gz

https://usegalaxy.org/trackview/356/388054622/388054622/388054622.fa.gz

The screenshot shows the Galaxy web interface at <https://usegalaxy.org>. The main content area displays the 'Change data type' tool configuration. The 'New Type' dropdown menu is open, showing a list of data types with 'Samvanger' selected. A warning message states: 'Dataset but not modify its contents. Use this if Galaxy has incorrectly guessed the type of your dataset.' The left sidebar contains a 'Tools' menu with categories such as 'Get Data', 'Text Manipulation', 'Genomics', and 'Statistics'. The right sidebar shows the 'History' panel with a search bar and a list of datasets, including one named '1. EN 356, 38024622' with a size of 394.8 MB.

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sniff  
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EMBOSS  
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FASTA manipulation  
Evolution  
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Microarray analysis  
Motif Tools  
NGS TOOLBOX BETA

Attributes Convert Format Database Reanalysis

Change data type

New Type

Text

Samvanger

Dataset but not modify its contents. Use this if Galaxy has incorrectly guessed the type of your dataset.

History

search datasets

Unnamed history  
1 shown

394.8 MB

Annotation  
Click here to full annotation

1. EN 356, 38024622  
File: /gal:/galaxy01/1. EN 356, 38024622  
38024622.fa.gz

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- NGS, Picard
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Attributes Convert Format Database Annotations

Change data type

New Type

Namesanger

This will change the database of the existing dataset but not modify its contents. Use this if Galaxy has incorrectly guessed the type of your dataset.

Save

History

search datasets

Unnamed history

1 shown

394.8 MB

Annotation

Click here to edit annotation

1. EN 356, 588054622. File: Pa1\_Tp13146146. wk:YSL\_Tp13146146\_588054,588054622; 588054622\_764646

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- Met. Tools
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Changed the type of dataset EB\_SRA\_SRR054622 File: ftp://ftp.sra.ftp.nih.gov/ftp2/SRA/SRR054/SRR054622/SRR054622.fastq.gz to fastq-compr

Attributes | Convert Format | Database | Picardtools

### Edit Attributes

**Name:**  
EB\_SRA\_SRR054622 File: ftp://ftp.sra/

**Info:**

**Annotation / Notes:**

Add an annotation or notes to a dataset; annotations are available when a history is viewed.

**Database / Build:**  
----- Additional Species Are Below -----

Save

Auto-convert

This will inspect the dataset and attempt to correct the above column values, if they are not accurate.

History

search histories

Unnamed history  
1 shown

194.8 MB

Annotation:  
Click here to edit annotation

EB\_SRA\_SRR054622 File: ftp://ftp.sra.ftp.nih.gov/ftp2/SRA/SRR054/SRR054622/SRR054622.fastq.gz

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- BBTools
- Genome Diversity
- EMBOSS
- Regional Variation
- FASTA manipulation
- Evolution
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- Microarray analysis
- Met. Tools
- NGS TOOLBOX BETA

Changed the type of dataset EN SRA, SRX014622 File: ftp://ftp.sra.ftp.nih.gov/1/fastq/ERR014/SRR014622.fastq.gz to fastq.gz

Attributes | Convert Format | Delete | Refresh

### Edit Attributes

**Name:**  
EN SRA, SRX014622 File: ftp://ftp.sra.

**Info:**

**Annotation / Notes:**

Add an annotation or notes to a dataset; annotations are available when a history is viewed.

**Database / Build:**  
----- Additional Species Are Below -----

Save

Auto-convert

This will inspect the dataset and attempt to correct the above column values, if they are not accurate.

History

search datasets

Unnamed history  
1 shown

194.8 MB

Annotation:  
Click here to edit annotation

1: EN SRA, SRX014622 (1) + ✕  
In: ftp://ftp.sra.ftp.nih.gov/1/fastq/SRR014/SRR014622.SRR014622.fastq.gz

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- NGS Peak
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**kmer-SVM**  
Galaxy server for identifying predictive regulatory sequence features in NGS data

**Galaxy / Beer Lab**

Yuan Le Bras (@yuanlebr) 26m  
With @cmorjeaud we love the new @galaxyproject release! Using Kmer-SVM from @khorling on @biogerovest Galaxy Instance [pic.twitter.com/NGkWhuqR](https://pic.twitter.com/NGkWhuqR)  
RT Retweeted by Galaxy Project  
Show Photo

Nate Conroy (@nconroy) 1h  
It's official, #usegalaxy finally has a new version! This is 15.03.1: RT @galaxyproject The March 2015 Release is here! [bit.ly/galaxy201503](https://bit.ly/galaxy201503)  
RT Retweeted by Galaxy Project  
Expand

Tweet to @galaxyproject

History

search history

Unnamed history  
2 shown, 1 hidden

721.5 MB

3. rmdup on data.3.16.8 EDI

1. RM\_SRA\_SRR054642.F1

10. 10.1.10.10.10.10.10.10

10.1.10.10.10.10.10.10

10.1.10.10.10.10.10.10

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https://usegalaxy.org/workflows/td\_published

Galaxy | Published Workflows

https://usegalaxy.org/workflow/list\_published

Galaxy [Analysis Tools](#) [Workflows](#) [Shared Data](#) [Visualization](#) [Cloud](#) [Help](#) [Users](#) Using On

### Published Workflows

Search name, annotation, owner, and tag

Advanced Search

Name	Annotation	Owner	Community Rating	Community Tags	Last Updated
<a href="#">Omics Preprocessing</a>		robertobouhail	☆☆☆☆		-3 minutes ago
<a href="#">Galaxy Variant 303</a>		galaxyproject	☆☆☆☆		Mar 06, 2015
<a href="#">Trim Reads Based on Quality (imported from uploaded file)</a>		jhm	☆☆☆☆		Feb 28, 2015
<a href="#">RNAseq/TRAFLINE</a>	RNA sequencing data analysis in a Transparent Reproducible and Automated Pipeline - TRAFLINE	meuffen	☆☆☆☆☆	<a href="#">RNA</a> <a href="#">FASTQ</a> <a href="#">FASTA</a> <a href="#">bioRxiv</a> <a href="#">summerburst</a>	Feb 25, 2015
<a href="#">TF_aylwin_302504940</a>		aylwin	☆☆☆☆		Feb 19, 2015
<a href="#">BioRxiv121_304</a>		ing-ef	☆☆☆☆	<a href="#">training</a>	Feb 09, 2015
<a href="#">Lab_3_Bioinformatics</a>		moreno	☆☆☆☆		Feb 07, 2015
<a href="#">Picard Quality stat on BAM</a>		umar	☆☆☆☆		Jan 26, 2015
<a href="#">Quality stat on BAM</a>		umar	☆☆☆☆		Jan 26, 2015
<a href="#">Quality Statistics work flow</a>		umar	☆☆☆☆		Jan 26, 2015
<a href="#">CATK 337 unified genoscope build</a>		umar	☆☆☆☆		Jan 26, 2015
<a href="#">Workflow: hg19 MQ 40 RQ 30</a>		umar	☆☆☆☆		Jan 26, 2015
<a href="#">SAM filter Picard duplicate removal CATK recalibration workflow hg19 successful run</a>		umar	☆☆☆☆		Jan 26, 2015
<a href="#">Workflow: BED test</a>		umar	☆☆☆☆		Jan 26, 2015

https://usegalaxy.org/libraries/index



Galaxy | Published Workflows

https://usegalaxy.org/u/nbertaboukhall/w/ginkgo-grep

Galaxy

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Published Workflows | nbertaboukhall | Ginkgo Preprocessing

Input workflow

### Galaxy Workflow 'Ginkgo Preprocessing'

Annotation

Step	Annotation
<b>Step 1: Input dataset</b>	
Input Dataset	select at runtime
<b>Step 2: Bowtie2</b>	
Is this library mate-paired?	Single-end
FASTQ file	Output dataset 'output' from step 1
Write unaligned reads to separate files?	False
Will you select a reference genome from your history or use a built-in index?	Use a built-in index
Select a reference genome	hg19
Specify the read group for this file?	No
Parameter Settings	Use defaults
<b>Step 3: Filter SAM or BAM, output SAM or BAM</b>	
SAM or BAM file to filter	Output dataset 'output' from step 2
Reader in output	Include reader

https://usegalaxy.org/workflows/imp/af/1024134462a1394

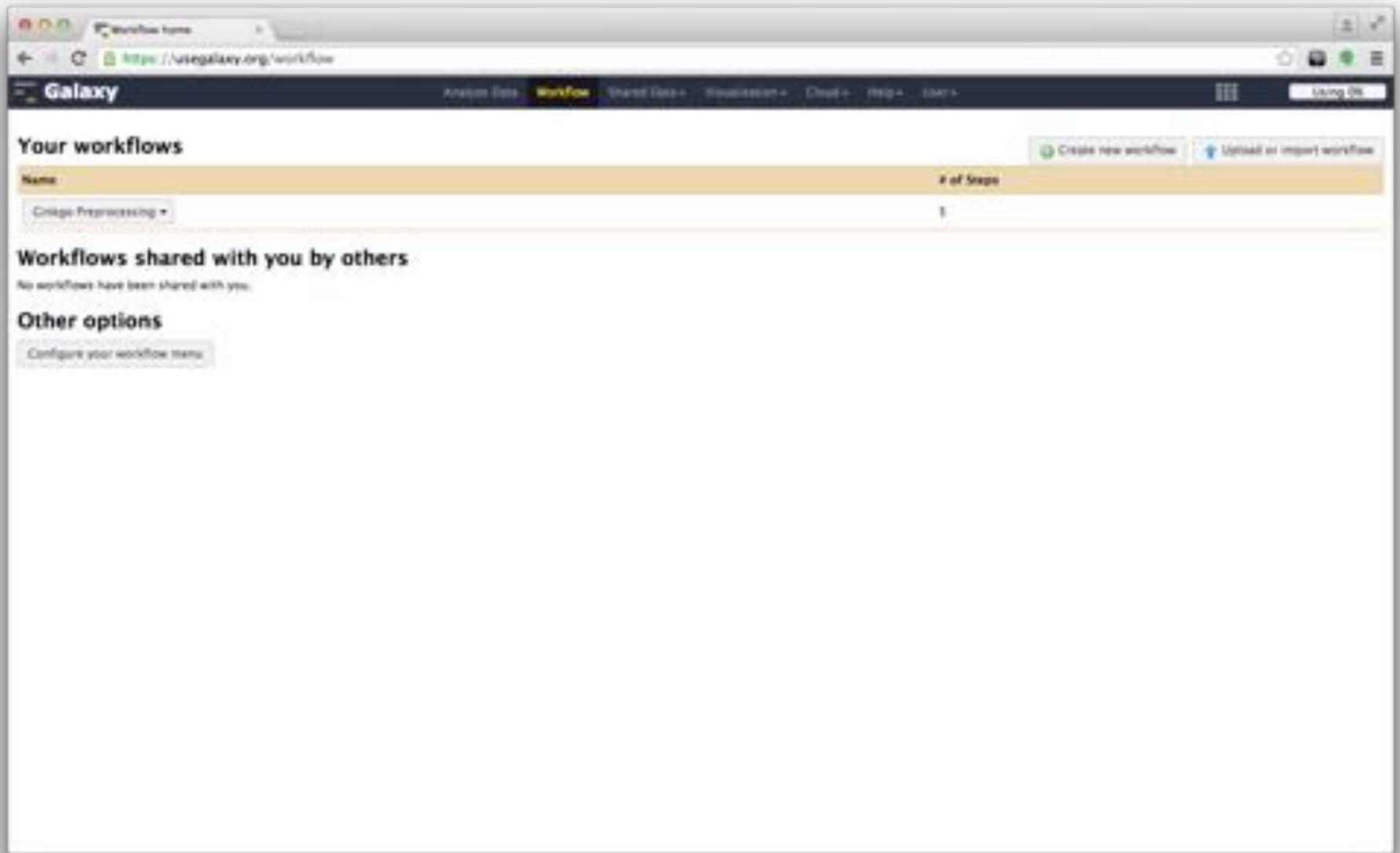
About this Workflow

Author: nbertaboukhall

Related Workflows: All published workflows published workflows by nbertaboukhall

Rating: Community: 0 ratings, 00 examples; Years: 000000

Tags: Community: none; Years: 0000



## Your workflows

[Create new workflow](#) [Upload as import workflow](#)

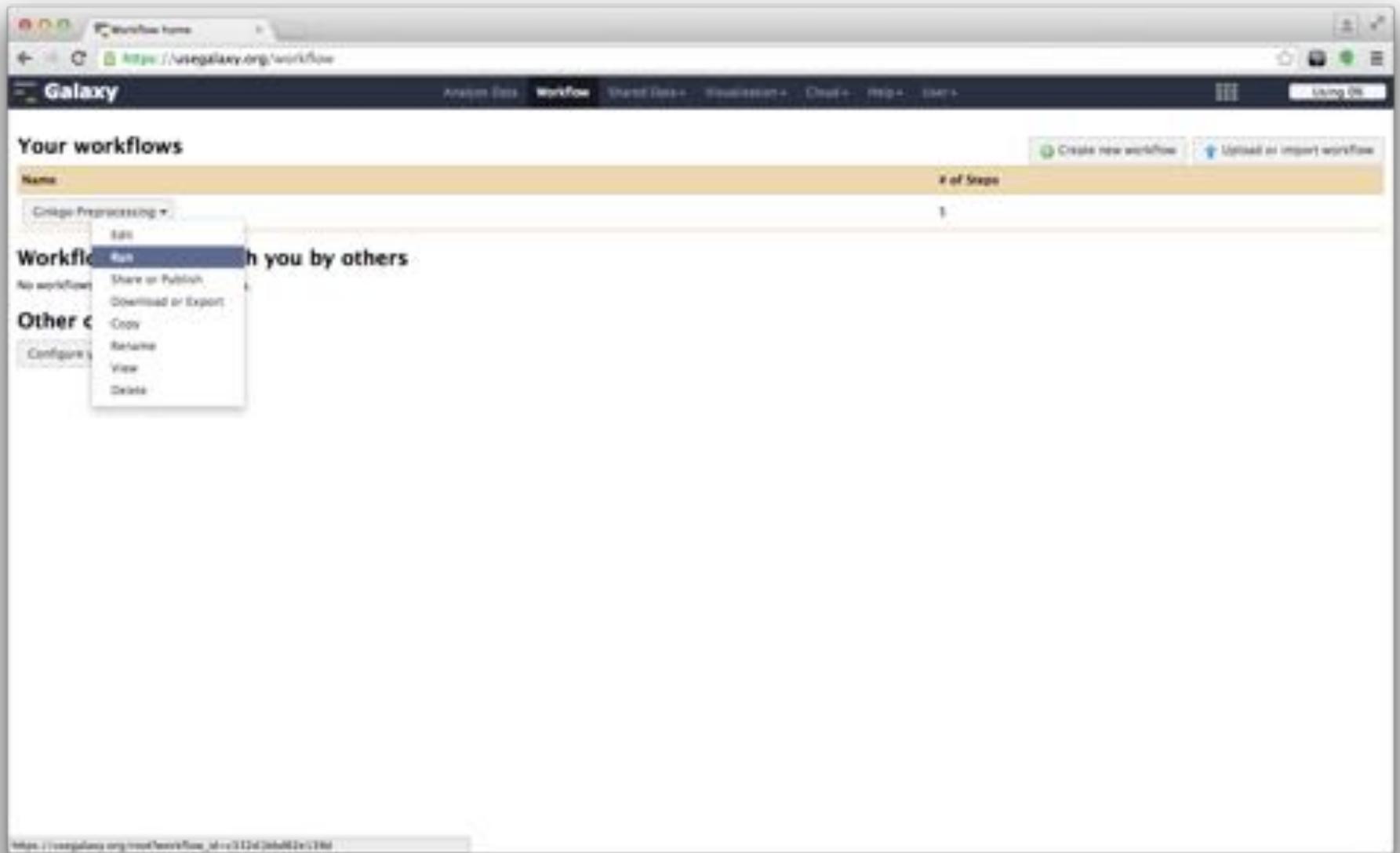
Name	# of Steps
Collage Preprocessing	1

## Workflows shared with you by others

No workflows have been shared with you.

## Other options

[Configure your workflow menu](#)



Galaxy

https://usegalaxy.org/workflow\_id=c332d1bbd62e139d

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### Running workflow "Ginkgo Preprocessing"

Expand All Collapse

Step 1: Input dataset

Input Dataset

1: SRX\_SRA\_SRR054622.Fix File: file:///home/afblac.uk/.galaxy/SRR054/SRR054622/SRR054622.fastq.gz

Fast to Filter

Step 2: Bwa2 (version 0.2)

Step 3: Filter SAM or BAM, output SAM or BAM (version 1.2.0)

Step 4: rmdup (version 1.2.0)

Step 5: Convert from BAM to BED (version 0.1.0)

Send results to a new history

Run workflow

History

search histories

Unnamed history

1 shown

394.8 MB

Annotation

Click here to full annotation

1: SRX\_SRA\_SRR054622.Fix  
file:///home/afblac.uk/.galaxy/SRR054/SRR054622/SRR054622.fastq.gz

Galaxy

https://usegalaxy.org/workflow\_id=c332d1bbd62e139d

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### Running workflow "Ginkgo Preprocessing"

Expand All Collapse

Step 1: Input dataset

Input Dataset

1: EN USA\_SRR054622.Fix.Pa.../Pa.ora.edu.au/seq1/fixpa/SRR054622.SRR054622.fastq.gz

View as File

Step 2: Bwa2 (version 0.2)

Step 3: Filter SAM or BAM, output SAM or BAM (version 1.2.0)

Step 4: rmdup (version 1.2.0)

Step 5: Convert from BAM to BED (version 0.1.0)

Send results to a new history

Run workflow

History

search histories

Unnamed history

1 shown

394.8 MB

Annotation

Click here to edit annotation

1: EN USA\_SRR054622.FI  
Pa. Pa.../Pa.ora.edu.au/seq1/fixpa/SRR054622.SRR054622.SRR054622.fastq.gz

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https://usegalaxy.org/root/workflow\_id=c332d1bbd62e139d

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- Regional Variation
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- Evolution
- Multiple Alignments
- Microarray analysis
- Metil Tools
- NGS TOOLBOX BETA

Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

1. EB\_SRA\_SRR054622.fastq file: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz
2. Bowtie2 on data 1: aligned reads
3. Filter SAM or BAM, output SAM or BAM on data 2: bam
4. rmdup on data 3
5. rmdup on data 3 (as BED)

History

search datasets

Unnamed history  
1 shown

394.8 MB

Annotation  
Click here to edit annotation

- 1. rmdup on data 3 (as BED)
- 5. rmdup on data 3
- 3. Filter SAM or BAM, output SAM or BAM on data 2: bam
- 2. Bowtie2 on data 1: aligned reads
- 1. EB\_SRA\_SRR054622 (1)  
file: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz

Galaxy

https://usegalaxy.org/root/workflow\_id=c332d1bbd62e139d

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Tools

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- NGS BAM Tools
- NGS Filter
- Extract Features
- Fetch Sequences
- Fetch Alignments
- Get Genomic Scores
- Operate on Genomic Intervals
- Statistics
- Graph/Display Data
- Phenotype Association
- snEff
- BEDTools
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1. EB\_SRA\_SRR054622.fastq file: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz
2. Bowtie2 on data 1, aligned reads
3. Filter SAM or BAM, output SAM or BAM on data 2: bam
4. rmdup on data 3
5. rmdup on data 3 (as BED)

History

search datasets

Unnamed history  
3 shown

394.8 MB

Annotation  
Click here to edit annotation

- 1. rmdup on data 3 (as BED)
- 1. rmdup on data 3
- 1. Filter SAM or BAM, output SAM or BAM on data 2: bam
- 1. Bowtie2 on data 1, aligned reads
- 1. EB\_SRA\_SRR054622.fastq file: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz

Galaxy

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- Evolution
- Multiple Alignments
- Microarray analysis
- Metil Tools
- NGS TOOLBOX BETA

Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

- 1: SRX SRX SRX014622 File: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR014/SRR014622/SRR014622.fastq.gz
- 2: Sorted on data 1: aligned reads
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 4: rmdup on data 3
- 5: rmdup on data 3 (as BED)

History

search datasets

Unnamed history  
4 shown, 1 collapsed  
394.8 MB

Annotations  
Click here to edit annotations

- 1: rmdup on data 3 (as BED)
- 4: rmdup on data 3
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 1: SRX SRX SRX014622 File: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR014/SRR014622/SRR014622.fastq.gz

Galaxy

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Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

- 1: SRX SRX0054622 File: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz
- 2: Sorted on data 1: aligned reads
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 4: rmdup on data 3
- 5: rmdup on data 3 (as BED)

History

search datasets

Unnamed history  
4 shown, 1 collapsed  
394.8 MB

Annotations  
Click here to edit annotations

- 1: rmdup on data 3 (as BED)
- 1: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 1: SRX SRX0054622 File: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR054/SRR054622/SRR054622.fastq.gz

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- Genome Diversity
- EMBOSS
- Regional Variation
- FASTA manipulation
- Evolution
- Multiple Alignments
- Microarray analysis
- Metil Tools
- NGS TOOLBOX BETA

Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

- 1: SR: SRR14622.fastq.gz [ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR14/588054/SRR14622/SRR14622.fastq.gz]
- 2: Sorted on data 1: aligned reads
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 4: rmdup on data 3
- 5: rmdup on data 3 (in BDT)

History

search datasets

Unnamed history  
3 shown, 2 hidden

394.8 MB

Annotation  
Click here to edit annotation

- 1: rmdup on data 3 (in BDT)
- 2: rmdup on data 3
- 3: SR: SRR14622.fastq.gz [ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR14/588054/SRR14622/SRR14622.fastq.gz]

Galaxy

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- NGS BAM Tools
- NGS Read
- Extract Features
- Fetch Sequences
- Fetch Alignments
- Get Genomic Stats
- Operate on Genomic Intervals
- Statistics
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- Phenotype Association
- snEff
- BBTools
- Genome Diversity
- EMBOSS
- Regional Variation
- FASTA manipulation
- Evolution
- Multiple Alignments
- Microarray analysis
- Motif Tools
- NGS TOOLBOX BETA

Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

- 1: EB\_SRA\_SRR054622.fastq.gz [v1.1/ftp.ncbi.nlm.nih.gov/trace/trace/SRA/SRR054622/SRR054622.fastq.gz]
- 2: Sorted on data 1: aligned reads
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 4: rmdup on data 3
- 5: rmdup on data 3 (in BDT)

History

search datasets

Unnamed history  
3 shown, 2 hidden

394.8 MB

Annotation  
Click here to edit annotation

- 1: rmdup on data 3 (in BDT)
- 4: rmdup on data 3
- 1: EB\_SRA\_SRR054622.fastq.gz [v1.1/ftp.ncbi.nlm.nih.gov/trace/trace/SRA/SRR054622/SRR054622.fastq.gz]

Galaxy

https://usegalaxy.org/root/workflow\_id=c332d1bbd62e139d

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- FASTA manipulation
- Evolution
- Multiple Alignments
- Microarray analysis
- Metil Tools
- NGS TOOLBOX BETA

Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

- 1: SRP SRX SRX054622 file: ftp://ftp.ncbi.nlm.nih.gov/trace/ftpdir/SRR054622/SRR054622.fastq.gz
- 2: Sorted on data 1: aligned reads
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 4: rmdup on data 3
- 5: rmdup on data 3 (as BED)

History

search datasets

Unnamed history  
2 shown, 1 hidden  
394.8 MB

Annotation  
Click here to edit annotation

1: rmdup on data 3 (as BED) + / x  
1: BED

1: SRP SRX SRX054622 (1) + / x  
file: ftp://ftp.ncbi.nlm.nih.gov/trace/ftpdir/SRR054622/SRR054622.fastq.gz

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- NGS Filter
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- Fetch Alignments
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- snEff
- BBTools
- Genome Diversity
- EMBOSS
- Regional Variation
- FASTA manipulation
- Evolution
- Multiple Alignments
- Microarray analysis
- Met. Tools
- NGS TOOLBOX BETA

Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

- 1: SRX SRX:SRX054622 File: ftp://ftp.ars.ubc.ca/ucsc/ver1/fasta/SRR054622/SRR054622.fastq.gz
- 2: Sorted on data 1: aligned reads
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 4: rmdup on data 3
- 5: rmdup on data 3 (in BDT)

History

search datasets

Unnamed history  
2 shown, 1 hidden  
394.8 MB

Annotation  
Click here to add annotation

- 1: rmdup on data 3 (in BDT)
- 1: SRX SRX:SRX054622 (1)  
ftp://ftp.ars.ubc.ca/ucsc/ver1/fasta/SRR054622/SRR054622.fastq.gz

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https://usegalaxy.org/root/workflow\_id=c332d1bbd62e139d

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- Phenotype Association
- snEff
- BBTools
- Genome Diversity
- EMBOSS
- Regional Variation
- FASTA manipulation
- Evolution
- Multiple Alignments
- Microarray analysis
- Metil Tools
- NGS TOOLBOX BETA

Successfully ran workflow "Omics Preprocessing". The following datasets have been added to the queue:

- 1: SRX SRX:SRX014622 File: ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR014/SRR014622/SRR014622.fastq.gz
- 2: Sorted on data 1: aligned reads
- 3: Filter SAM or BAM, output SAM or BAM on data 2: bam
- 4: rmdup on data 3
- 5: rmdup on data 3 (as BED)

History

search datasets

Unnamed history  
2 shown, 1 hidden

721.5 MB

Annotation  
Click here to edit annotation

3: rmdup on data 3 (as B

320

1: SRX SRX:SRX014622.FI

ftp://ftp.sra.ebi.ac.uk/vol1/fastq/SRR014/SRR014622/SRR014622.fastq.gz



Galaxy

https://usegalaxy.org/workflow\_id=332d1bbd62e139d

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Tools

search tools

- Get Data
- Send Data
- Lift-Over
- Text Manipulation
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- NCL QC and manipulation
- NCL Missing
- NCL BAM Tools
- NCL Picard
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- snEff
- BEDTools
- Genome Diversity
- EMBOSS
- Regional Variation
- EASTA manipulation
- Evolution
- Multicoll. Alignments
- Microarray analysis
- Metil Tools
- NCL TOOLBOX BETA

1	2	3	4	5	6
chr10	61988	62016	SRX054622.203867	30	+
chr10	69090	69141	SRX054622.2024140	30	+
chr10	69098	69146	SRX054622.481587	30	+
chr10	69099	69147	SRX054622.739359	30	+
chr10	72809	72907	SRX054622.1184885	30	-
chr10	82075	82083	SRX054622.2384537	30	-
chr10	101842	101810	SRX054622.2476079	30	-
chr10	108416	108464	SRX054622.301574	15	-
chr10	108416	108464	SRX054622.2321097	25	+
chr10	110108	110116	SRX054622.1050882	34	-
chr10	112529	112567	SRX054622.1459050	38	+
chr10	112525	112573	SRX054622.651721	28	+
chr10	113729	113771	SRX054622.2235401	30	-
chr10	114074	114122	SRX054622.2190838	30	+
chr10	114078	114126	SRX054622.1006253	30	+
chr10	124019	124067	SRX054622.2083880	42	+
chr10	130939	131007	SRX054622.213507	40	+
chr10	132896	132944	SRX054622.1964746	34	+
chr10	132905	132953	SRX054622.181640	35	+
chr10	132910	132958	SRX054622.1188143	34	-
chr10	135289	135337	SRX054622.1757379	42	-
chr10	137966	138014	SRX054622.1142440	42	+
chr10	141733	141781	SRX054622.1035785	42	+
chr10	143964	144012	SRX054622.1722643	42	-
chr10	148479	148527	SRX054622.879161	42	+
chr10	148669	148717	SRX054622.1863231	26	+
chr10	168517	168565	SRX054622.1815104	42	-
chr10	168526	168574	SRX054622.1863154	42	-
chr10	168611	168659	SRX054622.2720413	30	+
chr10	173788	173836	SRX054622.2499973	42	-
chr10	176020	176068	SRX054622.1812131	42	+
chr10	179265	179313	SRX054622.1989607	40	-
chr10	182939	182987	SRX054622.1731700	42	-
chr10	182940	182988	SRX054622.1479478	42	-
chr10	183960	184008	SRX054622.531468	48	+

History

search history

Unnamed history  
2 items, 7 jobs

721.5 MB

Annotations  
Click here to edit annotations

1. rmike on data 3 (cat, BED)

1. SRX SRX054622.F1

In: chr10:108416-108464.chr10:114074-114078.SRX054622.SRX054622.fastq.gz

https://usegalaxy.org/workflow\_id=332d1bbd62e139d



Galaxy [https://usegalaxy.org/workflow\\_id=332d1bbd62e139d](https://usegalaxy.org/workflow_id=332d1bbd62e139d)

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- EASTA manipulation
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- Metagenomic analysis
- Motif Tools
- NCL TOOLBOX BETA

1	2	3	4	5	6
chr10	61988	62016	SRX054622.2038647	30	+
chr10	69090	69141	SRX054622.2024140	30	+
chr10	69098	69146	SRX054622.481587	30	+
chr10	69099	69147	SRX054622.739359	30	+
chr10	72809	72907	SRX054622.1184885	30	-
chr10	82075	82083	SRX054622.2384537	30	-
chr10	101602	101610	SRX054622.2476079	30	-
chr10	108416	108464	SRX054622.301574	15	-
chr10	108416	108464	SRX054622.2321097	25	+
chr10	110108	110116	SRX054622.1050682	34	-
chr10	112529	112567	SRX054622.1459050	38	+
chr10	112525	112573	SRX054622.651721	28	+
chr10	113729	113771	SRX054622.2235401	30	-
chr10	114074	114122	SRX054622.2190838	30	+
chr10	114078	114126	SRX054622.1006253	30	+
chr10	124019	124067	SRX054622.2083880	42	+
chr10	130939	131007	SRX054622.213507	40	+
chr10	132896	132944	SRX054622.1964746	34	+
chr10	132905	132953	SRX054622.181640	35	+
chr10	132910	132958	SRX054622.1168143	34	-
chr10	135289	135337	SRX054622.1757379	42	-
chr10	137966	138014	SRX054622.1142440	42	+
chr10	141733	141781	SRX054622.1035785	42	+
chr10	143964	144012	SRX054622.1722640	42	-
chr10	148479	148527	SRX054622.879161	42	+
chr10	148669	148717	SRX054622.1863231	26	+
chr10	168517	168565	SRX054622.1815104	42	-
chr10	168526	168574	SRX054622.1863158	42	-
chr10	168611	168659	SRX054622.2720413	30	+
chr10	173788	173836	SRX054622.2499973	42	-
chr10	176020	176068	SRX054622.1812131	42	+
chr10	179265	179313	SRX054622.1989607	40	-
chr10	182939	182987	SRX054622.1731700	42	-
chr10	182940	182988	SRX054622.1479478	42	-
chr10	183960	184008	SRX054622.531468	48	+

History search history

Unnamed history  
2 items, 7 objects  
721.5 MB

Annotations  
Click here to add annotations

1. rmBam on data 3 (cat... BEDS  
-1,000,000 regions  
Format: bed, database: hg19

Annotations  
Click here to add annotations

display in IGV View  
display at Ensembl Current  
display at RefSeq track  
display at UCSC track

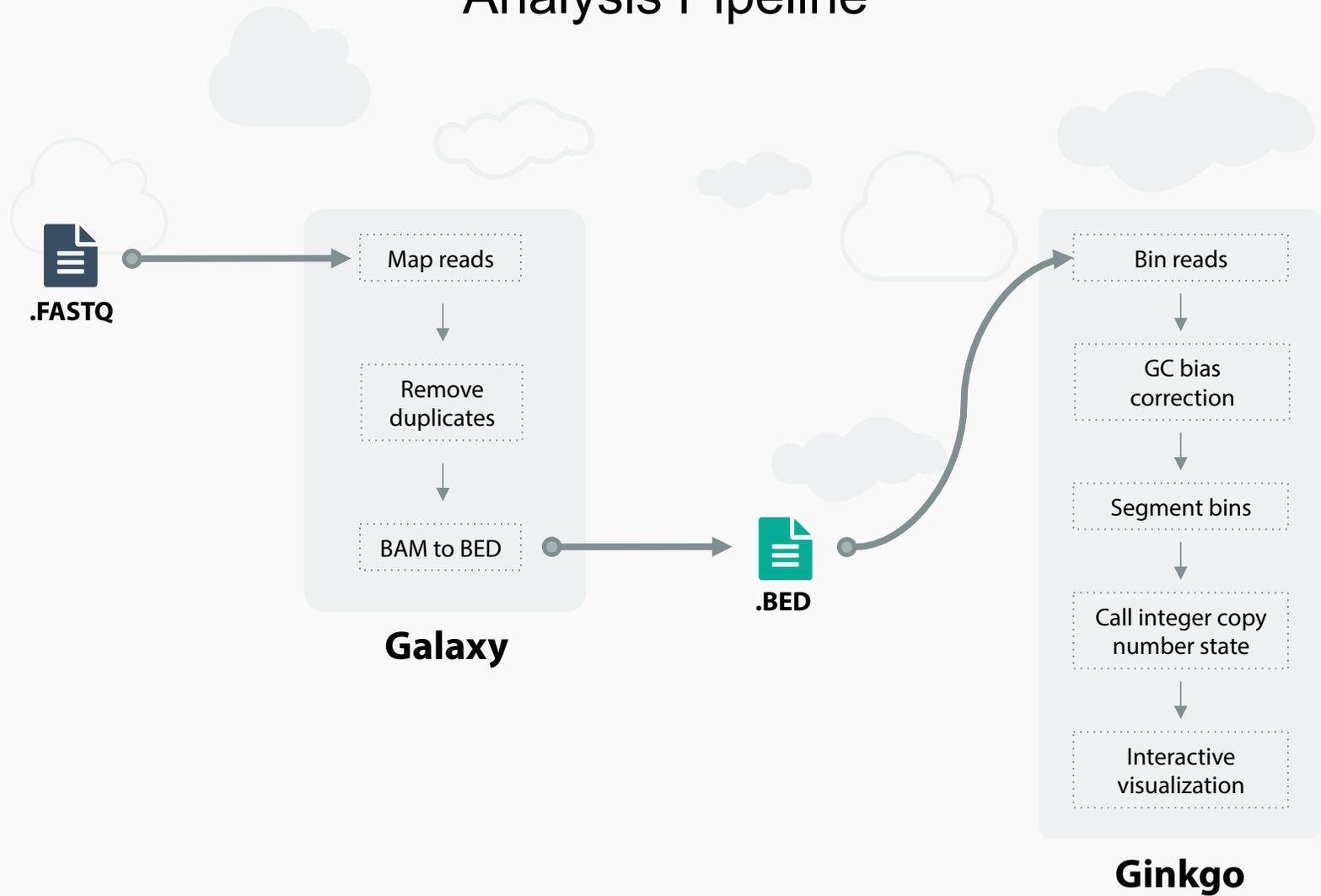
1. Chrm 1, Chr2 1, Chr3 1, Chr4 1, Chr5 1

chr10 61988 62016 SRX054622.2038647  
chr10 69090 69141 SRX054622.2024140  
chr10 69098 69146 SRX054622.481587  
chr10 69099 69147 SRX054622.739359  
chr10 72809 72907 SRX054622.1184885  
chr10 82075 82083 SRX054622.2384537

1. SRM SRM SRX054622.1  
W: 100 / 100000000000  
/var1/Tools/SRX054622.38  
R054622.fastq.gz

https://usegalaxy.org/workflow\_id=332d1bbd62e139d/PanelOf11/Display/na

# Analysis Pipeline



## Hands-on with Ginkgo



**Ginkgo:** [qb.cshl.edu/ginkgo](http://qb.cshl.edu/ginkgo)

**Sample Data:** [qb.cshl.edu/ginkgo/vizbi2015.tar](http://qb.cshl.edu/ginkgo/vizbi2015.tar)