Genome assembly for everyone:

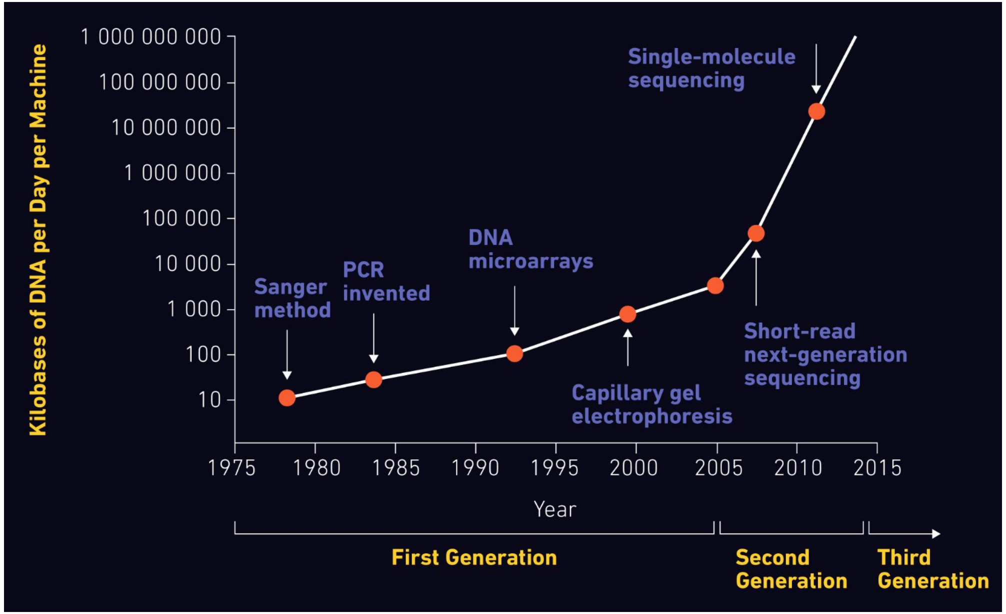
Galaxy Project-VGP collaboration

Cristóbal Gallardo
Galaxy Europe, University of Freiburg

Why do we need to make genome assembly accessible?

Fact:

Recent improvements in sequencing technologies and assembly tools promise to generate high-quality reference genomes for all species.



https://www.technologynetworks.com/genomics/articles/an-overview-of-next-generation-sequencing-346532

Why do we need to make genome assembly accessible?

Problem:

the genome assembly process is still laborious, costly, requires significant expertise.

Why do we need to make genome assembly accessible?

Solution:

make the pipeline freely accessible through the public computational infrastructure (**Galaxy**), and provide the required training (**Galaxy Training Network**).

VGP VERTEBRATE GENOMES PROJECT

A PROJECT OF THE G10K CONSORTIUM

Phase I

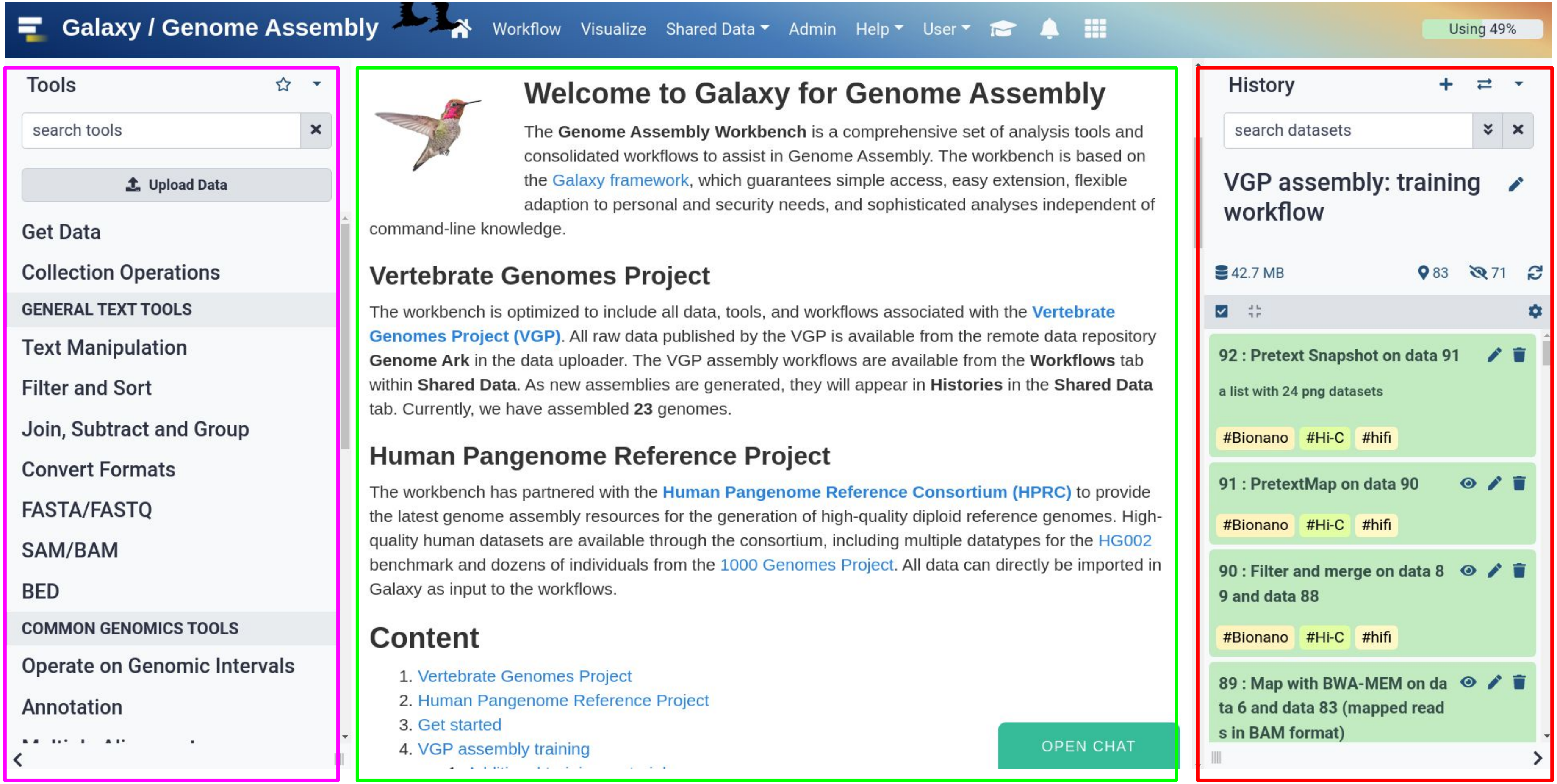
54	58	32	30	90	4
BIRDS	MAMMALS	REPTILES	AMPHIBIANS	FISH	INVERTEBRATES

OVER 260 SPECIES

Galaxy useGalaxy.eu

- **Open source platform** for accessible, reproducible, and transparent computational research
- **Public computational infrastructure** that provides a free analysis environment
    - European server: over 9000 CPU cores, 50TB of RAM, 4PB data storage
- The web-based graphical user interface allows interactive analyses

- **Training infrastructure Service (Tlaas)**
    - Private queue where only your training’s jobs will run
    - See how your students are progressing
- **Galaxy Training Network (GTN) provides training material**



Welcome to Galaxy for Genome Assembly

The **Genome Assembly Workbench** is a comprehensive set of analysis tools and consolidated workflows to assist in Genome Assembly. The workbench is based on the Galaxy framework, which guarantees simple access, easy extension, flexible adaption to personal and security needs, and sophisticated analyses independent of command-line knowledge.

Vertebrate Genomes Project

The workbench is optimized to include all data, tools, and workflows associated with the **Vertebrate Genomes Project (VGP)**. All raw data published by the VGP is available from the remote data repository **Genome Ark** in the data uploader. The VGP assembly workflows are available from the **Workflows** tab within **Shared Data**. As new assemblies are generated, they will appear in **Histories** in the **Shared Data** tab. Currently, we have assembled **23** genomes.

Human Pangenome Reference Project

The workbench has partnered with the **Human Pangenome Reference Consortium (HPRC)** to provide the latest genome assembly resources for the generation of high-quality diploid reference genomes. High-quality human datasets are available through the consortium, including multiple datatypes for the HG002 benchmark and dozens of individuals from the 1000 Genomes Project. All data can directly be imported in Galaxy as input to the workflows.

Content

1. Vertebrate Genomes Project
2. Human Pangenome Reference Project
3. Get started
4. VGP assembly training

92 : Pretext Snapshot on data 91

a list with 24 png datasets

#Bionano #Hi-C #hifi

91 : PretextMap on data 90

#Bionano #Hi-C #hifi

90 : Filter and merge on data 89 and data 88

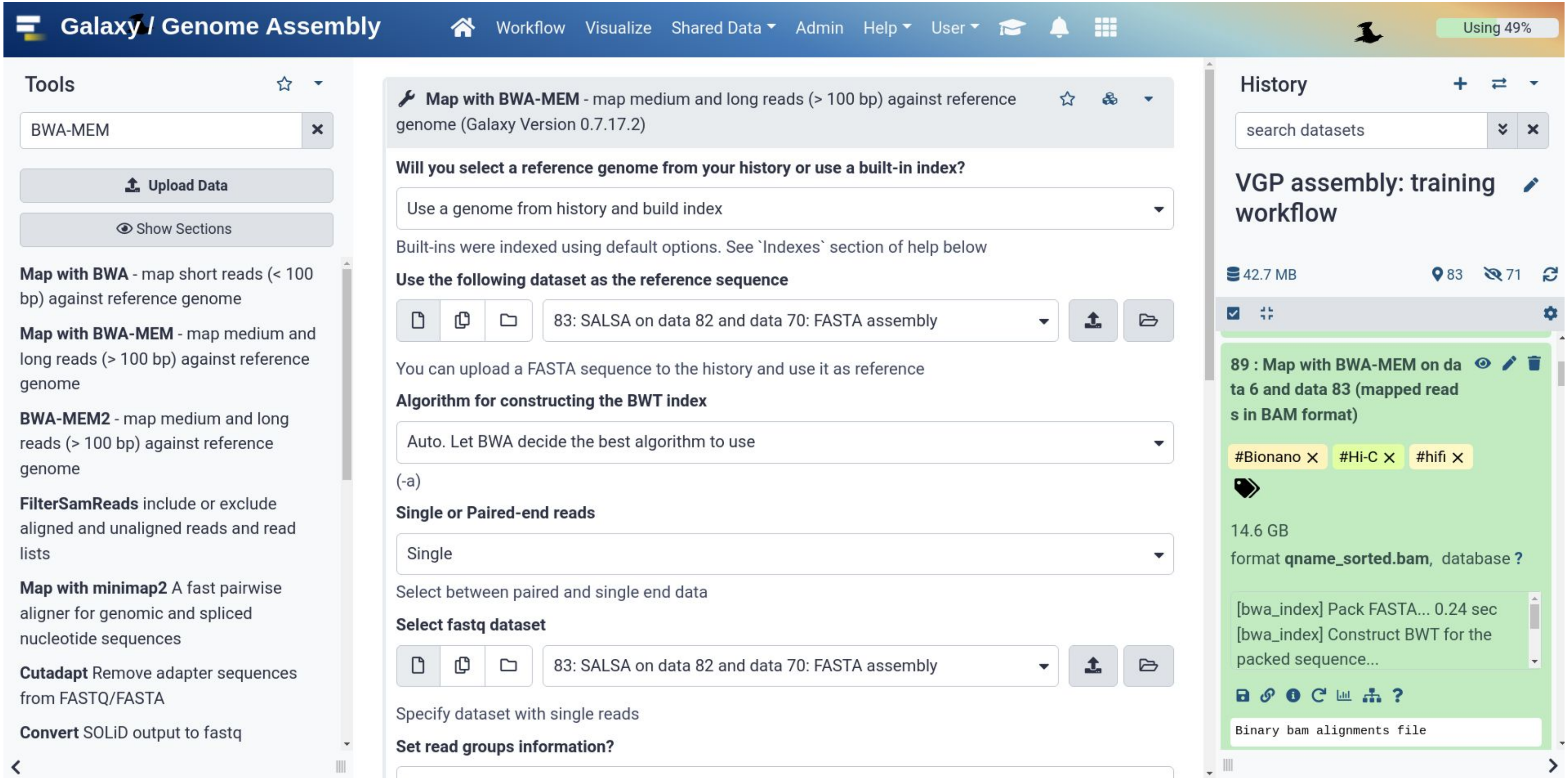
#Bionano #Hi-C #hifi

89 : Map with BWA-MEM on data 6 and data 83 (mapped reads in BAM format)

Tool search panel

View panel

History panel



Map with BWA - map short reads (< 100 bp) against reference genome

Map with BWA-MEM - map medium and long reads (> 100 bp) against reference genome

BWA-MEM2 - map medium and long reads (> 100 bp) against reference genome

FilterSamReads include or exclude aligned and unaligned reads and read lists

Map with minimap2 A fast pairwise aligner for genomic and spliced nucleotide sequences

Cutadapt Remove adapter sequences from FASTQ/FASTA

Convert SOLiD output to fastq

Map with BWA-MEM - map medium and long reads (> 100 bp) against reference genome (Galaxy Version 0.7.17.2)

Will you select a reference genome from your history or use a built-in index?

Use a genome from history and build index

Built-ins were indexed using default options. See `Indexes` section of help below

Use the following dataset as the reference sequence

83: SALSA on data 82 and data 70: FASTA assembly

You can upload a FASTA sequence to the history and use it as reference

Algorithm for constructing the BWT index

Auto. Let BWA decide the best algorithm to use

(-a)

Single or Paired-end reads

Single

Select between paired and single end data

Select fastq dataset

83: SALSA on data 82 and data 70: FASTA assembly

Specify dataset with single reads

Set read groups information?

VGP assembly: training workflow

42.7 MB 83 71

89 : Map with BWA-MEM on data 6 and data 83 (mapped reads in BAM format)

#Bionano #Hi-C #hifi

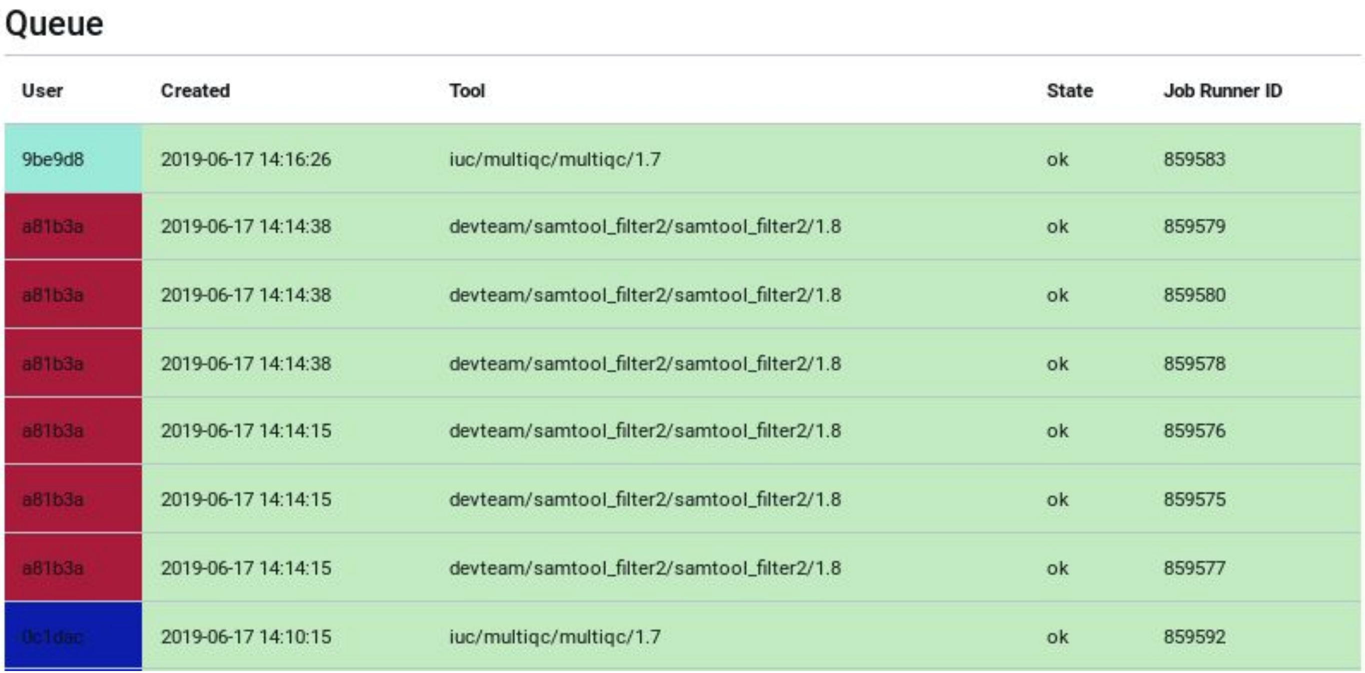
14.6 GB

format **qname_sorted.bam**, database **?**

[bwa_index] Pack FASTA... 0.24 sec
[bwa_index] Construct BWT for the packed sequence...

```
Binary bam alignments file
```

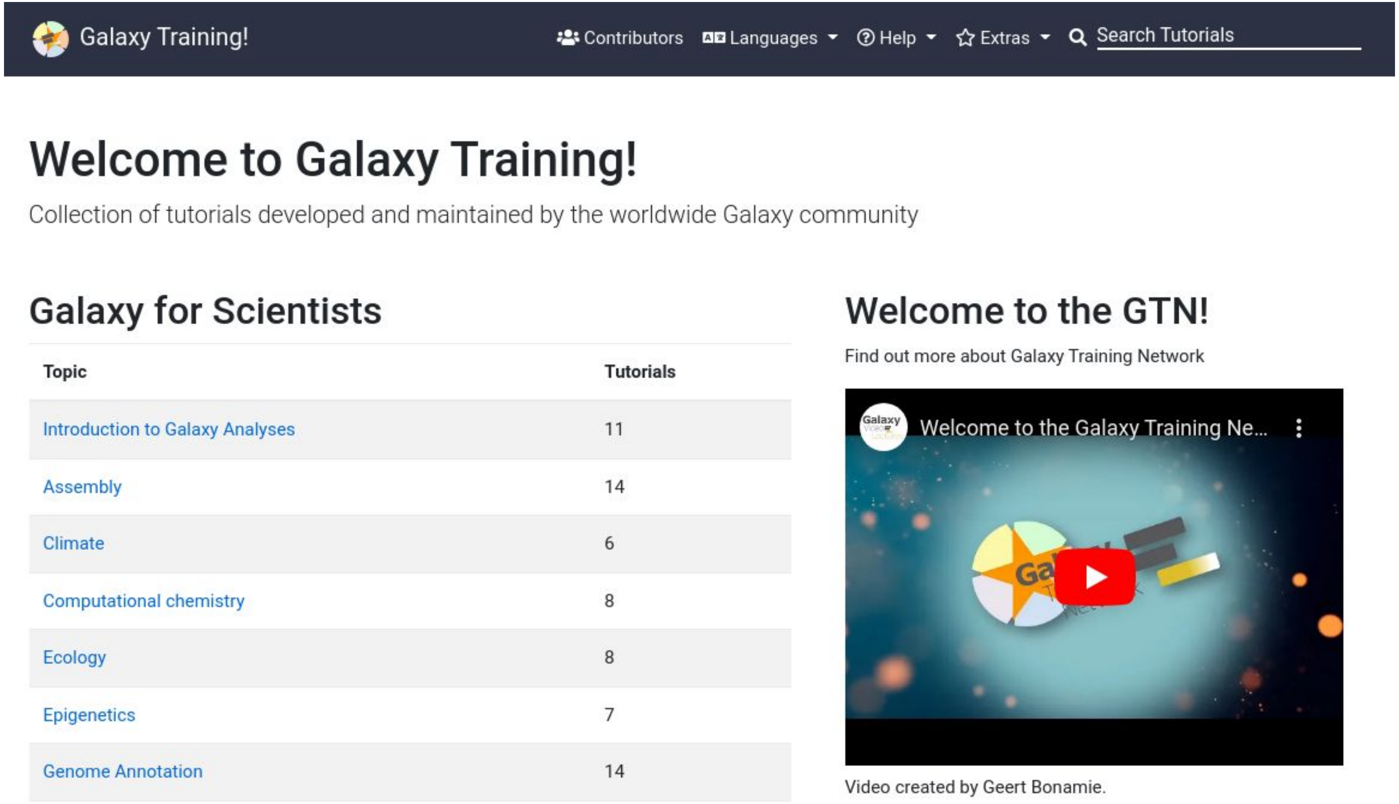
A web interface for each tool, so not **command line skills are required** for performing complex analysis



Queue

User	Created	Tool	State	Job Runner ID
9be9d8	2019-06-17 14:16:26	iuc/multiqc/multiqc/1.7	ok	859583
a81b3a	2019-06-17 14:14:38	devteam/samtool_filter2/samtool_filter2/1.8	ok	859579
a81b3a	2019-06-17 14:14:38	devteam/samtool_filter2/samtool_filter2/1.8	ok	859580
a81b3a	2019-06-17 14:14:38	devteam/samtool_filter2/samtool_filter2/1.8	ok	859578
a81b3a	2019-06-17 14:14:15	devteam/samtool_filter2/samtool_filter2/1.8	ok	859576
a81b3a	2019-06-17 14:14:15	devteam/samtool_filter2/samtool_filter2/1.8	ok	859575
a81b3a	2019-06-17 14:14:15	devteam/samtool_filter2/samtool_filter2/1.8	ok	859577
0c1dac	2019-06-17 14:10:15	iuc/multiqc/multiqc/1.7	ok	859592

Jobs assigned to training groups preferentially run on a training machine with **dedicated resources**.



Welcome to Galaxy Training!

Collection of tutorials developed and maintained by the worldwide Galaxy community

Galaxy for Scientists

Topic	Tutorials
Introduction to Galaxy Analyses	11
Assembly	14
Climate	6
Computational chemistry	8
Ecology	8
Epigenetics	7
Genome Annotation	14

Welcome to the GTN!

Find out more about Galaxy Training Network

Video created by Geert Bonamie.

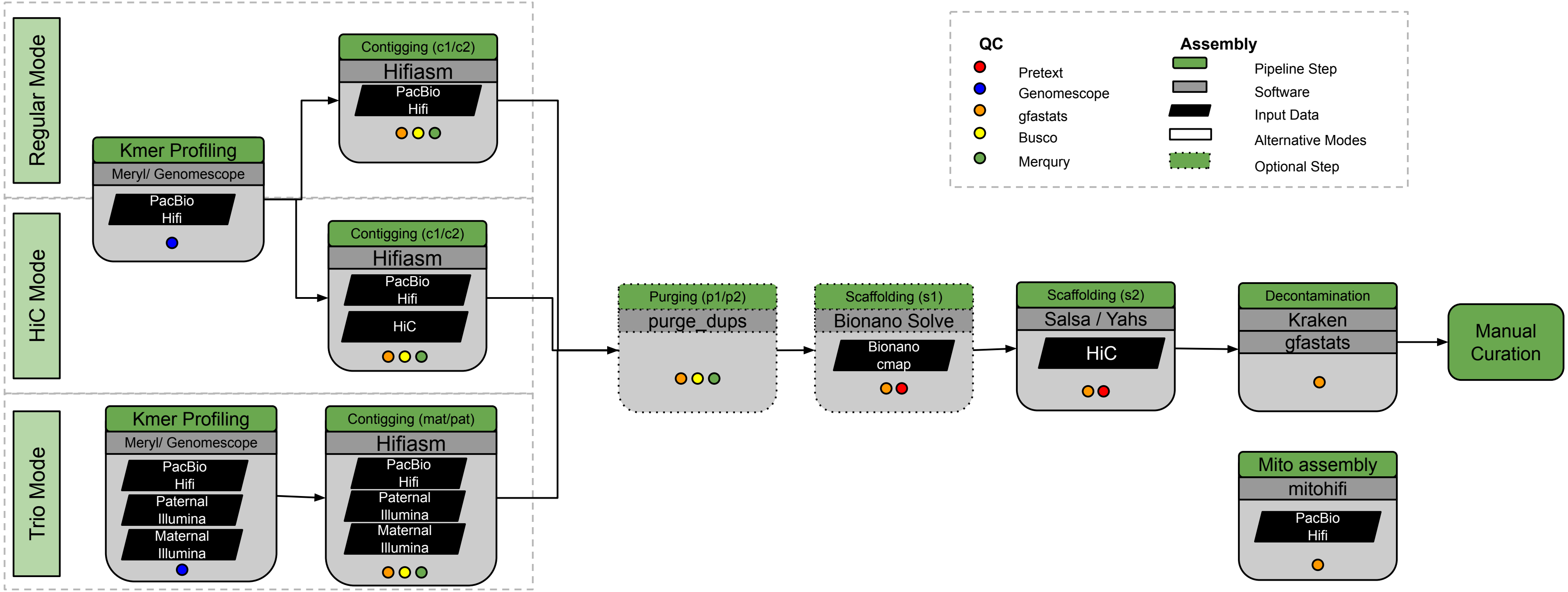
https://training.galaxyproject.org/

The GTN Materials in May 2022 has 260+ tutorials covering 23 topics, developed by over 260+ contributors!

GTN tutorial are characterized by:

- Constructed around a **real world research problem**
- **Self-contained**: [illegible] needed to complete them is bundled with the tutorial
- [illegible]
- No software installation is needed!

VGP VERTEBRATE GENOMES PROJECT



Regular Mode

HiC Mode

Trio Mode

Kmer Profiling
Meryl/ Genomescope
PacBio Hifi
●

Contigging (c1/c2)
Hifiasm
PacBio Hifi
● ● ●

Contigging (c1/c2)
Hifiasm
PacBio Hifi
HiC
● ● ●

Kmer Profiling
Meryl/ Genomescope
PacBio Hifi
Paternal Illumina
Maternal Illumina
●

Contigging (mat/pat)
Hifiasm
PacBio Hifi
Paternal Illumina
Maternal Illumina
● ● ●

Purging (p1/p2)
purge_dups
● ● ●

Scaffolding (s1)
Bionano Solve
Bionano cmap
● ●

Scaffolding (s2)
Salsa / Yahs
HiC
● ●

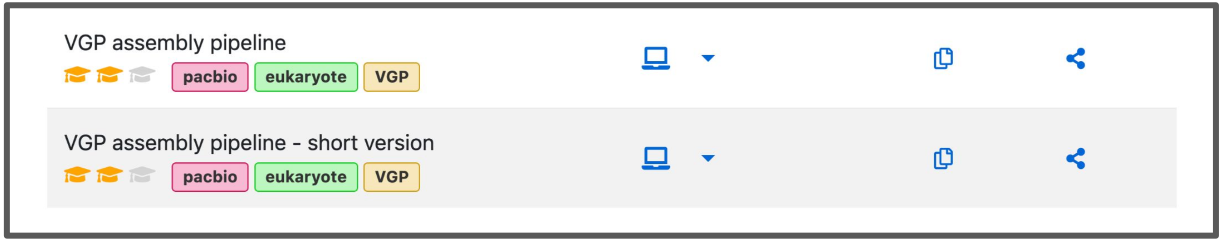
Decontamination
Kraken
gfastats
●

Manual Curation

Mito assembly
mitohifi
PacBio Hifi
●

QC		Assembly	
🟥	Pretext	🟩	Pipeline Step
🟦	Genomescope	▨	Software
🟧	gfastats	■	Input Data
🟨	Busco	□	Alternative Modes
🟩	Merqury	▩	Optional Step

Two training version available: extended and workflow-focused

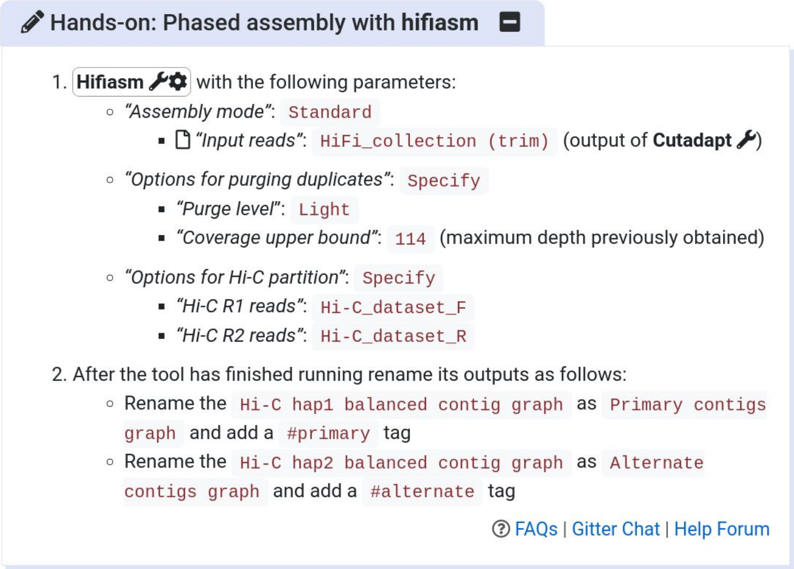


VGP assembly pipeline

pacbio | eukaryote | VGP

VGP assembly pipeline - short version

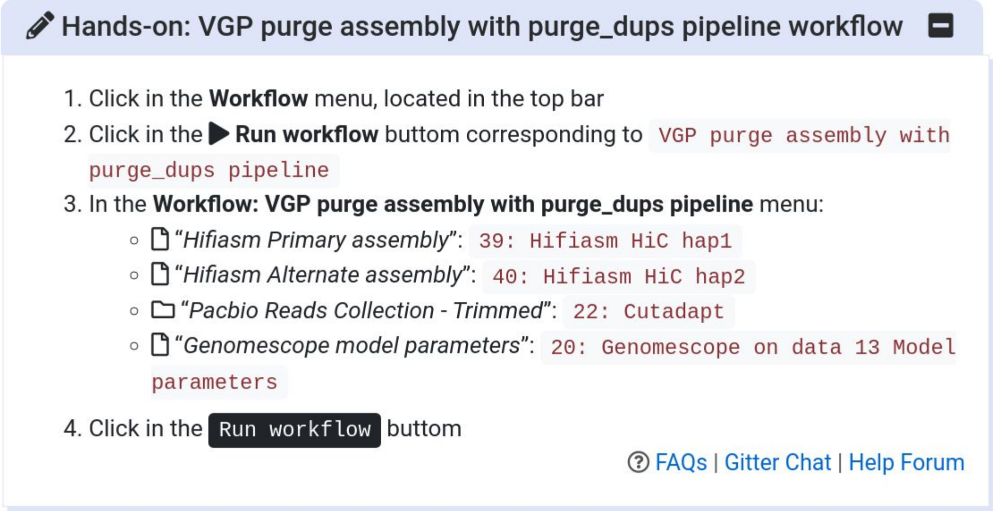
pacbio | eukaryote | VGP



Hands-on: Phased assembly with hifiasm

1. **Hifiasm** with the following parameters:
   - *"Assembly mode"*: `Standard`
     - *"Input reads"*: `HiFi_collection (trim)` (output of **Cutadapt**)
   - *"Options for purging duplicates"*: `Specify`
     - *"Purge level"*: `Light`
     - *"Coverage upper bound"*: `114` (maximum depth previously obtained)
   - *"Options for Hi-C partition"*: `Specify`
     - *"Hi-C R1 reads"*: `Hi-C_dataset_F`
     - *"Hi-C R2 reads"*: `Hi-C_dataset_R`
2. After the tool has finished running rename its outputs as follows:
   - Rename the `Hi-C hap1 balanced contig graph` as `Primary contigs graph` and add a `#primary` tag
   - Rename the `Hi-C hap2 balanced contig graph` as `Alternate contigs graph` and add a `#alternate` tag

FAQs | Gitter Chat | Help Forum

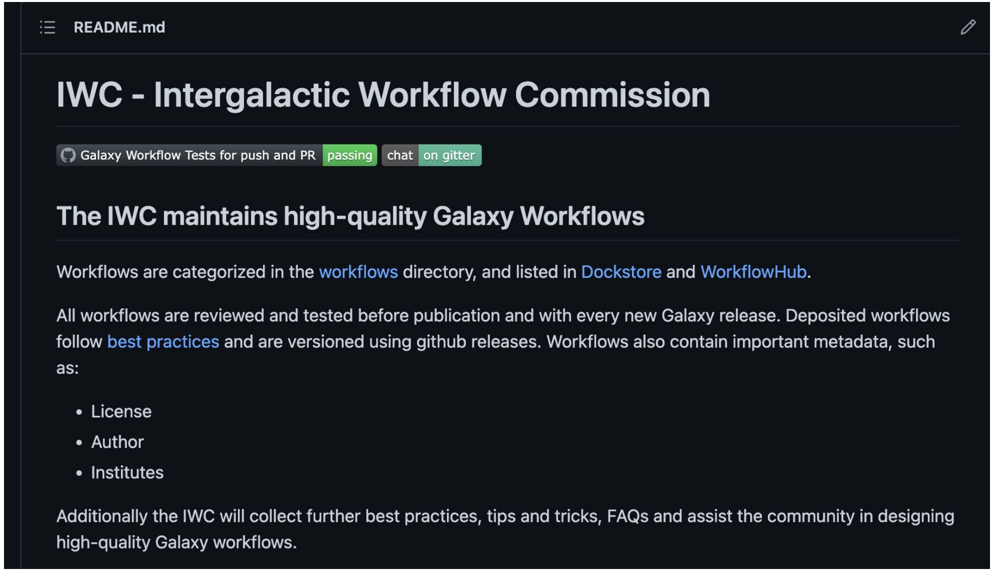


Hands-on: VGP purge assembly with purge_dups pipeline workflow

1. Click in the **Workflow** menu, located in the top bar
2. Click in the ▶ **Run workflow** buttom corresponding to `VGP purge assembly with purge_dups pipeline`
3. In the **Workflow: VGP purge assembly with purge_dups pipeline** menu:
   - *"Hifiasm Primary assembly"*: `39: Hifiasm HiC hap1`
   - *"Hifiasm Alternate assembly"*: `40: Hifiasm HiC hap2`
   - *"Pacbio Reads Collection - Trimmed"*: `22: Cutadapt`
   - *"Genomescope model parameters"*: `20: Genomescope on data 13 Model parameters`
4. Click in the `Run workflow` buttom

FAQs | Gitter Chat | Help Forum

Workflow Availability: IWC



README.md

IWC - Intergalactic Workflow Commission

Galaxy Workflow Tests for push and PR passing | chat on gitter

The IWC maintains high-quality Galaxy Workflows

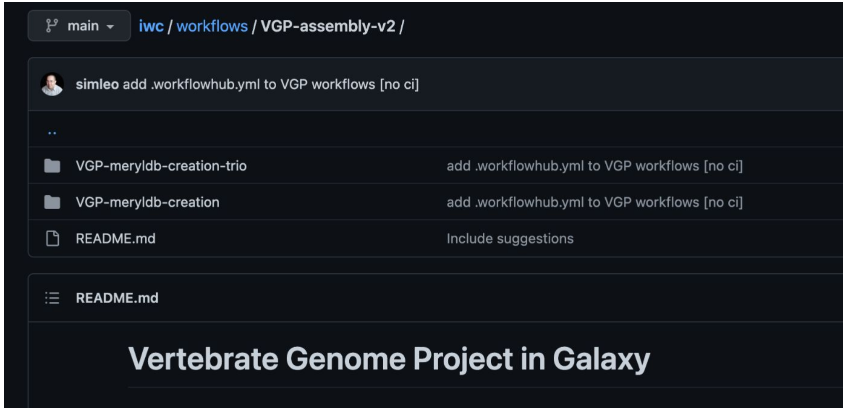
Workflows are categorized in the workflows directory, and listed in Dockstore and WorkflowHub.

All workflows are reviewed and tested before publication and with every new Galaxy release. Deposited workflows follow best practices and are versioned using github releases. Workflows also contain important metadata, such as:

- License
- Author
- Institutes

Additionally the IWC will collect further best practices, tips and tricks, FAQs and assist the community in designing high-quality Galaxy workflows.

https://github.com/galaxyproject/iwc



main iwc / workflows / VGP-assembly-v2 /

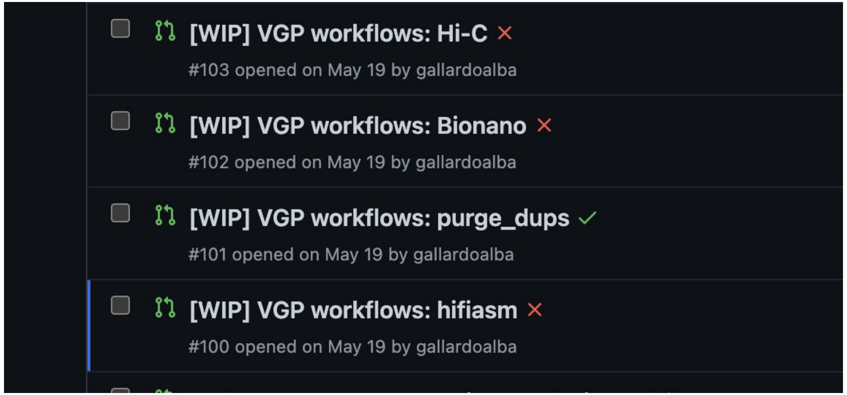
simleo add .workflowhub.yml to VGP workflows [no ci]

..

VGP-meryldb-creation-trio	add .workflowhub.yml to VGP workflows [no ci]
VGP-meryldb-creation	add .workflowhub.yml to VGP workflows [no ci]
README.md	Include suggestions

README.md

Vertebrate Genome Project in Galaxy



□ [WIP] VGP workflows: Hi-C ✗
#103 opened on May 19 by gallardoalba

□ [WIP] VGP workflows: Bionano ✗
#102 opened on May 19 by gallardoalba

□ [WIP] VGP workflows: purge_dups ✓
#101 opened on May 19 by gallardoalba

□ [WIP] VGP workflows: hifiasm ✗
#100 opened on May 19 by gallardoalba

Genomes Assembled on public Galaxy instances

- 21 Genomes in 6 months
  - 10 birds, 2 amphibians, 2 fish, 6 mammals, 1 reptile
  - 5 more in the works

Florida Museum photos by
Kenneth Krysko

Jacob Drucker
Hawaii, United States
Macaulay Library ML 141519531

Acknowledgments

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- Giulio Formenti
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- Bjorn Grüning
- Michael Schatz
- And everyone else