How to Use the Human Pangenome Reference in AnVIL

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Human Pangenome Reference Consortium (HPRC)

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

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Financial Disclosure

ALICE POPEJOY & JEAN MONLONG

Do not have any relevant disclosures.

Financial Disclosure

JULIAN LUCAS

Discloses the following relevant relationships with ineligible companies. Any potential interests have been mitigated:

Independent contractor (including contracted research) relationship with Prime Genomics (has ended).

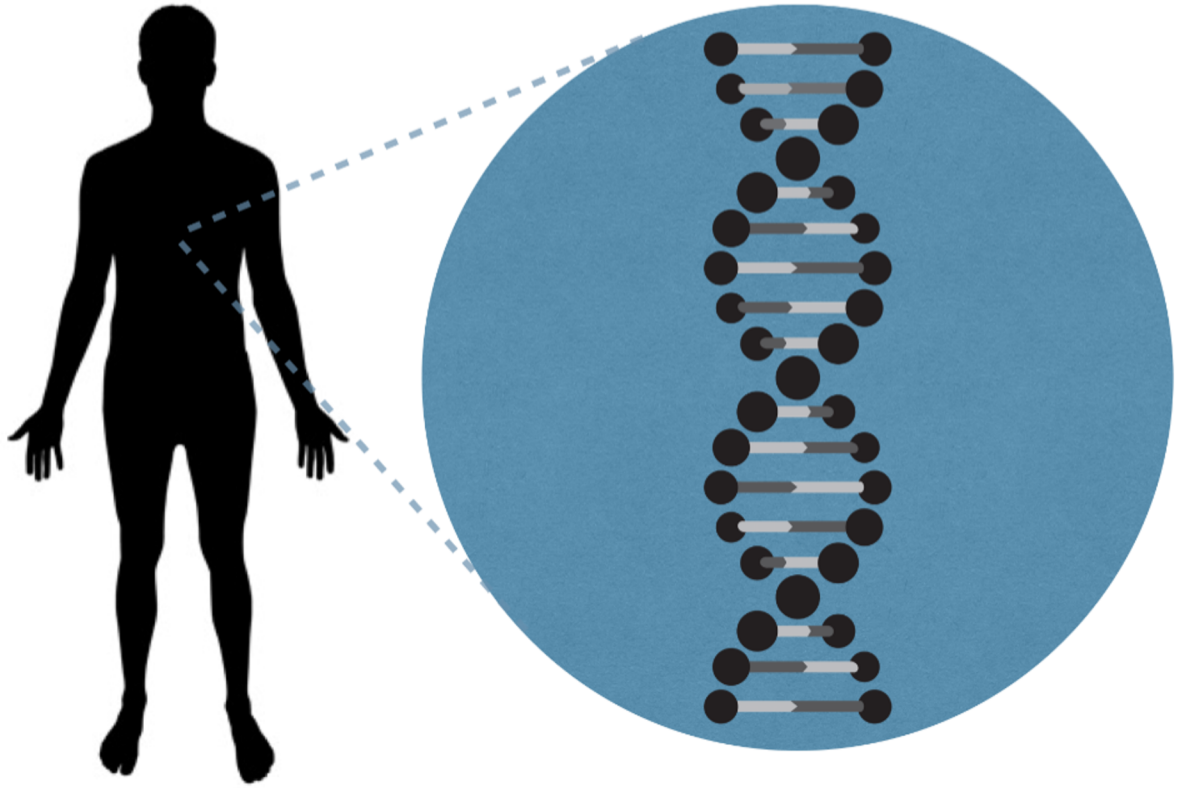
Workshop Logistics

Slides 10 minutes	Introduction To The Human Pangenome Reference Consortium
Discussion 15 minutes	Using The Human Reference Genome In Clinical Genetics
Slides 10 minutes	Overview of AnVIL and HPRC Resources
Demo 40 minutes	How To Use Pangenomes To Improve (Clinical) Variant Calling
Discussion 15 minutes	The Pangenome Reference For Clinical Variant Interpretation

Introduction To The Human Pangenome Reference Consortium

The Initial Human Genome Project

The Human Genome Project: **international, collaborative research program** whose goal was the complete mapping and understanding of all the genes of human beings.



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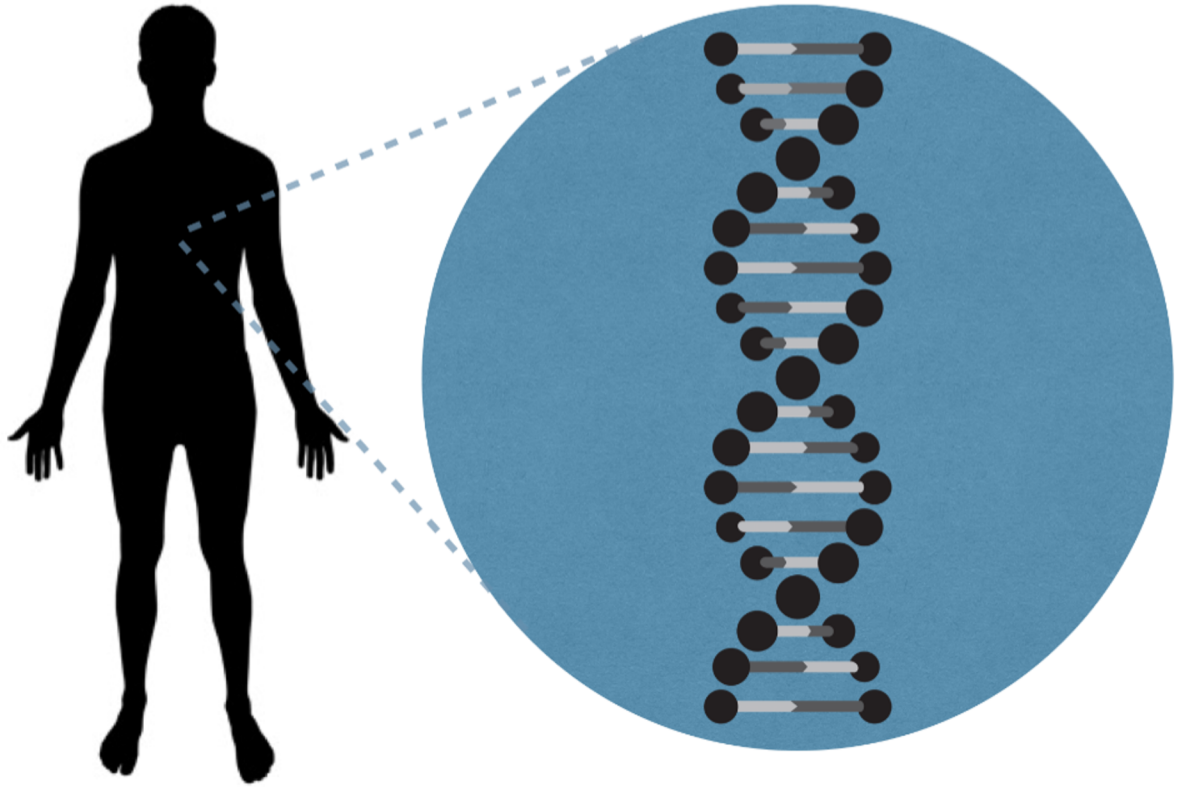
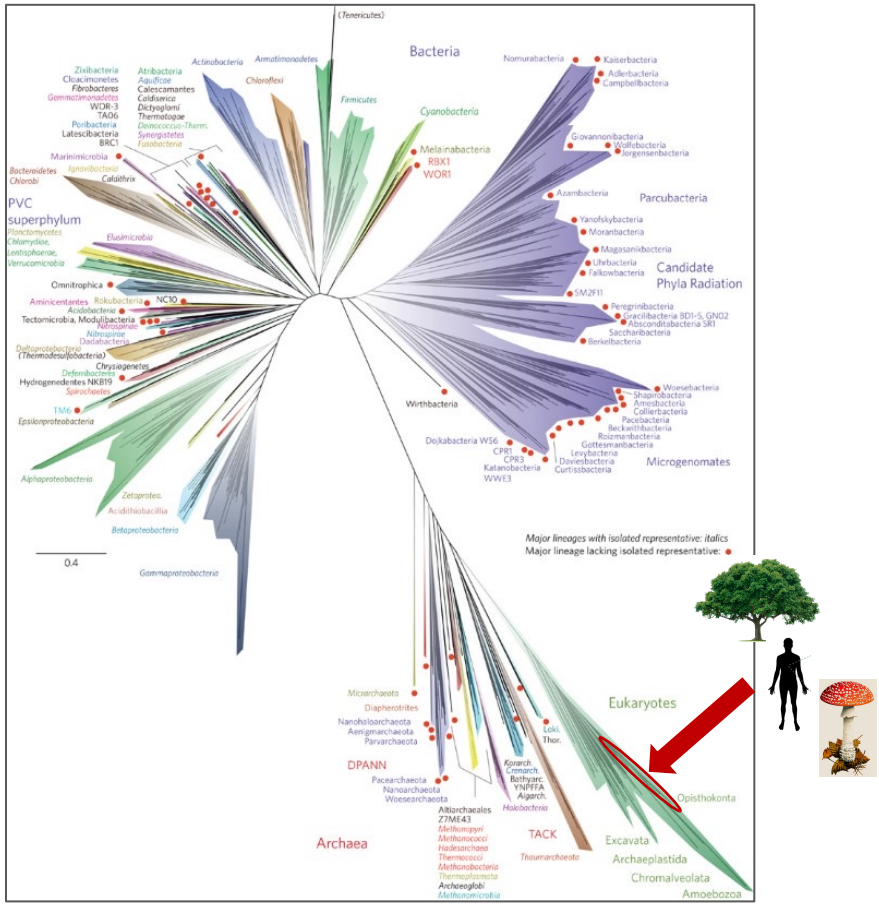
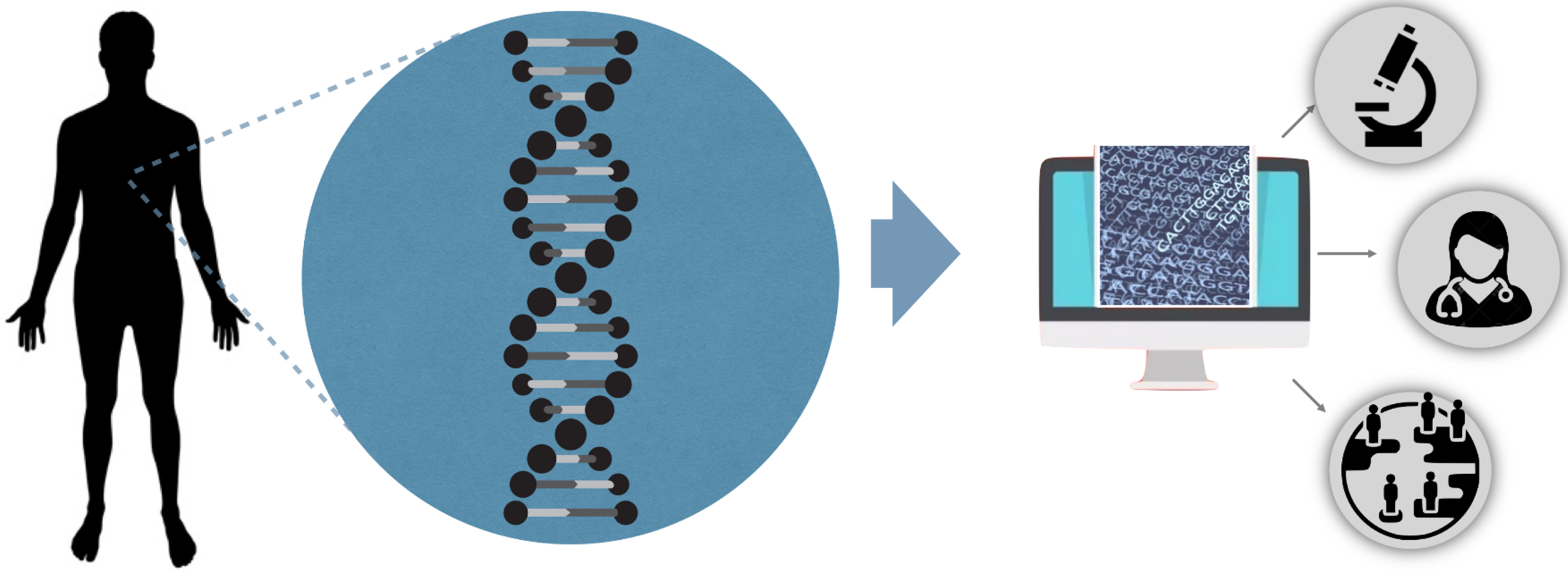




"Tree of Life" (Hug et al., 2016)

The Initial Human Genome Project

Centralized coordinate system: critical for **sharing genomic data and data analysis standards** among international researchers.



How do you use the human reference genome in your work (if at all)?

What do you find most useful or important about the current reference genome?

What concerns, frustrations, or limitations are you aware of regarding the human reference genome?

If you do use the human reference genome, which build is most frequently used?

GRCh37

GRCh38

Both GRCh37 and GRCh38

Neither (or do not use any)

Internal reference(s) to lab/company

Other reference(s)

UPDATE! Data Structure: C- to an **A+**

What is '*The Human Genome Reference*' ?

- live genomes are dynamic and actively changing in our cells
- data representations of genomes collapse complexity to 2D
- current genome representations are linear; misleading
- global *diversity must be integrated* into the reference genome

Data Structure: C- to an A+

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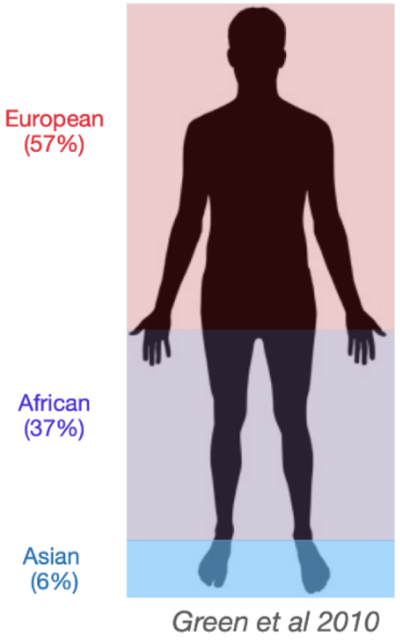
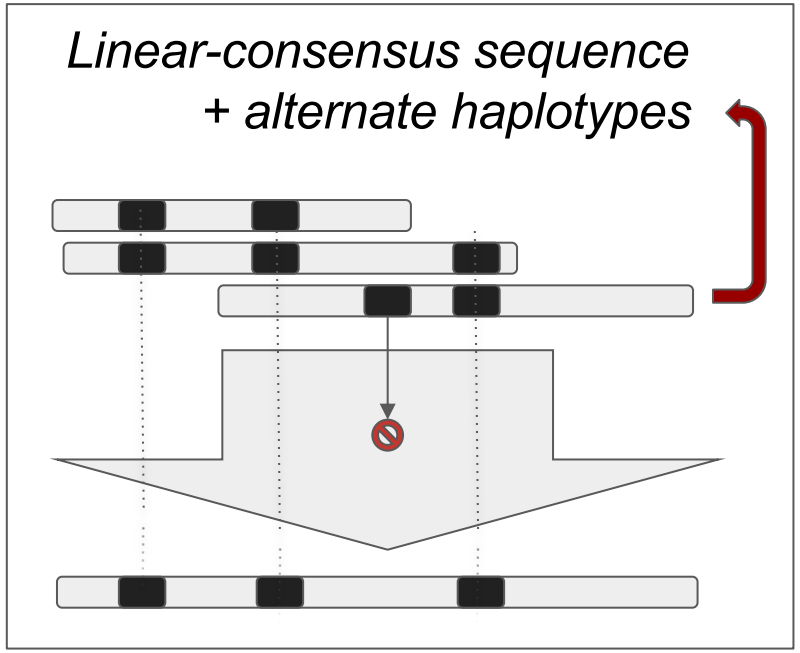


Green et al 2010



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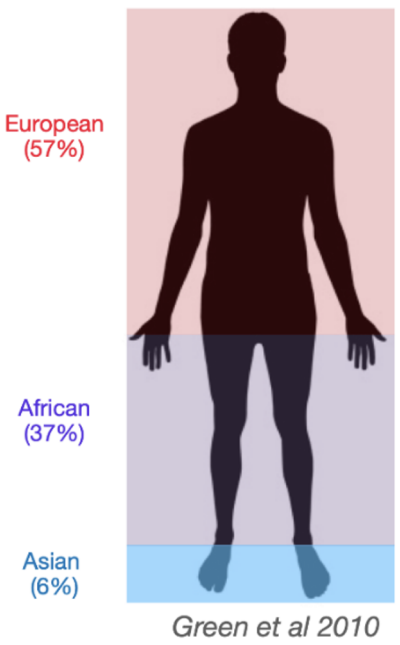

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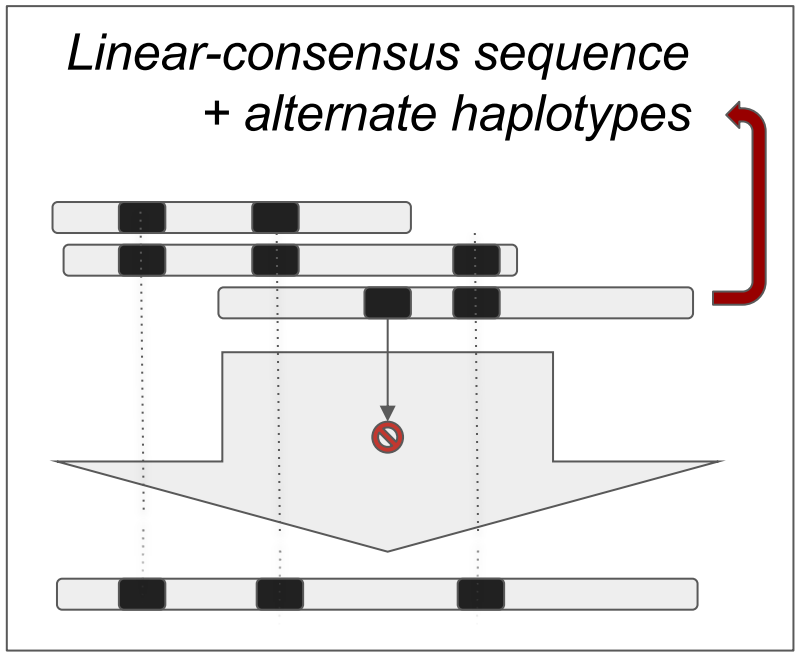
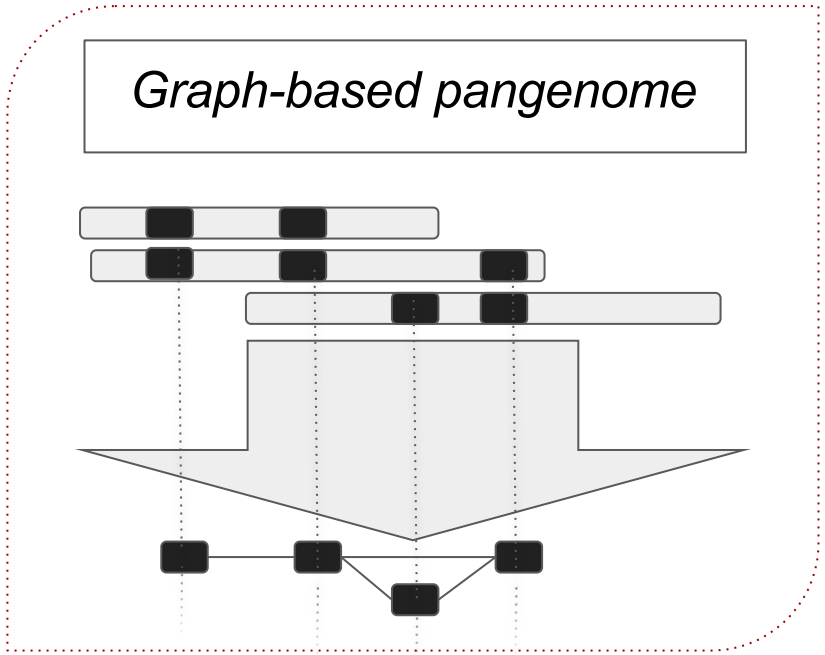
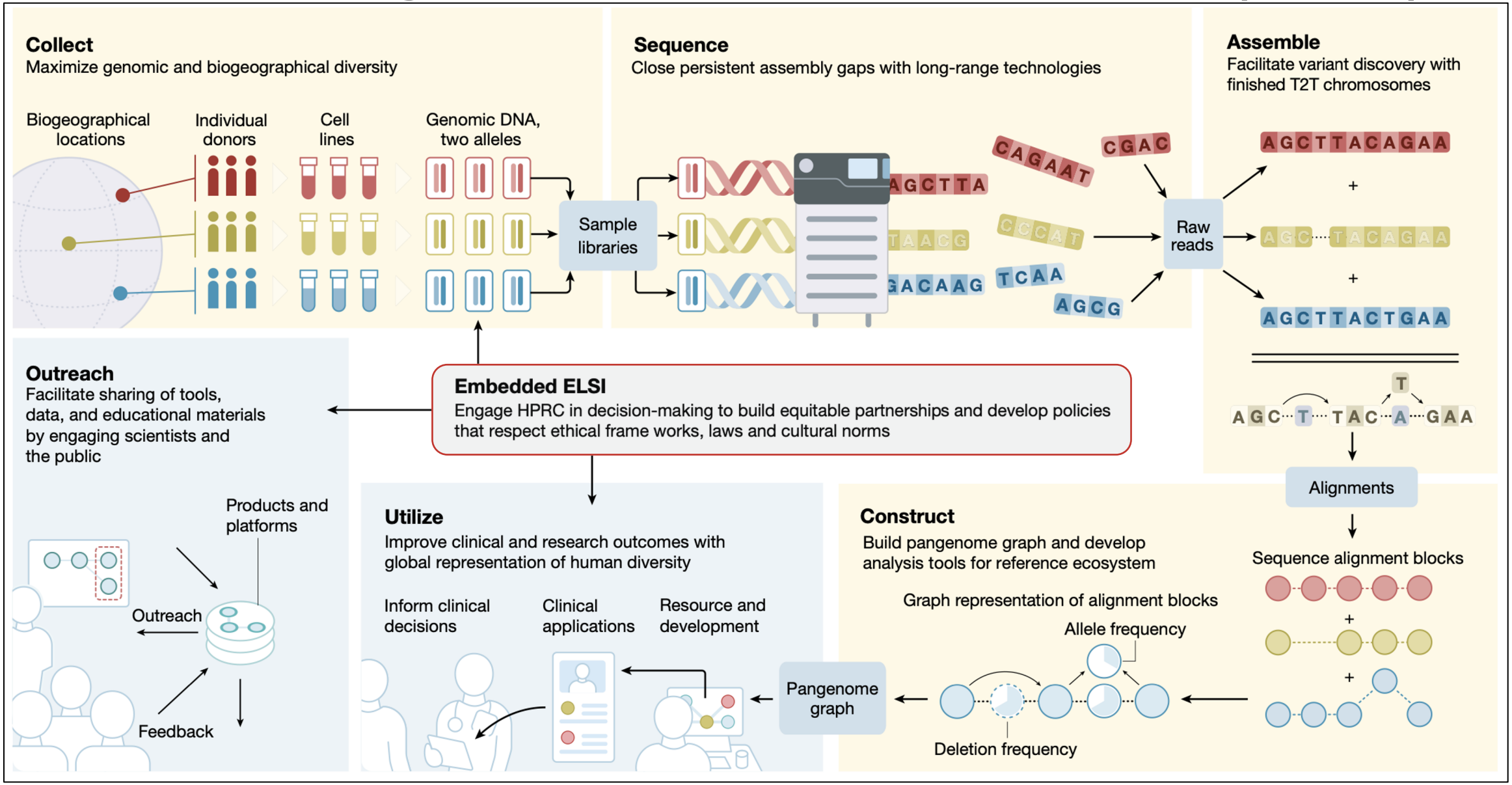




What questions do you have about a pangenome graph? (Please vote for questions that you would like answered or add a unique question.)

Top

Human Pangenome Reference Consortium (HPRC)



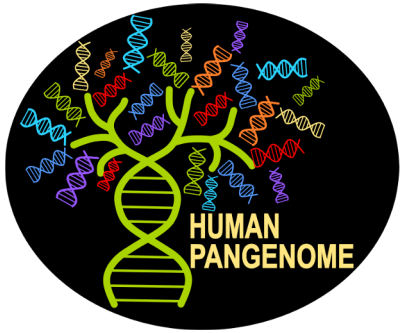
Wang et al., 2022

NIH-OSP Workshop (2021)
Integrating Bioethics and Biomedical Research

*Bioethics engagement over the lifecycle of biomedical research projects can enhance the conduct and **output of research**, ensuring **better science**, increased **inclusion** of participant, stakeholder, and societal values, and engendering greater **trust** in the biomedical research enterprise.*

*Bioethicists, biomedical researchers, institutions, and funders all have an important role to play in building the **relationships and structures** that can enable bioethicists and biomedical researchers to collaboratively advance science.*

"A Match Made in Science" (NIH-Office of Science Policy; July 20, 2021)



Building a Trustworthy Resource for Genomics and Medicine

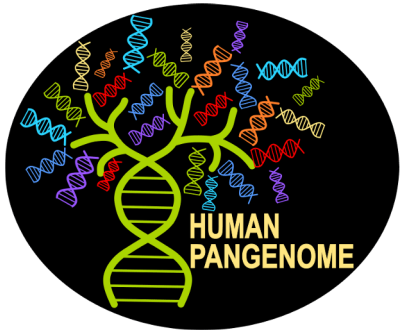
How might we represent global genomic diversity in a graph-based data structure while respecting ethical frameworks and data sovereignty?

Sampling and Representation

- selection criteria for diversity
- contributions to reference variation
- legacy sample reuse and immortal cell line creation

Engagement

- Communication with participants and the public
- clinical providers needs and constraints
- genomics researcher use interoperability
- cultural norms and expectations



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Recruitment
- informed consent for broad further use
- criteria for inclusion and exclusion

Justice
- equitable benefit sharing
- Indigenous data sovereignty
- commercial entities and interests

Collaborations
- international data sharing
- embargos and political implications

Technical – Logistical – Ethical – Social – Cultural – Legal – Regulatory – Commercial

Challenge: Diversity in Genome Reference Data

Known

Genomic variation in existing reference resources

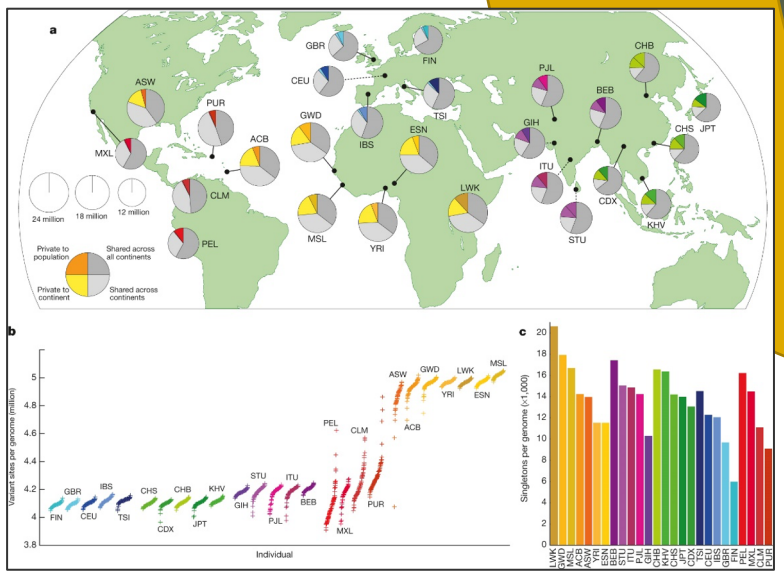
Known Unknown

Geographic regions
Global populations
Effective population size
Continental ancestry proportions
Self-identified group representation

Unknown

Global variation that has yet to be sampled or identified

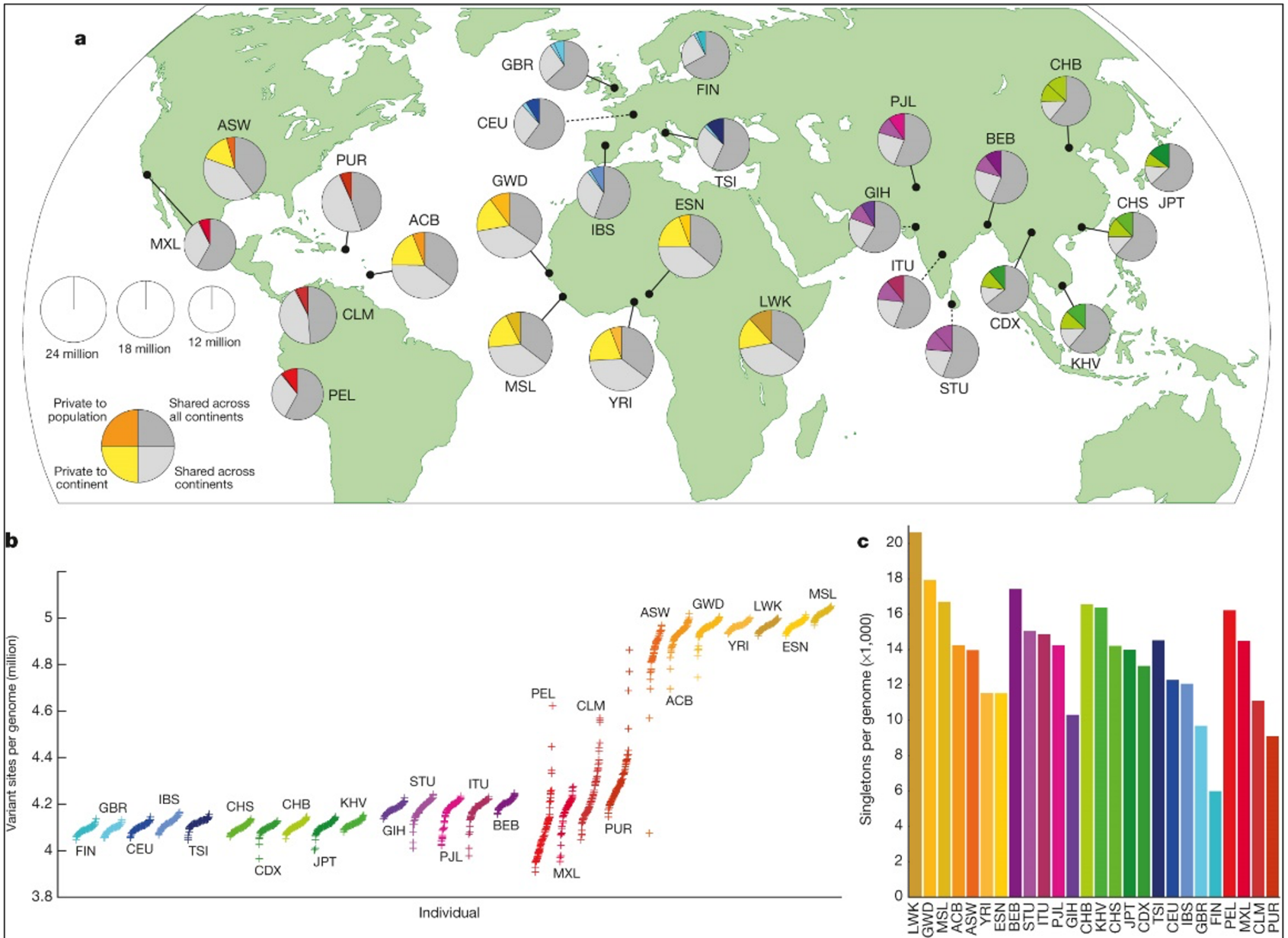
1000 Genomes
Auton *et al*. (2015)



Human Genomic Diversity is Shared (Gray Slices)

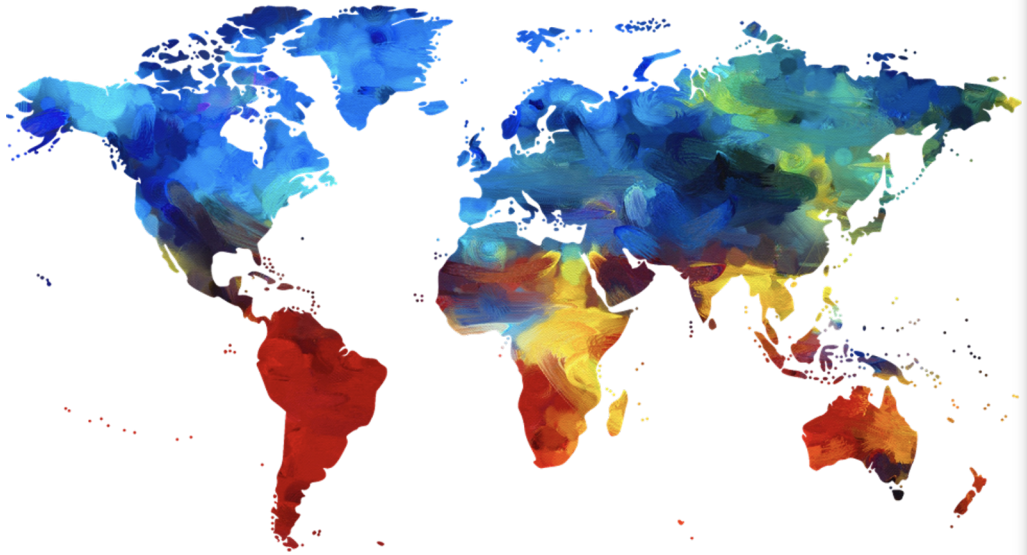
Population
African Caribbean in Barbados **[ACB]**
African Ancestry in SW USA **[ASW]**
Bengali in Bangladesh **[BEB]**
British From England and Scotland **[GBR]**
Chinese Dai in Xishuangbanna, China **[CDX]**
Colombian in Medellín, Colombia **[CLM]**
Esan in Nigeria **[ESN]**
Finnish in Finland **[FIN]**
Gambian in Western Division – Mandinka **[GWD]**
Gujarati Indians in Houston, Texas, USA **[GIH]**
Han Chinese in Beijing, China **[CHB]**
Han Chinese South **[CHS]**
Iberian Populations in Spain **[IBS]**
Indian Telugu in the U.K. **[ITU]**
Japanese in Tokyo, Japan **[JPT]**
Kinh in Ho Chi Minh City, Vietnam **[KHV]**
Luhya in Webuye, Kenya **[LWK]**
Mende in Sierra Leone **[MSL]**
Mexican Ancestry in Los Angeles CA USA **[MXL]**
Peruvian in Lima Peru **[PEL]**
Puerto Rican in Puerto Rico **[PUR]**
Punjabi in Lahore, Pakistan **[PJL]**
Sri Lankan Tamil in the UK **[STU]**
Toscani in Italia **[TSI]**
Yoruba in Ibadan, Nigeria **[YRI]**

Auton *et al*. (2015)



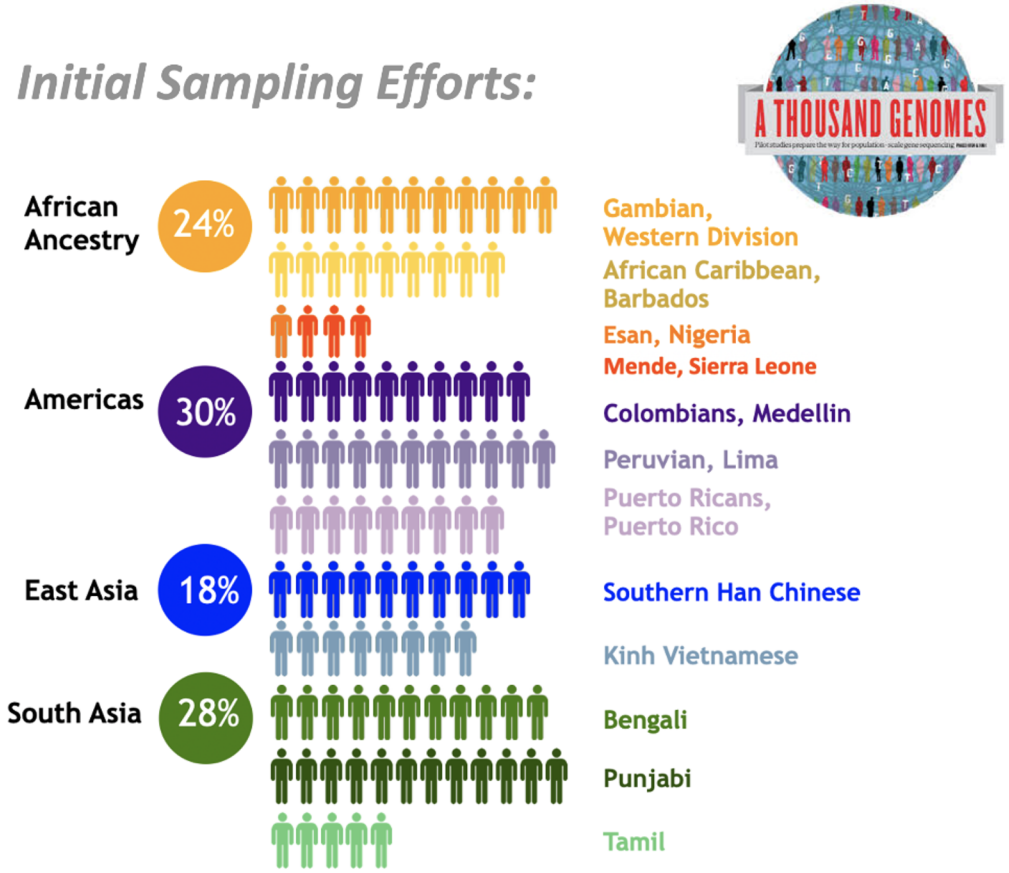
HPRC's Initial Sampling Efforts

Population Representation and Sampling



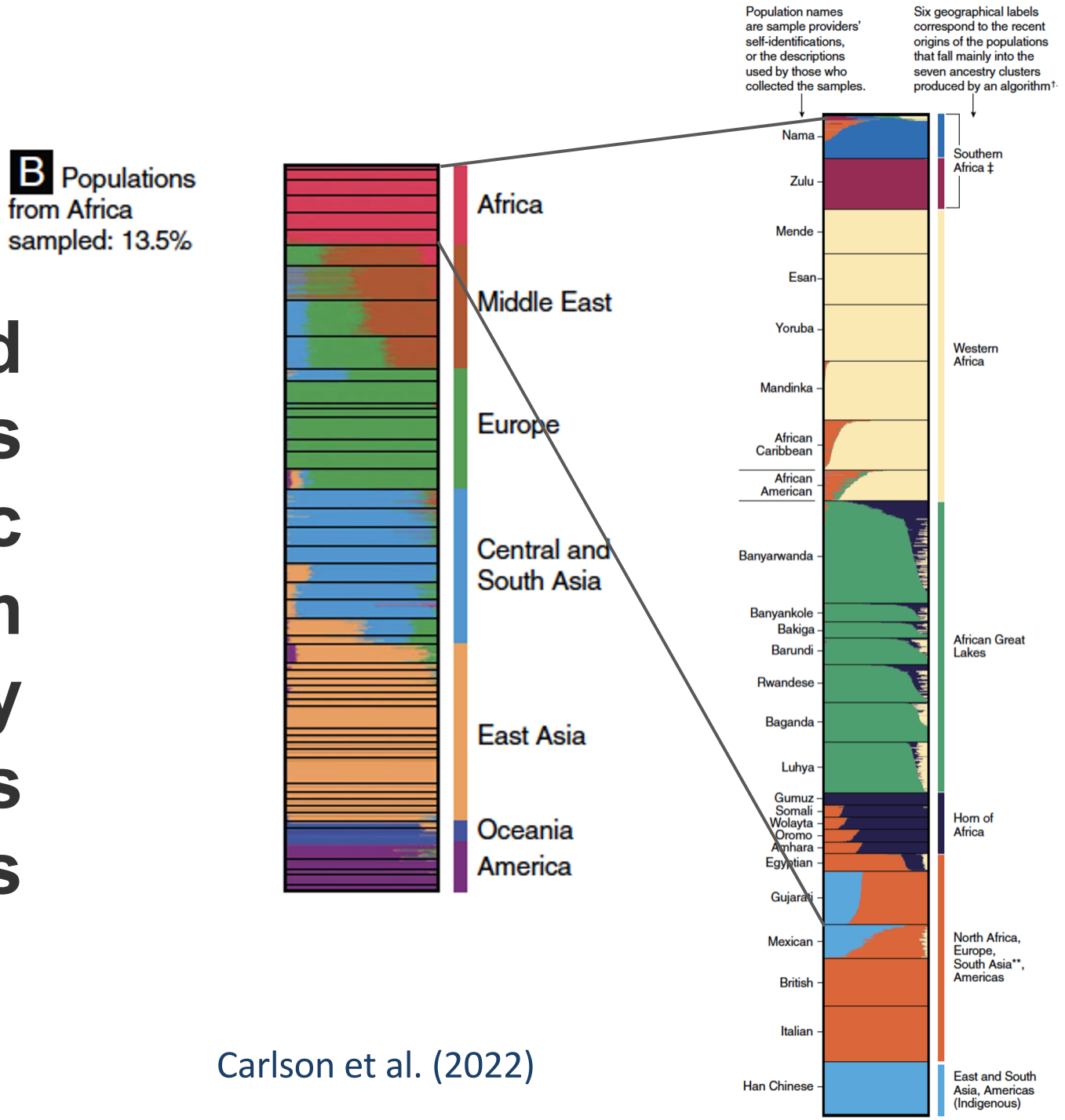
1000 Genomes data alone is insufficient to fully represent genomic diversity within the human population

Initial Sampling Efforts:



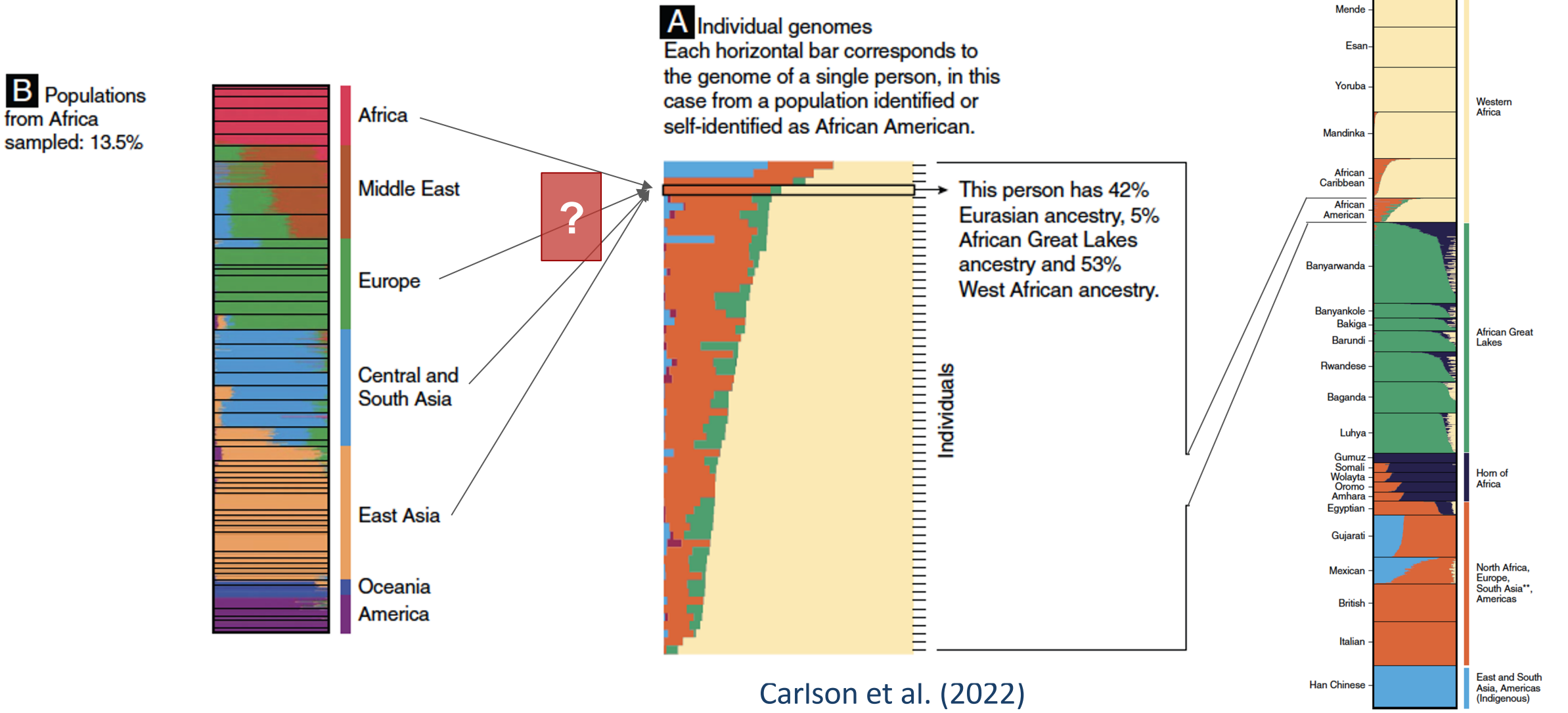
- ☑ Cover genetic and geographic diversity
- ☑ Availability of low passage cell lines
- ☑ Availability of trios/parental data.

Two randomly selected ‘African’ individuals have greater genetic variation between them than two randomly selected individuals from different continents



Carlson et al. (2022)

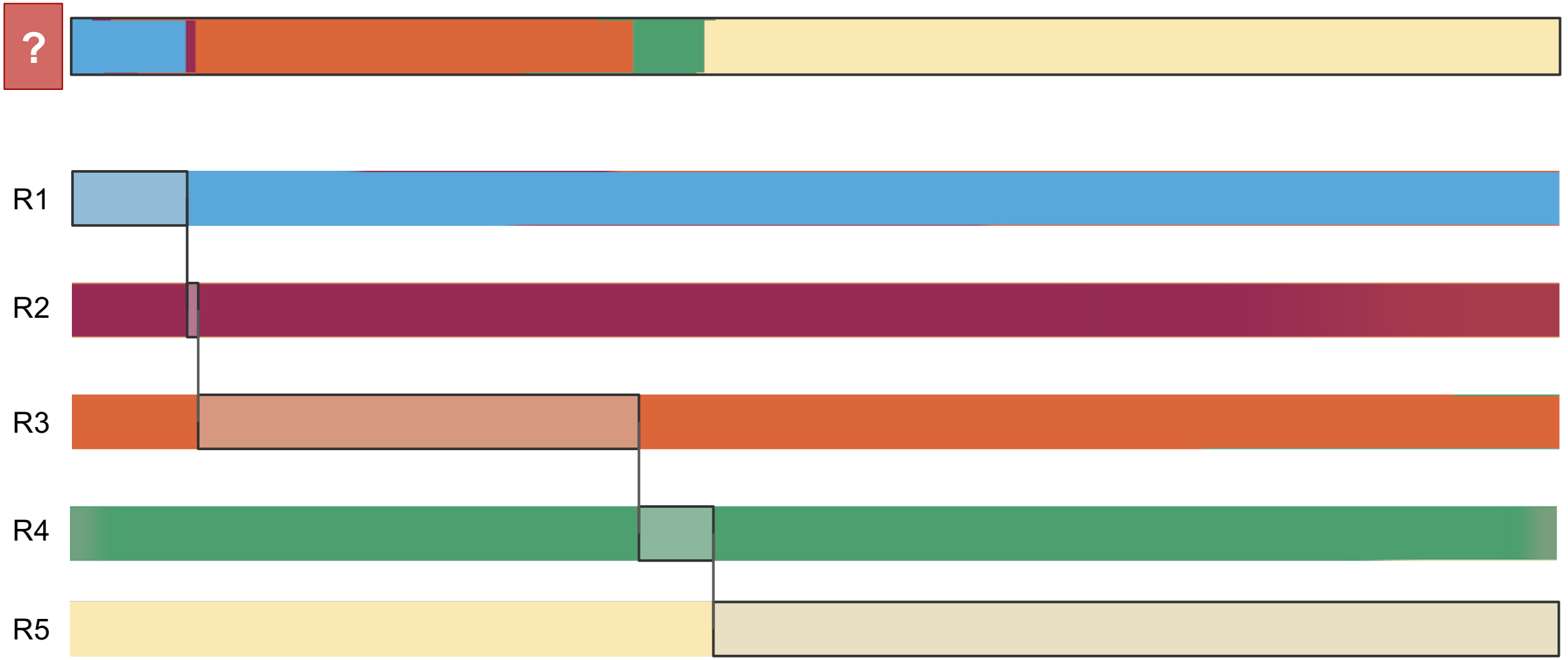
Why use a *pangenome graph* vs. population-specific references?



Carlson et al. (2022)

Why use a *pangenome graph* vs. population-specific references?

“African American” individual in the United States



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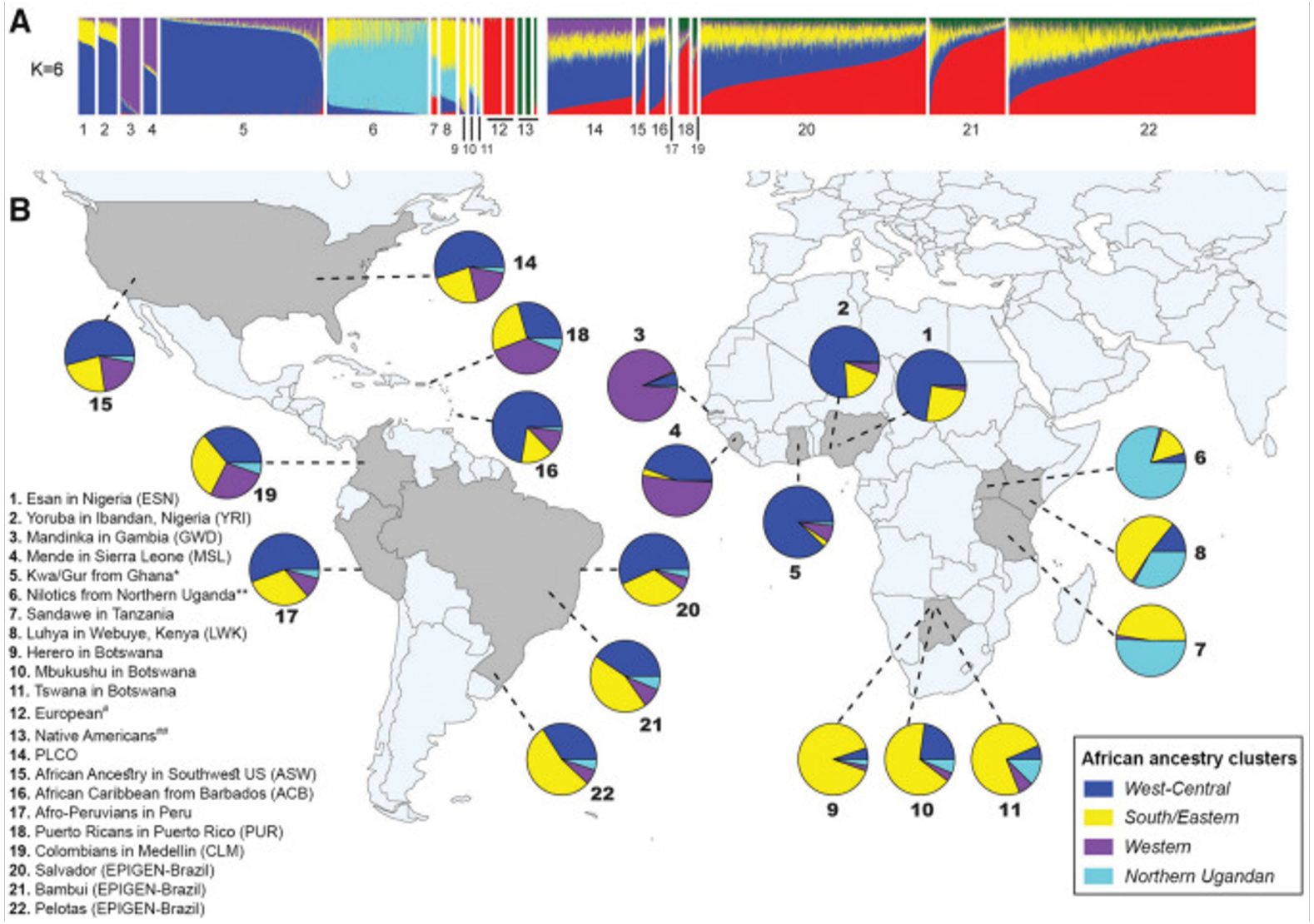
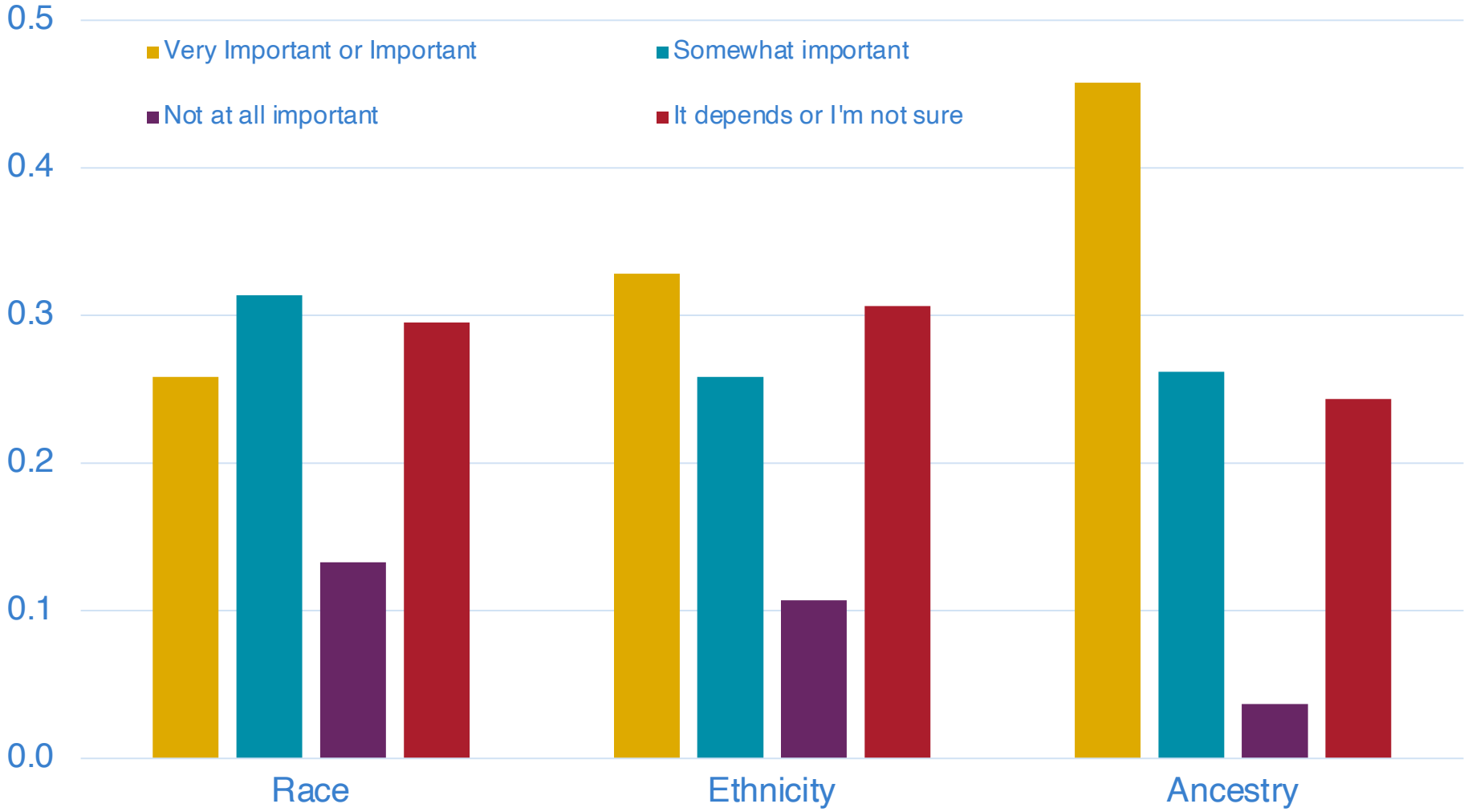


- Genetic ancestries are continuous and relative to reference data (circular logic)
- Most human genomic variation is shared globally across all continents/populations
- Allele frequencies vary gradually across geography and time

Gouveia et al. (2020)

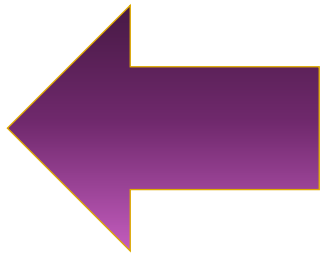
How important are race, ethnicity, and ancestry in the clinical interpretation of genetic variants? (N=271)

Popejoy et al., 2020



If information about a patient's race, ethnicity, or ancestry is used in any aspect of your clinical work, from where are these data obtained? (N=216)

Source of Data	No. Respondents	% Respondents
Race and/or ethnicity information is reported directly to me by the patient	**204**	**94.4%**
Data are obtained or provided from the patient's medical record	87	40.3%
Race and/or ethnicity are recorded by another care provider, possibly without asking the patient directly	**39**	**18.1%**
Genetic ancestry is inferred from the patient's DNA sample	10	4.6%
I do not use any of these measures in my work	5	2.3%
Other (Please specify)	6	2.8%
I'm not sure	4	1.9%



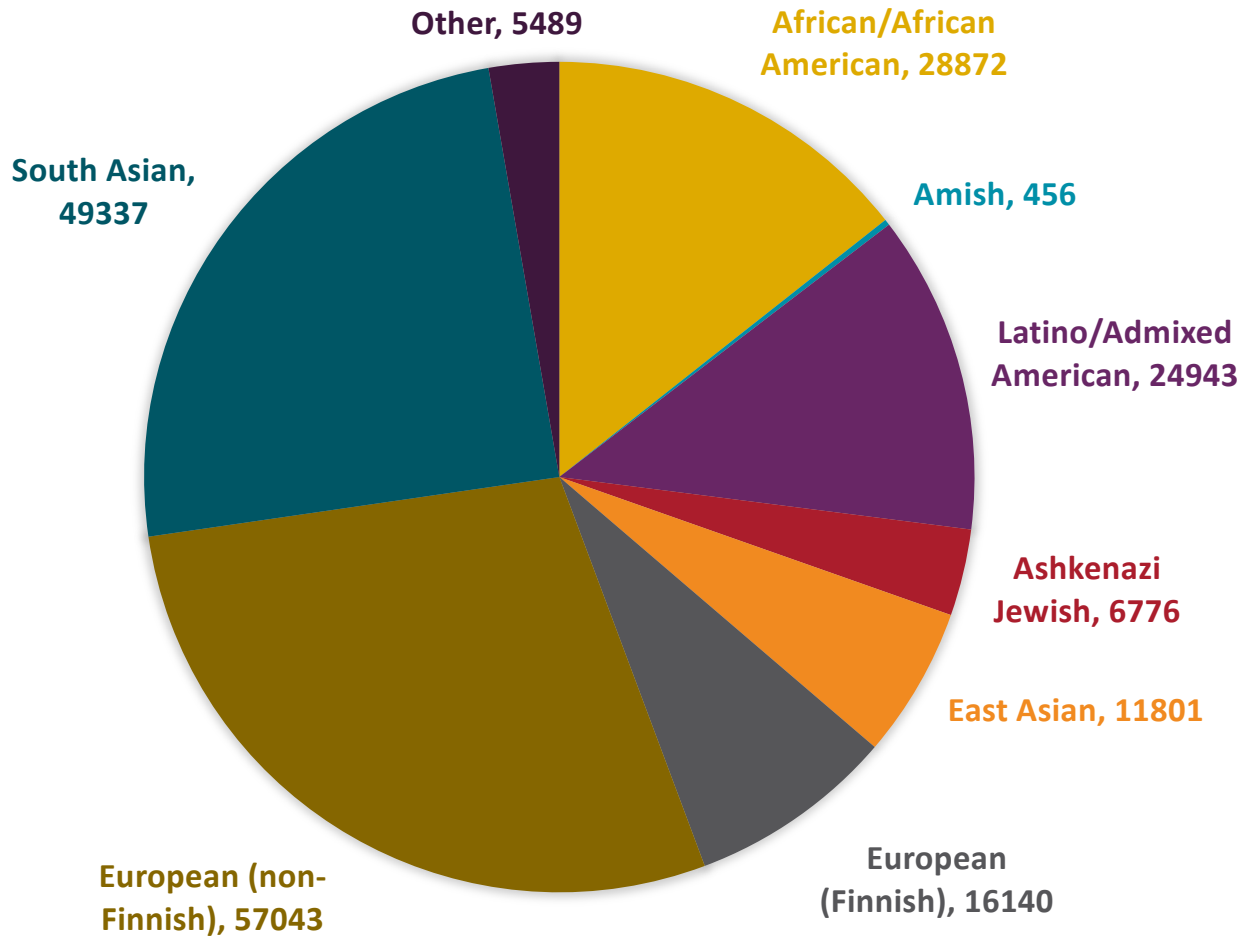
Popejoy et al., 2020

ClinGen
Clinical Genome Resource

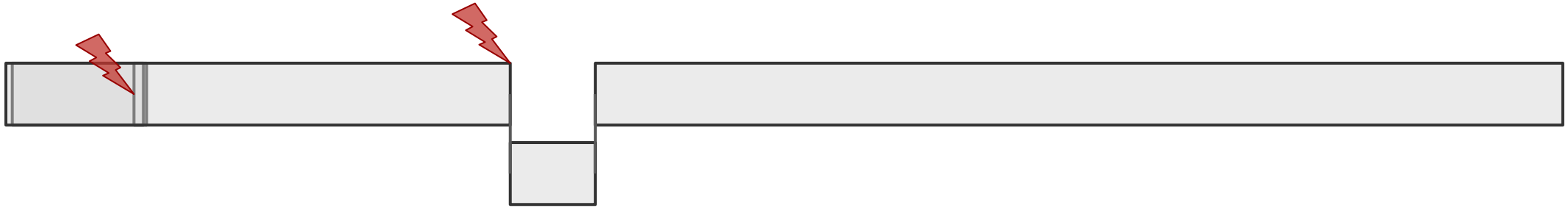
Populations represented in gnomAD allele frequency data

- African/African American
- Amish
- Ashkenazi Jewish
- East Asian
  - Japanese
  - Korean
  - Other East Asian
- European (Finnish)
- European (non-Finnish)
  - Southern European
  - North-western European
  - Other non-Finnish European
  - Bulgarian
  - Swedish
  - Estonian
- Latino/Admixed American
- Other
- South Asian

https://gnomad.broadinstitute.org/



Using The Human Reference [pan]Genome in Clinical Genetics (Discussion)

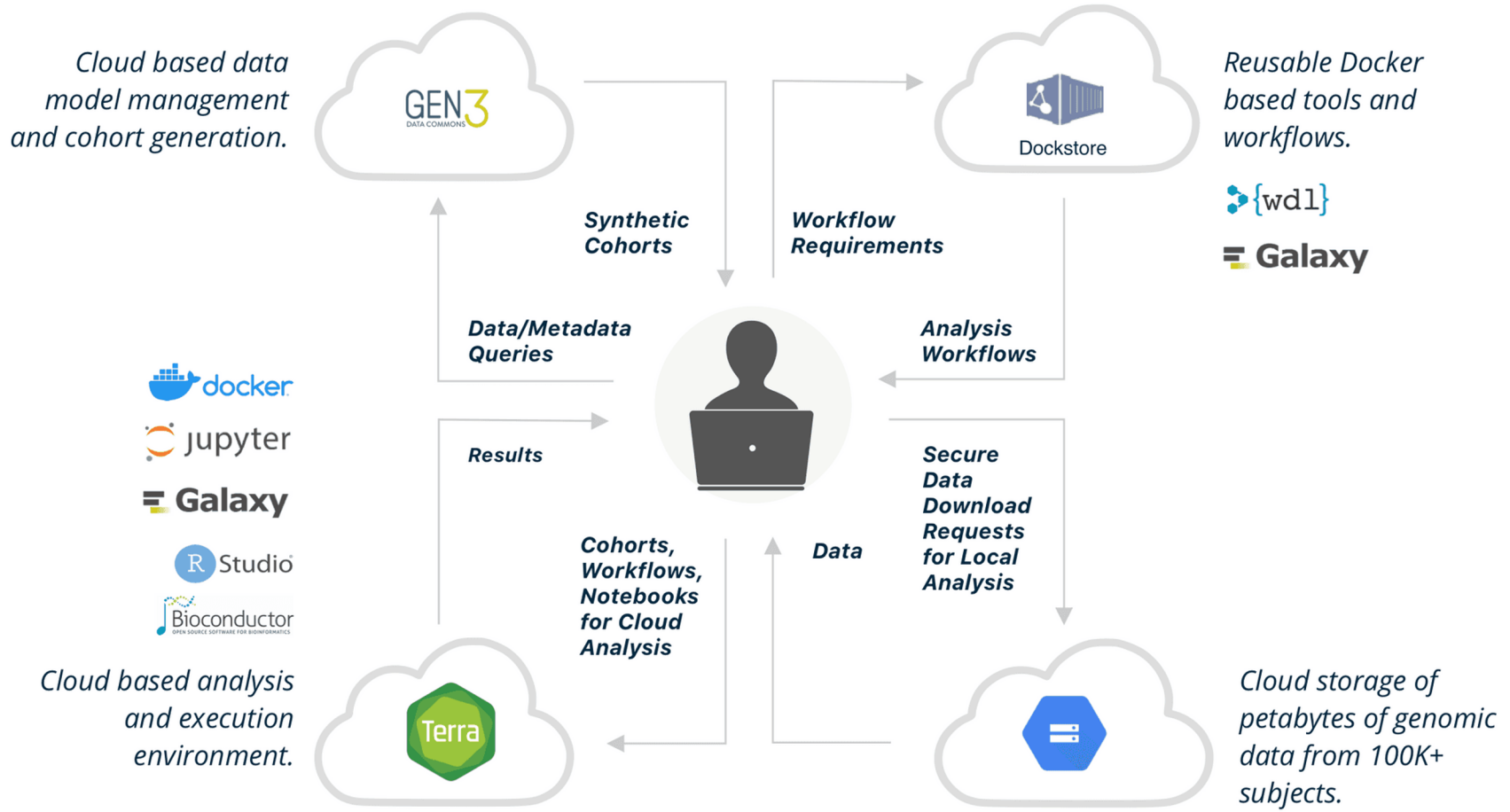


What is exciting or intriguing to you about the development of a graph-based pangenome reference, specifically as it relates to your work in clinical genetics?

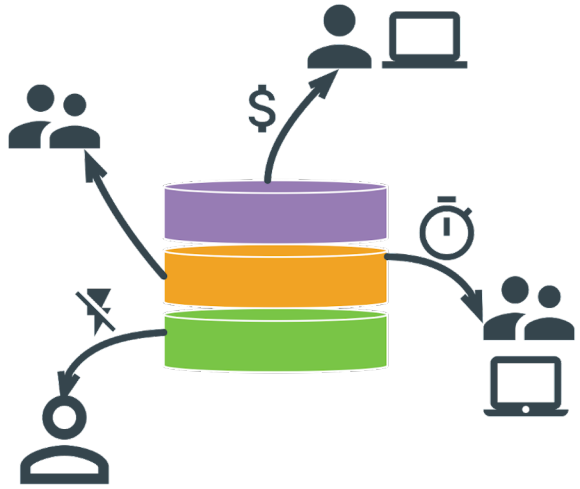
What questions or concerns do you have about the development of a human pangenome reference, and its implications for clinical genetics?

Overview of AnVIL and HPRC Resources

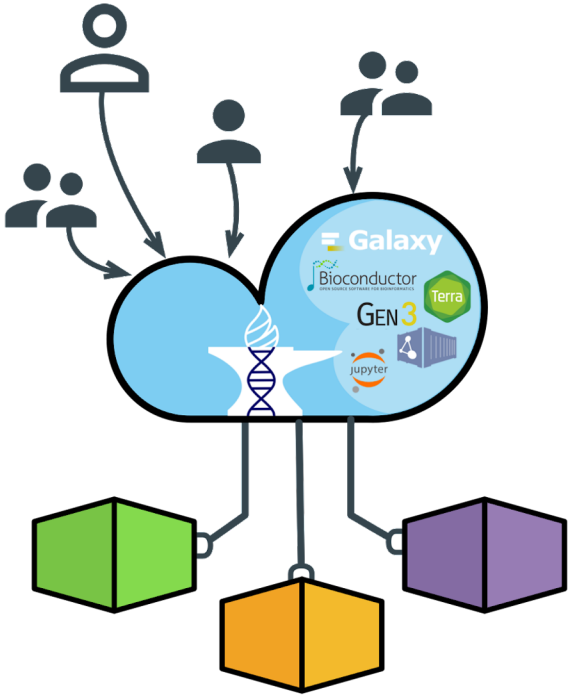
AnVIL: Ecosystem Overview



AnVIL: Inverting The Model Of Genomic Data Sharing



Traditional: Bring data to the researcher

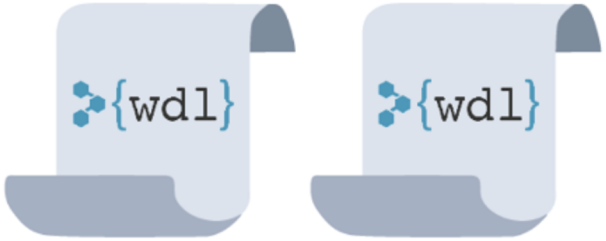


Goal: Bring researcher to the data

Schatz, Philippakis et al. (2022) *Cell Genomics*. https://doi.org/10.1016/j.xgen.2021.100085

Terra: Cloud Data Storage & Processing

Workflows

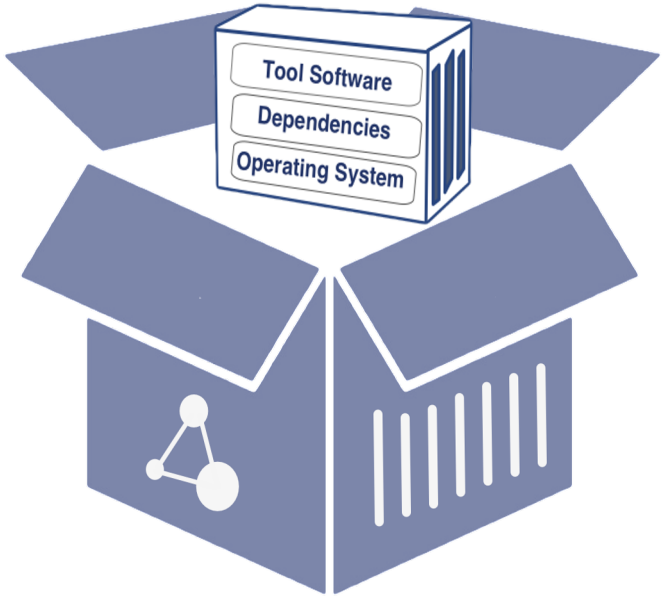


Data Tables

Analyses

Dockstore: AnVIL's Repository For Workflows

Galaxy {wdl}



AnVIL

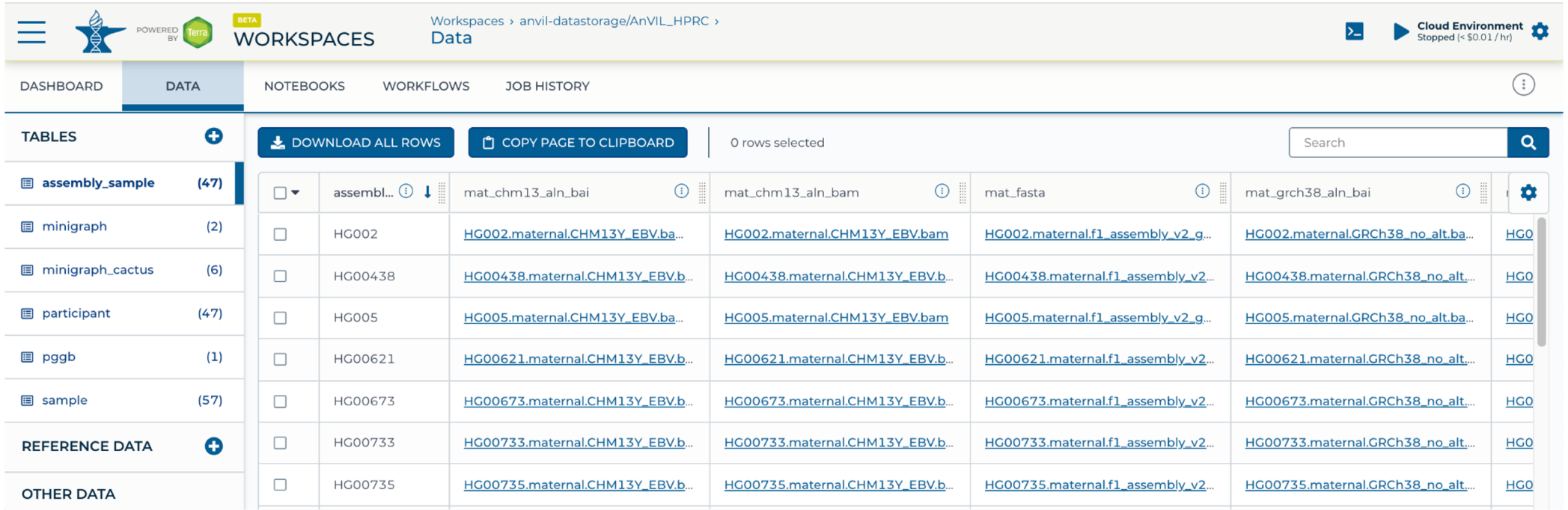
Terra

Launch with

dockstore.org

AnVIL Hosts HPRC Data

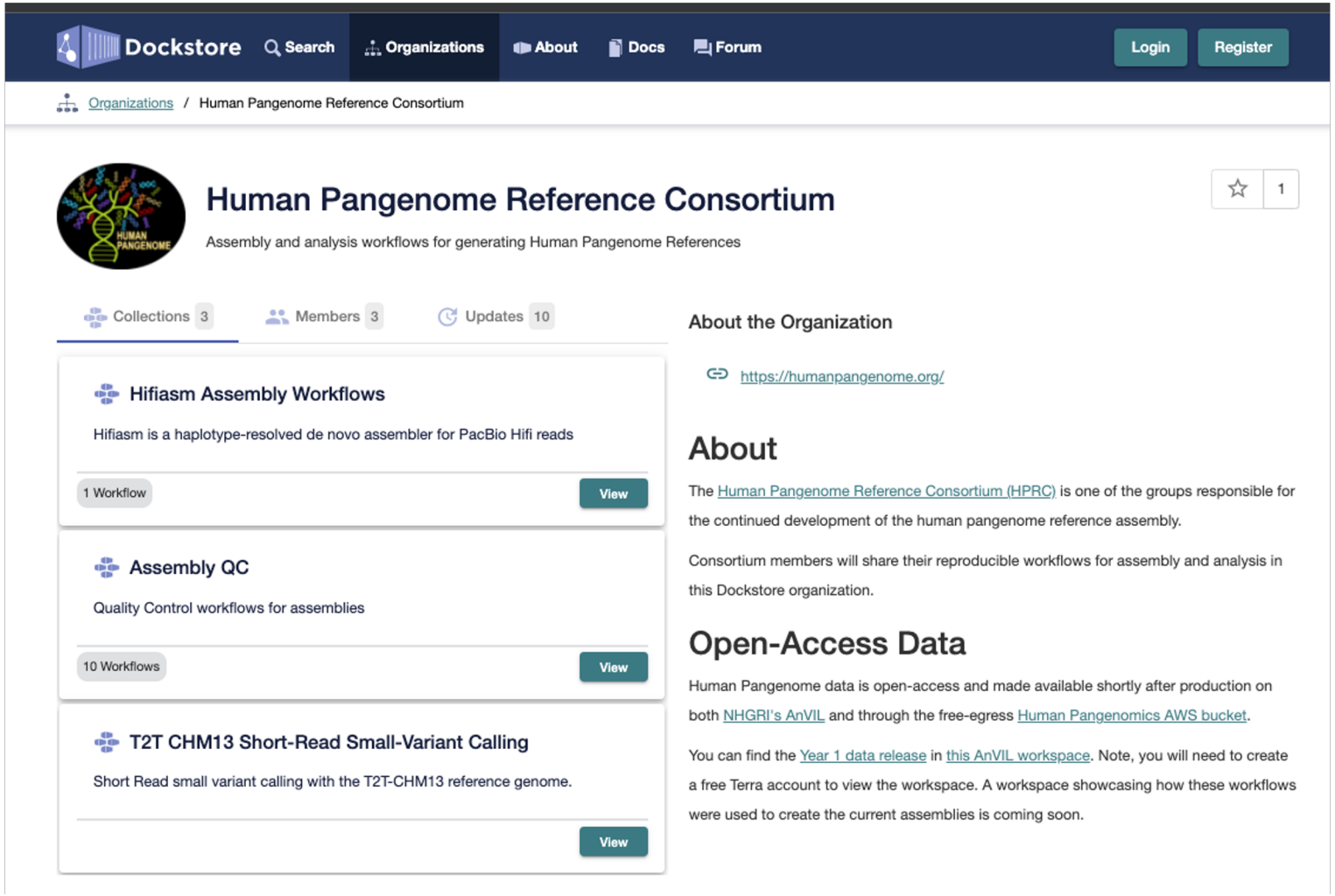
- Community can directly access and analyze data in the HPRC data workspace
- When you work in AnVIL or GCP, you don't pay to use the data (no need to copy or host individually)



https://anvil.terra.bio/#workspaces/anvil-datastorage/AnVIL_HPRC

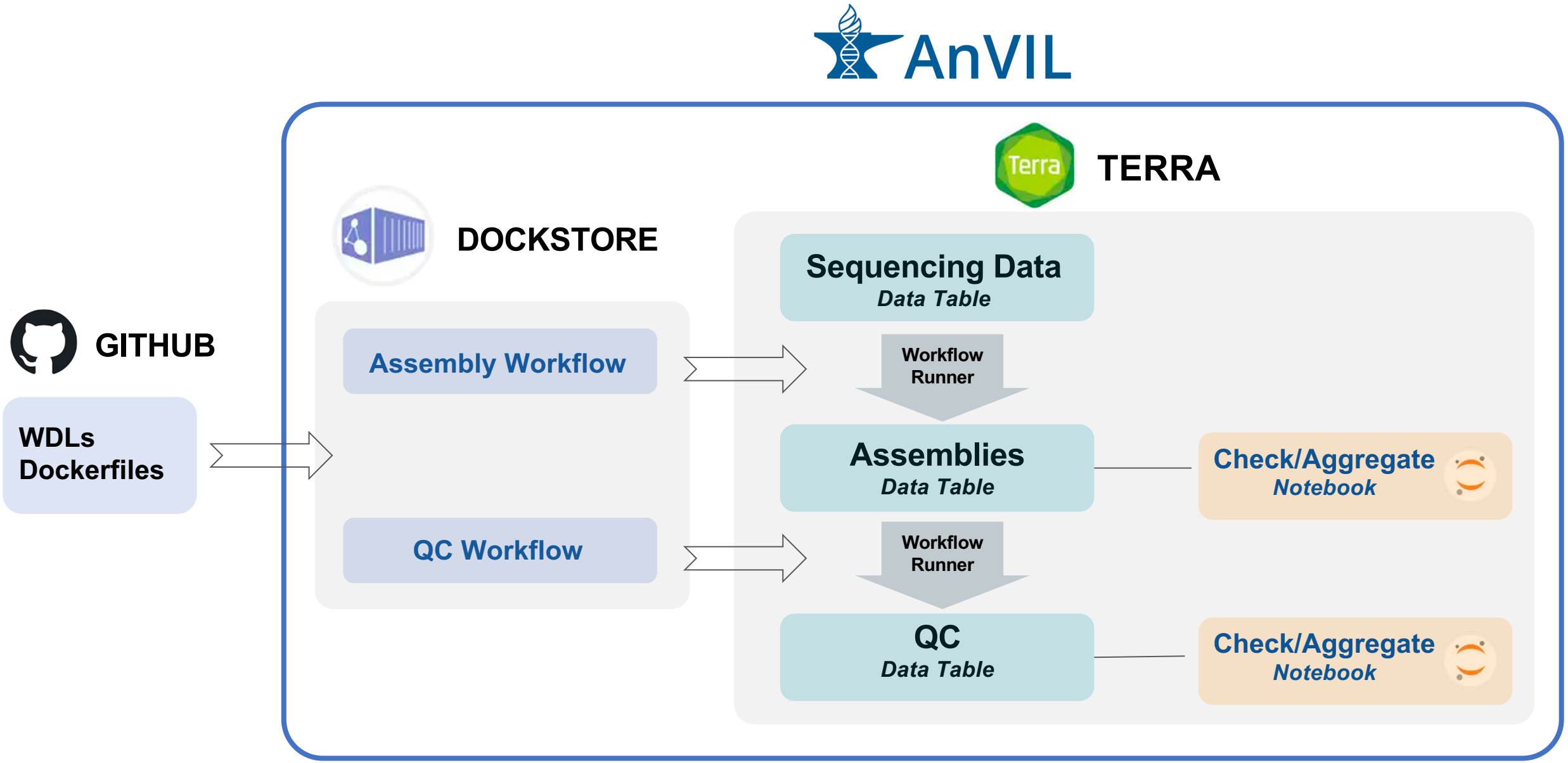
HPRC Shares Workflows In Dockstore

- Consortia share production workflows with the community using GitHub and Dockstore
- Leverage a container-based workflow language (WDL)
- Easily launch from Dockstore to Terra



https://dockstore.org/organizations/HumanPangenome

HPRC Created Year 1 Assemblies In AnVIL



HPRC's Data Flow

Dovetail GENOMICS

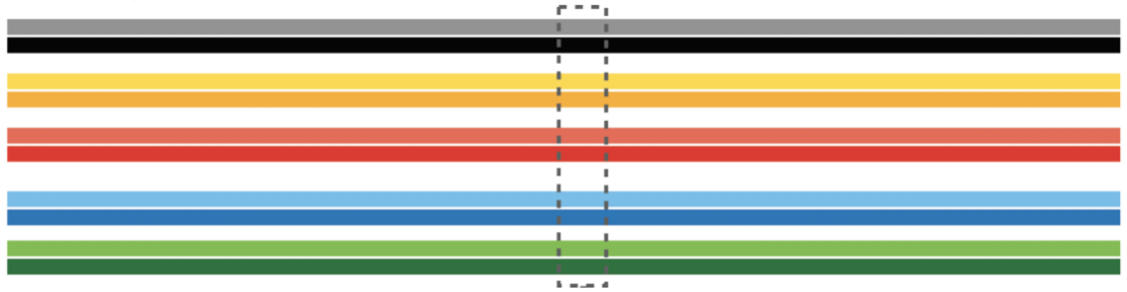
illumina

Oxford NANOPORE Technologies

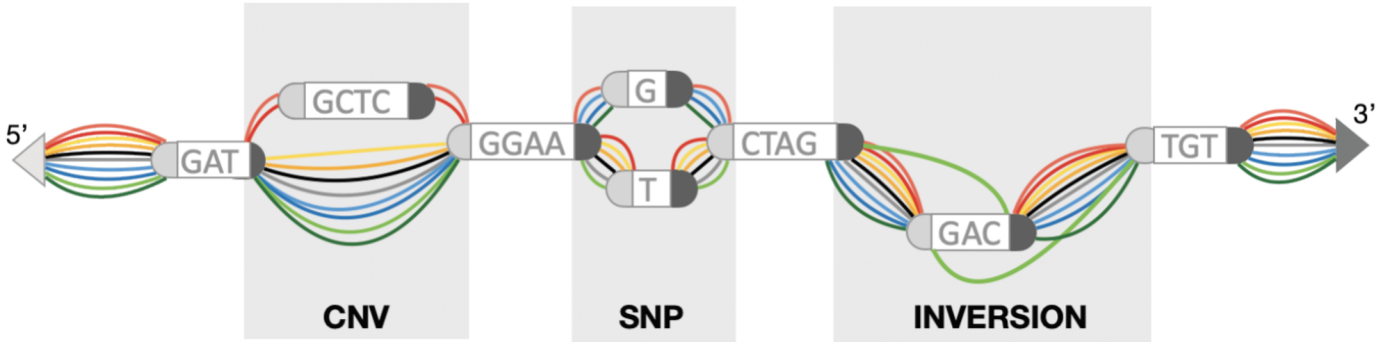
PACBIO®

Release → **Data/ Workflows**

Haplotype-Phased Assemblies:



Release → **Data/ Workflows**



Release → **Data/ Workflows**

HPRC Sequencing Data

The AnVIL

Public workspace (requires AnVIL account)

GitHub

Repository with data indexes for GCP and AWS files

Sequence Read Archive

BioProject with INSDC copies of raw sequencing data

HPRC Assemblies

The AnVIL

Public workspace (requires AnVIL account)

GitHub

Assembly repository with indexes for AWS & GCP files

Genbank

BioProject for HPRC assemblies

UCSC Genome Browser

Browser hub with year 1 assemblies & annotations

ENSEMBL

Project page containing assemblies & gene annotations

HPRC Pangenomes

The AnVIL

Public workspace (requires AnVIL account)

GitHub

Repository with data indexes for GCP and AWS files

European Nucleotide Archive

BioProject with pangenomes

First Data Release

sequencing data & QC metrics for the first 30 samples*

30 HiFi (30x, 17-20kb)	30 ONT Ultra-Long (~6x 100 kb+)	30 Hi-C (Omni-C, ~60X)
30 Bionano Maps (N50>250kb, ~100X coverage)	60 Parental Datasets (30x, 150 bp PE)	10 Strand-Seq single-cell libraries

Open Data Sharing and Cloud-based Data Management

amazon web services
S3://human-pangenomics

AnVIL
AnVIL_HPRC

Dockstore
Human Pangenome

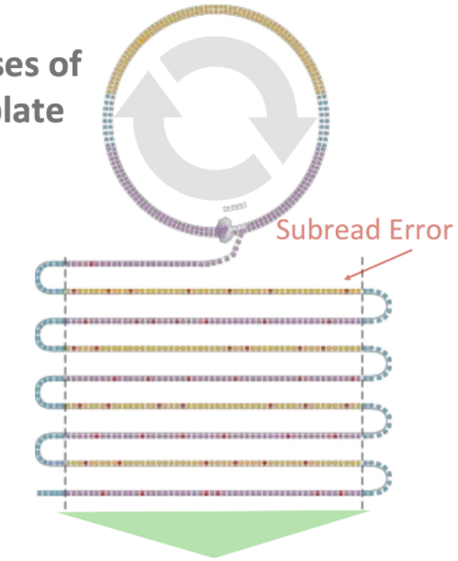
GitHub
Human-Pangenomics

Data Production Has An Emphasis On Long Reads

Advances in Long-Read Sequencing

PacBio High Fidelity (HiFi) Data

Multiple passes of a single template molecule

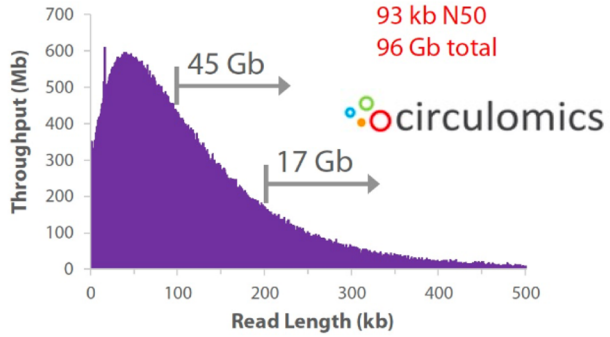


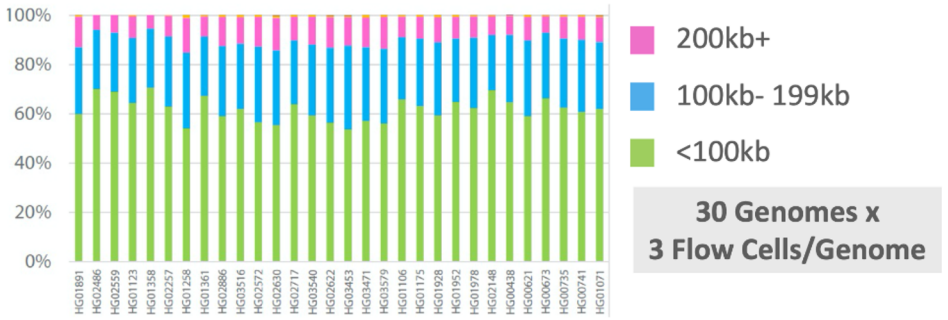
High-Quality Circular Consensus ("CCS") Read

99.9% Consensus Read Accuracy
(35-40x Coverage >Q20 HiFi Reads; 18-20kb)

Accurate circular consensus long-read sequencing improves variant detection and assembly of a human genome. Wenger et al. *Nature Biotechnology* (2019)

ONT Ultra-Long (UL) Data





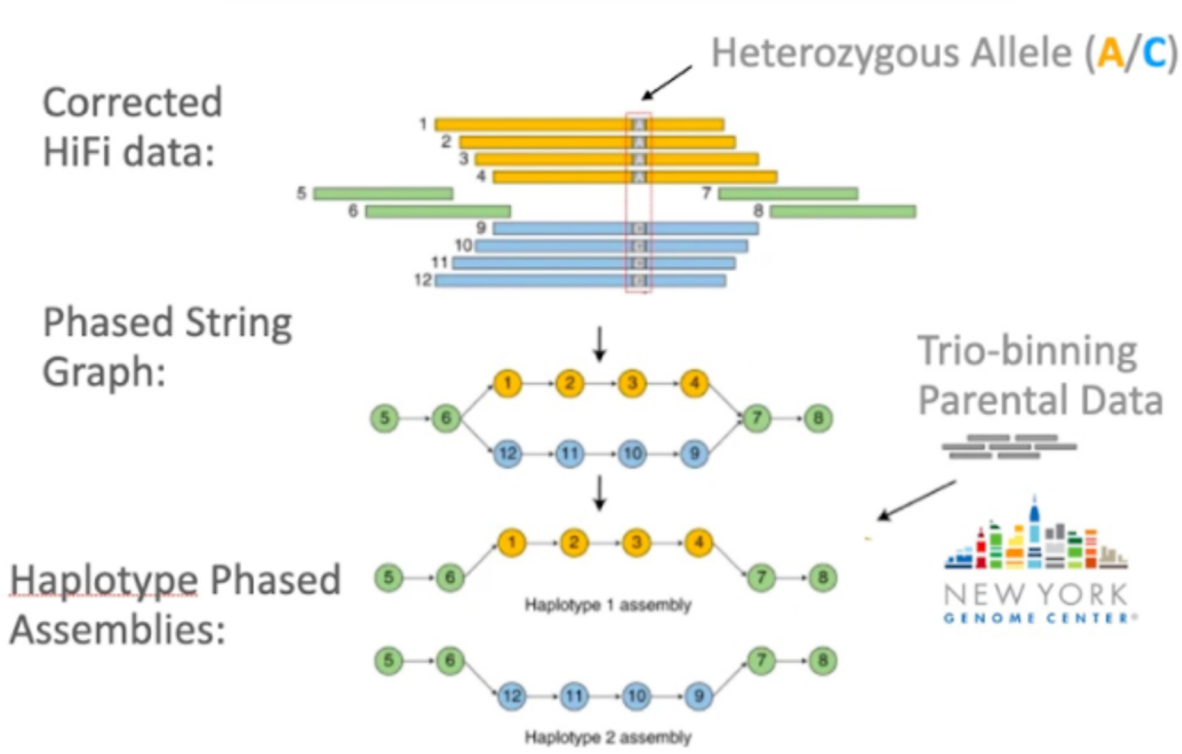
30 Genomes x
3 Flow Cells/Genome

Nanopore sequencing and assembly of a human genome with ultra-long reads. Jain et al. *Nature Biotechnology* (2018)

Assembly Strategy

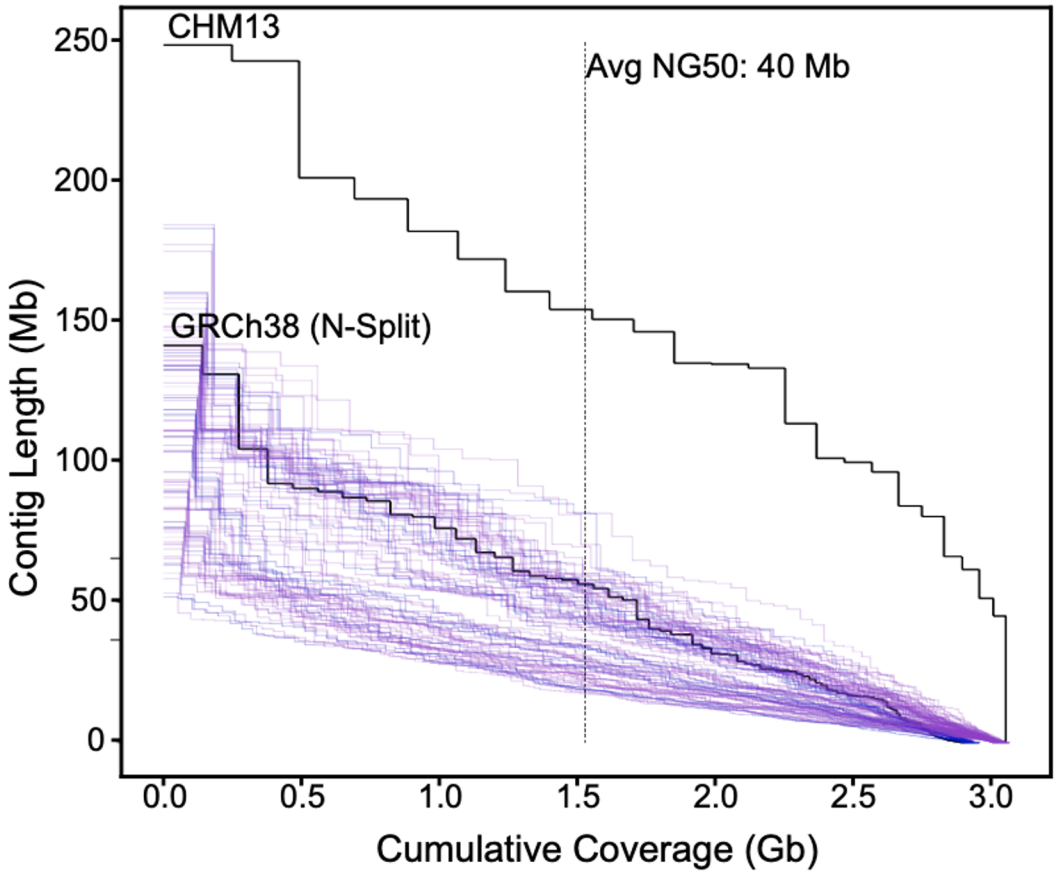
- **Needed a high-quality, automated workflow to create contig-level diploid assemblies**
- **Held a bake-off**
  - *Jarvis, et al. Nature 2022*
- **Developing methods for automated QC, scaffolding, T2T, & non-trio assembly**

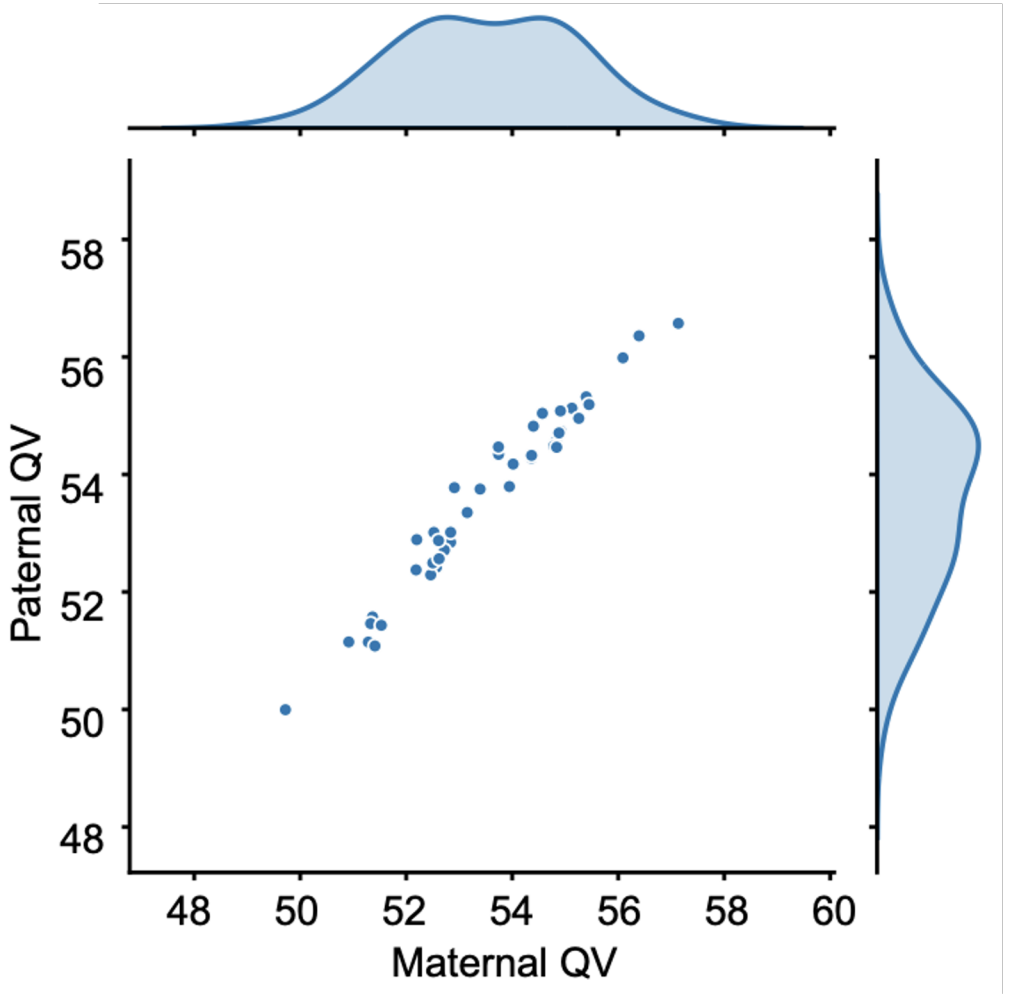
Hifiasm



Cheng, Haoyu, et al. *Nature Methods* (2021)

High-Quality Diploid Assemblies





Liao, Asri, Ebler, et al. *BioArxiv* (2022)

How To Use Pangenomes To Improve (Clinical) Variant Calling

Pangenome Creation From Assemblies

```
TCGGACTTGCGACCAGCAATGGAGATGCTCAGCGTTAGATACGCCGAATTAATGGACAA haplotype 1

TCGGACTTGCGACCAGCAATGGAGATGCTCAGTGTTAGATACGCCGAATTAATGGACAA haplotype 2

TCGGACTTGCGACCAGCAATGGA------------------------TAATGGACAA haplotype 3
```

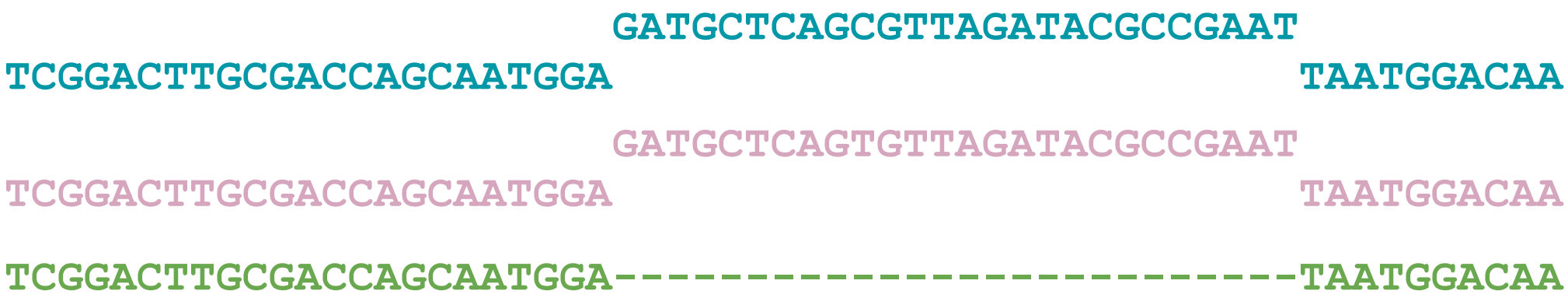
Pangenome Creation From Assemblies

```
TCGGACTTGCGACCAGCAATGGAGATGCTCAGCGTTAGATACGCCGAATTAATGGACAA

TCGGACTTGCGACCAGCAATGGAGATGCTCAGTGTTAGATACGCCGAATTAATGGACAA

TCGGACTTGCGACCAGCAATGGA-----------------------TAATGGACAA
```

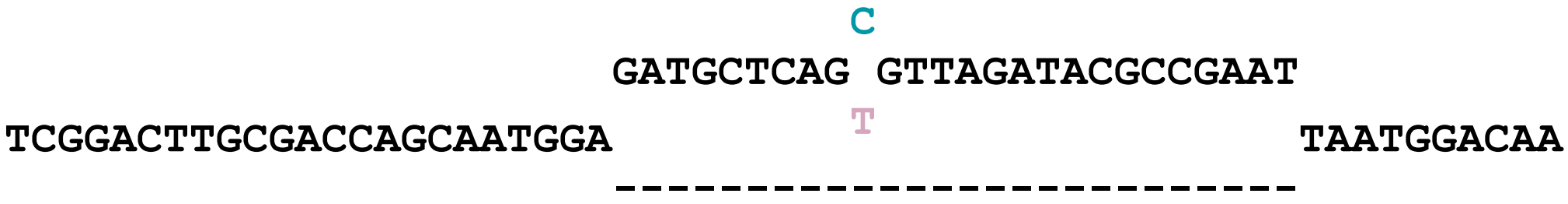
Pangenome Creation From Assemblies



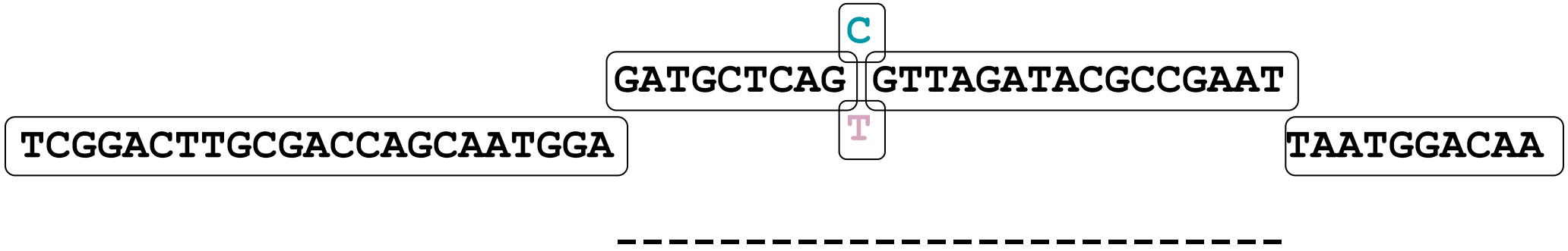
Pangenome Creation From Assemblies

Pangenome Creation From Assemblies

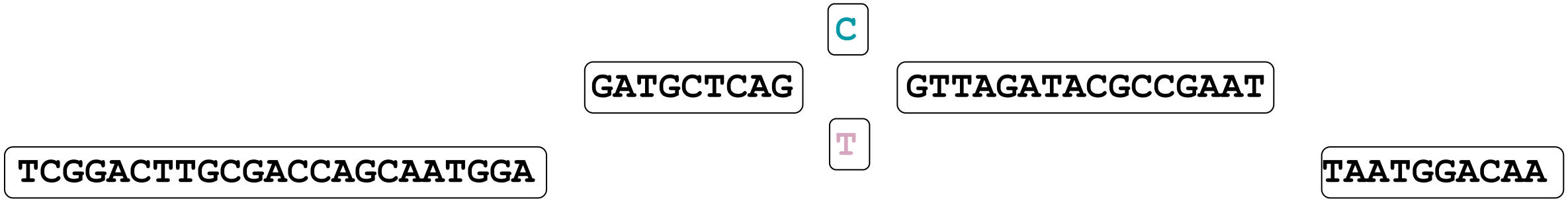
Pangenome Creation From Assemblies



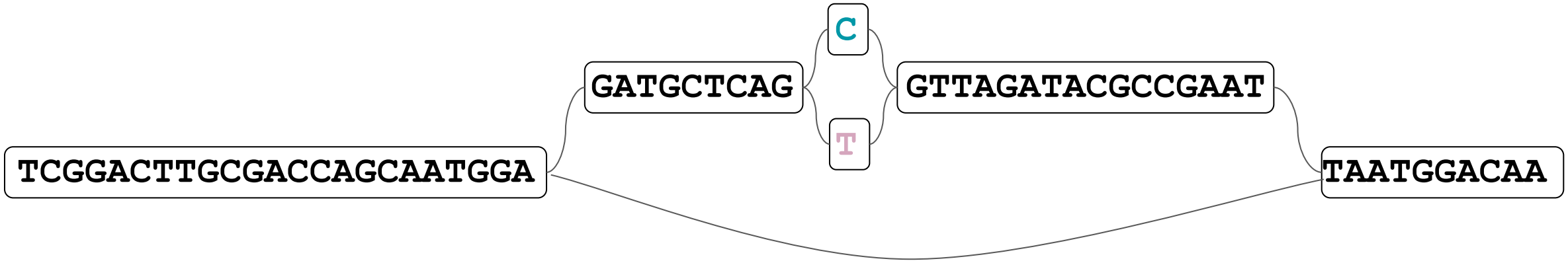
Pangenome Creation From Assemblies



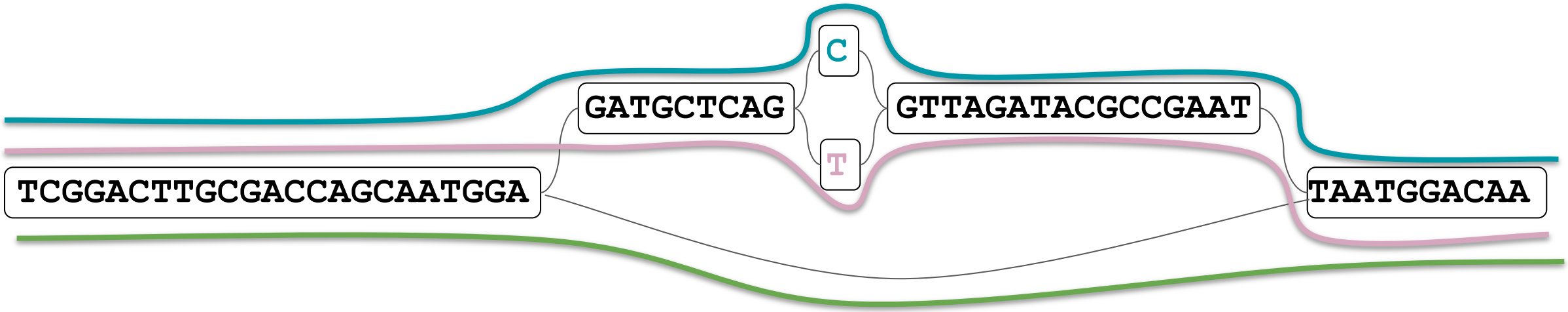
Pangenome Creation From Assemblies



Pangenome Creation From Assemblies



Pangenome Creation From Assemblies



Pangenome: Conceptual Approach To Creation

Pangenome: Three Approaches

- Pangenome team currently has “Freeze1” pangenomes
  - The best pangenomes we can create now with current data and tools
  - 90 haplotypes were included (includes GRCh38 and CHM13)

	Minigraph	**Minigraph/CACTUS**	**PGGB**
Short Read Mapping	Untested	**Yes (fast)**	Untested
Long Read Mapping	Yes (fastest)	Yes	Yes
Assembly Mapping	Yes (direct)	Untested	Yes (via injection)

Pangenome: Three Approaches

- **Generalization of minimap2**
- **Iterative construction**
- **SV only**

	Minigraph	**Minigraph/CACTUS**	**PGGB**
Short Read Mapping	Untested	**Yes (fast)**	Untested
Long Read Mapping	Yes (fastest)	Yes	Yes
Assembly Mapping	Yes (direct)	Untested	Yes (via injection)

Pangenome: Three Approaches

- **Adds base-level alignments to minigraph**
- **Omits centromeric variation**

	Minigraph	**Minigraph/CACTUS**	**PGGB**
Short Read Mapping	Untested	**Yes (fast)**	Untested
Long Read Mapping	Yes (fastest)	Yes	Yes
Assembly Mapping	Yes (direct)	Untested	Yes (via injection)

Pangenome: Three Approaches

- **Constructed with all-to-all pairwise mapping**
- **Contains centromeric variation**

	Minigraph	**Minigraph/CACTUS**	**PGGB**
Short Read Mapping	Untested	**Yes (fast)**	Untested
Long Read Mapping	Yes (fastest)	Yes	Yes
Assembly Mapping	Yes (direct)	Untested	Yes (via injection)

Pangenome: How You Can Use A Pangenome

1

Calling SNV/InDels From Short Reads
VG-Giraffe

2

Short RNA Mapping
VG

3

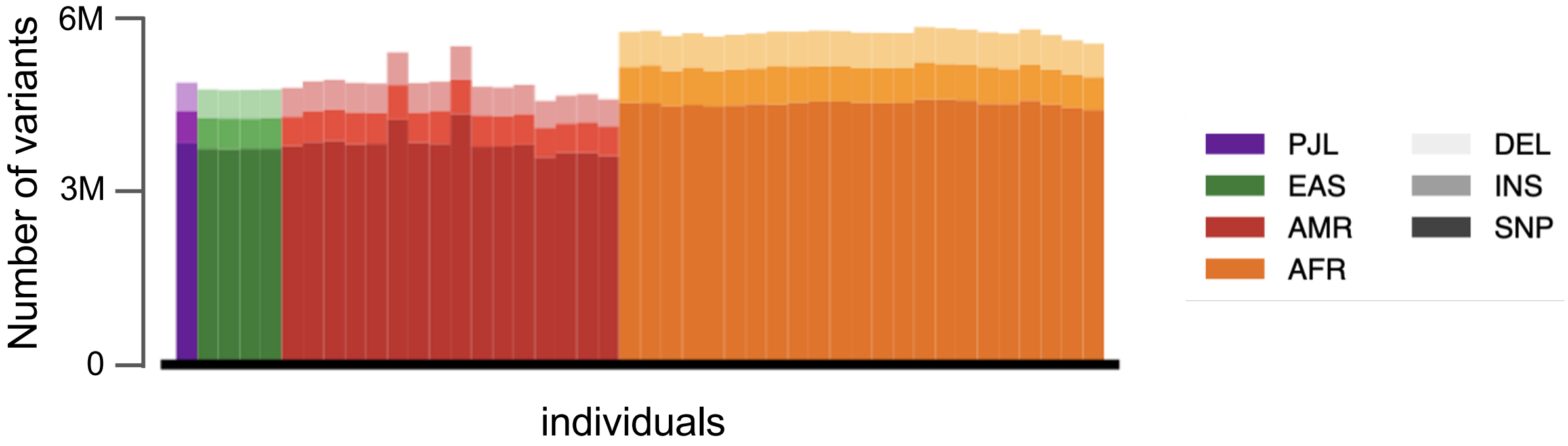
Comparative Genomics
Create browser hub (MC)
Liftover with Comparative Analysis Toolkit

4

Calling Structural Variants From Short Reads
Pangenie

Liao, Asri, Ebler, et al. ***BioArxiv*** **(2022)**

Pangenome: How You Can Use A Pangenome



Credit: Wen-Wei Liao

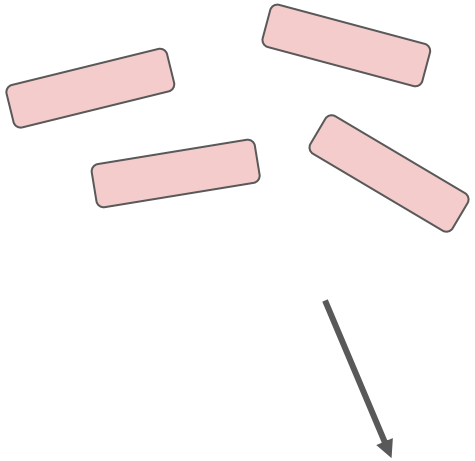
You Can Do This Too!

- Navigate to Demo Workspace:
  https://github.com/human-pangenomics/hprc-tutorials

Later (after workshop): Google “Sign Up For Terra”

  - FYI: need a Gmail account
- Use Demo Workspace (link above) to replicate analysis

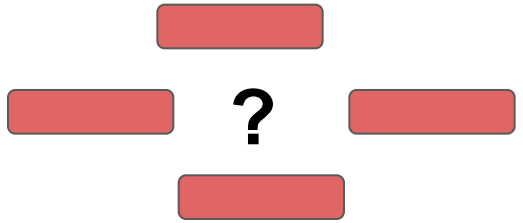
Variant Calling From Short Sequencing Reads

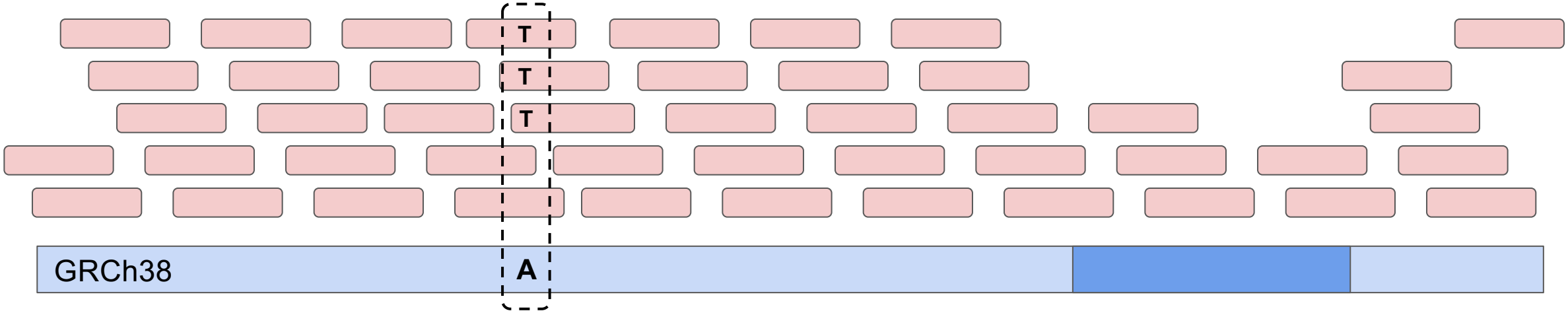


Sequencing reads mapped to a reference genome.

Variants called from the differences between reads and reference.

Unmapped when sequence not in the reference





Heterozygous variant A/T

Challenging region (e.g. repeat, highly polymorphic region)

DeepVariant

Machine learning approach to predict genotype at a candidate site from read pile-up images.

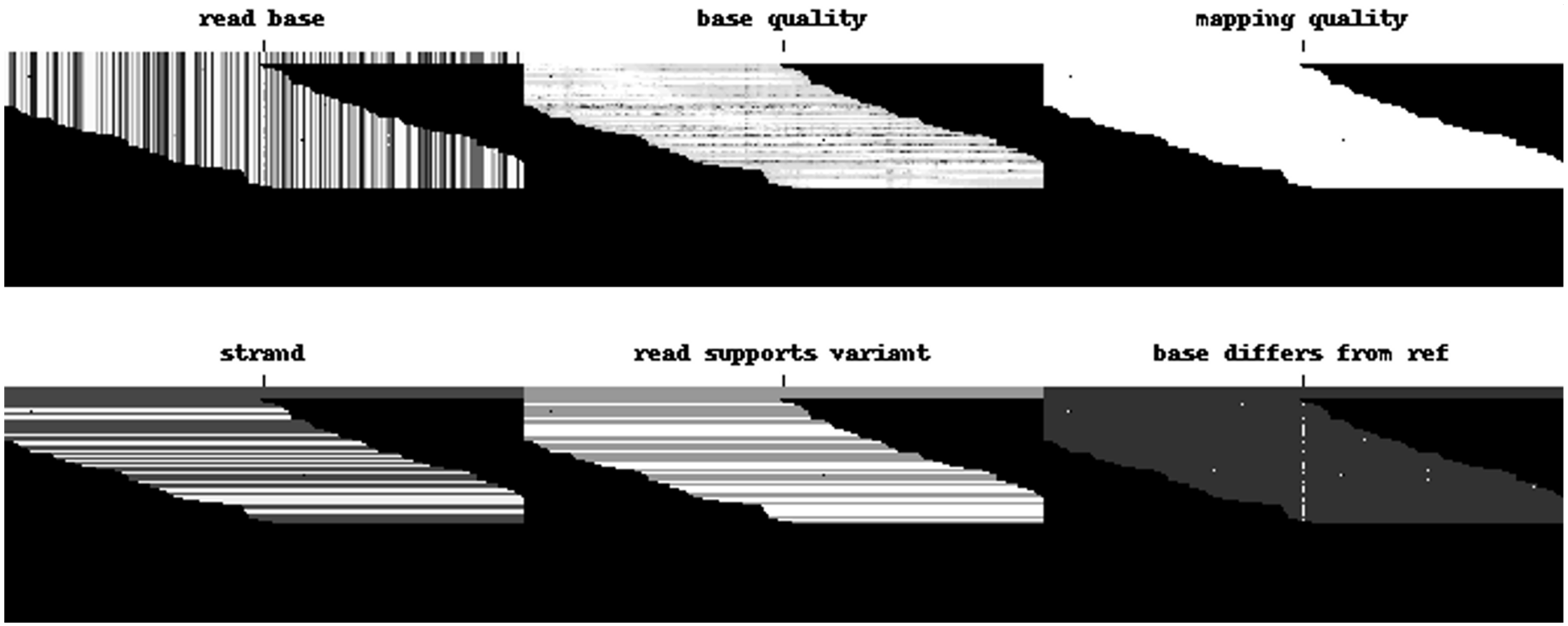
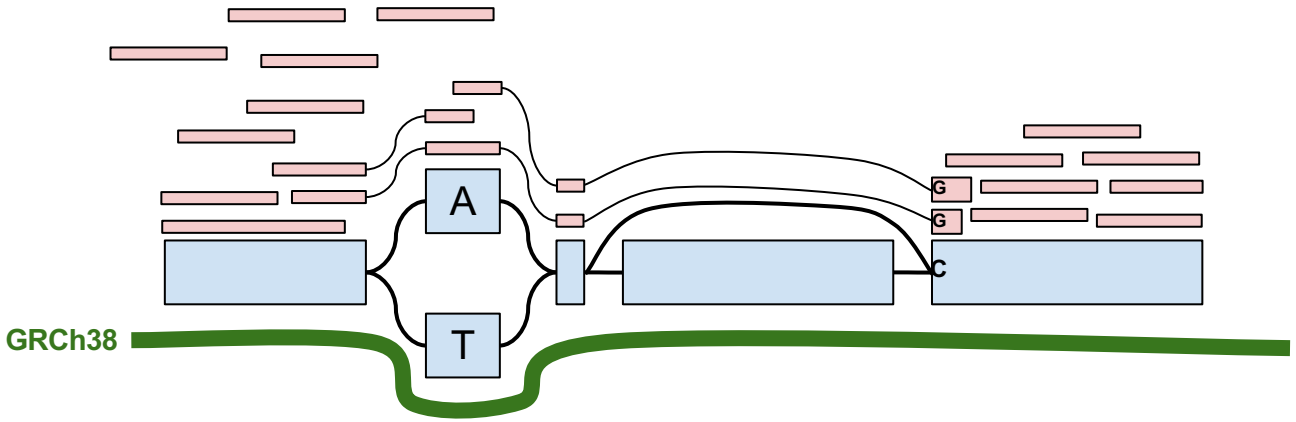
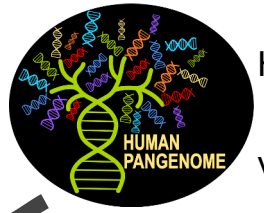


Image: https://google.github.io/deepvariant/

Giraffe/DeepVariant

1. Short sequencing reads **mapped to the pangenome** with *vg giraffe*





HPRC frequency-filtered Minigraph-Cactus pangenome v1.0 (90 haplotypes)

Giraffe/DeepVariant

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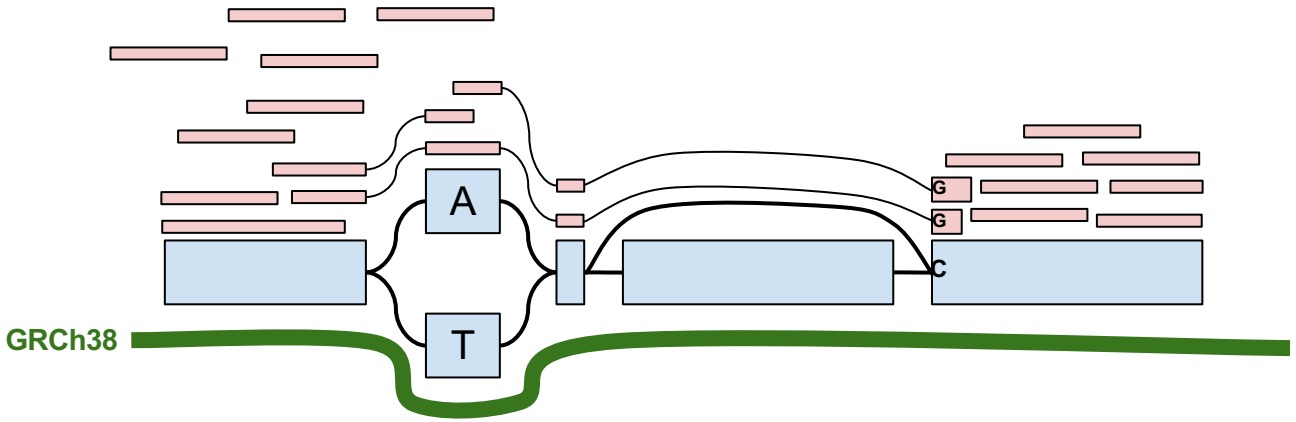
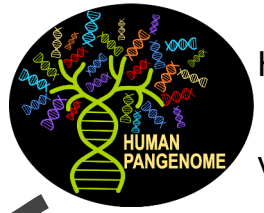
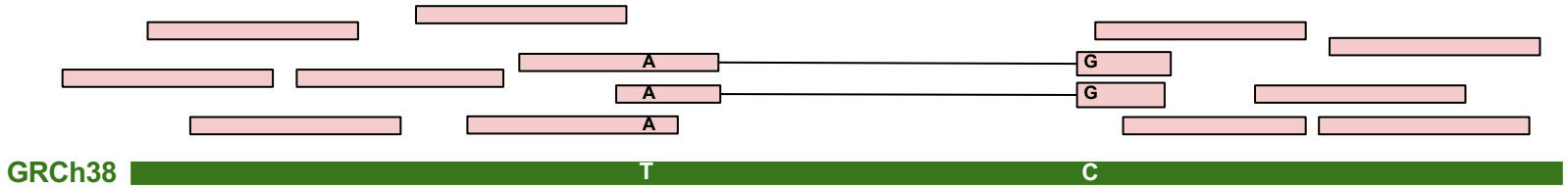




HPRC frequency-filtered Minigraph-Cactus pangenome v1.0 (90 haplotypes)

2. Aligned reads **projected to GRCh38** with *vg surject*



Giraffe/DeepVariant

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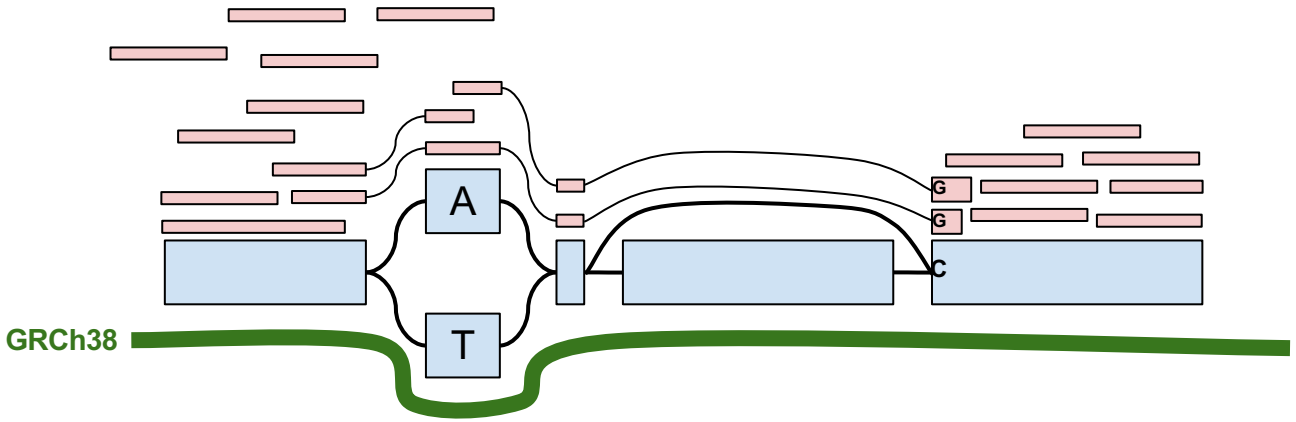

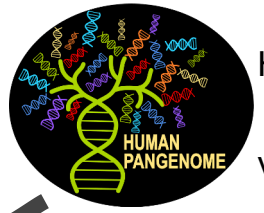

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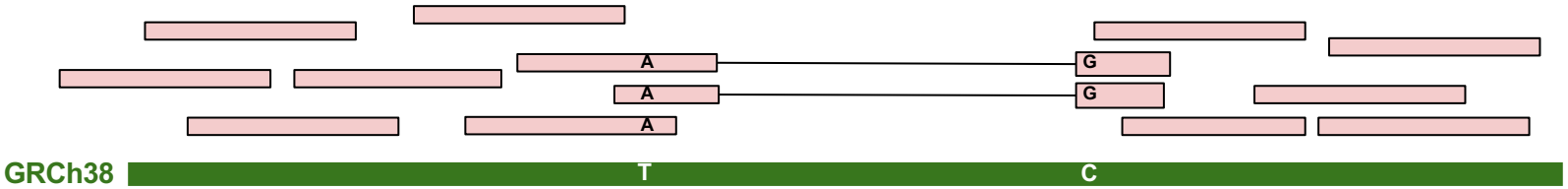
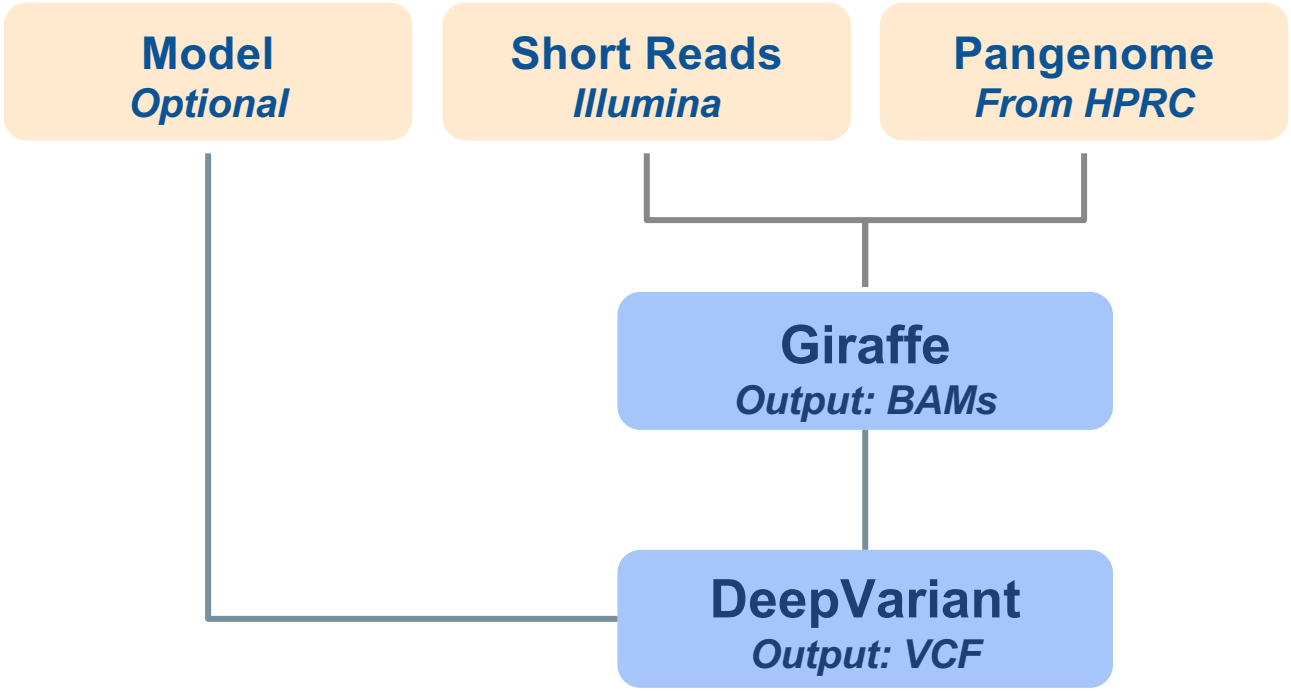
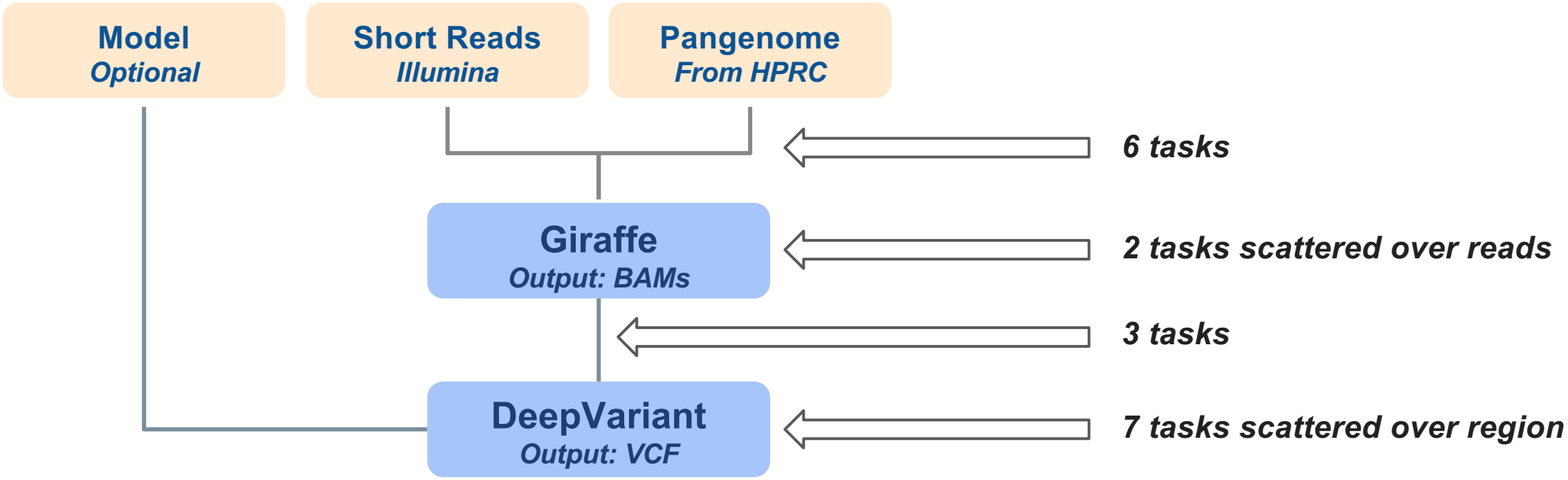


3. **Variants called with DeepVariant** trained on surjected alignments, after indel realignment.

Giraffe/DeepVariant Workflow (WDL)

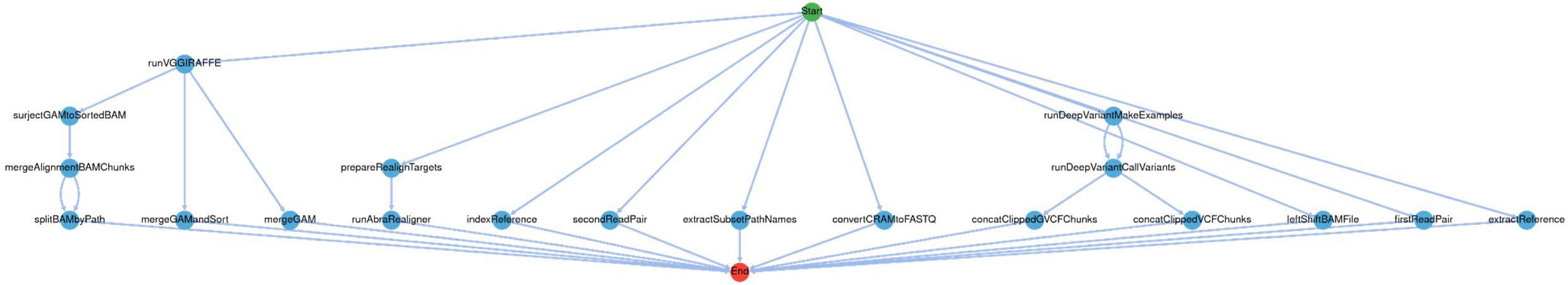


Giraffe/DeepVariant Workflow (WDL)



Giraffe/DeepVariant Workflow (WDL)

Can Be Treated By Users As One Step

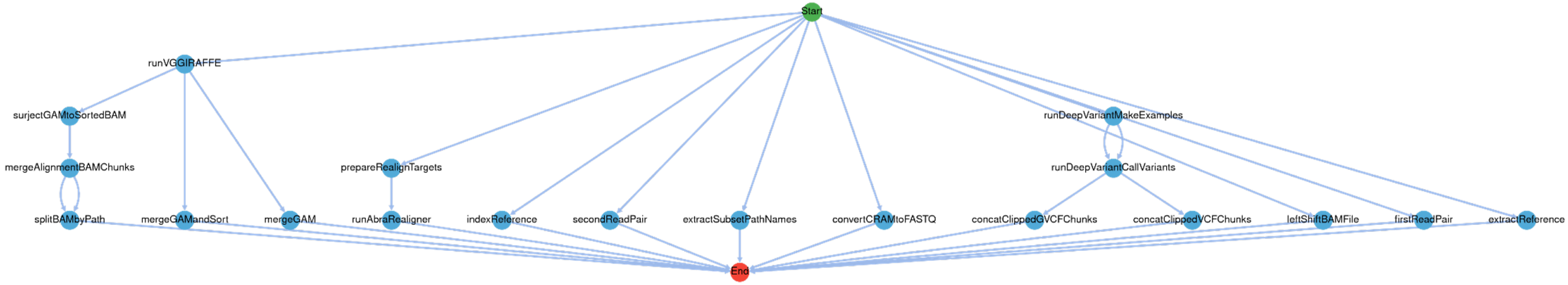


Motivation For Using Tools/Data In AnVIL

- Able to use pangenome in HPRC's workspace
- Easily find and import workflows from Dockstore
- Allows easy sharing of analysis results

Giraffe/DeepVariant Workflow (WDL)

Can Be Treated By Users As One Step



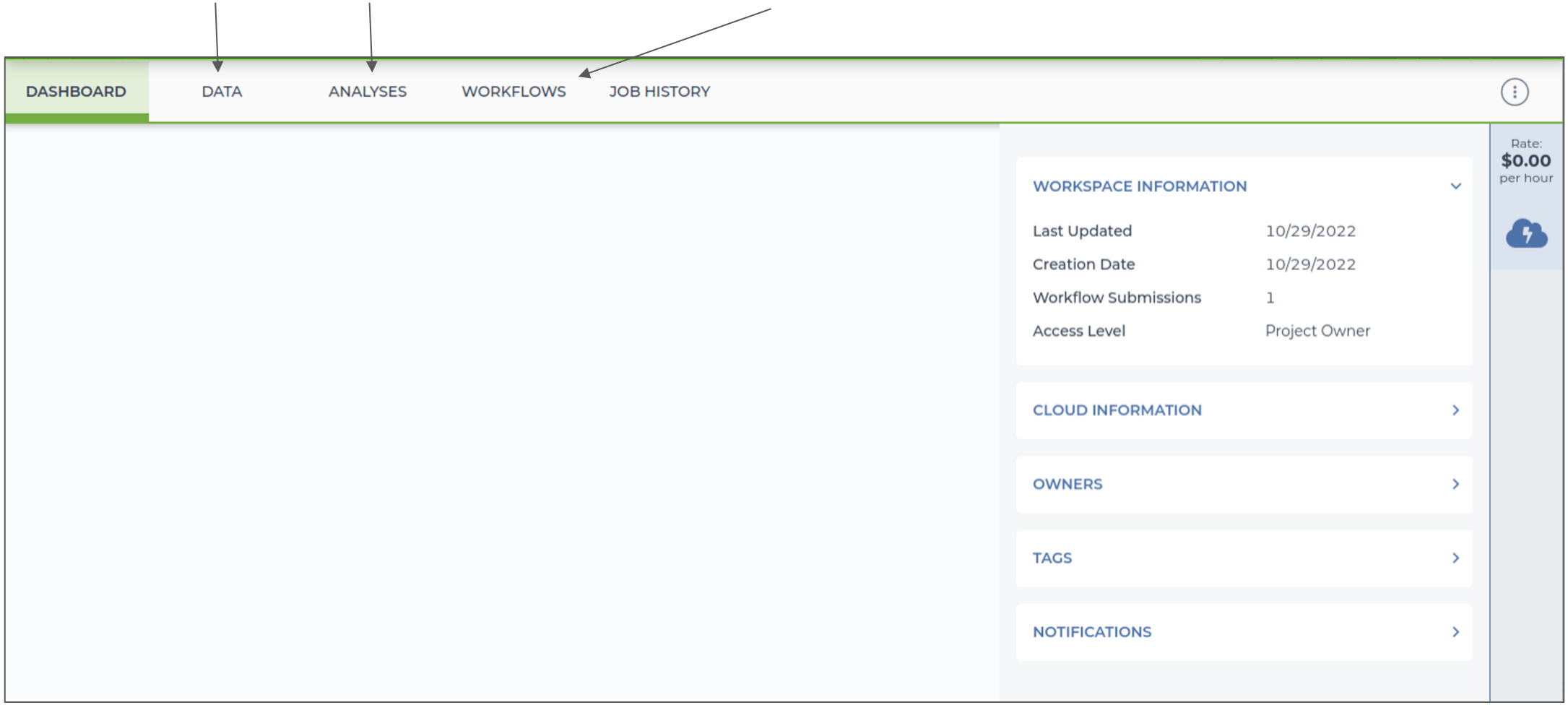
Motivation For Using Tools/Data In AnVIL

- Able to use pangenome in HPRC's workspace
- Easily find and import workflows from Dockstore
- Allows easy sharing of analysis results

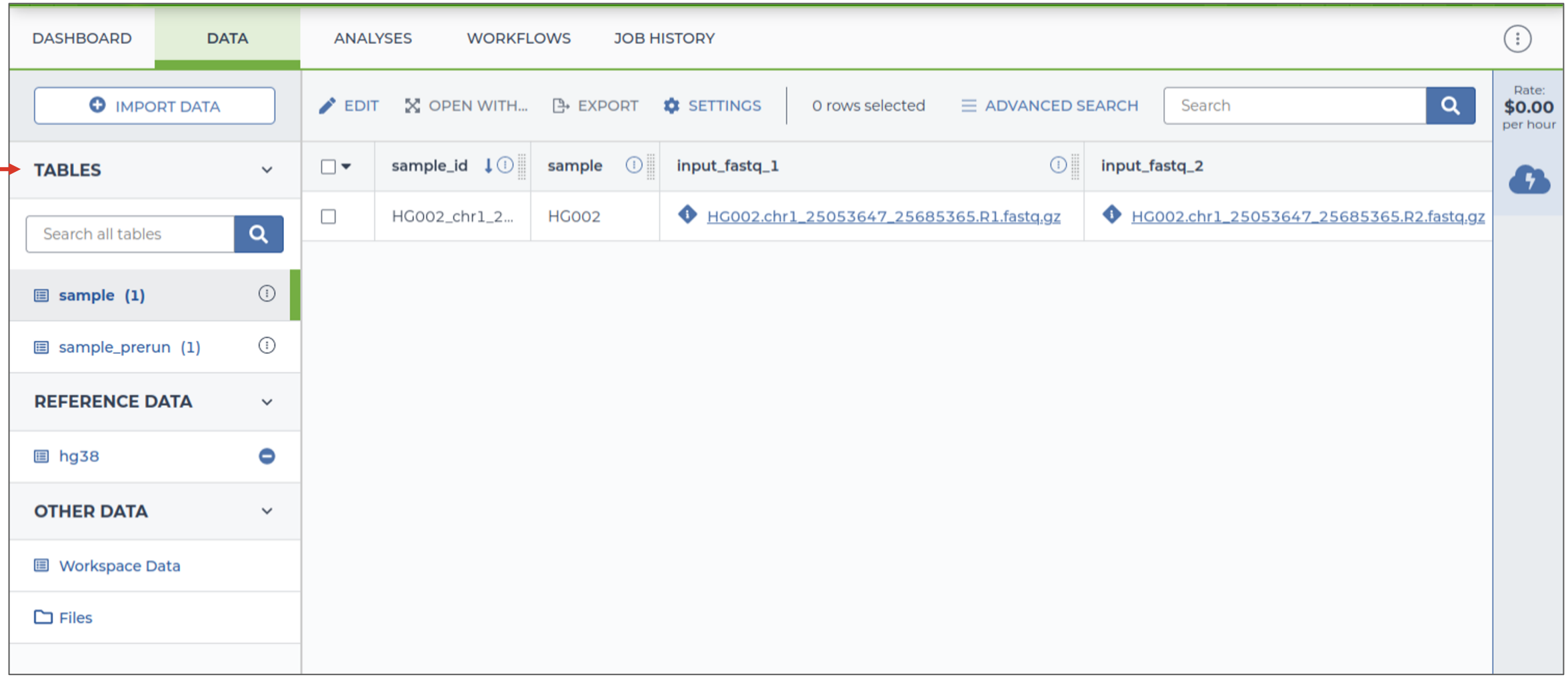
Running workflows with a workflow runner in the cloud (using data stored centrally in the cloud) is an important tool for genomic analysis.

First Look At a Workspace On Terra

Tabs to describe data, run interactive notebooks, or run workflows



Data Tab: Organizing The Data In Tables



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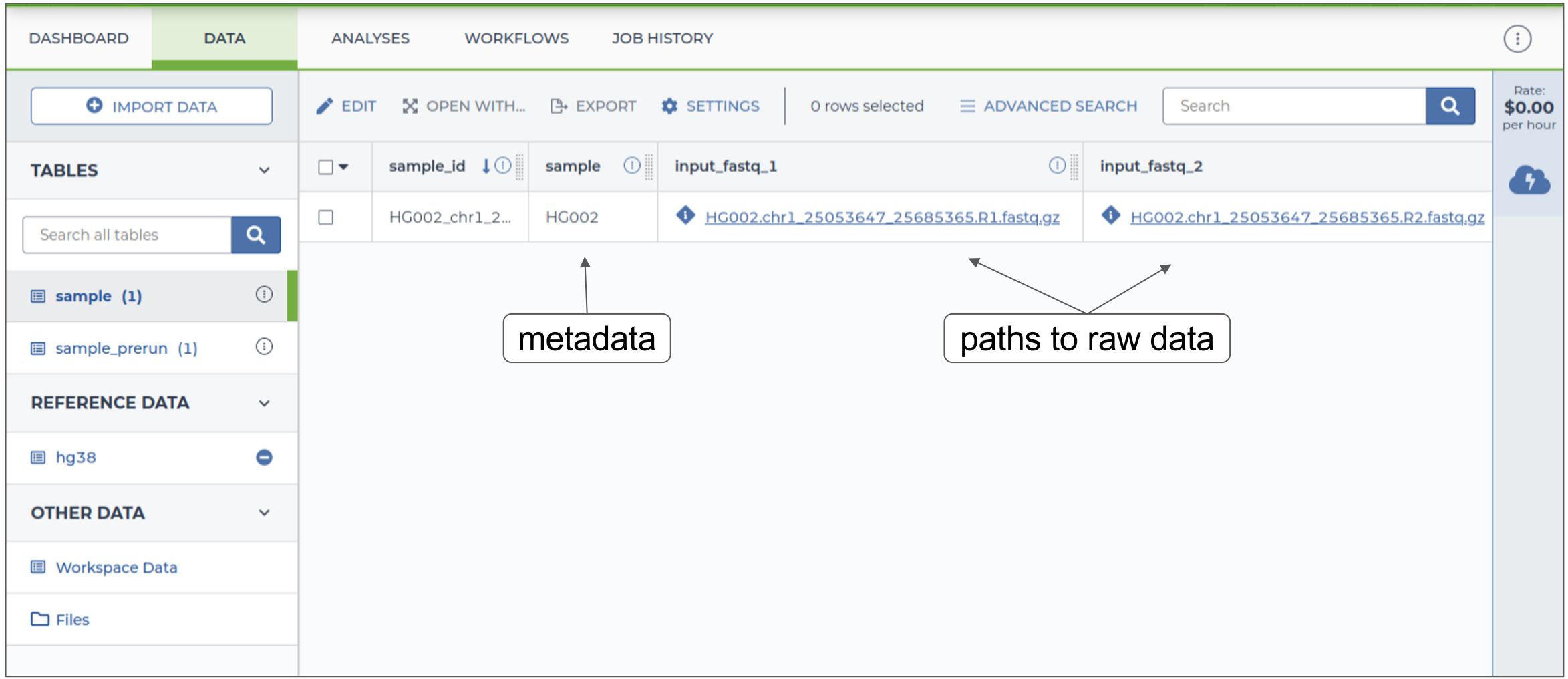
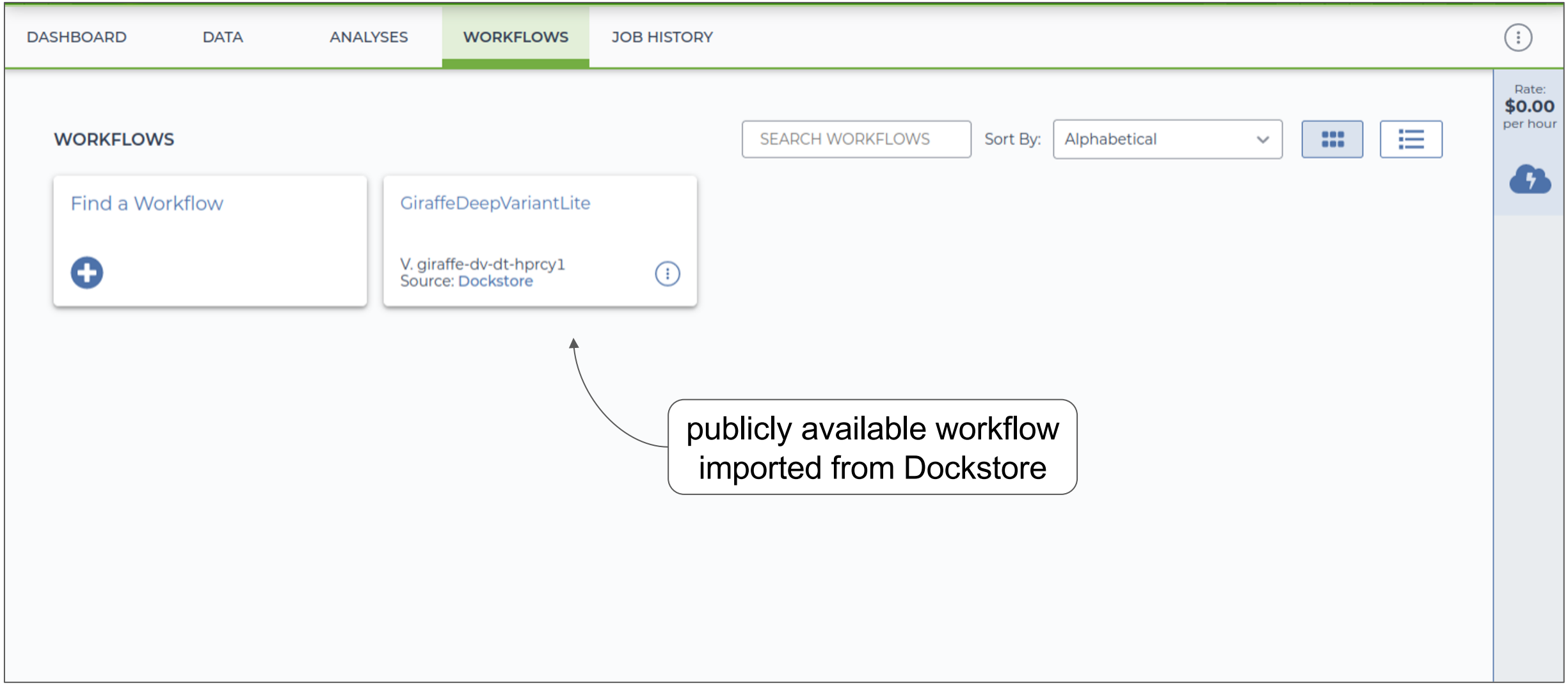
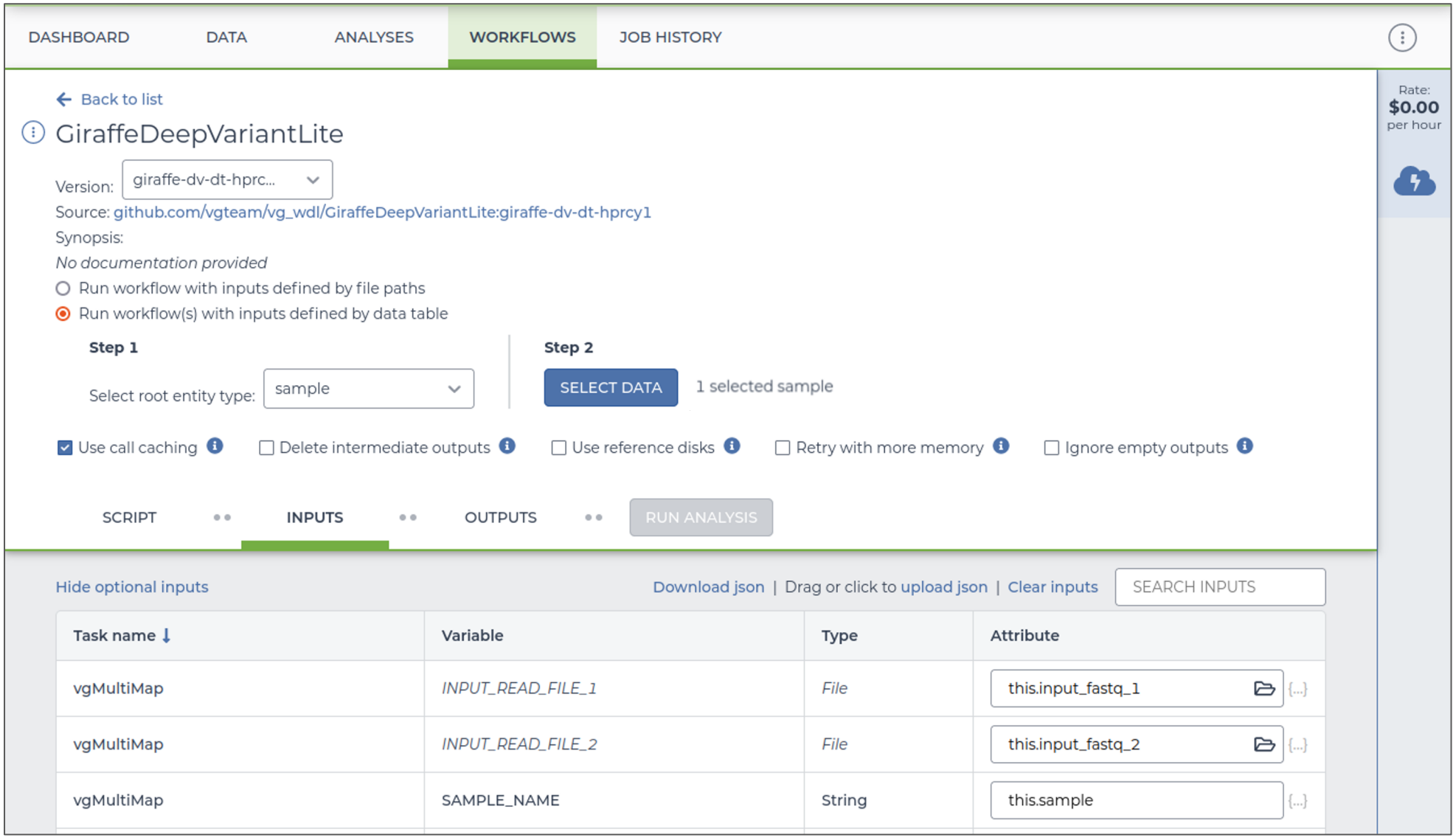


Workflows Tab: Launch Analyses



Configure Inputs Of The Workflow



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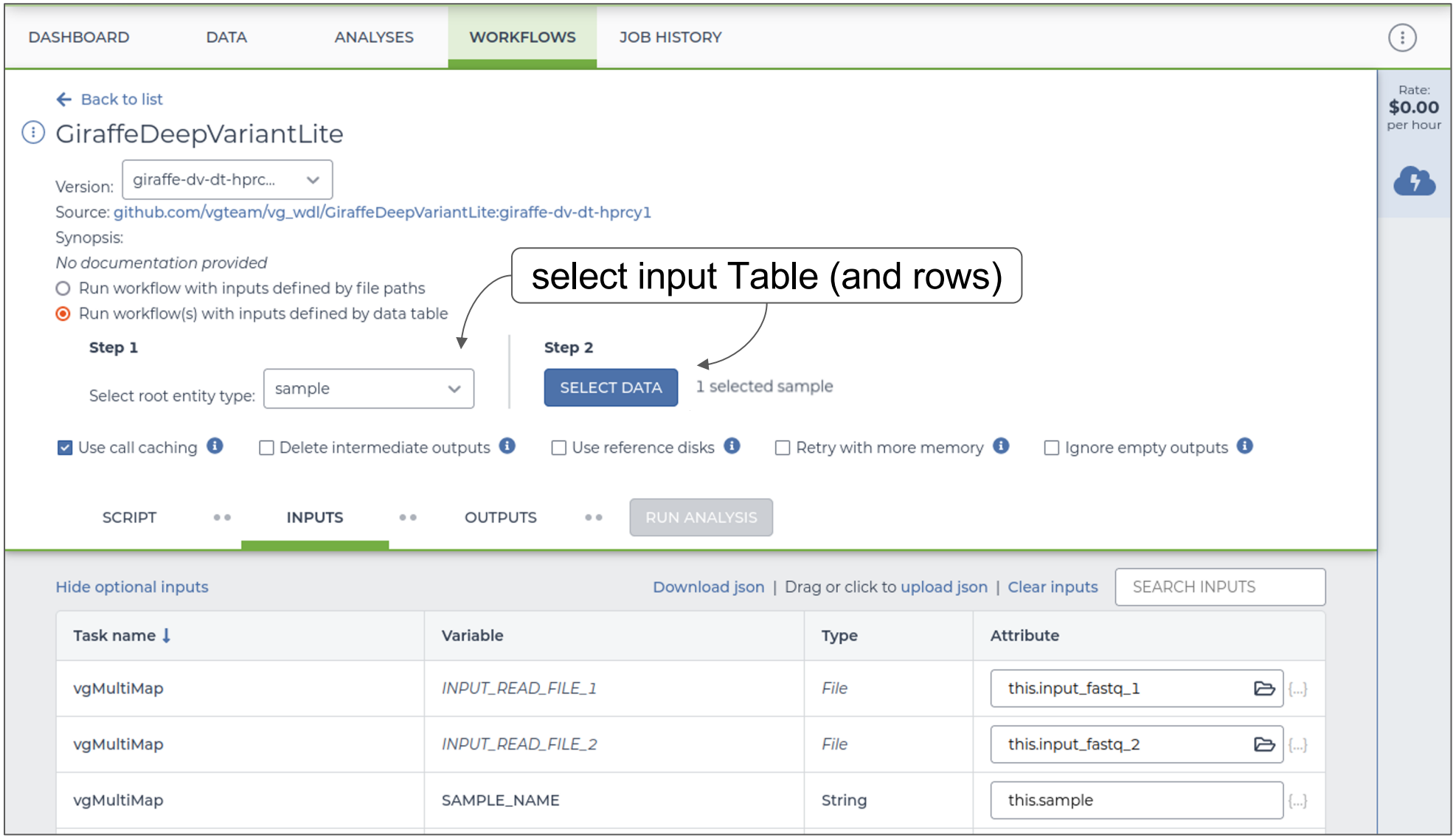
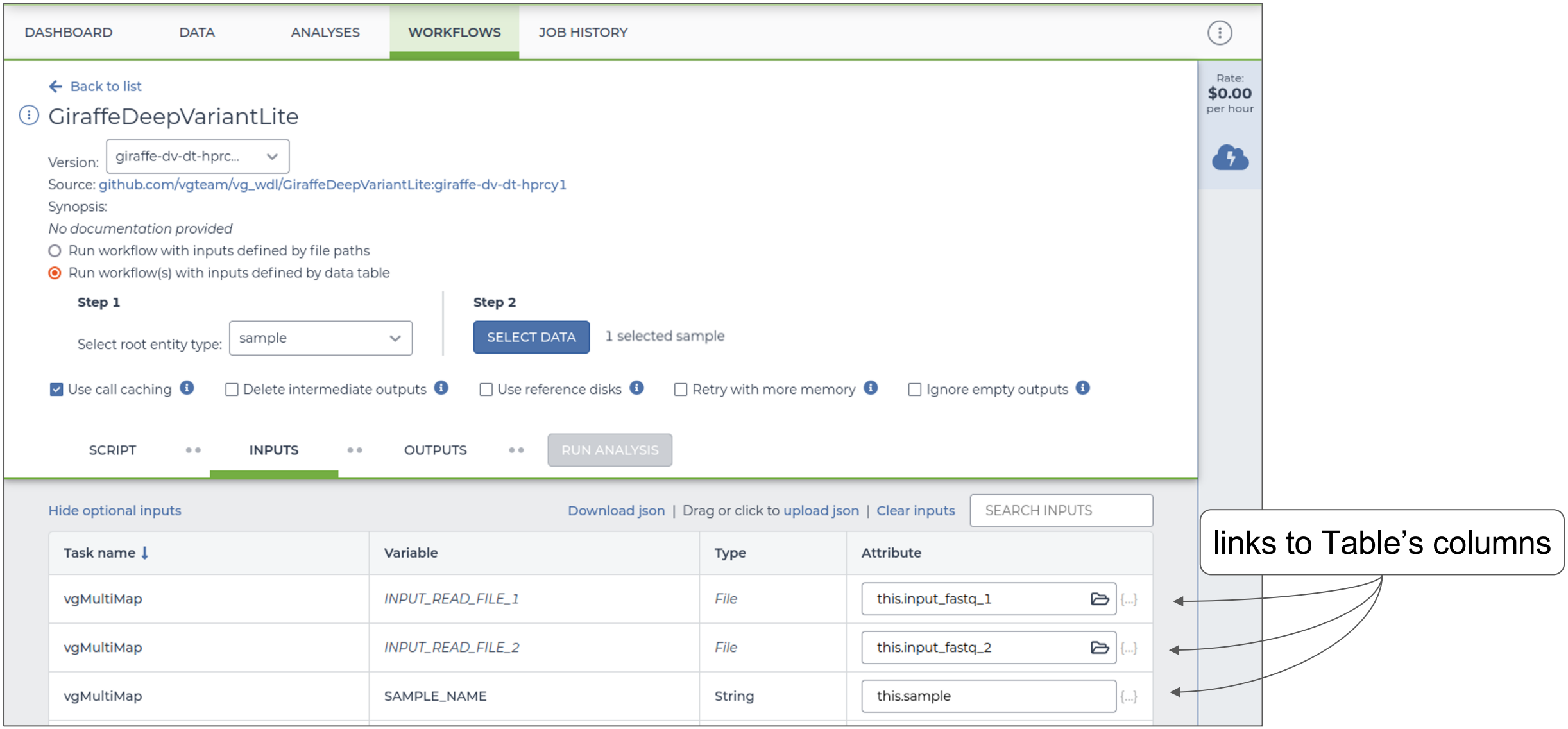
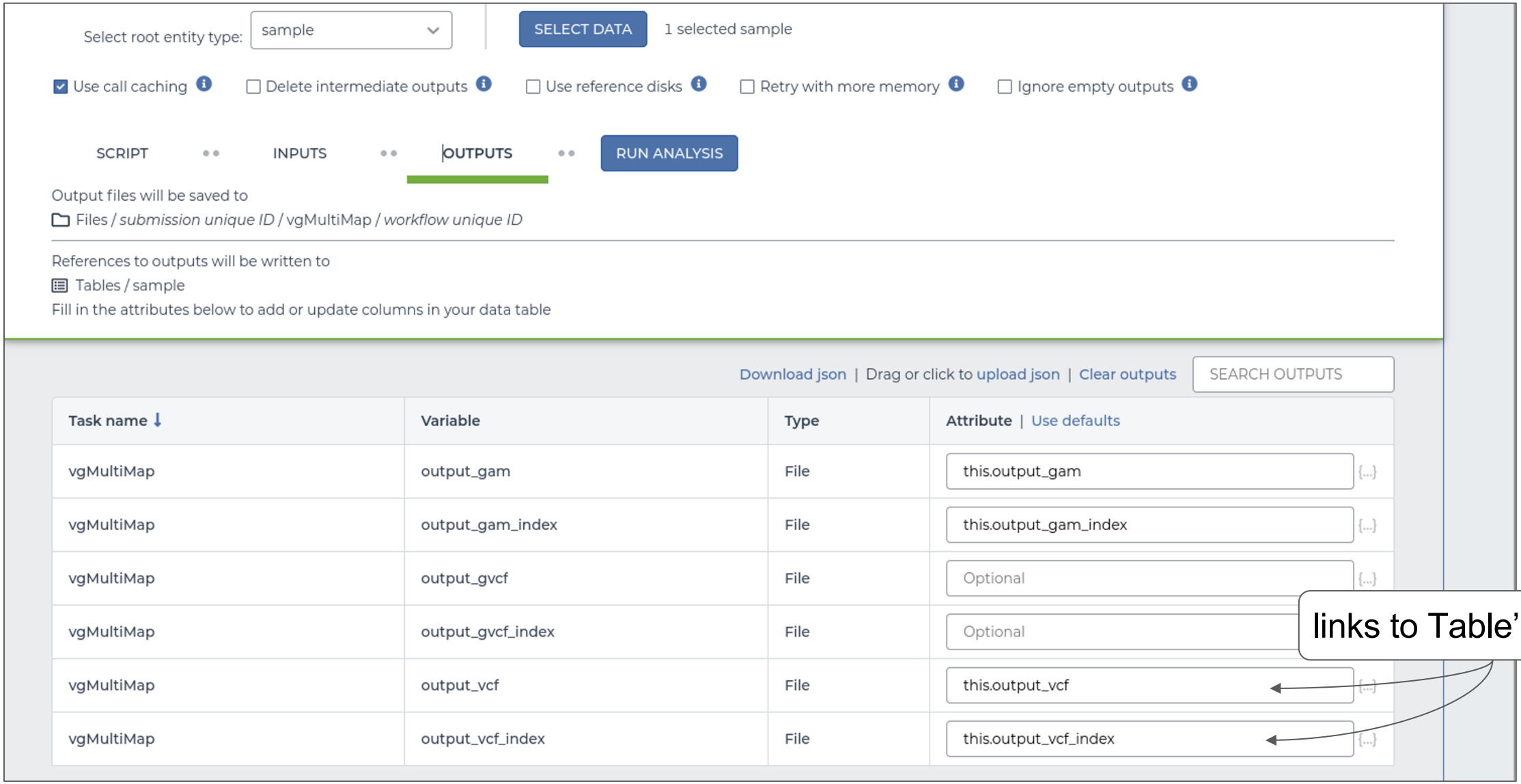


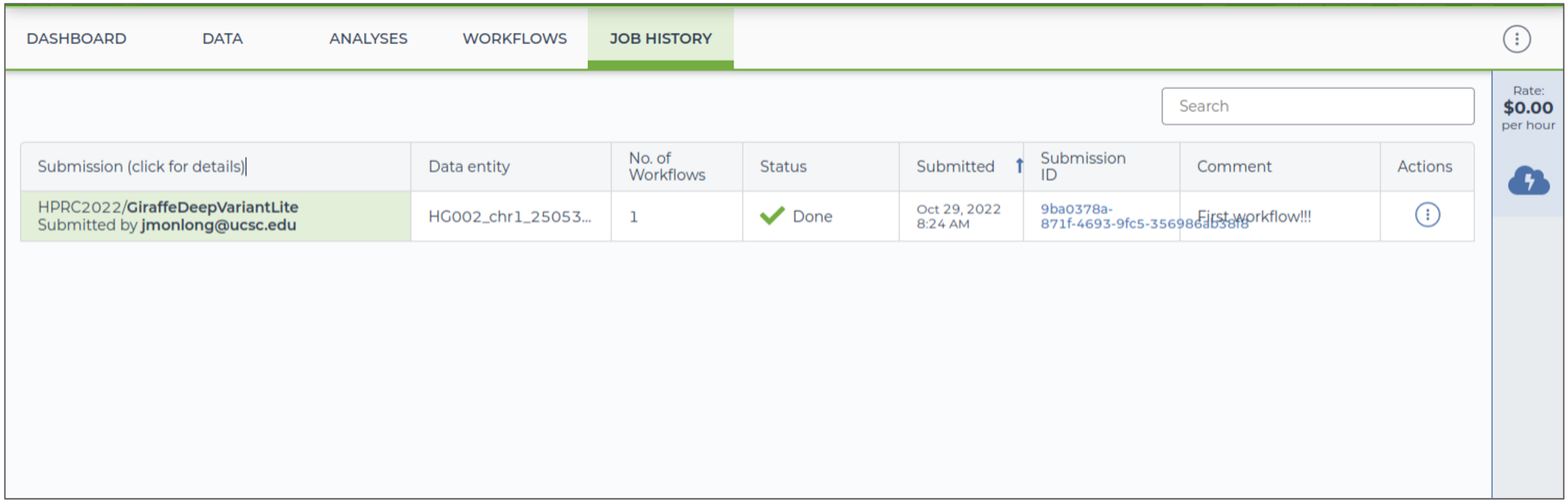
Configure Inputs Of The Workflow



Configure The Outputs Of The Workflow



Job History Tab: Monitor Running/Prior Jobs



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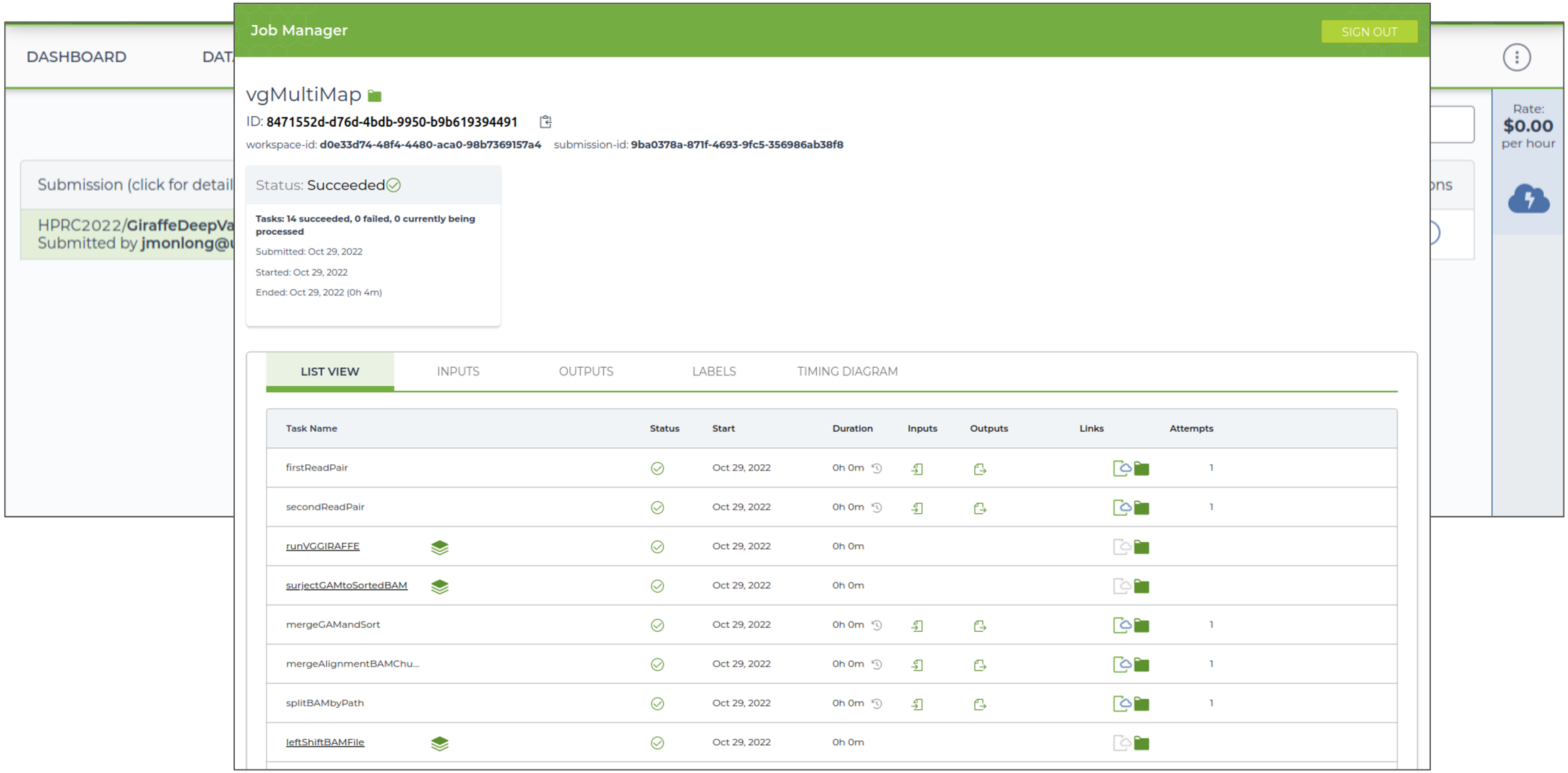
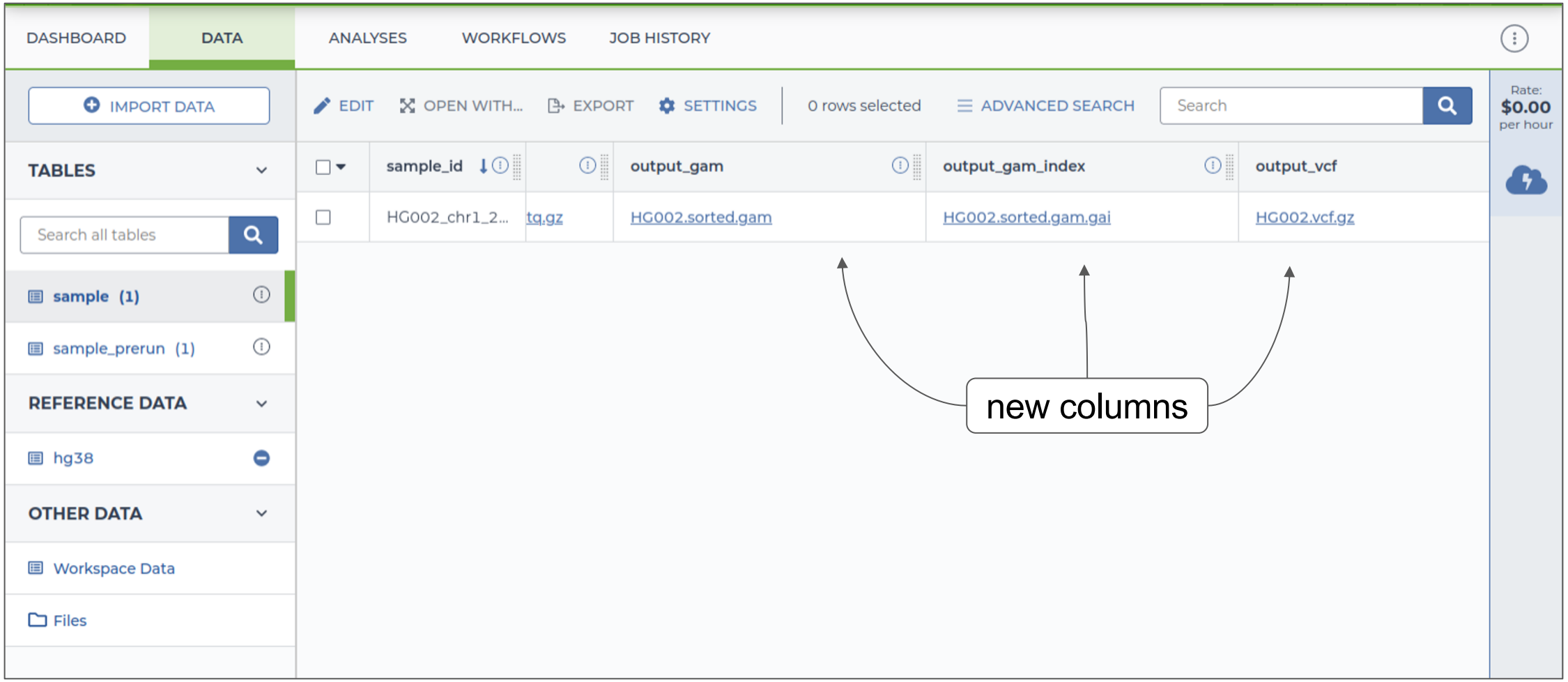
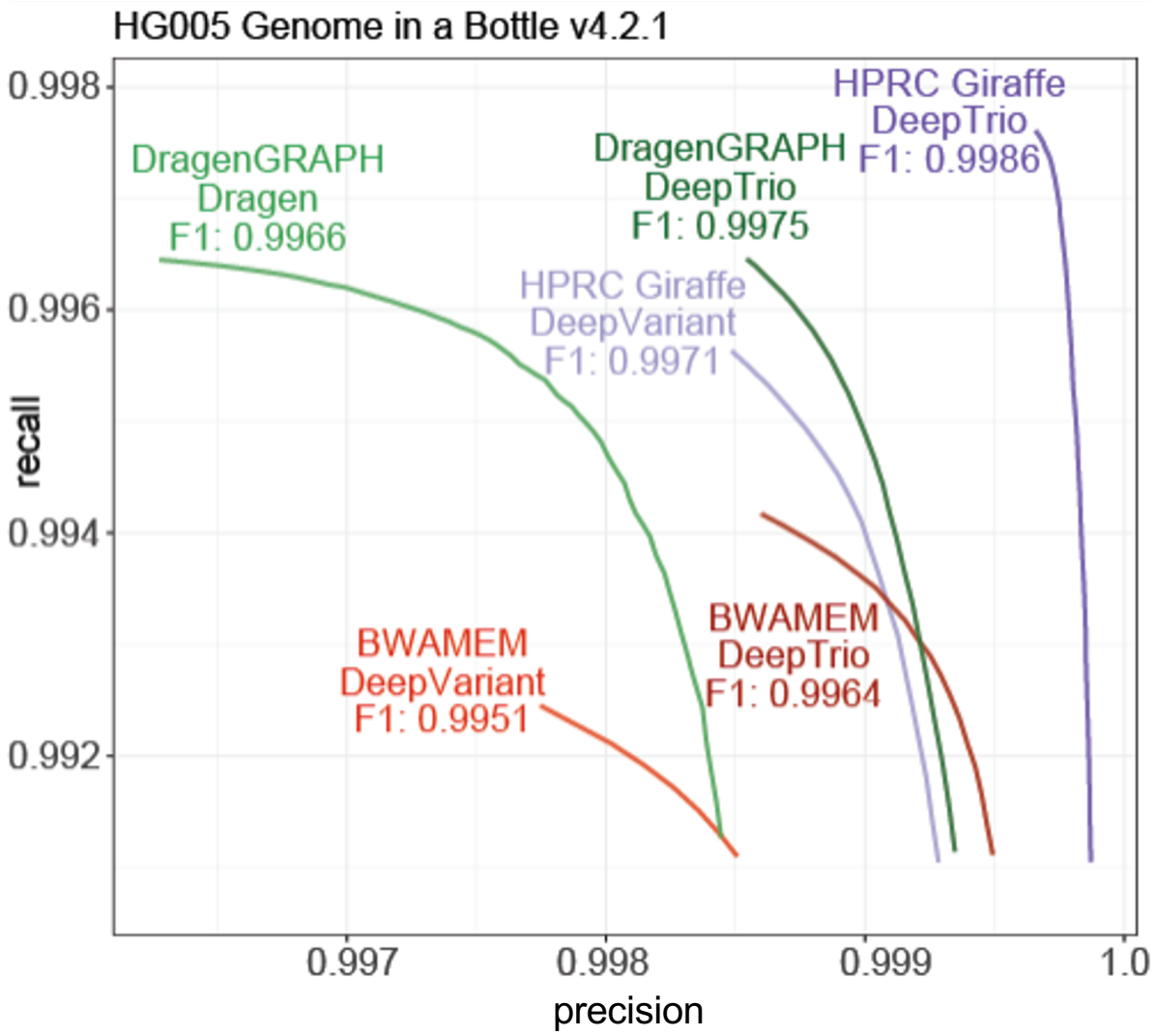


Results Are Also Organized in Data Tables



Giraffe/DeepVariant Improves Variant Calling

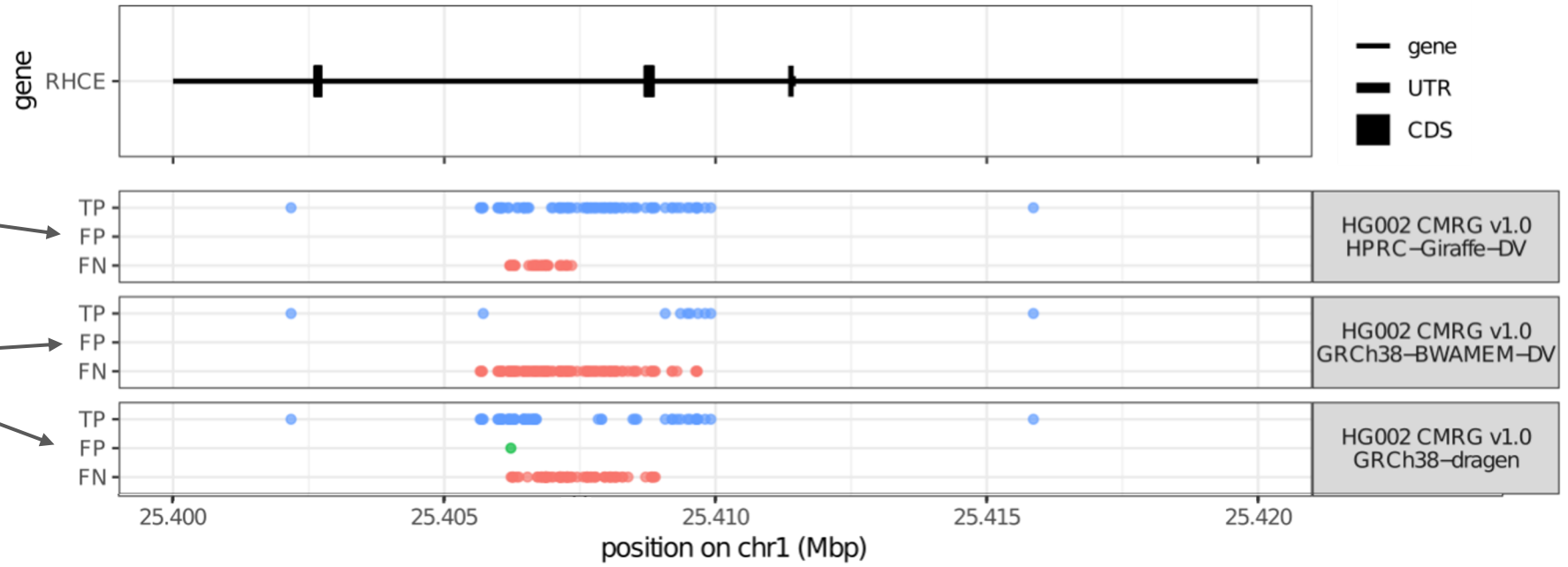
On average, 34% less errors compared to the linear reference approach (GRCh38-BWA-DeepVariant)



RHCE: A Challenging, Medically-Relevant Gene

Mostly true positives with HPRC-Giraffe+DeepVariant

Many false negative with BWA+DeepVariant or Dragen



Each point is a variant called on HG002

Complex Variation Around RHCE

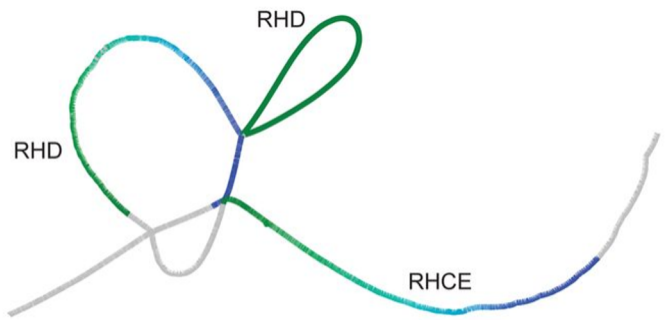
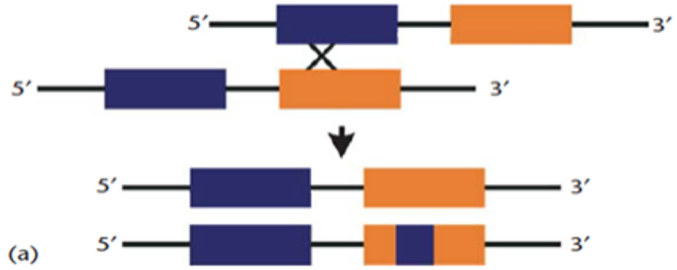


Image from Wikipedia

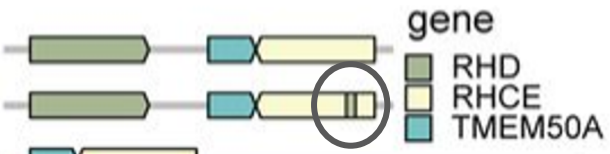


Gene conversion event

GRCh38

HG002

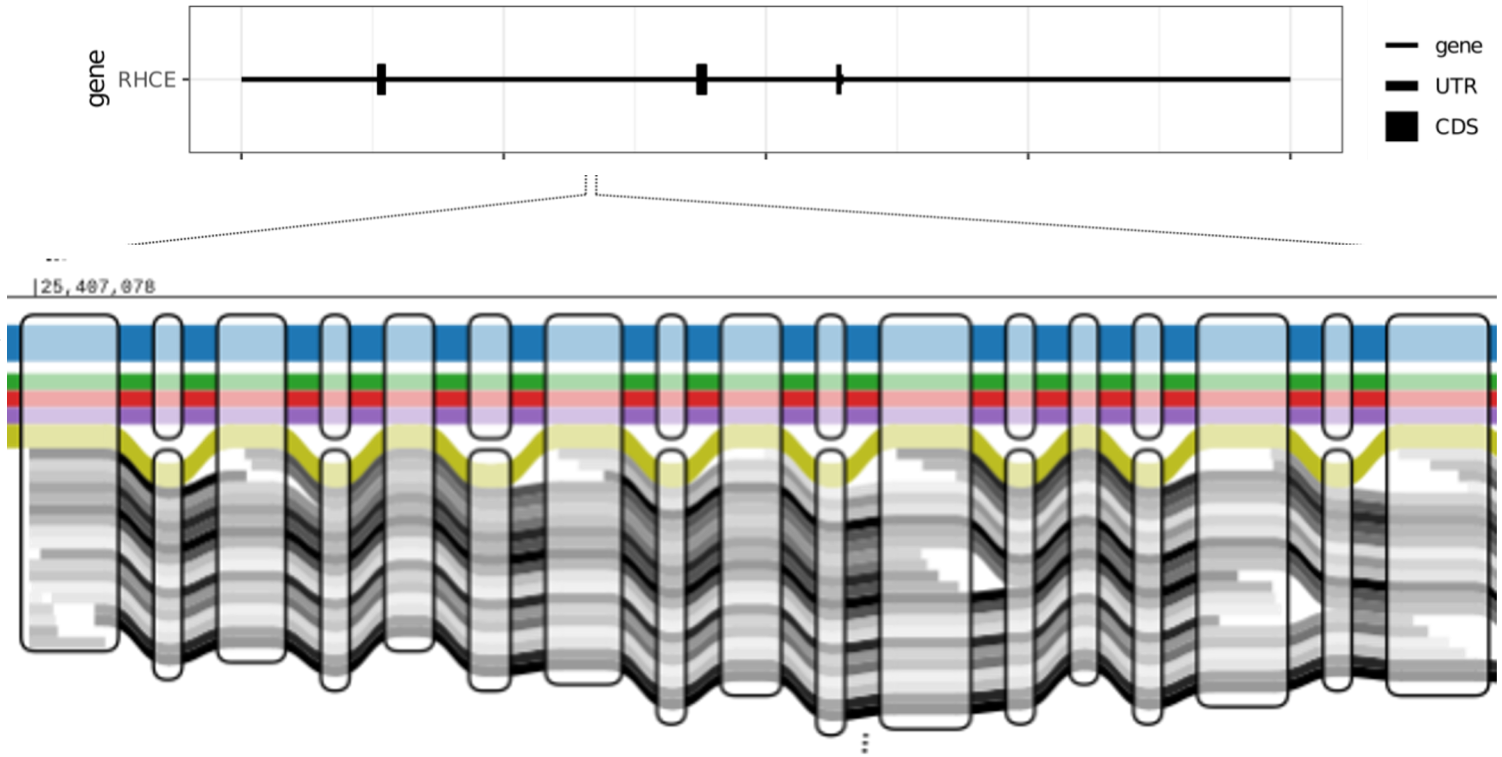
Count	Frequency	Haplotype name
43	0.48	RHD;RHCE
22	0.24	RHD;RHCE-RHD(2)-RHCE
15	0.17	RHCE
3	0.03	RHD-RHCE(2-3)-RHD;RHCE
2*	0.02	RHD-RHCE(8)-RHD;RHCE
1	0.01	RHCE-RHD(2)-RHCE
1*	0.01	RHD-RHCE(2-9)-RHD;RHCE
1*	0.01	RHD;RHCE-RHD(9)-RHCE
1*	0.01	RHD-RHCE(10);inv;RHCE-RHD(10)
1*	0.01	RHD;RHD;RHCE-RHD(9)-RHCE



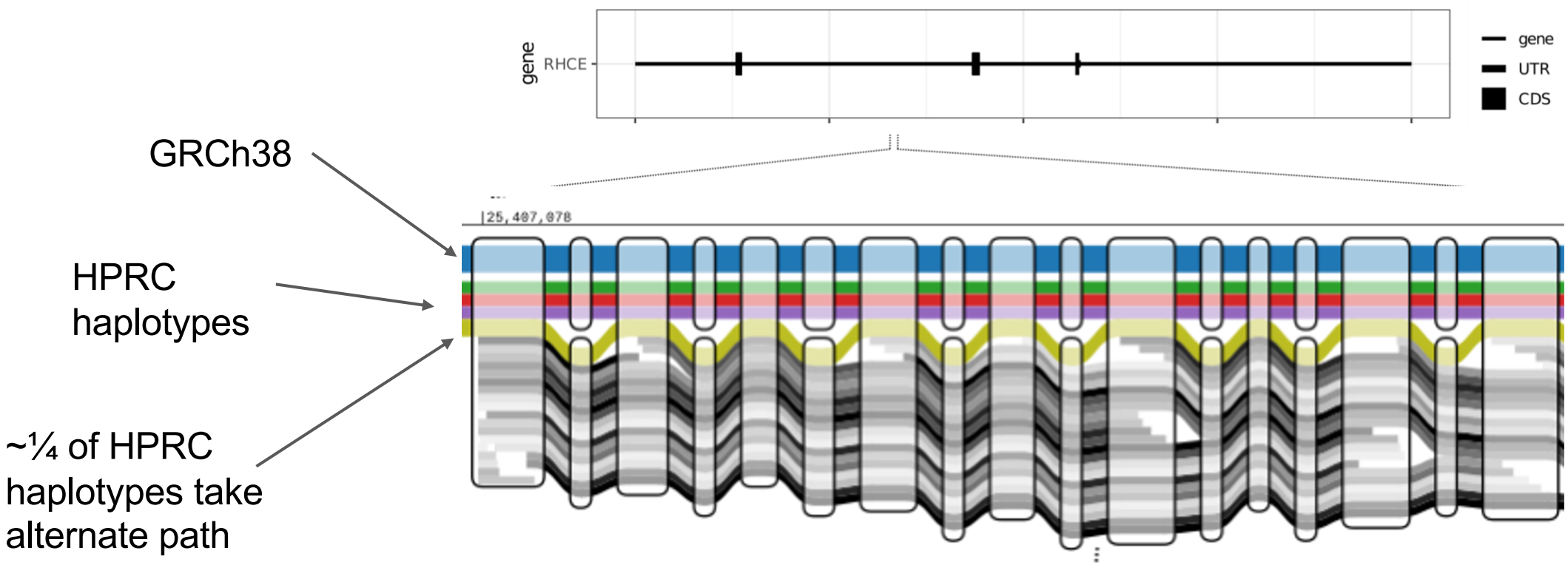
Small Reads Map To A Subset Of Haplotypes

GRCh38

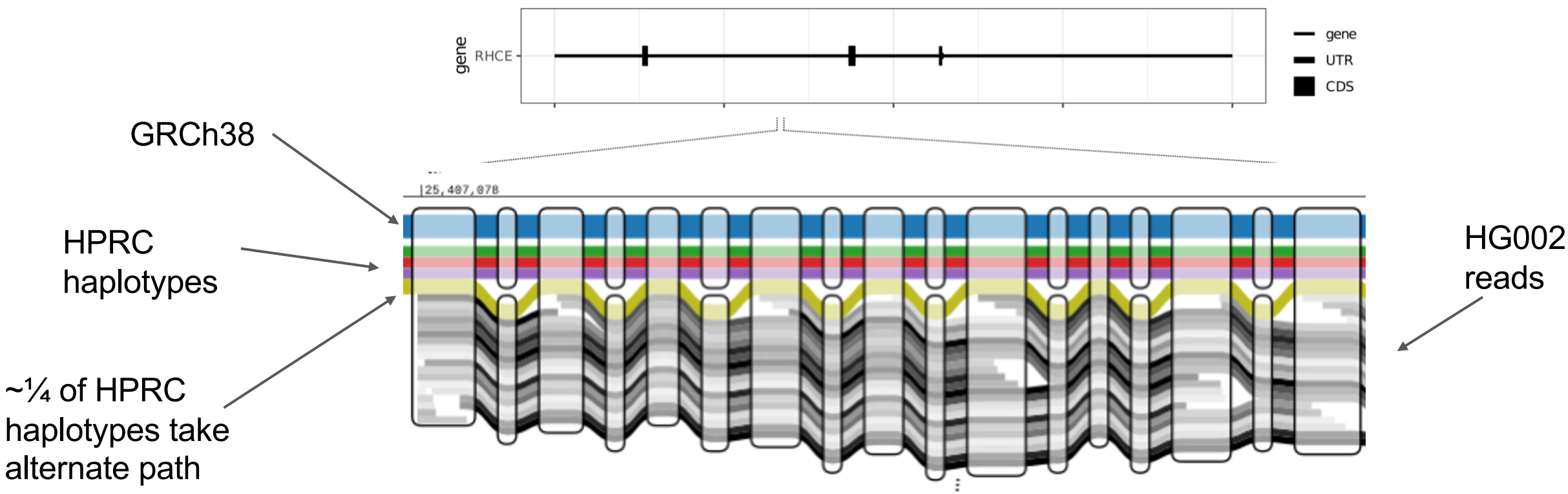
HPRC haplotypes



Small Reads Map To A Subset Of Haplotypes



Small Reads Map To A Subset Of Haplotypes



Another example: VUS observed in healthy individuals

rs1553862732 https://www.ncbi.nlm.nih.gov/snp/rs1553862732 **Current Build** 156 **Released** September 21, 2022

Organism	*Homo sapiens*	**Clinical Significance**	Reported in ClinVar
Position	chr2:151600710 (GRCh38.p14)	**Gene : Consequence**	NEB : Missense Variant
Alleles	A>G	**Publications**	0 citations
Variation Type	SNV Single Nucleotide Variation	**Genomic View**	See rs on genome
Frequency	None		

Variant Details | **Clinical Significance** | HGVS | Submissions | History | Publications | Flanks

Allele: G (allele ID: 516380)

ClinVar Accession	Disease Names	Clinical Significance
RCV000641401.5	Nemaline myopathy 2	Uncertain-Significance

Frequencies https://varsome.com/variant/hg38/rs1553862732

gnomAD Genomes

This variant does not have a gnomAD genomes entry, but its locus is covered in gnomAD genomes as follows.

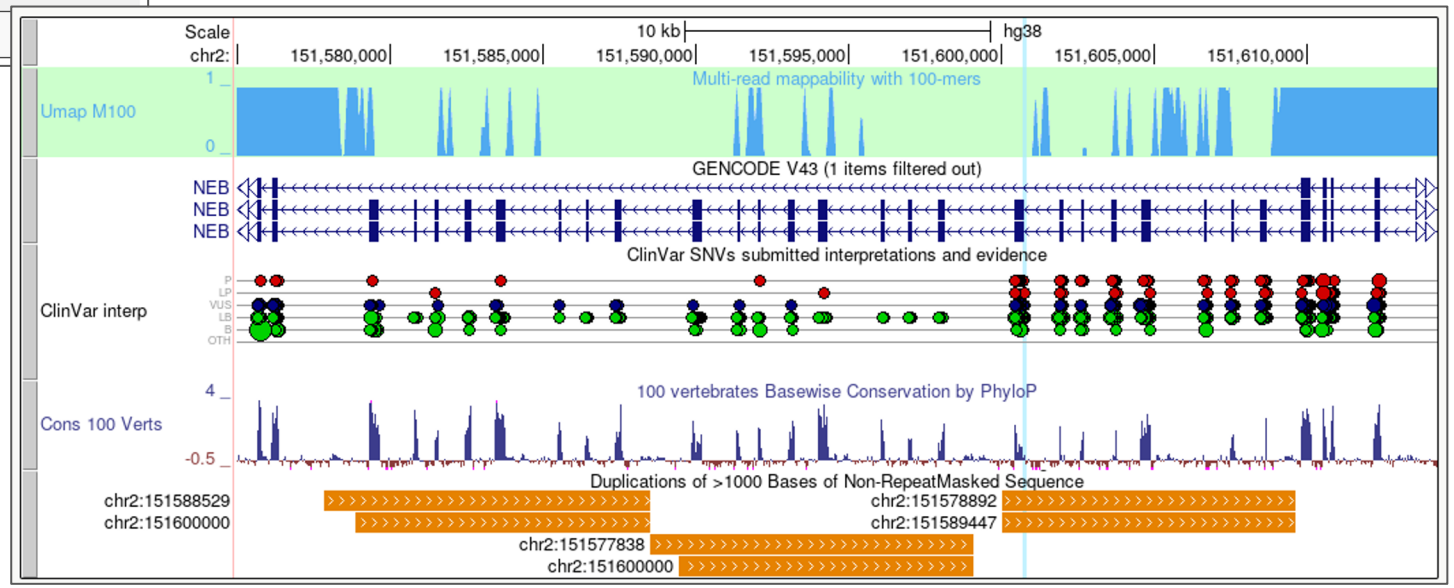
gnomAD Genomes Coverage Version: 3.0

Coverage in gnomAD Genomes samples

Mean coverage	Median coverage	% of samples over 20x coverage
3.1	3	0.0076%

Found in 3 individuals of the 1000 Genomes cohort when using Giraffe-DeepVariant on the HPRC pangenome (AF ~0.002)

Hard region to map short reads to because of segmental duplications.



The Pangenome Reference For Clinical Variant Calling and Interpretation (Discussion)

How might the adoption of a pangenome reference improve variant calling and/or interpretation?

What are the key barriers to adoption of a human pangenome reference in clinical genetics?

How might we design the pangenome reference tools to best serve the clinical genetics community?

Acknowledgements

AnVIL

HUMAN PANGENOME

HPRC Coordinating Center
Ting Wang, Heather Lawson, Nathan Stitziel, Lucinda Fulton, Robert Fulton, Tina Lindsay, Sarah Cody, Milinn Kremitizki, Eddie Belter, Derek Albracht, Haley Abel, Chris Markovic, Wen-Wei Liao

David Haussler, Benedict Paten, Karen Miga, Ed Green, Mark Akeson, Adam Novak, Glenn Hickey, Miten Jain, Hugh Olsen, Erik Garrison, Jean Monlong, Adirna Fuller, Xian Chang, Marina Haukness, Trevor Pesout, Beth Sheets, Tony Tsung Yu Lu, Jonas Sibbesen, Julian Lucas, Ryan Lorig-Roach, Jouni Siren, Kishwar Shafin, Charles Markello, Jordan Eizenga, Melissa Meredith, Brian Hannafious

Evan Eichler, Katy Munson, David Porubksy, Mitchell Vollger, Pete Audano, Arvis Sulovari

Erich Jarvis, Olivier Fedrigo, Giulio Formenti, Lauren Shalmiyev, Sadye Paez

Eimear Kenny, Lisa Wang

Matthew Mitchell, Brittney Martinez, Ellen Kelly

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Valerie Schneider, Chunlin Xiao, Terence Murphy, Paul Kitts, Francoise Thibaud-Nissent

Gene Myers

Ira Hal, Wen-Wei Liao, Shuangjia Lul

Paul Flicek, Susan Fairley, Daniel Zerbino

We would like to acknowledge the National Genome Research Institute (NHGRI) for funding the following grants which are in support of creating the human pangenome reference: 1U41HG010972, 1U01HG010971, 1U01HG010961, 1U01HG010973, 1U01HG010963, and the Human Pangenome Reference Consortium (https://humanpangenome.org/)

Partnerships

*Robert Cook-Deegan, Nanibaa' Garrison, Barbara Koenig, Alice Popejoy, Heather Lawson, Vasiliki Rahimzadeh, Ann McCartney, Joe Yracheta, Jonathan Lotempio, Pearl O'Rourke, Jean McKewen, Shawneequa Callier, Pilar Ossorio, Mahsa Shabani, Leroy Hubert, Clement Odebamowo

Pangenome Workshop ACMG 2023

0 done

0 underway