

**Connecting scientists to the patients,
datasets, and tools they need to do
life-changing research**

Consent for Research
(patients)



Data ingest
(hospitals and labs)



Data harmonization
(data stewards)



Data storage & maintenance
(data custodians)



DATA
GENERATORS

**DATA
PRODUCTION & CURATION**

INTEROPERABLE
SERVICES



BIOMEDICAL
RESEARCHERS

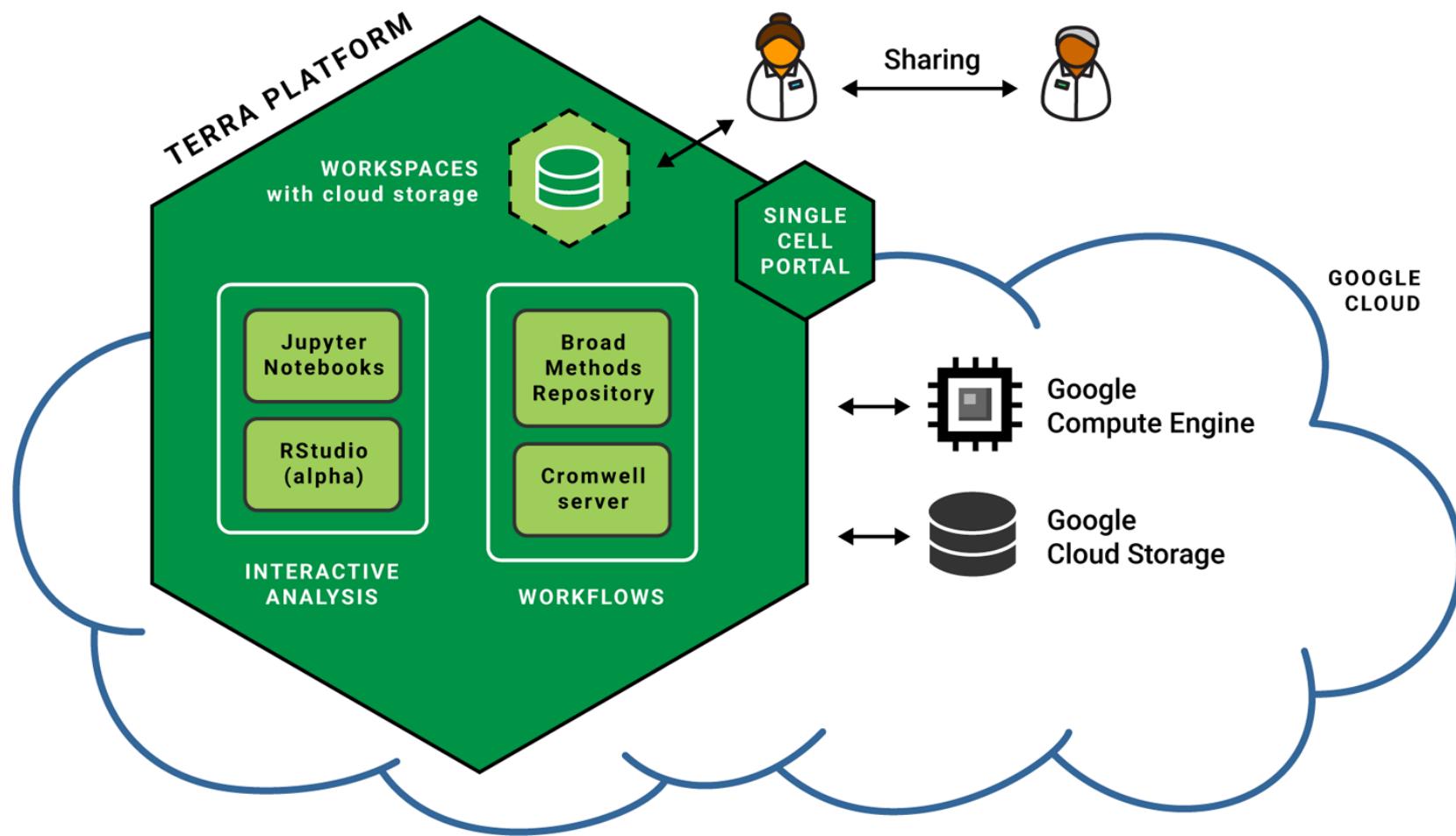


TOOL/METHOD
DEVELOPERS

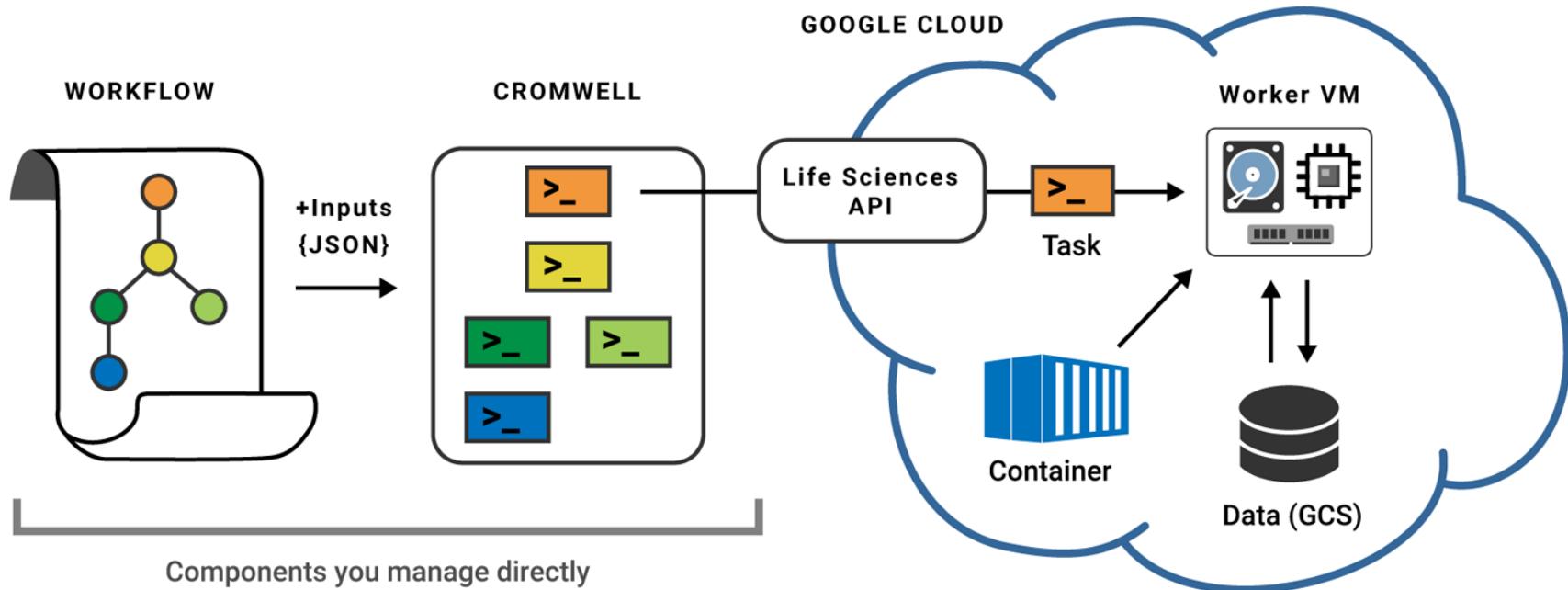
**TOOL
CREATION &
DISTRIBUTION**



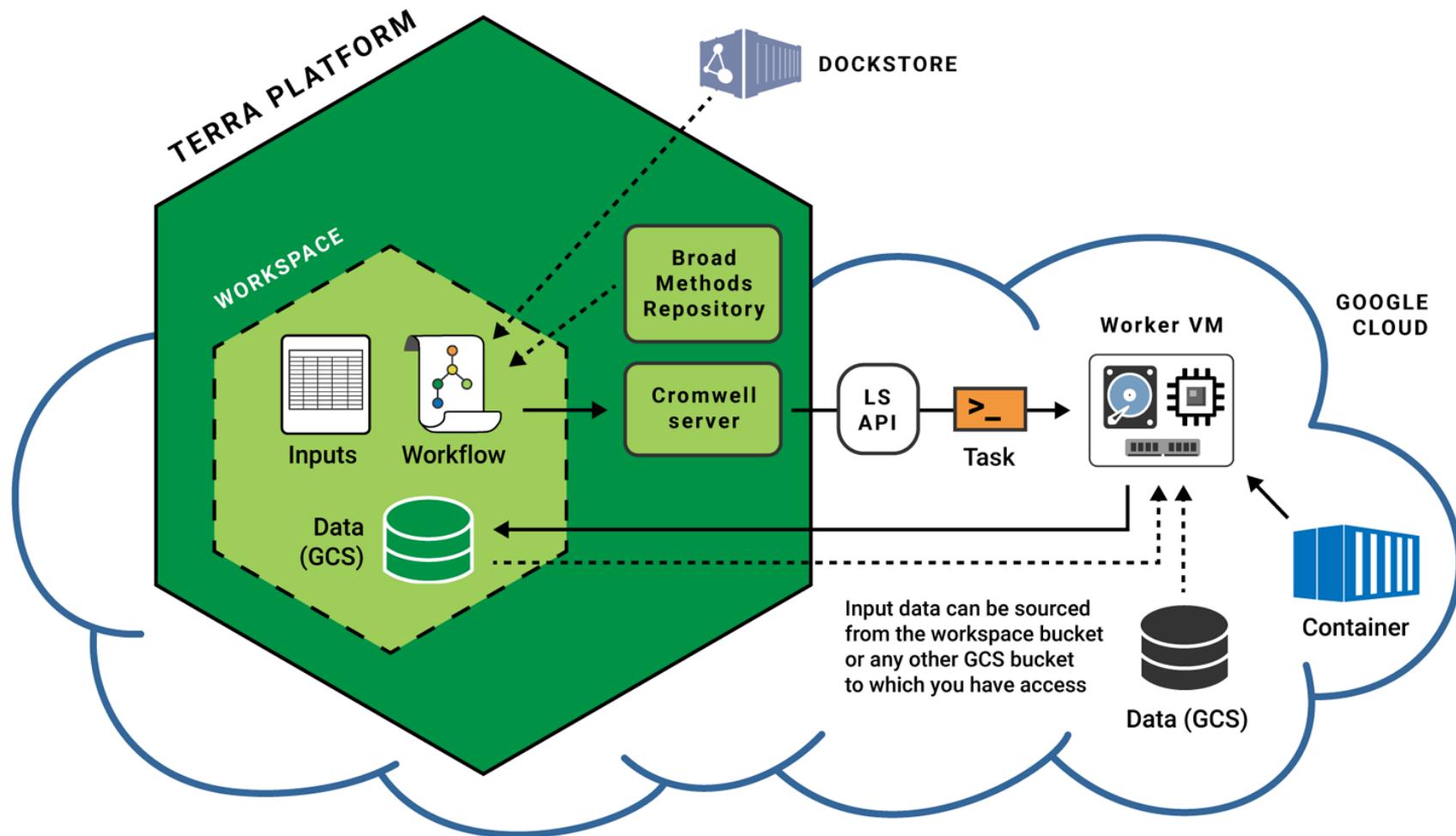
- Built in DSP
- 3P/Extensions



RUNNING WORKFLOWS



Cromwell dispatching workflows to Google Cloud



WORKFLOWS

SEARCH WORKFLOWS

Sort By: Alphabetical



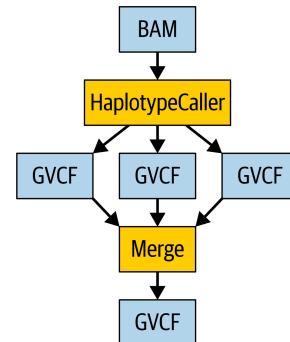
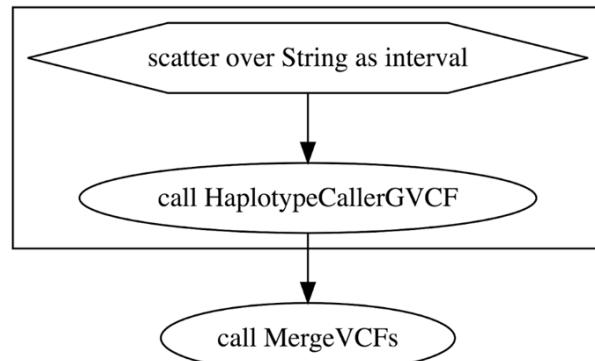
Find a Workflow



scatter-hc.data-table

V. 1
Source: Terra

scatter-hc.filepaths

V. 1
Source: Terra

SCRIPT

••

INPUTS

••

OUTPUTS

••

RUN ANALYSIS

```
1 ## This workflow runs the HaplotypeCaller tool from GATK4 in GVCF mode
2 ## on a single sample in BAM format. The execution of the HaplotypeCaller
3 ## tool is parallelized using an intervals list file. The per-interval
4 ## output GVCF files are then merged to produce a single GVCF file for
5 ## the sample, which can then be used by the joint-discovery workflow
6 ## according to the GATK Best Practices for germline short variant
7 ## discovery.
8
9 version 1.0
10
11 workflow ScatterHaplotypeCallerGVCF {
12
13     input {
14         File input_bam
15         File input_bam_index
16         File intervals_list
17     }
18
19     String output_basename = basename(input_bam, ".bam")
20
21     Array[String] calling_intervals = read_lines(intervals_list)
22
23     scatter(interval in calling_intervals) {
```

SCRIPT

INPUTS

OUTPUTS

RUN ANALYSIS

Download json | Drag or click to upload json

SEARCH INPUTS

Task name	Variable	Type	Attribute
HaplotypeCallerGVCF	docker_image	String	"broadinstitute/gatk:4.1.3.0"
HaplotypeCallerGVCF	java_opt	String	"-Xmx8G"
HaplotypeCallerGVCF	ref_dict	File	"gs://genomics-on-the-cloud/book-bundle-v0/data/germline/ref/ref.dict"
HaplotypeCallerGVCF	ref_fasta	File	"gs://genomics-on-the-cloud/book-bundle-v0/data/germline/ref/ref.fasta"

Direct
file paths

SCRIPT

INPUTS

OUTPUTS

RUN ANALYSIS

Download json | Drag or click to upload json

SEARCH INPUTS

Task name	Variable	Type	Attribute
HaplotypeCallerGVCF	docker_image	String	workspace.gatk_docker
HaplotypeCallerGVCF	java_opt	String	"-Xmx8G"
HaplotypeCallerGVCF	ref_dict	File	workspace.ref_dict
HaplotypeCallerGVCF	ref_fasta	File	workspace.ref_fasta
HaplotypeCallerGVCF	ref_index	File	workspace.ref_fasta_index
MergeVCFs	docker_image	String	workspace.gatk_docker
MergeVCFs	java_opt	String	"-Xmx8G"
ScatterHaplotypeCallerGVCF	input_bam	File	this.input_bam
ScatterHaplotypeCallerGVCF	input_bam_index	File	this.input_bam_index



References to data tables

this.input_bam**workspace.gatk_docker**

“Workspace data”

workspace.gatk_docker

Key	Value
gatk_docker	broadinstitute/gatk:4.1.3.0
intervals_list_full	snippet-intervals-full.list
intervals_list_min	snippet-intervals-min.list
ref_dict	ref.dict
ref_fasta	ref.fasta
ref_fasta_index	ref.fasta.fai

Input data

this.input_bam

<input type="checkbox"/> ▾	book_samples_id	⬇	input_bam	input_bam_index	⋮
<input type="checkbox"/>	father		father.bam	father.bai	
<input type="checkbox"/>	mother		mother.bam	mother.bai	
<input type="checkbox"/>	son		son.bam	son.bai	

SCRIPT

INPUTS

OUTPUTS

RUN ANALYSIS

Output files will be saved to

Files / submission unique ID / ScatterHaplotypeCallerGVCF / workflow unique ID

References to outputs will be written to

Tables / book_samples

Fill in the attributes below to add or update columns in your data table

Download json | Drag or click to upload.json SEARCH OUTPUTS

Task name	Variable	Type	Attribute Use defaults
ScatterHaplotypeCallerGVCF	output_gvcf	File	this.output_gvcf



TABLES +

DOWNLOAD TABLE TSV COPY TO CLIPBOARD OPEN WITH... Search 🔍 ⚙️

book_samples_id	input_bam	input_bam_index	output_gvcf
father	father.bam	father.bai	father.merged.g.vcf
mother	mother.bam	mother.bai	mother.merged.g.vcf
son	son.bam	son.bai	son.merged.g.vcf

REFERENCE DATA +

OTHER DATA

Workspace Data

Files

DASHBOARD DATA NOTEBOOKS WORKFLOWS **JOB HISTORY** ⋮

[← Back to list](#)

Workflow Statuses ⌚ Submitted: 2	Workflow Configuration dsp-comms-dev/scatter-hc.data-table	Submitted by genomics.book@gmail.com Dec 16, 2019, 5:46 PM	Total Run Cost N/A
	Data Entity scatter-hc-data-table_2019-12-16T22-45-48 book_samples_set	Submission ID adbaa401-af54-4c58-be37-bf552e69c77a	Call Caching Enabled

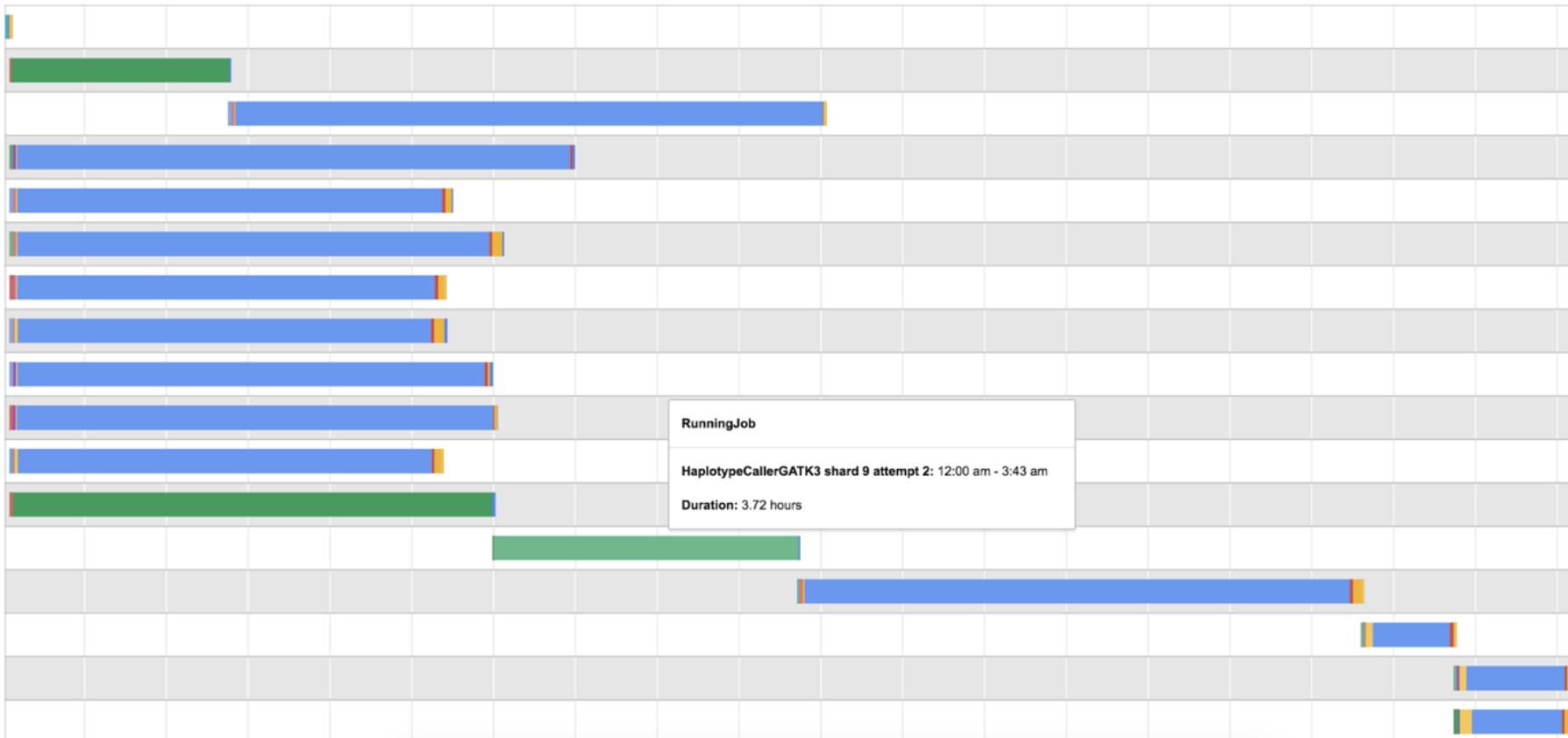
Completion status

[Queue Status](#)

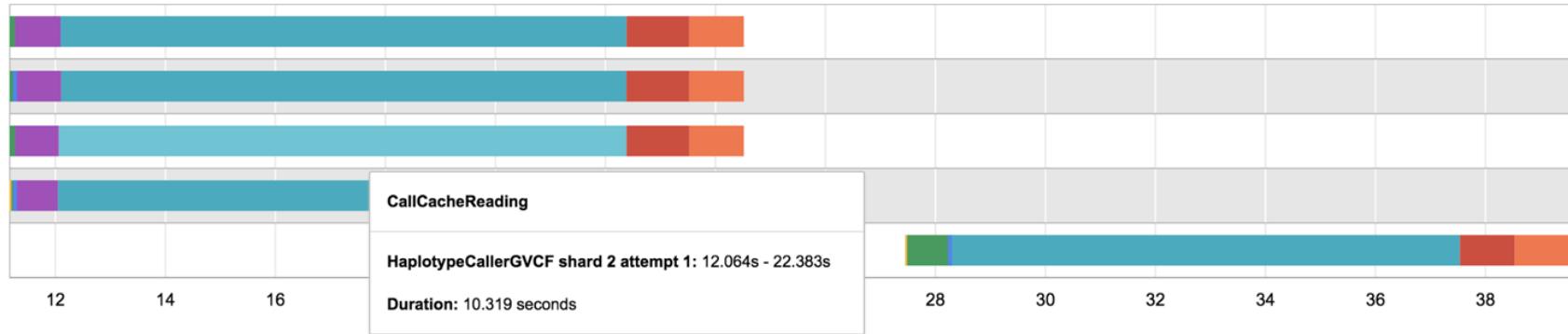
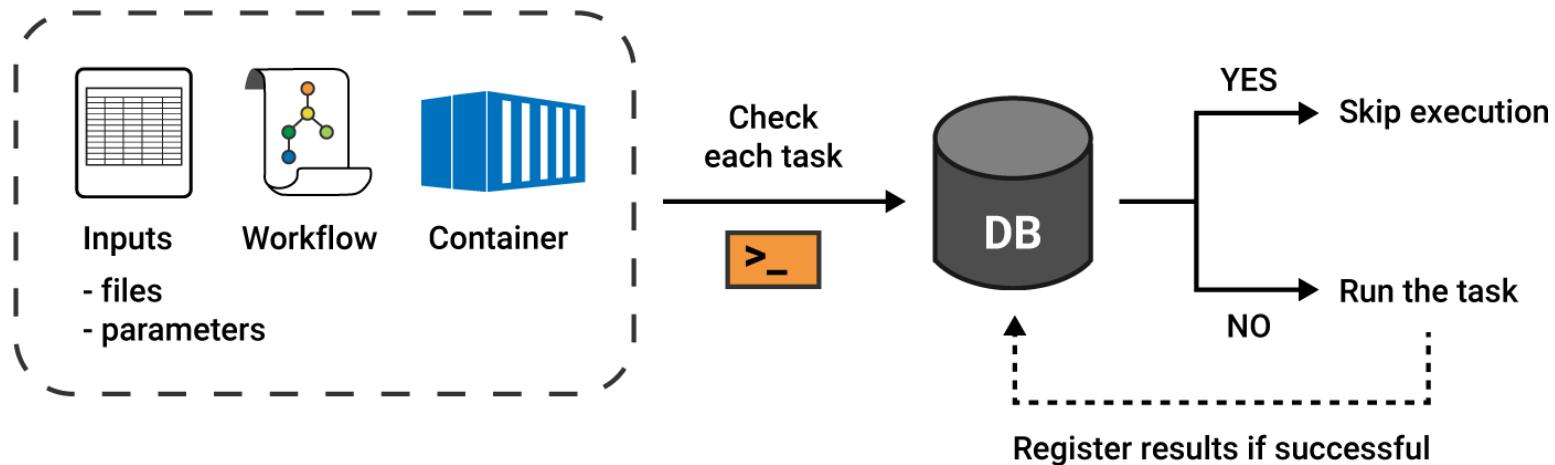
	Data Entity	Last Changed	Status	Run Cost	Messages	Workflow ID
View	father (book_samples)	Dec 16, 2019, 5:46 PM	⌚ Submitted	N/A		5a8ed577-1593-45f6-bc32-0c494a3dd638
View	mother (book_samples)	Dec 16, 2019, 5:46 PM	⌚ Submitted	N/A		b52bf87e-0b15-42fd-9f5d-b34706c5a243

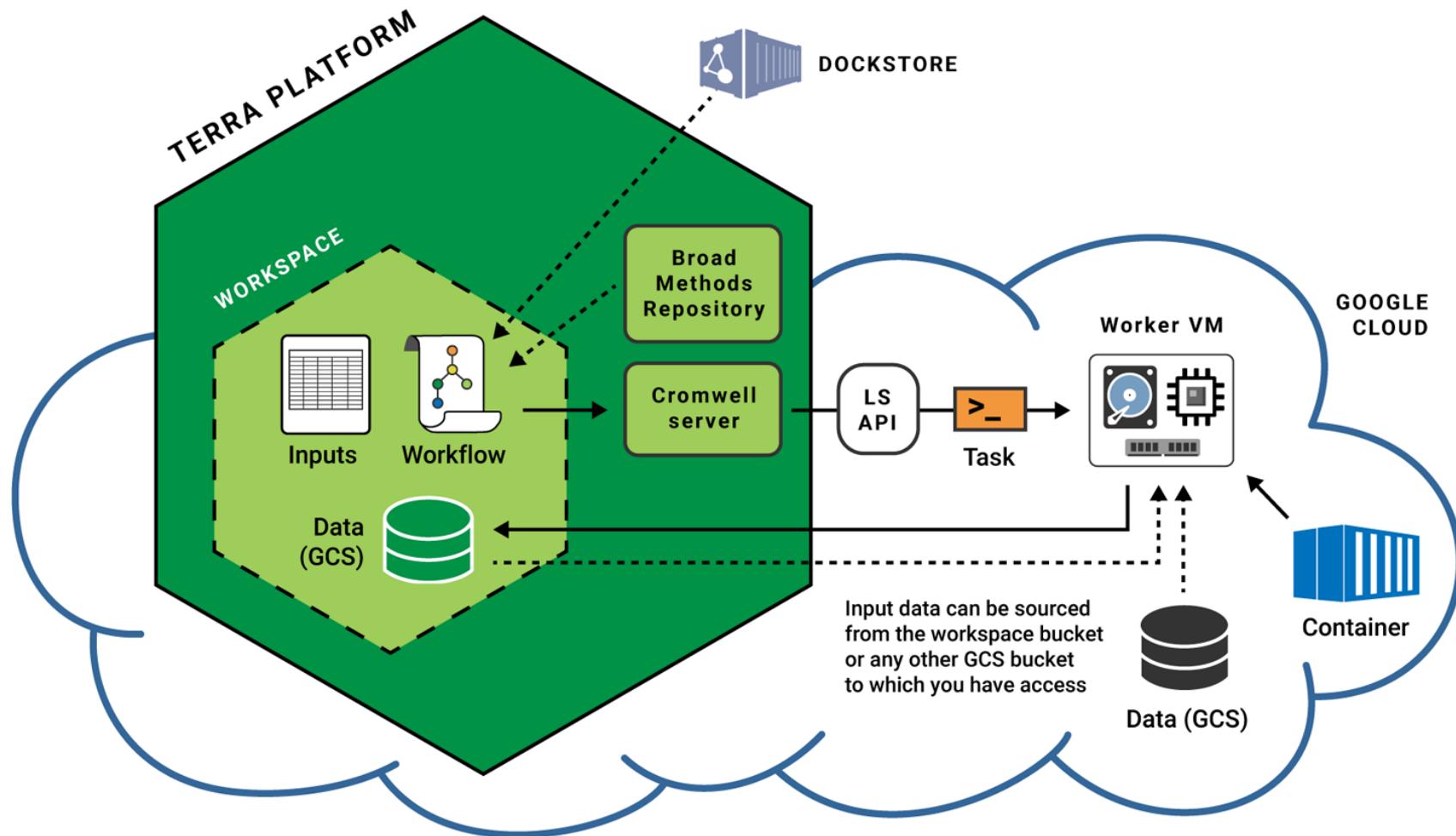
LIST VIEW INPUTS OUTPUTS LABELS TIMING DIAGRAM

Task Name	Inputs	Status	Start	Duration	Inputs	Outputs	Links	Attempts
HaplotypeCallerGVCF		✓	Today, 7:13 PM	0h 4m				
MergeVCFs		✓	Today, 7:18 PM	0h 5m				1

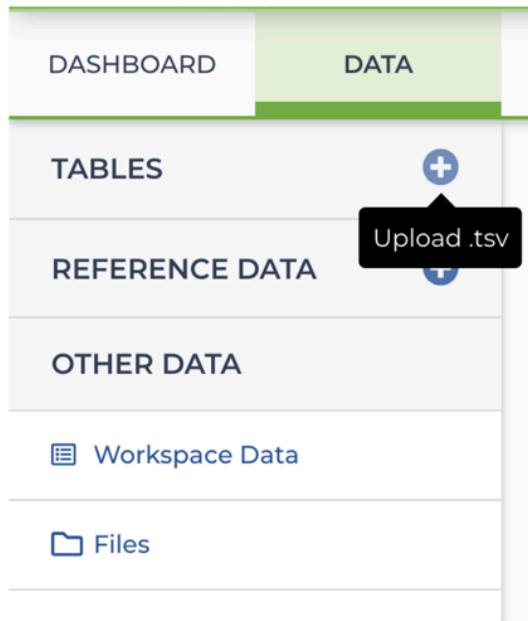
[LIST VIEW](#)[INPUTS](#)[OUTPUTS](#)[TIMING DIAGRAM](#)

Has this combination been run before?





GETTING DATA



B.

Import Table Data

Choose the data import option below. [Click here](#) for more info on the table.

FILE IMPORT **TEXT IMPORT**

Select the TSV file containing your data:

Drag or [Click](#) to select a .tsv file

Selected File: **None**

[CANCEL](#) [UPLOAD](#)

	A	B	C	D	E	F
1	entity:book_samples_id	input_bam	input_bam_index			
2	father	gs://genomics-on-the-cloud/	gs://genomics-on-the-cloud/book-bundle-v0/data/germline/bams/father.bai			
3	mother	gs://genomics-on-the-cloud/	gs://genomics-on-the-cloud/book-bundle-v0/data/germline/bams/mother.bai			
4	son	gs://genomics-on-the-cloud/	gs://genomics-on-the-cloud/book-bundle-v0/data/germline/bams/son.bai			
5						



1000 Genomes High Coverage
presented by NHGRI AnVIL

1000 Genomes project phase 3 samples sequenced to 30x coverage. This dataset is delivered as raw BAM files. You may clone this workspace to run analyses or copy specific samples to a workspace of your choice.

Participants: 2,504

BROWSE DATA



1000 Genomes Low Coverage

The 1000 Genomes Project ran between 2008 and 2013, creating the largest public catalogue of human variation and genotype data. The goal of the 1000 Genomes Project was to find most genetic variants with frequencies of at least 1% in the populations studied.

Participants: 3,500

BROWSE DATA

Copy Data to Workspace

Destination *

Select a workspace

Entries selected

SRS000030

SRS000031

WORKSPACES

Workspaces > anvil-datastorage/1000G-high-coverage-2019 > Data (read only)

DASHBOARD DATA NOTEBOOKS WORKFLOWS JOB HISTORY

TABLES		Actions	
participant (2504)	sample (2504)	Download All Rows	Copy Page to Clipboard
sample_set (1)			25 rows selected More Options
		Search	
REFERENCE DATA		Download as TSV Open with... Export to Workspace	
OTHER DATA		gVCF NA06985.gvc... NA06986.gvc... NA06994.haplotypeCalls.er.raw.gvc... NA07000.haplotypeCalls.er.raw.gvc...	

Send the selected data to another workspace [NA06985.gvc...](#)

ENCODE

6 Donors | 201 Files

EXPORT ➤Reference Genome Assembly=GRCh38 x Assay Type=ATAC-seq xSend to Terra

Donor Accession

Search...

- ENCDO000AAB
- ENCDO000AAC
- ENCDO000AAD
- ENCDO000AAE
- ENCDO000AAF
- ENCDO000AAG

Age



Age Units

Search...

- day
- month
- week
- year

Health Status

Search...

- abdominal sarcoma
- acute promyelocytic leukemia
- acute T cell leukemia
- Adenocarcinoma of cecum
- apparently healthy
- B cell lymphoma

Sex

Search...

- female
- male
- mixed

Assay Type

Search...

- ATAC-seq
- ChIA-PET
- ChIP-seq
- DNase-seq
- Hi-C
- microRNA-seq



BETA

IMPORT DATA



Importing Data

From: broad-gdr-encode.appspot.com

The dataset(s) you just chose to import to Terra will be made available to you within a workspace of your choice where you can then perform analysis.



Destination Workspace

Choose the option below that best suits your needs.



Start with an existing workspace

Select one of your workspaces



Start with a new workspace

Set up an empty workspace that you will configure for analysis



WORKSPACES

Workspaces > dsp-comms-dev/TEST-ANVIL-1234 >
Data



Notebook Runtime
RUNNING (\$0.76 hr)



DASHBOARD

DATA

NOTEBOOKS

WORKFLOWS

JOB HISTORY



TABLES



donor (6)

donor_set (1)

file (201)

file_set (1)

DOWNLOAD ALL ROWS

COPY PAGE TO CLIPBOARD

0 rows selected

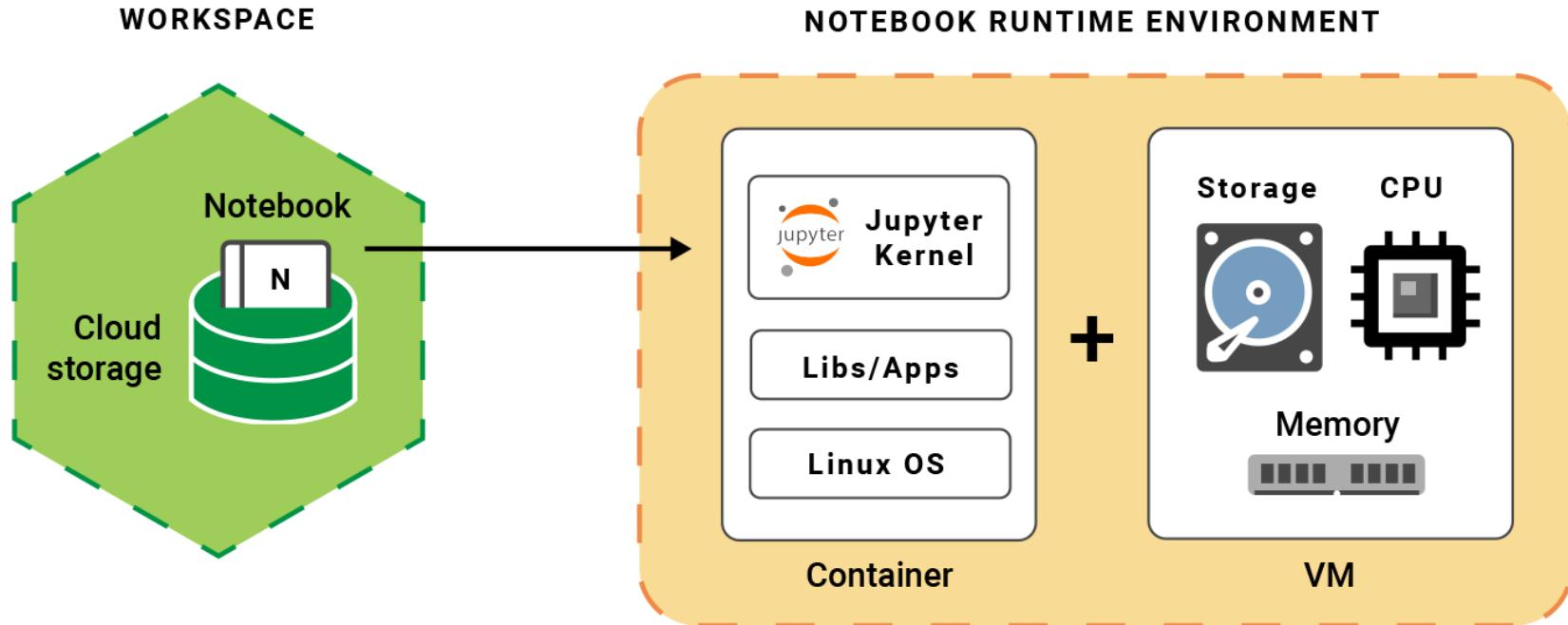
Search



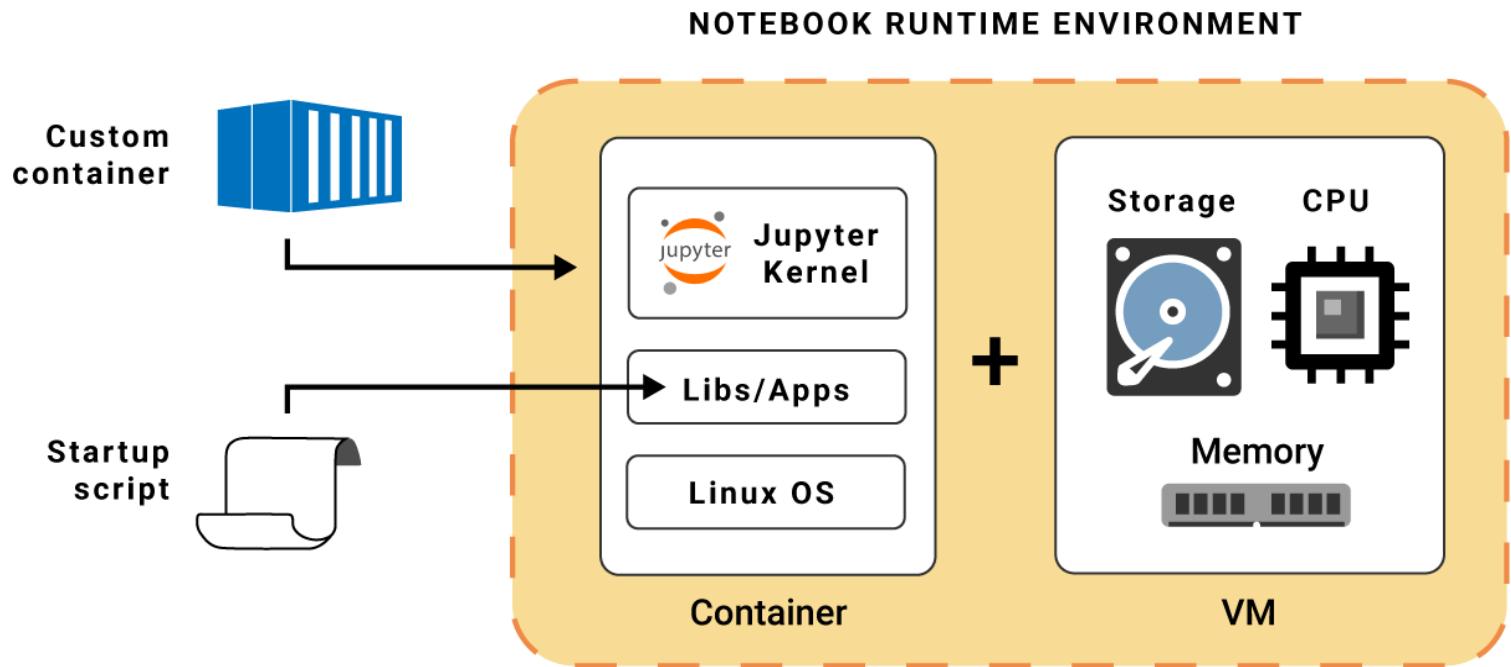
▾	file_id	assay_type	biosamples	biosample_term_id	
<input type="checkbox"/>	ENCFF013HFS	ATAC-seq	1 item	EFO:0001086	
<input type="checkbox"/>	ENCFF015FKA	ATAC-seq	1 item	UBERON:0001150	
<input type="checkbox"/>	ENCFF020COS	ATAC-seq	1 item	EFO:0001086	

INTERACTIVE ANALYSIS

(Jupyter Notebooks)



Interactive work is done in a runtime environment



Options for customizing the runtime environment

RUNTIME CONFIGURATION

Choose a Terra pre-installed runtime environment (e.g. programming languages + packages) or choose a custom environment

PRE-INSTALLED ENVIRONMENT **CUSTOM ENVIRONMENT**

Environment: Default (Python 3.6.8, R 3.5.2, Hail 0.2.11)

What's installed on this environment? Updated: Aug 25, 2019 Version: FINAL

COMPUTE POWER
Select from one of the compute runtime profiles or define your own

Profile: Default (Moderate) computer power

CPUs: 4 Memory (GB): 15 Disk size (GB): 50

Cost: \$0.19 per hour

CANCEL **CREATE**



INSTALLED PACKAGES

Default (Python 3.6.8, R 3.5.2, Hail 0.2.11)

Updated: Aug 25, 2019
Version: FINAL

Installed packages	Python
Package	Python
python	3.6.8
hail	0.2
wrapt	1.11.2
absl-py	0.7.1
arrow	0.14.5



COMPUTE POWER
Select from one of the runtime profiles or define your own

Profile: Custom

CPUs: 4 Memory (GB): 15 Disk size (GB): 50

Startup script: gs://genomics-on-the-cloud/book-bundle-v0/code/notebooks/insta

Configure as Spark cluster

COST: \$0.19 per hour

Terra BETA **WORKSPACES**

Workspaces > broad-firecloud-dsp-pm/20200120 genomics-book-10 > Notebooks

DASHBOARD DATA NOTEBOOKS WORKFLOWS JOB HISTORY

SEARCH NOTEBOOKS Sort By: Most Recently Updated

NOTEBOOKS

Create a New Notebook +

whatever Last edited: Today

GATK-tutorial-notebook-demo Last edited: Today

Genomics-Notebook Last edited: Today

Drag or Click to Add an ipynb File

SEARCH NOTEBOOKS Sort By: Most Recently Updated

NOTEBOOKS

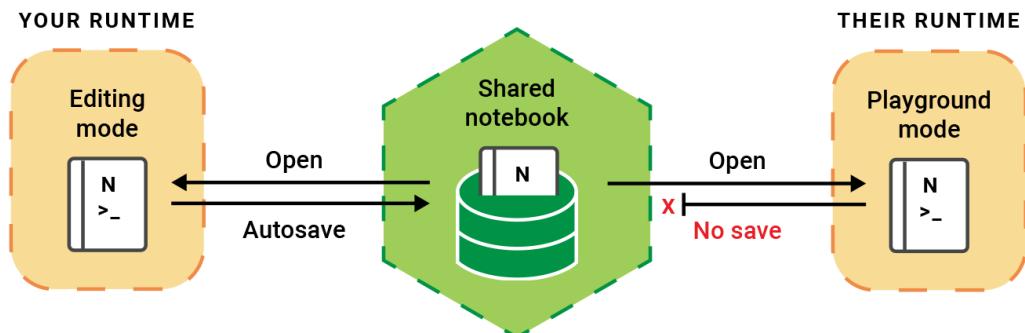
Create a New Notebook +

whatever Last edited: Today

GATK-tutorial-notebook-demo Last edited: Today

Genomics-Notebook Last edited: Today

Drag or Click to Add an ipynb File





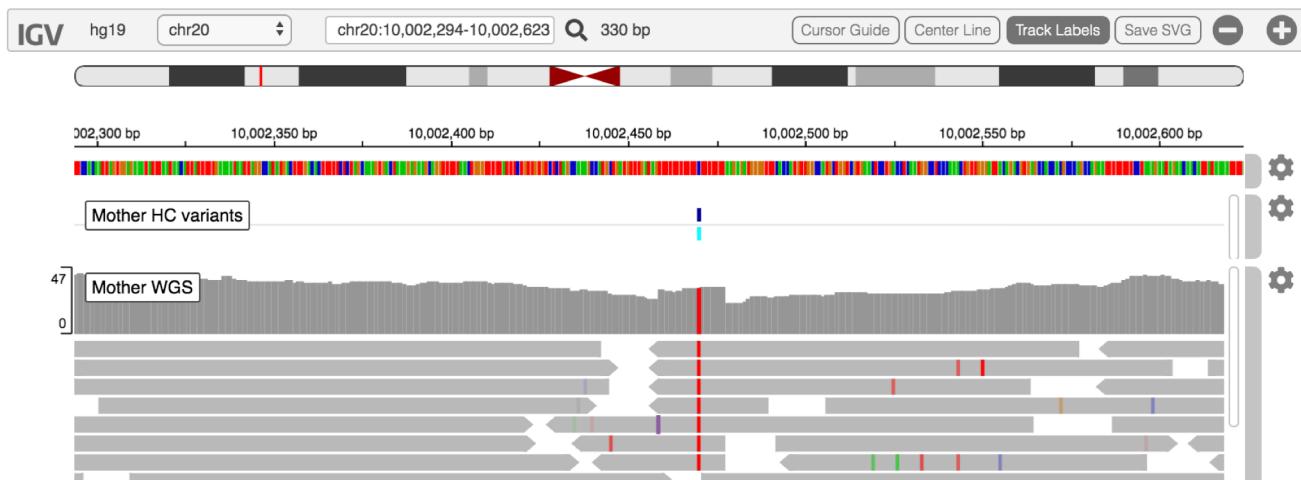
jupyter Genomics-Notebook (autosaved)

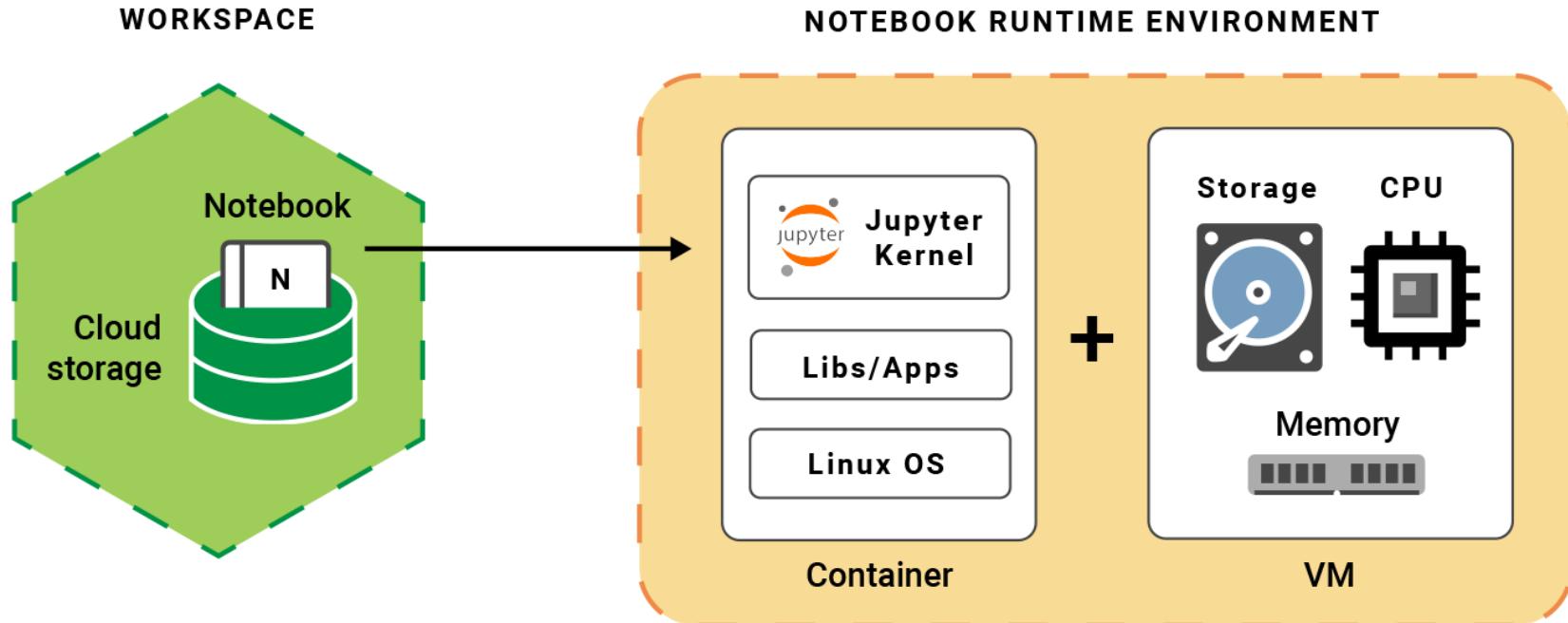


File Edit View Insert Cell Kernel Navigate Widgets Help

Not Trusted Edit Mode Python 3

```
In [21]: # Create an interactive IGV browser with genome reference and coordinates specified.  
IGV_InspectCalls = igv.Browser(  
    {"genome": "hg19",  
     "locus": "chr20:10,002,294-10,002,623"  
    }  
)  
  
# Tell Python to display it below  
IGV_InspectCalls.show()
```





Coming next: other apps for interactive analysis

**Connecting scientists to the patients,
datasets, and tools they need to do
life-changing research**

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(patients)



Data ingest
(hospitals and labs)



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