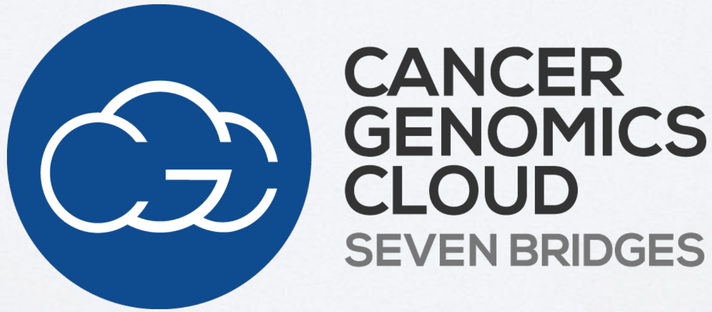
Cancer Genomics Cloud & R: Find, Access, and Analyze Petabyte-Scale Cancer Genomic Data on the Cloud

Nan Xiao
Seven Bridges



TCGA IS A TREMENDOUS GIFT TO THE CANCER RESEARCH COMMUNITY …

More than 11,000 cases representing 33 cancer types

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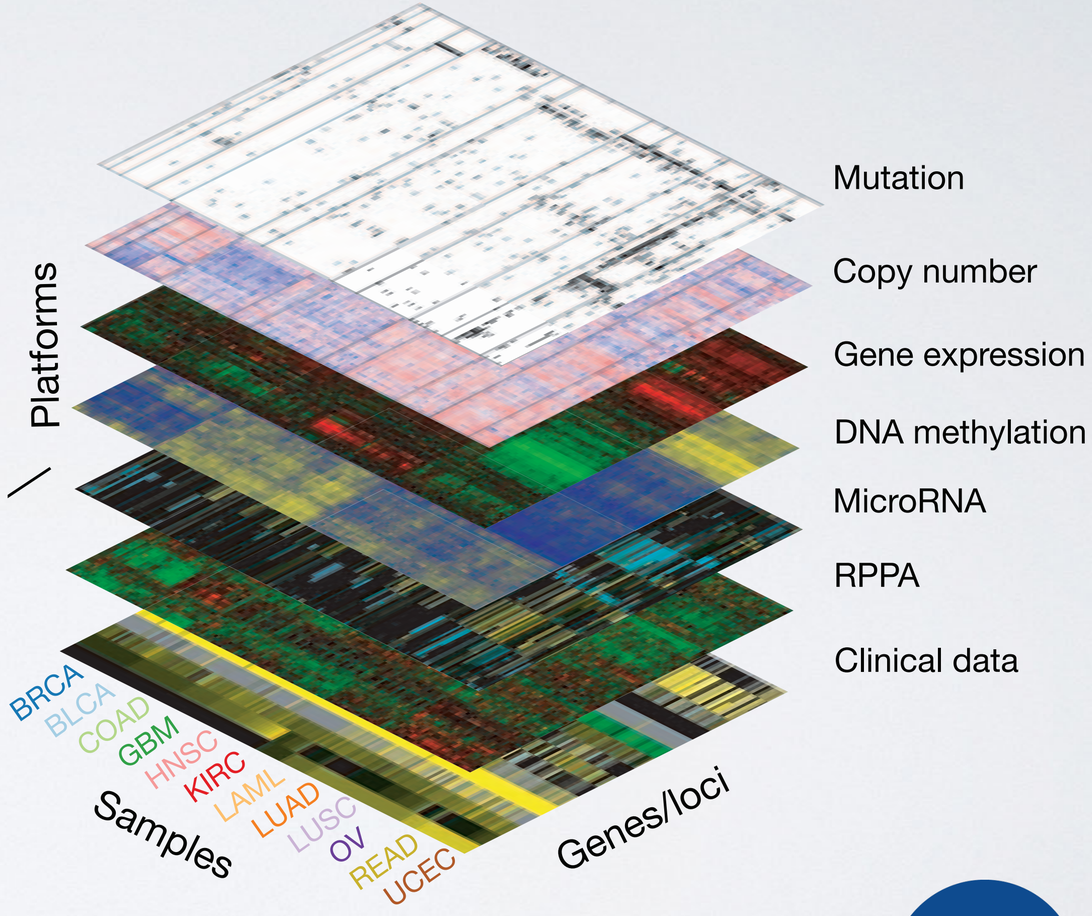
UNDERSTANDING TCGA DATASET

- Multiple Samples per Case
- Multiple Analyses per Sample
- Rich metadata: barcodes, UUIDs, XMLs...



Nature Genetics 45, 1113-1120 (2013)

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As the amount and diversity of data increases, it becomes more difficult to learn from them.

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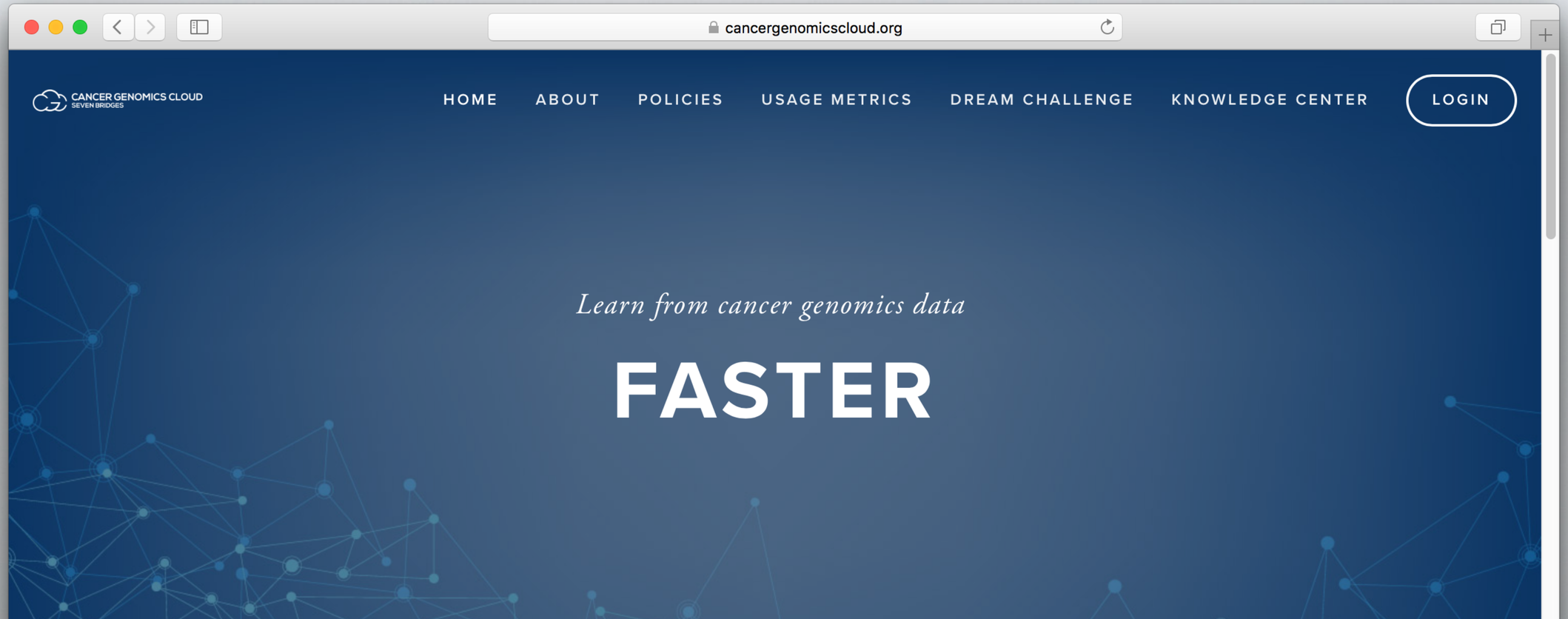
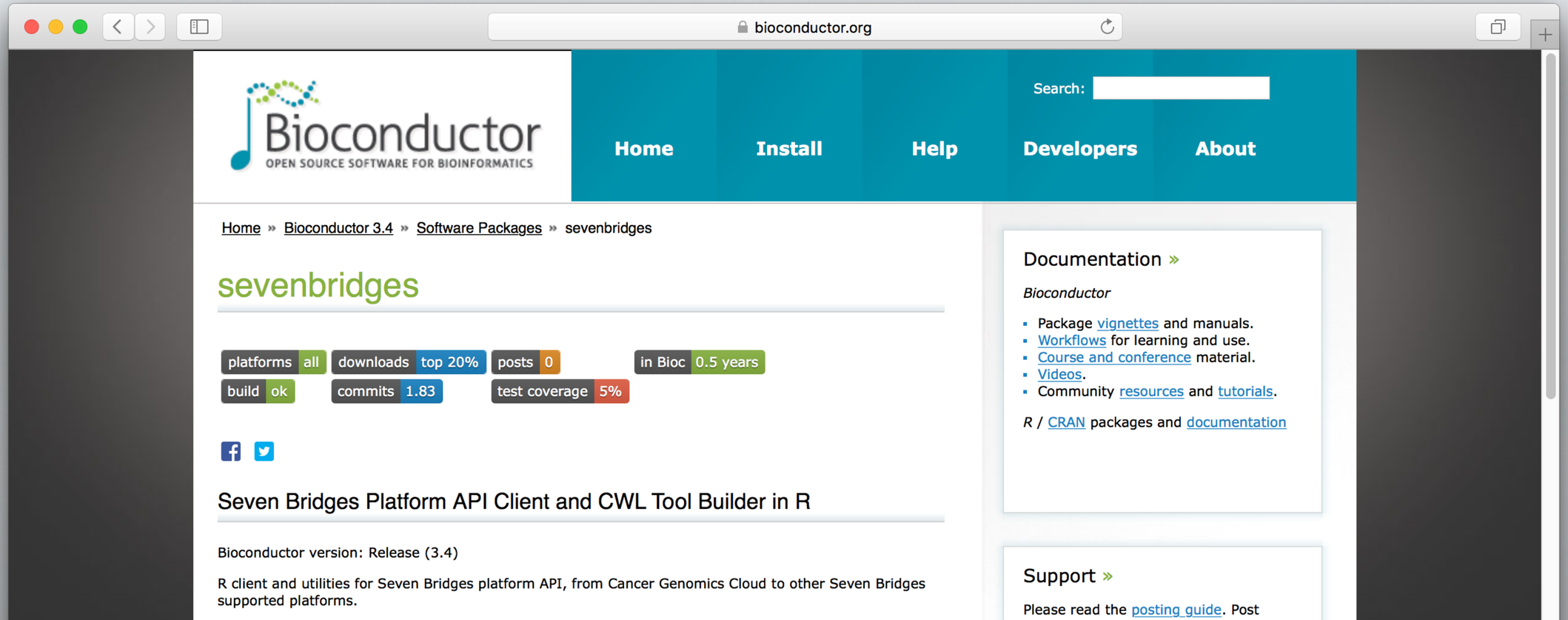
The CGC aims to provide a collaborative environment where researchers can take advantage of co-localized public data (like TCGA) and public tools; but also recombine these with their private data and tools.

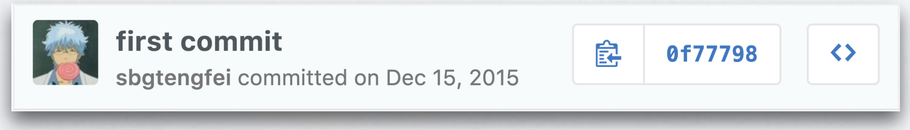


`sevenbridges` is an R/Bioconductor package offering easier programmatic access to CGC.



The `sevenbridges` package offers:

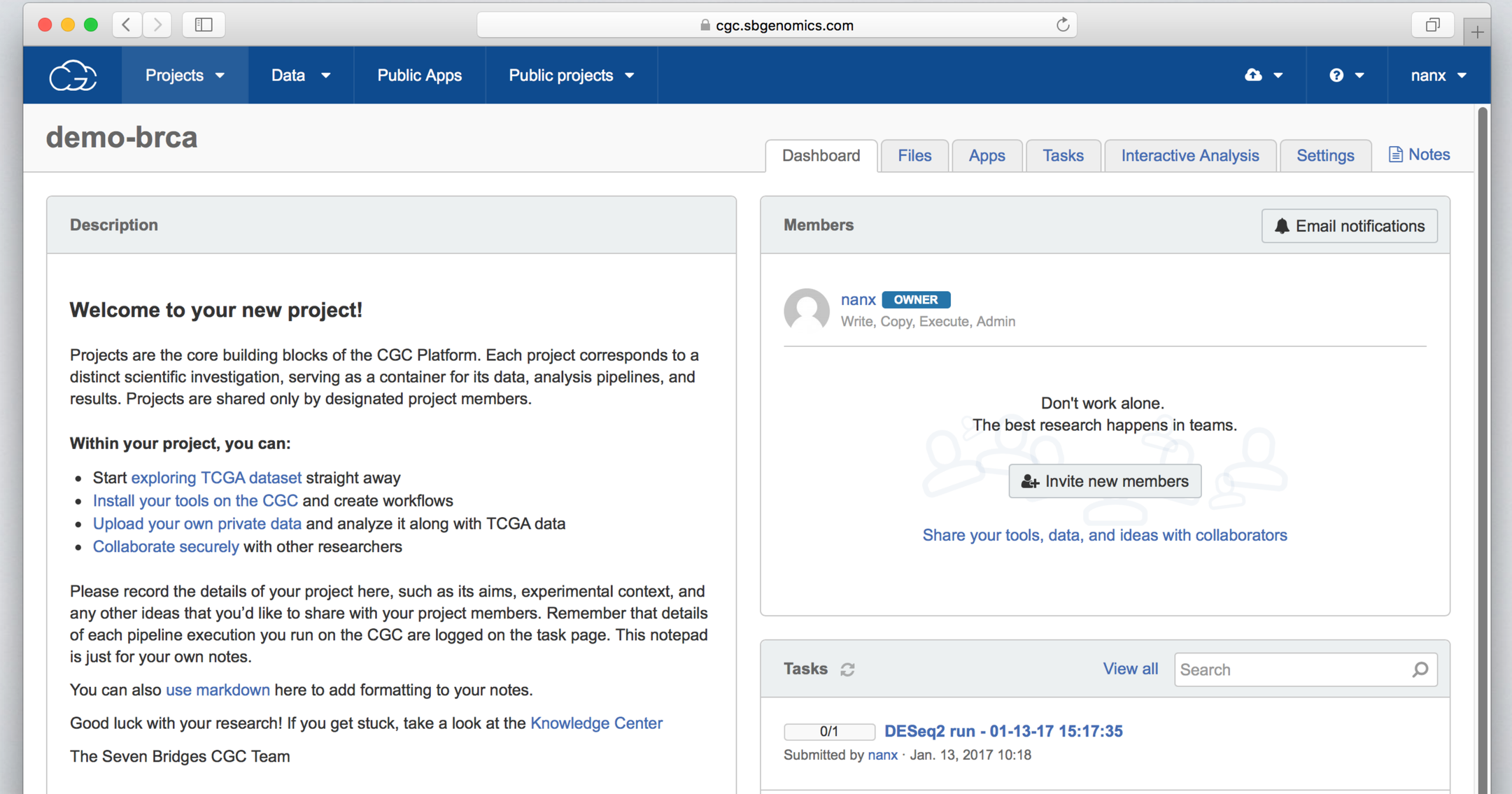
- Complete R API client for CGC/Seven Bridges API
- Common Workflow Language Tool Interface
- Task monitoring / Batch tasks support
- and many helper functions/utilities in R.



2015

2017

PROJECT MANAGEMENT

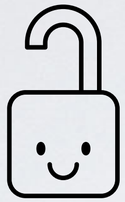


PROJECT MANAGEMENT VIA API

```
library("sevenbridges")

# Create Auth object
a = Auth(platform = "cgc", token = “your_token")

# List project and details
a$project(owner  = "user")
a$project(detail = TRUE)

# Get billing group ID
bid = a$billing()$id

# Create new project
a$project_new("brca_test", bid,
              description = "BRCA Test")

# Add member to project
m = a$project(id = "demo/brca_test")$
      member_add(username = "new_user")
```

FIND DATA ON CGC

MORE THAN ONE PETABYTE OF TCGA DATA AT YOUR FINGERTIPS



Open Data

Information NOT unique to an individual.

- de-identified clinical data
- gene expression data
- copy number alterations
- epigenetic data

Controlled Data

Information that IS unique to an individual.

- primary sequencing data
- raw & processed SNP6 array data
- raw exon array data
- mutation calls for an individual

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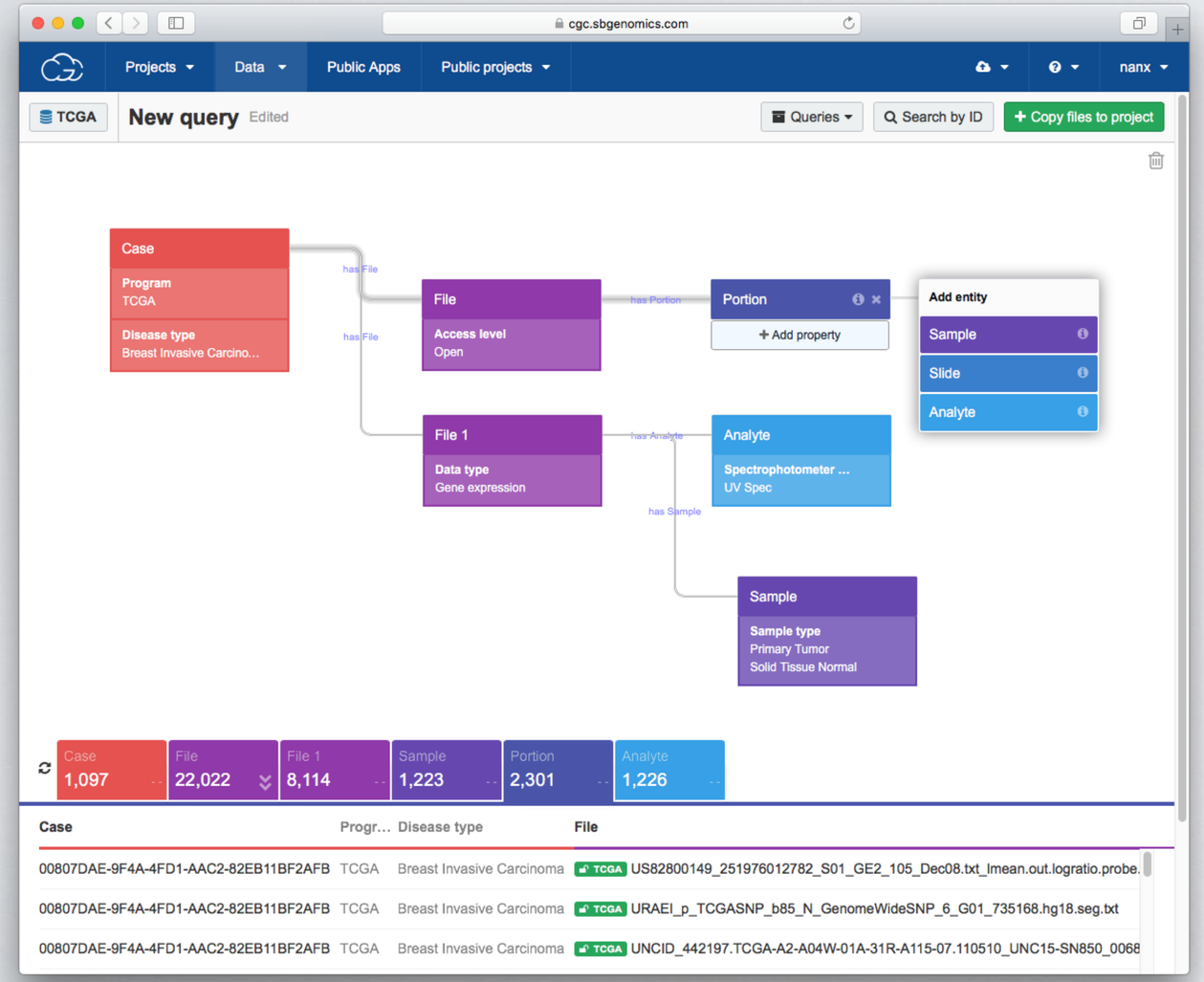
CGC DATA BROWSER



- Query metadata
- Add files to project

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SPARQL QUERY

```
library("SPARQL")
endpoint = "https://opensparql.sbgenomics.com/blazegraph/namespace/tcga_metadata_kb/sparql"
query = "
prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#>
prefix tcga: <https://www.sbgenomics.com/ontologies/2014/11/tcga#>

select distinct ?case ?sample ?file_name ?path ?xs_label ?subtype_label
where
{
 ?case a tcga:Case .
 ?case tcga:hasDiseaseType ?disease_type .
 ?disease_type rdfs:label 'Lung Adenocarcinoma' .

 ...

}
"
qd = SPARQL(endpoint, query)
df = qd$results
head(df)
```

DATASETS API

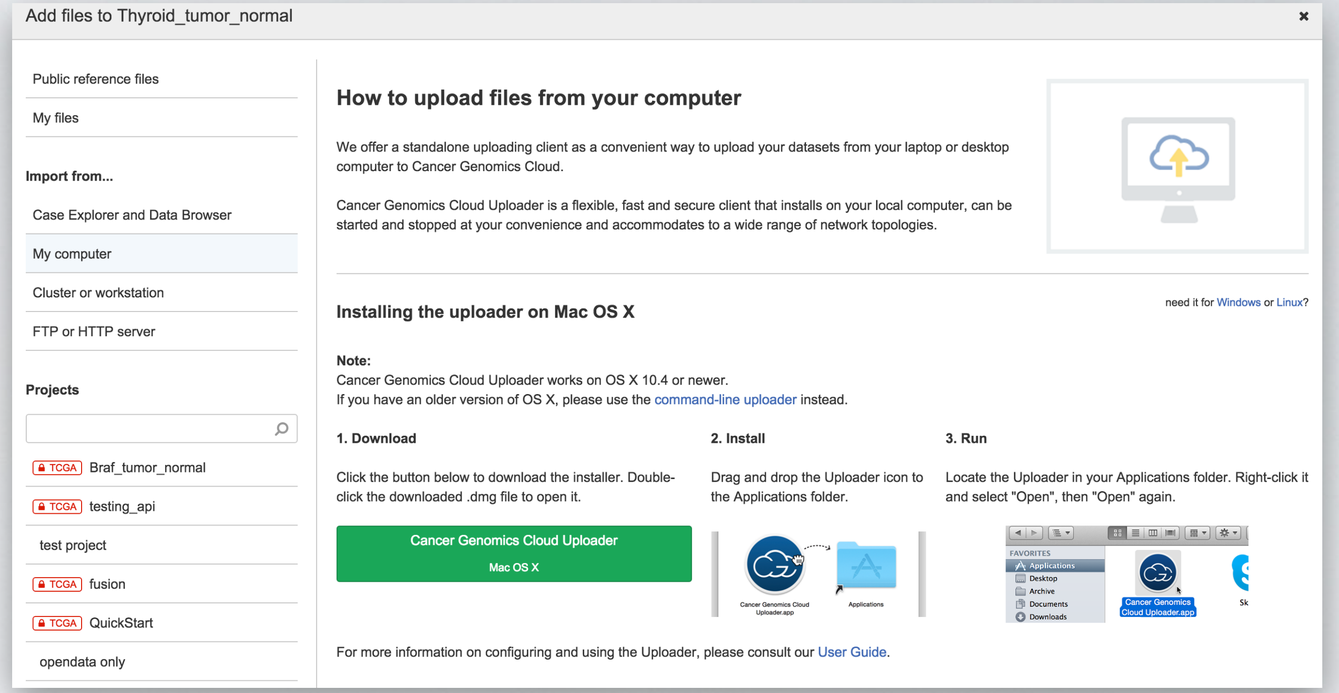
```
term = list(
    "entity" = "cases",
    "hasSample" = list(
        "hasSampleType" = "Primary Tumor",
        "hasPortion" = list(
            "hasPortionNumber" = 11
            )
        ),
    "hasNewTumorEvent" = list(
        "hasNewTumorAnatomicSite" = c("Liver", "Pancreas"),
        "hasNewTumorEventType" = list(
            "filter" = list(
                "contains" = "Recurrence"
            )
        )
    )
)

a$api(path = "query", body = term, method = "POST")
```

ADD & ANNOTATE DATA

UPLOAD YOUR DATA

- CGC Uploader (GUI)
- Command Line Uploader
- FTP / HTTP / S3 Volume
- API



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UPLOAD A FILE VIA API

```
myfile = "file_path.fastq"
p = a$project(id = "demo/tcga-demo")

# Load `.meta` for the file by default
p$upload(myfile, overwrite = TRUE)

# Pass metadata manually
p$upload(myfile, overwrite = TRUE,
         metadata = list(library_id = "test_id",
                         platform   = "Illumina x11"))

# Rename file
p$upload(myfile, overwrite = TRUE,
         metadata = list(library_id = "new_id")
         name = "sample_new_name.fastq")
```

UPLOAD FILES / FOLDERS

```r
# Upload a folder

dirpath = "path_to_dir"
list.files(dirpath)
p$upload(dirpath, overwrite = TRUE)


# Upload a list of files

dirpath = "path_to_dir"
myfiles = list.files(dirpath, recursive = TRUE, full.names = TRUE)
p$upload(myfiles, overwrite = TRUE)
```

ANNOTATE FILES VIA API

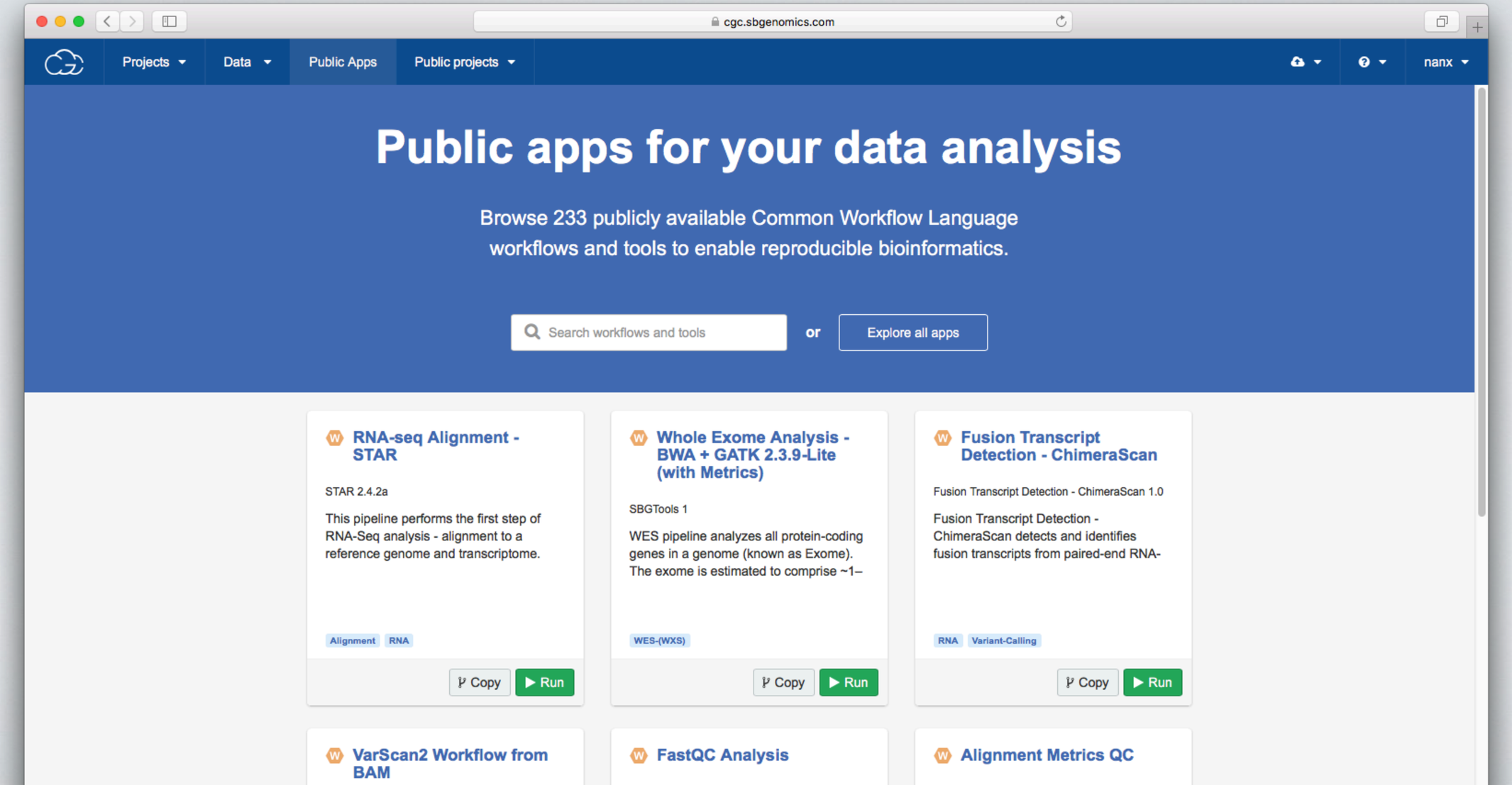
```
# Locate a sample BAM file in project
p  = a$project(id = "demo/tcga-demo")
fl = p$file("sample.bam", exact = TRUE)

# Show tags for single file
fl$tag()

# Add new tags
fl$add_tag("new year new tag")

# Set tags to overwrite existing
x = list("this", "is", 2017)
fl$set_tag(x)

# Set metadata
fl$meta()
fl$set_meta()
```

UPLOAD MANIFEST FILE (DEFINES METADATA OF FILES)

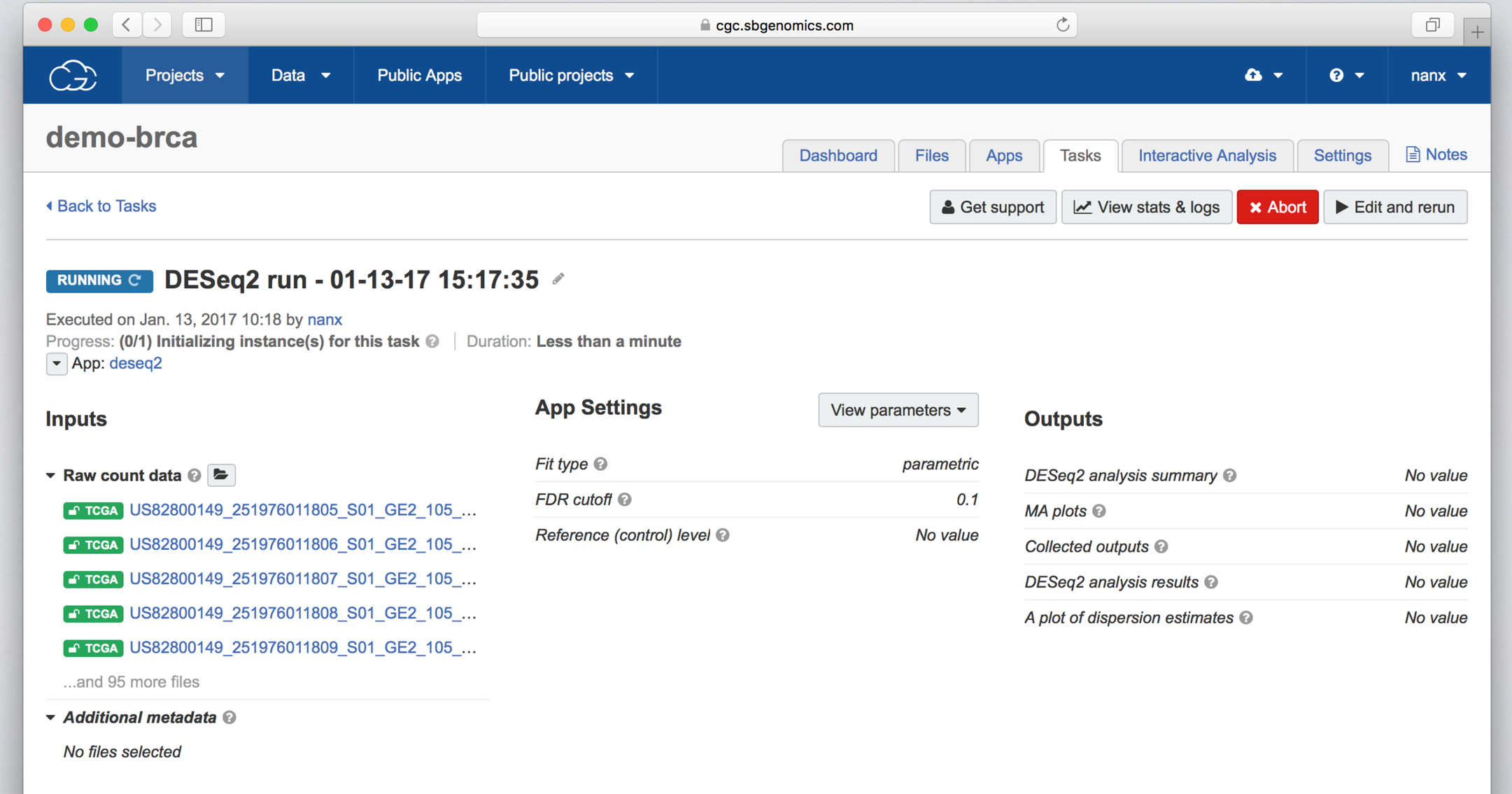
```
# attach all metadata except "bad_field" and "sample_id"

p$upload(manifest_file = "~/manifest.csv",
         overwrite     = TRUE,
         subset        = score < 0.5,
         select        = -c(bad_field, sample_id))
```

ANALYZE THE DATA

Run an analysis immediately, with ~230 tools and workflows on the CGC today.



RUNNING ANALYSIS TASKS



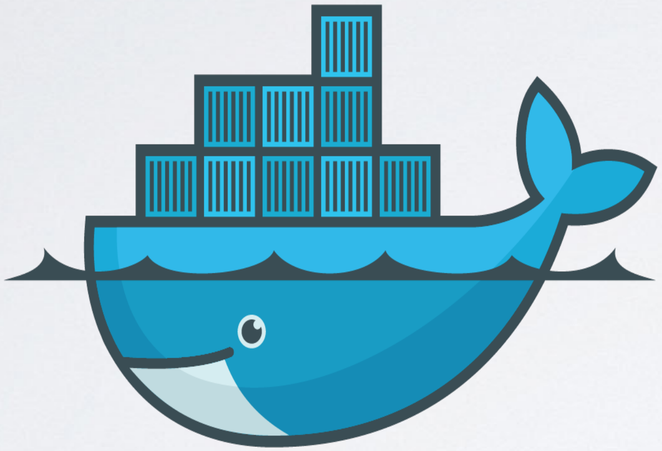
COPY APPS & RUN TASKS

```
# List public apps
a$app(visibility = "public")

# Copy an app to project
aid = a$public_app()[[1]]$id
a$copy_app(aid, project = pid, name = "new_app_name")

# Add & run new tasks
tsk = p$task_add(name        = "new_task",
                 description = "new task",
                 app         = "demo/tcga-demo/rna-seq-alignment-star/0",
                 inputs      = list(...))
tsk$run()
tsk$abort()
```

RUN TASKS IN BATCH MODE

```
# Batch by items
tsk = p$task_add(name        = "RNA DE Report Batch 1",
                 description = "RNA DE Analysis Report",
                 app         = rna_app$id,
                 batch       = batch(input = "bamfiles"),
                 inputs      = list(...))

# Batch by metadata
tsk = p$task_add(name        = "RNA DE Report Batch 2",
                 description = "RNA DE Analysis Report",
                 app         = rna_app$id,
                 batch       = batch(input = "fastq",
                                     c("metadata.sample_id",
                                       "metadata.library_id")),
                 inputs = list(...))
```

CONTAINERS + CWL MAKES IT EASY TO PUT NEW TOOLS ON THE CGC… AND OTHER PLACES.



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COMMON WORKFLOW LANGUAGE

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COMMON WORKFLOW LANGUAGE

Specification for describing scalable, portable and reproducible computational workflows.

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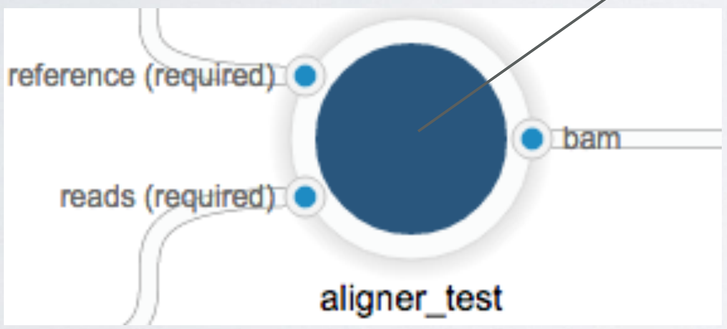
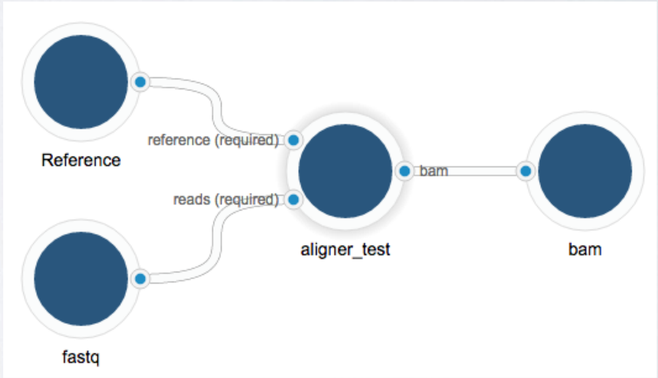
FOR DEVELOPERS, THIS MEANS…

- Easier and faster to deploy your tools.
- Write once, runs everywhere, regardless of operating system/infrastructure.

CWL TOOL: INPUTS, OUTPUTS, AND PARAMETERS




```
"softwareDescription": {
  "name": "myaligner",
  "description": "Aligns reads to a reference"
},
"documentAuthor": "kaushikghose@sbgenomics.com",
"requirements": {
  "environment": {
    "container": {
      "type": "docker",
      "uri": "",
      "imageId": ""
    }
  },
  "resources": {
    "cpu": 0,
    "mem": 5000,
    "ports": [],
    "diskSpace": 0,
    "network": false
  }
},
```



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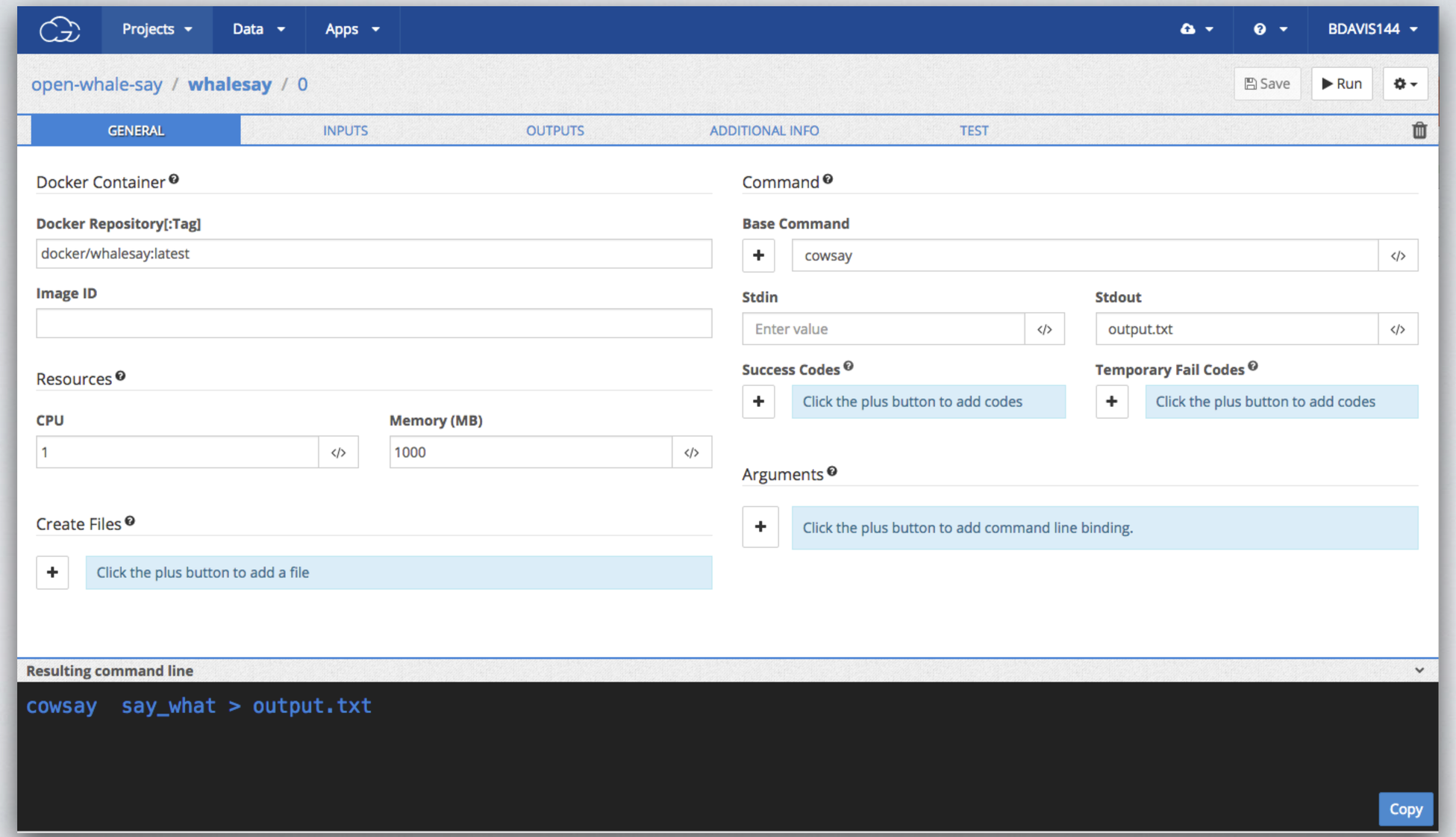
WRITE CWL WITH WEB IDE



BUILD CWL TOOLS WITH R

```
# Create CWL Tool
runif = Tool(id          = "runif",
             label       = "runif",
             hints       = requirements(docker(pull = "rocker/r-base")),
             baseCommand = "Rscript runif.R",
             stdout      = "output.txt",
             outputs     = output(id = "random", glob = "*.txt"))

# Convert to JSON or YAML
runif$toJSON()
runif$toYAML()
```

GETTING STARTED

- Try `sevenbridges` package from Bioconductor
- `docker pull sevenbridges/sevenbridges-r`
- Pull requests: github.com/sbg/sevenbridges-r

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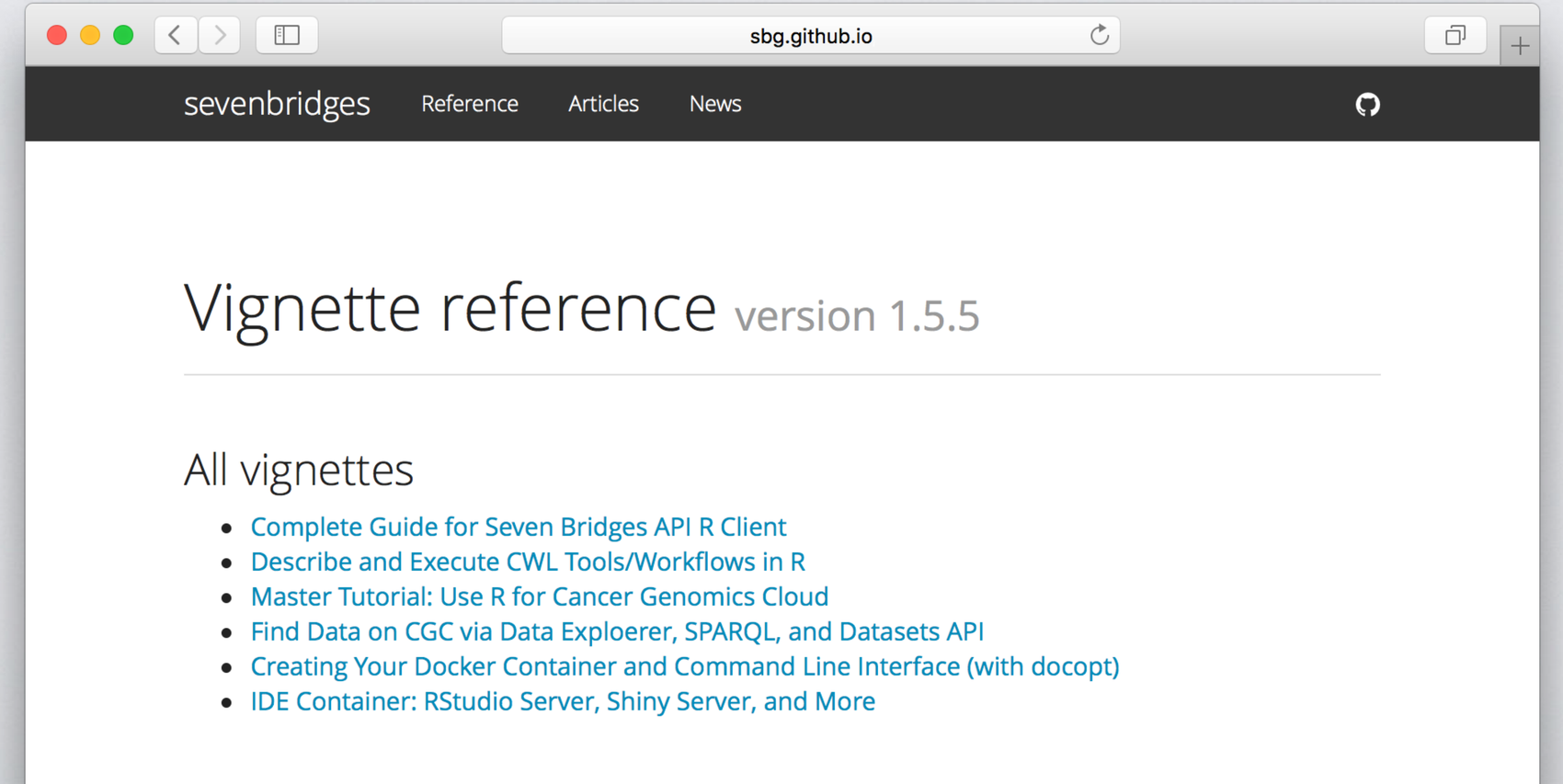
DOCUMENTATION

https://sbg.github.io/sevenbridges-r/



NEED MORE RESOURCES?

- Apply to NIHCommons Credits Pilot for new opportunities in cloud computing & storage before Jan 16:
- https://datascience.nih.gov/commons
- https://www.commons-credit-portal.org/

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MORE ON CANCER GENOMICS CLOUD AND SEVEN BRIDGES PLATFORMS

- www.cancergenomicscloud.org
- www.sevenbridges.com/tcga
- www.sevenbridges.com

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