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NCI Cancer Genomics Cloud Pilots (and Genomic Data Commons)

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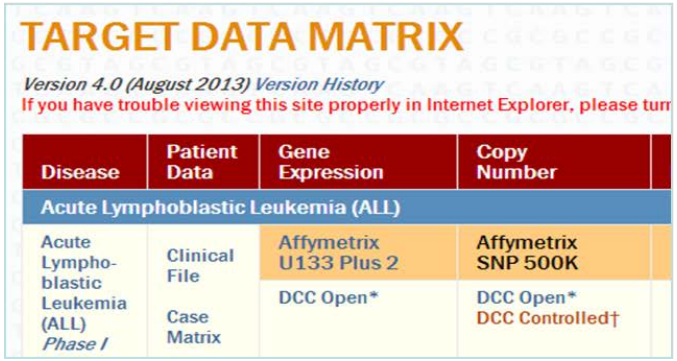
May 12, 2015

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
National Institutes of Health

Center For Cancer Genomics (CCG) Genomics Data Commons (GDC)

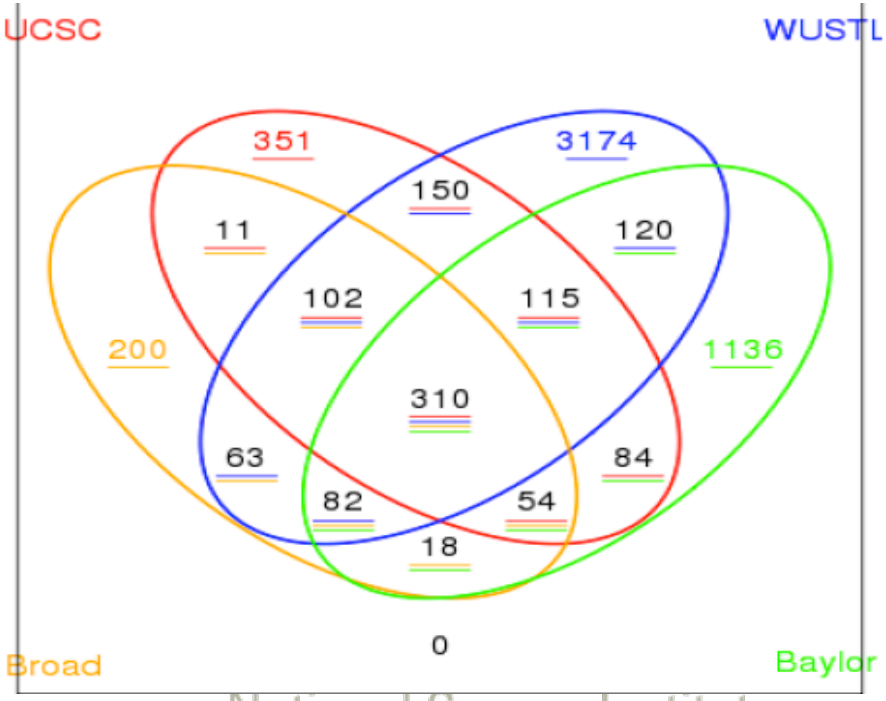
- Goal to unify fragmentary repositories at NCI
- TCGA, TARGET and CGCI have their own data repositories (DCCs)
- Sequencing data: BAM files at CGhub while VCF/MAF files at DCC



Genomics Data Commons (GDC)

- Harmonize diverse standards
- BAMs aligned to various references
- Mutations are called by various tools

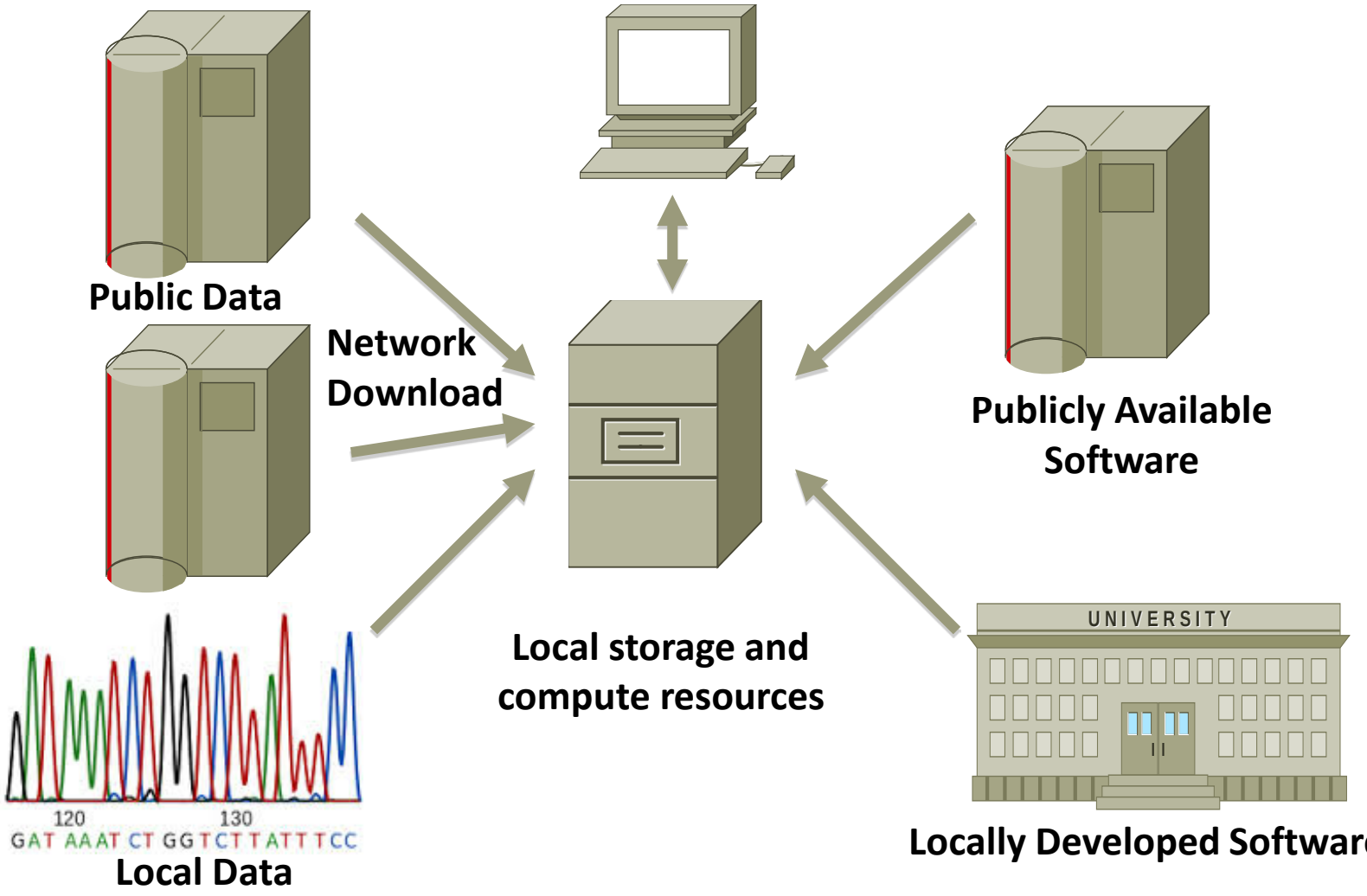
NCBI-human-build36
NCBI36_BCCAGSC_variant
NCBI36_BCM_variant
NCBI36_WUGSC_variant
HG18
HG18_Broad_variant
GRCh37
GRCh37-lite
GRCh37_BI_Variant
GRCh37-lite-+-HPV_Redux-build
HG19
HG19_Broad_variant



Genomics Data Commons (GDC)

- University of Chicago, PI: Dr. Robert Grossman
- Go live date: Late Spring 2016
- Not a commercial cloud: Free to download data

Standard Model of Computational Analysis



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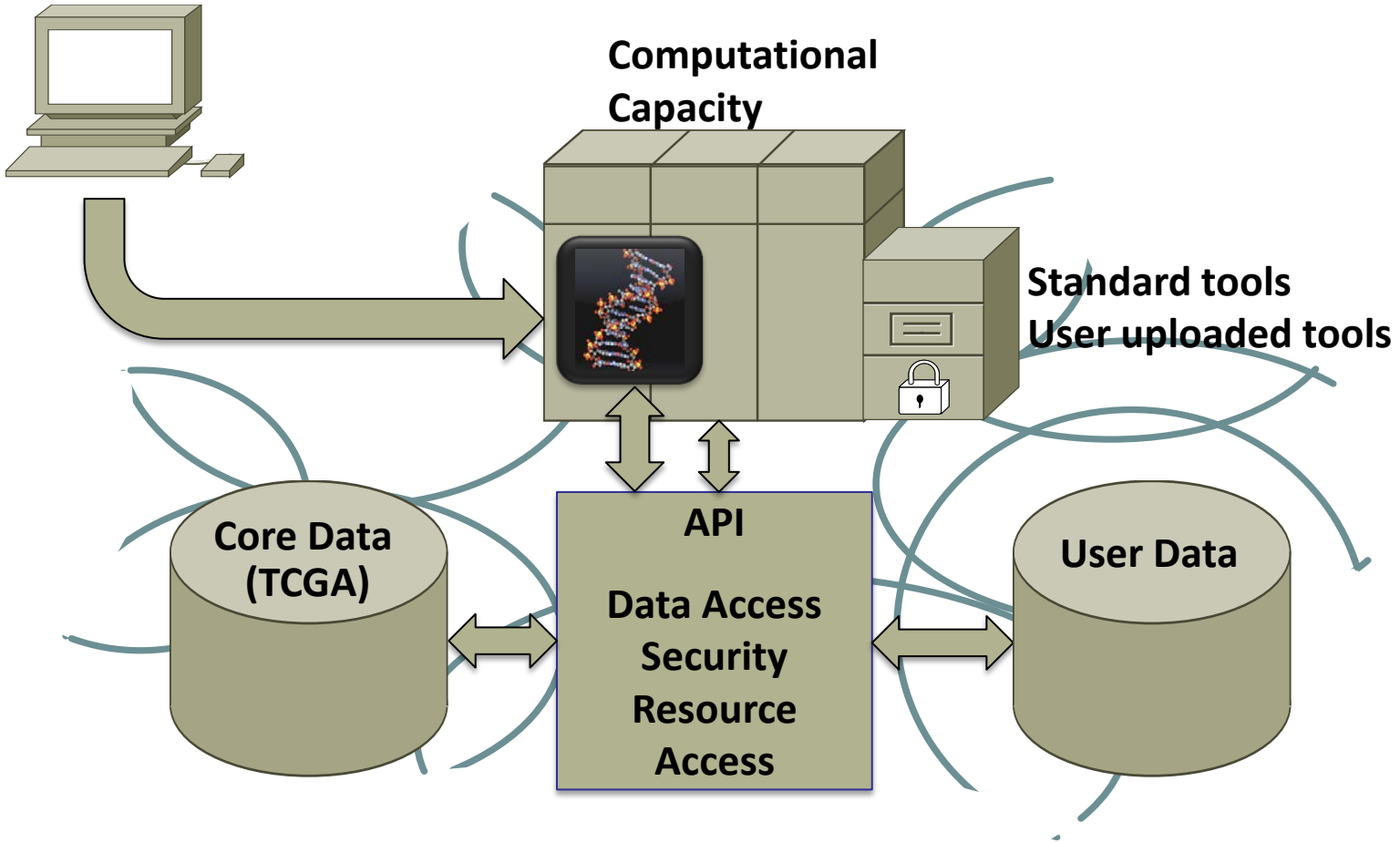
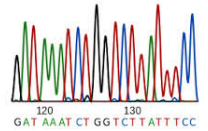
Limitations of the standard model for large data

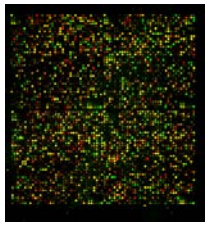
- Assuming the 2.5 PB TCGA data set
- Storage and Data Protection cost approximately $2,000,000 per year
- Downloading TCGA data at 10 Gb/sec = 23 days
- Only large institutions have the ability to utilize this data
- These datatypes will continue to grow

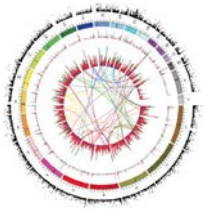
Co-located Compute & Data

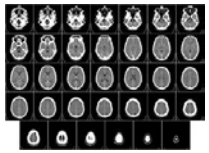


The Cloud Pilots in Context









Data Coordinating Center

QA/QC Validation

Aggregation

Authoritative NCI Reference Data Set

NCI Clouds

High Performance Computing

Analysis

Search/Retrieve

Download

NCI Genomic Data Commons

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Project Structure

Effort to democratize access to NCI genomics data

Managed through CBIIT in partnership with the Center for Cancer Genomics (CCG)

- Coordinating with the Genomic Data Commons (GDC)

Three contracts awarded to

- Broad Institute
- Institute for Systems Biology
- Seven Bridges Genomics

Period of performance: Sept 2014 – Sept 2016

- https://cbiit.nci.nih.gov/ncip/nci-cancer-genomics-cloud-pilots
- Anticipated launch date: January 2016

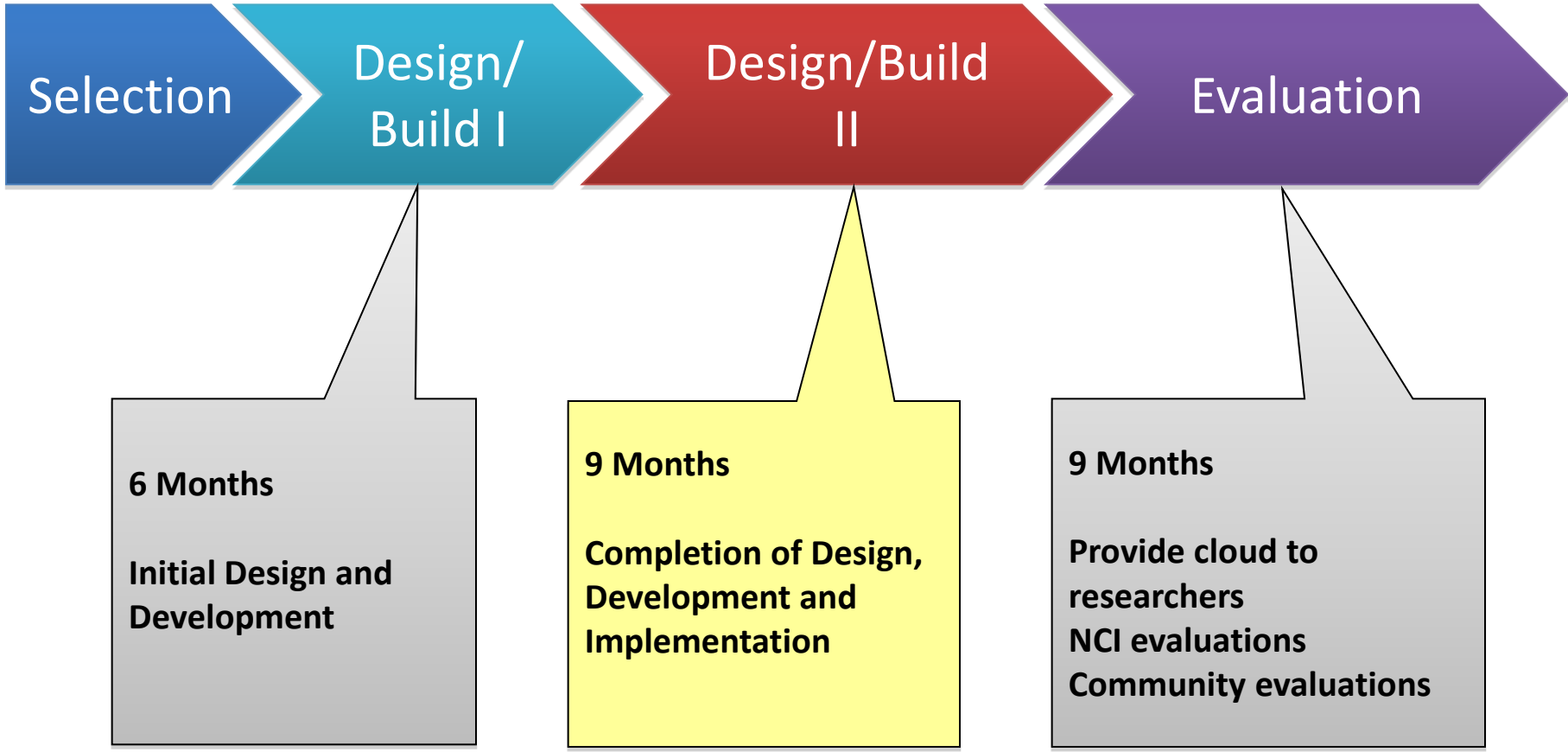
Considerations

- Design
  - Designs must be released under a non-viral, open source license
- Extensibility
  - Initial clouds will focus on a set of “core datatypes”
  - Extend to additional datatypes without major refactoring of the existing system
- Sustainability
  - Cost assessments for operating at current scale and at 10/100 fold increases in storage, compute and usage
- Security
  - FISMA moderate system, FedRAMP certified cloud provider, Trusted Partnership
  - Open v/s Controlled access data

Research and Technical Objectives

- Core Data
  - All three awardees will host a common core data set from TCGA
    - DNA-Seq binary alignment (BAM) files
    - RNA-Seq FASTQ and BAM files
    - SNP array (.cel) files
    - Somatic and germline mutation calls for each sample (.vcf, .maf)
    - Clinical data
  - Each awardee will include at least one additional TCGA data set
    - Broad: validation BAMs, miRNAseq, and methyl-seq
    - ISB: miRNAseq, and all L3 data (mRNA/miRNA expression, copy-number, DNA methylation, protein RPPA)
    - Seven Bridges: whole genome and exome DNA-Seq FASTQ, miRNAseq data, and methyl-seq

Project Schedule and Deliverables



Common to all three Cloud Pilots

- Core datasets
- Use Cases
  - Running preloaded pipeline on TCGA data
  - Uploading and processing user data
  - Uploading and running custom algorithms
  - Serve both biologists and bioinformaticians
- Workflow Language
  - Common Workflow Language (CWL) is being considered
- Docker containers
  - For improved portability and reproducibility
- Using emerging GA4GH standards
- Authorization and Authentication process

Broad Cloud Pilot

- PI: Gad Getz
- Collaborators: University California Berkeley, University California Santa Cruz
- Cloud Platform: Google
- Unique Technologies Used: ADAM/Spark
- Tools Incorporated: Firehose
- Cloud Pilot Website: http://firecloud.org

FireCloud

TCGA data

Your Data **Data**

Tools

Your Tools

ReCapSeg MuTect GISTIC PathSeq RNASeqQC SegSeq Gistic HapSeg MutSig2CV MutSigCVInterVar dRanger ABSOLUTE ContEst coMut InVexChainFinder MutSig1 MutSigCV RNASeQC CapSeg Oncotator

User or team Workspace

- Securely tracks and manages data, metadata, tools, job execution and results
- Captures provenance for each run (method versions, timestamps, input and output files)

Your Workflow

Workflows

Users

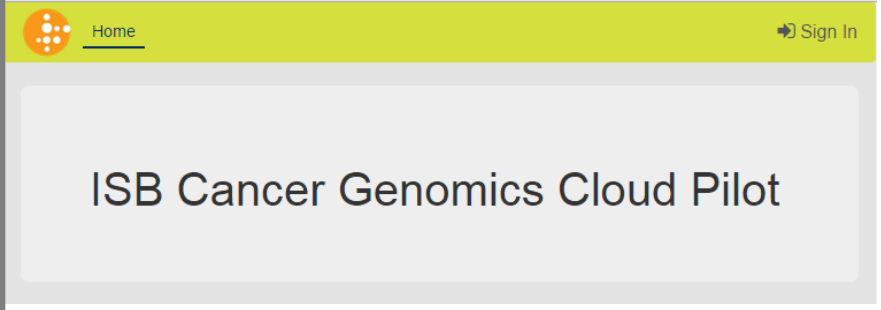
ProductionManager ProjectManager PI Group Lab ToolDeveloper Clinician Analyst

The FireCloud Platform

- Scaleable and elastic compute on Google Cloud Platform
- Versatile job executors:
  Docker, ADAM/Spark, Google Cloud Dataflow
- Data can be stored in scalable distributed 'stores':
  ReadStore, VariantStore
- Available Spring 2016
- GA4GH compliant data stores
- For more updates and information, leave your contact information at: *firecloud.org*

Institute for Systems Biology (ISB) Cloud Pilot

- PI: Ilya Shmulevich
- Collaborators: Google, SRA International
- Cloud Platform: Google
- Unique Technologies Used: Google Genomics Platform
- Tools Incorporated: Regulome explorer, Gene Spot
  - Focus on interactive data visualization, exploration and analysis
- Cloud Pilot Website: http://cgc.systemsbiology.net/



Interactive tools

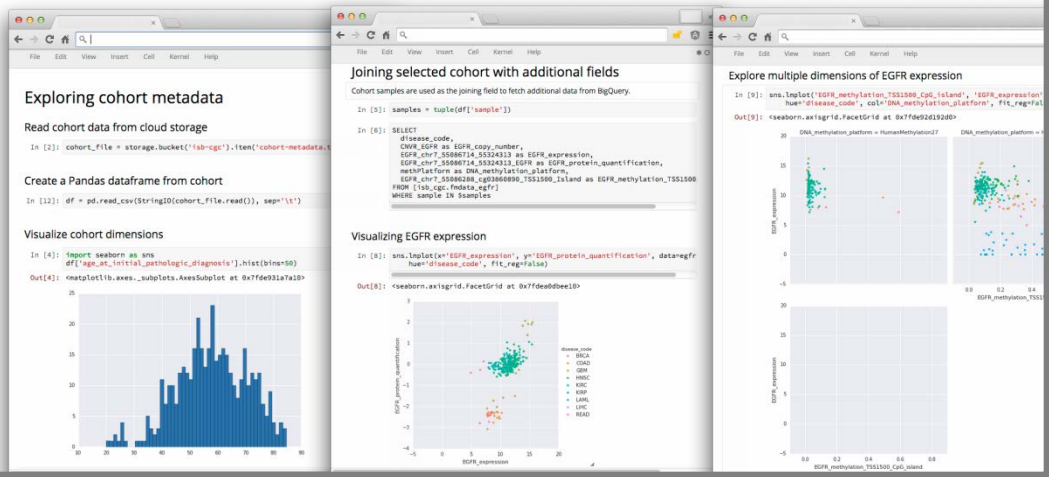
- explore all tumors or a subset
- define custom “cohorts”
- focus on specific molecular data types or platforms

Programmatic access

- REST APIs for Cloud Storage
- SQL-like queries for BigQuery
- GA4GH API for Google Genomics

Tutorials

- IPython notebooks
- RStudio (Rmd) files



Seven Bridges Genomics Cloud Pilot

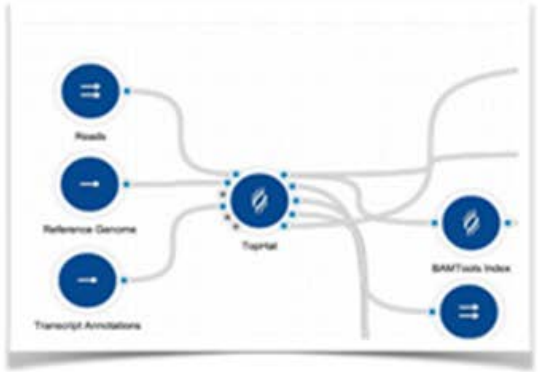
- PI: Deniz Kural
- Collaborators: None
- Cloud Platform: Amazon Web Services
- Unique Technologies Used: SBG platform
- Tools Incorporated: > 30 public pipelines
  - https://igor.sbgenomics.com/lab/public/pipelines/
- Cloud Pilot Website: https://www.sbgenomics.com/cancer-genomics-cloud/

CANCER GENOMICS CLOUD
SEVEN BRIDGES

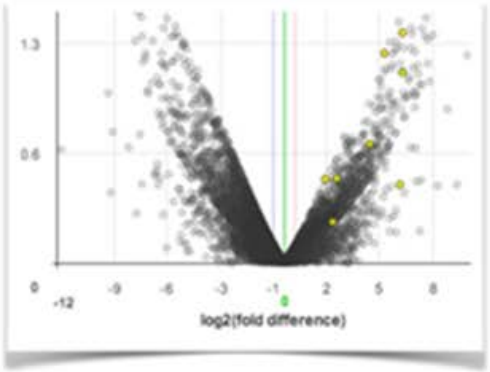
Run your tools on TCGA data. No waiting.

```
import os
from sbgsdk import define, Process


class SamtoolsSamToBam(define.Wrapper):
    class Inputs(define.Inputs):
        inp = define.input(required=True)

    class Outputs(define.Outputs):
        out = define.output()

    def execute(self):
        out_file_name = change_ext(self.inputs.inp,
        Process('samtools', 'view', '-S', '-b', sel
```

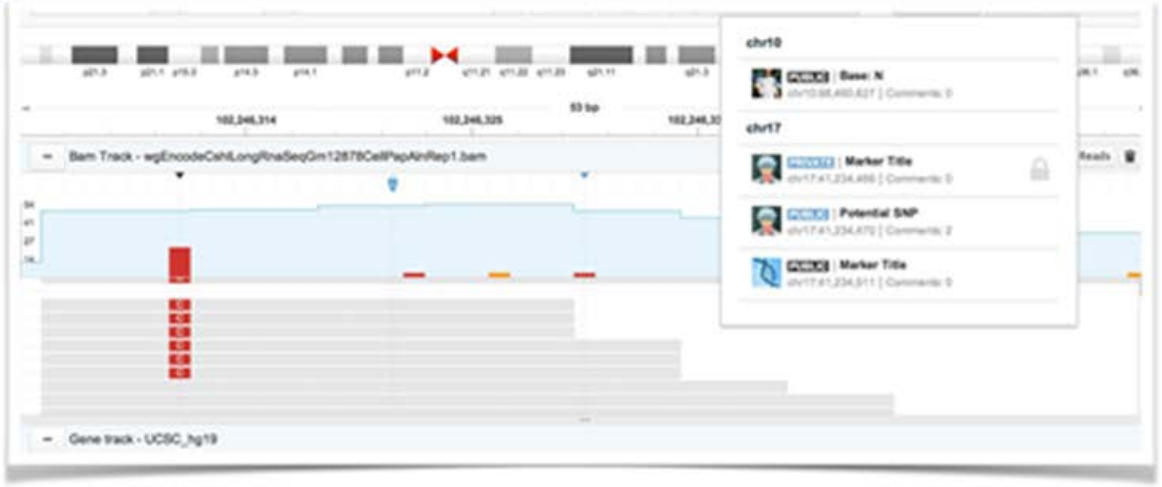
Wrap your tools



Mix and match with others …and TCGA data



Analyze



Collaborate and discuss what you discover with colleagues

Pre-register at http://www.cancergenomicscloud.org/

Cloud Pilot Workshop

- Additional details
- Today, 4-5 and 5-6pm
- Natcher Auditorium

Cancer Genomics Project Teams

CGC Pilot Team Principal Investigators

- **Gad Getz, Ph.D** - Broad Institute - http://firecloud.org
- **Ilya Shmulevich, Ph.D** - ISB - http://cgc.systemsbiology.net/
- **Deniz Kural, Ph.D** - Seven Bridges - https://www.sbgenomics.com/cancer-genomics-cloud/

NCI Project Officer & CORs

- Anthony Kerlavage, Ph.D – Chief Project Officer
- Juli Klemm, Ph.D – COR, Broad Institute
- Tanja Davidsen, Ph.D – COR, Institute for Systems Biology
- Ishwar Chandramouliswaran, MS, MBA – COR, Seven Bridges Genomics

GDC Principal Investigator

- Robert Grossman, Ph.D - University of Chicago

Center for Cancer Genomics Partners

- JC Zenklusen, Ph.D
- Daniela Gerhard, Ph.D
- Zhining Wang, Ph.D
- Liming Yang, Ph.D
- Martin Ferguson, Ph.D

NCI Leadership Team

- Warren Kibbe, Ph.D
- Lou Staudt, Ph.D
- Steven Chanock, Ph. D
- George Komatsoulis, Ph.D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