# The power of connection

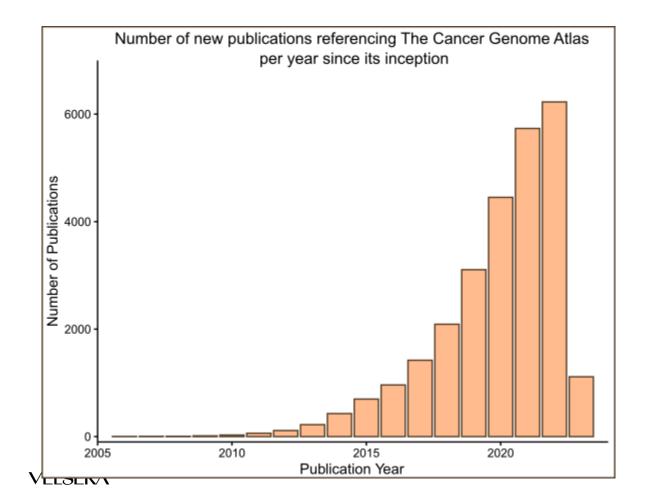
How the Cancer Research Data Commons enables researchers to connect data, computational tools, and collaborators to accelerate discovery

Brandi Davis-Dusenbery, PhD

Bioinformatics Training & Education Program (BTEP) May 4, 2023



# NCI has proven that large, shared data repositories accelerate discovery and improve patient outcomes.



#### March 4, 2013

- BACK TO PRESS RELEASES

Foundation Medicine Receives Accreditation from the College of American Pathologists

### Jan 22, 2019

# The FDA grants breakthrough device designation for TruSight Assay

Based on the content of TruSight Oncology 500, the proposed in vitro diagnostic will receive prioritized review and resources

Feb 16, 2023

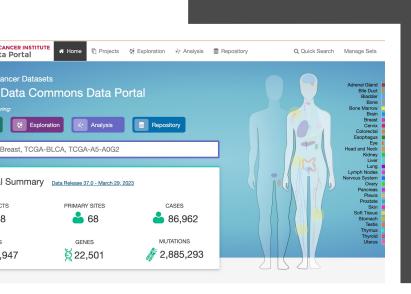
### Medicare Extends Coverage of Natera's Signatera<sup>™</sup> MRD Test to Breast Cancer

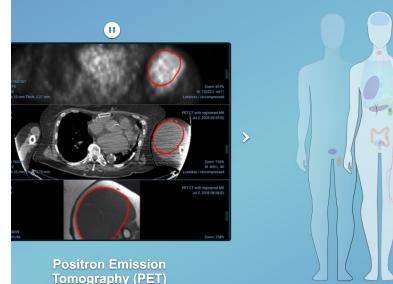
Coverage to include serial monitoring in all subtypes, including hormone receptor-positive, HER2-positive, and triple negative breast cancers

**NCI Cancer Research Data Commons** 

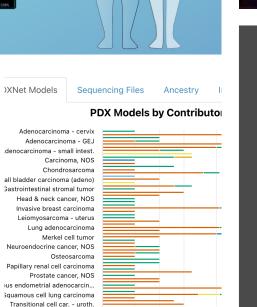
#### **NAL CANCER INSTITUTE**

Explore Images Collections Getting Started 🗹 User Forum 🗹 News About **Jing Data Commons** 









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10 Model Count per

### Human Tumor Atlas Network

HTAN is a National Cancer Institute (NCI)-funded Cancer Moonshot<sup>SM</sup> initiative to construct 3-dimensional atlases of the dynamic cellular, morphological, and molecular features of human cancers as they evolve from precancerous lesions to advanced disease. (Cell April 2020)

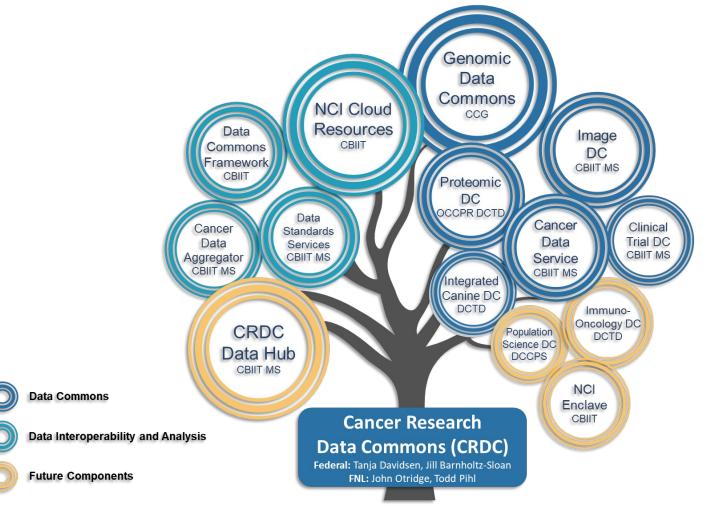
Explore latest Data Learn more about HTAN



**Proteomic Data Commons** 



# The NCI Cancer Research Data Commons connects diverse data with analytical power and tools.



VELSERA

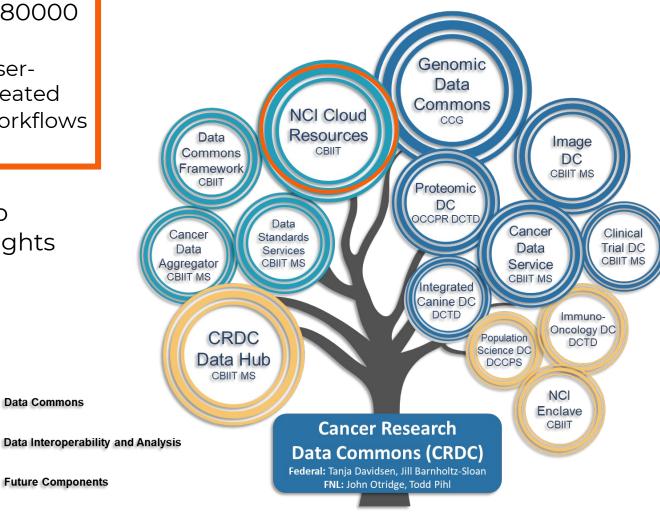
\*Data Hub, CDA, & CTDC have not reached go-live



3+	1600+	800+	8000+	80000
Petabytes Public Data		Public Tools & workflows	Users	User- created workflows

Provides powerful, yet easy to use interfaces to empower cancer researchers to draw new insights from petabyte scale data.

Stable, secure, and highly customizable cloud storage and computing platform



\*Data Hub, CDA, & CTDC have not reached go-live

## Who are the CGC Users?

The CGC is designed to serve a wide range of scientists and users with varying skill sets



#### ADMINISTRATORS

- Manage and Control Users
- Monitor and Control Institutional Assets
- Manage and Monitor Projects
- Monitor and Control Costs
- Create Reports



#### BIOINFORMATICIANS

- Store, Manage, and Share Data
- Access Public and Proprietary Datasets
- Query, Build, and Investigate Cohorts of Interest
- Access Optimized Tools
  and Workflows
- Create, Optimize, Maintain, and Distribute New Tools and Workflows

- Create Push-button Automation Solutions
- Analyze Data at Scale with Tools and Workflows
- Conduct Interactive Exploratory Analyses
- Explore/Visualize Results and Gather Insights
- Easily Collaborate with
  Other Stakeholders
- Integrate with External Systems



- Store, Manage, and Share Data
- Run Optimized Tools/ Workflows at Scale
- Conduct Defined Analyses
  via Push-button Solutions
- Investigate/Visualize Results
- Easily Collaborate with Other Stakeholders



#### CLINICIANS

- Conduct Validated Analyses via Push-button Solutions
- Query, Build, and Investigate Cohorts of Interest
- Create Reports
- Investigate/Visualize Results
- Easily Collaborate with Other Stakeholders



- Create, Optimize, and Maintain New Tools and Workflows
- Create Push-button
  Automation Solutions
- Create Custom Interfaces for Specific Use Cases
- Distribute Proprietary Tools/ Workflows
- Integrate with Upstream/ Downstream Systems

# Analyzing the genomic landscape of CHIP in solid tumor microenvironments.

Rutgers PhD Candidate Vaidhyanathan Mahaganapathy



### **Data Connectivity**

- Immediate access to TCGA data without needing to deal with long term storage.
- Originally proposed 5 tumor types, further optimization of tools allowed to extend to all qualifying tumor types.



## **Speed and Scale**

- Ran parallelized analysis over a weekend that would have taken 3+ months on HPC.
- Created millions of files over the course of the analysis which also excluded use of local resources.



### Community

"Office hours is a huge benefit, not only to help me, but also to not feel like I'm in a silo by myself."

• Optimized MERIT to be made available in tool repository to enable others to extend analysis.

# Accelerating collaboration, data harmonization, and sharing for large consortia.

Patient Derived Xenograft Network



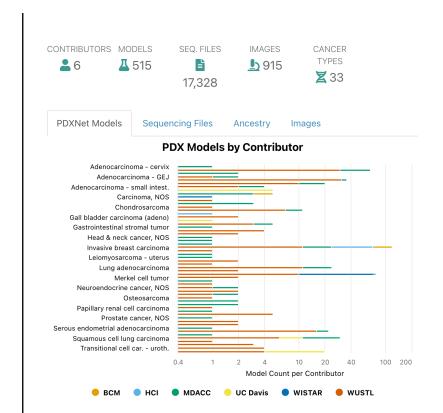
### **Data Connectivity**

- Multimodal data including: WXS, RNAseq, proteomics and Imaging
- 16 optimized workflows published in CWL.
- Cost predictor based on 7,000 observations



### Impact

- 60+ manuscripts published by PDXNet investigators over 5 years.
- Preclinical, *in vivo* evidence supporting 10 Clinical Trial LOIs incl ComboMatch & ETCTN



# Promoting bioinformatics education for the next generation of cancer researchers.

Diverse approaches and engagement strategies tailored to community needs



### **Train the trainer**

Min Zhang, M.D., Ph.D., researcher within the Department of Statistics at Purdue University

Developed and led an NCI-funded R25 initiative called "Big Data Training for Cancer Researchers".

After the 2022 workshop, **77% of the participants** stated in a survey that they were interested in or already using the Cancer Genomics Cloud to conduct their own research.



### Workshops

Judy Wawira Gichoya, MS, MD | National Institute of Biomedical Imaging and Bioengineering

Over **200 international scientists** and stakeholders from across Africa as part of DS-I Africa initiative.

Workshop on data harmonization using cloud resources, then a live demonstration showcasing a deep learning Al tool utilizing a Jupyter Labs Python Notebook to make predictions on one of the Covid-19 imaging datasets provided by Judy



### **Collaborative projects**

- Cost estimation, optimization, and planning support
- Great for researchers new to bioinformatics and cloud approaches
- Up to \$10k compute/storage costs
- Fast, rolling applications
- To date > 60 projects

# Introduction to the CGC

## Build and Execute Flexible Analytical Pipelines



### **User friendly portal**

Access petabytes of publicly available data, and analyze it alongside private data.



### **Unprecedented Collaboration Features**

Collaborate within and across organizations while keeping control of your assets with precise permission levels.



### Industry Standard Tools: Reproducibility

Execute, build, and customize analysis pipelines using popular tools such as CWL, WDL, Nextflow, Docker, RESTful APIs, and CLI.



### **Connected Cloud**

Keep data in your own buckets with storage support for AWS, and Google Cloud Platform.

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## Integrated Custom Tertiary Analysis Tools

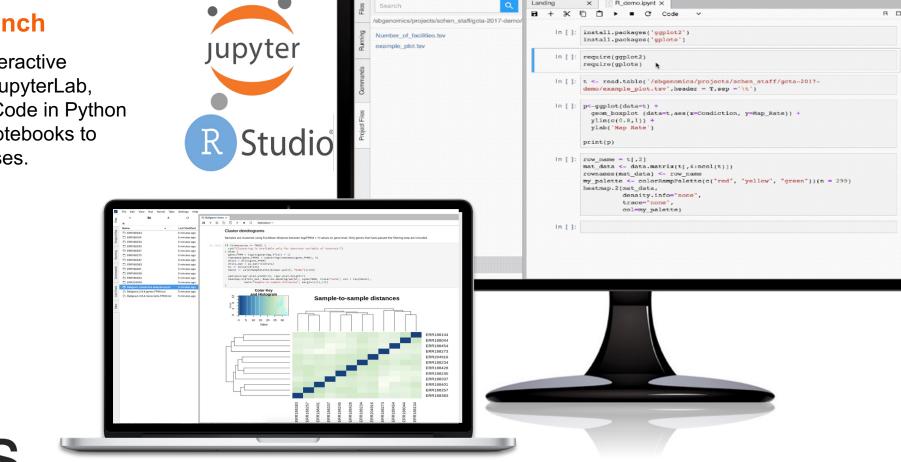
### **Data Science Workbench**

**COMING SOON** 

**=** Galaxy

PROIFC

Derive new insights using interactive analysis environments with JupyterLab, and RStudio environments. Code in Python and R, and create Jupyter Notebooks to record and share your analyses.

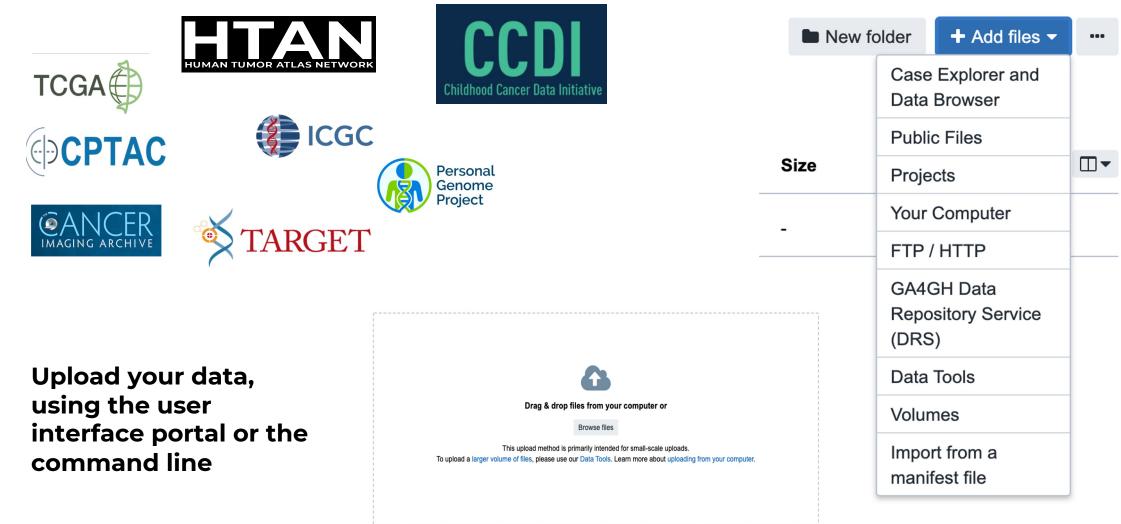


Stop Analysis

File Notebook

VELSERA

## Petabytes of public data, at your fingertips



VELSERA

# Making an impact

**112** Publications + Citations

**30+** Webinars + Video tutorials

**100+** Office hour sessions

# LINE-1 expression in cancer correlates with p53 mutation, copy number alteration, and S phase checkpoint

Wilson McKerrow<sup>a,b</sup>, Xuya Wang<sup>a,b</sup>, Carlos Mendez-Dorantes<sup>c,d</sup>, Paolo Mita<sup>a,b</sup>, Song Cao<sup>e,f</sup>, Mark Grivainis<sup>a,b</sup>, Li Ding<sup>e,f</sup>, John LaCava<sup>g,h</sup>, Kathleen H. Burns<sup>c,d</sup>, Jef D. Boeke<sup>a,b,i,1</sup>, and David Fenyö<sup>a,b,1</sup>

\*Institute for Systems Genetics, New York University Grossman School of Medicine, New York, NY 10016; <sup>1b</sup>Department of Bioch Pharmacology, New York University Grossman School of Medicine, New York, NY 10016; <sup>1b</sup>Department of Bioch Boston, MA 0215; <sup>1b</sup>Department of Phatology, Harvard Medicinel School, Boston, MA 0215; <sup>1b</sup>Department of Medicine and Gene Washington University in St. Louis, St. Louis, MO 63110; <sup>1k</sup>McDonnell Genome Institute, Washington University in St. Louis, St. Loois, St. Louis, St. Louis, St. Loois, St. Louis, St. Louis, St. Louis, St. Loois, St. Loois

Taylor & Francis Taylor & Francis Group

Contributed by Jef D. Boeke; received August 31, 2021; accepted January 14, 2022; reviewed by Molly Gale Hammell and Michav Xavier Bofill-De Ros 💕, Zhenyi Hong<sup>b</sup>, Ben Birkenfeld<sup>a</sup>, Sarangelica Alamo-Ortiz<sup>a</sup>, Acong Yang<sup>a</sup>, Lisheng Dai<sup>a</sup>,



Roozbeh Dehghannasiri, Julia Eve Olivieri, Ana Damljanovic & Julia Salzman 🖂

**GA4GH Standards** 

Genome Biology 22, Article number: 219 (2021) Cite this article

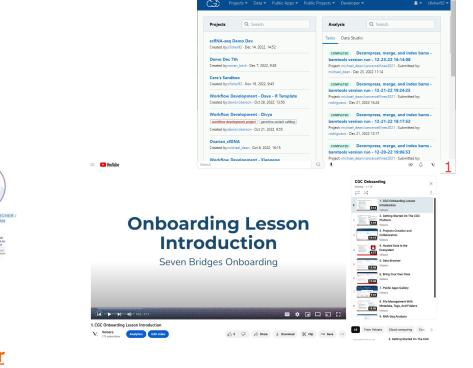
Specific splice junction detection in single cells with

Short Report | Open Access | Published: 05 August 2021

1336 Accesses | 30 Altmetric | Metrics

SICILIAN

\*RNA Mediated Gene Regulation Section, RNA Biology Laboratory, Center for Cancer Research, National Cancer Institute, Frederick, MD, USA; \*Neural Development Section, Mouse Cancer Genetics Program, Center for Cancer Research, National Cancer Institute, Frederick, MD, USA



# Live demo!!

# Trends in Cancer Supports open access

### *TP53* Mutations and Outcomes in Breast Cancer: Reading beyond the Headlines

Ashkan Shahbandi • Hoang D. Nguyen • James G. Jackson 🙁

Published: February 05, 2020 • DOI: https://doi.org/10.1016/j.trecan.2020.01.007 • 🔍 Check for updates

Submit

**Highlights** 

vords	TP53 is mutant in 30% of all breast cancers and is the most
rences	frequently mutated gene. The role of TP53 in the management
le info	of breast cancer remains unclear.
	Recent analysis reveals that mutant p53 can be detrimental or
ted Articles	beneficial to clinical outcome, depending on treatments given.

What gene expression changes are present in breast cancer tumors harboring p53 nonsense mutation compared to breast cancer tumors which express p53 at a similar expression level without mutation.

liahts

# Live demo!!

### **Differential expression analysis**

#### Table of adjusted p-values ordered features:

##		target_id	ext_gene	num_aggregated_transcripts	<pre>sum_mean_obs_counts</pre>	pval	qval
##	1	ENSG00000104419	NDRG1	16	62.18210	7.953447e-10	2.238339e-05
##	2	ENSG0000073849	ST6GAL1	12	42.41697	5.085899e-09	7.156623e-05
##	3	ENSG00000126217	MCF2L	19	78.84039	1.554294e-08	1.458083e-04
##	4	ENSG00000141552	ANAPC11	19	72.25131	3.160626e-08	2.223737e-04
##	5	ENSG00000242265	PEG10	5	28.77247	8.146654e-08	4.585426e-04
##	6	ENSG00000099204	ABLIM1	6	26.49753	1.930873e-07	9.056759e-04

> J Natl Cancer Inst. 2022 Apr 11;114(4):579-591. doi: 10.1093/jnci/djab222.

### NDRG1 in Aggressive Breast Cancer Progression and Brain Metastasis

Emilly S Villodre <sup>1</sup> <sup>2</sup>, Xiaoding Hu <sup>1</sup> <sup>2</sup>, Bedrich L Eckhardt <sup>1</sup> <sup>2</sup> <sup>3</sup>, Richard Larson <sup>2</sup> <sup>4</sup>, Lei Huo <sup>2</sup> <sup>5</sup>, Ester C Yoon <sup>5</sup>, Yun Gong <sup>2</sup> <sup>5</sup>, Juhee Song <sup>6</sup>, Shuying Liu <sup>1</sup>, Naoto T Ueno <sup>1</sup> <sup>2</sup>, Savitri Krishnamurthy <sup>2</sup> <sup>5</sup>, Stefan Pusch <sup>7</sup> <sup>8</sup>, Debu Tripathy <sup>1</sup> <sup>2</sup>, Wendy A Woodward <sup>2</sup> <sup>4</sup>, Bisrat G Debeb <sup>1</sup> <sup>2</sup>

Affiliations + expand

 PMID: 34893874
 PMCID: PMC9002276
 DOI: 10.1093/jnci/djab222
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#### Abstract

**Background:** N-Myc downstream regulated gene 1 (NDRG1) suppresses metastasis in many human malignancies, including breast cancer, yet has been associated with worse survival in patients with inflammatory breast cancer. The role of NDRG1 in the pathobiology of aggressive breast cancers remains elusive.



Create an account and access free credits at <u>CancerGenomicsCloud.org</u>

## Stay in touch.



Cera Fisher, PhD Community Engagement Manger Cera.Fisher@velsera.com



### **Every Week:**

- 10:00 am ET Tuesday
- 2:00 pm ET Thursday



Rowan Beck, PhD Community Engagement Manager Rowan.Beck@velsera.com



Zelia Worman, PhD Director of Researcher Engagement and Education Zelia.Worman@velsera.com